PRENATAL GENETIC TESTING PERSPECTIVES:

A LITERATURE REVIEW

By

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Abstract

This thesis examines stakeholder perspectives on prenatal genetic testing. The stakeholders include genetic counselors, other medical professionals, parents, the disability community, and disability activists and scholars. The review includes literature from academic sources that identifies and summarizes findings (e.g., surveys, interviews) related to genetic testing/counseling. Primarily, this literature review searched online databases such as Google Scholar and PubMed by MEDLINE. The main focus of this review is to increase understanding of the unique views of different stakeholders and compare and contrast these perspectives. Due to the increasing availability of less invasive forms of prenatal genetic testing, understanding different viewpoints can help direct when and how testing is utilized. By identifying these different viewpoints, future research can focus on the best approach to prenatal genetic counseling.

Keywords: prenatal genetic testing, genetic counseling, NIPT, disability

Prenatal Genetic Testing Perspectives

Congenital disability screening was first developed in the 1950s using ultrasound, and the technology has continued to evolve. This type of screening focuses on gestational age, malformation evaluation, fetal well-being testing, and fetal number. Amniocentesis was the first prenatal testing for chromosomal abnormalities. This test consists of removing amniotic fluid from the mother's uterus (Carlson and Vora, 2017). Chorionic villus sampling is another invasive screening option that is offered earlier than amniocentesis. Researchers in Hong Kong in 2011 developed a new technique called noninvasive prenatal genetic testing (NIPT). Commercial companies first introduced NIPT in the U.S. in October of 2011 (Allyse et al., 2015). This testing involves using the pregnant woman's blood to analyze cell-free fetal DNA to detect fetal chromosome aneuploidies. NIPT is a step between serum screening or ultrasound imaging, which has a high false-positive rate, and invasive diagnostic testing (Allyse et al., 2015). Prenatal genetic testing raises ethical concerns when used to screen for disability. One possible moral dilemma is the termination of a pregnancy that tests positive for disability. Because NIPT is increasing in availability worldwide, stakeholder perspectives are essential to provide a pathway for implementation (Hill et al., 2016). The stakeholder views considered are the medical community, the disability community, disability scholars and activists, and parents receiving genetic testing.

Literature Review

Genetic Counselors

Genetic counseling is the standard of care for genetic testing (Faucett et al., 2019). Genetic counselors are a part of the patient's healthcare team who meet with patients before and

after genetic testing to explain testing options and testing results. Genetic counselors educate pregnant women on their prenatal genetic testing options and help them decide whether they want to pursue testing (Agatisa et al., 2018). This includes educating them on the advantages and disadvantages of different genetic testing types and what type of information one can learn from the test results. Genetic counselors also meet with patients who have received prenatal genetic testing to discuss the implications of the products, help them prepare for having a child with a genetic condition, and share information about options to terminate the pregnancy (Agatisa et al., 2018).

The National Society of Genetic Counselors (NSGC) identifies essential considerations for the use of non-invasive prenatal testing (NIPT), which is increasing in availability worldwide. When their position was released in 2012, the NSGC supported NIPT as an assessment option during pregnancy (Devers et al., 2013). This is based on data showing that NIPT has been proven to detect aneuploidy in fetuses effectively. The NSGC believes that NIPT should be used as a screening tool only when there is a possibility of false-negative results. The NSGC does not recommend the use of NIPT as a diagnostic tool. Additional diagnostic testing should be used for patients that receive positive NIPT results (Devers et al., 2013). In 2012, the NSGC only recommended NIPT for high-risk patients, not as routine screening even though screening of the average risk population has been shown by one study to be effective. NSGC also recommends having pretest counseling to inform patients about disorders NIPT testing can detect and concomitant limitations. In addition to pretest counseling, post-test counseling by a certified genetic counselor is recommended by the NSGC for any patient with an abnormal or positive test result (Devers et al., 2013). As for future decisions, the NSCG recognizes that the NIPT's landscape is rapidly changing. It will be necessary to revisit these recommendations as the

technology evolves. Overall, the NSGC supports NIPT as an option for patients that have pregnancies that are at a higher-than-normal risk for chromosome abnormalities as long as these patients provide informed consent and are provided the education and counseling by an appropriately qualified medical provider, such as a genetic counselor (Devers et al., 2013).

In addition to their position statement regarding NIPT, the NSGC has also released a position statement on prenatal testing for adult-onset conditions. The primary ethical considerations for prenatal genetic testing for adult-onset conditions are the parents' reproductive rights, the rights of the future child, the right not to know, and the adverse effects on childhood (Hercher et al., 2016). The prospective parents have a right to know their fetus's genetic status, and NSGC's previous statements support this. The main controversy exists because prenatal testing results for adult-onset conditions do not generally affect medical care in childhood, so this testing has little benefit (Hercher et al., 2016). Also, it does not allow the child to decide whether they want to learn their genetic status for adult-onset conditions. The right not to know has been established so that an adult can deny predictive testing. This right is taken away from children tested for adult-onset conditions before they are born (Hercher et al., 2016). A child knowing that they have a genetic predisposition for an adult-onset disease can negatively affect the child, such as self-esteem issues, stigma, and being treated as a vulnerable child. The NSGC concluded that genetic counseling should be provided for all parents considering prenatal genetic testing for adult-onset conditions. Any conflict between the parent's rights and the future child should be resolved in the parent's favor (Hercher et al., 2016).

