

# percept™ NIPT fact sheet

## High risk for trisomy 21

This fact sheet is for women and their partners who receive a high risk result for trisomy 21 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 trisomy 21 **does not** mean the baby definitely has trisomy 21.

A high risk result for trisomy 21 indicates there is a very high chance that the baby has trisomy 21. 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 trisomy 21.

### What is trisomy 21?

Trisomy 21 (Down syndrome) is the most common chromosome condition found during pregnancy. It occurs when the baby has three copies of chromosome 21 instead of the usual two.

People with Down syndrome have characteristic physical, medical and intellectual features. Most have some degree of intellectual disability and may have other health and developmental challenges.

Many people with Down syndrome however are able to participate in school, work and social life. It is not possible to predict the level of disability during pregnancy. There is no cure for Down syndrome but early intervention can support development.

### Possible explanations for this high risk result:

The most likely explanation for this high risk result is that the baby has trisomy 21. Rarely, some high risk NIPT results are 'false positive' results. A false positive result means that although NIPT indicates a high risk of trisomy 21, 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 21 from NIPT include:

#### **Confined placental mosaicism (CPM)**

This is caused by a population of cells in the placenta with three copies of chromosome 21 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 21.

### What are the next steps?

All women who receive a high risk result for trisomy 21 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.

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

### 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 21.
- A CVS can be performed at 12-13 weeks to determine if the baby has trisomy 21.
- Alternatively, an amniocentesis at around 15-16 weeks can be performed.

## 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 21.

**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 also need to 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.

Only an invasive diagnostic procedure with chromosome testing can confirm if the baby has trisomy 21.

## Who can I speak to about this result?

It is recommended that women with a high risk trisomy 21 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  
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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.