Genetic Disorder as Metaphor and the Construction of the Abnormal Self

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Abstract

Narrative ethics is particularly important in examining and improving the experience of patients with genetic disorders. Such an exercise includes careful consideration of the figures of speech that influence people's beliefs and actions, including the metaphors which have arisen to help convey the semi-abstract nature of the gene. This thesis examines the development of genetic metaphors over the course of the 20th century, noting the significance of particular representations of genetics like those of virus, language, lottery and soul. Based on these findings, metaphors of genetic disorder are characterized into the opposing conceptions of "genetic disorder versus self" as independent entities and "genetic disorder as self" as a characteristic of one's identity. Each of these categories of metaphor carries its own unique bioethical consequences. Finally, this thesis discusses recommendations for influencing the future development of these metaphors and the practicality of such plans.

Résumé

L'éthique narrative est particulièrement importante dans l'examen et l'amélioration des expériences des patients avec des troubles génétiques. Cette examen doit inclure une étude attentive des figures de style qui influence les croyances et les actions, comme les métaphores qui aident à exprimer la nature abstraite des gènes. Cette thèse examine le développement des métaphores génétiques au cours du 20ème siècle, en notant l'importance des représentations particulières de la génétique comme ceux de virus, de langue, de loterie et de l'âme. Selon ces résultats, les métaphores de maladies génétiques sont caractérisées par les conceptions opposées de «trouble génétique par rapport à soi-même» comme des entités indépendantes et «trouble génétique comme soi-même» comme une caractéristique identitaire. Ces deux grandes catégories de métaphore entraîner des conséquences différentes pour la bioéthique. Enfin, cette thèse traite de recommandations préexistantes pour influencer le développement futur de ces métaphores génétiques et la faisabilité de ces changements.

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The initial idea for this project came to me this summer while I was in New Haven attending the lovely Summer Institute held by Yale's Interdisciplinary Center for Bioethics. One of the teachers had a large personal collection of bioethics-related books, which students were allowed to sign out. After perusing the shelves and reading Karen Joy Fowler's *We Are All Completely Beside Ourselves* and Albert Camus' *The Plague*, I noticed a copy of Susan Sontag's *Illness as Metaphor*. Her essay describes how illnesses of unknown origin have historically been understood through metaphor, and how that can have detrimental effects on our beliefs and behaviours. As a student of human genetics with an academic background in English literature, I naturally wondered whether a similar analysis had ever been written on genetic disorders. That question led to the formulation of this thesis.

Introduction

The central question this project asks is "How are genetic disorders represented in metaphor, and what are the bioethical consequences of such representations?" In order to set the stage for this discussion, I must begin with a brief overview of why we should be concerned with the functions of metaphor and their implications for the practice of medical genetics.

The first use of metaphor in biology has been dated back to Alcmaeon of Croton around 500 BC (Ouzounis & Mazière, 2006), while the earliest known philosophical study of metaphor comes down to us from Aristotle and his late fourth-century books *Poetics* and *Rhetoric* (Condit et al., 2002). Aristotle claimed that metaphors fostered thought, stating "It is a great thing, indeed, to make proper use of the poetic forms . . . But the greatest thing by far is to be a master of metaphor" (*Rhetoric* 1410b, quoted in Lakoff & Johnson, 1980, p. 190). However, Aristotle's equally influential teacher Plato argued that metaphor was misleading, as did the English philosophers Thomas Hobbes and John Locke two thousand years later. The study of metaphor was eventually reinvigorated by the Romantic literary movement in the early 1800s, sparked partly by poets such as Wordsworth and Coleridge (Lakoff & Johnson, 1980, p. 190-192).

Metaphors are often articulated through narrative; and like narrative they are both rooted in everyday experiences and involved in bringing meaning to those experiences (Kay, 2000, p. 22; Calsamiglia & Van Dijk 2004; Lakoff & Johsnon, 1980; Rommetveit, Scully & Porz, 2013). Mere "decorative" metaphors remain useful in rhetoric – and, indeed, poetry – as a way of adding variety to writing, but they can also profoundly influence our thought structures (Ivie, 1987; Condit & Condit, 2001; Condit et al., 2002) by bringing different domains of human experience into each others' context (Van Dijck, 1998, p. 23: Liakopoulos, 2002; see also Ortony, 1993). In creating connections between these areas and spurring our curiosity (Avise, 2001), they generate new meanings and understandings (Lakoff & Johnson, 1980, p. 139).

Although metaphors are now seen as crucial for discourse, there are many conflicting theories about how exactly they function (Condit et al. 2002). There is even disagreement about whether all language is metaphorical in some sense, conveying associations rather than concretely describing objects themselves (Condit et al. 2002; Baake, 2003, p. 60, 111; Sidler, 2006; Pramling & Säljö, 2007).

Based on the research conducted for this thesis, the work which has most influenced the literature of genetic disorder is Lakoff and Johnson's seminal 1980 book *Metaphors We Live By*, which defines metaphor as "understanding and experiencing one kind of thing in terms of another" (p. 5). The conceptual metaphor theory that Lakoff and Johnson (1980) advance is that, in their own words: "Our ordinary conceptual system, in terms of which we both think and act, is fundamentally metaphorical in nature" (p. 3). Their rationale is that we understand abstract or unfamiliar concepts by transferring personal experience in a "source domain" to construct the cognitive structures needed to create meaning in the "target domain" which we attempt to describe or understand. This process plays an essential role in structuring perception, thought and action alike, thus "determining what is real for us" (Lakoff & Johnson, 1980, p. 146). In fact, they claim that "Metaphors as linguistic expressions are possible precisely because there are metaphors in a person's conceptual system" (Lakoff & Johnson, 1980, p. 6).

In addition to being shaped by our experiences in the culture that surrounds us (Lakoff & Johnson, 1980, p. 19, 57), Lakoff and Johnson (1980) further argue that our conceptual structures are based on embodiment - that is to say, the experience of existing as a body in space and interacting with a physical environment. For instance, they argue that up is fundamentally seen as associated with good, and down with bad, partly because "serious illness forces us to lie down physically" (Lakoff & Johnson 1980, p. 14-15, 25; Lopez, 2007).[1]

The focus on embodiment as human experience runs contrary to many of the traditions of Western biomedical thinking, particularly with respect to the separation of self and body (Sharp, 2000; Everett, 2003; Gronnvoll & Landau, 2010). This dualism is largely attributable to the 17th-century French philosopher René Descartes and his reductionistic philosophy (Lippman, 1992; Vannatta & Vannatta, 2013).[2] Reductionism can be understood as the paradigm in which things are viewed with respect to their constituent parts rather than as wholes (Hubbard & Wald, 1993, p. 3).

[1] These sorts of associations can have practical uses; one study, for example, found that teaching people about genetic risk with visual reference to an elevator was more effective than using a bridge in the same way, possibly because risk visuals tend to assume the former shape but possibly because people naturally think of elevated risk levels on a vertical rather than a horizontal axis (Kaphingst et al., 2009).

[2] Lakoff and Johnson (1980) note that although Cartesian coordinates (one of the other great contributions for which Descartes is known, along with the line "I think, therefore I am") describe locations in space, they don't come with orientations and therefore cannot readily be used to represent a body in space as one experiences it (p. 56)

As Lewontin (1991) puts it, ever since Descartes' *Discourses* introduced the powerful organizing metaphor of the clockwork mechanism, "the individual bits and pieces, the atoms, molecules, cells and genes, are the causes of the properties of the whole objects and must be separately studied if we are to understand complex nature" (p.12). Descartes' distinction between cause and effect likewise diminished the role of the environment in favour of individual subunits (Lewontin, 1991, p. 12-13). Thus, historically speaking, Western models of science have promoted "biomedical" over "biopsychosocial" explanations of disease (e.g. Hull, 1978), form over matter and mind over increasingly mechanized body (Strathern, 1996, p. 5; Gannett, 1999; Pickering, 1999).

In her influential book *The Meaning of Illness*, Toombs (1992) wrote that the patient should be seen as equivalent to the body and not as its possessor (p. 81). By examining the experiences and meanings of illness as articulated through "qualitative, affective, and thick" narrative, physicians can improve their ability to interpret symptoms for diagnosis, choose the correct treatment to alleviate suffering, and come to a common understanding with their patients as persons (Toombs, 1992, p. 42-43, 46, 87-89; Nowaczyk, 2012; Vannatta & Vannatta, 2013). Narrative ethics is especially important in illuminating problems within the context of nuanced lives and relationships (Churchill, 2002, p. 183), and these methods are necessary in medicine because of "the overreaching of the scientific understanding into the realm of interpersonal experiences" (Vannatta & Vannatta, 2013).

However, those scientific understandings and processes are often informed by models based on metaphor (Pickering, 1999; Pramling & Säljö, 2007). Copland (2005), for instance, argues that metaphors are particularly useful in science because they are "naturally compatible with . . . reductionist methodology". While scientists are often perceived to use metaphors as heuristic tools for simply transferring meaning, these rhetorical techniques actually generate interactions and displacements of meaning that help develop new concepts and push science forward (Doyle, 1994, p. 58; Lopez, 2007).

On the other hand, one school of thought holds that, in a tradition not unlike Plato and his successors, metaphors are deceptive and therefore incompatible with the "rational enterprise" of science (Pickering, 1999). The limits of metaphor go hand in hand with their generative ability (Porta, 2003). Unlike an analogy, which infers a one-to-one correspondence between separate cases with known similarities from the same general domain of experience, a metaphor is not

exclusive, imaginatively generates new similarities and can be used to highlight specific features for comparison (Nordgren, 1998; Barry, Brescoll, Brownell & Schlesinger, 2009). Van Dijck (1998) writes that while neither metaphor nor analogy can provide the full account of a topic, at least "scientific models are based on verification and logical consistency [while] metaphors are often intentionally imperfect" (p. 22).

As much as they highlight some facets of the domain in question, metaphors obscure other ways of thinking, even within scientific disciplines (Lakoff & Johnson, 1980, p. 10, 52-53; Sherwin, 1999; Condit & Condit, 2001; Ceccarelli, 2004; Weigmann, 2004). As such, they naturally suggest their own truthfulness (Lakoff & Johnson, 1980, p. 157). Lakoff and Johnson (1980) wrote that: "the people who get to impose their metaphors on the culture get to define what we consider to be . . . absolutely and objectively true" (p. 160). By sanctioning action, metaphors can shift focus to certain realities and therefore act as a self-fulfilling prophecies (Condit & Condit, 2001; Gronnvoll & Landau, 2010). Metaphors influence scientists, administrators, and funders, thus affecting the kind of evidence researchers look for (Fox Keller, 1995, p. 21, 35) and directing study away from other directions (Avise, 2001). They can also take on associations unintended by their authors (Weigmann, 2004; Sidler, 2006).

Since the body is itself the instrument of perception, it has been argued that we tend to develop images of our bodies as other things through metaphor (Rommetveit et al., 2013), and this includes our illnesses. As Sontag argued in *Illness as Metaphor* (1978), "Any important disease whose causality is murky, and for which treatment is ineffectual, tends to be awash in significance" (p. 58), making it more susceptible to metaphorical interpretation. This certainly applies to the general category of genetic disorders. While it is true that scientists have identified the etiology of thousands of single-gene disorders, there remains something about the human genome itself that prevents it from being widely comprehensible. Lakoff and Johnson (1980) argued that "vaguer" concepts lend themselves to "more concrete" metaphors, as those provide a vantage point within our actual experiences that can be used to structure our thoughts about a less familiar topic (p. 112). Indeed, metaphors have been considered essential in helping laypersons to visualize and ultimately comprehend complex concepts like the human genome, which most people cannot interact with directly (Petersen, 2001; Gronnvoll & Landau, 2010). This can give people "at least ... the illusion" of understanding genetics (Van Dijck, 1998, p. 22) and help individuals create new "moral order" in their lives (Rommetveit et al., 2013).

In the context of medical conditions which are treated scientifically but popularly interpreted as a part of the humanities' domain, an approach to ethics which balances empirical facts with experiences and values may be particularly warranted (Vannatta & Vannatta, 2013). Narrative is also useful in examining the practices used in bioethics itself (Churchill, 2002, p. 183). For instance, metaphors can encourage creativity and make moral understandings more accessible to stakeholders in bioethical issues (Sherwin, 1999). Indeed, fundamental bioethical concepts like utility, integrity, care, and the principles of autonomy and justice, have been argued to be rooted in metaphor and shaped by the metaphors and analogies we use to understand them (Nordgren, 1998; Sherwin, 1999).[3] In the afterword added to the 2003 edition of their book, Lakoff and Johnson even claimed that science had shown "our basic understanding of morality arises via conceptual metaphor" (p. 250).

Any thorough approach to biomedical ethics should therefore take into account the importance of figurative language and its role in both shaping and sharing people's beliefs and behaviours in response to certain forms of illness. As will be argued in later sections of this thesis, metaphors appear to be particularly important in defining, depicting and even experiencing genetic disorder. However, very little of the huge amount of academic literature that has been written on the rhetoric of genetics or the human genome in general has focused specifically on genetic disorders and how they are depicted as a category of illness based on shared type of causality. While a number of individual papers and book chapters have been written on this topic, there does not seem to have been a work of any significant length integrating the existing data, theory and historical information into a unified account of the topic.

Greater understanding of how figurative language both indicates and affects how we think about diseases of genetic origin could make a very useful contribution to the practice of narrative ethics. Therefore, I decided that the best approach to this unmet need would be to begin with a complete literature review of the existing publications which had discussed genetic disorder in conjunction with metaphor. This would allow me to establish the common themes of these metaphors, identify how they have been treated in academia thus far, and integrate these findings

[3] Nordgren argues that moral situations are framed metaphorically, and we use our imagination in narrative context to extend our moral oncepts to nonprototypical cases. He describes autonomy as a metaphor from politics dating back to Greek city-states, utility as a commercial transaction, and justice as a variety of metaphors including commercial transaction, balance, and proportion. Metaphors of medicine as a whole include war, business, machine repair, system maintenance, and care in its personal sense.

into a context-based theory about the fundamental attributes they share and how these underlying conceptions might affect patients' self-concept in practice.

By restricting this analysis to peer-reviewed, academic publications, I made sure that my sources were structured and verifiable. While a qualitative study such as a survey or series of roundtables would also have been very productive, but I believe the themes elucidated by the current project can also provide the basis for a more informed set of questions for that sort of follow up research. I believe the current project, while complete in itself, offers results useful for research in several fields of inquiry related to inherited diseases and traits, including the following example.

One area where these results have relevance is that of rare disease, which are in many jurisdictions defined as affecting fewer than 1 in 2000 people. The collective burden of these illnesses is nevertheless quite important, as there are about 7000 known rare diseases and as many as 1 in 12 Canadians may be affected by them (Melnikova, 2012). Yet whereas relatively little has been written specifically about genetic disorder in metaphor, there do not appear to have been any similar studies of the narrative aspects of rare disease.

There are several reasons for which this line of inquiry bears relevance to rare diseases. Firstly, both represent specific categories of illness with both medical and social connotations, and as 80% of rare diseases are genetic disorders (Melnikova, 2012), many of the findings of this thesis likely apply to those patients as well, especially with respect to the biomedical aspects discussed and their effects on many of the psychosocial effects of disease.

Secondly, products for the treatment of these diseases are called orphan drugs due to their limited market size. They are an important issue in health policy for Canada and other countries with single-payer health care systems due to their expensiveness and high opportunity. The term "orphan" is itself a metaphor, and further analysis of how this sort of rhetoric is developed and articulated by stakeholders such as the government and the pharmaceutical industry should prove revealing.

Thirdly, narratives appear to be especially important for patients with rare diseases. By definition they are already have exceptional medical experiences, and the rare disease community is heavily involved in advocacy on behalf of a constituency they feel have been politically overlooked. It could also be very rewarding to examine how the abstract idea of rarity is both presented to the general public and interpreted by them, especially as their willingness to

pay for expensive treatments is the theoretical basis of our health care system's reimbursement policy.

For these reasons, rare disease could be an ideal topic to pursue further in my future research. In order to accommodate these applications, I have mentioned rarity wherever that theme emerged in the course of this literature review, according to the procedure outlined below.

Methodology

As one of my goals was to analyze the development of metaphors for genetic disorders over the course of the 20th century, I did not place any restrictions upon the publication dates of documents being included in the analysis, provided they were peer-reviewed, published in recognized academic sources, and available in English. Academic interest in the Human Genome Project produced a surge in publications on the cultural interpretation and significance of human genetics during the 1990s, and so it would have been very counterproductive to have excluded these books and articles from the study based on their date.

I located publications which fit these criteria by conducting keyword searches in the databases EthicShare, EthxWeb, PubMed, Web of Science, PhilPapers and JSTOR, as well as the research engine Google Scholar. Similar searches were carried out in the GenEthx and Philosopher's Index databases, but did not produce any novel results pertinent to this project. I also searched specific journals which had been useful to me in earlier bioethics research, finding new publications in sources such as the *American Journal of Bioethics*, *Bioethics*, the *Hastings Center Report*, the *Journal of Medical Ethics*, the *Journal of Medicine and Philosophy*, the *Kennedy Institute of Ethics Journal*, and *Theoretical Medicine and Bioethics*.

These keyword searches began with a combination of the terms "metaphor" and "genetic disease" [or] "genetic disorder" [or] "inborn error of metabolism" in addition to a series of further searches involving the added terms "ethics", "bioethics", "narrative ethics", "narrative bioethics", and (based on one of the key themes which characterized the previous searches) "embodiment". I also discovered a number of useful results by replacing "metaphor" with the more general term "rhetoric" in another set of searches. The publications which seemed most useful to this project were identified from among the results by appraising all titles and abstracts. Following this cursory examination I coded documents for high or low-priority review, based on their perceived quality, validity, and degree of relevance to the project's specific aims.

Each review began with a brief overview to determine whether the title and abstract of that publication had adequately represented the relevance of the publication to this project, as many articles were originally identified this way that only made brief reference to metaphor in the main body of the text. I took notes from each that remained, with emphasis on theoretical content which appeared unique to that publication. Publications by the same author or authors, or which appeared in the same journal issue, were reviewed together in sequence when possible. Finally, I searched each document's reference section for relevant publications which had not been identified by the previous keyword searches. Rather than coding whole publications into different categories, I compiled specific notes under a flexible set of headings representing the proposed sections and subsections of the thesis. New categories were added as areas of interest emerged while conducting the readings.

The newly-added categories included the use of key metaphors within academia, as articulated to the public in advertisements by the biotechnology industry, and expressed by patients and laypersons. This research process enabled a comparison of the surveys, roundtables and other audience studies of genetic metaphor which had been identified as part of the general review. Accordingly, I conducted a systematic review using this subset of the identified documents. As the data was predominantly qualitative, I adopted a meta-synthesis approach aimed at identifying and integrating the common themes emerging from these publications.

As the analysis progressed I also eliminated the proposed categories covering extracts from primary sources such as advertisements and press releases, prominent individual genetic disorders such as sickle cell and cystic fibrosis, and the cultural role of heritability in general. Some of this information was moved under different headings that better reflected the overall aims and structure of the project as well as the amount of data available.

There were several further subsets of sources which I ended up having to cut from the analysis entirely. Those who have read *Illness as Metaphor* will be aware that, along with tuberculosis, one of the key diseases covered in the essay is cancer. Cancer is, of course, a disease that arises when genetic alterations cause cells to divide out of control, and elevated risks of certain types of cancer are often hereditary. I decided not to focus on representations of cancer in this thesis for several reasons: firstly, because many of those mutations arise due to environmental influences in addition to mutations which arise randomly; secondly, cancer has

enough of a presence in the popular consciousness to be considered independently of disorders which are associated with genes; and thirdly, it had already been done (see Sontag, 1974).

Another topic covered by Sontag which I was forced to leave out of this thesis was the description of works of literary fiction which reflected beliefs about the conditions being discussed. Having long been a fan of classic literature, this had been one of my favourite parts of the original essay. The problem I encountered when trying to do the same early in the course of this project was that relatively few works of literature have been written specifically about genetic disorders. Genetics in general is a very prominent theme in every form of media, and has been for decades. As such, a number of literary novels have dealt with the genome, such as Richard Powers' fantastic achievement *The Gold Bug Variations*, which even deals explicitly with the theme of metaphor in science. But despite a number of standout novels, I concluded that this review could not be treated well within the bounds of the chosen topic.

This thesis begins with an overview of the history of metaphor in genetics in general, as it would be impossible to write about genetic disorders without first explaining the figurative language attached to the archetypal gene and the "normal" genome against which many of them are defined. However, in the interest of keeping the focus on genetic disorders I have tried where possible to introduce this background in a concise fashion.

Chapter 1 traces the development of metaphors in genetics from the origins of the discipline up to the beginning of the human genome project, with particular attention to the competing conceptions of genes as individual objects and the genome as a representation of identity. Key periods covered in this section include the invention of the concept of the gene in the early 1900s; Erwin Schrödinger's 1944 book *What is Life?* and its influence on numerous geneticists including Watson and Crick; the paradigms introduced to genetics by military-industrial "technoscience" in conjunction with emerging theories in other disciplines; the negative imagery attached to genetic disorder in the 1970s and the growing influence of the biotechnology industry on the language of DNA.

Chapter 2 briefly discusses the Human Genome Project and its effects on metaphor before attending to the implications of the increasingly common representation of the human body as information. This is related to the idea of an ideal abstract body against which fallible individual genomes must be compared in ascertaining genetic disorders, despite our inability to justify objective normality in genetics. The chapter also provides an overview of the concepts of

geneticization and genetic imperialism, which have resulted in a simultaneous expansion of the importance of genes in our understanding of disease and the number of diseases classified as genetic. Finally, the chapter discusses how the common metaphor of language is used to represent the genome and the implications this carries for representations of genetic disorder.

Chapter 3 and Chapter 4 are both split into two complementary sections. The first half of Chapter 3 covers the role of metaphors of genetic disorder that are articulated through the mass media, both as news and as commercial advertisement, in addition to the considerations that lead to their use in the first place. The second half of Chapter 3 covers the effects of metaphors on patients diagnosed with genetic disorders or hereditary risk factors, particularly with respect to self-concept, choice of testing and treatment, and coping strategy.

Chapter 4 is dedicated to the two fundamental conceptions of the relationship between one's genetic disorder and oneself which emerged during the research undertaken for this thesis: the idea of the mutated gene as a specific, separate entity maliciously imposing itself upon the actual person, and the idea of genetic disorder as a mark of difference characterizing one's identity. These two opposing representations are illustrated by, respectively, the common metaphors of the gene as virus or contagious disease and the genome as soul or essence of humanity.

Chapter 5 summarizes the recommendations for research and clinical practice which have been offered by the authors of publications on genetic disorders and metaphor. In particular, the chapter discusses plans and suggestions for influencing the development of the metaphors and terminology of genetics, chiefly in order to reduce their potential for detrimentally affecting patient beliefs and experiences. As an example of how this might occur in practice, I summarize the existing literature on the recent attempt to replace "blueprint" with "recipe".

As this thesis is being presented to the Department of Human Genetics, I have taken the liberty of writing these sections assuming the reader has a basic familiarity with genetics. A more thorough explanation of the core scientific concepts (such as what DNA actually is) will likely be added when this text is rewritten for publication.

Chapter 1 – Genetic Metaphors in the Pre-genomic Era

In order to understand the contemporary use of metaphors for genetic disorders, I believe it is essential to provide an account of how they, as well as metaphors of genetics in general, have developed over the course of the discipline. As we shall see, many popular metaphors from scientific discourse and pass into popular usage. Therefore, we can come to a better understanding of these metaphors' significance by analyzing the factors that have shaped the scientific context in which they originated. It is also worth noting the nuances of composing any narrative about the history of communication, scientific or otherwise. As Doyle (1994) notes, the evolving discourse of genomics itself "constitutes a story" (p. 52), thus making it a narrative of narratives. The need to keep such histories engaging also tends to result in authors emphasizing the more suspenseful aspects of scientific development like competition, which has been quite frequent in the rapidly-developing discipline of genetics (Van Dijck, 1998, p. 19). As this section attempts to trace a specific use of language, it would have been unreasonable to attempt a narrative completely without gaps. However, it should offer a coherent overview of the relevant aspects of medical genetics.

The primary lesson of the upcoming history is that not all metaphors are fixed in the discourse. Some have remained more or less ever-present, some fade in and out depending on the scientific and social forces which influence each era, and others simply "die" in conventional usage, ceasing to function as metaphors (Van Dijck, 1998, p. 22). Those that become codified in this way can be used literally in some contexts and figuratively in others (Sidler, 2006). Metaphors can also be "reopened" when their meaning is challenged, or (more frequently) in popular science, where authors tend to use more metaphors and to identify them explicitly through the use of quotes, italics and similes (Knudsen, 2005; Pramling & Säljö, 2007).[4]

Metaphors seem to have a greater chance of survival when they have "local plasticity" but "overall robustness" (Klitzman, 2009), allowing them to adapt to specific contexts while

[4] Stern's 2000 book *Metaphors in Context* argues that (unlike Lakoff and Johnson's assertion that metaphors' meanings transcended context) metaphors have a variety of potential meanings based on their historical uses, which audiences must sort out based on how they are brought together. The most "fruitful" metaphors are those with a particular link to the subject but many potential meanings (Stern, 2000, p. 251; Condit et al., 2002). The conceptual metaphor theory has also been criticized for not taking social context sufficiently into account. In Foucauldian discourse analysis, metaphors are quite polysemic, meaning that they can take on multiple related meanings due to historical and cultural factors including the traditions and constraints of discipline-specific writing like science communication (Lopez, 2007).

remaining conceptually coherent on a broad scale. However, Liakopoulos (2002) argues that, "No matter how much or how little each image is used, it is always there ready for use providing the circumstances are appropriate."

Some analyses have even considered metaphors as ideologies or systems of meaning that invade different social spheres and affect discourse within them (Maasen & Weingart, 2000, p. 28), and Lakoff and Johnson (1980) have even suggested that metaphor replacement results in much of cultural change, such as their nebulous example of Westernization (p. 145). This represents another reason it is particularly important to place these figures of speech within their historical and cultural context.

Metaphor in Early Genetics

While the notion of hereditary disease in general has existed for millennia (Chapple, May & Campion, 1995), the specific types of traits considered to be hereditary have depended on contemporary sociocultural concerns (Lippman, 1992). Doyle (1994) even makes the intriguing claim that "Heredity has always been metaphorical," as the idea that traits are transmitted between generations fundamentally consists of a comparison of like with like (p. 53). At the same time, genetics has always tended to focus on differences rather than similarities (Hubbard & Wald, 1993, p. 39); indeed, the famed early-20th century geneticist J.B.S. Haldane described his field as "the branch of biology which is concerned with innate differences between similar organisms" (Haldane, 1942, p. 11).

Recurring cultural ideas like "blue blood" and stories of lost and rediscovered royal children have historically used heredity to reinforce the premise of inherent class differences (Nelkin & Lindee, 1995, p. 15-6).[5] This presentation of the aristocracy as biologically superior was, in practice, a self-fulfilling prophecy given the environments in which the poor were forced to live (Hubbard & Wald, 1993, p. 14). As Haldane (1938) pointed out, nobody seemed to hold Queen Victoria's descendants' hemophilia against them at the same time as they asserted the genetic inferiority of lower socioeconomic classes (p. 88-9).The concept of the biological "message" also dates back to the Victorian era (Kay, 1997; Knudsen, 2005), influenced mainly by the

[5] One frequently-cited example is the remarkably "genteel" Oliver Twist, whose parentage is eventually revealed to be upper middle-class (Lewontin, 1991, p. 23; Hubbard & Wald, 1993, p. 61).

invention of the telegraph in the 1830s (Fox Keller, 1995, p. 81). This would be one of the first metaphors of information to be applied to genetics.[6]

Syed, Bölker & Gutmann (2008) demarcate the first three key periods of genetics as "genetics with neither genes nor information", "genetics with genes but no information", and "genetics with both genes and information". The first of these could well be argued to begin with the groundbreaking research of Gregor Mendel, published as *Experiments on Plant Hybridization* in 1865. His experiments laid the foundation for what we now call Mendelian inheritance, including the laws of segregation, independent assortment, and dominance. We now know that this model only describes traits caused by single genes, but this subset does include several thousand genetic disorders, including around 80 percent of rare diseases (Melnikova, 2012). Mendel never mentioned genes in his work, as the term had not even been coined yet, although he did make occasional references to "elements" or "factors" which could be seen to prefigure them. He was not the first to propose units of the kind, but rather the inventor of a methodology that allowed them to be examined. Although Mendel was aware that heredity had a material basis, he was more interested in pursuing the statistical laws governing their patterns of inheritance than their physical origin (Hubbard & Wald, 1993, p. 40; Moss, 2004, p. 23; Roof, 2007, p. 4, 73; Shea, 2008, p.20; Syed et al., 2008).[7]

Mendel's work was rediscovered in 1900, the beginning of a formative decade that would see key developments in cytology and embryology as well as the coining of the word "genetics" in 1905 (Hubbard & Wald 1993 p. 40-42; Chapple et al., 1995; Shea, 2001). One of the scientists whose work contributed to the rediscovery of Mendel was William Bateson, who had also been responsible for the discovery that inheritance produced discontinuous traits rather than continuums (Moss, 2004, p. 26). The term genetics was made official following a recommendation made by Bateson to the 1906 Third Conference on Hybridization and Plant

[6] Although it would eventually become highly conventionalized, Rolston (2006) states that the idea of information in DNA can be considered metaphorical as "Humans first known the meaning of the word 'information' in our own experience".

