

Is Genetic Screening Before Birth A Good Idea?

Genetic screening has become more available as time progresses, meaning more expectant mothers can get their unborn children screened for possible disorders. There are various tests that can be done to see if a child has or is at risk for certain genetic disorders or birth defects. Some of these tests are blood tests, non-invasive prenatal testing (NIPT), amniocentesis, chorionic villus sampling, nuchal translucency, and preimplantation genetic diagnosis (PGD). There are some perks of getting an unborn child screened for disorders as it allows the parents to know if their child will need extra support in the future, or if they need to terminate their pregnancy. There are also some downsides to genetic screening as there are possibilities of false positives and false negatives.

The tests available to intrauterine pregnancies are blood tests, non-invasive prenatal testing (NIPT), amniocentesis, chorionic villus sampling, and nuchal translucency. The first prenatal tests are blood tests which test for signs of certain conditions such as Trisomy 18, Down Syndrome, and neural tube defects like spina bifida. Blood tests can give false negatives or positives for any disorder. Non-invasive prenatal testing (NIPT) is a highly effective test that can detect Down Syndrome and other genetic abnormalities. It is a blood test that measures the cell-free fetal DNA that can be found in the mother's blood. Next, there is amniocentesis which is a test done using the amniotic fluid. A small amount of amniotic fluid is taken out of the uterus with the use of a needle inserted in the abdomen, with the assistance of a camera. The fluid is then tested for genetic abnormalities. If the test comes back positive for a genetic abnormality the parents have the choice to terminate the pregnancy. This test does have a low risk of miscarriage (0.5% - 1%). There is a chorionic villus sampling which is a test that takes a small piece of the placenta tissue. The tissue is extracted from the abdomen or cervix and is tested for genetic abnormalities. It can be done between ten to thirteen weeks of pregnancy. This test also has a risk of miscarriage (about 1-2%). The last prenatal test that can be performed is called nuchal translucency. This is a test to see if the fetus has a risk and is not a diagnosis. This test is done by getting a sample of fluid from underneath the skin of the fetus' neck. The fluid is sampled to measure the thickness of the fluid, the thicker it is the more likely the fetus could have Down syndrome or other genetic anomalies.

There is a test that is not done for intrauterine pregnancies but rather for in vitro fertilization (IVF) and it is known as preimplantation genetic diagnosis (PGD). It is an alternative form of prenatal diagnosis done through the embryo before it is placed inside the uterus. It was originally offered to couples who were at risk for single gene disorders such as cystic fibrosis, Tay-Sachs, Huntington disease, and muscular atrophy. PGD has been used frequently to assist with reproduction detection of chromosome aneuploidy from advancement in maternal age or structural chromosome rearrangements. It is not performed on an ongoing intrauterine pregnancy in the late first or early second trimester as the pregnancy had already begun. The main positive thing about preimplantation genetic diagnosis is that it is very accurate

and can be done early on in the pregnancy to detect a genetic or chromosomal condition before birth. The negative thing about this testing is that it is only carried out only if there is a genetic change for a specific condition that has been found in one or both parents through earlier testing. It is intended only to detect only disorders and the parents may find a condition that is unrelated to family history. The costs for preimplantation genetic diagnosis is added to fertility treatments and it depends on the test that is done. It does not guarantee that the embryo will implant correctly or if there will be a full term pregnancy therefore a miscarriage can still occur.

Genetic screening before birth is important as it allows the parent(s) to see if they need to have additional support for their child and ensure their child did not inherit a genetic disorder from one or both parents. This is good for parent(s) if they choose to keep the child if the test comes back positive to get additional support as some children with disorders require a lot more attention and assistance. For example, cystic fibrosis (CF) which is an inherited disease where the body makes very thick and sticky mucus causes problems in the lungs, pancreas, and other organs. Those with CF are more prone to breathing problems and infections in the lungs. Parent(s) can get additional help by having a care team to help them with treatments, therapies, etc. Parent(s) of children with CF can also learn more about treatment plans and how to teach their child self-care for this disorder. Genetic screening before birth is also beneficial for parent(s) who have a genetic disorder themselves to ensure it was not passed down to their unborn child. This is beneficial as some parent(s) will stress out that they have already reduced the quality of life for their child by having a genetic disorder. Therefore, genetic screening before birth is beneficial for the parent(s) and child as there can be additional support and ensured quality of life.

There are reasons that genetic screening should not be done before birth as it can cause emotional distress, the number of false positives and negatives, as well as how it gives limited information. First, this test can cause emotional distress for the parent(s) of the unborn child as some people may be angry, depressed, or guilty with the results they have gotten. Next, it is prone to giving false negatives and false positives. This is not good when it comes to life as a false positive can result in a termination of a perfectly good fetus or in the case of a false negative the birth of a child who will have a lower quality of life. Finally, the tests can only grant a limited amount of information. This means that the test is for certain inherited conditions, it cannot show if the baby will show symptoms of a disorder, how severe it may be, or if the disorder will progress more. All of these factors make it seem as if the genetic screening before birth is not good as it carries a lot of negative possibilities. The emotional distress, the end result of the pregnancy, and the amount of information that can be given to the parent(s). Therefore, genetic screening before birth should not be done for the sake of the parent(s).

There are some potential directions this field of study could go, one being improving the tests to grant more information on the diagnosis or for there to be an ability to test later on in the pregnancy. The first direction it could go is providing more information for the tests to provide a more refined and detailed description of the condition for the parent(s). Currently parent(s) only receive a very limited amount of information about progression, severity, or if the baby will

present symptoms of a disorder. Next, the field of study could provide more testing for the pregnancies that have progressed past the usual time frame of prenatal screening. This would be beneficial for the parent(s) as some disorders develop later on in the pregnancy. The testing done later on in the pregnancy could provide insight for the parent(s) as they can continue to prepare for their child and ensure they have all the necessary support they need. Therefore, the controversial field of study could potentially provide more information for genetic screening for unborn children.

Genetic screening before birth is beneficial for parent(s) who have genetic disorders in their families, to ensure their child's quality of life, and provides insight for the supports the child may need in life. The disadvantages of genetic screening before birth include that it can cause emotional distress for the parent(s), too many false positives and negatives, and it has limited information for the parents. There are many tests that can be done which may be helpful for the parent(s) that have a child with no genetic disorders, but distressing for the parents who have babies that might have a disorder. My opinion on the use of genomics to make "designer babies" is that it is wrong to do. I believe this as life was not made to be perfect and the use of genomics in this way is only to get the desired traits for a baby. This is extremely unethical as the baby will not have any original traits of its own as it was designed to fit what the parent(s) wanted. Therefore, I believe the use of genomics to make "designer babies" is wrong and unethical.

Sources:

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