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Genetics and the (social) body
Testing for the Cystic Fibrosis Gene

par
Wendy Hadd

Département de Sociologie

Faculté des arts et des sciences

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présenté par

Wendy HADD

a été évaluée par un jury composé des personnes suivantes:

Guy ROCHER	président du jury
Marc RENAUD	directeur de recherche
Bartha Maria KNOPPERS	membre du jury
Louise VANDELAC	examinateur externe

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Abstract

Being able to ascertain an individual's genetic status is a scientific possibility which has many personal and social ramifications. These outcomes must be explored from a social scientific point of view which incorporates a multidisciplinary grounding. This exploratory research uses Cystic Fibrosis as a case study for genetic screening and testing programs. An attempt is made to place scientific and technological innovations into social context. The focus is on if genetic screening for CF *should* be provided and how genetic screening *could* be provided. In addition, the moral ambiguities and ethical dilemmas engendered by this new technology are explored. Theories of social changes and Michel Foucault's theories of the body serve as a conceptual framework for understanding and enlightenment. The findings indicate that researchers are very aware of the ethical and moral questions to be resolved. In addition, the findings indicate some disagreement among researchers on several issues; namely, the necessity for population based genetic screening for CF and the value of screening children for CF.

Résumé

Au mois d'aout 1989, les scientistes ont découvert le gène de la Fibrose Kystique, une maladie héréditaire caractérisée par l'infection des poumons. Cette découverte a créé la possibilité de dépister la Fibrose Kystique (FK) sans tenir compte de l'histoire familiale des sujets et a amené avec elle des enjeux complexes. Présenté comme une procédure médicale routinière, en réalité le dépistage comporte de nombreuses dimensions sociales.

En tenant compte des autres technologies de reproduction et de la génétique, la capacité de discerner le code génétique des individus oblige à repenser la manière dont nous percevons le corps humain. Ces innovations changent non seulement l'individu et l'expérience de son corps mais changent également le corps social, c'est-à-dire toute la société.

Les scientifiques étant maintenant capables de déterminer le statut des porteurs de la FK pour n'importe quel individu, les questions quant à la manière dont notre société utiliserait cette information ont fait surface. Et tant que le sujet n'a pas été bien étudié, il est d'autant plus nécessaire de faire une étude exploratoire du sujet sous ses dimensions multidisciplinaires, sociologiques, déontologiques, médicales et légales, plutôt que de se concentrer à prouver une hypothèse très spécifique.

1. Revue de la littérature

La FK est une maladie des poumons qui affecte approximativement 3,000 individus au Canada. La région avec la plus haute concentration de porteurs de la FK est le Saguenay-Lac St-Jean au Québec. Il est estimé qu'un individu sur trente est porteur du gène de la FK sans en avoir nécessairement la maladie. Le traitement comprend une approche d'équipe clinique qui a comme but l'amélioration de la vie quotidienne et le prolongement de la vie moyenne des individus. La vie moyenne courante d'un nouveau-né avec la FK est entre 30 et 40 ans, par rapport à une vie moyenne de 5 ans pour un nouveau-né en 1960. Cette réelle différence est étroitement liée à la recherche, aux traitements et médicaments améliorés. Malgré tout, les enfants continuent de mourir à cause des complications de la FK.

Le gène de la FK sous 350 formes de mutation est présent dans la population, bien que moins de 20 se présentent fréquemment (Gilbert, 1994). Il est impossible d'affirmer avec une certitude absolue qu'un particulier *n'est pas* porteur du gène de la FK de par la quantité de mutations encore inconnues à ce jour. Ceci implique des questions concernant les notions de risque et la prise de décisions sur la reproduction. Quelques chercheurs ont suggéré que le dépistage ne dont pas être disponible tant et aussi longtemps qu'il y n'a pas de certitude à communiquer aux couples inquiets. Toutefois, en avril 1997, le NIH a recommandé que le dépistage soit offert aux couples désirant un enfant ou demandant des soins prénataux (NIH Consensus Statement, 1997).

Le point principal du désaccord est basé sur la méconnaissance du concept de risque génétique. Ceci sera aggravé par l'incertitude

révélée par un test négatif de la FK. Cette atmosphère d'incertitude plutôt que d'assurance peut être accablante pour les gens qui n'ont pas d'autres connaissances de la génétique. Michael Kaback, président élu de l'American Society of Human Genetics a peur qu'un dépistage peu concluant fasse plus de mal que de bien aux couples. Il croit bien que les résultats augmenteraient l'inquiétude et que quelques couples avorteraient des fœtus en bonne santé (Roberts, 1990). La recherche a démontré qu'entre 20 et 75% des gens qui reçoivent de la consultation génétique ne se souviennent pas, ou ne comprennent pas, l'information reçue. Néanmoins, il y a eu jusqu'à présent plusieurs projets pilotes afin de tester la distribution de services génétiques auprès de la population. Ces projets pilotes ont deux buts; s'informer de la meilleure méthode d'offrir des services génétiques; analyser la demande pour ces services et mesurer combien de gens décideront de les utiliser. En général, les répondants ont des attitudes positives envers le dépistage auprès des porteurs mais peu de gens veulent connaître leur propre statut vis à vis la FK.

Les services de santé offrent non seulement le dépistage mais aussi de l'information pour que toute personne puisse connaître son propre statut comme porteur si elle le désire. Il est très important de mesurer l'impact du dépistage pour l'individu et de réduire l'angoisse reliée au statut porteur de la FK. Peu d'individus démontrent des effets négatifs à long terme.

La dispensation des services de santé doit aussi inclure l'analyse des coûts du dépistage. Tandis qu'il est relativement facile de compter les coûts immédiats, les coûts moins tangibles sont aussi

très importants à considérer. Il est difficile de mesurer les coûts encourus par les individus et la société résultant des changements sociaux créés par le disponibilité des tests génétiques. Les coûts des services génétiques sont typiquement mis en comparaison avec les coûts des traitements, y compris les thérapies génétiques et la transplantation de poumons.

2. L'encadrement théorique

Les théories de la sociologie fournissent un encadrement essentiel à la thèse. Trois méthodes de travail sont utilisées. Les travaux de Michel Foucault sont indispensables si on veut comprendre les diverses façons dont le dépistage affecte le corps humain lui-même, et le corps social. Les gènes sont le fondement du corps et la capacité de déconstruire ce corps et de l'examiner est transformatif. En dépit du fait que le corps est constitué par l'ADN, chacun a sa propre perception du corps, du corps social et culturel.

Foucault écrit que nous créons le corps et qu'en même temps, nous le soumettons à un examen minutieux ; ce qui est très évident au sujet de la génétique. L'essentiel de notre corps est soumis à l'analyse et notre capacité de nous reproduire est exposée aux regards inquisiteurs des juges. En choisissant le dépistage, on choisit l'auto-surveillance de notre propre corps, une soumission volontaire au regard normalisateur. Donc il n'y a aucun besoin d'une contrainte extérieure. Quoique Foucault n'avait pas l'intention de questionner le corps engendré en entamant une discussion du corps malléable, il est manifeste que quelques méthodes d'instruction du corps sont plus appropriées soit aux hommes ou aux femmes. Étant donné que les

femmes sont traditionnellement plus aptes à l'auto-surveillance, il n'est pas étonnant que les services génétiques soient offerts plus souvent aux femmes.

Les théories des changements sociaux sont aussi nécessaires si on veut comprendre l'impact social de la génétique. Les théories du changement social démontrent qu'il y a un écart culturel entre les changements de la culture matérielle (les technologies) et la culture nonmatérielle (les attitudes). En analysant les enjeux sociaux des innovations médicales, Diana Dutton démontre que les nouvelles technologies ont un effet énorme sur le social. En plus, Dutton offre une analyse des effets négatifs de ces nouvelles technologies mises en place sans être mises à l'épreuve.

3. Méthodologie

Une méthode qualitative a été utilisée pour la compilation des données. Des entrevues ont été menées à Toronto, Ottawa et Montréal avec des experts de la génétique et de la FK.

4. Résultats

L'équipe qui travaille à la clinique pour enfants décrit la vie quotidienne d'une personne avec la FK. De plus, les répondants parlaient de la manière dont les gens prennent les décisions de reproduction lorsque qu'ils se savent porteurs du gène de la FK. Il convient de noter que si les parents ont déjà eu un enfant avec la FK, ils sont peu en faveur de l'avortement d'un fœtus subséquent avec la FK; d'autres membres de la famille seront plus favorables à l'avortement. Comment le public répondra est maintenant inconnu

mais la recherche montre qu'il y a moins de gens qui désirent savoir s'ils sont porteurs du gène de la FK qui l'inverse.

Un des enjeux les plus importants qui soit ressorti des données est le façon dont la FK sera présentée aux citoyens. La manière donc on répondait à cette question est significatif et il y a deux possibilités tout à fait différentes.

En fait, la FK est perçue sous deux formes, selon son degré de dangerosité. Pour certains individus, la FK est une maladie bénigne qui peut être soignée, mais pour d'autres la FK est une maladie sérieuse qui entraîne la mort dès le jeune âge. Les répondants dans cette recherche savent bien que le pouvoir de limiter le discours revient aux gens capables de décrire la maladie àl'intention du public. Les répondants intéressés au dépistage disent que la FK est une horreur et que tout le monde devrait faire ce qu'il peut pour la prévenir dès naissance d'un enfant. Ces répondants pensent qu'il est fort nécessaire d'établir des programmes de dépistage et d'éducation pour montrer aux gens le besoin d'utiliser de tels programmes. D'autres répondants croient que le statu quo fonctionne bien ; présentement, des gens avec une histoire familiale de la FK se présentent pour être examinés. Ces répondants pensent qu'il s'agit de la meilleure utilisation des fonds médicaux.

Malgré le désir exprimé par certains des répondants pour un système de dépistage de la FK sur le modèle de Tay Sachs, il n'y a aucune demande pour le dépistage de la FK hors des familles avec une histoire familiale à risque. Même ces répondants ont convenu que si on décide d'offrir le dépistage à la population, une sensibilisation dans les médias serait indispensable pour démontrer

aux gens que les programmes existent et qu'il sera avantageux d'en profiter afin que chacun connaisse son statut. Dans l'état actuel des faits, on constate que la plupart des gens au Canada n'ont aucune expérience de la FK. Que trois milles personnes soit atteintes de cette maladie ne parais pas important dans un pays de vingt-six millions. Alors cette recherche montre que le mouvement vers le dépistage est plus un "supply push" qu'un "demand pull".

Il y avait un désaccord marqué au sujet du dépistage pour enfants. Quelques répondants sont d'avis que les enfants peuvent avoir un test génétique si leurs parents le demandent. D'autres répondants pensent plutôt que le dépistage sur les enfants présente des difficultés morales de par le fait que les enfants ne sont pas en mesure de prendre de telles décisions sur la reproduction alors qu'une fois adultes ils amaient préféré ne pas connaître leur statut de porteurs. Ce désaccord est un enjeu décisif car il souligne clairement les difficultés que pose le dépistage sur demande.

Il est évident que les parents qui demandent le dépistage sur leurs enfants ne comprennent pas que cette connaissance ne peut être utilisée par les enfants que s'ils décideront à un moment dans le futur d'avoir leurs propres enfants. Connaître que son enfant est porteur de la FK n'avance à rien, ce n'est pas de l'information qu'on peut utiliser. En plus, décider de passer des tests génétiques est une décision personnelle et individuelle que seuls les adultes peuvent prendre pour eux-mêmes après avoir consulté un conseiller en génétique. Les parents qui font ces demandes agissent comme des propriétaires de leurs enfants et pensent faire ce qu'ils veulent. De plus, les chercheurs et médecins qui ne préviennent pas ce type de

décisions agissent de manière irréfléchie car ils sont prêts à accomplir des tests médicaux hors de tout contexte et sans aucun besoin et avantage pour les enfants. C'est un bon exemple de la surveillance que Foucault décrit ; la surveillance qui a pour but l'exercice du pouvoir, en ce cas le pouvoir des parents sur leurs enfants. Plusieurs répondants ont exprimé l'inquiétude que si un enfant dans la famille est porteur de la FK, le comportement des parents envers cet enfant ne sera pas le même que le comportement envers les autres enfants "non-porteurs". C'est réellement un enjeu qui demande davantage de recherches avant qu'aucune décision protocolaire ne soit prise.

Les questions concernant les coûts occupaient la pensée des répondants. Néanmoins, comme anticipé, il y avait un désaccord concernant la meilleure façon de dépenser les fonds. Quelques répondants pensaient que les gens qui refusent l'avortement du fœtus porteur du FK ne devraient pas avoir le droit aux tests génétiques parce qu'ils dépenseraient doublement l'argent tandis que d'autres étaient d'avis que le rôle des tests est de fournir de l'information et que c'est aux individus de choisir quoi faire avec les informations qu'on leur donne.

Des considérations éthiques étaient au premier plan pour la plupart des répondants. Presque tous ont exprimé un intérêt en matière d'éthique et de morale soulevée par le dépistage génétique. Le délai culturel était compris comme plein d'importance pour société. Par ailleurs, beaucoup de répondants ont exprimé une inquiétude face à la vitesse de l'innovation technologique et son impact sur la société. Ils désirent davantage de recherche en

sciences humaines qui puissent donner une direction aux innovations médicales.

La prèsente recherche n'est qu'un petit pas vers une compréhension contextuelle de cet enjeu, divers et important.

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Introduction

Genetics are an integral part of human life but until recently our genes have been hidden and remote. With the revolution in genetic research, our genes have become accessible: they can be coded, they can be 'tested', they can be manipulated. These abilities change the way we see and understand the human body.

The original impetus for this research was the discovery of the gene for Cystic Fibrosis on chromosome number 7 in August 1989. The sociological implications of this discovery are myriad and there has been a consequent plethora of interrogation as our society attempts to understand the social changes and personal changes wrought by new knowledge. Many of our genes have been mapped, and continue to be mapped each and every day, as part of the Human Genome Project (HGP). HGP intends to fully map the entire human genome and to provide, in a sense, a geographical map for human beings. This information will not just change our knowledge of the chromosomal structure of the human body but will change our very perception of our bodies and the meanings that the body holds for us.

The title of this thesis seeks to invoke the double meaning of genes and the body. On the one hand, it is clear that genetics is linked to the physical aspect of the individual body in that our bodies are each an individual outcome of a particular genetic mixture. On the other hand, it is equally true that the *social* body

will also be affected by genetics. That is to say, society as a whole must react to the knowledge generated by research into genetics. As Michel Foucault points out, there can be no conception of the individual body outside of a social conception because our bodies are constituted by and through culture. This thesis examines changes wrought by genetics on the individual body (the person) and the social body (society) using the specific case of the CF gene.

It is very important that social research examine changes brought about through scientific and technological innovations. It is not self evident that technological breakthroughs are always positive for human beings. As one respondent states, information has a price and we can extend that to state that knowledge has a price as well. We know more about our genes now than ever before but we also need to discover what we want to do with that knowledge and what price will be paid to attain this knowledge.

The location of the CF gene opened up the possibility for every person to be tested for carrier status of CF. Nevertheless, the subsequent discovery that many, perhaps hundreds, of mutations of the CF gene are present in the population mitigated the original excitement and anticipation of the discovery. The original plan was to have carrier screening programs which would allow every individual to know his or her genetic status for CF. However, time has shown that carrier screening for deleterious genes is much more complicated that the simple provision of information. Countless factors play a role in understanding how individuals and society will react to genetic knowledge. Understanding the most important of these factors is the ultimate goal of this dissertation.

Chapter 1 reviews the relevant literature concerning CF. This literature has several sub-sections. These sections are introduced in chapter 1 and integrated throughout the dissertation. The sections are as follows: 1. living with CF, 2. the social meaning of genetic testing and screening and 3. the provision of health care services.

The epistemological framework that unifies this dissertation is presented in Chapter 2. Here the multidisciplinary nature of the research is evident and three separate, yet linked, theoretical perspectives are presented to 'make sense' of the data.

Medical innovation as a social issue provides a background to the nature of the research and incorporates some common research. Sociologists recognize that the field of medicine is in many aspects a profoundly social field. Medicine may find it's cures and potions in research from the hard sciences, however, medical treatment is acted upon the physical and social body. Medical innovations may change how people perceive illness or health; they may change the way people live and how people perceive their own lives. Diana Dutton has written an important work on this topic which provides an indepth analysis of micro and macro ramifications of new medical techniques and medications. This serves to validate the underlying premises of this dissertation and to enhance the exploration of social change.

While often viewed as dichotomous and unrelated, both conflict and functionalist theory enlighten the research regarding the meaning and impact of social change. Sociologists recognize that technological change is quicker than is attitudinal change. Change can be seen as both detrimental and beneficial to society, depending

upon the perspective of the observer. Conflict and functionalist theoriests both posit that social change is inevitable.

Finally, the theory of Michel Foucault and his work on the body broadens the perspective and demonstrates the interior affect of social events on the individual and on the social body. Foucault is important to this research because it does not simply concern something that can be done to the body (i.e. check one's genes) but more importantly it concerns the ways in which the body is perceived by the individual and by society. How does one think about one's physical body when one knows deleterious genes are present and can be passed on to offspring? What does this mean for the individual and what meaning can it have for society in general; particularly a society that devalues the "non perfect" human being? Stemming from a philosophical tradition of mind/body dichotmization which enacts a separation of the physical from the spiritual body Foucault's work enables us to find meaning in these queries *because* he refuses to accept this dichotomy. We are our physical bodies and we must make meaning for ourselves and for others as physically and culturally constituted bodies.

Chapter 3 discusses the methodological criteria for the research. The qualitative method of data collection is discussed along with the rationale for using a case study method. The respondents, the interviews and the sampling method are presented herein. A non-random sample was used which prohibits generalizing from the data. However, given that the research was designed as an exploratory study, this is not an unanticipated drawback. Respondents are considered as "experts" in the field of CF; scientists,

nurses, laboratory personel were recruited through a snowball technique.

In addition to the interviews, archival research is presented as a secondary data collection method.

The archival method was used to gather information presented in Chapter 4. This chapter provides a historical perspective on genetic screening with a focal point on Tay Sachs and Sickle Cell Anemia. Both of these genetically transmissible disorders have previously been the focus of population based carrier screening programs. As such, the experience garnered from them has much to add to an understanding toward such programs for CF.

One of the most important things to remember is that our knowledge about the genetics of CF occurs within a particular social and scientific context. If we were only able to test for the CF gene, the impact may be minimal. However, when we add this to the already rapidly growing number of other abilities and knowing, the impact is more expansive. All around us there are fundamental changes in the area of genetic research happening in a context of profound changes in reproduction. Not only is the knowledge and meaning of the body changing but the essence of the way we reproduce our bodies is undergoing substantial change as well. The accumulated result is pushing the framework of our moral and ethical boundaries. To understand this necessary and broad context, Chapter 5 presents a brief contextual analysis of a wide spectrum of research along with some attempts by the Canadian government to come to grips with the social implications of science.

Chapter 6 is a presentation and discussion of the data. Given that the research was designed as an exploratory study, this data is incredibly extensive. The data touches on the practical such as how to train genetic counselors or how much it would cost to process a genetic test, to the more abstract and philosophical issues such as how do we as a society choose a moral agenda. The three categories established in the review of the literature are also present here: living with CF, the social meaning of testing and screening, health care services.

Data indicates that there is no one experience of living with CF. The daily care and the life experience for the affected child and the family of the child depend heavily upon the extent of CF. CF itself can be seen as different disorders because one person may be mildly affected while another can be severely dibilitated by CF. The variablity of CF is the source of much of the disagreement among respondents regarding the meaning of being a carrier.

These different meanings of living with CF are carried into the social aspects of CF. There is some interesting data which shows that attitudes toward carrier testing and screening for CF can be manipulated by information given or withheld from individuals. As well, the way in which individuals understand and articulate risk factors depend upon the way in which information has been presented which again is linked to the perception of CF as either a mild or a serious disorder. Attitudes toward abortion and willingness to abort depend heavily on factors that are difficult to discern and involve hard ethical choices that an ethicist interviewed feels may be outside the individuals' realm of capacity.

With regards to health care provision, issues centre on if and how our medicare system could or should respond to genetic testing. Respondents disagreed on whether genetic screening programs were necessary or desirable. While economic considerations were not a focal point of the research, they did arise as an outcome of interviews.

The depth and range of data presented in Chapter Six are a reflection of the incredible reach of genetic discovery, reminding us that the location of the gene was just the beginning of intense social changes.

The conclusion of the research forms Chapter 7. Included in this chapter are the limitations of the research and the implications for future research. This research is but one stepping stone on the way to further knowledge. In the future, more research is needed to discern the social impact of genetic discovery on the individual and on society in general.

Chapter 1

Review of Literature

Cystic fibrosis provides an excellent opportunity to examine the issues of genetic testing for several reasons: it is a common genetic disorder, only recently has the possibility of screening been made available, and such screening has become a practical reality on an exploratory or research basis in many communities. Because the area of reproductive technologies is so vast, encompassing and incorporating many different views and themes, narrowing the field to include only Cystic Fibrosis allows for a more nuanced understanding. Cystic Fibrosis is a disease which allows for an analysis of one particular reproductive technology. By more fully understanding the ethical, legal, social and medical issues in this one case, we can more fully understand the broader social context of which it is an important part.

Not only must Cystic Fibrosis be understood in its clinical manifestation but it must also be acknowledged as an example of how society constructs health and illness, how diseases are given priority within our health care system, the influence of media reporting in the medical field, and what meaning genetic testing has on a literal and on a symbolic level for the average lay person. Consequently what may seem at first to be a relatively simply issue is actually incredibly complex. It is this very complexity that makes Cystic Fibrosis such an exciting and important field of inquiry.

This chapter is a review of relevant literature. As such it places the subject matter into the context of other research and helps to situate the general categories to be explored. The review of literature is organized around three broad categories to help the reader by providing a framework within which to situate such a wealth of information. These three categories are 1. living with Cystic Fibrosis; 2. social meaning of genetic testing and screening; 3. health care services.

1.1 Living with Cystic Fibrosis

Cystic fibrosis was recognized as a disease in 1938 when Dorothy Anderson, a pathologist at the Babies' Hospital in New York gave the first detailed pathological description of it. Cystic fibrosis is "a progressive obstructive suppurative lung disease together with malabsorption due mainly to an insufficiency of pancreatic digestive enzyme secretion" (Goodchild, 1975). The name Cystic Fibrosis (CF) is a descriptive term in that it describes the changes that occur in the pancreas of affected individuals. The part of the pancreas that produces the proteins that digest food (digestive enzymes) is replaced by a fibrous scar tissue with fluid-filled cysts. The respiratory tract is also infected by a thick, sticky mucous that obstructs the airways. The presence of this mucous causes subsequent infections in the chest (Harris and Super, 1991)· These usually frequent chest infections are very damaging; over 90% of CF patients die from pulmonary insufficiency. In addition to the involvement of the pancreas and the lungs, five to 10% of newborns with CF are born with a type of intestinal obstruction called

meconium ileus, and 2 to 5% develop liver disease at some point in the course of the illness. As the life span for individuals with CF lengthens from about 5 years in the 1960s (Goodchild, 1975) to between twenty to thirty years in the 1990s, it has now been found that males with CF are infertile and females with CF are more likely to be infertile than are unaffected females (Collins, 1992).

Because the severity of CF varies from individual to individual, treatment is based on each persons' needs and is modified throughout the life span. In general, it has been found that treatment in a recognized Cystic Fibrosis clinic is preferable to treatment by individual family doctor in a private practice (Capewell, 1986). Treatment in a clinic allows a team of experts to pool their knowledge and to act together to decide effective treatments based on knowledge of many other cases. A general practitioner does not have this expertise because they will treat few patients with CF. In Denmark the establishment of a CF centre in Copenhagen resulted in a doubling of survival of treated patients confirming the belief that a separate CF clinic is preferable (Capewell, 1986).

While Cystic Fibrosis is commonly seen as a child's disease, improvements in treatment means that there are more and more adults with CF. Today approximately 32% of people with CF are over 18 years old.("You Were Asking", 1991) whereas thirty or forty years ago, an individual with Cystic Fibrosis would be certain to die in early childhood. In 1992, the median life span was 29 years. In addition, an individual born today with Cystic Fibrosis would be expected to survive 40 years based on the treatments now available ("You Were Asking", 1991).

1.1 (1) Genetics of Cystic Fibrosis

Cystic fibrosis is an autosomal recessive inherited genetic disorder. That means that an individual must inherit identical copies (alleles) of the CF gene from each of his/her parents in order to have the disease. Such an individual is said to be homozygous for that gene. If the individual inherits one copy of the CF gene from one parent and a copy of a normal gene from the other parent (non-identical alleles), s\he is heterozygous for that gene and is said to be a carrier of the CF gene. A carrier does not have the disease but if they have children with another carrier, they incur a risk of having a child with the disease.

FIGURE 1

Inheritance of the CF gene from two carrier parents

(CF= CF gene, N= normal gene)

	Mother	Father		
Parental Genotype	CF N	CF N		
Possible Genotypes of children	CFN NCF	CFCF NN		
Disease Status	CARRIERS	CYSTIC	NON-CARRIER	
Incidence	1/2 (50%)	1/4 (25%)	1/4 (25%)	

As this figure illustrates, two carriers have a one in four (or 25%) chance of having a child with Cystic Fibrosis, a two in four (or 50%) chance of having a child who is a carrier of the gene and a one in four (or 25%) chance of having a child who is neither affected with the disease nor a carrier of the gene.

Cystic Fibrosis is most common in the white (Caucasian) population; both in carrier frequency and in incidence rate as Table 1 indicates.

TABLE 1

Incidence and carrier frequency of CF (in select populations)

Population	Carrier frequency	Incidence
White	1/29	1/3,300
Black	1/65	1/15,300
Asian	1/90	1/32,100

Source: NIH Concens Statement, 1997.

The carrier frequency for CF in whites is approximately 1 in 29. This means that one of every twenty-nine white people have one copy of the CF gene which is about 4 percent of the total white population. In the North American black population, carrier frequency is one in sixty-five and in the North American Asian population it is one in ninety. Given that carrier frequency is much lower in these two latter populations, incidence of the disease is also much lower; the incidence of CF in the white population is 1/3,300, in the black population it is 1/15,300 and in Asian population it is 1/32.100. Interestingly, the incidence of CF in *Native* Africans and *Native* Asians is much lower, 1/50,000, suggesting that North American blacks and Asians have an increased incidence due to interracial reproductive patterns (NIH Consens Statement, 1997).

1.1(2) Identifying the CF Gene

The Cystic Fibrosis gene was identified in August of 1989 by a team of researchers headed by Lap-Chee Tsui of the University of Toronto and the city's Hospital for Sick Children, and Francis Collins of the University of Michigan. These two researchers isolated the gene on a region of chromosome 7 following what has been described as an intensive and frantic five year search. Laboratories worldwide were involved in the search for the CF gene; each hoping to be the first to make the momentous discovery. At stake was the enormous prestige of being first as well as unspecified financial rewards. Thus while the gene was isolated on 24 August 1989, Tsui and Collins hoped to postpone the announcement until the 8 September edition of Science because they did not want to jeopardize a patent application covering the gene sequence and its future applications (Davies, 1989).

1.2 Social meaning of genetic testing and screening

It is necessary to establish the difference between carrier *testing* and carrier *screening*. Carrier testing refers to the analysis of a particular individual's genetic status. It is conducted on a case by case basis, following genetic counseling. Testing is available to people with a family history of CF and, increasingly, as a part of preconceptual or prenatal diagnostic care for pregnant women. Screening refers to programs which would have as their goal to test the entire population for CF carrier status. Population based carrier screening is not yet a practice but is in an exploratory stage for many researchers.

1.2 (1) Cystic Fibrosis Mutations

Cystic Fibrosis is not one disease with one outcome as are diseases like Tay-Sachs or Lesch Nyman disease. Rather, CF can have very different outcomes for different individuals. Screening for CF involves not just the search for one genetic mutation but several, in fact, to date more than 350 different mutations of the CF gene have been found, although less than 20 occur with any frequency (Gilbert, 1994)· The implications of this are profound and far reaching with respects to population screening programs. The meaning of different mutations is beginning to be understood. A series of experiments conducted by US and UK researchers have discovered that individuals who are *carriers* of AF508 may be less susceptible to typhoid fever than people with other mutations (Bradbury, 1998). Hence, there may be positive benefits bestowed by the CF gene.

In general, a population screening program is expected to yield certain results. The impetus for an individual or a couple to be screened is to allay the fear of the unknown. However, the high number of mutations confound the search for absolute results. Additionally, there may be mutations which have yet to be discovered. Thus a couple presenting themselves for screening can find out for certain if they are carriers but can never be assured that they are *not* carriers. The implications for reproductive decision making in this situation are still relatively speculative.

The Cystic Fibrosis Genetic Analysis Consortium (1990) has compiled a frequency distribution table of AF508 worldwide based on 84 different studies.

TABLE 2

Frequency Distribution of AF508 In Selected Populations

FREQUENCY (%)	POPULATION	SOURCE
82	Northwest England	Super & Schwarz 1992
81	Northwest Brittany	Guillermit et al 1990
80	Belgium	Wauters et al 1991
75	France	Vidaud et al 1990
72.2	France	Simon-Bouy et al 1991
71	Urban Quebec	Rozen et al 1990
70	Louisiana	Rozen et al 1990
67	S.-W. Germany	Lindner et al 1992
64	Southern France	Claustres et al 1990
60	Norway	Eiklid et al 1993
58	Sag-LacStJ	Rozen et al 1992
58	Northern Italy	Restagno et al 1992
54	Poland	Witt et al 1993
47	Brazil	Raskin et al 1993
30	Israel	Shoshani et al 1992
0	Native Americans	Grebe et al 1992

Frequency of AF508 range from a low of 26% in Slovenia (formerly Northern Yugoslavia) to a high of 100% in North Africa. The overall average frequency worldwide of the AF508 mutation is 68%. Cystic Fibrosis has also been detected in populations previously considered to be unaffected by the disorder. This generally is the case for non-Caucasians, for example Native Americans in the southwest (Grebe et al. 1992) and Mexicans (Salcedo et al. 1993). Although the frequency of CF and its mutations are as yet unknown in Mexico, one postmortem study has revealed a child homozygous for the AF508 mutation. The authors hypothesize that adverse socioeconomic and environmental conditions may be a factor in the low frequency of CF cases diagnosed in living children; essentially they are suggesting

that the children with CF do not live long enough to be diagnosed (Salcedo et al. 1993).

These figures indicate that a worldwide statistical average of 68% of people who are carriers of CF carry the ◊F508 mutation. It also means that 32% of people who test *negative* for that mutation actually are carriers; they are carriers of another mutation. Therefore, if the test was only done for this one dominant mutation there would be a 32% false negative rate i.e. out of every 100 people who really are carriers, 32 people would be told they were not. The range of 40 to 88% given is an indication of the differences between different groups of people with regards to the dominant mutation. With a low 40% carrier rate, the hypothetical false negative would be 60% while with a high 88% carrier rate, the false negative rate would only be 12%. Given that many mutations are detectable, screening is able to raise the detection rate much higher than it would be if only one mutation were known. Still, the detection rate continues to vary between different peoples. And the number of mutations appears to be unpredictable as some mutations have been reported as single cases (Cutting et al. 1992) .

TABLE 3

Combined Frequency Rates for Select CF Mutations

TOTAL FREQUENCY	POPULATION	SOURCE
92 (5)*	Ashkenazi Jews	Shoshani et al 1992
91.5 (11)	Northwest England	Super an Schwarz 1992
89 (3)	Sag-LacStJ	Rozen et al 1992
85 (3)	Urban Quebec	Rozen et al 1992
80 (9)	France	Simon-Bouy et al 1991
79 (12)	S.-W. Germany	Lindner et al 1992
73 (30)	Southern France	Claustres et al 1992
70 (7)	Poland	Witt et al 1993
62.6 (3)	Norway	Eiklid et al 1993
8 (6)	Native Americans	Grebe et al 1992

*Numbers in brackets denote the number of mutations tested to achieve the total frequency. For example, 92 (5) means that when five mutations are tested for, 92% of carriers will be detected.

The Saguenay-Lac St. Jean region of Quebec has the highest reported rate of Cystic Fibrosis in the world, with a birth incidence of 1 in 891 births and a carrier frequency of 1 in 15 (Rozen et al., 1992). This high frequency has been contributed to the founder effect in a relatively remote community. In the S-LSJ area only 55% of CF families carry the dominant mutation. However with the additional testing for two other mutations, A455E and 621 + 1G -> T, 90% of CF chromosomes were accounted for and with the additional testing of five other mutations a detection rate of 93% was achieved (Rozen et al., 1992).

Similar results have been achieved with other groups of people. Whereas the ◊F508 accounts for only 23% of Ashkenazi Jews with CF, 60% have been found to have the W1282X mutation (Shoshani et al., 1992). And with the additional testing of three more mutations, a

92% detection rate has been achieved. The W1282X mutation is very rare in other people, occurring at a rate of only 1.6% (Shoshani et al., 1992).

In countries where the population is quite homogenous, high detection rates have been consistently achieved with only a few mutations. In the UK. and Denmark, for example, between 85-90% of CF chromosomes are identifiable. One study in Northwest England yielded a false negative rate of only 1.19% (Super and Schwarz, 1992) In other countries, however, a continually learning and discovery process has been going on. While it was originally believed that the disease was limited to Caucasians and therefore any population screening would also be limited to Caucasians, it has now been discovered that CF has appeared in Native Americans and in the American Black population, albeit at a lessor incidence level, and with six of the nine mutations found in American Blacks unique to that population (Grebe et al., 1992).

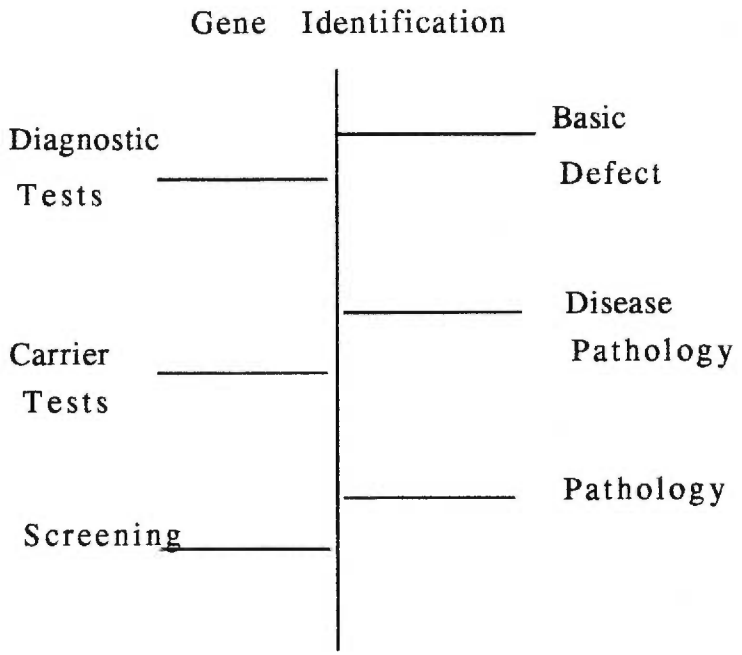
Without ongoing carrier screening programs, it is impossible to enumerate the different mutations existing in the population. To date, this enumeration has been confined to families with an identified proband leading some scientists to speculate that genotype/phenotype links are being given short shrift, inasmuch as many mild mutations may be missed by familial testing (Hamdi et al., 1993). Therefore, it is suspected that some individuals who carry two copies of the CF gene are not picked up as such because their expression of the disorder is so mild. It is not unheard of for people to be diagnosed as suffering from asthma as a child, only to be

identified in late childhood or early adulthood as having CF (Simon-Bouy et al, 1991).

1.2 (2) Genotype/phenotype linkage

The link of genotype to phenotype is an area of particular concern to physicians who work with CF patients. Genotype refers to a person's genes; in the case of CF genotype refers to the particular mutation that an individual carries. Phenotype is the characteristics of the disease as it is expressed by each individual. CF has a variable expression and the inability of current genetic screening to pinpoint accurately to what extent an individual will be characterized by any particular characteristic of the disorder adds an additional unknown to people making reproductive decisions. This void in knowledge needs to be filled.

Without a genotype/phenotype link scientist lack the ability to predict an individual's expression of the disorder. There may be pancreatic involvement, liver disease, links to diabetes or sterility, the individual with CF may be seriously ill and die in childhood or may live to adulthood with rare hospital treatment, yet these more specific factors are unknown and unknowable prenatally (Lanng et al, 1991; de Arce et al, 1992; Hamosh et al, 1992; Hamdi et al, 1993);. For parents forced to contemplate termination of a pregnancy, additional information regarding how severely their child would be affected by CF would be welcome. This information is invaluable in terms of treatment and cure such that this is one step on the time line of genetic identification (Worton, 1993) .



This time line can take years if not decades and to date there is no example where knowing the location of the gene and being able to detect it has led to a cure. However gene identification may lead not to a cure but to appropriate therapy which would be considered an acceptable goal (Elliot, 1993; Harris, 1993). Carrier testing is understood as a small step on the way to a cure. The need to enumerate carrier mutations and especially to establish a data bank of phenotype/genotype links is clear. However, carrier screening in the general population does not contribute to this knowledge bank because phenotype/genotype links can't be studied in carriers. It can only be studied where the disease is expressed. The American Society of Human Genetics issued a statement in which they expressed reservations concerning society's preparedness in terms of education, laboratory and counseling aspects of a wide scale program.

They unequivocally stated their position that "routine CF carrier testing of pregnant women and other individuals is NOT yet the standard of care in medical practice" (their emphasis) (American Society of Human Genetics, 1990). However, as more information is gathered from research protocols, position statements reflect changes in knowledge and practice. NIH released a consensus statement in April 1997 in which they recommended genetic testing for CF be offered to couples planning a pregnancy and/or seeking prenatal care (i.e. the woman is already pregnant) (NIH Consens Statement 1997).

1.2 (3) How risks are understood

Women are expected to make their own decision regarding testing and/or subsequent abortion based on a thorough knowledge of risk. The provision of such knowledge is to be provided them via the genetic counseling session. Genetic counseling is provided by the physician or, increasingly, by a specially trained genetic counselor. The counselor provides information regarding the general etiology of the disorder under investigation and tries to provide an accurate assessment of the particular risk level for each particular couple. The expectation is that women will respond rationally to statistical information concerning their risk of having a child with a particular anomaly. This belief in the rationality of the decision making process has propelled some researchers to create a "decision tree" that includes mathematical formulae showing the relevant probability and relative cost of each potential outcome (Perlmutter Pauker and Pauker, 1979). The research highlights that the response to

counseling differs from expectations. A counselors' most important task is to convey enough information to the client such that the client is capable of making an informed decision. Kessler states that anywhere from 20 to 75% of people counseled do not remember or understand the information they receive from a genetic counseling session (Kessler, 1980). Similarly, Rona et al (1994) found that while couples at risk of having a child with a genetic disorder appreciate the counseling they receive, many of them could not remember important facts concerning their risk status. Moatti et al (1990) found that education level correlated positively with one's ability to understand one's own level of risk but Ekwo et al (1985) found this to be untrue. The problem therefore is not so much one of understanding the mathematical nature of statistical probabilities but rather in the individual's interpretation of statistics. The concepts of a 1 in 4 risk or a 25% risk are often perceived differently by the same individual (Kessler and Kessler, 1988).

Research indicates that people do not necessarily make "rational" decisions based on quantitative analysis (Beeson and Globus, 1985). Lippman-Hand and Fraser (1979a; 1979b) found that people's assessment of risk takes the form of a "binary" interpretation in which they perceive themselves as having one of two possible outcomes: either they will or they will not have an affected child. The difference between acceptors and rejecters of amniocentesis is repeatedly seen to lie in the subjective interpretation of objective levels of risk; women who reject testing feel that their personal level of risk was not particularly high, at least not high enough to warrant testing (Marteau et al, 1991).

1.2 (3) Interest in CF Screening/Testing

There are varying patterns of interest in CF screening and testing in different populations. Distinctions are made between couples who have a family history of CF (including aunts, uncles and siblings of a child with CF), women planning a pregnancy or who are already pregnant, and the general population.

1.2(4) Carrier testing for CF based on proband

The uptake for PND and abortion has been much lower than expected in couples with a child with CF.

TABLE 4

Attitudes toward abortion for CF (Parents of CF children)

% would abort	Region	Source
95	France	Boue et al, 1986
72.9	Germany	Passarge et al, 1984
65	Belgium	Denayer, 1990
52	Wales	Al-Jader, 1990
42	Belgium	Evers-Kiebooms, 1987
32	(Mennonites) New York (U.S.A)	Miller et al, 1992
20	New England (U.S.A)	Wertz et al, 1991
0	(Amish, Hutterites) New York (U.S.A)	Miller et al, 1992

The percentage of couples who say they would abort a subsequent child if it was found to have CF ranges from 0% among the Amish and Hutterites (Miller et al, 1992) to 95% of parents in a study in France (Boue et al, 1986). Obviously, groups with strong religious beliefs would be expected not to see abortion as an option. Interestingly, the proportion who say they would abort is *lower* in studies conducted *after* the discovery of the CF gene in 1989. In a study conducted by Wertz et al (1991) of 227 families with a child with CF, 20% said they would abort for CF. It is important to note, however, that this figure includes people who were not planning to have any more children. When only those couples planning to have more children were considered, 58% had used, or were planning to use, prenatal diagnosis. Of these, 44% would use PND to prepare for the birth of a child with CF and 28% would prevent the birth of a child with CF. Prenatal diagnosis does not have a direct link with abortion for all couples although the tendency has been to assert this link over that of using PND as a way to be financially, emotionally and psychologically prepared. The parents in the Wertz study had high expectations for their children with CF. Most parents believed that their children would live independently (78%), hold a full time job (77%), and marry (67%) (Wertz et al, 1991). Given the sequelae of the disease, it is not unwarranted that many couples feel life with one or more affected children is far from impossible and with proper treatment and social support can be quite manageable.

Nevertheless all parents do not automatically rule out prenatal diagnosis and/or abortion of an affected fetus. A 1994 study in the U.K. of adult CF patients and their parents found that both supported

prenatal screening (88% and 90% respectively) and the option of terminating an affected pregnancy (68% and 84% respectively) (Conway et al, 1994). It could be that these parents are looking at the issue from a much longer term perspective than parents of very young children, they are past the age when they must decide whether or not to have more children. This may provide some emotional distance from, and thus depersonalize the issue of abortion. Or perhaps with changing times and increased information about genetics in the public media, parents have fewer concerns about possible stigmatization and other ethical issues.

This 1994 study of parents is similar to studies of the attitudes of other relatives of children with CF. More specifically, it has been found that siblings, aunts and uncles have different attitudes toward prenatal diagnosis for CF; they tend to be more in favour of prenatal diagnosis and abortion than parents of a child with CF (Denayer et al, 1992). Approximately 65% of aunts and uncles tended to rate the burden of the genetic disease as "moderately great" to "very great" although they stated that they felt they would be able to cope nonetheless (Conway, 1994).

1.2 (5) Population based carrier screening

Carrier screening is an issue separate from that of prenatal diagnosis. Although carrier screening *may* be linked to prenatal diagnosis, it is not *necessarily* so. Community carrier screening for the Cystic Fibrosis gene is not standard of practice in any country at this time. Nevertheless, there is widespread discussion of the possibility, weighing both pros and cons in terms of ethics and economics, medicine and law. Research in this area recalls many of the issues

which have been discussed in relation to families of children with CF as many of their concerns are the concerns of researchers and public policy makers as well. Specifically, there are concerns about stigmatization, confidentiality and privacy, the portrayal and the perception of the disease, possible eugenic implications of screening and for some, a concern that carrier screening not usurp the position of research designed to help combat or cure CF.

Even for those quite certain of the efficacy and the value of screening programs there are questions surrounding how best to offer such screening to the vast population that may be carriers. Thus, several pilot studies have been underway to ascertain how to teach people about their genes, how to offer services in such a way as to maximize uptake, and how large a segment of the general population will be interested in this type of genetic service. Consequently, there are two dilemmas or issues regarding carrier screening that are intertwined and must both be dealt with; firstly are the ethical concerns inherent in carrier screening and whether such screening should be provided and if so, what criteria should guide the provision of such and secondly there are practical concerns of how to package and deliver a service within the health field.

1.2 (6) Criteria for screening programs

Community carrier screening has received much attention from governing bodies. The WHO has outlined several preconditions for community carrier screening that have received support from many countries (Williamson, 1993). These preconditions take into account that the needs and resources of different countries can be vastly dissimilar. Accordingly, the four conditions are as follows:

1. The disease should be a major health problem *for a particular country.* WHO recognizes that for many countries still grappling with high morbidity and mortality and very high rates of infectious disease in children, genetic services cannot be accorded a high priority. The understanding of what constitutes a *major* health problem is open to some debate. Is a health problem major in terms of the number of people it affects, or in terms of the extent of illness caused by the disease or some combination of both? Given that CF affects approximately 2600 people in Canada but that many of them expect to live well into adulthood, can it be considered a major health problem?

2. The clinical course of the disease should be defined and severe diseases should be accorded priority. WHO also states that the priority should lie in preventive measures. For CF this poses some difficulty because the rapid advances in research mean that the treatment and life expectancy are constantly changing. However, the underlying understanding is present.

3. There must be a simple, acceptable, and inexpensive carrier test, which is accurate. The information must be clear to both the carrier and to the health care professional. Again, CF poses a particular case as there is a wide variance in the accuracy of the carrier test. What level of accuracy is to be considered accurate and acceptable is an issue of intense debate.

4. It should be possible to act on the knowledge of carrier status to help solve the problem. Action can take the form of early intervention or by termination of affected pregnancies. Both early intervention or termination of affected pregnancies is available for people who discover that they are carriers of the CF gene.

Whereas it is apparent that the preconditions set by WHO can be met for CF community carrier screening, there are still several

issues that must be dealt with and in fact, are being dealt with at this time.

1.2 (7) Identification of carriers

The most onerous problem is the fact that carrier screening for CF cannot identify every carrier because CF carriers may carry the gene in one of more than 350 mutations, not all of which can be detected. This then leaves many carriers who receive a negative result as *false* negatives; they are not found to carry any of the mutations they have been tested for but they still do carry the CF gene. This can pose severe problems for individuals whose partner has already tested positive for a particular mutation. For these couples, carrier screening does not decrease their risk but, rather, increases their risk. For example, with no carrier screening available, a couple's risk of having a child with CF is 1 in 3,300 (NIH Consens Statement, 1997). At a detection rate of 75%, that rate can increase or decrease. If both partners test negative, their risk of having a child with CF is decreased to 1 in 39 200, a substantial decrease. However, if one partner tests positive and one tests negative, their risk actually increases to 1 in 396 (Wilfond and Fost, 1990). Only at a detection rate of 96% does the level of risk return to the original level: if both partners test negative, their risk is 1 in 1 500 000; if one tests positive and one negative their risk increases to 1 in 2450, almost exactly that of the original risk of someone in the population who has not undergone testing (Wilfond and Fost, 1990). For this reason, members of a workshop on population screening for CF stated that screening should not commence until it is possible to attain a 96% detection rate (Workshop on Population Screening for the Cystic

Fibrosis Gene, 1990; Biesecker et al, 1992). The worry herein is how to counsel and educate people about the imperfectibility of screening when a negative result does not necessarily mean a person does not carry the CF gene. As discussed previously, individuals tend to make "binary" decisions; i.e. they think they either are carriers or they are not. Individuals who test negative may process the information in one of two ways; they may feel that they do not carrier the CF gene and therefore put it out of their minds or they may feel that they are carriers and proceed accordingly. Axworthy et al (1996) found that 50% of individuals receiving a negative screen erroneously believed that their negative result meant that they were definitely not carriers. That same study suggests that recall errors consistently occur in such a way as to reduce perceived risk (Axworthy et al., 1996). Micheal Kaback, president-elect of the American Society of Human Genetics fears that inconclusive screening will do much harm to couples. He suspects that test results will increase anxiety and that some couples may end up aborting healthy fetuses (Roberts, 1990).

Indeed, editorials in major professional journals indicate that the issue of sensitivity of CF screening is of primary concern to physicians and geneticists, as well as ethicists working in the field (Colten, 1990; Harper, 1990; Biesecker et al, 1992; Kerem and Lynch, 1991; Gilbert, 1994). The main concern is whether population screening for CF is premature given the uneven detection rates. This point may actually become moot as more mutations are isolated, yet there is a consensus that inasmuch as certain mutations are responsible for only a particular familial strain of CF, a detection rate

of 100% may be unattainable(Gilbert, 1994). Given the dissension concerning what is an appropriate detection rate, both the American Society of Human Genetics and the Canadian College of Medical Geneticists have issued statements that the routine screening in the community was not the standard of care for couples with no family history of CF (Caskey et al., 1990; L. Robb, personal communication, April 1993). Regardless, genetic tests are fast becoming the norm in health care and therefore more research is felt necessary to ensure responsible and professional actions (Motulsky, 1997). To date, several pilot projects are underway, and they provide a glimpse of how the general community, with little or no previous knowledge of CF, react to the offer of carrier screening.

