

Case Report

Association between cloacal exstrophy and two limited dorsal myeloschisis lesions: A case report and literature review

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Abstract

Background

The term limited dorsal myeloschisis (LDM) was coined by Pang in 1993. It involves incomplete obliteration of the dorsal neural fold in primary neurulation. On the other hand, Cloacal Exstrophy is one of the most complex endodermal anomalies and is usually associated with spinal anomalies; hence, the term OEIS complex was coined (Omphalocele, Exstrophy of the cloaca, Imperforate anus, and Spinal deformities complex).

Case Description

We report the coexistence of two LDM lesions in tandem (originating at L2 and S2 levels) in a child diagnosed with cloacal exstrophy. Initially, prenatal ultrasound detected the lumbar but not the sacral lesion. The patient was surgically untethered under intraoperative neurophysiologic monitoring (IOM) at four years of age, and this paper reports his one-year follow-up.

Conclusion

Cases of cloacal exstrophy must always be investigated for spinal cord malformations. LDM is rare and requires careful diagnosis. MRI should be done for the whole neuroaxis to rule out associated conditions, including multiplicity. Surgery should be done under IOM to avoid long-term complications.

Keywords: *Limited dorsal myeloschisis, Tethered cord, Cloacal exstrophy, OEIS complex.*

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1. Introduction

Pang invented the term ‘limited dorsal myeloschisis’ (LDM) in 1993, and it was later described as a distinct entity in 2010 (1). LDM occurs when the dorsal neural fold is not completely obliterated during primary neurulation, resulting in a discontinuity between the neural and cutaneous ectoderm layers (2). Closed myelomeningocele with fibroneural stalk formation resulting in spinal cord tethering is described as LDM (3). Glial fibrillary acidic protein (GFAP) immunohistochemistry findings are positive (2).

It is classified as saccular (stalk to the dome, basal neural nodule, and segmental myelocystocele) or nonsaccular (crater, pit, and membranous sac) according to Pang’s classification (4). It is often accompanied by a cyst linked to a subcutaneous lipoma (3). In addition, it has been linked to hydrocephalus and Chiari II abnormalities, both of which should be screened for (4). Surgery is undertaken to avoid problems in the child’s upgrowth and prevent the establishment of tumors (dermoid and epidermoid) in the neural tract (3). It entails untethering the stalk from the cord by laminectomies to access the pathology site, which is commonly done at four to eight months of age (4, 5). A pediatric cloacal exstrophy (CE) case with double LDM is presented.

2. Case Presentation

A case report is presented about a four-year-old child with double LDM coexisting with CE. Consent has been obtained from the parents according to ethical guidelines.

2.1. The case

The child was born prematurely at 35 gestational weeks and diagnosed prenatally with cloacal and

bladder exstrophy and paraspinal lumbar lipoma. After birth, the CE was revealed (Figure 1a). Upon examination of the back, there was a soft, shallow paramedian swelling, toward the left of the midline, at the lumbar area measuring 3 × 4 cm with a corresponding reddish discoloration (Figure 1b). She also had a slight asymmetry of the buttock and natal cleft in the sacral area. There were no skin defects, hair tufts, or dimples. The magnetic resonance imaging (MRI) of the whole neuroaxis (Figure 2) showed two LDMs at L2 and S3, associated with extensive syringomyelia and tethering at both levels. The CE required an initial surgical intervention at the age of nine days. Other surgical stages followed this in the following years. He had prenatal talipes equinovarus (Figure 1c), repaired at two months. Unfortunately, the patient’s planned surgical repair of the doubled LDM was postponed several times due to multiple hospitalizations for infections related to the CE and urinary tract infections. Further delays were caused due to the COVID-19 pandemic restrictions. At the time of admission, he was developmentally appropriate for his age, able to walk and run without any motor weakness, urination was through the extruded bladder, and a midline ileostomy at the umbilicus and CE. LDM with spinal cord tethering was portrayed on an MRI scan showing the stalks at L2 and S2, associated with extensive syringomyelia (Figures 3a, 4a, and 4c).

