EAJNS East African Journal of Neurological Sciences
Official journal of East African Association of Neurological Surgeons

Original Article EAJNS 5(2): 100-106

Anatomical characteristics, clinical features, and risk factors of spina bifida: A retrospective review of children at Cure Hospital, Uganda.

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Received: 07-08-2025; Revised: 02-10-2025; Accepted: 18-10-2025
DOI: https://dx.doi.org/10.4314/eajns.v5i2.6

ABSTRACT

Background: Spina bifida, a neural tube defect, continues to pose substantial public health challenges in Uganda despite multiple preventive efforts. An estimated 1,400 children are affected annually. We conducted a retrospective cross-sectional review of hospital records for children diagnosed with spina bifida between 2020 and 2023. **Methods:** Using systematic sampling , we selected 414 patient records. We analysed the data using SPSS version 20. We defined severe spina bifida as a lumbosacral lesion presenting clinically with hydrocephalus. **Results**: Males constituted 60.4% of cases (n=250). Infants aged <1 year comprised 36.2% (n=150). Most lesions (69.0%) were located in the lumbosacral region; sacral lesions accounted for 17.4%, lumbar 8.0%, lumbothoracic 4.6%, and occipital and cervical regions 0.5% each. Regarding lesion integrity, the highest proportion of intact lesions occurred in the lumbosacral region (48.1%), followed by sacral (16.2%), lumbar (15.2%), and lumbothoracic (12.4%); ruptured lesions were also observed in the thoracic region (7.8%). Maternal febrile illness was associated with severe spina bifida (adjusted OR 2.24, 95% CI 1.34–3.74; p=0.02). Tertiary maternal education was also associated with higher odds of spina bifida (adjusted OR 2.27, 95% CI 1.43–4.57; p=0.01). **Conclusion:** Myelomeningocele remains the predominant form of spina bifida in Uganda, chiefly affecting the lumbosacral region.

Keywords: Spina Bifida; Myelomeningocele; Lumbar-sacral hydrocephalus.

INTRODUCTION

Spina bifida (SB) is a neural tube defect (NTD) resulting from failure of neural tube closure within the first 25 days of pregnancy (1). It is considered a developmental disorder arising from the interplay of genetic and environmental factors (2). Established risk factors cluster around genetic predisposition and dietary insufficiency. Neurological manifestations commonly include pain, motor and sensory deficits, gait disturbance, and bowel and bladder dysfunction. Orthopaedic features such as limb-length discrepancy, unequal foot size, equinovarus/equinovalgus deformities, and clawed toes, are also suggestive of an underlying NTD.

Globally, the prevalence of SB remains high despite advances in technology and the implementation of mandatory food-fortification policies (3). The global incidence of spina bifida stands at 300,000 annually (4). Recent African

data indicate marked heterogeneity across countries, with reported prevalence as high as 0.43% in Algeria and as low as 0.06% and 0.09% in Libya and Tunisia, respectively (5).

In Uganda, an estimated 1,400 children are born with SB annually. Incidence and prevalence may be elevated due to inconsistent folate intake among pregnant women, limited access to antenatal care, absence of effective secondary prevention, greater exposure to environmental risk factors, and high birth rates (1). Against this background, the objective of the present study was to describe the anatomical distribution and clinical features of SB in children presenting to CURE Children's Hospital of Uganda (2020–2023) and to identify factors independently associated with clinical severity.

METHODOLOGY

Study Design and setting

This was a retrospective records review conducted at CURE Children's Hospital of Uganda.

Ethics

Ethical approval was obtained from the Makerere University School of Biomedical Sciences Research Ethics Committee (SBS-2024-529). A consent waiver was granted by the Higher Degrees Research and Ethics Committee. Administrative clearance was provided by CURE Hospital prior to data collection.

