Non-invasive Prenatal Testing (NIPT) Result
High Probability for Trisomy 18

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

Your NIPT result shows:
HIGH PROBABILITY for TRISOMY 18

What does this result mean?
A high probability 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 can look for 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.

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 – much lower than 1%. Results are ready within 14 days.

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

POSSIBLE REASONS FOR THIS RESULT
The most likely reason for this result is that the baby has trisomy 18. More than 90% of women with this result are carrying a baby with trisomy 18.

However, there is a small chance for a “false positive” result. A false positive result is when the test shows a high risk for trisomy 18, but the baby does not have this condition. We do not usually know the reason for a false positive result.

False positive results can be 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 with trisomy 18 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 18.
- 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. A small amount of fluid from the uterus (womb) is taken without touching the baby. The fluid has cells from the baby. The chromosomes in these cells can be examined.
**Non-invasive Prenatal Testing Result**

**High Risk for Trisomy 18**

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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. There are also support resources for parents and babies with trisomy 18.

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**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. Anyone can have a baby with trisomy 18, but the chance is higher as a woman gets older. Trisomy 18 usually happens randomly and does not run in the family. It is not caused by anything parents have or have not done. About 1 in 5000 babies is born with trisomy 18.

The most common features are very slow growth and heart defects. There may also be other birth defects, such as abnormal kidneys or brain differences. Many babies with trisomy 18 are miscarried or stillborn. Babies born with trisomy 18 may live for a few hours, days, or weeks. A small number of children survive to their first birthday and beyond, with medical support. All children with trisomy 18 have delayed development and intellectual disability.

There is no cure for trisomy 18. Treatment focuses on symptoms of the condition, such as breathing problems (apnea), feeding problems, heart conditions, and infections.

**Support resources:**

[SOFT – Support Organization For Trisomy 18 Foundation](#)