Prenatal Diagnosis (PND)

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Prenatal diagnosis or screening is testing for diseases or conditions in a fetus or embryo before it is born. By prenatal diagnosis (PND) many diseases or conditions can be diagnosed before birth of the child, such as - Thalassaemia, Sickle cell anemia, cystic fibrosis, hemophilia, fragile X syndrome, Down's syndrome, chromosomal abnormalities, genetic diseases, neural tube defect, Spina bifida, birth defects etc.

There are mainly three purposes of prenatal diagnosis a) To enable timely medical or surgical treatment of a condition before or after birth. b) To give the parents the chance to abort a fetus with the diagnosed condition. c) To give parents the chance to prepare themselves psychologically, socially, financially and medically for a baby with a health problem or disability or for the likelihood of a stillbirth.

Prenatal diagnostic tests are of two types a) Non invasive b) Invasive techniques employed for prenatal diagnosis include :

1. Ultrasonography
2. Amniocentesis.
3. Chorionic villus sampling.
4. Fetal blood cell in maternal blood.
5. Maternal serum alpha feto protein.
6. Maternal serum beta HCG.
7. Maternal serum Estriol.

Ultrasonography is a very good informative non invasive procedure for PND. It can be done at any stage of pregnancy and may be repeated as and when necessary without any hazards. Fetal well being and anatomical defects may be visualized correctly with this modality.

Chorionic villus sampling (CVS) under ultrasonographic guidance through trans-vaginal or trans-abdominal route, usually done in early part of first trimester of pregnancy. Mostly is done at 8-12 weeks of gestation[1].

Amniocentesis is usually done in early part of second trimester. Risks of these procedures are around 1%1.

In prenatal diagnosis often we use the term triple test, which means test for maternal serum alpha feto protein, Beta HCG, Estriol[2].

Increased level of alpha fetoprotein indicates neural tube defect (Anencephaly, spinabifida). Very high levels of HCG suggest trophoblastic disease (Molar pregnancy).

The absence of a fetus in ultrasonography along with an elevated HCG suggest hydatiform mole.

The amount of maternal serum Estriol depends upon a viable fetus, a properly functioning placenta and maternal well being. The measurement of serum Estriol level in the third trimester gives an indication of general well being of the fetus. If the estriol level drops then the fetus is threatened and delivery may be necessary urgently. Estriol tends to be lower in Down's syndrome, adrenal hypoplasia with anenecephaly[3].

In Multan, Pakistan though free prenatal diagnostic services are introduced but the response is not good[4].

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In India PND is done in many government and private sectors. Most of the care seekers comes for detection of Thalassaemia[1]. For Thalassaemia premarital screening is better and wiser than PND. Emphasis should be given in premarital screening in developing countries, as it is easily available, easier to do and cheaper. Detection can be done by Hemoglobin electrophoresis[5].

Advanced molecular, cytogenetic and biochemical techniques are useful addition for genetic counseling and prenatal diagnosis[6].

In Bangladesh we are still lagging behind in prenatal diagnosis. Neither in Government nor in private sector well organized, well equipped, sophisticated, true tertiary level lab facilities for prenatal diagnosis not yet developed. There is a wide scope for developing this sector in Bangladesh.

References

1. *Colah RB, Gorakshakar AC, Nadkarni AH; Invasive & non invasive approaches for prenatal diagnosis of hemoglobinopathies: experiences from India; Indian J Med Res; 2011 oct; 134:552-60.*

2. *Imudia AN, Kumar S, Diamond MP, Decherny AH, Armant DR; Transcervical retrieval of fetal cells in the practice of modern medicine: a review of the current literature & future direction; J Fertil steril; 2010;93(6):1725-30.*

3. *Ball RH, Caughey AB,Malone FD et al; First & second trimester evaluation of risk of Down's syndrome; J Obstet Gynecol; July 2007;110(1):10-7.*

4. *Mahmud Baig S, Sabih D, Rahim MK et al; Beta Thalassaemia in Pakistan : a pilot program in prenatal diagnosis in Multan; J pediatr Hematol Oncol, 2012 Mar; 34(2):90-2*

5. *Verma IC, Saxena R, Kohli S; Past present & future scenario of thalassaemic care & control in India, Indian J Med Res, 2011 Oct; 134:507-21.*

6. *Verma IC, Saxena R, Lall M, Sharma R, Genetic counseling & prenatal diagnosis in India: experience at Sir Ganga Ram Hospital. Indian J pediatr, 2003 Apr; 70(4):293-7*