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GUEST EDITORIAL

Prenatal screening and medical termination of pregnancy

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The Medical Termination of Pregnancy Act of 1971 deals with legal and safe abortion services based on eugenic, therapeutic, social and humanitarian grounds and ensures that trained healthcare professionals supervise the procedure.[1]

The MTP (Amendment) Act of 2021 permits both married and unmarried women to undergo termination of pregnancy up to the gestational period of 20 weeks in case of failure of contraceptive methods, and the opinion of a single Registered Medical Practitioner (RMP) is necessary for the same. For pregnancies between 20 to 24 weeks of gestation, an opinion of two RMPs is required. For MTP indicated for fetal anomalies, the opinion of State level Medical Board is mandatory. The board should consist of a gynaecologist, a radiologist, a paediatrician and any other member as notified by the State Government. The Act permits MTP beyond 24 weeks of gestation on the grounds of substantial fetal anomalies as determined by the Medical Board.[2] The MTP (Amendment) Act of 2022 permits abortion up to the gestational age of 24 weeks, irrespective of the woman's marital status.[1]

A situation may arise when severe fetal anomalies are diagnosed after 24 weeks of gestation, and the woman does not want to continue the pregnancy. In such situations, the clinician must approach the medical board constituted for this purpose. The board will have to refer to the list of serious fetal anomalies published by the government for this purpose and, after thorough discussion, may allow termination of pregnancy.[3]

It is, therefore, imperative that a systematic prenatal screening is essential for the detection of fetal anomalies as early as possible so that the obstetrician can do proper counselling with the expectant parents. Including a pediatric surgeon and a clinical geneticist in the counselling team is also necessary.

Prenatal screening for fetal anomalies is initiated during the first trimester for the detection of Neural tube defects, chromosomal anomalies like trisomy 21 (Down), 18 (Edwards), 13 (Patau), etc., structural anomalies like congenital heart disease, congenital diaphragmatic hernia, renal anomalies like agenesis and cystic disease, lung anomalies, limb anomalies, body wall defects like exomphalos and gastroschisis, single gene disorders like cystic fibrosis, fragile X syndrome, etc. Some anomalies are late fetal events not detected during early fetal screening. Such conditions include intestinal atresia, malrotation defect, CTEV, etc.

Prenatal diagnostic methods are of two types: Invasive and non-invasive. The invasive investigations are chorionic villus sampling and amniocentesis. Because of the chances of complications like bleeding and sepsis, invasive investigations are done in special situations with proper aseptic and antiseptic precautions. Moreover, they need professional expertise and technical details.[4]

The non-invasive methods are more extensively used for early detection of fetal anomalies. They are of three types:

1. Maternal-fetal markers
2. Ultrasound anomaly scan
3. Fetal echocardiography.

Markers are detected in the maternal serum. Commonly screened markers are:

(i) α-fetoprotein (AFP)

(ii) Maternal serum human Chorionic Gonadotrophin (hCG)

(iii) Free-β subunit of hCG (free β-hCG)

(iv) Pregnancy-associated plasma protein (PAPP)- A

(v) Unconjugated Estriol (uE3)

(vi) Inhibin A

(vii) Placental Growth Factor (PlGF)

The combination of free β-hCG, AFP, uE3 and Inhibin A is referred to as the Quad test and is regarded as reliable. A quad test and a first-trimester ultrasound scan can accurately identify the anomalies.

Ultrasound anomaly scan is generally performed at around 18 to 21 weeks of gestation to get maximum detection of fetal anomalies. However, earlier scans towards the end of the first trimester at expert hands may have an early diagnosis of many anomalies.[5]

The common Ultrasound markers are:-

(i) Nuchal Translucency (NT)

(ii) Absence of Fetal Nasal Bone (NB)

(iii) Tricuspid Regurgitation (TR)

(iv) Absent or reversed ductus venous blood flow (DV)

(v) Frontal Maxillary Facial angle (FMF)

(vi) Biparietal Diameter of the skull (BPD)

Second-trimester maternal serum AFP is the earliest example of screening for detecting Anencephaly and Spina bifida, which generally includes Meningomyeloceles and Encephaloceles.

Maternal serum AFP level increases throughout the second trimester. To allow for this increase, screening programs express results as multiples of gestation-specific median values (MoMs) in unaffected pregnancies. In a large multi-centric collaborative study in the UK, the average AFP level was 6.4 MoMs in Anencephaly and 3.8 MoMs in open Spina bifida. In contrast, levels in closed spina bifida were found to be near normal. Detection rates were 86% for Anencephaly and 76% for open Spina bifida.[6]

Detection rates have since increased with the addition of an "Anomaly scan" to measure the fetal skull's biparietal diameter (BPD) in 18 to 20 weeks. It has increased the detection rates for open Spina bifida to 85% with a 1.4% false positive rate. Several first-trimester ultrasound markers, such as Intracranial Translucency, BPD, scalloping of frontal bones and cerebellar anomalies, are reported. Since the fetal liver produces AFP, AFP screening also detects the presence of abdominal wall defects like Gastroschisis and open Omphalocele. AFP false positivity is more in the case of twins.

cfDNA test: Cell-free fetal DNA represents extra-cellular DNA originating from trophoblastic cells. cfDNA of fetal origin represents only 3% of the total cfDNA circulating in maternal blood in early pregnancy. cfDNA test is helpful for the detection of genetic sequences not present in the mother. Detection of mRNA from the PLAC4 gene, which is only expressed in the placenta, can also detect chromosomal anomalies in the fetus. Presently, the test is too expensive to be prescribed for routine screening.

It is helpful for the detection of chromosomal aneuploidy (Trisomy, monosomy). For Down syndrome, the detection rate may be as high as 99%. Other areas of promise are the detection of Rh status in D-negative mothers, Fetal sex determination in fetuses with genital ambiguity on ultrasound scan, Congenital adrenal

hyperplasia, Single gene disorders like Achondroplasia, Huntington's disease, Turner's syndrome and many more conditions.[7,8]

Congenital Heart Diseases

CHD accounts for nearly 25% of all fetal anomalies. Sonographic markers like increased Neuchal Translucency, abnormal ductus venous blood flow, Tricuspid regurgitation and abnormal cardiac axis increase the risk of CHD in the fetus. Early fetal echocardiography helps detect Tricuspid atresia, pulmonary atresia, Tetralogy of Fallot, Transposition of great vessels, hypoplastic left heart syndrome and several other cardiac anomalies.

Detection of a serious fetal anomaly causes a great psychological impact on the mother. Very careful counselling is necessary. At times, it is a difficult task for the treating obstetrician to decide whether to go for medical termination of pregnancy. With the advancement of Pediatric Surgery and Fetal Surgery, it is now possible to save many babies with serious congenital malformations. Hence, a careful assessment must be done before offering the final opinion to the expectant mother.

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