A study was conducted by Horsting et al. that explores the thoughts, opinions, personal experiences, and concerns of prenatal genetic counselors regarding NIPT. This study was performed by creating a 67-question survey answered by 236 individual genetic counselors who

saw at least one prenatal patient a week (Horsting et al., 2013). Generally, the respondents supported NIPT as an invasive testing alternative. Most of the genetic counselors felt that NIPT should only be used as an advanced screening tool rather than a diagnostic test, though 35.6% thought that it should be used to rule out chromosomal abnormalities. The genetic counselors expressed concern for the current and future uses of NIPT by patients and some obstetricians but expressed the belief that educating patients and providers will decrease current misconceptions about the capabilities and the intended use of this type of testing (Horsting et al., 2013). Ninety-eight percent of genetic counselors in the study agreed that pretest counseling is necessary for NIPT. This research study is limited in that it is a survey-based study, meaning the participants were interested in the subject matter enough to complete the survey. Also, about 8% of the respondents had not offered NIPT in their work. The survey was done with every effort not to have bias, but bias may have been inadvertently introduced (Horsting et al., 2013).

Another study regarding genetic counselor's perspectives on NIPT by Agatisa et al. conducted interviews with 25 genetic counselors. This study hoped to identify genetic counselors' views on expanding the use of NIPT screening over time (Agatisa et al., 2018). The study participants agreed with NIPT's use to screen for common autosomal aneuploidies such as Down syndrome. The genetic counselors had some reservations regarding using NIPT to identify the sex of the fetus and to screen for microdeletions in the genetic makeup of a fetus. The participants identified a barrier to ensuring that patients have the support and current information needed to make informed decisions about NIPT screening (Agatisa et al., 2018). The researchers believe that this problem should be addressed by adding additional education interventions for patients and educating obstetricians who work closely with patients regarding genetic risk and NIPT. The participants noted that genetic counselors' professional responsibility is to provide

this education to obstetricians regarding NIPT (Agatisa et al., 2018). The small sample size of 25 genetic counselors is a limitation of this study. Due to the sample size, the views reported in this study may not be representative of all United States genetic counselors. Overall, the respondents concluded that there are not enough genetic counselors to meet NIPT's counseling needs at its current trajectory and that educating obstetricians to provide certain aspects of counseling to patients about NIPT could help resolve this issue (Agatisa et al., 2018).

Susan Markens completed a study to explore genetic counselors' views on prenatal testing and genetics by conducting recorded interviews with 26 board-certified genetic counselors. These genetic counselors explained prenatal genetic testing as "It's kind of a fine line to draw," meaning that prenatal testing can lead to an increase in choice and empowerment for pregnant women but can be damaging if ambiguous results are received (Markens, 2013). Many genetic counselors interviewed described the costs and benefits from prenatal genetic testing. The genetic counselors weighed the benefit of autonomous choice while acknowledging genetic testing limitations (Markens, 2013). The genetic counselors were aware of genetic testing's disadvantages, such as the uncertainty and anxiety-provoking decisions it can cause. The genetic counselors in this study valued autonomous and persuasion-free choices for their patients over their personal beliefs. This study was limited due to the focus on genetic counselors in the United States, so further research is needed to ascertain a global perspective (Markens, 2013).

NIPT is also used in the United Kingdom. Genetic counselors from the United Kingdom support NIPT as an alternative to invasive prenatal genetic testing for high-risk patients. However, their beliefs differ on using NIPT as a genetic screening method for patients without high-risk family histories due to ethical, counseling, and practical concerns (Alexander et al., 2015). A study conducted on 20 genetic counselors in England found that they favor prenatal

genetic tests that are accurate and reliable. Areas of concern relative to accuracy include the possibility of false-negative or uncertain results. Roughly one-third of the participants in this study expressed concern about the anxiety patients feel due to inconclusive results (Alexander et al., 2015). The newer the type of prenatal genetic testing, the more reluctant genetic counselors are to utilize. Most participants from this study noted that patients must have adequate knowledge of the testing, testing limitations, and the opportunity to talk through this information with a genetic counselor (Alexander et al., 2015). Genetic counselors also noted that providing prenatal genetic testing through a blood test, which has no association with miscarriage, may cause more medical professionals and patients to think about prenatal genetic testing. The main limitation of this research is that the data comes from just four genetic centers, so the findings can not be an accurate representation of all genetic counselors.

Due to the nature of genetic counseling practices, issues surrounding disability and how it is perceived are very relevant. Genetic counselors play the role of advocate for the disability community because they work closely with parents who receive test results that reveal their child has a congenital disability. This close tie has raised questions about whether prenatal genetic counselors actively serve as disability community advocates (Farrelly et al., 2012). A study conducted by Farrelly et al. explores how genetic counselors talk about disability in prenatal genetic counseling. This was done by analyzing 93 transcripts from genetic counseling sessions with simulated patients from the Genetic Counseling Video Project. These researchers found that 95% of genetic counselors focused their sessions on the physical aspects of disability. Still, less than 30% had a discussion with their patients about the social aspects of disability (Farrelly et al., 2012). Less than 40% of the genetic counselors asked about the patient's personal disability experiences. When talking with a patient with a pregnancy diagnosed with a disability, 86% of

genetic counselors mentioned termination. In contrast, only 37% said continuing the pregnancy, and 13% mentioned adoption as an option. A limitation of this study is the use of simulated patients for the genetic counseling sessions (Farrelly et al., 2012). This allowed researchers to keep client factors such as age, culture, and beliefs the same but is not reflective of the patient population. To explore this limitation, the researchers had genetic counselors rate whether they felt the session seemed natural, of which 72% agreed that it felt real (Farrelly et al., 2012). Another study limitation could be that patients had a small risk of having a fetus with Down syndrome, so that the results may have varied for a high-risk patient. The researchers from this study concluded that genetic counselors should have deeper discussions with their patients regarding their willingness and ability to parent a disabled child. The researchers felt that this would allow the genetic counseling session to be more aligned with patient values (Farrelly et al., 2012).

