Genetic Discrimination:
Genealogy of an American Problem

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ABSTRACT

Genetic discrimination has been transformed from an isolated concern of a handful of professionals into a pressing civil rights and public policy problem in the United States over the last twenty years. My dissertation is a genealogical account of how genetic discrimination has been shaped into a problem of this stature. It answers two questions: Where did the problem come from? How has the problem changed over time?

In Part One, I trace the history of concerns about discrimination from the 1970s to the present. Drawing from oral histories with key actors and organizations that shaped early public understanding of the problem, I show that concerns about genetic discrimination originated in diverse practices. These practices include workplace genetic screening, insurer discrimination against individuals with AIDS, the rapid commercialization of genetic tests in the 1980s, and health care reform.

In Part Two, I present findings from a three-year ethnographic study of public policy hearings on genomic medicine in the United States that illustrate how new actors have been defining the problem of genetic discrimination since 1995. The hearings of the Secretary's Advisory Committee on Genetics, Health and Society were a site where participants legitimized genetic discrimination as a civil rights problem and developed lobbying tools to persuade Congress to pass federal nondiscrimination legislation. Participants framed fear of discrimination as a barrier to the nation's scientific progress and a significant threat to the lives of Americans.

I use the construct of genomic citizenship to draw out claims about the rights and duties of Americans in contemporary discourse on genetic discrimination. Passing federal nondiscrimination legislation is one way in which the civil rights of Americans appear to be expanding, while their responsibilities to act genetically are increasing. Advocates of nondiscrimination legislation, who use the language of genetic defect to argue that everyone is vulnerable to discrimination, geneticize all Americans by enrolling them into the biosociality of the flawed, transparent genome, with attendant duties. What these advocates do not also champion is the right of Americans to refuse to think or act genetically.

RÉSUMÉ

La discrimination génétique est passée du statut de préoccupation isolée parmi un petit nombre de professionnels à celui d'un urgent problème de droits civils et de politique publique aux État-Unis, depuis les vingt dernières années. Ma thèse est un compte rendu généalogique de la transformation de la discrimination génétique en un problème d'une telle envergure. Elle répond à deux questions : Quelle est l'origine du problème? Comment le problème a-t-il changé avec les années?

Dans la première partie, je retrace l'histoire des préoccupations au sujet de la discrimination, des années 70 à aujourd'hui. Je puise dans la tradition orale chez des acteurs clés et des organismes de premier plan qui ont informé la compréhension initiale du problème par le public. Je montre comment les préoccupations entourant la discrimination génétique sont issues de différentes pratiques.

Dans la deuxième partie, je présente les résultats d'une étude ethnographique d'une durée de trois ans, traitant des audiences publiques sur la médecine génomique aux États-Unis, et illustrant comment de nouveaux acteurs ont défini le problème de la discrimination génétique depuis 1995. Dans le cadre des audiences du Secretary's Advisory Committee on Genetics, Health and Society, les participants ont identifié la peur de la discrimination comme un obstacle au progrès scientifique de la nation, de même qu'une menace significative pour la vie des Américains et des Américaines.

J'ai recours à la construction de citoyenneté génomique dans le but de dégager des revendications au sujet des droits et devoirs des Américains et des Américaines, en rapport avec le discours actuel sur la discrimination génétique. L'adoption d'une législation de non discrimination semble contribuer à l'élargissement des droits civils des Américains et des Américaines, tandis que s'accroît leur responsabilité d'agir sur le plan génétique. Les défenseurs de la législation de non discrimination emploient le langage des défaut génétiques pour soutenir que toute la population est sujette à la discrimination. Selon ces mêmes défenseurs, les Américains et Américaines n'ont pas le droit de refuser de penser ou d'agir en termes génétiques.

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LIST OF ABBREVIATIONS

ADA:	**The Americans with Disabilities Act of 1990**
AHIP:	**America's Health Insurance Providers**
CDC:	**Centers for Disease Control and Prevention**
CEPH :	**Centre d'Etude du Polymorphisme Humain (France)**
CMS:	**U.S. Centers for Medicare and Medicaid Services**
DHSS	**U.S. Department of Department of Health and Human Services**
DoD:	**U.S. Department of Defense**
DOE:	**U.S. Department of Energy**
DTC:	**Direct-to-consumer**
EEOC:	**U.S. Equal Employment Opportunity Commission**
ELSI:	**Ethical, Legal, and Social Implications of the Human Genomic Project**
FACA:	**Federal Advisory Committee Act of 1972**
FDA:	**U.S. Food and Drug Administration**
GINA:	**The Genetic Information Nondiscrimination Act of 2008**
GRAD:	**Genomic Research in the African Diaspora (Howard University)**
HGP:	**Human Genome Project**

HIPAA:	**The Health Insurance Portability and Accountability Act of 1996**
NBCC:	**National Breast Cancer Coalition**
NCHGR:	**National Center for Human Genome Research**
NHGRI:	**National Human Genome Research Institute**
NIH:	**National Institutes of Health**
OBA:	**Office of Biotechnology Activities (NIH)**
OTA:	**U.S. Congressional Office of Technology Assessment (1972-1995)**
SACGHS:	**The Secretary's Advisory Committee on Genetics, Health, and Society (2003-2010)**
SACGT:	**The Secretary's Advisory Committee on Genetic Testing (1999-2002)**

Introduction

A PRESSING NATIONAL PROBLEM

Wednesday, May 21, 2008 was a day of victory for genetic discrimination activists across the United States. After working for thirteen years to persuade Congress to pass comprehensive federal legislation banning genetic discrimination by insurers and employers, they were finally celebrating. President George W. Bush had signed into law the Genetic Information Nondiscrimination Act (GINA) (See Appendix B). Genetic activists heralded GINA as "the first civil rights legislation of the new millennium" (Genetic Alliance 2008).

The signing came three weeks after the U.S. House of Representatives had voted 414-1 in a remarkable show of bipartisan support to pass H.R. 493, on May 1, 2008, and four weeks after the Senate had voted 95-0 to pass S. 358, a similar version of the bill, on April 24, 2008. GINA is the first comprehensive federal legislation to extend protections and remedies to individuals and families against the misuse of genetic information by third parties. The measure prohibits health insurers from using genetic information to deny coverage or to charge higher premiums to a healthy person or family. It prohibits employers from using genetic information in their hiring, firing, or promotion decisions. And it prohibits insurers and employers from requesting or requiring genetic testing from an individual or a family.

Member organizations of the Coalition for Genetic Fairness, who have been working since 2001 to pass federal nondiscrimination legislation, were quick to praise passage of GINA. "GINA will be the first civil rights

act passed by the Congress in almost 20 years," said Jeremy Gruber, legal director for the National Workrights Institute (Coalition for Genetic Fairness 2008). “This marks the first time in the history of our country that legislation has been passed to protect against genetic discrimination before it becomes deeply ingrained in the very fabric of our society,” he said. Angela Trepanier, president of the National Society of Genetic Counselors, and Joann Boughman, executive vice-president of the American Society of Human Genetics, voiced the oft-repeated hope that passage of GINA would dispel fears and remove a significant barrier to Americans seeking genetic testing and participating in genomic research and clinical trials. “The floodgates are about to open,” said Trepanier. “When GINA becomes law and genetic information is protected, we expect more people to seek out genetic testing that can help in the prevention or management of a broad range of diseases and conditions,” she said (National Society of Genetic Counselors 2008). Boughman’s message was similar. “With the long-awaited federal passage of GINA, researchers and clinicians can now actively encourage Americans to participate in clinical trials without the fear of genetic discrimination,” she said (Coalition for Genetic Fairness 2008). “Once this legislation has taken effect, clinicians will be able to order genetic tests for patients and their families in a manner that ensures the full realization of the advantages of personalized medicine, while also easing patients’ concerns about the risk of genetic discrimination by insurance companies and employers based on this data” (Genetic Alliance 2008). But it was Congressional Representative Louise Slaughter, a Democrat who has represented western New York State since 1986 and has sponsored versions of GINA in Congress since 1995, who

voiced the sentiment that went to the heart of the problem. Genetic discrimination is everyone's problem because all Americans carry genetic risks for disease. "This is a tremendous victory for every American not born with perfect genes—which means it's a victory for every single one of us," she said. "Since all of us are predisposed to at least a few genetic-based disorders, we are all potential victims of genetic discrimination" (Genetic Alliance 2008).

These accolades for GINA offer some indication of the stature that the problem of genetic discrimination has acquired in the United States over the last twenty years. The problem is unfamiliar to most Canadians, who greet the expression "genetic discrimination" not with anxiety or dismay, but with curiosity and puzzlement. Few Canadians are aware that genetic discrimination has become a major public policy issue in the United States, or that it has become the object of considerable activism by a diverse group of actors that today comprise the landscape of genetic politics in the United States (Health et al 2004; Taussig et al 2003; Taussig 2005). These actors include genetic advocacy organizations, federal science agencies, biotechnology firms, and the coalition and lobbying groups they have developed.

Genetic discrimination dominates public discourse about genomic medicine and genetic testing in the United States.[1] It has become impossible to hold a policy conversation about either of these without provoking a passionate dialogue about the need to protect Americans—and the nation—from this threat.[2] The issue overshadows and, at times, displaces, concerns about the utility of susceptibility testing for common

diseases like breast and colon cancer, the therapeutic gap between genetic tests and clinical therapies, the inability of the current health care system to deliver a vision of genomic medicine, and the fact that comprehensive reproductive services and prenatal care are unaffordable or unavailable for millions of pregnant women.[3] Discourse about genetic discrimination has become wedded to narratives of scientific progress, technological prowess, and the desire of all Americans to predict, prevent, and control disease.

The significance of the successful passage of GINA to advocates of the legislation cannot be overemphasized. Persuading Congress to pass comprehensive federal nondiscrimination legislation had become such a visible and important goal to the many individuals and organizations championing this issue that a story about their heroic efforts might seem like the one worth telling. Certainly, the story of legislative activism is complex and engaging, and deserves its own hearing. But the legislative activism story does not address underlying questions about genetic discrimination, particularly why it takes up so much space and attention in the American public sphere today.

The cultural significance of genetic discrimination—the power that the issue has acquired to dominate policy discussion, provoke activism, and displace other constitutions of American citizens as vulnerable—calls for investigation. Where did this issue come from? Why are Americans galvanized by the problem? Why are so many Americans—the majority of whom may never undergo susceptibility or diagnostic testing for gene-based disorders—considered to be at risk of discrimination? What kind of political work does the problem of genetic discrimination do, and what

opportunities does it create? What problems does attention to genetic discrimination obscure or absorb? In this dissertation, I tell a different story that begins to answer these questions.

I begin by adopting a political-economic approach that situates contemporary discourse about genetic discrimination in the history and rationales of the Human Genome Project (HGP). Drawing from the work of political scientist Rodney Loeppky on the HGP in the United States, and anthropologist Kaushik Sunder Rajan (2005, 2006) on genomics in India and the United States as a form of biocapital, I treat genomics in the United States as a post-Cold war investment of capital and infrastructure. Investment in genomics is intended to transfer technology to the private sector, develop the economy, bolster national prestige, and shore up the competitive decline of the United States in biotechnology. But rather than investigate how the United States is trying to "catch up" to other countries such as India (see Sunder Rajan 2005, 2006), as would be suggested by an economic ethnography of genomics, I examine how the United States has grappled with a domestic problem that is widely perceived to block progress in genomics: genetic discrimination.

For genomics to be successful in the United States, one of the tasks of federal scientific agencies, such as the National Human Genome Research Institute (NHGRI) and the Centers for Disease Control (CDC), is to shape Americans into two types of subjects: consumers who will use genome sequencing to identify their personal risks of disease and pharmacogenomic medicines to prevent or treat these diseases; and research subjects who will participate in large-scale genomic research

studies and clinical trials of pharmacogenomic medicines (cf. Sunder-Rajan 2005). Scientists at these agencies, as well as genetic activists who have worked closely with them, have gone on record as stating that public fear of genetic discrimination constitutes a significant barrier to the success of the genomics enterprise in this country. Americans are unwilling to undergo genetic testing or participate in genomic research because they fear discrimination by health insurers, they have declared. I examine these claims, and situate this analysis in a genealogy (Foucault 1977) of genomic discrimination that traces how different actors have shaped public understanding of discrimination over the last thirty years.

My dissertation makes two contributions to medical anthropology and the anthropology of new genetics. First, it brings the state into view as an actor with a great deal at stake in reassuring Americans that it is safe to participate in genetic testing and genomic research.[4] The state has been a neglected actor in studies of biopolitics (Foucault 1978a; 2008) and biosociality (Rabinow 1996) in the United States, particularly anthropological studies of genetic advocacy organizations and patient support groups (see, for example, see Heath et al 2004; Novas 2006; Rapp 2002; Rapp et al 2001, Rapp et al 2006; Taussig 2005; Taussig et al 2003). These studies have examined the activism of advocacy organizations and patient support groups apart from federal genomics, and outside of a political-economic analytical framework. Yet it is the state's commitment to the HGP and post-HGP research that has helped these organizations to thrive. While these studies help us to understand how genetic patient groups are becoming important players in the enterprise of scientific-knowledge production, we have to ask: Where is the

state in all of this? In the United States, genomics is the largest "big science" biology project the nation has ever undertaken. In this dissertation, I situate these genetic advocacy organizations and patient support groups, which have championed the passage of federal legislation banning genetic discrimination, within the broader context of genomics as an enterprise of the state.

Secondly, genomics in the United States is not just any large-scale scientific initiative. It is a nation-building project. With some important exceptions (see Biehl 2004b and Cataldo 2008 on Brazil, Brotherton 2008 on Cuba, and Petryna 2002, 2004 on the Ukraine), medical anthropologists have not theorized health discourses and biomedical initiatives through the nation-building lens. But I argue in this dissertation that this is precisely what is going on in the United States, with genetic discrimination in particular and genomics in general. The state is engaged in genomic nation-building in a number of ways: in annual Congressional appropriations to the NHGRI based on the NHGRI's promises of discoveries and treatments to come, and on the perceived need to "keep up" with the genomic initiatives of other nations (for example, by developing a large-scale biobank); in the NHGRI aggressively promoting its genomics initiatives, such as the large-scale population cohort study and the Surgeon General's "My Family Health Portrait," both of which I talk about in Chapter 7; and in state actors articulating imaginaries of the United States as a technological innovator and progressive nation through genomics. I argue in this dissertation that this nation-building work has become transparent on the public stage of the hearings of the Secretary's Advisory Committee on Genetics, Health and Society (SACGHS), during

discussions about genetic discrimination. It is here, in these presentations and testimonies, in claims about the rights of Americans to be protected against discrimination based on the genetic disorders and flaws that all carry, that the work of shaping of Americans into genomic citizens and ideal subjects of a genomic nation is evident.

Above all, this dissertation is an ethnography of the problem of genetic discrimination in the United States (as opposed to an ethnography of individual experiences of genetic discrimination, for example). Using qualitative data, the dissertation provides a substantive account of how the problem of genetic discrimination has acquired a prominent status in the United States since the 1970s. It examines some of the discourses and activities that have that have shaped this problem, and identifies the goals of individuals and organizations that have played key roles in shaping this problem. More precisely, it is an *institutional ethnography* of the SACGHS that shows how this committee legitimized genetic discrimination as a problem that required immediate action by Congress.

This dissertation is also reflexive and interpretive. Following in the tradition of ethnographic writing, I reflect on my own observations and assumptions. I interrogate certain ideas—for example, the idea that all Americans seek self-improvement through genetic testing, and have the resources and desire to act on genetic self-knowledge—as specific to the United States, rather than taking these for granted as universal or neutral. Finally, I reflect on the implications of these ideas.

GENEALOGIES OF NATURAL FACTS AND SOCIAL PROBLEMS

By a genealogical approach (Foucault 1977), I mean that I investigate where the problem of genetic discrimination has come from, why it has become synonymous with insurer and employer discrimination, and why it has become the subject of passionate debate and intense activism in the United States. For Michel Foucault, following Nietzsche, a genealogy is a search for origins, not in the form of a linear and progressive story, but as a partial and branching account in which the meaning of words change over time, and ideas lose their logic. Foucault emphasizes the importance of power relations in shaping discourse, and I bring that into focus in my ethnographic analysis of the SACGHS hearings.

I am treating genetic discrimination as a social construction: a real social problem that nonetheless has been named, defined, framed—and redefined—by many people with different interests. In adopting this approach, I am not denying that individuals and families have experienced discrimination by insurers, employers, and other institutions as a consequence of undergoing genetic testing. What I argue is that the definition and scope of genetic discrimination is plastic. Before 1992, genetic discrimination was not widely recognized as a problem of discrimination by health insurers (in particular) and employers against healthy persons who carry mutations for rare, genetic disorders. Moreover, since 1992, this understanding of the target of discrimination has expanded, such that all Americans are understood to be vulnerable to genetic discrimination, not just individuals who are carriers of rare mutations.

Ian Hacking's (1999:6) distinction of tacit claims is useful here.

Following Hacking, I note that genetic discrimination is a problem whose parameters seem to be well-established. I also note that it is not inevitable. I start from the position that practices of discrimination have not been sufficient for genetic discrimination to become a recognized problem. The public commentary on the stigmatization and discrimination of African-American sickle cell carriers during the 1970s clearly demonstrates this. (I discuss this further in Chapter 4). Genetic discrimination has become a widely-recognized problem because individuals and organizations have named, framed, and redefined the problem. One of the consequences of this shaping work is that today, genetic discrimination seems to have a life of its own, an appearance of inevitability. Following Ian Hacking's lead, I do not make any claims about whether genetic discrimination is good or bad, or any judgements about the merits of federal nondiscrimination legislation. I look instead at why genetic discrimination has acquired its stature as a significant problem in public discourse, when there is little evidence to suggest that Americans routinely experience discriminatory decision-making by health insurers as a consequence of undergoing genetic testing, or that fear of discrimination by health insurers and employers is the biggest barrier to Americans seeking genetic testing.

Although my account of genetic discrimination is properly termed a genealogy, it could also be called a "biography," for two reasons. Economic anthropologist Igor Kopytoff (1986) developed what he called a biographical method of tracking the life cycle of a single object through its changing identities as non-commodity and commodity. His biographical approach treats the process of commodification as dynamic, one in which objects acquire and shed identities as a consequence of being circulated by

people in their economies. In an analogous fashion, I can track the changing identity of a problem called genetic discrimination from 1992, when it was named, to the present.

But genetic discrimination did not exist before 1992. How then to understand the roots of genetic discrimination and the origins of concern about actuarial decision-making that labels healthy individuals diseased or at-risk? For this, I have done two things. I have looked at the three decades from the 1960s to 1992 to identity practices that were labelled discriminatory and that were directly tied to genetic screening and testing practices. This research led to a second step: identifying changing understandings of which experienced discrimination as a consequence of genetic screening and testing practices. This opened up a series of questions: What was genetic about the practices that produced discrimination? What was discriminatory about their experiences? Did the problems experienced constitute something called genetic discrimination? Why or why not? What I discovered was that this understanding of vulnerability to discrimination was indeed like a branching structure of a tree, in that one understanding would seem to take hold for a while in conjunction with a set of screening and testing practices, then fall off in public awareness, to be replaced by another understanding tied to a different set of screening and testing practices.

My other strategy has been to identify individuals (and organizations) who have drawn attention to what they perceived as a unique form of inequity. This dissertation is as much a story about the individuals who have given genetic discrimination shape and form because of their

interests, concerns, networks and resources, as it is about the changing problem of genetic discrimination. By tracking the careers of these actors and examining their statements about genetic discrimination, I have identified some of the practices and problems that were departure points for their shaping activities. This is the second way in which my genealogy is biographical.

Genealogies of natural facts and social problems are well-represented in medical anthropology and other fields. In the next section, I identify work that has provided models for this dissertation.

Medical Anthropology: Allan Young and Margaret Lock

Within medical anthropology, Allan Young and Margaret Lock have combined historical research with ethnographic study to provide powerful accounts of how seemingly timeless and universal medical objects, facts, categories, and diseases have been produced in specific times and places by experts and patients, political activity, discourse, and professional practices. Young (1995) challenges the notion that traumatic memory is a "found object" or a timeless natural phenomenon, by offering an account of the invention of post-traumatic stress disorder (PTSD) that locates the emergence of traumatic memory—today a well-known diagnostic category that has migrated into mainstream usage—in the late nineteenth-century litigation over railway spine. The phenomenon reappeared as shell-shock during the First World War, declined in diagnostic popularity, then was concretized as the psychiatric disorder PTSD in 1980 with the publication of the DSM-III, after activism to legitimize the suffering experienced by Vietnam War veterans and compensate them.

Young's ethnographic study of group therapy sessions at a psychiatric facility for Vietnam veterans vividly illustrates the reflexive subject-making process: talking about PTSD and its symptoms is a necessary part of creating the subjects who suffer from the disorder. Their experiences are framed psychodynamically and given meaning. While his concern is to show how therapists understand the mechanism of traumatic memory and interpret what he calls the double story of patients' narratives, his fieldwork also shows the role that narrative performance plays in making problems and their victims concrete. This narrative performance is a key element of the SACGHS hearings, and I show how, similarly, actors there gave concreteness to the problem of genetic discrimination and its victims.

Margaret Lock's unrivalled work on the medicalization of menopause in North America (Lock 1993) and the invention of brain death as an adjunct to organ procurement in North America (Lock 2002a) draw on comparative studies (in both cases, with Japanese practices and experiences) to show how so-called natural facts and categories are produced in specific political and cultural contexts, and promulgated by clinicians and medical practice, scientists, and sometimes, patients. *Encounters with Aging* is the best-known of a substantial body of work by Lock on the changing roles of Japanese women, women's experiences of menopause in Japan and North America, and medicalization of the life cycle. Its strengths are the powerful combination of quantitative research (a survey of 1,738 Japanese women reporting symptoms of konenki, the gradual process of mid-life transition between ages forty-five and fifty-five) with qualitative research (interviews with women undergoing koneki), a historicized treatment of the medicalization of menopause in

North America, and a critique of how women's bodies and their aging processes are pathologized by biomedical thinking and practice. Perhaps most useful for this dissertation, her book begins with a discussion of how researchers have created a language of menopause, and underlines the enormous authority of scientists and their discourses in creating and shaping problems. Where she focusses on the experiences of individual women with their bodies and lives as mediated by biomedical categories and intervention, I am concerned with the production of an American body that is both genetically-flawed (harbouring devastating time-bombs that need to be revealed so that individuals can intervene and control their health) and a resource for a nationally-defined genomics enterprise.

Twice Dead explores the invention of the brain death criterion and its routinization to facilitate organ transplantation. Her genealogy of the brain death criterion and subsequent routinizing of organ transplantation starts with the invention of the ventilator, which in turn created a new medical object: the living cadaver. Lock problematizes North American norms, asking why the criterion was so readily adopted in North America with little controversy, in contrast to Japan, where it has created controversy and sits uncomfortably with the deference that the medical profession shows to families of patients and the familial habitus that death is a non-medicalized process involving the whole body and person, not simply the brain.

Lock is confronting larger questions in *Twice Dead*: Where does life end and death begin? Is it an event or a process? Who has the authority to decide? How do different societies at different times designate boundaries

between the two? Her anthropological approach to constructionism is clear in comments she makes on the boundary-making around brain death and organ procurement: "My task as ethnographer is to go one step further to consider why certain responses, decisions, and commentaries have become dominant and 'naturalized' and why other possibilities may be either openly disputed or completely beyond consideration" (p. 51). In a similar fashion, I hope to show why, in contemporary genetic discrimination discourse, one construction of Americans as vulnerable in particular has become dominant and displaced others.

Beyond Medical Anthropology

The field of history and philosophy of science furnishes models of how ideas and problems have been constructed. Ian Hacking is well-known for his accounts of the origins of multiple personality disorder (Hacking 1995) and the emergence of statistical thinking in the nineteenth century (Hacking 1990). However, an earlier work on the sudden emergence of probability in the mid-seventh century, *The Emergence of Probability* (Hacking 1975) was an influential first exposure to a constructionist account. It modelled the strategy of tracing a concept along changing epistemological frameworks.

The large body of work on the construction of fetal personhood from history of medicine, sociology, and cultural studies also has been a persistent influence on my thinking. This literature has been useful for thinking about how technological, scientific, and legal practices can produce or concretize new subjects that are eligible for protection, even rights claims. Although American scholars dominate this scholarship, it

was German historian Barbara Duden, and her marvellous exploration of how female patients in early modern Germany experienced their bodies (Duden 1991), that was my introduction to this literature. Duden followed this with an account of how imaging technologies have shaped new ways of experiencing the pregnant body in *Disembodying Women: Perspectives on Pregnancy and the Unborn* (Duden 1993), and later work in the anthology *Fetal Subjects, Feminist Positions* (Duden 1999).

Alongside Duden's careful scholarship, American feminist scholars (Hartouni 1997, Petchesky 1987, and Stabile 1992) have explored how anti-abortion activists have used fetal images to pursue claims for fetal personhood. Inherent to the process of visually producing fetal personhood is a politics of erasure, in which the pregnant mother is made to disappear visually or discursively. Their work has been influential, but a more sophisticated approach to understanding how technological practices intersect with politics can be found in the work of sociologist Monica Casper on fetal surgery (1994, 1998). She offers a thoughtful account of how a controversial speciality, fetal surgery, has produced a new subject, the "unborn patient," with attendant needs for protection as a vulnerable subject.

ORGANIZATION OF THE DISSERTATION

This dissertation is divided into four parts: an Introduction (Chapters 1-3); Part One (Chapters 4-5); Part Two (Chapters 6-9); and a Conclusion (Chapter 10).

In Chapter 2 ("Finding the Problem"), I describe why I shifted my

research focus from the direct-to-consumer (DTC) genetic testing industry in the United States, to genetic discrimination. I outline my research design, and discuss the rationales and challenges of studying elites and social problems of the United States. In Chapter 3 ("Situating the Problem"), I outline the genetic privacy story, which is the dominant account of the origins of genetic discrimination, and identify its shortcomings. I situate my account of genetic discrimination within two sets of literature: the anthropology of new genetics, which is my primary set of literature; and the anthropology of citizenship.

The literature of the anthropology of new genetics is significant for examining the far-ranging individual and social impacts of molecular discoveries and interventions into life, death, reproduction, and disease. Anthropologists working in this field have examined technological practices such as the routinization of prenatal screening, the patenting of genes and DNA sequences, and stem cell research. They have examined the ideologies and political-economic imperatives driving national and international genomics ventures, the return of race to genetics, public enthusiasm for genetic explanations of disease and disorder, and ways in which genetic technologies and practices are reshaping notions of self, family, kinship, citizenship, and what it means to be human. And they have elaborated on how individual patients receiving genetic information and diagnoses interpret this information and bring to these encounters their own assumptions about inheritance.

One of the significant theoretical contributions of this literature has occurred at the intersection with the anthropological literature on

citizenship: the incorporation of "citizenship" constructs to describe new social formations, identity-practices, categories of the at-risk, and rights claims upon the state. We can see this hybridization in the work of medical anthropologists Adriana Petryna (2002, 2004) on biological citizenship, Vin-kim Nguyen (2005) on therapeutic citizenship, and João Biehl (2004b) on biomedical citizenship. Medical anthropologists Deborah Heath, Karen-Sue Taussig, and Rayna Rapp, who have been studying the impact of the HGP, have also introduced the organizing construct of genetic citizenship (Heath et al 2004; Rapp 2002; Rapp et al 2006; Taussig et al 2003) into their work. They use this construct to describe how genetic activists have built alliances with scientists and politicians to secure money and access to genetic research agendas. These anthropologists have argued that patient and genetics advocates have also inserted themselves into scientific knowledge-production by creating their own repositories of family medical histories and tissue samples and controlling access to them. In Chapter 3, I review their work and identify some concerns with their arguments about genetic citizenship. I also introduce Lawrence Cohen's (2004) "bioavailability" construct, which I use later in the dissertation to argue that the drive for bioavailable citizens by both the state and genetic advocacy organizations keeps genetic discrimination at the front and centre of policy discussions about genomics.

In Part One, "Roots of Concern", I present the findings of my archival research on, and interviews with, the actors who were instrumental in shaping of public awareness about genetic discrimination from 1970 to 2003. In Chapter 4, "Entry Points and Early Warnings (1970 – 1992)," I

draw on oral histories with six individuals who were the first to identify and shape public understanding of genetic discrimination: Jonathan Beckwith, Phil Bereano, Paul Billings, Neil Holtzman, Sheldon Krimsky, and Troy Duster. I describe when these actors first became concerned about discrimination as a consequence of genetic testing serious problem for the country, and what events prompted their concerns. I also identify the networks and organizations to which they belonged, and that influenced their concerns. In Chapter 5, "Naming and Framing the Problem (1992 – 2003)," I identify the second wave of actors who adopted the now-defined problem of genetic discrimination as their cause and took the problem in a different direction. I pinpoint events and practices that shaped their interests and concerns.

In Part Two, "Building a Genomic Nation," I turn to my ethnographic findings and theoretical analysis. In this section of my dissertation, I present the results of my ethnographic study of the SACGHS hearings, and situate my findings in a political-economic framework of genomics. To outline this framework, I draw on the work of political scientist Rodney Loeppky (2005) and anthropologist Kaushik Sunder Rajan (2005, 2006). Loeppky argues that the rationale for the HGP was to position the state as the architect of capital accumulation through technology transfer. Sunder Rajan extends Loeppky's political-economic analysis of the HGP to examine how textile workers in Mumbai, India are being configured as experimental and research subjects in producing biocapital, as opposed to American subjects, whom he describes as "sovereign consumers."

In Chapter 6, "Genomics Meets the U.S. Health Care System," I

describe the ways in which genomics research programmes have developed their own qualities in different national settings. I examine the political-economic origins of the HGP and review two cases of the localization of genomics outside of the United States (in France and Iceland) to highlight what is unique about genomics in the United States. I then look at how genomics in the United States has developed in a national direction.

In Chapter 7, "Legitimizing the Problem: Genetic Discrimination at the SACGHS Hearings (2003-2005)," and Chapter 8, "Voices of Discrimination at the SACGHS Hearings (2003-2005)," I present the findings from my ethnographic study of the SACGHS hearings. The hearings were a national arena at which federal scientists outlined a national imaginary of the United States as a scientific and technological innovator, with a sophisticated plan to intervene and prevent diseases of aging. Americans fit into this national imaginary as rational consumers of genetic testing, but vulnerable in specific ways. These scientists identified genetic discrimination as a central concern to the progress of the nation, and urged Committee members to legitimize the problem of genetic discrimination.[5] The Committee responded by soliciting testimony from the public of their experiences of genetic discrimination. In Chapter 7, I show how the Committee arrived at its decision to solicit public testimony, and that its actions set its treatment of genetic discrimination apart from any other issue it addressed. In Chapter 8, I present the testimonies of the seven victims of genetic discrimination who travelled to the hearings, and identify the key themes in their narratives. In testifying about genetic discrimination, they also negotiated genomic citizenship, by making claims

on the state to expand their civil rights, and by tacitly identifying their duties to themselves and to the nation.

In Chapter 9 ("Genomic Citizenship"), I offer a reflexive account of the testimonies of the seven victims of genetic assumptions who appeared at the SACGHS hearings, to draw out the assumptions they hold about genetic testing and their own duties, to themselves and the nation. I argue that these seven, in representing themselves as moral pioneers who have made sacrifices to prevent disease for themselves and future generations, are model citizens in an emergent genomic nation. I introduce the construct "genomic citizenship" to describe the claims of some Americans for expanding the civil rights of Americans—along with increasing their duties to practice self-surveillance and to participate in genomic research. The form of biosociality that underlies genomic citizenship is an inclusive identity of the flawed genome, which some genetic advocates have imposed on all Americans. By using the construct of genomic citizenship to describe these negotiations, rather than that of genetic citizenship (which was developed by medical anthropologists Rayna Rapp, Karen-Sue Taussig and Deborah Heath), I bring attention back to the role of state scientists and agencies in engineering a genomic nation. In this ideal genomic nation, Americans embrace their duties to prevent disease by undergoing predictive genetic testing, and to make their bodies and medical histories available for genomic research, while genomic industry flourishes.

I conclude in Chapter 10 by revisiting this genealogy of genetic discrimination and asking again what makes genetic discrimination a distinctly American problem. I also examine some implications of a

nascent genomic citizenship and identify them as directions for future research. Calls for legislation banning genetic discrimination appeal to an American ethos of egalitarianism while valorizing prudential Americans who embrace genomic testing and accept full responsibility for their latent genetic liabilities. I argue that this moral and political economy of hope (Novas 2006) hides the contentious ethical politics of abortion, disability, and eugenics that construct personal and societal choices to use genetic testing.

I outline and discuss my methods and their limitations in two places in this dissertation. In Chapter 2, I describe my research design and how events structured my choices of data. In Appendix A ("Data Collection and Analysis"), I describe my data in detail and the methods I used for data analysis.

Finally, a note about representation: Francis Collins is the person to whom I refer most often throughout my dissertation. Collins was Director of the NHCGR from 1993 to 1997. He then became Director of its successor, the NHGRI, from 1997 until his resignation in May 28, 2008, one week after the passage of GINA. In August 2009, he was appointed Director of the NIH. While I was conducting fieldwork for this dissertation and during most of my dissertation writing, Collins was still Director of the NHGRI. I decided not to alter my references to him in this dissertation to read "former Director." His name appears in so many places in this document that I believe this representation would have been confusing and cumbersome. Instead, I have chosen to refer to him in the present tense throughout the dissertation, as "Director." This reference reflects

not only his position during my fieldwork and most of my writing. It also reflects his position up until the passage of GINA, the event that "bookends" my chronicle of how and why genetic discrimination has been shaped into a particular kind of problem in the United States.

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[1] By "discourses," I mean all public-domain speech meant to persuade: written and oral conversations and arguments, press releases, web site statements, media communications (e.g. interviews, newspaper and magazine articles, television and radio talk shows), and other publications.

[2] Throughout, I use the colloquial expression "Americans" to refer to residents of the United States.

[3] Prenatal screening and testing are the most common forms of adult genetic testing.

[4] I use the term "the state" to indicate a complex set of institutionalized power relations at the federal level that wield tremendous power over the everyday lives of citizens. I explicitly reject a model of "the state" as a singular entity. Instead, I have in mind the many disparate individual and organizational federal actors (for example, institutes, agencies and departments, officials, legislators, senators, and executive officers) who enact competing, even conflicting, agendas, but sometimes work together. Where possible, in this dissertation I refer to the specific institutes or agencies in question (for example, the NHGRI, the NIH, the DoE, and so on), rather than generalize these actors as "the state." Where I resort to using the expression "the state" is in describing political-economic and legislative decisions in the nation's interest that have the support of many of these actors. These decisions give the appearance that there is a singular entity called "the state" operating. For example, the Congressional decision in 1988 to fund both the DoE and the NIH to lead a joint Human Genome Initiative is what I would call a "state" decision, because it had the support of a majority of relevant actors (Congressional Representatives and Senators, the DoE, the NIH, and other agencies), and because it was a political and economic commitment to enhance the nation's international competitiveness. However, as HGP chroniclers Robert Cook-Deegan (1994) and Rodney Loeppky (2005) have amply demonstrated, this "decision" had a tortured history, and was marked by contention, controversy, and failure. Finally, I note that this model of "the state" is specific to the United States. It is not generalizable to other nations, based as it is on my fieldwork in the United States, my observations of how these actors operate, and the origins of the HGP.

[5] I use the terms "SACGHS" and "the Committee" interchangeably throughout the dissertation.

INTRODUCTION

In this chapter, I describe my entry into the field, why I chose to study a social problem of the United States, the turns that my project took, and why I introduced "citizenship" as an analytical theme. I also outline the limitations of my research design, and discuss the methodological challenges of studying elites: their discourses, practices, and institutions.

The goal of my initial research design was to investigate the growing direct-to-consumer (DTC) genetic testing industry in the United States. I wanted to understand why consumers were purchasing these services, what commodities this industry was producing from consumers' tissue samples, and what material practices made it possible to produce these commodities. After I conducted preliminary fieldwork on this industry while based in Chicago, my project acquired a new dimension, an exploration of subject formation. I wanted to understand how federal scientists and policy-makers were representing (or imagining) Americans as consumers of genetic testing and personalized medicine, and how these representations were shaping policy recommendations about the regulation of DTC testing practices. My focus shifted again when I decided to make the hearings of the Secretary's Advisory Committee on Genetics, Health and Society (SACGHS) my primary field site, rather than the DTC companies. Once I began reviewing the SACGHS hearings, my research project changed altogether, to the problem of genetic discrimination.

My change in research project occurred because at the SACGHS hearings, I witnessed a form of genetic activism that had not been

described by medical anthropologists. This was activism to pass federal nondiscrimination legislation. Moreover, the activism was primarily by elite actors, rather than grassroots actors. Although medical anthropologists Deborah Heath, Karen-Sue Taussig and Rayna Rapp (Heath et al 2004; Rapp et al 2006, Taussig et al 2003) have studied the forms and impact of activism by genetic advocacy organizations and patient support groups in their innovative work on "genetic citizenship," they have not focussed on the elite activism around genetic discrimination that I witnessed at the SACGHS hearings. This genetic activism was not the empowering network-building and exercise of lay expertise by patient organizations advocating for families with rare, single-gene disorders, which these anthropologists observed in their fieldwork. Rather, the genetic activism I witnessed at the hearings by federal scientists, genetic advocates, and some industry actors (for example, Myriad Genetics), was directed at persuading Congress to pass a federal nondiscrimination law that would protect all Americans from discrimination by insurers and employers.

In the testimonies and presentations of these actors at the hearings (but also outside of the hearings), there was an implied biosociality that turned on the framing language that some of these actors used. This framing language was what I call "the language of genetic defect." The message that it conveyed was that all Americans were at risk of experiencing discrimination because all Americans had genetic flaws. Thus, the implied biosociality in the hearings was not the voluntary biosociality of affliction that Heath, Taussig and Rapp had described in their research on patient support groups. It was a biosociality that has

implications for all Americans to think and act genetically. These observations were what prompted me to make genetic discrimination, and how it has been shaped into a particular kind of problem, the focus of my research.

FINDING THE PROBLEM

Entering the Field with One Project in Mind (October 2002- September 2003)

How does a study of the DTC genetic testing industry in the United States turn into an account of how genetic discrimination has been shaped into a prominent policy and civil rights problem? In the fall of 2002, I moved to Chicago for eleven months of fieldwork to study how the DTC consumer DNA profiling industry produced commodities from tissue samples.[1] Eight months later, in June 2003, my attention had moved away from the production of bodily commodities, and towards the production of discourse by participants attending public hearings sponsored by a federal advisory body. These were the hearings of the Secretary's Advisory Committee on Genetics, Health and Society (SACGHS). SACGHS is a federal policy body that has been chartered by the Department of Health and Human Services (DHHS) to identify barriers to integrating the discoveries of the HGP (and future genomics research) into the health care system.

One of the tasks that Committee members identified as a priority was the regulatory oversight of the DTC genetic testing industry. Although I had intended to use the SACGHS hearings to understand the industry's status and its relationship with federal genomics agencies, I quickly

discovered that the Committee was devoting less time to the industry than I had anticipated, despite it having identified this as a priority concern. Genetic discrimination, which was not on my radar as an issue, came into view at a session held by SACGHS on October 18th, 2004, called "Perspectives on Genetic Discrimination."[2] At this session, SACGHS arranged for seven Americans to testify about their experiences of discrimination.

Finding Another: Genetic Discrimination and the Vulnerable American

I did not anticipate to what extent the problem of genetic discrimination would become a focal point for the work of SACGHS. Nor did I expect to hear participants characterize genetic discrimination as the most significant civil rights problem since segregation in the 1950s and 1960s. Initially, I was attuned to how participants represented Americans as a single kind of subject: a smart, savvy, and educated consumer. Participants imagined Americans as enthusiastic consumers of personalized medicine, eager to learn about their potential risks for future illnesses, and willing to participate in genomics research and clinical trials. I did hear some departures from this representation during my fieldwork at the hearings. For example, I heard some Committee members and one scientist challenge the idea that Americans had the abilities to make sound choices for themselves in the DTC marketplace for genetic testing services. I also listened to Committee members express their concerns about the low levels of scientific literacy amongst Americans. But I did not hear Committee members or participants challenge the idea that Americans would be (or could be) anything but enthusiastic consumers of

personalized genomic medicine.

Participants did represent Americans as vulnerable subjects, but again, in very specific ways. At an early stage of my fieldwork at the hearings, I had configured in my mind several categories of vulnerable Americans that I thought would appear, either as attendees, or in the discussions. These vulnerable Americans of my own imagination would require protection or special measures to ensure their participation in a genomic nation: the uninsured; the scientifically illiterate and/or poorly educated; those isolated and lacking access to health services; and visible minorities, particularly African-Americans, who shoulder the burden of health disparities in the United States.

In fact, some of these vulnerable subjects did make appearances in the hearings discourse. For example, some participants criticised unscrupulous vendors of medical goods and services for marketing junk science to American consumers, an image that reinforced the ever-present subjectivity of Americans as eager consumers of self-knowledge and risk assessment in the pursuit of good health (Crawford 2006). The scientific literacy of children and young adults received special attention from those who felt strongly about education. One person made an impassioned plea for national health insurance. (This was met with silence). Yet the key vulnerability that presided over the hearings from 2003 to 2005 was Americans' shared vulnerability to genetic discrimination.

What struck me in particular about this constitution of Americans as vulnerable subjects was the broad consensus that genetic discrimination had become a serious civil rights problem that warranted a new federal

law. This consensus seemingly effaced concerns expressed at the hearings about the uninsured, the medically underserved, and visible minorities, who shoulder the burden of health disparities in the United States. I observed widespread agreement with the argument that every American, regardless of colour, size, shape, origin, or ability, was vulnerable to genetic discrimination. What threat was this, I wondered, to have unified all Americans as potential victims of discrimination based on to invisible markers of defect?

As I began investigating the problem of genetic discrimination beyond the SACGHS hearings, I learned that passionate debate about this problem was not an artifact of the hearings. It also was taking place outside of the hearings. Moreover, this debate had preceded the SACGHS hearings by more than a decade. Genetic discrimination was a problem that had been on the radar of the NIH since the early 1990s. Political collaboration and coalition-building between the National Breast Cancer Coalition and federal agencies was evident in 1993. So what were the roots of this problem? And how had this problem changed over time? No one had told this story, I thought, leaving the impression that the problem of genetic discrimination had always been self-evident, while it had simultaneously "come from nowhere." By November 2006, with the decision to conduct archival research on genetic screening practices since the 1960s, and oral histories with actors from the 1980s and early 1990s, the last component of my project fell into place. What had been initially an investigation of the regulation of the DTC genetic testing industry became a genealogical account of the problem of genetic discrimination.

This decision brought back into my view a subject that had been the driving interest of my research into the DTC: the body. When I had expanded my study of the DTC genetic testing industry by turning to the SACGHS hearings, I had abandoned the commodification of the body as an organizing theme for my dissertation. But when I looked at the SACGHS hearings as part of the exercise of biopower, I again saw bodies: the bodies of Americans carrying genetic predispositions to disease, and needing powerful, predictive medicine tailored to their individual genomic profiles; the bodies of Americans as material resources for genomic researchers to conduct studies and pharmaceutical companies to conduct clinical trials; and the bodies of Americans as resources for profit, prestige, and international competitiveness. With this shift in perception, the body re-entered my analysis, aided by the addition of Lawrence Cohen's (2004) bioavailability construct. (I discuss this construct and my use of it in Chapter 3).

GOOD CITIZENSHIP IN AMERICA

I did not go to the hearings intending to explore how good citizenship is being constituted in the United States today. That is, I did not set out to find out what actors at the hearings thought about the rights and duties of Americans as potential users of personalized medicine, or what made for a virtuous citizen three years after the completed sequencing of the human genome and sixteen years after the inauguration of the Human Genome Project. When I first came to the SACGHS hearings, it was to understand what regulatory concerns federal policy-makers identified with the growing DTC genetic testing industry, and how they were trying to

reconcile this "rogue" industry with the "sanctioned" enterprise of genomics.[3] The idea of using citizenship as an analytical theme for the dissertation arose two years after I had watched a remarkable set of deliberations on genetic discrimination, and long after I had realized that these deliberations had been the dramatic focal point of this "nation's conversation with itself" (Ricci 2004:5). (I discussion Ricci's work and how I draw on it in Chapter 6).

The "Perspectives on Genetic Discrimination" session that SACGHS held on October 18th, 2004, made it apparent that the problem of genetic discrimination had to be situated in the state's commitment to building a genomic nation. The claims that these seven individuals made about genetic testing, and their rights and duties as Americans, were not simply the concerns of seven individuals. They were statements by model citizens about their responsibilities to think and act genetically, given the discoveries of the HGP, and the promises by the NHGRI that it would deliver personalized medicine.

The anthropological literature on citizenship turned out to be very useful in understanding what these individuals were articulating and what work it was doing. I found especially useful the work of policy anthropologist Phyllis Chock (1991, 1994, 1995, 1996, 1999) on the discursive construction of illegal aliens and good citizenship in Congressional hearings and media reports on immigration, and Aihwa Ong's (1996, 2003) on the efforts of Asian immigrants to achieve full cultural citizenship in the United States. My reading in this literature of citizenship, particularly on immigration debates and processes, shaped my

thinking about what I witnessed at the hearings.[4] Specifically, I began to think about the claims that participants made about the need for a federal law banning genetic discrimination as part of a process of assimilating Americans into an emergent genomic nation, and training them to think and act genetically.

Yet why should questions about what constitutes good citizenship in a genomic nation be relevant to a nation that has engaged in a prolonged period of self-examination about national security, borders, immigration policies, civil liberties, and patriotism, following the terrorist attacks of September 11, 2001? Surely "homeland security" is a more pressing civic concern than the rights and duties of Americans as potential consumers of personalized medicine. Why not simply ask individual Americans what, if anything, it means to be a good citizen in an age of genomic research and medicine, rather than listen on a set of hearings that were an extended national conversation among elites (see Ricci 2004)? (I define "elites" later in the chapter).

Claims about what constitutes good citizenship might seem to be outside the scope of the SACGHS hearings. Yet at the SACGHS hearings, I listened to participants outline their visions of the United States as a nation that would offer consumers predictive tools to prevent and control diseases such as cancers and mental illnesses. I watched participants at the hearings shape a shared understanding of how Americans would participate in a society that defined itself through its biotechnological prowess.

What I heard some participants at these hearings articulate, and what

the committee produced through its deliberations, were *norms* about Americans as genomic citizens. Although these norms filtered through many discussions during the hearings, such as those on DTC genetic testing, the genetic training of medical professionals, and the coverage and reimbursement of genetic tests by insurance companies, for example, these norms were most starkly articulated during deliberations about genetic discrimination. Claims about the rights and duties of Americans are part of what sociologists Nikolas Rose and Carlos Novas (2005) call a citizenship project, where Americans were defining the nation and themselves in biological terms. The SACGHS hearings were a site for "making up citizens" (Rose and Novas 2005; Rose 2007), in that citizens made claims on the state and lawmakers as victims of discrimination. To represent the elements of citizen-making that I witnessed at the hearings, I draw on political scientist David M. Ricci's (2004) model of public transcripts as well-publicized opinions about citizenship norms (see Chapter 6).

RESEARCH DESIGN AND LIMITATIONS

In this section, I describe my research design and its limitations, including my decision to make the SACGHS hearings my primary field site, and the ways in which time limitations affected my data gathering decisions.

This account of genetic discrimination is based on three stages of multi-sited research that I conducted over five years. The first stage consisted of eleven months' of fieldwork and participant-observation in Chicago from October 2002 through September 2003 while I was a

visiting scholar at the American Bar Foundation (ABF) and Northwestern University. There, I conducted the fieldwork on the consumer DNA profiling industry. This research included an inventory of the testing services offered for sale by approximately fifty companies nationwide and the tissue collection tools and methods they used; and an analysis of the claims that these companies made about the commodities they produced, and the utility and accuracy of their testing results. The participation-observation component was on both the health care system, and the material culture of health goods and services for sale outside of the health care system.

The second stage was ethnographic fieldwork that I conducted at the SACGHS hearings, from 2005 to 2007, in the Washington, DC area, and in Montreal. I initially focussed on the regulation of the DTC genetic testing industry at the hearings, but by June of 2005, I had turned my attention towards genetic discrimination. I conducted a discourse analysis of the data I collected from these hearings (see Appendix A).

The third stage consisted of twelve months' of archival research and oral histories that I conducted from January 2007 through May 2008, in four cities (Montreal, Baltimore, San Francisco, and Washington, DC). I conducted oral histories with some of the first actors to define the problem and shape public awareness of genetic discrimination. I draw heavily on my interviews with Neil Holtzman, Paul Billings, and Troy Duster because these men offered detailed accounts of their roles in shaping and framing the problem. My reliance on their interviews also reflects a bias in data collection: I conducted these oral histories in person. Consequently, their

reflections took on a more explicitly narrative form.[5] For these reasons, their comments dominate my account in Chapters 4 and 5.

I also conducted two types of archival research. I examined media accounts of stigmatization and discrimination from the 1960s onwards to understand when and how concerns about something that could be called "genetic" discrimination penetrated public consciousness. I also looked at clinical genetics and bioethics journals from the 1960s to understand how this debate unfolded among scientists and biomedical professionals. I present my findings from this archival research in Part One of the dissertation.

The SACGHS Hearings as a Field Site

SACGHS came to my attention in 2003 while I was a visiting fellow at the ABF in Chicago, trying to gain access to DNA profiling companies that offered DTC services. Although I had chosen a timely and interesting problem, I quickly discovered its limitations as a dissertation research project. My discovery of the SACGHS hearings came through the divine intervention of Robert Dingwall, a medical sociologist who directs the University of Nottingham's Institute for Science and Society. Dingwall also spent the 2002-2003 year as a visiting fellow at the ABF. During a presentation of my research at its seminar series, he asked me how the commercial DNA profiling industry was regulated. I told him, and the rest of my audience, that I had no idea. With great tact and kindness, he suggested that this was something that I really ought to know. This simple—or so I thought—question prompted me to look at regulatory practices and debates around genetic testing in the United States. This

inquiry in turn led me to discover the SACGHS hearings, which had just started. I learned that this committee would be deliberating on the regulatory gaps with DTC genetic testing. As other countries, particularly Australia and the UK, were also engaged in policy hearings on the regulation of the direct-access testing industry, I decided it would be useful to do a comparative study of the regulation of this industry. When I reviewed the transcript and webcast from its first hearing (June 2003), I realized that these hearings were opening up questions beyond consumption, and they were rich enough for me to focus entirely on the United States. With that decision, SACGHS became my primary field site, and the scope of my research expanded.

The SACGHS hearings turned out to be a productive site for many reasons. Since 1999, two federal advisory committees have held hearings on ethical, legal, medical and regulatory issues associated with the growth of clinical and commercial genetic testing practices: the Secretary's Advisory Committee on Genetic Testing (SACGT), and SACGHS.[6] Picking up where its predecessor SACGT left off (and reproducing some of its work), SACGHS was given a mandate by the DHHS to advise its Secretary on the key challenges to integrating genetic testing practices and genomic medicine into the U.S. health care system. So the SACGHS hearings have been the longest-running public arena for discussion of current and future challenges associated with genetic testing and genomic medicine.

Another reason the hearings were a productive site for research was because of their premise of facilitating deliberative democracy on genetics, genomics, and the health care system. SACGHS reports directly to the

Secretary of the DHHS. In other words, it is a committee with some influence. The hearings drew attendance from a cross-section of organizations and individuals with an interest in genomics, and drew representatives of many federal agencies and departments. For example, the non-governmental actors who attended the hearings included representatives from genomics and pharmaceutical companies, genomics researchers, attorneys, medical professional organizations (particularly genetic counsellors and nurses), and genetic advocacy organizations. The premise of deliberative democracy was challenged by the absence of actors who did not or could not attend or participate, thus contradicting a perception that the hearings were truly "public."[7] Even so, the spectrum of actors that attended the SACGHS meetings was unrivalled in its broadness by any other public or private forum taking place at the same time. Had I chosen any other site for my fieldwork, I would have overlooked the diversity of actors interested in the public policy of genomics. My fieldwork at the SACGHS hearings challenged my perception, based on research in medical anthropology and sociology (see, for example, Epstein 1996; Heath et al 2004), that laypersons and health activists exert considerable power and influence in making science policy. The SACGHS hearings suggested to me that power and influence are not as accessible to non-elites as these researchers have indicated. What I observed at the hearings is that power is concentrated in the hands of a few individuals with charisma, political connections, or both.

Thus, the SACGHS hearings were an arena for an array of actors and coalitions that constitute contemporary genomic politics to present their agendas and pursue their interests. What is different about the arena of

genomics from other forms of health politics is that genomics is a technological and economic enterprise that is highly differentiated, with many technology providers. Genomics companies include the many new genetic diagnostic firms and clinical laboratories that have sprung up over the last ten years, venture capital firms that bankroll the diagnostic companies, information technology and genetic informatics firms, and manufacturers of genomics technologies.

A third factor that made the SACGHS hearings a valuable choice for conducting fieldwork was logistics. Because SACGHS is a federal advisory body, its operations are regulated by the Federal Advisory Committee Act (FACA) of 1972. FACA was passed to enhance transparency and public access in the public policy process. Consequently, all of the advertised hearings of the Committee were open to the public. Hearings dates, locations, and agendas were advertised in advance in the *Federal Register*, the federal government's daily published bulletin of notices by committees and rules, and on the Committee's own website. All of the hearings were webcast live, and webcasts were archived afterwards on the Committee's website, along with transcripts of the proceedings and PowerPoint presentations delivered by participants. (The Committee also made draft documents available to the public for commentary). This transparency allowed me to listen to and watch the proceedings of the hearings through on-site observation, by watching the webcasts, and by reading the transcripts of the hearings and supporting documents. The availability of these many mediums was beneficial, because it allowed me to cross-reference my notes from the webcasts and on-site observation with the hearings transcripts to verify my findings and analysis.

Perhaps the most valuable aspect of attending the public hearings was the access they provided to participants and attendees. At every hearing of SACGHS, I had opportunities to introduce myself to anyone there. No one refused to talk with me. In fact, everyone I spoke to expressed an interest in my research. I spoke with SACGHS committee members and support staff, consultants, company representatives (from diagnostic laboratories, pharmaceutical companies, and genomics diagnostics firms), postdoctoral researchers, and genetic activists. Exchanges that I expected to be cursory often turned into longer conversations. Some participants used the opportunity to vent about the proceedings. Others corrected my assumptions about the reach of the hearings and their impact. These comments challenged my own observations, provided valuable information that I could not otherwise obtain, and opened up new lines of inquiry for me.

Why the United States?

This discussion of the SACGHS hearings raises a broader question: why conduct an institutional ethnography of genetic discrimination? And why choose the United States? Despite the increasing legitimacy of conducting anthropological fieldwork in the United States, by and large, anthropology students still tend to choose field sites outside of North America—and to "study down" (cf. Nader 1974). An abiding interest in American politics, particularly the contentious character of reproductive politics, has made the country a compelling choice for me. Other features that contribute to its interest to me are the scope and dynamism of social movements; the reach of legal institutions and practices; the ascendancy and influence of

Christian fundamentalism over the last twenty years; and the concentration of wealth and power.

My choice of the United States has also been influenced by Laura Nader's admonition (1974:292-293) to anthropologists to "study up," by examining elite institutions and practices, particularly within the United States. Writing more than thirty years ago, Nader provided a clear rationale for studying the processes of power in the United States. "A reinvented anthropology should study powerful institutions and bureaucratic organizations in the United States, for such institutions and their network systems affect our lives and also affect the lives of people that anthropologists have traditionally studied all around the world," she wrote. Although anthropologists have been devoting more attention to institutions and organizations in the United States in recent years (Forsythe 1999), anthropologists still tend to leave the study of elite institutions and practices in the United States to sociologists, in an unspoken disciplinary division of labour.

Despite its decline across multiple measures of "superpower" status (for example, annual military expenditures, military deployment, export and creditor status, economic growth, absolute size of the economy),[8] the United States is still a global centre of power and commerce. Few countries can afford to ignore policy decisions and legislation in the United States, or its initiatives. While many nations have committed themselves to a programme of genomic research and medicine, we should understand genomics in the United States as a post-Cold War, nation-building initiative of technology transfer and capital accumulation (Loeppky 2005).

At first glance, the United States does not seem to qualify for a spot in the "nation-building" camp, or even a candidate for being seen as practicing national science and medical modernization. Anthropologists interested in nationalism, citizenship, and self-making have focussed on post-Communist states, post-civil war and Cold War African nations, or countries transitioning from dictatorship and military rule to constitutional democracy. But the country's economic and political decline over the past twenty-five years has provided an important impetus for a commitment to new large-scale science projects (Loeppky 2005), as well as a renewed commitment to militarization in the interests of nation-building (Masco 2008). (I discuss genomics as a nation-building project in Chapters 3 and 5).

Defining Elites: Towards a Theory of Elites in Anthropology

Throughout the dissertation, I describe participants at the hearings as "elites." What do I mean by this? Sociologists have been theorizing about elites for over a century, starting with the work of Italian economists Vilfredo Pareto (1902[1976]) and Gaetano Mosca (1896) and German sociologist Robert Michels (1915) on ruling elites, and moving through C. Wright Mills (1956) on the institutional origins of power elites in the United States and George William Domhoff (1967) on the American business aristocracy. Over the last forty years, sociologist have developed pluralist accounts of elites, producing studies of how strategic elites (also known as experts) are replacing ruling classes (Keller 1963), the corporate elite (Useem 1980), and minority access to power (e.g. Zweigenhaft and Domhoff 1998), to name a few areas of inquiry.

Despite Nader's encouragement in 1974 to anthropologists to study what she called vertical slices of power, anthropologists have theorized very little about elites (Gusterson 2001a). Anthropologist Hugh Gusterson (2001a) attributes this neglect to the traditional division of labour between anthropologists and sociologists, in which the former focus on the marginal, ceding study of elite power relations, institutions, practices, and individuals to the latter. A representative theory of elites in anthropology drawing from the study of peasant societies can be found in James C. Scott's (1990) model of power relations, whereby elites form a clearly-bounded "dominant" group that is easily distinguishable from "subordinate" groups. This model, however, will not do to represent power relations in complex societies, where membership in a category of elites is more fluid and pluralistic.

Anthropologist George Marcus picked up the baton in 1983 by publishing a collection of essays and ethnographic case studies of elites. Anthropological studies of elites lagged until the 1990s, when anthropologists turned their attention to weapons scientists and militarization in the United States (Gusterson 2001b, Lutz 2002, Masco 2004, 2008), high-energy physicists (Traweek 1988), transnational entrepreneurs, (Ong 1999), and bureaucrats (Riles 2000, 2004). The anthropology of public policy, an outgrowth of British organizational anthropology, has also contributed to an anthropology of elites (see, for example, Heyman 2004; Marshall 1984; Shore 2002; Wedel et al 2005).[9] However, despite this encouraging growth, there is not a comparable theoretical discourse within anthropology about elites.

One of the problems with using a term like "elites" to characterize the actors who have shaped public understanding of genetic discrimination is that it has a self-evidentiary appeal. "I know one when I see one" comes to mind. On each of my fieldwork trips to the SACGHS hearings, I came away with the impression that every participant was an elite simply because he or she had the resources to attend hearings for two days in the Washington, DC area. (Whether attendees used their access to the hearings to influence public policy is another question.) Gusterson's (2001:4417) description of elites from the standpoint of anthropological access seems germane: "Elites are busy, important people who have the power to elude or obstruct anthropologists, and whose lives are usually unsuited to the anthropologist's classic (and time-intensive) technique of investigation—participant observation." All of the people who attended the hearings seemed to be important people with professional credentials. Anthropologist Catherine Marshall's (1984:236) definition of elites as "[p]eople in high positions" also fits the bill here.

But this contingent understanding of elites is problematic. By "elites," do I simply mean "professionals?" Could we call individuals who attended the hearings but did not seek an audience with decision-makers elites? Would a homeless person with no professional identity or status be considered an elite if she or he attended the SACGHS hearings and testified to the committee? What about the committee members and the ex-officio members (the individuals who represented government agencies and departments at the hearings)? Were they elites in their working lives beyond the hearings? Was I an elite?

Surely the elite status is more stable and less contingent. I also had to ask myself whether there is value in distinguishing between power elites, who have privileged access to decision-makers, and scientific elites, who have privileged access to scientific knowledge (these were, after all, hearings on genomics). Both, it seemed to me, were important. Some participants—the most influential and powerful ones, I thought—could claim both.

In the end, I decided that what counted for the elite status was having ongoing access to decision-makers outside of the hearings *and* having scientific expertise. Elites at the SACGHS hearings were those individuals who were positioned to influence public policy because of ongoing access they had to decision-makers through their professional or personal networks, and because of their scientific expertise. (This definition excluded me from elite status). By decision-makers, I mean a small group of extremely powerful people: legislators (Congressional representatives and Senators); federal officials such as Tommy Thompson and Michael Leavitt, the two heads of the Department of Health and Human Services from 2002 to 2006; and the president and his advisors. My definition of elites included some committee members, ex-officio committee members, and experts called upon to make presentations to the committee. (I discuss the constitution of the committee and the identities of public participants in Chapter 6.) Other elites were lobbyists, executives of health and genetic advocacy organizations, industry representatives with connections to the NHGRI or other agencies, and medical professional bodies.

Yet even with a formal definition of elites, determining who is or is not

an elite is often difficult, because power relations are largely invisible to outsiders. With some important exceptions, despite knowing their professional identities and affiliations, I could not know which of the participants at the hearings wielded power and influence (how much or what sort) in genomics decision-making beyond the hearings. Individuals who testified to the Committee, particularly the many representatives of biotechnology firms, practiced impression management (Goffman 1959) by credentialing themselves as experts, or dropping names to suggest they might be influential and well-connected. At the other end of the spectrum, some individuals who attended the hearings whom I knew to be influential did not advertise their credentials or connections. They might not strike an outsider to genomic politics as elites.

One example of the latter is Sharon Terry, who is president and CEO of the umbrella organization Genetic Alliance, and chair of the Coalition for Genetic Fairness, the two largest and most powerful genetic advocacy organizations in the country. In her public presentations, at the SACGHS hearings and other venues, she occasionally adopts the persona of an ordinary American by emphasizing her status as mother of two children. Even if Terry did not hold these positions, I would still consider her to be an elite because of her status as a scientific insider or lay expert (Novas 2006; see also Epstein 1995, 1996) and her close ties to the NHGRI. But not everyone who attended these hearings would have been aware of her insider status.

Access to Elites

Because elites often are public figures, their speeches, testimony, and

written comments are often widely available. This is the main benefit of studying elites. Choosing to work with or study elites, however, presents a raft of methodological challenges. Catherine Marshall (1984:237) outlines some of these:

My experience confirms Marshall's observations. Obtaining interviews with SACGHS participants and other professionals outside of the hearings was difficult; at times, it was impossible. I had anticipated this, but was not prepared for how often the non-availability of potential interviewees forced me to change course. More than half of the individuals and organizations that I contacted did not respond to my requests for interviews or visits. For example, one of the directors of the Genetic Alliance was very gracious when I introduced myself to her at a SACGHS hearing in March 2007. She expressed an interest in my project and a provisional willingness to be interviewed—but warned me that she was knee-deep in work and about to take a one-month vacation later that year. I took her business card, offered mine, and followed up with an email message asking her if she would be willing to set up an interview with me after her break. I also asked if I could drop into the Washington office of the Genetic Alliance during a visit there in April, to introduce myself to the administrative staff and pick up some of their literature. When I did not receive a reply from this director, I contacted the Genetic Alliance and asked the staff to forward my email message to her. Within days, I received a terse email response from that director. The priority of the Genetic Alliance, she wrote, was their lobbying efforts to persuade Congress to vote for GINA, not making themselves available to anthropologists who wanted to "study" them. The story does not end there. Months later, this person

contacted me and retreated somewhat from her previous position by referring me to a staff member at the Genetic Alliance. By this time, I was conducting my archival research and oral histories. I did not have the time to conduct an interview with staff at the Genetic Alliance or the resources to visit its offices. In the end, I was the one who did not make myself available to an elite.

There were some individuals who have been influential actors in the genetic discrimination public debate from whom I decided not to request interviews. For example, Francis Collins, the NHGRI Director, attended many of the hearings at which I was present. There were a few occasions when I had the opportunity to introduce myself to him and request an interview. I decided not to, because I did not believe that he would consent to being interviewed. Also, at this point in my fieldwork, I did not feel I had sufficient knowledge of my subject to conduct an interview with him. An interview with Collins, I decided, would be more valuable later in my fieldwork. As it turns out, later in my fieldwork I turned to setting up and conducting oral histories of older actors, and no longer had the time to interview the current actors, such as Francis Collins and Sharon Terry. That task would have to come under another research project.

Polymorphous Engagement

This leads to a question about how relevant, if at all, participant-observation is to ethnographic research on elite discourse, practices, and settings, and what methodologies might substitute for participant-observation. In describing the methodological challenges of gaining access to elites and elite institutions such as weapons laboratories and

companies, Hugh Gusterson (1997) suggests that it might be necessary to adopt some unusual tactics to conduct participant-0bservation:

> Participant-observation is not only difficult to achieve in elite settings, "where ethnographic access is by permission of people with careers at stake, where loitering strangers with notebooks are rarely welcome, and where potential informants are too busy to chat," it is, says Gusterson (1997:217), an inappropriate methodology for knowledge-production in a globalized world when elites are highly mobile, frequently dispersing and linking up, often electronically. As an alternative, he proposes something he calls "polymorphous engagement." This is a suite of research strategies that includes multi-sited ethnographic research, formal interviews, electronic data collection, archival research, and observations of popular culture. (Gusterson 1997:115)

I used all of these methodologies during my fieldwork (see Appendix A). I set up formal interviews (oral histories) with some of the first actors to shape public understanding of genetic discrimination, with one director of a consumer genetic testing company, and with one SACGHS staffer. I conducted informal interviews with individuals familiar with the DTC genetic testing industry and the history of health insurance in the United States. Informal conversations with approximately twenty hearings attendees, including one SACGHS committee member and a SACGHS chair, constituted an important part of my information-gathering. Electronic data collection and archival research were essential tools. I used these tools to retrieve speeches and policy statements of many actors in my account; track institutional histories and policies; review the webcasts and transcripts from all of the SACGHS hearings; retrieve all of the correspondence, testimony, policy statements and agendas from the SACGHS hearings (as well as from the hearings of its predecessor,

SACGT); and to trace the history of controversy over genetic screening practices from the 1960s to the present. Finally, observation was a key methodology I used for the SACGHS hearings. This observation took two forms: in-person, when I attended the hearings; and viewing the webcasts of the hearings, both when I could not attend them, and after I had attended them.

I also used participant-observation during my first year of fieldwork (2002-2003). Just after I arrived in Chicago in October 2002 to start my fieldwork on the consumer genetic testing industry, I became ill. My fieldwork took on an unplanned participation-observation component as I became a patient of the U.S. health care system. I experienced being treated as a "rational" consumer in my clinical encounters. My experiences as a patient in the U.S. made me aware of the transactional relationship between medical services and patients, and how insistently Americans are shaped into entrepreneurial, even actuarial, consumers in these transactions. Each time I contacted a new medical practice, I was enthusiastically welcomed by the administrative staff and my designated clinician as a potential client or even business partner who might offer long-term customer loyalty. At one point in my treatment, I discovered that as an uninsured, foreign patient, my treatment rates were negotiable to the Medicare rate, a detail not advertised by medical providers. From then on, I negotiated the Medicare rate with providers for all of my treatments. Essentially, I was rewarded for being entrepreneurial in my clinical transactions, an experience unfamiliar to me in the Canadian healthcare system.

A Canadian Conducting Fieldwork the United States

It is standard practice for doctoral students in anthropology to immerse themselves in the study of another cultural setting, usually in a country far from one's own home. Students typically live in one or more communities for a year (or longer) to generate the ethnographic field data that comprises conventional anthropological dissertations. My decision to study the contemporary discourse and politics shaping public understanding of genetic discrimination in the United States, and to conduct an institutional ethnography using the SACGHS hearings, went against the grain of typical anthropological fieldwork in many ways.

It also meant that I had to become familiar with two complex areas quite late into my PhD: the history and structure of the health care system in the United States in the twentieth and twenty-first centuries, particularly the changing nature of health insurance practices; and the history of the Human Genome Project and genomics policies. There is an argument to be made that an American, who has grown up within the U.S. health care system, would have an advantage writing about this subject purely from a technical perspective of the relationship between genetic discrimination and health insurance, over an outsider who has grown up elsewhere. I would argue that as a Canadian writing about a cultural problem of the United States, I was able to bring a unique perspective to the subject.

