MORE ANSWERS FOR MORE PATIENTS

Prenatal screening is testing offered to all parents who want to learn information about the health of their pregnancy. Most pregnancies have a low risk for birth defects and genetic conditions. Some women have a family history or medical history that identifies their pregnancy as "high risk". Even in "low risk" pregnancies, 2-3 out of every 100 babies are born with a birth defect. Prenatal screening tests are used to find pregnancies that may be at high risk for certain conditions that may require special care.

Footnotes:

1. Dahl F, Ericsson O, Karlberg O, et al.; Imaging single DNA molecules for high precision NIPT. Scientific Reports 2018;8:4549
2. Practice Bulletin No. 226: Screening for fetal chromosomal abnormalities. Obstet Gynecol 2020;136.
3. Gregg AR, Skotko BG, Benkendorf JL et al. Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics. Genet Med. 2016;18(10):1056-65.
4. Bianchi DW, Parker RL, Wentworth J, et al.; CARE Study Group. DNA sequencing versus standard prenatal aneuploidy screening. N Engl J Med 2014;370:799–808.
5. Norton ME, Jacobsson B, Swamy GK, et al. Cell-free DNA analysis for noninvasive examination of trisomy. N Engl J Med 2015;372:1589–1597.
6. Zhang H, Gao Y, Jiang F, et al. Non-invasive prenatal testing for trisomies 21, 18 and 13: clinical experience from 146,958 pregnancies. Ultrasound Obstet Gynecol 2015;45:530–538.

Prenatal PRECISE
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Patient Information

THE PRENATAL TEST PRECISELY MADE FOR YOU

Prenatal PRECISE
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Non-Invasive Prenatal Testing

One of the tests that is offered during pregnancy is NIPT (Non-Invasive Prenatal Testing). NIPT is a screening test for pregnant women to help determine their chances for having a baby with trisomy 21 (Down syndrome), trisomy 18, or trisomy 13. Each of these conditions are caused by the presence of an extra copy of a chromosome. These conditions can occur in any pregnancy, even without a family history. Trisomy 18 and trisomy 13 are typically considered more severe than Down syndrome.

How does NIPT work?

When a woman is pregnant, very small pieces of the baby's DNA enter the mom's bloodstream. NIPT is performed by drawing the mother's blood and analyzing the fetal DNA (see Figure 1). When the fetal DNA fragments are measured in mom's blood, we expect to see balanced counts for the each of the sets of chromosomes. When the amount of DNA is not balanced, this can indicate a possible chromosome condition.[1] NIPT can also provide information about the likely sex of the baby.

The data from the fetal DNA combined with other information such as the patient's age and family history are used to calculate the risk for trisomies 21, 18, and 13.

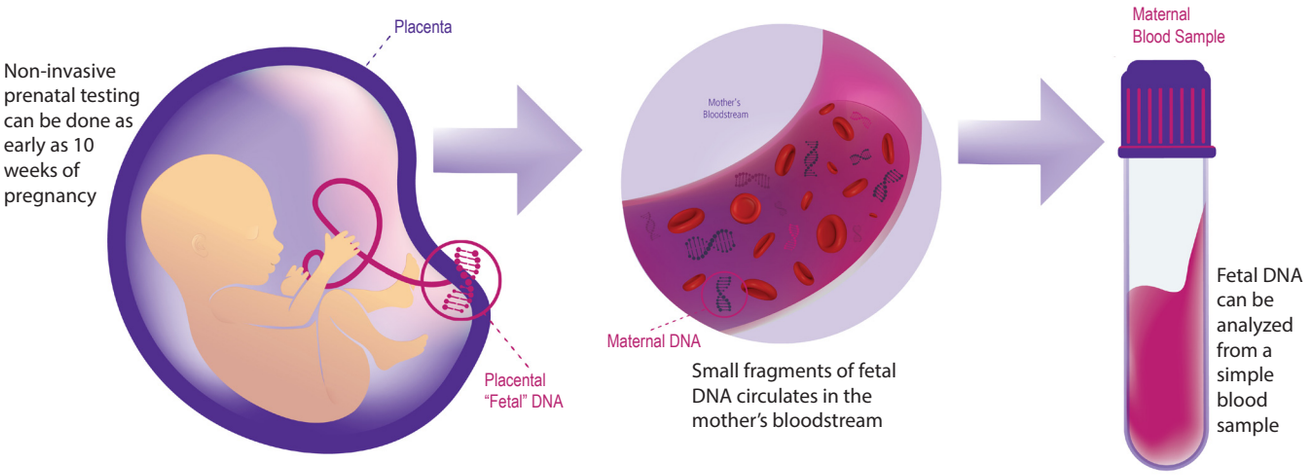
Why should I consider NIPT?

It is up to you to decide if you would like to have this screening test. NIPT is the most sensitive screening test available for the most common trisomies.[2-6] It is performed by a simple blood test so there is no risk to the pregnancy. The test is available any time after 10 weeks of gestation, and results usually take around one week.

Women may choose to get NIPT during the pregnancy if they would like more information about the health of the baby and if these specific chromosome conditions could be present.

NIPT can detect pregnancies at increased risk for trisomy 21, trisomy 18, or trisomy 13, but this is considered a screening test. Diagnostic testing, such as amniocentesis, is needed to provide a definitive yes or no answer. This early information can aid parents and doctors when making decisions about the pregnancy and pregnancy management which may include monitoring the pregnancy more closely or planning to give birth in a hospital that can provide special care for the baby.

Figure 1



What will my results mean?

Low Risk Result

The baby is unlikely to have trisomy 21, trisomy 18, or trisomy 13. A negative test does not mean the baby will not have a genetic condition or birth defect. There is no prenatal test that can detect all possible problems.

Increased Risk Result

There is a greater than 90% chance that the baby has the trisomy indicated on the report (21, 18, or 13). Screening ultrasounds or diagnostic procedures such as amniocentesis may be used for further evaluation. These options should be discussed with your healthcare provider following an increased risk result.

Inconclusive

There is a small chance for an inconclusive result. A result cannot be provided in less than 1% of cases.

For all women taking the test, there is less than a 1% chance that a false positive result will be reported.