The recommendation to have deeper discussions with patients about disability and whether they have the capacity to parent a child with a disability aligns with the common theme that genetic counseling is necessary for patients considering and receiving prenatal genetic testing. The studies above share a common theme that genetic counselors value the patient's right to make autonomous decisions regarding whether to receive NIPT and what to do with the information. The respondents from the studies also believed that NIPT should be used as an advanced screening tool rather than a diagnostic tool. Many of the studies also recommended increased education on NIPT for medical providers such as obstetricians. Genetic counselors also felt that ambiguous results or results about adult-onset conditions could cause increased stress and anxiety for patients (Agatisa et al., 2018; Alexander et al., 2015; Devers et al., 2013; Farrelly et al., 2012; Hercher et al., 2016; Horsting et al., 2013; Markens, 2013).

Other Medical Professionals

Many medical professionals can provide genetic testing other than genetic counselors such as medical geneticists, specialists such as obstetricians and gynecologists, nurse practitioners, primary care physicians, etc. (Faucett et al., 2019). A study conducted with various health professionals, including fetal medicine obstetricians and geneticists, noted that blood test prenatal genetic testing is an attractive alternative to invasive genetic testing methods, as it does not pose an increased risk of miscarriage (Hill et al., 2013). These health professionals also believed that testing should be "highly accurate" and that patients should have the capacity to make an informed choice. The participants expressed concerns about patients not taking the blood test as seriously as an invasive test (Hill et al., 2013). Because blood tests are easy to administer and access, several participants raised concerns that the testing may be trivialized and viewed as a routine test given during pregnancy. This is due to the many other blood tests offered during pregnancy. One participant of the study felt that "people could get on a path that they don't realize the consequences of that path until they're a long way down it (Hill et al., 2013)." The health professionals also felt that NIPT causes an increased pressure to test and terminate and expressed concerns about NIPT possibly providing less information than invasive testing. The benefits these health professionals saw in NIPT included non-invasive properties and early access for testing during pregnancy. Overall, these professionals viewed NIPT very positively and used terms such as "useful," "exciting," "huge impact," and "exciting" to describe it (Hill et al., 2013).

A study conducted on women and health care professionals from nine countries supports the opinions of the health care providers, as noted in the study above. One thousand two hundred

forty-five health care professions participated in this study. Results of the study indicated that healthcare professionals prioritize test accuracy and early testing. In this study, healthcare professionals were more likely to choose invasive testing than the patients, but both groups equally chose NIPT (Hill et al., 2016). It was limited due to questionnaire bias and concluded that healthcare providers and patients were interested in using NIPT as a screening option.

A study conducted by Ngan et al. in 2017 focused on obstetric professionals' perceptions of NIPT. These professionals raised concerns about an increase in earlier detection of Down's Syndrome by NIPT having a possible association with an increase in abortion rates. This study found that obstetricians had a stronger belief in NIPT's usefulness than midwives. Some of the professionals raised the notion that NIPT could provide psychological assurance to women with low risk. The professionals who raised this idea were less concerned about NIPT's ethical issues than other healthcare professionals (Ngan et al., 2017). The healthcare professionals in the private practice sector saw more usefulness of NIPT when compared to the public sector. These results suggest a need for public hospital guidelines regarding how and if a woman should be informed about NIPT as a screening test option. The researchers suggest there should be guidelines for all sectors regarding pre and post-test information on NIPT (Ngan et al., 2017).

Global perspectives on NIPT use were explored by Minear et al. by surveying 49 clinicians from 28 different countries. The majority of the clinicians held M.D. or Ph.D. degrees, though their exact job titles varied due to differences in medical systems between countries (Minear et al., 2015). This study explored how test prices vary between countries and that they can be a significant barrier for patients wanting access to NIPT. This study found that most clinicians have NIPT available in their country and currently offer it. Eighteen of the respondents said that there are plans in their country to induce NIPT into routine antenatal care. The vast

majority of these providers offer post-test counseling for NIPT when a positive result occurs. Only about half of the respondents provide pretest counseling for this form of screening (Minear et al., 2015). The clinicians' general views on NIPT were positive, but they had concerns about the effectiveness, cost, screening protocols, and physicians' education regarding NIPT. This study is limited because it only provides a peek into clinicians from around the world's experiences with NIPT (Minear et al., 2015).

United States obstetricians were surveyed regarding their opinions on general practice NIPT use. This survey found that the obstetricians thought that NIPT is a better screening tool when compared to traditional screening methods and that NIPT should be provided as a recommendation to pregnant women (Brewer et al., 2016). These obstetricians felt that cost, and therefore insurance coverage is the most significant barrier to NIPT. If the patient's insurance provided full coverage, 81% of the obstetricians would provide NIPT for patients as a screening test. Most of the providers who had provided NIPT in a clinical setting felt that patient care was improved (Brewer et al., 2016). Roughly 15% of the obstetricians misunderstood NIPT because they thought it could be used as a diagnostic test. Overall, these obstetricians felt that there was a need for change regarding insurance coverage and practice guidelines in which NIPT is not recommended for women who are low risk. The researchers concluded that continued NIPT education for obstetricians is needed (Brewer et al., 2016).