I grew up in a nation that is accustomed to being compared to its southern neighbour. Consequently, I have an outsider's perspective on the United States—a country that seems in so many ways similar to Canada, yet is guided by values and priorities that are distinctly different. These

values and priorities are not always evident to Americans as being specifically American. One example of such a difference is the self-representation by most Americans as middle-class—despite marked disparities in wealth and power across the populace. Another is the emphasis on consumer choice: the twin ideas that self-improvement is a universal goal for all Americans, and that all Americans have the capacity and desire to make informed choices in their consumption of medical goods and services. A third is the (mostly) unquestioned practice of framing what Canadians would call ethnic difference as racial difference—and thus treating difference as a set of biologically-determined categories that nature invented, rather than as social and historical constructs that serve political purposes (such as marking class differences).[10] It is not, for example, that discourses of health self-improvement and consumer choice are foreign to Canadians. What is remarkable—and often startling to Canadian ears—is the degree to which these values and priorities play out in national discourses in the United States. As a Canadian, I am attuned to these differences. As a social scientist, I am interested in how these values and priorities can displace—and even absorb—others.[11]

The approach that I decided upon was to write a dissertation that explores genetic discrimination as a cultural problem that has acquired a unique profile in the United States over the last thirty years, and to ask what is *specifically American* about the discourses and claims surrounding this problem. I was concerned to emphasize the theoretical work of the dissertation, and to develop the "genomic citizenship" construct from the construct of "genetic citizenship" introduced by medical anthropologists Rayna Rapp Karen-Sue Taussig and Deborah Heath. I use

the genomic citizenship construct to critique the limits of medical anthropological scholarship on genetic activism in the United States, to point out the creep of eugenics language into rights claims that insist that all Americans all equal (but genetically flawed), and to highlight what I think is a fundamentally-American insistence that scientific and technological innovation will eliminate health inequalities and deliver good health to all. It is for this reason that my dissertation on genetic discrimination is subtitled "Genealogy of an American Problem," rather than "Its Relationship to the U.S. Health Care System," "Developments since the Human Genome Project," or even "The Making of GINA."

There is a criticism to be made of my methodology and the data that it generated, that I did not conduct the sustained fieldwork (whether through participant-observation and interviews) of either SACGHS or the NHGRI required for an ethnography of genetic discrimination. Beyond attending the SACGHS hearings on six occasions, travelling to Washington, DC for short-term data collection, and conducting in-person interviews in that country, I did not return to the United States after I left Chicago in 2003 to conduct another period of sustained fieldwork. One reason was personal. We develop a research design with the hope that all will proceed as planned. But life sometimes has other plans for us. In June 2003, after I had conducted preliminary fieldwork in Chicago and was considering extending my stay for another year to begin my SACGHS fieldwork, my father died. Two months later, my mother was diagnosed with end-stage cancer. I returned to Canada, left my apartment in Montreal, and moved to my parents' house in Ontario to care for my mother and manage my parents' affairs. I remained there for the better

part of two years. Although I resumed my research in the fall of 2005, my duties to my parents continued to the end of my PhD. I travelled frequently between Ontario and Montreal during these years. This left only brief periods of time in which to travel to the United States. The other reason is financial. Fieldwork is time-consuming and, particularly in the United States, expensive. I did not have the financial resources to move back to the United States and conduct a second period of sustained fieldwork.

Promising Avenues of Research

The final project came into view in November 2006, which was late into my degree. Consequently, I did not have time to pursue some avenues of research that would have enriched my story. I reluctantly turned down an invitation from Sheldon Krimsky to use the archives of the Council on Responsible Genetics (CRG), in Cambridge, Massachusetts. Phil Bereano, a founding member of the CRG, had also mentioned the CRG archives to me in a phone interview in May 2007. The CRG played a central role in widening public awareness of genetic discrimination in the late 1980s and early 1990s, and their archives would have enriched my story.

There were many individuals and organizations that I wanted to interview, particularly those who became active in the mid-1990s. Either I did not have time to contact them or they did not respond to my queries. These include Arno Motulsky, who developed pharmacogenomics and was one of several scientists to flag discrimination as a consequence of genetic screening, in the 1970s; Richard Severo, who wrote the 1980 *New York Times* series on workplace genetic screening; Tania Simoncelli, a science

policy researcher at the ACLU; Karen Rothenberg, a legal scholar and Dean of Maryland State School of Law; and the California-based Center for Genetics and Society, an organization that has taken over from the CRG the task of keeping genetic discrimination in the public eye through a regular critique of genetic initiatives.

The voice of the health insurance industry is largely missing from my account. It is unfortunate, because this absence reinforces its role as the designated villain in the drama of genetic discrimination and the notion that the problem of genetic discrimination is largely a problem tied to the health care system. I contacted America's Health Insurance Providers (AHIP) and requested an interview (and a visit to their Washington offices). No one at AHIP responded to my request. The National Association of Insurance Commissioners (NAIC), the body that regulates the insurance industry in the United States is another organization that wrangled over the question of discrimination and health insurance in the 1980s. Members of the CRG attended their meetings to educate and lobby the NAIC, and Phil Bereano suggested that their archives might be worth exploring. However, I lacked the time to correspond with the NAIC.

Finally, I want to comment on a significant omission from this story. I wanted to investigate Shobita Parthasarathy's (2004) claim that the National Breast Cancer Coalition (NBCC) was the organization that was instrumental in reframing genetic discrimination as a civil rights problem after Myriad introduced its BRCA testing. While my own research corroborates Parthasarathy's claim that the NBCC framed genetic discrimination as a civil rights, I suspect that other organizations and

actors, including the NHGRI, helped with this framing. Because I did not conduct fieldwork with these organizations, this thread of my story, about the origins of a civil rights frame for genetic discrimination, is missing. This is a story that needs to be researched thoroughly, at another time.

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[1] My research base in Chicago was the American Bar Foundation (ABF), which is affiliated with Northwestern University.

[2] My fieldwork was interrupted from 2003 to 2005. (I say more about this towards the end of this chapter). Consequently, I was unable to attend the October 18th, 2004 SACGHS hearings. I attended the SACGHS hearings from June 2005 through November 2007. I watched those hearings that I could not attend, including the "Perspectives on Genetic Discrimination" session, by webcast. I also read the transcripts from those hearings, reviewed the written public commentary on genetic discrimination submitted to the Committee, and watched the DVD that the Committee produced from this session.

[3] This is my wording. Federal scientists and Committee members portrayed each in this manner, but did not use these words.

[4] Chock's research on the use of framing strategies by political actors at policy hearings also provided me with a valuable model for how to apply frame analysis to the SACGHS hearings. Medical anthropologists Lisa Wynn and James Trussell (2006) have also used applied frame analysis to their study of single-day federal policy hearings in the United States, to identify how elites and state actors shape public understanding of emergency contraception.

[5] My interview with Paul Billings was in two parts. We began our interview in San Francisco, where I met with him, but it was interrupted. We continued our interview by telephone. I conducted the remainder of my oral histories by telephone. My interview with Amanda Sarata, which was not an oral history, took place in person, with follow-ups by email.

[6] Although both committees emerged from the deliberations of an ELSI Task Force that was struck by the joint NIH-DOE Working Group on Ethical, Legal and Social Implications of Human Genome Research, neither are ELSI bodies. They were initiated by the Department of Health and Human Services (DHHS).

[7] Some voices were noticeably absent. One was the medical association professional bodies (the American Medical Association and the National Medical Association). Another was the direct-to-consumer industry, which stayed away from the hearings until 2008. Big Pharma, too, rarely made an appearance. Genetic counsellors and nurses specializing in genetics, on the other hand, were well-represented by their professional associations.

[8] See Fox (1944) on the origins of the superpower status of the United States.

[9] Gusterson (1997) notes that although anthropologists have begun to study capitalist elites, they have neglected military elites.

[10] For more on this, see especially Duster (2005).

[11] I am grateful to Abby Lippman for pointing this out to me.

INTRODUCTION

The dominant account of the origins of genetic discrimination, which I call the genetic privacy story, represents genetic discrimination as a problem of inadequate privacy protections that originated in the mid-1980s with the Human Genome Project (HGP). In this chapter, I identify the limitations of this story and outline how my account challenges it. I situate my account of genetic discrimination within two sets of literature: the anthropology of new genetics, which is my primary set of literature; and the anthropology of citizenship, my secondary set.

At the intersection of these two sets of literature, medical anthropologists Deborah Heath, Karen-Sue Taussig, and Rayna Rapp have introduced the construct genetic citizenship (Heath et al 2004; Rapp et al 2006, Taussig et al 2003). This construct builds on recent scholarship within medical anthropology that explores how communities and populations have turned biological claims of difference and vulnerability into rights claims on the state (see, for example, Biehl 2004b; Cataldo 2008; Petryna 2002, 2004), or on local instantiations of transnational initiatives (see Nguyen 2005). Genetic citizenship describes how genetic activists have mobilized their biosociality (Rabinow 1996) of embodied difference to forge political and economic alliances with scientists, building powerful networks within patient support groups, and controlling researcher access to valuable banks of tissue samples and family histories that they have themselves built. These new configurations of expertise, the three argue, open up democratic opportunities for marginalized Americans from communities of embodied difference (genetic patient groups and the

disabled) to work with elites and shape the directions of genomic research to their own purposes.

Using this work on genetic citizenship as my departure point in Part Two of my dissertation, I argue that the heightened concern about genetic discrimination as a civil rights issue for all Americans indexes a very different kind of emergent citizenship in the United States. I call this form of citizenship "genomic citizenship." Genomic citizenship is part of the state's effort to build a genomic nation and a political economy of hope (Novas 2006) that configures Americans as genomic consumers and research subjects. In other words, genomic citizenship confers certain responsibilities for Americans. Here I draw on Lawrence Cohen's (2004) "bioavailability" construct to argue that the drive for bioavailable citizens by both the state and genetic advocacy organizations has kept genetic discrimination at the front and centre of policy discussions about genomics.

GENETIC DISCRIMINATION AND THE GENETIC PRIVACY STORY

Over the last decade in the United States, advocates of federal nondiscrimination legislation have widely promoted two related claims about genetic discrimination. One is that all Americans are at risk of experiencing discrimination at the hands of insurers or employers because of invisible genetic markers that they cannot change or control. The other is that fear of genetic discrimination is preventing Americans from seeking genetic testing and from participating in research or clinical trials (and thereby hindering scientific progress).

For such a pervasive and significant problem, surprisingly little has been written about where genetic discrimination came from, which actors have been instrumental in shaping it and giving it a high profile, and what political interests are driving recent action on the issue. One doctoral dissertation in bioethics (Vazakas 1993) reviews antidiscrimination law in the United States, including the Americans with Disabilities Act (ADA) of 1990, and assesses whether these laws will provide protection to individuals with "atypical genotypes." Vazakas identifies some sources of genetic discrimination and social forces that have encouraged insurers and employers to use genetic information in their actuarial assessments. However, she does not offer an account of how the problem has been shaped or by whom. Nor does she situate her analysis in a political-economic framework of genomics as an ongoing commitment of the NIH and the NHGRI. What little we know about genetic discrimination comes from three scholars who have chronicled activism on genetic privacy since the 1990s (Everett 2004, 2007; Frankel 1999; Parthasarathy 2004). These scholars have produced an account that I call the genetic privacy story.

The genetic privacy story has two features. One is that concerns about genetic discrimination emerged in conjunction with the mapping and sequencing of the human genome in the early 1990s and the proliferation of susceptibility genetic testing (see also Gostin 1991, and Ostrer et al 1993). The other is that genetic discrimination is a problem of inadequate genetic privacy protections in law. Although the genetic privacy story could be called a legislative activism story because it describes efforts to pass antidiscrimination laws at the federal and state levels, I believe it is more appropriate to call it a genetic privacy story because the salient

explanatory feature is genetic privacy as a strategy or driver for passing legislation.

While this story is accurate in broad brushstrokes, it overlooks evidence that the first published commentaries abut genetic discrimination in the late 1980s and early 1990s were motivated by concerns about a disparate set of practices that emerged much earlier than the Human Genome Project. The story also overlooks how concerns and political interests unrelated to genetic privacy have shaped the problem and driven activism on it.

I devote some space to examining the three accounts. Each author covers essential historical ground by identifying important actors, practices, and events. Each helps to clarify what many perceive to be the problem of genetic discrimination. And each points out limitations with our current understanding about what kind of problem genetic discrimination is and where it came from. Despite their efforts, I argue in this chapter that thinking about genetic discrimination as a genetic privacy problem limits us. It obscures our understanding of when the first concerns emerged (and in what context), and which concerns are driving the issue today. The problem of genetic discrimination is today a moving target, still being legitimized in the public sphere, its scope still being shaped, and too complex to be captured by the privacy story. To understand where the problem came from and why it has acquired its current stature as a pressing problem for the nation, we need to locate the issue in both a longer history and a broader context.

But first, I briefly review some of the ways in which insurers and

employer in the United States can access medical records, taken genetic tests, or require that individuals share results from genetic tests with them.

THE ACTUARIAL AMERICAN

Insurers and employers have broad access to the medical records and genetic information of Americans through several mechanisms. They also have incentives to use that information to reduce their own risks. In the next section, I describe some of the mechanisms by which insurers and employers conduct actuarial assessments of Americans.

Risk Rating, Medical Underwriting, and Access to Medical Information

The chief problem that GINA is intended to address is tied to the delivery of health insurance in the United States, which is a mixture of private and public. Most Americans with insurance plans are covered by private plans, which are sold by approximately 1,100 for-profit health insurance companies in group and individual plans. A smaller number of Americans are covered under public health insurance programmes (Medicare, Medicaid, and veteran's assistance). These programmes are both entitlement and means-based assistance, and are funded and delivered by federal agencies and states.[1]

In 2006, 61% of the 260 million non-elderly (under age 65) Americans that were insured obtained their health plans through their employers, as a benefit of employment (Kaiser Family Foundation 2007). Workers who are ensured through employer-sponsored group plans are charged the same rates, which insurers set based on the experience of that group, and are not

required to undergo individual medical underwriting. In other words, 61% of the 260 million insured Americans under the age of 65 are not subject to medical underwriting.

Workers who are not eligible for group plans can purchase coverage in the individual insurance market. In 2007, this constituted approximately 5% of non-elderly Americans (Kaiser Family Foundation 2007). They are, however, subject to medical underwriting.

Medical underwriting is part of the rationale of risk rating, a practice that insurance companies adopted in the 1960s, after abandoning the earlier practice of community rating, whereby risk was spread evenly across communities. Risk rating pools together individuals of similar perceived risk based on a set of variables such as age, sex, and medical history, and assigns them similar premiums and coverage (The Ad Hoc Committee on Genetic Testing/Insurance Issues 1995).

In medical underwriting, insurers gather personal and family medical history, occupation, and lifestyle information to assess the risk status of applicants and subscribers and move them into different risk pools. Information about genetic conditions and susceptibility genes provides one more measure of risk for insurers to use. Healthier individuals are charged lower premiums, while sick people are charged higher premiums:

> Insurance companies routinely require applicants to release their medical records to the insurance carrier for individually underwritten policies. These records may indicate the results of genetic tests, a family history of susceptibility to a genetically influenced disorder, or simply casual remarks or inquires about disease with a significant genetic basis. The records may be interpreted as indicating the possibility of genetic risks for certain disorders. The insurance

> company viewing this perceived risk may then either reject the applicant altogether or charge higher premiums for the increased risk. ... Although the insurance industry does not *perform* genetic tests as a precondition to coverage, genetic information, including family history data, contained in medical records may be *used* as a risk-rating device as it becomes available to the underwriter (Brody et al 2001:344).

The rationale guiding risk rating is market-driven: healthier individuals should be rewarded for costing the health care system less, while lower premiums provide an incentive for less-healthy individuals to improve their health. But risk rating also provides profit-oriented insurers with strong incentives to disqualify individuals for coverage, or to increase their premiums when they are assessed as being at a higher risk of disease or disability (Ostrer et al 1993). In a competitive health insurance market, companies can decline to insure individuals whom they deem too risky, leaving them to be picked up by less profitable companies. And insurance coverage in the individual market is much more expensive than in the group (large employer) market.

Even though health insurers do not subject Americans enrolled in employer-sponsored plans to medical underwriting, insurers can gain access to the medical records of any American through the Medical Information Bureau, a centralized database of medical information that insurance companies share to detect insurance fraud. Bioethicist Baruch Brody (Brody et al 2001) explains how this works:

> Patients routinely sign an authorization to release medical information to third parties as a part of their insurance reimbursement scheme. Once the insurance company has this information, there are few legal controls on who has access to it. Insurers may acquire medical information, including genetic

> information, from medical information provided by the applicant, from physicians' medical records, from the Medical Information Bureau (MIB) and from inspection reports from consumer reporting agencies. Medical information from many sources is available to insurance companies, and information sharing is a routine part of the insurance industry. This free flow of information allows insurance companies to use genetic information about the patient and his or her family even though no genetic testing has been done (Brody et al 2001:344).

Competition between for-profit insurers in a risk rating market means that any distinguishing information about an individual that might be used to predict future health risks is valuable to insurers. Knowledge of positive genetic test results—or even that tests were conducted—provides for-profit insurers with a means to further differentiate their clients according to risk—even when their clients may never manifest disease, and when such tests communicate little or nothing about penetrance, age of onset, or course and severity of disease. For example, an insurer may choose to disqualify for coverage an individual with a positive test result for BRCA, who manifests no disease symptoms of breast cancer, by claiming that she has a pre-existing condition. Within the risk-rating rationale of for-profit insurance, genetic testing creates a category of individuals that clinical geneticist Paul Billings and his colleagues have aptly labelled "the asymptomatic ill" (Billings et al 1992:478).

Employer access to medical records

Employers, who purchase group insurance for their employees and provide most Americans with their insurance coverage, also practice actuarial assessments of employees and applicants. Employers can require applicants to disclose their medical records or their medical histories

during pre-employment screening, including genetic test results. They can also subject applicants and workers to genetic tests to identify susceptibilities to disease, a practice that came to light in the 1980s.

Some employers opt to provide health insurance coverage directly to their employees instead of purchasing group plans from insurers, a practice known as self-insurance (Park 2000). In this situation, the employer acts as the health insurer. Because self-insured employers manage their employees' insurance claims, they have direct access to information about workers' genetic tests, health conditions, and family histories. In this setting, results from diagnostic, predictive and susceptibility testing can provide employers in the self-insured market with two means to discriminate against workers. They can raise the insurance premiums of workers thought to be at higher risk of illness or disability because of the anticipated increased health care costs to employers, and they can fire employees or refuse to hire those with positive genetic test results (even if no illness or disability is evident) to avoid anticipated health care costs.

Genetic Discrimination Beyond the United States

Other nations have expressed concern about the potential for genetic discrimination. Norway, France, and Austria have banned the use of genetic testing for employment (Taylor et al 2004), while Australia and the UK have made genetic discrimination a policy issue. In 2003, the Australian Law Reform Commission and Australian Health Ethics Committee released its report "Essentially Yours: The Protection of Human Genetic Information in Australia," and recommended that the

definition of "disability" in the country's Disability Discrimination Act of 1992 be amended to include discrimination based on genetic status. Despite this change, insurers and employers can seek an exemption to the legislation if they can justify discrimination (Taylor et al 2004). In 2002, the Australian Research Council, an advisory body to the government, launched a two-year Genetic Discrimination Project (recently extended to three years) to investigate genetic discrimination among consumers, third parties, and the legal system (Taylor et al 2004). The commitment of government funds to empirical research provides some indication of the extent to which genetic discrimination has been established as a public policy priority in Australia. In the UK, the Human Genetics Commission, the country's policy equivalent to SACGHS in the United States, formed a Genetic Discrimination Monitoring Group in 2003, with a focus on life insurance and workplace genetic screening, which appears to meet infrequently. The UK has imposed a moratorium on using genetic information for insurance products.

THE GENETIC PRIVACY STORY: THREE SOURCES

Mark Frankel: Advancing Congressional Interests by Championing Privacy

Mark Frankel, a widely-published science policy critic who directs the Scientific Freedom, Responsibility and Law Program at the American Association for the Advancement of Science, repeats the concerns that Neil Holtzman (1989) first outlined in *Proceed with Caution*, about the potential impact on workers as susceptibility testing expands, testing costs decline, and health care costs rise. Employers with self-insured health care

plans, Frankel says, will be motivated to institute genetic testing for their employees, denials of insurance coverage will increase, and those individuals who would benefit most from the increased availability of susceptibility testing will be less likely to risk testing for themselves.

Frankel also portrays genetic information as powerfully deterministic:

> The power and potential of genetics rest in the knowledge it provides, which is personal, predictive, permanent (at least for now), and prejudicial, in the sense that the knowledge in and of itself can prompt people to think differently about you. People would ordinarily want to keep it private, but it also could be of great interest to others—family members, employers, insurers, schools, and legal institutions. (Frankel 1999:216)

Given his emphasis on the power of genetic information, is it not surprising that he argues that privacy and genetic discrimination are inseparable. The close relationship between the power of genetic information and a shared sense that it should be kept out of the hands of insurers and employers, he says, explains why legislation intended to prohibit discrimination by these parties typically employs privacy provisions.

For Frankel, the salient questions are those of timing and longevity: why did genetic discrimination become a Congressional issue in 1994, and why did the issue have such staying power, remaining on the federal agenda for five years?[2] According to Frankel, the Human Genome Project brought to light a number of ethical issues that Congress has since debated, and genetic discrimination and privacy have been the most prominent. Efforts to put genetic discrimination on the Congressional

agenda arose from the dissatisfaction a broad group of elite actors with the patchwork of protections afforded by state bans on genetic discrimination.[3] In the mid-1990s, these actors, "joined forces...to plead their case for federal legislation that would broaden the protections offered to larger numbers of people" (Frankel 1999:216). This coalition of actors consisted of government, public advocacy groups, private sector, professional, and a public/private partnership—all of whom, he notes, "are highly knowledgeable about genetics and its social implications."[4]

Frankel singles out the expansion of computerized databases and growing public dismay with the collection and storage of personal information as responsible for putting genetic discrimination on the Congressional agenda. "Information privacy in the United States," he say, "has, for many, become a civil rights issue with the power to arouse people whose concerns, while spread over a wide range of applications, e.g. law enforcement, finances, medical records, and so on, enable them to forge alliances and to appeal to public authorities to take action" (Frankel 1999:217). Consequently, he says, genetic discrimination, framed as the collection and use of personal genetic information, became "an attractive policy issue in the context of the broader political landscape" (Frankel 1999:217). The issue remained on the Congressional agenda because genetic privacy was "an attractive policy issue" that helped politicians to promote their own interests (Frankel 1999:217). In other words, Congress was "sold" on genetic discrimination as a pressing concern because it was framed as an information privacy issue, which has enormous political capital.[5]

Frankel also answers the question of which actors were responsible for bringing genetic discrimination to the attention of Congress and keeping it on their radar. He returns to his earlier observation that it is the champions of genetics research—powerful genetic coalitions and advocacy organizations—rather than civil liberties advocates who have been behind the push for the passage of federal nondiscrimination legislation:

> This is clearly an example of where science is driving policy and where the research community and public advocacy groups are joining forces to shape the content of such policy. The outcome will depend on these groups sustaining their collective focus on genetic privacy and discrimination as a civil rights issue that, if not resolved, will pose serious consequences for the conduct of research and the benefits it produces. That potent theme has helped to propel the issue on to the United States' political agenda and kept it there for the past five years. To secure passage of legislation, these groups will need to ensure the visibility of the issue in the public arena, a task that will undoubtedly be aided by the recent decision of the government and private sector to speed up the timetable for mapping the entire human genome. (Frankel 1999:221)

To understand better which actors have been driving policy on genetic discrimination at the federal level, and how these actors are maintaining the visibility of the issue in the public arena, it is helpful to look at the work of Shobita Parthasarathy.

Shobita Parthasarathy: Breast Cancer Activism and Contentious Politics

Sociologist Shobita Parthasarathy draws on her dissertation research (Parthasarathy 2003) comparing how the United States and the UK implemented BRCA breast cancer testing to explain why privacy advocates and genetic activists in the United States have worked towards legally

protecting genetic information as private and distinct. In a 2004 article based on her dissertation research, she compares the different approaches adopted in the United States and the UK towards genetic privacy, emphasizing the role of breast cancer activists in heightening public awareness about genetic discrimination and popularizing the issue in the mid-1990s. Her account deserves close attention because she provides an explanation for how and when genetic discrimination was transformed from a marginalized concern of Americans with rare genetic disorders into a mainstream social problem with the potential to affect a broad spectrum of Americans.

Parthasarathy locates the first concerns about genetic discrimination in the early 1980s, "in anticipation of widespread availability of genetic testing and increased understanding of the makeup of the human genome" (Parthasarathy 2004:334). The 1983 President's Commission for the Study of Ethical Problems in Medicine and Biomedical Research cautioned against disclosure of genetic information to third parties. Unfortunately, her discussion of this report is brief and she offers no context for the concerns or events that motivated this commission and its report.

From there, her story moves to the 1990s. She outlines activism by three actors that were instrumental in bringing federal genetic discrimination legislation to the Congressional table. These are the Council on Responsible Genetics (CRG), the National Breast Cancer Coalition (NBCC), and the Hereditary Susceptibility Working Group of the National Action Plan on Breast Cancer (NABPC), a private-public coalition sponsored by the NIH.[6]

CRG was the first organization to advance the position that genetic information needed legislative protection because of its view that "public excitement and attention to genomics had led to misinformation about the power of DNA and incorrect expectations among insurers about the relationship between genetic information and disease incidence" (Parthasarathy 2004:335). CRG published position papers and helped individual states and the federal government to draft legislation banning genetic discrimination. Colorado and California were among the first states to pass such legislation starting in 1994. CRG focussed on the results of genetic tests as the measure of genetic risk that needed protection, and not other information such as family history, which also has discriminatory potential.

Yet despite the efforts of CRG to push for state and federal legislation, argues Parthasarathy, genetic discrimination continued to be perceived as a problem affecting few Americans: those with rare genetic disorders. It was only with the mid-1990s discovery of genes for breast cancer that genetic discrimination was seen as a problem that could affect many Americans. "Suddenly, efforts to protect individuals with rare genetic disorders had transformed into a movement to protect relatives, friends, and neighbors who might have a mutation to a very common disease" (Parthasarathy 2004:336). This new urgency brought two major breast cancer coalitions and lobbyists—the NBCC and the NAPBC—on board. With their involvement came a number of influential individuals, including National Human Genome Research Institute (NHGRI) director Francis Collins.[7] In 1995, the group published a report in *Science* (Hudson et al. 1995) based on the recommendations from a workshop the

held in July of that year on genetic discrimination. The group adopted the position that genetic information was distinct and can indicate future health risks of family members, not only individuals. For these reasons, it argued, insurers should be prevented from using genetic information to increase premiums or deny coverage. According to Parthasarathy (2004:337), the NAPBC working group viewed "genetic information as inherently private" and that "genetic information was the private property of the individual alone."

Parthasarathy advances the argument that the United States is characterized by an adversarial political culture. For the issue of genetic discrimination, this means that privacy advocates faced off against insurers, with government taking the side of the former. The strategy that American activists adopted, she says, was to make the case that genetic information was a unique form of personal information and therefore warranted legislative protection. In other words, the adversarial style of advocacy politics in the United States informed activists' legislative strategy to treat genetic information as distinct and private. This style contrasts with what Parthasarathy calls the "more conciliatory and cooperative tone" of UK activists, who rejected the strategy of designating genetic information as distinct or private:

> In the United States, where patient advocacy groups and insurers engaged in adversarial politics, advocacy groups defined the results of genetic testing as new products of biotechnology that needed to be protected in direct opposition to the views of insurers. They argued that genetic information constituted a novel and distinct category that was inherently private, and fought to develop legislation at both state and federal levels that reflected this understanding. Government officials seemed sympathetic, accepting that genetic information

> occupied a distinct category of protected information and placing greater weight on the individual's right to privacy than the insurer's right to access genetic information. (Parthasarathy 2004:349)

A major victory for genetic discrimination activists was the passage of the federal Health Insurance Portability and Accountability Act (HIPAA) in 1996. It was the first federal measure to protect individuals from genetic discrimination in group health insurance plans, which is the market most employed Americans belong to. According to Parthasarathy (2004:338), HIPAA enshrined the status of genetic information as "both private and distinct from other types of medical information," and "privileged the individual's right to determine the meaning of genetic risk while de-emphasizing the insurers' right to control the use and meaning of genetic information." However, advocates such as the NBCC were unhappy with HIPAA's inadequacies and loopholes, and continued to work with Congressional representatives to draft and introduce comprehensive federal bills banning genetic discrimination:

> By the end of the 1990s, advocacy groups in the United States had taken advantage of public concern about the new technology of genetic testing and become quite successful in defining genetic information as both private and different from other types of medical information. This definition was codified in both the HIPAA regulations as well as many state laws, and both federal and state governments continued to explore the issue to see whether additional legislation was warranted. (Parthasarathy 2004:339)

Like Frankel, who argues that political issues stay on Congressional agendas because they serve politicians' interests, Parthasarathy adroitly observes that issues advance to legislative action because they acquire high-profile champions. Genetic discrimination stayed in the public eye

and moved into state and federal legislative arenas because of the advocacy of Francis Collins, the NBCC, and the NAPBC, and their *Science* article. (See Chapter 5 for further discussion). But activists usually have opponents, particularly in the adversarial political climate of the United States, and Parthasarathy introduces a key one into her story. Health insurers such as the Health Insurance Association of America and the National Association of Health Underwriters challenged efforts to pass new laws. They have objected, of course, to the idea of further regulations impeding their practice, but also to the twin ideas that genetic information is different from other kinds of medical information, and inherently private.

While Parthasarathy's chronicle of genetic discrimination activism begins to fill in the picture of how and why genetic discrimination became a pressing problem for Americans, and identifies key concerns motivating some of the actors who have championed the genetic discrimination cause, her analysis is limited in two ways. One limit is her focus on breast cancer politics. This filters out key actors and important events unrelated to breast cancer activism that have nonetheless shaped public understanding of genetic discrimination as a significant problem. The other is her tacit acceptance of privacy as a social good. Why do Americans venerate privacy, and what kind of political work does the privacy banner do? What interests and concerns does privacy mask or absorb?

Anthropologist Margaret Everett takes up some of these questions. Despite the fact that her analysis puts the problem of genetic discrimination into the realm of genetic privacy activism, I include her

publication in my review. Everett examines the complicated implications of property rights and personhood claims that have figured in policy debates on genetic privacy. These are concerns that I will return to in Part Two of the dissertation. Her analysis also illustrates the need for an historically-informed account of the origins and shaping of genetic discrimination as a problem.

Margaret Everett: The Genetic Privacy Movement

Margaret Everett is a cultural anthropologist who served for two years on Oregon's Genetic Research Advisory Committee, which amended the state's genetic privacy law in 2001 by removing its property rights provisions. Her interest in genetic privacy legislation and genetic discrimination grew out of this experience. This helps to explain why she regards genetic discrimination as a subset of the problem of genetic privacy. According to Everett, genetic privacy references "fears of discrimination, social stigma, familial problems, loss of control over one's identity, as well as assertions of the rights to know and not know, and to freedom from government interference in private choices, including abortion" (Everett 2004:277). Another clue to her concerns with confidentiality, personhood and identity is that she is writing for a genetic counselling audience in one of these publications (Everett 2004). She comments that privacy concerns related to "social stigma, self-identity, and psychological issues" are "well-known to genetic counselors" (Everett 2004:277).

Everett begins her analysis with a review of state and federal legislation that has designated a unique status for the genetic information of

individuals. While an insightful discussion (she notes, for example, that four states have stipulated that genetic information is the personal property of the individual), it quickly becomes evident that she too readily conflates concerns about genetic privacy with the problem of genetic discrimination. While some federal anti-discrimination legislation contains genetic privacy provisions, commentators do not typically label these laws "genetic privacy legislation". For example, Everett discusses the intent and impact of three pieces of federal anti-discrimination legislation that are often cited by genetic discrimination activists as providing important but inadequate protections against insurer and employer discrimination: the Americans with Disabilities Act (ADA), passed in 1990; HIPAA, passed in 1996; and President Clinton's executive order banning federal agencies from using genetic information to hire or promote employees, passed in 2000.

Where she does concentrate solely on genetic discrimination, she correctly notes that most of the evidence of discriminatory practices has been anecdotal. The exception is the widely-cited Burlington Northern Sante Fe Railway case.[8] Everett also challenges widespread claims that fear of discrimination by insurers is holding Americans back from seeking genetic testing. It is here that her anthropological concern with everyday practice and meaning distinguishes her commentary from others. While some surveys indicate that a majority of Americans are worried they may lose their insurance coverage or be fired if they seek genetic testing, Everett points out that one problem with these surveys is that genetic information as a category is difficult to define and might not be well understood by the public. She also cites the work of Hall and Rich (2000)

showing that patients worry more about the psychological impacts of genetic testing than the possibility of insurer or employer discrimination.

Everett is writing about what she calls the genetic privacy movement. According to her, this movement was spearheaded by bioethicists who wanted to pass federal privacy legislation. However, she singles out just one such bill: the 1995 *Genetic Privacy Act: A Proposal for National Legislation*. This bill was drafted by bioethicist George Annas and two colleagues at Boston University School of Public Health, and funded in part by the National Institutes of Health (NIH). The impetus for the bill, according to Everett (2004:274), was "the Human Genome Project and the growing anticipation of, and anxiety over, the importance of genetics for clinical medicine and biotech research." Annas and colleagues drafted the bill to protect individual privacy and enhance individual control over their genetic information, particularly in genetic counselling settings, but also in research settings. The bill outlined procedures for obtaining informed consent, disclosing risk, explaining the purposes of testing, and assuring confidentiality. It also outlined a provision to treat an individual's genetic information as personal property.

Everett's (2007) later account of the Oregon Genetic Privacy Act of 2005 offers not only a more cogent treatment of the issues that have engaged her, but a personal story that explains why she chose to serve on the Committee. In 1998, her newborn son Jack was diagnosed with Menkes disease, a rare, genetic disorder that is lethal in newborns. Two months after geneticists took more blood and tissue samples from her newborn son, and before they could deliver the results, he died

unexpectedly. The UK geneticist in possession of Jack's tissue samples then contacted Everett and husband, asking if he could send some of their deceased son's cells to an Italian laboratory. This request triggered discomfort for Everett and her husband:

> Somehow, this news brought up disturbing questions for us, and we were surprised by the emotional weight of the decision. I found myself trying to decide what part of Jack was contained in those cells. My husband and I talked about the strangeness of his immortal cells, growing in three different countries after his death. It made his life—or maybe it was the story of his life—feel unfinished in a way that was unsettling. Moreover, I did not like the idea of his life, short as it was, being reduced to a disembodied lab sample. (Everett 2007:378)

The anthropological strength of Everett's (20o4) analysis of the *Genetic Privacy Act* is that she probes the relationship between two ideologies that shape genetic privacy legislation. One ideology is genetic exceptionalism, the belief that genetic information is more sensitive and predictive than other medical information, and deserves special treatment. The other is genetic essentialism, the belief that genes largely determine how we are shaped. Everett questions some of the assumptions that Annas and colleagues make about genetic information, asking with scepticism whether this information truly can be considered a diary of future health conditions, as these bioethicists claim. She also asks to what extent "genetic information poses unique concerns about confidentiality and discrimination" (Everett 2004:282), given that third parties routinely discriminate on the basis of medical conditions unrelated to genetic illness (for example, HIV status). She sides with the many commentators who argue that practicing genetic exceptionalism reinforces the reductionism

inherent in genetic essentialism, and concludes that treating genetic information as special may lead to greater restrictions on its use and dissemination. Given criticisms that state genetic privacy laws fail to protect Americans from genetic discrimination, the most important question Everett asks with respect to federal draft legislation to protect genetic privacy and ban genetic discrimination is, "[o]ne might ask, then, what these laws are about—what new fears and anxieties do they reflect?" (2004:276). She might also have asked, "What political interests do these laws advance, and for whom?" While Everett (2007) rightly challenges the tendency among legal scholars and bioethicists to treat privacy claims and property claims to the body and its parts as separable claims, it is disappointing that in her analysis of personhood concerns that she describes from her own life, that she remains bounded by the legalistic and narrow framework of privacy rights. Although I disagree with Everett that there is such a thing as a genetic privacy movement, her discussion of extended personhood in "disembodied" tissue samples is relevant to questions about the assumed bioavailability of the American population for federal genomics research. I revisit her comments in my conclusion (Chapter 10).

Limitations of the Genetic Privacy Story

The three authors underline the fact that privacy is highly valued in the United States, as is the idea of protecting personal and medical information in general, and genetic information in particular. Clearly, protecting genetic information as something both private and different from other kinds of health information has been an important—and highly

visible—legislative goal for many genetic discrimination activists.

Frankel and Parthasarathy offer politicized accounts of genetic discrimination activism. They see genetic privacy as a particularly resonant and politically expedient strategy (or frame) that genetic discrimination activists and Congressional politicians have adopted to advance their causes. These activists consist of a wide array of federal scientists, genetic coalition groups, and genetic patient advocacy organizations according to Frankel. Parthasarathy singles out the efforts of breast cancer activists and NHGRI director Francis Collins in keeping genetic discrimination visible to federal lawmakers.

Everett, on the other hand, privileges genetic privacy as a driver, arguing that privacy concerns articulated by bioethicists in the mid-1990s have driven genetic discrimination activism. It is Everett who coins the term "the genetic privacy movement" to describe collective action aimed at passing state and federal legislation banning genetic discrimination. To my knowledge, no one else had adopted this label. It is difficult to know if she is subsuming all genetic discrimination activism to genetic privacy activism, or whether she is simply unaware of the history and extent of genetic discrimination activities. Regardless, her account of the relationship of genetic discrimination activism to genetic privacy is not supported by empirical research. Although protecting privacy has always been an important element of the genetic discrimination debate, contrary to what Everett tells us, genetic discrimination has not been widely viewed as a problem of protecting individual dignity or personhood, improving informed consent, or controlling the circulation and ownership of tissue

samples. Rather, it has been characterized largely as a problem of protecting individuals from discriminatory decisions by institutions affecting their insurance prospects, livelihood, or future opportunities.

Enhancing genetic privacy protections is one political strategy among many for resolving or preventing genetic discrimination. Most genetic discrimination activists have converged upon the goal of passing comprehensive federal legislation banning discrimination by insurers and employers. Draft federal legislation employs genetic privacy provisions. These provisions are a mechanism to circumscribe the actions of insurers and employers, so that Americans do not lose their health insurance or their jobs, do not pay more for their insurance premiums or face demotions, and are not made ineligible for insurance coverage or employment, as a consequence of seeking genetic testing or having a positive result. As Frankel in particular, but also Parthasarathy, argues, genetic privacy should be viewed as a successful strategy—one among many—that actors have wielded to galvanize public attention and Congressional action.

While acknowledging that privacy is an important feature of why genetic discrimination is perceived to be a threat to many Americans, I suggest that the genetic privacy story obscures reasons why genetic discrimination emerged as a problem twenty years ago, and why federal scientists and genetic activists today are characterizing it as problem affecting all Americans. As an endpoint or solution, genetic privacy and efforts to pass federal legislation banning genetic discrimination tell us little about how the problem of genetic discrimination has been made.

Because it is a compelling frame with populist appeal, genetic privacy has absorbed or obscured some of these concerns. Genetic discrimination activists have been a heterogeneous set of actors with diverse interests. Other concerns and interests, besides genetic privacy specifically and protecting the civil liberties of Americans more generally, have been at work in shaping public awareness and understanding.

There is a more interesting story to tell about how genetic discrimination became a problem in the United States and why it has acquired its current stature. First, I locate the emergence of concern and activism on genetic discrimination in the 1970s with practices that predate the Human Genome Project: the development of recombinant DNA (rDNA) and concern by scientists over its potential to foster discrimination; and in the adoption by major chemical and manufacturing firms of genetic screening of employees for "hypersusceptibility" to occupational diseases. Secondly, I situate my story in the anthropology of new genetics. I draw out two aspects of this field that the privacy authors have overlooked: the political economy of the genomics enterprise in the United States; and the ways in which genetic discrimination discourse is tied to the execution of biopolitics (Foucault 1978a; 2008) in the United States. By situating my analysis in the anthropology of new genetics and adopting a political-economic perspective, and focusing not on individual practices but how the state is building a genomic nation and thereby shaping shared understandings of what it means to be a good American, I offer a biopolitical (Foucault 1978a) analysis of genetic discrimination.

I draw on two anthropological constructs for my analysis. One is

genetic citizenship (Heath et al 2004; Rapp et al 2006, Taussig et al 2003), which lies at the intersection of two sets of literature: the anthropology of genetics, and the anthropology of citizenship. The other construct is bioavailability, from the work of medical anthropologist Lawrence Cohen (2004) on organ harvesting in India (and hence, not from either of these two sets of literature). In the next section, I review these two sets of literature and explain more clearly the utility of these constructs to my work.

LOCATING THE PROBLEM: THE ANTHROPOLOGY OF NEW GENETICS

My investigation of the origins of genetic discrimination can be regarded as a study of contemporary biopolitics in the United States, following the work of Nikolas Rose (2001, 2007) and sociologist Carlos Novas (2006), particularly on biological citizenship (Rose and Novas 2005). However, the dissertation is above all a contribution to the anthropology of new genetics, using a political-economic framework that represents biopolitics in relation to state functions such as economic and technological development. In support of this approach, I draw on the complementary work of political scientist Rodney Loeppky (2005) on the political-economic origins of the HGP, and Sunder Rajan (2005, 2006) on biocapital.

Anthropologists began investigating what is called the new genetics in the late 1980s, and critiquing multiple practices and phenomenon: the routinization of genetic diagnostic practices such as amniocentesis; the advent of the HGP and national genomic enterprises; and the increasing embrace of medical (and molecular) explanations of disease (see

Pálsson2007). In the next section, I review this literature along four organizing themes: patient experiences of genetic testing and screening; complexity and uncertainty; global genomics and biocapital; and biosociality and genetic citizenship. I single out some of the questions and constructs that intersect with my findings and analysis.

Patient Experiences of Testing and Screening

Rayna Rapp's decade-long study of the social impact and cultural meanings of prenatal diagnosis amongst women and couples in New York City (e.g. Rapp 1987, 1988, 1997, 1998, 1999) set the standard for empirical investigation of patient encounters with genetic technologies.[9] Rapp has explored in exquisite detail the moral nuances and ambiguities of how and why women choose to undergo or refuse amniocentesis, and among those who do, how do they integrate the probabilistic information they receive with their personal and familial risk calculi.

Rapp's long-term work identifies the many variables, including reproductive history, previous experience with disability, religion, ethnicity, and education, that structure moral decision-making for women (and men) around whether to accept or refuse amniocentesis, and whether to bring to term or abort a fetus with a diagnosed abnormality. "Those who refuse the test, no less than those who accept it, are, therefore, responding to a complex, highly-structured social nexus within which they negotiate and exercise personal choice," she says (Rapp 1998:62). In light of her extensive work on genetic decision-making, it seems correct to question the utility of survey results that identity fear of genetic discrimination as the most significant barrier to Americans seeking genetic

testing. It is difficult to accept at face value the validity of survey results that have not also probed through ethnographic study the role of fear at *all* stages of genetic decision-making. Such research would need also to distinguish between diagnostic and predictive testing for rare single-gene disorders, and susceptibility testing for common disorders.

It was Rapp who coined the phrase "moral pioneers" (Rapp 1987, 1999), drawing attention to the complexities of genetic decision-making that women and couples experience as prenatal diagnostic testing has becoming routinized. In a society that venerates the decision-making autonomy of the individual, the expression also conveys the loneliness of shouldering this burden. Her finding, that "[t]hose without much privileged scientific education are most likely to reject testing altogether" (Rapp 1998:67), calls into question the NHGRI's optimistic vision of personalized medicine, which assumes that Americans will acquire the genetic literacy, and have the desire and time, to pursue probabilistic genetic decision-making as part of their routine clinical care (see Chapter 6 for a discussion of personalized medicine).

Rapp reminds us that clinical genetics is not simply a narrow technical medical speciality with little relevance to everyday life. It is, she says, "a field that provides powerful and proliferating discourses on the state of being human" (Rapp 1998:47). These powerful discourses on genetic discrimination are what I examine in Chapter 8. What they highlight is not so much the state of being human, but the state of being American in a nation committed to being genomic.

Complexity and Uncertainty

Margaret Lock's recent work (Lock 2005a, 2005b, 2007; Lock et al 2006a, 2006b, 2007) extends Rayna Rapp's pioneering research on the cultural meanings of diagnosis into the realm of susceptibility testing. Using late-onset Alzheimer's disease as her case study, she examines the epistemological frameworks that shape molecular research, and tracks what she calls the "penetrance and uptake of genetic knowledge" (Lock et al 2006b) among clinicians, patients, and the public. She argues that social scientists must begin examining how patients integrate the rapidly-shifting risk knowledge produced from susceptibility testing into their own multi-causal theories of disease and inheritance (Lock et al 2006b).

Lock's important work on the genetics of AD highlights several themes: the persistence of the genetic reductionism paradigm even as researchers embrace epigenetics; the provisional and shifting nature of genetic knowledge and the difficult of producing risk estimates; and the frequency of biosocial groupings (Rabinow 1996). I briefly examine each of these themes in turn.

Noting the declining power of genetic reductionism as an explanatory paradigm in molecular biology—"the rise and fall of the genotype/phenotype dogma" (Lock 2005a:S48)—and the corresponding embrace of epigenetics by molecular scientists, Lock wonders about the staying power of genetic determinism, so entrenched in popular conceptions of disease:

> Is the idea of a controlled life and mastery of risk, so central to modernization and the production of Beck's risk society, being reinforced through the allure of DNA testing? Or, on the other hand,

> as information about genetic susceptibility with its inherent uncertainty is increasingly disseminated, is a belief in a technologically assisted future of bodily mastery disappearing below the horizon, to be replaced by a Postgenomic angst of uncertainty? (Lock 2005b:S66)

She notes that "[w]e would do well to understand the extent to which basic scientists, clinicians, patients, families, advocacy groups, and the public are captivated by genetic determinism and how these same groups of people are likely to respond to an emerging discourse in which the gene no longer reigns supreme and the limitations of current genomic knowledge cannot be denied" (Lock 2005a:S48-S49). The SACGHS hearings offer some glimpse into the stamina and explanatory power of genetic determinism in a nation that has declared it will transform understandings of human disease and the delivery of health care through genomics. In the realm of public discourse, narratives of genomic nation-building draw heavily on the salvational imaginary of genetic determinism.

Lock points out that the task of producing risk estimates for diseases such as AD is challenging since no single susceptibility gene will account for all cases of the disease (Lock et al 2007). Consequently, risk estimates will always be provisional, making extrapolations from populations to individuals difficult. She adds another layer of complexity to the picture with the finding that relatives of individuals diagnosed with AD grasp the complexity of AD genetics but they integrate risk estimates with their own models of who is most at risk (Lock et al 2006b). One conclusion she draws from this research is that family history will continue play the most important role in shaping perceptions of who is genetically at risk in AD, and "people whose family lives are affected by AD will persist in doing

what they have always done—divine the future on the basis of family histories" (Lock et al 2007:272). I wonder to what extent this finding will hold true in the United States in the coming years, where citizens have paid with their tax dollars and hope for scientists to deliver divinatory tests to the clinic, so as to fulfil the dream of individualized (and not family) medicine. How Americans will integrate the multiple risk calculi that whole-genome sequencing (if it arrives—see Chapter 6) will produce into their personal risk models will provide a large terrain for ethnographic investigation.

One question Lock takes up across her work (Lock 2005a, 2005b, 2007, 2008) is whether biosociality (Rabinow 1996) is always seen with genetic diagnosis. Paul Rabinow's biosociality concept builds on Foucault's central interest in the constitution of the modern subject through the adoption of technologies regulating processes of life, reproduction, and death (Lacombe 1996; see Foucault 1983). Biosociality describes the enrollment of individuals into collective identities around specific biomedical categories or diagnoses. These identities become politicized and increasingly central to the social lives of patients, suggests Rabinow. Lock asks whether diagnosed individuals and their family members always form kin and political alliances around their molecularized identities. Her research finds that most families associated with AD do not participate in support groups, because of the stigma and shame of that disease (Lock 2007; Lock et al 2007). If anything, she suggests, an AD diagnosis produces a reduced sociality. Lock's observation mitigates against the uncritical embrace of biosociality by social scientists. It also throws into relief the bold predictions by champions of federal nondiscrimination

legislation, that passage of GINA will open the floodgates to American participation in genetic testing, research, and clinical trial. These predictions bank on Americans embracing, through personalized medicine and expanding susceptibility testing, a nationalized biosociality that goes beyond participation in support groups.

The story of how genetic discrimination has been shaped into a major social problem indexes both the political economy of genomics as a nation-building project, and the shaping of rights and responsibilities of Americans as participants in a genomic society. I locate my dissertation in these last two areas, and draw attention to two constructs that I use to analyse my findings: bioavailability (Cohen 2004), and citizenship.

Genomics and Biocapital

One of the deficiencies of the genetic privacy story is the absence of a political-economic framework. Such a framework is needed to link activism on genetic discrimination within the United States to the goals of establishing genomics as a successful commercial venture and research paradigm in that country. This absence of a political-economic framework is also evident in the risk literature, which I discuss at the end of this chapter. As the work of several anthropologists—Paul Rabinow, Gisli Pálsson, and Kaushik Sunder Rajan—shows, genomics must be understood as both a form of global capitalism that many nations are pursuing, along with the United States, and as a set of technologies and practices that have developed in distinct national directions.

Even within anthropology, genomics has been undertheorized as the intersection of private global capital and national political-economic

interests. Although genetic screening and individual testing predate the HGP and genomics by forty years, discourses of genetic screening and testing are being absorbed into personalized medicine, as genomics has become a focus of wealth generation and is influencing public health. In Chapter 6, I discuss the work of Paul Rabinow on France's notable genomics controversy, Rabinow and Pálsson on Iceland's National Health Sector Database, and Sunder Rajan (2005, 2006) on how genomics constructs subjects into consumers and experimental subjects.

Biosociality and Genetic Citizenship

I have already outlined Rabinow's biosociality construct. But how have anthropologists adopted it, and what relationship does it have to genetic citizenship? Medical anthropologists Deborah Heath, Karen-Sue Taussig, and Rayna Rapp have explored the complex sociality and politics of patient support groups and genetic coalitions that have developed around genomic research (see Heath et al 2004; Rapp 2002; Rapp et al 2001, Rapp et al 2006; Taussig 2005; Taussig et al 2003). Here I review their work while providing a summary of the different kinds of organizations.

Genetic patient support groups for rare, single-gene disorders have existed since the late 1950s. These organizations include the American Hemochromatosis Society, the Cystic Fibrosis Foundation, and the Little People of America, to name just a few of the approximately 600 genetic patient interest groups in the United States. The U.S. commitment to fund the HGP and pursue postgenomics research has been a boon to patients and families of patients with single-gene disorders. As researchers have located genes responsible for these disabling disorders, patients and

families have benefited from the acknowledgement of their diseases, from a commitment to ongoing research and funding for their diseases, from the development of prenatal and diagnostic tests, and from the promise that genomics can eventually develop treatments for these diseases. Not surprisingly, most of the 600 genetic patient support and advocacy organizations that now exist only began in the 1990s, after the launch of the HGP.

In their work on genetic patient groups, Deborah Heath, Karen-Sue Taussig and Rayna Rapp draw attention to what they call "technosocial networks of association" (Heath et al 2004:159) and "rapidly emergent social forms in the age of genetics" (Rapp et al 2001:392). These networks have formed in the wide ambit of the HGP and the increasing geneticization (Lippman 1991) of health and disease. The anthropologists have coined the term genetic citizenship to capture these "new forms of science-health activism" (Rapp 2002)—an activism that sociologist Carlos Novas (2006) in a similar study, calls a "political economy of hope." This activism is characterized by the democratic participation of laypersons in setting genetic research agendas, controlling the material resources essential to genomic research, and enrolling the support of critical actors, including clinicians, scientists, and politicians, as advocates for research in their rare diseases.

Their thinking about genetic citizenship is based on three years of fieldwork that the medical anthropologists conducted with three genetic patient interest groups that provide information and support to families with heritable connective tissue and skin disorders (Rapp et al 1998). Little

People of America represents families with dwarfism, including achondroplasia (Rapp et al 2001; Taussig et al 2003); DEBRA (Dystrophic Epidermolysis Bullosa Research Association) is a patient support group for families with epidermolysis bullosa, which causes painful skin blistering (Heath et al 2004); and the National Marfan Foundation represents families who suffer from Marfan Syndrome, a connective tissue disorder associated with long limbs. The anthropologists also investigated the workings of PXE International, an organization started by Sharon Terry with her husband Pat, which coordinates research for patients with pseudoxanthoma elasticum (PXE).

These organizations, and others that they mention, such as The National Down Syndrome Society, the Coalition of Heritable Connective Tissue Disorders, the Coalition of Patient Support Groups for Skin Disease Research, The National Organization of Rare Diseases, and the Genetic Alliance (Rapp et al 2001), are remarkable for having developed new strategies and tools to promote their members' interests and secure resources for research. For example, many have used information and communication technologies, such as online forums or chat groups, to create communities and open up "participatory knowledge-making" (Heath et al 2004:156) for patients and families. The electronic forums facilitate kin relations and information sharing amongst patients and their family, who are geographically dispersed. DEBRA, for example, runs EBmommas, "a family-driven electronic self-help group" (Heath et al 2001:155). These electronic forums are also a means to enlist the advocacy of researchers. For example, Little People of America collaborated with geneticists interested in studying heritable dwarfism to create a chat group

that brought "clinicians, patients, and researchers together on-line to compare notes on 'their' conditions" (Rapp et al 2001:392).

The organizations have also successfully mobilized the biosociality (Rabinow 1996) of their shared disease identities to secure interest from, and support by, scientists, clinicians, politicians, and industry, who have the resources to channel research dollars into their diseases. The anthropologists define biosociality variously as "the conscription into a new identity politics as people come to align themselves in terms of genetic narratives and practices" (Taussig et al 2003:60), and a "kinship of affliction" (Heath et al 2004:155; see also Rapp et al 2006). The Little People of America, for example, was one of the first patient interest groups to forge a "biosocial coalition" with genetic researchers and clinicians (Taussig et al 2003:6). In another article, the authors describe how parents affiliated with DEBRA brought their bandaged and suffering babies into the offices of Senators in the late 1970s—using "'their bloody, blistering babies like a battering ram'" in the words of a former director of DEBRA (Heath et al 2004:155)—to enlist politicians' support for research into an otherwise unknown and marginalized disease (see also Rapp et al 2006). In doing so, say the medical anthropologists, members of DEBRA "are making citizenship claims on behalf of their genetically vulnerable offspring" (Heath et al 2004:155). That is, they are making claims on the state and scientists for resources, recognition, and the power to direct research that reaches far beyond what they could muster on their own.

These organizations have also inserted themselves directly into the enterprise of science knowledge production. The most remarkable story is

that of Sharon Terry and her husband Patrick, two non-scientists who transformed themselves into researchers after their two children were diagnosed in 1994 with PXE. After discovering that little was known about this disease (Novas 2006), the two formed PXE International and developed a blood and tissue repository, and a patient registry. Sharon Terry was also named co-inventor on the patent for the gene associated with PXE (ABCC6).[10] DEBRA, too, created its own registry of patients' tissue samples. By controlling researchers' access to the material resources essential to research (banked tissues samples and family histories of rare diseases), as well as their products (patents, for example), these organizations have been able to make themselves an obligatory passage point (Callon 1986) for anyone wanting to study the diseases they represent.

Heath, Taussig and Rapp make several claims about the significance of these biosocial networks. They "challenge conventional notions of a divide between lay people and experts" (Heath et al 2004:152) and they represent new models of scientific knowledge-production in the realm of genomics research around single-gene disorders. The anthropologists suggest that one aspect of this genetic citizenship is the democratization of science, and is a hopeful sign that those previously marginalized from scientific decision-making have developed some means to participate in knowledge-production.

As promising as these cases are for the inclusion of single-gene organizations and their members into the mainstream of genomic research in the United States, however, I would like to throw some cold water on

the idea that patient activism has produced scientifically-competent lay experts in significant numbers, or that their activities signal the democratic inclusion of marginalized populations, including the disabled, in mainstream science. Even if we agree that lay experts exist (see Prior 2003 for a dissenting view), the lay incursion into knowledge-production and agenda-setting that Heath, Taussig and Rapp describe is, and continues to be, exceptional. The organizations that Heath, Taussig and Rapp worked with represent a minority of Americans. Little People of America, for example, has 6,000 members. Moreover, few laypersons—activists or otherwise—acquire sufficient expertise, cultural capital, and opportunity to alter federal clinical trial practice, set research agendas, or transform themselves into published scientists who also hold patents on their discoveries (Hess 2004). Sharon and Patrick Terry are two exceptional individuals who lead four genetic advocacy and research organizations. Sharon Terry's achievement in co-discovering and co-patenting the PXE gene has yet to be reproduced by other genetic activists.

While some social scientists (for example, Schaffer et al 2008) have extended the genetic citizenship construct to characterize patients and family members who acquire and share medical knowledge through online support groups and access to research databases, I am sceptical, like medical sociologist Anne Kerr (2003a, 2000b) of the extent to which these activities truly open up "democratic possibilities" (Taussig et al 2003:62, quoted in Schaffer et al 2008:157) for non-scientists.

My ethnographic research on the shaping of genetic discrimination by state actors and genetic advocacy organizations suggests that the

celebratory tone of Heath, Rapp and Taussig on the possibilities that genetic activism offers for agenda-setting needs to be tempered. Their evident enthusiasm for the democratic engagement of genetic activists with researchers obscures the ways in which a small number of genetic activists have represented themselves as the voice of the genetics community, while securing their own power base and marginalizing other public health concerns.

Here I return to Lock's (2005a) interrogation of the utility of the biosociality construct. A key finding of my ethnographic research is that biosociality is alive and well in the United States, particularly around genetic discrimination. Testimony at the SACGHS added two layers of biosociality to the disease-specific form that is the engine of genetic citizenship. One was a biosociality of family: of consanguinal kin ties, of sharing disease alleles and the risks of genetic testing. The other was what could be called a nationalized biosociality of all Americans as carriers of flawed genes and deserving of legislated protection from discrimination. Individuals who testified to the committee about their experiences of discrimination turned their biological claims of difference and misfortune into a positive citizenship claim on the state for all, comparing the presence of disease alleles to other immutable markers of difference that have become the centre of civil rights battles in the United States and efforts to legislate equality. This message, which was repeated by several organizations that also submitted testimony, amounted to an involuntary biosociality imposed on all Americans by a few. As I will argue in Chapter 9, these discourses suggest that a different formulation of citizenship—what I call genomic citizenship—is emerging alongside genetic citizenship.

This formulation of citizenship, I argue, ultimately serves the interests of federal genomics agencies such as the NHGRI, and genetic advocacy organizations. It does not open up democratic possibilities of engagement for marginalized populations that have no interest in becoming genomic citizens or no ability to become such subjects.

Along with the recent involvement of advocacy organizations for common diseases (such as the NBCC) in genetics advocacy and lobbying, a second type of genetics organization has appeared, which Heath, Taussig and Rapp have not examined. These are the single-interest coalition groups that have formed around promoting personalized medicine and removing barriers to its delivery. One of these is the Coalition for Genetic Fairness, an interest group comprised of industry, government, consumer groups, civil liberties organizations, health professionals, and genetic advocacy organizations that formed in 2000 to lobby Congress to pass federal nondiscrimination legislation. Another is the Personalized Medicine Coalition (PMC) which formed in 2004 and bills itself as "a non-profit group that works to advance the understanding and adoption of personalized medicine for the ultimate benefit of patients."

The PMC's diverse membership provides a snapshot of the landscape of genomics actors in the United States: patient advocacy groups (for example, Sarcoma Foundation of America), diagnostic firms and commercial laboratories (for example, Quest Diagnostics), insurers (for example, Aetna), big Pharma (for example, AstraZeneca and Pfizer), universities and research centres (for example, Duke, the Mayo Clinic), and the major governmental players (for example, the CDC, FDA, NHGRI,

NCI, and CMS). However, a look at its board membership reveals that the organization is controlled by the biotechnology industry. The PMC is an advocacy organization that represents biotechnology interests in advancing personalized medicine (and ensuring the production of consumers for its products and services), but it is also the promotional arm of government agencies committed to personalized medicine: the NHGRI, the FDA, and the DHHS.[11]

These coalition organizations tend to blur the once-comfortable lines between expert and layperson, insider and outsider (Hogle 2002). When Sharon Terry, a former nun and now a mother, a co-discovered of a gene, and President and CEO of the umbrella organization Genetic Alliance, speaks as chair of the Coalition for Genetic Fairness, whose interests are on the table? The entrance of these coalition groups suggests not only that genetic politics in the United States is becoming more complex, but also that genetic activism in the United States presents less of an opportunity for democratic participation than Heath et al (2004) suggest.

THE ANTHROPOLOGY OF CITIZENSHIP

The appearance of the genetic citizenship construct can be better appreciated when set against two decades of citizenship inquiry within anthropology. Citizenship studies have undergone a renaissance in the last decade, not only among political theorists (see Kymlicka and Norman 1994) but also social scientists. Anthropologists have been especially industrious in developing new uses for the construct in their work. Cultural anthropologist Aihwa Ong (2004, 2006) notes that uses of the citizenship construct have moved beyond the dominant juridical

interpretation of the 1970s, where it signified the possession of legal rights, to the more inclusive signification of membership, identity, and entitlements. The hallmark of contemporary uses citizenship, says Ong (2006:499) is the disarticulation of its properties (territoriality, rights, duties, membership, identity) and re-articulation "with universalizing forces and standards" such as human rights.

Renewed interest in citizenship follows an intensification of political events and social dynamics at the regional, national and transnational levels since the 1990s. These include nationalist and independence movements that have shifted state boundaries and membership (in the Ukraine, the former Soviet Union, and the Balkans); claims for entitlement and recognition being made by identity social movements (for example, legalized marriage for gays and lesbians); Mexican and Latino immigrant and refugee experiences in the United States; and claims to health care, therapeutic treatments, compensation for occupational injury and disease, and control of the production of genetic knowledge.

Two decades of vibrant anthropological inquiry has produced proliferating citizenship forms that reference different aspects of belonging, exclusion, identity, and claims on the state. These include "cultural citizenship" (Rosaldo 1997) as the right to express ethnic and cultural differences while participating in the process; "flexible citizenship" (Ong 1993, 1999, 2004) as the security of juridical citizenship for Hong Kong entrepreneurs who have immigrated to the United States and whose dual citizenship allows them to pursue business opportunities at home; and "negative citizenship" (Biehl 2004a) to describe the lives of the

unwanted and abandoned in Brazil, who can produce only disease and remind citizens of how not to live. Moving into medical anthropology, we have "biomedical citizenship" (Biehl 2004b) marking the shift in Brazil from political to biological rights with the birth of a constitution that makes free AIDS therapy a right of citizens; "therapeutic citizenship" (Nguyen 2005) as a stateless form of citizenship whereby HIV-positive citizens in local West African setting mobilize their social networks to access treatment; and Adriana Petryna's (2002, 2004, 2005a, 2005b) complex "biological citizenship" as the emergence of new categories of the at-risk and entitlement negotiated by survivors of the Chernobyl explosion during political transition and the erosion of scientific certainty.

The citizenship theorizing of medical anthropologists has tended to focus on the negotiation of, or claims for entitlements. But what about the duties of citizens? I also address the question of moral duties and virtues of Americans. One of the questions I ask is, what constitutes good citizenship within a country that is committed to being a genomic nation? What does it means to be a virtuous American?

The genetic privacy story highlights concerns about individual and familial bodily privacy and integrity, but it overlooks the state's interest in the bodies of its citizens as resources for nation-building and developing a thriving biotechnology industry that is internationally competitive. In the place of bodies of the population in this narrative, we find sensitive genetic information, abstracted from its human sources, political-economic imperatives, and modes of production. We need to take a step back and look again at the bodies of Americans. How do their bodies feature in

discourse about genetic discrimination and personalized medicine?

Useful here is Laurence Cohen's (2004) notion of bioavailability. One of the valuable—and often, scarce—resources critical to build a thriving national genomics enterprise is human bodies (see also Epstein 2007; Lock 2001; Petryna 2005a). They are needed for clinical trials of new therapeutics, and to carry out large population cohort studies. As Margaret Lock reminds us throughout her work on biomedical practices and technologies (see especially Lock 2000, 2001), these practices always have materiality. Individual bodies, which are the sources for aggregates (such as the human genome), are a key element of this materiality. They are both a resource of biomedical practices and policies, and a site of consumption.

Cohen describes kidney-removal surgery and transplantation in India and places the bioavailability of organ donors in tension with "operability." In his research on the commercialization of organ transplantation in India, he is using the term to describe populations of the poor and vulnerable who are seen by organs entrepreneurs as resources for their trade. To be bioavailable is to be available, within the apparatus of state biomedicine, as a body for the extraction and redistribution of human tissue into the bodies of worthy citizens. I use the term to describe the demand for, and scarcity of, American bodies, which are needed to conduct population studies and clinical trials to advance genomics research, commerce, and medicine. Anthropologist Alvarro Jarrin's (2006), who incorporates Cohen's term into his analysis of the stratified cosmetic surgery industry in Brazil, clarifies the meaning of term while providing a sharp contrast for

the state's demand for bodies in the United States. In contrast to Cohen, who sees in bioavailability the possibility of being reduced to bare life (a biological form at odds with rights-bearing citizenship) (Agamben 1998), I suggest that in the United States today, bioavailability and rights-bearing citizenship are closely linked: the assumption that most Americans want to (or should) volunteer their bodies for genomics research is a key component of an emerging genomic citizenship.

A Comment on the Risk Literature

Given that this dissertation explores an aspect of contemporary biopolitics, it might seem appropriate to situate it in the risk (or governmentality) literature. Associated with UK scholars Nikolas Rose and Carolos Novas (see Novas 2006; Rose 2001, 2007; Rose and Novas 2005), and with Australian scholars Deborah Lupton (1995), Pat O'Malley (1996, 2000), and Alan Petersen (Petersen and Bunton 2002), this literature singles out risk identification and management as the salient features of public health and biomedical initiatives. However, while these are the central logics of genetic screening and testing practices, the risk approach is limited in several ways.

One problem with theorizing genomic politics in the United States through the lens of risk and governmentality is that this literature is a response to the growth of neoliberalism in the UK and Australia in the 1980s and the privatization of social services (see especially Peters 2001). I question how appropriate it is to treat the United States as subject to the generic forces of neoliberalism, when privatization of "public" services has a much longer history in the United States than in the UK or Australia.

While the 1980s in the United States did bring a revival of an economic liberalism that favoured greater privatization and de-regulation, an emphasis on the self-regulating properties of the free market, and the promotion of individual self-reliance, the driving forces for the "Reagan Revolution" were different from the neoliberalism that found expression in the UK through Margaret Thatcher and in Australia under Labour Prime Minister Bob Hawke. The moral conservatism behind political and judicial appointments, policies, and spending priorities in the United States during this decade grew out of the rapid ascendancy of a Christian fundamentalism that secured political power—a dynamic that has no parallels in the UK or Australia. Nowhere is the disjuncture between the United States, and the UK and Australia, greater than in the realm of health care insurance, which has never undergone nationalization in the United States. These are significant differences that argue for treating the United States as a distinct society with its own political-economic history and priorities.

Secondly, Rose and Novas single out governance of the life processes of citizens as the central task of the state (see, for example, Novas 2006; Rose and Novas 2005). In so doing, they relegate economic and military priorities to the sidelines. While this may be an artifact of their analytical approach, I am uneasy with an approach that prunes economic and military rationales from biopolitics. A starting point for any representation of genomic politics in the United States is the military origins of the HGP and other large-scale science projects (such as the Manhattan Project and the U.S. space program). An account of genomic politics should also acknowledge that post-Cold War declines in military research and

development, and in technological innovation and manufacturing (in which countries such as Japan, China, India, and Brazil have surpassed the United States), have become key drivers of policy decisions (Du Boff 2003; Fry 2007; White 2005).

A major thrust of the risk literature is the delineation of "governance at a distance" that characterizes neoliberal democracies. Patient support groups have become favoured subjects of studies of governance at a distance (see, for example, Heath et al 2004; Rapp et al 2006; Novas 2006; Rose and Novas 2005; Taussig et al 2003). My fieldwork at the SACGHS hearings confirms what Rose and others have been saying: patient groups and advocacy organizations *have* played an important in shaping public understanding of genetic discrimination as a civil rights problem. They also constitute themselves as particular kinds of subjects in this political economy of hope (Novas 2006). But in studies of genomic politics in the United States, there is still too great a focus on these biosocial forms (Rabinow 1996), and not enough attention to the state's activities and interests. My goal is to show that the state's investment in genomics is a nation-building project. As part of that nation-building project, the state seeks to encourage Americans to consume genetic testing, and to source the bodies of its citizens for research and clinical trials. This is why I do not focus on the workings of patient groups and advocacy organizations, but on public discourse about genetic testing and genetic discrimination. While it might be tempting to regard claims about the rights of Americans to undergo genetic testing without fearing genetic discrimination as yet another instantiation of the pursuit of special interests in a rights-obsessed society, I argue that this discourse must be

seen as part of the state's efforts to build a genomic nation and to secure the participation of citizens in this nation as consumers and research subjects.

