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Genetic Bodies and Genetic Families:
Social and Material Constructions of Prenatal Genetic Testing

by

Kristen Karlberg

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I dedicate this dissertation to my son Owen Hansen Skriloff and to the hope of a sibling for him.

GENETIC BODIES AND GENETIC FAMILIES

SOCIAL AND MATERIAL CONSTRUCTIONS OF PRENATAL GENETIC TESTING

KRISTEN KARLBERG

ABSTRACT

This dissertation is a sociological examination of prenatal genetic testing (PGT) experiences of pregnant women and the genetic providers who care for them. First it analyzes the biomedicalization and geneticization of American culture as normalizing and routinizing PGT; it then compares and contrasts the perceptions and interpretations of pregnant women and genetic care providers about the practice of genetic medicine, anxiety in pregnant women, the non-directive tenet of genetic medicine, embodied knowledges of pregnant women, and the consequences for families of prenatal genetic testing. This multi-site project analyzes data from ethnographic fieldwork at 11 biomedical conferences, 7 HMO Genetics Department meetings, 3 HMO interdepartmental meetings related to PGT, 3 clinic days of amniocentesis and CVS, one week shadowing a medical geneticist, immersion in the popular culture of pregnant women during my own pregnancy, 10 in-depth interviews with perinatologists, medical geneticists, genetic counselors and research scientists, and 20 interviews with pregnant women who had PGT. I examine how the work of genetic care takes place and its influences on both pregnant women and their providers. The key findings from this project include: 1) Provider discourses of the "gray zone" of ambiguous genetic information and of genetics as omniscient are all-encompassing, permeating prenatal genetic care; 2) PGT produces "genetic bodies" and "genetic families" through the

identifying and labeling of entities as "genetic" regardless of whether anything abnormal is detected; 3) providers applied a flexible interpretation of nondirectiveness, tailoring the information provided to each woman to what they perceived were her needs; 4)pregnant women's embodied knowledges of health, age and body were challenged through the experience of PGT, mainly through the material and discursive constructions of "genetic bodies" and "genetic families"; and most importantly 5) pregnant women, with the help of genetic providers, *shape* their families through determining genetic acceptability of the fetus from PGT results and use abortion as a mechanism of prevention of births of genetically unacceptable babies. Shaping genetic families through PGT and abortion is increasingly legitimated in American culture, enabled through biomedicalization, geneticization, and the routinization and normalization of prenatal genetic testing and screening technologies as standard obstetrical care.

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1 WOMEN, FETUSES AND FAMILIES MAY BE ALTERED BY GENETIC MEDICINE THROUGH PRENATAL GENETIC TESTING

A woman decides she would like to have a child. She would like this child to be healthy, intelligent, with certain physical characteristics and a specific sex. She discusses the options with her doctor and opts for preimplantation genetic diagnosis (PGD) and in vitro fertilization (IVF). She chooses her sperm donor, the one in the catalog with most of the desired genes for characteristics she prefers. She follows the ovulation drug regimen and her eggs are harvested. Her eggs with the desired genes are chosen and fertilized with the sperm to form embryos (in vitro fertilization). The embryos are tested (preimplantation genetic diagnosis) to verify they are genetically desirable and then implanted in her uterus. Selective reduction (removal of all but one embryo from the uterus) is used to guarantee only one fetus. The baby is born and appears to have the traits she requested.

While this scenario may seem to represent an extreme pursuit of perfection, the only part of it that is not possible today is identification in egg and sperm of desirable genes for intelligence and physical characteristics. While "health" cannot be guaranteed through genetic testing, many genes for fatal, near fatal or disabling health problems are presently detectable along with less medically complicated diagnoses, benign chromosomal rearrangements and sex.

BACKGROUND

The practice of American medicine has been altered dramatically by developments ongoing in genetics research. While genetic influences have long been acknowledged in medical practice through the taking of family histories, one of the first arenas of medical practice *directly* shaped by genetic sciences was obstetrics, through new technologies of prenatal genetic testing (PGT). The availability of such technologies has been dramatically changing the experiences of pregnancy for women, their families and for medical providers as well for over two decades (Beeson 1983; Beeson 1984;

Becker and Arnold 1986; Rothman 1989; Kolker and Burke 1994; Rothenberg and Thomson 1994; Karlberg 2000). One social/cultural consequence of this permeation of genetic medicine is the assumption that a normal pregnancy should produce a "healthy" baby *if* one utilizes the technologies correctly (Rothman 1989; Nelkin and Tancredi 1994; Rothenberg and Thomson 1994; Hubbard 1995; Gottweis 1997; Hartouni 1997; Beckwith 2002).

Prenatal genetic testing (PGT) is diagnostic testing of amniotic fluid or fetal material to determine the discernable genetic components of the fetus. The information obtained can determine whether there are chromosomal abnormalities in the fetus. Testing can be done for specific genetic abnormalities if individual genes have been identified in the family, but standard PGT determines only chromosomal abnormalities. The most common types of PGT are amniocentesis (amnio) and chorionic villus sampling (CVS). Amnio involves removing amniotic fluid from the amniotic sac using a syringe guided by ultrasound. Amnio is conducted between 15 and 20 weeks gestation. Depending upon the source, there is a 1% chance of miscarriage with this procedure (CGDB 1995a:3) or a 0.5% risk (ACOG 1999:2) or "only 0.3% to 0.6%" (Blackwell, Abundis et al. 2002:1). CVS requires biopsy of the villi, which turn into the placenta, conducted either via insertion of a needle through the abdomen into the uterus or a small catheter through the cervix into the uterus. CVS is also guided by real-time ultrasound, but can be conducted as early as 9 weeks. With CVS there is a 1%-3% miscarriage rate (CGDB 1995a:4) or a risk "slightly higher than amnio" (ACOG 1999:3). Both procedures are 98%-99% accurate in diagnosing chromosomal abnormalities (CGDB 1995a:3,4) but "no test is 100% foolproof" (ACOG 1999:3). According to one meta-

analysis of three studies involving over 9000 pregnant women, CVS is associated with more sampling and technical failures, more false positive and false negative results, more pregnancy loss, and "a suggestion (though not statistically significant)" of more stillbirths and neonatal deaths (Alfirevic, Gosden et al. 2001:1). For a comprehensive history of the development of PGT technologies please refer to Appendix A.

A unique facet of PGT is that despite its designation as a type of diagnostic testing, which implies a definitive answer in the test result, "normal" amnio and CVS test results do not guarantee a "normal" baby. These tests read chromosomes and can provide with certainty only confirmation that the fetus is chromosomally normal, that is, it possesses 23 pairs of chromosomes, one of them a sex pair of either XX or XY. There can be many variations from the norm in *genes* that will not be detected by either test. Genetic variations that are specifically tested for, such as genes known to cause sickle cell disease, can be ruled out only when all of the known genes have been examined. This usually requires knowing which disease genes have already been discovered in the family, because most diseases have multiple genes that are implicated in the disease.

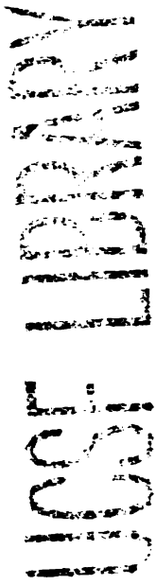
Further, there is no test yet available that tests for all disease genes known to date. At this point, it would be prohibitively time consuming and expensive. While the most common chromosomal rearrangements are coupled with diagnoses of prospective mental and physical characteristics, there is also the possibility of detecting a chromosomal rearrangement that has not yet been documented in the medical literature. In this case, there is no premise on which to base a diagnosis. Such cases usually result in a compiled diagnosis of similar chromosomal rearrangements with the caveat, "We can't know for sure." Even in a diagnosis of a relatively common genetic disease such as Down

syndrome, possessing three copies of chromosome 21 instead of the normal 2, there is no way to tell what level of mental retardation or physical disability will be associated with the disease. For PGT, there is no individual to be examined in conjunction with test results, because the test is for *fetal* abnormalities. Thus amino and CVS test results are inherently uncertain.

American culture, the focus of this dissertation, has been altered by the introduction of PGT. As early as 1974 (Pearson 1974), and officially beginning in 1983 by the American College of Obstetricians and Gynecologists and the American Academy of Pediatrics (ACOG and AAP 1983), physicians caring for pregnant women were advised to offer or refer their patients for prenatal diagnostic services. The routinization of PGT is an important feature and part of the first surge of the "geneticization" (Lippman 1991) of Western society. Lock (Lock 1997) argues that geneticization is accomplished through reducing bodies to their genes, effectively ignoring their cultural and other social elements. Geneticization refers to the ways in which genetic technologies are increasingly used to manage problems of health. It can also refer to the reductionism of genetics when applied to individuals: differences between individuals are reduced to DNA codes, with most disorders, behaviors and other variances attributed some genetic origin. This is part of what I define as the discursive production of a genetic body, one that has been diagnosed as carrying specific genes and chromosomes, be they "good" or "bad".

Of the many ways the experience of pregnancy has been affected by genetic medicine, of particular interest to me is the geneticization of bodies and families implicated through these technologies, the lived experience of pregnancy with a

genetically marked female body, family and fetus. The risks of (bio)medicalization (Zola 1994; Lupton 1997; Clarke, Shim et al. 2003) and geneticization of life in the West include the oversimplification of complex information and the inadequate handling of complicated emotional identities. Genetic information can potentially evoke major and multiple emotions in those undergoing counseling and/or testing and their families and friends, including anxiety, fear, and questions of self-worth (Marteau, Duijn et al. 1992; Rona, Beech et al. 1994; Tibben, Duivenvoorden et al. 1994; Seibert 1995; Lerman, Narold et al. 1996; Markens, Browner et al. 1999; Tercyak, Johnson et al. 2001). The fear of a parent passing on negative genetic properties is further complicated by the fact that particular patterns exist. For example, the most common forms of genetic mental retardation are X-linked, therefore affect male children more often, and the genetic propensity is contributed solely by the mother (Epstein 2002). There are also instances where the sex of the parent contributing a given chromosome influences the occurrence and transmission of chromosomal abnormalities, such as Prader-Willi syndrome (paternal) and Angelman syndrome (maternal) (Epstein 2002). The genetic "culpability" of one parent adds to the emotional strain of reproductive risk. Families play an immeasurable role in the PGT process. These domains of emotion are further complicating the already typically emotion-laden aspects of pregnancy.



Prenatal genetic testing (PGT) technologies allow for testing of the fetus, equating health with normal/'good' genes and illness with mutant/'bad' genes. A positivist discourse exists which encourages labeling individuals in terms of genes. What emerges as a result of this discourse is a perspective that suffuses genetics with conceptions of normality and deviance, particularly socially constructed identities of what I call "genetic

bodies". Rose (Rose 2001) argues that selfhood has become increasingly defined in somatic terms, encouraging the molecular vocabularies currently en vogue to (re)shape self, "actually reorganizing it a new way and according to new values about who we are, what we must do, and what we can hope for." My conception of genetic bodies includes those found to have "bad" or "good" genes, or those being tested for deviant variations (mutations) in genes or chromosomes. This definition could encompass all individuals, because all bodies have potential genetic information discoverable through genetic testing of that person. But there is another unique consequence of the permeation of genetics into maternal-fetal medicine--a pregnant woman's body is potentially *two* genetic bodies simultaneously: hers and the fetus's.

Genetic bodies are situated culturally, historically and textually in American society, typically gendered, raced and classed (Collins 1999). Genetic bodies are sociocultural phenomena that must be managed-a kind of "technoscientific identity" (Clarke, Shim et al. 2003) that requires negotiation by those so identified.

A challenging site for genetic bodies is their intersection with emotions in the situation of PGT. In any pregnancy there is a potential heightened emotional state for the pregnant woman, her partner, family and genetic care providers. What is unique about emotionality in PGT is the potentially greater incitement of guilt/excitement/trepidation in both biological parents in respect to their individual contributions of genetic material. Anxiety and depression are some of the emotions attributed to prospective parents having "bad" genes in their genetic bodies (Kenen and Schmidt 1978; Beeson and Golbus 1979; Beeson and Golbus 1985; Marteau, Duijn et al. 1992; Mennie, Compton et al. 1993; Hill 1994; Jorgensen 1995; Duster 1999; Beeson and Duster 2002). Rose's (2001)

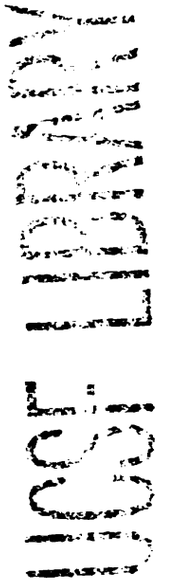
'ethopolitics' lends itself well here to the ways emotions in biological parents are complicated and conflicted in relation to the urge to judge themselves. Parents are influenced by the "knowledges and beliefs about ones biological and genetic complement [that] become integrated into the complex choices that *prudent* individuals are *obliged* to make in their life strategies, biological identity generates biological responsibility" (Rose 2001:17 emphasis added). Foucauldian "technologies of the self" (Foucault 1988)—ways the self polices itself in society which often seem to be natural but in reality are reflections of power --abound in pregnancy.

The market for prenatal genetic testing is infinite, as there is no way other than pregnancy to produce children as of yet. The link between social pressures toward the biomedicalization and geneticization of pregnancy and the ongoing creation of the market for genetic testing dramatically enhances the social power of genetic care providers. Rose's (2001) argument about the "politics of life itself" is relevant here, for he says that such politics are shaped by the funding entities who provide the technologies that *define* what is genetic and molecular through choosing to study a particular family lineage or marker and to fund particular biomedical research and development. Life at the molecular level is only knowable through such technologies, and those who market these technologies are therefore defining "normal."

A telling emotional facet of PGT is kinship. When dealing with genetics, the links between families and individuals are magnified. The uniqueness of genetic diagnostic tools is that if one individual in the family possesses a genetic trait, it is extremely likely that someone related to them also has that trait. Thus, the genetic body is not one's own when viewed through the lens of kinship and familial heredity. Hence

my conception of genetic families: those whose members or a member has experienced prenatal genetic testing or some other type of genetic testing which explicitly identifies "genetic" information about the family. A genetic family does not have to have a genetic anomaly to be "marked," merely it must have been examined through the lens of genetics. Once this genetic information is "in the family," the genetic family will forever be marked by that genetic knowledge, be it benign or life-altering.

PURPOSE OF MY RESEARCH



Biomedicine, through the infusion of genetics into pregnancies, is actively creating and maintaining emotional genetic bodies and, in conjunction, genetic families. Broadly speaking, American culture allows for the shaping of genetic families through PGT, encouraging the quest for knowledge and legitimating abortion when the knowledge is not "good." But also in some sectors, there exists intense social condemnation of the option of abortion for any means, heightening the emotional experience of PGT for most who have it.

Constructions of pregnant women's bodies, their fetuses, and their families are discursive through the knowledge bases of genetics, obstetrics, perinatology, and other related medical practices as well as through socio-cultural realms. Such constructions are material through labeling of pregnancies as "high-risk" or otherwise at risk, designating pregnant women's bodies as requiring PGT, labeling fetuses as non-viable or possessing a genetic mutation, identifying families as "high-risk" for genetic disorders, possibly stigmatizing women who have children with genetic disorders, and not providing adequate resources for those with genetic disabilities.

My project here is to critically examine the meanings and consequences of PGT in the lives of pregnant women, their families, and their providers. I hope to provide a detailed examination of how these material and discursive constructions of pregnant women's bodies, genetic selves, and those of their fetuses and families are created and negotiated within the maternal-fetal medical realm and also in the lives of pregnant women and their families.

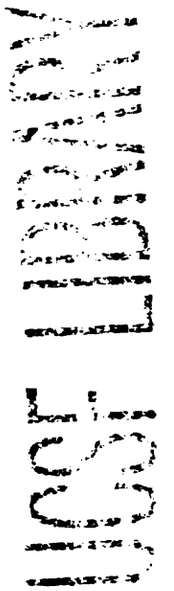
RESEARCH QUESTIONS

The existence of PGT technologies as part of routine prenatal care of American women today provides a unique situation for the study of the intersections of genetic technologies, identities, and subjectivities. My dissertation research focused on these intersections and addressed the following questions:

(1) Have genetic technologies and related scientific knowledges altered (are they altering) understandings of what "normal" pregnancies entail, what "normal" female and fetal bodies are, and what "normal" emotional responses should be to personal genetic information? If they have, how?

(2) Are PGT technologies transforming the lived experiences of selves, subjectivities, genders, bodies, and/or emotions? How?

(3) More specifically, how do women who have experienced PGT technologies perceive themselves, their bodies, and their children in relation to the new genetic knowledge available to them?



(4) What do the embodied experiences and knowledges of these women who have had PGT tell us about the ways such genetic information changes identities and subjectivities?

(5) What do current trajectories of the use of PGT technologies reveal about changing cultural definitions of healthy and ill?

(6) What affects (if any) do these possible alterations in pregnancy and health experiences and definitions have on the lived realities and definitions of families?

OVERVIEW OF CHAPTERS

Chapter 2 discusses literatures useful to my understandings of PGT. Section I reviews the sociological and anthropological theoretical literatures I used to inform my data analysis. Section II explores the scientific production and social construction of PGT. Section III discusses women and prenatal genetic testing literatures from both medical and social science backgrounds. Section IV reviews literature available about providers.

Chapter 3 explains why I chose to study in the area of prenatal genetic testing. I tell my story of genetic identities and reconstructions, including my personal experiences of prenatal genetic testing, to "out" myself as not only a social science researcher interested in PGT, but also a woman with a genetic body, genetic family, and one who has utilized PGT.

Chapter 4 is an explanation of the methods, data sources, modes of data collection and data analysis that I employed while conducting this research.

Chapter 5 begins the data analysis by discussing biomedicalization and geneticization. I begin by defining the types of genetic care providers included in the data. I move into the discourses used by these providers when conducting genetic medicine, and explore their understandings of geneticization and the media. I then shift to pregnant women's experiences of biomedicalization and geneticization of their pregnancies. This chapter concludes with the nostalgia both providers and pregnant women expressed when talking about their understandings of PGT, and "the way things used to be" before pregnancy became so complicated.

Chapter 6 offers a back-and-forth discussion between providers and pregnant women contrasting their perceptions of prenatal genetic care. I compare women's and providers' perceptions of the practice of genetic medicine, the impact of the creation of medical pedigrees in the genetic counseling session, anxiety about PGT, and non-directiveness as the mode of practice for genetic care.

Chapter 7 explores the embodied knowledges pregnant women possess. The chapter begins with pregnant women's initial exposures to PGT and how they navigate genetic medicine protocols to receive PGT. How pregnant bodies are constructed as genetic bodies is examined. Then I discuss why women have testing and how they describe coping with the experience of making the decision to have PGT and then actually having it. The chapter concludes with providers' understandings of pregnant women having PGT.

Chapter 8 explains how pregnant women and providers together accomplish the goal of shaping families through PGT. Through both providers' and women's explanations of why PGT is utilized, abortion is discussed as the method of shaping a

genetic family to attain the kind of family desired by the pregnant woman and her partner. The ways pregnant women describe the processes of making the decision to have PGT in order to avoid the births of babies with particular genetic disorders is compared to what providers say about why they practice genetic care and provide PGT.

Chapter 9 weaves together the findings from chapters 5-8 and synthesizes my conclusions from the data. I assert that pregnant women and genetic care providers work together to enable pregnant women to exercise the options they presently have to avoid having babies with certain specific genetic conditions. The individualized choices women make about PGT are intimately linked to their willingness or lack thereof to have abortions in the event of an undesirable fetus being detected.

2 THEORETICAL ENGAGEMENTS AND MEDICAL LITERATURE

My research is framed primarily through the theoretical perspectives of feminism, science and technology studies, and symbolic interactionism/social constructionism. In this chapter I briefly discuss the theoretical frameworks central to this dissertation: biomedicalization; situated knowledges; theories of bodies and embodiment; theories of emotions and bodies; and theories of families and kinship. The examination is supported through a more in-depth analysis of social science literatures on geneticization and stigma.

I then move on to literature discussing prenatal genetic testing, demonstrating how utilization of PGT has increased with the biomedicalization and geneticization of American society. I examine abortion literature relating to prenatal genetic testing to frame the options available to pregnant women who have adverse diagnoses from PGT. I then outline the biomedical routinization of such testing including issues of transferring responsibility for abnormal genetic children from the biomedical industrial complex to their mothers. A summary of publications related to the emotions and familial issues of women who have prenatal genetic testing lends itself to the discussion of how women deal with this enormous responsibility to shape their genetic families. I conclude with a brief discussion of the literature on providers of genetic care, examining their constructions of non-directive approaches to care and how they indirectly and sometimes directly influence women in the prenatal genetic testing arena.

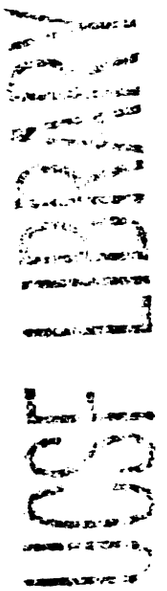
SECTION I: SOCIOLOGICAL/ANTHRPOLOGICAL THEORIES

Social Constructionism and Biomedicalization

Symbolic interactionism addresses how relationships lead to production of knowledges that in turn lead to new relationships (Blumer 1969). In medical sociology, this approach conceives the patient as an active agent in personal constructions of illness, as well as diagnosis and treatment (Strauss, Fagerhaugh et al. 1982; Strauss, Fagerhaugh et al. 1982; Strauss and Corbin 1988). The construction of knowledge is an important theme here. The social construction of biomedical knowledges and practices asserts that the meanings of illnesses and health are created based on material, cultural, social, experiential and other knowledges explicit and implicit to the biomedical experience. The cultural constructions of illness include the different beliefs, traditions and worldviews of the individuals involved and how these play an integral role in shaping ideas, constructs and experiences of illness (e.g. Charmaz 1983; Olesen, Schatzman et al. 1990). Illness is both individual and shared.

Within the sociology of health and illness, social constructionism has also contributed greatly to the concept of medicalization (Freidson 1970). Zola (Zola 1994:404) defines medicalization as, "largely an insidious and often undramatic phenomenon accomplished by "medicalizing" much of daily living, by making medicine and the labels "healthy" and "ill" relevant to an ever increasing part of human existence." Medicalization changes people's perspectives on their bodies, and on how they experience the world (Englehardt 1986). It can restructure individual's realities by intruding on the taken-for-grantedness of everyday life regarding the body.

In contrast, following Foucault, Lupton (Lupton 1997) emphasizes that medicalization is a product of the ways that society is structured, including the influential social role played by members of the medical profession, a powerful and high-status occupation group. Clarke and colleagues (Clarke, Shim et al. 2003:161) elaborate medicalization, articulating "biomedicalization" as the "increasingly complex, multisided, multidirectional process of medicalization, both extended and reconstituted through the new social forms of highly technoscientific biomedicine." This is obviously the more appropriate term for the kind of medicalization produced through the routinization of PGT technologies.



Technoscience Studies and Situated Knowledges

Technoscience studies add further dimensions to the explication of biomedicine in today's society. While this interdisciplinary domain has various titles and specificities,[1] I prefer technoscience studies because it encompasses the study of science and the study of technologies, both of which are essential in the study of genetics. Technoscience studies began with the purpose of exploring "the way in which social interests, values, history, actions, institutions, networks and so on, shape, influence, structure, cause, explain, inform, characterize, or co-constitute the content of science and technology" (Hess 1997:82).

Following Latour (Latour 1990) and Casper and Koenig (Casper and Koenig 1996) and others, I use "technoscience studies" to explicitly capture the merging of the terms "technology" and "science" which in the past were kept separate. Clarke cites

[1] Technoscience studies has many names, among them "science and technology studies" (STS, S&TS), "sociology of scientific knowledge," "science, technology and society studies," "science, technology and medicine studies" (ST&MS), and "science studies." Technoscience studies both draws upon and dwells in sociology, philosophy, history, anthropology, engineering, legal studies, feminist studies, critical studies, and cultural studies (Jasanoff, Markle et al. 1995; Hess 1997).

Pickstone who complicated what "technoscience" means: "the term has a 'specific historical meaning for fields where knowledge and practice and the economy were intimately related, where knowledge was saleable,' where science involved 'the creation and sale of knowledge products'" (Clarke 1998:13). Conceptually and analytically, the terms "science" and "technology" are co-constituted in my work.

Technoscience studies foundations lie in the belief that science is social, rather than purely rational. My study of prenatal genetic testing technologies and the emotional and corporeal issues requires the exploration of the uses and cultural and social meanings attributed to these medical technologies that cut across social, cultural and corporeal boundaries. Technoscience studies is uniquely equipped with the "right tools for the job" (Clarke and Fujimura 1992) for understanding PGT, especially through its interdisciplinary slant and the view that science, technology and medicine are cultural phenomena (Franklin 1995). Basic assumptions of interpretive technoscience studies include the locality of knowledges, scientific "facts" as social constructions, science and society as co-constituted, and skepticism regarding the taken-for-granted nature of meanings (Hess 1997). The co-constituitive nature of science and society is particularly important to my interpretation where content and context are viewed as mutually shaping each other, with causal links as multi-directional (Latour 1987; Woolgar 1991; Clarke and Fujimura 1992; Bijker 1995).

Establishing the priority, relativity and situatedness of knowledges and allowing for diversity of such knowledges are foundational to my approach. Constructions of knowledge come from multiple actors and voices, "generated through collective interaction over time, in communities of discourse and practice" (Clarke and Montini

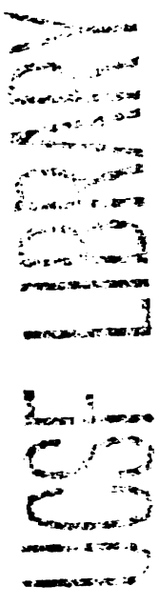
1993:69). Many feminist epistemologies recognize the irreducible multiplicity of beliefs and cultural frameworks grounded in particular historical and sociopolitical locations. Situated knowledges (Haraway 1991) are produced from the standpoints of groups or individuals. The prenatal genetic testing examined in this study includes the pregnant women, their partners and families, the medical settings through which they navigate, the genetic counselors, medical geneticists, perinatologists, obstetricians, radiologists and/or laboratory staff, to name a few.

Haraway's insistence on knowledges as embodied ensures that the partiality, locality and circumstances of knowledge claims are examined. Through such situated knowledges, partiality instead of universality becomes the acknowledged basis for knowledge-making claims (Haraway 1991:195). This is particularly relevant when addressing genetic issues, as genes are components of chromosomes, which are components of individuals, but which do not define who one is or how "healthy" or "ill" one is or will become. That is, PGT diagnoses are particular bits of genetic information about the fetus and therefore the parents' genetic make-up, but do not determine what kind of individual that fetus will become, nor do they fully indicate the health or well-being of the fetus or the parents.

How the situated knowledges of genetic care providers become THE knowledge—THE genetic information related to particular individuals in the situation of PGT--is a major focus of this study. But more particularly, the ways such knowledges are then (re)negotiated by pregnant women and their families, and reinterpreted to become their own knowledges, with very different meanings and actions associated, are also of primary interest. These recycled and revised knowledges allow us to understand

how genetic bodies and genetic families are co-constituted through interactions among genetics professionals, the women who utilize genetic testing technologies, others with whom they interact, and pertinent discourses available in the broader culture.

In a similar fashion, Novas and Rose (Novas and Rose 2000) identify the changes of an individual who "becomes" genetically at risk as part of a larger somatic transformation and "mutation" in conceptions of life itself. The authors also suggest that life itself is now "imagined, investigated, explained and intervened upon at a molecular level, what they call the new "molecular optics" (Novas and Rose 2000:487). Within the molecular optics, Novas and Rose propose that there is a wider mutation in personhood, a "somatic individuality" which forges new and direct relations between body and self. They argue that "genetic ideas of personhood are already beginning to infuse these languages of somatic individualization, inscribing an indelible genetic truth into the heart of corporeal existence" (Novas and Rose 2000:489), making sense of individuality through reorganizing according to new values about who we are, what we must do and what we can hope for including decisions on how to conduct one's life, have children, get married or pursue a career. In conjunction with Haraway's situated knowledges, Novas and Rose (2000:506) state: "Knowledge comes to be regarded as residing in multiple sites, which are to be actively sought and assimiliated for purposes of the care of the self and the care of others. Somatic individuals, in this case those genetically at risk, engage with this knowledge as interested and avid consumers."



Bodies and Embodiment

Foucauldian (Foucault 1975) bodies are normalized, surveyed and disciplined through relations of power. The body is always a social representation, and particular

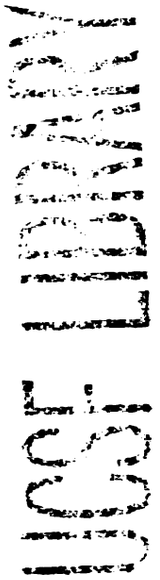
representations can transform bodies. Normative bodies are "healthy" in more delineated ways because and as a reflection of technologies of seeing (e.g. the map of the Human Genome, prenatal genetic testing). The genetic body is created through discourse and biomedical sciences, with the body as a representation of the "human genome." When one's genotype does not match the cartography of the "human genome," the individual embodies the "genetic body" with "abnormalities" based on the diagnosis of genetic mutation/deletion/translocation.

Following Rose's (Rose 2001) premise that selfhood has become inherently somatic, his argument that the new genomic and molecular "vocabularies" of ourselves are becoming "self" is useful in interpreting these transformations of bodies into genetic bodies through PGT. Rose (2001) believes that individuals are increasingly being defined by others, and by ourselves, in terms of both the possibilities and limitations of our corporeality. He suggests that "dilemmas about what we are, what we are capable of, and what we may hope for, now have a molecular form," and as such human existence is now molecularly biopolitical (2001:16). The molecular optics Novas and Rose (2000) discuss influence the ontology of human beings by shaping which family lineages are investigated, which markers are chosen, and which genes are researched by which methods.

Body theories help explain the social and discursive creation of an embodied, emotional, labeled body-an individual's genetic body. The genetic body is challenged through the "embodied character of social processes and with individual agency expressed through the body" (Lock 1997). Genetic bodies are more than mediators between subjectivity (embodiment) and representation. Instead, I believe that individuals

whose genetic bodies have been labeled experience a trans/re/formation of self and must grapple with identity issues that awaken emotions not addressed by those dwelling in unlabeled, not (yet) geneticized bodies living in the "normal" world.

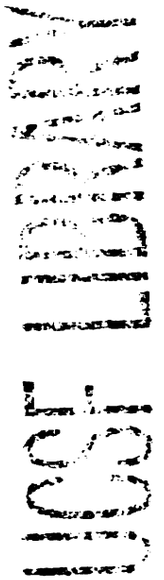
When discussing embodiment, consideration of the changing nature of the experience is essential. Bodily knowledge represents a lifetime of self-understanding, including the past and our culture as embodied (e.g. Becker 1990; Becker 1997; Becker 2000). When new information or a new experience of the body occurs which is incommensurate with previous notions of embodiment, an altering of the self takes place. The physical self, the tangible human body, is "one of many selves, which evolves and is transformed not only in interaction with others, but in the processes between self as knower and the body which draw upon subjective and cultural resources" (Olesen, Schatzman et al. 1990:451). The evolution of the physical self and transformations of meanings are facilitated through the self-reflexive viewing of body states and condition. Embodiment may be a critical link to self-reflexivity for those who have the life disruption of startling new genetic information from their prenatal genetic testing experiences.



Gender is yet another consideration one must address in body theory. Women's bodies are inherently multiply defined because of their common capability to produce other bodies through their own. Geneticization and the current dominating constructions of flawed genomes and bodies are predominantly represented as dwelling within the *female* genetic body. This, I suggest, is a result of the histories of the body as gendered. In short, Gatens (1992) and others, including Grosz (Grosz 1994), Balsamo (Balsamo 1996) and Butler (Butler 1993), through providing alternative conceptual frameworks,

argue that sex differences are not located (only) in the biology of the body, but are created through gendered discourses, experiences and performances. One of these gendered experiences is pregnancy accompanied by prenatal genetic testing.

Grosz's (1994) interpretation of the materiality of the body is of great importance to my analysis of genetic bodies. Her "corporeal feminism" conceives the body as *simultaneously* volatile and shifting, durable, corporeal, and delimited. She proposes that the specificities of the materials being inscribed (the body) have an effect on the kind of text produced (the embodied body) through lived experience and self-reflexivity. Thus the beginnings of the kind of body being shaped by society and culture are not more important than the shaping, but they do impact it.



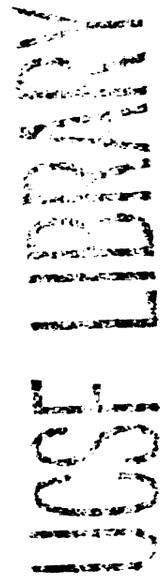
This supports my argument about genetic entities being created through the technologies and experiences of prenatal testing. That is, the prenatal genetic testing experience is such that women are given new genetic information about the self that is usually incommensurate with previous notions of embodiment, which leads to altering of the notions of self. The whole individual is linked together by these facets of self (Olesen, Schatzman et al. 1990), and embodiment may be a critical link to self-reflexivity for those who have the disruption of new genetic information.

Theories of Embodied Emotions

My interest in genetic bodies cannot be comprehensively articulated without attention to emotions. Emotion is our experience of the body, and embodied emotion is individually articulated. The interactionist stance in the sociology of emotions echoes my understanding of the embodied emotions implicated in prenatal genetic testing. Interactionist emotions theory incorporates the social, cultural, physiological and

biological facets of emotionality and effectively embraces the embodied nature of such emotions. Individual experience creates emotion and is created by the social interpersonal emotion work present in most social interactions, most specifically for my argument, in the family.

It is through bodies that people act and feel, according to Lyon (Lyon 1996), and emotionality is a mode of being, a way for people to experience selfhood as relational to society, in Freund's (Freund 1990) estimation. These two theoretical constructions link identity and selfhood to emotional experience and capability, suggesting an array of intriguing conceptual and theoretical issues. An exploration of the complex relationships among bodies and embodiment, emotions and health/illness in women diagnosed with genetic bodies raises fundamental questions about constructions and experiences of bodies, the relationships of those to emotions and identity.



Bodies are the locations where prenatal testing technologies are experienced. Following Williams and Bendelow (Williams and Bendelow 1996), I suggest that the embodied nature of emotions may be a "missing link" for explicating the ties between health and the social body, as emotionality is the experience of emotions throughout the body representing ongoing lived experience and culturally sensitive self-reflexivity. In Olesen and Bone's (Olesen and Bone 1998) discussion of emotion labor in nursing under the new managed care system, they support the contention that health care provision is influenced by the emotions of both the patient/"client" and the health care provider. The embodied nature of emotions allows for the inclusion of considerations of gender, race and class because of the ways bodies are constituted by these inscriptions.

Martin (Martin 2003) reveals that white, middle-class, heterosexual women attempt to control the emotions of childbirth because of an "internalized sense of gender" which disciplines these women and their bodies during childbirth to avoid screaming and to try to stay "nice". Emotions as embodied are pivotal in my argument about the ways that emotions change the experience of prenatal genetic testing for pregnant women and also to some extent, the experiences of their partners, families, and providers.

Theories of Family/Kinship

In American society until recently, family and kinship constructs were based on consanguinity, being related by blood through procreation (Schneider 1980), but this is not necessarily the case in other parts of the world. Schneider (1980) argues persuasively that kinship is a Western construct. Another concept peculiar to the Western world is bilateral descent, where relatively equal weight is given to the mother's and the father's descent lines, with identity derived from each parent (Finkler 2001). Family and kinship are culturally bound together by responsibility, love and identity (Schneider, 1980). Loosening of kinship ties has been perceived as an indicator of a complex society, most common in urban areas, but notable everywhere (Finkler, 2001).

Today's family is not easily defined. Today, affect and choice determine who precisely is defined kin, bound through blood as well as other linkages such as friendship (Finkler, 2001). Stacy (Stacey 1990:17) notes that, "no longer is there a single culturally dominant family pattern to which the majority of Americans conform and most of the rest aspire." This is due to divorce, sexual partner choices, and other "non-traditional" methods of forming families. I define "family" as a social reality constructed through shared meanings and experiences. Through shared discourse, family members shape

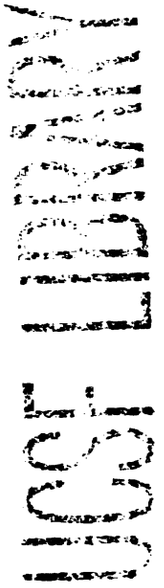
their individual identities by creating a personal narrative that must be compatible with a grand narrative of the family, at least in terms of certain constants, such as who the family members are and how they are related. Altering that reality may instigate a (re)adjusting of the family definition. The personal and family narratives, cultures and concepts of a family are mutually constituted through ongoing (re)construction which is dramatically illustrated through an alteration of the shared reality of who family is, such as when a genetic mutation is discovered or a child with a genetic disease is born.

The family can be seen as the locus of primary socialization. The family we are part of is also central to our individual identity (Stone 1988). Social reality and self-identity may be primarily defined in interaction with the social group of the family. As Stone (Stone, 1988:7) asserts, "the family's survival depends on the shared sensibility of its members." Although most families have spoken stories, the family does not have to articulate its culture; it may use a variety of unspoken communication modes to convey meanings. Communication of social realities within the family provides a collective understanding within which personal identities are negotiated. No matter how the family is construed, it still carries loyalty, love and responsibility as primary cues to its identity.

Strathern argues that kinship is the connection between the domain of society and the domain of procreation, and thus is the cultural construction of family (Strathern 1992). Becker (Becker 1990) believes it is an American value to "be a close-knit family" and that when people prepare to reproduce, they want to perpetuate the bond between their nuclear families and the next generation. When we partner we bring our families with us (Becker, 1990), so family is indelibly linked to the children we produce. Biology

grounds some cultural notions of kinship (Edwards, Franklin et al. 1993), and as such is inescapable.

In U. S. culture today, biomedicalization and geneticization have had a distinct impact on the definition of family. The biomedical understanding of disease etiology calls for close scrutiny of family and kinship (Finkler, 2001). The traditional understanding of "parent" now has been generally subcategorized into "biological-" and "social-", with the "genetic" parent as a facet of the biological parent. Nelkin and Lindee (Nelkin and Lindee 1995) define a new family that is gradually replacing the nuclear family of mother, father and children related by blood. They call this the "molecular family," defined as "one bound together less by history, tradition, or common experience than by shared DNA" (Nelkin and Lindee, 1995:58). The emphasis of the new molecular family is strictly on genetic ties between parent and child, disregarding emotional connections and social bonds. I build on Nelkin and Lindee's concept to explain the generation of a family through prenatal genetic testing and screening. This new molecular family, that I call a "genetic family," is one fashioned by genetic technologies to produce the desired kind of family, be it one with one boy and one girl, or one free of genetic disease, or one with a genetic condition, such as achondroplasia or deafness.



SECTION II: THE SCIENTIFIC PRODUCTION AND SOCIAL CONSTRUCTION OF PRENATAL GENETIC TESTING

Amniocentesis was initially developed to determine fetal sex. Later it was used to detect Down syndrome, further evolving more recently to test for additional known, mapped genetic disorders. CVS and other genetic testing and screening procedures followed soon after. For a comprehensive explanation of the specifics of these tests,

please refer to Appendix A. In brief, technologies were developed that allowed all *known* chromosomal abnormalities to be detected, and later registered gene abnormalities could also be diagnosed. The purposes of this kind of testing vary. When examined through the sociological lens, this testing is a socially constructed good for many pregnant women, helpful to some who want to prevent births of disabled babies or to know before birth of congenital problems. But it is also complex and confusing for some, with new responsibilities for decision-making about issues once completely beyond human control. This type of testing calls into question what being a good parent means and how family is defined.

Next I address the ways I believe prenatal genetic testing has been socially constructed and scientifically produced in American culture. I begin with a discussion of geneticization, linking that with stigmatization of genetic bodies and families. I move on to an examination of the role the media play in aiding geneticization and biomedicalization through research publications about the relationship of the media with statistics, public understandings of genetics and genetic public interest stories.

Geneticization

Biomedicalization theory has argued that new "technoscientific identities," produced through technoscientific means (e.g. DNA testing, genetic testing) are today more commonly produced in the West (Clarke, Shim et al. 2003). My argument in this dissertation extends this point: through biomedicalization, the individual technoscientific identity of a genetic body and the *collective technoscientific identity of a genetic family* are produced through the technologies of prenatal genetic testing. These individual *and familial* transformations are enabled through the availability of testing to determine who's

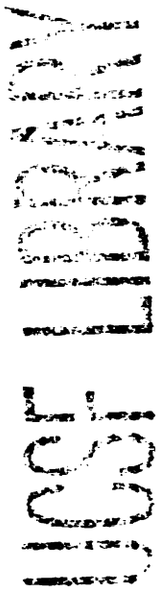
who in a "family"--blood members and not-- and who does and does not have genetic ties and/or genetic abnormalities. Such biomedicalization is constituted in part through geneticization.

Geneticization (Lippman 1991:19) is the:

> 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
>
> .

In this medically prescribed rhetoric, genes are framed as the principle factors influencing health, improving the way humans reproduce through enhanced technologies to diagnose, treat, and predict disease. While this is disturbing to those who understand how ambiguous genetic information truly is, such thinking also has great influence over how disease is defined, viewed, and managed. A disease has not changed in manifestation or symptoms once it has been labeled "genetic," it merely has been reconstructed and often "reduced" as such by biomedicine. What I am arguing is that the label "genetic" effectively creates a different disease from the non-genetic one it was before the designation. The label "genetic" applied to a disease can distract from other important factors involved in the individual's lifestyle and other instances that contribute to that person having a particular "genetic disease." The more genes identified, the more genetic disease, the more likely one is at risk for something. Genetic predictability and accountability successfully transfers health responsibility to the individual, effectively erasing social responsibility to eradicate factors that contribute to health, likely including genetic health, such as poverty, racism and gender discrimination.

Lock supports Lippman's assertions in her treatise on bodies. She believes geneticization is accomplished through normalizing what bodies are. "The body of matter-the body that matters-must...be interpreted as something more than a site of mediation between subjectivity and representation. The new body of surfaces grapples with the embodied character of social process, and with individual agency expressed through the body" (Lock, 1997:269). While some have criticized the overarching net of Lippman's "geneticization" as too broad, negative, colonial, philosophical and ethically loaded (see Hedgecoe, 2001 for a review of these discussions), there is no doubt that her concept has enabled ongoing critique of the ways in which medical genetics has pervasively altered social perceptions of health and illness. Hedgecoe (Hedgecoe 2001:876) argues that a molecular interpretation of geneticization is more fluidly integrated into arguments in the social studies of science around genetics: "in medicine, geneticization takes place when a condition is linked to a specific stretch of DNA." However it is defined, geneticization is changing the way society and individuals view themselves, facilitated in part by the information age. Stigmatization of individuals based on genetic information is one result of geneticization. I also think the media play an extrememly influential role in the geneticization of the West, particularly in America.



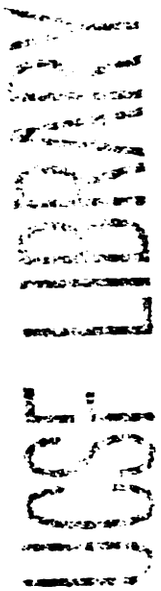
Stigma

Erving Goffman (Goffman 1963:3) in his classic work on the topic, defined stigma as an "attribute that is deeply discrediting" and that reduces the bearer "from a whole and ususal person to a tainted, discounted one." There has been considerable

research on the potential stigmatization of genetic information.[2] However, when addressing genetic stigmatization I believe the process is more complex. I argue that one of the consequences of (bio)medicalization (Conrad and Schneider 1980; Zola 1994; Clarke, Shim et al. 2003) of American culture is that *genetic status* has become a stigmatizable entity, establishing a "virtual social identity" (Goffman 1963), a "technoscientific identity" (Clarke, Shim et al. 2003) vis-à-vis genetics, a "somatic identity" (Rose 2000) that can also be the basis for "biosociality" (Rabinow 1996).

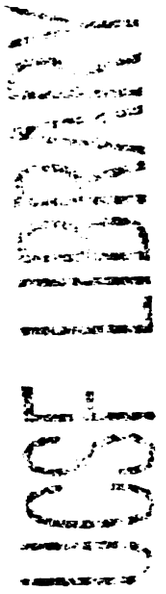
These new identities are consequential for stigmatizing processes. Individuals devoid of physical manifestations, but who possess mapped mutations can now be distinguished, thus possibly stigmatizing those individuals who are *established carriers* of genetic mutations. Link and Phelan's (Link and Phelan 2001:367) conception of stigma is useful here: "...stigma exists when elements of labeling, stereotyping, separation, status loss, and discrimination occur together in a power situation that allows them." These authors further emphasize that stigma exists as a matter of degree. Genetic stigmatization is unique in that it is dependent upon public knowledge of inheritance patterns, the "known-about-ness". Thus the dissemination of genetic information through the media is an essential contribution to the "geneticization" of society and the potential stigmatization of carrier individuals.

Turner reads Goffman's sociology of self in everyday life as: "the performance of the self through the medium of the socially interpreted body" (Turner 1996:68). This socially interpreted body is itself subject to Rose's (2001:17) "somatic individuality"



[2] Please see: (Wexler 1979; Billings, Kohn et al. 1992; Nelkin 1992; Parsons and Atkinson 1992; Draper 1993; Mennie, Compton et al. 1993; Wexler 1995; Lerman, Narold et al. 1996; Mitchell, Capua et al. 1996; Condit and Williams 1997; Hoedemaekers, Have et al. 1997; McConkie-Rosell and DeVellis 2000; Geller, Alper et al. 2002)

which renders the novel, unique molecular aspects of self visible and open to interpretation in relation to "new values about who we are, what we must do and what we can hope for." Stigmatization based on genetic *responsibility* is one possibility, in that if one has knowledge about potential offspring being potentially genetically "abnormal" the implication is that one should prevent that life from existing, suffering and affecting the public good through draining tax and other dollars. This is reminiscent of eugenic standards. For a brief discussion of the history of genetics and its multifaceted relationship with eugenics, please see Appendix B.



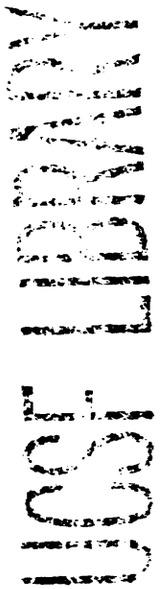
Genetics in the media; the media in genetics

Lupton (Lupton 1994:17) argues that "the study of the ways in which medical practices and institutions are represented in the mass media and the reception of such representations by audiences is integral to interpretive scholarship attempting to understand the socio-cultural aspects of medicine and health-related knowledges and practices."

> The worlds of medicine and media exist in a unique symbiosis. The media not only cannibalize the medical world for their news stories and fictional plotlines, but health care institutions, medical researchers, and academic physicians themselves manipulate the media, inviting the spotlight to shine on their successes and deflecting the glare away from the less savory aspects of their work. The delicate balance between using the media as an outlet for legitimate stories and exploiting it for particular agendas raises a host of ethical issues (Friedman and Jones 2003).

This quote was part of a 2004 conference announcement for the American Society for Bioethics and Humanities (ASBH). While the conference was not focused specifically on the media and genetics, concern with the influence of the media is particularly relevant to this section.

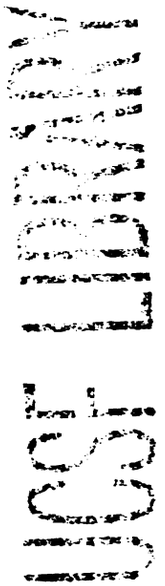
Obviously, the ASBH has a different perspective on the interactions between media and medicine than a medical organization would. In September 2002, the Mayo Clinic, one of the preeminent hospital consortiums in the world, held the "Mayo Clinic National Conference on Medicine and the Media." This conference was convened to study "the accurate, timely and responsible reporting of medical news to the public" and discovered, among other things, that "medical news reports may be confusing because the underlying scientific issues are unresolved and open to multiple interpretations" (Lantz and Lanier 2002). Most people, *including physicians and scientists*, first learn about the newest developments in medicine through news reports, according to articles published in medical peer-reviewed journals and presented at professional genetics conferences (Caulfield and Bubela 2002; Voss 2002; Stamm, Williams et al. 2003).



Here, I briefly discuss the interactions of the media with genetic science and genetic discoveries, first addressing current work on the media and genetics. I then spend some time on the role of public interest stories and the media. Any discussion of the media and genetics would be glaringly incomplete without addressing Nelkin and Lindee's 1995 treatise *The DNA Mystique: the Gene as a Cultural Icon* (Nelkin and Lindee 1995). Genetic essentialism, the reduction of humans to their genetic components, is the focus of their examination of cultural, social and media interactions with genes and DNA. The authors explore how genes have been given power (through science, the media, and popular culture) to alter the scope of individual identity. In their discourse analysis, they assert that "the narratives of mass culture give shape to what is seen in the world" (Nelkin and Lindee1995:198). Nelkin and Lindee's research spanned examining newspapers and periodicals , interviewing specialists, searching through

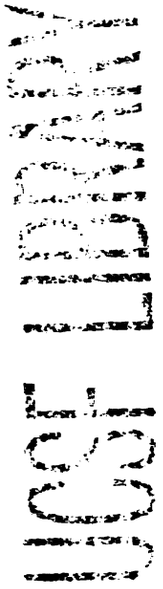
publications of special interest groups, and attending discussions and debates around topics related to DNA, genes and the Human Genome Project.

Nelkin and Lindee (1995) successfully argue that the status of the gene in society is not only a product of the prestige of science, but also of its position as a "cultural icon" which mirrors public expectations, social tensions and political agendas. This, they argue, is the appeal of genetic essentialism, which lies behind the success of the gene in our American society: it reflects traditional American values and social agendas. The gene promises "reassuring certainty, order, predictability and control" (Nelkin and Lindee 1995:194). The social construction of "the gene" that the authors outline subsumes personal responsibility, in that it is nature and cannot be altered (given the medical technologies available today). They conclude that in media, genes are often granted the capability to determine family, sex, race, sexual orientation and certain personality characteristics such as aggressiveness and creativity, for example. "Genetic essentialism is a narrow way of understanding the cultural meaning of the body---it erases complexity and ambiguity...Problems and opportunities both disappear behind the double helix that has loomed out of proportion in the social imagination" (Nelkin and Lindee 1995:196). There are natural limits constraining possibilities when one adheres to the idea of a preordained by science "genetic destiny" (Nelkin and Lindee 1995:100). In line with Nelkin and Lindee's genetic essentialism, Alper and Beckwith (Alper and Beckwith 1995) define a kind of "genetic fatalism" in public discourse: an assumption that a genetic association is deterministic and means a trait or behavior is unchangeable.



