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Case Report

Mind the gap: an unusual case of a cervical lipomyelocele

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ABSTRACT

Cervical dysraphism is rare, and the 3 recognized subtypes manifest as cystic, skin-covered masses. To our knowledge, no case of cervical lipomyelocele has been reported in the literature so far. We present a case of surgically and pathologically confirmed cervical lipomyelocele in a patient with spondylocostal dysostosis and multiple other congenital anomalies and a brief review of the literature. In this case, magnetic resonance imaging demonstrates fat extension into a dysraphic cervical spinal canal, allowing for preoperative diagnosis. Computed tomography using 3-dimensional reconstruction serves to more clearly characterize the extensive spine malsegmentation characteristic of spondylocostal dysostosis. The use of this technique is suggested to benefit the orthopedic or neurologic surgeon confronted with such complex malformations.

Introduction

Spondylocostal dysostosis (SCDO) is a form of short-trunk dwarfism characterized by multiple vertebral and rib abnormalities. Other congenital anomalies may coexist, for which these patients should be thoroughly screened, and include congenital heart disease, genitourinary abnormalities, umbilical, diaphragmatic or inguinal hernias, Chiari malformation, and rarely neural tube defects [1,2]. Almost 400 cases of SCDO have been described, rarely with associated diaphragmatic hernias and neural tube defects; these 2 defects are, however, present in this patient [1]. To date, 18 cases associated with neural tube defects are described, 4 of which are terminal lipomyelomeningoceles [2]. We present a case of cervical lipomyelocele, which to our knowledge has not yet been described as a form of cervical dysraphism, in a case with typical features of SCDO.

Case report

A female with multiple congenital anomalies was born at 38-1/7 weeks gestational age to a 22-year-old, gravida 2, para

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0 (G2P0) mother. The mother had a history of first-trimester marijuana, cigarette, and alcohol use, which was stopped once she was aware of pregnancy and intermittent prenatal vitamin use. A first trimester and 24-week prenatal ultrasound were reportedly normal, and a quad screen was also reportedly negative. However, a 34-week prenatal ultrasound performed for preterm premature rupture of membranes was notable for findings suggestive of left congenital diaphragmatic hernia, pelvic kidney, and congenital heart disease.

After birth, physical examination revealed a very short neck with a reddish skin lesion over the upper cervical spine. The patient moved all extremities but favored her right arm over her left. Radiographs demonstrated cervical dysraphism with numerous cervicothoracic vertebral segmentation defects, and a small left chest with multiple absent and fused left ribs (Fig. 1). Genetic testing did not reveal an abnormality, but given her constellation of spinal segmentation and rib anomalies, a diagnosis of SCDO was made, along with pulmonary hypoplasia, congenital heart disease with a ventricular septal defect, bicuspid aortic valve, transverse aortic arch hypoplasia and severe aortic coarctation, pelvic horseshoe kidney, uterine didelphys, and inguinal hernia (with subsequent obstruction).

Magnetic resonance imaging (MRI) demonstrated mid-cervical spinal dysraphism with a dorsal soft-tissue mass containing linear bands of fat that insinuated through the dysraphic posterior elements. A fat-neural placode interface was present at the level of the spinal canal (Fig. 2). In addition, a separate cervical intradural and/or extramedullary cyst with mass effect on the cord, and T1 hyperintensity of the filum consistent with a filar lipoma, were also present (Figs. 3 and 4). MRI of the brain was normal.

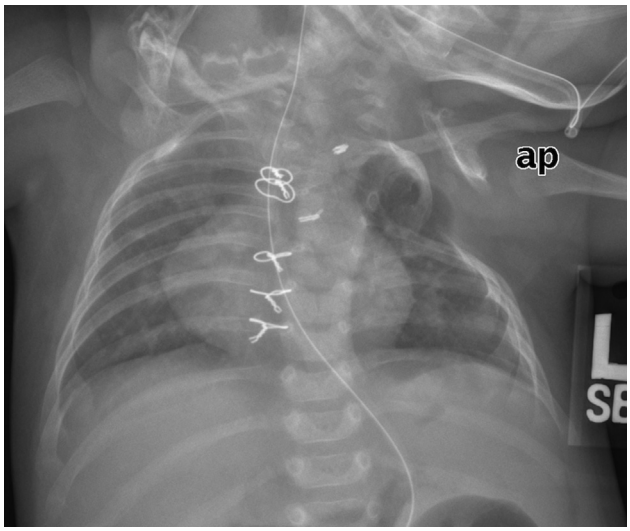

Fig. 1 – Chest radiograph demonstrates cervical spinal dysraphism with multiple cervical and upper thoracic segmentation anomalies. The left thorax is asymmetrically small with hypoplasia and fusion of the left first and second ribs, absence of the third and fourth left ribs and probable fusion of the left fifth and sixth ribs. Mediastinal clips and wires are present from hypoplastic aortic arch repair.