[7] Mendel believed in a roughly preformationist conception of development in the Cartesian tradition, which argued that development basically consisted of the expansion of a tiny homunculus. This belief was historically opposed by the epigenesist view similar to that of Aristotle, which held that some sort of organizational force patterned development (Moss, 2004, p. 8-9; Roof, 2007, p. 155). Moss draws an important distinction between the different types of information used in preformationist views, which focused on phenotype, as compared to epigenesist views, which focused on the gene as defined by molecular sequence (Moss, 2004, p. 45-50)

Breeding (Roof, 2007, p. 75; Shea, 2008, p. 28). Although some authors have erroneously claimed that "genetics" arose from the word "gene", a misunderstanding which promotes the latter's centrality, the concept of the individual gene was not even invented until several years later (Shea, 2001; Roof 2007 p. 76; Shea 2008, p. 16).

The second of Syed et al.'s three periods, that of "genetics with genes but no information", actually began when Danish botanist Wilhelm Johannsen introduced the term "gene" in 1909.[8] Interestingly, he deliberately avoided assigning the term "gene" to a material object or biological mechanism. Johannsen used the word to represent the "hidden or covered reality" that was manifested in observable traits (Nelkin & Lindee, 1995, p. 3; Shea, 2001). In 1911, he described the "gene" as "nothing but a very applicable little word, easily combined with others" (Johannsen, 1911, p132-133). Moss (2004) describes it as "a kind of placeholder" (p. 2).[9]

This freed the term from being seen as part of any particular theory, allowing it to remain abstract and receive changing material definitions across time periods and differing organisms (Shea, 2001). Fox Keller (1995) suggests that the lack of knowledge about what genes actually were for the next few decades may have kept the focus on their action, allowing scientists to attribute them with "miraculous" powers (p. 10). Later in 1911, Johansen gave us the terms "genotype" and "phenotype" to describe the distinction between genetic makeup and observable traits. Although most of the early geneticists followed in Mendel's mathematical views, Johannsen had now established a metaphysical link between the two concepts (Fox Keller, 1995, p. 4; Syed et al., 2008), which would help genetics to emerge as its own separate discipline (Moss, 2004, p. 29). The first genetics journal, *Genetics*, would be founded in 1916 (Fox Keller, 1995, p. 4).

In the 1920s, embryologist Thomas Hunt Morgan, a pioneer of genetic linkage in fruit flies known for his mechanistic and reductionist outlook (Moss, 2004, p. 37) depicted the gene as "part physicist's atom and part Platonic soul" (Fox Keller, 1995, p. 9). He claimed that "at the level at which the genetic experiments lie, it does not make the slightest difference whether the

[8] He was inspired by Dutch botanist Hugo DeVries, a fellow champion of Mendel's work who had previously introduced the earlier concept of "pangenes". That term was itself derived from the tentative Darwinian concept of pangenesis (Nelkin & Lindee, 1995, p. 3; Shea, 2008, p. 16, 21, 30-32). Darwin had suggested that particles called gemmules migrated from all parts of the body into the germ cells, enabling them to produce another full organism (Moss, 2004, p. 20; Shea, 2008, p. 33).

[9] However, Johannsen was careful to declare that it did represent a reality, unlike the fictions of prior theories; he criticized Mendel, Bateson, De Vries and other previous geneticists for failing to differentiate between the inheritance of a trait and the trait itself (Moss, 2004, p. 29; Shea, 2008, p. 43-45).

gene is a hypothetical unit, or whether the gene is a material particle" (quoted in Shea, 2008, p. 51). It has been suggested that Morgan's work mapping genes on chromosomes marked the beginning of modern genetics proper, and also led to the marginalization of factors discovered not to be heritable (Moss, 2004, p. 36-37).

The first major popular science article to discuss the "gene" rather than genetics in general was a 1934 *Time* magazine article, which depicted them primarily as things that were tiny and therefore difficult to study visually. The frequency of such articles increased through the late 30s and 40s, amidst growing scientific commitment to their material existence (Shea 2008 p. 52-53, 76). During this time, the gene was often described as a discrete particle similar to an atom (Condit, 1999a, p. 71), a metaphor that would be quickly supplanted in scientific discourse by that of communication and the idea of DNA as a complex chemical molecule (Condit 1999a, p. 101-102).

This hugely influential genetic "code" metaphor was first popularized in the 1944 book *What is Life?* by Erwin Schrödinger, a Nobel Prize-winning physicist whose interests also ran towards philosophy and the immortality of "genealogical memory" (Fox Keller, 1995, p. 45). In addition to related metaphors like "architect's plan" and "builder's craft", Schrödinger (1992) described the basis of heredity as "some kind of code-script" (p. 21-22) or more simply as a "miniature code", illustrated by the example of Morse code (p.61).[10]

What is Life? was adapted from a series of lectures Schrödinger had given in Dublin the previous year (Fox Keller, 1995, p.46; Doyle, 2004, p.104), during the same stage of World War II in which the British team at Bletchley Park were in hard at work in the process of decrypting Germany's Enigma cipher. It was a year characterized by the impulse to dispel secrecy (Doyle, 2004, p.104). Carrying over metaphors similar to those found in the nineteenth-century thought experiments of Laplace's and Maxwell's Demon (Fox Keller, 1995, p.75), Schrödinger (1992) suggested that an "all-penetrating mind" could deduce the resulting organism by reading the code alone (p. 21). As such, this "code-script" was portrayed as *containing* the organism's ultimate four-dimensional patterning (Schrödinger, 1992, p. 21), both providing the instructions and building the final product (Roof, 2007, p. 79). As Doyle describes it, this conception "condenses a lifetime of development into a moment" and transfers patterning from the level of

[10] Schrödinger was not the first to make such a comparison (Olby, 1974, p. 246), and it is worth noting that the book has been critiqued both by contemporaries and modern authors for being both unoriginal and occasionally scientifically inaccurate (Ceccarelli, 2001, p. 67-69).

the phenotype to that of the genotype (Doyle 1994, p.55; Doyle 1997 p. 28-29) as defined by Johannsen. The "code" metaphor has therefore been critiqued as promoting a "one-to-one" model of genes' relationship to phenotypes (Weigmann, 2004; Sidler, 2006).

To his credit, Schrödinger was generally quite careful to avoid implying that his metaphors were anything other than figures of speech (Doyle 1994, p.55); he described his use of coding terminology as "a simile" and "too narrow" (1992, p. 22). Although Schrödinger (1992) made reference to the Habsburg Lip that affected European monarchs as an example of a hereditary defect (p.41) he also rejected the term "error" and described mutations as alternate "readings", surrounding the word with quotes to avoid a literal interpretation (p. 37).

Doyle suggests that Schrödinger's overall position as a "dilettante" (p. 20) allows us to read the work as a summary of the "rhetorical reservoir" of the 1940s (Doyle 1994, p.54; Doyle 1997, p. 27). Besides capturing the zeitgeist of genetic metaphor, Schrödinger's book proved to have significant impact of its own. With a few exceptions, *What is Life?* would end up being described by most historians of science as one of the most widely read and influential works of the 20th century (Ceccarelli, 2001a, p.63- 65). This was assuredly Schrödinger's intention; one of the goals of the book was to make physics and biology mutually intelligible, with an eye to inspiring interdisciplinary collaboration (Ceccarelli, 2001a, p. 87, 93). One of the book's greatest impacts may have arisen from the fact that it influenced both Watson and Crick, providing one of their only pieces of shared background (Moss, 2004, p. 53). James Watson, who began his education in zoology, stated that "I spotted this slim book in the Biology Library, and upon reading it was never the same" (Watson, 2000, p. 5), while Francis Crick allegedly changed his research interests from physics to biology after reading *What is Life?* (Watson, 1998, p. 13).

Crick was not the only scientist to make the switch between disciplines in that era. Schrödinger had made genetics appealing to physicists by "recouching it in terms of the physics of stability and predictability" (Moss, 2004, p. 56). Furthermore, by the 1950s life science was easily contrasted with nuclear physics, which – in the midst of nuclear weapon accumulation for the Cold War – had become widely seen as the study of death (Van Dijck, 1998, p. 34). This may also have resulted in a psychological motivation for many physicists to cross over to a more affirming line of research, in addition to the typical economic and academic motivations for entering a burgeoning new discipline (Ceccarelli, 2001a, p. 62). Regardless of the motivation, these ex-physicists' research interests would focus far more on the nature of the so-called

information stored in the nucleus of the cell than on its biochemical context (Fox Keller, 1995, p. 88; Moss, 2004, p.44).

The 1950s and 1960s

The last of Syed et al.'s three periods, representing the introduction of the "information" metaphor, coincides with the first of four conceptual periods identified by Van Dijck (1998) – that of the New Biology in the 1950s and 1960s.

Although the "code" metaphor has not received much study specifically as a rhetorical device, its success has been analyzed by a number of historians and philosophers of science, largely with respect to its ambiguity and the influence of the Cold War (Sidler, 2006). It was a time in which the tensions of the Cold War and the power of the Western military-industrial complex had huge ramifications for the development of science. Just as Schrödinger's influential writings took place simultaneously to the efforts of the British codebreakers at Bletchley Park, the growing emphasis on cryptography, cybernetics, systems theory and digital technology, along with the related overarching theoretical paradigm of structuralism, would influence both the terminology and practice of genetics in the Western world (Doyle, 1997, p. 86; Kay, 1997; Fox Keller & Winship, 2000, p. 110; Kay, 2000, p. 132, 221; Roof, 2007, p. 60-61).[11]

In order to take full advantage of these preoccupations, genetics had to represent itself as a communication science (Kay, 1997). This was, perhaps, not so difficult: the association of DNA and linguistics was made easier by the fact that both had simultaneously begun to adopt "cybernetic and informational representations" (Kay, 2000; Moss, 2004, p. 64). Furthermore, the development of cybernetics had already been informed by the analogy of system as organism (Fox Keller, 1995, p. 90-91), suggesting the feasibility of the reverse.

Information theory, the mathematical study of quantified information, had become prominent by the early 1950s (Fox Keller, 1995, p.18-19), contributing to the establishment of terminology like the "sense" and "nonsense" strands, "reading" and "proofreading" (Moss, 2004, p. 66). Scientists put aside their suspicions of metaphor in order to benefit from the swift rise of the "compelling and deceptively accessible" information theory and carve out a boundary for the

[11] Although Lysenkoism would fall from favour during the mid-1950s, the USSR would remain formally committed to their own "new biology" until 1964. These conflicting theories were popularly described as a conflict between nature-centric and nurture-centric theories of inheritance, the latter of which denied the existence of genes that could not be visualized (Condit, 1999a, p. 69-71; Shea, 2008, p. 56-60)

new discipline of genetics. Although its usage and meanings remained inconsistent, the language metaphor proliferated in the 1950s due to military-industrial funding (Gilbert, 1992; Nelkin & Lindee, 1995; Hedgecoe, 1999; Sherwin, 1999; Kay, 2000, p. 2; Petersen, 2001; Ratto, 2006). Genetics thus became a form of "technoscience", operating under a paradigm with a complex network of stakeholders and activities including scientific study, technological development, and public culture. Research now had to be evaluated outside of the scientific community, necessitating the use of metaphor as a tool for broader communication (Hellsten, 2005).

During the first two decades following World War II, science journalism displayed little carryover between the implications for science and those for society in general. It was instead marked by what Van Dijck (1998) calls a paradigm of "awe-and-mistrust" (p. 50-1). Both halves of this combination were influenced by the lasting cultural trauma of the conflict. Much of the rhetoric used by geneticists in the 1950s and early 60s, including the success narrative of Watson and Crick, was used to dissociate the field from the eugenic horrors of Nazi Germany (Van Dijck, 1998, p. 35, 61). Genes were no longer seen as subject to control by scientists, but rather as implacable controllers in their own right, with the environment as a secondary influence (Condit, 1999a, p. 83-85).

Watson and Crick's 1953 paper "A Structure for Deoxyribose Nucleic Acid" has received a great deal of rhetorical analysis (Gross, 1990, p. 54; Sidler, 2006), and many have argued that the paper's wording was heavily influenced by *What is Life* (Hedgecoe, 1999; Kay, 2000 p. 3, 23-33; Weigmann, 2004).[12] Although their research program was driven by the idea of a code (Rolston, 2006), it was not until their subsequent paper that the duo wrote of "the code which carries the genetical information" (Watson & Crick, 1953).[13] The "code" term soon became widely-used,

[12] Interestingly, Watson and Crick's iconic discovery took place in the same year as J. L. Austin's influential lectures on performative language – that is to say, statements which actually accomplish what they say. In a similar vein, DNA's structure has been compared to both meaning and function – and this may have helped it to make the eventual jump between structuralism and the more variable paradigm of poststructuralism (Roof 2007 p. 3, 31-2, 54).

[13] The revival of the code metaphor has also been attributed to George Gamow, an interdisciplinary scientist inspired by the Watson and Crick model who in 1954 suggested the more abstract, cryptanalytic term of "translation" to describe the production of protein directly from DNA (Knudsen 2005). The specifics of Gamow's model ultimately proved to be incorrect, and his approach to the coding problem as language analysis produced few accomplishments (Moss 2004 p. 64-66; Doyle 1994, p. 60, 65: Doyle 1997 p. 41). Although his metaphor had to be revised once the two-step process from DNA to mRNA and mRNA to protein had been verified, "transcription" and "translation" replaced the vaguer words like "correlate", "specify" and "determine" that had previously been used (Knudsen 2005).

making it the only popular informational metaphor prior to the 1960s (Syed et al., 2008).[14] As Condit (1999a) described it:

> "A coded message could simultaneously be passed on to others intact and provide guidance and direction for action. Like life itself, codes were orderly and yet complex, real and yet intangible. Codes also had the unique ability to feature both relative stability and openness to change. A message could be altered or corrupted . . . it was also important that the vehicle of this metaphor was widely comprehensible" (p. 101).

Moss (2004) wrote that the code-script exhibits slippage from a context "in which it can be held accountable – to one in which it appears to be self-sufficient" (p. 62), while Doyle (1997) claims that this discourse "translates the molecular as no different from the living" (p. 42). The image of material DNA itself also became increasingly popular, in some ways surpassing and incorporating that of the abstract gene (Condit, 1999a, p. 102-104). Some critics see the primacy of DNA in metaphor as a factor in the rise of molecular genetics as its own discipline (Gannett, 1999)

The molecule was attributed with human activity, imposing a sort of structure on the biology of the cell in which DNA remained central and regulation acted as a bridge to the external (Doyle, 1997, p. 80-81; Van Dijck, 1998, p. 36-38). This had the result of "ascribing to DNA the implicit power of an origin . . . making possible the idea that a straight line leads from DNA to proteins to 'us'" (Doyle, 1997, p. 58). The claim that language speaks people and vice versa is representative of postmodernism (Doyle, 1997, p. 61), a paradigm which emerged simultaneously to the influential disciplines of computer sciences and cybernetics (Roof, 2007, p. 3). Indeed, the information metaphor may also have been imported from cybernetics to genetics by Watson and Crick themselves. Certainly, Crick's paper "On Protein Synthesis" (1958), which first articulated the "central dogma" of genetics, used the term "information" to describe a linear transmission from gene to protein (Fox Keller, 1995, p. 18, 93; Van Dijck, 1998, p. 48; Syed et al., 2008).

[14] Not every metaphor applied to new developments in genetics ended up catching on; the New York Times initially described Watson and Crick's discovery as a "wire hawser" pulling traits between generations (Condit, 1999a, p. 101-102).

The central dogma of genetics gave "ontological priority" to DNA, according genes "directive agency" (Gannett, 1999).[15] Under the "central dogma" paradigm, gene products were often erroneously considered independent from the environment and other forms of biochemistry, even though some evidence had to be ignored to support this claim (Doyle, 1997, p. 72, 93, 98; Kay, 2000, p. 174-5; Sidler, 2006). Indeed, the term "information" was agreed upon in the 50s and 60s despite not having been tested experimentally in the same way other metaphors had been (Knudsen, 2005). Despite this perceived lack of rigor, the central dogma retained significant cachet within the discipline up through the completion of the Human Genome Project (Sidler, 2006).

The notions of genetics as information (and the code in particular) became dominant in all areas of discourse starting around 1960, and was used in reference to both the material of DNA and the concept of the gene (Van Dijck, 1998, p. 22, 123; Condit, 1999a, p. 270; Knudsen, 2005; Syed et al., 2008). Syed et al. (2008) suggest this may have been because the "information" metaphor has a more flexible meaning than "gene". For example, it has different connotations in communication science and informatics. In practice, this versatility more than compensates for the problem of lacking definitions for either term, connecting "narratives" from subdisciplines such as biochemistry, cytology, and physiology. Doyle (1997) even writes that "the trope of the 'code' has been as crucial to nascent molecular biology as the more obvious gadgets of ultracentrifuges, electrophoresis gels, and electron microscopes" (p. 25). "Code" was in the process of becoming both a conceptual archetype and a root metaphor (Van Dijck, 1998, p. 22) from which similar conceptualizations sprung.

The mechanism of gene regulation was discovered in 1961 by the *E. coli* experiments of Jacob and Monod. In addition to disproving the universality of the central dogma, they described their findings both as a "genetic program" and as a "series of blue-prints" (Jacob & Monod, 1961), marking the first entry of these popular metaphors into genetics (Nordgren, 2003; Fox Keller & Winship, 2000, p. 80).

[15] Interestingly, although "genetic authority" was expanded to mRNA transcripts, it was never applied to protein (Fox Keller, 1995, p. 95-96). One technical explanation may be found in the fact that protein function cannot be deduced directly from the genome as this contains no information about the rules of protein folding (Weigmann, 2004); I also think it is likely that proteins were excluded on the basis that they are popularly perceived as having actual biochemical functions lacked by RNA.

As a way of explaining development, the programming metaphor equated genetic material, written in a sort of alphabet of nucleotides, with the magnetic tape that was then used to process information in computers (Fox Keller 1995p19, 94-95; Fox Keller & Winship 2000 p. 81). Doyle (1997) suggests that this "linguistic turn" in genetics displaced Cartesian metaphysics onto the program (p. 108), acting as a sort of animating force within a machine: indeed, genetic metaphors tend to simultaneously portray the body as more mechanical while the object of comparison is depicted as more biological (Van Dijck, 1998, p. 23).[16] The information metaphor also helped meanings to be transmitted between disciplines during the 1960s (Fox Keller, 1995, p. 104), and associated metaphors like "alphabet" and even "grammar" also became more prominent during this time period. Popular new theories like Noam Chomsky's generative grammar and Marshall McLuhan's approach to media may have helped provide the intellectual context for such a paradigm shift (Van Dijck, 1998, p. 36-38, 123; Searls, 2002).

Genetics had a limited presence in the media until the mid-1960s (Van Dijck, 1998, p. 34); a study of the Science Citation Index by Knudsen (2005) found that articles referring to the code metaphor in their title or abstract rose from 20 in the 1956-1960 period to 153 in the 1961-1965 period, during which it became established and conventional. Prior to around 1960, the metaphors used both within science and for popularization were generally used to interpret research in terms of a new hypothesis, or to suggest the broader implications of new developments. Early papers tended to use explicit similes rather than metaphors, and those that were used were always marked out as figurative language through the use of text, quotes or italics. However, these practices faded away after 1962 (Knudsen, 2005). Although popularization became key to funding in the 1960s (Van Dijck, 1998, p. 24), metaphors that had been conventionalized and fully integrated into the biochemical explanations within scientific discourse became less and less frequently explained in text intended for laypersons, and by 1965 they had ceased to be explained or even marked out by quotes (Knudsen, 2005).

Scientists began to use the language domain as a basis for both popularization efforts and new scientific theories, such as the pioneering "DNA linguistics" subspecialty (Van Dijck, 1998, p. 123; Kay, 2000, p. 1), and it has been argued that the information and coding paradigms contributed to actual advances in genetics (Condit & Condit, 2001). For example, Avise (2001)

[16] As Van Dijck points out, the "code" and "computer" frameworks differ much more than might be expected in practice (Van Dijck 1998, p. 22-23). Indeed, the advent of the computer ultimately changed our definitions for many words like "message" and "information" (Fox Keller, 1995, p. 81).

suggests that the metaphor of encrypted language may have represented a useful conceptual step towards the cracking of said code. Furthermore, the prioritization of DNA in metaphor may have contributed to the rise of molecular genetics as its own subdiscipline, giving geneticists what Gannett (1999) calls a "professional stake" in the casual nature of DNA.

How did these evolving notions of genetics in general affect the way scientists wrote about genetic disorder? Limoges (1994) notes, with reference to the emergence of the concepts of genetic errors and lesions, that "some of the most crucial ethical issues are implicitly decided upon not in the context of philosophical discussions using the conceptual tools of ethicists, but in that of scientific research and discourse itself." (p. 123). However, it does not seem as though there was very much focus on mutation in the scientific literature during the time period. Early experiments in radiation damage and the discovery that damage could be reversed were performed in the late 50s and early 60s, but did not attract much attention. Fox Keller and Winship (2000) argue that the central image of DNA depicted it as stable and immutable, not subject to metabolism and revision (p. 27-30). In contrast, the concept of mutation had become associated with the dangers of the atomic bomb in the media and the public sphere. "Mutants" came to act as a label for "monstrous beings which plagued human civilization" in science fiction scenarios (Condit, 1999a, p. 71-72). The perceived stability of the gene lent itself to a conception of heredity in which radiation damage in the present would be carried forward to future generations (Condit, 1999a, p. 85).

At the same time, there was an intentional move away from eugenic discourse in the postwar era, and one effect of this was a move away from the ideal of "superior" children in favour of simply "healthy" and "normal" ones (Condit, 1999a, p. 90-91, 271). The notion of "genetic error" first arose during the latter half of the 1950s. For instance, Vernon Ingram's classic 1958 paper on sickle cell anemia used the term "abnormal" with respect to "mutations" and "differences" (Limoges, 1994, p. 114-15). Although sickle cell has a broad range of health effects depending on other factors, most of the attention focused on the fact that it arose from a single altered "letter" rather than that mutation's context (Condit 1999a, p. 104). Concurrently, the concept of "inborn error of metabolism" was being converted to that of "molecular disease" (Stanbury, Wyngaarden, Fredickson, Goldstein, & Brown, 1983, p. xv).

However, one of the first significant uses of the word "mistake" to describe a mutation was by Freese in 1959, during the race to solve the so-called coding problem (Freese, 1959; Limoges,

1994, p. 114-15). Khorana and Nirenberg's 1961 accomplishment in finally "decoding" or "deciphering" how DNA nucleotides corresponded to the amino acids of protein (Weigmann, 2004) produced an answer to the definition of the gene, which could now be conceptualized as coding for a particular enzyme whose defects produced corresponding defects in traits (Fox Keller & Winship, 2000, p. 54). The "patriotic and heroic" codebreaking metaphor was widely used in popular magazines (Condit, 199b, p. 101-102). However, this accomplishment resulted in few changes to the predominant metaphors (Knudsen 2005). In 1963, five years after its use by Freese, the term "mistake" had become entrenched as an extension of the information paradigm (Limoges, 1994, p. 114-15).

Popular culture has long associated genes with success or failure due to "tainted blood", justifying inequalities on a fated basis (Nelkin & Lindee, 1995, p. 80, 94-5, 100). Nelkin and Lindee (1995) compare the tainted blood paradigm to that of the XYY "supermale" karyotype, which emerged into popular culture after Patricia Jacobs' 1965 study in an Edinburgh prison suggested that it was associated with aggressiveness and criminal behaviour. These claims received significant attention in the media as well as films and crime novels (p. 83). These provocative pop culture conceptions emerged due to the scientists' publicity strategies (Green, 1985) and perhaps because, as a result of medicalization, genes were seen as "more newsworthy" than socioeconomic conditions (Nelkin & Lindee, 1995, p. 89, 91).

A probabilistic view of hereditary contribution to disease was first recognized after World War II as part of the move away from the intentional human manipulation advocated by eugenics, although the idea of susceptibility did not extend to rare disease and other Mendelian traits. The use of the "lottery" metaphor, which rose to significance over this time period, will be discussed further during the following section, as it has been most noted as a feature of discourse in the 1970s (Condit, 1999a, p. 86-87).

The 1970s and 1980s

For many years after World War II, journalists generally tended to uncritically recite the images offered to them by scientists (Van Dijck, 1998, p. 25). In the 1950s and 60s they had acted more like messengers on behalf of geneticists, and were not usually trained in science. However, in the years following Watergate, science journalism became more investigative and journalists began to see themselves more as working for the public than for the scientists

(Juengst, 2004). Although previous years had seen a steady increase in articles discussing ethical concerns, this increase was particularly pronounced (Condit, 1999a, p. 140).

As such, another development which may have increased geneticists' concern for their image during this time period was the emergence of bioethics, which was partly attributable to developments within their field of study. Alongside organ transplantation, psychosurgery and mechanical ventilation, concerns relating to genetics played an instrumental role in establishing bioethics as its own field in the late 1960s (Juengst, 2004). This profession came to fill the role played by theologians in science criticism before the secularization of the 1960s. Accordingly, geneticists and other scientists increasingly employed public relations experts to prepare material (Van Dijck, 1998, p. 25, 49-50, 70-71, 78-90). Perhaps as part of this effort, scientists of all kinds were highly esteemed and began to be photographed much more often for the media in the 1970s (Van Dijck, 1998, p. 18; Condit, 1999a, p. 107, 134).[17] Despite this, Liakopoulos' survey of biotechnology metaphors in British media (2002) found that the 1970s saw relatively little coverage of genetics, with the focus kept mainly to potential hazards. Indeed, the 1970s was "dominated by negative images" including eugenics and mad scientists, although Nazi imagery faded away fairly quickly during the first few years of the decade (Liakopoulos, 2002). Intriguingly, the 1970s also saw the beginning of an increased academic interest in metaphor (Stern, 2000, p. xi).

The key discourse during the 1970s seems to have been the media attention garnered by high-profile disputes over the safety of genetic research. It was even debated whether certain types of experiments ought to be banned , resulting in the appearance of what seemed to be "pro" and "anti" DNA lobbies (Van Dijck, 1998, p. 68-70, 74; Condit, 1999a, p. 147-151; Liakopoulos, 2002). This was the decade in which the gene both "took center stage" and became subjected to increasing social, ethical and political scrutiny, marking the second of Van Dijck's conceptual eras (1998, p. 30, 64).

Richard Dawkins' 1976 bestseller *The Selfish Gene* personified genes by casting them with human characteristics, even going so far as to call humans "lumbering robots" and "machines created by our genes". Although he claimed to be skeptical of metaphor, Dawkins used terms like "computer programmer", writing that genes "think" and "gamble". He also reintroduced a

[17] These depictions replaced the images of animals that had previously been more common, suggesting that "humans were understood to be the coding animals par excellence" (Condit, 1999a, p. 107), despite the fact that humans are rather poor representatives of organisms in general (Juengst, 2009, p. 133).

number of "factory" metaphors originally dating back to the 1960s, using terms like "business", calling genes the "policy-makers" to the brain's "executives" (Dawkins, 1976, quoted in Lewontin 1991, p. 13; Van Dijck 1998, p. 92-4; and Roof, 2007, p. 121). By describing the human body as a mere ephemeral container for something immortal, Dawkins' rhetoric even took on a sort of theological bent reminiscent of religious design (Nelkin & Lindee, 1995, p. 53; Van Dijck, 1998, p. 92; Moss, 2004, p. 7-8). Further discussion of this topic will be found in the section below on Theological Metaphors, beginning on page 88.

As Doyle (1997) puts it, "Bodies have been overlooked and recast as an effect of a molecule, an extension or supplement to the real, timeless, deathless bit of immanence known as DNA" (p. 8-9). This relationship between humans and genes implied that the structure of society was the sum of individual behaviours which were themselves the sum of individuals' selfish genes (Lewontin, 1991, p. 13). It was even suggested that, being a self-reproducing machine, an organism could even be viewed as a message in and of itself (Fox Keller, 1995, p. 109; Doyle, 1997, p. 96; Sidler, 2006).

During the 1970s, the concept of "genetic disease" began to expand both institutionally, culturally and economically (Fox Keller, 1992, p.291-293). Fox Keller (1992) attributes this to a combination of:

> "increasingly general acceptance of the explanatory framework of molecular biology; the postwar diminution of the burden of acute disease; intensification of scientific training for medical practice; changing expectations for health in the general public; and patterns of resource distribution for scientific research" (p. 293).

The results, unfortunately, have been pretty much unanimously declaimed as inaccurate, counterproductive and morally fraught.

During the 1970s the gene was, broadly, seen as one's fate (Liakopoulos, 2002). Perhaps for the political reasons which will shortly be discussed, sociobiologists often intentionally promoted the idea of DNA as "superior" and "all-controlling" (Van Dijck 1998, p. 92). Figurative expressions of the 1970s likened genetic disorders to the idea of a tainted bloodline, linked them to "primordial sin" and moralized them as somehow inherently evil (Condit, 1995). This language of "inevitable tragedy" and "natural disaster" makes stigma towards illness seem like a

natural outcome of the disorder itself rather than a byproduct of social conditions; and this paradigm directs genetic screening towards controlling reproduction by carriers of disease alleles (Steinberg, 1996). From 1971 through 1976, the media's expression of concerns about population genetics included "distinctly eugenic" statements to the effect that certain people should stop having children (Condit, 1995).