The American Medical Association, through its Council on Ethical and Judicial Affairs has gone so far as to say that "There is a substantial body of misleading

information relayed to the public almost daily about the implications of genetics" (Council on Ethical and Judicial Affairs 1998:19). The AMA also believes that, as a result of this misinformation, many in the general public believe "genes are...the source of inevitable outcomes and predetermined conditions...[and] to many [genes are] the essence of human beings" (Council on Ethical and Judicial Affairs 1998:19). As a result of such misrepresentations, the Council suggests that current social conceptions of self have shifted to place "tremendous significance" on genetics (Council on Ethical and Judicial Affairs, 1998:19). This supports my argument that the media are socially significant in the determination of the role of genetics in society---playing a definite role in the geneticization of society.



Translating Genetic RISK: The Media and Numbers

A topic related to the misinformation disseminated by the media concerns genetic risk. Our "risk society" (Beck 1992) is partially a product of risk reporting-the extended coverage of risk in the mass media. Of interest here is why and how particular risks become topics of focus. Rose (2001) extends Beck's (1992) concern with risk society, arguing that contemporary biopolitics is "risk politics" linked to the molecularization of truth regimes in the life sciences, and that biomedical developments are deeply intertwined with technologies of the self. He (Rose 2001:17) coins "ethopoltics" which "concerns itself with the self-techniques by which human beings should judge themselves and act upon themselves to make themselves better than they are."

Kitzinger and Reilly (Kitzinger and Reilly 1997) examined media coverage of human genetics research to determine how risk is presented and when and why the media focus on a particular risk issue. These authors argue that theoretical accounts of "risk

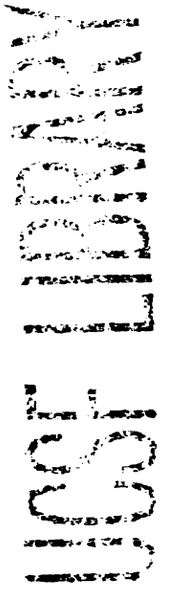
society" often oversimplify the media's role in the creation of such, demonstrating how "source competition, journalists' training, 'newsworthiness,' news momentum and the organization of news beats and media outlets encourage certain risks to be highlighted at particular times, but encourage other risks to be entirely overlooked" (Kitzinger and Reilly 1997:319). Risk is unavoidable in the prenatal genetic testing arena: risk for miscarriage, age-related risk for genetic abnormalities in pregnancy, and risk for genentic disorders in the fetus based on family history. I assert that part of the biomedical construction of PGT as a social good is facilitated through the social construction of PGT risks as "reasonable" in order to predict the health of the baby and to avoid having babies with disabilities.

A recent study examined genetic news articles published in the US, Canada, the UK, and Australia between 1995-2000 (Caulfield and Bubela 2002:poster) and found the majority of newspaper articles had no exaggerated or erroneous claims (62%), while only 11% of newspaper articles were categorized as having moderately highly exaggerated or erroneous claims. The authors concluded that scientists' beliefs that their work was being distorted through the media are unjust, despite the fact that the remaining 27% fell somewhere in between exaggeration and no exaggeration.

Genetic risk is commonly quantified numerically, and numbers are difficult to translate into everyday, practically applicable language. This is problematic because the healthcare system in the US requires that we make more of our healthcare choices independently, particularly for genetic decisions where the providers are "non-directive" and therefore unwilling to tell you to do something (Rapp 1999; Karlberg 2000). Deciding what is best for our health has become such a problem, that the National

Institute on Aging has developed a fact sheet for consumers called "Understanding Risk: What do those headlines really mean?" (NIA 2003). It outlines the basics of epidemiology and explains relative risk and absolute risk. Kertesz (Kertesz 2003) argues that translations are necessary because research studies are written for academic audiences, and the journalists who translate them are not academicians. Due to journalists' lack of expertise, many of us do not get the information we need in order to make informed decisions, and occasionally we are alarmed or confused unnecessarily. She asks: "How much of the responsibility for reporting public health information should fall to journalists?" (Kertesz 2003:10).

Pregnant women are inundated with numbers regarding their individual specific risks for miscarriage and genetic abnormalities in pregnancy (see Chapter 6). The media's framing of PGT as a social good enables some women to accept these risks and have PGT without further research. Other women play with the numbers to make them discernable to their particular definitions of acceptable risk. Regardless, the way the numbers are presented in the media, as either "big" risks or "insignificant" risks, inevitably influences the way women perceive these numeric quantifications.



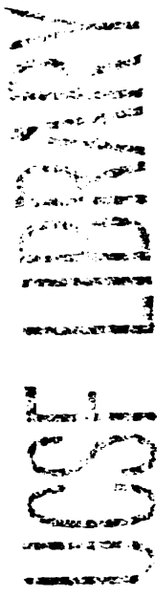
Scrutiny Of Media Coverage of Genetics

Media coverage is generally assessed in the genetics arena through measurements of public comprehension. Below, I briefly examine recent research on genetics in the media chronologically to provide an overview of the flow of these discourses. I propose that media coverage is pertinent to the understandings and misunderstandings between pregnant women and their genetic care providers regarding the capabilities and limitations of prenatal genetic testing technologies.

Durant, Hansen and Bauer describe public understandings of the new genetics as: "active constructs, the products of multiply-mediated historical and cultural (including mass media) influences, which may be expected to diverge significantly from those professional understandings of science with which they coexist" (Durant, Hansen et al. 1996:236). They argue that public understandings of science are important because they are capable of exercising a powerful influence over both biomedical research and clinical practice.

Alan Petersen examined how the print news media "framed" stories on genetics and medicine by analyzing genetics articles in three Austrailian newspapers between August of 1996 and April of 1999 (Petersen 2001). Petersen argues that because of the unique position journalists hold, standing between the genetic scientists and the public, they are in a position "to play an important role in shaping public perceptions of genetics and its value and applications, by selectively presenting some subthemes and not others" (Petersen 1997:1256). He found that genetic research is generally positively portrayed and is featured prominently in newspapers. A common technique to draw readers in is the human interest element, either interviewing those with a genetic disease or individual researchers studying genetics. This strategy simultaneously universalizes and personalizes human experience. Genetic researchers are portrayed as defenders of the public's health. There is little debate about the value of pursuing a particular line of research, and rarely is there any refutation of earlier reported findings. "In all the newspapers, there is a strong bias in favour of the positive outcomes of genetic research, and an apparent disinclincation to report negative or inconclusive findings" (Petersen 2001:1265).

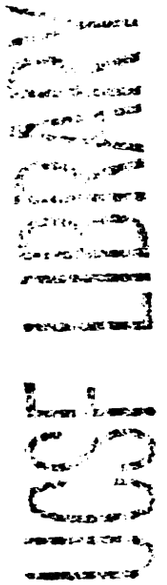
Overall, Petersen's research confirms findings of previous research on media and genetics which show that, in general, the media portray genes as omnipotent and reinforce the view that nature and culture are independent phenomena (e.g. Nelkin and Lindee 1995; Van Dijk 1998). The main point of media coverage about genetics, in Petersen's view, is that genes are the *foundation* of illness and health, and by unlocking the secrets of the genes, the world will be free of disease. "As new genetic technologies become more and more integrated in preventive medicine and public health---in screening , testing , counseling, and treatment---genetics will radically alter the way we view our bodies, and our ability to manipulate and control our environment" (Petersen 2001:1267).



Conrad (Conrad 2002) points to four implications of the media portrayal of genetics issues for public understanding of genetics. He found that the "genetic optimism" of the media serves to raise public expectations of medical genetics, encouraging the public to expect treatments and cures based on the discovery of yet another gene that may be linked to a specific behavior or disease. Another problem Conrad found repeatedly was the simplification of scientific findings so that many of the scientific caveats or qualifications are lost. Disconfirmations and lack of replications of research citing genetic connections to behaviors are rarely printed. He argues that this can produce "errant cultural residues, obsolete ideas that remain part of public knowledge" (Conrad 2002:74). His critique of the news media catch phrase to signify complex phenomena is exemplified in what he calls the "OGOD model-one gene, one disease" assuming that one gene determines the disease or trait (Conrad 2002:66). While Conrad cautions that the media often simplifies to OGOD, such as "the gay gene", he

allows that it may be scientists' exuberant optimism that is reflected in the journalists' portrayals. The OGOD model obviously contributes to the geneticization of human problems. As a result, attention shifts away from analyzing social and environmental conditions that may contribute to poor health to an emphasis on individual responsibility for health through genetic analysis and possible treatment.

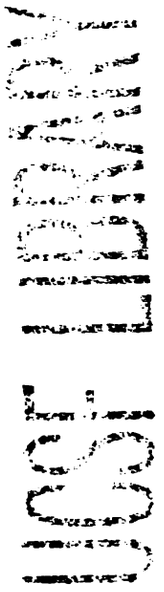
Conrad concludes with a harsh judgement of media portrayals of genetics. "The media's coverage of genetics---whatever excellence is achieved in terms of information, technical accuracy, and clarity of presentation---manifests journalistic conventions that ultimately misrepresent the role of genetics in human behavior....Media presentations of genetics are the raw materials by which individuals and communities create their own understandings of the role of genes in disease and behavior"(Conrad 2002:76-77). He argues that because news depictions are a key source for shaping public understanding, community perceptions will reflect media perspectives on the role of genes in life.



Public Interest Stories and Genetics In the Media

Studies of public understandings of genetics have highlighted how fictionalized accounts of individual encounters with genetics may be referred to in discourse. Further, they have shown that non-news outlets such as women's magazines have an impact on readers (Richards, Hallowell et al. 1995; Kerr, Cunningham-Burley et al. 1998). For example, in an attempt to further clarify the impact of the media on public perceptions of breast cancer genetics, Henderson and Kitzinger (Henderson and Kitzinger 1999) examined both traditional news media outlets and explored human interest stories, women's magazines, 'true-life' tales and fiction. Human interest stories are of consequence because, based on the focus group data the authors collected, it was these

types of stories that had made the "most impression on women's understanding of genetic risk" (Henderson and Kitzinger 1999:65). These stories often focused on family dynamics, dilemmas, and relationships. Non-traditional venues for breast cancer genetic information were significant: a storyline in a medical soap opera, a radio play, a BBC documentary, and a human interest story that followed a women through the testing for breast cancer genes on television. The volume of human interest stories on genetics is also of interest because of the media's "carrying capacity." That is, if an issue is simultaneously addressed in a range of different forms/outlets, then the media's carrying capacity for that issue is greatly increased (Hilgartner and Bosk 1988). Also, an interactive media momentum may be generated (Kitzinger and Reilly 1997). These nontraditional formats for news dissemination are likely to reach different and wider audiences and may generate distinctive types of audience engagement (Henderson and Kitzinger 1999).



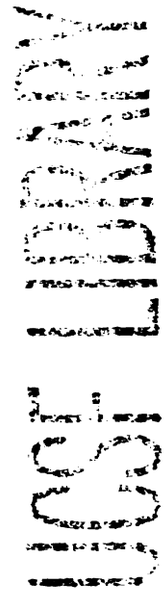
"Personal stories can put a 'human face' to the science and were seen as an accessible way of exploring social implications and personal dilemmas as well as capturing audience attention"(Henderson and Kitzinger 1999:68). Human interest stories focused on genetic inheritance can evoke powerful emotions, from fear to tragedy, as well as disrupting relationships and invoking decision-making, secrets, and narrative suspense. Further, the contrast between 'hard' and 'soft' news stories is becoming increasingly blurred. Henderson and Kitzinger suggest that soft reporting might be more accessible and better suited to the important task of addressing psycho-social issues, but may also mask scientific information behind emotion and distort the issues into clichés. The authors argue that soft news prioritizes ordinary women's accounts while hard news

uses scientifically rigorous sources, but often the hard news is black and white while some of the appeal of soft news is its messy gray areas. They conclude that attention to the culturally representative soft forms of genetic news is "particularly important" in the changing climate of scientific development and increasing interest in the social and psychological aspects of genetics.

National Public Radio (NPR) produced a group of radio talk shows called The DNA Files with one of its sections titled: "Prenatal Testing: Do you really want to know your baby's future?" The show, like most NPR segments, contained an enormous amount of information in a compact one hour time slot. There were experts in the fields of law, genetics, genetic counseling, genetic law, abortion, racism, feminism, and many others, as well as women who had had testing and women who were parents of children with genetic diseases all talking in a conversational style about prenatal genetic testing technologies. The framework allowed for the public interest stories to highlight what the experts were saying, in a way that did not exploit the women's experiences outright. But the women were explicit about what they felt. One woman who had had amnio without giving it much prior consideration was told the fetus carried a chromosomal defect: "I felt almost poisoned inside-that I had this information and if I gave birth to a child that was severely disabled, having had this prior information, I don't know how I would have dealt with it." (McAnnally 1998:21). There is little doubt that this kind of heart-wrenching description would capture some people's imaginations.

Choosing Naia: A Family's Journey is the chronicle of a couple having prenatal testing and making the decision to continue the pregnancy knowing their future child will have Down Syndrome. This book was written by Pulitzer Prize finalist Mitchell Zukoff,

who contacted the couple through their genetic counselor just after they received the diagnosis and followed them throughout the pregnancy, delivery, and Naia's first few years of life. The book began as a weekly series in The Boston Globe. The family narrative is interspersed with facts about Down syndrome, prenatal genetic testing, general genetics, and interviews with other families with children with Down Syndrome as well as with people who opted to abort Down syndrome fetuses. The website: www.beacon.org/naia posts pictures of Naia throughout her life, linked to the titles of the chapters. Upon publication of the book in 2002, Dateline NBC did a one-hour segment on the family. The book's jacket says: "...Choosing Naia examines the exploding world of prenatal information-and the emotional maelstrom that ensues from an unwanted test result-through the prism of the Fairchild family's searing experience" (Zuckoff 2002).



These examples of media coverage of PGT and its consequences and of genetic science more generally provide a sense of the range of genetic discourses to which people are exposed. While my research in this dissertation does not focus on the media per se, such discourses were routinely mentioned in the interviews.

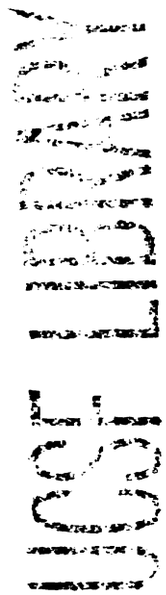
SECTION III: WOMEN AND PRENATAL GENETIC SCREENING AND TESTING

The existing literature on women and prenatal genetic testing is a vast array of publications in venues ranging from traditional peer-reviewed medical journals to women's magazines. I focus primarily on medical journals and the sociological/anthropolical literature, but delve occasionally into mainstream publications regarding my topic. I begin the literature on the use of testing, and then turn to the social construction of testing as routine and a "necessary" choice. This discussion leads into the

literature on the PGT and abortion issue. From there I move to the literature on the emotional nature of testing, emphasizing the multiple ways women and their families express knowledges about the choices and decisions they make with this kind of information available to them. Finally I delve into the complex relations between prenatal genetic testing and families.

Utilization

American society has been altered by the revolution in prenatal genetic testing. In 1999, there were genetic tests for 700 inheritable conditions, testing for 344 of which was available from a physician (rather than in a clinical trial setting), and 211 of them were available prenatally (Singer, Corning et al. 1999). The number of amniocenteses performed in the U.S. jumped from 8,700 in 1980 to 74,000 in 1989 (Meaney, Riggle et al. 1993). A 1992 survey conducted by Lewis Harris and Associates for the March of Dimes Birth Defects Foundation found that 99% of Americans thought they would have PGT during pregnancy to determine whether their future children would be likely to have a "fatal" genetic disease.



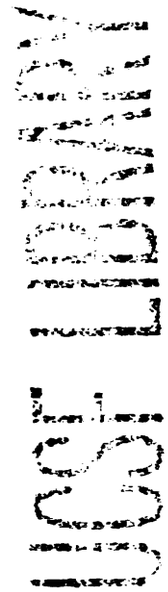
Of women who presented for genetic counseling in the first trimester, 96% of them had testing, while 85% of women coming in for counseling during their second trimester had the tests (Pryde, Drugan et al. 1993:498). A more recent study examining PGT utilization in New York state in 1983-1993 noted that the number of women having amnio or CVS increased between 1979 and 1993(Olsen and Cross 1997). Between 1986 and 1993, the number of women having PGT stabilized at 44.7% of women who carried a live fetus to 20 weeks. Interestingly, the number of women under 35 years old having PGT steadily increased (Olsen and Cross 1997).

In Victoria, Australia in 1998 approximately 60% of older pregnant women (women over 37 who are provided free testing) were having amniocentesis or CVS: 54% of women aged 37-39 and 73% of women older than 39 (Halliday, Warren et al. 2001). In England, a study of over 4,000 pregnant women, found that of women offered CVS after a positive nuchal translucency test or positive serum screen, 83% accepted CVS (Spencer, Spencer et al. 2000). Rapp (Rapp 1999) cites a personal communication with E. B. Hook, a prominent prenatal epidemiologist who believes that rates of acceptance of PGT will never be higher than 70%.

The question of why women chose to have or not to have amniocentesis or chorionic villus sampling, alone or coupled with the array of prenatal genetic screening tests, is one that has challenged medical and social science researchers since testing became available. I do not believe there is only *one* answer for any one woman. My research is concerned only with women who have PGT, so the research I cite is interested in this as well. The choices tangled with the decision to have testing are numerous, usually complex and interrelated. Many suggest that women have testing for reassurance, seeking to rule out genetic disease, or seeking knowledge about the genetics of the fetus, be it something mundane like sex or something many consider more serious like Down syndrome.[3] Other concerns found in this review include the miscarriage risks from having the testing, the timing of testing in terms of making an abortion decision, prevention of bearing and raising a child with disabilities, religion, pressure from providers and many others.

[3] Please see: (Beeson 1983; Beeson 1984; Rothman 1986; Rapp 1988; Kolker 1989; Rothman 1989; Scholz, Endres et al. 1990; Richards 1993; Singer 1993; Kolker and Burke 1994; Rapp 1994; Rothenberg and Thomson 1994; Jorgensen 1995; Balsamo 1996; Green and Statham 1996; Press and Browner 1997; Rapp 1997; Rapp 1999; Andrews 2001; Markens 2002; Suter 2002; Zuckoff 2002).

There are two issues that are not obvious considerations in my study because of the homogeneity of my sample: race and socio-economic level. Race and PGT has been addressed by many.[4] Rapp (1993,1994,1998) challenges traditional upper-middle class, white understandings of mental retardation, showing through her research in diverse populations of black and Spanish speaking peoples that some cultures do not place such a high value on normal intelligence. She found that culturally for Hispanic and some African-American populations, obvious physical impairments are more significant considerations when thinking about PGT and abortion. Duster (1990), Roberts (1997) and Nsiah-Jefferson (Nsiah-Jefferson 1993) all assert that people of color experience greater barriers to PGT because of access, cost and education, while also framing PGT as a covert mechanism for the social control of racial minorities. Hispanic and African American women are generally less likely to have PGT than white or Asian women.[5]



The financial implications of testing are linked directly to insurance status and socioeconomic levels.[6] Interestingly, no American studies evaluated insurance coverage as a factor in having PGT, muchless abortion coverage. I believe the lack of attention to this issue is linked to the higher socioeconomic levels of the types of women assumed to have PGT, implying that typically women who have PGT have insurance or can afford to pay out of pocket (e.g.Evans, Pryde et al. 1993; Pryde, Drugan et al. 1993).

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[4] Refer to: (Beeson and Golbus 1985; Duster 1990; Duden 1991; Rapp 1993; Press and Browner 1994; Rapp 1994; Browner, Perloran et al. 1996; Kuppermann, Gates et al. 1996; Roberts 1997; Browner and Preloran 1999; Rapp 1999; Beeson and Duster 2002; Zlotogora 2002)

[5] Please also see: (Browner and Press 1995; Browner, Perloran et al. 1996; Kuppermann, Gates et al. 1996; Beeson and Jennings 1998).

[6] Refer to: (Rothman 1989; Kevles and Hood 1992; Nsiah-Jefferson 1993; Rapp 1993; Andrews, Fullarton et al. 1994; Julian-Reynier, Macquart-Moulin et al. 1994; Kolker and Burke 1994; Rothenberg and Thomson 1994; Rapp 1999).

Routinization

Once women are offered prenatal genetic testing, a decision-making process is set in motion. Given my theoretical stance on the social construction of genetics within the biomedical realm, I assume that the decisions women make about testing are not independent of the culture in which they live, their families, the biomedical industrial complex, the particular medical environment, and partners' positions on the matter. The social arena of prenatal genetic testing is the space within which the pregnant woman makes her decision. This "choice" is a question much debated in the literature. Many researchers have examined refusal of PGT, and the body of research is thoughtful.[7] One line of argument builds on the problematization of choice and links it with a parallel problem in the biomedicalization of genetics: the routinization of prenatal genetic testing. Among others, these authors argue that the ways women are made aware of testing and its risks and benefits encourage them to "take advantage" of the technology, and "learn all you can" about the pregnancy, making the decision not to have testing very difficult if not impossible (Rothman 1989; Cowan 1992; Lippman 1994; Rothenberg and Thomson 1994; Rapp 1998; Rothman 1998; Duster 1999; Bennett 2001; Asch 2002; Markens 2002). Routinization encourages the acceptance of testing because offering PGT now falls under the normal "standard of care" for pregnant women in much of the U.S.

With the routinization of prenatal diagnostic procedures such as amniocentesis, pregnant women may be more inclined to undergo testing. Assumptions may be made

[7] For examples, please see (Beeson and Golbus 1985; Rapp 1998; Markens, Browner et al. 1999; Rapp 1999; Beeson and Doksum 2001).

that women who are over 35 and thus "high risk" *should* have amniocentesis.[8] Prenatal screening and testing seem to be supported in the U.S., and the general belief is that such actions are "good" (Beeson and Jennings 1998; Suter 2002). A former genetic counselor who is now a law professor, explained her experience:

> As a genetic counselor, I observed that most, though not all, patients to whom we offered genetic screening or testing accepted it, often with seemingly little difficulty. Now as a law professor who teaches and writes about the ethical and legal aspects of genetic testing, I witness the same attitudes among my students...And finally, as I gained a new facet to my identity (the pregnant woman of advanced maternal age) I was struck by how many people, no matter how familiar, raised the subject of prenatal testing, and even more by the way they phrased their query. Most did not ask *whether* I underwent amniocentesis or CVS, but instead, *what kind* of testing I chose...In short, my professional and personal experiences suggest that reproductive testing for "older" mothers, and genetic screening for all pregnant women, has become almost as routine as forsaking alcohol and caffeine during pregnancy (Suter 2002:241, italics added).

This routinization allows for the translation from testing to responsibility: a woman must be responsible for the type of child she bears. A woman with a baby born with Down syndrome is automatically asked whether or not she had a prenatal test, implying that if she had, the baby would have been normal or not born at all. The implication is that women now have control over what type of child they bear. Two studies (Marteau and Drake 1995; Menec and Weiner 2000) found that women who did not have prenatal genetic testing were blamed when a child was born with a disorder that could have been detected. *Genetic Delimmas: Reproductive Technology, Parental Choices, and Children's Future* (Davis 2001) argues that because children are entitled to open futures,

[8] Routinization has been problematized by many, arguing that it limits choice (Press and Browner 1994; Browner and Press 1995; Green and Statham 1996; Press and Browner 1997; Suter 2002).

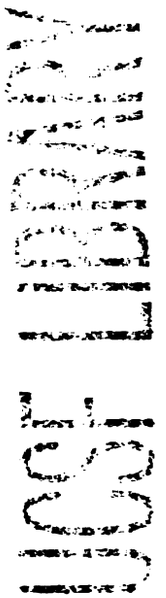
unethical to deliberately opt for a disabled child to be born. Does prenatal
esting then mean one "should" abort if something abnormal is found?
app (1999:306) called women who have PGT "moral pioneers" and "moral
ners of the private." Because of the control women have over reproducing the
app argues that women are "culturally positioned to think about their
tive capacities, desires, and decisions as a private dimension of public life"
7). She proposes that reproduction itself is located at the heart of what a culture
tself to be, a representation of self.

n

renatal genetic screening and diagnostic tests do not provide many alternatives
n who carry an abnormal fetus. The most common "treatment" for a negative
iagnosis is abortion. There are very few other options, and they are usually
extraordinarily expensive, such as fetal surgery (Casper 1998). Presently, fetal
s only offered at four U.S. hospitals (University of California San Francisco,
s Hospital of Philadelphia, Vanderbilt, and George Washington) and is only
in clinical trials. This means one cannot request it and have it, but rather must
ain criteria to be allowed to enter into the trial. At the 2002 annual meeting of
ican Society for Human Genetics in a session on fetal therapy (Wilson 2002), it
ested that despite a number of obstacles, in utero fetal surgery is, at the present
linical trials for treating the following congenital malformations:
ningocele, cystic adenomatoid malformation of the lung, sacroccygeal teratoma,
twin transfusion syndrome. Fetal surgery carries risks of premature delivery,
h, maternal complications, maternal death, neurodevelopmental delay in fetus,

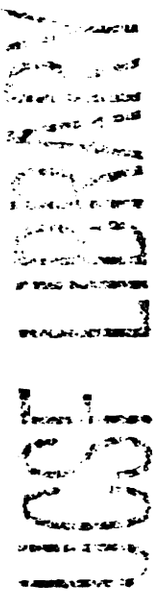
vascular compliance in infancy, and infant death after birth, among many others (Wilson 2002). However, *none* of the congenital anomalies currently being treated by fetal surgery are indicated directly by PGT, which means that it is not directly useful based on information received from PGT. With abortion as the only "treatment" option today for women who are diagnosed with a fetal abnormality, women's attitudes about abortion are of great import.[9]

The timing of PGT is an important consideration because of the link to abortion, with the belief that an early abortion is easier and safer, both emotionally and physically. Two studies found that women were more likely to abort the earlier in the pregnancy the diagnosis was made (Pryde, Drugan et al. 1993; Zlotogora 2002). Many studies determined that women preferred CVS testing because it could be conducted earlier in the pregnancy (Lippman, Perry et al. 1985; Abramsky and Rodeck 1991; Evans, Pryde et al. 1993). Lippman's research revealed that women who were concerned about the risks of PGT preferred amnio because it had a lower quoted risk for miscarriage (Lippman, Perry et al. 1985). In other research, some women simply said they wanted an abortion if there was something wrong (Rapp 1999; Borsack, Metzenberg et al. 2002). The legal landscape of abortion is particularly pertinent for women who chose to terminate a pregnancy, whether because of a genetic diagnosis or by choice, if the termination takes place later than 18 weeks. (For a full discussion please see Appendix C.)



[9] For a complete review of the history of abortion and the challenges to it from a sociological perspective please see Luker (1984), Reagan (1997), and Petchesky (1990). There is a whole body of literature in ethics regarding abortion in prenatal genetic testing covering wrongful birth and wrongful life statutes. Wrongful birth occurs when parents are not warned that they may be at risk for conceiving and/or bearing a child with a serious genetic disorder. Wrongful life is when the claim is based on the belief that one would be better off if life had never been granted. Wrongful life and wrongful birth are evaluated on the debilitating factor and whether or not life is truly harmed by it. For an example of these please see Botkin and Mehlman (1994), Jacton (1996) and Robertson (1996). And for a note on legal rights to regulate a woman's behavior during pregnancy, see Solomon (1991).

The severity of the anomaly diagnosed through PGT is a significant factor for some when considering abortion. Multiple studies cited severity as the deciding factor in whether to consider selective termination: (Drugan, Greb et al. 1990; Evans, Pryde et al. 1993; Pryde, Drugan et al. 1993; Singer, Corning et al. 1999; Zlotogora 2002). In Hawaii between 1986 and 1997, 50% of the pregnancies found to have Trisomy 18 and 40% of the pregnancies that had Trisomy 13 were electively terminated following PGT (Forrester and Merz 1999). In another study, the birth prevalence of Down syndrome dropped by a stunning 80% between 1979-1999, and the likelihood of termination of pregnancy after PGT was much higher for chromosomal anomalies (64%) than for nonchromosomal congenital anomalies (10.2%) (Alembik, Roth et al. 2002). Linked with severity of diagnosis, two studies (Pryde, Drugan et al. 1993; Sagi, Meiner et al. 2001) further stated that when a genetic diagnosis was *not* clear-cut, women were more inclined to consider abortion.



In terms of race, another recent study (Lee, Khoshnood et al. 2001) showed a disturbing trend of increased mortality of non-white infants from congenital birth defects: in 1970 the congenital birth defect rate was higher among whites (3.1/1000 live births) than non-whites (2.6/1000), while in 1997 the rate in whites (1.6/1000) was lower than in non-whites (1.7/1000). The authors suggest the discrepancy could be due to racial differences in access to and use of prenatal screening and pregnancy termination, indicating that whites utilize prenatal diagnosis more and are more inclined to terminate than non-whites.

While the severity of the diagnosed disease weighs heavily, religion has also been found to affect abortion decisions. In American society, political opposition to abortion

is frequently linked to religious beliefs, so I include in this summary those studies of PGT examining women opposed to abortion. Overall, women who said they were opposed to abortion or would not have an abortion because of religious considerations interestingly still tend to have prenatal genetic testing or screening(Beeson and Golbus 1985; Pryde, Drugan et al. 1993; Singer 1993; Beeson and Jennings 1998; Rapp 1999; Halliday, Warren et al. 2001). Historically, those opposed to abortion were assumed to be uninterested in prenatal screening, as they would not abort an affected fetus. This has been refuted by Pryde and others (Pryde, Drugan et al. 1993). A group in Australia found that even though women in their sample refused amnio or CVS, a "vast majority" of them (82%) accepted prenatal screening, despite 43% saying they were opposed to abortion (Halliday, Warren et al. 2001). Another study found that about 60% of Catholics, who are traditionally assumed to be opposed to abortion, would want prenatal testing and 37% would abort in the case of a serious defect(Singer 1993).

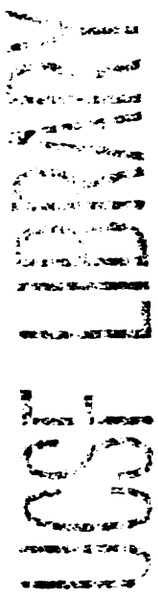
Decision-making around having PGT is thus indelibly linked to abortion, be it through timing of the testing, personal feelings about abortion, or a desire to prevent births of disabled children. This yearning to have a child free of genetic abnormalities has been explored in a body of research that found that women who have had exposure to people living with a particular genetic disease are generally less inclined to consider abortion of a fetus with that disorder (Saxton 1984; Beeson and Golbus 1985; Evers-Kiebooms, Denayer et al. 1990; Pryde, Drugan et al. 1993; McAnnally 1998; Rapp 1999; Asch 2002; Beeson and Duster 2002).

A related aspect is the role of money and the distribution of public funds for those with disabilities. There are studies assessing economic savings from the prevention of

births of children with costly-to-care-for birth defects, many of which can be detected prenatally (Henderson 1991; Ennever and Lave 1995; Kuppermann, Gates et al. 1996; Shackley 1996; Jallinoja 2001; Cusick, Buchanan et al. 2003; Grimshaw, Szezepura et al. 2003). Rapp (1999) points out, along with numerous disability rights advocates like Saxton (1984), that blatantly inadequate support of disabled individuals and their families makes it extremely difficult to opt to have a disabled child.

Last, studies that have examined women's experiences of a selective termination following a genetic diagnosis found that many women suffer severe emotional consequences from choosing termination of a desired fetus, sometimes impacting everyday activities and lasting as long as a year after the abortion (Black 1989; Black 1994; Kolker and Burke 1994; Green and Statham 1996; Rapp 1998).

In sum, women who have had an abnormal diagnosis from PGT face a complex decision-making process involving many personal, familial, cultural, biomedical, and social factors. Women's decisions about abortion in the case of fetal abnormality are not predictable, as the literature shows. Yet the assumption is still commonly made that abortion will be performed in the event of a birth defect diagnosis.



Emotions

Regardless of the outcome, women's feelings about the very experience of pregnancy have been altered by PGT. The emotions provoked by PGT do not only occur when there is a positive result. Much research is devoted to the waiting period between having the testing/screening and receiving the results. Two pioneering and influential scholars in the sociology of prenatal genetic testing suggested two decades ago that women attempt to withhold attachment to the developing fetus until the results come

back from PGT (Beeson 1984; Rothman 1986). Beeson (Beeson 1984) termed this "suspension of pregnancy," while Rothman (Rothman 1986) called it "the tentative pregnancy." Rapp's decade of ethnographic research in the prenatal genetic testing arena supports their findings (Rapp 1999). This anxiety pregnant women experience reflects the desire to know that the baby will be healthy and well before committing to the pregnancy. These yearnings are considered "normal" in our society, and expected.

Prenatal genetic testing and screening options induce anxiety during a critical time in pregnancy. Considerable research has been conducted that found anxiety in women who have a positive screening/diagnostic test or a postive carrier screening test, indicating that they carry a defective gene.[10] Mennie and colleagues (Mennie, Compton et al. 1993) examined carrier testing for Cystic Fibrosis in pregnant couples, where the woman was found to be a carrier and her partner a tested non-carrier, meaning the fetus could *not* have CF. At the time of the screening, 23% of the women reported feeling anxiety. Women who were diagnosed as carriers were distressed; they had higher anxiety and depression than women who were not carriers.

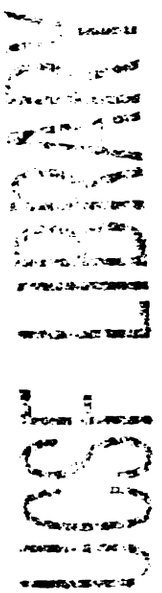
There is also a significant body of literature addressing how self-identity is affected by new genetic knowledge, such as a positive carrier screening test. These studies found negative affects in self-perception after a positive diagnosis.[11] Moreover, the genetic information does not even have to be disease related to be damaging to self-concept. One study examined non-disease genetic information relating to the ability to gain strength and muscle mass. People were told what genes they had and answered a

[10] See for example: (Kenen and Schmidt 1978; Beeson and Golbus 1979; Mennie, Compton et al. 1993; Rona, Beech et al. 1994; Tibben, Duivenvoorden et al. 1994; Jorgensen 1995; Lerman, Narold et al. 1996; Rapp 1999; Ettorre 2002; Green, Murphy et al. 2002)

[11] Please see: (Kenen and Schmidt 1978; Marteau, Duijn et al. 1992; Seibert 1995; Lerman, Narold et al. 1996; Karlberg 1999; McConkie-Rosell and DeVellis 2000; Ettorre 2002)

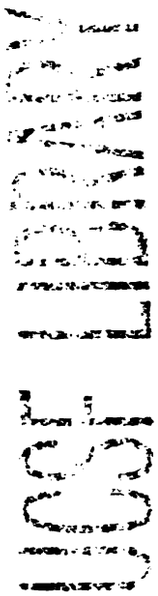
series of questionnaires about self-identity. The authors found that 14% of respondents felt that the genetic information they received changed their self concept, and 33% said that the information would change their behavior (Gordon, Devaney et al. 2002).

Significantly, it is not only women who have testing who worry about PGT and screening. Marteau and others (Marteau, Johnston et al. 1989) examined pregnant women who were eligible for but chose not to have MSAFP and amniocentesis. They found significantly more anxiety in these women interviewed at gestations too late to change their minds and have the testing. The reasons women refuse are diverse (Rapp 1998; Markens, Browner et al. 1999; Rapp 1999; Beeson and Doksum 2001). Markens and colleagues (1999) found that women refused because they perceived the testing as outside the realm of normal prenatal care and thus were avoiding risk by avoiding testing. Rapp's (1998, 1999) decade long research project on women and PGT found that state funding, community definitions, miscommunication between provider and woman, religious beliefs, women questioning scientific discourse, male partners, women's reproductive histories, missing the prenatal appointment, or presenting for prenatal care too late all played a role in women refusing amniocentesis. The most common reason Rapp (1999) heard for refusing PGT was risk of miscarriage from the procedure. Beeson and Doksum (2001) cited "family values" as the foundation for resistance to genetic testing, specifically framed as "culturally sanctioned resistance" such as religion and romantic love and "experiential resistance" expressed by those who knew or lived with someone with genetic disease.



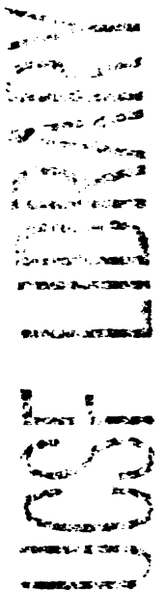
Families

Part of the anxiety women experience vis-à-vis PGT is related to how their families will relate to the new member in their genetic family and what that new family member will mean for the family. Medical genetics is by nature a science of families and relatedness. In order to provide clinically useful genetic information, often the family history of the trait in question must be mapped by inheritance, requiring other family members to provide not only information, but often samples of blood or other DNA related material. Genetic testing implicates families in numerous ways, the most obvious being familial emotional relationships (Wexler 1995; Rothman 1986; Rothman 1989; Rothenberg and Thomson 1994; Ginsburg and Rapp 1995; Rothman 1998; Rapp 1999; Andrews 2001). The emotional consequences of family genetics are vividly illustrated by Wexler's (1995) recollection of her experiences with Huntington's disease. Her mother died of Huntington's disease, but she and her sister only learned it was hereditary late in this tragic story. Wexler herself could be tested, as her sister helped discover the gene connected to the disease. In her book she describes her complicated decision-making about not having testing, and concurrently also deciding not to have children.



Family and kin definitions are challenged when genetics is introduced (Richards 1996; Rapp 1999. Others (Anderlik and Rothstein 2002; Finkler 2001; Peters, McAllister et al. 2001) suggest that there are legal ramifications of "family" in terms of the source and nature of genetic parental obligations, specifically paternity. These varied implications for family relationships are further complicated when genetic decisions must be made.

Complex relationships among family members alter decision-making. Genetic related decisions are tied to severity of the disease, consequences for self and family, and links to the family's values in important matters of life (Shiloh 1996; Rapp 1999; Beeson and Doksum 2001). Such decisions are not necessarily made independently by pregnant women, but often within familial discourse. Beeson and Doksum (Beeson and Doksum 2001) suggest that "family values" as "differing constellations of values" distinct to specific families and socially pattered, provide a framework for families making genetic decisions, incorporating religion, romantic love, everyday experiences and practices, and emotions. Further, understanding the inheritance patterns of disease may not be as easy as presenting statistics. Richards argues that family members commonly attempt to overlay their understandings of family traits passing down through members with the genetic inheritance patterns, with the results being incompatible with medical genetics (Richards 1996). For example, in order to explain the inheritance patterns of a particular disorder in a family, members may adhere to beliefs such as that the disorder skips a generation, appears only in first-born children, is associated with particular physical or personality types in the family, and so on. More research is needed on family and kinship understandings of the genetic issues they confront through PGT.



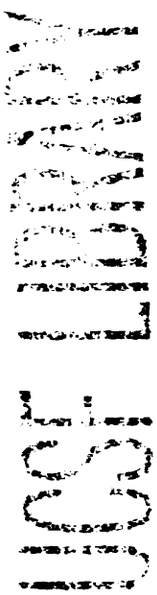
Comprehending one's risk for genetic disease is complicated by responsibility to one's family and the relevance of risk for one's position in the family and the family's position in society. Laurie (Laurie 2001) provides an overview of the relationship between individual and familial entitlement to genetic information. Juengst (Juengst 1999) suggests that genetic testing challenges the family through threatening family loyalty, intimacy and security. He suggests that prenatal testing can be perceived as a

protection the parent must offer the child, meaning that if the test shows an abnormality one should abort. Clinicians traditionally frame PGT as a service to the prospective parents "which allows them to reduce the risk of burdening themselves and their family with more than they can handle"(Juengst 1999:199). This is enhancing familial, rather than fetal, security. At the same time, it challenges parents to define the parameters of an acceptable life for their child. The literature shows that especially for women, the self in relation to the family, or the interdependent nature of the group to self, caused decisions about testing to be foregrounded, despite personal risk (Hallowell 1999; Rapp 1999; Nippert, Teige et al. 2002). This, I would argue, is because of the parenting responsibilities women feel even during pregnancy. The parenting pressure is such that women feel they should do everything they can to produce a "healthy" baby to join the family (McGee 1997; Suter 2002; Robertson 2003). Ida Bell(Bell 2003:55), the pseudonym of a woman who terminated a pregnancy just past 24 weeks, recounted her feelings of family responsibility and loss:

> I remember sitting in the clinic...feeling Ezra kick, and thinking that he wanted to be with us. For weeks afterwards, I think about that kicking. Small muscle twinges, even elsewhere in my body, remind me of that small but insistent presence, and I fall into the huge hole of this loss, Ezra's absence. But knowing how likely he was to suffer even if he survived the birth and the NICU, we decided to terminate the pregnancy. It's a parent's job to make the hard decisions. To make them go to bed, eat their vegetables. And we decided to end his life, before it had even started. It's very hard to feel lucky to have done this.

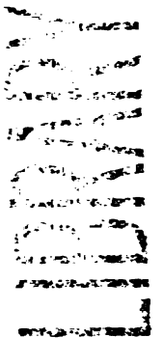
Risk can be closer or further away depending upon, for example, a diagnosis of the genetic disease in a sibling, or deciding to take a predictive test or have prenatal genetic testing. Genetic information is unique in that the actual diagnosis can be new information, but the genetic anomaly has been there in one's family for generations. Cox

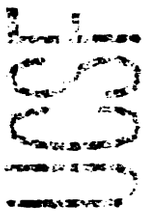
and McKellin's work support this point, noting that a genetic disease does not come about because of predictive testing, as one's genes are already there, but the brand new knowledge of one's status makes it feel as though it has just occurred (Cox and McKellin 1999). Women who have PGT and find out they are carrying a fetus with a genetic disease, and who then opt not to terminate the pregnancy, have health information about their future child that they normally would not have. If a woman found out she carried a child with the HD gene, and did not abort, that child would be born into a family whose members knew that s/he would develop the neurological illness and die at a relatively young age. Even if the genetic information gained will not have health implications, such as if the child is a carrier of a CF gene, the potential remains that the social relations within the family toward the child will be colored by genetics. Novas and Rose (2000:490) call this a "genetic network" and say that the "web of genetic connectedness" is "overlaid upon a web of family bonds and family memories, with their burden of mutual obligations and caring connectedness, and with all the ethical dilemmas they entail." They argue that within these genetic networks, those at genetic risk reevaluate their relations in terms of risk and inheritance.



One way to predict one's genetic status without testing is by doing a medical pedigree. Familial links and kinship are explored, enhanced, and defined through the creation of a family tree and its corresponding medical pedigree (Nukaga and Cambrosio 1997). Medical pedigrees have implications for defining "family" and "risk". Through this process, individuals are located in their family's genetic-based riskscapes, creating new notions of family, "thus modifying simultaneously the context and the relation of the individual to that context"(Nukaga and Cambrosio 1997:30). The authors argue for

viewing the medical pedigree as a boundary object (Star and Griesemer 1989) because it links different professional practices, and external, internal, collective and individual aspects of the body. Atkinson and colleagues (Atkinson, Parsons et al. 2001) also use this pedigree-as-boundary-object theoretical tool when examining the medical pedigree construction among genetic care providers. Nukaga and Cambrosio (1997:50) conclude that pedigrees are co-constituitive in relation to families, in that kinship relations inform pedigrees while pedigrees render kinship as "embodied in notions such as family disease". The medical pedigree is a "genetic remapping of a person's life in a biological and temporal space which contains the potential to reconfigure identity in terms of a genetic past, a genetic present and a genetic future" (Novas and Rose 2000:495).





Finkler (Finkler 2001) explores relations among kinship, family and genetics. She introduces the concept of the *perpetual patient*, someone who has entered the medical stream despite being healthy, which she theorized from interviews with women who were at genetic risk for breast cancer and adoptees who were searching for their birth parents because they wanted to know their health histories. Finkler found that genetic inheritance forges a lasting bond with blood kin because of shared genetic inheritance, even if they have little else in common. For the adoptees, kinship meant a biological connection which was validated by the medical history that established continuity with the present and the past. The biomedicalization of kinship can also create tensions between individualism and choice and an orientation to family and kin. With the dominance of the new genetics, there is a movement away from the what Finkler terms the modern family, where individuals choose their kin on the basis of affective ties, to one's kin being defined on the basis of birth and blood ties. Finkler goes so far as to say, "...the medicine

future will be the medicine not of the individual but of the family" where duals will no longer be able to make medical decisions independent of the family Finkler 2001:245).

In this brief review of social science literature pertaining to families and genetics, al has been to frame these issues so that the relationship between bodies and ons and families in prenatal genetic testing can be made clearer. I argue that h geneticization, women's pregnant bodies are produced as vividly including the and fetuses they contain, and not merely the bodies of individuals. One plishment of geneticization is to attribute genetic cause to social issues, including rmal" and potentially "abnormal" pregnancies. Families are altered by genetic oses, through personal responsibility to other family members, emotional nships, shared risk, and the social shaping of the definition of "family" by genetics. changes in perceptions of self and family are in part facilitated through prenatal c testing.

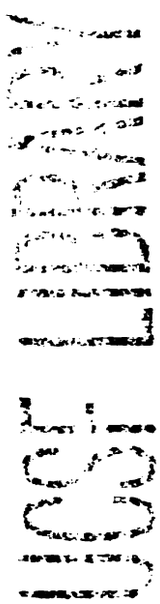
TION IV: PROVIDERS OF PRENATAL GENETIC TESITNG

The social science literature on genetic care providers is not extensive. It has d largely on non-directiveness as the philosophy of care and on how providers nce patients' decision-making.

irectiveness

Providing genetic care is different from general medical practice because of the eness of genetic information and its consequences for identities, families and s. In general medical practice, the physician makes a diagnosis based on the gs in a medical examination or test and suggests a course of action for treatment of

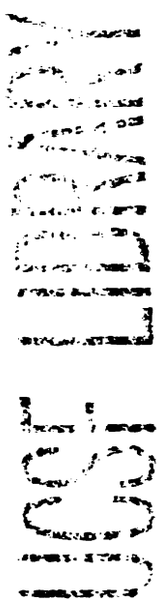
the problem. In sharp contrast, genetic care by medical geneticists and genetic counselors has not centered on recommendations for action. In most cases, the philosophy has been that providers are to present information and allow the patient to make an "informed" decision. Practicing genetic care "nondirectively" is a challenge for the providers. Patient autonomy is the foundational ethical principle of the practice of genetic care today (Bosk 1992), and the nondirective tenet was established in part to avoid the past abuses of genetics and eugenics (Johnson and Brensinger 2000). The National Society of Genetic Counselors' code of ethics states that genetic counselors should: "enable their clients to make informed, independent decisions, free of coercion, by providing or illuminating the necessary facts and clarifying the alternatives and anticipated consequences" (NSGC 2003).



Medical geneticists and genetic counselors generally follow the "nondirective" tenet, which asserts that the providers should not direct patients to make any particular decision in response to a genetic diagnosis. The principle of nondirectiveness in genetic counseling is based on "the assumption that most families would act responsibly, not on a principle of procreative liberty" (Paul 1998:147) or the principles of reproductive rights. Couples were assumed to act rationally in situations to achieve a normal child. Nondirectiveness today, according to Kessler, is "any procedure used in genetic services that promotes the autonomy and self-directedness of the client" while directiveness refers to "any procedure used in genetic services that uses one or more means to persuade a decision that might not otherwise have been made by the client" (Kessler 2001:188). Thus in a nondirective counseling session, "the good subject...becomes the individual

who will modify their lifestyle responsibly in relation to their genetic risk" (Novas and Rose 200:495).

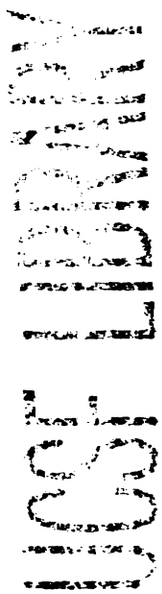
Yet, nondirectiveness, defined as counseling without being directional, is ultimately an unattainable goal, as all information is socially filtered through the individual presenting that information. The beliefs and experiences of the counselor/doctor and the pregnant woman and her partner and their lived realities may all be consequential in the interaction. Kessler's suggestion of counseling with the goal of assisting women to achieve personal reproductive-related goals seems reasonable, but requires excellent counseling skills which some if not many genetic counselors today lack (Michie, Smith et al. 1999; Kessler 2001). Moreover, some analysts suggest that the mere offer of prenatal genetic testing is, in essence, endorsement of the practice, thus encouraging pregnant women to have the amnio, CVS or prenatal screening offered to them (Suter 2002; Clarke1999; Bosk 1992; Johnson and Bresinger 2000;Veach, Bartels, et al. 2001; Bennett 2001). Bennett (2001) argues that the routinization of prenatal genetic testing itself hinders nondirectiveness because the goal of any routine test is to encourage utilization-"it's routine."



The stance on nondirective information about genetic disorders should then be reflected in patient materials. Most women who have prenatal screening or testing are provided written materials of some kind describing the testing and the disorders sought through the tests. Lippman and Wilfond (Lippman and Wilfond 1992) found a difference in the tone of materials provided prenatally and postnatally about cystic fibrosis and Down syndrome: prenatal information was more negative in tone and focused on medical complications associated with the disorder while postnatal information

emphasized medical and social advances in the treatment and management of Down syndrome and cystic fibrosis. A more recent study found that overall a negative image of Down syndrome was conveyed in printed patient information, but the cystic fibrosis information was more neutral (Bryant, Murray et al. 2001). Interestingly, with the Down syndrome information, the median number of sentences describing the condition was one, with 33% of the leaflets containing no descriptive information on the condition at all (Bryant, Murray et al. 2001).

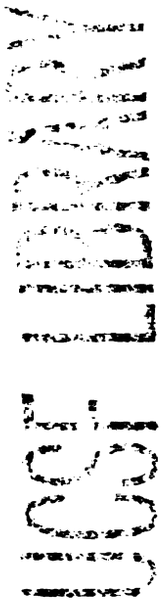
In practice, nondirectiveness is interpreted differently by different practitioners. Michie (Michie, Bron et al. 1997) found there was an average of 5.8 directive statements per counseling session in which the counselor provided advice about what he or she thought was the best decision for the patient. Another study found that the issues on which individuals are encouraged to focus can have a substantial effect on the decision made, suggesting that careful consideration may need to be given to the way in which counseling is carried out (Salkovskis, Dennis et al. 1999). That is, the very organization of the information by the providers is itself intrinsically directive. In sum, the interactions between pregnant women and their genetic counselors and physicians are inevitably colored by the perspectives of those involved. We can now turn to an explicit consideration of how providers influence the decisions of pregnant women about PGT.



