Clinico-epidemiology of neural tube defects and associated anomalies in pediatric patients in north west Rajasthan

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ABSTRACT

Background & objectives: Having adequate surveillance data is essential for the purposes of prevention and evaluation, given the substantial influence of birth defects on the death rate of children below the age of 5. The purpose of this study was to assess the clinical and epidemiological features of neural tube abnormalities and associated anomalies in pediatric patients residing in the north-western region of Rajasthan. **Methods:** Each attendee of the patients provided a well-informed written consent. Following that, a clinical evaluation was performed to assess the functioning of the sensory and motor systems, any anomalies related to the musculoskeletal system, as well as the functioning of the bladder and intestine, taking into account the presence of the spina bifida defect. The results were subsequently documented. **Results:** 56% of the persons were between the age range of 0 to 1 year, whereas 68% of them resided in metropolitan regions. Preterm children and multipara individuals had a greater frequency of neglected tropical diseases (NTDs), with rates of 56% and 58% respectively. Among the entire population, 70% were found to have myelomeningocele, 24% were found to have meningocele, and 84% had surgical intervention. The recorded survival rate stands at 76%. **Conclusions:** Male individuals exhibited a higher prevalence of NTDs, while spina bifida demonstrated a higher incidence rate.

Key words: Neural tube defects, incidence, clinico epidemiology, management

INTRODUCTION

Neural tube defects (NTDs) are serious birth disorders of the central nervous system that occur during the development of an embryo and are caused by the inability of the neural tube to close properly. NTDs are prevalent among the majority of human birth abnormalities. The prevalence of spina bifida is believed to be between one and two cases per 1000 individuals in the general population. However, specific populations may have a notably greater prevalence due to genetic predisposition. Incidence also varies significantly across different geographic regions.[1]

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Neural tube defects are often categorized as either open or closed, depending on whether neural tissue is exposed or not.

Open neural tube defects encompass anencephaly, spinalrachischisis, spina bifida aperta/cystic (such as myeloschisis, myelomeningocele, and mening ocele), and encephalocele. Examples of closed neural tube anomalies include spina bifida occulta, which encompasses lipomatous malformations such as lipomas and lipomyelomeningoceles, split cord malformations like diastematomyelia and diplomyelia, neurenteric cysts, dermal sinuses, tethered spinal cord, and sacral agenesis (caudal regression). Among the various types of open neural tube defects, myelomeningocele is both the most prevalent and the most severe birth defect that can be survived.[2]

Due to the significant impact of birth abnormalities on the mortality rate of children under the age of 5, it is crucial to have sufficient surveillance data for the goals of prevention and evaluation. Nevertheless, the precise determination of the frequency of NTDs in the majority of nations remains uncertain mainly as a result of inadequate and fragmented data gathering. Therefore, we undertook this study to evaluate the clinical and epidemiological characteristics of neural tube abnormalities and related anomalies in pediatric patients in the northwestern region of Rajasthan.

MATERIALS AND METHODS

This study was conducted on a cohort of 50 pediatric patients aged between 0 and 14 years, from August 2020 to December 2022, at PBM Pediatric Hospital in Bikaner. A well-informed written consent was obtained from the attendants of each patient. Subsequently, a clinical assessment was conducted to evaluate sensorimotor function, orthopaedic abnormalities, and bladder and bowel function, while also considering the spina bifida defect. The findings were then recorded.

Diagnostic procedures including X-ray of the spine, ultrasonography of the kidneys, ureters, and bladder (KUB), and magnetic resonance imaging (MRI) of the brain were performed. Surgical procedures were performed based on the patients' clinical status and radiological findings.

The patients were monitored for an average follow-up duration of 1-2 years. All patient data was obtained in its entirety according to the proforma, and necessary tests were conducted using the Epi Info software, version 3.5.3, whenever applicable.

