Non-invasive Prenatal Testing Result
High Risk for a Sex Chromosome Disorder

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

Your NIPT result shows “HIGH RISK” for a SEX CHROMOSOME DISORDER

What are sex chromosome disorders?
Sex chromosome disorders are a group of genetic conditions caused by changes in the number of chromosomes that determine the fetal sex. These disorders happen randomly and are not inherited. About 1 in every 500 newborns has a sex chromosome disorder. There are three common sex chromosome disorders: Turner syndrome, Klinefelter syndrome, and trisomy X. A difference the number of sex chromosomes does not affect the sex of the baby. The sex organs and gender typically match the expected sex. However, changes in the sex chromosomes may affect learning, height, and fertility. And some babies with Turner syndrome may need extra care during pregnancy or after delivery.

Does “High Risk” mean that the baby really has a sex chromosome disorder?
Not always…many women with this result have babies with normal chromosomes. However, it does mean your baby has a much higher chance to have a sex chromosome disorder than the typical pregnancy. Your NIPT result estimates how many X and Y chromosomes the baby has, but this is not a direct test of the baby’s chromosomes.

Why did my test show a high risk for sex chromosome disorders?
NIPT screening looks at tiny pieces of fetal DNA from the placenta as well as your DNA. A high risk result happens when there seems to be either more or less than the expected amount of DNA from the X chromosome. Sometimes this is because the baby has an extra or missing X chromosome, but there are other reasons for this result. The mother or the placenta may have some cells with an atypical number of X’s (either more or less than the usual number). This is fairly common and not usually related to a medical condition or health problem.

What can I do next?
You will be offered diagnostic testing. A diagnostic test can determine whether or not the fetus has a sex chromosome disorder. These tests can also find other chromosome disorders. Results are ready in about 10 to 14 days.

Diagnostic 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. Ultrasound (sonogram) may be offered in some cases. Fetuses with Turner syndrome sometimes show findings on ultrasound. Ultrasounds are usually normal in pregnancies with Klinefelter syndrome or trisomy X.

DIAGNOSTIC TESTING:
PREGNATAL TESTS THAT DIAGNOSE CHROMOSOME DISORDERS

Chorionic Villus Sampling (CVS)
This diagnostic test can be done between 10 and 14 weeks. The procedure removes a small sample from the placenta without touching the fetus. The sample includes cells that have the same chromosomes as the fetus. The chromosomes in these cells can be examined.

Amniocentesis
This diagnostic test can be done between 15 and 22 weeks. The procedure removes a small amount of fluid from the uterus. The fetus is not touched. The fluid has cells from the fetus. The chromosomes in these cells can be examined.
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How safe are the diagnostic tests (CVS and amniocentesis)?
CVS and amniocentesis are considered safe tests when done by a specially trained medical doctor. The risk of miscarriage due to either a CVS or amniocentesis is small – much less than 1%.

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 only happens in females. It is caused by a missing X chromosome in some or all of a person’s cells. The most common features in Turner syndrome are short stature, puberty problems and infertility. Girls 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 only happens in males. It is caused by an extra X chromosome. Boys with this condition are often tall for their age compare to boys with typical chromosomes. 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). This can lead to puberty problems. In addition, most males 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 only happens in females. It is caused by having an extra X chromosome. Girls with this condition are more likely to have learning and behavior issues than girls with typical chromosomes. These girls also 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.

This information is not intended to diagnose health problems or to take the place of medical advice or care you receive from your physician or other health care professional.
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