

## Original Research Article

# Epidemiological study of neural tube defects in Jammu division, India

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## ABSTRACT

**Background:** Prevalence and spectrum of neural tube defects has undergone seasonal and secular variations in different regions of the world since the turn of last century. However precise etiology inspite of lot of research has not been clearly elucidated.

**Methods:** The study was conducted in Government Medical College Hospital and SMGS Hospital Jammu in patients diagnosed to have a neural tube defect from birth to 19 years. History regarding sociodemographic profile, antenatal history and significant birth history was elicited. Complete general physical examination, Local examination of the lesion was carried out and neural tube defects were classified. All the cases were subjected to ultrasound head to determine ventricular size and rule out hydrocephalus. The data was entered in Microsoft Excel and results were presented in form of percentages and proportions.

**Results:** Maximum children admitted were in the age group of <1 month and belonged to Jammu district. Commonest anomaly seen was lumbosacral meningocele with associated hydrocephalus and neural deficit. Mothers who gave birth to an NTD affected child were commonly in the age group of 20-30 years, with commonest NTD affected sib being 1stborn and most of parents being from very low socio-economic strata of society. Only 7 mothers had received antenatal advice with ultrasound and almost none had received folic acid either before or after conception.

**Conclusions:** Neural tube defects appear to be fairly common anomaly among females of lower socioeconomic strata, living in far flung mountainous regions of the state, devoid of proper health care facilities and education.

**Keywords:** Encephalocele, Jammu, Meningocele, Meningocele, Neural tube defect

## INTRODUCTION

Neural tube defects (NTDs) are one of the most common congenital anomalies reported so far. They are malformations secondary to neural tube closure that occur between 3<sup>rd</sup> and 4<sup>th</sup> week of gestational age. The precise aetiology and specific genes that may be involved during this abnormal neural ontogenesis and resulting in various NTDs have not yet been elucidated. However, over the last century teratogens implicated in the

aetiology of NTDs include potato blight, hyperthermia, low socio-economic status and drugs like antihistamines, sulphonamides, anticonvulsants etc. Of all the suspected teratogens, carbamazepine, valproic acid and folate deficiency, have most strongly been identified with the development of NTDs.

Variation in incidence of NTDs is multifactorial worldwide and epidemiological data from the period before in utero diagnosis and termination of affected pregnancies established two trends. First illustrated a

varying worldwide epidemic that cycled over years in the past and second established a general decline in birth incidence over recent several decades. Incidence varied from 0.03-0.21/1000 live births in North West Pacific to 2.5-3.5/1000 live births in Nova Scotia. Highest reported incidence of NTDs is among Celts in UK, Canada and USA.<sup>1</sup> which suggested importance of a genetic component that has been borne out in family studies and fall in incidence has been attributed to termination of affected pregnancies and periconceptional use of folic acid. The spectrum of anomalies included in neural tube defects is true occult spina bifida, meningocele, meningomyelocele, encephalocele, anencephaly and craniospinalrachischisis.<sup>2</sup> Spina bifida and anencephaly comprise 90% of NTDs and encephaloceles 10%.

The diagnosis of NTDs comprises two aspects, first being antenatal assessment and second post-natal evaluation of baby. Regarding antenatal assessment two tests commonly done are, serum  $\alpha$ -fetoprotein and ultrasound. Multiple measurements of  $\alpha$ -fetoprotein are taken between 15<sup>th</sup> and 20<sup>th</sup> weeks of gestation and a significant rise >1000 ng/ml at 20 weeks gives 75% accuracy whereas amniotic fluid  $\alpha$ -fetoprotein gives 95% accuracy in diagnosis of NTDs, although latter is not the preferred method.<sup>3</sup>

A significant reduction in occurrence and recurrence of NTDs has been seen following periconceptional use of folic acid both in females with NTDs affected pregnancy and other females of reproductive age group which has further decreased the world-wide incidence of these dreaded malformations. Epidemiological data regarding prevalence of neural tube defects in different geographical regions can give valuable information regarding its determinants which may further aid in proper planning of health services.

The aim of the present study was to describe the spectrum of neural tube defects among children in Jammu division.

## METHODS

The study was conducted in Government Medical College Hospital and SMGS Hospital Jammu in patients diagnosed to have a neural tube defect. Detailed History was taken from parents or family member or guardian accompanying the patient regarding age and sex of child, age at presentation, age of parents, occupation etc. Interval between pregnancies, abortions/still births in early pregnancies, Diabetes, Hypertension, Tobacco smoking, exposure to irradiation, epilepsy, thyroid disease, TORCH was taken. History of any drug intake during three months of age, Ultrasound findings, use of iron folic acid.