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[1] Medicaid is a means-based programme that provides health insurance to low-income Americans (for example, parents, children, seniors, and people with disabilities). It is jointly-funded by the federal government and the states, and administered by the states. Medicare is a federal-funded entitlement programme for the elderly (persons aged 65 years or older). Veterans are also eligible for health care benefits from the U.S. Department of Veteran Affairs based on their status in a priority ranking system. However, a recent study (Himmelstein et al 2007) counters a widely-held belief that all Veterans receive coverage. In fact, 1.8 million veterans are uninsured and lack access to health care.

[2] This five-year period is from 1994 to 1999, when Frankel published his article. However, the issue has remained in Congress, bolstered by genetic nondiscrimination bills sponsored nearly every year since 1995.

[3] Frankel notes that many states did not pass laws, protections at the state level did not include the category of self-insured, and legislation focussed narrowly on genetic tests rather than sensitive information generated by medical exams or family histories.

[4] Frankel identifies the following groups in this effort: *Government/science*: National Advisory Council for Human Genome Research, NIH-DOE Working Group on Ethical, Legal and Social Implications of the HGP, NIH Task Force on Genetic Information and Insurance. *Public advocacy groups/private sector*: American Cancer Society, National Breast Cancer Coalition, Council for Responsible Genetics, Biotechnology Industry Organization, Health Insurance Industry. *Professional*: Institute of Medicine, American Society of Human Genetics. *Public–private partnership*: National Action Plan on Breast Cancer. Sources: http://www.4woman.gov/napbc/catalog.wci/napbc/history.htm, http://www.4woman.gov/napbc/catalog.wci/napbc/closeout.htm, accessed Wednesday, 25 April 2007.)

[5] Here I am drawing on framing theory from the sociological study of social movements and collective action (e.g. Benford 1997; Benford and Snow 2000). Framing is used to explain how social movement actors identify and interpret a problem, attribute blame, and motivate people to act on a particular solution. Collective action frames are interpretive devices that tell people why a particular grievance is not just an unfortunate event but also an intolerable injustice that calls for collective action. They are "action-oriented sets of beliefs and meanings that inspire and legitimate the activities and campaigns of a social movement organization" (Benford and Snow 2000:614).

[6] The National Action Plan on Breast Cancer was formed in 1994 during the Clinton

administration by the U.S. Secretary of Health of Human Services, in response to a petition presented to President Clinton in October 2003 by the National Breast Cancer Coalition for a national strategy to fight breast cancer. It disbanded in 2000.

[7] Other members of the NAPBC that Parthasarathy does not identify are University of Maryland legal scholar and Law School Dean Karen Rothenberg, and Kathy Hudson, director of the Johns Hopkins University-affiliated Genetics and Public Policy Center. Rothenberg and Hudson have been influential activists on genetic discrimination over the last decade.

[8] In the first prosecuted case of employment discrimination, the U.S. Equal Employment Opportunity Commission (EEOC) charged Burlington Northern in 2001 with illegally testing workers for genetic defects. The company had subjected 36 of its employees to genetic tests for carpal tunnel syndrome as part of a comprehensive medical exam, without their knowledge or consent. These employees had lodged claims of work-related carpal tunnel syndrome injuries against the company, and the company threatened disciplinary against those who did not comply with the exam. The EEOC argued that the company's testing programme violated the ADA's prohibition against collecting the DNA from employees.

[9] While Margaret's Lock recent work on patient interpretations of Alzheimer's disease (AD) testing results also falls into this area, I examine her work on AD in my discussion of complexity and uncertainty.

[10] Under U.S. intellectual property law, individuals and groups that isolate and patent a gene are called "inventors," because of the ingenuity required to isolate the gene and identify its function. The reasoning is that a gene isolated by researchers does not exist alone in nature. The isolated gene therefore is not considered a "product of nature" under patent law, but a manufacture. As a manufacture, it is patentable (as long as it satisfies other criteria of patent law that is also new, useful, and non-obvious). This reasoning was made explicit in 1980 with the controversial U.S. Supreme Court decision Diamond v. Chakrabarty, a decision that opened the floodgates in the United States to patenting genes and gene fragments. In the case of Terry's PXE gene invention, the patent was assigned to PXE International.

[11] The publications page of the PMC website features federal agency publications on genomics and links to the agencies' own websites.

Part One:
Roots of Concern

INTRODUCTION

During the 1970s and the 1980s, genetic discrimination did not exist as a discrete problem. But the roots of concern about Americans being denied entitlements based on genetic status were fomenting in what sociologist Troy Duster calls multiple "entry points" to the problem: workplace genetic screening, health insurance, disability rights, and privacy.[1] In this chapter, I examine the earliest of these entry points—workplace screening—against the mandatory sickle cell screening programs of the 1970s.[2]

Both sickle cell screening and workplace screening produced prominent instances of discrimination against individuals because of their status as carriers of benign traits. Yet neither episode provoked a social consensus that Americans were in danger of being treated differently by insurers and employers because of their genetic makeup. However, workplace screening, which came to light in 1980 and was less controversial than sickle cell screening, sowed the seeds of concern that healthy Americans could be denied employment simply for harbouring hidden markers of potential disease. In this chapter, I argue that one of the responses to workplace screening was the identification of genetic makeup as a key marker of difference—alongside race, colour, sex, religion and national origin—that employers were using to deny jobs to workers. This response produced a new category of American who was deemed vulnerable to discrimination: the healthy, asymptomatic individual with a genetic makeup deemed "high-risk."

Workplace screening was also the entry point for geneticist and paediatrician Neil Holtzman to the dangers of genetic screening and testing. His book, *Proceed with Caution* (1989), one of several critiques of the social construction of risk during this period, introduced the public to genetic discrimination as part of a larger problem of the revival of eugenic reasoning. I discuss the events that led to him publicizing his concerns about the deleterious impacts of genetic screening and testing.

1970 – 1981: SICKLE-CELL SCREENING

Genetic screening was routinized in the United States in the 1960s through newborn screening for phenylketonuria (PKU). PKU, a rare autosomal recessive disorder causing mental retardation and seizures, can be detected using a simple biochemical enzyme assay—the Guthrie test, developed by Robert Guthrie in 1960—and controlled with a special diet. Over the objections of the American Medical Association and the American Academy of Pediatrics, as well as state medical societies and researchers in human metabolism, from 1964 to 1975, forty-three states passed laws mandating PKU newborn screening—a test that was promoted in part because it was seen as cost-effective (Paul 1994, 1997).[3] Screening programs for PKU did produce reports of stigmatization and insurer discrimination (Holtzman 1989).[4] But it was not until the following decade, with the proliferation of well-intentioned but disastrous sickle cell screening programs, that a clear pattern of discrimination related to genetic diagnosis emerged in the form of denials of insurance and jobs, and increased insurance premiums.[5]

Sickle Cell Screening

Sickle cell anemia is an autosomal recessive blood disease that follows Mendelian patterns of inheritance. Carriers, who inherit one copy of the sickle cell beta-hemoglobin gene (HbS) have sickle cell trait and remain healthy. Those who inherit two copies of the variant can experience a range of health problems that include painful haemolytic sickling crises and a shorter life span.

Following the successful implementation of mandatory newborn screening and pressure from black leaders after the civil rights movement to improve the health of African-Americans, states embarked on a new experiment in genetic screening: mandatory sickle cell screening of African-Americans. Between 1970 and 1972, nine states and the District of Columbia passed laws mandating sickle screening, making sickle cell screening the first instance in the United States of mandatory carrier screening (Rutkow and Lipton 1974). Four states also passed non-mandatory screening laws (Rutkow and Lipton 1974). By 1973, thirty-four states had implemented mandatory and voluntary screening services (Schmidt 1974). These programs largely targeted African-American schoolchildren (Markel 1997), but also targeted newborns, couples applying for marriage licenses (e.g. New York State), and inmates (in Virginia) (Rutkow and Lipton 1974).

The Federal Government also responded to pressure from black leaders to improve the health and well-being of African-Americans.[6] In 1971, the Nixon Administration allocated $6 million for research into sickle cell anemia, and held Senate hearings on the merits of a national sickle cell anemia program (Markel 1997). The following year, the Administration

passed the National Sickle Cell Anemia Control Act, "to TTreverse the record of neglect on this dread disease" (U.S. Congress 1972). Describing sickle cell as a "debilitating menace to many Americans," the Act provided $115 million over three years for screening, counselling and education, through voluntary-only programs (Curran 1974).

Problems with Screening Programs

Unlike newborn screening, sickle cell screening programs were fraught with problems that became evident in delivery and impact. From a contemporary perspective, it is remarkable how widespread was the ignorance amongst the public, clinicians, community leaders, politicians, and legislators about differences between the carrier status and the homozygous state. The use of the Sickledex haemoglobin solubility test only confirmed the presence of sickling: it did not distinguish between the heterozygous and homozygous states (Markel 1997). Consequently, healthy carriers were told they had the disease when they had the trait, and they were regarded as if they were ill or prone to serious health problems (e.g. Bowman 1977; Murray 1972). Inadequate patient counselling reinforced this error, as did unfamiliarity with the trait and disease in the general population (Bowman 1977; Motulsky 1974; Powlege 1972; Schmidt 1974). Even the text of the 1972 National Sickle Cell Screening Act demonstrated confusion between the trait and disease forms (Bowman 1977). Other problems were the absence of privacy and confidentiality protections (e.g. Hilton 1972; Kenen and Schmidt 1978), the lack of effective treatments for sickle cell, and the dubious merits of public health spending on sickle cell when African-Americans faced more

pressing health problems (e.g. Murray 1972).

The social impacts of conducting a mandatory genetic screening program in the absence of an adequate social understanding of genetic inheritance and expression were disastrous. Some carriers of sickle cell trait were refused life and health insurance; others faced increased health insurance premiums after they had been diagnosed. Schoolchildren who were identified as carriers were prohibited from participating in sports by some schools. Volpe (1984:43-44) observes that some people believed that genes were germs, and that individuals with the trait were carrying an infectious germ. Georgia legislators, for example, believed that sickle cell was a communicable disease that could be fought with immunization.

Attention came to focus on problems with employment, particularly in aviation. Carriers were denied jobs, reassigned, or fired. Major airlines grounded or dismissed African-Americans flight attendants with sickle cell trait (Bowman 1977). Although all branches of the U.S. military prohibited sickle cell carriers from pilot and flight crew training (Draper 1991), the Air Force Academy practices the one identified most often as egregious. These practices eventually prompted legal action. Beginning in 1972, after two cadets with sickle cell trait became ill after training at 7,000 feet, the Academy banned African-American carriers from entering as cadets (Severo 1981). The Academy based its exclusion policy on a flawed study from the National Academy of Sciences (1981) that recommended all recruits be screened for sickle cell trait and disease before starting basic training (Duster 1990). The Academy argued that working conditions for its pilots, who flew at high altitudes under low oxygen, were too dangerous

for sickle cell carriers (Bowman 1977; Draper 1991). From 1972 to 1981, the Air Force Academy turned down forty-two candidates with sickle cell trait (Hoffman and Slade 1981). In 1979, it rejected six African-Americans with sickle cell trait, including 20-year old Stephen Pullens. Pullens, a talented athlete who had qualified for the pilot trainee program, was rejected from the Air Force Academy after a blood test during a mandatory physical examination revealed he was carrying sickle cell trait. The *New York Times* (1981) reported the story and quoted Pullens:

> "'I was told by a doctor that I might die if I stayed,'" Mr. Pullens said. "I asked for a second test, which also was positive, and at that time I was told to leave. I was given an escort to assure I would not discuss my case with anyone and was denied further appeals."

A class-action lawsuit field by Pullens in 1981 forced the Air Force Academy to revoke its policy (Severo 1981).

Criticism of Screening Programs

Black leaders, who had pushed for the screening programs, roundly condemned them, as did medical professionals, bioethicists and lawyers (e.g. Bowman 1977; Curran 1974; Hampton et al 1974; Hilton 1972; Kenen and Schmidt 1978; Motulsky 1974; Murray 1972; Powlege 1972; Rutkow and Lipton 1974; Schmidt 1974). Public commentary on the screening programs, which were implemented less than ten years after passage of the 1964 Civil Rights Act, communicated anger, embarrassment, disgust—and shame. In fact, it is difficult to find anything to praise about the programs. Powlege (1972:3), for example, pointedly criticized the proliferation of new state screening laws, which she attributed to "the recent availability of a

cheap, easy, mass testing process that is fairly reliable:"

> Many of the laws may be unconstitutional; an appalling number are biological nonsense. They have spread as rapidly as the phenylketonuria laws a few years ago, and with equally good intentions, despite sporadic opposition from the profession. (Powlege 1972:3)

Critics blasted the inherent racism of the legislation and programs (e.g. Powlege 1972; see also Markel 1997), because they targeted African-Americans to the exclusion of other minorities who also had an elevated incidence of the trait. Again, it was Tabitha Powlege (1972:4) who suggested that the mandatory screening programs functioned as a proxy for a "racial classification test" (see also Markel 1997):

> In addition to being of doubtful constitutionality and genetic content, many laws pussyfoot around the prickly question of race to an almost comic degree. One New York State law, for instance, mandates the testing only of *urban* school children. ... New York also has a marriage license law; *it* mandates testing of "each applicant for a marriage license who is not of the Caucasian, Indian or Oriental race"—which is a bit of anthropological mumbojumbo that doesn't even specify which kind of Indian. Perhaps the legislators meant both. Laws in some states—Virginia is an example—shift responsibility by letting the physician decide who should be tested, thus taking the onus of racial classification off the legislators, or so they hope. (Powlege 1972:4)

Early Focus on Stigmatization

Critics initially focussed on the psychosocial and economic consequences that resulted from confusing the trait for with the disease. These consequences were compounded by inadequate genetic counselling to help patients, institutions and the public understand the significance of

their test results. At the beginning of the 1970s, critics talked broadly in terms of "stigmatization" rather than discrimination. They described the psychosocial impact of positive test results on self-esteem, as well as disadvantages in marriage and employment prospects, and schooling (e.g. Motulsky 1974; Powlege 1972). For example, medical geneticist Robert Murray (1972:11) worried about what he called "the dilemma of the carrier" as the psychological impact of a positive result on carriers:

> But what of the person detected as the carrier? He is usually unprepared for the disappointing news that he receives unless he already knows that he has a positive family history. Even individuals who are knowledgeable about a particular condition, like for example sickle cell trait, may well be unable to readily accept the news that they are a carrier. (Murray 1972:11)

Stigmatization was also the primary concern of the media and the public in the early part of the decade. A December 1972 letter to the editor in the *New York Times* by Alene B. Bennett (Bennett 1972) illustrates how the concern with stigmatization as an effect that encompassed children as well as adults, and extended beyond insurers and employers to schools and neighbourhoods:

> As a black physician and mother I have had personal experience with the whole supermarket mentality surrounding this new craze to perform genetic screening for sickle cell anemia. ... How dare the Federal and state officials and politicians legislate genetic screening for the most unpopular minority in this country. It will be many generations before this country is racially matured and educated enough to screen any group for any disorder. ... As a black physician I would refuse to engage in genetic screening of school-children and I would discourage parents from participating in these stigmatizing programs. As a black mother of a school-age child, I would file the first suit if such genetic screening becomes law in the state in which I

> reside, because such activities are stigmatizing; an invasion of privacy; and must be unconstitutional. This is without a doubt the most dangerous thing to hit the black community yet. (Bennett 1972)

The Shift towards a Discourse of Discrimination

Towards the mid-1970s, with increasing evidence that African-Americans with carrier status were being treated as diseased or ill by insurers and employers, commentary shifted away from the psychosocial effects of a positive diagnosis and towards denials of entitlements for African-Americans. "Discrimination" has a specific cultural and legal reference. Following the civil rights movement of the 1950s and 1960s, the Civil Rights Act was passed explicitly to outlaw segregation in schools and public places. Title VII of the Civil Rights Act prohibits employment discrimination on the basis of race, colour, sex, religion and national origin. To critic of sickle cell screening, the blatant treatment by insurers and employers of African-Americans as "diseased" embodied the very problems that the Act was intended to rectify.

Rutkow and Lipton (1974:218) noted that that "persons found to have sickle cell trait, which many experts believe to be an essentially benign condition (except under extremely unusual circumstances), can have trouble receiving life insurance and health insurance, and some have been discriminated against on the job." Claims of discrimination were authenticated by the National Research Council's Committee for the Study of Inborn Errors of Metabolism. In 1975, the Committee published an extensive report reviewing the history, implementation, and ethics of genetic screening practices in the United States for diseases such as PKU, sickle-cell, Tay Sachs, and thalassemia, and Down Syndrome (using

amniocentesis). In the section on sickle-cell, the authors summarized discrimination by employers and insurers, as well as stigmatization of schoolchildren:

> Evidence of stigmatization in the United States is seen in job discrimination, in proposals to limit admission to the armed forces to noncarriers, and in increases in insurance premiums. Nine of twelve insurance companies in one sample charged higher rates for individuals with sickle cell trait even though mortality curves for such individuals do not differ significantly for blacks without the trait. The screening of school-age children for sickle cell trait is subject to particular hazards, since the natural cautions and fears of parents and teachers may lead to unnecessary but unavoidable restrictions on activities and unconscious but irreversible curtailment of expectations for performance and achievement. All of these results of "labeling" therefore reflect misunderstanding about the significance of being a carrier. (National Research Council 1975: 126)

And in 1977, African-American physician James Bowman (1977), the most outspoken commentary on sickle cell screening programs, produced the first lengthy analysis of the Air Force Academy's decision to ban carriers from pilot training and flight crews.

While most critics focussed on the racialized aspects of discrimination tied to sickle cell screening, some were more concerned with the actuarial and eugenic reasoning that informed screening, and the denials of entitlements to asymptomatic, healthy carriers. Twenty-five years before Neil Holtzman (1989) and Troy Duster (1990) sounded their warning that genetic screening and testing practices were constructing categories of high-risk persons, lawyer and public health critic William Curran (1974) suggested that all screening was driven by the desire to remove defective persons from the population because they were costly to institutions:

> It can also be said that the thrust of all genetic screening and counseling programs of a public, legal nature, whether compulsory of not, leaves an aftertaste of genocide, of a desire to rid the world of imperfect people, particularly those who may become a burden on society (here read: "on my tax bill") through hospitalization or institutionalization for long periods of time. (Curran 1974)

The injustice that Curran perceived was not racial in nature. Rather, it took the form of the denial of entitlements to individuals who were neither handicapped nor diseased—because their genetic differences might cost institutions money. Another warning voice was that of Hastings Center ethicist Bruce Hilton (1972:9). He did not phrase his criticism quite as strongly as Curran (who suggested that screening programs were genocidal). But he did use the language of genetic defect to suggest that each person had several flawed genes—a message that genetic activists would deliver thirty years later, for a different purpose—and warned that all Americans should consider themselves vulnerable to the abuse of their private medical information:

> Since each of us has on the average three to five deleterious genes, all of us are potentially affected by such questions as: Who owns the information about our genetic shortcomings? Who has access to it? Does the doctor have a duty to inform our relatives of what he finds, since they may also be affected? If he does so, can we sue him for breach of confidence? Privacy is an increasingly slippery commodity, which does not, despite popular belief, enjoy the full broad protection of either common law or the Constitution. Thus, it should be no surprise when we find that our medical records are already likely to be in the hands of school administrators, the Army, several life insurance companies, the Blue Cross, and of course, the family doctor and several specialists. (Hilton 1972:9)

Physician James Bowman (1977) feared the reprisal of negative eugenics

by insurers should voluntary programs fail to reduce the incidence of sickle cell among African-American children:

> Since many mass education, testing and counseling programs are formed under the assumption that there will be a decrease in the number of matings and consequently offspring with sickle cell anemia, I fear that when this does not take place, that more stringent proscriptions will follow, such as sterilization of carriers or mandatory abortion. This is in the realm of reality. There have also been suggestions that insurance companies may demand higher premiums in certain families unless women in high risk families submit to amniocentesis and agree to abortion of affected fetuses. (Bowman 1977:136)

Tay-Sachs screening, by contrast, suffered from none of the problems of sickle cell screening, despite the fact that it targeted Ashkenazi Jews, another vulnerable and historically stigmatized population. Screening for Tay-Sachs began in 1970. However, the screening programs were voluntary from the start. Also, they were delivered under a different model than sickle cell screening. Community leaders worked alongside physicians and genetic counsellors, and emphasized informed decision-making (Duster 1990; Goodman and Goodman 1982). The comparative success of Tay Sachs screening bolstered the perception that the problems associated with sickle cell screening were rooted in a misplaced enthusiasm for mandatory genetic screening of a stigmatized population (African-Americans), rather than genetic screening per se.[7] Workplace screening, however, would challenge the strong association of discrimination with race and ethnicity, and produce an understanding that something called "genetic makeup" in workers could be a basis for discrimination—along with race, colour, sex, religion and national origin.

1980 – 1990: WORKPLACE SCREENING

Workplace susceptibility screening came to light in 1980 when *New York Times* reporter Richard Severo revealed that some of the country's largest employers were routinely screening workers for genetic traits associated with hypersusceptibility to industrial disease. In a four-part feature published in 1980, Severo reported that chemical companies had denied jobs to workers who screened positive for genetic traits thought to confer hypersusceptibility to industrial disease (Draper 1991; Severo 1980a, 1980a, 1980c, 1980c). The federal government investigated and found the practice to be more widespread than Severo's series suggested.

Background

Genetic models of occupational risk date to the first half of the twentieth century, when scientists and industry officials proposed links between occupational disease and inherited abnormalities (Draper 1991:23-24). British geneticist and evolutionary biologist J.B.S. Haldane (1938) is credited with promoting the screening of applicants to exclude susceptible workers from hazardous workplaces (Gochfeld 1998). However, Haldane emphasized improving working conditions along with implementing susceptibility screening (Murray 1983). Moreover, industrial hygienists of the era endorsed environmental and social interventions, rather than interventions directed at the individual (Draper 1991).

In the 1960s and 1970s, this public health model gave way to an individualizing framework that privileged individual genetic susceptibility to occupational disease as the salient risk factor in occupational disease

(Brandt-Rauf and Brandt-Rauf 2004; Draper 1991).[8] Occupational genetic screening received a boost from pharmacogenetics research from the 1950s, when researchers discovered that American soldiers in Korea with the inherited metabolic disorder Glucose-6-Phosphate Dehydrogenase (G6PD) developed hemolysis to the anti-malaria drug primaquine (Murray 1983). G6PD, which confers resistance to malaria, occurs at a higher frequency in African-Americans and people with Mediterranean ancestors. The reaction was labelled a "hypersusceptibility." The rationale for workplace susceptibility screening was based on this finding:

> By the early 1960s, a handful of prominent American toxicologists had drawn the straightforward analogy from drug ingestion to chemical exposures in If drugs can cause hemolysis in people with G6PD, isn't it likely that chemicals with similar structures and toxicological properties will have the same effect? (Murray 1983:5)

Herbert Stokinger (Stokinger and Mountain 1963, 1967; Stokinger and Scheel 1973; Stokinger et al 1968), chief toxicologist for the U.S. Public Health Service, endorsed hypersusceptibility testing for applicants to the chemical and manufacturing industries, to help management with its bottom line (Draper 1991; Gochfeld 1998). Stokinger and his fellow toxicologists, who argued that "the industrial physician could employ to advantage such tests to distinguish heredity-based disease from job-claimed disability" (Stokinger et al 1968:970), promoted three traits for workplace screening: sickle cell trait, glucose-6-phosphate dehydrogenase (G6PD) deficiency, and serum alphaRR_1-antitrypsin (SAT) deficiency. [9]

In 1980, nearly a decade after companies in the chemical and manufacturing industries had implemented pre-employment genetic

screening, *New York Times* journalist Richard Severo brought the practice to the attention of the American public. In a four-part, front-page series called "The Genetic Barrier: Job Benefit or Job Bias," Severo (1980a, 1980b, 1980c, 1980d) described two controversial screening practices in place since 1972 at E.I. Du Pont and since the mid-1970s at Dow Chemical Company, two of the country's largest chemical employers. At Du Pont, applicants were subject to a one-time, pre-employment susceptibility screening; at Dow, workers endured genetic monitoring for signs of chromosome damage.[10] Severo reported that Du Pont screened African-American applicants for sickle cell trait, thought to heighten susceptibility in workers to occupational disease such as anemia in the presence of compounds such as oxidizing aromatic nitro and amino, and dyes. Through a practice called cytogenetic screening, Dow monitored existing workers for signs of chromosome breakage to assess genetic damage from exposure to carcinogens such as benzene. Other companies screened for G6PD deficiency and SAT deficiency (Draper 1991). The companies also implemented fetal exclusion policies, barring fertile women in their reproductive years from working in production jobs with suspected teratogens (Draper 1991).[11]

Controversial Policies

The screening practices were controversial for different reasons. A common criticism was that the screening practices shifted responsibility for occupational disease from management to the workers, thereby penalizing healthy applicants, and leaving healthy workers vulnerable to a dirty workplace. Scientists were sceptical of claims linking genes with

hypersusceptibility to industrial disease, and neither Du Pont nor Dow could provide data demonstrating the utility of the screening practices and the benefits to workers, despite claiming to have screened thousands of workers during the 1970s. Du Pont scientists and management offered contradictory accounts for the origin and rationale of their pre-employment screening policies, but defended the policies as voluntary and not contested by African-Americans.

In fact, the impetus to establish sickle-cell pre-employment screening came from Dr. Alston Meade, a research biologist who also headed Du Pont's Black Employees Association (Severo 1980e, 1981b). After the Nixon Administration passed the National Sickle Cell Screening Act in 1972, Meade wrote to Du Pont's public relations and employee relations departments, requesting the company test all existing African-American employees for sickle cell anemia (Severo 1980e, 1981b). The company decided instead to institute pre-employment screening of African-Americans for sickle cell trait, not sickle cell anemia.

Genetic monitoring, developed by Dow scientist Dr. Jack Killian, took the form of research studies whose sole beneficiaries appeared to be Dow scientists. Workers whose chromosomes showed signs of breakage after working with substances such as benzene were not informed of the results. Dow scientist Dr. Dante J. Picciano left the company after management refused to let him tell workers about their results, reduce benzene levels, or report the findings to regulators and the petrochemical industry (Severo 1980a).

Why were the Policies Discriminatory?

Were the policies considered discriminatory? The answer is yes, but for different reasons. Some critics saw pre-employment susceptibility screening as an instance of racial discrimination because most of the traits selected for screening are found disproportionately higher among minorities.[12] For example, Thomas Murray, a Hastings Center bioethicist who has written extensively on insurer and employer discrimination throughout the 1980s and 1990s, expressed this view:

> Genetic traits naturally fall along racial lines. When the trait in question occurs disproportionately often among members of historically mistreated groups, there is likely to be suspicion and mistrust on the one hand, and a feeling that this is just one more obstacle placed in the way of fair and equal treatment. We should scrutinize with great care any exclusionary screening program having a focus or disproportionate impact on such groups. (Murray 1983:8)

Severo's own reporting was probably the most influential in shaping public perception that the policies were discriminatory. Because his articles focussed on the selective application of sickle cell screening to African-American applicants by Du Pont, a practice that resembled the Air Force Academy's policy of rejecting sickle cell carrier trainees, his accounts portrayed susceptibility screening as a clear-cut case of racial discrimination. Subsequent analysis reinforced this portrayal. For example, sociologist Elaine Draper, who has analyzed workplace screening as an example of the social construction of occupational risk (1991, 1993), has argued the policies were discriminatory because companies applied them selectively, using race, ethnicity, and sex as naturalized markers of risk (see also Holtzman 1989). Companies adopted genetic screening for

traits that are found at a higher incidence among certain ethnic populations (such as sickle cell and G6PD deficiency among African-Americans, or Beta thalassemia trait among Italians and Greeks) only "when a group is relatively new to a specific workplace or occupation" (Draper 1991:83-85). Such was the case when African-Americans entered the chemical industry in large numbers in the 1980s, and companies began screening for sickle cell trait and G6PD deficiency.[13]

But not all scholarly analysis has treated susceptibility screening as a racially discriminatory practice. Legal scholar Mark Rothstein (1983:1491), for example, has argued that susceptibility screening created a category of "high-risk" persons. The salient trait of these high-risk persons, according to Rothstein, is not that they were historically-mistreated minorities, but that they were healthy applicants with no discernible illness or handicap:

> When an individual's present physical condition, even with reasonable accommodation, precludes the performance of an essential job function it is neither illegal nor unfair for the individual to be denied that particular job. High-risk persons, however, are generally healthy and currently able to perform the job. If they are to be denied employment on some rational basis, it must be because of an unacceptable risk of future illness. (Rothstein 1983:1491)

Rothstein's concern, that workplace screening discriminated against the healthy but high-risk person, would be picked up and developed by the U.S. Congress Office of Technology Assessment (OTA) the following year, in its report on industry susceptibility screening (OTA 1983).

Fetal exclusion policies, although they did not involve genetic screening, were considered to be an egregious example of sex

discrimination. Forty workers—all women—filed complaints with the federal Equal Employment Opportunities Commission (EEOC) against a host of companies, including American Cyanamid Company, for excluding them from jobs working with reproductive hazards (Severo 1980a). In 1977, American Cyanamid instituted a policy excluding women "of childbearing capacity" (Draper 1991:13) from jobs that would expose them to chemicals that to harm the fetus at its Willow Island, West Virginia plant since 1977 (Uzych 1986). The company required that female production workers had to "provide evidence of permanent infertility by April, 1978" or transfer to another department. Thirty women were affected by the policy but only seven transfer positions were available (Uzych 1986:39). In response to this policy, five female American Cyanamid employees underwent sterilization to work at these jobs, "which, as it turned out, the company later eliminated altogether (Draper 1991:13). The women chose to undergo sterilization only because their jobs were threatened by the company's policy. According to Severo (1980a), "[r]eports of women having themselves sterilized so they could continue to work were not new. What was new was that the company had allegedly made sterilization a formal part of employment policy." [14]

Impact of the Severo Series: The U.S. Congress Responds

Severo's reporting brought workplace screening to the attention of the U.S. Congress (Draper 1991; Uzych 1986). In 1981 and 1982, the U.S. Congress Committee on Science and Technology held hearings on the practice (U.S. Congress 1981; U.S. Congress 1982a, 1982b). The Committee engaged the now-defunct U.S. Office of Technology Assessment (OTA), a

bipartisan agency that provided technology analysis and policy advice to Congress, to conduct a survey of 500 of the largest industrial companies in the U.S., fifty large utilities, and the presidents of eleven unions that represented the companies' workers (Uzych 1986). Of the 366 Fortune 500 industrial companies that responded to the 1982 OTA questionnaire (U.S. Congress, Office of Technology Assessment 1983), six companies (1.6%) indicated they were currently using either pre-employment susceptibility screening or cytogenetic monitoring; seventeen companies (4.6%) had used these tests within the past twelve years (between 1970 and 1982); and fifty-nine companies (16.1%) reported plans to use the tests or consider using them (Draper 1991; Uzych 1986).

Unlike Severo's reporting, which had focussed on screening of African-American applicants (and women, for fetal exclusion policies) and portrayed the Du Pont and Dow practices as racial and sex discrimination, the OTA survey found that race and ethnicity were criteria only for sickle cell trait screening. For screening of all other traits, "employees were selected on the basis of job category" (U.S. Congress, Office of Technology Assessment 1983:9). This is a critical piece of information missing from Severo's representation.

The authors of the OTA report assessed the merits of workplace screening. The authors concluded the ability of genetic screening "to predict future disease has not been demonstrated" (U.S. Congress, Office of Technology Assessment 1983:15), but they were particularly critical of the tacit claim of proponents of workplace screening, that it could identify those at risk of "future illness." While acknowledging that genetic makeup

could indeed be a risk factor for future illness, along with "age, sex, medical history, nutritional status, lifestyle" and "prior exposure to hazardous agents" (U.S. Congress, Office of Technology Assessment 1983:7), they cautioned that genetic testing was in too early a developmental stage to accurately predict which individuals within high-risk groups might be at increased risk of developing occupational disease. The authors indirectly raised the question of whether employers should be entering the divination business. If the courts decided that risk of illness was a valid concern for employers, said the authors, then it would be up to employers to demonstrate that "genetic screening techniques were reasonably predictive of illness" (U.S. Congress, Office of Technology Assessment 1983:13).

The most important aspect of the report was the introduction of a new category of difference in its legal and policy analysis of whether genetic screening of applicants constituted discrimination. The authors of the OTA report singled out "genetic makeup" predictive of future illness as the criterion on which employers were discriminating. They noted that like other markers of difference protected under the Civil Rights Act, genetic makeup was not within the control of workers. This language marks an important shift in expanding the scope of intractable biological markers of difference beyond race and ethnicity to include genetic makeup and the prospect of future illness or disability.[15]

Response of the American Public Health Association

The American Public Health Association (APHA) also responded to the report, taking a strong position against workplace screening. In 1984, the

APHA it released a policy statement (American Public Health Association 1984:281), titled "Guidelines for genetic testing in industry." The APHA policy statement drew on Severo's reporting, the 1981 and 1982 Congressional hearings, and the 1983 OTA findings. In a statement acknowledging workplace genetic screening put workers at risk of discrimination, the APHA recommended that industry "cease genetic testing for inherited genetic traits for the purpose of job exclusion," and urged the development of ethical and scientific guidelines for workplace genetic testing (American Public Health Association 1984:281). Nowhere does the APHA policy statement mention racial or ethnic discrimination. The focus is simply on workers who are not presently handicapped or ill. Like the 1983 OTA report, the 1984 APHA statement notes the absence of protection for genetic testing under Title VII of the Civil Rights Act and the 1973 Rehabilitation Act (which offered some protection to handicapped workers). This comment suggests that the APHA viewed workers with genetic hypersusceptibility to occupational disease as a vulnerable population in need of legislated protection.

1987 – 1991: CRITIQUING THE SOCIAL CONSTRUCTION OF GENETIC RISK

The end of the 1980s and the early 1990s saw the publication of several analyses of the social impact of genetic testing and screening technologies: Lori Andrews's TT*Medical Genetics: A Legal Frontier* (1987), Neil Holtzman's *Proceed with Caution* (1989), Dorothy Nelkin and Laurence Tancredi's *Dangerous Diagnostics* (1989), Troy Duster's *Backdoor to Eugenics* (1990), and Elaine Draper's *Risky Business* (1991). These authors, whom I call "the risk critics," warned that the rapid adoption of

genetic testing and screening technologies, and the growing embrace of genetic explanations for health and social problems, were producing categories of high-risk individuals and groups who were seen as the source of their own health problems. In this section, I focus on Neil Holtzman's *Proceed with Caution*, identifying the practices and events that led him to worry about discrimination. I also bring in Troy Duster's perspective on the difficulty of defining genetic discrimination as a problem.

Neil Holtzman and *Proceed with Caution*

It is unlikely that policy analyses of workplace screening had any impact on public awareness about genetic makeup as a basis for discrimination by employers. Troy Duster, for example, believes that the Severo series and the Congressional investigation had a "minimal" impact on public consciousness, both at that time, and today:

> People who knew, knew. People in the field knew about that series. I would say, even people who are now talking about health insurance and genetics, most of them would not have known what went on in that period. (Interview with Troy Duster, November 30, 2007)

However, public attention to workplace screening, along with Severo's series, did have a profound impact on one person: Johns Hopkins geneticist Neil Holtzman. It was Holtzman who would introduce the public to the problem of genetic discrimination in 1988 and again in 1989, although not by that name. Although credit rightly goes to Paul Billings and his colleagues for naming the problem of genetic discrimination in a 1992 *American Journal of Human Genetics* (*AJHG*) article (Billings et al 1992), it was Holtzman's work that seeded the ground for public reception

to their arguments. Holtzman, unlike commentators following him, would also explain the actuarial reasoning that guided health insurers' decision-making.

Retired in 2005 from Johns Hopkins School of Medicine and School of Public Health, where he was professor of pediatrics and genetics, Holtzman is best known for his book on the impact of recombinant DNA technology and the unintended social impacts of the growth of genetic testing for common, multifactorial disorders. *Proceed with Caution* (Holtzman 1989) offered a stark warning to geneticists, clinicians, and bioethicists: as medical treatment costs rise and as genetic testing becomes a profitable industry, the ability to test for the presence of common genetic "defects" in otherwise asymptomatic persons, with no treatment available for these diseases, would lead to increased pressure on individuals to conduct prenatal testing and abort fetuses carrying mutations. It would also lead, warned Holtzman, to denials of insurance and employment to individuals carrying these mutations. Holtzman's message, that the technological ability to identify an individual's risk of genetic disease had vastly outstripped the therapeutic capacity to treat most of these conditions, was directed at the United States, where enthusiasm for technological innovation and commercialization of genetic testing were developing in a climate of cost-cutting. The insurance industries (health and life) and employers were looking for incentives to reduce their expenses and risk exposure.

Even before Holtzman published *Proceed with Caution*, he set out in a short article in the *AJHG* (Holtzman 1988) the growing problem of

insurers using genetic testing information to lower their costs and increase their competitiveness:

> People suffering from genetic or gene-influenced conditions whose management is costly (frequently because no definitive treatment is available) cannot obtain individual medical, disability, or life insurance at the standard rate... Insurance companies could deny or limit insurance provided to apparently healthy people in whom genetic tests predict the occurrence of these or similar conditions. By excluding such individuals from the standard class, insurers could lower the standard rate, thereby gaining an advantage on competitors. Therefore, once one company starts to require the results of tests, others are likely to follow suit. (Holtzman 1988:629)

In the article, Holtzman identified nine "conditions with genetic components" that were deniable by health and disability insurers: sickle cell anemia, aplastic anemia, angina pectoris, arteriosclerosis, Huntington chorea, insulin-dependent diabetes, Down Syndrome, polycystic kidney, and muscular dystrophy. This was the first concrete enumeration is in a clinical genetics journal of discriminatory practices by insurers towards not only carriers of sickle cell trait, but individuals suffering from rare, single-gene disorders. However, it was Holtzman's book *Proceed with Caution* that brought to the attention of a wider audience the looming threat of discriminatory practices against otherwise healthy individuals.

In his book, Holtzman made a crucial link between the radical technological innovation that had swept the field of biology in the 1970s and the 1980s, and the increasing availability of genetic tests that was also increasing the vulnerability of Americans to being labelled at-risk. Holtzman singled out recombinant DNA (rDNA), developed in 1973 by Stanley Cohen and Herbert Boyer as the pivotal technology that made

modern genetic diagnostics possible (Holtzman 1988).

rDNA technology, which cuts human DNA into small segments and uses microorganisms to quickly clone the DNA segments into large amounts, allowed researchers to transform human DNA into DNA probes which could be used to locate individual variation in DNA sequences. These DNA sequences variations could then be used as markers, allowing researchers to build genome maps and identify the specific locations on chromosomes of genes responsible for inherited disorders. The introduction of rDNA technology, which allowed biologists to develop probes to locate known mutations, in particular marks a turning point towards "modern" genetics.[16] Before the adoption of rDNA, tests for "genetic" diseases, such as sickle-cell anemia, Tay-Sachs disease, and PKU were widely not considered to be genetic tests.[17] In his account of the transformation of post-war biology, Sheldon Krimsky (1999) argues that the invention of rDNA technology in 1973 radically transformed biology from an analytic to a synthetic science. This invention oriented scientists and research bodies towards the commercialization of molecular biology, which was realized through increasing partnership between academic scientists and industry.

rDNA is arguably the innovation with the greatest single impact on the development of genetic diagnostic tests, but two other innovations—positional cloning and the polymerase chain reaction, or PCR—also magnified the potential of molecular biology to generate clinical applications. These technologies, like rDNA, helped pave the way for the aggressive commercialization of genetic testing through the growth of

private laboratories and biotechnology companies such as Genentech (Krimsky 1999). Positional cloning, developed in 1980 by four researchers (David Botstein, Ron Davis, Ray White, and Mark Skolnick, the founder of Myriad Genetics), was an early genomic-sequencing technique that used restriction fragment length polymorphisms (RFLPs) to map the entire human genome and locate specific genes. PCR technology, which was developed by Cary Mullis in 1983, allowed researchers to amplify specific segments of DNA, making billions of copies for analysis. Other developments which also accelerated during the 1980s include the production of the first human genetic map using restriction enzymes, which helped to locate genes responsible for disease (1987); the use of microsatellites, which are repetitive DNA sequences, as genetic markers (1989); and the development of expressed-sequence tags (ESTs) to zero in on the "expressed" sequence of a genome (1991). These developments expanded the number of genetic tests available for rare diseases that lacked treatment, such as sickle cell disease, Huntington Disease (HD), Cystic Fibrosis (CF), and Tay Sachs Disease (TSD). These techniques accelerated the search for susceptibility genes for common diseases, such as breast cancer, prostate cancer, colon cancer, and Alzheimer's disease.[18]

It was a series of twists and turns over the course of Holtzman's career from 1962 when he finished his pediatric residency to retirement in 2005, rather than a single event, that aroused his concern about the social impact of runaway genetic testing in the United States. While acknowledging his role in putting the problem of genetic discrimination before the public, he portrays himself as a reluctant geneticist, insisting that genetics was a small part of his career, something that he kept getting "dragged back

into."[19]

Holtzman says that the problem of genetic discrimination "really antedated the [Human] Genome Project." "Its roots go *way* back," he says. And I think I contributed to making it an issue [laughs]." (Interview with Neil Holtzman, May 31, 2007).

In conversation, Holtzman alludes to sickle cell screening as the first instance of genetic discrimination. He was, after all, a member of the Committee for the Study of Inborn Errors of Metabolism, which produced the National Research Council (1975) report on genetic screening. However, he dates the roots of his own awareness of genetic discrimination to events in the 1980s. Holtzman singles out the Severo series as providing the public with the first substantial media exposure to a problem that would be called genetic discrimination starting in the 1990s. He also credits Severo's reporting with raising his own awareness that genetic discrimination was a threat to Americans beyond specific populations, such as African-Americans and Jews who were the targets of sickle cell and Tay Sachs screening programs:

> That was a very impressive series. It certainly influenced my thinking, that series. And it is a very important link, to screening in the workplace. I think the workplace is a critical issue. (Interview with Neil Holtzman, May 31, 2007)

Well before the Severo series appeared, Holtzman had become active in shaping public policy and law on genetic screening. In the early 1970s, he left bench science at Hopkins, where he had been working on Wilson's disease, a rare genetic disorder, to become a primary-care physician at an

HMO for foster children in east Baltimore. There he cared for children with phenylketonuria (PKU). PKU was the first disorder that was subject to routinized genetic screening in the United States, and its success encouraged the institutionalization of other screening programs, such as sickle cell and Tay Sachs, in the 1970s. During this time, while working as a primary-care physician, Holtzman accepted an invitation from the Baltimore and Maryland State Department of Health and Mental Hygiene Health to run its Division of Hereditary Disorders and develop screening programs. As states began drafting laws on genetic screening for sickle cell, thalassemia, and Tay Sachs in the early and mid-1970s, however, Holtzman says that he became concerned about their rush to mandate genetic screening:

> As diseases came up there was this tendency to pass laws about them. And it wasn't long before I realized it was a pretty bad thing to do. So I got involved in rewriting the Maryland law in 1975. (Interview with Neil Holtzman, May 31, 2007)

Also at this time, Holtzman became interested in the problem of laboratory proficiency for genetic testing for PKU. In a 1992 interview, Holtzman explains:

> Now, about the time that I was finishing my training and doing this, there was a large collaborative project organized in the United States in which our University, Johns Hopkins, was asked to participate to look at the effectiveness of therapy for genetic disease.
>
> The genetic disease was phenylketonuria (PKU) for which at that time all newborns essentially were being screened in the United States. And a very puzzling thing had come out of the early findings of that project. This is a disease that you would expect to find equal numbers of males and females, but in the first 90 children who were identified

> through population-based screening, for PKU, there were twice as many males as females. And this was very puzzling and as a participant in this collaborative project I volunteered to look into this further and did this by a survey of the laboratories that were providing genetic testing. (University of California San Francisco 1992)

Holtzman credits his investigation of laboratory proficiency for newborn screening as the experience that alerted him to the profound and potentially negative consequences of genetic screening:

> ... all of that was sort of a long prelude to tell you that when we did this survey and collected information one of the appalling things was the quality of laboratories that were performing newborn screening tests around the country and this has been confirmed by many other people since then. And I began to be worried as to whether these advances that were just beginning at that time and that offered hope that there would be other diseases that could be as effectively treated as PKU that as we moved from the laboratory into the clinical part of practice that these applications would not be handled appropriately, that there would be a poor quality of labs which means that some infants who could benefit from a treatment would be missed by a test and that other infants who didn't have the disease that they were tested for would be mislabeled as having the disease. And it's actually that experience that made me aware that in the practice of medicine there were a number of problems that were going to interfere with the appropriate use of new science, new technology ... I continued from there and I found many other problems just in the area of genetics that indicate that we need to pay careful attention to the clinical application of advances in science. (Interview with Neil Holtzman, University of California San Francisco 1992)

Although his work on state genetic screening legislation and PKU laboratory proficiency provided the foundation for *Proceed with Caution*, Holtzman developed the book's arguments while working at the Congressional Office of Technology Assessment (OTA) in the early 1980s.

Holtzman describes the events that led him to become a member of the advisory panel on occupational genetic screening that investigated pre-employment genetic screening and produced its landmark report (U.S. Congress, Office of Technology Assessment 1983):

> An opportunity came up to go to the Office of Technology Assessment as a senior analyst, again on leave from Hopkins. I was doing that again with the expectation that I would *not* work on genetics, when I was at the OTA.
>
> So, after the Severo Report, the next thing, I was on the OTA Committee, which published this report, U.S. Congress Office of Technology Assessment, in 1983, on genetic testing and the workplace. They did a survey and found there was very little being done, and began to lay out some of the questions about the validity of genetic tests. (Interview with Neil Holtzman, May 31, 2007)

While at the OTA, Holtzman moved away from genetics and became involved with toxicology:

> I did not expect to work on genetics. I wasn't particularly eager to work on genetics at OTA. And in fact, I did two projects there, one of which I am a co-author of, on the National Toxicology Program, which is the animal-testing that's used to look for mutagens and other toxins in the environment. Genetics was a very minor part of it. And then I also was involved with studies on Agent Orange, dioxin, in Vietnam veterans. So, this was far removed from genetics. I learned a lot about the way policies worked, and the Veterans Admin didn't want to get involved. They didn't want to see any relationship or association because they'd have to pay for it. And, you know, it's difficult to build up a strong case for an association. But I think it was there. And I think the aspects that suggest it was greatly underplayed when the report was finally published. (Interview with Neil Holtzman, May 31, 2007)

But while at the OTA, he bumped up against concerns about

discrimination, and made the link to genetic testing:

> It was at OTA that the issue of discrimination and insurance first came to my attention. There was another project going on that was looking at AIDS discrimination. Remember that was the time where, mid-1980s, all this question of denying insurance to the HIV-positive people at risk of AIDS was coming on. And the same issues came up, with adverse selection. So I sat in on a number of those meetings, and it became very apparent to me that this was the same issue that would arise with genetic testing. And there were already examples of it when we started to look. (Interview with Neil Holtzman, May 31, 2007)

Holtzman says that *Proceed with Caution* began initially as a report on a survey of biotech industries for the OTA that he had conducted with Maria Hewitt, a genetic counsellor at the University of Maryland. The OTA had just rejected a report that he had written on the insurance industry:

> I produced a draft of the [insurance] report in the summer of 1987. And it was sent out to about 65 reviewers. But I left to go on vacation in August, and I came back, and was fired. (Interview with Neil Holtzman, May 31, 2007)

Holtzman reflects on what was behind his employer's decision to fire him:

> I really think it was internal politics. They said that the report was not up to the quality of the OTA reports. Mind you, we sent it out to 65 reviewers. A lot of the reviewers came back with minor comments, but most of them thought this was a good report and something that was needed. I really think what happened was there was a guy at OTA whose toes I had stepped on, who was involved with this insurance report. I think he was very resentful that we had moved ahead very quickly, and I had taken a lot of stuff I'd learned from sitting on his committee, and talking to the people that were talking to his committee as well. I think he just felt I was intending to invade his territory. I liked the OTA work, I thought it was very close to policy-making, and I really would have been very pleased to stay there [laughs], and not to come back to Hopkins. (Interview with Neil

> Holtzman, May 31, 2007)

The report on the insurance industry "never came out," Holtzman says. Instead, his survey of biotech industries was released under Maria Hewitt's name and published by the OTA. "By mutual agreement with OTA, that was the first, early draft of *Proceed with Caution*," he says. Holtzman sums up the impact of his OTA experience on his thinking:

> So, my experience at OTA was where I got interested in a whole series of issues, of which discrimination and the insurer, particularly from the insurer's point of view, is one. And then, following that, I came back to Hopkins and got involved with the ELSI group. (Interview with Neil Holtzman, May 31, 2007)[20]

Towards the end of his career, he say, he "really lost interest in the discrimination issue" and "wasn't persuaded by this questions of genetic exceptionalism" or by privacy concerns. He turned his energies towards investigating media coverage of genomics (see, for example, Holtzman et al 2005) and commenting on the complexity of the genetics of common disorders (see especially Holtzman 2001):

> In my last few years of being active in genetics, I was much more concerned to show that the genetics of complex disease—with a few exceptions like BRCA1 and colon cancer, early-onset, rare forms of Alzheimer's, maybe prothrombin, factor 5 and a few things like that, but even that gets to be chancy—are much too complex that in the foreseeable future, the possibility of developing tests for these complex diseases, with a few rare exceptions—*rare* exceptions—was remote. I went so far to say that I didn't think that government funds should be used to look for genes for common disease. It was a waste of money. If private industry thought it was so great, let them invest in it. (Interview with Neil Holtzman, May 31, 2007)

DISCUSSION

One of the questions that arises from the history of sickle cell screening is why the ramifications of sickle cell screening programs in the 1970s did not start a national conversation about something called genetic discrimination. I asked three people who played a role in shaping public awareness of genetic discrimination about this: geneticist Paul Billings, the lead author of the 1992 *American Journal of Human Genetics* article that formalized a definition of genetic discrimination; sociologist Troy Duster, who examined sickle cell screening in his book *Backdoor to Eugenics*, and Phil Bereano, a lawyer and retired professor at the University of Washington, who was a founding member of the biotechnology public interest organization the Council for Responsible Genetics (CRG) and was active with its predecessor, Science for the People. (I discuss their activities in Chapter 5).

Paul Billings suggests that there were two different interpretive frameworks for the sickle cell experiences. Neither suggested that discrimination could become a broader problem tied to genetic diagnosis:

> I think one interpretative framework was there was racial discrimination, and that racial discrimination was obviously playing a role in health care, and so the problems of the sickle cell programs were just reflective of ambient racial discrimination. The other interpretative framework was that carriers of sickle-cell genes were not properly counseled, did not understand that there were not diseased. So it had been portrayed as a misunderstanding, really, rather than an example of genetic discrimination. (Interview with Paul Billings, July 20, 2007)

Phil Bereano argues that there is often too much risk for minorities in challenging biological determinism, because it also reinforces difference:

> There's a real concern among groups that have been historically discriminated against to highlight differences. We see this in the gay community. It's the exact same dynamic. Definitely something you can see in communities that have been historically subject to discrimination. It's clearly a double-edged sword, because you want to protect your people; the other time, you're giving fodder to the bigots. (Interview with Phil Bereano, May 17, 2007)

He suggests that African-Americans had conflicting responses to sickle cell screening:

> One is, "this is yet another example of discrimination against black people," versus "They're going to use this to say yes, we are different and we are defective." So the ideology of defect, which is critical to genetic discrimination, was one of the currents there. Discrimination from sickle cell screening didn't become more salient because this second prong caused people to moderate expressing concerns about discrimination on this basis. The idea that there was something bad about an African-American blood. You know, blood is used—at least in the States—was used as a synonym for genetic history, or heritage. Syphilis was called, colloquially, having bad blood. And of course, the Tuskegee experiment. You have these linkages here. So, you'd give pause to getting up and saying how this is another form of discrimination, because what you're also saying is "Our blood is different; it's defective; we have bad blood; and you're discriminating against us." Well, do you want to say that? Do you want to take that position? And, so, to get up and, try to fight discrimination *because* you had bad blood was very, very tricky for an African-American politician or leader to do. Or want to do. (Interview with Phil Bereano, May 17, 2007)

The other reason why sickle cell screening didn't translate into a broader concern with genetic screening widely available, says Bereano, was due to the perception that sickle cell screening was based on a laboratory test and was therefore not genetic testing:

> Why didn't this happen in 1972? What we call modern genetics didn't

> exist then. We didn't talk about sickle-cell disease because it wasn't modern genetics. Sickle cell testing was done not through modern genetic testing. It was more crude and not really a genetic test. (Interview with Phil Bereano, May 17, 2007)

Troy Duster deflates any notion that sickle cell screening mobilized concern about genetic discrimination, from either the perspective of protecting the rights of minorities or the generating awareness about the social impacts of genetic screening:

> There was never a moment in which race came forward, except for racialized groups. So, some African-Americans who were in the Sickle-Cell Foundation were very much concerned about the Stephen Pullens case, this Air Force case, but not many others were. I didn't see a general concern about sickle-cell anemia discrimination. African-American activism was focussed, as you might expect, on African-American mobilization—and that's what happened. Finally, the Pullens case was resolved, and the Air Force did admit Blacks back into the Academy. But no. This was more of a scattergun. It was never a coherent mobilization around genetic discrimination. (Interview with Troy Duster, November 30, 2007)

Unlike sickle cell screening, workplace screening registered as a departure point for later conversations about genetic discrimination for everyone that I spoke to, with the exception of Paul Billings. Billings stressed that the Severo series had no impact on the development of his concern about genetic discrimination. "I don't think I was even aware of it," he says.[21] However, like sickle cells screening, it too never provoked activism or generated widespread alarm. Workplace screening was practiced only by the manufacturing and chemical industries, so the discriminatory effects were localized. Troy Duster is emphatic that throughout these two decades there was no consensus about something

that could be called genetic discrimination. He attributes the absence of mobilization around either sickle cell screening or workplace screening to the individualizing power of genetic screening and testing:

> I think that the peculiar feature of genetics is that it fragmented the public health consensus. Before genetics, you can have a general notion of the public health around contagion, or problems of smallpox, or flu or cholera, or yellow fever, or tuberculosis. What that did, beginning in the middle of the nineteenth century, was to have this huge debate about "the public health." When you get to genetics, you get splintering and fragmentation. Because now it's, "my disease." It's "my Tay Sachs and your Cooley's Anemia." It's your beta-thalassemia; it's my alpha-thalassemia. It's your Huntington's, and it's my hemachromatosis. And these are located in different parts of the populations, [with] different capacities, different understanding, different education level, different social class positions, different ethnicities, races and so on, different genders. So, you don't get anything resembling a movement around public health. (Interview with Troy Duster, November 30, 2007)

Like the other risk critics, Troy Duster also flagged the production of risk categories through genetic screening and testing. In his 1990 book *Backdoor to Eugenics*, Duster echoes an earlier warning from Dorothy Nelkin and Laurence Tancredi (1989) that genetic testing would create a "biological underclass," when he warns that one impact of the differentiating power of genetic testing is the creation of a "genetic underclass" through the identification of alleles associated with disease.

Duster agrees that workplace screening played a greater role in heightening concern about discrimination than health insurance:

> The workplace issue seemed to be more salient in terms of actual cases that people were concerned about. The health issue was always the threat. It was the thing hovering over the conversation. It wasn't

> the real thing, while as Severo was pointing out, it was actually happening in some places, people were being discriminated against in the workplace. (Interview with Troy Duster, November 30, 2007)

However, he challenges the idea that it is possible to locate the origins of genetic discrimination or the point at which multiple interests converge into a single problem called genetic discrimination:

> You know, there's no one spot where this starts. It's not a case where one can have a chronological account where, you know, at point A and B and C, people's interests end and shape, and you have one story. There are at least five different entry points on gene discrimination. There's the workplace. There's health insurance. There's privacy, with respect to one's own local scene or family structure, or kinship group, or you name it—or disease group. So, you've got a combination where genetic discrimination has so many different meanings for people. It's sort of like diversity. If you ask someone, are they in favour of diversity, many will say, yes they are. But when you begin to explore what they mean by it, then you see that some people mean by diversity individual capacity to be enriched by different foods, and so on. And others mean, they want institutionalized transformation.
>
> Well, gene discrimination has a very similar feel to it. When someone says they're against it, you have to probe. You don't know whether that means they're against screening for Down Syndrome. You don't know if it means screening for that kind of phenomena. Is that what they mean by it? It's a Rorschach. But I think, immediately, what I have to say is that this is—and always has been—a problem where you can't assume your audience knows what the concept is. So if you ask me what the history is, I gotta say, "Which history?" [laughs] (Interview with Troy Duster, November 30, 2007)

The questions relevant to understanding genetic discrimination, says Duster, are not, "what are the roots of discrimination practices by insurers and employers," but rather, about the collection and control of personal genetic information. For Duster, privacy and control of information

became a salient concern following DOE and NIH talks to launch a Human Genome Project (HGP):

> Okay. Fine. Now, with that in mind, as soon as it became clear that knowing about the decoding of the genome was going to provide personal information, one of the questions which came up was, was "Who's going to keep the information?" So, that's the first question that came up. "Where's your DNA?" (Interview with Troy Duster, November 30, 2007)

The practice that Duster identifies as being a departure point for privacy concerns about genetic discrimination following DOE-NIH talks is not, surprisingly, anything tied to the insurance industry or employers; rather, it is a government DNA collecting initiative, the U.S. Department of Defense (DoD) DNA Registry. The DoD initiated the DNA Registry in June of 1992 to collect and store blood samples and genetic information from all military personnel, as well as civilian employees and contractors, to aid in post-battle identification of soldiers (Nelkin and Andrews 1999).[22] The DNA Registry, says Duster, was a significant departure point into genetic discrimination for individuals and organizations that were concerned with individual privacy, intersecting with what scholars Dorothy Nelkin and Lori Andrews (1999) have called "surveillance creep": the expansion of surveillance and research capacities of databases beyond their original purpose:

> [Privacy concerns related to genetic discrimination] started off with the Department of Defense. And the first question for the DoD was, "What are you going to do with this information?" And their answer was, "Dog tags! We're only going to use this in a time of war, to locate a soldier." That came up immediately, when I was on ELSI. We used to get presentations by Lieutenant Weiner, I think his name was. He

> would come in and tell us, "Don't you worry about a thing, because it's only going to be used in the military database. We're not going to use it to do any research." Now, of course, it's a different issue, because the purpose did erode, and there are people hovering around that database and saying, "Well, let's take a look at this." That was one whole arena: the fear from some people that the military and the government was collecting a lot of data on people. How were they going to use it? And of course, the subterranean issue was, right away, might it be used for discrimination? (Interview with Troy Duster, November 30, 2007)

In 1996, two Marines, John C. Mayfield and Joseph Vlacovesky, challenged the constitutionality of the DNA Registry when they filed a civil suit in Hawaii claiming that the mandatory collection of their blood samples violated their Fourth Amendment protection to privacy and right against unreasonable searchers and seizures. The two men argued that the information stored in the repository could be used to identify their genetic susceptibility to disease, which might in turn be used by health insurers or employers to discriminate against them. The courts ruled against the soldiers and they were reprimanded, but as a result of the court challenge, the DoD changed its policy and agreed to destroy individual tissue and blood samples on request when military personnel withdraw from service (Nelkin and Andrews 1999).

One of the points that Duster makes in telling this story is, of course, that the private sector does not have a monopoly on collecting, storing or using genetic information. This point is, however, absent from contemporary genetic discrimination discourse, which has framed the problem as one of misuse of genetic information by insurers and employers discrimination. I return to the question of privacy and ownership of genetic material in Chapter 10.

CONCLUSION

Neither sickle cell screening nor workplace screening generated a public consensus that genetic screening was a threat to many Americans. While discrimination against African-Americans as a consequence of mandatory sickle cell screening began as early as the early 1970s, sickle cell screening was depicted by commentators as a disastrous engagement in American public health that furthered societal and institutional mistreatment of African-Americans using a new tool, and not the harbinger of a wider problem with genetic diagnosis. Sickle cell screening grew out of a long history of public health portrayal of African-Americans as asymptomatic disease carriers and "disease vectors," and problems with the screening programs were attributed to a profound ignorance about the differences between the mostly benign heterozygous trait and the serious homozygous disease. Critics saw insurer and employer discrimination as the oft-unintended consequences of an unbridled enthusiasm for genetic screening without a clear endpoint or treatment, coupled with widespread ignorance and racist assumptions about which populations were at risk. Few today label this episode in screening history as an instantiation of genetic discrimination, even though one of the problems with the programs was the failure to distinguish between the mild sickle cell trait and the serious disease. The outcome—healthy individuals who were carriers were viewed as seriously ill and a positive genetic diagnosis was seen as proof positive of a debilitating health status warranting differential treatment—is a facet of how genetic discrimination has been defined since 1992, whereby presence of an allele for single-gene disorder or susceptibility genes for common disorders is reason for insurers, but also

employers, to treat individuals as high-risk.

Workplace screening departed in many ways from sickle cell screening. The impetus was not political pressure from black leaders to improve the plight of African-Americans, but the enthusiasm of toxicologists to apply the findings of pharmacogenomics research to industry, to remove workers who were likely to develop occupational disease from exposures to toxic substances and reduce operating costs. The targets of workplace screening were not just African-Americans; they were all applicants for productions jobs that entailed exposure to toxic chemicals within the chemical and manufacturing industries. Most importantly, workplace screening introduced the notion of "hypersusceptibility," a term that implies a more diffuse gradient of vulnerability to disease, unlike screening for single-gene disorders where carrier status is often a benign condition. With workplace screening, denial of jobs resulted not from gross misunderstanding about the difference between the heterozygous and homozygous states, as it was with sickle-cell; it resulted from evidence of the presence of an allele deemed high-risk, which the carrier frequently had no awareness of and was not handicapped by.

Responses to workplace screening produced a burgeoning awareness of a new category of Americans vulnerable to discrimination that was race-neutral: healthy, asymptomatic individuals with a genetic makeup that made them "hypersusceptible" to occupational disease. This response, I suggest, weakened the close association of genetic disease with race and ethnicity that had seemed self-evident in the 1970s with sickle cell screening, and forged the first awareness, however limited in its impact,

that the category of "genetic defect" applied to Americans who were not minorities. Any American entering a production job in the manufacturing or chemical industries could be deemed an unacceptable occupational risk and denied a job because of an inherited susceptibility, hidden even to himself or herself.

Both Neil Holtzman and Tory Duster have linked the seemingly disparate and diffuse occurrences of African-American and workplace discrimination to a growing enthusiasm for genetic explanations for disease and social problems. *Proceed with Caution* and *Backdoor to Eugenics* served both as sophisticated introductory texts to genetic testing, and as warnings about the eugenic potential of the diffusion of genetic screening and testing. Both elaborated on the power of genetic testing to entrench and naturalize the ideology of the genetic defective, and to create a social underclass of genetic undesirables, as well as incentives for abortion and discrimination. As more genetic linkages were found for common disorders, such as breast cancer and depression, argued Holtzman and Duster, individuals testing positive for a mutation were being penalized for their inborn defects, through denials of insurance claims and employability, or through workplace practices that restricted their employment. These commentators worried about the normalization of financial and societal disincentives to reproduce, and tacit pressures on carrier of defective genes to abort fetuses carrying mutations.

For Holtzman, the possibility that individuals might be denied insurance and employment because they carried mutations that might lead to disease costly to treat was a subset of a larger and graver problem: a

revival of eugenic imperatives. His concern that genetic testing and screening are ushering in a new era of eugenicist practices, which was shared by Troy Duster, is largely absent from contemporary discourse about the problem of genetic discrimination. Yet even without using the term "genetic discrimination," *Proceed with Caution* offered the most sophisticated and cogent warning about the looming problem that Paul Billings and colleagues would call genetic discrimination just three years later.

Commentary during these two decades indicates that two interpretations of discrimination were simultaneously in use which mapped onto two distinct populations of the vulnerable. The dominant meaning of discrimination was racial discrimination, as codified in Title VII of the Civil Rights Act: denials of entitlements or differential treatment of individuals based on visible markers of difference, such as skin colour and ethnic identity. This mapped onto racial discrimination of minorities, particularly African-Americans in both sickle cell screening and workplace screening. Susceptibility screening targeted minorities to some degree, because the traits selected for screening are found at higher incidences among minority populations. According to Draper (1991), this was no coincidence: minority workers who entered highly-paid production jobs in the chemical and manufacturing industries threatened the status quo. Because screened traits lined up with visible markers of race and ethnicity, using screening results to make employment decisions was at best, an unintended violation of the Civil Rights Act, and at worst, a clever way to get around Title VII prohibitions against discriminating by visible markers. This was the primary concern of critics such as Severo, that

screening was bypassing Title VII prohibitions of the Civil Rights Act. Thus, there was no societal consensus before the late 1980s that something called "genetic" discrimination was afoot, because genetic screening was simply a proxy for racial and ethnic discrimination.

But during the 1980s, "discrimination" developed a second meaning in response to workplace screening. The growth of concern about discrimination towards healthy, asymptomatic carriers is evidence that, by the time of workplace screening, some were seeing genetic screening as constructing categories of high-risk populations with no visible markers of difference (such as skin colour or religious affiliation) who were not handicapped, but who nonetheless lacked protection from discrimination under existing laws. This is the population today considered at risk of genetic discrimination.

The commentaries on workplace screening also reveal a sentiment that is central to contemporary discourse and framing of genetic discrimination: a profound sense of betrayal that powerful technologies with the potential to identify health problems were being used against healthy individuals to label them as diseased. This sentiment surfaced following sickle cell screening, but was a minority response. Underlying this betrayal is the assumption that genetic technologies are fundamentally benign, and their use by institutions to reduce their own exposure to risk is unethical. Despite the American experiences with sickle cell and workplace screening, this sentiment is operating just as powerfully today, as I will show in Part II.

[1] Interview with Troy Duster, November 30, 2007.

[2] For an analysis of the intersection of genetic discrimination with the Americans with Disabilities Act, see Vazakas 1993.

[3] See Paul (1994 and especially 1997) for a history of PKU screening.

[4] Interview with Paul Billings, July 20, 2007.

[5] See Duster (1990) for a comprehensive discussion of the origins of sickle cell screening, and a comparison to Tay-Sachs screening.

[6] Randall (2006) argues these efforts were aimed primarily at changing the reproductive behaviour of African-Americans.

[7] Paul Billings describes this as "ambient discrimination." Interview with Paul Billings, July 20, 2007.

[8] See Brandt-Rauf and Brandt-Rauf (2004) for an overview of workplace genetic screening and testing in the United States.

[9] G6PD deficiency was associated with a predisposition to increased risk of anemia among individuals working with the oxidizing chemicals aromatic nitro and amino, while SAT deficiency was associated with the development of emphysema and chronic bronchitis with exposure to pulmonary irritants (Draper 1991:25).

[10] The term "susceptibility" in the context of workplace genetic screening refers to the presumed heightened susceptibility—"hypersusceptibility"—of individuals to occupational diseases because of predisposing genetic traits.

[11] For a review of U.S. workplace fetal exclusion policies, see Uzych (1986).

[12] The exception was SAT, which is found at a higher incidence among northern Europeans.

[13] Women who work in the chemical, oil, steel, auto and textile industries, and who are at an age that is considered to be reproductively active have been excluded from production jobs using chemicals such as lead, mercury, benzene, acrylamide, vinyl chloride, carbon disulfide, and carbon monoxide by companies claiming to want to protect the health of fertile women and the fetus. Draper (1991, 1993) identifies flawed assumptions of fetal exclusion policies: they assume that all fertile women are potential mothers and that reproduction is the exclusive domain of women. The conclusion is that only fertile women need to be protected from fetal hazards. Male workers are not excluded from exposure to these substances, despite evidence that many of these substances can damage sperm, that male workers can be vectors of fetal hazard by exposing their spouses to harmful substances, and that paternal exposure can pose greater risks to the fetus than maternal exposure (Draper 1991, 1993). Moreover, women in industries such as healthcare are not so rigorously excluded from fetal hazards such as x-rays. "Women,"

argues Draper (1993: 97), "are told they must be denied jobs in order to protect the fetus; they are treated as mothers who require protection until they are proven sterile."

[14] In fact, General Motors Company implemented a fetal exclusion policy for lead exposure in 1952, which was been challenged as sexually discriminatory. Female workers had to demonstrate their infertility to a plant physician (Uzych 1986).

[15] The authors explored at length whether genetic makeup could be deemed a handicap, even in the absence of existing disability, under the 1964 Civil Rights Act and the 1973 Rehabilitation Act. This turned on the question of whether workplace genetic screening was predictive of future illness. Treating genetic makeup as a handicap offered protection for workers (but workers would have to claim their genetic makeup was a handicap, even if they didn't consider themselves disabled), and a legitimate basis for job exclusion by employers (but employers would have to demonstrate the genetic screening was predictive of future illness). The passage of the Americans with Disabilities Act in 1990, which protected employees with an existing disability caused by an inherited disorder from workplace discrimination, closed a gap that was left open by the two federal acts.

