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The Visual Transcription of "Family Disease":

A Comparison of the Use of Medical Pedigrees in Genetic Counseling Practices in Canada and Japan.

by

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ABSTRACT

In recent years, with the development of DNA tests and genetic knowledge, there has been a growth of genetic counseling services and research in Canada and Japan. Although the uniqueness of genetic services in medicine lies in the preliminary assessment of the entire family rather than a single patient, few attempts have been made by social scientists to examine the technical and social construction of family trees and medical pedigrees. The purpose of this thesis is to analyze how the family data taken by genetic counselors are transcribed as medical pedigrees and used by associated health care workers in different cultural settings. The comparative analysis was based on an ethnographic approach that included participant-observation in genetic counseling sessions, interviews with clinical workers, and content-analysis of medical textbooks. The findings include three major points: 1) cultural views of the family are taken for granted by genetic counselors; 2) the process of documenting family data consists of four stages: primary transcription, secondary transcriptions, combination and publications; 3) the clinical workers' use of medical pedigrees results in the construction of family history as part of the present family illness.

Résumé

Au cours des dernières années, suite au développement des tests d'ADN et des connaissances génétiques, on a assisté au Canada et au Japon à une forte croissance des services de conseil génétique et de la recherche en génétique humaine. La génétique médicale se distingue des autres pratiques médicales dans la mesure où elle porte non pas sur un patient individuel mais sur la famille du patient dans son entier. En dépit de cette caractéristique, aux lourdes implications culturelles, de la génétique médicale, peu de chercheurs en sciences sociales ont examiné les pratiques de cette discipline, et, notamment, la construction technique et sociale des arbres généalogiques et des pedigrees médicaux. L'objet de ce mémoire est d'étudier la façon dont, dans des milieux culturels différents, les données familiales recueillies par les conseillers génétiques sont transformées en pedigrees médicaux et utilisées par les travailleurs de la santé. L'analyse comparative Canada-Japon est fondée sur une approche ethnographique incluant l'observation-participante de séances de conseil génétique, des entrevues avec des cliniciens, des infirmières et des conseillers génétiques, et l'analyse de contenu des publications médicales pertinentes. Les résultats de cette enquête comprennent trois éléments majeurs: 1) des conceptions culturelles différentes de la famille sous-tendent le travail des conseillers génétiques au Canada et au Japon; 2) le processus de documentation des données familiales comprend quatre étapes, à savoir une étape de transcription primaire, une étape de transcription secondaire, la combinaison de différents types de données et le remaniement de ces données en vue de la publication; 3) l'utilisation des pedigrees médicaux par les cliniciens et les conseillers génétiques a pour résultat la transformation de l'histoire de la famille en un aspect de la maladie qui affecte présentement certains de ses membres.

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CHAPTER1. INTRODUCTION

> I went to the woods because I wished to live deliberately, to front only the essential facts of life, and see if I could not learn what it had to teach, and not, when I came to die, discover that I had not lived.
>
> Walden, or Life in the Woods, Henry David Thoreau, 1854.

This Master's thesis deals with social and cultural issues about "medical pedigrees" that have been virtually ignored by social scientists and health care workers. Medical pedigrees are genetic counselors' visual tools which serve as the clinical record of patients and family in genetic counseling services and research. The focus of the thesis will be on a comparison of the role and value of medical pedigrees in genetic counseling practices in Canada and Japan. The genetic counselors' transcriptions and publication of medical pedigrees are not only a technically central part of genetic counseling, but they also constitute a practice involving key social and cultural issues such as the notion of the family, the ownership of family information, and the production of "family disease."

In recent years, with the development of genetic technology, there has been a growth of genetic counseling services and research in Canada and Japan. The development of amniocentesis, certain techniques of chromosome identification, and recombinant DNA technologies have led to the establishment of a growing number of genetic centers. This has, as a result, led to an increase in the importance of genetic counseling services and research. "Genetic counseling" has been defined as "a communication process which deals with the human problems associated with the occurrence, or the current risk of occurrence, of a genetic disorder in a family" (Fraser, 1974: 637). One of the most widely published research projects related to genetic counseling is "The Human Genome Project," described as "an attempt to map

the human genome, the distribution of genes on chromosomes, to find where each gene lies, and, ultimately, what each does" (Rothman, 1995: 1). The Human Genome Project, started in the 1980s, is an international collaborative enterprise dealing with genetic knowledge for biological research, which has medical and social applications. In 1986, as part of the human genome mapping, family pedigree analysis was started (Matsubara, 1992). The family pedigree analysis urged counselors and researchers to take more comprehensive medical pedigrees which would be used as a starting point for genetic tests.

A distinctive characteristic of genetic counseling services and research is to be found in the preliminary data-collection. Genetic counselors and researchers collect data concerning the entire family rather than data on an individual patient. Medical geneticists suggest that genetic counseling is different from traditional medical practices because genetic counselors assess not only a single patient but also the other family members (Fraser, 1963; Gelehrter, 1983; Gelehrter and Collins, 1990; Nora and Fraser, 1994; Ohkura and Kimura, 1989). Genetic counselors and researchers often request medical information on other family members, or ask the relatives to have a DNA test, if the tests are deemed to be relevant for the patient. Thus, it is very important for an understanding of the social and medical practices of genetic counseling to study how genetic counselors and researchers transcribe family data using medical pedigrees.

Although medical practitioners as well as social scientists may take "family pedigrees" for granted, pedigrees are not a self-evident, static tool. They can be made into empirical objects for sociological and anthropological inquiry. Pedigrees are the "visual tools" of clinical and laboratory workers. They make visible "the invisible knowledge of the family." In the social studies of genetic counseling, however, little attention has been given to this point. One reason for this is that social scientists tend to study genetic counseling in relation to early eugenics, which is the use of genetic measures to attempt to improve the genetic nature of a whole population and/or a society, usually of a certain *ethnic group*. Therefore, they also tend to argue about the social issues of genetic counseling in terms of ethnic groups. My present purpose is not to explore issues pertaining to ethnic groups, but, rather, to examine issues related to a smaller social unit, the family, on the basis of a sociological analysis of medical pedigrees in genetic counseling.

Numerous attempts have been made by both social scientists and medical practitioners to investigate the social issues involved in genetic counseling practices, mainly because of the possible wide-ranging social consequences of genetic counseling. In *Prescribing Our Future* (Bartels et al., 1993), biomedical ethicists, genetic counselors, and a clinical ethnographer discuss the issues raised by genetic counseling and research in the Human Genome Project. The authors contrast the possible "dramatic" impact of the Human Genome Project on society, and the "prosaic" features of genetic counseling services. Orr (1993), a genetic counselor, suggests that the Human Genome Project is likely to be the most influential factors affecting the demand that society as a whole will place on genetic counseling services. Bosk (1992), a clinical ethnographer, describes genetic counseling as a routine, prosaic process, during which genetic counselors provide only general genetic information. Genetic counselors claim to offer "non-directive" counseling, and genetic counselors are, therefore, described as "information-givers" (Hisa, 1979; Bosk, 1992), or "decision-facilitators" (Antley, 1979). Although social scientists are concerned with the new social power of genetic knowledge, many seem to fail to provide a detailed description of the visual tools used in genetic counseling practice.

In particular, research on genetic counseling often fails to grasp the importance of pedigrees in the counselors' practices. Thompson et al. (1991), in their book *Genetics in Medicine*, one of the most popular textbooks among genetic counselors, identify five stages of clinical genetic services: preassessment, clinical diagnosis and management, recurrence risk estimation, genetic counseling, and follow-up care.[1] The *preassessment stage* includes taking a "family history," collecting "family history information," practicing "clinical examination and laboratory tests of relatives if indicated," as well as asking the "reason for referral." The *genetic counseling stage* deals with the "nature and consequence of the disorder, recurrence risk, means of modification of consequences, means of prevention of recurrence (prenatal diagnosis and counseling)" (Thompson et al., 1991). In other words, prior to

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[1] According to the genetic counselors' terminology, the concept of genetic counseling can be used in two ways; clinical genetic service in a general sense and a communication stage (Thompson and Thompson, 1986) in a specific sense. In this thesis, the former is called "genetic counseling" or "genetic counseling services," the latter is called "genetic counseling stage" or "genetic counseling as communication."

"genetic counseling," the family assessment is carried out as a routine clinical practice by the counselors. Social and medical scientists have paid far less attention to the stage of pre-counseling assessment. Without either questioning or investigating the "preliminary" and "taken-for-granted" aspects of genetic counseling services, they jump to an overall assessment of genetic counseling (Bosk, 1992; Caplan, 1993; Holtzman, 1989; Kessler, 1980; Kessler and Levine, 1987; Lippman-Hand and Fraser, 1979; Rothman, 1986; Wertz and Fletcher, 1989; Yoxen, 1982).

This neglect is unfortunate, since the process of taking a pedigree is not only a technically central part of genetic counseling, but also a socially vital part of clinical practice and laboratory work. For example, the development of large pedigrees as tools in genetic research raises the question of who owns the family information (Frankel and Teich, 1993) assembled by various associated clinical workers: This inquiry into the construction of medical pedigrees will go to the core of this and related problems.

My thesis will address the following questions: How do genetic counselors know what they know? How do genetic counselors produce data out of the narratives of their clients and family members? How do they construct medical pedigrees as visual documents relevant to a diagnosis of genetic disease and genetic tests? How do the other clinical workers such as molecular laboratory researchers reconstruct medical pedigrees? How do medical pedigrees find their way in clinical publications? These questions should allow us to reconsider a phenomenological theme: how and why genetic knowledge has gained social power in everyday life.

My analysis is grounded on two approaches: (i) the theoretical analysis of visual representations and (ii) the use of comparative research methods. As explained in chapter 2, recent contributions in philosophy, anthropology, history, and sociology have discussed the issue of visual representations in scientific practice (Lynch and Woolgar, 1990). This analytical focus on visual representations is related to the fact that the latter embody tacit knowledge (Fleck, 1986 [1947]; Polanyi, 1966); visuals also entail practical knowledge and persuasive power (Gordon, 1988; Latour and Woolgar, 1979). I will apply to genetic counseling Amann and Knorr-Cetina's (1990) model of how "proto-

data"[2] become fixed as visual evidence for scientific belief. From this perspective, I will carefully examine how geneticists make "visible" the invisible knowledge of the family, the body, and even the nature of disease.

In order to explore "tacit" knowledge,[3] my research will rely on a comparative, ethnographic approach. Comparative methods make tacit knowledge explicit by articulating "taken-for-granted" elements from different social settings (Fleck, 1935). In the present case, an ethnographic approach (interviews and participant observation) is used in order to research "pedigrees in action." Medical pedigrees are not static objects but, rather, part of genetic counseling practices. The first step of my project was to carry out a small number of interviews with genetic counselors as well as participant observation in a human genetics course at McGill University in Montreal. Subsequently, ethnographic research was performed in the Tokyo metropolitan area of Japan. After comparing the data, I started a new cycle of participant observation and interviews in Canada. Finally, I carried out follow-up research in Japan. My comparative approach is thus characterized by an ongoing comparison of data produced through recurrent "theoretical sampling" procedures (Strauss, 1987), processes aimed at collecting data on the basis of a particular theoretical focus.

Why do Canada and Japan provide an interesting contrast? Although I am going to discuss this question in chapter 3, here I would like to mention that the secondary purpose of this thesis is to analyze the "internalizing-and-externalizing" aspects of biomedical discourses in different cultural settings. According to anthropological work on medicine, Western medicine is characterized by an internalizing discourse (Young, 1978), while medical discourse in Japan shows, by comparison, externalizing characteristics (Lock, 1986). If this is so, how then are genetic knowledge and family pedigrees constructed and used in both Western and non-Western contexts? Since my comparative research remains exploratory, this thesis will provide a limited analysis of the cultural aspects of genetic services and research. This research question, however, will affect my interpretation of the collected data.

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[2] Proto data are sense data activated by some "perceptible manifold-out-there." Amann and Knorr (1990) distinguish "proto-data" from "evidence": proto-data are produced in laboratory work, while evidence is published in scientific papers.

[3] See Cambrosio and Keating (1988) for a discussion of the ambiguities surrounding the use of the term of "tacit" in sociological research.

In Chapter 2, I will review the relevant literature concerning genetic counseling services and research, raising two questions: how is genetic counseling performed and why are family trees or pedigrees important for social and cultural inquiries. Beginning briefly with the historical background of genetic counseling, I will present an overview of the debates concerning genetic counseling in North America. These debates can be reduced to a confrontation between two models of genetic counseling: the medico-scientific perspective and the psycho-social perspective. Social scientists have tended to stress the pscyho-social perspective, portraying genetic counseling as a communication process. As a consequence, ethnographers have not analyzed the precounseling stage of genetic counseling. Secondly, I will discuss why the analysis of pedigrees is important for sociological and anthropological inquiries. Referring to the sociology of scientific knowledge, I will suggest that visual tools, especially medical pedigrees, play an important role in the technical and social construction of family diseases. In order to examine the relationship between visual tools and human actors, I will focus on the notion of *inscription devices* (Latour and Woolgar, 1979) and on a theoretical model of visual documentation in genetic counseling practices. The model consists of three levels of visual documentation; at the level of proto-data, family narratives are transcribed as *family trees*; at the level of evidence, *family trees* are retranscribed as *medical pedigrees*; and at the level of genetic theory, *medical pedigrees* are published as *family history*.

Furthermore, using medical texts and literature, I will show how medical pedigrees are important in genetic theory and practices. I will highlight four aspects of pedigrees. First, pedigrees are visual tools resulting in the production of phenotypic knowledge; they make visible the invisible genes described by genetic theory. Second, pedigrees are legitimated as clinical records, that is, as evidence in a clinical context. Third, pedigrees function as "boundary objects," that is, they mediate divisions between medical professionals. Finally, pedigrees express the power of genetic counselors to manipulate the body of the patients and the family.

Chapter 3 discusses methodological issues. I will describe my ethnographic approach that involves interviews with clinical workers, participant-observation, and content-analysis of medical textbooks.

Chapter 4 is centered on comparative analysis. It will focus on the cultural contexts of the family, and on their representations in the production of visual documentation. First, I will analyze the genetic counselors' encounters with the family, looking at who records the family data. I will also examine various notions of the family. I will describe some cultural differences in the notions of the family in Canada and Japan. In Canada, the prototype of the family is mainly the nuclear family consisting of a couple and child(ren), while in Japan it is the extended family that includes not only the couple and child(ren) but also the grandparents and even ancestors. In addition, in Canada, the concept of ethnic background is stressed by the counselors in recording procedures, while in Japan, the issue of marriage is one of the main topics emphasized in genetic counseling. These different cultural views surrounding issues of the family are often taken for granted by genetic counselors. How these ideas and values are reflected in the medical pedigrees is a question that will be discussed in an exploratory way at the end of the chapter.

Subsequently, I will analyze the four stages of the process of visual documentation of family data: primary transcription, secondary transcriptions, combination and publication. In *primary transcription*, family data are *translated* by genetic counselors from oral narratives into a handwritten family tree. Through this process, the genetic counselors contrast normal and abnormal icons, ignoring the family's stories of the illness. In the primary transcription, counselors tend to focus on "*the family as an expression of illness.*" In *secondary transcriptions* the primary trees undergo various transformations in the hands of clinical workers within local medical communities, and develop into various edited versions: an updated version, a personal version, a team version, and a version for requisition sheets. Through secondary transcriptions, pedigree information is stabilized and maintained as a clinical case file, i.e., as medical evidence. This explains why some counselors call for the establishment of standardized, universal pedigree symbols. In the secondary transcriptions genetic counselors are likely to perceive the "*family pedigree as an expression of the illness.*"

Combination of various types of medical evidence is a powerful technique leading to "fact mobilization," in which the clinical workers collectively reconstruct stabilized data. I found two types of combination: large pedigrees and laboratory pedigrees. *A large pedigree* consists of individual

family pedigrees. The construction of a large pedigree can be equated to the birth of the research pedigree. Because the patient and the other family members are not aware of the content of the large family pedigree, only researchers can obtain that information for research purposes. *A laboratory pedigree* is generally stored as a master file in a molecular biological laboratory. The laboratory pedigree serves as the coding of the phenotype-genotype relationship. The combination of medical evidence helps genetic counselors to see "*the family history as part of the present expression of illness.*" One of the reasons for combinations presumably lies in the uncertainty surrounding genetic evidence. Genetic counselors prefer to combine different kinds of evidence rather than to attempt to certify isolated family data.

Publications are also the result of collective work, leading to the establishment of coherent categorical knowledge of genetic diseases, on the basis of an accepted theory. The role of visual documentation in publications is to produce an integrated image of genetic theory through the combination of relational, external, and internal images of the patients' bodies. The family history used in medical textbooks plays an important role in introducing biology students to the perception of the family history as parts of the present expression of illness. Moreover, publications raise the issue of ownership of the research pedigrees. Issues of ownership are important, because the ownership includes a new social power to affect the personal life of the family. Although such pedigrees enable clinical workers to access information on the entire family, there is no clear answer as to who owns the family history.

In chapter 5 I will briefly discuss the cultural aspects of family data, the social construction of pedigrees, and the body politics of medical pedigrees. I will focus on the relationship between genetic counseling and ancestor worship in Japan, arguing that therein lies the main reason why Japanese have reservations about pedigrees and genetic counseling. Second, I will argue that visual representations of family data are not only the result of a process of simplification of culturally complex family data, but that they also mobilize the construction of new evidence. Third, I will claim that the construction and use of medical pedigrees involves the manipulation of family diseases. The construction of medical pedigrees may lead to the overproduction of "family diseases." As part of this process, genetic counselors tend to

perceive family history as part of the present expression of family diseases rather than as the product of interaction between genetic counselors and family.

In the conclusion I will return to three main points: 1) the role of cultural notions of the family as taken-for-granted proto-data, 2) the stabilization and mobilization process of medical pedigrees, 3) the manipulation of family diseases. The power of genetic knowledge resides not only in technological developments in laboratory work, but also in clinical work where the body politics of the family is practiced in the taking of medical pedigrees. These constructed family pedigrees can easily be combined with laboratory work, detached from the family's emotions and narratives.

CHAPTER2. LITERATURE REVIEW

In this chapter I will show that medical practitioners have historically focused on the genetic counseling stage rather than on the preliminary clinical services. Furthermore, I will discuss the controversy between the two models of genetic counseling: the medico-scientific model and the psycho-social model. Social scientists have also generally imitated medical practitioners, by focusing solely on the genetic counseling stage. The chapter will also review the general background of genetic counseling in North America, and discuss the importance of pedigrees for sociological and anthropological analysis.

A. What is Genetic Counseling?

Several key advances in genetic technology have been instrumental in leading to the widespread practice of medical genetics[4] and genetic counseling in the 1950s in North America. During that era, many developments were seen, such as the identification of the human diploid chromosome number (Kevles, 1985: 238), the identification of chromosomal disorders (such as Down's syndrome) by Lejuene and his colleagues in 1959 (Kevles, 1985: 247), the development of techniques for chromosomal analysis and of methods for estimating the recurrence risk for several Mendelian disorders (Fraser, 1979).

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[4] The concepts of Genetics, Human Genetics, Medical Genetics and Clinical Genetics in this thesis follow the terminology defined by McKusick, the President of the American Society of Human Genetics (1992: 668):

Genetics: the science of biologic variation.
Human Genetics: the science of biologic variation in the human.
Medical Genetics: the science of human biologic variation as related to health and disease
Clinical Genetics: that part of medical genetics concerned with disorders in individual patients and their families. Or the science and practice ("art") of diagnosis, prevention, and management of genetic disorders.

The development of a means to treat some inherited disorders and to identify affected individuals and carriers allowed medical practitioners to apply genetics to clinical practice.

In this period, genetic counselors were almost all academically-based Ph.D. geneticists, often in biology and zoology departments. Sorenson (1993) notes that this generation of genetic counselors practiced genetic counseling "almost by accident." These scholarly-oriented professionals tended to view their obligations toward the use of their expertise largely as one of presenting the facts and information. One of the factors contributing to the origins of a "non-directive" approach in genetic counseling may be found in the scholarly orientation and role definition of the first set of genetic counselors.

The next period extends from the late 1960s to today. This era witnessed the first institutionalized attempts to define genetic counseling. The growth of the discipline brought about a rapid increase in the number of counseling centers in the USA: from ten counseling centers in 1951 (Reed, 1974: 335), to over 150 centers by 1968 (Lynch et al., 1983: iii). Although these numbers are of varying reliability, all researchers agree that there has been a tremendous growth of genetic services since the late 1960's (Mitchell, 1987).

The definition of genetic counseling, which was devised in the 1970's by a committee of the American Society of Human Genetics, is as follows:

> Genetic counseling is *a communication process* which deals with the human problems associated with the occurrence, or the current risk of occurrence, of a genetic disorder in a family. (Fraser, 1974: 637, emphasis added).

This definition has been widely cited by genetic counselors (Mitchell, 1987). It implies that genetic counseling starts after the identification of the genetic problems of the client and of the family. The definition shows that medical practitioners and social scientists see genetic counseling as a communication process.

Since the mid-1970's, during a period of rapid change in the character of genetic counseling, many articles have discussed the medico-scientific orientation of genetic counseling and have criticized the lack of a psycho-social perspective. Socio-psychological issues such as religious beliefs, financial constraints, the emotional perception of genetic disorders, and the

reproductive goals of the clients had been neglected (Sorenson and Culbert, 1979). One of the important articles advocating a psycho-social perspective was Kessler's "The Psychological Paradigm Shift in Genetic Counseling" (1980). In this article the author criticized the paradigm of traditional genetic counseling explaining that little attention had been given to the process of communication and to the way information was acquired, processed and translated into a usable form. According to Kessler, "the psychologically-oriented paradigm moves genetic counseling closer to the world-view of the psychotherapists or mental health workers rather than that of the biologist" (1980: 182) and the paradigm "emphasizes the fact that communication is a two-way street" (Ibid.).

In addition to medical practitioners, social scientists have also investigated the psycho-social aspects of medical discourse. For example, Yoxen (1982), reviewing clinical genetics in the twentieth century, pointed to the existence of a division of labor in genetic counseling: "The legitimacy of this role as counselor and diagnostician is continually maintained by research production in which technical knowledge is seen as primary, and other skills, such as psychological insight, or an understanding of welfare entitlement, are seen as secondary and able to be entrusted to lower status members of the clinical group" (Ibid.: 152). He emphasized the dualistic perspective characterizing genetic counseling; technological knowledge played a primary role, while the psychological perspective played a secondary one.

Ethnographers have also drawn our attention to the psycho-social issues of genetic counseling through their analysis of the communication process. Bosk (1992, 1993), as a participant-observer, investigated M.D. genetic counselors in a pediatric hospital in the USA. His rich ethnography describes the psychological dilemmas faced by genetic counselors in their routine practice.

It is noteworthy that although ethnographers described the communication stage, they tended to skip the pre-counseling stage. Bosk took for granted the existence of everyday problems, such as client complaints. Consequently, his analytical focus was "on the solutions that the workgroup evolves to these everyday problems" (Bosk, 1992: 21). In other words, he did not pay attention to the process by which the problems were defined as genetic disease and assessed by genetic counselors in the pre-counseling

stage. Rather, he tended to describe genetic counseling as "routine practice," "dirty work," or "a form of abortion counseling."