Providers' Influence

Through the language they use, the presentation of information and intrinsic judgments about the emotional impact of the information presented, providers of genetic services are in a position to significantly influence pregnant women's perceptions of themselves and their potential children. Marteau and Drake (Marteau and Drake 1995)

found that obstetricians and the public when faced with different outcome scenarios were more blaming towards mothers who gave birth to a child with Down syndrome who had not participated in screening, than they were towards mothers not offered screening who gave birth to affected children. There is also some evidence from a preliminary study by Marteau that indicates that some parents blame their physicians for not detecting affected children during pregnancy (Marteau and Anionwu 1996). A German court awarded the parents of a disabled child 275,000 euros with an additional 10,225 euros for the mother for her depression(Cleaver 2002). The parents argued that the mother would have had an abortion if the defects had been detected via ultrasound around the 20th week of pregnancy. This decision came just after French court rulings made individual doctors liable if they fail to notify a pregnant woman of problems with the fetus that she might have otherwise aborted. Women are informed mainly through providers about the prenatal genetic testing and screening available, and providers may be held accountable if things go wrong.[12]



Bernhardt and Bannerman (Bernhardt and Bannerman 1982) studied PGT referral patterns of obstetricians and found that board certified obstetricians were significantly more likely to refer women for testing. Catholic obstetricians were significantly less likely to refer. Bernhardt and other colleagues (Bernhardt, Geller et al. 1998) found that obstetricians were more likely to make recommendations and less likely to present the testing as voluntary and that obstetricians' and genetic counselors' personal feelings

[12] For example, as early as 1978, a Philadelphia court found in favor of the defendants in *Gildiner v. Thomas Jefferson University Hospital* citing that the parents deserved medical cost compensation for the birth of their son who suffered from Tay Sachs disease after they had mistakenly been told amniocentesis results were normal. More recently, in a May 2004 Minnesota Supreme Court decision on *Molloy v. Meier, Backus and North Memorial Medical Center*, the court held in favor of the defendants suing because after being told their first child who suffered from undiagnosed Fragile X was genetically normal, they had a second child who also suffered from a more severe form of the disease.

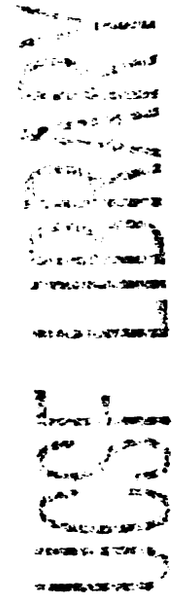
about abortion changed the information they would provide to pregnant women. Heckerling and others (Heckerling, Verp et al. 1998) found that despite patients' beliefs that their obstetricians played a role in the choice of prenatal genetic test, the physicians' preferences did not significantly impact the choice of prenatal test made by their patients. However, a survey of American obstetricians in 2001(Egan, Kaminsky et al. 2002) found that obstetricians use amnio 83% of the time over all other available diagnostic tests for Down syndrome, which would mean that the women who saw these obstetricians were possibly influenced by that preference. Rona and colleagues (Rona, Beech et al. 1994) found that genetic counseling after the birth of an affected child did not aid in understanding the genetic component of the disease, and even caused a deterioration in social interaction with friends and family. Press and Browner (Press and Browner 1993) argue that providers gave women information about the expanded alpha-feto protein test in such a way as to convince them the test was something they should have, rather than providing them with a "neutral" presentation and allowing the women to make their own informed decisions. Richards and Green (Richards and Green 1993) found that the routinization of prenatal diagnosis into antenatal care by genetic providers causes women who refuse amniocentesis to be labeled as "abnormal".

Wertz and colleagues have conducted a series of surveys of medical geneticists and genetic counselors world-wide to frame an ethics of the new genetics and found that overall, culture and professional training were more indicative of ethical slant than gender. This set of studies (Wertz and Fletcher 1989; Wertz, Fletcher et al. 1990; Wertz 1993; Wertz 1993a; Wertz 1997; Wertz and Fletcher 1998) extended over nearly ten years and examined the perceptions of medical geneticists from 37 nations on a number

of ethical issues. Of great interest here is the finding that, "Worldwide, 94-98% of the respondents would disclose ambiguous or conflicting prenatal test results to patients" (Wertz 1993a:7). For the United States, 75% of medical geneticists reported they would disclose colleague disagreement about ambiguous or "artifactual" test results, while 89% would disclose new or controversial interpretations of test results (Wertz 1993a:27). In the most recent of these studies (Wertz and Fletcher 1998), Wertz and Fletcher focused on ethical dilemmas of prenatal sex selection and found that 29% of geneticists queried would provide prenatal genetic testing and abortion services for male sex selection.

A recent survey of the American Society for Human Genetics members, who are qualified as genetics experts, stressed patient choice and patient education as essential in any kind of genetic testing (Rabino 2003). These responsibilities of allowing patient choice and educating patients fall primarily to genetic specialists, a growing field, but one which is not nearly large enough to handle the increasing demand for genetic testing. (Please see Appendix D for a complete discussion.) When asked specifically about pregnancy termination using hypothetical situations in the questions, around 80% of genetics experts felt it was most acceptable when there would be severe mental retardation, death by age 4, or development of a chronic childhood disease. Approximately 40% of genetics experts thought abortion was less acceptable if there would be death by disease in young adulthood or mild mental retardation (Rabino 2003):375. Abortion was considered unacceptable by genetics experts if the deciding factor is sex (88%), homosexuality (83%), depression (70%), obesity (70%), or developing a debilitating disease around age fifty (52%) (Rabino 2003:376). The genetics experts surveyed were overwhelmingly convinced that parents should have the

decision-making power in these issues around reproduction. Concern was expressed, however, when the selection and termination involved attempts to acquire preferred traits in a child, such as sex, level of intelligence, physical characteristics, etc.. Where does the boundary lie between personal choice and eugenics? Rabino argues, "Without an objective definition of "normal" it will become increasingly difficult to differentiate between prevention [of births of affected babies] and enhancement [of the genetics of families and offspring through selective abortion]" (Rabino 2003:378). Keller goes further to say that the geneticization of health and disease presents the problem of an "invitation to biologically and socially unrealistic standards of normalcy" (Keller 1994:97). Keller's and Rabino's studies suggest that there is concern among genetics professionals about the outcomes of the genetic care provided in prenatal genetic testing and the ethics around choices pregnant women and their families make about fetuses that are allowed to continue into babies and those that are not.



Another study of genetic counselors, primary care physicians and nurses with experience in genetics examined the professional and ethical issues these providers confronted in everyday practice (Veach, Bartels et al. 2001). The authors found 16 ethical and professional areas where the providers believed there were issues. The most interesting in terms of this research are the domains of withholding information, facing uncertainty, and determining the primary patient. The discussions of providers withholding information included lying; withholding unanticipated information (such as a finding other than the one tested for on a genetic panel); assuming the patient could not handle the truth; restricting patient's access to their own information; and protecting the patient. The providers' concerns with uncertainty were focused on the uncertainty of

long-term outcomes; limited information-the number of 'unknowns' in relation to genetics; lack of guidelines; and patient inconsistency or indecisiveness in decision-making. The domain addressing providers' capabilities and limitations when determining the primary patient considered whose wishes to follow in terms of a disagreement as well as the obvious issue of who was the primary concern between parents and child and within the extended family.

The power of genetic providers extends into social definitions in Ettorre's research. Her premise is that genetics experts are storytellers who conjure ideologies through their knowledge of genetics, risk and cultural constructions of families and kinship (Ettorre 1999). She found that in order for genetic professionals to be successful storytellers, they had to utilize normalizing strategies, including genetic foundationalism. She argues that within foundationalism, experts are eugenic through presenting a

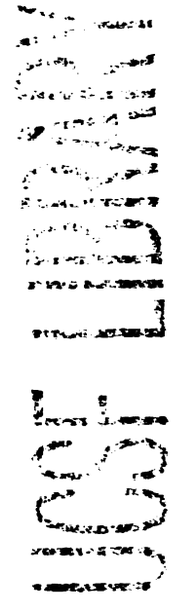
> genetics moral order in which one's genetic capital (i.e. genes) becomes a reproductive resource within the family. Simply, they want to priviledge a morality of the body which upholds the standard for conventional (i.e. non-diseased, genetically 'normal') offspring and society's need for citizens who are fit to be born (Ettorre 1999:50).

These genetic experts' judgements permeate society and encourage the belief that all illness has genetic components. Ettorre furthered her research when examining the organization of genetic work within the site of reproduction. She asserts "It is the belief among some physicians that the health of a community will only be ensured if the whole of the population comes within the range of genetic surveillance" (Ettorre 2002:59).

Kerr and colleagues (Kerr, Cunningham-Burley et al. 1998) delve into the eugenics issue. They interviewed clinicians and scientists working in the new genetics, which the authors define as involving "research into the genetic components to a range of

disease, illness, and behavior, and its application in the clinic in the form of testing, screening and treatment" (Kerr, Cunningham-Burley et al. 1998:194). They found that these British genetics researchers believed there were two main differences between eugenics and the new genetics: (1) new genetics is considered to be based on individual informed choice, not coercion; and (2) the new genetics is based on claims concerning a combination of nature (heredity) and nurture (environment) as opposed to nature only.

Heath is an anthropologist who conducted field work in the lab that discovered the key protein in the genetic disease Marfan syndrome while simultaneously doing anthropological research (Heath 1998). Depending on the situation and the observer, many different interpretations and shifting meanings of Marfan syndrome are possible. Thus, Health attributes the beginning of the geneticization of the syndrome and the normalization of the phenotypic expression of the gene for Marfan syndrome to genetic scientists. Through studying members of an advocacy group for individuals with Marfan syndrome Heath found that individuals with Marfan syndrome want to be "ungeneticized" and viewed as individuals, rather than as phenotypic representations of a particular cluster of genes.

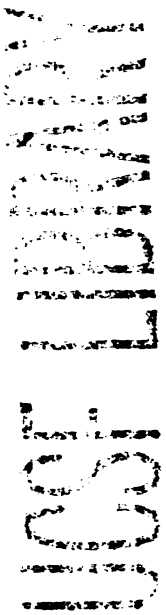


The position of genetic care providers is one of importance to my argument in this dissertation both about the geneticization of society and its consequences for bodies and families. The ethics of genetic care providers is not one ethic but multiple, dependent, as one would expect, upon gender, culture and other social factors (Wertz and Fletcher 1998). Genetic care providers maintain their positions as purveyors of genetic information by maintaining ownership over genetic knowledge, among other things (Ettorre 1999). Genetic capital, one's genes, is shaped as a resource with which to

ılate a "morality of the body" (Ettorre 1999) and normalize what is fit to be born. c ists try to distinguish themselves from the eugenics of old through their ethic of ed choice and an emphasis on nature (genes) and nurture (environment), while ppear oblivious to their geneticization of bodies and families by labeling traits and ors as "genetic" (Kerr, Cunningham-Burley et al. 1998).

usions On The Literature And Its Relation To My Research

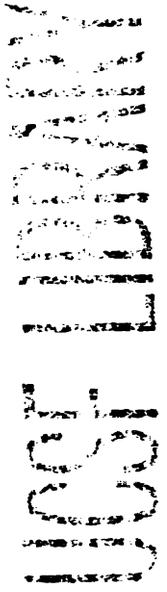
Throughout this extensive literature review related to social science, genetics, and families I have emphasized key concepts for understanding where my ch begins and how it builds upon and enhances understandings of women's and ies' experiences with prenatal genetic testing. The foundation of my argument in issertation is that through biomedicalization and geneticization, the routinization of is facilitating a shift in responsibility to pregnant women for making decisions about kinds of babies are acceptable in their individual families. The media coverage of ics overall, and prenatal genetics specifically, inevitably has some affect on the mination of genetic information and the perceptions of what genetics can do for ant women and their families.



Through the experience of PGT, pregnant women are given genetic information their fetuses, but also about themselves, their partners and their extended families. ant women's bodies are transformed into what I refer to as genetic bodies, dying the ambiguities and uncertainties of genetic knowledge while challenging ns of self as "normal." Pregnant women's genetic bodies have an intimate ction with their families, because of the reproduction of culture and mores through

procreation and the blood ties of shared genetics. When a family is exposed through genetic information, it becomes what I call a genetic family.

The very act of having PGT engenders anxiety in pregnant women, resulting for many in altered experiences of pregnancy. Some pregnant women suspend their commitment to the pregnancy until they have genetic knowledge about the fetus and accept it as a good "fit" for the family (Beeson 1984; Rothman 1986). PGT also causes anxiety through fear of miscarriage, waiting for results, and worry about the decision about abortion if there is something wrong. If something adverse is detected through PGT, women have to decide if it is something they can deal with in their future child, or if they cannot. The stigma of genetic disease, and even genetic knowledge, often influences these decisions. If it is not something that fits with the family, the only option for the woman is abortion. Rapp's (1999) conception of pregnant women who have PGT as "moral pioneers" is particularly relevant to my examination. While I believe pregnant women are conflicted about the choices they make, they are also largely relieved on some level that they have access to the information that prompts such complex choices. These choices lead pregnant women to shape their families according to their personal preferences of what types of individuals "fit" within the existing familial parameters, thus shaping their genetic families.

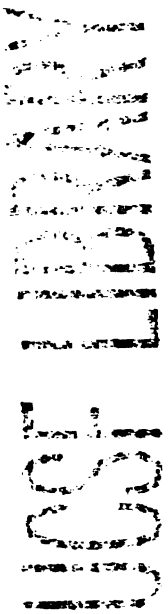


EXTENDING REFLEXIVITY

RESEARCH AND ME

ENETIC FAMILY

The issues of research and personal experience blur for me. My interest in
cs and families began while I was in college, studying biology. While enrolled in a
ed genetics course, my personal definition of family was given a genetic addition.
arents revealed that they had conceived a child when they were sixteen, and had
her up for adoption. She had recently contacted them because of a genetic
ion in her family that may have come from ours, her birth family, her genetic
y. Before telling my brother and me about my "new" genetic sister, my parents had
rgone carrier testing for the Robertsonian translocation for Down syndrome my
and her son carried. My sister had been made aware of the translocation when she
daughter with translocation Down syndrome. The genetic counselor she met with
eive the news that she was a carrier told her she should notify her birth mother
se she could also carry the translocation. Thus was the construction of my genetic
y. When my mother was determined to be a carrier of the translocation, my parents
ny brother and I and we too were tested. He and I both carry the translocation also.
ranslocation testing was done through a research study, so was free to us, and was
tached to our medical records in any way.



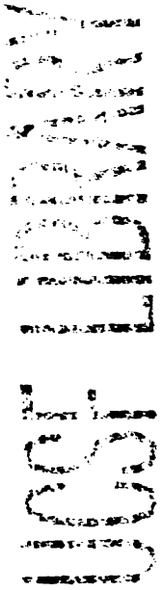
When I was told I carried the balanced translocation that made me at much higher
or having a child with Down Syndrome than any age-related risk, I was immediately
tated. I felt defective, broken and very different. The knowledge was a weight,
ng me think that other things must also be wrong with me if this hidden information

genes had been there all along and I had not know about it. I remember crying
ıy boyfriend at the time that I would never have normal children. It was a tad
tic, but the news that I had this thing inside me, as an essence of me according to
netics class, made me afraid.

At that point in my life, though, the changes in my family structure were more
ɔing to me in terms of everyday life than the genetic translocation. I could forget
something that was obvious to no one and was not relevant to my current
oning in society. But the appearance of a "new" sister was a shock. It was odd to
to terms with a blood relative who materialized out of nowhere when I was 19
old. The molecular bond that we shared was the link, as there was no other
non ground. Yet both my family and hers acted as though we should just transition
ıccepting each other as true "family" and move on as though nothing unusual or life-
ng had occurred. To somehow digest this, and as part of my fledgling career as a
cal sociologist, in 1996 I interviewed my family---mom, dad, brother, and new
--- for a symbolic interactionism class. I presented the resulting paper in 1999 at the
ic Sociological Association (Karlberg 1999) where I did not identify the family I
e of as my own. For me at that time, it was just too weird to admit that kind of
mation in public. The social pressure to have a "normal" family was enormous, so
ımily became me, my brother and my sister, even though my sister was not a sister
y of the traditional ways other than the genetic, blood relationship. The paper's
ise:

This is an empirical analysis of a nuclear family that has experienced a dramatic
ge in its self-definition based on movement from the social definition of a nuclear

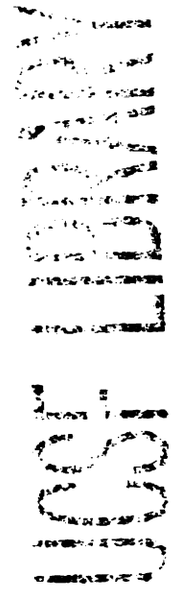
family to a new group identity based on molecular biological (genetic) information. It is a case study situated within broader cultural and technoscientific shifts from the nuclear age in the decades after World War II to the molecular genetics age which will dominate the next century. Within this shifting terrain of "family," individual narratives constructed by its members reveal sociocultural assumptions, themes and motives. There are two sites of disruption (Becker 1997) corresponding to this shift: "family" as married adults and nuclear family as disrupted by knowledge of a previously unknown (by my brother and me) *and/or unacknowledged* (by my parents) *child and secrecy of that individual; and dominant family definitions disrupted by enlisting genes as the foundation of relatedness, as opposed to life experienced together.*



The genetic knowledge I was now privy to, through this twist of genetic fate in my sister's life, that I carried a balanced translocation, was an anomaly in 1990 for a 20 year old. I knew no one who had any knowledge of his or her individual genetics, or even family genetics for that matter. My genetics professor's wife was a genetic counselor, and she drew my family tree for me—the first time I saw a medical pedigree. It was disturbing to hear of my 10-15% increased risk for Down syndrome, combined with my age-related risk, every time I conceived. At that point in my college career, I was more concerned with the social aspects of education and life. I was not thinking about having children at that point, so the new genetic knowledge was merely interesting to me, not debilitating in terms of my reproductive capacities.

It did, however, foster in me a new interest in genetics. I played with the idea of becoming a medical geneticist or a research scientist in genetic testing, but decided instead to go into social research. I pursued an MPH focused on women's health and

pregnancy, and then sought a position in social research that overlapped with genetics. I found a home at the Division of Research for an HMO on the West Coast. There I worked with a reproductive epidemiologist who was both a medical doctor and a Ph.D. in epidemiology as a project coordinator on two different projects related to prenatal genetics. In order to do my own research, I knew I needed to be a Ph.D., so I sought a program that would allow me to do something other than epidemiology. I was interested in more than statistics, and believed prenatal genetics involved social experiences on some level, I just was not sure how. I found a medical sociology doctoral program.



My genetic mutation was in the back of my mind in 1994 when I met someone I thought I might want to spend the rest of my life with. When we started talking about moving in together, I told him about the increased risk in my family for Down syndrome and we had a conversation about the lack of options other than abortion to deal with this knowledge during a pregnancy. I believed, based on the ongoing reading I was doing about prenatal genetic testing, that fetal cell sorting would be available by the time I was interested in getting pregnant, so I did not think I would be faced with the choice of having an abortion or a baby with Down syndrome. Again, the abstractness of the conversation made the genetic knowledge not as significant as it would later become when we decided to have children.

But I did start again to feel defective on some molecular level. This was reasserted when our commitment became more serious and at a dinner with his parents, we told them about my genetic history. I felt like I was confessing something, like I should have been able to control or alleviate this risk somehow. Knowing that when I did reproduce, I was at a much higher risk of having a Down syndrome child like my niece

was disturbing to me, especially after hearing from my "new" sister about the emotional, time-consuming struggles of surgeries, managing health issues and raising someone with special needs, despite the sweetness and pure love that came from her and contributed to their family. It did not seem to affect my future husband, however, and things moved along in our lives.

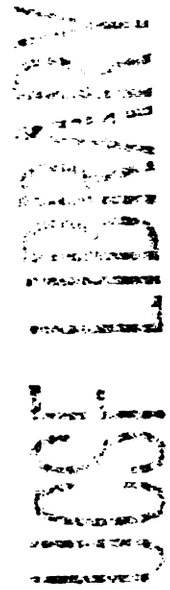
DOWN SYNDROME IN THE FAMILY: FACING MY FEARS

While researching and writing this dissertation, whose main questions are those I faced when coming to terms with my own genetic knowledge, we decided to have a child. Over the years we had abstractly spoken about what we would do when we pursued pregnancy. We had played through various scenarios and with little discussion had decided we would test as early as possible and abort a Down syndrome fetus. This was an experience I dreaded. Rob was more clear-cut about it being the only option. I was ambivalent about the choice, but knew I would not want to experience what my sister had told me in 1996 about the birth of her daughter who was then eight years old:

It was a Monday. She was born, and she was blue, so they took her away. We had her for five minutes and then they wanted to get her into the nursery and give her oxygen. They finished with me and I went into recovery and we were sitting there for about an hour maybe. Kurt was making all the phone calls telling everyone that we had had the baby and that she was good. Then the doctor came in, a different doctor, and he had her with him. He gave her back to me. He just said, "I want to point a few things out to you. Her ears are small and lower on her head than normal. The bridge of her nose is really flat." And I said, "Well, what does that mean? What is that indicative of?" He said, "We think she might have Down syndrome. She has a number of the

characteristics. The probability is very high, but we need to do a blood test to be sure. And we need to do a couple of other tests, like check her heart, because babies with Down Syndrome have more things wrong with them." We said that was fine. I don't remember what was said after that. We had her with us for about half an hour and then he came back and took her away. She came back later.

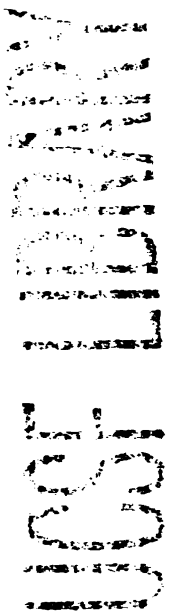
We didn't leave the hospital until Thursday because she had a high hematocrit and she had to have light therapy. The also had to take out her blood and put it back in twice, with synthetic plasma. They couldn't even get a sample because it would clot up into a little ball. We didn't find out until Friday that it was definitely Down's. They said it was translocation Down syndrome, which was pretty rare, and that could indicate that one of us would have the translocation too. That's the first we knew that people might be predisposed to having it.



When they found out she had translocation Down syndrome, they took blood from both of us that same day. I guess I didn't think that we would have it. You know, it's very rare, like 1 % have inherited translocation Down's, so I wasn't even thinking about it. It wasn't until right before I had to go back to work, so she was probably 5 weeks old, that we found out that I had the translocation. I remember that day and it was really bad. I just cried the rest of the day. I didn't stop crying when we were at the office. After she told me I just kept thinking, "I did this! I did this! Oh my god!" I was driving home and I was crying and I couldn't even see the road. I just kept looking over at her in her carseat going, "I'm sorry! I'm sorry! I'm sorry!" I couldn't even see. We had two cars, because we'd met there, so I had to drive home. That day was really bad.

Oh, and when we were at the genetic counselor's office when she told us I had the ocation, she did a family history. When she found out I was adopted, she said that I contact the adoption agency and tell them. That I should. She offered to do it if I want to. She said, "You really should do this." She didn't force me or anything, e stressed that I should do it. She said, "You would be helping them by telling " I just thought, "Well fine. It's not going to bother me."

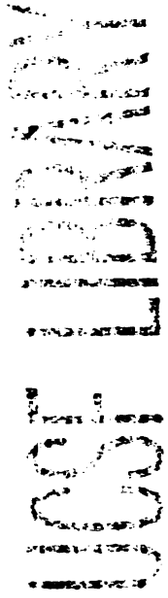
So we contacted your Mom right away, but I didn't' send anything back to you out a year. I couldn't handle it then. It took a year or more, about a year and a half, t everything wrong with her diagnosed and fixed, the stuff that could be fixed. And s what I was concentrating on then, during that first year. The first two years were y bad, as far as that. Plus, just adjusting to all of the different things was hard. It devastating to have this baby. I had this baby that I was carrying for nine months, hen I gave birth to this other baby, one I didn't know.



Hearing this in 1996, six years after meeting my "new" sister and her family for rst time because of the birth of this daughter, I remember feeling completely whelmed with grief at her description of what she and her husband went through. I married by then and although we were not talking about having children, we knew ventually wanted them. The fear I felt about having that experience was primal, and usband was also very worried. I also, in a strange way, felt grateful to my sister for ng out for us about the genetic "thing" in our family and contacting us about it. out that knowledge, her story could have been mine.

MY PREGNANCY AND CVS

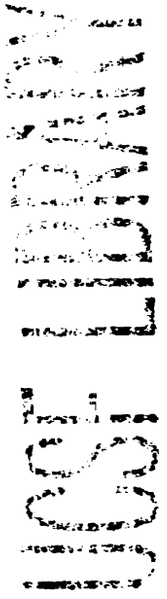
We moved to New York in 1998, and with Rob's job supporting us comfortably while I worked on this dissertation, we decided to get pregnant in the spring of 2000. Remarkably, we had no problem conceiving. I found out I was pregnant on July 23, 2000. Finding out I was pregnant for the first time ever was an awesome experience, full of trepidation, excitement, and a bit of outright fear. For both Rob and I the specter of genetic doom was always present when we thought about what we called "the pod." We very much exercised Beeson's (1984) "suspension of pregnancy" by not telling anyone right away, and eventually only telling our parents and a few friends before we knew for sure the "pod" did not have Down syndrome or another chromosomal abnormality.



I chose a maternal-fetal medicine specialist at one of the preeminent New York hospitals to do my CVS. He had trained with someone I met through my research who I respected and had seen conduct many PGT procedures. I have heard since from women who have gone to him that he is the "best." I felt like this man would know what to do. The entire first trimester of pregnancy was full of ups and downs in relation to my genetic status. We decided not to tell anyone other than our parents and best friends about the pregnancy—I needed a network of support, but couldn't handle telling the world if I then decided to abort a fetus when I truly wanted a baby. I chose not to have our insurance cover the procedure because I did not want to have the translocation in my medical record. In 2001, it cost $1,375.00 for us to have the ultrasounds required for dating the pregnancy, genetic counseling and CVS.

The genetic counseling session was exactly what I expected. The counselor, Toni, drew a family tree and discussed genetic risks. We were offered the panel of tests

for Jewish genetic diseases because my husband is Jewish, even though I am not. I noted that this was not the case in the HMO I worked for, because of cost containment issues. Our insurance would have covered it if we were using it. The money was not the reason we did not have the tests. I knew that the likelihood of both of us being a carrier of one of those disorders was very slim, so decided it was not necessary. We had to sign a release acknowledging that we were offered the tests and declined them. My researcher mind registered this as a way to avoid legal liability if our potential baby was born with one of these diseases. Rob was well-versed in what to expect from the session from the hundreds of conversations we had had about my work. I had at various times told him there was a 3% background risk of genetic disease in any pregnancy. He was very concerned by that number from the counselor, saying it seemed very high. I was so accustomed to it, it meant nothing to me. I was much more concerned with the considerably higher risk of a Down Syndrome fetus for us---around 10% plus my age-related risk. Our interpretations of the same issues ended with different concerns.



Because I did not want the insurance company to know of the translocation, I decided not to receive any prenatal care until after I had the results of the PGT. Mainly, this was to prevent any possibility of insurance discrimination in terms of a preexisting condition or other potential genetic discrimination[1]avoid being pregnant for prenatal care and then not being pregnant if I aborted the fetus because of a genetic diagnosis. It seemed too odd and convoluted, so I avoided the situation altogether until I was sure I was going to continue the pregnancy. I have never been a dreamer, but I had vivid, terrifying dreams of our dog and my husband dying throughout the first twelve weeks of

[1] For exploration of genetic discrimination see for example: (Bradby 1996; Shakespeare 1999; Geller 2002).

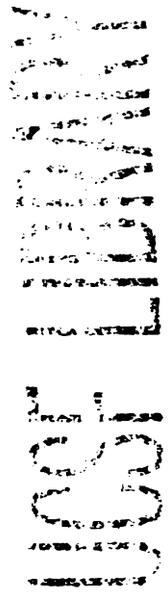
my pregnancy. I was certain this fetus would have Down syndrome and I would abort. But I desperately wanted that not to be true. I had the CVS on Tuesday September 4th, 2001. My journal entry from the sixth:

Rob drove us to the CVS appointment fairly early in the morning. I was really nervous and I cried in the car. I knew exactly what was coming and was terrified with the knowledge, but then strangely calm about the entire process. We had to wait quite a while in the waiting room, with women who looked really pregnant. I did not look pregnant at all. I know everyone was looking at everyone else, and I just kept thinking how thankful I was that I was doing this so early because these women looked REALLY pregnant, and no one could tell I was. In that room all of the women were pregnant, but at least the outside world didn't know I was. I went in knowing I wanted the procedure to be done transabdominally. I had a feeling that transcervical procedures have more miscarriages, because I had recently been reading medical literature for background information for the dissertation. I was going to request that he attempt it transabdominally if he could. But I was willing to be flexible to a point.

When I was called back to a room, I dressed in a gown and an ultrasound tech came in to gauge the position the fetus. I didn't want to see it, but I looked anyway. He confirmed the dating. He didn't turn on the noise of the heartbeat like they do in the normal ultrasounds, but I could see it beating, with the blood coursing around in there.

I could hear a couple talking in the hallway and she was crying because she had been in last week and wanted amnio and couldn't have it because of something. She was really upset that she wasn't in a room getting the testing now.

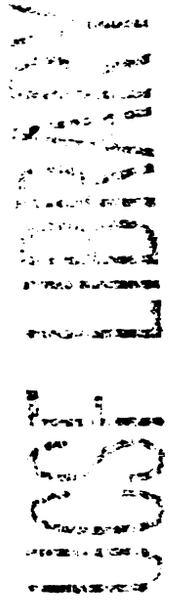
The ultrasound tech said my bladder was really full, something I was acutely aware of. The doctor came in and said he was going to do it trascervically. I said I would prefer it transabdominally, if he could. He told me to pee out a bit and then be checked again, and that he thought we could position it so it could be done my way. I must have gone back and forth to the bathroom 5 times. And was checked by ultrasound after each one. It felt so good to pee I could hardly stop it! By the time he was ready to do the procedure I had calmed down a lot. Rob wouldn't watch because the needle is really long. It hurt quite a bit, and the needle was inserted for an eternity. I thought while he was doing it that he was probing a bit more than necessary. I was at the point of asking him if there was something wrong when he took the needle out. The sample looked miniscule. But he inspected it and seemed satisfied there was enough there, so we were done. Very anticlimactic. Now we had to wait, up to 2 weeks.



Tuesday September 11, 2001---9/11--- occurred a week after I had the procedure. We were living in Brooklyn, close to the Manhattan and Brooklyn Bridges. It was too close to lower Manhattan on that day. I wrote about it in my journal on the 15th, the day after we received the results from the CVS.

Rob and I talked about the results, trying to figure out if there was any way this (the explosions of the World Trade Center) *would affect us getting them by the weekend. I broke down and called Toni, our genetic counselor, on Wednesday, and she said we would have the results by Friday. It rained on Thursday. There was sludge on all of our windows. The particulates in the air made sludge on the ground, on cars, and on windows. The sight of it made me cry. And the whole city smelled of burning.*

Rob's company was donating time and trucks and equipment to the recovery effort. He volunteered to drive a truck on Friday, picking up food from the restaurants and taking it down to what was designated Ground Zero. I decided rather than sit here all day and wait, I would go along. When we were at the warehouse, loading the equipment people had requested, I called Toni and told her to call us on the cell with the results. We went into the city. We were stopped at the midtown tunnel and searched. There were national guardsmen with machine guns positioned along all of the entrances to the city. Uptown, where all the fancy restaurants were that were donating food, things seemed remarkably normal. Traffic was a mess, and people were going to work and taking lunch and moving as quickly as they normally did. Then we drove by the armory in midtown. The entire block was plastered with paper—pictures of people missing and ways to contact their loved ones if you had seen them. I started crying. There were flags everywhere. Union Square was the cutoff for traffic, so that's where people put up memorials. The park was white with notes and flowers and papers.



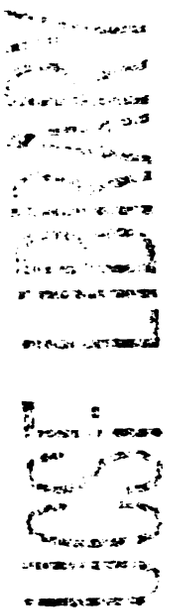
We were at a restaurant on the Upper East Side, parked in the street with me sitting in the cab by myself while Rob loaded the mountains of food when the genetic counselor called. I answered the phone and it was Toni. My heart started pounding and she quickly said everything was fine; there wasn't even the translocation. And we did not want to know the sex, right? "Right," I said. And I said, "Thank you." And that was it. I hung up. And then I cried. Rob was outside, so I got out of the truck and walked over to him sobbing and he said "What?" and I said, "Everything's okay." We hugged really hard and I cried a lot. The reality of the fact that I would stay pregnant seemed insignificant in the face of so many dying and the devastation that was our home. But I

was happy and so was Rob. I think we were happy. I know we were numb. I was almost 12 weeks pregnant.

After going through the emotional roller coaster that was prenatal genetic testing, the rest of my pregnancy seemed very easy, and I had a healthy child.

MY MISCARRIAGE

When we were pregnant the next time, I decided we would use the insurance, because I thought everything would be okay. I found out I was pregnant on December 13, 2003. The process of making appointments and getting ultrasounds was much easier with a referral in hand from my obstetrician. With my first pregnancy, my focus was on the Down syndrome and not on the risk of miscarriage. It never entered my mind that I would miscarry with the first pregnancy. The second pregnancy felt very different from the beginning. I wrote about it almost daily in my journal. The December 17 entry:



I called Toni and left a message. She called back very quickly. She pulled our file and reminded me that we did not have the Jewish panel, and asked again if we'd like that or the CF screening test. I declined. Then, after I've hung up with her, I wrote a little and started to freak out about the risks of CF and Gaucher's disease and Canavan and Tay-Sachs. I emailed Rob and relaxed a little because it really is ridiculous to worry about that. And I know it's going to be fine.

And December 20:

The timing couldn't be more perfect, actually. I can finish the dissertation, still not be too pregnant to fly, have the baby late August, surviving the heat with my new house's central AC, and then go to Susie's wedding in late October with a new baby, but

not too new. So it is truly ideal. I can't believe how perfect it is. And I desperately want it to work out. Please don't let there be anything wrong! And yet I find myself thinking it is too perfect, and therefore it will probably be a DS girl and we'll have an abortion. And then next year we'll have a boy. That's me for you, always the worst-case scenario. I would be happy if it's a healthy boy, but I would really like a girl. Be careful what you wish for, I know.

Monday December 29,2003:

Well, I know I'm pregnant because I keep thinking Owen is dead. The whole time I was pregnant with Owen I kept dreaming during the night and day that Rob or the dog or mom and dad or someone was dead or dying. And I dreamed that the baby I was carrying was a girl with Down syndrome so we had to abort. Nice thoughts while I'm on our holiday trip for Christmas with my family.

Monday January 5,2004

I had a dream sometime last week I forgot to record. I dreamed that the baby was chromosomally normal, but when it was born it had ambiguous genetailia.

We moved from an apartment in Brooklyn to a 100-year-old farmhouse in Westchester County on January 17th. The move was uneventful, but because we were new to the area, I had not interviewed doctors or researched anything. I had a few names to call from friends of friends who lived near where we moved, but had met none of them.

Friday January 23rd

About 3pm I noticed a splurt come out of me. Wasn't really concerned as I'd been dripping pee since I found out I was pregnant. But when I went to the bathroom

there was brown blood dried in the pantyliner. My heart started beating really fast, but I rationalized it could just be spotting which is not unusual. I called Rob and read the book on pregnancy. I decided to call Dr. Fuchs (old obstetrician). *Her partner was on call, and told me exactly what the book did: if I experienced heavy bleeding or cramping I should go the emergency room. Otherwise just monitor the bleeding and make an appointment with the doctor. I DON'T HAVE A DOCTOR!! I have an appointment for 3 weeks after the CVS I have next Wednesday, but I don't have one* **now**. *So I decided to just wait until Saturday a.m. and reevaluate. But the blood when I wiped was pink/red, so I know there's something going on.*

Saturday the 24th

We woke to 45 degrees inside. It's freezing! Owen was shaking when I picked him up from the crib. The boiler was off. Rob couldn't make it restart. He called the boiler people. When I went to the bathroom, there is still no blood in the urine, but there are tiny little clots in the bottom of the toilet. I freaked out a bit, and then calmed myself down and decided I'd better just go in. I told myself that it was probably a miscarriage because of a chromosomal defect and therefore better that it happen this way than that we have to wait to find out from CVS. Did I really believe this? I'm not sure. But I had myself convinced as I left the freezing house that it was okay if it was a miscarriage. I don't think I believed that it was.

Rob stayed home with Owen and waited for the boiler people to come. I got to the emergency room and there was no one waiting, and a nurse helped me immediately. I was in a room in about 10 minutes, after all of the initial paperwork was filled out. I was thankful I was there rather than in Brooklyn when I had to do this. I explained that I had

no doctor, had just moved here, and my husband was at home with our child. I felt alone, but strong. The doctor examined me and told me my cervix was still closed, a good sign. They ordered blood work and an ultrasound. They told me I have to be full for the ultrasound and started an IV. All the while, I'm thinking how strong I am to be here by myself and how good it is that we can handle this.

I had an ultrasound. The woman came in just for me. She was young and very nice. She tried to do it abdominally and failed. Then she did it transcervically, having me insert the wand thing. I thought this was crazy, but it worked quite well. She was very quiet while she was looking at the screen. She commented that my uterus was retroverted, which I've heard before, but other than that, said nothing. She called in a radiologist to check because she was having a hard time with the orientation. They both were very quiet. I asked if they could tell me what they saw. He (the radiologist) said they needed to finish the whole exam before they can say. My heart beat kind of fast, but I was still thinking things could be okay. Then he said, " Well, it looks like there is no heartbeat, and the fetus is measuring about 8 weeks, so it looks like it expired a couple of weeks ago. These things are usually because there is an anomaly in the fetus." When he started talking I started crying and couldn't stop. It was embarrassing and painful. I was overwhelmed with disappointment and hurt and sadness and helplessness and loneliness. I wanted Rob. The radiologist left, and the tech patted me and helped me calm down. She let me use her phone to call Rob. I was crying and he knew right away what it was. He was great. I can't remember what he said, but the invisible dog fence people had come for their appointment, so he found out we were no longer technically pregnant while dealing with a sales call. The boiler people had informed him that the gauge on the

oil tank was broken and we'd just run out of oil. They filled the boiler and the house was warm. One problem solved.

I went back to my ER room and waited for the ER doctor to call my new doctor who I'd never met and find out what to do next. Dr. Tessler called and was lovely. She said I probably needed a D&C and would try to schedule it today. She would call the ER and call me back. She did, and the operating room wasn't open so I had to wait until Monday, when Dr. Eng would do the procedure. I should call if I started bleeding heavily or had severe cramping, but other than that, just show up at the hospital on Monday a.m. with no food in me.

I drove home dry-eyed, still not really believing it, I think. When I saw Rob, he was talking to the dog fence woman, so I just picked up Owen and we went upstairs to play. Rob came up when he was finished with her and hugged me and I cried a little. I just couldn't let go. I think I thought maybe it was all a mistake, a dream of some kind like all the daydreams I'd been having recently of bad things happening. I still wasn't really bleeding, but was having mild cramping.

Sunday the 25th

When I woke I had to pee. There was a lot more blood. And I couldn't flush the toilet. It wouldn't go. And then I couldn't wash my hands. Rob had just gotten up and taken Owen upstairs to play in the playroom. I walked up there crying, and said, "I hate to do this to you but we have no water." I lost it, sobbing while poor Owen just looked on, trying to figure out why I was so sad. I just said mommy was sad because she didn't feel good, and that a hug from Owen and Daddy would make me feel better.

Rob started calling people to tackle the water situation. Everyone said they couldn't help because it was probably the pipe from the well, and that we needed a welder who could shock it. Rob's parents came up because of the water and the fact that I was having more cramping. I just didn't want to have to go to the bathroom and have to keep track of Owen in the midst of it. Thank god they were willing to come out. They brought water and bagels and a chocolate chip muffin. The water was of course the most needed, but also the comfort food. I stayed in bed.

At around 11:30 Rob got some guys to say they would assemble their team and come to us. We felt relieved, and hopeful about getting water. My bleeding was getting worse, and I just lay in bed all day. I was glad I had the DaVinci Code to read, as it was a thriller with lots of interesting information, and it kept me distracted. The cramping would get really bad and then subside. I actually broke down and took Tylenol at one point. I was really disturbed by the fact that every time I went to the bathroom there was more blood and we couldn't flush the toilets. It was truly awful. I kept thinking how all this blood was going to be a baby at some point and now it wasn't. I was really freaked out.

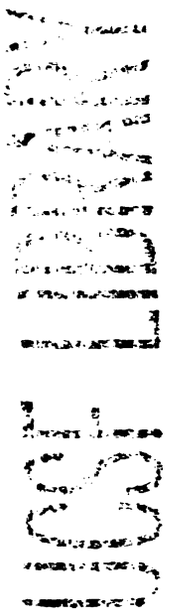
While we were eating dinner I got some really bad cramps and felt something slip out. I knew it had to be bad, so I went upstairs to my bathroom where only I had been going all day long. I felt it come out, and then had to dig around to find it. I put it on some tissue and laid it in the sink. It was the sack, still with fluid in it and the tiny—about black-eyed pea sized—embryo attached to a clump of red goo. It made me cry, but it was also kind of scientifically interesting that it would come out looking so completely perfect. There were even little dots for eyes on it. Everyone looked at it, and

we talked about what an odd day it was, kind of removed from the situation. I think Rob was a little freaked out by it. It made it real for him and he was very sad.

By 7ish we had someone else here, an arc welder guy who was willing to try to shock the pipes between our house and the well. At 11:30 we had water again.

Monday 26th

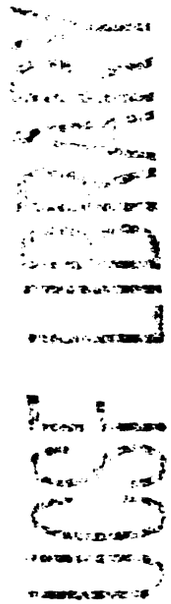
I slept fitfully, truly scared about having to have the D&C. It just seemed crazy to have surgery after such a loss. We got up and drove to the ER. Rob and Owen dropped me off and went to get Chyrel (babysitter) *and then Rob was to come back and get me. He wanted to be with me before and after the surgery—they use general anesthesia and I'm a little freaked out about it.*



Again, I was alone in the hospital. I got into the ambulatory surgery area fine. And got all the paperwork done okay. And then they sent me down to ultrasound pretty much right away. They were still very nice, and I didn't wait more than 5 minutes before I was in on the table. The tech was surprised by the retroverted uterus, and then she too wanted to call in the radiologist. The radiologist this time was a woman, and she had much better bedside manner than the male I had on Saturday. They looked together at the screen and then started talking to me about the fact that I had a bicornuate uterus. And it looked like there was a septum there. They suggested that I have a hystosalpinogram with an MRI to confirm this. While they're telling me this I'm just tearing up again, as this is yet one more thing to deal with in the pregnancy arena. I felt very alone, again. And on top of that, there was still blood pulsing through the uterus, so I had to have the D&C.

After the ultrasound I went back up to ambulatory surgery and tried to call Oxford insurance to change the provider and make sure they knew about the ER visit. I was on the phone with them when they called to take my vitals and move me to a room. I started crying again, just feeling overwhelmed with the whole mess of our lives right now. And frustrated that things hadn't happened differently, somehow. I don't know what would have been better, but overall the weekend was really crappy. I called Rob and was really crying then. He was dealing with the plumber and would come as soon as he could for the surgery.

Dr. Eng came in to talk to me pretty soon after that and was as nice as Dr. Tessler. I had an IV and blood drawn and then Rob showed up. I cried again and Dr. Eng came back in to go over things again and update us on times. I was to go in at 11. I encouraged Rob to go home and deal with the plumber more and then come back to get me. He left and soon after I went in. It seemed like seconds and I woke up. After that I felt very tired, but okay. And sad. Very, very, very sad.



Rob took me home. Chyrel had planned to stay the night because she and Owen were to go into the city for a party on Tuesday. They forecast tons of snow, so they probably wouldn't go, but she agreed to stay anyway. It was a godsend that she did. I was wiped out to the point that I basically slept that night and into the next day.

Tuesday 27th

Chyrel took Owen from me right after he woke and I went back to bed for a while. I felt very tired, and again, very sad. It was a hard day, as I kept thinking about the embryo just sitting in my makeup drawer. It was too much to deal with it today, and Rob was at work for the first time, so I just tried to keep it out of my mind.

Wednesday 28th

I cried this morning for so many reasons, not the least of which is the fact that we still haven't buried the embryo. I just want to put it in the backyard and have a place to plant a tree or something. I know it's weird, but I just couldn't flush it down the toilet. Again, when I was crying this morning, Owen said, "Mommy sad," and I said I wouldn't be sad much longer.

Finally, I felt ready to find a place for embryo. I just don't know what to call it. It's an embryo, but I had placed such potential in it. I'm glad we decided not to have pathology done. It would have hurt more to know what it was—or could have been. So I chose a tree that I can see from the kitchen, one that is young, but looks strong. And I had Rob and Owen come down from the playroom and watch me truck out there through the 15 or so inches of snow—I was really huffing. I carried it in my glass root beer mug from when I was a kid. I put some water in with the fluid so none of it would stick. And I cried as I was walking down. When I cleared all the snow away, I just poured it in and buried it back in the snow. It will be our tree of hope now, for much better things to come. And when I feel sad about the missed birth, I can look out there and feel a little better. Rob was great. I waved at them just before I buried it and really started crying. He met me at the door and held me while I cried. Even if he's not the one who carries it, I know he feels very similar feelings to me of loss and disappointment.

2 February, Monday

I am feeling an overwhelming sense of loss. I've been so sad lately, missing what might have been. I just feel like I missed out on what should have been, like I've been cheated. I was at the mall on Sunday with Mom and there are babies and pregnant

women and maternity stores, all of it working against me trying to pretend everything is normal. It's all just very, very sad. It was such a perfect time and place and year to have a baby, and now we won't.

WHY INCLUDE *MY* GENETIC STORY?

The pervasiveness of genetics in my narratives about my new sister, prenatal genetic testing, and miscarriage lends itself to my argument about biomedicalization and geneticization of American society broadly, and pregnancy more specifically. As a medical researcher, I have access to an extensive body of literature on genetic risk and statistics about cause of miscarriage. Yet is difficult for me to incorporate what I know of the social constructions of genetics and pregnancy into my embodied experiences of pregnancy, those of CVS and miscarriage. I attribute both to genetic knowledge and leave little room for other explanations. I flow with the biomedical paradigm and seem not to challenge it. My fears of genetic abnormality are similar to those of women I have spoken with who have no knowledge of genetic disease in their family or their partner's family. I rationalize that at least I have an excuse.

When I analyze this writing as a social scientist, I am tempted to categorize myself as a technocratic birth woman (Davis-Floyd 1994), one who consumes any technology available during pregnancy. But I know I am not that. I probe into the workings of prenatal genetic testing for just this reason. The choices I made about reproduction were made in part because of the knowledge I had available to me: my genetic translocation, its incumbent risk, the testing available to determine the genetics of my fetus, and the option of abortion if Down syndrome was present.

I also know that the translocation Down syndrome my niece has is severe, requiring open heart surgery, possible reconstruction of ear-nose-throat connections, and constant health monitoring by a pediatric cardiologist, pediatric ENT, and other specialists including speech pathologists and teaching aids in school. My exposure to Down syndrome has convinced me that the way the family must work around it is too much responsibility for the person with Down's, and takes a lot away from the children without it. I know my sister and her husband would *not* have aborted my niece. She is their child and an integral part of their family. What they did do was have another child to help their older son care for my niece in the event of their deaths. And they had CVS with their third child. I know they contemplated abortion with the third pregnancy if the fetus had been diagnosed with Down syndrome.

The purpose of my story is to show that my experiences have given me a deeply personal knowledge about genetic information in families. I know now that prenatal genetic testing is a complex, multifaceted, difficult, life-altering choice. And that families are the ones who live with the choices, whatever they are. This is useful for my research because I know the pitfalls of thinking you know what will happen and what the real story is. What I've learned in my life is that you never really know anything, and only by asking and waiting will you find out. That is what I hoped to do with this research, and I think I accomplished it.

4 MY RESEARCH METHODS

THE QUALITATIVE PARADIGM AND QUALITATIVE METHODS

This research is a multi-sited ethnography (Marcus 1995; Gupta and Ferguson 1997), utilizing research from different sources and locations to present a picture of manifestations of prenatal genetic testing in America today. Rapp's (1999:120) approach was to examine "the many pieces of a cultural puzzle" through empirical localization and studying "all over the place" within the designated locale. Understanding the local manifestation is crucial, as interpretations vary across temporal, geographic and cultural boundaries. The multiple interpretations of prenatal genetic testing technologies by those who administer and utilize the technologies, as well as the social and biomedical constructions of the testing itself, are testimony to the heterogeneity of interactions in the creation and continuing modification of this type of genetic technology.

The pieces of the puzzle I have assembled here as key to understanding genetic bodies and genetic families enabled and shaped through prenatal genetic testing technologies are many. They include peer-reviewed biomedical journal articles, literature produced by specific advocacy groups, conferences where medical investigators exchange information and interpretations, and lived experiences of those who administer, counsel and undergo prenatal genetic testing, as well as submersion in the local pregnancy experience in Brooklyn and Manhattan. In true feminist fashion, the personal is political, and the personal is also sociological. My interest in this topic is due to personal experience with a genetic mutation in my family, as discussed in Chapter 3, and this research project is interwoven with my self-reflexive search for a better understanding and interpretation of embodied knowledge.

THE INEVITABLE MERGING OF DATA AND SELF: PERSONAL EXPERIENCES

The methodological advantages and disadvantages of personal involvement with the research topic have been examined extensively by feminst researchers and center on the issues of reflexivity, distinguishing insider/outsider status, and voice, among others. Many researchers are, as I am in this study, a convoluted hybrid or insider/outsider. Naryan (Narayan 1997):23-24 emphasis in original) in response to these issues suggested the "*enactment of hybridity* in our texts; that is writing that depicts authors as minimally bicultural in terms of belonging simultaneously to the world of engaged scholarship and the world of everyday life." Even if the research enacts hybridity, the voices of all of the participants in the research process, especially the respondents, are still framed through the researcher. The expression of voice is a challenge even if one does not have personal experience as a lens of expression as I do.

My research inquiry was designed to capture the prenatal genetic testing experiences of women and genetic care providers, representing their voices while attempting to create a research text that will speak to and reflect upon the voices of my dissertation committee and the social worlds of academia, genetic medicine, and pregnancy in America. I reveal myself in the writing through my selection of conversations cited as "data" (Fine 1992). Fine (Fine 1994):72) suggests that researchers "work the hyphen" by "prob(ing) how we are in relation with the contexts we study and with our informants, understanding that we are all multiple in those relations." My positions as feminist, genetic epidemiological researcher, sociological researcher, graduate student, pregnant woman, genetic body, daughter, sister, wife, mother, member

of a genetic family, friend, colleague, and stressed out graduate student all factor into the experiences conveyed in this research. I also am constantly aware of the multiple locations of the informants in this research as well. In baring my personal stories about genetics and families and pregnancies, I am attempting to avoid the appearance of an "innocent" ethnographer/researcher (Rosaldo 1989).

