KAISER PERMANENTE®

Cell-Free DNA (cfDNA) Screening Result
Screen positive (high risk) for trisomy 18

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 18

What does this result mean?

A screen positive result for trisomy 18 happens when the blood test shows more DNA from chromosome 18 than expected. This may be because the baby has trisomy 18, but more testing is needed to be sure.

What is trisomy 18?

Trisomy 18 is a genetic condition that causes severe intellectual and physical disabilities due to an extra chromosome 18. Chromosomes are the packages of genetic information found in every cell of the body. Babies with trisomy 18 often grow slowly during pregnancy and are small at birth. This condition includes birth defects in many parts of the body. Babies with trisomy 18 usually die before birth or shortly after birth, but some children live longer.

What can I do next?

You will be offered an ultrasound and diagnostic testing. Ultrasound looks for physical signs of trisomy 18. Ultrasound may show that the baby is not growing well or find differences in the baby's heart, brain, or hands and feet. 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

The most likely reason for this result is that the baby has trisomy 18.

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 trisomy 18. 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 18. These cells are only in the placenta and not in the baby.
- **Early twin demise**
  This happens when a twin embryo with trisomy 18 is lost very early in the pregnancy.
- **Technical interference**
  This happens when your DNA has harmless differences that cause an over-estimate of chromosome 18.
- **Sample variability**
  This is an inaccurate result with no clear biological reason.

A diagnostic test is needed to know for sure if the baby has trisomy 18. 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 ready within 14 days.

Follow-up testing is optional. You can decide not to have testing or 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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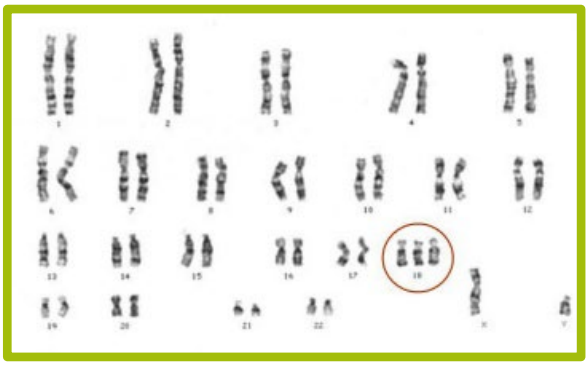
What if trisomy 18 is found?

A doctor or genetic counselor will talk with you about trisomy 18. 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 18.

More about...

Trisomy 18 (Edward syndrome)



Trisomy 18 is a genetic condition caused by an extra chromosome 18 in some or all of a person's cells. Trisomy 18 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 18, but the chance is higher if you are older when you get pregnant. About 1 in 5000 babies is born with trisomy 18.

More than 9 out of 10 (90%) babies with trisomy 18 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. The most common findings are very slow growth and heart defects. Many babies with trisomy 18 are miscarried or stillborn. Babies born with trisomy 18 may live for a few hours, days, or weeks. Some children survive to their first birthday and beyond, with medical support. All children with trisomy 18 have very delayed development and usually need frequent doctor visits for ongoing medical care.

There is no cure for trisomy 18. Medical care focuses on treating symptoms, such as breathing problems (apnea), feeding problems, heart conditions, and infections.

Support resources:

SOFT – Support Organization For Trisomy
Trisomy 18 Foundation