Social scientists have not yet focused on how genetic counselors construct and use medical pedigrees. For instance, although visual representations in genetic counseling were described to some degree, ethnographers have not provided a detailed analysis of visualization practices. The visual representations described were limited to the visual aids used by genetic counselors in the communication stage. For example, Bosk mentioned the counselors' resort to visual displays as a form of scientific dramatization:

> This routine display of photographs and the expert decoding of them is a regular feature of counseling for couples who come in for advanced maternal age (woman over 35 years old are by far the largest single segment of clients who used counseling services). Ultrasound photographs and karyotypes are shown to parents to help aid their understanding of how prenatal diagnosis work. They are evidence of the real problem. Of course, such photograph display can only be a feature of chromosome disorders. Other types of disorders call for different dramatizations of scientific expertise (Bosk, 1993: 31).

According to Bosk's account (1992), visual displays are used as part of a dramatization of scientific practices that avoids the emotional issues raised by genetic counseling. His analysis does not elaborate on how and why scientific representations are used and dramatized in practice. Why do genetic counselors need to resort to these visual representations, or, if Bosk's analysis is right, to resort to scientific dramatization? How and why do genetic counselors avoid the emotional issues of clients through visual representations? It is necessary to have a further look at this process by focusing on visual representations as a starting point.

Rapp's (1988) article "Chromosomes and Communication" is the only exception to this trend. Although her interest is in the communication process, she describes the use of visual displays and mentions the importance of the family tree in the pre-counseling period:

> Genetic counselors generally begin communication with pregnant patients with three goals in mind; to convey significant information about the risks of birth defects and the availability and nature of amniocentesis; to take *a health and family history* and to communicate with the patient well enough so that her questions and concerns can be addressed(Ibid.: 146, emphasis added).

Rapp (1995: 180) also suggests that "[i]n a forty-five-minute genetic intake interview, the counselor must: take an individual and family reproductive health history; ascertain 'background factors' (e.g., age, parity, ethnicity, familial causes of morbidity and mortality) which might affect appropriate screening and diagnostic tests to be offered." It goes without saying that her ethnographic descriptions have contributed greatly to the discussion of the ethical and psycho-social problems of genetic counseling, genetic testing, and eugenics. However, since their emphasis was not on the pre-counseling process, they have neglected to analyze how "genetic counseling as a communication process" is grounded in and proceeds from a preliminary clinical diagnosis based on visual documents.

This literature review shows that social and medical scientists have not dealt with the primary stage of genetic counseling services and the use of medical pedigrees. Some readers may argue that this is hardly surprising, since there is no need for sociological and anthropological inquiry on the relationship between a visual tool (the pedigree) and clinical actors. In the next section, I will address this question, by discussing some recent contributions in the sociology of scientific knowledge. Then I will show how genetic theory and practices stem from visual representations.

B. Why is the Analysis of Pedigrees Important?

The analysis of pedigrees is important for sociological and clinical reasons. From a sociological and anthropological point of view, the pedigree is

one of the fundamental tools through which genetic counselors and researchers can make visible the invisible proto-data of the family. Through visual representations, family data are transformed into medical evidence and medical theory. In addition, in terms of genetic counseling, a pedigree is a useful tool because it enables clinical workers to visualize phenotypic aspects of genetic diseases. A pedigree also allows genetic counselors to legitimate their practices by acting as a form of clinical record. Furthermore, a pedigree can be conceived of as a "boundary object" that connects different professional disciplines.

B-1. Social Science and Pedigrees

1) The Sociology of Visual Representations

Since the 1970s, philosophers, anthropologists, historians, and sociologists of scientific knowledge have actively investigated the previously neglected domain of scientific constructions and beliefs. Recent work in this area has increasingly focused on the issue of visual representations in scientific practice, adopting various perspectives and investigating various fields and practices: the development of a visual language in geology (Rudwick, 1976), the socio-historical development of images as icons of objectivity (Daston and Galison, 1992), the role of inscription devices (Latour and Woolgar, 1979), the actor-network theory of inscriptions (Latour, 1990), the ethnomethodological inquiry into representations (Lynch, 1990), and the studies of the visual fixation of proto-data (Amann and Knorr-Cetina, 1990). Following a short summary of Latour's notion of inscription device, I will focus on Amann and Knorr-Cetina's model of visual fixation.

Latour and Woolgar (1979) introduced the notion of *inscription device* as a new tool for exploring laboratory life. *Inscription* is "an operation more basic than writing," used to "summarize all traces, spots, points, histograms, recorded number, spectra, peaks, and so on" (Latour and Woolgar, 1979: 88). Latour and Woolgar's notion calls for social scientists to observe the scientists' use of visual tools in daily laboratory practices rather than simply to examine published versions of their scientific results and theories. The notion of the inscription device can be applied to clinical practices and used to account for

the production and mobilization of visual documents, such as the pedigrees produced by genetic counselors. The notion will be of particular value in examining the process by which associated health care workers produce and stabilize the preliminary data of the family.

Berg (forthcoming) is one of the medical sociologists who applied the notion of an inscription device to the medical field. It is true that medical sociologists and historians had already discussed the construction of clinical records. However, as Berg (1992) points out, medical sociologists until recently generally separated the '"content" of medical activities from their "social" aspects, the former constituting a domain inaccessible to sociological investigation. Traditional medical sociology assumed that clinical data could be regarded as "facts" that physicians only needed to "reveal," and regarded medical criteria and disposal options as scientific, fixed "givens" (Wright and Treacher, 1982; Armstrong, 1983; Berg, 1992). Sociologists inspired by ethnomethodology, however, have shown that clinical records are constructed in the doctor-patient interaction. This has led to the analysis of the production of medical discourses (e.g., Raffel, 1979; Silverman, 1987). In particular, Silverman (1987) carried out a discourse analysis of clinical dealings, comparing parents with a Down's syndrome child with those with a non-Down's syndrome child. On the basis of that analysis, Silverman suggested that the medical discourse fixes the proper forms for constituting Downs' children. Although this thesis will not resort to discourse analysis, it will reject the traditional assumptions of medical sociology. By studying the production of pedigrees, I will investigate the actual content of the recorded family data, which was taken for granted by traditional social scientists.

2) A Model of Visual Documentation

Amann and Knorr-Cetina (1990) have proposed an analytical model of visualization to explain how scientists manage to produce consensual evidence out of a set of perceived data. Going beyond the Duhem and Quine thesis of underdetermination, "according to which data can never conclusively prove or disprove a particular theory" (Ibid.: 86), Amann and Knorr-Cetina suggest that this thesis is not concerned with how sense data may be problematic.

Their strategy was to focus on the fixation of proto-data rather than on the later stages of conventional judgments of scientific beliefs and theories.

As part of a laboratory study of molecular genetics, Amann and Knorr-Cetina (1990) provide us with a description of the initial processes involved in the fixation of evidence of visually flexible documents. They suggest that there exists a preliminary process in which molecular geneticists tinker with visible proto-data, prior to reducing the modified evidence. The model of visual fixation can be applied to the process of visual documentation in the pre-counseling stage of genetic counseling.[5] For example, the process of "taking family history" can be reformulated as follows:

> **Proto-data:**
> A family tree is constructed through "family data."
> **Evidence:**
> The family history is represented as a "medical pedigree."
> **Accepted Theory:**
> The family history is a "tool" for determining the diagnosis of a genetic disease, particularly the phenotypic expression of a family disease.

Although many genetic counselors prefer to use the word "family history" when mentioning the collection of family data, we will refrain from such a terminology. One reason is that the word "family history" is too general a concept for an ethnographic description and analysis. More importantly, this terminology hides the very fact that it is impossible for genetic counselors to take all the stories of the family; they just take a part of the family narratives. It is, therefore, important to define the words *family history*, *medical pedigree*, and *family data*. Usually the *family history* consists of a medical pedigree and the list of family members by sex, age, and state of health, and it is often utilized by the clinical workers in publications. The *medical pedigree* is the clinical inscription that is recorded as medical evidence during and/or after a genetic counseling session. *Family Data* are all

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[5] There are some obvious differences between laboratory life and clinical life. In laboratory life, scientists play a decisive role in constructing their scientific data, while in clinical life, not only health care workers but also clients who are presumably laymen participate in the construction of genetic data. In this thesis, I will focus on the genetic counselors' construction of clinical pedigrees.

the sense-data taken during the encounter with the family. The relationship between a *medical pedigree* and *family data* is as follows: "(f)amily data can be summarized in a pedigree, which is merely a shorthand method of classifying the data for ready reference" (Thompson and Thompson, 1986: 45). I will clearly distinguish between the notion of medical *pedigree* and the notion of *family tree*. The *medical pedigree* is presented as medical evidence in medical records and/or journals, while *family trees* are proto-data that will subsequently be summarized in a medical pedigree. Family trees are for the counselor's personal use. Although genetic counselors might not be sensitive to the difference, it is a valuable one insofar as it allows one to clarify both how the biomedical map of the family is taken and how that map is subsequently transformed.

3) Visual Tools as Boundary Objects

The pedigree is not only a technically central tool in genetic counseling but also a socially fundamental tool which mediates between different health care workers and generates medical power. One of the central arguments in this thesis is related to Akrich's (1992) question: "how do technical objects take part in building a heterogeneous network that brings together actants of all types and sizes, whether human or non-human?" Sociological theories such as symbolic interactionism and constructivism are grounded in the concept of "negotiation" which stresses the role of human actors rather than the interactive relationship of human and non-human actors (e.g., Strauss, 1978). On the other hand, Latour and Woolgar (1979) pointed out that the construction of facts is not simply the result of a technological or social negotiation; according to these two authors, "*[o]ur arguments are not just that facts are socially constructed, [....]; [w]e also wish to show that the process of construction involves the use of certain devices whereby all traces of production are made extremely difficult to detect*"(Ibid.: 176, emphasis in original). Following their "symmetrical" model, Akrich (1992) states that:

> [b]ecause the answer [to the previously mentioned question] has to do with the way in which they (technical objects) build, maintain, stabilize a structure of links between diverse actants, we can adopt

> neither simple technological determinism nor social constructionism (Ibid.: 206).

From this perspective, "[j]ust what the disciplining of a practice, or the localization of a tool, exactly means can never be understood from standpoints which center merely on e.g. the nature of a medical problem or, conversely, the actions of medical personnel" (Berg, forthcoming). Akrich and other sociologists of science and technology are interested with how facts are stabilized in a local context and how facts are related to scientific networks. My analytical focus is not only on the "fact stabilization" process in a local setting, but also on the collective work of network formation in relation to pedigrees.

In order to analyze this collective work, I would like to introduce Star and Griesemer's(1989) notion of *boundary objects*. These authors want to explain how scientists manage both diversity and cooperation across scientific worlds. Boundary objects are "scientific objects which both inhabit several intersecting social worlds and satisfy the informational requirements of each of them" (Ibid.: 393). Star and Griesemer (1989) were concerned with the problem of how various professionals managed to construct scientific representations despite different viewpoints and agendas: "The creation of and management of boundary objects is a key process in developing and maintaining coherence across intersecting social worlds" (Ibid.: 393). Pedigrees can be defined as boundary objects allowing several health care workers to cooperate.

To sum up, I have argued that visual tools such as pedigrees are legitimate empirical objects for sociological and anthropological study. However, so far I have not discussed how and why medical pedigrees are central for clinical and laboratory work. In the next section, I will look at the relationship between genetic theory and medical pedigrees.

B-2. Genetic Counseling and Pedigrees

1) Genetics and Medical Pedigrees

> Nature is nowhere accustomed more openly to display her secret mysteries than in cases where she shows traces of her working apart from the beaten path; nor is there any better way to advance the proper practice of medicine than to give our minds to the discovery of the usual law of Nature by careful investigation of cases of rare diseases. For it has been found in almost all things, that what they constrain of useful or applicable nature is hardly perceived unless we are deprived of them, or they become deranged in some way. --William Harvey, 1657.

The remark by Harvey, a famous anatomist of the 17th century, was cited in the introduction to the textbook *Principles of Medical Genetics* by Gelehrter and Collins (1990). It is interesting to see how genetics may be related to an anatomical perspective. The perception of the "law of *Nature* " through the study of genetic disease may be related to the visual representations of the heredity traits of the family. The natural history of genetic disease ultimately depends on the accumulated storage of clinical case files and family pedigrees. "Human genetics was for most of its history more or less restricted to studying pedigrees which stood out in the way that the geneticists discover an abnormality of a rare disease" (Jones, 1993: 17). Indeed, from a historical point of view, the use and development of pedigrees are coincident with the rise of human genetics and eugenics (Resta, 1993: 236).

Two of the most fundamental notions in genetics are the notions of *phenotype* and *genotype*. Harper (1993) defined these concepts as follows:

> **Phenotype:** the visible expression of the action of a particular gene; the clinical picture *resulting from* a genetic disorder (emphasis added).
> **Genotype:** the genetic constitution of an individual (either overall or referring to a specific gene locus).

These definition contain two important elements: causality and visibility. According to genetic theories, a phenotype represents the external features of a genetic disease while a genotype represents its internal characteristics. Thus, the genotype is prior to the phenotype; the externalized phenotype results from the internalized genotype. I am going to discuss the issue of causality in genetic theories later. Here I would like to focus on the issue of visibility.

It may safely be argued that the relationship between phenotype and genotype is regulated by ideas concerning the visibility of Nature. Genetic theories suggest that the phenotype is the visual manifestation of the genotype. A pedigree is considered a visual tool for representing the phenotype of the family affected by a certain disease. Genetic tests, particularly the recently developed molecular biological test kits, are used to show genotypes (Yarborough et al., 1989). It is thus possible to say that both concepts are based on the visual tools by which geneticists make "visible" the invisible knowledge of the body, of the family, and even of "Nature." In this respect, geneticists' visual images can be compared to the anatomical gaze at the body, even though their arguments are grounded in a different discipline. According to Foucault (1973), a new way of conceptualizing the body resulted in a radical shift in both medical knowledge and medical practice, and even in representations of the world. In this sense, the "anatomical gaze" can be associated with the development of geographical "maps" of the world.

A 'map' is one of the most popular analogies in genetics; for example, geneticists often use the word gene mapping, or human mapping. In the book *The language of genes*, Jones (1993: 41) explains how genetics is rooted in map imagery:

> Genetics, like geography, is about maps; in this case the inherited map of ourselves. Not until the invention of accurate clocks and compasses two thousand years after Herodotus was it possible to measure real distances on the earth's surface. Once these had been perfected, good maps soon appeared and Herodotus was made to look somewhat foolish. Now the same thing is happening in biology. Geneticists, it appears, were until only a few years ago making the same mistakes as the ancient Greeks. Just as in

> mapping the world, progress in mapping genes had to wait for technology.

However, Jones fails to mention a taken-for-granted aspect of geographical maps, namely that there are no comprehensive geographical maps which accurately represent all components of a fact such as shape, direction, area, and distance. All maps embody a certain degree of bias, because a geographical map involves a reduction from a three-dimensional reality to a two-dimensional language (e.g., Rudwick, 1976). From this point of view, it is also to be expected that a genetic "map" will not represent all the components of the genetic make-up.

Wood (1992: 116), in *The Power of Maps*, analyzed the different functions of maps:

> (A) [m]ap [...] is comprehended in two ways. As a medium of language (in the broad sense) it serves as a visual analogue of phenomena, anticipation of experience, to compare or contrast, measure or appraise, analyze or predict. [....] As myth, however, it refers to itself and its makers, and to a world seen quite subjectively through their eyes. It trades in values and ambitions; it is politicized.

According to Wood (1992), a map acts as a focusing device between two significations; as a medium of language, a map internalizes signs, whereas, as myth, a map externalizes signs. Jones (1993) appears to focus on the former aspects of genetics as a medium of language, without addressing the issue of genetics as myth.

A central question will be thus as follows: Given that "pedigrees attempt to compact complex relationships and information into a graphical simple model" (Resta, 1993: 236), what is the bias of pedigrees? Fleck (1947 [1986]: 134) suggested that "[t]o see, one has first to know, and then to know how, and to forget part of the knowledge." What do genetic counselors know before taking a family history? How do they know a family history? How do they forget part of the family history? It is important to understand how a medical pedigree marks the transcription of family narratives of illness into a visual document.

2) Clinical Practices and Pedigrees

In the book *Ethics and Human Genetics*, Fletcher recalled his personal experience with genetic counseling services. "I and countless others were told that my father lost his hearing in 1904, when at age four, he was 'struck by lightning' standing on the back porch of his Alabama farm home from watching a storm. [....] The story of my father's miraculous survival was literally a legend in his time" (Wert and Fletcher, 1989: xxviii). However, "[b]y 1980, my growing involvement with medical geneticists, unanswered questions about the actual cause of my parent's deafness, and three maturing children led me to seek help from genetic counselors to assess the genetic risks of deafness in our family" (Ibid.: xxviii). The story of his father's deafness was examined through discussions with neurologists and with his father's cousin, and Fletcher realized that the lightening story was false. His cousin said that his father had become "very sick, was taken to the doctor for a long time, and came back deaf" (Ibid.: xxviii). The "old" family story was examined, rejected and transformed into a medical pedigree.

In terms of a clinical perspective, pedigrees are one of the most important documents in genetic counseling services and research. Without any prior knowledge of the family history, the geneticists can hardly verify and diagnose "genetic diseases." Gelehrter and Collins (1990: 255-256) clarify the three reasons why taking a family history is important:

> First, the family history provides an aid to reaching a correct diagnosis. [....]
> The second reason for obtaining the family history is to determine an accurate prognosis. [...]
> The third and most important use of the family history is for a presymptomatic diagnosis of genetic disease and the prevention or avoidance of clinical disease.

It is thus hardly surprising that genetic counseling practitioners are usually required by clinical guidelines to take a family history when they first meet with a patient.

Pedigrees are also important tools for connecting the various sectors of the geneticists' community. A striking feature of the development of genetic services has been the growth of national and international collaborative networks (Thompson et al., 1991). Pedigrees have been instrumental in establishing links between genetic counselors and researchers, and they have been useful in forming a medical network: "Correct interpretation of family pedigree is essential for *human genetic research* and is particularly challenging when reviewing the pedigree diagrammed within *professional publications* or when research teams collaborate to study large families" (Bennett et al., 1995: 745. emphasis added). It is thus important to examine how a family tree is used as a boundary object in different settings and disciplines. This kind of analysis will allow us to perceive the socio-political aspects of pedigrees, as well as their role in medicalizing the family.

Finally, the role of pedigrees in publications need examination. The use of published medical pedigrees can not only lead to the production of genetic knowledge but can also be instrumental in reconstructing social organizations. A historically notorious example can be found in the Nazi's abuse of pedigrees. The Nuremberg Racial Law of 1935 illustrates the Nazi's view of what determined Jewishness or Aryan background and what kinds of marriages were allowed only under special circumstances (Geiehrter and Collins, 1993; Yonemoto, 1989). Cook-Deegan, a vice-president of the National Academy of Sciences, discussed how pedigree-based research will create new "problems" and lead investigators to cross the increasingly blurred line dividing clinical care from "social gate-keeping," He gave the following example:

> [O]ne group studying Huntington's disease has discussed a case in which an air traffic controller is at risk. The people appear to be responsible and the investigators have talked about when it might be necessary for the patient to notify the employer, but what should investigators do if the patient refuses despite symptoms of cognitive decline? [...] The long-term relationships and specialized knowledge of the condition under study make it impossible to draw a bright line between the domains of research, clinical care, and nonmedical social and gatekeeping functions (Cook-Deegan, 1994: 83).

Recently, human geneticists have started to discuss the issue of confidentiality in publications: "Unlike most other research where subjects have no ties among themselves except for their publication in the study, the subjects of pedigree research share a genetic heritage. This means that when one subject learns something about herself, she also learns about other family members" (Frankel and Teich, 1993: 6). The clinical issues may go beyond the rule of "informed consent" in publication, which concerns an individual patient's decisions. Some researchers experience a dilemma over the "duty to warn" the other family members, if they find out the "problems" of diseases (Cook-Deegan, 1994). As genetic tests have became popular in medical practice, the storage and retrieval of pedigrees from data-bases on computer network systems have became a routine practice. "The potential misuse or abuse of new genetic technology that will identify heterozygote carriers or people with genetic diseases is frequently contrasted and compared with previous events in the historical record pertaining to the isolation of the sick" (Markel, 1992: 209). This clearly calls for sociological investigation, because the use of biological technology is social and cultural rather than biological.

My review of the books and articles in social and medical disciplines shows that little research has been done on how the network of genetic data in pedigree research could influence social issues and how constructed medical records will be used for therapeutic purposes in the future. I will argue in this thesis that pedigrees embody a new social power that focuses on the health and illness of the family, rather than that of the individual patient.

CHAPTER 3. METHODOLOGY

A. Comparative Research Methods

I have resorted to a comparative approach because of my interest in analyzing "cultural differences" in medical discourse. Young (1978), a medical anthropologist, contrasted different medical discourses as follows:

> We can imagine the medical discourses of the world as being strung along a continuum. At one end are the discourses that rely on etiological explanations. For convenience sake, we refer to these as externalizing discourse since they concentrate on events and relations that are external to the sick person's body. At the opposite end of the continuum is discourse relying on functional explanations, i.e., internalizing discourses. While most of the world's medical discourses are located somewhere between the extremes, Western medicine is an extreme example of internalizing discourse (Young, 1978: 113).

In spite of its generality, Young's symmetrical model of the internalizing-externalizing medical discourses is useful for understanding the production of genetic knowledge in different cultures. According to the genetic discourse in Western medicine, some diseases are caused by genes inside the body. Although some geneticists are critical of the accepted theory (e.g., Wolf, 1995), many people believe that the power of genetic knowledge is derived from technological developments allowing to accurately represent internal "maps" of genes, or DNA markers (see Jones, 1993).

On the other hand, medical discourse in Japan tends to be externalizing, at least compared to "Western" medicine (Lock, 1986). Despite a long exposure to genetics, genetic training in Japanese medical schools is not as well

established as in North America (Takebe, 1994). This situation is partly due to the fact that genetic discourses are associated with negative social images and family problems (Fukushima, 1988). Given the presence of such an externalizing discourse, it is interesting to investigate the genetic counselors' and researchers' use of family pedigrees and family history in a cultural context different from the Western one.

In brief, my thesis also involves an analysis of the relationship between biomedicine and culture. By focusing on a "visual tool," the medical pedigree, in different cultural contexts, I intend to examine some of the links connecting the invisible and the visible world, as well as internalizing and externalizing discourses. Although a comprehensive examination of the link between biomedicine and culture is beyond the scope of this thesis, it can be argued that by focusing on inscription devices, a culturally-sensitive comparative methodology can lead to some fresh insights into the practices of genetic counseling services and research.

1) Comparative methods

The comparative research methods used in my project include interviews with genetic counselors, participant observation, and the content analysis of clinical documents and medical textbooks. Most of the findings originate in ethnographic fieldwork performed from January 1994 to August 1995. The fieldwork was conducted through semi-structured interviews in Canada with twenty nine clinical workers: four Ph.D. researchers, eight M.D. genetic counselors, fourteen M.S. genetic counselors, and three nurses, and in Japan with twenty seven clinical workers: three Ph.D. researchers, seventeen M.D. genetic counselors, seven public health nurses, and seven patients. My interviews involved both general questions and specific, open-ended ones. An example of the former is a question concerning the purpose of genetic counseling. An example of the latter is a request to describe how genetic counselors take down a "family history." Some interviews, namely those with laboratory workers, were carried out in informal conditions. In these cases, I did not use a tape recorder because I thought that it would interfere with the communication with the interviewee. As soon as I finished the interview, I recorded my recollections of the exchange and my impressions of the

interviews. Throughout this thesis, for confidentiality reasons, the names of the interviewees are pseudonymous. Real names are used only for the cases taken from published papers. The names of family members, practitioners, and hospitals in figures have been erased for the same reason.

