Victorian Clinical Genetics Services
Murdoch Childrens Research Institute
The Royal Children's Hospital
Flemington Road, Parkville VIC 3052
P (03) 8341 6201 W vcgs.org.au

# percept™ NIPT fact sheet

## High risk for trisomy 13

This fact sheet is for women and their partners who receive a high risk result for trisomy 13 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 VCGS genetic counsellor at no additional cost. Genetic counsellors are experts in assisting people to understand genetic 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, 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 trisomy 13 **does not** mean the baby definitely has trisomy 13.

Some women who have a high risk result for trisomy 13 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 trisomy 13.

## What is trisomy 13?

Trisomy 13 (also called Patau syndrome) occurs when the baby has three copies of chromosome 13 instead of the usual two. Babies with trisomy 13 have severe intellectual and physical problems. Many pregnancies with trisomy 13 will miscarry and babies that are born with trisomy 13 usually do not live beyond the first few weeks of life.

## Possible explanations for this high risk result:

There is a high chance that the baby has trisomy 13 however, some high risk results for trisomy 13 may be 'false positive' results.

A false positive result means that although NIPT indicates a high risk of trisomy 13, the baby does not have this condition.

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

### Possible causes of false positive results for trisomy 13 from NIPT include:

**Confined placental mosaicism (CPM)**
This is caused by a population of cells in the placenta with three copies of chromosome 13 instead of the usual two. These cells are confined to the placenta and are not present in the baby.

**Statistical false positive result**
This is an incorrect result with no apparent biological cause.

**Co-twin demise**
When one twin was lost earlier in pregnancy due to trisomy 13.

## What are the next steps?

All women who receive a high risk result for trisomy 13 should discuss this with their doctor.

Women should be offered the option of further testing; 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.

Continued overpage...

## Further testing options:

- A 12-13 week ultrasound is recommended if not already performed. However, it is important to be aware that a normal ultrasound at 12-13 weeks does not exclude trisomy 13.
- If abnormalities are seen on the ultrasound, it is more likely that the baby has trisomy 13. A CVS can be performed at 12-13 weeks to provide a definitive result.
- If no abnormalities are seen on the ultrasound, it is more likely that the baby does not have trisomy 13, 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.

# 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 trisomy 13.

**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 suggests waiting for an amniocentesis if the ultrasound is normal. However, the woman should also discuss this option with her doctor, as CVS is still an appropriate procedure if information is wanted earlier in the pregnancy.

**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.

Only an invasive diagnostic procedure with chromosome testing can confirm if the baby has trisomy 13.

# Who can I speak to about this result?

It is recommended that women with a high risk trisomy 13 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

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

P (03) 9936 6402
F (03) 8341 6366
E perceptNIPT@vcgs.org.au
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.

percept® cell-free DNA prenatal test