



# Cell-Free DNA Prenatal Screening Test

## How the Test Is Done

### What is it?

The cell-free DNA prenatal screening 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?

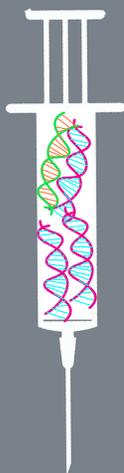
A cell-free DNA test can be done as early as 10 weeks of pregnancy and up until delivery.

### How is it done?

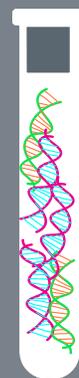
Some of the genetic material (DNA) from the pregnancy circulates in the pregnant woman's blood. The cell-free DNA test is done on a sample of her blood.

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

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



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



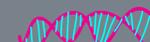
The sample is analyzed in a laboratory to check for an abnormal amount of DNA from chromosomes 21, 18, and 13.

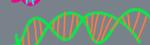
#### Major conditions screened for:

- 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

 = woman's 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.



# Cell-Free DNA Prenatal Screening Test

## Understanding Your Results

### What do the results mean?

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

For the cell-free DNA 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)

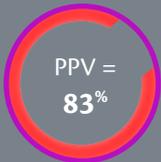
#### High-risk\* group

The disorder occurs more frequently in this group.

#### Low-risk group

The disorder occurs less frequently 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.

#### 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.

An ultrasound exam and diagnostic testing also 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, 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 cell-free DNA test does not test for.
- If you have a positive result, a diagnostic test is needed to determine if the fetus is truly affected.
- 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.