1.2 (8) Pilot Projects

There have been, to date, in the United States and Europe, several pilot projects designed to assess attitudes toward carrier screening in the general population: that is, people who have little or no previous knowledge of, nor experience with, Cystic Fibrosis (Mennie et al., 1993; Botkin & Alemagno, 1992; Decruyenaere et al., 1992; Green, 1992; Mennie et al, 1992b; Watson et al., 1992; Cobb et al., 1991; Williamson et al., 1989). Many pilot projects involve the appraisal of attitudes toward the offering of genetic services, and serve to help professionals in the design of educational materials such as films and written pamphlets to be used should carrier screening become the standard of care (Myers et al., 1994; Mennie et al., 1992b; Watson, 1992). Measuring uptake rates has proven to be difficult as many factors must be considered. Recent reports indicate a wide

discrepancy in uptake rates for several pilot projects (Axworthy et al., 1996; Brock, 1996; Loader et al., 1996).

1.2 (9) Attitudes toward screening

Overall, respondents show a positive attitude toward screening for carrier status of CF. Research measures the preferred location for screening and the theoretical and practical uptake rates for screening.

Preferred location for screening

Many studies have focused on an assessment of *where* respondents feel carrier screening would best be conducted. A study by Mennie et al (1993) indicated that the majority of respondents were in favour of screening being done in the office of a General Practitioner (GP); between 87-93% felt this was the most appropriate location. In addition 81-86% responded that they felt a Family Planning Clinic (FPC) to be the best place, and only 30-38% agreed that screening should be done in school. This latter statistic was supported by Green (1992) wherein only 34% of respondents felt that carrier screening should be done in schools. However, when approached in school, students felt that it was suitable for screening to be offered; 78% wanted to know their carrier status in a study by Williamson et al (1989); Cobb et al (1991) found that 86% of school children aged between 14 and 16 felt screening should be offered.

Uptake rates for screening: theoretical and practical

Interestingly, in a pilot project in Montreal, there was an *actual uptake* of 42% of school children aged 15 to 17 years old (Mitchell et al, 1993). The difference between a theoretical acceptance of

screening and its actual uptake is to be expected; even with parents of CF children, more feel that screening, PND and abortion should be available than avail themselves of these services. The dilemma of offering screening in schools is fraught with many concerns and will be explored in detail below. In the same study mentioned above, Williamson et al (1989) found that 89% of respondents approached at the GP wanted to know their carrier status as well as 91% approached at a FPC. Green (1992) states that in a population of new parents 83% (60% definitely, 23% possibly) were interested in screening when they were told that the detection rate was not 100% accurate. In a study of community members, 63% expressed a desire to know their carrier status (Decruyenaere et al, 1992). There was a theoretical uptake of 86% in screening in an antenatal clinic between October 1990 and April 1992, although only 73% of women were actually screened (Mennie et al, 1992a). It can be supposed that these women had a more immediate interest in their carrier status as they were in their reproductive years (Eng et al., 1997; Bekker et al., 1993).

Exposure to and knowledge of genetic disease may serve too increase acceptance of screening. Eng et al (1997) offered CF carrier testing to 2824 Ashkenazi Jewish individuals (1412 couples) who were already being tested for Tay Sachs disease and had an uptake of 97% for the CF test. In a large study conducted in an HMO, 78% of pregnancy women offered carrier tests accepted (Witt et al., 1996). Nonetheless, the majority of individuals feel that screening should be done before pregnancy (Botkin and Alemagno, 1992) Still, given the impossibility of this scenario, most are accepting of screening during

pregnancy, although this presents a more stressful event for those who are found to be carriers of the CF gene. The interest in carrier status is also be reflected in the uptake of 82% in a FPC in a study of individuals aged 16-44 with no previous knowledge of CF compared with only 66% in a GP and a mere 19% approached by written invitation (Watson et al, 1992). A study in Scotland where prenatal screening is routinely offered had an uptake of 70% (Brock, 1996). However, Clayton et al (1996) discovered a definite lack of interest in population based carrier screening for CF even when it was offered free of charge. Although 90% of respondents felt that genetic testing should be offered, less than 1% of those offered a free test elected to be tested. Loader et al. (1996) had an uptake rate of 57%, a low rate they link to lack of willingness to participate by prenatal care providers. The study was designed to offer free testing to pregnant and nonpregnant women through their health care providers. The studies coordinator approached 124 health care providers, however only 37 were willing to participate; most decline citing lack of time to explain genetics to their patients (Loader et al, 1996). This could prove to be a very important factor in genetic screening programs because the current rate of uptake for CF screening may very well be a result of "supply push" as opposed to "demand pull" (Bekker et al., 1993). Indeed, in one study 6.2% of women tested for CF indicated they did so because their doctor highly recommended it (Loader et al, 1996).

The main reasons given for wanting to be tested were to obtain reassurance and to avoid the birth of a child with CF but the

distribution of these factors varied greatly from study to study as Table 5 indicates:

Table 5

Avoidance of child with CF as a factor in genetic testing

Proportion citing factor (%)	Study source
27.8	Loader et al. 1996
5.8	Witt et al. 1996
20	Wertz et al. 1992
23	Conway et al. 1994

Witt et al (1996) found that 5.8% of women cited "I wanted to have the option to avoid having a child with CF" as a factor in decision making whereas Loader et al.(1996) cite 27.8% of women saw it as a factor. In addition, 50.6% of women in the Loader study cited they choose testing "for reassurance that my chance of being a carrier is low" (Loader et al., 1996). Most women who declined to be tested did so because either they felt their chance of being a carrier was very low (32.2% in Loader et al., 1996; 30% in Witt et al., 1996) or because they would not terminate a pregnancy even if the fetus was found to have CF (32.4% in Loader et al., 1996; 27.3% in Witt et al., 1996.

1.2 (10) Knowledge about CF

One of the primary tasks of pilot projects is to determine the extent of knowledge participants have about CF. Knowledge levels

vary considerably. Mennie et al (1992b) found that 92% of respondents had heard of CF but the extent of their knowledge prior to reading a leaflet describing CF was not measured. A similar study by Decruyenaere et al (1992) found that while 59% of respondents had heard of CF, only 38% could give a feature of the disease. This suggests that there is less actual *knowledge* of CF than there is *awareness* of the existence of CF. This could prove to be a significant factor, as Mennie et al (1992b) discovered in their study designed to measure how well respondents understood an information leaflet outlining CF carrier screening. This study indicated that three quarters of women who did not understand the purpose of carrier screening had not previously heard of CF. Providing information is therefore an important element of screening and testing programs.

1.3 Health care services

The category of health care services incorporates several aspects of genetic services. One of the services is that of providing information about genetic screening and carrier status to interested individuals. Health care services must also collect information of the impact of screening on these individual participants, including measuring any anxiety experienced by participants. Costs associated with screening must be addressed as well as costs of other avenues such as gene therapy and lung transplantation for people with CF.

1.3 (1) Providing information

Designing educational materials is an important preliminary step to establishing screening programs. As professionals, those involved in such programs are concerned that individuals have adequate information to make an informed decision about whether or not to be

tested for carrier status (Myers et al, 1994). To this end, leaflets, pamphlets and videos have been developed (Mennie et al, 1992b; Watson, 1992; Myers et al, 1994). The necessity for secondary methods of knowledge transmission arises directly from the fact that the resources of genetic counseling are inadequate to meet the needs of personal one to one counseling for large numbers of people.

In the meantime, most educational programs emphasize written materials and have found them to be for the most part adequate for the needs of the clientele. In a study by Myers et al (1994) in which they used focus groups to help design an information leaflet, they found that 75% of respondents without a family history of CF would choose CF screening it they were offered it, and that 60% of those reached their decision after reading the answers to the question "What is Cystic Fibrosis?". This study also found that different people have different needs in terms of education and knowledge about CF. For example, some people were primarily concerned with their chance of being a carrier, while others expressed more interest in their reproductive options including how a child with CF would affect the rest of the family. The authors of this study, with the help of the focus groups, were able to design materials that was able to respond to these various needs. It is an important, yet often overlooked, element in carrier screening that not all individuals would want the same information and would have the same needs vis a vis learning their carrier status. In general, pilot studies provide two elements; one is the actual genetic tests being provided, the other is the opportunity to

perfect educational materials and instruments designed to test the knowledge of participants.

1.3 (2) Impact of screening on respondents

With the completion of some preliminary pilot studies, it is now possible to assess the impact of the experience of being screened for carrier status on carriers and non-carriers alike. This is a significant component of screening as is generally understood that screening should not expose the individual to excessive stress nor should it cause them to experience a decline in their level of self-esteem. Hence, many pilot studies include a component designed to measure the psychological impact of knowledge and experience.

1.3 (3) Anxiety associated with screening

To date there has been little evidence that any long term anxiety is caused either by being tested or by being found to be a carrier of the CF gene. Indeed, Bekker et al state that "In the longer term the greatest problem of population screening would appear to be one of false reassurance rather than anxiety" (1994: 364). They make this statement on the basis of the fact that even though those with a negative test were told that there was still a 1:135 chance that they were carriers of the CF gene, fully 17% receiving a negative result believed that they were at no risk of having a child with CF.

Several of the studies found that there was some anxiety in those being tested but it was, for the most part, temporary. A study of women in an antenatal clinic found that 41% of the women felt "anxious" or "slightly apprehensive" about being screened, while 59% said that they felt reassured (Mennie et al, 1992b). In a similar

study it was found that female carriers had more ambivalent feelings about having been screened; 80% stated they were glad they had had the carrier test compared with 97% of women who were found not to be carriers (Mennie et al, 1993). Mennie et al (1992a) also detected that between 20-30% of women in a trial experience stress but that their stress levels were significantly reduced once their partners were found not to carry a detectable CF gene. Watson et al (1992) showed similar results: while 54% of carriers stated that they were anxious, worried or depressed initially following their test, two weeks later 32% stated that they were not worried and 37% stated that they were less worried: only 4% claimed that they were now more worried. In a six month follow-up, a full 55% stated that they were now not worried and 30% expressed that they were slightly anxious, worried or depressed. All but one of the people who remained worried after six months were planning to have children in the future which may account for some of their concern. Significantly, it must be noted that 64% of carriers said that after attending counseling, they felt less worried or not worried (Watson et al., 1992) This would seem to confirm the need not just for pre-screening counseling and information sessions but for post-screening counseling as well, especially for carriers. However, more long term study is needed. While no difference in anxiety levels between carriers and non carriers was found in a 3 year follow up of the psychological impact of population based testing for CF, Axworthy et al. (1996) did find that carriers had a poorer perception of their current health than did non carriers.

In summary, pilot projects conducted to date have provided invaluable data and information regarding methods of delivery of genetic screening as a component of a health care policy. These projects have indicated the need for educational materials (films and pamphlets) that respond to the different needs of the clientele as well as provided a forum for individuals to voice opinions on the best locale for such services.

Pilot projects indicate that theoretical uptake levels are consistently higher than actual uptake. Nevertheless there are considerable concerns that high uptake levels would strain human and financial resources and may result in inadequate counseling being made available. On a positive note, pilot projects indicate little long term negative impact of screening and/or learning one's carrier status on the individual.

The Montreal series of CF pilot studies is fundamentally different from other studies discussed above and therefore merits careful consideration on it's own.

1.3 (4) Montreal CF Pilot Studies

For the past 20 years, Montreal has been the site of an extremely successful screening program for Tay Sachs (Kaplan, 1992). Building on this earlier success, a series of pilot studies for CF have been ongoing in select Montreal High schools since 1990 (Kaplan et al, 1991).

The CF pilot studies involve three stages: education, screening, and evaluation. Students in high schools, aged fifteen and over, attend an information session designed to provide them with information about CF, the genetics of Cystic Fibrosis, and the limits of

the CF carrier screening in terms of mutations that are detectable and the meaning of a negative test result. Following the educational component of the study, students are invited to be tested. The first two trials, in the spring of 1990 and the spring of 1991 required parental consent for a student to be tested, however this requirement was abandoned when it was found to be an impediment to participation. In three separate trials conducted in three concurrent years, participation rates rose from 40% to greater than 70%. This dramatic rise in participation rates is linked to the withdrawing of the requirement for parental consent as well as to the switch from blood sample collection to the use of a buccal cell rinse (a mouth rinse). This latter change was indicated when many non-participants indicated that they refrained from participating because they were afraid of having blood drawn (Kaplan, 1992).

Limited published results indicate that in the Montreal studies nearly half of the carriers (4 out of 9) stated that they experienced great or modest anxiety upon learning that they had tested positive. Yet this initial anxiety has dissipated in all individuals at the follow up interviews (Mitchell et al, 1993). Advocates of screening in high school dismiss worries about the age and maturity of participants as "patronizing" (Scriver, 1992), claiming that Tay Sachs studies have dispelled this worry and arguing in my interview that people under eighteen are able to enjoy other privileges of society such as "drink[ing] and drive[ing]" .

The optimism of researchers regarding CF testing in high schools is linked to their success with similar testing for Tay Sachs and Thalassemia. With a twenty year history of testing providing

pre-established protocol, a framework is already in place within which to place CF testing.

1.3 (5) Costs

There are two ways of looking at costs associated with CF. There are the purely economical costs and there are the more intangible psycho-social costs.

Intangible costs associated with screening include the psychological health of the individual as noted above. In addition, especially where the reproductive plans of a woman are interrupted, Mooney and Lange (1993) have identified a list of tangible and intangible costs and benefits of prenatal diagnosis.

Intangible costs	Intangible benefits
Anxiety aroused through being informed of risk	Greater information
Discomfort of diagnostic procedure	Wider choices
Worry about test results	Reassurance
Qualms about contemplating abortion	Reduction of uncertainty
Wrong decisions caused by false results	In event of termination: avoid distress by not having handicapped child; greater likelihood ultimately of having non-handicapped child

Mooney and Lange (1993) highlight the fact that even when women choose not to participate in screening programmes they are affected by its very existence and therefore still incur costs (and to a lessor degree benefits). This research points to an important facet of medical innovation: technologies must be assessed not simply on their monetary costs but on other costs as well (Grimes, 1993). It is a point of fact that high cost technologies do not necessarily meet the needs of society and may, in fact, take money from more simple and necessary programs (Rutten & Bonsel, 1992).

Exact costs for CF screening programs are difficult but certainly not impossible to calculate. In 1993, Williamson calculated that a CF test to search for as many as 12 mutations cost between $100-300 (US dollars). He broke down costs in the following manner: enzyme, tubes, reagents would cost $7 (US, per test for 6 mutations) but if more than 250 tests were done per week it would be necessary to add $3 per test for lab salaries, overhead would add an additional $10 per test, another $5 would be paid as a royalty for the test. Counseling would add $25 to the total costs for a final cost of $50. Williamson believes that this figure would stand for European health systems but that in the United States, with it's preponderance of profit generating laboratories, costs will run three to four times higher, in other words between $150-200 (US). Although the NIH states that costs are changing rapidly, four years after Williamson's initial estimates, costs hadn't fallen very far: the NIH estimates a cost between $50-150 (US)(NIH Consens Statement, 1997). This can be compared with per person costs for Tay Sachs of $36.32 and Thalassemia of $16.33 (1994 Canadian dollars) (Mitchell et al., 1996).

These figures must be analyzed in relation to costs associated with treatment of a person with CF. Estimates are that it costs $40,000 per year in direct medical costs and $9,000 in ancillary costs per individual with CF. (This is the case in the United States, which has much higher medical costs than Canada and European countries). This represents a "net present value of approximately $800,000 for direct and ancillary costs associated with a CF birth" (NIH Consens Statement, 1997: 9). This can be compared with cost per identified CF fetus averted which are placed in the range of $250,000 to

$1,250,000 in the Caucasian population (NIH Consens Statement, 1997: 9).

Data do not indicate that screening is the best use of technology on a cost benefit analysis. Unfortunately, there has been no effort to quantify costs associated with intangible costs and benefits of screening. It is difficult to place an economic sum on moral and ethical issues. As indicated in the results and discussion in Chapter 5, the very idea of discussing costs in relation to the life of an individual with CF is distasteful to many.

1.3 (6) Lung transplants and gene therapy

The Canadian Cystic Fibrosis Foundation (CCFF) states that it is "making a major long term contribution to the enhancement of lung transplantation as a viable option for Canadians with CF" (Hancock, 1991: 6). To this end the foundation has allocated over $1.6 million to transplantation research, training and support services in the years 1985-94 (Hancock, 1991). There are currently five centres in Canada offering lung transplantation services to persons with CF: two in Ontario, and one each in Manitoba, British Columbia and Quebec. Lung transplantation is offered only when there are no other alternatives : the Toronto General Hospital, for example, accepts only individuals with 12 to 18 months life expectancy (Shennib, 1994). A rigorous assessment is undergone by all potential patients to ensure optimal chances of surviving the surgery; the survival rate for CF lung transplantation patients is about 60% (Shennib, 1994). The assessment alone can take as long as a week and a half to two weeks. Presently at the Montreal General Hospital, there is a six

month waiting period just for the assessment (Shennib, 1994). At any given time there are approximately thirty people on the Montreal waiting list for lung transplantation, of these about eight people would have CF. As for most organ donor plans, the wait for a suitable donor can be quite lengthy. Although there are two options for surgery; heart-lung or bilateral lung (double lung), the bilateral lung is most prevalent as there is also a lengthy waiting list for heart donors (Shennib, 1994).

Lung transplantation is not a cure but it may help prolong the life of an individual who has little hope left and who has exhausted all other treatment resources. Human interest stories in the CCFF newsletter, *Candid Facts* concentrate on survivors and their satisfaction with the procedure. Regardless, this option is not available as a widespread treatment of CF. With many more persons with CF hoping for a cure, many have turned their attention to the possibilities of gene therapy.

Although gene therapy, like lung transplantation, appears at first blush to offer a cure for CF, it cannot cure the disease at this time. Gene therapy would certainly help the CF lung but these lungs are already damaged and would therefore continue to be susceptible to infection. "Gene therapy would likely reduce the frequency of lung infections and slow down the rate of lung function loss..." (Cantin, 1992: 6) but chronic infections would still have to be controlled with other therapies. This is not to say that gene therapy has made no progress in the treatment of CF. While not offering a cure, research in gene therapy has yielded some new treatments and

some new drugs that help to control the worst effects of CF. One such treatment is DNase, marketed by Genentech, and sold under the name Pulmozyme (Hamilton, 1994). DNase helps to break up viscous secretions thereby making them easier to clear (Cantin, 1992). The downside of this is that the new drug is expected to cost between $10 and $20 thousand (US.) per year per patient (Medline, 1993), a cost that would be well beyond the reach of most individuals and the Canadian government which subsidizes drug costs for the chronically ill.

While the advances made by gene therapy may not have led to a cure at the present, each new find aids in this final, ultimate goal. In 1992 scientists were working on the insertion of an engineered virus (adenovirus) into the lungs of rats with CF. This adenovirus caused the respiratory cells to begin to produce a normal human CFTR protein, the effects of which lasted for two weeks to a month (Cantin, 1992). By the fall of 1994, scientists proved that they could transfer normal genes into the lungs of humans (*The (Montreal) Gazette*, 1 sept. 1994). By December of the same year, scientists had abandoned the use of the adenovirus and had switched to the use of microscopic bubbles called liposomes to implant the genes. This was the first time the gene had delivered without the use of a virus, an important milestone in gene therapy research (*The (Montreal) Gazette*, 31 Dec. 1994).

Treatments and advances in treatment are ongoing for CF patients. Indeed, it is the overwhelming advances in treatment that have contributed to raising the survival rate and extending the life span of persons with CF.

Conclusion.

At this time [population] carrier screening is not standard of practice. Population based screening for CF is a complex issue encompassing many concerns not least of which are those of stigmatization, confidentiality and privacy and possible eugenic implications of screening.

The World Health Organization (WHO) has published a set of preconditions which should be met before a community carrier screening program is established. Society has yet to deal adequately with these preconditions at this time; however, this is an ongoing project.

The most daunting task is that of dealing with the high number of false negative results that would be found given the current possible level of detection. Of major concern is how to counsel and educate people about the imperfectibility of screening when a negative result does not necessarily mean that an individual is not a carrier of the CF gene.

As a means of dealing with the logistics of this testing, several pilot projects have been conducted or are ongoing. These projects attempt to measure several things, including attitudes toward screening of the general population and knowledge and understanding of genetics and the limits of CF screening. Overall, respondents have shown a positive attitude toward screening and some researchers have moved on to trying to determine the best time and place for such screening. Furthermore, educational

materials are being designed and tested to determine how best to communicate to the general population.

By piggy backing on a well established Tay Sachs screening program, researchers in Montreal have been able to set up a successful program of CF testing in area high schools. This has not been without criticism however, as many researchers continue to feel that the lack of specificity of the CF test means that testing, especially of teenagers, should be on hold. An argument that has yet to be resolved centres on whether or not there is a "consumer" demand for screening, and if so whether or not the medical community has an obligation to respond to this demand. Preliminary findings indicate that there is no such demand but rather that proponents of screening are trying to create a demand. This is of vital importance as the history of Tay Sachs and Sickle Cell screening indicate that for a screening program to work, it must have the solid support of the community. Social change that appears to come from the top down rather from the grass roots up will be more strenuously resisted. Changing practices and changing attitudes are both crucial elements to a smooth integration of any new social phenomenon.

Chapter 2

Theoretical Framework

This chapter provides the theoretical framework that guides this exploratory study. There are three key sections: 1. Michel Foucault's theories relating to the body, power and knowledge; 2. Theories of social change; 3. Rationale for presenting medical innovations as a social issue which includes a summary of Diana Dutton's research examining positive and negative consequences of medical innovation.

Michel Foucault acknowledged that the physical body is constrained by cultural forces. In addition, the body is created by social context and is lived according to dictates of this knowledge. Foucault did not see such a thing as a natural body but rather all bodies are cultural representations and are culturally and socially defined and created.

The application of this theory to genetics is essential. Foucault analyzes the normalizing gaze and it's links to eugenics in ways that are elemental to genetic testing. The framework and the boundaries provided by his theories serve to encode the data with meaning.

Theories of social change provide a sociological perspective from which to understand how societies change when confronted with technological breakthroughs. For example, the arrival of personal computers on the mass market has precipitated many changes including but not limited to their intended use of computing and word-processing: telecommuting has changed the individual's relationships to work and to work colleagues; personal computers in

the home and in school change the ways in which our children are educated and even the meaning of "education"; social relationships that once were conducted in person or over telephone lines can now take place completely "on-line" where individuals may have an intimate relationship but never be in the same room together. Most of the changes were not manifest changes anticipated by the creators of computer technologies and yet these latent changes also have serious ramifications for society. So, too, medical technological change does not limit itself to changes in the way doctors "do" medicine. The ability to detect the presence of a gene such as Cystic Fibrosis brings with it a plethora of manifest and latent functions. Thus the repercussions of such a discovery are felt at many levels and in many areas of the social world and it's institutions including religion, health, economics and the family.

Given this, it is theoretically and methodologically sound to present medical innovations as a social issue. Essentially, individuals in society have a vested interest in medical innovation even when they, as individuals, may not need or use those particular innovations.

In the area of the relationship between social issues and medical innovations, Diana Dutton has written a seminal work: *Worse than the disease: Pitfalls of Medical Progress* (1988) . Using four case studies, she elucidates the decision making process regarding technological breakthroughs. She outlines the forward and backward steps of specific medical innovations: the artificial heart, DES, the swine flu immunization program and genetic engineering. These last two case studies in particular are relevant and of interest for this

research project. The experience garnered by the swine flu immunization program may provide insight into the difficulties inherent in attempting to establish a widespread program within the population aimed at "improving" health and/or preventing illness: this experience is relevant to this analysis of a screening program for CF. Dutton's analysis of genetic engineering is insightful in many ways that are relevant to this topic as well, especially in the sense of context she provides for social-medical-economic realities. Dutton's scholarship helps to form and to validate the work undertaken and presented herein.

2.1 Foucault, Genetics and the (social) Body

The works of Michel Foucault are indispensable to any project that looks at medicine, power and the body. While many of this original ideas have been integrated into current theories, their emergence nearly 20 years ago was revolutionary. Foucault was one of the first theorist/philosophers to articulate a theory of medicine and power and to show the social origins of discipline and punishment not just as they relate to the penal system but to the physical social body. It is this facet of Foucault's theories that makes it such an important element for this thesis. Genetic research has the capacity to strengthen power dynamics that are vested in the medical profession and through this to increase the effects of the discipline of the individual and the social body.

Genes exist at the level of the individual body. Each person has a specific genetic "code" that plays a part in determining the workings of their physical body. The extent to which any person is

personally affected by genetic research will vary in terms of whether that person *in particular* carries harmful genes *that can be detected.* Yet at the same time, everyone will collectively bear the burden of our genetic knowledge, for the social body is also deeply affected and this is most apparent in changes in our attitudes and behaviors, as they are manifested in economic policy, law and standards of care in medicine.

2.1 (1) Political technology of the body

Foucault depicts individuals as subjects/objects of strategic games of power/knowledge: subjugation is a political technology of the body obtained through violence and through the rationalization of the lifeworld articulated by the organization of disciplinary knowledge

> ...there may be a 'knowledge' of the body that is not exactly the science of its functioning, and a mastery of its forces that is more than the ability to conquer them: This knowledge and this mastery constitute what might be called the political technology of the body (Foucault, 1977: 26).

This political technology of the body refers not only to the individual body but also to the social body. A reading of Foucault is vital to analyzing how the body, sexuality, and gendered subjectivity are produced within specific networks of power relations. This is the basis of a Foucauldian theorization in this thesis. Foucault elucidates the history of the body and also makes clear that this history is written not only on the body but through the body. The subjects in whom power is vested are not outside the sphere of those upon whom power is exerted but are included within this sphere. Thus physicians and researchers as well as the men and women who may,

in the near future, have their genes "screened" are all part of the same system. And while it may be true to state that society, or relations of power, construct our choices for us, the allure of genetic testing and screening to some groups cannot be explained by so limited a theory. While scientists and physicians have their own specific agenda, for the layperson it is helpful to understand the desire for genetic screening as an indication of inner disciplinary techniques that Foucault describes.

2.1 (2) The docile, gendered body

Foucault has been criticized for his over-reliance on the concept of docility and for neglecting to entertain ideas of *agency*. This is an important omission particularly when he places so much emphasis on the cultural constitution of our bodies, for what is culture, if not a human creation. Indeed, if there is no "natural" body it is because all human bodies are experienced through their inclusion in a particular society, or culture. The tangible artifacts of each culture are contingent upon the existence of human bodies upon which to hang these artifacts. The choices made, consciously and subconsciously, are made through human agency; that is, they are able to be made because humans have a sense of agency.

However, we cannot discard the idea of docility out of hand, for it is one result of discipline, especially inner disciplinary techniques that Foucault brings to our attention. Our choices are limited by factors and forces outside of our command. It must be determined to what extent our capacity for choice is usurped by power relations within society. In other words, yes, we can make choices and those choices are socially constructed but they are not always knowingly

and meaningfully constructed. It is important to avoid theories of social construction which consistently portray women as doubly victimized: victimized by plots to control them and victimized by their own ignorance.

Is Foucault's concept of docility a gendered one? If it is, we would have to accept that it is a strictly male concept and we know by experience that this understanding of the world is false. We must therefore accept that while he does not make it explicit, docility is meant to take into account both men and women. By no means to I mean to say that men and women express the docile body in the same way, simply that both are affected by and party to the abstraction of the docile body. The body, according to Foucault, is always in the grip of cultural practices--there is no "natural" body. Cultural practices, far from exerting their power against spontaneous needs, basic pleasures or instincts, or fundamental structures of body experience, are already and always inscribed, as Foucault has emphasized, "on our bodies and their materiality, their forces, energies, sensations, and pleasures"(Foucault, 1978: 155). Our bodies are constituted by culture.

Foucault does not take gender into account in his theoretical analyses but it is possible to elaborate on the gendered subjectivity that arises in particular with the geneticization of women. While geneticization refers to both men and women there is little discussion of men's participation in genetic screening programs. Women were consistently referred to as giving researchers an "in" to the family: women are the target group for research programs designed to predict uptake levels for genetic testing and screening services.

2.1 (3) Foucault and the Body

Foucault's conceptualization of the "docile" body is crucial. "A body is docile that may be subjected, used, transformed and improved" (Foucault, 1977: 136). Foucault terms "disciplines" the methods through which the body is used, transformed and improved. Disciplines incorporate " a policy of coercions that act upon the body...The human body was entering a machinery of power that explores it, breaks it down and rearranges it...Thus discipline produces subjected and practised bodies, `docile' bodies" (Foucault, 1977: 137-8). Tied in closely with the idea of the physical setting of the panopticon as a disciplinary tool is the image of discipline of the body in it's more conventional sense. The body here is seen as a machine and therefore able to be trained much the same way that machines are "trained" or programmed. Discipline is achieved by active training, much the type of training we would now associate with army training. In this disciplinary technique, bodies are all trained to be the same. Each motion, each step of each activity, has been timed and perfected so that the body is operating as a perfect machine. Each body has the same clothing, the same haircut, they perform each function in exactly the same way. Schedules and timing are distinct aspects of training and the training of the body becomes the most important form of domination of the body (Shumway, 1989: 124).

The assumption of the docile body negates any degree of agency and Foucault has been criticized for disallowing any notion of agency in human actions. If we allow for a certain amount of agency

in individual action and activity, the docile body as a interpretive concept still has meaning for us at the same time that we are able to allow for individuality and personal expression. For even within a state of totalitarianism, one may catch glimpses of agency in individuals. Individuals who question the dominant discourse are still subjects/objects of disciplinary techniques but at the same time, they are able to form an image of their domination and to question its' origins and its' source of power. The body as subject is not only subject *to* power but is also a part of the process of power relations. For Foucault, power was understood as being both productive and repressive.

Individuals as consumers and subjects/objects of medical expertise and medical technology provide an excellent example of docile bodies. And although the docile body of Foucault's theory was consistently desexualized, this to be especially true for women and reproductive health care. Women who incorporate genetic screening technology as a part of their reproductive health care plans have consistently been portrayed as quite simply docile bodies acted upon by outside disciplinary forces. The idea of the docile body is predicated on the notion of malleability; i.e. that a body can be pressed into a particular shaped, power can be exerted upon a body to ensure that it correspond to a previously determined form, that it can be and should be normalized. The malleable body is a gendered body and the form and the techniques by which the particular forms are realized are differentiated by gender. Thus, while the military may be seen as the legitimate place in which to train the male body, the medical system is implicitly understood as the training grounds

for the female body. Although power is understood by Foucault as being vested in the very bodies it controls, under a patriarchal ideology, the power of the medical profession falls disproportionately to men even while it is exerted disproportionately over women.

2.1 (4) Surveillance

For Foucault discipline is an unambiguous concept. Yet it is vital that we not misinterpret what is meant by discipline. Not only does he mean discipline in the ordinary way but he also means *surveillance.* This meaning is often lost in the interpretation from the original French. The term *surveiller,* is translated as discipline but it must also contain in translation the idea of the original which is surveillance. Thus discipline incorporates surveillance which in turn subsumes the idea of the gaze.

The Panopticon serves as an apt metaphor. The panopticon is a model for prisons that is built in such a way as to afford no prisoner the luxury of privacy. While it is impossible, and each prisoner is aware of this impossibility, for the guards to see each prisoner at all times, no prisoner can ever be certain that he isn't being watched at any particular time. Therefore he must assume he is being constantly surveyed and govern himself accordingly. The result: each prisoner is actually engaged in self-surveillance. Yet this architecture is not limited in its use for the prisoner only, but for others; the school child, the insane, the medical patient.

This model, wherein everything is controllable, can be comforting because there is no room for chance, everything can be predicted with certainty. When disciplinary techniques are ordered from above, the individual can see power as manifest in someone

else. But for the subjects, discipline and surveillance become self-discipline and self-surveillance. Discipline is not bound to any particular institution and this makes actions appear to be voluntary since the anonymity of power leads to invisibility. Bartky (1989) asserts that women in particular are committed to self-surveillance, characterizing it as a form of obedience to patriarchy. This is certainly true when women demand such services as prenatal diagnosis, ultrasound and amniocentesis. In making these demands as an expression of one's agency, she has become "just as surely as the inmate of the Panopticon, a self-policing subject" (Bartky, 1989: 81). In doing so, she is also tacitly accepting two tenets of the Panopticon, visibility and unverifiability. *Visibility* in that the woman's pregnancy is an outward fact, physicians and others are aware of her condition and she in turn is aware of the possibility of their surveying her behaviour at any one time. *Unverifiability* in that she does not know for a fact that she is being watched. I posit another sense of these terms for the pregnant woman especially if she is at risk for a genetic disorder in her fetus. If she chooses not to use the prenatal technologies available to her, her choice is made "visible" to her physician and she is unable to verify beforehand if she is carrying a child with a disorder therefore if she does give birth to a handicapped child, she will carry that child with her as a verification of her inadequacy of self-surveillance; that is the child is a public verification that she didn't do all that she could to have a healthy child. This demonstrates how discipline and power are linked. Although women do make individual choices about prenatal diagnosis, these choices are part of the system of discipline and

therefore are constrained choices. And yet the very anonymity of power leads to invisibility and the negation of the existence of power outside one's own agency.

2.1 (5) The practice of power

To understand power, the key question we must ask is not "What is power and where does it come from?" but rather "How is power practiced" (Deleuze, 1986: 71). Power is maintained not through the use of force or through the imposition of laws "but the regulation of the population through various techniques, such as the stimulation of the birthrate or the improvement of the health and longevity of the population" (McNay, 1992: 68). Foucault(1978) asserts that power is organized around the management of life and therefore sex becomes a crucial target. Sex is understood as the origins of life and the regulation of sex and sexual relations allows for the regulation of life itself. According to Foucault's *The History of Sexuality, Vol. 1*, the regulation of sexuality is a result of the importance given the blood relation and it is here that he first links sexuality with eugenics. Foucault equates eugenics with racism and racism with eugenics (1978: 149). Early nineteenth century eugenics did have it's basis in the regulation of sexuality, more specifically, the regulation of who could and could not have sex and thereby reproduce. Control was not merely centered on sexuality but also focused on all aspects of marriage, family, education and property. Thus eugenics necessitated the extension and intensification of micro-powers. In fact, Foucault (1978) claims that eugenics was the original justification for surveillance and regulation of the body, health and private conduct. The concern was with the *kind* of people being

born. The solution is a form of governmentality--targeting the individual as a means to maintain social control. Social control, in early eugenic terms, is the control, not so much over who is being born, but over who is giving birth. This is a subtle distinction that is often missing from current debate. Foucault links Eugenics with it's political counterpart, Nazism. For what is Nazism but a political expression of the belief in the purity of blood and the triumph of the race. And how best to achieve this lofty goal than through the ultimate satisfaction of governmental control (Foucault, 1978). Yet a society does not to espouse Nazi beliefs to believe that there should be some government control over citizens. Governments do exercise influence over the family and reproduction through tax grabs or tax breaks. In Quebec, women were paid cash bonuses for babies; a third child received 4500$. This is a clear expression of power exerted. The crucial difference is that such bonuses were made available to all women with no racial or class distinctions.

2.1 (6) Discourse

To properly understand how the twin concepts of power and (self) discipline can be practiced, it is necessary to understand the concept of discourse. Foucault's method of genealogy is a method of looking at discourse that incorporates history. Discourses have uses for particular groups and therefore we will hear the discourse of the powerful, all other discourses with be marginalized or silenced (Shumway, 1989: 11). The first principle of genealogy is therefore that of reversal. In this way, Foucault searches for alternative interpretations of history, interpretations that may not follow the traditional interpretation, as outlined by the traditional, accepted

discourse of the powerful. In doing so, not only does Foucault discredit the notion of one interpretation of historical events, he also calls into the question that history is a linear progression from the past to the present to the future. Because we attempt to understand the past as an indication of linear progression to the present, we are doomed to misunderstand (Shumway, 1989). We give history a sense of order and meaning that isn't there through our insistence on the "truth". Nevertheless, the truth does not exist apart from it's creation as the discourse of the most powerful.

This has important ramifications for understanding the debate over genetic testing. The very framing of the issue has tried hard to stay away from the subject of eugenics. The discourse has focused on the benefits of genetic research and has consistently denied that there are links between the mentality behind early eugenics and genetic research. That link is the belief that our genes are responsible for social ills and that by controlling our genes, we can exercise control over society. Arguments to the contrary have been marginalized, most often by confining them to the small corner of ethics or bioethics. The power of dominant discourse to marginalize others is demonstrated the fact that the Human Genome Project has allocated only 3% of its' total budget for bioethics (Lippman, 1992: 1469).

The example of eugenics points to the fact that power can be understood as a "normalizing" technique. The judicial institution operates to regulate, to establish norms (Foucault. 1978: 144). The complexity of disciplinary and discursive actions is aimed at the establishment of norms. The discipline of the body, and the creation

of the docile body, are achieved on the basis of an idea of the norm. Without having first established what is "normal", for example, in terms of how to carry out a particular task, there is no sense of how the body should be disciplined. Once norms have been established, however, there is something against which everything else can be measured to see if it measures up. Eugenics is just such a normalizing technique. The first step in deciding who should reproduce, or more recently, who should be born, is first in establishing norms. What is "normal" for a human being? What genetic make-up falls outside the boundaries of our agreed upon norms? These are the first questions in establishing a eugenic policy or in undertaking genetic screening programs. These assumptions about normalcy are then transferred to the legal arena. Laws exist to identify the normal and the not normal (abnormal).

Surveillance, power and control are not limited to the prison but are found everywhere in society. The judicial system is a judge of who is normal but so too is the medical system. While the doctor is also the judge, power is not vested in the individual doctor but rather in the institution of medicine, and "since no one holds power, no one can seize it" (Shumway, 1989: 139). Just as the prisoners in the Panopticon are subject to the power of the gaze stemming from the inner tower, so too, everyone is subject to the power of the medical/scientific gaze. The gaze has the power to objectify all that it sees, the power of the gaze over its' objects is clear and the object is then constituted by the observer in his terms (Shumway, 1989). The very act of seeing is conditioned by the discourse and therefore to be examined, as by a doctor, is to be subjected to the power,

control, surveillance of institutional privilege and to be subjected to the norm. This has profound implications for medicine and provides us with a way to understand the "gene screen" in such a way to minimize our reliance on social construction theories and theories that preach the power of patriarchy and the victimization of women faced with the dominant medical profession. This is not to suggest that these elements do not exist, merely to suggest that they are lacking in their ability to analyze the complexities of power and control in our society. This theory locates [the position of] power as outside of individuals at the same time as it recognizes that power is vested in individuals. Can we be both oppressed and oppressor? Foucault definitely says yes.

2.1 (7) Genetics and the social body

A Foucauldian analysis of genetics is enlightening because Foucault provides a way to understand events that goes beyond theories of some great power ruling over individuals in the name of some giant unknown goal. Patriarchal forces are at work in our society but we cannot deny that not all men hold power nor that not all men want power nor that not all men would abuse power if they did have it. When we discuss reproductive technologies, we cannot simply state that men are creating these technologies to control women. While that may well be one end result, there is not a group of men sitting together in a lab somewhere drawing up plans to take over women's choices. That type of explanation that was so popular in the sixties and the seventies is much too simplistic for the nineties.

Foucault allows us to examine issues of power and dominance as they are experienced through the individual body and through the

social body. We are wise to remember that the individual body and the social body are more than just a physical entity. Because this is so, ethical and moral considerations are very important.

Discourse has intricate links with power because those who have the ability to control, or guide discourse have the ability to control or guide what others see and believe. In *The Birth of the Clinic* (1973) Foucault points out that changes in medicine are linked to changes in discourse, not changes in actual bodies. Where it was once believed that a disease had infiltrated the body and had to be found, now it is believed that the body itself is ill. In the modern language of genes and genetic research, a body can be ill without expressing any manifestations of illness but merely by "carrying" genes for particular disorders. Abby Lippman (1992) argues that discourse can actually "create" illness by framing what it means to be ill or framing what an illness is. She asserts that the medical profession has the power to create illness through discourse. This is actualized when research groups, or individual researchers decide to take on the task of "discovering" a gene for a particular disorder. The very act of receiving a grant for the research legitimates their belief that the disorder they are researching is serious; it is dangerous enough, or lethal enough that it warrants the expenditure of monies and it needs a "cure".

Conclusion

Foucault provides a window through which to see the competing concerns of genetic research. A Foucauldian analysis of genetics allows for the discernment of nuances of how power is practiced by the individual and *on* the individual. Essentially, as

culturally constituted bodies, we create the idea of the body and we assume the discipline of the body such that it adheres to some defined norm. The body is subject to the normalizing gaze of genetics in the same way that the prisoner is subject to the normalizing gaze of the guard. The difference is that for genetics we are all both prisoner and guard at the same time and in the same space.

2.2 Theories of Social Change

Generally speaking, social change is a shift in the characteristics of culture and society. I will discuss herein the three sociological theories of social change in greater detail so that a better grasp of the purpose and relevance of social change in medical innovations is understood: 1. evolutionary theory, 2. conflict theory, 3. functionalist theory.

2.2 (1) Evolutionary Theory

Evolutionary theory stems from Charles Darwin and holds that there is a continuing progression of successive life forms. Auguste Comte (1798-1857) and Emile Durkeim (1858-1917) furthered this notion. Comte stated that human societies move forward in their thinking from mythology to scientific method while Durkheim believed that society progressed from simple to complex forms of social organization. Both Comte and Durkheim believed that change was unilinear: change progressed in one direction. However, modern theorists advocate a multilinear theory: change occurs in several areas of a society at once and all change does not necessarily lead in the same direction.

2.2 (2) Conflict Theory

Conflict theory affirms that social institutions and practices continue because the status quo is maintained by those who hold power. Change is needed to correct social injustices and inequalities and conflict is a instigator of these changes and therefore is a normal and desirable aspect of social change.

The origins of conflict theory are attributed to Karl Marx (1818-1883). Conflict theory regards the history of human society as being characterized by struggle over scarce resources which leads inevitably toward social change. The doctrine of historical materialism posited by Marx highlights his belief that "the mode of production of material life conditions the general process of social, political and intellectual life. It is not the consciousness of men(*sic)* that determines their existence, but their social existence that determines their consciousness" (1972: 20-21). Hence, we become who we are because of the political, economic and social conditions in which we exist and struggle.

Although Marx's original impetus for the formulation of his theory was his interpretation of labour relations in an increasingly industrial society characterized by alienation and inequalities between capitalist and worker, these theories have been restructured to provide a framework for understanding gender relations in today's society (Tong, 1989). In addition, feminist theorists writing about reproductive technologies and women's relations with the discipline of medicine have found conflict theory with it's emphasis on issues of power and control to be helpful and enlightening (Corea, 1985, Rowland, 1985 are two of the earliest

writers on this topic). In this analysis, doctors are placed in the role of capitalist and women in the role of the worker. Doctors control access to resources which include not only the technologies of medicine but medical knowledge as well (Overall, 1989). The rationale behind this is to maintain control over professionalization, economic reward and (as some feminist scholars maintain) the female body; a rationale that is made obvious by the example of the history of midwifery in Canada, a history characterized by territorial struggle between doctors and midwives (Burtch, 1994). Women, in turn are subject to the will of the doctor and limited in their own sense of autonomy in terms of making health care decisions (Merrick, 1993). The control of medicine over birth control and abortion services is characterized as a means to control women by controlling their reproduction (McDonagh, 1993). Even when women become doctors, the culture of medicine remains decidedly male, so that female doctors are portrayed as having capitulated to a male-centered belief system that guides their own practice of medicine.

Conflict theory portrays social change as occurring when different groups compete for resources in an unequal situation. Change functions as an upheaval of the status quo. Feminist theory has built upon this concept to create an understanding of women's social position. Social change is the result of women struggling with a patriarchal power structure to bring about a system of gender equality.

2.2 (3) Functionalist Theory

Functionalist theory and conflict theory are frequently portrayed as opposite: conflict theory highlights the need for social change whereas functionalist theory emphasizes the necessity of social stability. However, these two theories are compatible in that society may be characterized as having periods of stability in turn with episodes of serious conflict. Indeed Marx advocated social change not as an end in itself but as necessary so that society could run more smoothly and more fairly for all citizens. In turn, Talcott Parsons (1902-1979), the "father" of functionalist theory also felt that social change could lead to new and important functions in society.

Parsons declared that society is naturally in a state of equilibrium; a state of balance and stability. Each element of society is part of a system that functions smoothly: the family, religion, economics, education, deviance for example all have their separate, equally important functions to fulfill. Any change that occurs in one system, one part of society, engenders adjustments in other parts of society. If this adjustment does not occur, there will be strains in society that will threaten the stability of that society. Parsons (1966: 21-24) maintained that four processes of social change are inevitable:

1. Differentiation: the increasing complexity of social organization,

2. Adaptive upgrading: social institutions become more specialized,

3. Inclusion: accepting groups that were previously excluded due to gender, race, social class background,

4. Value generation: the develop of new values that tolerate and legitimate a greater range of activities

All four of these processes emphasize consensus and critics argue that they disregard the use of coercion by the powerful to maintain the illusion of a stable society and that they appear to stress that social institutions won't survive unless they continue to contribute to the good of society which flies in the face of many experiences. However, functionalism does allow for the fact that although altering institutions will result in disequilibrium, over time, as each social system restabilizes through change, equilibrium will be restored and the social change will become part of a newly ordered society.

2.2 (4) Sources of Social Change

Sociology recognizes five sources of social change. 2. The Physical Environment, 2. Population, 3. Social inequality, 4. Youth, 5. Science and Technology. This last source of social change is of relevance to the research presented herein for reasons to be elucidated below.

Science is the body of knowledge obtained by methods based on systematic observation while technology is the application of such knowledge to the making of tools and the utilization of natural resources (Henslin & Nelson, 1996). Advances in science and technology can have enormous implications for social change.

With specific application to science and technology, William Ogburn(1922) saw two parts to social change. Writing in a time in which the amazing advancements in science and technology we are

dealing with today were barely even imaginable, Ogburn's analysis has proven to be apt and to withstand the tests both of time and increasing sophistication with regards to technologies invented. These processes are Innovation which is comprised of either invention or discovery and Diffusion.

Innovation is the process of introducing an idea or object that is new to a culture. Discovery is making known or sharing the existence of an aspect of reality, such as a new gene marker, while invention is when existing items are combined into a new form, such as an MRI machine. Diffusion, the second step, occurs when the cultural item is spread from group to group or society to society. This can involve something as simple as the hula hoop or as complex as new ideological thought such as a belief in alternative medicines.

A new technology can transform society in five different ways (Henslin & Nelson, 1996).

1.. Transformation of existing technologies (from a rotary dial to push button phones),

2. Changes in social organization (machine technology which gives birth to the factory),

3. .Changes in ideology (from communism to capitalism)

4. Transformation of values (what we value as a necessity is a function of what is made available to us by technology),

5. Transformation of social relationships (changes in family structure as a result of men and women working away from the home).

Nevertheless, there is some resistance to these kinds of social change. Resistance to social change is seen as occurring either

because some individuals or groups are frightened of social change or because they have a vested interest in the status quo. It is also important to understand that cultural factors shape change as well as resistance to change.

William Ogburn (1992) has pointed out that we cannot devise methods for controlling and utilizing new technology before the introduction of a technique. This means that nonmaterial culture (norms, values and mores) are forced to respond to changes in material culture (techniques and tools). What occurs is a "culture lag": what Ogburn characterized as a period of maladjustment during which nonmaterial culture is still adapting to new material conditions. This is a vital element in terms of reproductive technologies and genetic research. The cultural lag between material culture (technology) and nonmaterial culture (religious and secular values and beliefs) is the essence of this research and how this lag will be resolved is one of the foundations of this dissertation.

Conclusion

There are several theories that help to explain the phenomenon of social change. For the particular needs of this research conflict theory and functionalist theory, although frequently depicted as opposites, are both useful to further an understanding of how medical innovations enact social change. Feminist analyses of conflict theory are useful in that they provide a framework from which to examine the power struggles enacted over reproductive issues and in which to focus criticisms of many changes heralded by these new technologies. Functionalist theory demonstrates the ways in which institutions within society are connected. This leads to an in-depth

perception of how changes in one institution can lead to turmoil within other institutions. In addition, functionalist theory shows how these conflicts can be resolved. The concept of cultural lag to indicate the time gap between developing a new technique and developing the moral and ethical response to that technique is at the heart of this research.

2.3 Medical innovation as a social issue

Medical innovation are an integral social phenomenon and as such warrant discussion and analysis. Medical sociology has emerged as one of the fastest growing sub-disciplines in the field of social research. This section describes why this is so and also looks in detail at the work of Diana Dutton in the field of multidisciplinary analysis of medical innovations.

2.3 (1) Interpretations of technology

As elucidated above, science and technology are interpreted as being two sides of a coin with science representing the abstract knowledge and technology representing the concrete tools or techniques made possible by that knowledge. One common limitation in interpretations of technology and their effect on people is that the analysis is frequently limited to a discussion of particular techniques such as the ability to clone or experimentation with fetal tissue. But as Ursula Franklin demonstrates in *The Real World of Technology* (1990), technology is not limited to techniques or machines but more importantly encompasses a particular frame of mind or mind set. This mind set is so deeply imbedded in our own social world that it is

difficult if not impossible for many people to imagine a world in their absence.

