By Lexy Campbell

In a world that feels increasingly uncertain and unpredictable, humans instinctively search for any knowledge that will give us a sense of control over the future. We search for the latest diet fads and weight loss trends all in the hopes of making ourselves live longer, healthier lives. Now, we turn to genetic testing in an attempt to make sense of the complexities of our health. The allure of genetic testing becomes increasingly stronger with innovative genetic companies, such as 23and Me, easily accessible to the public. Through our genetics, we hope to unveil the secrets our bodies hold and be able to control our future health.

Synonymously, with genetic innovations, we seek to influence our unborn children's futures in the hopes they may live healthier lives. These technologies are referred to as prenatal screening, screening for risks of genetic diseases in a fetus. These technologies are growing in popularity around the world. About one-half of all pregnant women in the United States use non-invasive prenatal tests (NIPT).[1] Parents-to-be and lawmakers' decisions regarding genetic screening in unborn children will shape society and all future generations. Although genetic screening has several benefits, the risks associated with screening and future disparities must be further addressed.

Prenatal testing was first developed to tell the parents-to-be the risk that a fetus has of chromosomal disorders, genetic disorders caused by a change in a chromosome(s) in number or structure. These chromosomal disorders included Down Syndrome (trisomy 21) and Turner syndrome (missing X chromosome). The technology has advanced since its inception and now can screen for various non-chromosomal disorders, such as cystic fibrosis and sickle cell anemia. Today's most common prenatal test is non-invasive prenatal test (NIPT). These tests are easily performed through analysis of a blood sample from the mother after ten weeks of pregnancy. The results of these tests tell the parents-to-be the probability that the unborn child has a genetic disorder, like Down Syndrome.

Often, companies advertise their results as highly accurate, meaning that results are interpreted as diagnostics of disease rather than probabilities of disease by future parents. However, the results of screening are not as accurate as advertised. “Private companies have a very big influence a very big influence. The first studies on the market had been from the companies,” said Marion Baldus, a mother and the author of the paper published in *Medicine, Health Care, and Philosophy* on February 2023 that examines the ethical conflicts surrounding prenatal screening. However, according to researchers, the test has only an 80% accuracy for predicting Down syndrome and is less for other diseases like trisomy 18, with only a 60% accuracy in predicting the genetic syndrome.[1]

Additionally, the prediction accuracy can vary depending on the mother's age and pregnancy risk, which may make results less accurate. “If the woman is 40, then the test will have 93% positive predictive value. But if you take the same test and the same condition and have a woman who is 25 years old, the rate goes down to 50%,” said Baldus.[1] The tests are more

1. Baldus, M. (2023). “overestimated technology – underestimated consequences” – reflections on risks, ethical conflicts, and social disparities in the handling of non-invasive prenatal tests (nipts). *Medicine, Health Care and Philosophy*, *26*(2), 271–282. https://doi.org/10.1007/s11019-023-10143-1
2. Liehr, T. (2022, August 18). *False-positives and false-negatives in non-invasive prenatal testing (NIPT): What can we learn from a meta-analyses on > 750,000 tests?*. Molecular cytogenetics. https://pubmed.ncbi.nlm.nih.gov/35986330/#:~:text=Results%3A%20A%2027%3A%201%20rate,been%20aborted%20spontaneously%20before%20birth.

accurate in women of advanced age because older women are more likely to have a child with a genetic disorder than a younger woman. As women age, their eggs age with them and decrease in quality which may result in more genetic disorders.

Several studies predict that literature surrounding the accuracy of prenatal tests is not completely scientifically correct and may biased towards the benefit of the genetic testing corporations.[2] "Greater than 90% of the studies report on NIPT without distinguishing NIPT testing only trisomy 21, 13, and 18 plus sex chromosomes, or NIPT testing all chromosomes for trisomy's…." said Liehr, "All [testing] is discussed together (independent of the method used) and this is not scientifically correct," said Thomas Liehr, a clinical geneticist working in human genetics and prenatal diagnostics since 1992. With a mixture of scientifically incorrect methods and "financial interest of some authors," said Liehr, the results of several NIPT studies can be biased towards the accuracy of prenatal screening.

