

A Review of Prenatal Genetic Testing and its Ethical Issues

Prenatal genetic testing is just one of the medical practices that is questioned and regulated by ethical debate. It is used to screen for birth defects and diagnose any other disorders or traits during pregnancy. The hope is to provide families with the capabilities to make well-informed and appropriate decisions about pregnancy and reproduction. This testing also helps doctors have the most accurate and helpful information to assist their pregnant patients. Although prenatal screening is not diagnostic, there are a multitude of other tests that are controversial yet provide the patient with further information. People choose to have tests for many reasons. Perhaps the mother is older and so has a greater chance of having a child with defects, or the family may have a history of genetic related conditions, or because prior screening showed an indication. Issues include social, legal, and ethical concerns. Although there are ethical issues such as selective abortion, parental rights, stress of choosing tests, risks, fears, and the role of the law, prenatal genetic testing is a technology that can be implemented in clinics and be a helpful tool for planned prenatal care.

Screening tests may determine if a fetus has a high or low probability for certain conditions. There are over 1000 genetic tests that are either currently applied or being developed (Hudson). Doctors are encouraged to record family histories, their genetic risks, and consider the age of the mother and ethnic background, in order to make a proper decision on which tests to conduct and what to look for (Hudson). Tests may not only discover conditions the baby would be born with, but also those that may present later in life, such as Huntington's disease (Hudson). Also, some tests may show any predispositions and increased risks of certain adult diseases, such

as breast cancer (Hudson). The fact that a cure for the condition may be found during the individual's lifetime makes abortion more difficult. Also, testing does not guarantee with 100 percent certainty that the condition will develop (Hudson). In any case, prenatal genetic testing allows parents to gather information to help with difficult decisions, especially in high-risk cases.

There are many tests involved in prenatal genetic testing. Maternal serum screening views the mother's blood for various proteins, the most common being AFP (alpha-fetoprotein), hCG (human chorionic gonadotropin), uE3 (unconjugated estriol), and DIA (dimeric inhibin A) (Common). These tests are useful for determining the degree of risk the fetus has for conditions. However, these are not diagnostic. A negative test helps by indicating a significantly lower probability that the fetus may develop the condition (Common). The American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) encourage maternal serum screening if one parent is Caucasian (Common). One type of maternal serum screening is AFP X-tra that specifically looks for open neural tube defects, Down syndrome, and trisomy 18 (Common). Table I shows various maternal serum screening tests and the detection rates according to these three conditions.

Table I:

| Maternal Serum Screening Test | Gestational Age (in weeks) test can be done | Open neural tube defects | Detection Rates* | |
|---|---|--------------------------|---------------------|------------------|
| | | | Down syndrome | Trisomy 18 |
| AFP (tests only for AFP) | 15.0-23.9 | 85% ¹ | N/A | N/A |
| AFP-Xtra (includes AFP, hCG, and uE3) | 15.0-21.9 | 85% ¹ | 60% ¹ | 60% ¹ |
| AFP Tetra (includes AFP, hCG, uE3, and DIA) | 15.0-21.9 | 85% ¹ | 75-80% ² | 60% ¹ |

* Detection rate refers to the percentage of affected pregnancies that will have a positive maternal serum screening test.

Source: (Common)

Another procedure that is very common during pregnancies is amniocentesis. This prenatal diagnostic testing uses amniotic fluid as it contains cells that have been shed by the fetus (Common). These skin cells allow for studies related to fetal chromosome abnormalities. One common example is an extra chromosome 21, indicating Down syndrome (Common). Other diagnoses involve open spina bifida, abdominal wall defects, and inherited faults (Common). Amniocentesis is commonly conducted during the second trimester, between weeks 16 and 18 (Common). It tends to be successful by detecting about 99% of fetal chromosome abnormalities (Common). Testing the amniotic fluid AFP (alpha fetoprotein) can also be done by amniocentesis, which if elevated, can test for AChE (acetylcholinesterase) (Common). About 98% of the open neural tube defects, including spina bifida, are seen by testing AFP and AChE (Common). Although this test is successful for the majority of the time, it does have the potential to cause a miscarriage, but only at a low rate of 0.5% (Common). Amniocentesis, as well as an ultrasound examine, can determine the sex of the child, creating further ethical turmoil (Sandel).

