CASE REPORT

Sacral dermal sinus associated with lumbosacral dermoid cyst in an adolescent: a case report

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ABSTRACT

Background: Dermoid cyst is a rare, slow-growing spinal lesion that accounts for less than 1% of intraspinal tumors. It is most frequently located in the lumbar region and may be associated with a dermal sinus, which increases the risk of infection and neurological complications. Although it is usually diagnosed in childhood, it may remain asymptomatic for years and present later in life.
Case description: We present the case of a 14-year-old male patient with a sacral dermal sinus present since birth, previously unevaluated, who presented in adolescence with low back pain and purulent discharge. At admission, the patient had no neurological deficits or sphincter dysfunction. Physical examination revealed a sacral dermal sinus with hypertrichosis. Lumbosacral spine magnetic resonance imaging showed a dermal sinus extending to the dura mater, associated with an intraspinal intradural lumbosacral dermoid cyst. Surgical treatment was performed by lumbar laminectomy and complete excision of the dermal sinus and dermoid cyst, with favorable postoperative evolution and no complications.
Conclusions: Dermoid cyst represents a challenge in surgical treatment. Although recurrence is low, it may affect patients' quality of life; therefore, complete resection is the treatment of choice when feasible. This case highlights the importance of early recognition of dermal sinus and prompt evaluation with magnetic resonance imaging to prevent complications.

Keywords: Spina Bifida Occulta; Dermal Sinus; Spinal Dysraphism; Case Reports (Source: MeSH)

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Seno dérmico sacro asociado a quiste dermoide lumbosacro en un adolescente: reporte de caso

RESUMEN

Antecedentes: El quiste dermoide es una lesión espinal infrecuente y de crecimiento lento, que representa menos del 1 % de los tumores intraespinales. Se localiza con mayor frecuencia a nivel lumbar y puede asociarse a un seno dérmico, lo que incrementa el riesgo de infección y complicaciones neurológicas. Aunque suele diagnosticarse en la infancia, puede permanecer asintomático durante años y manifestarse de forma tardía.
Descripción del caso: Se presenta el caso de un paciente varón de 14 años con seno dérmico sacro desde el nacimiento, no evaluado previamente, quien debutó en la adolescencia con dolor lumbar y secreción purulenta. Al ingreso, el paciente no presentaba déficit neurológico ni compromiso esfinteriano. El examen físico evidenció un seno dérmico sacro con hipertricosis. La resonancia magnética de columna lumbosacra mostró un seno dérmico que se extendía hasta la duramadre, asociado a un quiste dermoide intrarraquídeo e intradural lumbosacro. Se realizó tratamiento quirúrgico mediante laminectomía lumbar y exéresis completa del seno dérmico y del quiste dermoide, con evolución postoperatoria favorable y sin complicaciones.
Conclusiones: El quiste dermoide representa un reto en el tratamiento quirúrgico. Aunque la recurrencia es baja, puede afectar la calidad de vida de los pacientes, por lo que la resección completa es el tratamiento de elección cuando es factible. Este caso resalta la importancia del reconocimiento temprano del seno dérmico y de la evaluación precoz mediante resonancia magnética para prevenir complicaciones.

Palabras clave: Espina Bífida Oculta; Seno Dérmico; Disrafia Espinal; Informes de Caso (Fuente: DeCS)

INTRODUCTION

Congenital dermal sinus (DS) is a cutaneous stigma of occult spinal dysraphism and a rare congenital malformation resulting from incomplete separation of the cutaneous ectoderm and neuroectoderm during the first weeks of gestation. Its incidence is estimated at approximately one in every 2,500 live births. It is most frequently located in the lumbar region, although it may also occur in the thoracic and cervical regions (1,2).

DS consists of an epithelial tract that extends from the skin surface, usually in the midline, traverses soft tissues, and may reach the vertebral column, the intraspinal space, and the dural sac, becoming intradural and potentially involving neural tissue. DS is more frequently associated with dermoid cyst (DC) than with epidermoid cyst. The clinical presentation of DS may be due to pressure from the associated cyst, the presence of a tethered cord, or secondary infection; the latter may lead to recurrent meningitis. Likewise, infection of the DS may extend through the tract to the intradural DC, producing an intradural abscess and leading to neurological complications (3).

