

Non-Invasive Prenatal Testing

Every pregnancy, regardless of a woman's age, has a chance to have a chromosome condition. The most common chromosome condition is Down syndrome, though there are other chromosome conditions that can be more or less severe. Non-Invasive Prenatal Testing is a blood test available to women who would like to learn more about their chances to have a pregnancy with one of these conditions. All genetic testing is optional and this information is intended to help you understand your options. Keep in mind, no test or combination of tests can screen for all genetic conditions and birth defect.

What is Non-Invasive Prenatal Testing and when is it done?

Non-Invasive Prenatal Testing, also called NIPT, is a blood test performed during pregnancy that screens for Down syndrome (also called trisomy 21), trisomy 18, and trisomy 13. Several laboratories offer this test. It may be referred to by several different names, including: Harmony, MaterniT21, Verifi, or Panorama. Additionally, NIPT screens for conditions caused by extra or missing sex chromosomes. NIPT does not screen for all genetic conditions. NIPT can be drawn as early as 10 weeks gestation and results generally take about 2 weeks to return.

How accurate is NIPT?

NIPT is a highly accurate screen, however, it is not 100% accurate. It is not considered a diagnostic test (see *What are my other options?*). The detection rate is laboratory-dependent but is typically between 90-99% for high-risk, singleton (only one baby) pregnancies with false positive rates of less than 1%.

What are the possible results?

Negative/Low Risk: A negative or low risk result means that it is very unlikely the pregnancy has one of the conditions NIPT screens for. It is again important to remember that this test is not diagnostic and a relatively low chance still remains for the pregnancy to have one of the conditions.

Positive/High Risk: A positive or high risk result means the pregnancy is at increased risk for a particular condition. Only diagnostic testing can provide a definite answer. Genetic counseling is recommended to discuss positive/high risk results along with further testing options.

No Result/Inconclusive: There is a small possibility that results may not be reportable or may be inconclusive. In these cases, your healthcare provider may recommend you have your blood redrawn, have different testing, or have genetic counseling to discuss your options.

False positive and false negative results are possible with NIPT, therefore a result may be negative/low risk and a baby be born with one of these conditions, or may be positive/high risk and the baby not have that condition.

Can anyone have the NIPT test?

The accuracy of NIPT is best understood in have an increased risk for their pregnancy to have one of these chromosome conditions. This would include women who are 35 years or older at the time of delivery, who had a previous pregnancy or child with a chromosome condition, who have a positive screening test in this pregnancy, or when there are concerns on ultrasound in their current pregnancy.

The accuracy of NIPT in the general population, or low risk women, is not well understood. The test still appears to have a 90-99% detection rate, however, when a test comes back positive/high risk for a low risk woman, the chance that the pregnancy has a chromosome condition is not as high as it is for high risk women. Furthermore, insurance coverage may vary for low risk women.

Limited information is available on the accuracy of NIPT in twin pregnancies. While it is believed that NIPT is more accurate than other screening tests available, a positive result cannot distinguish if only one or both twins are considered increased risk.

What are my other options?

Maternal Serum Screening (First Trimester Screening and Quadruple Marker Screening): screens for Down syndrome, trisomy 18, and sometimes open neural tube defects (called spina bifida). These tests provide an estimate of risk, or chance, that the pregnancy has one of these conditions, however they are not diagnostic. Depending on the test, approximately 75-90% of pregnancies with these conditions will screen positive and 1-5% of pregnancies will receive a false positive result.

Diagnostic Testing: provides a “yes” or “no” answer regarding chromosome conditions during the pregnancy. There are two options. Both diagnostic procedures are performed by an experienced obstetrician under ultrasound guidance; however, both options carry a risk for pregnancy complications and/or miscarriage, less than 1%.

- Chorionic villus sampling (CVS): invasive procedure performed between 11 weeks and 13 weeks 6 days of pregnancy that samples a small piece of the placenta to evaluate the chromosomes of the pregnancy.
- Amniocentesis: invasive procedure typically performed between 16 and 20 weeks of pregnancy that samples a small amount of amniotic fluid to evaluate the chromosomes of the pregnancy.

Ultrasound: screens for birth defects and markers for genetic conditions. Approximately 50% of pregnancies with Down syndrome and 90% of pregnancies with trisomy 18 and trisomy 13 have findings on ultrasound that raise concern. Thus, a normal ultrasound can be reassuring and lower the risk for these conditions, but it cannot rule out all birth defects or genetic conditions.

How do I know if NIPT is right for me?

Talking to a genetic counselor may be beneficial in helping you decide on a personal screening plan. Genetic counselors are healthcare professionals trained to discuss the various risk factors during pregnancy and review the risks, limitations, and benefits of screening and diagnostic testing, as well as what it may mean for a family if test results are concerning for a condition. It is also important to remember that while most babies are born healthy, there is always a 3-5% risk for birth defects in any pregnancy.

If you have additional questions about these testing options and would like to speak to a genetic counselor, please inform the front desk and this will be arranged for you.

Kindest regards,

The Prenatal Genetic Counselors and Care Team