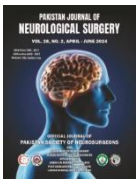

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Original Research

Comprehensive Analysis of Spinal Dysraphism: Early Institutional Experience at Pediatric Neurosurgery of Punjab Institute of Neurosciences

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ABSTRACT

Objective: To examine the demographic and clinical characteristics of pediatric patients presenting with spinal dysraphism.

Material and Methods: This retrospective study was conducted at the Department of Pediatric Neurosurgery, Punjab Institute of Neurosciences in Lahore and included data from patients aged <16 years with spinal dysraphism over 7 months from March 2023 to September 2023. Data analysis was performed using SPSS version 27.0.

Results: Among 32 patients, age distribution: <1 year (37.5%, mean age: 5.58±2.31 months), 1-4 years (25%, mean age: 3.25±0.6 years), 5-15 years (37.5%, mean age: 11±2.6 years). Gender: 22 males (68.75%), 10 females (31.25%). The most prevalent clinical presentations were Myelomeningocele (18.75%) and Meningocele (15.63%), both predominantly observed in patients under 1 year of age. Among the cutaneous findings, Fluid sacs (34.38%) and Tuft of Hair (15.63%) were the most common. In terms of other clinical findings, Lower Limb Weakness (50.00%) and Lower Limb Muscle Atrophy (21.88%) had the highest percentages.

Conclusion: The study revealed a diverse age distribution, with Myelomeningocele and Meningocele being common clinical presentations, especially in infants. Some of the physical signs of the condition included a fluid sac. a tuft of hair on the back, and weak and underdeveloped leg muscles.

Keywords: Spinal Dysraphism, Pediatric Patients, Clinical Presentations, Cutaneous Findings.

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INTRODUCTION

Spinal dysraphism encompasses a group of congenital disorders affecting the development of

the spinal column and spinal cord. These conditions stem from disturbances during early embryonic development, specifically, the incomplete closure of the caudal neural tube. This crucial event, occurring during the fourth week of gestation, is essential for proper spinal formation. Disruptions within this process lead to a range of malformations involving the ectoderm, mesoderm, and neuroectodermal tissues, resulting in a spectrum of disorders classified as spinal dysraphism.[1,2] There are two main categories of spinal dysraphism: exposed spinal dysraphism and occult spinal dysraphism. Exposed spinal dysraphism is a prime example of the intricate interaction between genetic and environmental factors during embryonic development. It frequently manifests as noticeable edema on the back at birth. The prevalence rate of neural tube defects, which includes spinal dysraphism, is higher than that of congenital cardiac disease, making them a significant category of birth abnormalities.[3-5]

Even though these disorders have grave consequences, there is still a dearth of thorough research on spinal dysraphism, particularly in particular institutional settings. Maternal nutrition, access to prenatal care, and geographic location are just a few examples of the variables that can significantly affect patient demographics and clinical presentations. The co-occurrence of spinal dysraphism with other congenital anomalies is common, indicating the intricate embryological origins and interrelated nature of these disorders.[6,7]

To fill in knowledge gaps about spinal dysraphism, we intend to perform a thorough examination of the clinical and demographic features of the pediatric patient population treated at the Punjab Institute of Neurosciences. A deeper understanding of this complicated illness is needed to improve our capacity to identify, treat, and deliver the best possible care. This is the driving force behind this research. We hope to provide insight into the range of presentations and possible variations related to spinal dysraphism by closely examining our institutional experience. This research has the potential to significantly improve patient outcomes and healthcare practices for individuals with this condition.

MATERIAL AND METHODS

Study Setting and Duration

This retrospective case series spanning 7 months (from March 2023 to September 2023) was conducted at the Department of Pediatric Neurosurgery, Punjab Institute of Neurosciences (PINS), Lahore after taking ethical approval along with the waiver of consent (vide letter no. 1713/IRB/PINS/Approval/2024, Dated 07/02/2024).

Sampling Technique

This study used a non-probability convenience sampling technique, including pediatric patients with spinal dysraphism who presented at the Punjab Institute of Neurosciences, Lahore, from March to September 2023.

Inclusion Criteria

Pediatric patients aged less than 16 years, diagnosed with spinal dysraphism, including myelomeningocele, spina bifida occulta, meningocele, and other forms of neural tube defects, patients whose complete medical records, including demographic data, clinical presentation, cutaneous manifestations, and other relevant clinical findings, were available.