RESULTS

The highest percentage was observed in the age category of less than one year (56%), while the lowest percentage was recorded in the age range of 6-12 years (6%). The male to female ratio was 3:2. 68% (34) of the individuals were residing in urban regions, while 32% (16) were residing in rural areas. (Table 1)

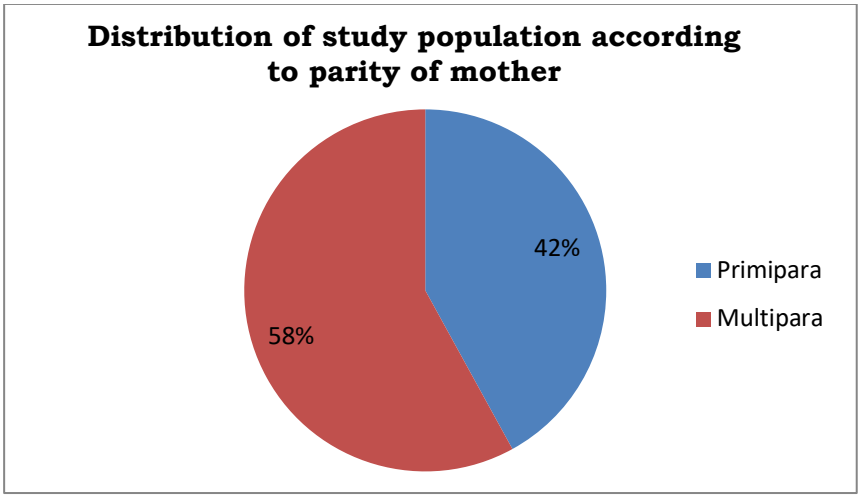
Table 1. Socio-demographic profile of study participants

Age group (years)	No. of cases	Percentage
≤1	28	56.0
1 – 2	10	20.0
2 – 5	9	18.0
6 – 12	3	6.0
Sex		

Male	30	60.0
Female	20	40.0
Residential Area		
Rural	16	32.0
Urban	34	68.0

Among the total of 50 cases, the prevalence of NTDs was greater (56%) in preterm children compared to term children (44%). The prevalence of NTDs was higher in multipara born children (58%) compared to primipara born children (42%). Before and throughout pregnancy, 96% of individuals did not receive folic acid supplementation, while only 4% received supplements during pregnancy. (Figure 1)

Figure 1: Pie chart showing parity of mother



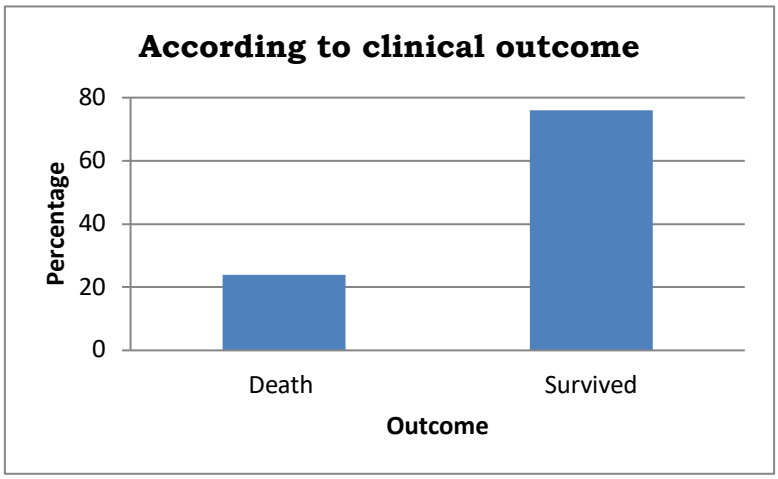
The biggest proportion, 70%, had myelomeningocele, while 24% had meningocele, and encephalocele was present in 6% of cases. A majority of children (56%) experienced urinary incontinence, 44% had bowel problems, and 76% were unable to walk. 84% had surgical procedures, while the remaining 16% utilized alternative methods such as physiotherapy. (Figure 1)

Table 2: Distribution of study population according to Neural Tube Defects and associated anomalies

Neural Tube Defects	**No. of cases**	**Percentage**
Myelomeningocele	35	70.0
Menigocele	12	24.0
Encephalocele	3	6.0
Associated anomalies		
Urinary incontinence	28	56.0
Bowel dysfunction	22	44.0
Unable to ambulate	38	76.0

