Killing Mendel

Prenatal Testing and its Effects on Family and Society

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Abstract

Our goal was to learn about modern prenatal testing and its impact on children and families as well as on society. We did this through philosophical analysis of the research we conducted prior, regarding prenatal testing and modern genetic counseling techniques, and wrote a paper presenting all sides of the issue. We concluded that there exist positive and negative repercussions to testing future children for genetic disease.

Executive Summary

Our goal was to gather information about current prenatal testing and to learn about its possible impact on children, families, and society. Recently a number of advancements have been made in genetics including the completion of the sequencing of the human genome, better methods for sorting X- and Y-carrying sperm, and cloning. Some of the possible uses for this technology are to test unborn children for genetic defects, or to sort fertilized eggs for desirable characteristics prior to implanting one into the mother's uterus. As more people use prenatal testing it is important that we understand what sort of impact it will have on families and on the children who are tested. When these techniques even more widely used there may be an associated change in society.

In order to create our report we first conducted research on the history of genetics, specifically prenatal testing, and on genetic counseling. Most of our research came from books, but we also used recent magazine publications, old periodicals, recent government publications, and an email interview with a professional genetic counselor. The purpose of our research was to familiarize ourselves with the current technology and how it has evolved over time, along with the social changes that have allowed us to advance to a point where genetic testing is valid. We also traced the evolution of the genetic counselor and his techniques. From the more recent publications, we drew a picture of the state of technology now, its limits, and its capabilities. Our final sources were bioethical considerations from both past and more recent writers on the possible social implications of genetics as well as how it will affect the family.

The next step was to combine our sources into a cohesive paper. The final product was broken down into historical, philosophical/ethical, and technological sections. Each team member was assigned sections to write. We, then, worked as a group to make the individual components blend into one cohesive piece of work. We, also, collaborated to develop the introduction and conclusion to the paper in order to ensure that elements of each group member's sections were included.

At the end of the project we concluded that prenatal testing can have a great effect on the ways that a family will perceive their children, how the children will grow up, and how a society of children who have gone through these tests will differ from one that has not. In looking at genetic counseling, we can see that a lot of training and practice must go into this profession in order to offer nonbiased, yet helpful, information that allows the patients to fully understand their choices without feeling pressured. Additionally, we realized that no one opinion regarding the ethical debate of prenatal testing will ever be agreed upon due to the religious and cultural diversity that exists not only nationally but, also, globally. As more variances are introduced into the problem, such as religious variety within a population, a greater amount of differences will exist among the opinions expressed by that society.

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Introduction

Since the turn of the century there has been an escalating interest in the field of genetic engineering, a flood of new inventions technologies and methods, and a whole host of ethical questions. In more recent years the 'what if?' and 'if we could?' questions have started to turn into 'should we?' and 'now what will happen?'. In the past few years, genetic screening and testing, the ability to analyze and choose the qualities of a child, and the statistics to predict the life of a person based on genetic information has become a reality. We have gotten to the point where early detection of many diseases is either ready or at the door. Insurance companies and adoption agencies already consider genetic information to be relevant and even critical for approval. There are also more ethical, social, and personal questions that are perhaps more important than the technologies themselves. The question of how such information impacts the family and the development of the child is a great concern for genetic counselors. The strain of choosing which tests to take before giving birth and what to do with the information a counselor may provide are hard-hitting issues that will only become more and more prevalent in society as genetic detection technologies become more widespread.

The purpose of this paper is to explore the potentials of prenatal testing looking at the topic from multiple angles including historical, technological, ethical, philosophical, social, and personal. The possibilities as well as the limitations of current and future technologies will be considered based partially on analysis of past technological advances and their effects. It is important as students of science and engineering to be concerned with the impact that we can have on people and society. Numerous courses and classes are dedicated to educating students in the areas of business conduct, ethics, and

professional behavior. Understanding how we can change the world for better or for worse is an important part of anyone's education. The arguments and information given herein are to provide information for anyone working in the field and for anyone wanting to be knowledgeable for personal involvement in the future of technology. The burden of understanding technology, especially involving personal issues like childbirth, cannot fall solely on the physician. It is common for patients to make decisions solely on the word of their doctor without completely understanding the implications of their choices. In order to make intelligent decisions in life we must all make an effort to be well informed. To this end, our discussion will not uphold or defend one side of the debates we present; instead we will analyze both sides to present all possibilities and views. The question with prenatal testing is whether it is better to know or not to know each child's genetic state. Then, if it is better to know, how much information should parents and children receive?

Factors that effect this choice include the certainty to which we can predict genetic diseases and other traits with current technology. Genetic counselors must at times make decisions as to what information to report and what to omit when offering guidance. What basis should they use for this choice? Do parents have a similar option? In order to make these decisions easier, it is important that the physician, as well as the patient, be well educated on genetic testing and the impact it can have before making a choice. Therefore, beneficiaries of this study will be both genetic counselors and their patients. Now, as the 'genetic age' approaches, it seems to be of paramount importance that the general public, not only knows, but has a good understanding of prenatal diagnosis capabilities and implications of choices made thereof.

For the purposes of our discussion, we will be omitting the ethical analysis of abortion. Although issues concerning abortion in conjunction with prenatal screening are relevant, it is another project in and of itself.

History of the Development of Technology and Counseling

Key dates / What events allowed other events to occur

Present day technologies are yesterday's future developments. They once began as ideas that initiated an interest in the research of the respective topic, and eventually leading the project into the experimentation phase. It is this process that has continued to evolve the world of science and technology. It was Reinier de Graff's interest in the human egg in 1672 that initially paved the way for the future of genes and genomes. De Graff's description of the egg initiated further research and eighty-seven years and several scientists later Caspar Wolff uncovered the mystery behind the ovum's function. Genes would not be understood if the sperm had not first been explored and understood. Antoni Van Leeuwenhook's description of the sperm in 1767, five years after understanding the functionality of the egg, opened the doors to understanding the reproductive process and its constituents. (Genetic Engineering: A Documentary History, p. xxi-xxii)

Understanding the human embryo required exploration within other living and non-living systems at various levels of development. One of the main contributions in genetics was the discovery and understanding of traits. This discovery is attributed to Gregor Mendel. Mendel had an interest in nature and questioned the atypical characteristics that plants possessed. He planted an atypical plant next to its typical variety and allowed them both to flourish. After examination of the offspring and comparison to their parents, Mendel found that the offspring were similar to the parents and introduced the idea of heredity. His enthusiasm spurred further experiments with mice and peas. The result of crossing peas and mice showed a numerical significance,

such that ratios were observed in according with the inheritance of traits. Once again an idea was instigated, the idea of dominance and segregation of genes. Using peas to test this proposal, he was able to map the laws of inheritance by tracing characteristics of the pea plants across several successive generations. Thus, in 1865 Mendel's Laws were founded. (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80).

Nearly four decades later, Mendel's influence was finally accepted by the world of science. In 1902, Walter S. Sutton, a graduate student of Edmund Wilson's in the Zoological Lab at Columbia University, developed the basic presupposition of the chromosome theory of heredity based upon studies done on locust chromosomes. Additionally in 1902, McClung, in the Zoology and Histology Lab of the University of Kansas, discovered sex chromosomes but was unable to resolve how they functioned. Edmund Wilson focused on the cellular basis of heredity, specifically the importance of the chromosome, between 1903 and 1938. His work included investigation of the physiology of the chromosome, chromosome movements, spindle formation, and the independence and replication of chromosomes. He worked independent of Nettie Stevens, a female student from Standford, to determine what McClung was unable to conclude about chromosome functions, as well as to figure chromosomal distinctions within a variety of species of insects containing XY and XO males. Nettie Stevens and Edmund Wilson described the behavior of sex chromosomes and distinguished between males, XY, and females, XX. (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80).

Previously unconvinced of Mendelian findings, Thomas Morgan, a PhD. recipient from Johns Hopkins who taught at Columbia and conducted his research in the "Fly Lab" there, utilized Drosophila in 1908 in order to test for the origin of variations within a species for the purpose of understanding how evolution occurs. In 1910, Morgan discovered a recessive sex-linked mutation in the flies, white eye color, thus showing that crossing a mutated chromosome will allow for the possibility of future generations to inherit the mutation. This and other concurrent experiments with Drosophila confirmed the Chromosome Theory of Heredity during what is known as the "Drosophila era". Finally, in 1913 Alfred Sturtevant, an undergraduate at Columbia who worked alongside Thomas Morgan in the fruit fly lab at Columbia, completed the first gene map while he was working in Morgan's lab. (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80).

Clinical medicine and experimental physics had been using X-ray technologies since their inception in 1895. It was only in 1927 that this technology was employed by the genetics field of research and development in order to further explore genes, heredity, and mutations. Hermann Muller, an American geneticist who conducted research at Columbia University, used radioactivity in order to create point mutations in Drosophilae for the purpose of exploring the theory that radioactivity would interfere with the normal molecular structure of single genes. He attempted mapping genes to particular chromosomes in hopes that these principles could be applied to improve the human race. Inspired by Muller's use of X-rays, in 1934 John Bernal, a physics professor at Cambridge University, utilized X-ray crystallography in examining protein structures. These advances in science demonstrated an evolution in the application of technology in

genetics and the biological sciences. (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80; Prenatal Testing And Disability Rights, 44-53).

George Beadle, a geneticist who worked in Thomas Morgan's lab at Columbia University working with fruit flies, proposed the idea that the color of one's eyes is characterized by a series of genetically determined chemical reactions. Working with Edward Tatum, a biochemist,he further examined the theory that chemical reactions establish inherited characteristics. In 1941, Beadle and Tatum turned to Neurospora, a fungus, in order to more easily examine metabolic products by producing mutant genes in some and crossing them with the non-irradiated Neurospora. This experiment succeeded in representing the concept that genes produce enzymes, as Garrod's, a physician in London, unrecognized contributions had previously represented. Beatle and Tatum's work influenced what has become today's knowledge in the fact that the individual genes each specify the production of a single polypeptide. Oswald Avery, an immunochemist at Rockefeller Institute for Medical Research, was responsible for the detection of DNA's responsibility in determining the chemical foundation of transformations in bacteria. He found that deoxyribonucleic acid holds a primary role in determining characteristics during reproduction. Initially his writings were not well read by others but he was an instigator in future DNA research. (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80).

Avery's findings in the significance of DNA led to James Watson, an American who had received his doctorate, and Francis Crick's, a graduate student at Cavendish Laboratory in Cambridge, conclusions, nearly a decade later in 1953, that DNA was composed of complementary base pairs. It was seen that the order of the bases within the

DNA encoded the genes. The chromosomes were known to house the genes. The composition of the genes was known to be comprised of proteins and DNA. Together, Watson and Crick uncovered the truth that DNA was the "master molecule". Through their interest in DNA's mysterious elementary importance, it was found that this master molecule not only contained the genes but was capable of self-replication and was responsible for merging again during reproduction. Their success did not come without a two year period of experimentation. They followed Linus Pauling's, who was a graduate student in Chemistry at the California Institute of Technology, lead in attempting to describe DNA. They built a three- dimensional model of the chainlike structure using cardboard and sheet metal. Recognizing the possibility that the molecular structure might be of a winding helix shape, another model was constructed. Finally, two years after the commencement of their studies, Watson began to understand the mystery behind the arrangement of base pairs about a sugar and phosphate backbone. He realized that if the complementary base pairs were held together by a hydrogen bond then they would be identical in shape, and if they were arranged in long chains then it was a good assumption that they would form a double helix. Understanding the base pairs and genes themselves helped to demonstrate that separation of the genes allowed for replication and reproduction. It was Watson and Crick's discovery of the molecular structure of DNA that initiated the future boom in science. (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80; Prenatal Testing And Disability Rights, 44-53; Genetic Engineering: A Documentary History, p. xxi-xxii).

