

## What happens to the sample?

The sample is taken to our laboratory and cells are grown or DNA is extracted to complete the test.

A rapid test is used to provide a quick answer (usually within two days) about Down syndrome and two other chromosomal abnormalities – this will identify about half of all chromosomal problems we can detect.

The longer test is more thorough as it can look at all 46 chromosomes in more detail. This may take 10-14 working days to be reported.

## Deciding to have an amnio or CVS

Whether to have an amniocentesis or CVS is a very personal decision. You may decide to have one of these tests because:

- The screening test you had for Down syndrome reported a high risk result and you want to know for sure whether your baby is affected.
- You don't want to rely on a screening test for Down syndrome (that may miss the occasional case) but would rather have a diagnostic test without any previous screening.
- You want to have information about additional chromosomal abnormalities that can be detected through CVS or amniocentesis but that are not traditionally screened for.
- You are known to have a risk of passing on a genetic condition (for example cystic fibrosis, thalassaemia or sickle cell anaemia) that can only be diagnosed by collecting a sample of DNA from the pregnancy.

The midwives and doctors that are looking after you in pregnancy will be happy to talk about the pros and cons of these tests and to help you make a decision. Your GP may be able to provide you with more information as well.

## Contacts

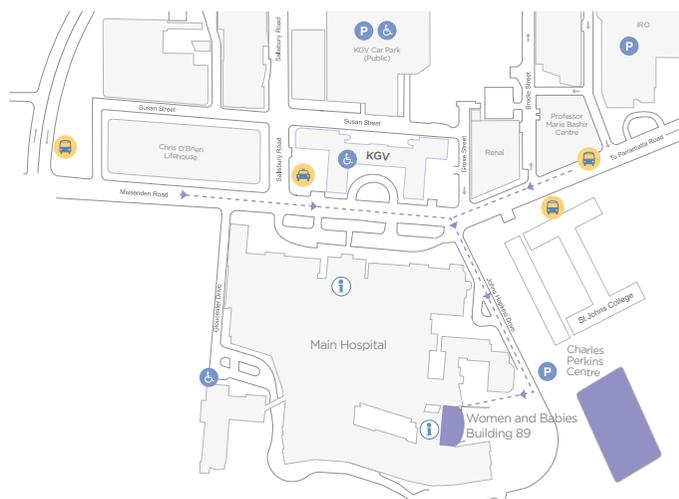
### Departments of Obstetric and Gynaecological Ultrasound and Fetal Medicine

RPA Women and Children's Health  
Royal Prince Alfred Hospital  
Level 5 Building 89  
Missenden Road Camperdown, NSW 2050  
Phone: (02) 9515 6042  
Email: SLHD-RPAFetalMedicineUltrasound@health.nsw.gov.au

### Charles Perkins Research Centre

John Hopkins Drive (off Missenden Road)  
The University of Sydney NSW 2006  
Phone: (02) 9515 6042

## 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



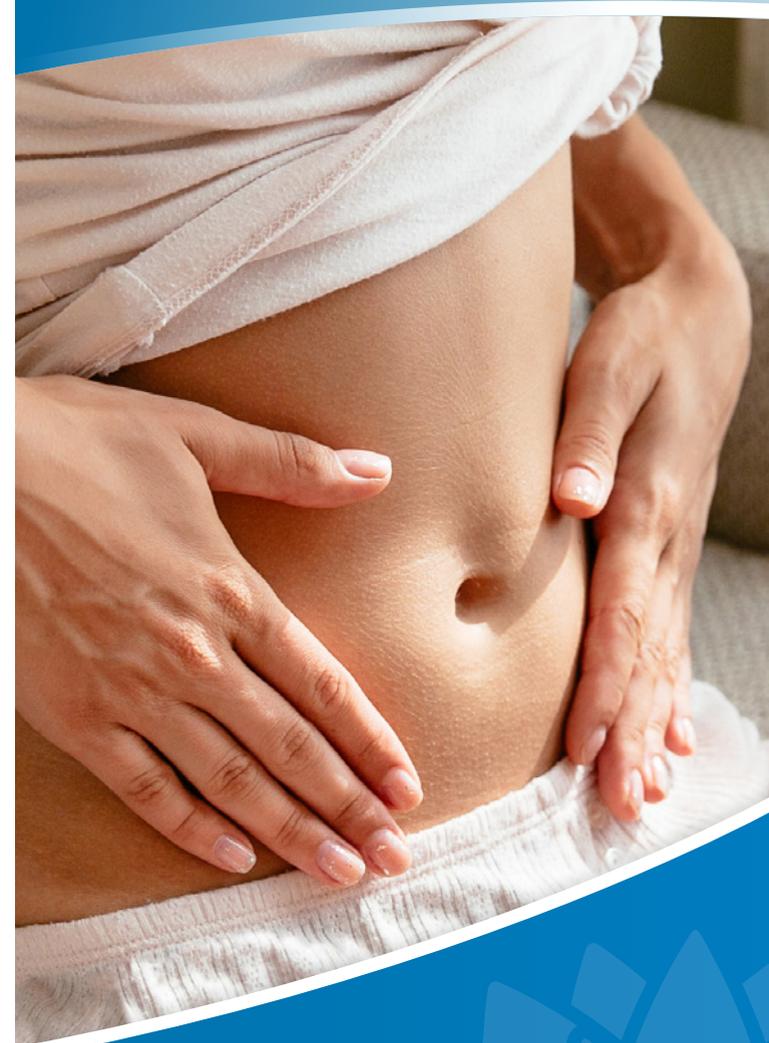
Plan your public transport travel. Timetable, fare, wheelchair access details contact the Transport Infoline 131500 [www.131500.com.au](http://www.131500.com.au)