Exclusion Criteria

Medical records with incomplete or missing data, patients who were diagnosed with conditions other than spinal dysraphism, and cases where the diagnosis was uncertain or not confirmed by clinical or imaging studies.

Data Collection Tool

The data of the patients i.e., name, age, gender, clinical presentation, cutaneous, and other clinical findings were noted. All data collected during this study was analyzed using SPSS version 27.0.

Data Analysis

Categorical variables i.e., gender, clinical presentation, cutaneous, and other clinical findings have been presented as frequency and percentages, while numerical variables i.e., age have been presented as mean and standard deviations. The chi-square test has been applied for the association of age groups/gender with clinical presentation, cutaneous, and other clinical findings. A p-value of ≤0.05 has been taken as significant.

RESULTS

Age Distribution

A total of 32 patients were included in the study. Among them, 12 patients (37.5%) were under 1 year of age with a mean age of 5.58±2.31 months, 8 patients (25%) fell within the 1-4 years age group with a mean age of 3.25±0.6 years, and the remaining 12 patients (37.5%) were aged between 5 and 15 years, having a mean age of 11±2.6 years.

Gender Distribution

There were 22 male patients (68.75%) and 10 female patients (31.25%), resulting in a male-to-female ratio of 2.2:1.

Clinical Presentation

Myelomeningocele was observed in 6 (18.75%) cases, meningocele in 5 (15.63%) cases, lipomeningocele in 7 (21.88%) cases, dermal sinus in 2 (6.25%) cases and split cord malformation in 5 (15.63%) cases (Table 1).

Table 1: Distribution of Clinical Presentation according to age groups.

Clinical Presentation	n	%age
Myelomengiocele	6	18.75%
Meningocele	5	15.63%
Lipmeningiocele	7	21.88%
Dermal Sinus	2	6.25%
Split cord malformation	5	15.63%
Thickenened Filum Terminale	7	21.88%
Total	32	100%

Cutaneous Findings

Fluid sac was observed in 11 (34.38%) cases, tuft of hair in 5 (15.63%) cases, subcutaneous mass in 3 (9.38%) cases, lumbosacral dimple in 3 (9.38%) cases, previous scar in 3 (9.38%) cases and dermal sinus in 2 (6.25%) cases (Table 2).

Table 2: Distribution of Cutaneous Findings.

Cutaneous Findings	n	%age
Fluid sac	11	34.38%
Tuft of Hair	5	15.63%
Subcutaneous Mass	3	9.38%
Lumbosacral dimple	3	9.38%
Previous scar	3	9.38%
Dermal Sinus	2	6.25%

Other Clinical Findings

Lower limb weakness was observed in 16 (50.00%) cases, scoliosis in 7 (21.88%) cases, lower limb muscle atrophy in 7 (21.88%) cases, talipes equinovarus in 6 (18.75%) cases, graded sensory loss in 5 (15.63%) cases, hydrocephalus in 5 (15.63%) cases and sphincteric dysfunction in 4 (12.50%) cases (Table 3).

Table 3: Distribution of Other Clinical Findings.

Other Clinical Findings	n	%age
Lower Limb Weakness	16	50.00%
Scoliosis	7	21.88%
Lower Limb Muscle Atrophy	7	21.88%
Telepes Equinovarus	6	18.75%
Graded Sensory Loss	5	15.63%
Hydrocephalus	5	15.63%
Sphincteric Dysfunction	4	12.50%

Association Between Clinical Parameters and Age Groups

The chi-square tests conducted to evaluate the relationship between clinical presentation, cutaneous findings, and other clinical findings with age groups revealed distinct patterns. A highly significant association was observed between clinical presentation and age groups, indicating a strong relationship ($\chi^2(10) = 58.29$, $p < .001$, Cramér's V = 0.95). This suggests that specific clinical presentations vary significantly across different age groups. Conversely, cutaneous findings did not exhibit a significant association with age groups ($\chi^2(12) = 7.17$, $p = .846$, Cramér's V = 0.33), implying that age may not significantly impact these dermatological manifestations. However, other clinical findings displayed a moderate association with age groups ($\chi^2(26) = 27.28$, $p = .395$, Cramér's V = 0.65), suggesting a notable but less pronounced relationship compared to clinical presentation (Table 4).

Table 4: Distribution of Clinical Presentation, Cutaneous findings, and other clinical findings according to age groups.

