# Facts about
# Cell-Free DNA Screening

## Cell-free DNA (cfDNA) screening is a blood test done during pregnancy. This test checks the developing baby for certain chromosome conditions.

DNA is the genetic information we inherit from our parents. It is found in most cells of the body and is also in your blood. When you are pregnant, a small amount of the DNA in your blood comes from the placenta. This is called fetal DNA. Cell-free DNA screening checks for differences in the amount of fetal DNA. This can let you know about the chance for birth defects caused by an abnormal number of chromosomes.

### How is cfDNA screening done?

cfDNA screening is done with a single blood test from your arm. It can be done any time after 10 weeks in pregnancy and is usually done before 21 weeks.

### What can cfDNA tell me?

cfDNA can tell if your pregnancy has a low risk or high risk for certain chromosome conditions, including: Down syndrome (trisomy 21), trisomy 18, and trisomy 13. cfDNA helps find almost all pregnancies with these 3 conditions. The test can also predict the sex of the baby.

**DID YOU KNOW?**

**Chromosomes are the packages of DNA found in most cells of the body. There are typically 46 chromosomes in each cell.**

**Chromosome conditions can be caused by having an extra or missing chromosome in each cell (ex: 47 chromosomes). These conditions happen by chance and do not usually run in the family.**

### What are the limits of cfDNA?

**cfDNA only looks for 3 chromosome conditions.** This test does not find all chromosome conditions and does not test for any other birth defects or genetic conditions.

**No screening test is 100% accurate.** This test can raise false alarms in healthy pregnancies (false positive results). It can also miss a pregnancy that has one of the 3 conditions (false negative results).

### Who can have cfDNA?

cfDNA can be done if you are pregnant with one baby or with twins. cfDNA cannot be done with triplets. A small number of pregnancies are not able to have cfDNA. If this is the case for you, your doctor will let you know about other screening options.

### How do I get results from cfDNA?

Results from cfDNA usually take about 2 weeks. You will get a secure email message or a phone call when the result is ready. You can also find your test result on kp.org (if you have an account).

### What if my cfDNA result is screen negative (low risk)?

Most people who have cfDNA get a screen negative result. A screen negative result is reassuring. It means there is a very low risk for Down syndrome, trisomy 18, or trisomy 13. It is rare for a baby with one of these 3 conditions to have a negative result and be missed by this test. This result does not guarantee that the baby has no health problems or birth defects.

### What if my cfDNA result is screen positive (high risk)?

A screen positive result means the test found a difference in the amount of fetal DNA. This may be because the baby has a chromosome condition, but there are other reasons for a positive result. If you have a positive result, you are offered a test that can show if the baby really has a chromosome condition.

### IMPORTANT THINGS TO CONSIDER

**Cell-Free DNA Screening:**

...has no risk for miscarriage.

…does not test for all chromosome conditions.

…does not find all pregnancies with Down syndrome, trisomy 18, or trisomy 13.

…does not test for other genetic conditions or other birth defects.

…may show a high risk in a healthy pregnancy (false-positive result).

...requires more testing to diagnose a chromosome condition.

…is unable to give a result in a small number of pregnancies.

## What tests will I be offered if cfDNA shows a screen positive (high risk) result?

If you have a screen positive result you will be offered CVS or amniocentesis. Both CVS and amniocentesis accurately diagnose chromosome conditions by studying fetal cells in the laboratory. These tests have a very small risk of miscarriage (about 1 in 1000).

You can decide if you want one of these tests.

- **Chorionic villus sampling** (CVS) takes a small sample of the placenta between 10 and 14 weeks in pregnancy. It is done by passing a needle through the lower belly or a flexible tube through the vagina – without touching the baby.
- **Amniocentesis** takes a small sample of the fluid around the baby between 15 and 22 weeks in pregnancy. It is done by passing a thin needle through the belly – without touching the baby.

## What if I get "No Result" for fetal sex?

cfDNA screening can predict the sex of the baby, if requested. However, a small number of people will get "no result" for fetal sex on the lab report. This is not a sign of a problem with the baby. It is just a limitation of the test. A routine ultrasound done about 20 weeks in pregnancy is usually able to predict the baby's sex.

## What if cfDNA does not give ANY result?

A small number of pregnancies do not get any result from cfDNA screening. This could happen if the sample does not pass all of the quality control steps at the lab. This does not usually mean a problem with the baby. If there is no result, you can do the test again or choose a different test.

**IT'S YOUR CHOICE. You can decide if you want to learn about chromosome conditions during pregnancy. This test may help you prepare for a child with special needs. You might use the information to decide whether or not to continue the pregnancy. But not everyone wants to know about chromosome conditions.**

## More about…

**Down syndrome** is a chromosome condition that includes mild to moderate intellectual disability and a characteristic appearance. Babies with Down syndrome also have a higher chance to be born with physical birth defects, such as a heart defect or intestinal problem.

**Trisomy 18 and trisomy 13** are two different chromosome disorders that include severe intellectual disability and medical problems in many parts of the body. For either condition, survival beyond the first year of life is uncommon.