[16] Interview with Phil Bereano May 17, 2007. See also Holtzman 1989.

[17] Sickle-cell was detected in a laboratory using several methods on blood samples, including a blood count, hemoglobin electrophoresis, and inducement of sickling. Tay-Sachs was detected using an inexpensive biochemical test—an enzyme assay that measures levels of the enzyme hexosaminidase A in blood serum.

[18] In 1982, prenatal diagnosis of sickle cell disease was introduced. In 1983, a marker for HD was discovered on chromosome 4, and the first predictive test for the condition became available in 1986. In 1985, markers for CF were discovered on chromosome 7. Four years later, in 1989, Canadian researchers and current NHGRI Director Francis Collins isolated the associated gene and mutations for CF. And in 1987, the *hexA* gene and mutations for Tay Sachs were located, four years after Josef Ekstein launched Dor Yeshorim to screen for "Jewish genetic diseases" after losing four children to TSD.

[19] Interview with Neil Holtzman, May 31, 2007.

[20] Holtzman chaired the NIH Task Force on Genetic Testing in 1995. The Task Force published a report in 1998 (Promoting Safe and Effective Genetic Testing in the United States) that addressed genetic discrimination.

[21] Interview with Paul Billings, July 20, 2007.

[22] The Registry was established by the Armed Forces Institute of Pathology of Walter Reed Army Medical Center in Washington. The repository for the blood samples is in Gaithersburg, Maryland. It holds more than one million samples from former and active-duty military personnel, and civilian employees.

INTRODUCTION

In 1992, following warnings from the risk critics that the expansion of genetic testing was creating a genetic underclass that would be disenfranchised from insurance coverage and employment opportunities, clinical geneticist Paul Billings and five colleagues presented evidence to readers of the *American Journal of Human Genetics* (*AJHG*) that Americans who are carriers for rare genetic disorders were being treated as seriously ill by major social institutions (Billings et al 1992). The vulnerable Americans that these authors identified were not job applicants being screened for inherited hypersusceptibility to industrial disease, or minorities being screened as carriers for genetic disorders. Nor were they disabled persons. They were healthy individuals who had been identified, through their own medical care, as carrying rare, single-gene disorders such as Huntington disease or Charcot Marie Tooth, and their genetic information was being used by other institutions for their own actuarial decision-making. And the primary realm in which these individuals with disease-linked genotypes were experiencing discrimination was not their employability, but their insurability.

The publication, which drew on reports from clinicians and patients of institutions denying entitlements to healthy individuals, was instrumental in putting genetic discrimination on the map as a problem of the ongoing insurability of Americans. It garnered media attention, spawned a lively debate in the *AJHG*, and helped to secure a permanent place for genetic discrimination as a policy issue on the agenda of the ELSI (Ethical, Legal and Social implications) Working Group of the Human Genome Project

(HGP), a joint project of Department of Energy (DOE) and the National Institutes of Health (NIH).

Yet only a few years after this *AJHG* publication appeared and introduced genetic discrimination to the public as a discrete problem of health insurance for individuals with rare, single-gene disorders, new actors came on board to champion—and reframe—the issue. The remainder of the nineties saw these actors define genetic discrimination as a pressing problem for all Americans and for the nation. Their collaborations mark a significant institutionalization of genetic nondiscrimination activism (see Rabeharisoa 2008).

In this chapter, I describe the genesis of the *AJHG* publication and its impact, showing why the authors came to focus primarily on health insurance. I then turn to developments from 1992 to 2003, identifying new actors who adopted the problem, and how they have continued to shape public understanding of this problem.

1992: NAMING THE PROBLEM

In the March 1992 issue of the *AJHG,* clinical geneticist Paul Billings and five colleagues introduced the clinical genetics community to a new problem: genetic discrimination. They defined this as "discrimination directed against an individual or family based solely on an apparent or perceived genetic variation from the 'normal' human genotype...in many social institutions, especially in the health and life insurance industries" (Billings et al 1992:476). They also identified a category of Americans experiencing discrimination as a consequence of genetic testing: healthy,

mostly asymptomatic individuals with genotypes linked to rare genetic disorders.

The authors presented evidence from a pilot study showing that major social institutions were treating these healthy individuals with genotypes for specific single-gene disorders as high-risk prospects. These individuals, whom the authors labelled "the asymptomatic ill" (Billings et al 1992:478), could be characterized as phenotypically asymptomatic or experiencing only mild symptoms of their conditions. From twenty-nine responses to a solicitation of 1,119 genetics professionals and patients, the authors found evidence that insurers, employers, adoption agencies and health care providers were treating genetic conditions "as extremely serious, disabling, or even lethal conditions" (Billings et al 1992:481).

The twenty-nine respondents described thirty-two incidents of insurer discrimination and seven incidents of employer discrimination (Billings et al 1992:478). For example, health insurance subscribers with genetic conditions but who were asymptomatic experienced difficulty obtaining new coverage when they changed their existing policies through moves or new jobs. Respondents related withholding essential "medical or family history information to physicians, employers or insurers" because they feared discrimination (Billings et al 1992:478). Insurance agents encouraged some respondents to withhold medical information or to falsify it on application forms (Billings et al 1992:478). The urgency of the problem lay in the increasing number of laboratory tests for rare inherited disorders, and the anticipated growth of genetic tests for common conditions such as "cancer, cardiovascular diseases, and mental disorders"

(Billings et al 1992:476). The authors warned that the rapid growth of genetic tests of all types (predictive, diagnostic, prenatal, and susceptibility) would create a category of "the asymptomatic ill" that would become uninsurable.

Features of Genetic Discrimination

The common denominator of genetic discrimination, according to the authors of the 1992 *AJHG* article, is the inability of social institutions to grapple with the complexity of genetic diagnoses and appreciate the significance of variability in gene penetrance and gene expression. Most institutions, they said, unfairly simplify genetic diagnoses, treating a genetic condition, even amongst asymptomatic healthy individuals, as proof positive of a disease state—an assessment redolent of the problems with sickle cell screening in the 1970s. Institutions fail to understand that "many individuals with 'abnormal' genotypes will either be perfectly healthy, have medical conditions which can be controlled by treatment, or experience only mild forms of a disease" (Billings et al 1992:481). For example, one asymptomatic individual who had lived for years with hereditary hemochromatosis and was denied insurance despite taking preventive measures to prevent iron overload, stated, "I might as well have AIDS"—referring to a condition that was invariably fatal in an era before the advent of affordable antiretroviral therapy. A woman with mild symptoms of Charcot Marie Tooth (CMT), a neuromuscular condition that manifests in wide individual symptomatic variability, was denied a job offer after her employer educated himself about the condition using a medical text.

Another feature of genetic discrimination, said the authors, was the tendency of institutions to give more weight to genetic information than to other medical information, so that "certain conditions categorized as 'genetic' are viewed as special and handled differently in terms of social decision making" (Billings et al 1992:480). For example, a couple with a family history of Huntington Disease, a fatal neurological disorder that strikes those carrying the gene later in life, had decided to adopt a child without undergoing confirmatory genetic testing for presence of the gene. (It is not clear from the authors' anecdote whether one or both of the spouses had a family history of Huntington). The adoption agency denied the family's application because of the 50% risk that one parent might carry the gene and develop the disease. Driving this decision-making, they said, was an over-emphasis on the relationship of genetics to good health, what they called "the myth of genetic perfection":

> The agencies assume that the best possible family is the one least likely to face medical adversity—the "perfect" family with a disease-free genome. Unfortunately, all families are at risk. The comparison made by one respondent, of being at risk for Huntington disease with susceptibility to diabetes or cancer, highlights a prejudice—that the chance of developing a genetic condition is perceived differently from a similar probability of contracting an illness not produced primarily by a gene. (Billings et al 1992:480)

Some cases the authors included pointed to explicit eugenicist reasoning. For example, a couple with a child who had cystic fibrosis (CF), a progressive and debilitating disease affecting the lungs and other systems, was given a positive CF prenatal diagnosis for the second pregnancy. The couple's Health Maintenance Organization (HMO), or

health care provider, threatened to withdraw health coverage for the existing child, as well as prenatal, postpartum and pediatric care for the second child, if the couple did not abort the pregnancy.[1]

Operationalizing Genetic Discrimination

The authors operationalized genetic discrimination as a concrete problem with three features. One, genetic discrimination was an equal-opportunity threat that skirted visible markers of difference and susceptibility to disease. From their study, the authors discovered that most individuals experiencing discrimination had neither visible signs of illness or disease, nor visible markers of susceptibility to illness or disease. Thus, discrimination on the basis of genotype was not a risk confined only to minorities, such as African-Americans and Jews who had been targets of sickle cell and Tay Sachs screening programs respectively, nor for populations known to have a higher incidence of certain genetic conditions, such as Italians and Greeks for beta-thalessemia. All individuals from families with rare disorders were potentially at risk of being deemed high-risk by institutions.

Two, unlike the two screening practices that had produced notable instances of discrimination—sickle cell screening and pre-employment workplace screening—there was no mandatory mechanism linked to genetic discrimination. Discrimination was an unanticipated consequence experienced by individuals and families who had chosen to undergo voluntary genetic testing for personal medical decision-making. This outcome pointed to the increasing actuarial value of genetic information in the risk-assessment practices of a variety of institutions, and the widening

ambit of personal medical information (e.g. Nelkin 1996).

Three, the primary domain of exclusionary decision-making based on genetic information was not with employers and the workplace, as it had been with sickle cell screening and workplace susceptibility screening, but with health insurers. This pattern of genetic discrimination that the authors described departed from discrimination tied to both sickle cell and workplace screening.[2]

Notably, the authors did not use a privacy frame to portray the problem. Their theoretical perspective was not libertarian: they did not present genetic discrimination as stemming from the too-easy flow of information, nor did they call for stronger privacy protections for individuals and genetic information. The authors' stance was consonant with that of the risk critics, who were concerned with the runaway effects of genetic determinism. Their message was a warning based on empirical findings: that the acceleration of our ability to detect genetic difference, in the absence of both treatments and the ability of institutions to grapple appropriately with the significance of genetic diagnoses, was causing institutions to mistakenly classify healthy individuals as diseased. This would be the last major appearance of an anti-determinist framing of genetic discrimination. It would be superceded by the mid-1990s by a rights discourse around both information privacy and the individual's right to genetic self-knowledge in the pursuit of good health as a civic duty (e.g. Novas 2006; Rose 2001).

A New Problem?

The authors of the *AJHG* publication were not the first to use the term

"genetic discrimination." The Social Issues Committee of the American Society of Human Genetics, which publishes the *American Journal of Human Genetics*, sponsored a meeting in November of 1986 on genetic discrimination as a direct consequence of genetic screening in the workplace and insurer access to genetic testing results (Rowley 1988). However, the report from this workshop does not provide attribution or define the expression, and no one seems to have picked up on this workshop or the report.

The next major appearance of the term was in the 1987 book *Medical Genetics: A Legal Frontier* by legal scholar Lori Andrews. Andrews used the expression to capture a wide range of practices, from workplace screening for occupational disease to denials of coverage by insurers, as genetic discrimination, and to argue for policies to prevent private institutions from using genetic information to "disadvantage currently healthy and asymptomatic individuals" (1987:19). Andrews herself attributed the phrase to James E. Bowman, the African-American physician and scientist who had written extensively on sickle-cell screening in the 1970s, in an unpublished conference paper he wrote on sickle cell disease (Bowman 1972).[3]

The expression also appeared in several newspaper articles in the 1980s, beginning in 1983 (e.g. Globe and Mail 1983; Henig 1989; Weiss 1989). Genetic discrimination in the print media was not a well-defined or discrete problem. Like Andrews, reporters used the phrase to highlight an array of alarming practices tied to genetic determinism, ranging from workplace genetic screening for occupational disease, to negative eugenics.

For example, in a 1989 *New York Times* article titled "High-Tech Fortunetelling," reporter Robin Morantz Henig explored "the questions raised by the new power of prediction" through comments by Nancy Wexler, Lori Andrews, Laurence Tancredi, Dorothy Nelkin, Thomas Murray, and Paul Billings. One theme of the article was genetic uncertainty. Henig faithfully parlayed Billings's message about the contingency of genetic diagnosis:

> From a societal point of view, the true danger of genetic tests is not that they convey too much information, but too little—and that the information is far more ambiguous than it first appears. "There is virtually no genetic condition in which the genes alone determine outcome," says Dr. Paul R. Billings, director of the Clinic for Inherited Diseases, affiliated with Harvard Medical School. "Even in Huntington's disease, there is much uncertainty. Yes, you can say whether or not the individual appears to have the gene, and you can say that as far as we know all who have been screened who have the gene have gone on to develop Huntington's. But you still can't say anything about when the disease will start, what will be the course of the disease, and what will be the relevant aspects of the illness." (Henig 1989)

The other theme in Henig's article was the tension between individual rights to (genetic) self-knowledge and the potential of institutions to abuse that information. In her discussion of genetic discrimination, Henig mentioned the preliminary results collected by Paul Billings that would become the basis for the 1992 *AJHG* article. She also discussed the 1983 OTA findings about workplace genetic screening for occupational disease alongside a warning from Dorothy Nelkin that alluded to social pressures to curtail reproductive choices. As a reporter, Henig's job was to raise questions about the potential social impact of genetics, and she drew upon

the social forecasting of the risk critics. But her futuristic portrait genetic discrimination strained credulity:

> The danger will come when imprecise tests are used as though they can predict the future, and when institutions actually use them to construct the future: when court decisions are based on the genetic profiles of the accused; when employers refuse to hire or train individuals at high risk of dying in their prime; when health insurance companies insist on knowing the genetic profiles of their potential subscribers before paying for "pre-existing" genetic conditions; when schools require a permanent genetic record to anticipate which children will exhibit behavioral problems or learning disabilities. (Henig 1989)

Identifying and Diagnosing a Social Problem

If the Screening Group authors shared the same concerns as the risk critics about genetic determinism, why did they succeed in legitimizing discrimination as a real problem and creating an association in the public mind with health insurance? One answer is that the Screening Group named and framed (Brown 1995) the problem, producing what medical sociologist Phil Brown identifies as the first stage of social construction of a disease: the identification and diagnosis of genetic discrimination to the clinical genetics community and to the public.

The authors legitimized genetic discrimination as a social problem in four ways. One, they named and defined the problem to the clinical genetic community, providing criteria for social diagnosis. Although Neil Holtzman (1988) had been the first to elaborate on the problems with health insurance experienced by individuals with rare genetic disorders in the AJHG, he had not singled this problem out or named it.

Two, the authors carefully circumscribed the problem. By describing

genetic discrimination as the *unintended consequences* of genetic testing, they excluded any non-clinical genetic screening or testing practices designed to assess individuals for employment or insurance benefits. This shaping of the problem excluded workplace genetic screening for hypersusceptibility to occupational diseases from its scope—a screening practice that earlier commentators and reporters had included within the ambit of genetic discrimination. What did fall within the scope of genetic discrimination, according to the authors, were cases where employers used knowledge of individuals' genotypes—produced outside of the employment context—to deny jobs or other benefits. By circumscribing the problem as the unintended consequences of genetic testing, the authors narrowed the problem to one dynamic: the inappropriate uses of appropriate medical diagnosis. This was an alarming enough prospect.

Three, the authors focussed on presenting existing cases rather than elaborating on possible futures. Their study substantiated the existence and scope of the problem to a community that had been sceptical that genetic discrimination existed, and even hostile to such claims. Four, the methodology that Billings and his colleagues used to authenticate the problem—the anecdotal case report—put a human face on the problem and made their claims difficult to dismiss outright.[4]

While the authors did weave negative eugenics into their portrayal of genetic discrimination—one of the cases that Billings and colleagues presented was that of an HMO pressuring a couple to terminate their pregnancy—they did not emphasize the overarching prospect of negative eugenics to the extent that Neil Holtzman and Troy Duster had. The

message that the authors delivered was less a warning about the rationalization of reproductive imperatives than a concrete illustration of how some Americans with rare genetic disorders were losing their benefits, particularly health insurance. This emphasis on the tenuousness of health insurance mirrored widespread frustration with managed care in the country, but it also drew on the authors' personal experiences and the influence of their professional networks.

In the next section, I discuss the origins of the study and the article. The authors of the publication were members of a small Boston organization, the Genetic Screening Study Group, which, along with the Council for Responsible Genetics (CRG), had grown out of a leftist organization, Science for the People. Members of all three groups were experienced social activists. Drawing from interviews with members of the Screening Group, Science for the People, and CRG, I trace how genetic discrimination became a focal point of the Genetic Screening Study Group and shaped the roots of a contemporary understanding of the problem of genetic discrimination. I also show that the roots of concern about genetic discrimination for some of these individuals date to the 1970s.

Origins: The Genetic Screening Study Group and Science for the People

Although Parthasarathy (2004) credits the Council for Responsible Genetics (CRG) with transforming genetic discrimination into a policy issue in the 1990s, it was the Genetic Screening Study Group that created a solid association in public consciousness between genetic discrimination and vulnerability to insurance problems in the 1980s. And the group's ideological concerns about genetic discrimination can be traced to an

earlier time: to activism in the 1970s over the emergence of recombinant DNA (rDNA) technology.

Given the current stature today of genetic discrimination as a significant civil rights and policy problem, it might seem surprising that a small group of scholars and activists, some with ties to anti-Vietnam war activism in the 1970s, was instrumental in defining genetic discrimination as a problem of insurability. But members of the Genetic Screening Study Group had decades of experience in social critique and activism (e.g. Beckwith 2002). The problems with managed care and the entrenchment of inequities of a privatized health care system that left millions of Americans uninsured was one focus of a small network of scholars and activists in Cambridge, Massachusetts who met biweekly to discuss the social impact of genetic technologies. It was their collaborations in the 1980s that would shape genetic discrimination into a problem of health insurance coverage, and put the issue on the map as a federal policy concern.

The Genetic Screening Study Group, formed in 1986, grew out of an older group—the Sociobiology Study Group—whose members had belonged to the left-wing organization Science for the People. Jonathan Beckwith, a Harvard microbiologist and geneticist, started the Sociobiology Study Group in 1975 to assess the merits of E.O. Wilson's arguments and provide a counterbalance to the overwhelming popular and media interest that greeted *Sociobiology* (Beckwith 2002). Members of the Sociobiology Study Group included biologists Richard Lewontin and Ruth Hubbard, both members of Science for the People, and paleontologist

Stephen Jay Gould. The group later expanded to include members from other academic disciplines, as well as clinicians and students (Beckwith 2002).

Beckwith himself had been an active member of Science of the People, and he brought to the group, as did other members, that organization's concern (and his own personal concern) about the abuses of genetics in the United States and Germany, and the revival of eugenics in contemporary science research agendas.[5] From 1975 to 1986, members met to discuss the new evolutionary biology being readily embraced by the media, and to publish their commentaries in the *New York Review of Books* and in their own book (Sociobiology Study Group 1984).

In 1986, the U.S. Department of Energy began efforts to launch a national project to sequence the human genome. Around this time, the interests of the Sociobiology Study Group members shifted from behavioural genetics to advances in human genetics. The group changed its name to the Genetic Screening Study Group (Beckwith 2002) (hereafter, the "Screening Group"). Still headed by Jonathan Beckwith, it welcomed new members, among them chemistry professor Joseph Alper, geneticists Marvin Natowitz and Paul Billings and later, lawyer Lisa Geller.[6] Paul Billings, who had graduated from Harvard University in 1979 and completed his clinical training in medical genetics at the University of Washington in 1983, joined the Screening Group in 1986, two years after moving back to Boston to become director of the Clinic for Inherited Diseases and an instructor at Harvard Medical School. He describes the Screening Group as "reading broadly in the social impact of

genetics," particularly eugenics and sociobiology.[7]

Amongst some members of the Screening Group, concern about discrimination can be traced to the 1970s with their involvement in Science for the People over the development of rDNA technology. Science for the People was one of the principle bodies to challenge the use of rDNA technology in the 1970s. Jonathan Beckwith explains that rDNA triggered a strong concern about discrimination given the complicity of geneticists in the United States in legitimizing eugenics policies in the twentieth century, because of fears the technology might permit the genetic manipulation of people.[8] Other past members of Science for the People offered similar accounts. For example, Phil Bereano, a retired professor at the University of Washington and a founding member of CRG when it split off from Science for the People in 1983, said members of Science for the People were attuned to the potential for what would be called genetic discrimination following debate about NIH guidelines for rDNA. He argues that the shift towards thinking about discrimination as something related to modern genetics occurred in 1977. That year, several members of Science for the People—Sheldon Krimsky, Jonathan Beckwith, Jonathan Newman, and Phil Bereano—flew to Washington, DC, funded by the environmental group Friends of the Earth, to testify on the NIH draft guidelines on rDNA technology that had been published the previous year, in favour of public participation on the institutional review boards. Bereano explained the association members made between rDNA and discrimination of individuals based on their genetic makeup:

> I don't think we used the term genetic discrimination. We said that unless this is carefully regulated, information will come up that will

> lead to the negative impacts on peoples' abilities to get entitlements. We talked very generally about what might be found. But there already was discussion that the impact of recombinant DNA might lead to discrimination. (Interview with Phil Bereano, May 17, 2007)

rDNA technology was also a point of ideological departure for members of Science for the People over the political significance of the technology. Science for the People subsequently developed into two different organizations: the Sociobiology Study Group, and CRG. Members who joined the Sociobiology Study Group were concerned about the eugenic potential of rDNA technology, according to Jonathan Beckwith, while CRG members privileged health concerns related to genetic engineering and genetically-modified foods.[9]

Beyond the concerns about the social impact of rDNA that had travelled from Science for the People to the Screening Group, contemporary developments in genomic mapping became a focus of members' concerns, with genetic discovery of diseases rapidly accelerating during the mid- to late-1980s. Jonathan Beckwith describes the trepidations of Screening Group members about the pace of genetic discovery:

> From the very beginning, there was this sense that we've entered this new era where genes associated with known diseases were being found every week. Some of them fell apart, but it did appear that the amount of genetic information that was becoming available was going to expand very rapidly, as it has. There was this really strong feeling that, even though genetic discrimination might have not been a big issue at the time, that inevitably it going to become a big issue. (Interview with Jonathan Beckwith, May 6, 2008)

The Screening Group attacked some well-travelled areas. Workplace

genetic screening for hypersusceptibility to occupational disease, for example, became a target of the Screening Group's efforts. In 1988, members wrote a position paper on workplace genetic screening, which they distributed at a meeting of the American Association for the Advancement of Science (AAAS) meeting that year.

Other members of the Screening Group, who had not been members of Science for the People, made concerns about the health care system a focal point. Paul Billings, who had encountered the inflexibility of the health care system in his own clinical practice, recalled that health care reform and the single-payer movement were important influences on the Screening Group:

> There was a lot of influence by Steffie Woolhandler and her husband [David Himmelstein] who led the single-payer system. So discussions about genetic discrimination partly came out of general discussions about health care reform, health care financing, out of talking about new technologies. We talked about just how broken the health care system was, and how people were finding it difficult to navigate the U.S. health care system, and financing things. And then in my clinical practice, I heard of some of these cases.
>
> I had been a general intern, and then I was trained in this very rare, mostly pediatric sub-specialty of medical genetics. And everyone was aware that there was a problem of the uninsured and people falling out of the American health care system. There was no great surprise. I think that the cases that really got to me were these people where they were having such a struggle paying for the diet, to keep their kid on some diet who needed it for a biochemical genetic disorder. And the system was so hostile to them. So these were cases that I was dealing with in my clinic, and that enlivened my thinking about it. (Interview with Paul Billings, July 20, 2007)

Describing himself as "the sole clinician in a small group," Paul Billings

recalled that discussions about genetic discrimination came out of what he called "a melding" of the Screening Group's long-time interest in the social impact of genetics and his own clinical experience:

> The idea clearly came from a nexus between general interest in this range of social impacts of more genetic information becoming available, which was a basic interest of the group, and my own experience with patients having problems of one sort or another. For example, people with phenylketonuria not being able to get the diet because they didn't have insurance and people being denied genetic tests as not medically-necessary. (Interview with Paul Billings, July 20, 2007)

The Screening Group came to focus on the insurance industry, because that appeared to members as the most obvious source of discrimination.[10] Paul Billings explains that the risk practices of insurance companies allowed for no nuance in assessing individuals with genetics disorders:

> The policy of insurance companies deals with pools of information. Variability is handled in the insurer system by pooling. So the question is, "What pool do people with genetic disorders fall in?" And, "Do they fall into the average risk, the high risk, the low risk, or do they not get risk-rated at all?" What was happening was there was a view that everybody with a genetic disorder was seriously ill. The seriously ill in the system can either be placed in a very high-risk pool, or they get red-lined and get kicked out of the pool altogether. This was the situation that we were dealing with. (Interview with Paul Billings, July 20, 2007)

Impetus for the Study

In 1988, the same year that the Screening Group wrote their position paper on workplace genetic screening for the AAAS meeting, members talked about how they might put together a study to document evidence of discrimination by health insurers and other institutions. Paul Billings

recalls the genesis of the idea:

> I can remember then there was this discussion of how we would go about finding other evidence of it. Or find evidence that genetic testing might be a kind of a gating phenomenon for access to the health care system. I remember the discussion not going very far. I suggested, "Well, maybe I'll just ask around, or something like that." (Interview with Paul Billings, July 20, 2007)

Legal scholar Phillip Reilly offers a somewhat different account of how these events transpired, one in which he credits himself with the first published usage of the term "genetic discrimination" in 1985, and with the idea to conduct a study:

> I trace the origins of the survey research that Billings and his colleagues undertook to a small meeting in about 1987 of the Genetic Screening Study Group, a Boston-based organization of concerned scientists and others to which I was invited to make a presentation and at which I first met Dr. Billings. At that meeting I suggested that there was a need for a study like the one that they later planned, organized, and conducted (and which they may well have already conceived). In making the suggestion I was motivated by the same concern that motivates me today. No published evidence of genetic discrimination existed, and I wondered whether my worries about the potential for abuse of genetic data were justified (Reilly 1999:108).[11]

In his memoir, Jonathan Beckwith (2002:163) confirms Reilly's recollection of events, while recounting his version of the discussion that members had during an interview:

> We asked Phil whether there was any evidence that people had suffered negative consequences as a result of the availability of genetic information about them. We knew of isolated incidents—the problems associated with XYY screening, the use of sickle cell testing to reject applicants from health insurance and employment. Were these only isolated incidents? Phil told us that no one had ever asked

> us this question. For example, he knew of no surveys that had looked for cases of discrimination by surveying people who had undergone genetic tests. He also pointed out that there were no laws to protect people against such discrimination. It seemed obvious that employers and insurance companies could have a strong interest in new ways of predicting who would remain healthy and who would get sick. We wanted to know if people who were perfectly healthy, but had received genetic tests indicating susceptibility to a particular disease, suffered any discrimination, such as losing insurance or a job. But without any evidence or examples, "genetic discrimination," as we called it, remained a theoretical problem. Before we could discuss public policy on genetic discrimination, we had to know whether it existed. We would make our own attempt to gather data on this subject.

However, in an interview, Beckwith notes that the year Reilly visited the Screening Group was 1988.[12]

In the end, it was Paul Billings who decided to document cases of discrimination. A few weeks after Reilly's visit to the Screening Group and its discussion about the absence of any studies, Billings drafted an ad to solicit cases of discrimination and placed it in the *American Journal of Human Genetics*.[13] The ad ran for a few months:

> At the time, you could post for free short research requests, and publish them in the *American Journal of Human Genetics*. I put it on the bulletin board of the National Society of Genetic Counselors, and posted it in the *American Journal of Human Genetics*. This whole flood of stuff came in. And that was the genesis of some of the initial work. Then we applied for an NIH grant and we were NIH- funded for the second study. (Interview with Paul Billings, July 20, 2007)

From that first ad, Billings and members of the Screening Group collected approximately 100 anecdotal cases that illustrated the problem they had identified. These formed the basis of their 1992 manuscript. In the first

funded follow-up to this study, the DOE-NIH awarded a grant to the Shriver Center in San Francisco for Paul Billings and Marvin Natowitz as principal investors, along with other Screening Group members Lisa Geller and philosopher Carol Barash, for a systematic investigation and case study analysis of genetic discrimination. The authors mailed 27,790 questionnaires, and followed up 206 of 917 responses with telephone interviews. The results, published as Geller et al 1996, found that 22% of survey respondents who self-identified as being at risk for a genetic condition had experienced discrimination from "health and life insurance companies, health care providers, blood banks, adoption agencies, the military, and schools" (Geller et al 1996:71). The authors concluded that "[t]he presence of abnormal genes in all individuals makes each person a potential victim of this type of discrimination (Geller et al 1996:71).

Why Publish in the *AJHG*?

What did members hope to accomplish by publicizing their findings in the *AJHG*? They did not oppose genetic testing or genomics research. Beckwith (2002) in particular takes great care to make this point in describing the relationship of his activism to his work as a genetics researcher. Members of the Screening Group were, in fact, cautious proponents of genetic testing. One of their goals in publishing their findings was to alert clinicians and geneticists to the possibility that genetic information was creating a social underclass of the asymptomatic ill.

According to Jonathan Beckwith (2002), members of the Screening Group chose the *AJHG* for publication because they held a long-standing

commitment to educating geneticists about the social impacts of their research. Also, says Beckwith 2002, they "thought the journal would be receptive."[14] At an earlier point, in his memoir, Beckwith (2002) has also described having a strong personal and professional commitment to alerting his colleagues to the dangers of revisiting and naturalizing eugenics reasoning in their research findings. Throughout his career, he has expressed alarm that geneticists did not show the same commitment as physicists to recognizing—or challenging—the social impact of their work. He reiterated this motive during an interview:

> I certainly have a strong feeling about trying to alert scientists that what they're doing can also have some serious negative consequences, and they should become involved in trying to ensure that these things don't happen. The American Society for Human Genetics set up an ethics committee that started publishing articles like that. And it's a journal that, particularly in the genetic era, started to get a lot of attention from the media. You'd see articles that were published in the journal being picked up by the newspapers. (Interview with Jonathan Beckwith, May 6, 2008)

Beckwith also observes that the media were picking up articles in the *AJHG* at this time. They knew that publishing in the *AJHG* would ensure a wide audience for their findings and their message.[15]

But another goal of the Screening Group appears to have been health care reform. Although the authors proposed passage of legislation protecting the privacy of genetic information as one mechanism to address genetic discrimination, and argued the need for "broad-based public consensus" on the appropriate use of genetic tests, they also emphasized the need for "alternative medical care systems" and "changes in the

prevailing American health care system" lest "many healthy and potentially productive members of our society will suffer genetic discrimination" (Billings et al 1992: 481-482).

This aspiration is reinforced by Paul Billings's comments about who Screening Group members perceived as vulnerable and why.[16] To say that members of the Screening Group were concerned exclusively with the practice of genetic exceptionalism by insurers would be misleading. According to Billings, the vulnerability that concerned them was that of individuals with rare disorders to the health care system in general. Billings is unequivocal: the reason that individuals experienced discrimination by insurers was not because their disorders were genetic, but because the insurance industry simply was unfamiliar with rare disorders:

> I think that we have to be careful. Rare disorders are not handled well by the private health insurance system, whether the rare disorder is genetic or otherwise. It has nothing to do with the genetic nature of the problem; it has to do with the rareness of it, and that people have fear about rare disorders. It's a special situation, let's put it that way. The question is, as genetics becomes more common in medical practice because it's relevant more to common diseases, that genetic discrimination will change. And I think that's true. But in the meantime, we need to protect the people with the rare genetic variants. It just happens that the technology developed in such a way that it was better for identifying small groups of people with rare disorders, while it was working up the methods to be able deal with more common diseases, and multiplex variability. In the meantime, these people with these rare things are having all these problems. So the idea is to protect them. By protecting them, and by showing allegiance to them, you're also creating a world in which the common variants will feel safer and more inclined to search out those risks and act on them. (Interview with Paul Billings, July 20, 2007)

Reception to the Study and Publication

Reception to the group's findings and their article was mixed. While the publication garnered tremendous media attention (Beckwith 2002; Reilly 1999), with Paul Billings appearing on the ABC News Nightline program, scientists and academics offered more measured, even critical, responses. Neil Holtzman, in an editorial with Mark Rothstein in the same issue (Holtzman and Rothstein 1992), reiterated his earlier position (Holtzman 1989) that insurer-based genetic discrimination was one aspect of a renewed eugenics brought about by expansions in and commercialization of genetic testing. The more alarming prospect, according to Holtzman, was that insurers might deny coverage to families who knowingly brought fetuses with rare genetic-based disorders to term.

Skepticism greeted the authors' claim that genetic discrimination was widespread—or even a genuine problem. As might be expected, one source of this skepticism was the insurance industry itself. J.A. Lowden, an insurance professional with Crown Life Insurance in Toronto, argued that the twenty-nine cases of discrimination that fit the authors' own criteria was a "remarkably small" number (Lowden 1992:902), given the numbers of insured individuals in the northeastern United States, where the study had focussed. He suggested that these cases indicated "mistakes in judgment based on lack of information," rather than intentional discrimination on the part of insurance companies, and that errors in judgment could be corrected through better education of medical directors in the insurance industry by the genetics community.

Another source was legal scholar Phillip Reilly (1999), who criticized the group's methodology, particularly the absence of any follow-up

interviews, and the use of overly-broad categories to capture instances of discrimination. Citing empirical research he conducted with legal scholar Jean McEwen on the Screening Group's findings, in which they surveyed life insurance companies and insurance commissioners in all states (McEwen and Reilly 1992; McEwen et al 1993, 1994), he argues there is no evidence that consumers had filed suits against insurance companies over genetic discrimination, nor any evidence that life insurers make use of genetic information.[17] Reilly concluded that the authors' study illustrated that genetic discrimination is in fact rare, and attributed the perception that insurers widely discriminate against individuals based on genetic information to how strongly Americans value access to health care (and implicitly, how greatly they fear losing it).

Paul Billings is sensitive to criticisms of the Screening Group's methodology:

> This is something I've had to deal with since before that first publication. There were levels of hostility to that. I'm the first to admit that the study was not a perspective double-blinded. This was an anecdotal study. First there was hostility within the science establishment. I've discussed some of this in subsequent publications, this debate about what genetic discrimination was, and was this illness discrimination that we were talking about instead of genetic discrimination, and debate about the term. Then, of course, there was hostility from the insurance industry. Insurers said, one, that this did not occur, and two, that if did it occur, that it was rogue insurance salesmen and certainly didn't reflect the policy on the part of the insurance industry to discriminate against people on the basis of genetics, even when there were examples where there clearly was a policy. At one time we produced letters from family members with Huntington's Disease. The insurers were saying, "Well, we can't insure you, but if you get a genetic test and it turns out negative then, maybe we will insure you." Such letters were generated by the insurance industry. (Interview with Paul Billings, July 20, 2007)

One surprising defender of the insurance industry is Neil Holtzman (see, for example, Holtzman 1987). He has argued that discrimination is to some extent a legitimate practice of insurers, and that some genetic tests have actually reduced the incidence of unfair discrimination. For example, before 1986 when a predictive genetic test for Huntington's disease became available for presymptomatic individuals, the insurance industry routinely refused to insure anyone with a family history of the disorder. The availability of the test means that approximately half who undergo testing for the allele are eligible for insurance coverage (rather than none).[18] This pragmatism is not evident in the 1992 *AJHG* article. This suggests that the chief goal of the Screening Group was to urge health care reform, not simply to target genetic exceptionalism. However, Paul Billings points out that this conclusion is simplistic. The two issues, he says, are intertwined:

> The point is that both are true. I think that there *is* a systemic problem of private health insurance and its dealing with groups that are identified in which they're trying to marginalize and control their costs of tariff. Those groups could be people infected with HIV or people with cancer. It's also true that genetics is a special case, that people have strange ideas about the import of genetics, and that genetics was being hyped a good deal in the nineties. So genetics was both a special case, and a case that illustrated a broader set of issues. (Interview with Paul Billings, July 20, 2007)

Impact of the Publication

The *AJHG* article is the most frequently-cited piece on genetic discrimination, and most writers attribute the expression "genetic discrimination" to the authors.[19] Paul Billings assesses its impact:

> First of all, it fostered a good deal of follow-up work and funding by the ELSI program. It gave some credence to the fact that this issue was the first major policy thrust of the Genome Project, and that caused its own set of controversies. There were plenty of people who thought that the Genome Project was very ineffectual in creating effective public policy on issues like genetic discrimination. It certainly played a big role in the creation of all the state legislation that now exists. And it brought together coalitions that have been working for federal legislation. So, I think that it was the first big public policy discussion that coincided with the funding of the Human Genome Project. (Interview with Paul Billings, July 20, 2007)

The ELSI program that Billings refers to is the Ethical, Legal and Social Implications program of the Human Genome Project (HGP). ELSI was authorized by Congress in 1989 in response to a suggestion by James Watson. In its original conception, it began as a venture in which both the Department of Energy (DOE) and the National Institutes of Health (NIH), joint partners in the HGP, set aside 3-5% of their annual sequencing budgets for task forces, workshops, and studies to explore ethical, legal, and social concerns raised by genomics.

In fact, even before the Screening Group published their findings in results in the *AJHG,* Jonathan Beckwith and Paul Billings had become members of the ELSI Task Force on Genetic Information and Insurance (1991-1993). The body was one of many that the DOE-NIH ELSI Working Group would establish between 1989 and 1997 to explore policy and ethical issues raised by the HGP, including how the insurance industry used genetic information. (I discuss this more in the next section). But Jonathan Beckwith insists that he did not introduce the issue of genetic discrimination to the Task Force; rather, he says, "it was an obvious issue,

it seemed to me, to anybody who'd been thinking about these concerns." (Interview with Jonathan Beckwith, May 6, 2008.)

CRG adopted genetic discrimination as a policy issue from the Genetic Screening Study Group in 1991, just before publication of the *AJHG* article. CRG was for many years the sole U.S. public interest group committed to raising public awareness about the social impact of genetic engineering, particularly with respect to agriculture.[20] Members of Science for the People and CRG included physicist Sheldon Krimsky, biologists Stuart Newman and Ruth Hubbard, and lawyer Phil Bereano. Paul Billings, who was a member of both the Screening Group and CRG, describes how the issue came to the attention of CRG:

> In 1989, Ruth Hubbard, who was a board member of the CRG and one of the founders, and Nahama Wilker, who was at that time the Executive Director of the CRG, invited me to participate in the Human Genetics Committee at CRG. So I started attending their meetings. They were broad-ranging discussions of the social issues around, human genetics and reductionism. I think I shared with them a manuscript at some point, probably around that time or maybe in 1990. They became interested in the issue and wanted to do their own collection of cases, or create a registry. The board of CRG in the early nineties adopted genetic discrimination as an issue that they thought was worth paying attention to, and applied for foundation money to get involved in that during that time. (Interview with Paul Billings, July 20, 2007)

Once it had adopted the issue, CRG became quite active in organizing efforts to pass state legislation:

> CRG took some role in collecting cases. Some cases were directly reported to CRG—people would call CRG and ask for help in one way or another—and CRG also then got involved with discussing this with state legislatures. They were very active in Massachusetts and in New

> Hampshire, and various other states, and in talking to interested staff people about legislation on genetic discrimination. For most of the nineties, it was one of the issues that CRG spent time thinking about and working on. (Interview with Paul Billings, July 20, 2007)

Genetic Discrimination becomes a Public Policy Issue: The ELSI Working Group

Many of the critics from the 1980s and early 1990s who framed their concerns about the rapid development of genetic tests around eugenics and the creation of risk categories continued to work on insurer and employment discrimination throughout the 1990s, particularly at the federal level, with the ELSI Working Group. From 1989 to 1997, the ELSI Working Group, a joint initiative of the NIH and the DOE in their governance of the HGP, provided policy advice to these two federal bodies on a range of issues, especially genetic discrimination, which was flagged as a policy priority from the start.[21] From 1991 to 1993, Jonathan Beckwith, Paul Billings, and Phillip Reilly worked on the ELSI Task Force on Genetic Information and Insurance, one of two task forces struck by the ELSI Working Group, and the first ELSI body to issue policy recommendations on genetic discrimination.[22] In 1993, Lori Andrews, along with Arno Motulsky, sat on a National Academy of Sciences panel that warned about the growing prospect genetic discrimination; she also chaired the ELSI Working Group in 1995 until her resignation in 1996, taking over from Huntington researcher and ADA champion Nancy Wexler.[23] Troy Duster replaced Andrews as ELSI Working Group chair, and from 1995 to 1997, Neil Holtzman chaired the second task group struck by the ELSI Working Group, the ELSI Task Force on Genetic Testing. This task force made minimal recommendations on genetic

discrimination compared to its predecessor, and focussed mainly on laboratory proficiency for rare inherited disorders (Holtzman and Watson 1998).

Timing: Renewed Calls for Health Care Reform

Why were the authors successful in establishing genetic discrimination as a problem of health insurance, beyond the fact that most of the cases they gathered illustrated problems with the health and life insurance industries? I suggest one reason was the timing of the article's appearance. Widespread discontent with the health care system in the late eighties and early nineties, amongst business, physicians, labour, and the middle-classes, prompted renewed calls for health care reform. These calls came primarily from big business (Bergthold 1991; Hacker 1997; Maher 1989; Skocpol 1992; Starr 1991; Swenson and Greer 2002), and from the single-payer movement. The latter emerged in 1987, led by Physicians for a National Health Program (Voelker 1998) and Harvard physicians Steffie Woolhandler and husband David Himmelstein.[24]

Health care reform has been a prevailing theme of the United States in the twentieth century (Rothman 1993), with six major attempts to introduce some form of national health insurance (NHI), beginning in 1915 in the Reform Era with a campaign by the American Association for Labor Legislation for compulsory health insurance for workers, and ending in 1994 with the Clinton Administration's failed Health Security Act (Gordon 2003; Hoffman 2003, 2006; Skocpol 1992; Starr 1982).

The inflation and spiralling health care costs that were hallmarks of the U.S. economy in the 1970s and the early 1980s prompted health care

"cost-containment" measures to restructure the health care system (Quadagno 2004; Rowland et al 1988). The centrepiece of these efforts was managed care, introduced in 1973 through the Nixon Administration's Health Maintenance Organization bill. Managed care, a term coined in 1970 by physician Paul Ellwood, describes a number of employer-funded health care plans including Health Maintenance Organizations (HMOs), Preferred Provider Organization (PPS), and Point-of-Service Plans (POS). It is a model of health care delivery and payment intended to control costs by setting caps on the fees that health care providers can charge for their services. Insurers sell managed care plans to employers, who in turn insure their employees as a group. The 1973 HMO bill increased the number of managed care organizations providing prepaid health plans to Americans through employers, providing an alternative to conventional fee-for-service care and indemnity health insurance plans, which did not reimburse for preventive care, prenatal care, or immunizations. Americans and employers switched to managed care plans gradually: not until the 1980s, during the Reagan Administration, did managed care became the dominant health care delivery and insurance model in the United States.

While managed care has become the only affordable form of health insurance for most Americans (Becker 2007; Dudley and Luft 2001), and has covered subscribers for services not available through indemnity plans, such is the high cost of health care in the United States that health insurance is still out of reach for some forty-seven million (18%) of Americans under the age of 65 because they cannot afford the monthly insurance premiums or cannot find a plan to cover them (Kaiser Family Foundation 2007:1). Most Americans who hold one or more part-time jobs

are ineligible for managed care plans, as are the self-employed, small business owners, and workers in small firms that do not offer insurance (Becker 2007). Even those fortunate enough to receive full-time benefits that include health insurance can find themselves locked out of their policies when insurers impose large increases in monthly premiums following exceptional health care treatment (such as hospitalization or chronic care).

The dynamics of managed care characterize a unique health insurance and health care delivery model in the developed world (Becker 2007). The United States is the only major developed nation where for-profit insurers are the primary gatekeepers to the health care system: they decide which patients can be treated and by whom; what level of service is appropriate; and which treatments are reimbursable and for how much (Becker 2007; Quadagno 2004). Moreover, the U.S. insurance industry is unique among developed countries in practicing medical underwriting to determine eligibility for coverage, insurance premiums, and reimbursement rates in the individual health market (Quadagno 2004).

Despite the adoption of managed care plans by many employers in the late eighties and early nineties, health care costs have continued to soar, placing an increasing financial burden on employers and workers. In 1990, a recession year, health care costs outstripped profits in most U.S. businesses (Swenson and Greer 2002). During this decade, workers had to contend with not only the increases in premiums that employers have passed on to them in lieu of wage cuts (Swenson and Greer 2002), but also increasing unemployment and the replacement of full-time positions with

part-time work, both of which cut them out health insurance coverage altogether. Growing numbers of middle-class Americans were finding themselves shut out of health insurance coverage because of rising health insurance premiums and increasing unemployment (Hacker and Skocpol 1997; Rothman 1993; Schwartz 1988; Steinmo and Watts 1995). The anecdotal reports that the authors of the 1992 *AJHG* publication presented, of individuals being denied health insurance by insurers despite being healthy or having mild impairments, reflected a current of fear among middle-class Americans of being left unable to afford basic health care or being bankrupted by unexpected health care costs.

These fears are well-founded. Since the early 1990s, the number of uninsured Americans had steadily grown for the first time in several decades, with working families making up more than 80% of the uninsured (Kaiser Family Foundation 2007:4). Illness and unpaid medical bills are the leading cause of personal bankruptcy in the United States, and most Americans who experience medical bankruptcy are home-owners and have health insurance (Himmelstein et al 2005). Although the authors of the 1992 *AJHG* publication identified a range of social institutions, including life insurers and employers, as practicing unfair actuarial decision-making based on the genetic information of their clients, it was adverse decision-making by health insurers that became the hallmark of genetic discrimination.

Sixteen years after the *AJHG* publication, genetic discrimination is still largely associated in public consciousness with the misuse of genetic information by health insurers. This is despite the fact the first case of

genetic discrimination was successfully prosecuted by the federal Equal Employment Opportunity Commission (EEOC), the agency that enforces the Americans with Disabilities Act (ADA), in 2001, against Burlington Northern Santa Fe Railway for illegally genetic testing of employees for carpal-tunnel syndrome in violation of the ADA. What has changed since the 1992 *AJHG* publication is the appearance of new actors who have introduced new frames to the national conversation about genetic discrimination. In the next section, I identify these actors and how they have framed genetic discrimination as a problem, to set the stage for Part Two of the dissertation.

1995-2003: NEW ACTORS AND NEW FRAMES

Of the many individual and organizational actors that entered the public policy field on genetic discrimination in the nineties, a small number came to dominate federal activity on the issue and shape public understanding of the problem. One was Francis Collins, who was appointed Director of the National Center for Human Genetics Research (NCHGR) at the NIH in 1993, and its successor, the National Human Genomic Research Institute (NHGRI) in 1997. Another was Kathy Hudson, who currently directs the Genetics and Public Policy Center, and was an Assistant Director of the NHGRI from 1995-2002.[25] A third was Sharon Terry, who directs the largest and most powerful genetics advocacy organization in the country, Genetic Alliance, a coalition of genetics advocacy organizations that conducts research and lobbies politicians. Medical anthropologists Rayna Rapp, Deborah Heath and Karen-Sue Taussig (Heath et al 2004) and sociologist Carlos Novas (2006) have

documented how Terry has transformed herself from a mother of two children with pseudoxanthoma elasticum (PXE), a rare genetic disorder, to scientist and a director of Genetic Alliance.

Organizational actors that had an interest in seeing genomics research move forward also stepped into genetic discrimination politics: the National Breast Cancer Coalition (NBCC), probably the most influential health advocacy organization in the United States; Genetic Alliance, which represents primarily families with rare, single-gene disorders that affect small numbers of people; and the Coalition for Genetic Fairness. These organizations, through their collaborations with the NHGRI and industry on genetic discrimination, exemplify the "biomedical risk thinking" that Nikolas Rose (2001, 2007) describes as indexical of contemporary biopolitics. Other social scientists have explored in detail biopolitics among specific genetic interest groups (Heath et al 2004; Novas 2006; Taussig et al 2003). What I want to stress here is the new message that these organizations began delivering around the issue of genetic discrimination: that everyone is genetically at risk for disease, and everyone is a candidate for genetic testing.

Francis Collins and Personalized Medicine

The appointment of Francis Collins to lead the NCHGR in 1993 and direct HGP efforts under the NIH created a welcoming environment for health and genetic advocacy organizations to step into genetic discrimination politics.[26] Collins, a cystic fibrosis and Huntington's researcher who developed positional cloning, became the most influential and visible advocate for passing federal genetic nondiscrimination

legislation during the 1990s. From the start of his tenure at NCHGR, Collins flagged genetic discrimination as a central policy issue for genomic medicine, and promoted the cause of federal nondiscrimination legislation publicly to the media, to interest groups, and to Congress during annual appropriations speeches.[27] His willingness to work closely with the many highly professionalized health and genetic advocacy organizations on this issue, and his diplomacy and media skills set him apart from James Watson, his predecessor at the NIH.

Personalized medicine emerged in the mid-1990s as both a central platform in the NIH genomics research programme, and a discourse of individual responsibility among advocacy organizations. In 1995, as head of the NCHGR, Francis Collins began promoting personalized medicine as a central pillar of the NIH vision for genomics research in press releases, speeches, and publications. The emphasis on individual responsibility for health, often framed as consumer choice, found natural allies in health and genetic advocacy organizations. These organizations have endorsed the goal of individual prevention through early detection, along with treatment that includes individual responsibility for diet and lifestyle modification.[28]

The National Breast Cancer Coalition and the Hereditary Susceptibility Working Group

Following the discovery by researcher Mary-Claire King of the BRCA1 gene on chromosome 17, which conferred heightened susceptibility to breast cancer, the National Breast Cancer Coalition (NBBC) led the coalition-building on genetic discrimination with NIH scientists and the

DOE-ELSI Working Group. In 1993, the NBCC formed the National Action Plan on Breast Cancer (NAPBC) with the DHHS and with funding by the NIH (National Action Plan on Breast Cancer 2004). The NAPBC identified the issue of genetic discrimination and health insurance as a high priority. The same year, Francis Collins took the helm of NHGRI in 1993 when it was still known as the National Center for Human Genetics Research (NCHGR). In 1995, the NAPBC formed the Hereditary Susceptibility Working Group to discuss policy and legal solutions to the perceived growing problem of genetic discrimination (Parthasarathy 2004).[29] Members of the ELSI Working Group (under the leadership first of Lori Andrews, then Troy Duster) also participated in this public-private partnership. In July of 1995, the ELSI Working Group and the Hereditary Susceptibility Working Group held a workshop on genetic discrimination and health insurance. These efforts produced two publications in the journal *Science* that urged Congress to pass federal legislation to keep genetic information out of the hands of insurers and employers (Hudson et al 1995; Rothenberg et al 1997).[30]

The push by the Hereditary Susceptibility Working Group for Congress to pass federal nondiscrimination legislation was one sign that activism on genetic discrimination in the United States was being institutionalized (see Rabeharisoa 2008). Here was Francis Collins, federal scientist and head of a $3.5 billion federal genomic research project in the United States, along with luminaries such as breast cancer patient advocate Mary Jo Ellis Kahn, legal scholars Lori Andrews, Karen Rothenberg, and Mark Rothstein, and sociologist Troy Duster, advocating passage of federal legislation to keep genetic information out of the hands of insurers and employers so that

Americans could benefit from genomic research (Hudson et al 1995; Rothenberg et al 1997). While their endorsement of federal legislation to ban unfair use of genetic information, theirs was certainly the most public and influential. Passing federal legislation to ban genetic discrimination at the instigation of breast cancer activists had become a common and unifying goal, endorsed by no less than the NIH's genome institute.

The Hereditary Susceptibility Working Group accomplished something else: it reframed the problem of genetic discrimination and introduced a new understanding of who was vulnerable. Compare the framing language and claims in these two *Science* articles to the 1992 *AJHG* article to see how different a problem genetic discrimination had become in just three years. The authors of the 1995 *Science* publication framed their arguments in terms of the utility of genetic testing for individualized, preventive medicine:

> As at-risk populations are identified, research can be done to determine effective prevention and treatment strategies that will lower the personal, social, and perhaps the financial costs of disease in the future. We all carry genes that predispose to common illnesses. In many circumstances knowing this information can be beneficial, as it allows individual strategies to be designed to reduce the risk of illness. (Hudson et al 1995:391)

The "at-risk" populations that the 1995 *Science* article authors referred to were, presumably, women with a family history of breast cancer. However, the authors' inclusive language invited all Americans to think of themselves as "at-risk"—or to imagine that they might belong to a population that would one day be considered at-risk, since all Americans harboured alleles for common diseases. Thus, by 1995, it was not just

individuals with rare genetic disorders who were at risk of genetic discrimination, but potentially, all Americans. This shift in focus away from single-gene disorders and towards multifactorial disorders reflected growing excitement about the discovery of susceptibility genes for common disorders and the promise of genomics for treating cancer and heart disease, but it also implied that Americans with a known elevated risk of disease like breast and colon cancer might be at greater risk of genetic discrimination than individuals with single-gene disorders.

This was quite a different message than the one delivered by the Screening Group in the *AJHG* three years earlier. The vulnerable Americans that the Hereditary Susceptibility Working Group authors identified were individuals with genetic predispositions to common diseases such as breast and colon cancer. What made them vulnerable to genetic discrimination was not that breast and colon cancer were not handled well by insurers, but the presumed willingness of at-risk Americans to seek individual preventive strategies—including susceptibility genetic testing for multifactorial disorders—to identify and reduce their individual risks of developing debilitating diseases. This imagined relationship between Americans and genetic testing, with Americans depicted as entrepreneurial agents managing their health from prevention through treatment, is consonant with what Robert Crawford (2006:419) describes as the "self-making salvation" of health practices and Nikolas Rose (2001) describes as the contemporary biopolitical emphasis on the pursuit of maximal fitness of the population through the molecular reshaping of the individual.

The tone of the 1995 *Science* article was also different from the 1992 *AJHG* piece: optimistic, even heroic. By privileging susceptibility testing for common disorders over testing for single-gene disorders, the authors brushed away the historic import of eugenicist concerns to genetic discrimination, emphasizing instead a future in which individuals would take control of their health—even though rare, single-gene disorders have been the locus of historic genetic discrimination practices through mandatory sickle-cell screening programmes and workplace susceptibility screening practices.[31] Similarly, it is the disabling or lethal rare genetic and chromosomal disorders which have few available treatments—cystic fibrosis, Down's syndrome, sickle cell anemia, Tay Sachs disease, and spinal bifida—that are the targets of reproductive screening and prenatal testing practices.[32] This forward-looking tone was also evident in the 1997 press release for the second publication from the Hereditary Susceptibility Working Group (whose membership had by this time changed), on workplace discrimination, which made no reference to past workplace genetic screening practices (Rothenberg et al 1997). Author Francis Collins emphasized the central place of personalized medicine in genomic research, describing genetic information as "a valuable part of individualized, preventive medicine that focuses on keeping a person well" (National Human Genome Research Institute 2008).

The authors of the 1995 *Science* article introduced something that had not been present in the 1992 *AJHG* article: a citizenship frame. The inclusive language of "we" invited ordinary Americans without rare, single-gene disorders to imagine themselves entering the arena of genetic testing for common disorders. But the "we" was not just a populist

invocation of "us Americans." The authors singled out fear of genetic discrimination as a significant barrier, not just to Americans reaping the collective benefits of genomic research, but also to their participation in ongoing research (Hudson et al 1995:291). What was at stake here was not only the rights of Americans to profit from their tax investment in a national genomics enterprise, but also the need of federal scientists and health advocacy organizations for Americans to participate in ongoing research to sustain this enterprise. This discourse of citizenship is fully elaborated during the SACGHS hearings. I explore this in Part Two of the dissertation.

Passing Federal Legislation becomes a Unifying Goal

Other events during this decade indicate that legal and regulatory approaches to protect workers and the insured gained momentum. In 1995, the federal EEOC redefined disability to extend ADA coverage to protect workers from discrimination based on genetic information in the workplace. However, many legal scholars have agreed that while this decision may protect workers with an established disability caused by a genetic condition against workplace discrimination, the ADA is unlikely to protect healthy, asymptomatic individuals who carry an allele associated with a disorder, despite the argument made by Billings et al (1992) that genetic discrimination turns on the perception that an asymptomatic carrier is seriously ill or potentially disabled (Gostin et al 1999; Holtzman and Rothstein 1992; see Alper 1996 and Natowicz et al 1992 for a dissenting view). The perceived failure of the ADA to protect asymptomatic or healthy individuals carrying a genotype associated with a

disorder against workplace discrimination is cited by activists as a reason to push for federal nondiscrimination legislation.

In 1995, legal scholar and bioethicist George Annas introduced the *Genetic Privacy Act: A Proposal for National Legislation* (Annas et al 1995), which he subsequently published in 1996 with his colleagues (Roche et al 1996). The Act was a broad-reaching bill designed to regulate not only the use of genetic information but also the collection, analysis, and storage of DNA samples.[33] The Genetic Privacy Act was an example of a prominent use of a privacy frame (Frankel 1999) to set out special status for genetic information (Everett 2004; Parthasarathy 2004). Of the three authors, only Frankel notes that the Act was drafted during a period in which information privacy had become a major public policy and civil rights issue.

Although the Genetic Privacy Act was not passed into legislation, the Health Insurance Portability and Accountability Act (HIPAA) was passed the next year, in 1996. HIPAA was considered a milestone by genetic discrimination activists and offered some of what they were seeking: a prohibition against health insurers treating genetic diagnoses of Americans in group insurance markets as evidence of a pre-existing condition. HIPAA amended an earlier federal act, the Employee Retirement Income Security Act (ERISA), by closing some loopholes that allowed health insurers to discriminate against participants in group health plans based on health information. This effectively protected the majority of already-insured, working Americans against genetic discrimination. What HIPAA did not do was protect Americans in

individual health plans against the unfair use of genetic information by insurers (because it only applied to workers enrolled in group plans). Nor did it address employers' ability to access workers' medical records or medical histories. HIPAA also did not apply to workers enrolled in ERISA-exempted employer self-insured group plans. This was problematic as some of these self-insured plans had the appearance (to subscribers) of regular group plans administered by private insurers (and therefore, subject to HIPAA rules). Finally, HIPAA did not provide Americans with protections specific to genetic information: it did not prohibit insurers from requiring genetic testing or denying insurance coverage using genetic information (however obtained). These problems became widely known in the genetic activist community as "the HIPAA gap."

The year 2000 brought another legislative milestone, when U.S. President Bill Clinton signed an executive order banning all federal departments and agencies from using genetic information in any hiring or promotion decisions. A number of medical professional organizations endorsed the order, including the American Medical Association, the American College of Medical Genetics, the National Society of Genetic Counselors, and the Genetic Alliance (U.S. Department of Energy 2008).

During this time, forerunners of the Genetic Information Nondiscrimination Act (GINA) were introduced annually into Congress, starting in 1995-1996 and continuing until May 2008, when GINA was passed by both houses and signed into law. Much of the work to persuade Congress and the Senate approve GINA was done by two, close-knit organizations: the Genetic Alliance, and the Coalition for Genetic Fairness.

The Genetic Alliance and the Coalition for Genetic Fairness

In 1995, the NBCC had begun the Sisyphean task of persuading Congress to pass a federal law banning insurers and employers from using genetic information in their decision-making. Towards the end of the nineties and into the twenty-first century, the Genetic Alliance and the Coalition for Genetic Fairness—both lead by Sharon Terry—took over this task and became the lead advocacy and lobbying organizations for federal legislation banning genetic discrimination.

The Genetic Alliance, an umbrella genetic advocacy organization for mostly rare genetic disorders that began in 1988 as the Alliance of Genetic Support Groups, exemplifies what Carlos Novas (2006) calls "the political economy of hope" among genetic advocacy groups in the United States. The organization works to "accelerate translational research; improve the climate for the development of technologies; encourage cohorts for clinical trials; increase the availability of linked, annotated biological resources; and ultimately lead to improved human health" (Genetic Alliance nd). It lobbies politicians and scientists on behalf of its 600 member organizations for the translation of research into treatments for "individualized decision making;" it also facilitates networking, partnering, and training opportunities for its members. Some of its member organizations have existed for decades (e.g. the Little People of America and the National Ataxia Foundation, both founded in 1957). Most, however, sprang up in the 1990s, when the federal government committed $3 billion to the HGP.

The Coalition for Genetic Fairness is a single-issue organization that formed in 2000 to lobby Congress and the Senate to pass comprehensive

federal legislation outlawing genetic discrimination. It continues to be the sole genetic discrimination activist organization. Members initially came from patient advocacy and civil rights organizations, but in 2005, it began accepting industry and employers as members.[34] In the same year, it hired a lobby firm (B&D Sagamore, now Baker & Daniels) to develop a strategy for passing GINA (Center for Genetics Research Ethics & Law 2005).

By collaborating directly with the NCHGR and the NHGRI on policy and legal solutions, and building allies among the many industries that support genomics research, the Genetic Alliance and the Coalition for Genetic Fairness became the lead organizations outside of the NHGRI on genetic discrimination. Yet despite efforts that these organization made to keep genetic discrimination uppermost in the minds of Congressional Representatives and Senators, when the SACGHS hearings opened in 2003 with genetic discrimination at the top of its agenda, the committee was forced to acknowledge there was little empirical evidence that many healthy Americans had suffered unfair decision-making at the hands of insurers and employers. The exceptional measures that the committee took to legitimize genetic discrimination as a public policy problem for the future of genomics further helped to further frame it as a civil rights problem for Americans.

CONCLUSION

Current accounts of how and why genetic discrimination became a significant policy issue in the United States locate its emergence with the DOE and NIH plans of a human genome initiative from 1985 to 1989 (e.g. Frankel 1999; Parthasarathy 2004). It is accurate to say that

organizational talks by the DOE and the NIH for a large-scale project to sequence the human genome were an important impetus for concerns about the collection and use of genetic data. The HGP, through the DOE-NIH ELSI Working Group, also provided an arena for debating genetic discrimination as a policy issue and bringing it more fully into public light. But this explanation does not tell us who defined the problem or shaped public consciousness.

Members of the Boston-based Genetic Screening Study Group, several of whom were activists and members of the organization Science for the People in the 1970s, formally defined genetic discrimination and legitimized it as a problem to the clinical genetics community and the public in 1992. They acted on long-standing personal and professional concerns about the complicity of American geneticists in legitimizing eugenics policies throughout the twentieth century, and on fears about the social impacts of sociobiology and the rapid expansion of genetic testing. Their decision to document instances of genetic discrimination was also informed by their professional experience with insurers unfamiliar with rare disorders, and by discussions with leaders of the single-payer health reform movement.

By publishing the results of the first study of genetic discrimination in the *American Journal of Human Genetics*, the authors named and framed (Brown 1995) genetic discrimination as a discrete problem, largely one of insurability, whereby individuals with rare genetic disorders experienced discrimination as the unintended consequences of clinical genetic testing. The 1992 *AJHG* article provided an ordering category for the many

problems associated with genetic determinism, and wove them into something more readily identifiable as a single problem. The publication translated Screening Group members' concerns about the revival of eugenic directions in genetic research and social policies into something more specific: unfair and prejudicial practices by health insurers, as well as employers and life insurers, against otherwise healthy individuals who were known to have genetic markers for diseases. This framing moved discrimination away both from its earlier associations with workplace genetic screening and race-based discrimination against minorities, and narrowed its scope somewhat from the broader ambit of eugenicist practices that both Neil Holtzman (1989) and Troy Duster (1990) had outlined.

The remainder of the 1990s saw new actors raise the public profile of genetic discrimination and bring activism into the federal level. Breast cancer activists were instrumental in forging coalitions that would lead to the institutionalization of action on genetic discrimination. The Hereditary Susceptibility Working Group, the Genetic Alliance, and the Coalition for Genetic Fairness led efforts to have Congress pass legislation banning genetic discrimination, along with Francis Collins, Director of the NCHGR and the NHGRI. The new emphasis on personalized medicine, which wed the NHGRI and genetics advocacy organizations, described an imagined relationship between Americans and genetic testing. This was a relationship in which Americans could maximize both the benefits of their tax investment into federal genomics research and their personal fitness through the pursuit of good health and risk reduction (see, for example, Crawford 1994). However, it also left Americans vulnerable to

discrimination by insurers and employers.

With the institutionalization of genetic discrimination as a public policy issue through the 1990s, it might be expected that by the beginning of the twenty-first century, genetic discrimination was a clearly established problem. Yet by the start of the SACGHS hearings on genomic medicine and the health care system in 2003, the problem was still being legitimized as such to the public and in Congress. During the SACGHS hearings, the testimony of participants articulated a set of rights and responsibilities that was hinted at in 1995 by the Hereditary Susceptibility Screening Group. In Part Two of the dissertation, I turn to the SACGHS hearings to show how actors worked to legitimize genetic discrimination as a problem, and identify what was at stake for them in doing this.

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[1] The family in turn threatened legal action against the HMO, and the HMO withdrew its threat.

[2] Although the stigmatizing and discriminatory impact of sickle cell screening permeated social and institutional practices, it was the workplaces cases that critics cited as the most egregious.

[3] If true, this would be the first appearance in print of the expression. However, Bowman did not use the expression "genetic discrimination" in this 1972 conference paper, nor in his other work on sickle cell screening published in the early seventies.

[4] Jonathan Beckwith says that the article "attracted attention because it described individuals who suffered." Interview with Jonathan Beckwith, May 6, 2008.

[5] See especially Beckwith 2002.

[6] Interview with Paul Billings, July 20, 2007.

[7] Interview with Paul Billings, July 20, 2007.

[8] Interview with Jonathan Beckwith, May 6, 2008.

[9] Interview with Jonathan Beckwith, May 6, 2008.

[10] Interview with Jonathan Beckwith, May 6, 2008.

[11] In her dissertation on genetic discrimination and the Americans with Disabilities Act, Susan Vazakas (1993:24) identifies the first published usage of the expression in a 1977 article jointly authored by Leonard Riskin and Philip Reilly (Riskin and Reilly 1977:489). Riskin and Reilly use the term to describe the widespread institutionalized discrimination experienced by African-Americans diagnosed with sickle cell trait in the early 1970s.

[12] Interview with Jonathan Beckwith, May 6, 2008.

[13] Interviews with Jonathan Beckwith, May 6, 2008 and Paul Billings, July 20, 2007.

[14] Interview with Jonathan Beckwith, May 6, 2008. "At the time, I don't think we thought it would be a hostile audience," Beckwith said. "At least my attitude was, we weren't attacking the work they were doing."

[15] Interview with Jonathan Beckwith, May 6, 2008.

[16] It is also reinforced in a 1998 article by Screening Group members Jonathan Beckwith and Joseph Alper. They argue that legislation singling out genetic discrimination is fundamentally flawed, and a better solution would be for the country to move towards a single-payer system (Beckwith and Alper 1998).

[17] One flaw of their methodology is that the authors relied on self-reporting from insurance companies and insurance commissioners.

[18] On this, Holtzman says:

> One has to acknowledge that there's such a thing as fair discrimination at some point. So if the risks get very high from an insurance point of view, the insurer says, "We'll charge a higher premium or we'll exclude them from insurance." For single-gene conditions, take for instance the example of Huntington's disease. I use this example in *Proceed with Caution*. Before the test for Huntington's disease, *every* offspring of a mother or father with Huntington's was ineligible for both health and life insurance. But the test exonerated half of them, so to speak. (Interview with Neil Holtzman, May 31, 2007)

[19] A recent (April 18, 2008) Web of Science search located 248 citations of the Billings et al (1992) publication in the sciences literature. Reilly (1999) calls their work the "seminal study" of genetic discrimination.

[20] Interview with Jonathan Beckwith, May 6, 2008.

[21] Interviews with Jonathan Beckwith, May 6, 2008 and Paul Billings, July 20, 2007.

[22] In its final report issued in May 1993 (NIH-DOE Working Group on the Ethical, Legal and Social Implications of Human Genome Research 1993), the Working Group recommended that the U.S. adopt a system of national health insurance. Alternatively, it urged health insurers to ban the use of genetic tests in their underwriting practices. Although the report was prefaced with a letter from Francis Collins to Donna Shalala, President Clinton's Secretary of Health and Human Services, it made no mention of

genetic discrimination as a barrier to implementing personalized medicine.

[23] Andrews has written critically of the events and working culture at the NIH in her book *The Clone Age* (Andrews 1999), which led to her resignation from the ELSI Working Group.

[24] In 1992, presidential hopeful Bill Clinton campaigned successfully on a platform of health care reform.

[25] The Genetics and Public Policy Center, which is affiliated with Johns Hopkins University, conducts public opinion research and produces policy statements on genetics.

[26] The NCHGR was established in 1989 to govern the HGP for the NIH. In 1997, the NCHGR was promoted into an NIH institute, the NHGRI, with Francis Collins at the head. On May 29, 2008, Collins announced his resignation as director from the NHGRI.