Bayefsky et al. explored American obstetricians' and gynecologists' ethical views on prenatal whole-genome sequencing (PWGS). This study found that most obstetricians and gynecologists (O.B./GYNs) rely heavily on genetic counselors due to limited genetic knowledge. Considering the clinical adoption of PWGS, the O.B./GYNs would like to be provided with more information and guidance (Bayefsky et al., 2016). The most significant ethical issue was concern

that sharing complex information about a child's genome may heighten parental anxiety and lead to increased costs from overtreatment and concerns for the fetus's future autonomy. The doctors were also concerned that the test might reveal a learning disability or non-medical trait. This survey was limited in its potential for bias because the questions could induce discomfort about PWGS due to explicit reference to ethical concerns (Bayefsky et al., 2016).

The studies above regarding the views of medical professionals demonstrated that, overall, they view the testing positively. These providers felt that non-invasive prenatal testing is better than traditional testing due to the ease of the blood test and zero increased risk of miscarriage. The ease of testing did raise concerns about whether the test would be trivialized because the providers valued informed consent and provided participants with information about the impact of the testing. Another concern raised in many of the studies was the cost of testing and the lack of insurance coverage. With PWGS specifically, discomfort was mentioned due to the potential for increased patient stress and uncertainty about the fetus's future autonomy. In conclusion, these providers felt that there is a need for increased provider education regarding prenatal genetic testing options and stressed the importance of pre and post-test counseling for patients (Hill et al., 2013; Hill et al., 2016; Ngan et al., 2017; Minear et al., 2015; Brewer et al., 2016; Bayefsky et al., 2016).

Disability Community

Because NIPT can screen for genetic impairments, the disability community has varying attitudes about screening. The majority of studies regarding genetic screening and testing have focused on the views of the medical community. Still, it is important to consider other opinions. Few studies have been conducted regarding the attitudes of people with disabilities on prenatal

screening and diagnosis (Chen and Schiffman, 2000). Prenatal screenings can now be used for fetal aneuploidy, certain types of genetic deafness, spinal muscular atrophy, and even the genome as a whole. While parents are valuable stakeholders in genetic screening debates, people with disabilities have lived experiences and knowledge regarding their condition (Boardman et al., 2018).

From 1995-1996 a study was conducted at Northwestern Memorial Hospital in which 15 individuals were interviewed over the phone. The participants were chosen because they had a physical disability, although not necessarily genetic, were of childbearing age, and had some understanding of prenatal diagnosis and genetic counseling (Chen and Schiffman, 2000). The five-part questionnaire included demographic data, participants' views on genetic counseling, personal experiences, eugenics and prenatal diagnosis, disability in general, and self-esteem. The results indicated that participants had favorable genetic counseling views (Chen and Schiffman, 2000). Ten of the study participants had previously received genetic counseling. All of them felt that it is helpful for others, and 80% of them said that genetic counseling was moderately beneficial. 80% viewed prenatal diagnosis positively, while the other 20% had unfavorable views due to their opposition to abortion (Chen and Schiffman, 2000). The researchers found that much of the opposition to prenatal testing was based on the incorrect assumption that the only option is to terminate the pregnancy if the fetus is found to have a fetal abnormality. Nearly all of the study participants did not view genetic testing as eugenics but instead felt the testing increased knowledge and promoted health. The small sample size is this study's main limitation. More extensive studies should be conducted to see if these views are consistent with larger numbers of people with disabilities (Chen and Schiffman, 2000).

A study was conducted specifically addressing the views of people with Spinal Muscular Atrophy (SMA). The types of genetic testing for SMA are whole genome sequencing and NIPT. The three genetic screening programs for SMA are prenatal, newborn, and preconception screening (Boardman et al., 2018). There are four main subtypes of SMA, which vary in age of onset and severity. SMA type I is the most severe SMA form, which usually results in death before 18 months of age. SMA type II is considered the intermediate form with a lifespan near-normal with medical support, though complications and respiratory infections can lead to premature death. In SMA types III and IV, there is usually a deterioration in the affected person's abilities over time. SMA IV is not diagnosed until adulthood (Boardman et al., 2018). Using a survey and qualitative interviews, researchers found that people with Type II SMA with more severe symptoms and earlier-onset symptoms tended to view SMA more positively and prenatal screening for SMA more negatively than people with later-onset, milder forms of SMA such as Types III and IV. Aligning with this, people with Types III and IV tended to support all forms of screening for SMA. People with Type II SMA favored newborn screening over prenatal screening, which is not surprising given their more positive attitudes towards SMA (Boardman et al., 2018). While fewer people with Type II SMA favored prenatal or preconception screening than other SMA types, 53% felt prenatal screening and 63% felt preconception screening should be an option for the general public. People with Type I SMA were not included in this study as they do not live long enough to reach adulthood. A limitation of the study is there was nothing to prevent individuals from taking the survey multiple times since no identifiable data was collected due to confidentiality and data protection issues. Finally, an unforeseen source of bias was the parental status of individuals with SMA who completed the form. A higher number of parents in Type III and IV groups could have changed their prenatal testing views (Boardman et al., 2018).