Indeed, according to the conception of genes as causative, genetic disease can only be stopped by controlling reproduction and eliminating the prospective patient.[18] In combination with the negative metaphors and "voluntary hereditarianism" paradigm which characterized the 1970s and 1980s[19], prejudice was exhibited to many people on the basis of genetic disorders as well as certain heritages (Hull, 1978; Steinberg, 1996; Juengst, 2004).

As a result of these misconceptions, individuals with the sickle-cell trait were fired or barred from certain types of employment (Hubbard & Wald, 1993, p. 34). Even the unaffected carriers of diseases like sickle-cell anemia and Tay-Sachs (which are respectively associated with African and Ashkenazi Jewish ancestry) were subject to stigma, distress and feelings of "strangeness". Meanwhile, those who had no family history of disease were potentially subject to undue optimism caused by the erroneous belief that they could not possibly be affected (Kenen & Schmidt, 1978; Condit & Williams, 1997: Condit, 1999a, p. 142-144).

Even though similar concerns are not usually expressed by scientists, Huntington's is broadly considered to be among "the most persuasive" examples of genetic determinism (Churchill, 2002, p. 185). In a paper published in 1979, one person with a 50% chance of developing Huntington's is quoted as saying: I feel like I'm "playing Russian roulette with a two-barreled gun and somebody else's hand on the trigger". Others associated the risk with a lottery (Wexler, 1979) bearing out the popularity of the genetic-disease-as-gambling metaphor which had been popular for decades before (Condit, 1999a, p. 270). Although variability of onset and development remain possible, the only type of disease an individual can know their exact odds of contracting is a hereditary, Mendelian genetic disorder (Wexler, 1979).

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[18] As Condit (1999a) notes, it is important to consider the legal context – therapeutic abortion first become legal in certain states of the United States starting in the 1960s and nationwide in 1973, allowing parents to terminate pregnancies that amniocentesis had shown not to be "normal" (p. 125-126).

[19] In various publications, Condit has written that voluntary hereditarianism remained dominant from as early as 1960 through 1973 (Condit & Williams, 1997), from 1971 through 1981 (Condit 1995), and throughout both the 70s and 80s (Condit, 1999; Condit, 2004). Despite this seeming lack of certainty about when the paradigm began and when it faded away, it is fairly clear that this was an important influence and that at bare minimum it was prominent during much of the 1970s.

The central metaphorical premise of voluntary hereditarianism was a sort of lottery of undesirable traits in which those unlucky enough to have "lost" were erroneously seen as liabilities to the "gene pool" yet whose responsibility not to reproduce was nevertheless a matter of individual choice[20] (Wexler, 1979; Condit & Williams, 1997; Condit, 1999a, p. 133; Juengst, 2004). Continuing the gambling metaphor, the "genetic lottery" was sometimes described as being based on a "roll of the dice" (Condit, 1995).[21]

The gambling metaphor persisted longer in the popular consciousness than voluntary hereditarianism itself did. Modern carriers of *BRCA1/2* mutations, which increase the risk of breast and ovarian cancer, describe their heredity in metaphors like "part of the cards I was dealt" (Werner-Lin et al., 2012). The laypersons interviewed in Gronnvoll and Landau's 2010 study used similar metaphors, expressing sentiments that the individual loses and "the house always wins". Gambling was also the third most common metaphor mentioned by their participants, behind those of viruses and time bombs. Further discussion of those topics will be found in the section below on the Virus Metaphor, beginning on page 77.

However, during the 70s the notion of passive vulnerability to random chance was seen by patients at risk of Huntington's as "psychologically unacceptable . . . or unassimilatable". They preferred to view their lives as "conforming to the laws of cause and effect", and most people who believed they either would or would not get the disease expressed "magical" reasoning to support this outlook. Almost every patient interviewed by Wexler expressed the idea of preventing the disease through "strength of will" and "positive thought". Many felt they had stronger chances of evading the disease because they had "always been lucky" (Wexler, 1979). As Wexler noted (1979), we tend to speak of luck as a personal attribute which opposes randomness. The opposition between selfhood and disorder evidenced here is an extremely important one, and further discussion of this topic will be found below beginning at page 72.

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[20] In the 1970s, Wexler noted that many individuals had children despite their genetic risks because of the "symbolic and magical significance of the child" for the parent's health and "immortality". Intriguingly, Wexler's research interests were informed by the fact that her own mother had Huntington's. Wexler would later be appointed the chair of the Human Genome Project's ELSI (Ethical, Legal and Social Issues) Committee (Van Dijck, 1998, p. 143; Couser, 2001). By dedicating 3% of its total budget for the purpose, the project would enable the largest single bioethical analysis ever undertaken (Lippman, 1992).

[21] As summarized by Condit (1999a), the patients to whom this responsibility was designated readily misunderstood information about the statistical likelihood of passing on genetic disorders (p. 135-136). Laypersons' limited ability to understand statistics and probability in genetics has remained an issue up to the present day (Klitzman, 2010).

While the belief that illness can be avoided if we wish hard enough offers an avenue away from mere passivity, those who take this position and subsequently develop the disease may take it as a condemnation of the patient's lack of faith or self-control (Wexler, 1979).[22] Several studies have also shown that mothers who did not use genetic diagnosis tend to be seen as more culpable and less deserving of social aid following the birth of a child with a genetic disorder, even among health professionals (Lawson, 2003; Shiloh, 2006). In the 1970s and 80s, failure to take the necessary precautions to avoid passing on a genetic disorder was even described as a form of abuse or violation of the rights of the prospective child, a sort of individualistic recasting of the older eugenic discourse (Hubbard & Wald, 1993, p. 25-26). The rise of personal autonomy as a bioethical principle may also have contributed to the moralizing of individual responsibility for genetic risk (ten Have, 2001). Indeed, Smiley has claimed that Americans see causation and blame as "interchangeable" (Smiley, 1992, p. 13). This viewpoint may have resulted in a focus on personal responsibility as a solution for biomedical problems, making genetic essentialism a useful ideological tool for neoconservatives who opposed social welfare (Nellkin & Lindee 1995, p. 128-129).

Genetic screening raised the issue of individuals' financial burdens on their communities, contributing to the incentive to control reproduction under voluntary hereditarianism (Condit 1999a, p. 131; Juengst, 2004). Responsibility for medical cost-containment was shunted to the faulty individual rather than the society (Lippman, 1992), turning social problems into individual problems, delegitimizing the notion of public health efforts through sociocultural, environmental or economic changes, and thereby depoliticizing the causes of disease (Hubbard & Wald, 1993, p. 5, 60-61; Nelkin & Lindee, 1995, p. 101-103; Rosner & Johnson, 1995; Condit, 1999b, p. 146-147; Churchill, 2002, p. 187; Everett, 2003; Melendro-Oliver, 2004).

The shift back to pre-World War II hereditarian ideas in the early 1970s was partly attributable to opposition against the growing civil rights and women's rights movements, which emphasized the role of nurture over nature in shaping the socioeconomic order (Hubbard & Wald, 1993, p. 9). Naturalizing these outcomes through genetics would "exonerate" the prevailing culture from wrongdoing in producing inequities while simultaneously implying the tractability of the trait (Rosner & Johnson, 1995; Roof, 2007, p. 97, 133-4). This was quite

[22] On the other hand, genetic mutations diffuse a certain form of responsibility from parents whose children's disorders might once have been seen as results of their poor parenting ability (Hubbard & Wald, 1993, p. 10).

harmful, as interventions like providing better education, healthy diets and places to exercise would have been much more effective in reducing mortality than individual genetic testing and behavioural changes. As one of many authors who have criticized voluntary hereditarianism (Hubbard & Wald, 1993, p. 5-6, 67, 77; Condit & Condit, 2001; Melendro-Oliver, 2004), Lippman (1992) argued that public health ought to be improved by addressing variations not in genetic sequence, but within "the distribution of wealth and power in North America".

While the intellectual disabilities which so concerned eugenicists prior to World War II were generally caused at least partly by genetic factors in the middle and upper class, the lower classes tended to develop them as a result of malnutrition, poor schooling and other environmental factors (Robertson, 1983).[23] Control of behavioural risk factors like weight, exercise and food choices are, of course, more readily available to more wealthy people in the first place (Hubbard & Wald 1993 p. 63). In addition, blaming behaviour on one's genes provides a better defense for wealthy criminals who cannot blame their crimes on a poor socioeconomic background (Nelkin & Lindee, 1995, p. 144).

This paradigm, which Lewontin (1991) describes as a genetic argument for capitalism, was only strengthened by the business metaphors that rose to the fore during the 1980s (p. 83), during which conceptions of individual responsibility and "voluntary hereditarianism" would remain dominant (Condit, 1999b; Condit, 2004). By 1980 the socioeconomic explanations for educational performance that were prevalent in the 1960s had been replaced by biological ones. Disabilities were seen as costs to the state, and the disabled communities expressed quite reasonable concern about a discourse that implied they should not have been conceived (Nelkin & Lindee, 1995, p. 163, 174-6).[24] However, the metaphorical landscape also shifted drastically, beginning in the latter 1970s. New technology contributed to computer science and

[23] Hubbard and Wald (1993) give the example of nutrition programs to treat pellagra (Vitamin B3 deficiency) not being funded by the United States' Republican government in the 1920s because it was presumed to be hereditary (p. 17).

[24] However, it is important to note that Condit (1995) claims there was no change in essentialism or the notion of genes as sole controllers between 1971 and 1991. She writes that media reports of human genetics during the medical genetics period responsibly avoided implying that genes acted as the full cause or automatically caused disease, instead focusing on risk and predisposition. Condit claims that findings otherwise by authors including Nelkin & Lindee, Lippman and Fox Keller reflect unrigorous procedures on their part, including a lack of systematic counting and low inter-coder reliability. She also argues that although the goal of classifying and reducing the number of handicapped persons suggests the condition is less desirable, the claim that this practice might have social effects is purely an assumption on the part of ideological critics (Condit, 1995).

bioinformatics, helping information metaphors like "code" to beoame fully literalized in scientific discourse (Fox Keller, 1995, p. 113; Kay, 2000, p. 1) and in magazine articles (Condit, 1999a, p. 160); while the lottery and odds metaphors decreased significantly (p. 270).

Indeed, the 1980s saw a shift from generally negative genetic imagery in the media towards metaphors of progress (Liakopolous, 2002), corresponding to a significant increase in public appreciation of the discipline (Van Dijck, 1998, p. 31). The primary reason for this shifting reception seems to have been the emergence and valorization of an economically successful biotechnology industry, which began mainly with the sale of diagnostics targeted towards women concerned about their children's health (Van Dijck, 1998, p. 97-98).

Interestingly for our purposes, the field of narrative ethics began to rise in prominence around 1980 (Churchill, 2002, p. 183-4). In that year, the US Supreme Court's narrow 5-4 decision in *Diamond v. Chakrabarty* (1980) confirmed the patentability of genetically modified organisms. Business prospects were further boosted by deregulation initiatives from the conservative ruling parties in the UK and USA (Van Dijck 1998, p. 91), which came into power in 1979 and 1981 respectively. Liakopoulos (2002) found that as genetics became more economically important in the UK, the British media reported many success stories and genetics gained a "feel-good factor". Condit (1999a) describes a similar enthusiasm in the American media (p. 169-171). Indeed, the so-called "pro-DNA lobby" would eventually be described as having won the debate (Van Dijck, 1998, p. 90).Van Dijck (1998) called this shift in public perception the "geneticization of the social mind", characterized by increased awareness of genetic risks and vulnerabilities (p. 97-98)

Metaphors relating to the categories of "progress", "economics" and "super" were most commonly observed by Liakopoulos (2002), while Condit (1999a) identifies the leading metaphor as that of the blueprint and its "manufacturing mentality" (p. 157). In accordance with the business focus of the decade, the 1980s also saw increases in figurative language like "harvest", "mining", "banking", "manage" and "factory". This continuation of the sort of economic language Dawkins had used in the mid-70s, acted as a way to describe the value of genes as property (Van Dijck, 1998, p. 95; Andrews & Nelkin, 2001; Nelkin, 2001).

Popularization metaphors like language, programming and personification also rose during this time period (Liakopoulos, 2002). The textual metaphors in particular were seen to enable the idea of gene as property, based on the knowledge that creations like computer programs and

works of fiction can be copyrighted (Roof, 2007, p. 107, 109). Roof suggests that narrative itself conveys a sort of "instant and naturalized causality" in which conflict ends in production, a gendered notion which "parallels our ideas of capitalist investment and payoff" (p. 18).

The metaphors of DNA as commodity reject the metaphors of theology and identity (discussed in later chapters of this thesis), instead casting genes as property to be owned and managed by the biotechnology industry (Andrews & Nelkin, 2001; Nelkin, 2001), who were "less interested in what DNA might 'mean' than with what it can do".[25] As Doyle (2004) puts it, industry is less interested in the stability implied by the theological metaphors compared to a valuable potential for self-reproduction and mutability. And by the 1980s, technology like recombinant DNA and the polymerase chain reaction began to provide this sort of functionality while dispelling the idea of genes as somehow secret (p. 106-107).[26] The promotion of genetics to the public in order to make DNA seem less dangerous and more tractable for human purposes made the biotech industry "image-dependent" (Van Dijck, 1998, p. 99-100). Even as they emphasized their financial viability to investors, these rising biotechnology companies emphasized the academic qualifications of their staff to the public, with many scientists returning to academia specifically to retain that image (Van Dijck, 1998, p. 106-107).

Commercial metaphors, such as describing cells as factories controlled by the genes, diminished around the early 1990s (Van Dijck, 1998, p. 120). Although the 1990s saw a great rise in British media coverage of genetics[27], as new products and technologies raised ethical concerns, there was also an increase in attention to public understanding, including both engagement initiatives and perception research on laypersons' views of genetics. Negative images of fear and mad scientists actually disappeared in this time period, and the predominantly negative metaphors of the 70s and positive metaphors of the 80s were finally replaced by a more even balance between the two (Liakopoulos, 2002). The most popular metaphors noted in the

[25] Medical anthropologists have asserted that the scientific language which has contributed to the commercial commoditization of an increasingly mechanistic body stems ultimately from the mind-body dualism advocated by Descartes (Andrews & Nelkin 2001, p. 5; Everett, 2003).

[26] Biochemist Kary Mullis (who won the Nobel prize for inventing the DNA-amplification technique of PCR and specifically cited Schrodinger and *What is Life?* in his acceptance speech) vividly described DNA in its natural state as "like an unwound and tangled audio tape on the floor of the car in the dark" (Mullis, 1993; Doyle, 2004 ,p. 103). Rather than searching for the meaning of DNA, Mullis' goal was to produce huge amounts of it mechanically (Doyle, 2004, p. 18)

[27] It should be noted that around this time genetics discourse had begun to occupy a take a more nationalized form, as debates developed on different ways in different topics from country to country (Bauer, Durant & Gaskell, 1998).

press during this period were ones useful for popularization, namely personification, language and programming (Liakopoulos, 2002). Indeed, one factor which contributed to the popularity of genetics in the early 1990s was the increasing prominence of computers in biotechnology along with popular culture as a whole (Van Dijck, 1998, p. 120, 181).

Over the course of this chapter, we saw the origin of concepts like the biological code and plan, as well as the individual gene, whose intentional abstractness allowed it to be both long-lasting and interpreted in many different ways. Many of these rhetorical developments, including both informational and economic metaphors, promoted the centrality of DNA in our understanding of biology and medicine. The ideas of genetic "error" and the "molecular" disease category took hold around 1960, and genetic explanations were seen as increasingly newsworthy by a public who had grown to associate the idea of mutation with monstrosity. The extremes of negativity and stigma towards genetic disorder noted above, as well as the disempowering metaphor of the lottery, would be reduced as genetics took on new connotations. However, many of the themes which arose in the pre-genomic era would become even more important in the decade to come, including the terminology of abnormality and the expansion of the category of genetic disease.

Chapter 2 – Genomics and the Body as Information

The Human Genome Project, Normality, and Categorization

Whereas the images prominent in the 1980s had presented DNA as an economic resource to be exploited by scientists, in the latter half of the decade the focus of the popular press swiveled to "medical genetics" as it applied to the health of the lay citizen (Condit, 1995; Condit & Williams, 1997; Condit, 1999b; Condit et al., 2002; Condit, 2004; Condit, 2009). Indeed, Fox Keller (1992) analyzed Medline and found that the number of "genetic disease" reviews had risen from 51 articles in 1986 to 366 in 1989, indicating attitudes increasingly receptive to the Human Genome Project (p.292). Unlike the focus on prenatal genetics and clear-cut genetic disorders which had particularly characterized the 1970s, the media of the 1990s displayed an increased interest in the testing of adults for their own disease predispositions so that they could adjust their lifestyles accordingly (Condit, 1999a, p. 181-183, 270).

This perspective was significantly more progressive and tolerant of "biological diversity" than preceding discourses. The references to tainted blood and making a reproductive sacrifice for the collective good that had been prominent under voluntary hereditarianism had disappeared by 1991, replaced by the idea that people with such conditions could potentially have healthy children (Condit, 1995). The medical discourse that arose in the mid-1980s reduced the identification of genes with "fate" or the "essential qualifiers and identifiers" of a person (Condit & Williams, 1997). Condit wrote that this paradigm "narrows the character of defect" and "makes genetic conditions simple, accidental illnesses that are tied neither to sin nor to the fundamental and permanent identity of persons" (Condit, 1995) while suggesting to the public that no individual was genetically perfect in the first place and everyone stood to benefit from genetic medicine (Condit, 1995; Condit, 1999a, p. 184).

Given its portrayal as a tool for the treatment of genetic disorders, the importance of the Human Genome Project has frequently and somewhat paradoxically supplanted the discourse surrounding individual genes causing Mendelian diseases by the rising concepts of genomics and the genome as a whole (Van Dijck, 1998, p. 119-120). A"holistic" and context-dependent approach to genetics was represented by the blueprint metaphor, which had begun to be used significantly more often starting in the 1980s. As Condit (1999a) writes, this metaphor suggests a

collaborative plan for the creation of an entire organism rather than a set of individual genes coding for specific traits. Like the metaphor of the genome as data in a computer, which had also become more common, this image has been noted for its implied passivity. The idea of this information being used by external agents reflected an increasing knowledge of the regulation of genes in different cells and contexts (p. 160-161, 270).

One study of undergraduates from a southern US university found only 39 out of 137 students found more recent metaphors like "blueprint" deterministic (including some who saw genetics as God's plan), and they did not always agree with that implication. Indeed, laypersons had more diverse opinions and were more sensitive to the nuances of metaphor than expected. Regardless of their rational analogical meanings, most participants interpreted these images as probabilistic and subject to the mitigating influence of technology or individual decisions (Condit, 1999b). Many laypersons understand a blueprint as a mere beginning, rather than a complete process, and some saw them as leaving blank spaces to be filled later (Condit & Condit, 2001). One respondent, for instance, stated that "a blueprint is a plan and not necessarily a reality" (Condit, 1999b).

Although some social critics viewed them as similar to the hereditarian concepts, the public tended to interpret the newer metaphors as less judgmental and perfectionistic (Condit & Williams, 1997). In particular, laypersons tended to find the blueprint metaphor more humane and less discriminatory than the preceding lottery metaphor, given the latter's implications of passive victory or loss (Wexler, 1979; Condit & Williams, 1997; Condit, 1999a, p. 166). Medicalized discourse reduced negative perceptions of people with genetic conditions, while those who were asked to read hereditarianist discourse expressed more judgmental opinions (Condit & Williams, 1997). Some described the metaphor of lotteries and luck as "cold" (Condit, 1999b). Rather than being affected by choice of metaphor, determinism was correlated with reading fewer materials and having right-wing political beliefs (Condit & Williams, 1997). However, the students did not make references to social or cultural factors (Condit, 1999b).

Hall (2003) claims that the new era of genetics actually "challenged many long-standing claims of social construction of gender, race and age, in addition to health and illness". Indeed, the Human Genome Project has also been described in terms of the individualization that marked the 1970s (Hubbard & Wald, 1993, p.60-61). Formally approved by the United States Congress in 1990, the Human Genome Project pulled much of the scientific community together with both

theoretical and practical aims. However, it has been argued that the shift to medical discourse undermined the legitimacy of research into nonmedical applications of genetics (Condit & Williams, 1997). The project was articulated to the public mainly in terms of identifying the molecular etiology of diseases, helping to diagnose them and ultimately to prevent congenital mutations. These medical images led it to be received enthusiastically by the public (Lloyd, 1994, p. 101; Van Dijck, 1998, p. 119-120).

The Human Genome Project made great use of pop culture for self-promotion, including television programs and documentaries funded by the National Center for Human Genome Research (Van Dijck, 1998, p. 25, 139). The project was depicted through metaphors like the quest for the Holy Grail or the Apollo mission (Rosner & Johnson, 1995). As Van Dijck (1998) writes that the images used to justify the Human Genome Project were "simultaneously dispersed through the discourses of science, journalism, advertisement and public relations, to the extent that [these media] become almost interchangeable". These images, she argues "are as important as 'real products'" (p. 138-140). For instance, the informational metaphors offered to the public by scientists – such as "mapping" – make their research sound precise, coherent, rigorous, and objective (rather than socially constructed) while simultaneously making use of popular rhetorical imagery (Petersen, 2001; Gronnvoll & Landau, 2010).

Patients and their experiences of suffering from genetic disorder also moved to the foreground, along with their treatment, in a sort of medical drama (Van Dijck, 1998, p. 137-8). Abstract notions like gene therapy were also made digestible and desirable through narrative (Van Dijck, 1998, p. 13), and the word "therapy" itself has very positive connotations as opposed to "treatment" or "surgery" (Hubbard & Wald, 1993, p. 110). However, the limitations of our ability to treat genetic disorder have tended to be downplayed in the media (Raspberry & Skinner, 2007), making them more appealing than actual surgical interventions. Very little of the discourse referenced any potential dangers caused by genetics research (Petersen, 2001), although Condit has pointed out coverage of ethical issues including privacy, genetic discrimination, and (given the debate over universal health insurance in the United States) equal access to health care (Condit, 1995; Condit & Williams, 1997).

Indeed, one of the side effects of the Human Genome Project was its effect on bioethics. By the end of the project, the discipline was firmly established in academia, and many popular books had been written about human genetics and the ethics of the Human Genome Project in

particular (Van Dijck, 1998, p. 141; Churchill, 2002, p. 183-4). Interestingly, from the mid-90s to the mid-2000s, human genetics was the best-funded research area in bioethics despite not actually producing the most academic publications (Juengst, 2004).

Although Nobel laureate Walter Gilbert (a co-founder of Myriad Genetics) noted concerns about social effects such as the use of genetics in medical insurance, he expressed optimism that the Human Genome Project's ability to produce "the information content of the genome" would help identify the genetic causes of both rare and common disease (Gilbert, 1992, p.92). Despite this focus on information, most publications focused on the medical rather than the technical side of the project, which had the effect that scientists were rarely portrayed to the public as information specialists (Van Dijck, 1998, p. 131).

In many ways, the competitive ability of the field seems tied to the self-promotion ability of its stars (Van Dijck, 1998, p. 17). Harold Varmus, a former Director of the NIH, was quoted towards the end of the project in 2000 as saying "There's a metaphor contest going on" (Angier, 2000). In addition to the theological metaphors discussed later in this thesis, scientists of the Human Genome Project expressed metaphors suggesting that knowledge of the genome would allow us to predict the future, such as "Delphic oracle", "time machine" and "crystal ball" (Nelkin & Lindee, 1995, p. 7). In line with the language metaphor (another upcoming section), it was also called a "future diary" (Annas, 1993). These metaphors of predictive readability can suggest a sort of inevitability about the future even when such notions are not backed up by scientific results (Nelkin, 2001). A great deal of this sort of gene-centric rhetoric was invoked in favour of the Human Genome Project, despite the fact that scientists were becoming ever more aware that genes themselves were subject to control during development (Fox Keller, 1994, p. 94) and had recently increased their focus on crucial biochemical activities in the cytoplasm (Fox Keller, 1995, p. 24-25). As Juenst (2009) put it, "the essentialistic genetic metaphors that animated the promotion of the Human genome Project . . . were already outdated by the science they attempted to illustrate" (p. 135).

If anything, the results of the Human Genome Project further underscored how much more difficult it would be than expected to leverage genetic knowledge into useful applications. Many have claimed that we are in a "postgenomic age" where we look beyond the genome to interactions among proteins, cells and tissues (Sidler, 2006; Juengst, 2009, p. 136). After the discovery that humans had far too few genes for a one-gene, one-function model to be accurate,

the leaders of the Human Genome Project promised that causal explanations would be forthcoming (Melendro-Oliver, 2004). Indeed, following the main phase of the Human Genome Project, the language in official documents largely switched to describing more practical applications and their impact upon systems as a whole (Roof, 2007, p. 112). While some authors have claimed that both academic and popular texts avoided determinism and emphasized the interaction of multiple factors (Condit, 1999a, p. 160-163; Nordgren, 2003), others have asserted that the growing scientific knowledge of complexity was not borne out in popular discourse (Churchill, 2002, p. 186).

Numerous authors have commented on how the development of genetic technologies and the Human Genome Project in particular have affected our conceptions of normality. Writing during the course of the project, Rosner and Johnson (1995) predicted that "the Project will determine what is 'correct,' what is 'real.' It will necessarily set standards, defining and cataloging what it means to be human, limiting what range of diversity is acceptable". Previous representations of the relationship between genes and environment, like the former acting before the latter or setting the boundaries for the latter's effects, tended to be replaced with comparisons of elevated or decreased disease risk when compared to an "imaginary norm" (Condit, 1999a, p. 163-164).[28] As Fox Keller (1992) wrote:

> "the very notion of 'culture' as distinct from 'biology' seems to have vanished [and was replaced by] a demarcation between the normal and the abnormal; the force of destiny is no longer attached to culture, or even to biology in general, but rather more specifically to the biology (or genetics) of disease . . . the new human genetics turns on the elucidation not of human order but of human disorder" (p. 297-298).

This has allowed allowing normality to be defined only in negation, in a way that has allowed it to elude study (Fox Keller, 1992, p. 297-298). Fox Keller (1994) further suggests that the healthy organism is the referent of disease in the same way that a "normal allele" is the referent of mutation – the entity to which it refers and by which it is defined (p. 95-96). This

[28] However, as Couser (2001) notes, although the standard genome was rhetorically constructed as a "definitive master text against which to match individuals", it was in fact already a "pastiche" of samples from numerous imperfect individuals. Continuing the literary metaphor, he describes it as "fictive" and "like passing off the works of a narrow tradition of Western male writers as universal".

makes the classification of "abnormal" with respect to the healthy ideal a comparative judgment (Lloyd, 1994, p. 100).

The gene, in its cultural sense, has been seen as promoting conceptions of social normalcy over diversity (Shea, 2001). As early as the 1970s, it was argued that terms like "abnormality", as well as "maladaptive", "malformation", "defect", "deleterious" , "aberration" and even "inborn error of metabolism" created what they called a "disvalued divergence" from normality (Hull, 1978). Even the fact that the cell has "error- correction" machinery implies that there is a correct, "intentional" and even teleological form of each gene, which mutations represent a divergence from (Rolston, 2006). Wilson (2001) calls this the creation of a "Genetic Other".

The term "disease-causing genes" is argued to be a mere euphemism, and the idea of normalcy as a lack of such genes is an abstract idea. It is also an implausible one, given our knowledge of the number of variants possessed by every member of the population (Fox Keller, 1994, p. 96). Many of the "defects" identified, such as the "premutations" of Huntington's, are benign (Juengst, 200, p. 140), and genetic predispositions for complex disorders like hypertension or depression can arise from "normal variants" of genes (Klitzman, 2009). Some medicalized traits – such as height, which can be manipulated to some extent via growth hormone treatment – vary naturally across the population in a "normal distribution" (Hubbard & Wald, 1993, p. 68-69). But it is highly debatable what threshold of height should be considered normal, healthy, or disadvantageous. As Condit (1999a) puts it, "there is no clear line between that which is a genetic disease and that which is simply . . . undesirable" (p. 138).

Similarly, in response to the issue of whether an objective normal even exists (see e.g. Gifford, 1990), Limoges writes that "Normality has no stable referential meaning: the reference is the experience of the individuals in the population" (Limoges, 1994, p. 124). According to Hubbard and Wald (1993):

> "DNA, the molecule, is material and real . . . However, our understanding of DNA and genes incorporates ideological baggage derived from our concepts of health and disease, normality and deviance, and what we can be or ought to be." (p. 6-7).

Although Moss (2004) agrees that medical normality arises from "taken-for-granted presuppositions", he notes that new alleles which affect phenotype generally eliminate a certain

biochemical activity (p. 41). It is tempting to accept a definition of disorder that simply suggests we look to a loss of function, given that it is easy to agree about when a protein is absent or is not actually involved in any processes. However, other diseases arise as the result of genes that produce proteins with gained functions, and aneuploidies like Down syndrome simply consist in there being too many chromosomes that are otherwise structurally normal.