2.2. Surgical Management of the LMDs

The purpose of the procedure was to untether the cord so that normal growth and development could take place. As a result, both LDMs were planned for surgical repair. A midline incision was done under general anesthesia and IOM to accommodate both neighboring sacs (Figure 5a).



Figure 1. Post-natal clinical photographs show: (a) Cloacal extrophy complex anomaly component, showing the omphalocele (O) with the remnant of the umbilical stump (US), below the omphalocele, is the bladder extrophy as two exstrophied hemi-bladders (B) separated by exstrophied ileocecal region (IC) with some fecal (F) content, also showing the splitting of male external genitalia (G) and two hemiscrotums (S). (b) Slight swelling in the lumbar region with reddish skin discoloration.

Subcutaneous dissection was performed to liberate the sac (Figure 5b) from surrounding tissue, taking care not to puncture the sac, which would result in early CSF leaking. The stalk neck was also depicted across the myofascial plane. The lumbosacral fascia was opened, then laminotomies were performed above and below the stalk's neck. The sac was opened and followed down to the

cord's junction (Figure 5c). The stalk was detached and freed at the dorsal side of the cord with neurophysiologic monitoring. A watertight dural closure was made after nerve stimulation using sutures. When compared to baseline, intraoperative neurophysiology demonstrated an enhanced response. The recovery period was unremarkable. The postoperative MRI of the lumbosacral sagittal

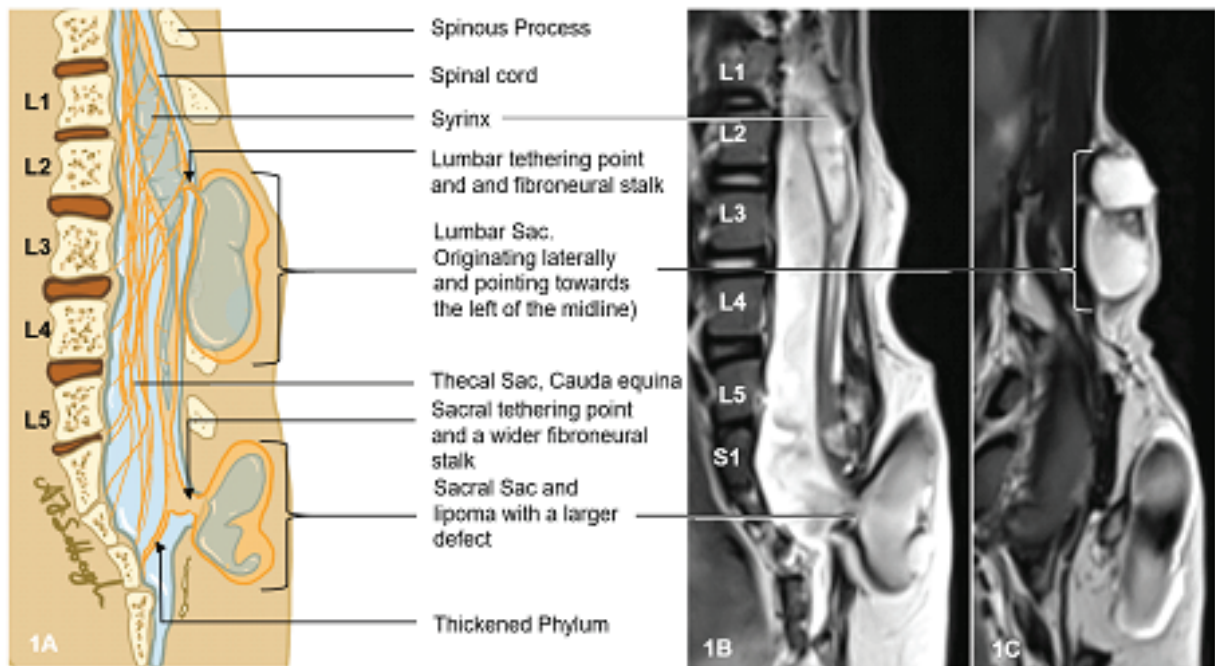


Figure 2. (a) Illustration showing Limited dorsal Myeloschisis (LDM) with stalks originating at L2 and S2 on the left side. (b) A corresponding midline Sagittal T2 weighted MRI at the midline. The lumbar LDM is not demonstrated at the midline as it is off the midline. (c) A left lateral sagittal MRI image shows both lesions.

cut shows untethering of the cord and lipoma removal (Figures 3, 4). Histopathology showed a fibroneural stalk.