Middleton et al. conducted a study of Deaf adults' attitudes regarding genetic testing for deafness. More than 60 genes have been discovered to cause nonsyndromic and syndromal deafness, so it is possible to use prenatal genetic testing to screen for deafness-causing genes (Middleton et al., 1998). Deafness is an important impairment to study because, for many Deaf individuals, it is their cultural identity. Therefore, some individuals view deafness from a sociological perspective. This leads many culturally Deaf people to be proud to be Deaf and see Deafness as a significant part of their identity (Middleton et al., 1998). Eighty-seven people who attended the conference "Deaf Nation" participated in this study. All of the participants identified as either deaf or hearing impaired, with the majority identifying as deaf. 82% did not tick any of the positive words on the questionnaire when asked how they felt about the discoveries in genetics regarding deafness. Over half of the participants felt that genetic testing would do more harm than good, and only 16% of individuals said they would consider having prenatal genetic testing for deafness (Middleton et al., 1998). Almost 30% of the participants said they would prefer to have children who are deaf. Overall, participants viewed prenatal genetic testing negatively.

When examining the views of the disability community, it is essential to consider the opinions of family members. Friedman and Owen published a study in the Disability Studies Quarterly regarding sibling attitudes on prenatal genetic testing. This study found that siblings of people with disabilities have a broad range of beliefs regarding prenatal genetic testing. On average, participants believed that prenatal testing should be used to screen for disability but that they were unlikely to use prenatal screening themselves in the future (Friedman and Owen, 2016). There was a relatively weak correlation between a participant's likelihood of using prenatal testing and their views on whether this testing should be used for disability. Through

open-ended questions, the researchers found that participants viewed genetic testing as a way to prepare for having a child with a disability by having more knowledge about the disability. Some participants regarded this preparation and knowledge as a negative instead of a positive because it could lead to abortion (Friedman and Owen, 2016). Participants then explained how complicated they felt the prenatal genetic testing issue is due to their belief in choice, parental autonomy, and personal concerns regarding pregnancy termination. This data concluded that siblings of people with disabilities have a nuanced understanding of prenatal genetic testing and disability but do not necessarily predict sibling attitudes. Bias is present in this study due to participant self-selection, and the subjects were all white women.

As the above studies show, the views of people with disabilities about prenatal genetic testing vary widely. These views range from believing that genetic testing and genetic counseling are beneficial and would help parents to over half of deaf individuals thinking that genetic testing would do more harm than good. This range of views was also demonstrated by people with SMA who are more likely to have positive views if they have a late-onset, less severe form of SMA. Regardless, more than half believed prenatal genetic testing should be available to the public. Siblings of people with disabilities echoed this by expressing a range of viewpoints but felt strongly about parental autonomy and availability of screening for those who want it (Chen and Schiffman, 2000; Boardman et al., 2018; Middleton et al., 1998; Friedman and Owen, 2016).

Parents

Parent perspectives on prenatal genetic testing are important to explore and consider. NIPT has been primarily used to screen for fetal chromosome abnormalities such as Down syndrome. The parents of children with Down syndrome expressed their opinions and attitudes in

a study conducted by Schendel et al. This study conducted individual interviews and focus groups with Dutch parents of children with Down syndrome (van Schendel et al., 2017). Earlier testing, accuracy, and safety were noted as NIPT advantages. While participants raised concerns about NIPT's practices in screening for Down syndrome, they acknowledged the benefits for people to know and to prepare for having a child with Down syndrome. A fear that the parents acknowledged was an increase in abortions of fetuses found to have Down syndrome (van Schendel et al., 2017). This idea also correlated with the notion that NIPT could lead to a decrease in facilities and a loss of societal diversity and acceptance for people with Down syndrome, which could increase decisions to undergo the screening. The participants expressed a need for accurate and balanced information and personal counseling (van Schendel et al., 2017). Increasing testing to cover more syndromes could take some of the focus away from Down syndrome, but where to draw the line was a concern. Parents described their concerns as a circle where NIPT availability leads to an increase in screening, which leads to more abortions, which leads to fewer people who have Down syndrome, which leads to fewer facilities and less acceptance for people with Down syndrome, and which leads back to increased NIPT (van Schendel et al., 2017).

Another critical parent perspective to consider is pregnant women and their partners. Focus groups and interviews were used to conduct this research (van Schendel et al., 2014). The most significant disadvantages the participants identified were the risk of miscarriage from follow-up diagnostic tests and uncertain results. The NIPT characteristics of safety, accuracy and early testing should be considered to decrease these disadvantages. Participants in this study noted that NIPT's scope should be limited, which could avoid testing for minor abnormalities (van Schendel et al., 2014). The increase in prenatal testing scope could make it challenging to

draw the line for patients but could be beneficial in screening for severe disorders. The less concern about using prenatal screening tests, the greater the possibility for individuals to feel pressure to test. Information that is necessary to participate and to make informed decisions voluntarily should be provided (van Schendel et al., 2014).

A similar study conducted by Farrell et al. used focus groups to access pregnant and new parent perspectives regarding NIPT. This study found that NIPT's main advantages are early timing, testing ease, accuracy, and determination of fetal sex compared to other screens (Farrell et al., 2014). Subjects noted that the non-invasive nature of NIPT is an advantage. NIPT's main disadvantages were false-negative and false-positive results, cost, insurance coverage, and anxiety. The most unlikely women to undergo NIPT were women who did not want information regarding fetal aneuploidy (Farrell et al., 2014). The participants in this study valued their ability to make private, informed, and autonomous NIPT decisions. Overall, this study highlighted the advantages and disadvantages pregnant women listed about NIPT.

