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Atlantoaxial limited dorsal myeloschisis: A report of two cases and review of literature

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ABSTRACT

Introduction: Limited dorsal myeloschisis (LDM) is a rare form of spinal dysraphism that is characterised by a distinctive fibroneural stalk connecting the spinal cord to the overlying skin lesion. The skin lesions associated with LDM can appear benign clinically and careful evaluation with an MRI scan is essential for diagnosing LDM and to differentiate this entity from other forms of spinal dysraphism and benign causes of skin lesions.
Research question: There is a lack of reported atlantoaxial LDM in the literature. We sought to report the clinical presentation, radiological features and surgical management of the first two reported atlantoaxial LDM.
Material and methods: Clinical findings and radiological images of the two cases of atlantoaxial LDM that underwent surgical intervention at our institution were retrieved from the medical notes, operative records and imaging system.
Results: Both cases of atlantoaxial LDM (C0-1 and C1-2 respectively) underwent successful resection of the overlying cutaneous lesions and stalks to release the tethered spinal cords.
Discussion: The surgical management of LDM have been associated with good outcomes and consists of resecting the fibroneural stalk close to the underlying cord, releasing the tethered spinal cord and removing the overlying cutaneous lesion.
Conclusion: These are the first two reported cases of atlantoaxial LDM in the literature. We aim to raise awareness of this pathological entity and highlight the importance of establishing the correct diagnosis to guide definitive management, and report the favourable neurological outcome in these cases despite the rostral location.

1. Introduction

Limited dorsal myeloschisis (LDM) is a rare form of spinal dysraphism characterised by a focal midline skin lesion and fibroneural pedicle or stalk that connects the spinal cord to the overlying skin lesion (Pang et al., 2013). This distinct clinicopathological entity was first described in 1993 and the persistent fibroneural pedicle has been hypothesised to be the result of an incomplete dysjunction between cutaneous and neural ectoderms (Pang et al., 2013). The surgical management of LDM involves disconnecting the fibroneural stalk that links the spinal cord and the dorsal cutaneous appendage in addition to untethering the spinal cord (Batista Cezar-Junior et al., 2020). Limited dorsal myeloschisis in the cervical region is a less common subgroup of LDM when compared to the thoracolumbar spine. In this article, we present two cases of LDM involving the atlantoaxial spine, which has not been reported in the literature to date.

2. Cases

2.1. Case 1

A 3-year-old girl was referred with a midline 'tail-like' cutaneous appendage at the nape of her neck that was present since birth (Fig. 1a). She was neurologically intact with no other stigmata of congenital anomaly. She had an MRI scan at the age of 24 months which demonstrated that the overlying cutaneous appendage was in continuity with the tethered spinal cord (Fig. 1b and c). She underwent surgical resection of the LDM and untethering of the spinal cord with continuous intraoperative neuro-monitoring of motor-evoked and somatosensory-evoked potentials at the age of 38 months. The fibroneural stalk from the base of

