# NEURAL TUBE DEFECTS : PREVALENCE AND OUTCOME IN A SOUTH INDIAN HOSPITAL

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## SUMMARY

**Neural tube defects (NTD) were the commonest anomalies encountered in our hospital with a prevalence of 6.3 per thousand births. Spina bifida and anencephaly constituted 50% and 44% of the NTDs respectively. Rare froms of NTD like exencephaly and iniencephaly were seen in one fetus each. Majority (78%) of the patients reported to the hospital only in the third trimester. Malpresentations were seen in 31% and hydramnios in 28% of the cases. Second trimester maternal serum alpha fetoprotein estimated in 6 patients, was found to be elevated. Fetal outcome has been poor with NTDs. Fetal demise occurred in utero in 62% while 16% had neonatal death. Early second trimester termination after prenatal diagnosis was done in 8% of cases while 30% had late termination/induction of preterm labour. Early booking for antenatal care, ultrasound and alpha feto-protein screening, termination of affected pregnancies and folic acid supplementation of affected pregnancies and folic acid supplementation of diet of all women in the reproductive age group, may have to be undertaken to bring down the prevalence of NTD in our population.**


## INTRODUCTION

Neural Tube Defects (NTD) are not only the most common fetal malformations but also the most devastating congenital anomalies. The birth prevalence of NTD varies in different countries. In India it is said to be high among Sikhs - 7 per 1000 births (Regine et al 1993). As a result

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*Accepted for Publication on Nov' 96*

of maternal serum alpha fetoprotein (MSAFP) screening and ultrasound (USG) prenatal diagnosis, births of babies with NTD is decreasing in the West. However in developing countries majority of the NTDs are detected late in pregnancy or after birth.

## *MATERIAL AND METHODS*

To study the prevalence and to evaluate the outcome of pregnancies with neural tube defects a retrospective analysis of 50 Neural tube defects encountered in 7977 deliveries and 504 second trimester abortions over a period of six years and four months (1989 January - 1995 • April)Obstetric and Gynaecology department of KMC Hospital, Manipal was done. Information was taken from the confinement register and case sheets.

The factors analysed were maternal age, parity, period of gestation, predisposing factors, type of NTD, clinical markers and fetal outcome.

## *RESULTS AND OBSERVATIONS*

The prevalence of NTD in our study was 6.3 per thousand births. NTDs constituted 41% of all the anomalies detected during the study period.

Table I shows that majority were open spina bifida (50%) with meningoceles / meningomyelocoeles. Anencephaly was seen in 44%. Exencephaly was seen in one case at 20 weeks of gestation. Iniencephaly aperta was seen in a case of missed abortion.

There were 4 women aged less than 20 years and one aged 40 years. Fifty six percent of the women were primigravidas and there were 2 grandmultiparas.

History of consanguinity was identified in 2 , they had married their first cousins. There were 2 frank diabetics. One was

**Table I**
**TYPES OF NEURAL TUBE DEFECTS**

| Types of NTD | Number - 50 | Percentage |
|---|---|---|
| Anencephaly | 20 | 40% |
| Spina bifida | 23 | 46% |
| Anencephaly with Spina bifida | 1 | 2% |
| Anencephaly with Rachischisis of upper spina | 1 | 2% |
| Encephalocele | 3 | 6% |
| Iniencephaly aperta | 1 | 2% |
| Exencephaly | 1 | 2% |

an epileptic who had taken anticonvulsants in the first trimester.

Table II shows that 70% of the women were in third trimester at presentation to the hospital, majority of them at term.

Twenty eight (56%) of the babies were females. Ambiguous genitalia was seen in 3 babies with spina bifida. Males and females were equally distributed among the spina bifida babies whereas there was a female preponderance seen in anencephalics.

Table III shows that fetal outcome was very poor in the presence of neural tube defects. There were only 4 terminations following prenatal diagnosis in early second trimester. In 15 patients induction of preterm labour or late second trimester abortions were done after the diagnosis. Thirty one (62%) had fetal demise in utero. There were 7 survivors with meningocele. They were asked to come for surgery at specified dates. One of them had residual problems following surgery. One baby died at the age of 8 months. Two are alive and well while the other 3 didn't come for surgery and were lost for follow up.

Second trimester MSAFP levels were estimated in 6 patients and were raised to more than 2.5 MOM in all. In the case

**Table II**
**PERIOD OF GESTATION AT PRESENTATION**

| Period of gestation (weeks) | Number - 50 | Percentage |
|---|---|---|
| 37 - 40 weeks | 23 | 46% |
| 28 - 36 weeks | 16 | 32% |
| < 28 weeks | 11 | 22% |

**Table III**
**FETAL OUTCOME**

| Fetal Outcome | Number - 50 | Percentage |
|---|---|---|
| Abortions | | |
| (i) Induced | 4 | 8% |
| (ii) Missed | 7 | 14% |
| Intrauterine deaths | 24 | 48% |
| Neonatal Deaths | 8 | 16% |
| Survivors | 7 | 14% |

**Table IV**
**NTD WITH OTHER ANOMALIES**

| Other Anomalies | Number | Percent |
|---|---|---|
| Multiple anomalies | 2 | 4% |
| Ambiguous genitalia | 3 | 6% |
| Gastroschisis | 1 | 2% |
| Omphalocele | 1 | 2% |
| Cleft palate | 1 | 2% |
| Spina bifida with related anomalies | | |
| (i) Ventriculomegaly | 9 | 18% |
| (ii) Talipes | 7 | 14% |

with exencephaly it was more than 10 MOM for the period of gestation.

