

How do I have the test

We currently send NIPT blood samples to the United States for analysis.

Women need to be seen in our Fetal Medicine clinic to discuss the test in more detail and to perform an ultrasound scan before taking the blood sample to send to the United States.

It takes approximately 7-10 working days to get the test result. A 'low-risk' result is very reassuring. If the result is 'high-risk' we recommend amniocentesis to confirm the findings.

What is the cost of this test

This test is not currently covered by the public health system. The cost is approximately \$350/\$400 and this is payable to the lab directly.

Important things to remember

- NIPT is not the same as amniocentesis. NIPT does not have the risk of miscarriage, but some chromosomal abnormalities that would be detected by amniocentesis are not detected by NIPT.
- 1-2% of samples sent for NIPT cannot be analysed. This is because the level of fetal DNA in maternal blood is too low to allow accurate counting. Women in this group will not get a result from the test.
- NIPT compares fetal and maternal DNA levels. In rare circumstances, previously unrecognised maternal chromosomal re-arrangements have been found during testing.
- More information regarding prenatal screening and diagnosis is available at: <http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/non-invasive-prenatal-testing-nipt>

Contacts

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How to get to RPA Hospital



 The hospital is a 10-minute walk from most bus stops and 20-30 minute walk from major train stations

 Plan your public transport travel. Timetable, fare, wheelchair access details contact the Transport Infoline 131500 www.131500.com.au

 Newtown Station is a 20 minute walk along King St and Missenden Rd. Redfern Station is a 30-minute walk through Chippendale and Sydney University

 Buses run between the city and inner west from Central Station



Chromosomal abnormalities

Our genetic information is stored inside the cells of our body in a very orderly manner. The *genes* that tell our cells how to function are made up of DNA and this information is organised into chromosomes. Most individuals have 23 pairs of chromosomes- one of each pair is inherited from each parent. The pairs are numbered 1-22 (from biggest to smallest) then the final pair determine gender- women have two X chromosomes, men one X and one Y.

About 1 in 200 babies have a chromosomal abnormality. The most common chromosome abnormality is Down syndrome, which is caused by an extra chromosome 21 (*Trisomy 21*). Other common trisomies are caused by an extra chromosome 18 (*Trisomy 18*) or an extra chromosome 13 (*Trisomy 13*). Both these conditions are more severe than Down syndrome. Other types of chromosomal abnormality may also occur but are less common. In some cases the 'sex' chromosomes (X and Y) are affected.

Prenatal testing

Women have the option of testing to see whether or not their baby is affected with Down syndrome or another chromosome abnormality. This commonly involves *combined first trimester screening* - where the risk of a chromosomal abnormality is based on maternal age, ultrasound assessment of fetal nuchal translucency (NT) and measurement of proteins produced by the placenta; PaPP-A, free- β hCG and PIGF. This test is very effective - producing a high risk group that includes only 5% of all women but 90% of the fetuses affected by Down syndrome.

A diagnostic test - CVS or amniocentesis - is then available for women with an increased risk result on screening to definitively inform them whether or not their baby has Down syndrome. This diagnostic test carries a 0.2% risk of causing miscarriage.

Non-invasive Prenatal testing

Non-invasive prenatal testing (NIPT) is a new test that can tell women whether their baby has Down syndrome, Trisomy 18 or Trisomy 13. It has a high degree of accuracy and avoids the risk of miscarriage.

NIPT works by counting pieces of DNA found in the mothers' blood. During pregnancy some of this DNA comes from the fetus. By making millions of counts, the test is able to see small changes in the relative amounts of DNA that occur if there is a chromosomal abnormality.

The test detects 99% of babies that have Down syndrome. Less than 1% of women with a normal pregnancy will be identified in this high risk group (a false positive result). In other words, for detection of pregnancies affected by Down syndrome, NIPT is almost as effective as amniocentesis but does not carry the risk of this procedure.

Who should have NIPT?

We are recommending that women still have combined first trimester screening (11-13 weeks) and then consider NIPT on the basis of their result.

NIPT is not a replacement for combined first trimester screening - because there are several aspects of the scan that will not be covered by this blood test. Similarly, measurements of the biochemical markers (PaPP-A, PIGF and free- β hCG) have some value in screening for other pregnancy abnormalities apart from chromosomal problems.

NIPT is best suited to women whose combined first trimester screening risk is between 1 in 50 and 1 in 2500. Some women who have a risk in this range may want to proceed with CVS or amniocentesis. Others may want to get further information from NIPT. A third group may be happy to continue the pregnancy with no further testing.

We advise all women who have a very high risk (>1 in 50) from combined first trimester screening to consider CVS or amniocentesis. This is because the chance of finding chromosomal abnormalities other than trisomy 21, 18 or 13 is higher in this group of women.

Women who have a very low risk (<1:2,500) from combined first trimester screening are usually reassured by this and do not want to proceed with NIPT.

Accuracy and limitations

NIPT is designed to detect Down syndrome, Trisomy 18 and Trisomy 13. The test is very effective in identifying Trisomy 21 and Trisomy 18 (99% detection), but less effective at detecting Trisomy 13 (90% detection).

NIPT is also able to determine the baby's gender and detect variation in the sex chromosomes. In most circumstances NIPT will recognise if one of the sex chromosomes is missing or extra. The most common sex chromosome condition is called Turner syndrome (45X) and affects 1 in 2500 female births.

NIPT is **not** the same as a CVS or amniocentesis, which looks at all 46 chromosomes in detail. However chromosome abnormalities other than Trisomy 21, Trisomy 18, and Trisomy 13 are uncommon, affecting less than 1 in 1,000 pregnancies.