The American College of Obstetricians and Gynecologists
WOMEN'S HEALTH CARE PHYSICIANS

CELL-FREE DNA PRENATAL SCREENING TEST

How the Test Is Done

What is it?

The cell-free DNA prenatal screening test (also called "cfDNA test") screens for certain conditions caused by an abnormal number of chromosomes. It does not test for all types of chromosomal disorders.

When can it be done?

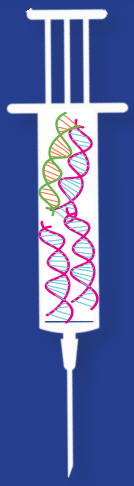
It can be done as early as 9–10 weeks of pregnancy and up until delivery.

How is it done?

Some of the genetic material (DNA) from the pregnancy circulates in the mother's bloodstream. The cfDNA test is done on a sample of the pregnant woman's blood.

Screening tests are used to estimate whether your baby is at higher risk or lower risk of having a certain condition.

Diagnostic tests can give a definite answer about whether the baby has a certain condition. These tests include amniocentesis or chorionic villus sampling (CVS).



A blood sample is taken from the pregnant woman that contains maternal DNA and DNA from the pregnancy.

The sample is analyzed in a laboratory to check for the presence of an abnormal amount of DNA from chromosomes 21, 18, and 13.

Major conditions screened for:

- Trisomy* 21 (Down syndrome)
- Trisomy 18
- Trisomy 13

Conditions not screened for:

- Problems that are screened for by ultrasound, such as neural tube defects, heart defects, and abdominal wall defects
- Many other chromosomal and genetic disorders

= maternal DNA
= DNA from the pregnancy

*Trisomy means that there are three copies of a particular chromosome instead of the normal two copies. For instance, trisomy 21 means that there are three copies of chromosome 21.

PFSI008: Designed as an aid to patients, this document sets forth current information and opinions related to women's health. The information does not dictate an exclusive course of treatment or procedure to be followed and should not be construed as excluding other acceptable methods of practice. Variations, taking into account the needs of the individual patient, resources, and limitations unique to the institution or type of practice, may be appropriate.

National Society of Genetic Counselors

Society for Maternal-Fetal Medicine

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Understanding Your Results

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What do the results mean?

If you get a positive result, how likely is it that the baby actually has the disorder? This is called the positive predictive value (PPV) of the test.

If you get a negative result, how likely is it that the baby actually does not have the disorder? This is called the negative predictive value (NPV) of the test.

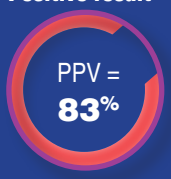
For the cfDNA test, the positive predictive value depends on how frequently the disorder occurs in a group of people similar to you.

For Example: Trisomy 21 (Down Syndrome)

High-risk* group

The disorder occurs more frequently (e.g., 1 in 100 or higher) in this group.

Positive result



Out of 100 women with a positive result, 83 will have babies with Down syndrome, and 17 will not have babies with Down syndrome.

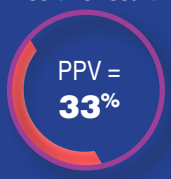
Negative result

Women with a negative result will only rarely have a baby with Down syndrome.

Low-risk group

The disorder occurs less frequently (e.g., 1 in 1,000 or lower) in this group.

Positive result



Out of 100 women with a positive result, 33 will have babies with Down syndrome, and 67 will not have babies with Down syndrome.

Negative result

Women with a negative result will only rarely have a baby with Down syndrome.

Sometimes the test does not yield a result or is indeterminate. In this case, you should receive further genetic counseling, and an ultrasound exam or diagnostic testing should be offered due to an increased risk of a chromosomal disorder.

*You are at "high risk" if you are 35 years or older; you have had an ultrasound exam that shows a possible problem with the fetus; you have had a previous child with one of these disorders; you have a chromosomal problem that increases your risk of having a child with trisomy 21 or trisomy 13; or you have had a positive first-trimester or second-trimester screening test result.

BOTTOM LINE:

- Cell-free DNA testing is a very good screening test to detect common chromosomal disorders in high-risk women, but it has limitations.
- A negative result does not rule out the possibility of having a baby with a chromosomal disorder or other disorders that the cfDNA test does not test for.
- If you have a positive result, a diagnostic test is needed to determine if the baby is truly affected. False-positive results are more frequent among low-risk women than those at high risk.
- If you have cell-free DNA screening, a blood test or ultrasound exam should be offered to screen for neural tube defects and other conditions.

http://www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-On-Genetics/Cell-Free-DNA-Screening-for-Fetal-Aneuploidy

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