Ultrasound had been done in 39 (78%) of these women. In the others it had not been done as they came in labour or in the process of abortion. In 5 women, spina bifida was missed despite the scan and these were isolated lesions. All nine cases of spina bifida with ventriculomegaly were diagnosed. All the anencephalic fetuses were also diagnosed by scan. Among the 22 anecephalics 6 were face presentations. There were 6 breech presentations in the spina bifida group. Hydramnios was seen in 28% of the cases in the third trimester and there was severe oligohydramnios in one case. There were 2 cases of twins where one of the babies had NTD and the other was normal. There were 5 cases where caesarean section had to be done. One of them was twins with one anencephalic baby with malpresentation. Other cases were either done for foetal distress or breech presentation where spina bifida was not detected.

Table IV shows associated anomalies. One of the patients whose foetus had multiple anomalies was a case of exencephaly with gastroschisis, talipes and cardiac defects. She was a frank diabetic. The other patient with a multiple anomaly foetus, had conceived following clomiphene induction for infertility. The fetus had features suggestive of trisomy 18. There was omphalocele, cardiac defects, overlapping of fingers and meningocele. Karyotype could not be performed. Abdominal wall defects were also seen in two other cases with spina bifida while one anencephalic baby had cleft palate.

## *DISCUSSION*

Our incidence of NTD is high compared to reports from other countries (Xiao et al 1990). However the prevalence of NTD varies not only from country to country but also between places within a country. This is evident from the fact that Kulkarni et al (1989) reported a high incidence of 11.4 / 1000 births in central Karnataka while our incidence in the southern part

of the same state is 6.3 / 1000 births. A general decline in birth incidence of NTDs is reported in the West mainly because of MSAFP screening and termination of pregnancy. Nutritional supplementation with folic acid may also be responsible for this declining trend (Andrew et al 1993). Neural tube defects are of multifactorial etiology. Some of the predisposing factors are diabetes, anticonvulsant exposure in early gestation, teenage pregnancies, hyperthermia, ingestion of analgesics, antiemetics and paternal occupation, with a higher incidence noted in painters (Lemire 1988, Brender & Snarez 1990). Pregnancy following induction of ovulation is said to have a higher incidence, but this is not proved (Lemire 1988). Kulkarni et al (1989) found higher incidence of NTDs in consanguineous families which suggests that there may be a genetic component. We had only 2 patients married to their first cousins. Majority of our cases did not have any predisposing factors. Reports regarding NTD and maternal parity are conflicting. We found higher incidence (56%) in primigravidas.

Ultrasound screening of pregnancies is very essential for early diagnosis of this disorder so that pregnancies can be terminated early. Anencephaly is easily diagnosed but isolated spina bifida is difficult to diagnose. In such cases MSAFP screening is of enormous value. Spina bifida has to be looked for carefully in the presence of breech presentation, hydramnios, and ventriculomegaly. Six of our cases with isolated spina bifida who were scanned did not have any other ultrasound sign and the lesion was missed in 5 ot them. Unfortunately none of them had presented in the 16 - 20 weeks of gestation when we do MSAFP screening. Cranial markers for diagnosis of spina bifida like the lemon sign, banana sign and absent cerebellum, aid in the diagnosis even prior to 17 weeks of gestation by transvaginal sonography (Blumenfeld et al 19993). Iniencephaly aperta and exencephaly are rare forms of NTD. We encountered one case each. Exencephaly, where a disorganised mass of cerebral tissue is exposed due to absence of cranial vault is said to be precursor of anecephaly. Usually by the end of first trimester, exencephaly progresses to anencephaly due to physical and chemical trauma of the exposed cerebral tissue. Why in some cases like ours, it persists well into the second trimester is not known (Yi-Chich et al 1992). Iniencephaly is characterized by an exaggerated dorsal flexion of the head associated with cephalocele and spina bifida. The spina is often abnormally short and deformed (Romero et al 1988). NTDs most often occur as isolated anomalies. Ventriculomegaly and talipes occur as a result of spina bifida. Other anomalies occurred in only 16% of cases in our series.

Fetal outcome is poor in babies with NTD as shown in this study. However cases with isolated spina bifida may do well with surgery after birth. Prognosis depends on the size and position of the lesion. The last word has not been said regarding the optimal mode of delivery for these fetuses. Breech presentation is common in these pregnancies and if delivered vaginally the prognosis may be bad. Small (<4 cm) lumbosacral lesions seem to have good prognosis regardless of mode of delivery. The contemporary approach is to perform abdominal delivery for fetuses affected with meningomyelocele larger than 4 cms or

when nonvertex presentation exists. Vaginal delvery can be allowed for small lesions with vertex presentations (Charles et al 1993). In view of the high incidence of NTD in our study, awareness regarding early booking for antenatal care had to be created in the population. MSAFP and USG screening should be done. Folic acid supplementation for all women in the child bearing age can be tried, as primary prevention is now possible for a potentially lethal malformation. This may go a long way in decreasing the prevalence.

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