Feminists view their sociological analysis of technology as fundamentally incorporating issues of power and control Marie Mies argues that "technology is *per se* political because it deprives us of control over events, or centralizes them in fewer and fewer hands" (1985). Thus those who acquire the techniques necessary to manage the technology also acquire a legitimacy to manage the individual and hence, society. While claiming to be value free, science is, in fact, inevitably value laden. Sylvia Tesh asserts that "there is no science uninfluenced by politics" (1988: 177). Franklin demonstrates two ways in which technology has been developed: work related (to make work easier) and control related (to increase control over the operation). In addition, she combines these with a growth model and a production model. The growth model tries to find the best conditions to encourage growth and tries to provide them. This model leaves much to chance. The production model, on the other hand, creates a situation where all conditions are (theoretically) entirely controllable and predictable. In this case the physician's power over the ordinary citizen is rarely questioned. The focus on individual cause and a subsequent individual plan of treatment is both a cause and an effect of the politics of science.

2.3 (2) "Technophilia"

Barbara Wright (1989) created the term "technophilia" to describe that a technological solution is often society's first choice when faced with human problems such as disability, illness, death. Technology

is a seductive force in medicine. Developments are pushed by science on one side and pulled by forces of demand on the other (Rutten & Bonsel, 1992: 567). According to Kathryn Ratcliff (1989) the discipline medicine is especially favourable toward the use of technology because medical education, public policy and the profit motive have as one common result the view that new and more technologies are a measure of medical progress. Physicians and the lay public appreciate the scientific method of inquiry. The scientific method is the foundation of both knowledge and technological precision, objectivity and the ability to make diagnoses and finally to treat illnesses. However, many new technologies are made available without being subjected to randomized control trials (RCT) as an objective and precise method of evaluation (Rachlis & Kushner, 1989: 71). In his classic work, John B. McKinlay (1981) outlined seven steps in the process of technology adoption:

1.a promising report; 2. professional and organizational adoption; 3. public acceptance and state (third-party) endorsement; 4. standard procedure and observational reports; 5. randomized controlled trial; 6. professional denunciation; 7. erosion and discreditation.

These seven steps highlight that technologies are not evaluated until they have established credibility in the marketplace. It thus becomes very difficult to withdraw them from use because doctors and the general public have developed faith in their efficacy. The "glamour of high-cost sophisticated approaches deflects the medical profession's attention away from more useful interventions (Rachlis & Kushner, 1989: 74) As well, in the case of many childbirth

technologies, low-intervention, hands-on skills are lost because they are not passed on to the next generation of physicians. . Once a technology exists and hospitals have paid for the tools in the form of the machinery, physicians feel a compulsion to use it for every possible scenario whether or not they personally feel it will ameliorate the outcome. As Rachlis & Kushner (1989) relate, the culture of the medical profession also makes it difficult for one physician to question what his/her colleagues are embracing wholeheartedly. When the norm favours the use of a particular technology, the compulsion comes not just from the profession, but from the patient as both parties want to be assured that the physician is *doing everything possible*.. In his book *What kind of life?* Daniel Callahan states we are in "a crisis about the meaning and nature of health and about the place that the pursuit of health should have in our lives" (1990: 42). As Callahan notes, there is not an intrinsic limit to health care needs, all new technologies are "needed" and the impetus thus arises to provide them regardless of cost. However, he argues that limits to needs must be set from the outside and imposed by political force: "otherwise, as we see in the United States, health :"needs" take on a life of their own, constantly escalating" (1990: 98). This regulation must come from a combination of efforts by government, industry, health care providers and consumers in order to be truly effective (Rutten & Bonsel, 1992). Without regulation technology is allowed to create and recreate its' own need. Two examples taken from reproductive technologies will clearly illustrate these points.

2.3 (3) Sonography and EFM

The seven steps in the process of technology adoption that McKinlay delineates highlight the way in which a technology begins with a specific mandate for use within a particular health need or population and then spreads to other areas. This is a common route for medical technologies and is made clearly visible in sonography and EFM where noncoercive and necessary medical technologies for the few result in being universally and coercively applied to the general population of childbearing women (Beck-Gernsheim (1989).

The National Institutes of Health (NIH) and the federal Food and Drug Administration (FDA) in the United States have recommended that ultrasound NOT be used as a method of routine screening, nevertheless it continues to be used in this way (Ewigman et. al, 1993). Criticism has been levied at the continued use of ultrasound as a routine diagnostic tool by many feminist critics (for one early example see Rothman, 1989). One reproach is that ultrasound has never been proven to be safe and effective. In addition, many conditions identified by ultrasound at 20-24 weeks self correct by the time labour begins and therefore do not need to be managed by medical intervention. Finally, ultrasound is routinely performed without obtaining informed consent from the women; they are, for the most part, unaware of any dangers or debate of the procedure and neither are they aware that they have a right to refuse the procedure (Gold, 1984).

Between November 1987 and May 1991, the Routine Antenatal Diagnostic Imaging with Ultrasound (RADIUS) trial was conducted by a group of more than one hundred physicians and

clinicians involving 15, 151 women at low risk (Ewigman et al, 1993). Pregnant women were randomly assigned to either a screening group which underwent two "routine" ultrasounds, or a control group who only underwent ultrasound for medical indications. The researchers stated uncategorically that *"screening ultrasonography did not improve perinatal outcome as compared with the selective use of ultrasonography on the basis of clinical judgment""* (my emphasis) (Ewigman et al, 1993). Despite these findings, routine ultrasound continues to be the norm, and in fact appears to be on the rise (Susan James, June 1994, personal communication). One of the main reasons for the continued use of a technology that has no medical benefits for more than 80% of the women who use it is that these women themselves have been instructed in a culture of childbirth that regards sonography as an important *rite de passage* for all pregnant women. While all women have the right to refuse an ultrasound, very few are aware of this right, and still fewer wish to avail themselves of this right. The technology of sonography is so deeply imbedded in the cultural values of our society that most women cannot conceive of a pregnancy that is not anointed with this experience (Anne Quéniart, 1988. This issue will be further pursued in later chapters). Before the advent of the routine ultrasound, the quickening of the fetus was a magical moment in a woman's pregnancy signifying the reality of the growing life inside her, this experience has been superseded by the verification by machine. Ursula Franklin points out "..the downgrading of experience and the glorification of expertise is a very significant feature of the real

world of technology" (1990: 40). The second example to illustrate this point is that of EFM (External Fetal Monitoring).

Again, the collusion of women with physicians in terms of their belief in and reliance on technology points to the fact that as a society, we embrace technological innovation. In her interviews with pregnant women Anne Quéniart (1988) found that women want to be "normal". They want to have an experience of pregnancy that is similar to other women they know. In addition, the women she interviewed were concerned that the fetus be "normal": i.e. neither malformed nor handicapped in any way. Women receive much, if not most, of their information concerning pregnancy from their own physicians who, in turn, make decisions in accordance with a value structure reflective of existing social biases particularly the bias towards medical management (Sherwin, 1989). Women feel, and/or are made to feel, responsible for abnormal experiences and the impetus falls on them to arm themselves--to use technology for the benefit of their fetus (Quéniart,1987). A woman who does not "choose" to use technology is confronted with the "danger of death and disability...as the price to be paid" (Oakley, 1987) The extensive use of routine External Fetal Monitoring(EFM) also bears witness to this phenomenon. Shy et al (1990) report that comparing EFM to auscultation does not result in improved neurological development in children born prematurely and that 20% of children whose mothers had EFM were diagnosed with cerebral palsy compared to only 8% whose mothers has auscultation. Freeman (1990) also found that there were no benefits of EFM compared to auscultation although there was a higher c-section rate for EFM but he advocates that

routine EFM be continued because it is good medical practice from the medical-legal standpoint vis a vis medical liability. Burtch (1994) contends that physicians have quite simply lost the ability to use hands-on low tech treatments, a view that is repeated in calls for the reintegration of midwifery in our health care system

The examples of sonography and EFM demonstrate how a reliance on technological innovation can arise and how such reliance may lead to the usurpation of traditional, less high tech skills. These examples also show that this change is a complex one and cannot be reduced to simplistic explanations relying solely on one point in the politico-economic-social trilogy. Rather, we, as a complex society engender such changes. Again, once technology exists physicians feel compelled to use it and patients begin to demand it. The key issue is to ensure that technological innovations that reach the public have been properly evaluated, and have been proven to be of benefit before they become standard of care.

2.3 (4) Improving our health

Perhaps one of the most universally believed untruths concerning the role of technology in medicine is that improved technology automatically leads to vast improvements in the health of individuals. The public supports technological solutions based on their understanding of the greater benefit these solutions offer. Yet while technology has transformed medicine and contributed to it's spiraling costs, it offers no real improvement for health in the overall population (Rachlis & Kushner, 1989). This occurs because even in this modern age, in a society as well-off as Canada, the most

fundamental "cause" of disease and ill health is poverty (Tesh, 1988). Thus, poor people are more likely to get ill, to remain ill longer, and to die sooner (younger) of their illness (Rachlis & Kusher, 1989). The association of poverty with illness stems from poor housing conditions, inferior nutrition, inferior education. Indeed, the drop in the death rate over the past century can be attributed to improvements in nutrition, living standards, and sanitation and not to an increase in technological innovation (Dutton, 1988). In fact, "...medicine's contribution to the health of the population as a whole is really rather small in comparison to the role of social and environmental conditions" (Dutton, 1988: 4). The key to further improvements in health lies than in socio-political-economic solutions rather than an increasing reliance on technological ones (Tesh, 1988). Or as Louise Vandelac (1986) has pointed out, "La trilogie science-économie-technique englobe l'ensemble du social"; it is part and parcel of the same phenomenon. Nevertheless, it is this kind of low-tech social solution that is missing from our health care agenda. Tesh makes the claim that one part of the problem with the way we approach health care and formulate our health care policies is that our societal belief in the individual permeates our decision making. Therefore, because we believe in individualism, our health care policies hold individualism as a fundamental tenet and proceed from this point. Thus by placing emphasis on the individual, the need for social change can be ignored. Hence no real structural changes are called for but rather changes are advocated at the individual level.

The ideology of science as neutral has a specific historical development and the questioning of that neutrality began in the late 1960s and early 1970s with a critical examination of the uses and abuses of science . This was considered at the time a radical shift in which science could no longer be understood simply as the discovery of reality. With the publication of *The Structure of Scientific Revolutions* (1970), Thomas Kuhn launched a new field of study known as the sociology of scientific knowledge. He asserted that scientific knowledge, like all other forms of knowledge is produced and shaped by social interests and is, therefore, affected at the most profound level by the society in which it is conducted (Wajcman, 1991). Tesh (1988) agrees, asserting that although science continues to maintain that it is essentially value free it is impossible to justify this fallacy. We should not attempt to maintain this justification; rather the values that are inherent in science should be brought out into the open as an integral part of health care policies. It is important to note that not all scientist share the same values much like not all sociologist share the same values. However, the subjectivity of each individual is value laden which is to say each individual has values, morals and norms that guide them be they secular or religious.

Our focus on the individual and our political unwillingness to effect meaningful social change provides a partial explanation for the present state of health care. But to understand more deeply the use of technology, it is important to understand the meaning of technology for individuals within and without the health care profession.

2.3 (5) The dilemma of modern medicine

From the point of view of medical technology, the dilemma of modern medicine is defined as ensuring that new innovations respond to and reflect the interest of society. This is not the same focus that social scientists have in relation to medical innovations as will be discussed throughout this thesis. Tesh (1988) has pointed out that science is not value free, rather science is fraught with value judgments. It is not possible to conduct research without casting an eye toward what uses said research will be put to. Increasingly in the latter part of this century, research is becoming more and more profit oriented. This is aptly illustrated by the influx of industrial dollars into the previously aloof realm of university research facilities. As Dutton (1988) sketches the scenario, industry-university collaboration embodies a profound difference in their "aims, methods, and values". Industry is in the research business to make money whereas universities have traditionally been interested in research for the pursuit of knowledge--a pursuit that necessitates and demands openness and a sharing of information. Industry does not share information because information is money; secrecy is necessary to protect their investment and (future) profits. The collaboration between industry and university has arisen partly because of the inability of the government and private investors to meet the funding needs of research projects. Consequently, this union has resulted in the fact that "scientific knowledge is no longer considered a collective good but, rather, private property that can be owned and traded for profit" (Dutton, 1988: 209). This perceived

ownership of knowledge threatens the basic norms and values of science.

The main aim of biotechnology companies is to turn a profit. Therefore they choose projects based on their ability to provide gain in the shortest term possible and not on the basis of their social value or need. Therefore the needs of the poor will continue to be neglected. Again, the high tech, high cost solution is preferably to business that the low tech, low cost, social change solution.

2.3 (6) Geneticization and Medical Innovation

Geneticization is a word coined by Abby Lippman (1991) to refer to:

> an ongoing process by which differences between individuals are reduced to their DNA codes, with most disorders, behaviors and physiological variations defined, at least in part, as genetic in origin. It refers as well to the process by which interventions employing genetic technologies are adopted to manage problems of health. Through this process, human biology is incorrectly equated with human genetics, implying that the latter acts alone to make us each the organism she or he is".

The trend toward geneticization of health problems and social problems is worrying. In a social state of flux such as we are experiencing and the apparent rise in social problems, pointing to a genetic root to problems is appealing because it gives the legitimation of scientific inquiry; it is objective, controlled and technical. Thus all aspects of our social and biological lives are geneticized: "biological reductionism is part of the pervasive tendency to medicalize social problems" (Nelkin & Tancredi, 1989: 10). Karen Messing (1993) agrees, warning that by "biologizing"

social and political issues we "depoliticize" them: this depoliticization leads to an individuation of problems. Individuation, according to Turner (1983) is "a set of practices by which individuals are identified and separated by marks, numbers, signs and codes which are derived from knowledge of the population and related to the establishment of norms" (cited in Shilling, 1993: 78). The establishment of norms is itself formed by what Michel Foucault (1977) called the "normalizing gaze"; a crucial aspect of his theory of power and control. For Foucault, discipline incorporates surveillance which in turn subsumes the idea of the gaze. He writes: "the judges of normality are present everywhere. We are a society of the teacher-judge, the doctor-judge, the educator-judge, the social worker-judge" (1977: 183). In terms of genetics, discipline has become *self-discipline*: discipline is not bound to any particular institution and this makes actions appear to be voluntary since the anonymity of power leads to its invisibility. Therefore, individuals become "just as surely as the inmate of the Panopticon, a self-policing subject" (Bartky, 1988: 81).

By urging individuals to become self-policing we are urged to take individual responsibility for our bodies (our health-care) at a time when the global threats to our health are increasing. As a fall-out from this, we are made to feel personally responsible for ill health (Shilling, 1993).

As Nelkin and Tancredi (1989) make clear, diagnostics have found their way into all areas of our lives with alarming results. They show how genetic information is used to establish norms for individuals and how the norms and the techniques take precedence

over any individual person. Thus people become subject to the test rather than the test serving individuals. This translates into workplace policy that puts the impetus on the worker to opt out of a work environment that may be hazardous rather than on the employer to clean up the workplace for all workers (Holtzman, 1989). This is also true of workplace "fetal-protection" policies which are aimed at excluding women of childbearing age from jobs involving exposure to toxins or situations dangerous to a developing fetus (Gonen, 1993). Genes are seen as the root of everything whereas other factors, especially environmental ones, are ignored.

Media reports are rarely able to present an issue in context but rather extract simplified explanations from complex phenomena such that most lay persons receive an extremely simplified understanding of the role of genetics in our lives. The language used by media relate that scientists have discovered the gene "for" something. In the past few years headlines have heralded the discovery of the genes for a variety of ailments such as fragile X syndrome, Canavan, Waardenburg's Syndrome (deafness), myotonic dystrophy, ALS (Lou Gehrig's disease). In addition, the gene has been "discovered" that "causes" colon cancer and cancer of the uterus, stomach, ovaries, small intestine, gall bladder, kidney, ureter, and breasts. These "cancer causing" genes don't cause cancer but the tone and style of media reports posit a strict causal relationship. Other human problems have also been given a genetic link, such as alcoholism, schizophrenia and manic depression. Genetics is only partially responsible for these mental illnesses however, with environment, upbringing and experiences all having a strong role to play. As a

consequence psychiatric ailments that afflict the very complicated brain are not as amenable to DNA analysis as other disorders such as Cystic Fibrosis are (Angier, 1993). A print media series in the *Montreal Gazette* on the "Biology of Violence" placed the origin of aggressive behaviour on genetics even while admitting that any genetic component would have to be combined with a deprived or violent environment to be manifested: violence then is not in our genes but rather is a direct response to and result of social problems (Kotulak, 1993). The nature of media coverage of genetic breakthroughs is important because most lay individuals receive their information about genetics and technology through media and not through more complex and thorough scientific journals. Therefore, the intricacies of what genetics can and cannot tell us about our predisposition to particular diseases or disorders are lost in the language of discovering the gene "for" something.

Geneticization produces simplistic explanation for complicated phenomena. As Hubbard & Wald state:

> "Social and economic circumstances affect our body states and also shape the ways we perceive and categorize them. Biology cannot be separated from social and economic realities, because they are intertwined in complex ways and build upon each other. We cannot isolate the biological factors, and when we try we oversimplify and distort reality" (1993: 58).

Often at the forefront of the crusade for genetic explanation are the economic concerns of interested parties.

As stated above, the business of biotechnology companies is to make a profit. Given the proliferation of biotech companies in the past two decades, and the huge amounts of money being put into this

market, critics have expressed concern over the prioritizing of financial gain (Hamilton, 1994). Companies with names like "Biogen", "Amgen" and "Genentech" have profits in the hundreds of millions of dollars (Hamilton, 1993). The profit motive can be worrying if concerns of the individual such as insurance, health care and stigmatization, take a back seat to the business aspect of technology.

Neil Holtzman, in his book *Proceed with caution*, highlights that not only must individual freedom be guarded with regard to genetic testing, but as a society we must be on guard against the misuse of such testing. In addition, society has a collective duty to ensure that health care is available to all. This means that as a society, decisions must be made regarding health care. These decisions must incorporate an understanding of fiscal responsibility as well as the meaning and nature of health and the place that the pursuit of health should have in our lives (Callahan, 1990). "Ideally, the new genetic technologies would be directed toward trying to eradicate critical social problems such as world hunger, epidemic diseases and environmental pollution. However, for many of these problems, the limiting factors are social and economic, not technological" (Dutton, 1988: 217). This returns us to the arguments made above, principally that medical innovation has in a sense reached a saturation point with regards to technology's ability to ensure greater health care benefits to the majority of people. Thus, any further push toward technological innovation can be seen as a push for profit and not one aimed purely at ensuring greater health and social standards for individuals. The health care budget is a finite one and essentially the question in regards to medical innovation and

the proliferation of technology in the health care system comes down to the very simple and yet, at the same time, the most complex question of all, "Who decides?". However, as Diana Dutton elucidates, that may not be the right question to be asking.

2.3 (7) Diana Dutton: *Worse than the disease*

Diana Dutton has written an enlightening book on the subject of the social aspects of medical innovation. Her work helps to provide boundaries and justification for the research undertaken herein. For this reason, an expanded summary of *Worse than the disease; Pitfalls of Medical Progress* is presented.

Dutton's primary thesis is that medical innovation and technologies impact upon society as a whole and therefore must respond to the needs of society. Dutton acknowledges that society is not homogenous; the needs of different groups are not always the same, nor are they always compatible or complementary. Noting that confidence in medicine by the general public has fallen drastically, Dutton points out that rarely is the public asked to give opinions regarding which medical innovations are wanted or needed even though the public is expected to be consumers of and to benefit by those innovations. One source of the problem is the emphasis on scientific and economic considerations to the detriment of social and moral ones. Decisions about medical care and innovation will "influence who will live and who will die. They are, ultimately, society's alone to make"(p.5). Collective decision making is lacking from most medical processes. In presenting four case studies from which lessons can be drawn, Dutton noted that one common feature of all four was of the "dominant role of technical and scientific

experts in decision making, even on issues with important social or ethical components." (p.9) In fact, social or ethical issues were never viewed as an integral part of any decision making, but in each instance were viewed as add-ons or extra bits. This separation of science from ethics, of technology from the uses of that technology is a central issue of concern.

Four case studies are presented from which to analyze the process of innovation and decision making: DES, a drug used to prevent miscarriage; the artificial heart; the swine flu immunization program; and genetic engineering. These four cases share some common characteristics as well as offer some divergent issues of medical innovation. Dutton's investigation focuses on the social actors involved in the events depicted, and on the decision making processes, including decisions and "nondecisions". She demonstrates that decisions taken are not inevitable but rather are the result of particular instances in history converging with particular individual personalities and needs. What emerges is an image of how decisions which affect all society can be influenced by individual personalities; decision making by the very few for the very many.

Medical research and care are "typically massive enterprises involving high-cost capital equipment and skilled personnel" which "constitute a major public financial investment" (p.14). Health is no longer centred on the individual but is rather an international enterprise with huge economic impact. Ethical considerations must compete with economic ones. As government control cedes to industrial control, economics is driving the car and ethics is but the back seat driver. However, ethical considerations do not centre

solely on whether particular innovations *should* exist but also pose questions about how those innovations should be used; in cases of economic disparity is it fair to fund medical innovations that will only be available to the wealthy, either the wealthy in a particular country or to a country which is wealthy as a whole? Increasingly, the car is speeding away before these ethical issues are discussed, leaving concerned groups and individuals hanging on tightly and cautioning, slow down, slow down.

At the heart of some of these differences is a very different world view of participants. The mechanistic view of the human being as a bunch of parts, mechanical and chemical is at the heart of medical research. This contradicts the more esoteric view of the human being as something not necessarily definable, as not just made of parts, but of something more lofty including a soul and an essence of humanity.

Economic conditions are a driving force behind much of medical research and in the case of genetic engineering, are the prime source of consternation of critics. Technological innovation is used to bolster the economy as well as to provide optimum health care. This dual role is the source of some tension when either the economy or health must be prioritized.

> "When health and economic goals are merged, these various conflicts are inevitably obscured. More importantly, the fundamental purpose of medical and scientific innovation becomes confused. Economic considerations begin to shape health priorities, even though the types of innovations needed to stimulate the economy may *not*, in many cases, be those that would yield the greatest benefits to health" (p. 25 emphasis in original).

The question posed by Dutton of "Who shall decide" appears to already have been answered. She notes that the public has little or no voice in shaping the direction of medical innovation and that projects that may be socially worthwhile are not undertaken if there is no economic gain to be had. In addition, even those shaping the future may deny their own role in the process, "Some academic scientists explicitly disavow responsibility for the practical implications of their work" (p.26). The analogy is often made with scientists who worked on the technology of the atom bomb. They provided the technology but denied any responsibility for the deaths caused by it. The separation of science and ethical responsibility is thus complete.

2.3.(8) Four Case Studies

1. *DES or deithylstilbestrol* is a drug created in 1938 as a synthesis of natural estrogen and a nonsteroidal compound. Initially, the news of this innovation provoked excitement because it was felt that DES could be used for a myriad of ailments related to imbalances in sex hormones, imbalances that previously had been treated by natural estrogens which were scarce and expensive. From these beginnings, Dutton outlines the history of a drug as a tale of colossal failure of drug safety and the use of an unwitting clientele as experimental cases. The thirty odd year history of DES as presented by Dutton, presents an interesting look at the ineffectiveness of regulatory agencies in the face of medical and technological innovation. Dutton also shows how criticisms of the drug were brushed aside in favour of the new wonder drug. This discounting of

critical viewpoints will prove to be a common theme in all four case studies.

The FDA gave approval for DES to be used for treatment of four indications with a maximum approved dosage of 5 milligrams (mg): menopausal symptoms, gonorrheal vaginitis, senile vaginitis and suppression of lactation after childbirth. Nevertheless, by the early forties, the drug was being used for nearly every "female" problem possible (and was also used to treat prostate cancer in men) at doses considerably higher than those recommended.

The decision to use DES in pregnancy was pioneered by Dr. Karl J. Karnacky of Houston. Karnacky used dogs for his initial experiments but stopped because the dogs were dying quickly. He was convinced by the drug company, E. R. Squibb and Sons, to continue his experiments on women and was given free samples to do so. Doses given ranged from 25 mg up to 6,000 mg (and by 1947, Karnacky was giving some women up to 24, 050 mg) and became standard of care in the hospital Karnacky was affiliated with. The FDA had not given approval for the drug to be used for pregnant women. There had been no Random Controlled Trials for use of the drug in pregnancy and doctors admitted they didn't know why the drug worked, just that it did work. When the FDA finally did give approval for the drug to be used in pregnancy, there were already indications of serious side effects and problems with the way the drug was being used, including questions about the effect of DES on the fetus and links of the drug to cancer. Regardless, the FDA approved the new indications for the drug and in doing so "authorized what amounted to mass experimentation on pregnant

women-the sanctioned use of a drug with known risks whose effectiveness had not yet been fully proven" (p.54). When clinical trials were finally undertaken, it was revealed that DES was not an effective treatment for problems associated with pregnancy. Despite these controlled experiments, many doctors continued to prescribed the drug.

Dutton offers several reasons for the continued use of DES. Firstly, many physicians felt that their own experience contradicted the controlled experiments. Secondly, some physicians felt that they must support their colleagues who were standing by the drug. Thirdly, drug company "detail men", or salesmen actively pushed the drug and drug company literature downplayed adverse outcomes. Fourth, women had come to know of the drug through media reports and put pressure on their physicians to prescribe it. Fifth, the FDA failed to provide leadership by removing pregnancy from its' list of indications for DES nor did it force drug companies to send out letters to physicians correcting previous misinformation.

Although studies indicating negative conclusions about DES began to surface in 1952, the FDA did not change it's indication for the drug until 1972 when doctors began to report cases of clear-cell vaginal adenocarcinoma in daughters of women who had received DES in pregnancy. This "represented the first known human occurrence of transplancental carcinogenesis-the development of cancer in offspring due to exposure in utero to a substance that crossed the mother's placenta" (p.32).

When the links between cancer and DES were found DES was already being used as a post-coital contraceptive, again without any

clinical studies to justify it's validity. However, the FDA was stonewalled in 1976 when it sought to approve an application for DES as a morning after pill. Opposition came from within the FDA itself, interestingly enough, when the Director of the Generic Drug Division and his medical officer, Dr. Marvin Seife and Dr. Vincent Karusaitis refused to approve of the drug. However, DES continued to be prescribed as late as 1985 up to 15% of college and university health services in the United States were still advocating DES to women.

The story of DES continues to unfold as affected women and their daughters seek compensation from drug companies for their use as human guinea pigs without their knowledge or consent. Problems continue to multiply with higher levels of breast cancer in women who took DES, a variety of complications in their daughters including cancer, troubles conceiving and structural abnormalities of the cervix or vagina, and structural abnormalities of the genital tract in their sons.

The experience of DES shows what can happen when regulatory agencies do not carry out their jobs and when the public is dependent on the advice of experts to give treatment and information. The story of DES "reveals a regulatory system that functioned less as public protector than as a willing and cheerful ally in the pursuit of corporate profits through technological development" (p. 89) This same chain of events is seen in the example of the development of the artificial heart.

2. The origins of the *artificial heart* was a time in which confidence in medical innovation and technology was very high. The mid sixties were a time of great achievement in medicine with

kidney dialysis and transplant, heart pacemakers, open-heart surgery and artificial heart valves. In the midst of this optimism the artificial heart program was approved in 1964. It was to proceed in four phases with the final phase in 1970 resulting in the mass production, installation and maintenance of artificial hearts. Similar to the case of DES, initial euphoria and expectations for the innovation quickly gave way to a more realistic analysis of the problems involved. But again like the case of DES, the program continued in spite of these problems although by 1968 the program had stalled, primarily due to technical problems.

Despite the seeming setback to the official artificial heart program on April 4, 1969, Dr. Denton Cooley implanted an artificial heart into Mr. Haskell Karp. Dr. Cooley performed the operation without ever asking for or receiving approval for the experimentation on a human subject. This disregard for ethical review stunned the research committee as there was no reasonable expectation of scientific advancement nor was there any reasonable expectation of benefit for the patient. Mr. Karp underwent a second operation on 7 April in which the artificial heart was replaced with a transplanted heart but he died the following day.

Dr. Cooley's defiance of ethical norms and the norms of scientific behaviour cast a pall over the entire artificial heart program. While experimentation was to proceed, the Special Advisory Committee "urged that future planning should include psychological, social, ethical, legal, and religious concerns, in addition to medical issues" (p. 104) and when an Artificial Heart Assessment Panel was established in 1972 to make recommendations it consisted

of "two lawyers, two economists, three physicians (one a psychiatrist), a sociologist, a political scientist, and a priest-ethicist" (p. 105). Nevertheless, the goal of the panel was not to discuss if plans should go ahead for work on the artificial heart but rather to make recommendations for it's use given that it would become a reality in the near future. However, the panel itself broadened it's goals to include issues related to quality of life for artificial heart recipients, violations of social justice which would occur if only the very rich could afford to have transplants (at a cost of 15,000-25,000 1973 dollars), and dared to ask the ultimate question:" Should the artificial heart be developed?" especially when so many individuals lack even basic health care.

These issues were soon of no importance as the artificial heart program funded by the government was usurped by a private group at the University of Utah. It was this group, under the leadership of Dr. William DeVries, who implanted an artificial heart in Barney Clark.

> The breakaway of the Utah heart is a case study of how personal and institutional incentives, politics, and high-level finances can influence the selection of medical technology. It is also the story of how, under the loosely knit American system of federal assistance to private medical researchers, one group of investigators was able to exploit the federal program to its own particular ends. It is the story of the uneasy marriage of commercialism with established scientific methods and governmental safeguards for the introduction of new and unproven medical technologies (p. 109).

The heart that was implanted was developed by Robert Jarvik and his mentor, Dr. Willem Kolff.. These two individuals nearly single

handedly highjacked the artificial heart program and in doing so left an indelible mark on the economic aspects of the technology. Through shrewd investment and corporate know how and sleek marketing, the principal players made their fortunes with the artificial heart program. Dutton characterizes the case of Barney Clark "as much an experiment in the use of the public media to market a medical technology as it was a scientific experiment". The media was used to influence public acceptance of what was still an unproven and experimental procedure, a procedure that many felt had not yet been proven to be wanted or needed by the population. The central concern is whether small groups of individuals with an economic investment should be allowed to create policy for the rest of society, a society that will have to live with the consequences.

3. Like the two case studies outlined above, *the swine flu immunization program* provides lessons in the way pivotal decisions are made to proceed with a particular course of action and how difficult it may be to stop machinations once they are in place.

The swine flu of 1976 was believed to be similar to the pandemic of 1918-19 which killed over 20 million people worldwide, including 500,000 Americans. When the virus was believed to be located at an army base in New Jersey, health officials began to plot a course toward dealing with what they anticipated would be a public health care crisis.

The United States response to fears of a repeat of the swine flu was to begin a program to inoculate every man, woman and child against the disease before the end of the 1976 calendar year. This monumental undertaking was the direct result of decisions made by

David Sencer, Director of CDC (Center for Disease Control) and his colleagues. In order to counteract criticism and to bring other players on board, the risk of an epidemic of swine flu was consistently overstated such that whereas the original case in New Jersey which indicated that an epidemic was unlikely (odds were given of 2-20 percent of occurring) was quickly sublimated by reports that there was a "strong possibility" an epidemic would occur to the certainty that it would occur "there *will* be a major flu epidemic" (p. 139, emphasis in original). When the President of the United States, Gerald Ford, came on board, it was a definite go. The goal was to immunize every citizen of the United States by the fall of 1976.

The immunization program was beset by problems including the inability of drug companies to manufacture enough of the drug in the time frame available. Thus even when personnel was mobilized in communities to ensure turn-out by citizens, the immunization clinics often had to be postponed when the drug was not available. Added to that was the general disinterest of the population and the decision not to inoculate children because they would need two sets of inoculations. In the short time span in which this scenario was played out, it became known to the public that there were two areas of uncertainty. First there was disagreement over the necessity of the inoculation program and secondly a general belief (aided by the media) was that adverse reactions to the inoculation had been downplayed by medical personnel. Adverse reactions ranged from the relatively innocuous such as sore arms and fever, to the association of swine flu vaccine with Guillain-Barré syndrome. This

negative publicity was highlighted by the inability of the government to find an insurance company to cover liability, in the end, the government itself had to assume liability for the drug's use. By the time the program got seriously underway, doubts from many physicians and public health officials, legal and logistical uncertainties and the media coverage had already combined to indicate that the program was not needed. Regardless, the machinery proceeded forward. Altogether more than 45 million people were immunized, less than one quarter the total population. Had the swine flu epidemic materialized the ambitious program would have been deemed a failure. As it happened, there was no swine flu and the immunization program was still a failure.

One of the biggest sources of it's failure was the inability of the social actors to agree on a course of action and once a course of action had been decided upon, the inability of individuals to recognize that the wrong decision had been made. Dutton highlights why things went so wrong with the swine flu immunization program.

The main source of problems was that planning was based on overly optimistic assumptions: the vaccine would be effective, there would be few side effects, production would be possible, there would be no liability problems etc. A worse case scenario would probably provide a more realistic way to plan, but this optimism was not checked for three reasons: 1. the substantial influence of medical scientists in aspects of the program that fell outside their expertise, 2. the role of the president. Once the president was behind the idea, it became difficult to opt out, 3. lack of public input. Dutton charges this third reason as the most important source of error. The refusal

of experts to debate publicly on the merits of the program indicated that decision making was suspect because they all had something to gain from going ahead with the immunization campaign. The failure of the swine flu immunization program "highlights the danger of relying too heavily on the views of technical experts, whose unswerving confidence in the safety and efficacy of medical intervention (even in the name of prevention) seemed to blind them to impending problems, and of insulating national health policy from public scrutiny" (p. 173).

4. *Genetic engineering* provides a case study in which ethics is consistently placed outside the boundaries of scientific research. The development of techniques and the application of the techniques are not viewed by researchers as part of the same process. Dutton demonstrates how researchers consistently flouted government regulations regarding genetic engineering research when those regulations infringed upon their research needs. The scientific community involved in the decision making process of the early seventies regarding research safeguards overwhelmingly favoured self regulation and they continually "were opposed to the idea of sharing control over science policy with lay citizens" (p.183). Researchers who did try to draw the attention of their colleagues to problems and concerns with research being undertaken were usually of the opinion that their concerns were not taken seriously.

Recombinant DNA research is important to everyone in society because "society is the patient" and as such is entitled to informed consent for any procedure being done. However, the problems that society needs to discuss are not problems of technical ability but

rather moral, religious and ethical issues. These issues need time to sort through, there can be no quick fix. The dizzying pace of science left social issues behind, and while lay citizens were still discussing whether techniques were necessary, they were already being perfected in the research labs. The main problem was that scientists refused to acknowledge the larger context of their research; that the ability to do something is acted out in a social context and not just in a laboratory.

The underlying question with regards to genetics is "who has the right to control this powerful new technology which will affect all of our lives?" (p. 224). Genetic research poses individual risks and a collective risk to society as a whole and we are as yet unsure of what exactly those collective risks may turn out to be. One risk that has been discussed is the danger of genetic engineering compromising the integrity of the gene pool. "By eliminating 'defective' genes, the new technology might gradually reduce genetic diversity" (p. 242). Because our knowledge of the functioning of genes is still quite primitive, research is being done with only a partial understanding, too little understanding to allow us to predict what may happen when we manipulate genes.

In addition genetic manipulation "poses a variety of risks to traditional social and moral values" which is why many religious orders are wary of such research and have consistently called for a moratorium on 'playing god' (p. 218). Genetic research, more so than any other medically innovative technology, allows scientists to manipulate the very essence of what makes us human and the collective risks to society are very real.

The economic benefits to be gained by genetic engineering are also a source of criticism. The commercial applications of genetic engineering could result in substantial economic benefit and these opportunities have resulted in a "molecular goldrush". The commercial future of biotech companies was ensured when the U.S. Supreme Court ruled that genetically altered life forms could be patented. This has resulted in the ownership of knowledge and human genes by particular companies. This decision has profoundly altered the way in which research has traditionally been conducted. Many scientists themselves object to the commodification of the very essence of human life on two grounds; firstly, no one should be able to exploit the human gene pool for economic gain and secondly, no one should be able to own such an important resource.

This ability to own knowledge has altered the ties between industry and university. Biotech industries now fund much of the university research into genetics and one result has been an increase in trade secrecy. Knowledge is no longer something to be shared as a professional courtesy between scientists but rather something to be hoarded until money can be made. The idea of secrecy goes against one of the principle tenets of scientific research and it also weakens the ability of the public to have a voice in what research will be done. Industry -university ties also mean that production becomes more important than the pursuit of knowledge. Industry needs products to sell and they are more likely to pursue profitable enterprises than they are to pursue long term projects with social worth but little economic potential. Thus it is unlikely that genetic technologies would be used to ease the world's social problems or to

cure world hunger or epidemic diseases, especially in poorer countries. Dutton quotes the head of Schering-Plough, a major pharmaceutical company: "Schering-Plough is not in business to do research, it's in research to do business" (p. 208).

Dutton feels that biotechnology needs greater input from the public than any other industry for three reasons. 1. The basic developments of biotechnology emerged from federal funding, 2. Many aspects of biotechnology will not be pursued by private industry because it is not profitable to do so and therefore public interests will have to step in, 3. This research carries tremendous risks and responsibilities for all of society and therefore public priorities should be paramount.

Diana Dutton's analysis of four case studies in medical innovation highlight important issues. From these cases we can glean some important lessons. The most important of these lessons is that of public participation

It is vital that the public have a role in decision making regarding medical innovation. At the very least, doctrines of informed consent should be strictly adhered to so that individuals can make decisions based on what knowledge is available. Paternalistic policies enacted for the benefit of the public at the behest of the expert have to part in medical decision making today. In addition, the public has a vested interest in being able to influence the decisions that are made on issues affecting them if only tangentially. This means that biotechnological developments should be subject to the scrutiny of the public not only scientists. Science

exists within a particular social context and must acknowledge that society may not be enamored of science for the sake of science.

Conclusion

There is no doubt that medicine in the twentieth century has been transformed by technology. The use of technology in the field of medicine is so prevalent that technology and medicine have come to be seen as conjoined; medicine is the use of technology to combat the frailties of the human body. Yet this reliance has not been without both economic and social costs. The reliance on technology is made possible *because* as a society we value technological intervention. In addition, the use of technology to solve medico-social ills complements an individualist approach to the causes and hence the cures of the human body. Nowhere is this more evident in this last part of the twentieth century than in the realms of reproductive technologies and genetic research. These two burgeoning areas of research are the front-line of technological advancement and as such, we must pay careful attention to the lesson to be garnered from the choices that we, as a society, are making. Rapid progress in these areas means that it is difficult to have the benefit of hindsight yet only by having a clear understanding of the role of medical technology both positive and negative, can we make a clear path toward choices that will benefit us all.

Medical innovation is a social issue because medical techniques and technology are paid for, either directly or indirectly, by individuals who form our society and they are used on or by those same individuals. However, few individuals get a say in what

technologies are made available and what constitutes a social health care need.

The main determinant of health is social class. This is true both of Canadian society which has a universal health care system and the United States where millions of individuals are without any health care coverage (Callahan, 1990). The class based difference arises not because of differential health care coverage but more due to other aspects of health and well-being; level of nutrition, housing standards, access to education and type of employment (or lack of employment). These crucial discrepancies cannot be solved through medical innovation and the use of technology. However, the money spent on technology may be of more help were it channeled to other, more socially relevant changes such as mentioned above: measures to combat poverty, adequate housing, more education and better living standards in general.

Once technology becomes available, there is an impetus to use it, even if it's efficacy has not been proven, or even if, as the twin examples of sonography and EFM highlight, they are proven to not be valuable tools. The adhesion to a technological solution is one result of our collective mind set that prioritizes science over experience and sees value in a production based model of society. Thus, it would be simplistic to state that physicians rely overtly on technology without also acknowledging that the individual patient frequently pushes for a high-tech, do-something, approach. Technological innovation has to proceed at a slower pace. This slower pace would allow for more time to examine the impact of technologies on the individual and his/her state of health and also the impact on society as a whole.

Genetics is a relatively new area of research but one that has made impressive progress in a short time. This area of research has attracted substantial debate regarding the values inherent in such research as well as regarding which values we wish to promote as a society. Society must make choices about such abstract and intangible ideas. Given that there are collective risks there perhaps should be collective decision making. The problem is that although we all share in the risk, we do not all share the same values. Compromises must be made on all sides by all interested parties. This requires more than just simple intent. It requires a moral force and a political will to ensure standards are met and kept.

Chapter 3

Methodology

The choice of method for any research project is firmly rooted in the nature of the question that is to be explored. The research herein was envisioned from the outset as exploratory in nature and as such several avenues of information gathering were deemed essential.

The research project examined social changes arising from medical innovation: in particular, social changes related to and stemming from the medical technological ability to screen any individual in a population for carrier status of the Cystic Fibrosis gene. Because the medical community was making a space for itself, the research project was taking place in "real time"; that is, it was not intended as a historical account of what happened and why, but rather an ongoing account of what was happening at a particular junction of time, space and place. Given constraints of time with regards to the actual writing of an academic dissertation, at the final stages of writing , the events in question are necessarily in the near past.

This chapter outlines the methods used for data collection. Triangulation was used to compensate for any methodological weaknesses that may have arisen given the use of only one method. Triangulation is the use of multiple techniques for data collection (Sommer & Sommer, 1997). The focus of data collection rested on a qualitative approach. This choice was compatible with the fact that the research was not testing formulated hypotheses, but rather was

guided by research objectives developed through a review of the relevant literature. Qualitative research designs use systematic observation and focus on the meanings people give to their social actions (Palys, 1997). Three different qualitative research methods were engaged, with a different emphasis given to each. The three methods are 1. Case study; 2. Interviews; 3. Archival (Historical) Research.

3.1 The case study

Case studies involve intensive observation of a particular person, group or event. This intensive observation allows the researcher to maximize information gathered which then enables the formulation of general theoretical and abstract research conclusions (Sommer & Sommer, 1997). The case study used for this research project allowed for the elaboration of the macro social processes that are ongoing within a particular community.

Yin discusses the case study as a means of enlightenment:

> "case studies...are generalizable to theoretical propositions and not to populations or universes [and] the investigator's goal is to expand and generalize theories (analytic generalization) and not to enumerate frequencies (statistical generalization)" (1984: 23).

Given the closed nature of the "community" that was the focus of this research-- a research community defined (by the author) and bounded by it's interest in and work on one particular gene, it is apparent that attempts at generalization would be inadequate and inappropriate. The respondents themselves, while acknowledging the "small world" of research, did not always readily identify themselves as a community. They did, however, identify that

individuals working in the field of CF in varied capacities knew one another by name and/or by reputation. In addition, respondents were aware of the role others played in the definition of and the process of CF programs related to screening and testing for the CF gene. The case study method is particularly helpful when examining rare or unusual events and as such can lead to the formulation of testable hypotheses at a later point in time (Cozby, 1997). The essence of the research was an in-depth understanding of processes. Schramm (1971) elucidates that

> "the essence of a case study...is that it tries to illuminate a decision or set of decisions: why they were taken, how they were implemented, and with what result" (Quoted in Yin, 1984: 24).

The case study approach, therefore, is ideal for a situation such as encountered with the new information regarding genetics and CF. For this research, the "case" being studied was an event: the discovery of the CF gene and the subsequent technological ability to begin genetic screening.

3.2 Interviews

In some instances, interviews are conducted as a form of survey in which the respondents are read a set of prepared questions and their responses recorded on a prepared response sheet. However, for this research *ethnographic* interviews were conducted. James Spradley in *The Ethnographic Interview* points out that "Rather than studying people, ethnography means learning from people" (1979: 3). An ethnographic interview is one in which the researcher attempts to learn as much as possible about the respondents and their behaviour by talking to the respondents. Typically, ethnographic interviews

focus on a particular culture, or, as in this research, community and learn what people do, and what meanings their behaviours have for themselves. Ethnographic interviews depend more so than other interviews on locating experts or leaders in a particular group or community to act as informants or interpreters for acts and meanings of those acts.

3.2 (1) General categories explored in interviews

While public representation is advocated in all areas of genetic screening, individuals who have the power to affect research criterion ultimately are able to direct policy. Three separate yet linked broad areas of interest arose from the literature review:

1. *The social meaning of testing.* This encompasses a large catchment area addressing such diverse and varied issues as the ethics of testing, queries about stigmatization, the meaning of the genome project itself, the implications of PND for reproduction now and in the future.

2. *Health care* . Included in this general category are questions regarding cost and expenditures for health care which includes concerns about lab capacity. Broad understandings of what we mean by health care and more abstract evaluations of the meaning of health and illness for individuals and for our society in general form an important element of this category.

3. *Living with Cystic Fibrosis* The day to day experience of living with a disease such as CF is important to understand because it

brings a concrete reality to abstract concepts like "the disabled". The meaning that individuals bring to their lives and the ways in which they perceive and validate their own experiences is important to understand.

3.2 (2) Location of interviews

Interviews were conducted in three cities; Montreal, Ottawa, Toronto. The choice of cities was not random but rather purposive. Montreal and Toronto have the two largest populations of individuals with CF and consequently their medical facilities are also two of the largest in Canada. Interviews began in Montreal and through a "snowball technique" interviews were booked in Toronto and finally in Ottawa. There was no methodological rationale for interviewing in these particular cities, rather they were the sites of personnel conducting research and available for interview.

3.2 (3) Interviewing procedure

Establishing contacts

A list of potential interviewees was assembled of individuals known to be working in the area of CF and/or genetics. This list was made available to the researcher through *GRASP*, a research group affiliated with the Université de Montréal. *GRASP* had originally compiled the list for a research project similar to that which this researcher has undertaken. Using this list as a starting point, initial contact was made to each individual by telephone or by letter. A "snowball" technique was employed in which each interviewee was asked for the name of someone who could be interviewed. Eighty percent (sixteen of twenty) of the interviewees made suggestions as to potential respondents. These potential respondents were then

contacted with the name of the referring respondent as a "door opener". In this manner, interviewing moved from Montreal, to Toronto and then to Ottawa.

Characteristics of respondents

Limited demographic information was collected for respondents. As per agreement, respondents are identified only by professional position and gender. All respondents were university graduates. They had held their professional positions for a minimum of three years and a maximum of "more than thirty-five" years. Seven of the respondents were male and thirteen were female. Women were sole representatives of traditionally female occupations such as nursing and social work, however other occupations were gender diverse.

Position	Number and Gender
Ethicist	1 male and 1 female
Executives from Canadian Cystic Fibrosis Foundation (CCFF)	1 male
Genetic counselor	1 male and 2 female
Nurse	1 female
Physician	1 male and 1 female
Physiotherapist	1 female
Researcher in community/public health	2 male and 3 female
Scientist	1 male and 3 female
Social worker	1 female

Rationale for choice of respondents (informants)

As indicated above, informants (respondents) were representatives of a wide and diverse range of knowledge of and experience with CF. Following is a short rationale for the choice of the types selected.

1. Laboratory personnel. Today's research will shape the future and one of the research objectives was to examine the ongoing research and to explore it's meaning(s) for the future. While economic issues were not paramount herein, they were raised as an important concern in this time of shrinking health care budgets. It was necessary to look at how screening could be paid for. In addition the availability of trained laboratory personnel and the physical capacity of laboratory space to be expanded if screening programs were a reality needed to be pursued as vital elements in the research process. Do we have the skilled personnel, the space and the financial resources that would be necessary to expand lab facilities? Laboratory personnel have first hand knowledge of what strains and limits the increased demand would put on the current situation. Laboratory personnel formed part of the interview population.

2. Researchers conducting pilot screening programs. Research projects are already under way to study the best way of setting up screening programs. The social meaning of screening programs is paramount. The interest in these programs is not simply in the nuts and bolts but rather in the motivations and meanings that the researchers place on the project. What benefits do they hope to provide and to what purpose is their research aimed? Researchers

involved in pilot screening programs were also included in the sample.

3. Front line medical personnel. While technicians, geneticists and medical researchers deal only peripherally with individuals who have Cystic Fibrosis, the doctors, nurses and social workers work as a team in the CF clinic of the hospital and have a "hands on" approach to individuals. How do these people interpret the discovery of the CF gene and what meaning do they give to the proposal to put into place population screening? This series of interviews was expected to result in a better understanding of the medical and social needs of the CF patient. Is the understanding and beliefs about Cystic Fibrosis that these "team" members have different from the perception of laboratory workers and researchers? If so, what are the implications of this? Interviews with members of CF teams were an important part of the research.

4. Ethicists. Ethicists help to provide the "big picture" when examining the moral and ethical ramifications of events and processes. Criticism of screening programs invariable rely upon an ethical and/or religious foundation.

5. Researchers in public/community health. Like Ethicists, this group of informants helps to provide a macro level analysis of processes and events. These researchers concern themselves not with the individual and individual response but rather with the needs and necessities of the community at large. They help to place information into a sociological context.

