

## Fees payable

Your doctor will provide you with details of the fee for chromosome testing. When the testing is complete you will be sent an invoice.

## Does Medicare cover the fee?

Your account will show a charge for chromosome analysis and should be paid in full. Medicare covers a portion of the fee for chromosome analysis and the Medicare Plus safety net 80% rebate out-of-pocket costs will assist even further, once the annual threshold has been reached.

## Where can I get more information?

You can either talk to the doctor who performed the aminocentesis/chorionic villus sampling, contact the laboratory by telephone or visit our website [www.sydneygenetics.com](http://www.sydneygenetics.com)

### This is what is involved in your test

1. Your tissue sample is identified and carefully labelled.
2. The specimen is prepared by isolating your baby's cells from the sample.
3. The cells are placed in nutrient culture medium, which will help them to grow.
4. The cell cultures are kept at 37 degrees (which is normal body temperature) and the cells must divide many times. This takes time and cannot be hurried.
5. After about a week or more in culture, the cells are ready to be examined. Their cell-division cycles are stopped so that the scientist can see the chromosomes. This is a special stage of the cell cycle called "metaphase". We spread the chromosomes onto a microscope slide.
6. The scientists use a special treatment on the chromosomes to give them a striped appearance. Each chromosome has a specially banded pattern, rather like a bar code. The scientists are skilled in recognizing these patterns.
7. At least 15 metaphase cells are examined by looking down the microscope.
8. Using a special computer we take a "photo" of the chromosomes, which can be rearranged to match each chromosome with its pair. The pairs of chromosomes are lined up, identified, placed in order and examined closely for any error. The final picture is called a karyotype.

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*The information in this brochure does not replace medical advice. Medical and scientific information provided in print and electronically by Sydney IVF might or might not be relevant to your own circumstances and should always be discussed with your own doctor before you act on it.*

MKTO0393 | August 2006

# Prenatal Chromosome Testing



SYDNEY GENETICS

Sydney Genetics' cytogenetics diagnostic laboratory in Australia offers timely, accurate chromosome testing.

## What are chromosomes?

The cells of your body contain DNA, which codes for all the parts of your body and its functions. The DNA is packaged into chromosomes. Most people have 46 chromosomes. Two chromosomes code for sex. Females usually have two X chromosomes and males usually have one X and one Y chromosome. There are 22 pairs of other chromosomes, which are numbered from 1 to 22. Chromosomes are not visible in cells all the time, but when a cell is dividing they can be seen briefly.

## What are chromosome abnormalities?

Chromosome errors can include having the wrong number of chromosomes or having chromosomes that are not completely intact. Having either a chromosome missing or an extra chromosome is called aneuploidy. This is the most common type of chromosome abnormality. You may be familiar with Down syndrome, which is caused by having an extra chromosome 21. Your doctor will have discussed with you your specific risk for a chromosome abnormality in your pregnancy.

## What is the effect of a chromosome abnormality?

If there is an extra chromosome the cells have extra DNA and if there is one missing there will be less. There can also be the wrong amount of DNA if the chromosomes are not completely intact. This is rather like having either too many or not enough ingredients for a recipe. Having such an error in the chromosome can result in abnormalities in your baby.

## How do scientists check the chromosomes?

The testing of chromosomes is one of the most labour intensive (and therefore one of the most expensive) pathology tests. Chromosome analysis is performed by highly trained scientists (called cytogeneticists) who are skilled in checking the chromosomes' fine detail. Standard prenatal chromosome testing is performed on samples of amniotic fluid or chorionic villus (placenta).

## When will I get my result?

As soon as your test is complete and has been checked we will contact you or your doctor to let you know the result. Sydney Genetics is the only cytogenetics laboratory in Australia that contacts patients directly. We think that this is an important service because it means that you will hear your very important result much faster. Most doctors are happy for us to phone you with the news. A few doctors prefer to give the results to their patients themselves. We try very hard to contact you as soon as possible. If you haven't heard from us and think your result might be ready, please do not hesitate to contact the laboratory.

## What if the test is abnormal?

Your doctor will contact you and discuss with you what the scientists have found. They will be able to explain to you what the abnormality means for your baby.

## What if my test takes less than 2 weeks?

Sydney Genetics' reporting times are among the fastest in Australia. We are always trying to improve our reporting times even further. Usually the results are available in 1 - 2 weeks. Please do not be concerned if our secretary contacts you before 2 weeks.

## What if my test takes longer than 2 weeks?

This happens very occasionally. The main reason is that the cells are taking a bit longer to grow in culture.

Just as we are all different in our height or weight, cells can also differ in how quickly they will grow in culture. Other times we do not get enough cells to look at initially to allow us to complete our analysis and we need to grow some more. This does not mean that the result is more likely to be abnormal.

## What if I am worried about how my test is going?

Most patients find it difficult waiting for the result of the chromosome test. We understand that you will be anxious about your result. We understand that it is even harder when your doctor has found a problem with your baby on ultrasound. Sydney Genetics is the only cytogenetics laboratory that welcomes you to make contact if you are concerned about the progress of your test. However, until we have grown your baby's cells, prepared the chromosome spreads and started to examine them through the microscope, we will not be able to tell you how long your test will take to complete.

## When will the result be ready?

It takes 1-2 weeks for the cells to grow and to examine them through the microscope and prepare the karyotype. For an extra charge, there are other rapid tests that check for the most common errors of chromosomes X, Y, 13, 18 and 21 in 1-2 days. Please see our "Fetal Aneuploidy Screening Test" brochure for more information.

## Is the result always correct?

In almost all cases the result is correct. However, for most scientific and medical tests there is a small risk of an abnormal result when there is not really an abnormality (false positive). There is also a small risk of getting a normal result when there is really an abnormality (false negative). From published studies and our own experience, we know that the chance of a false result is less than 1 in 4,000, but the risk is not zero.

## What if the test fails and there is no result?

It is extremely rare for a test to fail at Sydney Genetics. The chance of this happening in our laboratory is less than 1 in 1,000. We would usually be able to advise your doctor about two weeks after taking the sample if you need to have the test repeated.