My personal experiences with genetic knowledge, the creation of a genetic family, pregnancy, CVS, and miscarriage caused me to be particularly aware of my research questions and their interacting and overlapping complexities. My position as genetic researcher when speaking with the genetic care providers, and as a pregnant women who had had CVS when speaking with pregnant women, gave me a type of insider status with my informants. Riessman (Riessman 1993) refers to this relationship, recognizing the importance of the underlying propositions that make talk sensible, including what is taken for granted by speaker and listener. Here, my proximity to biomedical genetic knowledge was useful with genetic care providers, and my personal experiences with CVS, pregnancy and miscarriage allowed me to directly empathize with pregnant women. Other social scientists who have personal experience with their research topics (Becker 1990; Ellis 1991; Ronai 1992; Naples 1995; Becker 1997; Rapp 1999) have also incorporated their personal experiences into their data or used self as data.

I chose not to tell the women or providers I was talking to about my own experience with prenatal genetic testing until after we had conducted the "official" interview. I thought this approach would serve to allow me to collect less influenced data, in that my informants would not color their responses to what they thought I wanted

to hear. Occasionally, in interviews arranged through personal contacts, these women or providers would have some rudimentary facts about my personal genetics, and I found that these interviews were more "real" in that both of us were aware of my history. I now regret that I did not tell more people about my own situation before we talked, just to make the interaction less structured by self and other, rather than by "us."

DATA SOURCES AND DATA COLLECTION

Participant Observation

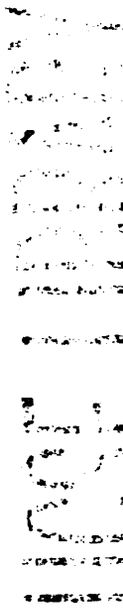
A large part of my ethnographic fieldwork involved immersion in the social worlds (Spradley 1980) of the experts involved in the genetics revolution around prenatal genetic testing and the pregnant women who choose to have it. In-depth observations and informal interviews were conducted at medical genetics conferences and professional organization conferences. Textual and visual materials (including scientific data presentations and promotional material from genetics research companies) were also collected at these conferences. Extensive fieldnotes were recorded during these conferences as well. Professional conferences are gathering places for perinatologists, medical geneticists, and genetic counselors among other medical and research professionals to learn about the newest technologies available. Please see Appendix F for a list of medical professional conferences attended throughout the study period. Through participant observation of clinical providers, an understanding was derived of how the caregivers---geneticists, genetic counselors, and perinatologists---frame their work and themselves construct genetic bodies and genetic families. This facet of the study describes the social and discursive construction of genetic entities, the ways in which the professionals in medical genetics as specialized providers enable the redefinition of

bodies and families through increasing availability of new genetic technologies in genetic medicine.

I collected most of the provider data while living on the West Coast of the United States. I attended seven HMO Genetics Department meetings lasting about two hours each. These meetings involved genetic counselors, at least one perinatologist and a medical geneticist in which they discussed ongoing clinical cases of both prenatal testing and childhood diagnoses related to genetics. I also sat in on three HMO interdepartmental meetings, each approximately one and one half hours long, with representatives from radiology, obstetrics, perinatology and genetics present. I attended 3 different genetic counseling informational meetings for pregnant women considering amniocentesis or chorionic villus sampling. I observed amnio and CVS being conducted at both a university hospital for a full clinic day (approximately 20 procedures) and an HMO clinic for two clinic days (approximately 60 procedures). I spent a week shadowing a medical geneticist in the prenatal diagnosis clinic of a university hospital. On the East Coast I observed the workings of a Division of Human Genetics at a metropolitan public teaching hospital and observed in the waiting rooms of a medical genetics department where women wait to have prenatal genetic testing procedures.

The other significant block of observational and participant observation data is from examination of the social worlds of pregnancy. My personal experience of pregnancy enabled me access through interacting with pregnant women in social, on-line, and organized meeting places to begin to parse the social and biomedical constructions of prenatal genetic testing in pregnancy. I observed pregnant women at birthing classes, prenatal yoga classes, and in the waiting rooms of obstetricians and the waiting rooms of

medical geneticists/genetic counselors. I attended classes on infant CPR, mommy and me yoga, Music Together, Music for Aardvarks, and signed up for playgroups organized by due date. For sixteen months I participated in a weekly playgroup of eight women and eight babies beginning when the babies were six weeks old. I observed other mothers' groups in social situations, at a regular coffee shop I frequented and talked with them informally about prenatal genetic testing. I monitored two online resources for pregnancy in the New York metropolitan area: the Urban Baby website for New York City and the Park Slope Parents Group for discussions relating to prenatal genetic testing. This immersion in the culture of pregnant women and recent mothers afforded me an intimate portrait of pregnancy and the experience of prenatal genetic testing in the New York metropolitan area.



Qualitative Interviews

I conducted open-ended qualitative interviews with genetic care providers and pregnant women. As part of an earlier study, thirteen semi-structured in depth interviews were conducted with genetic providers and researchers in the field of prenatal genetic testing. I obtained interviewees through work contacts while I was a project coordinator at an HMO's division of research, and through contacts made at conferences. Interviewees include perinatologists, medical geneticists, genetic counselors and research scientists, both medical doctors and master's level epidemiologists. I asked providers questions about their work, what they thought of prenatal genetic testing, how they believed it affected pregnant women and their perceptions of themselves, and how it affected the providers. The open-ended interview guides I used with both genetic care

providers and pregnant women are listed in Appendix E. I have published one paper based on parts of this data set (Karlberg 2000).

In order to paint a more complete picture of prenatal genetic testing in America today, I also conducted twenty-one interviews with pregnant women who had recently had prenatal genetic testing. I posted fliers throughout my Brooklyn neighborhood, at pediatrician and obstetrician offices, and at gathering places for pregnant women at strategic places throughout Manhattan and Brooklyn. I posted invitations to participate in my study on-line at Urban Baby New York and the Park Slope Parents Group and weekly added a synopsis of the flier into the message boards on these websites. Please see Appendix G for the flier. I responded to all women who showed interest, and screened them to find out if they had had prenatal genetic diagnostic testing, if they were still pregnant, and if they had received the results of the amnio or CVS. If a woman contacted me before she received the results, we scheduled a time to meet after she was notified.

Medical sociological literature supports the argument that patients and providers often have very different perceptions of a particular experience (Lupton 1996). Analyzing women's accounts of their prenatal genetic testing is a necessary means for understanding the constructions of genetic bodies and families and the subjectivities they produce. These interviews focused on eliciting responses that allow informants to discuss the subjective and embodied experiences of their desired pregnancies, potential genetic diagnosis of the fetus, and corresponding labeling/diagnosing/ reconstructing of their bodies as genetic bodies and their families as genetic.

RESEARCH INFORMANTS

Providers

The majority of the provider research took place at a managed care facility, a not-for-profit health maintenance organization (HMO) on the West Coast of the United States. Genetic services within the multi-facility HMO are coordinated by a group of medical geneticists who both provide genetic care and plan for future services by working with the HMO administration. My research was focused at one site, interacting with individuals mainly from the Genetics and Perinatology Departments. I also conducted some research at a metropolitan public teaching hospital on the East Coast, and a university hospital on the West Coast.

In total, I interviewed twelve genetic care providers: six genetic counselors, three medical geneticists, two perinatologists, and an obstetrician who performed prenatal diagnosis procedures. I also interviewed one master's level genetic researcher who had been trained to do genetic counseling and worked in the research division of the HMO. During three of the genetic counseling interviews, the taping malfunctioned and the data were lost. The sample I use for data analysis reflects ten transcribed interviews. The interviews ranged in length from twenty minutes to one hour and twenty minutes, and were conducted in the interviewee's offices or in a departmental conference room. The interviews were tape recorded and transcribed. The data were collected between November 1997 and April 2003.

Of the ten genetics professionals I interviewed, five were women and five were men. They ranged in age from 25 to 69, the male age range was 42-59 and the female age range was 25-69. I chose to allow self-identification of race/ethnicity of both providers and pregnant women. All of the people I interviewed were "American." There were eight providers who identified themselves as white, one who self-identified as Asian

Indian and one who refused to answer. Seven genetic care providers were married, one separated, one not married and one had a regular partner.

Pregnant Women

I conducted twenty-one interviews with women who were either pregnant or recently pregnant and had had some kind of prenatal genetic testing or screening. These women all lived in the New York metropolitan area and received care at New York City hospitals. One interview was not able to be transcribed because of excessive noise on the tape. The data analysis reflects twenty interviews. All of the women were either married or had male partners, and 18 had held professional jobs at some time. The other 2 did not provide employment information. In terms of self-described race/ethnicity, all the women were "American". I spoke with one women who self-identified as Asian, one who was Arab, one who was Greek, one who was Portugese, seven who were Caucasian/European, one who was Italian/Irish, one who was _ Mexican, _ Irish, _ Jewish, one who was Irish, and six who were Jewish. Some of the women provided more than one race/ethnicity identifier.

Sixteen women were pregnant when I spoke with them. Eleven were between 18 and 28 gestational weeks, and five women did not say what week they were when we spoke. One woman had had a miscarriage by the time we conducted the interview. Three women had already delivered their babies. The twenty women had 12 children among them, not counting the three women who had just delivered. The women had cumulatively experienced nine miscarriages. Two women had terminated previous pregnancies because of genetic abnormalities. I did not ask about abortions for reasons other than genetic indications.

Nine of the women were discussing with me their first viable pregnancy, meaning they had not carried another pregnancy to term. Two women were pregnant with twins, but one lost one embryo at about 10 weeks. The age range of the women was 31-47. Six of the twenty women were under the age of 35, the point at which women are required medically to be *offered* amniocentesis or chorionic villus sampling and all six were having prenatal genetic testing for the first time. One of those women had the testing because she would be 35 before delivery. One had testing to rule out cytomegalovirus, and one had genetic screening only. Of the remaining three women under 35, two had testing because of an abnormal ultrasound finding and one because of an abnormal serum screening result.

DATA ANALYSIS

The data, all interview transcripts and fieldnotes, were analyzed using the general principles of grounded theory (Glaser and Strauss 1967; Strauss and Corbin 1990; Strauss and Corbin 1997). Grounded theory focuses on uncovering the social processes and conditions that lie behind the phenomena in question, in this research, prenatal genetic testing. Constant comparison of data is central to the analysis, which allows code categories and analytic concepts to be compared to one another to examine the robustness and framing of the developing analytic model. Grounded theory offers systematic procedures that move beyond description into analysis, and through which substantive theories can be inductively developed. Analysis begins with data collection, and proceeds into a sequence starting with open coding---labeling and grouping concepts into categories. Those categories are then developed in terms of properties and dimensions, eventually forming the basis for constructing major concepts and determining the

conditions within which key phenomena unfold in different ways (Glaser and Strauss 1967). The idea of grounded theory as a transactional system was very useful to me in this research:

> All phenomena and their related action/interaction are embedded in sets of conditions. Action/interaction also lead to specifiable consequences. These in turn may become part of the relevant conditions that bear upon the next action/interactional sequence (Strauss and Corbin 1990:159-160).

Because my interviews with the providers took place well before I was able to speak with pregnant women, my analytic concepts about the ways providers structured their work in relation to the pregnant women they dealt with created through open and analytic coding helped me frame the types of questions I wanted to use to stimulate conversations with the pregnant women about their understandings not only of the biomedical constructions of PGT but also their personal and familial relationships to genetic testing. It also allowed me to more clearly pursue the idea of encouraging the providers to move beyond the biomedical constructions of prenatal genetic testing and convey their interpretations of pregnant women's emotional and physical experiences of PGT.

Codes and categories were developed around the formation of definitions and conceptualizations of genetic technologies in pregnancy, genetic bodies, emotions, families and how they affect or were affected by categorizations of, research on, and treatments for disability, pregnancy, and genetics in society. In particular, attention was paid to the specific content of the definitions of genetic technologies in pregnancies and how those conceptualizations prevalent in medical research and medical genetics practice during pregnancy are formed and used in pregnancy and obstetrical care. The way the pregnant women related to the identification of their fetuses as genetic entities to be

screened, themselves as genetic bodies, and their families as genetic families were revealed in the data. Strauss' (Strauss 1993) techniques for analyzing "trajectories" provided further assistance in this project, encouraging me to follow the genetic technologies through their beginnings to their present state as standard of care, as well as following the pregnant woman's experiences of prenatal genetic testing from when she first became aware of it during her first pregnancy until after she received the results for the current pregnancy.

In conjunction with grounded theory, as a part of the analysis I incorporated social worlds analysis. Social worlds characteristically have at least one primary activity, and division of labor, which generate ideologies. Social worlds are structural units within which the negotiated social order is itself constructed and reconstructed (Clarke 1990). The social world of genetic care providers is a production world, producing genetic knowledge. The pregnant woman's world is a communal world where the activities focus on establishing and maintaining a community of people committed to each other and a shared goal—a family. One cannot study a such worlds in isolation (Clarke 1991).

These two data analysis methods, both intimately linked with symbolic interactionism, allowed me to construct a map of the ways pregnant women and providers work together to achieve the goal of an acceptable genetic family for the individual pregnant woman. The data sources, including everything from television shows to articles from the *New England Jounal of Medicine* and my interviews with women, as varied as a music critic for a national magazine to a children's book illustrator, combined with interviews with genetic counselors who had been working only a year to a medical geneticist who was one of the first to have that title, provided unique combinations of

perspectives, types of data, and levels of education, experience, and exposure to PGT. These differences enhanced the constant comparative elements of grounded theory and enabled me to feel confident about the data analysis that emerged as the research project progressed.

VERIFICATION: TRIANGULATION FOR ADEQUACY AND CREDIBILITY

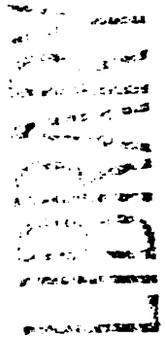
Olesen (Olesen 1994) argues that the corresponding parallels to validity in quantitative research are adequacy and credibility. Achieving this for my research entailed a multilevel approach. I first revealed myself as a genetic body, member of a genetic family, pregnant woman, feminist and qualitative researcher, and include that in the discussions of the data to some extent, incorporating a reflexivity and awareness to data analysis. Secondly, I follow an "across-method triangulation"(Denzin 1989) through combining dissimilar methods of data collection to "illuminate" the same phenomenon. This has some similarities to multi-site research. I not only interviewed and observed, I experienced, and I have been collecting anecdotal data on this subject for over thirteen years. My awareness of the personal meanings of the experience of prenatal genetic testing is something I am constantly aware of and I think that makes me a better researcher. I return to Olesen (Olesen 1994) who points out that the struggles between "intersubjective reporting in feminist qualitative research foreground ethical issues" which is where I turn next.

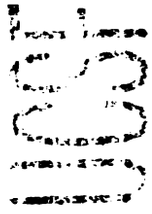
ETHICAL CONSIDERATIONS

The ethics of my research on prenatal genetic testing are less clear. A caveat that should be considered is that descriptions of observations and experiences are never value-neutral, and may well be discordant with the views of those being observed or observing

the same event (Emerson 1983). My goal of painting a picture of prenatal genetic testing in American society today through the experiences of pregnant women and genetic care providers failed. I could not collect interviews from a diverse population of pregnant women or providers. Most of my respondents are white, middle-class or upper-middle class, heterosexual, highly-educated people. I am such a person as well, and as such had easy entree with my informants as one of them, be it pregnant women or medical professionals.

However, my findings are not necessarily complimentary to either group, and would probably not be appreciated by some, because of my complicated and somewhat challenging assertion that people who participate in prenatal genetic testing are choosing babies to fit their families, shaping their genetic families.

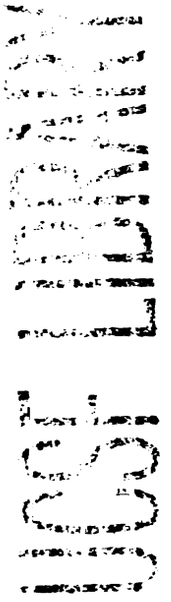




Everyone I interviewed was provided informed consent, and through this process made aware that some of the questions I asked could cause anxiety. That was the only potential harm of my research. No one refused any questions or stated they were uncomfortable with a particular question, with the sole exception of a medical geneticist who would not provide his race/ethnicity. I was studying "up" with the genetic care providers. The pregnant women, on the other hand, received information they might not have had during the conversations I had with them arranging and actually interviewing. Despite the fact that they knew I was not a medical practitioner of any kind, they often would ask my opinion or a question about genetics or obstetrics that I would answer if I could. Was it ethical for me to provide information to them about genetic testing or pregnancy care? I think so. I did nothing that I would not do with a friend or co-worker

who wanted to draw upon my knowledge and understanding of some pretty complicated issues.

In sum, this study relied on extended participant observation, ten in-depth interviews with genetic care providers and twenty in-depth interviews with pregnant women. While these numbers are relatively small, the data were carefully analyzed. The project suffers most from the limitations of a lack of diversity among both providers and pregnant women.



5 BIOMEDICALIZATION AND GENETICIZATION

CONSTRUCTIONS OF PRENATAL GENETIC TESTING

With all genetic testing I feel like we're on a train that's rolling down the track, and we just have to do the best we can. And that there's no possibility of stopping it. Clarissa~genetic counselor, 35, North American

Medicine is so different than any other industry, even though it's now a corporate industry, rather than the cottage industry it used to be. But you can never take medicine and standardize it like you can an assembly line. There is not a single fastest way to counsel a patient. Jacob~medical geneticist, 42, refused race/ethnicity

I think technology has kind of messed things up. Dane~perinatologist, 42, European

A caveat: This data analysis is conducted in relation to pregnant women and their experiences of prenatal genetic testing and screening. When I began this research I thought I could examine the providers and the women separately, but this was not possible once I had begun to analyze the data more critically. The providers' perceptions of the women and their experiences are very useful as a biomedically informed perspective on the same genre of interaction, even though the specific providers I interviewed did not personally provide medical care for the women I interviewed. Throughout, then, the analysis of interviews with women is interwoven with the analysis of interviews with providers.[1] All interviewees identified themselves as "American." The cited race/ethnicities are self-described ancestries.

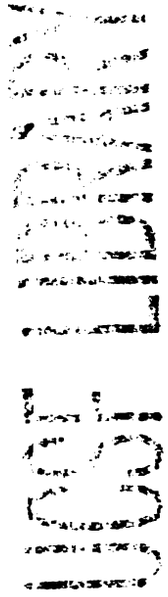
[1] None of the names used are the true names of my informants. I gave everyone I spoke to a pseudonym and used that throughout my analysis.

THE PLAYERS IN GENETIC MEDICINE

Genetic Care Providers[2]

Genetic counselors are masters' level trained in a program emphasizing molecular genetics and medical counseling. Most practicing genetic counselors are board certified.

> Genetic counselors work as members of a health care team, providing information and support to families that have members who have birth defects or genetic disorders, or who may be at risk for a variety of inherited conditions. They identify families at risk, interpret information about the disorder, analyze inheritance patterns and risks of recurrence, discuss the risks, benefits, and limitations of genetic testing, review available options with families and provide supportive counseling. They also serve as patient advocates, educators, administrators, researchers and resource people for health care professionals and the public(SLC 2003).



Clarissa, a genetic counselor told me, "I feel like we are definitely a resource. We are one of the last holdouts in medicine where we can still meet with people for an hour or longer. If they're really upset we can stay with them all day." Bosk (1992:xix) described *genetic counseling* as a "service" which "is generally a matter of transferring information to individuals alone to make the tragic choices based on that information." The profession of genetic counseling is nearly all female. Genetic counseling serves as an "auxiliary occupation, [a] deliberately fashioned lower status group" to genetics (Bucher 1988:143). The counselors do work that medical geneticists used to do. When that work became routine to the specialty, it was relegated to a subprofession-- genetic counseling: medical record chart review, initial meetings with patients, and family history taking.

Perinatologists are subspecialists who attended medical school, were granted a residency in obstetrics and gynecology and then pursued a subspecialty in perinatology.

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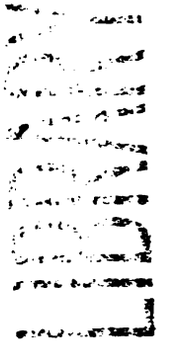
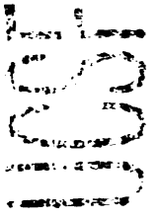
[2] This section builds on parts of my paper (Karlberg 2000) and is reprinted with revisions and additions with permission from Elsevier.

Board certification is available for this subspecialty. They are specifically trained to manage high-risk pregnancies, treating both mother and fetus until delivery. Some perinatologists are referred to as maternal-fetal specialists. Dane, a perinatologist, described the difference between his role and those of geneticists: "We take care of clinically what goes on on the labor floor while the [medical] geneticists deal with the cerebral issues of what the prognosis is, but they (medical geneticists) don't really participate in clinically managing the problem." The perinatologist's role in genetic care provision is actually performing the prenatal diagnosis procedures, amniocentesis and chorionic villus sampling. In the cases of "abnormal" results, they manage pregnancies if women choose to continue them. Perinatologists are trained to manage pregnancies, while genetic counselors and medical geneticists are trained to diagnose and manage genetic diseases.

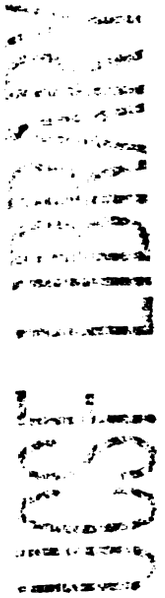




Medical geneticists are medical doctors who completed residencies in a specialty, such as pediatrics, internal medicine or obstetrics, and then chose a specialty in genetics. Most practicing medical geneticists are board certified. They diagnose and manage genetic diseases. Robert, a medical geneticist who has been in practice for thirty years described his job as, "not only challenging, but a tremendous variety of interesting things...so I can stay interested in what I do." Another medical geneticist, Jacob, called his practice, "very non-routine." The main role of the medical geneticists in prenatal diagnosis is the interpretation of results from the tests to make a diagnosis. They also provide management suggestions following the diagnosis, if desired by the pregnant woman, but do not directly provide pregnancy care for pregnant women.

PROVIDER DISCOURSES OF PRENATAL GENETIC MEDICINE[3]

I found two overarching discourses common to the practice of genetic medicine that are fundamental to understanding the ways providers actually practice genetic care and pregnant women receive such care: the ambiguous nature of genetic information and the omniscience of genetic knowledge. I identified these two discourses through my analysis of several kinds of data: participant observation at professional meetings (see Appendix F); participant observation at case study meetings at the HMO; following perinatologists on procedure days and observing their communication with colleagues, such as genetic counselors and medical geneticists; and the interviews with providers.



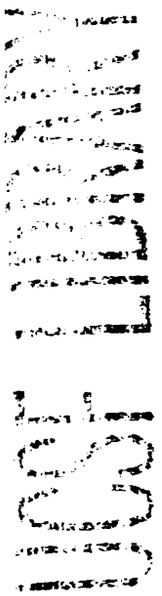
Genetic medicine is very much an emerging field, even though amniocentesis has been available since 1970. The genetic care providers I surveyed and observed doing their work are a professional, empathetic and highly specialized group of clinicians. Their work is evolving daily with discoveries of new genetic markers for particular predispositions, and pregnant women presenting with unique diagnoses not previously recorded. It is a challenging and life-altering field of medicine, and its discourses of ambiguity and omniscience run in contrast to each other, yet in tandem—yolked to each other in important ways. The challenge of ambiguity with the dominance of omniscience makes the experience for pregnant women a varied and anxiety producing one, as we shall see.

THE GRAY ZONE

I argue that one overarching ideology or discourse common to the prenatal genetic testing arena is the belief in the ambiguity of genetic information. This ambiguity is a

[3] This section builds on parts of my paper (Karlberg 2000) and is reprinted with revisions and additions with permission from Elsevier.

situated knowledge that impacts the shared work practices among medical geneticists, genetic counselors and perinatologists. Ambiguity is a "gray zone" of diagnosis, when there is a genetic "abnormality", but one not typified by a medical definition or known medical significance. Handling ambiguous information is part of the routine and challenge of practicing genetic medicine, including the ever-changing diagnosis and new findings. Clarissa, a genetic counselor told me, "*Most* of the areas in genetics are not absolutely lethal or absolutely fine." But ambiguity is frustrating for most patients, and can generate emotional turmoil for some. The providers I studied recognized this ambiguity and its impact on patients and incorporate handling it into their work ideologies.



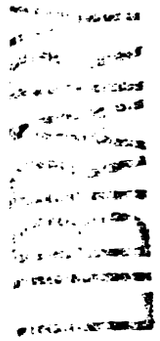
Jared, a perinatologist, noted the complexities of providing information that is subjectively interpreted:

> It's a very difficult situation for the patient. We're doing procedures a lot for women who choose to terminate when the prognosis is good and choose to continue when the prognosis is bad. I think a lot of times the patient doesn't understand, especially with Down Syndrome (DS). I think they may not know what they're getting into. I personally feel carrying a pregnancy should be a woman's or a couple's choice. I think it's more difficult when the pregnancy is not clear-cut, like when the father's not the father, or with surrogates. These are social issues too. There are a variety of things that can complicate genetic diagnoses. Jared~perinatologist, 42, white

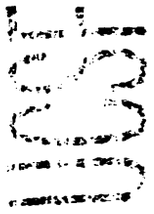
Most geneticists and genetic counselors understand and expect uncertainty and have considerable empathy for the women and their families who have to make life-altering decisions based on such tenuous results. Below Robert, Isabel and Jacob illustrate the life-altering impact of uncertain genetic information on pregnant women and their genetic families. Gertrude's focus is more on coping with genetic diagnoses, but she perceives this as challenging.

What is most difficult is seeing a child or a fetus where you know there is something wrong but you cannot make a diagnosis. You have to deal with the uncertainty of what that is. I think those gray zone cases where we can't give them a clear picture of what their child is going to be like are the most difficult. I can prove to the parents, or I can point out to the parents where it might even be better not to know the diagnosis. But it's still better probably in the long run to know the diagnosis; they just might not want to know what the eventual outcome will be. Robert~medical geneticist, 60, North American/European

I had a woman with an AFP of 10.5 and normal average is 1. And we couldn't say that this baby wouldn't make it to term or have some sort of syndrome that would make it mentally retarded. We had to really give her the range of outcomes in that sort of situation, and she had a really hard time with it. And I think part of it was the situation; it was just so ambiguous. But it was also her personality. She was very focused on finding answers. And she got many different opinions, but nobody could tell her the right thing to do. She wondered how we could not have the answer. And she ended up not continuing....She felt guilty about terminating, but she knew she did not want to deal with a baby that had multiple problems. We didn't have any guarantees that that would be the case either, so it was much more difficult. Isabel~genetic counselor, 25, Asian Indian



I think the other area that is very difficult is sex chromosome aneuploidies. Even though there is a literature, it's still hard. It's dealing with uncertainty about behavioral issues, learning issues, which most people who are having children are not thinking about. They're thinking about the "normal" child, whatever that means. They aren't thinking about the 20% of the United States with learning disabilities. They're not thinking about their family history of reading problems. I guess the hopes and dreams and everything depend on the immediate, as compared to trying to understand over a lifetime what can happen to people and what role genes play in that. Gertrude~medical geneticist, 69, white, Jewish



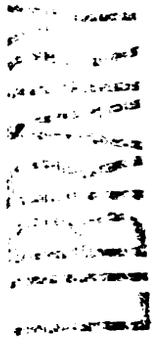
We see so many kids without a diagnosis, they sort of all lump together. Probably 50% of the born children with multiple anomaly conditions, about half of them we don't know what the cause of it was. And part of that is because the science is so young. Robert~medical geneticist, 60, North American/European

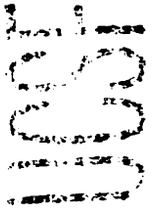
While ambiguous information is normal and expected, the professional interpretations made can be tenuous. This is evidenced by *unexpected* outcomes:

> We have the "hall of shame" where we hang pictures of all the babies of women we told their babies should be aborted and they [the babies] came out fine. Dane~perinatologist, 42, European

> I rarely take home my highly emotional, stressful cases. If I make a mistake, which I've made over the years, but not too often, I [do] take that home. [I've made] mistakes like missing a diagnosis, or maybe not ordering or interpreting lab work properly. I'm very good at turning that off as I walk out the door. The counselors tend to ruminate more and to allow that to affect their personal feelings and lives. Robert~medical geneticist, 60, North American/European

> I think that's just the style of genetics, period. That there is no black and white, and we of all people know that. It's scary sometimes to compare what we say to what happens. Not in what we say, because we do know to stay away from black and white, but in what happens: a lot of cases a baby will be born with things we didn't expect and without things we did. There's a good reason to be cautious. The problem is again that the burden falls on the family of having to make sense of that. Clarissa~genetic counselor, 35, North American





While Dane and Clarissa talk about ambiguities in genetic diagnoses that sometimes create medical mistakes as personal failures, they seem disturbed by their profession's uncertainties and how the gray zone affects pregnant women. Robert manages this proudly by attempting to leave his work at the office. While Robert's quote may seem callous, I observed that he was an incredibly sensitive, empathetic and comforting physician when dealing with his patients.

The gray zone is well recognized by genetic care providers on a professional level, as it is the nature of the care they provide. When the diagnosis falls within this zone, however, they expressed frustration and uneasiness with the capabilities of genetic knowledge to predict the outcomes of such gray zone diagnoses. The "hall of shame" created by Dane the perinatologist is a perfect expression of this ambiguity. My data

support the literature's assertion that providers tell women the information available to them, even if it is ambiguous (Wertz, 1993; Veach, Bartels, et al. 2001).

While providers acknowledge their limitations in terms of predicting the manifestations of a genetic diagnosis, they still cling to the idea that genetics is a universally true predictor of health.

THE OMNISCIENT GENE

The discourse of genetics as all-knowing also pervades the practice of genetic medicine. Among those providers and pregnant women I interviewed, the priority of genetic knowledge over the understandings of environmental influences on health was a routine undertone. I found the practice of genetic medicine does not allow for environmental influences to be recognized, evidenced by the power of the genetic diagnosis, regardless of how uncertain, to alter perceptions of pregnant women and fetuses. Nature, conceptualized as restricted to the actual genes and chromosomes of the fetus, was the basis of the diagnosis made. Environment as nurture was not generally allowed for or taken into account in the diagnosis. Not once during the meetings I observed was their any backing away from a genetic diagnosis to allow for the ways a fetus could develop differently due to environmental influences. While the providers seemed to acquiesce somewhat to environment by mentioning it, their explanations negate any substantial influence.

> Most frankly, everything that you do in medicine is either environmental or it's genetic or it's a combination thereof. It can't be anything else. If it wasn't for genetics, we wouldn't do family histories. It is relevant because if you look at the data, the best single predictor is genetics. You can tailor what you tell patients specific to what *their* needs are. Right now we tell a lot of people in society things that are frankly worthless to them. My family has no history of hypertension, so if you start telling me how I need to worry about cholesterol and my salt intake, I think that's crap. If it was specific genes it

> may become a much more real thing to do. Jacob~medical geneticist, 42, refused race/ethnicity
>
> I still believe there's as much to nurture as there is to nature. Our genetic make-up is only a part of who we are. Environment and nurturing is still a very strong part of who someone becomes. I'd like to think that what we learn in genetics will be helpful in treating diseases and understanding more about diseases and how to prevent them, but not in creating society that's based on genetic discrimination. Elaine~genetic counselor, 53, North American/European, Jewish

While Jacob appears to allow for environment in this thinking, he still says the "best" predictor of health is genetics. Elaine allows for nurture, and paints a realistic picture of what genetics is capable of doing for society, but throughout my interview with her she did not provide an example of environmental influence.

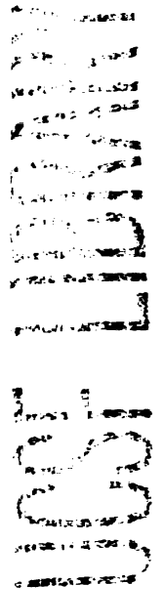
The belief that "genes-r-us" is not new, and while the primacy of genetics in medicine, does not blatantly exclude the possibility of environment, it does not incorporate it. I found that these two genetic discourses, the gray zone and omniscience, permeate the practice of genetic medicine.[4] This was true regardless of where the woman received her care, and what type of health care model was in place. My findings here also generally echo the extant literature.

For example, I believe this is a part of Nelkin and Lindee's (1995) argument regarding genetic essentialism, which conveys power to genes that encompasses far more than clinical genetics is capable of presently, or will ever be able to provide. Even more than that, Alper and Beckwith's (1995) genetic fatalism echoes omniscience of genetics with the assumption that a genetic association is deterministic and unchangeable.

[4] These ideologies are congruent with the research on the media by Petersen (2001) and Conrad(2002). Both argue that media portrayals of genes as omnipotent and genetic optimism shift the discursive focus from possible environmental or social considerations in "genetic" disease a naïve and unrealistic to the "one-gene-one-disease" model.

The all-knowing gene ideology is supported by Ettorre (1999) in research on genetic experts whose judgments included the belief that all illness has genetic components.

My data refute the assertion by Kerr and colleagues (1998) that new genetics is different from eugenics in that it allows for nature and nurture. These providers allowed for environment in tone, but in practice there was no environmental consideration taken. And Nelkin and Lindee's (1995) point that genetic essentialism erases ambiguity is strong support for my argument that while genetic care providers acknowledge the ambiguity of genetic medicine, genetics is still omniscient in the practice of genetic care.



PROVIDERS: GENETICIZATION AND THE MEDIA

While the pregnant women were focused on their experiences of biomedicalization and geneticization, the genetic care providers presented biomedicalization and geneticization from medical perspectives. They were more concerned with misinterpretations of the possibilities of genetic medicine, rather than the actual biomedicalizing of pregnancy itself.[5] The genetic professionals expressed concern over the misinterpretations and limitations of prenatal genetic testing, fed in part by media coverage of genetics. If news coverage of scientific breakthroughs is truly the way most medical professionals and lay people alike are informed (Caulfield and Bubela 2002; Conrad 2002; Voss 2002; Stamm, Williams et al. 2003), then genetics professionals are likely for many years to experience people coming to them for answers they cannot provide.

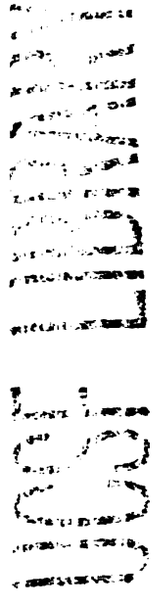
> God, the next 20 years are going to be horrible for us [as genetic care providers]. Because everyone is going to think we know everything. I think

[5] This biomedical interpretation of geneticization falls more in line with Hedgecoe's (2001) reinterpretation of Lippman's (1991) concept: that geneticization occurs when a particular condition is linked to specific DNA structures.

it's going to be really difficult trying to get people to understand that all this stuff in the media means nothing. You know it's really sort of sad because testing is one area where people think, "Oh, I can do some tests and I can get some control over my life and what's going on in my family." Often you can't do that. You have to help them in a different way. Isabel~genetic counselor, 25, Asian Indian

Right now genetic testing is very glamorous. It's in the media a lot. I think genetic testing is really high profile right now. I think there's a certain crystal ball aspect of this that people are very drawn to. And I think it's possible that it will just sort of fade away and it will become useful for people who are in these very high risk families, but it won't in fact be universally applied to people who aren't. That it won't in fact change the lives of people who aren't at extreme risk of certain things by virtue of their family history. Jane~genetic researcher, 32, European

Everybody thinks with all these breakthroughs that they read about and hear about with cloning and gene therapy and organ transplants that we can do more than we actually can. How many people are really going to get these things? Not very many. For instance, I saw a girl yesterday who is almost fully grown at 4 feet 8 inches and she read in "Seventeen" magazine about the use of growth hormone. So she came in to talk about that for her. The art of counseling comes in when you have to explain why that is not right for her. Robert~medical geneticist, 60, North American/European



These genetic professionals mention the media, as did five others I spoke with. Many of the providers referred to pregnant woman wanting "the" DNA test that incorporated testing for everything available. I have had many people anecdotally tell me they had this test, or planned to have it during pregnancy.[6] If it is possible for people to still believe that there is "A" test for genetic abnormalities, then the dissemination of information by the media is failing miserably. I do not hold the media wholly responsible for the genetic education of the public. However, if new technologies are more likely to be introduced to the public through media as the literature suggests (Patterson 1982; Rowley 1984;

[6] I tend to agree with the AMA's Council on Ethical and Judicial Affairs (1998:19) who believe that the media relays "misleading information to the public almost daily" rather than with Caulfield (2002) who found that scientists' belief that their work was being distorted was unfounded.

Singer, Corning et al. 1999), then the media does bear some responsibility as Singer (1999) alleges.

One genre that seems to be successful for the media is public interest stories relating to genetics.[7] But not all such media representations are valued by genetic care providers. Ironically, two of the genetic counselors described particular entertainment to me, outlining what they thought were outlandish approaches to creating families:

> Have you seen "GATTACA" [the movie] yet? Well, if you want to see the possible effect of prenatal diagnosis on society that's the movie to see. It's incredibly frightening if anyone thinks that could really happen. It's a society in which there's a way to manipulate conceptions and choose the best features of a couple so that they get the best possible offspring.
> Elaine~genetic counselor, 53, North American/European, Jewish

> This play, "The Twilight of the Gold's" was about this young couple and he was in a biotech firm who developed genetic tests. When she got pregnant they just tested for everything they could, and one gene was for homosexuality. Her brother was a homosexual, and the fetus carried the gene, and they decided to end the pregnancy. It was devastating to the whole family, the brother especially, but she was a mess at the end. If it gets to that point, I don't want to be a part of that kind of genetic testing.
> Clarissa~genetic counselor, 35, North American

While these specific approaches are not currently available, the distinctions the genetic counselors made about the differences between these futuristic prenatal genetic tests and those presently available are not readily apparent to me. In both these scenarios, as well as what happens in present day PGT, pregnant women have testing, make a judgment of the fetus after finding out its knowable genetic information, and make a decision about that fetus's fitness for their family.

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[7] Henderson (1999) argues that it is through these types of stories that people retained most useful information.

The ways providers approach genetic care are also not immune from personal emotional experiences of making these kinds of decisions about desired pregnancies. Providers also have questions about what kinds of impacts PGT as a pregnancy technology has on women and their families, and they problematize the possibilities of PGT in their musings.

> I once did a study where we asked pregnant women to draw themselves. We got to see their perceptions of the babies inside, after the ultrasound results were given. It made me understand what I was doing. I was really upsetting people. And I hate to admit this, but for all of us, technology does change your life! It doesn't have to be bad, but it has an impact. Gertrude~medical geneticist, 69, white, Jewish

> There will certainly be more and more things we can test for, that's true. But I find today that there are still many couples, more than I knew 15 years ago, who are opting to continue pregnancies with genetic abnormalities or opting to not have testing. They seem to want to return to "let nature take its course." Maybe this is a swing of the pendulum that's happening because of the increase in technology. There's a sort of resistance to it. Elaine~genetic counselor, 53, North American/European, Jewish

> I'm conflicted about the field of genetic research. I think that on the one hand it's incredibly important and incredibly exciting and at the same time we're moving very quickly. I think it's also very terrifying because of the idea of identifying somebody at risk for a disease that you can't do anything about. It's very disturbing. And I'm not sure that we have quite caught up with the technology yet. I'm not sure that the people aspect, the social aspect, has caught up with the technology. Jane~genetic researcher, 32, European

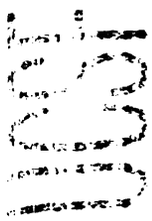
> If we could tell people specific genes that they had that caused them to be at risk for certain cancers, for instance, it may become much more applicable to those people's lives. The down side to that is then you disable people in ways which you may not really want to. That's one of the downsides to what we do in genetics. Jacob~medical geneticist, 42, refused

> If society is going to pay for those people that want prenatal genetic testing, then they should be part of the information gathering, voices that say that this is the direction that we really want things to happen. If we want, as a society, to have healthy children, if we want our society to look after children with all needs, how do we deal with this? We cannot offer these prenatal genetic technologies without that philosophical, ethical bed being considered first,

because we do not want people punished for not doing the testing.
Gertrude~medical geneticist, 69, white, Jewish

The providers I quote above all express concern about how pregnant women process the information available to them through PGT. Elaine even suggests that women are choosing to avoid the situation altogether, or to continue pregnancies even if they have an adverse diagnosis. Ethics are mentioned by Jane, Jacob and Gertrude, questioning the situations pregnant women as decision-makers are placed in with the genetic knowledge provided through PGT.

Throughout my ambitious literature review, I did not find descriptions such as these of how genetic care providers feel about the work they do and its impacts on pregnant women. The ways the providers I spoke with managed their work in their lives seemed to allow them to be more empathetic with the women for whom they provided care. They too experienced some of the anxiety, fear and confusion about genetic medicine, and thus could communicate more effectively with the pregnant women they served. They also had their own ethical questions and saw policy dilemmas in the genetic care situation today.



PREGNANT WOMEN: BIOMEDICALIZATION AND GENETICIZATION

I argue next that biomedicalization and geneticization of American society primes the social environment for geneticizing bodies and shaping the genetic family. I assert that biomedicalization (Clarke, Shim, et al. 2003), for my purposes the medicalizing of pregnancy through technoscientific means, is a mechanism through which geneticization –the attribution of genetic cause to disorders, behaviors and physiological variations to manage problems of health (Lippman 1991) is accomplished. Pregnancy is no longer a

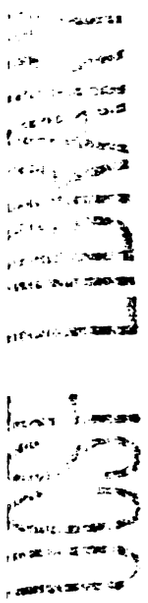
process experienced by women without medical intervention for most of the nine months.[8]

Biomedicalization and geneticization of pregnancy are essential to my argument that pregnant women are encouraged by the mere availability of prenatal genetic testing to self reflexively identify their pregnant bodies as genetic and make pregnancy decisions about termination and continuation of pregnancy resulting in genetic families based on genetic information provided through PGT. I argue that once pregnant women perceive themselves as genetic bodies, they desire the most attainable "normal, healthy" genetic bodies for their fetuses, attainable through shaping the type of fetus they give birth to. For many women, the option of shaping their families genetically is an enormous, challenging responsibility, regardless of whether they opt to terminate. The pregnant women who have PGT have consciously chosen to test the genetics of their potential family member. This "choice" is possible only through the availability of prenatal genetic testing technologies and the socially accepting climate of using them in America, tenable because of biomedicalization and geneticization. Novas and Rose (2000) suggest that society is now viewed through "molecular optics" where life itself is correlated with molecular/genetic conceptions. Rose (2001) argues that human existence is now molecularly biopolitical. PGT instantiates these assertions.

The decisions women make about aborting fetuses are dependent upon what technology can tell them about their potential child. "The way things used to be" women

[8] As described in Chapters 1 and 2, there is a prescribed pattern of prenatal care that now includes a battery of genetic screening and testing procedures recommended for different pregnant women based on risk factors predicted through identifiers such as age and genetic disease in the family (ACOG and AAP 1983; ACOG 1985).. The literature review in Chapter 2 outlined the ways normalization (Lock 1997) and routinization (Press and Browner 1997; Suter 2002) of prenatal genetic testing enables the biomedicalization and geneticization of pregnancy, which I argue paves the way for the creation of pregnant women's genetic bodies and the shaping of genetic families.

had no options other than to find out about the sex and health of their child at birth. The current state of prenatal genetic testing technologies lies somewhere between fate and design. There are presently three options available to pregnant women: 1) no testing and take what you get; 2) testing and know more about what you are getting; and 3) testing with the option of aborting a problematic fetus. One can also currently utilize in vitro fertilization (IVF) with preimplantation genetic diagnosis to implant only those embryos with screened genetics (see Appendix A). If the technologies continue to be developed for this market in the "brave new world" direction they are headed, there may be a day when a couple could choose the genes their child should have, create an embryo, and through IVF have the embryo implanted in the woman's uterus.



The perceived biomedicalizing and geneticizing of pregnancy through the cultural normalization and routinization of PGT was obvious when speaking with both genetic care providers and with pregnant women. The women were more specific about processes of the biomedicalization and geneticization of their experiences of pregnancy. Some women framed pregnancy as "natural" and PGT as disrupting naturalness, inserting science into what was once a woman's domain:

> The whole thing about having the baby, I mean, I know there's this kind of thing about Elizabeth Seton mentality about having the natural childbirth and you know, women have been doing this for however long human beings have been around and that kind of thing, but I think realistically, it's all a big science experience, and so the amnio is just kind of, to me, part of that continuum.— Nissa, 36, Mexican/Irish and Jewish

> Everything is so natural, just like what it's supposed to be, but then stepping into the medical realm and getting all these tests, you start to feel like you're poisoning the natural joy of it all with this clinical crap. It's like a medical procedure rather than a natural happening.—Rox, 38, Arab

Others decided that having prenatal genetic testing was *the* thing to do, so they had the tests. This does not meant to imply that these women believed they had succumbed to peer pressure. They experienced the normalizing and routinizing strategies of geneticization and just flowed with the socially accepted path.

> There is certainly a certain amount of peer pressure in the sense that everybody is doing it [having amnio], everybody you know is doing it pretty much.—Kate, 35, Jewish

> I think in this state [NY] and the medical world, it's just so common [to have amnio]. I know a million women now under 35 who are getting all this prenatal testing for a variety of reasons.—Jill, 47, Jewish

> I think all of the information that pregnant women get is very biased and very aimed at constructing a particular experience in pregnancy.—Hilary, 36, Irish

> I think pregnancy doesn't need to be this experience where you're just in the dark. I think the days of hoping for the best—it doesn't have to be like that. You can actually find out what's going on. There are so many things that aren't in your control, but there's a lot more that could be.—Tasha, 40, Caucasian

> I looked at it very comfortably. Things could happen. We need more information. The information is there. The technology is there so let's take advantage of it. They're not incredibly risky tests. For me, pregnancy came relatively easy, so the risk of miscarriage wasn't that threatening like it is for some people. The positives of having the tests done far outweigh any of the negatives.—Jennifer, 35, Asian

The themes of "peer pressure" and the "commonness" of having PGT are aspects of the routinizing of testing. The "control," "constructing a particular experience," and quest for "information" all function as facets of biomedicalization and geneticization through normalization. These women were informed, and felt they knew what they were getting themselves into, but in a Foucauldian way, they were normalized through "technologies of the self" (Foucault 1975; Foucault 1988; Balsamo 1996; Shildrick 1997; Sawicki 1999).

"THE WAY THINGS USED TO BE"

While biomedicalization and geneticization permeated conversations I had with both genetic professionals and pregnant women, there was also a romantic nostalgia expressed by both for "the way things used to be" with pregnancy. Many wished that there was not the option of PGT or interventions because it made things easier to just take what one got. They would rather play the genetic lottery as opposed to deciding if a genetic disease diagnosed prenatally is something the family can cope with and accordingly shaping the family through abortion. Elaine, the genetic counselor quoted in the section above, said she felt women were rebelling against technology somewhat by not having testing and not aborting fetuses with adverse genetic diagnoses. I found the providers' perspectives on this much more telling than the pregnant women's because I assumed that people who provide these services must think they are good or they would not do the work they do. But my interviews revealed that the situation is more complicated than that.

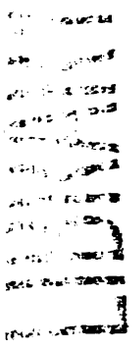
> I actually don't think prenatal testing is a good thing. I think it would be better if there wasn't this kind of testing. I think that you kind of alter how society works and functions. I think technology has kind of messed things up. Many of these babies would have just died, but now technologically they're able to intervene and keep them alive. I kind of think it was simpler and maybe more beneficial when we didn't have the technology to keep sick kids alive and then the technology to interrupt pregnancies that were going to have certain problems. Dane~perinatologist, 42, European

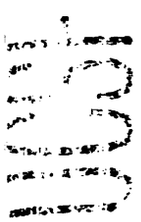
> What's that great quote, something like, "The decision of the group is better than the decisions of individuals for people who live in that group." I myself kind of miss the idea of having a more family group. I think we lose out on a lot by being so individualized. Clarissa~genetic counselor, 35, North American

> Fifty or one hundred years ago you basically accepted what came. The higher percentage of people accepted what life gave them. Not today. And

there's a lot of people today who want to take control of it. Jacob~medical geneticist, 42, refused race/ethnicity

Dane is a perinatologist who spent over ten years training to complete his subspecialty, and yet he is conflicted about the work that he does everyday. He takes care of those women who are at highest risk for genetic problems, preterm delivery and other fears of pregnancy. The irony in Clarissa's quote is that genetic counseling is focused on the individual---the decisions pregnant women make are to be independent of everything but the correct information about the genetics of the pregnancy. Not even the opinions of the counselors are supposed to influence the women's decisions, and yet Clarissa wishes society had more say about what happens in PGT than the individual women she works with daily. Jacob laments women's wishes for control in their pregnancies, not acknowledging or realizing that his diagnosing power for genetic disorders is the supreme exercising of control over a particular situation. He also echoes Clarke's (1995) argument that there are now enhanced social/cultural desires for control in American culture.





The way these providers framed PGT reveals the complexities of personal feelings intermingling with professional experience. The individual focus of pregnant women on the fetus they are carrying and its "fitness" combined with the technologies available today allow decisions to be made that were not even imaginable forty years ago. The desire for control echoed in these quotes is one that permeated the discussions I had with pregnant women as well as genetic care providers. If the information and technologies are there, many people feel compelled to take that advantage and utilize it. Interestingly, the providers of the care realize and appreciate that desire, despite their ambivalence about its utility and ethicality in the society in which we live.

The pregnant women were much less existential in their reflections on "the way things used to be." They were more focused on the difficulty of the decisions they were forced to make because of the availability of PGT, and how that made them feel that there was no alternative but to have testing.[9] As the providers above noted, the mere existence of PGT meant to some of the pregnant women that they were obliged to have testing, just because it was available.

