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Case Report

Spondylocostal dysostosis with type II split cord malformation: A report of a rare case and brief review of the literature ☆

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ABSTRACT

Spondylocostal dysostosis (Jarcho Levin syndrome) is a rare costovertebral malformation syndrome that will result in restrictive pulmonary physiology. It manifests its major components at birth. Split cord malformation, together with spondylocostal dysostosis, is even rarer. We hereby report our experience with diagnosing 1 infant with spondylocostal dysostosis and type II split cord malformation using computed tomography and magnetic resonance imaging. We also present a concise summary of previously published case reports and case series involving patients with concurrent spondylocostal dysostosis and split cord malformations.

Introduction

Spondylocostal dysostosis (SCD) is a rare congenital disease of the ribs and spine [1]. It is also called Jarcho Levin syndrome and is different from spondylothoracic dysostosis (STD), a morphologically related but distinct entity [1,2]. SCD causes variable degrees of rib and vertebral anomalies [1–3]. The rib anomalies occur in an asymmetric manner, which allows for a more voluminous thoracic space and better respiratory function when compared to STD [2–4].

Spina bifida occurs in a substantial number of SCD patients, and this association is embryologically explainable [2,5,6]. Split cord malformations (SCM) in SCD patients are extremely rare, at the level of a small number of case reports and series. SCM are designated types I and II, depending on the presence and type of the splitting mesenchymal structure [5–7].

Abbreviations: CT, computed tomography; DLL3, Delta-like ligand 3; MRI, magnetic resonance imaging; SCD, spondylocostal dysostosis; SCM, split cord malformation; STD, spondylothoracic dysostosis; VEPTR, vertical expandable prosthetic titanium rib.

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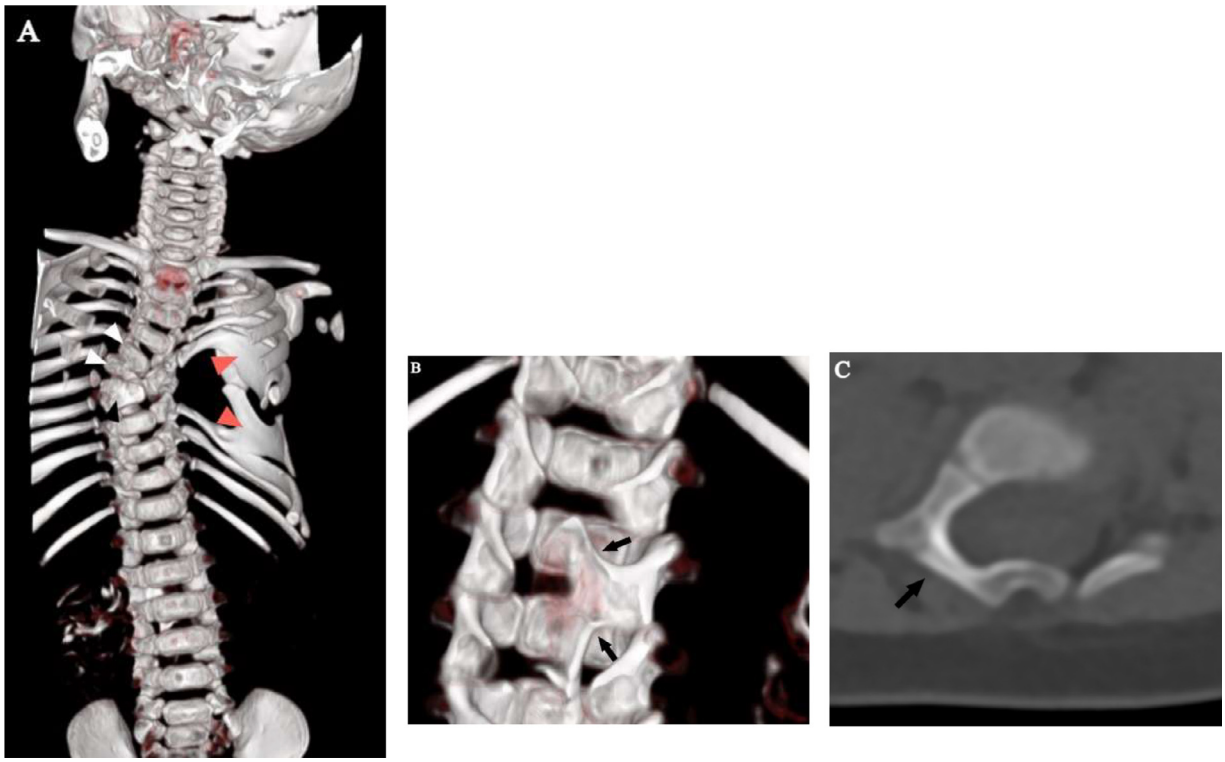