	Age Group								Chi-square
	<1 Year		1-4 Years		5-15 Years		Total		
Clinical Presentation	**n**	**%**	**n**	**%**	**n**	**%**	**n**	**%**	
Myelomengiocele	6	18.75%	0	0%	0	0%	6	18.75%	$\chi^2(10) = 58.29$, p = <.001, Cramér's V = 0.95
Meningocele	5	15.63%	0	0%	0	0%	5	15.63%	
Lipomeningiocele	1	3.13%	6	18.75%	0	0%	7	21.88%	
Dermal Sinus	0	0%	2	6.25%	0	0%	2	6.25%	
Split cord malformation	0	0%	0	0%	5	15.63%	5	15.63%	
Thickenened Filum Terminale	0	0%	0	0%	7	21.88%	7	21.88%	
Cutaneous Findings									
Fluid sac	5	15.63%	2	6.25%	4	12.50%	11	34.38%	$\chi^2(12) = 7.17$, p = .846, Cramér's V = 0.33
Tuft of Hair	2	6.25%	2	6.25%	1	3.13%	5	15.63%	
Subcutaneous Mass	2	6.25%	0	0%	1	3.13%	3	9.38%	
Lumbosacral dimple	1	3.13%	1	3.13%	1	3.13%	3	9.38%	
Dermal Sinus	1	3.13%	1	3.13%	0	0%	2	6.25%	
Previous scar	0	0%	1	3.13%	2	6.25%	3	9.38%	
Other Clinical Findings									
Lower Limb Weakness, Lower Limb Muscle Atrophy	2	6.25%	0	0%	1	3.13%	3	9.38%	$\chi^2(26) = 27.28$, p = .395, Cramér's V = 0.65
Lower Limb Weakness, Hydrocephalus	1	3.13%	0	0%	0	0%	1	3.13%	
Graded Sensory Loss, Scoliosis	2	6.25%	1	3.13%	0	0%	3	9.38%	
Hydrocephalus	1	3.13%	0	0%	0	0%	1	3.13%	
Telepes Equinovarus, Hydrocephalus	1	3.13%	0	0%	0	0%	1	3.13%	
Lower Limb Weakness	2	6.25%	3	9.38%	3	9.38%	8	25%	
Hydrocephalus, Lower Limb Muscle Atrophy	0	0%	1	3.13%	1	3.13%	2	6.25%	
Lower Limb Weakness, Scoliosis	1	3.13%	0	0%	2	6.25%	3	9.38%	
Sphincteric Dysfunction, Telepes Equinovarus	0	0%	0	0%	1	3.13%	1	3.13%	
Sphincteric Dysfunction	1	3.13%	0	0%	1	3.13%	2	6.25%	
Telepes Equinovarus	0	0%	0	0%	2	6.25%	2	6.25%	
Telepes Equinovarus, Lower Limb Muscle Atrophy	1	3.13%	0	0%	1	3.13%	2	6.25%	

Graded Sensory Loss	0	0%	2	6.25%	0	0%	2	6.25%
Sphincteric Dysfunction, Scoliosis	0	0%	1	3.13%	0	0%	1	3.13%

Association Between Clinical Parameters and Gender

The chi-square analysis for the association between clinical presentation, cutaneous findings, other clinical findings, and gender did not reveal any significant associations in all comparisons. The clinical conditions versus gender ($\chi^2(5) = 0.86$, $p = .973$, Cramér's $V = 0.16$) and cutaneous findings versus gender ($\chi^2(6) = 1.36$, $p = .968$, Cramér's $V = 0.21$), were not significant, indicating that the variables were not related. Likewise, other clinical findings by gender ($\chi^2(13) = 11.83$, $p = .542$, Cramér's $V = 0.61$) did not show a significant association, suggesting that gender is not the determining factor for these clinical outcomes (Table 5).

Table 5: Distribution of Clinical Presentation, Cutaneous findings, and other clinical findings according to Gender.

