

What Conditions are Commonly Tested for During Pregnancy?

During your pregnancy, you will likely be offered tests that can provide information about whether or not certain birth defects or genetic conditions may be present in your baby. Tests you may be offered include blood tests, ultrasound or a procedure, such as amniocentesis. It is important to keep in mind that most babies are born without any type of condition or birth defect. However, with all pregnancies, there is a 4 out of 100 chance for this to occur.

This video will provide a brief overview of some of the conditions that may be tested for during pregnancy. This information can help you have a more in-depth discussion with your doctor about the testing options and the conditions that are tested for. When it comes to tests during pregnancy – there are typically two types of conditions that are commonly discussed - birth defects AND chromosome conditions.

Birth Defects

Birth defects can occur in anyone and do not always run in the family. You may have heard of some structural birth defects such as heart defects or cleft lip and palate. The severity of birth defects is variable, and they can often be treated.

Birth Defects- Neural Tube Defects

Other examples of birth defects are neural tube defects. Neural tube defects involve an opening in the spine or the skull. One example of a neural tube defect is spina bifida. Another example is anencephaly, which is a more severe, life-limiting neural tube defect.

Chromosome Conditions

Other types of conditions that may be tested for during pregnancy are chromosome conditions. Chromosomes are structures that hold our genetic information, the information that tells our bodies how to grow and function. Chromosome conditions occur when the baby has extra or missing genetic information. These conditions usually do not run in families.

Although the chance to have a baby with a chromosome condition increases with the age of the mother, babies with chromosome conditions are born to women of all ages.

Down Syndrome (Trisomy 21)

One of the most common chromosome conditions is Down syndrome, also known as Trisomy 21. Trisomy 21 means there are 3 copies of the 21st chromosome instead of the typical 2 copies. Individuals with Down syndrome have some degree of intellectual disability that can range from mild to severe. This condition is also associated with medical concerns that are often treatable.

Trisomy 18 & Trisomy 13

Other chromosome conditions that may be tested for include Trisomy 18 and Trisomy 13. Both of these conditions are associated with life-limiting medical complications.

Sex Chromosome Variations

Some prenatal tests can look for milder chromosome conditions related to differences in the number of X and Y chromosomes; these conditions are called sex chromosome variations. Children with sex chromosome variations usually have typical intellectual ability, however, they may have learning differences. Some of these conditions are associated with a higher chance for health concerns that are often treatable. Because these conditions can be so mild, some individuals may go through life without ever even knowing they have a sex chromosome variation.

In summary, prenatal testing can look for a wide range of conditions, from mild to severe. It is also important to know that no prenatal test can rule out all genetic conditions or birth defects. If you have a family history of a specific genetic condition or birth defect or questions about anything presented in this video, we recommend you talk with your doctor or genetic counselor.

You can find more detailed information about these conditions on Genetic Support Foundation's website. See the other videos in our series to learn more about specific prenatal testing options and how to decide if prenatal testing is right for you.

Credits

This video was created by the Washington State Department of Health and Genetic Support Foundation. You can find more information and additional videos about prenatal genetic testing options at: www.doh.wa.gov, www.geneticsupportfoundation.org

Disclaimer

This project was supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under grant number (93.994) Title V Maternal and Child Health Block Grant. This information or content and conclusions are those of the authors and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government. The content of this video is for informational purposes only and is intended to educate consumers about health care issues and choices. This information is not a substitute for professional medical advice, diagnosis, or treatment. You should always consult with your physician before making medical decisions or electing to undergo any medical testing or treatment, or if you have any questions or concerns about your health.

*Highlighted Version Not Spoken