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ANTENATAL DIAGNOSIS AND PREVALENCE OF NEURAL TUBE DEFECTS IN NORTHWESTERN INDIAN STATE (RAJASTHAN): A TERTIARY CARE CENTER STUDY

Radiology

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ABSTRACT

Introduction – Neural tube defects (NTDs) which are congenital abnormalities of the brain and spinal cord are among the leading causes of infant morbidity and mortality and continue to be a serious public health problem. They are caused by a failure of the neural tube to close completely, which results in the incomplete formation of the brain or spinal cord. **Methods-** This was a hospital- based cross- sectional study carried out in the Hadoti region of Rajasthan covering a period of 2 years (Jan 2023 to Jan 2025). All pregnant women referred from antenatal clinic were screened for congenital anomalies throughout their pregnancy irrespective of the gestational age. Gravid females between age 18 to 45 years were included in the study. **Results** - The study surveyed 39,600/-women, total of 33 neural tube defect cases are detected. The overall prevalence rate of neural tube defect was 8.33 per 1000 birth outcomes. Acrania-anencephaly was the most prevalent neural tube defect (17), followed by spina bifida (8), occipital encephalocele (6). **Conclusion-** The study found high prevalence rate of neural tube defect in Hadoti region which is far more compared with other states of India. There is urgent need to strengthen the quality of antenatal care services and prophylactic folic acid supplementation programme for prevention of neural tube defects.

KEYWORDS

neural tube defect, spina bifida, antenatal prevalence.

INTRODUCTION

Neural tube defects (NTDs), which are congenital abnormalities of the brain and spinal cord, are among the leading causes of infant morbidity and mortality and remain a significant public health concern. They occur due to the incomplete closure of the neural tube, resulting in the improper formation of the brain or spinal cord. NTDs are generally classified into two categories: brain defects—such as encephalocele, anencephaly, and cranial meningocele—and spinal cord defects—such as myelomeningocele, spinal meningocele, and lipomeningocele[1]. A 2024 systematic review and meta-analysis encompassing 27 studies across India reported a pooled NTD prevalence of 9.46 per 1,000 births (95% CI: 8.01–10.91). The study highlighted significant regional variations, with some areas exhibiting higher rates than others[2].The estimated global prevalence of NTDs is approximately **2 per 1,000 births,** equating to around 214,000–322,000 affected pregnancies annually. Neural Tube Defects (NTDs) arise due to a combination of genetic and environmental influences. Non-genetic risk factors include inadequate maternal nutrition, pre-existing diabetes, exposure to valproic acid (an antiepileptic drug) during early pregnancy, and a history of a prior NTD-affected pregnancy. Among these, insufficient folate levels in the mother prior to conception and during the early stages of pregnancy is the most significant—and preventable—contributor[3]. Neural tube defects (NTDs) continue to pose a significant public health challenge in low- and middle-income countries. Ultrasonographic diagnosis of NTDs typically occurs during the second trimester of pregnancy. The diagnostic sensitivity of prenatal ultrasonography (USG) for detecting NTDs in high-risk populations is approximately 97–98%, with a specificity of 100%[4].

METHODS

This hospital-based cross-sectional study was conducted at J.K. Lone Hospital, Kota, located in the Hadoti region of Rajasthan, over a two-year period from January 2023 to January 2025. The study population comprised all married women of reproductive age (18–45 years) who attended the antenatal clinic during this period. The study was conducted in an urban setting.

All pregnant women referred from the antenatal clinic to the radiology department for ultrasonography were screened for congenital anomalies throughout the course of their pregnancy, irrespective of gestational age. Prenatal ultrasonographic evaluations were performed by experienced radiologists.

A standardized data collection protocol was used. Information collected included socio-demographic characteristics, folic acid supplementation, medication use during or early in pregnancy, underlying maternal health conditions, timing and location of ultrasonography, and the timing of NTD diagnosis.

Data were entered into Microsoft Excel 2019 and analysed using IBM SPSS Statistics version 22.0. The prevalence of neural tube defects (NTDs) was estimated among all ever-married pregnant women aged 18–45 years during the study period. Quantitative variables were expressed as frequencies and percentages. Prevalence was calculated by dividing the number of identified NTD cases by the total number of birth outcomes. Ninety-five percent confidence intervals (95% CI) were computed. Additional data from hospital records concerning the mothers of NTD-affected pregnancies were also collected and descriptively analysed.

