

Non-Invasive Prenatal Testing (NIPT)

Non-invasive prenatal testing uses cell-free fetal DNA from maternal plasma to screen your baby for fetal aneuploidy, or chromosomal abnormalities. NIPT is the most accurate screening test for Trisomy 21 (Down syndrome), which comprises more than 50% of all chromosomal abnormalities and 8-10% of all significant birth defects.

What does NIPT screen detect?

This relatively new screening option is constantly adding new disorders to its testing menu.

Currently, *Harmony*TM screens for:

- Down syndrome (Trisomy 21)
- Edward's syndrome (Trisomy 18)
- Patau syndrome (Trisomy 13)
- Turner and Klinefelter syndromes
 - These disorders involve the X/Y chromosome. If you wish, you can find out the fetal sex with this testing.

*The NIPT does not screen for neural tube defects, such as Spina Bifida. Ask your provider about additional screening options for these anomalies.

How is this testing performed?

NIPT is blood draw performed between 10-14 weeks of pregnancy. Two tubes of blood will be drawn and can be obtained at the same time as your initial labs. The lab will receive the samples and report in about a week.

What happens next if the test is positive?

In the event of a positive result, you will be referred to a genetic counselor. This counselor will help you navigate the next steps, such as additional testing.

NIPT is a screening test, so false negative and false positive may occur. Women who desire definitive information about chromosomal conditions in their pregnancy will be offered a follow-up Amniocentesis or Chorionic Villus Sampling (CVS).

For whom is this test recommended?

Currently, NIPT is recommended for high-risk patients. You are considered high risk if:

- You will be over 35 years old when your baby is delivered
- You have had another child with a chromosomal abnormality or other genetic anomaly
- You have a family history of chromosomal abnormality or other genetic anomaly.

Use in "low-risk" population is still something that is evolving and is not regularly reimbursed by insurance companies as of yet. You may incur out of pocket costs for NIPT testing.

Should I get this test?

All genetic screening is elective. Whether a woman chooses to have aneuploidy screening, prenatal diagnostic testing, or no testing is a personal decision and any of these is a reasonable option. Explore your feelings about the testing and discuss with your family. Some important questions to consider are:

- What will I do with the information that the test provides?
- In the event of a positive result, would we ever consider terminating the pregnancy?
- Will I stress and worry if I get the test?
- Will I stress and worry if I don't get the test?
- Would I potentially change my plan for delivery if I were positive?