Most DCs are congenital non-neoplastic lesions and account for less than 1% of intraspinal tumors (4); however, they represent approximately 20% of intradural tumors in the first year of life (5). Although most DCs are congenital, acquired cases have been described, potentially related to lumbar punctures or injuries (5).

DC belongs to the group of occult spinal dysraphisms and consists of a capsule containing epidermal elements and dermal derivatives. It is believed to originate from totipotent ectodermal cells that remain within the developing neural tube between the third and fifth weeks of gestation and coexists with a DS tract in approximately half of cases. The most frequent location is intradural lumbosacral (60%), followed by thoracic (10%) and cervical (5%) (5–8).

DC differs from epidermoid cyst by the presence of hair follicles, sebaceous glands, and other components. Epidermoid cysts are more frequent than DC in the lumbar region. DC is most commonly located in the conus medullaris, and its growth is due to the accumulation of desquamation products and glandular secretions within the cyst (6).

The differential diagnosis includes spinal lipoma, epidermoid cyst, teratoma, and myxopapillary ependymoma (6).

From a clinical perspective, spinal DCs may remain asymptomatic for long periods or present with radicular pain, spinal cord compression, or neurological deficits depending on their location. In some cases, they may present acutely, generally in association with cyst rupture or infectious processes related to DS (7,8). Although DC may be present since childhood, in some cases it becomes symptomatic in early adulthood (9).

Imaging studies of DS in children may begin with ultrasound, especially during the first year of life, due to the lack of ossification of the posterior elements of the spine, which provides an adequate acoustic window for evaluation of the spinal canal (3,10). This method also allows identification of associated anomalies, such as tethered cord, inclusion cysts, lipomas, and syringomyelia (3). In older patients, magnetic resonance imaging (MRI) is the method of choice for the diagnosis of these lesions and for preoperative surgical planning (4).

The treatment of choice for DC is complete surgical resection; however, when the capsule is adherent to neural structures, resection may be challenging due to the risk of neurological injury (6).

We present the case of a 14-year-old male patient with sacral DS present since birth, who presented in adolescence with low back pain and purulent discharge through the dimple. Contrast-enhanced lumbosacral MRI showed DS, tethered cord, sacral spina bifida, and the presence of a lumbosacral tumor mass suggestive of DC.

CASE DESCRIPTION

We present the case of a 14-year-old male patient with no relevant family or perinatal history, with a sacral dimple present since birth that had not been previously evaluated or studied with imaging. One year before admission to a national pediatric referral center located in Lima, increased hair growth at the level of the dimple was noted. Two months before admission, the patient presented with low back pain, as well as pain and burning during urination, for which he was diagnosed with a urinary tract infection and received antibiotic treatment. One month prior, he developed yellowish discharge through the DS, which later became purulent, prompting specialized evaluation. He received antibiotic treatment with resolution of pus and was advised to undergo contrast-enhanced lumbosacral MRI.

At hospital admission, the patient had no neurological deficits or sphincter dysfunction. Physical examination revealed hypertrichosis and a DS in the sacral region, with a cutaneous opening measuring approximately 3 × 3 mm in diameter and a depth of approximately 4 mm, without signs of inflammation or discharge on digital pressure. No purulent discharge was observed due to prior oral antibiotic treatment before hospitalization (Figure 1).

Preoperative lumbosacral MRI showed the DS tract from the skin to the S3 level, with contrast enhancement on T2-weighted sequences. Additionally, an intraspinal intradural cystic lesion from L5 to S1 was identified, measuring approximately 32 × 10 × 11 mm in the craniocaudal, transverse, and anteroposterior axes, respectively, showing heterogeneous high signal on T1- and T2-weighted sequences, without enhancement after contrast administration, associated with a DS. Furthermore, lack of fusion of the posterior arch of S2 and S3, consistent with spina bifida, was observed (Figure 2).