[27] Interview with Amanda Sarata, November 28, 2007, and comments from anonymous informants.

[28] I discuss the place of personalized medicine in the NHGRI's plans in Chapter 6.

[29] I review Parthasarathy's account in Chapter 1.

[30] See also Lauren McCain's (2002) excellent synopsis of these events, and the workings of ELSI during this period.

[31] The authors identify mandatory sickle cell screening programs practices from the 1970s and Huntington's disease from the Billings et al (1992) publication as early and well-documented examples of genetic discrimination.

[32] After newborn screening for metabolic disorders, prenatal genetic testing constitutes the most common form of genetic testing practice in the United States—and has been critiqued by disability activists and scholars as eugenic and class-driven (see Garver and Garver 1994).

[33] See my discussion of this in Chapter 1.

[34] For example, 23andMe and IEEE-USA are industry members. Source: "About the Coalition for Genetic Fairness." Coalition for Genetic Fairness website. Electronic document, http://www.geneticfairness.org/about.html, accessed June 5, 2008.

Part Two:
Building a Genomic Nation

INTRODUCTION

April 14, 2003, marked the finish line of the most ambitious biological research project the United States had undertaken. On this day, the National Institutes of Health (NIH) announced the successful completion of a thirteen-year, international effort by six nations to map and sequence the human genome, the Human Genome Project (HGP)—a project in which the United States had invested heavily, in tax dollars, and in hope.

One of the challenges facing the Department of Health and Human Services (DHSS), as well as scientists at the National Human Genome Research Institute (NHGRI), would be to translate the discoveries of the HGP into clinical and public health practice. To facilitate this work, the DHHS has chartered two committees since 1999 to assess the challenges of genetics discoveries for the health care system: the Secretary's Advisory Committee on Genetic Testing (SACGT, 1999-2002), and the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS, 2003-2010).

On June 11, 2003, just two months after the NIH announcement, the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) held its inaugural meeting. NIH Director Elias Zerhouni stood in for DHHS Secretary Tommy Thompson and delivered the Committee's mandate to SACGHS Chair Edward McCabe and his Committee members. From June 2003 to December 2006, Elias told the Committee, it would make recommendations to the DHSS Secretary about the financing of genetic technologies in the health care system, the merits of undertaking a

large-scale population cohort study, the oversight of direct-to-consumer (DTC) testing, the impact of patenting and licensing practices on access to genetic tests, and the therapeutic potential of pharmacogenomic testing. The Committee would also examine issues and opportunities arising from genomics unrelated to the health care system, such as bioterrorism. Drawing from expert testimony and the public participation solicited for each hearing, the Committee would sift through a range of perspectives, some conflicting, on what priorities the DHHS should address. It would determine how to fit existing federal regulatory apparatuses to these challenges, and advise the Secretary on how to mould a notoriously unwieldy, heavily bureaucratized, and inequitable health care system to reap the promise of genomic medicine (see Appendix D, SACGHS Charter).

A sense of optimism coloured the hearings at their outset in 2003, the deliberations of participants evoking the cumulative hopes of a nation that had promised its citizens powerful new tools to diagnose and treat chronic diseases. But as the hearings unfolded over the next four years, I watched the Committee's deliberations magnify the limited capacity of the U.S. health care system to deliver the NHGRI's vision of genomic medicine. The Committee identified major barriers, in the form of deeply-rooted administrative and organizational problems that plague the current health care system. Some of these problems seemed insurmountable, such as the antiquated and patchwork system of coverage and reimbursement set by the Centers for Medicare and Medicaid Services (CMS).[1] The state of genetics literacy in the country, not only in the public education system, but also amongst most family practitioners, coupled with a shortage of

genetics counsellors for interpreting test results, challenged a presumptive vision of personalized medicine in which millions of Americans would hold informed conversations with their doctors about the probabilistic role of genetics in their susceptibility to multifactorial disorders. Compounding the difficulties of delivering personalized medicine was the unknown—and possibly, unknowable—epigenetics of these disorders (Holtzman and Marteau 2000), making predictions based on genotype uncertain at best (Lock et al 2007). Another problem facing the integration of genomic medicine to the health care system was the threat of a growing DTC testing industry. At the start of the hearings, NHGRI Director Francis Collins singled out DTC genetic testing as "junk science," reminding Committee members of the need to rein in unscrupulous vendors to preserve public faith in the nation's (authentic) genomic enterprise. Yet the active reluctance of the two agencies with jurisdiction over this practice—the Food and Drug Administration (FDA) and CMS—to assume responsibility for this hot potato generated palpable frustration and exasperation amongst industry representatives, public interest organizations, and Committee members. Amidst all these challenges, however, the problem that stole the show at the SACGHS hearings was genetic discrimination.

I will present the discourse on genetic discrimination at the SACGHS hearings in Chapters 7 and 8. But first, I want to situate these hearings in a broader context: the political-economic rationales for the HGP, and the Americanization of genomics.[2] I do this as a reminder of why the United States decided to undertake genomics research in the first place, and to illuminate what is at stake for the NHGRI in resolving public fears about genetic discrimination. To understand the discourse on genetic

discrimination at the SACGHS hearings, I examine the political-economic origins of the HGP and discuss the local development of genomics. I review two cases of the localization of genomics outside of the United States (in France and Iceland), which were characterized by the rejection of American ownership and monopoly of "national" genetic material and information. I then look at how genomics in the United States has developed in its own direction. By the end of Chapter 8, it should be clear why deliberations on genetic discrimination at the SACGHS hearings were woven so tightly into the NHGRI's prerogatives, rather than tied simply to heightened concerns about the fragility of health insurance.

THE HUMAN GENOME PROJECT: HOPES AND RATIONALES

One of the enduring myths about the HGP is that it was launched as a response to a public health objective: to improve health outcomes for Americans by furnishing the knowledge and tools to prevent and treat disease (for example, Collins and Mansoura 2001; Watson 1990). Since the launch of the HGP, the NIH, along with the science and mainstream media reporting on sequencing and mapping developments, has promoted a singular vision of the HGP as a project that would lay bare the molecular structure of human disease and revolutionize medicine. One keenly-articulated hope was that a roadmap to the human genome would lead to the development of treatments, if not cures, for common, degenerative disorders like cancer, heart disease, and late-onset Alzheimer's disease. [3] The HGP was envisioned not only as a project of discovery, the largest "big biology" project that the United States had undertaken; it was also, implicitly, a public health venture. The metaphors that project architects

and politicians used to describe the human genome—a "book of life," a "map," a "code," and "a blueprint of humanity" (Nerlich and Hellsten 2004; Nerlich et al 2002)—communicated expectations that the HGP would furnish powerful tools for improving human health.

This popular origin story ignores evidence that the project was conceived in 1984 by the Department of Energy (DOE), the federal agency that conducts weapons research and development for the Department of Defense (DoD), to test the effects of radiation on the human genome, following studies of genetic damage to survivors of the Hiroshima and Nagasaki bombings (Barnhart 1989; Cook-Deegan 1994). Also missing from this public health story is the fact that Congress approved the HGP to support the development of the biotechnology sector of the economy (Loepkky 2005). A key rationale for the HGP, less widely reported by journalists in thrall with scientific discovery, was technology transfer to the burgeoning U.S. biotechnology industry. As Sheldon Krimsky (1999, 2001, 2003) has argued, genetics is not a stand-alone research enterprise. Since the 1980s, it has developed in a highly-commercialized socio-economic milieu that is oriented towards profit (see also Loeppky 2005, and Sunder Rajan 2005, 2006).

Political scientist Rodney Loeppky (2005), who provides a political-economic account of the origins of the HGP, argues that the state saw its role in the HGP as the architect of capital accumulation through technology transfer. Public funding of the HGP was necessary to build the research infrastructure for the biotechnology industry to develop applications and treatments, thereby "preserving and advancing America's

most strategic competitive assets" (Loeppky 2005:106). The HGP was a calculated gamble by the DOE, and by politicians who pushed to allocate funds for the DOE-NIH partnership, that investing in genomics research would boost biotechnology development in a flagging economy and shore up post-War competitive losses in the manufacturing industry (Loeppky 2005). This political and economic commitment to the HGP was a bold step for a nation that had seen its technological and military supremacy diminished at the end of the Cold War, and one that might reconstitute its national imaginary as a pioneer in science, medicine, and public health.[4]

Loeppky illustrates his argument using Congressional testimony on the HGP from the mid-1980s to the 1990s. For example, in his testimony at the Hearing on the Human Genome Project, held by the Congressional Subcommittee on Energy Research and Development in 1990, James Watson—co-discoverer of the double helix and head of the Human Genome Initiative and HGP from 1988 to 1992—invoked the vision of the Japanese competitive threat to the United States. Watson argued that United States should build a competitive genomics research enterprise to bolster its economic health:[5]

> As we all know, America is currently the world leader in biotechnology. This leadership is unequivocally being threatened by the Japanese. The human genome project, both through technology and the creation of a powerful infrastructure, is helping to insure this future world leadership. ... Hence, when we are technologically competitive, we can generate positive trade balance. The human genome project to develop a variety of new biological tools and technologies is going to spawn new industrial opportunities that will on the one hand create new industries and on the other hand will give the old industries new opportunities. ... [The] ... genome project will prime the American economic pump. It is a critical time to develop

> these new technologies. If we decline to do so, rest assured our competitors will fill the vacuum. (Testimony of James Watson to the Subcommittee on Energy Research and Development, Hearing on the Human Genome Project. U.S. Congress 1990:91-92)

While the NIH has resolutely championed the public health vision of the HGP and genomics research, particularly under Francis Collins's leadership of the HGP, the DOE has never shied from trumpeting the economic rationales for the HGP or its commercial applications. On its genomics website, which it maintains separately from that of the NHGRI, in a section titled "On the Shoulders of Giants: Private Sector Leverages HGP Successes," the DOE outlines its vision for the HGP:

> The deluge of data and related technologies generated by the Human Genome Project (HGP) and other genomic research presents a broad array of commercial opportunities. Seemingly limitless applications cross boundaries from medicine and food to energy and environmental resources, and predictions are that life sciences may become the largest sector in the U.S. economy.
>
> From the start, HGP planners anticipated and promoted the private sector's participation in developing and commercializing genomic resources and applications. The HGP's successes in establishing an infrastructure and funding high-throughput technology development are giving rise to commercially viable products and services, with the private sector now taking on more of the risk. [6]

Nor has the DOE masked the original radiation research goals that drove the project. David Smith, who founded and directed the Human Genome Program at DOE, explains why the DOE committed to genomics research:

> DOE had been supporting mutation studies in Japan, where no heritable mutations could be detected in the offspring of populations

> exposed to the atomic blasts at Hiroshima and Nagasaki. The program really grew out of a need to characterize DNA differences between parents and children more efficiently. DOE led the development of many mutation tests, and we were interested in developing even more sensitive detection methods. Mortimer Mendelsohn of Lawrence Livermore National Laboratory, a member of the International Commission for Protection Against Environmental Mutagens and Carcinogens, and I decided to hold a workshop to discuss DNA-based methods. ... We concluded the obvious: that if you really wanted to use DNA-based technologies, you had to come up with more efficient ways to characterize the DNA of much larger regions of the genome. And the ultimate sensitivity would be the capability to compare the complete DNA sequences of parents and their offspring.[7]

A key driver of the HGP was the DOE's Charles DeLisi, who was Associate Director of Energy Research at its Office of Health and Environmental Sciences (OHER, now the Office of Biological and Environmental Research). According to HGP chronicler Robert Cook-Deegan (1991, 1994), the DOE's interest in initiating a genome project followed the decline of its Cold War dominance in managing the country's nuclear weapons and safety programmes. The HGP was transformed into a public health project only after the NIH contested the DOE's authority to develop a big science project centering on molecular biology and insisted on coming on board as a joint sponsor with the DOE (Cook-Deegan 1994; Loeppky 2005). Congressional funding for the HGP was legitimized with explicit appeals to shore up America's flagging competitiveness in the global economy, and to build a thriving biotechnology industry with federal investment (Cook-Deegan 1994; Loeppky 2005). Cook-Deegan (1991, 1994) conjectures that the DOE wanted to understand the survivability of humans to a direct nuclear attack. A somewhat broader

research agenda, consistent with the DOE's mandates to investigate the health impacts of energy production technologies and to build a better soldier (e.g. Bickford 2008), is suggested by a 2002 comment by Ari Patrinos, then Associate Director for the DOE's Biological and Environmental Research department, of its interest in "individual susceptibilities to environmental toxins and ionizing radiation."[8]

However, the HGP did not remain solely a public venture. In 1998, Celera Genomics, a private biotechnology company headed by former NIH scientist Craig Venter, stepped into ring by declaring it would complete sequencing of the human genome by 2001—four years ahead of the consortium's schedule. Venter's motivation was both commercial gain and technology acceleration (Fortun 2006). He was interested in patenting the gene sequences that Celera mapped, an approach that the first HGP Director, James Watson, had forcefully rejected. (Under its second Director, Francis Collins, the NHGRI had declared it would make the human genome map and sequencing information publicly available). But Venter's ambition was also to develop more efficient sequencing technologies—a goal, argues Michael Fortun (2006), that was congruent with the original rationale of the HGP, of technology transfer to the private sector. Says Fortun (2006:1), "[i]n a very real sense, then, Venter's history is one not so much of competing with the HGP, but of extending and intensifying some of the original rationales for the HGP."

By challenging the consortium's monopoly on the sequencing efforts, Venter turned the HGP into a public-private race, one that saw the NIH and Celera sling the occasional mud at each other (Fortun 2006). The

significance of Celera's entry into genome-sequencing efforts is that it put pressure on the NIH to accelerate its own efforts (and increase its funding commitment to the HGP). The result that the HGP map was completed two years ahead of schedule, in 2003, with President Bill Clinton recognizing both Francis Collins and Craig Venter as the lead scientists of the HGP.[9]

Governance of human genomics shifted fully to the NIH in 1997, under the NHGRI. That year, the National Center for Human Genome Research (NCHGR) was promoted into a full NIH institute, the NHGRI. Also that year, the DOE created its Joint Genome Institute and applied its resources towards non-human genomics. Because the NIH governs human genomics research (through its genomic institute, the NHGRI), it is tempting to ignore the original military research and technology transfer rationales of the HGP, and see genomics as a public good: a biomedical and public health project. But given the U.S. recommitment to military activity since 9/11 (e.g. Lutz 2002; Masco 2008), its tremendous trade deficit, and the size of its foreign-owned debt, I suggest it is more appropriate to view genomics as a national enterprise that is developing in a climate of economic insecurity and renewed militarization, with research agendas inherited from both the NIH and the DOE. Genomics may revolutionize health care delivery in the United States. But for the time being, it remains primarily a vehicle for building private enterprise and capital accumulation (Loepkky 2005), by encouraging Americans to become genomic consumers—or what anthropologist Kaushik Sunder Rajan (2005:22) calls "patients-in-waiting." It is also a vehicle for reconstituting its global dominance in science and technology.

Genomics has been developed in distinctive national directions. In the next section, I look at two cases of local response to genomics—in France and in Iceland—to highlight what is distinctive about genomics in the United States.

GENOMICS: NATIONALIZED RESPONSES TO THE HGP

Genomic medicine has been labelled a "promissory science" (see, for example, Hedgecoe 2004; Rapp 2006), because its proponents have banked its value on its promised or anticipated, rather than actual, clinical and public health benefits (see also Sunder Rajan 2005, 2006). In his research on postgenomics in the United States and India, anthropologist Kaushik Sunder Rajan (2005, 2006) extends Loeppky's political-economic analysis of the HGP, and treats genomics as a particular form of capitalism: biocapital. To realize the promissory value of genomics, biotechnology and pharmaceutical start-ups must provide technology and capital to transform discoveries into diagnostics, drugs and other treatments. This model, whereby research institutes partner with biotech and pharmaceutical start-ups, was developed in the United States in the 1980s (Sunder Rajan 2006). It followed the liberalization of patenting doctrine, which permitted new forms of biotechnology patenting and licensing. Two cases of commercial genomics partnerships, one in France and one in Iceland, illustrate the clash of this American-bred model with other national practices and values.

France: CEPH and Millenium Pharmaceuticals

In France, a partnership to identify genes responsible for non-insulin

dependant diabetes between the country's main genomic institute, Centre d'Etude du Polymorphisme Humain (CEPH), and the American start-up biotechnology company Millenium Pharmaceuticals, was scuttled in 1994 over a dispute about ownership of genetic material collected from French families. As chronicled by anthropologist Paul Rabinow (1999), the partnership would have given the American company Millenium exclusive access to CEPH's gene bank, built with tissue samples from hundreds of families with diabetes. The principal CEPH diabetes researcher, Philippe Froguel, who opposed the commercialization of the gene bank, appealed to French Prime Minister Edouard Balladur to prevent "French DNA" from being sold to American capitalists. According to Rabinow, the dispute hinged on French patrimonial attitudes towards blood as a substance that was independent of commerce. It also recalled the national blood scandal of the 1980s when officials knowingly allowed blood supplies contaminated with HIV to be transfused into hemophiliacs.

Iceland: The National Health Sector Database

Iceland's controversial National Health Sector Database, which most commentators refer to as the deCODE project, attracted wider and more sustained international attention than the French case. In December 1998, the Icelandic Parliament passed the Health Sector Database Act, granting a monopoly to the American-registered, Iceland-based company deCODE Genetics to develop a large-scale population study, known as the Health Sector Database. deCODE's goals were to isolate genes responsible for disease and develop drug treatments in partnership with Swiss pharmaceutical firm Hoffman-Roche. To do this, deCODE proposed to

link three data sets: Iceland's detailed genealogical records; genetic samples from individuals; and health records at the National Health Service (Árnason and Simpson 2003:537).

The venture would draw on practices and characteristics unique to Iceland, such as the Islanders' long-standing tradition of keeping extensive genealogical and health records, the relative genetic isolation of the population, its small size and cultural homogeneity, and the high level of literacy among Icelanders (Árnason and Simpson 2003). Controversy centred on three issues. One was the monopoly that a single company (deCODE) would have on the database and ownership of genetic information. A second was the presumed consent clause of the Act. This clause required Icelanders to opt out if they did not want to release their medical records, but did not permit them to opt out their deceased relatives. A third was the violation of the privacy and confidentiality of patients' medical records (Árnason and Simpson 2003; Pálsson and Rabinow 2001; Sigurdsson 2001). In November 2003, the Icelandic Supreme Court annulled the Health Sector Database by ruling it as unconstitutional. Some 20,000 Icelanders (approximately 7% of the population) had already chosen to opt out (Canellopoulou-Bottis 2005).

In the next section, I look at two aspects of American genomics: the expanding meanings of personalized medicine, and the "molecular reinscription" (Duster 2006) of race as a marker of genetic difference.

The United States: The Four Ps of Genomic Medicine

The NHGRI has always been clear about the goals and deliverables of genomic research, but somewhat vaguer about the substance of genomic

medicine and the logistics of its execution. What we are told is that genomic medicine, according to NHGRI Director Francis Collins, is just around the corner (see Petersen 2006). In his budget speech to the U.S. House of Representatives in 2003, Collins elaborated on the NHGRI's vision of how genomic medicine will unfold:

> While it always is somewhat risky to predict the future, I want to leave you with my view of where I believe genomic medicine is headed. In the next ten years, I expect that predictive genetic tests will exist for many common conditions in which interventions can alleviate inherited risk, so that each of us can learn of our individual risks for future illness and practice more effective health maintenance and disease prevention. By the year 2020, gene-based designer drugs are likely to be available for conditions like diabetes, Alzheimer's disease, hypertension, and many other disorders. Cancer treatment will precisely target the molecular fingerprints of particular tumors, genetic information will be used routinely to give patients more appropriate drug therapy, and the diagnosis and treatment of mental illness will be transformed. [10]

The crown jewel of NHGRI's clinical deliverables is personalized medicine, which it has tagged "Preemptive, Predictive, Personalized, and Participatory."[11] While personalized medicine is not unique to the United States, in countries such as the UK, it is synonymous with pharmacogenomics, the tailoring of drug treatments to individual genomic profiles. One promising application of pharmacogenomics is the identification of individual variations in the Cytochrome P450 enzyme system, which is responsible for metabolizing a wide range of drugs for depression, anxiety, seizures, blood pressure, coagulation, diabetes, and pain relief (e.g. SSRIs). The potential benefits of pharmacogenomics are clinical and financial: reduced adverse drug reactions, increased drug

efficacy, and cost-savings to institutions and consumers.

Until the mid-1990s, personalized medicine in the United States also meant pharmacogenomics: assigning individuals into risk groups for better drug prescribing. But over the last ten years, personalized medicine has come to mean a model of health care delivery that shifts the focus of clinical medicine from curative to preventive. The centrepiece of personalized medicine today in the United States is the $1,000 personal genome chip, which would identify the approximately ten million single-nucleotide polymorphisms (SNPs) that serve as biological markers for alleles that are associated with diseases. In his 2005 budget appropriations speech to Congress, NHGRI Director Francis Collins explained how the genome chip would be the key to revolutionizing health care:

> The ability to determine the complete genome sequence of an individual could revolutionize medical care. ... Ultimately, NHGRI's vision is to cut the cost of whole-genome sequencing to $1,000 or less, which would enable the sequencing of individual genomes as part of medical care. The ability to sequence each person's genome cost-effectively could give rise to more individualized strategies for diagnosing, treating, and preventing disease. Such information could enable doctors to tailor therapies to each person's unique genetic profile.[12]

Whole genome sequencing, says Collins, would allow individuals to take greater control of their health and even prevent disease using the predictive knowledge generated:

> This approach will allow new treatment strategies that rely on choosing the right medicine for the right person at the right time. In addition, personalized disease risk prediction profiles will allow individuals to make lifestyle and medical choices that delay, or even completely prevent, the onset of many common diseases.[13]

Collins's vision for a personalized health care system is, like the rest of genomic medicine, promissory. The low-cost sequencing technologies that would permit the routine manufacturing of a $1,000 whole-genome chip do not exist: it costs approximately $350,000 to sequence a whole genome. But there is currently a race to produce the technologies that could sequence the whole genome quickly and inexpensively, and some scientists project that a $1,000 whole genome chip will be available by 2015. There are other obstacles to executing the NHGRI vision of personalized medicine. Most doctors do not have the training to interpret the results from sequencing even part of a genome. Indeed, few doctors today might recognize an SNP (Nainggolan 2008). Genetic counsellors are the health care professionals who most likely would be charged with the task of interpreting the whole genome results and counselling clients. Yet there is currently a severe shortage of genetic counsellors in the United States, with only 2,200 currently practicing. Moreover, it is difficult to imagine that insurers might one day reimburse for the cost of whole-genome sequencing, which is the only way that most Americans could embrace it. Given the logistical challenges routinizing whole-genome sequencing for clinical care, the NHGRI's promotion of personalized medicine should be understood as a strategy to encourage Americans to become genomic consumers. In this role, the NHGRI is supporting the state's agenda to further technology transfer and capital accumulation.

And although personalized medicine has not yet been integrated into the health care system, the NHGRI has succeeded in creating markets for services that do not yet exist. Some companies have begun offering DTC profiling services that will fulfil a portion of the NHGRI's promise. Three

companies—23andMe, Navigenics, and deCODEme—will sequence a small percentage of an individual's genome (between 600,000 and 900,000 SNPs) for $1,000.[14] deCODEme, for example, will assess individual risk for twenty-nine diseases (including Alzheimer's disease, breast, colon and prostate cancer, multiple sclerosis, rheumatoid arthritis, and Types 1 and 2 diabetes) and traits (e.g. male pattern baldness). Ironically, in the absence of deliverables for personalized medicine, the promotion of personalized medicine by the NHGRI may have helped to create a market for the unregulated and "rogue" DTC industry.

Sovereign Consumers and Research Subjects

Sunder Rajan's work is essential reading for anyone working on genomics, because he brings a Marxian analysis of industrial and merchant capital (which is absent from ethnographic analyses of genetic testing and screening) to biopolitics. That is, Sunder Rajan examines the promissory value of these genomic practices and how they construct particular subjects. Genomics is one element of a recent phase of capital that he calls biocapital, which emerged in the twentieth century and is technologically driven. He situates genomics in the global marketplace, as a set of practices that produces value through speculation on the production of future commodities—and future consumers.

What is important for my work is the sharp contrast that Sunder Rajan makes between the American sovereign consumer of personalized medicine and the experimental subject in Mumbai, India. Sunder Rajan (2006) argues that personalized medicine configures Americans into what he calls "sovereign consumers." Experimental subjects, on the other hand,

are textile workers that are "recruited into clinical trials" (Sunder Rajan 2006:191). While Sunder Rajan does not explain what he means by the sovereign consumer, he may be referencing the work of economist William Harold Hutt (1936), in suggesting that citizens exercise a limited form of democratic power over (or influence of) producers through their purchasing decisions in the market (see Persky 1993).[15] Sunder Rajan does make it clear that the sovereign consumer is a liberal subject, an interpretation in accordance with Hutt's (1936:257) insistence that the power of the sovereign consumer "maximizes liberty and justice." Thus, the contrast that Sunder Rajan is making is a stark one: between being a liberal subject in a market of choice who exercises some power over producers by forcing them to cater to his or her tastes, and being "consumed" (Sunder Rajan 2006:99) as the raw materials to produce the future commodities of biocapital.

I agree with Sunder Rajan that the idea of consumer sovereignty is a prominent feature of American genomics. The speeches and press releases from the NHGRI on personalized medicine suggest that consumer satisfaction in the form of good health and a satisfying clinical encounter are the ultimate goals of genomic medicine, and that American consumers are autonomous agents who participate in these economic exchanges by making rational, informed, and responsible health care decisions. But Sunder Rajan neglects to draw out the moral duties of being a consumer of personalized medicine, at least in the United States. As anthropologist Nadia Abu El-Haj (2007:291) says, "[t]he concept of the individual at medical risk also presupposes a distinct moral economy that calls on the patient-in-waiting to act responsibly by tailoring her lifestyle to the

specific genetic risk that she bears." I also disagree with how Sunder Rajan contrasts sovereign consumers to experimental subjects, as if Americans (and Indians) might occupy only one of these subject positions, and the two subject positions would be diametrically opposed. The NHGRI's vision of genomics, which the genetic advocacy organization Genetic Alliance also shares, assumes that Americans will be both consumers of personalized medicine *and* research subjects whose bodies are "consumed" (Sunder Rajan 2006:99). I discuss the place of Americans in genomics as research subjects after the following section.

The Revival of Race

Social scientists (e.g. Abu El-Haj 2007; Duster 2005, 2006; Fausto-Sterling 2004; Fullwiley 2007; Kahn 2006; Palsson 2007; Reardon 2004) have observed that race has re-emerged as a "legitimate" category of biological difference in genomics research for studying the distribution of health and disease in populations. What Troy Duster (2006) calls "the molecular reinscription of race" has appeared in numerous international and American research projects. From the ashes of the controversial Human Genome Diversity Project came a new international genetic diversity research initiative, the haplotype map, launched in October 2002 by the International HapMap Consortium (International HapMap Consortium 2003).[16] The project was designed to collect 270 tissue samples from four populations: 90 Yoruban from Nigeria, 45 Japanese, 45 Han Chinese, and 90 U.S. residents of Northern and Western European ancestry. As sociologist Jenny Reardon (2007) points out, the Consortium attempted to address the failures of the Diversity Project through a process

of community engagement. However, Reardon argues, it failed to examine just how it would constitute these populations, and why exactly they would represent homogeneity. In so doing, the HapMap Project opened the door to "re-biologiz[ing] identity along racial and national lines" (Reardon 2007:251). As Troy Duster (2005:1051) reminds us, it is not that race is meaningless; as a lived category of socio-economic stratification and discrimination, it is an important variable in producing health disparities. The danger lies in "reifying race" by creating tissue sample repositories from populations that are defined as biologically distinct and treating these samples as "racially categorized populations" (Duster 2005:1051).

Nowhere has the debate over race and its place in genomics been more vociferous than in the United States.[17] As Harvard historian Evelynn Hammonds (2006) notes, "we are living in the midst of a raging debate over the existence of human races"—"we" meaning "Americans." In a 2005 *New York Times* op-ed, evolutionary developmental biologist Armand Marie Leroi (2005) launched a new sally into the race wars by defending race as a valid marker of genetic variation. If there had been any doubt about the volatility of this terrain, the animated dialogue that this op-ed produced banished that doubt.[18] Based on the work of statistician A.W.F. Edwards (2003), who has found that by examining many genetic loci instead of a single locus, individual geographic ancestry can be classified to five geographic regions, Leroi claimed that the world's people could be classified into five genetically similar groups that are "native to Europe, East Asia, Africa, America and Australasia—more or less the major races of traditional anthropology."[19] Leroi's finding contradicted Richard Lewontin's (1972) classic debunking of the genetic

significance of racial classification—not to mention the earlier contribution of anthropologist Franz Boas (1911) to empirically dismantling the prevailing scientific theories of racial difference.

Race is being used in genomics research in the United States as a proxy for genetic variation, to explain disparities in health and disease that are rooted in biological difference, not socio-economic status. In 2003, the National Human Genome Center at Howard University initiated a project called Genomic Research in the African Diaspora (GRAD), an African-American biobank and health database. Its goal is to collect, by 2008, samples and medical histories from 25,000 volunteers who self-identify as African-American. The NHGRI is also using race a research variable. In May of 2004, the NHGRI announced plans to undertake a large, longitudinal, population-based cohort study to understand the role of genes and environment in health and disease (Collins 2004). This large-scale cohort study, which would recruit 500,000 or more volunteers to provide medical histories, and tissue and blood samples, would track a "representative" sample of the population before disease onset to identify environmental risk factors and biomarkers (Manolio et al 2006).

As Francis Collins (Collins et al 2003) points out, many countries have established large population cohort studies. These include Iceland, the UK, Estonia, Japan, and Canada (Quebec). What is different about the proposed U.S. population study, he notes, is the NHGRI's intent to represent the country's ethnic diversity by over-sampling minorities. According to Collins and his colleagues (Collins et al 2003:84), "if the multiple population groups in the United States and elsewhere in the

world are to benefit fully and fairly from such research... a large population-based cohort study that includes full representation of minority populations is also needed." But the intent of project architects is confusing because they use the words "race," "minority," and "ethnicity" interchangeably. What do they mean? Anthropologist Duana Fullwiley (2007:221) observes that "many geneticists today work with the assumption that human biology differs by race as it is conceived through American census categories."[20] A closer look at the cohort study proposal shows that this logic informs the design. The NHGRI proposes to measure diversity using the 2000 U.S. census classification, with its six racial and ethnicity categories: Hispanic, White, Black, Native American, Asian/Pacific Islander, and Other.[21]

From where comes the impetus to make "group-based diversity" a mandatory locus of genomics research (Abu El-Haj 2007:292)? Anthropologist Nadia Abu El-Haj (2007) cautions us against automatically assuming that institutions such as the NHGRI are behind the push to treat race as a variable in genomics research. Echoing the work of sociologist Steve Epstein (2007) on the institutionalization of inclusion practices in research and clinical trials in the United States, she argues that minority physicians are urging genomic researchers to include race. "Medicine has met identity politics, and out of that meeting point novel practices of both race and medicine (as 'expertise') have been borne," she says. This may well be true. What Abu El-Haj neglects to mention is that none other than the U.S. Congress legislated inclusion practices at the NIH. In 2000, it passed the Minority Health and Health Disparities Research and Education Act, which requires the NIH to conduct research on health

disparities, using race and ethnicity as markers for group difference (Duster 2006:428-429).

The Bioavailability of Americans

Social scientists will, no doubt, continue to debate the merits of using race as a proxy of biological difference. Amidst this debate about the place of race in genomics, a key issue has escaped scrutiny: the assumed availability of Americans to participate in large population cohort studies and clinical trials. This is what I call the "bioavailability" of Americans, following Laurence Cohen's (2004) introduction of the term.[22] The NHGRI's proposed cohort study presents a daunting logistical and ethical challenge: sourcing the 500,000-plus consenting American bodies needed for study. Given that presumed consent will not successfully cross the Atlantic from Iceland (not that it was successful in Iceland), that genetics literacy in the United States is widely perceived as poor, that information privacy is a paramount concern, and that the study data will be publicly accessible—including, one assumes, by other federal departments and agencies, including the FBI and the DoD—one wonders: how will the NHGRI convince 500,000 Americans to hand over their genetic material and medical histories to the U.S. government?

As I will show in Chapters 7 and 8, assumptions about the bioavailability of Americans surfaced during presentations and testimonies at the SACGHS hearings. These assumptions were woven into claims about the right of Americans to consume the benefits of genomics research without losing their health insurance or employment benefits. Race, which was almost entirely absent from this discourse, made one striking

appearance as a metaphor for the second-class citizenship of Americans who have adopted the subject position of "victims" of genetic discrimination. I discuss the use of race as a metaphor during the hearings in Chapter 8.

THE SACGHS HEARINGS

What were the SACGHS hearings, and how did SACGHS operate? In the next section, I describe the origins, scope and functioning of the hearings, and the constitution of the Committee members and the public participants. I then turn to the work of political scientist David M. Ricci (2004) on public citizenship discourse in the United States, to represent the hearings as a site of nation-building and citizenship discourse.

Origins and Scope

The SACGHS hearings, the site at which I examined how public understanding of genetic discrimination is currently being shaped, were an initiative of the Secretary's office of the DHHS. SACGHS was chartered by the DHHS in September 2002, for four years. In August 2006, the Committee's charter was extended for two more years, until December 2008.

The Committee's creation followed on the heels of its predecessor, the Secretary's Advisory Committee on Genetic Testing (SACGT), also a DHSS initiative. SACGT held its hearings between October 1999 and July 2002. While SACGHS inherited some of the issues that SACGT had explored, notably genetic discrimination and the regulation of DTC testing practices, SACGHS addressed a broader set of concerns and issues than SACGT.

Reporting to the U.S. Secretary of Health and Human Services (hence its name, the "Secretary's Advisory Committee"), its mandate was to identify barriers and challenges to integrating genomics into the U.S. health care system, as well as opportunities and ethical concerns arising from genomics. The Committee held public hearings two or three times each year, starting in June 2003 and ending in November 2006. It operations have been managed by a full-time office of four staff members working for the NIH Office of Biotechnology Activities (OBA).

The relationship between SACGHS and the ELSI programme of the NIH needs to be clarified. Given the mandate of SACGHS, it might seem that the Committee was a direct ELSI initiative, or alternatively, that it was chartered by the NIH or the NHGRI. Although SACGHS was not a formal ELSI (Ethical, Legal, and Social Implications of the Human Genomic Project) initiative, and was not mandated or governed by the NIH, its provenance is the NIH-DOE ELSI Joint Working Group, which met from 1989 to 1997. In December 1996, just before the Working Group disbanded, the evaluation committee of the Working Group issued its final report. It recommended that the activities of the Working Group be divided amongst "different committees and at various levels within the government" (National Human Genome Research Institute 2009). The evaluation committee recommended "the formation of three ELSI committees" (National Human Genome Research Institute 2009)—the third of which was "a genetics and public policy advisory committee in the Secretary of the Department of Health and Human Services (DHHS) Office" (National Human Genome Research Institute 2009). This third "ELSI committee" would become the SACGT in 1999, followed by SACGHS

in 2003.[23] Thus, the vision of the Working Group was that the DHHS policy committee would have an ELSI-driven mandate. The relationship of the DHHS policy committee to the mandate of the ELSI Working Group is further clarified in the recommendations section of the evaluation committee's final report (National Human Genome Research Institute 2005). The authors of the report state that "[a] forum at the DHHS-wide level is needed to promote public awareness of ELSI issues," and that this forum "should identify emerging ELSI issues" (National Human Genome Research Institute 2005). Thus, while SACGHS has never been an ELSI body, nor has it been governed by the NIH or the NHGRI, it is arguably a defacto ELSI body, in that it has promoted and addressed ELSI issues—with input from NHGRI Director Francis Collins and NIH Director Elias Zerhouni.

As an advisory committee, SACGHS has operated under the aegis of the Federal Advisory Committee Act (FACA) of 1972. FACA was enacted in an attempt to formalize transparency and democratic engagement in the public policy advisory process, and to guard against complaints of special interest influence. Bioethicist Bethany Spielman (2003: 346), who has examined the origin and rationales of the legislation and its role in bioethics commissions, says that "FACA was enacted in 1972 amidst concerns that some special interests had come to enjoy unchecked and secret access to federal executive decision-makers, thereby subverting the public interest." FACA mandates that federal government consultations with various bodies (e.g. task forces, boards, and commissions) and cross-sections of public interest (e.g. public interest groups, scientists, industry, business, and citizens) on policy issues must be transparent and balanced.

In practical terms, this means that all documents (for example, membership roster, charter, agendas, reports, and correspondence) must be made publicly available, meeting dates and agendas must be advertised so that anyone interested has the opportunity to attend, and Committee membership must be balanced with a range of perspectives so that no single interest or viewpoint dominates.[24] In practice, while SACGHS satisfied this black-letter requirement, it did not achieve the overarching goal of democratic engagement with the public at large. Committee members also found ways to sidestep FACA prohibitions against lobbying efforts—an unexpected move that nonetheless was not protested or challenged by any party. I discuss this first concern later in this chapter, and the second in Chapter 7.

SACGHS Membership

The Secretary of the DHHS appointed two types of members to SACGHS (see Appendix E, SACGHS Roster). The Committee proper consisted of between thirteen and seventeen sitting members, including the chair, who held staggered terms of up to four years. Most members were scientists and clinicians; often a bioethicist, social scientist, and lawyer were also on board. Two of the members were chosen to represent consumer issues and the views of the public. In addition to these sitting members, at least nine federal agencies and departments that have an interest in genomics and genetic testing appointed representatives to the Committee. These federal representatives, called “ex-officio members,” attended all the hearings. Indeed, they were on call at all times to answer Committee members’ questions about the policies and practices of their

agencies or departments.

Under FACA rules, SACGHS had wide latitude to interrogate federal agencies on their mandates and activities, to invite experts to make presentations, and to solicit public testimony on any issue under discussion. The Committee held eleven public hearings of two days each over four years, for a total of twenty-two days of hearings to the end of 2006.[25] It solicited testimony from interest groups, industry, professional bodies, individual experts, and citizens.[26] This made the hearings a key site at which to observe the interests and strategies of those members of the public with an interest in understanding or influencing genomics policy—and the means to attend the hearings.

The SACGHS Public

Who were the public participants at the SACGHS hearings, and how did they participate? In principle, anyone—including non-Americans—could attend the hearings, testify to the Committee in person or in writing, or comment on the Committee's draft reports. Members of the public participated in the workings of the Committee in two capacities: as invited speakers designated as experts in their fields who made formal presentations to the Committee; and as interested members of the public who submitted comments or suggestions on any agenda item, or who testified directly to the Committee (in writing or in person).

While the model for the hearings may have been democratic engagement with the public, in practice, class assumptions were built into the hearings (for example, where hearings were advertised and held) that restricted attendance and participation. The SACGHS staff advertised the

hearings in two federal government venues: the *Federal Register*, "the official daily publication for rules, proposed rules, and notices of Federal agencies and organizations, as well as executive orders and other presidential documents,"[27] which is available in print form and online; and the SACGHS website, which is run by the NIH OBA. Agendas for each hearing were publishing on the SACGHS website two weeks prior to the meeting. Participation, therefore, was generally limited to those who already knew about these venues and had access to them, and to those who could either physically attend the hearings or submit their comments in writing to the Committee. Hearings were held two or three times a year in Washington, DC and surrounding areas, a part of the United States where hotel rooms are very expensive. Attending the hearings also required taking time off from work, because the hearings were held on weekdays during business hours. Although the hearings were webcast on the SACGHS website, they were not televised (as Presidential debates have been since 1960, for example). Anyone interested in watching the hearings via webcast, either live or archived, would have required Internet access.

However, interest groups and advocacy organizations played a role in publicizing the hearings and generating commentary on issues that concerned them, and their efforts brought the workings of SACGHS to a wider audience than would have SACGHS's own limited publicity arms. For example, Sharon Terry of the Genetic Alliance sent formal comments to SACGHS on all of its draft reports. The Genetic Alliance posted Terry's letters on its website, and informally publicized these updates through monthly newsletters for members. These letters were also available to website visitors. The SACGHS staff also publicized their call for public

testimony on genetic discrimination by posting directly on disease-specific list-serves and chat groups, such as the Huntington's Disease Society of America, according to Amanda Sarata.[28] (I discuss this in more detail in Chapter 7.)

Participation at the hearings was restricted also by the complexity of discussion, and by the ambitious agenda, which required that the Committee often move through discussions quite quickly. Discussion at the hearings was shaped by the knowledge, questions, and concerns of the Committee members, all of whom were educated professionals, and some of whom were biomedical specialists. Frequently, the discussion was so complex that only specialists could understand and participate. Examples of this occurred not only during discussions of genetics, but also during sessions on patenting and licensing, and on health insurance coverage and reimbursement. With only one exception that I witnessed, all of the participants who testified in person to the Committee were articulate, their speech suggesting the educational attainment of middle-class professionals. Although the hearings provided an instructive "boot camp" into the workings of the health care system (and this was one of their functions for me), it was clear to anyone attending that they were not the place to ask rudimentary questions about the workings of the health care system, or even genetics.[29]

The Nation's Conversation with Itself

If the SACGHS hearings had been a theatrical spectacle, they might have been called *The Human Genome Project meets the U.S. Health Care System* given what a vast and daunting undertaking they were, even for a

body that had been given four years to fulfill its mandate. Erving Goffman's (1959) social interactionist model of impression management comes to mind. STS scholar Stephen Hilgartner (2000), for example, has used Goffman's dramaturgical approach to show how scientific experts at the National Academy of Science credentialed themselves to their audiences during hearings to produce science advice on diet and health. In the case of the SACGHS hearings, the dramaturgical approach offers a way to analyze, at the level of discourse, the in-person testimony of Americans who adopted the role of victims of genetic discrimination. I do this in Chapters 7 and 8, treating individual testimonies as performances designed to persuade other participants and the audience of the authenticity of their roles and the merits of their claims. Like Hilgartner, I note the importance of in-person testimony to the Committee, and I describe how participants credentialed themselves to the Committee as "ordinary" Americans who were victims of genetic discrimination.[30] Participants also mobilized powerful tropes and mythologies about Americans and the United States that helped to persuade the Committee that genetic discrimination was a threat to all Americans.

But the SACGHS hearings were not simply a set of social interactions where participants managed impressions. They were a formal, public site where participants—Committee members, representatives from federal departments and agencies, industry representatives, individuals, and organizations—configured a vision of genomics in American life. Moreover, the SACGHS hearings were not the only public site for discussion about genomics or genetic discrimination. For example, the Washington, DC-based Genetics & Public Policy Center, headed by Kathy

Hudson, has held frequent policy seminars for the public since 2003. It has also sponsored public town hall meetings on genetics in six communities across the country. How then to characterize the SACGHS hearings amidst all the public discussions in the United States about genomics?

A perspective that is political, rather than social-interactionist, is needed to frame the SACGHS hearings for this dissertation. In his study of norms of good citizenship in America, political scientist David M. Ricci (2004:5) re-interprets anthropologist James C. Scott's (1990) model of public and hidden transcripts, and extends this model the political worlds of elites in complex societies. Ricci portrays public hearings and their documents as "the visible part of any nation's conversation with itself." For Ricci (2004:5-6), official documents as well as "widely publicized expressions of opinion" constitute the public transcript in the United States. They offer "a representative sample of documents and studies bearing on American citizenship." Ricci includes among these public transcripts "the Mayflower Compact (1620), the Declaration of Independence (1776), and the Constitution (1789)" as well as political speeches and significant Supreme Court decisions.

Following Ricci's characterization of public transcripts, I argue that the SACGHS hearings should be regarded as the visible part of this nation's conversation with itself about good citizenship in an era of genomic medicine.[31] Seen through Ricci's lens, the hearings were a prolonged, public conversation where participants collectively imagined the United States as a kind of nation and Americans as a kind of citizen, in dialectic

with genomics. The national and citizen imaginaries that participants outlined during the hearings were not artifacts of the hearings. These imaginaries and messages have appeared before, in discourse by the NHGRI, by the Genetic Alliance, the National Breast Cancer Coalition, the Partnership on Women and Families, and other genetic and health advocacy organizations. What the hearings did, however, was to bring many of these actors to the same stage over a period of four years and amplify these messages and imaginaries in the nation's conversation with itself. What I argue in the dissertation is the SACGHS hearings, as the stage for this conversation and the vehicle for bringing together so many actors, made visible the power relations between the state, health and genetic advocacy organizations, industry, and citizens around genomics as a national enterprise. The hearings allow us to see that there is something else is going on besides activism on genetic discrimination. This would not be evident, I argue, if we were to view the actors and their discourse in separate settings. What takes place on this stage is a process of genomic nation-building. Listening in on this conversation provides some idea of what is expected of Americans as citizens in an emergent genomic nation: who can claim membership in this nation, what rights they can claim, and what duties are expected of them.

CONCLUSION

The SACGHS hearings, which began two months after the announced completion of the HGP, can be treated as a public site of policy-making where experts appointed by the DHHS, as well as government representatives and members of the public, made recommendations to the

Secretary of the DHHS on how to engineer the U.S. health care system to deliver genomic medicine. However, following from David Ricci's (2004) work on norms of good citizenship, I suggest that the SACGHS hearings should be regarded more broadly as this nation's conversation with itself about the place of genomics in American life and good citizenship in an era of personalized medicine. This perspective follows from a political-economic understanding of the HGP as a large-scale biology endeavour that the DOE first initiated to understand the impact of ionizing radiation on inheritance (Cook-Deegan 1994), and that Congress financed (as a joint DOE-NIH undertaking) to provide technology transfer to the biotechnology industry and reinvigorate America's economic competitiveness and technological supremacy (Loeppky 2005).

The NHGRI promissory vision of personalized medicine in the United States is a comprehensive and preventive model of health care delivery in which clinicians will use the individual's whole genome profile to tailor diagnostics and treatments—a vision that Sunder Rajan (2006) argues configures Americans as sovereign consumers necessary to the successful consumption of biocapital. While the "molecular reinscription" (Duster 2006) of race in genomics research is another notable feature of Americanized genomics, overlooked in the public debate about the re-emergence of race is the assumed bioavailability of Americans for research purposes and as experimental subjects, indispensable to the production of biocapital. The NHGRI intends to recruit at least 500,000 American bodies into a large-scale population study that will oversample racial and minority populations. It will also need American bodies for clinical trials to test drugs. These ambitions, in tension with the demands by individuals,

patient support groups, and genetic coalitions for the right to benefit from genomic medicine without fearing genetic discrimination, suggest that more was at stake in the discussions about genetic discrimination at the SACGHS hearings than just the fragility of health insurance in the United States.

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[1] One problem is inefficiency. Clinicians must select multiple clinical and laboratory codes from this hodge-podge system to order a single test from laboratories for which insurers will reimburse. Another problem is that the 1,100 private insurers in the country set their reimbursement rates based on CMS rates. Yet the rates that CMS sets are far below current laboratory testing costs. The outcome is that either laboratories must absorb the costs of the genetic test themselves, or pass the non-reimbursed portion of the test onto the patient.

[2] While "postgenomics" is the name for current efforts to reveal gene function and expression now that genome mapping and sequencing is competed, for simplicity, I use the expression "genomics" to refer to both sets of practices.

[3] In 1989, *Time* magazine noted that the HGP "would launch a new era in medicine" (Jaroff 1989). In 1990, HGP head James Watson (1990:44) said, "When finally interpreted, the genetic messages encoded within our DNA molecules will provide the ultimate answers to the chemical underpinnings of human existence. They will not only help us understand how we function as healthy human beings, but will also explain, at the chemical level, the role of genetic factors in a multitude of diseases, such as cancer, Alzheimer's disease, and schizophrenia, that diminish the individual lives of so many millions of people." In his January 27, 2000 State of the Union address, President Bill Clinton said, "Later this year, researchers will complete the first draft of the entire human genome, the very blueprint of life. It is important for all our fellow Americans to recognize that federal tax dollars have funded much of this research, and that this and other wise investments in science are leading to a revolution in our ability to detect, treat, and prevent disease." "The Human Genome Project, Benefitting All Humanity," The White House Office of the Press Secretary, March 14, 2000. Electronic document, http://clinton4.nara.gov/WH/New/html/20000315_3.html, accessed June 17, 2008. In 2001, Francis Collins (Collins and Mansoura 2001: 221) reminded scientists that "[t]he HGP is laying the foundation for a 21st century revolution in biomedical research and medicine that promises longer, healthier lives for everyone."

[4] See, for example, the DOE's description of the place of the HGP in the American imaginary. "Ethical, Legal, and Social Implications." *To Know Ourselves*, Human Genome Program, U.S. Department of Energy, 1996. Electronic document,

http://www.ornl.gov/sci/techresources/Human_Genome/publicat/tko/08_ethical.html, accessed June 12, 2008.

[5] Parts of Watson's testimony to the Subcommittee matched, nearly word for word, a 1990 editorial in *The Scientist* by Leroy Hood, a molecular biologist and champion of the Human Genome Project. In his editorial, Hood (1990:13) wrote the following:

> America currently is the world leader in biotechnology. This leadership is being threatened by the Japanese. The HGP, through both its technology and the creation of a powerful biological infrastructure, will help ensure this world leadership in the future. In this regard, let me make two points. First, biotechnology offers an opportunity to redress America's failure in industrial competitiveness and the negative trade balance. … The HGP will prime the American economic pump. It is a critical time to develop new biological technologies. If we decline to do so, we can all rest assured that our competitors will fill the vacuum.

[6] "The Human Genome Project & the Private Sector. A Working Partnership." U.S. Department of Energy, Human Genome Project Information. Electronic document, http://www.ornl.gov/sci/techresources/Human_Genome/project/privatesector.shtml, accessed June 9, 2008.

[7] "Evolution of a Vision: Genome Project Origins, Present and Future Challenges, Far-Reaching Benefits." Interview with David Smith, The Seventh International Genome Sequencing and Analysis Conference, Hilton Head, South Carolina, September 1995. http://www.ornl.gov/sci/techresources/Human_Genome/publicat/97pr/evolve.html. June 12, 2008. See also "The Genome Project—Why the DOE?" *To Know Ourselves*, Human Genome Program, U.S. Department of Energy, 1996. Electronic document, http://www.ornl.gov/sci/techresources/Human_Genome/publicat/tko/02_why.html, accessed June 12, 2008.

[8] "Research Abstracts from the DOE Genome Contractor-Grantee Workshop IX." Welcome statement by Ari Patrinos, Associate Director of Science for Biological and Environmental Research, Office of Biological and Environmental Research, U.S. Department of Energy, January 27-31, 2002, Oakland, California. Electronic document, http://genome.gsc.riken.go.jp/hgmis/publicat/02santa/index.html, accessed June 12, 2008.

[9] Margaret Lock, personal communication.

[10] "The Future of Genomics." Statement of Francis S. Collins, M.D., Ph.D., Director National Human Genome Research Institute, National Institutes of Health, Department of Health and Human Services, in Testimony Before the SubCommittee on Health Committee on Energy and Commerce United States House of Representatives, Thursday, May 22, 2003. Electronic document, http://www.genome.gov/11007447, accessed August 9, 2006.

[11] In 2006, the NHGRI began substituting the word "preemptive" for "preventive." Also, in 2008, personalized medicine became "a personalized health care system." The "preemptive" element of the NHGRI slogan appeared in different NHGRI communiqués throughout 2006, 2007 and 2008. See, for example, Zerhouni 2006. See also "Director's Overview" in the NHGRI 2009 budget justification (U.S. Department of Health and Human Services 2008a:12), in which the Institute said, "NHGRI supports the development of resources and technology that will accelerate genome research and its application to human health, thus enabling truly preemptive, predictive, personalized, and participatory health care."

[12] Source: "NHGRI Director's Statement to Congress on the FY2006 Budget Request." Witness appearing before the House SubCommittee on Labor-HHS-Education Appropriations, March 9, 2005 and Senate SubCommittee on Labor-HHS-Education Appropriations April 6, 2005. Electronic document, http://www.genome.gov/13514249, accessed Sunday, July 30, 2006.

[13] See "Moving toward Genomic Medicine: An Innovative and Proactive Approach" (U.S. Department of Health and Human Services 2008a:14).

[14] Since writing this dissertation, sequencing prices have fallen dramatically. Companies have also changed their marketing strategies. In January 2010, 23andMe was offering a package called "23andMe Complete" that combines ancestry testing with its Health Edition (sequencing for disease markers) for USD $499.

[15] According to economist Joseph Persky (1993:185), Hutt's vision of the sovereign consumer was a liberal subject who enacted democratic principles and "voted" with his dollars. "For Hutt," says Persky (1993:186), "consumer sovereignty is a mechanism developed through social evolution to discipline producers to the wants of consumers without threatening political legitimacy."

[16] The Human Genome Diversity Project, or HGDP, was a controversy-plagued initiative by population geneticists to conduct a large-scale study of human genetic diversity in select populations. Some critics, including members of indigenous advocacy organizations, dubbed the HGDP "the vampire project" and "biopiracy" because the project architects planned to collect commercially valuable tissue samples from remote populations that were in danger of extinction. The HGDP also came under fire for a project design that treated isolated populations as genetically-distinct racial groups unaffected by recent human migration, and for presuming that informed consent was a universally-valid concept. See, for example, Marks 1995.

[17] Margaret Lock, personal communication.

[18] To offer one example: the Social Science Research Council sponsored a web forum on the question of race in response to Leroi's op-ed. This forum generated fourteen rebuttals to Leroi's claims. See "Is Race 'Real'? A web forum organized by the Social Science

Research Council," June 7, 2006. Electronic document, http://raceandgenomics.ssrc.org/, accessed June 13, 2008.

[19] See also Excoffier (2003:R134), who says that "self-reported ancestry is a good predictor of one's genetic make-up."

[20] See Epstein (2007:148-150) on the development of racial and ethnicity categories for use in the U.S. census, and how the DHHS and the NIH have adapted to these changes.

[21] Source: "Design Considerations for a Potential United States Population-Based Cohort to Determine the Relationships Among Genes, Environment, and Health: Recommendations of an Expert Panel." Source: National Human Genome Research Institute, National Institutes of Health (nd.) Electronic document, http://www.genome.gov/Pages/About/OD/ReportsPublications/PotentialUSCohort.pdf, accessed June 21, 2008.

[22] I use the term to describe the demand for, and scarcity of, American bodies needed to conduct population studies and clinical trials to advance genomics research, commerce, and medicine. See my discussion of bioavailability in Chapter 3.

[23] I am grateful to Sarah Carr in the NIH Office of Biotechnology Activities for clarifying the origins and administration of SACGHS.

[24] Public Law 92-463, 5 U.S.C., App. Electronic document, http://www.gsa.gov/gsa/cm_attachments/GSA_BASIC/without_annotations_R2G-b4T_0Z5RDZ-i34K-pR.pdf, accessed June 8, 2006.

[25] This figure only refers to the public hearings. Committee members and sub-Committees met outside of the public hearings.

[26] By "testimony" and "testimonies," I mean the written and oral statements that members of the public submitted or delivered to the Committee. I use the term "presentations" to describe the formal presentations that guest speakers made to the Committee, and "comments" to describe reaction by Committee members (regular and ex-Officio), or discussion amongst them. I use the term "discourse" to describe all of the written and oral conversations, arguments and speeches that comprise the hearings, as well as the documents the Committee produced.

[27] Source: "Federal Register: About." Electronic document, http://www.gpoaccess.gov/fr/about.html, accessed July 3, 2008.

[28] Interview with Amanda Sarata, November 28, 2007. Sarata was a full-time SACGHS employee from 2003 to 2006 and co-ordinated the genetic discrimination public commentary.

[29] However, there were ways in which anyone attending could obtain information and ask questions away from the "front stage" (Goffman 1959) of the formal hearings themselves. One was through the SACGHS staff. At every hearing, the SACGHS staff maintained a

reception and information table outside of the conference room. One of their jobs was to answer questions and respond to requests for information, which they frequently did by email. The SACGHS staff members who worked inside in the conference room during the hearings were also approachable, if busier. A second means to obtain information was by downloading the reports that SACGHS produced on topics such as coverage and reimbursement, which included primers on the health care system and health insurance.

[30] In treating the hearings as a site where actors made claims and employed rhetoric to persuade their audiences that genetic discrimination poses an immediate threat to Americans, I remain on the "front stage" (Goffman 1959). With the exception of my interview with Amanda Sarata to understand how the Committee arranged the public testimonies, for the most part, I do not try to go "backstage" to identify what happened during the hearings and how or why Committee members acted.

[31] I am adopting Ricci's interpretation of the public transcript as public statements produced by elites in complex societies. I do not adopt Scott's model of power relations, which is based on his research of peasants in the Malaysian state of Kedah. In Scott's model, the public transcript is the public exchange between dominators and the oppressed, while the hidden transcript is the critique of power by the oppressed, which is hidden from elites.

Chapter 7. Legitimizing the Problem: Genetic Discrimination at the SACGHS Hearings (2003-2005)

INTRODUCTION

In this chapter, I draw from my ethnographic research of the SACGHS hearings to show how genetic discrimination arrived on the Committee's agenda and how participants at the SACGHS hearings framed it as an impediment to progress in medicine and genomic research from the outset.[1] I begin the chapter by describing two imaginaries that were articulated by participants at the start of the hearings: a national imaginary of the United States as a particular kind of nation, and an imaginary of Americans. These imaginaries, which communicated a set of imperatives around the problem of genetic discrimination, set the tone for the October 18, 2004 "Perspectives on Genetics Discrimination" session on genetic discrimination, which I examine in Chapter 8. I then show why the Committee committed itself to gathering stories from Americans about their experiences of genetic discrimination, and show how the October 2004 "Perspectives on Genetic Discrimination" session was produced.

I make two arguments in this chapter. One, participants at the hearings, including federal scientists Elias Zerhouni and Francis Collins and genetics advocates, made a clear link between the public's fear of genetic discrimination, and the nation's progress in genomics and personalized medicine. They argued that fear of genetic discrimination was causing Americans to forgo the genetic testing that would save lives—that Americans were not reaping the benefits of the Human Genome Project (HGP). They also argued that scientific progress was at stake if Americans continued to fear genetic discrimination, because they were declining to

participate in studies and clinical trials. The messages that these participants delivered, about the United States as a nation committed to technological innovation, genetic testing as an essential tool to prevent or control disease, and genetic discrimination as a barrier to progress, have been voiced outside of the hearings since 1995, by the actors that I identified in the latter part of Chapter 5. What the SACGHS hearings did was to amplify these messages, by bringing many of these actors to the same stage over a period of several years. This amplification, and the testimonies that I present in Chapter 8, shows that something else is going on besides activism on genetic discrimination at the hearings, which is not evident when viewing the actors and their discourse in their own domains. This "something else" is a citizenship project, which I discuss in Chapter 9.

Two, the SACGHS hearings illustrate the ongoing plasticity of genetic discrimination. The work that the Committee and staff performed, to legitimize genetic discrimination as a "real" problem affecting Americans, was itself part of the shaping of the problem. The series of steps that Committee members and SACGSH staff took, from discussing the lack of evidence for discrimination with participants at the hearings, to interpreting comments by the SACGHS Chair about the desirability of soliciting public comments on experiences of genetic discrimination, to using the text of the proposed federal nondiscrimination bill (GINA) to word the call for public commentary to the Committee, illustrate how social problems are shaped by the interpretations of individuals and organizations.

GENETIC DISCRIMINATION ARRIVES ON THE SACGHS AGENDA

Genetic discrimination was a priority for the Committee from the start. In the first few minutes of his introductory speech to SACGHS at its inaugural meeting on June 11, 2003, NIH Director Elias Zerhouni singled out genetic discrimination as a "core issue for the country," characterizing it as a major impediment to the practice of medicine and the delivery of health care.[2] Noting that President George W. Bush had endorsed passage of S. 1053, the Genetic Information Nondiscrimination Act (GINA) of 2003, Zerhouni, who handed SACGHS its mandate but did not sit on the Committee, described passage of the legislation as crucial to fulfilling the promise of genomic research and medicine. "There's no progress possible unless we can provide assurances to individuals that their genetic information will never be misused against them," he told the Committee members and attendees. This theme, that the scientific and medical progress of a nation was at stake, and American's fear of genetic discrimination was hampering that progress, would be repeated throughout the hearings.

SACGHS was not the first federal advisory committee to try to define the scope of genetic discrimination. Nor was it the first to try to persuade the Secretary of the Department of Health and Human Services (DHHS) to lobby Congress and the Senate to pass federal nondiscrimination legislation. The Committee had inherited genetic discrimination as an agenda item from its predecessor, the Secretary's Advisory Committee on Genetic Testing (SACGT). SACGT, also a DHHS initiative reporting to the Secretary of DHHS, held its hearings from October 1999 to July 2002.

Genetic discrimination was one of three issues that SACGT had identified as priorities, along with the oversight of genetic tests, and gene patents and licensing. But SACGT's charter expired in 2002 without seeing DHHS implement any of the Committee's recommendations on the regulation of genetic testing. The work of SACGT capped off nearly a decade of failed efforts during the Clinton administration by elite actors to persuade Congress to enact comprehensive federal legislation banning genetic discrimination.[3] These elite actors include NHGRI Director Francis Collins, Sharon Terry and the Genetic Alliance, Kathy Hudson and the Genetics and Public Policy Center, the National Breast Cancer Coalition (NBCC), Congressional representative Louise Slaughter, and legal scholar Karen Rothenberg at the University of Maryland.

In fact, despite its status at the outset as an issue of concern, genetic discrimination competed for the Committee's time and attention with other high-priority items that were perceived to be barriers to delivering genetic technologies through the health care system.[4] But two events lent some urgency to the deliberations on genetic discrimination at the SACGHS hearings. One was the introduction of a new federal genetic nondiscrimination bill in the Congress and the Senate. On May 1, 2003, Congressional Representative Louise Slaughter had introduced H.R. 1910, the Genetic Nondiscrimination in Health Insurance and Employment Act. Three weeks later, on May 27, 2003, Senator Olympia Snowe introduced the Senate version, S.1053, the Genetic Information Nondiscrimination Act of 2003.[5] Given that Congressional subcommittees had stalled similar nondiscrimination bills in other years, that President Bush had declared

his support for federal nondiscrimination legislation, and that the SACGHS charter would expire just after the 2006 midterm elections, there was a palpable sense of urgency at the start of the hearings about seeing a sitting Congress finally pass a nondiscrimination bill into legislation.

The other event was the announcement on April 14, 2003 by the International Human Genome Sequencing Consortium and the NIH that efforts to sequence the human genome were successful. This was not simply the culmination of thirteen years of international collaboration in human genomic sequencing, it was a triumphant declaration that scientists had deciphered the "code of codes" and had found the "holy grail" unlocking the secrets of human disease.[6] The sequenced human genome, declared federal scientists, would usher in a revolution in health care to Americans. Over the next several years, both at the hearings and in NHGRI speeches outside of the hearings, the public would hear that what was holding back this revolution was not the absence of national health insurance for an estimated 46 million Americans, or a health care system woefully unprepared for the arrival of genomic medicine. What has blocked progress, according to Elias Zerhouni, Francis Collins, the Genetic Alliance, the Coalition for Genetic Fairness, the National Breast Cancer Coalition, the National Partnership for Women and Families, Myriad Genetics, and other champions of nondiscrimination legislation, is the public's fears that insurers and employers will misuse their genetic information if they undergo genetic testing, or have a positive genetic diagnosis, or participate in genomic research. The revolution in personalized medicine would happen, advocates argued, when Congress

and the Senate passed federal legislation banning discrimination by insurers and employers.

Given these pressures, it was not remarkable the genetic discrimination was a high-priority item for SACGHS. What was remarkable was the action that SACGHS took on genetic discrimination as an advisory body subject to Federal Advisory Committee Act (FACA) rules.[7]

The Committee took some exceptional measures to document genetic discrimination and persuade Congress and the Senate to pass GINA. By June of 2004, in a formal rankings process of potential agenda topics, members of the SACGHS Committee had identified public concern about genetic discrimination as one of twelve high-priority items, and ranked it highest among five items requiring short-term monitoring and action. The same month, the Committee asked its staff to solicit experiences of genetic discrimination from the public and medical professionals. Amongst those invited to present their testimony to the Committee in person at the Perspectives on Genetic Discrimination session in October 2004 were seven individuals who were called "victims of genetic discrimination." No other issue received this kind of treatment by SACGHS during the hearings, and no victims of any other problem associated with the health care system or genetic testing testified to the Committee.

Although FACA rules prohibit individual Committee members from lobbying politicians, the Committee found creative ways to sidestep this restriction. By April 2005, six months after the Perspectives on Genetic Discrimination session, the Committee had produced a ten-minute DVD

video montage of the in-person testimony from the October 2004 session on genetic discrimination called "Voices of Discrimination." Set to music with a voice-over by the second SACGHS chair Reed Tuckson, the DVD was circulated as a companion to a compilation of public comments on genetic discrimination called "Public Perspectives on Genetic Discrimination: September 2004-November 2004." Because of its size and heft, the volume of public commentary came to be known by the Committee and participants as the "telephone book." The "Voices of Discrimination" DVD was displayed in a plastic pocket on the front cover of the compilation volume.

The Committee also commissioned a legal analysis on existing protections from genetic discrimination from the NIH Office of Biotechnology Activities, which was published in May 2005 (Lanman 2005). The report concluded that "current law does not adequately protect against genetic discrimination based on genetic predisposition" (Lanman 2005:23). The Committee submitted the DVD-public commentary set, along with the legal analysis of current law, to DHHS Secretary Leavitt in May 2005. It also made the DVD-public commentary set available to attendees at the hearings, exhorting them during one set of hearings, to deliver it to recalcitrant Congressional representatives and persuade them to vote for GINA. With this production and circulation of a Congressional lobbying tool, the Committee fully shifted from a facilitator of deliberations on genomics and the health care system, to a participant in collective action on genetic discrimination in its own proceedings.

Chapter 7. Legitimizing the Problem: Genetic Discrimination at the SACGHS Hearings (2003-2005)

SETTING THE STAGE: IMAGINING THE NATION

Before looking at why the Committee decided to solicit testimony on genetic discrimination from the public, I want to explore a national imaginary that participants introduced at the start of hearings. This imaginary set a tone for the hearings and provided a context for the testimonies on genetic discrimination delivered the following year. I identify four nesting themes that comprised this imaginary. Participants revisited these themes over the course of the hearings. The overarching theme in this imaginary was the imperative of American scientific and technological progress. A second theme was that Americans are rational, yet vulnerable, consumers. A third theme was the insistence that everyone has flawed genes. A fourth theme was that genetic discrimination was blocking Americans from participating as research subjects in genomic research and clinical trials, and hence blocking the momentum of genomic research.

The Imperative of American Scientific and Technological Progress

During his opening remarks and induction of Committee members on the first day of hearings, NIH Director Elias Zerhouni left no doubt in the minds of those listening that technology was the real star of the American genomics enterprise. Zerhouni reminded Committee members and the public that the United States was a leader in science and technology:

> On behalf of the Secretary, I want you to know that he supports genetic research and clearly embraces the plan that NIH has developed, in particular focused on the Human Genome Project and its consequences. As Francis and I talk about the future, we like to say that, in fact, 2003 is an important year because of the completion, full

> completion on April 14th of the human genome, 50 years after the discovery of the structure of DNA. My advice to him is to change the calendar for science and start with April 14 being Day 1 A.G., after genome, and anything before that was B.G. (Opening comments by Elias Zerhouni to SACGHS, June 11, 2003)

Zerhouni emphasized the tremendous reach of the Committee's mandate:

> Well, in fact, you are an important Committee in this new era, and your advice and direction is going to be extremely critical. There isn't one aspect of what you're touching that will not affect both research and health care in this country for years and years to come. So it's important, I think, to understand and to realize that the Secretary, myself, all of us are really supportive of your efforts and look forward to your contributions. (Opening comments by Elias Zerhouni to SACGHS, June 11, 2003)

He then reiterated his message that diagnostic technologies were reshaping the United States and that the Committee needed to consider their various impacts:

> I don't want to make a long speech about the varied applications that are going to define the way we perform medicine that are arising from the Human Genome Project, but more importantly it's not just the Human Genome Project that we need to consider. It's all of the diagnostic technologies that are related to the downstream technologies that are emerging from rapid DNA sequencing, from DNA arrays, from proteomics. All of those technologies have an impact, I think, in our era. (Opening comments by Elias Zerhouni to SACGHS, June 11, 2003).

