

A Patient's Guide to Understanding Non-Invasive Prenatal Testing

Screening for genetic conditions is offered to pregnant women because all pregnancies have a small chance for a genetic condition regardless of maternal age, family history, or personal health. Some screening methods are routine, such as an ultrasound. Other screening tests are optional, such as blood tests for Down syndrome.

When considering prenatal screening options, you may want to discuss your thoughts, feelings, and how prenatal screening results may affect your pregnancy with your obstetric provider or genetic counselor. During this discussion, please consider the following questions.

Should I undergo prenatal screening for genetic conditions?

Based on your values and needs, you may choose whether or not to undergo these screening tests. For some, the benefits of prenatal screening might include receiving reassurance from the results; preparing emotionally to raise a child with a health issue or disability; or learning more about the condition. Some might want to arrange specific birth plans; start treatments as soon as possible after birth; or, in some cases, prepare for a baby who is not likely to survive. Some might create an adoption plan for a child with a disability or decide not to continue their pregnancy. For others, prenatal screening may cause unwanted stress and worry during pregnancy, and they may prefer to wait until delivery to find out if their baby has a genetic condition.

What are non-invasive prenatal screening tests?

You may be offered non-invasive prenatal testing (NIPT) as a way to screen for some specific genetic conditions, such as Down syndrome, the most common chromosome condition. NIPT may also be referred to as non-invasive prenatal screening (NIPS), cell-free DNA testing (cfDNA), or other specific brand names. NIPT is performed on a blood sample from a pregnant woman and poses no risk of miscarriage to the pregnancy. NIPT can better estimate the chances for some chromosome conditions, but is not 100% accurate. NIPT can miss a condition that is present (false negatives) or can incorrectly show high chances for a condition when none exists (false positives). In addition, NIPT does not detect all genetic conditions or risk factors that might be present in a pregnancy. Therefore, diagnostic testing is recommended for those who want to be certain or for those who would like to test for more conditions. A medical professional should review the results with you.

Alternatively, there are several other blood tests that you may be offered, such as a first trimester screen, second trimester screen, sequential screen, or integrated screen. For questions about these screening tests, please contact your obstetric provider.

What is diagnostic testing, and how is it different from screening?

Diagnostic testing is used to confirm or rule out chromosome conditions with the most accuracy. These tests are also more comprehensive and can detect other genetic conditions not found by screening tests. Depending on how far along you are in pregnancy, two diagnostic testing options may be available, including chorionic villus sampling (CVS) or amniocentesis. These are procedures in which a small sample of placental tissue or amniotic fluid is obtained to examine the baby's chromosomes. Because these procedures are invasive, there is a risk, likely less than 1%, for complications that can lead to miscarriage.

What conditions can NIPT identify?

NIPT routinely screens for conditions such as Down syndrome, trisomy 18, and trisomy 13. Screening for gender, sex chromosome conditions, and several other genetic conditions may also be included. Even though it is not diagnostic, NIPT has been shown to be the most sensitive screening test for Down syndrome. More information about the accuracy of screening for other chromosome conditions is needed. Prenatal screening options are

constantly evolving to include more conditions, so your medical providers can explain which conditions are included in your screening test.

Individuals with chromosome conditions can experience a broad range of outcomes. For example, serious medical and neurological issues occur in babies with trisomy 13 and 18, with about 10% living past the first year. Individuals with Down syndrome typically have mild to moderate intellectual disabilities and some treatable medical issues. They usually become active members of their communities and live an average of 60 years. The effect of an extra or missing sex chromosome may be so mild that it goes undiagnosed throughout a person's life.

How long before I receive my NIPT results?

NIPT results are typically available in 5-10 days. You can ask your obstetric provider or genetic counselor how and when you will be receiving your NIPT results.

How do I interpret my NIPT results?

A negative or low risk NIPT result indicates the pregnancy is unlikely to be affected by any of the conditions included in the screen. It does not eliminate the chance, and NIPT does not screen for all genetic conditions. A positive or high risk NIPT result indicates an increased chance your pregnancy has a specific genetic condition. Your actual chances for the condition after a positive or high risk screen depend on several factors such as the particular condition, maternal age, timing during the pregnancy, family history, and ultrasound results. In rare instances, NIPT results can raise concern for an unexpected condition in a pregnancy and/or in a mother. Sometimes, an NIPT result cannot be obtained for a variety of reasons. In these cases, we encourage you to have a discussion with your genetic counselor or obstetric provider.

How do I get information and support if my screening test comes back positive or high risk for a genetic condition?

While prenatal screening offers more information about your pregnancy, it can also lead to many questions such as: what does the screening result or diagnosis mean? How do I determine if my baby has this condition? What quality of life does a person with this diagnosis have? Where can I find reliable information about this condition?

Sometimes expectant parents find incorrect or out-of-date information when trying to learn about different conditions. The outcomes and attitudes about many conditions have improved greatly in recent years. This means you need current information about genetic conditions so that you can make informed choices about your pregnancy and find any needed services, resources, and support. Your obstetric provider or genetic counselor can direct you to resources with accurate and up-to-date information.

If you would like additional information, you can request a referral to a genetic counselor. Genetic counselors are health care professionals with specialized training in prenatal genetics and in the emotional complexities surrounding genetic testing and screening. Genetic counseling is available to help you understand your options and facilitate a decision about testing as well as provide accurate information about your test results. A genetic counselor can be located by your obstetric or medical provider or by using the "Find a Genetic Counselor" link on the www.nsgc.org website.

Where can I find more information about prenatal screening, testing, and various genetic conditions?

You can find more detailed information about prenatal screening, testing, and chromosome conditions at www.lettercase.org/prenataltesting/. This information is an introduction to your prenatal testing options, and you can discuss them further with your genetic counselor or obstetric provider.