Even if normality is defined on the genetic level where any variation is considered abnormal, this tells us nothing about the physiological effects we must take into account in actual medical practice (Lloyd, 1994, p. 104). The other option is for disease to be classified medically based on patient functionality within "socially negotiated standards", and researchers often and easily seem to slip between the two models (Lloyd, 1994, p.106, 109). Fox Keller (1992) summarizes her view:

> "Genetics merely provides the information enabling the individual to realize an inalienable right to health, where 'health' is defined in reference to a tacit norm, signified by 'the human genome,' and in contradiction to a state of unhealth (or abnormality), indicated by an ever growing list of conditions characterized as 'genetic disease.'" (p. 295).

The rhetorical images offered by advocates of the Human Genome Project (including James Watson and the National Research Council's Committee on Mapping and Sequencing the Human Genome) offered the two central images of "a base-line norm" and a "panoply" of genetic diseases (Fox Keller, 1992, p. 294). Indeed, as technology developed, there was a corresponding increase in the number of diseases considered genetic and people labelled genetically abnormal (Lippman, 1992; Juengst, 2000, p. 129).

Hall (2003) writes that "the atomising of the body into separate parts that make sense in the individualism of the capitalist economy" changes the power of the body "from a whole power to a fragmented power, opening up the body and the genes to domination and ownership"[29] Producers naturally have an incentive to maximize the number of users of their product, leading to a broadening of individuals included in the disease category. And of course, the most broadly

[29] From a Foucauldian perspective in which medicine is "the political technology of the body", this constitutes an epistemological shift towards knowledge of the interior of the body (ten Have, 2001).

applicable use of biotechnology is as a diagnostic (Hubbard & Wald, 1993, p. 70, 118; Condit, 1999a, p. 184). This also shifts the responsibility for detection of disease from the patient and physician to the geneticist who sequences their genes (Fox Keller, 1994, p. 97).

The fact that the emphasis had turned to testing adults for probabilistic predispositions meant that in practice everybody could be considered particularly susceptible for some disease (Couser, 2001; Stempsey, 2006). However, the term "abnormal" itself was used significantly less often in magazine articles (Condit, 1999a, p 271). As Condit put it, "Abnormalities were now the norm . . . Everyone was normal". It was the genes themselves rather than the affected people who were considered defective (p. 187), with the label transferred from the person to the specific gene.

The word "genetic" itself presently has two meanings. In addition to the one describing genes, it is also used to refer to things relating to origins. Indeed, it ultimately derives from a Greek root word meaning "generation". As such, a gene causing a trait is genetic in both senses of the term (Gifford, 1990; Roof, 2007, p. 83). The term "genetic" privileges genes as underlying causes and makes them seem tractable as solutions – a conceptual, institutional, cultural and philosophical phenomenon sometimes called "geneticization" (Lippman, 1992; Rosner & Johnson, 1995; Condit & Williams, 1997; Gannett, 1999; Hoedemaekeers & ten Have, 2001). Van Dijck (1998) defines geneticization rather obscurely as the "gradual expansion of loci of contestation where meanings of genetics are weighed" (p. 29), while ten Have (2001) describes geneticization as a heuristic that "reintroduces moral methods often forgotten". This shift resulted in a loss of opposing conceptual tools, producing an effect on both research and social paradigms (Griesemer 1994) in which differences between humans and even human biology as a whole are attributed to genetics (Nijhout, 1990; Lippman, 1992).

Genes are, of course, insufficient to produce traits independent of biochemical context and cannot reasonably be taken as proxies for the organism itself (Fox Keller, 1994, p. 90). It is true that they are likely to be involved in some way in basically every trait, but so are proteins (Smith, 2007, p. 89-90), and if any disease with a genetic component is defined as a genetic disease, then all of them are (Condit, 1999a, p. 138; Juengst, 2000, p. 129, 132). Smith (2007) writes that "The concept of a genetic disease is neither well-developed nor generally defensible as it is employed in the literature".[30] Geneticists tend to focus on the questions of "causal connection" but simply

[30] It is difficult to establish a good definition for "genetic disorder", as it depends largely on the population being considered (Gifford, 1990). Gannett (1999) gives the example of lactose intolerance,

resort to a gene-centric metaphor when "causal selection" is necessary to explain something (p. 83-84). Many authors have been concerned by the idea that genes should be considered the "specific causes" of disease, in a manner not unlike the early "beanbag" analogy used to disparage similar beliefs among early Mendelian geneticists (Juengst, 2000, p. 129, 132; Juengst, 2009, p. 135).

Under this paradigm, genes are seen as "irreducible and immutable" units of selfhood (Horton, quoted in Conrad, 1999), each responsible for separate traits (Conrad 1999) which collectively account for the entire organism. This is perhaps evidenced by the fact that DNA without genes eventually came to be referred to metaphorically as "junk" (Richards, 2001). Although these conceptions seems directly opposed to that of the more common passive and context-based metaphors discussed earlier in this chapter, Condit (1999a) suggests that they coexisted and brought an increasing sense of ambiguity to genetics (p. 161-163).

Over the course of the Human Genome Project, announcements of newly-identified genes frequently linked them to specific diseases, partly as a method of self-promotion (Wilson, 2001; Raspberry & Skinner, 2007). This offered the public a monogenic, Mendelian paradigm of genetic disorders, despite the fact that such diseases are relatively rare (at about 2% of total morbidity) and most traits are influenced in a complex, probabilistic manner by multiple genes and by epigenetic modifications (Conrad, 1999; Fox Keller & Winship, 2000, p. 68; Petersen, 2001; Melendro-Oliver, 2004; van der Ploeg, 2007). Everybody has some recessive genes or genes that contribute to disease or unwanted traits, and there are many mutations that can even be helpful (Lewontin, 1991, p. 50; Hubbard & Wald, 1993, p. 24; Conrad, 1999; Roof, 2007, p. 100). As such, this determinism incorrectly presumes the penetrance and expressivity of all

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which might be seen as a genetic disorder in populations accustomed to drinking milk whereas the milk might be seen as an environmental factor in cultures that never do. A similar argument could even be used with respect to dietary intake of phenylalanine and the inborn error of metabolism phenylketonuria. Numerous methods of establishing causality have been applied to genetic disorders, but none of them appears to be satisfactory to philosophers. The difficulty of establishing normality in a biological population prevents genes from being readily described as more "abnormal", as the immediately "precipitating factors" or even as "sufficient" for a trait (Smith, 2007, p. 91-92). Gifford (1990) and Smith (2007, p. 90) also question whether a specific gene could ever be proven to be the only factor necessary for the development of a certain trait, as the same could be said of absence of 1000-degree temperatures. Gifford (1990) argues that, rather than individual traits, only differences exhibited across a population can be attributed to a single factor. Smith recommends classification based on a pair of population-based, epidemiological criteria measuring the number of people in the population with the gene who have the corresponding disease because of the gene's involvement and the number of the people with the disease who have it without the gene's involvement (Smith, 2007, p. 102).

genetic disorders as well as the presence of a "normal" environment (Lloyd, 1994, p. 109; Porta,, 2003).

One reason for this perception may be that the limited number of diseases which are "predictable", "intractable", and inherited in a distinct pattern have long served as the "clearest examples" of genetic disorder to the public (Juengst, 2000, p. 133). It has been argued that these sorts of exceptional diseases, like cystic fibrosis and Huntington's can be considered unambiguously out of the biochemical context (Fox Keller, 1994, p. 96). Yet even Mendelian disorders like sickle cell disease, cystic fibrosis, and Huntington's are also multifactorial[31] and can result in a range of different phenotypes (Hubbard & Wald, 1993, p. 64-65; Limoges, 1994, p. 119-120; Juengst, 2000, p. 139). About 2000 mutations in the CFTR gene, which causes cystic fibrosis, have been documented (Hospital for Sick Children, 2011), and it has been suggested that the disease is actually "a group of related conditions" rather than a single monolithic entity (Hubbard & Wald, 1993, p. 37). The identification of the CFTR gene actually made the concept of the disease less concrete, as it expanded to include the entire phenotypic spectrum of identified cause (Hedgecoe, 2003).

Although several thousand monogenic disorders have been identified, numerous polygenic and multifactorial diseases have also been included under the umbrella of "genetic disorder" (Melendro-Oliver, 2004). Whereas the word disease has often been seen literally as "dis-ease" based on its etymological origins (Hull, 1978), the term "disorder" likely suggests not an experience of suffering so much as a fundamental defect in the underlying patterning of an organism. The uptake of the latter term represents a shifting of the idea of illness from a category of syndromes which are actually experienced by the patient to an explanation model that identifies them using criteria based on causal mechanisms within the body (Hull, 1978; Fox Keller, 1994, p. 97; Lloyd, 1994, p. 109; Steinberg, 1996; Melendro-Oliver, 2004).

Genetic variants resulting in mere risk predisposition for complex diseases and traits offer more room for environmental input than Mendelian disorders and others that highly correlate with the presence of disease (Klitzman, 2009). However, conditions like diabetes, schizophrenia, heart disease, Alzheimer's and alcoholism were advocated to be "genetic" diseases and disorders (Chapple et al., 1995; Rosner & Johnson, 1995; Gannett, 1999; Juengst,

[31] Multifactorial traits are those which are caused by multiple genes or by both genetic and environmental conditions.

2000, p. 131), as well as many forms of cancer which are thought of as being caused by exposure to carcinogens in one's environment (Fox Keller & Winship, 2000, p. 68; Lloyd, 1994, p. 109; Gannett 1999). Any conceivable deviation from a norm could be pathologized (Juengst, 2000, p. 145).

This process of expanding the umbrella of genetic nomenclature through this reclassification has been described as "genetic imperialism" (Juengst, 2000). It has provided additional work for geneticists and justified their efforts (Lippman, 1992), potentially defining what was seen as a problem, affecting funding priorities and elicit promises of clinical intervention (Hull, 1978; Freund & McGuire, 1995, p. 193-96; Juengst, 2000, p. 130; Melendro-Oliver, 2004).

Of course, many critics have challenged the causal role of DNA and the exaggerated idea that we ought therefore to be focusing on genetic solutions (Cranor, 1994, p. 130-131). Medicine involves prevention and treatment of disease rather than mere identification of their causes (Conrad, 1999). However, during the 20th century our ability to treat the actual effects of genetic disorders did not rise as quickly as our ability to diagnose them (Stempsey, 2006). Many genetic disorders that are treatable are not even treated at the genetic level. To reiterate the most commonly-offered example, phenylketonuria (the inability to metabolize the amino acid phenylalanine) uses a non-genetic test and a dietary treatment (Hull, 1978; Gifford, 1990; Richards, 2001; Moore, 2008). The infections that result from sickle cell and cystic fibrosis are treated with antibiotics (Hubbard & Wald, 1993, p. 65, 109; Hedgecoe, 2003). In many cases, our knowledge of how to relieve the symptoms is not predicated on eliminating or even fully understanding the genetic cause (Lippman, 1992).

Cranor (1994) argues that attending to a single causal condition is not inappropriate in context and can be illuminating, although complexity is important to take account of for the purposes of organizing research efforts and developing superior therapeutic targets. However, he doubts that conceptual claims should affect pragmatic research to any significant degree (p.131-134). It is also not true that molecularization inevitably causes determinism in medicine (Limoges, 1994, p. 121), and even subtle implications of determinism do not necessarily mean that recipients will agree with the message or be influenced by it (Condit, 1999b; Hedgecoe, 1999; Petersen, 2001).

The results of the Human Genome Project in some ways suggested both that genes were less monolithic in determining human traits than they had been assumed to be, and that natural

variation between individuals precluded the establishment of an ideal or normal genome. However, the project involved a heavy marketing push to the general public, and the images offered in support of the undertaking tended to emphasize our ability to predict phenotypes and treat diseases based on genetic knowledge. The corresponding medicalization of genetic imagery during the 1990s seems to have been associated with both a decrease in the stigma of genetic disorder and an increase in the sheer number of disorders known to science and explicitly labelled as such. The increased emphasis on genetics as a medical opportunity naturally drew focus to what should be considered a healthy genome, and the genome as a whole was frequently represented in metaphor over the course of the Human Genome Project. One of the two most prominent of these metaphors of the 1990s was that of the blueprint. The other was the code (Syed et al., 2008). While both of these are metaphors of information, the former is more visual and the latter is more readily interpreted as a form of language.

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The Language Metaphor

The code metaphor is increasingly argued to be not merely "literary" but "theory constitutive" (Baake, 2003, p. 72; Sidler, 2006). Indeed, the objections to "coding" are generally applicable to all metaphors relating to information and information processing (Condit & Condit, 2001), which served "as a kind of unifying key-metaphor" for the Human Genome Project (Syed et al., 2008) However, no consensus had developed concerning the units of information conveyed by the genome, nor how they should be measured within a biological system (Nijhout, 1990). In this context, the increasing knowledge of complexity lying beyond Crick's central dogma caused cyberscience to replace early forms of information theory as a framework for thinking about genetics. Along with the related disciplines of systems theory, cybernetics and computer science, this had the effect of transforming the body into an "informational network" (Van Dijck, 1998, 120-1).

By the 1990s, language-related terms like "editing" and "copying" had become literalized as a part of scientific informatics (Van Dijck, 1998, p. 123-125) along with those of proof reading, the open reading frame, the transcript, translation, and code itself (Syed et al., 2008). Although they were no longer intentionally used as metaphors in their scientific context, over the course of the Human Genome Project metaphors like "language" and "alphabet" were revived in the public

sphere (Van Dijck, 1998, p. 123-125; Nerlich, Dingwall & Clarke, 2002). As such, most of the genetic metaphors in use at the turn of the century involved information (Condit & Condit, 2001). In 2002, Condit et al. found that the most frequent metaphor for the genome was "code", which emerged unprompted in 16 of 17 focus groups. Writing of the other highly popular information metaphor, Van Dijck (1998) elaborates that:

> "the genetic blueprint is not a transcription of a real body, but the projection of an 'ideal body', constructed as a univocal formula, a mathermatical language. Biotechnology combined with informatics transforms the body as an (organic) object of knowledge into an ordered collection of biotic components" (p. 124).

She described this representation as the "collapse of digital, organic and metaphysical signifieds of the body" (p. 123). A prominent example of this Walter Gilbert's much-discussed claim that "one will be able to pull a CD out of one's pocket and say, 'Here is a human being; it's me!'", which he was known to do at his speaking arrangements (Gilbert, 1992 p. 96; discussed in Nelkin, 1994; Nelkin & Lindee 1995, p. 7; Fox Keller & Winship, 2000, p. 6; Kay, 2000, p. 1, 327; Weigmann, 2004). The implication of this sort of statement is that a human is reducible to the information stored in their genome and fully separable from the body.[32] Raspberry and Skinner (2007) comment that the idea "information extracted from the body can then be used to stand in for the whole of the person – is made possible by shifts in concepts of what the body is", towards a self represented by medical genetics as "textual", "transformable", and "visible".

Indeed, the overarching root metaphor of "information" suggests that it can be shared between many interested people (Stempsey, 2006), making it more valuable than the material itself in our information-based society (Everett, 2007), and stripping information from its material form makes it easier to communicate (Parry, 2004, p. 7). The fact that genomic information was indeed stored in a purely digital format suggested that the rhetoric of information was "no longer exclusively representational" and might be interpreted in legal or

[32] This sort of reductionism has even been described as the "effacement . . . of the patient" (Fox Keller, 1994, p. 97). Doyle (1997) wrote that "the implosion of a body and its description into coding and mapping . . . is the end of a story", pointing out the dual meaning of the world "resolution" (p. 18-19) and helping to explain why narrative is key in examining the bioethical issues predicated on genetics.

actuarial contexts in its own right rather than as a mere indicator of material reality (Van Dijck, 1998, p. 124-126).[33] The metaphor of life as information had now become materialized in such a way as to be susceptible to commodification (Rajan, 2006, p. 16-17). van der Ploeg (2007) writes that the "informatization of the body" as useful, machine-readable data blurs the classical dichotomy between material, biological reality and immaterial, social language. This challenges the construction of identity, while enabling "body data" to be readily accessed and assessed as normal or abnormal. She argues that genetics represents the epitome of "body-as-information", partly due to its conceptualization in terms like "information" and "coding", and partly because of its extensive use of computer science (van der Ploeg, 2007).

Although the metaphor of the computer program suggests that at least some genes are active agents in a regulatory role, the idea of the genome as a computer program was invoked much less frequently during the Human Genome Project than the older and more tangible informational metaphor of books and printed text, which depict genes as completely inert (Nordgren, 2003; Doring, 2005).[34] This is partly because of the fact that the four principal bases found in DNA are represented by letters (A, C, G, and T), readily allowing them to be presented as if they were either an alphabet (Copland, 2005; Gogorosi, 2005; Pramling & Säljö, 2007; Roof, 2007, p. 16) in both scientific and lay communication (Condit & Condit, 2001). The fact that these "letters" are grouped into codons within genes within chromosomes has suggested they be described as words, sentences and chapters respectively, with DNA itself as the language in which this text is written (Condit, 1999a, p. 102; Wilson, 2001; Nerlich et al., 2002; Copland, 2005; Vieth, 2010). Copland (2005) even compares transcription factors to rules of grammar and housekeeping genes to "very common words such as 'a', 'and', and 'the'".

Before delving deeper into the implications of the language metaphor, I should emphasize that these claims relating DNA to language are not literally correct. According to this logic, as Roof (2007) points out, chemical elements ought to be conceptualized in the same way, as they are represented by letters on the periodic table and could be argued to form molecules by combining as words (p. 16). Moreover, genes are not expressed from one end of the chromosome

[33] Interestingly, Couser (2001) notes that a legal defense is also a culturally-significant form of narrative.
[34] Hubbard and Wald (1993) wrote that the "program" was inappropriate because "DNA doesn't 'do' anything; it is a remarkably inert molecule. It just sits in our cells and waits for other molecules to interact with it." (p. 11) – at best, it "participates" in reactions along with other components (p. 64). Both Nijhout (1990) and Sidler (2006) have also suggested that genes be depictive through "passive" metaphors.

to the other in the same way that chapters are read from start to finish (Copland, 2005). Although it is true that bioinformaticians and linguists use similar computer-aided statistical methodologies, meaning that the language metaphor may be justified in these contexts (Searls, 2002; Syed et al., 2008), from an actual linguistic or cryptographic standpoint DNA out of its biological context lacks semantics and should not be considered a code so much as a "table of correlations" (Sherwin, 1999; Kay, 2000, p. 2; Petersen, 2001). Roof (2007) states more bluntly that "None of these analogies is accurate in terms of how DNA works or even what it accomplishes. All of them import values, meanings, mechanisms, and possibilities that are not at all a part of DNA." (p. 7). Unfortunately, these insights are not readily evident to the casual observer without knowledge of linguistics or semiotics.

The rhetorical features of metaphor do not exist independent of the audience, but depend on their cooperation; those that are obviously metaphorical, such as the comparison between genetic risks and ticking time bombs, are not likely to be taken literally. However, the notion of genetics as the study of "communication" and related metaphors is not so clearly rhetorical (Halloran & Bradford, 1984, p. 186-188; Shea, 2008, p. 114-117).[35] This confusion is not helped by the fact that numerous authors addressing lay audiences have explicitly stated that genetics *is* language. For example, in the preface of his popular 1999 book *Genome: the Autobiography of a Species in 23 Chapters*, science journalist Matt Ridley wrote:

> "The idea of the genome as a book is not, strictly speaking, even a metaphor. It is literally true. A book is a piece of digital information, written in linear, one-dimensional and one-directional form and defined by a code that transliterates a small alphabet of signs into a large lexicon of meanings through the order of their groupings. So is a genome." (p. 6-7)

Indeed, the metaphor is extremely common in popular science publications; other examples include Campbell's *Grammatical Man*, Jones' *The Language of the Genes*, Bodner and McKie's *The Book of Man*, and Wade's *Life Script* (Roof, 2007, p. 83). The fact that each of these,

[35] The materiality of DNA does not necessarily prevent this metaphor from broadly making sense. To quote Lakoff and Johnson (1980), "We speak in linear order; in a sentence, we say some words earlier and others later. Since speaking is correlated with time and time is metaphorically conceptualized in terms of space, it is natural for us to conceptualize language metaphorically in terms of space. Our writing system reinforces this conceptualization. Writing a sentence down allows us to conceptualize it with words in a linear order. Thus our spatial concepts naturally apply to linguistic expressions." (p. 126)

including Ridley's book and its subtitle, have used the metaphors in their very names suggests that, at the very least, their publishers thought this would be an effective way to market information about genetics to the interests of general public. Even Francis Collins (the scientist who led the NIH's involvement in the Human Genome Project) followed his 2006 book *The Language of God* with another nonfiction book unironically titled *The Language of Life* (Collins 2010).

In 2000, Collins appeared alongside US President Bill Clinton and Craig Venter (the founder of private sequencing company Celera) to announce the first genome assembly. The announcement marked an explosion of worldwide public interest in genetics. The *New York Times* ran 108 articles on the Human Genome Project in that year alone (Weigmann, 2004), resulting in numerous academic studies of how the achievement was presented. Language metaphors appeared in the media more frequently around "strategic" events such as the beginning and completion of the Human Genome Project, the publication of the first full genome sequence and the shift towards annotation, functional genomics and proteomics as the next phase of inquiry (Hellsten, 2005).[36]

Accordingly, comprehensive reviews of metaphorical references to genetics in media such as newspapers have been undertaken in Australia, Finland, Germany, Greece, the Netherlands, Spain, Sweden and the United Kingdom. Although regional references such as references to Greek mythology in Greek newspapers were of course noted (Gogorosi, 2005), there was remarkable similarity in the metaphors used by each country studied, likely because the publications' authors tended to refer to the same English-language sources and translate them literally (Gogorosi, 2005). Easily the most common type of metaphor across countries and languages were related to language (Doring, 2005). The most frequent of these was the "book of life" written at conception (Fox Keller, 1995; Kay, 2000; Nerlich & Hellsten, 2004; Copland, 2005), which had been a "media favorite" since the debut of the Human Genome Project (Fox Keller & Winship, 2000, p. 55) and existed long before being applied to genetics (Copland,

[36] As a result of its narrative formulation, a number of completion announcements of different kinds were made in order to bring closure to the project; despite scientists' knowledge that it would be a continuing endeavor, a "clear and achievable end goal" was needed to secure funding. This focus on achievement may have drawn focus from the project's social, economic, and political contexts and conflicts (Hall, 2003).

2005)[37]. The book metaphor has been argued to lend legitimacy and flexibility, as books can be conceptualized as anything from instructions to recipes to occult magic (Shea, 2008, p. 117). However, it is impossible to discuss this metaphor and its use in the Western world without reference to the Bible, and those connotations are developed in the upcoming section on theology.

Other terms used from the same domain included letters, spelling, words, sentences, paragraphs, passages, pages, chapters, a draft, a story, allusions to other works, paper and print, books, an encyclopedia, a dictionary, a library, an instruction manual, a cookbook, an autobiography of humanity and even a best-selling novel with the genome's donor as the protagonist (Nelkin & Lindee, 1995, p. 8; Petersen, 2001; Nerlich et al., 2002; Calsamiglia & Van Dijk; 2004; Doring, 2005; Gogorosi, 2005; Hellsten, 2005; Pramling & Säljö; 2007). Since the use of metaphors to describe properties tends to lead to their use in describing their associated acts, it was not surprising that verbs like "reading" and "writing" were also used frequently in these publications to refer to techniques such as sequencing, genetic testing or gene therapy (Calsamiglia & Van Dijk, 2004; Doring, 2005).[38]

Although the discovery that scientists had overestimated the total number of human genes and underestimated the sheer amount of "junk DNA" led to the adjustment of some of these metaphors and the gradual addition of some new ones to address this unexpected complexity, overall the language metaphor was "flexible enough to be extended" and became even more deeply conventionalized in the years following Clinton's announcement (Nerlich & Hellsten, 2004; Hellsten, 2005). The lack of change in these metaphors may have been partly due to the

[37] Derrida (1974) wrote on the ramifications of the conception of nature as book promoted by Descartes, pointing out the tension between this view and the technological activity of writing (p. 15-16). In general, post-structuralist perspectives on the language metaphor tend to emphasize the unknown over the idea of an absolute text (Hedgecoe, 1999). As such, Couser (2001) notes: "DNA is a post-structuralist's nightmare, a unique and constant signifier of the individual" in a postmodern era that tends toward the deconstruction of the subject.

[38] A number of different metaphors were also noted in the countries studied, sometimes occurring within the same publication or even within the same sentence (Doring, 2005). The second most common metaphor tended to be that of a map, sometimes containing "junk DNA" "wastelands" (Petersen, 2001; Doring, 2005; Gogorosi, 2005; Pramling & Säljö, 2007). The idea of the genome as a blueprint or construction plan with "building blocks" also appeared frequently (Doring, 2005; Pramling & Säljö, 2007), as did comparisons to other famous technological developments like the industrial revolution or the moon landing (Nerlich et al., 2002; Doring, 2005). Metaphors of space exploration have been noted as being particularly common in the literature (Ouzounis & Mazière, 2006). These sorts of spatial metaphors have historically played an important role in engendering values and emotions, as a huge endeavor is likely to appear more noble than a physically minuscule one (Van Dijck, 1998, p. 26).

limited number of authors who drew any attention to their inaccuracy over the period of time studied, even though Craig Venter did state his opposition to them (Nerlich & Hellsten, 2004; Doring, 2005). But the durability of these expressions was perhaps to be expected, given that the general idea of genetics as language had already lasted over 60 years.

In *Who Wrote the Book of Life?*, Kay (2000) suggests that language and its subdomains in general were particularly ripe for application toward the scientific domain of genetics. In addition to being highly symbolic, the "slippages" and "ambiguities" that occurred when the language metaphor lost its original reference created "space . . . that served as a repository for the scientific imaginary of the genomic book of life" (p. 3, 34). Commenting that "nature has always been textualized", she chronicles the language metaphor all the way from Roman times through the invention of the printing press and Schrödinger, in far greater length and detail than could be reasonably summarized here (Kay, 2000, p. 3, 23-33).

Another key text that discusses the metaphor of genetics as language is Robert Pollack's 1995 book *Signs of Life: The Language and Meanings of DNA.* Pollack argued that DNA has "the richness of a real language" (p. 153), with words, syntax, and meanings, likening protein domains to verbs and nouns and regulatory regions to grammar (p. 13, 72-75). Calling this "far more than a metaphor" (p. 21), he describes the genome as a "close to an encyclopedia in design and content", written in "magically small" text (Pollack, 1995, p. 11, 19) consisting of "a single string of letters", separated by sequences that serve as punctuation marks in a language like English or Hebrew rather than a logographic one like Chinese (Pollack, 1995, p. 21-22).[39]

However, this language has to be felt by the cell like Braille to be read rather than from a distance (Pollack, 1995, p. 22), and Roof (2007) argues that the physical contiguity of the genetic machinery used in transcription and translation places it in the realm of the mechanical, which we often mask with metaphor (p. 26). Others have described sequence translation as a movement

[39] This necessarily casts the language of life as a Western one, which carries some vexing implication especially with respect to the language-related theological metaphors which will be discussed later. As far as I am aware, there have not been any studies on the language metaphor in China, Japan, South Korea, or Vietnam, but one would certainly make an interesting addition to this field of study. As a further note on the comparison between heredity and specific human languages: Linguists sort languages into "families" connected by "genealogical family trees" (Searls, 2002; Ouzounis & Mazière, 2006), and Pollack (1995) notes that different languages tend to keep people apart and preserve genetic differences between them (p. 168-9).

from digital to analog, words to deeds (Moss, 2004, p. 70).[40] Pollack (1995) himself compares the biochemical process of translation (in which mRNA sequences are used as a template for the sequences of amino acids in protein) to actual bilingual translation (p. 81), with the relation between amino acid sequences and protein conformation being that of signifiers to meanings (p. 68). Indeed, language carries meaning, and the ability to read it suggests that scientists might be able to study it as a way of understanding the meaning of the organism itself (Condit, 1999a, p. 102). The term "reading", especially in conjunction with related metaphors like "our own instruction book" has also been criticized for elevate genes over the other biochemical apparatus of the cell and implying a sort of context-free uniformity of outcome (Rosner & Johnson, 1995; Van Dijck, 1998, p. 125, 152; Morange, 2001, p. 22-23; Ceccarelli, 2004; Nerlich & Hellsten, 2004).

These metaphors create the image of the genome as a "comprehensive and unbiased resource" which defines the "natural order"; a simple, uniform entity written for experts to decode and use in predicting and controlling the human body (Nelkin, 1994; Nelkin & Lindee, 1995, p. 8; Van Dijck 1998, p. 129; Weigmann, 2004; Calsamiglia 2005; Doring, 2005; Gogorosi, 2005; Hellsten, 2005; Vieth, 2010). Likewise, Roof (2007) suggests that DNA is in many ways "mechanical with none of the arbitrariness or indeterminacy of language: C always requires G, for example." (p. 88). And Doyle (1997) argues that despite its linguistic qualities, the genome has the "inability to articulate a complete description" like the "human finitude", "limits, deaths, and forgetting . . . that also comprise the human narrative" (p. 108).