Participant-observation was undertaken in Canada in two children's hospitals and a general hospital. In Japan two children's hospitals, a general hospital and a public health center were examined. Participant observation started with training courses, university lectures and a seminar for genetic counselors in Canada, and an intensive training seminar for genetic counseling in Japan. After I had acquired a basic knowledge of genetics and counseling, I began intensive participant observation at one site in both Canada and Japan; I attended more than ten sessions in a Children's hospital in Montreal and Tokyo. I communicated not only with genetic counselors but also with medical students in the hospitals. I subsequently participated in several sessions at various hospitals in Japan and Canada. I spent about a week at two other hospitals in Toronto. I attended a few sessions in a children's hospital, a general hospital, and a public health center located in the Tokyo metropolitan area.

The observations were often carried out by adopting the role of a student. For example, I participated in a training session for M.S. genetic counselors at a Children's Hospital in Montreal. During the fieldwork, I usually observed all the aspects of genetic counseling sessions including the pre-counseling stage, team conferences, the communication stage, and follow-ups. Given the delicate nature of the counseling sessions, I opted for taking notes as opposed to using tape recordings. Whenever I judged that my taking notes during a counseling session would disrupt the counseling process, I refrained even from taking ethnographic notes and wrote them down after the session on the basis of my recollections.

Finally, I have analyzed clinical documents and medical textbooks. During the fieldwork, I collected several examples of visual documents in Canada and Japan. I asked several counselors and researchers how and why they used a particular type of pedigree. When I found idiosyncratic symbols, I asked them why they used such symbols.

I also read and analyzed medical textbooks used by genetic counselors in clinical settings. The purpose of this analysis was to help me to understand clinical metaphors and behaviors. By informally socializing myself, as if I

were a medical student, I tried to understand their ways of thinking. I checked the list of books that they used. For example, although Harper's(1993) textbook is based on genetic counseling in England, I will use it extensively in this thesis, because it is one of the most popular textbooks in both Canada and Japan. Another example is Jones' (1993) *The language of genes.* A Japanese M.D. genetic counselor was absorbed in reading the book and told me that he was interested in translating it. Thus, I concluded that the book could also be considered as an important reference for Japanese geneticists.

2) Exploratory Approach

The research process can be described as exploratory and recursive insofar as I tried to discover the important role of a visual tool, by following genetic counselors' interests and practices. I started by studying several visual tools, namely family trees, family photographs, chromosome pictures, and ultrasound pictures. The subsequent focus on the family tree came from the fact that during my field work, the family tree was one of the most common and important tools used by genetic counselors and researchers in Canada and Japan. In addition, during the first phase of my research, I was not interested in how researchers other than genetic counselors -- for instance, laboratory workers and population geneticists -- used the pedigree for their research interests. However, during the second phase of my research I paid more attention to these other workers, since I had noticed that genetic counselors negotiate with laboratory workers to redraw and reconstruct the pedigree. In short, my research approach was exploratory because its focus has been determined by and through the process of conducting comparative research.

3) Limitations of the Research

As is to be expected for an M.A. thesis, my research had several limitations. The first concerns the amount and quality of the comparative data that I was able to collect in Canada and Japan.

Participant-observation was conducted in different ways in Japan and Canada. In Japan, I carried out an intensive observation of a small number of

genetic counselors, while in Canada, I was able to observe a greater variety of counseling cases with small group sessions. Except for an intensive observation at a children's hospital in Montreal, I carried out short-term observations, spending, for instance, a week observing genetic counseling in hospitals in Toronto. In Japan, thanks to the cooperation of an M.D. geneticist, I was able to select the counseling cases and even interview the patients after counseling. In Canada, I did not have the opportunity to select cases; rather the cases were chosen by the genetic counselors. These differences may bring about selection bias.

The interviews were not always the same, because my research questions differed according to the research contexts and phases. The interviewees in Japan were mostly medical geneticists, largely pediatricians and gynecologists. In Canada, the interviewees were often licensed genetic counselors rather than physicians. This bias may affect units of comparative analysis of genetic counseling.

My analysis of medical texts and articles resorts not only to Canadian and Japanese medical literature, but also to American texts. One reason for this is that the concept of genetic counseling is a North American construct. Another reason is that I tried to collect the medical textbooks which influenced genetic counseling in Japan. Many Japanese genetic counselors refer to the textbooks used in North America. As for articles, many Canadian geneticists and researchers and some Japanese genetic counselors are trained in the US and they often publish their articles in American medical journals.

CHAPTER 4. COMPARATIVE ANALYSIS

A. Encounters with the Family

It is reasonable to assume that there exists a difference between, on the one hand, the clinical settings, namely the production of clinical knowledge and, on the other hand, the family settings, namely the family accounts. In this thesis, I do not deal *directly* with the family settings, or the narratives given by the patient(s) and the family. Rather, my main focus in this thesis is on the clinical side, that is, the process of constructing of medical pedigrees in the preparatory stage of genetic counseling services. In this section, the first question to be discussed concerns the social and cultural settings of the genetic counselors and of the family, prior to the clinical interaction between them. In order to investigate the production of proto-data in genetic counseling, I will begin by considering two basic questions: Who records the family trees? What is the notion of the family like? My underlying hypothesis is that in both Canada and Japan, genetic counselors and the family negotiate the construction of medical pedigrees, although medical pedigrees have different meanings in these different contexts.

1) Who Records Family Trees?

Genetic counseling is performed by various types of health care workers: in Canada by Ph.D. geneticists, M.D. geneticists, M.S. counselors, and nurses, and, in Japan, commonly by M.D. counselors and public health nurses. Many genetic counselors do not seem to think that taking down family trees is a very exciting activity; they rather see it as one of their routine tasks (Resta and Wcislo, 1993: 9). The primary professional interests of M.D.'s, Ph.D.'s, and M.D.-Ph.D.'s genetic counselors were found to be significantly "more research-

oriented than clinically-oriented" (Sorenson and Culbert, 1977: 138). According to my research, M.S. genetic counselors (in Canada) and public health nurses (in Japan), rather than medical doctors, tend to specialize in taking family trees. An M genetic counselor in Canada claimed that "my particular job is to take the family history." Some M.S. counselors contrasted the role of M.D. geneticists and M.S. counselors as follows: "M.D. geneticists specialize in the diagnosis. We specialize in counseling and so we interpret the family history."

In Canada, 60 M.D. geneticists, 41 Ph.D. geneticists and 57 genetic associates are involved in prenatal diagnosis at genetic centers (Hamerton et al., 1993). "Genetic associates" are the health care workers who have either a background in counseling, such as social work or psychology, and learn genetics on the job, or those who have training in nursing, genetics, or other paramedical skills. Genetic associates are trained to play the practical role of counseling on the job. A training program for M.S. genetic counselors in Canada is at the present offered only by McGill University. Many associates who work in Canada are trained in the USA. The Canadian College of Medical Genetics (CCMG), more than a decade ago, established a system of guidelines and accreditation for the provision of prenatal diagnosis services. The CCMG requires all genetic centers to maintain clinical records: "identifying data on the patient and referred family, adequate for subsequent contacts with the consultands or for genetic or other follow-up." According to the CCMG requirements, "[r]ecords (files, charts) should be retrievable, at minimum, by some workable diagnostic category. [...] A basic, but conventional pedigree [should] at least [extend] to second degree relatives" (Unpublished Document). The clinical record legitimates genetic counseling services as medical treatment.

According to my observations, in Canada, medical teams are so organized that both M.D. and M.S. genetic counselors participate in the sessions, and after the sessions, they usually discuss patients' files in team conferences. In spite of some variation, the typical genetic counseling sessions consist of M.S. and M.D. genetic counselors interacting with a couple or a single patient.

In Japan, there is a division of labor between public health nurses and doctors; public health nurses who are trained in the "Family Planning Special Counseling Project" specialize in taking family trees, while M.D. doctors

establish the diagnosis and give medical information to the clients. The work performed by public health nurses is called "primary counseling." Nurses take family data as a preliminary to the general practitioners' "secondary counseling" in the general institutions. "Third counseling" is practiced by genetic counselors in specialized institutions; it is usually done by an M.D., on the basis of the medical pedigree constructed by the public health nurse. Yet, it is also true that because only a small number of trained public health nurses are available, many doctors take family trees.

According to my observations, teams of genetic counselors are poorly developed in Japan, contrary to the situation in Canada. The usual counseling sessions involve a single M.D. and more than two family members, such as relatives. One of the most popular policies of genetic counseling in Canada is "non-directive" counseling. Almost all Canadian genetic counselors I interviewed stressed the importance of "non-directive" counseling. In Japan, the doctors take more of an authoritarian role. A leading Japanese genetic counselor I interviewed suggested that a doctor's position of power over patients should be fostered in genetic counseling practice.

Taking family trees is not only the practitioner's role but also that of the patients. Many Japanese genetic counselors ask patients to draw their own family tree and bring them to the counseling session. Several reasons were given for this request. First, it saves time and energy for actual genetic counseling. Since genetic counseling fees are not covered by insurance, it is an important consideration.[6] In addition, practitioners claim that patients have difficulty in recalling accurate family data during the short counseling period. Moreover, patients may also be reluctant to discuss their family data in a crowded medical office where their privacy is usually poorly protected. In many clinics, genetic counseling offices are separated only by a curtain and so the patients' voices can be heard in the next room. For these reasons,[7] to take family data during the counseling session is by no means an easy task. The family trees, when transcribed by family members, are transformed into medical pedigrees by the counselors. In this thesis, I will consider the patients'

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[6] Because genetic counseling in Japan is not supported by the government as a medical task, genetic counseling is on a voluntary basis. Some counselors ask the clients to pay for it, but most do not.

[7] Another reason was that family data have historically been abused and used as a basis for discrimination in Japan.

transcriptions as part of the family data, since my focus is on the practitioner's construction of the family tree.

2) What is a Family?

Although the notion of the family seems at first to be unproblematic, there is a clear difference in the idea of family in Canada and Japan. For Canadians, the prototype of the family is the nuclear family consisting of a couple and child(ren), while for the Japanese, it is the extended family which consists not only of the couple and the child(ren) but also includes grandparents and even ancestors. Before proceeding to the analysis of the clinical side and of the processes of visual documentation, it is necessary to describe a few cultural aspects of the family and of the negotiations between genetic counselors and family members in Canada and Japan.

2-1) Nuclear Family and Ethnic Background: Canada

In Canada, genetic counselors pay attention to the couple's reproductive plans, since a family is defined by the presence of children. Indeed, the underlying assumption of genetic counseling is that the consultand wishes to have another child. This idea is supported by the members of a Canadian focus group:

> Results of the focus groups in preparation for the mailout survey indicated that a heterosexual married couple with children was what most participants considered to be "a family." During this preliminary investigation, other family structures were acknowledged. Nevertheless, what was fundamental to most participants' perception of a family was the presence of children (Decima Research, 1993: 63)

Although it is necessary to further examine the Canadian concept of the family, it can safely be said that in Canada, the notion of a family in connection with reproductive technology refers to the "nuclear" family.

Genetic counselors, therefore, generally assume that a couple who had an affected child will expect to be able to reckon the risk of malformation or any other effects on the next child. Indeed, genetic counselors strive to help the couple make reproductive plans for a new child as a result of counseling, especially when the couple is to have a child. If the couple is to have a child, genetic counselors are willing to help them to assess the risk. When I attended the final stage of counseling sessions, I noticed that genetic counselors were likely to ask the parents whether they were interested in having another baby. When parents said "Yes" counselors appeared to engage in more active counseling. However, when parents said "No" counselors seemed more reluctant to positively engage with them, especially when they could not offer any particular clinical treatment or genetic test. In one counseling session in a Canadian children's hospital, the following exchange took place:

> A father with a child diagnosed with a brain dysmorphism visited the counseling office to get the results of a genetic test and a prescription. The M.D. geneticist explained that there were no special treatments for the disease and asked whether he was going to have another baby. The father quietly said "No". He explained that he and his partner were too old and could not afford to have another child. Then the M.D. counselor said, "I am sorry, but there is nothing we can do." The father took a look at his child and muttered to himself, "So, no hope."

This case was not unusual and I have observed similiar cases in another hospital in Canada. During a counseling session an M.D. geneticist inquired about a couple's reproductive plan and tried to help them by performing a detailed physical examination of their first child. After the counseling, the geneticist told me that the physical examination was not necessary, but, because the parents expected a treatment, she had changed her mind and examined the child. It appears that genetic counselors in Canada are interested in assisting couples who plan to have a child in spite of being classified in the "high risk" category. The practice of genetic counseling is, therefore, influenced by what genetic counselors think is important, and in this sense, their work can be said to be shaped by cultural assumptions.

The concept of "family" in Canada is often associated with the notion of "ethnic group." Since many Canadian families have a history of immigration, patients tend to link their family to its ethnic origin. According to a survey of Canadian reproductive plans, over half (59%) of the respondents agreed that: "It is very important to carry my culture and ethnic heritage on through my children" (Decima Research, 1993: 65).

In genetic counseling in Canada, genetic counselors *always* ask the consultands about their ethnic background and put that information on the top (or the bottom) of the individual family tree. At a general hospital, the guidelines for making a pedigree issued by the genetic department contained the following item: "[Put the] [c]ountry of origin and ethnic origin for each branch of the family at top of pedigree for each division of the family i.e.; born Ont.(Italian)."

According to M.S. genetic counselors, the reason for writing down the ethnic background is to offer genetic tests which are related to a given ethnic background. For example, genetic counselors usually offer to patients of Afro-Canadian background a specific genetic test for "sickle-cell anemia," since the rate of this particular disease is higher among Afro-Canadians than among other ethnic groups.

I did *not* observe such procedures in genetic counseling in Japan at all. Japanese counselors take the origin of the consultee for granted and thus they do not ask questions about the ethnic origin. Furthermore, Japanese genetic counselors do not offer additional genetic tests linked to a particular ethnic background.

2-2) Extended Family and Marriage Counseling: Japan

The concept of family in Japan refers more often to the extended family than to the nuclear family. Although the number of nuclear families is increasing in Japan, the notion of the family in relation to genetic issues still refers to the extended family. Ohkura, a representative of the Japanese Association of Clinical Genetics and one of the most active genetic counselors in Japan, commented on the difference in the notion of family between Japan and America as follows:

> The Japanese notion of the family includes a couple, their children and even grandparents, while the American one does not appear to include grandparents, but only refers to the unit of "a couple and children." [...] When I question patients about their family, asking, for instance, "How many family members do you have?," they start to talk about the health of their grandparents and sometimes of their great-grandparents. Genetic counseling is supposed to be a family plan for the couple; however, in Japan grandparents also participate in genetic counseling (Ohkura, 1989: 9, my translation).

To be sure, given the lack of a firm empirical basis, Ohkura's emphasis on the difference between Japan and the USA cannot be divorced from its ideological connotations. It should, however, be noted that Japanese genetic counselors who participated in clinical sessions in the US do mention such differences in the attitudes of patients (e.g., Kawajima, 1992). One of the characteristics of genetic counseling in Japan is that the consultands are likely to come with a grandparent(s) (Saito, 1992). At times, it may happen also in Canada that grandparents visit a genetic counseling office with the couple. Genetic counselors in Canada indicated that the families of some ethnic groups, for instance, those of Middle Eastern background, come to the counseling session with their parents. Genetic counselors in Canada agreed, however, that these cases were rare. Even if the grandparents visit the office in Canada, the counselors will focus their attention on the proband, or the couple. During my field-work in Canada I have never observed grandparents attending genetic counseling sessions, while in Japan I observed that grandparents participated in the counseling and expressed their opinions to the counselors.

It would appear that cultural concepts of the family influence genetic counselors in their efforts to respond to the patient's needs. These include, in Japan, negotiations concerning marriage. Ohkura and Kimura (1989: 304, emphasis added) suggest that "in Japan, there is still the belief that the marriage of a man and a woman is also the union of their *entire family* unit." Indeed, genetic services in the pre-war period in Japan originated from what was called "eugenic marriage counseling" (*Yûsei kekkon soudan*) (Matsunaga,

1994).[8] Thus, it should not be surprising that modern counseling services in Japan are still often consulted on marriage issues. Dr. Sato, a gynecologist and one of the leading genetic counselors in Japan, defines genetic counseling as "the place to consult on heredity issues concerning marriage" (1988: 52). M.D. genetic counselors point out that the parents are concerned with the couple's marriage (Ohkura and Kimura, 1989; Sato, 1988; Saito, 1992; Iinuma, 1990). Fujiki et al. (1991), having examined 2376 cases of genetic counseling, showed that about half of the cases were related to marriage issues. In fact, as Sato points out, genetic counseling is used by family members to negotiate the marriage of the couple:

> [I]t happens that a partner's parents visit, as clients, the counselor. In particular, it should be noted that, especially when the parents appear to be interested in the family background, they may be planning to break up the proposed marriage (Sato, 1988: 56, my translation).

Iinuma (1990) reported on a counseling case in which parents tried to interfere with a proposed marriage by resorting to genetic counseling, because the parents thought that genetic disease was associated with so-called "Buraku-min," or the people who have been labeled as outcasts throughout Japan's history until recently. Kawamura (1994) suggests that once genetic problems, related to the wife, are noticed, many couples are likely to experience severe family reactions resulting in divorce.

One of the most popular clichés characterizing the relationship between a family and genetic counselors in Japan is: "heredity issues (*iden*) are taboo." Fukushima, an M.D. genetic counselor who observed and took genetic counseling courses in Canada and the US, pointed out that the Japanese word *iden* means both heredity, or inheritance and genetic disease; he believes that the Japanese word *iden* leads to a misunderstanding of genetics. For example, he suggests that many Japanese entertain false ideas such as the following: "if the parents are normal, they will not have any diseased

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[8] According to my interview with Matsunaga, the relationship between eugenic marriage counseling in the pre-war period and modern genetic counseling in the post-war period is questionable. My point, however, is that genetic services in Japan started under the name of "marriage counseling".

children" and "it is not 'ordinary' people who suffer from genetic disease and malformation" (Fukushima, 1988: 7). Accordingly, "the existence of genetic disease in a family is thought to be shameful, not only for the person affected, but for all members of the family, like 'losing the family name,' or having 'bad blood'" (Ohkura and Kimura, 1989: 304). From this cultural perspective, it seems that Japanese families do not make a distinction between phenotype and genotype. Rather, these notions are mutually adjusted to each other so that metaphorical expressions such as "bad blood" or even scientific terms such as "DNA" are used to indicate the problems of parents, other siblings, and even ancestors.

According to my fieldwork, Japanese genetic counselors, except in special cases, do not take detailed family data to the same extent as do Canadian genetic counselors. Although many leading genetic counselors insisted repeatedly, in both textbooks and medical journals, on the importance of taking family trees, the counselors, in practice, appeared to be reluctant to take detailed family trees for all patients. It is also possible that genetic counselors do not like to talk about the construction of family trees, probably because they were aware of the gap between ideal counseling procedures as explained in medical textbooks and their own practice. Many genetic counselors asked the patients to transcribe their own family trees; other Japanese genetic counselors took a *short pedigree* which consisted of first degree relatives (siblings, parents, offspring), or second degree relatives (uncles, aunts, nephews, nieces, and grandparents), but not third degree relatives (first cousins). In other words, genetic counselors in Japan tend to transcribe only the members of the nuclear family rather than that of the extended family. According to my observations, in Canada genetic counselors usually spend at least about fifteen minutes with all patients for the primary intake, while in Japan, the counselors, particularly M.D. counselors, are inclined to spend only about four or five minutes. I also interviewed a small number of patients in Japan who had experienced genetic counseling at various sites. Five of the seven patients I interviewed told me that they were rarely asked by the counselors about their family history. The point is that in Japan taking family trees is not perceived as part of routine clinical practice, and the process is often ignored in practice.

Some readers may note that detailed family histories are published in Japanese medical journals. To be sure, some Japanese geneticists do take down

detailed family trees. However, they mostly do so for research purposes. In terms of routine genetic counseling practices, Japanese counselors are not accustomed to taking down detailed medical pedigrees. One of the members of the Japanese Society of Human Genetics told me that Japanese geneticists performed poorly as far as the application of scientific knowledge to medical practice is concerned. Indeed, Takebe (1994) insists that there are few medical schools and institutions which offer a course on clinical genetics.

How different cultural understandings of the family have affected the manufacture of pedigrees is still an open question. Certainly, both genetic counselors in Canada and Japan take medical pedigrees. A comparative analysis of the genetic counselors' encounter with the family in Canada and Japan raises, however, several questions: Why do Canadian genetic counselors produce detailed transcriptions of family pedigrees, while the Japanese counselors do not do that? How are the notions of the family related to these different genetic counseling practices? How are different notions reflected in medical pedigrees? Given the prototype of the nuclear family in Canada, how do the genetic counselors represent that prototype on medical pedigrees? Given the prototype of the extended family in Japan, why do genetic counselors not construct comprehensive medical pedigrees in their daily practice?

3) Negotiations Between Genetic Counselors and Families

In my comparative analysis I consider that genetic counselors and family members carry out explicit and implicit negotiations during the construction of medical pedigrees. The manufacture of medical pedigrees can represent a threat for the whole family, since comprehensive pedigrees stigmatize all family members. A family pedigree may be used to argue that the illness is not a personal but a family issue. At least, the product of medical pedigrees may be construed as an expression of medical power over family relationships. For example, Saito (1992) described the medical power over the narrative of an "angrioneurotic edema" case as follows:

> The illness is so rare that few researchers and research facilities are concerned about it; there are also few support networks. The primary doctor suggested that the woman should not have many children because of the high risk of having a child with the same disease. Through primary counseling she came to believe that her illness was negatively weighted. Before the counseling, however, her own image of the illness was not particularly judgmental. To the contrary, especially when she was a child, she felt somehow comfortable, because there was something inside her body which was the same as her grandfather and father (Saito, 1992: 41, my translation).

Given the social and cultural emphasis on the importance of family ties, it should not be surprising that families try to conceal the relationship between illness and family. Therefore, both the family members and the genetic counselors are very sensitive to the making of medical pedigrees.

The same may be said, no doubt, of genetic counseling in Canada. A genetic counselor in Canada told me of the delicate aspect of the first phone call, in which genetic counselors usually ask for basic family information:

> How difficult it is for those people [the patients] to make a phone call [to us]! [When I make a first phone call to them], I do not want to spend 10 or 15 minutes, saying, "I am really busy; I can't talk now." So, if they want to talk about and are very anxious, you know, I will try to address the issues on the phone. The initial phone call is really, really important. A lot of times, the people [the other family members] know they [patients] have Huntington's disease. Many times they can't talk to the family. It is a taboo subject [that] the people are afraid of, and they have denial. So, they can't necessarily talk to their family. Physicians may not have much experience. So, a lot of the time patients have never really found the place where they could talk to professionals who understood the disease--sort of no threat, no

part of the family.[9] So, I think we are trying to give a good, comfortable, easy, supportive impression on the first contact.

The counselor has to establish a good relationship during the first contact with a patient, in order to be able to establish a better relationship later on with the family members, and by so doing, to obtain more comprehensive family information. Even though genetic counselors in both Canada and Japan may seem to share a similarly sensitive approach to the taking of medical pedigrees, their attitudes toward the latter are different with regards to the actual collection of data and the use of family records. Thus, the contents and meanings of family pedigrees should not be left out of a sociological analysis. In the following sections, I will focus on how medical pedigrees play a central role in the negotiations between family members, genetic counselors and other associated workers. My discussion of medical pedigrees will first examine primary transcription, then secondary transcription, combination, and, finally, publications.

