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Epidemiology of NTD and Down's Syndrome

Neural Tube Defects (NTD) are one of the most common birth defects affecting the human fetus with significant morbidity and mortality. An important distinction from the point of view of prenatal diagnosis is that between 'open' and 'closed' spina bifida. Open in this context means that there is some exposure of neural tissue or the lesion is completely covered by a thin transparent membrane, and closed means covered by skin or a thick opaque membrane. Open spina bifida can be more readily diagnosed by biochemical tests and has worse prognosis. About one-third of infants with open lesions survive to 5 years and most survivors have severe handicap due to hydrocephalus, urinary and anal incontinence and paralysis of the lower limbs. For closed lesions, about two-thirds survive to 5 years and one-third of survivors have severe handicap. About one in six spina bifida lesions are open. The incidence of NTD in India varies from 1.7 to 12.2/1000 live births. *BDR News*.[1] These include anencephaly, open and closed neural tube defects.

Down's syndrome (trisomy 21): Trisomy 21 is the most common autosomal aneuploidy with significant impact on the fetal and neonatal well-being, burden on the families and society in form of moderate to severe mental retardation and variable expression of several malformations such as congenital heart disease, hypothyroidism as well as predisposition to certain cancers like leukemia. Morbidity

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Markers used are pregnancy associated plasma protein A (PAPP-A), free β-human chorionic gonadotropic (fβ-hCG), alpha fetoprotein (AFP), unconjugated estriol (uE3), inhibin (Inh-A) which are combined into dual (PAPP-A, fβ-hCG), triple (AFP, fβ-hCG, uE3), and Quadruple (AFP, fβ-hCG, uE3, Inh-A) screen as their efficacy lies in combined results. The mathematical calculation involves the levels of these substances after considering other factors namely maternal age, date of birth, weight, diabetes status, previous pregnancies with trisomy 21, number of fetuses, smoking status, ethnic origin, type of pregnancy (natural/induced/IVF). Inclusion of nuchal translucency (NT) finding in double marker test, increases the accuracy of the test (Table 8.1). The main objective of the tests is to offer pre-test genetic counseling; for effective diagnostic and therapeutic options, having a child who would have special needs and explain the fundamental difference between the screening and the diagnostic tests.

Review of current scenario from few countries and our experience regarding the uptake of its diagnostic ability and impact on Down syndrome outcome are discussed below. Overall increase in women of elderly age group is increased due to more couples starting or spacing birth of a child and possibility of parenthood by way of reproductive technology for infertile couples even at a later age.

Introduction of MSS and its impact on the birth prevalence of Down syndrome, follow-up of invasive procedures like CVS and amniocentesis is widely studied. A population based study in Australia has shown the uptake of prenatal testing went up from 7% (which was mainly for elderly woman undergoing CVS/amniocentesis) to 84%. Maternal serum testing followed by invasive procedures has shown that about half the terminations were in younger women.[3] Similar studies on 26,488 antenatal and postnatal diagnoses of Down syndrome made in cytogenetic laboratories in England and Wales in the year 2001, the UK national committee advised that all pregnant mothers should be

Annexures

- Information booklet for public
- Handbook for screening program staff
- Consent form
- Referral proforma
- Result proforma and clinical explanatory document
- Data compilation proforma

ANNEXURE 1: First Trimester Combined Screening Sample Referral and Patient Consent for First Trimester—Form*

Date:

Note: *Mandatorily to be filled

*Name: * DOB: Date:
*Address:
Religion: Caste: *Age: * Weight:
*Clinician in charge:
E-mail ID:
Hospital address:
*Contact numbers:

Confounding factors:
*Pregestational diabetes: Yes/No Smoking: Yes/No
Periconceptional folic acid: Yes/No
Previous chromosome abnormaly/NTD: Yes/No
If yes, specify:

Brief obstetric history:
* Gestational age by LMP: * LMP
* Sonography findings: * Scan date