RESULTS

- During the study period from 1 JAN 2023 to 31 JAN 2025, a number of 39,600 women were screened by USG for antenatal detection of neural tube defects. A total number of 33 NTD cases were detected. Out of which 6 cases were Live birth, 10 were Still birth and 17 mothers underwent MTP following antenatal diagnosis. The overall prevalence rate of NTD in our study was **8.33 per 1000** birth outcomes with 95% CI (0.4187 to 0.853).
- **Distribution and types of NTD**: In the years Jan 2023–Jan 2025, a total of 33 cases of NTD were analysed. The most prevalent NTD was Acrania-anencephaly, with the prevalence **4.29 per 1000** birth 95% CI (0.2115 to 0.5423) (Figure 1), followed by spina bifida **2.02 per 1000** birth 95%CI (0.1082 to 0.3667) (Figure 2), occipital encephalocele (Figure 3) with a prevalence of **1.52 per 1000** birth 95%CI (0.01093 to 0.1453) and lastly craniorachischisis **0.25 per 1000** 95%CI (0.0035 to 0.1128).
- **Socio-demographic and obstetrical characteristics of mothers having NTD child:** A detailed overview of various factors was associated with a cohort of 33 pregnancies, offering insights into maternal characteristics, medical history, folic acid consumption, antenatal care and USG evaluations. **Maternal age** distribution indicated that the majority of participants fell within the 20–24 age range (48.5%), followed by 25–29 (30.3%). **Parity** analysis revealed that half of the pregnancies were from women with a parity of 1 (50.0%), with decreasing frequencies for higher parities. Medical history during pregnancy highlighted diverse conditions such as fever, cold, cough, anticonvulsant consumption, gestational diabetes, anaemia, mental disorders and thyroid issues, each with varying prevalence.
- Regarding **folic acid** consumption, 48.7% of participants took iron folic acid, while 51.3% did not. The timing of folic acid intake

varied, with 40.0% taking it regularly during pregnancy. All pregnancies were singleton.

- The provided table summarised key variables related to a study population of 33 individuals, shedding light on the demographic and clinical characteristics of the sample. **Birth order** analysis revealed that half of the individuals were firstborns (50.0%), while the second birth order constituted 40.0% of the sample. Notably, there was a decreasing frequency with higher birth orders Examination of birth years demonstrated variability. Delivery settings predominantly favoured hospitals (96.7%), underscoring the importance of institutional birthing environments. Regarding **birth outcomes**, the majority resulted in MTP (51.51%), with 30.3% ending in SB and 18.2% in LB. Furthermore, the table outlined the distribution of NTD types, revealing anencephaly as the most prevalent (51.51%), followed by spina bifida (24.24%). Occipital encephalocele (18.2%) and craniorachischisis were less common, comprising 3.0%.

Figure 1 – Shows complete absence of cranial vault with relatively disorganized brain tissue which is exposed.

Figure 2 – Showing lower occipital bone defect with outward protrusion of the brain tissue.

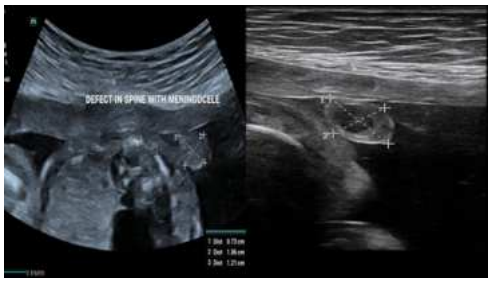

Figure 3- shows posterior spinal defect myelomeningocele- closed spinal dysraphism.

Table 1. Socio-demographic and obstetrical characteristics of women reported with a pregnancy with neural tube defect

Variable	category	Frequency (%) (N=33)
Maternal age	15-19	1
	20-24	16
	25-29	10
	30-34	4
	35-40	2
Parity	1	17
	2	13
	3	3
	4	0
Medical history during pregnancy	Gestational diabetes	2
	Hypertension	1
	Viral fever	1
	Thyroid	0
	Anaemia	2
Previous history	Family history	0
	Mother previous birth history	1
Folic acid consumption	Taking iron folic acid tablets	16
	Not taking iron folic acid tablets	17
Birth Outcomes	MTP	17
	Still birth	10
	Live birth	06

Table 2. Types of Neural tube defects

Category	Number	Percentage (%)
Anencephaly	17	(51.51%)
Spina bifida	8	(24.24%)
Encephalocele	6	(18.18%)
Craniorachischisis	1	(3.0%)
Combined	1	(3.0%)