	Gender						Chi-square
	Male		Female		Total		
Clinical Presentation	**N**	**%**	**n**	**%**	**n**	**%**	
Myelomengiocele	4	12.50%	2	6.25%	6	18.75%	$\chi^2(5) = 0.86$, $p = .973$, Cramér's $V = 0.16$
Meningiocele	3	9.38%	2	6.25%	5	15.63%	
Lipomeningiocele	5	15.63%	2	6.25%	7	21.88%	
Dermal Sinus	1	3.13%	1	3.13%	2	6.25%	
Split cord malformation	4	12.50%	1	3.13%	5	15.63%	
Thickenened Filum Terminale	5	15.63%	2	6.25%	7	21.88%	
Cutaneous Findings							
Fluid sac	8	25%	3	9.38%	11	34.38%	$\chi^2(6) = 1.36$, $p = .968$, Cramér's $V = 0.21$
Tuft of Hair	3	9.38%	2	6.25%	5	15.63%	
Subcutaneous Mass	2	6.25%	1	3.13%	3	9.38%	
Lumbosacral dimple	2	6.25%	1	3.13%	3	9.38%	
Dermal Sinus	2	6.25%	0	0%	2	6.25%	
Previous scar	2	6.25%	1	3.13%	3	9.38%	
Other Clinical Findings							
Lower Limb Weakness, Lower Limb Muscle Atrophy	2	6.25%	1	3.13%	3	9.38%	$\chi^2(13) = 11.83$, $p = .542$, Cramér's $V = 0.61$
Lower Limb Weakness, Hydrocephalus	1	3.13%	0	0%	1	3.13%	
Graded Sensory Loss, Scoliosis	3	9.38%	0	0%	3	9.38%	
Hydrocephalus	1	3.13%	0	0%	1	3.13%	
Telepes Equinovarus, Hydrocephalus	1	3.13%	0	0%	1	3.13%	
Lower Limb Weakness	6	18.75%	2	6.25%	8	25%	
Hydrocephalus, Lower Limb Muscle Atrophy	2	6.25%	0	0%	2	6.25%	
Lower Limb Weakness, Scoliosis	2	6.25%	1	3.13%	3	9.38%	
Sphincteric Dysfunction, Telepes Equinovarus	1	3.13%	0	0%	1	3.13%	
Sphincteric Dysfunction	1	3.13%	1	3.13%	2	6.25%	
Telepes Equinovarus	1	3.13%	1	3.13%	2	6.25%	
Telepes Equinovarus, Lower Limb Muscle Atrophy	1	3.13%	1	3.13%	2	6.25%	
Graded Sensory Loss	0	0%	2	6.25%	2	6.25%	
Sphincteric Dysfunction, Scoliosis	0	0%	1	3.13%	1	3.13%	

DISCUSSION

Spinal dysraphism is among congenital disorders of the spinal cord and column that are caused by incomplete closure of the caudal neural tube in early embryogenesis.[7,8] It includes both the exposed and the occult types in which edema at birth is a prevailing feature and it also co-occurs with other congenital anomalies.[9-11] In our study, we found that clinical manifestation is associated with age groups but cutaneous findings do not seem to be affected by age. Gender does not have a significant association with either clinical manifestations or cutaneous features. Nevertheless, the other clinical findings have a moderate relationship with gender, but this relationship is less pronounced than that of age groups. Such findings indicate that age is the main factor in determining the clinical presentation, but gender might have a more complicated influence on some clinical aspects, apart from the dermatological manifestations. In the research conducted by Gohar et al., the mean age of the participants was 1.5 years with a standard deviation of 5 months. The gender distribution consisted of 68.7% males and 31.3% females.[12] Clinical presentations revealed intriguing patterns, with myelomeningocele being most prominent in both the <1 year and 1-4 years age groups (18.75% each), suggesting the early onset of this condition. Meningocele, seen in 15.63% of patients aged <1 year, may indicate its occurrence during early development. The lower prevalence of lipomeningocele in <1 year (3.13%) may be attributed to its later presentation. Dermal Sinus, observed in 6.25% of patients aged 1-4 years, possibly reflects its emergence during this age range. Notably, split cord malformation (15.63%) and thickened filum terminale (21.88%) were more prevalent in patients aged 5-15 years, potentially indicating their delayed onset or diagnosis.

In the study conducted by Khan et al., the average age at treatment was 23 months with a standard deviation of 39.77 months. The most common presenting symptom was back swelling, reported in 52.8% of cases, followed by lower limb weakness. The distribution of specific conditions among patients included meningocele in 11.2%, myelomeningocele in 47.2%, myelomeningocele with hydrocephalus in 12.4%, tethered cord syndrome in 25.8%, and diastematomyelia in 3.4% of patients.[13] In our study lower limb weakness (50.00%) and lower limb muscle atrophy (21.88%) predominated among other clinical findings, highlighting the substantial motor impact of spinal dysraphism. In the study by Gupta et al, a significant portion of patients presented with specific symptoms: 94% reported back swelling, 47% experienced CSF leaks, 30% reported bladder and bowel incontinence, and 3% had bladder incontinence without bowel involvement. There were no cases of upper limb weakness or deformity observed, but lower limb weakness and deformities. Hydrocephalus was reported in 77% of the patients.[14] In the study by Rehmani et al., the most prevalent clinical presentations among patients were muscular weakness (70%), foot deformity (40%), bladder disturbance (36%), and skin abnormality (28%). Myelomeningocele emerged as the most common pathological pattern (56%), followed by lipomyelomeningocele (16%), diastematomyelia (10%), congenital dermal sinus (8%), hypertrophied filum terminale (8%), and meningocele (2%).[15] Cutaneous findings in our study were led by fluid sac (34.38%) and tuft of hair (15.63%), likely reflecting their visibility and ease of detection.