Study Population and selection criteria

The study included medical records for all children diagnosed with spina bifida who received care at CURE Children's Hospital of Uganda between 2020 and 2023. Records covered both patients who underwent surgery and those managed non-operatively. Inclusion required: a confirmed diagnosis of spina bifida; comprehensive past medical history for the child and caregiver; detailed physical examination findings; complete demographic details (age, sex, residence); and adequate clinical documentation, including pre-operative and post-operative notes. Using systematic sampling, 414 eligible records were selected to ensure even representation across the four-year period (Figure 1).

Sample Size Determination

The sample size (anatomical and clinical features) was determined using the Kish–Leslie (1965) formula for a single proportion, with Z=1.96 for a 95% confidence level, an estimated prevalence of 5.7% reported in Kampala (6), and a desired margin of error of 5%. A 10% allowance for missing data yielded a final target sample of 414 records.

Sample Size Determination (severe spina bifida)

For analyses of severe spina bifida, the required sample size per group was estimated using a two-proportion comparison with $Z\alpha/2=1.96$ (95% confidence) and $Z\beta=0.84$ (80% power). The assumed exposure proportion among individuals with severe spina bifida was 53.8% based on African data (5), and 25% among those without severe spina bifida from a case–control study in north-eastern Ethiopia (2). This yielded $N\approx41N$ per group.

Sampling technique

Systematic sampling was applied to obtain a representative set of records. Approximately 100 records were selected from each calendar year (2020–2023). Within each year, files were ordered and grouped, and every second file was chosen.

Data collection procedure.

A comprehensive review of medical records was performed using a standardised data-extraction tool adapted from the CDC (7). Variables captured included demographic characteristics, anatomical site of the lesion, clinical features, and factors hypothesised to be associated with clinical severity.

Data management and analysis.

Two trained research assistants independently reviewed each file and extracted data, resolving discrepancies by consensus. Four team members entered the data, which were checked daily for completeness and accuracy; identified errors were corrected before finalisation. Data were entered and cleaned in Microsoft Excel (version 2019), validated for inconsistencies, and exported to SPSS (version 20) for analysis. Descriptive and inferential statistics were conducted to characterise the cohort and evaluate associations of interest.

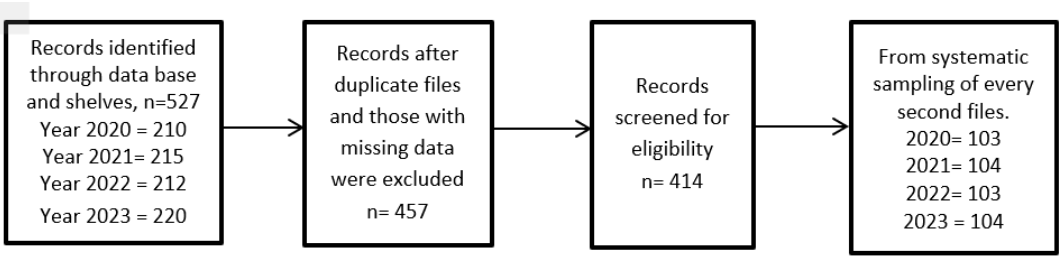

Figure 1: Flow chart showing the sampling strategy

RESULTS

General characteristics

A total of 414 records were reviewed. Males constituted 60.3% (n = 250). The age range was 5 days to 12 years. Birth order was most commonly first-born, followed by second- and third-born, respectively.

Regarding parental characteristics, most mothers had no formal education (reported as 60.3%, n = 210), with 23.9% (n = 83) having primary education, 11.2% (n = 33) secondary education, and 4.6% (n = 16) tertiary education. The majority of mothers were married (91.5%, n = 379); 5.1% (n = 21) were separated and 3.4% (n = 14) were single. Half of the mothers were housewives (50.0%, n = 207), and 5.3% were engaged in business. Among fathers, 72.5% (n = 300) were peasants, 15.9% (n = 67) were businessmen, and 5.8% (n = 24) were in other occupations (Table 1).

Types of spina bifida

Myelomeningocele was the most prevalent and severe type, followed by meningocele; lipomyelomeningocele and spina bifida occulta were less frequent (Figure 2).

