Risk Factors and Prevalence of Spina Bifida in Children Reported at Children Hospital (Lahore): A Mix Method Analysis

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Abstract

Pakistan is the country with more than 100 million children and spina bifida is a neural tube defect that commonly affects children. Folic acid deficiency during the initial three weeks of pregnancy has been identified as a significant causative factor. However, other factors and their effects vary based on geographic location, maternal pregnancy history, child sex, and race. This cross-sectional study aimed to investigate the factors contributing to the incidence rate of spina bifida in Lahore, Pakistan. A total of 200 cases of neural tube defects, including spina bifida, were studied and reported from October 2012 to August 2013 at Children's Hospital Lahore. Various factors were examined, such as the age of mothers, with 50% of babies with neural tube defects born to mothers aged between 26 and 36 years. The incidence of spina bifida was found to be higher in boys (59%) compared to girls (41%). Hydrocephalus, a common complication of spina bifida, was more prevalent in boys (51%) than in girls (14%). Spina bifida cystica, another form of the condition, occurred more frequently in girls (38%) than in boys (33%). In this study group, 161 out of 200 women did not use or properly use folic acid during pregnancy, indicating a lack of awareness regarding its importance. The study also revealed that 119 cases of spina bifida occurred during winter, suggesting a seasonal influence on the condition. In addition to other identified risk factors, this study underscores the critical need for raising awareness about the use of folic acid before conception and during the initial three weeks of pregnancy.

Keywords: Neural Tube Defects (NTD), Spina Bifida, Hydrocephalous, Meningomyelocele.

Introduction

Spina bifida is a complex congenital condition characterized by the incomplete closure of the spinal column during early fetal development (Fathi et al., 2023). It is a neural tube defect that can lead to significant physical and neurological disabilities in affected children (Berry, 2023). Understanding the risk factors associated with spina bifida and its prevalence is crucial for effective prevention, early detection, and appropriate management strategies.

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This introduction provides an overview of the risk factors and prevalence of spina bifida in children based on available information up until September 2021, as the model's knowledge cutoff date. Several research studies and publications have contributed to our understanding of these factors during this period (Sun et al., 2023). Numerous risk factors have been identified as potential contributors to the occurrence of spina bifida. One of the most significant risk factors is maternal folic acid deficiency during pregnancy. Adequate folic acid intake before conception and during early pregnancy has been shown to reduce the risk of spina bifida and other neural tube defects. Other risk factors include maternal obesity, diabetes, certain medications, exposure to environmental factors (such as pesticides and solvents), and genetic predisposition. The prevalence of spina bifida varies across populations and geographical regions. Global estimates suggest that the prevalence ranges from approximately 0.5 to 5 cases per 1,000 live births (Blencowe et al., 2018). However, prevalence rates can vary significantly based on factors such as ethnicity, socioeconomic status, and access to prenatal care. This report aims to provide a comprehensive understanding of the risk factors and prevalence of spina bifida in children. It will review relevant research studies and publications conducted up until September 2021 to identify key risk factors associated with the condition. Additionally, the report will present available data on the prevalence of spina bifida in different populations, highlighting any notable trends or variations.

By examining the risk factors and prevalence of spina bifida in children, we can enhance awareness, promote preventive measures, facilitate early intervention, and improve the overall management and outcomes for affected individuals. Ongoing research and monitoring are essential to further our understanding of this complex condition and develop effective strategies for its prevention and treatment.

Objectives of the Study

- To identify and analyze the risk factors associated with Spina Bifida in children.
- To estimate the prevalence of Spina Bifida in children reported at Children Hospital, Lahore.
- To provide a comprehensive understanding through a mixed-methods approach.

Literature Review

Spina bifida is a neural tube defect that commonly affects children in the Pakistani population. Folic acid deficiency during the initial three weeks of pregnancy has been identified as a significant causative factor (Tsegai, 2023). However, other factors and their effects vary based on geographic location, maternal pregnancy history, child sex, and race (Payne-Sturges et al., 2023). This cross-sectional study aimed to investigate the factors contributing to the incidence rate of spina bifida in Lahore, Pakistan.

Spina bifida is a severe congenital disorder due to neural tube formation defects. Spina bifida is one of the most common congenital disorders, with an average incidence of 1 in every 1000 births (Cragan et al., 1995). Spina bifida is a series of malfunctions of neural tube formation and human structure. This defect is more common in girls than boys by birth (Daltveit, 2023) This condition presents both in developed and developing countries as well as urban and rural areas (Aruleba & Jere, 2022).