Fig. 1 – 3D volume rendered recontruction (A and B) of spinal CT shows thoracic dextroscoliosis with right side T5 and T6 (white arrowheads in A) hemivertebrae as well as right side fused T7-T8 (black arrowhead in A) vertebral bodies with unilateral hemivertebrae. On the left side, the 6th- 8th ribs are fused anterolaterally (red arrowheads in A). B and C depict bony fusion involving the right lamina of L1 and L2 (black arrows in B and C).

Treatment is done both by medical and surgical means. Early on, attention is given to respiratory failure and pulmonary infections. Various surgical methods also attempt to address the root cause of mechanical restriction from anomalous chest wall structures. SCM is primarily treated with surgical correction [8–10].

Clinical presentation

A 6-month-old female, a product of a non-consanguineous pregnancy, was born to a Para 3 mother. She was born by spontaneous vaginal delivery, without the need for resuscita-

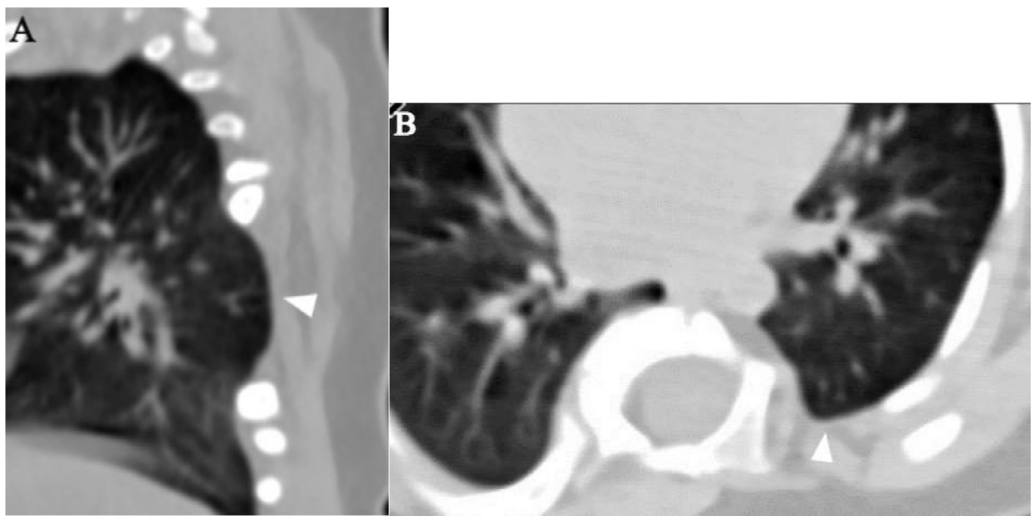

Fig. 2 – Sagittal (A) and axial (B) chest CT shows volume loss of the left lung following herniation (white arrowheads in A and B) through a posterior rib defect.