> It's sad the way we live now, that we're forced to be subjected to these emotional tests. Whereas, in our mother's day, we didn't have any of these tests and they didn't think twice about their pregnancies. Maybe there's something to be said for that. Maybe some of these children deserve to come into the world whether they're perfect or not.—Susan, 33, Italian/Irish

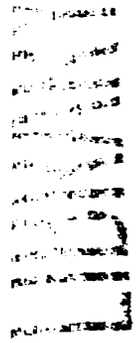
> I feel more confident because we have the tests available to us, but I also feel less confident. I feel that women thirty years ago before all these tests must have had to relax. They had nine months, and they just had to have faith. The more testing I have done, the more I feel that they do it because they can...I think the nice thing about pregnancy is despite all these tests, you're still basically in control of your body and it's your body. There's nothing you can really do. It's times like when I had the amnio when you feel helpless. When you're just lying there thinking, am I having something done that I'm not sure about? That's when you feel like you're kind of losing control or giving up control.—Meg, 39, Northern European

> Everything now about having a baby, you know, people have been doing this since the beginning of mankind, and it's like you just rolled with whatever happened. It happened. The human race has gone on for better or for worse. It's like all this stuff, this fetal monitoring and all this stuff is just making everybody more insane about it. You know? And in some ways is it putting them at risk more because people are so crazy about it? And then there's also the "perfect baby" thing. If my baby is not going to be perfect should I terminate now? Who gives you the right to make that decision? I don't want to be that all-powerful. It's a weird see-saw.—Rox, 38, Arab

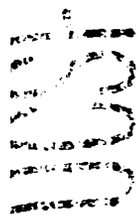
[9] These women's feelings support the literature saying that the mere offer of prenatal genetic testing is an endorsement of the practice, thus legitimating it and encouraging pregnant women to have PGT (Clarke 1991; Johnson and Bresinger 2000; Bennett 2001).

Susan laments that the technology *is* available, suggesting that abortion of genetic disorders is not necessarily the answer to adverse diagnoses. She was the only pro-life woman I spoke with. Rox and Meg both talk about control, and how testing is not "real control" because of the hard decisions that come with the diagnoses. The anxieties these women feel about the normalization, routinization, biomedicalization and geneticization of PGT is apparent from these quotes.

These women and others I spoke with were not really questioning the utility of the testing, more they were challenging the usefulness of the application of a genetic diagnosis to their lives. Most of them worried about the decisions they would make regarding abortion in the event of an adverse diagnosis, a conundrum I discuss in depth in chapter 8.



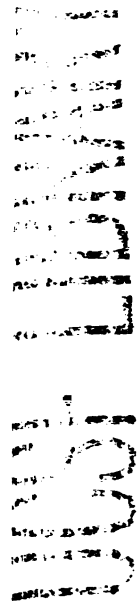
Given the existence of prenatal genetic testing in today's American consumer-driven culture, in love with all things technical, biomedicalization and geneticization are socially legitimated, mediated and enabled. Those navigating the social worlds of PGT, especially genetic care providers and pregnant women, seem especially aware of the social construction of pregnancy as biomedicalized and geneticized. Because the culture is moving and changing in the technological frame anyway, these movements within the experience of pregnancy and prenatal genetic care are accepted with little challenge by most who move within these worlds.



6 SHE SAID, S/HE SAID

WOMEN'S AND PROVIDERS' PERCEPTIONS OF PRENATAL GENETIC CARE

While providers understand to some extent the emotional immobilization PGT can engender in pregnancies, they believe they must approach their work in very specific ways. The work providers do informing women about the testing, conducting the testing and notifying women of the results is widely practiced, but *how* this work is carried out varies by training of provider, setting, and personal preference.[1] In this chapter I examine the actual practices of providing genetic care from the perspectives of providers and pregnant women. First, the practice of genetic medicine is outlined from my data findings that genetic care providers assess pregnant women and tailor the information they provide to them based on that assessment. Then I discuss the pervasive nature of anxiety in prenatal genetic testing, first through the pregnant women's descriptions and then through providers' understandings and experiences. I conclude with an examination of the mode of practice of non-directiveness, challenging its possibilities and using data to show that some women experienced being directed to have PGT in their genetic care experience.



THE PRACTICE OF GENETIC MEDICINE: WHAT PROVIDERS BELIEVE THEY DO[2]

Assessing the pregnant woman

[1] See these authors for different perspectives on specific types of genetic care providers: (Bosk 1992; Michie, Bron et al. 1997; Salkoviskis, Dennis et al. 1999; Johnson and Brensinger 2000; NSGC 2003).
[2] An earlier version of this section was published in part (Karlberg 2000) and is here with permission from Elsevier.

My data showed that some providers assess pregnant women on different elements to decide how to approach the genetic medicine they practice (Karlberg 2000). In their views, assessing the woman enables the providers to practice more "competent" care. There were five main elements about the pregnant women that the genetic care providers I studied suggested were useful to their assessments: 1) knowledge level about genetic topics; 2) apparent comprehension of the information about the testing available for prenatal diagnosis; 3) whether a pro-choice or anti-abortion (pro-life) stance is favored; 4) the comfort level in dealing with uncertainty and ambiguity; and 5) a broad sketch of the pregnant woman's personality type. In most cases, the assessment is very tacitly and informally performed by genetic providers in their initial visits with pregnant women by asking them questions and observing overall attitudes. Different providers also had different assessments of the same woman. Much of the interpretation was based on social position, including such variables as education level, religiosity and other relative measures. Clarissa, a genetic counselor, noted that one woman she met with brought her pastor's wife with her to the meeting and was clasping a Bible on her lap throughout the session. To this genetic counselor, her observations meant the woman was not going to consider abortion if there was an adverse diagnosis. Another genetic counselor, Elaine, provided an example of a woman dealing with different variables to make a PGT decision:

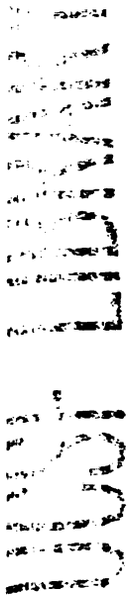
> A couple came in and they were very ambivalent about having an amnio—they were very worried about the risk of miscarriage. I just remember meeting with them for a long time and laying out the pros and cons and helping them to think in several different ways to determine which would be harder for them. This was a couple who really though they would not continue a pregnancy that had a serious birth defect. They had also had years of infertility and this was a very wanted pregnancy. She finally decided to have an amnio, and she did miscarry. Those kind of things stick with you.

> And you wonder how she weighed it out and was she second guessing the decision she made?—Elaine~genetic counselor, 53, North American/European, Jewish

A medical geneticist described her assessment approach:

> An atmosphere of neutrality is what I'm striving for. If I can. And I'm just trying to understand the people, where they're coming from, what's on their minds. Where they're coming from may be places I don't know much about. Gertrude~medical geneticist, 69, white, Jewish

The patient assessment, examining conditions and contingencies that can affect the course of the interaction, is a complex process involving the genetic care providers ordering their work around the ambiguities and uncertainties of genetic diagnoses. The extensive list of issues thought to impact how a woman receives the information about genetic testing or the results from such testing qualifies the structure of the provider/patient interaction. The ambiguity of test results makes this work task extremely important because of the uncertainty which must be conveyed to most patients with a genetic diagnosis.



Tailoring the information to the pregnant woman

Once the assessment of the pregnant woman has been conducted, the information relevant to that woman's case is often tailored by genetic care providers based on the providers' feelings and pieces of personal information gleaned from previous interactions. Providers actually anticipate being able to tailor the information about a prenatal diagnosis procedure being considered or a genetic disease or birth defect being diagnosed.

> Going [in to meet with] a woman who's carrying a baby with a problem, just the knowledge of whether she was considering abortion or not changes how I approach the situation. Having that information ahead of time makes it easier to develop an approach, a strategy. Now I've been fooled before, and I've had

> to change in midstream when I've realized that I've made the wrong decision or got the wrong information, and I've misread the patient. And I have to sort of back-pedal and start all over again. Robert~medical geneticist, 60, North American/European

Tailoring the information involves presenting the options available, such as the various tests, abortion, adoption, and the multiple ways of managing the pregnancy, in a specific way to fit the individual pregnant woman's needs.

> There's so many differences [in the patients] because there's differences in diagnoses, there's differences in family's attitudes, there's difference in couple's attitudes, in couple's relationships to their pregnancy, relationships to feelings about abortion, about the types of decisions they would make. Elaine~genetic counselor, 53, North American/European, Jewish

> We try very hard to take a 3 generation pedigree line and really tailor make this, and have them understand what the testing is about. A lot of people think they're having their DNA analyzed for every gene. Gertrude~medical geneticist, 69, white, Jewish

Such tailoring of the information may also include a specialized description of the implicated disease. A disease description could highlight the positive or negative outcomes associated with the disease while minimizing the opposite. Full disclosure of diagnostic information was favored by the providers. They overwhelmingly stated that their policy was to not withhold any information.[3]

> I believe patients should be told as much as we know. I don't withhold information. Dane~perinatologist, 42, European

> Our policy...is that they're gonna know everything that we know about their child or child to be whether it's good or bad. Jacob~medical geneticist, 42, refused race/ethnicity

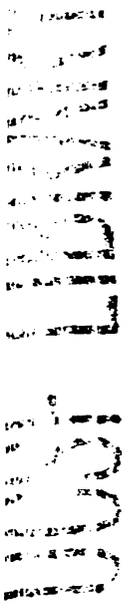
> I don't ever try to protect my patients. I always tell them the hardest news. Because I want them to see if they can deal with the worst case scenario. Because if they can deal with that they can deal with anything. So that's

[3] My findings of work practice of delivering all the information to the pregnant woman is also supported by the literature on provider disclosure (Wertz 1993; Wertz 1993a; Veach, Bartels et al. 2001).

> usually a choice. It's not difficult to know what to say, it's more difficult to know how to say it. Isabel~genetic counselor, 25, Asian Indian

However, while the genetic care providers believed they were being forthcoming with diagnosis information, how they represented this information work as "tailoring" reveals that in subtle ways the information a patient receives is not "all" of the information available. Jacob, a medical geneticist, explains his tailoring approach:[4]

> ...I try to make a judgment call. I'm probably right more than I'm wrong. But it's still a judgment call about what sort of information they need... So it's possible that somebody, two different families with children having the same condition may get different spins based upon what their perceived needs are...If somebody's in denial to the point of that creating perpetual problems, ... I may swing some verbally, to try and get them to come to the reality that you can't deny this, that your child has this...The other parent may be so focused on the negative side that I'll spin very positive....So the way I say it will sound very different. But I'm trying to bring them to what I think is the correct place--something that's intellectually correct. Jacob~medical geneticist, 42, refused race/ethnicity



This "intellectually correct" position is, of course, very subjective, but a common goal of medical geneticists and perinatologists. The "correct" position is founded on the assessment of the particular provider of the pregnant woman and her partner, so it may differ based on who is the diagnosing medical geneticist.

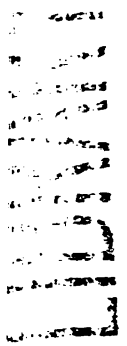
The ambiguity of diagnoses and the impossibility of a guarantee of a "perfect baby" even with the "normal" genetic testing results further *destabilize* the concept of a "firm" diagnosis of test results. Genetic counselors problematize their approach to the situation:

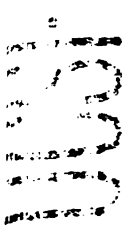
> The possibility that I always grapple with is that we give people too much information that they may not be prepared to deal with, and that sounds a

[4]This medical geneticist worked mainly with children who were diagnosed with a genetic disease after birth, rather than fetuses diagnosed in utero.

> little patronizing and I don't mean it that way. One of the points of counseling people, especially before prenatal procedures, is getting some sense of what this might mean to them. To get an idea so that that can just help you frame things. When you have anything with gray information, because it *is* gray so you do have to try and play both sides, but more so because people are looking for it to be black and white. No one wants to be in a gray area. So they're going to take out of that [information] *one* version. That's human nature to make sense. No one wants to deal with mushy, gray, ambiguous information. Clarissa~genetic counselor, 35, North American

The process of presenting the information in a patient-specific manner is the epitome of negotiation in this arena. The genetic care providers' jobs change depending upon the pregnant women they are dealing with, as the skills required vary with each individual. I argue that all of the social worlds in the prenatal genetic testing arena affect the outcome of the negotiation, as the assessment of the pregnant woman is altered based on the pregnant woman herself, the providers and each of their beliefs, feelings and ideologies/discourses which are shaped by their work, professional, home and social environments.[5]





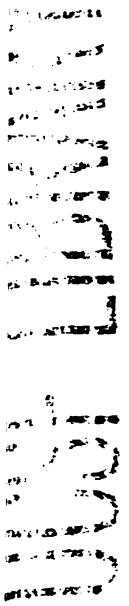
While my data found providers were filtering the information to individual pregnant women based on what those providers believed was relevant for them to know to make the "intellectually correct" decision, Ettorre (1999,2002) focused on the agenda of genetic medicine at large. She (2002) found that some genetic care providers believe the only way to ensure society's health is to subject everyone to genetic surveillance-genetic screening. The first step in this approach would be to have every pregnancy screened prenatally. The biomedicalization and geneticization of pregnancy and the resulting routinization and normalization of prenatal genetic testing technologies supports this position by making it difficult to avoid genetic surveillance of pregnancy. This

[5] These findings are supported by the literature (e.g. Bernhardt and Bannerman 1982; Ritchey, Yoels et al. 1995; Bernhardt, Geller et al. 1998; Heckerling, Verp et al. 1998; Egan, Kaminsky et al. 2002). Ettorre's (1999, 2002) work is particularly relevant here.

social pressure to have prenatal genetic testing and screening breeds anxiety in already "on-guard" pregnant women.

CONSTRUCTING THE GENETIC FAMILY TREE

For many pregnant women and their families, a genetic counseling session is the first time the thought of passing down ill health affects takes on shape and consequences. Vaguely, people know that what runs in families can be transferred to future generations, but the specifics of genetics are complex and unavailable to most. The counseling session includes the genetic counselor or medical geneticist asking the woman and her partner questions about immediate family members, branching out to include three generations back in the maternal and paternal lines to construct a medical pedigree. Thirty-five percent of the women I talked with had specific knowledge of genetic diseases in their families, and thirty-five percent of women also had friends who had had positive genetic screening or testing results. These experiences no doubt played a role in women's decision-making about PGT.

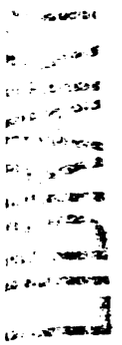


For many women I spoke with, this medical pedigree construction experience was disturbing in its connotations for their future child. Twenty percent of the women were factoring in fears of having a child with disabilities and the weight of responsibility that would place on their existing child/ren. But for most, the realization that genetic family histories have an impact on the future child's health was scary and anxiety-producing.

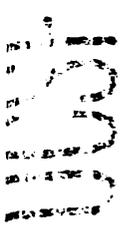
> The only thing that made me feel funny was she [the genetic counselor] has you go through the family tree, and my husband's family is a very traditional family. His parents are still married, he has two siblings, so there are two boys and a girl, very traditional. His dad's a doctor, his mom stayed at home. She was very educated through the whole thing and they're this very traditional nice Jewish family, you know. And my parents are younger...and they're divorced, and my dad was a substance abuser and has one child out of wedlock. So I have a half-brother, and you know, just like in going through

the family tree, it's like apples and oranges, which of course, we recognize. It's made me feel different, but it's just kind of really out there when that happens.—Nissa, 36, Mexican/Irish and Jewish

We already had a child and I just felt that if there was going to be some kind of serious problem, that there was more concern. It wasn't like it was going to affect me and my husband so much as that it would affect our other child. When we talked to the genetic counselor we were certainly aware that the risks were very real, but because we had this big genetic difference, I didn't know if we had very much risk. We realized we were not as healthy as we thought. Once you start thinking about your family tree, you realize there are a lot of people who have health issues. When you start thinking about your family in a clinical way, you realize you're at risk. Having to kind of parse out my mother's AD, my sister's MS, my dad's cancer, and you suddenly think, "Why are we reproducing?" And you're adding to it a multi-cultural mix. [Her husband is Jewish.] We keep telling ourselves this isn't really how it's supposed to work. This isn't how people are supposed to do things. It's a little unnerving to be thinking in those terms, "Well, I'm healthy and he's healthy, but she's not."—Hilary, 36, Irish



You just assume everything's going to be fine, so suddenly you say [after meeting with the genetic counselor], "Oh, I could be a carrier of this dreadful thing and pass this on to a child!" It's terrifying, you know?—Kate, 35, Jewish



When we sat down and did our family history it sort of made us both stop and think. You see how many people had high blood pressure. You see how many people had cancer. There is no breast cancer in any of our families. Until you really sit down and have someone lay that out for you, you don't really know, you don't really see what you have to watch out for.—Delilah, 36, Eastern European

These pregnant women brought up issues of culture, social definitions of family, and health. While Kate and Delilah mentioned fear and realization, Hilary and Nissa were more concerned with the immediate ramifications of the pedigree analysis. Nissa pointed out the stark differences between her family and her husband's family and expressed discomfort. Hilary, however, noted contemplatively that she and her husband were "not as healthy" as they thought they were and wondered, "why are we

reproducing?" The issues these women raised when constructing family pedigrees are far reaching, even beyond the scope of medical genetics.

While those with knowledge of genetic family histories found themselves anxious when the realization of the impact of those histories on the lives of their future children was thrust to the forefront, two women I spoke with had little or no knowledge of their family health histories. They were particularly concerned with the realization that this lack of knowledge could be transferred to their fetus and have health connotations for the future child.

> I think trying to figure out the family history with the genetic counselor, things like that always make me feel like a piece of my history is missing, any kind of medical background stuff. My dad passed away 13 years ago and we don't have any other relatives here. Everyone is in Baghdad right now. So I can't get that information. It's like there's a hole there. And then it also makes me feel like, you know, a "normal" American family documents these things, but mine didn't.[laugh] So I guess when you're trying to figure out how to put rice in your mouth you don't really worry about what diseases people have had. It kind of panics me when they ask questions like that, like they're doing a background check. And with my partner, it just made me really sad, and it was like he doesn't have *any* information. His dad just ditched out. So he doesn't have anything. And asking the questions about Down syndrome in the family, it's almost like rubbing a raw wound. In terms of him [the fetus] we're not going to be able to answer these questions. Because I don't have any information, it was like, "Oh god, he could have this." He's just not going to have a lot of information on that front. It kind of makes me proud in a certain way because it's going to be a wild card—whatever it is, it is.—Rox, 38, Arab

> Meeting with the genetic counselor brought a lingering concern to my mind, in that we are from Holocaust survivors on both sides. We have no history. That gives me cause for concern. And it brought back up an uneasiness that will always lay on me. Prior to having kids, or being married, it was never a big deal.—Ruby, 42, Jewish

It is often difficult for pregnant women and their partners to conjure family histories out of their limited knowledges of relatives they have little contact with.

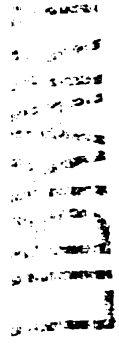
e genetic care providers have tools they incorporate when taking a medical

ligree to enable the pregnant woman to remember things she might otherwise

ve considered irrelevant. A medical geneticist described an outline of a family

digree to me:

> We review their family history. And we check off what's pertinent to the pregnancy and what other things we might need. We put down things like a family history of breast cancer. We take all of those elements, the information about her and her partner and their families in order to better understand what things they might be at risk for or are at risk for with relation to the pregnancy. We check to see if they have been carrier tested and found to be carriers. Did anyone tell them about the risk prognosis of carrying the gene for that disease? We check blood types. So the elements are who they are, and where they're coming from. Do they understand the task, the risk of the procedures versus the risk of whatever problems they are concerned about, plus the limitations of the tests? A lot of people think having genetic testing means they're having their DNA analyzed for every gene. We ask if there's anything special they think we should know about. Gertrude~medical geneticist, 69, white, Jewish



A genetic counselor told me a story that represents the lighter side of genetic medicine while also pointing out that people often have little understanding of the genetic implications for differences in family members.

> I was asking this woman who had her mother with her about a family history. She said, "Oh, there's nothing unusual. Everything's fine." And in this instance there was just something...every family member had one thing more bizarre than the next. Somebody was missing a leg, and somebody had a missing kidney and someone else had something. There was just birth defect after birth defect [laugh]. When we went through family member by family member she and her mother went into elaborate detail about testing that the people had had done, and it became so funny. It was the most amusing family history I've ever taken. There was nothing really that seemed significantly inherited from the family, but the stories—it just got into the kind of detail that became unbelievable. And she said, "No, everything's fine!" Elaine~genetic counselor, 53, North American/European, Jewish

Through the pedigree construction, pregnant women's families are made genetic families, linked by genetic traits they share and do not share with other family members

l their partners' families. Interpreting the medical pedigree as a boundary object, king different professional practices with external, internal, collective and individual ects of the body accomplishes the embodiment of genetic information as family ukaga and Cambrosio 1997; Atkinson, Parsons and Featherstone 2001).[6] As Rapp Rapp 1999):76 says, "Learning to think through medicine entails the recording of the ody: its ills, systemic connections, and intergenerational history all take on new and specialized meaning." Geneticizing family on this very personal level enables pregnant women to imagine the kind of family they would like to have, one with or without specific detectable genetic traits or disease. And through PGT, they can, to some extent, shape their genetic family.

ANXIETY: A RESULT OF PRENATAL GENETIC MEDICINE

Both genetic care providers and pregnant women believe PGT produces anxiety in pregnant women. Most women expressed feeling emotions of stress, anxiety and/or guilt. To counteract that, some sought enhanced control in a situation that was scary and unknown. Others combined these feelings with a quest for peace of mind. Whether specific anxieties about the test or general anxiety about PGT, there is little doubt most women who have prenatal genetic testing have some anxiety. These emotions are complex and bring with them the possibility of a variety of psychosocial and physical difficulties, such as problems functioning in everyday life and nausea.

> It's a level of stress that you probably don't need when you're pregnant.—Sotiria, 37, Greek

> I think for me the stress of not knowing would have caused more damage.—Jennifer, 35, Asian

[6] Nukaga and Cambrosio (1997) and Atkinson and colleagues (2001) coopt Star and Grisemer's (1989) concept of a "boundary object" as objects that are simultaneously shared by different parties, and often interpreted differently by them.

I think there might have been anxiety about simply not knowing.—Jill, 47, Jewish

ANXIETY About The Test Itself

Many women had specific fears about the actual amnio or CVS, worries that the test would harm the fetus or the needle would hurt going into the uterus or pierce the fetus. These fears made the testing time extremely upsetting.

> It was a terribly anxious time. I do think I was nervous about the amnio, I mean, I was breathing fast and I was trying to relax. I knew that if I didn't relax it could make the procedure worse, but I managed to relax OK.—Kate, 35, Jewish

> I was nervous! I mean I was nervous going in because I thought, "Am I doing some weird selfish thing that I have to know? And therefore I'm going to put this baby's life in danger or am I actually possibly going to find out something that I could fix early on with the baby, so therefore it's better for the baby?" I just thought of it as the whole weighing decision.—Rox, 38, Arab

> I was nervous. I absolutely vacillated during the whole time I was waiting for the amnio. I really didn't think anything was wrong with the baby.—Susan, 33, Italian/Irish

> The sensation was unnatural, and bad. Even though I was prepared for it, it was very unnerving. It didn't hurt, it just felt wrong. We spent all this time trying not to have a miscarriage, and then they stick the needle in there.—Hilary, 36, Irish

Kate, Rox and Susan all use "nervous" to describe their feelings during the amnio. They were focused on getting through the procedure, using breathing techniques and rationalization. Hilary voiced what most of the women were nervous about: miscarriage.

One woman I spoke with had a particularly horrible experience during her amnio.

> The doctor came in with someone doing a residency who was there to practice. When the doctor came in she looked haggard and frenzied. I asked her about how long it would take because I needed to prime myself for this. I

was not happy to be there, and I was very anxious about it. She said, "As long as it takes." The resident was on one side of me and she was on the other, and she's talking to him. I'm not looking, I've got my eyes closed and my head turned away because I'm not good with needles. So he inserts the needle on my right side and she says to him, "did you feel it penetrate the muscle?" and she says something else which I couldn't make out which made it sound like she was not pleased. Before you knew it, I feel another needle enter on my left side, where she is! They put two needles in! And she won't tell me a damn thing. And the next thing, I'm saying, "How's it going?" and she says, "OK, it's going fine. You just have to relax or this is going to take a long time. The more you relax the faster it will go." I'm starting to count now because I want to tell her to take the needle out and go screw herself. And just then, it's so strange, my stomach muscle, I assume that's what it was, involuntarily spasmed. And she says, "What was that? Did you move?" And she's standing right over me. I'm laying right there! You can tell if I'm moving or not. So that was that. She takes it out and that was the end of it. And I felt like I was going to throw up, I felt nauseous. And it was an awful experience.—Sydney, 41, English

After Sydney described this experience to me, she wanted to know if this was "normal" because this was her first amnio. I have never heard of different people inserting two needles simultaneously. It is not uncommon to have to do multiple insertions, but they are usually conducted by the same physician and with notice to the woman. Sydney survived and had a healthy child, but was terribly disappointed in the bedside manner of her genetic care providers and enhanced risk.

ANXIETY About Receiving Results

While fears about the test itself were very real, the anxiety women had about receiving the results had much wider reaching repercussions. Women were worried about what they would do if there were something wrong, and how they would handle the information.

> The actual process wasn't nearly as bad as just the mental anguish. It was a terrible stress to manage....And when I got the results, I was definitely relieved...Ummm, I felt like I had gotten over a hurdle. That I could relax a little, you know?—Maya, 34, Jewish

> I was scared to death, you know. Paralysis is the only way that I could really describe it while I was waiting for the results. I was so upset, so worried, you know, because your whole life, your whole baby's life is like waiting on a phone call. [when she got results] I felt like, OK, now I'm back in the normal pool of normal people.—Carla, 31, Jewish
>
> I had lots of anxiety that it wouldn't come out so good. I mean, I spent those two weeks like, "Oh shit!" I was crying. It was a very, very emotional time.—Sydney, 41, English
>
> You're anxious enough when you're pregnant, you don't need that sort of stress.—Tasha, 40, Caucasian
>
> I had reached the point of no return, where I couldn't stop obsessing over this possibility that these horrible thoughts could come true. I just decided that that kind of stress was not going to be productive. I remember being extremely nervous before and during the procedure. I just closed my eyes.—Mary, 34, European

Maya and Carla described waiting as "mental anguish" and "paralysis" while Sydney, Tasha and Mary used "stress," "emotional", and "obsessing" to explain how they experienced the fear while waiting for PGT results. These words are often used in the context of overreaction and mental illness scenarios. Their usage here is indicative of the nerve-wracking experience of PGT for these women.

My data about anxiety are not distinguishable from most other research querying pregnant women about prenatal genetic testing. Rapp (1999:106) asserts that prenatal genetic testing technologies "feed upon the more universal state of liminal pregnancy anxiety" and address women's "personal and social aspirations embodied in producing normatively accepted babies." Anxiety is one of the primary embodied emotions of these pregnant women, who are acutely aware of their pregnant bodies and the emotional experience of PGT. Several women referred to having PGT because they could not bear

the stress if they did not have testing of worrying the entire pregnancy.[7] These women would have worried more without PGT. I argue that this discursive position is available due in part to the biomedicalization and geneticization of American culture and the parallel routinization of PGT which makes the perception of this kind of testing "normal" and "necessary" as opposed to optional. The connotation of familial responsibility makes women feel they should have testing. For many of these women, the future of the child hangs in the balance of the results from PGT, and they were, in their minds, appropriately concerned about the outcome.

ANXIETY—Providers' Perspectives

It appears from the data that the providers understand this is an extremely emotional time for women. Their jobs are made more difficult by the heightened emotional states of the pregnant women, and the knowledge that sometimes the results from PGT are not what women desire or expect. Genetic professionals worry about the emotional impacts of their work on the pregnant women who have PGT, and how to make it an easier process.

> What one is dealing with are people in one of the most heightened anxious times. The women that are undergoing the procedure are extremely anxious. It's like nothing they've ever experienced before, so there's a lot of unknown involved. They you also have to deal with people's fear of needles, which is a whole other separate facet. So one has to in a short amount of time, make the situation tenable for the patients. Donald~obstetrician, 42, North American/European

> I think the screening programs are not well done. I think there has been very little attention paid to informed consent and education. And I think therefore it creates even more anxiety when people get abnormal results and then the

[7] The review in the "Emotions" section of chapter 2 summarizes the findings similar to mine: pregnant women who have PGT are anxious about what they will face with a genetic diagnosis, and worry that there will be something wrong found in their fetus. Interestingly, the literature also discussed women who wished they had had PGT and had experienced anxiety because they had not done so (Marteau, Johnson et al. 1989).

> OB doctors have to pick up the pieces and deal with them. I have not seen an analysis of anxiety, particularly, you know, how many car accidents there could be, or fights with your spouse or partner. I think it does create a lot of tension. Gertrude~medical geneticist, 69, white, Jewish

> The most important aspect of my work is getting the patient to a point where they can understand the information that's been given to them. Because a lot of what we do is so hard, and is so horrible, if you don't remember that component of it, the emotional component, it's really going to be completely lost. I think it's really helping the patient assimilate the news that you're giving them into their own life. Isabel~genetic counselor, 25, Asian Indian

> I'm not sure we have all the pieces to help people. And if we're possibly not causing more psychological harm than good. Clarissa~genetic counselor, 35, North American

Elaine, a genetic counselor, remembered a woman whose story epitomizes the complex dichotomy of the positive aspects of prenatal genetic screening and testing: abortion of a desired pregnancy to prevent emotional and physical trauma to the woman by the birth of a child who will not survive.

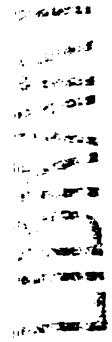
> There's this woman I remember who I really love. She'd had an undiagnosed pregnancy with anencephaly. She had gone to 42 weeks, had a C-section and had to deliver a baby with anencephaly through a real birth without knowing it was anencephalic. She was just wonderfully refreshing to speak to because she was so relieved that they'd come up with a way to check for women at increased risk for this kind of defect in their pregnancy, even if there were false positives. She said, "You know, at least you're picking out some of these pregnancies that have these birth defects and preventing women from having to go through what I went through. Please tell women who are thinking about not having the screening test to call me and I'll tell them what I went though." Elaine~genetic counselor, 53, North American/European, Jewish

Anxiety experienced by providers is another facet of the prenatal genetic testing experience. Genetic care providers expressed that they personally experienced anxiety when practicing genetic medicine because of the gravity of the decisions pregnant women

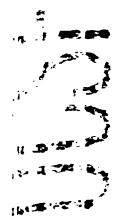
were making, and the stress of helping them to make these decisions. Some providers took on the PGT experience and made it personal.

> Your life experiences do impact your work, and for every child I see, for every pregnancy that's lost, naturally or whatever, you're still potentially pregnant here. You worry constantly. Some of the people who come here are colleagues and relatives. You're about as removed from this as the man in the moon. It has a dampening thing sometimes, but each day you just get up and just find the bright notes of the day. Gertrude~medical geneticist, 69, white, Jewish

> My boyfriend claims that I need to quit my job if I ever become pregnant. He wants to know how I could possibly be surrounded by this if I were pregnant. It's very true. I do know that it's hard on patients when they have a pregnant genetic counselor and they're dealing with an abnormality and the possibility of termination. I joke that I want to go to a deserted island when I'm pregnant. I'm impacted by seeing how ambivalence plays out in my patients and therefore, for me, I just want to avoid it all. I'm really afraid of the responsibility of making a decision if there was a problem. I'm afraid of what that would do to me personally. Clarissa~genetic counselor, 35, North American



> You know, I don't know what I would do in a lot of these situations. I just don't think I could ever have an abortion because I'm just so sensitive I think it would traumatize me. So I don't make any assumptions at all when I'm counseling my patients. Isabel~genetic counselor, 25, Asian Indian



Other providers, especially those who actually conducted the amniocentesis and CVS, had anxiety about the responsibilities they held to provide safe, effective genetic medicine for the pregnant women they care for.

> Doing over 2,000 amnios has the effect of separating one from your patients a little bit. Along the order of how do you take in the process of putting a sharp metal object through somebody's abdomen while they're pregnant. How do you deal with the losses the have happened after your procedures? I have a knowledge [laughing]. I've seen through the years many times that the diagnosis was made four weeks later, the baby was dead at 16 week size. So the loss isn't diagnosed immediately after the amnio. I have a little more caution when I see people after their amnios. I tend to do an office ultrasound to make sure that the baby's still alive. I do that for my patients because I want to make sure it's okay. [laughing] So I bring a little caution,

where I know the negatives. Donald~obstetrician, 42, North American/ European

The most important facet of my work is don't let mothers die and don't let babies die. Dane~perinatologist, 42, European

Donald's laughs were ironic, indicating his discomfort with the situation he and the pregnant women were in. Dane was sincere when he answered my question: "What is your role in this position and please describe your primary activities?" pointing out the most obvious basic job of a specialist in high risk pregnancies.

The complex nature of genetic medicine is illustrated by the range of emotions expressed by both pregnant women and their providers. The providers are in the situation of dealing with pregnant women in heightened emotional states, with the futures of their fetuses weighing in the balance. Often women want an opinion about what they should do. Genetic medicine is by tradition fundamentally non-directive, meaning information should be delivered in an informative way, not to guide or direct. Is this possible?

A CONUNDRUM OF PRENATAL GENETIC MEDICINE: TO BE DIRECTIVE OR NON-DIRECTIVE—THAT IS THE QUESTION

The mode of practice of genetic medicine regarding directiveness or non-directiveness is another issue made more complex by the data. This study demonstrates that the directive/non-directive dichotomy is not one. I found it is nearly impossible to practice genetic care in a non-directive manner because genetic information is loaded with uncertainty. The very nature of the work tasks revealed through the data is subjective. Based on an assessment of individual patients, the providers make decisions regarding what information is best suited to those patients. As pure non-directiveness is not possible, the dichotomy is deeply problematized. Problematizing non-directiveness is

not new (Clarke 1991; Veach, Bartels et al. 2001; Suter 2002), but the idea of "non-directiveness" must be further interrogated as prenatal genetic testing becomes more common.

Providers' Perspectives[8]

Each of the following providers has his/her own way of interpreting non-directiveness. Some problematize the non-directive stance by looking for what "fits" with their assessment of the particular pregnant woman to whom they are providing care:

> So I'm seeing a pretty select group of people who for the most part have made their decision that if anything's wrong with this kid they're not going to continue [the pregnancy]. On the other hand there is a small group who will change their mind based on the degree of severity of the condition and even a smaller group who are having the amnio just to find out if there's anything wrong so they can prepare for that. And I just have to know what group they fit in, or what counseling is going to give them the most support and benefits. So although we still say we're non-directive we do--are directive--in the fact that we might tailor the counseling for what we know the family is going to do no matter what we tell them. Robert~medical geneticist, 60, North American/European

> I just think counselors really know how to allow people to have their own feelings and their own decisions. We really try to focus on what's best for this particular person and family. And we can't always get to that, but it's kind of a matter of what's going to fit for this family. Clarissa~genetic counselor, 35, North American

> It's hard to know how much responsibility you take. You try and be non-directive, but is there some direction in something you say that influences someone to make a decision or not make a decision. I certainly counsel lots of couples who decide to have amnio and lots who decide not to have amnio, but it does make you wonder how do you make these decisions? And how do you help people make them? Elaine~genetic counselor, 53, North American/European, Jewish

[8] An earlier version of this section was published (Karlberg 2000) and is reprinted here with permission from Elsevier.

But other providers have particular instances that, in their minds, are the time for directiveness. In these instances, there is little or no factoring in of the pregnant woman's opinions. For example, in a genetic intake meeting there was a discussion between Dane, a perinatologist, and Clarissa, a genetic counselor, regarding their case of a woman pregnant with a fetus with a sex chromosomal and other anomalies:

> D: Did you tell her this baby's no good so she should terminate? You know this is the time for directive counseling.
>
> C: I just really think it will die.
>
> D: If the baby doesn't die it's gonna be a big problem. It's better off dead. The further you go in the pregnancy the higher risk you have for serious problems if you carry to term. Is she stacking a full deck?
>
> C: She's only twenty, and she seems to deny the baby is as bad as it is, but she does understand there are serious issues.
>
> D: So if it doesn't die, you're going to adopt it?

During my interview with Dane, he again expressed his directive stance:

> There are always ethical issues about the baby's right to live, and that society is going to bear the burden of a severely handicapped child that has no qualitative benefit from living. If I perceive it as a hopeless situation, then I try to have the patient look at it logically. Some people don't logically look at things, you know. You need to make sure they understand what is medically feasible. Dane~perinatologist, 42, European

Other genetic care providers agree that directive genetic care does is sometimes appropriate.

> I don't agree that you should *always* be nondirective. I think there are rare situations where you can be. I have on occasion recommended to parents, fairly strongly, that we do or do not do certain things. I had a family 8 years ago now, who had a baby with DS who had a complex heart condition who needed surgery. They wanted him to die. To be truly non-directive, you would have allowed nature to take its course and he would have died. But that, first didn't feel right, and second there's been enough court cases in the

> country saying you can't do that. Jacob~medical geneticist, 42, refused race/ethnicity
>
> There are times when I recommend termination. If it's clear cut like Trisomy 18 or Trisomy 13, prognosis is 0. I can direct the patient to make choices about the pregnancy. Jared~perinatologist, 42, white

The genetic researcher I spoke with recognized the problems with combining the information about the diagnosis with what the diagnosis means personally for the pregnant woman and her family. It complicates the tailoring of information approach.

> I think that it's very important that education and counseling be recognized as being two different facets of the same thing. It's very important that the information be presented separately. The information comes first, then how it's going to impact you and your life and your family. I think that those get a little muddied. You need to separate the information from the personal application of the information.—Jane~genetic researcher, 32, European

It is obvious from the data above that all genetic care providers do not believe they must be purely non-directive. I therefore argue that Kessler's (Kessler 2001) definition of non-directive counseling is impossible based on my data. Most of the providers I spoke with at some point in the discussion gave an example of a time when s/he attempted to persuad a pregnant woman of something during a genetic care interaction. My data revealed that providers tend to think abortion of fetuses with Down syndrome, anencephaly, Trisomy 13 or Trisomy 18 is acceptable, some even implying it is socially necessary.[9] The line between being directive and non-directive moves—is constructed only in the moment of practice. This point is made more obvious by pregnant women's experiences of genetic care during their prenatal genetic testing experiences.

[9] This line of thinking, that aborting fetuses with specific disorders is warranted, is supported in the literature, shown by the patient information pamphlets describing Down syndrome in clearly negative tones (Lippman and Wilfond 1992; Bryant, Murray et al.2001) and the providers' opinion on abortion (Veach, Bartels et al. 2001; Ettorre 2002; Rabino 2003).

Pregnant Women's Perspectives

While the goal for providers was to avoid making direct suggestions most of the time, women had different experiences of the testing. Many women recounted experiences of the providers telling them what they should do. Most of the doctors referred to here are obstetricians, supporting the literature that obstetricians are more likely to be directive than genetic professionals (Bernhardt and Bannerman 1982; Bernhardt, Geller et al. 1998; Egan, Kaminsky et al. 2002). This raises an interesting question: even if genetic care providers are non-directive most of the time, does the directiveness of the obstetricians counteract this?

Obstetricians

Meg had told her obstetrician she would not have an abortion for a DS baby, and therefore wasn't interested in amnio. She proceeded to tell me what happened after she said she didn't want the testing:
The obstetrician said, "Here's some other chromosomal abnormalities that can occur where the child will live for a year, be really uncomfortable and then die after a year." And then, I realized I didn't want that to happen, because we've had problems with my daughter since she was six months old. She was in and out of the hospital for a while, and I have seen a lot of sick children. I just knew I didn't want to go through that or put a child through that.—Meg, 2nd pregnancy, 39, Northern European

The results of the CVS came back a week later and it was Trisomy 18. So the doctors basically said that the pregnancy should be terminated because there was only a 5% chance it could go to term, and if it did, the baby wouldn't last a year. So I had the baby terminated the next week. My doctor recommended it. And the doctor who did the procedure recommended it, and I had to see a different doctor because my doctor was out, and he also said, "This is what you should do."—Amy, 38, Portuguese

I felt pressure to have the testing. Well, I felt like coming from a pro-life position...the doctors assume every woman is pro-choice. New York City isn't really a pro-life environment, so I never even felt like my physician questioned what my decision would be until I told him. And he was a little bit taken aback and probably would have advised me more strongly to terminate the pregnancy if the results had been positive.—Susan, 33, Italian/Irish

I think my husband had more fear than I did, so I would take pressure [to have the testing] more from him, definitely. And from the providers, well, they do pressure. They pressured me. You can be sure if you're over 35...I think for anybody over 35 they just start pouncing on you about the risks and scaring you. And part of it is probably legitimate, and part of it is the medical establishment.—Sotiria, 37, Greek

The doctor said, when I asked him about amnio and what he would do, he said, "We didn't have that option when my wife and I were having kids, but that's the only thing that I would do differently in our pregnancies. I would have her have an amnio. Even if she were under 35, I'd still have her have an amnio."—Robin, 33, Jewish

I think I was 34 at the time just about to turn 35 and she [Dr.] said that the results came back 1 in 39 chance of Down Syndrome, and she said, "You told me if something came back from the AFP you would have the amnio," and I said, "OK." She said, "Get in here right now."—Meg, 1st pregnancy, 39, Northern European

Directive techniques used by obstetricians included very specific negative descriptions of genetic diseases, pro-life discrimination, citations of alarming risk ratios, and a blatant directive statements like "this is what I would do if I were you," or "you should do this." There is little doubt these obstetric providers were certain they were providing direction when speaking with these pregnant women.

Perinatologists

The first specialist said "If you're not going to have amnio to rule out CMV, the only thing I can suggest is that you come in for a 28 week growth ultrasound. However, if at that point we see growth retardation which means there's a problem, then you need to know it's too late to terminate." When I had the amnio part of me felt like I was doing what they wanted, because that way they didn't have to worry about it, especially the first specialist. The second doctor I thought was great. He said, "If you were my wife, I wouldn't advise you to do this. If you want to, you can, but I'm not advising you." The first guy was like reading something out of a rule book, "I have to advise you of your rights." I think I've learned a lesson: even in the same practice you can get a lot of very different advice. Probably the one doctor who told me I didn't need the amnio gave be the best advice. But psychologically for me, it was too late at that time, because I had two other doctors telling me all these bad things.—Mary, 34, European

This perinatologist experience encompasses both the specific descriptions of scary genetic diseases and the "this is what I would do" scenario.

Genetic Counselors

The genetic counselor was extremely pushy about seeing if you could test for this syndrome having something to do with emphysema because of my partner's family history. And it seemed inconclusive that it could tell you whether the baby would develop emphysema from this gene. If the baby had the gene, we would go ahead with the pregnancy anyway, but the genetic counselor was extremely pushy about getting tested for that.—Jill, 47, Jewish

Radiologists

The radiologist did tell us, she said, "I do recommend an amnio." And so I was asking questions about that, and ummm, she just kept saying, "Well, you'll have the amnio and everything will be fine." And that was the first time that I was, not pressured, but I felt like that was the first time I was getting an opinion. My OB doctor is much more like, "We'll wait and see, we'll look at the results." And that was the first time I had anybody telling me, "You should do this."—Carla, 31, Jewish

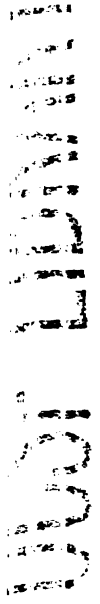
Both Jill and Carla found themselves in situations where the genetic care provider was more interested in the information coming from their testing than they were. Both the genetic counselor in Jill's quote and the radiologist in Carla's wanted the information for

themselves, and did not seem to incorporate the risks to the pregnant women they were speaking with into the decision.

The ways women were pressured were but a few examples of how genetic care is inherently directive. It is impossible for women to have information about PGT provided to them without someone making a judgment of some kind about whether they should have this testing or not. The women I spoke with who said they had not felt pressure to have PGT were women who went into pregnancy thinking they would have the testing, regardless of what the provider said.

This discussion of the differences and similarities between providers' perceptions of genetic care and pregnant women's experiences of it illustrates some basic points about prenatal genetic testing. This kind of testing engenders anxiety in the shape of stress, fear, and blatant, life-altering anxiety about the continuation of the pregnancy, the health of the woman and her fetus, the testing itself, and whether the woman made the right decision to have PGT in the first place. Providers recognize the complex nature of the decisions and choices pregnant women make and try to do their work in a way that enables the women to make sound, information-based decisions, but this is not always possible. The information they provide the women is often tailored to what they perceive are the needs of the pregnant woman and her family, and therefore is subjective. The providers themselves take on emotional stress when dealing with a pregnant woman with a genetic diagnosed fetus, trying to walk the line of non-directiveness, without short-changing the pregnant woman. The anxiety that weighs so heavily on women and genetic professionals is created because of the gravity of the decisions these pregnant

women and their families are making about the futures of their families and the lives of their potential children.



7 PREGNANT WOMEN

BALANCING GENETIC CARE AND EMBODIED KNOWLEDGES

Pregnant women who have genetic testing or screening are becoming more and more common (Olsen and Cross 1997; Spencer, Spencer et al. 2000; Halliday, Warren et al. 2001). It is increasingly rare to be pregnant and not have had at least one of the multitude of genetic screening tests available. Please see Appendix A for a discussion of the prenatal genetic testing and screening currently available and refer to the Glossary in Appendix H for brief definitions of terms.

What a woman experiences when she enters the world of medical genetics varies based on the type of health care facility she utilizes. It also varies by location in the country, availability of genetic specialists, funding for particular types of research in her area, the training of her provider, her insurance coverage, and many other elements. With this in mind, I first describe the three main types of providers active in genetic medicine for pregnant women. Next, I outline the approaches of two different kinds of health care organizations' to prenatal genetic testing to frame the experience for most women: an HMO and a large public teaching hospital. The two types I discuss are similar, but with important variations. I then turn to an overview of women's descriptions of their interactions with genetic care providers to frame the experience of genetic medicine for pregnant women.

OBTAINING GENETIC CARE: NAVIGATING DIFFERENT MEDICAL SYSTEMS

Today, informing the pregnant woman and her partner about prenatal genetic testing and its possibilities and limitations falls to many different kinds of providers,

depending upon the medical system the pregnant woman uses. In most instances, a pregnant woman is referred by an obstetrician to a genetic counselor and then meets with a medical geneticist or perinatologist to have the procedure. The results from PGT are usually delivered by a medical geneticist or a genetic counselor or both.

The bulk of my research with providers was conducted at a managed health care facility, a not-for-profit health maintenance organization (HMO) on the West Coast of the United States. Genetic services within this multi-facility HMO are coordinated by a group of medical geneticists. My research was focused at one site, interacting with individuals mainly from the genetics and perinatology departments. In this HMO, all pregnant women were offered prenatal screening, and women over 35 were offered amnio. Pregnant women attend a pregnancy class when they present as pregnant and intending to keep the pregnancy, whether through a pregnancy test or a visit with a nurse practitioner or obstetrician, at approximately 6-10 weeks gestation. In this pregnancy class, all prenatal genetic screening and testing is briefly discussed. There are forms included in the "pregnancy packet" that the woman is to fill out and provide to her OB. One form pertains to genetic risk, asking family history questions to identify women who may have genetic disease in their families and who would be considered "high risk." Then, at the appropriate point in the pregnancy, women are told by their nurse practitioner or OB how to make an appointment for amnio or CVS.

All women who have a prenatal genetic diagnostic test (amnio or CVS) see a genetic counselor either in a class or individual setting. If the woman is having amnio/CVS only because she is over 35, she is referred to the class conducted by a genetic counselor. If the woman is considered "high risk" for genetic problems,

indicating that she checked something on the genetic risk form, she will meet individually with a counselor. The class covers the basics of the testing, including diagrams of what the testing will look like, what the test can detect, how the woman may feel during and after the testing, and what side-effects there are to testing.

The amnio/CVS is performed by a perinatologist on one of the designated procedure days. The same genetic counselor then calls her with the results of the amnio/CVS. If the results are "normal" the interaction ends there. If the results are adverse[1] they then make an appointment to discuss them more completely with both a geneticist and the same genetic counselor. The call is never made on a Friday. The genetic counselors try to time the call so that the woman's family is home and she can ask as many questions as she likes surrounded by the support of her family. They keep appointments open every day for women who need to come in immediately who have had adverse results.

I also conducted a small amount of research at an East Coast division of human genetics in a large, public, metropolitan teaching hospital. Several thousand patients are seen every year at this clinic. The division does prenatal testing five days a week. Women are referred, usually by their obstetrician or self-referred to the division. Here, any patient interested in prenatal testing has a counseling session with an MD geneticist and possibly a genetic counselor and a resident or fellow. An informed consent process is undertaken at this meeting. All prenatal patients from the geographic area (except for private patients) are seen at this teaching hospital, regardless of insurance status. They

[1] My thanks to a medical geneticist I spoke with who explained that the connotations of using the word "abnormal" or "negative" to describe a genetic diagnosis were just too horrible to contemplate for pregnant women, so the use of "adverse" is substituted. Adverse is the new euphemism for abnormal.

are provided with written materials about testing options, and attend a class about prenatal genetic testing. The class is run by genetic counselors and contains diagrams of the procedures and basics about genetic inheritance and what happens with the sample at the laboratory. The risk rates for the facility for the specific procedures are also provided. After the class, each patient makes an appointment and later meets with a medical geneticist. This meeting consists of taking a family history, obtaining information on all relevant billing and insurance forms, and constructing a pedigree for three generations. Each interaction is individualized to that pregnant woman and her partner, assessing their risks, understanding their dates, their demographics, and their family history.

The prenatal genetic test is then conducted by a perinatologist or obstetrician. After the testing, the Division of Human Genetics encourages the primary provider, the one who referred the patient or who has an established relationship with the patient, to deliver the results. The contact of the patient is timed so that they can come in immediately with their questions if they wish. Most women with adverse results come in to meet with the medical geneticist again to review the results in detail, along with options.

Bearing in mind that these two different kinds of genetic centers are on opposite sides of the country, and may have different philosophies about medical care, the similarities and the differences between the two facilities are surprising. Both rely on pregnant women to trigger genetic care. The HMO model relies on the woman to screen herself in or out of the genetic system by filling out the genetic risk form, one of approximately 20 forms she is required to sign and submit to her OB after her pregnancy

class. The teaching hospital also does not seek women out; they are referred by non-genetic specialties like obstetrics and fertility specialists.

Once women are in the system, in the HMO they see first only a genetic counselor while at the teaching hospital they meet with a geneticist. The HMO pregnant women only see a geneticist if there is something wrong with the pregnancy. The prenatal genetic testing class format is similar. But again, the HMO women are screened and only see a genetic counselor if they are "high risk," thus assuming they answered the genetic risk form thoughtfully. In contrast, at the teaching hospital, pregnant women by now have seen both a geneticist and a genetic counselor. Risk rates for miscarriage after the different procedures are provided in both facilities, but facility-specific risk rates are only provided at the teaching hospital. The HMO has the data, even down to provider-specific risks, but does not share it with the pregnant women having the procedures.