A study conducted in the United Kingdom surveyed 1,131 women and partners and found that 95.7% of respondents viewed NIPT as a positive development in the field of prenatal care. 88.2% of respondents felt that they would use this screening method (Lewis et al., 2014). 70% of the participants felt an essential NIPT attribute is the baby's safety. Interestingly roughly the same number of respondents said that they were likely to terminate a pregnancy affected with trisomy (30.7%) compared to respondents who were not likely to terminate (36.5%) (Lewis et al., 2014). Because the survey respondents overwhelmingly felt positively about NIPT, the test uptake will likely be high. Women who recently chose to decline the test indicated that they would consider NIPT, so the uptake may be higher than expected for this group. Uptake may also be higher than previously expected for women who only want the test for information rather

than termination decisions. Finally, it is essential to have pretest counseling provided to ensure that the women understand the difficulty and the implications of its possible results (Lewis et al., 2014).

NIPT is widely used to screen for fetal aneuploidies. Scientifically, NIPT could be used to sequence the entire fetal genome. This type of testing is referred to as non-invasive prenatal whole-exome sequencing. A survey of 553 pregnant women found that most women want information about treatable childhood-onset severe conditions, but fewer are likely to want information regarding non-medical traits (Sullivan et al., 2020). The most common reason that they wanted prenatal whole-genome sequencing was to prepare for a child with special needs medically, psychologically, and financially. Some women didn't want to receive genetic information because they would not consider aborting the fetus and did not want the increased stress that data could cause (Sullivan et al., 2020). Almost half of the women wanted clear recommendations from their clinicians about which categories of data to test for, and they wanted all options presented. These results showed that, generally, pregnant women wish to receive genetic results regarding medical conditions in order to prepare. Over half of the women wanted their providers' recommendations regarding what information to obtain from whole-genome sequencing (Sullivan et al., 2020).

NIPT can be used for many different applications, including whole-genome sequencing. This presents a host of ethical dilemmas for pregnant women. Bowman-Smart et al. conducted a study to explore the views of women who have previously undergone NIPT. They explored women's ideas on using NIPT to screen for adult-onset, neurodevelopmental, and psychiatric conditions (Bowman-Smart et al., 2019). Women expressed interest in using NIPT to screen for conditions that have an onset earlier in life and that severely impact a person's quality of life.

They stated that test accuracy was critical. Using the screening for non-medical traits raised concerns for them (Bowman-Smart et al., 2019). Also, they indicated that pregnancy termination was not their sole rationalization for receiving NIPT. 71.3% of the women supported NIPT's use to test for deafness, and 67.8% were interested in using this testing themself. Screening for non-medical traits raised concerns for the women participating in this study (Bowman-Smart et al., 2019).

The studies above show that parents of children with Down syndrome believe that NIPT's advantages are test accuracy, safety, and the early timing of the testing. These parents valued parental autonomy and the right to know their fetus's genetic status. This group raised concerns regarding abortion and believed this could lead to a decrease in social diversity and acceptance of people with disabilities. Pregnant women considering NIPT saw the same benefits, valued the test's information, and the opportunity to make independent decisions. The concerns raised were about cost, false results, and anxiety. Over 95% of women in the U.K. viewed NIPT positively, and roughly 90% said they would use this testing themselves. Pregnant women expressed interest in genetic testing for treatable childhood-onset conditions but not for non-medical traits. They felt this testing could help them prepare for having a disabled child. Overall, this testing was viewed positively, despite concerns, due to the beliefs about parental autonomy (van Schendel et al., 2017; van Schendel et al., 2014; Farrell et al., 2014; Lewis et al., 2014; Sullivan et al., 2020; Bowman-Smart et al., 2019).

Disability Activists/Scholars

Disability studies is the study of disability through the lens of society, culture, and politics instead of through psychology or medicine. This field acknowledges that disability is a fact

rather than an exception to the human experience (Rice, 2018). People who focus on disability studies are referred to as disability scholars. Disability activists organize and advocate for the equality of people with disabilities and make up the disability rights movement, which began in the 1960s (Meldon, 2019). The perspectives of disability activists and scholars are essential when looking at genetic testing views because these individuals approach this subject with a very different conceptualization of disability/impairment than the dominant societal narrative (sociopolitical view vs. medical/individual view).

Chris Kaposy authored a disability critique of prenatal testing for Down Syndrome. In his analysis, Kaposy argued that people who have Down Syndrome, and the people close to them, oppose developments in prenatal testing due to bias against people with cognitive disabilities, which influences an individual's decision to use genetic tests and leads to termination (Kaposy, 2013). This critique continues to present the arguments that people living with Down syndrome are negatively affected by bias. Selective termination of a fetus diagnosed with Down syndrome does not cause bias in itself but is a product of discrimination. Finally, a consequence of bias is high rates of termination, so it is justified to object to prenatal testing. Kaposy concluded that an increase in understanding and awareness of Down syndrome and prenatal genetic testing is important (Kaposy, 2013). Women must be well informed about what life is like with Down syndrome. This information should not be exclusively centered around cognitive deficits or medical problems. It is crucial for prenatal testing not to be an aspect of routine prenatal care but rather an intentional and informed decision to counteract bias (Kaposy, 2013).