B. Primary Transcription

In both Canada and Japan, primary transcriptions can be distinguished from secondary transcriptions in terms of the degree and amount of manipulation of family-data. Primary transcriptions are the first inscription derived from oral family stories. Secondary transcriptions are the processes by which genetic counselors correct, edit and reorganize the primary family tree into medical pedigrees. The difference is that the primary tree is usually grounded in the interaction between the genetic counselors and the patients, while the secondary trees are produced mainly by the counselors and other

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[9] According to Tibben et al.(1990), non-carriers in their study of Huntington's disease (HD) found that relatives reacted to their "privileged" position by "banning" them from the family because the HD tie had been severed.

associated workers. In this section, I will examine the process of primary transcription.

Primary transcription involves the first fixation of family data. Genetic counselors draw the family tree, listing the patient's near relatives by sex, age, state of health, and, in Canada, religion and ethnic background. Particular attention is devoted to the occurrence of relevant illnesses in the family. Primary transcription involves three components: the institutional format of the pedigree, the communication process between genetic counselors and the patients, and the genetic counselors' use of pedigree symbols and abbreviations. Genetic counselors usually bring the clinical format of local hospital charts to an intake office. Genetic counselors begin by asking several questions to the patients, either on the phone, or in a face-to-face situation, and transcribe what the patients say.

Primary transcription is characterized by the *translation* of the oral family narratives into clinical inscriptions. Through the use of a medical pedigree format and/or template, genetic counselors sketch a "collective" prototype, or a phenotypic representation of the family illness, by ignoring large parts of the subjective experiences of illness. Through this process, genetic counselors tend to perceive *the family as an expression of the illness.*

1) How do Genetic Counselors Start?

Harper, in *Practical Genetic Counselling* (1993), which was used as one of the most common textbooks among clinical workers in Canada and Japan, argues that the process of transcribing family trees usually follows a basic rule: counselors start from what they call the *proband* (or index case). The proband -- called the propositus (male) or proposita (female) -- is clearly indicated with an arrow. Harper (1993: 335, emphasis added) defines the *proband* as follows: The proband is "the affected individual through whom a family with genetic disorder is *ascertained.*"

When there is no proband, genetic counselors begin by transcribing the consultand couple. A genetic counselor in a children's hospital described the process of taking family data (Figure 1) as follows:

> Usually the male goes on left and the female on right. So let's say, *a little girl with Down's syndrome.* She would be colored in and in a legend would be Down's syndrome. There are different types of Down's syndrome. I usually write the type. The most common type is called trisomy. [....] Then we'll get different names and ages [of the children]. It is important. I would ask her if there were any other pregnancies. Let's say she said that she had three miscarriages before the child. I would ask for each miscarriage when it happened. Then I draw small black spots. Underneath, I usually write SAB, which means spontaneous abortion. And then the time; ten weeks whatever, each. [....] I find out when they happened, and whether or not they have hidden illnesses or any other exposures, whether or not she received any explanations, when the couple had three miscarriages along with Down syndrome (emphasis added).

Starting from the affected child transcribed as a shadowed icon with an arrow, genetic counselors go back at least two generations in the father's and mother's families. The concept of the proband seems to be clear as a starting point, but in practice it is problematic because genetic counselors often neglect the difference between the proband and the consultand. One molecular biology laboratory worker who had taken many varieties of pedigrees pointed out to me that many genetic counselors tend to forget this difference, and argued that proband and consultand should be described by different symbols. According to my observations, some counselors actually did indeed start from the couple rather than the proband. In their article "Recommendations for Standardized Human Pedigree Nomenclature," Bennett et al. (1995) defined the proband as the "first affected family member coming to medical attention" and the consultand as the "individual(s) seeking genetic counseling/testing." Nevertheless, the symbol of the arrow is still used in both cases, and the only difference is that the proband is signaled by a capital "P" near the arrow (see Figure 2). These facts imply that genetic counselors in practice do not mark the difference between proband and consultand, and that, as a starting point, they recognize both the proband and the consultand as being affected by a similar category of illness.

In the next section, starting from the descriptions found in textbooks and from hypothetical cases, I will analyze how genetic counselors put questions to the patients and, in so doing, elicit information from them.

2) How do Genetic Counselors Take the Family Data?

One of the important characteristics of primary transcription is that genetic counselors skip all the family data which are not useful as elementary data. A Canadian genetic counselor with a nursing background contrasted genetic counseling with nursing practice in a community care unit as follows:

> Maybe [genetic counseling] is a little bit more in-depth, and is more focused, because you are probably focusing on one particular aspect: [for example] prenatal and Huntington's disease. So, in a way, the difference is that a community care unit takes a more holistic approach.

Genetic counselors usually count the number and age of first degree relatives, second degree relatives, and third-degree relatives, and the counselors ask questions about their health and illness status. "In obtaining the family history one should ask about the same or related diseases found in the index case or patient, rather than about a list of diseases of 'familial tendencies'" (Gelehrter and Collins, 1990: 259). If counselors do not find any relevant information associated with the illness of the proband, they tend to skip the other details of family information. One of the procedural guidelines for taking pedigrees in a general hospital suggests that genetic counselors "[m]ay use a diamond to summarize, if family members do not have noteworthy problems and greater than first degree relatives" (see Figure 9).

Similar attitudes and procedures can be found in the department of Obstetrics and Gynecology. For example, when obstetricians and/or gynecologists practice genetic counseling they pay attention to the family members as carriers of genetic disease. "For all pregnancies, it is standard practice to determine whether a couple or anyone in either of their families has a disorder that might prove heritable" (Simpson and Golbus, 1992: 53).

Simpson and Golbus (1992) continue by stating the importance of recording abnormal features relevant to illness:

> Record abnormal reproductive outcomes such as repetitive spontaneous abortions, stillbirths, and anomalous liveborn infants. Couples with such histories should have chromosomal studies and other evaluations. Any patient who replies "YES" to a question should be offered appropriate counseling. If the patient declines further counseling or testing, this should be noted in the chart (Ibid.: 53).

Some counselors certainly think that what they do is to transcribe a "biological" family history (Bennett et al., 1995), so that the intake process is discipline-specific. However, we could ask to what extent the actual transcription is related to biological features only. In order to examine this question I would like to describe the questionnaires used during an actual intake process in Canada and Japan.

In a Canadian children's hospital, an M.D. geneticist, after a physical examination, started to take family data, by asking a native [illegible]an the following questions:

1 Where do you live?
2 Do you work?
3 Does your husband work?
4 What's his name?
5 What's your name?
6 How is it spelled?
11 Are you related to him?
12 Do you have other children?
13 How many?
14 How old are the boys?
15 The girl?
16 Is she O.K.?
17 Do you have brothers and sisters?
18 Do they have children?
19 How many?

20 How many children do your parents have?
21 Are all the children alive?
22 Are there any problems?
23 Are your parents alive?
24 Your mother died for what reasons?
25 How old was she?
26 How old are you?
27 Do you know someone who has a brain problem?
(--No, except my aunt.)
28 How old is she?
29 She is mentally retarded?
30 She can talk?
31 This is the only one you know?
32 Are your grandparents alive?
33 How about your husband's family?
34 How many?
(--Two sisters.)
35 Do they have children?
36 How old is your husband?
37 Is he healthy?
38 How old are his grandparents?
39 Is his grandmother healthy?
40 Died of what?
(--Lung disease.)
41 Lung cancer?
42 Is there any other mental problem?

These questions can be classified into two types: demographic questions, and questions about health and illness. The counselors tried to sort out the number, age, and sex of the family, and then to investigate the general features of the family such as mental problems. The interaction between the counselor and the consultand was one-way; the genetic counselor asked closed questions about family relationships and open-ended questions about the health condition of relatives. The particular case described may slightly differ from other counseling sessions, but, by-and-large, it is not very different from other practical cases I observed in Canada. Genetic counselors ask both

demographic and health-related questions. Family trees, in other words, are as close to "sociological" or "socio-demographic" research as they are to "biological" research. The sociological nature of this process will be further discussed in the final chapter.

National differences may be at work here so that the style of questioning I just described is probably typical of North American genetic counseling. By contrast, in Tokyo, in a general hospital, an M.D. genetic counselor asked the following questions to patients whose fetus had died during pregnancy:

1. Is your marriage a "blood-related" (consanguineous, *chi no tsunagari no aru*) one?
2. What is your husband's date of birth? I know your date of birth.
3. How was the pregnancy process?
4. Do you know why the fetus died?

In practice, M.D. genetic counselors are aware of the diagnosis because of a primary care doctor's referral letter, and so there is no need to take the history for genetic counseling. Rather, a short family tree is constructed, based on the diagnostic information provided by the primary care doctor. It could be argued that short history taking occurs only in special cases. However, according to my observations, this is not so. The M.D. counselor I observed did not usually ask for a detailed family history but limited himself to a couple of questions. Nevertheless, he always asked whether the couple had a consanguineous marriage or not.

To sum up, my description of the primary transcription of family data has shown the existence of differences between Canada and Japan, to which I will return in subsequent sections.

3) The Visual Construction of a 'Family Phenotype'

The construction of family trees results in a new visual representation of the family data. A symbol such as a square or a circle on the family trees is the icon of an individual body. The visual linkages among the icons promote both the idea of the family as a whole and of the illness as a property of the

whole family network. Shaded squares and/or circles in family trees provide additional symbolic tools to further characterize a number of icons, some of which can also be further distinguished by the use of colors. The juxtaposition of black and white icons results in a rhetoric appealing to our understanding and memory. The new representations externalize not only the visible, external disease as a shadowed icon, but also the invisible, not yet manifest illness of the carrier as identified by a partly shaded icon. In this sense, family trees lead us to perceive a collective "phenotype" of diseased bodies.

Some readers will object to this idea because the phenotype is usually associated with individual body features, elicited by physical examination. Indeed, medical geneticists usually make a physical examination after taking the family data, and the physical examination also indicates part of the phenotype of illness. However, the results of the physical examination are only used to develop an "individual" phenotype of the diseased body and, more importantly, the data of the physical examination are likely to be combined and symbolized as colored icons on a family tree. To put it another way, the family tree summarizes and aggregates the illness information related to the family members by contrasting normal and abnormal features of the bodies. The point is that the notion of "a family phenotype" is derived from the graphic language of family trees. The family tree as a visual tool allows genetic counselors to construct a "collective" phenotype of the family instead of an "individual" phenotype of the patient(s), or the proband. The collective phenotype objectifies the family relationships with regards to proband and carriers, while paying less attention to social and cultural aspects of family life.

In addition, family trees help genetic counselors to acquire a *visual memory* of the family phenotype. Visual memory is made up of an associative network of colored icons which, taken individually, have a low information content (Nakayama, 1990). A genetic counselor stressed the importance of visual memory in the following way:

> I use different colors to indicate developmental delay. So, in this way, a look at the pedigree, and these individuals will stand out. So you know what you are doing.
> [Q; In order to see . . . ?]

> In order to concentrate on a particular case. Because a lot of times I did the family history over the phone. Let's say [I take a family history] on April 13th. They'll come a week later. When I see them, I want to remember it. I have shaded in a color and by looking at the legend I know what I need to ask them. ..... I think the reason for doing a pedigree is that you have all the information to inform and summarize, and that everything about the family is there on paper.

Family trees, thus, provide visual information that can be easily memorized by genetic counselors. They act both as a tool and a constraint, allowing the counselor to focus on particular aspects of the illness common to family members, but also to set aside social histories of the family such as divorce, adoption, and other life-cycle events. Latour (1990: 42) describes the rhetoric of inscriptions as follows:

> [O]ne more inscription, one more trick to enhance contrast, one simple device to decrease background, or one coloring procedure might be enough, all things being equal, to swing the balance of power and turn an incredible statement into a creditable one that would be passed along without further modification.

Power relationships are absent from family trees. Family relationships are reconstructed as a linkage network. Compared to the other senses, vision presents the most static picture, revealing a structure more than a process (Gordon, 1988).

A family tree may lead genetic counselors and researchers to see the family in a particular way. Gray and Conneally (1993: 90), commenting on a pedigree study of Huntington's disease, claimed that:

> Huntington's disease is a family disease. Every member of the family is affected-emotionally, physically, socially--whether patient, at risk, or spouse.

They insisted that the disease lies not only in the patients, but also in the entire family. In other words, and as already mentioned, genetic

counselors and researchers in the primary stage tend to perceive *the family as an expression of the illness.*

The symbols and the formats used in the construction of a collective phenotype in primary transcription are not necessarily the same. In the next section I would like to show that there are some differences in the use of symbols and formats in Canada and Japan.

4) Diversity of Symbols and Formats: Canada

All medical institutions require medical practitioners to maintain medical records in order to document and legitimize their practice. Each institution and hospital I surveyed in my research had a pedigree format that provided for different shapes and meanings of the pedigree symbols. Despite some similarities, a surprising large variety of formats were found to exist in Canada and Japan.

Primary transcriptions are performed by individual counselors. According to each counselor's preferences, the meaning of a given shaded icon can be different. A Canadian M.D. geneticist explained to me that "as long as you put a mark in the legend box, you can use any kind of symbol you want." Although the family tree is often referred to as a "visual language," the visual translation process is not standardized. For example, in North America, pregnancy was symbolized in at least 17 different ways (Bennett et al., 1993), and some of the symbols used for pregnancy were attributed a another meaning by other genetic counselors. In particular, the spontaneous abortion of a fetus of unknown sex was symbolized in 12 different ways (Bennett et al., 1993). A genetic counselor with a nursing background claimed that whenever she looked at a pedigree, she could easily imagine who, in her department, had done it. The production of a family tree is part of the "art" of a counselor. In Canada, family pedigrees are required to include the genetic counselor's signature, because, without it, it would be hard to determine the meaning of the symbols.

The same is true of the overall format. Hospitals have different file formats for family trees. Among genetic counselors in Canada, two standardized types of family trees are, however, dominant: the straight and the semicircular one. A straight family tree (see Figure 3) is an ordinary diagram

constituted by various icons and straight lines that branch out like a tree. This type was utilized in general hospitals in Montreal, Ottawa and Toronto. A semicircular type was used in a pediatric hospital in Montreal (see Figure 4).

The semicircular type was designed in 1950s by Dr. Light, an M.D., Ph.D. geneticist. According to him, this particular type of family tree has since then been utilized by many medical practitioners in the Montreal area. While Dr. Light was working in a US university, he had to use a different format of family tree. After coming back to Canada, he returned to the semicircular one, because of his preferences. An M.S. genetic counselor explained the advantages of the semicircular family tree:

> My supposition is that it is so because you want to put as many people on as small a space as possible. Because you are limited to one page you use a half-circle.

It is noteworthy that the argument that one should "put many people on as small a space as possible" only applies to the younger generations. For the older generations there is no space for adding many people. Thus, the semi-circular family trees were suitable for representing nuclear families rather than extended families. The semi-circular type focuses on the child and the couple, and it does not make room for older generations as well as for complex family relationships such as consanguineous marriage.

The semi-circular family tree has other characteristics. The distance between the husband's family and the wife's family is large, whereas the distance in the grandfathers' generation is smaller. The semi-circular tree helps counselors to formulate questions about each family *separately*. The separation of the couple on the family tree also facilitates the task of asking separate questions about each member of the couple's ethnic background. It can thus be said that the semi-circular family tree reflects, to some degree, the notion of the nuclear family and the importance of ethnic background.

5) Symbols and Formats: Japan

The use of icons has, to some degree, been internationally standardized; the meanings of icons, however, were not, at least in Japan. Japanese genetic counselors utilize different types of symbols according to the academic groups to which they belong. Generally speaking, Japanese genetic counselors belong to two main groups: human geneticists and medical geneticists. Only three medical schools out of eighty schools in Japan offer a genetics course. Accordingly, most genetic counselors learn on their own, relying greatly on textbooks used in North America. Consequently, the association of human geneticists follows the style and methods of human genetics in North America and Europe, resorting to icons used in such textbooks as Thompson & Thompson (1991) and Harper (1993).

The medical geneticists' group has for a long time followed a paternalistic orientation. Dr. Ohkura, a medical geneticist and founder of the association, every year organizes training seminars for doctors and public health nurses who want to learn genetic counseling. This is the only Japanese public institution that provides an educational course in genetic counseling. Through Dr. Ohkura's efforts, the number of medical geneticists has increased; as a consequence, his particular way of using symbols and his approach have been widely reproduced by practitioners, particularly by health care nurses (see, for example, Uchida, 1987). All public health nurses I interviewed had adopted his style of pedigree symbols.

Since the two associations have a different history, and they work separately, their use of icons on family trees is clearly different. For example, the symbol "ø" is used as "diagnosed person" or "examined" among the medical geneticists, while among the human geneticists it refers to a "diseased person" or "dead." For the medical geneticists, the symbol of "disease" is the cross symbol "†." The interesting point is that both groups of practitioners seemed not to be aware of the difference in the symbols they used until I pointed it out to them. This supports my claim that the two geneticists' communities are isolated from each other.

Furthermore, genetic counselors in Japan do not resort to templates in transcribing family tree symbols, while in Canada, templates are often used by counselors. The use of templates result in a certain degree of standardization.

The use of different pedigree symbols and the rare use of templates indicate that in Japan primary transcriptions of family data cannot function as boundary objects connecting different social worlds. This function is played by secondary transcriptions, as discussed in the next section.

As for the overall format, family trees in Canada are drawn on an independent sheet of paper, while the clinical file in Japan includes a family tree as part of the background information sheet. The Japanese case files are characterized by a small blank space on the chart, reserved for the pedigree information. This implies that genetic counselors are not expected to take a detailed family tree even in primary contact.

I will argue that institutional factors account for the differences of family trees between Japan and Canada. In Japan, the two professional groups do not regulate the counselors' treatment of family trees, and rely on the individual practitioner's work, while in Canada the Canadian College of Medical Genetics requires genetic counseling centers to maintain family trees as primary records, and regularly monitors the practice.

C. Secondary Transcriptions

Secondary transcriptions involve the additional editing of family trees. They correspond to the passage from hand-written trees to a printed, more formal pedigree. The latter's extent and character are the outcomes of interactions between genetic counselors and other associated health care workers. While primary transcriptions are to some degree regulated by notions of the family, secondary transcriptions are regulated by clinical standards.

Secondary transcriptions involve the following processes:

Updated Versions
from old family data to new family data
Personal Versions

from a working draft to a final version

Team Versions

from an M.S./Nurse Genetic counselor to M.D. Geneticists

Requisition Versions

from a clinical pedigree to a requisition and/or fax sheet

One of the problems involved in obtaining family data is that the latter are not static (Gelehrter, 1983), although, once written down, the family trees become a static document. Given that genetics is a field in a state of flux, alterations or updates of the form will be periodically required. Because of the often tentative nature of recorded family information, genetic counselors usually tinker with previously gathered data.

Secondary transcriptions are grounded in clinical standardization (see, for example, Figure 5). Clinical standardization resorts to conventions of communication within the local clinical department of genetics, especially when genetic counselors are required to use a special format in the clinical context, when the practitioners hold their team conferences, and when they send the information to other practitioners on a requisition form. Although some genetic counselors sometimes use a working paper to transcribe family data, the working paper is transformed into a clinical paper after the counseling. The reformulated pedigree is used during clinical team conferences where it functions as a tool that allows team members to obtain "at a glance" an overview of the clinical situation under discussion. Furthermore, one of the important jobs of genetic counselors is to transmit the relevant family information to other laboratory workers involved in genetic tests. In particular, molecular biological researchers or clinicians are likely to request an accurate family pedigree, prior to DNA tests. I will discuss the combination of an individual pedigree with other pedigrees and with DNA tests in a later section. In this section, I will focus on the construction of an individual pedigree in local clinical contexts.

Some practitioners may reject the idea of secondary transcription, arguing that they take the family tree only once. To be sure, occasionally this can indeed be the case; however, when family trees are used as medical evidence, that is, if they have to be shown to associated health care workers, or if they are to be published in conference proceedings or in medical journals,

the original inscriptions have to be corrected or reworked. But how do genetic counselors construct secondary transcriptions?

According to my observations, the process of clinical transcription was characterized by standardization. For example, the chronological or generational order will be organized from the top to the bottom, and index numbers added to each icon. The underlying issue is that the purpose of the transformation of primary data is not to communicate with the family, but to share the information with other practitioners. This process leads counselors to perceive of *medical pedigrees as visual expressions of illness.*

I would like to discuss four versions of secondary pedigrees and to analyze how these secondary transcriptions intervene in the prediction of clinical facts.

1) Updated Versions: the Complexity of Family Data

One reason for producing secondary transcriptions lies in the complexity of family data, particularly in the case of consanguineous marriages. In Japan, the rate of consanguineous marriages, although decreasing, is higher than in Canada (Imaizumi, 1986; Ohkura and Kimura, 1989). When I attended an intensive training seminar for genetic counselors in Japan, the first topic covered was how to use the family tree symbols. Having explained the meanings of the symbols and icons, the teacher gave us an exercise involving the transcription of three cases of consanguineous marriage. For example:

> I am going to marry a woman whose mother is my father's younger sister, and my mother is her (the woman's) father's older sister.

I tried to transcribe this kind of family trees, but this was not easy, not only because of the complexity of the extended family relationships, but also because of variations in their expression. Indeed, when asked to draw the above-mentioned example on the board, three participants did it differently. The teacher told us that "even such simple family data enable us to make six

versions of the family tree, according to changes in the symbols' order." The point is that consanguineous marriages violate the usual rules of generation and sex order; in the same generation, older family members are usually transcribed on the right and younger on the left; in addition, men are usually on the right and women on the left. Since several versions can be produced, it is hard for even skillful counselors to make the best, clear family tree in a short period. In the case of consanguineous marriages, counselors are very likely to redraw the diagram. In addition, given the complexity of the extended family, it is almost impossible to transcribe a well-organized family tree in a one-shot transcription. Consanguineous marriage cases in Japan are a good example of how the notions of the family effect the process of transcription. Since many Japanese counselors are likely to encounter complex extended families, it does not make much sense to use a fixed, pre-organized medical format such as the Canadian one. Japanese counselors prefer to use the blank spaces on medical records, which allow them a certain degree of flexibility for representing complex family relationships.

The complexity of family data is not only a problem related to consanguineous marriages; it also shows up when updating family information. New information on the family often becomes available over time. Primary family data, sometimes of questionable value, can modify the family tree, thus affecting the diagnosis and risk calculations. The following case was observed (see Figure 6). Although a public health nurse took much time to take family data from a young woman whose father and two brothers suffered from phenylketonuria, she did not ask about other affected family members during the first intake. During the second interview, the nurse was told that the patient's cousin had the same disease. During the third interview, this information turned out to be incorrect and the nurse realized that it was the father's cousin who had the disease.

2) Personal Versions

The clinical case record format of pedigrees affects secondary transcriptions. An M.S. genetic counselor who specializes in prenatal diagnosis in a children's hospital in Montreal prefers to use a "straight" family

tree for primary transcriptions, although hospital files require the use of semi-circular medical pedigrees. Therefore, the original family tree produced during the counseling session has to be transformed into a semi-circular format. The original working sheet is destroyed and the second drawing is inscribed in the case record file.

In a general hospital in Montreal, an M.S. genetic counselor explained to me that he "personally" used blank papers for the "two copies" of the family tree: a "working copy" and a "good copy." During his first intake, he completed the primary transcription on the "working" paper. Then he would insert, "on the 'good copy,' circles and squares, and correct lines," adding "the name on the top, the date, the family name." He explained the reasons for the two versions as follows:

> Something comes up during the interview, which is relevant information like high-blood pressure. And afterwards, you usually add it in formally so that other people can read it. So I write the family tree again.

The transcription is not corrected during the first interview, since the family history has to be formalized and what the counselors perceive as important elements have to be emphasized. On the "good copy," blank spaces are left for further information that has yet to be provided, and the "good copy" is kept as the medical record file. The "good copy" is quite often organized chronologically. The other counselors can then examine the "good copy," and use it for their team conference.