2.3. Follow-up

A three-month follow-up was done, and the patient was ambulating freely with no infection. A staged treatment for bladder exstrophy was planned.

3. Discussion

Pang reported just one incidence of ‘double’ dorsal myeloschisis in the thoracic and lumbar areas in a case study of 51 individuals (1, 4, 5). Here is another example of double dorsal myeloschisis in the lumbar and sacral regions. Pang defines the sac level as the stalk cord interface corresponding to the vertebral level (5). In the author’s viewpoint, this description of the level is less obvious and may cause

confusion; the lower fibroneural stalk seen in our patient, according to this definition, is opposite the sacral vertebrae, giving the spectator the impression of a tethering below the conus. However, this is not true because the cord, in this case, is low-lying due to the stretched effect created by the stalk. Furthermore, it was reported that other authors found difficulty in appreciating the exact location of the attachment of the fibroneural stalk, in part due to the stalks being challenging to appreciate on conventional MRI, its isointense appearance to the cord, and its small size in some young children.

Our patient additionally exhibited an undetected spinal dysraphism with a midline defect in the form of bladder exstrophy and a cutaneous hemangioma in the dorsal lower sac. Pang classified LDM as saccular or flat based on its cutaneous appearance. He described it as a primary neurulation failure of fusion at a circumscribed cord segment, thus the term ‘limited.’ This failure occurs at the final stage

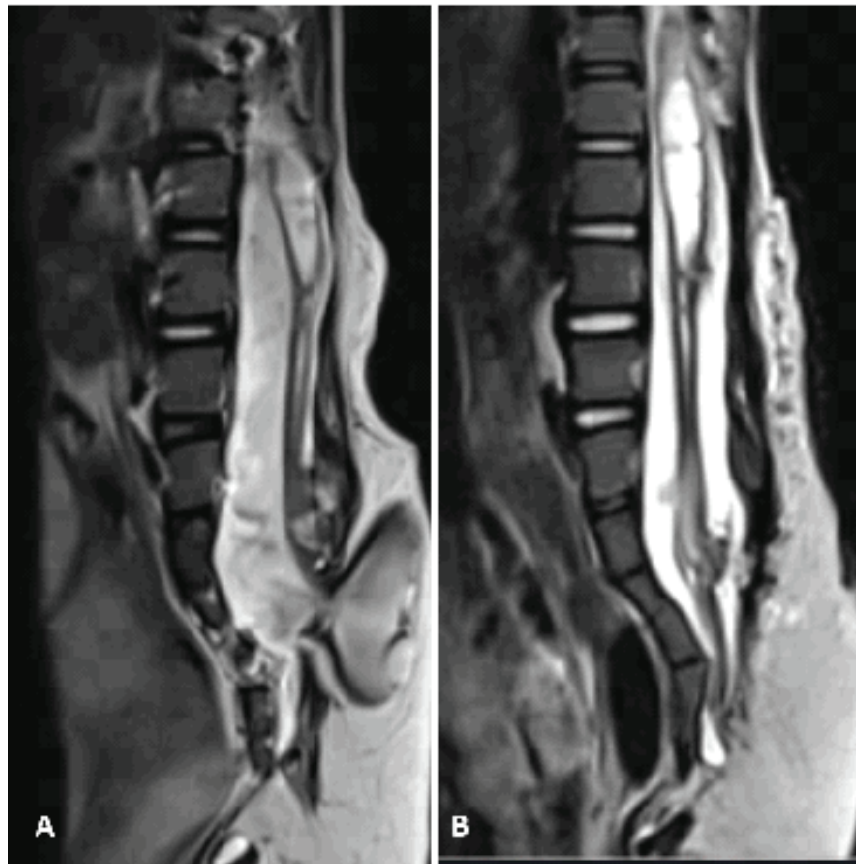


Figure 3. (a) A sagittal T2 waited MRI showing the two tethering points associated with syringomyelia. (b) Two-week postoperative corresponding T2 waited MRI illustrates untethering of the cord at both points as well as the filum with residual syringomyelia.