This point of view is not universal. Richards (2001) argues that the model of development as reading of text remains true to the basics of molecular biology while providing rhetorical space for developmental processes and other influences upon phenotype. Similarly to the polysemic nature of language (and metaphors themselves) in which meaning is influenced by context, the genome produces different results based on its environment (Ceccarelli, 2004)[41]. Schmitt & Herzel (1997) wrote that "DNA sequences possess much more freedom in the combination of the

[40] "Translational research" from basic science to clinical intervention has, similarly, been interpreted via the linguistic metaphor (Juengst, 2009, p. 131).

[41] As Ceccarelli (1998) noted, "the term 'polysemy' is itself polysemous". Most of these interpretations refer to the fact that language can have a finite variety of distinct meanings. The authors cited in this thesis have generally used the term to indicate the fact that a metaphor can be understood differently depending on their context, including both the language surrounding it and the individual who is reading.

symbols of their alphabet than written language or computer source codes". As Sidler (2006) describes it:

> "Much like written words and sentences, genes are read and reread continuously, producing different actions, meanings, and narratives. Like the contextually-influenced reading of text, some genes are "skimmed over" while others have great emphasis, depending on the context".

Comparing DNA to actual human languages can help to contextualize it, conveying the idea of a stream of a limited number of symbols carrying information in a flexible manner that provide a multitude of potential semiotic interpretations in context (Pollack, 1995, p. 12, 64-67). Several authors have suggested emphasizing the fact that language and genetics are both products of history, fundamentally ambiguous, and subject to social context (Nelkin, 1994; Nelkin & Lindee, 1995, p. 9; Hedgecoe, 1999).

This set of metaphors, of course, extends to genetic disorders as well. Pollack (1995) argues that mutations "provide natural opportunities for interpretation" (p. 85). He described disorders resulting from chromosomal translocations and duplications as "like blocks of articles accidentally bound out of alphabetical order", which may result in congenital defects because "users cannot easily find an article if it is not in its proper – though arbitrary – alphabetical place" (p. 21). In a similar vein, Copland (2005) likens an individual with genetic disorder to a "book of life" telling a good story despite the lack of certain nouns. Point mutations are often depicted as "typographical errors" and "misprints" (Lippman, 1992; Pollack, 1995, p. 39), and the cell's "DNA repair machinery" (itself a metaphor) has been described as a "spell-checker" (Gura, quoted in Wilson, 2001). These unassuming figures of speech seem to belie the fact that when the "meaning" of the text changes in a way relevant to patient experience, the new meaning is most commonly cancer (Roof, 2007, p. 94).

Terminology like the description of mutations as "spelling errors" implies that an objectively correct version of the genome exists (which of course is not true, as discussed in the preceding section) and that there may be an opportunity to "edit" and correct the mistake (Wilson, 2001; Gogorosi, 2005). It is possible for the environment to be seen as having a role in rewriting the genetic code. However, from early on in the development of the discipline articles focusing on

the code metaphor tended to represent "the opportunities for dynamic interaction between conscious, individual human minds and the gene"; journalists of the 1960s routinely suggested rewriting the genetic language as a way of returning meaning to it (Condit, 1999a, p. 106-107).

Roof (2007) writes that disease genes are generally "presented as if science has found an on-off button or a talisman" resulting in "belief in a more powerful and instant science that gives humans shamanistic control over complex phenomena" (p. 66). This depiction can be misleading regarding the actual role of genetics in health and could inadvertently raise patients' expectations for successful treatment (Condit, 1999, p. 108; Gogorosi, 2005; Klitzman, 2010). Raspberry and Skinner (2007) found that parents of children with genetic disorders expressed great optimism in the ability of genetic research to produce cures such as "deleting" the defect in question.

By the end of the 20th century, the information metaphor had created a sense of the genome's vulnerability either to "corruption" or "augmentation" (Condit, 1999a, p. 88), with disease as "textual irregularity" and health researchers as "editors" (Wilson, 2001). Roof (2007) argues that the genome became "a language that genetic medicine hopes to rewrite" (p. 23), describing the "code/language analogies" and "narrative of writing" as implying intentional control and manipulation (p.83, 177). Indeed, according to Syed et al. (2008), genetic information as a metaphorical construction "refers to human action and not primarily to natural entities". Pollack (1995) even offers the idea of technology as a "molecular word processor" offering the creation of new "literature" (p. 96).[42] This sentiment is mirrored by Roof (2007), who concludes that "If DNA is a language, we can make it say things. We can control it, not it us" (p. 65). She suggests that being bought, owned and sold is "the logical extension" of the "book of life" metaphor (p. 66), as language implies an ability to be authored, edited and copyrighted (p. 16, 90).

In this section, we discussed two consequences of the information or language metaphor, both of which carry implications for lay beliefs and use of medical services: the diminishment of the importance of the rest of the body in favour of a shareable and readable information-self, and the implication that genetic variations are equivalent to typos that we should correct to form the words they were meant to be. The latter encourages the use of genetic testing and an overly-optimistic interest in gene therapy, and as the following chapter notes, the language metaphor was indeed used for commercial purposes by biotechnology companies.

[42] Rather amusingly, Pollack (1995) concluded that Italo Calvino's characteristics of great literature from *Six Memos for the Next Millenium*, lightness, quickness, exactitude, visibility and multiplicity, all applied to the genome (p. 176).

Chapter 3 – Uses of Metaphor in Genetic Disorders

Metaphors Directed at Laypersons

A large proportion of scientists work in the biotechnology industry, which has its own motives for the use of rhetoric and metaphor; corporate entities must raise expectations among both potential customers and investors by implementing positive and desirable imagery for their products (Hubbard & Wald 1993, p. 2; Van Dijck, 1998, p. 12, 104-110; Petersen, 2001). A major goal of the Human Genome Project was to transfer genetic technology to private industry (Gronnvoll & Landau, 2010), and over 100 genetic testing companies had emerged by 2003 (Gollust, Wilfond & Hull, 2003). Many of these companies and products, including 23andMe, deCODEme, Knome, Mycellf, and Mygenome, include first person pronouns in their names (Nordgren & Juengst, 2009), and deCODE adopted both its name and its slogan, "decoding the language of life", from the common text metaphor just discussed (Weigmann, 2004).

These corporate brands and slogans emphasize the uniqueness of one's genome even as the companies' services their services offer kinship via genetic similarity. Placing emphasis on biological "heritage" and "roots" rather than cultural legacies, their advertisements imply the conflation of genome and selfhood. Although the name of 23andMe suggests juxtaposition rather than equivalence, the company itself claims to offer "self-knowledge". Some of these companies also add a moral message of responsibility for one's own health. As such, personalized medicine has raised concern about promoting more individualistic conceptions of identity (Klitzman, 2009; Nordgren & Juengst, 2009; Widdows, 2009).

Another issue raised by depictions of genes out of their social context is that clinical communication and direct to consumer advertisement both tend to imply medicine should be used to treat heredity, addressing the etiology rather than the symptoms of illness (Parrott & Smith, 2014). Companies offering genetic diagnostics have disclaimers saying not to use or take action without healthcare advice (Nordgren & Juengst, 2009). However, there have been surges in both physician-directed and direct-to-consumer genetic testing, with few genetic counselors are available to be consulted (Klitzman, 2009). According to the United Health Center for Health Reform and Modernization, almost $5 billion USD was spent on genetic testing in the first half

of 2012 and this figure is predicted to grow to between $15 and $25 billion by 2021 (UnitedHealth, 2012).

Although promotional material for biotechnology companies undoubtedly convey images of genetic disorder to many people, the primary way in which genetics enters public awareness is through the news media (Conrad, 1999; Petersen, 2001).[43] As such, the remainder of this section will address representations that are intended to be primarily informative rather than (explicitly) commercial.

Individuals with greater experience in an area tend to use metaphors more often as they attempt to share meanings with others (Parrott & Smith, 2014), and metaphors can be especially important in science communication when they are already common in "elite discourse and media representations" (Barry et al. 2009). Metaphors can be used to help educate laypersons by articulating complex or technical information to people who lack much background or interest in the area (Lakoff & Johnson, 1980, p. 115; Van Dijck, 1998, p. 21; Nelkin, 2001; Liakopoulos, 2002; Barry et al., 2009), placing concepts within their sociopolitical experience (Condit & Condit, 2001) and shaping public opinion with respect to emerging concerns (Barry et al 2009). Narrative shapes the ways in which genetic research and medical anomalies are presented to the public in the media, as well as the ways in which they are interpreted and shaped into personal stories of illness that fit individuals' own conceptual frameworks (Van Dijck, 1998, p. 15). Beyond the mere explanation of research findings, metaphors convey beliefs about the nature of science and technology and the role they are to play in our lives (Nelkin, 2001; Petersen, 2001).

As such, Van Dijck (1998) calls science not just a "fact-producing process" but also an entwined "image-producing process" (p. 11). Indeed, science writers use similar strategies as authors of fiction (Van Dijck 1998, p. 26).[44] Knudsen (2005) comments that:

> "writing textbooks, popular scientific articles or review articles, in which the specific task is to identify central scientific narratives and to have the individual research fragments

[43] Representations of genetics are also common in other types of television (Condit 1999). For instance, DNA is used as a metaphor in advertisements; from relatively early on in the Human Genome Project genetic metaphors were being used to sell products like cars (Nelkin & Lindee, 1995, p. 96-97).

[44] In a sort of call to action, Gross (1990) once wrote that "it is necessary to show that science shares essential characteristics with other routes to knowledge, routes as different as autobiography, history and literary criticism" in order to strip science of pure ontological privilege and allow epistemological pluralism (p. 55)

make sense . . . presents an ideal situation for the creation of metaphor, as both the genre invites them and the writing specialist approaches the task with a mind prepared for identifying the larger picture".

Van Dijck (1998) writes that, since narrative "serves as a mode of cognition", "It is through the lens of story-telling that we can perceive communal rhetorical strategies and images in the various discourses on genetics" (p. 15). Of course, one specific way that we can do this is to examine the use of metaphor. Analyzing British media articles from 1973 through 1996, Liakopoulos (2002) divided the principal metaphorical themes of genetics into:

- Promise (metaphors of progress like revolution and key and metaphors of economics like business and gold rush)
- Scare (metaphors of Pandora's Box and eugenic dystopias)
- Other Science (popular metaphors of information technology like programming, codes, engineering, and archaeological discovery of an ancient library)
- Religion (metaphors of Holy Grail, "essence" and immortality, touching on issues like evolution and free will)
- Natural Order (metaphors of monsters, aliens and superhumans)
- Gene Person (metaphors of genes as personified organisms and people as genes), and
- Scientist (metaphors of mad scientists and designers)

Those of the "Gene Person" and "Religion" bear the most importance for this topic, and they are further discussed below beginning on pages 80 and 88 respectively. As is evident, there were many positive as well as negative metaphors presented in the media over the time span analyzed. These were roughly balanced overall: Liakopoulos (2002) notes that the four most popular, those of personification, IT, engineering and the library could be interpreted either way.

Nevertheless, many studies have remarked on the positive representation of genetics in the media (Durant, Hansen & Bauer, 1996, p. 242; Petersen, 2001) and suggested that inaccurate beliefs about science could be spread by overly optimistic depictions of genetics in news media and popular films (Klitzman, 2010). The social concerns of the day tend to aggregate genetic explanations (Nelkin & Lindee, 1995, p.16; Shea, 2001), and these are often sensationalized in

newspaper and magazine headlines as cultural icons (Shea, 2001), leading to an unscientific portrayal of genes as a predictive, readable reference about the inevitable future (Nelkin, 2001).

Although scientists are, of course, aware of the importance of gene-environment interaction in disease, media stories generally present a much more simplistic version of disease etiology and, indeed, fail to capture the complexity of the research process itself (Petersen, 2001). The multifactorial, multigenic or non-genetic causes for most disorders are rarely mentioned in the media, potentially giving the public the false impression that genetic diseases all arise from a single mutation alone and diminishing the role of culture in health (Kay 2000, p. 326; Petersen, 2001). For similar reasons, publicity for the identification of disease loci and other promising findings that suggest the imminent development of treatments are often far more prominent that their subsequent retractions and refutations, and negative or inconclusive results are often buried, if they are reported at all (Fox Keller, 1992, p. 296; Conrad 1999; Petersen, 2001).

Part of this distortion is likely due to journalists' need to advance their careers by producing stories of greater importance and significance to the general public (Thompson, 1994, p. 118), as discoveries with practical implications make for more interesting reading for a general audience (Hubbard & Wald, 1993, p. 5; Nelkin, 2001). Likewise, programming choices have been influenced by the fact that the public tends to be more interested in explicitly medical research than other types of science (Durant et al., 1996, p. 235; Petersen, 2001).[45]

The media's selection of themes and issues to focus on limits the scope of public debate (Priest, 1994, p. 168), and the topics emphasized depend largely on journalists, who strategically use literary techniques including metaphors to articulate complex scientific concepts (Thompson, 1994, p. 118; Van Dijck 1998, p. 23; Petersen, 2001; Petersen, Anderson & Allan, 2005). Although journalists sometimes introduce new metaphors to keep their writing interesting and accessible to the lay public (Hellsten, 2005), the metaphors used in the West tend to be the same between countries since they rely on the same primary sources (Condit, 1999b; Condit, 2009). Even when they do not have the time or academic background to search beyond a few well-known sources or rigorously double check their scientific claims, journalists often appeal to

[45] Durant et al.'s study of British newspaper coverage (1996) found that (despite its promotional efforts) as of 1996 references to the Human Genome Project were infrequent compared to other applications of the new genetics, and that none of the non-specialists interviewed in their focus groups were even aware of the project. Instead, most thought of genetic testing's roles in screening for specific disorders like Down syndrome and cystic fibrosis as well as its use in forensic DNA profiling (p. 240-242).

scientific impartiality to protect themselves from criticism (Thompson, 1994, p. 117; Petersen, 2001).

Journalists can only use what is given to them or validated by credible sources like scientists, so they typically seek out scientific information from governments, hospitals, universities, well-known journals, political leaders, well-known experts, professional associations and these groups' press conferences and press releases. These are often conducted by public relations experts, who have been employed by scientists for many years to strategically improve their image, gain a competitive edge against other scientists and promote their research to funders and publics (Nelkin, 1994; Thompson, 1994, p. 117; Nelkin, 2001; Petersen, 2001; Doring, 2005; Gogorosi, 2005; Petersen et al., 2005). Furthermore, journalists are often pressured to write according to the values of the scientific community rather than representing the public's concerns, and are motivated to maintain a good relationship with their sources (Petersen, 2001).

Hence, the perspectives expressed in lay media tend to reinforce both the perspectives of well-established scientists and the purported objectivity of science (Petersen, 2001). The use of only these scientifically credible sources has resulted in systematic underutilization of contexts from the humanities, including culture, sociology, history and ethics (Logan, 1991, p. 48). As a result, the media in the countries studied by Petersen (2001) rarely lent attention to related social or ethical concerns which may have been more personally relevant to the public (Petersen, 2001).

When scientists themselves appear in the news, they typically explain why discoveries are significant to the viewer or to correct the public's perceived misconceptions of science (Conrad, 1999; Petersen, 2001). Scientists have a number of additional motives for the intentional use of metaphor, including the need to improve the public image of genetics and to promote the importance, legitimacy and sheer amount of their own work (Nelkin, 1994; Nelkin, 2001; Petersen, 2001; Hellsten, 2005; Ratto, 2006). Publically-funded researchers may feel the need to justify their use of taxpayers' money, while those from private industry may use compelling metaphors to raise their investors' expectations of profit and boost the company's stock price (Petersen, 2001). In 1994, Nelkin stated that scientists had become much better at preparing imagery for use by the media, and by 2005, Petersen et al. expressed their belief that scientists were increasingly becoming "expert storytellers". Scientists often use pop culture metaphors and even fiction to communicate complex concepts to diverse and increasingly distrusting audiences (Nelkin, 1994; Petersen et al., 2005), as this helps laypersons to integrate new knowledge with

their pre-existing experiences (van Dijck, 1998, p. 21; Calsamiglia & Van Dijk, 2004). Their strategic use of rhetoric, including metaphors, has been remarked on by numerous authors including Nelkin (1994) and Van Dijck (1998, p. 11-12, 23).

Science is often inaccurately seen as empty of metaphors (Gronnvoll & Landau, 2010). However, many metaphors are also used within scientific discourse (Van Dijck, 1998, p. 22; Richards, 2001). For instance, when discussing the genetics of human behaviour, Nordgren (2003) even argues that metaphors appeared to be "vital for the scientific understanding of the role of genes". According to Nijhout (1990), metaphors like "control" and "program" "are no more than working hypotheses", but many biologists forget that metaphors represent the model rather than the actual mechanism and take them literally. The "program" metaphor, in particular, has required qualification in popular science in order to avoid contradicting empirical findings (Nordgren, 2003).

Similarly, metaphors like "code", "book" and "map", as well as their associated verbs, are common in scientific journals, popular science magazines, and news media alike. It is possible that this is because all derive their terminology from the same scientific discourse (Petersen, 2001).[46] However, it is also possible that geneticists' use of language merely reflects the available models and meta-narratives, ones which are generally shared by the public due to shared cultural values and ideologies (Roof, 2007, p. 10, 15).

Nordgren (2003) found no difference in the type of metaphors used by the same authors in texts intended for the public compared to texts intended for other scientists, suggesting the importance of understanding the historical context of this language. Others report that metaphors become even more "colourful" when articulated to the public (Weigmann, 2004), and that genetic metaphors are used much more frequently and more flexibly in writing for laypersons than in purely scientific discourse (Hellsten, 2005, Nerlich & Hellsten, 2004). Van Dijck (1998) comments that "the choice of metaphors is always strategic" (p. 23), and according to Condit & Condit (2001), there was "every reason" to believe that metaphor will continue to be used as a way to articulate genetic concepts to the public. Roof (2007) writes that:

[46] Interestingly, as the journal in which the actual results of the Human Genome Project were published, Nature used the metaphors of "map" and "code" in their conventionalized forms, as well as the language metaphors of "reading" and "writing" the "letters" of DNA, but never used "program" or "book of life" (Nerlich & Hellsten, 2004).

> "Although scientists engaged in DNA research long ago elaborated on and departed from the structuralist models that dominated mid-twentieth-century thinking, representations of DNA operation aimed at the general public today have retained their structuralist and primarily textual and linguistic quality". (p. 65).

This practice "allays suspicion and fear, encourages acceptance, fosters hope, and discourages more probing questions and the necessity of inquiry" (Roof, 2007, p. 65). This sort of outdated scientific terminology can return in the form of popular metaphor occurs because such figures of speech reach into "established stocks of cultural knowledge" which may be both relevant and accessible in the social context but no longer up to date with modern science (Doring, 2005). Current beliefs are often shaped by preceding conceptions from eras in which genetics mainly addressed single-gene disorders within clearly-defined groups (Durant et al., 1996, p. 238)

In this context, the news media generally fails to give an account of multigenicity (Petersen, 2001), and although they rarely suggest that gene variants are equivalent to disease this can nevertheless lead to a perception of "one size fits all" genetics whereby even mild predispositions are seen as guarantees (Condit, 2009; Gronnvoll & Landau, 2010). Indeed, opinion polls have frequently shown that between a quarter and a third of Americans (depending on the condition in question) endorse genetic determinism by believing that having a "gene for disease" guarantees being afflicted by it (Singer, Corning, & Lamias, 1998; Condit, 2007), perhaps due to the sheer simplicity of the idea (Moore, 2008).

The idea that a disease gene might be equivalent to the disease itself is very unhelpful from a public health perspective, since it would seem to deny the ability to protect one's health through behavioural or environmental means, even if the symptoms could perhaps be delayed (Hubbard & Wald, 1993, p. 12; Gronnvoll & Landau, 2010). Few interviewees have seemed to have integrated accounts of the relationship between genes and environment (Condit, 2007). Indeed, there is good reason to believe that a large segment of the public has limited genetic literacy[47], understanding the science in an oversimplified way and making them especially vulnerable to rapid changes in science and technology (Chapple et al., 1995; Condit & Condit, 2001; Johnson,

[47] It hardly needs to be pointed out that the word "literacy" itself implies the nature of genetics as language, although of course the term has been applied to other areas like climate and science in general.

Case, Andrews & Allard, 2005; Kaphingst et al., 2009; Klitzman, 2010; Reydon, Kampourakis & Patrinos, 2012). Klitzman (2010) recruited New York patients at risk for a genetic disease and found that they exhibited a number of scientific misunderstandings that reinforced each other, from the basics of where DNA is located and how it is inherited, to an overestimation of their own risks combined with high expectations for the effectiveness of genetic testing, diagnosis and treatment. Representations that cause people who are not at high risk to consume medical services may result in waste and unnecessary risk (Shiloh, 2006) through inappropriate healthcare decisions with respect to testing, treatment, coping, and reproduction (Klitzman, 2010). As such, many critics have criticized such promotional strategies as potentially socially harmful (Sidler, 2006).

In summary, many studies have shown laypersons to have relatively poor understanding of genetics in disease, a fact which is not helped by the factors which shape their simplistic presentation in the popular media. The images presented to the public carry significant consequences for both their beliefs about genetic disorder and their self-concept. Yet, as will be explained in the following section, patient understandings of these conditions tend to develop in a much more nuanced way than mere absorption of the broadly-communicated metaphors we have so far discussed.

Lay Representations of Genetic Disorder

The actual influence of news media on public thinking is not easily encapsulated. While it is important to convey correct medical models to patients in order to guide them towards appropriately beneficial behaviours (Toombs, 1992, p. 47), laypersons' interpretations of popular genetic metaphors are varied. They seem to be influences primarily by individual knowledge and personal experiences, followed by sociocultural context (Condit, 1999b; Condit et al., 2002; Nelkin, 2001; Petersen, 2001).[48] These factors include level of education, religious values and membership in religious communities, family interests and attitudes, and popular media including both fiction and various forms of news (Thompson, 1994, p. 113-114).

[48] Interestingly, when metaphors are offered to laypersons, the framing and order in which the domains are presented play a role in their effect (Condit 2009). Indeed, comparisons made in a different order (X is Y versus Y is X) seem to produce different understandings (Parrott & Smith, 2014).

Most rhetorical analyses in this area have studied media relaying messages in which scientists and other authority figures explained their discoveries, under the implicit assumption that the language they use is taken up by laypersons in an equivalent fashion. However, there is reason to think that the metaphors articulated by the public may have different functions and imagery than those used by scientists, despite the efforts of the latter (Gronnvoll & Landau, 2010). For example, critics have suggested that metaphors like "code", despite being widely used by professionals, do not successfully bridge the gap to laypersons (Gronnvoll & Landau, 2010).

The public tends to be more interested in the participants, social context and implications of science than the details of the science itself, so the scientific concepts in mass media often remain vague; the symbolic power of the language metaphor, for instance, relies much more upon its social meanings than its scientific ones (Nelkin, 1994; Calsamiglia & Van Dijk, 2004). As such, audiences have expressed a desire for the use of culturally meaningful language (Krieger, Parrott, & Nussbaum, 2012), and laypersons with genetic conditions seem more concerned about aspects of identity and potential stigma than about more scientific distinctions such as that of nucleic and mitochondrial DNA (Klitzman, 2009). Their goal is less to understand the state of scientific discovery and more to understand enigmatic perceived risks to themselves personally (Gronnvoll & Landau, 2010).

Rather, the public seems to choose metaphors from a very broad range of fields (including architecture, technology, medicine and politics) (Klitzman, 2009) based more on associated emotion (using what Condit calls a "biosymbolic" paradigm) than the specific, rationalist analogical parallels advanced by Lakoff and Johnson. However, they still reject metaphors that do not make any semantic sense to them (Condit, 2009). As Durant et al. (1996) wrote:

> "public understandings of the new genetics are not passive reflections of professional, scientific understandings; rather, they are active constructs, the products of multiply-mediated historical and cultural (including mass media) influences, which may be expected to diverge significantly from those professional understandings of science with which they coexist" (p. 236).

As a result, the information received is modified to allow it to be integrated into personal representations which are in turn part of larger belief systems. This can include beliefs in

personal responsibility for a child's disorder or religious notions like being chosen or blessed, and may be part of a coping strategy (Weil, 1991; Shiloh, 2006). However, people are capable of holding contradictory theories depending on the context (Chapple et al., 1995; Condit, 2007); some can simultaneously offer conflicting interpretations from both technical and nontechnical sources (Weil 1991). Some studies have indicated that patients end up mixing lay and scientific explanations through the use of their own "mental models", motivations and expectations, with subjective representations more influential than objective facts (Shiloh, 2006; Klitzman, 2010).

According to self-regulatory theory, patients' decision-making processes and coping mechanisms depend on these representations of illness, including genetic disorders. These arise not just from the meanings assigned to direct experience or that of friends and family, but also on depictions in the media, cultural beliefs and language (Leventhal, 1997; Shiloh, 2006). Representation involves identifying the threat, its cause, its timeline, its effects and its treatability (Shiloh, 2006), which can include notions relating to behavior, environment, biological, mystical and psychosocial factors. Interestingly, biology tends to be grouped with the latter two as being "hidden" and having "wonder, uncertainty and little perceived control", causing genetic disorders to be perceived as "abstract, uncontrollable and inexplicable" (Shiloh, Rashuk-Rosenthal & Benyamini, 2002; Shiloh, 2006),[49] likely allowing patients great leeway in how to interpret their experiences of genetic disorders.

These personal experiences and stories became even more important as people with genetic illnesses began to survive much longer than they had done before viable health care was available for those conditions (Nowaczyk, 2012). Narratives are critical to understanding how knowledge of one's own genes affects their self-perception (Quaid, 1994, p. 5), and Nowacyzk (2012) comments that "Clinical genetics, like few other specialties, is a specialty of narratives". DNA has even come to function as a sort of autobiography,

> "first, the sense in which genes themselves may decisively influence one's identity and life course; second, the sense in which gene tests may influence one's sense of identity

[49] Medical professionals have also been noted for a tendency to treat genetic disorders as different from other types of illness (Quaid, 1994, p. 6). These narratives can depend greatly on the storyteller's own experiences, views and fundamental values (Thomson, 1994. p.173-174), even when she or he is an expert in their field. For instance, descriptions of Down syndrome and cystic fibrosis were more technical and negative in tone when health care providers were speaking to prospective parents considering prenatal testing when compared to the parents of affected newborns (Lippman & Wilfond, 1992).

and life course; and third, the way in which the cultural construction of genetic information may influence identity and life course" (Couser, 2001).[50]

The effect of chronic genetic illness has been described as "biographical disruption" (Petersen, 2006); unlike most narratives of illness, people with genetic disorders rarely "get well". Rather, narrative closure can be achieved by conceiving the experience as an active quest to "find some kind of meaning in illness" (Nowaczyk, 2012). Indeed, one of the recurring themes in patients' experiences of genetic conditions is beginning with a "quest" for information about their illness (Petersen, 2006). It is therefore extremely important that we devote energy to understanding laypersons' beliefs about genetic disorder and how they affect individuals' conceptions of the self.

Genetics can affect both subjective individual identities and social roles as the sick person (Klitzman, 2009). Some researchers have found that carriers, people with risk predispositions and even noncarriers may also experience threatened identity in various forms (McConkie-Rosell, Spiridigliozzi, Melvin, Dawson & Lachiewicz, 2008; Klitzman 2009). As Petersen (2006) puts it, genetic conditions can be seen as "posing a substantial threat to one's sense of self". According to McConkie-Rosell and DeVellis (2000), there are four mechanisms by which genetic information acts on self-concept:

- "altered perception of genetic identity (the individual's perception of defects in his/her inherited endowment)
- diminished social identity (the perception of self as less adequate and worthy in the interaction with others)
- altered perception of health (self perceptions as defective through the connections between the self, health and illness) and
- threat to the parental role (perceived barriers to attain the valued normative role of parenthood)" (summarized in Shiloh, 2006).

[50] Indeed, Couser (2001) writes that knowing of one's predisposition for disease "can induce intense, even excruciating, self-consciousness" that lends itself to literal autobiography. These works can be very useful both in helping the authors to "repair the damage that illness wreaks", as well as in helping others to understand the experiences of individuals with genetic disorders (Nowaczyk, 2012).

To these, Petersen (2006) adds the potential loss of gender identity. In response to these threats, patients with genetic disorders try "thinking of alternatives, wondering if it was someone else's problem, and calculating the odds of avoidance" plus wondering about the removal of the condition (Armstrong, Michie & Marteau, 1998). Eventually, affected individuals generally attempt to regain "some sense of normality" in the context of the disease community (Petersen, 2006). Yet many newly-tested patients with genetic disorders will already have had first-hand experience of the illness. Most people at risk for certain types of genetic disorder (especially autosomal dominant ones) have seen the condition's medical, psychological and social effects upon their close relatives; as Quaid (1994) writes, "Genetic diseases are truly a family affair" (p. 6). Genetic identities tend not to be wholly individualistic (Klitzman, 2009), and the narratives of people with genetic conditions "are almost invariably partly stories about the lives of others in the family" (Petersen, 2001). Family history can even affect the identity of people without symptoms and shape the meaning individuals assign to more or less universal symptoms (Toombs 1992, p. 36; Klitzman, 2009). As Couser (2001) notes, the border between disability and "normality" is particularly fluid when one only suspects having a gene.