With existing advertising and biased results that praise non-invasive testing, many parents-to-be have full trust in their results. Especially for parents with a positive screening result, prenatal testing can result in emotional distress and anxiety throughout a pregnancy. With a positive result, "all feeling of security goes. The pregnancy becomes a tentative pregnancy, woman and partner may not start bonding to the unborn," said Baldus. Then, women with a positive screening result must go through diagnostic testing to determine if the positive is a false-positive. In the case of a false-positive, the unborn child likely does not have genetic disorder; however, the damage done to the bonding experience of parent and child remains. If the result is a true-positive, the parents-to-be must make the life-deciding decision to keep the child or abort the pregnancy.

Diagnostic testing does not come without its own risk. Diagnostic testing is an invasive procedure. Invasive procedures are associated with a risk of miscarriage between 0.5% and 2%.[1] Although this risk of miscarriage is small, it should still be communicated with mothers before they consent to a prenatal test. Diagnostic testing should always be confirmed if a parent is concerned about having a child with a disability and wants to make decisions based on prenatal screening.

If the diagnostic test confirms the positive prenatal screening, parents-to-be must decide whether to keep the child, knowing they have a disability, or to abort the pregnancy. This is often a complex decision related to the parent-to-be's religion, beliefs, external pressures, and financial state. Parents must also consider several ethical questions: Is it ethical to bring a child into the world who might suffer? Does aborting a child with a disability mean our society does not value the lives of those with disabilities? Do all disabilities cause suffering? What does aborting children with disabilities mean for the disability community? These are all complex questions relevant to the ethical dilemmas of prenatal screening.

Prenatal screening perpetuates the idea that children with disabilities, hence adults with disabilities, are less desirable in our society. With the increased popularity of prenatal screening

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in the United States, it promotes the idea that having a child with a disability is not normal. This shift in normalcy, can make it more difficult for those with disabilities to exist in our society in the future. Baldus states, "In this respect, prenatal selection can be seen as a modernized form of exclusion, brought forward by the prenatal sphere." At its core, prenatal tests provide information about potential disabilities, allowing parents to make informed decisions about the pregnancy. However, this framing can inadvertently suggest that children with disabilities are less valued.

For some, the practices of prenatal screening evoke uncomfortable parallels with eugenics. Eugenics, a practice used by the Nazi party in World War II, is the belief and practice that aims to improve the genetic quality of the human population. Some believe that prenatal screening is a modernized, more socially acceptable form of eugenics, aiming to test and prevent "genetically inferior traits" within the unborn population. However, this is highly debated. Others argue that it is not eugenics because the goal is to prevent suffering in the unborn child who may live a life with a disability.[1]

The ethical questions of prenatal testing channel to the question: does having a disability mean that a child's life will be filled with more suffering than joy? Additionally, this question can change based on the potential disability the child may be born with. For example, the life of a child with Down Syndrome will be vastly different than the life of a child with cystic fibrosis, and the suffering each experience may be different.

Genetic screening on unborn children is more complex than a blood test and receiving a positive or negative result. It is difficult, and ethical questions and implications for the future of humanity must be considered. For parents-to-be, the choices made because of a positive genetic result will affect the rest of their life and determine their children. Ultimately, it is an individual choice to have genetic screening, so all must be informed of the emotional and ethical risks.

When asked for advice for future parents considering non-invasive prenatal screening, Liehr and Baldus both had guidance to give that paralleled one another

Liehr, pulling from his years of genetic research advised, "take care with that test; understand what is can do and what it cannot do; understand the biological background why is can be never a 100% test – it is only a screening and it is not covering ten-thousands of reasons for genetic diseases."

Baldus, with her experience as a mother and a social worker in her early career advised, "keep calm, think at least twice, look for your resources, find an independent counselor who can counsel without bias, start to think from the end of the process. How could I imagine birth or not birth in my life and how could I stand my decision."

1. Baldus, M. (2023). "overestimated technology – underestimated consequences" – reflections on risks, ethical conflicts, and social disparities in the handling of non-invasive prenatal tests (nipts). *Medicine, Health Care and Philosophy, 26*(2), 271–282. https://doi.org/10.1007/s11019-023-10143-1
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