All congenital anomalies about neural tube closure which occur during the first three weeks of embryogenesis include neural tube defects, spina bifida, and anencephaly are severe forms of neural tube defects (Ten et al.,2022). Spina bifida falls into three types due to its severity conditions: Spina bifida occulta, spina bifida cystica and spina bifida myeloschisis. The Spina bifida or malformations can occur at any point but mostly occur in lumbar and sacral areas (Moore

and Persaud, 1993). Meningomyelocele is more common than meningocele. Central nervous system disturbance can be death-leading, such as meningomyelocele (spinal cord area exposure) and anencephaly (brain absent), which a lethal defects and death-leading (Campbell et al., 1996). Spina bifida is a multifactorial disease in which these factors mostly related to spina bifida like maternal age, social class, maternal health, maternal nutrition, maternal obesity, education of mother and fathers, pregnancy histories, folic acid consumption, vitamin B12, drug (anticonvulsant, valproic acid) usage, varies in Ethnic group, altered sex ratio, genetic syndromes, geographical location and parents occupation (De Marco et al., 2011). It is worldwide accepted that the risk of spina bifida and anencephaly is strongly associated with low or inadequate uptake of folic acid (Wald, 1993). Spina bifida is worldwide one of the most common congenital disabilities. The average incidence rate is one to two cases per 1000 births (Parvin & Hasan, 2023). The rate of spina bifida is high risk in some populations like southern Wales (7.6 per 1000) and Northern Ireland (8.6 per 1000) (Hail et al., 1988). According to one study of NTDs in Pakistan, the incidence rate of NTDs in our setup was 13.90 per 1000 deliveries (Khattak et al., 2008). In the UK, USA and Denmark, it is around 1-5 per 1000 deliveries. Lowry (2019) "Prevalence rates of spina bifida in Alberta, Canada: 2001–2015. Can we achieve more prevention?" *Birth Defects Research* 111.3 (2019): The prevalence of neural tube defects in Swat and Peshawar, Pakistan, is 3.2-13.9 per 1000 deliveries (Khattak et al., 2010; Khattak et al., 2008). So, study these risk factors to evaluate the relation with spina bifida in Lahore, Pakistan. Because Lahore is a populated area of Pakistan, such type of study was not conducted here.

Research Scenario 1: Genetic Predispositions and Prevalence Trends
Background
This scenario focuses on investigating the role of genetic factors in the occurrence of Spina Bifida among children in Lahore. It also aims to analyze the prevalence trends over a specific period.

Methodology
Quantitative Strand
- *Data collection*: Retrospective analysis of medical records of children diagnosed with Spina Bifida over the past decade.
- *Variables of interest:* Family history of neural tube defects, presence of genetic mutations.
- *Data analysis*: Logistic regression to identify significant genetic risk factors. Prevalence rates will also be calculated.

Qualitative Strand
- *Data collection:* In-depth interviews with genetic counsellors at Children's Hospital, Lahore, to gain insights into familial patterns and genetic counselling practices.
- *Data analysis:* Thematic analysis to extract patterns and recommendations related to genetic factors.

Expected Findings
This study anticipates finding a higher prevalence of Spina Bifida among children with a family history of neural tube defects. Additionally, insights from genetic counsellors will provide valuable recommendations for genetic counselling practices.

Research Scenario 2: Socioeconomic Factors and Access to Care
Background
This scenario aims to explore how socioeconomic factors influence the prevalence and management of Spina Bifida in children at Children's Hospital, Lahore.

Methodology
Quantitative Strand
- *Data collection:* Surveys administered to parents/caregivers of children with Spina Bifida, assessing socioeconomic status, access to healthcare, and utilization of available resources.
- *Data analysis:* Correlation analysis between socioeconomic indicators and clinical outcomes.

Qualitative Strand
- *Data collection:* Semi-structured interviews with families from diverse socioeconomic backgrounds, focusing on experiences in accessing healthcare services.
- *Data analysis*: Thematic analysis to identify barriers and facilitators.

Expected Findings
This study expects to find that families with lower socioeconomic status face challenges in accessing specialized care and resources. Understanding these barriers will be crucial for designing interventions to improve healthcare accessibility.

Research Scenario 3: Maternal Age and Prenatal Care Practices
Background
This scenario investigates the influence of maternal age on the occurrence of Spina Bifida and the prenatal care practices of mothers in Lahore.

Methodology
Quantitative Strand
- *Data collection:* Analysis of birth records and medical histories to categorize cases based on maternal age.
- *Variables of interest:* Maternal age, prenatal care utilization, incidence of Spina Bifida.
- *Data analysis:* Comparative analysis of Spina Bifida incidence among different age groups. Assessment of prenatal care practices.

Qualitative Strand
- *Data collection:* Focus group discussions with mothers from various age groups to explore their experiences with prenatal care.
- *Data analysis:* Thematic analysis to identify common experiences and perceptions.

Methodology
In this mixed-method research, a comprehensive questionnaire was developed to investigate the prevalence and risk factors associated with Spina Bifida within the Pakistani population. The questionnaire encompassed variables related to Spina Bifida, including social class, pregnancy history, family history, maternal health, and dietary or medication intake. Data was gathered from mothers of children affected by Spina Bifida, who were either attending routine check-ups or had their children hospitalized for Spina Bifida-related surgeries at Children Hospital, Lahore. This

study was conducted over ten months, spanning from October 2021 to August 2022. Data collection was performed through face-to-face interviews. Subsequently, the gathered data was subjected to analysis using SPSS 19.0, enabling a rigorous examination of the identified factors within a mixed-method research framework.