Conducting the interviews

Interviews were conducted in March and April 1993. The majority of interviews (fifteen of twenty) took place in the private offices of the respondents. Individuals with whom I spoke on the telephone were on occasion unable to find time for a formal interview yet were able to give some time for a telephone "interview". Telephone respondents as well as interview respondents sent printed material to make clear their own position on the issues involved. Interviews were frequently combined with a guided tour of facilities at the instigation of respondents. For example, laboratory personnel were willing to show their lab facilities and to provide information as to how genetic samples were taken and analyzed: the researcher was provided with a tour of the physiotherapy rooms where children with CF were treated. These impromptu tours were greatly appreciated and extremely informative for the researcher.

Interviews were tape recorded for accuracy with the consent of each respondent. One interview was recorded by the researcher *and* by the respondent who stated that he had frequently been misquoted by the press and was wary of being misrepresented by this researcher. The interviews were transcribed onto computer by the researcher verbatim with some "cleaning"; that is speech patterns with many hesitations and/or fillers such as "um", "aah", "like" and "you know" were edited out unless they help to convey meaning to statements. One interview with a researcher in community and public health could not be used because background noises in the office where the interview took place and was recorded completely

obscured the voices on the tape. In some cases, follow up telephone calls were made to interviewees during the transcribing period to clarify particular remarks or to seek additional information. Permission for this follow up was obtained from respondents at the time of the initial interview. Respondents were assured of confidentiality but did not sign consent forms. They were assured that at no point in the final work would their names appear. Comments included in this work are attributed to individuals through a system wherein only the researcher can identify the name of respondents. With regards to the issue of confidentiality, several individuals gave two versions of accounts: an officially sanctioned version (an "open door" account and a private, personal version (a "closed door" account).

Non-response

Six people who were contacted by letter and by telephone either refused to be interviewed or did not respond at all. There were two refusals and four non-responses. Two individuals were out of the country on sabbatical leaves and/or working on joint ventures abroad. After three attempts at contact, non-responses were struck from the list. This was unfortunate because I was unable to learn the reasons for their desire not to be included as respondents in the research project. While it is not possible to know the motivations for refusal of some individuals, several participants did remark that there were several ongoing in-house research projects and it was likely that certain individuals had no time or no desire to participate in yet another project. Unfortunately these refusals may point to a bias in the research procedure in that individuals with particular

beliefs with regards to genetic screening for CF may have uniformly refused to respond. In addition, acceptances may also contain be an indication of those with particular biases agreeing to participate in the research. However, given the exploratory nature of the project, and the built in lack of generalizability due to non random sampling methods, this potential bias does not skew results in a serious manner. In future projects aimed at establishing a more generalizable statistical analysis, this bias would have to be overcome by a more extensive covering of the social, medical, and scientific community.

Interview schedules

As previously noted, the variety of interview respondents meant that one interview schedule could not possibly capture the wide range of knowledge gathered. Therefore, each interview was approached as a unique source, while some issues were included on all or several interview schedules, each schedule was tailor made for the particular respondent. Tailoring each schedule to the particular niche of each respondent allowed for a thorough tour of the subject. Consequently, where necessary, an in depth examination was used to procure a more detailed investigation of the subject. Composites of interview schedules are included in Appendix A.

Analyzing qualitative data

Interviews were transcribed verbatim by the interviewer in preparation for analysis. This analysis was done using the cut and paste method and not with the use of new software packages designed to analyze qualitative data. Categories of response were established by the researcher to correspond with the three broad

areas that guided the creation of the interview schedules: the social meaning of testing, health care, and living with Cystic Fibrosis. Responses were ordered on a topic by topic basis. This enabled the researcher to identify important issues that had been indicated in the literature review which may have arisen in one or two interviews. Again, because the research project was designed as an exploratory research, singular views were as important as those held by many respondents. Common and uncommon themes of interviews form the backbone of the main chapters of this thesis. The interviews in conjunction with the literature review provide an overview of the present state of genetic screening for CF in Canada today.

3.3 Archival research

Given that the phenomenon to be studied for this project was approached as a case study, it made methodological and theoretical sense to compare this particular case with two other similar instances of screening programs made available in the past. Archival research is the use of previously collected data or information to answer research questions (Cozby, 1997). Thus case study and archival research were combined through an examination of Tay Sachs screening programs and Sickle Cell anemia screening conducted in Canada and the United States. These two cases provide historical context of genetic screening programs. A comparison between Tay Sachs screening and Sickle Cell screening and between those two earlier screening programs with CF screening enlightens us to the discrepancies in the rhetoric of screening and the actual hands-on

screening as it is experienced in the community. The comparison to Sickle Cell points out many of the pitfalls of screening a community for genetic susceptibility. Tay Sachs screening has been met with little resistance and much praise, yet it is the very success of Tay Sachs screening that has engendered support for CF screening in some scientific and medical corners. Given that many respondents pointed to the success of Tay Sachs as a reason why CF screening should also be undertaken, it became essential that the Tay Sachs screening be thoroughly understood. This comparison points out many discrepancies between CF and Tay Sachs that serve as important cautionary notes.

Summary

As an exploratory research project, qualitative methods of data collection are a well established means of eliciting information. Given the unexplored nature of this research subject a triangulation method of research was chosen.

Firstly, it was decided to interrogate the subject of genetic screening for CF as a case study which involves intensive observation of a particular event.

Secondly, twenty interviews were conducted with "experts" in the field of genetics as they pertain to CF and in the field of CF in general. These interviews were conducted in Montreal, Toronto and Ottawa during March and April of 1993. The transcribed interviews provide the original data for this research project.

Thirdly, archival research was undertaken to provide a historical background and to allow for comparison between two

previous attempts at genetic screening for Tay Sachs and Sickle Cell Anemia with that of Cystic Fibrosis.

The use of multiple research methods or *triangulation*, helps to minimize any weaknesses in any one method. Triangulation also provides a vantage point for a multi-level and multidimensional analysis of social change.

Chapter 4

Historical Perspectives of Genetic Screening Programs

Cystic Fibrosis will set the new standard for screening programmes as more are made available with each new genetic discovery. Testing and screening programs being developed for CF are not the first in modern medical care. Genetic screening programs have a historical background grounded in the attempts in the past to ensure greater health and social well-being for individuals and for society. Present day programs emerge from past health care programs that sought to eradicate disease. This chapter briefly describes the genesis and history of genetic screening as an integral component of health care. Early screening programs dealt with diseases that were transmissible person-to-person. Technological breakthroughs in the nineteen sixties allowed for testing of *genetic* diseases. On the basis of these first steps, the ambitious programs for Tay Sachs and Sickle Cell Anemia were conceived: a history of these programs is the focus of this chapter.

Archival research indicates that there is a general consensus in the medical community of Tay Sachs screening programs as "good" because the programs had positive results and Sickle Cell screening programs as "bad" because the fall out from the community was very negative. Focusing on this dichotomy results in the neglect of other central issues, specifically an analysis of ethics pertaining to screening programs. An analysis of broad ethical principles removes the focal point from techniques of delivering a service and re centers

around questions pertaining to the fundamental rights and needs of human beings in relation to genetic screening. These quintessential dilemmas uncovered by Tay Sachs and Sickle Cell screening programs remain unanswered and therefore are inextricably part of CF.

4.1 (1) Early Screening Programs

Screening the population for disease began in the early part of this century. The first screening programs were attempts to control communicable diseases like malaria, syphilis and tuberculosis: diseases which spread quickly and easily through a population. This type of screening can be likened to our present day widespread vaccination programs; indeed, we routinely vaccinate children against Tuberculosis. Individuals who travel to countries where malaria is present are vaccinated before their voyage. Screening for syphilis is no longer necessary as an easy, non-intrusive method of treatment means syphilis is no longer the deadly serious disease it was at the turn of the century.

A basic criteria of early screening was the existence of an accepted treatment for the disease in question. Thus, screening for the sake of enumeration was not endorsed. Rather, individuals were urged to seek screening in order to benefit from available treatments. In addition, by seeking screening and treatment, individuals helped halt the spread of communicable diseases.

Following the second World War and a decline in the spread of communicable disease, screening began for chronic non-communicable disease, such as diabetes screening in 1947.

Concurrently, multi phasic screening was developed, allowing for several tests to be run at the same time, on the same sample. This screening was officially endorsed by the American Public Health Association in 1960.

In early screening as one type of disease began to respond to detection and treatment and therefore to diminish in the population, it was then replaced by another disease. Thus when diseases like syphilis, and TB were under control and no longer posed the serious health threat they once had, attention shifted to other non communicable diseases. In this way, the market for screening was maintained, attention simply shifted to less and less urgent cases.

4.1 (2) The history of PND in Canada

The history of PND in Canada is dated to the late 1960s with the introduction of amniocentesis to Canadian hospitals. The extent of services was limited by the lack of qualified personnel and also because there was limited awareness of the service by consumers (MacKay and Fraser, 1993: 28).

While the benefits for mass screening of communicable disease have been amply demonstrated this was not necessarily so for non communicable chronic disease and even less so for some genetic disorders. One of the most important screening criteria, as stated above, was that treatment be made available to affected individuals. This criterion was later amended to read `there must be some treatment-or at least some kind of benefit for the patient'.(my emphasis). The phrase, "Some kind of benefit" has lent itself to lose

interpretation. Over time and in different situations, it has meant many different things to different people.

Genetic screening began in earnest nearly thirty years ago, in 1964 with screening for PKU. Persons homozygous for PKU lack adequate phenylalanine hydroxylase activity. This enzyme is critical to proper metabolism of phenylalanine in the body. The condition leads to severe and irreversible intellectual deterioration. The treatment takes the form of a diet low in phenylalanine instituted shortly after birth. The diet itself poses difficulties since phenylalanine is present in just about every food. However, by closely following the diet, a child's intellectual abilities will remain in the "normal" range. The initial success of PKU screening led both the American and the Canadian governments to implement laws for mandatory newborn testing which were implemented in virtually every province and state between 1963 to about 1973. Nonetheless, debate continued concerning the merits and wisdom of mass screening. There was additional concern over treatment methods; physicians and scientists felt that their knowledge of PKU was preliminary and there was much more to be learned about the disease, its' origins and the range of treatments that could be made available before one blanket solution could be advocated. Physicians were certain that PKU did not affect all people equally but rather was found predominantly in one population; people of northern European descent. The legislated mandatory screening could well have been unnecessary; rather, voluntary screening of particular populations could have achieved the same results. In fact, Washington DC. gave up its' mandatory program of screening after

three years of testing mostly black newborns failed to find one single baby affected with PKU.

Notwithstanding attendant doubts and criticisms, PKU screening has proceeded for three decades. PKU screening is accepted as a standard component of childbirth and neonatal care. Many women giving birth in hospital are unaware that their newborns are being screened for PKU and a host of other diseases as a matter of law and/or routine. In Quebec, no requirement of informed consent exists in which new mothers are asked for their informed consent to newborn testing. PKU screening has become hospital policy, the same as weighing and measuring a child, or doing an APGAR. The only time a mother would necessarily be made aware of the testing is if her newborn were found to carry a disorder.

In Montreal, the very first PND was done on an experimental basis by Dr. Louis Dallaire in 1968. His success was the impetus for him to continue offering the service (MacKay and Fraser, 1993: 44). At the end of 1971 there were 13 genetic centres in Canada continually expanding and improving upon their services. Yet this expansion was on an ad hoc basis and the source of some worry by professionals that standards of care and practice were lacking in this new field. The Medical Research Counsel (MRC) appointed a working group to investigate the safety, efficiency, benefits and limitations of amniocentesis. The first report making recommendations was tabled in 1977. It's aim was to establish guidelines for the practice of amniocentesis under the auspices of three professional organizations: The Canadian Paediatric Society (CPS), the Society of Obstetricians

and Gynecologists of Canada (SOGC) and the Genetics Society of Canada (GSC) . The report made recommendations regarding the use of amniocentesis as an effective technique, albeit with a small associated risk factor; genetic counseling was cited as an important component of the delivery of services as well as the need for provincial funding. With these guidelines amniocentesis moved from the realm of experimental procedure into that of standard of practice (MacKay and Fraser, 1993: 28-30).

Although it is widely believed that Chorionic Villus Sampling (CVS) is a relative newcomer to the PND family of techniques, it came into existence at the same time as amniocentesis, that is, in the 1960s. CVS was developed to ensure an earlier diagnosis than amniocentesis, while an amniocentesis is performed at between sixteen and eighteen weeks of pregnancy, CVS can be performed between the ninth and the twelfth week (Blatt, 1988). Procedural difficulties hampered the use of CVS but by the 1980s, these difficulties were largely overcome. Canada has recently completed the first randomized clinical trial of CVS in comparison to amniocentesis. This trial concluded that there was a slightly greater chance of miscarriage in women undergoing CVS as compared to those undergoing amniocentesis which has to be weighed against the advantages of earlier diagnosis afforded by the technique (MacKay and Fraser, 1993: 32; 113-14).

Further advances in the field of PND include the discovery that maternal serum alpha-fetoprotein (MSAFP) was elevated in association with the presence of fetal neural tube defects (MacKay and Fraser, 1993: 4). MSAFP screening is believed to detect up to

90% of all cases of spina bifida and anencephaly in the fetus (Blatt, 1988: 46). Fetoscopy, ultrasound and DNA technology are three more techniques used since the 1970s to aid in PND. The 1980s saw the increasing technical ability of scientists to perfect these techniques and this continues in to the 1990s (MacKay and Fraser, 1993). PND is now a routine part of obstetrical care for Canadian women.

4.1 (3) Genetic centres in Canada

There are twenty-two genetic centres in Canada: one in Manitoba, Nova Scotia and Newfoundland, two in each of British Columbia., Alberta, and Saskatchewan, three in Quebec and ten in Ontario. In 1990 five percent of all pregnant women (22,222 women) were referred to a genetic centre. All centres offered amniocentesis, only twelve offered CVS. Of the women referred to centres, 78% were referred because of advanced maternal age (AMA), 3.6% for abnormal MSAFP, 3.1% for abnormal ultrasound, 2.4% for a previous chromosome abnormality, 2.3% for familial chromosome abnormality, 1.6% for single gene disorders , 1.4% for possible teratogen exposure and 7.6% for other reasons. (Hamerton et al, 1993: 1).

There were obvious regional differences in the delivery and use of genetic services across Canada. While some of the differences are clearly related to the availability of service, in particular to rural or remote areas, other differences appear to be related to underlying attitudes and values of physicians and/or the women themselves.

Obstetricians accounted for over 56% of referrals in the Canadian aggregate. The range for obstetrician referral is from over

84% in North York, Ottawa and Saskatoon to a low of 25% in British Columbia (Hamerton et al, 1993: 24).

The most common reason for referral is advanced maternal age (AMA) which is itself subject to interpretation. With regards to amniocentesis, AMA is considered to be 35 years at the time of delivery. Interestingly, one centre in Quebec lowered this age to 34.2 for a brief time in 1990 but reinstated the 35 year limit shortly thereafter. There is much greater variety with CVS. The standard cut-off is 35 years but several centres restrict this procedure to older women: 37 at Toronto General, 39 at Université de Montréal and Laval and 40 years at McGill. There were also differences in rates of referral: 87.5% of all referrals in BC. were for AMA compared with only 53.3% in Newfoundland. It was estimated that approximately 52% of all women eligible for referral due to AMA were actually referred to a genetic centre (Hamerton et al., 1993: 33-36).

Most women referred to genetic centres undergo amniocentesis (70%) while a small number have CVS (9.4%). The highest rate for CVS was in BC. (27.1%) and the lowest was in Quebec (2.1%). Indications for CVS were overwhelmingly AMA (86.6%) with a range of 98.6% in Toronto to 66.1% in Quebec. The second most common reason for CVS was having a previous child with a chromosome anomaly (7.5%) (Hamerton et al., 1993: 49).

In all centres the most common reason for undergoing an amniocentesis was AMA (82,2%) with a range of 88.8% in BC. to 67% in Newfoundland. The second most common reason for amniocentesis was abnormal levels of MSAFP (3.9%) followed by

abnormal findings on an ultrasound (3.6%) (Hamerton et al., 1993: 57-58).

It is also noteworthy that approximately 52% of all women over age 35 were recommended to a genetics centre. The proportion of eligible women referred vary greatly across Canada, with a high of 65% in Quebec and only 15% in Newfoundland. This is an inequity that the authors of the study felt was imperative to rectify to ascertain that all Canadian women were given the same opportunities (Hamerton et al, 1993: 104).

Counseling services are an integral part of the genetics centres, allowing women (couples) to make informed, consensual decisions regarding their reproductive behaviour. Regrettably, Hamerton et al felt that counseling services were somewhat lacking and again, there were very large regional disparities: "responsibility for this activity [counseling] rested with the primary care physician" although "it would seem appropriate that [genetic counselors] be encouraged to take a more active role in prenatal counseling and patient support" in centres where they are not already doing so (1993: 105). It is most important that the counseling be non-directive which appears to be the case in most centres. However, one centre maintained that testing be discouraged for women who had made it clear they would not consider termination (abortion) as an option (Hamerton et al, 1993: 103) This is by far a minority position and one that has received some criticism from the public and from other centres as it undermines the very notion of informed consent by potentially withholding information from some individuals. It is precisely this kind of ethical and moral issue that has alarmed many special

interest groups and individuals. Whereas the centres are concerned with providing a professional service, others have questioned the very idea that such services should be provided at all. It was the attempt to understand these disparate viewpoints that led to the establishment of a royal commission to study the rapid changes in reproductive technologies.

4.2 (1) Tay-Sachs Disease

Tay-Sachs disease was first recognized by the British doctor, Tay in 1881 and by an American neurologist, Sachs, several years later. The familial nature of the disease was first remarked upon by Sachs, who clearly noticed that Jewish families were more likely to be affected by the disorder he called "amaurotic familial idiocy" Yet it wasn't until 1942 that the cause of the disorder was discovered to be an excess of a lipid called "ganglioside" by Klenk, the physician who made the discovery(Kolodny, 1979: 217).

The infant with Tay-Sachs disease usually appears to be completely normal in the first months of life, although some subtle differences are noticeable by experienced physicians or parents. Increasingly, the child appears hypotonic and apathetic. Towards six or eight months the child loses it's previously developed ability to hold up it's head or to roll over or to sit up. This deterioration continues such that as the child approaches its' first birthday, he or she rarely engages in any purposeful movement, loses whatever motor skills he or she had acquired and becomes more spastic and rigid.

Following the child's first birthday and into the second year of life, excessive drooling, bouts of unmotivated laughter and generalized convulsions become common. At about 18 months, the head begins to enlarge out of proportion to other growth and the child is usually blind.

After the second birthday and into the third year of life, the child remains in a vegetative state, requiring nonstop nursing care for feeding and the prevention of infection. If the child survives to its' third birthday, the hands become pudgy, the fingers taper, and the skin assumes a yellowish colour. Generally, the child's convulsions increase in severity throughout this time period but progressive cachexia and aspiration pneumonia are usually given as the cause of death. Most children with Tay-Sachs die before the age of five.

There is no cure for Tay-Sachs. In the past, before screening programs, parents who had a Tay-Sachs child could only stand by helplessly as their seemingly normal infant deteriorated within its' short lifetime. Treatment has generally consisted of tending to proper nutrition, usually through a permanent gastrostomy tube, and administering to skin infections (bed sores). Treatment also helps to control seizures through the use of medications. Essentially these treatments are palliative: existing to ease the discomfort of the child and his or her parents and to allow the child a relatively painless and untraumatic death. The impetus for screening programs for Tay-Sachs disease and for the availability of therapeutic abortion for women diagnosed prenatally as carrying a fetus with Tay-Sachs has been made widely on humanitarian grounds. The disease is

particularly harsh on the parents as they must watch the unstoppable decline of an infant that appeared normal at birth. The added grief stemming from the knowledge that every subsequent child would have a one in four chance of also having the disease was considered to be too much for a couple to bear.

Tay-Sachs is rare in the general population; the newborn incidence is 1:360 000, but is much more frequent in Ashkenazi Jews with a newborn incidence of approximately 1:3600. It is an autosomal recessive disorder, and approximately 3-4%(1:30) of Ashkenazi Jews are carriers of the gene(NAS, 1975: 129). This incidence rate is similar to that of CF, which has been put at 1:25 or 1:30.

4.2 (2) Tay-Sachs Screening

Tay-Sachs, along with sickle cell anemia and thalassemia, was one of the first autosomal recessive conditions that warranted consideration for population screening. With these screening programs, the emphasis again shifted from treatment to prevention. There was no need for newborn testing for Tay-Sachs as the disease would very quickly make itself known and there was no treatment available for affected children.

Rather than treatment of affected individuals, the goal of Tay-Sachs screening was to "treat" couples who were both carriers of the Tay-Sachs gene and who wanted to have children who would be unaffected with the disease. The "treatment" was the identification of couples in which both partners carried the Tay-Sachs gene and the extension of prenatal diagnosis of the fetus to ascertain it's carrier

status. A fetus with two copies of the gene could be aborted. The intention was that with these available testing procedures, there was no pressure put on people regarding mate selection or reproductive decisions; that is, the decision to have or not to have children. To this end Tay-Sachs screening programs have traditionally been viewed as a great success. They have allowed many couples to avoid the heartbreak of giving birth to a Tay-Sachs child while at the same time allowing them to have healthy children.

Tay-Sachs is a horrendous disease that offers no possibility for meaningful human engagement on the part of the child and absolutely no quality of life whatsoever. The child who is born is already dying as it has been determined that the deterioration made apparent at the sixth month actually begins from the moment of birth or even before birth. Given the sequalae of this disease, the consensus is Tay Sachs is an excellent candidate for genetic screening and therapeutic abortion (this latter only if one has no religious or secular objection to abortion). Nonetheless it is fruitful to explore the mechanisms of testing and the practice of population screening. The analysis of this must first separate the mechanisms of testing from the disease. The organization of screening services and the provision of this particular health care service could conceivably be envisioned in many distinct manners. Thus it becomes informative and essential to examine the particular mechanisms instated to encourage testing within the relatively small community of Ashkenazi Jews.

While Tay Sachs has been the focus of concentrated population screening, the population is question is actually a sub population; it is

not nearly as large as the population that is considered to be at risk for CF. Two different projects, one in the US and one in Canada are widely seen as particularly successful.

4.2 (3) Baltimore/Washington, DC

The Baltimore/Washington, DC. pilot study is widely hailed as one of the best examples of how genetic testing services should be offered(Kaback et al, 1974; McQueen, 1975; Childs et al, 1976). Conducted over a two year period between 1971-1973, this screening program tested 6938 individuals the first year, with a two year total of 17,000 people screened. The entire process was planned with extreme attention to detail through a process of "community education" that began approximately eight weeks before the screening designed as a mass blanketing of the community of Ashkenazi Jews. For a period of approximately 14 months before the initiation of public education, "leadership" training was conducted with volunteers who were to help with education and manpower organization. These volunteers were to be indispensable when the actual testing began as they were necessary for the proper processing of those wanting to be tested. Kaback and his colleagues (1974) described the educational process as follows:

> "Educational mechanisms utilized: letters from rabbinical leadership to their congregants, fliers from community organizations, medical presentations in the community, telephone calls from squads of specially trained volunteers, brochures and information provided by medical practitioners and special mailings from other community leadership".

The Baltimore/Washington study had an almost military like strategy for getting people to "volunteer" to be tested. The research design even allowed for a follow up on non-participants. Because there was a master list of individuals who had received the educational program, it was easy to determine which individuals had opted out of testing. These "noncompliants" were sent the same questionnaire that had been filled out by participants and their answers were processed in the same way as participants. Interestingly, 82% of non participants complied with the request to participate by filling out the questionnaire.

The level of participation in the community was very high. This is considered to be a direct result of the educational process-a multi-media blitz designed to convince individuals and couples to be tested and appealing to them on a personal and a broader community level. Of the 17 000 people tested, 17 couples (0.001%) were found to be at risk; that is, both partners carried the Tay-Sachs gene. Of these 17 couples, 8 had pregnancies and one affected fetus was discovered and aborted. Because they were disproportionately affected by the disease, the awareness level of Tay-Sachs among Ashkenazi Jews was relatively high even before the additional educational mechanisms of the pilot project. It was not reported if the couple with the affected fetus had a family history of the disease. If they had, it would be safe to assume that they would have presented for testing regardless of the existence of the pilot project.

4.2 (4) Screening in Montreal

The Montreal study differed from that of Baltimore/Washington in the targeted population (Beck et al, 1974; Clow et al, 1977). Whereas the US study had concentrated on couples to the extent that they would not test unmarried individuals, the Montreal study concentrated its efforts on adolescents.

The Tay-Sachs screening program in Montreal was initiated by the Jewish community itself and the input and cooperation of the community was vital to the success of the program. Tay-Sachs screening is set in the high schools where there is easy access to the students, creating what has been described as a "captive" population. The screening was introduced to students as part of a biology lesson. Researchers with the study would make arrangements with individual schools to give a presentation to the students regarding genetic inheritance patterns in general and Tay-Sachs in particular. Following this presentation in which it was impressed upon students that they were at risk of one day having a child with Tay-Sachs, the students were asked to volunteer to be tested. While there was no covert pressure to be tested, the high school milieu is well known as a place that has elements of collective behaviour and peer pressure. To what extent these elements impact on the students' decision is unclear. Following the test, students were advised by letter mailed to their home as to their carrier status. Carriers were invited to contact a genetic counselor for further information if they felt inclined. Parents of carriers were also advised to be tested although what purpose this would serve is questionable. We can assume that only rarely would parents of a 17 or 18 year old be contemplating

additional children. Siblings of carriers were also advised to be tested before they began planning their own families.

There has been much concern about the psychological impact on adolescents of genetic testing. A study comparing 45 carriers with 45 noncarriers whose median age was sixteen and a half showed that there was some psychological or emotional disturbances. 15% of both groups said that they found the prospect of screening to be frightening. Curiously, although Tay-Sachs is found almost exclusively in Ashkenazi Jews, and the students were presumably aware of this, more than half of carriers and noncarriers felt that the screening should not be confined to Jewish students. They felt that screening would be helpful to others. One must wonder then, if the students truly understood the message being conveyed to them regarding the notion of risk. Although Tay-Sachs does appear, albeit rarely, in the general population, it would be very difficult to attempt to screen for it. Given this trend to advocate screening for individuals who would not really be at risk, one must wonder about the relevance of claims that high school students are favourably disposed to genetic testing. Could it be that they are just taken in by the techniques themselves without an adequate understanding of the true issues involved?

One of the most compelling issues is that of self-esteem in carriers. Heterozygotes were initially more upset than were noncarriers; this makes perfect sense. 88% of carriers said that their self-image was unchanged by this new information but nearly 9% stated that their self-image had diminished. Curiously, 9.5% of non carriers stated that their self-image had *improved* after receiving the

test results. In addition, twice as many carriers as noncarriers did not tell their friends about their test results. The implication is that noncarriers felt that not being a carrier was a *good* thing, and good news was to be shared, whereas 33% of carriers were reluctant to share the news of their heterozygosity. In addition, 12% of noncarriers stated that they would not marry a carrier, indicating either ignorance of genetic inheritance patterns or bias against carriers. Unfortunately this wasn't explored further.

A follow-up study revisited carriers eight years after they were screened in the high schools(Zeesman et al, 1984). The researchers surveyed 264 persons with a response rate of 42% (38 carriers and 45 noncarriers). While carriers recalled that they had been worried by their test results initially, they reported that now they were less worried and most were characterized as being indifferent to their status. The survey sought to understand how or if knowledge of genotype might affect choice of a marriage partner. 95% of female carriers said that they would not modify their plans if they discovered their intended partner was also a carrier but only 69% of males made the same statement. A full 92% of noncarriers thought that a carrier couple would separate if they learned of their joint status, although only 70% said that they themselves would do so.

The data from the survey appeared, at first glance, to demonstrate that concerns about adverse affects of screening on individuals was misplaced. However, there are some alarming findings in the results. How do we interpret the fact that nearly 10% of noncarriers had an improved self image after getting test results?

This must be interpreted as an indication that it is clearly not a good thing to be a carrier of Tay-Sachs. Even though these people were in the minority, the impact of their feelings could prove to be important. Given that 9% of carriers felt a diminished sense of self-image, we can conclude that for approximately 10% of the population, carrying a gene for Tay-Sachs is associated with negative image for oneself and for others. The seeds for animosity toward carriers is further elaborated by the 12% of noncarriers who would not marry a carrier. Given that one has a wide choice of whom to marry, we can image that this part of the population would rather stick with undamaged goods. There is no breakdown of statistics on the basis of sex, but this would be very interesting to see. It is likely more males than females would refuse to marry a carrier given that 31% of male carriers said that they would modify their marriage plans if they discovered their partner was a carrier compared with only 5% of female carriers. These figures call attention to the idea that carrier status is not considered as a simple fact for many individuals but that it has implications for social relationships that are very real and very insidious. It is striking that 92% of noncarriers thought a carrier couple would separate; the unasked follow up question would be what percent thought a carrier couple *should* separate. This 92% rate reemphasizes our pronatal social policy while at the same time drawing attention to the pronatalist belief in producing good quality children. In doing so, these beliefs highlight that most people feel a relationship is for the purposes of reproduction. When nearly everyone surveyed agrees that two carriers will separate, it shows that nearly everyone agrees that the reasons for them to be together

in the first place is to have children. This ignores the other equally important aspects of a relationship; love, companionship, friendship and gives a very disjointed portrayal of what adults' lives are really like. The emphasis here is firmly on the genetic profile of the individual and on what steps they can and should take to prevent that genetic profile from being reproduced. And while concerns about stigmatization appear to be unfounded for the most part, the entire program of screening is based upon the acceptance of the belief that certain genes are not welcome. Thus any improvement in self-esteem among some individuals can only be realized by negating the value of others. As Goodman and Goodman have pointed out "The psychological effects of Tay-Sachs screening on the majority of the carriers, who will never produce an affected child, is a cost that must be paid for the location of a small minority who will" (1982: 24). The policy has been to accept these costs because we have assigned a high value to the prevention of a small number of affected births and we see the negative effects of education campaigns and screening as being relatively minor. This is a choice that we have made on a *social* level, and it is not one that has an absolute truth to it. We have *chosen* to proceed on the basis that the suffering that would be incurred to one carrying a Tay-Sachs fetus would be more than the suffering that occurs to nearly 10% of all carriers of the gene. We have *chosen* to construct our technological apparatus in such a way that our ethical considerations would seem to support its' use. This shows, not that the technology is necessary, vital and minimizes suffering but that we know how to rationalize our choices before and after the fact.

Tay-Sachs is a success story of screening although it does encompass some areas of dispute in the evaluation of success. Sickle Cell Anemia screening programs could never be called a success, although it differs very little in substance or form from Tay-Sachs. It does provide some important lessons for us that the success of the Tay-Sachs experience may have allowed us to overlook.

Tay Sachs screening programs were and continue to be a resounding success in terms of their establishment and ability to reach and process large numbers of individuals in such a way as to have seriously influenced a decrease in birth rate of babies with the disease. They are also a success in that, unlike sickle cell, Tay Sachs programs continue to share the good graces of the community. Nevertheless, it has increasingly been noted that the amounts of human energy and financial expenditure required for prevention may not have been well spent when weighed against the costs (financial and human) of the few babies who would have been born without these programs.

4.3 Sickle Cell Anemia

The misguided attempts to screen for sickle cell anemia in the American Black population have been chronicled in nearly every article and book to discuss genetic disorders. The failure of the programs is predominantly understood as being an organizational failure and one that may have been avoided given adequate preplanning. However, the failure was more deeply rooted; the program was a failure at the very heart because it was based on the

belief that technology could be used, not only *for* people, but *on* people.

4.3 (1) Sickle cell anemia in the population

Sickle cell anemia is a blood disorder that affects roughly 1 of every 400 to 600 blacks in the US. Like Tay-Sachs and CF, sickle cell anemia is an autosomal recessive disorder--a person must inherit one sickle cell gene from each parent in order to have the disease. People who inherit one affected gene from a parent and a normal gene from the other parent are said to have sickle cell trait--they are carriers of the disease even though they do not have the disease itself and are at no risk of ever developing the disease

Approximately 1 in 10 to 12 American blacks are carriers. With sickle cell anemia, normally rounded blood cells take on a crescent (sicled) shape under periods of stress, which can limit the supply of oxygen needed for the body to function. Medical problems that occur include pain from swelling joints and sickle cell crises. While fatal infections can occur, sickle cell anemia is not considered to be a fatal disease. Still, sickle cell is a painful and debilitating disease that has no cure. Some people are moderately affected, have few crises and can live well into adulthood. A child born with the disease has a 50% chance of living into middle age. Based on these factors, the disease has more in common with Cystic Fibrosis than it does with Tay-Sachs. However, many social scientists have highlighted the fact that both Tay-Sachs and Sickle Cell Anemia and their screening programs have occurred in well defined minority communities. The implication of this is that public policy is seen as focusing attention on a community that is usually ignored and that

this sudden attention to the problems within a community can be looked upon with great suspicion. In addition, such attention may result in members being ostracized from society

4.3 (2) Screening for Sickle Cell Anemia

While screening for Tay-Sachs was done on a voluntary basis, the United States was very quick to make sickle cell testing mandatory. By the Mid 1970s, several states had passed laws making testing a requirement for getting a marriage license or entering school. The children were being tested to determine if they carried the gene, not to determine if they had the disease. The knowledge was useless to young children who surely were not about to make reproductive decisions. The total lack of sense that went into the creation of the law was lost to the lawmakers. In his book, *Backdoor to Eugenics*, Troy Duster (1990) elucidates the point that even though Tay-Sachs was a much more fatal disorder, testing for sickle cell became mandatory. With no input from the Black community, there seemed little doubt that the legislation was a form of legalized racism. Blacks were being told that there was something very wrong with them and that they should be taking steps to avoid having children if they carried this disease. Focusing on the black population, the law makers ignored the fact that sickle cell disease is also common among Greeks, Arabs, and Sicilians. Charges of racism greeted the white doctors who arrived in the community to do the tests. There were accusations that not enough was being done to get everyone tested and there were accusations that requiring everyone to be tested was a form of genocide. The principle problem was that the community had little control over the programs but a more

fundamental problem was linked to ongoing discrepancies in the black and white communities. Socio-economic status of blacks was well below that of whites and it is these wide gaps in the income and education levels that accounted for much of the problems. Trying to build a genetic screening program on the precarious basis of black/white race relations in the US at that time was doomed for disaster. Added to this was the fact that the two separate statuses of being a carrier and having the disease were conflated into one undesirable status.

4.3 (3) Stigmatization and racism

Stigmatization of both those with the disease and carriers of the gene was fast and furious. The US Army began to screen all black recruits for both the trait and the disease and to place limits on carriers even though a carrier did not have, nor would ever have, the disease. Interestingly, one of the exclusions was copiloting airplanes. The Air Force carried out a policy of excluding altogether blacks who carried the gene. The policy was in effect from 1973 until 1979 when they were successfully sued by a recruit.

Insurance companies initially charged higher rates to blacks who were carriers although this policy has been stopped as a result of governmental intervention. More insidious is the fact that employers have tried, sometimes successfully, to prevent carriers from taking jobs in certain areas. DuPont, for example, has a policy that restricts sickle cell carriers from handling certain chemicals for fear that they would be more at risk for an on-the-job accident (Holtzman, 1989).

The disaster of sickle cell anemia screening occurred on many levels. It was poorly organized and it was done in too much of a rush without time to adequately educate people. It was done from the outside, with whites coming in to tell blacks what was wrong with them. It suffered from inadequate understanding of the difference between carrying the gene and having the disease and it provided an outlet and a justification for previously existing racism.

Conclusion

The twin examples of screening for Tay Sachs and Sickle Cell are precise reminders of the dual aspects of genetic screening. For some, genetic screening programs are a benign force that enable them to make informed reproductive decisions; for others, they are a mechanism for racism, segregation and stigmatization. The most fundamentally telling element is the support of the community in the origins of testing programs. Without community support and involvement, without the express desire of the community to avail themselves of genetic services, genetic screening programs risk appearing as an ordinance brought down from an uncaring bureaucratic government.

The past twenty years of ongoing Tay Sachs screening services as part of health care genetic services is an indication that individuals are capable of making use of such services in their reproductive decisions. The legacy of sickle cell screening reminds us that technology can serve individuals or individuals can be made to serve technology. It also serves as a caution flag; genetic testing must be a component of global health care provisions rather than as a method of separation and segregation.

Screening programs for the control and eradication of communicable diseases began in the early part of this century but it wasn't until 1964 that genetic screening began with the first PKU screening programs. Tay Sachs was one of the first autosomal recessive conditions that was considered for population screening, along with Sickle Cell Anemia and Thalassemia. The first Tay Sachs programs were established in the early seventies. Tay Sachs was an excellent candidate for screening for two reasons: it was a severe, fatal disorder with no chance of a cure and the population it affected was a small cohesive population that was in favour of the use of such screening in their community. These two factors alone distinguish Tay Sachs from Sickle Cell. In the case of Sickle Cell, the disease itself was not always fatal and while there was no cure, it was possible to control the disease, and perhaps more damaging, the affected community was not largely in favour of screening programs in that many felt they were designed as a racist policy of outside intervenors.

The nearly thirty years of research into screening for Tay Sachs has yielded a wealth of information that will impact on CF screening. However, some large discrepancies exist which mitigate against a comparison. These discrepancies concern the etiology of the two diseases and the population of potential carriers. CF has more in common with Sickle Cell than with Tay Sachs in that it is a treatable disorder. Furthermore, the potential population "at risk" is huge, encompassing virtually the entire Caucasian population. The logistics of organization alone are daunting. Added to this are concerns raised by Sickle Cell screening with regards to stigmatization, insurance

difficulties and lack of community control over programs. The importance of screening only being offered on a voluntary basis was highlighted by the Sickle Cell experience as was the danger of misinformation and misunderstanding, not just of carriers themselves, but of society in general. This is of the utmost importance for CF given the uncertainty of many screening results that would arise due to the many mutations in the population.

Tay Sachs and Sickle Cell screening programs were the real pioneers in genetic screening programs. Any move to this type of program for CF will have to carefully weigh the actions, reactions and consequences of what history has to show.

Chapter 5

Framing the larger context of reproductive technologies

Genetic and reproductive technologies are burgeoning fields not only for science and technology but in the social sciences as well. Specifically, such medical innnovations challenge society to keep abreast of breakthroughs and to assimilate new capabilities with ongoing concerns of values, mores and ethics. However, the question is not one of simple assimilation but rather crucial decisions must be made regarding these innovative technologies and techniques. The most fundamental question is, "Do we want this?" and this is followed by other questions concerning how we will incorporate this knowledge into society while maintaining the integrity and well-being of social members.

This chapter explores the broad context of reproductive technologies. It situates the research on genetic screening within a wider arena and in so doing demonstrates the importance of understanding the social context of changes that are ongoing. If we limit the discussion to genetic screening, it is possible that the widerscope of social changes will not be recognized. Genetic screening is part of other technological breakthroughs that affect all the members of society even those unaware or unconcerned about such issues. These technologies create an entirely different way of looking at the body and at our social and family relationships.

Rapid changes in ability tend to be followed more slowly by changes in values and mores. Thus society must strain it's collective

capabilities to accomodate changes in the way that human beings can be made, are made and can and are treated. Some critics have cautioned against the rampant commodification of the human body and hence of humanity itself, whereas proponents of the new technologies herald the wide range of hitherto impossible possibilities.

Commissions were set up to deal with these dichotomous viewpoints and to try to formulate guidelines for dealing with these delicate subjects. Granted, ethical guidelines are already in existance with regards to the use of humans for science and research but it was felt that these new matters were important enough and compelling enough to warrant a very close look and how and what they proposed to accomplish. Many countries have established their own guidelines for dealing with genetic and reproductive technologies. The Canadian Commission involved a comprehensive overview of research coupled with face-to-face presentations by interested parties. It yielded a wealth of written material and culminated in the creation of recommendations for all reproductive technology services in Canada.

5.1 The Royal Commission on New Reproductive Technologies

Although it is clear that PND is not a new phenomena, the relatively quick advances and expansion that have been realized within the past decade have thrust it into the public consciousness seemingly for the first time. As such, the public debate of the merits and demerits of such procedures occupy much of our collective interests. The intense examination of PND was the initiative for the

establishment of the Royal Commission on New Reproductive Technologies, a commission that could listen to and respond to the concerns and worries of individuals and special interest groups in society regarding their hopes and fears of the future that PND was heralding as a mythic and as a concrete phenomenon.

5.1 (1) The Committee and it's Mandate

The Royal Commission on New Reproductive Technologies was established by the Canadian (federal) government in October 1989. Established under Part 1 of the *Inquiries Act*, it was to

> "inquire into and report on current and potential medical and scientific developments related to new reproductive technologies, considering in particular their social, ethical, health, research, legal and economic implications and the public interest, recommending what policies and safeguards should be applied" (Mandate, 1989).

The scope of the commission was broad; the press release announcing it's establishment outlined it's mandate as follows:

> The Commission will examine in particular:
>
> a) implications of new reproductive technologies for women's reproductive health and well-being;
>
> b) the causes, treatment and prevention of male and female sterility;
>
> c) reversals of sterilization procedures, artificial insemination, *in vitro* fertilization, embryo transfer, prenatal screening and diagnostic techniques, genetic manipulation and therapeutic interventions to correct genetic anomalies, sex selection techniques, embryo experimentation and fetal tissue transplants;

d) social and legal arrangements, such as surrogate childbearing, judicial interventions during gestation and birth, and "ownership" of ova, sperm, embryos and fetal tissue;

e) the status and rights of people using or contributing to reproductive services, such as access to procedures, "rights" to parenthood, informed consent, status of gamete donors and confidentiality, and the impact of these services on all concerned parties, particularly the children;

f) the economic ramifications of these technologies, such as the commercial marketing of ova, sperm and embryos, the application of patent law, and the funding of research and procedures including fertility treatment

Nine commissioners from a varied background were appointed to the task to ensure a wide spectrum of participation and perspectives. Dr. Patricia Baird, with a background in science and medicine, was appointed chair of the commission. She was joined by Dr. Bruce Hatfield, a physician, Martin Hébert, a lawyer and member of many hospital ethics committees, Grace Jantzen, a lecturer in the Department of Theology and Religious Studies at University of London, Bartha Knoppers, a professor of law, Susan McCutcheon, a past Chairman of the Board of Directors of the Women's College Hospital, Maureen McTeer, a member of the Law Society and more relevently, an active member of groups that lobbied for the establishment of the commission, Suzanne Scorsone, Director of the Office of Catholic Family Life for the Archdiocese of Toronto and Dr. Louise Vandelac, a professor of sociology widely published in the field of reproductive technologies.

The Commission established a public consultations program and a research program in order to carry out their mandate of gathering information and responding to public opinion. The Commission traveled to 17 cities across Canada between September 11-November 29, 1990, hearing from more than 550 people who appeared on their own behalf or as representatives of nearly 250 organizations.

5.1 (3) Recommendations of the Royal Commission

Commission guidelines recognize that reproductive technologies have both potential harms and potential benefits. Given the vested interests that society, as a whole, has in such technologies, the commission felt that "it is the role of governments as guardians of the public interest to ensure that individuals and society as a whole are not harmed by inappropriate use of reproductive technologies" (Baird et al, 1993b: 1020). Inasmuch, the commission's guidelines recognize the shared interest of many segments in society in providing leadership, thus many different groups are expected to play a role in the regulation of reproductive technologies and in the education of members of society regarding their rights.

The commission named several actors as having a joint responsibility toward regulation and education: the federal and provincial/territorial governments, the health professions, private sector interests, and various advocacy and public interest groups.

The recommendations are quite lengthy, addressing many different levels of concern by area of responsibility. These will be discussed in detail below.

5.1 (4) Dissension within the ranks

After four years, three deadline extensions and $25 million spent, the 1 200 plus page report was released 1 December 1993. Even prior to it's release, the report and the Commission had been embroiled in controversy that stemmed from attempts by four commissioners to gain access to the commission's budget and research plans, access that had been denied them by the chair, Dr. Baird (Branswell & Bueckert, 1993). On 6 December 1991, four commission members, Martin Hébert, Louise Vandelac, Bruce Hatfield and Maureen McTeer, brought a complaint to the federal court listing Dr. Baird as defendant. They charged that Dr. Baird had assumed an authority that effectively prevented them from fully participating in the commissions work as set out by the government mandate. The brief filed in federal court alleged that "the plaintiffs have been progressively distanced and prevented from participating in every important decision concerning the Commission's on-going operations including the nature of the Royal Commission's research, its consultation and communication program and its organizational and financial priorities..."(Basen et al, 1993: 276). Dr. Baird's authoritative reach was so broad, the plaintiffs claimed, that they were not privy to

> "the determination of how, and under what conditions, research projects would be formulated, and to whom they would be contracted, and for what amounts" and further that the Chair "systematically refuse[d] to provide, or allow staff members to provide, to all Commissioners, clear and complete information on the entirely of the research program" (Basen et al, 1993: 281).

Unfortunately none of these issues was ever addressed. Before the suit could come up in court, all four commissioners were fired and the Royal Commission proceeded without them. There were additional charges by the Social Science Federation of Canada that the research undertaken did not meet accepted standards for the research community (Branswell & Bueckert, 1993). Added to these allegations are the stories of six anonymous individuals who worked in various capacities for the Royal Commission. They describe a Kafkaesque environment in which activities and phone calls were monitored, offices were swept for "bugs", dissenting opinions were not tolerated and were punished, and people could be and were, suddenly fired which contributed to an ongoing atmosphere of fear in which very little work was done (Basen et al, 1993: 223-236).

Critics of the Royal Commission voiced objections concerning their perception of the scientific bias of the commissions work; specifically, critics argue that the chair of the commission, as a scientist, prioritized a scientific agenda over a thorough analysis of ethical and moral criterion of the reproductive technologies. The focus of the Commission's recommendations was on guaranteeing effective management of technological services. The two volume report enumerated clinics and available services but made little critical analysis of these services; the underlying assumption garnered was that reproductive technologies are benign and need only be properly monitored and licensed. When the commission acknowledged critics of reproductive technologies, their acknowledgment took the form of general reassurance rather than specific answers to queries. "By increasing the volume and

accessibility of objective information about new reproductive technologies, such publication will also help Canadians make informed decisions about whether and under what circumstances to consider using these technologies" (Baird et al, 1993: 119). This course of action serves to advocate a program of *individual* responsibility, wherein each person must choose from the services made available by science and research.

An important recommendation that arose from report was for the establishment of a National Reproductive Technologies Commission organized along the same lines as the National Transportation Agency and the Canadian Radio-Television and Telecommunications Commission with a mandate for "licensing and monitoring; guideline and standard setting; information collection, evaluation and dissemination; records storage; consultation, coordination and intergovernmental cooperation; and monitoring of future technologies and practices(Baird et al, 1993: 115-116). It is clear that the Royal Commission views most reproductive technologies as an accepted standard of practice in Canadian society.

5.1 (5) Bill C-47

The commissioners state in the last line of their report that "the next steps are not ours-they belong to governments, the professions, and individual Canadians" (1993b: 1050). Bill C-47 is a response to this mandante. The *Human Reproductive and Genetic Technologies Act* was introduced and first read 14 June 1996 by Health Minister David Dingwall. This act "prohibits the use of certain reproductive and genetic technolgies in relation to human beings as well as certain

commercial arrangements relating to human reproduction". The Act would prohibit the following practices:

Sex selection for non-medical purposes

Buying and selling of eggs, sperm and embryos.

Germ-line genetic alteration

Ectogenesis

Cloningof humans

Creation of animal-human hybrids

Retrieval of sperm or eggs from cadavers or fetuses

Surrogacy arrangements

Transfer of embryos between human and other species

The use of human sperm, eggs or embryos for assisted human reproduction procedures or for medical research without the informed consent of the donor(s)

Research on human embryos later than 14 days after conception

Creation of embryos for research purposes only

Offer to provide or offer to pay for prohibited services

Persons who contravene this act are liable for fines up to $500.000 and jail terms up to ten years. Unfortunately, the legislation died after the election. This left no legislation to curb the above practices. Furthermore, on 9 July 1998 the *Globe and Mail* reported that female students at elite universities were being actively recruited to sell their eggs to infertile couples. With no legislation in place, the market place mentality is able to reign.

Reproductive technologies have the power to transform society. What society must do is to rein in that transformation so that it reflects as much as possible common societal desires and values.

5.2 Federal Government

With regards to the most potentially harmful practices the commission felt that there is a need for criminal legislation to set boundaries regarding the use of reproductive technologies. Some actions are so potentially harmful that they must be prohibited. The commission cited the following:

1. human sygote/embryo research related to ectogenesis, cloning, animal/human hybrids, the transfer of zygotes to another species, the maturation and fertilization of eggs from human fetuses.
2. the sale of human eggs, sperm, zygotes, fetuses, and fetal tissues.
3. all aspects of surrogacy arrangements.