Here again, the format of the case record leads to some degree of local standardization. Secondary transcriptions, as prescribed by the medical format, organize and classify the chronological order of generations, so that the position of individuals is indicated by Roman numerals for each generation. This particular format thus emphasizes the "historical" sequence of the family.

3) Team Versions

Genetic counseling is not always carried out by a single practitioner, but, often and at least in Canada, by a team of several counselors, usually a geneticist and a genetic associate, who collaborate in the transcription of the pedigree. In a general hospital, as part of a screening program, a secretary takes a primary intake over the phone, an M.S. genetic counselor adds more information in the face-to-face interaction with the patient, and then a geneticist checks and completes the pedigree in the subsequent counseling session. In a children's hospital, a senior genetic counselor told me jokingly, "I don't know how many people are working in this department." Not only genetic counselors, but also other biologists might cooperate in this collective endeavor; thus, the basic information contained in the pedigree is expected to bridge the various medical workers.

One of the most important aspects of the medical pedigree is its "at a glance"quality. A public health nurse in Japan emphasized this aspect when discussing how to transcribe the family tree as a clinical case file:

> The most important thing is not to put down too much information, but rather to select the information in order for the other associates to see and understand it *at-a-glance* (emphasis added).

In a Canadian children hospital I often noticed that during a case conference genetic counselors drew a rough sketch of the pedigree on the board to explain the family history to other participants. I also observed the participants correcting the pedigree drawn by the speaker. Thus, the pedigree actually becomes the focus of the discussion.[10] Indeed, on the basis of the medical pedigree, genetic counselors usually discuss, during team conferences, whether "the family history is positive or negative" in terms of diagnosis.

The "at-a-glance" requirement suggests that the amount of information displayed on secondary transcriptions is limited. The medical pedigree so constructed becomes the means of communicating with associates. Secondary

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[10] Suchman (1990: 314) has an insightful discussion of interaction at the "white board" using shared conceptual tools such as pedigrees.

transcriptions also help practitioners to keep their distance from complicated family illness episodes; medical practitioners consider that seeing the image is better than reading a report, and better than seeing the patient (Kaplan, forthcoming).

In this respect, the medical pedigree is an abstract ideogram, or the graphic display of certain ideas and certain meanings (Fleck, 1947). It results in selective interpretations where relevant meanings are displayed as a property of the icons. The medical pedigree can be interpreted as a visual language through which scientists communicate with each other and understand the genetic meanings of family data.

4) Requisition Versions

The pedigree is not only used by genetic counselors within a single department, but also by other associated workers in different departments or even hospitals: "The history must be recorded so that it communicates information to all health professionals caring for the patient" (Gelehrter, 1983: 122). Because of the variety of rare genetic diseases, genetic counselors sometimes have to contact a geneticist who specializes in a particular disease and who may be working in another medical institution. Some practitioners, therefore, will fax a medical pedigree to one of their colleagues. In these situations, they will resort to secondary transcriptions.

Figure 7-a and Figure 7-b show the difference between a primary transcription and a secondary transcription in Japan. In a Japanese children's hospital an M.D.-Ph.D. genetic counselor had transcribed the family tree in a medical record. As can be seen from Figure 6-a, he did not put the pedigree number and the generation number on the family tree. When he asked his colleagues for suggestions concerning a diagnosis of hemophilia, however, he added more information; he inserted the generational order and individual numbers, thus standardizing the family information (see Figure 6-b). In this case, both primary and secondary transcriptions were included in the blank space. The transformation from primary to secondary transcription was characterized by the standardization of the chronological order.

In Canada, requisition forms requesting family information for laboratory work include a pedigree. In Ontario, molecular diagnostic

laboratories require genetic counselors to complete a requisition form for DNA testing. The requisition form includes a note stating that "a pedigree must be provided before analysis can begin." The requisition form also includes the following list of items, to be checked by the counselor:

Reason for Referral

Documented family history of indicated disease
Possible family history of indicated disease
Symptoms of indicated disease in this individual
Other:-------

Provided details on pedigree.

It is interesting to see how, prior to DNA tests, the family history as documented by a pedigree is recognized as the indication of a disease. In using a requisition format, a family pedigree does not simply show family relationships, but, more importantly, it points to the presence of the indicated disease. The preliminary work for DNA tests, thus, highlights the relationship between the disease and the family. For the purpose of laboratory work, family data are summarized as follows:

Family Information

Have samples from this family been sent to a DNA lab before? Yes No. If Yes, specify------
Individual born in ------(place)
Ethnic background------
This individual in the index case
Or Name of Index case in the Family
Relationship to this patient.

The content of the family information stresses components such as a previous experience of DNA testing, birth place, ethnic background, and the relationship with the index case. The latter is a clinical construct produced by the primary transcription. Since the only concrete, demographic information

on the family is limited to such elements as birth place and ethnic background, it may be argued that the image of the family is likely to be associated with its ethnic background.

But how do laboratory workers deal with the pedigrees that are transmitted to them? This issue will be discussed in the section on "Combination." Before that, it is necessary to examine how pedigrees are stabilized in a clinical context.

5) Fact Stabilization

As described above, there are various versions of secondary transcriptions: updated, personal, team, and requisition versions. These versions are clearly different from primary transcriptions in three ways. First, secondary transcriptions have the legitimate status of "clinical evidence" as opposed to the proto-data status of primary transcriptions. Some counselors destroy the primary transcriptions, while others do not. In both cases, however, primary transcriptions are regarded as tentative 'clinical data' that have not yet been confirmed by genetic counselors. In other words, the process of secondary transcription involves the transformation of proto-data into clinical evidence. For example, the standardization of chronological order in requisition forms, by emphasizing a visible linear order, shows how medical pedigrees become a visual record rather than simply a sketch of the family. Moreover, genetic counselors are usually required to sign on secondary transcriptions, in order to show their responsibility for the clinical case "record."

Second, when reordering and stabilizing the initial family data, the focus of genetic counselors shifts from the family members to the family "pedigree." Clearly secondary transcriptions aim at and result from interactions with other clinical workers rather than with family members. Pedigrees in secondary transcriptions are regulated by local hospital standards rather than by the encounter with the family. From this perspective, it can be argued that secondary transcriptions imply a shift in

the genetic counselors' perception from "the family as an expression of illness," as embodied in the original transcription, to "the family *pedigree* as an expression of illness."

Third, secondary transcriptions increase the distance between the abstract icons and symbols used in pedigrees and the concrete contexts of primary data collection. The abstract chronological order of the symbols is also emphasized by secondary transcriptions. By standardizing family pedigrees, genetic counselors produce an objective image of the phenotypic aspects of a given illness and they thereby attain the anonymity of clinical evidence. The objective imagery of family pedigrees erases concrete individual narratives about illness. As pointed out by Berger and Luckmann, 1966: 64):

> An objectively available sign system bestows a status of incipient anonymity on the sedimented experience by *detaching* them from their original context of *concrete individual biographies* and making them generally available to all who share, or may share in the future, in the sign system in question (emphasis added).

It is rather interesting, from this point of view, that many practitioners believe that family tree inscriptions are universal, although, in reality, there are several small differences (Kelly, 1986). A genetic counselor expressed her hope that a universal pedigree format be adopted, and this discourse is indeed very popular among Japanese counselors, especially in medical textbooks (Ohkura, 1991; Iinuma et al., 1990; Uchida, 1987; Higurashi, 1976). An international, standardized pedigree would certainly contribute to an increase in the authority and legitimacy of genetic counseling.

In short, the fact stabilization produced by secondary transcriptions allows practitioners to construct the medical pedigree as a visual tool for communication. Secondary transcriptions focus our attention on the clinical evidence about phenotypic aspects of "family illness."

D. Combination

Combination is the process of articulating an individual family pedigree with other evidence such as other pedigrees and genetic tests. Combination is also a powerful technique for mobilizing "facts" that had been previously stabilized (Latour, 1990). Primary and secondary transcriptions are characterized by the genetic counselors' efforts to translate and stabilize complex family data in a clinical context. Combination is defined by the collective work across clinical worlds to mobilize and manage facts. As such, it functions as a "boundary object" (Star and Griesemer, 1989) that allows divergent uses, interpretations, and reconstructions to be practiced by the various health-care workers.

During my fieldwork, I encountered two main types of combination: "large pedigrees" and "laboratory pedigrees." A large pedigree is reconstructed by genetic counselors and/or pedigree researchers who routinely collect several individual pedigrees from various clinical offices and laboratories. The combined pedigree is used for the study of "family diseases" such as Huntington's disease and colon cancer. A laboratory pedigree results in a new, simplified pedigree transcribed mostly by molecular biologists. It is usually stored in what laboratory workers call a "Master File" in a molecular biology laboratory, and it is combined with genetic test results.

D-1. Large Pedigrees

1) Reconstruction of Individual Pedigrees

A large pedigree is the outcome of the combination of individual pedigrees. The resulting chart is likely to be kept as a new record, separated from individual case records. The construction of a large pedigree is most often part of a research project or a screening program for a certain disease (e.g., Figure 8).

In a general hospital in Canada, following the development in the late 1980's of a screening program for Huntington's Disease (HD), clinical charts and databases have been stored in the local computer network. As explained by the local genetic counselors, the head of the department focused on the screening program, and the program became one of the main departmental endeavors. Not surprisingly, the program has resulted in the rapid accumulation of data including many individual family pedigree files, to which were added the results of the improved ability to examine human chromosomes and markers in the laboratory, and data resulting from the combination of the collected individual pedigrees.

An M.D. genetic counselor, a member of the HD screening program's team, commented on the recording procedures in the following terms:

> What we do in Huntington's cases is surely tricky. Usually, I was speaking to the family with Huntington's Disease, certainly showing them a pedigree, but that turned out not to be a good thing to do. Because everybody understands things that we just add to a pedigree. But it turns out that maybe a family member A did not know what family member B had told us, and so [he/she will be] suspicious of how we got the information. What we do now is to construct a pedigree for each person who comes in, and that information is taken from their point of view. And then we put together a general pedigree and we don't show it to anybody else in the family. Each one has a separate chart, and we may have a big, big chart.

The interesting point is that the recording activity of the screening program has led to the birth of a large research pedigree that is quite different from a set of individual pedigrees. While an individual pedigree is transcribed as a clinical case file in the interactive process between the family and the genetic counselors, a large pedigree is the result of the collective work of genetic counselors and pedigree researchers. The goal is not clinical practice, but rather clinical research. Seven research-oriented counselors I interviewed noted that they would refrain from showing the combined pedigree produced by the screening program because of

confidentiality issues. Individual patients and family members, therefore, are not aware of the content of the large family pedigree.

As pointed out by Lisa Madlensky, a genetic counselor at the Mount-Sinai Hospital in Toronto, the production of a large, combined pedigree is a difficult and time-consuming endeavor. Madlensky deals on a daily basis with families affected by "hereditary, non-polyposis, colo-rectal cancer," which adds up to "roughly five percent of all colon cancer." She has been building a database since she joined the registry in April 1994. According to her, gathering a complete family history is critical, as with any adult-onset, genetically-linked disease. "It takes, on average, five to six months to work up a complete family history" (Roig, 1995: 17).

Population geneticists agree on the difficulties involved in completing a whole family pedigree. For example, Fujiki, a population geneticist and genetic counselor responsible for the construction of a very large family pedigree of an isolated village in Japan, noted that "[i]t is clear that it takes much time and energy to make complete pedigree charts for an entire village manually" (Fujiki, 1980: 12). He insisted that basic data must be combined, rearranged, and unified to form a new entity. These processes are rather complicated and they can be equated to puzzle-solving whereby the original data act as the individual pieces of the puzzle.

With the birth of the research pedigree, genetic counselors have refocused their attention on the collection of particular family data; when transcribing family trees, they are likely to ask new questions or to pay more attention to the answers to old ones. For instance, when I asked a genetic counselor whether she could make a complete pedigree, she answered: "Yeah. That's why a name and birth date are extremely important." This information is necessary in order to connect individual pedigrees with one another. Name and date of birth will allow researchers to locate a certain person in relation to the family. In practice, however, not all the counselors in a screening program are careful to take the necessary information concerning a person's name and date of birth. A genetic counselor told me that they would not do so out of "laziness." Obviously, it is not an easy task to put all the names and dates of birth on a pedigree. As a result, these data are often taken during secondary or subsequent intakes. With the unfolding of the screening programs, new local policies on how to construct pedigrees are developed by research teams. Figure 9 shows that in a hospital's guidelines for pedigrees, the requirement of

including names and dates of birth has been added by hand to the printed guidelines. This procedural revision supports the idea that research objectives can re-shape the questions asked during individual counseling encounters.

The accumulation of clinical data produces not only intentional research combinations, but also coincidental combinations. A genetic counselor in a children's hospital told me the following story:

> I saw a woman who has Myotonic Dystrophy. She had affected relatives who had not been tested in the lab. We got a clinical record; she arranged for us to have the clinical record of the relative who was affected. We sent her DNA blood sample to Ottawa, to DNA lab for genetic testing. I got a call from the lab there because they had another family of the same name. They wanted to know if they were part of the same family. So I pulled out the pedigree. And in Ottawa they pulled out their pedigree. They have enough laboratory records. I pulled out the record I had in my chart. And we had a couple with a similar name. [However,] I did not have birth dates. I did not hang onto uncles and cousins. Because they were not relevant to the counseling situation I had drawn, there were no uncle cases. I had to call back my patient and fill in the details on the pedigree. She was unable to give us the name and date of birth of many people, but at least [she gave us] the name of the grandparents. [Many details were lacking, but at least we had some sort of a pedigree], so that we could compare it to the pedigree in Ottawa. They sent it to me, but these pedigrees did not match. If they had matched to some extent, then [you would have to decide] how many names and dates of birth are needed to be similar. You are talking about *coincidences*. If you have got the same names and the same uncles and grandparents, you would have a certain number of criteria so that you could make a judgment. (emphasis added)

The accumulation of individual pedigrees will enhance the opportunities for associated health care workers to "communicate" with each other, to "discover" resemblance between family pedigrees, and even to engage in the construction of a new large pedigree.

Although I have described some features of large pedigrees, I have not discussed the social and cultural variations affecting their production. In the next section I will examine different reconstruction strategies.

2) How are Pedigrees Reconstructed?

Fujiki (1980: 1) distinguished two approaches for the reconstruction of pedigrees:

> There are two basic ways to reconstruct pedigrees: the conventional one is to obtain information on all relatives by questioning individuals, starting with one's parents and siblings and tracing lines back to all known ancestors; the second method requires several kinds of documentary files for a community covering a long period of time.

The first approach involves the cooperation of genetic counseling services, while the second approach is based on the availability of documentary files (see Figure 10).

According to the evidence I collected, the first approach seems to be popular in Canada where clinical workers collect family data directly from the patients and other relatives. In contrast, in Japan research geneticists tend to look for data in the documentary files of medical institutions. For example, in a general hospital in Canada, family data on Huntington's disease were usually collected by genetic counselors' intake; on the other hand, Kanazawa (1994) reported that his research group investigating the Huntington's disease in Japan collected the data by approaching many practitioners and was thus able to establish a data bank of about forty family pedigrees. Of course, there are also examples in Canada, namely in Quebec, of the use of church records in cooperation with historians and demographers. However, legal and ethical requirements such as the informed consent of the patients limit the use of documentary files in genetic research. This kind of legal and ethical requirements are stronger in Canada, severely limiting the use of private data. In Japan, on the other hand, geneticists, with the cooperation of the Minister

of Law, can easily access so-called household registers maintained by local administrations.[11]

In the next sections, I will look more closely at how the geneticists reconstruct large pedigrees in Japan.

3) Pedigrees and Household Registers in Japan

As mentioned above, Japanese genetic counselors, compared to Canadian ones, take less family data in clinical contexts. This being the case, how can they produce a large pedigree? In this section, I will further examine the methods used by Dr. Fujiki for his pedigree study, that focused on an inland community. Dr. Fujiki is a medical and population geneticist in the department of Genetics and Epidemiology at Fukui University. My account is based on my interviews with him and on an article entitled "Using the Family Linkages to Reconstruct the History of an Isolated Japanese Village" (1980).

According to Dr. Fujiki, a pedigree is defined as "a table or list of individuals known to be related through descent or marriage" (1980: 1). The purpose of his pedigree study of Laurence-Moon-Biedl syndrome, a rare autosomal recessive disease discovered in an isolated Japanese community, was "to investigate biological and sociological characteristics of an isolated community" (1980: 4). For this purpose, Dr. Fujiki's team "designed and revised a pedigree chart so as to make the best use of the data" (1980: 4). This involved the combination of the pedigree with the household register.

The utility of the household register (*koseki*) is to be found in the fact that:

> The *koseki* records vital events in the lives of family members (such as births, deaths, marriages, divorces, and adoptions), in a prescribed sequence in relationship to the family head. Because these records cover more than 100 years, pedigrees can be reconstructed without resorting to direct interviews (Fujiki, 1980: 1).

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[11] It should, however, be noted that a Japanese population geneticist suggested to me that while this was the case until about 10 years ago, it has now become more difficult to carry out this kind of research.

Various sources of information were used: the modern resident card (*juminhyo*), the present family register (*genkoseki*), an inactive register on which all individuals had been crossed off (*joseki*), the old registration files dating from the 1880s to 1946 (*hara koseki*), the restricted old family registers, which began in 1872 and continued to about 1886 (*jinshin koseki*), and the death registers maintained by Buddhist temples (*kakocho* and other documents). This shows that in Japan, family data centered on the household have been extensively documented by local administrations.

Fujiki adopted the following operational procedures to construct an "overall" pedigree:

> The information on the *koseki* record of each household is transcribed onto a blank sheet, showing the person and his parents, siblings, and children together with the name of the head of the house and the *koseki* number. It is similar to a family tree and includes as many relatives as possible. The old *koseki* records included many family subunits on one file, but recent *koseki* tend to show only a nuclear family, distributing data over several files. However, these files are numbered in order, so one can easily trace family to the other files and this also includes persons who have moved or died, so it is possible to trace back from the *koseki* records to the *joseki* and the *harakoseki* (Fujiki, 1980: 3).

His approach to data collection was to use data "chiefly from official records." More specifically, he investigated family names and registered numbers in order to trace back several household registers. The next step was to combine procedures leading to the production of a large pedigree:

> Next, pedigree sheets with the same surname are connected vertically, tracing a single family unit, and then horizontally, connecting all family units with the same parents and culminating in the compilation of an overall pedigree. In this way, all blood relationships between individuals, including those deceased, can be ascertained, resulting in a population three or four times as numerous as the present population (Fujiki, 1980: 3).

By resorting to official records rather than clinical intakes, Dr. Fujiki reconstructed and reorganized family relationships, ending up with the entire pedigree of a village. By using the household register, Dr. Fujiki was able to work around the difficulty of getting family data directly from the people. This does not mean, however, that he never tried to speak with the villagers. Indeed, as a preliminary step in this research, he worked as a primary care practitioner in the village and he then asked the village representative for permission to conduct the research. The methodological approach he adopted was the outcome of negotiations between his research group and the village people. Accordingly, the resulting large pedigree bears the marks of the social and cultural setting of which it is a simplified representation.

Indeed, the notion of *population* he adopted corresponds to a particular social and cultural viewpoint. Not only was the large pedigree reconstructed through the use of household documents, but the resulting representation was reconceptualized as a "village pedigree." The population inscribed on the large pedigree, including present inhabitants and dead ancestors, was recognized as a *village*. Figure 11 visualizes the history of the village by connecting the present-day family to a small number of ancestors. The village pedigree led to the calculation of the degree of inbreeding and thus to the construction of *common ancestors* in the previous generations. The population as represented on the pedigree became a new kind of visual evidence for the community. The notion of population, as used in Dr. Fujiki's research, is quite different from the notion of population used by Canadian geneticists; for instance, while the issue of ethnic background was absent from the former, it is an important part of the latter. Requisition sheets and clinical pedigrees used in Canada explicitly mention the ethnic background of family members. The point is not simply that Dr. Fujiki's research, having been conducted in remote Japanese villages, presupposes the idea of ethnic homogeneity as opposed to the idea of ethnic heterogeneity characterizing a country of immigration such as Canada. Rather, the important issue is that even the apparently simple notion of *population* can be grounded in different social and cultural understandings that need to be examined by comparative analysis.

D-2. Laboratory Pedigrees

A laboratory pedigree involves the combination of *pedigree icons* with the results of genetic tests, or DNA markers. The pedigree icons are obtained from secondary transcriptions. The term *laboratory pedigree* is not an official one but, rather, a colloquial term used by genetic counselors and molecular biologists. Before I analyze the laboratory pedigree in molecular biology, it is necessary to discuss the phenotype-genotype relationship, because the laboratory pedigree is used to articulate the phenotype and genotype of the family.

1) Phenotype-Genotype Relationship

The development of molecular biological technologies has allowed researchers to deal directly with genotypic information. Accordingly, some molecular biologists believe that the new genetic knowledge of the body will restructure our understanding of what it means to be human. Yonemoto (1994), a historian of science, noted that while many geneticists believe that DNA is the blueprint of life (i.e., that all human traits are inscribed in the DNA), all we can say at present is that DNA determines the structure and regulation of proteins. It is, however, generally accepted that there is a hierarchical relationship between genotype and phenotype: the internal genotype causes the external phenotype, or, in other words, the latter is the expression of the former.

In clinical contexts, however, without any preliminary knowledge of the family pedigree, a researcher cannot interpret the results of genetic tests. A clinical geneticist involved with DNA work pointed out to me that: "the pedigree information is critical to any DNA test." The important point is that the clinical use of genetic knowledge presupposes the availability of the proto-data elicited during genetic counseling sessions. With the development of an increasing variety of genetic tests, genetic researchers are likely to become more aware of the clinical importance of pedigree information. For example, during a conference concerning the Human Genome Project, Eunpu and Weiss (1993: 95) pointed out that "[m]olecular diagnostic techniques are being used for a growing number of inherited diseases; however, their use to

date has been almost entirely restricted to families in which there is prior knowledge of a specific genetic risk." Yarborough et al. (1989: 141) described the relationship between genetic counseling and the DNA marker method in the following terms:

> [t]he marker is not the gene, but is so close to it that it can be used to follow genes from parents to offspring. Markers can be used only in informative families, i.e., those in which the abnormal gene is present and associated with an identifiable marker that is distinguishable from the marker carried out by other family members. Before the marker method can be used in a given family, multiple family members must be tested to determine if the pattern or marker transmission can be used in the family to trace the abnormal genes and thus identify carriers.

With the growth of the marker method, genetic counselors are required to ask for specific kinds of family data. As was the case with pedigree researchers, genetic counselors who resort to genetic tests pay more attention to the name and the date of birth of the family members. For example, an M.D. geneticist in Canada commented on the growing importance of family names and dates of birth in the following way:

> Q; Do you put the name and birth date on pedigree?
> A: That's an interesting question. We are ..... I honestly do not, because I am used to [taking] a pedigree, based on my impressions. But now I am doing more and more DNA work. Then we have to do that. I have been very lazy up to now. For me, the pedigree is a clue to the family, to look at the pedigree and remember the family, but in the lab I forgot [the existence of] those families. So, now I try to be better. The [M.S.] counselors had to remind me as well. It's a lot of work. You are talking to the person, who often does not know the birth day of the siblings, parents, grandparents. It is hard to get that. ... We tried to be better because of DNA work.