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



A small proportion of women who have combined first trimester screening, NIPT or a scan at 18-20 weeks will be considered to be at high risk of having a baby with Down syndrome or another chromosomal abnormality. These women may be offered a CVS or amniocentesis test which can tell for sure whether a fetus is affected by one of these conditions.

## Down syndrome and other chromosome abnormalities

Our bodies are made up of cells. Each cell has a central library which contains instructions controlling how it works. The 'library' is called the nucleus and the instructions are written on 'books' called chromosomes. A single 'sentence' within a book may contain a specific instruction and is described as a gene.

A normal cell contains 46 chromosomes. Sometimes mistakes are made in copying this information between generations (i.e. from mother and father to child). A cell could for example have an extra chromosome (trisomy), or a small piece of a chromosome missing (a deletion).

Children with Down syndrome have an extra chromosome in all their cells. A normal cell has twenty three pairs of chromosomes. In Down syndrome, there is an extra chromosome 21 in each cell.

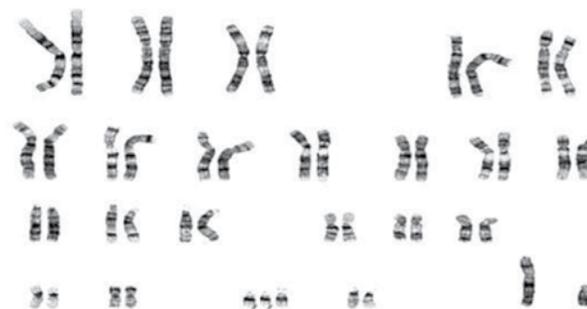


Figure: The chromosomes in a cell with an extra chromosome 21 (Down syndrome)

Down syndrome is the commonest chromosome abnormality we see. The additional genetic information leads to developmental changes that result in the characteristic appearance, physical and mental health problems that we see in children with this condition. Other chromosome additions or deletions also cause recognisable patterns of physical and mental abnormalities. Some are more severe than Down syndrome whilst others are less significant.

There are a number of screening programs designed to inform women about their risk of having a child with Down syndrome. Some of these programs provide risk information about other chromosomal abnormalities as well. These screening programs are described in more detail in our leaflets related to "combined first trimester screening" and "non-invasive prenatal testing (NIPT)".

If a mother is identified as being at high risk of having a baby with chromosomal problem, then she can choose to get additional diagnostic information by having an amniocentesis or CVS test.

## Amniocentesis or CVS?

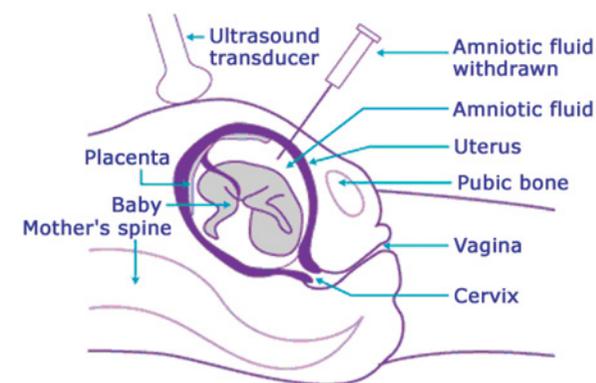


Figure: A schematic diagram showing how an amniotic fluid sample is obtained.

Amniocentesis and chorionic villus sampling (CVS) both involve taking a sample directly from the pregnancy. This involves introducing a needle into the uterus so that the sample can be collected.

Most of these tests are done by passing the needle through the mother's abdomen.

The amniotic fluid, which surrounds the baby, includes cells that have come off the baby's skin. These cells can be extracted from a fluid sample (20mls) and examined to identify how many chromosomes are present. The CVS test is similar - but in this case the cells are collected from the placental rather than the amniotic fluid/baby.

A CVS test is usually preferred at 11-14 weeks of pregnancy and a amniocentesis at >15 weeks of pregnancy - but there are some exceptions to this which your doctor would explain in more detail.

Both tests involve passing a needle through the abdomen into the uterus and there is a small risk of causing a miscarriage whilst taking the sample. This risk is approximately 0.2% (1 in 500 tests). It is actually a lower risk than the risk of spontaneous miscarriage at 12/16 weeks (1 in 50 and 1 in 100 pregnancies respectively).