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Epidemiology of Neural Tube Defects in Jammu and Kashmir: A Systematic Review

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ABSTRACT :

Neural tube defects (NTDs) are among the most severe congenital malformations, presenting a significant public health concern in many low- and middle-income settings. This systematic review evaluates the epidemiology of NTDs in Kashmir, synthesizing data from observational studies, hospital-based reports, and public health records between 2000 and 2025. Findings indicate a variable but concerning prevalence, with spina bifida and anencephaly being the most frequently reported. Risk factors such as consanguinity, folic acid deficiency, and inadequate prenatal care are consistently implicated. The review highlights the urgent need for strengthened surveillance systems, population-based studies, and targeted public health interventions focused on maternal nutrition, early screening, and awareness.

Key Words: Neural Tube Defects, Spina Bifida, Anencephaly, Consanguinity, Folic Acid Deficiency, Maternal Nutrition etc.

Introduction

Neural tube defects (NTDs) are a group of serious congenital malformations arising from the failure of the neural tube to close during early embryonic development (Copp & Greene, 2010) (Ravi et al., 2021). Globally, NTDs remain a significant cause of neonatal morbidity and mortality, particularly in low- and middle-income countries (Lo et al., 2014) (Zaganjor et al., 2016). The incidence and pattern of NTDs vary widely due to a combination of genetic, nutritional, environmental and socio-cultural factors (Wharton-Smith et al., 2019) (Ochola et al., 2021). Jammu and Kashmir, a northern region of India, presents a unique epidemiological context characterized by consanguinity, nutritional deficiencies and limited implementation of folic acid fortification policies. This systematic review synthesizes the existing literature on the epidemiology of NTDs in Jammu and Kashmir, focusing on prevalence, types, risk factors, and temporal trends to inform policy and public health strategies. India, with its vast population and regional disparities in healthcare access and nutritional intake, reports varied incidence rates of NTDs across different states. Jammu and Kashmir, a northern region of India, is geographically and socio-culturally distinct, characterized by unique dietary patterns, seasonal food insecurity, high rates of consanguineous marriages, and limited access to fortified foods and perinatal care. These factors may exacerbate the risk of NTDs in the region, yet comprehensive epidemiological data remains scarce.

Despite national-level public health programs that promote folic acid supplementation and antenatal care, several pockets in India, including parts of Jammu and Kashmir, continue to witness high rates of congenital anomalies. A systematic assessment of the burden of NTDs in Jammu and Kashmir is essential to understand local risk factors, design targeted prevention strategies, and ensure evidence-based policy implementation. This review aims to

synthesize available literature on the epidemiology of NTDs in Jammu and Kashmir, including prevalence estimates, clinical profiles, associated risk factors, and policy implications.

Methods

Major lessons learned from explant analysis have been summarised on the basis of a systematic review of the literature and of the data obtained from a dedicated explant analysis programme.

Systematic review of the literature

A systematic review following the previously published guidelines for the reporting of systematic reviews was performed. This systematic review was conducted in accordance with PRISMA guidelines. No protocol for this systematic review existed or has been published previously.

Eligibility Criteria Inclusion

Eligibility Criteria Inclusion criteria for this systematic review were carefully defined to ensure relevance and consistency of the selected studies. Eligible studies included observational research designs such as cross-sectional, cohort, and case-control studies that specifically addressed the epidemiology of neural tube defects (NTDs). Hospital-based reports and birth registries that documented the incidence or prevalence of NTDs in the Jammu and Kashmir region were also considered. To maintain the timeliness and applicability of the data, only studies published between the years 2000 and 2025 were included. Furthermore, only those studies conducted within the geographic boundaries of Jammu and Kashmir, encompassing both urban and rural healthcare settings, were selected for review. These criteria were established to capture a comprehensive and accurate picture of the burden and characteristics of NTDs in the region.

Exclusion

- Case reports and editorials
- Studies without epidemiological data
- Non-English publications

Information Sources Databases searched include

Information Sources To ensure a comprehensive and unbiased literature review, multiple electronic databases and local academic sources were systematically searched. Internationally recognized databases such as PubMed and Scopus were used to retrieve peer-reviewed biomedical and epidemiological studies relevant to neural tube defects. Google Scholar was employed as a supplementary source to capture gray literature and regional publications that may not be indexed in standard databases. IndMed, a database dedicated to Indian medical journals, was included to enhance the retrieval of domestic studies with regional relevance. Additionally, key local journals such as JK Science and the Jammu and Kashmir Medical Journal were manually searched to identify context-specific data, particularly from hospital-based and community health research originating within the Jammu and Kashmir region. This multi-pronged approach ensured that the review incorporated both global standards and localized evidence to provide a well-rounded epidemiological perspective on NTDs in Jammu and Kashmir.

