What is Prenatal Diagnosis?

by Joan Rye

During pregnancy, a woman receives antenatal screening, to assess the risk of her unborn foetus developing genetic abnormalities or conditions, such as Down syndrome, neural tube defects and chromosome abnormalities

All future parents hope for a healthy bouncing baby, but unfortunately, for some, there is more chance of having a child with serious mental or physical disabilities than there is for others. As congenital anomalies account for 20-25% of prenatal deaths, without the knowledge gained from prenatal diagnosis, there would be more chance of an unfavourable outcome for the foetus, the mother or both. Prenatal diagnosis is especially helpful for determining the outcome of the pregnancy, deciding whether to continue or terminate the pregnancy, making plans for possible complications during labour, managing the remaining weeks of the pregnancy, making plans for problems that may occur after the birth and discovering conditions that may affect future pregnancies.

If the results of antenatal screening show that a foetus has a greater than average risk of developing birth defects, the woman's doctor will offer her a definitive diagnostic test. Doctors may also offer these tests to women with a history of foetal abnormality, or an inherited condition in their family. Listed and briefly described below are some tests doctors may offer, including links if you wish to gain more in-depth information about the procedures used and the conditions they are used to detect.

Amniocentesis,
Chorionic villus sampling (CVS)
Fetoscopy
Foetal blood sampling
Foetal Radiology
Maternal blood tests
Ultrasound guided percutaneous skin and organ biopsy

Amniocentesis:

The test is usually carried out from the fourteenth week of pregnancy. A needle is inserted through the wall of the placenta into the amniotic cavity, in order to gain a sample of the amniotic fluid, to allow cultivation and assessment of foetal cells. Foetal cells in the amniotic fluid can then be analysed to defect specific conditions.

Chorionic villus sampling (CVS):

This test is usually carried out between the tenth and thirteenth week of pregnancy. Samples are taken of the developing placenta and foetal cells are analysed in the same was as in the amniocentesis procedure.

Fetoscopy:

Endoscopic techniques are used to allow visualisation and inspection of the foetus, and are usually carried out between the eighteenth and twentieth week of pregnancy.

Foetal Blood Sampling:

Foetal blood cells are collected from the umbilical cord by ultrasound guidance. The blood cells are then assessed for specific conditions. Intrauterine blood transfusions may also be performed using this procedure.

Foetal radiography:

X-rays are taken for investigation, around the same time as a routine ultrasound scan. If specific conditions are suspected, Prenatal MRI and foetal echocardiography are also used.

Maternal serum blood test:

Blood samples are taken to Measure the maternal serum alpha-fetoprotein levels. The test is usually performed between the fifteenth and twenty-second week of pregnancy to aid the diagnosis of specific conditions.

Prenatal ultrasound guided percutaneous biopsy:

This Procedure is carried out to allow foetal organs such as the skin, liver and muscle to be analysed for specific conditions.