Location and characteristics of the lesions

Most lesions were located in the lumbosacral region (69.0%), followed by the sacral (17.4%) and lumbar (8.0%) regions. Lumbothoracic lesions accounted for 4.6%, while occipital and cervical lesions were least common (0.5% each). With respect to lesion integrity, the lumbosacral region had the highest proportion of intact lesions (48.1%), followed by the sacral (16.2%), lumbar (15.2%), and lumbothoracic (12.4%) regions. Ruptured lesions were also observed in the thoracic region (7.8%) (Figure 3).

Associated clinical features included hydrocephalus (n = 105, 25.4%), truncal hypotonia (n = 76, 18.4%), neurogenic bladder (n = 68, 16.5%), clubfoot (n = 102, 24.6%), and seizures (n = 51, 12.3%) (Table 2).

Factors associated with severe spina bifida

On bivariate analysis, maternal tertiary education was associated with severe spina bifida (unadjusted OR 2.76, 95% CI 1.56–4.88; p < 0.001). Maternal febrile illness was also significant (unadjusted OR 2.40, 95% CI 1.50–3.05; p < 0.001) (Table 3). In multivariable analysis, maternal febrile illness remained independently associated with severe spina bifida (adjusted OR 2.24, 95% CI 1.34–3.74; p = 0.002). Maternal tertiary education was likewise associated with higher odds of severe spina bifida (adjusted OR 2.55, 95% CI 1.43–4.57; p = 0.001) (Table 3).

DISCUSSION

In this cohort, males were more frequently affected by spina bifida than females, with the highest burden among infants under one year and first-born children. Sex-related differences in early embryogenesis may partly explain this pattern, consistent with a Zambian review of 253 cases (55% male; 46% <1 year) and a Nigerian series that also noted an excess of first-born children (8, 9). These observations suggest that birth order and early infancy may mark periods of heightened vulnerability.

Most mothers were married, had no formal education, and were housewives, reflecting lower socio-economic status and potentially constrained access to nutrition and healthcare. These findings align with prior work reporting high unemployment,

low educational attainment, and a predominance of married status among mothers of children with spina bifida (10). Socio-economic disadvantage may reduce folate intake and healthcare access, both of which are recognised risk factors for neural tube defects.

Table 1: Child demographics, maternal and paternal characteristics of spina bifida children admitted at cure childrens hospital (n=414)

Category	sub-group	percentage %
Age group	< 1 year	150 (36.3)
	1-5 year	120 (29.1
	5 year	144 (34.6)
Birth order	1st	120 (28.9)
	2nd	90 (21.7)
	3rd	75 (18.1)
	4th	30 (7.3)
	5th	25 (6.0)
	6th & above	16 (3.9)
	Unknown	58 (14.0)
Sex	Male	250 (60.5)
	Female	164 (39.5)
Representation by region	Eastern	175 (42.5)
	Central	79 (19.1)
	Western	66 (15.9)
	Northern	48 (11.6)
	Unknown	22 (5.3)
	Non- Ugandans	18 (4.3)
	West- Nile	6 (1.5)
Educational level of mothers	Non- formal	210 (50.7)
	Primary	83 (20.1)
	Secondary	39 (9.4)
	University	16 (3.9)
	Unknown	66 (15.9)
Marital status of mothers	Married	379 (91.5)
	Separated	21 (5.1)
	Single	14 (3.4)
Occupation of mothers	house wife	207 (50)
	Peasant farming	141 (34.1)
	Vocational jobs	17 (4.1)
	Business	22 (5.3)
	Professional jobs	15 (3.6)
	Student	12 (2.9)
Occupation of fathers	business	66 (15.9)
	Peasant farming	300 (72.5)
	Professional jobs	24 (5.8)
	Unknown	24 (5.8)

Table 2: Clinical presentation of spina bifida children at Cure Children's Hospital

Clinical feature	Count (%)
Hydrocephalus	105 (25.4)
Trunk hypotonia	76 (18.4)
Neurogenic bladder	68 (16.4)
Clubfoot	102 (24.6)
Seizures	51 (12.3)

Figure 2. Distribution of Spina Bifida Types among Children admitted at Cure Children's Hospital.

