VCGS
Leaders in Genetic Health

Victorian Clinical Genetics Services
Murdoch Children's Research Institute
The Royal Children's Hospital
Flemington Road, Parkville VIC 3052
P +61 1300 11 8247 W vcgs.org.au

percept™
cell-free DNA prenatal test

What is the percept™ cfDNA prenatal test?

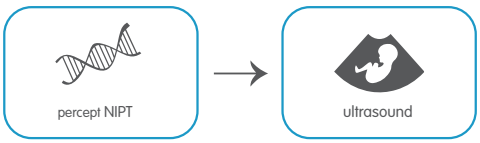
percept™ is an Australian based non-invasive prenatal test (NIPT) offered by Victorian Clinical Genetics Services (VCGS). This test uses cell-free fetal DNA (cfDNA) found in maternal blood to identify pregnancies with an increased chance of chromosome conditions. It is the most accurate type of prenatal screening test for Down syndrome.

percept™ by VCGS is Australasia's most advanced non-invasive prenatal test: *simple, fast, reliable*

When can I have this test?

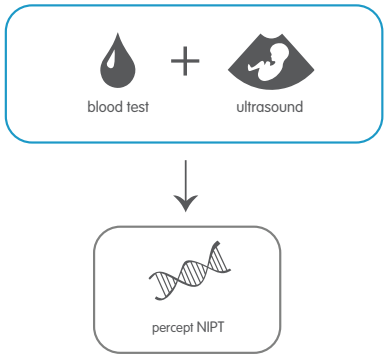
The *percept* blood test can be performed from 10 weeks of pregnancy onwards, as confirmed by an acceptable dating method like ultrasound. Test results are usually available within 3-5 working days from the time we receive the sample (typical courier times from New Zealand are approximately 1-2 working days).

1. You can choose *percept* as your initial screening test from 10 weeks. VCGS recommends that you also consider having a 12-13 week ultrasound to check the growth of your baby.



2. Alternatively, you may choose to have *percept* after combined first trimester screening (CFTS). CFTS includes an ultrasound and a blood test.

If you are not reassured by the results of your CFTS, further screening with *percept* may be helpful.



What will this test tell me?

Conditions screened	Detection Rate
Trisomies	
Down syndrome (trisomy 21)	>99%
Edwards syndrome (trisomy 18)	>98%
Patau syndrome (trisomy 13)	>98%
Rare trisomies	LD
Sex chromosome conditions	
Turner syndrome (monosomy X)	>95%
Klinefelter syndrome (XXY)	LD
Triple X (XXX)	LD
XYY	LD
Partial chromosome changes	
Missing/extra genetic material*	78%
Fetal sex	**Accuracy**
Male XY, Female XX	>99%

*Only large changes detected (>10 Mb) LD = Limited data available

- ✓ Results within 3-5 working days (from lab receipt)
- ✓ Available from 10 weeks of pregnancy
- ✓ IVF pregnancy
- ✓ Egg donor pregnancy
- ✓ Twin pregnancy (trisomies only)

You should discuss your options with your healthcare provider. Genetic counselling is available through VCGS. Please see vcgs.org.au/perceptNIPT for more information.

How does percept™ work?

During pregnancy, some DNA from the placenta crosses into your bloodstream. *percept* tests this cell-free DNA in your blood to look for certain chromosome conditions that may be affecting your pregnancy.

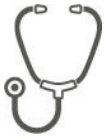
When should I consider diagnostic testing instead of NIPT?

NIPT identifies a limited range of chromosome conditions. In some situations, your doctor may recommend a diagnostic test such as chorionic villus sampling (CVS) or amniocentesis with chromosome analysis. This provides more detailed and accurate information about your pregnancy. These situations may include:

- If the ultrasound shows a fetal abnormality.
- If the combined first trimester screening risk is very high (>1 in 10).
- If the nuchal translucency is increased at the 12 week ultrasound.

NIPT is an advanced screening test but is NOT a replacement for diagnostic testing through CVS or amniocentesis.

How do I arrange this test?



Discuss *percept* with your genetic counsellor, doctor or NIPT provider.

A test request form will be completed by your genetic counsellor, doctor or NIPT provider.

Have your blood collected. See the VCGS website for collection sites.

Most results available within 3-5 working days (from receipt of sample).

How are percept™ results reported?

Low risk

Most results are reported as low risk. This means it is very unlikely (less than 1 in 10,000 chance) that your pregnancy is affected by the specific conditions screened. However, other conditions or abnormalities may still be present. We recommend all patients have an 18-20 week ultrasound.

High risk

A high risk result means that there is an increased chance of the chromosome condition in your pregnancy. **CVS or amniocentesis with chromosome analysis is strongly recommended to confirm a high risk result.** In some cases the diagnostic test will show your baby is not affected by the condition. In this case, the NIPT result is considered a 'false-positive' result.

No result

In some cases, no result is obtained. This is very uncommon (<1% of samples). If this occurs the laboratory will request a repeat specimen for testing at no additional charge.

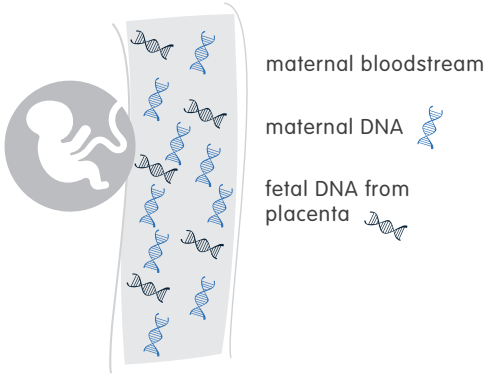
Contact details

VCGS is a not-for-profit provider of a comprehensive range of clinical and laboratory genetics services. We can provide pre-test genetic counselling for *percept* and will liaise with your doctor or genetics service for any high risk results.

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

P +61 1300 11 8247 W vcgs.org.au/perceptNIPT
F +61 3 8341 6366 E perceptNIPT@vcgs.org.au

We're Australian & not-for-profit – all proceeds support medical research.



What is Down syndrome (trisomy 21)?

Down syndrome is the most common chromosomal condition seen in children and adults. It is caused by an extra copy of chromosome 21. People with Down syndrome have some degree of intellectual disability and may have other health and developmental challenges.

What are Edwards syndrome (trisomy 18) & Patau syndrome (trisomy 13)?

Edwards & Patau syndromes are less common than Down syndrome and pregnancies with these conditions usually miscarry. Babies that are liveborn rarely survive long and will have significant physical and intellectual problems.

What are rare trisomies?

Rare trisomies are caused by three copies of any chromosome other than 21, 18, 13, X or Y. These are rarely seen at birth and can be associated with an increased chance of an early miscarriage, growth problems in the fetus and premature delivery. Babies that are liveborn may have a chromosomal condition.

What are sex chromosome conditions?

The X and Y chromosomes are called the sex chromosomes because they determine if a baby will be male or female. Sometimes there are extra or missing copies of these chromosomes. Development may be affected in a variety of ways, depending on the chromosome condition.

What are partial chromosome changes?

In rare cases, large parts of the chromosomes may be missing or extra. These changes will be reported as additional findings as they can be associated with health concerns for your baby.

NOTE: *percept* may be used to screen a pregnancy where one parent is a known translocation carrier. Testing only performed with prior arrangement.

What does percept™ cost?

This test is not covered by the National Screening Program or private health insurance. Fees are payable on the day of blood collection. Please contact your provider for current pricing.

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