The survival rate was 76%, with only 24% of children experiencing mortality. Among the 30 male cases, the biggest proportion, 73.33%, had myelomeningocele, while 23.33% had meningocele, and encephalocele was present in 3.33% of cases. Among the 20 female instances, myelomeningocele was the most prevalent condition, occurring in 65% of cases. Menigocele was present in 25% of cases, while encephalocele was found in 10% of cases. (Table 2 and Figure 2)

Figure 2: Bar chart showing distribution of subjects according to clinical outcome



DISCUSSION

Within our research, 28 instances (equivalent to 56%) occurred in individuals under the age of 1. Additionally, 10 instances (equivalent to 20%) were observed in individuals between the ages of 1 and 2, 9 instances (equivalent to 18%) were observed in individuals between the ages of 2 and 5, and 3 instances (equivalent to 6%) were observed in individuals between the ages of 6 and 12. The greatest values were observed in the age category of less than one year, while the lowest values were found in the age category of 6-12 years. In a similar vein, Basma Sadik and colleagues (2017)[3] discovered that the majority of cases were in infants younger than 6 months old, accounting for 57.7% of the total.

Our study found a higher prevalence of male patients (60%) compared to female cases (40%). The male to female ratio was 3:2. In a study conducted by Vijayalakshmi Kandasamy et al. (2015)[4], it was discovered that 55% of the participants were male. In contrast, Garg et al. (2020)[5] discovered that a total of 9 infants were born with neural tube defects (5 female and 4 male).

Our study found that 96% of participants did not receive folic acid supplementation prior to and during pregnancy, whereas only 4% took supplements throughout pregnancy. A minuscule number of mothers opted for folic acid supplementation. According to Basma Sadik et al (2017)[3], although 69.0% of the mothers took folic acid, the majority did not take it regularly.

Our investigation revealed that the biggest proportion, 70%, had myelomeningocele, while 24% had meningocele, and encephalocele was present in 6% of cases.

In a similar vein, Basma Sadik et al. (2017)[3] discovered that spinal defects were more prevalent than cranial lesions. Spina bifida was the most prevalent anomaly, accounting for 73.2% of cases, while encephaloceles accounted for 26.8%. Theophilus et al (2016)[6] discovered that 72% of the cases were Myelomeningocoeles.

The majority of children in our study (56%) experienced urinary incontinence, while 44% had bowel dysfunction and 76% were unable to ambulate. In a similar vein, Basma Sadik and colleagues (2017)[3] discovered that 30% of the subjects had sensory loss or bladder bowel incontinence.

Within our study, 84% of participants underwent surgical procedures, while the remaining 16% pursued alternative modalities such as physiotherapy. Another study (2016)6 discovered that, among 53 cases of spina bifida, all patients except two had surgical intervention to repair and close the spinal defect.

The study revealed a notable survival rate of 76%, with a corresponding mortality rate of 24% in youngsters. Similarly, Another study (2016) discovered that 90% of the patients experienced uneventful procedures.

There is a lack of randomized control trials in India investigating the impact of folic acid on neural tube defects (NTDs). Indian women are still recommended to take folic acid before pregnancy. In India, there are currently no official initiatives promoting the addition of folic acid to food products as a means of preventing neural tube defects (NTDs).

CONCLUSION

Early diagnosis of the illness can be achieved with screening tests like as AFP (alpha-fetoprotein) and early USG (ultrasound) performed between 16-20 weeks. This allows for the earliest feasible decision on termination. The occurrence of NTDs has decreased in comparison to previous studies, primarily due to factors such as prenatal screening for fetal developmental abnormalities and the administration of folic acid supplements before and throughout pregnancy. Additionally, it highlights the higher prevalence of NTDs in boys and the increased occurrence of spina bifida.

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AUTHORS' CONTRIBUTION

All the authors have contributed equally.

CONFLICT OF INTEREST

The authors declare no conflicts of interest.

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