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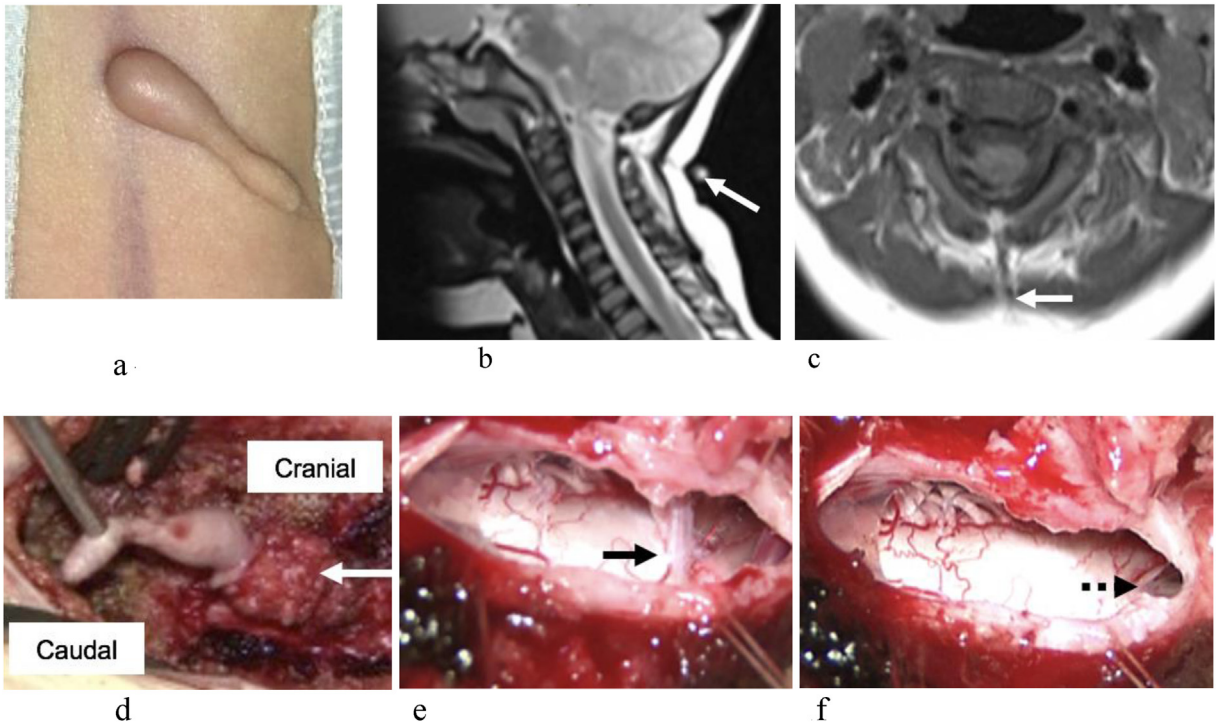

Fig. 1. a – 'Tail-like' skin appendage on the nape of the neck
Fig. 1b and c – Sagittal and axial MRI scan demonstrating the C1/2 limited dorsal myeloschisis with evidence of tethered cord. White arrow indicates the skin appendage from which the stalk (black arrow) is going through the soft tissue. Fig. 1d–f – Fibrous stalk connecting the cutaneous appendage to the underlying spinal cord (white arrow). The visible stalk (black arrow) was disconnected flush to the cord to release the tethered cord below the obex (dotted black arrow).

the appendage was followed through to the bony defect of the C2 lamina. Limited bony decompression was performed around the defect until normal dura was exposed. The dura was opened, and the fibrous tract was visualised to extend intradurally (Fig. 1d). The spinal cord was