Later that day, Francis Collins, Director of the National Human Genome Research Institute (NHGRI), elaborated on Zerhouni's message that technology was the centrepiece of the nation's plan for tackling disease and producing targeted drugs. Collins delivered a presentation to

the Committee titled "Future Directions in Genetic and Genomic Research," which he had drawn from an article he had published with colleagues (Collins et al 2003) in *Nature* earlier that year. The article had been subtitled "A blueprint for the genomic era." In his presentation to the Committee, Collins outlined the NHGRI's vision of the United States as a genomic nation. Like other scientists who made formal presentations throughout the hearings, Collins had prepared a PowerPoint presentation to accompany his talk, with images to represent the complex vision of the NHGRI for the future of genomics and the nation. Collins began by comparing the genomics enterprise to an architect's designs for a building. He displayed a series of slides that rendered the present and future of genomics into a multi-floor building. Genomics, he told his audience, was a "set of grand challenges:"

> Well, in this article which you have, we depicted this future that we're aiming to try to develop as a metaphorical building, a building resting upon the foundation of the Human Genome Project, as you see here, and consisting of three floors of this rather Frank Lloyd Wright-inspired-looking edifice here. One floor is genomics to biology, another floor is genomics to health, and the third is genomics to society. (Presentation by Francis Collins to SACGHS, June 11, 2003)

Collins acknowledged that some of these "grand challenges" which were the product of "input from more than 600 scientists and ethicists and public policy experts over the course of almost two years," were "perhaps a little on the audacious side." The grand challenges that Collins described to the Committee were technology-driven and relied on the assumption that the cost of technologies would go down. For this to happen, the nation

had to continue investing in these technologies, he said:

> In order to achieve that, we believe that we have to drive the cost of sequencing ever downward. It currently would cost me about $50 million to sequence one of your genomes to a high degree of accuracy, and we clearly can't afford to keep doing it at that rate. So the technology not only for sequencing, which is highlighted here, but for many other applications, like genotyping and looking at gene expression, has to come down in cost, and we will only see that happen if we invest in it. (Presentation by Francis Collins to SACGHS, June 11, 2003).

The NHGRI had a role for Americans as consumers in this blueprint. Collins explained that the NHGRI hoped that a $1,000 personal genome chip would be available by 2015. He suggested that Americans might be keen to have a glimpse of the conditions lying in wait for them, indirectly suggesting that they might be like what Sunder Rajan (2005:22) calls "patients-in-waiting." Americans would want to undergo whole-genome sequencing, Collins suggested, because everyone carries risks for disease. In the past, Collins has used this language of genetic defect to hint at what lies in store for most Americans, and described Americans as having "glitches" in their DNA. In his presentation to the SACGHS hearings on its first day, Collins also used the language of defect to explain what lay in store for most Americans, but referred to "risks" instead of "glitches:"

> Frankly, most of us are carrying risks for future illness somewhere in our DNA, and at the moment we don't have the ability to know very precisely what those are except in instances ... where we already are beginning to get a handle on those conditions. (Presentation by Francis Collins to SACGHS, June 11, 2003)

The appeal of the genome chip, Collins told his audience, was that it would create a permanent record of individual predispositions to illness. This record would also provide a template for interventions:

> But imagine how things would change if we could today sequence the genome for $1,000. Imagine how that would change the way we practice medicine. You'd be very tempted, with appropriate restrictions on access to who gets to peak [sic] at it, to just get the sequence done once and for all and keep it as part of your medical record, and not have to go back and do specific genetic tests on the germline DNA for particular applications. You'd just have it all there, and as new information came along you could quickly in silico determine the consequences and the possible interventions for that individual. (Presentation by Francis Collins to SACGHS, June 11, 2003)

Collins's presentation to the Committee on the opening day of the hearings outlined a future of genomic medicine that revolved around the use of diagnostic and pharmacogenomic technologies not yet available (or affordable) to routinize the delivery of personalized medicine. Collins's vision of a health care system driven by technological innovation and perhaps more appropriately, *reformed* by the genomics initiative, was also shared by members of the Committee. For example, Committee member Reed Tuckson, who would replace Ed McCabe as SACGHS chair in October 2004, offered his thoughts on the task of the Committee on June 11. Tuckson voiced what many tacitly acknowledged in their speech and actions as Committee members: that the American genomics train had already left the station:

> I continue to think that if we're going to as a Committee do right by the American people, there is an inevitable march of science going

> forward. We could and should be advocacy-oriented towards having the basic science research budget being robust in this field. I think that is in America's interests, and if we need to make statements about that, that's terrific, but that's moving on its own course. (Comments by Reed Tuckson to SACGHS, June 12, 2003)

These opening speeches and comments served as a reminder to those gathered, and those watching by webcam, that the United States was a nation committed to technological innovation and to progress in science and medicine. Its genomic agency, the NHGRI, had engineered an ambitious blueprint worthy of architectural great Frank Lloyd Wright to identify the causes of debilitating, common diseases. The nation had the technological prowess, resources, and political commitment to execute this plan. The NHGRI's plan did not address key epidemiological markers of a nation's fitness, such as newborn and infant mortality. But it promised to deliver tools to predict adult-onset risks of many of the chronic, degenerative diseases that occur after the prime reproductive years.

Americans as Rational and Vulnerable Consumers

Participants also produced an imaginary of American as consumers at the hearings. Americans were depicted as two types of consumers: rational consumers, and vulnerable consumers. Both constitutions revolved around Americans adopting genetic technologies in the pursuit of good health (cf. Crawford 2006). Participants at the hearings seemed not to entertain the notion that some Americans might be uninterested in using genetic technologies, incapable of understanding the significance of genetic diagnoses, or simply unable to step into the role of health consumers who exercise choice and make decisions in the best interests of

their health.

The rational consumer that participants at the hearings depicted resembles what governmentality scholar Pat O'Malley (2006) calls the "prudential subject": a calculating and moral actor who locates responsibility for personal health and well-being with himself, and meets those needs by making rational market choices. This rational consumer bears less of a resemblance to the American "sovereign consumer" of personalized medicine in Sunder Rajan's work (2005, 2006), because Sunder Rajan does not depict the sovereign consumer as a moral subject in his model of biocapital production and consumption.[8] Moreover, for Sunder Rajan, the American sovereign consumer of personalized medicine is distinct from the genomic research subject, who is an Indian textile worker and enrols in clinical trials of pharmacogenomic drugs. As I noted in Chapter 6, these distinctions and characterizations are too polarized.

In the guise of rational consumers, Americans were depicted at the SACGHS hearings as intelligent and discerning individuals seeking empowerment, who wanted to make smart choices in the best interests of their health. They were keen to embrace technologies that might offer give them a glimpse of the dangers lurking in their bodies so that they could take action to prevent the onset of disease. Committee member Reed Tuckson told those assembled that the main challenge facing Americans with respect to genomic medicine will be to make smart choices about the new technologies soon available to them:

> I become, as I listen to the conversation, more and more focused on patient education. How do you teach them about direct-to-consumer

> advertising, marketing, expectations? How do you help patients to have a conversation with their physicians and their other health care team so they can make rational, intelligent decisions about the use of this new technology? (Comments by Reed Tuckson to SACGHS, June 12, 2003).

The imaginary of the rational consumer also appeared during a panel discussion on "The Importance of Family History in Health," on October 18, 2004, when Alan Guttmacher, Deputy Director at NHGRI, introduced an initiative developed by the NIH, CDC, and Surgeon General's Office. Called "My Family Health Portrait," the initiative is a web-based pedigree that families can download from the Surgeon General's website to organize and store their medical histories. Guttmacher, who was a much quieter presence at the hearings than Francis Collins, outlined plans by the three agencies to encourage Americans to adopt the tool:

> If we're going to get the public ready to use some of these new genomic tools as they become more available, genetic testing, et cetera, it would be a good idea to have folks become more familiar with some of those concepts by using the old tried and true family history. And preparing both the American public and their health professionals for this coming era of genomics in which we believe that will be a regular part of health care. (Comments by Alan Guttmacher to SACGHS, October 18, 2004)

Guttmacher then introduced My Family Health Portrait as "the first little product of this initiative:

> This is going to be a web-based tool that will be unveiled very shortly -- I'll tell you a bit more about that in a moment -- where individuals and families will be able to download directly to their computer so that this information lives only on their computer, not on some government site, which would be illegal amongst other things, and

> allow people – not just allow people, but give people, we believe, an easy, interactive kind of way to gather their family health information. Then once they've gathered it, in fact, give them some guidance about what they might do with that information. (Comments by Alan Guttmacher to SACGHS, October 18, 2004)

Guttmacher told his audience that the agencies would announce and release the initiative on November 8th, to prepare the public to use the tool on Thanksgiving Day. Their idea was to create a new Thanksgiving Day tradition, in which families collected their family histories of illness, disease, and death to create an online family pedigree at the Family Health website:

> The idea is to make Thanksgiving Day that day when American families by and large traditionally gather together to eat a lot, to watch the Packers on TV and do other kinds of things, to use that family event to actually talk about family history and to gather family history information, the idea that people would have this web-based tool. They could use it that day, they could gather some of the information before, they could gather it afterwards. But the time when the family is really together, when you have Aunt Gladys around who can actually tell you about what you thought you had heard about Uncle Joe or something like that, to get more accurate information. (Comments by Alan Guttmacher to SACGHS, October 18, 2004)

One Committee member, mindful perhaps that recent U.S. Census data indicated that just over half (54.7%) of U.S. households had Internet access, voiced scepticism at the NHGRI's proposal:[9]

> I just wanted to ask a question. Are you going to provide some kind of a mailout so that you could take a pamphlet or something and mail it to your elderly relatives and ask them at their leisure, for those of them that aren't computer literate and aren't able to deal with that, to

> send information back? (Comment by Emily Winn-Deen to SACGHS, October 18, 2004).

Guttmacher replied:

> There will be a pamphlet which will explain sort of the role of family history, why it's important for specific disorders, and has a template in which you can record information that we made available. (Comment by Alan Guttmacher to SACGHS, October 18, 2004).

But Americans could also be indiscriminate consumers of "junk science." In his presentation to the Committee one year earlier, on its opening day, NHGRI Director Francis Collins had warned about the dangers of the growing direct-to-consumer (DTC) genetic testing industry. He turned to a slide in his PowerPoint presentation that showed the home page of web site for a DTC company called DocBlum, which sold "Nutraceuticals for the Millenium" to help with problems ranging from substance abuse to premenstrual syndrome. Although Collins could have represented this industry with any of the dozens of DTC companies that market the same clinically-validated genetic and paternity tests that clinicians order from laboratories, Collins selected a company that markets tests of dubious merit. DTC testing, Collins told listeners, was an unregulated practice that preyed upon the naiveté of consumers:

> A special concern in that regard is the proliferation, mostly on the World Wide Web, of direct-to-consumer marketing of genetic tests, some of which, I must say, are of deeply questionable validity and for which at the present time there seems to be no particular oversight whatsoever. I show you as an example this one from the Web. This is a company that is offering to concerned parents genetic testing for the millennium, as it says here. I'll quote from their Website: "Are

> you concerned about your children's future? Does your child have the genetic trait that leads to disruptive and addictive personalities?" I'm not quite sure how the parent was supposed to know if the child had that genetic trait. Maybe they had a bad day in school. (Presentation by Francis Collins to SACGHS, June 11, 2003)

Collins then told his audience that the real problem with the DTC industry was that it threatened the integrity of the genomics enterprise:

> This is junk science, and it is not the only example that one can find out there on the Internet of similar such things that are happening in greater and greater profusion, and they run the risk, I think, of perhaps fouling the nest here in terms of convincing the public that genetics is junk science in general. If we don't have the ability to restrict in some way the marketing of such information, we may later find out that the public has concluded that this whole field is not something to be trusted. (Presentation by Francis Collins to SACGHS, June 11, 2003)

Four months later, in October 2oo3, the rational consumer and the vulnerable consumer came face-to-face in a discussion by Committee members about DTC testing services and the integrity of the genomic enterprise. Committee member Emily Winn-Deen upheld the sharp distinction that Francis Collins had made on the opening day of the hearings, between the "good science" of genomics researchers and the "junk science" marketed by some of the DTC testing companies. Winn-Deen's comment suggested not only that Americans should reject "junk science," but that they also have an obligation to use "good" science to help themselves:

> I guess one of my big concerns is in order to have the public believe in and reap the medical benefits that genetic testing we hope will offer in the future, not just for highly penetrant monogenic disease but for

> the common complex diseases where your genetic heritage is one component of your health management of your future, I have concern that if genetic testing is used for a lot of "junk science" and consumers lose confidence in its abilities and what it can really deliver because of junk science, then when the good science comes along they won't use it as they could and should to take better care of themselves. (Comments by Emily Winn-Deen to SACGHS, October 22, 2003)

Winn-Deen's comment set off a debate amongst Committee members, with Brad Margus leading the charge. Margus was CEO of Perlegen Sciences, a commercial genetic testing provider that developed off from Affymetrix, and had been appointed to SACGHS from June 2004 until October 2004 as one of its designated consumer voices.[10] He was also a father of two children with Ataxia-Telangiectasia, a rare neurological genetic disorder that impedes motor control and immune function in infants and children. Margus challenged the portrayal of Americans as marketing dupes to his fellow Committee members:

> I'm going to dissent from everyone by saying that I think the web is here to stay, and on average I would argue that consumers are smarter by being able to look up stuff on the web than only relying on their physicians and genetic counselors and all that. All the genetic counselors who are going to speak tomorrow are going to kill me, but I don't think you can avoid the web. Don't forget that everyone in this room is a consumer, and to assume that no one can handle the information themselves... (Comments by Brad Margus to SACGHS, October 22, 2003)

Striking a conciliatory tone and reiterating Margus's point, Reed Tuckson suggested that the task of federal agencies and health care providers was ultimately to empower consumers to take control of their health:

> I think Brad's point is important, and I would think that all of us

> around the table on the Committee would share that the concern in this area is not because we feel that consumers and the American people are not bright and that the inevitability for the movement for more consumer empowerment and more access to information is not only inevitable, as Brad has described, but also desirable.
>
> ... I want to make sure that the sense of the Committee -- and I'm looking for dissent -- is that we absolutely respect the intelligence of the American people and their ability to need to be able to take control over their own health. (Comments by Reed Tuckson to SACGHS, October 22, 2003)

But Tuckson ultimately sided with Collins's mission, of protecting consumers and defending the integrity of the genomics enterprise:

> But as Emily I think rightly points out as well, what happens is that if we don't get on top of this, if people are provided with misleading information, it makes it hard for them to do what they're trying to do. If information is deceptive in this growing area, the natural distrust in this area could also lead to unfortunate decisions being made and an unfortunate level of distrust out there. (Comments by Reed Tuckson to SACGHS, October 22, 2003)

These exchanges demonstrated several things. One, Committee members adopted a limited imaginary of Americans as "the consumer." To the Committee, the American consumer was a homogenous entity, rather than diverse citizens with different health needs and abilities to consume, based on measures such as class, income, age, ethnicity, educational attainment, health insurance coverage, residence (rural or urban), and so on. None of the Committee members demonstrated empirical knowledge of which consumers, in which parts of the country, would be more likely to embrace genomic medicine and why. Nor did their comments indicate they had an understanding of how health disparities impacted Americans differently,

or how the distribution of health disparities might affect participation in this genetic revolution in health care. Secondly, in these exchanges, Committee members indicated that they saw their mandate partly as protecting consumers who might mistakenly seek "bad science" in their zeal for diagnostics and treatments, but more appropriately, to "empower" consumers to reap the benefits of the Human Genome Project. Their comments showed a stark polarization between the "good science" of sanctioned genomic discoveries and clinically-validated tests, and the "bad science" of unvalidated and unregulated DTC testing. Third, it showed that Committee members, like federal scientists Elias Zerhouni and Francis Collins, saw the genomics enterprise as an unquestionable social good that was also unstoppable. The question before them was how to get rational American consumers on board for their maximum "empowerment."

The Language of Genetic Defect

Another theme in the hearings discourse was the persistent reference to the genetic flaws everyone carries, which was a form of genetic egalitarianism. One expression of this genetic egalitarianism was the statement that all Americans are susceptible to genetic disease. For example, during opening day of the hearings, Francis Collins told attendees that "[f]rankly, most of us are carrying risks for future illness somewhere in our DNA, and at the moment we don't have the ability to know very precisely what those are."[11]

This language of genetic defect took a starker form, in the insistence that all Americans have flawed genes. For example, in her September 3,

2004 letter to the Committee, Congressional Representative Slaughter, who has introduced federal nondiscrimination bills to Congress, wrote,

> No human being has a perfect set of genes. In fact, every one of us is estimated to be genetically predisposed to between 5 and 50 serious disorders. Every person is therefore a potential victim of genetic discrimination. (Written testimony of Louise M. Slaughter to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services)

Sharon Terry, writing on behalf of the Genetic Alliance, emphasized the dual dangers of disease and discrimination:

> We also represent those who do not yet understand that 'Genetics is about ALL of us.' [emphasis in the original] Because every man, woman and child has some genetic predisposition, condition or disease resulting from inherited or acquired genetic changes. (Written testimony of Sharon F. Terry to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services)

Terry referred to both "mutations" that everyone carries, and "flawed genes":

> We all possess mutations that will become equally and increasingly transparent with tomorrow's technologies.
>
> ... we all have flawed genes. With so many predictive tests already on the radar screen, we will all be at risk for genetic discrimination. (Written testimony of Sharon F. Terry to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services)

These comments suggest the ongoing cultural power and appeal of the gene as the explanatory unit of biomedical disorder (Nelkin and Lindee 1995). They also suggest that proponents of federal nondiscrimination legislation place a premium on genetic testing, with its promise of delivering actionable knowledge. The assumption is that everyone is ready to be diagnosed for the 5 to 50 serious disorders that they may be carrying. What Slaughter and Terry do not mention is that for many disorders, there are few or no treatments, and the treatments that do exist are not highly-effective. Knowledge of one's susceptibility alleles for disorders like Alzheimer's disease, which cannot be treated, may create despair, not empowerment. For disorders such as breast cancer, there are interventions, but they may be painful, high-risk, and expensive. One prophylactic intervention for women with a family history of breast cancer and a positive BRCA diagnosis is a double mastectomy, followed by reconstructive surgery.

These statements invite another critique. The claim that all Americans carry flawed genes, and that they have mutations that will become transparent, confuses fundamental distinctions between single-gene mutations and susceptibility genes.[12] It denies the complexity of gene expression and epigenetics, including the possibility that susceptibility genes implicated in disease may also have a protective or salubrious influence under certain conditions. Moreover, this claim and its analogues overlook the fact that disease is an object produced through invented categories, shifting scientific facts and diagnostic criteria, changing norms of disorder, and biomedical actors pursing their interests. Finally, it

represents the human genome as a natural object with fixed disease markers, rather than an artifact of human technological intervention that points to hundreds of thousands of years of genetic variation in response to selective environmental pressures.

Genetic Discrimination is Blocking Progress

On the first two days of the hearings, the Committee and the public heard that the United States has a sophisticated blueprint for tackling diseases and enhancing prevention in clinical care, but progress in genomic discovery and delivery of genomic medicine was blocked. The country needed to remove barriers to Americans participating in research and clinical trials, they heard, starting with public fear of genetic discrimination. In his opening speech to the Committee and public, NIH Director Elias Zerhouni stated point-blank that "[t]here's no progress possible unless we can provide assurances to individuals that their genetic information will never be misused against them." He emphasized the importance of passing GINA and volunteered that the legislation "will be a landmark if it goes through." Zerhouni encouraged Committee members to support the process, telling those assembled that "Francis and the Genome Institute and I and many people have contributed in informing both the Senate and the House of the importance of this bill." (Presentation by Elias Zerhouni to SACGHS, June 11, 2003).

Francis Collins followed Zerhouni's presentation on this first day of the SACGHS hearings with details about what form scientific progress would take, and what was at stake for the country if it did not remove barriers to

progress. Presenting the NHGRI's vision of how genomics will translate into health benefits for the country, Collins pressed upon those present the need to conduct a large-scale prospective cohort study to study gene-environment interactions and identify the causes of health disparities among minorities. The chief selling point of a large prospective cohort study, according to Collins, is that it will produce a better understanding of health disparities. Collins told the Committee that "[i]f we're serious about health disparities, for instance, we need to have a plan that involves adequate sampling of some of the minority populations in this country."

The Committee's own estimate of the cost of such a study, which would recruit a minimum of 500,000 Americans as research subjects, was $3 billion—as much as the HGP. Several have argued that the study is methodologically problematic, and that this much money will generate better returns on health outcomes if invested elsewhere (see, for example, Foster and Sharp 2005; Willett 2007). Muin Khoury, a genetic epidemiologist and Director of the CDC's National Office of Public Health Genomics, opposes the study on the grounds that it would be expensive and take years to implement (see Khoury 2004; Khoury et al 2007).[13] Khoury (2004:1) argues that instead of a large-scale cohort limited to the United States, "a coordinated global initiative to carry out and synthesize human genome epidemiologic research worldwide" would better translate genomic discoveries into population health benefits. Collins, however, had a different vision, one that perhaps better captured the imagination of those attending:

> ...here is a case where I think many of us are looking in some

> optimistic way for perhaps a really new and bold enterprise to emerge, we really need in this country a large cohort study of perhaps half a million individuals who are carefully followed over the course of several years for whom a consent has been obtained in a fashion that is able to stand up to all possible standards. They will be involved in an ongoing way in such a study, if it could be mounted, so that it's not a one-time analysis. (Presentation by Francis Collins to SACGHS, SACGHS, June 11, 2003)

In a comment that was reminiscent of James Watson's 1990 admonition to Congress that Japan was outstripping the United States and stealing its crown as the world leader in biotechnology (see U.S. Congress 1990:91-92), Collins reminded everyone that several countries had already launched their own large-scale cohort studies, and the United States was lagging in its efforts:[14]

> It is fine to do a lot of disease-specific case/control studies, and we're all doing lots of those as well, but they're often chosen in a fashion that they emphasize the more severe end of the spectrum of the disease, and therefore they may tend to overestimate the genetic contribution. If we're ever going to sort that out for common diseases, this kind of a large-scale cohort, as is currently being contemplated in the U.K. with their BioBank program, as Iceland is doing in terms of the whole country in collaboration with a company called Decode, as the Japanese are just beginning to mount with their own BioBank program which is about to get underway, but here in the U.S. we do not have such a plan. (Presentation by Francis Collins to SACGHS, SACGHS, June 11, 2003)

Towards the end of his presentation, Collins stressed that the study was something that "we really need if we're ever going to sort all of this out." Sorting it out, for Collins and the NHGRI, meant identifying genetic contributions to common diseases of affluence, and even to mental illness.

Scientists could hit the jackpot, his speech suggested. A PowerPoint slide that accompanied Collins's talk read, "If we do this right, the major contributing genes for diabetes, heart disease, cancer, mental illness, Alzheimer's and Parkinson's disease, asthma, and response to major drug classes will be identified within the next 5 to 10 years."[15]

A little later in his presentation, Collins identified genetic discrimination as a barrier to launching this study and to genomic progress in general. Going through a list of issues he felt that the Committee could examine, he identified genetic discrimination as the top priority. A slide that was labelled "Possible areas to focus by SACGHS" accompanied his comment and provided bulleted instructions to the Committee. The first bullet on the slide read, "Achieving a legislative solution for health insurance and the workplace." In other words, not only was Collins was urging the Committee to puts its energies towards helping to pass GINA, the federal nondiscrimination bill under consideration. He was also directly linking the absence of a federal nondiscrimination law to genetic discrimination as the chief barrier to conducting the large-scale study. If the Committee could work to achieve a legislative solution to genetic discrimination, they would make it possible for genomics researchers to get on with their important work of identifying the "major contributing genes" of a wide range of serious and degenerative illnesses.

Collins was not the only participant at the hearings to argue that fear of genetic discrimination was stopping Americans from reaping the benefits of the HGP and blocking progress in disease discovery and drug treatment. Many of the patient and health advocacy organizations, particularly those

representing susceptibility testing for breast and ovarian cancer, also made these arguments in their written comments to the Committee. For example, in her submission to the Committee on genetic discrimination, Fran Visco, president of the NBCC, argued that "the fear of potential discrimination threatens both a woman's decision to use new genetic technologies and to seek the best medical care from her physician." [16] Visco observed that women are "afraid to enroll in research and clinical trials, and this in turn threatens the ability of the scientific community to conduct the research necessary to understand the cause and find a cure for breast cancer."[17] Myriad Genetics, the only commercial genetic testing provider to submit a written comment to SACGHS, also championed efforts to eliminate fear of discrimination:

> Perception if [sic] reality, and the public's perception is that genetic discrimination is a serious threat. People have allowed an essentially nonexistent or limited risk for discrimination to prevent them from managing a very real risk of developing cancer. We must eliminate the fear of genetic discrimination to allow the public to participate in the benefits of genetic medicine. (Written testimony of Myriad Genetics to SACGHS, Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services)

It was left to Sharon Terry, head of the umbrella organization Genetic Alliance, to stress the responsibility of the broader genetics community to step up and facilitate progress in genomic research for the benefit of all, and not just for women or victims of breast and ovarian cancer. Terry wrote that "it is important that we who carry mutations for disease are

encouraged to participate in genetic research. A fear of discrimination discourages that participation—adding another hurdle to the pathway from basic science and health care services."[18] Terry made the position of the Genetic Alliance clearer in a second statement she submitted to the Committee. "In the midst of the Genetics Revolution, people who could benefit from the new technologies are afraid to use them," she said. Terry warned that "without nondiscrimination assurances, people will not participate in the very studies that could lead to more precise interpretations of 'risk' measures, better understanding about interplay between gene and environment and other genes, and the development of preventative treatments—sometimes for their own condition."[19]

Promoting Scientific Progress beyond the SACGHS Hearings

These themes of the American and consumer imaginary, and their linkages to genetic discrimination, were not artifacts of the SACGHS hearings. Proponents of federal nondiscrimination legislation, particularly NHGRI Director Francis Collins, Congressional representative Louise Slaughter, and legal scholar and law school dean Karen Rothenberg, have been delivering these messages since the mid-1990s. What the SACGHS hearings did was to amplify these messages, by bringing these actors together onto the same stage over a three-year period.

Francis Collins has been warning Americans that they are vulnerable to genetic discrimination for years. In 1997, he delivered the bad news to the Congressional Task Force on Health Records and Genetic Privacy. "Each of us has an estimated five to 30 serious misspellings or alterations in our

DNA; thus, we could all be targets for discrimination based on our genes," he said.[20] The journal *Science* is another forum in which Collins and other advocates of federal legislation have used this language of genetic defect. For example, in an October 2003 editorial authored with his NIH predecessor, James Watson (Collins and Watson 2003:745), Francis Collins hit all of these points home to make a case for Congressional passage of GINA. "All of us carry dozens of glitches in our DNA sequence," the two luminaries told readers, referring to the idea that everyone presumably carries susceptibility genes predisposing them to disease, and therefore, to genetic discrimination. But what really mattered, they said, was the vulnerability of the American genomic research enterprise to genetic discrimination:

> It can slow the pace of the scientific discovery that will yield crucial medical advances. We know that many people have already refused to participate in genetic research for fear of genetic discrimination. Without protections in place, individuals who do agree to participate will represent a self-selected group that could skew research results, producing a negative impact on all of us who look to genetics to help find better ways of diagnosing, treating, and preventing disease. ... The longer this problem remains unresolved, the greater the damage that will be done to U.S. science and medicine. (Collins and Watson 2003:745)

It was in this public statement that the two federal scientists unequivocally declared genetic discrimination to be "a civil rights issue."

Collins continued to drive home his message about the threat of genetic discrimination to scientific progress after the SACGHS hearings had begun. For example, in a February 2005 address to the U.S. Senate on

GINA, he told Senators that "genetic discrimination affects more than jobs and insurance. It also slows the pace of science." Without federal nondiscrimination legislation, he said, "our clinical research protocols will lack participants, and those who do participate will represent a self-selected group, thus further compromising research."[21]

Legal scholar Karen Rothenberg has also been an ardent champion of federal nondiscrimination legislation. In a 2002 article in the journal *Science*, the year before the start of the SACGHS hearings, Rothenberg and Sharon Terry (Rothenberg and Terry 2002) explained why it was so important to allay public fear about genetic discrimination. They played up the enormous promise that genomics discoveries would lead to the delivery of a personalized medicine that focusses on the individual:

> The application of genetics to human health is poised for dramatic expansion. The draft sequencing of the human genome has already led to discoveries about some of the genetic factors contributing to heart disease, diabetes, Parkinson's disease, asthma, and other common illnesses. Before 2010, people may be able to learn their genetic susceptibilities to common disorders, allowing for design of individualized preventive medicine through life-style changes, diet, and medical surveillance. (Rothenberg and Terry 2002:196)

What was needed, they argued, was to "ensure public confidence in genetics research and use of genetic information" (Rothenberg and Terry 2002:196-197). In 2007, Rothenberg delivered a similar message to a different audience, a House Committee holding hearings on the threat of genetic discrimination to workers:

> Fear of genetic discrimination is widespread in the American public.

> ... Fear of genetic discrimination has a negative impact on biomedical research and potentially, healthcare decision making. Genetic research holds tremendous promise to unlock new diagnoses and new treatments, and even to assist in the creation of pharmaceutical therapies tailored to an individual's genetic makeup. However, scientific research and development cannot progress without clinical trials, and these trials can move forward only if individuals who could benefit are willing to participate. Fear that information will become available to and be misused by health insurers or employers has chilled participation in many studies of genetic conditions.[22]

Sharon Terry and the two advocacy organizations she leads, the Genetic Alliance and the Coalition for Genetic Fairness, also have been relentless in alerting the public and politicians to the grave problem posed by genetic discrimination (or the fear of it) to the progress of genomic research. Terry, like Francis Collins, makes an effort to remind those listening about the increasing transparency of the genetic flaws that all Americans carry. In January 2007, the Coalition for Genetic Fairness published a press release on its web site in response to the House of Representatives holding hearings on federal nondiscrimination legislation that reiterated the messages that the Coalition delivered at the SACGHS hearings in 2003 and 2004:

> Fear of the misuse of genetic information has been demonstrated to cause many individuals to choose not to take advantage of genetic tests, information from which could be used to manage their health proactively. The potential for discrimination continues to grow as the number of tests available increases and electronic health information proliferates. ... Studies have shown that fear of discrimination also causes a large numbers of people to opt out of clinical trials. This lack of participation in research has a negative impact on researchers, clinicians, and industry, slowing the research and development process for targeted drugs and treatments. (Coalition for Genetic Fairness 2007)

One year later, in a revised policy statement on genetic discrimination published on its web site, the Genetic Alliance pointed out that everyone is susceptible to genetic discrimination because "each of us carries a number of mutated genes."[23]

A CREDIBILITY PROBLEM

By the second day of the hearings, the idea that genetic discrimination was a formidable barrier to progress in genomics appeared to be self-evident. But if genetic discrimination was truly blocking large numbers of Americans from seeking genetic testing and enrolling in genomic studies, where was the evidence? There were no empirical studies, beyond those published by members of the Genetic Screening Study Group in 1992 (Billings et al 1992) and 1996 (Geller et al 1996), indicating that Americans were being affected by unfair genetic decision-making in insurance and employment genetic discrimination (see also Reilly 1999). The small number of diagnostic and predispositional tests performed by laboratories each year in the country cast doubt on that claim (Reilly 1999).

Only three cases of workplace genetic discrimination have come to light since the Office of Technology Assessment Report published its 1983 report on workplace pre-employment genetic screening (U.S. Congress, Office of Technology Assessment 1983). The U.S. Equal Employment Opportunity Commission (EEOC) had litigated in the Burlington Northern Santa Fe Railway case (see Chapter 3). Another case occurred when employees at Lawrence Berkeley Laboratory accused their employer of conducting pre-employment screening for sickle cell trait, syphilis, and

pregnancy. The company settled out of court in 1999. A third case was that of Terri Seargent, who was diagnosed in 2000 with alpha-1 antitrypsin deficiency. She had been undergoing preventive treatment for the condition, which was covered by her employer-sponsored health insurance plan, and was fired when her employer received the first bill for her prophylactic treatment. Seargent, who was asymptomatic for the condition, subsequently lost her health, life and disability insurance. In this case, the EEOC agreed with her allegation of discrimination under the Americans with Disabilities Act (ADA), a law that protects Americans from workplace discrimination (and not discrimination by health insurers).

Moreover, by the summer of 2001, two years before the start of the SACGHS hearings in 2003, the Council for Responsible Genetics was reporting that forty-two states provided some protection against genetic discrimination in health insurance, and twenty-one states had passed laws prohibiting genetic discrimination by employers (Council for Responsible Genetics 2001). The fact that no lawsuits have been brought under these state laws also cast some doubt on claims that genetic discrimination was a significant problem. Moreover, the federal Health Insurance Portability and Accountability Act (HIPAA) of 1996 extended protection against genetic discrimination in health insurance to the majority of Americans who are insured in the group health insurance market. Thus, despite a decade of efforts by legislators, legal scholars, health and genetic activists, and the NHGRI to legitimize genetic discrimination as a significant and growing problem affecting Americans, by the start of the SACHGS hearings in June 2003, the merits of this claim were still in doubt.

Supporters of GINA liked to point to a 2004 NIH survey (Apse et al 2004) of 777 individuals with a family history of colorectal cancer as evidence that concern about genetic discrimination was a significant barrier to Americans interested in genetic testing. The survey, which was designed to measure awareness and concern about genetic discrimination for hypothetical genetic decision-making, found that 47% of the 470 respondents would ask for results from genetic tests to be left out of their medical records. The survey showed that 45% of respondents who rated their concern about genetic discrimination as high would be more likely to pay for tests themselves rather than submit their claims to insurers, or to use an alias for genetic testing. The survey also showed the 55% of respondents rated their concern about genetic discrimination as low or non-existent.[24]

These survey results are inconclusive. First, the sample size is too small to draw conclusions about most Americans. Secondly, the survey measured hypothetical decision-making, not actual decision-making. Third, the survey did not measure concern about genetic discrimination against other reasons for refusing testing, such as the absence of treatments or the desire not to know one's genetic status. Rayna Rapp's extensive ethnographic research on amniocentesis decision-making (for example, Rapp 1987, 1988, 1997, 1998, 1999) shows that genetic decision-making is extremely complex and shaped by many variables. Her work should put to rest the assumption that a survey tool can represent genetic decision-making.

Was fear of genetic discrimination stopping so many Americans from

seeking genetic testing? Were insurers and employers routinely using genetic information to make actuarial decisions about healthy Americans, and excluding them from benefits or charging them more than their peers? Elias Zerhouni and Francis Collins had charged the Committee with persuading Congress to pass GINA so that genomics research, with its set of "grand challenges," as Collins had put it, could move forward. In its first year of operations (2003), the Committee discussed what it should and could do. In its second year (2004), it turned its efforts towards documenting—and legitimizing—genetic discrimination as a significant civil rights problem. In its third year (2005), the Committee produced the ten-minute DVD video montage called "Voices of Discrimination," the compilation of all public comments on genetic discrimination called "Perspectives on Genetic Discrimination," and the legal analysis that argued existing state and federal protections from genetic discrimination were inadequate. In the remainder of this chapter, I will examine how the Committee arrived at its decision to solicit public testimony. In Chapter 8, I present and analyze the in-person testimonies.

The Need for Testimony

In fact, while the public heard from federal officials that genetic discrimination was a significant problem, members of the Committee and SACGHS staff had heard two very different messages. The source of these messages was the U.S. Chamber of Commerce, the most persistent and vocal opponent of federal nondiscrimination legislation.[25] One message was that there was no evidence that genetic discrimination existed. The

other was that current law provided adequate protection against genetic discrimination. Yet Committee members felt genetic discrimination was an issue that they should investigate. Although NIH scientists Elias Zerhouni and Francis Collins had instructed SACGHS that genetic discrimination was a priority, the Committee and staff were at liberty to decide how much time to allocate to discussions on genetic discrimination, and how best to use that time.

Some of this co-ordination happened on the frontstage (Goffman 1959) of the hearings. On the second day of the Committee's hearings (June 12, 2003), Paul Miller, Commissioner of the U.S. Equal Employment Opportunities Commission (EEOC), floated the idea that the Committee solicit stories about genetic discrimination from the public. As the federal agency charged with enforcing the 1990 Americans with Disabilities Act (ADA), the EEOC had become the de facto policing body for employer genetic discrimination. During this day's hearing, the inaugural SACGHS chair Ed McCabe asked Miller if his agency had documented instances of genetic discrimination beyond the three documented cases (Burlington Northern Railway, Lawrence Berkeley Laboratory, and Terri Seargent). In a long discussion that revealed how little the EEOC knew about the size of the problem or who it was affecting, Miller told the Committee that more evidence needed to be collected. He suggested that this might be an appropriate task for the Committee. Miller and McCabe tossed the idea back and forth:

> [Miller] ... there's really a dearth of information, of experiential information. There's not a whole lot of sort of cases coming forward

complaining of discrimination and everybody is sort of foreseeing this as a problem sort of around the corner. I wonder whether it might be an appropriate use of this time to sort of get to use the Committee and the various different points of view from the Committee to really get a sense of what is the concern out there, how deep does that concern go, how many people do feel that they're currently experiencing discrimination, to get a larger sense for the scope of the problem as it exists today because there's not a lot of information out there about that.

[McCabe] Now, these data are old. They're probably three plus years old and they were anecdotal.

[Miller] Right.

[McCabe] But when we approached the public, this was a major concern, and as has been mentioned, while the number of cases are relatively small and tended to be among the self-insurance where the employer is also the insurer and you're certainly aware of that since you've been involved in those.

[Miller] Right.

[McCabe] The public has extreme concern and is having the testing done anonymously or under pseudonyms because of their concerns and that creates certain problems as well.

This would be the kind of thing that we could certainly explore, though I wonder if perhaps you and your colleagues have already explored this and written about the cases that have come before the EEOC.

[Miller] ... There has been anecdotal evidence and stories and some of the consumer groups talk about it and there's a lot of anecdotal talk about the existence of discrimination, but there isn't really a lot of cases that have been coming forward in any sense and there's a great deal of confusion and question about why that is. (Comments by Paul Miller and Ed McCabe to SACGHS, June 12, 2003)

McCabe and Miller then brought up the idea of collecting stories of genetic discrimination experiences. McCabe suggested that it might be useful to recruit the Genetic Alliance in the effort because it had already undertaken something similar. Miller added, "Or the genetic counselor group or some of the other groups that are represented by your Committee." (Comments by Paul Miller and Ed McCabe to SACGHS, June 12, 2003)

But not everyone thought that genetic discrimination was a problem, or deserving of the Committee's time. Paul Zurawski of the U.S. Department of Labor told the Committee that in 2002, his department "had 184,000 consumer inquiries about their health plan and having almost none of those register as having a genetic information type of concern."[26] Others, such as Committee member Cynthia Berry, questioned whether collecting evidence of genetic discrimination was the best use of the Committee's time and if it had the resources for the task. Following Berry's comments, SACGHS Chair Ed McCabe and staff director Sarah Carr conferred on the Committee's purview and whether it could collect data. Carr confirmed that the Committee could "certainly consult with the public and ask for their input on issues." (Comments by Sarah Carr to SACGHS, June 12, 2003). Following this exchange, Alan Guttmacher, Deputy Director at NHGRI, chimed in with a comment that he was not aware of any surveys of genetic discrimination:

> [Guttmacher] If you're asking whether the current grants that are surveying cases of discrimination, none that I know of. I don't believe anybody's --
>
> [Carr] Did you ever do that in the past, Alan?

> [Guttmacher] I don't remember ever receiving any applications to do that. (Comments by Sarah Carr and Alan Guttmacher to SACGHS, June 12, 2003)

Later in the discussion, another Committee member, Debra Leonard, also questioned whether documenting cases of genetic discrimination would be a valuable use of the Committee's time, given that federal legislation was already under review. Miller's response to Leonard further suggested there was uncertainty at the Department of Labor about the scope of genetic discrimination:

> If I can just jump in, one of the concerns that's been raised around the bill is or one of the issues around the bill is that there really isn't evidence or data that discrimination is currently occurring, and so I think that one of the issues that's going to be thrown into the mix into the House as the House begins to consider whatever bill they begin to consider is, in a sense, is this timely? Is this a problem going on out there in the world?
>
> While there are some cases of great notoriety, including the one brought by my agency, there isn't sort of a landslide of these kinds of cases yet and that, I think, raises two questions. One is sort of to what extent beyond anecdotal stories here and there do you use to build a case or to understand the problem, and secondly, if there are a lot of anecdotal stories but nothing's turning into complaints in any formal kind of way, are there barriers out there that preclude complaints?
>
> Well, either it's because genetic information is so amorphous and it's all over the place, people don't know that that's the reason why they're not getting hired or not getting promoted. People aren't saying we're not hiring you because of your predisposition to cancer. People just don't know why and maybe that's a barrier or maybe there are all sorts of other barriers. People may feel that, gee, this is such private information to come forward and make a complaint, that's going to reveal information about me and my family that I simply don't want to put out there in the public realm. Maybe that's a barrier.

(Comments by Paul Miller to SACGHS, June 12, 2003)

If the head of the EEOC did not know how big the problem of genetic discrimination was or to what extent it shaped genetic testing refusal, and the NHGRI had not conducted any studies of its own, this was a good indication that no one in any federal agencies had a grasp on the problem. Following Miller's comment, Ed McCabe agreed with Miller that the Committee should document instances of discrimination to help to move the legislation forward. The Committee struck a Genetic Discrimination Task Force, with Agnes Masny (as chair) and Cynthia Berry.

Soliciting the Public's Testimony

As with all of the Committee's operations, even this seemingly straight-forward decision by the Committee to solicit public commentary had its backstage (Goffman 1959) elements. SACGHS staffer Amanda Sarata shed some light on how the Committee came to its decisions. Sarata, who worked for SACGHS from August 2003 to December 2005 through the NIH Office of Biotechnology Activities, managed the Task Force and co-ordinated all of the activities for the "Perspectives on Genetic Discrimination" session at the October 2004 SACGHS hearing. Sarata says that the job of deciding which issues the Committee focussed on often fell to Sarah Carr, who led the SACGHS staff team. Carr had tremendous latitude to prioritize the many concerns that Committee members identified, and the resources to translate ideas into action. But Carr also faced the difficult task of interpreting the concerns of Committee members fairly and ensuring that the process was transparent:

> Sometimes, after our meetings, the staff was left with the job of establishing a process that could be used to determine the course the Committee would take in fair, rigorous and transparent manner. Sarah [Carr] always wanted the Committee's input in any decision that was made and she always created mechanisms, such as task forces, to make sure the Committee guided all the decisions and all of the work. I do believe, though, that as staff, we had at least some part of the responsibility for "framing" the issues. I would say we had a role, deliberately or not, in informing where the Committee went. (Interview with Amanda Sarata, November 28, 2007)

According to Sarata, it was Chair Ed McCabe who pushed the Committee to solicit testimony from individuals who had encountered genetic discrimination. Sarata recalls McCabe telling the SACGHS staff, "It would be great if we could hear their stories." Sarata says that this was a good example of how a Committee member might make a comment or a suggestion, and the SACGHS staff would translate it into a concrete initiative.[27]

As the staffer responsible for the Perspectives on Discrimination session, Sarata drafted the short statement describing genetic discrimination in the call for public commentary (see Appendix F). She describes how challenging this task was:

> This was a very tricky issue. If you're soliciting public testimony about genetic discrimination, how do you frame it appropriately? What counts as discrimination? There were many examples that staff and the Task Force didn't necessarily consider to be discrimination. It was really, really difficult. The staff and Task Force were looking for cases of insurer and employer discrimination from predictive and susceptibility testing. (Interview with Amanda Sarata, November 28, 2007)

In developing the call, Sarata and the Task Force used the language and parameters of the federal nondiscrimination legislation in the House (GINA) as their guide. This legislation was narrow in scope. Consequently, the call for public testimony focussed on predictive and susceptibility testing, rather than a broader definition of genetic testing that would include prenatal diagnosis and newborn screening. The language of the call also had to be appropriate for the public, says Sarata. This meant they excluded such terms as "predisposition," "susceptibility," and "pre-existing condition" that are unfamiliar to most people.

The Task Force publicized its call for public testimony on genetic discrimination on August 2004, posting it directly on disease-specific list-serves and chat groups of organizations such as the Huntington's Disease Society of America. They also circulated it to organizations like the Tuberous Sclerosis Complex, which published the call in *TSC Alert*, its online newsletter. In all, twenty-six individuals, fourteen health-care providers, and twenty-one institutions and professional organizations submitted statements to the Task Force. Even though some cases did not fit the profile of genetic discrimination they had in mind, the Task Force included all of the written testimony that it received in the published compilation of testimony.[28]

Sarata and the Task Force then contacted a handful of people who submitted written testimony and invited them to deliver their statements to the Committee in person on October 18th, 2004. None declined. One of the seven individuals who testified, Rebecca Fisher, did not submit a written statement; it was Tim Leshan, chief of the Policy and Program

Analysis Branch at the NHGRI, who recommended Fisher to the Task Force.[29] Fisher was already known within the genetics community as an outspoken patient advocate on breast cancer, and an equally passionate advocate of federal nondiscrimination legislation.

CONCLUSION

By the start of the SACGHS hearings in June 2003, it was evident that genetic discrimination suffered from a credibility problem. Three notorious cases of workplace discrimination had been established as "genetic" discrimination, but most of the evidence for genetic discrimination, particularly with the insurance industry, was anecdotal. There was no empirical evidence that insurers or employers were systematically discriminating on the basis of genetic information. Yet comments made by NIH Director Elias Zerhouni and NHGRI Director Francis Collins in their formal presentations to the Committee on the opening day of SACGHS hearings indicate that these federal scientists considered passing a federal law banning the use of genetic information to be a priority for Congress. They also saw it as a priority concern for the Committee to address, emphasizing the importance of resolving this concern to keep genomics research moving. Committee members subsequently ranked the issue as a top priority in their own meetings.

The Committee decided, with some urging from EEOC Commissioner Paul Miller, to solicit testimony from the public to substantiate claims that individuals were being affected by genetic discrimination and that a federal law was needed. From the public commentary that the Committee

received, it selected six individuals to testify in-person to the Committee as members of the public and victims of discrimination. An NHGRI staffer put forward Rebecca Fisher's name to the Committee as a seventh. (I examine these public commentaries to SACGHS on genetic discrimination in Chapters 8 and 9.)

Thus, from 2003 to 2005, SACGHS took some remarkable steps to persuade Congress to pass federal nondiscrimination legislation. It solicited testimony from the public and devoted a half-day of its hearings to in-person testimony from victims of discrimination, healthcare providers, and organizations that have lobbied to see Congress pass federal legislation on genetic discrimination. It then produced lobbying tools based on the in-person and written testimonies (the "Voices of Discrimination" DVD, the compilation of written testimony), and a legal analysis that argued for new legislation. The Committee, whose members were forbidden by FACA rules to directly lobby politicians personally, managed to side-step this restriction, by encouraging participants to use these lobbying tools themselves.

In Chapter 5, I argued that activism on genetic discrimination had become institutionalized at the NIH in the early 1990s through, first its ELSI Working Groups, and then through the National Action Plan on Breast Cancer, a joint partnership between the NHGRI and the NBCC. The actions that the Committee members and SACGHS staff took to legitimize genetic discrimination and to produce lobbying tools suggest that activism on genetic discrimination continues to be highly institutionalized through the early twenty-first century (see Rabeharisoa 2008). Their actions also

show how individuals can shape the meaning of a social problem, by interpreting instructions to solicit public comments, by using language that narrowly frames a call for public comments on genetic discrimination in terms of problems with health insurance, and by selecting certain individual testimonies to present as the public "face" of experiences of genetic discrimination. In the next chapter, I look at the testimonies that the Committee chose to present as this face of genetic discrimination.

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[1] The Committee's most active deliberations on genetic discrimination spanned the period 2003-2005, but it continued to include updates on the progress of GINA at hearings after 2005. My SACGHS fieldwork began in 2005 and continued until the end of 2007.

[2] Testimony of NIH Director Elias A. Zerhouni to the Secretary's Advisory Committee on Genetics, Health, and Society. Inaugural Meeting, Wednesday June 11, 2003, Washington, DC.

[3] The last nondiscrimination bill to be introduced into Congress during SACGT's charter was S. 318/H.R. 602, a measure jointly sponsored by Senator Thomas Daschle and Representative Louise Slaughter on February 13, 2001. While S. 318 made it to hearings, H.R. 602, like many measures before it, stalled in the SubCommittee on Employer-Employee Relations. Source: THOMAS. Electronic document, http://thomas.loc.gov/cgi-bin/thomas, accessed October 3, 2006.

[4] See my discussion of these barriers, as identified by participants at the SACGHS hearings, in Chapters 6 and 7. To reiterate, judging by the attention that the Committee gave to these issues, some of the biggest barriers to integrating genomics into the health care system are the coverage and reimbursement of genetic tests, the validation and regulation of new genetic tests, and the education (or, genetic literacy) of clinicians and the public. Francis Collins has also identified these issues as barriers to delivering personalized medicine, both at the SACGHS hearings, and in his presentations to professional organizations. See, for example, Collins 2001 and Collins 2005.

[5] On October 14, the Senate passed S. 1053 by a vote of 95 to 0. H.R. 1910 had a less successful outcome. On May 27, 2003, the bill was referred to the House Energy and Commerce SubCommittee on Health, the House Education and the Workforce SubCommittee on Employer-Employee Relations, and the House Committee on Ways and Means. No further action was taken on the bill.

[6] This triumphant tone was also evident at a White House press conference on June 26, 2000 announcing the completion of a first draft of the human genome. At the press conference, President Clinton declared that the sequenced genome was "the most important, most wondrous map ever produced by humankind" and "the beginning of a new era of medicine." NHGRI Director Francis Collins, also at the press conference, described the work as "the revelation of the first draft of the human book of life" and "the first glimpse of our own instruction book, previously known only to God." With characteristic confidence, Collins proclaimed that "[r]esearchers in a few years will have trouble imagining how we studied human biology without the genome sequence in front of us." Source: "Remarks Made by the President, Prime Minister Tony Blair of England (via satellite), Dr. Francis Collins, Director of the National Human Genome Research Institute, and Dr. Craig Venter, President and Chief Scientific Officer, Celera Genomics Corporation, on the Completion of the First Survey of the Entire Human Genome Project." The White House Office of the Press Secretary, June 26, 2000. Electronic document, http://www.genome.gov/10001356, accessed October 8, 2006.

[7] FACA rules prohibit individual members of federal advisory Committees from lobbying politicians, and require all Committees to select members so that no single perspective dominates. See my discussion of FACA rules in Chapter 6.

[8] I discuss the origins of the term "sovereign consumer" and Sunder Rajan's use of this term in Chapter 6.

[9] U.S. Census data for 2003 strongly correlates household Internet access with age, educational attainment, and household income, with wide disparities in access along these variables (Day et al 2005). Household internet access is highest amongst those ages 35-44 years (65.3%), those with a Bachelor's (76.8%) or Advanced degree (81.1%), and families with an income of $100,000 or more (92.2%). Household Internet access is lowest amongst those 65 years and older (24.4%), high-school dropouts (20.2%), and families with an income of less than $25,000 (30%).

[10] Affymetrix is a leading manufacturer of DNA microarrays. Molecular biologists routinely use DNA microarrays to examine a whole genome at once and observe which genes are active and inactive at any given time.

[11] Presentation by Francis Collins to SACGHS, June 11, 2003.

[12] Mutations are rare genetic variants whose effects are deleterious. The diseases they cause are often lethal to infants. Their distribution is limited to certain populations, which is why there are only thousands of people in the world with any one single-gene disorder. Susceptibility genes are widely-distributed genes that are linked to common (not rare) diseases, such as breast and colon cancer. The diseases they are linked with usually develop in adults, not in children. Susceptibility genes do not directly cause disease. They only confer an increased risk of disease—and then, only in conjunction with

environmental factors that are yet to be identified. (Margaret Lock, personal communication)

[13] Khoury may be the only federal scientist to publicly challenge the NHGRI's plans for a separate large-scale prospective cohort study (see Khoury 2004). Khoury's opposition to the large-scale cohort study hints at competing visions of the place of genomics in public health and clinical medicine in the United States. As the director of the CDC's National Office of Public Health Genomics, Khoury has advocated bridging the gap between medicine and public health by adopting population approaches to genomics, focussing on prevention (rather than individualized drug treatments), and re-engineering the health care system to accommodate genomics discoveries (see Khoury et al 2007). Collins, as NHGRI director, has promoted a vision in which genomics discoveries and improved sequencing technologies drive the delivery of personalized medicine for the individual.

[14] See my discussion of the rationales for funding the HGP in Chapter 6.

[15] Source: "Future Directions in Genetic and Genomic Research." PowerPoint presentation by Francis Collins to SACGHS, June 11, 2003, Washington, DC." Electronic document, http://www4.od.nih.gov/oba/SACGHS/meetings/June2003/Presentations/Collins_s.pdf, accessed October 30, 2008.

[16] Written testimony of Fran Visco, President, National Breast Cancer Coalition, to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[17] Written testimony of Fran Visco, President, National Breast Cancer Coalition, to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[18] Written testimony of Sharon F. Terry, Genetic Alliance, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[19] Written testimony of Sharon F. Terry to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[20] Source: "Preventing Genetic Discrimination in Health Insurance." Statement of Francis S. Collins to Congressional Task Force on Health Records and Genetic Privacy, July 22, 1997. Electronic document, http://www.genome.gov/10002352, accessed August 16, 2008.

[21] Source: Comments by Francis S. Collins to the Senate on the Passage of Genetic Information Nondiscrimination Act of 2005 (S. 306), February 17, 2005. Electronic document, http://www.genome.gov/13014311, accessed August 16, 2008.

[22] Source: "Protecting Workers from Genetic Discrimination." Testimony of Karen H. Rothenberg before the House Committee on Education and Labor SubCommittee on Health, Employment, Labor, and Pensions. January 30, 2007. Electronic document, http://www.geneticalliance.org/ksc_assets/publicpolicy/hr493hearingrothenbergtestimony.pdf, accessed August 16, 2008.

[23] Source: "Genetic Discrimination." Genetic Alliance Policy Statement, 2008. Electronic document, http://www.geneticalliance.org/policy.discrimination, accessed August 16, 2008.

[24] See Peterson et al (2002) for a study of how fear of genetic discrimination affects actual (not hypothetical) decision-making for genetic testing. The authors studied a cohort of 384 patients who attended a clinic for BRCA1 and BRCA2 testing. They found that approximately 25% of patients declined testing because of their combined concerns about the cost of tests, confidentiality, and insurer discrimination. The authors were unable to document any cases of insurer discrimination based on test results, and concluded that there was a discrepancy between patients' fears of insurer discrimination, and patients' actual experiences with insurers after testing.

[25] Interview with Amanda Sarata, November 28, 2007. Other organizations that opposed GINA were America's Health Insurance Providers (AHIP), the National Association of Manufacturers, the Society for Human Resource Management, and a coalition of trade organizations called the Genetic Information Nondiscrimination in Employment (GINE) Coalition.

[26] Zurawski was commenting on the perception that health insurers were violating the 1996 federal Health Insurance Portability and Accountability Act (HIPAA). This Act forbids insurers from using genetic information to set premiums for individual subscribers in a group plan. See Chapters 3 and 5 for a discussion of HIPAA.

[27] Interview with Amanda Sarata, November 28, 2007.

[28] Interview with Amanda Sarata, November 28, 2007. Federal Advisory Committee Act (FACA) rules governing the conduct of SACGHS required staff to publish all public commentary they received.

[29] Interview with Amanda Sarata, November 28, 2007. The Policy and Program Analysis Branch, now under the direction of Phyllis Frosst, develops policy positions for the NHGRI on genetic discrimination, but also on personalized medicine, pharmacogenomics, coverage and reimbursement of genetic tests, and human subjects research.

INTRODUCTION

On the morning of Monday October 18, 2004, 70 people gathered in the small Congressional Ballroom at the Bethesda, Maryland Marriott for a session unlike any other the Committee had held, called "Perspectives on Genetic Discrimination." Those gathered included members of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS), representatives of federal departments and agencies, and the public. They listened intently as Agnes Masny, chair of the SACGHS Task Force on Genetic Discrimination, introduced the seven Americans who had travelled to Bethesda to testify to the Committee about their experiences of genetic discrimination. The six women and one man, who formed a panel called "Members of the Public," sat side by side at a long table across from Committee members and the webcam. Place names identified each. Behind them, members of the public and invited speakers sat in four rows. Heidi Williams, a Kentucky resident and one of the seven on the panel, spoke first.

"My name is Heidi Williams and my children, Jayme, 8, and Jesse, 10, were recently victims of genetic discrimination," is how she began her story. With her sons Jayme and Jesse sitting behind her, the younger one twisting around his mother to peer into the webcam broadcasting her testimony, Williams told the Committee how she had tried to apply for health care insurance for her children, who are carriers of the genetic disorder alpha-1 antitrypsin deficiency, with the insurer Humana. Despite her providing the insurer with evidence from the National Institutes of Health (NIH) and the Alpha-1 Foundation that carrier status would not

cause symptoms of the progressive liver disorder, Humana denied Williams's application and rejected her appeal.

Williams described in vivid terms the perils for a family of sharing its medical history and genetic diagnoses with a health insurer:

> Today, there is a current of fear reverberating throughout the genetic community. It is not just a fear of loss, but it is a fear of retribution. It is a fear that forces many within this particular community to accept what should be unacceptable: discrimination by genetic status. Many people are afraid to come forward and fight for their rights to employment and health insurance coverage because they are afraid of the retribution that may not only be taken against them, but could be taken against their families, as well. (Testimony of Heidi Williams to SACGHS, October 18, 2004).

These seven in-person testimonies were only a portion of the stories that the Committee collected for the "Perspectives on Genetic Discrimination" session. But their effects were potent, and they were the dramatic focal point of the entire hearings. In all, the Committee collected written and in-person testimony from twenty-six individuals, fourteen health-care providers, and twenty-one institutions and professional organizations attesting that genetic discrimination was a serious problem facing the country.

The collected testimony accomplished several things within the setting of the hearings. It legitimized genetic discrimination as the most pressing civil rights concern affecting Americans today (see Collins and Watson 2003). It also crystallized a specific subject that had already made discursive appearances at the hearings: the vulnerable American as a middle-class consumer seeking genetic self-knowledge and practicing self-

surveillance. As testimony from victims of discrimination made clear, this vulnerable American was a moral subject who expected—and was expected—to use genetic testing in the care of the self (cf. Foucault 1978). This construction of Americans as potential victims of genetic discrimination displaced other potential constitutions of American vulnerability at the hearings, such as the disabled, the poor, the estimated 46 million (or 16%) uninsured Americans (U.S. Department of Health and Human Services 2005), and an estimated 56 million "medically underserved" Americans who live in regions of the country with no health care providers (National Association of Community Health Centers and The Robert Graham Center 2007).[1]

In this chapter, I present the in-person testimonies from these seven individuals and identify key themes in their stories. I argue that in presenting these testimonies, SACGHS did not simply legitimize genetic discrimination as a real problem affecting Americans. It succeeded in framing it as the most significant civil rights problem in the United States since segregation.

VOICES OF DISCRIMINATION

The October 18, 2004 session that SACGHS called "Perspectives on Genetic Discrimination" consisted of three panels. The first was a panel of seven "members of the public" testifying about their experiences of genetic discrimination. This was followed by a "provider panel" that consisted of three healthcare providers, one of whom was an NHGRI researcher. The third panel comprised four "additional stakeholders." These included Kathy Hudson of the Center for Genetics and Public Policy and Joanne

Armstrong, representing America's Health Insurance Providers (AHIP), the chief lobbying organization for the country's 1,100 private health insurance companies.

The seven members of the public who travelled to Bethesda that day represented two distinct categories of genetic disorders. Three of the seven who testified to the Committee (Heidi Williams, Phaedra Malatek, and Phil Hardt) have family histories of single-gene disorders (alpha-1 antitrypsin deficiency, hemochromatosis, hemophilia B, and Huntington's disease). The other four (Rebecca Fisher, Tonia Phillips, Paula Funk, and Maria Carolina Hinestrosa) have all tested positive for mutations in the tumour-suppression genes BRCA1 or BRCA2, which heighten risk of breast and ovarian cancer.

The testimonies were complex in two other ways. The tacit understanding of genetic discrimination that was in use at the hearings corresponded to the 1992 American Journal of Human Genetics definitive article (Billings et al 1992), as the treatment of healthy, *asymptomatic* individuals with a genetic diagnosis as ill by insurers or employers. Yet only three of the seven who testified (Phillips, Malatek, and Funk) to the Committee were asymptomatic for their conditions. Fisher and Hinestrosa are breast cancer survivors, Hardt has active hemophilia but is symptomatic for Huntington's, and Williams is symptomatic for alpha-1-antitripsin deficiency. (Williams's children are asymptomatic carriers of the allele).

Moreover, Agnes Masny, the chair of the SACGHS Task Force on Genetic Discrimination, did not distinguish between diagnostic and

confirmatory testing for rare, single-gene disorders, and susceptibility testing for more common, multi-factorial disorders, such as breast cancer. Nor did any of the Committee members.[2] This is critical in listening to the testimony of the seven, because one of their messages is that genetic testing has strong predictive power.

The meaning of "predictive" must be queried here. Diagnostic and confirmatory testing for single-gene disorders, particularly for autosomal recessive disorders that have high gene penetrance, such as cystic fibrosis, sickle cell anemia, Tay Sachs disease, and AAT, has a high predictive value. Tests for these disorders can predict whether an individual will develop that disease—but not at what age, with what severity, or how the disease will progress. These tests deliver precise values that indicate whether or not the patient will develop the disease, based on Mendelian inheritance rules of single-gene disorders. At the opposite end of the predictive spectrum lies susceptibility testing for common gene-linked disorders that have lower penetrance, such as breast, ovarian and colon cancer, and diabetes.[3] Susceptibility testing for these disorders produces probability ranges that express a heightened risk for developing the disease. But susceptibility genes only account for a small percentage of the cases of disease. For example, the BRCA1 and BRCA2 mutations associated with an increased risk of breast cancer only account for between 5% and 10% of all cases of breast cancer. This means that between 90% and 95% of all cases of breast cancer are not explained by inherited mutations in the BRCA1 or BRCA2 genes.[4] In other words, susceptibility testing for common disorders is limited in its predictive power and the results are characterized by tremendous uncertainty (Lock 2005a).

To speak of the "predictive" power of susceptibility testing, therefore, is clinically inaccurate and misleading. However, it is consistent with how some scientists, the mass media, and the NHGRI have portrayed genomic medicine in a promissory light, with the hopeful expectation that researchers will refine these probabilities and permit more precise clinical interpretation and decision-making. It is also, as these testimonies make clear, consistent with public understandings of susceptibility genetics and inheritance. In these testimonies, the assumption that even genetic testing for low-penetrance susceptibility genes confers some certainty about the future hints at the valorization of scientific and medical knowledge on the one hand, and a desire for technological interventions that offer some semblance of individual control over complex and fundamentally unknowable events.

I present the testimonies in a different order from the hearings, grouping the stories by the type of disorder. I first present testimonies from the three individuals with single-gene disorders (Heidi Williams, Phaedra Malatek, and Phil Hardt). I then follow with the testimonies from the four women diagnosed with mutations in BRCA1 and BRCA2 genes, some of whom developed breast cancer (Paula Funk, Tonia Phillips, Maria Carolina Hinestrosa, and Rebecca Miller). From each person's testimony, I draw out one or more themes that were common to many of the testimonies.

Heidi Williams: The Uncertain Futures of Our Children

Heidi Williams, whose two children are asymptomatic carriers of alpha-1 antitrypsin deficiency, is herself symptomatic for the disease. The

disease is an inherited disorder that affects approximately 1 in 2,500 Americans, and is characterized by the production of low levels of the liver protein alpha-1 antitrypsin. This can cause lung disease in adults, and lung and liver disease in adults and children. Like cystic fibrosis, sickle cell anemia, and Tay Sachs disease, AAT deficiency is an autosomal recessive disorder, meaning that a child must inherit two copies of a mutant allele (one from each parent) to develop the disease.

The story that Heidi Williams told the Committee underlined the point that members of Genetic Screening Study Group had made a decade earlier (Billings et al 1992). Insurers, in particular, treat individuals with rare disorders as seriously ill, even if they are asymptomatic carriers. Listening to Williams's story, one of the messages seemed to be that intake staff at the call centres of insurance companies are ill-equipped to interpret family medical history and to understand the significance of carrier status for rare disorders.

Williams told the Committee that after watching a television commercial in August 2003 for affordable health insurance offered by the health insurer Humana, she called the company and spoke with a woman who told her the monthly cost to insure both her children would be $105. Williams decided then and there to go ahead with the application to start coverage as soon as possible. The Humana employee then began asking Williams whether her children had a pre-existing condition. Under "a threat of a fine and incarceration for falsifying information," Williams told the employee that her children were carriers of AAT and that they would never experience any health problems from their carrier status, unlike

Williams herself. This information set off a series of exchanges between Williams and the intake staff at Humana:

> The young woman, who wasn't quite sure what to do with this information, asked me to hold on the line while she contacted her supervisor. As I spoke with her supervisor, I again explained how my children were only carriers of the AAT gene and that my children themselves would never suffer from any aspect of the disorder as I am suffering, and that they are exceptionally healthy and active children. Again, I was told to hold the line because, as this gentleman was uncomfortable with the information I had imparted, he needed to contact his supervisor. As I spoke to the senior supervisor, I once again relayed the information about alpha-1 and how my children were only carriers. (Testimony of Heidi Williams to SACGHS, October 18, 2004)

In the end, the company finalized Williams's application for health insurance for her children, and told her it would confirm the application within 24 hours. "After five days of waiting," Williams said, "I knew instinctively that there had been a problem with my children's application." In a letter that she received from Humana two days later, Williams read that the insurer had rejected her application for her children because of their AAT deficiency carrier status. Williams enlisted the support of the Alpha-1 Lungs and Life Chat Group, the genetic advocacy umbrella group the Genetic Alliance, and a Washington, DC law firm to fight Humana to change its decision. They sent a letter to Humana with information from the NIH and the Alpha-1 Foundation explaining the carrier status of AAT. Despite their efforts, Humana declined her children coverage on the basis of their carrier status. After media attention to Humana's decision, the insurer reversed its decision and offered full coverage to Williams' children.[5]

What was at stake for Williams, along with others testifying that day, was her hope that her children would be able to realize their dreams. Despite having only carrier status for ATT, her children were at risk, she told those listening:

> As they get older and they choose their careers, my daughter is a competitive gymnast. She's 8 years old. She wants to grow up and be a gymnast. But there is a chance that she could be discriminated against because she is a carrier of alpha-1. My son, he wants to be a research scientist. He wants to build habitats on the moon. He's 10 years old. There's a chance that when he gets to that point, he may not be hired in his chosen field. He may have to, God forbid, flip burgers at McDonald's. (Testimony by Heidi Williams to SACGHS, October 18, 2004)

Williams' worries about her children's futures hinted at the tremendous ongoing power of genetic diagnosis in the public imagination (and amongst insurers) to define individuals and their potential (see Nelkin and Lindee 1995). "We are all viable members of a community with contributions to make and shouldn't have to be afraid that our genetic anomalies, in whatever form they arise, will be held against us," Williams told the Committee:

> I should not have had to spend the better part of six months wondering if the decision to have my children's genetic status verified by their pediatrician was a huge mistake. I should not have to wonder if my children's genetic status is going to follow them into the workforce and render them unable to become employed in their chosen fields. And I certainly should not have to feel guilty for unknowingly passing this genetic anomaly on to my children. (Testimony of Heidi Williams to SACGHS, October 18, 2004)

Williams closed her testimony with a passionate appeal to the

Committee to help pass federal nondiscrimination legislation. As she did, she described genetic discrimination in dire terms. “As each day passes and the genetic community waits for the House to bring this bill to a vote, scores of people across this nation are being persecuted on the basis of their genetic status,” she told the Committee. “It is completely reprehensible that any type of discrimination still exists and has to be legislated against in this day and age. But since discrimination still exists, it must be swiftly eradicated in any form that it is found before its destructive force has had the chance to harm anyone else.”

Phaedra Malatek: Genetic Testing is the Weather Tracking Device of Health

Weather forecasting was the metaphor that Phaedra Malatek used to describe the utility of genetic testing. Malatek, who teaches at two community colleges in Illinois, has a family history of hemochromatosis, a common inherited metabolic disorder that causes the body to absorb too much iron. The condition, which affects an estimated 5 in 1,000 Americans, is inherited in an autosomal recessive pattern with a defective HFE gene. It is easily treated, but if left untreated, it can lead to organ damage. Malatek’s father was diagnosed in 1991 and died eleven years later. His diagnosis, and the subsequent diagnosis of Malatek’s two siblings, alerted her to the possibility that she and her two sons were at risk for developing the disorder.