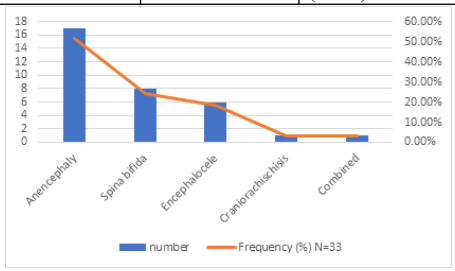

Chart 1- bar chart showing the distribution of various types NTDs

DISCUSSION: -

- The present study, conducted at a tertiary care centre in Rajasthan, highlights the regional prevalence and antenatal detection of neural tube defects (NTDs)—a significant group of congenital anomalies associated with high rates of morbidity and mortality.
- During the study period from January 2023 to January 2025, a total of 33 cases of NTDs were identified among 39,600 screened pregnancies, yielding an overall prevalence of 8.33 per 1,000 birth outcomes. This finding aligns with previous Indian studies, which have reported prevalence rates ranging from 1.5 to 11 per 1,000 live births, varying by geographic region and healthcare accessibility.
- In contrast, Mahalik et al. (2024) [4] reported a lower prevalence of 0.59 per 1,000 birth outcomes in the eastern Indian state of Odisha, based on a large population-based cross-sectional study involving 49,215 women and over 50,000 birth outcomes. Rai et al. (2016) [5], studying a socioeconomically similar population in rural Eastern Uttar Pradesh, reported a substantially higher prevalence of 7.48 per 1,000 live births. Internationally, Berhane and Belachew (2022) [6] reported an alarmingly high incidence of 107.5 per 10,000 live births in Eastern Ethiopia, coupled with a low antenatal detection rate of only 21.6% and virtually no periconceptional folic acid use.
- These regional differences within India (Table 3) can be attributed to variations in folic acid supplementation compliance, dietary patterns, and socio-cultural factors. In contrast, studies from developed countries consistently report lower NTD prevalence, which may be explained by mandatory folic acid supplementation, improved health-seeking behaviours, access to preconception care, food fortification policies, early screening, and therapeutic interventions during pregnancy. This stark contrast underscores the global disparity in NTD burden and prevention efforts, particularly in resource-limited settings.
- The detection rate in our study was 0.0833%, which is comparable to detection rates reported in other developing countries.
- Our study reaffirmed the most prevalent NTD subtypes, with spina bifida and anencephaly being the most common, consistent with findings from Morocco (Forci et al., 2021)[7], where spina bifida accounted for 38.6% and anencephaly for 50% of cases. Similarly, Berhane and Belachew (2022)[6] reported anencephaly in 48.1%, spina bifida in 22.6%, and myelomeningocele in 10.5% of NTD cases. However, our findings differ from those of Mahalik et al. (2024) [4], who reported spina bifida as the most common subtype (56.6%), followed by anencephaly (33%) and encephalocele (6.7%).

Table 3 - Table representing the type of study and the prevalence of NTDs across various places in India

Place of Study	Type of Study	Prevalence (per 1000)
Pondicherry	Hospital based	5.7
Balrampur	Population based	8.21
Lucknow	Hospital based	3.9
Odisha	Hospital based	9.12
West Bengal	Hospital based	8.2
Andhra Pradesh (united)	Hospital based	5.08
Kerala	Hospital based	10.6
Karnataka	Hospital based	3.15
Gujarat	Hospital based	6.4
Maharashtra	Hospital based	1.68

Strengths And Limitations

1. This is the first study of its kind in Rajasthan to estimate the antenatal prevalence of neural tube defects (NTDs).
2. It is among the few studies conducted to identify congenital anomalies—particularly NTDs—through antenatal ultrasonography.
3. As this is a single-centre, tertiary care study, the prevalence rate observed may not be generalizable to the wider population.
4. The study excluded unmarried pregnant women, and a few cases were lost to follow-up.

CONCLUSION

The consistently high occurrence and limited prenatal detection of neural tube defects (NTDs) in Rajasthan highlight both medical and systemic shortcomings within the healthcare framework. To address this, efforts should focus on promoting early antenatal registration, ensuring routine second-trimester anomaly scans, and strengthening folic acid supplementation programs. Key strategies include enhancing the training of rural sonologists, improving the accuracy of public health records, and increasing awareness among women about the significance of preconception health.

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Conflicts Of Interest - Nil

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