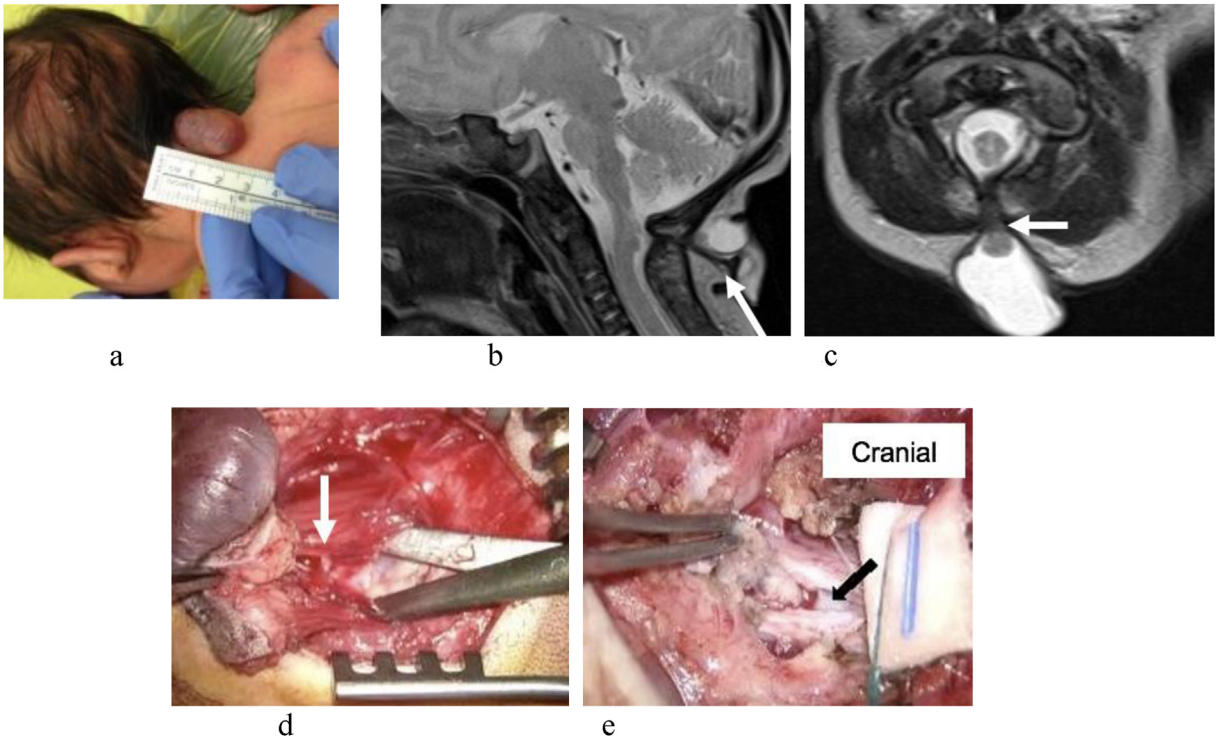

Fig. 2. a. Soft, pedunculated, transilluminable, mobile cystic swelling on the posterior aspect of the neck at the cranio-cervical junction.
Fig. 2b and c – Sagittal and axial MRI scan demonstrating the C0/1 LDM with associated tethering of the cord. Fig. 2d: and e – The overlying cutaneous lesion was connected to the deeper structures by a fibrolipomatous stalk. The deep portion of the stalk (white arrow) extended into the intradural space. Normal spinal cord (black arrow) was visualised between the dural opening cranial to the stalk.

tethered 3 mm inferior to the obex (Fig. 1e). The stalk was fully excised from the cord by sharp dissection and resected along with the cutaneous appendage (Fig. 1f). The tethered cord was carefully released, and the dura was closed in a watertight fashion. There were no compromise in motor-evoked and somatosensory-evoked potentials throughout the procedure. She remained neurologically well and was discharged on postoperative day four. Histopathological examination of the excised skin lesion and stalk was consistent with a benign fibroepithelial polyp and fibroconnective tissue devoid of acute or chronic inflammation.

2.2. Case 2

A newborn baby boy with normal antenatal scans was noted to have a 4 × 2cm, soft, pedunculated, transilluminable, mobile cystic swelling on the posterior aspect of the neck at the cranio-cervical junction (Fig. 2a). He was neurologically intact, and the rest of his spine was normal. On day one of life, an ultrasound of the swelling demonstrated a septated cystic mass with possible attachments to posterior cervical muscles and no deeper extension. On day three of life, he underwent an MRI scan which revealed a C0/1 LDM with associated tethered cord and an overlying sac measuring 11 × 27 × 21mm which contained meninges, CSF and fat (Fig. 2b and c). He subsequently underwent excision of the LDM and untethering of the cord on the 6th day of life. Intraoperatively, a fibrolipomatous stalk measuring 8mm in diameter that connected the cutaneous lesion to the underlying cord was identified. The stalk extended deep through a midline defect in the ligamentum flavum and dura (Fig. 2d and e). The dura was opened cranially and caudally to the stalk that was subsequently divided. The cutaneous lesion, a thin rim of normal skin and the superficial part of the stalk were removed *en bloc*. A near-total resection of the abnormal tissue was performed, with a small portion that was adhered to the cervico-medullary junction left *in situ*. The cervical cord was untethered. The patient had an uneventful postoperative course and was discharged 2 days later. Histopathological examination revealed neural tissue surrounded by fibrous tissue and blood vessels. He remains neurologically well at subsequent follow-ups.