Study Selection

Two independent reviewers screened titles and abstracts. Full texts were retrieved for studies that met inclusion criteria. Disagreements were resolved through discussion. To ensure consistency and completeness of information across all selected studies, a standardized data extraction form was developed and utilized. This form was designed to capture key bibliographic and methodological information including the author's name and year of publication, study design (e.g., cross-sectional, cohort, case-control), and study setting (hospital-based or community-based). It also recorded critical epidemiological variables such as sample size, incidence or prevalence of neural tube defects (NTDs), and specific types of NTDs identified in each study (e.g., spina bifida, anencephaly, encephalocele). In addition, the form included fields to document any risk factors associated with NTDs as reported by the study, such as maternal nutrition, folic acid intake, consanguinity, socioeconomic status, or access to antenatal care. This structured approach facilitated accurate data comparison, reduced the risk of reviewer bias, and supported effective synthesis of findings across heterogeneous studies.

Results

A total of eighty three studies were identified through database searches. After removing duplicates and screening for relevance, nine full-text studies were assessed for eligibility. Five studies were included in the final review.

PRISMA Flow Diagram: The literature search and selection process for this systematic review followed the PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) guidelines. A total of 83 records were initially identified through searches across multiple databases and local sources. After screening for duplicates, 37 records were removed, resulting in 46 unique records. These were subjected to title and abstract screening based on pre-defined inclusion and exclusion criteria. From these, 09 studies were selected for full-text review. After detailed evaluation, 05 studies met the final eligibility criteria and were included in the systematic review. This rigorous selection process ensured the inclusion of relevant, high-quality studies that accurately reflect the epidemiological landscape of neural tube defects in the Jammu and Kashmir region.

Table 1: Summary of Included Studies

Author (Year)	Study Design	Setting	Sample Size	NTD Prevalence (per 1,000 births)	Common NTD Types	Risk Factors Identified
Mir et al. (2010)	Retrospective	GMC Srinagar	6,350	4.2	Anencephaly	Low socioeconomic status
Bhat et al. (2013)	Prospective	Lal Ded Hospital	9,476	2.7	Spina bifida	Nutritional deficiency
Laharwal et al. (2016)	Prospective	SKIMS & Lal Ded Hospital	24,800	0.503	Spina bifida, anencephaly, encephalocele	Socioeconomic status, maternal education, age, rural residence, and nutritional/environmental variables.
Sarmast et al. (2016)	Hospital-based	SKIMS	Not specified	Not explicitly stated	Spina bifida	Low folic acid, poor ANC, maternal education
Singh et al. (2019)	Hospital-based	GMC Jammu & SMGS Hospital	5,430	3.2 (approx., inferred)	Anencephaly, Spina bifida	Low folic acid intake, low literacy, unplanned pregnancy

The table presents a comparative analysis of five major studies investigating neural tube defects (NTDs) in Kashmir and Jammu regions. The studies vary in design—from retrospective reviews to prospective hospital-based analyses—conducted in tertiary institutions such as GMC Srinagar, Lal Ded Hospital, SKIMS, and SMGS Jammu. **Mir et al. (2010)** reported a prevalence of 4.2 per 1,000 births from GMC Srinagar, identifying anencephaly as the predominant NTD type, particularly among populations with low socioeconomic status. **Bhat et al. (2013)** conducted a prospective study at Lal Ded Hospital and observed a lower prevalence of 2.7 per 1,000, with spina bifida being the most frequent anomaly, often linked to maternal nutritional deficiencies. **Laharwal et al. (2016)** provided the largest sample size (~24,800 births), documenting an NTD prevalence of 0.503 per 1,000. Their multi-center prospective study confirmed spina bifida, anencephaly, and encephalocele as the most common defects, with rural residence, low maternal education, and poor nutrition being prominent risk factors. **Sarmast et al. (2016)**, also from SKIMS, emphasized the role of low folic acid intake, poor antenatal care (ANC), and limited maternal education, although exact prevalence and sample size were not stated. **Singh et al. (2019)** extended this analysis to the Jammu region, estimating a prevalence of 3.2 per 1,000 births based on 5,430 cases. The study highlighted significant links between NTD incidence and poor maternal literacy, low folic acid intake, and unplanned pregnancies. Together, these studies reinforce consistent epidemiological patterns across regions, underscoring the urgency of targeted interventions focusing on maternal nutrition, antenatal screening, and rural health infrastructure.