Over the next few years, Arthur Kornberg, who attended the University of Rochester for his medical degree and worked in various hospitals and health services,

worked to crystallize DNA polymerase. With crystallization of this DNA synthesizing enzyme in 1956, many more doors would open in the years to come. In 1957, Crick changed the world of biology. He proposed that the primary function of genes is to manufacture proteins. He also suggested that the proteins, in compilations of twenty amino acids, form the basis of the processes of life. The genes were seen to be responsible for the order and assembly of amino acids, otherwise referred to as the building blocks of proteins. Additionally, he proposed the theory of the sequence hypothesis and the theory of the central dogma. The idea of the sequence hypothesis is that each order of complimentary bases along a section of the DNA symbolizes the amino acid sequence of a particular protein. This hypothesis helped to create the idea of "triplets", combinations of three bases, which sequenced the code for a sole amino acid. The central dogma represented the theory that DNA and RNA relay information to the proteins, but the proteins cannot reciprocate this function. His insight into the role of cytoplasmic RNA and the process of transmitting information from DNA to the cytoplasm of the cell prefaced the developments that followed. (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80; From Genes to Genomes, 5-19).

Beginning around 1960, the structural difference of RNA from DNA became known. Uracil in RNA was substituted in place of the thymine in DNA. Examination of the cells revealed that the cytoplasm contained ribosomes, which housed the RNA, whose presence is essential to protein synthesis since it can not occur without them. Following experiments with E.Coli as subjects, it was determined that there existed an additional form of RNA. This molecule was referred to as messenger RNA, or mRNA. Eventually,

several forms of RNA were discovered and distinguished. Messenger RNA, ribosomal RNA (rRNA), and transfer RNA (tRNA) each carry out a particular function specific in the process of protein synthesis. By 1961 the previously proposed idea of the "triplet" became confirmed. Marshall Nirenberg was responsible for the discovery of the first "triplet", or triple base DNA sequences. Within the next five years the genetic code as a whole was interpreted. Nirenberg, a biochemist at the National Institute of Arthritic and Metabolic Diseases, and Johann Matthaei, a German scientist, added RNA chains containing a sole base to a test tube comprised of a cell-free system and E. coli "sap". Radioactive tagging allowed for the identification of the synthesis of molecules similar to protein, which were completely comprised of phenylalanine, and for conclusions to be drawn regarding the effects of homogeneous uracil sequences on phenylalanine and protein chains. The purely uracil RNA sequences directed the addition of phenylalanine to any protein chain that was not complete. . (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80; From Genes to Genomes, 5-19).

Nirenberg had interpreted thirty-five triplets by 1963 and sixty by 1966. Codons had the possibility of forming 64 triplets since only three bases out of the four possible are used in each codon chain. It was found that three of the codons signaled the end of an amino acid chain. Har Gobind Khorana, who conducted his research at the University of Wisconsin, was responsible for adjusting Nirenberg's experimental system in order to further the work previously done. Howard Temin, who was a graduate student at the California Institute of Technology and worked in the lab of one of his professors, and David Baltimore, who received his PhD from Rockefeller University and later taught at MIT, both individually discovered reverse transcriptase. It was found that the reverse

transcriptase enzyme made DNA from RNA. (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80).

Experiments done on bacteria, that were performed thirty years prior, were further examined in 1970 when bacteriophages helped develop molecular biology. Bacteriophages are responsible for destroying bacteria with these parasite-like constituents. It was these experiments done one bacteriophages that led to the discovery of restriction enzymes, which bacteria use for protection. They assisted in manipulating DNA and provided the base from which genetic engineering grew. (The Code of Codes: Scientific and Social Issues in the Human Genome Project, 37-80).

Paul Berg, a professor at Stanford University, combined genes from various organisms to assemble a DNA chain in 1972. He developed methods to individually isolate genes and put them into mammalian cells for the purpose of examination and allowing their protein products to be expressed and manufactured. Berg created the cut-and-splice method which led to "sticky ends", in which complementary DNA bases were added to ends of molecules using the enzyme transferase. Before utilizing this method in bacteria, Berg realized that there existed potential hazards. In 1974, he wrote of the potential dangers of recombinant DNA research. This led to the regulations on research, which were devised in 1976. (From Genes to Genomes, 41-63).

In 1973, Herbert Boyer, a graduate of the University of California at San Francisco, and Stanley Cohen, a student at Stanford University, contributed to the utilization of living organisms in genetic engineering that Paul Berg had just recently explored. They used Berg's techniques for inserting viral DNA into bacterial DNA to inform on the construction of organisms that joined and replicated genetic information

from various species. This showed the influence that recombinant DNA engineering could have on various medical fields, industry, and agriculture. But there existed difficulties in reading DNA sequences. In 1977 Walter Gilbert, a doctorate recipient from the University of Cambridge, and Frederick Sanger, a biochemical PhD. student at Cambridge, worked independently to develop a way to rapidly sequence DNA. Their techniques allowed for identifying the nucleotide sequence for complete genes in compilations of 1,000 to 30,000 bases. (From Genes to Genomes, 161-177).

1978 signified an advancement in gene mapping. David Botstein, a genetics professor at Standford, used restriction fragment length polymorphisms, or RFLPs, to identify differences in genetic codes among individuals. By 1983, replicating DNA had reached a new front. Kary Mullis, who received his PhD. in biochemistry under J.B. Neilands in Berkeley, California, found and developed the technique that polymerase chain reaction would help to quickly multiply sections of DNA. Applied Biosystems Incorporated was responsible for developing the first automatic DNA sequencer in 1986. The inventor, Leroy Hood, a biologist at the California Institute of Technology, followed advancements made by Walter Gilbert and Frederick Sanger in order to create an automatic human genome sequencer, which was just an improved method of enzymatic sequencing. This instigated the idea that the entire human genome could be sequenced by decoding the entire base-by-base complement of human DNA for medical benefits and in 1990 the Human Genome Project was funded and launched. (Prenatal Testing And Disability Rights, 44-53; From Genes to Genomes, 161-177).

Shortly after launching the Human Genome Project, J. Craig Venter, with the National Institute of Health, introduced a way to increase gene discovery. He developed

Expressed Sequence Tags (EST's) to find genes and investigate their roles. They were accurate and efficient, demonstrating accuracies comparable to that of automated sequencing technology. In 1995, J. Craig Venter and his colleagues patented the original completely sequenced genome, Haemophilus influenzae Rd., a self-replicating, free-living bacterial organism. Continuing the use of Drosophilae in genetic research and experiments, Celera genomics and the Berkeley Drosophila Genome Project sequenced and assembled the complete Drosophila's genome within eight months in 1999. Due to the similarities between Drosophila genes and human genes, Drosophilae expressed the potentials that comparative genomics possessed in conjunction with medicine. (From Genes to Genomes, 259-304).

The turn into the next millennium, 2000, prompted competition between The Human Genome Project, which was funded by the government, and Celera Genomics to sequence the human genome in its entirety. Speaking of their advancements at the White House on June 26, 2000, Celera Genomics announced that they had completely sequenced the human genome within four months and assembled the DNA base pairs within the following six months. Additionally, The Human Genome Project announced their developments in regards to the project at hand, however they had only sequenced part of the human genome. Since Celera had made such advancements, the primary focus of the project was modified so that the emphasis was placed on interpreting the genome rather than its sequencing. This re-directed the project in order to assist in understanding variances within genes and gene mutations.

Even in present day the controversies over cloning of humans and animals, stem cell research, and genetic modifications in humans, animals, and agriculture continue.

Knowledge and application of science is always growing; especially in today's society, it is expanding faster than the majority of the population is prepared to face it. The research and developments that result in furthering existing technologies in the field of genetics are bringing about many concerns regarding the intentions of use and potential abuse of the techniques. The questions that arise will be discussed in the following sections.

Social Attitude Changes Permitting People to Undergo Prenatal Testing

One major consideration to keep in mind when discussing genetic engineering and the changes in lifestyle that technological advances and new discovories have afforded us, is that unlike many other areas of development, genetic engineering is a very people centered technology. Whether it's diagnosing genetic defects or treating them using engineered cells or bacteria, genetics is very personal and makes a great deal of difference to a great numer of people. This is not to say that early detection of genetic disease will not have any far-reaching effects or possible impact on society as a whole, but it must be remembered that at its core it is a personal and a family matter. This means that people will react differently to this budding technology based on more than accuracy and availablility. Issues like race, culture, and religion will be very large players in future debates on the uses of the technology we are currently developing. These debates are not new either, since the earliest societies, there have been debates on ethics and eugenics by one name or another and we cannot expect cultural differences to melt away with newer technological advances or education. We can however expect integration and change.

In the vein of historical changes, society too has evolved along with, and at times because of, technology. It would be impossible to integrate something like the computer

into a culture that had not yet grasped the meaning and power of electicity. Modern medicines would not be as beneficial to people from the distant past who had not yet accepted the science of medicine and the trust that people have for their doctors. Genetic engineering and testing will and has worked in the same way. In the days when women rarely saw doctors for serious and "private" needs, because most doctors were male, genetic screening would have been impossible even if they had the technology. Society needed to change in order to allow for issues like pregnancy and human pedigree before the idea of taking samples from a pregnant woman's uterus would be considered acceptable.

Integration of technology and interpritation of genetic morality is not only a matter of evolution but a matter of society and culture. A standard code of morality has never been, and most likely never will be, universal. Key cultural differences, especially religion, normally form people's interpritations of right and wrong. This is not to rule out individuality and free thought, but on the average there are cultural lines drawn on most moral issues. Culture and religion are of special importance to an issue as personal and spiritual as childbirth. Many view it as the ultimate act of love and human creation. Where do doctors and probes fit into this picture? Would the sanctity of life and of procreation be soiled by such technological intervention as prenatal testing and evaluation? If it is God's will that a family have a child with genetic disease, who are we to interferre? These are the sort of questions that await us as we develop the technologies of prenatal testing and screening. This is not to say that these persuits should be abandoned, but the feelings and beliefs of others must be considered and respected at all times.

Societal and Cultural Views

Some of the earliest views on genetics and eugenics come from the ancient Greeks. Based on the simpler relationships of heredity by observation, i.e. blond parents have mostly blond children, etc, it was common knowledge to the educated at the time that healthy strong parents more likely give birth to healthy strong children. This was tied closely to moral perfection and proper upbringing. Morally upstanding and "good" people more likely gave birth to and raised moral and "good" children. It was understood that biological and social are not exactly the same but they were very closely tied. This may not seem important but keep in mind that the general culture of the United States does not usually include good breeding as part of being a good person. This is not always the case but more often money is an important factor in modern North-American culture. The point is that cultural differences must be understood and considered in matters such as genetic testing. For instance, many of the dialogues of Plato begin ethical debate with the assumption that euthenasia is a completely acceptable and socially beneficial practice. He also proposed that citizens of his ideal city to be carefuly evaluated in secret. All children were to be taken from their parents at birth and those from parents most likely to produce the best children were allowed to live and be raised while others were left outside the city walls to die of exposure. The very best of the children were then raised to be the rulers of the city. Most people today would be appalled at such a suggestion but it is a good example of how an enlightened culture can seem barbaric from the point of view of others. (Plato).

Modern China is a good example of a cultural difference from people of North America today when it comes to children and selection. With only one child per family

allowed it is prefrable to have a male child who can work and be of more economic use to the family. Due to this regulation on the number of children a family can have, it is common to abort female children and try again for a male. This may also be common in societies where marrying off daughters costs includes a dowry. Logically, in cultures where the dowry goes the other way, i.e. the family with the son pays for the daughter, the opposit preferance may exist. Examples of countries like this are Zimbabwe, Botswana, Zambia, and Mozambique, where it is common practice for the family of the groom to present a large monetary sum to the family of the bride. We would expect for there to be preference for children with qualities like stature, good looks, intelligence, etc, but it may surprise people to see hidden cultural differences when it is preferable for children to have specific qualities beyond what we may expect like hair color, eye color, neck length, nose size, and other things not common to North American culture.

There will also be cultural tension surrounding this new technology when we get to the simple matter of prenatal testing and the doctor's relationship with the patients of different countries. The ways in which people of various nations view the acts of sexual intercourse, child birth, and child rearing are also very important factors to consider in relationship to past and new technologies. In the United States, birth control methods have been an important factor in the modern sexual revolution. Devices like the condom help prevent the spread of disease and allow people to easily control pregrancy with fair reliability. After such success in most of the world, United Nations peaceworkers were shocked when they found out that many of the condoms they were distributing in the third world were being regularly pierced with needles before use. It was belived that semen was a manifestation of the male spirit and it would be wrong to imprison it within

the condom. Although this seems like a simple lack of education, it is in fact an important cultural difference. In order to understand the beliefs of other people it is vital to step out of one's own cultural bubble. Although difficult, this is the cornerstone of diplomatic relations. As the world continues to be separated by technology, it will also be the cornerstone for understanding cultural assimilation (or not) of these technologies.