Surgical treatment was indicated due to the presence of low back pain, recurrent purulent discharge through the dimple, and MRI findings. Surgery was performed under microscopic visualization, without intraoperative neurophysiological monitoring, due to the absence of neurological impairment and MRI findings (lesion level). A midline incision was made, isolating and surrounding the DS tract, following its

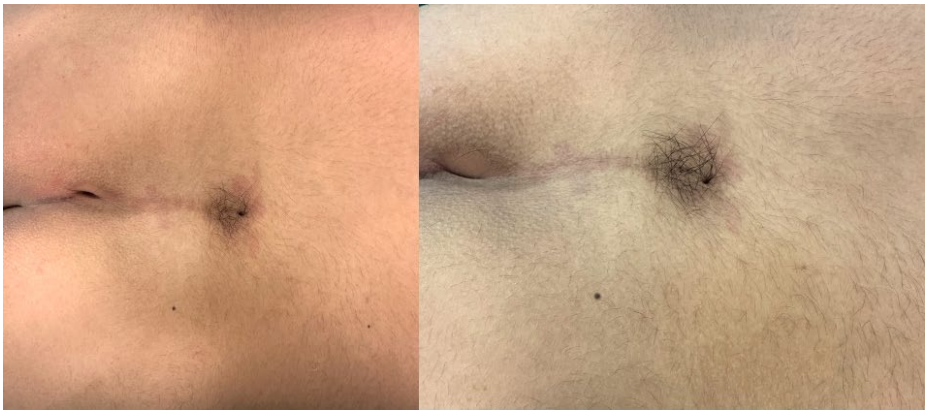

Figure 1. Image of the sacral region showing a tuft of hair and a DS in the midline, above the intergluteal cleft