Perceptions of hereditary illness can also be influenced by the number of relatives affected, as well as the patient's perceived similarities to those relatives (Walter, Emery, Braithwaite & Marteau, 2004). For example, shared personality, physical characteristics or even a close relationship can be seen by laypersons as indications of shared genes (Shiloh, 2006). When informed about their conditions by genetic counselors, the patients studied by Armstrong et al. generally made use of their family's pre-existing stories about the inheritance pattern of the condition and/or looked for physical representations of the illness, which "enabled the problem to be located in others" rather than as individual responsibility (Armstrong et al., 1998)

Unlike diseases caused and/or acquired later in life, genetic disorders and risk factors are generally present as part of the patient's genome from conception onwards, and hereditary illnesses naturally carry health implications for other family members including one's present or future children (Quaid, 1994, p. 6; Green & Botkin, 2003). Illness affects not just our experience of ability, intentionality and relationship to physical space, but also the significance of past, present and future (Toombs, 1992, p. 62-65, 68-69); as Lakoff and Johnson (1980) put it, "we

seek out personal metaphors to highlight and make coherent our own pasts, our present activities, and our dreams, hopes and goals as well" (p. 133).[51] Raspberry and Skinner (2007) wrote that:

> "the experience of the genetic body is situated in real time and familial relations . . . with reverberations that extend across generations into the past and future. Specifically, experiencing the genetic body confers a sense of predetermined identity and profound historicity."

They describe genetic diagnosis as an opportunity for families to "re-envision themselves, their bodies, and their health" as well as to reconsider their definitions of normal. Hence, testing often marks a nexus for the reconstruction of narratives both forward and retroactively, as the patient situates themselves with respect to earlier generations and those yet to come. Of course, this depends on the sort of genetic disorder which runs in a family (Raspberry & Skinner, 2007). Mutations less predictive of disease were found to allow patients more flexibility in how they perceived influences on their health, with more patients at risk of breast cancer than those at risk of Huntington's disease or alpha-1 antitrypsin deficiency patients citing the social and environmental factors in their health (Klitzman, 2009).

These sorts of perceptions of susceptibility influence people's desire to receive genetic testing, exert personal control over their health through methods like diet and exercise, and likely increase their beliefs in the efficacy of genetic research (Parrott, Kahl, Ndiaye & Traeder, 2012; Parrott & Smith, 2014). Preventability has been found to increase willingness to undergo predictive genetic testing (Shiloh, Ben-Sinai & Keinan, 1999). Somewhat ironically, the public also tends to see diseases diagnosed through genetic testing as less preventable (Shiloh, 2006); Parents whose children were tested for genetic disorders describe the results as more "definitive", "firm" and "clear-cut" than other types of tests (Raspberry & Skinner, 2007).

[51] One of the metaphors specifically discussed by Lakoff and Johnson is that "life is a story", which they describe as "rooted deep in our culture. It is assumed that everyone's life is structured like a story, and the entire biographical and autobiographical tradition is based on this assumption", including the assumptions of identifiable participants, parts, stages, linear sequence, causation and overall purpose (Lakoff and Johnson 1980, p. 172).

Predictive results like the detection of unpreventable diseases like Huntington's disease can seem like a "preview of upcoming chapters in people's life stories that threaten their own authorship" (Juengst, 2004)". As such predictions prove accurate, inefficient coping behaviours may be revised to better reflect the objective facts. Many develop "self-protective" misconceptions as part of the coping process, and these are especially important when there are no methods for prevention (Shiloh, 2006), as is generally the case for Mendelian genetic disorders.

Beliefs also appear to affect health outcome and well-being in some illnesses, including Huntington's disease, cystic fibrosis and some hereditary cancers (Cooper, 1997; Helder et al., 2002; Shiloh, 2006). People who subjectively view disorders as more severe are correspondingly more likely to make reproductive plans accordingly, including aborting an affected fetus (Shiloh, 2006). Rarer diseases can be perceived as more severe, while those relating to oneself can be perceived as less severe as a way of coping through "minimization" (Croyle & Jemmott, 1991; McConkie-Rosell, Spiridigliozzi, Sullivan, Dawson & Lachiewicz, 2001; Shiloh, 2006; Klitzman, 2010). Coping mechanisms for risk diagnosis tend to involve the assertion of control, with failure potentially being transmuted into self-blame. Yet despite the notable exception of the pregnancy decisions discussed earlier at page 31, genetic disorders tend not to be seen as not blameworthy in and of themselves (Shiloh, 2006). Indeed, more "inherited" conditions were also seen as less contagious and were therefore more conducive to other peoples' interactions with those affected when compared to other types of disease (Bishop, 1991).

Despite the metaphors of genetics presented to the public, they tend to place more focus on the human rather than the technical side of such topics and to express metaphors reflecting personal experiences and emotions. These likely depend largely on the numerous potential effects these conditions can have upon self-concept. In the following chapter, I discuss the two overarching types of metaphor into which I believe these representations fall.

Chapter 4 – Relationships between Self and Genes

It is the central finding of my analysis that metaphors of genetic disorder can be broadly grouped into two principal, and fundamentally opposed, conceptions of the relation between one's genes and oneself. These can be summarized as "genetic disorder versus self" and "genetic disorder as self", and I suggest that incompatibility of the two is responsible for some of the problems involved in articulating these illnesses in narrative. These categories can be said to emerge from the two metaphysical ways to classify perceptions of disease - ontologically as distinct entities, or physiologically as traits of the body. The previously discussed notion of genes as causative in and of themselves represents a "neo-ontological" version of the former type of thinking (Stempsey, 2006).

I do not believe I am the first person to identify this underlying duality with respect to popular conceptions of genetics. As Shiloh (2006) notes, some representations are compared to the self and others as external. Examining several popular and academic texts on behavioural genetics, Nordgren (2003) found that the metaphors used fell into the two overarching categories of "text" (including program) and "agent" (including creator), and that there was considerable – potentially productive – tension between the respective passivity and activity of these two categories. I suggest that the former can be understood as a representation of genes as part of one's identity and the latter as independent entities.

Genetic Disorder versus Self

Ontology is the branch of metaphysics which studies being and existence. Hence, the concept of genetic mutations existing separate from and possibly in opposition to the self is an ontological metaphor – one which asserts that the disorder in question is an entity in its own right rather than a quality or a description of another entity.

According to Lakoff and Johnson (1980), ontological metaphors for a thing "allow us to refer to it, quantify it, identify a particular aspect of it, see it as a cause, act with respect to it, and perhaps even believe that we understand it." (p. 25-26). For instance, genes can be conceptualized in a similar way as other things inherited from our parents and ancestors (Richards, 2001). Pollack (1995), for instance, describes alleles as "gifts we receive from our parents and give to our children" (p. 40). A common example of this framework is language that

asserts “entities within a person”, as seen in phrases like “He has a pain in his shoulder” and “Don’t give me the flu” (Lakoff and Johnson 1980, p. 50). Indeed, Lakoff and Johnson (1980) assert that “Each of us is a container, with a bounding surface and an in-out orientation” (p. 29).

DNA is, of course, a material that can quite literally be found within the body. Terms like “found”, “located”, “isolated” and “pinpointed” are frequently used in the media to describe the identification of genes involved in disease. These may give a sense of genes’ presence, minuteness and location within the human body, diminishing the perceived role of external factors (Petersen, 2001). Furthermore, conditions like Down syndrome which result from the presence of additional chromosomes are easily conceptualized as entities, making them easier to fit into this type of metaphor than, for example, genetic disorders in which one nucleotide is exchanged for another in the way of a “typo”.

According to Hall (2003), the process of spatialization allows entities to be manipulated and “transforms the body into a bounded [and] potentially knowable space”, telling a “spatial story about the genes and the body” that objectifies our imagining of the genome and changes the story of genealogy into one relating genotype to phenotype. This results in the fetishization of the gene as a materialized entity, powerful yet physical, while ignoring the rest of the body (Hall, 2003). These metaphors lack any equivalent for biochemistry and interactions between genes and other aspects of biology (Richards, 2001).The view of genes as real, material, objects unrelated to intangible environmental effects carries the “allure of specificity” (Conrad, 1999) and has been argued to appeal to Western notions of individualism and personal responsibility (Conrad 1999; Nelkin & Lindee, 1995).

When applied to genetic variation in disease this conception also affects the way in which medical narratives are articulated to health care providers. Patient experiences are structured differently in their narratives when the condition in question is conceived in ontological terms rather than as a mere symptom (Toombs, 1992 p. 48). Toombs (1992) further argued that, under the prevailing biomedical Western model, scientific constructs overtake patients’ experience such that illnesses must be pathologically classified to be considered valid (Toombs, 1992, p. 39, 45). After diagnosis the symptom or experience may be thematized as the object disease, even without access to the direct diagnostic information from which the physician or geneticist made their decision (Toombs, 1992, p. 41). Patients often attach more value to the official diagnosis than to their feelings, so the labeling of the disorder can have a great effect on how they

subsequently feel about the illness and themselves. Furthermore, this ontological view deflects research from the way in which diseases are integrated into individuals' life experiences (Hubbard & Wald, 1993, p.58-59).[52]

Roof (2007) argues that we imagine the activities of genes based on our preconceptions of "the way stories usually go". Narrative, she argues, "reproduces basic ideologies about life, behavior, motivation, values, proper gender roles, and the necessity for closure" on a subcellular scale (p. 117). As Pramling and Säljö (2007) write, "intentionality, animism, and reification . . . are resources for telling coherent stories."

In 1951, the Nobel-winning geneticist H.J. Muller observed that early theorists of pangenes and other sorts of self-propagating particles were "still, subconsciously, so close to the ancient lore of animism" (p. 95), the belief that parts of the natural world have souls of their own. Prior to the 1970s, individual genes were rarely the object of attention (Conrad, 1999). During that decade, though, they often were depicted as "tiny agents" with "intellectual human powers" that acted as the protagonists of a narrative, while simultaneously remaining unpredictable and mysterious (Van Dijck, 1998, p. 64, 91-2). Apparently, little has changed since; Liakopoulos (2002) noted in his review of biotechnology metaphors in the British media that "genes are portrayed as individual organisms [with] physical or behavior characteristics", which withdraws personal responsibility.

In some ways, genes have become even more personified: as the medical paradigm took hold, terms like "culprit", which were once attached to individuals who had negligently reproduced and passed down faulty genes, were used to describe "guilty" genes themselves (Condit, 1999a, p. 190; Couser, 2001). Condit (1999a) describes this as "The movement of potentially discriminatory descriptors to the microscopic level," writing that "Persons with genetic disorders were now most often described as afflicted or affected by a disease, rather than as diseased or unfit." (p. 190).

Indeed, one of the key contributor to paradigms of genetic determinism has been the inaccurate notion of genes as "active agents" (Fox Keller, 1994, p. 90), which tends to result in their being attributed with human motivations and psychology of their own as a way to help us

[52] The ontological view has also been disputed by such prominent figures as Tolstoy and Rilke, whose literary works covered topics like the disrupted relationship between self and disease (Hubbard, 1993, p. 59).

identify with them (Roof, 2007, p. 116-121). Several authors have described the personified gene as "homunculi" rather than mere contributions to probability (Fox Keller, 1995, p.xvi; Roof, 2007, p. 74, 118), and Roof (2007) argues that "The inevitable effect of agency is consciousness" (p. 116). Genes are still referred to as "the cell's brain" on occasion (Fox Keller & Winship, 2000, p. 47), and Fox Keller (1995) calls the paradigm that the cell is organized and maintained by genes the "genetic psyche" (p. 97). In their analysis of genetic metaphors in Swedish media, Pramling and Säljö's (2007) found that "mini-narratives . . . about miniature human-like actors" were the most prevalent, giving the examples of genes said to "know", "remember", "decide", "steer", be "alert" and "prepared", and to do "both evil and good".

Some authors have noted that "good" and "bad" genes have morality attached to them (Nelkin, 2001). People tend both to distance the self from the body and the body from the gene – and that which is outside the self is considered untrustworthy (Gronnvoll & Landau, 2010). This has led participants in audience studies to express a need to "stand vigil" over one's body to product it from the negative influence of genes (Lippman, 1992; Gronnvoll & Landau, 2010). Genes are often anthropomorphized as "rogue" or "killer" in this respect, an image which also plays the role of casting the scientist as a "warrior" protecting the patient (Petersen, 2001). Furthermore, "bad" genes have tended to be the first ones studied and covered in the news (Couser, 2001), meaning that much of public thinking about genes in general has been shaped by negative associations. The easier a gene is to associate with disease, the earlier and more prominently it may make its way into the lay consciousness.

As Lakoff and Johnson (1980) wrote, by personifying our adversaries we shape our way of thinking about and acting towards them (p. 34). Thus, laypersons tend to see genetic disorder as a battle between disease in the form of genes and the self in the form of behaviour, via personal attributes such as strength of will, or, as previously mentioned in the 1970s section, luck (Wexler, 1979; Klitzman, 2009; Gronnvoll & Landau, 2010). Complex interactions between genes and environment rarely enter into the battle metaphor, and this sort of battle tends to be seen as prolongable but ultimately futile (Gronnvoll & Landau, 2010). Thus, bodies have been argued to be the subject of a "division" (Everett, 2003), which Sharp describes as "bodily fragmentation" (Sharp, 2000). By redirecting focus to individual body parts, this fragmentation can complicate the relationship of the body narrative to narratives about personhood and responsibility even as it generates value for the biotechnology industry (Rabinow, 1992).

Authors in various disciplines have suggested that scientists and physicians tend to view "the" human body and its constituents as abstract scientific objects, or representatives of disease categories, rather than an individual (Zaner, 1981, p. 53; Toombs, 1992, p. 70-71, 77-78, 87; Quaid, 1994, p. 11). Likewise, patients may express this perceived or attempted division through language like "the" rather than "my" body part, perhaps as a way of minimizing trauma (Cassell, 1985, p. 55-65). Indeed, Klitzman (2009) noted that some patients tried to view their genetic conditions as restricted to the organs most affected by them; one stated that "If I didn't have these two lungs, I'd think of myself as healthy". This sort of division can result in the loss of the intimate awareness of the limitations and implications of one's body which characterizes embodiment (Zaner, 1981, p. 52; Toombs, 1992, p. 60). In the case of genetic disorder, this is exacerbated when those administering care do not have personal experience with the people who have that condition, shifting focus from personal, social and environmental factors towards pure biology (Quaid, 1994, p. 11) The body thus presents itself as a "faulty" "mechanical, physical object" or, in technologically advanced cultures, a "physico-biological 'thing' among other objects to be felt, seen, and acted upon" (Toombs, 1992, p. 59, 71, 91).[53]

The Western biomedical approach has thus been criticized for seeing the body as a closed system containing specific internal factors that cause bodily malfunctioning – ones that avail of technological treatment and that conveniently coincide with the socioeconomic interests of academics, universities, businesses and government (Lippman, 1992; Gannett, 1999). Diseases that affect the lives of large numbers of people but are not medically tractable, like Down syndrome, are seen as less worthy of research (Gannett, 1999).

The relationship between body and self is most complicated in chronic illness, during which the body is more likely to be seen as transformed into the "diseased body" (Toombs 1992, p. 75-6, 91), and this is quite relevant to patients with inborn errors of metabolism and other unrepairable genetic disorders. Klitzman (2009) suggests that his interviewees expressed "a yearning for coherence" in the face of finding that parts of themselves has 'altered" against their will. In particular, symptoms like motor disturbances, which characterize well-known inherited

[53] Jean-Paul Sartre discussed this topic in *Being and Nothingness* (1956). While we normally overlook our bodies rather than experiencing them as organisms, reflecting actively on pain or illness transforms it into a psychic object separate from the body. Knowledge derived from others allows us to apprehend this psychic object as an objective and objectified disease. This results in an alienation from the body, the experience of what Sartre called "Being –for-the-Other" (p. 429-30, 440, 461, 466).

disorders such as Huntington's disease, can make patients see their bodies as oppositional and malevolent, having a will of its own. A loss of bodily control can contribute to a sense of aloneness, especially in highly technological cultures, making this an important threat to selfhood (Toombs 1992, p. 88-97). Roof (2007) writes that "The post-DNA subject has perhaps ironically become a composite of forces perceived as 'outside' . . . a pastiche of virulent fragments" (p. 97). This is a very appropriate adjective.

We have established that ontological conceptions of illness which arise from certain experiences cast the body as a spatial container in which discrete objects of disease are located, especially as validated by the process of medical diagnosis. Since genes are frequently depicted as tiny living beings opposed to the individual patient, it is natural that one of the most common metaphorical representations of genetic disorders and risk factors seems to be that of the virus. The following section examines the historical background and elements of the disease experience which have led genes to be readily conceived in a model analogous to that of germ theory.

The Virus Metaphor

Genetic disorders can be particularly difficult to understand and rationalize because they are treated as concrete despite being invisible in everyday life (Chapple et al., 1995; Shea, 2001). Historically, evil has been associated with invisible and abstract entities like demons and curses like the "evil eye", all of which displace control over human fate (Nelkin & Lindee, 1995, p. 83). Holm (1999) suggests that the "act of boundary breaking" needed to bring hidden information from within the body to light gives us a sense of something unpredictable, "valuable and potentially dangerous" like the secrets common in the world's folk tales (1999). Langauge like "secrets" and "mysteries" are popular in media representations of genetics, often at the suggestion of the scientists themselves (Petersen, 2001). Indeed, Van Dijck (1998) associates genetics with the age-old "desire to penetrate the invisible to turn it into the knowable" (p. 130). But the only way for most of us to know the invisible and intangible is through "language and representation" (Everett, 2007).

From this point of view, thinking of genes as viruses makes sense, since they are another common, invisible, intangible and internal health threat that fits both the analogy and the emotional valence of the threat (Condit, 2009). Gronnvoll and Landau (2010) conducted a study in three cities in the southern United States during which some participants, rather than merely

describing genes as similar to viruses, expressed the two concepts interchangeably. Indeed, it was the most commonly-expressed metaphor in their study. Genes were seen as foreign, invasive, separate from the otherwise healthy human body, and yet within it waiting for the right moment to strike in the same way viruses come out of dormancy (Gronnvoll & Landau, 2010). Likewise, in audience studies conducted in Georgia by Condit (2009), the most frequently offered metaphor of genetic disorder was the ontological one of the virus or communicable disease (Condit, 2009).

Parrott et al. (2012) demonstrate that metaphors that produce "threat beliefs" influence "essentialist beliefs". The virus metaphor is, of course, rather inaccurate in the case of risk predispositions, given that these genetic variations are not necessary to disease the way viruses are (Condit, 2009). Genetic disorders do not, of course, meet criteria equivalent to Koch's famous postulates for bacterial causation of disease (Stempsey, 2006; Smith, 2007, p. 95-96); as Condit (2009) points out, one does not need a "gene for heart disease" to get heart disease.

Pernick (2002) comments on the relation of the information metaphor to those of being "communicable" and the notion of hereditary "germ plasm". Certainly, the model of germ theory is readily used to interpret genetic disorder (Juenst, 2000, p. 138). Indeed, numerous authors have compared the explosion of the discipline of genetics and process of geneticization in the 20th century to the emergence of germ theory in the late 1800s. Both paradigms shared a focus on etiology arising from a specific and identifiable cause, shifted study to the internal environment of the body independent of environment, and resulted in the human body being frequently articulated in metaphor as a machine with individual replaceable parts (Conrad, 1999; ten Have, 2001; Melendro-Oliver, 2004). But unlike germ theory, genetics became credible long before it had produced useful health results (Conrad, 1999).

The reification of disease under germ theory presented them as separate and threatening to the patient, just as genetics suggested a localized target of the "genetic lesion" for treatment (Juengst, 2000, p. 136-137, 140; Juengst, 2009, p. 138). It is also likely that the identification of bacteria as causative agents of disease lent credence to the idea that minute things could have enormous health effects (Hubbard & Wald, 1993, p. 3). As a result, the uptake of genetics was facilitated and complemented by the pre-existing paradigm of germ theory (Conrad, 1999); and was initially portrayed in bacteriological terms, including the introduction of terms like "carriers". This resulted in similar fears and stigma, including the image of the "unclean" (Paul,

1998, p. 160; Juengst, 2000, p. 137-140, 146; Lemke, 2002). Even in the present decade, *BRCA1/2* mutation carriers often describe guilt about introducing their "dirty" genes into "clean" families (Werner-Lin et al., 2012).

The comparison between disease-related genes and microorganisms developed further during the 1970s, during which time negative metaphors were prominent and there was significant concern about harmful genes escaping from labs like bacteria or viruses. Like germs, genetic variants were often seen as a sort of invading army, but unlike germs, they were seen as an inherently internal threat (Montgomery, 1991; Van Dijck 1998, p. 91-2). However, these metaphors were short-lived compared to others (Van Dijck 1998, p. 181), and the conceptualization of genes as germs dissipated to some extent after the 1970s. This was perhaps due to new findings about the extent of genetic variation and perhaps due to the rise of immunology (Paul, 1998, p. 167; Lemke, 2002). The phenomenon of AIDS may later have helped return the virus to metaphorical prominence, and indeed several of the laypersons Condit (2009) interviewed explicitly referred to HIV as a comparison to genetic disorders (Condit).[54]

Despite this, it is at least mildly suspicious that the study claiming to be the first to report the prevalence of the virus metaphor came out only five years ago (Condit, 2009). One explanation might be the increased public attention to viruses as a result of the H1N1 "swine flu" pandemic in that year. The fact that such metaphors are articulated "tentatively" in a "groping" manner (Condit, 2009; Klitzman, 2009), might also suggest that participants are not repeating a specific ingrained metaphor so much as rapidly developing one to meet a underlying conceptual framework that favours ontological conceptions of genetic disorder.[55] If this is true, we would expect to see other metaphors of genetic disorder as discrete objects articulated frequently in interviews with laypersons.

Indeed, the second most common metaphor noted by Gronnvoll and Landau (2010) was also ontological: the rather inaccurate image of a fire or bomb waiting to explode given the right circumstances. The time bomb, unlike the virus, was not a relatively new association – Kahn and Wexler, for instance, had previously described the time-bomb metaphor for impending risk of

[54] As the representation of the body's own defense mechanism, Lemke (2002) argues that immunological discourse promotes the establishment of a control regime rather than simply accepting a "pathological identity".

[55] Indeed, most laypersons expand their thinking to include more favourable behavioural influences on health when they are probed about their use of metaphor (Condit, 2007; Gronnvoll & Landau, 2010).

conditions like Huntington's disease that do not affect the body in the present but represent a future harm (Wexler, 1979; Kahn, 1991). Gronnvoll and Landau (2010) found that these lay viewpoints were "more complicated and less fatalistic" than expected, and occasionally seen in relation to environment: although the bomb metaphor, like the virus, is pessimistically conceived as separate from and damaging to the body, the authors note that the image of the smoldering fuse implies a single potential harm that might be averted, rather than a guaranteed harm.

The focus on the internal body imparted by ontological conceptions of disease carries the metaphor of incorrectly programmed information or of a malfunctioning machine: simplified, objectified, composed of "tiny interchangeable parts" with distinct roles, and potentially repairable through gene therapy (Toombs 1992, p. 71; Lupton, 1994, p. 61; Rosner & Johnson, 1995; Stacey, 1997, p. 159; Conrad, 1999; Hall, 2003; Melendro-Oliver, 2004).[56] This may give laypersons the idea their genes can be controlled (Condit, 2004), raise hopes about treatment (Petersen, 2001), or create misperceptions about the predictive accuracy of genetic tests (Richards, 2001).

The ontological approach allows us to feel that we have identified an intangible condition as part of one's corporeal body yet separate from oneself, allowing us to interact with it in a way we could not imagine if it were conceived to be an attribute of the self rather than an independent object. Yet from a narrative perspective, the externalization of disease can prevent it from being integrated into the story of one's own experience; when personified, it is the genetic variant which is acting out its own story of its own accord.

Stempsey (2006) has suggested jettisoning the concept of ontological disease altogether, recommending more developed thought about the implications of such concepts. While I don't believe this to be particularly plausible, I suspect such an accomplishment would result in a swing towards the type of metaphor discussed in the following chapter.

[56] The machine metaphor is robust and has survived a long time (Klitzman 2009) in various contexts. Mechanical concepts of "DNA repair" overlap with informational ones like "proofreading", even mixing in the same sentence on occasion (Limoges, 1994, p. 115). Syed et al. (2008) suggest that this sort of stability is due to "the relation between metaphors and models" but that mechanical metaphors may nevertheless be helpful for encouraging innovation in fields like molecular computing.

Genetic Disorder as Self

Despite the psychological appeal of the aforementioned ontological metaphors, it is impossible to fully dissociate one's self from one's body (Toombs 1992, p. 73). While one of the key images of the self versus genes paradigm was that of the futile battle, Churchill argues that the central idea of determinism is "The idea that we are our genes" in the first place (Churchill, 2002, p. 184), a conceptualization which would seem to preclude such opposition. Indeed, there is a persistent belief that one's genome can be the most personal and unique way of considering them (Richards, 2001). As Quaid (1994) wrote:

> "Information that one has, or is at risk for, a genetic condition is more intensely personal than information about an illness contracted as a result of contact with an external cause, such as a virus. Genetic information is widely viewed as saying something about who the person is at some fundamental, if unarticulated, level." (p. 5-6).

One of the oldest ways of thinking about disease is the doctrine of intrinsic, "constitutional pathology", which dates back to Galen and imbalances in the four humours. Juengst (2000) suggests that, unlike the category of monogenic disorders which lend themselves to specific causes, vague predispositions, susceptibilities and risk factors are readily interpreted in a constitutional manner, affecting the whole body in a manner more open to personal agency (p. 141-146) and shifting personal responsibility further toward the patient (Juengst, 2009, p. 140).

One of the constitutional theories which survived the turn to ontological concepts of disease in the 19th century was the metaphor of "bad blood" (Chapple et al., 1995; Juengst, 2000, p. 141-143). Before DNA was well-known, blood had represented an animating force akin to the soul, as well as kinship within families and ethnicities, performing a metaphoric function in social structures which suggested a metaphysical difference between groups. Blood type (which was discovered in 1900) was sometimes used for identification as well as paternity testing. Both of these characteristics have now been subsumed into the purview of DNA, whose relation to parent-child relationships is not metaphorical and can be used to represent actual proof of relationship between individuals. Roof (2007) argues that the assertion of origin transforms into

that of identity since the two are intrinsically connected in narrative, contributing to DNA's position as "the almost sole representative figure of both individuality and identity" (p. 166-170).

Genetic testing readily differentiates people based on specific biological characteristics like blood type and sex rather than ones like age, helping to shape our conceptions of what traits are part of one's identity (Roof, 2007, p. 68). The notion of identity as it relates to identification through forensic DNA analysis has been further strengthened by its portrayal in crime and mystery media since the late eighties, when DNA "fingerprinting" first became admissible as evidence (Martin, 2007).

Soon after, official documents about the Human Genome Project began to promote the endeavour as what Doyle (1997) called "ontological research" with respect not to individual genes but to the human condition (p. 26). And as genetics became increasingly relevant to medicine, some have argued that genetic profiling will have a rising effect on personal identity (Everett, 2007). Raspberry and Skinner (2007) found that parents of affected children described the untreatable genetic disorders in their family as a fixed "part of who we are", although not in a singular or a negative way.

In accordance with the language metaphor, Roof (2007) even claims that "DNA, conflated with the idea of the gene, has taken over older metaphoric functions such as the name" (p. 164), while Shea (2001) suggests that the concept of the gene has more recently become "a new rhetorical foundation for manifesting cultural phenomenon in the individual". She describes genes as "exemplary boundary objects" that "allow for cooperation among diverse groups without necessitating consensus about meanings" (Shea, 2008, p. 69-70; see Rheinberger, 2000).

Indeed, the concept is sufficiently malleable to allow different communities to apply it in many different ways (Nelkin & Lindee, 1995, p. 16). Although the word "gene" is often used as a concrete noun in common parlance (Shea, 2001), it can be used to describe both the material, the information, and the "unit of inheritance", creating uncertainty between the name, function and ontology of the gene (Hedgecoe, 1999; Shea, 2001). As Hubbard and Wald (1993) explained:

> "To molecular biologists, a gene is a stretch of DNA that specifies the composition of a protein and may affect whether and at what rate that protein is synthesized, as well as sometimes affecting the synthesis of proteins specified by nearby genes. To geneticists,

> genes are parts of our chromosomes that mediate heritable characteristics of traits. To population biologists, genes are units of difference that can be used to distinguish various members of a popular from each other. To evolutionary biologists, genes are historical records of the changes organisms have undergone over time." (p. 11)

They conclude that in some contexts, "the 'gene' no longer has a physical meaning" (Hubbard & Wald, 1993, p. 43). Gross (1990) even suggests that "the sense that a molecule of this structure exists at all, the sense of its reality, is an effect only of words, numbers, and pictures judiciously used with persuasive intent" (p. 54). As such, it is no surprise that the conceptual and molecular definitions of the gene are both difficult to communicate to laypersons (Van Dijck, 1998, p. 21), even though DNA is easily the most prominent biological molecule in popular culture (Richards, 2001). The "gene" has from its outset been a figurative concept at the borderline of the material and the abstract, and it takes on additional function and meaning in the public sphere (Shea, 2001). Nelkin (1994) states that the metaphors chosen by scientists in lay media present the genome as "the essence of identity", further stating that "DNA is text without context, data without dimension. To locate complex human behaviour in a molecular entity is to ignore its social context". Thus, deterministic and reductionistic views of the body passed from science into lay culture such that people are increasingly seen genetically (Vieth, 2010). Doyle (1997) even writes that the force and reference of "genes are us" as a scientific statement "depends on the amputation of the body that it heralds" (p. 7). Nelkin and Lindee (1995) write that "the gene of popular culture is not a biological entity", but rather a symbol or metaphor used to construct social meanings in identity and relationships, including morality and fate (p.16).