Sex, Race, Wealth, Education, Religion

Beyond cultural background, race, social status, income, religion, and education are important features that will cause differences in interpritation and use of genetic technology. The ongoing debate on stem cell research is not limited to Republican vs Democrat, liberal vs conservative, it is also a religious and cultural battle. Even with a seemingly simple issue of personal choice like prenatal testing, serious religious and social issues erupt. It will be considered murder by many to search through prospective fetuses and select the "best" to keep and the rest to get rid of. Going beyond the expected debate on abortion, what about cultures in which monitoring or controlling other aspects of a child is just as unacceptable.

Differing levels of education will play a major role in how these new technologies are accepted as it has always in the past. Ideas like percentages, chance, medical risks, and the care required by a child with genetic defects may be difficult for some families to understand depending on their background. Although it is true that simply understanding statistics cannot lessen the blow of a lost child, it is still important to grasp the risks at hand. People with less education may be inclined to take the word of their doctor at face

value, or may simply assume that any tests he recommends are nonessential and are just an attempt to make more money for the medical business.

Economic status too will play a major role in the way that this new technology is accepted. Extensive medical treatments beyond healing the sick have historically been reserved for the middle to upper class. Countries like Canada have worked hard to guarentee health care for every citizen, but things like genetic counseling, which is more of a luxury, will continue to be reserved for the well-off. In past times it became tradition for mothers of families with enough money to spare to turn care of infants over to wet nurses, even for simple things like breast feeding. Those who could not afford a wetnurse would have to care for their own children. In this manner certain family behavioral trends will tend to follow along economic lines.

Views of the Tested

It is difficult to say the exact effect that prenatal testing and evaluation have on people and their families. The technology is fairly new and, although it has been applied, it has not had the test of time. There are few children who can say "Yes, my parents tested me at 2 months, and decided to keep me despite the risks; this is how it affected my life...". Since we do not have a wealth of first-hand information like this, it is much more feasible to go on accounts from genetic counselors, doctors, and parents who have had experience with these situations. In 1992 the American Journal of Human Genetics did a general reseach program into genetic discrimination. The study was not a comprehensive tabulation of data but more of a sampling of specific situations people have to report. Even at that time, people reported genetic discrimination in the workplace

and in attaining medical assistance. On the upside, they also reported great benefit from the increased certainty with which doctors would diagnose children who had prenatal screening evidencing a condition.

Changes Over Time

The general trend with technology and society is that time and education breed acceptance. Ideas like nuclear power is slowly being accepted by most americans as an efficient and safe way to produce energy. Chemical additives in food are a part of daily life in the United States when once only home-grown and natural would do. Genetically engineered foods have been in existance for quite a while, and things like extensive prenatal testing may one day become commonplace for all pregnancies. The question is, what effect will this have on society as a whole? Will we benefit from this or will society be changed in a negative way? What cautions and understandings must be made in order to preserve some of our current ways of life as we move ever forward into the genetic tomorrow?

Path to Technology

Human Genome Project

The focus toward human genes from animal genes shifted over the past few decades, showing sciences' increased interest in the 1980s. The Human Genome Project (HGP) has paved the way for future technologies in diagnosing and treating physiological, as well as behavioral, diseases. Genes influence talents, traits, addictions, and diseases, or more generally they help to identify us as individuals. Genes, although they influence our characteristics, do not determine who we are. Persons can work to develop talents or may just happen to develop an addiction that their parental alleles did not possess. For example, someone may be a slow reader but they can learn to become faster by taking courses and practicing new techniques. Or, someone may start drinking at a young age and, even though alcoholism does not run in their family, become addicted to the way it makes it them feel. The concern of the HGP was to determine human conditions in present time and in the future through knowledge about determinants of or influences on human disease. Along with the use of the HGP as a path to technology came, and continue to come, many concerns.

Current beliefs have long emerged from the belief in the genetic composition of physiological components to the belief in genetic influence on behavior, and more recently to the knowledge that genetic components can be individually identified and modified. Since the 1920s, there has existed a general belief in regards to intellectual and physiological attributes as inherited traits. In addition to the inheritance of DNA, it was a topic for discussion whether morals and behavior, as well, were hereditary. Diseases,

whether they be physiological or behavioral, consist of genomic make-up that often stems from the parents' alleles. These concerns are what helped to shape the goals of the Human Genome Project and continue to be the driving force behind understand the human chromosome. (The Code of Codes, 281-299).

One of the primary focuses of the HGP was to increase the geneticists' knowledge of humans as a compilation of genomes. Watson and Crick's contributed efforts in identifying genes as sequences of DNA set the foundation upon which future developments in the genome project were built. Beginnings in the project included developments in sequencing, synthesizing, and altering the "master molecule". Genomes are mapped in order to investigate the codes of diseases and other traits. In identifying diseases through their composition of genes, early detection and prevention measures can be evaluated and further developed. Techniques of detection include prenatal diagnosis and findings throughout the term of life, generally utilizing ultrasounds, blood tests, biopsies, and other tests in order to best determine the expression or possible expression of the diseased gene or genes. Through the HGP it is hoped that diseases will be able to be better understood and detected. Additionally, the project hopes to lead geneticists to gene therapies so as to fix the codons and gene sequences which could otherwise cause future illnesses.

The future of healthcare is continually becoming more sophisticated with the advances in the Human Genome Project. In better understanding what sequences correspond to which diseases, it is hoped that physiological and behavioral diseases will someday be prevented by prenatal detection and gene therapy. However, concern also exists for the proper use of genetic screening and therapy. New eugenics raises the issue

that when science advances itself to the level where gene sequences can be rearranged, will there exist immoral use of the techniques in order to create the perfect human? Will gene therapy be practiced in converting all less-than-perfect genes to the utmost genetic potential? At present, gene therapy is only in its beginning stages. Prenatal diagnosis is still under development, yet it is growing quickly, as will be discussed further on. Several contributing factors influence or will influence the availability and the use of these developing diagnostic techniques, such as health insurance agencies' policies and the availability of trained clinicians and doctors. The Human Genome Project is furthering scientific developments in gene mapping as well as creating several ethical conflicts among patients, physicians, and society. In order to impact society in a positive manner, HGP technology and its diagnostic information should be established in combination with effective treatment, so as to emphasize benefits to the patient. The HGP prompts two additional issues involving justice and equality in utilizing the concerned health care, as well as the distinction between gene therapy and gene enhancement, as will be discussed in a later section. (The Code of Codes, 281-299).

Genetic Counselors

A visit to a genetic counselor is a goal oriented visit to educate the counselee in the ways in which their genetics will influences their lives. The specific knowledge to be given out involves the technologies available to help the counselee, the ways in which their genetics will and won't affect their lives, and the uncertainties of their unique situation. The information that each counselee needs to know is very specific and the challenge comes from not giving out extraneous and confusing information. The genetic

counselor must do all of this while not imparting any morals or opinions on the counselee (Capron, 85).

Genetic counseling typically consists of only one or two sessions with a follow up letter summarizing what was talked about. The limited amount of time prevents an extensive amount of learning. This necessitates efficiency and accuracy in the counseling process. It also prevents the counselor from having time to give emotional support or advice on morals. This burden is placed upon to the patient or those close to the patient such as family or close friends. The overall counseling approach is then a hands-off attempt to educate in order to allow for better-informed decisions.

The term genetic counseling can be used to make reference to a large number of things. Genetic counseling in this paper is meant to refer to the advice and knowledge that a trained professional gives out to an adult in reference to the adults genetic condition or that of their unborn child. Genetic counseling covers the faults and successes of modern technology (Rothman, 40).

Genetic counselors like all other professionals make mistakes. These mistakes can lead to a less than optimal counseling outcome. There are also situations in which the limited counseling time prevents the counselor from achieving their desired results. Some of the situations that can cause poor outcomes are the patient's previous knowledge of genetic screening, the patient's perception of the risks of genetic screening after they have been given the facts, and the complex ways in which patients make decision.

One thing that can get in the way of good genetic counseling is a counselor with poor communication skills. Someone who cannot communicate effectively would keep the patient from having a clear idea of what is going to happen to them. In addition to not

receiving all the knowledge, a genetic counselor with poor communication skills can give out misinformation, or give the wrong impression or ideas.

Doctors who provide genetic counseling fall into two different types of categories. In more expensive clinics physicians with lots of technical knowledge provide counseling on the facts and risks associated with various operations. For the most part they are very well informed on the facts and present them to the patients. These doctors very rarely provide any sort of emotional support or advice in order to distance themselves from the decision making progress as much as possible. The other type of counseling that is provided is from those who are much less technically qualified to do so. Because of the increasing need for genetic counseling without a proportional increase in those who are fully qualified to do, social workers, nurses, psychotherapists and sometimes those with graduate training in genetics are having genetic counseling forced upon them. While these professionals often do the best job possible, at times they are unable to do justice to the complexity of genetics and the diseases. These workers however are more likely to help their patients deal with their emotional trials because with their lack of training, the best support they sometimes can give is emotional. While this is sometimes just what the patient needs, other times it is lacking in factual information that can influence the patients to go against their original intentions or morals. Surrogate counselors who do not have training in how to avoid changing their patient's disorders will continue to do so (Bosk, 25).

Ignoring the problems that can occur from the genetic counselor, the background of each patient needs to be taken into account. Race, sex, religion and economic class divide potential counselees into different groups. While members of these different

groups may each be a unique experience for the counselor, there are general problems that each group faces collectively that others do not. Two particularly important social groups to look at are those who are female, and those of African American descent.

Female clients, and more specifically female clients who come alone, or with a husband who does not really care enough to contribute to his wife's decision are a specific group that needs to be given special consideration. The burden of the decisions in the process of genetic counseling lies solely on the shoulders of the mother (Alper, 106). This significance of this is that they are more susceptible to the emotional effects of their decisions and need more emotional support than women with supportive husbands who are actively involved in the process. In addition these women because of the magnitude of their decision are more susceptible to outside influences from doctors, friends, family and society (Alper, 104) (Rothman 53-55). Careful care must be taken in order to help guide them to a decision that is of their own choosing that they will be satisfied with.

Members of different racial groups have preexisting biases towards genetics and those that might give them counseling. Members of different races, and in this case specifically African Americans have an innate mistrust of genetic counseling as a whole because of its control mostly by white people. This distrust stems from incidents such as the Tuskegee Syphilis study and an overall mood that could be created from a fear of being tested upon like a guinea pig (Alper 162-164). Many African Americans also have a fear and distrust of the science and the power behind the science that might make their lives even more difficult. African Americans in addition have an increased fear of being identified and discriminated against because of a genetic risk. In turn with being more afraid of the technology they are less likely to take any advice seriously which can end up

lowering their perception of their genetic risks which can then cause them to make choices adversely affecting them(Alper 166-168). Two examples of this are how African Americans could more regularly use amniocentesis and selective abortion if they so chose to, and how they could consider genetic risk when choosing a mate.

Teaching methods are also important when it comes to genetic counseling. Two techniques that can be used to help clients get a better grasp on the complexities of genetics and hereditary diseases are analogies and pictures. One example of an analogy that is commonly used is the two by two grid commonly used in high school biology to represent dominant and recessive genes. Often this can be extended slightly past its points of accuracy to simplify a much more complex genetic issue aiding the understanding of the clients. A common visual tool used in helping people to understand how genetic diseases are passed down from generation to generation is a family tree with special notation at each member to denote if they have the disease or not, and if they are a probably carrier (Marteau, 256) (Bosk, 31). Seeing who the carriers are in the family's tree often demystifies the pattern of disease.

Even the best genetic counselor in the world will not always be able to get across the message that they hope to. The process of geneticing screen evokes a different emotional response in everyone that goes through it. A calm, very factual approach that may be perfect for some patients might only amplify the fears of others. Because a genetic counselor cannot perfectly get to know their patient, their ability to customize their educational approach will be limited, and all patients will not receive the optimal amount of information. The degree to which the patient's emotions will effect their perception of the information presented can range from seeing things in a slightly

different light to missing the point completely, to being offended and turned away from the entire idea.