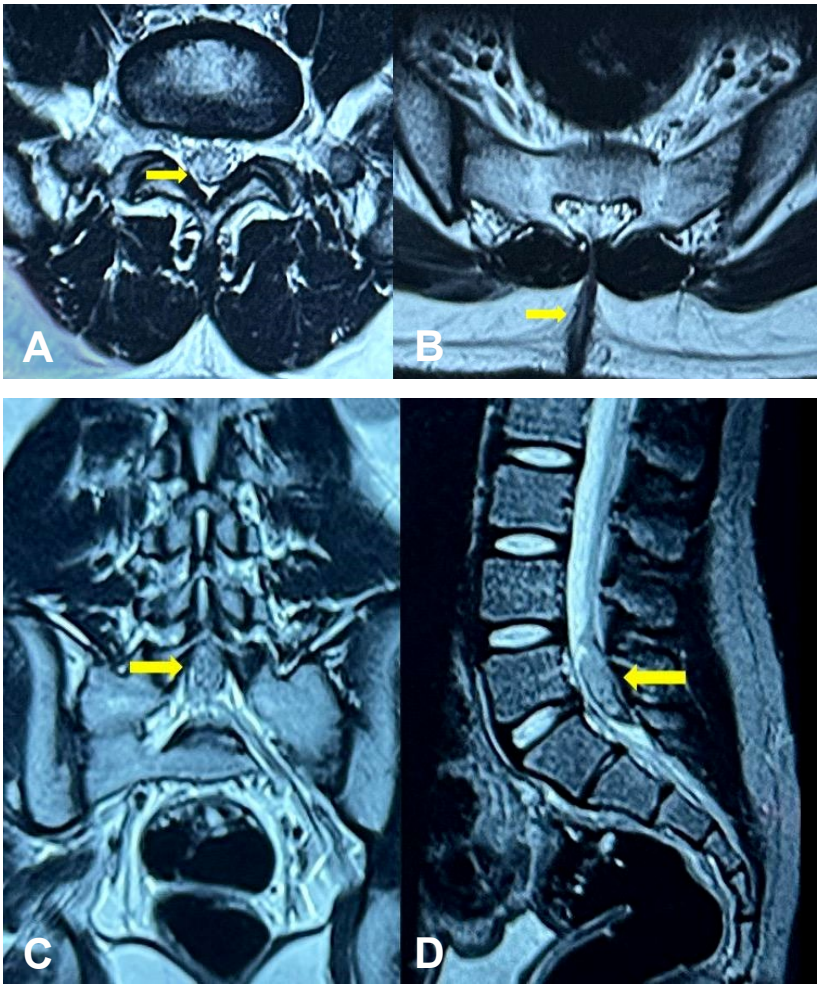

Figure 2. Contrast-enhanced lumbosacral spine MRI

A. Axial T2-weighted view showing a heterogeneous signal intradural lumbosacral cystic lesion. B. Axial T2-weighted view showing the tract from the skin to the posterior sacral region in relation to a DS. Additionally, spina bifida is observed. C. Anteroposterior T2-weighted view showing the lumbosacral cystic lesion. D. Sagittal T2-weighted view showing an intraspinal intradural lesion at the L5–S1 level.

course until reaching the extraspinal level. The DS lumen was not irrigated due to the risk of superinfection. Subsequently, S1 laminectomy and L5 hemilaminectomy were performed, visualizing the lumbosacral dura mater and the DS tract, within which hair was observed. Ligation of the DS was performed to prevent purulent discharge, followed by its excision. Dural opening was then performed in the midline in a craniocaudal direction, revealing an intradural DC projecting toward the conus medullaris. Dissection and complete excision of the DC were performed without intraoperative complications (Figure 3).