Legislation should also be enacted to prohibit:

4. unwanted medical treatment and other interferences with the physical autononomy of pregnant women.
5. the confinement or imprisonment of a woman int her interests of her fetus
6. the criminalization of any conduct of a woman in relation to her fetus.

Apart from the outright criminalisation of particular practices, the commission recommended the establishment of a National Reproductive Technologies Commission "to ensure that reproductive technologies are provided in a safe, ethical, and accountable way

within legal boundaries. The commission should be representative of advocacy and special interest groups and should always include at least fifty percent female representation. Members should demonstrate a range of backgrounds including medicine, ethics, law and social sciences. In addition, six sub-committees should be formed focusing on:

1. infertility prevention
2. assisted conception
3. assisted insemination
4. prenatal diagnosis and genetics
5. human zygote/embryo research
6. the provision of fetal tissue for use in research.

(For the purposes herein, solely the recommendations concerning prenatal diagnosis and genetics will be discussed in detail.)

The recommendations issued by the commission regarding federal responsibility for prenatal diagnosis and genetics are concerned principally with the licencing requriements of facilities that wish to provide such services. Licencing would be required for any facility providing amniocentesis, CVS, and any other prenatal testing of women for the purposes of obtaining information regarding the genetic health status of the fetus. However, these recommendations do not apply to MSAFP programs, ultrasound programs and blood tests.

Licencing conditions also include the requirement that counseling be made available and be non-directive; that pregnancy termination not be a determing factor in the provision of prenatal testing; that record keeping protect patient confidentiality; that there

be annual reporting to the National Reproductive Technologies Commission.

With regards to the testing of late-onset disorders, the recommendations are that special counseling be made available in such instances.

The commission states that the detection of susceptibility genes not be a part of prenatal diagnostic services. Additionally, fetal sex selection should be disallowed, with fetal sex being revealed only if it is medically relevant.

Finally, in addition to licencing, the sub committee would also be responsible for monitoring ongoing clincial research trials; determining the effectiveness and safety of new services and monitoring developments in DNA testing and gene therapy.

5.3 Provincial government

The commission felt that improvements should be made in the area of counseling. To this end, it recommended that further research be conducted with regards to the best ways to present information and how individuals make decisions with the given information. The value of non-directive counseling is one that must be continually upheld by provisioners of prenatal diagnosis. As well, the Canadian College of Medical Geneticists is called upon to establish a coordinated effort by genetics centres in collaboration with special interests groups to accurately portray the capabilities and life-styles of people living with disabilities that are being diagnosed. To ensure the provsion of these services, it is expected that medical training

will incorporate more teaching and information regarding the provision of genetic services to individuals and their families.

The recommendations offered by the Royal Commission on Reproductive Technologies recognize that technology is at it's very essence a social issue. Individual members and special interest groups in society have a vested interest in the outcome of medical innovation that touches so widely on our human-ness; that asks of society to define what it means to be human. Given the complexity of such interests, it is certain that there are many dissenting viewpoints with regards to the level of intervention necessary to protect the interests of society.

5.4 Issues examined by the Royal Commission

Criticisms of prenatal diagnosis are many and varied and cluster around micro and macro levels of analysis; that is concerns on individuals and concerns on a societal level. Naturally these concerns are separately only artificially. On a practical level, individual concerns are societal concerns and societal concerns affect the individual. That said, positions critical of PND can be divided as follows: concerns surrounding societal attitudes towards persons with disabilities; women's status in society; pressure to abort; the geneticization (individualization) of health problems; medical values/attitudes/terminology; economic concerns such as the proliferation of technology for financial gain, life and health care insurance; and other related issues such as PND for sex selection, for late onset disorders, for genetic susceptibility; and somatic and germ line gene therapy.

5.4 (1). Societal attitudes towards persons with disabilities

Persons with disabilities are among the most vociferous critics of PND in Canada. Their contention is that the very basis of PND is a foundation of negative social attitudes toward disabilities. Societal intolerance of physical and mental disabilities, and a horror of all imperfectness lead individuals to the belief that the disabled live lives of diminished quality that those without disabilities and a presumption that individuals with disabilities would have been happier had they never been born. The disabled are traditionally a marginalized and disenfranchised group but they have made attempts in the past years to counteract these negative beliefs with testimony ascertaining that they are capable of living happy and full lives and that their main obstacle is the discrimination faced daily by a society preoccupied with perfection.

For persons with disabilities, the crux lies in social definitions of "normal". One of their fears is that of a domino effect wherein if we are able to sidestep the "natural lottery" of reproduction:

> our understanding of what a genetic defect is will actually begin to slip. For example, if the genetic engineer can offer a means of guaranteeing that a child has a higher IQ, then today's "normal" level would certainly be considered deficient tomorrow(Elkington, 1985).

Kenneth Vaux deplores the lack of human element, or element of individuality he sees in a strict interpretation of normal. Vaux is critical of the move toward homogeneity:

> The danger of all eugenic proposals is that they drive toward homogeneity. It is a short step from the desire to eliminate defect in the interests of normality to the desire to eliminate diversity in the interest of some norm. Anyone with scientific appreciation knows the value of hybrid vigor and the rigor of diversity but technology and law thrive on regularity and uniformity. Indeed, any bureaucratic society seems to crave regularity for its own purposes of organization and administration (Vaux, 1989: 125).

Vaux raises the point that society is unable to answer; are what we call defects accidents of nature to be eliminated or are they merely manifestations of the true diversity of the human form? For persons with disabilities, increasingly, the tendency is to view them as terrible accidents with no intrinsic value.

The medical management of obstetrics/gynecology reinforces the implicit illusion that medicine can provide a guarantee of health. For some doctors, it is an obligation to "beat" nature at her own game. Says Joseph Fletcher, "We cannot accept the invisible hand of blind chance or random nature in genetics"(cited in Vaux, 1989: 129).

5.4 (2) Women's status in society and the pressure to abort

Societal attitudes towards persons with disabilities provide the framework within which maternal-fetal relationships are experienced. Within this matrix, there can be pressure to abort a fetus that has a disability. This pressure has attained a legitimacy in the courts in the in such a manner as to redefine the maternal-fetal dyad as one that may be constructed as an oppositional relationship rather than as a symbiotic one. The legal argument employed in these circumstances explicitly advocates the intrinsic rights of the fetus including the right to live a good life and the right not to live.

By refusing to abort a fetus with a genetic defect, the individuals involved are denying the fetus "the basic right to prefer death to an unacceptable form of life" (Mason, 1990: 132) Several wrongful birth suits have allowed parents to sue a doctor if their child is born with a handicap that they believe the doctor could have prevented had s/he used PND and offered the woman the chance to abort (Mason, 1990). These cases ask the courts to weigh the value of a handicapped life against non-existence. Is it better to never have been born than to have been born with a handicap? The difficulty in making this judgment did not prevent one court in the United States from ruling that "it cannot be held as a matter of law that an impaired life is preferable to non-life in all situations". The interpretation of this ruling has been that the child has a right to be born "whole" or not to be born at all, "not to be born unless it can be born perfect or "normal", whatever that may mean" (Mason, 1990: 134)

This position has the support of others in society. George Smith states that "the right to procreate may not include the right to breed without restrictions" and that "society now has a compelling interest in restricting reproduction by those who...perpetuate human suffering by giving birth to genetically defective offspring" (1985: 439). John Robertson concurs and suggests that technology can be used to solve the problem that technology creates. While he argues for a couple's constitutional status of a right to procreate, he also suggests that women should routinely use *in vitro fertilization*: "IVF provides a window on the embryo that will eventually enable prenatal diagnosis to occur before implantation, and thus avoid the

stresses of abortion for genetic reasons later in the pregnancy" (1988: 446) Clearly, even for Robertson, who upholds the right to procreate, this right does not include the right to have defective children. This theme is also taken up by Linus Pauling, Nobel prize winner, who condemned couples who continued to have children after the first one was diagnosed with Cystic Fibrosis: "A father and a mother who, with this knowledge, continue to produce defective children and in this way add to the amount of human suffering should feel guilt for their actions" (cited in Holtzman, 1989: 216) Lawyer-Geneticist Margery Shaw also criticizes couples who have children with genetic diseases. Going further she maintains that some wanted children:

> will be physically or mentally defective or diseased and some will have physical pain and suffering that is no less acute than the pain of child abuse...The real tragedy of these tragedies is that the conception or birth of many of these unfortunate children could have been prevented because of advances in our ...ability to predict many of these adverse conditions...The burden has shifted from the physicians to the parents to act in reasonable ways to prevent the birth of a child who would certainly or likely suffer postnatally...parents might incur a legal duty to obtain an abortion [when a fetus is discovered to have a defect] (cited in Holtzman, 1989: 217).

What is to be considered "reasonable" behaviour on the part of women? We could see the time when a woman could be charged with prenatal abuse for refusing to have prenatal diagnosis and/or refuses to abort a "defective" fetus (Taub & Cohen, 1988: 130).

In a study conducted by Dr. Karen Grant of women's perceptions, attitudes and experiences of PND, few women felt their doctors were pressuring them to abort. Nevertheless, there were

cases where women were told that if they did not intend to abort a fetus with a disability, there was no point in them having PND (Grant, 1993: 221). More importantly, women spoke of pressure to undergo testing. This pressure was not tied to any one individual, but rather was a subtle pressure expressed in diverse manners. Women felt that testing would offer a certain level of comfort, even while, in the same breath they expressed the wish that there was no choice to make, i.e. that testing was not an option (Grant, 1993: 227). Women feel a compulsion to choose testing and abortion once that technology becomes available (Hubbard, 1986). In her study of women and prenatal diagnosis, Barbara K. Rothman (1988) remarked that many women said they choose to abort because "they didn't have a choice". What is highlighted is the sense of *personal* responsibility that women experience. Having to accept the responsibility for choosing to test or not to test, to abort or not to abort is difficult for women especially given that they *want* to be pregnant. The situation gives rise to what Rothman (1988) has called "the tentative pregnancy". When a woman experiences a tentative pregnancy she tries to avoid emotional ties with the fetus until after she is certain that it is fine and will not have to be aborted. She is caught in a limbo where she is pregnant but is unsure if the pregnancy will continue, unsure of whether or not to tell family and friends, unsure of whether or not she will soon be a mother. Her experience of the fetus is mediated by technology. A woman who does not "chose" to use technology is confronted with the "danger of death and disability...as the price to be paid" (Oakley, 1987: 49). This perception of the woman as detrimental to her fetus refocuses our attention onto the desire of

women to be "normal" in particular to how the concept of normalcy goes beyond what is "natural". The normal mother is the good mother who submits to the authority of the physician. The physician is cast in the role of knowing more about a woman's body and her fetus than the woman herself and as "protector" of the fetus and its rights (Rothman, 1987). A "collusive" relationship is formed in which the mother is regarded simply as the receptacle of a life form and wherein physiological demands on her are thought secondary to the perceived welfare of the fetus(Finkelstein & Clough, 1983). The legal ideological understanding of this medical-political process is that it is legitimate to control pregnancy and birth in the name of fetal protection (Law Reform Commission of Canada, 1989).

Using the law as a tool for reproductive technologies to protect fetal rights not only forces the woman to adhere to particular set of behaviour but has an affect on the way that the physician perceives his or her role as well. Lippman writes of "defensive medicine" which highlights the tendency to make clinical decisions based on the possibility of a future lawsuit. In the wake of "wrongful life" suits brought against physicians in the United States and the skyrocketing rate of malpractice insurance for ob/gyns the routinization of prenatal diagnosis can be seen as a way for physicians to limit their own sense of risk. Thus PND is used by physicians who simply want to comply with perceived medical-legal standards of care rather than to reduce birth disorders or to help women control the "quality" of children they bear (Lippman, 1989).

There are limits to what a woman can reasonable be required to do to ensure the health of a wanted fetus; women have a right to

body integrity and the right to informed consent should override considerations of the fetus. However, some advocates of mandatory genetic screening believe that social problems favour the collective interest over the interests of the individual. Thus while a woman may be undaunted by the prospect of caring for a handicapped child, state interests in the problems of population control, the general welfare of the population and the high cost of supporting the handicapped create a justification for the infringement of an individual's civil liberties (Smith, 1985: 442-3). Legal recognition of this justification is evident in recent developments in legal doctrine that allow interference with a pregnant woman's autonomy wherever her behaviour is seen to be detrimental to the fetus she carries. Increasingly, academics and judges have taken the view that pregnancy results in diminished juridical capacity and that pregnant women are not entitled to all the rights and privileges as other individuals (Rodgers, 1989: 174). This trend has had some very unsettling outcomes especially in the United States but in Canada as well. The courts have been used by physicians to limit women's choices of health care options. Several court orders have been obtained by physicians claiming that women could not give birth vaginally, had refused to sign consent forms for caesarian delivery, and were endangering the baby. In nearly every case cited, the women eventually gave birth vaginally, the babies were healthy and the physicians had been, quite simply, mistaken (Roth, 1993). In Canada fetuses have been made wards of the courts under provisions of the Child and Family Services Act and the Mental Health Act. Women have also been charged with criminal injury allegedly

committed against their fetus (against themselves?). The possibility has been raised of prosecuting or incarcerating pregnant women for smoking, social drinking, strenuous physical activity and having sexual relations against the doctor's advice. (Shevory, 1993). While actual numbers are small what is disturbing is the trend here in which the rights of the would-be mother are perceived as opposing the rights of the fetus. The right of the fetus to be born normal then can be seen as overriding a woman's right to make her own reproductive and family planning choices. Prenatal diagnosis is portrayed as offering women choice in their reproductive decisions yet women are concurrently portrayed as unable to distinguish rationally between their choices (Sutton, 1990). The only good choice, the proper choice, is to not give birth to a child with a disability or a genetic disorder.

5.4 (3) Medical values and attitudes

> "The Canadian Disability Rights Council maintains that the medical model or paradigm of health, disease, and disability, which views biology as destiny, contributes to a perception of persons with disabilities as passive, imperfect beings in a society that admires perfection" (Milner, 1993: 463).

The influence of the woman's physician is an important indicator as to whether she will be tested and is also an important influence on her decision to abort or not. Referral for amnio is related to the physicians' attitudes toward abortion which in turn is related to age, gender and religion (Bernhardt & Bannerman, 1982)

Attitudes toward abortion vary between different anomalies. In a study in France, 78% of physicians approved of abortion for Down's syndrome and only 46% for cystic fibrosis (Julian et al.,

1989). Weitz found in the U. S. that 85% of physicians approved of abortion for Down's (missing info). In Quebec, 71% approved for Down's and 46% for cystic fibrosis and there was still much debate surrounding the use of abortion for gonosomique disorders and heart disease. In addition, 46% of physicians in Quebec agreed with the statement that "Giving birth intentionally to a child with a genetic defect at a time when both prenatal diagnosis and abortion are available is socially irresponsible" (Renaud et al., 1993)

The majority of these studies questioned physicians on the use of amniocentesis, a procedure which is performed between the 16th and 18th week of pregnancy. The cultivation of karotypes takes between 2 and 6 weeks. Therefore any abortion done on the basis of results of these tests are going to be late ones. Preliminary studies show that some physicians opposed to abortion at such a late date may change their minds if chorionic villus sampling becomes widely available thus allowing for first trimester abortions. CVS is ideally performed between 9 and 12 weeks (Perry et al., 1985) Julian et al (1989) state that 30% of physicians opposed to abortion on the basis of amnio would change their positions is CVS was available.

The influence of physicians attitudes is expected to be diffused through the criterion of nondirective counseling. The prevailing belief is that genetic counseling either by physician or by trained counselor should correspond to a norm of nondirectivity that allows the woman or the couple to come to their own decision. Even if a couple were to ask, as they often do, What would you do in my position, counselors are not supposed to respond with their own opinion. Julian et al (1989) found that only 33% of physicians would

give their own opinion if asked. Variables influencing the likelihood of giving one's opinion include age, gender and specialty. Older, male obstetricians or general practitioners were more apt to give their opinion (Julian et al., 1989; Renaud et al., 1993). As noted above, younger physicians are more likely to refer women to amniocentesis perhaps an indication of their stronger belief in the importance of autonomous decision-making by women.

5.4 (4) Geneticization

Geneticization, as previously discussed, is a process by which individual differences are defined as genetic in origin with little or no relevance given to competing theoretical perspectives. As Nelkin and Tancredi (1989) make clear, diagnostics have found their way into all areas of our lives with alarming results. They show how genetic information is used to establish norms for individuals and how the norms and the techniques take precedence over any individual person. Thus people become subject to the test rather than the test serving individuals. This translates into workplace policy that puts the impetus on the worker to opt out of a work environment that may be hazardous rather than on the employer to clean up the workplace for all workers (Holtzman, 1989). This is also true of workplace "fetal-protection" policies which are aimed at excluding women of childbearing age from jobs involving exposure to toxins or situations dangerous to a developing fetus (Gonen, 1993). Genes are seen as the root of everything whereas other factors, especially environmental ones, are ignored.

Geneticization produces simplistic explanation for complicated phenomena. As Hubbard & Wald state:

> "Social and economic circumstances affect our body states and also shape the ways we perceive and categorize them. Biology cannot be separated from social and economic realities, because they are intertwined in complex ways and build upon each other. We cannot isolate the biological factors, and when we try we oversimplify and distort reality" (1993: 58).

Often at the forefront of the crusade for genetic explanation are the economic concerns of interested parties.

5.4 (5) Economic concerns

There is strong concern over the issue of economic considerations in the biotechnology industry; an industry that exists to secure profits from research. This profit motive worries critics of genetic technology who feel that individual concerns, concerns with insurance, with health care and with stigmatization, take a back seat to the business aspect of technology.

Insurance companies also stand to gain from the institutionalization of genetic testing. Health insurance is a growing problem in the Untied States as citizens depend on private insurance companies for their health care needs. Nevertheless, Canadians have a legitimate concern with regards to their own health care system, and also with the business of life insurance. Life insurance firms have begun to explore the possibility of using genetic tests as an aid to the assessment of mortality risks. According to the vice president of the Canadian Life and Health Insurance Association, insurance companies are entitled to all the information that the individual has and that includes any information about genes that may predispose a person to a particular ailment (Bueckert, 1993). Insurance companies don't want to insure people who are at a greater than

average risk of dying. Dr. Ronald Worton, chief geneticist at Toronto's Hospital for Sick Children and head of the Canadian Genome Analysis and Technology project has stated for the record that insurance companies should be worried if they are not able to gain access to individual genetic profiles. He states that "it could destroy the life insurance industry in the sense that people who got bad news from their gene screen would load up on insurance and those who got good news would either not insure themselves or wait until later in life" (Bueckert, 1993). In the United States, health and life insurance have been denied to individuals who carried particular genetic disorders. In 1989, Neil Holtzman listed nine conditions for which insurers had denied medical or disability insurance, including sickle-cell anemia, Down Syndrome, Huntington and type-1 diabetes (cited in Hubbard & Ward, 1993: 141). Other genetic conditions considered unacceptable of medical or disability coverage include autism, spina bifida and duodenal or gastric ulcer (Nelkin & Tancredi, 1989). The denial of benefits also affects children who are normally covered on their parents policies. In addition, women carrying fetuses diagnosed as having a genetic disorder have been denied medical coverage for their pregnancy, delivery and care of the child after it's birth (Hubbard & Ward, 1993). Billings et al. (1992) found that genetic discrimination does exist and is manifested mostly in the health and life insurance industries.

While Canadians have socialized Medicare, the government makes choices about what it will fund. In the past few years several services have been de-listed and thus are no longer available through the Medicare system but must be paid for out of pocket by

the individual. In addition, social services have experienced cut-backs and this can have a profound effect on persons with disabilities and their families. The Quebec government was able to save some $6 million dollars from 1991 to 1993 by closing two schools for intellectually handicapped adults. The closures meant that 350 students no longer had anywhere to go during the day when most of their parents were at work (Baker, 1991) We have to position this against the rhetoric that the government has no policy of encouraging people to screen and abort fetuses with Down's syndrome. Yet the government health care policy continues to pay for amniocentesis. Faced with the uncertainty about where their future children will be educated and where they will live as adults, parents choices become more limited. Financial considerations do play a part in a couple's decision making, even when they do not have to worry about paying for health care. Statistics from 1984 show that whereas 45% of women with children aged 0-6 worked outside the home, this decreased to 22.5% when the child has a disability (these figures would be higher in 1994 where 65% of women with children under age 6 worked outside the home)(Barden, 1984). Couples must consider how they will afford to care for their children, especially if the children in question need special care. This can be no small task for some families. In November of 1993, the Quebec Health Minister, Marc-Yvan Côté, outlined a plan that would have seen hospital outpatients paying out of pocket for medications that had been available free or for a minimal charge Individuals with CF were facing costs of between $5,000 and $14,000 a year for their medication (Thahn Ha, 1993a; Wells & Thahn Ha, 1993).

Following widespread outcries and condemnation, the minister rescinded his plan (Thahn Ha, 1993b). This example, however, points to the underlying motivation of many implicit therein, specifically, that governments must make financial choices. PND appears to offer an easy solution to this fiscal responsibility, opening what Troy Duster (1990) has called the *Backdoor to Eugenics.*

5.4 (6) Sex Selection

There are concerns world wide about the use of amniocentesis and other PND technologies for the use of sex selection of fetuses. This concern revolves primarily around the fact that male children enjoy a world wide preference and allowing sex selection would be tantamount to "Gynicide"; the large scale abortion of female fetuses, which would tip the sex ratio balance (see Corea, 1985; Arditti et al., 1989; Rowland, 1992 for a seminal treatment of this subject).

While a slight preference for male children has been indicated in Canada, this preference is usually expressed for a first-born child and this pro-son bias exists among potential fathers and not mothers. Subsequent pregnancies result in a variety of preferences, with most couples having more than one child expressing a strong preference for children of both sex (Thomas, 1993). Thomas (1993) also found that few of the individuals who were aware of sex selection techniques and who approved of them in principle would be will to use the techniques themselves. Thus the expression of a sex preference does not immediately translate into a willingness to use technology to that end.

Miller and Fraser (1993) conducted a study of the attitudes of genetic counselors with respect to sex selection. They found that only 2% of genetic counselors approved of PND for sex preference. It seems highly unlikely that sex selection techniques will become a major factor in PND as few people would willingly abort a fetus based on sex preference for non-medical reasons.

Sex selection for medical reasons falls into a separate realm. Disorders such as hemophilia and Duchenne's muscular dystrophy affect only male fetuses. Couples are able to use PND technologies for sex selection but this is a secondary motivation; their motivation is not to avoid male children per se, but rather to avoid male children because they have a 50-50 chance of carrying a disorder. For these couples, risk factors indicate that they have only female children to avoid genetic disorders.

5.4 (7) PND for late onset disorders

PND for late onset disorders presents a particular set of dilemmas for counselors and individuals alike. Perhaps the most well known late onset disorder to present this dilemma is Huntington Disease. Huntington Disease usually manifests between thirty and fifty years of age; it is a degenerative disorder that may last a decade or more during which a the affected person gradually becomes totally incapacitated and unable to communicate culminating in death (Post, 1992; Terrenoire, 1992). Each child of an affected person carries a fifty percent chance of inheriting the gene responsible for the disorder and thereby inheriting the disease itself. Unfortunately, by the time most affected people become affected, they have already had children.

Given that there is no treatment for HD, the protocol for detection is based on alleviating the psychological distress that a positive diagnosis will generate and enabling individuals to deal in a positive manner with life events.

With regards to prenatal diagnosis, the Canadian Collaborative Study of Predictive Testing for Huntington's Disease (CSHD), which involves centres across Canada, "specifically states that predictive testing should not be performed for minor children, nor prenatally in cases where parents do not consider termination of pregnancy as an option" (Cooke, 1993: 3). This stance contradicts non-directive approach of genetic counseling which generally makes no suggestions to couples concerning their decision making. This contradiction exists *because* of the nature of HD: as Post points out: " ...a life of thirty or fifty years duration is potentially a fully good one" (1992: 77). Furthermore, counselors feel that only the individual him or herself can make the decision to be tested; this decision can only be made when the individual is an adult and understands the full implications of the decision and the disorder: "...testing may be disadvantageous for the child, either because of possible distortion of parent/child or sib/sib relationships or because of limitation of resources for the child shown to be at risk. The self-esteem and sense of worth of a developing child may be profoundly and negatively affected" (Bloch & Hayden, 1990: 2 cited in Thomas, 1993).

Prenatal testing for late onset disorders is of value in two circumstances: firstly, if parents are prepared to abort on the basis of a positive test, secondly, if there exists a treatment that may cure or alleviate the symptoms of the disorder.

5.4 (8) PND for genetic susceptibility

Screening for genetic susceptibility for common diseases could have benefits but these benefits may be outweighed by their disadvantages. Benefits include allowing individuals to take preventive measures; disadvantages include discrimination against carriers by employers, insurance companies, or peers; harm to self-image and happiness; damage to parent-child interactions; and stigmatization (Prior, 1993: 41).

Pinpointing an individual as being genetically susceptible to a particular disorder is a factor in geneticization or the individuation of health care issues as discussed above. Susceptibility genes are linked to such common diseases as lung cancer, heart disease and mental illness. However, the expression of a particular cancer is also strongly linked to environmental factors and the exact interplay of biology and environment exists in an unknown quantity. The danger is in the potential overemphasis on genetic factors to the detriment of environmental ones such that individuals are expected to make changes whereas environmental damages can be ignored. The impetus then falls on the individual to make "life-style" changes to lower their risk of disease; changes such as inappropriate diet, lack of exercise and smoking. Nevertheless, as Hubbard and Wald (1993) point out, life-style factors are not always the result of free choice but rather are affected by economic, cultural and other social factors. Lower income individuals, for example, may have difficulty maintaining a healthy diet and will not have funds to joining exercise programs or gyms. The focus on life -style and an "orientation toward genetic susceptibility can easily begin to dominate our ways

of thinking about disease prevention, until it becomes *the way* that we come to think about the problem: (Duster, 1990: 123, italics in original).

There is danger also in work place discrimination wherein some individuals will face unemployment because they are considered more likely to develop a disease in a carcinogenic environment. This has already led some companies, such as Dupont, to instigate workplace restrictions on women of reproductive age (Gonen, 1993). This is reminiscent to the 1970s wherein carriers of the sickle cell gene were banned from the US. Air Force on the grounds they were at greater than normal risk for becoming dizzy and disoriented at high altitudes (Holtzman, 1989). Testing for genetic susceptibility carries with it complex moral, legal and social criterion that must be adequately understood. As noted by Billings et al. "stigmatization, and denial of services or entitlements to individuals who have a genetic diagnosis but who are asymptomatic or who will never become significantly impaired" does exist and must be dealt with in order to avoid creating a social underclass based on genetic discrimination (1992: 476).

5.4 (9) Somatic and germ line therapy

In human gene therapy a genetic defect resulting from an alteration in the DNA of a specific gene is corrected by inserting a normal DNA sequence for that gene into the cells of the patient (Prior, 1993b: 234). There are two types of gene therapy: somatic and germ line gene therapy. The somatic or body cells are non reproductive therefore the alteration is not inherited by the offspring of the

individual treated. Germ line cells are the reproductive cells; any change invoked will be passed on to future generation.

Somatic gene therapy is the only type feasible at the present time. It involves gene insertion whereby a normal version of a gene is introduced into the chromosomes of an affected cell. The insertion process can take one or more of several techniques: viruses, microinjection, physical and chemical treatments and membrane fusion. This treatment is only feasible for recessive mutations and not for dominant disorders (Wills, 1991). To date it has been used experimentally on such disorders as CF, ADA, Lesch-Nyhan syndrome, PKU and some cancers, with hopes that it will one day provide a cure for AIDS (Wills, 1991; Prior, 1993b).

Whereas somatic germ therapy is relatively unproblematic in that it incurs no strong moral or legal dilemmas, the same is not true of germ line gene therapy. Even though germ line gene therapy is not yet feasible, there is considerable debate surrounding it's moral and legal status. The concerns centre around two main queries: firstly, that it would produce changes in the genetic material that would be passed on to future generations and would therefore affect the human gene pool [by decreasing genetic diversity]; and secondly, subsequent generations could be exposed to risks that could not be foreseen by the results of testing in experimental animals (Prior, 1993b).

In response to concerns regarding the tampering with the very essence of humanity, Munson & Davis claim that "medicine itself has a prima facie moral obligation to pursue and employ germ line gene therapy" (1992: 153). The authors equate germ line therapy with

positive eugenics and argue that it is in our best interests as humans to "transmit the capability to live a superior kind of life" to our offspring (1992: 151). Rosenkranz (1987) affirms that we may have a moral duty to genetically engineer our children. Harris (1993) concurs, establishing a moral compunction of parents to use germ line therapy to ensure the well being of any offspring. This compunction includes the use of gene therapy which could "retard the aging process and so lead to greater health longevity, or which might remove predispositions to heart disease or which would destroy carcinogens or maybe permit human being to tolerate other environmental pollutants" (Harris, 1993: 183). Harris feels that parents owe their children the best chance in life as possible and this includes arming them genetically to exist in a world that has been environmentally contaminated by humans.

Critics of germ line therapy worry that tampering with reproductive cells amounts to "playing God" and will have unforeseen and perhaps unwelcome consequences. The permanence of the changes poses problems as it is not unusual for medical therapies to have unanticipated and undesirable effects. To date these unanticipated side effects, such as those with DES or thalidomide have not had a permanent effect on humans but side effects associated with germ line gene therapy would be permanent (Hubbard & Wald, 1993: 114).

Although there are concerns that germ line gene therapy would result in a decrease in genetic diversity, Shapiro (1991) takes on opposing stance. He postulates that germ line therapy decisions would be make on a local basis and would therefore result in

worldwide diversity: "The options made available by germ line therapy may produce a new division of the human race...based upon freely selected values" (1991: 373). To do otherwise, i.e. to prohibit germ line therapy would necessitate a global police state to enforce genetic prohibitions. Following his proposed scenario, those opposed to germ line therapy could opt out: "They should certainly be allowed to act upon their beliefs within their own nations, although not to enforce their opinions on others" (Shapiro, 1991: 373).

Granted, germ line gene therapy is not yet feasible, nevertheless it is important to examine the moral, social, ethical and legal issues in advance of scientific progress to prevent our technical capabilities from overriding these concerns. Maurice A.M. De Wachter (1993) examined reports dealing with germ line gene therapy from European countries and found that their dealings with the subject matter were sparse, they preferred to confine the analysis to somatic gene therapy since it is already a scientific possibility. He notes that attention to germ line therapy "almost invariably leads to rejection" (1993: 167). Most conferences and working groups to address the subject felt that there were too many gaps in knowledge to enable sound decision making to take place. De Wachter concludes that "the deficiencies of our present knowledge make it impossible to clarify the morality of germ line therapy" (1993: 175). Germ line gene therapy is an issue about which there will be much future debate.

Conclusion

Prenatal diagnosis exists within a broad context of reproductive technologies. Within this vast area there is extensive debate

regarding the moral and ethical ramifications of particular technologies. The criticisms surrounding PND revolves around a mass of information that is difficult to synthesize as it spans the continuum from women's status in society to concerns with the very essence of what it means to be human. Economic concerns, medical values and social attitudes are all part and parcel of the vast amounts of knowledge that comes into focus when one addresses PND. The overriding conclusion is that continued, ongoing debate and discussion is needed with the participation of all members of society who share in the concern for the future.

The Royal Commission on Reproductive Technologies was a massive research undertaking. It was set up in an attempt to consolidate knowledge and opinion on the new advances in medical innovation before such advances could no longer be confined to socially desirable boundaries. The Commission dealt with such a vast array of subject matter that it is next to impossible to given a short concise summary. Suffice to state that the Commission dealt with every facet and level of reproductive technologies, including that of prenatal diagnosis and genetics.

The very act of establishing such a commission points to the fact that medical innovation is a social issue in that it touches upon society in such a way as to transform ways of seeing and doing. It was precisely to guide this transformation that the commission issued a lengthy series of recommendations. The impetus was to ensure that society's values are reflected in the medical techniques that are legitimize by their very use.

With regards to prenatal diagnosis and genetics, the major recommendations centred around the licencing and monitoring of prenatal diagnosis facilities. No limits were placed on the establishment of prenatal diagnosis programs; no suggestions were made as to whether population screening programs should form an integral part of health care services. The absence of any such prohibitions clears the way for such provision of prenatal diagnostic services to eventually become a part of the health care services offered to individuals. Therefore it becomes even more imperative that ongoing study and monitoring with experimental trials be closely watched.

Chapter 6

Results and discussion

This chapter presents an analysis and discussion of the findings of this research. The main purpose of the research is to investigate Cystic Fibrosis as a case study through interviews with experts in field. As a way of managing information, three broad categories or areas of interest were created: what living with CF entails on a daily basis, the social meaning of genetic testing, and issues related to the provision of health care. These three general categories are placed within a theoretical perspective of social change theory and are enlightened by theoretical perspectives provided by a Foucauldian analysis of the body.

6.1 Living with Cystic Fibrosis

The meaning of living with CF was explored with members of a team devoted to caring for individuals with CF. This team consists of physicians, nurses, physiotherapists, social workers and genetic counselors. The following information is garnered directly from these interviews. Direct quotations are neither used nor attributed in this section only due to the nature of the information; it appeared best in terms of readability of the information. As well, information was factual as opposed to the expression of opinions, beliefs and attitudes and as such these facts are presented by several of the respondents. The data is contained in the interviews with respondents numbered three (3), four (4), six (6), seven (7), eight (8) and nine (9).

Approximately 2860 individuals with Cystic Fibrosis are registered with the Canadian Patient Data Registry of the Canadian Cystic Fibrosis Foundation (personal communication, Ian MacKintosh, 29 Sept. 1994). These individuals are treated at 32 clinics country-wide. Nearly half of the total population with CF are treated at three hospitals who serve primarily children: five hundred patients are treated at the largest hospital--Toronto's Hospital for Sick Children; the second largest clinic, Ste. Justine children's hospital in Montreal, treats about 250 patients; the Montreal Children's Hospital treats around 100 children. When a CF patient turns 18 they are then treated at separate Adult clinics [where available]. The Adult Clinics are established as departments in other hospitals for example, in Montreal, the CF Adult clinic is a department at Hôpital Hôtel Dieu.

Treatment of CF includes chest percussion to improve clearance of the mucous that builds up in the lungs, antibiotics to fight infection, pancreatic enzyme treatment (usually in the form of pills or capsules taken with food), and strict attention to nutrition. Treatment is ongoing and will continue throughout the individuals entire life as there is no cure for cystic fibrosis. The parents of affected children have an important part to play in their children's treatment as the disease is managed for the most part at home with periodic admittance to hospital for intensive treatments or to fight particularly dangerous infection. A staff team at a CF clinic is generally led by a paediatrician and includes a nurse, a physiotherapist interested in sports medicine and lung function, a dietitian, a psychologist and a social worker. The emphasis is placed on helping the person with CF to live as normal a life as possible.

The daily routine for the parents and the child is not a difficult one, so much as it is time consuming. Treatment is given one to three times per day and is done in three phases. First is a ventalin treatment which takes about ten to fifteen minutes and which is designed to help clear the lungs. Following the ventalin is the physiotherapy which is the percussion treatment in which the parent physically tries to loosen the secretions to help the child cough them up. This is done by putting the child into different positions and clapping them with a cupped hand. The treatment is not at all painful and the children are taught different breathing techniques so they can better bring up the secretions. This takes about twenty to thirty minutes and is followed by an oral or an inhaled antibiotic. The antibiotic is given when the lungs are the most clear because they work more efficiently at that time. The entire routine takes between sixty and ninety minutes. It is difficult for families to organize the treatment around work and school. The hospital team works with them to create a schedule the family can adhere to. The more severely ill children follow these treatments three times a day. About 5% of children are unable to attend school. Roughly 95% of all CF kids attend school regularly. Younger children can attend daycare and the hospital teams have worked with daycare workers to teach them the process of the treatments. In other cases, they have worked with baby-sitters, volunteers or friends. It is stressed by the hospital team that the treatment is not complicated and anyone can be taught to do it.

Most children with CF are on replacement enzymes because their pancreas is unable to secrete the enzymes necessary for

digestion. These oral replacements are taken with meals. Children may also take vitamins. While a few children don't need the replacement enzymes, most of them carry medications with them everywhere they go. Not all children are on all the medications, antibiotics, ventalin etc. but most children use some medications.

Children with CF do not have to follow a special restrictive diet, in fact they are encouraged to eat lots of anything and everything as weight gain is a problem for them. They can eat foods that are considered "unhealthy" in a non CF afflicted person, e.g.. high fat foods. The dietitian plays an important role because there is a danger that a child can become malnourished and be unable to fight off infection. In addition, malnourishment can prevent a child from reaching puberty and will restrict natural growth into adulthood. Infants have often been diagnosed as having CF after first being diagnosed as "failure -to-thrive", no matter how much they eat they are unable to gain weight. A few children will take a nutritional supplement, *Ensure Plus* which helps to prevent malnourishment.

Families do not contend with many financial costs but they must contend with the emotional and physical costs of the ongoing treatment. The effects on the families of having a child with a disease in the family are varied. As with all chronic illnesses, whether genetic or not, congenital or not, the divorce rate is higher than it is for the general population. But many families jump right into treatment and are very conscientious about the routine and work hard to make sure their child will be in good health when the cure is found. Because most families are hoping for a cure. Some families go overboard and the child gets *too much* treatment. Over

treatment results when a child is treated excessively with drugs and/or therapy both of which can be deleterious in excessive amounts. Over treatment with drugs is unhealthy as the body must work to process the excess; overtreatment of therapy drains parental energy and leaves the child with little time to partake of normal activities, while producing no extra beneficial results. As such, illness then becomes the sole definitive characteristic of the child. In some cases the child doesn't get enough treatment, the parents do the minimum possible. These latter type of parents are thought to be ignoring the situation either because they are too tired to do treatment or because they don't want to believe that their child is really very ill.

If an individual has a bad infection that won't clear they are admitted to hospital for a clean up. Winters are worse because everyone gets colds and the end of the school year can be a time of high admission because children are worn out. Some adolescents admit themselves in late August before the school year begins again because they want to be in good shape for the coming term. There they receive IV antibiotic therapy and postural drainage three times a day with the physiotherapist. Treatment usually requires ten to fourteen days. Only 7 to 10% of the CF population is regularly hospitalized, the other 90% only show up for regular clinic visits. In the routine visit, the patients are monitored. In addition, every six months X-Rays are done to help the team to identify any deterioration as soon as possible in order to modify the treatment. Although only a small percentage are hospitalized, some of these children never make it to a regular clinic visit. Instead they are

admitted to hospital and after the fourteen day treatment are given a follow up appointment at the clinic in one month's time but are readmitted to hospital before they can make it to the clinic. This is striking proof that the disease is variable; some children are very ill and others are actually quite well.

6.1 (1) Medical costs

The monthly bill for one person with CF is about three thousand dollars. These costs can reach upwards of four or five thousand dollars depending on the severity of the individual case. These costs do not factor in hospitalization costs. Cost itself is a social issue in Canada with Medicare. Cost/benefit analyses could be based on the ratio of treatment costs to the costs of screening and abortion.

The costs of the medications are subsidized by the government. Each subscription costs two dollars to a maximum of eight dollars per month regardless of family income. The Canadian Cystic Fibrosis Foundation fought for ten years by lobbying the government to get this help. Some families have health insurance but for those who don't the hospital will offer additional subsidies. People who receive welfare payments are covered by the health insurance made available to them and it would typically be the working poor who would need additional help* . No one ever goes without their necessary medication and most of the medication is distributed by the hospital pharmacy which is able to buy in quantity and thus able to minimize costs. Parents also receive a supplement to the family allowance of one hundred dollars per month to help defray the costs

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* This information from interviews was given before the establishment of the province wide drug insurance plan in Quebec created by health minister Guy Rochon in 1996.

of equipment or travel expenses. These supplements are available to all children with a handicap or disability. Personnel at the Montreal Children's hospital stressed that the monetary issue was not an issue per se here in Canada in the same way as it is in the United States. In the US. private health insurance present many obstacles to parents trying to pay the high costs of medication. In Canada, the issue is that of *agency* in that problems arise with regards to jurisdiction and responsibility. For the most part, however, any problems arising are dealt with by the medical community and never become an issue for the family as they do not deal directly with the pharmacy. The CF team intervenes on behalf of the families as a group.

6.1 (2) Social construction of illness

The meaning of illness can be socially constructed by the actors involved. As noted above, some parents have a tendency to over-treat their children whereas others under-treat their children. In this way, the severity of CF is constructed and managed by the parent regardless of the true physical manifestation of illness in the child. In such a way, parents can create a meaning for the disease: they can create CF as a disease that necessitates constant treatment and management or they can create CF as a disease that needs little care. So too, the adolescents themselves have leeway in terms of hospitalization: they can admit themselves to the hospital for preventive treatment. Additionally, physicians construct meaning for CF.

> Kids come every two or three months regularly. Every time they come, they make an appointment for the next

> time. Most kids, the physicians feel that the two month interval is the optimal, I don't know how they came to that, *I think part of it is also income.* If it was three months it wouldn't make that much difference for some kids. (my emphasis) (R6).

The reference is to the physicians' income. This respondent felt that physicians had decided on a two month interval to insure their own level of income and not for reasons of health care. This indicates how illness is constructed *socially*. Specifically, although it appears CF is an illness that requires monitoring at two month intervals, this interval could be extended without compromising health care if physicians were not concerned with establishing a two month interval billing procedure. An illness that requires visits to the hospital six times a year may be perceived differently than one that requires hospital visits four times per year. The social perception of CF is of utmost important for genetic testing and screening programs. When the social meaning of testing is explored, the severity of a disease is an indication of how important individuals will feel genetic testing or screening is. The severity of CF is variable and very severely affected children may die despite efforts at treatment. It is difficult for the hospital team not to get into what they call a "denial phase" by focusing so much on the positive even though this focus on the positive is a necessary component of treatment. While the hospital team does not deny that CF is a life threatening degenerative disease, they try to focus on the improved life expectancy when they are dealing with parents, particularly newly diagnosed families. Families can use denial in a positive way to keep going with the treatments and hospital staff tries to maintain a high level of

awareness of the general state of mind of the various family members.

Conclusion

Because there is such a variable penetrance of CF, daily living with CF is different from individual to individual and the experience of CF varies from one family to the next. The daily care needed for a child with CF becomes a part of the family routine and according to the respondents, rarely results in undue hardship for family members. Members of the CF team interviewed all believed that caring for a child with CF was not much more difficult than caring for a child in general. Nevertheless, some children are severely affected and during the course of my interviews two children died from complications related to CF. The team was saddened by these deaths; the first deaths in nearly two years at that particular hospital.

Medical personnel describe different diseases almost as some children have diabetes, others have liver disease, others have a heavy gastrointestinal involvement, and some are pancreatic insufficient. A small group of children rarely need medical care and are infrequently present at the CF clinic so that the CF team does not know them or their family very well. Different families therefore perceive the risks on very different levels. This discrepancy is of importance because it highlights the difficulties in establishing common understanding about CF. This lack of commonalty of experience may be one factor in the individual's decision making process regarding genetic tests and may be of utmost importance in the establishment of a genetic screening program. Where the power to describe and delineate the limits and parameters of CF is seated

will have a great impact on uptake levels of genetic screening. How society constructs meaning for illness in general and for CF in particular is a dialectical relationship. This subject will be discussed in further detail below.

6.2 The social meaning of genetic testing and screening

The social meaning of genetic testing and screening encompasses several areas of interest. Many issues are explored within this broad category. Firstly, the way in which CF is portrayed an interesting and charged topic. Secondly, an analysis of links between testing for genetic status and abortion of an affected fetus is necessary. Thirdly, the way in which individuals understand the concept of risk and their own risk in particular is especially vital for CF with its' high false negative tests. Fourthly, debates over who should be offered testing and whether carrier screening should be the norm are central to this discussion.

6.2 (1) Portrayal of the Disease

The variance in the severity of CF is a striking feature of the disorder and a key issue in genetic screening.

> I think we could describe different diseases almost, some kids have diabetes or some have a heavy GI involvement, they have liver disease and stuff like that and there's other kids we see that are quite healthy and at 18 we transfer them off. You could never pick them out as being any different from anybody else. So there's a lot of differences. (6).

The understanding that parents of children with CF have of the disorder is heavily influenced by their own child's health status.

> ...you're going to have very different views from a parent whose child died very young versus parents whose child is living to 22 and only been in the hospital once or twice...(R4).

Regardless of, or perhaps because of, the different experiences that people have with CF, how the disorder is portrayed to the general public, and more specifically to individuals contemplating either prenatal testing or carrier testing, is of utmost concern to parents of affected children, professionals working with those children and groups such as the Canadian Cystic Fibrosis Foundation. The concern is that of balancing the portrayal of the disease in order to give an "accurate" account. But who will determine what is accurate and how will a consensus be reached (if indeed, that is a goal)? The issue of who will have the power to define Cystic Fibrosis to the general public is an important one to the CF community. This has been an issue in previous years in relation to fund raising campaigns wherein the organizers have to make sure that children look sick enough to need funds but not to distort the reality of their lives and to make them victims of misguided pity.

> The Cystic Fibrosis Foundation does a good job of raising money, but they talk about how sick kids get and they have to, every group does. And so people say, why don't they show healthy kids playing hockey and the answer is then no one will donate! so there is some of that feeling, even in terms of fundraising efforts, is that we show kids who are sick. It's a breath of life, *sick but cute, cute and pretty sick!* So of course, it would have to be done and some people worry that it makes their child look sicker than they are. (R8).

To this end, the discussion revolves around whether the disorder is and should be portrayed as a "killer" disease, or if it

should be portrayed as a disorder that cannot be cured but can be controlled.

> It's very hard to explain to anybody what it must be like to have a child with CF...I find even among a number of physicians you see a wide range of variability and how they perceive the burden of the disease. There are some that will say look we can keep half of them alive on the average till they're almost thirty and the other position will say it's not good enough. How would you like it if you knew your child was only going to live till 30. (R17).

The uncertainty with which the general public approaches CF may mean that they will have a more negative understanding of the disease than will those with some experience with the disease. This can be important in terms of preconception counseling and carrier screening.

> I think the unknown is always more frightening. We talk about illness, illness always has a negative connotation. I try to be careful when I'm talking to people in a counseling session to not use words like sickness or disease, I try to use the word condition...no matter what you want to have unbiased counseling and you want to give people unbiased information but the use of a word like disease or disorder could weight things a certain way (R4).

Thus, in order to preserve the unbiased nature of counseling, the disorder, or condition must be discussed in an almost benign manner. There is no talk even of disease or disorder. The term condition implies something that requires some care but that is not as serious as a disease may be perceived to be. It incorporates the less serious nature of CF.

> ..you're saying that with the treatment now versus when that child [was] born, the treatment is obviously better now, the children are obviously living longer but it's extending the inevitable basically still. We don't have a cure, we're just avoiding dying young, but kids still die...(R3).

Children are dying of CF but this cannot be the only image of the "condition" received by individuals within the public. The counselors implicitly recognize their own power to influence individuals through discourse and value laden language. The counselors are in a position to construct reality for individuals seeking counseling. They try to adhere to a belief in non directive counseling by choosing neutral words but this does not change the basic fact that language does construct reality. Therefore, counselors are not avoiding influencing the individual by using words such as "condition" instead of "sickness" or "disease", they are constructing a different reality. Nevertheless, language is a tool that must be used. Whatever terms the counselors decide to use or discard, they are in the business of constructing reality for many individuals who have no prior knowledge of CF:

> ...the general public doesn't really know much about CF but if you say CF they automatically assume, yeah, that horrible, I wouldn't want to have a child with CF. They generally don't know someone who has it already and they probably assume it's worse than it is. (R17).

Yet, ethical concerns must mediate the way the disease is portrayed.

> ..you can't in order to help people with their decision making, take them to see a really sick kid and say this is what will happen, you can't. A, because of the privacy of the really sick kid and B, you could also take somebody, and this is a little crazy but not untrue, you could take somebody up to the ward where the head injury kids are and say this could happen to your child the next time it runs across the street. Because you don't know that it's going to happen. It may but it may not, [it's not] absolute...nobody knows at the beginning how it's going to be ...there is reason in CF to be hopeful, either for a cure or a significantly better treatment. (R8).

It is this balancing of the reality of an illness with the hopefulness of improved treatment that creates the dilemma. As many people expressed to me, CF is not like Tay Sachs, where the course of the disorder and therefore the decision making process is much more absolute.

> CF is very very different [than Tay Sachs]. These people [with CF] are not all people who are going to die in childhood, with Tay Sachs disease...there's no quality of life, this child is going to die and die young...It's a pathology...which...cannot be treated. (R14)

Professionals who work closely with parents who already have a child diagnosed with CF are especially careful about how the disorder is portrayed but at the same time they recognize the limitations inherent in what one nurse characterized as "positive denial".

> I think it's absolutely necessary that we try to portray it in a positive way because it's such a shock to families to have the diagnosis...We talk a lot more about the improved life expectancy...And there's a lot of research done and there's a lot of hope in terms of treatment. I

> think that's absolutely essential.... Our families do use denial in a positive way, I think it's very necessary in order to keep functioning. (R6).

