

# Non-invasive Prenatal Testing Result

## Sex Chromosome Aneuploidy or Fetal Sex Inconclusive

You recently had a blood test to screen for chromosome disorders in the fetus and were told:

**NIPT is unable to provide a clear answer about fetal sex or sex chromosome changes (or both)**

The main purpose of NIPT is to screen for major chromosome conditions (Down syndrome, trisomy 18, and trisomy 13). Your test result shows that your pregnancy is at low risk for these three conditions.

NIPT also includes a study of the sex chromosomes, but a result is not always possible. Babies can be born with a change in the total number of sex chromosomes, such as only one X (X), an extra X (XXX or XXY), or an extra Y (XYY). NIPT checks for the presence or absence of the Y chromosome in the fetus and estimates how many are present. The test also measures how many X chromosomes are present. Most often an extra X or Y does not have a big impact on a baby's development.

### Did you know?

- There are two different sex chromosomes: the X chromosome and the Y chromosome.
- Most males have one X and one Y chromosome (XY). Babies with a Y chromosome develop as a male.
- Most females have two X chromosomes (XX).
- A difference in the number of X and Y chromosomes is called sex chromosome aneuploidy or "SCA".
- Most of the DNA in the NIPT sample comes from the woman being tested. Only a small amount of DNA is from the pregnancy.

**Why was the fetal sex not reported?** The Y chromosome is much smaller than any of the other chromosomes being studied. The smaller size means there is less "Y" DNA to examine at the lab. This makes it harder to confirm the presence or absence of the Y chromosome in some cases. The lab is careful to report fetal sex only if the result is very clear. The lab reports the result as "inconclusive" when there is not enough information from the sample to provide a reliable result.

**Why was a "sex chromosome aneuploidy" (SCA) result not reported?** Trying to estimate the number of sex chromosomes from a fetus can be complicated. The Y chromosome is small and can be hard to measure, but the X chromosome can sometimes be even harder to measure. Not only does the fetus have one or two X chromosomes, but there are also X chromosomes from the mother. To complicate things even more, 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. However, it can make it hard to measure how many X chromosomes a fetus has. When the sample does not have clear information to give a reliable result for SCA, the lab reports the result as "inconclusive". There may still be enough information to predict the fetal sex for some pregnancies. Twin pregnancies do not include testing for SCA conditions.

**Does this mean a problem in the baby?**

**Usually not.** While there is still a small possibility for a chromosome condition in the baby, most women who get this result have healthy pregnancies.

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Page 2

Can I take the test again? When the fetal sex or the sex chromosome number cannot be reported, re-testing is not offered. Repeat testing is unlikely to give a reliable result about the sex chromosomes in this situation.

Should other tests be done if fetal sex or sex chromosomes cannot be reported? You could consider having a procedure like chorionic villus sampling (CVS) or amniocentesis to examine the fetal chromosomes. It may also be possible to request a chromosome study after delivery. The decision to have additional testing will depend on the details of your pregnancy and the questions you want answered. You may want to discuss this with your genetic counselor. All pregnancies have a routine ultrasound typically scheduled between 18 to 22 weeks. Ultrasound is often able to predict the fetal sex and checks for physical birth defects. However, ultrasound cannot identify most babies with SCA conditions.

### More about SCA conditions...

About 1 in every 500 newborns has a sex chromosome aneuploidy (SCA) condition. These conditions happen randomly and do not run in the family. The three most common SCA conditions are:

#### Turner syndrome (45,X or Monosomy X)



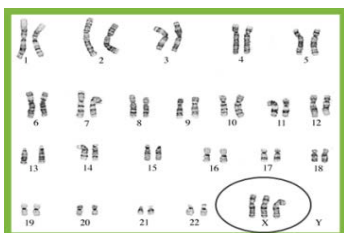
Turner syndrome is a genetic condition that only happens in females. It is caused by a missing X chromosome in some or all of a person's cells. The most common features in Turner syndrome are shorter height, late puberty and infertility. Girls with this condition have normal intelligence, but learning delays are possible. Turner syndrome in a fetus may lead to an early miscarriage. Birth defects, such as heart defects and malformed kidneys, also 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 compared 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 delays in puberty. 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.