Ultrasound is a noninvasive examination that develops a picture of the fetus. During the second trimester, it can determine 30-50% of birth defects (Common). It does not analyze chromosomes or genes, but may reveal conditions resulting from chromosome abnormalities.

Chorionic villus sampling (CVS) diagnoses chromosome abnormalities, inherited diseases, and other birth defects (Common). CVS is especially recommended for women above the age of 35 and those with suggestive screening test results (Common). It is commonly tested between weeks 10 and 12. By using the placentas' fetal cells, 99% of chromosome abnormalities can be diagnosed, with the exception of open neural tube defects (Common). This test too can cause a miscarriage or other complication, but at a rate of 0.5-1.0% and mostly before week 10 (Common). Amniocentesis is needed in 1-2% of CVS tests to confirm results (Common).

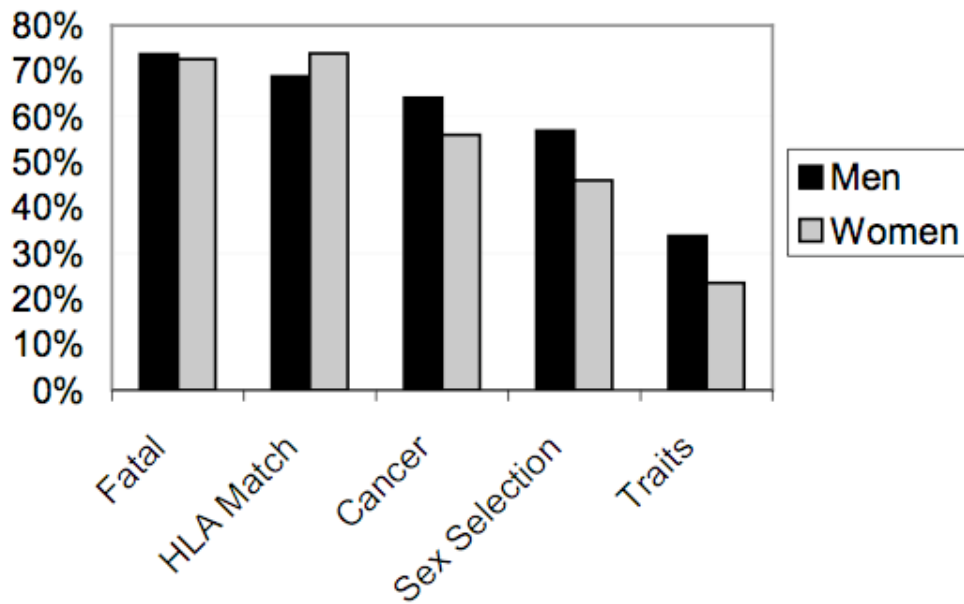
Two forms of analysis are used for prenatal genetic tests. Cytogenetic analysis views the chromosomal number and structure. Molecular genetic testing examines the DNA sequence of genes (Hudson). Prenatal genetic tests tend to be reliable and are rarely false (Hudson).

However, due to the gravity of the decision based on the results, it is critical that tests are valid before going on the market, that all laboratories conduct the tests properly, and that federal agencies ensure that each laboratory will properly use each test for accurate results (Hudson).

There are multitudes of ethical questions that arise from the capabilities of these prenatal genetic tests. Many people find it necessary for society to take responsibility, define what is morally right, and what interests to preserve. In 2004, the Genetics and Public Policy Center conducted a survey to see what Americans thought about prenatal genetic testing. Two thirds of the people were in favor of testing for fatal childhood diseases and for tissue matching (Hudson). A lower number, yet still majority, were comfortable with testing for diseases that develop in adulthood. Much less people approved testing to find or select the gender of the fetus and even more people were against genetic testing to find traits such as intelligence or strength (Hudson). In general, Americans feel that prenatal genetic testing should only be used for health reasons to not take advantage of science for personal gains beyond what is healthy and natural. Figure 1 displays the results of the study.

