

DNA testing

DNA is analysed using fluorescence PCR (polymerase chain reaction), which targets specific regions (markers) on the chromosomes of interest. Data from all the markers tested is then analysed for chromosome dosage by quantifying the peaks generated by the DNA analyser. Below are some examples of the output and what they tell us.



chromosome 21

This output shows two copies of chromosome 21.



chromosome 21

This output shows one single and one double copy of chromosome 21 - a total of three copies.

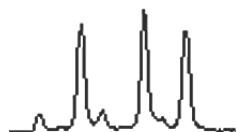
This fetus has trisomy 21 (Down syndrome).



chromosome 21

This output shows three distinct copies of chromosome 21.

This fetus also has Down syndrome.



chromosome 13

This output shows three copies of chromosome 13. This fetus has trisomy 13 (Patau syndrome).

Where can I get more information?

You may either talk to the doctor who performed the amniocentesis / chorionic villus sampling, contact the laboratory by telephone, or visit our website.

Fetal Aneuploidy Screening Test

by

FASTDNA™ or FASTPLUS™



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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.

MKT0060 | Version 5, October '07



SYDNEY GENETICS

FASTDNA™ or FASTPLUS™

Fetal aneuploidy screening test (FAST) by DNA analysis.

These are tests that can rapidly identify the most common chromosome abnormalities. Sydney Genetics was the first laboratory in NSW to offer these tests as routine clinical services.

Most people find it difficult waiting two weeks or more for the result of the chromosome test. We know that it is even harder when your doctor has found a problem with your baby on ultrasound. The tests allow us to find the majority of the more common chromosome abnormalities quickly - generally in one to two working days.

What is aneuploidy?

The cells of your body contain DNA, that codes for all the parts of your body and its functions. The DNA is packaged into chromosomes. Most people have 46 chromosomes. Aneuploidy is having either a chromosome missing or an extra chromosome. You may be familiar with Down syndrome, which is caused by having an extra chromosome 21.

What is the effect of aneuploidy?

An extra or missing chromosome can result in abnormalities in your baby. If your doctor has seen something unusual about your baby with ultrasound, or you have had a high risk result from the earlier pregnancy test for Down syndrome, there may be a greater risk that your baby could have an extra or missing chromosome. The chance of such an error increases with the mother's age.

How can scientists check for aneuploidy?

Chromosome analysis is performed by trained scientists (cytogeneticists) who are skilled in checking that the chromosome number is correct and that each chromosome is intact. This test involves growing the cells in culture because chromosomes can only be seen when cells are dividing. Because of the extra time needed for the cells to grow, a result is usually available in two weeks.

How are these tests done?

Your doctor will take a sample of amniotic fluid or chorionic villi and send it to our laboratory.

Option 1 - FASTDNA™ - DNA is removed from a number of cells. Using special markers for distinctive pieces of DNA, we can distinguish the different chromosomes X, Y, 13, 18 and 21. The fluorescent data generated by DNA is then analysed to determine the number of each chromosome present (see picture overleaf). Results are usually available in one to two working days.

Option 2 - FASTPLUS™ - In addition to aneuploidies, the FASTPLUS™ screen checks for the presence of Delta F508, the most common mutation that causes cystic fibrosis (CF). There are over 1,000 mutations that cause CF, however the Delta F508 mutation accounts for approximately 70% of the CF mutated genes in an Australian population. The Delta F508 mutation is less common in other ethnic groups.

What is cystic fibrosis?

Cystic fibrosis (CF) is an inherited disorder that principally affects the lungs and digestive system. Despite advances in treatment, CF remains incurable. Sufferers face a shortened life filled with physiotherapy and drugs.

One-in-25 Caucasians carry a CF mutation. When two carriers come together, their offspring have a 25% chance of having CF, and a 50% chance of being carriers.

If my result for CF is negative, does that mean my baby will not have CF?

The risk is never zero as not all mutations can be screened for. Approximately 10% of affected children do not carry the Delta F508 mutation.

If my result is not abnormal, does that mean my baby has normal chromosomes?

Not necessarily. About 90% of the chromosome errors seen by cytogeneticists involve extra copies or loss of chromosomes X, Y, 13, 18 or 21. If we do not find a problem with your FAST test for these chromosomes, it is unlikely (but not impossible) that your baby has a chromosome abnormality. The other chromosomes will be revealed, however, by the standard karyotype that always follows.

Is the result always correct?

In the majority of cases the results are 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 we know that the chance of a false result is less than 6 in 10,000.

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 tell you what the abnormality means for your baby.

Do I still need to have routine chromosome analysis as well as aneuploidy screening?

Yes. We must still check all of the other chromosomes and check that the chromosomes X, Y, 13, 18 and 21 look completely intact.

What if the test fails?

If we do not get a meaningful result from the FAST test, we will still get a result from the routine chromosome analysis. About 1 test in 2000 fails to give a result. There is no charge for the FAST test if it fails.

Does Medicare cover the fee?

There is no Medicare cover at the present time for aneuploidy or CF screening. However, Medicare covers a portion of the chromosome analysis. Please send us your credit card payment details on the payment form card found in the patient information pack. Alternatively we will send you an invoice upon receipt of your sample. Your account will show a charge for chromosome analysis including FAST screening 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.