### Examination

Following history, complete general physical examination was conducted. Local examination of the

lesion was carried out and neural tube defects classified as

- True occult spina bifida,
- Occult spina bifida,
- Meningocele,
- Meningomyelocele,
- Encephalocele,
- Anencephaly,
- Spinalrachischisis,
- Cranioschisis.

All the cases were subjected to ultrasound head to determine ventricular size and rule out hydrocephalus. The data was entered in Microsoft Excel and analysed. Results were presented in form of percentages and proportions.

## RESULTS

Authors studied 50 children admitted in Govt Medical College Jammu from 10 December 2016 to 31 December 2017 diagnosed with neural tube defects.

Regarding age distribution of cases with Neural tube defects, Mean age was 13.56 months with age range of 1 day and 19 years. Maximum number of children i.e. 26 (52%) were in age group of less than 1 month, both in males and females with Male to female ratio of 0.9:1 (Table 1). Commonest age group of parents was 20-30 years (48%) followed by 10-20 years (27%), 4 persons were more than 40 years and oldest was 46 years old male from Surankote Poonch (Table 2).

**Table 1: Age Distribution of children diagnosed with neural tube defects.**

Age	Male	Female	Total
≤1 month	13 (54.2%)	13 (50%)	26 (52%)
2-4 months	5 (20.8%)	2 (7.7%)	7 (14%)
4-6 months	1 (4.2%)	4 (15.4%)	5 (10%)
>6 months-19 years	5 (20.8%)	7 (26.9%)	12 (24%)
Total	24 (100%)	26 (100%)	50 (100%)

**Table 2: Distribution of subjects according to age of their parents.**

Age of parents	Father	Mother	Total
<20	11	16	27
20-30	22	24	46
31-40	13	10	23
>40	4	0	04
Total	50	50	100

Regarding birth order, in 21 cases (42%) 1<sup>st</sup> child was affected, in 13 cases (26%) 2<sup>nd</sup> was affected, in 8 (16%) 3<sup>rd</sup> child was affected, in 4 (8%) cases 4<sup>th</sup> and in 4 (8%) cases 5<sup>th</sup> child was affected.

Maximum interval between NTD affected pregnancy and previous pregnancy was 10 years in a patient belonging to Budhal Rajouri. Commonest interval between NTD affected pregnancy and normal pregnancy was (<1) year post marriage, with an average of 10.2 months and second common was of (2-4) years, average being 28.8 months.

Regarding geographical distribution of cases, maximum 21 (42%) belonged to Jammu district followed by Rajouri district 9 (18%) and only 1 case (2%) from Doda district was reported. Maximum cases i.e. 32% belonged to socioeconomic status class V as per modified BG Prasad classification of socioeconomic status (Table 3).

Regarding education status of parents 22 (42%) of fathers and 39 (78%) of mothers were illiterate, 25 (50%) of fathers and 10 (20%) of mothers had an educational qualification below 10th standard, whereas only 3 (6%) fathers and 1 (2%) mothers had received education beyond 10<sup>th</sup> standard. Maximum qualified person was a graduate from Jammu city.

**Table 3: Distribution of subjects according to sociodemographic characteristics.**

Region	No.	Percentage (%)
Jammu	21	42
Rajouri	9	18
Poonch	7	14
Udhampur	5	10
Kishtwar	4	8
Reasi	3	6
Doda	1	2
Total	50	100
Socioeconomic status		
I	13	26
II	10	20
III	11	22
IV	10	20
V	16	32

**Table 4: Type of neural tube defects in children.**

Type of neural tube defect	n	%
Meningocele	18	36
Meningomyelocele	24	48
Encephalocele	8	16
Total	50	100

Antenatal advice was sought by 7 mothers and antenatal USG was done in 2<sup>nd</sup> and 3<sup>rd</sup> trimester only in 6 cases. Out of these, three showed a normal fetus, three showed meningomyelocele and two showed hydrocephalus.

Out of 50 cases of Neural tube defects, Total numbers of cases in meningomyelocele (MMC) group were 24 (Table 4, 5).

**Table 5: Distribution of subjects according to types of meningomyelocele.**

Type	n	%
Meningomyelocele (N=24)		
Lumbosacral	9	18
DorsoLumbar	4	8
Lumbar	4	8
Cervical	3	6
Sacral	3	6
Cervical Thoracic	1	2
Meningocele (N=18)		
Type	n	%
Occipital	3	6
Cervical	3	6
Thoracic	2	4
Lumbar	3	6
Lumbosacral	1	2
Sacral	1	2
Lipomenigocele		
Lumbosacral	4	8
Cervical	1	2
Encephalocele (N=8)		
Type	n	%
Occipital encephalocele	6	12
Occipital meningoencephalocele	1	2
Spina bifida	1	2
Total	50	100



**Figure 1: Lumber meningomyelocele.**

Associated by hydrocephalus was present in 13 cases. Commonest type was lumbosacral followed by dorsolumbar, lumbar, cervical, sacral and cervical thoracic. There were 18 cases of meningocele. Associated hydrocephalus was seen only in 1 case of sacral meningocele.