All of the advice that a genetic counselor can give a patient is under the constraints of technology. The specific technologies that have been developed limit what can be done to test for a genetic disease. The technology also limits the accuracy and the risks associated with each of the choices that the patient faces. As technology develops so will the abilities of doctors to predict, prevent and cure genetic diseases.

Every potential client for genetic counseling is coming from a different place with different opinions and a different point of view. This makes the job of a genetic counselor particularly difficult because each and every new client is a unique challenge to counsel and educate.

Current Technology

The public right now is probably most familiar with mass population screenings or surveillance, as well as surveillance of individuals for a particular disease. This familiarity could also vary depending on age groups and the types of epidemiological studies done in different areas at different times. What the public might not be as familiar with as they should be is prenatal screening. These screenings can be interpreted to mean mass population screenings (surveillance), but rather they are individual medical assessments of a woman's pregnancy. In addition, screening is usually a short-term cross-sectional activity while surveillance tends to be a long-term vigil on the health of a person or population (Scriver 1979) (Sadick, page 19).

To begin with, a distinction between prenatal screening and prenatal testing and diagnosis should be understood. Prenatal screening is the programmatic search for fetal abnormalities such as congenital malformations, chromosomal disorders, neural tube defects and genetic conditions among the asymptomatic population of pregnant women. Through prenatal screening, women in high-risk groups are identified for additional testing (Ettorre, page 24). On the other hand, prenatal diagnosis is undertaken to determine whether a pregnant woman with a fetus considered to be at risk of being abnormal by prenatal screening process does, in fact, carry a fetus with the disorder in question (Ettorre, page 27). In this section, the focus will be on the techniques used for the prenatal screening as well as for the further testing, which both comprise of fetal analysis. The technologies used can be either non-DNA-based (i. e. unrelated to genetics, such as ultrasound scanning) or DNA-based (i. e. related to genetics and blood or serum collection, such as chorionic villus screening, maternal serum screening or amniocentesis). Both non-DNA and DNA-based practices are used in conjunction with each other in the search for fetal abnormalities.

Prenatal screening covers many diseases, as evidenced by the variation of technologies in which they can be DNA-based or non-DNA-based. It will be apparent that, in this study, the focus will progressively be narrowed down to genetic screening. As examples are given in this section, most of them will be technologies that mainly affect women. Although pregnant women are those who are mentioned and most affected in most of the techniques mentioned, it can be noted that, especially during the prenatal screening process, both pregnant women and those who are not can be indispensably involved. The involvement of women who are not pregnant, although not mentioned as

much in this study, has been enhanced by the paradigm shift from predominantly curative health care to a health care provision that emphasizes preventive measures.

This is not to give an impression that only women are considered when using prenatal screening technology. They are the most intricately related to the prenatal processes that are undertaken and that will be given as examples. Despite all this, men are also important in this process and are considered in some processes. An example is in determining the heterogeneity of a mother and deeming her an obligate carrier for Duchene Muscular Dystrophy. This determination can only be certain if brothers and sons of a particular woman both express symptoms of the disease. This can be kept in mind as the apparent, predominant focus on women is realized in this section. This is so because the more detailed aspects of the technologies will be described in this section. Some techniques used during prenatal screening are described as follows:

Diagnostic ultrasound (not inherently a genetic test):

Diagnostic ultrasound is often used routinely in pregnancy as an all-purpose guide to fetal development. It is a procedure that forms an image of the fetus (i. e. fetal imaging) by using sound waves. A video image called a sonogram is displayed on a monitor as the sound waves can be converted into an image. Physical features of the fetus can be seen and often a photo can be produced from the monitor image for prospective mothers or parents. It is common belief that ultrasound hastens maternal bonding (Green 1990a). Ultrasound scans can help to diagnose a number of birth defects including hydrocephalus, limb or organ deformities, some heart defects (Buskens et al. 1995), neural tube defects such as spina bifida and renal tract anomalies (Malone 1996). Skupski

et. al (1994) argue that while ultrasonagraphy never will be perfect in the detection of anomalies, it is viewed as a cost effective procedure (Ettorre, page 26).

Maternal Serum Testing:

The most usual form of maternal serum screening is the triple test or triple screen. The triple test is a blood test that examines the level of alpha-fetoprotein or AFP (a protein produced by the fetus) and two pregnancy hormones estriol and human chorionic gonadotrophin (HCG). This test does not give a definite diagnosis of abnormalities and further tests would need to be done to confirm any indications of fetal disease. An example of when this kind of test would be done is when trying to detect Down's syndrome. In this case, particular markers are used to indicate the most probable presence or absence of a disease. High levels of alpha-fetoprotein may indicate the presence of a neural tube defect, such as spina bifida or anencephaly. Low levels of alpha-fetoprotein and estriol combined with high levels of HCG may indicate Down's syndrome (Reid 1991). Abnormal levels of these proteins or hormones are not diagnostic themselves, but when abnormal levels of these proteins are found, the test merely indicates a potential risk and further tests such as ultrasound and amniocentesis are indicated (Ettorre, page 25).

A pregnant woman is given a risk calculation based on analysis of the serum in her blood. For example, she may have a 1 in 250 risk of having a Down's syndrome baby. Usually, prenatal services have a 'fixed' cut off figure (such as 1 in 250) at which point physicians will prescribe prenatal diagnosis if risk calculations fall below the cut-off point. One expert, an obstetrician, implied that good screenings meant that you would

successfully detect Down's syndrome with the further test of amniocentesis. He emphasized that serum screenings were not going to give pregnant women a definite diagnosis of abnormalities (Ettorre, page 25).

Prenatal screening using molecular genetic tests:

Pregnant women can also be screened for recessive conditions such as haemoglobinopathies (sickle cell disorders, beta thalassaemia major, etc.) or Tay-Sachs disease that involves molecular genetic tests. Analysis from blood samples, CVS or amniocentesis is used where a disease causing mutation has been identified in a family. Screenings do have limitations and can only be accurate to a certain degree. With the analysis of certain levels of biochemicals being a common technique during screening, as shown above, the level distribution is continuous and although there are extremes, there are people who will have levels of these biochemicals that are in-between or around cut-off levels for either a positive or negative result. There are situations where it is pretty much certain that a person is a carrier, for example judging from the level of a particular biochemical which can be deemed extreme enough. On the other extreme, there would be people who have levels that are within the 'safe' range and it can pretty much be determined that they are not carriers for that particular disease. It would be nice if these distinctions were as discrete as that. But there are people who may be in-between and it can be difficult to counsel these people with regards to the potentials that they relate to. They potentially are carriers, and if they are, there is a potential that their children may have a particular genetic disease.

The main reason for this continuity is that most things in nature are continuous rather than discrete. An example that relates directly to genetic screening that has been studied is that of the phenomenon of lyonization. Lyonization is the random inactivation of one X chromosome in each cell of the fetus, with the possible exception of the ovarian cells. A particular example to keep in mind is how lyonization affects carrier detection for Duchene Muscular Dystrophy. The level of creatine phosphokinase (CPK) in serum is used to determine the carrier state in females. As with most carrier tests for X-linked disorders, it is not a very exact one, since many carriers test within normal limits. This is due to lionization (after Mary Lyon, who first described it in 1961) (Sadick, page 52). Because of lyonization , on average, females have one half of their cells with their maternal X chromosome active and one half with their paternal X chromosome being the functioning one. If the maternal X chromosome contains a gene for Duchene Muscular Dystrophy, about half of the muscle cells will have the membrane defect which leads to elevated CPK levels in the blood serum. However, since the number of cells that will develop to muscles are comparatively few at the late blastocyst stage, it is possible that by chance a significantly larger proportion than 50% of the activated X chromosomes in these cells will be paternal (which wouldn't contain the gene for Duchene Muscular Dystrophy). In those cases, the elevation of CPK may be very slight or, in fact, CPK levels may be normal. Regardless of this X chromosome distribution, the chance of a carrier transmitting the gene to her offspring is still 50%. The problem with lyonization is present in most X-linked diseases in which the product measured is not the primary gene product (Lubs et al. 1979) (Sadick, page 52).

With aspects like this to take into consideration, some definitions to describe the efficiency of a genetic screening method are prescribed. The efficiency of a genetic screening method is said to be related to its specificity and its sensitivity. Specificity is defined as the ability to exclude from classification as heterozygotes, those who have the normal homozygous genotype. In the screening process, the non-carrier may yield either a normal test result (with frequency a) or a positive result (with frequency b), the latter being a false positive test result. The specificity of screening is then $\frac{a}{a+b}$. Sensitivity is defined as the ability of the test to identify those who possess the mutation. The latter yield either a positive test (with frequency d) or a normal (false negative) test result (with frequency c). The sensitivity of screening is then $\frac{d}{c+d}$. Binary test systems can yield perfect specificity and sensitivity but quantitative testing may not (Scriver 1979) (Sadick, page 23).

It may be noted from the above discussion that for prenatal screening, the tests tend to be done on the pregnant women themselves for a preliminary sampling. But as will be discussed in the following paragraphs, tests for prenatal diagnosis are done more directly on the fetus as opposed to the pregnant mother. Prenatal diagnosis is the identification of an abnormal condition in the fetus (Beekhuis 1993). With the current technology, there are several methods used for extracting genetic samples from fetuses during the prenatal diagnosis. The methods of prenatal diagnosis include second trimester ultrasound screening, amniocentesis, chorionic villus sampling (CVS), placentocentesis and fetoscopy in conjunction with fetal blood sampling and fetal skin biopsy. In biomedical terms, the primary aim of prenatal diagnosis is 'to provide an accurate

diagnosis that will allow the widest possible range of informed choice to those at increased risk of having children with genetic disorders, within the boundaries established by society' (Advisory Committee on Genetic Testing 2000) (Ettorre, page 27). Descriptions of some techniques used are as follows:

Second trimester ultrasound:

Starting with the broader picture, direct examination of the fetus can be carried out by second trimester ultrasound, which can discover developmental lesions and major congenital malformations. High-resolution equipment can accurately assess fetal development and anatomy at 18– 20 weeks of gestation (Ettorre, page 27).

Amniocentesis:

Amniocentesis may be performed in the third or second trimester. It should be noted, however, that when particularly focusing on prenatal testing, amniocentesis is mostly performed during the late first to early second trimester to avoid having too late an abortion when that is an option. Amniotic fluid is drawn from the amniotic sac around the fetus with a long needle through the pregnant woman's stomach. The fluid contains fetal cells and that can be used to obtain genetic knowledge about the fetus. The culture obtained through amniocentesis can be used to detect Down's syndrome, blood type, metabolic problems (i. e. Tay-Sachs disease) and neural problems. The results of this test are available within seven to ten days (Ettorre, page 28).

Chorionic villus sampling:

Chorionic villus sampling (CVS) is another way to look at fetal chromosomes and it is performed at the tenth to twelfth week of pregnancy. In this procedure, physicians remove a tiny sample of chorionic tissue with a small tube that is inserted into the vagina through the cervix to collect (with suction) a tiny sample at the edge of the placenta. The sample can also be taken in a similar way to amniocentesis. Karotyping, the arrangement of chromosome pictures in a standardized way, is prepared from the tissue sample. Results are available in seven days. Unlike amniocentesis, CVS is unable to detect neural tube defects and the risk of miscarriage is slightly higher than amniocentesis.

For both amniocentesis and CVS, there is a risk for miscarriage and fetal loss rate at 0.5–1.0 per cent and 1– 3 per cent respectively (Advisory Committee on Genetic Testing 2000). In biomedical terms, it has been argued that invasive genetic testing is the gold standard in fetal diagnosis (Kuller and Laifer 1995) (Ettorre, page 28).