Surgical excision was performed at 6 months of age to address potential tethering and found a dural defect with subcutaneous fat protruding through the defect into the spinal canal. Surgeons also found and fenestrated, the subarachnoid cyst displacing the spinal cord. Pathology revealed a lesion lined by meningothelial-like tissue and containing ependymal canal and fibrous tissue, fat, skeletal muscle, and scattered meningothelial and neuroglial elements.

Cervical spine radiographs were deemed inadequate so cervical spine computed tomography with three-dimensional (3D) reconstruction was performed and demonstrated cervical scoliosis with numerous vertebral segmentations defects and fusion of nearly every level that progressed on follow-up computed tomography (Fig. 5). Her pulmonary function remained relatively normal, and no orthopedic correction of her spine or rib deformities was performed. At the age of 6 years, the patient was lost to follow-up when the family relocated but was doing well neurologically with the exception of left-sided hearing loss and a right-sided chin tilt.

Discussion

SCDO is a rare anomaly of the axial skeleton characterized by multiple vertebral and numerical or structural rib anomalies that result in thoracic asymmetry, short neck, and kyphoscoliosis [1–3]. Saul Jarcho and Paul Levin originally described this entity in 1938 as an entity distinct from Klippel–Feil syndrome. Since that time, five subgroups of SCDO have been recognized based on genetics and the Jarcho–Levin syndrome is considered SCDO1 [2]. Four genes with roles in the notch signaling pathway have been identified—*DLL3*, *MESP2*, *LNFG*, and *HES7*—, and inheritance is typically autosomal recessive, but autosomal dominant cases are also described [1,4].

Typical vertebral anomalies in SCDO are decreased number of vertebra, block, and wedge vertebra. Rib anomalies include numerical anomalies or structural anomalies such as broadening, bifurcation, or fusion. These abnormalities are often more advanced on one side of the thoracic cavity leading to asymmetry [1]. The thorax is shortened, but the limbs grow normally resulting in a characteristic short stature with limbs that appear long [1]. Cognitive function is reportedly normal in these patients [1]. Eighteen cases of associated neural tube defects are reported, but none are cervical in location [2].

Dysraphism of the cervical and upper thoracic spine is rare, and 3 variants are described which are distinctly different than our case of cervical lipomyelocele. These 3 types present as skin-covered, cystic masses overlying the cervical or upper thoracic spine and in decreasing order of prevalence, are: (1) meningocele with stalk (cystic mass containing a fibroglial stalk bridging the dorsal spinal cord and the posterior cyst wall); (2) myelocystocele (cystic mass containing the dorsal wall of a hydromelic spinal cord); (3) meningocele (cystic mass containing only cerebrospinal

