PRENATAL DIAGNOSIS OF NEURAL TUBE DEFECTS

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ABSTRACT

14 cases of neural tube defect (NTD) were detected on screening of 1950 pregnant women over a period of 30 months by antenatal ultrasonography (USG) examination, suggesting an incidence of 7.1 per thousand conceptions. The distribution of types of NTD were occult dysraphism 14%, meningocele 14%, myelocele 7%, myelomeningocele 14%, encephalocele 14% and anencephaly 37%. The study established high accuracy of USG screening for antenatal diagnosis of NTD with sensitivity of 85.71% and specificity of 100%.

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KEY WORDS :Neural tube defect.

Introduction

The NTDs results from failure of neural tube to close in early embryogenesis. It is a syndrome of malformation of CNS that includes spina bifida, encephalocele and anencephaly. The spina bifida, also called as spinal dysraphism, is a midline defect of fusion of vertebra resulting in exposure of contents of neural canal commonly at dorsal aspect. It includes meningocele, myelomeningocele, myelocele, lipomeningomyelocele and rachischisis. Encephalocele also called as cranium bifidum or cephalocele is a condition of defective fusion of cranium resulting in protrusion of intracranial contents through the defect. Anencephaly is an anomaly, characterized by absence of cranial vault and cerebral hemispheres. The exact aetiology of NTDs is still unknown, however it could occur as a part of chromosomal anomalies or due to exposure to teratogens. The reported incidence of NTD is 0.4 in USA [1], 8.7 in Chandigarh and 4.0 in Delhi per thousand live births [2].

The conservative therapy of 1950s resulted in mortality of 90% by 10 years of age and of the survivors, only 70% remained ambulatory and had a mean IQ of 89. Even at present, intensive therapy still results in mortality of 14% by 3-7 years of age and of the survivors, 80% are ambulatory and only 73% have mean IQ of > 80 [3]. Hence the occurrence of NTD is not only disastrous emotionally and financially to the family and society but is also an unrewarding challenge to the medical profession. Therefore there is urgent need of exact early antenatal diagnosis of NTD and appropriate intervention in pregnancy.

Material and Methods

The study was conducted over a period of 30 months in the imaging department of a large service hospital. The study material involved all the pregnant women attending the hospital and the peripheral family clinics in the area. The pregnant women who were referred in the second trimester for routine USG were studied. Detailed history and clinical data were recorded which included history of epilepsy, alcohol intake, diabetes mellitus, fever and any nutritional deficiencies in first trimester. History of abortion and previous pregnancy affected with NTD, family history of NTD with respect to woman and her husband were also recorded.

USG examination was conducted with 5 MHz sector transducer on Siemens Sonoline SL. First the fetal skull was scanned, BPD was recorded and compared with nomogram. If there was failure to demonstrate cranial vault superior to orbit in coronal view then volume and acoustic characteristics of the soft tissue were recorded. The brain was screened in ventricular, thalamic and cerebellar views. Subsequently, fetal spine was scanned in descending sequence starting from craniocervical junction to sacral region. Bony and soft tissue signs, associated anomalies of nervous system and others were observed carefully. Termination of pregnancy was advised and morphological details of all defects were noted after expulsion of fetus and correlated with USG findings.

Results

This study screened 1950 pregnant women and detected 14 cases of NTD, bringing the incidence to 7.1/1000 conceptions. The distribution of types of NTD revealed (Table-1) occult dysraphism 2(14%), meningocele 2(14%), myelocele 1(7%), myelomeningocele 2(14%), encephalocele 2(14%) (Fig-1) and anencephaly 5 (37%). In maternal age analysis 12 mothers were between 20-30 years and 2 were between 31-40 years of age in NTD positive mothers. Out of them 4 were primigravida and 10 were multigravida. Only 2 mothers had past history of NTD, 1 had anencephaly and other had meningomyelocele; in both the same NTD recurred during the gestation under study. 3 mothers had medical disease during gestation, 1 had malaria and the second had viral fever both associated with anencephaly while the third mother had gestational diabetes mellitus and was associated with myelocele. Maternal hemoglobin (Hb) correlation revealed that 10 mothers had Hb > 10 gm/dl, 3 had 8-10 gm/dl and only 1

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Fig. 1 : Fetal cranial scan showing bilobed cystic brain structure protruding out from occiput

had 6.9 gm/dl. The fetal BPD correlate revealed decreased BPD in 2 cases of encephalocele, increased BPD in 2 cases of meningocele, meningocele each and 1 case of myelocele. BPD was normal in cases of occult dysraphism, it could not be recorded in cases of anencephaly. Anterior Ventricular Hemispheric ratio (AVHR) analysis revealed < 50% in cases of occult dysraphism, could not be recorded in anencephaly and was > 50% in remaining NTDs.

