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Single Case

Spinal Dysraphism Presenting as Neuropathic Ulcers: A Case Report of a Delayed Diagnosis

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Keywords
Neuropathic ulcers · Spinal dysraphism · Congenital spinal defect · Tethered cord syndrome · Case report

Abstract
Spinal dysraphism (SD) refers to the abnormal fusion of dorsal midline structures during embryogenesis. It encompasses a variety of congenital spinal defects, ranging from an overt defect in which neural tissue is exposed with no overlying skin (open SD) such as myelomeningoceles to skin-covered malformations (closed or occult SD). A 13-year-old boy presented with recurrent multiple painless ulcers and erosions over the tips of the toes, mainly involving the right foot with hemorrhagic crusts for 5 years. A review of systems revealed back pain, urine incontinence, and numbness in his right knee. He was diagnosed with peripheral neuropathic ulcers and tethered cord syndrome secondary to SD and confirmed by MRI. He underwent cord detethering and lipoma resection as well as expectant therapy with satisfying outcomes. Physicians should consider early diagnosis of SD to avoid later neurological complications of SD (traction and/or pressure on the spinal cord) when infants are presented with such anomalies: MRI, close follow-up, and neurosurgical intervention may be recommended.

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Introduction

Spinal dysraphism (SD) refers to abnormal fusion of dorsal midline structures during embryological development. The skin and the nervous system share an ectodermal origin, emphasizing that concurrent anomalies of these tissues are common. The cutaneous markers of SD are located in the midline, usually on the lower back, thus serving as a valuable marker for SD. In the majority of patients, these markers lead to the diagnosis of occult SD. The skin complications related to SD are due to neuropathy, mainly neuropathic ulcers.

Here we report a case of a 13-year-old boy with an unusual and late presentation of SD, which required neurosurgical intervention to prevent potentially irreversible neurological damage. Our case report highlights the importance of early detection of SD to avoid delayed diagnosis and complication.

Case Report

A 13-year-old boy was brought by his family to the outpatient dermatology clinic complaining of recurrent painless skin lesions on his feet for 5 years. According to his parents, he improved during the COVID-19 quarantine when he stopped playing outside. A review of systems revealed back pain, urine incontinence, and numbness in his right knee. Past medical history of lipoma on his lower back, imperforated anus, and hypospadias since birth; surgical management was made with good outcomes. He was admitted to NICU and then continued follow-up for 4 years. No significant history of medication uses or similar cases in the family. The patient recorded good family support and has suitable relationships with his peers.

Skin examination revealed multiple ulcers and erosions over the tips of the toes, mainly involving the right foot with hemorrhagic crusts (Fig. 1). Nails are thin and brittle, and there was a linear vertical scar over the lower back.

Neurological examination revealed hypesthesia of the right leg at L5, left leg at L4 and S1, and bilateral power of the lower limbs 5/5. He was diagnosed with peripheral neuropathic ulcers and tethered cord syndrome secondary to SD. He was referred to a neurosurgery clinic, and MRI confirmed the diagnosis of transitional lipomyelomenigiocele with tethered cord. No diagnostic challenges were faced. He underwent cord detethering and lipoma resection (Fig. 2).

The neuropathic ulcers are treated with a topical Fucidin cream for 2 weeks, proper wound dressing, off-loading measures, and avoiding trauma. The patient reports improvement of neuropathic ulcers without new skin lesions and good adherence to management during the follow-up appointments (Fig. 3). In patient words, the overall management has a satisfying impact on his quality of life. The CARE Checklist has been completed by the authors for this case report, attached as online supplementary material (for all online suppl. material, see https://doi.org/10.1159/000533517).

Discussion

SD refers to the abnormal fusion of dorsal midline structures during embryogenesis [1]. It encompasses a variety of congenital spinal defects, ranging from an overt defect in which neural tissue is exposed with no overlying skin (open SD) such as myelomeningoceles to skin-covered malformations (closed or occult SD). The latter includes diastematomyelia (a split in the spinal cord), meningoceles, intraspinal lipomas, lipomyelomeningoceles (protrusion of the cord extra spinally into an attached lipoma), tight or fatty filum terminale, dermoid cysts, dermal sinuses, and isolated posterior spina bifida [2].

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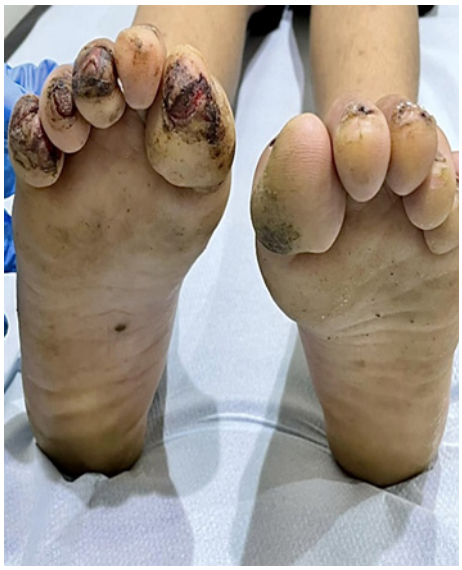

Fig. 1. Multiple ulcers and erosions on the tips of the toes, mainly over the right foot, with some hemorrhagic crusts.