3. Discussion

Limited dorsal myeloschisis (LDM) is a rare form of closed spinal dysraphism that can be associated with tethered cord, lipoma, lipomyelomeningocele, split cord malformation, teratoma and other congenital anomalies (Donovan and Pedersen, 2005; Park et al., 2005; Pang et al., 2010; Izci and Kural, 2019). The embryogenesis of LDM has been proposed to be secondary to incomplete fusion of the neural folds in primary neurulation resulting in incomplete separation between the cutaneous and neural ectoderm (Pang et al., 2010). This theory is supported by Pang et al.'s study whereby all 51 cases in their series had neural stalk attachment to the cord superior to the conus that forms during secondary neurulation (Pang et al., 2010). However, in a more recently published series by Kim et al., the authors reported that 11 out of 28 patients with LDMs had interspinous defect in levels below S1-2, which is formed during secondary neurulation. Kim et al. hypothesised that LDMs may occur during early stages of secondary neurulation due to incomplete dysjunction between the caudal cell mass and the skin or during later stages of secondary neurulation due to incomplete separation between the 'terminal balloon'(CSF-filled dilated caudal end of the secondary neural tube) and cutaneous ectoderm (Kim et al., 2020; Lee et al., 2013). LDMs are more commonly seen in the thoracolumbar spine when compared to the cervical spine (Pang et al., 2013; Izci and Kural, 2019; Chatterjee et al., 2021; Lafitte et al., 2018; Chatterjee and Rao, 2015; Mohindra, 2007; Lee et al., 2017). In Pang et al.'s series, all eleven cervical LDMs were located in the subaxial C3-7 levels (Pang et al., 2013). Our cases showed that LDM can occur in the atlantoaxial cervical spine, which have not been previously reported. In Case 1, there was a skin appendage, which perhaps be an external feature in its own right, because it does not fit into the external features of saccular LDMs described by Pang et al.'s (Pang et al., 2013). Case 2 was a typical saccular LDM consisting of one myelocystocoele but was unique in that it was the first C0/1 LDM. Both cases were diagnosed postnatally.

3.1. Proposed embryological explanation for rare occurrence

The reason for the rare occurrence of atlantoaxial LDM is unclear. While it is likely to be underdiagnosed and underreported because it is misdiagnosed as other cutaneous lesions, there may be other developmental and embryological explanations. Embryological studies have shown that human neural tube closure initiates at multiple sites (Nakatsu et al., 2000). The closure at the occipito-cervical region results from the joining of the closures from two initiation sites: Site A – cervical level, Site B – mesencephalic-rhombencephalic boundary (Fig. 3). We postulate that disruption of neural tube closure/dysjunction at the occipito-cervical region is less likely than other regions, because if the process fails at one of the two contributing initiation sites, the other process will still continue at the other site. However, if a neural tube defect does occur, it is likely to result in conditions much more severe than LDM, including spontaneous abortion, as described by Nakatsu et al. as Type III defects (Nakatsu et al., 2000).