The third main mode of pregnancy care in the U.S. is private health insurance. Here a woman would likely determine that she was pregnant through a pregnancy test at home and then confirm with her OB or primary care provider. That primary care physician would then refer her, if she intended to continue the pregnancy, to an OB. The OB would then be the provider to offer PGT and to refer the pregnant woman to a genetic care provider should she opt for such testing.

WOMEN'S EXPERIENCES OF PRENATAL GENETIC CARE

Pregnant women bear much of the responsibility to navigate through the maze of genetic care. This weight of responsibility for obtaining proper genetic care is combined with what women in American culture already feel about the "musts" of prenatal care in pregnancy. There were 32 pregnancies discussed in the 20 women I interviewed. These

women had varying assessments of their genetic medical care during the prenatal genetic testing experience. Please see Table 7A for statistics regarding genetic care in the women I interviewed.

Table 7A: Pregnant Women's Medical Experiences of PGT

Received genetic counseling *before* PGT	
Did not receive any genetic counseling *at all*	
Received care *primarily* from obstetricians during pregnancy	
Received care at some point in pregnancy from maternal-fetal medicine specialist/perinatologist	
Received care *primarily* from midwives during pregnancy	
Insurance covered PGT and screening	
Used fertility services to achieve pregnancy	

Some women expressed that they felt disdain from the providers they encountered:

> I do have to say that when you're dealing with the medical world and doctors, they really tend to make you feel like you don't know what the hell you're doing. They act like, "We're not going to explain anything to you." Like you really have to ask questions and they're not exactly friendly about it.—Rox, 38, Arab

> I'm sort of evangelical anytime I talk to somebody who's not having a straightforward pregnancy. I say, "Don't go to an ordinary OB, don't go to a perinatologist, go to a maternal-fetal medicine specialist. Let them guide you about whether you need them or not. Don't just bleed, find out why you're bleeding. I'm a little bit cynical about ordinary obstetricians.—Tasha, 40, Caucasian

One woman thought the entire genetic counseling process was useless:

> I thought genetic counseling was the thing to help. I can't believe how concerned people are with them. For me, it was literally a 10-minute experience, draw my family tree and ask if anyone had any major malformations. I had already told my doctor all of that in his office. Unless you were already predisposed to something, I don't really see the necessity of making the appointment, taking time out of my day to go, drag my husband to it.—Susan, 33, Italian/Irish

Overall, the women were more positive than negative when speaking about providers of genetic medicine. Forty percent of women said genetic counselors provided them useful information, while fifteen percent of women said they had negative experiences with genetic counselors. The negative experiences included time spent with the provider was not helpful, and the counselor was pressuring them to do testing. Jill expressed what most women I spoke with insinuated if they did not state it outright: for the most part genetic care providers were helpful and supportive.

> I trust my doctor so I think he's been a tremendous source of information, comfort, security. And the genetic counselor, despite her being a little nutty, was actually extremely informative. The doctors that performed all the sonograms have also been varying degrees of helpful and not helpful.—Jill, 47, Jewish

Interestingly, the pregnant women I interviewed frequently referred to their obstetricians as sources of information on genetic topics. Most obstetricians do not have genetic training, and this can cause problems in genetic medicine, because they may provide incorrect information. One problem with the field is that other medical specialties do not clearly understand the complexities of medical genetics, and there are not enough genetics professionals to address the growing need for education. (See Appendix D for further explanation of this point.) There are even referrals of patients for tests that do not

exist. Gertrude a medical geneticist stated, "People are sent in by their doctors to have these tests. It may not even be a test."

Sources of useful information varied, and were different and multiple for the women I interviewed. Some found genetic care providers useful while others did not. Please see table 7B for a summary of sources of information. Most women looked to friends or relatives as well as medical practitioners, but they also relied on their bodies and feelings about pregnancy to help them make decisions regarding prenatal genetic testing.

Table 7B: Sources of Information About PGT

Useful Source	Percentage of 20 Women
Genetic counselors	40%---8 women
Obstetricians	55%---11 women
Internet	45%---9 women
"What to Expect When You're Expecting" pregnancy book	40%---8 women
Friends/Relatives	100%---20 women
Not Useful Source	**Percentage of 20 Women**
Genetic counselors	15%---3 women
Obstetricians	15%---3 women
Internet	25%---5 women

The different types of medical model shape the way genetic care is provided, but do not erase the discourses permeating genetic care discussed in Chapter 5. Women experienced their providers as helpful on many levels, but superior and unavailable on others. The primary experience of genetic medicine for women was the gathering of

information about PGT in order to make decisions about testing and what it would mean. This information gathering included interactions with providers as well as researching outside sources of information. As I discuss below, women also relied on embodied knowledges to make these critical decisions.

EMBODIED KNOWLEDGES

From the moment the pregnancy test is positive, most women are consciously pregnant. If they continue the pregnancy, they commonly embody pregnancy by living the awareness through doing what one "should" do during pregnancy, such as take prenatal vitamins, and avoiding "should nots" like consuming alcohol and caffeine. This embodied knowledge of a growing entity inside her body alters perceptions and experiences for the newly pregnant woman, even though the world may not know she is pregnant. This understanding and changed perception affects the PGT decision-making process and the experience of testing itself. In this section I explore data relating to pregnant women's understandings of their bodies and why they decided to have testing. I examine their knowledge quests, use of statistics, and searching for peace of mind throughout the PGT experience. I discuss the coping mechanism of the "suspension of pregnancy" (Beeson 1984) employed by many of the women I spoke with to deal with the waiting game between having testing and receiving the results. I also briefly examine the ways providers perceive pregnant women at this crucial time.

Overall I collected data on 32 pregnancies among the twenty women I interviewed. Some of these women had other children and had had genetic testing or screening in those pregnancies as well. Please see table 7C for specific statistics on genetic testing and screening. One pregnancy had no testing or screening. Screening was

a common theme among the women I spoke with, and many of them had more than one genetic screening test in conjunction with the diagnostic procedure of either amnio or CVS. Overall, there were 43 genetic screening tests conducted on these twenty women in the first 24 weeks of their pregnancies. There were 69 genetic screening and diagnostic tests conducted over the course of 32 pregnancies, an average of 2.2 genetic tests per pregnancy.

Table 7C: Types of PGT and Screening Utilized by Women

Amnio or CVS	81%---26 pregnancies
Amnio	62%---20 pregnancies
CVS	16%---5 pregnancies
Attempted CVS and then had to have amnio	3%---1 pregnancy
Jewish panel for carrier screening	25%---8 pregnancies
Specific disorder screening, e.g. CF or Tay Sachs	19%---6 pregnancies
Triple screen	22%---7 pregnancies
Quad screen	25%---8 pregnancies
Nuchal translucency ultrasound screening	28%---9 pregnancies
FISH	9%---3 pregnancies

A woman who recently moved to New York attributed the eagerness of women to be tested in New York to a pervasive attitude in the area, while a New York geneticist said it was a knowledge quest:

I don't have that many friends who've done amnio where I used to live (in the San Francisco Bay Area). We're just kind of getting to the age now where my peers are doing it. In New York, though, most of my friends in New York had it, and they were all very adamant that they would have terminated had they had a Trisomy 21 fetus, so that was interesting. You know, their reaction was more like you would have to know because you don't want to have this compromised baby. We just felt we needed to be prepared. I had two friends in the last year who had Trisomy 18 miscarriages. One was 30 and the other 34. In New York everybody has amnio at 32. I find New York's a very tech heavy city.—Nissa, 36, Mexican, Irish and Jewish

I want people to feel that they are in charge of their lives as best they can be. Which is of course the New Yorker way. It's not control, it's called knowing everything you need to know. Gertrude~geneticist, 69, white, Jewish

PREGNANT BODIES: EMBODIED KNOWLEDGES

The pregnant women I spoke with earnestly launched themselves into the matrix of medical genetics in their pursuit of a textbook pregnancy, one without complications and genetic abnormalities. Overall, these women were *not* confused about what the PGT could offer them, and most had a clear understanding of the limitations of the testing. They opted for testing because they thought they should know everything they could know about the pregnancy. They did so despite feeling they were healthy and therefore not at risk for anything in particular. I call such feeling "embodied knowledges." I frame embodied knowledges as, in part, produced through the biomedicalization and geneticization of pregnancy. That is, "embodied knowledges" are self-knowledges that women distinguish from the information and assessments provided by the various medical professionals with whom they interacted during pregnancy. These women knew testing was a choice, believed they were healthy, and yet still had testing.

Some women felt they were mere vessels for the fetus growing inside them, especially during the testing procedure, when there were ultrasounds and needles involved. They removed themselves from the experience and focused on the fetus.

> When I realized I was pregnant, it's such a surrendering of your body. So I was like, it was like this alternate self [when having amnio], and I kind of feel like that about the pregnancy. It's all so weird, it's so foreign from your own knowledge of your body. It didn't bother me, though, and getting to see the baby on screen was so great.—Nissa, 36, Mexican, Irish, and Jewish

> I did sort of feel like I'm just this vessel for now. I was so focused on the uterus and the fetus that I didn't think about the way the doctors were also [focused on the uterus and the fetus].—Kate, 35, Jewish

> They're not looking at me, they're looking at him [fetus]...so it was kind of like it wasn't really even about me. I was just the outside casing, you know? It's almost like I wasn't there. I don't know how to explain it. It's like focusing on him and all this stuff, and I was just like the layer in between, you know? It just felt like a really cold detached thing.—Rox, 38, Arab

Nissa explicitly states that she is "surrendering" her body to the pregnancy, and ruminates on how she is separated from the pregnancy self. Kate recognized that she and her doctors were more focused on the fetus than the fact that it was housed in her body. And Rox called herself the "casing" for the fetus. These women experienced a disconnect between their bodies and their selves as individuals, effectively creating genetic bodies through choosing to have PGT.

Women also expressed surprise that they were having testing, almost as though it happened of its own accord. A recurrent theme was, "I'm healthy, why should anything go wrong?" Yet the women still had genetic screening and testing conducted during their pregnancies.

> I'm like, oh, I'm 31, I'm young, I'm healthy, blah, blah, blah...And the doctor said, "OK, in the future you know these are the tests we're going to do and an amnio may be one of them." And it kind of went in one ear and out

> the other because I'm 31....And then when we went in for the first ultrasound the doctor said the baby's kidneys are enlarged. We met with the genetic counselor that afternoon and when you put all the pieces together, I'm the equivalent of a 40 year old. So [laugh] I'm not the healthy 21 year old, but I still think I'm young, and this isn't us.—Carla, 31, Jewish

> Well, I already have a healthy child, and I know that most children are healthy [laugh]---something like 95% or 96%, even at my age [37]. So I felt that the baby would probably be healthy. I wasn't going to worry about it. And you know, I do what I can to encourage that with taking my prenatal vitamins and stuff. I wasn't obsessed with the baby's health. My family is very healthy....At some point I wondered, "Did I make a mistake? Would it have been better to stick with one kid who's healthy?" Somewhere in the middle I started to wonder and then [after test results] I was relieved that "oh, once again I'm back on course with probably, hopefully having a healthy baby." But it made me a little bit angry at myself for having given in to the testing and to my husband. But I understand his concern.—Sotiria, 37, Greek

> I come from a very healthy family. We're strong as an ox. There's not a lot of sickness or illness. And my partner's family is, from what I can tell, except for the emphysema thing, very healthy as well. So I always assumed I was not in that percentage of people that were likely to have these kinds of things.—Jill, 47, Jewish

Another recurring theme was age, intimately related to gender and bodies. Forty-five percent of women made specific comments about their age in relation to prenatal genetic testing, separate from the sixty percent who supplied "age" as the medical reason for having PGT. The biomedical construction of 35 as the age at which women become high risk for genetic abnormalities and thus should have PGT was challenged by some of these women.

> There's been a recognition of an aging body since we entered into this pregnancy thing, because I was over 35 when we started this. So an aging body with regard to fertility has been over us for a long time. But I can separate it from the rest of me. It's just a fact of life.—Sydney, 41, English

Jill, the oldest woman I spoke with, said she was getting younger through pregnancy: "I'm Superwoman." In sum, many of these women say, "I'm young,

healthy" yet they submit to PGT because it is framed as "routine" and women who do not have it are perceived as "abnormal" (Richards and Green 1993).

PREGNANT BODIES AS GENETIC BODIES

Despite their embodied knowledges of health and well-being, these women worked to separate self from body on some levels. All the women I talked to felt they willingly and knowledgeably had entered into prenatal genetic testing, despite some pressure from outside sources examined in Chapter six. Theoretical issues around embodiment including Foucualdian (1975) normalization of bodies and Grosz's (1994) corporeal feminism all echoed in the data. The volatility and changing nature of pregnant women's bodies is evidenced through the quotes above about women thinking their bodies were vessels, and their perceptions that they were not really present during testing. Their perceived separation from the embodied experience of pregnancy and from being pregnant is revisited later in this chapter when I discuss suspension of pregnancy. Here I first discuss the geneticization of the pregnant body.

Pregnant women's bodies are inscribed through PGT and the resulting genetic body, the pregnant body, is volatile and shifting for the pregnant woman herself, unless or until she is reassured of the normalcy of the genetic body through "normal" fetal genetics results. When the results of PGT are not "normal," the stigma of genetic information is yet another vehicle for the geneticizing of pregnant women's bodies and families. While I argue that genetic stigmatization is in practice ultimately dependent upon public knowledge of inheritance patterns, through the geneticization of American culture everything is potentially genetic. Thus when a baby is born with something wrong, one of the first questions is: "Did the mother have prenatal genetic testing?" I

suggest that stigmatization based on the assumption of *maternal* genetic responsibility for each pregnancy is a real possibility, and a potential motivator for pregnant women to have PGT.[2] The body awareness of the women I spoke with reflects how women incorporate information about themselves, their bodies, their genetics and the ramifications of genetic information for their pregnancies and their families into their embodied knowledges. The women generally believed they had healthy bodies, yet, through the availability of the technologies of prenatal genetic testing, women's bodies and the futures of their families are reconstituted as genetic entities, revealed as potentially abnormal.

CHALLENGING EMBODIED KNOWLEDGES: WHY HAVE TESTING?

While the women I spoke with are in no way a generalizable sample, they do offer a range of perspectives on the acceptance of PGT. Only ten percent of women believed they had to have prenatal genetic testing and screening. A quarter of the women went into their pregnancies believing they would have testing. Forty percent of women told me they did not really want testing in the beginning of their pregnancies, but changed their minds during the course of the pregnancy. Ten percent said they wished they had not had testing this pregnancy. One woman will never have PGT again, even though she plans on having more children. Twenty percent of women explicitly stated they were glad they had had the testing.

The reasons the women gave me for having amnio or CVS varied greatly. Of the twenty-six pregnancies that had amnio or CVS, forty-three percent were tested because of

[2] The literature supports this assertion through the idea of stigmatization based on genetic knoweldges altering self-concept and life choices (Kenen and Schmidt 1978; Marteau and Anionwu 1996; McConkie-Rosell and DeVellis 2000; Cleaver 2002; Geller, Alper et al. 2002; Gordon, Devaney et al. 2002).

the age of the woman. Eighteen percent had abnormal ultrasounds, ranging from abnormal nuchal translucency tests to the detection of choriod plexus cysts. Eighteen percent of pregnancies had abnormal screening results, either from a quad screen or a triple screen. One woman had CVS in her third pregnancy after amnio in the first two purely out of fear because a sister-in-law had had a child with disabilities. One pregnancy had amnio to rule out exposure to cytomegalovirus. Seven percent of pregnancies had CVS because of genetic abnormalities detected in previous pregnancies. But these medical indications for PGT were not the only reasons these women had testing. The other reasons included a quest for knowledge, seeking peace of mind and, in some ways, control of an uncontrollable situation. Those who were on a knowledge quest were often searching for some semblance of control.

> The only reason I decided to do the amnio was just for my own knowledge, to make preparation if there was something wrong. I've already decided after this whole experience that I'm not doing any of the prenatal testing in my next pregnancy. I'd rather just try to get through the pregnancy without any of that stress.—Susan, 33, Italian/Irish

> Well, I just wanted to know. I wanted the security of knowing...I just wanted all the information I can get, which I also understand can be a problem....But I'm very curious and I always want as many answers as I can get, and I just wanted to know exactly where I was going.—Meg, 39, Northern European

Women who were searching for peace of mind through PGT wanted to know everything was okay in their pregnancies, as far as the testing could predict.

> I think I just wanted some peace of mind. And to know that he was, as much control as I could have of knowing he [the fetus] was healthy. Maybe control is not the right word, as much knowledge as I could have to know that he was healthy. I wanted it to just give me some peace of mind that everything was OK.—Rox, 38, Arab

> I'm a big worrier, so I had it because otherwise I just would've been panicked throughout my entire pregnancy. I just really gained peace of mind. You

> know that there are other things that can go wrong, it's not telling you you're going to have the perfect healthy baby. I just would rather know than not know.—Delilah, 36, Eastern European

> I was looking for peace of mind. There was a very high chance that I would have a DS baby given my age, so I wanted to know that I was free of everything because, quite frankly, if it was a DS or born with some other type of disease, I probably would not have continued.—Tiffany, 39, Caucasian

The peace of mind these women sought was to quell the anxiety around unhealthy or abnormal fetuses. I do not think the women I spoke with were more anxious than the average pregnant woman, and I believe this quest for peace is ultimately about a search for "normal" healthy children.

For the two women who had had previous pregnancies with genetic disease and opted for termination, the prenatal genetic testing was a balancing of risks and responsibilities.

> When I had the CVS, I felt kind of good in a sense because I felt like I was taking responsibility. When I had the termination, I really did feel like I was doing the right thing. I should just face up to the risks and meet them head on and go with whatever the results are. I felt kind of strong, not particularly happy about it, but I definitely did not feel like a victim or anything. I felt more like I was taking charge, I think.—Tasha, 40, Caucasian

Tasha framed her decision to have CVS as a responsibility, almost a parenting decision. The termination was empowering because she felt she had made the right choice for her family, not to include a member with Down Syndrome. Tasha "took charge" by deciding the DS fetus was not a good fit for her family.

Some women were particularly focused on the statistics available to them regarding risks of amnio for miscarriage and for genetic defects in general. They used the numbers to rationalize their decisions to put their desired fetuses at risk to gain genetic knowledge.

I was more concerned that I'd have a healthy child. I guess I always look at the .05% risk of miscarriage. To me that just seemed so little, and the risk of having another baby with Trisomy 18 is much greater to me. What I've learned about myself is that I have no problem getting pregnant, so I can always try again, and I just relax.—Amy, 38, Portuguese

I did debate it with my husband [whether to have amnio], and I did agonize over it for many days. One thing we resolved was, they tell you that the risk is 1 in 400...and to me that sounded not such great odds, and then my husband turned that around and said, "What that's really saying is that 99.75% of the time there is no problem. So those odds seemed very good. You know?—Maya, 34, Jewish

The risk of miscarriage just added to the confusion, because if you look at my risks, for everything else, if you just looked at the quad screen thing between 1/200 and 1/400, so, OK, confused!!! Um, and that's why I had such a hard time making the decision. Originally when we just had one soft sign [for Down Syndrome from ultrasound], I took the risk of miscarriage as being pretty high, and then when we had two soft signs, the risk of miscarriage became almost incidental. It kind of made me shift how I was thinking about what could be wrong, and I figured, "I'm not going to have a miscarriage." That was basically it...My chances became about 1/142 for DS and 1/100 for any genetic disorder. And that sounds scary. But if you flip that around and say, "ok, I have a 1 in 141 out of 142 chance of having a healthy baby...well nobody told me that! So that's a 99.3% chance of having a healthy baby, you know? [laugh] And I wish somebody would have told me that.—Carla, 31, Jewish

I had to kind of give myself a talk on statistics. We probably won't have a miscarriage. I also knew I was at an excellent facility—that helped me. I know that they don't publish their own statistics, so they had to quote me 1/300, but I was pretty confident that it was less, even less. I thought it was probably like one in four or five hundred.—Mary, 34, Jewish

Thus we can see a very intense focus among some women on playing the statistics game. The risks of miscarriage from the PGT procedure are no doubt in part a response to risk reporting and a consequence of living in a risk society. Carla and Maya's husbands played the numbers game and shifted the statistics around to demonstrate that there were not significant risks.

From the time of the introduction of genetic screening/testing into a woman's pregnancy, the implications of PGT commonly influence the way she feels about the fetus. For many of the women I interviewed, once the decision has been made regarding the testing, the timing of the testing and receipt of results rule the woman's world.[3]

QUESTIONING EMBODIED KNOWLEDGES:

SUSPENSION OF PREGNANCY

Embodied knowledge was further represented by many women waiting to bond with the fetus until after they received "its okay there are 23 pairs of chromosomes" result from amnio/CVS. I use Beeson's (Beeson 1984) term "suspension of pregnancy" to express the women's fear and unwillingness to *be* pregnant as a lived experience until the test results were normal. This hesitation arises from the possibility of genetic problems with the fetus and the ramifications of that knowledge for the pregnancy itself. One strategy to endure the waiting period before the results were available was to attempt *not* to bond with the fetus.

> I just felt a little more like I was trying to not get attached to the baby as early as I did the last time, just because there's that possibility. I didn't really start telling the public until I got the results back.—Delilah, 36, Eastern European

> I think I felt more detached from him [the fetus] at that moment [having the testing] because I kind of had to be. Waiting for the results I was just in limbo. Am I really pregnant? Am I not? What's going on? And once I got the results back it was like, "OK, now I can officially tell people I'm pregnant." I'm going to be the pregnant lady now.—Rox, 38, Arab

> I guess I felt up until the amnio that I could lose this baby any day. Not that I wasn't bonding with the baby or happy with the pregnancy. I was very cautious and not as excited as my husband. I still feel like that sometimes. The amnio made me feel like it's OK to be more comfortable with being

[3] These data support the literature discussed in Chapter 2 "Women's utilization of prenatal genetic testing." The women's choices to have PGT reflected Wertz and Fletcher's(1993:183) finding that, "most women, including most who consider themselves feminists, seem relieved to be able to make the choices implied in prenatal diagnosis."

> pregnant, as opposed to being on guard. It kind of helped me make the baby more real, more believable, and that it's actually happening, that there will be a baby.—Tiffany, 39, Caucasian

> When I got pregnant with him, I was worried. It's funny, because I don't think that it affected getting attached to him at all. We just kind of took it day by day, but I think it was kind of affecting us still. You try not to bond too much, but I did. You don't want to, but you just do it anyway. And then I even thought, I just can't go through a late termination ever again, so we decided to have CVS.—Tasha, 40, Caucasian

> I didn't want to find out the sex of the baby with my first pregnancy because it allowed me to maintain a certain amount of emotional distance. I was concerned that if I found out the sex of this baby I would then think about it as a person.—Hilary, 36, Irish

Another mechanism employed to suspend the pregnancy was to wait to acknowledge it to the world until the genetic testing results were back and acceptable to the woman and her family.

> I had been sort of tempted to just not tell people until we got the AFP results back, but my husband really didn't want to wait. I just didn't want it to be a public event, you know, I didn't want to have to come up with a lie if there was something wrong and we terminated.—Robin, 33, Jewish

> That was upsetting [having to wait for amnio because the CVS was impossible to do], and it was upsetting for a lot of emotional reasons because of where we were in the process. We hadn't told anybody at all that we were pregnant. We were going to wait until the results from the CVS were in, so the idea of keeping this a secret until I was almost five months pregnant was very difficult. I, for one, was feeling a great need to start to talk to other people and tell people. And I was beginning to show, in my mind. That was the harder part, I think. I was disappointed that this meant another five weeks of being very private about what was going on, you know?—Jill, 47, Jewish

These tactics to deal with the fear of a genetically abnormal fetus and the possibility of choosing a termination are also supported in the literature.[4] This suspension of pregnancy I assert is due in part to the routinization of PGT procedures. Such

[4] For example see Rapp's (1999) extensive body of ethnographic research. Both Beeson (1984) and Rothman's (1986) work has been supported in other literatures, and I found similar explanations by women who waited to commit to their pregnancies because they might terminate if something was found.

routinization of prenatal genetic testing leads to an increased responsibility for the pregnant women in regards to what type of child she bears[5]. Combined with the extremely controversial cultural status of abortion in the U.S. today, this makes pregnant women uncomfortable to admit they are pregnant before they can decide if they want the option of abortion. (Please see Appendix C for more on this topic.) Most genetic care providers recognize suspension of pregnancy and other coping mechanisms and do their best to help the woman deal with their individual situations.

OVERRIDING EMBODIED KNOWLEDGES: PROVIDERS' UNDERSTANDINGS OF PREGNANT WOMEN HAVING PGT

Providers usually recognize the dilemmas women confront when making choices about having testing and try to empathize with them.

> Some [pregnant women] appreciate knowing and some wish they'd never know for the rest of their pregnancy. We try to encourage people to think about that before they make a decision. Many people will say, "Well, I'm not sure it would help me to be prepared." You try to think whether knowing something like that for the rest of your pregnancy, not being able to go through the normal kind of pregnancy anticipating a normal baby—would that be a really heavy burden to carry? Do they feel they could adjust? Or would they rather have the experience of pregnancy be what they thought it would be? It's hard to know those things. It's often hard to predict how you're going to feel with one piece of information or another. Clearly people coming in for this kind of testing, what they really want is some assurance that everything we can test for is fine. That influences a lot of decisions. They're just optimistic, hoping for good news, and it's really hard to think in advance. Elaine~genetic counselor, 53, North American/European, Jewish

> Human nature is always amazing. You just can't predict what any one person is going to do or want to do here until you hear the story afterwards. You can think you can. Sometimes you kind of guess. But you can't know. Gertrude~medical geneticist, 69, white, Jewish

[5] For examples please see (Marteau 1995) and (Menen and Weiner 2000).

The perspectives of providers on these issues has not been explored in the literature I examined. The empathy of the genetic care providers I spoke with about the dilemmas pregnant women face in making the decision to have testing was revealing. The genetic professionals take seriously the situations the pregnant women find themselves in when they have PGT, and try to help them make the most informed decisions for themselves and their families. As discussed earlier, the women's decisions are often a surprise to the providers, which calls into question some of the techniques the providers use.

Pregnant women are unique in that they are faced with making decisions about the potential of a possible life they have growing inside them. They live the knowledges and experience the knowledges of their bodies and the growing bodies inside them. They question health, identity, and place in the world while still adjusting to the idea of housing another being for nine months. Making the decision to have PGT is an arduous one, and the process draws on the embodied knowledges only they are wholly privy to. Pregnant women also decide when they will announce that they are pregnant to everyone but themselves. Suspension of pregnancy and not announcing it serve as boundaries between women and the emotional responsibility of presenting as pregnant in the world. It enables pregnant women to cope with the responsibilities they bear as a result of becoming pregnant in this age of genetic technological availability. The ways they navigate the genetic medicine at their disposal depends in part upon the types of providers they encounter during their pregnancies, and what information they are provided with or can cull on their own from the massive amounts of media about prenatal genetic testing.

8 BRAVE NEW FAMILIES

WOMEN'S AND PROVIDERS' IDEAS ABOUT SHAPING FAMILIES THROUGH PRENATAL GENETIC TESTING

In this chapter I explore the changing nature of family, through discussions of the role of abortion in prenatal genetic testing and the production of genetically shaped families through the experiences of pregnant women and genetic care providers' interpretations.

Today, family and genetic medicine are melded in the minds of many physicians, and it appears from my data that pregnant women see this to be the case as well. Genetic medicine has permeated pregnant women's experiences of pregnancy as discussed in chapter 7 and these findings were supported in the literature review. While research that confirms prior results is in itself important, one of the most exciting new findings in this research is that genetic medicine is changing pregnant women's notions of family.

What "family" is has been altered by the possibilities of shaping family into what one desires, rather than taking family as what one is given. My definition of family, a social reality constructed through shared meanings and experiences, incorporates considerations of what Nelkin and Lindee (1995) call "molecular families" where members are linked through genetics. But my term "genetic families" also allows for some broader interpretations. What is interesting from my data is that both the pregnant women and genetic care providers I spoke with believe that "genetic parents" are constructing families through their *shared meanings of genetic fitness for that particular family*. Thus they are fulfilling Strathern's (1992) ideas about the cultural constructions of the family by linking society and procreation through kinship. The desire to

perpetuate the bonds of family through generations Becker (1990) is epitomized in the choices women make during the PGT process.

The genetic family is facilitated by prenatal genetic testing and screening, most obviously through defining the family as genetic---its members as genetically linked---and therefore malleable by screening out fetuses with undesirable traits that can be detected. These undesirable traits are traits that may be subject to genetic stigmatization. Further, the genetic stigmatization of certain detectable genetic disorders can be viewed as a bridge to the potential stigmatization of genetic irresponsibility. Here, for example, women who have children with Down syndrome are queried as to whether they had a test to detect the disorder prenatally. The query implies that if they did not, they failed in their parental responsibilities to the future child. The reverse of this holds for providers: if they do not provide information about testing to pregnant women, they can be held liable for births of children that parents argue would not exist if they could have detected the disorders prenatally and aborted the fetuses (Cleaver 2002; Marteau 1996).

The uniqueness of genetic families is their multiple social realities: they are traditional and nuclear in the blood relation of members, yet they are technological and biomedical through obtaining actual "genetic" information using genetic sampling and testing techniques. The linkages sustained through the strengths of these socially mediated and legitimated blood relations and biotechnological verification are the sources of the power of the genetic families. Genetic families are "true" families because they meet the traditional definitions, but they are also postmodern because they are defined through biotechnology and genetic linkages. Pregnant women are in the tenuous position

of mediating the needs and desires of individual families in relation to the social acceptability and social demands of American society in the 2000's.

GENETIC FAMILIES: CONSIDERING ABORTION

The sense of family, whether the first pregnancy or not, was an important factor for these pregnant women in making the decision to have PGT. The primary way to shape a family in the case of negative testing outcomes is to abort fetuses that are not in line with expectations of "family" for the pregnant woman and her husband/partner and other children. In terms of their intentions in the event of an abnormal diagnosis, 40% of the women said they *would* have abortions, ten percent had had previous abortions, and another twenty-five percent thought they *would probably* abort. Fifteen percent of the women said they were *not considering* abortion, and ten percent either did not know or did not say during the interview. One of the women who would not consider abortion described herself as "pro-life". Overall 75% of the sample stated they would have or would consider having an abortion if a genetic abnormality was detected through PGT. Ten percent of the women I interviewed had terminated pregnancies because of a genetic diagnosis.[1]

Women expressed fear and anxiety about making decisions to abort their very wanted pregnancies. Some hedged the decision, not committing to abortion prior to knowing the diagnosis:

> I remember thinking to myself [while having the amnio], "I'm so pregnant already, and I look so pregnant"...I kept thinking, "What am I going to do if this pregnancy either miscarries or I decide to terminate. What am I going to tell everybody?" Yeah, oh god, I can't even imagine, and people do it too. I

[1] These women followed the literature (Alembik, Roth et al. 2002) and aborted fetuses with chromosomal abnormalities, and expressed the emotional trauma of the experience, as does the literature (Black 1994; Green and Statham 1996).

> know people have done it because there are all kinds of horror stories, but I just could not even imagine how one could do that.—Kate, 35 Jewish

> I know that it would've really been hard if something was wrong and I decided I did want to terminate. You see it there and you see it kicking, you know? [During the ultrasound before the amnio] I've always been pro-choice, but when you are actually there you kind of realize that "oh my god that really is a person" you know?—Maya, 34, Jewish

Others were certain that if there were a diagnosis that was not genetically normal or acceptable to their idea of family, they would abort:

> I suppose if the decision whether to keep or terminate wasn't predicated on the outcome of such testing we wouldn't have done it.—Sydney, 41, English

> It was strongly encouraged that I have the amnio done, and for my husband and I it was really a big decision because we had decided that if the baby did have a really good chance of having DS we would not have the baby.—Tiffany, 40, Caucasian

Still others were certain they would do something, but did not want to state it, feeling this would be tempting fate:

> I think it was more understood. I said, "You know, the doctor said that if we're not going to do this [terminate a pregnancy with an abnormality], we shouldn't have amnio. Are we going to do something?" And he said, "I think so, don't you?" And I said, "Yes." I feel a little bit guilty that that's my answer, but that's my answer. I'm sure it would have been a complicated and very emotional decision. We didn't go there.—Hilary, 36, Irish

The two women who had terminated pregnancies because of adverse genetic diagnoses talked about abortion in different tones. They were thankful for the option and availability of termination. Tasha explained the complex decision-making she and her husband went through, despite the fact that they agreed they should terminate the Down Syndrome pregnancy.

> We were both pretty confident on our views on things, and it's funny, because even though we kind of intellectually knew that we would want to terminate the pregnancy, we went through a whole process of making a decision about it. I had lost four pregnancies and we desperately wanted a

> baby, but we just didn't think it was right to have a child knowing what we knew. It was just a terrible, gut-wrenching decision. It was probably the hardest thing we've ever been through. I don't think we ever thought about not terminating, but we still had to go through that whole process.—Tasha, 40, Caucasian

For Amy who terminated a Trisomy 18 pregnancy, there was little discussion about whether they would terminate. Her decision was easy because the doctors told her the fetus's condition was incompatible with life. The directiveness of her genetic care providers removed the responsibility from Amy, so the decision-making about abortion was not *the* issue for her and her husband. Her solution to the emotional loss of the pregnancy was to get pregnant again. She felt the only way to get over the termination was to have a baby.

> I had a nuchal translucency test at 12 weeks and they found something on the neck. My doctor told us we should have CVS to find out the extent of the problem. My husband's cousin is a pediatrician and he said we should come to Delaware to have it done. So, interestingly, I went there to have the procedure on September 11, 2001. They couldn't fly the results to Baltimore where they do the analysis so we didn't have it done that day, but because we were there they did an ultrasound. The doctor said, "I think the baby's going to be fine. I don't see anything. Maybe it just worked itself out." The next day I actually had the CVS done and we drove the results ourselves to Johns Hopkins. It came back a week later that it was Trisomy 18. So they basically said that we should terminate because there was a 5% chance the baby would come to term and if the baby comes to term it wouldn't last a year. So I had the baby terminated the next week. The termination was emotionally horrible, but the healing of my body and all that was quick. I got pregnant as soon as we could have sex again, right away, and it was probably too soon for me, so I had a miscarriage then. And then it took five months, which isn't that long, but at the time that was the only thing that was going to get me over the termination. I made my husband get his sperm tested, I got tested and everything because I was consumed with getting pregnant.—Amy, 38, Portuguese

The women who shared their feelings about abortion and prenatal genetic testing with me are not different from the women whose feelings about abortion have been

recorded in the literature. The women who had CVS ---thirty percent of the sample---did so because of the timing of testing.[2]

Personal feelings about abortion and religion were not common considerations for the pregnant women I spoke with because most were pro-choice and did not mention religion. However, Susan declared herself pro-life, and yet still had testing.[3] Hilary, who was raised Roman Catholic said her mother and cousins were surprised that she had had PGT, and that she would not tell them if an adverse diagnosis was made because she would probably abort. The overwhelming reason the women I spoke to considered abortion when they had PGT was the desire to prevent births if there was something detected.[4]

PROVIDERS AND ABORTION

Overwhelmingly, the providers I interviewed were "pro-choice", although one medical geneticist described himself as "pro-life" at home and "pro-choice" at work. I found that the providers I spoke with had very specific ideas about when abortion is warranted and when it is not. In one genetic intake meeting at the HMO, I noted that providers question why pregnancies are not terminated when a "severe" diagnosis is made. If the diagnosis is severe in their minds, such as Down syndrome and anencephaly, and the woman decides not to abort, they want to know if the woman is extremely religious. It is not only genetic counselors asking this, but perinatologists, and occasionally the medical geneticists. It seems the providers favor abortion in these cases,

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[2] This agrees with the extant literature (Lippman, Perry et al.1985; Abramsky and Rodeck 1991; Evans, Pryde et al.1993; Pryde, Drugan et al.1993; Zlotogora 2002).
[3] This supports some research (Pryde, Drugan et al. 1993; Singer 1993; Beeson and Jennings 1998; Halliday, Warren et al. 2001) that found that women opposed to abortion do still have PGT.
[4] These findings are also supported by other research (Rapp 1999; Borsack, Metzenberg et al. 2002).

and seek a rationale from those women who have not followed the diagnosis with termination.[5]

Many providers reflected on the ways women make abortion decisions, some framing it the way they perceived women deciding.

> Whether or not you chose prenatal diagnosis mostly depends upon your worldview side. It weighs heavier. The worldview side is where you start to bring in the religious views and the pregnancy termination. In the sense that that determines whether the other side—the abortion—is even an option. If you believe that pregnancy termination is murder, you're not going to ask for prenatal diagnosis, because you just don't want to deal with it. Jacob~medical geneticist, 42, refused race/ethnicity

> In this sort of field, I just think there are so many decisions that have so much personal investment in them dealing with life-and-death situations and making decisions about continuing or not continuing a pregnancy. A lot of religious and personal feelings are involved, people's own social consciousnesses and personal beliefs that go with and make the decision that they are ultimately going to have to live with. It's hard. Elaine~genetic counselor, 53, North American/European, Jewish

> Families love directiveness. Part of the reason people want to be told what to do is that they don't want that responsibility. It's an awful burden to live with, that that choice to abort. Everyone relies on medical people to be the experts on medical information, so if you're saying this is a genetic condition we don't really know what it is, how helpful is that? I think abortion is an incredibly difficult process, even when it's a chosen abortion for your own reasons in the first trimester. We're talking about second trimester abortions because the baby has possible problems. There are lots of layers of things that can really haunt people for the rest of their lives. Clarissa~genetic counselor, 35, North American

These providers have ideas about how pregnant women frame the decisions that they make about abortion. They believe the women incorporate cultural, social and personal

[5] The appropriateness of abortion for specific anomalies is examined in the literature, which found that genetics professionals do have strong opinions about when women should terminate and when they should not, including that women should terminate Down Syndrome fetuses and those with other "severe" genetic disorders (Wertz and Fletcher 1989; Wertz, Fletcher, et al. 1990; Wertz 1993; Wertz 1993a; Wertz 1997; Wertz. and Fletcher 1998; Veach, Bartels et al. 2001; Rabino 2003).

ideas about religion, abortion, and responsibility, weighing them and making definite decisions in light of those beliefs. I believe this conceptualization itself makes the genetic care providers feel more confident about the work they do. They believe the women go through complicated, multi-faceted decision-making processes as opposed to quickly deciding to abort because they feel the resulting baby will not be good for their families.

At a quarterly genetics meeting at the HMO, there was a discussion about the availability of abortion, noting that at three HMO facilities, the providers did not want to provide abortions, so women who wanted to terminate their pregnancies had to travel to another HMO facility to have an abortion. The consensus of the representatives of all the HMO facilities was that abortion was a full service of the HMO. With that status, the providers are responsible to provide it regardless of personal preference. This feeling that providers must practice ethical genetic care is echoed by Jacob and Clarissa:[6]

> Although I tell women they can terminate their pregnancies, I fall very much on the right side of the political spectrum. In my personal life, I am very much a right-to-life person who believes abortion is murder. Some of my friends think I'm a real hypocrite because I don't tell women that. Outside of this office, I think it is entirely reasonable for me to try to convince those women otherwise. But I'm not seeing these people in my personal life, I'm seeing them in my professional life. Jacob~medical geneticist, 42, refused race/ethnicity

> That's the whole crux of prenatal testing, not abortion, but giving them the option. That's what people relate to about playing god. It's that telling them the results is not just telling them what to do with them. Clarissa~genetic counselor, 35, North American

[6] How the personal preferences of providers play a role in the availability of medical care for pregnant women is found in the literature as well (Bernhardt and Bannerman 1982; Press and Browner 1993; Richards and Green 1993; Bernhardt, Geller et al. 1998).

The difficulty in making a decision about aborting from a genetic diagnosis is summed up by Clarissa, noting that without the "baby" there to judge whether it's the kind of person that fits with the family, these decisions can be next to impossible to make:

> No one wants to deal with the gray areas. Part of that is because we don't want that responsibility. But it's telling them [the diagnosis] and asking them to make a judgment and decision on that particular condition before they've ever had a chance to deal with what we're talking about. They don't have that child there. That's an awful burden to live with, making the choice for abortion. It's a pretty unique form of responsibility. The only other thing that I can think of is deciding to take someone that you love off life support. This opens up a whole realm of decision making that I really feel people have never had to deal with before. Clarissa~genetic counselor, 35, North American

The "burden" of making the choice for abortion that Clarissa mentions is a personal perception of what a pregnant woman who chooses to terminate might feel. But Clarissa's framing of the decision making is in line with Rapp's (Rapp 1999) characterization of women who have prenatal genetic testing as "moral pioneers." These types of decisions that shape the family are loaded with personal and social consequences, and must be recognized as such.

In sum, abortion is the main technology through which PGT enables women to shape their genetic families. Yet, alternatively, knowledge of a genetically abnormal baby about to be born could also allow the pregnant woman to shape her family in terms of preparation. Pregnant women struggle with the thought of aborting desired fetuses, and some cannot even face the idea. But if an abnormal diagnosis is made, termination often occurs. Providers tend to favor abortion for what they term "severe" diagnoses, and often attribute refusal to terminate to religion or some other variable that would explain

why a woman would not abort a fetus that is "bad" and incompatible with a quality of life acceptable to these providers. I argue that the conceptualization of abortion as a mechanism to shape one's family in our society is facilitated through the biomedicalization and geneticization of American culture. The first step is usually the construction of family as genetic, accomplished in part through meeting with a genetic counselor to produce a family tree.

BRAVE NEW GENETIC FAMILIES

Genetic Providers as Assistants in the Shaping Process

This new kind of genetic family is different *in life*. The parents know things about their own and their offsprings' genetic constitutions that historically most families have *not* known, or certainly have not known in such detail. It is in the shaping of family members, what types of people are and are not deemed acceptable, where the genetic aspects are important. Such families can be contoured through PGT. Genetic care providers play a crucial role in this shaping of family because as I have shown, they are the ones providing information to pregnant women about the types of fetuses they carry, and what these fetuses will become. Robert, a medical geneticist, acknowledged the "emotional problems and grieving response" that is created in a family when given a genetic diagnosis, as well as the way society has been affected, through people selecting particular types of children and also selecting "against having children with serious problems." Jacob, also a medical geneticist, suggested that genetic knowledge "changes people's lives and/or perceptions of themselves." Genetic care providers' feelings about family and what genetic disorders are difficult to deal with or are incompatible with life shape the information they convey to pregnant women.

> And you know that's what's hard, because sometimes people really do push my buttons, "Oh, absolutely not. We can*not* deal with *that* in *my* family." Clarissa~genetic counselor, 35, North American

> Working with these high-risk breast cancer women, it does make you appreciate not being in a family like that. It makes you appreciate your health and the health of your family. Jane~genetic researcher, 32, European

> Well, obviously the bottom line idea is that you're culling out abnormals. And the general feeling of genetics people is that you should abort these abnormals. I have 4 children. I couldn't imagine raising a baby that I knew was abnormal. There are so many difficulties that it causes in people's lives: marital break-ups and separations and anxieties and lifetime difficulties. I think it's really a service to be able to separate that out. Donald~obstetrician, 42, North American/European

Not only do personal feelings affect how and what information is given to pregnant women, but genetic care providers also question their practice of genetic medicine when women are given genetic diagnoses. Genetic medicine is never clear-cut, even for genetic care providers. Some providers even question the utility of genetic testing for pregnant women.

> This was when we were testing for the BRCA1 gene in very high-risk families, 20 cases or more of breast cancer in a family. The older sister who had had breast cancer manipulated her family, her two sisters who had not had breast cancer. They had testing at the oldest sister's request, and decided, with the older one urging them on, to get the results together. The middle sister turned out not to have the mutation, and she was the one who really should have had it, because she was married, had her kids. It was the younger one who carried the gene, who was not married. This was a Catholic family for whom getting married and having children was a major part of who you were. And when we told her, she was very quiet. She clearly understood what the story was, but she wasn't emotional. The other two were weeping and very upset. The youngest sister then decided what she needed to do was find herself a husband, have a couple of children and have her breasts and ovaries removed. And she spent the next couple of years trying to do that. She had a really difficult two years, during which she did not find a husband and have children. Then she was diagnosed with stage four ovarian cancer! And you wonder whether the screening was such a useful process for her. Nobody could've predicted she was going to get ovarian cancer so young. It seemed that we were opening doors for her and

> that we were giving opportunities, and after all that I don't know that we did. Jane~genetic researcher, 32, European

Other genetics professionals feel thatr the responsibility for decision-making with the genetic information they provide may be too much for pregnant women. There is a hint of the idea that women may not be able to interpret the information in a way that enables them to make the right decisions here. There also is some acknowledgement of the conflicting interpretations of the situation by the provider and the pregnant woman, in which case the pregnant woman's decision stands.

> I had a patient who had a fetus with a malformation of the lung. The prognosis was excellent, and the couple chose to terminate. It was a desired pregnancy. I found that difficult because I felt very conflicted about having found that for them and offered the whole spectrum of options including termination. They made that choice. They wanted the pregnancy until they found out it had an abnormality. Jared~perinatologist, 42, white

> One of my biggest issues is that people made decisions to end pregnancies based on things that I wouldn't end a pregnancy on. And it's sort of hard to deal with. One of the hardest situations was sex selection for me. I had an Indian couple tell me that they would definitely abort the baby if it was a girl. As an Indian American it was really hard for me because I don't subscribe to a lot of those beliefs. But I came to terms with that because I think that for a lot of couples, the sex of the child is as bad as having Down syndrome or another disability. I certainly don't think that I can judge whether one situation is right or wrong. Isabel~genetic counselor, 25, Asian Indian

The genetic researcher I spoke with outlined the eugenic facet of shaping families through prenatal genetic testing.

> We would like to think in some sort of way that you would improve the lot of people if you could weed out people who are not perfect. But I don't think that's actually true. I think that evolution has carried on very effectively without our having absolute control over it. I don't think we've done a very good job of determining what are good characteristics and what are bad characteristics so far. It's very terrifying, of course, the idea of being able to decide on the features of people that you would choose to maintain in your population. Jane~genetic researcher, 32, European

Jane's opinions seem to ignore the reality of extant PGT and the shaping of genetic families based on sex, disease status or disability through abortion. I am not arguing that the current state of PGT is not useful for pregnant women, as should be obvious by my own story of using these technologies. However, I do not believe the providers or pregnant women are willing to explicitly identify it as shaping the family based on personal preference. This framing connotes a eugenic facet of PGT that makes most people uncomfortable. While "personal preference" is socially and culturally constructed as acceptable, it is an individual choice that allows women to chose the genetic acceptability of their fetuses.

Family considerations are also a focal point for genetic professionals in shaping the family through PGT. Family was mentioned in every interview I did with genetic care providers, and most acknowledged the complicated definitions of family in genetics. The genetic researcher I talked with said it best:

> In genetics, particularly in these high-risk breast cancer families, the unit is no longer the person, the unit becomes the family. And there is a whole gamut of complicated issues that arise from dealing with a number of related people as opposed to an individual. Like discovering non-paternity in a family, which certainly comes up. Jane~genetic researcher, 32, European

This argument, that family is the unit of examination in genetics, is precisely Finkler's (2001) point. Finkler believes that future genetic medicine will incorporate families physically into medical decision-making rather than just factoring in family genetics as practiced presently. This could include making genetic decisions only when multiple family members are present and requiring blood testing from a number of family members before allowing testing.

Elaine, a genetic counselor, provided an anecdote of the coping aspects of an adverse genetic diagnosis through PGT for shaping a genetic family through preparation and planning rather than abortion:

> A woman had a pregnancy diagnosed with a sex chromosome abnormality (XXY male) and she continued. She didn't want this to be in his medical record—mainly to avoid him being stigmatized in a school program—but she continued the pregnancy. It was the type of sexual abnormality that nobody would necessarily be aware of if it hadn't been found through amnio. He's sixteen now and he's doing great. He has had learning problems in school, and she does see differences between him and her other son. She has been able to make changes in his school for him without letting them know his chromosomal standing. She said it was just so hard when she got this information in the middle of her pregnancy, figuring out how to accept this information, how to interpret it. And it *is* hard for people. People aren't anticipating this kind of result when they come in for amnio. Elaine~genetic counselor, 53, North American/European, Jewish

My assertions about the work tasks of genetic care providers place them in the center of shaping of the family: pregnant women use the skills of the genetics professionals to make their families what they want. If the fetus does not lie within the parameters of genetic acceptability for a family, it is aborted. This may sound extreme, but in reality, this process is followed over and over again by pregnant women and genetic care providers during PGT. From genetic care providers' standpoints many issues are raised: personal feelings about what should be done in particular diagnoses; frustration at pregnant women's lack of understanding or different interpretations of diagnoses; blocking the somewhat eugenic nature of the job of genetic medicine; evaluating the family as the unit in genetics, especially prenatal genetics; and the feeling of satisfaction at the good the skills of genetic medicine can provide families when they can avoid the potential tragedy of adverse genetic diagnoses.

PREGNANT WOMEN AND PGT: SHAPING FAMILIES

While the genetic care providers bear responsibility for the information they convey to pregnant women, the women themselves are the ones who may live everyday with the good and bad of life with a child with a genetic disability. The decisions about what kind of family member will fit with a family are unlike most decisions one is forced to make in a lifetime. The women I interviewed all felt very strongly about this issue, and were clear about the ways they had struggled with making decisions about what type of fetus they would consider terminating. The women mentioned family considerations including factoring other children into the decision to have testing, the wish to avoid a child with disabilities that would complicate family life, and parenting responsibilities to the unborn child. The women who were hoping to avoid disabilities in their fetuses had very specific, family-based definitions of which genetic diagnoses would not be acceptable for their families:

> With my first two pregnancies I was hoping to be prepared, if the child needs a lot of special care. If you don't have a physically and mentally flawless child from the onset, you know what you're kind of getting yourself into. This pregnancy is very different. Now, we've been living with Peter [a nephew] and all of his special needs. They figured out there was something wrong with him in the past five years. We see the hardships on the family. I was like, I know this pregnancy is going to be Down's or some abnormality that's going to cause the rest of its life to be very difficult, and that's it. I knew that I would terminate the pregnancy, so it was a very different expectation.—Jennifer, 35, Asian

> My husband was very concerned about having a healthy baby. He teaches children and said, "It's really, really hard to raise a child that is not a healthy child. So we talked about it, and we decided to go for the screening.—Sotiria, 37, Greek

> I'm sure we would have probably had to go talk to a Rabbi or something, you know, and really get some help making that decision. The clearer the data, the clearer our choices would have been. I'm sure if our kid was going to have Tay Sachs or something, we would have terminated. We wouldn't have

> to really make a big debate about it. Similarly, if we knew the child was going to be mildly retarded or something, obviously, we wouldn't. We would have terminated for Down syndrome, even though I know Down's children are very easy to love and I would have been torn about it, but I probably would have done it.—Robin, 33, Jewish
>
> We talked about it, and neither of us thought we could handle it. It was an awful thing to realize about one's self. You hear these wonderful stories of people who have these great kids, but we're just really scared. We're *so* scared.—Heather, 33, Caucasian

Jennifer's concern was for the child that might suffer from disabilities while Heather, Robin and Sotiria each admitted their focus was more on themselves and their partners and how a genetic disorder would affect them.[7]

The women who cited other children as the reason they wanted PGT were thoughtful about how their existing children's lives would be altered with a sibling with disabilities. Weighing the responsibilities of the children without disabilities to care for those with genetic disease after the parents were gone was a concern, but also how the financial implications of a child with genetic disease would alter family life.