In a symposium published in 1979, it was said that despite the relative safety of amniocentesis and the high accuracy rate of over 99%, difficulties arise occasionally when no amniotic fluid is obtained or if suboptimal cell growth does not allow proper interpretation (Pueschel 1979) (Sadick, page 66). There have been significant advancements since then and it seems safe to presume that this does not happen that often today. However, we are still not immune from such difficulties, and the following is a description of what would be done as time went on. Although this might not happen that often now, this is probably the procedure that would happen today. Because of a dry tap in 2-5% of cases a second amniocentesis will have to be performed and in 5-10% of cases cells from the amniotic fluid fail to grow in the laboratory culture media again

necessitating a second and sometimes even a third amniocentesis (Pueschel 1979) (Sadick, page 66).

Other complications are bloody amniotic fluid samples, contamination with skin bacteria, and technical or clerical errors of reporting the results of the chromosomal analysis (Pueschel 1979) (Sadick, page 66)These may be rare nowadays, but they are complications nonetheless. In addition difficulties arise in the interpretation of cytogenetic observations from amniotic fluid cell cultures in instances of mosaicism, polyploidy, spontaneous translocations, and subtle chromosomal abnormalities (Pueschel 1979) (Sadick, page 66).

Fetoscopy, fetal blood sampling, and skin biopsy:

After several years of using larger endoscopes with attendant high risks to the pregnancy, perinatologists turned to a small diameter fetoscope which can be inserted into the uterus through the abdominal wall with minimal trauma. A major problem of the small instrument, though, is a limited field of view. The instrument has a diameter of 2.0 mm or less and, when inserted into the amniotic cavity, is housed in a cannula of 2.0-3.0mm. The cannula is large enough to allow passage of a blood sampling needle or biopsy forceps alongside the lens. Light is taken into the uterus on fibers and the image is returned either by a fiber optic lens or a solid self-focusing lens. The angle of visualization is about 70^{o} and objects are in focus through a depth of 2cm. These parameters permit 2-4cm^{2} of surface area to be in view at one time (Mahoney 1979) (Sadick, pages 89-91).

Prior to insertion of the fetoscope, careful ultrasound information is obtained using either gray scale or real time sonography. This information enables the fetoscopist to select an entry site which will be as safe as possible for the fetus and which will place the desired parts of the fetus or placenta into view. Real time ultrasound is also used during the procedure to help locate specific fetal parts. Local anesthesia is injected at the entry site on the mother's abdomen and is usually the only drug given during the procedure. If fetal sedation is desired, diazepam or meperidine can also be given to the mother.

Visualization of the fetus is optimum between 15 and 18 weeks gestation. At this stage the fetus is relatively small compared to the surrounding amniotic fluid volume and the fluid is very clear. Below is an extract of a report by Mahoney and Hobbins (Mahoney 1979) (Sadick, page 91).

"We and others have been able to identify and study specific parts of the fetus and several fetal behaviors. Localized areas have included close view of digits, joints, external genitalia, and umbilical cord. Details of the skin surface including hair, nails, and pores can be appreciated. At the head and face one can see ears, closed eyes with fused eyelids, the nose and mouth, and even the palate, but the parts can only be seen singly and a panoramic view of the face is not possible. Fetal movements such as thumb sucking, swallowing, grasping, and defecation have been observed through the fetoscope in the 16 to 18 week fetus." (Mahoney 1979) (Sadick, page 91)

Total visualization of the fetus is rarely attempted, but instead focus is on parts of the fetus which may yield diagnostic information regarding a specific anatomic defect.

With this limited purpose, fetoscopy can successfully examine the fetus about 90% of the time (Mahoney 1979) (Sadick, page 91).

Below are examples of the diseases for which limbs, digits and trunk have been examined.

Limbs or digits examined because of risk for:

- Arthrogryposis multiplex congenital
- Laurence-Moon-Biedl-Bardot syndrome
- Absent limb
- Meckel syndrome
- Hold-Oram syndrome
- Split hand syndrome
- Ellis-van Creveld syndrome
- Polydactyly (with other major defects) Trunk examined because of risk for:
- Spina bifida
- Exomphalos (Mahoney 1979) (Sadick, page 92)

Below is a commentary on the above examinations by Mahoney and Hobbins. "In the majority of these examinations normal fetal parts have been seen and normal infants were born at the end of gestation. The first positive diagnosis using fetoscopy was in a pregnancy at risk of the Ellis-van Creveld syndrome. This autosomal recessive disorder consists of dwarfism, congenital heart disease, and polydactyly. The presence of an extra digit established the diagnosis." (Mahoney 1979) (Sadick, page 92)

It can be seen that sonography is used in conjunction with fetoscopy in order to have a safer and more efficient process. But for diagnosis that is focused more on genetic diseases, fetal blood sampling may be done. When this is done, it is done with the use of a fetoscope as well. The fetal diagnostician is probably more interested in the placenta with its circulation of fetal blood, rather than just images of the fetus from the fetoscope. Small veins and arteries, which branch from the umbilical vessels, course along the chorionic plate at the inside surface of the placenta. These vessels with connecting capillaries form a closed circulation of fetal blood. In the placental substance, surrounding the fetal capillaries are pools of maternal blood. It is from the fetal circulation in the placenta that successful attempts at sampling fetal blood have been made rather than from the body of the fetus. Two methods for obtaining fetal blood have been developed. One, as mentioned above, utilizes fetoscopy. But there is another which consists of ultrasonically directed placental aspiration.

For the first, fetoscopy affords direct visualization of the fetal vessels on the chorionic plate and vessel puncture can be accomplished in a controlled manner. A 25-27 gauge sampling needle is inserted into the cannula alongside the lens of the fetoscope. When a vessel is located the sampling needle is advanced to puncture it under direct vision. Most often, blood cannot be aspirated directly from the lumen of the vessel because of the small bore and long length of the sampling needle. Instead, the needle is withdrawn from the vessel and blood is aspirated with a syringe as it spurts out into the surrounding amniotic fluid. Samples of 0.05-0.15ml are collected and bleeding stops after several seconds. The syringe and sampling needle must contain anticoagulant. Heparin and citrate have been used successfully. Although blood sampling via fetoscopy can be

difficult, especially when the placenta is anterior, it can be satisfactorily accomplished with experience. Samples obtained by an experienced operator using this method are often uncontaminated by maternal blood. If the gestational age of the fetus is 18-19 weeks, the most common time for attempting fetoscopic sampling, fetal blood volume depletion is expected to be less than 3% (Mahoney 1979) (Sadick, page 93). The above just shows how even with optimal conditions, with regards to the experience of the operator as well as gestational age, there is still room for error due to contamination with the maternal blood.

The second, placental aspiration is performed with a 19-21 gauge spinal needle. The position of the placenta and the depth of the chorionic plate are estimated from the sonogram and the needle is advanced to the interface of the chorionic plate in the amniotic cavity. Blood is then aspirated and immediately checked by a Coulter electronic cell sizer for the presence of fetal cells. Fetal red blood cells have a mean volume about one and a half times larger than adult red cells at mid-gestation. The cell sizer would detect a 5% contamination of fetal blood by maternal blood or maternal blood by fetal blood. If a satisfactory sample was not obtained, the needle is redirected and another sample is aspirated. Contamination with maternal blood, often in large amounts, is common with this method, although pure samples of fetal blood occasionally result. A satisfactory blood sample for fetal diagnosis of the hemoglobinopathies will be obtained on first attempt about 90% of the time. A second attempt will succeed in most of the initial failures so that diagnostic information is usually available to the requesting family. A good plasma sample is more difficult to obtain than a good red cell sample

because of dilution with amniotic fluid. Experience with obtaining plasma is still small, but presumably the failure rate will be higher (Mahoney 1979) (Sadick, page 95).

Most of the information and examples given above came from a symposium from 1979, 'Genetic Diseases and Developmental Disabilities; Aspects of Detection and Prevention'. Some technologies described were at the earlier stages of being researched and refined. It may also be evident that the more experienced and skilled the hands that perform the particular procedures described above are, the less the apparent risk on the pregnancy, the woman's health, and the fetus' life. And with such a marked increase in the frequency of performance of these procedures in recent years, the hands that perform them seem to be generally more familiar with the procedures. These are the hands of obstetricians, fetoscopists, physicians and those who perform the laboratory testing that is required. There may have been some major advancements from the given description of certain techniques, but one thing that still remains the same is that the processes, although they can be very accurate, they have limitations, risks and uncertainties. Some of this is confirmed in the other resource used (2002), 'Reproductive Genetics, Gender, and the Body'. That makes a situation where particular techniques are best suited for detecting certain diseases. Furthermore, combinations of techniques can be used to improve the accuracy as well as reduce the uncertainties related to the diagnosis.

Future Technology

There currently exist many ways to screen and test a mother and her unborn child. The future holds innumerable possibilities for advancing today's technologies and techniques. Many of these possibilities are quickly becoming a reality and opening doors

within medicine and technology. What was once unknown is quickly becoming well known and new ideas are constantly being explored. Many of the current studies in progress for improving upon the care and treatment of the unborn child revolve around the continual advancements in the Human Genome Project. Mapping the chromosome will allow further understanding of the genetic composition of specific diseases as well as treating the mutated genes responsible for the illness. Ultimately, these treatments would allow for the chromosome to be repaired in the fetus so that the child would be unaffected after birth. This technique is not a long way away. Additionally, as technology advances more DNA tests will become available and the challenges faced will be related to education, such as ethical, legal, and social dilemmas that will be encountered.

As DNA testing becomes more sophisticated, the genetic counseling by professionals in the field will also become more complex. The availability of the developing knowledge is primarily available via computer resources, thus, in order to avoid the perceived negative aspects in DNA testing, health professionals will need to implement a broad based educational strategy. At present, counseling must accompany the DNA tests in order to provide the patient with knowledge. The difficulty encountered comes in answering the questions: Who does the counseling? What intensity of counseling is required? How should the required level of counseling be provided? In turn, a team of professionals would need to be collaborated in order to optimize the patient's knowledge of and care in prenatal testing. Conflicts will arise and will need to be addressed regarding DNA testing for clinical versus personal value. In addition, issues regarding testing for clinically relevant versus trivial diseases will need to be explored.

As possible therapeutic treatments improve, the use of DNA tests will need to be restricted to those with the greatest medical good.

The success of the implementation of techniques such as DNA testing is dependent upon one's geography and income. Many of the advanced techniques in prenatal testing are costly and not readily available. There are few offices that practice such techniques and are limited to areas of great technological development. Thus, some third world countries would not have the accessibility to provide certain advanced testing techniques. Since insurance does not cover these advanced methods in screening, typically only those families who are well off can afford such tests, and those from poverty will not benefit from these advancements. These limitations bring about other concerns as well. DNA testing may introduce a field that others will claim to be discriminative. This issue needs to be carefully addressed in order to improve the ignorance through education. Legislative matters may also need to be addressed in order to acquire a common understanding.

What tomorrow holds is fast approaching. Several advances in prenatal screening are underway. One of the concerns being examined is that of earlier detection of risks. Through advancements in serum screening, ultrasound, and noninvasive techniques it is hoped that the time it takes to identify pregnancies at risk will be significantly reduced. Techniques incorporating nuchal translucency, or swelling of the visible nachal area in ultrasounds, maternal age, and integrating beta-hCG (human chorionic gonadotropin) and PAPP-A (pregnancy associated plasma protein-A) into serum screening are aimed at the early detection of abnormal gestations. Inhibin, PAPP-A, and urinary hCG, in combination with maternal age, are being developed for the purpose of utilizing them as

maternal serum markers, which are independent predictors of aneuploidy, in order to increase sensitivity rates. These markers, used alone or in combination, would be capable of increasing the sensitivity rate of detection by approximately 5%.

There exist noninvasive methods in prenatal diagnosis; however, improvements are underway to make this technique more readily available. Noninvasive techniques make use of the isolated fetal cells and tissue sample testing in order to alleviate the risks posed on the fetus that exist in other methods of prenatal testing. This concept is not often used due to the limits of availability of testing centers and trained technicians. Further developments within this area are presently undergoing clinical tests. Another area under development is presymptomatic testing. This particular type of testing brings about social and ethical issues that address the conflicts of testing for untreatable diseases and the use of this information in the decision making process.

