

With either CVS or amniocentesis, cells are studied while they are dividing, at which time the chromosomes can be seen, the number of chromosomes in each cell can be counted, and the individual chromosome pairs can be examined. The number and appearance of an individual's chromosomes are described as a karyotype (see our brochure on **Prenatal Chromosome Testing**).

Collecting the sample by CVS or amniocentesis carries a very small risk of miscarriage caused by the sampling procedure (generally less than 1%) and any couple considering a fetal chromosome study should first be counselled by a geneticist, genetic counsellor or other suitably qualified person.

Does Medicare cover the fee?

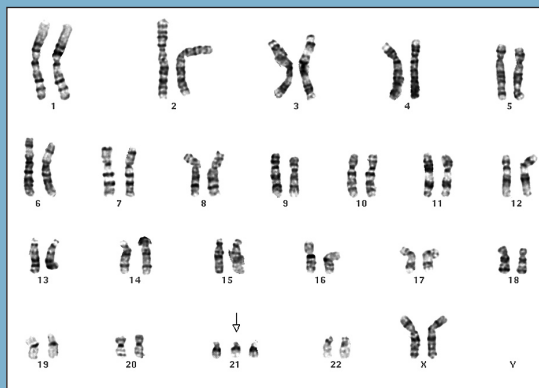
Your account will show a charge for biochemical screening and should be paid in full. Medicare covers a portion of the fee for biochemical screening and the Medicare Plus safety net 80% rebate out-of-pocket costs will assist even further, once the annual threshold has been reached.

For current information please ask your doctor, contact the laboratory or visit our website www.sydneygenetics.com.

Down syndrome screening

This screening test for Down syndrome has been developed to increase the proportion of Down syndrome cases that will be detected in the first trimester of pregnancy. The test involves combining the results of a blood test and a special ultrasound screening test.

The **screening** test is different from a **diagnostic** test for Down syndrome, which (with rare exceptions) would definitely show whether a fetus is affected or not. The screening test identifies babies at sufficiently high risk to move on to the diagnostic test, which is a fetal chromosome study (see brochure on **Prenatal Chromosome Testing**).



Trisomy Karyotype 21

A division of Sydney IVF

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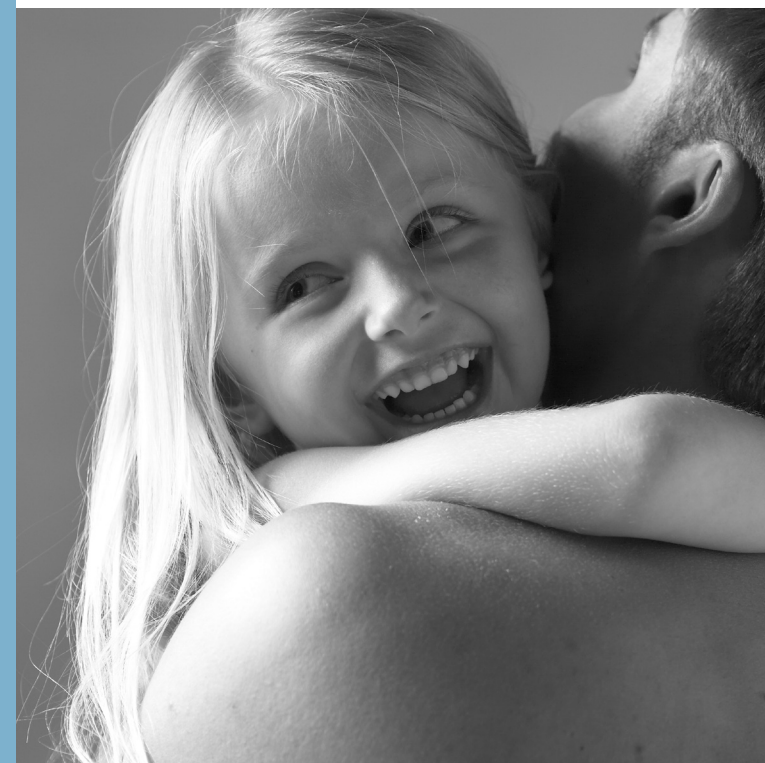
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The information in this brochure does not replace medical advice. Medical and scientific information provided in print and electronically by Sydney IVF might or might not be relevant to your own circumstances and should always be discussed with your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