Figure 1:

Approval of prenatal genetic testing by purpose



2004 Survey of 4,834 Americans

Source: (Hudson)

Some people feel as if there are no ethical issues with prenatal genetic testing as it would limit human suffering, remove conditions that would be eventually eliminated by natural selection, and end pregnancies that have a predisposition to terminate rapidly (Campbell). Others argue any termination is an evil act against life and human dignity. However, the other side argues the testing will provide the best quality of life possible and will make sure parents have a child they can raise with hope to be happy, healthy, and successful. Five main issues surround prenatal genetic testing: selective abortion, parental rights, the consent and stress during testing, the risks versus the benefits, and the chance of designer babies becoming a wide spread trend.

Abortion is an issue not limited to prenatal genetic testing. However, the results of the testing can be convincing for parents to end a fetus's life if that fetus would grow to be an abnormal and unhealthy child. Yet prenatal genetic testing can also assist parents who would not

abort, but would have time to prepare emotionally, financially, and medically for the birth and life of their child (Hudson). Some people choose not to have testing done so it would not influence their decision on having the baby or not. Yet for those who consider abortion, testing is extremely helpful. Many encourage parents to seek information on their genetic risks and their options to testing before conception (Hudson). This is important for those who find it difficult to terminate a pregnancy. The testing results may be a deterrent for potential parents considering pregnancy as they find out what the risks and likelihood of genetic abnormalities would be for their children. Some feel that aborting a sick fetus is prenatal euthanasia, bringing this other controversial issue into the light (Campbell). Some believe the law should require termination of abnormal fetuses to relieve the child of a handicapped, sick, and painful life, as well as to alleviate society's place in supporting the handicapped emotionally and financially (Campbell). However, many people question whether the law should have a say in personal and religious matters such as this. One opinion is that medicine is used to help people live the best life possible. Therefore, if a child cannot prosper in society, some believe it is best to give the position of a child in a family to one that will be able to thrive and look forward to a long life. As many families have a set number of children they are willing to have, or are limited by their government in the number they can have, it is crucial to many to make sure that child has the best chance of surviving, caring for the family, and having a happy and successful life. Therefore, some find that aborting a fetus would not simply be ending one life, but easing the anxiety of a family's dynamic and allowing others to be conceived and grow.

The rights of the parents must also be considered when discussing prenatal genetic testing. These tests may provide parents with reassurance and help them make a decision that is best for them, their family, and their bank accounts. Although prenatal genetic testing should be

available to all parents, many agree they also should have the right to refuse it (Hudson). In order to give parents this choice, there needs to be accessible and informative counseling available to those considering children and to those who are pregnant. Often, parents go through with testing and don't realize the consequences of the results that may lead them to making decisions they hadn't previously considered (Hudson). Also, clinics are working to present the results in appropriate manners, to keep the parents relaxed, well informed, and not under pressure. Some healthcare providers may assume that parents with affected children will automatically choose abortion, therefore limiting other options such as having the child and preparing a medical plan in advance (Hudson). Many believe that information on how to raise affected children and the responsibilities and common challenges be given to all parents, even if they have already made a decision (Hudson). Also, some agree that parents should have access to stories from people living with the genetic defects as well as the family's' opinions to see a real life account of what they are faced with and determine if they can handle it, treat the child equally, and learn how to adapt (Hudson).

Parental rights tie directly into the issue of stress during prenatal genetic testing. Parents have an obligation to raise their children, and to do it in the best way they can. Although prenatal genetic testing has non-directive genetic counseling, in which the parents are given information and they choose on their own as the subject is very personal, many people claim that the healthcare providers push patients to continue with prenatal genetic testing (Hudson). This stressor comes from society as well as healthcare providers and increases as the technologies and number of tests escalate. The Genetics and Public Policy Center found that 64% of Americans agree that due to how personal these issues are, people should decide for themselves to have testing done (Hudson). Consequently, most Americans feel that the government should not make decisions on the matter but should instead focus on providing parents with unbiased information.

