



Genetic counselling with a VCGS genetic counsellor (03 9936 6402) is available to anybody considering or undergoing this screening test. VCGS genetic counsellors can also discuss other prenatal screening and testing options with you. More information about this test and the chromosome conditions included in the test is available at [vcgs.org.au/perceptNIPT](http://vcgs.org.au/perceptNIPT)

## Patients undergoing percept™ non-invasive prenatal testing should be aware of the following key points:

### Purpose of the test

This test identifies pregnancies at 'high risk' of:

- The common trisomies 21 (Down syndrome), 18 (Edwards syndrome) and 13 (Patau syndrome);
- Conditions caused by too many or too few of the sex chromosomes (X and Y);
- Rare autosomal trisomies (trisomies involving chromosomes other than 21, 18, 13, X and Y).

### Test process

- This test is intended to be performed from the 10<sup>th</sup> week of pregnancy onwards, as determined by a dating ultrasound.
- A sample of your blood will be collected and sent to VCGS who will issue a report to your healthcare provider. Your healthcare provider is responsible for interpreting and explaining your test results. VCGS genetic counsellors are also available to discuss your results with you.
- The test results will include the sex of the pregnancy. If you do not wish to know the sex you can ask your healthcare provider not to disclose it to you. However, if the results show too many or too few of the sex chromosomes, you may not be able to avoid learning the sex of your pregnancy.
- As this is a screening test, it is recommended that all high risk test results are confirmed by chromosome analysis through CVS or amniocentesis.

### Limitations of the test

- This test screens for extra or missing copies of chromosomes in the fetus and is not designed to detect small genetic imbalances, single gene disorders or non-genetic causes of fetal abnormalities.
- Low risk test results do not eliminate the possibility that your pregnancy may have other chromosomal abnormalities, birth defects, or other conditions, such as open neural tube defects. A 'low risk' result does not guarantee a healthy pregnancy or baby.
- As this is a screening test, there is a small possibility that the results could be incorrect. It is possible to receive a high risk result even though a chromosomal abnormality is not present in the fetus. This is called a 'false positive' result. It is also possible that the chromosomal abnormality being tested for could be present even if the result is low risk. This is called a 'false negative' result.
- Some high risk test results may be due to chromosomal changes in the mother. Further testing of the mother may be required in some circumstances.
- The ability of this test to accurately report fetal sex chromosome abnormalities (too many or too few sex chromosomes) is not well known. Incorrect test results may occur more frequently for these conditions.
- For technical and biological reasons, the fetal sex is reported with >99% accuracy (not 100%).
- percept NIPT may be used to screen a pregnancy where one parent is a known translocation carrier. This testing is only performed by prior arrangement.

### Privacy, confidentiality and use of information

- Your test results will be kept confidential. Results will only be released to your healthcare provider, other healthcare providers involved in your medical care, or to another healthcare provider as directed by you, or otherwise as required or authorised by applicable law.
- Collecting information on your pregnancy after testing is part of our laboratory's standard practice for quality purposes and test evaluation. VCGS may contact your healthcare provider to obtain this information.

### Retention and use of samples

- In line with best practices and clinical laboratory standards, leftover de-identified specimens (unless prohibited by law), de-identified genetic material, as well as other information learned from your testing, may be used by VCGS for purposes of quality control, laboratory operations, laboratory test development, laboratory improvement, and generation of new scientific knowledge. All such uses will be in compliance with applicable law.

### Financial responsibility statement

- You are responsible for fees incurred with VCGS for services performed.