Parents and professionals seek to achieve a balance wherein children are well cared for, their special needs are met but they are given hope for improved treatments and controls, if not cures, for the future. The power of discourse lies within a tense interplay between professional and family member. Families may seek to create their own sense of reality of living with a child with CF by using denial, constructing a discourse that creates of CF an illness that has hope for the future. Professionals actively encourage this use of denial, "healthy denial" and in doing so, they too have power to create reality through discourse. This dilemma highlights the fact that CF has a physical reality in the physical body, but it also has a social reality that is constructed and reconstructed by society. This reality is taken apart and reconstructed with the help of professionals for each individual case in a way that serves to help the individual family deal with their own particular experience of CF. The power to create reality through discourse resounds in the dialogue surrounding the choice to abort a fetus with a genetic disorder.

6.2 (2) Genetic testing/screening and abortion

The Canadian Cystic Fibrosis Foundation is a volunteer run organization whose constituency includes "Catholics and pro-choice elements" and they do not want to get involved in a debate about abortion. However, the CCFF is equally adamant in insisting that:

> even though this has not been formulated firmly in a policy sense...it would be reasonable to say that the foundation is dedicated heart and soul toward a cure and not screening the disease out....The foundation is dedicated to finding a cure for the disease...our mandate is to find a cure or a control for Cystic Fibrosis not to screen it out of the population...the foundation is here to find a cure for this disease.[To that end]all of the research we're funding has direct relevance to CF and to developing cures or control. (R20).

The primary reason for the CCFF's insistence on cure and control stems from their long history as not simply a volunteer organization, but a *parent* volunteer organization. The members of the CCFF are for the most part parents (and other relatives) of children with CF. They come to the organization for support from others who understand their experience and they stay to raise awareness and funds for the disease.

> The people who built this foundation are people who have children with CF and if you tell those people that it's a desirable goal to screen those people out of the population what you're telling them is that their children who they love desperately should not have been born. (R20).

Prenatal diagnosis of Cystic Fibrosis has traditionally been based on presentation of the disease in a family via the proband, i.e. a child born with the disease in the immediate or extended family. There has yet to be a population screening program to detect carriers before the presentation of the proband. Thus most research up to this point has been concentrated on the decision making of couples who already have at least one child with Cystic Fibrosis. Parents

have had to make reproductive choices subsequent to the birth of the proband. Parents may then feel that to actively avoid the birth of a child with CF is a commentary on the life of the existing child.

> It's hard with loyalties to their kids. You see a lower uptake in families who already have a child with CF and that child is four years old and sure they have CF but that's just part of their day (R4)

A family with a child with CF begins to see itself as a normal family and dealing with CF becomes routine rather than extraordinary. Given the affection they have for the existing child, it becomes difficult to willingly choose to avoid the birth of a similar child.

> That's the big issue with many families in terms of aborting a second child. You've got one and then how do you explain to that one that you would not have another one (R8).

> Families [are] put into a very difficult situation and ...the uptake of any of those kinds of services [PND and abortion] would clearly be limited because these families would have to say "How can we choose to abort the second one when we didn't the first one?"...it's a very hard decision (R11).

Another mitigating factor in this decision making is that CF *is* an extremely variable disease and the first child that the parents have who has CF may not be very ill. This is especially true when the first child is quite young and it is then that the parents will be deciding to have or not to have another child. If the child with CF seems rather healthy and the family is able to cope well with treatments, then they may feel able to take on the care of a second one if need be. And yet this leads back to what was previously

discussed as "positive denial", the denial needed so parents have the will to continue with treatments.

> It can be [when the first cystic child is young] it doesn't seen so bad...But ..they're exposed to kids who have been admitted so they've seen some of the kids. They have to be fooling themselves a little bit whether intentionally or not...I don't know if it's conscious or not, somewhere back there they must know but they just decide, they're going to go through with it {another pregnancy] and the odds aren't so bad and they really want to do it. And we have a few families with two kids [with CF] (R7)

People may misinterpret the odds in their own favour as a coping device or they may believe that CF isn't such a terrible disorder after all.

> The parents of a CF child for the most part don't want prenatal diagnosis, I know they are less likely to be advocates of screening, because their attitude is, well if you are suggesting that my appropriate move is to terminate a future pregnancy that suggests there is no hope for child number one...[One woman] said "who could look at my child here, (who was about four years old and running around as cute as a button) how can anybody look at my [son] and say his life isn't worth living...[Parents} get caught up in relating whatever you might talk about for a future pregnancy to the existing child and in general at the time that they may be thinking about a pregnancy again their children are young and relatively well, and CF kids when they are young are doing very well...[parents] are seeing their kid as basically a normal kid and [they're] just waiting for a cure...(R17).

The balancing of the knowledge that their child has a serious disease with the hope for a future cure, or at least a future management of the disease has a great impact of the perceptions of and attitudes of parents of children with Cystic Fibrosis.

Siblings attitudes toward prenatal diagnosis and abortion are very positive for the most part. It is hypothesized that siblings are greatly affected by the experience of living with an affected brother or sister, with witnessing the seemingly large burden of care that had to be expended by their parents, and perhaps by feelings of neglect when the bulk of family energies was spent on the affected child. In addition, siblings are greatly affected if a child dies from the disease.

> When it comes to brothers and sisters, they're the ones that are just dying to be tested, at least in our age group, people in their 20s at the moment have generally watched the demise of their brother or sister with CF over a long period of time, probably watched them die. In any case their perception of the burden of this disease is quite different than the parents of a young child...their perception of it is much more horrible (R17).

There has been a "split" in the population with regards to attitudes toward prenatal diagnosis and abortion for CF. This split is characterized by parents of affected children on one side and relatives, siblings and others on another side. Parents have been found to be unique in a sense in their low uptake of available services. The more distance one has from an individual with the disease, the more likely a person is to approve of PND and abortion. Without a "buffer" of experience with the disorder, the perception of risk may be greatly increased, along with the willingness to make use of genetic services by the general population.

The fact that CF has many different mutations resulting in many different levels of expression of the disorder is one vital difference between it and a disease such as Tay Sachs. Added to this

is the fact that parents can choose to deal with a child with CF in many different ways, such as overtreating, undertreating, using "positive denial": all of these various actions demonstrate that to a large extent the experience CF is socially created and not physically mandated in the way that Tay Sachs is. The perception of the disease can vary and therefore it is not surprising that the way in which individuals perceive risk of carrier status and risk of bearing a child with CF also varies considerably.

6.2 (3) Understanding risks

The ability to understand one's individual, statistical risks may exist on several different levels. This is one of the most contentious issues specific to CF given that the number of mutations will yield a high false negative rate. There was some discrepancy in the data as to whether people were able to understand the necessary statistical information and different views on how people would create meaning from that information and thereby be able to make meaningful choices regarding their reproductive behaviour. Some respondents clearly believed that most individuals could not understand the information presented to them regarding carrier status.

> The base rate for cystic fibrosis isn't well understood. The base rates are subject to a lot of misinterpretation. The base rate being the probability in the population. The base rate of carriers in the population is something that people don't really know. (R12)

The perception of what a risk means for each individual is different even if the statistical information is static.

> One of the things ...with genetics and genetic disease is the difficult concept of this whole issue of probability....if it's black and white they can deal with it but probability.... Some people think that a 25% risk is different from one in four, some people think one in four is a very small risk and some think one in four is a horrendous risk and they wouldn't take a chance. (R18).

Regardless of the information provided them, some people will continue to think that the risk for them is very low.

> People are good at understanding some things about risk but they think they have very low probability, they're not very good at calculating, there's a systematic bias depending always on what they're comparing their risk to. (R12)

Nevertheless, others believed that different levels of understanding were possible and that all levels were valid and acceptable.

> ...there are quite a few people we've seen that have quite minimal education levels, they might not understand all the details but it's not important for them to know the details. Really what's important for them to understand is, is there a risk, what is the risk and what can I do about it, what do I need to know? (R5)

In addition there is no consensus as to which is the best way to communicate probabilistic information nor on how people make decisions regarding genetic risks. This problem is especially acute when parents are being given information at what may be a very stressful time in their lives.

> [When the anxiety level is high] you just don't filter the information, or you think you have and when you get home...everything's gone. (R4)

This may arise quite simply because human beings are not accustomed to making complex decisions.

> ...it seems that as a species we're not hard wired into making choices that involve multiple variables with multiple levels of certainty associated with them. And so the natural tendency is to pretend that some of that uncertainty is resolved or to work on the basis of the best case scenario or the worse case scenario, limited to one narrow question when in fact there are multiple questions....We're at a point I believe where most of the discoveries of genetics involve the creation of uncertainty much more than they resolve the reduction of uncertainty....You've identified a fetus who is affected with ...CF...and what does that mean? Well, it means we don't know what to say, we don't know what to think and we certainly don't know what to do about it. (R1).

Given that this kind of complex decision making does involve the creation of uncertainty for most couples, it is much more difficult to ascertain in any scientific, objective manner how and why different couples undertake different decisions even if they are faced with the same level of risk.

Some women feel that any level of risk is too high for them.

> One family who had a child screened...were told that it wasn't 100% sure but 99% that the child was a carrier and the mother said the hell with damn statistics, my kid's got CF so the statistics don't mean a bloody thing to me....There's all kinds of statistics one can produce but it doesn't always mean anything. (R6)

This returns us to the literature which stated that individuals perceive a binary decision making process: either their child will have CF or their child will not have CF. The intricacies of statistical analyses don't have much meaning for most individuals. In addition,

individuals do not make their decisions in a vacuum. They do not simply respond to statistical analysis of their risk but rather to the interplay of risk with other events in their lives.

> [Perception of risk depends on] How they perceive their reproductive futures and to what extent they need to feel in control over their lives. The need for control is something that is instrumental. Many families are putting off having children, the average age of first childbirth is going up, particularly in upper classes...the average age of first birth is 33 [years]. So these are women with not a lot of childbearing time left and they already feel squeezed....So faced with a one in four chance of a kid with CF versus never having any kids at all that's also going to affect their decision.
>
> The final thing that's important is there need to have control, people vary in the degree to which they feel the need to be in control over their environment...One of the ways they can maximize or improve their sense of control is to gather as much information as possible. Most people rather than wanting control they're very uncomfortable with the threat of having it taken away. They want to know if the test is available should they want to have it, that doesn't necessarily mean that they're going to have it. But should they be in a position of uncertainty, not knowing what's going to happen to them, feeling they've lost control over their future, then they want to be able to exercise that control. (R12)

The statistical risk is less a factor, according to this respondent than is the individual response to that risk. In other words, it is not just the number that is vital but where that level of risk fits into a person's life overall. Risk analysis must be looked at in a very broad spectrum in order to understand the decision making process. It is folly to assume that being at risk for a child with CF has the same meaning for each individual or couple. In addition, given the way in which people understand or don't understand statistics, it should not

be surprising that numbers may play less of a role in decision making than other less 'rationale' criteria such as age or status of relationship. While it may appear to researchers that individuals do not make rational decisions, to the individual him or her self who is in possession of more personal information, the decision may indeed appear to be very rational.

The understanding of risk and subsequent decision making is a personal experience aided by information from genetic counselors. It is the job of genetic counselors to make these details meaningful and to impart the choices available for the individual. Genetic counseling services are part of health care provision and will be discussed below.

6.2 (4) Carrier screening programs

Pilot studies based in Montreal high schools have generated controversy *because* they are being conducted in high schools with students who are under eighteen, the legal age of majority in Quebec. The rationale for targeting students lies in the fact that the majority of individuals in Quebec will attend high school. Thus, in terms of accessibility:

> ...it's an ideal time...it's a window of opportunity that you just don't have again. (R14)

Few dispute this fact, however they question whether it is ethical to use a captive population for research and the subsequent implications of doing so.

> I think perhaps the only reason it's worked is because it's a captive population. I don't know if it's right that you treat them as a captive population or whether they're capable of making independent decisions at that age and amongst their peer group.(R11).

Another concern that arises is whether students will remember their test results given that for most of them the information has no immediate value.

> ...and then there's the question, well are they going to remember and are they going to remember correctly. (R14).

The ability to remember information that has no immediate value is of concern, yet there is little indication, based mostly on Tay Sachs screening in high schools, that students forget vital information.

> (In my experience) the people who are carriers do remember, the people who are not carriers say, yeah, I was tested but I don't remember if I was a carrier or not and generally we go back and we find out they weren't a carrier. It's because the information wasn't really important for them whereas it would be important if they were a carrier. (R3).

Although many teenagers act like and are treated like adults, they are not yet adults capable of taking on adult decisions and responsibilities. While individuals are able to make health care decisions for themselves at the age of fourteen, they may be unprepared to deal with the intricacies of the uncertainty imparted by CF genetic tests and they may be uninterested in the information as it has a purely reproductive focus.

> You know when you're 15, 16 you're very fragile about [feelings associated with knowing you are different] and there's a lot of concern about that...there is concern because they're not of *legal* age of consent, they're not 18 years old. (R4).

Health care professionals involved in the Montreal pilot studies interviewed consistently equated CF screening with that of Tay Sachs, for example, talking about 20 years of results. However they are in reality, two very different diseases, with different pathologies and vastly different outcomes. This comparison, while easily made, is, essentially false. Thus:

> he's [Dr. X] a very strong advocate of [CF screening] and has been for years and most of the rest of the community disagrees with him on this. I don't have anything against screening high school students per se but when it comes to CF it's not a good idea for a lot of reasons....CF is much different from Tay Sachs and thallasemia in that it's a much less specific test and it's a whole lot more complicated to interpret the results of any specific screening test...(R17)

Again, the critiques return to the issue of the sensitivity of the test and the ability of the population to comprehend the non-specificity of screening at this time. As stated above, this can be perceived as essentially a time bound issue.

In the interim, in reaction to criticisms of the high school based programs, a new pilot project is due to commence in Montreal, based in local clinics with a target of preconceptual women (women who are planning to conceive). This study is expected to test 4 000 women over a three year period, using essentially the same

education, screening, and evaluation protocol as the high school projects.

> What we want to do is work within a family planning clinic, ...very large clinics in which several thousand women would come in on an annual basis and when they come in they automatically meet with someone before they see a physician and a blood sample is taken automatically. ... we would have a counselor or coordinator who would then provide written information about CF who would ask the individual ...simple questions [such as] Have you heard about CF?... If they are then interested, we would give them an information packet and we will also have a video....Then if they were interested, we would ask them to sign a consent form and we would tell them that a portion of the blood that you have already given...will be used to test for CF mutations. (R14).

This family planning clinic based study would avoid many of the criticisms aimed at the high school projects. Nonetheless, it does not resolve the issue of population screening, but merely moves the discussion into another arena. The essential and underlying context does not change. The focus remains on the provision of genetic services with no consideration of moral and philosophical meanings of geneticization. It is likely that genetic testing and screening would have a large impact on parent/child relationships before and after birth. This was explored by Barbara Katz Rothman in *The Tentative Pregnancy* (1988), in which she discovered through her interviews that the act of testing a fetus for a disorder changed the experience of pregnancy for women. Providing genetic services in family planning clinics represents a change in the way that family planning

is generally understood. It hypothesizes that a fetus must meet particular criteria before being accepted into the family.

Data from the interviews indicate that this is a concern for many individuals working in the field of genetics. Respondents discussed the fact that they felt there were concerns related to testing or screening children for carrier status. Most of their concerns were related to any change in the way in which the child was perceived and treated within his or her family environment.

6.2 (5) Screening children for carrier status

Screening children for carrier status of CF is linked to the marketing of genetic services because it allows for the expansion of a consumer base. To date, there have been no pilot studies involving children although some researchers have suggested that newborn screening for CF be added to the protocol of ongoing newborn screening as a means of maximizing coverage and minimizing financial outlay.

Screening children was frequently cited by respondents as a sensitive issue within the research community; an issue that had given rise to professional disagreements between laboratories.

The Montreal research laboratory had no policy against testing children for carrier status and stated that they would test a child at their parent's request:

> No, I don't think we should refuse if parents want their children tested. We assume that the parents will inform these children...when the child becomes an adult and can handle the information....I don't think there should be an age limit....who are we to intervene if the family thinks it's appropriate. So I wouldn't say no if a family felt strongly about having a child tested. (R19).

The course of action is to allow parents to make the decision for the child, even if that decision goes against the judgment of the researcher.

> We'll do it but we would really not like to....I don't know that we'd actually refuse. (R18)

This acquiescence to parental demand has caused much controversy in Ontario where laboratories that conduct CF carrier testing have been debating the issue with an eye toward establishing an age limit for the procedure that would allow all labs to refuse parental requests. There are several components to this concern; the concern that children who are identified as carriers will be stigmatized by society and/or by their parents, the time lag between testing and when the child would be able to make use of the knowledge, and the fact that genetic screening for CF should be undertaken only as a reproductive decision making tool.

One concern is that parents who find that a child is a carrier may treat that child differently from his or her siblings. This differential treatment could be either positive or negative.

> If they had a child who had CF and one was a carrier and three of them weren't you know they might treat the child [the carrier] differently. That's certainly gone through my mind when I've thought about them having their children tested (R4).

There is some fear that the child who is a carrier may suffer from stigmatization from his or her own family members.

> There's no need to know about it [carrier status] for a five year old, our concern is that parents might protect the child more, treat the child differently, focus energies on another child....We don't want to have any influence like that in the family, and it's no benefit to the child....I would never think of doing screening in childhood. (R5).

Carrier status for CF has no impact on an individual until such time as they decide to have children of their own. The importance of carrier status is not to the individual carrier but rather to their potential offspring, as being a carrier has no impact whatsoever on one's physical health. In addition, deciding to be tested for a particular genetic disorder is a decision that must be taken by the individual and like all health related decisions, it is predicated on the assumption of informed consent. This is not a decision that a parent can or should make for a child, given that the affect will occur only in adulthood.

> Then it becomes a whole story if you were tested for something when you were a kid then your parents have to say, well, at what age do we tell her and what if they don't think you're ready to have [sexual]relations and they decide you shouldn't know till you're twenty one or something. I think the person themselves should decide for themselves about testing. (R4).

The individual may not want to know their genetic status when they become adults.

> What happens if you grow up and by that time you decide that there's a disadvantage to knowing...and you've decided I don't want to know this information...well if your parents have already made the decision so that you know you're a carrier, it's too late. (R17).

The consensus apart from one Montreal lab is that CF carrier screening is purely a reproductive tool. Only if and when and individual begins a family are they in need of this information.

> The information is important when you are going to have children. It's important to have the information somewhat before so that you can use it effectively. (R11).

Nevertheless, it is difficult to know how much in advance of reproductive decision making the information must be given.

> I don't encourage people to have their kids tested unless they're at an age where they're going to have children themselves. (R4).

And if the individual choses not to have children then they need never be tested at all.

> ... it's entirely a reproductive issue so there no benefit to knowing on behalf of your child...there's no benefit to knowing you're a carrier except from the point of view of reproduction, so I think that when you're old enough to deal with reproduction, that's a better time to be doing carrier testing. (R17).

The willingness of the Montreal lab to test children for carrier status at the instigation of their parents is indicative of the primacy placed on genetic services in that particular milieu. Several respondents felt that a parents' need to know their child's carrier status was indicative of a lack of knowledge of what genetics can and cannot do for us. There are many things our genes cannot tell us. The refusal to test children firmly focuses attention on the fact that CF carrier screening is a reproductive tool. By providing an open market place in which children could be tested despite any tangible

benefits, researchers risk trivializing the seriousness of the requested information and it's impact on the individual. In addition, it risks drastically altering the relationship between parent and child.

> This is just the tip of the iceberg, we can predict with some ability whether the child will have CF...but how many genetic diseases are we going to look for?...I think that all of this somehow goes toward this kind of process that we're trying to create perfect kids, that I don't see as a goal to strive for.(R18)

If we do strive for perfection in our children, the emphasis on our children shifts from the joys of being parents to a type of pride of ownership in a perfect model of a human.

> What's precious about being a parent and what's likely to be lost with the genetic marketplace, is the concept that you do the best you can with what you're given. The commitment that you have to the child, and the love that you have for the child is an unconditional love. It's not a love that's predicated on the child's qualities or essence but on the child's existence and your relationship to him....Given social trends ...what I'm afraid is going to win out...is the quality control mentality. Such that ultimately the children that are born and are there and really have claims on your love, your respect, your attention will be shoved aside as mistakes of the genetic celluli.(R1)

The quality control mentality threatens to move society further in the direction of a production model of childbearing where the child is the product. This ties in with the idea of increased individualization and increased individual responsibility for the maintenance of the physical body; in essence, this responsibility for one's own body extends to a responsibility for any new bodies (children) created.

The quality of the child becomes a factor in the family relationship. This is an important aspect of genetic services that has not been fully explored by social scientists and which is unfortunately outside the framework of this thesis. Nevertheless it is a topic which warrants more study and more research in order to provide a full picture of how genetics may impact upon the family relationships; parent/child, parent/parent, sibling/sibling relationships will all be implicated within this subject. The respondent quoted above highlights the important fact that parents are supposed to want a child, not a specific child but nevertheless many parents feel that they are ill-equipped to deal with a prospective child who may need care outside the boundaries of 'normal', howsoever they define that for themselves. These are profoundly moral issues about what it means to be a parent and what it means to be a part of a family. In our pluralistic society, choosing a moral agenda is a difficult path.

6.2 (6) Choosing a moral agenda

Technological advances and medical innovation have provided our society with the ability to conduct population based carrier screening programs for Cystic Fibrosis. Nevertheless, no such program is yet in place. The main reason for this discrepancy is not so much linked to what technology can't do (i.e. achieve 100% sensitivity) as it is a result of moral and ethical hesitancy.

> It's a complex issue, very multi disciplinary and clearly people in your type of practice have a much better idea of what's needed. In medicine we haven't looked at these tools about how these services are generated or given.(R15)

It is fully recognized that technology must be approached from a holistic point of view. Not only must we contemplate the techniques but the uses of those techniques. In general, respondents felt that scientists did not have the tools to deal adequately with moral and ethical issues. Some spoke of their own ongoing efforts to understand this side of medical innovations, yet others believed that there was a clear separation between science on one hand and ethics on the other.

> Those in the basic sciences don't deal with ethics because they're working away totally removed from reality. Again it's the ivory tower syndrome.(R10)

Understanding the techniques involved was easier than coming to grips with the social meaning of those techniques.

> It's too easy to provide information, whether you are a carrier or you are not a carrier....It's much easier to do that work than it is to educate people [about what it means].(R18)

One respondent spoke at length about what they felt was the inconvenience of having to constantly deal with ethical issues in their research when what they really wanted to do was continue genetic testing. The symbiotic relationship between technique and the uses of those techniques was not a concern.

> We spend a lot of time dealing with ethical and social issues and so on when all we really want to do [is] test more families. I prefer to spend my time testing more families than working at the specific conditions of the consent forms, reading lawyers letters, taking care of the bureaucracy and administration that's involved with medical tests. I don't want to deal with that, I don't want

> to do anything amoral, I just want to be left alone to do what I'm supposed to do and we have to spend--not just spend but waste--waste time doing these things. There's a disproportionate amount of time and money that's devoted to these issues. (R19)

What this respondent missed was that it is not possible to continue testing without acknowledging that the tests themselves generate moral, ethical and social issues.

A common theme in social analysis of medical innovation and technology is the way in which the fast pace of science outruns the ability of society to make moral and ethical decisions and to change attitudes if need be.

> Technology can happen quicker than the change in society. It's much easier to have these new developments in technology than it is to change attitudes. Clearly technology can outpace all of that much faster. So I think it's good that we have these checks and balances all the time. The technology is moving very rapidly and you can have a test today and within a very few years that test can be automated and the cost can be cut down.(R18)

Choosing a moral agenda then becomes a question of catching up to the technological aspects and the techniques that are already available and in place. The very existence of the technology exerts a pressure for its' own use. A moral agenda is not paramount, it exists only in relation to the scientific agenda.

> People to a degree are either sensitized or conditioned by society to recognize some things as moral issues and not others. And so it's a question of whether something is going to be identified as a significant moral issue As far as that goes I trust society less to chose a moral agenda than I trust it to chose a scientific agenda. (R1)

Respondents point out the fact that techniques evolve much faster than do attitudes toward those techniques. Morals and ethics must of necessity play catch up to science. A moral agenda can only be established one the technique is invented. We cannot have a moral position on a theoretical technology. Thus, rather than morals setting the agenda, they are responding to a scientific or technological agenda. This results in what functionalism terms a "culture lag". Shown clearly by the respondents, a culture lag refers to the time lag when nonmaterial culture, (morals, values and norms) is in the process of responding to material culture (techniques). Applied specifically to genetic screening, the culture lag reflects the fact that we can screen yet we are still examining the context of such screening. What effect will screening have on people with CF, on their families, on reproductive decision making, on our view of health, on the individual body and the social body? These questions of nonmaterial culture must be resolved apart from material culture.

Social change involves a transformation of values stemming from a transformation of techniques, abilities and innovation. Techniques for genetic screening were not invented because society had decided they reflected our moral agenda, rather these techniques were invented as a reflection of scientific enterprise and now our moral agenda must wrestle with the consequences. Respondents had conflicting viewpoints on this matter. Several spoke of the need for scientists to reflect on the ethical implications of their research, the need for them to come down from their ivory tower and yet one respondent lamented the fact that they had to

deal with ethical issues when they would rather concentrate on doing tests. In this point of view, material and nonmaterial culture are not necessarily the business of everyone, but rather can be seen as quite distinct from one another. Following this belief, it is possible to develop technologies without concern for the uses of those technologies. This was a minority response but it does reflect an important opinion. It also reflects the increased specialization in our society wherein some will concern themselves with developing technologies and others with developing an ethical response to those technologies. This is revealing somewhat of a strange dichotomization of abilities and morals and does not reflect the careful and measured responses of most respondents to ethical concerns. Given the relative newness of genetic services, it is natural that society must now grapple with changes in society and develop moral and ethical guidelines that may be time consuming, (but not time wasting!). Nevertheless, it is in dealing with ethical issues and with the transformation of social values and of social relationships that social change will lead to a new social equilibrium. Values must, and will, change to reflect the new technological capabilities of society.

Conclusion

The category of the social meaning of genetic testing and screening is comprised of many different and, at times, disparate, sub-issues. Regardless of their initial seeming disparity, each of these issues is linked to the other in ways that may not be immediately apparent but are irrevocable.

To fully understand what genetic services means for society and what meaning we, as a society, will give to genetics, it is of utmost importance to firstly understand the implications of the power of discourse. One of the most influential factors is how CF is understood by the general population. This understanding will necessarily be influenced by the information given and the kinds of information given will in turn be determined by those who have the power of discourse. How is CF to be framed and understood by society at large? Is it a horrible illness that must be avoided or is it a condition that responds to treatment? While understanding that all parents want a healthy child, to what extent are we as a society willing to go to make guarantees?

It is no small matter that parents with a child with CF are least likely to abort another under similar circumstances. Lack of exposure to CF impacts upon a person's understanding of risks associated with carrier status and may influence a person's willingness to abort a fetus with CF. Additionally, experience with CF and knowledge of CF is vital to the establishment of a carrier screening program. A lack of knowledge of basic genetics and reproduction, in conjunction with the level of false negative tests results from CF testing, indicate possible future problems in terms of how individuals in society respond to genetic research and how they interpret the ability of science to discern truths from the individual's genetic structure. Carrier screening of children for CF is a further intrusion into the physical and social body that creates a deeper moral dilemma. The social meaning of testing must incorporate an understanding of both science and technology and moral and ethical

decision making. It must not simply be relegated to discussions as to how best to provide genetic services to a population.

6.3 Health care services

Issues related to the provision of health care services include epidemiological studies and their association with genotype/phenotype links; the provision of genetic counseling services and concerns about the costs of genetic screening.

6.3 (1) Counseling services

But many genetic counselors feel that written information is not adequate to deal with the intricacies of CF carrier screening.

> The Caucasian population in North America is just huge, there's no where near enough people to counsel and you need to counsel because people need to understand the implications of the test. There's not nearly enough counselors or the money to do it ...Here in Canada it's very difficult, we have Medicare, we all pay for the medical system. If this [carrier screening] were to become standard practice we would all have to pay more into the system. (R4).

Lack of financial resources does not just translate in a lack of genetic counselors, nor are financial issues the only reason for the relative small numbers of counselors practicing today. The program in genetic counseling at McGill was established in 1985 and a second one was started in 1993 at UBC. The McGill program graduates six or seven counselors a year from its two year program while the program at UBC had an initial goal of only four students.

> If you wanted to do major population screening, there wouldn't be enough people. ...There are not enough genetic counselors to go around to do something like that [population screening]. (R5).

Counseling can be a relatively time consuming process, particularly with a disease like CF where the test for carrier status is not 100%, thus patients have to be made aware that a negative test result does not completely rule out carrier status.

> An average first time session would be 45 minutes to one hour...[We see] the average person [not] more than two times. ...probably 80% to 90% of people we see only once....there's two of us who work full time and a genetic counselor who works one day a week and from us together we saw 521 patients in the last year...We could handle a little bit more but at a certain point it'd get crazy and then we would ask for another genetic counselor. (R5).

An alternative would be to put some of the burden of genetic counseling onto other health practitioners, however as the literature review indicates, physicians have been reluctant to take on the responsibility.

> We need to educate the rest of the medical community about what the story really is and who's at risk and what the considerations are in the context of that...The only practical way of dealing with that ... is through the family physicians, it's going to have to be part of their responsibility and their team's responsibility.(R17)

When asked if they thought GPs were qualified to do this job the respondent gave an unqualified No.

One solution could be to train individuals in counseling solely for CF.

> What they've done in the States in different areas is that they've done specific training for a particular disease, where they train somebody, they teach them all about that one disease. [But] to be accredited...you can't be a single gene counselor...But it's an option for screening to train people like that in one area so they know that one specific disease with some genetics background. (R5).

Nevertheless others seem to feel that the very nature of carrier screening for CF demands a much more rigorously professional approach.

> People have to present themselves to a genetics professional because the information they're going to get is not simple and that's part of the dilemma of CF, you can't be certain of a person's carrier status to a level of 100%. There's a certain percentage risk so from that perspective anyone who needs this type of information should really be counseled first....But it has to be someone who can at least counsel the person properly so CLSCs are not appropriate. (R7)

It could be possible to replace one to one counseling with written information or a video screening as has been done in some pilot programs.

> I don't think that's socially responsible because you're not giving people information. ...It isn't that easy with CF so I think it would be responsible to not five people counseling. Admittedly what is done in the schools is they give a group talk and people come away with a fair bit of information...people seem to get a lot out of it and to understand it. (R4).

To this genetic counselor, group counseling may be an adequate solution to the problem lack of resources and time. Unlike reading a pamphlet, a counselor is available to present the basic facts and to answer questions that arise. However, this scenario of extending the services of group counseling has not been studied. It would be interesting to discover which method succeeds in conveying information most adequately. Granted, when written materials are distributed, there is invariably a contact person from whom

additional information can be solicited. Yet, perhaps studies could demonstrate which method is preferable to the general population and to health care professionals themselves. The problem unique to CF is that the sensitivity of the tests is not infallible and there would be a high level of false negative tests as well as many cases where the tests does not reduce one's risk but raises it.

But others feel that the issue of sensitivity is of concern more to physicians than to individuals.

> Specific problems with regards to screening for CF has to do with...sensitivity and specificity of that test. In my mind that's pretty much a time bound issue, in other words, today we get X%, at some point we'll probably get the sensitivity which will probably make it acceptable to whoever feels if a screening test would be done it should have a particular sensitivity. I'm not convinced that that's a major issue for the screenees or for those who will or will not be interested in screening. I think that's much more of a major issue for the physicians and geneticists. (R19).

Because all of the CF mutations have not yet been located, carrier screening would not offer a level of reassurance that individuals would expect to come from the medical community. Rather there are too many unknowns to offer. A person may test negative for one or more mutations, but that would not rule out for absolute certain carrier status for them. What effect this would invariably have on a person trying to make reproductive decisions is hard to discern. For this reason, it has been suggested, and, for the most part, accepted by the medical community that carrier tests should be delayed until a 96% detection rate has been realized. Once

this rate has been achieved the risk for a couple where one partner is negative and one is positive will be 1 in 2 000. This risk is approximately the average risk in the general population. The risk of a couple where both partners tested negative would be appreciably lowered. Providing adequate genetic counseling to all participants of a CF genetic screening program would be financially prohibitive. It would fall to cost-benefit analysis to determine if those costs would be outweighed by savings garnered. However, as discussed below, some professionals question the ability of cost-benefit analyses to factor in intangible costs to the individual.

6.3 (2) Research into CF

One of the most compelling reasons given for ongoing carrier screening for CF is that of information gathering. Given that CF is a disease with very different levels of severity and with many different mutations more needs to be known about the distribution of the gene and about links between genotype and phenotype.

> ...the third rationale [for carrier screening] which is perfectly valid is for enumeration and genetic epidemiology... we know that the distribution of genes in CF is not random in this province [Quebec]....That's very valuable information. (R16).

> We try to be as impartial as possible. It's the same for any disease. ...You can give the horrible [scenario]...or you can give a very good rosier picture and for parents who are faced with the decision of terminating or not terminating that's going to play an enormous role in their decision and you don't want to do that...we have no way of predicting. Which is another reason to see the genotype/phenotype relationship because perhaps we

> [will] have a way of predicting. With some mutations of the CFTR gene which don't have any lung phenotype at all, they're associated with absence of the vas deferens, these people are infertile and that's the only way they get picked up, that's a very mild picture. (R3).

Although it is generally recognized that it is virtually impossible to completely eliminate the CF gene in the population, one respondent wondered about the hopes for cure given the direction screening is taking:

> There are people who will make the argument about if you keep doing this [aborting CF fetuses], you're just going to eliminate the gene pool before you can cure it. That argument has a role in a case like CF. These are not all people who are going to die in childhood [like] Tay Sachs...(R11).

One hope is that when firm links can be made between different mutations and different expressions of the disease, treatments can be better tailored to individual need and researchers will be one step closer to curing the disease altogether. First strides have been taken in this direction. How this proceeds depends upon the specific strategy of testing that is foreseen. Presently testing is limited to individuals with a family history of CF and in the case of Sagenay/Lac St. Jean area a regional disparity is also found to exist owing to that regions particular historical circumstances. Phenotype/genotype matching has shown that ◊F508 is associated with severe disease as is the W1282X mutation common in Ashkenazi Jews. In addition, the CF mutations found in Native Americans is linked to a severe phenotype often with microcephaly. The step by step linkage of phenotype to genotype is an important part of research. In addition there are uses for this knowledge other

than research for a cure. Given that one of some of the opposition to testing and aborting fetuses for CF is that some of those fetuses will be reasonably healthy, phenotype/genotype linkage could result in only the very severely affected fetuses being terminated. If this were possible, the comparisons of CF to Tay-Sachs would be more true than they are at the present time. Tay-Sachs is an always fatal disease of progressive deterioration that begins even before the affected child is born. Cystic Fibrosis is not nearly as severe, nor is it considered to be as lethal as Tay-Sachs yet the comparisons are continually made in discussions of screening programs. In my interviews, it was accepted as a given that since Tay-Sachs screening programs had such success that it would naturally follow that CF programs would also have wide success. But in terms of outcome, it would make more sense to compare CF with T21 (Down's Syndrome).

Although T21 is a mental disorder and CF a physical disorder, there are striking similarities in how the two are treated. T21 also has an extremely variable penetrance. Some individuals will be severely handicapped both intellectually and physically, some will only have a mild learning disorder and will be able to learn to care for themselves, some babies born will only survive a few months and some will live well into adulthood. But there is no way of knowing in advance how severely a particular fetus will be affected.

Amniocentesis for women over the age of 35 has become the normal standard of care and the termination rate for fetuses found to carry the third chromosome is consistently placed upwards of 80%, some studies have placed it as high as 98%. Nevertheless, there is no program in place to eliminate T21 from the population. In fact,

although older women have a higher risk of having a child with T21, birth patterns result in more children with T21 being born to younger women who are not routinely offered amniocentesis unless there is a family history of T21.

Research geared toward therapies and treatments for CF are the hope of individuals with CF and there families. Lung transplantation represents one of the more extreme treatments available.

6.3 (4) Lung transplantation

Despite the positive manner and the money that the CCFF puts into lung transplantation services, there are some reservations surrounding this procedure

> ...[the] issue of lung transplants ...is very controversial. I know two kids who have had a transplant...One was about 21 and one about 18....The debate is, I don't know what the statistics are but for some it works well, for some they die. The debate is, it's not a cure, it's a continuation. The GI problems don't change and they still can have liver problems, they wouldn't have to go through the postural drainage but there are still allot of problems and a lot of complications like any transplant, rejection....It's a big decision, ...it's not like you get a transplant and out the door you go....a lot of people die on the table or shortly after or within a short period of time. It makes the debate harder, where do you stop treatment, where do you continue on...How long do you wait? (R7).

Being eligible for a lung transplant requires a person to be sick but not too sick. It is a cure that may prove to some recipients to be worse that the disease they have been living with. At this point, lung transplantation must be looked at as a measure of last resort.

6.3 (5) Cost concerns

Some professional believe that genetic testing and/or prenatal diagnosis should only be made available to couples who are planning to abort if the fetus is found to have some disorder.

> What comes out is the cost part...Would you permit someone to have PND who clearly tells you I'm not going to act on that information I just want to know if my child has whatever disorder. I have difficulties with that because information is not free. To put aside the funds and resources to do one prenatal, it's quite expensive in the lab....information has a price. (R10).

The assumption that this doctor has made is that only by choosing abortion is the couple making an *active* decision; the only way to *act* on the information is to abort. This clearly does not speak to the parents who consider preparing for the birth of a CF child to be *acting* on the information they receive. Research has shown that many individuals choose carrier screening and prenatal diagnosis to avoid the birth of a child with CF but many others choose to prepare for the birth of a child with CF. In addition, counseling is intended to be strictly non directive with its' sole purpose to provide information that the individual (or couple) can choose to use in the manner which best suits their needs. Only allowing people who want to abort to have genetic tests done is a very dangerous and potentially harmful direction to take for genetic screening. It presupposes that genetic screening exists to avoid the birth of individuals with genetic diseases. The logical extension of this direction would be that anyone who opposes abortion would then oppose genetic screening.

Politically and socially that would be an incredible lobby against monies being spent for genetic services and genetic research.

In other areas, nevertheless, cost is a topic for discussion. In terms of lab facilities, as opposed to those of counseling facilities, it may simply be a case of more efficiency being able to pick up the increased needs.

> ..it depends first of all on how much uptake there really is. ...If there were universal uptake...there would have to be much more in the way of lab resources but I think anybody doing it now could do tremendously more samples than what they do. Because a lot of time it doesn't take any longer to do a hundred than it takes to do ten.(R17)

The largest laboratory in Canada was, at the time of the interview, only processing about 120 samples in a year. The cost per test would be expected to come down if bulk samples were processed.

> ...it would be much better [financially] if we were doing a lot [of tests] at the same time....there could be tremendous more efficiency and there's lots of opportunities for automation.(R17)

However, another respondent felt that there was a serious shortage of resources.

> We should think about expanding screening into the general population although I don't think we have the resources to do it, we don't have anywhere near all the resources to do it, we don't even have the resources to do all the family members of CF children. (R19)

Costs on a purely financial side are not necessarily what's most important as a consideration.

> It's not a cost issue from society's point of view really. I don't really look at that although I acknowledge that dollars and cents are incredibly important right now but they are important to the ministry of health who is paying for all of this, this is the only way that they can relate to the possibility of setting up wide scale screening so far as I can tell. (R17)

Other costs, such as the emotional costs to families are difficult to measure.

> If there were an overwhelming compassionate issue in this situation like if we were going to go out and do all these tests that cost a lot of money cause we may find a child that we could prevent from getting a disease by treating them or something, maybe that would be a little bit different. (R17)

Nevertheless, costs have to be calculated.

> There have to be cost analyses whether you want to relate them to the benefit, there's so many intangibles that it's very difficult to assess the other side of the coin but we do need to know how much the tests are going to cost. It may be that it's utterly ridiculous, some studies show that it's going to be utterly ridiculous...it's going to cost so much that if you only think about it for a minute you'll say it only makes so much sense to target the high risk population, people where just one member of the couple or someone in the family somewhere has CF.(R17)

It is difficult to know what costs are acceptable and what costs we are willing to take on as a society.

> Cost containment is a problem but the reality is there's only so much money. ... You have to ask, "how are you going to value someone's life" ...will socialized medicine take care of that? ... The reality is we make those decisions all the time....There's always decisions being made [in medicine about]what's practical and what's

> impractical. ...Put it in the States where they don't have Medicare, the CF children there who don't have insurance don't get care. That's the decision.
>
> The reality is [costs] are driving the system and I would rather bring it out into the open, address it fully and rationally rather than forget about it.(R15)

As a society, we should be able to determine where money is allocated and to what ends.

> Given the limited number of financial resources that are offered and what Canadian society will choose to do with its' money and where it will choose to put its' emphasis ...it's a question of allocating resources....One scenario is that instead of targeting just the population at large you target extended kindred of CF families because you know you might get more carriers that way and provide them with more information. (R18)

For this respondent then, the status quo makes more sense than population based carrier screening. Given the pressures on the health care system, resource allocation must proceed from a guarded consensus on what society will choose to do with it's money. Proponents of screening will highlight the savings possible with a screening program. One respondent separated her *professional* opinion from her *personal* opinion with regards to the severity of CF in the population.

> I feel uncomfortable about this and I think it's a very difficult thing when you ask a person what are your personal feelings as opposed to what are your professional feelings. It's difficult to know at that level. As director of research or as director of a diagnostic lab I really don't think it's necessarily appropriate to express personal opinions. ..But actually, you have to say is it [CF] that common, how many people do you know? When you were growing up, did you know anybody with CF? (R18)

This respondent emphasizes an important issue in that society may not deem as vitally important an genetic disorder that is carried by only 4 or 5 percent of the population and is expressed by fewer than 3.000 individuals country-wide. Indeed, when placed into that context, the prevalence of CF seems low to the respondent above.

6.3 (6) Genetic services for women

One issue that has engendered controversy is the extent to which genetic services (specifically testing and/or screening) are designed to be used predominantly by women. This may act to increase the burden and responsibility placed on women as gatekeepers for the health of their families.

> There is a gap between the theory that women are exploited [by geneticization] and the way they [women] seem to want these services. (R16)

The gap to which this respondent refers is an indication of the bind that women are placed in when health services are available and they must choose whether to use them. As has been previously discussed, women may feel pressured to use services even though the services may act to exploit them as women. Genetic services do not create this exploitative relationship, nevertheless, they do play upon a previously existing inequity in health care.

> Women *have* the burden of genetic health already. ...no one's going to say, "hey, we'll pay twice as much and we'll do them both at the same time". So who comes in for any kind of care, I mean, how often do men go to have their genitals checked? But women who look after their health see a gynecologist once a year, more or less, or once every 2 or 3 years. So these women are coming into a family planning clinic, they're coming in anyway, their

> blood is being drawn anyway so what agency is going to say OK we'll spend twice as much and we'll do them and their husbands. When we're going to make it a service and it's going to be a cost/benefit analysis and it's going to be how much does it cost to treat a CF patient versus how much does it cost to do testing. ...in the main, the initial concerns in terms of health etc. is usually carried by the women, it's a biological fact. It's a fact of life. If men would get pregnant and carry then maybe they'll do it but I think it's ... She's the one who's coming in to the physician, she's the one who's going to be the first hook-up. ...It is true that women will have the burden but we're not creating that burden. ...by having the woman walk through, have the test and walk out, it's more cost effective. Yeah, there might be an inequity in terms of who's carrying the burden but the other side of the coin is in terms of what you can deliver in terms of health care at the present time this is the most effective way of doing it. I think one could make an argument that women carry the genetic burden but I think they always have and I don't think that's going to change. Not just the burden of genetics, but the burden of child health care in general. ...if you start with the goal of providing the best kind of health care in a given situation, you immediately have all these limitations and you have to work with them. You have to work with them, you have no choice. (R14).

This response hearkens back to Foucault and his concept of the docile body. As Foucault states, there is no natural body but rather all bodies are inscribed by cultural practices and constituted by culture. This is made explicit in the use of women's bodies as an entry point for the provision of genetic services to a population. The female body is constituted as available for health care services. But how did the female body come to be so defined? The perception of the female body as available to science and medicine has a history(see Hubbard, 1990 and Russett, 1989 for a historical analysis of this

concept). The male body lacks this definition of availability to medical science, unless they are being paid to be a part of a research project. Whereas women are expected to show up for gynecological checks or obstetrical visits, there is no equivalent check point for men. While the logic of cost effectiveness appears valid on first blush, there is no sense of accountability in terms of the other social costs and consequences that arise in the definition of the role of women.

The respondents accept the status quo for women. A continuation of the status quo in this instance has as a result the continued perception of women as *available* to the medicine. Because women are made to be available to health care providers, those same health care providers have no qualms about requiring women to be *more* available and *more* responsible, not just for their own health and their children's health but also in acting as a screen for their male partner's genetic status. Many men will only learn their own carrier status as a function of their female's partner testing positive.

The perpetuation of women as responsible for the health care of men and children indicates one way in which women's agency is coopted by and for others. This is a way that the female body is culturally constituted apart from the male body. Although Foucault does not depict a gendered theory, it is possible to read his theories of the body as implicitly gendered. Clearly the female body is subject to surveillance in ways that the male body is not.

The gaze within which the female body is subsumed acts to create of the individual body a social body: the female person finds

her body acting as a gatekeeper to the genetic status of her children and her male partner. In this way, the woman learns to practice self-surveillance. This is key to understanding the gap one respondent referred to as existing between theories of exploitation and the willingness of women to embrace genetic services. Foucault points out that self-surveillance does away with the need for policing by others. This is especially true of women who have been subjected to the normalizing gaze to such an extent that they have internalized it as a core element of their own being. This can be seen in such mundane details as the fact that (heterosexual) women claim they dress to impress other women (and not men) as well as in more complex issues such as women using genetic services as a self-policing technique related to reproducing children. This element of self-surveillance is crucial to keep in mind when exploring the issue of supply push or demand pull for genetic screening programs.

6.3 (7) Supply push or demand pull

A crucial area of dissension is whether carrier screening programs are an outcome of supply push or demand pull. In other words, is there a demand for carrier screening by society or is the need for screening pushed or created by researchers with a vested interest in such programs? In addition, the basic question hinges on whether carrier screening is health care or not.

> I came away from the CF workshops that I was involved in last fall, thinking that people who are actively involved in investigating this are thinking that this [carrier screening] is not a great idea.(R17)

Individuals with a vested interest in maintaining and enlarging genetic services adamantly propound the existence of a "consumer"

demand for these services. However, it becomes clear that the demand is more an indication of some medical professionals desires than it is an indication of the desires of the community.

> The data are there that citizens don't come on their own initiative (R16)

Having said that, this same respondent slammed policy decisions that would narrow genetic services to those individuals who actively sought them.

> So , we're dealing with politics, we're not dealing with reality. We're not dealing with what citizens want, we're dealing with what some individuals wish to exert as power. (R16)

Nevertheless, it was commonly reported in my interviews that the above respondent was instrumental in the establishment of the Quebec Network of Genetic Medicine and had a vested interest in screening programs in the province of Quebec. The respondent went on to suggest that genetic services were being cut because of ethnic considerations.

> But our society has said it's OK to do something for Jews, it's OK to do something for Italians and Greeks, ...and the reason why all the fuss has started with CF is it's oh my god, it's not them, it's us that we're talking about now, the genes in us. We don't like that. ...When you put that out on the table people have trouble dealing with that hypothesis. And then they realize oh, my god [the respondent] is going to be looking at my genes instead of some minority groups' genes. So we better take care of [respondent]. And his colleagues. (R16)

The personal attack felt by this one respondent was not legitimized by any other of the respondents, not even those who work closely

with the respondent. There was no evidence given for this feeling other than a personal belief. Despite the respondents bitter vitriolic, history has demonstrated that screening programs that do not have a solid generation in the community do not fare as well as those that do.

> Many of the other genes for which there is community screening are ones in which there is an identifiable community so if the community stands behind it--there is support from the community--people in the community understand what it means to be a carrier and it's accepted--then I think programs can take place fairly effectively but when it's something like CF where it's common through almost all of the Caucasian population there's no support group, there's no structure and from that point of view I think...in terms of there being any kind of community based screening thing it's going to be rather difficult.(R18)

Creating this base, and establishing a niche for continued medical services, amounts to the marketing of genetic services to the public.

> Reality is medicine right now is driven by individual interest groups. Should that be the way it should be driven? Of course not. It should be driven on a rational basis because you cannot allow one small interest groups to act on its' own isolated from the rest of society. And it's unfortunate that those who squeak loudest get what they want but that's the way our system is driven.(R15)

Part of the reason for the supply push is that medicine creates for itself a niche. With a healthier population, medicine moves into preventive health care and now moves toward lifestyle management.

> Medicine is turning into preventative medicine [and] lifestyle management and I think that genetics has always thought of itself as a lifestyle [component] in terms of reproduction, what plans you're going to make, what your future is.(R10)

Lifestyle needs are increasingly defined by medicine and by technology.