> In response to a question about what the family of the nephew was like, Jennifer said, "Our family has a lot of kids, and we get together a lot. We see the kids progressing every year and we see Peter not moving forward. I think they initially wanted a third child. They have an older daughter and they didn't want to leave her with the possibility of taking care of Peter. They wanted to have a normal sibling for her. But I think at this point they've ruled that out because of the risk to have another child like Peter.—Jennifer, 35, Asian
>
> Nissa, 36, Mexican, Irish and Jewish, discussed her first child and how she and her husband had wondered when deciding to have the amnio, "Were we ready to sacrifice his life in that way?" referring to her son and his potential responsibility to care for this "other child" if it suffered from Down syndrome.

[7] Junegst (1999) would call Heather, Robin and Sotiria's usage of PGT a pursuit of familial rather than fetal security. This framing is palatable because the genetic care providers suggest their work is a service to prospective parents to enable them to avoid the burden to the family of more than they can handle, implying a genetically abnormal member of the genetic family is too much to bear.

> We were looking at the really life altering aspect of it—financially devastating. We thought we couldn't do that, so we needed to know. And of course, we had to worry about it because we had both baby A and baby B.—Heather, 33, Caucasian (carrying twins)

> Amy, 38, Portuguese, told me the story of a friend who found out she was carrying a Down syndrome baby and made the choice to continue the pregnancy. It wasn't their first child, and it was a huge struggle. "She did a lot of research on it. They were only going to have two children, but they made the decision to have three, because she doesn't want him to be left alone."

Parenting responsibilities to the fetus were another reason cited by women who thought they might shape their families through PGT. Tasha chose to terminate a Down syndrome fetus and rationalized that decision saying, "it just wasn't right to have a child knowing what we knew" implying that Down syndrome was not an acceptable fit for a child in her family. The power of making the life or death decision for the fetus was overwhelming to Rox. Kate framed the decision to have PGT as a parenting decision, implying that amnio was beneficial to the fetus, despite the connotation that it could end its existence.

> All of that stuff is just really not in your control. And it was weird to be making this big, sort of powerful decision over someone's life.—Rox, 38, Arab

> I think maybe (having amnio) is sort of like you make one of the first parenting decisions early, you know, and in a sense it feels like a premonition of what's to come.—Kate, 35 Jewish

Families are different today than before the genetic technological revolution in biomedicine. How families are defined depends upon the members of particular families, but many families now are genetic families, shaped through technomedicine. The major mechanism through which genetic families are created and maintained is prenatal genetic

testing and screening. Once PGT has been conducted, abortion is the only available technology utilized to prevent fetuses that will not fit with a particular family from being born. Abortion per se is a heated issue in American culture without the added complexities of choosing which types of fetuses are acceptable and which are not. Biomedicalization and geneticization of American cultural values enables pregnant women who opt to have PGT to more comfortably make these difficult decisions within the confines of acceptability and normativity. While there is nostalgia for the days when pregnant women did not have to face such choices, the overall feeling about the availability of PGT as a technoscientific means for shaping one's family is positive. After an adverse genetic diagnosis, pregnant women and their families make judgment calls on what types of genetic disorders are acceptable within their families, and make abortion decisions based on those parameters. Thus, I found that genetic families are shaped through prenatal genetic testing.

9 CONCLUSIONS

THE EXPERIENCE OF PRENATAL GENETIC TESTING & PRACTICE OF PRENATAL GENETIC CARE

Many pregnant American women today make decisions daily about right and wrong for their fetuses, from what to ingest to what they must purchase before birth. Through the biomedicalization of health (Clarke, Shim et al. 2003) American culture clearly demarcates what is and is not condoned during pregnancy. My research examined one of the life-altering decisions many pregnant women make: prenatal genetic testing. Sometimes this decision is made before her kith and kin are even informed or can mark her as "pregnant." This research discerned, largely in support of the literature reviewed in chapter 2, that the accessibility of PGT depends upon the type of insurance the woman has, her geographic location, the type of obstetric provider she uses, and her personal motivation to utilize these technologies, among others. I also found, like others who have studied PGT, that the decision to have PGT is fraught with many anxieties including whether to have testing or not, what the testing experience will feel like, fears of risks of miscarriage from the procedure, receiving the results, and deciding what to do regarding abortion if something abnormal is diagnosed.

My research also contributes to the small amount of published literature on prenatal genetic care providers though examining how they manage, practice, and feel about the work that they do. I found that genetic care providers believe they provide a useful and necessary service to pregnant women. They determine, through an assessment of the individual woman, exactly how to tailor the genetic information they deliver in a way they believe is most suited to her specific situation. They diligently work to clarify

complicated scientific scenarios into realistic, available information about the capabilities and limitations of PGT to detect genetic disorders. This is accomplished in part through creating what I call the genetic family by constructing a medical pedigree of three generations for both the pregnant woman and her partner.

My research revealed that non-directiveness, the established framing of the appropriate mode of practice for genetic care providers, was *not* a useful representation of the actual tailoring of information work these providers do. This finding is supported not only by providers themselves, but also by the experiences of pregnant women who have PGT (see chapter 6). In the abstract, genetic care providers following the non-directive mandate would present the information in a unbiased manner so that the pregnant woman could make an informed, yet personal and uninfluenced decision about PGT technologies based on interaction with her partner and anyone else she chooses to include. The assumption in the medical genetics field that providers adhere to non-directiveness is proven false by my data, not only through the fact that some genetic providers do not attempt to *always* be nondirective, but also because the idea of nondirectiveness is challenged by providers and the very practice of providing PGT care. The line between being directive and non-directive moves—is constructed only in the moment of practice. That is, the information presented in a prenatal genetic counseling session is potentially always already directive, merely by being presented in that setting. It exists precisely so that pregnant women can make judgments on the types of fetuses they ultimately deliver as babies, selecting out those deemed unacceptable.

Moreover, through tailoring, the genetic care providers themselves filter the information they provide to pregnant women based on an assessment they make

including their understandings of what the woman plans to do with the information, whether she wants merely to be informed and prepared in the event of a genetic diagnosis, or if she intends to abort a genetically abnormal fetus. This is important because it refutes the construction of the informed consent process within the field that is currently framed through the non-directive tenet that providers should present all the information available. The information is sometimes consciously filtered by the providers, so it is not *all* of the information. Further, some pregnant women are truly exasperated by the providers who do practice without *overt* direction, expecting a more traditional model of medical providers suggesting modes of action for a particular problem. My findings suggest that even if providers are not obviously directive, the ways they assess the pregnant woman and her partner and make choices about the information they provide to them are in fact directive in some senses.

THE MATERIAL AND DISCURSIVE (RE)CONSTRUCTIONS OF GENETIC BODIES AND GENETIC FAMILIES

I argue that gendered technologies, such as prenatal genetic testing, may have enormous consequences, one of which is the formation and inscription of a "genetic body" and corresponding "genetic family" for pregnant women and their relatives. Women's pregnant bodies are under observation by the medical community and society at large, and are marked through the destabilizations and disruptions of pregnancy and prenatal diagnosis. Normalization (Balsamo, 1996; Sawicki, 1999; Shildrick, 1997) of prenatal care is accomplished through biomedicalization and geneticization. This is where my conceptions of genetic bodies and genetic families rest. These entities exist only through genetic technologies, and are created by the corporeal experience of the

woman utilizing prenatal testing technologies. The uniqueness of the genetic body and the genetic family is that it is only "genetic" once the genes have been examined, assayed and compared to "normal". Interestingly, even if the genetics are "normal" the "genetic" label is there, because the knowledge of genes has been produced and consumed. The technologies of karyotyping and assays are the vehicles of the genetic body and its corresponding genetic family. Constructions of genetic families are created and destroyed through the experience of PGT, by definitively identifying blood-related members who share DNA and those who do not but may have believed up to that point they did.

This research extends Clarke and colleagues (2003) argument that new technoscientific identities produced through technoscientific means are today more commonly produced in the West, asserting that through biomedicalization and geneticization, the individual technoscientific identity of a genetic body and the *collective technoscientific identity of a genetic family* are produced through the technologies of prenatal genetic testing. These individual and *familial* transformations are enabled through the availability of testing to determine who's who in a "family"—blood members and not—and who does and does not have genetic ties and/or genetic abnormalities.

The experience of PGT implicates both bodies and families, despite the reality that the bodies and families had these genetic pedigrees before the testing occurred. The embodiment of a genetic body is facilitated through the technologies of genetics, and the experience of a genetic family is similarly enabled through PGT or other genetic testing technologies. Primarily through clarifying the relationship between the lived experiences of women and their families in prenatal genetic testing, and correspondingly examining

their production by the (bio)medicalization/geneticization of pregnant bodies and families, the bodies, selves and families of pregnant women become understood as both material/corporeal and discursive/represented/produced/created. My point supports the larger argument by Novas and Rose (2000) that life itself is now explained and intervened through molecular optics enabling a mutation of selfhood that challenges traditional notions of embodiment.

The embodiment of women's genetic bodies can be most clearly understood through an examination of the material experiences of the body as it is being embodied and the discursive construction of the body as being embodied. Self-reflexivity is the theoretical panacea for the disruption of corporeality experienced by the diagnosis of a genetic body. It is through self-reflection that a genetic diagnosis is accepted as self, a genetically defined body in the self that is, has been, and will be "normal" to self, whether defined as abnormal or normal through genotype. Thus the creation of a genetic body. Embodiment is individually articulated, but influenced culturally, socially and epistemologically. The pregnant women I interviewed incorporated pregnancy into their self-identities through their experiences of embodied knowledges. The most obvious re/constructions of self in relation to pregnancy and PGT were the examinations of "bodies," "health" and "age" that are discussed in chapter 7. Rose's (2001) suggestion that molecular vocabularies of ourselves are *becoming* self explains how the women I interviewed could challenge their embodied understandings of health, age and bodies when presented with the omnipotent genetic knowledge of American culture today.

American cultural constructions of genetics, especially prenatal genetics, as a panacea for preventing disabled babies enables individuals to feel abortion after a

positive PGT diagnosis is not only acceptable, it is normalized and condoned through the institutions of biomedicine with the legitimation of the state via research support, public health authority to offer testing, and other mechanisms. Genetic essentialism (Nelkin and Lindee 1995) is revealed through my research as remarkably applicable and in practice today. Most obviously, the practice discourses of the providers represent genetic essentialism in its purest form through the omniscient gene ideology/discourse. The appeal of the personal interest stories described in chapter 2 is in part the ambiguous nature of genetic information, but never is the genetic aspect dismissed as irrelevant—it remains the center of the discussion. Some women are so convinced of the omniscience of genetics that if the diagnosis from PGT falls in the gray zone they are *more* inclined to abort (Pryde, Drugan et al. 2001).

Through the routinization and normalization of PGT, American culture has effectively condoned the use of prenatal genetic testing technologies and abortion of genetically abnormal fetuses. The transfer of responsibility for the type of child born from the biomedical community to the pregnant woman is facilitated through the geneticization, medicalization, routinization and normalization of such testing. Suter's (2002) recollection of people asking her, as a pregnant woman over 35, not did she have testing but which tests is an indication of the pervasiveness of the reach of genetic care. This supports my point that there is an assumption, particularly prevalent among the population of middle and upper-middle class, educated women, which encompasses my sample of women, that one has PGT to prevent births of unacceptable babies through abortion. The quotes from women in the "biomedicalization and geneticization" section of chapter 5 further illustrate this.

Throughout this dissertation I have argued that pregnant women who choose PGT, with the help of genetic care providers, make decisions about the types of fetuses that are an acceptable fit for their families and those that are not. The fetuses deemed unacceptable based on genetic diagnosis provided through PGT are intended to be aborted by some pregnant women. Abortion is the only available mechanism to prevent the births of unacceptable babies when PGT is the technology of identification. Appendix A lists and explains other genetic technologies such as IVF with PGD that avoid abortion, but these technologies have their own problematic issues. Rapp's (1999) conception of pregnant women who make these complicated and multifaceted judgments of their desired fetuses as "moral pioneers" encompasses the weighty individual aspects of the PGT decision-making process expressed vividly by the women quoted in chapters 7 and 8.

This research supports Rapp's constructions of women as moral pioneers and adds to her conclusions by revealing the potential cultural shift represented by the American cultural acceptability of pregnant women shaping their families through PGT. The existence of PGT technologies is intimately linked with the availability of abortion, represented in chapter 8 most eloquently by Sydney who said, "I suppose if the decision whether to keep or terminate wasn't predicated on the outcome of such testing we wouldn't have done it." Pregnant women have very specific parameters outlining what diagnoses are acceptable and which are not and why. Genetic care providers also have particular criteria regarding when abortion is warranted. When these personal constructions are challenged by individual women's choices, providers feel frustration. This occurs whether it was a woman not terminating a fetus when the provider believed

she should or a woman opting to abort when the provider though she should not. Important in these scenarios is the primacy of the pregnant woman's decision about the future of her fetus and, correspondingly, the future of her family. This represents the co-constituitive nature of genetic bodies and genetic families, through interactions among genetic professionals, the women who utilize PGT, others with whom they interact, and pertinent discourses available in the broader culture.

My construction of families is families as genetic entities, impacted in some way through genetic identification, be it constructing a medical pedigree in a genetic counseling session or through identifying a balanced translocation or a disease gene through genetic assays. Even if nothing genetically abnormal is detected, the family is forever aware and educated on its genetics. Finkler (2001) theorizes that shared genetic inheritance between family members forges a lasting bond with blood kin, even if there is little else in common between family members. During the construction of the medical pedigree, kinship relations inform medical pedigrees just as the medical pedigrees embody kinship (Nukaga and Cambrosio 1997). Medical pedigrees also (re)construct genetic families into entities with a genetic past, present and future (Rose 2001). Novas and Rose (2000) argue that this genetic connectedness in families and the complexities of it comes in conjunction with the burden of mutual obligations and caring that exists in the traditional nuclear family. My discussion of medical pedigrees in chapter 6 revealed not only the importance of accurate information for the providers, but also that pregnant women's identities were altered through their creation. My research clearly revealed the co-constitutive nature of the experience for the pregnant women and their partners.

These women were materially on paper and discursively through the counseling session reconstructed as genetic bodies and their families were represented as genetic families.

The process of (re)constructing bodies and families through PGT has potential to be stigmatizing. I argue that one of the consequences of (bio)medicalization and geneticization of American culture is that *genetic status* has become a stigmatizable entity, establishing a "virtual social identity" (Goffman 1963), a "technoscientific identity" (Clarke, Shim et al. 2003) vis-à-vis genetics, a "somatic identity" (Rose 2000) that can also be the basis for "biosociality" (Rabinow 1996). These new identities are consequential for stigmatizing processes. Individuals devoid of physical manifestations, but who possess mapped mutations can now be distinguished, thus possibly stigmatizing those individuals who are established carriers of genetic mutations.

This is most clearly illustrated in my research in the final section of chapter 8 where the women outline what was and was not genetically acceptable in their fetuses. Fitness was determined by the women I interviewed on the genetic identity of the fetus as "normal" or something other than "normal". While there was a range of acceptability, many women expressed a desire for a mutation-free fetus, effectively stigmatizing all genetic abnormalities even if there was no phenotypic expression. To further support this point, the gray zone discourse among providers exemplifies the potential stigmatization and ambiguity of *any* chromosomal or genetic rearrangement that does not have a specific diagnosis attached.

The ways genetic care providers and pregnant women work together to shape genetic families through PGT technologies is the most salient finding from my data. The goal of PGT is to provide women "options" when faced with having a child with a

genetic disorder/disease prenatally detected, and the only option available is abortion. Through the PGT technologies available, pregnant women and their families can determine *to a point* whether their fetus is genetically acceptable to their family. The process of PGT creates genetic bodies and genetic families through the identification of individuals and families as genetic entities, shaped and determined by what the genetics of their cells indicate, no matter how tenuously, about the possibilities of genetic risk to offspring and themselves.

FUTURE RESEARCH AGENDAS

A missing link in my research and the literature I surveyed could be provided through in depth study of obstetricians and their role in prenatal genetic testing. Women often referred to their obstetrician as part of their families, and relied on her/him for trustworthy advice in many instances. Yet most obstetricians do not have genetic training. This is one of the critiques of genetic medicine from genetically trained professionals. (See Appendix C for a discussion of the need for more genetic training in related specialties.)

Another relatively unexplored facet of this research is the partners' contributions to the decisions about shaping the family. All of the women I interviewed mentioned their partners at some point in the interview, but most said they deferred to her in the final decision. The published research on this topic does not clearly illustrate the level of involvement in the deicison-making process and whether partners prefer to be included on the emotional, anxiety-ridden level of abortion decisions or if they feel more comfortable passing on the ultimate responsibility to the pregnant woman.

The genetic families I write of here are on the cusp of a new wave of pregnant women's potential parental responsibilities. New reproductive technologies, including IVF, preimplantation genetic diagnosis, *Microsort* sex selection and other pending techniques promise a much more challenging decision-making process for the eventual tailors of our society's genetic bodies and families.

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APPENDIX A PRENATAL GENETIC TESTING TECHNOLOGIES

Following is a brief overview of the historical development of prenatal testing, primarily focusing on amniocentesis (amnio) and chorionic villus sampling (CVS). I start by framing the beginnings of prenatal testing. I then discuss early amniocentesis, molecular tests, serum screening, fetal cell sorting, preimplantation genetic diagnosis and carrier screening, including how each is conducted and its risks and benefits. When the data reported in this dissertation contain references to such tests, I do not explain them there.

AMNIOCENTESIS (DIAGNOSTIC)

According to Cowan (Cowan 1992; Cowan 1993), the first fetal property that could be prenatally tested was sex. In 1960, four separate research groups, two in Europe and two in the U.S., were credited with applying feline research techniques to human amniotic fluid to determine the sex of a fetus (Cowan 1993). The amniotic fluid was removed using amniocentesis. This procedure involves inserting a needle through the abdomen into the uterus and withdrawing a small amount of amniotic fluid. (Today needle placement is guided by real-time ultrasound.) Almost immediately, genetics became involved, because this kind of testing could be used to indicate the presence of sex-linked diseases in a fetus. A sex-linked disease gene is located on a sex chromosome, either an X or Y. Hemophilia, for example, is passed through an X-linked recessive pattern, where males with the gene will be affected because all the genes on their single X chromosome will be expressed. A woman with a history of hemophilia could test the fetus through this technique, and if it was male and she was unwilling to have a child suffer from the disease, she could abort. The small number of women who

could use amnio for diagnosing sex-linked disorders increased gradually during the next decade as amnio became known as a method of detecting chromosomal problems in a fetus (statistically more likely as the woman ages).

The limitations of amniocentesis as a genetic test used only for sex-linked disorders were addressed in the next decade by three advances: (1) the development of media to culture cells; (2) karyotyping of cultured cells to reveal chromosomes; and (3) legalizing abortion to prevent the birth of a fetus diagnosed with a genetic disorder (Cowan 1992). The development of media to culture cells in 1967 provided the possibility of karyotyping, which could diagnose disorders in chromosomes. In 1959, a common type of Down syndrome was determined to be caused by the presence of three copies of chromosome 21, diagnosable using karyotyping. Abortion, legal in Scandinavia for many years, became legally available in the U.S., Britain and Canada during the time amnio was being tested for safety in the U.S. Wrongful birth lawsuits in 1978 and 1979 were won by parents who claimed that their obstetricians had failed to refer them for amniocentesis which could have prevented the births of their severely disabled children (Cowan 1993). Together these developments produced amniocentesis as an important prenatal genetic test.

As early as 1974 (Pearson 1974), and officially in 1983 by the American College of Obstetricians and Gynecologists and the American Academy of Pediatrics (ACOG and AAP 1983), physicians caring for pregnant women were advised to offer or refer their patients for prenatal diagnostic services. By the time amniocentesis was endorsed by the medical community, the key factors were in place to make it a desirable test for pregnant women. Amnio is usually performed between 15 and 20 weeks gestation. It is more than

99% accurate, according to two multi-center studies (NICHD 1976; CEMAT 1998). Amniocentesis has a miscarriage rate of 1% (CGDB 1995a) or 0.5% based on a recent review (Eisenberg and Wapner 2002) and a prominent obstetrics textbook (Cunningham, Gant et al. 2001). The fetal loss rate (miscarriage rate) varies by provider experience and facility, and has been attributed to preexisting abnormalities in the fetus and/or uterus including placental abruption, abnormal placental implantation, uterine anomalies and infection (Cunningham, Gant et al. 2001). Other risks of having this procedure are an association with decreased lung growth, and development in utero and after birth of respiratory and pulmonary problems (for a list of studies see (Wilson 1995)).

Today amnio is routinely recommended for women who fall into a set of defined categories. The most common category is "high risk:" here risk of fetal aneuploidy is greater than the fetal loss rate associated with the diagnostic procedure being considered. "Advanced maternal age" categorizes one as "high risk" and is related to increased risk of Down syndrome and other chromosomal abnormalities complicated by age. It generally means a woman over 35. " Advanced paternal age" is not considered a factor because there is dispute about what constitutes the diagnosis. Numerical risks for Down syndrome and all aneuploidies (chromosomal or genetic abnormalities) differ based on the age of the pregnant woman and the source of the information.

Table AA. Down syndrome risk by age

The first provider to see a pregnant woman for prenatal care is the one who usually refers her for prenatal genetic testing. In a recent nursing publication (Kenner and Dreyer 2000:629-630), amnio was recommended for the following:

- carriers of a metabolic or autosomal recessive disorder;
- advanced maternal age;
- maternal history of infertility, stillbirths, or multiple spontaneous abortions;
- previous child with chromosomal abnormalities, NTD or congenital anomaly;
- family history of a known parental balanced translocation;
- family history of mental retardation;
- parental (either) exposure to teratogens before or during pregnancy; and
- abnormal ultrasound or AFP results.

Interestingly, the leading obstetrical textbook frames the qualifications as "Women with Risk of Fetal Aneuploidy High Enough to Justify Risk of Amniocentesis" (Cunningham, Gant et al. 2001:975) and lists:

- single pregnancy with age 35 or greater at delivery;
- twin pregnancy with age 31 or greater at delivery;
- previous trisomy birth;
- previous X-linked anomaly birth;
- "patient" or partner with chromosomal translocation;
- "patient" or partner with chromosomal inversion;
- history of triploidy;
- repetitive early pregnancy loss;
- "patient" or partner with aneuploidy; and

- major fetal structural defect detected by ultrasound.

CHORIONIC VILLUS SAMPLING (DIAGNOSTIC)

Chorionic villus sampling (CVS) technology followed in the larger, more successful footsteps of amniocentesis. The advantage of CVS compared to amnio is that it can be conducted much earlier in the pregnancy. Therefore, if a therapeutic abortion is sought, it can be performed at an earlier gestation with a simpler and safer procedure. CVS is generally performed at 10-13 weeks gestation. The CVS technique involves removing a small amount of the villi, which turn into the placenta. It can be done in two ways. One uses a technique similar to amnio where the tissue is biopsied via needle and syringe through the abdomen and uterus (transabdominal TA method). The sample may also be obtained by inserting a small catheter through the cervix into the uterus, taking the sample, and withdrawing the catheter (transcervical TC method). Both types of CVS procedure are guided by real-time ultrasound today.

The first attempts to conduct this procedure occurred in the late 1960's in Copenhagen (Cowan 1993). Many attempts to perfect it followed, most plagued by two serious problems: unacceptably high spontaneous abortion rates and incorrect diagnoses. Between 1975 and 1982 the literature is void of CVS articles. The diameter of the biopsy instrument was the crux of the problem. Since 1982, there have been two international conferences on first-trimester fetal diagnosis and an enormous amount of developmental research has been conducted on CVS (Cowan 1993). A meta-analysis of CVS studies found that the choice of the type of procedure conducted (TA vs. TC) and the choice of instruments is based on operator's personal preference rather than any statistical data in favor of one method or instrument (Alfirevic, Gosden et al. 2001). Cowan (Cowan 1993)

posits that the reasons for the delay of CVS diffusion into the marketplace are varied, but include: competitive advantage of the first test on the market (amnio); more difficult techniques for the physicians learning CVS; higher loss rates by a new CVS provider due to the difficult techniques; and less fetal research funding because of social pressures in the U.S.

Other factors are also important to chorionic villus sampling's development and dissemination. There were issues with congenital malformations including digital and limb defects, oromandibular defects and cavernous hemangiomas in babies born from pregnancies that had CVS. These malformations were attributed to the procedure (Firth, Boyd et al. 1991; Burton, Schulz et al. 1992). More recently, these problems have been attributed to physician error, especially correlated with practitioner expertise related to the number of procedures performed (Modell 1992; Andrews, Fullarton et al. 1994). Kuliev and colleagues (Kuliev, Jackson et al. 1996) showed that the global 10-year experience with over 139,000 CVS procedures conducted after 9 weeks gestation had an incidence of limb reduction defects the same as the background incidence: 6 per 10,000 births. The association of CVS with these various defects caused women to opt for amnio later in the pregnancy rather than incur the risk of such defects compounded by the risk of miscarriage. One study that compared the rate of CVS in a Chicago hospital before and after publicity about the association between CVS and limb reduction defects found a 24.4% reduction in the number of CVS procedures performed (Heckerling, Verp et al. 1997). There is also a greater risk of congenital malformations linked to CVS when it is conducted prior to 10 weeks (Wilson 1995; Eisenberg and Wapner 2002). Unlike

amniocentesis, CVS has not been associated with any increased risk for an abnormal delivery (Cederholm, Haglund et al. 2003).

The difference in physician practice technique, whether transabdominal or transcervical, appears to be related to the risks associated with CVS. When both types of procedure, transabdominal (TA) and transcervical (TC), are examined together, the procedure is considered safe with a miscarriage rate of 0.5% (Eisenberg and Wapner 2002), more sampling and technical failures and more false positive and false negative results than amnio (Alfirevic, Gosden et al. 2001). Another study found the diagnostic accuracy and fetal loss rates to be equal in CVS and amnio (Ammala, Hiilesmaa et al. 1993). A patient education pamphlet cites the accuracy for detecting chromosomal anomalies at 99.9% (GeneCare 2002). One study cited a .03% false negative rate for CVS with a sensitivity (the ability to measure true positive results) of 98.9-99.8% (Hahnemann and Vejerslev 1997). The transcervical method has been found to have a higher fetal loss rate when compared to TA (Papp, Beke et al. 2002). A review study found that TC CVS has an excess fetal loss rate of 3.7% compared with TA CVS or amnio (Wald, Kennard et al. 1998). The transabdominal method did not have rates above normal for preterm delivery or stillbirths associated with it (Papp, Beke et al. 2002), or post-procedure complications (Brambati, Tului et al. 2002). Brambati and colleagues conclude from their study of 1844 CVS procedures conducted between 13-20 weeks gestation that TA-CVS is a valuable alternative to early as well as mid-trimester amnio (Brambati, Tului et al. 2002). An obstetrics textbook also asserts that TA-CVS performed after 9 weeks is as safe as amnio (Cunningham, Gant et al. 2001).

Evans and colleagues (Evans, Pryde et al. 1993) add to the historical account of fetal diagnostic technologies. In the early years of performing prenatal diagnostic technologies, both amniocentesis and CVS, there were limits on the number of providers and labs capable of performing the procedures and processing the cultures. This caused the testing to be "significantly prioritized to patients felt to be at highest risk, and those for whom management decisions would be changed by the data obtained" (Drugan, Greb et al. 1990):71. A "change in management" constituted an abortion for abnormalities. It was felt at that time, that it would be a "waste" of scarce diagnostic resources if the patient were not willing to abort when an abnormality was found. As services became more widely available and the field of prenatal diagnostic technologies grew, the fundamental tenets of genetic counseling began to be applied. Presently, the foremost consideration in genetic counseling is that "testing was for information purposes only and that there is no linkage between what a patient is told about the status of her pregnancy and what she elects to do about it"(Drugan, Greb et al. 1990):71.

EARLY AMNIOCENTESIS (DIAGNOSTIC)

Furthering the search for an earlier, efficient and safer prenatal diagnostic test, early amniocentesis (EA) was proposed in the late 1980's. Early amnio had the purpose of combining the advantages of CVS, early gestational age, and amniocentesis, accuracy and safety (Nagel, Vandenbussche et al. 1998). There are varying definitions of when early amniocentesis is conducted, ranging from 11-12 weeks(Wilson 1995), 11-14 weeks (Nagel, Vandenbussche et al. 1998; Cunningham, Gant et al. 2001) and 10-14 weeks (Daniel, Ng et al. 1998). Accuracy and spontaneous abortion rates vary according to needle gauge, amount of fluid aspirated, gestational age at procedure, and number of

attempts needed to obtain the sample (number of times the needle is inserted through the woman's abdomen into the uterus)(Wilson 1995). Early amniocentesis performed before 13 weeks gestation has been shown to have an increased risk of talipes equinovarus (foot deformities such as clubfoot)(CEMAT 1998; Eisenberg and Wapner 2002). When the risks for maternal complications following amniocentesis were evaluated, one study found that while it was not associated with an increased risk for severe pregnancy complications, women who had amnio before 15 weeks were more likely to have delivery problems. These included abnormal delivery, specifically one using forceps, vacuum extraction, or elective caesarean section, and an elevated risk of complications related to amniotic cavity, membranes and hypotonic uterine dysfunction (Cederholm, Haglund et al. 2003).

Daniel and colleagues (Daniel, Ng et al. 1998) compared the outcomes of women who had EA (10-14 weeks) with women who had amnio (15 weeks +) with the same doctors, techniques, and laboratory. The authors found that EA had smaller samples and thus fewer cells to culture and test, a longer waiting period for test results, more multiple insertions, more bloody samples, and a higher rate of pregnancy loss (2.2% compared to amnio 0.6%). Despite these data, the study concluded that EA was a viable alternative to CVS. Nagel and her colleagues (Nagel, Vandenbussche et al. 1998) compared women who had undergone either EA (11-14 weeks) or CVS (11-14 weeks) for differences in spontaneous abortion and child morbidity. The authors found a spontaneous abortion rate of 6.2% for EA, with no such miscarriages for CVS. They concluded that CVS is the preferred method for first trimester diagnosis. Again comparing EA to CVS, Delisle and others (Delisle and Wilson 1999) found that EA procedures demonstrated a higher rate of

total pregnancy loss, a significant increase in musculoskeletal foot deformities, a significant increase in culture failure rate and an increase in the postamnio rate of fluid leakage.

FISH AND Q-PCR (DIAGNOSTIC)

Molecular testing (FISH and Q-PCR) of the samples taken by tests described above (amnio, CVS and EA) performs chromosomal analysis for the five most common chromosomal disorders and is offered in conjunction with amnio or CVS. These disorders and their incidence in newborns are (Scott, Disaia et al. 1994): Down syndrome (trisomy 21)(1/800), sex chromosome abnormalities (XXX, XYY, XXY)(1/1000), trisomy 18 (1/8000), Turner syndrome (45, X)(1/10,000), and trisomy 13 (1/20,000). These tests provide results in 2-3 days and are conducted using fluorescence in situ hybridization (FISH) or the quantitative polymerase chain reaction (Q-PCR)(Grimshaw, Szezepura et al. 2003). Chromosomal errors other than the top five will not be detected, but the authors of one extensive study support this risk with the data that these other abnormalities are extremely rare (4/1000) and some of these abnormalities could be detected through other means standard in prenatal care, such as ultrasound (Grimshaw, Szezepura et al. 2003). The Grimshaw study used FISH and Q-PCR in 1576 cases and found that FISH detected the chromosomes screened 86% of the time when one of the abnormalities was present, and 99% of the time when there were none of the chromosomal abnormalities present, with Q-PCR the numbers were 82% and 99% respectively (Grimshaw, Szezepura et al. 2003). The accuracy of the tests is good, but not so good that most people would be satisfied without the confirmation from amnio or

CVS results. Molecular tests are presently conducted only in conjunction with one of the diagnostic tests discussed above.

SERUM SCREENING

While the diagnostic tests discussed here are complicated and time sensitive, blood screening tests exist that can be used for most pregnant women with no risk to the woman or fetus. These screening tests indicate risk of open abdominal wall defects (AWD), neural tube defects (NTD—like spina bifida), trisomy 18 and Down syndrome. These tests are the expanded alpha-fetoprotein test (xAFP or AFP), the triple screen, and the quad screen. They are normally conducted between 15 and 20 weeks gestation. The information from these screening tests hypothetically can enable a woman to make an educated decision about having amniocentesis. These tests were developed to screen out women at normal risk and identify those at higher risk than other women in their category for anomalies based on the levels of certain blood serum components.

AFP, Expanded AFP (xAFP or Triple Screen)

The xAFP test has its roots in the maternal serum alpha-fetoprotein test (MSAFP) which tested a pregnant woman's blood for hormone levels linked to AWD and NTD, with a 20% chance of detecting Down syndrome (Haddow, Palomaki et al. 1992). When MSAPF was expanded, becoming the expanded AFP test (xAFP) to test the woman's blood for human chorionic gonadotrophin (hCG) and unconjugated estriol with the alpha-fetoprotein, the test's capabilities for detecting defects was greatly increased (Haddow, Palomaki et al. 1992). The test was then referred to as the triple screen. Because the xAFP is a blood test involving only a normal blood sample drawn from the arm, it confers no risk to the woman or her fetus, and thus can be offered to all pregnant women,

regardless of age. In 1985, the American College of Obstetricians and Gynecologists issued a "professional liability alert" advising obstetricians that it is "imperative" that they discuss the availability of this screening test with their patients and document it in their charts (ACOG 1985).

While these screening tests have the advantage of being noninvasive, they also have many liabilities. Abnormal xAFP results may result from misdating the pregnancy, a multiple pregnancy, or errors in recording the race/ethnicity, diabetes mellitus status, or body weight of the woman, causing false positive results (Andrews, Fullarton et al. 1994). The xAFP test has a potential 25% false positive rate for Down syndrome detection (Haddow, Palomaki et al. 1994), while overall detection of the disorders tested for has a 4% false positive rate (Goodburn, Yates et al. 1994). The xAFP test detects 97% of the cases of anencephaly , 80% of the cases of open spina bifida, 85% of AWD, 50% or more of cases of trisomy 18 and 40%-66% of Down syndrome (CGDB 1995). If the woman receives a negative diagnosis, indicating that there are only normal variations in the levels of the hormones measured, she is not guaranteed a "normal" fetus. It only indicates that she is within normal limits for the birth defects for which the test screens.

Quad Screen

More recently, new blood components have been added to this type of screening to create the quad screen. The test now examines levels of alpha-fetoprotein, unconjugated oestriol, free beta-hCG and inhibin-A between 14 and 20 weeks (Wald, Huttley et al. 2003). Wald and his colleagues found that, for Down syndrome, the quad test represented a 7% false positive rate and an 81% detection rate (Wald, Huttley et al. 2003), much better than the numbers associated with the triple screen. There are

numerous other blood components under consideration for screening including the proform of eosinophil major basic protein (proMB) (Rode, Wojdemann et al. 2003) and pregnancy-associated plasma protein-A (PAPP-A) (Cicero, Bindra et al. 2003). However, the research shows that even more accuracy can be achieved if one utilizes a nuchal translucency measurement with some kind of serum screening in what is referred to as an integrated test.

INTEGRATED TEST (SCREENING)

The integrated test is defined variably by different research groups, but concerns a particular combination of serum markers and ultrasound findings. The definition of the integrated test followed by Wald and his colleagues, prominent researchers in the field of antenatal screening, includes: nuchal translucency (NT) measurements by ultrasound and blood serum testing for PAPP-A levels conducted at 10 completed weeks with AFP, unconjugated oestrial, free beta-hCG and inhibin-A examined at 14-20 completed weeks (Wald, Rodeck et al. 2003). Wald's integrated test is conducted over the first and into the second trimester. If a woman receives a "screen positive" result from the integrated test, it means she has a 1/270 or greater risk of a Down syndrome baby (Lenetix Medical Screening Laboratory 2002a). But while 1/50 women receive this result, only 1/20 women will actually have a pregnancy with Down syndrome (Lenetix Medical Screening Laboratory 2002a).

Nuchal fold translucency measurement

The main factor that has been added to make the test "integrated" is the nuchal translucency measurement read through ultrasound. Nuchal translucency (NT) thickness is used as a measurement of risk of fetal aneuploidy, and is measured most appropriately

at 10-14 gestational weeks (Chitty 1999). NT focuses on the area at the back of the fetal neck, the black area lying between the subcutaneous fascia and the skin, the size of which increases with gestational age. The measurement is considered abnormal if it is "greater than the 95th centile"(Comas, Torrents et al. 2003; Zoppi, Ibba et al. 2003). While NT is useful as a Down syndrome risk indicator, it is also associated with cardiac abnormalities, abnormalities of the extracellular matrix of the nuchal skin of the fetus, and abnormal lymphatic development (Haak and Vugt 2003). One study found that using NT alone as an indicator for Down syndrome could achieve prenatal detection rates for Down syndrome greater than 95% with a 5% false positive rate if conducted between 10 and 14 weeks gestation (Comas, Torrents et al. 2003). Wald's study (Wald, Rodeck et al. 2003) found a 1.2% false positive rate with an 85% detection rate for their integrated test. Overall, Wald argued that the triple screen and the NT test alone were not as cost effective as the integrated test because of their higher false positive rates (9.3% and 20% respectively). Cusick and colleagues (Cusick, Buchanan et al. 2003) found that first-trimester screening like Wald's integrated test was 29.1% less expensive than second-trimeseter screening, factoring in costs of fetal Down syndrome, live-born Down syndrome and screening plus live-born costs.

Another ultrasound finding being used in conjunction with serum screening as an indicator for Down syndrome is nasal bone absence at the ultrasound screen performed at 10-14 weeks gestation. This is thought to be the result of a developmental delay in Down syndrome fetuses. One study examining Down syndrome fetuses in utero and then after elective termination, found the nasal bone absent in about one-half the 21 fetuses examined (Larose, Massoc et al. 2003). Using nasal bone absence in conjunction with

NT and serum markers, researchers have managed to improve the test's capabilities for detecting Down syndrome. A study using maternal age, NT, free beta-hCG and PAPP-A markers at 11-14 weeks gestation achieved a detection rate of 90.5% and a false positive rate of 0.5% (Cicero, Bindra et al. 2003). Through computer modeling, another group predicted a detection rate of 95% with a false-positive rate of 2.9% combining nasal bone absence, maternal age, NT, free beta-hCG, and PAPP-A (Orlandi, Bilardo et al. 2003).

The serum screening tests and the integrated test are still only used as indicators of risk. They are continuously being improved upon, as this review suggests, building on findings from previous studies to reduce error and increase both the specificity and sensitivity of the tests. When one of these screening tests indicates an abnormality, women are encouraged to have a diagnostic test, usually amnio or CVS, to clarify and verify the findings. Thus, amnio and CVS and the screening tests discussed above work in conjunction with each other as tools for women to find out about the chromosomes and sometimes the genes of their fetuses.

This testing has become increasingly popular with pregnant women. In a Swedish study examining births to women 35-49 from 1991-1996, while 67% of the women did not have PGT, 30% of women had amnio and 3% had CVS (Cederholm, Haglund et al. 2003). A Japanese study found that of women having PGT, 99.6% of them had amnio and only 0.4% had CVS (Matsuda and Suzumori 2000). Alembik and colleagues examined trends in the diagnosis of congenital anomalies and found that between 1979 and 1999 the number of anomalies diagnosed prenatally jumped from 11.7% to 31.9% (Alembik, Roth et al. 2002), indicating a jump in PGT. The serum screening tests and the integrated test do not satisfy the need for a noninvasive genetic test because they only

screen. These tests are not diagnostic. The search for a non-invasive diagnostic test continues with fetal cell sorting.

FETAL CELL SORTING (DIAGNOSTIC)

Fetal cell sorting is available most commonly in clinical trials and research protocols. It is not available to most pregnant women, and it is never provided without amnio or CVS to confirm the findings. Scientists have been manipulating fetal cells found in maternal blood since the late 1970's (Campagnoli, Multhaupt et al. 1997). The idea is to identify genetic abnormalities in the fetus through isolating fetal cells from maternal blood and performing analysis on these cells. Fetal cells are present in the maternal circulation as early as 5 weeks gestation (Thomas, Williamson et al. 1994), and virtually all women have at least a small number of fetal cells in their bloodstream(Goldberg 1997). Problematic issues include the "sparse concentration" of fetal cells available from maternal blood and the separation of the fetal cells from maternal cells (Campagnoli, Multhaupt et al. 1997). Other problems presented are isolation of the fetal cells, enrichment of those cells, and discrepancies in the diagnosis of the fetus (Harper 1995). Rodriguez de Alba and colleagues (Rodriguez de Alba, Palomino et al. 2001) found through comparing fetal cell concentrations in maternal blood in first and second trimesters that the optimal week for a "reliable" non-invasive prenatal diagnosis is week 15.

Isolated fetal cells have been used to determine the presence of autosomal recessive diseases. Camaschella and colleagues (Camaschella, Alfarno et al. 1990) used isolated fetal cells to diagnose beta-thalassemia. One study found that there was a sixfold increase in fetal cells in the maternal circulation if the fetus is aneuploid (Bianchi,

Williams et al. 1997). A study in late 2002 cited at "50-75% detection rate" for Down syndrome and other fetal aneuploidy through fetal cell diagnosis from maternal circulation (Bischoff, Sinacori et al. 2002). While research has been unsuccessful in providing a definitive approach for this procedure thus far, some hold that fetal cell sorting "represent the future of prenatal screening and diagnosis" (Shulman 2003). There is an ongoing multi-site trial sponsored by the National Institutes of Health which proposes to perfect the technique for widespread application (Simpson and Elias 1998).

NEWBORN SCREENING

Newborn screening was the first genetic screening conducted on a massive scale in the United States, and with over 4 million babies screened annually, it is the most common type of genetic testing today (Andrews, Fullarton et al. 1994). It was established in the 1960's through the urging of parents of children with mental retardation to prevent the consequences of untreated phenylketonuria (PKU) (McCabe 2002). In 1961, a test that could detect severe, treatable inborn errors of metabolism was developed and this type of screening was initiated (Caskey 1992). The newborn screening system in place in the US today includes screening, follow-up, diagnosis and management as well as education and funding (McCabe 2002). Newborn screening for genetic disease responsibility lies primarily with states, as state legislatures and boards of health determine what is screened for in a particular state (McCabe 2002). All states screen for PKU and congenital hypothyroidism. Beyond these, some screen for as few as three disorders, while others screen for twenty-seven. While there is no question that screening for genetic diseases such as sickle cell and cystic fibrosis provides immeasurable benefit for the children who suffer from the disease, the debate is whether

screening for autosomal recessive traits actually accomplishes much, as there is little benefit for the newborns or their families to know carrier status.

CARRIER SCREENING AND TESTING

Carrier status screening is one facet of screening which can take place in health care organizations and other community group gatherings. It consists of mandating or making available a genetic test, the results of which should indicate whether or not a person has a specific disease gene. This type of testing has three broad categories: 1) autosomal recessive in inheritance, meaning that individual will never manifest the disease itself; or 2) presymptomatic, indicating an increased risk for a disease, but not a guarantee of manifesting it; or 3) a dominantly inherited condition, where if the person has the gene s/he will inevitably develop the disease. There are various optional carrier screening programs that have been available for cystic fibrosis (Marteau, Duijn et al. 1992; Balfour-Lynn, Madge et al. 1995), breast cancer(Lerman, Narold et al. 1996), beta-thalessemia and Tay-Sachs (Mitchell, Capua et al. 1996), Huntington's Disease (Harper 1992), and many other diseases for which particular genes have been identified.

Testing/screening is not universally advocated. Holtzman (Holtzman 1996) outlines several reasons why genetic tests/genetic screening are of such great concern:

- Such tests can predict risks of disease in healthy people without, in some cases, certainty of disease manifestation.
- There is no independent means to verify this prediction.
- For some disorders, there are no interventions to reduce risk.
- Genetic test results implicate relatives by the nature of the information.
- There is, for some people who are tested, psychological distress with genetic knowledge.
- There are ethnic differences in disease frequencies that could stigmatize certain groups further.
- There are not enough medical genetics professionals to provide the necessary resources to fill the needs of those who are receiving genetic tests.

When carrier testing is conducted, there are many different reactions to the results among those tested. Individuals who are tested in this manner have been judged purely by the *presence* of genes, in the *absence* of phenotypical expression. Yet those found to carry genes for late onset diseases may be treated as though they are ill and have the disease, even when such genes do not guarantee disease manifestation, as in the instance of BRCA1 and BRCA2 (Lerman, Narold et al. 1996). Those carrying genetic mutations may be perceived as "diseased" or the "presymptomatic ill" (Lippman 1992; Marteau, Duijn et al. 1992; Nelkin 1992; Hubbard 1995). All this may occur despite the fact that nothing has been documented up to this point which links being a carrier of a recessive gene to any fully predictable negative health consequences. Geneticists estimate that each individual carries four to eight recessive deleterious genes (Kenen and Schmidt 1978), thus making nearly everyone a "potential carrier" of something. The Council on Ethical and Judicial Affairs of the American Medical Association suggests that (Council on Ethical and Judicial Affairs 1998:19) :

> even seemingly benign tests that provide information about genetic status but that have no bearing on phenotypic expression or reproductive decisions may have a psychological impact on the patient who interprets the presence of a mutation as a deep-seated flaw.

For such reasons, some individuals at risk of serious disease choose not to be tested and also not to bear children who would at least be carriers (e.g. Wexler 1995).

In 1998 ACOG suggested that Ashkenazi Jewish couples should be offered Canavan disease carrier screening, ideally prior to pregnancy (MSSM 2003). In October 2001 the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Geneticists (ACMG) published clinical guidelines for implementing cystic fibrosis (CF) screening (ACOG and ACMG 2001). The practicing

ob/gyn is now responsible for offering screening to the following patients before conception or during prenatal care: those with a history of CF, reproductive partners of individuals with CF, and couples with one or both partners of European Caucasian or Ashkenazi Jewish descent.

There is a panel of screening tests now available in large metropolitan hospitals for "Jewish genetic diseases". These are disorders that occur more commonly in the Jewish population than in the population at large. This panel screens for nine diseases as of 2003 (MSSM 2001; Lenetix Medical Screening Laboratory 2002; Therapeutics 2002; MSSM 2003) listed in table AB.

Table AB. Jewish Genetic Diseases

DISEASE TYPE—description	CARRIER RISK	Inheritance Pattern
Tay-Sachs disease: progressive neurological problems leading to death in early childhood(by age 5), deterioration in mental and physical characteristics, range of disabilities affects quality of life, no treatment available	1/30 or 1/25	Autosomal recessive
Familial Dysautonomia: neurological disease that includes insensitivity to pain, unstable blood pressure, frequent pneumonia and poor growth, feeding difficulties, normal intelligence, frequent hospitalization, shortened life span (death in 50% of patients by age 30), surgical and other treatments available	1/30	Autosomal recessive
Canavan disease: degenerative neurological disorder (brain and nervous system degenerate), fatal in childhood, no treatment, death between 1 year and adolescence	1/40	Autosomal recessive
Niemann-Pick disease: progressive mental and physical deterioration, large liver, poor growth, no treatment, death by age 4, birth incidence is 1/32,000	1/90 or 1/80	Autosomal recessive
Gaucher disease: most common genetic disorder affecting the Ashkenazi Jewish population (1/450 has the disorder), fatigue, anemia, bone pain, retarded growth in children, enlarged liver and /or spleen, and easy bruising or bleeding, treatable with enzyme replacement therapy	1/10 or 1/15	Autosomal recessive
CONTINUED ON NEXT PAGE		

DISEASE TYPE—description	CARRIER RISK	Inheritance Pattern
Fanconi Anemia: severe anemia, learning disabilities or mental retardation, short height, often abnormalities of the heart, kidneys or limbs, high rates of cancer, especially leukemia	1/89 or 1/100	Autosomal recessive
Bloom Syndrome: growth retardation, poor immune system, normal intelligence, affected individuals usually die of cancer before age 30	1/100 or 1/110	Autosomal recessive
Mucolipidosis IV: affects brain and nervous system, severe psychomotor retardation and impaired vision, no known treatment, some milder forms of disorder	1/120	Autosomal recessive
Torsion Dystonia: affects movement control, appears between ages 6-16, progresses rapidly from impairment to complete physical disability—limbs rigid and contracted, 1/3000 carriers manifest symptoms, normal life expectancy, normal intelligence	1/900	Autosomal dominant

PREIMPLANTATION GENETIC DIAGNOSIS

Preimplantation genetic diagnosis (PGD) is a process that tests an embryo that was created outside a woman's body for genetic abnormalities before it is implanted in her uterus. It was first successful, meaning a woman who had embryos tested and implanted achieved a live birth, in 1990 (Egozcue, Santalo et al. 2000). The most common technique for PGD utilizes the 3-day embryo (6-10 cell stage), taking a biopsy of one of the cells, all of which are totipotent, so theoretically the test does not impact the ultimate development of the fetus (Cunningham, Gant et al. 2001). The cell is cultured and tested for genetic disorders. PGD was initially developed to identify embryos with sex-linked or autosomally inherited genetic abnormalities (Egozcue, Santalo et al. 2000). Preimplantation genetic diagnosis is presently used for detecting preexisting genetic conditions, preselecting embryos of a particular sex, preventing transfer of embryos with age-related aneuploidies, and identifying specific genetic disorders (Kuliev and Verlinsky 2003). An added benefit from PGD is the predicted increase in successful pregnancies

for women with age-related aneuploidies and recurrent spontaneous abortions, although there is controversy as to whether this is actually possible in practice (Egozcue, Santalo et al. 2000; IWGPG 2000; Geraedts, Harper et al. 2001; Kovacs 2003).

The advantage of PGD is that it avoids both pregnancy and abortion after the diagnosis of a genetic abnormality; the embryo with the genetic disorder is simply discarded. PGD is up to 100% accurate (IWGPG 2000). PGD is also considerably more expensive than traditional prenatal genetic testing. The number of clinics offering preimplantation genetic diagnosis is expanding worldwide, but each clinic has its own list of chromosomes it analyzes and anomalies it can detect. In a worldwide survey of 20 PGD centers, all but one center offered sex determination, 13 of 20 screened for aneuploidy, all but one center offered detection of translocations and other structural chromosomal abnormalities, and the number of centers offering monogenic disease screening varied considerably (Geraedts, Harper et al. 2001). PGD has been used to test for some controversial disorders:

Table AC. Controversial Usage of Preimplantation Genetic Diagnosis

DISORDER	SOURCE	CONTROVERSEY
[illegible] syndrome	[illegible]	[illegible]
Early Alzheimer's	[illegible]	[illegible]
HLA matching	[illegible]	[illegible]

DISORDER	SOURCE	CONTROVERSEY
		syndrome, with suggested use in Huntington Disease.