As with many ethical issues, there are a number of risks that contrast with the benefits. First off, there is a fear of liability, where healthcare providers could be held responsible for not providing or considering all genetic issues (Hudson). This could provoke people to give as much information as possible about genetic testing (Hudson). However, with the number of tests that are frequently released to the market, it may be difficult to expose all couples to all tests right away. Also, this fear would promote healthcare providers to highly encourage testing, when many find that this decision should be the parents'. Another risk is providing results of early pregnancy tests, which are not as accurate as the later stage tests (Campbell). Also, some emerging tests may become preferable. However, since they are less tried, the issue comes up of allowing mothers to be research subjects, who may be necessary for scientific reasons to help future mothers, and understanding that since there haven't been many trials, this test seems dependable and extensive, but the uncertainties are great (Campbell). Another risk involved to the parents or the healthcare providers is that the affected individual may later sue for being given a "wrongful life" when they had the opportunity to save him/her (Campbell). The low chance of hurting a fetus during the test is also a risk parents must be aware of. Yet, despite these issues, the primary focus has been to maintain the health of the mother and fetus (Campbell).

As genetic testing technologies increase, more information on specific traits and genes will be revealed. Therefore, genetic testing and manipulation lead to the ethical question of designer babies, where parents choose which genes to pass on their children. The controversy lies in whether is it ethical to alter a child's genes, for medical reasons, aesthetic purposes, and/or physical and mental capabilities? Society may be more tolerant when people understand their own genome. They may also accept differences more readily as they come to understand there is no perfect being (Caplan). Others fear this will turn into a form of eugenics. However, keeping the choices free from the states' decisions and everyone having different opinions of the best

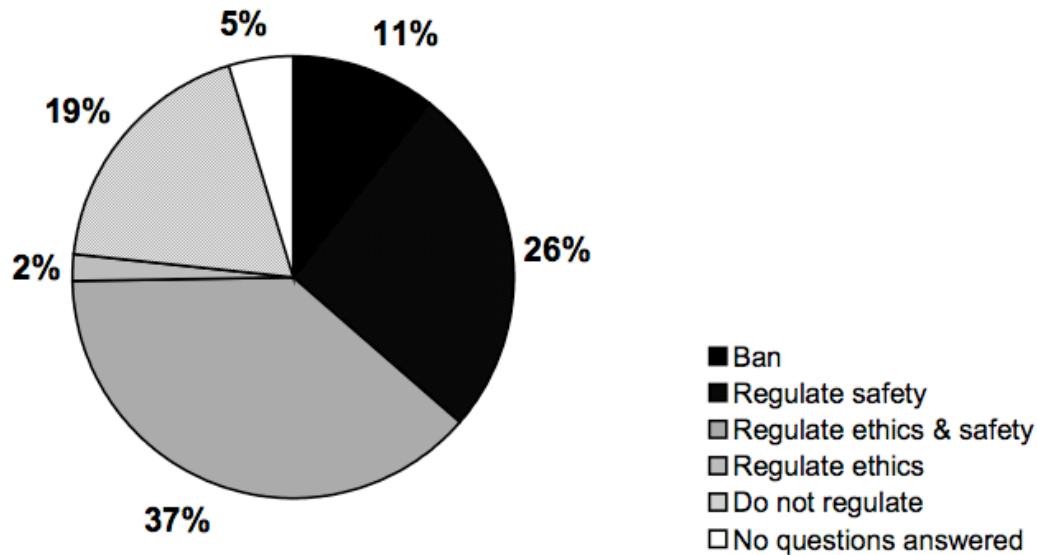
traits, will not be the same type of eugenics which bears a bad name due to instances such as the Holocaust (Caplan). Yet more issues arise, such as the social implications. For example, it is probable that since the healthy subsidize the handicapped with health costs in many places, perhaps the commitment to disability facilities will be less likely since people had the choice to have a child born without genetic abnormalities and should be responsible for this decision (Sandel). Both the individual rights and societal rights are considered, each advocated by various views of academia, such as the first encouraged by Arthur Caplan and the later supported by Michael Sandel. This would be a result of society forming genetic testing as an obligatory and necessary act, when it should be the parent's choice (Hudson). Since amniocentesis can determine the gender of a fetus, and in vitro fertilization can choose a sex, it is probable that fetuses be terminated based on gender (Sandel). The choice children have for how they want to live their lives and their autonomy may be hindered by genetic alterations, disallowing the freedom others get from discovering their individual talents and faults in their lifetime (Caplan). This may decrease the amount of creativity as children are compiled for a concrete and known future. Parents tend to want the absolute best for their children, and if they could choose to give them an advantage at birth, many would (Caplan). The danger in this is the possibility of creating two classes, one with the capabilities to choose and enhance their offspring, and another that cannot afford or have access to the technology and so are born with normal or unhealthy attributes (Caplan). Although it seems helpful, some believe society should not allow parents to choose the genetic makeup of their children as the fears of societal impact and the natural essence of a human being are too risky to interfere with (Sandel).