DNA testing has grown since its conception in the late 1970s. It is currently used in several areas, such as: predicting the development of genetic disorders, screening populations, confirming clinical diagnosis, prenatal testing, and improving medical treatment. This area continues to grow as technology advances and societal, as well as ethical, concerns are faced by the community with the help of professionals. Screening tests are currently available, however they are also underway for improving upon them. These screenings identify genetic mutations in order to recognize carriers who could pass the mutation, which could be expressed as a genetic disorder, on to their offspring.

Beyond the tests that involve screening and testing the mother for information regarding the health of her unborn child, there exist tests performed on the fetus itself.

Fetal tissue research is currently taking place with the aim of treating genetic conditions in the future.

Decreasing the time between the test and the diagnosis of genetic mutations in an unborn child is a continuous goal in scientific research. Enabling tests to occur earlier in pregnancy, will allow for a greater scale of women to participate, or at least have the opportunity to participate, in prenatal genetic testing. Current tests involving invasive methods, such as amniocentesis and chorionic villus sampling, are not commonly performed on women under 35 nor those who are not tagged as high risk cases. This selective screening is due to the associated risk of miscarriage and the cost of testing. This allows for undiagnosed abnormalities in babies born to younger mothers. The improvements are intended to develop less costly and more efficient procedures for diagnosis. The non-invasive method involves obtaining foetal cells, isolating them from the PAP smear, using PCR techniques to classify them genetically, known as DNA fingerprinting, and then screening them for defects using SNP. This is a simple procedure that can be performed by general practitioners. The samples are then sent to labs for analysis. This test is designed not only in hopes to be performed earlier in the pregnancy but also to decrease the wait period for the results from 2-3 weeks to the same day.

Gene manipulation is developing to improve screening for fetal disorders and prevent future generations from inheriting diseases. Gene therapy involves injecting a healthy gene into organ cells in order to intervene and replace the abnormal cells. This allows for the fetus to be rid of abnormalities and lowers the risk of future generations of offspring inheriting diseases. Moral issues are of concern in this method of treatment due to interference with natural evolution and heredity.

A unique advancement in scientific technology is known as boutique ultrasounding. Boutique centers are going up all across America. These commercial facilities are designed to attract curious expecting mothers. The ultrasounds produce detailed pictures of the parents' unborn child(ren). Using high-density 3-D technology, the baby's features are accentuated as compared to conventional ultrasounds. The features imaged range from physical traits, such as facial expressions, to behavioral traits, such as thumb-sucking. These boutiques are utilizing conventional methods of medicine for personal benefit. They allow people to see physical, and some behavioral, features of their unborn children without the hassle of having to go to the doctor. They allow customers to print out a sharper imaged picture of what their baby looks like. In return, they are bringing about concerns due to their use of such technologies and the motivations that they may promote.

With the above mentioned developing technologies, as well as those that have not yet commenced, the future of medicinal technology and prenatal care and diagnosis holds innumerable and even unimaginable possibilities that one day soon will become a reality. Along with this reality, though, will also come ethical questions and social concerns that the world of science is not yet prepared to face.

Ethical And Philosophical Issues

It is all well and good to speculate on the benefits of future technology. However, there will always be those who shout out cautionary words and cryptic phrases to remind people that not all technology improves people's lifestyles. These sorts of things usually begin with "when I was your age we didn't have..." and proceed from there. There is merit in the idea, however, and any great change in technology should be considered for its possible impact on our society. Far from cries of doom and gloom, these are serious considerations based on a long history of human societal evolution both dependently and independently of technology. Advances like automation, interchangeable parts and the assembly line, the telephone, the television, and the computer have not just changed the way that people do business and contact each other but have also affected personal lifestyles and burrowed deeply into society to forever change it. The basic idea is that technology should not exist for technology's sake; it should exist towards an end, not be an end in and of itself. Although technology itself is incapable of being good or bad, it is possible that a certain development could alter society for the worse or help to improve it based on the way people utilize that technology.

The trend thus far has been towards mechanization, towards automation, creating more tools that replace functions once performed by humans. This has generally lead to a decrease in physical activity and labor, but an increase in lifespan and education. Problems like obesity and suicide have increased while problems like starvation have decreased in the developed world. Communications have brought poeople closer together but have also served to increase the fast pace at which people, especially people in the

United States, are forced to work and live their lives. The most difficult question would be does the increased technology make people happier as a whole? It's all well and good to ask this question and expect the answer to solve the whole problem. However the question itself is inherently flawed. How can we ask what benefits a society if we don't first establish a solid grounds for testing the happiness of society? Is lesure time available a better judgement of happiness than the suicide rate and number of people who go to counseling? Is counseling just part of the modern lifestyle and more a cultural phenomena than a true indication of our happiness as a society? The answer, although not at all satisfying, is that it may be impossible to really know if people are happier as a society, and it would be out of the question to quantify such a measure if we could find it. Perhaps the best way to approach the issue of societal happiness is to look at the individuals in the society and impacts on them, and then compare to what most people consider our "social problems". If a change increases these problems and does not make people happier then it is a bad change. If we look at our developing technology and see the good and the bad from them as it applies to the personal as well as the social, we can still gleam at least a good guess as to what would benefit society.

Do We Benefit From This as a Society?

The development of navigational tools and ships capable of crossing the sea between North America and Europe allowed people to explore new and excitiing parts of the world. Now faster forms of communication allow the easy exchange of information across this same expanse and beyond. Space flight may soon allow us to colonize the moon or even leave our solar system. For each benefit to be drawn from these

technologies there is the the same potential to increase the capacity of some people to gain an edge of others. The television enabled people to "see" other countries or even space without moving and to see presidential candidates on the campaign. Television though is now believed by many to be a catylist in the growing problems of inactivity, obesity, and the gradual social deadening to violence and sex. Cellular phones connect people and save lives in emergencies, but are now believed to be the cause of many automobile accidents. In the same way, prenatal screening is nothing more than another tool, which could present postive benefits or create new problems. There always exists the potential to be misused. The benefits and risks must be weighed before the technology is allowed to run rampant. Essentially the possibly impacts on society and their relative likelyhoods should be considered in order to fairly assess the technology.

The Good

Since children and family are such personal issues, the psychological benefits of knowing a future child's condition is a good place to start analysis. A family that has time to plan and prepare may be able to come to grips with the situation, allowing the child to grow up and develop in an environment more like that of any other child. For genetic defects that only manifest themselves given certain environmental conditions, it would be possible to minimize the chance of these events or situations occuring which would again reduce the chance of loosing a child to genetic disease. As it stands with current technology this is the most common use for prenatal screening. In most cases a screen will turn up negative. Even in those cases that show an unusual chance of genetic defect there is normally no guarentee that it will manifest itself in any given child. This means

that very few prenatal tests will end in an early termination, as for most people this may not be the best solution to having a child with genetic defect. Most people are willing to care for and love an abnormal child so the screen is not usually used to find and weed out children with genetic defects. (Carrie Haverty) Instead it is used as an early identification system so that parents will be prepared for the special needs of their new child. Part of the job of the genetic counselor lies here in helping to provide information the parents of a defective child will need if they choose to keep it, which is normally the case. There is also the great question of knowing whether or not a future child will have a fatal genetic disorder. The potential for this and its impacts are discussed in a later section.

Since society is made up of people, the impacts of prenatal testing on society can be traced from the effects it will have on families and people. Simply extend the changes on the family to include nearly everyone (or a large percentage) to see the possible widespread effects of this advance. Although most couples who undergo prenatal screening either find no problem or choose to keep the child anyway (a positive flag does not prove that the genetic disease will manifest), as the technology becomes more accurate and available, more and more people will decide to abort based on genetic condition. The ability to choose between "normal" and "abnormal" children would reduce the population with genetic defects which would increase the number of productive members in society. This would also reduce health care costs and decrease the juvenile death rate. For many genetic conditions, such as diabetes, early medical attention as simple as a special diet or medication can be the difference between a normal lifestyle and a childhood of emergency room visits. For defects such as dwarfism or similar hormoneal deficiencies, early treatment is the only way to treat the child. Knowing that a

child has a deficiency versus just being "a little short for his age" can make all the difference for treatment. (Billings, Kohn, Cuevas) The opposit is true as well; if a child is short but prenatal screens reveal a very small chance of dwarfism it may help to lay parents fears to rest and prevent unneccesary treatment. This again will save money on health care for both the family and the insurance companies. Oftentimes treatments like horomones are very expensive especially when the treatment extends for several years can be a great financial strain on a family. The potential to avoid such costs would be a great boon to society.

As far as society is concerned, increased prenatal testing could lead to a number of changes. Being able to detect genetic disorders is only the first step, the second is deciding what to do with this information. If we are able to treat conditions early, it is possible that it will not be as detremental. This may decrease the number of people who suffer from genetic defects and will help to decrease the juvenile mortality rate. As mentioned before, this could increase the number of active productive members of society. On the other hand, if children with genetic defects are treated early and kept alive, we run the risk of actually increasing the number of people who are dependent on special care, medications, classes, and living arrangements. Socially prenatal testing could be a real Pandora's box. This will be discussed more below.

The Bad

Immediatly problems that could arrise from widespread prenatal testing are also apparent. Genetic disorders that are untreatable could cause families that are unable to cope with potential loss to have problems developing and living together. If a family is

told to expect to have a child for only a few years it could lead to a sad detatchment from that child in order to preemptively accept the loss, or it could lead to a sick child being showered with attention "while they're still around" leaving other children, spouses, jobs, and problems on the wayside. As our ability to test for and choose genetic traits expand to things like hair color, skin tone, eye color, intelligence, and physical perfection, children maybe feel more pressured to live up to their genetic standards. Parents who choose their child in detail may put more pressure on their children to continuously look and act in a certain manner desirable to them. This potential is technologically a ways off but worth considering before we reach that point.

Once again, the larger social issues should be of imporance to us as well as the impact on the individual family. Although our research here largely ignores abortion, in this consideration it is not possible. Choosing to abort children based on genetic defects will undoubtably impact our culture. The ancient Spartans were famous for only allowing the children of strong women to live, thus increasing the strength of their own people. This same principle can apply once again. If we eliminate all of the genetically "inferior" children what will society become? Will we convert to racing and competing for the best genes? How would movements such as Naziism have been affected if it could be proven that a given race is indeed genetically "superior"? Establishing such a convention could ruin competitive sports forever since it would all be a race to find the best possible parents to breed.

Fictional predictions and Ideas

Although it may seem to be trivial, it may be worthwhile to deduce possible future trends in reaction to technological advances from literary predictions. Especially during the so called "Golden Age" of science fiction, a great deal of these works were produced not only as fictional literature but also as serious commentary on human society. In addition, most of the most famous authors of science fiction from that era were well-educated scientists and psychologists. Books like *1984*, *Foundation*, and *Brave New World* were written to create a view into a possible future for mankind. Getting away from specific examples, trends in futuristic fiction can indicate culteral predictions or beliefs for the possible outcome when we move deeper into the 21st century. At the time when Orwell wrote *1984* there was growing concern for an overly powerful and controlling centralized government. Even to this day people read the book and shudder with the realization that it is still a future possiblilty. Modern issues like Homeland Security and the Patriot Act brings the dark images of Orwell to the surface reminding us to think carefully about what we do and the possible future ramifications of our actions. Even more pertinant to the idea of prenatal screening and selection is *Brave New World*. In this book the human race is grown in vats and those that are to become the ruling class are carefully screened for genetic purity. Those who are doomed to be members of a lower worker class are stunted during development with alcohol and lowered oxygen supply to make them smaller and mentally defective. This sounds far-fetched but what will our world come to if more perfect genetic screening and prenatal genetic alterations become possible? Wouldn't those who could afford it make sure that their children have the very best bodies and minds possible? Tall, athletic, attractive, smart, with strong

healthy organs? If people of the lower or middle classes could not afford such treatment wouldn't there indeed be a biological class structure?

As television and cinema grow to replace the written word, the tradition of bringing cultural fears and predictions into story form continues. The world of cinematic science fiction present the views of the future that many people hold as assumption. A modern example is the movie *Gattaca* which takes place in the very near future (50 years at the most). In this world the process of genetic identification is a standard everyday process. Drivers licence, birth certificate, and all other forms of identification are replaced by a simple blood test. Resumes are augmented by a complete genetic review covering likelihood of any number of defects, predicted lifespan, and many other bits of information before left to chance. In this rendition of the future, health insurance coveraage, job placement, physical qualifications, competency, and other factors are all calculated from percentages predicted from simple blood tests and genetic analysis. It is apparent that some people fear that with the dawn of genetic predictions with a great level of certainty, all other evidence as to a person's quality will fall by the wayside. We will then be reduced to the sum of our genetic markers. Truly this is a frightening concept.