PGD requires that the woman be artificially impregnated using invitro fertilization (IVF), intracytoplasmic sperm injection (ICSI), or related procedures (assisted reproductive technologies-ART). ART is a fast growing industry in the United States. Nationwide data comparing 1996 and 1998, show that the number of clinics offering ART services increased 18%, with the number of reported procedures jumping 26.5% (MMWR 2002). Thus, one can assume the demand for PGD will grow as the demand for ART increases. In 2000, the overall number of PGD cycles conducted worldwide may be more than 2,500, with approximately 600 clinical pregnancies resulting in close to 500 children born (IWGPG 2000). There is also a new test for embryos before implantation called comparative genomic hybridization (CGH), which analyzes the all of the chromosomes present in the polar body before conducting the standard FISH test on a blastomere from each embryo (Wells, Escudero et al. 2002). This technique is presented as more thorough than the standard preimplantation testing currently available.

APPENDIX B HOW IT ALL BEGAN

A VERY BRIEF HISTORY OF GENETICS AND EUGENICS

Gregor Mendel is commonly credited with establishing the fundamentals of modern "Mendelian" genetics through his experiments with pea plants, first published in 1865. Such patterns of inheritance can be determined by looking at large numbers of offspring in generations (Kevles and Hood 1992). Today this is revelatory in its simplicity and comprehensive understanding of heredity, but Mendel's work went largely unnoticed until 1900, when several botanists independently rediscovered it.

Meanwhile, in 1877, cell biologists discovered chromosomes. By 1892, August Weismann, a German physiologist, was confident enough in his research to conclude: male and female parents contribute equally to the heredity of their offspring; sexual reproduction therefore generated new combinations of hereditary factors; and chromosomes were the bearers of heredity (Kevles and Hood 1992). Drawing upon this work and within two years of the rediscovery of Mendelianism, Archibald Garrod discovered an inborn error in metabolism---alkaptonuira---and became the first person to demonstrate Mendelian inheritance in man (Kevles and Hood 1992).

By 1910, Mendel's model of units of heredity, now chromosomes, had been extensively researched, and found to contain genes. Genes were necessary in the explanation of heredity because the number of chromosomes was statistically too few to account for the vast number of traits that need to be represented and passed down. No gene had yet been associated with a particular chromosome. In that year, Thomas Hunt Morgan discovered sex-linkage through his research on fruit flies. He proved that in fruit flies, the gene for red eye or white eye traveled on the X chromosome, the sex

determinant chromosome. The first instance of a sex-linked trait in humans was red-green colorblindness (Kevles and Hood 1992). Gene mapping had begun.

From that point, geneticists realized that genetic inheritance was to a great extent predictable following Mendelian lines. What makes genetics *unpredictable* is mutations--losing, gaining, substituting or rearranging genes or pairs of genes on the same chromosome or between two chromosomes. Mutations occur rarely and at random. Mutations are likely to be recessive to the traits the organism normally exhibits. Recessive means that in order for the trait to be expressed, both genes for the trait must be present.

Ironically, these mutations were what allowed genetic research to proliferate, because the examination of mutations was what enabled scientists to determine inheritance of the trait in question (Kevles and Hood 1992). Mutations are called polymorphisms when they exist in more than 2% of the population. Polymorphisms are common variations in the sequence of DNA among individuals (Witherly, Perry et al. 2001). To define a variation (a reoccurring mutation) as a polymorphism, researchers consider the biology behind the gene.

Francis Galton, coined the term and became the "father" of "eugenics" in 1883 (Allen 1994). Galton defined his term as "'the science of improvement of the human germ plasma through better breeding'; also as 'the study of agencies under social control which may improve or impair the racial qualities of future generations'" (Paul 1992:666). Galton intended eugenics to be a science and helped organize professionals to further it through the eugenics movement. Galton hoped that "what Nature does blindly, slowly and ruthlessly, man may do providently, quickly, and kindly" (Larson 1995:19). Overall,

eugenicists advised two different kinds of interference in human reproduction: "positive eugenics" to manipulate human reproduction to produce superior people, and "negative eugenics" to improve the quality of the human race by eliminating genetically inferior people from the population (Kevles and Hood 1992).

In early eugenic circles, very little attention was paid to economic, environmental or socio-cultural influences; nor was there attention to polygenic complexities (inheritance dependent upon many genes rather than one) (Kevles and Hood 1992). Eugenic "science" was rife with class and race prejudice. The founders and supporters of eugenics were Northern European and American, predominantly white, upper-middle class Protestant males with standards of fitness and social value representative of their culture.

The eugenics movement in the United States was led by biologist Charles Davenport (1866-1944), who defined it: "the science of the improvement of the human race by better breeding" (Allen 1994:168). He and Harry Hamilton Laughlin (1880-1943) conducted research at the Eugenics Records Office (ERO) at Cold Spring Harbor, Long Island, including the "collection of thousands of family pedigrees, cross-referenced by family name, hereditary conditions and geographic locality" (Allen 1994:168). Leon Whitney, a former dog breeder turned unsophisticated eugenicist and secretary of the American Eugenics Society, advocated "fitter family" contests at county fairs in the Midwest (Allen 1994). The most eugenically positive families were determined by judges who reviewed human pedigrees similar to the ways cattle, chickens and pigs were awarded blue ribbons. What made these families fit is difficult to discern, but some evidence is suggested by the fact that all entrants had to take an IQ test and the

Wasserman test for syphilis (Kevles 1994). Other notable eugenic research was carried out at Princeton, Harvard, the University of California at Berkeley and the American Museum of Natural History (Allen 1994).

Some of the successful negative eugenics programs were eugenic sterilization laws, which by the late 1920's were active in 24 states. After the Supreme court upheld involuntary sterilization laws in *Buck vs. Bell* in 1927, sterilization laws were passed or active in thirty states, and the number of sterilizations performed each year "rose substantially" (Reilly 1991). By 1933 California had subjected more people to eugenic sterilization than all other states combined (Kevles 1994).

While eugenicists were the first geneticists, they did not keep up with genetic science, and in the U.S. by the end of World War I, they were being challenged in the lab. However, they remained active politically, and in 1924 helped pass the Johnson Act. This act drastically limited immigration to the U.S. from southern and central Europe, the Balkans and the Soviet Union (Allen 1994). The rationale behind such legislation was the assertion that there was a higher incidence of genetic abnormalities among the poor, immigrants and ethnic minorities, including Jews, blacks and Native Americans. Many have argued that millions of Germans died in the Holocaust because there were such restrictions.

Some of the most notorious eugenic acts are the Nazi sterilization efforts in World War II that culminated with the Holocaust. In 1933, the fascist tendencies of eugenics became crystal clear when the Nazi Sterilization laws were passed. They were based on---and nearly a direct translation of---the California sterilization laws. Under these laws several hundred thousand people were sterilized in Germany in the 30's and early 40's

(Kevles 1994). Sterilization was ultimately rejected as the way to rid Germany of the Jewish population, in part because the mechanism to gas people was already established (Proctor 1995).[1]

But it was the Nazi's "Final Solution" of murdering millions of people in the name of eugenics that caused the essential demise of the eugenics movement *under that name* throughout the world. There had been eugenics societies in Germany, France, Brazil, Russia, the U.S., Great Britain and elsewhere (Adams 1990).

Diane Paul provides a history of the early eugenicists' pursuit of a major goal---ridding society of "feeblemindedness" by preventing those with the trait from breeding. Gradually geneticists realized that even if all were prevented from breeding, the heterozygotic (see Glossary in Appendix H for definitions) carriers of the gene(s) would allow feeblemindedness to survive for an extensive period of time. By one estimate 8,000 years would only decrease the feebleminded to 1/100,000 from the original frequency of 3/1000 (Paul 1998)! There were attempts made to identify and sterilize those who were heterozygotic for "feeblemindedness", assuming that they would exhibit less intelligence than "normal". H.S. Jennings, a eugenicist, said in 1927, on mutations (Paul 1998:142):

> a defective gene--such a thing as produces diabetes, cretinism, feeble-mindedness--is a frightful thing; it is the embodiment, the material realization of a demon of evil; a living self-perpetuating creature, invisible, impalpable, that blasts the human being in bud or leaf. Such a thing must be stopped wherever it is recognized.

[1] The euthanasia operation for the destruction of the "mentally ill" began in 1939 and by 1941 had successfully killed more than 70,000 patients from mental hospitals in Germany (Proctor 1995).

Paul concludes that the early eugenicists were not statistically challenged. Instead, they believed that despite the long time required to wipe clean the slate of human undesirables, if any could be prevented it was worth the task.

While the eugenics movement in its own name was ebbing after World War II, eugenicists were moving into other social movements they believed would further their negative eugenic goals---the population control movement (Allen 1991). Here they hoped that by promoting birth control for all that fewer undesirable people would be born. Eugenicists and others in this movement promoted means of contraception that could be imposed on women (e.g. sterilization, the Depo Provera shot, the I.U.D., the Norplant implant) rather than contraceptives women could control—start and stop using at will (e.g. the Pill, the diaphragm, cervical caps) (Clarke 2000).

Another direction in which eugenics moved after World War II was toward sociobiology. When viewed historically, eugenics is repugnantly race, class and ability biased, and raises the specter of Nazi terror. Most would like to believe eugenics has disappeared. But many scientists in the fledgling field of medical genetics in the not so distant past, and some presently, believe that the field should serve to improve the race. In 1960, the journal *Human Heredity* published an article that stated that people carrying the gene for "infantile anaurotic family idiocy" should never bear children, while in 1968 Linus Pauling advocated that carriers of sickle-cell be tattooed on their foreheads (Paul 1997). In the presidential address of 1970 to the American Association for the Advancement of Science, Bentley Glass speculated, "No parents...have a right to burden society with a malformed or mentally incompetent child" (Paul 1997:106).

Even in present day medical genetics there are eugenic undertones. For example, in 1984 geneticist Marjorie Shaw asserted "the law must control the spread of genes causing severe deleterious effects, just as disabling pathogenic bacteria and viruses are controlled" (Shaw 1984:113). Quite active today, bioethicist Gregory Stock, director of the Program on Medicine, Technology and Society at the University of California, Los Angeles and author of the 2002 book *Redesigning Humans: Our Inevitable Genetic Future* is in everything but title a eugenicist. He said in a 2003 seminar that "Genes are the biggest window into who we are, and we are drawing back the curtain. This will call into question what it means to be a human being" (Flam 2003). According to Dr. Stock, sex may become purely recreational, and reproduction will be best achieved in a laboratory. He argues that it could be conceived as "reckless and primitive" to produce a child without genetic testing. He believes that the prenatal genetic testing of today will ultimately lead to germline alterations in the future, with the positive consequences of choice and control:

> These developments will write a new page in the history of life, allowing us to seize control of our evolutionary future. Our coming ability to choose our children's genes will have immense social impact...Biological enhancement will lead us into unexpected realms, eventually challenging our basic ideas about what it means to be human (Stock 2002:iv).

Troy Duster (Duster 1990) has called this "the back door to eugenics." Here the promotion of eugenic goals shifts from populations to individuals, though there may still be discriminatory genetic screening programs directed at particular social groups.

A third, more professional and scientific direction that some eugenicists took was the development of medical genetics. Here the shift of focus was away from applying eugenics to populations and instead focusing on individuals. My work is focused in the

individual scenario. Generally, individuals who utilize prenatal genetic testing do not *intend* to produce population effects. Their goal is to exert greater control of the kind of family they produce. In many cases, the collective result of their individual actions would be unpalatable. But a definition of eugenic consequences broad enough to incorporate unintended consequences will necessarily involve medical genetics (Paul 1992). Today, prenatal genetic testing allows for some individual options regarding what kind of offspring is desired, or more commonly is *not* desired. In the future families may well be able to design the kind of child they want through genetic manipulation of the embryo and IVF. Duster calls this the "backdoor" approach to eugenics, through screening, treatments and therapies (Duster 1990).

The backdoor has been opened through the promise of medical genetic technologies, enhanced by the completion of the Human Genome Project in April 2003. The completion of the project to map the location of genes on the 23 pairs of human chromosomes coincided with the fiftieth anniversary of the Watson and Crick discovery of the double helix of DNA. American researchers joined those from other countries to map the 2.85 billion base-pairs of DNA comprised of the 23 pairs of chromosomes that each carry 50,000,000 to 300,000,000 base pairs and hundreds to thousands of genes (Ault 2003). The understanding of medical genetics has grown considerably through research furthering the completion of the HGP, and promises of disease gene discoveries are plentiful. Over 1000 disease genes have been located, but without phenotype-genotype correlation, their medical usefulness remains very limited (Ostrer 2002). Behavioral and environmental considerations must ultimately be factored into these equations. One example of this is the complex linkage medical genetics research

reported in the presidential address of the American Society for Human Genetics in 2002 that found alcoholism is approximately 40% environmental and 60% genetic (Conneally 2002). Considerable research must be conducted to clarify findings such as these and understand them in terms of the biomedicalization and geneticization of American culture. Policies for medical interventions will need to be crafted for insurance purposes at the very least.

The main promise of medical genetics lies in the translation of the location of a disease gene to a treatment of the disorder caused by the genetic defect. This has not proven to be a linear progression. There are today no known cures for diagnosed genetic diseases, and very few possibilities for treatments. There are exceptions, of course, such as newborn screening programs that identify children with diseases that can be treated through diet, pharmacology and physical therapy (for more on this see the newborn screening section in Appendix A). At the annual meeting of the American Society for Human Genetics Albert de la Chappelle queried his colleagues about gene therapy (de la Chappelle 2002): "What have we accomplished in the way of helping our patients? Very little indeed." He went on to say the only effective innovation has been the pharmaceutical Gleevak being developed for chronic myelogenous leukemia (CML) through research that spanned 1960-1998. Optimism is high among genetics researchers, but in practice, the applications are few, and some argue that hype is endemic to recruit funding and to enhance cultural legitimacy.

Medical genetics is a burgeoning field, attached most precipitously to obstetrics through the genetic counseling process for prenatal genetic testing offered to pregnant women over 35 and to any pregnant woman considered high risk for genetic problems.

Any pregnant woman who can pay for prenatal genetic testing can have it. The social and biomedical constructions of pregnancy as an experience that requires genetic monitoring is changing the experience of pregnancy for pregnant women and their families. This is what my research examines.

APPENDIX C ABORTION CONSIDERATIONS FOR WOMEN WHO UTILIZE PGT

EDUCATION FOR PREGNANT WOMEN ABOUT "ABORTION" AND PGT

Abortion is generally not explicitly discussed with pregnant woman who present for PGT. During the genetic counseling session, most women are informed of the risks of the procedures and, less commonly, the dearth of treatments for diagnosed disorders. Some women are provided written material before the session to prepare questions, or after the session so they can review what was discussed. The actual term "abortion" is rarely used, even in the event of a diagnosis of a birth defect. This is illustrated by a pamphlet distributed in California to most pregnant women:

> Information will be given to the woman...by a doctor or genetic counselor. They will discuss the type of birth defect that has been found and any available treatments. They will also discuss options for continuing or ending the pregnancy. The woman can then make a decision (CGDB 1995:9).

The euphemism "ending the pregnancy" makes it sound as though one thinks, "I am no longer pregnant," and magically it is true. In a booklet distributed to pregnant women who are enrolled in Oxford Health Plans, the only reference to what could occur in the event of an abnormal finding is found under "What is amniocentesis, and why is it done?"(Plans 2000:12): "Before the procedure you should have an opportunity to discuss the test with your provider. Genetic counseling before testing ensures that you have all the information you need to make sound decisions." One reads between the lines: but we are not going to provide you with any information now. And sound decisions about what? In a patient education pamphlet published by a genetic testing laboratory and research center on amniocentesis this is written about adverse results (GeneCare 2002):

"If the test shows your fetus has a disorder, the nature of the disorder will be discussed with you. Prenatal diagnosis allows couples to make informed decisions and helps with the management of their pregnancy. Supportive genetic counseling is available." The innuendo is that there may be options and decisions to make, but no clear indicator of what that could be is provided. Another patient education pamphlet produced by a genetics lab says, "The compelling reason for carrier testing is that prenatal diagnosis is available for carrier couples to determine whether or not their fetus has the disease" (Lenetix Medical Screening Laboratory 2002) and says nothing more about the decision-making process that follows. Only one patient oriented pamphlet that I found mentioned abortion, and it was produced by the American College of Obstetricians and Gynecologists:

> You may choose to continue the pregnancy and have the baby. Or, you may choose to have an abortion...If you choose to have an abortion, you should decide as soon as possible. The earlier an abortion is done, the safer it is for you (ACOG 1999).

THE MORAL AND LEGAL LANDSCAPES FOR ABORTION IN THE U.S.

The moral and legal landscapes of the United States vis-à-vis abortion are pertinent to pregnant women having the alternative option of abortion to shape their families when using PGT. Women's option to terminate a pregnancy after 18 weeks, whether because of a genetic diagnosis or by choice, could be in jeopardy. Here I offer only a very brief overview.[1] In terms of legality, the current Bush administration is vehemently pro-life, and has been consistently pursuing legislation that limits accessibility and availability of abortion services. The Bush administration seeks to

[1] For complete reviews of the history of abortion and challenges to it from sociological and historical perspectives please see Petchesky (1990), Luker (1984) and Reagan (1997).

redefine "child" as "an individual under the age of 19 including the period from conception to birth" (Hibbert 2002). This is ostensibly a change to include fetuses in the State Children's Health Insurance Program (SCHIP), but what the move truly accomplishes is to create a "disincentive for pregnant women carrying SCHIP-covered prenatal "children" by disregarding the carrier's health and, potentially, creating a conflict between the carrier, a non-patient, and the SCHIP-covered prenatal patient" (Hibbert 2002:10).

The Bush administration is also pursuing the notion of "fetal personhood," seeking to bestow *in- and ex- utero* embryos and fetuses citizenship, heretofore rejected by both the Supreme Court and the Constitution. In conjunction with the pressure for fetal personhood, the Bush administration successfully passed a ban on a specific kind of abortion procedure, the partial birth abortion or dilation and extraction (D & X) which is not performed until after 16 weeks gestation. Eighty-eight percent of abortions are performed within the first 12 weeks of pregnancy, with 55% performed before 8 weeks (MMWR 2000). Most of these are performed by vacuum aspiration. A dilation and evacuation (D & E) abortion is performed after 16 weeks, and is accomplished by dilating the cervix extensively, mechanical destruction of the fetus, evacuating *fetal parts*, and, using a vacuum curette to remove the placenta and remaining tissue (Cunningham, Gant et al. 2001). The D & X abortion is similar except that part of the fetus is extracted through the dilated cervix first, thus the title "partial birth abortion." The problem with the bill limiting use of the "partial birth abortion" is that there is no such thing: abortion opponents coined the term and Congress used it in the bill (Parker-Pope 2003). While most experts agree the term is meant to apply to a D & X procedure, critics warn the

language could be widely interpreted to apply to the D & E and other second trimester procedures (Parker-Pope 2003; Rovner 2003). Another encompassing concern is that there is no exception to allow the procedure in cases where the woman's life is in danger. The Bush Administration is also slowly working towards making all abortion illegal. Only about 1.6% of all abortions are of the D & X type (Parker-Pope 2003).

The availability of the full range of abortion services is important to women who have PGT because sometimes they do not receive a diagnosis from amnio until around 20 weeks gestation, and therefore must have a D & E or D & X abortion. A late diagnosis from amnio could be because the woman presented late for her amnio or because the cultures of the amniotic fluid grew slowly. A recent article in the *San Francisco Chronicle* recounted a woman's pursuit of a D & E from her HMO, one of the largest in California, that had no trained providers in the three closest facilities to the woman (Ryan 2004). She had to travel from Sacramento to San Jose to get the abortion. The only time a D & X is done is when there is no way to do a D & E. The D & E is considered a safer procedure for the woman than the D & X.

In a study of late abortion, defined as 18-34 weeks gestation, 11.6% of the procedures were conducted because of a diagnosed fetal anomaly or fetal genetic disorder (Hern 2001). There are only two clinics in the western hemisphere and Europe that provide abortion services after 24 weeks, despite the fact that some women do not receive results until after this gestational age. These clinics do not advertise and they are difficult to track down without help. One woman recounted her story in a parenting magazine (Bell 2003:55): "We were lucky to have access to the information about Wichita, which one of my beloved midwives and several of the specialists we saw declined to give us,

even when we asked directly." Abortions conducted after 24 weeks are surely in grave danger in this political climate. A study of 124 late term abortions (after 14 weeks) because of fetal anomaly, diagnosed genetic disorder or fetal death were most commonly conducted for neural tube or central nervous system defects (27%) and trisomy 21(Down syndrome---24%) (Hern 2001). Another study examining 130 pregnancies diagnosed with perinatal lethal conditions before 24 weeks gestation found that 92% of those with central nervous system anomalies aborted, as did 70% of those with chromosomal anomalies (Hassed, Milleret et al. 1993). This decision by the Bush administration to eliminate the "partial birth abortion" will no doubt impact the way PGT is practiced in the future.

The moral climate vis-à-vis abortion in the U.S. has also been affected by the anti-abortion activism of the past twenty years or so. From bombing clinics to shooting abortion providers, anti-choice extremists make their views known: abortion is killing a baby and it must be stopped. Abortion providers are in jeopardy not only from organizations like Operation Rescue, but also because of the lack of training available to new physicians in abortion techniques and the reluctance of the medical profession to incorporate abortion as a routine aspect of women's healthcare.[2] The question of whether abortion is murder is the center of the pro-choice or pro-life dichotomy. A recent sociological study of emergency contraception argues that this perceived dichotomy between contraception and abortion is more of a continuum from potential fertility to pregnancy (Simonds and Ellertson 2004). The continuum framework could be carried further to contrast the bifurcation of choice/life.

[2] For an excellent study of abortion providers both before and after legalization, see Joffe (1995). See Dresser (1994) for a bioethical examination of providers' conscientious objections to perform abortions.

Bioethicists find much to discuss about abortion. When life begins and whether a conceptus is "alive" at conception are popular debate topics. Bioethics frames the struggles around abortion as conflicts primarily about privacy, autonomy and individualism.[3] Recently there were a series of articles and commentaries on another bioethics morality slant on abortion, the "futures like ours" arguments, presenting the fetus as an individual capable of having a future and abortion as deprivation of that future.[4] These bioethics arguments for the most part all return to the idea that abortion is either killing or it is not.

The contentious discourses swirling around the abortion issue are intense in terms of issues of morality. A recent proclamation by a British bioethicist (Vehmas 2002:84) concluded that because procreation assumes parental responsibilities, and those responsibilities include an implied duty of caring for any kind of child, with very few exceptions, there is no "morally sufficient reason to terminate [a] pregnancy on the grounds of fetal abnormality." Another recent bioethical examination of preimplantation genetic diagnosis concluded that PGD was a better option than abortion of genetically abnormal fetuses because while abortion could be considered "killing," with PGD the spare embryos are "allowed to die" (Cameron and Williamson 2003:92).

Situating *all* women considering abortion for whatever reasons, such discourses must somehow be negotiated by women using PGT. This is one element of such women being positioned as "moral pioneers" (Rapp 1999) by PGT.

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[3] For examples of explanations of bioethics arguments concerning abortion, please see (Callahan 1970; Annas 1993; Callahan 2003).

[4] For a complete discussion, please review (Brown 2000; Marquis 2001; Brown 2002; Parsons 2002).

APPENDIX D GROWING NEED FOR GENETIC CARE PROVIDERS

The professions of "geneticist" and "genetic counselor" are relatively new as medical specialties. The American Board of Medical Specialties (ABMS) recognizes a number of medical genetics specialties, one subspecialty and masters' level genetic counselors. Only since 1982 has there been board certification for medical genetics specialties, and the first genetic counselors were certified the same year. Medical geneticists and other M.D. and Ph.D. genetics specialties are certified by the American Board of Medical Genetics (ABMG). Genetic counselors were certified by the ABMG until 1990, when the American Board of Genetic Counseling (ABGC) took over the certification process. Certification for ABMG and ABGC are only available every three years.

The total number of genetic counselors as of September 2003 is somewhere around 1675 (ABGC 2003). [Please see Table DA.] This number is from the counselors certified by the ABGC in 1993, 1996, 1999, 2002/3 combined with charter members certified by ABMG and allowed to acquire ABGC certification in 1993 (ABGC 2003). Considering the demand for genetic counseling for not only prenatal testing, but also for carrier, mutation testing, and presymptomatic genetic disease testing, these numbers seem very low.

TABLE DA. Certification of Genetic Counselors

American Board of Genetic Counseling Statistics for ALL Years									
Genetic Counseling	1982	1984	1987	1990	1993	1996	1999	2002/ 2003	Next exam 2005
Genetic Counselors certified by The American Board of Medical Genetics (ABMG)	167	144	179	141					
Genetic Counselors certified by The American Board of Genetic Counseling (ABGC)					181	258	340	401	
Charter Members					495				
Charter members are ABMG certified diplomats who became members of the ABGC in 1993									

(ABGC 2003)

The total number of practicing clinical medical geneticists is a bit more complicated to determine. [Please see Table DB.] There are 1773 total diplomats of the ABMG, but this includes those who are trained to run laboratories and conduct research as well as those who practice some kind of genetic medicine. Another complicating factor is that 989 of those certified are also certified by one of the 21 other ABMS member boards, 155 are certified by 2 other ABMS member boards, and 12 ABMG diplomats are certified by 3 other ABMS member boards (ABMG 2003). Logically, the top three other specialties ABMG diplomats are certified in are: 94 in internal medicine, 126 in obstetrics and gynecology and 668 in pediatrics (ABMG 2003). Multiple certifications make it difficult to determine if the AMBG certification is the one being exercised, such certification may be an aid only to practice and not utilized daily as the primary practice of the individual.

TABLE DB. Certification for Medical Geneticists

American Board of Medical Genetics NUMBERS OF CERTIFIED SPECIALISTS IN GENETICS									
Medical Genetics Specialties	1982	1984	1987	1990	1993	1996	1999	2002	Total
Total number of certificates	519	263	268	254	384	302	201	161	2352
Total number of individuals	392	226	241	210	331	251	174	146	1773
Subspecialty certificates							2001	2003	Total

(ABMG 2003)

Over the past three decades, genetic services delivery has been relatively successful, with the exception of access. Genetic services have not been available equitably because of geographic, sociocultural, socioeconomic, and linguistic barriers (Guttmacher, Jenkins et al. 2001). Genetic care is expensive, requires comprehension of complex concepts, and is most widely available in large metropolitan areas.

With the elaboration of genomic medicine, targeting those who have predispositions to cancer or other detectable disorders, and addressing multifactorial genetics, the roles of the geneticist and genetic counselor will be changing dramatically in the coming years. The roles of genetic specialists will be redefined in response to the development of genetics-based tests and therapies, as well as the expansion of nongenetic specialist providers' use of genetics. A group of genetics professionals with government and non-government affiliations recently suggested that in light of the changes in

genomic medicine, genetic professionals should help establish a set of criteria for different scenarios, including when a genetic test requires referral to a genetic specialist, what tests should be offered and to whom, what a traditional genetic counseling session should accomplish, and what types of genetic tests require informed consent (Guttmacher, Jenkins et al. 2001:219). What is most essential to implementing these recommendations, however, is an understanding of genetics and its applications by referring physicians.

Studies of obstetricians' knowledge of genetics have not been encouraging. When asked if newborn screening was relevant to their practice, 41% of obstetricians surveyed stated they were not sure, and 31% knew very little about newborn screening, despite the design of newborn screening which requires that obstetricians educate pregnant women about the screening (Dolan, Gross et al. 2002a). Moreover, the biomedical industrial complex traditionally relies on obstetricians to refer pregnant women for genetic services. One study found that general practice obstetricians considered their knowledge of clinical genetics to be a 5 on a scale of 1 to 10 (Aalfs, Smets et al. 2003). The same study concluded that "limited alertness and awareness" among general practice obstetricians about genetic risk factors in their patients played a major role in less appropriate timing of a referral for reproductive genetic counseling (Aalfs, Smets et al. 2003).

Others have suggested that genetics is more a component of primary care (Scanlon and Fibison 1995) and that primary care will quickly be required to change in relation to use of genetic tests and technologies (Callahan, Durfy et al. 1995). One clinical geneticist interviewed by Ettorre (2000) envisioned the monitoring of genetic risk

as soon becoming a routine part of everyday medical practice. The implication is that primary care and other specialists will engage in counseling their patients, communicating genetic risks. This geneticist said: "the whole counseling concept should be changed to be more informative and more informing the general public and test users" (Ettorre 2002:67).

Medical curricula are currently expanding to address such needs. During 1992, 63% of U.S. medical schools required a human or medical genetics course, with the average course entailing 30 hours of study (Andrews, Fullarton et al.1994). There are also continuing medical education courses to update practice on genetics offered regularly, and practice guidelines are issued by professional associations to instruct and engage providers in genetics. The new National Coalition for Health Professional Education in Genetics (NCHPEG) (Bergeron 2002) is an interdisciplinary organization composed of over 100 diverse specialty organizations devoted to health professional education in genetics. The NCHPEG has enumerated the core competencies in genetics that it sees as essential for all health professionals (Guttmacher, Jenkins et al. 2001). These can be found on the web: http://www.nchpeg.org. Web-based education will likely be important in this domain as well.

APPENDIX E INTERVIEW GUIDES

Professional Interview Guide

1. Professional History

    a. What is your educational/ professional background? What areas of expertise and training did you focus on, what degrees do you hold, and which if any medical specialties are you board certified in?

    b. How and why did you become involved in prenatal genetic testing? What about this kind of testing interests you?

    c. What is your role at this position? Please describe your primary responsibilities.

    d. How do you feel about the kind of work you do?

    e. What effects does this work have on you?

    f. Are you in a relationship?

    g. Do you have children? Did you/your partner have prenatal testing with the pregnancies?

2. Current situation of prenatal genetic testing

    a. How would you define prenatal genetic testing? What is its primary purpose? What role does it play in pregnancy for a "normal, healthy" woman?

    b. What do you think are the ethical aspects of prenatal genetic testing?

    c. How do you think insurance companies organize your work around prenatal genetic testing?

d. What do you think are the implications for society of prenatal genetic testing?

3. Current research needs in prenatal genetic testing
    a. What do you think are the research issues around prenatal genetic testing that are, for the most part, widely accepted?
    b. What in your opinion are things that need to be further researched related to prenatal genetic testing? How could these things be best studied?
4. Patients in prenatal genetic testing
    a. How would you describe women who come in for prenatal genetic testing?
    b. What social or demographic factors do you think are important in understanding women who have or do not have prenatal genetic testing?
    c. What do you think should be included when pregnant women are provided information about prenatal genetic testing?
    d. What do you provide women with in terms of written material about prenatal testing? Do you refer them to websites?
    e. How do you think individual women are affected by the testing?
5. The future of prenatal genetic testing
    a. Do you think prenatal genetic testing plays an important role in medicine? Is this kind of testing relevant and necessary for most pregnancies today?
6. Demographics
    a. How do you describe your race/ethnicity?
    b. What is (are) your professional title(s)?

c. What is your birth date?

Pregnant Women Interview Guide

1. First exposure to prenatal genetic testing.
    a. Are you in a relationship?
    b. How many children do you have?
    c. How did you get pregnant?
    d. Are you insured?
    e. How many times have you had prenatal testing?
    f. Please tell me the story of your first encounter with prenatal genetic testing.
    g. When did it happen? How did you see yourself during the process? Was it strange for your body to be focused on so intently?
    h. Where were you?
    i. How did you come to know about the testing?
    j. Why were you offered or pursuing testing? Did you feel pressure in any way from anyone?
    k. Did you have difficulties gaining access to the testing?

2. Knowledge about prenatal genetic testing.
    a. What is prenatal genetic testing?
    b. What kind of prenatal testing did you have?

c. What did you want to gain from having the testing? What does this mean to you? What do you think were your partner's feelings about it? And for your family?

d. What do you think are the risks involved in prenatal genetic testing? Were they relevant to you? How? Which risks, if any, were important to you and how did you manage them?

3. Information sources about prenatal genetic testing.

   a. Where do you get your information about prenatal genetic testing? Did you use the internet? Did you see things on TV? Was a particular book helpful?

   b. Do you think the information you got was useful? Why or why not?

   c. What sources have you found to be most trustworthy? Why?

   d. Did you talk to other pregnant women or women who had been pregnant who had prenatal genetic testing? What did they tell you? How did their experiences compare to yours?

4. Personal interpretations of prenatal genetic testing.

   a. Tell me about yourself at the time you had the testing. How did you see yourself? How has this experience affected the feelings you have about yourself as a woman?

   b. Do you see yourself differently now that you have the test results?

c. How did the questions asked about family biographies, such as birth defects and other health issues in your family and your partner's family, make you feel about your body? And your future child?

d. How did prenatal genetic testing affect your beliefs about pregnancy and perceptions of your child?

e. How did it affect your opinions of yourself and your body? Your health?

f. How did the testing affect your overall experience of pregnancy?

g. What would you tell a pregnant woman if she asked you about having prenatal genetic testing?

5. Demographics

   a. How do you describe your race/ethnicity?

   b. What is your birth date?

APPENDIX F CONFERENCES ATTENDED DURING THE COURSE OF RESEARCH

"Managed Care and Genetic Services: Translating Needs into Action." Berkeley Marina Marriott, Berkeley, California, September 28, 1995.

Pacific Southwest Regional Genetics Network

California Public Health Foundation

"American Society of Human Genetics 46th Annual Meeting." Moscone Center, San Francisco, California, October 30-November 2, 1996.

American Society of Human Genetics.

"The Human Genome Project: Science, Law and Social Change in the 21st Century." MIT Campus, Cambridge, Massachusetts, April 23-24, 1998.

American Society of Law, Medicine and Ethics

Whitehead Institute for Biomedical Research

"Building Bridges IV: Improving the Public's Health Through Research Partnerships." Oakland Marriott City Center, Oakland, California, May 6-8, 1998.

American Association of Health Plans

HMO Research Network

Agency for Health Care Policy and Research

Centers for Disease Control and Prevention

"Individual Genetic Variation: Implications of the Coming Transformation of Medicine." Stanford University, Palo Alto, California, October 17, 1998.

Stanford University School of Medicine

Stanford University Center for Biomedical Ethics/Program in Genomics, Ethics and Society

"Genes and Society: Impact of New Technologies on Law, Medicine and Policy." MIT Campus, Cambridge, Massachusetts, May 11-12, 2000.

American Society of Law, Medicine and Ethics

Whitehead Institute for Biomedical Research

"A Decade of ELSI Research: A celebration of the first ten years of the Ethical, Legal and Social Implications (ELSI) Program, NIH Campus, Bethesda, Maryland, January 16-18,2001

National Human Genome Research Institute

National Institutes of Health

Department of Energy

"People's Genome Celebration: Genetics is about ALL of us! Understanding and Celebrating our Shared Inheritance." Smithsonian Institution, National Museum of Natural History, Washington, D.C., June 8-10, 2001

Genetic Alliance

"Human Genetics, Environment and Communities of Color: Ethical and Social Implications." Columbia University, New York, New York, February 4, 2002.

WE ACT, West Harlem Environmental Action, Inc.

National Institute of Environmental Health Sciences

U.S. Environmental Protection Agency

NIEHS Center for Environmental Health in Northern Manhattan

at the Mailman School of Public Health, Columbia University

"Strategies in Genetic Counseling: Beyond the Basics." Hyatt Regency, Phoenix, Arizona, November 8-9, 2002.

National Society of Genetic Counselors

"American Society of Human Genetics 52nd Annual Meeting." Baltimore, Maryland, October 15-19, 2002.

American Society of Human Genetics

APPENDIX G

Are you pregnant?

Did you have amnio or CVS?

Volunteers needed for an interview
You must be currently pregnant

I am conducting dissertation research on prenatal genetic testing, asking questions about your feelings regarding the procedure, why you had testing, and the process of having the testing. Were you glad you did it?

The _ hour interview will be scheduled at a place and time convenient for you.

If you are interested in participating in my medical sociology dissertation research, please call Kristen at (718)783-1012

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APPENDIX H GLOSSARY

The focus here is on the necessary terms for a layperson to have an understanding of prenatal genetic testing. I largely avoided using medical textbooks. I also, when useful, provide more than one definition from different sources to avoid overly scientific framings.

Allele One of several alternative forms of a gene occupying a given locus on the chromosome. A single allele for each locus is inherited separately from each parent, so every individual has two alleles for each gene (Kevles and Hood 1992) . Variant forms of the same gene. Different alleles produce variations in inherited characteristics such as eye color or blood type (NIH 1995).

Alpha-feto protein test A noninvasive screening blood serum test given to women at 15 to 20 weeks gestation, providing an indicator as to whether a woman is at higher risk than other women her age, weight and gestational age for three anomalies: Down Syndrome, Trisomy 18, and neural tube defects. The test evaluates the amount of alpha-fetoprotein, human chorionic gonadotrophin and unconjugated estriol, all substances produced in the woman's body during pregnancy. This screening test has a 25% false positive rate, indicating that of 100 women who have the test and receive a positive result 25 will really be negative (Karlberg 2000). *also called the* ***expanded alpha-feto protein test****; see also* ***triple screen*** *and* ***quadruple screen*** *and* ***nuchal translucency test***

Amniocentesis A procedure, usually carried out at about the sixteenth week of pregnancy, that obtains a sample of the fluid (amniotic fluid) surrounding the fetus. The fluid is collected by insertion of a needle through the mother's abdominal wall and into the sac immediately surrounding the fetus. Studies of the fluid and the fetal cells contained within it can provide information about the fetus's chromosomes, genes, and chemical makeup (Zallen 1997). While there are many different miscarriage rates cited, depending upon the source, the range is between 0.5%-2%.

Aneuploidy A change in the number of chromosomes present in a fetus's cells (Andrews, Fullarton et al. 1994). The incidence of aneuploidies in newborns is approximately 9 in 1,000 births, making the risk per birth 1 in 109 (Cunningham, Gant et al. 2001). At least 8% of conceptuses are aneupolid, and aneuploidies account for 50% of first trimester spontaneous abortions (miscarriages) and 5-7% of all stillbirths and neonatal deaths (Cunningham, Gant et al. 2001). Aneuploidy in a fetus usually results in abnormalities.

Autosomal dominant A pattern of inheritance attributed to genes located on chromosomes other than the X and Y (sex) chromosomes. The trait or disorder will appear even when only one copy of the gene for that trait or disorder is present. Males and females are equally likely to be affected, and the trait can show up in successive generations of a family (Zallen 1997). Autosomal dominant disorders are produced by a single mutated dominant allele, even though its corresponding allele is normal (NIH 1995). *see also* ***dominant allele***-same

Autosomal recessive A pattern of inheritance attributed to genes located on chromosomes other than the X and Y (sex) chromosomes. Both copies of the gene in a gene pair must be flawed for a disorder to appear. Males and females are equally likely to be affected. The disorder can appear suddenly with no prior history of it in the family (Zallen 1997). Autosomal recessive disorders develop in persons who receive two copies of the mutant gene, one from each parent who is a carrier (NIH 1995). *see also* ***recessive allele***-same

Autosome A chromosome not involved in sex determination. The diploid human genome consists of 46 chromosomes: 22 pairs of autosomes and 1 pair of sex chromosomes (the X and Y chromosomes). Each parent contributes one haploid set of chromosomes (22 autosomes and 1 sex chromosome) to each offspring (NIH 1995).

Balanced Translocation (balanced chromosomal translocation) A transfer of parts of one chromosome to another; one chromosome will be shortened while the recipient chromosome, which may be part of a different pair, will be lengthened. If the number of chromosomes and the total amount of genetic material is not changed, the person will *usually* be phenotypically "normal". Some offspring of parents who have a balanced translocation will inherit the balanced translocation, meaning that they are "normal", but they can pass on the translocation to their children in both balanced and unbalanced form (Kolker and Burke 1994). *see also* ***unbalanced translocation***

Carrier A person who has a recessive mutated gene, together with its normal allele. Carriers do not usually develop disease but can pass the mutated gene on to their children (NIH 1995). An individual who has a gene pair in which one of the genes is flawed. The presence of the flawed gene is masked by the dominant functional gene (Zallen 1997).

Carrier test a genetic test performed to determine if a healthy individual has a flawed gene which, if expressed in his or her children, could lead to a genetic disorder (Zallen 1997).

Chorionic villus sampling (CVS) CVS is performed at 9-11 weeks gestation, either transabdominally, by inserting a needle through the woman's abdomen into the uterus to extract a sample of the villi surrounding the fetus, or transcervically, which involves inserting a long plastic catheter through a woman's cervix into the uterus and withdrawing a sample of the villi (Karlberg 2000). The cells are cultured and tested for information about fetal genes, chromosomes and chemical makeup. With CVS there is a 1-3% spontaneous abortion rate (CDGB, 1995a).

Chromosome Structures found in the nucleus of the cell, which contain the genes. Chromosomes come in pairs, and a normal human cell contains 46 chromosomes, 22 pairs of autosomes and two sex chromosomes (NIH 1995).

Down Syndrome The medical diagnosis is Trisomy 21, meaning there are 3 copies of chromosome 21 present in the individual, rather than only the necessary 2. The incidence

of Down syndrome is 1 in 800 to 1,000 newborns, and as such it is the most common non-lethal trisomy (Cunningham, Gant et al. 2001). Associated major abnormalities include heart defects in 30-40%, gastrointestinal atresias, neonatal or childhood leukemia, and thyroid disease. The average IQ of individuals with Down syndrome is between 25 and 50. Alzheimer's disease at much earlier ages than typical is common in Down Syndrome as well.

DNA Deoxyribonucleic acid; the substance of heredity; a large molecule that carries the genetic information that cells need to replicate and to produce proteins (NIH 1995). DNA has the form of a double-stranded helix. Each strand contains a long sequence of four types of chemical bases (denoted as A, C, G, and T). The sequence of bases makes up the genetic code containing the information for all of the proteins that an organism can produce. The helix is held together by strand-to-strand bonds, following the chemical rule that A connects to T, and G connects to C. DNA is located in the chromosomes within the organism's cells (Zallen 1997).

Dominant allele An allele whose phenotypic effect is expressed regardless of whether the organism is homozygous or heterozygous for that allele (Cobb 1997). *see also* ***autosomal dominant***-same

Early amniocentesis Same as amniocentesis, but conducted between 9 and 14 weeks gestation. Its safety and accuracy have not yet been adequately evaluated (Andrews, Fullarton et al. 1994).

Fetal cell sorting A *pending* possible prenatal diagnostic genetic test which holds the promise of noninvasive prenatal diagnosis by genetic analysis of fetal cells isolated from a sample of the mother's blood. This can be conducted as early as 9 weeks gestation. Limitations include the very small number of fetal cells present in woman's blood and the possibility of contamination from previous pregnancies (Andrews, Fullarton et al. 1994).

FISH Fluorescence in situ hybridization (FISH) is a modified cytogenetic technique intended for more rapid detection of chromosomal disorders in the fetus. FISH allows rapid cytogenetic assays based on in situ hybridization of chromosome-specific DNA probes that can be visualized by fluorescence methods. This technique can be used to count the number of copies of a specific chromosome present, to identify unknown chromosomes present in metaphase spreads, and to identify predefined chromosome translocations. At this time FISH is only available with a complete karotype analysis (Andrews, Fullarton et al. 1994).

Gamete A male or female reproductive cell. In the female, an ovum (or egg); in the male, a sperm (Zallen 1997).

Gene The fundamental physical and functional unit of heredity. A gene is an ordered sequence of nucleotides located in a particular position on a particular chromosome. Each gene encodes a specific functional product (Kevles and Hood 1992).

Genetic counseling A multifaceted interaction between a genetic professional and a client in which information about individual and family genetic risks is provided along with related information about tests, treatments, and reproductive options (Zallen 1997).

Genetic counselor Masters' level trained in a program emphasizing molecular genetics and medical counseling, they meet with pregnant women and explain the tests available, the risks involved, and the options after testing. Other work includes medical record chart review and family history taking (Karlberg 2000).

Genome All the genetic material in the chromosomes of a given organism (Cobb 1997).

Genotype The actual genes carried by an individual (NIH 1995). The total genetic, or hereditary, constitution that an individual receives from his or her parents (Kevles and Hood 1992).

Integrated test A screening test which combines ultrasound findings (NT) with various blood serum measurements (such as the triple or quad screen) to determine an approximate risk for Down Syndrome.

Heterozygote An individual having different alleles of a particular gene (Cobb 1997).

Homozygote An individual having identical alleles of a particular gene (Cobb 1997).

Karyotype An organized picture showing all of the chromosomes in a cell (Zallen 1997).

Medical geneticist Medical doctor with a residency in a specialty such as pediatrics, internal medicine, or obstetrics and then a specialty in genetics. They diagnose and manage genetic diseases. The main roles include interpretation of results of genetic tests, making a diagnosis and management following the diagnosis (Karlberg 2000).

Multifactorial disorder A disorder which is brought on by the joint action of multiple factors. The contributing factors include several different genes as well as various types of agents from the environment. (Zallen 1997)

Mutation Any permanent change or alteration in the genetic material (gene)(Zallen 1997). A change in the number, arrangement, or molecular sequence of a gene (NIH 1995).

Nuchal fold translucency Nuchal translucency (NT) is the area at the back of the neck, the size of which increases with gestational age. Nuchal fold translucency measurement is an indicator of risk of fetal aneuploidy, and is conducted at 10-14 weeks gestation (Chitty 1999). This test is most commonly used to detect Down syndrome, and is most effective when used in conjunction with other screening tests. *See also* ***integrated test***

Phenotype The appearance and other physical characteristics of an organism, a result of the interaction of an individual's genetic constitution with the environment. Phenotype differs from genotype in that it includes only the outward manifestations of genes (Kevles and Hood 1992).

Polygenic Pertaining to the combined action of alleles of more than one gene. Height is an example of a polygenic trait, as are predispositions to different types of heart disease (Kevles and Hood 1992).

Perinatologist A medical doctor who completed a residency in obstetrics and gynecology and then earned a subspecialty in perinatology. The training is specific for managing high-risk pregnancies and treating both mother and fetus until delivery. Normally, this is the physician who performs the prenatal genetic testing (Karlberg 2000).

Predictive genetic tests Tests to identify gene abnormalities that may make a person susceptible to certain diseases or disorders (NIH 1995).

Preimplantation diagnosis Tests a woman's removed, fertilized egg which has divided into four to eight undifferentiated cells by removing one of these cells for genetic testing. The tested egg---soon to be embryo--- is then implanted in the woman's uterus to continue development. Risks include inability to implant the fertilized, tested egg and

very little is known about the technical and biological safety of these techniques (Andrews, Fullarton et al. 1994).

Prenatal test A genetic test performed during pregnancy to obtain information about the chromosomes or genes of a fetus (Zallen 1997).

Presymptomatic genetic test A genetic test performed to determine if a gene(or genes) are present which will bring on a health problem later in an individual's life (Zallen 1997).

PUBS Percutaneous umbilical blood sampling (PUBS) is used to obtain a fetal blood sample at approximately 18 weeks gestation through inserting a needle guided by ultrasound into the umbilical chord. The blood can be tested to clarify ambiguous chromosomal analysis from amnio and CVS and also to evaluate for hematological abnormalities and diagnosis of some inborn errors of metabolism. PUBS has a 5% rate of fetal loss, so is used only when there is no other method appropriate (Andrews, Fullarton et al. 1994).

Quadruple Screen Quad screen. A genetic screening test which examines levels of alpha-fetoprotein, unconjugated oestriol, free beta-hCG and inhibin-A between 14 and 20 weeks to determine risk for Down syndrome, Trisomy 13, Trisomy 18 and neural tube defects (Wald, Huttley et al. 2003).

Recessive allele An allele whose phenotypic effect is expressed only in the homozygous state (Cobb 1997). *see also* ***autosomal recessive***—same

Screening Looking for evidence of a particular disease such as cancer in persons with no symptoms of disease (NIH 1995). A search in a population for persons possessing certain genotypes that (1) are associated with disease or predispose an individual to a disease, (2) may lead to disease in the individual's descendants, or (3) may produce other variations not known to be associated with disease (Nantowicz and Alper 1991).

Sex chromosomes The X and Y chromosomes. Females have two X chromosomes; males have one X and one Y (Zallen 1997).

Sex linked Denotes a relationship to a gene located on a sex chromosome (usually the X chromosome) (Cobb 1997).

Spontaneous abortion The expulsion of products of conception without mechanical or pharmacological intervention (Wilson 1995). This experience is more commonly referred to as miscarriage.

Susceptibility test A genetic test for a gene whose presence can increase the chances of developing a health problem later in life. The problem may not develop even if the damaged gene is present, and it may occur even if the gene is absent (Zallen 1997).

Translocation (chromosome translocation) A transfer of parts of one chromosome to another; one chromosome will be shortened while the recipient chromosome, which may be part of a different pair, will be lengthened (Kolker and Burke 1994). *see also* ***balanced translocation*** *and* ***unbalanced translocation***

Triple screen *see* ***alpha-feto protein test*** *-* same

Unbalanced translocation (unbalanced chromosome translocation) A transfer of parts of one chromosome to another; one chromosome will be shortened while the recipient chromosome, which may be part of a different pair, will be lengthened. If only one of the two defective chromosomes is inherited, a change is made in the total genetic material, resulting in an "abnormal" fetus/baby. The type of "abnormality" depends upon the amount and types of genetic material missing or added. An unbalanced translocation may be incompatible with life. Some offspring of parents with a balanced translocation receive an unbalanced translocation, which results in "abnormality" while others receive the balanced translocation in the same configuration as the parent (Kolker and Burke 1994). *see also* ***balanced translocation***

Ultrasound A noninvasive process which can assess gestational age, position of the fetus and sometimes sex, evaluate fetal growth and development, guide the instruments in amniocentesis and chorionic villus sampling, identify certain structural birth defects (such as missing limbs, some cleft lip, and spina bifida), and can help identify certain

high-risk pregnancies for which cesarean delivery would be appropriate-for example with some neural tube defects such as hydrocephalus (Andrews, Fullarton et al.1994).

X-linked dominant A pattern of inheritance attributed to genes located on the X chromosome. A disorder will appear when one copy of the gene for that disorder is present. Affected males pass X-linked dominant genes to all of their daughters but none of their sons. Affected females pass X-linked dominant genes, on average, to half of their daughters and half of their sons (Zallen 1997).

X-linked recessive A pattern of inheritance attributed to genes located on the X chromosome. Males with the gene will be affected because all the genes on their single X chromosome will be expressed. Females who have two X chromosomes, can be carriers. Affected males in a family are related through females (Zallen 1997).