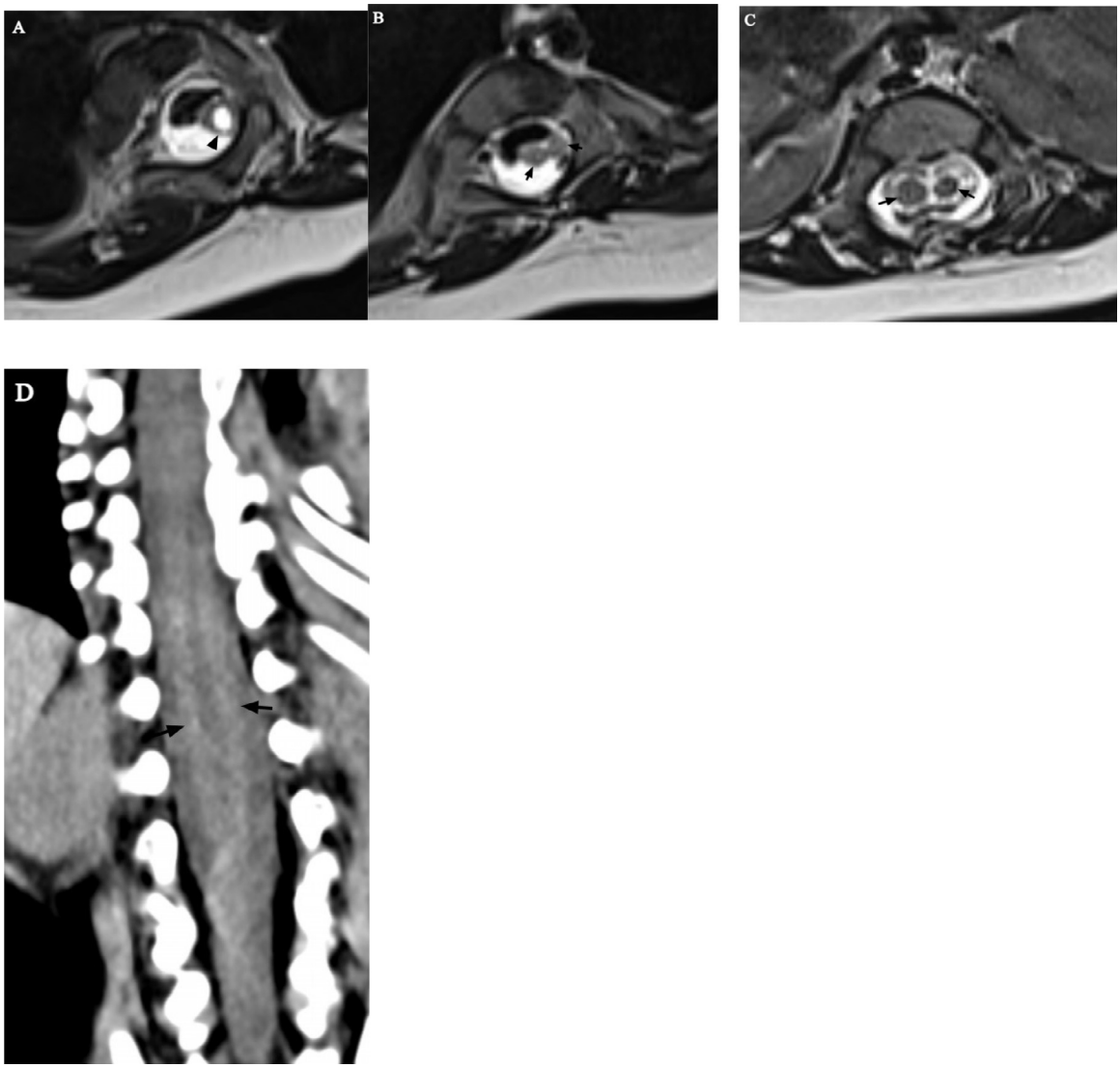

Fig. 3 – Cranial to caudal axial FSE T2W MRI (A-C) and coronal CT (D) clearly shows the 2 hemicords at the thoracolumbar level (black arrows in B-D) without a separating structure. There is also a focal syrinx proximal to the cord division at T9 level (black arrowhead on A).

tion treatments. When her parents noticed tufts of hair in the lower back region, they initially brought her to a local hospital, and she was subsequently referred to our center. There was no history of respiratory complaints, acute fever, or urinary incontinence. She also had no history of hospital visits or admissions before her current presentation. The patient's mother had no deformities, other chronic medical conditions, or history of teratogenic drug exposure. The patient's father and 2 other siblings were also free of any congenital conditions.