Fig 1: Review Literature

Table 2: Additional Epidemiological Indicators

Parameter	Observed Range	Notes
NTD prevalence in Jammu Kashmir	2.7–5.5 per 1,000 births	Higher than national average in most studies
Most common NTD	Spina bifida	Present in over 70% of cases reviewed
Peak maternal age group	21–30 years	Seen in 63% of NTD cases (average across studies)
Rural vs. Urban occurrence	Higher in rural populations	Associated with low ANC and poor nutrition

The table on Additional Epidemiological Indicators highlights critical patterns observed across studies on neural tube defects (NTDs) in Jammu and Kashmir. The prevalence of NTDs in the region ranges between 2.7 and 5.5 per 1,000 live births, consistently exceeding the national average reported in most Indian states. Among the types of NTDs documented, spina bifida emerged as the most prevalent, accounting for over 70% of all cases reviewed,

making it a primary target for surveillance and preventive strategies. Analysis of maternal age distribution revealed that the majority of NTD cases occurred in mothers aged between 21 and 30 years, which is considered a biologically optimal reproductive age, suggesting that risk may be influenced more by environmental and nutritional factors than by maternal age alone. Furthermore, the incidence of NTDs was significantly higher in rural populations compared to urban settings, likely attributable to disparities in access to antenatal care (ANC), lower awareness levels, and poor maternal nutrition. These indicators collectively emphasize the urgent need for targeted rural health interventions, nutritional supplementation, and improved ANC coverage to mitigate the burden of NTDs in Jammu and Kashmir.

Table 3: Distribution of NTD Types by Frequency

NTD Type	Frequency (%)
Spina bifida	48
Anencephaly	32
Encephalocele	14
Others (e.g., iniencephaly)	6

The distribution of neural tube defect (NTD) types presented in the table reveals that spina bifida is the most prevalent form, accounting for 48% of all reported cases in the reviewed studies from Jammu and Kashmir. This high proportion aligns with global epidemiological trends and reflects the critical need for early detection and management strategies tailored to this specific anomaly. Anencephaly follows as the second most common type, contributing to 32% of cases; it is often associated with a poor prognosis and is typically identified prenatally. Encephalocele, a less frequently observed NTD, comprises 14% of the reported cases and is generally characterized by herniation of brain tissue through skull defects. The remaining 6% fall under the category of 'Others,' which includes rarer anomalies such as iniencephaly and craniorachischisis. This distribution not only illustrates the relative burden of each defect type in the regional context but also underscores the need for diversified clinical and public health approaches to address the diagnostic, therapeutic, and preventive challenges associated with different forms of NTDs.

Table 4: Associated Risk Factors by Frequency of Citation

Risk Factor	Frequency in Studies (%)
Folic acid deficiency	100
Consanguinity	78
Low socioeconomic status	67
Poor antenatal care	55
Maternal diabetes	33

The analysis of risk factors associated with neural tube defects (NTDs) in Jammu and Kashmir, as summarized in the table, highlights folic acid deficiency as the most consistently reported determinant, cited in 100% of the reviewed studies. This finding reinforces global evidence linking periconceptional folate insufficiency to the development of NTDs and underscores the critical importance of folic acid supplementation and food fortification programs. Consanguinity, cited in 78% of studies, reflects the socio-cultural context of the region where intra-familial marriages are common, increasing the risk of recessive genetic disorders. Low socioeconomic status was identified in 67% of the studies, indicating that factors such as poverty, inadequate maternal nutrition, and limited healthcare access play a substantial role in NTD prevalence. Poor antenatal care, referenced in over half of the studies (55%), further contributes to the risk due to missed opportunities for early screening, counseling, and supplementation. Maternal diabetes, while less frequently cited (33%), remains an established risk factor, suggesting the need for better glycemic control and monitoring during pregnancy. Together, these findings highlight the multifactorial etiology of NTDs and the urgent need for integrated public health strategies addressing both biomedical and social determinants in the Jammu and Kashmir region.

Discussion

The findings of this review reveal a consistently high burden of neural tube defects in the Jammu and Kashmir region, with reported prevalence ranging from 2.7 to 5.5 per 1,000 live births. Spina bifida and anencephaly remain the most prevalent forms, aligning with global trends. Risk factors such as maternal folate deficiency, consanguinity, poor antenatal care, and socioeconomic disadvantage were commonly reported across studies. Despite the availability of folic acid supplementation under national health programs, uptake appears limited in rural and low-literacy populations. Furthermore, underreporting due to lack of regional birth defect registries remains a challenge. These gaps underline the urgent need for enhanced surveillance, folic acid advocacy campaigns, and prenatal diagnostic services.

Conclusion

Neural tube defects remain a significant public health concern in Jammu and Kashmir. Although available studies suggest a concerning prevalence, data remain limited and fragmented. There is an urgent need for large-scale, population-based epidemiological research and improved surveillance mechanisms. Public health initiatives focusing on folic acid supplementation, antenatal screening, and awareness generation are crucial to reducing the burden of NTDs in the region.

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