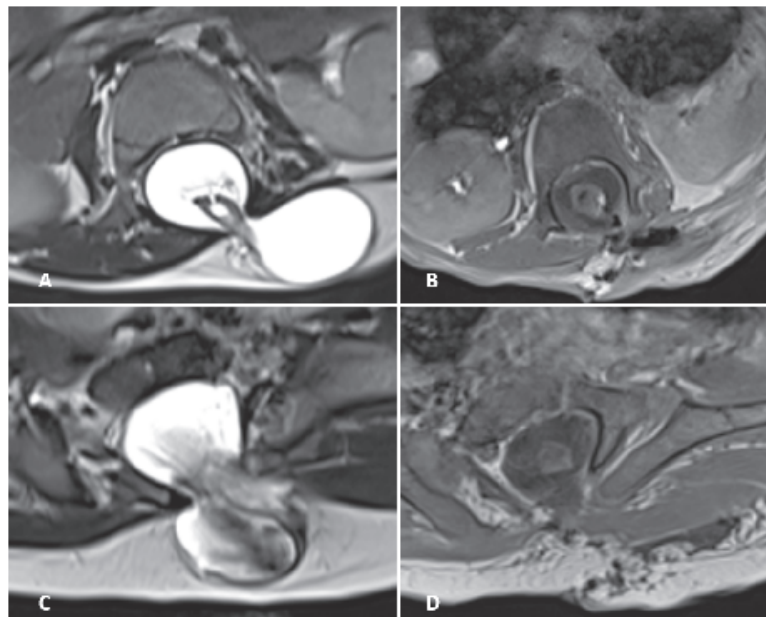


Figure 4. (a) An axial T2 waited MRI showing the lumbar LDM pointing toward the left. (b) Two-week postoperative T1 waited MRI showing the resection of the lumbar lesion and cord untethering. (c) An axial T2 waited MRI showing the sacral LDM. (d) Two-week postoperative T1 waited MRI showing the resection of the sacral lesion and cord untethering.

of dorsal fusion, which results in a tethering effect related to its fibroneural stalk that exerts tension on

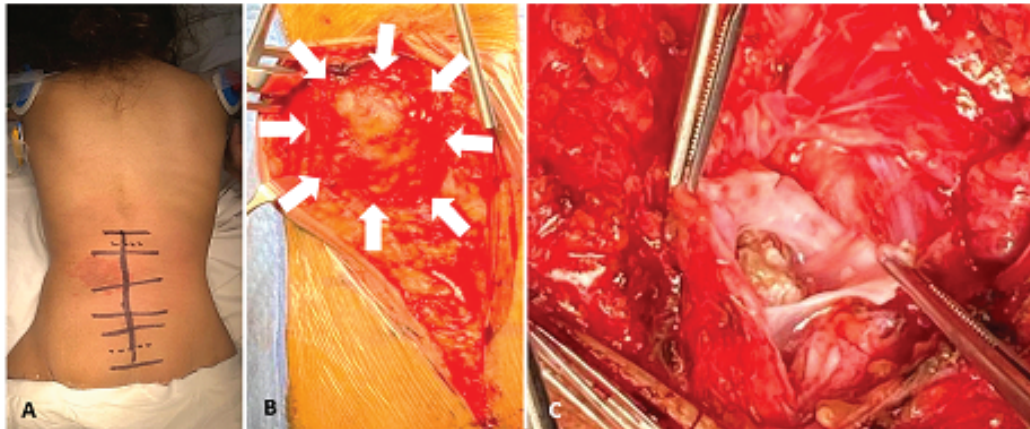


Figure 5. (a) A picture of the back showing the reddish skin pigmentation indicating the lumbar LDM. The sacral LDM is unclear and is only associated with a slight deviation of the natal cleft. A line was drawn to plan the surgical incision. (b) The lipoma of the lumbar LDM is exposed (indicated by white arrows). (c) The remnant of the lumbar sac of the LDM was resected and the cord untethered from the associated lipoma in preparation for closure.

the cord and is considered the hallmark of LDM (4). The nomenclature of cystic spinal dysraphism requires special attention. LDM is differentiated from myelomeningocele by the absence of the placode and from myelocystic by the presence of the fibroneural stalk that links the skin to the cord and causes tethering (6–8).

