

percept™ NIPT fact sheet

High risk for triple X

This fact sheet is for women and their partners who receive a high risk result for triple X 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 triple X **does not** mean the baby definitely has triple X.

A high risk result indicates the baby has a high chance of having triple X. 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 triple X.

What is triple X?

Usually a female has two X chromosomes. Triple X occurs when there is an extra X chromosome. A baby with triple X therefore has three copies of the X chromosome instead of the usual two.

The main effects of triple X are that girls and women with this condition may be taller than average for their family and may experience some learning difficulties.

These learning difficulties are usually mild and do not prevent women with triple X from living a full and normal life.

Possible explanations for this high risk result:

There is a high chance that the baby has triple X. 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 triple X may represent 'false positive' results. A false positive result means that although NIPT indicates a high risk of triple X, the baby does not have this condition.

Possible causes of false positive results for triple X from NIPT include:

Statistical false positive result – this is an incorrect result with no apparent biological cause.

Maternal mosaicism – when the mother has an extra X chromosome in some of the cells in her body.

Confined placental mosaicism (CPM) – this is caused by a population of cells in the placenta with an extra copy of the X 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 triple X 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 have 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.

In some situations, a maternal chromosome test (karyotype) may be recommended to help determine whether triple X in the mother has contributed to the increased risk result. This will be highlighted on the percept™ report where indicated.

Occasionally, a maternal chromosome test may show that the woman herself has triple X. Many women with triple X may have no physical problems and be unaware that they have an additional X chromosome.

What are diagnostic procedures?

CVS and amniocentesis are diagnostic procedures available during pregnancy that can identify for certain if the baby has triple X.

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 high risk percept™ 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

VCGS Prenatal Testing Team
Murdoch Childrens Research Institute
The Royal Children's Hospital,
Flemington Road Parkville 3052

P (03) 9936 6402 **E** perceptNIPT@vcgs.org.au

F (03) 8341 6366 **W** vcgs.org.au/perceptNIPT

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 triple X.

Who can I speak to about this result?

It is recommended that women with a high risk triple X 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.