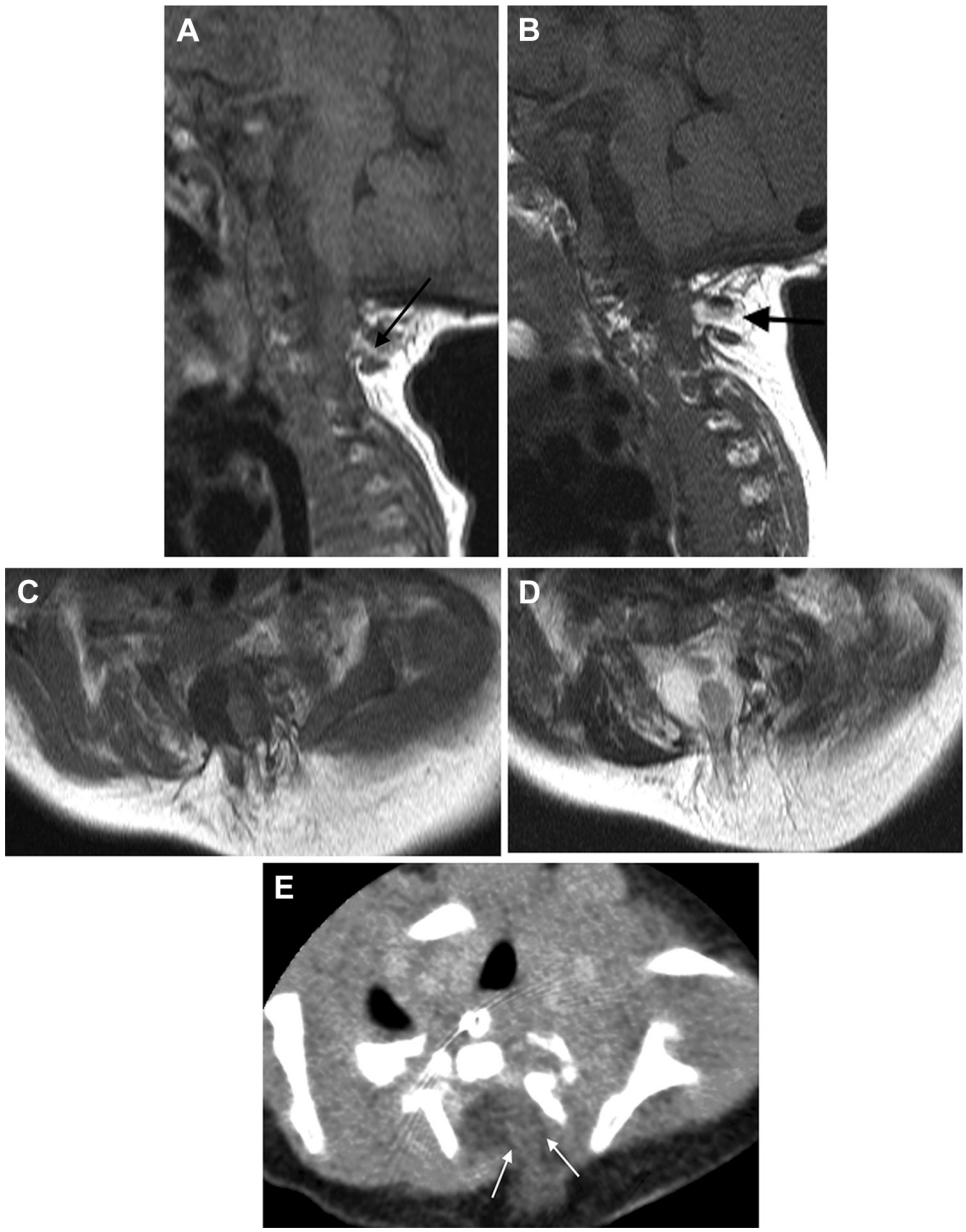

Fig. 2 – (A, B) Sagittal T1 MRI demonstrates posterior dysraphism and scoliosis. Along the dorsal dysraphic midcervical spine, there is a soft-tissue mass with linear T1-hyperintense bands (black arrow) representing fat that extend into the spinal canal. (C, D) Axial T1 and T2 MRI demonstrate widening of the spinal canal with T1-hyperintense tissue extending through the dysraphic posterior elements and dural defect. The neural placode-lipoma interface is at the level of the spinal canal. (E) Axial contrast-enhanced computed tomography shows low attenuation fat (white arrows) extending through the dysraphic posterior elements into the spinal canal. Note the displacement of the spinal cord to the left by a cystic structure in the spinal canal.

fluid) [5]. According to Steinbok and Cochrane's unifying hypothesis, the 3 types represent different manifestations along the spectrum of limited dorsal myeloschisis, and the final outcome is dependent on the presence or absence of hydromelia [6]. These differ from the much more common lumbosacral dysraphism in structure and have a better prognosis due to lack of functional neurologic tissue in the dysraphic sac, and absent or less severe intracranial anomalies [5]. Interestingly, although our case of cervical lipomyelocele is distinctly different from other cervicothoracic dysraphism subtypes, it also demonstrated a good neurologic outcome.