Results

Gender	Frequency	Percent
Boy	118	59.0
Girl	82	41.0
Total	200	100.0

Table 1: Frequency of neural tube defects (NTDs) in girls and boys

200 cases of neural tube defects were studied. The mothers aged from 26 to 36 years had 50% of babies with neural tube effects. Both genders are affected by neural tube defects, but boys (59%) are more affected as compared to girls (41%), as shown in table 1.

The difference in severity of defects was observed in both genders. Like girls have high percentage of spina bifida while boys have high percentage of hydrocephalous. Winter season has strong effect on 119 patients (59%) for spina bifida occurrence. Ratio of different neural tube defects frequency was different in girls and boys. Hydrocephalous occurrence was more in boys 51% as compared to girls 14%. While spina bifida cystica occurrence frequency was 38% in girl as compare to 33% in boys. Spina bifida cystica with hydrocephalous cases were present in 24(12%) patients while microcephaly cases were 7 (3.5%) less than others neural tube defects.

Figure 1: Percentage of different NTDs in girls and boys
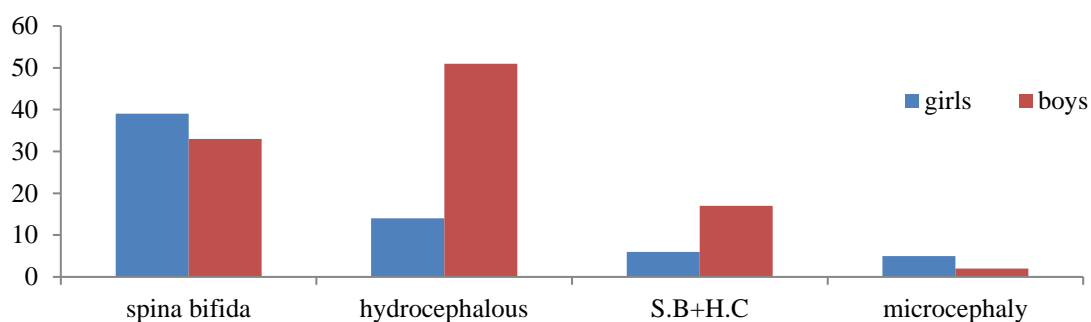
No relation between folic acid and canned food intake and spina bifida occurrence was observed (Martinez et al, 2021). Strong relation between preconception folic acid and neural tube defects is present. 134 mothers (67%) do not use folic acid preconception and during pregnancy while 27 women do not used properly folic acid. Only 39 mothers used folic acid properly.

Table 2: Frequency of mothers taking folic acid during pregnancy

Folic acid intake during pregnancy	Frequency	Percentage
Yes	39	19.5

No	134	67.0
Sometimes	27	13.5
Total	200	200

According to study as illiteracy rate of parents and poverty increases the rate of spina bifida

Table 3: Frequency of Spina Bifida in rural and urban areas

Resident Area	Frequency	Percent
Rural	113	56.5
Urban	87	43.5
Total	200	100.0

increases. Both in urban and rural areas spin bifida cases were present. But more spina bifida

	Previous NTD pregnancy history	Valproic acid intake	Vitamin A intake	Improper diet	Canned food
11 (5.5%)	3 (1.5%)	7 (3.5%)	33 (16.5%)	12 (6%)	8 (4%)

cystica cases were residents of rural cases as shown in table 3.

Table 4: Relationship of other factors with occurrence of Spina Bifida

Maternal health during pregnancy has varied percentage for affecting neural tube defects. Blood pressure during issue is common as shown in table 5.

Table 5: Relationship of maternal health with occurrence of Neural Tube Defects

Disease	Good	B.P	Diabetes	Gestational diabetes	T.B	Other diseases	Total
Frequency	111	75	3	4	2	4	200
Percentage	55.5	37.5	1.5	2.0	1.0	2.0	100.0

Discussion and Conclusion

The incident rate of neural tube defects, especially spina bifida, is increasing in Pakistan due to many factors. The main important factor is the use of folic acid during preconception and pregnancy. According to study results, 67% of females affected with spina bifida do not use folic acid during pregnancy, and 13.5% of females do not use it properly. Poverty and illiteracy rates are also important factors causing the spina bifida rate to be high. 70(35%) mothers were uneducated, only 11 mothers graduated, and only 5 mothers had master's degrees. 27% of parents have jobs while a high percentage of parents have daily income-based jobs, due to which they do not use proper diet. The majority of females go hospital for checkups during pregnancy but do not use medicine properly. And 44% of females are not concerned with doctors. During face-to-face conversations, most females do not know the names of the medicines which they use during pregnancy. 88% of females do not know they are carrying a spina bifida-affected child during pregnancy. Pakistan has agricultural land so canned food relation with spina bifida is not strong. 56.5% of parents of affected children come from rural areas where medical facilities are not good.

Due to unawareness spina bifida rate is also high in both rural and urban areas. At the same time, other factors like maternal health during pregnancy, drug intake, genetics, and previous NTD pregnancy history have varied moderate effects. Awareness about folic acid use before baby conception and during pregnancy, especially the first 3 weeks after conception, is very important. As spina bifida is becoming common in Pakistan therefore, medical facilities need to be improved (Kancherla et al., 2022).

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