TABLE 1
Neural tube defects

NTD Type	No of cases	Percentage
Occult dysraphism	2	14
Meningocele	2	14
Myelocele	1	7
Myelomeningocele	2	14
Encephalocele	2	14
Anencephaly	5	37
Total	14	100

The scalloping of frontal bones "Lemon sign" was seen in all cases of myelomeningocele, myelocele and meningocele (Fig-2). "Banana sign" the typical crescentic deformation of cerebellum with concavity pointing anteriorly was seen only in 1 case of myelomeningocele. The IV ventricle size correlation revealed inability to record in case of anencephaly (Fig-3), normal in 2 cases of occult dysraphism, increase in 1 case of encephalocele and decrease in the remaining cases of NTDs. Bone segment defect site correlate showed affection of cranium exclusive in all cases of anencephaly and encephalocele, thoracic spine in 1 case of myelomeningocele, thoracolumbar in 1 case of myelomeningocele, lumbar spine in 1 case of myelocele and lumbosacral spine in 2 cases each of meningocele and occult dysraphism.

The other neural defects correlation revealed Chiari II malformation in all cases of myelomeningocele, myelocele and meningocele. Chiari III and Dandy Walker anomalies were seen in one case each of encephalocele. Amniotic fluid volume correlate revealed increase in amniotic fluid volume in all cases of NTD except in occult dysraphism. Medical termination of pregnancy was performed in all cases of NTDs except the cases of occult dysraphism. The NTD morphological correlate with USG antenatal detection revealed that USG picked up all the cases except 2

Fig. 2 : Fetal head scan showing "Lemon sign" as a result of small posterior fossa and cerebellum.

Fig. 3 : Fetal head scan showing orbits but absent cranial vault.

cases of occult dysraphism, out of total 14 cases of NTDs. This gave accuracy to USG for antenatal diagnosis as sensitivity was 85.71%, specificity 100%, positive predictive value 100% and negative predictive value of 99.89% (Table-2).

TABLE 2
Characteristics of USG screening for NTD

Accuracy	Values (%)
Sensitivity	85.71
Specificity	100.00
Positive predictive value	100.00
Negative predictive value	99.89

Discussion

The study has brought out an incidence of 7.1/1000 conceptions in a composite population of Armed Forces. The incidence reported in western literature is much lower ranging from 0.3-4.1/1000 conceptions [4]. However, an Indian study by Sood et al [2] has published varying incidence ranging from 0.9 in Calcutta to 8.7/1000 in Chandigarh. The present study data is comparable to Indian data. The incidence was

much higher in mothers < 30 years of age, possibly due to higher conception rates in this age group. Multigravida were more affected than primgravidas. The higher incidence in this study could be due to the subjects studied living in highly polluted environment and stressful life style.

Amongst the various stypes of NTDs, anencephalies were highest being 37% followed by encephalocele, myelomeningocele, meningocele and occult dysraphism each being 14% and the least was myelocele being 7%. Winsor et al [5] found 35% anencephaly and 11% encephaloceles. Present study data is nearly comparable to that of Winsor [5]. The study revealed recurrence in 2 cases of previously affected gestations with NTDs. It is also in consonance with the known increased recurrence rate of 4% of NTDs with previously affected gestations [6]. This study did not have NTDs affected parents or thier blood relations. Other etiological associations observed in the study though already reported in the literature, had been febrile illness [7], gestational diabetes mellitus and anemia [8]. In this study, 4 women had anaemia. The USG signs in form of increased BPD, AVHR, Lemon sign and decreased IV ventricle in all cases of myelomeningocele, myelocele and meningocele were strongly suggestive of Arnold Chiari Type II malformation, an association well established in literature.

Thiagarajah et al [9] found Lemon sign more reliable before 24 weeks period of gestation alongn with other signs. Arnold Chiari type II malformations - 5 cases , Type III malformation 1 case and Dandy Walker malformation 1 case were the other nervous system associated defeects with NTDs in this study. These nervous system defects have been reported in literature [10].

This study did not detect any non nervous system associated malformations. This study has established the diagnostic accuracy of antenatal USG after morphological post expulsion correlation as sensitivity of 85.71%, specificity of 100%, positive predictive value of 100% and negative predictive value of 99.89%. The reported accuracy data from various studies [6,8,11] reveal sensitivity of 30-98%, specificity of 96-100% positive predictive value of 80-100% and negative predictive value of 99-100% to which the

data of this study is comparable.

With the advent of three dimensional ultrasound the diagnostic potential for NTD has further enhanced as it delineates the exact nature and magnitude of the defect [12], however the limitation being high cost and limited availability of three dimensional ultrasound equipment.

Hence early prenatal USG is a very useful tool for detection of NTDs and thus providing early intervention in form of termination of pregnancy which prevents burden on society in care of a handicap. It is hoped that more advancement in the USG technology and progressively increasing user experience will permit earlier diagnosis making intervention more safe and acceptable.

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