

ORIGINAL ARTICLE

Cutaneous Markers of Spinal Dysraphism: A Cross-Sectional Study in 230 Children

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Pak Pediatr J 2026; 50(1): 20-27

ABSTRACT

Objective: The objectives of the study were to identify the pattern and distribution of cutaneous lesions linked to spinal dysraphism in newborns and children with neurological complaints or spinal defects.

Study Design: Cross-sectional observational study.

Place and Duration of Study: Department of Pediatric Neurology and Pediatric Neurosurgery, Children Hospital & Institute of Child Health, Faisalabad, from January 2023 to December 2024.

Material and Methods: 230 neonates and children (aged 0-3 years) were enrolled. Inclusion criteria were the presence of evident midline cutaneous lesions as well as neurological or spinal abnormalities. Detailed demographic and clinical data were collected, with emphasis on the number, type, and anatomical location of skin lesions.

Results: Among the 230 subjects, 67% were female, and 76% presented within the first month of birth. Lumbosacral lipomas (44%), hypertrichosis (12%), sacral dimples (8%), vascular nevi (7%), and other lesions (9%) were the most common skin markers. There were several cutaneous abnormalities in 64% of children, with the bulk of lesions (93%) located in the lumbosacral region.

Conclusion: Spinal dysraphism in early childhood is significantly indicated by cutaneous symptoms, especially in the lumbosacral region. So any child presenting with a cutaneous marker, especially in the lumbosacral region, spinal dysraphism should be ruled out.

Key Words: *Spinal dysraphism, Congenital abnormalities, Lumbosacral, Lipoma*

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Received for publication: Dec 03, 2025
Revision accepted: Jan. 08, 2026

How to cite this: Rana FF, Bibi A, Javed I, Iqbal F, Fazal G, Rubab H
Cutaneous Markers of Spinal
Dysraphism: A Cross-Sectional Study
in 230 Children

DOI:

<https://doi.org/10.66347/ppj.v50i1.643>

INTRODUCTION

Spinal dysraphisms (SD) include a variety of congenital spinal and spinal cord malformations as a result of defective embryonic development. Both open and closed (occult) variants are included in the term, which W. B. Leichenstein used initially in 1940.¹ The most prevalent kind of open SD, myelomeningocele, accounted for

52.63% of cases in a study including 38 children. Meningocele and myelomeningocele are two further forms of open SD that involve revealed neural tissue.² However, closed or concealed spinal dysraphism (OSD), which encompasses tethered cord syndrome and split spinal cord malformation, is characterized by skin-covered neural tissue.³

With SD, the most common congenital disorder of the central nervous system, significant neurological dysfunctions are often caused. Magnetic resonance imaging (MRI) is the primary diagnostic method because it provides high-resolution imagery for surgical planning, diagnosis, and postoperative monitoring.⁴ In OSD in particular, cutaneous markers are crucial external indicators of spinal abnormalities that exist underneath. According to Lotfinia et al. (2014) and Guggisberg et al. (2004), lesions such as hypertrichosis, lipomas, dermal sinuses, and midline dimples are frequently the first noticeable signs, necessitating further neurological and radiological testing. While not all skin lesions are pathognomonic, when multiple or unique midline lesions are present, the likelihood of spinal abnormalities is significantly raised.^{5,6}

Congenital skin lesions like facial angiofibromas, neurofibromas, and café-au-lait macules are seen in neurocutaneous disorders such as tuberous sclerosis complex (TSC) and neurofibromatosis Type 1 (NF1), and neurodevelopmental problems like autism spectrum disorder.^{7,8} Given the severe neurological consequences of SD, early detection of cutaneous symptoms can be a valuable tool for early diagnosis. They may be early indicators of spinal dysraphism to facilitate timely diagnosis and treatment.^{2,9}

Mild cutaneous complaints, like a tuft of hair or a dimple, at birth are maybe early indicators of spinal abnormalities, thus increasing suspicion of severe diseases. Proper dealing and early detection of these skin lesions can timely diagnose and treat them. If left unchecked, they can avert crippling neurological consequences. This study sought to determine the patterns of cutaneous lesions associated with neural tube abnormalities in babies and children in order to increase clinical awareness and expedite identification in patients who are at risk. The main focus of the study is to understand the most common site and type of cutaneous lesion associated with spinal dysraphism.

