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™ cell-free DNA prenatal test

What is the percept™ cfDNA prenatal test?

percept™ is an Australian non-invasive prenatal test (NIPT) offered by the Victorian Clinical Genetics Services (VCGS). This test uses cell-free fetal DNA (cfDNA) found in maternal blood to identify the most common chromosome conditions seen in newborns. This test is the most accurate prenatal screening test for Down syndrome.

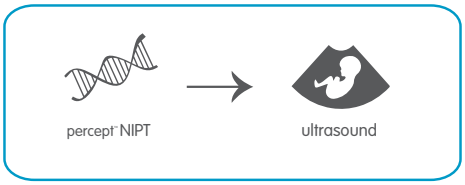
The percept™ blood test can be performed any time from 10 weeks of pregnancy, as confirmed by a dating scan. You will get your test results quickly (within 3-5 working days) because your blood sample is not shipped to an overseas laboratory.

percept™ by VCGS is an Australian non-invasive prenatal test: simple, fast, reliable.

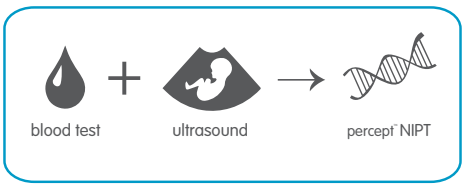
When can I have this test?

percept™ is a highly accurate test for identifying pregnancies at increased risk of chromosome conditions such as Down syndrome.

1. You can choose percept™ as your first screening test. VCGS recommends that you also consider having a 12 week ultrasound to check the structural development of your baby.



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



If you are not reassured by the results of your CFTS, further screening with percept™ may help with your decision making.

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%
Sex chromosome conditions	
Turner syndrome (monosomy X)	>95%
Klinefelter syndrome (XXY)	LD*
Triple X (XXX)	LD*
XYY syndrome	LD*
Gender	**Accuracy**
Male XY, Female XX	>99%

*Limited data available

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

You should discuss your options with your doctor or with one of our genetic counsellors at VCGS.

Visit 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 identify certain chromosome conditions in your pregnancy.

When should I consider invasive testing?

percept™ only detects a limited range of chromosome conditions. In some situations, your doctor may recommend an invasive test such as chorionic villus sampling (CVS) or amniocentesis. These tests will give more detailed and accurate information about your pregnancy. These situations include:

- If your ultrasound finds a structural abnormality.
- If a combined first trimester screening risk is very high (>1 in 50).
- If the nuchal translucency is very large at the 12 week scan.

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

How do I arrange this test?

Discuss percept™ with your genetic counsellor or doctor.

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

Have your blood sample collected. For collection sites, see the VCGS website.

Most results will be available from your genetic counsellor or doctor within 5 working days (from receipt).

How are percept™ results reported?

Low risk

Most results are reported as low risk. This means it is very unlikely your pregnancy is affected by the specific conditions screened for. However, other conditions or abnormalities may still be present. We recommend all patients have an 18-20 week ultrasound.

High risk

A high risk or intermediate (borderline) risk result means that your pregnancy is at increased risk for the chromosome condition reported. **CVS or amniocentesis is strongly recommended to confirm a high or intermediate risk result.** These diagnostic tests identify women with a 'false positive' finding (meaning their high risk screening result was incorrect and their pregnancy is not affected).

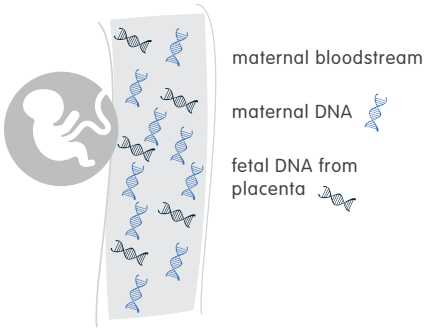
No result

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 provide genetic counselling support before and after testing.

VCGS Prenatal Testing Team
Murdoch Childrens Research Institute
The Royal Children's Hospital, Flemington Road Parkville 3052
P (03) 8341 6201 F (03) 8341 6366
W vcgs.org.au/perceptNIPT E perceptNIPT@vcgs.org.au



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 is Edwards syndrome (trisomy 18)?

Edwards syndrome is less common than Down syndrome and is caused by an extra chromosome 18. Pregnancies with trisomy 18 usually miscarry and babies that are liveborn rarely survive for long. All babies with Edwards syndrome have significant physical and intellectual problems.

What is Patau syndrome (trisomy 13)?

Patau syndrome is even less common than Down syndrome and Edwards syndrome. It is caused by an extra copy of chromosome 13. Pregnancies with trisomy 13 usually miscarry and babies that are liveborn are not expected to live. All babies with Patau syndrome have serious health problems.

What are sex chromosome abnormalities?

The X and Y chromosomes are called the sex chromosomes because they determine if a baby will be a boy or a girl. Sometimes there are extra or missing copies of these chromosomes. Normal development is affected in a variety of ways, depending on the chromosome abnormality.

How much does percept™ cost?

The percept™ cell-free DNA prenatal test is $489. This cost is subject to change. Please visit vcgs.org.au/perceptNIPT to confirm.

The test is not covered by Medicare or private health insurance. Fees are payable via credit card (Mastercard and Visa only), with details to be provided on the day of blood collection.

VCGS Leaders in Genetic Health