3.2. Histopathological features of LDM

The histopathological examination of LDM stalk revealed glial fibrillary acid protein (GFAP)-immunopositive neuroglial tissues in 50% of cases (Morioka et al., 2019). Morioka et al. reported that peripheral nerve fibres were observed in all of the six fibroneural stalks examined and numerous melanocytes were found along the long-axis of the stalk in three out of four patients with dermal melanocytosis (Morioka et al., 2019). Dorsal root ganglion cells, fibrous bands, fat, skeletal muscles, blood vessels and dermoid or neuroenteric cysts have been detected surrounding the stalk and/or within the stalk (Pang et al., 2010). The presence of dermoid elements has the potential to cause neurological deterioration with development of dermoid cysts following partial resection of LDM stalk (Eibach et al., 2017).

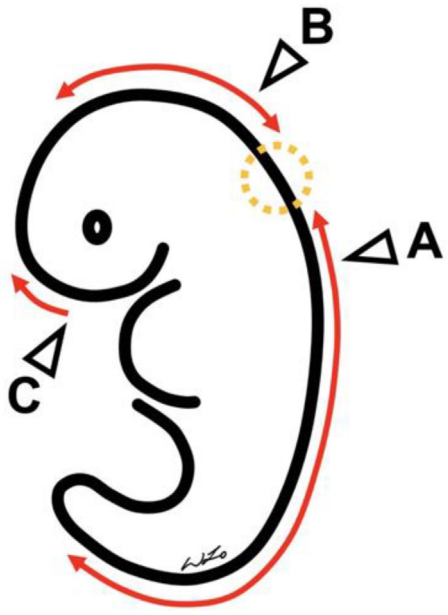

Fig. 3. Schematic diagram showing the current model for multi-site neural tube closure in human embryo. The *black line* represents the craniocaudal arrangement of the embryo. *Open triangles* indicate the three initiation sites: A – cervical level, B – mesencephalic-rhombencephalic boundary, and C – rostral tip of the neural groove. *Arrows* indicate the directions of neural tube closure. *Dotted circle* indicates the occipito-cervical region where the reported atlantoaxial LDMs are located. (Adapted from Nakatsu et al. (2000).

3.3. Antenatal radiological differential diagnosis of LDM

The antenatal diagnosis of LDM can often mimic myelomeningocele (MMC) and it is important to differentiate between these two entities due to the differences in prognosis (Friszer et al., 2017). In a prospective prenatal ultrasound study by Friszer et al., the authors found that diagnosis of MMC was revised to LDM in 7 out of 29 cases (Friszer et al., 2017). It is important to note that myelomeningocele is often associated with cerebral abnormalities such as hydrocephalus or Chiari malformation (Morais et al., 2020; Russell et al., 2013). It is essential to establish the correct diagnosis of LDM as missed LDM can lead to delayed neurological deficits, recurrent tethered cord syndrome and need for further surgical intervention (Chatterjee and Rao, 2015). LDMs are usually associated with a better prognosis in comparison to MMC and foetal MRI should be performed if there is any diagnostic uncertainty (Pang et al., 2013; Russell et al., 2013; Spoor et al., 2019). On the MRI scan, the fibroneural stalk or tract can usually be traced from the base of the skin lesion to the underlying cord (Pang et al., 2010).

3.4. Cutaneous stigmata of LDM

Skin tags are usually benign pedunculated lesions that can be found anywhere on the body. However, the presence of midline dorsal skin tags or neuroectodermal appendages is not always benign and should prompt further investigations to detect the presence of underlying spinal dysraphism (Guggisberg et al., 2004). Pang et al. elegantly described that the skin lesions associated with LDMs can be subdivided into four types namely saccular, crater, pit and membranous sac with saccular skin lesions being most commonly found in cervical LDMs (Pang et al., 2010). The 'tail-like' appendage described in Case 1 of our series has not been included in Pang et al.'s classification. Therefore, we propose an addition of 'appendage' to the four external features of the saccular subtype described by Pang et al.: thick squamous top, thin squamous top, dome pit, and membranous sac. There are other reported cases of 'tail-like' appendages in the literature (Batista Cezar-Junior et al., 2020; Sarukawa et al., 2019; Gaskill and Marlin, 1989; Cai et al., 2011; Canaz et al., 2018; Turk et al., 2016; Abe et al., 2020). By the 8th week of intrauterine life, the tail of the human embryos usually regresses. Failure of regression the distal remnant of the embryonic tail has been proposed to result in a persistent 'human-tail', more commonly located in the lumbosacral region (Klinge et al., 2020; Mukhopadhyay et al., 2012). Including our case, only two cases of 'tail-like' appendages in the cervical region have been reported in the literature to date (Mohindra, 2007) and the pathoembryological explanation for these occurrences are unclear.