Saccular skin lesions are caused by CSF hydrostatic pressure squeezing around the sleeves of the dura enclosing the stalk, resulting in a saccular CSF-containing pouch (4, 5). Both sacs in our case fit the description of saccular LDM, with a clear connecting band of tissue or fibroneural stalk that connects the inner aspects of the dome to the dorsal spinal cord above the conus, producing a tethering effect of the cord as seen on the patient's lumbar MRI, resulting in syringomyelia. In our case, it was located in the thoracolumbar lesion. However, Pang reported 51 cases, in which the most common location was cervical, followed by upper thoracic (4).

CE is the most complex congenital anomaly in neonates (9, 10). Cloaca means having a common channel between gastrointestinal and urogenital tracts. CE indicates a lower abdominal wall defect where urinary and gastrointestinal tracts are exposed

and are opened directly into the anterior abdominal wall with multiple associated anomalies, including spine and sacrum. Most of these cases are associated with spinal malformations if well investigated. The Published: percentage of the associated spinal anomalies with CE is reported to be 46% (9); however, the actual rate is believed to be much higher (10). Most spinal anomalies are closed spinal dysraphisms involving spinal lipomas and almost always involve the conus and/or filum terminale (10).

Some authors name the CE as OEIS complex (omphalocele, exstrophy of the cloaca, imperforate anus, and spinal deformities complex) (11). This rare anomaly is found in 1:200,000 to 1:400,000 live births and is more commonly seen in males (9–11).

The etiology of CE is unknown. Nonetheless, genetic and environmental factors might play a role in this etiology. Certainly, this complicated anomaly occurs during early fetal life due to failure of normal development of cloacal membranes; failure of uro-rectal septum to separate the urogenital sinus anteriorly and ano-rectal canal posteriorly at fourth to sixth weeks of gestation, then the rupture of cloacal membranes between sixth and eighth

gestational week causing CE complex with other fetal deformities to adjacent structures (9, 10).

The spine's dysraphisms are related to abnormalities of the embryologic development occurring between the second and seventh weeks of gestation, starting from gastrulation between the second and third weeks, primary neurulation between the third and fourth weeks, and secondary neurulation. The association of CE with neural tube defects is very poorly understood. Still, there is a timeline overlap between the embryologic development of the two areas (10).

This severe congenital disability consists of the following: 1) omphalocele, 2) bladder exstrophy (two exstrophied hemi-bladders), 3) blind-ended foreshortened intestinal opening, usual cecum to anterior abdominal wall defect with imperforate anus, 4) separated pubic bones, and 5) splitting of the external genitalia and spinal defects. Diagnosis is possible during early gestation with antenatal ultrasound showing anterior abdominal wall defect with omphalocele and myelomeningocele and non-visualized bladder. This complex anomaly is easily identified clinically and requires a multi-disciplinary team approach.

In our case, the skin overlying the sac was of the mature epidermis, dermal appendages, and subcutaneous composition, contrary to reports of a thin epithelialized cap of tissue overlying the dome (4). In addition, we did not find any associated SCM, dorsal lipomas, or dermal parts inside the stalk suggestive of a dermal sinus tract. In the literature, the most common anomaly found in lumbar and thoracolumbar LDM was thickened filum. As with any tethering lesion, the severity of the neurological deficit increases with age, as grades 0–1 were the most common neurological grading in 5–10-year-old children (5).

4. Conclusion

CE is a rare condition that can be associated with LDM. Therefore, imaging the whole neuroaxis is important to rule out multiple lesions in these cases.

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Conflict of Interest

None.

Authors' Contribution

Abdulahman J. Sabbagh and Jamal S. Kamal contributed to the research idea; writing, drafting and revising the manuscript; and overall supervision. Rana H. Moshref contributed to data storage, writing, revising and editing the manuscript. Faisal Asiri, Abdulrahman M.R. Arafah and Abdulaziz Basurrah contributed to the manuscript writing and revision.

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