Parens et al. published a disability rights critique regarding ethics, reproduction, and prenatal testing. The social context involves discrimination against people with disabilities, the multiplicity of prenatal diagnosis attitudes and disability traits, and reproductive freedoms

(Parens et al., 2002). The ethical arguments include whether it is morally problematic to perform prenatal genetic testing and whether misinformation is a basis of prenatal testing. A central argument of disability scholars regarding prenatal testing is that there is misinformation in society regarding the lived experience and impact on family. The disability rights critique claims that prenatal genetic testing would not be a need if people with disabilities were ultimately integrated into society. This critique concludes by explaining that the arguments presented are not meant to justify prenatal genetic testing wholesale restrictions (Parens et al., 2002). Alternatively, these arguments should allow prospective parents to take time to think about their decision and for professionals to help individuals examine their decisions. These arguments should also help people to be more informed and thoughtful when making decisions. As prenatal genetic testing technology continues to advance, critical conversations are needed, and disability rights arguments are a valuable resource for these conversations (Parens et al., 2002).

Parens and Asch presented reflections and recommendations on their previous disability rights critique about prenatal genetic testing described above. In this paper, they reflected on morality, misinformation, attitudes, selective abortion, and discrimination (Parens and Asch, 1999). While the authors of this paper could not reach a consensus regarding where to draw the line between reasonable and unreasonable prenatal genetic tests, they did agree that testing for uniformly fatal conditions was morally defensible. Disability activists raised concerns about the inability to form a consensus, indicating it could lead to increased testing, attempts to screen out specific attributes, and increased intolerance for human diversity (Parens and Asch, 1999). The scholars were able to agree that genetic counselors should help their clients make informed decisions about how to use test results. They also agreed that patients should be informed about programs that help prospective parents obtain information about the lived experience of

disability (Parens and Asch, 1999). They also argued that disability education should be integrated into medical genetics, genetic counseling, and obstetrics programs. This could help combat the ignorance regarding disability, one of the primary sources of discrimination against people with disabilities (Parens and Asch, 1999).

Martha Saxton's piece called "Why Members of the Disability Community Oppose Prenatal Diagnosis and Selective Abortion," includes a powerful quote by Deborah Kaplan, who states, "If persons with disabilities are perceived as individuals who encounter insurmountable difficulties in life and who place a burden on society, prenatal screening may be regarded as a logical response. However, suppose persons with disabilities are regarded as a definable social group who have faced great oppression and stigmatization. In that case, prenatal screening may be regarded as yet another form of social abuse (Saxton, 2001)." This explains the disability studies argument against prenatal genetic testing - negative attitudes toward people with disabilities influence decision-making regarding pregnancy and enforces the disability stereotypes held by the general public. While the American public seems to have accepted that prenatal genetic testing is a good thing, disability scholars argue that they came to this conclusion based on misinformation, including the idea that disabled people have a lower quality of life, that having a child with a disability is undesirable, that the burden of raising a disabled child could be avoided by selective abortion, and that a society should be able to decide who should and should not be born (Saxton, 2001).

The Disability Studies Quarterly published an article by Guillemin et al. that explores deafness, current technologies, and genetic testing for deafness. Nineteen individuals from South Eastern Australia were interviewed (Guillemin et al., 2005). Sixteen of the study participants supported newborn and prenatal genetic testing for deafness. Some participants supported

prenatal screening for hereditary deafness for those who want it, even though they would not use this screening themselves. Limitations of this study included a lack of insight into the respondents' deliberation process or critical engagement. Upon further investigation, it was found that participants identified beneficial factors of genetic testing and then expressed their concerns (Guillemin et al., 2005). The benefits mentioned were early and definitive diagnosis, parents' ability to plan, prepare for the child's school and language needs, early intervention opportunities, medical intervention, and assist in developing a possible cure. Participants who identified as Deaf were less likely to bring up the idea of a cure as a potential benefit but were interested in parents' opportunities to prepare for a child who is deaf (Guillemin et al., 2005). The main concern mentioned was the possibility of pregnancy termination resulting from a positive prenatal test for deafness. This concern stemmed from personal beliefs about abortion and the idea that terminating a deaf fetus insinuated that a deaf child is worth less than a hearing child. Another problem identified was that parents who find out they are pregnant with a deaf child are not provided with enough information. This study was limited because of its size but found no significant difference in the views of participants who identified as Deaf, hearing impaired, or deaf. Overall, these individuals felt pessimistic about the idea of terminating a pregnancy due to prenatal genetic testing. Still, the individuals advocated strongly for parental autonomy (Guillemin et al., 2005).

Disability scholars and activists present the most negative views on prenatal genetic testing of any of the groups studied. This was based on the belief that prenatal genetic testing is founded on bias against people with disabilities. Disability scholars objected to this form of testing due to the possibility of bias leading to pregnancy termination. It was mentioned that this bias could stem from misinformation regarding what it means to live with a disability. The

disability scholars suggest that there should be an increase in awareness of disability in the early stages of testing and that these conversations should not solely focus on deficits. This would allow parents to take time to critically consider their decision on whether to test rather than make a decision without forethought. Deaf scholars acknowledge that they did not support testing in order to obtain a cure but rather as a way for parents to prepare for a child who is deaf. These scholars raised concerns about eliminating deafness. They stated that professional programs should include disability education. Their challenge was reaching a consensus on where to draw the line when it comes to prenatal genetic testing (Kaposy, 2013; Parens et al., 2002; Parens and Asch, 1999; Saxton, 2001; Guillemin et al., 2005).