It is true, of course, that DNA is a material molecule. Yet extracting information from DNA requires its physical transformation (Hedgecoe, 1999), reducing our ability to take it merely as itself. Tomasula (2004) calls DNA "both material and message, both the book and its content: a book that is its message embodied" (p. 249). Roof (2007) writes that two categories of gene as "organized operation" and "chemical material" have merged conceptually (p. 6-7), producing a fusion of "animate and inanimate", "conclusion and solution", "the particular and the general" (p. 31-32, 37), "the social and the biological, the symbolic and the material, species and individual, life and death, Being and speech, existence and signifier" (p. 49). These dualities are,

of course, rather rhetorically overblown. But much of it rings true with actual metaphorical representations of genetic disorder.

In "postmodern science" where abstract and unobservable phenomena are studied with the aid of information technology, they can be conceptualized using metaphor (Baake, 2003, p. 83; Sidler, 2006). It is quite easy to understand how genes could be understood in an abstract manner. DNA is indeed found in all parts of our bodies, not unlike the gemmules of Darwin's pangenesis theory (Roof , 2007, p. 189-190), and it is an extremely stable molecule resistant to environmental change (Sidler, 2006). As such, we tend to have the idea of the personal genome as a stable sequence of bases, even though there are billions of actual, individual cells in the body and they are replaced many times over (Chadwick, 2001, p. 341). Hence, Rolston (2006) states that our notion of the gene requires "cybernetic identity superimposed on a material identity"; to ask where a gene is would be like asking "where is the book, *War and Peace*"?

Elliot (1993) writes that "genotype is an abstract object, a complex universal, which may be instantiated in any number of different organisms". Monozygotic twins, for example, are called "identical" on the basis that they share the same genome (Richards, 2001; Elliot 1993). In a similar line of thinking, it has been argued that it is incoherent to conceptualize oneself without an inborn genetic disorder in the same way as an acquired disease, since being born with a different genome you would require you to be a different person entirely (Kahn, 1991; Zohar, 1991; Elliot, 1993; Chadwick, 2001, p. 345).[57] However, it is easier to think of individual traits, rather than identity as a whole, when genes (or additional chromosomes) are represented as discrete, ontological units capable of being manipulated (Condit, 1999a, p. 129).

It is not clear how much of the genome's "informational structure" or of the resulting traits would need to be modified in order to consider one's identity changed, drawing comparison to the classical sorites paradox. Is it material, structural, or causal continuity that is constitutive of one's identity? Elliot (1993) concludes that, while it is possible to say that genotype constitutes essence inasmuch as identity is determined by genotype, not all genetically determined things are essential to identity, and memory and other key components of identity are mediated by human or environmental influence upon the organism (Zohar, 1991; Elliot, 1993).

[57] Some philosophers emphasize "person-regarding" reasons for genetic therapy, with respect to the identity of the future person, over "organism-regarding" ones, with respect to the future human regardless of identity (Elliot, 1993).

Obviously, patient attitudes with respect to the problem of genetic disorder and identity vary greatly. Klitzman (2009) suggests that genetic conditions can result in "more fluid" correlates on which to base subjective identities when compared to other diseases, since there can be a range of "inchoate and fine-grained" symptoms, genotypes, phenotypes and test results to consider. As a result, there can be a great deal of fluidity and variation expressed depending on the time and the patient's present company. Many also have nuanced genetic identities that can seem to encompass mutually exclusive categories like "healthy" and "sick" or "normal" and "abnormal", depending on those contexts and on the extent to which the patient considers genetic risk a part of his or her identity (Klitzman, 2009).

On the furthest extreme, reducing identity to DNA such that it is only accessible through the biotechnology industry is perhaps equivalent to losing identity (Everett, 2003), and this conception is incompatible with the view of personhood advanced by virtue ethics. These implications also present a potential threat to voluntary, informed consent if patients are thought of in terms of a genetic model rather than as individuals with a personal narrative beyond that which popular metaphors insinuate they carry in their genome (Nordgren, 1998).

If the genome is considered to be the self, then negative traits (such as alcoholism), while being removed from blame for lack of self-control, may be simultaneously added to a person's very essence (Stempsey, 2006). Just as scientists and physicians often describe individuals in terms of their conditions (as "a diabetic" rather than a person with diabetes, or as adjectives like "a Down syndrome child" or "the Marfan syndrome family") (Toombs, 1992, p. 86; Thomson, 1994, p. 175), the disease itself becomes a property rather than an ontological entity of its own. Juengst (2000) writes that terms like "Down's babies", "sicklers," and "phenylketonurics" play a role in reducing identity to genetic status (p. 134).

Of course, it has been argued that patients tend not to adopt the appropriate attitudes towards illness until they do see themselves as, in the case of one of the examples just given, "diabetics" (Toombs, 1992, p. 47). Organizations like Alcoholics Anonymous have already played a large role in defining alcoholism as a permanent state of the individual rather than a mere behaviour (Hubbard & Wald, 1993, p.99). Patients with genetic disorders often reason that their genes constitute their identities and refer to themselves using the name of the disorder – for instance, some people with alpha-1 antitrypsin deficiency self-identify as "alphas" (Klitzman, 2009). This, of course, contributes to the formation of a patient community. But categorizing traits like

alcoholism as an illness can either, polysemically, protect the afflicted from stigma or generate further stigma depending on the context (Condit & Condit, 2001).

Some people feel they have genetic "conditions" rather than "diseases", suggesting the role of social input in making the distinction. Others feel that even the term "predisposition" implies limited personal control of health outcomes. Still another response is to direct attention to the practical consequences of their symptoms rather than the underlying genetic etiology by describing themselves as "disabled" rather than "diseased". Indeed, some support groups for genetic conditions actively encourage people to avoid using the word "disease" (Klitzman, 2009). Many identify themselves as asymptomatic, "healthy" members of patient communities (Hubbard & Wald, 1993, p. 38; Klitzman, 2009). Some people with *BRCA1/2* mutations even identify as "healthy" based on their present experience despite having previously experienced breast cancer as a result of their hereditary risk factors (Klitzman, 2009). However, the difficulty of defining one's own situation can be unsettling (Quaid, 1994, p. 15).

As mentioned in the first chapter, the image of the "mutant" has long been associated with "monstrous deformities". However, the term 'mutation" is pervasive in describing both harmful changes in one's genetic makeup and abnormalities in general. Questionnaires and focus groups have shown that the term carries "strong emotional coloration" which most laypersons respond to quite negatively, likely influenced by works of popular culture (Condit, Dubriwny, Lynch & Parrott, 2004) ranging from the radioactive monsters of the 1950s to the benevolent yet freakish X-Men. Indeed, Klitzman (2009) found that patients with a history of trauma and/or mental illness are more likely to describe themselves in negative terms as "mutants". One woman with a *BRCA1/2* mutation who had "a history of depression and suicidality" and saw herself as a mutant.[58] On the other hand, one engineer from the same study claimed he did not mind being a mutant because mutations are normal from an evolutionary standpoint. Although Limoges (1994) notes in this vein that "errors" result in the evolution that produced modern humans in the first place (p. 124), terms like "evolutionary error", "mistake" or "freak of nature" tend to feature in very negative ways, resulting in people seeing their condition as a "flaw" and making the

[58] Medical providers would be best to avoid the term, at least when introducing a patient to their genetic disorder for the first time (Condit et al., 2004). Based on her work with focus groups and community organizations, Condit (2007) has even suggested replacing the term "variant gene" with "version of a gene", partly because the former implies certain genes are not shared by all people and partly because it also sounds a little like the word "mutant".

construction of a genetic identity problematic (Klitzman, 2009). These sorts of beliefs about the self result a discounting of the subjective experience of the patient (Melendro-Oliver 2004) in favour of a negative abstraction that is likely irrelevant to their actual life.

In addition to psychological risks like anxiety, depression and "family discord" (Quaid, 1994, p. 9); threatened self-concept and other identity issues raise significant implications for patients' methods of coping, choices about disclosure of their condition, testing, and treatment (Quaid, 1994, p. 5; Klitzman, 2009). These effects appear to be in many ways independent of actual risk levels (Quaid, 1994, p. 5). Klitzman (2009) suggests they could depend on "the strength of previous narratives and the perceived penetrance, predictiveness, and lethality or severity" of the newly revealed condition, and Quaid (1994) writes that the "aura of hopelessness" that health care providers can transmit may cause people to feel "tainted" (p. 9).

Yet it is also possible to have a genetic disorder without knowing about it, and patients rarely want to change their self-perception (Klitzman, 2009). Does diagnosis result in the experience of having a hidden underlying identity revealed or simply adjusting the old one based on new self-perception (Armstrong et al., 1998, Chadwick, 2001, p. 346)? Genetic counselling has been described as the opposite of stigma, as it can give the sense of a real inner identity coming to the fore instead rather than a false external identity being forced upon the patient from outside (Armstrong et al., 1998). Genetic counselling can also help promote the unfamiliar idea that common genetic variations may not be "abnormal" and elicit more helpful conceptualizations of the relationship between that which is genetic and that which is actually pathological (Limoges, 1994, p. 116, 123). As such, ever since the 1990s, the need to address self-concept issues has been influential in the construction of policies such as those of the American Society of Human Genetics and American College of Medical Genetics (Shiloh, 2006).

In this section, we saw how the innate abstractness of one's genetic makeup has readily allowed it be conceived as equivalent to identity, tracing these associations from ancient notions of the bloodline to technological applications like the Human Genome Project. This has raised questions about how genetic deviations with respect to perceived norms affect patients' self-concept. Indeed, the resulting illnesses are often described as characteristics of people rather than mere aspects of their experience, with both motivational and stigmatizing effects. Alongside previously-discussed metaphors like that of language, this sort of metaphysical labeling of individuals carries many religious connotations, which are investigated in the following section.

Theological Metaphors

In *Metaphors We Live By*, Lakoff and Johnson (1980) wrote that:

> "Metaphor is one of our most important tools for trying to comprehend partially what cannot be comprehended totally: our feelings, aesthetic experiences, moral practices, and spiritual awareness." (p. 193).

Spiritual connotations are often attached to metaphor (Hellsten, 2005), and "superordinate categories" of metaphor like religion can be more flexibly mapped onto new domains (Liakopoulos, 2002). At the same time, Roof (2007) writes:

> "The primacy of textual analogies of various sorts – from codes to letters to history books – suggests that DNA is imagined as something that stands for something else in the same way that words or images refer to concepts or objects by representing them." (p. 73)

The two would seem to be naturally suited for each other, and indeed, the conception of self as genes is intricately entangled with theology. Western thinkers have attributed generative power to the word ever since Ancient Greece (Roof, 2007, p. 31). And like the inherently meaningless magic spell "abracadabra", the genetic code's lack of referent allows it to represent a sort of magic and an immediate cause of other things (Roof, 2007, p. 176-177); Roof adds that "As the Word, as a powerful, magic signifier, DNA becomes available to almost any account of the universe, the meaning of things, or the nature of life" (Roof, 2007, p. 173).

For instance, Nelkin and Lindee (1995) suggest that DNA acts as "a secular equivalent of the Christian soul" in popular culture (p. 2). The comparison does have a reasonable basis: like the soul, the genome is implied to be sacred, immortal and independent of the human body, yet essential to human life and individual identity. Like the promise of the Resurrection, the genome offers the potential for the return of a new body, albeit that of a clone. It comes across as a guide to moral order and destiny (Nelkin & Lindee, 1995, p. 40-1; Roof, 2007, p. 68) that internally

"bears the marks of good and evil" in the form of genes' influence upon us (Nelkin & Lindee, 1995, p. 40-1).

Of course, the correspondence between soul and genome is not so much a coincidence as a result of the fact that the latter has been frequently articulated in Christian imagery since at least the 1950s (Nelkin & Lindee, 1995, p 40; Kay, 2000, p. 36).[59] This practice is used as way to reconcile modern science and religion (Roof, 2007, p. 173), giving genetic research meaning and helping to justify it to religious people (Nelkin, 2004). Geneticists and journalists alike have described the genome in explicitly religious language such as a "Bible" and a person's "soul" (Nelkin, 1994; Nelkin & Lindee 1995, p. 8; Van Dijck, 1998 p. 45-49; Calsamiglia & Van Dijk, 2004), and some writers have even remarked that "new genetics" displaces the human soul into DNA (Sharp, 2000; Chadwick, 2001, p. 340-341; Richards, 2001). When Schrödinger's philosophy led him to believe that selfhood lay in the molecular structure of DNA, he compared this theory to "proving God and immortality at one stroke" (1992, p. 87). However, Schrödinger also wrote of genetic variation that "it would be entirely wrong to regard the original version as 'orthodox,' and the mutant version as 'heretic'" (p. 37).

During the 50s and 60s, as genetics reclaimed its image from the profoundly negative one of eugenics, religious figures were often quoted in support of the field (Van Dijck, 1998, p. 46-7). Genomics has been affiliated with the "Christian rhetoric of the Word" ever since the translation became standardized, simultaneously suggesting the possibility of some final ending or closure and casting scientists as some sort of "guardians" (Doyle, 1994, p. 67-8). Interestingly, some critics have suggested that the term "dogma" in Crick's central dogma was intentionally conceived as a religious metaphor for the carrying of an altogether different kind of "message" (Locke, 1992, p. 50-51; Van Dijck, 1998, p. 48-49), although Crick was an agnostic. His collaborator James Watson has tended to avoid overtly religious metaphors (Hubbard & Wald, 1993, p. 4) as he is a well-known atheist; so is Craig Venter of Celera, who has explicitly rejected the "language of God" metaphor (Nerlich & Hellsten, 2004). But many of the other prominent individuals involved with the Human Genome Project had no issues with casting genetics in theological terms.

[59] The relation between the material body in general and abstract cultural representations of personal identity can be traced as far back as Ancient Egypt, where specific human organs were considered sacred. (Eliade, 1957, p. 11). Descartes himself believed that the soul interacted with the body at the location of the pineal gland (Fox Keller, 1995, p 77)

In his speech on June 26, 2000 announcing the human genome assembly, Bill Clinton stated that "Today we are learning the language in which God created life" (Collins, 2010, p. 304; Ceccarelli, 2004). Francis Collins, a devout Christian who was often portrayed as a sort of missionary (Van Dijck, 1998, p. 132), stated that "We have caught our first glimpse of our own instruction book previously known only to God" (quoted in Nelkin, 2001). Following this announcement, and possibly partly as a result of it, the news media cast the achievement in metaphors which merged the familiar with the holy through deeply-ingrained theological imagery like "the handwriting of God" (Nerlich et al., 2002; Doring, 2005; Van Dijck, 1998, p. 129-130). According to Nelkin (2001), the four primary types of genetic metaphor in public communication in 2001, shortly after the completion of the Human Genome Project, were "the essence of personal identity", "sacred", "destiny" and "commodity".

The phrase "book of life", which was frequently articulated by scientists, universities, museums and NGOs in addition to the media, carries religious significance in the Judeo-Christian tradition as a variation on the theme of "natural, eternal and universal writing" (Doyle, 1994, p. 52; Kay, 2000, p. 31; Gogorosi, 2005; Doring, 2005; Hellsten, 2005). This particular metaphor helped to "focus and sell" the Human Genome Project (Avise 2001), imparting it with a kind of mystical significance or "quasi-religious sanction" (Nerlich et al., 2002; Gogorosi, 2005). The more explicit reference to the book of Genesis was also made (Van Dijck, 1998, p. 21). The Bible states "In the beginning was the Word", linking the notion of language to both genetic and cosmological origins (Doyle, 1997, p. 99), and Moss (2004) argues that the information metaphor resulted in a paradigm in which:

> "[DNA is] temporally, ontologically, and causally antecedent to organismic becoming . . . envisaged as context-independent information for how to make an organism [it] appears to have become the new heir to the mainstream of Western metaphysics." (p. 3)

Pollack (1995) – an endless source of this sort of rhetoric, who is well-known for his writing on science and religion – writes that "If a cell were as big as the Old City of Jerusalem, each chemical "letter" in the cell's DNA text . . . would be about as big as a letter in a word of any familiar book" (p. 18). Tomasula (2004) draws comparisons to the angels' eternal "language

without any syllables" described by St. Augustine and the language used by Adam to name the animals, both of which acted as "visible traces of God's mind" (p. 251).

Wilson (2001) argues that the idea of a "correct" genome is inherently teleological, implying a "final intention". Indeed, images of the genome have commonly been criticized for implying that it is the voice of God, mediated to the public by scientists as a "guide to moral order" (Weigmann, 2004; Nelkin, 1994; Nelkin & Lindee, 1995, p. 8; Van Dijck, 1998, p. 48-49) which threatens patient autonomy in genetic testing and decision-making (Nerlich et al., 2002).

Another metaphor which cast genetics as a sort of holy vocation, according to Van Dijck (1998, p. 21, 130) is the conception of the Human Genome Project as a quest for the fabled Holy Grail (see, for example, Walter Gilbert's 1992 book chapter "A Vision of the Grail"). Dar-Nimrod and Heine (2011) found that exposure to metaphors like "holy grail" tends to cause laypersons to see genetic influence in a more absolute fashion, potentially contributing to stigma against those with certain genotypes (Parrott & Smith, 2014).

Religious language is associated with "definitive power" rather than open to interpretation (Ceccarelli, 2004). As such, genetic determinism is reminiscent of Calvinistic predestination, in which the "plot" conveyed by this language determines one's fate (Churchill, 2002, p. 185). However, Tomasula (2004) also points to the evolving tradition of Biblical commentary and explication as the texts were proliferated (p. 252-253), while Pollack (1995) compares alleles to different versions of the New Testament (p. 37-38). Both of these metaphors suggest the room for latitude in the interpretation of religious texts.

The magicality of genetics implies the potential for supernaturally effective cures to be developed for diseases considered to be genetic (Roof, 2007, p. 181). Indeed, the notion that these types of condition could be controlled metaphysically through willpower and "positive thinking" were voiced by some of the patients interviewed by Klitzman (2010), as was the belief that they represented a form of punishment for "living an immoral life". Wexler (1979) also noted that some families viewed the illness as a "family curse" or as punishment for "the sins of the fathers". Sontag (1978) wrote that:

> "Ceasing to consider disease as a punishment which fits the objective moral character, making it an expression of the inner self, might seem less moralistic. But this view turns out to be just as, or even more, moralistic and punitive . . . the romantic idea that the

disease expresses the character is invariably extended to assert that the character causes the disease." (p. 46).

Indeed, some of the patients quoted by Wexler (1979) related Huntington's to anger or other unpleasant personality traits. However, I do not think this can be said to occur in the same way for genetic disorders which are inherited, as any disease that could be tested for via amniocentesis could be considered present before the fetus has a character. The defect must in these metaphors be imposed externally, and in the theological context that suggests the involvement of God.

Somewhat alarmingly, Doyle cites the ominous Biblical verse from Revelation, "And whosoever was not found in the book of life was cast into the lake of fire" (Doyle, 1994, p. 52; Doyle, 1997, p. 62). He argues that the existence of a "book of life" (which, as previously mentioned, was the single most popular specific metaphor of the genome in many countries) enables the revealing of a theological Truth as narrative closure (Doyle, 1997, p. 62). We would do well to question what this particular set of metaphors imply about genetic disorder and the soul. Does it represent a departure from the image or the plan of God, as the Revelation quote suggests, or a trait deliberately imposed by him? One survey of the general public in Georgia found that those who saw the "blueprint" metaphor in a theological manner as God's plan were more likely to accept deterministic view of the genome – one stated that "God by giving the genes that we have . . . has already decided for us what diseases we will be suffering from" (Condit et al., 2002). The question of how genetic disorder relates to religious sensibilities given the frequent spiritual interpretation of the genome is definitely a topic that deserves further investigation.

Another important consideration in the narrative significance of genetics is the particular religious meanings which metaphors of genetic disorder imply for members of under-represented cultural groups outside the Judeo-Christian tradition. Genetic testing is an ethically-sensitive issue for many indigenous peoples due to their involvement in genetic research projects whose results conflicted with their beliefs (see Reardon & TallBear, 2012); however, the adoption of the genetics-is-language metaphor has also allowed "crucial" tribal creation narratives to be aligned with modern scientific theories in an acceptable fashion (TallBear, 2007). She quotes a filmed

interview in which one member of the Diné (Navajo) tribe discussed his perspectives on the "code of life":

> "And we have the narrative about creation and in that creation there is a divvying up of information and knowledge . . . And when we're talking about those things we're actually remembering it because it's already imprinted within us in our DNA and our RNA. So we do chants, we do songs, we do prayers and all we're doing is reciting those reference points within our existence . . ." (TallBear, 2007).

Although New Zealand is among the least "geneticised" countries of the developed world, focus groups held with the Māori in New Zealand have indicated that popular metaphors such as "the book of life" or a "script" had contributed to views of genetic exceptionalism, in combination with local beliefs. Participants voiced beliefs that genome determines identity and can be "read" to predict the future. The genome was also seen as meaningful in relation to the past, carrying spiritual significance through comparison with the genealogical system of *whakapapa* as "a really, really sacred link with the ancestors". Some participants drew parallels between the genome and the practice of *mihi*, a formal verbal introduction and statement of identity including "their ancestry, their tribal and genealogical connections, and their connections to particular mountains, rivers and landscapes". Hence, the idea of performing genetic testing, even in order to terminate fetuses with significant genetic disorders, was described by participants as "like editing bits of ourselves" (Scott 2006).

Contrary to some of the aforementioned North American groups for whom genetic backgrounds contradict beliefs about creation, the significance of *whakapapa* coincides with a curiosity about the ability of genetic testing to provide information about one's more distant ancestors, as well as the use of genetic diagnostics offers the ability to benefit one's *whanau*, or extended family. This potential was considered especially important by the Māori because of the emphasis their culture places on continuing one's family line (Evans, 2012).

Several studies have also discussed Māori beliefs with respect to genetic disorders and risk predispositions. One Maori family carrying a mutation for otopalatodigital syndrome, which led to the early deaths of male babies, believed that they were subject to a curse, or *makutu* (Evans, 2012). In another example, the finding that an allele for low activity of monoamine oxidase

(MAO) which was associated with aggressiveness and addictive behaviours led to it being described as a "warrior gene", linking it in a positive fashion back to the Māori belief in their descent from the Polynesian trickster god Maui (Perbal, 2013).

As we have just discussed, there are a number of religious traditions from around the world which offer differing takes on the spiritual significance of genetics. However, another feature of this discourse is the implication that genetics unites all humans by revealing a shared vitality rather than a set of individual genetic souls. Despite findings such as humans' genetic similarity to chimpanzees, many writers have remarked on the characterization of the genome as the collective essence of the species (Chadwick, 2001, p. 347; Richards, 2001). James Watson has called the genome "what makes us human" (quoted in Jaroff, 1989), and Sharp (2000) wrote that "Current trends in genetics . . . locate the essence of our humanity within our DNA".

In a corresponding fashion, despite the fact that population biologists think of a species as encompassing variants (Lloyd, 1994, p. 107-8), in the words of Gilbert (1993) "Molecular biologists generally view the species as a single entity, sharply defined by a set of genes" (p.84). Pollack (1995) claimed that DNA had "sustained and linked all people since our beginnings as a single species" (p. 18), comparing the development of life across earth to that of a single embryo (p. 155). Indeed, DNA can be interpreted as a history text, answering the question of origin from immediate parentage back through the common beginning of life itself (Roof, 2007, p. 33, 92).

Most people assume that species are defined by a set of knowable characteristics which apply to all members except for a few deviant exceptions. This understanding is partly attributable to language like "the" human genome, as the definite article suggests specificity as an entity rather than as a descriptor (Hull, 1994, p. 209, 214-215).[60]

The idea of genome as species raises the uncomfortable suggestion that departures from the so-called norm, even if they do not represent a flawed soul, might be taken as a deviation from the standard of humanity. Juengst (2004) dismisses this possibility, stating that "species are not static collections of organisms" and that essentially no conceivable germ-line genetic modification could threaten the species of sacred human genome's species. Critics who assume

[60] Technically, the "type specimen" used as the formal biological example of *Homo sapiens* is Carl Linnaeus, the 18th-century taxonomer. Although this selection was made principally a posthumous honour for him, Hull notes that as a short-statured Caucasian Swede he would be a relatively poor representative of the vast majority of human beings (Hull, 1994, p. 215).

otherwise, he argues, seem to confuse the scientific and the more colloquial meanings of "human".

While theological images are typically framed positively as in the promotional material for the Human Genome Project, the metaphor of DNA as a source of religious meaning may have unintentionally resulted in increased public resistance to research involving "playing god" by "tampering" with a holistically designed genome (Nelkin & Lindee, 1995, p. 54; Nelkin, 2001; Richards, 2001; Nelkin, 2004; Weigmann, 2004; Lopez 2007). The idea of a sacred genome stands in opposition to the mechanistic view of genes as tiny objects that can be manipulated (Nelkin & Lindee, 1995, p. 56-57). In 1983, for example, twenty-one Catholic bishops wrote a statement asking for a ban on genetic engineering on that basis that humans should not "play God", a sentiment which was reflected in numerous articles from religious magazines (Nelkin & Lindee, 1995, p. 54-55). Many secular authors of both popular and scientific publications have also expressed similar views (Nelkin & Lindee, 1995, p.56). In a similar vein, the pro-life movement has taken the combination of genetic material into a new genome at conception to indicate the creation of a new human identity – and therefore soul – drawing on the metaphors which brought Christian imagery into genetics (Nelkin & Lindee, 1995, p. 42).This raises the interesting question of what can actually be considered the beginning of one's own life story (Chadwick, 2001).

As discussed in this chapter, the inherent abstractness of the genome has allowed numerous communities to apply their own interpretation to its role in identity. In the Judeo-Christian tradition and others, the metaphor of genes as self is fundamentally tied to that of genes as words in a language, meaning that some of the beliefs and experiences discussed earlier likely apply to this related metaphor as well. One potential difference might be in the idea of those words lacking a referent like a magic phrase, which might counteract the notion of an objectively correct version of a word – in its typical usage, "abracadabra" is already intentionally meaningless.

However, the personal control implied by the metaphor of a magic word is likely not carried over to the metaphor of text written by the hand of God, which implies conformity to a divine plan. It also makes the body extraneous to that plan; the metaphor refers to the information content of the gene rather than its countless tiny material bearers. It is the single intangible entity

of the individual soul or humanity as a whole, not a complex body or even a socioeconomic situation, to which theological metaphors and the like apply.

This conceptualization raises problems when that selfhood appears to be flawed; there are no subunits which can be externalized as in the "versus self" category discussed earlier. It would be difficult to conceive of somebody as part "mutant" or as having an alcoholic subsection. Discovering that one has a risk factor for disease might be more accurately thought of as switching one adjective with another. And as discussed in the following chapter, replacing terminology is exactly what many authors have recommended in response to these sorts of findings.

Chapter 5 – Action Plans

The study of how genetic disorders are understood through metaphor naturally raises the question of how this knowledge can be used to help meet the psychosocial needs of the people grappling with these conditions and their family members. In this chapter, I will discuss some of the methods that have been offered for leveraging metaphor in practice.

To begin with, numerous authors have proposed further interdisciplinary research on problems of self-concept. Klitzman (2009) recommends involvement by experts in the humanities including social scientists, philosophers, and linguists in addition to health care providers. There have, for instance, been calls for further empirical research on public understanding of genetics, the pragmatics of metaphor, rhetorical representations of specific genetic disorders and their effect on identity and behaviour (Condit, 1999b; Condit et al., 2002; Shiloh, 2006; Zeiler, 2009), including the implications of "perceived" and "actual" genetic identities, the understudied area of identity change over time (Klitzman 2009) and how social contexts, previous experiences, emotions and "cognitive-emotional interactions" influence behavior (Condit, 1999b; Condit et al., 2002; Shiloh, 2006). This sort of knowledge could help people to develop more useful coping mechanisms and improve the public's ability to provide feedback to the media, thus helping to balance out the influence of scientific experts and institutions (Condit, 1999b; McConkie-Rosell & DeVellis, 2000; Shiloh, 2006).

Indeed, a common suggestion is for laypersons to play a greater role in shaping the rhetoric used to present biomedical information. In order to counter genetic determinism, Condit (2007) has called for text intended for the public to be presented to them for specific feedback on whether the metaphors used are helpful or confusing with respect to the intended meaning. She wrote that public knowledge must be taken seriously and without "flippancy" while still being "challenged and refitted to new contexts and experiences" (Condit, 1995). As such, laypersons ought to be seen as information "processors" rather than mere "receivers" (Shiloh, 2006).

