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

Sara contacted us on the MotherToBaby live chat service after being referred by her OB/GYN who had told her that the Paxil (drug name paroxetine) she was taking could cause a heart defect. She was 14 weeks along and wanted to learn more, but was confused because all her genetic testing results had come back normal. She asked: "How could my baby have a heart defect if my non-invasive prenatal testing (NIPT) was negative?"

During pregnancy, there are many tests done to check on the health and development of your baby. It can be difficult to keep track of what they are all looking for and why they are important. To understand these tests, it can be helpful to know the differences between a birth defect and a chromosomal abnormality and what they each mean for the health of your baby.

All pregnancies have a background risk for birth defects of 3-5%, but certain exposures can increase the risk of birth defects above this background estimate. Most birth defects occur during the first trimester while the baby is growing and developing. A certain body part – such as the heart, brain, or limbs – might not develop correctly, and the baby's body may look or function differently than it should. You may have heard of a baby being born with a hole in the heart, or with something called a cleft lip. These are two examples of birth defects that can occur during pregnancy. Birth defects can range from mild to severe, and the health of the baby will be dependent on where the birth defect occurs and how severe the problem is.

Certain tests done during pregnancy can look at your baby to see if there are birth defects. For example, most pregnant individuals will go in for an anatomy scan between 18 and 22 weeks where the healthcare provider will look at the baby using an ultrasound. Most people know this as the time when they can learn the gender of the baby, but the ultrasound will also take a detailed look at the baby's organs and body parts, including the heart, brain, face, and stomach, to check for birth defects. While this is an important screening test, it is not perfect, and more minor defects may go unnoticed until birth.

Chromosomal abnormalities are changes in the baby's DNA that happen at the time of conception. The best way to understand DNA is to think of it like a recipe book that holds all the recipes for the growth and development of different body parts. If a certain recipe calls for one stick of butter, but the printed book accidentally says two sticks of butter, the recipe will turn out different. In the same way, sometimes people have extra or missing amounts of DNA that can cause changes in development. For example, people who have a chromosomal abnormality known as Down syndrome have three copies of their 21st chromosome rather than only two. This extra amount of DNA is what causes the developmental differences in individuals with Down syndrome.

During your pregnancy, your healthcare provider may suggest that you meet with a genetic counselor to have non-invasive prenatal testing, or NIPT, performed. This test can be done as early as 10 weeks. During pregnancy, some of the baby's DNA enters the pregnant woman's bloodstream. By taking a small blood sample from mom, a lab can take a look at the baby's DNA and tell if they have certain chromosomal abnormalities, such as Down syndrome. When you meet with a genetic counselor, they will go over all of the pros and cons of this test as well as your family history, and will meet with you again to review the results.

Back on the live chat service, Sara asked: "So because my NIPT results were normal, the baby is unlikely to have a chromosomal abnormality. However, a birth defect still could have happened in the first trimester, and I need to wait until my anatomy scan to get those results, is that right?" That's exactly right, I replied. I then went on to explain that

although Paxil (paroxetine) has been shown to increase the risk for heart defects in some studies, other studies do not suggest a risk, so the overall chance of the baby being affected is low.

As we ended our chat, Sara shared that she was feeling much more knowledgeable about the difference between a birth defect and a chromosomal abnormality. She was able to breathe a sigh of relief that her NIPT results came back normal, indicating a low risk for conditions like Down syndrome. She also felt much more confident heading to her anatomy scan in a few weeks knowing that this test, while not perfect, would be the best way to identify birth defects before her baby is born.

If you have any questions about birth defects or exposures during pregnancy, speak with a MotherToBaby specialist via phone, text, live chat, or email. For any questions regarding genetic testing in pregnancy, or to find a genetic counselor in your area, visit the Find a Genetic Counselor page on the National Society of Genetic Counselors website: https://findageneticcounselor.nsgc.org/