Discussion

The literature review demonstrates that genetic counselors and other medical professionals view genetic testing positively and value parental autonomy. Both groups also argue for increased provider education and pre and post-test counseling for patients. These groups also view NIPT as better than traditional invasive testing. The genetic counselors stress that NIPT should be used as a screening tool rather than a diagnostic test. The primary concerns are stress on individuals due to ambiguous test results, testing being trivialized, and the cost of testing due to a lack of insurance coverage. The ethical concern of these stakeholders is that using testing for adult-onset conditions could lead to the loss of autonomy of the fetus to make these decisions in the future.

The views of parents echo those of the medical professional groups in that they value parental autonomy and NIPT's safety, accuracy, and early test timing. The parents of children with Down syndrome express uneasiness regarding the abortion of fetuses that test positive for

chromosome abnormalities. This raises the concern that an increase in abortion rates could decrease social diversity and societal acceptance and programs for people with disabilities. Pregnant women also view NIPT positively due to the same benefits described above and value the information testing could provide. This group feels particularly strong about the ability to make non-coerced autonomous decisions regarding testing. These parents have the same apprehensions about NIPT as the medical providers in that testing could cause increased anxiety relative to false results and cost. Many women view NIPT as help for them to prepare for having a child with a disability and prefer testing for treatable childhood-onset conditions over adult-onset conditions. This view matches the view of medical professionals.

The views of the disability community vary widely between and among disability groups. These views range from positive, that genetic testing is helpful, to negative, that genetic testing would do more harm than good. About half of people with disabilities state that genetic testing should be available to the public. Siblings of people with disabilities also have varied views and echo the belief in parental autonomy and that testing should be available to women who want it. Interestingly, the disability communities' viewpoints vary from the perspectives of disability scholars and activists. Disability scholars generally oppose prenatal genetic testing in that it is based on bias against people with disabilities and that this bias leads to pregnancy termination. They also note that this bias could stem from misinformation surrounding disability and the disability experience. Additionally, they discuss that this could be solved by increasing disability awareness in the early stages of testing, thus allowing parents to make informed decisions. The concerns of this group align with the apprehensions of parents that prenatal genetic testing could lead to the elimination of disability and a decrease in social acceptance. This group feels strongly that professional programs, such as genetic counseling and medical schools, should provide

disability education in their curriculums. Overall, the scholars did not form a consensus about where to draw the line when it comes to prenatal genetic testing.

From these results, it is clear that medical professionals and genetic counselors view prenatal genetic testing primarily from a medical model of disability and in a positive light. In contrast, disability scholars view this testing from a social model of disability and in a negative light. These groups do agree on increasing education for providers on testing, disability, and the disability experience. All of the groups value parental autonomy and the assurance that pregnant women are not coerced into testing. These results are significant because they show that, while the groups did not share the same views on genetic testing, all agree on transparent, actionable ideas to improve prenatal genetic counseling. Another significant finding of this literature review is that the views of disability scholars and the disability community are not the same. The opinions of the disability community are more diverse than expected. These differences could be due to whether people view disability from a medical model or social model perspective. This is demonstrated by people who identify as culturally Deaf, as they have the most negative views on prenatal genetic testing. In contrast, people with late-onset SMA are more positive about testing and view their disability as a medical condition and not due to societal oppression (Guillemin et al., 2005; Bowman-Smart et al., 2019; Boardman et al., 2018).

These results are limited to thirty studies included in the literature review. These sources primarily cover U.S., U.K., and European viewpoints and may not be representative of world perspectives. Many of these studies were conducted as surveys or interviews, which may be biased based on the participants who chose to complete these studies. The viewpoints of people who do not feel strongly one way or another about prenatal genetic testing may be missing from these results. Survey and interview-based research studies also can be biased based on the kind

of questions asked and the way they are asked. These results do not tell us the individual views of society or the personal views of individuals from groups covered. While this literature review has limitations, the research conducted provides an overview of stakeholder attitudes regarding prenatal genetic testing. It opens the door for additional research in this area.

Conclusion

This literature review's purpose is to gain an understanding of different stakeholders' views on prenatal genetic testing. It is clear from the research reviewed that genetic counselors, other medical professionals, and parents view prenatal genetic testing positively based on parental autonomy and the safety and accuracy of NIPT. The views of the disability community varied widely, and disability scholars viewed this testing negatively overall due to moral concerns regarding the elimination of disability through abortion. Concerns regarding an increase in abortion rates due to NIPT were raised by all stakeholders researched. This research is significant because it allows for insight into how to best approach NIPT. By understanding the wide range of views regarding this testing, prenatal genetic counselors can tailor their approach to best counsel these groups. While the stakeholders did not share the same views regarding prenatal genetic testing, these results are notable because they show that all groups agree on transparent and actionable ideas to improve prenatal genetic counseling. These ideas include a need for increased provider education regarding prenatal genetic testing options, the importance of pre and post-test counseling for patients, and disability education in professional programs. Additional research is needed to determine how to best implement disability education into professional programs and on the views of groups not covered in this paper. Overall,

understanding different stakeholders' perspectives on prenatal genetic testing provides a framework for how to best approach genetic counseling for NIPT.

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