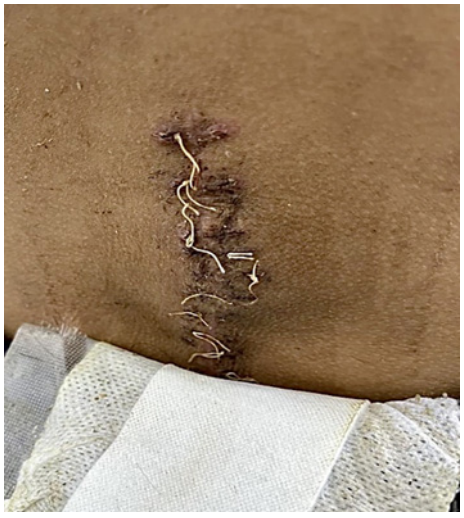

Fig. 2. Post-operative solitary vertical scar over the lower back.

SD lesion could produce traction and/or pressure, leading to progressive spinal cord damage, resulting in neurologic symptoms. Isolated posterior spina bifida is an incidental radiographic finding that is not clinically significant. Other forms of SD may become symptomatic by childhood, adolescence, or rarely by adulthood. When the diagnosis is delayed, as happened with our patient, many of the aforementioned malformations can result in tethered cord syndrome, which occurs when the conus medullaris is low-lying and/or immobile. This syndrome is characterized clinically by back pain, urinary incontinence, neurological deficits of the lower extremities, and orthopedic deformities. Early diagnosis allows neurosurgical intervention, which can prevent potentially irreversible neurologic damage [3].

Approximately 80% of individuals with closed SD have a cutaneous marker of underlying SD, and the majority have more than one type of skin lesion Lumbosacral lipomas (similar to our case) are the most commonly associated skin lesions with SD. An intraspinal lipoma or lipomyelomeningocele often represents a portion of a larger subcutaneous lipoma, typically presenting as a soft mass located above the gluteal cleft and extending asymmetrically into one buttock. A curved gluteal cleft is suggestive of such a lesion, but clinical findings may be subtle initially, becoming more noticeable with time.

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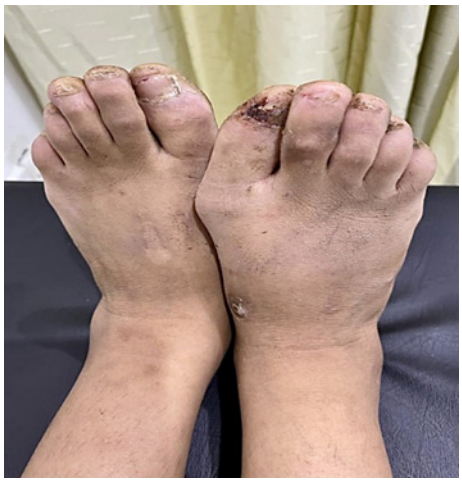

Fig. 3. Patient reports improvement on post-operative follow-up appointment.

Other cutaneous markers of SD in the lumbosacral area include deep dimples superior to the gluteal cleft, hypertrichosis with a long, coarse, or silky hair on the dorsal midline known as a faun tail, infantile hemangiomas >2.5 cm in diameter, especially the segmental pattern or those with minimal or arrested growth and vascular malformations [4–10].

LUMBAR syndrome is an acronym that has been proposed for: L, lower body/lumbosacral hemangioma and lipomas or other cutaneous anomalies as skin tags; U, urogenital anomalies and ulceration; M, myelopathy (SD); B, bony deformities; A, anorectal and arterial anomalies; R, renal anomalies. Our patient has most of these features (lipoma, SD, imperforated anus, and hypospadias). However, he lacks infantile hemangioma. It could be incomplete LUMBAR syndrome.

Cutaneous lesions of the spinal axis should alert the clinician to the possibility of SD [7]. In one study, 39% (22/56) of neonates with high-risk stigmata such as hypertrichosis, subcutaneous masses, infantile hemangiomas, tails, or dimples above the gluteal cleft were found to have SD. The presence of two or more skin lesions is a particularly strong sign of the presence of underlying SD; in another series, 61% (11/18) of such patients had SD, compared to 8% (3/36) of those with only one skin lesion [11].

High-risk cutaneous stigmata overlying the spine (like our patient) represent an indication for radiologic evaluation, and the excision for cosmetic reasons is contraindicated until an underlying abnormality is excluded. MRI is the modality of choice for occult SD patients of all ages. Unfortunately, our patient's lipoma was surgically excised without an MRI of the lumbosacral area. That behavior resulted in a late diagnosis of underlying SD; hence, the patient later developed a neurological complication of SD (traction and/or pressure on the spinal cord) and presented to us with neuropathy manifestations (numbness, hypesthesia, and neuropathic ulcers).

When infants are found to have anomalies with the potential to cause traction and/or pressure on the spinal cord, MRI, close follow-up, and neurosurgical intervention may be recommended. Our patient had surgical intervention, hopefully to stop the progression of the disease and prevent potentially irreversible neurologic damage.

Statement of Ethics

The ethical approval is not required for this study in accordance with local/national guidelines. Written informed consent was obtained from the parent of the patient for publication of the details of their medical case and any accompanying images.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

The authors of this article included Razan S. Al-Luhaibi, Shahad T. Khayyat, Suha H. Al-Sayed, Waseem K. Alhawsawi,' and Khalid A. Al Hawsawi; all of them were participated in conceptualization, project supervision, data/evidence collection, acquisition, analysis, and interpretation, investigation, and data curation, as well as writing the original draft, critical revision, editing, and final approval of the version to be published.

Data Availability Statement

All data generated or analyzed during this study are included in this article and its online supplementary files. Further inquiries can be directed to the corresponding author.

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