Prenatal Screening Program

Useful information

The majority of pregnancies will lead to the delivery of a healthy newborn. However, a minority of approximately 3% will unknowingly deliver a child with some form of anatomical defect of disfigurement/dysmorphia/deformity. Prenatal screening programs have been developed to decrease this risk. Abnormalities present in the newborn are usually owed to some form of chromosomal or gene aberration in the fetus which may arise either spontaneously upon conception, during embryogenesis or via a hereditary mechanism.

Amongst such genetic aberrations are the syndromes which arise from Trisomy 21 (Down's), Trisomy 18 (Edward's) and Trisomy 13 (Patau's).

Chromosomal Abnormalities

The functions of the human body and its physical characteristics are all predetermined by the set of chromosomes found in the nucleus of all its cells whereby the transfer of all genetic information is performed. Each chromosome is made up of hundreds of genes of which each is responsible for the expression of a single protein which contributes to a very specific function within the body. The nucleus of each cell within our body contain 23 pairs of chromosomes (46 chromosomes) apart from sperm cells and ova which contain 23 chromosomes. During conception, the fusion of the sperm and ovum contribute all of their chromosomes to make the new genetic material of the fetus which will in turn develop to have 23 pairs of chromosomes in each of its cells.

If for any reason a fetus ends up with more or less than 46 chromosomes in each cell then functional and developmental abnormalities arise. The most commonly occurring chromosomal abnormality in the general population is Trisomy 21 (Down's syndrome). Its pathogenesis is owed to the presence of an extra chromosome 21 besides the pair which already exists in each cell, thus coining the term Trisomy 21.

All set of parents bear a risk of having a child with chromosomal abnormality. This risk increases as the age of the mother increases.

Modern diagnostic methods

Chromosomal abnormalities in a fetus are diagnosed with cytogenetic techniques which can be applied after the 10^{th} week of gestation. However, these invasive techniques carry up to a 1% risk of miscarriage. For this reason, such techniques are reserved for pregnancies which carry an increased risk following prenatal screening of bearing a fetus with a chromosomal abnormality as described below:

Screening for chromosomal abnormalities is achieved by combining the measurements of two types of markers: maternal biochemical markers and fetal ultrasound measurements. The biochemical markers are two hormones in the pregnant woman's blood.

The risk of chromosomal abnormalities in a pregnancy is determined through the combination of the biochemical blood test results, the ultra sonographic fetal measurements and the age of the mother. The first trimester fetal ultrasound provides a thorough examination of the fetus together with the measurement of the fluid collection seen behind the fetus's neck which is directly linked with the presence of chromosomal and/or anatomical defects.

In the case of Trisomy 21, a risk greater than 1 in 300 (e.g. 1 in 250) is considered as high risk for the fetus as being affected with this syndrome. In such cases genetic counselling is offered to the parents before they take any further action. A risk less than 1 in 300 (e.g.1 in 600) is considered as being low risk and no further action is required. Nevertheless, any risk, however small still leaves a negligible chance of the fetus being affected with a chromosomal abnormality.

By following the above described procedure, the pregnant population is actually grouped into a low risk category or a high risk category, whereby it is estimated that approximately 95% will fall into the former group and 5% into the latter. Those falling into the high risk group are subsequently offered a diagnostic cytogenetic examination. In this way, 9 out of 10 Trisomy 21 cases will be diagnosed early on in pregnancy, i.e. prenatally.