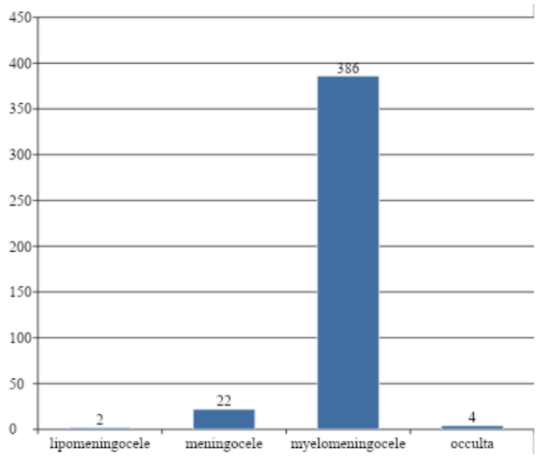

Figure 3. Description of spina bifida lesions by type and location

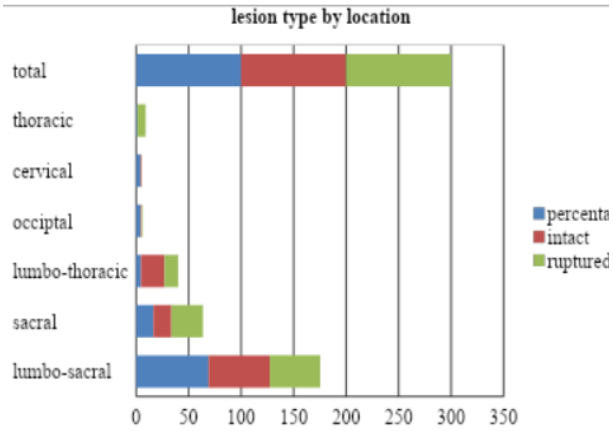

Table 3: Factors associated with severe Spina Bifida

Variable	Bivariate analysis			Multivariate analysis		
	cOR	95%-CI	P-value	aOR	95%CI	P-value
Sex						
Female	-	-	-	-	-	-
Male	1.04	0.74-1.46	0.802	-	-	-
Child age						
< 1 year	-	-	-	-	-	-
1-5 years	0.9	0.33-2.48	0.846	-	-	-
>5 years	1.96	0.72-5.30	0.185	-	-	-
Marital status						
Married	-	-	-	-	-	-
Separated	1.16	0.41-3.04	0.762	-	-	-
Single	0.77	0.22-2.70	0.688	-	-	-
Education level						
No formal	-	-	-	-	-	-
Primary	0.59	0.30-1.18	0.141	0.61	0.31-1.19	0.152
Secondary	1.27	0.67-2.42	0.458	1.45	0.75-2.80	0.75
Tertiary	2.76	1.56-4.88	**<0.001**	2.55	1.43-4.57	**<0.001**
Febrile illness in pregnancy						
Yes	2.4	1.50-3.05	**<0.001**	2.24	1.34-7.4	**0.002**
No	-	-	-			
Family history of spina bifida						
Yes	0.97	0.29-3.27	0.965			
No	-	-	-			
ANC attendance						
Yes	0.72	0.49-1.06	0.098			
No	-	-	-			
Folate consumption						
Yes	1.13	0.34-0.65	0.532	-	-	-
No	-	-	-	-	-	-

cOR- crude odds Ratio aOR-adjusted Odds Ratio

Myelomeningocele predominated (93.2%), in keeping with the pathophysiology of failed neurulation and with prior regional reports (3). Lesions were mainly lumbosacral (69%), followed by sacral and lumbar levels; this distribution mirrors published series of open spina bifida in which lower spinal levels are most commonly involved (12). The variability in the site of the lesion could be explained by the staggered closure points and rostrocaudal progression of primary and secondary neurulation (13).