As technology stands today these frightening images are a long way off if ever to be possible. However it does speak to public opinion that writers conjure up these ideas in the first place. If people truly believed that insurance companies and buisinesses would not latch onto these technologies then these fears would not be voiced. Even without the ability to accuratly predict lifespan and intelligence, being able to screen employees for a number of provable diseases would be beneficial to any company for liability reasons.

There is also the issue of genetic identification which goes deeply into our legal system and may not be very far off as well. Today genetic evidence in criminal cases is often unreliable and many judges are wary of using DNA tests as absoloutly conclusive. Genetic information is much the same way. As it is we cannot say conclusively who will have a manifestation of a genetic disorder. Because of this it is not yet used in a concrete manner. However as courts are setting precident for reliance on DNA evidence as being nearly flawless, so too may genetic markers be enough proof of genetic malformality. When this barrier is crossed, when we know enough to be more certain than uncertain, that is when the biggest changes will come about.

Relationships

Perhaps the most important consideration when prenatally screening is one that we have not yet had the time to see realized; the impacts on the parent/child relationship and on the child as a growing and develping individual in society. Plenty of research has been dedicated to understanding the stresses and pains of choosing whether or not to undergo some sort of prenatal testing for disease, and the subsequent choice to abort or not. Due to its relatively new nature, there has not been enough time to see the effects those choices have on children and the family in the long term. Naturally the choice to abort would result in their being no child. The considerations herein assume that the parents choose to keep their child whether they are informed of a chance for abnormality or not. The psychological health of the entire family should be considered by geneticists and genetic counselors alike as well as that of just the parents. Immediatly a number of concerns can spring to mind reguarding the effects of knowing; Is it better to expect the worst and possibly be proven wrong or to suspect nothing and be surprised? Does it benefit the life of the child or the family to know that the child will not live for long? How is a child's development affected by believing (truly or falsely) that he has a genetic disorder? These are all very important questions that must be asked before counseling is offered to expecting parents.

How the Child and Parent Interact with Each Other Differently

Once the decision is made, the parents are no longer the only component nor the primary concern for genetic counseling. It then becomes a matter of family and of child development. Naturally a disabled child has a great many needs that other children do not have and often require special attention as well as medical care. This can create a great strain on the family that can cause an extraordinary number of problems and hardships. Knowing beforehand of a genetic condition and preparing for this sort of lifestyle can be of great benefit to both the child and the family as a whole. Some disorders may take years to manifest and it would be beneficial to know from birth that a child has special needs. On the other hand prediction techniques are not perfect and could give a false positive. This would lead to a situation where a child is treated as if there is a condition even though it does not exist. It could substantially hamper the development of a child to be treated as a genetic "inferior" even if the intentions of the parents are only for the child's welfare. One example of this happening today is with Attention Defecit Disorder. Often a hyperactive child is diagnosed with this disorder and put on medication or sent to special classes that are not neccesary. This is not to say that ADD is not something that needs attention, but it is possible to accidentally treat those who do not require it which, in some cases, can make the child sick or deprive them of valuable experiences in life.

In order to analyze the impacts on the family, the most common cases will be looked at. That is, the most common scenarios that could result from genetic testing on a prenatal child. First we shall consider the situation where the predition turns out to be valid; the test results correctly predict a defect and the child is born. The family would have time to seek further counseling on the special care their child will need. Other

preperations such as medical advice, medical equipment, and altered lifestyles could all be prepared before the child's arrival. It may also lessen the emotional impact of realizing after perhaps years that a child has a genetic defect. The family would be prepared for the change in lifestyle which would, presumably, allow for an easy transition with perhaps no negative developments. Quite often it is the case that genetic disorders are not fatal if they are detected and treated. In the case of some genetic disorders, the only treatment required is a special diet. If this is already established before birth the child and the family should be able to continue and live normal and healthy lives. In this sort of case it is far better and easier to know in advance about the child's condition. The situation where the condition is fatal presents a different set of problems.

The loss of a child is a heavy blow for a family to endure in any situation. That the loss will be deep and longfelt is not in question. What is to be considered is whether in that situation it is better to have known beforehand. Certainly the family will have time to assimalate the notion that they don't have much time with a new member, but does this really mean that their lives will be happier and the time will be better spent? It is quite possible that knowing ahead of time would be like dooming a family to endure a hovering black cloud of worry and the biting truth that their child or sibling will be leaving within a very short period of time. If it were assumed that a child would die before the age of eight, would his parents bother to enroll him in school or would they just spend all the time they had with their child? A common reaction to the death of a family member is to wonder whether there was time that could have been spent with that family member that was 'wasted' on something else. It is likely that this feeling is just so common a human reaction that no amount of preparation will alleviate it.

Situation Where the Test Says No but the Child is Abnormal

The last scenario is one that many people may not at first consider. Even with recent advances it is still impossible to make a perfect prediction with 100% accuracy. Knowing this, doctors and genetic counselors alike are trained to notice things that will cause problems and those that will not and every potential for a problem is not cause for alarm. It is possible then for a genetic defect to be overlooked or missed completely if we do not have the technological means for detecting it. Genetic screening should in no way be thought of as a guarentee or absolout proof of anything. An attitude like this would lead to parents blaming doctors when a child proclaimed to be "absoloutly normal" develops a genetic defect or dies suddenly. It may become the case that genetic counselors are held accountable for the accuracy of their predictions and if that happens they may go the way of the weatherman and fall into disreguard. Genetic prediction and testing are simply tools and should not be blamed for results. In the same vein, these tools should they be trusted entirely.

The genetic counselor must be trusted to make decisions concerning what information is relavant to patients and what information is either too questionable or too insubstantial to mention. For instance let us consider a hypothetical test for a specific genetic disease. On one occasion this test comes up with a 10% chance that a child will have that genetic disease. Is 10% a large enough chance to worry about? The severity of the disease, the accuracy of the test, and the history of the family in question are all factors a genetic counselor must weigh because the wellfare of the family could depend on this choice.

Child

Expecting mothers who get their children tested for genetic diseases open them up to a number of potential problems. The children may be vulnerable to new kinds of discrimination all because they have the wrong sequence of genes. These children are brought into the world facing new kinds of discrimination and self esteem problems. This is more than cause for analysis of the problems associated with genetically screening children. In addition to problems that occure concerning the child and their family, there are a number of things that take place between the child and the rest of the world as a result of genetic screening.

The self-esteem of a child is fragile and can be damaged by bad results from genetic screening. Bad test results affect self-esteem with a feeling of inadequacy from not having genes that are as good as someone else. Instead of being inadequate in a way that could be overcome, it points out a way in which that child will never measure up to the norm. Acceptance of personal faults is something very hard to, something that most adults do not even master. Asking a child to be able to do so is unreasonable. This raises questions as to how children should be informed and treated if it is confirmed that they have a genetic disease. One such solution could be to wait a while before anyone in the public were to be told. This would allow for the child to mature into a young adult until they are at a point in which knowing about their genetic disorder would be more helpful that harmful. Children who have been tested for genetic disorders fall either into the category of those who were tested for certain conditions prior to birth through technologies such as amniocentesis because of a concern over a potential genetic

disorder, or through a post birth test in response to a physical condition that the doctors have suspected is of genetic origin. In either case the child's genetic risks have been determined. If money for the test was obtained by a health insurance agency then those test results are usually subject to review by the agency that paid for them. This is now a problem because insurance agencies use this information in determining not only how much health insurance will cost but if they will give health insurance to that person (Alper, 268-279).

Giving people insurance rates based off of genetic conditions has its faults. When testing people for genetic diseases, tests on certain sections of chromosomes are used in order to determine if someone is at risk for developing a genetic disorder. In some cases it is very clear and can be determined with 100% accuracy that someone will have a condition, although the severity of it will be unpredictable. In other cases the expression of certain alleles means only that the person could develop a condition either soon, or later in life. The ability to detect this is important because health insurance agencies tend only to cover conditions that didn't exist before the start of the plan. Most insurance companies consider a genetic disease that someone is at risk for a pre-existing condition, which would not be covered even though the client exhibits no physical symptoms at the time. One thing to consider is the morality of something like this. Having a certain set of genes is not the fault of the person bearing them. They were born into a certain condition and in past times that wasn't a disadvantage. If health insurance companies charge those that have these chances at developing physical symptoms even though they may not then that more unevenly distributes the cost of sickness on those that are, and also sets a

dangerous precedent in which people can be discriminated against for things that might happen even if they also might not ever happen.

A frightening example of this genetic discrimination along with an excessive amount of genetic screening is presented in the movie Gattaca. In this fictitious world most children who are born have been picked as the best possible combinations of their parent's genetic material. Those who were not hand picked embryos faced the challenge of being discriminated by their parents since they did not have the same chance of succeeding. Those who were not hand picked faced possible discrimination by employers if their genes showed an unacceptable probability of developing certain diseases or physical ailments. Those who were not hand picked also faced the prospect of potential mates rejecting them solely because of their inferior genetic material. What this fictitious world seen in a movie shares in common with the real world is the ways in which parents put demands on their children, or treat them unfairly because they do not meet their expectations of performance. A parent's evaluation of their child's performance too often is related to physical indications of ability and excellence. Sporting events, competitions and grades set definite benchmarks on performance. A game can only be won or lost, as is a competition. Grades place a numerical mark on perfection. With such definite and strict indications of performance one might ask whatever happened to parents being satisfied with having happy children. It seems that often parents having otherwise happy children have pushed them so far as to make them unhapy. Especially in our modern society, parents can tend to value performance and achievement above all else. The constant pressure from parents to do better can be enough to ruin a child's livelyness and rob them of their precious youth. We now apply the thought of competitive genetic

performance to the constant need to be better and to win. How would a child feel who has "lost" in their parent's eyes because of a genetic trait? It it any better for a child who needs to constantly outperform others because their genes are "better" and thus more is expected of them?

One unanswered question is to what degree are parents self defeating in their search for happyness for their children. One large motivation prospective parents have behind going through genetic counseling is to provide the best for their children. They theorize that in physically making their children better people that they will have an easier time finding happieness in the world. While this might be true, a common occurring case to the contrary is when parents take to improving their children in an extreme fashion. These parents either end up treating their child as an object that they generated, or they end up pushing them so hard to achive that the child is not happy. Two ethical considerations come to mind after looking at the situation. The first is what gives the parents of a child the right to test their child for a genetic disease? (Marteau, 167) Knowing that this action could potentially cause that their child could be blacklisted from health insurance or certain jobs and future positions, should it be left to the parents to decide?

As previous sections have touched on the affects that genetic counselors play in the decisions that prospective parents make, this section will look at situations that occur in genetic counseling that effect no one but the parents. The situations are outside influences that effect the decisions of the parents through pressure from society, close friends and family. Commonly accepted among genetic counselors is that the best way to approach counseling is to try and educate in a non-directive manner. Although non-

directive education of patients has been agreed upon by doctors two problems arise. The first is that not all doctors do so. Some make direct suggestions because they don't know what they are doing, and some do so because they think that their opinion is the correct one. The second problem is that the acquaintances of the parents and society as a whole tends to be very directive in their advice. Both of these situations result in there being a loss freedom of choice by the patients.

Many prospective mothers trying to make decisions for themselves regarding their pregnancies are looking for acceptance from those around them. Their husbands, friends and families all influence the decision that they make in their comments. Supportive non-directive comments from those close to the mother aid in her decision much like those of a doctor. However, unlike comments from those of doctors which are usually mediated by organizational codes of ethics. Unfortunately for expecting mothers, friends can make not so subtle hints, families can place expectations or put pressure to make certain decisions, and husbands can be anywhere from unsupportive to manipulative.

