Genetic Screening: An Ethical Viewpoint

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Genetic screening has been having a growing interest in the United States, and worldwide. It allows the mother and family to test the genetic makeup of a fetus for possible genetic anomalies and hereditary conditions. This practice was being used in the United States as early as the latter half of the twentieth century (Luque-Bernal & Buitrago-Bejarano, 2017). This can be done through many types of testing, including the widely used amniocentesis, and others such as chorionic villus testing, and cell free DNA sampling (cfDNA). With the knowledge an expectant mother gets with this testing, she can decide if she wants to terminate the pregnancy if she wishes. There are many factors that determine a woman's decision to get genetic screening, including cultural and societal norms, autonomy, and potential harms.

It is important to examine the advantages and disadvantages related to the use of genetic screening, and have the ability to inform an expecting mother so she can make a decision of whether or not to undergo genetic screening testing, and based on the results, if she wants to terminate the pregnancy. It is also important to inform a mother of the types of prenatal testing available and the possible risks associated with each. The most well known is the amniocentesis, in which a needle is inserted into the uterus and extracts amniotic fluid, which is then tested for abnormalities. The chorionic villus sampling is another invasive technique, but can be done earlier than an amniocentesis. This tests the chorion, the fetal side of the placenta, by removing a small piece via a needle. The final often used invasive technique for genetic screening is the percutaneous umbilical blood sampling, in which a needle is used to draw blood from the umbilical cord, which contains the genetic makeup of the fetus (Lowdermilk, Perry, Cashion, & Alden, 2016). A noninvasive technique which is prevalent in the literature is the cfDNA

screening, which compares the DNA in maternal blood, can be used to test Rh factor and suggest possible trisomies, but it is not a diagnostic test compared to the invasive screenings (Luque-Bernal & Buitrago-Bejarano, 2017).

A woman's culture and society can have one of the largest impacts on care and the decision to undergo genetic screening. In surveys completed by Latina and non-Hispanic White women, the Hispanic women were less likely to have the testings done, especially when their preferred language was Spanish because of held cultural beliefs (Floyd, Allyse, & Michie, 2016). On the other hand, women of East and Southeast Asian descent were more likely to follow the trends from such countries. For instance, Chinese women were more likely to undergo screening similar to what the Chinese government encourages them to use, while countries with strong religious trends, such as Malaysia and the Philippines, were less likely to use screening due to the possibility of termination of pregnancy (Tsai et al., 2017). It is important to be aware of a woman's culture and nurses and other health professionals must be ready to understand and accept cultural implications.

American society is currently in a heated debate about the legal and ethical aspects regarding abortion, which plays into genetic screening. After positive results for a genetic abnormality, a woman is often given the decision to either continue or terminate the pregnancy, so society's views of abortion play a vital role in this step of the process. The ability to terminate the pregnancy given positive test results, however, has led to some believing this is a stepping stone to eugenics.

This is from the belief that women would terminate their pregnancy if they were to have positive results for a genetic anomaly. Some have a fear that the ability to terminate this type of

pregnancy could cause harm to the people currently living with such a disability. For example, one of the commonly tested genetic traits is for trisomy-21, Down syndrome. Once a mother learns the infant will have Down syndrome, she is given the choice to terminate, but some believe if this mother terminates this pregnancy, it will harm the community living with Down syndrome by stigmatizing the disability further. Many women, when given a positive genetic screen for trisomy-21, will decide to have a therapeutic abortion, which makes the community of those living with Down syndrome smaller (Floyd et al., 2016). On the other hand, some think it is not technically eugenics, due to their definition of eugenics. Eugenics is a state-sponsored event, according to Luque-Bernal and Buitrago-Bejarano, so it would only be considered eugenics if the government decided if the woman was to keep the child, rather than this being a decision for the woman and her partner (2017).

Society's views may seem to largely oppose the use of genetic screening, but it can have a highly important benefit. Once a woman has undergone genetic screening of the fetus, if an abnormality is discovered, she has the option to continue with the pregnancy. The ability to prepare can have an impact on the woman in the postpartum period. It has been found parents of infants with congenital abnormalities are more susceptible to postpartum depression (Cole, Olkkola, Zarrin, Berger, & Moldenhauer, 2018). By acknowledging this, it can be easier for a parent to receive treatment, and nurses can pay close attention to these women. Postpartum depression has been gaining awareness as of late, and society being vigilant about it can benefit these mothers.

Although cultural and societal pressures can play a major role in a woman's decision to undergo prenatal testing and possibly terminate a pregnancy based on the results, it is her

decision and it is important to maintain the autonomy of the mother. A woman may decide to undergo this testing in order to be able to prepare. Closely related to the ability to prepare for possible difficulty coping is the ability to prepare for a child with a disability. If a woman chooses to continue the pregnancy, she can prepare for the added financial costs, and can give her a feeling of control over herself and her pregnancy. It also allows women with a background with children with disabilities the autonomy of choosing to keep the child or not.

