

# percept™ NIPT fact sheet

## High risk for XYY

This fact sheet is for women and their partners who receive a high risk result for XYY from the percept™ non-invasive prenatal test (NIPT) offered by Victorian Clinical Genetics Services (VCGS). This information may not be applicable to test results reported by other NIPT service providers.

As part of the service offered by VCGS, women and couples have the opportunity to discuss their result with a genetic counsellor at no additional cost. Genetic counsellors are experts in assisting people to understand genetic test results and make informed decisions about further testing options.

## What does this result mean?

NIPT is a way for women to get an accurate estimate of the chance that their baby has one of the most common chromosome conditions: trisomy 21 (Down syndrome), trisomy 18 and trisomy 13. The test may also detect whether there are extra or missing copies of the sex chromosomes, X and Y.

A high risk result for XYY  
**does not** mean the baby  
definitely has XYY.

A high risk result for XYY indicates the baby has a high chance of having XYY. However, NIPT is a screening test and only a diagnostic procedure (such as chorionic villus sampling (CVS) or amniocentesis) can confirm if the baby has XYY.

## What is XYY?

Usually a male has two sex chromosomes: an X and a Y. Males who have XYY have an X chromosome and have two copies of the Y chromosome.

Most babies with XYY do not have any medical problems at birth. In childhood, boys with XYY may be tall for their age with an increased chance for acne and may have some mild learning, speech or behavioural problems. However, many males with XYY do not have these problems and most males with XYY never know they have the condition. XYY is also sometimes called Jacobs syndrome.

## Possible explanations for this high risk result:

There is a high chance that the baby has XYY. However, the only way to provide a definitive diagnosis is to have a diagnostic procedure (CVS or amniocentesis) with chromosome testing.

In some cases, high risk results for XYY may represent 'false positive' test results. A false positive result means that although NIPT indicates a high risk of XYY, the baby does not have this condition.

## Possible causes of false positive results for XYY from NIPT include:

### Statistical false positive result

This is an incorrect result with no apparent biological cause.

### Maternal DNA variation

Rarely, a high risk result on NIPT may be caused by a variation in the mother's DNA. If this is suspected, the laboratory will suggest further investigations.

### Confined placental mosaicism (CPM)

This is caused by a population of cells in the placenta with an extra copy of the Y chromosome. These cells are only in the placenta and are not present in the baby.

## What are the next steps?

All women who receive an increased risk result for XYY should discuss this with their doctor.

Women should be offered the option of further testing (including testing in pregnancy or testing their baby after birth); however women do not need to have further testing. It is their choice.

Referral for genetic counselling may be helpful in supporting women and their partners with making decisions about further testing options.

## What are diagnostic procedures?

CVS and amniocentesis are diagnostic procedures available during pregnancy that can identify for certain if the baby has a chromosomal condition such as XYY.

**CVS** involves passing a fine needle into the developing placenta (chorion) under ultrasound guidance and drawing a few small fragments of tissue into a syringe. CVS carries a small risk of miscarriage (1 in 500 or 0.2%)\*, generally within two weeks of the procedure. CVS analyses placental tissue. If the cause of the increased risk NIPT result is confined placental mosaicism, there is a chance the woman will need to also have an amniocentesis.

**Amniocentesis** involves the removal of a small sample of fluid from the sac around the developing baby. Under ultrasound guidance, a fine needle is inserted through the mother's abdomen to obtain the fluid. The fluid contains cells that are shed naturally from the fetus. Amniocentesis carries a very small risk of miscarriage (1 in 1000 or 0.1%)\*, generally within two weeks of the procedure.

## What are diagnostic tests?

After the CVS or amniocentesis procedure, chromosome analysis is performed. This can include FISH, karyotype, and microarray. These laboratory tests are considered diagnostic tests.

**FISH:** stands for 'Fluorescence In Situ Hybridisation'. This test looks at whether the cells contain the correct number of chromosomes for the common conditions (chromosomes 13, 18, 21, X, Y). This is a rapid, preliminary test with results available in 1-2 working days.

**Karyotype:** looks at the size, shape and number of chromosomes to determine whether there are any extra, missing or abnormal chromosomes present. Results are usually available in 10-14 days.

**Microarray (or molecular karyotype):** a detailed test that provides more information than the karyotype. This very sensitive test looks for extra or missing segments of DNA in the chromosomes. Results are usually available in 10-14 days.

\*Akolelar R et al. *Ultrasound Obstet Gynecol*, 2015, Jan;45(1): 16-26.

## Contact details

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The information on this fact sheet is based on a comprehensive assessment of outcomes from 15,000 women who have had percept™ NIPT.

Only an invasive diagnostic procedure with chromosome testing can confirm if the baby has XYY.

## Who can I speak to about this result?

It is recommended that women with a high risk XYY result discuss this with their doctor. Their doctor can refer them to speak with a VCGS genetic counsellor at no additional cost.

Alternatively, women and their partners can contact a VCGS genetic counsellor directly on 03 9936 6402.

Genetic counsellors are experts in communicating genetic information and assisting people to understand complex genetic results. They can support women in making informed decisions that are right for them and their family.