The baby had stable vital signs on physical examination, with a pulse rate of 100/min, a respiratory rate of 24/min, and a temperature (armpit) of 36.2 degrees Celsius. She cannot sit unsupported, but her other developmental milestones were normal. Except for mild chest wall asymmetry, signs of respiratory distress were absent. She showed a short neck, scoliosis, a patch of hair in the lower back region, and a mildly underdeveloped right leg with normal tone and power. A chest radiograph from the referring hospital reported curvature of the thoracic spine with multiple broad left side ribs. The imaging work-up continued with spinal computed tomography (CT) and magnetic resonance imaging (MRI). A 3D CT scan (Fig. 1) revealed dextroscoliosis and several rib problems on the left side, including fusion and absence defects. The types of vertebral abnormalities were hemivertebrae and anomalous fusion (Fig. 1). A rib defect caused a focal herniation of the left posterior lung (Fig. 2). MRI and complementary CT scans clearly showed 2 hemicords from T10–L4 levels with no midline bony spur and contained within a single dural covering and a focal proximal syrinx (Fig. 3).

After considering the imaging findings, a surgical correction of the SCM was proposed to the family. Unfortunately, due to financial constraints, the patient has not yet undergone the procedure. At the time of writing this document, the patient has not shown any worsening in her respiratory or neurologic conditions.

Discussion

Jarcho Levin Syndrome is a rare congenital anomaly that causes a spectrum of rib and spine anomalies. Associated cardiothoracic complications result in respiratory deficiency and recurrent lung infections [22].

A review by Berdon and colleagues [1] analyzed the original description of SCD by Jarcho and Levin from 1938 and made a distinction from STD, first described in 1966 by Levy and in 1969 by Moseley. SCD is marked by scoliosis, asymmetric rib anomalies, and multiple vertebral deformities. It is linked to the Delta-like ligand 3 (DLL3) gene on chromosome 19, which is associated with the Notch gene signal pathway. In STD, the thoracic spine is fused, and all the ribs are fused posteriorly, giving rise to a symmetric fan-like or "crab" configuration. The DLL3 mutation has not been found in STDs.

A study that reviewed over 1.4 million live births registered by the Spanish Collaborative Study of Congenital Disorders found the rate to be 0.2/100,000 [23]. It has been commonly reported in females [24,25]. SCD has been reported in both consanguineous and non-consanguineous marriages [10]. The majority of cases have autosomal recessive inheritance, and these will have a more severe disease compared to the few autosomal dominant cases [26].

Newborns with SCD have a short neck and trunk, a protuberant abdomen, and long limbs. Previous studies have reported facial dysmorphism in a small number of cases [1,3,14,24]. The most common vertebral anomaly is the hemivertebra, which is commonly seen in the thoracolumbar segment [3,10]. Approximately 75% of cases exhibit scoliosis [10]. Intrinsic and asymmetric rib anomalies include fusion (near the costovertebral junction), absence, orientation, shape, and size malformations [10,22]. Typically, the rib anomalies do not exhibit the fan-like orientation originally classified by Solomon et al. In some cases, a quandary arises as patients present with fan-like rib orientations of intrinsically deformed ribs, and in such cases, they should be classified as SCD [3].

SCD is more common than STDs and is more compatible with life [4,10,26]. This disparity is mainly due to the severity of thoracic complications in the 2 groups [22,26]. But these patients still suffer from recurrent pulmonary infections and have a poor quality of life [10]. Other congenital anomalies associated with SCD can be genitourinary, cardiac,

Table 1 – Summary of cases with concurrent SCD and SCM.

