KAISER PERMANENTE®

Cell-Free DNA (cfDNA) Screening Result

Screen Positive (high risk) for trisomy 13

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 TRISOMY 13

What does this result mean?

This result means that your blood test showed more DNA from chromosome 13 than expected. This could be because the baby has trisomy 13, but more testing is needed to be sure.

What is trisomy 13?

Trisomy 13 is a genetic condition that causes severe intellectual and physical disabilities due to an extra chromosome 13. Chromosomes are the packages of genetic information found in every cell of the body. Babies with trisomy 13 often have birth defects of the brain, face, and heart. Pregnancies with trisomy 13 usually end in miscarriage or loss of the baby soon after birth. However, some babies with trisomy 13 live longer.

What can I do next?

You will be offered an ultrasound and diagnostic testing. Ultrasound can look for signs of trisomy 13. If the baby has trisomy 13, ultrasound may show that the baby is not growing well or find birth defects related to the condition. 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.

POSSIBLE REASONS FOR THIS RESULT

It is possible that the baby has trisomy 13.

However, many times this is a "false positive" result. A false positive result is when the test shows a high risk for trisomy 13, but the baby does not have this condition. We do not usually find 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 13. These cells are only in the placenta and not in the baby.
- **Early twin demise**
  This happens when a twin embryo with trisomy 13 is lost very early in the pregnancy.
- **Technical interference**
  This can happen when your DNA has harmless differences that cause an over-estimate of chromosome 13.
- **Sample variability**
  This is an incorrect result with no clear biological reason.

A diagnostic test is needed to know for sure if the baby has trisomy 13. This type of test also finds other chromosome conditions. 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 (about 1 in 1000). Results are usually ready within 2 weeks.

Follow-up testing is optional. You can decline testing, or you can choose to wait until after delivery to have testing.

PRENATAL DIAGNOSTIC TESTS - Tests that diagnose chromosome conditions during pregnancy

Chorionic Villus Sampling (CVS)
CVS is done **between 10 and 14 weeks**. A small sample of the placenta is taken using a thin needle through the belly or a flexible tube through the vagina - without touching the baby. The sample includes cells that usually have the same chromosomes as the baby. The chromosomes in these cells can be studied.

Amniocentesis
Amniocentesis is usually done **between 15 and 22 weeks**. A small amount of fluid from the uterus (womb) is taken using a thin needle through the belly - without touching the baby. The fluid has cells from the baby. The chromosomes in these cells can be studied.

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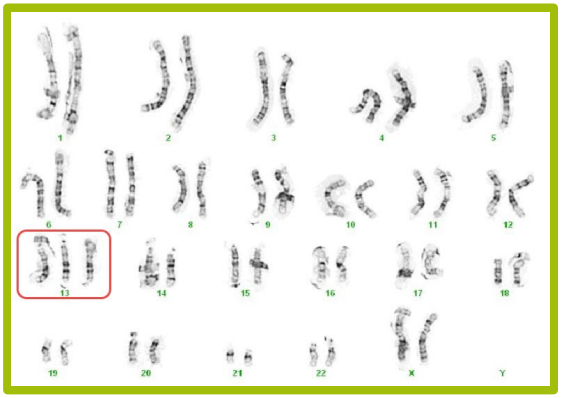
What if trisomy 13 is found?

A doctor or genetic counselor will talk with you about trisomy 13. You will be offered support and information. 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. There are also support resources for parents and babies with trisomy 13.

More about...

Trisomy 13 (Patau syndrome)



Trisomy 13 is a genetic condition caused by an extra chromosome 13 in some or all of a person's cells. Trisomy 13 usually happens randomly and does not run in the family. It is not caused by anything parents have done. Anyone can have a baby with trisomy 13, but the chance is higher as a person gets older. About 1 in 10,000 babies is born with trisomy 13.

More than 9 out of 10 (90%) of babies with trisomy 13 have findings on a prenatal ultrasound. The ultrasound may show differences in the fetal brain, heart, hands, or other parts of the body. These differences are easiest to see around 20 weeks or later in pregnancy. Most babies with trisomy 13 are miscarried or stillborn. Babies that are born alive may live for a few hours, days, or weeks. A small number of children survive to their first birthday and beyond. All children with trisomy 13 have very delayed development and usually need frequent doctor visits for ongoing medical care.

There is no cure for trisomy 13. Medical care focuses on treating symptoms, such as breathing problems (apnea), feeding problems, seizures, and heart conditions.

Support resources:

SOFT – Support Organization For Trisomy