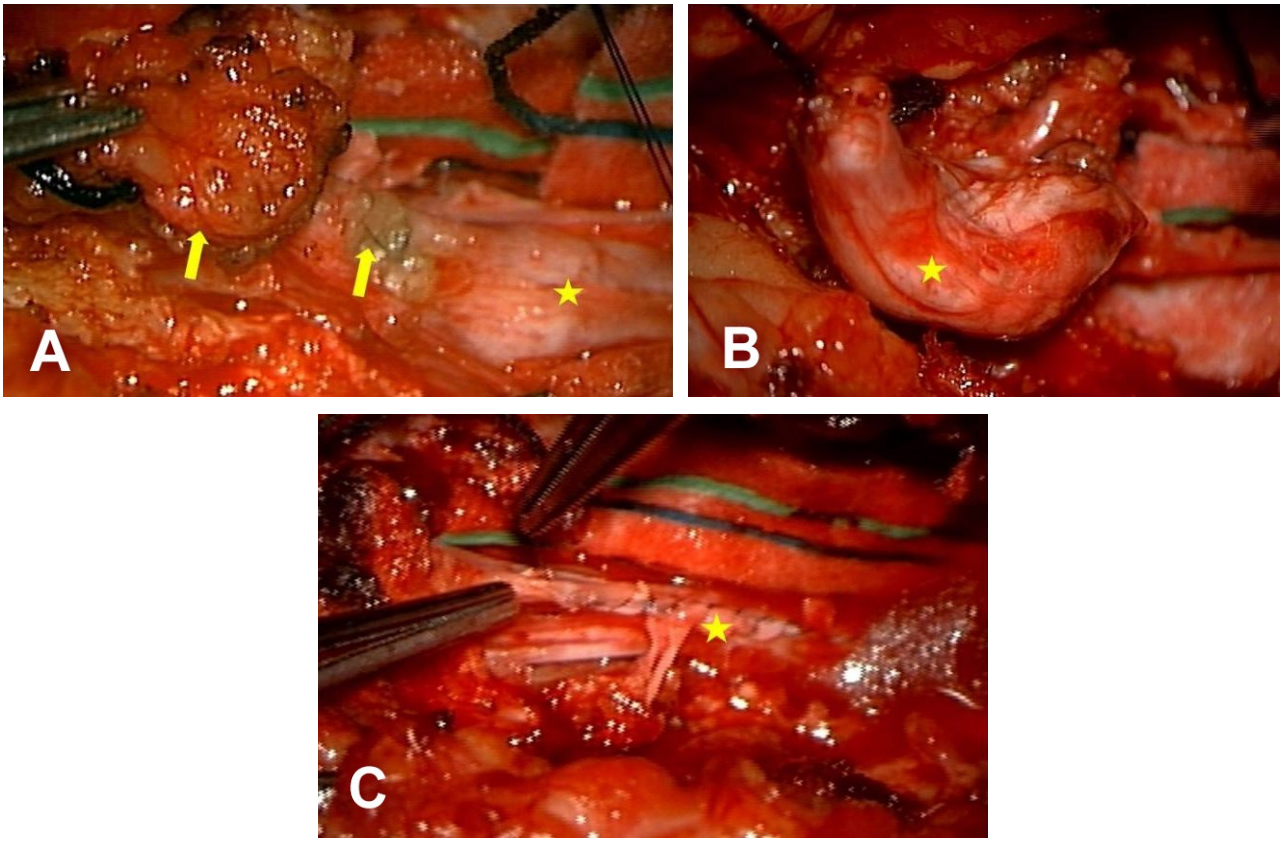

Figure 3. Intraoperative images

A. Arrows indicate the DS tract and its sebaceous content with the presence of hair; the star indicates the lumbosacral dura mater. B. The star indicates complete excision of the lumbosacral DC. C. Complete closure of the lumbosacral dura mater.

Histopathological examination of the excised intradural lesion, well-defined and encapsulated, measuring approximately 3 × 2 × 1 cm, showed a wall lined by mature keratinizing squamous epithelium, with the presence of hair follicle appendages at different stages of maturation, multiple keratin scales, and sebaceous glands. The histopathological diagnosis was DC (Figure 4).

The postoperative course was favorable and without complications, and the patient was discharged early. During outpatient follow-up over eight months after surgery, including evaluation of motor function, sphincter function, and surgical wound status, no surgery-related complications were observed. A delayed follow-up lumbosacral MRI (approximately one year after surgery) was recommended to avoid misinterpretation of findings related to early postoperative changes.

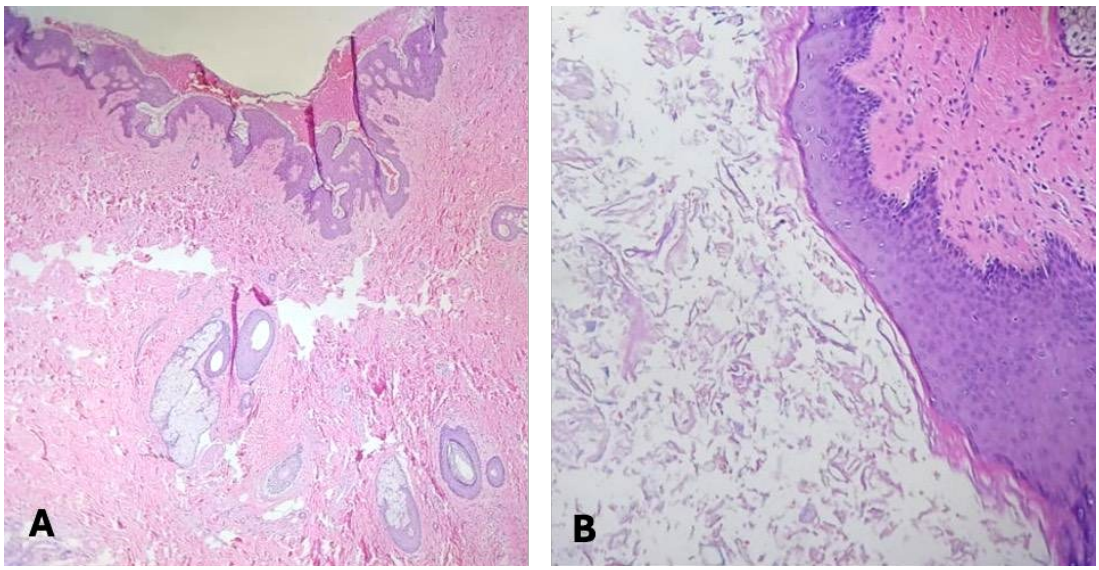

Figure 4. Optical microscopy of the DC

A. Cyst wall lined by keratinized squamous epithelium, in which pilosebaceous follicles are observed (hematoxylin and eosin staining, original magnification ×100). B. Cyst wall with squamous epithelial lining and abundant desquamation of mature keratin into the lumen (hematoxylin and eosin staining, original magnification ×200).