Malatek told the Committee that it was unfair for insurers to punish individuals who used genetic testing to avert disease, when their actions reduced the potential costs and damage from illness:

> For me, genetic testing and the protection offered by S. 1053 can be compared in an analogy to weather tracking or storm prediction. Imagine, if you will, that we had no knowledge of the storms that recently swept through the southeastern United States and the Caribbean. How would the death toll change? How would the damage assessment change? How would the insurance industry have changed? Now imagine never having any information about any storm ever. Well, I think our understanding and consideration of genetic testing can be compared to those "what ifs."
>
> What if people were given the knowledge of the potential storms in their lives? How would they be able to protect themselves? What would serve as the plywood for the windows, and what evacuation routes would be made available to them? More importantly, how many lives would be saved? Because that really is the question, isn't it? How many lives can we save by what we're doing here and through the enactment of S. 1053? (Testimony by Phaedra Malatek to SACGHS, October 18, 2004)

The advance warning that genetic testing provided, said Malatek, allowed individuals to practice better self-surveillance. “Those who are informed about their risk can be proactive and take either prophylactic measures or be monitored more closely, increasing their ability to entirely avoid developing a disease or having it detected in its earliest, most treatable and survivable stages,” she said. Not only do these measure save lives, she testified, but they save money for everyone. (Testimony by Phaedra Malatek to SACGHS, October 18, 2004)

Malatek also invoked the struggles of civil rights activists for equal treatment of African-Americans in her testimony:

> All of this is reminiscent of a series of choices that were being made 40 years ago. In the late '50s and early '60s, my parents fought diligently for the rights of people who were genetically different from them. They were not different in that they were at higher risk for obtaining hemochromatosis or ovarian cancer but that their skin was

> a different shade of beautiful. My parents, along with many others, won that fight. The Civil Rights Act amendments are there to protect people from discrimination based on genetic makeup that we can see, be it skin tone, gender, or disability. A person's genetic makeup that isn't visible should be equally protected under the same terms and can be through S. 1053.
>
> It's remarkable for me to realize that the work my parents did for the Civil Rights Act in the '60s was not complete. Here I am, 40 years later, working on the same issue, equal rights and protection under the law no matter the genetic makeup of a person. The fact that we can look inside the DNA of a person to know more about them should not preclude them from the protection that was fought for so valiantly. As I see it, genetic testing is the weather tracking device of health. Just as we rely on weather tracking technologies to predict and to allow us to protect ourselves from hurricanes or other weather-related storms, I urge you to allow us to do the same for genetic diseases.
>
> We must move forward in protecting people from the potential storms in their lives. (Testimony by Phaedra Malatek to SACGHS, October 18, 2004)

Phil Hardt: Choosing the Safety of Anonymous Testing

Phil Hardt inherited two diseases from his parents: hemophilia B from his mother, and Huntington's disease from his father. His two biological daughters and his granddaughters are hemophilia B carriers. As well, all of his three biological children were considered to be at risk for Huntington's disease until they underwent genetic testing and learned that they do not carry the allele. Hardt told the Committee that “our story is one of continuing genetic discrimination even though we have laws that are supposed to protect me, my children, and my grandchildren.” (Testimony by Phil Hardt to SACGHS, October 18, 2004)

Hardt was warned by the human resources manager at his workplace

never to tell his boss about his hemophilia because it would jeopardize his training and promotion opportunities. “Consequently,” said Hardt, “all future bleeding episodes had to be hidden from him.” This was just the tip of the iceberg. His daughter and grandson experienced difficulty obtaining insurance because of their family history. For example, when his oldest daughter applied for mortgage life insurance in 2000, her application was rejected by “every major insurance company.” “Each of her rejection letters state two pertinent facts that are important,” Hardt told the Committee. “Number one, they each state that they will not insure her until she has tested for Huntington's disease, and two, that she is found to be negative.” But this was not all. On one of the letters, the insurance agent wrote a note telling his daughter and her husband that the company would insure their children only when she learned her HD status. This indicates, said Hardt, “that the discrimination is down to the third generation now.”

Hardt’s children resorted to anonymous testing for Huntington’s disease, and Hardt established an anonymous testing service to help others who do not want their genetic information made available to others. However, says Hardt, this is not an ideal solution. For one, he told the Committee, it is very costly. “It's around $900 out of pocket to find out,” he said. But it also means having to hide information from medical providers. Hardt told the Committee that “it's a shame that we have to do this.”

Paula Funk: The Family Burden of Genetic Testing

One of the persistent images of these testimonies was the vulnerability of the entire extended family: siblings, parents, aunts, uncles, and cousins.

This family vulnerability had two facets. When one member of a family receives a positive genetic diagnosis, members of the extended family must contend with the possibility that they, too, may have inherited an allele for a disorder. But family members also face uncertainty and anxiety about whether to undergo genetic testing, be it for presymptomatic testing, diagnostic testing, or susceptibility testing.

None of the seven who testified to the Committee depicted this as uncertainty and anxiety arising from a lack of treatment options. Rather, they related their feelings to their concerns about how insurers might use their test results. Some, such as Paula Funk, identified members of their extended families as current and potential victims of genetic discrimination. With great dignity, Funk described a family that had been already devastated by the inheritance of alleles for breast and ovarian cancer and faced the further burden of weighing the risk of discrimination:

> My dad is one of ten children, and he has five sisters. All five of his sisters have had breast cancer, and the current count right now is that eight of my cousins have had breast cancer as well. The number breakdown there, that is 13 women out of 24 that have had breast cancer. This disease is something that the women of my family have to constantly think about. There's a constant threat. (Testimony by Paula Funk to SACGHS, October 18, 2004)

Describing how her "sweet aunt Dorothy" had twice survived breast cancer and was currently fighting "aggressive form of ovarian" cancer, Funk told the Committee that her aunt's experience made her feel "really sad. It makes me feel like I have to aggressively fight my possibility of cancer."

But it was not only cancer that had reached deep into their lives. Funk

described how her family had been held hostage to their fears that health insurers might make prejudicial decisions if any of them sought genetic testing or had a positive diagnosis. Funk told the Committee that she had made the decision ten years earlier to undergo genetic testing. This required that she approach several of her female relatives to donate blood samples to determine if there was a mutation. But Funk did not succeed in her request to her family. "I approached my aunts and my cousins about this, and they talked to their physicians, and their physicians recommended that they not pursue genetic testing because at the time people could deny insurance and the discrimination could be even worse in the future as more was learned about being genetically positive," she said.

Funk, who was 23 years old at the time, decided to postpone genetic testing. Now 33 years old, she is at the average age when women in her family begin developing breast cancer. In May 2003, she reconsidered undergoing genetic testing and spoke with a genetic counselor, who asked Funk "a lot of questions about insurance." She and her husband, who had just opened their own business and did not have health insurance, were accepted by United HealthCare as a two-person group. This group health insurance coverage meant that Funk would not undergo medical underwriting, and would have her genetic testing covered without prejudice. "I'm thankful that they accepted me as a small group," Funk told the Committee, "but I live with the fear every day that I could be rejected."

Funk described genetic testing as an invaluable, life-saving tool—but

one that called for the sacrifice of body parts. “I'm so grateful that I have an opportunity to save my own life, though,” she said. “I hope to have a prophylactic mastectomy this fall, and I've been told that it gives me a 95 percent chance that I will never have breast cancer. After I'm finished having children, I plan on having my ovaries removed as well.” These surgeries would offer Funk “a 95 percent chance” that she would never develop ovarian cancer, either. (Testimony by Paula Funk to SACGHS, October 18, 2004).

The cost of genetic discrimination, according to Funk, was lives lost to indecision about undergoing genetic testing:

> We put off being tested for 10 years because of what the physicians recommended because of the potential discrimination. Countless women in my family during the last 10 years have been diagnosed with breast cancer, and several of them have lost their battle to breast cancer. That could have all been prevented if we had pursued testing then. (Testimony by Paula Funk to SACGHS, October 18, 2004)

And while a shared allele reaches into the extended family, so too does the fear of discrimination, Funk told the Committee. “That really grieves me thinking about the loss of life there that could have been avoided,” she said. “I've decided that knowledge about my health is a gift. I want everyone to feel the freedom to have that gift.”

Funk closed her testimony by reminding listeners that once the decision is made to reveal one’s own genotype through genetic testing, neither that decision nor the knowledge it brings can be undone. Here lies the vulnerability of those who have committed to knowing their genotypes. “Finding out your genetic status is permanent,” she told the Committee.

"You can't take it back, and it isn't something that you can change your mind on." She implored those listening to pass protective legislation. "What I really need, and what we all need, is a law that clearly defines the safety and the fact that you cannot be discriminated against genetically."

Tonia Phillips: Being Penalized for Practicing Prevention and Self-Care

Tonia Phillips, who was diagnosed with the BRCA1 mutation in 2003, told the Committee that Americans who work in small companies are particularly vulnerable to the economic consequences of a positive genetic diagnosis. The diagnoses and illnesses of employees in small organizations are more difficult to hide. When health insurers increase annual premiums for a group because one member of that group undergoes expensive treatments, these increases cannot be spread out amongst a large pool of workers in a small company.

Phillips works in a "small company of about four people, including my two bosses, the owners." She describes them as "a tight-knit family" who were with her when her mother died of ovarian cancer in 2002 and when she underwent her own testing for the BRCA mutations. "I was very open with my experience just because we are a small company and there was no way to hide it," she said. In March 2003, she learned she had a positive diagnosis for the BRCA1 mutation, which translated into an "80 percent lifetime chance of getting ovarian cancer and a 45 percent lifetime chance of getting breast cancer." She made the decision to undergo a prophylactic hysterectomy in October that year. She followed with a prophylactic mastectomy in March 2004, and then reconstructive breast surgery, which she was still undergoing by the time of her testimony to the Committee.

In July 2004, four months before this testimony, her company's group health insurance bill had increased by $13,000 a year for four people. Phillips learned about the company's increased premium immediately from her boss, who asked her to switch to her husband's health insurance policy so the other employees would not have to shoulder the increase. Phillips told her boss that she was reluctant to change insurers because she was in the process of reconstructive surgery. "It was like pulling teeth to get the insurance company to pay for these procedures, and switching would confuse and complicate everything," she told the Committee. "I didn't think it was in my best interest to switch while I still needed more surgery." They arrived at a compromise, whereby the other employees would pay half of the premium. This satisfied Phillips, although, as she acknowledged, "I'm sure the other employees weren't too happy with me."

The injustice for Phillips was that she had made the difficult decision to undergo prophylactic surgery to prevent the onset of a serious and costly disease. She felt that she had been penalized by the insurer and her employer for taking extensive measures to prevent disease from occurring:

> It seems unfair to me that I am taking steps to keep myself healthy and to prevent cancer in the future, and I am being singled out and made to feel I am a liability. I also don't smoke, I work out, I eat right most of the time. If someone in the company were diagnosed with cancer or some other disease, they would not have been asked to switch insurance companies as I was asked. I hope that me coming here and telling my story will help with defining the problem and passing laws against genetic discrimination of any kind. (Testimony of Tonia Phillips to SACGHS, October 18, 2004).

Phillips's story thus reminded listeners that one of the moral bases for a

claim to genetic discrimination is being a prudential subject (O'Malley 1996) and practicing self-care. In a nation where Myriad Genetics aggressively promotes BRCA testing to consumers and clinicians, practicing genetic prudence (Rose and Novas 2005) means using susceptibility testing to predict future risk of disease and making lifestyle changes to stave off disease.

Maria Carolina Hinestrosa: Fear Impedes Testing and Research

Maria Carolina Hinestrosa's testimony was unusual, in that she appeared to the Committee as both a "two-time breast cancer survivor...a mother of a 13-year-old daughter," and as an executive officer of the National Breast Cancer Coalition (NBCC), the main breast cancer lobbying force in the country. Like others sitting at the table with her that day, she opened her story with her personal and family histories of diagnosis:

> My first diagnosis with breast cancer was at the age of 35. My second diagnosis was at the age of 40. My younger sister was also diagnosed twice, first at age 29, and then at 34. Over Christmas last year, two of my cousins and an aunt were diagnosed with breast cancer as well. Of course, we suspect there is a genetic mutation that predisposes members of my family to breast cancer. (Testimony of Maria Carolina Hinestrosa to SACGHS, October 18, 2004).

Like members of Paula Funk's family, who feared for their health insurance coverage if they underwent genetic testing for a family history of breast cancer, and like Heidi Williams, who feared her children's carrier status might follow them into the workforce, Hinestrosa worried that she might jeopardize daughter's future if she chose to be tested. "I sought genetic counseling as part of a study," she told the audience. "After

carefully weighing the potential benefits and harms of genetic testing, I decided not to undergo testing for fear of potential consequences to my daughter."

Hinestrosa outlined two fears. One was that "the information may not be protected and might even be misused." The other was that if she tested positive, her "daughter might be obligated to disclose the presence of a genetic mutation and that she might suffer future discrimination in health insurance and employment as a consequence." Yet Hinestrosa was not the only family member who held these fears. "I have four sisters and a brother," she said. "We all worry about our risk for breast cancer and the potential risk for our daughters, yet none of us feel safe enough to undergo genetic testing. My family experience illustrates why our nation needs strong nondiscrimination laws."

Despite the promises of the HGP, said Hinestrosa, fear of discrimination was a liability for all women with a family history of cancer. "The mapping of the human genome has brought with it the promise of reducing human suffering by targeting interventions for those at risk for disease," she told the Committee. But "[f]ear of potential discrimination threatens both a woman's decision to use new genetic technologies and to seek the best medical care. Women are also afraid to enroll in research and clinical trials that involve genetic studies, and this in turn threatens the viability of the scientific community to conduct the research necessary to understand the cause and find a cure for breast cancer. Many of the women testifying at present in this audience today have experienced exactly those concerns." Genetic discrimination, Hinestrosa told the

Committee, "is a real and growing problem that needs an immediate solution, not one that should wait until we have further cases of women and men who have experienced this type of discrimination that is so detrimental to the ability to seek quality health care."

Rebecca Fisher: Refusing to be Second-Class Citizens

Rebecca Fisher, a medical librarian from Virginia, was diagnosed with breast cancer and underwent treatment while working for "a small community hospital in south Florida." The company was self-insured, and a third party managed its health insurance plan. One year after she had finished her treatment, Fisher told the Committee, she received a phone call from a "flustered young woman" who had been trying to locate Fisher. The woman calling, who did not realize that she was talking to Fisher, told Fisher that Fisher's "bone marrow transplant and other health care costs exceeded the calendar year cap last year." She wanted to know if Fisher's treatment costs would again exceed the cap. Fisher told the Committee how she responded: "'I'm Rebecca Fisher, I said, and I really hope not.'" (Testimony by Rebecca Fisher to SACGHS, October 18, 2004)

Fisher's anger was obvious as she told the Committee that it was clear what mattered to the company that she worked for. "This experience taught me something," she pointedly told the Committee. "It taught me that there are people who are paid to look at me and see not my ability to contribute to a community, not my honesty, my integrity and my faith, not my education, hard work, and social conscience, not my family members and the ways in which I have helped each of them succeed, but dollar signs, costs, increased liability, and the odds of my dying an expensive

death."

Responding to speculation by Nobel laureate Sidney Brenner that fifty years' time might bring a public health transformation in which "those who have a genetic predisposition to disease will learn how to take extra care," Fisher told those assembled that that day had already come. "Dr. Brenner needn't wait 50 more years to see this prediction realized," said Fisher. "Some of us, those who possess BRCA1 or 2 mutations, known to predispose us to breast and ovarian cancer, are already taking extra care." Women with a family history of breast and ovarian cancer and who are, in addition, diagnosed with either the BRCA1 or BRCA2 mutant alleles, already shoulder the burden of "taking extra care," Fisher told the Committee. They should not be punished or treated as an extra actuarial risk by their insurers and employers for taking the necessary steps to prevent illness. Fisher elaborated:

> A recent study established that 67 percent of women with this mutation are diagnosed with breast cancer by the time they're 50 years old. But I have a cousin who died of it when she was 28. I have another who is battling Stage 4 ovarian cancer as we sit here today. She has a 4-year-old. My mother had breast cancer at 35. Her mother died of ovarian cancer at 41. Her sister had breast cancer at 32. I was 31 when I was diagnosed with Stage 3 breast cancer. My daughter, a 21-year-old, is in this line, too. She tested positive for BRCA1. She will also have to learn how to take extra care. (Testimony by Rebecca Fisher to SACGHS, October 18, 2004)

One aspect of this extra care that individuals with a family history of cancers have to practice, Fisher said, is hiding the results of their genetic tests, even from health care providers, for fear that insurers will penalize them. She described this as an additional burden of care that her daughter,

who testing positive for BRCA1, will have to practice:

> But the care that Katie will have to learn how to take includes not only the low-fat diet she's already eating and the daily exercise regimen she's undertaken. It includes more than the breast self-exam she's required to perform monthly, and believe me, I do remind her. It even goes beyond the MRIs of her breasts she will start receiving when she turns 25. The extra care she will have to learn how to take demands that she, like me and like everyone in our family who has this mutation, hide -- that is, hide, H-I-D-E -- her genetic information even, and perhaps especially, from those health care providers most likely to help her manage her lifelong predisposition to disease. (Testimony by Rebecca Fisher to SACGHS, October 18, 2004)

"Unfortunately," Fisher said, "that's what we're reduced to. Hiding integral health information is the only fail safe way we can avoid discriminatory practices such as the loss or denial of health insurance or the loss or denial of employment, because there simply is no comprehensive federal legislation that patently forbids insurance or employment discrimination on the basis of genetic information."

Fisher also repeated the familiar message that fear of genetic discrimination was preventing people from taking advantage of the genetic testing that might save their lives by giving them advance knowledge of the risks they carried in their individual genomes. "Fear and innuendo surround the brave new world of genetic information," she told the Committee. "People are afraid. Their fear keeps them from being tested, even when this test might make the difference between whether they live or die." But it was for children that Fisher feared the most, especially her daughter, "who must live not only with an exponentially higher risk of developing a terminal disease but also with the burden of never knowing

whether or when she will legally be asked to take a genetic test as a condition of employment, be lawfully fired from a job because she's very likely to get breast cancer, or be legitimately denied health insurance or life insurance on the basis of her genetic predisposition to disease."

Fisher's testimony, like that of the other six who spoke that day, emphasized the dual properties of genetic testing. Testing provided powerful, even life-saving, knowledge of future risk of disease. But it also increased the risks to individuals seeking that extra knowledge and taking that extra care that insurers and employers might use that information against them. Like the others who testified, Fisher's testimony also conveyed the anguish of genetic decision-making and a shared family history of disease. But what stood out in her testimony was her use of rights-based language to describe what was at stake for her and others in what Heidi Williams and SACGHS Chair called "the genetics community."

Like Heidi Williams and Phaedra Malatek, Fisher characterized genetic discrimination as a civil rights problem. She invoked Title VII of the Civil Rights Act, which prohibits discrimination by employers on the basis of race, colour, sex, religion, and national origin. "It is not a function of insurance companies' and employers' decisions to take the moral high road and, out of the kindness of their hearts, remain disinterested in this information in the same way that they are legally obliged to remain disinterested in information such as race, gender, creed, or sexual preference," she told the Committee. Fisher's voice rose, and she pounded the table to make her point:

> In my opinion, genetic information is no different from any other

> essential distinguishing information about any human being, all of which is by law kept off the bargaining table that bears up this human rights-based society. But if this argument is truly different -- okay, I'll give you this. If this argument is truly different, if because of its fiscal component, as the United States Chamber of Commerce might argue, we must locate this debate within the framework of an implicit utilitarianism, I would point to professional contributions I and other genetically vulnerable people have been able to make because we've been lucky enough to remain considered employable. (Testimony by Rebecca Fisher to SACGHS, October 18, 2004)

Fisher was not simply advancing the argument that government should defend the civil rights of Americans with positive genetic diagnoses because their immutable markers for disease were "essential distinguishing information," like skin colour and sex. She also alluded to a "social contract" whereby those with genotypes that caused disease or heightened their risks of disease had a civic duty to offer up their bodies and family medical histories to researchers:

> I would point to the contributions my daughter, 21 years old, hopes to make with her two degrees in public policy and economics from Duke University. I would point to the way in which our family's completion of innumerable psychological questionnaires, the donation of tissue from our bodies, and the giving of our blood have advanced medical science. I would argue that we are, in fact, making a difference for the health of all people, everyone in this room, that we've lived up to our end of the social contract and deserve the same fundamental legal protections that are extended to all Americans. (Testimony by Rebecca Fisher to SACGHS, October 18, 2004)

Race, which had been absent from most of the discussion on genetic discrimination throughout the hearings, made a dramatic reappearance as Fisher concluded her testimony with the most powerful image of the day: the image of Rosa Parks refusing to give her Montgomery, Alabama bus

seat to a white passenger in 1955:

> We with strong family histories of disease in which the baton of illness has been passed from generation to generation are simply the first line of defense against a staggering spectrum of possible abuses. We want to be heard, we want to be protected, and we don't want to sit in the back of the bus anymore. (Testimony by Rebecca Fisher to SACGHS, October 18, 2004)

With the implication in her comment that members of this genetics community were potential second-class citizens, race became the most powerful mobilizing metaphor of the day.

Responses to the Testimony

Committee members and attendees responded to these testimonies with sympathy, outrage over the injustices that these individuals had experienced, and promises to do more to protect individuals from fearing genetic testing. Ed McCabe, speaking with characteristic fervour, dismissed criticisms that genetic discrimination did not exist:

> We as a genetics community, and also as members of the public, have been told that genetic discrimination does not exist. We've been told that over and over. In fact, scholarly articles have been written and are referenced in the genetics literature where the authors made inquiries to insurance companies, and guess what? They said there is no genetic discrimination. Yet, all of us know that it exists, and that's why this is so important today. (Comments by Ed McCabe, SACGHS hearings, October 18, 2004)

Francis Collins concurred with McCabe, telling the seven who testified and those present that the problem needed to be fixed:

> I also want to thank all of you for the very powerful and moving

> stories that you have told, which certainly underline in stark and compelling terms the need to do something about a situation which grows worse every day. It is, I'm sure, a great disappointment for all of you that we haven't fixed this by now, when the arguments are compelling, when you can see that the likelihood of more and more genetic testing being offered is inevitable, and therefore the likelihood of more and more people facing up to the dilemmas that you have faced also becomes inevitable. (Comments by Francis Collins, SACGHS hearings, October 18, 2004)

Yet not everyone present agreed that genetic discrimination was a real or pressing problem. Donald Hadley, an associate investigator and genetic counselor at the NHGRI, also testified to the Committee at the October 18, 2004 "Perspectives on Discrimination" session as a member of the healthcare providers panel. He described what he thought was the real problem. "In summary," Hadley said, "the prevalence of genetic discrimination by insurance companies does not appear to be the key issue. The real issue is that the public perceives that the potential for genetic discrimination by insurance companies is an overwhelming risk, and in my experience this fear provides a barrier to genetic research and clinical genetics care." Hadley told the Committee that this fear "limits our potential for research and basic sciences and social and behavior research." But an even greater problem, he said, was "the missed opportunity to prevent cancer or diagnose it early in persons at high risk who are unwilling to risk the potential of discrimination." Hadley was unequivocal: "Providing federal legislation prohibiting genetic discrimination will reassure the public that genetic discrimination is not a risk, provide an increased opportunity for research to address other, more significant issues, and most importantly reduce mortality and morbidity

associated with cancers diagnosed at later stages." (Comments by Donald W. Hadley, SACGHS hearings, October 18, 2004).

At this point in the hearings, Hadley's opinion seemed to be in the minority. But this changed by late 2005, when the Committee had finished with genetic discrimination as a priority issue. After the Committee had wrapped up its efforts to circulate the "Voices of Discrimination" DVD, the compilation of the testimony, and the legal analysis of current law to genetic activists and champions of legislation, Committee members began talking about these measures as necessary to "reassure the public," as Donald Hadley put it. From the Committee's perspective, the goal of passing federal legislation was no longer to prevent insurers and employers from discriminating against Americans, but to allay Americans' fears of discrimination so that they could benefit from, and contribute to the progress of, genomic research.

DISCUSSION

The seven individual testimonies in the "Perspectives on Genetic Discrimination" session at the SACHGS hearings accomplished two things. One, the discrimination narratives challenged the NHGRI's vision of personalized medicine as a bounded encounter between patient and clinician. The stories that these six women and one man told made it very clear that it is families, and not individuals, who bear the burden of deciding whether to undergo genetic testing, handling a positive diagnosis and its implications for the extended family, and anticipating adverse outcomes of testing, such as discriminatory decisions by insurers or employers. The imaginaries of family inheritance that the seven outlined

on October 18th, 2004, featured a strong sense of shared responsibility for disease and a family burden of genetic decision-making. Decisions by one family member (to undergo testing and risk exposing other family members to possible discrimination; to choose anonymous testing; to refuse genetic testing and risk dying prematurely) reverberate across the entire extended family. To risk the safety and future of oneself, by undergoing genetic testing, is to risk the safety and future of the entire extended family.

Secondly, the testimonies that the seven delivered made it clear that Americans with a known disease genotype—members of the genetics community, as several described themselves—were vulnerable subjects, deserving of legislated protection. They told stories that emphasized the seriousness of genetic discrimination as a serious civil rights problem that warranted the same attention as discrimination against African-Americans did in the 1960s, with the passage of the Civil Rights Act. Other vulnerable subjects that might have competed for the title of "victim of discrimination," particularly the uninsured, racial minorities, and the disabled, were erased throughout the hearings or simply not present. Even Francis Collins's vulnerable consumer—the consumer of junk science—receded against these stories.

I want to explore more carefully why these seven were able to seize the mantle of "victims" at the hearings, and why the testimony established genetic discrimination as a pressing civil rights problem. First, these testimonies were performative. They were the singular human drama that unfolded at the hearings. The seven who testified to the Committee on

October 18th, 2004, told stories of misfortune, suffering, and loss that communicated fear, worry, guilt, anger, betrayal, and suffering. These six women and one man described watching family members sicken and die from devastating illnesses such as breast and ovarian cancer; anguishing over whether to participate in potentially life-saving genetic testing and research that might bring treatments; contemplating prophylactic surgeries; planning for the onset of degenerative and fatal illnesses such as Huntington's; feeling guilt for passing their potentially crippling or lethal alleles to their children; and worrying about whether their children's bright futures would dim because of their own testing decisions and the alleles they passed on. In their raised voices and passionate pleas to the Committee to pass federal legislation, their anger, fear, and desperation were palpable. It was impossible not to be moved by listening to the emotion in their voices. What made this emotion so stark is that their testimonies were also a respite from the mostly technical and procedural discussions at the hearings. The emotional content of these testimonies unified everyone, reminding them of their shared humanity. The presence of living, breathing victims of discrimination at the hearings elevated the threat of discrimination from something that might happen to a few people, to a real problem that any American with a family history of a genetic disorder could experience.

The language choices of the seven also reinforced their status as victims. Comments like "Scores of people across this nation are being persecuted on the basis of their genetic status" (Heidi Williams) and "People are afraid" (Rebecca Fisher) invited comparisons to victims of hate crimes, or political refugees. Others, such as Phaedra Malatek's ("The Civil

Rights Act amendments are there to protect people from discrimination based on genetic makeup that we can see, be it skin tone, gender, or disability. A person's genetic makeup that isn't visible should be equally protected"), compared genotype to other immutable markers of difference and discrimination in the United States, like sexual orientation and skin colour. These framing comments communicated the idea that Americans with a genetic diagnosis are marked subjects who must be protected from predatory institutions at all costs.

But the main reason why the seven were able to monopolize the territory of victims is because there was no real competition for this title from other actors, including (and especially) disability activists.[6] Their voices were some of those "missing" from the hearings. In her studies of Congressional hearings, policy anthropologist Phyllis Chock (1991, 1994, 1995, 1996) has documented how certain constitutions of a subject can be made to "stick" in policy settings when competing constitutions of that subject are erased or ignored from the discourse. At the SACGHS hearings, one of the ways this erasure occurred was by absence: other vulnerable subjects did not participate in the hearings. I have already discussed, in Chapter 5, how the logistics of the hearings made it difficult for certain categories of people (namely, the poor, the illiterate, and the working) from attending. But some communities, such as disability advocates, evidently chose not to participate in the hearings.

Throughout the hearings, I struggled to understand why representatives from organizations that might have competed for the status of "vulnerable Americans" did not show up. The SACGHS hearings

struck me as an important forum for social movement actors with an interest in changing the health care system, to attend, to lobby the Secretary of the Department of Health and Human Services (DHHS), to network with scientists and federal representatives, and to keep their interests in the public spotlight. The hearings were, after all, being webcast. It was not until after I attended the SACGHS hearings on the DTC genetic testing industry in June 2005 and puzzled over the absence of that industry from the hearings, that I considered the possibility that social movement organizations representing other vulnerable subjects, particularly disability activists, regarded the SACGHS hearings as irrelevant to their interests.

This possibility had not occurred to me until after the June 2005 hearings on DTC genetic testing, which I attended. I was surprised by the absence of representatives of this industry from those hearings, given that their industry was under attack (from the NHGRI and the Committee) as hawking junk science to consumers. In my view, some of the DTC businesses that offer the same genetic testing services directly to the public that clinicians order from laboratories, and have genetic counsellors and clinicians on staff to interpret the results for customers, had missed a unique opportunity not only to set the record straight about how they operate, but to increase public awareness about the industry and promote their services.

I asked some of the other attendees if they had any idea why this industry was so noticeably absent from the hearings. Carol, a bioethics consultant, told me that she thought many of the DTC companies didn't

know about the hearings, an explanation that I doubted. Greg, a consultant to a medical device manufacturer's association, offered me a more convincing explanation. In hushed tones, he said, "You have to be careful who you talk to here and what you say. The industry thinks that the scope of the Committee is too broad and the discussion on DTC testing too narrow. They don't see this forum as relevant to their interests." This made more sense. Elizabeth, a consultant for Roche Pharmaceuticals taking notes for the company, offered another explanation. She told me that Roche had adopted a wait-and-see stance on attending the hearings, wanting to see first whether the Committee would have any effectiveness or clout in making recommendations on DTC testing and having them implemented, before participating.

It was some time before I realized that, despite their standing as a federal advisory committee open to the public, one that took care to solicit the views of the public on diverse issues affecting the health care system, attendance and participation at the SACGHS hearings was very insular. These hearings represented the interests of what Chair Ed McCabe and participant Heidi Williams called "the genetics community:" genomic researchers, genetics health care professionals, industry, genetic patient support groups and coalitions, and of course, the NHGRI. [7] Because organizations like the National Society of Genetic Counselors and the International Society of Nurses in Genetics made frequent appearances at the SACGHS hearings to promote their interests, particularly the need to dramatically increase the number of certification programmes for genetic counselors in the United States, I had assumed that other reform-minded organizations and advocacy groups would also come to the SACGHS

hearings to promote their interests or voice dissent. To be more specific, I had assumed that I would hear strong voices of public dissent at the hearings. But no actors with broader reform goals made presentations at the hearings. For example, there were no representatives from Physicians for a National Health Program, the oldest advocate of a single-payer, national health insurance program in the United States, urging the Committee to recommend that the country adopt a single-payer system to facilitate the integration of personalized medicine into the health care system and deliver the benefits of the revolution in medicine to the uninsured. The Black Women's Health Imperative, an advocacy organization whose goal is to "[m]ake Black women's health an imperative for federal and state governments and communities," did not come to the SACGHS discussions on the proposed large-scale cohort study to ask NHGRI Director Francis Collins how the study would improve health disparities amongst African-American women.

It was the absence of a disability rights voice from discussions about genetic discrimination and personalized medicine that surprised me most, even though this is a large and diverse set of actors with competing interests in genetic screening and genomics. Some disability activists have been critical of the eugenic implications of using prenatal testing and genetic screening to prevent the birth of disabled babies (see Parens and Asch 2003). Others, such as deaf activists and members of the Little People of America, have insisted on their right to use prenatal diagnosis to ensure their children are born with a disability, like them (see, for example, Taussig et al 2003). While attending the hearings I wondered: Where were the voices of disability activists at the hearings, those who

would seem to have much at stake in having their interests represented in a nation committed to routinizing genomic medicine as part of clinical care? Advocates for the disabled made only one appearance in the hearings, and it was nearly invisible. United Cerebral Palsy and The Arc of the United States, member organizations of ADA Watch/National Coalition for Disability Rights (NCDR), submitted a joint written statement to the Committee on genetic discrimination. Their statement appeared in the "telephone book," the compendium of all public testimony submitted to SACGHS on genetic discrimination.[8] Acknowledging the long history of insurer and employer discrimination against "[p]eople with mental retardation, cerebral palsy and other disabilities," the two organizations pledged their support for passage of federal nondiscrimination legislation in the hopes that such a law would reassure Americans and encourage them to seek genetic testing.[9]

Clearly, large disability advocacy organizations, such as the National Coalition for Disability Rights and the American Association of People with Disabilities, which work to defend or reform the ADA, would not use their resources to attend the SACGHS hearings, where the ADA was not under discussion. But it was less obvious to me why some of the smaller disease-specific disability organizations that have been some of the most outspoken critics of prenatal screening and genetic testing would not attend the SACGHS hearings to challenge some of the assumptions about personalized medicine and whole-genome sequencing. I have no answers to this question, and it needs investigation.

CONCLUSION

The in-person testimonies to SACGHS on October 18th, 2004, did not simply legitimize genetic discrimination as a real problem affecting Americans. They helped to frame it as the most significant civil rights problem in the United States since segregation. Their testimony elevated the threat of discrimination from something that might happen to a few people, to a serious problem that any American with a family history of a genetic disorder could experience. In their raised voices and passionate pleas to the Committee to pass federal legislation, their anger, fear, and desperation were tangible.

However, what was remarkable about the testimonies of the seven Americans is not that they described discrimination by insurers and employers. Discriminatory decision-making informs medical underwriting in the U.S. health insurance industry. Employers also practice discriminatory decision-making in pre-employment screening of candidates and family medical histories, and in their promoting, compensation, and firing practices. Had the Committee broadened its call to include experiences of discrimination from healthy, asymptomatic individuals with invisible, non-genetic health risks (for example, from HIV-positive individuals), it is likely it would have uncovered experiences of discrimination from equally angry and outraged individuals. It is also not surprising to hear individuals express hostility towards health insurers. Insecurity about health care coverage in the United States is widespread. Coverage is tied to full-time employment, but even being employed is no guarantee of maintaining coverage: the insured can face the prospect of personal bankruptcy from unanticipated medical costs, and

monthly premiums can be prohibitively expensive. What makes these stories merit closer examination are the assumptions that these seven victims of discrimination hold about the personal and social good of genetic testing, and their articulation of their rights and duties as citizens living in a genomic nation.

These seven did more than just tell stories that legitimized genetic discrimination as the most serious civil rights problem since segregation. Drawing on their biological claims of immutable difference and vulnerability to persecution, they made citizenship claims on the NHGRI and on Congress. They claimed their rights as consumers and prudential subjects to use genetic technologies to predict and control disease. They articulated a civic duty to make their bodies and family medical histories available to researchers—what Rebecca Fisher called a "social contract." They called on Congress to pass legislation to protect their entitlements, and those of their children. The entitlements they identified were to not just health insurance and jobs, but to the promised goods of the genomic revolution: genetic testing as a tool to generate knowledge about their risks of disease, and to predict and control their futures.

To regard these expressions of hope, fear, rights, and duties as simply an instantiation of the pursuit of special interests in a rights-driven society would be to miss the broader political and moral economies of hope created by the Human Genome Project and the NHGRI. The claims that these individuals made about the promise and predictive power of genetic testing must be located within the national imaginaries expressed throughout the hearings, and the NHGRI's commitment to genomics as a

large-scale scientific initiative. The SACGHS hearings, which amplified this discourse, reveal that something else is going on besides the individual expression of rights claims to equal treatment, or the pursuit of a fair American society that rejects discrimination in any form. This "something else" is a citizenship project, part of the state's efforts to build a genomic nation.

This citizenship project is what I call "genomic citizenship." In Chapter 9, I explain what I mean by genomic citizenship and contrast it to the construct of genetic citizenship advanced by medical anthropologists Deborah Heath, Karen-Sue Taussig, and Rayna Rapp. I examine the logic and implications of genomic citizenship, starting with the idea that there is something special about discrimination based on a genetic diagnosis.

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[1] Lack of access to health care providers is not the same problem as lack of insurance coverage, although the two can overlap. Millions of insured Americans who live in poor or rural areas of the country lack access to basic health care because these areas are underserved by physicians. The National Association of Community Health Centers calls these Americans "the medically disenfranchised" and "the medically underserved." Compounding the problem, some health care providers refuse to accept Medicaid patients because Medicaid reimburses physicians at lower rates than do private insurers (Fuhrmans 2007). Medicaid patients who are rejected by health care providers, particularly those living in areas with few health care services, have few choices. They can go to hospital emergency rooms and endure long waits for basic medical care, pay out-of-pocket for medical services, or forgo medical care altogether.

[2] The expression of single-gene disorders follows Mendelian patterns of inheritance. Genetic tests to detect the presence of autosomal dominant and autosomal recessive genes can be said to have predictive power for the expression of these genes—but not for the severity of illness or age of onset.

[3] "Penetrance" describes the percentage of individuals with a particular genotype who also develop the phenotype, and is expressed as the lifetime risk that an individual with that genotype will develop the phenotype. In a single-gene, autosomal dominant genetic disorder with 95% penetrance, for example, 95% of individuals with the dominant gene

will develop the phenotype, and 5% will not. The alleles associated with single-gene disorders are considered to be highly penetrant, because they often, or almost always, result in a disease phenotype. Susceptibility genes, on the other hand, are generally classified as having low penetrance. However, the picture is more complex than this. There can be enormous variation in penetrance within disorders. For example, mutations in the BRCA1 and BRCA2 tumour-suppressor genes are associated with an increased risk of breast cancer. However, estimates of the penetrance of these mutations have ranged from a high of 90% penetrance by 70 years of age, to more recent estimates of 45% to 68% penetrance (Burke and Austin 2002).

[4] With breast cancer, there may be one or more still-unknown inherited susceptibility genes that also increase the risk of the disease. However, genetics still accounts for a small percentage of all cases of breast cancer. Environmental, physiological, and lifestyle factors appear to account for most cases.

[5] After Williams's testimony to SACGHS on October 18, 2004, Heidi Margulis, senior vice-president of Humana, wrote to Sarah Carr, the SACGHS staff coordinator, clarifying Humana's decision and outlining the corrective steps it had taken following the denial of coverage to Williams. In her December8, 2004 letter to Carr, Margulis stated that Humana had "discovered that an underwriter had incorrectly declined Ms. William's [sic] application for dependent coverage with us." According to Margulis, "[i]t has never been Humana's policy to make a coverage determination based on someone's status as a carrier for genetic disease or based on the results of a genetic test" and the company had since introduced a training program to educate its underwriters. Source: Letter from Heidi Margulis to Sarah Carr, "Public Perspectives on Genetic Discrimination: September 2004 – November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[6] I acknowledge that there is no singular "disability community" or "disability rights movements" in the United States. Disability organizations number in the hundreds, and represent diverse interests and priorities.

[7] For example, Chair Ed McCabe made the following comment during this session: "We as a genetics community, and also as members of the public, have been told that genetic discrimination does not exist." (Comments by Chair Ed McCabe, SACGHS hearings, October 18, 2004). Heidi Williams used the expression the "genetic community."

[8] The Arc, formerly known as the Association for Retarded Children, is an advocacy organization that formed in 1950 as to support families dealing with mental retardation. ADA Watch/National Coalition for Disability Rights, which formed in 2001 as ADA Watch, is a coalition of disability and civil rights organizations that work to prevent politicians from eroding the ADA.

[9] Letter from The Arc and United Cerebral Palsy Public Policy Collaboration, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's

Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

INTRODUCTION

In Chapters 7 and 8, I argued that the SACGHS hearings were not just a site for Americans to air their grievances about insurers and employers, or for the Committee to identify barriers to integrating genomic medicine into the health care system. The SACGHS hearings were a site at which the NHGRI and its allies worked to persuade the public (and Congress) that the country needed federal nondiscrimination legislation. The "Perspectives on Genetic Discrimination" session that SACGHS held on October 18th, 2004 was the most performative element of these efforts.

In this chapter, I offer a reflexive account of the testimony of the seven Americans who appeared at the hearings that day. I argue that these seven Americans were model citizens for an emergent genomic nation, and that they were negotiating for expanded civil rights protection for all Americans, to include genetic status as a marker of difference. In negotiating this right, they also acknowledged their duties to practice genetic self-surveillance and to participate in genomics research. This set of rights and duties constitutes what I call "genomic citizenship." Genomic citizenship is a model of good citizenship for Americans in a nation committed to developing personalized medicine as norm of clinical care. I examine the collective messages and complaints that these seven individuals delivered to the Committee to flesh out the parameters and implications of genomic citizenship.

I contrast genomic citizenship to genetic citizenship (Heath et al 2004; Rapp 2002; Rapp et al 2006; Taussig et al 2003), drawing on the work of

medical sociologist Anne Kerr (2003a, 2003b) to sharpen these contrasts. The two citizenship forms turn on different claims and different types of biosociality (Rabinow 1996). Genetic citizenship describes the claims for scientific inclusion by marginalized populations with rare genetic disorders, some of whom have forged kinship ties through their shared disease identities and experiences. These individuals have mobilized a self-ascribed biosociality to produce new networks, social forms, and models of knowledge-production. Genomic citizenship, on the other hand, describes the geneticization of all Americans by some genetic activists. In their statements about why all Americans need protection from genetic discrimination, these activists enroll everyone into a biosociality of a flawed genome that is being made transparent by researchers. According to these activists, all Americans are members of the "genetics community," whether they realize it or not. What this implies is that all Americans, who share a kinship tie through their inherently flawed genomes, should practice genetic self-surveillance and act pre-emptively on their individual risks of disease.

LISTENING TO THE VOICES OF DISCRIMINATION

Since 1993, activism in connection with genetic discrimination designed to pass federal nondiscrimination legislation has been led by the NHGRI, in partnership with genetic interest groups, breast cancer organizations, legal scholars, and industry (for example, companies such as 23andMe, Affymetrix, and Myriad Genetics). This group of actors has defined genetic discrimination around a political and moral economy of personalized medicine. Proponents of personalized medicine imagine

Americans as two types of subjects, and in fact, needs Americans to step into both roles. One type of subject at the heart of personalized medicine is the "patient-in-waiting" (Sunder Rajan 2005:22), who takes responsibility for his or presumed genetic liabilities by acting as a consumer of sequencing and drug treatments. This is the imagined subject of the future that Francis Collins describes when he outlines the virtues of routinized whole-genome sequencing at the SACGHS hearings, or when he outlines a scenario in which the compliant patient "Betty" undergoes whole-genome sequencing and uses pharmacogenomic medicines "tailored" to her genomic profile to prevent heart disease. (I discuss this scenario at length later in the chapter).

The other subject of the political and moral economy of personalized medicine is Americans who will make their bodies and family histories available to genomic researchers for studies (such as the NHGRI's proposed large-scale cohort study) and drug trials (although activists do not specify which drug trials they have in mind). They, too, are entwined in the public discourse on genetic discrimination. These imagined, bioavailable subjects are one of the necessary material resources to the future of genomic research and the clinical trials that will deliver personalized medicine.

The link between genetic discrimination and the bioavailable subject is quite clear in claims that activists make. Fear of genetic discrimination stops Americans from participating in research and trials, they claim, and impedes progress. For example, Fran Visco, president of the NBCC, writes in her testimony on genetic discrimination to the Committee that women

are "afraid to enroll in research and clinical trials, and this in turn threatens the ability of the scientific community to conduct the research necessary to understand the cause and find a cure for breast cancer."[1] Sharon Terry echoes Visco by stating that "[a] fear of discrimination discourages that participation—adding another hurdle to the pathway from basic science and health care services," and that "without nondiscrimination assurances, people will not participate in the very studies that could lead to more precise interpretations of 'risk' measures, better understanding about interplay between gene and environment and other genes, and the development of preventative treatments—sometimes for their own condition."[2]

From 2005 to 2007, I attended six SACGHS hearings of two days each, watched webcasts for the hearings from their start in June 2003 through the end of 2007, and read the available transcriptions for this same five-year period (2003-2007). I examined all of the testimony submitted to SACGHS on genetic discrimination and other issues, such as coverage and reimbursement. I read the reports and correspondence produced by the Committee, talked to participants at the hearings, and in other settings, and hunted down policy statements and press releases made by participants outside of the hearings. What I saw during the three-year period in which the Committee was actively deliberating on genetic discrimination (2003 to 2005) was the discursive construction of a "vulnerable" American around the problem of genetic discrimination. This vulnerable American was not racially-marked, poor, disabled, illiterate, or uneducated, but white and Latina, middle-class, educated, articulate, and driven to seek good health through technological intervention and lifestyle

changes.

The machinations on genetic discrimination at the SACGHS hearings were part of the genomic nation-building that the NIH has undertaken since 1986, and the NHGRI since 1993. Assimilating Americans into a genomic nation as consumers and bioavailable subjects is a citizenship project, and the SACGHS hearings, which were a site for complaints and claims about genetic discrimination, were also a site for "making up citizens" (Rose and Novas 2005; Rose 2007). In the course of setting out their imaginaries of the nation and Americans, and in offering their testimonies on genetic discrimination, participants at the SACGHS hearings articulated norms of good citizenship in a nation where health and disease have been undergoing geneticization (Lippman 1991) since the launch of the Human Genome Project in 1990.

In Chapter 8, I identified one theme in each of the seven testimonies delivered to SACGHS on October 18th, 2004. In the next section, I offer a reflexive account of the testimonies and identify the collective messages that these Americans delivered to the Committee, to understand what they were negotiating. I argue that the seven Americans who testified to the Committee on October 18th, 2004 were model citizens for this emergent genomic nation. The stories that they told were an allegory for the struggles of the middle-class. They could have been told only in a post-Human Genome Project era, with the promises that the NIH and the NHGRI have made to Americans about delivering a revolution in health care. What rights and duties are attached to genomic citizenship? Who belongs in a genomic nation, and who is excluded from membership?

Misfortune was an unmistakable image of these narratives. It took two forms: the personal and familial devastation from disease (and the fear of future disease), and the unexpected treatment by insurers and employers of these individuals as actuarial subjects. It was genes that unified both forms of misfortune. “Genes,” as biological anthropologist Alan Goodman (2007:228) so elegantly puts it, “portray stable histories and predicable [sic] destinies.” In these stories, the stable histories that genes portrayed were consanguinal histories of disease transmission, worry, fear, and suffering.

Genes also held a special place in these stories as markers of difference and disorder, like skin colour or sex. As Phaedra Malatek told the Committee during her testimony, “The Civil Rights Act amendments are there to protect people from discrimination based on genetic makeup that we can see, be it skin tone, gender, or disability. A person's genetic makeup that isn't visible should be equally protected.”[3] Paula Funk emphasized the immutability of one’s genotype, and the permanence of making it visible. “Finding out your genetic status is permanent,” she told the Committee. “You can't take it back, and it isn't something that you can change your mind on.”[4] The predictable destinies that genes indexed in these stories were the duties of seeking pharmaceutical and surgical interventions, and a lifetime of “taking extra care.”[5]

While genes are a source of misfortune, the seven who testified to the Committee that day made it clear that genes were also a source of hope. Whether genes are the high-penetrance rare mutations that follow Mendelian patterns of inheritance, or low-penetrance susceptibility genes

that heighten the risk for acquiring certain diseases in conjunction with unknown factors, knowledge of their existence helped these individuals to make sense of the misfortune and disorder that has visited these lives, and to ward off the seeming randomness of disease (cf. Evans-Pritchard 1937). In these stories, genetic explanations for disease transformed accidents of replication and inheritance into rule-bound, ordered events, into which humans can intervene (cf. Evans-Pritchard 1937). Genes also shift blame away from the individual, albeit temporarily (Nelkin 1996).

To intervene into accidents of replication and inheritance requires making personal and biomedical negotiations. The personal and biomedical negotiations that these individuals have made include anguishing over whether to undergo genetic testing, making lifestyle changes, monitoring themselves for signs of disease, and sacrificing body parts to surgery. These interventions and negotiations turn on the assumption that the individual is the locus of disease.

The testimonies recall anthropologist Nadia Abu El-Haj's (2007:2191) observation that "[t]he concept of the individual at medical risk also presupposes a distinct moral economy that calls on the patient-in-waiting to act responsibly by tailoring her lifestyle to the specific genetic risk that she bears." By this, I do not mean that these seven who testified delivered atomistic stories to the Committee about their experiences of disease. On the contrary, their stories put the bonds of familial affliction front and centre of the experience of disease, well beyond what the NHGRI has depicted in its vision of personalized medicine. Rather, I mean that these testimonies illustrated what Nikolas Rose (20o1:12) calls the

"individualizing and clinical" focus of biopolitics. More precisely, these testimonies articulated an internalizing medical belief system (Young 1976) that privileges the gene as the causal agent and locates responsibility for preventing future disease with the individual.

This internalizing perspective was most apparent in the testimony of the four women who had tested positive for inherited mutations in BRCA1 and BRCA2 genes that increase susceptibility to breast cancer (Rebecca Fisher, Tonia Phillips, Paula Funk, and Maria Carolina Hinestrosa). None questioned whether occupational or environmental exposure might have affected the functioning of their tumour-suppressing BRCA1 and BRCA2 genes. Nor did they entertain any environmental etiologies, such as the endocrine disruptor hypothesis (see, for example Ley 2006; McCormick et al 2003; Zavestoski et al 2004).

I was astonished by the commitment of some of these women to a profoundly internalizing etiology that did not admit any gene-environment interactions. They appeared unwilling to assign any blame to third-parties for exposure to carcinogenic agents. The exclusion of a role for environmental contaminants in these stories was remarkable to me, given four decades of feminist and environmental health activism since the 1960s that has challenged researchers and epidemiologists to study the role of suspected carcinogens and other triggers in disease, particularly cancers.[6]

It turns out that I am not "with the programme." The exclusion of environmental etiologies of breast cancer, such as the endocrine disruptor hypothesis, from some of the testimonies at the SACGHS hearings is

consistent with how the U.S. print media has marginalized environmental etiologies of breast cancer and instead emphasized individual responsibility for disease (see Brown et al 2002). My own understanding of the etiology of cancer was influenced by the efforts of grassroots environmental and feminist activists of the 1960s, 1970s, and 1980s, who drew attention to the role of industrial contaminants in the development of cancer. It turns out that this explanatory model, in which contaminants play a central role in turning off genes that manufacture tumour-suppressing proteins and turning on genes that cause cells to reproduce wildly, is no longer mainstream.

In fact, there is a perfect correspondence between the internalizing narratives that were at the centre of these testimonies on genetic discrimination, and the NHGRI's own vision of personalized medicine. To illustrate: when Francis Collins delivers talks on personalized medicine, he often brings a PowerPoint presentation to his guest appearances. Whether the setting is a meeting of the SACGHS hearings, the Personalized Medicine Coalition, or the Physician Assistant Education Association, his PowerPoint show depicts the same scenario of the future. It is called "Betty's Story."[7] In 2015, a 25-year old patient named Betty goes to her doctor (or physician assistant), who recommends that she undergo whole-genome sequencing and complete the Surgeon General's Family Health Tool. Based on her family history of heart disease and her genotype, which indicates an elevated risk of heart disease, her doctor recommends "a program of prevention based on diet, exercise, and medication precisely targeted to her genetic situation." When, at age 75, Betty develops a pain in her left arm, her health care provider examines her genotype to select an

appropriate drug treatment that is tailored to her enzyme profile. The drug prevents Betty from having a heart attack. This NHGRI presentation selectively reinforces what Brown et al (2002) call the dominant epidemiological paradigm in medical and popular explanations for disease. Granted, Betty has an elevated risk of cardiovascular disease, not breast cancer. But in Betty's story, the clinician does not discuss her work environment to determine if she has had occupational exposure to danger levels of carbon monoxide or lead, which are associated with cardiovascular disease (see, for example, Gustavsson et al 2001). Nor does Betty question whether any of the medications she is taking are responsible for her symptoms. Ultimately, Betty, a compliant patient, accepts all responsibility for unwittingly carrying a genetic propensity to heart disease.

Because all of the stories told to the Committee on October 18th, 2004 touched on insurer or employer decisions, or on fears that third parties would shut them out of their entitlements in the future, the seven individuals who testified that day seemed to be protesting the inherent unfairness of the health insurance industry. They voiced their objections to institutions valuing them, as Rebecca Fisher angrily put it, in "dollar signs, costs, increased liability, and the odds of my dying an expensive death," instead of their potential to contribute to society.[8] In the next section, I draw on the work of political scientist Anna Kirkland (2008) on obesity to challenge the perception that the primary complaint of these individuals was the inherent unfairness of the health insurance industry.

In her analysis of obesity as a civil rights problem in the United States,

Kirkland describes several cases in which obese workers have challenged employers' refusals to hire or promote them into positions for which they are otherwise qualified. The complaint of these obese workers is that employers are applying the logic of actuarial personhood by using uniformly standardized measures of health and disease, such as body mass index (BMI), in an arbitrary way, to evaluate them and subsequently disqualify them. These workers have argued that they are in fact healthy, not disabled, and can perform their jobs. In making this complaint, says Kirkland, these workers have invoked the counter-claim of functional individualism: that employers should evaluate each worker based on her skills as an individual and her abilities to contribute to her workplace.

In the testimonies that these individuals delivered at the hearings, there was a similar complaint about actuarial personhood and a counter-claim of functional individualism. Some who testified to the Committee pointed to their contributions, and the potential contributions of their children, to their workplaces, to advancing research, and to society. Yet if their primary objection was to the logic actuarial personhood at work in decision-making by insurers and employers, we would reasonably expect them to call for health care reform that disentangles health insurance from employment, or for a more equitable system of health care coverage that does not exclude healthy Americans based on any indicator of past or future health problem. But this did not happen. Nor did these Americans seek solidarity with other Americans who have experienced non-genetic cases of insurer discrimination, such as obese workers or HIV-positive workers.

What this suggests to me is that their complaint goes beyond actuarial personhood. These individuals were complaining that the NIH and the NHGRI have promised a genetic revolution, but they have not done their parts to protect citizens from penalty and ensure they retain their entitlements to insurance and jobs, even as they have valiantly participated in this health care revolution. These Americans made it clear that they have been the valiant foot soldiers in building a genomic nation by enlisting in the war on disease. They have done their parts, they told the Committee, by adopting the role of patient-in-waiting, by embracing genetic testing, and by taking responsibility for their genetic diseases. The message that they delivered was this: "We're personalizing the risk and consequences of disease as genetic, as something that we carry and we have to fix. We're committed to practicing good health. We're committed to protecting our children and future generations of Americans. In exchange, we want you to pass legislation to preserve our entitlements to health insurance and employment." And Francis Collins, the NHGRI Director, acknowledged their commitment to the programme of genomic medicine, and their sacrifices. As he told them, following their testimony at the October 18th session in 2004, "It is, I'm sure, a great disappointment for all of you that we haven't fixed this by now, when the arguments are compelling, when you can see that the likelihood of more and more genetic testing being offered is inevitable, and therefore the likelihood of more and more people facing up to the dilemmas that you have faced also becomes inevitable."[9]

What is different about genetic discrimination from other forms of discrimination such as obesity discrimination, and why does the former

warrant a federal nondiscrimination law while the latter apparently does not? Individuals claiming workplace discrimination based on genotype have greater protection and recourse to remedies under state and federal laws than do individuals claiming workplace discrimination on the basis of body weight and size.[10] What distinguishes genetic discrimination from obesity discrimination is that the former, unlike the latter, is perceived to be a barrier to the hope promised by genomic discoveries and drug treatments for Americans who already have a genetic diagnosis, and a barrier to the ambitions of the NHGRI to sustain the enterprise of genomic research that began with the HGP.

WHY "GENOMIC CITIZENSHIP"?

Why do we need a new construct called "genomic citizenship," when so many citizenship forms proliferate within medical anthropology? We have biological citizenship to describe the emergence of new categories of the at-risk and entitlement to state benefits (Petryna 2002, 2004; see also Rose and Novas 2005), biomedical citizenship to describe the constitutional rights of Brazilians for free AIDS therapy (Biehl 2004b), therapeutic citizenship to describe how patients mobilize their social networks to access pharmaceutical treatment (Nguyen 2005), and genetic citizenship (Heath et al 2004; Rapp 2002; Rapp et al 2006; Taussig et al 2003) to describe the claims of genetic patient support groups for greater control of genomic research. To understand why I have introduced the construct, I return to the arguments that medical anthropologists Deborah Heath, Karen-Sue Taussig, and Rayna Rapp make about genetic citizenship (see (Heath et al 2004; Rapp et al 2006, Taussig et al 2003),

which I examined in Chapter 3.

Rayna Rapp's work on genetic citizenship with her colleagues Deborah Heath and Karen-Sue Taussig extends some of the arguments and sympathies in a prior essay on disability that she wrote with Faye Ginsburg (Rapp and Ginsburg 2001). In their 2001 essay, Rapp and Ginsburg argue that a powerful disability rights movement in the United States has grown out of social networks of support for families. As a consequence, disability has become more visible over the last four decades. Disability rights has become one of several "social movements that demand inclusion" (Rapp and Ginsburg 2001:543). Greater acceptance of Down syndrome, for example, has also meant more frequent portrayal of individuals with Down syndrome in popular culture. Disability, say Rapp and Ginsburg, is not deviation; it is part of the normal life course of humans. But this increasing visibility has also heightened the tensions with medical practices that pursue "the fantasy of bodily perfectibility through technological intervention" (Rapp and Ginsburg 2001:552). Fantasies of bodily perfectibility and the array of technological tools for reproduction not only obscure the work of raising and integrating a disabled child into social life, they also contradict the increasing traction of disability rights activists for "rights, entitlement, and citizenship," they argue (Rapp and Ginsburg 2001:538).

Rapp and Ginsburg are arguing for more than simply the acceptance of disability as part of the life course. They are championing "a broader understanding of citizenship in which disability rights are understood as civil rights" (Rapp and Ginsburg 2001:545). This is what is at stake for

these medical anthropologists. In this broader understanding of citizenship, deviation from the entrenched fantasy of the perfectible body is embraced, becoming a basis for "democratic inclusion" (Rapp and Ginsburg 2001:552). Although they do not spell out what they mean by democratic inclusion, or what rights and entitlement constitute full citizenship for the disabled, it is clear that they are invoking the idea of a biosociality (Rabinow 1996) of disability, in the claim that bodily differences can become a powerful basis for rights claims amongst marginalized populations.[11]

These arguments and themes make their way into Rapp's theorizing about genetic citizenship as the activism of genetic patient groups and organizations, with Deborah Heath and Karen-Sue Taussig. Their fascination with the politicizing potential of kinship as a basis for social networks is a bridging theme. For example, in one article (Rapp et al 2001:392), they describe their motivation for studying "rapidly emergent social forms in the age of genetics" as an interest in exploring "what makes a relative" and how American notions of kinship are changing. The standpoint of disability advocacy that Rapp and Ginsburg display in their essay also carries over to the sympathetic perspective that Rapp, Heath and Taussig adopt in describing the goals of genetic patient interest groups and in the hopeful conclusions they draw about the democratic potential of genetic citizenship for communities of the disabled.

I would like to be as hopeful as these anthropologists that the activism of genetic patient support groups has opened up democratic opportunities of participation for the disabled. But in the setting of the SACGHS

hearings, I saw something at work that suggested to me a different set of opportunities for engagement and inclusion. Where Heath, Rapp, and Taussig saw in their fieldwork members of rare disease groups forming partnerships with scientists and clinicians, and even developing new models of knowledge production, I saw in the setting of the SACGHS hearings a self-interested genetics community aggressively promoting a federal law by insisting that all Americans are genetic subjects. I watched this drama unfold against the NHGRI's arguments that personalized medicine is inevitable and that launching a prospective large-scale population cohort study is desirable. The biosociality that underlines the claim that all Americans are genetic subjects, which was voiced in many ways, is not the self-ascribed biosociality that Heath, Taussig and Rapp describe in their work with genetic patient interest groups. It is a mandatory kinship of affliction that encompasses all Americans, willing or not. This geneticization of the entire American population shuts down opportunities for the disabled to refuse to be genetic subjects, even as it enacts the democratic ideal of public participation in genomic policy-making.[12]

The sharpest contrast between what these medical anthropologists call genetic citizenship and what I am calling genomic citizenship lies in the respective scopes of their impact. Activism by genetic patient interest groups to control the tools and directions of genomic research has implications primarily for this community of individuals and families who are seeking cures and treatments for their rare disorders. But some members of this genetics community are also making claims about the rights and duties of Americans as genomic subjects, *on behalf of* all

Americans. Their claims about the rights and duties of all Americans, which are thoroughly wedded to their genetic discrimination discourse, have implications not just for themselves, but for the entire nation. The reach of these claims is underscored by efforts to pass a federal nondiscrimination law. This is not simply activism by the NHGRI, the Genetic Alliance, and the NBCC to ban the use of personal genetic information by insurers and employers because genetic discrimination is an intolerable reminder that United States is an unjust society. This is an attempt to remove, through legislation, a perceived objection by the American public, to encourage broader participation in genetic testing and genomic research.

For example, the federal agency that monitors workplace discrimination and enforces the ADA is the U.S. Equal Employment Opportunities Commission (EEOC). The EEOC is the chief federal body with a mandate to litigate cases of workplace genetic discrimination. Yet during discussions by the Committee at the SACGHS hearings on its second day (June 12, 2003), Paul Miller, Commissioner of the EEOC, demonstrated in his responses to questions about genetic discrimination that neither he nor his agency had any sense of its scope. In fact, Miller suggested to Chair McCabe that the Committee might want to conduct some empirical research on the problem. During that same discussion, Alan Guttmacher, Deputy Director of the NHGRI, told the Committee that the NHGRI had not conducted any studies of its own on the scope or impact of genetic discrimination.

What is significant here is that both the EEOC, which is the chief

federal body with the authority to police workplace genetic discrimination, and the NHGRI, which has consistently advanced the argument since 1995 that genetic discrimination is blocking progress in genomic medicine, were telling Chair McCabe that neither had conducted any empirical studies to demonstrate the scope or impact of genetic discrimination. This information appeared to signal to McCabe that the Committee needed to generate its own data, in its own fashion.

In fact, the NIH has funded such studies through its ELSI programme, a fact that Guttmacher did not point out. Since 1995, Francis Collins has clearly linked public fear of genetic discrimination to limits on the progress of genomic research, and advocated for state and federal policies and legislation to limit the use of genetic information by insurers and employers. He has made these statements in the journal *Science*, where he wrote, with his colleagues, that "[a]s genetic research progresses, it will be increasingly important that discrimination and fear of discrimination not be a roadblock to reaping the benefits" (Hudson et al 1995:391). He has also made these statements in NHGRI press releases; in addresses to U.S. Senators on federal nondiscrimination legislation in 2004 and 2005, where he told them that genetic discrimination "slows the pace of science."[13] At the SACGHS hearings, on June 11, 2003, Collins identified genetic discrimination as the most important barrier to discovering the genetic bases of disease, and encouraged the Committee, in one of his PowerPoint slides accompanying his presentation, to "Achiev[e] a legislative solution for health insurance and the workplace" for genetic discrimination.[14] Also in 2003, in a one-page editorial in *Science* written with James Watson (Collins and Watson 2003:745), Collins endorsed

GINA. In an explicit advocacy statement to Congress in this editorial, Collins and Watson called on the U.S. House of Representatives to pass GINA "as soon as possible." The two federal scientists stated that genetic discrimination "can slow the pace of the scientific discovery that will yield crucial medical advances," that "many people have already refused to participate in genetic research for fear of genetic discrimination," and that without a federal law in place, "many in the public will be reluctant to enter into the genome era, and we will not fully reap the rewards of the investment already made in human genome research." Statements such as these indicate that the NHGRI has a strong investment in seeing a federal nondiscrimination law pass to encourage Americans to become participants in the "genome era."[15]

This exchange, between Chair McCabe, Paul Miller, and Alan Guttmacher, reinforced the appearance of the Committee's supposed impartiality as a policy and fact-gathering body reporting to the Secretary of the DHHS and acting independently of the NHGRI. The Committee would take on—apparently for the first time—the task of documenting the problem of genetic discrimination in order to legitimize it to Congress. Yet the head of the NHGRI already had been legitimizing this problem to the public and Congress for ten years.

In the uncertain daily life of post-Chernobyl Ukraine, writes Adriana Petryna (2004:261), "the injured biology of a population" became "the basis for social membership and for staking claims to citizenship." Biological citizenship in Ukraine describes the negotiations of the injured for disability benefits in a terrain of changing biological standards of

radiation injury and the individualized judgment of clinicians.

The model of citizenship that was negotiated during the SACGHS hearings seems, at first glance, to have nothing in common with the biological citizenship negotiated by the state, scientists, clinicians, and the disabled in Ukraine. Seven healthy individuals who carried mutations for rare disorders and susceptibility genes for common disorders, argued at the SACGHS hearings for their rights to participate in genetic testing and research without fearing recrimination by insurers and employers. They argued, along with federal scientists, legal scholars, and leaders of genetic and breast cancer organizations, that they represented all Americans in their right to protect their valuable medical information—which they themselves had commissioned, with their clinicians—from being used against them.

But this claim, that all Americans should be protected against discrimination, rests on the assumption that all Americans are genetically flawed. This is the biosociality of the inherently flawed, transparent genome, which was described by genetic advocates and federal scientists. Whether it was Francis Collins telling attendees that "most of us are carrying risks for future illness somewhere in our DNA," Louise Slaughter writing that "[n]o human being has a perfect set of genes," and that "every one of us is estimated to be genetically predisposed to between 5 and 50 serious disorders," or Sharon Terry writing that "we all have flawed genes," and "[w]e all possess mutations that will become equally and increasingly transparent with tomorrow's technologies," everyone, it seemed, is presumed to be genetically flawed—a form of injury. The implication is

that legislated protection against genetic discrimination should be extended to all individuals, not because they are healthy, but because all carry genetic flaws that will become transparent over time.

This is the language of genetic defect, and its use turns on an imagined "normal" genome that does not exist (see Lewontin 1992). In this world of genetic flaws, disorders, risks, and glitches, deviance from an imagined normal genome become a target not just for insurers and employers, but also for genomic research and medical intervention (see Lewontin 1992). No one at the hearings seemed startled or distressed by this language of genetic defect; no one critiqued the presumption of a fixed and knowable genomic norm as the template against which certain deviations (and not others) would register as genetic flaws. Rather, the claims that everyone is genetically flawed, or that everyone carries risks for future disorders, were well-received by participants. The appeal of this imagined biosociality of all Americans as genetically disordered lies in its rejection of elitism: no one is perfect; no one is better than anyone else. But to my ear, this message of genetic inevitabilism and its corollary, that but for their fears of genetic discrimination, Americans would be flocking to genetic testing, research projects, and clinical trials, had a bullying sound to it. It conjured a vision of Americans as bodies loaded with defective genes that are ticking time bombs, who were also zealous participants in the genomic enterprise. "Whether you realize it or not, whether you like it or not, you're a member of the genetics community," was the message that came across in these written and spoken comments.

I was disturbed, too, by the stories of heroism and courage in the

testimonies. I heard in some of them a barely-concealed condescension towards Americans who have not mustered the courage or the resources to undergo genetic testing to discover the 5 to 50 genetic disorders they are harboring. This sentiment crystallized in the written testimony that Sharon Terry submitted to the Committee on behalf of the Genetic Alliance. In it, she wrote that "[w]e also represent those who do not yet understand that 'Genetics is about ALL of us'," [emphasis in the original]. Terry's statement invoked a social hierarchy of diagnosis-seeking behaviour, in which those outside of the genetics community remain oblivious to their own genetic liabilities.[16]

There are two messages being delivered here. The explicit message is that all Americans have a reason to demand that Congress protect them from genetic discrimination. The tacit message is that having an inherently flawed (and transparent) genome comes with obligations. Knowing one's own genetic predispositions to disease is a moral imperative towards self, family, future generations, and society. This framing language encourages Americans to think of themselves in genetic terms, and chastises those who will not take responsibility for their genetic liabilities. To what extent this second message will be transmitted to the public at large, it is too soon to say. Because it is the Genetic Alliance that has communicated this message most forcefully, it will be worthwhile watching that organization to see where it puts its energies, now that GINA has been passed and the organization no longer needs to lobby for nondiscrimination legislation, and what strategies it develops around the NHGRI's proposed large-scale study. Another place to watch for the transmission of this message is the Surgeon General's Family Health Initiative, a project intended to train

Americans to take responsibility for their inherited predispositions for disease by documenting their family histories and discussing their pedigrees with their clinicians.[17]

Thus, while genomic citizenship appears to be an inclusive and egalitarian model of expanding civil rights protections, it is coercive and divisive. It is coercive because it insists that all Americans are genetically flawed and should take responsibility for their genetic liabilities. It is divisive because the egalitarianism that drives it masks inequalities of access to genetic testing and health care in the United States. The model of genomic citizenship assumes that all Americans have the desire, skills, and resources to pursue genetic self-knowledge and prudential care.

From this perspective, genomic citizenship begins to resemble the biological citizenship of post-Chernobyl Ukraine. Policies on genetic testing and genomic medicine that are formed around the subject of the deliberations at the SACGHS hearings, the rational consumer who is also genetically flawed, will not easily accommodate those Americans who do not aspire to genetic prudence and responsibility, who cannot make rational choices, or who lack the resources to participate in the care of the self.

