

Cell-Free DNA(cfDNA) Screening Result

Screen positive (high risk) for Down syndrome

As part of your prenatal care, you had a blood test to screen for chromosome conditions in the pregnancy.

Your cfDNA screening result is:

SCREEN POSITIVE for DOWN SYNDROME

What does this result mean?

This result means that your blood test showed more DNA from chromosome 21 than expected. This is usually because the baby has Down syndrome, but more testing is needed to be sure.

What is Down syndrome (trisomy 21)?

Down syndrome is a genetic condition that causes physical differences and intellectual delays due to an extra chromosome 21. Chromosomes are the packages of genetic information found in every cell of the body. About 1 in 2 (50%) babies with Down syndrome are born with a heart defect. However, each person with Down syndrome is different, and not everyone with this condition will have serious health problems.

What can I do next?

You will be offered an ultrasound and diagnostic testing. Ultrasound may show extra fluid at the back of the baby's neck, extra fluid in the brain, a heart defect, or other signs of Down syndrome. Some of these differences are easier to see in the second trimester of pregnancy, but there are limits to how much an ultrasound can see or predict for the baby. Many babies with Down syndrome have a normal ultrasound.

A diagnostic test is needed to know for sure if the baby has Down syndrome. Testing during pregnancy is considered safe when done by a specially trained doctor. The risk of miscarriage due to a prenatal diagnostic test is very low (less than 1 in 1000). Results are ready in about 14 days. You could also choose to wait and test the baby after delivery.

POSSIBLE REASONS FOR THIS RESULT

The most likely reason for this result is that the baby has Down syndrome.

However, it is possible that this is a "false positive" result.

With any screening test, some results are false positive. This means the test can show a high risk even though the baby does not have Down syndrome. We do not usually know the reason for a false positive result.

Some false positive results are caused by:

- **Confined placental mosaicism (CPM)**
This happens when part of the placenta has cells with three copies of chromosome 21. These cells are only in the placenta and not in the baby.
- **Early twin demise**
This happens when a twin with Down syndrome was lost very early in the pregnancy.
- **Technical interference**
This happens when a mother's DNA has harmless differences that cause an over-estimate of chromosome 21.
- **Sample variability**
This is an incorrect result with no clear biological reason.

PRENATAL DIAGNOSTIC TESTS - TESTS DURING PREGNANCY THAT DIAGNOSE CHROMOSOME CONDITIONS

Chorionic Villus Sampling (CVS)

This diagnostic test can be done **between 10 and 14 weeks**. A small sample of the placenta is taken without touching the baby. The sample includes cells that usually have the same chromosomes as the baby. The chromosomes in these cells can be examined.

Amniocentesis

This diagnostic test is usually done **between 15 and 22 weeks**, but it can be done later. A small amount of fluid is taken from the uterus (womb) without touching the baby. The fluid has cells from the baby. The chromosomes in these cells can be examined.

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What if Down syndrome is confirmed?

You will be offered support and information. A doctor or genetic counselor will talk with you about Down syndrome. When this condition is found during pregnancy, the options of continuing or ending the pregnancy are discussed. You may be referred to a high-risk pregnancy doctor to discuss pregnancy care and delivery plans, especially if a heart defect is seen by ultrasound. There are also support resources for parents and babies with Down syndrome.

More about...

Down syndrome (trisomy 21)

Down syndrome is a genetic condition caused by an extra chromosome 21 in some or all of a person's cells. It is also called trisomy 21. Down syndrome usually happens randomly and does not run in the family. It is not caused by anything parents have or have not done. Anyone can have a baby with Down syndrome, but the chance is higher if you are older when you get pregnant. About 1 in every 700 babies is born with Down syndrome.



Facts about Down syndrome:

- All children with Down syndrome have delayed development and some amount of intellectual disability. It varies from person to person, but is usually in the mild to moderate range.
- A few common physical traits of Down syndrome are: low muscle tone, slow growth, short height, and a typical facial appearance.
- Certain medical conditions happen more often with Down syndrome, like heart disease, thyroid disease, hearing loss, and vision problems. Many of these conditions are treatable, so people with Down syndrome can lead healthy lives.
- There is a higher rate of miscarriage and pregnancy loss when a fetus has this condition.
- Life expectancy for people born with Down syndrome is in the 50's and 60's.
- For more facts about Down syndrome, visit the National Institute of Health (NIH) website:
 - Down syndrome: <https://rarediseases.info.nih.gov/diseases/10247/down-syndrome>

Support resources: [National Down Syndrome Society](#)
[Down Syndrome Connection of the Bay Area](#)