DISCUSSION

This case illustrates a late presentation of sacral DS associated with spinal DC, an uncommon and clinically relevant combination due to its potential to develop infectious and neurological complications. Unlike most reports, in which diagnosis is made at early ages, clinical manifestation in adolescence highlights the risk of delayed diagnosis when cutaneous stigmata suggestive of occult spinal dysraphism are not evaluated in a timely manner. It should also be considered that spinal DCs are rare entities and that there are few publications addressing this association, reinforcing the relevance of this report (5,11). Previously reported case reports documenting the association between DS and spinal DC correspond to infant patients, with ages at presentation less than or equal to 26 months (infants and young children). Some reports describe presentations complicated by central nervous system infection and early neurological involvement, including fever, episodes of recurrent meningitis, regression of motor developmental milestones, and difficulty walking; in contrast, others report predominantly local manifestations characterized by discharge through the DS with preserved neurological examination at admission (5,11–13). In these cases, DS was located at the lumbar, dorsal, or lumbosacral level (5,11–13).

In contrast, the patient presented in this report had a late onset in adolescence, with a less severe initial clinical picture, mainly characterized by low back pain and purulent discharge through the DS, without established neurological deficit or episodes of fever or meningitis at diagnosis. In this case, the DS had a low sacral tract, from the skin to the S3 vertebral level, associated with an intradural lumbosacral DC, which could help explain the less aggressive and later-onset clinical presentation observed, although this association should be interpreted with caution, given the limited available evidence. Additionally, differences in age at presentation, anatomical location, and clinical onset highlight the heterogeneity of the clinical expression of this association.

Regarding case series, two studies describing the characteristics of the association between DS and spinal DC in the pediatric population have been identified. In 2021, Sauradeep Sarkar & Vedantam Rajshekhar reported a retrospective series of 59 patients with spinal DC, of whom 45 were under 18 years of age, treated between January 1999 and December 2017. In this cohort, the male-to-female ratio was approximately 2.5:1 in the pediatric population; 60% were associated with DS, the most frequent symptom was decreased muscle strength, 33% had a history of meningitis, and the most frequent associated anomaly was lipoma in 28.9% of cases (8).

Similarly, Shabari Girishan and Vedantam Rajshekhar described a retrospective series of 10 patients with intramedullary DC, in which the initial clinical presentation was acute, characterized by paraparesis or quadriparesis. In 9 patients, DC was associated with DS, and the most frequent location was lumbosacral in 40% of cases, followed by dorsolumbar (30%), dorsal (20%), and sacral (10%). Nine patients received antibiotic treatment prior to surgery; 7 underwent early surgery and 2 late surgery, with loss to follow-up in one case (7). In contrast to previously published reports and case series describing infectious manifestations and/or

neurological deficits associated with DS, the patient presented with local manifestations characterized by low back pain and purulent discharge through the DS, without neurological deficit at diagnosis.

Both in case reports and case series, MRI played a central role in diagnosis and surgical planning and was used for postoperative evaluation in patients with available follow-up (5,7,8,11–13). Depending on their content, these cysts may show variable MRI characteristics. Classically, DCs are described as hyperintense lesions on T1-weighted sequences due to their lipid content derived from sebaceous secretions and cholesterol; however, the signal may be heterogeneous when there is a mixture of fat, keratin, cellular debris, or inflammatory content. On T2-weighted sequences, the signal may also be variable (4). In previously reported case reports, MRI allowed identification of heterogeneous intradural lesions associated with DS, as well as findings suggestive of infection, such as spinal cord edema, intramedullary abscesses, or peripheral enhancement after contrast administration.

Furthermore, in several cases, MRI enabled visualization of the DS tract from the skin to the spinal canal and definition of its relationship with associated intradural lesions, which is essential for surgical planning (5,11–13). In the present patient, MRI showed a lumbosacral intradural cystic lesion with heterogeneous signal on T1- and T2-weighted sequences and no enhancement after contrast administration, findings previously described in the literature for spinal DC. Additionally, it allowed identification of the DS tract from the skin to the spinal canal, consistent with what has been reported for this malformation.