For laboratory work, the pedigree is not a reminder of the family but becomes, instead, preliminary evidence for genetic tests. In many

laboratories, laboratory workers are not allowed to contact the family. The family pedigree as edited by genetic counselors becomes the primary connection between the family and the biologists. Family accounts are entirely absent from the laboratory. This fact is important because it indicates the transformation from the family's "thick" accounts into laboratory workers' use of "flat" inscriptions.[12]

There is, of course, an important difference between biology and clinical medicine. Molecular biologists have traditionally relied on experimental work with viruses, bacteria, and animals, while genetic counselors deal with humans and thus cannot control the conditions of fact production in the same way. While genetic disorders in laboratory animals are widely used as models for homologous defects in the human species (Wolf, 1995), the difference between human genetics and molecular biology is that the former deals more directly with issues related to the family, while the latter tends to overlook the notion of family. It could be said that molecular biologists, by resorting to laboratory organisms such as viruses and transgenic mice, have managed to control the social and cultural aspects of the body. When applied to human genetics, however, molecular biology needs more specific information on the family, in order to control for social and cultural 'artifacts'.

2) The Laboratory Pedigree

A laboratory pedigree contains a summary of genetic tests and it is usually stored in a master file (see Figure 12). The usual starting point of a laboratory pedigree is a requisition sheet that is sent by clinical departments or other laboratories. The laboratory pedigree is used by Ph.D. researchers, population geneticists, and molecular biologists.

The laboratory pedigree is basically a clinical pedigree to which various columns of genetic tests have been added. The focus of the laboratory pedigree, however, is no longer on chronological order, but on indexical categories of indicated diseases. Thus, the visual representations of the entire family relationships tend to be deconstructed in the laboratory pedigrees. However,

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[12] Latour (1986, 1990) insists that inscriptions are characterized by "flatness" in laboratory work.

the symbols and icons referring to individual family members still retain the same consistent meaning as in clinical pedigrees. The icons are used to distinguish "normal people" from "carriers" and "diseased people." Laboratory pedigrees link phenotypic differences to the genotype.

Indeed, the laboratory pedigree is constructed so as to represent a coherent phenotype-genotype relationship. In order to "see" this relationship, clinical workers mix the medical pedigree with the results of several genetic tests. Paul (1994: 52) clearly explained how the mobilization and the combination of stabilized, heterogeneous evidence is at the core of human genetics:

> [H]uman genetic information is more accurately defined as the composite product of several data sources and processes including: the elucidation of family history; the examination of genetically related phenotypic manifestation in family members; the results of biochemical, cytogenetic and DNA analytic testing; the genetic counseling of families and their responses to it; and the continuing care of those undergoing genetic examination to establish genotype/phenotype correlation.

Although Paul speaks of the "composite product of several data sources and processes," it is noteworthy that a laboratory pedigree corresponds more to the idea of "several data sources" than to the notion of data "processes." Fact mobilization constrains genetic counselors to focus their attention away from the "qualification" of proto-data to the "quantification" of the stabilized evidence. A laboratory pedigree, in other words, does not question the process by which family data are produced by genetic counselors. Some genetic counselors I interviewed suggested that laboratory workers often misinterpret pedigrees, because they are not familiar with the way genetic counselors take pedigrees and, often, with the counseling process as a whole.

A laboratory pedigree can be seen as a boundary object that bridges different social worlds. A laboratory pedigree provides evidence for the clinical workers' genetic interpretations. While individual icons of a pedigree are visual expressions of family illness, genetic tests show the *a priori* existence of genetic disease. Simultaneously, the laboratory pedigree erases

the documentation process that was involved in the production of the initial pedigree.

3) Why Do Researchers Mobilize Evidence?

I have described two tools used in fact mobilization: the large pedigree and the laboratory pedigree. But, why do counselors and researchers combine medical *evidence*? Why do they prefer to *quantify* visual evidence, rather than to *qualify* family data? One possible answer lies in the "uncertainty" of medical evidence, a factor openly recognized by geneticists and researchers. One of the most common statements during counseling sessions is that "there is no 100 percent certainty in medicine." Not only do M.D. geneticists use this message during counseling; but such sentences often appears in textbooks and articles. Harper (1993, emphasis added) in his textbook suggested that:

> [T]he chance that a definitely inflamed appendix will be found in an appendectomy is far from 100 percent, while the entire process of clinical diagnosis is based on the combination of numerous pieces of information, each with a degree of *uncertainty*, though this is often unappreciated by those involved."

And Fraser (1988: 204) noted that: "All tests have uncertainty, and it seems to me that the thing to do is to convey the uncertainty, as well as the information."

As a remedy for such "uncertainty," researchers attempt to increase the robustness of the data, by combining various approaches and by using the resulting, composite image as evidence of a given genetic disease. The mobilization of combined evidence is particularly evident in publications, to which the next section is devoted.

E. Publications

> The questions [of how to make the pedigree public] are far from clear,[...], since information is neither fully private nor fully public, but often somewhere in between (Cook-Deegan, 1994: 85).

The publication of pedigrees amounts to the presentation of an integrated visual image of genetic disease. Publication signals the transformation of medical documents such as secondary transcriptions and laboratory pedigrees into printed evidence. While secondary transcriptions and laboratory pedigrees are grounded in the negotiations between health care workers in clinical and laboratory contexts, publication presupposes a set of different negotiations among genetic counselors, pedigree researchers, clinical laboratory workers, journal editors and even family members as readers. The publication of a pedigree, therefore, raises new issues about the confidentiality and the ownership of the pedigree information. These issues have been recently discussed by some pedigree researchers. However, so far, there have been no clear answers as to the question of how to deal with the ownership of pedigrees.

1) Data Stabilization in Journals and Textbooks

Medical journals require researchers to follow a fixed format for publication. For instance, the *American Journal of Medical Genetics* (1995) has instructions for contributors about pedigree symbols (see Figure 13). Publication requirements are different from those directing the hand-written production of a locally standardized pedigree. The use of computer software programs introduces an additional dimension to the editing process and may prevent practitioners from publishing the locally standardized version of a pedigree. For instance, semi-circular family trees were at this stage transformed into straight family trees, because only the latter fit the format of the computer program. Medical journals insist that the medical pedigrees be chronologically standardized with Arabic numerals for an individual within a single generation. Clinical workers, accordingly, are required to modify the

style of pedigree manuscripts to fit the publication format. As noted in an article: "For a pedigree in physicians' records or hospital charts, it is useful to note the person's age next to, not inside, the symbol. In formal published pedigrees, however, the number usually refers to the person's number within that generation" (Gelehrter, 1983: 122). It should, however, also be noted that journal requirements have not achieved universal standardization. Even in publications, some differences can be seen in the medical pedigrees published by different academic teams.

The shift from manuscripts to publication raises the important issue of data stabilization. One of the major questions discussed by pedigree researchers is the dilemma between family confidentiality and the accuracy of scientific data. The accuracy problem in published pedigrees is linked to the fact that pedigrees can or should be altered to secure anonymity. However, because of the necessary breadth and depth of information conveyed by pedigrees, it may be difficult to preserve anonymity. Family members as well as other acquaintances may learn about the family history and the current health status of other family members who might prefer to maintain the confidentiality of that information. Some counselors developed a few "tricks" to modify published family pedigrees. Simpóson (1993: 49), a pedigree researcher on Bipolar Mood Disorder, mentioned that "[a]nother practice is to disguise the sex of family members, but this may decrease the scientific value of the pedigree." Frankel and Teich (1993: 31-32) argued that "disguising the identities of study participants by, for example, changing gender or age, or shifting birth order, has been a practice followed by some clinical researchers in reporting traditional medical case histories [...] and could be applied to pedigree studies."

Some readers could assume that, as far as pedigree researchers are able to obtain the informed consent of patients, there should be no problem in publishing pedigrees. However, relatives who are included in the study but have not been directly interviewed do not have the opportunity to give or withhold their informed consent to their "participation" in pedigree research. The issue of informed consent from non-participants in pedigree studies has been discussed at various conferences but no generally accepted solution has been found (International Committee of Medical Journal Editors, 1991; Frankel and Teich, 1993). The issue of family confidentiality is thus one of the factors involved in the fixation of medical pedigrees as published medical evidence.

Data stabilization is also performed in medical textbooks. Medical textbooks often use medical pedigrees to illustrate the family history of a particular genetic disease. For example, Harper (1990) explains the risk and nature of genetic diseases by using a family pedigree. For instance, the caption of pedigrees (see Figure 14) used to describe Mendelian disorders reads as follows:

> Figure 2.3 Polygenic inheritance simulating a mendelian pattern. Manic-depressive illness. (a) The superficial pedigree, with two generations affected, suggests dominant inheritance (autosomal or X-linked). (b) The recognition of affected individuals in both parental lines makes polygenic inheritance more likely than mendelian. (*Pedigree details have been modified for illustration purposes*) (Harper, 1993: 20, emphasis added).

In this "family history," all the concrete information was erased but the schematic expression of the pedigree was displayed in the textbook. As a result of this practice, geneticists have become accustomed to conceiving of a medical pedigree as the representation of illness. This is an important step in the socialization of geneticists and genetic counselors. Before clinical workers even start to treat family members in the counseling sessions, they have internalized a representation of genetic diseases that takes the form of a medical pedigree. Medical pedigrees are one of the most important tools for the socialization of human genetic students. As noted by a genetic counselor:

> My first exposure [to medical pedigree] was in a biology class on medical genetics, [although], I would say that the ability to use them was developed by on-the-job-training.

Medical pedigrees play an important role in reproducing a particular point of view: Printed pedigrees introduce the students to a new vision of genetic diseases. I would argue that the perception of the family history as the present expression of illness does not result from a short exposure to patients during clinical sessions; rather, it is the outcome of a long process of socialization. In this sense, the "family history" used by textbooks, which takes the form of medical pedigrees, leads genetic counselors and other associated

workers to develop and maintain a particular "way of seeing," the geneticists' vision of the family disease.

2) The Mobilization of Evidence in Medical Journals

In medical journals, geneticists and researchers tend to mobilize medical evidence and produce an integrated visual image of genetic disease. While individual bits of evidence are separately constructed in their practices, the mobilization of composite evidence constrains us to perceive a coherent image of genetic disease. For example, Figure 15 was published in the Japanese journal *Knowledge of Medicine* under the title "Seeing Genetic disease: Fragile X Disease."[13] The photographs represent both the inside and the outside of the body. The external body photographs show both the front and side of the external features as well as the particular feature of the penis. The internal photographs consist of several chromosome pictures. The first one represents the first proband case in the upper family history. The second one shows the proband in the second family history. The third one is that of the carrier. In addition to these body photographs, the figure shows the diagrams of two "family histories" whereby the colored icon refers to the illness. These different types of visual representations, taken separately, explain some aspects of the present Fragile X disease. As Kaplan (forthcoming) notes, readers may think of the imaging system as an integrated whole. By juxtaposing the various elements, the researchers construct an objectivated aggregate of an illness, each element of which can be separately produced by collaborating researchers.

What counts as "family history" is an abstract map of the family, dissociated from the meanings of the family narratives. The picture of the family history is nothing but a functional diagram that aims at presenting an objective image of the phenotypic expression of Fragile X. Gelehrter (1983: 121), a medical geneticist and genetic counselor insists that "the family history is most relevant when it is considered a part of the 'present illness' rather

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[13] This journal was used as material for an intensive genetic counseling training session in Japan. Although Fragile X is not a widespread disease in Japan, it is used in this paper as an example of combination.

than of the 'review of systems'."[14] Fact mobilization has the effect of dramatizing the "present" of the illness: "out there" and "now." The black-and-white icons work as an integrated image system associating the internal and external imagery of the genetic disease. Through the visual efficacy of a series of combined photographs, the family history is recognized as part of the "present" disease. The combination of medical evidence leads to the reclassification of the family history as disease, or, more precisely, of *the family history as part of the present expression of illness.*

Fact mobilization has a strong explanatory power because of its integrated aspects. The internal photographs can be associated as causal factors with external features, and the family history mediates between these two images. As described before, in the process of producing these images, the family history and the external body features are prior to the internal images; however, the juxtaposition of these pictures presents all these elements as simultaneous. One possible interpretation is that the internal photographs of the body are primary factors, while the discussion of the family history as represented by a pedigree is an icon of the collective body, and the external photographs are the direct expression of the body. The important point is not whether this interpretation is true of not. Rather, the issue is that the processes leading to the combination of visual evidence are erased and the photographs conjure up a deductive image of the genetic disease, divorced from the concrete meanings for the family of the affected family members.

3) Who Owns the Family History?

I have already briefly discussed the issue of ownership of family data in the section on data stabilization. Here I would like to return to this issue, from a slightly different point of view. In recent years, pedigree researchers have engaged in open discussions about the issue of who owns the collected pedigree data. Both pedigree researchers and journal editors have discussed the problem during an international conference (International Committee of

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[14] The "review of systems" refers to inquiries about the various diseases affecting the family members.

Medical Journal Editors, 1991; Frankel and Teich, 1993). However, they were not able to provide any clear answer to this issue. There is an important qualitative gap between the individual information for which a patient can sign an informed consent form and the collective information that adds up to a medical pedigree (Powers, 1993). The information gathered by informed consent forms concerns data independent from other people's personal data, while the information contained in the medical pedigrees, by necessity, includes other personal information. In pedigree research and counseling, however, the clinical practitioners tend not to distinguish between the two. Family data are stored under the name authorized in the informed consent forms, reconstructed as large, laboratory pedigrees and published as family history.

More importantly, there remains the question of who owns the constructed family history when the research ends. Since pedigree research is expensive and funded by grants, research projects may come to an end for lack of funds. According to Cook-Deegan (1994: 84),

> [W]hat happens to the data generated at considerable expense when a grant ends? Many call for destroying such information to protect family privacy, but this can fly in the face of the purpose for which the family members contributed data and samples; to find a disease-associated gene as quickly as possible.

An organization that funded a research project decided that they would own those charts at the end of the project (Frankel and Teich, 1993). However, the following questions remain: Do they also "own" the family history? Is there any guarantee that they can safely store and share the information? Is there any possibility for abuse of pedigrees?

It would seem that the research pedigree is neither the ownership of genetic counselors, nor simply of the family members, because it is a collective production of a set of persons, including various family members, genetic counselors, geneticists, laboratory workers, and even journal editors. Of course, the data on the family history are basically derived from the information provided by several family members. However, given that medical pedigrees are constructed by different workers, it is impossible to reduce the final result to the contribution of a single individual or group of individuals.

In other words, genetic counselors cannot simply give the research pedigree to an individual family member, because the research pedigree contains information concerning the entire family. Clinical workers do not routinely show pedigree data to each family members. Furthermore, since the information is constructed by many associated workers, it is hard to negotiate the ownership and the use of information. The interests of associated clinical workers are different. For example, laboratory workers who never meet family members may be interested in the accumulation of family data in order to acquire new genetic knowledge. On the other hand, genetic counselors may be more interested in issues of family privacy; how can they protect their privacy without endangering the counseling process?

The ownership issue is an important one because of the various social uses of the pedigree. Indeed, a family history can be used not only by medical, but also by non-medical institutions. McEwen et al. in an article entitled "A Survey of Medical Directors of Life Insurance Companies Concerning Use of Genetic Information" (1993), show that all the survey respondents in North America indicated that many life insurance companies already use "genetic" information in making their underwriting decisions, even if they do not perform or require "genetic tests" as such. On the basis of the applicants' existing medical records, many insurance companies rate the applicants in relation to selected genetic conditions. Of course, the medical insurance system in Canada and Japan is different from that in the US. However, it is important to stress that the ownership of the family history entails the actual "power" to control not only the patients, but also the family members. These issues can be related to the discrimination and stigmatization of the family, if the information is abused by non-medical institutions such as insurance companies. There are actually precedents for this, such as the abuse of the family history of Afro-Americans in the 1960s that led to the stigmatization of families (Markel, 1992).

Knowledge of the family history appears to be a new and potential political power and the basis for a new form of medical control. The "black box" of the ownership of the family history in genetic counseling services and research has not yet been fully examined by medical practitioners and social scientists.

CHAPTER 5. CONCLUSION

In conclusion, I would like to discuss three main points. The first issue concerns the cultural aspects of genetic counseling, and namely the fact that in Japan genetic counseling conflicts with ancestor worship. The second issue concern the process of fact stabilization and mobilization in relation to their "sociological" dimension. The third issue concerns the power of genetic knowledge, and it will be discussed in relation to the problem of the ownership of pedigrees.

A. Discussion

1) Genetic Counseling and Ancestor Worship in Japan

As described above, social and cultural notions of the family are erased during the visual fixation of medical pedigrees. Although my analysis is far from complete, I would like to focus on a specific aspect of genetic counseling, namely the fact that in Japan, the practice of genetic counseling is, in comparison with Canada, poorly accepted by family members and even by genetic counselors. Families hesitate to talk about genetic disease even during counseling sessions. Although Japan has had a long exposure to human genetics (Matsunaga, 1992), clinical genetics has not yet been recognized as a medical specialty by professionals and patients. Indeed, Japanese practitioners are eager to introduce new reproductive technologies, but they tend to neglect genetic counseling, and particularly, tasks related to the transcription of medical pedigrees.

The reasons for this negative attitude towards medical pedigrees, I have argued, is to be sought at least in part, in "ancestor worship." Smith has also argued that ancestor worship (1974, 1983) is one of the most important

foundations of contemporary Japanese culture which to this day strongly affects social values and attitudes towards everyday life activities, particularly reproductive plans. From a sociological perspective, Yagi (1984: 217, my translation) accounts for the marriage discrimination between so-called untouchable (*buraku*) people and *non-buraku* people in the following terms:

> Pre-modern society respected family discipline and a policy grounded in ancestor worship, but even in modern society this pre-modern tradition has been maintained, so that a form of paternalism has been institutionalized, combining a nationalistic ideology with the image of the emperor as the father of the nation, or as the "national family."

According to Yagi's analysis, marriage discrimination relies on traditional ancestor worship. Arranged marriages are still popular in Japan, and marriages are a matter of central importance to the family, because family members wish to have children who worship and protect them as well as the ancestors.

Now, there is a clear relationship between medical pedigrees and ancestor worship in Japan. Nakane (1967: 140, emphasis added) points out that:

> What is termed ancestor worship in Japan .... *visualizes* and is based on a concrete conception of ancestors in a series of generations going back directly from the dead parent to the founder of the house in which the family has its domicile. The recognized lineage of ancestors is fairly short, hardly going back further than that forefather who lives in the memory as a quite concrete personal figure.

Given that visualization of the family is part of ancestor worship,[15] medical representations of family data are problematic insofar as they stigmatize ancestors. Japanese people appear to be quite sensitive to any visual

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[15] Although Nakane literally refers to the form of a photograph, it is important to note that when constructing medical pedigrees, genetic counselors usually ask family members to bring family pictures (see Harper, 1993). My point is that both family photographs and medical pedigrees are characterized by externalizing discourses on the basis of visualization.

representation of negative family data. A DNA study group has grounded its critique of genetics research in the fact that the latter will connect illness to family data (DNA study group, 1994). Mass media such as NHK, the *Nippon Housou Kyoukai* (Japan Broadcast Cooperation), aired in 1994 a special program on genetic research, claiming that a genetic company in the USA had bought the family history of all the people in a small isolated island, and had been using these data on a computerized network.

To be sure, the formal rituals of ancestor worship are no longer practiced by many Japanese family in their daily life. Many publications has shown that modern Japan is a secular society: Secularization or the "disenchantment of the world" have taken places (e.g., Nudeshima, 1991). Nevertheless, the ideology of ancestor worship are still important in Japan (Smith, 1995). Namihira (1988) described that ancestor worship play an important role in medical fields where the concepts of death and life of patients are treated by practitioners.

Berger and Luckmann (1966: 75) have suggested that the political order is legitimated as a representation of the cosmic order:

> [T]he political order is legitimated by reference to a cosmic order of power and justice, political roles are legitimated as representation of these cosmic principles.

If ancestors play a vital role as cosmic principles, it should not be surprising that medical pedigrees represent part of the cosmic order. Thus, the construction of medical pedigrees in Japan is challenged on the ground that the cosmic order of the family is likely to end up under the control of medical practitioners.

2) Fact Stabilization and Mobilization

I analyzed my research findings by resorting to the empirical concepts of primary transcription, secondary transcription, and combination. These concepts can be interpreted in terms of Amann and Knorr-Cetina's model of

visual fixation. In primary transcription, the family proto-data are transcribed as family trees. In so doing, genetic counselors focus on the illness of the family. Secondary transcriptions complete a "local pedigree" through various types of editing processes. Then, in the clinical laboratory, the local pedigree is further reorganized by processes such as combination.

The fixation of facts discussed in this thesis corresponds not only to a simplification process as described by Star (1983), but also to a combination process whereby single pieces of evidence are recombined in order to produce theoretical interpretations. To put it another way, it is not simply a question of "fact stabilization" but also of 'fact mobilization' so that, by combining different sources of evidence, new concepts such as "population," "genotype," and even "genetic disease" appear. Genetic counselors do not limit themselves to the translation of clinical data. They also re-categorize these data to make them compatible with the requirements of laboratory researchers. In this way, genetic counselors negotiate with both the family members and the other clinical and laboratory workers.

One of the most important claims of this thesis is that although the phenotypic knowledge of the family comes first in the clinical and research processes, in the published visual representations, genetic knowledge appears as prior to phenotypic knowledge. Genetically-based diseases are recognized only after their existence has been described clinically or after genetic facts are used to define diagnostic or treatment groups. Thus, the relationship of genetic variation to disease or illness is always highly dependent on a variety of contextual factors including those which modify clinical signs and symptoms, and determine access to diagnostic or therapeutic services. Nevertheless, visual mobilization as used in publications erases these contextual factors and constructs a coherent image of the genetic disease which is derived from the *a priori* internalizing discourse of genetic theory.

The basic method of collecting family data in genetic counseling services and research resorts to a "sociological" approach insofar as the counselors focus on specific elements of the family data, code the family information as pedigree symbols, and produce a standardized "family history." The term "sociological" has been explicitly used by genetic counselors and human geneticists, for instance during the Annual Education Conference which was concerned with the Human Genome Project. On that occasion, two

genetic counselors, Bennett and Steinhaus (1992: 312, emphasis added), referred to the family history in the following terms:

> The family history is the most powerful tool of the genetic counselor. It serves not only as a diagnostic tool, but also as a *sociological* aid in counseling by serving as a record of family relationships.

In Japan, Fujiki (1981) also pointed out that genetic research is sociological. Genetic counselors and researchers are aware of the sociological nature of the family histories they construct. Of course, the exact meaning of the terms "sociological approach" and "sociology" is debatable. However, it is still noteworthy that genetic counselors think that the data collection in genetic counseling and research cannot simply be reduced to a biological dimension. Rather, the content of the questionnaires used by genetic counselors and researchers appears to be largely demographic.

3) The Body Politics of Medical Pedigrees

Although I suggested that there are some similarities between the "sociological" and "genetic" approaches, there is a clear difference between sociology and genetics, and the difference lies in body politics. The term body politics refers to the fact that individual and social bodies can be used as an artifact of social and political control by social institutions (Scheper-Hughes and Lock, 1987). Genetic counselors have the power to access the family members' body, as long as patients agree to collaborate. Indeed, genetic counselors ask their patients to bring family photographs along to the clinic in order to examine the bodily features of the family members.