Spinal lipoma with dural defect encompasses both lipomyelocele and lipomyelomeningocele. It is an abnormality of primary neurulation whereby premature dysjunction of the cutaneous ectoderm from the neurectoderm allows

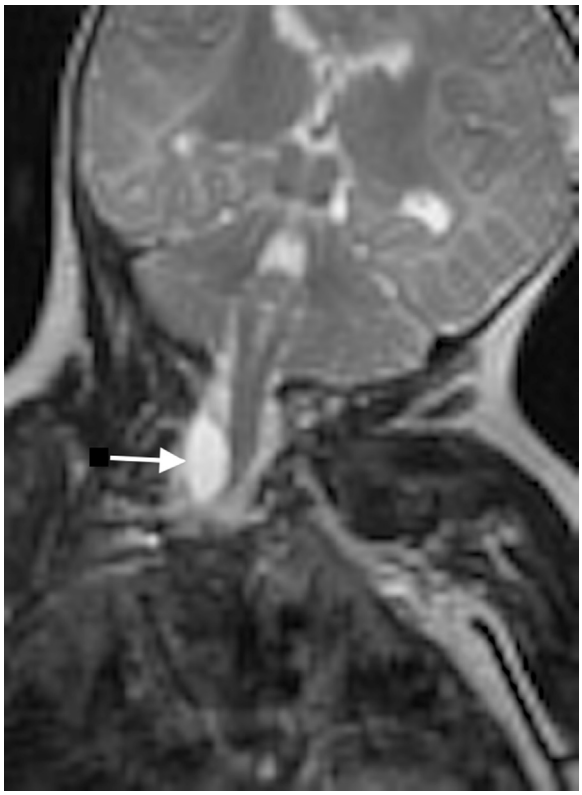

Fig. 3 – Coronal T2 MRI shows an isolated T2-hyperintense cyst (white arrow) in the right half of the spinal canal with mass effect on the cord. This subarachnoid cyst was fenestrated surgically.

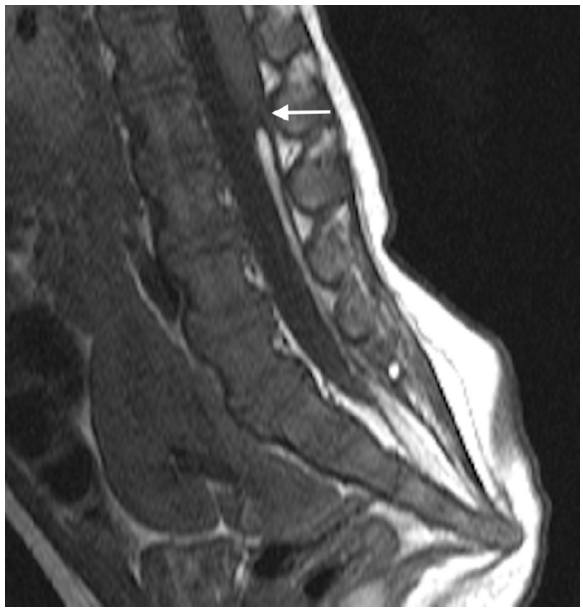

Fig. 4 – Sagittal T1 MRI shows abnormal T1 hyperintensity of the filum compatible with a filar lipoma. The conus terminates at L2 (white arrow). It is postulated that early errors during gastrulation may progress to involve primary and secondary neurulation, which may explain the presence of a filar lipoma in this case.

mesenchyme to enter the neural tube. Under the influence of the overlying dorsal neurectoderm, it differentiates into fat that prevents further neural tube closure. The lipomyelocele is more common demonstrating a neural-lipoma interface in or at the edge of the spinal canal. The lipomyelomeningocele is less common demonstrating a placode-lipoma interface dorsal to the spinal canal due to expansion of the subarachnoid space [7].

The association of costovertebral segmentation abnormalities with neural tube defects is plausible understanding that the neural tube stimulates somitic segmentation. A defect beginning early in gastrulation, during conversion of the bilaminar disc to a trilaminar disc, could progress to involve primary neurulation and thereby affect somitogenesis. In the case of a lipoma with dural defect, widening of the spinal canal from ingrowth of mesenchymal tissue laterally displaces the paraxial mesoderm containing the sclerotome to form the vertebra and ribs [8]. Disturbed somitogenesis then results in vertebral anomalies such as butterfly, hemi or block vertebra, and ribs are malformed or absent due to deficient sclerotome [2,3].

The excellent soft-tissue contrast of MRI is well suited to demonstrate the intraspinal fat extension of a lipomyelocele. The "placode" may be eccentric if premature dysjunction involves only one edge of the neural plate and may be irregular with stripes of fat permeating the spinal canal. The spinal canal may be expanded by the spinal lipoma, but the subarachnoid space ventral to the spinal cord should be normal [7]. Screening of the entire central nervous system with MRI is recommended for the classic types of cervical and upper thoracic dysraphism to identify intracranial abnormalities or additional sites of spinal cord tethering, and our case of associated filar lipoma would support this practice [5].