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Early Pregnancy Test for Down Syndrome



SYDNEY GENETICS

DNA, genes, chromosomes and Down syndrome

The cells of your body contain DNA, that codes for all the parts of your body and its functions. The DNA is packaged into separate stick-like chromosomes. Most people have 46 chromosomes. Two chromosomes code for sex (the sex chromosomes). 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, called autosomes. Chromosomes are not visible in cells all the time, but when a cell is dividing they can be seen briefly, through a microscope. Each chromosome includes a very long strand of a molecule of DNA. The DNA is a genetic code or recipe that provides the cell with information on how to function and what to make. Small regions on each chromosome are the genes. Because the chromosomes mostly come in pairs, there will be two copies of most genes in every cell, one gene from each parent.

Down syndrome results from an individual having three copies of chromosome 21 instead of two. This is called trisomy 21. It nearly always results from either the egg or the sperm containing two copies of chromosome 21 instead of one. Individuals therefore have 47 chromosomes instead of the usual 46 chromosomes. Much less often, having three working copies of chromosome 21 results from an extra chromosome 21 being attached to another chromosome. This is called a translocation.

People with Down syndrome have unusual physical characteristics, including a facial appearance that is recognisable. Mental handicap is always present but it varies in severity. Affected people, to some degree, will benefit from intervention to help them to reach their potential.

In the absence of a widely accepted screening program, the frequency of Down syndrome will be about 1 in 660 births.

Are some couples at higher risk of having a baby with Down syndrome?

The frequency of Down syndrome increases with increasing age of the mother. It is a widely accepted policy that mothers aged 35 years or more be offered a fetal chromosome study. Couples who have had one baby with Down syndrome often choose to have the more accurate fetal chromosome study in subsequent pregnancies for greater diagnostic certainty.

How are the screening tests and diagnostic tests carried out?

Two tests are undertaken for Down syndrome screening. A blood test measures the level in the serum of two substances called PAPP-A and free beta-hCG. A special ultrasound examination measures the thickness of the soft tissue under the skin at the back of the baby's neck. On ultrasound, this tissue is more translucent than the adjacent tissues and the examination is called a test for nuchal translucency. The depth of nuchal translucency tends to be greater in babies with Down syndrome than in other babies. The results of the blood test and the nuchal translucency measurement are combined in a calculation to give a final risk-estimate of your baby having Down syndrome.

The blood test must be taken after 10 weeks and before 14 weeks of gestation (at the border of the first and second trimesters). If you have your blood taken at least 5 working days before your nuchal translucency test, your ultrasound specialist will have your blood result at the ultrasound rooms and will be able to give you your final risk rate at that appointment. Otherwise, you can have your blood taken on the day of your nuchal translucency measurement and Sydney Genetics will send your results to your doctor the following working day.

Where can I have blood taken?

Your blood can be collected by your doctor or any collection centre of Symbion or, by special arrangement, at other pathology centres.

What does it mean if I get a high-risk result?

A high risk result is not always a cause for alarm. The most likely outcome is that the baby is normal. The result of the screening test merely suggests that a diagnostic chromosome study would be a reasonable further precaution.

When fetal chromosome studies are recommended on the basis of the screening test, in about 20% of cases the fetus will be found to have Down syndrome or another chromosome error. A "high risk" result therefore does not mean that the fetus has Down syndrome: in about 80% of cases the chromosome study is normal.

Screening for Down syndrome is capable of detecting about 90% of fetuses with Down syndrome, leaving about 10% undetected. A low risk result does not mean that the fetus does not have Down syndrome.

The diagnostic test for Down syndrome is a fetal chromosome study. The study is done on either a sample of the placenta (afterbirth), which is called chorionic villus sampling (CVS), or on a sample of the amniotic fluid (amniocentesis). CVS is obtained ten to twelve weeks into the pregnancy, usually by a needle through the abdominal wall, or occasionally, by a soft catheter through the vagina.

Amniotic fluid is collected by amniocentesis, with a needle through the abdominal wall.