MATERIAL AND METHODS

This descriptive cross-sectional study was conducted from January 2023 to December 2024 at the Department of Pediatric Neurology and Pediatric Neurosurgery, Child Health & Institute of

Child Health, Faisalabad. The goal of the study was to determine the distribution of cutaneous indicators linked with spinal dysraphism in newborns and children with neurological symptoms and spine defects. Inclusion criteria included neurological abnormalities (such as limb weakness, hypotonia, or aberrant reflexes) or spinal deformities, as well as evident midline cutaneous lesions on the back. The parents who did not give consent and healthy babies or infants with incidental skin lesions and no neurological signs were excluded. The study included 230 neonates and children aged birth to three years who were sent to the neurology or neurosurgery departments.

Informed consent was taken from the parents of each child at the time of enrolment. Participants' demographic information included age and gender. A thorough physical examination was performed, with a special emphasis on the spine and surrounding skin, to find and document any midline cutaneous indicators suggestive of concealed spinal dysraphism. The cutaneous markers examined were lumbosacral lipomas, hypertrichosis, sacral dimples, vascular nevi, and a range of other abnormalities. Each lesion's anatomical position was described as lumbosacral, cervical, thoracic, cranial, or mixed if it affected more than one region. The number of lesions observed (single or numerous) was also recorded. The lesions were also classified on the basis of structural type as a lumbosacral lipoma, hypertrichosis, sacral dimple, vascular nevi, and others. The collected data were systematically tabulated and descriptively analyzed.

RESULTS

A total of 230 participants were included in the study. Of these, 67% (n = 154) were female and 33% (n = 76) were male, indicating a female predominance among patients presenting with spinal dysraphism and visible cutaneous markers. The age of participants ranged from birth to three years, with a mean age of 26 days. The majority of children (76%) presented before one month of age (n = 175), while 19% (n = 44) were between 1 month and 1 year, and 5% (n = 11) were above 1 year of age (**table 1, fig 1**).

TABLE 1: Sociodemographic characteristics of study participants (n = 230)

Variable	Category	Frequency (n)	Percentage (%)
Sex	Female	154	67%
	Male	76	33%
Age Group	< 1 month	175	76%
	1 month – 1 year	44	19%
	> 1 year – 3 years	11	5%

(3%, n = 7), cranial region (2%, n = 5), and thoracic region (1.5%, n = 3). Only 0.4% (n = 1) had involvement of multiple regions (i.e., mixed) (fig 2&3).

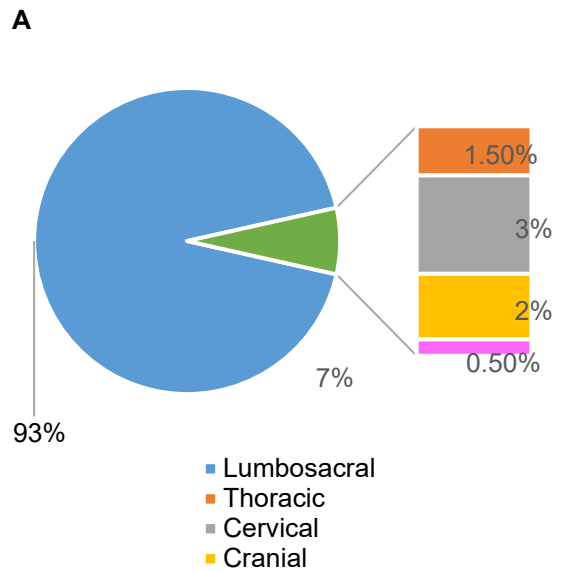
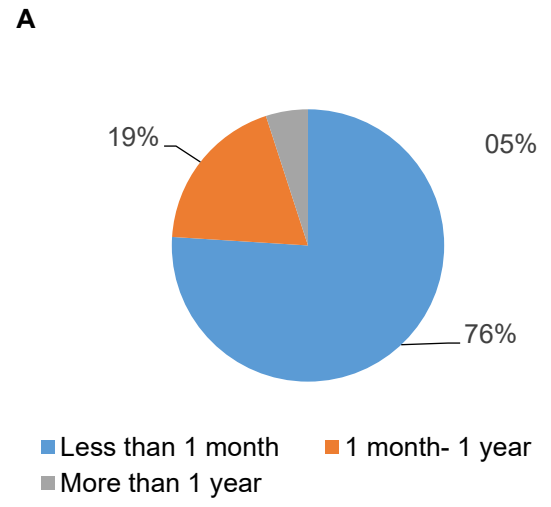
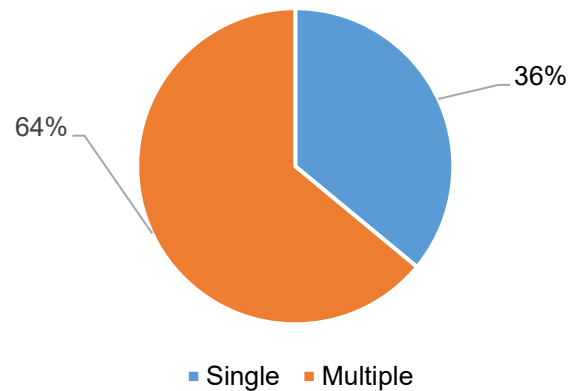
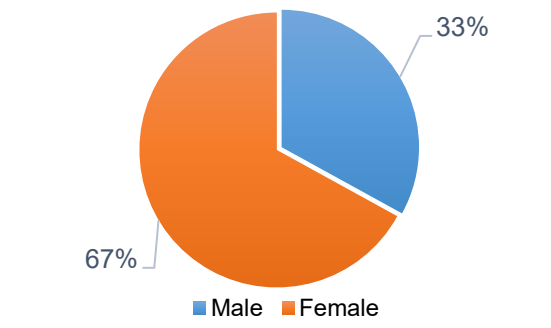