Learning more about how the public receives and responds to metaphor ought to help us more present genetic information in more effective ways (Shiloh, 2006). Based on his finding that laypersons with more science education had fewer misconceptions of genetics, Klitzman (2010) suggests implementing "targeted public education" in genetics. In addition to urging better genetics education in schools, Reydon et al. (2012) suggest that "as part of the scientific

professional ethos" scientists and science educators should help the public understand not just their misconceptions but also the philosophical and historical trends which initially led to the dissemination of misleading ideas about genetics. By pointing out research results that run counter to positivistic[61] and religious interpretations, Shiloh (2006) argues that we can produce a more multifaceted discourse that admits of the genre's eugenic and military-industrial past. Indeed, it has been argued that the recontextualization of medically significant research in a consistent and well-communicated manner is a key part of a democratic society, also making it a political responsibility (Pramling & Säljö, 2007; Vieth, 2010). Unfortunately, little of this work has been recognized in the overall discourse, perhaps because many scientists avoid interpretations contrary to popular perceptions of the gene-phenotype relationship underpinning the research of the biotechnology industry (Sidler, 2006).

As such, many specific recommendations have been made with respect to educating the public about the actual role of genes in illness. For instance, journalists could emphasize that genes in general are not more important than environment and that genetic diseases are not fundamentally different from other conditions (Condit, 1999b; Condit, 2007). Wilson (2001) advocates emphasizing that to the public that variation is "normal". Along those lines, media attention could be given to genetic advantages in addition to weaknesses so that genes are not seen as equivalent to disease (Gronnvoll & Landau, 2010). As Couser (2001) noted, "we don't hear much about the discovery of genes for friendliness, optimism, and using one's turn signal".

In parallel, health communicators should attempt to give the public a "sense of personal control" (Gronnvoll & Landau, 2010), striking a balance between avoiding genetic essentialism and the important goal of bolstering susceptible individuals' motivation to seek out protective care (Parrott & Smith, 2014). Many authors have suggested more substantial meetings with genetic counselors and other health care providers trained in lay beliefs and psychosocial issues, such as the representation of genetic disorders and the ways in which testing and treatment decisions are affected by fluid self-concept (Wexler, 1979; Chapple et al., 1995; Shiloh, 2006; Klitzman, 2009; Klitzman 2010; Vieth, 2010).

Juengst (2000) writes that symptoms expressed in patient accounts of illness could serve as more clinically-useful "organizational tools" for medicine than "mutation-based" definitions of

[61] According to the philosophical tradition of logical positivism, largely stemming from the Vienna Circle of the 1920s, truth can be judged only based on empirical evidence via the scientific method (Pickering, 1999; Vannatta & Vannatta, 2013).

disease, especially following the effects of genetic imperialism (p. 148-150). Narrative ethics is all the more important because issues of personal identity may not be amenable to objective analysis in the way that physicians are typically trained (Klitzman, 2009), making narrative ethics all the more important. Some authors have noted that even health care providers are insufficiently trained in the social and cultural significance of genetics. Indeed, language metaphors are used in academic biochemistry and genetics textbooks (Hedgecoe, 1999; Nordgren, 2003; Reydon et al., 2012). Furthermore, many physicians have limited ability to interpret and use genetic testing results. If professionals are insufficiently familiar with the scientific or the metaphorical sides of genetic testing and decision-making, it is difficult to expect them to articulate all the necessary information to their patients (Reydon et al., 2012).

Ought this to be the responsibility of therapists and psychiatrists instead? Perhaps not. Lippman (1992) and Armstrong et al. (1998) wrote that the process of geneticization moves identity issues from psychology into the field of genetics, while Toombs (1992) argues that people who consider themselves to be sick as a result of genetic disorders must be helped through the irreplaceable "healing relationship" expected from healthcare providers. However, an emphasis on curative treatment does not help the majority of patients who come in with genetic disorders. Instead, the goal must be to reduce fear and anxiety while restoring the patient's feelings of integrity as a human being (Toombs, 1992, p. 112-118).

Medical anthropologists have been active in trying to reconceptualize the body away from the Cartesian dualist paradigm in favour of embodiment (Strathern, 1996, p. 181; Everett, 2003), which is sensitive to contextual human variability in a way that the "authoritative, standardized text" of the genome cannot be. Hubbard and Wald (1993) wrote that "We urgently need to demedicalize our relationship to our bodies and our state of health" (p. 162). By implementing a narrative approach and using embodiment as a frame of reference for understanding the illness experience, health care providers can learn more about how patients with genetic disorders reconstruct their identities and improve their ability to provide information and support (Toombs, 1992, p.100-101; Petersen, 2006; Nowaczyk, 2012). Storytelling can be a way of regaining agency by constructing an empowering personal narrative, and might be particularly effective when individuals' autonomy or identity is threatened by a conflicting metaphor (Petersen, 2006).

Finally, in recognition of the "biosymbolic" paradigm in which metaphors are more representative of emotional effects than pure relational analogy, critical frameworks could be

expanded to better encompass and address emotional effects of clinical genetics (Condit 2009, Klitzman, 2010). This approach may be particularly important when attempting to actively introduce new metaphors, as is discussed in the following section.

New Metaphors

The identification of poorly-chosen and misleading metaphors could also result either in the rejection of metaphor in genetics communication, or – more likely – to new metaphors being suggested (Condit & Condit, 2001). After the results of the Human Genome Project dispelled much of the preceding paradigm, mainstream research was for the first time "poised to consider new metaphors" (Sidler, 2006).Even prior to that, many authors had recommended further research into the use and understanding of metaphor followed by the elimination of dysfunctional or inaccurate metaphors in favour of more accurate or effective terms (Ivie, 1987; Lippman, 1992; Rosner & Johnson, 1995; Condit & Condit, 2001; Richards, 2001; Lopez, 2007; Moore, 2008; Klitzman, 2009; Gronnvoll & Landau, 2010).[62] As Nordgren (2003) notes, the question of whether a metaphor is appropriate could refer to its effectiveness at communicating the author's thought, its rhetorical success or its scientific pragmatism.[63] However, in practice our goal must be to identify and prioritize metaphors which represent and structure patients' thinking about genetic disorders in a constructive manner. More qualitative studies could be used to determine which specific metaphorical frameworks are not progressive (Condit, 1999b).

New metaphors could then improve the understanding of genetics among bioethicists, scientists in other fields and laypersons alike (Copland, 2005). They might help improve coping mechanisms (Klitzman, 2009), convey the interdependence of nature and nurture (Moore, 2008), reduce fatalism, and encourage the public motivations to work towards more healthy environments (Gronnvoll & Landau, 2010). As genes are generally seen as increasing risk while behavior decreases risk (Ponder, Lee, Green & Richards, 1996), representations that play down

[62] However, Condit (1999a) argues that many of the earlier authors were not so much effecting change as criticizing metaphors that were already outdated and being replaced independent of their influence.
[63] Syed et al. (2008) write that the misuse of metaphor, and the t ethical issues that result, generally arise when writers inappropriately conflate metanarratives and subnarratives, which they describe in terms of "language games".

the genetic nature of certain illnesses may also help improve the sense of controllability or keep people from assigning blame to others (Shiloh, 2006).

Nordgren (2003) rejected the metaphors of program and code, writing that metaphors for genes that affect behavior must "imply a rejection of genetic determinism and emphasize the interaction between multiple genes and multiple environmental factors". One early example of such a metaphor is Fox Keller's recommendation (1988) of "glue", "nexus" or "linchpin" as a way of representing the genome's interactive relation to other factors.

Another example more populist flair is Condit's proposed metaphor (1999a) of genes as individual members of a football team whose effect on phenotype is likened to the outcome of a game, as the composition and efforts of a team influence the result without making it completely predictable. She argues that this metaphor would lessen the perception that genes "produce regular, linear life histories" (p. 233).

In order to achieve a similar effect, Mauricio (2005) and to a lesser extent Rolston (2006) have offered the interesting image of genome as an ecosystem complete with numerous networks of interacting factors. This metaphor has also been applied as well as the field of metagenomics – specifically, the genetic sequencing of the microbiota that inhabit the human body and influence our health (Juengst, 2009, p. 137-139).[64]

In a more concerted effort at metaphor generation, van der Weele (2005) asked a dozen biologists and philosophers of biology for images that could represent a reaction norm (the range of traits produced by a single genotype in different environments), receiving many "decentralized and potentially chaotic" metaphors including a village, a parliament and a jazz orchestra. Likewise, Porta (2003) recommends the metaphor of an expressive interpretation of a jazz score rather than the pure technical repetition of music to represent the relation between genotype and phenotype in complex chronic diseases. This was not the only musical metaphor offered.

Similarly, Gronnvoll and Landau (2010) suggest using metaphors like "dance" or "band" to represent gene-environment interaction, as these are both collaborative undertakings which bring to mind human agency over one's behavior and environment. Indeed, those authors showed

[64] Juengst (2009) also describes how the Human Microbiome Project challenged the "bodily boundaries" and the related ontological categories that shape our understanding of concepts like "purity" and "wholeness". Rather, it encourages more fluid and plural conceptualization in which predators are no longer malicious but play an important role in the system as a whole (p. 133-134, 139).

laypersons a video representing the genome as dance found that this did not increase expressions of genetic determinism. Others have also suggested the genetic metaphor of a musical score, mingling disparate elements to produce a complex and unique performance (Condit & Condit, 2001). Under this sort of metaphor, I imagine a genetic disorder might be seen as a note missed or played out of tune. While similar to the "typo" metaphor in many ways, this auditory metaphor strikes me as slightly more aesthetic and subjective. It would also imply that, while the written score may have been accurate, the fault was attributable to human error. Finally, almost everyone who undertakes genetic testing can read while not as many individuals can interpret musical notation.

In recognition of these aspects of the music metaphor one popular science book, which offered a less deterministic "musical instrument" metaphor. They note that instruments "don't determine exactly what music is played – or how well – but they do determine the range of what is possible", making it a less deterministic metaphor (Hamer & Copeland, 2000, p. 12). Interestingly, the image of the instrument suggests neither text nor agent (Nordgren, 2003), setting it apart from both of the major categories of metaphor discussed in this thesis and suggesting that this sort of alternative metaphor could conceivably be useful in combating their undesirable effects.

Another interesting metaphor that might be used to address these conceptualizations is Copland's (2005) interesting spin on the book metaphor. He suggested that the genome be seen as merely "the dictionary to the language of life", rather than the text itself. He writes that:

> "Like a book, life is a linear process that builds and develops as time passes. Indeed, if anything, it is the process of development that is the author of the book of life that is an individual organism. The book of life is effectively written by our lives" (Copland, 2005).

This would be "compatible with the practice of bioethics", highlighting the genome as necessary but insufficient for development of an organism and thereby enabling a narrative view of the process. However, it is difficult to say whether introducing a metaphor based on the same common domains of text and language would stand out sufficiently from the preceding discourse to be recognized as having different implications. As discussed in the section that follows, this has previously been a problem for critics of genomic metaphor.

Blueprints and Recipes: An Example of Metaphor Replacement

At this point, I will provide an overview of what is probably the most prominent and certainly the best-studied of these attempted metaphor replacements – that of the genome as blueprint or as recipe for the human being. Neither relates directly to genetic disorder, but they certainly carry implications for how we imagine genetic disorder, and I hope they illustrate the discourse sufficiently well to indicate how a proposed replacement for disorder metaphors might occur in practice.

Like the metaphor of language, blueprints serve as "concurrently material and representational inscriptions" of information (Van Dijck, 1998, p. 125). Both Lippman (1992) and Rothman (1998, p. 23) have identified "blueprint" as the most common metaphor for the genome, although it varies by time period and did not overtake the other images until the 1980s (Condit, 1999a, p. 270). During the Human Genome Project, about two hundred news articles a year used the blueprint metaphor (Gronnvoll & Landau, 2010).[65] Genes were often portrayed as the blueprint for the machine of the body, with gene therapy as the replacement of faulty parts (Conrad 1999). As we know, metaphors can affect research paradigms, and Avise (2001) writes that images of blueprints or machines "may have blinded many biologists to genomic imperfections attributable to phylogenetic constraints and evolutionary-genetic tradeoffs".

A few months after the announcement of the first assembly of the human genome, Craig Venter and Eric Lander (founder of the Broad Institute and first author of the draft genome which was published in *Nature*), stated the opinion that "We don't think blueprint is the right metaphor".[66] As mentioned in Chapter 2, many critics also disagreed with the blueprint metaphor, and indeed used it as a representative against which to direct attacks targeted at genetic metaphors in general (Lippman, 1992; Hubbard & Wald, 1993, p. 64; Nelkin & Lindee, 1995, p. 164-8; Condit & Condit, 2001). Starting around the mid 1990s, critical ideological

[65] Interestingly, in the early 1990s there was a call by social critics to replace the code metaphor with ones like the blueprint, contributing to a more "contextualized" image of the genome (Condit, 1999a, p. 160). However, Condit et al.'s focus group (2002) found that the term still occurred less frequently than those of code, plan, and map. Although earlier military-industrial metaphors like "command center" were considered even worse (Hubbard & Wald, 1993, p. 64; Condit & Williams, 1997; Condit & Condit, 2001), the code and blueprint metaphors are often critiqued similarly anyways (Gronnvoll & Landau, 2010).

[66] Lander preferred the metaphor of a parts list, which strips the image of "order, instruction, or meaning" (quoted in Roof, 2007, p. 110), although Venter rejected that metaphor as well (Nerlich & Hellsten, 2004).

research increasingly decried the "blueprint" metaphor as reductionistic, deterministic, and hierarchical (Lippman, 1992; Hubbard & Wald, 1993, p. 64; Condit, 1999a, p. 165; Condit et al., 2002; Everett, 2003; Condit, 2004). Condit & Condit (2001), for instance, called it "static, product oriented, uniform, deterministic, and excessively simple".

It was claimed that the blueprint metaphor presented the idea of a beanbag-like collection of single genes each responsible for a single trait regardless of context. The public may readily take up such images precisely because they cleave genetics from environment and coding sequences appear to be more important in medicalized paradigms (Fogle 1995; Spanier, 1995, p. 93). In a recent study of 324 American undergraduates, Parrott & Smith (2014) found that "blueprint" language tended to increase essentialist beliefs about the efficacy of genetic research in improving health. The metaphor did not improve perceptions of personal control as much as "instruction" metaphors, possibly because the latter involves more human agency (Parrott & Smith, 2014). Certainly, more people follow instructions in their daily lives than build things from blueprints. However, other audience studies (summarized well in Condit, 2004) have found that laypersons do not see blueprints as deterministic. Despite this lack of certainty, several replacements have been offered in place of the blueprint metaphor. One of these was of DNA as a language, with the development process occurring through a "reading of the text or performance of the script" (Ceccarelli, 2004). As discussed in the section on that topic, this metaphor comes with its own set of very influential associations.

Also as a result of these concerns, Rothman suggested replacing the term "blueprint" with "recipe" in order to draw more attention to process (Rothman, 1998, p. 23; Condit & Condit, 2001; Moore, 2008). The "recipe" metaphor was first offered as one of many images in the intensely productive era following the discovery of the double helix (Condit, 1999b). As Pramling & Säljö (2007) point out, recipes can be considered a variant of the text metaphor, whereas blueprints are more visual (Condit et al., 2002). It has been described as a better representative of external influence, human activity, growth over time, and a lack of "one-to-one" correspondence between input and results. It has also been described as conveying more creativity, flexibility, variability and complexity (Hubbard & Wald, 1993, p. 11-12; Rothman 1998, p. 23; Condit & Condit, 2001; Condit et al., 2002).

Ethologists Bateson and Martin (2000) also offer a cooking and, indeed, "cookbook", metaphor for genes and environment in development, stating that it places importance both on

the raw ingredients brought together inseparably as well as the method and timing needed to cook them (p. 16). Interestingly, this metaphor is similar to familiar, long-established idioms like "a bun in the oven" for pregnancy. Hubbard and Wald (1993) wrote that:

> "We need a cookbook if we want to make a complex dish, but it does not make the dish, nor can it determine which dish to make or whether the dish will come out right. The cook and the ingredients will determine whether and how a recipe is used, whether we end up eating soup or cake, and how the food tastes. Cookery is also an apt metaphor because it introduces an element of adaptability and flexibility. A good cook can deviate from the recipe and fudge the outcome is she or he lacks some seemingly essential ingredients or implements. Similarly, cells and organisms can compensate for 'genetic mistakes'. Moreover, if cells and organisms are the cooks in this metaphor, many ingredients, among them genes and environmental factors, combine to produce a 'dish' that could not have been predicted by looking at the ingredients separately." (p. 11-12).

The recipe and cooking metaphors were praised by Nordgren (2003), among others. In accord with these recommendations, the recipe metaphor was quickly taken up by the media and scientists conducting outreach – rising through the mid-nineties from twenty to sixty academic uses per year by 2000 (Condit & Condit, 2001; Condit et al., 2002). One survey taken by 122 undergraduates concluded that, on average:

> "the participants perceived *blueprint* as connoting less variability and being more fixed, as more controlling, more uniform, and more determined than they perceived *recipe* [while they] saw the recipe metaphor as more closely associated with smallness and with their own use, as well as substantially more personal, more simple, more friendly, more familiar and more female" (Condit et al., 2002).

However, Condit has criticized the recipe metaphor for three primary reasons. Firstly, the specific meanings read into "recipe" by academics did not equate to laypersons agreeing with them. Members of the public thought of recipes more as lists of multiple ingredients, implying a mix of many factors or of one's ancestors, than the series of instructions most academic critics

conceived. Some disagreed with it because it implied things coming together to form a homogeneous whole rather than an assembly of different systems (Condit et al., 2002). One focus group found that American undergraduates thought recipes suggested the alteration of humans, rankling pre-existing concerns about genetic manipulation (Condit & Condit, 2001). Many members of the public actually preferred the blueprint metaphor because they had already heard it so many times it sounded more scientific than the new metaphor (Condit et al., 2002).

Secondly, "recipe" appeared to be fairly meaningless in practice. The metaphor was mainly used in a mere "decorative" manner in texts addressed to the general public. Only 15 of the 58 articles sampled by Condit et al. (2002) used it in a heuristic manner, and none for longer than a single paragraph. In 41 of those articles, it stood without elaboration such that it could easily have been replaced by other metaphors (Condit et al., 2002). As such, the recipe metaphor did not appear to reduce determinism among laypersons as its advocates had predicted. Condit et al. (2002) conducted a Likert-scale study in which participants read identical passages differing only in the use of "blueprint" and "recipe" and both groups expressed equally reduced levels of determinism. This is likely because it was insufficiently different from a blueprint in the first place, as both are essentially "static" instruction manuals (Condit & Condit, 2001). Indeed, the two often appeared interchangeably in the same articles (Condit et al., 2002).

Thirdly, a survey of 95 members of the general public gave the impression that these metaphors were hugely polysemic, with so many potential meanings that, in accordance with Josef Stern's theory of metaphor, their impact would be determined by the clusters of meanings (like religious views) that people brought to them. They concluded that the term "recipe" had failed because the critics who had advocated it did not understand the polysemic, context-based nature of both metaphors (Condit et al., 2002).

Despite the fact that the participants in these studies expressed considerable variability and made references to ingredients, neither were reflected in the media's use of recipe metaphors. Rather, articles were "structured by other, more powerful discursive units" from their well-established context. Condit et al. (2002) suggest these patterns of discourse emerged because science writers formed a "small and close-knit community, writing within a well-defined genre", one which portrays genetics as "elite, masculine or at least androgynous" as opposed to the "socio-ethically fashionable" recipe metaphor which was "familiar, friendly and female".

Indeed, the only perceived difference between the blueprint and recipe metaphors which did not differ significantly between individuals was that of gender association (Condit et al., 2002). Recipes were more "personal", "nurturing", "homey", "creative" and "feminine" in flavor. Condit et al. (2002) also noted that most of the critics who preferred the recipe metaphor have been female. However, based on the authorship of books and papers read for this thesis, it seems that most of the academics who have made major contributions to the study of genetics in metaphor were female in the first place, so there is likely no more than a correlation.

There is, of course, a significant undercurrent of gender in the evolving discourse of genetics (see Spanier, 1995). Based on the fact that maternal and paternal chromosomes combine during fertilization, genes are often attributed "gendered behaviour" (Roof, 2007, p.120) as "heterosexual couples" (p.94), while the gene "becomes an attractive little personal representative on the level of the molecular" (p. 94). Roof (2007) calls this "the imaginary embodiment of a binary principle never detached from ideas of gender [or] the logic of heterosexual reproduction" (p. 49).

Prior to World War II, the cell nucleus and cytoplasm were respectively (and hierarchically) associated with male and female, and the eventual change away from this metaphor may have arisen from some combination of the women's movement and hiatus of bacterial genetics, (Fox Keller 1995, p. 38, 41; Spanier, 1995, p. 149-50) both of which resulted from the war's disruption to the status quo. However, Van Dijck suggests that the character category of "scientist" in the genetic narrative has remained gendered as male (1998, p. 24), as are the explorers conjured up by the rhetoric of treasure hunts and new frontiers (Rosner & Johnson 1995).

The metaphors of treasure map and the exploration of a new continent were used in promotional videos from the early stages of the Human Genome Project (Hubbard & Wald, 1993, p. 158). Ceccarelli has noted the frontier language expressed by prominent figures such as Bill Clinton, Tony Blair and Francis Collins in the 2000 announcement of the Human Genome Project completion, stating that this sort of mapping allows "its owners to conquer the land for use and for profit . . . with the express purpose of aiding in the alteration of that territory" by human agency (Ceccarelli, 2004). Rosner and Johnson (1995) concur that mapping is inextricable from the rhetorical assertion of ownership through representation. These metaphors have drawn comparison both to the triumphant, colonialist discovery of the New World and the

penetration of pioneers into the American West (Van Dijck, 1998, p. 126-127) as well as colonization by European empires (Jones, 1994, p. 56; Rosner & Johnson, 1995). In response, some authors have advanced "counter-metaphors" by drawing negative attention to the commercial activities of "bio-colonialism" and "gene prospecting" (Rosner & Johnson, 1995; Nelkin, 2001). Rather than attempting to replace outright the language that was seen as damaging, this strategy involved subverting it.

As Knudsen (2005) comments, "the scientist can either discard the metaphor because of its metaphoricity or because of it being an inaccurate one, or she can repair and re-interpret the metaphor, thus providing the concept with new meaning". Van Dijck (1998) concurs that images always avail of reinterpretation. She concluded not we should not try to eliminate pre-existing images so much as use our "imagenation" to "reconfigure" science and society (p. 197-198). Similarly, Copland (2005) argues that replacements "must to a certain extent incorporate the previous metaphor and explain how our understanding has progressed".

This may be more effective in practice. As noted above, many laypersons tend to prefer established metaphors because they sound more scientific. Furthermore, no single metaphor is capable of conveying all the relevant aspects of a genome (Avise, 2001; Porta, 2003), and most popular science authors thus tend to mix metaphors in their work (Cecccarelli, 2004; Roof, 2007, p. 85). While Ceccarelli (2001b) maintains that the use of multiple metaphors can help provide a more rounded view and avoid simplistic explanations, she subsequently revised her original claim by noting that mixed metaphors often reduce each other to their lowest common denominator, filtering out more desirable associations (Ceccarelli, 2004). Since newly-introduced metaphors tend to appear alongside others (Condit et al., 2002), they may not convey specific connotations so much as strengthen underlying relationships like those of genetic disorder as a separate entity or as an aspect of selfhood.

As this chapter has shown, there are numerous ways in which a metaphor could conceivably be replaced, from the substitution of a word similar enough that the two could be easily substituted in most contexts to the development of a pre-existing metaphor in a new direction. In addition to promoting embodiment and avoiding theological or ontological metaphors, these recommendations have typically involved de-emphasizing genes in favour of personal control over one's health. But this is not particularly useful in the case of many single-gene disorders,

which are not particularly tractable for the people who have them and continue to influence the thinking of people who do not.

It is not clear that simply using new words can prevent the old ones from continuing to be used or diminish their previous effects, and it appears to be very difficult to predict those effects and interpretations anyways. While I do not doubt that medical genetics could be very different today if DNA had never been seen as information, language, or code, it is doubtful whether the metaphors and interpretations described in this thesis are even vulnerable to intentional manipulation, even for the best of intentions.

Conclusion

When I first decided on the topic of genetic disorder as metaphor, I had no idea whether there were even enough academic publications on the subject to synthesize into a discussion of their common themes. Although it is certainly true that a limited number of authors have addressed this area, and much of the important material has had to be recontextualized from publications addressing the rhetoric of the genome as a whole, I believe the above discussions have shown that the topic is sufficiently important (and interesting) to be studied in its own right.

Although I initially had some concerns about the actual importance of metaphor in the bioethics of genetic disorder, these concerns were allayed by my research. Metaphors, far from being mere rhetorical devices, both generate new understandings and obscure other parts of the subject. According to conceptual metaphor theory, metaphors shape the way that people both think and act. This makes their study fairly important for biomedical ethics in general and narrative ethics in particular, which is likewise concerned with how people experience and describe their situations. Narrative sensibility allows health care providers to reach past the difficulty of self-concept that emerges along with genetic disorder and collaborate on a course of action that both interprets and addresses these experiences.

Furthermore, patients tend to have limited genetic literacy and use metaphors when trying to understand their genetic disorders and risk predispositions in a meaningful way, who tend to mix technical and non-technical sources of information and choose different sorts of metaphors on the basis of emotional valence rather than on the strength of the analogy. Narratives, and the rhetorical devices used within them, influence people's perceived susceptibility to diseases of genetic origin, their beliefs about the tractability of those conditions, and their eventual decisions with respect to testing and treatment, in addition to their coping methods.

More than other areas of medical practice, genetics is associated with the important narrative influences of origins, families, identities and – through numerous popular metaphors - the idea that it is a language or narrative itself. The notions of text and identity are entwined through metaphors of theology and the Word, which cast the genome both as the soul of the individual and as a defining property of humanity in general. Indeed, it is against a representation of the ideal or the normal human that genetic disorder must then be defined, even though we have no reasonable way of identifying what this should actually be.

Over the course of this thesis, I have advanced the theory that metaphors of genetic disorders typically present them either as independent agents in opposition to the self or (almost the opposite) as attributes of one's personal identity. Just as metaphors of genetics in general have shifted in tone and import, these two overarching conceptions have varied in popular discourse over the course of the 20th century based on the scientific and cultural context.

Accordingly, I traced the development of genetic metaphors from the initial formulation of the gene, through the influences of Erwin Schrödinger in the 1940s, military-industrial funding priorities and the advent of related disciplines in the 50s and 60s, voluntary hereditarianism and the "lottery" metaphor in the 70s, commercialization in the 80s, medical genetics and the Human Genome Project in the 90s, and the "postgenomic" 21st century.

The image of the gene has long remained concurrently material and abstract, resulting in a great deal of metaphorical flexibility that has allowed genetic disorders to be conceptualized as an aspect of identity imposed upon or revealed from within a patient. Under this conceptual framework, the names of diseases are applied – in both affirmative and denigrating ways – as descriptors to both individuals and their patients. The conception of the genome as representative and essence of the body as a whole is also associated with the informatization of the body, compressing the individual into data in a way that literalizes the metaphors of program and code.

Although they are rarely treated at the level of DNA or RNA, genetic disorders are widely defined with respect to their etiology independent of social or even biochemical context. More and more conditions have been included under the category of genetic disorder, increasing both the number of actual patients subject to these metaphors and the number of people forced to adjust their self-concept despite lacking symptoms (and potentially never developing any). This paradigm directs focus away from actual effects on patient experience and suffering, complicating these conditions' relationship with narrative ethics and making the application of those techniques all the more important.

The influence of previous, and frequently scientifically outdated, discourses remain influential in shaping the way that laypersons conceive of genetic disorder. The fact that they are so frequently viewed as the result of single genes, despite the fact that such disorders are comparatively quite rare, derives perhaps partly from the popular model of an unpreventable one-to-one correspondence between genes and traits, and partly from the fact that single-gene disorders are more easily categorized as abnormal than complex predispositions.

The so-called fragmentation of the body that emerges from reductionist portrayals and diagnoses of disease contributes to metaphorical representations of genes as independent objects like time bombs and viruses. In addition, they are often personified and granted agency separate from that of the actual patient. Under the conception of genetic disorder versus self, the desires, willpowers and personality characters of the affected individual are set against an enemy, one which is often vilified but less frequently triumphed over.

Another key role of metaphor in genetics is in the explanation of biomedical research, both as models within scientific discourse and as part of popularization efforts directed at laypersons for the purposes of education, self-promotion or advertising. An understanding of metaphor is thus necessary in order to communicate important information in the culturally-meaningful form desired by the public and to recognize when alternative rhetorical strategies would be more effective.

The metaphors which characterize both of the two principal conceptions of genes in relation self have been argued against by authors in this area. In replacement, it is frequently claimed that prioritizing embodiment, a paradigm which the body is identified as the self rather than as a separate metaphysical entity, can both help in this goal and return attention to the importance of actual patient experiences of chronic genetic illnesses. Another of the key strategies that has been suggested is to emphasize the passivity of the gene and the active agency of the patient in controlling behaviour and environment, a task which could potentially be aided by the adoption of appropriate metaphors for genetic disorder.

In conclusion, I think it is fair to say that there is plenty of opportunity for future audience studies, surveys, and other qualitative research into the metaphors of genetic disorder, both in terms of how they affect laypersons' behaviour with respect to these conditions and how they might feasibly be altered to improve patient experiences. The categories presented in this thesis would be interesting to test in further research, and given the amount of literature on this topic, it would not be that difficult to make a significant contribution to this field of study.

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