During the survey to determine the likelihood of terminating a pregnancy if the fetus was found to have a genetic defect, three of the respondents had direct experience with children with such disabilities. One woman had an autistic son, and stated her desire to undergo testing in order to know, but she was still undecided if she would have kept a fetus or terminate the pregnancy. The other women wanted the choice to undergo the screening due to their experience in pediatric healthcare and did not wish to cause pain to a potential child. All three women had experience and knowledge about the subject, so they felt prepared to make an informed decision if they were to be put into such a situation (Floyd et al., 2016).

The autonomy of the mother should be taken into account, but the mother is likely to have several reasons to either do the screening or not, specifically the disadvantages of both sides. One may refuse genetic screening because of potential harm to the fetus, a possible negative pregnancy experience, and the costs associated with the testing. On the other hand, the costs of a child with a disability may be taken into account by a woman having a genetic screen.

When a woman decides to undergo genetic screening, she is informed of the possible risks associated with whichever testing she chooses to use. With the use of invasive testing, such as the amniocentesis, chorionic villus sampling, or percutaneous umbilical blood sampling, there

is a greater risk of injuring the fetus. The risk of a spontaneous abortion during invasive screening techniques such as these is between 0.5 to 2% of those tested (Luque-Bernal & Buitrago-Bejarano, 2017). This is due to difficulties with the needles used typically and improper placement of such needles, but the risk is an important factor for many women. For this reason, a mother may be fearful to have genetic screening performed, including when they are at a higher risk of possible genetic anomalies. The use of noninvasive genetic screening tests may seem a better decision, but they are not diagnostic, and provide a limited amount of knowledge, with the possibility of inaccurate results (Floyd et al., 2016).

An expecting mother can have a negative pregnancy experience if she has positive results from genetic screening. The possibility of harm during an invasive procedure may cause stress to the mother, but the highly reported reason behind a possible negative pregnancy experience is the mother's worry about her fetus once receiving positive results. The knowledge of genetic abnormalities is a double edged sword, it allows the family to prepare for a child with a disability, but the mother also worries about the fetus's health and future. One woman stated she would not have wanted to have a wide array of diseases tested for because she felt it would give her more reason to worry, rather than feeling well-informed (Floyd et al., 2016).

The cost of these screenings can be expensive, and depending on a person's income and socioeconomic status, it may be a barrier to care. The cost of genetic testing can range anywhere from $100 to greater than $2,000, which is a significant financial strain on expecting mothers (U.S. National Library of Medicine, 2018). The costs of the testing may also not be covered by insurance. Most insurance companies assess the risk of a couple for genetic anomalies, and based on the risk will cover partial costs, but they do not typically cover the entire cost. During

interviews, most women were not aware of the costs of the genetic screenings, and were guessing lower out of pocket costs than the actual amounts, even with insurance coverage (Floyd et al., 2016).

The costs of genetic screening can be a major deterrent to using them, but it has been proven to help women in financial strain to have the testing done. Some women have the testing done in order to decide if they can provide for the child. Many women undergoing the screening do not have the financial resources necessary to provide for a child with cognitive disabilities. By utilizing genetic screening, a woman is able to decide if she can provide for a child, and if she cannot, she has the ability to terminate the pregnancy. In the long run, the cost of the therapeutic abortion and testing is significantly less than the costs associated with a child with disabilities. In a 2000 study, the projected costs of removing the choice of a therapeutic abortion in Michigan alone would be an average of $74 million per year in added healthcare system costs. In the United States as a whole, it would be about $2 billion (Luque-Bernal & Buitrago-Bejarano, 2017). The choice of a mother to terminate a pregnancy based on genetic screening can eliminate the cost and strain on the healthcare system that would be used if the child was born.

It is important to acknowledge the many factors in a woman's decision to undergo genetic screening and possibility of terminating a pregnancy with positive test results. The three main factors researched are the societal pressures, the need to maintain autonomy, and the harms and risks associated with these decisions. It is important to address cultural concerns on both sides, the importance of society's view on abortion, and the belief that such genetic screening can lead to eugenics. The autonomy of the woman is one of the major benefits of performing the genetic screening tests. They allow for the woman to make informed decisions regarding her

pregnancy and the care she is provided. The autonomy of choice also gives the woman the opportunity to prepare for a child with a genetic or hereditary condition, but it is important to help the woman understand and cope with this news, rather than allowing the information to become a burden. It is important to adjust care so the woman is given opportunities to cope so the information does not negatively impact her pregnancy experience. The final factors are the costs associated with the screenings. The cost of the screening itself may be a barrier to care, but some women view the cost of the screening is outweighed by the potential costs of care for a child with a disability. At the end of the day, it is the role of the nurse and healthcare team to educate and inform the woman in order to allow her to make a well-informed decision.

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7/22/2018