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Prenatal Screening for Birth Defects

Many pregnant women have concerns about the health of their unborn children. But prenatal testing can help reduce these concerns. Prenatal testing is done before birth. It helps find out if the unborn child is at greater risk for, or has, certain birth defects. These include open neural tube defects (eg, spina bifida), Down syndrome, trisomy 18, and other birth defects.

Types of prenatal testing

There are 2 basic types. The first is *prenatal screening*. These tests can find out if an unborn child is at greater risk for certain birth defects. But screening cannot tell if the unborn child actually has a birth defect. More testing is needed for that.

The other type of prenatal testing is *diagnostic testing*. This type can tell if the unborn child actually has a birth defect. Most of the time, diagnostic testing is done to follow up a positive screening test.

Prenatal screening options

There are 2 types of prenatal screening tests. One type is testing that detects certain substances in the mother's blood. These are called *maternal serum screening (MSS) tests*. They can test for AFP, hCG, h-hCG, estriol, inhibin, and PAPP-A. The amount of these substances in the blood can show if the unborn child is at a higher risk for certain birth defects.

The other type is testing that looks for certain changes in the unborn child's DNA. This DNA is found in the mother's blood—more about this later. If changes are found, it's a sign of a birth defect. Currently, this test is only for pregnant women whose unborn child is more likely to have a birth defect.

A positive result in either one of these tests means that the unborn child has a higher risk of having a birth defect. Diagnostic testing is the next step. It can tell if the unborn child actually has a birth defect.

More about the DNA screening test

This test is called by a couple names:

- *Cell-free fetal DNA test*
- *Noninvasive prenatal test (NIPT)*

DNA makes up genes, which make up chromosomes. Chromosomes are part of a cell. When the DNA leaks out of a cell, it's called cell-free DNA.

Who can have the NIPT?

Pregnant women whose unborn child is more likely to have a birth defect can have the test. These include women who:

- Are 35 years old or older
- Have had an abnormal ultrasound
- Have had a previous pregnancy with one of these birth defects
- Have a positive MSS test

The NIPT is not recommended for pregnant women whose unborn child has a low risk of a birth defect.

If this DNA comes from the unborn baby, it's called fetal DNA. Cell-free fetal DNA gets into the mother's blood, so the test can be done using a sample of the mother's blood.

In the past, the only way to test fetal DNA was to take it from the placenta or the fluid surrounding the unborn child. So, collecting the sample is relatively invasive. It is also risky—there is a small chance that the unborn child could die. Taking a sample of the mother's blood is almost risk-free. It's much less invasive. That's why the test is sometimes called the noninvasive prenatal test, or NIPT.

Which type of screening test is best?

Data so far show that the NIPT is more accurate than the MSS screening tests. It can also give information about more birth defects. These include trisomy 13 and Turner syndrome. But, the NIPT cannot detect neural tube defects.

The NIPT has only been studied in pregnant women whose unborn child is more likely to have a birth defect. So it's not recommended for all pregnant women.

The table below gives an idea of how the NIPT compares to the best of the non-DNA screening tests.

Birth Defect	MSS Test	NIPT[1-3]
% of open neural tube defects detected	80	0
% of Down syndrome cases detected	92	>99
% of trisomy 18 cases detected	90	>99
% of trisomy 13 cases detected	0	>99
% of Turner syndrome cases detected	0	92

Which prenatal screening tests does Quest Diagnostics offer?

Quest Diagnostics offers both types of prenatal screening tests:

- MSS tests that measure the amount of certain substances in the mother's blood:
  - First Trimester Screen
  - Triple Screen
  - Quad Screen
  - Penta Screen
  - Integrated Screen (4 different ones)
- Test that looks at the cell-free fetal DNA (ie, the NIPT):
  - Panorama™ Prenatal Test

Quest Diagnostics also offers all the diagnostic tests your doctor needs.

Learn more about prenatal testing

You can find out more about prenatal testing from

- Quest Diagnostics QuestDiagnostics.com/home/patients/tests-a-z/prenatal/during-pregnancy
- March of Dimes marchofdimes.com
- WebMD® webmd.com/baby/guide/prenatal-tests

References

1. Zimmermann B, Hill M, Gemelos G, et al. Noninvasive prenatal aneuploidy testing of chromosomes 13, 18, 21, X, and Y, using targeted sequencing of polymorphic loci. *Prenat Diagn.* 2012;32:1233-1241.
2. Levy B, Demko Z, McAdoo S, et al. Use of targeted sequencing of SNPs to achieve a highly accurate non-invasive detection of fetal aneuploidy of 13, 18, 21, and sex chromosomes. Paper presented at: 33rd Annual Meeting of the Society for Maternal-Fetal Medicine; February 2013; San Francisco, CA.
3. Nicolaides KH, Syngelaki A, Gil M, et al. Validation of targeted sequencing of single-nucleotide polymorphisms for non-invasive prenatal detection of aneuploidy of chromosomes 13, 18, 21, X, and Y. *Prenat Diagn.* 2013;33:575-579.