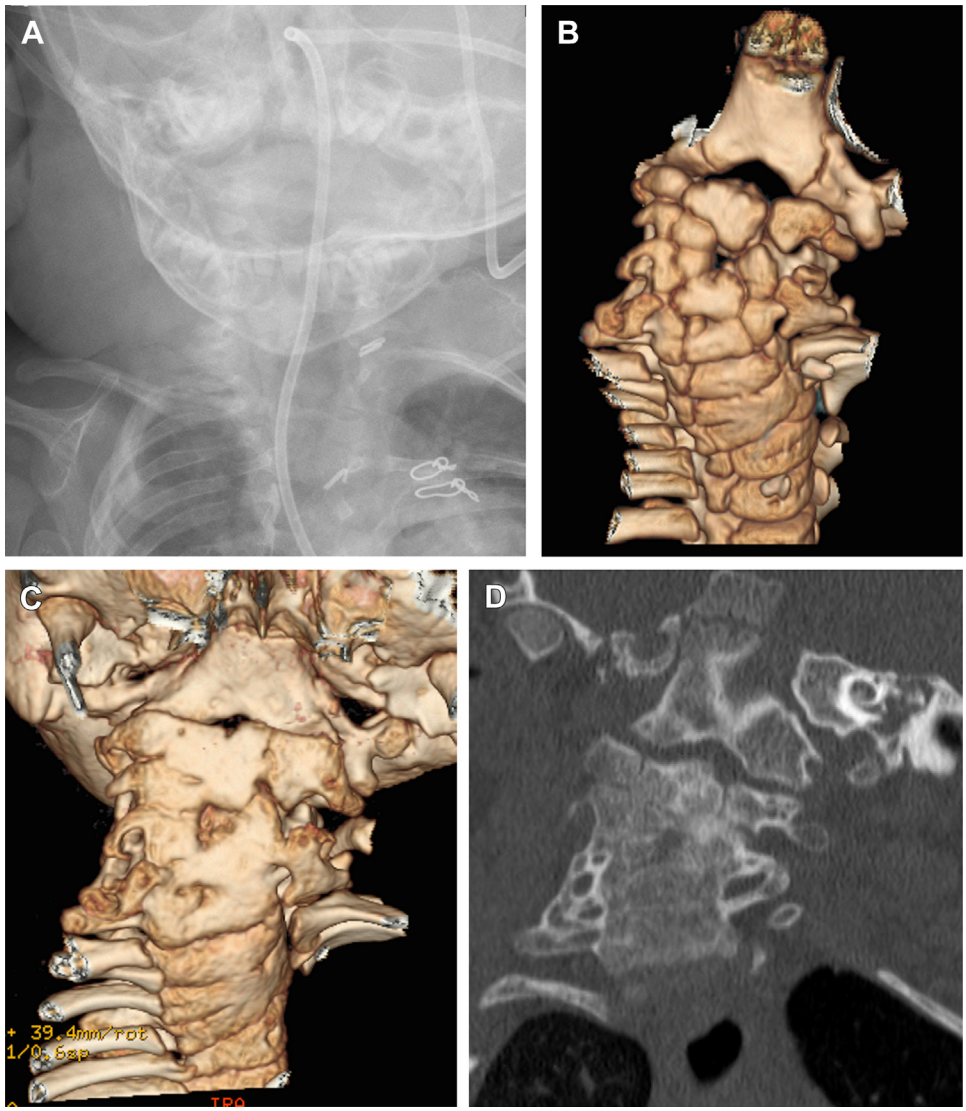

Fig. 5 – (A) Anterior-posterior cervical spine radiograph is of little diagnostic use given the markedly shortened neck and superimposition of the mandible. Not shown is the lateral view which was also limited by scoliosis and superimposition by the left scapula (Sprengel's deformity). (B) Three-dimensional reconstructions of cervical spine computed tomography performed at 22 months of age showing rotatory scoliosis and multiple segmentation anomalies including fusion of C1 and C2 and multiple midcervical and upper thoracic hemivertebra. A clear disc space is present between C2 and C3, but remaining levels are fused. Note multiple absent left ribs. (C, D) Three-dimensional computed tomography reconstruction and coronal image from cervical spine computed tomography at the age of 6 years shows progression of bony fusion, with a clear separation between C2 and C3 but confluent fusion of vertebral bodies and transverse processes throughout the rest of the cervical spine.

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