> Are those needs going to stay the same? We can't tell because the technology is going to change and many needs are technology driven. But at this current point as best we can what are the needs? And what are the patient's needs? What do they really want? ...People have to become educated consumers about their own health because the reality is bottom line, no one else is going to care about your health as much as you.(R10)

If the individual does not construct their own needs, they will be constructed for them by doctors, health care professionals and politicians. Foucault stated quite clearly that the doctor would become the judge and this is made visible in our society which has increasingly medicalized life and placed more spheres of life into the hands of medical professionals.

> Policy is driven now from the ivory tower and no one ever goes down to the people cause they say, "What do the people know?" ...Politicians don't know what's happening but they control research funds and they are learning from their advisors where the money should go.(R15)

Some people simply believe that individuals are unable to make these important decisions for themselves. The power to shape discourse and in doing so to shape the very social reality of the

populace falls not to the individual but to those acting in the interests of 'other'.

> There's clearly a group of people out there who think that the patient doesn't know and so we have to tell the patient. ...In medicine we have to deal with the individual so we go back to decisions like that are based on the individual and it's also based on an informed basis. In medicine we tend no to inform people because we assume they won't understand and we know best for them. This is sort of the health professional attitude. That's slowly changing.(R15)...

Even if there were a consumer demand for genetic services, as a society we may not be under a moral and ethical obligation to provide these services.

> To the extent that we consider it on a doctor/patient medical model, then it's absolutely appropriate to consider it on the basis of consumer demand and we have considered genetic services in that way, as services between a geneticist ...and a couple...certainly not on a social level. It's because of considering it in that way, and grounding the thing in consumer demand that it's so simple and obvious that you want to screen for gender if that's what people request. It's exactly the same model. ...If people are going to be caused anguish by having another female in the same proportion that they would by having a child with some disease then that demand which can be satisfied medically through testing and abortion needs to be done. ...[However] a lot of people think that the desire and the consumer demand in question is an illegitimate one. It's illegitimate because it involves a form of choice that cannot be kept within the bounds of the doctor/patient relationship. and to legitimize that desire is necessarily going to have implications beyond that relationship and therefore society is justified or may be justified in saying this is not an area where we would allow consumer preference to hold sway. (R1)

While functionalism does assume that society naturally exists in a state of equilibrium, it does not negate the existence of periods of social change. Social change may be the result of the discovery of a new phenomena or the creation of new phenomena; both are twin processes of innovation. Innovation is the precursor to diffusion; the spread of an innovation from group to group or society to society.

The discovery of the CF gene in 1989 led to the technique of being able to test the general population for carrier status of the gene. This present an interesting case because while this discovery was new for Cystic Fibrosis, it was already a reality for other diseases such as Tay Sachs. In broad terms, the ability to detect carrier status in the population was not new, it was only new for this particular disorder. Social change associated with Tay Sachs had already a twenty year period of adjustment. The diffusion of this 'new' discovery was predominantly into the Caucasian population; Jewish peoples had already spread the knowledge of genetic screening throughout their communities. (Interestingly, one of the respondents believed that part of the rectitude toward CF screening programs hinged on the fact that it dealt with a gene in the dominant, or majority, population. This respondent felt that policy makers were not as anxious to acknowledge defects in their own genetic patterns as they were to recognize them in a minority group. However, this was an extreme viewpoint, not shared by any other respondents nor found in any of the literature.)

Genetic screening has been an accepted practice in the Jewish community and in the Greek and Italian communities affected by Thalassemia and programs are in place to deal with the needs of

those communities. There are few dilemmas presented by these programs.

Cystic Fibrosis does present considerable dilemmas for the community. One of the main sources for these dilemmas is the fact that the disease itself is not perceived to be as bad as Tay Sachs and Thalassemia. Additionally, there is no community support for screening programs. Any attempts at establishing testing or screening has always come from the researchers themselves. The respondent quoted above seemed oblivious to the fact that while he stated there was a demand for genetic services, he also stated that there needed to be an education campaign to show people that they needed genetic services. This points to the fact that the act of diffusion has not extended far into the community. In addition, it signifies that there is more of a supply push for genetic services for CF than there is a demand pull.

Conclusion

An analysis of health care services related to genetics risks begging the question of whether the priority should be in discovering how to provide genetic services or if the priority should be in learning whether such services should be provided at all. In addition, the uncritical acceptance of genetic services may result in an unwillingness to address substantial issues related to changing values and mores especially as concerns human social relationships. Still it is important to examine health care services in terms of the ability to provide necessary professional care and facilities. Furthermore, the need for genetic services must be evaluated from a social science viewpoint.

There was some discrepancy as to whether current counseling and laboratory facilities would be able to respond to an increased workload related to population based genetic screening. It is apparent that while laboratory facilities would be easily upgraded to deal with increased sample processing, counseling services would have to be changed from the present format to meet the need of a larger sample size. This could mean an increase in the number of genetic counselors made available, but would probably actually translate into a substantially different method of information dissemination; this could include giving information through pamphlets and videos and/or training para-professional counselors to impart information concerning CF uniquely and/or enlisting the cooperation of general practitioners or family doctors as quasi-counselors.

Any analysis of health care services must deal with cost concerns and when discussing CF, which is an autosomal recessive gene, the costs incurred by genetic testing or screening are examined in relation to costs saved by not having to pay for the health care of individuals with CF. On a purely economic basis, this analysis may be practical, yet many respondents felt that such analyses disregarded the human costs associated with one or the other choice. When dealing with human decisions, the data suggests that purely budgetary discussions are too cold hearted.

Finally, the manner in which the discourse is framed leads to speculation as to whether the offering of genetic services as part of health care is a result of a demand pull by consumers or rather a result of a supply push by the provisors of such services.

Conclusion

The breadth and depth of information garnered by the data presented herein is such that conclusions must necessarily be broad. The data collected for this research helped to answer many questions and generated many more questions in its' wake. This will be discussed in the general conclusions as implications for future research. This conclusions section serves to sum up the wealth of information presented in this chapter of results and discussion.

One of the most striking features of CF is its' extreme variability and the subsequent implications of this fact. Respondents provided details of the daily life of a person with CF (focusing on children and their parents) and this presentation showed the significance of the individual case and the individual response to illness. Spoken plainly, respondents stated that it is as though there are many different diseases under the heading of Cystic Fibrosis rather than just one disease of CF. This leads to a situation wherein it is possible to socially construct CF in many different ways. These social constructions all bring with them different meanings. Understanding these meanings is vital.

One of the first and clearly the major tasks of understanding Cystic Fibrosis as a case study for the provision of genetic services is to identify precisely what CF is. This means knowing the way in which the CF gene(s) is carried and passed on through reproduction and, perhaps more importantly, knowing how CF is manifested in the body. The question becomes "how is CF lived in the physical body?" but this cannot be answered without the addendum of "how is CF

lived in the social body?". Because humans are social creatures who create their own reality, there can be no physical illness that does not have a social interpretation for that illness.

Data from the CF team working with children with CF helped to clarify the everyday routine demanded of an individual with CF. This serves to clarify some of the social interpretations of CF. Based on this data several conclusions can be made. It is clear that not every person with CF has the same physical symptoms and therefore does have to adhere to the same health care regimen. One individual with CF may be able to experience life the same way a person without CF does but another individual may spend much of their time in hospital and may die young. However, when dealing with CF on a broad spectrum, it is a choice as to which interpretation of living with CF must be paramount. As one respondent stated, the kids must be portrayed as "sick, but cute".

If children with CF are portrayed as being very very ill and destined to die young, then more individuals may want to be tested for carrier status in the hopes of preventing the birth of such a child. However, if children with CF are portrayed as having a disorder that requires some special treatment but that enables them to have a happy life, then perhaps fewer individuals would choose genetic testing. The power of this discourse cannot be understated as it forms the crux of the issue. The very act of choosing to research the CF gene is an act of discourse and an act of power. All else follows from this one act of definition.

Genetic counselors stress that they maintain an belief in non directive counseling while stating emphatically that they refrain

from calling CF a disease. Instead they use terms such as "condition" or "disorder". The choice of words, the act of discourse, by it's very nature overrules the goal of non directive counseling. The words condition or disorder are not neutral terms. They have their own implications in much the way that disease does; implications brought on by the very neutrality they are trying to create. By avoiding the word "disease" for "disorder", counselors create the impression that CF is not so very serious. Genetic counselors help to create the social meaning of CF through their use of non directive discourse.

A missing link in this discourse is the linkage of choices with available technology and a lack of understanding of such ties. In other words, the very fact that counselors are available to help individuals make decision for CF necessitates the previous presence of technologies to test for CF carrier status in the population. It is the technology, and not the nature of the disease (or disorder) itself that allows these decisions to be made. Other, perhaps more seriously debilitating genetic diseases (or disorders) may affect more people or present more of a drain on the emotional resources of parents or on the social health care budget but until and unless we have the technology to locate these genes, no choices or options can be made.

The science of gene location is sometimes presented as purely methodical and logical but chance and serendipity have always played a part in research in both social science and physical science. It is misinformed to surmise that genetic research responds directly to the needs of the population. The needs of the population may be shaped to respond to the abilities of genetic research. Scientific and technological discoveries do not exist on a supply and demand

system. If that were so, we would have a cure for cancer or AIDS. Instead, technologies are created as scientific knowledge provides the ability to do so and the needs of society are created around those technologies with a subsequent change in attitudes and values providing the justification for such needs.

Foucault's theoretical analysis of the discourse of the body and the source of power and knowledge help to move the subject of genetics away from questions of how to provide such services to more complex and abstract questions of genetic testing and screening. Foucault provides a sense of how power and dominance are experienced through the individual and the social body. In this matter discourse provides power through influencing how and what others see and believe. In any analysis of social construction, it is vital to emphasize that the subjects in whom power is vested and who control discourse are not found outside of society but are an integral part of society. Thus power is exerted upon individuals within the social sphere by others who also exist within this sphere. In some instances power is exerted by individuals upon themselves. For Foucault, we can all be judged but we are also all judges and our court relies upon inner disciplinary techniques and self surveillance rather than outside controls.

Choosing genetic screening can be seen as an act of self surveillance and the body is subjected by the individual to the normalizing gaze. Thus there is no need for obvious coercion from outside because discipline is internalized. While Foucault did not focus on a gendered body, in his discussion of the malleable body, it is apparent that particular forms of training are more or less

applicable to either men or women. Women have traditionally been subjects of self surveillance of the body from a medical standpoint. It is not surprising then that genetic services are offered predominantly to women. As one respondent stated women are more likely to present themselves for regular medical check ups than are men. Yet women are not more likely to be ill than men are. Nevertheless the social construction of the female body allows for more inner and outer surveillance of the physical body than that of men. Offering genetic services to women does not create this reality, it merely reflects it in an uncritical way. This surveillance of the body as its' own justification is highlighted by the fact that some respondents felt is was valid to test (and in some instances to screen) children for a gene whose presence would only have ramifications for choices made in their future. At this time, we lack the ability to surmise how this knowledge would influence parent-child relationships and what meaning parents would garner from this information. It may simply be a case of power to frame issues by the parent.

In conclusion, the data indicate the complexity of the subject matter from concrete matters such as how many samples could be processed in a lab to intangible abstract issues such as how power is expressed on and through the physical, social body. It is clear that the provision of genetic services will alter our conception of the body in many ways and will result in a new moral agenda. It is also clear on a more practical note that the data indicate some ambivalence regarding the need for and the demand for population based carrier screening.

Chapter 7

Conclusion

The main purpose of this research was to explore the social aspects of genetic research. Rather than testing a hypothesis, given the uncharted territory, the purpose was to generate questions and attempt to seek some possible answers. The literature concentrated on how to provide genetic services but this research was organized to look beyond these practical matters and to question the social meaning associated with such services.

A qualitative methodological approach was used to reflect the exploratory nature of the research design. Experts in the field were interviewed with open ended interview schedules to allow them to voice their own concerns and opinions. Cystic Fibrosis was chosen as a case study because the CF gene (in many mutations) had recently been located enabling the detection of carrier status in the population.

Data was categorized in three areas: practical aspects of living with CF, social perceptions of CF and the provision of genetic services related to CF. Each of these three categories is linked to the other. Theoretical boundaries were vital in bringing cohesion and understanding to the data. To this end, several different theoretical perspectives helped to form the information.

Diana Dutton's work on emerging medical technologies and their social impact helped to situate technological innovations related to genetics as a fundamental social issue. The acknowledgment of

medical innovation as a social inquiry is an substantial first step to the legitimation of this research. The field of knowledge described as medical sociology delineates just that interdisciplinary understanding. The practice of medicine is as much a social practice as it is a scientific one.

With regards to changes brought about by these medical innovations, theories of social change serve to frame the ways in which society reacts to change. Technology is often the arbiter of attitudinal change. Morals change much slower than technological abilities. There is often resistance to change and resistance to particular technological innovations. Necessity is not always the mother of invention; inventions may be born orphans.

Finally, in terms of theoretical framework, Michel Foucault's theories of the body, of power and knowledge and of disciplinary techniques and self surveillance guide the analysis of what meanings genetic research have for the individual and the social body. Foucault act as counterpoint to ideas of patriarchal dominance and control as outside forces. Instead, power and its' discourse are envisioned as coming from within the sphere upon which power is exerted. This helps to explain power dynamics in the medical profession in ways more sophisticated (and more apt) than a reliance on social construction theory would be able to do.

The initial purpose of the research was to examine the context of genetic screening. However, given the exploratory nature of the project, it is not surprising, nor unexpected, that the collection of data led the researcher in many different directions.

7.1 Portrayal of CF

The fact that CF is a variable illness and the subsequent debate over whether it should be portrayed as a serious illness or a treatable condition was unsurprising. This fact signifies that illness is socially constructed and meanings are created by social actors. Still, it was surprising to discover how much the nature of genetic services hinged on the way in which this one crucial issue was resolved. The way in which CF is portrayed stems directly from and in turn, affects, the social meaning of genetic testing. The way in which CF is described in information packets given to individuals to inform them about carrier testing will have an effect on population based carrier screening programs because most people have no secondary source of information. If people are taught that CF is a disorder that responds well to treatment and that does not compromise a happy and full life, they will be less likely to want carrier testing for themselves. On the other hand, if individuals are taught that CF is a very serious disorder which requires time consuming daily treatments and frequent hospitalization and which often results in death at a young age, there would be a high demand for carrier testing. At this point there is no direct cause and effect relationship between these two variables at this time. Nevertheless, the literature did show that individuals who felt that their chance of being a carrier and/or of having a child with CF was low were less likely to want to be tested. Respondents were very aware of this dilemma regarding how to fairly characterize CF. As one respondent stated when discussing fund raising for CF the children have to be cute but sick but not too sick, "cute but pretty sick". She recognized

the fine line that has to be walked so that the public is aware that CF is a serious disorder but not wanting to dramatize nor to alienate families who object to the image of their child as being on death's door. More information is needed on how individuals understand and assess risks related to carrier status and in how individuals process the information that a prospective child has a one in four or 25% chance of having CF. It seems clear from the interviews that experts feel there is a gap in their own knowledge of how people understand statistical information and what other contexts are brought into the decision making process.

7.2 "Acting" on information

In couples where both individuals know they are carriers of the CF gene, difficult reproductive decisions have to be. This research did not interview such couples therefore this information is second-hand and to some extent must be speculative. Respondents who work closely with such couples report that it is very difficult for a couple to choose to abort a fetus with CF if they have already had one living child with CF. Exposure to and experience with a child with CF appears to act as an inhibitor in terms of willingness to abort for some individuals. It appears that siblings and other relatives (especially aunts and uncles) perceive the physical and emotional effort and extra care that is required to properly care for a child with CF one step removed from the parent/child relationship within which the love the parent feels for the child overrides all else. The literature suggests that siblings and other relatives of children with CF are favourable toward abortion of a fetus with CF. Given that

most individuals have no prior knowledge of CF, it would be expected that there would be a willingness to abort.

This research found that some respondents believed that carrier testing should only be offered to people who would be willing to act on the information; that is, to abort an affected fetus. Any other form of "acting" on the information was not perceived to be cost effective. This is counteracted by others who felt that the provision of knowledge (i.e. that the future child would have CF) is an important and meaningful end in itself and parents were warranted in having the knowledge to prepare for the birth of an baby who will have extra financial and medical needs. This finding reflects the diversity apparent in the literature on this issue. At its' core, this dissension is both financial and philosophical in nature.

Respondents who believe that only couples who would abort a fetus with CF should be tested base their beliefs on the idea that then the acts have been cost efficient: the money spent on testing is counterbalanced by the money saved by not paying health care costs of a person with CF. Subsequently, a couple who is tested and who has a baby with CF has incurred double costs: the cost of the genetic test and the cost of raising a child with CF. From this point of view information in and of itself has a cost and cannot be provided free.

The main difficulty is in demonstrating that this information is vital and cost efficient even if a couple does not abort. It is impossible to put a dollar amount on the value of this information in terms of psychological preparation of the couple but this is something that does occur with other congenital disorders that are detected through amniocentesis and/or ultrasound, for example.

7.3 Calculating costs

If cost efficiency is an overriding concern, then population based carrier screening would definitely not be warranted. The high costs associated with detecting the small rate of "couple carriers" would not mitigate the costs of caring for a person with CF. It makes more epidemiological and economic sense to maintain the status quo where only people with a family history of CF are tested for carrier status. This course of action represents the best use of limited economic and professional resources in the present day health care system in this country.

There are costs beyond the purely economic ones and these must also be addressed. It is difficult to attribute costs to abstract ideas such as the change in morals and attitudes and our experiences of living our bodies. However, it is possible to understand these things. Social change theory shows how there is a cultural lag between technological advances and subsequent attitudinal changes. Our reliance on technology as a cure to social and spiritual ills is an indication of the prominence placed on the science of medicine at the present time. This reliance blinds us to the fact that our genes are not the single telling point of our existence and it emphasizes the extent to which medicine has become an overriding belief system for many members of society. However much of what is presented as an individual problem can also be cast as a social problem. As a social problem quite different solutions would be offered than the individualized approach of being solely responsible for one's physical body and it's real or imagined ills. One of the principle social changes regarding the human body is the change from depicting the body as

a physical manifestation of god to depicting the body as a machine with interchangeable parts that must be managed and maintained by the owner him or herself. Individualization and geneticization depoliticize the body and in doing so depoliticize many social problems by recreating them as individual problems fixed by individual solutions. Instead of asking how society can change to better accommodate individuals with various health statuses and needs, the individual is asked to accommodate him or herself to our discriminatory society by policing their own genetic make up and making reproductive decisions accordingly.

7.4 Ethicists response to technology

Ethical issues were important to many of the respondents as integral components to understanding their own work. The few respondents with a professional interest in the ethical issues generated by genetics are an unintended reflection of the marginalized status of ethicists in genetic research in general. Ethicists do not set the agenda, they must respond to the technological agenda set for them. As such, an ethicist interviewed said that he had little confidence in society's ability to set a moral agenda while he recognized it's ability to choose a technological one. The main problem is that society responds to ethics in much the way it responds to technology. Technology is seen as a sphere of knowledge that belongs to particular experts and the rest of us only respond to what those experts generate. Ethics is viewed in much the same way; that is, as a specialized area of knowledge best left to experts. But ethical and moral decision making must be practiced by everyone in order for it to have any value in society. The abandonment of moral questions to

experts leads to the situation where the only question we can ask is how do we best implement technologies rather than asking do we, as a society, need or want these technologies.

It is easier to deal with practical problems associated with the use of technology than do grapple with abstract problems such as how does genetic testing and screening affect a person's sense of self or the way in which a person lives his or her body. These are complex problems but they are also necessary and interesting and enlightening problems as well. The beginnings of dealing with this type of issue arose with the discussion surrounding carrier testing for children.

7.5 Carrier testing of children

Carrier testing of children for the CF gene was not a part of the initial research proposal but it emerged during the course of the interviews and became a flash point for discordance within the group of respondents. This topic illuminated two disparate beliefs about genetic testing. One belief is that any knowledge is good and individuals are entitled to all knowledge medical technology can give them. The other belief is that knowledge without context and meaning is useless and can be dangerous. This debate also indicated our lack of knowledge regarding the topic of negative self image, self esteem and self perception that may be related to knowing one is a carrier for a particular genetic disease.

Essentially, one side of this debate felt that if parents wanted to know the carrier status of their children then they were entitled to that knowledge. Proponents of this felt that the transmission of such knowledge to the child should be left to the discretion of the

parent. However, apart from satisfying parents' curiosity, little justification of testing children was offered. Any benefits would be better garnered by waiting until the child were an adult. This was the opinion of the other side of the debate.

Respondents opposed to testing children did so on the basis that harm may come to a child who tested positive for carrier status of CF. This potential harm was envisioned as stemming from several sources. Firstly, it was felt by some that the parents themselves may have a negative view of the child who was a carrier and might treat them differently than other children who were not carriers. More worrisome, however, were implications that would arise when the child became an adult. For example, the child him or herself may not want to know his or her carrier status but the decision had already been made on their behalf. Knowledge about carrier status could not be concealed from insurance companies therefore the individual was now forced into a position of disclosure. In addition, knowledge about carrier status may affect the child's self image, self perception and/or self esteem. Finally, it is widely acknowledged that knowing one's carrier status is only beneficial if one is deciding to reproduce. Since children are not in a position to reproduce, they do not need that information. When they are in such a position, or *if* they are ever in such a position, they will choose for themselves whether they want or need such information. Genetic counselors felt that the demand for testing of children is an indication of a lack of understanding on the part of the parents. Their demands for testing for their children showed that they did not recognize that knowing

one's carrier status has no use outside of reproductive decision making.

7.6 Supply and demand for genetic services

The discussion of supply and demand indicates that respondents themselves are of two minds simultaneously. While saying that there is a demand for genetic services, some of the same respondents said that education programs were needed to convince people that they should be tested for carrier status. Low uptake levels in many pilot projects discussed in the literature review show that the demand for carrier testing for CF is actually very low. Respondents who want education programs to increase that demand are offering contradictory information. It appears most likely that they have a service they want to offer and not that they have a service in demand. This is a contradiction found in many of the medical technologies and relates back to the fact that medical innovations do not necessarily respond to the needs of society but once a technology becomes available there arises a technological imperative to use the technologies. The discovery of the CF gene on chromosome 7 was made by scientists working toward a cure for CF and not working toward the eradication of CF from the gene pool. In other words, CF will not disappear because individuals or entire populations can be screened for the existence of the gene. Anyone with an understanding of the science of genetic diversity recognizes that eradication of disease is neither feasible nor desirable. In essence, the ability to screen populations arose out of research aimed at a cure for CF. Now that the technology exists, some individuals feel that there should be widespread use of the technology and therefore

approve of a program to educate individuals as to the merits of such screening.

If the relationship between supply and demand were a fulfilled one, CF would be a curable disease. A cure is what the CCFF and its' members are waiting for. Unfortunately, science and technology are unable to fulfill this demand at the present time. To pretend that there is a huge demand for carrier testing or screening of CF is to ignore the realities of the research. Individuals with a family history of CF want and receive counseling and testing; other individuals do not desire testing nor is it economically feasible or socially and medically beneficial to provide such testing. To put it bluntly, there is no demand for population based carrier screening for cystic fibrosis beyond the research desires of particular individuals with a vested interest in such programs.

The findings of this exploratory study are enlightening and help to bridge research gaps. Naturally, the findings also point the way to new areas that need to be explored. The sociological implications of genetic screening are many and varied. The ability to determine genetic information has already proven to profoundly alter our experiences and our perceptions of the human body and the social body.

Limitations of the research

The main limitation of this research is the small sample size and the fact that the sample of respondents was not randomly selected. Therefore, the views of the respondents cannot be said to reflect the views of the community as a whole. Since the respondents were self selected and since many individuals invited to participate as

respondents declined, there is no way of knowing if the sample is reflective of a particular type of individual. It may be that individuals who were not interviewed would have voiced views very different from those discussed herein. Nevertheless, generalizability of data was not anticipated from the outset. The research project was designed from the beginning as an exploration of a case study. As such, the information provided by the sample of respondents was fruitful.

A second limitation of the research centres around time. When the project was first started, the gene for CF had just been located and research was going at a fever pitch. The research herein was intended to describe a process in motion. However, the lengthy time delay from the beginning to the final writing of the project changed the focus somewhat from what *is* happening to what *was* happening. Still, the added time meant that some hindsight could be used to see that initial expectations were not realized in terms of the rapidity of a change in genetic screening.

A final limitation is the decision to focus on experts and not to interview individuals with CF, families with a child with CF or the general population. Experts obviously are knowledgeable about the concerns of the first two groups mentioned as they work with them, however with regards to the general population, information was speculative based on published studies and anecdotal evidence. This limitation did leave the door open for further studies.

Implications for the future

This research points to a clear need for a large scale study, combining quantitative and qualitative data collection, aimed at uncovering and

assessing the needs of the general population regarding genetic screening. As an exploratory case study, this research has succeeded in describing the social context of genetic screening, examining the direction screening could and will take in the future and illuminating how power and discourse act upon our representations of the body. Yet fundamentally, the changes wrought by genetic research and related technological abilities are not about the provision of health care services but about our image of ourselves as human beings.

Future research is needed in assessing these fundamental human changes and their effects on human (and therefore, social) relationships. It was outside the scope of this research to analyze the effects of increased geneticization on male and female relationships, on the choices of partners, on the concept of self and other as carriers of the CF gene and on the concepts of love and the effects it would have on the choice to reproduce and the meaning of reproduction. These are all questions for another project focusing on the individual experience of living in a time of increased reliance on and belief in genetics. This project focused on experts in the field. It is now vital to go to those outside of this limited sphere.

More research is also needed to analyze the idea of costs incurred by genetic services. This must be a project that looks at human and social costs. It is easier to calculate economic costs but the idea of cost must be approached from viewpoints other than an economic one. How individuals perceive and make choices would be an integral aspect of understanding costs.

While it is important to maintain the multidisciplinary nature of the research, it would be fruitful to break down the various

categories of exploration to look at each in much more microscopic detail before coming back to assemble the whole once again. One truth emerges as evident: we are only just beginning to explore the social meaning of genetic knowledge and the implications of gene technology.

Appendices

Appendix A

List of Respondents by Professional Position and Gender

Number	Professional Position	Gender
Respondent 1	Ethicist	M
Respondent 2	Ethicist	F
Respondent 3	Genetic Counselor	M
Respondent 4	Genetic Counselor	F
Respondent 5	Genetic Counselor	F
Respondent 6	Nurse	F
Respondent 7	Physiotherapist	F
Respondent 8	Social Worker	F
Respondent 9	Physician	M
Respondent 10	Physician	F
Respondents 11-15 are researchers in community and public health.		
Respondent 11		M
Respondent 12		F
Respondent 13		M
Respondent 14		F
Respondent 15		F
Respondents 16-19 are research scientists.		
Respondent 16		M

Respondent 17		F
Respondent 18		F
Respondent 19		F
Respondent 20	Executive Director of the Canadian Cystic Fibrosis Foundation	M

Appendix B

Interview Schedule

Interviews were conducted broadly within the mandate of three categories of information: living with Cystic Fibrosis, the social meaning of testing and screening, and health care issues. This schedule was used as a general guideline. All respondents were not asked all the questions. The interview schedule was not present in the hand of the researcher at the time of interviews; rather the researcher conducted interviews as a form of conversation.

1. Living with Cystic Fibrosis

Could you describe to me the daily routine of an individual with CF?

What kind of diet is needed? What types of activities can be undertaken?

What kinds of medication would a person take?

How much time does therapy take every day?

When would an individual with CF be hospitalized?

How does schooling work for the affected child?

What costs are incurred by the family?

How many individuals do you see in your clinic? How many families?

How do parents react when they learn their child has CF?

Is there a high divorce rate in families?

What types of support groups are available to the individual and their families?

Are the children you see generally well? What happens when there is a death? How does that affect other children and their families?

How would you characterize CF in terms of seriousness relative to other genetic diseases?

2. The social meaning of testing

Should we be thinking about population based carrier screening for CF?

Should we restrict testing to couples with a family history of CF?

Should we allow anyone who wants to to be tested?

Is the choice to be tested solely a reproductive decision?

Should we allow children to be tested(screened) if their parents so desire?

Who should make decisions regarding screening and policy decisions? Doctors, politicians, the consumer.

What are some of the ethical implications for screening?

Should we require individual's to act on the information they receive? What would this consist of?

Is screening a moral issue? Should it be?

3. Health care services

Is population based carrier screening for CF feasible? Do we have the laboratory facilities, including personnel?

How many tests does your lab process in a year? Would it be possible to do more? How many more? How would this affect costs?

Are cost/benefit analyses valid? Who should make them?

What is the goal for CF? Is it treatment, therapy, to eradicate the disease etc.

Is consumer demand a legitimate basis for establishing screening programs?

Describe a typical genetic counseling session.
Describe a typical prenatal counseling session.

Should we have education programs designed to make people want to be screened for CF?

What kinds of educational materials should be developed?
Who should develop these materials?

How do individuals and couples understand risk factors?

Could genetic counselors meet the needs of a screening program?
Could GPs step in to take some of the overload?

How do we reconcile that the needs of the community may not be the same as the needs of the individual or the needs of medicine?

References

Acker, Joan, Barry, Kate and Johanna Esseveld. 1983. Objectivity and Truth. Problems in Doing Feminist Research. *Women's Studies Int. Forum,6* (4), 423-35.

Alcoff, Linda. 1989. Justifying Feminist Science. In Nancy Tuana (Ed.). *Feminism and Science.* Bloomington and Indianapolis: Indiana University Press.

Al-Jader, L. N., Goodchild, M. C., Ryley, H. C. and P. S. Harper. 1990. Attitudes of parents of cystic fibrosis children towards neonatal screening and antenatal diagnosis. *Clin Genet, 38*, 460-465.

Anderson, Christopher. 1992. Widespread CF testing inevitable, Congressional report finds. *Nature, 358*(13 August), 529.

1990. Genome project to tackle mass screening. *Nature, 348* (13) December, 569.

Anderson, Ian. 1991. Simple test screens genetic disease. *New Scientist, 132*(1792), 25.

De Arce, M., O'Brien, S., Hegarty,J. O'Mahoney, S. M., Cashman, S.H. , Martinez A., Delgado, M., and M.X. FitzGerald. 1992. Deletion ◊F508 and clinical expression of cystic fibrosis-related liver disease. *Clin Genet, 42*, 271-272.

Arditti, Rita, Klein, Renate and Shelley Minden. 1984. *Test-Tube Women. What Future for Motherhood?* London: Pandora Press

Arnault, Lynne S. 1988. The Radical Future of a Classic Moral Theory. In Alison M. Jaggar & Susan R. Bordo (Eds.). *Gender/Body/Knowledge. Feminist Reconstructions of Being and Knowing.* New Brunswick, NJ: Rutgers University Press.

Axworthy, David, David J. H. Brock, Martin Bobrow, and Theresa M. Marteau.1996. Psychological impact of population-based carrier testing for cystic fibrosis: 3-year follow-up. *The Lancet, 347*, 1443-46.

Baird, Patricia. 1993a/b. *Proceed With Care.* Volumes 1 and 2.

Baker, Geoff. 1991. Peter Hall cuts school. *The Gazette*(Montreal), 23 October.

Barden, Howard S., Raymond Kessel and Virginia E. Shuett. 1984. The Costs and Benefits of Screening for PKU in Wisconsin.*Social Biology, 3* (spring/summer), 1-17.

Bartky, Sandra. 1988. Foucault, Femininity, and the Modernization of Patriarchal Power. In Irene Diamond and Lee Quinby (Eds.). *Feminism and Foucault: Reflections on Resistance.* . Boston: Northeastern University Press.

Bartholome, William G. 1987. Ethics and the Termination of Pregnancy: The Physician's Perspective. In William B. Weil, Jr. M.D. and Martin Benjamin, Ph.D. (Eds.). *Contempory Issues in Fetal and Neonatal Medicine.* Boston and London: Blackwell Scientifique Publications.

Baruch, Elaine Hoffman, D'Adamo, Amadeo F. & Joni Seager. (Eds.) 1988. *Embryos, Ethics and Women's Rights. Exploring the New Reproductive Technologies.* New York: Harrington Park Press.

Basen, Gwynne, Margrit Eichler and Abby Lippman (Eds.) 1993. *Misconceptions: The Social Construction of Choice and The New Reproductive and Genetic Technologies, Volume 1*, Hull: Voyageur.

Beck, Ellen et al. 1974. Advocacy and compliance in Genetic Screening: Behaviour of Physicians and Clients in a Voluntary Program of Testing for the Tay Sachs Gene. *New Engl J Med, 291*(22), 1166-1170.

Beeson, Diane and Mitchell S. Globus. 1985. Decision Making: Whether or Not to Have Prenatal Diagnosis and Abortion for X-Linked Conditions. *Am J Med Genet, 20,* 107-114.

Bekker, Hilary, Michael Modell, Gill Denniss, Anne Silver, Christopher Mathew, Martin Bobrow, and Theresa Marteau. 1993. Uptake of cystic fibrosis testing in primary care: supply push or demand pull?.*BMJ, 306,* 1584-6.

Bekker, Hilary, Denniss, Gill, Modell, Michael, Bobrow, Martin and Theresa Marteau. 1994. The impact of population based screening for carriers of cystic fibrosis. *J Med Genet, 3* (5), 364-368.

Benson Gold, Rachel. 1984. Ultrasound Imaging During Pregnancy. *Family Planning Perspectives, 16* (5), 240-243.

Bercowitz, Gertrude S. et al. 1990. Delayed Childbearing and the Outcome of Pregnancy. *N Engl J Med, 322*(10), 659-64.

Berger, Peter L. and Thomas Luckmann. 1969. *The Social Construction of Reality.* New York: Anchor Books.

Berhardt, Barbara. 1991. Population Screening for the Cystic Fibrosis Gene. *New Eng J Med, 324*(1), 61-2.

Berman, Ruth. 1989. From Aristotle's Dualism to Materialist Dialectics: Feminist Transformation of Science and Society. In Alison M. Jaggar and Susan R. Bordo (Eds.). *Gender/Body/Knowledge. Feminist Reconstructions of Being and Knowing.* New Brunswick and London: Rutgers University Press.

Bernhardt, Barbara A. 1991. Population Screening for the Cystic Fibrosis Gene. *New Eng J Med, 324*(1), 61-62.

Bernhardt, Barbara A. and Robin Bannerman. 1982. The Influence of Obstetricians on the Utilization of Amniocentesis. *Prenatal Diagnosis, 2,* 115-121.

Biesecker, Leslie, Bowles-Biesecker, Barbara, Collins, Francis, Kaback, Michael and Benjamin Wilfond. 1992. General Population Screening for Cystic Fibrosis is Premature. *Am J Hum Genet, 50*: 438-439.

Billings, Paul R. , Kohn, Mel A., de Cuevas, Margaret, Beckwith, Jonathan, Alper, Joseph S., and Marvin R. Natowicz. 1992. Discrimination as a Consequence of Genetic Testing, *Am. J. Hum. Genet., 50*: 476-482.

Blacker, C.P. 1952. *Eugenics: Galton and After.* Cambridge, Massachusetts: Harvard University Press.

Blanc, Marcel. 1986. *L'Ére de la Génétique.* Paris: Éditions La Découverte.

Blatt, Robin J.R. 1988. *Prenatal Tests. What They Are, Their Benefits and Risks, and How to Decide Whether to Have Them or Not.* New York: Vintage Books.

Bleier, Ruth. 1984. *Science and Gender. A Critique of Biology and Its Theories on Women.* New York: Pergamon Press.

Bleier, Ruth (Ed). 1986. *Feminist Approaches to Science.* New York: Pergamon Press.

Boone, C. Keith. 1988. Bad Axioms in Genetic Engineering. *Hastings Center Report,8* (4)., 9-13.

Botkin, Jeffrey R., and Sonia Alemagno. 1992. Carrier Screenig for Cystic Fibrosis: A Pilot Study of the Attitudes of Pregnant Women. *Am. J. Pub. Health, 82*(5), 723-725.

Boué, A., Françoise Muller, C. Nezelof, J. F. Oury, F. Duchatel, Y Dumez, M. C. Aubry and Joëlle Boué. 1986. Prenatal diagnosis in 200 pregnancies with a 1-in-4 risk of cystic fibrosis. *Hum. Genet., 74*, 288-297.

Boulton, Mary Georgina. 1983. *On Being a Mother.* London: Tavistock Publications.

de Braekeleer, M. and M. Mélançon. 1990. The Ethics of Cystic Fibrosis Carrier Screening: Where do we stand? *Am J Hum Genet, 47*, 581-582.

Brock, David J. H. 1996. Prenatal screening for cystic fibrosis: 5 years' experience reviewed. *The Lancet, 347*, 148-50.

Brody, Eugene B. 1987. New reproductive Technologies and Women's Rights: Framing the Ethical Questions. *Health Care for Women International, 8,*, 277-286.

Bueckert, Dennis. 1993. Life-insurance firms eye genetic testing for clients. *The Gazette*(Montreal), 29 December, B-1.

Bula, Francis. 1992. Gene affects grammar skills, expert says. *The Gazette*(Montreal), February.

Callahan, Daniel. 1990. *What Kind of Life. The limits of medical progress.* Toronto: Simon and Shuster.

Cantin, André. 1992. Research in cystic fibrosis: Immediate and future implications. *Candid Facts, spring.*

Capewell, Gillian. 1986. *Cystic Fibrosis.* London: Office of Health Economics.

Caplan, Arthur L. (Ed.).1978. *The Sociobiology Debate. Readings on Ethical and Scientific Issues.* New York: Harper & Row.

Carter, C.O. 1983. *Developments in Human Reproduction and their Eugenic, Ethical Implications.* London: Academic Press.

Caskey, C. T. et al. 1990. The American Society of Human Genetics Statement on Cystic Fibrosis Screening. *Am. J. Hum. Genet., 46,* 393.

CIBA Foundation Symposium. 1990.. *Human genetic information: Science Law and Ethics.* Ciba Foundation Symposia. New York: John Wiley & Sons. A Wiley-Interscience Publications.

Centre de bioéthique de l"institut de recherches cliniques de Montréal. 1980. *Le diagnostic prénatal.* Cahiers de Bioéthique 2. Québec: Les Presses de l'Université Laval.

Chan, C. K. 1985. Eugenics on the Rise: A Report From Singapore. *International Journal of Health Services, 15,*(4)., 707-712.

Chase et al. 1986. Assessment of Risk by Pregnant Women: Implications for Genetic Counseling and Education. *Social Biology, 33*(spring/summer), 57-64.

Cheney, Peter. (1998, 9 July). Human egg trade lures elite students. *The Globe and Mail,* pp. 1A, 4A.

Chervenak, Frank A. and Laurence B. McCollough. Does obstetric ethics have any arole in the obstetrician's response to the abortion controversy? *Am J Obstet Gynecol, 163*(5 part 1), 1425-9.

Childs et al. 1976. Tay Sachs Screening: Motives for Participating and Knowledge of Genetics and Probability *Am J Hum Genet, 28*, 537-549.

Choronic Villus Sampling CVS.: World Health Orgainzation European Office WHO/EURO. Meeting Statement onthe Use of CVS in Prenatal Diagnosis. *Journal of Assisted Reproduction and Genetics, 9*(4): 1992, 299-301.

Claustres, Mireille, Desgeorges, Marie, Kjellberg, Paule, Tissot, Catherine and Jacques Demaille. 1992. Analysis of 30 known cystic fibrosis mutations: 10 mutations account for 27% of non-◊F508 chromosomes in South France. *Hum Genet, 90*, 464-466.

Clayton, Ellen Wright, Vickie L. Hannig, Jean P. Pfotenhauer, Robert A. Parker, Preston W. Campbell III, and John A. Phillips III. 1996. Lack of Interest by Nonpregnant Couples in Population-Based Cystic Fibrosis Carrier Screening. *Am. J. Hum. Genet., 58*, 617-627.

Clow, Carol et al. 1977. Knowledge About and Attitudes Toward Genetic Screening Among High School Students: The Tay Sachs Experience *Pediatrics, 59*(1), 86-91.

Cobb, E., S. Holloway, R. Elton and J. A. Raeburn. 1991. What do young people think about screeing for cystic fibrosis? *J. Med. Genet., 28*, 322-324.

Code, Lorraine. 1988. Credibility: a double standard. In Lorraine Code, Sheila Mullett and Christine Overall. (Eds.). *Feminist Perspectives. Philosophical Essays on Method and Morals.* Toronto: University of Toronto Press.

Code, Lorraine. 1991. *What Can She Know? Feminist Theory and the Construction of Knowledge.* Ithaca and London: Cornell University Press.

Collins, Francis. 1992. Cystic Fibrosis: Molecular Biology and Therapeutic Implications. *Science, 256*(8 May), 774-9.

Colten, Harvey R. 1990 Screening for Cystic Fibrosis:; Public Policy and Personal Choices. *N Engl J Med, 322*(5), 328-329.

Comité consultatif nationnal d'éthique pour les sciences de la vie et de la santé. 1985. *Journées annuelles d'Ethique 1985.* Paris: Comité consultatif nationnal d'éthique pour les sciences de la vie et de la santé.

Conrad, Peter. 1987. The Noncompliant Patient in Search of Autonomy. *Hastings Center Report, 17*(4), 15-17.

Conrad, Peter and Joseph W. Schneider. 1980. *Deviance and Medicalization: From Badness to Sickness.* Toronto:The C.V. Mosby Company.

Conway, S. P., Allenby, K. Pond, M. N. 1994. Patient and parental attitudes toward genetic screening and its implciations at an adult cycstic fibrosis centre. *Clin Genet, 45*, 308-312.

Conseil du statut de la femme. 1988. *Sortir la maternité du laboratoire.* Gouvernement du Québec.

Cooke, Michael. 1993. Ethical Issues of Prenatal Diagnosis for Predictive Testing for Genetic Disorders of Late Onset, vol. 14, *Technologies of Sex Selection and Prenatal Diagnosis,* Research Studies of the Royal Commission on New Reproductive Technologies: Ottawa.

Corea, Gena. 1985. *The Mother Machine. Reproductive Technologies from Artificial Insemination to Artificial wombs.* New York: Harper & Row.

Corea, Gena et al. 1987. *Man-Made Women. How New Reproductive Technologies Affect Women.* Indianapolis: Indiana University Press.

Couture, A., Veyrac, C. et al. 1985. *Les malformations congénitales. Diagnostic anténatal et devenir.* Paris: Sauramps Medical.

Cozby, Paul C. 1997. *Methods in Behavioral Research.* (6th ed.) Toronto: Mayfield Publishing Company.

Cutting, Garry R. et al. 1992. Analysis of Four Diverse Population Groups Indicates that a Subset of Cystic Fibrosis Mutations Occur in common among Caucasians. *Am J Hum Genet, 50,* 1185-1194.

Cystic Fibrosis Genetic Analysis Consortium. 1990. Worldwide Survey of the ◊F508 Mutation--Report from the Cystic Fibrosis Genetic Analysis Consortium. *Am. J. Hum. Genet., 47*(2), 354-359.

Danziger, Sandra Klein. 1986. The Uses of Expertise in Doctor-Patient Encounters During Pregnancy. In Peter Conrad and Rochelle Kern. (Eds.). *The Sociology of Health and Illness.* New York: St. Martin's Press.

Davies, Kevin. 1989. The search for the cystic fibrosis gene. *New Scientist, 124*(1687), 54-58.

Decruyenaere, M., Evers-Kiebooms, G., Denayer, L. and H. Van den Berghe. 1992. Cystic fibrosis: community knowledge and attitudes towards carrier screening and prenatal diagnosis. *Clin Genet, 41,* 189-196.

Denayer, L., Evers-Kiebooms, G., De Boeck, K. and H. Van den Berghe. 1992. Reproductive Decision Making of Aunts and Uncles of a Child with Cystic Fibrosis: Genetic Risk Perception and Attitudes Toward Carrier Identification and Prenatal Diagnosis. *Am J Med Genet, 44,* 104-111.

Denayer, Lieve, Evers- Kiebooms and Herman Van den Berghe. 1990. A child with cystic fibrosis: I. Parental knowledge about the genetic transmission of CF and about DNA-diagnostic procedures. *Clin Genet, 37*: 198-206.

Deleuze, Gilles. 1986. *Foucault.* Minneapolis: Unversity of Minnesota Press.

Devault, Marjorie L. 1990. Talking and Listening from Women's Standpoint: Feminist Strategies for Interviewing and Analysis, *Social Problems, 37*(1), 96-116.

Digest. sept./oct.1984. Most Americans Remain Opposed to Abortion Ban. *Family Planning Perspectives, 16*(5), 233-234.

Digest. may/june 1988. CVS Prenatal Diagnosis Seen to Have Low Rates of Failure, Fetal Loss. *Family Planning Perspectives, 20*(3), 145-6.

Digest. july/august 1989a. Chorionic Villus Sampling is Useful Alternative to Amniocentesis. *Family Planning Perspectives, 21*(4), 188-9.

Digest. july/august 1989b. Performing a Second Ultrasound Exam Markedly Improves Prenatal Detection of Fetal Malformations. *Family Planning Perspectives, 21*(4), 280.

"Don't participate in midwife projects: two doctors' groups", *The Montreal Gazette*, 18 September 1990, A-6.

Dugger, Celia W. 1991. Infant Mortality in New York City Declines for First Time in 4 Years. *New York Times.* April 20.

Dutton, Diana B. 1988. *Worse than the Disease. Pitfalls of Medical Progress.* New York: Cambridge University Press.

Ehrenreich, Barbara and Deirdre English. 1978. *For Her Own Good. 150 Years of the Experts' Advice to Women.* New York: Doubleday.

Eiklid, Kristin et al. 1993. Frequency of the ◊F508 and exon 11 mutations in Norwegian cystic fibrosis patients. *Clin Genet, 44*(1), 12-14.

Ekwo, Edem E., Brenda F. Seals, Jae-On Kim, Roger A. Williamson and James W. Hanson. 1985. Factors Influencing Maternal Estimates of Genetic Risk. *Am J Med Genet, 20,* 491-504.

Elkington, John. 1985. *The Gene factory. Inside the Biotechnology Business.* London: Century Publishing.

Ellul, Jacques. 1980. *The Technological System.* New York: Continuum.

Eng, Christine M., Clyde Schechter, Jane Robinowitz, George Fulop, Tania Burgert, Brynn Levy, Randi Zinberg, and Robert J. Desnick. 1997. Prenatal Genetic Carrier Testing Using Triple Disease Screening. *JAMA, 278*(15), 1268-1272.

Evers-Kiebooms, Gerry. 1987. Decision making in Huntington's disease and cystic fibrosis. In G. Evers-Kiebooms, J. J. Cassiman, H. Van den Berghe and G. d'Ydewalle (Eds.). *Genetic Risk, Risk Perception and Decision Making. Birth Defects, 23*(2), 115-149.

Ewigman, Bernard G. et al. 1993. Effect of Prenatal Ultrasound Screening on Perinatal Outcome. *New Eng J Med, 329*(12), 821-7.

Farganis, Sondra. 1989. Feminism and the Reconstruction of Social Science In Alison M. Jaggar and Susan R. Bordo. (Eds.). *Gender/Body/Knowledge. Feminist Reconstructions of Being and Knowing.* Boston and London: Rutgers University Press.

Farrant, Wendy. 1985. Who's for amniocentesis? The Politics of Prenatal Screening. In Hilary Homans. (Ed.). *The Sexual Politics of Reproduction.* Aldershot: Gower Publishing.

1980. Importance of Counselling in Antenatal Screening. *MIMS* Magazine. June 15, pp. 55-63.

Fee, Elizabeth. 1986. Critiques of Modern Science: The Relationship of Feminism to Other Radical Epistemologies. In Ruth Bleier. (Ed.). *Feminist Approaches to Science.* New York: Pergamon Press.

Finger, Anne. 1984. Claiming all of our bodies: Reproductive rights and disabilities. In Rita Arditti, Renate Klein & Shelley Minden. (Eds.). *Test-Tube Women. What Future for Motherhood?* London: Pandora Press.

Finger, Anne. 1990. *Past Due. A Story of Disability, Pregnancy and Birth.* Seattle: Seal Press.

Finkelstein, Janice and Patricia Clough. 1983. Foetal Politics and the Birth of an Industry. *Women's Studies International Forum, 6* (4).

Foucault, Michel. 1978. *The History of Sexuality. Vol.1.* New York: Vintage Books.

1977. *Discipline and Punish.* New York: Vintage Books.

1973. *The Birth of the Clinic.* New York: Vintage Books.

Forester, John. 1986. On a Critical Theory of Health Care and Social Policy: A Comparison of Habermas and Illich. In John w. Murphy, Algis Mickunas and Joseph J. Pilotta. (Eds.). *The Underside of High-Tech. Technology and the Deformation of Human Sensibilities.* New York: Greenwood Press.

Fox Keller, Evelyn. 1989. The Gender/Science System: or, Is Sex to Gender as Nature Is to Science? In Nancy Tuana. (Ed.). *Feminism and Science.* Bloomington and Indianapolis: Indiana University Press.

Fox Keller, Evelyn. 1985. *Reflections on Gender and Science.* New Haven and London: Yale University Press.