In a study conducted by Taj, a total of 1000 neonates underwent an examination, revealing cutaneous signs suggestive of spinal dysraphism in 135 (13.5%) of the newborns. Among these, the most prevalent sign was a sacral dimple, observed in 128 (12.8%) neonates, while 5 (0.5%) exhibited features consistent with meningomyelocele, 1 (0.1%) had a dermoid cyst, and 1 (0.1%) neonate presented with acrochordons.[16] Choi et al, conducted a study involving 6,558 neonate or infant patients with cutaneous stigmata, finding a pooled proportion of occult spinal dysraphism

(OSD) cases at 2.8%. Patients with combined stigmata had a significantly higher OSD association (10.5%) compared to those with a single stigma (2.3%), and atypical dimples were more strongly associated with OSD (8.8%) than simple dimples (0.6%).[17] Nasir et al. stated that there were more cases of congenital spinal malformations among females (60.4%) and individuals aged under 20 years (93.9%). The most common spinal cord anomaly was tethered cord, followed by spina bifida, diastematomyelia, vertebral segmentation anomalies, myelomeningocele, meningocele, lipomyelomeningocele, lipoma of filum terminale and sacral agenesis. Observed associated abnormalities were scoliosis (42.4%), syrinx (32.6%), and dural ectasia (27.8%) among many others.[18]

According to Ranjan et al., out of 50 patients, the following percentages of primary pathologies were detected: 98% were diagnosed with hydromyelia, 94% were tethered, 92% had lumbosacral MMC, and 88% had AC. The main lesions in only 10% of patients with cervicothoracic meningomyelocele, diastematomyelia, and intraspinal lipoma were the primary pathologies.[19] Guggisberg et al. study reported the cases of 2.3 years old patients on average presenting with lumbosacral cutaneous lesions (range, 2 days–16 years). The 67% of the patients had an isolated lumbosacral cutaneous lesion while the other 33% had 2 or more lesions. The most commonly found lesions, either isolated or in combination, were port wine stains (n=26) and deviation of gluteal furrow (DGF) (n=15). In addition, there were 11 cases of lipomas. In 5 patients, dermal sinuses were depicted as a punctum that was not explored during the consultation.[20]

These studies altogether draw attention to various aspects of spinal dysraphism as it presents in many different forms. Sometimes the different manifestations of the disease are seen earlier in development, while others may emerge later in childhood or even delay their diagnosis. The effect on patients is immense, causing motor impairments, bladder and bowel dysfunction, and consequential conditions such as hydrocephalus. Cutaneous signs range from simple to complex, and they can be used as an indicator of early disease.

LIMITATIONS

The study is limited by its single-institutional context and a relatively modest sample size of 32 patients. The retrospective design precluded the evaluation of long-term outcomes and intervention efficacy.

CONCLUSION

The study revealed a diverse age distribution, with a significant proportion of patients under 1 year of age. Myelomeningocele and Meningocele were the predominant clinical presentations in this cohort, especially among infants. Notable cutaneous findings included Fluid sac and Tuft of Hair. Clinical findings like Lower Limb Weakness and Lower Limb Muscle Atrophy were prevalent, emphasizing the condition's impact on motor function.

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Additional Information

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Human Subjects: Waiver of Consent was obtained from the ethical review board.
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AUTHORS CONTRIBUTION

Sr.#	Author's Full Name	Intellectual Contribution to Paper in Terms of:
1.	Hassaan Zahid Muhammad Irfan & Syed Shahzad Hussain	1. Study design and methodology.
2.	Hassaan Zahid & Hassan Ali	2. Paper writing.
3.	Hassan Ali & Asad Iftikhar Shah	3. Data collection and calculations.
4.	Usman Ahmad Kamboh & Hassaan Zahid	4. Analysis of data and interpretation of results.
5.	Asad Iftikhar Shah & Usman Ahmad Kamboh	5. Literature review and referencing.
6.	Hassaan Zahid & Syed Shahzad Hussain	6. Editing and quality insurer.