The question of how involved the government should become in prenatal genetic testing is one that has many opinions and answers. Currently, there is no governmental review of

prenatal genetic tests by the Food and Drug Administration or other agencies before being put onto the market (Hudson). This disconnects the government with the ethical issues, but also may be frightening to those who are unsure of the health risks involved. However, the testing does have some regulation. The Centers for Medicare and Medicaid Services (CMS) works through the Clinical Laboratory Improvement Amendments of 1988 (CLIA) to regulate prenatal genetic testing (Hudson). However, it is difficult to determine how efficiently the tests are being conducted because CLIA has no explicit requirements for the molecular aspect of testing (Hudson). Congress has the potential to force the FDA and CMS to further investigate prenatal genetic testing and ensure its proper implementation (Hudson). This is necessary since professional groups such as the American College of Obstetricians and Gynecologists and the American College of Medical Genetics are highly depended on, especially with the influx of tests, and do not have all of the necessary capabilities to properly monitor genetic testing (Hudson). Federal funding is necessary to keep up with the science and so the government may employ the Department of Health and Human Services to assist these groups. The issue of regulation arises when exactly how far should these regulatory groups should go. Figure 2 displays results based on a study done by the Genetics and Public Policy Center to see where Americans stand on governmental regulation of prenatal genetic testing (Hudson). The majority believes the only regulation should center on the ethics and safety, ensuring the testing does not become undisciplined and the safety and health of the child and parents are the highest priority.

Figure 2:

**Regulating Prenatal Genetic Testing:
Distribution of opinions among Americans.**



Source: (Hudson)

In England, the Abortion Act of 1967 allowed selective abortion to be permitted as long as the fetus was expected to have serious abnormalities once born (Campbell). This included both mental and physical conditions as well as if the affected child would hurt the status and mental health of the mother or family (Campbell). In the United States, no such act has been made, but the issue is highly discussed. Only twenty-eight states mandate that consent must be given by the mother to perform genetic testing or to obtain genetic information (Newborn). These laws may not be subjected to prenatal genetic screening, but are general genetic privacy laws (Newborn). However, a few states with genetic privacy laws do not consider prenatal screening a part of this and so do not have such high demands for the consent of the mother (Newborn). Twenty-three states allow prenatal genetic screening to be waived if the reason is due to religious views (Newborn). Only twelve states have prenatal screening laws that require confidentiality to be

observed for decisions and results (Newborn). Some states require that genetic tests that are refused be reported through the law system (Newborn). Recently, on October 8, 2008, former President Bush signed the Prenatally and Postnatally Diagnosed Conditions Awareness Act, which gives parents more support and information about specific genetic conditions (Prenatally). This is aimed to make sure that parents understand the gravity of their decisions and all of the options they have. The law also creates a relationship guided by the Secretary to fund collaboration between healthcare professional groups and disability advocacy organizations (Prenatally). Therefore, the gap between regulation and information can start to be closed and parents will be given the necessary means to have the most accurate and reliable tests to make the best decision for them.

Prenatal genetic testing brings up many ethical issues. Society must be aware of the risks, fears, and consequences of prenatal genetic testing so they can choose whether to permit or prevent genetic selection in designing children, but allowing them to prosper in a healthy manner. It is commonly believed that parents should be given information on all of their choices and what each entails. The people want the government to become more involved in the future with funding, monitoring tests, and develop a system to ensure the safety of new tests and the proper implementation of current tests. Also, as designer babies become more probable, clinics are discouraged to advocate the practice as the public displays their views on limiting prenatal genetic testing for health purposes. Keeping a steady set of regulations for ethical and safety purposes, and confining genetic manipulations for only health reasons will maintain the usefulness these tests provide children, parents, and families.