KAISER PERMANENTE®

Non-invasive Prenatal Testing Result

High probability (high risk) for a sex chromosome condition

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 a SEX CHROMOSOME CONDITION

What does this result mean?

This result means that your blood test showed a difference in the expected amount of the X or Y chromosome. This could be because the baby has a sex chromosome condition (SCA). However, this is not a direct test of the baby's chromosomes. More testing is needed to know for sure.

What are SCAs?

SCAs are a group of genetic conditions caused by differences in the number of X or Y chromosomes. These conditions happen randomly and are not inherited. About 1 in every 500 newborns has an SCA. The X and Y chromosomes are called sex chromosomes because they help determine the fetal sex. However, a difference in the number of sex chromosomes does not change the predicted sex of the baby. The sex organs and assigned gender typically match the predicted sex. Changes in the sex chromosomes may affect learning, height, and fertility. Some babies may need extra care during pregnancy or after delivery. There are three common SCAs: *Turner syndrome, Klinefelter syndrome, and trisomy X*. Your result shows which SCA is suspected in your pregnancy.

POSSIBLE REASONS FOR THIS RESULT

It is possible that the baby has a sex chromosome condition.

It is also 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 a sex chromosome condition. We do not usually know the reason for a false positive result.

False positive results can be caused by:

- **Confined placental mosaicism**
  This happens when part of the placenta has cells with a different number of X chromosomes. These cells are only in the placenta and not in the baby.
- **Early twin demise**
  This happens when a twin with an SCA was lost very early in the pregnancy.
- **Technical interference**
  This happens when a pregnant person has harmless genetic differences or an SCA that interferes with the estimate of the sex chromosomes.
- **Sample variability**
  This is an incorrect result with no clear biological reason.

What can I do next?

You will be offered prenatal diagnostic testing. A diagnostic test is needed to know for sure if the baby has an SCA. 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 500). Results are ready within 14 days.

Ultrasound (sonogram) may be offered in some cases. A baby with Turner syndrome may show findings on ultrasound. Ultrasounds are usually normal in a baby with Klinefelter syndrome or trisomy X.

All follow-up testing is optional. If you want, you can choose to wait until after you deliver to check the baby's chromosomes or decline any further testing.

PRENATAL DIAGNOSTIC TESTS - Tests that diagnose chromosome conditions during pregnancy

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

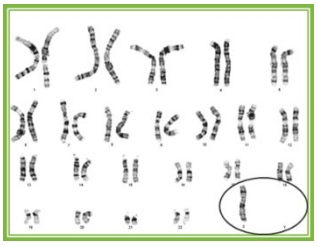
This diagnostic test is usually done **between 15 and 22 weeks**. A small amount of fluid from the uterus (womb) is taken without touching the baby. The fluid has cells from the baby. The chromosomes in these cells can be examined.

What if a sex chromosome disorder is found?

A doctor or genetic counselor would talk with you about the specific sex chromosome disorder. Each condition has its own set of medical issues. When the condition is found during pregnancy, options for continuing or ending the pregnancy are discussed as part of counseling. There are also many support resources for parents and individuals with a sex chromosome disorder.

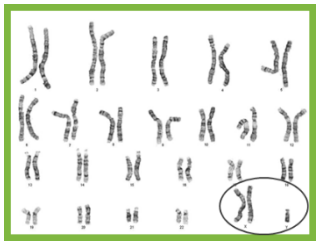
More about...

Turner syndrome (Monosomy X)



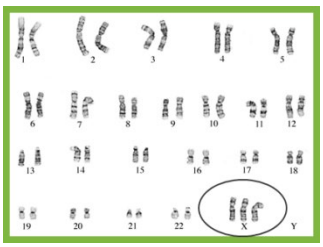
Turner syndrome is a genetic condition that happens when there is just one X chromosome in some or all of a person's cells. The predicted sex is female. The most common features in Turner syndrome are short stature, puberty problems and infertility. People with this condition have normal intelligence, but learning delays are possible. Many pregnancies with Turner syndrome miscarry. Birth defects, such as heart defects and malformed kidneys, happen more often in babies with Turner syndrome.

Klinefelter syndrome (XXY)



Klinefelter syndrome is a genetic condition that happens when there are two X chromosomes and one Y chromosome. The predicted sex is male. Children with this condition are often taller than average. Motor skills like sitting and walking may happen slightly later than usual. Learning disabilities, like speech delays, are also more common. The extra copy of the X chromosome also causes low testosterone (the male sex hormone). Most adults with this condition are infertile due to poor sperm production.

Trisomy X or Triple X syndrome (XXX)



Trisomy X (or triple X syndrome) is a genetic condition that happens when there are three X chromosomes. The predicted sex is female. Learning and behavior issues are more common, but vary from person to person. People with this condition tend to be tall for their age. This condition does not usually affect puberty or fertility. Most individuals with trisomy X never know they have this condition unless special testing is done.