There are a number of cases in which the doctor has little to no choice in providing a non-directive approach. In a number of cases patents felt obliged to go along with whatever the doctor was recommending because they felt it was the right thing to do and that the doctor knows best (Alper 115-117). This kind of behavior was described as typically occurring in those that had little or no education in the science of reproduction. These cases are sometimes the hardest because any explanation of genetics, or biology has to start from the absolute basics. When all of this occurs the patient's decision is solely based on the advice of the doctor, which may or may not be the best solution for the patient's physical or mental health, or for that of the potentially unborn baby.

Conclusion

At the moment, the advances in prenatal screening and diagnosis have been very significant. The methods and combination of methods used currently, can predict the possibility of a child having one of a number of particular genetic diseases. It is obvious that not all genetic diseases can be detected, and for those that can be, the degree of accuracy still varies from one disease to the next with no guarantees. Significant technological advancements have been crucial in the development of prenatal testing. Due to this fact, there are even greater expectations for the rate at which technology will advance from now on. Every year we can we know more, we can detect more, but how these capabilities are intergrated is up to society.

Biotechnology is a constantly changing field through the new technologies and methods of biological manipulation. The current trends in technology could lead to better, more accurate methods and could eventially be developed to the point at which the the presence of certain sequences of genes could be used to accurately predict if someone would have a certain condition, and how severe it would be. In the cases of genetics increasing or decreasing the chance that certain environmental conditions would trigger chronic diseases such as cancer, better odds could be given as to the increased risk to the patient. The societal trends that have been taking place are much more difficult to classify and little certainty can be given in the scope of a report such as this. The only certainty is that the current biotechnological trends of invention and innovation will lead to many more choices that society will continue to face. As with any advancement, prenatal

testing will be used by people towards whatever goals they see fit, which will differ depending on cultural and social background.

Improvements in biotechnology will, without a doubt, lead to change in society. One direction of change could be in the direction of the science fiction movie, 'Gattaca', in which people's possibilities are based upon their genetic make-up. Potentially positive outcomes could be found anywhere ranging from being able to reduce the number of people with genetic diseases to being able to more accurately predict when genetic diseases will manifest themselves as chronic illnesses. These potentials come with risks and problems. It will be up to our and future generations to decide which of these paths will be the most beneficial to society.

One of the first things that shocked us was the massive number of choices and pressures that are involved with any sort of genetic manipulation. Before genetic manipulation was possible there was relatively no choice at all. Doctors didn't have to educate patients and patients didn't have to make decisions about their fetuses. Things happened that were apparently completely out of their control.

As we progressed through research and critical evaluation, there were a number of transformations of ideas that took place. Because of our different backgrounds, none of us reached the same conclusion at the end of this project. We all have the simmilar concern that the developing technologies will not be used in moral or societally benificial ways. However, those terms may be personally defined. These concerns stem from the addition of choices that did not exist prior to the creation of this technology.

One of the group members started off with a curious and optimistic outlook, wondering how the technology could be used to benefit everyone in society. He ended the project thinking that this technology will not be used equally and was left wondering what specific abuses will occur.

Another member began with 'big-picture' moral issues in mind believing that it was a question of doing things for the greater good. Throughout the research, he realized the importance of the resulting impact on individuals. He ended the project more concerned about personal issues, especially those of the family and children.

The remaining two members began the project looking at the issues of prenatal testing from a strictly religious point of view. After researching the scientific and philosopical aspect of the topic at hand, it was found that they came to conclusions different from each other and, individually, they were able to look at the issue from a more open point of view. One of the members became more interested in the scientific involvement in prenatal testing rather than its interference with religious beliefs. He found the possibilities of the technologies to be intriguing, but their applications are more worrying than initially anticipated. The other member, instead of being concerned about whether prenatal testing and counseling itself is right or wrong as he originally was, he ended up being most concerned about the motive for using all or particular prenatal testing technologies. In light of this, he had the proclivity of being skeptical about prenatal counseling, an intricate part of prenatal testing. This propensity stemed from a concern that counselors most probably would not be able to give advice in a manner that appropriately handles the various motives that people or families would have.

When we started this project we came from different scientific, philisophical and religious backgrounds. We started off relatively optimistic about the technology as a whole. We were awed by its potential uses and benefits. With a group as small as ours, we had multiple views and multiple ways of interpreting the same information. When prenatal testing becomes a social and a world issue eventually, the number of interpretations will be staggering. This means that there will never be any universal opinion on this matter. No one way that this will affect society. The world will most likely remain divided on the issue. In fact, society will be affected on so many fronts that it is very difficult at this point to predict what will change as a result of these advances. All we can do is educate ourselves so that when the time comes we can all make the best choices possible. And Life.

Timeline of Genetics:

Important Events

DATE	EVENT
1859	Charles Darwin wrote "On the Origin of Species".
1865	Mendelian Laws were developed.
1876	Francis Galton proposed the "law of ancestral inheritance".
1882	Walther Flemming discovered chromatin and saw that the chromatin separated into threads known as chromosomes.
1888	Theodor Boveri proposed the idea that chromosomes were involved in heredity.
1895	X-ray technologies were invented.
1902	Archibald Garrod proposed the theory of "inborn errors of metabolism". Walter S. Sutton and Theodor Boveri independently proposed the chromosome theory of heredity. McClung discovered sex chromosomes but did not decipher their function.
1904	William Bateson showed that more than one gene may be required for certain characteristics or traits.
1910	Morgan discovered a recessive sex-linked mutation in Drosophila and confirmed the Chromosome Theory of Heredity.
1913	First linkage map created by Alfred Sturtevant.
1925-27	H. Muller used x-rays to cause artificial gene mutations in Drosophila.
1931	Harriet Creighton and Barbara McClintock showed that chromosomal pieces can be exchanged, causing genetic recombination.
1934	John Bernal used X-ray crystallography to examine protein structures.
1941	George Beadle and Edward Tatum proposed that one gene encodes one protein.
1944	Avery, MacLeod, and McCarty suggested that DNA is the molecule that mediates heredity.
1950	Erwin Chargaff discovered a one-to-one ratio of adenine to thymine and guanine to cytosine in DNA.
1951	Rosalind Franklin used X-ray diffraction to take photos of DNA.
1952	DNA was confirmed to be the molecule that mediates heredity.
1953	Francis Crick & James Watson discovered the three dimensional double helix shape of DNA. It was seen that the order of the base pairs within DNA encoded the genes.
1955	13 National Genetic Counseling Clinics in the US were open.
1957	Crick proposed that the primary function of genes was to manufacture proteins.
1958	Arthur Kornberg purified DNA polymerase I from E. coli.
1959	Messenger RNA was found to be the intermediate between DNA and protein.
1961	François Jacob and Jacques Monod developed a theory showing how certain genes are activated and suppressed.
1963	Nirenberg had interpreted thirty-five triplets.
1966	Marshall Nirenberg & H. Gobind Khorana discovered that triplet mRNA codons specify each of the twenty amino acids.
1969	Jonathan Beckwith isolated a bacterial gene.
1970	Hamilton Smith & Kent Wilcox isolated the first restriction enzyme.
1972	Paul Berg & Herb Boyer produced the first recombinant DNA molecules.
1973	Annie Chang & Stanley Cohen showed that a recombinant DNA molecule could be maintained and replicated in E. coli.
1977	Fred Sanger developed DNA sequencing technology.

1980	450 Human genes mapped.
1983	James Gusella demonstrated that the Huntington's disease gene is on chromosome 4.
1985	Lloyd Smith and Michael and Tim Hunkapiller invented the first automated sequencer.
1986	PCR is developed by Kary Mullis.
Mid 80's	1,500 human genes mapped.
1988	The Human Genome Project began.
1989	Francis Collins & Lap-Chee Tsui identified the gene coding for the protein on chromosome 7 that causes cystic fibrosis.
1990	2,000 human genes mapped.
1996	The yeast genome was completed.
1998	The C. elegans genome was completed.
1990's	Pat Brown et. al invented DNA microarrays.
1995	J.Craig Venter et. al patented the first completely sequenced genome, Haemophilus influenzae Rd.
1996-7	Ian Wilmut and colleagues cloned the first mammal (Dolly the sheep).
1999	Fully automated instrument could sequence up to 150,000,000 base pairs per year.
2000	Celera Genomics announced they had completely sequenced the human genome within four months and assembled the DNA base pairs within the following six months. The Human Genome Project announced their developments in regards to the project at hand, however they had only sequenced 85% of the human genome.

Bibliography

He hAlper, Joseph S., et al. The Troubled Helix. Maryland: John Hopkins University Press, 2002

Alpern, Kenneth D. Ethics of reproductive technology. New York: Oxford University Press, 1992

Asch, Adrienne and Erik Parens. Prenatal Testing And Disability Rights. Washington, D.C.: Georgetown University Press, 2000.

Bergsma, Daniel, M.D. Ethical, Social and Legal Dimensions Of Screening For Human Genetic Disease. New York, NY: Stratton Intercontinental Medical Book Corporation, 1974.

Bosk, Charles L. All God's Mistakes: Genetic Counseling in a Pediatric Hospital. Illinois: The University of Chicago Press, 1992

Capron, Alexander M., et al. Genetic Counseling: Facts, Values and Norms. New York: Alan R. Liss, 1979

Dale, Jeremy W. and Malcolm von Schantz. From Genes to Genomes: Concepts and Applications of DNA Technology. West Sussex: John Wiley & Sons, LTD., 2002.

Drlica, Karl A. Double-Edged Sword. New York: Helix books. Addison-Weseley Publishing Co, 1994.

Dworkin, Roger B., Gregory P. Gramelspacher, Judith A. Grandbois, Kimberly A. Quaid, David H. Smith, and Gail H. Vance. Early Warning: Cases And Ethical Guidance For Presymptomatic Testing In Genetic Diseases. Bloomington, IN: Indiana University Press, 1998.

Ettorre, Elizabeth. Reproductive Genetics, Gender and the Body. Florence, KY, USA: Routledge, 2002. http://site.ebrary.com/lib/wpi/Doc?id=10016889&page=33

Frankel, Mark S. and Albert H. Teich. Genetic frontier: Ethics, Law, and Policy. Washington DC: American Association for the Advancement of Science, 1994

Fuhrmann, Walter and Friedrich Vogel. Genetic Counseling. 2nd Ed. Heidelberg Science Library vol 10. New York: Heidelberg 1976.

Haverty, Carrie. Prenatal Testing Counselor at Vincent Obstetrical Ultrasound and Prenatal Diagnostic Center at Massachusetts General Hospital. Email interview, April 20, 2004.

Heyman, Bob. Risk, Age and Pregnancy : A Case Study of Prenatal Genetic Screening and Testing. Gordonsville, VA, USA: Palgrave Macmillan, 2000. http://site.ebrary.com/lib/wpi/Doc?id=10044905&page=107

Hood, Leroy and Daniel J. Kevles. The Code of Codes: Scientific and Social Issues in the Human Genome Project. Cambridge, MA: Harvard University Press, 1992.

Kalb, Claudia. "Brave New Babies." Newsweek 26 January 2004: 45-53.

Kaplan , Jonathan Michael. The Limits and Lies of Human Genetic Research; Dangers for Social Policy, New York, 2000.

Kuhse, Helga and Peter Singer. Should The Baby Live?. Oxford: Oxford University Press, 1985.

Marteau, Theresa and Martin Richards. The Double-Edged Helix. Cambridge, Great Britan: Cambridge University Press, 1996

Murray, Thomas H., Robert F. Murray, Jr., and Mark A. Rothstein. The Human Genome Project and the Future of Health Care. Indianapolis: Indiana University Press, 1996.

Oakley, Ann. The Captured Womb. Oxford: Basil Blackwell Inc., 1984.

Rothman, Barbara Katz. The Tentative Pregnancy. New York: Viking, 1986

Sadick, Tamah L. and Siegfried M. Pueschel. Genetic Diseases and Developmental Disabilities: Aspects of Detection and Prevention. Boulder, Colorado, USA: Frederick A. Praeger, 1979.

Shannon, Thomas A. Genetic Engineering: A Documentary History. Westport, CT: Greenwood Press, 1999.

The President's Council on Bioethics. Beyond Therapy: Biotechnology and the Pursuit of Happiness. Washington DC: The President's Council on Bioethics, October 2003. Accessed 1 May 2004. Available at: http://www.bioethics.gov/reports/beyondtherapy/index.html