More importantly, the clinical use of medical pedigrees can also be ascribed to body politics insofar as it provides the most graphic illustration of the subordination of the bodies of which the family is comprised. I have described these processes in the section on the visualization of pedigrees, claiming that genetic counselors and researchers tend to pay more attention to

the family history as part of the present expression of illness rather than as the interactive production of family narratives about illness. In medical textbooks, the prototype of genetic illness is often explained by the illustration of medical pedigrees. When clinical workers are socialized in genetic departments, the pedigrees are used not as an expression of family relationships, but as an icon of "genetic disease." In primary transcription, the family body is translated into schematic icons, which signify "normality," "abnormality" and even "potential" features of the illness. The visualization of healthy and unhealthy bodies leads to the overproduction of illness in which not only the individual body but also collective bodies are stigmatized as "family disease."

I speak of overproduction, because medical pedigrees promote, for instance, the existence of new pathological categories such as "disease carrier" (Markel, 1992). The carrier status is linked to unexpressed features of genetic disease, and thus carriers and their offspring might be required to estimate the potential risks of developing a genetic disease. Genetic counselors are particularly concerned about those who are seen as the carriers of genetic disease. The following example illustrates the notion of "overproduction." An M.D. genetic counselor explained the nature of a particular genetic disease to the parents, by saying, "This disease was not known ten years ago. The disease may not be known in developing countries." The number of new genetic diseases, including these diseases which can only be "seen" in developed countries, has been increasing, particularly in recent years. Medical pedigrees reproduced in medical textbooks and journals are used to illustrate a growing number of family diseases which can be "uncovered" in modern developed societies.

It could be objected that I am overstating my case, since the number of families currently resorting to genetic counseling is relatively small. To be sure, the number of people and services to which medical pedigrees may be presently applicable is limited. However, the potential danger is there, so that in the near future, genetic knowledge is expected to be applied to common diseases such as various forms of cancer, heart disease, and even psychiatric diseases. If these predictions are realized, the issue of medical pedigrees will no longer affect a small number of people, but will raise with renewed vigor the question of the medicalization of life (Freidson, 1970; Zola, 1972; Conrad and Schneider, 1980).

To sum up, although this thesis is only a preliminary study of the production and use of family pedigrees in Canada and Japan, my comparative analysis has led to three major conclusions: 1) the existence of different cultural interpretations of the notion of family affects the genetic counseling process; 2) the counseling process proceeds through five different stages of visual documentation that can be sociologically examined; and 3) behind the deceptive simplicity of medical pedigrees as clinical and research tools lies the largely unexplored field of body politics. First, cuitural notions of the family pre-date and shape the encounter between genetic counselors and family members in Canada and Japan. The notion of family in Canada is associated with the nuclear family, while that of the family in Japan is mostly related to the extended family. Family narratives in both Canada and Japan include not only negative images of family illness, but also positive elements that help family members to identify with the family. In Canada, counseling is mostly related to a couple's reproductive plans, while in Japan genetic counseling is associated with marriage counseling, because marriage involves family politics aimed at maintaining and managing heredity; thus, genetic issues are recognized as family problems. In Japan, ancestor worship conflicts with the production of medical pedigrees, since the latter visually objectify and even stigmatize the illness of the family and, thus the ancestors. Medical pedigrees are therefore not so popular as in Canada.

Second, my comparative analysis of clinical contexts shows that visual representations are not only the result of a simplification process, but also the outcome of local and interlocal adjustments between visual tools and various professional workers. More precisely, the process of visual documentation does not only lead to fact stabilization, but also achieves fact mobilization, or the combination of various forms of constructed evidence. Family stories are "translated" by genetic counselors into a primary transcription based on the local, personal use of the symbolic language of family trees. Through various negotiations between the family, M.D. geneticists, M.S. genetic counselors, and other associated workers, the pedigree is standardized as a secondary transcription on a clinical chart. Family pedigrees then assume the properties of boundary objects and are further reconstructed by associated health care workers and/or members of the research team. Finally, the publication of family pedigrees stresses an indexical interpretation of genetic disease

whereby genetic disease is reconstructed as an integrated visual representation. The family history, based on medical pedigrees, is categorized as the present expressions of the disease. Although family pedigree information pre-dates genetic knowledge in preliminary documentation, textbooks and journal articles present a visual image in which the genetic understanding of disease is prior to the phenotypic manifestation of disease as expressed in family pedigrees.

Third, the construction of medical pedigrees can be seen as part of the body politics of the family; the medical control of the pedigrees can lead to the social control of the family bodies. In practice, the taking of medical pedigrees implies a certain degree of stigmatization of the family; in primary transcription, the *family* is likely to be perceived as "an expression of illness;" in secondary transcriptions, the *family pedigree* is recognized as "an expression of illness," and in the combined imagery found in publications, the *family history* is looked upon as "part of the present expression of illness." Through the entire process, genetic counselors have the possibility of diverting the family narratives about illness.

The power of genetic knowledge lies not only in technological developments in laboratory work, but also in clinical work where the body politics affecting the family is practiced. A socio-demographic approach to the family practiced by the counselors can be combined with laboratory results. The reconstructed family history, insofar as the research pedigree is an outcome of collective work across diverse social worlds, is no longer the sole production of family members.

One of the implications of this thesis is that genetic counseling and the early analysis of pedigrees constrain clinical workers not only to erase the family background but also to equate the patients and the family with the disorder of which they become an expression. It is important to bear in mind that this leads to the possibility of overproducing family diseases. Finally, it cannot be emphasized strongly enough that this thesis is only a first step toward the analysis of the construction of the family history in genetic counseling practices. Although this thesis may have contributed an original perspective on the social and cultural issues pertaining to the clinical construction of medical pedigrees, the analysis of the data remains exploratory. The relationship between notions of the family and the clinical use of the family history needs further explorations. This thesis has not

examined genetic counseling from the point of view of the family. How do family members feel about their illness? How can their emotions be transcribed by medical practitioners and social scientists? How does the manufacture of medical pedigrees affects families? These and other questions await further investigation.

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All the Japanese titles were translated by myself.

Appendix: Figure 1-15

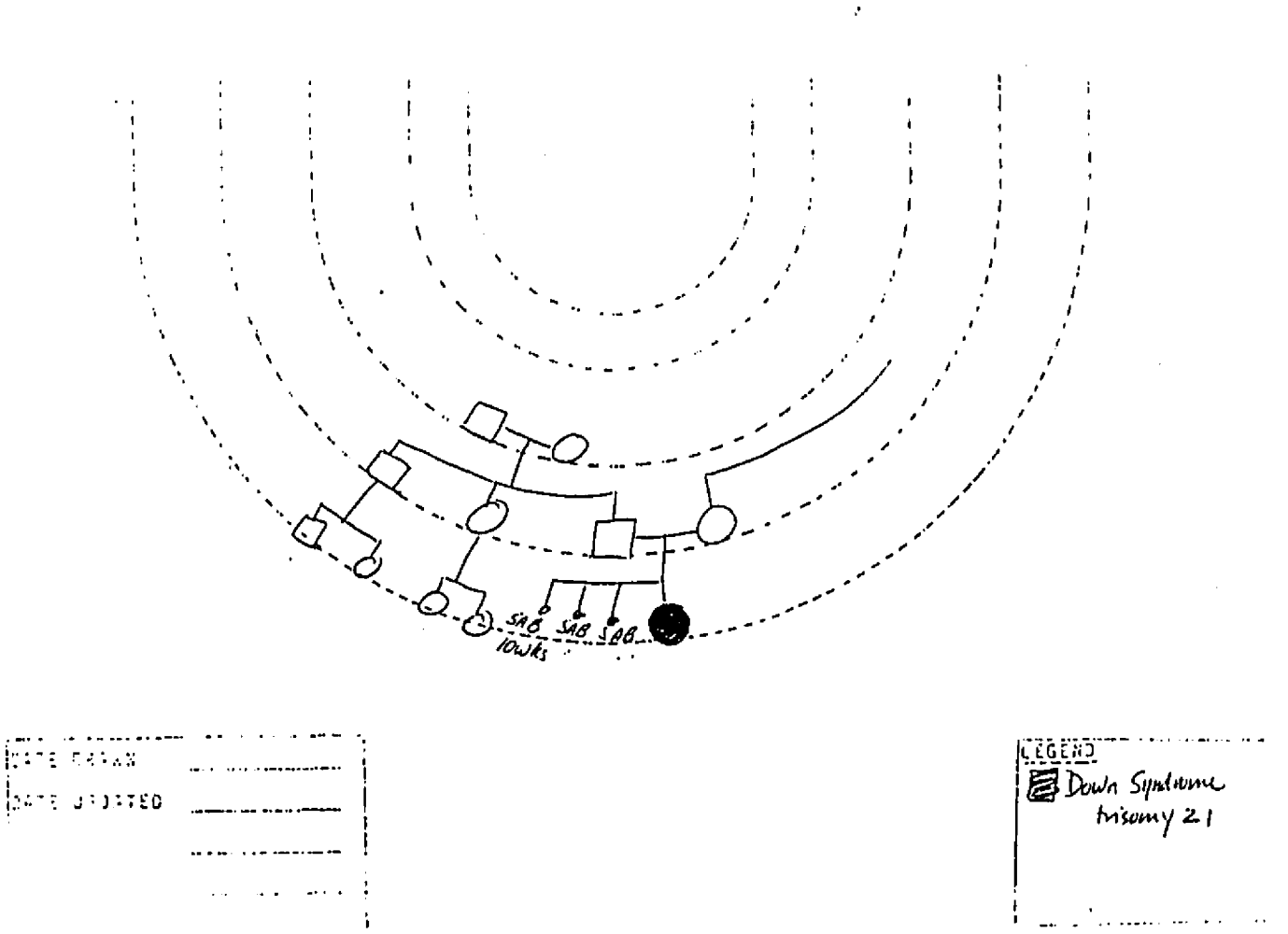


Figure 1: A Hypothetical Example of a Family Tree

Instructions:
— Key should contain all information relevant to interpretation of pedigree (e.g., define shading)
— For clinical (non-published) pedigrees, include:
a) family names/initials, when appropriate
b) name and title of person recording pedigree
c) historian (person relaying family history information)
d) date of intake/update
— Recommended order of information placed below symbol (below to lower right, if necessary):
a) age/date of birth or age at death
b) evaluation (see Figure 5)
c) pedigree number (e.g., I-1, I-2, I-3)

	Male	Female	Sex Unknown	Comments
1. Individual	b. 1925	30 y	4 mo	Assign gender by phenotype.
2. Affected individual				Key/legend used to define shading or other fill (e.g., hatches, dots, etc.).
				With ≥2 conditions, the individual's symbol should be partitioned accordingly, each segment shaded with a different fill and defined in legend.
3. Multiple individuals, number known	5	5	5	Number of siblings written inside symbol. (Affected individuals should not be grouped.)
4. Multiple individuals, number unknown	n	n	n	"n" used in place of "?" mark.
5a. Deceased individual	d. 35 y	d. 4 mo		Use of cross (†) may be confused with symbol for evaluated positive (+). If known, write "d." with age at death below symbol.
5b. Stillbirth (SB)	SB 28 wk	SB 30 wk	SB 34 wk	Birth of a dead child with gestational age noted.
6. Pregnancy (P)	P LMP: 7/1/94	P 20 wk	P	Gestational age and karyotype (if known) below symbol. Light shading can be used for affected and defined in key/legend.
7a. Proband	P	P	P	First affected family member coming to medical attention.
7b. Consultand			Individual(s) seeking genetic counseling/testing.	

Common pedigree symbols, definitions, and abbreviations

Figure 2: Symbols of Proband and Consultand
(Source: Bennett et al., 1995)

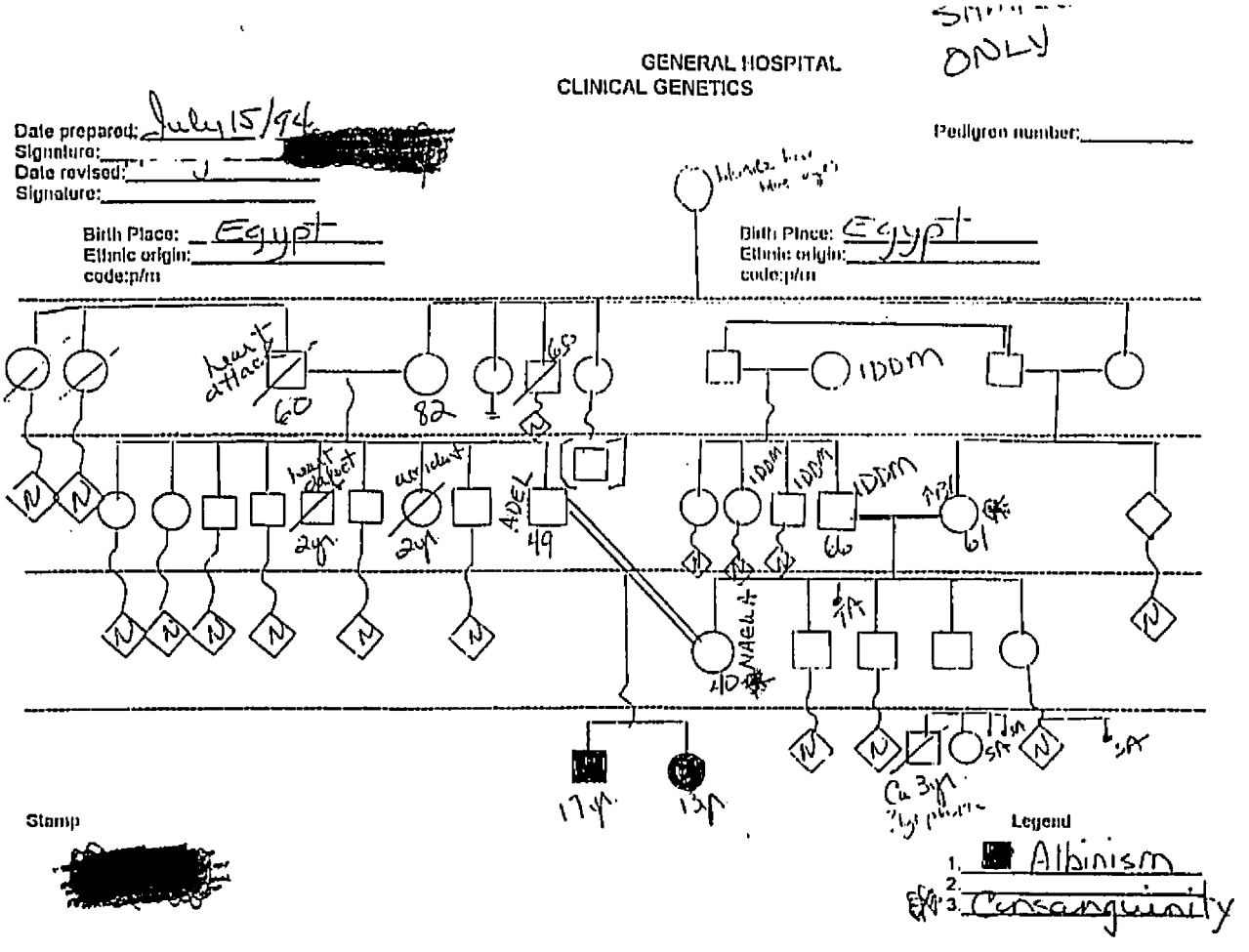


Figure 3: A Straight Family Tree

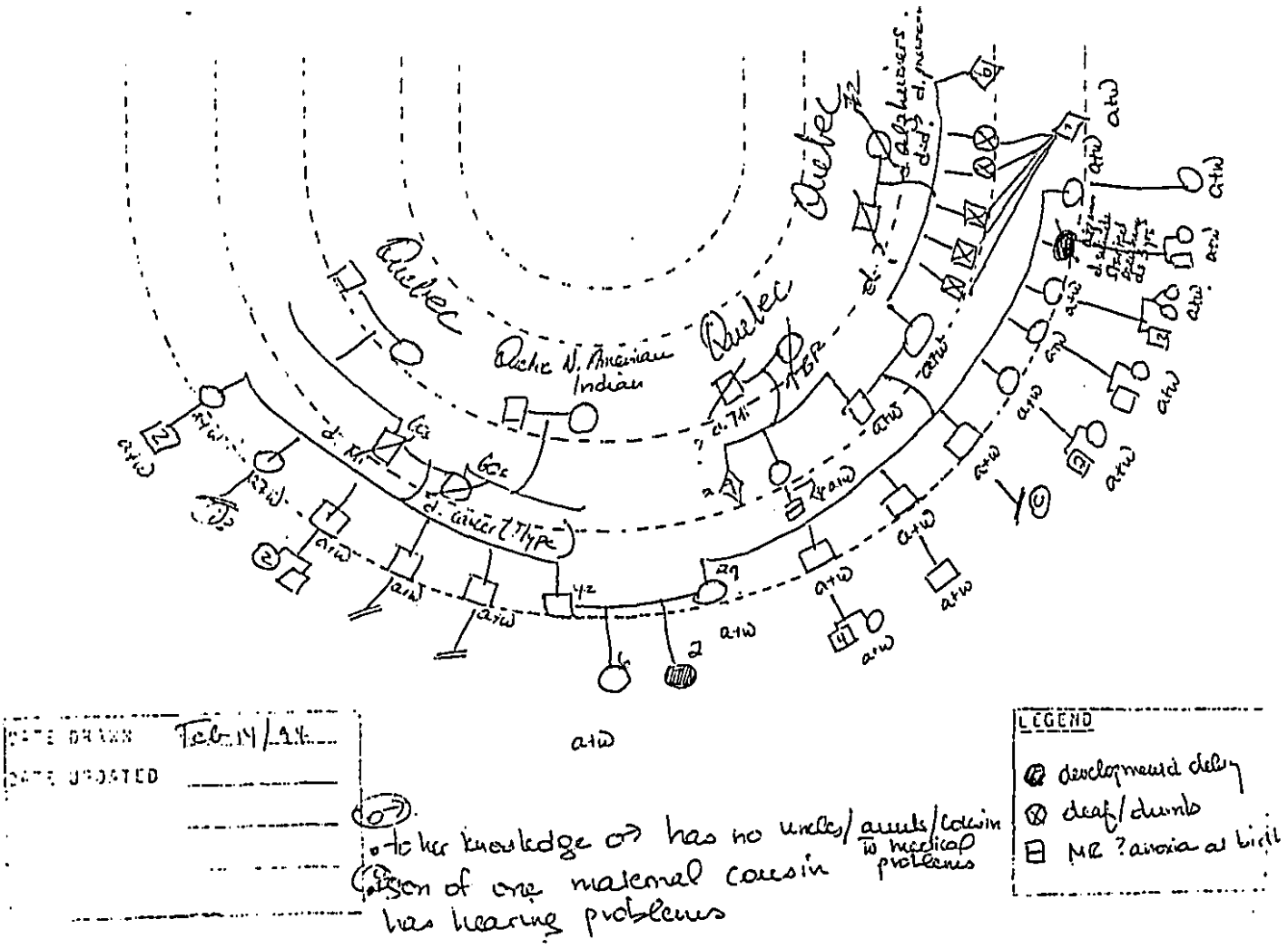
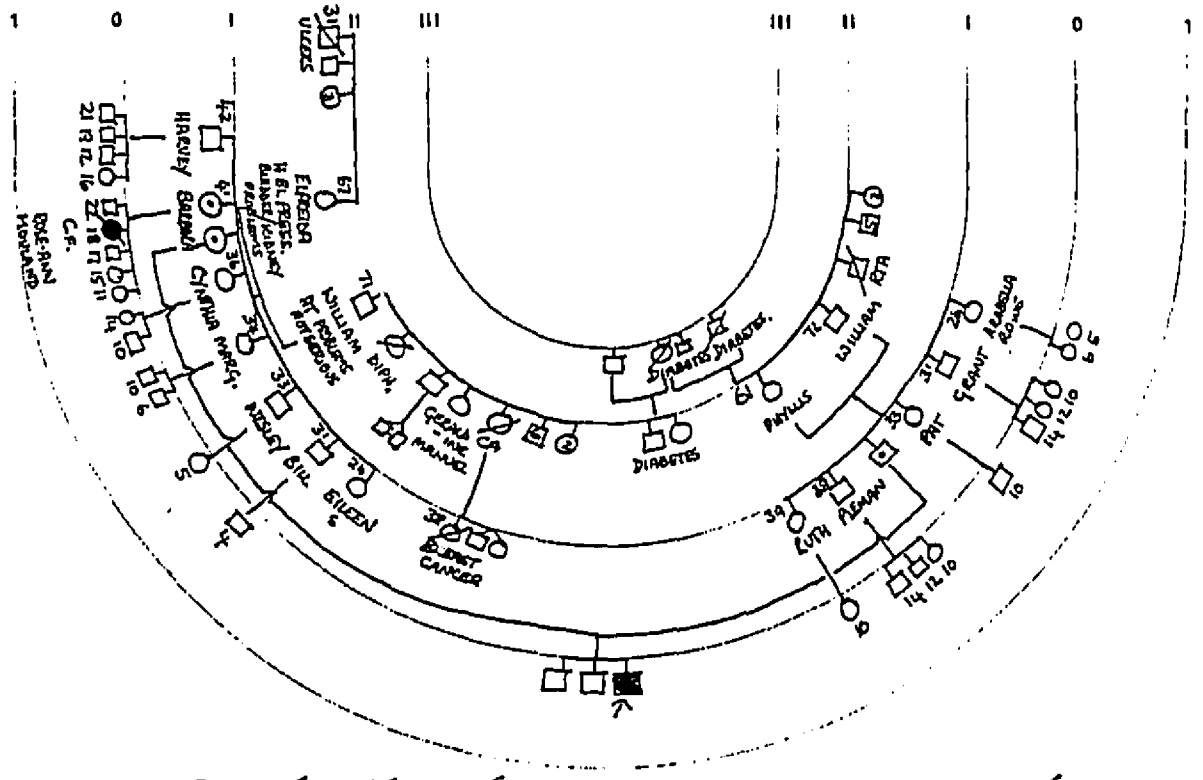


Figure 4: A Semi-Circular Family Tree



Record – abortions, stillbirths, cancer, cong. defects, diabetes, epilepsy, short stature, twins, mental retardation, conditions like proband's

Pedigree Number	Name	Address, hospital, diagnosis, confirmation of diagnosis, etc.
0 — 6		CYSTIC FIBROSIS DIED AGE 18 1982
1 — 16		HAD PROBLEMS WITH HIS HEART AS A BABY ? HOLE IN HEART
		IN HOSPITAL FOR A LONG TIME.
1 — 17		HAS TO HAVE A NEEDLE EVERY MONTH ? VIT. B. ? FOR LIVER
		GETS TIRED.
II — 8.		FINGERS LOOK LIKE TOES. MIDDLE FINGERS O.K. OTHERS JOINED
		TOGETHER. .. "FEET LOOKED FUNNY" "TOES JOINED TOGETHER"
		MARRIED - LIVES IN BISHOP FALLS.

