Case Report

Mind the gap: an unusual case of a cervical lipomyelocele

Natalie S. Valeur MD*, Ramesh S. Iyer MD, Gisele E. Ishak MD

Department of Radiology, Seattle Children’s Hospital, 4800 Sand Point Way NE, Seattle, WA 98105, USA

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.

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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...
was present at the level of the spinal canal (Fig. 2). In addition, dysraphic posterior elements. A fat-neural placode interface containing linear bands of fat that insinuated through the cervical spinal dysraphism with a dorsal soft-tissue massquent obstruction).

kidney, uterine didelphys, and inguinal hernia (with subse-

hypoplasia and severe aortic coarctation, pelvic horseshoe

ular septal defect, bicuspid aortic valve, transverse aortic arch

monary hypoplasia, congenital heart disease with a ventric-

anomalies, a diagnosis of SCDO was made, along with pul-

given her constellation of spinal segmentation and rib

clips and wires are present from hypoplastic aortic arch

probable fusion of the left fifth and sixth ribs. Mediastinal

sound performed for preterm premature rupture of mem-

were reportedly normal, and a quad screen was also

At the age of 6 years, the patient was lost to follow-up

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 ultra-

sound were reportedly normal, and a quad screen was also

reported negatively. However, a 34-week prenatal ultra-

sound performed for preterm premature rupture of mem-

branes 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 de-

fects, 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 pul-

monary hypoplasia, congenital heart disease with a ventric-

ular septal defect, bicuspid aortic valve, transverse aortic arch
hypoplasia and severe aortic coarctation, pelvic horseshoe

kidney, uterine didelphys, and inguinal hernia (with subse-

quent 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
consistently with a filar lipoma, were also present (Figs. 3 and 4).

MRI of the brain was normal.

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 neuro-
glial 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 neurologi-
cally 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 origi-
nally 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 iden-
tified—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 vertebrae, block, and wedge vertebra. Rib anom-

(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. 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.](image-url)
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...
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 lipomyelocoele is more common demonstrating a neural-lipoma interface in or at the edge of the spinal canal. The lipomyelomeningocoele 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 lipomyelocoele. 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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