

# percept™ NIPT fact sheet

## High risk for Monosomy X

This fact sheet is for women and their partners who receive a high risk result for monosomy X from the percept™ non-invasive prenatal test (NIPT) offered by the 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 their partners 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 monosomy X **does not** mean the baby definitely has monosomy X.

Most women who have a high risk result for monosomy X do not have a baby with this condition. NIPT is a screening test and only a diagnostic procedure (such as chorionic villus sampling (CVS) or amniocentesis) can confirm if the baby has monosomy X.

### What is monosomy X?

Usually a female has two X chromosomes (XX) and a male has one X and one Y chromosome (XY). Monosomy X, also called Turner syndrome, occurs when only one X chromosome is present. Babies with monosomy X are female. Intellectual capabilities of girls with monosomy X are usually within the normal range. The main effects of monosomy X are that girls and women may be shorter than average for their family and are usually infertile. Monosomy X may also cause problems with the heart and kidneys. Treatment options are available to help girls and women with monosomy X reach their potential in all aspects of life.

There is an increased chance of miscarriage in pregnancies where the baby has monosomy X.

### Possible explanations for this high risk result:

There is a chance that the baby has monosomy X however, only about 1 in 5 (or 20%) high risk monosomy X results are associated with a true finding of monosomy X in the baby.

The only way to provide a definitive diagnosis is to have a diagnostic procedure (CVS or amniocentesis) with chromosome testing.

High risk results for monosomy X are often 'false positive' test results. A false positive result means that although NIPT indicates a high risk of monosomy X, the baby does not have this condition.

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

#### Statistical false positive result

This is an incorrect result with no apparent biological cause.

#### Maternal mosaicism

When the mother has a population of cells in her body with one X chromosome instead of the usual two.

#### Confined placental mosaicism (CPM)

When there is a population of cells in the placenta with one X chromosome. These cells are only in the placenta and are not present in the baby.

In cases of CPM, the baby may have two X chromosomes (female) or rarely, one X and one Y chromosome (male).

#### Co-twin demise

When one twin was lost earlier in pregnancy due to monosomy X.

## What are the next steps?

All women who receive a high risk result for monosomy 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 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.

### 12 week ultrasound is recommended if not already performed:

- If abnormalities are seen on the ultrasound, it is more likely that the baby has monosomy X; a CVS can be performed at around 12 weeks to provide a definitive result.
- If no abnormalities are seen on the ultrasound, it is more likely that the baby does not have monosomy X, and it is suggested that the woman consider having an amniocentesis at around 15-16 weeks gestation. Amniocentesis will give the most definitive result in these circumstances.

### Maternal chromosome analysis:

- In some situations a maternal chromosome test (karyotype) may be recommended. This will be highlighted on the percept™ report where indicated. Maternal chromosome analysis is done to determine if the high risk monosomy X percept™ result may have been caused by the mother having mosaicism for monosomy X (some cells in her body with one X chromosome). Some women with monosomy X mosaicism may have short stature, experience early menopause and/or may be at higher risk of kidney abnormalities and/or heart problems. Other women will have no medical problems.

## 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 monosomy 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 increased risk NIPT result is confined placental mosaicism, there is a chance the woman will need to also have an amniocentesis. It is for this reason that VCGS recommends waiting for an amniocentesis if the ultrasound is normal.

**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.

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

## Who can I speak to about this result?

It is recommended that women with a high risk monosomy 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.

## 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.

 percept  
cell-free DNA prenatal test

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