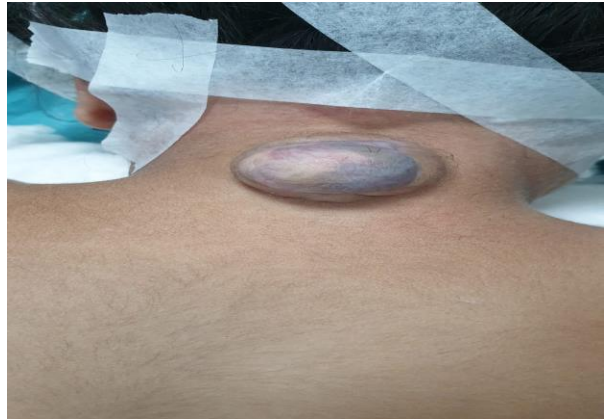
Fig 1(A & B): Gender and age distribution among participant

Out of 230 children, 64% (n = 147) exhibited multiple cutaneous lesions, while 36% (n = 83) had a single lesion. The lumbosacral region was the most frequently involved site, seen in 93% (n = 214) of cases, followed by the cervical region

Fig 1: Percentage Distribution of Cutaneous Lesions by Site and Number



A



D



B



E



C



F

Fig 2: Cutaneous marker at A&B - Lumbosacral; C - Thoracic; D - Cervical; E - Cranial; F - Mixed

The most common cutaneous marker was lumbosacral lipoma, found in 64% (n = 147) of the participants. This was followed by: hypertrichosis in 12% (n = 28), Sacral dimples in 8% (n = 18), vascular nevi in 7% (n = 16), and mixed lesions in 9% (n = 21), which included sinus tracts, atretic meningoceles, and dermal pits (Fig. 4&5).

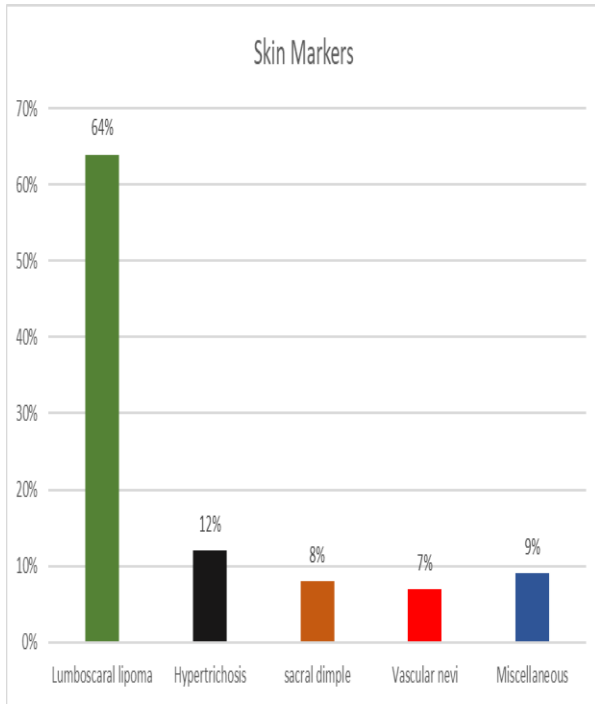


Fig 3: Percentage distribution of the type of cutaneous marker



A



B



C



D

Fig 4: A-Lumbosacral lipoma; B - Hypertrichosis; C - Sacral Dimple; D - Vascular nevi

DISCUSSION:

In this study of 230 pediatric patients with spinal dysraphism and apparent cutaneous signs, there was a 67% female predominance, with the majority of children (76%) presenting within their first month of life. The most prevalent skin marker was lumbosacral lipomas (64%), and the majority (64%) had several cutaneous abnormalities, most of which were in the lumbosacral region (93%). The observed female predominance (67%) in the current study is consistent with previous findings. Female patients are more likely to experience cutaneous signs of spinal dysraphism.¹⁰ This conclusion, though, varies from study to study. A prospective study conducted in India found a 2:1 male-to-female ratio among patients with spinal dysraphism and cutaneous lesions, indicating a higher frequency in men.¹¹ These variations show that the gender distribution of spinal dysraphism may be influenced by geographic, ethnic, or genetic variables.

The mean age of our study populations was 26 days (76%), i.e., arrived before the age of one month. This early appearance matches clinical expectations because spinal dysraphism's cutaneous stigmata are usually visible at birth and necessitate prompt medical assessment. As early visual indicators, lesions — particularly lipomas and regions of hypertrichosis — are a reason for major concern, referral, and diagnosis. Multiple cutaneous lesions were present in 64% of the individuals in this investigation. This research indicates that the predictive value for occult spinal dysraphism is significantly increased when two or more midline skin indicators are present.¹² Evidence thus suggests that in patients with several lesions, comprehensive and early imaging is essential.

According to O'Neill et al. (2017), dimples in individuals with occult spinal dysraphism (OSD) were generally observed to be around 15 mm above the coccyx.¹³ This suggests that there is a strong relationship between the position of lesions and the possibility of underlying spinal abnormalities. According to Taj, the sacral area is the most commonly involved, affecting 12.8% of newborns.¹² The most common site in our study is also the lumbosacral region in accordance with the literature.

The most common marker in this study is lipomas, which appear as subcutaneous lumps. Deeg et al. reported that lipomas are the most common cutaneous markers linked to tethered cord syndrome.¹⁴ Deeg et al. discovered that lipomas are one of the best indicators of underlying dysraphism.¹⁴ According to Taj (2018), a dimple in the sacral region was the most common skin lesion associated with neurological diseases.¹² The differences in the results may be due to different populations being studied. Since cutaneous lesions may have significant neurological repercussions, more research is required to ascertain whether they exist. Neurosurgical advice must be sought quickly to prevent irreparable issues such as lower limb motor impairments, bladder and bowel problems, progressive tethered cord syndrome, and repeated meningitis.¹⁵ These results highlight the importance of early imaging and newborn screening for children with prominent cutaneous stigmata suggestive of spinal dysraphism.

This study emphasizes the value of early screening, imaging, and multimodal therapy for all children presenting with cutaneous midline lesions at birth. A practical diagnostic step-by-step approach and anatomical clinico-radiological classification should be applied.^{16,17} All the suspected cases should be sent to a multidisciplinary team before opting for surgery.¹⁸⁻²⁰ However, there are certain limitations, such as the data is collected from patients presenting in the Neurology department, which constrains generalizability. Further population-based research is required to determine the true distribution and diagnostic use of these markers. Future studies should examine the relationship between these skin indicators and other systemic or dermatological conditions, such as latex allergy, which has been linked to spinal anomalies in some patients.^{21,22} Improved clinical assessment and long-term care plans for impacted children may result from an understanding of these linkages.

Furthermore, there is a pressing need to raise primary care and obstetricians' understanding of the clinical significance of midline cutaneous signs in newborns. Obstetricians can intervene in utero management of myelomeningocele with better reported outcomes.²³ Rapid identification and

repair of occult spinal dysraphism can be achieved through early detection and proper referral for imaging and professional evaluation, which can significantly improve outcomes.^{24,25} It follows that frontline healthcare providers should have access to education and training programs.

CONCLUSION

This study provides crucial details regarding the distribution and pattern of skin lesions associated with spinal dysraphism in young children. According to the data, lumbosacral lipomas are more common than other lesions, and the majority of instances happen early on, usually in the first month of life. Enhancing clinical identification and guiding early diagnostic testing requires an understanding of these patterns. The findings emphasize the need to raise awareness among obstetricians and primary care physicians so that all children with cutaneous lesions should be referred for thorough evaluation.

Source of funding: None

Conflict of interest: None

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Author's Contribution

FFR: Conception and design

AB: Data analysis and interpretation

IJ: Manuscript writing, proof reading

FI: Data collection

GF: Data collection

HR: Data collection

All the authors have approved the final manuscript draft and accept the responsibility of research integrity.