

## Second trimester screening – triple test

If you miss combined first trimester screening (done between 11 and 13<sup>+6</sup> weeks) you can have a triple (blood) test at 15-18 weeks. This test uses biochemical markers alone, but is still quite robust, and will identify 70% of affected pregnancies.

Alternatively, you can opt to have the NIPT test, which will identify 99% of babies affected by Down syndrome. This test is not funded by Medicare and costs \$350/\$400 out of pocket expense.

## Increased nuchal translucency/ screening for heart defects

One of the sonographic features examined for Down syndrome screening is also known to be abnormal in fetuses that have other structural problems, such as heart defects. If the NT is increased (broadly speaking above 2.5mm) then we will arrange for you to have an extra scan at 14-15 weeks to check the baby's heart development more carefully.

## Screening for early onset pre-eclampsia

A minority of women develop high blood pressure that may lead to very early (<34 weeks) birth (pre-term birth). This condition is described as pre-eclampsia.

During combined first trimester screening, in addition to measuring PIGF (proteins produced by the placenta), we can assess blood flow to the uterus and your blood pressure. These factors will be used to define the risk of early onset pre-eclampsia.

Women with an increased risk (>1%) will be advised to take 150mg Aspirin every night to reduce the risk of early onset pre-eclampsia. Where necessary, we will also arrange closer follow-up at the hospital antenatal clinic that specialises in this condition.

## Contacts

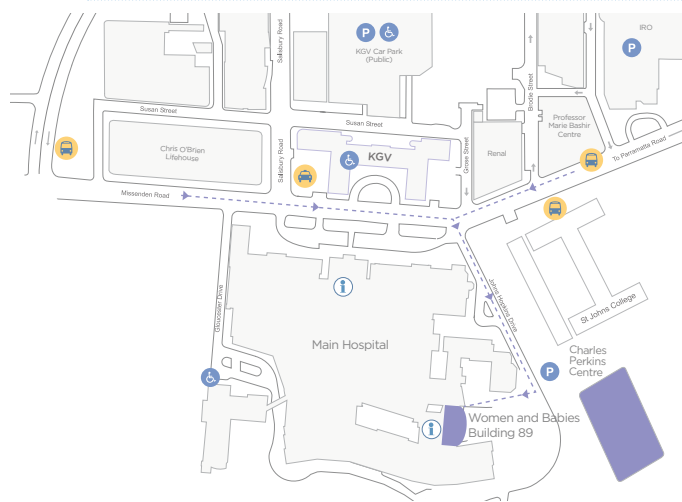
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### Charles Perkins Research Centre

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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](http://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



## The 12 week scan

All pregnant women are routinely offered ultrasound scans at 11-13<sup>+6</sup> weeks and 18-20 weeks to check the development of the pregnancy.

The 11-13<sup>+6</sup> week scan can:

- Confirm your baby's heart is beating.
- Check when your baby is due.
- Find out whether you are having twins.

It is also used to:

- Screen for Down syndrome and other chromosomal abnormalities.
- Detect some major structural problems.
- Screen for pre-eclampsia (high blood pressure that typically develops later in the pregnancy).

## What is Down syndrome?

Down syndrome is a genetic condition associated with moderate intellectual delay. Children with Down syndrome have characteristic facial features and some have problems with the heart or the digestive tract.

Down syndrome is caused by the presence of an extra chromosome (number 21) in all the body's cells. This means there are extra genes and this affects development. There is no cure for Down syndrome, but many symptoms can be treated and with early intervention children with Down syndrome can be helped to reach their potential.

Down syndrome is the commonest form of intellectual delay seen in children. It occurs in all races and cultures at around the same rate. Approximately 1 in 1,000 infants born in New South Wales have Down syndrome.

## What is my risk of having a baby with Down syndrome?

As women get older the chance of having a baby with Down syndrome increases (see chart below).



## Screening tests for Down syndrome

Screening tests aim to identify a small group of pregnancies at higher risk of having Down syndrome or other chromosomal abnormalities. These women can then be offered a diagnostic test (CVS or amniocentesis - refer to the separate brochure for information). At RPA, we normally use the combined first trimester screening test - which uses a number of factors (maternal age, ultrasound factors and biochemical factors) to calculate the risk of a pregnancy being affected.

## Do I want a screening test for Down syndrome?

Not all women want to have a screening test for Down syndrome - as the information would not affect decisions they would make about their pregnancy. This is a very personal decision and we respect the fact that not all couples would want to have this test.

If you want to discuss your options in more detail you could approach your GP or we can arrange for you to see a genetic counsellor.

## Combined first trimester screening

Combined first trimester screening test involves an ultrasound scan and a blood test at 11-13<sup>+6</sup> weeks pregnancy. The ultrasound examines the fluid-filled space at the back of a baby's neck, called nuchal translucency (NT), and the development of the baby's nasal bone (NB). The chance of Down syndrome is higher if the NT measurement is larger and/or if the nasal bone is not readily visible.

The blood test measures the levels of three proteins in the mother's blood: PaPP-A, PIGF and free-βhCG.

Your age, the NT and NB measurement and the blood test results are combined to develop a risk that describes how likely it is that the baby is chromosomally normal or has Down syndrome. This can be presented as a fraction (1 in 500, 1/500), a percentage (0.2%) or a description (low risk or increased risk).

If the adjusted risk is less than 1 in 2500 it is considered very low risk. Most women have a very low risk result. This is generally considered to be reassuring and no further testing is needed. If the risk is between 1 in 300 and 1 in 2500, this is considered low risk. There is no need to do anything but some women may choose to have a new blood test, called NIPT [that is very accurate (>98%) for Down syndrome] for further reassurance. All low risk screening results will be communicated to your GP - who should have a result within seven days of the test being completed. If you want the NIPT test we can arrange this for you. This test costs approximately \$350/\$400. There is a separate information sheet describing this test.

An adjusted risk >1 in 300 (0.3%) is considered an increased risk. 5% of women (1 out of 20) get an increased risk result and may choose to have further testing (CVS, amniocentesis or NIPT). Most babies at increased risk of Down syndrome are completely normal. If your test gives an increased risk result, we will support and help you understand the next steps.