In this model of genomic citizenship, too, there is an echo of eugenicist thinking from the early twentieth century, when families with good genes were encouraged, through better breeding practices, to populate the nation, while "hereditary defectives" (Danielson and Davenport 1912) and the "socially inadequate" (Laughlin 1921) were the subject of sustained campaigns and laws to curtail their reproduction. In May 2007, on the 80th

anniversary of the Buck v. Bell decision by Supreme Court Judge Oliver Wendall Holmes, disability activists Andrew Imparato and Anne Sommers (Imparato and Sommers 2007) challenged both the temptation to distinguish between "good" genes and "bad" genes, and the tendency to assume that eugenics initiatives targeting disability are a relic of the past. "So long as we speak in terms of good genes and bad genes, recognize a life with a disability as an injury, and allow health policies to value some lives over others," they wrote, "we continue to create human rights violations every day."

The explicit egalitarianism of the message that all Americans harbor bad genes may placate those who fear that the Human Genome Project has revitalized a negative eugenics. But this egalitarianism creates an uneven playing field, in which only the most diligent and resourceful can vie for the title of good citizen in a "genetically optimized population" (Sperling 2007:282). Genomic citizenship lays the foundation for a different kind of genetic discrimination: discrimination against those who do not conform to the norms of good citizenship. What is not being valorized here is, of course, the right not to make genetic choices (Kerr 2o03b; see also Sperling 2007), including the right not to know.

One of the rights that the seven Americans who testified at the SACGHS hearings on October 18th, 2004, articulated was the right of Americans to know their genetic liabilities, without being penalized or "persecuted" by insurers or employers. What, then, of the right of Americans not to make genetic choices, or to remain oblivious to what susceptibilities their genomes hold?

In her research on public involvement in genetic policy-making in the United Kingdom, medical sociologist Anne Kerr (2003a, 2003b) outlines the tensions between the expectation that citizens will participate more in science policy-making, and their increased responsibility towards themselves to prevent disease. She too uses the construct of genetic citizenship, but in a much different way than Heath, Taussig and Rapp. Kerr uses the construct to describe the discourse of individual rights and obligations to participate in genetic self-surveillance, a discourse that, she writes, appears routinely in clinical and policy settings in the UK.

Although Kerr does not refer to the work of Heath, Taussig and Rapp on genetic citizenship, she is skeptical of similar claims made by other social scientists (for example, Petersen and Bunton 2002) that expertise is being reshaped as patient groups form partnerships with medical experts, and that this lay expertise constitutes a new form of empowering citizenship. She questions this "prevailing emphasis on transformation" (Kerr 2003a:210), particularly the danger it presents of overlooking continuities between eugenics of the past and present. While the contemporary emphasis on the individual's right to choose was not an element of earlier eugenics campaigns, she says, "the emphasis upon personal responsibility for the prevention of genetic disease is not new" Kerr (2003a:220).

Kerr also challenges claims about the prospects for democratic inclusion in decision-making and agenda-setting in the UK. She observes that public participation in science policy-making is often elitist, in that genetic experts dominate agenda-setting and decision-making. Moreover,

the language of entitlement obscures the right of socially excluded groups not to make genetic choices. She warns that "we ought to be cautious about demands for individuals' rights to genetic choices and citizens' rights to involvement in policy making about genetics, given the obligations that the language of entitlement can mask" (Kerr 2003b:50). This is especially the case with marginalized populations, she writes. "Their rights not to make genetic choices, and not to participate in 'public debates' that are of little interest or importance in the context of their everyday lives, are being overlooked" (Kerr 2003b:50).

Kerr's thinking about genetic citizenship more closely resembles what I observed at the SACGHS hearings, especially in discussions about genetic discrimination, than the genetic citizenship of expanding opportunities for marginalized populations that Heath, Taussig and Rapp describe. As Kerr (2003b:49) writes, "[t]he discourse of individual entitlement to genetic services and genetic choices readily translates into a discourse of individual obligation to participate in self-surveillance." In the United States, passing federal nondiscrimination legislation is one way in which the civil rights of Americans appear to be expanding while their responsibilities to act genetically are increasing. The insistence that Congress pass protective federal legislation removes a key objection to seeking genetic testing, with no allowance for the possibility that testing may not provide clear answers and may make decisions more difficult, or that there will always be those who do not want to know what genetic flaws they might be carrying.

But there is an important difference between what Kerr describes as

genetic citizenship, and what I observed in my fieldwork in the United States. The model of good citizenship that I observed taking shape at the SACGHS hearings was around the demand that Congress pass civil rights legislation to protect Americans whose genetic activities leave them vulnerable to unfair decisions by insurers and employers. Specifically, in their testimonies to the Committee, the seven Americans turned their biological claims of difference and misfortune into a citizenship claim on the state for all in a uniquely American way. They compared the presence of disease alleles in their bodies to other immutable markers of difference that have become the centre of civil rights battles in the United States and efforts to legislate equality of opportunity. The representation of genetic discrimination as a civil rights problem that is comparable to racial discrimination, and the largely state-driven efforts to pass a federal law banning genetic discrimination, are absent in the UK.

CONCLUSION

The seven Americans who testified at the SACGHS hearings on genetic discrimination are, to borrow an expression from Rayna Rapp (Rapp 1987, 1999), "moral pioneers" of America's genomic age. They are citizens who have endorsed their responsibilities to tailor their lifestyles to the genetic risks they bear (Abu El-Haj (2007:2191). Their narratives were, above all, moral tales: of parental duty and guilt; of individual responsibility for maintaining good health, even if it means sacrificing body parts through surgery; and of the duty of the state, which has promised new tools for good health, to protect them from discrimination by insurers and employers. These moral pioneers are model citizens for a genomic nation

flying the banner of "Preemptive, Predictive, Personalized, and Participatory" medicine (see Zerhouni 2006) because they do not entertain externalizing disease etiologies that locate cause or responsibility with third-parties, such as the workplace or the government. They have embraced their moral responsibilities to produce genetic knowledge of themselves, and to prevent disease by intervening.

There was tremendous pathos in this rights-bearing testimony. The stories that these seven told to the Committee and the public conveyed the anguish of suffering and the dilemmas of genetic decision-making. They also expressed embarrassment and indignation that a wealthy nation would permit its middle classes to suffer at the hands of insurers and employers for trying to prevent disease before it occurs. But there was also hopefulness in this testimony. This hopefulness was evident in the extraordinary but misplaced optimism these individuals expressed for the technological mastery of body, disorder, and disease.

Better tools for predicting disease risk are the promised goods of the genetic revolution, and Americans want them, was a message that filtered through the testimonies at the SACGHS hearings. Those Americans who want these tools appear to be educated consumers who have a family history of genetic disease. But this message ignores the wants and needs of the "have-nots:" those individuals who lack the cultural and intellectual capital to be skilled health consumers, or those who have no interest in practicing risk-assessment. Those individuals are not demanding greater access to diagnostic tools, or legal protection from genetic discrimination. They are not a constituency that matters in a nation that valorizes taking

responsibility for one's genetic liabilities—except as potential subjects for research and clinical trials.

In their work on genetic citizenship, anthropologists Deborah Heath, Karen-Sue Taussig and Rayna Rapp have argued that the activism of genetic patient interest groups like the Little People of America and organizations like PXE International has afforded members of the genetics community greater input into the research enterprise. My SACGHS fieldwork suggests that some members of this genetics community are promoting a coercive and divisive form of good citizenship in the guise of expanding civil rights, by emphasizing the obligations of all Americans to act on their supposed genetic liabilities.

The genomic citizenship construct has an important function: it directs critical attention away from the socio-economic inequities that structure access to the health care system (including genetic testing) and health outcomes, towards the alarming prospect that the engines of scientific progress will be slowed by widespread fears of losing one's health insurance coverage. It conveniently displaces the structural and logistical problems of delivering genetic testing to the populace within the current health care system that the Committee itself identified. Calls for Congress to pass a law banning genetic discrimination rest on an appeal to American egalitarianism, yet they valorize the good genomic citizen: those Americans who are willing to take full responsibility for their genetic liabilities and enlist for duty in genomic research. What this egalitarianism disguises are class differences that shape not only which Americans can afford to undergo genetic testing and follow-up treatments, but which

Americans value the pursuit of good health (cf. Crawford 2006) and the right to make genetic choices (cf. Kerr 2003b). Such calls sidestep the work required to improve access to basic health care services for the many disenfranchised Americans.

To return to Margaret Lock's (2005b:S66) question about whether genetic determinism still holds sway over public discourse, even as researchers embrace the study of epigenetics and reject the genotype/phenotype dogma: there were no environmental or epigenetic discourses operating here, unless we count the oft-articulated moral imperative of caring for oneself by monitoring diet, exercise, and lifestyle. The "belief in a technologically assisted future of bodily mastery" that Lock (2005b:S66) queries not only remains in plain sight in genomics discourse in the public policy setting in this country, it shows no signs of relinquishing its hold in public understandings of health and disease. Genetic divination appears to be rooted in the American imaginary as an essential risk management tool, as indispensable as hurricane forecasting.

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[1] Written testimony of Fran Visco, President, National Breast Cancer Coalition, to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[2] Written testimony of Sharon F. Terry to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[3] Testimony by Phaedra Malatek to SACGHS, October 18, 2004.

[4] Testimony by Paula Funk to SACGHS, October 18, 2004.

[5] Testimony by Rebecca Fisher to SACGHS, October 18, 2004.

[6] The publication of Rachel Carson's (1962) *Silent Spring* on the dangers of pesticides spawned a heightened awareness of environmental toxicity and a generation of environmental and feminist health activists. This wave of feminist and environmental health activism produced the Boston Women's Health Book Collective (1973) and their pioneering *Our Bodies, Ourselves*, educating women about the role of environmental exposures to carcinogenic agents and some medications (for example, first generation oral contraceptives) in an increased incidence of reproductive cancers. Heightened concern about the relationship between carcinogens and environmental health hazards continued into the late 1970s when the Love Canal saga unfolded as a testament to the health hazards of building a residential community on a toxic waste site. Media coverage of the Love Canal episode framed it not as a story about the unusual genetic predispositions of residents in the Love Canal community to cancer, asthma, and birth defects, but about the liability of a manufacturing company for health problems decades after operation, and the wisdom of building a residential community and schools on a chemical waste disposal site. Love Canal is a residential community in New York State were Hooker Chemical and Plastics Corporation had buried 21,000 tons of chemical waste in the 1940s. The discovery of this history and high rates of illness made daily news in 1978.

[7] See "Betty's Story in 2015," slides 53-59 (Collins 2007).

[8] Testimony by Rebecca Fisher to SACGHS, October 18, 2004.

[9] Comments by Francis Collins, SACGHS hearings, October 18, 2004.

[10] The Rehabilitation Act of 1973 and the Americans with Disabilities Act (ADA) of 1990 in principle protects employers against obesity discrimination but only if plaintiffs can demonstrate they are disabled. One state (Michigan, under the Elliott Larsen Civil Rights Act of 1976) and three cities (San Francisco, Washington DC, and Santa Cruz, California) have enacted anti-obesity discrimination statutes.

[11] In my reading of their work, Rapp and Ginsburg appear to be advancing an understanding of the disability rights movements as a singular community of people with shared interests, rather than a diverse community of dozens of organizations with competing interests.

[12] Again, I acknowledge, as I did in Chapter 8, that there is no single community of the disabled.

[13] The NHGRI released the following comments by Francis Collins on Senate passage of the Genetic Information Nondiscrimination Act of 2005 (S. 306):

Since the completion of the Human Genome Project in 2003, we have seen a crescendo of advances in medical research. We have seen the development of new diagnostic tests, preventive strategies, and treatments for genetically based diseases. I am concerned, however, that this progress will be greatly hampered if the American people do not feel comfortable obtaining genetic information about themselves. S. 306 will clearly protect all

of us from genetic discrimination in health insurance and employment, and would free the American people from the fear of such discrimination. (Comments by Francis S. Collins to the Senate on the Passage of Genetic Information Nondiscrimination Act of 2005 (S. 306), February 17, 2005. Electronic document, http://www.genome.gov/13014311, accessed August 16, 2008)

See also comments by Francis Collins to the U.S. Senate in 2004, where he made a similar statement on the Genetic Information Nondiscrimination Act of 2003 (S. 1053):

The National Human Genome Research Institute and the National Institutes of Health have been working on the issue of genetic discrimination since the very beginning of the Human Genome Project. … If this bill doesn't pass, my concern is that we won't be able to realize the full potential of advances in genetic science, because people will be afraid to participate in clinical trials or obtain genetic tests out of fear of discrimination. Most people have not yet had a genetic test, so the opportunity for genetic discrimination has not occurred in most people's lives. But as genetic tests become more widespread, the risk will be quite real. (Comments by Francis Collins, Director, National Human Genome Research Institute, Regarding Genetic Nondiscrimination, April 1, 2004. Electronic document, http://www.genome.gov/11511396, accessed August 16, 2008)

[14] Source: "Future Directions in Genetic and Genomic Research." PowerPoint presentation by Francis Collins to SACGHS, June 11, 2003, Washington, DC." Electronic document, http://www4.od.nih.gov/oba/SACGHS/meetings/June2003/Presentations/Collins_s.pdf, accessed October 30, 2008.

[15] See Chapter 7 for a survey of similar statements made by genetic advocates.

[16] Written testimony of Sharon F. Terry to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[17] The NHGRI also promotes patient and clinician responsibility for genetic predisposition to disease in its presentations about personalized medicine to professional organizations. When Francis Collins shows "Betty's Story" in his PowerPoint presentations on the future of personalized medicine to the Personalized Medicine Coalition and the Physician Assistant Education Association, he also shows a slide sequence called "Betty's story gone wrong." In this sequence, which begins with a slide labelled, "Personalized Medicine: Could the dream become a nightmare?" Betty does not participate in the Surgeon General's Family Health Initiative, and does not bring a family pedigree to her clinician. Her clinician, therefore, never learns about her family history of heart disease. Betty also declines to undergo whole-genome sequencing, even though it is available in the year 2015, because no genetic nondiscrimination law was passed and Betty's brother lost his health insurance after undergoing genetic testing. Betty then "eats an unhealthy diet, gains weight, and develops high blood pressure." When her clinician treats her with a drug for hypertension, she develops a hypersensitivity reaction and discontinues its

use. (Apparently in 2015, there are no other medications to treat hypertension). With her hypertension uncontrolled for ten years, Betty then develops pain in her left arm at age 50, which her clinician misdiagnoses as muscular, and she dies of cardiogenic shock. See “Betty’s story gone wrong,” slides 56-61 (Collins 2007).

Conclusion

There are several stories I could have told that would have produced a narrow exploration of genetic biopolitics in the United States. I could have told a privacy story about how genetic activists and legal scholars have made a compelling case for genetic information as uniquely personal and private information that requires federal protection. Alternatively, I could have told a legal activism story that situates GINA in the history of civil rights activism in the United States. Or, I could have told a health reform story that portrays genetic discrimination as part of the long history of dissatisfaction with managed care, and GINA as an incremental health reform measure. All of these stories would fall short, because they would not be able to explain why contemporary discourse on genetic discrimination is tied to a political economy of hope in genomics, whose proponents insist that genetic testing unequivocally saves lives and that all Americans are genetic subjects. Nor could these stories explain why contemporary discourse on genetic discrimination turns on the language of shared genetic defect, and why this language has ramifications for all Americans.

All of these stories would miss this one observation: that there is no single problem called "genetic discrimination." Rather, there has been a set of actors over the last twenty years, with different interests and concerns, who have shaped public concern about problems tied to genetic screening and testing. Genetic discrimination looks like a different problem, depending on who has championed it.

The biography of genetic discrimination in the United States showcases what historian Allan Brandt (Brandt and Gardner 2000:711) describes as "a particularly American fascination with scientific and technical remedies

for complex social problems as an approach to reform." To expand on Brandt's observation, what has animated the biography of genetic discrimination from the 1970s through the present is a technology fetishism tied to a national imaginary of scientific innovation. This fetishism simultaneously makes genetic technologies appear to be the source of their own transformative value, while disguising the power relations and human labour that shape how these technologies are used (cf. Marx 1867). The dismay and betrayal that greet institutions that subvert the "inherent" good of these technologies for their own benefit are expressions of this fetishism, as is the reification of a disordered genome that no one, apparently, can escape.

The biography of genetic discrimination describes not only different ideas about the merits and dangers of genetic screening and testing. It also describes changing notions of which populations are at risk of discrimination, and what forms discrimination takes. We could argue that the stigmatization and employment discrimination of African-American sickle cell carriers in the 1970s was the first widespread instance of genetic discrimination. But such a public consensus never developed. As Troy Duster (1990) has argued, in the 1970s and 1980s, genetic screening and testing developed around (and targeted) ethnic populations and specific diseases. Thus, early on, genetic screening and testing fragmented the public health model and thwarted any understanding of sickle-cell discrimination as a society-wide problem that could be labelled "genetic" discrimination.[1]

New York Times reporter Richard Severo and the now-defunct Congressional Office of Technology Assessment forged the first public

awareness that the scope of discriminatory practices was wider than the sickle cell screening programmes of the 1970s. Following Severo's series (Severo 1980a, 1980b, 1980c, 1980d, 1980e, 1980f, 1981a, 1981b) in the *New York Times* on hypersusceptibility screening at DuPont and other chemical and manufacturing companies, the OTA surveyed employers in 1982 (U.S. Congress, Office of Technology Assessment 1983) to determine the extent of this practice. Severo, and later, sociologist Elaine Draper (1991, 1993), interpreted workplace screening as a form of ethnic and racial screening. However, the authors of the OTA report determined that employers were screening all applicants for certain genotypes, not just members of minority groups. The OTA report authors singled out "genetic makeup" as the basis for discrimination, and not race or ethnicity. Their language was important, because it identified workers with no visible markers of difference (such as skin colour or religious affiliation) as the population at risk for discrimination. The OTA report authors were also the first to explore the question of whether workers who experienced discrimination based on their genetic makeup were protected under existing laws, including disability statutes.

Less than ten years later, geneticist Neil Holtzman (1989) and sociologist Troy Duster (1990) published the first comprehensive analyses of the profound social impacts of genetic screening and testing practices. Their concerns that the rapid expansion of genetic screening and testing practices would create a "genetic underclass" (Duster 1990) were part of broader warnings they delivered about a revival of eugenic imperatives. Without using the term "genetic discrimination," Holtzman's (1989) *Proceed with Caution* seeded the ground for reception to the arguments

and evidence that Paul Billings and members of the Genetic Screening Study Group would present in 1992 (Billings et al 1992).

Yet while Neil Holtzman and Troy Duster shared the same concerns that enthusiasm for genetic screening and testing was a revival of eugenics, the departure points for their concerns were different. Richard Severo's reporting on workplace screening was a departure point for Holtzman's concern about discrimination. In fact, occupational screening remained a lifelong interest for Holtzman. His work to reform Maryland state law and efforts to assess the proficiency of PKU screening laboratories were also influential in drawing his attention to problems associated with genetic screening. But it was at the OTA, where he sat in on meetings about insurance denials to HIV-positive individuals, that he consolidated his observations into the first draft of *Proceed with Caution*. Troy Duster's concern about sickle cell discrimination, which was not shared by Holtzman, developed into a broader concern about government surveillance and control of genetic information when the DOE-NIH began talks in 1986, and when the Department of Defense began its DNA Registry in 1992. For Duster, the key questions have been, "Who is going to control the collected information, and how are they going to use it?" Duster also insisted, more than anyone else, that genetic discrimination is not one problem but many, and that it becomes salient whenever an institution has the power to collect and control the genetic information of individuals.

Genetic discrimination became a discrete and recognizable problem in the genetics community in 1992 when the Genetic Screening Study Group published its survey results and case reports of institutional discrimination

in the *American Journal of Human Genetics* (Billings et al 1992). Their "naming and framing" (Brown 1995) of genetic discrimination provided a set of diagnostic criteria that formalized the problem and identified the "asymptomatic ill" as the vulnerable population. This publication attracted media attention, spawned debate amongst geneticists, and helped to secure a permanent place for genetic discrimination as a policy issue on the agenda of the ELSI Working Group. The publication translated the concerns of members of the Genetic Screening Study Group about the revival of eugenic thinking in genetic research and social policies into something more specific. This "something more specific" was unfair and prejudicial practices by health insurers, as well as employers and life insurers, against otherwise healthy individuals who were known to have genetic markers for diseases. This framing of what they perceived as the problem moved discrimination away from its earlier associations with workplace genetic screening and race-based discrimination against minorities, and narrowed its scope from the broader ambit of eugenic practices that both Neil Holtzman (1989) and Troy Duster (1990) had outlined.

Yet only a few years after the Genetic Screening Study Group defined genetic discrimination as a discrete problem of health insurance for individuals with rare, single-gene disorders, new actors came on board to champion and reframe the issue. The rest of the decade and the beginning of the twenty-first century saw the Hereditary Susceptibility Working Group, the National Breast Cancer Coalition, the Genetic Alliance, and the Coalition for Genetic Fairness work with the NCHGR and the NHGRI to shape public awareness of genetic discrimination as a serious problem for

the nation. These actors shaped a new understanding of vulnerable Americans to include not only healthy carriers of mutations for rare disorders, but healthy individuals with susceptibility genes for common disorders—in other words, all Americans. Using the language of genetic defect to argue that all Americans were vulnerable to discrimination, these actors insisted in 1997 that "[e]ach of us has an estimated five to 30 serious misspellings or alterations in our DNA,"[2] in 2003, that "[a]ll of us carry dozens of glitches in our DNA sequence" (Collins and Watson 2003:745), in 2004, that "every one of us is estimated to be genetically predisposed to between 5 and 50 serious disorders,"[3] and also in 2004, that "[w]e all possess mutations that will become equally and increasingly transparent with tomorrow's technologies."[4] They also framed genetic discrimination as a civil rights problem (see Collins and Watson 2003).

The testimonies on discrimination at the SACGHS hearings illustrate how the scope of the problem continues to be defined. The "victims of discrimination" who presented their testimonies at the SACGHS hearings on October 18th, 2004, were both individuals carrying the rare mutations that directly cause serious, sometimes lethal, diseases, and individuals with susceptibility genes, which may or may not play a role in their family histories of breast and ovarian cancer. It is clear from these testimonies that the scope of genetic discrimination has expanded, to include punitive action by insurers and employers against individuals who have used the knowledge of their susceptibility genes they are carrying and their family histories of disease to take pre-emptive action against the onset of disease.

The eugenicist critique that was central to Neil Holtzman's and Troy Duster's commentaries on the expansion of genetic screening and the

commercialization of genetic testing has all but disappeared from contemporary discourse on genetic discrimination. Advocates of federal nondiscrimination legislation, who are committed to building a nation that continues with genomic research and delivers personalized medicine, express an unbridled optimism in the power of genetic testing to predict disease and save lives, and a genetic inevitabilism that the mutations that all Americans carry will become transparent over time. In championing the right of American to consume the promised goods of the Human Genome Project without fearing discrimination by insurers or employers, advocates geneticize all Americans by enrolling them into the biosociality of the inherently flawed and transparent genome. What these advocates do not also champion is the right of Americans to refuse to think or act genetically.

If genomic citizenship describes the expansion of civil rights protection for all Americans against discrimination based on their genotypes, while increasing their duties to pre-emptive care and make themselves bioavailable, what will this mean for those Americans who have not yet realized that "[g]enetics is about ALL of us?"[5] [emphasis in the original] In the remainder of this Conclusion, I imagine some of the implications of genomic citizenship, and outline directions for future research.

A Bioavailable Population?

In Chapter 1, I stated that the need for a bioavailable population for genomics research and clinical trials is a key driver of genetic discrimination activism. Both the NHGRI and the Genetic Alliance (the latter representing hundreds of single-gene patient groups as well as

organizations like the NBCC) have an investment in encouraging Americans to make their bodies and medical histories available, for clinical trials of pharmacogenomic medicines and large-scale genomics studies.

Since 1992, genetic discrimination has been framed as a problem of discrimination by insurers and employers. But in their efforts to see Congress pass a comprehensive federal genetic nondiscrimination law to keep genetic test results out of the hands of insurers and employers, genetic discrimination activists have not addressed a related concern: the potential reluctance of some Americans to share their tissue samples and medical histories with a government agency, even the NHGRI, even under the protection of GINA. The ambitions of the NHGRI to enrol up at least 500,000 Americans into a long-term population study open up other areas of ethical concern beyond insurer and employer discrimination. Advocates of nondiscrimination legislation have argued that a federal genetic nondiscrimination law will protect the genetic privacy of individuals, allowing them to feel safe enough to undergo genetic testing and participate in studies and trials. And a recent electronic survey of American adults indicates that a majority of those surveyed support the study and would be willing to participate in the study by supplying DNA and tissue samples (Kaufman et al 2008). But what measures will the NHGRI take to address concerns by participants that arise before, during, and after the study, about the collection, control, circulation, and use of genetic material?

Here it is helpful to return to Margaret Everett's (2007) discussion of personhood dilemmas, which I reviewed in Chapter 1. A request by a researcher to send the tissue samples from Everett's deceased newborn,

Jack, to an Italian laboratory and beyond, prompted moral and emotional wrangling in Everett and her husband. They anguished about whether they had an obligation to share Jack's tissue with researchers and aid research into a rare disease, about what part of Jack was in his cells, and about whether they were inappropriately keeping Jack "alive" long after his death by allowing researchers to transform his cells into immortalized cell lines that could travel around the world from lab to lab. "Some of us, myself included," Everett (2007:384) writes, "may be especially aware of the place of genes (as thing, symbol, and idea) in our biographies, but we are all struggling to figure out what part of ourselves is our genes and vice versa." Everett's poignant story illustrates how a tissue sample from a family member can acquire layers of meaning and value that may be completely at odds with the regimes of value in the world of researchers.

The reservations that Everett and her husband shared, over whether to permit tissue samples from their deceased son to circulate anywhere without their knowledge or control, are not isolated. Nor are they unique to them. This is not the place to review the large literature discussing property rights concerns in the human body, or claims to extended personhood in human genetic material.[6] But if recent debates about property rights and personhood dilemmas regarding alienated tissues (including DNA) are any indication, along with controversies over federal government collection and control of DNA samples, the NHGRI's proposal to collect tissue samples from some 500,000 Americans for a large-scale gene-environment study may prompt similar dilemmas for participants. What measures will the NHGRI take to convince Americans to share their tissue samples and medical histories with researchers who want them?

What controls, if any, will these research subjects have over the circulation and use of their biomaterials? Will families be permitted to withdraw tissue samples and medical histories of deceased relatives from researchers, once collected?

If the NHGRI wins Congressional approval and funding for its study, what incentives and public health appeals will the Institute and its partner agencies use to recruit Americans, particularly African-Americans, into the study? More broadly, how will the NHGRI and other agencies encourage Americans to become consumers of personalized medicine and participate in clinical trials of pharmacogenomic drugs? GINA, I have suggested, is one tool of this citizenship project. Another tool is the U.S. Surgeon General's Family Health Initiative, which is teaching Americans to think genetically through the use of a family health pedigree and public health posters.[7] What other public health initiatives will the NHGRI and other institutes with a commitment to genomics, such as the CDC and the Surgeon General's Office, launch to teach Americans to think and act genetically?

The Survival of Single-Gene Patient Interest Groups

Earlier, I observed that it has been a small group of elites that has spearheaded efforts to have Congress pass a federal law banning genetic discrimination, and to keep the issue at the top of the ELSI policy agenda. The Genetic Alliance, which represents 600 single-gene disorder patient groups, is one of these actors. However, while the Genetic Alliance is a key ally of the NHGRI, it is unclear what relationship the patient support groups that it represents have with federal agencies that control the purse-

strings of much of the genomics research that takes place in the United States. For this reason, the conclusions of Heath, Taussig and Rapp about the reach of genetic citizenship need to be revisited with further research that considers the relationships of these patient organizations to the NHGRI.

How do patient groups position themselves with respect to research and funding priorities at the NHGRI and other institutes with genome programmes? Research to investigate this question must acknowledge the power relations that have shaped genomics research and policy in the United States for the last fifteen years—specifically, that small groups of tightly-networked elites have managed genomics policy. In light of these concentrated power relations, do the genetic citizenship activities of patient interest groups truly open up "democratic possibilities" (Taussig et al 2003:62) for themselves and other populations?

I wonder, too, about the prospects for long-term survival of the hundreds of disease groups that advocate for patients and families with rare inherited diseases. Although the Genetic Alliance has formed a temporary alliance with the NHGRI to pursue its own interests (and those it shares with the NHGRI), I am sceptical of the extent of influence and control that patient interest groups have over the directions that genomics research takes. Single-gene disorder groups and organizations risk being marginalized by the NHGRI's personalized medicine platform, and by the interest among genomics researchers in studying the epigenetics of common disorders. The emphasis of genomics research—and personalized medicine—is on diagnosing, preventing, and treating common disorders, not rare diseases. The fifty-eight diseases and traits that researchers have

investigated through genome-wide association studies (GWAS) as of July 2008 are mostly common degenerative disorders or traits, not rare disorders (Manolio 2008).[8] Given that this is the direction in which genomic research is heading, what will happen to the interests of the single-gene disorder community and research into their "orphan" genes?

Accommodating Disability in a Genomic Nation

The vision of Americans eagerly peering into their own genomes to discover if they are susceptible to Alzheimer's disease, colon cancer, diabetes, or schizophrenia is at odds with a society that readily accommodates disability. The rallying cry behind the momentum to pass a federal nondiscrimination law, that everyone has flawed genes, generates not one but two disturbing visions. It suggests that disease awaits us all. And it hints at a lifetime of disability for some. Behind the curtain of the flawed genome lies an aversion not only to aging and infirmity, but also to disability and its costs.

The NHGRI vision of personalized medicine is rooted in an ideal of the perfectible, disease-free body (Nelkin and Lindee 1997). What this ideal and its hopeful vision obscure are the politics of abortion and the history of eugenics in the United States.[9] Certainly, some genetic interest groups that have embraced the revolution in genetics also worry about what impact this research will have on their survival. For example, in their research on the Little People of America (Rapp et al 1998, 2001), Heath, Taussig, and Rapp reported that following the isolation of achondroplasia gene causing dwarfism in 1994, some members of the Little People of America appeared at an annual meeting wearing t-shirts labelled

"endangered species." These members were making the point that their survival was threatened by the very tools that offered their community some hope of intervening into their conditions, specifically, the prospect of using prenatal diagnosis of achondroplasia and choosing whether to bring a fetus to term that is determined to carry the gene.

This story is a useful reminder of the eugenic potential of genetic diagnosis. It offers some idea of which populations might fear the introduction of diagnosis and screening technologies, and why. But we need to look beyond the survival concerns of patients with rare disorders, to eugenic pressures on an entire nation. When the same actors who promote personalized medicine and whole-genome sequencing also push for the passage of a law to remove barriers to wider participation in genetic testing and genomic research, what kind of society are they promoting?

At the SACGHS hearings, there were no discussions about how personalized medicine will intersect with reproductive decision-making, particularly abortion and the avoidance of disability. The interventions that Francis Collins has described in his presentations about personalized medicine are non-reproductive. They consist of whole-genome sequencing, preventive lifestyle changes, and pharmacogenomic interventions, all tailored to the "individual" profile. Yet the seven individuals who testified to the Committee made it abundantly clear that decisions they faced about whether to undergo genetic testing or seek prophylactic surgery were not individual decisions. These were decisions that rebounded across the extended family. It would be naïve to think reproductive decision-making would not be one of the conversations that members of the extended family would have with each other in a health care system where whole-

genome sequencing is a norm of clinical practice.

What was missing from these discussions at the SACGHS hearings was any mention of the reproductive pressures that inevitably accompany the introduction of new genomic screening technologies and the expansion of target disorders. For some idea of what these pressures might be, we need only revisit Neil Holtzman's (1989) analysis of the social impacts of expanding genetic screening and testing practices to include disorders that lack treatment. One of the reproductive pressures that Holtzman identifies in association with screening and testing for such disorders is the pressure to abort a fetus that is found to carry a genetic anomaly (however we define a genetic anomaly). Another is the pressure not to reproduce (cf. Holtzman 1989).

We need not entertain the prospect of compulsory sterilization to argue that new screening technologies and new disease targets continually revitalize a persistent eugenicist ethos that has never disappeared from the United States. As Rapp (1988) and others (see, for example, Hubbard 1985 and Nelkin 1996) have observed, the assumption that some lives are not worth living is built into reproductive practices such as prenatal diagnosis. Patients undergoing "nondirective" reproductive counseling experience pressure—sometimes subtle, sometimes not—to make certain choices (for example, to abort a fetus with a chromosomal anomaly such as Down syndrome). Outside of the reproductive diagnostic setting, political and economic priorities also constrain individual reproductive choices (Hubbard 1985). How free are families to bring an "abnormal" fetus to term, if there is no postnatal support (such as special education) for raising a disabled child, or if a health insurer refuses to cover costly

lifelong treatments for a disabled child?

The prospect of using whole-genome sequencing to peer inside our genomes and detect susceptibility genes for the dozens of disorders that we all presumably carry has the potential to dramatically expand the category of intolerable disability. What new calculi of human worth will whole-genome sequencing bring when it becomes integrated into clinical care as a norm of preventive medicine? When this technology becomes available for $1,000 or less, and price is no longer an objection for most people, who will refuse to have their personal genomes sequenced, for fear of being labelled genetically irresponsible, except the poor and the uninsured? On what shrinking moral ground will defiant or recalcitrant Americans reside, those who refuse to learn their risks, or those who insist on reproducing their defective genes and bearing children who may become disabled or "prone" to disease (cf. Hubbard 1985)? And in a country where abortion is not a choice for many, and where a well-organized anti-abortion movement has succeeded in reducing or removing access to abortion in many states, what reproductive choices will there be for women and families who know that they carry susceptibility genes for diseases such as breast cancer or schizophrenia? Will they feel pressured not to reproduce, in order not to pass on their "flawed" genes, even though susceptibility genes do not cause disease?

Whether or not whole-genome sequencing will have any predictive value, will families that carry susceptibility genes for schizophrenia, bipolar disorder, or Alzheimer's disease feel entirely free to reproduce? When high-penetrance mutations for lethal single-gene disorders are confused with low-penetrance and non-lethal susceptibility genes in public

discourse about genomics, as they have been at the SACGHS hearings, will families "diagnosed" with susceptibility genes consider themselves to be genetically flawed? These reproductive pressures, which lie in the shadows of discussions about the rights of Americans to seek genetic testing and the promise of personalized medicine to predict and prevent degenerative diseases, comprise the wider ambit of genetic discrimination. They are part of a much longer history in the United States of coercive pressures that coalesce each time a new genetic screening practice is routinized, or a new population is targeted for genetic screening. But these reproductive pressures, and the wide scope of genetic discrimination that they comprise, were not part of the nation's conversation with itself at the SACGHS hearings.

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[1] Duster explains:

> Sickle-cell was perceived as a problem only by a small percentage of the black community, and grew into a larger socio-political issue because of the times. But notice that the scale is much greater when we talk about insurance, and privacy, and the worksite. These three penetrate through the whole population, and are not ethnic or racial specific. Indeed, one of the arguments that I made in *Backdoor* was that genetic diseases and screening and testing fragmented the traditional public health model. With tuberculosis, smallpox, and so on, we are all vulnerable, so one can make a strong general public health appeal. With Tay Sachs limited to Ashkenazic Jews, cystic fibrosis to northern Europeans, Beta thalessemia to the Mediterranean population, and so on, you can see the point about the fracture of the public health consensus." (Troy Duster, personal communication, January 16, 2008)

[2] Source: "Preventing Genetic Discrimination in Health Insurance." Statement of Francis S. Collins to Congressional Task Force on Health Records and Genetic Privacy, July 22, 1997. http://www.genome.gov/10002352, accessed August 16, 2008.

[3] Written testimony of Louise M. Slaughter to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[4] Written testimony of Sharon F. Terry to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[5] Written testimony of Sharon F. Terry to SACGHS, "Public Perspectives on Genetic Discrimination: September 2004-November 2004," Secretary's Advisory Committee on Genetics, Health, and Society, U.S. Department of Health and Human Services.

[6] The last twenty years has seen increasing commentary and concern about the fragmentation of the body through biomedical and reproductive technologies and the alienation and circulation of body parts and products (for example, ova, sperm, genes, blood, organs, tissue samples, and genes) for sale, exchange, research, patenting, or conversion into research tools. Much of this commentary has fallen under the rubric of "commodification of the body." These concerns have developed at the intersection of (1) the rapid commercialization of biological research since the 1970s (see, for example, Krimsky 1999), (2) the proliferation of technologies that alienate body parts and their products, such as human tissues, immortalized cell lines, and genetic samples, for use as research tools or for profit, and (3) the liberalization of the U.S. patenting law that has encouraged the patenting of immortalized cell lines, genes, and genetic fragments. For representative commentary, see Andrews and Nelkin 2001, Kimbrell 1993, Lock 2001, Scheper-Hughes 2001, and Sharp 2000. As well, medical anthropologists have documented experiences of extended personhood in organs amongst both family members of organ donors and transplant recipients (see, for example, Lock 2002b and Sharp 1995).

[7] The Family Health Initiative offers an online, fillable family pedigree called "My Family Health Portrait." (See discussion of My Family Health Portrait in Chapter 7). Users are prompted to enter their family histories of health problems and diseases and bring their completed Family Health Portraits to their clinicians to discuss how to predict and prevent disease. The promotional component of My Family Health Portrait includes public service posters that target breast cancer, colorectal cancer, heart disease, and diabetes. One of the goals of the Initiative, according to the Surgeon General's Office, is to "[p]repare the public and health professionals for the coming era in which genomics will be an integral part of regular health care" (U.S. Department of Health and Human Services 2008b). For an overview of the NHGRI's educational goals to integrate genomics into health care, see Guttmacher et al 2007.

[8] The genome-wide association study (GWAS) is a new tool in genomics that builds on the mapping and sequencing of the human genome, and the HapMap project. These studies scan markers across the entire human genome, using two populations for comparison: those with a target disease, and those without the disease. Variations that appear more frequently in those with the disease are then associated with the disease. These variations are not necessarily causal; they simply point to regions of the genome associated with the disease.

The diseases and traits examined through GWAS include cancers (lung, prostate, breast, colon, as well as melanoma), autoimmune disorders (for example, rheumatoid arthritis and systemic lupus erethematosus), circulatory diseases, height, and mental illness and neurological disorders. The latter category includes Alzheimer's disorder, bipolar disorder, schizophrenia, neuroticism, and something called "family chaos" which is related to "environmental confusion in the home" (see Butcher and Plomin 2008).

[9] Notably, the preamble of HR 493, the bill that passed into law in May 2008 as GINA, includes a paragraph that mentions the eugenics movement in U.S. history (see Appendix C, Preamble, HR 493).

Appendix A.

Data Collection and Analysis

SACGHS FIELDWORK (JUNE 2005 – NOVEMBER 2007)

Data Collection

The SACGHS hearings were held two or three times each year for two consecutive days each, except for the July 10, 2007 hearings, which lasted for one day. Between June 2003 and November 2007, SACGHS held fourteen hearings. I attended six of these hearings in-person: June 15-16, 2005 (North Bethesda, MD); October 19-20, 2005 (North Bethesda, MD); March 27-28, 2006 (Bethesda, MD); June 26-27, 2006 (Bethesda, MD); November 13-14, 2006 (College Park, MD); and March 26-27, 2007 (Adelphi, MD). For the eight hearings I did not attend between June 2003 and November 2007, I reviewed the webcasts, transcripts, and supporting materials (PowerPoint presentations and other documents) associated with each of the hearings.

I collected or made use of six types of data generated by these hearings:

1. Field notes based on in-person observations of each of the six hearings that I attended, and on my conversations with attendees and participants at the SACGHS hearings.
2. Email correspondence with administrative staff, committee members, and attendees and participants at the SACGHS hearings.
3. Written documents produced or made available by SACGHS (e.g. meetings agendas, roster, mandate, written testimony to the committee, reports, recommendations, correspondence between SACGHS and officials, and copies of participants' presentations). I obtained many of these as hard copies at the SACGHS hearings and printed others directly off the NIH Office of Biotechnology Activities (OBA) website.
4. PowerPoint presentations shown at the hearings. There are archived on the OBA website as "supporting materials" by hearing date.

5. Verbatim transcripts of twelve of the fourteen hearings between June 2003 and November 2007, in Adobe Portable Document Format (.pdf), totalling 3,221 pages. (Transcripts from the hearings on July 10, 2007 and November 19-20, 2007 are not available on the OBA website, but the webcast and supporting materials, such as PowerPoint presentations are).
6. Webcasts of each of the fourteen hearings in Real Audio Metadata (.ram) format.

Other primary data external to the SACGHS hearings were also important to my analysis. These include:

7. Verbatim transcripts from the fifteen meetings of the Secretary's Advisory Committee on Genetic Testing (SACGT) from 1999 to 2002.
8. Archived speeches, press releases, policy statements, and annual budget statements of the NHGRI and the Department of Energy (available on their websites).
9. Archived speeches, press releases, and policy statements of the Genetic Alliance, Coalition for Genetic Fairness, National Breast Cancer Coalition, Council for Responsible Genetics, National Partnership for Women and Families, and American Civil Liberties Association (available on their websites).

Data Analysis

My analysis of the SACGHS hearings took place over five years (2004-2008), most of it prior to my chapter writing. The transcripts, webcasts, and PowerPoint presentations from the SACGHS hearings were my primary sources of data, along with my field notes from the six hearings that I attended. From June 2003 to June 2005, I reviewed the transcripts from the SACGT hearings and the first two hearings of SACGHS. In addition to reading through the transcripts, I also conducted keyword searches of the documents using Adobe Acrobat Reader, to search for every reference to "consumer," "American," "DTC," "direct-to-consumer"

and "regulation."

By June 2005, when my focus had shifted from the DTC genetic testing industry to genetic discrimination, I was already familiar with the first two years of the SACGHS hearings from reading the transcripts and watching the webcasts. In addition to conducting keyword searches on comments at the hearings related to genetic discrimination, I began to use the hearings materials in a wheel-and-spoke fashion. I identified actors who made statements on genetic discrimination, personalized medicine, Americans as particular kinds of subjects, and genomics and the nation. I tracked their speeches, presentations, and affiliations outside of the SACGHS hearings, from 1990 to 2008 (as appropriate). I also tracked these actors across the hearings, noting the dates of their appearances, the subjects of their comments, who they exchanged comments with, and which organizations (if any) they represented. I compared the comments they made, to see if their language had changed. This analysis generated knowledge about these actors' networks and their patterns of language use over time and place.

I often analyzed the transcripts and webcasts of the hearings together. For example, I would watch part of a webcast to identify the emotions and nuances in individual speech delivery, the tone of the Committee's discussion, or the dynamics between individual speakers, which were invisible in the transcripts. Towards the end of my SACGHS fieldwork, as I collected different types of data, I moved back and forth between these different data more often, using them to verify and expand on my observations and conclusions.

Appendix A. Data Collection and Analysis

ARCHIVAL RESEARCH AND INTERVIEWS (JANUARY 2007 – MAY 2008)

Data Collection

My goal in conducting archival research was to locate all public commentary between 1963 (the year that Massachusetts mandated newborn screening) and 1992 where the expressions "discrimination," "genetic discrimination," and "stigmatization" were used with respect to genetic screening and testing practices. A second goal was to identify the actor(s) who coined the expression "genetic discrimination."

I began by following the citations in the Billings et al (1992) *American Journal of Human Genetics* article to locate prior discourses about discrimination, to understand the particular events that had shaped the authors' concerns, and to locate the origins of the expression. I then conducted a search of the *New York Times*, *Hastings Center Report*, and *American Journal of Human Genetics* from 1963 to 1992, using electronic and physical archives, to locate these commentaries. I focussed initially on the history of the Guthrie test and its implementation, but when commentary on this screening practice failed to turn up uses of the terms "discrimination" or "genetic discrimination," I focussed on debates about sickle-cell screening and then Richard Severo's commentaries on the *New York Times*. Severo's articles in turn led me to the 1983 OTA report. In addition, I used the PubMed and MedLine databases to conduct keyword searches for these terms.

While I was conducting archival research, I began to conduct oral histories with Phil Bereano, Jonathan Beckwith, Paul Billings, Troy Duster, Neil Holtzman, and Sheldon Krimsky. I conducted two of these interviews entirely in-person (Duster and Holtzman), and three entirely by

telephone (Beckwith, Bereano, and Krimsky). My interview with Paul Billings began as an in-person interview on one date, but was interrupted. It continued as a telephone interview on another date. These interviews lasted between 60 and 90 minutes each, and I digitally recorded each, with the subject's consent. While conducting these interviews, I asked these subjects questions about the practices and events I was tracking in my archival research, and their networks and associations. Specifically, I asked each what events had galvanized their concern. However, I also solicited their comments about sickle cell screening in the 1970s, and Richard Severo's series in the *New York Times*, asking specifically whether either of these had been influential. These questions often generated new departure points that I had not considered or known about (for example, insurer discrimination against individuals with AIDS, or the DoD DNA Registry). These departure points, in turn, provided me with more practices to investigate through archival research.

I also interviewed Amanda Sarata, a former employee of the NIH Office of Biotechnology Activities and a SACGHS staffer who co-ordinated the "Perspectives on Genetic Discrimination" session. This was an in-person, semi-structured interview that lasted 60 minutes. Sarata did not consent to digital recording of the interview, but did consent to me taking notes. I later verified the accuracy of my notes with her.

Data Analysis

I transcribed each of the interviews myself, highlighting sections that were relevant to the archival research I was conducting and the arguments I was making. These interviews did not produce enough biographical

information about the subject on their own. For example, dates and places were frequently missing. To "fill in" this missing information, I drew on articles about the subjects and published interviews with them, or contacted them directly to clarify.

In contrast to the analysis for Part Two of the dissertation, which I did before I wrote the dissertation, the analysis for Part One of the dissertation occurred while I wrote Chapters 4 and 5. I synthesized my archival research with my interview findings to produce a narrative—which I then rejected. I repeated this process over and over, until I had produced a narrative that made sense of my findings.

Appendix B.

Summary of H.R. 493, the Genetic Information Nondiscrimination Act of 2008

Appendix B. H.R. 493, the Genetic Information Nondiscrimination Act of 2008, Congressional Research Service Summary

Source: "H.R. 493: Genetic Information Nondiscrimination Act of 2008." GovTrack.us. H.R. 493--110th Congress (2007): Genetic Information Nondiscrimination Act of 2008. Electronic document, http://www.govtrack.us/congress/bill.xpd?bill=h110-493&tab=summary, accessed November 27, 2008.

5/21/2008--Public Law.

(This measure has not been amended since it was passed by the Senate on April 24, 2008. The summary of that version is repeated here, with changes reflecting enrollment corrections.)

Genetic Information Nondiscrimination Act of 2008 -

Title I - Genetic Nondiscrimination in Health Insurance

Section 101 -

Amends the Employee Retirement Income Security Act of 1974 (ERISA), the Public Health Service Act (PHSA), and the Internal Revenue Code to prohibit a group health plan from adjusting premium or contribution amounts for a group on the basis of genetic information.

Prohibits a group health plan from requesting or requiring an individual or family member of an individual from undergoing a genetic test. Provides that such prohibition does not: (1) limit the authority of a health care professional to request an individual to undergo a genetic test; or (2) preclude a group health plan from obtaining or using the results of a genetic test in making a determination regarding payment. Requires the plan to request only the minimum amount of information necessary to accomplish the intended purpose.

Allows a group health plan to request, but not require, a participant or beneficiary to undergo a genetic test for research purposes if certain requirements are met, including: (1) the plan clearly indicates that compliance is voluntary and that noncompliance will have no effect on enrollment status or premium or contribution amounts; (2) no genetic information collected or acquired is used for underwriting purposes; and (3) the plan notifies the Secretary of Health and Human Services that it is conducting activities pursuant to this exception and includes a description of the activities.

Prohibits a group health plan from requesting, requiring, or purchasing genetic information: (1) for underwriting purposes; or (2) with respect to any individual prior to such individual's enrollment in connection with such enrollment (provides that incidentally obtains such information is not a violation).

Applies such prohibitions to all group health plans, including small group health plans.

Provides that any reference to genetic information concerning an individual or family member includes genetic information of: (1) a fetus carried by a pregnant woman; and (2) an embryo legally held by an individual or family member utilizing an assisted reproductive technology.

Authorizes a penalty against any sponsor of a group health plan for any failure to meet requirements of this Act. Allows a waiver or limitation on such penalty if the failure was not discovered after exercising reasonable diligence or was due to reasonable cause.

Section 102 -

Amends the PHSA to prohibit: (1) a health insurance issuer offering health insurance coverage in the individual market from establishing eligibility rules for enrollment based on genetic information; (2) discrimination on the basis of genetic information for health insurance offered in the individual market in the same manner as such discrimination is prohibited for group coverage; and (3) the imposition by a health insurance issuer offering health insurance coverage in the individual market of a preexisting condition exclusion on the basis of genetic information.

Applies such requirements to nonfederal governmental plans.

Section 104 -

Amends title XVIII (Medicare) of the Social Security Act (SSA) to prohibit an issuer of a Medicare supplemental policy, on the basis of genetic information, from: (1) denying or conditioning the issuance or effectiveness of the policy, including the imposition of any exclusion of benefits based on a preexisting condition; or (2) discriminating in the pricing of the policy, including the adjustment of premium rates.

Prohibits an issuer of a Medicare supplemental policy from: (1) requesting or requiring an individual or a family member to undergo a genetic test; or (2) requesting, requiring, or purchasing genetic information for underwriting purposes or for any individual prior to enrollment.

Section 105 -

Amends title XI (General Provisions, Peer Review, and Administrative Simplification) of SSA to require the Secretary of Health and Human Services to revise Health Insurance Portability and Accountability Act of 1996 (HIPAA) privacy regulations to: (1) treat genetic information as health information; and (2) prohibit the use or disclosure by a group health plan, health insurance coverage, or Medicare supplemental policy of genetic information about an individual for underwriting purposes.

Section 106 -

Requires the Secretaries of Health and Human Services, Labor, and the Treasury to ensure that their regulations, rulings, and interpretations under this title are administered so as to have the same effect at all times and that they adopt a coordinated enforcement strategy.

Title II - Prohibiting Employment Discrimination on the Basis of Genetic Information

Section 202 -

Prohibits, as an unlawful employment practice, an employer, employment agency, labor organization, or joint labor-management committee from discriminating against an employee, individual, or member because of genetic information, including: (1) for an employer, by failing to hire or discharging an employee or otherwise discriminating against an employee with respect to the compensation, terms, conditions, or privileges of employment; (2) for an employment agency, by failing or refusing to refer an individual for employment; (3) for a labor organization, by excluding or expelling a member from the organization; (4) for an employment agency, labor organization, or joint labor-management committee, by causing or attempting to cause an employer to discriminate against a member in violation of this Act; or (5) for an employer, labor organization, or joint labor-management committee, by discriminating against an individual in admission to, or employment in, any program established to provide apprenticeships or other training or retraining.

Prohibits, as an unlawful employment practice, an employer, employment agency, labor organization, or joint labor-management committee from limiting, segregating, or classifying employees, individuals, or members because of genetic information in any way that would deprive or tend to deprive such individuals of employment opportunities or otherwise adversely affect their status as employees.

Prohibits, as an unlawful employment practice, an employer, employment agency, labor organization, or joint labor-management committee from requesting, requiring, or purchasing an employee's genetic information, except for certain purposes, which include where: (1) such information is requested or required to comply with certification requirements of family and medical leave laws; (2) the information involved is to be used for genetic monitoring of the biological effects of toxic substances in the workplace; and (3) the employer conducts DNA analysis for law enforcement purposes as a forensic laboratory or for purposes of human remains identification.

Section 206 -

Requires an employer, employment agency, labor organization, or joint labor-management committee that possesses any genetic information about an employee or member to maintain such information in separate files and treat such information as a confidential medical record.

Prohibits an employer, employment agency, labor organization, or joint labor-management committee from disclosing such genetic information, except: (1) to the employee or member upon request; (2) to an occupational or other health researcher; (3) in response to a court order; (4) to a government official investigating compliance with this Act if the information is relevant to the investigation; (5) in connection with the employee's compliance with the certification provisions of the Family and Medical Leave Act of 1993 or such requirements under state family and medical leave laws; or (6) to a public health agency.

Section 207 -

Sets forth provisions regarding enforcement of this Act.

Section 208 -

Provides that disparate impact on the basis of genetic information does not establish a cause of action under this Act.

Establishes the Genetic Nondiscrimination Study Commission six years after enactment of this Act to review the developing science of genetics and to make recommendations to Congress regarding whether to provide a disparate impact cause of action under this Act. Authorizes appropriations to the Equal Employment Opportunity Commission (EEOC) to carry out this section.

Section 212 -

Authorizes appropriations.

Title III - Miscellaneous Provisions

Section 301 -

Provides that if any provision of this Act, an amendment made by this Act, or the application of such provision or amendment to any person or circumstance is held to be unconstitutional, the remainder of this Act shall not be affected.

Section 302 -

Amends the Fair Labor Standards Act of 1938 to increase the maximum employer penalty for violations involving oppressive child labor provisions or certain child labor safety requirements. Establishes an additional civil penalty for any such violation that causes the death or serious injury of an

employee under the age of 18, which may be doubled for a repeated or willful violation.

Defines "serious injury" as: (1) permanent loss or substantial impairment of one of the senses or of the function of a bodily member, organ, or mental faculty; or (2) permanent paralysis or substantial impairment that causes loss of movement or mobility of a body part.

Increases the maximum civil penalty for any repeated or willful violation of minimum wage or maximum hours requirements.

Appendix C.

Preamble, H.R. 493, the Genetic Information Nondiscrimination Act of 2008

H. R. 493

One Hundred Tenth Congress of the United States of America

AT THE SECOND SESSION

Begun and held at the City of Washington on Thursday, the third day of January, two thousand and eight

An Act

To prohibit discrimination on the basis of genetic information with respect to health insurance and employment.

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

SECTION 1. SHORT TITLE; TABLE OF CONTENTS.

(a) SHORT TITLE.—This Act may be cited as the "Genetic Information Nondiscrimination Act of 2008".

(b) TABLE OF CONTENTS.—The table of contents of this Act is as follows:

SEC. 2. FINDINGS.

Congress makes the following findings:

(1) Deciphering the sequence of the human genome and other advances in genetics open major new opportunities for medical progress. New knowledge about the genetic basis of illness will allow for earlier detection of illnesses, often before symptoms have begun. Genetic testing can allow individuals to take steps to reduce the likelihood that they will contract

a particular disorder. New knowledge about genetics may allow for the development of better therapies that are more effective against disease or have fewer side effects than current treatments. These advances give rise to the potential misuse of genetic information to discriminate in health insurance and employment.

(2) The early science of genetics became the basis of State laws that provided for the sterilization of persons having presumed genetic "defects" such as mental retardation, mental disease, epilepsy, blindness, and hearing loss, among other conditions. The first sterilization law was enacted in the State of Indiana in 1907. By 1981, a majority of States adopted sterilization laws to "correct" apparent genetic traits or tendencies. Many of these State laws have since been repealed, and many have been modified to include essential constitutional requirements of due process and equal protection. However, the current explosion in the science of genetics, and the history of sterilization laws by the States based on early genetic science, compels Congressional action in this area.

(3) Although genes are facially neutral markers, many genetic conditions and disorders are associated with particular racial and ethnic groups and gender. Because some genetic traits are most prevalent in particular groups, members of a particular group may be stigmatized or discriminated against as a result of that genetic information. This form of discrimination was evident in the 1970s, which saw the advent of programs to screen and identify carriers of sickle cell anemia, a disease which afflicts African-Americans. Once again, State legislatures began to enact discriminatory laws in the area, and in the early 1970s began mandating genetic screening of all African Americans for sickle cell anemia, leading to discrimination and unnecessary fear. To alleviate some of this stigma, Congress in 1972 passed the National Sickle Cell Anemia Control Act, which withholds Federal funding from States unless sickle cell testing is voluntary.

(4) Congress has been informed of examples of genetic discrimination in the workplace. These include the use of preemployment genetic screening at Lawrence Berkeley Laboratory, which led to a court decision in favor of the employees in that case Norman-Bloodsaw v. Lawrence Berkeley Laboratory (135 F.3d 1260, 1269 (9th Cir. 1998)). Congress clearly has a compelling public interest in relieving the fear of discrimination and in prohibiting its actual practice in employment and health insurance.

(5) Federal law addressing genetic discrimination in health insurance and employment is incomplete in both the scope and depth of its protections. Moreover, while many States have enacted some type of genetic non-discrimination law, these laws vary widely with respect to their approach, application, and level of protection. Congress has collected substantial evidence that the American public and the medical community find the existing patchwork of State and Federal laws to be confusing and inadequate to protect them from discrimination. Therefore Federal legislation establishing a national and uniform basic standard is necessary to fully protect the public from discrimination and allay their concerns about the potential

for discrimination, thereby allowing individuals to take advantage of genetic testing, technologies, research, and new therapies.

TITLE I—GENETIC NONDISCRIMINATION IN HEALTH INSURANCE

SEC. 101. AMENDMENTS TO EMPLOYEE RETIREMENT INCOME SECURITY ACT OF 1974.

(a) NO DISCRIMINATION IN GROUP PREMIUMS BASED ON GENETIC INFORMATION.—Section 702(b) of the Employee Retirement Income Security Act of 1974 (29 U.S.C. 1182(b)) is amended—

(1) in paragraph (2)(A), by inserting before the semicolon the following: "except as provided in paragraph (3)"; and

(2) by adding at the end the following:

"(3) NO GROUP-BASED DISCRIMINATION ON BASIS OF GENETIC INFORMATION.—

"(A) IN GENERAL.—For purposes of this section, a group health plan, and a health insurance issuer offering group health insurance coverage in connection with a group health plan, may not adjust premium or contribution amounts for the group covered under such plan on the basis of genetic information.

"(B) RULE OF CONSTRUCTION.—Nothing in subparagraph (A) or in paragraphs (1) and (2) of subsection (d) shall be construed to limit the ability of a health insurance issuer offering health insurance coverage in connection with a group health plan to increase the premium for an employer based on the manifestation of a disease or disorder of an individual who is enrolled in the plan. In such case, the manifestation of a disease or disorder in one individual cannot also be used as genetic information about other group members and to further increase the premium for the employer.".

(b) LIMITATIONS ON GENETIC TESTING; PROHIBITION ON COLLECTION OF GENETIC INFORMATION; APPLICATION TO ALL PLANS.—Section 702 of the Employee Retirement Income Security Act of 1974 (29 U.S.C. 1182) is amended by adding at the end the following:

"(c) GENETIC TESTING.—

"(1) LIMITATION ON REQUESTING OR REQUIRING GENETIC TESTING.—A group health plan, and a health insurance issuer offering health insurance coverage in connection with a group health plan, shall not request or require an individual or a family member of such individual to undergo a genetic test.

"(2) RULE OF CONSTRUCTION.—Paragraph (1) shall not be construed to limit the authority of a health care professional who is providing health care services to an individual to request that such individual undergo a genetic test.

"(3) RULE OF CONSTRUCTION REGARDING PAYMENT.—

"(A) IN GENERAL.—Nothing in paragraph (1) shall be construed to preclude a group health plan, or a health insurance issuer offering health insurance coverage in connection with a group health plan, from obtaining and using the results of a genetic test in making a determination regarding payment (as such term is defined for the purposes of applying the regulations promulgated by the

Appendix D.

Charter of the Secretary's Advisory Committee on Genetics, Health and Society (2004)

National Institutes of Health
Bethesda, Maryland 20892
www.nih.gov

CHARTER

SECRETARY'S ADVISORY COMMITTEE ON GENETICS, HEALTH, AND SOCIETY

Purpose

Considering that extraordinary scientific advances in biology and human genetics, including the sequencing of the human genome, are speeding the development of new technologies to prevent, treat, and cure disease; that these technologies also have nonmedical applications in areas such as education, employment and law; and that the technologies have substantial benefits, but also the potential to be applied in ways that can be harmful to individuals or society, the Secretary's Advisory Committee on Genetics, Health, and Society (Committee) is established to: (1) provide a forum for expert discussion and deliberation and the formulation of advice and recommendations on the range of complex and sensitive medical, ethical, legal and social issues raised by new technological developments in human genetics; (2) assist the Department of Health and Human Services and, at their request, other Federal agencies in exploring issues raised by the development and application of genetic technologies; and (3) make recommendations to the Secretary of Health and Human Services (Secretary) concerning how these issues should be addressed.

Authority

42 U.S.C. 217a, section 222 of the Public Health Service (PHS) Act, as amended. The Committee is governed by the provisions of the Federal Advisory Committee Act, as amended (5 U.S.C. Appendix 2), which sets forth standards for the formation and use of advisory committees.

Function

The Committee shall explore, analyze and deliberate on the broad range of human health and societal issues raised by the development and use, as well as potential misuse, of genetic technologies and make recommendations to the Secretary, and other entities as appropriate. The scope of the Committee's charge includes assessing how genetic technologies are being integrated into health care and public health; studying the clinical, ethical, legal and societal implications of new medical applications, such as preimplantation genetic diagnosis, and emerging technological approaches to clinical testing; identifying opportunities and gaps in research and data collection efforts; exploring the potential misuse of genetics in bioterrorism; examining current patent policy and licensing practices for their impact on access to genetic

technologies; analyzing uses of genetic information in education, employment, insurance; including health, disability, long-term care, and life, and law, including family, immigration, and forensics; and serving as a public forum for discussion of emerging scientific, ethical, legal and social issues raised by genetic technologies.

The Committee may call upon special consultants, assemble ad hoc working groups, and convene conferences and workshops, as necessary, to assist in the work of the Committee.

Structure

The Committee shall consist of 13 members, including the Chair, appointed by the Secretary, or designee, from authorities knowledgeable about molecular biology, human genetics, health care, public health, bioterrorism, ethics, forensics, law, psychology, social sciences, education, occupational health, insurance, and other relevant fields. Of the appointed members, at least two members shall be specifically selected for their knowledge of consumer issues and concerns and the views and perspectives of the general public.

The following Federal officials, or their designees, shall serve as nonvoting ex officio members of the Committee: Assistant Secretary for Children and Families; Assistant Secretary for Health; Administrator, Agency for Healthcare Research and Quality; Director, Centers for Disease Control and Prevention; Administrator, Centers for Medicare & Medicaid Services; Commissioner of Food and Drugs; Administrator, Health Resources and Services Administration; Director, National Institutes of Health; Director, Office for Civil Rights; Director, Office for Human Research Protections; Attorney General of the United States; Secretary of Commerce; Secretary of Defense; Secretary of Education; Secretary of Energy; Secretary of Labor; Secretary of Veterans Affairs; Chair, Equal Employment Opportunity Commission; Chairman, Federal Trade Commission; and any other officers or employees of the United States, as the Secretary determines are necessary for the Committee to effectively carry out its function.

Members may be invited to serve for overlapping terms of up to four years; terms of more than two years are contingent upon the renewal of the Committee by appropriate action prior to its termination. Members may serve after the expiration of their term until their successors have taken office. A member who has been appointed for a term of four years may not be reappointed to this Committee before two years from the date of expiration of that term of office. Any member appointed to fill a vacancy occurring prior to the expiration of the term for which that member's predecessor was appointed, shall serve for the remainder of that term; persons appointed to complete an unexpired term of less than four years may be reappointed immediately for a full term following completion of the unexpired term.

A quorum for the conduct of business by the full committee shall be a majority of the appointed members. A quorum for each subcommittee shall be three members.

As necessary, standing and ad hoc subcommittees may be established to perform specific functions within the Committee's jurisdiction. The Department Committee Management Officer

will be notified upon establishment of each standing subcommittee and will be given information on its name, membership, function, and estimated frequency of meetings.
Coordination of agenda development and management and operational services shall be provided for the Committee by the Office of Science Policy, Office of the Director, National Institutes of Health.

Meetings

Meetings shall be held not less than two times a year at the call of the Chair, with the advanced approval of a Government official who shall also approve the agenda. A Government official shall be present at all meetings.

Meetings shall be open to the public except as determined otherwise by the Secretary. Notice of all meetings shall be given to the public.

Meetings shall be conducted and records of the proceedings kept, as required by applicable laws and Departmental policies.

Compensation

Members shall be paid at the rate of $200 per day, plus per diem and travel expenses, as authorized by section 5703, Title 5 U.S.C. as amended, for persons in the Government service employed intermittently. Members who are officers or employees of the United States Government shall not receive compensation for service on the Committee.

Annual Cost Estimate

The estimated annual cost for operating the Committee, including compensation and travel expenses for members, but excluding staff support, is $332,244. The estimated annual person-years of staff support required is 3.1, at an estimated annual cost of $297,735.

Reports

In the event a portion of a meeting is closed to the public, an annual report shall be prepared which shall contain, at a minimum, a list of members and their business addresses, the Committee's functions, dates, places of meetings, and a summary of committee activities and recommendations made during the fiscal year. A copy of the report shall be provided to the Department Committee Management Officer.

Termination Date

Unless renewed by appropriate action prior to its expiration, the Charter for the Secretary's Advisory Committee on Genetics, Health, and Society will expire on September 23, 2006.

Approved:

JUL 2 2 2004
Date

Director, NIH

Appendix E.

SACGHS Roster, 2005

CHAIR

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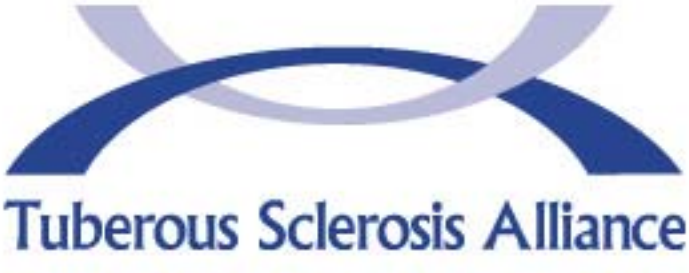
Appendix F.

SACGHS Call for Public Commentary on Genetic Discrimination



TSC ALERT

Edited by Vicky Holets Whittemore, Ph.D. & Cheryl Dunigan, Ph.D.

August 2004

Welcome to the August 2004 edition of *TSC Alert* – an online research newsletter for individuals interested in Tuberous Sclerosis Complex (TSC) research and clinical care. This online newsletter contains information of interest to the TSC research and health care community. Please forward this newsletter to colleagues who are interested in TSC. To be added/deleted to/from the mailing list for *TSC Alert* and/or to submit information for the September 2004 *TSC Alert* contact: Vicky.Whittemore@tsalliance.org

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[Clicking on one of the headings takes you directly to that section of TSC Alert]

IMPORTANT DEADLINES:

AUGUST 31, 2004: Deadline for registration to attend International TSC Conference in Cambridge, UK in September 2004. See information below under Conferences.

NIH ANNOUNCEMENTS:

The Secretary's Advisory Committee on Genetics, Health, and Society Hearing on Genetic Discrimination, October 18, 2004 The issue of genetic discrimination is a high priority for the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS). In an effort to raise awareness about the problem, the Committee is seeking public comments from individuals who:

- Have experienced genetic discrimination in health insurance or in employment.
- Fear the potential impact of genetic discrimination on either health insurance or employment.
- Have paid out-of-pocket for services to exclude genetic information from medical records.
- Are health care providers and have had patients experience genetic discrimination; express concern about genetic discrimination; or taken steps to avoid genetic discrimination (for example, not undergoing genetic testing or keeping the results out of a medical record).

SACGHS will be holding a hearing on October 18, 2004 to gather information from members of the public about the scope and nature of genetic discrimination. The Committee is particularly interested in learning about cases of genetic discrimination that are based on predictive genetic information, pre-symptomatic genetic disease, or carrier status.

In October 2003, the Senate unanimously passed the Genetic Information Nondiscrimination Act, and advocates are pressing for action in the House of Representatives. In July 2004, the House Subcommittee on Employer-Employee Relations of the Committee on Education and the Workforce held a hearing on the issue of genetic discrimination. In spite of broad bipartisan support for the legislation, there is opposition and it appears to be preventing further progress in the House. The fear of genetic discrimination and its adverse consequences is well-documented and regarded by many as sufficient justification for Federal legislation. However, opponents argue that there is insufficient evidence that genetic discrimination is occurring and thus legislation is not warranted at this time. SACGHS hopes that the information gathered during the hearing will help address the concerns of the bill's opponents.

SACGHS was established to serve as a forum for deliberation on the ethical, legal and social issues at the intersection of genetics, health and society and to advise the Secretary of Health and Human Services about these issues. For more information about the Committee and its meetings, as well as copies of the Committee's correspondence with the Secretary on this issue, please visit http://www4.od.nih.gov/oba/SACGHS.HTM

Please send your written comments to SACGHS by **September 17, 2004** in care of Amanda Sarata at sarataa@od.nih.gov or by fax to 301-496-9839.

Amanda Sarata, M.S., M.P.H., Secretary's Advisory Committee on Genetics, Health, and Society, 6705 Rockledge Drive, Suite 750, Bethesda, MD 20892, 301-496-7009 (ph), 301-496-9839 (fax)

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