Both chorionic villus sampling and amniocentesis have risks for the developing baby. Chorionic villus sampling is performed in the 10th to 13th week of pregnancy and has a 1 in 100 risk of causing a miscarriage. Amniocentesis is performed between the 15th and 18th week of pregnancy and has a 1 in 200 risk of causing a miscarriage. Amniocentesis is routinely offered to mothers who are more than 35 years because the risk of Down Syndrome in the baby is greater than the risk of adverse effects from the procedure.

The sample taken from the developing baby is sent to the laboratory for a chromosome test. It is necessary to grow the cells before they can be karyotyped, i.e., analysed. This may take as long as two weeks. A preliminary test (called FISH) allows limited analysis of the baby's cells within 24-48 hours. It is important to understand that testing is not complete until the usual karyotyping has been carried out. Some abnormalities will only be found by the standard testing. Couples must also remember that a normal chromosome test does not guarantee a normal baby. Some genetic diseases such as cystic fibrosis cannot be detected by chromosomal analysis and specialised tests are required.

What if abnormalities are detected?

The approach taken depends on the type of abnormality found. Chromosome testing of the blood of the parents may be necessary. Once all information is available the doctor will discuss the implications of the abnormality. This will include the extent of physical malformation and/or intellectual impairment expected in the baby.

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Pregnancy Screening

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Prenatal diagnosis of chromosomal abnormalities

The expected outcome of pregnancy is the delivery of a normal healthy baby, and in most cases this will happen. However, there are some instances when birth abnormalities will occur due to an incorrect amount of genetic material carried by the baby's chromosomes. Chromosomal abnormalities can occur with babies of mothers of any age and in any pregnancy irrespective of previous normal babies.

About 1 in 5 pregnancies are chromosomally abnormal but the vast majority of these are miscarried spontaneously in the first weeks of the pregnancy. The liveborn incidence of chromosomal abnormality is approximately 1 in 200 births but not all these abnormalities lead to intellectual impairment or deformities. Chromosomal abnormalities can cause a variety of disorders with varying degrees of severity. Some abnormalities will cause severe intellectual impairment or physical deformity while others will have relatively mild effects.

What is a chromosome?

Chromosomes carry the hereditary factors or genes and are present in every cell of the body. A normal cell has 46 chromosomes in 23 matching pairs. One chromosome of each pair is provided by the mother (in the egg) and the other is provided by the father (in the sperm). The sex of the unborn child is determined by the sex chromosomes – the female has two X chromosomes and the male an X and a Y chromosome.

What is a chromosomal abnormality?

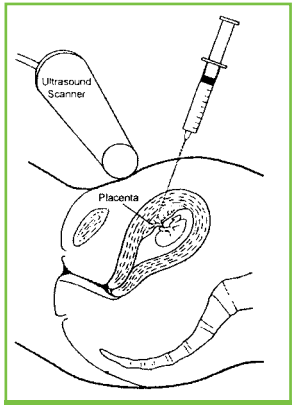
Normal growth and development of the baby requires the correct amount of genetic material. Occasionally, the egg or sperm has more or less genetic material than normal, resulting in a baby with abnormal chromosomes. Sometimes the egg and sperm are normal but an abnormality of cell division occurs just after fertilisation. The abnormality may involve the loss or gain of a whole chromosome or part of a chromosome. An incorrect amount of genetic material carried by the baby disturbs its normal growth pattern.

What is Down Syndrome?

The most common chromosome disorder is Down Syndrome. A Down Syndrome individual usually has an extra chromosome in every cell. This explains another name for the disorder – Trisomy 21 Syndrome. A small number of people have the extra chromosome 21 attached to one of the other chromosomes. They have three chromosome 21s but only 46 chromosomes in total. This is sometimes known as a Down Syndrome Translocation. An even smaller number of individuals with Down Syndrome do not have the extra chromosome in all cells - some cells are normal. This is called Mosaic Down Syndrome and is usually not as severe as the classic type.

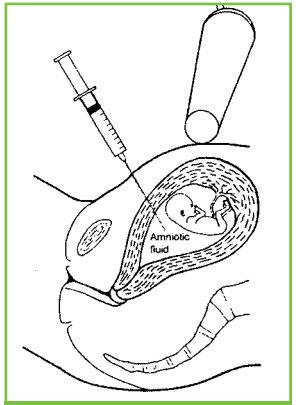
The overall incidence of the disorder is approximately 1 in 700 births but the incidence rises with the age of the mother. For mothers less than 25 years, the incidence is very low (less than 1 in 1000), but for mothers of 40 years the incidence is about 1 in 100. The age of the father is of no significance.

The risk of recurrence of Classic or Mosaic Down Syndrome in a subsequent pregnancy is slightly increased above the age related risk. There is a greater risk for Down Syndrome if the mother or father has a particular chromosomal abnormality known as a balanced translocation involving Chromosome 21.



Transabdominal Chorionic Villus Sampling

(Brock, D.J.H., Rodeck, C.H., & Ferguson-Smith, M. Prenatal Diagnosis and Screening, p.29)



Amniocentesis

(Brock, D.J.H., Rodeck, C.H., & Ferguson-Smith, M. Prenatal Diagnosis and Screening, p.29)

How can I check my pregnancy?

There are screening tests available. These tests cannot diagnose chromosomal abnormality in the developing baby. However, they can indicate the risk of Down Syndrome and whether further tests (chorionic villus sampling or amniocentesis) are worthwhile.

Maternal blood screening – can be performed for all pregnancies in the 8th to 18th week of pregnancy. Hormonal substances are measured in the blood of the mother and a risk for the developing baby having Down Syndrome is calculated.

Ultrasound – can be used as a screening test - some appearances raise the suspicion of Down Syndrome. Nuchal fold thickness (Nuchal Translucency) – the thickness of a particular layer at the back of the baby's neck - correlates with the risk of Down Syndrome. This screen is done between 10 and 13 weeks.

If these tests show that the developing baby is at increased risk of having Down Syndrome, then a chromosome test can be performed – prenatal diagnosis. A sample of cells is taken from the developing placenta (chorionic villus sampling) or from the fluid surrounding the baby (amniocentesis).