

Cell-free DNA (cf-DNA) Screening

No Result after Repeat Testing

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

Your result shows:

NO RESULT after REPEAT TESTING

Why did repeat testing not give a result?

There are many reasons cfDNA screening may not give a result. About 1 in 3 (33%) **repeat** cfDNA samples come back with no result. That means whatever caused no result the first time is still interfering with the test. This is unlikely to be a lab mistake. Sometimes it is because there is not enough fetal DNA in the sample, but there are other reasons the test may not work. Usually, we do not know exactly why repeat testing failed.

Does this mean there is a problem with the baby?

Usually not...most pregnancies that get no result from cfDNA screening are healthy. However, getting no result may increase the possibility of a chromosome condition in the baby by a small amount.

What can I do next?

When you are unable to get a result from cfDNA screening, you will be offered other ways to test the baby for chromosome conditions. You may choose one or more of the following:

- **Take a different screening test** – You could have AFP4 (quad marker) screening. This is a general screening test for birth defects. It screens for Down syndrome and trisomy 18. Other chromosome conditions may be identified. It also screens for neural tube defects (an opening in the spine). In twin pregnancies, ultrasound is done to screen for trisomy 18 and other conditions.
- **Have an ultrasound** – Ultrasound can look for physical differences in the baby and other signs of chromosome conditions. Ultrasound can be scheduled at any time in pregnancy, but is usually done between 18 to 22 weeks in pregnancy. There are limits to how much an ultrasound can see or predict for the baby. Some things are easier to see in the second trimester.
- **Have a diagnostic test** – A diagnostic test, like CVS or amniocentesis, is the only way to know for sure if a baby has a chromosome condition. These tests have a very small risk (about 1 in 1000) for miscarriage.
- **Test the baby after birth** – You can request chromosome testing after you deliver. This is done on a small blood sample from the baby.

Possible Reasons for this result:

- **Early embryo (fetal) loss**
The loss of an embryo can happen before the first ultrasound and may not be detected. When an embryo is lost very early in the pregnancy, DNA from the lost embryo can interfere with the test.
- **Technical interference**
A pregnant person's DNA may have harmless differences that interfere with the test
- **Not enough fetal DNA**
The sample may not have enough fetal DNA to give a reliable result. This is more likely to happen in a person with high body weight, when the placenta is small, or in an IVF or twin pregnancy.
- **Chromosome conditions**
Some chromosome conditions interfere with the test and lead to no result.