Regarding types, commonest anomaly was lumbosacral lipomenigocele (Figure 1). Out of total no of 18 cases, 5

cases i.e. 27.7% had lipomeningocele. Associated neural deficit was seen in 22 (44%) of cases, out of which 16 (32%) were in meningomyelocele group, 5 (10%) were in meningocele group. There were total 8 cases of encephalocele, commonest was occipital encephalocele. One case of occipital encephalocele had associated hydrocephalus (Figure 3).



**Figure 2: Thoracolumbar meningocele.**



**Figure 3: Occipital encephalocele.**

## DISCUSSION

In the present study, the spectrum of Neural tube defects along with epidemiological features was studied. Regarding sex, maximum children were females 26 (52%), though sex difference was not statistically significant. As observed in the context of available literature Airede Ik, Mahadevan and Bhat et al, also found higher incidence in female sex.<sup>4,5</sup> Maximum number being from Jammu district could have been due to close proximity of our institution to these areas and inaccessibility of other areas might have resulted in their lesser number in the study sample. Rest of the children were almost equally distributed among other districts and

most of them came from far flung mountainous areas of the state.

Commonest anomaly found in the study sample was meningomyelocele 24 (48%). Other authors also reported similar result.<sup>4,6,7</sup> However Mahadevan and Bhat found spina bifida (54.8%) as the most common anomaly followed by anencephaly (31.6%) and encephalocele (11.6%).<sup>5</sup>

Low frequency of spina bifida occulta may have been due to inability of patients to come from far flung and mountainous areas of state, as this anomaly is not associated with major morbidity or mortality and few cases are referred to tertiary institutes.

Aguiar found low incidence of NTDs among women who had more than three pregnancies, same being the case in our study also.<sup>7</sup> Campbell et al, found 1st child to be affected with NTDs in maximum number of cases.<sup>8</sup> Todroff et al, found an increased NTD risk for mothers with an interpregnancy interval of ( $\leq 6$ ) months compared with ( $>12 - \leq 24$ ) months, with the risk being greatest among mothers whose previous pregnancy was a live birth and not spontaneous abortion or elective termination.<sup>9</sup> Present study also showed commonest interval of  $\leq 1$  year between NTD affected pregnancy and marriage, however NTD affected pregnancy was commonly preceded by normal pregnancy or marriage. Commonest age group of mothers to have an NTD affected pregnancy was 20-30 years (48%), 10-20 years (32%) followed by (20%) in  $>30$  years age group. This corroborated with findings of other study as well.<sup>4</sup>

Campbell et al, have shown a progressive increase in prevalence rate of neural tube defect from higher to lower socio-economic class as determined by father's occupation and income. Most of parents in our study group were either farmers or labourers which was consistent with findings of Campbell et al.<sup>8</sup> Vrijheid et al, also suggested higher NTD risk among families of lower socio-economic status.<sup>10</sup>

Antenatal advice was sought by 7 women only and antenatal USG was done in 2<sup>nd</sup> and 3<sup>rd</sup> trimester only in 6 cases, out of which three showed a normal fetus, rest 3 showed meningomyelocele with 2 showing hydrocephalus. This again indicates ignorance of people and lack of health infrastructure in remote and inaccessible areas of the state which resulted in late diagnosis of these congenital malformations.

Cherian et al, in a door to door survey of mothers in remote village clusters in Balrampur district in Uttar Pradesh also found poor health care and drug delivery system which resulted in high incidence (6.52-8.21/1000) live birth's of NTDs.<sup>11</sup>

As is evident from our study, most of these NTD births have occurred in children of parents residing in far off, inaccessible and mountainous areas of the state with poor infrastructure for health and education, with low socio-economic status.

## CONCLUSION

Authors conclude from the present study that neural tube defects appear to be fairly common anomaly among females of lower socioeconomic strata, living in far flung mountainous regions of the state, devoid of proper health care facilities and education.

Authors emphasize upon a healthy policy for folic acid fortification of food or folic acid tablets to be distributed by health care workers to all females in reproductive age group, as also antenatal  $\alpha$ -fetoprotein estimation, USG examination of all pregnant females and termination of NTD affected pregnancies which will significantly reduce the incidence of these dreaded malformation and associated neural deficits.

However, as present study sample was small we suggest a large study over an extended period of time to know about prevalence/incidence of these congenital malformation in our state, which will also help to formulate a comprehensive policy for prevention of NTDs in future.

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