Franklin, Ursula. 1990. *The Real World of Technology.* CBC Massey Lecture Series. Anansi: Concord, Ont.

Freeman, Roger. 1990. Intrapartum Fetal Monitoring-A Disappointing Story, *New England Journal of Medicine, 322*(9), 624-666.

Frézal, Jean & Briard, Marie-Louise 1985. Malformations congénitales et conseil génétique. In *Le Conseil Génétique,* 61-64.

Frézal, Jean & Briard, Marie-Louise 1986. Aspects éthiques du diagnostic prénatal. Chapitre 21. In *Le Diagnostic prénatal.* Mattei, J. F., & Dumez, Y. Progrès en pédiatre, 1 1986. pp 269-276. Paris: DOIN éditeurs.

Furhmann, W. 1989. Impact, logistics and prospects of traditional prenatal diagnosis. *Clinical Genetics, 36,* 378-385.

Galton, Francis 1907 [1883]. *Inquiries into Human Faculty and its Development.* New York: MacMillan & Company.

Gavarani, Laurence. 1987. Les enfants que je veux si je peux. In *Sortir La Maternité du Laboratoire.* Quebec: Conseil du statut de la femme.

Gerson, Kathleen. 1985. *Hard Choices. How Women Decide about Work, Career, and Motherhood.* Berkeley: University of California Press.

Gilbert, Fred. 1994. Cystic Fibrosis Carrier Screening: Comparability of Data and Uniformity of Testing. *Am J Hum Genet, 54,* 925-927.

Gilligan, Carol. 1982. *In a Different Voice: Psychological Theory and Women's Development.*Cambridge, MA: Harvard University Press.

Ginzberg, Ruth. 1989. Uncovering Gynocentric Science, In Nancy Tuana. (Ed.). *Feminism and Science.* Bloomington and Indianapolis: Indiana University Press.

Goffman, Erving. 1968. *Asylums.* Chicago: Aldine Publishing Company.

Gonen, Julianna S. 1993. Women's Rights vs. "Fetal Rights": Politics, Law and Reproductive Hazards in the Workplace. In *The Politics of Pregnancy: Policy Dilemmas in the Maternal-Fetal Relationship*, eds. Janna C. Merrick and Robert H. Blank, New York: Harrington Park Press.

Goodchild, Mary C. 1975. *Cystic Fibrosis: A manual of diagnosis and management.* Philadelphia: Bailliere Tindall.

Goodfield, June. 1977. *Genetic Engineering and the Manipulation of Life.* New York: Random House.

Goodman, Madeline J. and Lenn E. Goodman. 1982. The Overselling of Genetic Anxiety.*The Hasting Center Report*, (Oct.), 20-27.

Gordon, Tuula. 1990. *Feminist Mothers.* New York: New York University Press.

Grant, Karen. 1993. Perceptions, Attitudes, and Experiences of Prenatal Diagnosis: A Winnipeg Study of Women Over 35, vol. 12, *Prenatal Diagnosis: Background and Impact on Individuals.* Research Studies of the Royal Commission on New Reproductive Technologies: Ottawa.

Grebe, Theresa A. et al. 1992. Mutation Analysis of the Cystic Fibrosis Transmembrane Regulator Gene in Native American Populations of the Southwest. *Am. J. Hum. Genet., 51,* 736-740.

Green, Josephine 1992. Principles and practicalities of carrier screening: attitudes of recent parents. *J Med Genet, 29,* 313-319.

1990. *Calming or Harming? A critical review of pychological effects of fetal diagnosis on pregnant women.* Galton Institute Occasional Papaer, Second Series No.2.London: The Galton Institute.

Grimes, David A. 1993. Technology Follies. The Uncritical Acceptance of Medical Innovation. *JAMA, 269*(23), 3030-3033.

Guillermit, H., P. Fanen, and C. Ferec. 1990. A 3' splice site consensus sequence mutation in the cystic fibrosis gene. *Hum. Genet., 85,* 450-453.

Hamdi, I., Payne, S.J., Barton, D.E., McMahon, R., Green, M., Shneerson, J.. M., and C. N. Hales. 1993. Genotype analysis in cystic fibrosis in relation to the occurrence of diabetes mellitus. *Clin Genet, 43* (4), 186-189.

Hamerton, John L., Evans, Jane A. and Leonie Stranc. 1993. Prenatal Diagnosis in Canada--1990: A Review of Genetics Centres, vol. 13, *Current Practice of Prenatal Diagnosis in Canada,* Research Studeis of the Royal Commission on New Reproductive Technologies: Ottawa.

Hamilton, Joan O'C. (26 sept.) 1994. Biotech: An Industry Crowded With Players Faces An Ugly Reckoning, *Business Week,* 84.

Hamosh, Ada et al. 1992. Cystic Fibrosis Patients Bearing Both the Common Missense Mutation Gly->Asp at Codon 551 and the ◊F508 Mutation Are Clinically Indistinguishable from ◊F508 Homozygotes, Except for Decreased Risk of Meconium Ileus. *Am. J. Hum. Genet., 51*, 245-250.

Hancock, R.E.W. 1991. CCFF program designed to enhance access to transplantation services. *Candid Facts, spring*:2, 6-11.

Handyside, Andy H. et al. 1992. Birth of a Normal Girl After In Vitro Fertilization and Preimplantation Diagnostic Testing for Cystic Fibrosis. *New Eng J Med, 327*(13), 905-909.

Hanmer, Jalna. 1987. Transforming conciousness: women and the new reproductive technologies. In Genea Corea et al. (Eds.). *Man-Made Women. How New Reproductive Technologies Affect Women*. Bloomington and Indianapolis: Indiana University Press.

Haraway, Diane. 1985. A Manifesto for Cyborgs: Science, Technology and Socialist Feminism in the 1980s. *Socialist Review, 80*, 65-107.

Harding, Sandra. 1986. *The Science Question in Feminism*. Ithaca and London: Cornell University Press.

1987. Introduction. Is There a Feminist Method? Chap. in *Feminism and Methodology. Social Science Issues*. Bloomington and Indianapolis: Indiana University Press.

1989. Is There A Feminist Method? In Nancy Tuana. (Ed.). *Feminism and Science*.. Bloomington and Indianapolis: Indiana University Press.

Harper, Peter S., 1990. Genetic services in the community. *J Med Genet, 27*, 473-474.

Harris, John. 1993. Is Gene Therapy a Form of Eugenics? *Bioethics, 7* (2/3): 178-187.

Harris, Ann and Maurice Super. 1991. *Cystic Fibrosis*. Oxford: Oxford University Press.

Harrison, Dr. Michael R. 1982. Unborn: Historical perspective of the fetus as a patient. *The Pharos, 45*,(19), 19-24.

Harsanyi, Dr. Zsolt & Richard Hutton. 1981. *Genetic Prophecy. Beyond the Double Helix.* New York: Rawson, Wade Publishers.

Heirich, Max. 1979. Why We Avoid the Key Questions: How Shifts in Funding of Scientific Inquiry Affect Decision Making about Science. In David A. Jackson & Stephen P Stich. (Eds.). *The Recombinant DNA Debate.*. Englewood Cliffs, New Jersey: Prentice-Hall Inc.

Henifin, M. S. 1987. Prenatal screening. Special issue. *Genewatch, 4*,(1).

Holtzman, Neil A. 1989. *Proceed With Caution. Predicting Genetic Risks in the Recombinant DNA Era.* Maryland: John Hopkins Unversity Press.

Hubbard, Ruth. 1989. Science, Facts and Feminism. In Nancy Tuana. (Ed.). *Feminism and Science*..Bloomington and Indianapolis: Indiana University Press.

1987. Eugenics: New Tools, Old Ideas. *Women & Health, 13* (1-2), 225-235.

1986. Eugenics and Prenatal Testing *International Journal of Health Services, 16*(2), 227-242.

1985. Prenatal Diagnosis and Eugenic Ideology, *Women's Studies International Forum, 8*, (6), 567-576.

1984. Personal courage is not enough: some hazards of childbearing in the 1980s. In Rita Arditti, Renate Klein & Shelley Minden. (Eds.). *Test-Tube Women. What Future for Motherhood?* London: Pandora Press.

1982. The Fetus as Patient. *Ms.* (October), 28-32.

Hubbard, Ruth, Mary Sue Henifin and Barbara Fried (Eds.) 1979. *Women Look at Biology Looking at Women.* Boston: Schenkman Publishing Co.

Hubbard, Ruth and Elijah Wald. 1993. *Exploding the Gene Myth.* Boston: Beacon Press.

Huntington, Ellsworth. 1935. *Tomorrow's Children The Goal of Eugenics.* New York: John Wiley & Sons.

Illich, Ivan. 1975. *Medical Nemesis. The Expropriation of Health.* Toronto: McClelland and Stewart.

Isambert, F.A. 1980. Éthique et génétique. De l'utopie eugénique au contrôle des malformations congénitales. *Revue française de Sociologie, XXI*:331-354.

Isambert, F. A. et al. 1980. *L'amniocentèse et les médecins. Le diagnostic prénatal des malformations congénitales.* Ethique et Pratiques symboliques. Cahier no 1. Paris: CNRS, centre d'Etudes Sociologiques.

Jacquard, Albert 1984. La tentation eugénique. *Futuribles, Juillet-Août,* 61-71.

Jackson, David A. and Stephen P. Stich. (Eds.) 1979. *The Recombinant DNA Debate.* Englewood Cliffs, New Jersey: Prentice-Hall, Inc.

Jaggar, Alison M. and Susan Bordo. (Eds.). 1989. *Gender/Body/Knowledge.Feminist Reconstructions of Being and Knowing.* New Brunswick and London: Rutgers University Press.

Jayaratne, Toby Epstein and Abigail J. Stewart. 1991. Quantitative and Qualitative Methods in the Social Sciences. Current Feminist Issues and Practical Strategies. In Mary Margaret Fonow and Judith A. Cook.(Eds.). *Beyond Methodology. Feminist Scholarship As Lived Research.* Bloomington and Indianapolis: Indiana University Press.

Johnsen, Dawn. 1987. A New Threat to Pregnant Women's Autonomy. *Hastings Center Report,* (August), 33-40

Julian, Claire, Marie-Claude Tordo, Geneviève Macquart-Moulin, Jean-Paul Moatti, Francis Giraud and Ségolène Aymé. 1989. Factors Influencing Genetic Counseling Attendance Rate: A Geographically Based Study. *Social Biology, 36*(3-4),240-247.

Kaback et al. 1974. Tay Sachs disease: A model for the control of recessive genetic disorders. In Arno G. Motulsky and W. Lenz. (Eds.). *Birth Defects* . Excerpta Medica: Amsterdam, 248-261.

Kaplan, Feige. 1992. Cystic Fibrosis: Schoolyard Experiences. presented at the Conference on Cystic Fibrosis: "La découverte du gène: qu'est ce que cela change?" organized by the Centre interuniversitaire de recherches sur les populations, SOREP, Montreal, May 29 1992.

Kaplan, Feige, Clow, Carol and Charles C. Scriver. 1991. Cystic Fibrosis Carrier Screening by DNA Analysis: A Pilot Study of Attitudes among Participants. *Am. J. Hum. Genet., 49*, 240-242.

Katz, J. 1984. *The Silent World of Doctor and Patient.* New York: The Free Press.

Kerem, Eitan and Abbyann Lynch. 1991. Screening for Cystic Fibrosis: Ethical and Social Issues. *Am Rev Resp Dis, 143*(3), 457-460.

Kessler, Seymour. 1980. The Psychological Paradigm Shift in Genetic Counseling. *Social Biology, 27*(3), 167-185.

Kessler, S. & H. Kessler. 1988. Psychosocial Developments in the Field of Genetic Counselling.In E.K. Hicks & J.M. Berg. (Eds.). *The Genetics of Mental Retardation. Biomedical,Psychosocial and Ethical Issues.* Boston: Kluwer Academic Publishers.

Kevles, Daniel. 1986. *In the Name of Eugenics.* New York: Alfred A. Knopf.

Kitch, Sally L. 1991. Does War Have Gender? In Anne E. Hunter. (Ed.). *Genes and Gender VI: On Peace, War and Gender. A Challenge to Genetic Explanations.* New York: Feminist Press.

Klein, Renate. (Ed.). 1989. *Infertility. Women speak out about their experiences of reproductive medicine.* London: Pandora Press.

Kolata, Gina. 1990. *The Baby Doctors. Probing the Limits of Fetal Medicine.* New York: Delacorte Press.

Kolodny, Edwin H. 1979. Tay Sachs Disease In R.M. Goodman and A.G. Motulsky.(Eds.). *Genetic Diseases Among Ashkenazi Jews.* Raven Press: New York.

Kuhn, Thomas. 1970. *The Structure of Scientific Revolutions.* Chicago: Chicago University Press.

Lanng, Susanne, Schwartz, Marianne, Thorsteinsson and Christian Koch. 1991. Endocrine and exocrine pancreatic function and the ◊F508 mutation in cystic fibrosis. *Clin Genet, 40,* 345-348.

Law Reform Commission of Canada. 1989. *Crimes against the foetus.* Working Paper 58, Canada.

Leclerc, B., Mélançon M.J. & R. Gagné 1989. Les Cahiers scientifiques de l'ACFAS. *Génétique et Éthique. identification et thérapie des maladies génétiques.* Montreal: Groupe de Recherche en Génétique et Éthique.

Levine, Sol. 1987. The Changing Terrains in Medical Sociology: Emergent Concern with Quality of Life. *Journal of Health and Social Behaviour, 28*(march), 1-6.

Lindner, M. , Wolf, A., Moh, B., Steinbach, P., Kleihauer, E., Bartram, C. R., and A. E. Kulozik. 1992. The specturm of CFTR mutations in south-west German cystic fibrosis patients. *Hum Genet, 90,* 267-269.

Lippman, Abby.1991. Prenatal Genetic Testing and Screening: Construcing Needs and Reinforcing Inequalities. *American Journal of Law and Medicine, 17*(1-2), 15-50.

1989. Prenatal Diagnosis: Tough Questions For All Women. *Current Therapy, March*:24-25.

1987. Prenatal Diagnosis: Reproductive Choice? Reproductive Control? In Christine Overall. (Ed.). *The Future of Human Reproduction.* Toronto: The Women's Press.

Lippman-Hand, Abby and Martha Piper. 1981. Prenatal Diagnosis for the detection of Down's syndrome: Why are so few eligible women tested? *Prenatal Diagnosis, 1,* 249-257.

Lippman-Hand, Abby and F. Clarke Fraser. 1979a. Genetic Counseling--The Postcounseling Period: I. Parents' Perceptions of Uncertainty. *Am J Med Genet, 4*:51-71.

1979b. Genetic Counseling--The Postcounseling Period: II. Making Reproductive Choices. *Am J Med Genet, 4*:73-87.

Loader, S., P. Caldwell, A. Kozyra, J. C. Levenkron, C. D. Boehm, H. H. Kazazian, Jr., and Peter T. Rowley. 1996. Cystic Fibrosis Carrier Population Screening in the Primary Care Setting. *Am. J. Hum. Genet., 59*, 234-247.

Lorenz, Robert P., John J. Botti, Cheryl M. Schmidt and Roger L. Ladda. 1985. Encouraging Patients to Undergo Prenatal Genetic Counseling Before the Day of Amniocentesis. *The Journal of Reproductive Medicine, 30* (12), 933-935.

Ludmerer, Kenneth M. 1972. *Genetics and American Society. A Historical Appraisal.* Baltimore and London: Johns Hopkins University Press.

MacKay, Ian Ferguson & F. Clarke Fraser. 1993. The History and Evolution of Prenatal Diagnosis, vol. 12, *Prenatal Diagnosis: Background and Impact on Individuals.* Research Studies of the Royal Commission on New Reproductive Technologies: Ottawa.

Mahowald, Mary. 1989. Beyond Abortion: Refusal of Caesarean Section. *Bioethics, 3*(2), 106-121.

Marion, Janet P. et al. 1980. Acceptance of amniocentesis by low-income patients in an urban hospital. *Am J Obstet Gynecol, 138*(1), 11-15.

Marshall, Catherine and Gretchen B. Rossman. 1989. *Designing Qualitative Research.* London: Sage Publications.

Marteau, Theresa, Jane Kidd, Rachel Cook, Susan Michie, Marie Johnston, Joan Slack and R. W. Shaw. 1991. Perceived risk not actual risk predicts uptake of amniocentesis. *Brit. J. Obs. Gyn., 98*, 282-286.

Mason, J.K. 1990. *Medico-Legal Aspects of Reproduction and Parenthood.* Dartmouth: Aldershot.

McCarthy, E. Jerome, Stanley J. Shapiro and William D. Perreault, Jr. 1986. *Basic Marketing*, (4th edition). Homewood, Illinois: Irwin.

McLaren, Angus. 1990. *Our Own Master Race. Eugenics in Canada, 1885-1945*. Toronto: McLelland & Stewart.

McNay, Lois. 1992. *Foucault and Feminism*. Boston: Northeastern University Press.

McQueen, David V. 1975. Social Aspects of Genetic Screening for Tay Sachs Disease: The Pilot Community Screening Program in Baltimore and Washington. *Social Biology*, *22*(2), 125-133.

Medline, Elaine. 1993. Canada, U.S. approve new drug treatment for CF. *The Gazette*(Montreal), 31 December.

Mennie, Moira et al. 1993. Prenatal screening for cystic fibrosis: attitudes and responses of participants. *Clin Genet*, *44*(2), 102-106.

Mennie, Moira E. et al. 1992a. Prenatal screening for cystic fibrosis. *The Lancet, 340*, 214-216.

Mennie, Moira E. et al. 1992b. Prenatal cystic fibrosis carrier testing: designing an information leaflet to meet the specific needs of the target population. *J Med Genet, 29*, 308-312.

Meryash, David L. 1989. Perception of burden among at-risk women of raising a child with fragile-X syndrome. *Clinical Genetics, 36*, 15-24.

Metheny, William P., Gerald B. Holzman, Jeffrey Taylor, William Young and James V. Higgins. 1988. Amniocentesis Use and Risk Awareness: Comparison of Knowledge and Beliefs Among Older Gravida. *Social Biology*, *35*(1-2), 50-61.

Mies, Maria. 1985. Why do we need all this? A call against genetic engineering and reproductive technology. *Women's Studies International Forum*, *8*(6), 553-560.

Mies, Maria. 1991. Women's Research or Feminist Research? The Debate Surrounding Feminist Science and Methodology. In Mary Margaret Fonow and Judith A. Cook. (Eds.). *Beyond Methodology. Feminist Scholarship As Lived Research.* Bloomington and Indianapolis: Indiana University Press.

Miles, Matthew B. and A. Michael Huberman. 1984. *Qualitative Data Analysis. A Sourcebook of New Methods.* London: Sage Publications.

Miller, Shelley R. and Robert H. Schwartz. 1992. Attitudes Toward Genetic Testing of Amish, Mennonite, and Hutterite Families with Cystic Fibrosis. *Am J Public Health, 82*: 236-242.

Miller, Z. G. and F. C. Fraser. 1993. Attitudes of Genetic Counsellors with Respect to Prenatal Diagnosis of Sex for Non-Medical Reasons, vol. 14, *Technologies of Sex Selection and Prenatal Diagnosis,* Research Studies of the Royal Commission on New Reproductive Technologies: Ottawa.

Millman, Marcia and Rosabeth Moss Kanter. 1987. Introduction to Another Voice. Feminist Perspectives on Social Life and Social Science. In Sandra Harding. (Ed.). *Feminism and Methodology. Social Science Issues.* Bloomingtn and Indianapolis: Indiana University Press.

Milner, Joanne. 1993. A Review of Views Critical of Prenatal Diagnosis and Its Impact on Attitudes Toward Persons With Disabilities, vol. 12, *Prenatal Diagnosis: Background and Impact on Individuals.* Research Studies of the Royal Commission on New Reproductive Technologies: Ottawa.

Minden, Shelley. 1984. Designer Genes: A View From the Factory. In Rita Arditti, Renate Klein & Shelley Minden. (Eds.). *Test-Tube Women. What Future for Motherhood.* London: Pandora Press.

1985. Genetic Engineering and Human Embryos, *Science for the People, 17*(3).

Mitchell, John J., Annie Capua, Carol Clow and Charles R. Scriver. 1996. Twenty-Year Outcome Analysis of Genetic Screening Programs for Tay-Sachs and ß-Thalassemia Disease Carriers in High Schools. *Am. J. Hum. Genet., 59*, 793-798.

Mitchell, John, Scriver, Charles R., Clow, Carol L. and Feige Kaplan. 1993. What young people think and do when the option for cystic fibrosis carrier testing is available. *J Med Genet, 30*, 101-105.

Moatti, J.P., Le Gales, C., C. Julian, J.P. Durbec, J.F. Mattei and S. Ayme. 1990. Socio-cultural Inequities in Access to Prenatal Diagnosis: The Role of Insurance Coverage and Regulatory Policies. *Prenatal Diagnosis, 10*(5), 313-325.

Modell, B. and A.M. Kuliev. 1989. Impact of Public Health on Human Genetics. *Clinical Genetics, 36*, 286-298.

Mooney, Gavin and Mette Lange. 1993. Ante-Natal Screeing: What Constitutes Benefit? *Soc. Sci. Med., 37*(7), 873-878.

Moore, Thomas J. 1989. *Heart Failure. A critical inquiry into American medicine and the revolution in heart care.* New York: Random House.

Motulsky, Arno G. 1997. Screening for Genetic Diseases. *New Eng. J. Med., 336*(18), 1314-1316.

Munson, Ronald and Lawrence H. Davis. 1992. Germ-Line Gene Therapy and the Medical Imperative. *Kennedy Institute of Ethics Journal, 2*(2): 137-158.

Murphy, Edmond, A. 1978. Eugenics. An Ethical Analysis. *Mayo Clinic Proc, 53*, 655-664.

Murphy, Jane. 1984. From mice to men? Implications of progress in cloning research. In Rita Arditti, Renate Klein & Shelley Minden. (Eds.). *Test-Tube Women. What Future for Motherhood?* London: Pandora Press.

Murphy, John, Algis Mickunas and Joseph J. Pilotta 1986. Conclusion: Fundamentals of a Responsible Technology In John Murphy, Algis Mickunas and Joseph J. Pilotta. (Eds.). *The Underside of High-Tech. Technology and the Deformation of Human Sensibilities.* b New York: Greenwood Press.

Myers, Melanie F., Bernhardt, Barbara A., Tambor, Ellen S. and Neil A. Holtzman. 1994. Involving Consumers in the Development of an Educational Program for Cystic Fibrosis Carrier Screening. *Am. J. Hum. Genet., 54,* 719-726.

Najman, J.M., D. Klein and C. Munro. 1982. Patient Characteristics. *Social Science and Medicine, 16*(20), 1781-89.

Nelkin, Dorothy and Laurence Tancredi. 1989. *Dangerous Diagnostics. The Social Power of Biological Information.* New York: Basic Books.

Newman, Lucile F. 1987. Framing the Ethical Issues in New Reproductive Technologies. *Health Care for Women International, 8,* 287-292.

New Scientist. 2 June 1990. Gene Screen. *126*(1719), 27.

Nielsen, Caroline C. 1981. An Encounter With Modern Medical Technology: Women's Experiences With Amniocentesis. *Women and Health, 6*(2), 109-124.

NIH Consens Statement Online 1997 April 14-16[98 06 08]; 15(4): in press.

Nolan, Kathleen & Swenson, Sara. 1988. New Tools, New Dilemmas: Genetic Frontiers. *Hasting Center Report, oct/nov.,* 40-46.

Nuttbrock, Larry A.1986. The Growth and Control of Medical Technology, In John W. Murphy, Algis Mickunas, and Joseph J. Pilotta. (Eds.). *The Underside of High-Tech. Technology and the Deformation of Human Sensibilities.* . New York: Greenwood Press.

Oakley, Ann. 1987. From walking Womb to Test-Tube Babies. In Michelle Stanworth. (Ed.). *Reproductive Technologies. Gender, Motherhood and Medicine.* Minneapolis: University of Minnesota Press.

1986. The History of Ultrasonography in Obstetrics. *Birth, 13*(1 March), 8-13.

1981. Interviewing women: a contradiction in terms, In Helen Roberts. (Ed.).*Doing Feminist Research.* Boston: Routledge & Kegan Paul.

Opatrny, Dennis. 1991. Are we coded to die around the age of 85? *The Gazette*(Montreal), 7 December, H-11.

Overall, Christine. 1989. *The Future of Human Reproduction.* Toronto: The Women's Press.

1987. *Ethics and Human Reproduction. A Feminist Analysis.* London: Allen & Unwin.

Palys, Ted. 1997. *Research Decisions: quantitative and qualitative perspectives.* (2nd ed.) Montreal: Harcourt Brace Canada.

Pannisset, Isabelle. 1992. Le Dépistage des porteurs: Aspects socio-juridique et éthiques. *Santé Vous Bien* .

Passarge, E., B. Eckerland and U. Stephan. 1984. Genetic counselling in cystic fibrosis. *Eur J Pediatr, 143*(1), 54-57.

Patychuk, Dianne. 1985. Ultrasound: The First Wave. *Healthsharing, 6*(4).

Pauker, Susan Perlmutter and Stephen G. Pauker. 1979. The Amniocentesis Decision: An Explicit Guide for Parents. In Charles J. Epstein et al. (Eds.). *Risk, Communication and Decision Making in Genetic Counseling.* The National Foundation-March of Dimes. Birth Defects: Original Article Series. Volume XV, Number 50. Alan R. Liss, Inc.

Perry, Terry B., Michel J.J. Vekemans, Abby Lippman, Emily Hamilton and Paul J.R. Fournier. 1985. Chorionic villi sampling: Clinical experience, immediate complications, and patient attitudes. *Am J Obstet Gynecol, 151*(15 january), 161-5.

Petchesky, Rosalind Pollack. 1987. Foetal Images: the Power of Visual Culture in the Politics of Reproduction. In Michelle Stanworth. (Ed.). *Reproductive Technologies. Gender, Motherhood and Medicine.* Minneapolis: University of Minnesota Press.

Phipps, Sean. 1981. Mourning Response and Intervention in Stillbirth: An Alternative Genetic Counseling Approach. *Social Biology, 28*(1-2), 1-13.

Pickens, Donald K. 1968. *Eugenics and the Progressives.* Nashville, Tennesee: Vanderbilt University Press.

Post, Stephen G. 1992. Huntington's Disease: prenatal screening for late onset disease. *Journal of Medical Ethics, 18*, 75-78.

Postman, Neil. 1992. *Technopoly. The Surrender of Culture to Technology.* New York: Alfred A. Knopf.

Porter, Ian H. 1982. Control of Hereditary Disorders. *Ann Rev Public Health, 3*, 277-319.

President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research. 1983. *Screening and Counseling for Genetic Conditions. A Report on the Ethical, Social, and Legal Implications of Genetic Screening, Counseling, and Education Programs.*

Prior, Lynn. 1993a. Screening for Genetic Susceptibilities to Common Diseases, vol. 14, *Technologies of Sex Selection and Prenatal Diagnosis*, Research Studies of the Royal Commission on New Reproductive Technologies: Ottawa.

Prior, Lynn. 1993b. Somatic and Germ Line Gene Therapy: Current Status and Prospects,vol. 14, *Technologies of Sex Selection and Prenatal Diagnosis*, Research Studies of the Royal Commission on New Reproductive Technologies: Ottawa.

Purdy, Laura M.1990. Are Pregnant Women Fetal Containers? *Bioethics*, *4*(4), 273-291.

Quéniart, Anne. 1988. *Le Corps Paradoxal. Regards de femmes sur la maternité*. Montreal: Éditions Saint-Martin.

Quéniart, Anne. 1987. La technologie: une réponse à l'insécurité des femmes? In Francine Saillant and Michel O'Neill. (Eds.). *Accoucher Autrement. Repères Historiques,Sociaux et Culturels de la Grossesse et de l'Accouchement au Québec*. Montreal: Éditions Saint-Martin.

Rachlis, Michael and Carol Kushner. 1989. *Second Opinion. What's wrong with Canada's Health care system and how to fix it.* Toronto: Harper Collins.

Rapp, Rayna. 1984. XYLO: a true story. In Rita Arditti, Renate Klein & Shelley Minden. (Eds.). *Test-Tube Women. What Future for Motherhood?* London: Pandora Press.

1988. Moral Pioneers: Women, Men and Fetuses on a Frontier of Reproductive Technology. In Elaine Hoffman Baruch, Amadeo F. D'Adamo & Joni Seager. (Eds.). *Embryos, Ethics and Women's Rights. Exploring the New Reproductive Technologies*. New York: Harrington Park Press.

Raskin, S., Phillips, J. A., Krishnamani, M. R. S. et al. 1993. DNA Analysis of Cystic Fibrosis in Brazil by Direct PCR Amplification From Guthrie Cards. *Am. J. Med. Genet, 46*, 665-669.

Renaud, Marc et al. 1992. *Les Médecins Canadiens Devant le Diagnostic Prénatal: Prudence et Ambivalence*, Rapport de recherche à la Commission royale d'enquête sur les nouvelles techniques de reproduction. Montreal.

Renaud, Marc, Bouchard, Louise, Bisson, Jocelyn, Labadie, Jean-Francois, Dallaire, Louis and Natalie Kishchuk. 1993. Canadian Physicians and Prenatal Diagnosis: Prudence and Ambivalence, vol. 13, *Current Practice of Prenatal Diagnosis in Canada*, Research Studies of the Royal Commission on New Reproductive Technologies: Ottawa.

Reiser, Stanley Joel. 1978. *Medicine and the reign of technology.* Cambridge: Cambridge University Press.

Restagno, G., Garnerone, S., Gennaro, C., Ferrone, M., and A. Carbonara. 1992. Analysis of CA/GT microsatellite polymorphism in IVS8 of the cystic fibrosis transmembrance conductance regulator (CFTR) gene: a study of Italian CF families. *Clin Genet, 42*, 309-313.

Riessman, Catherine Kohler. 1983. Women and Medicalization: A New Perspective. *Social Policy,*(Summer), 3-18.

Riessman, Catherine Kohler. 1987. When Gender is Not Enough: Women Interviewing Women *Gender and Society, 1*, 172-207.

Roberts, Leslie. 1990. CF Screening Delayed for Awhile, Perhaps Forever. *Science, 247*, 1296-1297.

Rona, R. J. et al. 1994. The influence of genetic counselling in the era of DNA testing on knowledge, reproductive intentions and psychological wellbeing. *Clin Genet, 46*(2), 198-204.

Robertson, John. 1988. Procreative Liberty, Embryos, and Collaborative Reproduction: A Legal Perspective. In Elaine Hoffman Baruch, Amadeo F. D'Adamo & Joni Seager. (Eds.). *Embryos, Ethics and Women's Rights. Exploring the New Reproductive Technologies.* New York: Harrington Park Press.

Rodgers, Sandra. 1989. Pregnancy as Justification for Loss of Juridical Automony. In Christine Overall. (Ed.). *The Future of Human Reproduction.*Toronto: The Women's Press.

Rose, Geoffrey. 1985. Sick Individuals and Sick Populations. *International Journal of Epidemiology, 14*(1), 32-38.

Rose, Hilary 1987. Victorian Values in the Test-Tube: the Politics of Reproductive Science and Technology. In Michelle Stanworth. (Ed.). *Reproductive Technologies. Gender, Motherhood and Medicine.* Minneapolis: University of Minnesota Press.

1986. Beyond Masculinist Realities: A Feminist Epistemology for the Sciences, In Ruth Bleier. (Ed.). *Feminist Approaches to Science.* New York: Pergamon Press.

Rossiter, Amy. 1988. *From Private to Public. A Feminist Exploration of Early Mothering.* Toronto: The Women's Press.

Roth, Rachel. 1993. At Women's Expense: The Costs of Fetal Rights. In Janna C. Merrick and Robert H. Blank. (Eds.). *The Politics of Pregnancy: Policy Dilemmas in the Maternal-Fetal Relationship* New York: Harrington Park Press.

Rothman, Barbara K. 1989. *Recreating Motherhood. Ideology and Technology in a Patriarchal Society.* New York: W.W.. Norton & Company.

1988. Reproductive Technology and the Commodification of Life. In Elaine Hoffman Baruch, Amadeo F. D'Adamo & Joni Seager. (Eds.). *Embryos, Ethics and Women's Rights. Exploring the New Reproductive Technologies.* New York: Harrington Park Press.

1986. *The Tentative Pregnancy. Prenatal Diagnosis and the Future of Motherhood.* New York: Viking.

Rothschild, Joan. (Ed.). 1983. *Machina Ex Dea: Feminist Perspectives on Technology.* New York: Pergamon Press.

Rowland, Robyn. 1985. A Child at ANY Price? An overview of issues in the use of the new reporductive technologies and the threat to women. *Women's Studies International Forum,8*(6), 539-546.

Roy, David J. 1987. L'éthique du diagnostic prénatal. *Le Clinicien, mai,* 23-26.

Royal College of Physicians 1989. *Prenatal Diagnosis and Genetic Screening. Community and service implications.* London: The Royal College of Physicians of London.

Royal Commission on New Reproductive Technologies, 1993. *Proceed With Care,* vols. 1 & 2, Final Report of the Royal Commission on New Reproductive Technologies: Ottawa.

Rozen, Rima et al. 1992. Cystic Fibrosis Mutations in French Canadians: Three CFTR Mutations are Relatively Frequent in a Quebec Population with an Elevated Incidence of Cystic Fibrosis, *Am J Med Genet, 42,* 360-364.

Rozen, Rima et al. 1990. Cystic Fibrosis Mutations in North American Populations of French Ancestry: Analysis of Quebec French-Canadians and Louisiana Acadian Families, *Am J Hum Genet, 47,* 606-610.

Ruddick, Sara.. 1989. *Maternal Thinking. Towards a Politics of Peace.* New York: Ballantine Books.

Russett, Cynthia Eagle. 1989. *Sexual Science. The Victorian Construction of Womanhood.* Cambridge: Harvard University Press.

Rutten, Frans F. H. and Gouke J. Bonsel. 1992. High Cost Technology in Health Care: A Benefit or a Burden?, *Soc. Sci. Med., 35*(4), 567-577.

Salcedo, Mauricio, Chávez, Margarita, Ridaura, Cecilia, Moreno, Manuel, Lezana, José L. and Lorena Orozco. 1993. Detection of the Cystic Fibrosis Delta-F508 Mutation at Autopsy by Site-Directed Mutagenesis. *Am. J. Med Genet, 46,* 268-270.

Saleeby, C.W. 1912. *Woman and Womanhood: A Search For Principles.* London: William Heinemann.

Sarah, Rebecca. 1988. Power, Certainty and the Fear of Death. In Elaine Hoffman Baruch, Amadeo F. D'Adamo & Joni Seager. (Eds.). *Embryos, Ethics and Women's Rights. Exploring the New Reproductive Technologies.* New York: Harrington Park Press.

Saxton, Marsha. 1988. Prenatal Screening and Discriminatory Attitudes About Disability. In Elaine Hoffman Baruch, Amadeo F. D'Adamo & Joni Seager. (Eds.). *Embryos, Ethics and Women's Rights. Exploring the New Reproductive Technologies.* New York: Harrington Park Press.

1984. Born and Unborn: Implications of the Reproductive Technologies for People with Disabilities. In Rita Arditti, Renate Klein & Shelley Minden. (Eds.). *Test-Tube Women. What Future for Motherhood.* London: Pandora Press.

Schneider, William H. 1986. L'eugénisme en France: Le Tournant Des Années Trente. *Sciences Sociales et Santé, IV,* (3-4), 81-114.

Scriver, Charles and T. Mary Fujiwara. 1992. Invited Editorial: Cystic Fibrosis Genotypes and Views on Screening Are Both Heterogeneous and Population Related. *Am J Hum Genet, 51*: 943-950.

Seals, Brenda F., Edem E. Ekwo, Roger A. Williamson and James W. Hanson. 1985. Moral and Religious Influences on the Amniocentesis Decision. *Social Biology, 32*(1-2), 13-30.

Shannon, Thomas A. 1988. In Vitro Fertilization: Ethical Issues. In Elaine Hoffman Baruch, Amadeo F. D'Adamo & Joni Seager. (Eds.). *Embryos, Ethics and Women's Rights. Exploring the New Reproductive Technologies.* New York: Harrington Park Press.

Shapiro, Thomas M. 1985. *Population Control Politics. Women, Sterilization and Reproductive Choice.* Philadelphia: Temple University Press.

Shapiro, Robert. 1991. *The Human Blueprint.* Toronto: Bantam Books.

Shennib, Hani. 1994. Lung transplantation in CF patients. *Candid Facts, winter*:2-4.

Sherwin, Susan. 1989. Feminist Ethics and New Reproductive Technologies. In Christine Overall. (Ed.). *The Future of Human Reproduction.* Toronto: The Women's Press.

Shevory, Thomas C. 1993. The War at Home: Positivism, Law, and the Prosecution of Pregnant Women. In Janna C. Merrick and Robert H. Blank. (Eds.). *The Politics of Pregnancy: Policy Dilemmas in the Maternal-Fetal Relationship.* New York: Harrington Park Press.

Shilling, Chris. 1993. *The Body and Social Theory.* Newbury Park, CA: Sage Publications.

Shoshani, Tzipora et al. 1992. Association of a Nonsense Mutation (W1282X), the Most Common Mutation in the Ashkenazi Jewish Cystic Fibrosis Patients in Israil, with Presentation of Severe Disease. *Am. J. Hum. Genet, 50,* 222-228.

Shoenfeld, Miriam DiMaio et al. 1987. Screening for Fetal Down's Syndrome in Pregnancy by Measuring Maaternal Serum Alpha-Fetoprotein Levels,.*N Engl J Med, 317*(6), 342-6.

Shy, Kirkwood et al. 1990. Effects of Electronic Fetal-Heart Rate Monitoring, As Compared with Periodic Ausculatation, on the Neurologic Development of Premature Infants,.*N Engl J Med, 322*(9), 588-93.

Shumway, David R. 1989. *Michel Foucault.* Charlottesville: University Press of Virginia.

Simon-Bouy, Brigitte, Mornet, Etienne, Taillandier, Agnes, Serre, Jean-Louis, and Andre Boue. 1991. The ◊F508 mutation in mild adult forms of cystic fibrosis (CF). *Clin Genet 39,* 304-305.

Simon-Bouy, B., Mornet, E., Sere, J. L., Taillandier, A., Boue, J., and A. Boue. 1991. Nine mutations in the cystic fibrosis (CF) gene account for 80% of the CF chromosomes in French patients. *Clin Genet, 40,* 218-224.

Smith, Dorothy E. 1987. *The Everyday World As Problematic. A Feminist Sociology.* Boston: Northeastern University Press.

Smith, George P. II, 1985. Genetics, Eegenics, and Public Policy *Southern Illinois University Journal, 3,* 435-453.

Sommer, Barbara and Robert Sommer. 1997. *A Practical Guide to Behavioral Research.* (4th ed.) New York: Oxford University Press.

Spallone, Patricia. 1989. *Beyond Conception. The New Politics of Reproduction.* London: MacMillan Education.

Stanley, Liz. 1984. Open Secrets: What they are and what we should do about them. In Olivia Butler. (Ed.). *Feminist Experience in Feminist Research. Studies in Sexual Politics No. 2*, BSA sexual divisions day conference.

Stanworth, Michelle. 1987. Reproductive Technologies and the Deconstruction of Motherhood. Chap. in *Reproductive Technologies: Gender, Motherhood and Medicine*. Minneapolis: University of Minnesota Press.

Sutton, Agneta. 1990. *Prenatal Diagnosis: Confronting the Ethical Issues*. London: The Linacre Centre for the Study of the Ethics of Health Care.

Super, M. and M.J. Schwarz. 1992. Mutations of the cystic fibrosis gene locus within the population of the Northwest of England. *Eur J Pediatr, 151*(2), 108-111.

Suzuki, David and Peter Knudtson. 1988. *Genethics: The Ethics of Engineering Life*. Toronto: Stoddart.

Synapse 1989. L'Éthique et la Génétique., 7-12.

Taylor, Katherine M. and Kelner, Merrijoy. 1987. Informed consent: The Physicians Perspective. *Social Science and Medicine, 24*(2), 135-142.

Taub, Nadine, Cohen, Sherrill. (Eds.). 1988. *Reproductive Laws for the 1990s. A Briefing Handbook*. Newark, N.J.: RutgersA Project of Women's Rights Litigation clinic, School of Law-Newark Institute for Research on Women.

Taussig, Michael T. 1980. Reification and the Consciousness of the Patient. *Social Science and Medicine, 14*(B), 3-13.

Terrenoire, Gwen. 1992. Huntington's Disease and the ethics of genetic prediction. *Journal of Medical Ethics, 18*, 79-85.

Tesh, Sylvia Noble. 1988. *Hidden Arguments: Political Ideology and Disease Prevention Policy*. New Brunswick and London: Rutgers University Press.

Thanh Ha, Tu. 1993. Chronically ill face new drug costs. *The Gazette*(Montreal), 16 November, A-1.

1993. Côté backtracks on proposal to charge chronically ill outpatients for medication. *The Gazette*(Montreal), December, A-1.

Thomas, Martin. 1993. Preference for the Sex on One's Children and the Prospective Use of Sex Selection, vol. 14, *Technologies of Sex Selection and Prenatal Diagnosis*, Research Studies of the Royal Commission on New Reproductive Technologies: Ottawa.

Thousands of handicapped waiting for assistance to live at home: group. 1991. *The Gazette*(Montreal), 23 October, A-5.

Tronto, Joan C. 1988. Women and Caring: What Can Feminists Learn about Morality from Caring? In Alison M. Jaggar & Susan R. Bordo. (Eds.). *Gender/Body/Knowledge. Feminist Reconstructions of Being and Knowing.* New Brunswick, NJ: Rutgers University Press.

Turner, B. S. 1983. *Religion and Social Theory.* London: Heineman Educational Books.

Vandelac, Louise. 1986. L'enceinte de la maternité: sexes et sexualité. In Anne Marie de Vilaine, Laurence Gavarini and Michèle Le Coadic eds., *Maternité en mouvement.* Grenoble: Editions St. Martin, 220-235.

Vaux, Kenneth L. 1989. *Birth Ethics. Religious and Cultural Values in the Genesis of Life.* New York: Crossroad.

Vidaud, M., C. Ferec, O. Attree, O. Pascal, H. Guillermit, N. Ghanem, J. Martin, J. P. Moisan, J. Feingold, and M. Goossens. 1990. Frequency of the cystic fibrosis AF508 mutation in a large sample of the French population. *Hum. Genet., 85,* 434-435.

Volodkevich, Helen and Carl A. Huether. 1981. Causes of Low Utilization of Amniocentesis by Women of Advanced Maternal Age. *Social Biology, 28*(3-4), 176-186.

de Wachter, M.A.M. 1988. Recent Developments in Genetic Counselling: Ethical Viewpoints. In E.K. Hicks & J.M. Berg.(Eds.). *The Genetics of Mental Retardation. Biomedical, Psychosocial and Ethical Issues.* Boston: Kluwer Academic Publishers.

de Wachter, Maurice A. M. 1993. Ethical aspects of Human Germ-Line Gene Therapy. *Bioethics, 7*(2/3): 167-177.

Wajcman, Judy. 1991. *Feminism Confronts Technology.* Cambridge: Polity Press.

Wald, George. 1979. The Case Against Genetic Engineering. In David A. Jackson & Stephen P. Stich. (Eds.). *The Recombinant DNA Debate.* Englewood Cliffs, New Jersey: Prentice-Hall Inc.

Watson, Eila K., Mayall, Edward S., Lamb, Judith, Chapple, Jean and Robert Williamson. 1992. Psychological and social consequences of community carrier screening programme for cystic fibrosis. *The Lancet, 340,* 217-220.

Watson, Elia K., Marchant, Joanna, Bush, Andrew and Bob Williamson. 1992 Attitudes towards prenatal diagnosis and carrier screening for cystic fibrosis among the parents of patients in a paediatric cystic fibrosis clinic. *J Med Genet, 29,* 490-491.

Wauters, Jan G.et al. 1991. Frequency of the phenylalanine deletion (◊F508) in the CF gene of Belgian cystic fibrosis patients. *Clin Genet, 39,* 89-92.

Watson, James. 1968. *The Double Helix: A Personal Account of the Discovery of the Structure of DNA.* New York: Atheneum.

Weil, Dr. William B. Jr., & Benjamin, Martin. (Eds.). 1987. Ethical Issues at the Outset of Life. Volume 3. *Contemporary Issues in Fetal and Neonatal Medicine.* Boston and London: Blackwell Scientific Publications.

Weiner, Carolyn, Fagerhaugh, Shizuko, Strauss, Anselm and Barbara Suczek. 19??. Patient Power: Complex Issues Need Complex Answers. *Social Policy, 11*(2), 30-38.

Weisman, Carol S. & Teitelbaum, Martha Ann. 1985. Physician Gender and the Physician-Patient Relationship: Recent Evidence and Relevant Questions. *Social Science and Medicine*, *20*(11), 1119-1127.

Wertz,, Dorothy C., Sally R. Janes, Janet M. Rosenfield and Richard W. Erbe. no date. Attitudes Towards the Prenatal Diagnosis of Cystic Fibrosis: Factors in Decision Making.

Wertz, Dorothy C., Rosenfield, Janet M., Janes, Sally R. and Richard W. Erbe. 1991. Attitudes toward Abortion among Parents of Children with Cystic Fibrosis. *Am J Public Health, 81*:992-996.

Wells, Paul and Tu Thanh Ha. 1993. Cutbacks on free drugs will force people onto welfare, critics say. *The Gazette*(Montreal), 17 November.

Wertz, Dorothy C. and John C. Fletcher. 1993. Proposed: an international code of ethics for medical genetics. *Clin Genet*, *44*(1), 37-43.

1993. Geneticists approach ethics: an international survey. *Clin Genet*, *43*(2), 104-110.

Whitbeck, Caroline. 1988. Fetal Imagining and Fetal Monitoring: Finding the Ethical Issues. In Elain Hoffman Baruch, Amadeo F. D'Adamo & Joni Seager. (Eds.). *Embryos, Ethics andWomen's Rights. Exploring the New Reproductive Technologies.* New York: Harrington Park Press.

Whiteford, Linda M., Poland, Marilyn L. 1989. *New Approaches to Human Reproduction. Social and Ethical Dimensions.* Boulder, Colorado: Westview Press.

Wilfond, Benjamin S. and Norman Fost. 1990. The Cystic Fibrosis Gene: Medical and Social Implications for Heterzygote Detection. *JAMA, 263*(20), 2777-2783.

Wilken, U. 1988. The Necessity For Ethical Reflection In Genetic Counselling: Advice For Professionals. In E.K. Hicks & J.M. Berg.*The Genetics of Mental Retardation. Biomedical, Psychosocial and Ethical Issues.* Boston: Kluwer Academic Publishers.

Williamson, Robert. 1993. Universal community carrier screening for cystic fibrosis? *Nature Genetics, 3*: 195-201.

Williamson et al. 1989. Community attitudes to cystic fibrosis carrier testing in England: A pilot study. *Prenatal Diagnosis, 9*: 727-734.

Wills, Christopher. 1991. *Exons, Introns, and Talking Genes.* New York: BasicBooks.

Wishnia, Judith. 1991. Pacifism and Feminism in Historical Perspective, In Anne E. .Hunter. (Ed.). *Genes and Gender VI: On Peace, War and Gender. A Challenge to Genetic Explanations.* New York: Feminst Press.

Witt, M., Jaruzelska, J., Kuczora, I., Matuszakr, R., Cichy, W., and K. Borski. 1993. A Simplified method for detection of the mutations predominantly causing cystic fibrosis and phenylketonuria in Polish Families. *Clin Genet, 44*(1), 44-45.

Witt, David R., Catherine Schaefer, Patricia Sallam, Soora Wi, Bruce Blumberg, Andrea Fishback, Judy Holtzman, Shoshana Kornfeld, Robin Lee, Laurie Nemzer, and Robbin Palmer. 1996. Cystic Fibrosis Heterozygote Screening in 5, 161 Pregnant Women. *Am. J. Hum. genet., 58*, 823-835.

Workshop on Population Screening for the Cystic Fibrosis Gene. 1990. *New Eng J Med, 323*(1), 70-1.

Worton, Ron. 1993. Genome programs: Why? Symposium of the Academy of Science, The Royal Society of Canada Annual Meeting, 26-29 May.

Wright, Anne and Wayne J. Morgan. 1990. On the Creation of Problem Patients. *Social Science and Medicine, 30*(9), 951-59.

Yin, Robert K. 1984. *Case Study Research. Design and Methods.* London: Sage Publications.

"You were asking". 1991. Canadian Cystic Fibrosis Foundation. Montreal.

Yoxen, Edward. 1983. *The Gene Business. Who Should Control Biotechnology?* New York: Harper & Row.

Zeesman, Susan et al. 1984. A Private View of Heterozygosity: Eight-Year Follow-Up Study on Carriers of the Tay-Sachs Gene Detected by High School Screening in Montreal. *Am J Med Genet, 18*, 769-778.