Figure 5: A Medical Pedigree

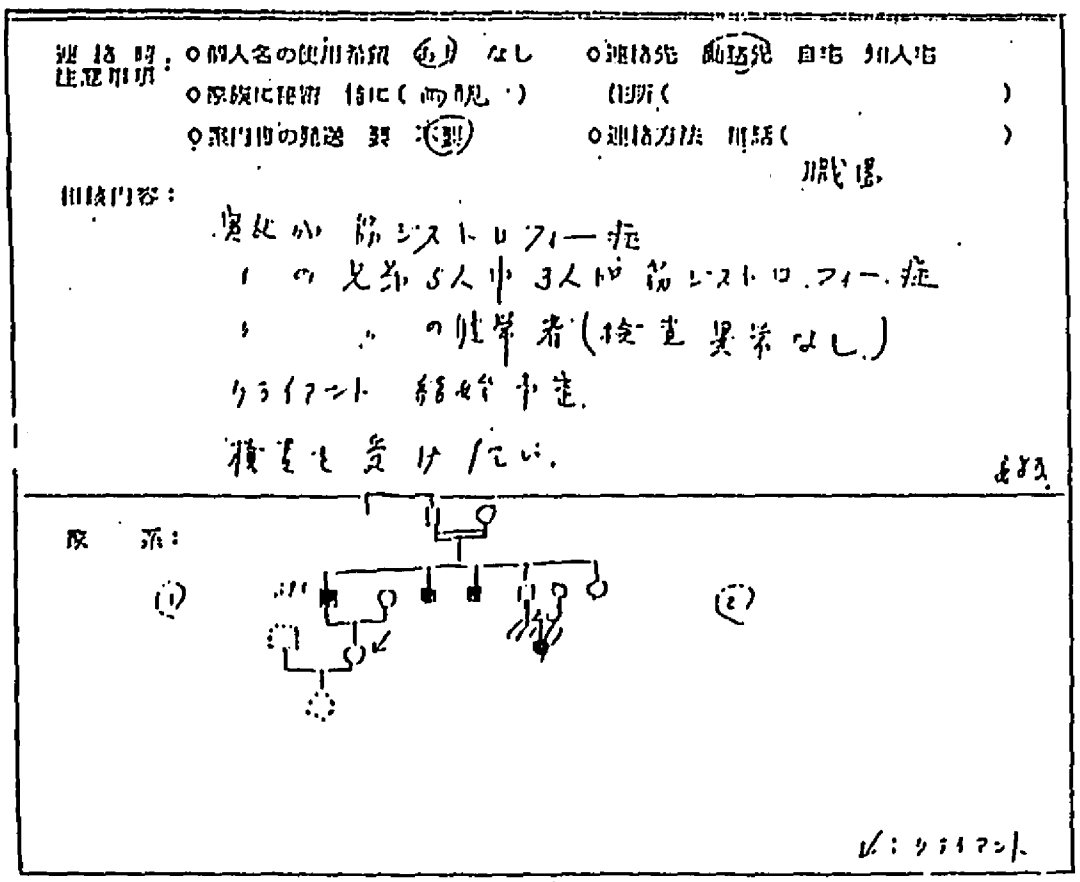


Figure 6: An Example of Secondary Transcription

出身地（本籍）

紹介機関　　同伴者

相談の要旨

家系図　いとこ婚 高齢妊婦.

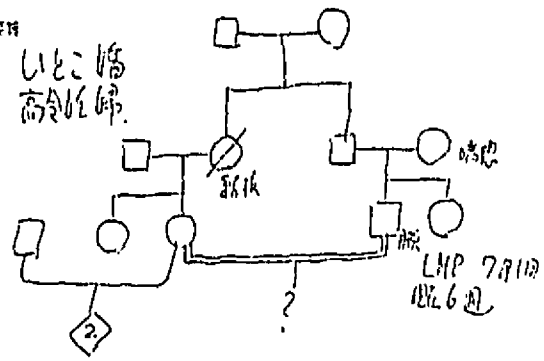


Table a

家系内疾患　無・有（近親婚・遺伝性疾患・先天奇形・染色体異常・重性肺疾・流死産・その他　）

医療情報（診断）

TELEFAX TRANSMITTAL FORM

TO: [redacted]

Name [redacted]　FAX No. [redacted]

Department　Phone No.

FROM:

Japan　Phone:
FAX:

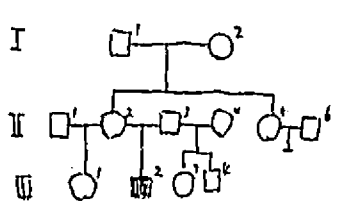
DATE: 1993. 2. 18

Number of Pages: 0 + COVER SHEET

TYPE OF DOCUMENT:

SHORT MESSAGE:

前略　　月　日の院内会議の際 おたずねしようと思っていたのですが、
月 日に退院されてしまいましたので FAX で失礼致します。



左図のような 血友病Aの家系の遺伝相談につき お教え下さい。 P.S%。
III-2 は 第8因子活性 %% 出血時間 2.5分
PT 11.0秒 APTT 150秒以上 の血友病Aの患者さんです。
II-2（母） と II-5（伯母） が保因者診断を希望されております。
III-2 以外に患者さんはいません。II-2, II-5 の第8因子活性を測定しておりません。どのような順序ですればよろしいでしょうか。また 保因者診断をお願いできますでしょうか。おつかましいお願いですがどうぞ宜しくお願い申し上げます。

[redacted]

Table b

Figure 7: Primary Transcription vs. Fax Version

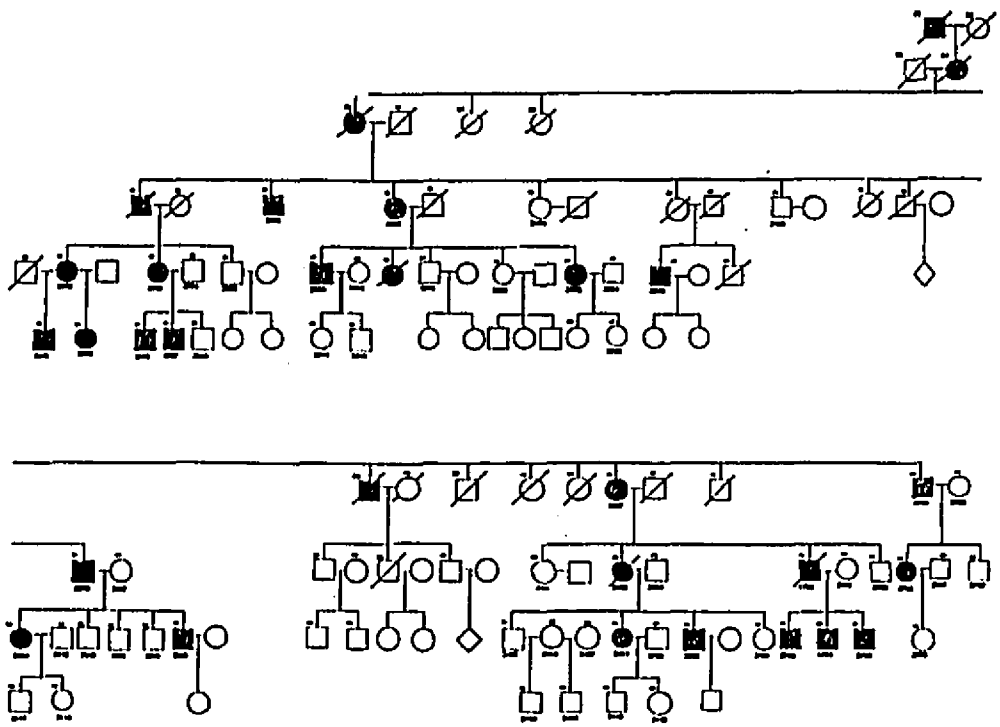


Figure 8: An Example of a Published Large Pedigree
(Source: Leppert, 1993)

PEDIGREES : Guidelines

CLINIC PATIENTS:

- Sign and date all pedigrees, and stamp with addressograph.
- Identify all family members who have condition for which patient is referred. Include first & last Names and clinic # if
  Include fourth degree relatives of the proband if possible.
- Identify anyone with birth defects, mental retardation, neurological signs, infertility, or anyone who is institutionalized.
- Identify anyone not living and indicate cause of death if possibe..

  Indicate all pregnancy losses, including: miscarriage, therapeutic abortion, stillbirth and neonatal death.
- Not necessary to identify Cancer, Arthritis, heart problems etc. if not related to reason for referral.
- Country of origin and ethnic origin for each branch of family at top of pedigree for each division of the family ie; born Ont.(Italian)
- May use diamond to summarize,if family members do not have noteworthy problems and are greater than first degree relatives.
- Should include names of couples and children, mother's maiden name if appropriate, any common family names, proband's name.
- Identify any family members that have had genetic counselling or prenatal diagnosis. Include first & last Names and clinic # if
- It is important that the geneticist be able to review the questionnaire and pedigree before the appointment.
- Any changes to the pedigree should be initialled.
-

Figure 9: An Example of Pedigree Guidelines

Using Koseki to link families

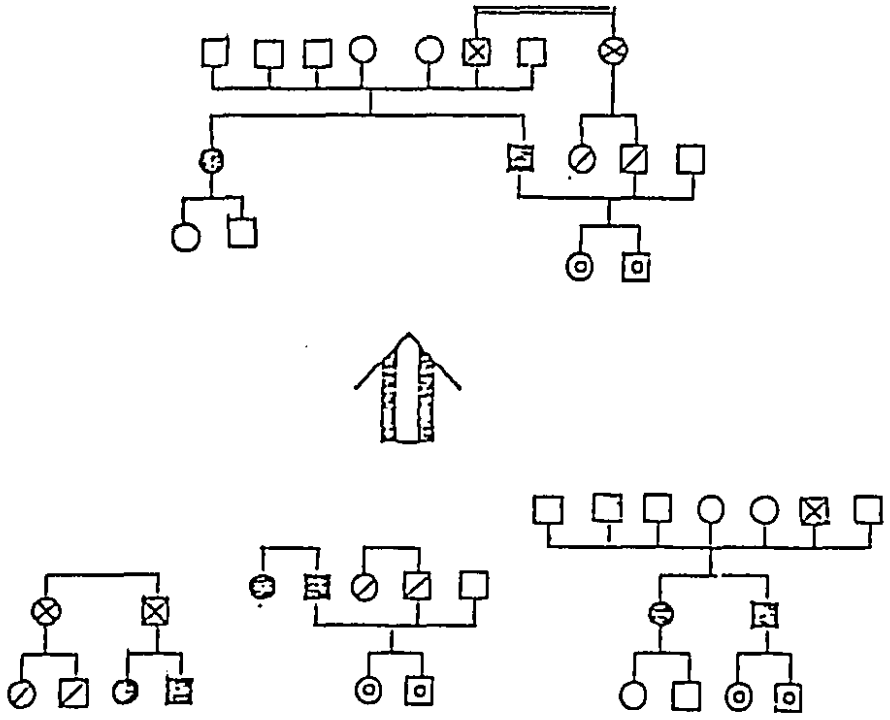


Figure 10: Reconstruction of a Large Pedigree
(Source: Fujiki, 1981)

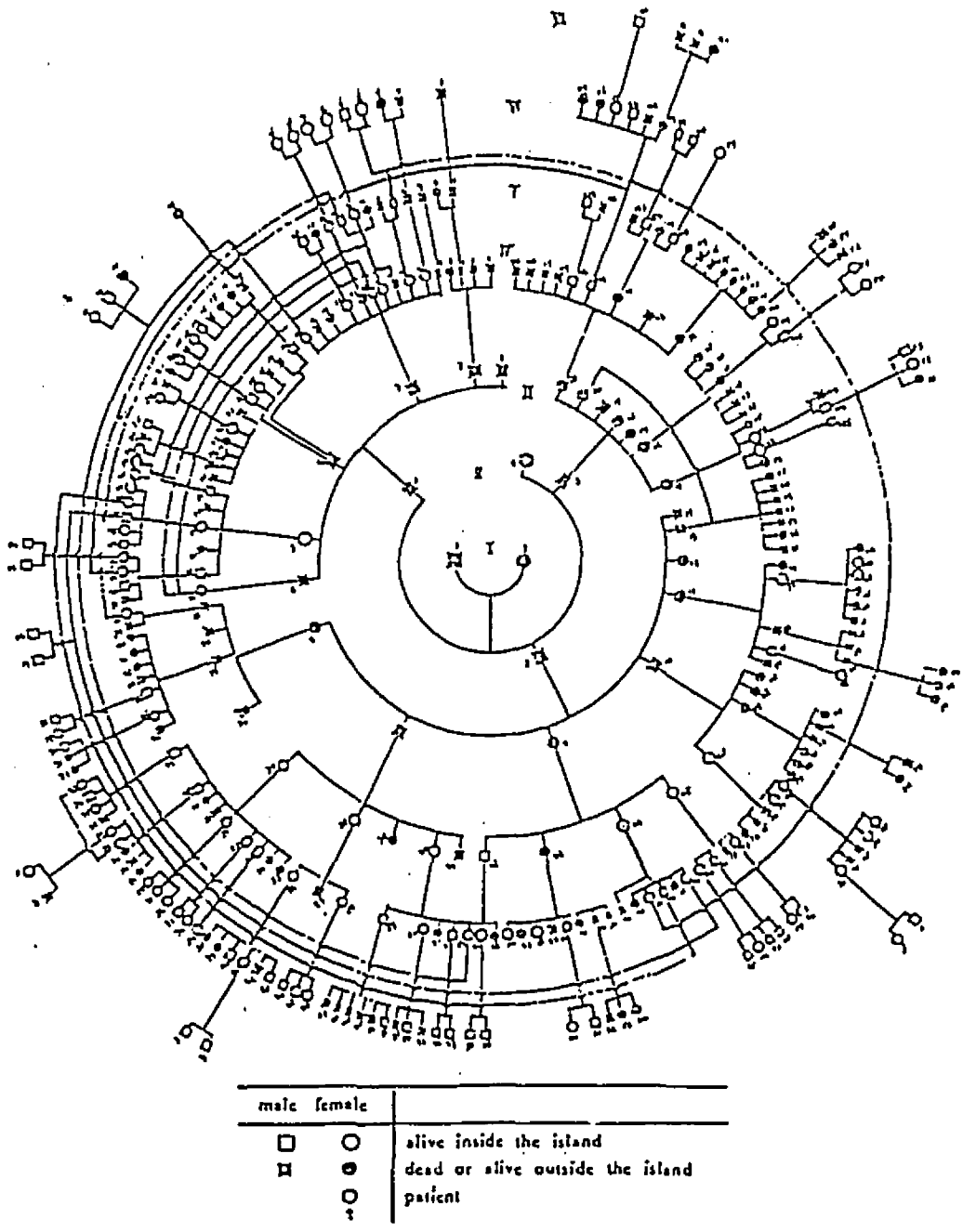


Figure 11: The Pedigree in a Village
(Source: Fujiki, 1981)

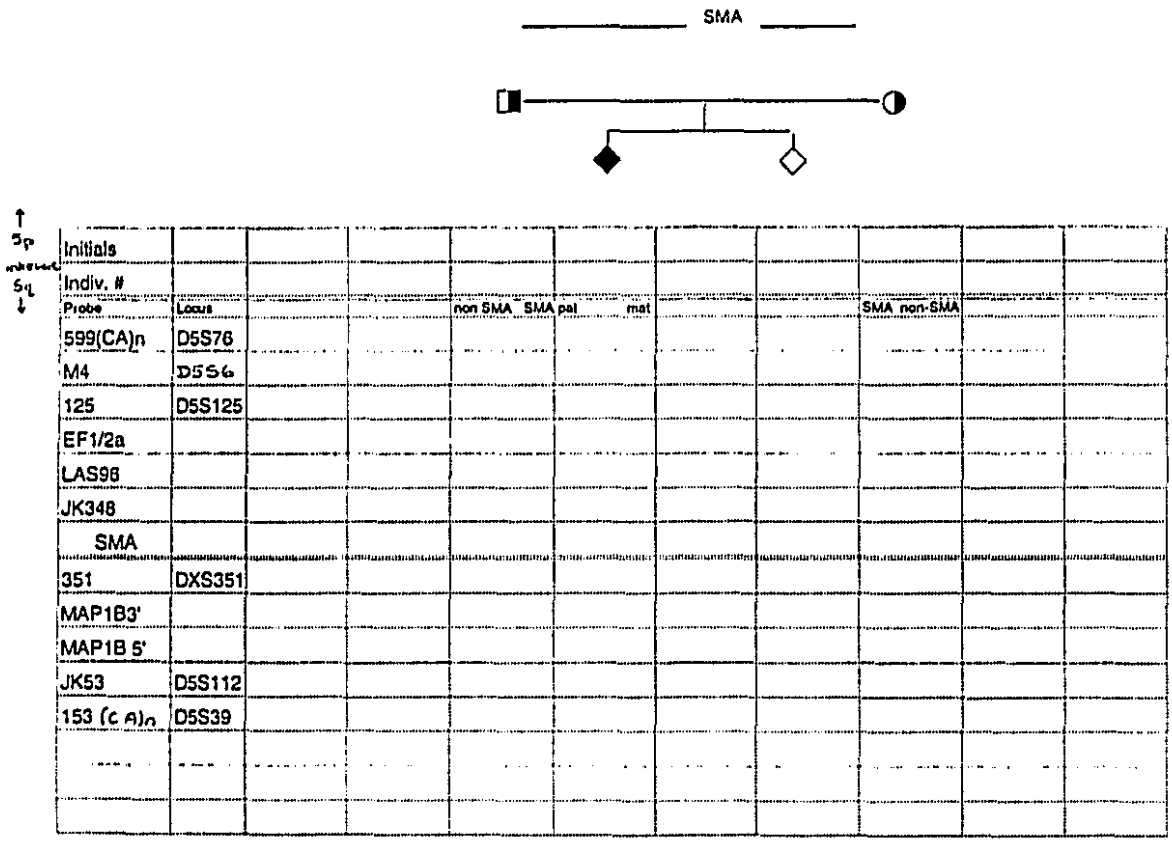


Figure 12: An Example of A Laboratory Pedigree

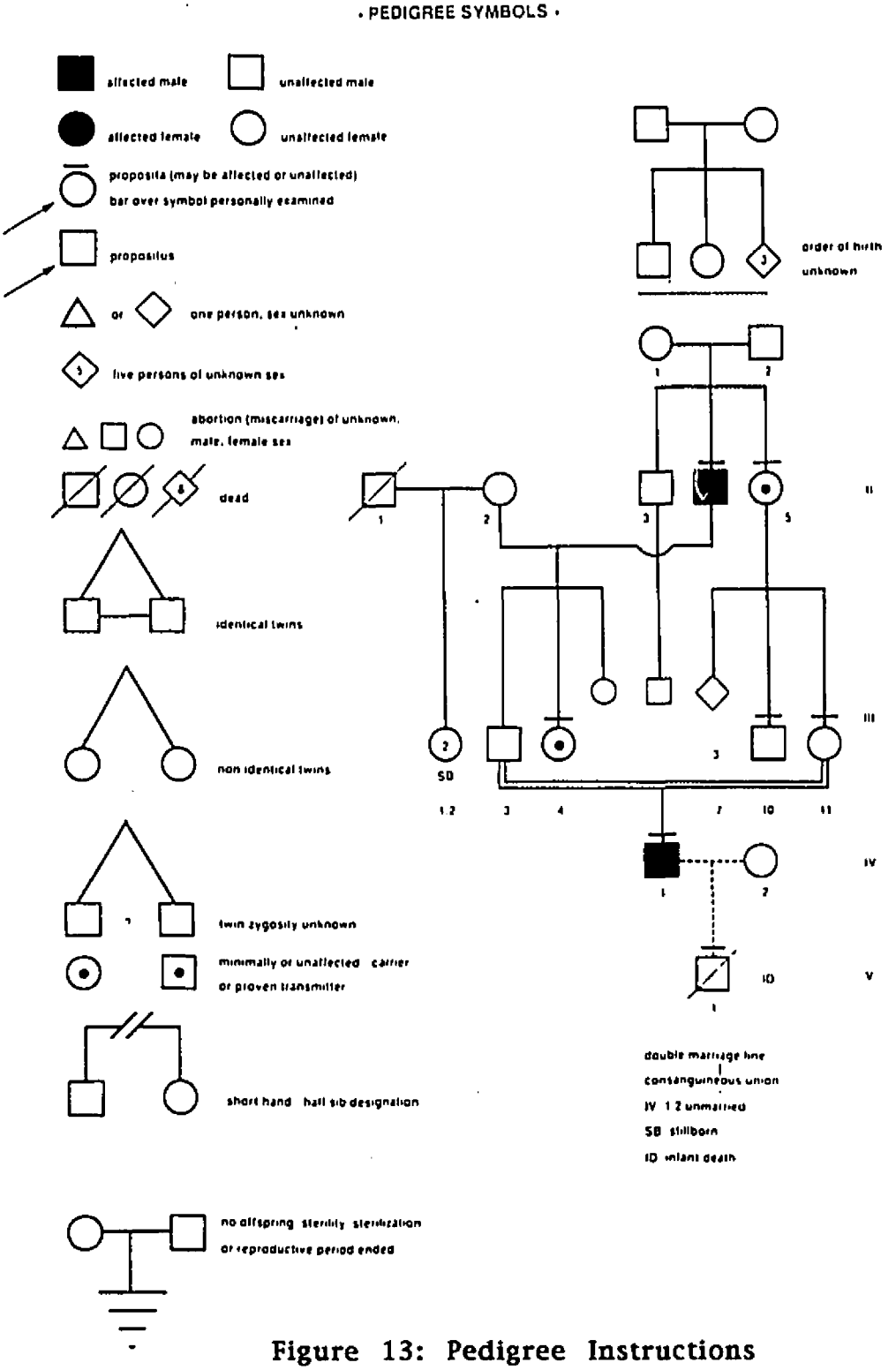


Figure 13: Pedigree Instructions

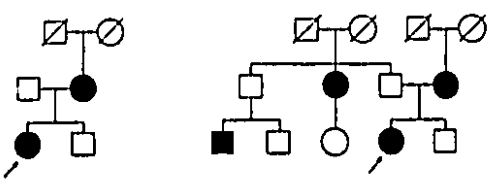


Figure 2.3 Polygenic inheritance simulating a mendelian pattern. Manic-depressive illness. (a) The superficial pedigree, with two generations affected, suggests dominant inheritance (autosomal or X-linked). (b) The recognition of affected individuals in both parental lines makes polygenic inheritance more likely than mendelian. (Pedigree details have been modified for illustration purposes)

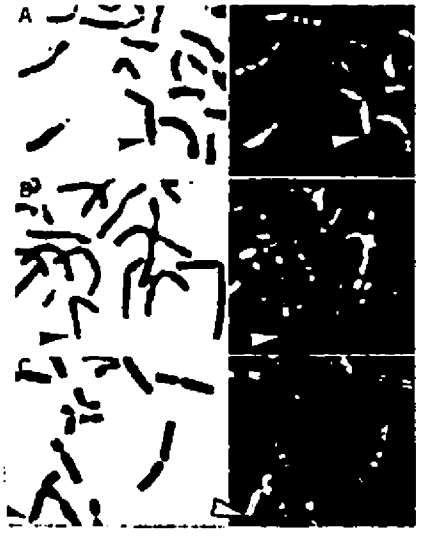
Figure 14: Pedigrees in a Medical Textbook
(Source: Harper, 1993)

脆弱X症候群
Fragile X syndrome

図1　症例I，30歳♂，長い顔，眉弓と下顎の突出，大きな耳介。

図2　巨大睾丸（症例I）
右はorchidometer
の最大球（20mℓ）
左睾丸 32mℓ
右睾丸 15mℓ

図3　脆弱X染色体（矢印）
脆弱部位はq28である。
縦A：症例I，B：症例II，C：保因者
横　左列，ギムザ染色
　　右列，キナクリン染色



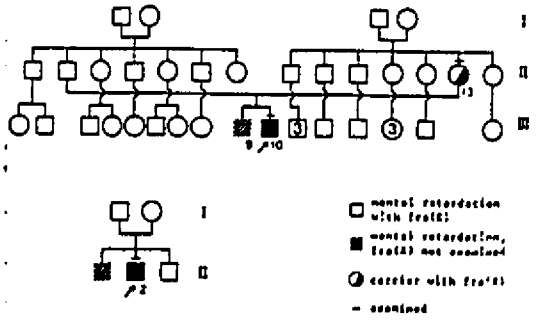


図4　家系図（上，症例I；下，症例II）

Figure 15: A Family History in a Medical Journal