Author	Year	Patient	Type of SCM	CNS or extra- CNS associations (scoliosis not included)
Our case		6 months old, female	II	Syringomyelia, Laminar Fusion
Sura and colleagues [11]	2023	7-year-old, female	II	Spina bifida, Epidermoid cyst
Das and colleagues [12]	2022	9-month-old, female	I	Syringomyelia, Aneurysmal dilation of the right posterior spinal artery
Milić and colleagues [13]	2019	3-year-old, female	I	Partial sacral agenesis, Thoracic rachischisis, Posterior neural arch defects, Meningocele, Spinal lipoma, duplicated and thick filum terminale, Syringomyelia, foot deformities
Padma and Sundaram [14]	2015	Newborn, male	I	Facial dysmorphism (low set ears, hypertelorism, high arched palate), Hypospadias with chordee and penile torsion, Imperforate anus, Sacral agenesis, Calcaneovalgus, Overriding feet, Sacral agenesis, Tethered cord
Alatas and colleagues [15]	2015	7 cases (age and sex not stated)	Not specified	1 Tethered cord, Syringomyelia, Mega cisterna magna 2 Myelomeningocele 3 No concurrent CNS anomalies 4 Myelomeningocele 5 Chiari malformation, Myelomeningocele 6 Syringomyelia, Myelomeningocele 7 Hydrocephalus, Chiari malformation, Tethered cord, Myelomeningocele
Muthukumar [9]	2015	7 months old, female	I	Spina bifida, Syringomyelia, Tethered cord, Imperforate anus
Kim, Young-Seon and colleagues [16]	2015	37 years old, female	I	Spina bifida
Srinivas and colleagues [17]	2014	8 years old, female	II	Abdominal wall defect, Tethered cord, Spina bifida
Yilmaz and colleagues [4]	2013	2 years old, female	I	Double right nipples, Tethered cord
Kansal and colleagues [18]	2011	1.5 – years old, male	I	Tethered cord, Spina bifida
Saini and Singh [19]	2010	22 days old, male	I	Lipomyelocele
Etus and colleagues [5]	2003	7-year-old, female	I	Spinal lipoma, Tethered cord, Syringomyelia
Giacoia and Say [6]	1991	Newborn, male	I	Aqueductal stenosis, Cleft palate, Atrial septal defect, Imperforate anus, Undescended testes
Reyes and colleagues [20]	1989	Newborn, female	I	Frontal bossing with wide nasal bridge, Talipes equinovarus, Shallow posterior fossa
Eller and Morton [21]	1976	Newborn, female	I	Lacunar skull, Thoracolumbar rachischisis, Talipes equinovarus, Persisting urachus, Rectovaginal fistula (Lysergic acid diethylamide-LSD exposure present)

gastrointestinal, hernias (diaphragmatic, umbilical, inguinal), central nervous system, and cutaneous in nature [2,4,26].

Spina bifida occurs in 40.6% of patients with SCD, and the association does not appear to be accidental [5,6]. Others have also reported this important association [2,27–29]. Based on a review of 41 cases, Pang and colleagues [7] proposed using SCM to replace the customary terms diastematomyelia and diplomyelia. According to the new proposal, these two types are considered a result of a common embryologic aberration, and the morphologic changes represent variations of a common entity. Hence, type I SCM is defined as consisting of 2 hemicords, each residing within its own dural tube and separated by a dura-covered, rigid osseocartilaginous median septum. Type II SCM also consists of 2 hemicords contained in a single dural tube separated by a nonrigid, fibrous median septum. In the same year, Dias and Walker [30] proposed disruption of one or more of the 3 germ cells during gastrulation as the primary event to explain multiple complex spinal dysraphic anomalies, as well as secondary failure of neural tube closure to account for the associated overt and occult neural tube malformations.

The association of double cord anomalies with SCD is rare. We discovered 21 cases from 1976 to 2023 (Table 1). Except for 2 cases [11,17], the rest had type I SCM. To the best of our knowledge, our case is the third reported case of SCD with type II SCM.

In the largest case series of SCD with concurrent CNS abnormalities to date, Alatas and colleagues [15] reviewed 28 cases and documented associated central nervous system abnormalities. SCM was diagnosed in 7/28 cases, and 6 of these occurred together with other CNS anomalies. 1 case lacked additional CNS anomalies. The type of SCM in the 7 cases was not specified. In one article [16], type I SCM was associated with SCD in a 37-years-old female patient. This case was unique as it showed the life span that can be reached with this complex abnormality.

The main focus of medical care in SCD is the respiratory system, which suffers from recurrent infections and respiratory insufficiency [22]. Multiple types of chest wall surgeries are performed in the hopes of expanding the thoracic cavity and continuing pulmonary development [10]. SCMs are treated with surgical resection of the spur, release of arachnoid adhesions, and tethering, as well as sectioning of associated anomalies such as fatty filum terminal and sinus tracts, are done [9]. Vertical Expandable Prosthetic Titanium Rib (VEPTR) expansion is used in patients with congenital scoliosis and fused ribs, which aims at improving cervical tilt as well as head and truncal alignment [31].

In conclusion, the association of SCD with SCM, especially type II, is very rare. Well-coordinated, long-term care involving both medical and surgical care will be required. Radiology is required for an accurate pre-operative assessment.

Patient consent

Written informed consent was obtained from the patient's parents for anonymized information to be published in this article.

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