Lesion integrity and associated clinical features

Intact lesions (72.2%) were more frequent than ruptured/ulcerated lesions (27.7%), contrasting with a Cameroonian series that reported more ruptured lesions (11). The determinants of lesion integrity remain poorly characterised and warrant further study; traumatic delivery and secondary infection may contribute to rupture.

Hydrocephalus occurred in 25.4% of cases, lower than rates reported in some surgical series (e.g., 49% in Kenya) (14). The association of hydrocephalus with myelomeningocele is well described, plausibly mediated by impaired CSF dynamics due to hindbrain and spinal malformations (15). Clubfoot was observed in 24.6%, lower than proportions reported in Austria and Pakistan (16, 17); differences likely reflect case mix and neurological level, as clubfoot prevalence increases with higher (thoracic/lumbar) lesion levels (18). Neurogenic bladder was present in 16.5%, below estimates from US clinic populations (19), which may reflect under-diagnosis in routine records or differing assessment protocols. Truncal hypotonia was seen in 18.4%; although cervical lesions were rare in this cohort, thoracic and high lumbar involvement can also impair trunk control, consistent with gait and postural asymmetries reported in ambulant SB cohorts (20). Seizures (12.3%) were infrequent, in line with small series elsewhere (21); seizure aetiologies are multifactorial and not specific to spina bifida.

Factors associated with severity

Maternal febrile illness showed an independent association with severe spina bifida, corroborating evidence that hyperthermia, particularly in the first trimester, is linked to increased risk (22). Potential mechanisms include heat-induced perturbation of signalling pathways involved in neurulation. In this study, tertiary maternal education was also associated with higher odds of severe spina bifida. This counterintuitive finding contrasts with a Sudanese report and may reflect residual confounding (e.g., referral patterns, differential health-seeking, or documentation quality) or small subgroup numbers. It should therefore be interpreted cautiously and explored in prospective studies.

Strengths and limitations

Strengths include a relatively large, multi-year sample and systematic sampling across the study period, enabling description of anatomical patterns and clinical features. Limitations arise from the retrospective design and reliance on secondary data, with incomplete documentation in some records. Potential misclassification of clinical variables and unmeasured confounding (e.g., folate intake, timing of antenatal care, and socio-environmental exposures) may have influenced associations.

Conclusion: In this hospital-based retrospective series, myelomeningocele predominated and lumbosacral lesions were most frequent. Maternal febrile illness during pregnancy was independently associated with greater clinical severity. Prospective studies incorporating standardised severity metrics, detailed exposure assessment (including folate status and infection history), and robust confounder control are warranted.

Recommendations: The findings of this study highlight several measures to improve the prevention and early detection of spina bifida. Newborns, particularly first born infants, should undergo careful examination for spina bifida at birth and during the first year of life to enable early diagnosis and timely intervention. Targeted health education is also needed in regions with a higher recorded burden, such as the Eastern region, with emphasis on periconceptional folate supplementation during the critical period of neural tube formation (days 0 to 28), appropriate dietary supplementation, timely healthcare seeking, and awareness of possible genetic and environmental risk factors. Antenatal services should incorporate routine screening and prompt management of febrile illnesses during pregnancy. In addition, women with higher educational attainment should not be presumed to be at lower risk on the basis of education alone; therefore, preconception counselling and folate guidance should be provided to all women of reproductive age.

Funding: No external funding was received.

Conflicts of interest: The authors declare no conflicts of interest.

Authors' contributions: Chebet Frida led conceptualisation, data collection, analysis, drafting, revision, and submission. Kiryowa Haruna, Catherine Lutalo Mwesigwa, and Ochieng Joseph provided academic and methodological oversight.

Data sharing: Data are available from the lead author (chebetfrida43@gmail.com) on reasonable request, subject to institutional approvals to ensure confidentiality and data-governance compliance.

Acknowledgements: The authors thank the administration of CURE Children's Hospital for access to records, and Dr KH (Lecturer, Department of Anatomy, Makerere University), CLM (Senior Lecturer, School of Dentistry, Makerere University), and Assoc. Prof OJ (Department of Anatomy, Makerere University) for their contributions to study development, revision, and manuscript preparation.

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