From a therapeutic perspective, total microsurgical excision of the DC, including the capsule, is the treatment of choice (4,9). However, the extent of resection must be individualized according to clinical presentation and the degree of capsule adherence to the spinal cord or adjacent neural structures, as total excision may carry a significant risk of permanent neurological deficit. In cases where the DC is firmly adherent to the spinal cord or adjacent neural structures, partial resection may be considered to minimize the risk of neurological injury (5,7,8,11).

In patients with DS associated with DC, definitive and timely surgical intervention is recommended to confirm diagnosis, eliminate the potential source of infection (the DS), and prevent complications such as meningitis. The approach may be performed by laminectomy or laminoplasty, depending on the location of the lesion and the extent of the DS tract. The literature describes resection of the DS tract together with associated intradural lesions as part of definitive surgical treatment (5,7,11–14). In some case reports, partial resection of the DC has been described when adherence to the spinal cord or adjacent neural structures exists; in contrast, case series report complete resections when anatomical conditions allow in a significant proportion of patients (33.3% and 35.6%, respectively) (7,8). In the present case, complete microsurgical excision of the DC and associated DS was achieved, consistent with the literature indicating complete excision as the treatment of choice when anatomically feasible.

In the present case, concomitant surgical management of the DS and DC was performed through a single approach and microsurgical excision. The approach included following the DS tract to its intraspinal component and resection of the associated DC. Postoperative evolution was favorable, and no neurological or infectious complications were recorded during the evaluated period. This finding is consistent with previous case reports and case series, in which surgical treatment is associated with favorable initial clinical outcomes in a relevant proportion of patients. In particular, available series describe neurological improvement mainly in patients with acute presentation, as well as the absence of long-term tumor recurrence in those undergoing complete resection, highlighting the importance of the extent of resection and the need for prolonged follow-up.

Spinal DCs should be differentiated from other intradural lesions that may present with similar clinical and imaging findings, such as spinal lipoma, epidermoid cyst, teratoma, and myxopapillary ependymoma (6). In the present case, definitive diagnosis was established by histopathological examination, which showed a cyst wall lined by keratinizing squamous epithelium with the presence of cutaneous adnexa, a characteristic finding of DC.

STRENGTHS AND LIMITATIONS

This report has several limitations. First, it describes a rare presentation, which limits the generalizability of the findings. Additionally, the patient had a DS since birth that was not evaluated or studied at early ages, despite being a cutaneous stigma suggestive of occult spinal dysraphism, resulting in delayed diagnosis. Finally, postoperative follow-up was short, preventing assessment of long-term outcomes, including lesion recurrence or late complications.

Nevertheless, the case has relevant strengths, including complete clinical, radiological, and histopathological characterization of the lesion, as well as detailed documentation of a rare association between sacral DS and spinal DC. Furthermore, surgical management resulted in clinical resolution without immediate complications, providing useful information for the recognition and management of this uncommon entity.

CONCLUSION

This case report describes a 14-year-old male patient with DS since birth that had not been evaluated in the early stages and became clinically evident at a later stage, associated with a spinal DC. Although this association is uncommon, the case highlights the importance of timely recognition of cutaneous stigmata suggestive of occult spinal dysraphism and their evaluation through imaging studies, particularly MRI, before the onset of symptoms or complications. Surgical management through excision of the DS and DC allowed resolution of the clinical condition during the evaluated period. However, when the cyst capsule is firmly adherent to neural structures, partial excision may be considered as an alternative, depending on the potential risk of neurological deficit.

Author contributions

ARE: Conceptualization, Data curation, Formal analysis, Investigation, Methodology, Project administration, Supervision, Validation, Visualization, Writing – original draft, Writing – review and editing.

NLMS: Investigation, Writing – original draft.

Conflicts of interest

The authors declare no relevant financial or non-financial conflicts of interest.

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Data availability

The data supporting the findings of this study are available upon request from the corresponding author.

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