3.5. Surgical treatment and neurological outcome

The surgical management of LDM have been associated with good outcomes and consists of resecting the fibroneural stalk flush/close to the underlying cord, releasing the tethered spinal cord and removing the overlying cutaneous lesion (Batista Cezar-Junior et al., 2020; Donovan and Pedersen, 2005; Pang et al., 2010; Nakatsu et al., 2000; Russell et al., 2013; Guggisberg et al., 2004; Sarukawa et al., 2019; Gaskill and Marlin, 1989; Cai et al., 2011; Canaz et al., 2018; Turk et al., 2016). It is essential to open the dura to expose the intradural component of the stalk and ensure that it is adequately disconnected from the cord. Based on Pang et al.'s series, seven out of eleven patients with cervical LDM presented with neurological deficit (Pang et al., 2013), which is in contrast to our cases of two young children, who were both neurologically intact at presentation. The difference may be explained by the young age of our patients (38-months and 6-days-old respectively), which means subtle neurological deficit or decline were not detected. Without releasing the tethered cord caused by the fibroneural stalk, Pang et al. reported that five out of six patients returned with progressive neurological deficits (Pang et al., 2010). We are in agreement that fibroneural stalk should be completely excised to minimise the risk of recurrence especially in the presence of any dermoid elements, although our series of two have not included cases of inadequate resection, and thus no direct comparison can be made (Eibach et al., 2017).

The surgical management of LDM at the atlantoaxial spine poses an additional risk of causing atlantoaxial instability as these levels are more heavily reliant on the ligaments and facet joints to prevent dislocation (Yang et al., 2014). Atlantoaxial instability is more common in the paediatric population due to the increased ligamentous laxity, more angled facet joints and incomplete ossification of the odontoid process when compared to the adult population (Lustrin et al., 2003). At the level of C1, the anterior arch usually completes fusion with the posterior neural arches by 7 years of age. At the level of C2, posterior fusion of the neural arches occur by 3 years of age and fusion of the neural arches with the body of the odontoid process occurs between 3 and 6 years of age (Lustrin et al., 2003). Therefore, careful bony exposure with preservation of the facet joints and lateral masses should be performed to maintain stability of the atlantoaxial spine. One should also be mindful of the close proximity of V3 and V4 segments of the vertebral arteries as they pierce the atlanto-occipital membrane and the dura to avoid vascular injury during the surgical approach.

Due to the tethering of the cord by the fibroneural stalk, the risk of neurological deficit increases over time with longitudinal growth of the spine, even though the change in the relative positions between the tethered cord and the overlying dura is modest in cervical cases (Pang et al., 2010). Urgent surgery should be offered in the presence of cerebrospinal fluid leakage and/or neurological deficit.

4. Conclusion

These are the two first reported cases of atlantoaxial LDM in the literature. We advocate that all dorsal cervical midline cutaneous lesions should be investigated further with MRI scan to look for evidence of spinal dysraphism and tethered cord. The presence of a stalk or tract between the cutaneous appendage and the spinal cord should be recognised pre-operatively and complete disconnection and resection of the fibroneural stalk should be performed to optimise surgical outcomes and prevent delayed neurological deterioration.

Conflict of interest

On behalf of all authors, the corresponding author states that there is no conflict of interest.

Consent

Verbal parental consents have been obtained for the publication of this manuscript.

Declaration of competing interests

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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