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Perspective

A Short Note on Prenatal Diagnosis

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DESCRIPTION

Prenatal diagnostic refers to a diagnosis done before the baby is born. It allows the doctor to determine whether the growing baby has a problem. Amniocentesis and chorionic villus sampling are the two basic procedures. These tests aid in the detection of genetic abnormalities before to birth. Some people are at a higher risk of having a child with a genetic illness or other issue. Knowing about potential issues before the baby is born may be beneficial. They may be able to make better judgments concerning their infant's health care. Certain issues can be addressed before to the baby's birth. Other issues may necessitate immediate medical attention.

DIAGNOSIS

For prenatal diagnostics, a number of non-invasive and invasive procedures are used. Prenatal diagnostic procedures include.

- Ultrasonography
- Amniocentesis
- Chorionic villus sampling
- Fetal blood cells in maternal blood
- Maternal serum alpha-fetoprotein
- Maternal serum beta-HCG
- Maternal serum unconjugated estriol
- Pregnancy-associated plasma protein A

Ultrasonography

This is a non-invasive treatment that is safe for both the mother and the foetus. High frequency sound waves are used to generate visual pictures from the pattern of echos produced by various tissues and organs, including the baby in the amniotic cavity. At around 6 week's gestation, the growing embryo may be seen for the first time. Between 16 and 20 weeks of gestation, the major internal organs and extremities can be identified to detect if any are defective.

Although an ultrasound examination may be very helpful in determining the size, position of the foetus, and the location of the placenta, the amount of amniotic fluid, and the appearance of embryonic anatomy, it has several limitations. Subtle abnormalities may not be noticed until later in the pregnancy. Down syndrome (trisomy 21) is an excellent illustration of this, as the morphologic defects are typically mild, such as increased nuchal translucency (the subcutaneous space between skin surface and underlying cervical spine).

Amniocentesis

This is an invasive technique in which a needle is inserted into the amniotic cavity within the uterus through the mother's lower abdomen. Enough amniotic fluid is present for this to occur at 14 weeks gestation. The majority of amniocenteses for prenatal diagnosis are conducted between 14 and 20 weeks gestation. However, an ultrasound examination is usually performed before to amniocentesis to assess gestational age, the location of the embryo and placenta, and the presence of adequate amniotic fluid. Fetal cells (mainly generated from foetal skin) are found in amniotic fluid and may be cultured in culture for chromosomal analysis, biochemical analysis, and molecular biologic study.

The amniotic fluid can be examined in the third trimester of pregnancy to determine foetal lung maturity. When the foetus is less than 35 to 36 weeks gestation, the lungs may not be developed enough to maintain life after birth. This is due to a lack of surfactant production by the lungs. The newborn may suffer respiratory distress syndrome after birth as a result of hyaline membrane illness. The amniotic fluid may be studied by counting the number of lamellar bodies. Fluorescence polarisation (fpol), Lecithin Sphingomyelin (LS) ratio and Phosphatidyl Glycerol (PG) are other assays for foetal lung maturity. Because these tests have a low positive predictive value for respiratory distress, the decision to do amniocentesis might be decided by taking into account concerns related to gestational age and urgency of delivery. It is quite effective in detecting genetic problems.

Amniocentesis risks are infrequent, although can include foetal loss and maternal Rh sensitization. The risk of foetal death after amniocentesis is roughly 0.5% higher than would be predicted. Rho Gam can be used to treat Rh negative mothers. It is exceedingly improbable that amniocentesis fluid will be contaminated by maternal cells. If oligohydramnios is present,

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obtaining amniotic fluid is difficult. Instilling saline into the amniotic cavity and then removing fluid for examination is occasionally achievable. Amniocentesis or CVS is performed when there is a high possibility that the baby may have genetic diseases or birth defects. There are a few minor problems associated with amniocentesis and CVS.

- Miscarriage
- A baby's infection or damage.
- Amniotic fluid leakage
- Abnormal vaginal bleeding