

Prenatal diagnosis for abnormalities detection



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PRENATAL DIAGNOSIS

The incidence of major abnormalities apparent at birth is 2 to 3 percent.

These anomalies cause a significant portion of neonatal deaths, more than a fourth of all pediatric hospital administration results from genetic disorders.

Prenatal diagnosis is the science of identifying structural or functional abnormalities, birth defects in the fetus. With this information clinicians can hope to provide appropriate counseling and optimize outcome. Birth defects can arise in at least three ways. The malformation i. e structural fetal abnormality, then the deformation, then the third type is disruption.

Sometimes multiple structural or developmental abnormalities occur together in one individual . A cluster of several abnormalities can be a syndrome. Prenatal diagnosis helps to detect these abnormalities.

Thus prenatal diagnosis basically comprises of different techniques and methods used to determine any diseases or health condition of the unborn fetus or embryo.

SOME PROCEDURES FOR EARLY DETECTION OF FETAL 1)GENETIC 2) CHROMOSOMAL 3)STRUCTURAL ABNORMALITIES

Amniocentesis

Triple test

Chorion villus sampling

Cordoncentesis

Ultrasonography

Fetoscopy

Maternal serum alpha fetoprotein

Peri-implantation genetic diagnosis

Fetal cell isolation from maternal blood

3-D or 4-D ultrasound with increased resolution

a) Amniocentesis: This test is developed by Richard Dedrick. Examination of a sample of amniotic fluid makes possible the prenatal diagnosis of chromosomal abnormalities and certain metabolic defects. The procedure can be used as early as 14th week of pregnancy when abortion of the fetus is still feasible. The diagnosis of chromosomal abnormalities is made by culture and karyotyping of fetal cells from the amniotic fluid, and of metabolic defects by biochemical analysis of the fluid. Karyotyping is a test used to detect genetic problems. Before the procedure begins a local anesthetic is given to the mother to get relief from the pain, a needle is inserted into the abdominal wall and then the amniotic fluid is withdrawn. The fetal cells are distinguished from the extract and the cells are cultured in medium, further stained and examined under microscope for abnormalities. Amniocentesis is very accurate in detecting the abnormalities in fetus as well as to find the gender of the fetus, hence is banned in many countries. Amniocentesis is called for in the following circumstances if the parents are prepared to consider abortion. A mother aged 35 years or more (because of high risk of down's syndrome with advanced maternal age). Patients who have had a child with Down's syndrome or other chromosomal abnormalities. Parents who are known to have chromosomal translocation. Parents who have had a child with metabolic defect-detectable by amniocentesis. The

most common are defects of the neural tube, anencephaly and spina bifida which can be detected by an elevation of alpha fetoprotein in amniotic fluid

b) Chorionic villus sampling(CVS): This is another prenatal diagnosis used to find chromosomal or genetic disorders in the fetus. CVS was first described in China in the mid-1970s. This technique is also called as chorionic sampling. This is usually performed at 10 to 13 weeks of pregnancy. This new technique allows prenatal diagnosis at 9 to 11 weeks of pregnancy. By this test the chromosome status can be easily determined. Prenatal diagnosis of congenital abnormalities offers the parents the option of therapeutic abortion. Samples may be obtained transcervically or transabdominally, depending on which route allows easiest access to the placenta. Relative contraindications include vaginal bleeding or spotting, active genital tract infection, extreme uterine ante or retroflexion, or body habitus precluding easy uterine access or clear sonographic visualization of its contents. The indications for CVS are essentially the same as for amniocentesis, except for a few analysis that specifically require either amniotic fluid or placental tissue. The primary advantage of villous biopsy is that results are available earlier in pregnancy, which lessens parental anxiety when results are normal. It also allows earlier and safer methods of pregnancy termination when results are abnormal. Complications of CVS are similar to those of amniocentesis. There is an understandable desire to perform CVS as early as possible. Technically, this can be done successfully as early as six weeks' gestation. However, a few clusters of limb reduction defects have been reported following CVS, with a trend toward an increased incidence of these defects when CVS was done before nine weeks gestation.

Subsequent, large epidemiological follow-up studies failed to confirm this association, but most clinicians delay this procedure until after 10 weeks gestation. The incidence of amniotic leakage or infection is less than 0.5 percent.

c) Alpha fetoprotein: Neural tube defects can be detected by measurement of a specific protein of foetal origin called alpha fetoprotein in maternal blood and amniotic fluid during pregnancy. A neural tube defect is termed as an opening in the brain or spinal cord that occurs very early in the developmental stage of human. Neural tube defects include spina bifida.

d) Ultrasound: This can be used to visualize the foetus and detect many abnormalities of the foetus. Ultrasound is the method of choice for detection of anatomical problems (e. g. absent kidneys, spina bifida), but provides no information on the genetic constitution of a fetus. Maternal serum screening, alone or in combination with ultrasound, is often used to identify fetuses at risk of Down's syndrome, but the definitive chromosomal diagnosis can only be made from fetal cells.

e) Fetal cells from maternal blood can be isolated for prenatal diagnosis during pregnancy. Fetal trophoblast, lymphocytes, granulocytes, and nucleated red blood cells are studied. Generally, 1ml of maternal blood contains one fetal cell.

f) Peri-implantation genetic diagnosis(PGD): This is done by polar body biopsy, blastomere biopsy, trophectoderm biopsy. Polar body biopsy is done by removing first or second polar body in the preconceptional phase.

Paternal genotype is not assessed here. Blastomere biopsy -one or two cells

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are aspirated through a hole made in zona pellucida by mechanical, laser or chemical means. This does not effect the normal embryonic development.

g) Triple test: This is basically a screening test. It mainly detects the presence of three substances in the maternal blood, i. e of alpha fetoprotein, human chorionic gonadotropin(hcg)which is basically a hormone in placenta, and estriol. The triple test detects the presence of high level or low level of these substances. Both high and low level can creat abnormalities.

h) Cordocentesis - It is also called as Percutaneous Umbilical Cord Blood Sampling (PUBS), this is a test that mainly examines the blood from the fetus to detect fetal abnormalities. The procedure carried out is quite similar to amniocentesis. This test helps in finding any malfunction and abnormalities of the fetus.

i) Fetoscopy -This procedure provides a direct visualisation to the fetus, amniotic cavity, umbilical cord, and fetal side of placenta. It does this by ultrasound scanning. Here an endoscope is inserted into the abdomen of the mother which acts as an analyzer.

Thus many prenatal diagnosis are available nowadays which allows to detect any kind of abnormalities in the fetus. Once diagnosed, some genetic abnormalities can be treated with partial or complete success by medical and surgical measures. Genetic counseling can also have impact when individuals or couples at risk are identified.