Birth Defects vs. Chromosomal Abnormalities: What's the Difference?

Sara contacted us at the recommendation of her OB/GYN to talk about the paroxetine she takes for her depression. Sara wanted to see if taking paroxetine can increase the chance of having a baby with a heart defect. She was confused because her genetic screening result was normal. She asked: "Is it possible for my baby to have a heart defect if my non-invasive prenatal testing (NIPT) was negative?"

There are many tests that can check on the development of your baby during pregnancy. It can be difficult to keep track of what they are looking for. Knowing the differences between a birth defect and a chromosomal condition can help you understand these tests.

What are birth defects and how can you look for them?

Out of all babies born each year, about 3 out of 100 (3%) will have a birth defect. Certain exposures, like medications, can increase the chance of birth defects. Most birth defects happen during the first trimester when the baby's organs (body parts) are developing. A certain organ might look or work differently. You may have heard of a baby being born with a hole in the heart. This is a common birth defect. Ultrasounds during pregnancy can look for birth defects. Read more about ultrasounds through our blog here. Most women will have an anatomy ultrasound between 18 and 22 weeks of pregnancy. You might know this as the time when you can learn the sex of the baby. The goal of this ultrasound is to take a detailed look at the baby's organs to check for birth defects. It will also check the growth of the baby. While this is an important screening test, it is not perfect. A normal anatomy ultrasound does not rule out the baby having a birth defect or other pregnancy-related problems.

A birth defect can be an isolated finding, meaning that there are no other related health problems. A birth defect might be caused by a change in the baby's genetic information, such as a chromosomal condition.

What are chromosomal conditions and how can you look for them?

Chromosomal conditions are genetic, meaning they are a change in the baby's genetic information. Chromosomal conditions do not usually run in families and are not inherited. Chromosomes are like a recipe book that holds all the recipes for the development of the baby. If a certain recipe calls for one stick of butter, but the book accidentally says two sticks of butter, the recipe will turn out differently. In the same way, sometimes people have extra or missing chromosomes that can cause changes in development. For example, people who have a chromosomal condition known as Down syndrome have three copies of chromosome 21 rather than two. This extra amount of genetic information causes developmental differences. Babies that have Down syndrome sometimes also have birth defects that are seen on ultrasound. Other babies that have Down syndrome can have normal ultrasounds. Therefore, ultrasound is not the best way to screen for chromosomal conditions.

Your healthcare provider will offer you screening for the most common chromosomal conditions. Testing for a chromosomal condition includes a screen called NIPT or diagnostic testing through procedures called CVS (chorionic villi sampling) or amniocentesis. NIPT involves drawing blood from you as early as 8-10 weeks of pregnancy. NIPT can tell you if you have a high or low chance of having a pregnancy with certain chromosomal conditions. Diagnostic

testing can tell you “Yes” or “No” about your baby having a chromosomal condition. Diagnostic testing involves a procedure that might come with small risks to the pregnancy. These screens and tests are optional. Your healthcare provider might suggest you meet with a genetic counselor. Genetic counselors can talk with you about the pros and cons of these tests, help with decision making and interpret the results.

During our conversation, Sara asked: “So because my NIPT results were low risk, the baby is unlikely to have a chromosomal condition. However, a birth defect could have happened in the first trimester, and I need to wait until my anatomy scan to get those results. Is that right?” That is exactly right! Although some studies show an increased chance of heart defects with paroxetine use in pregnancy, most other studies do not show an increased risk. If there is an increased chance of birth defects with paroxetine use in the first trimester, it is expected to be small. See our factsheet on Paxil® for more detail.

Sara shared that she was feeling more knowledgeable about the difference between a birth defect and a chromosomal condition. She was relieved that her NIPT results showed low risk. She also felt more confident heading to her anatomy scan knowing that this test, while not perfect, would be the best way to identify birth defects during pregnancy.

If you have any questions about exposures during pregnancy, contact MotherToBaby. To find a genetic counselor in your area, visit the Find a Genetic Counselor page on the National Society of Genetic Counselors website.

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