

Case Report

Spondylocostal dysostosis: A rare case report

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ABSTRACT

Spondylocostal dysostosis (SCD), also known as Jarcho Levin syndrome, is a rare congenital dysostosis characterized by multiple congenital vertebral and rib anomalies. The child usually presents with respiratory distress at birth with eventual development of short trunk dwarfism. Associated congenital anomalies are rare; however, they need evaluation. We report a 6-month-old Indian girl with SCD who presented with respiratory distress and an asymmetrical small thoracic cavity. The diagnosis was made with radiographs, computed tomography scans, and magnetic resonance imaging (MRI) showing multiple rib and vertebral segmentation anomalies. We would also highlight the importance of MRI in these patients and illustrate other associated brain and spine anomalies.

Keywords: Spondylocostal dysostosis, Congenital, Vertebral, Rib anomalies, Magnetic resonance imaging

INTRODUCTION

Spondylocostal dysostosis (SCD) is a rare skeletal dysplasia, first described by Jarcho and Levin in 1938.^[1] Multiple congenital vertebral and rib anomalies, short neck and trunk, abnormal segmentation of the spine and ribs, and normal-length limbs manifest the disease condition. The anomalies include overgrown, fused or missing ribs and segmentation defects of the vertebrae such as fusion, hemivertebrae, or block vertebrae. Other associated congenital anomalies are rare; however, they need evaluation.^[2] The exact prevalence of SCD is not well-established, but it is considered an extremely rare condition.^[3] Here, we describe the clinical presentation and imaging characteristics of a 6-month-old female child with SCD who presented with respiratory distress at birth.

CASE REPORT

A 6-month-old female child was referred to the pediatric clinic with a history of respiratory distress since birth. The child was born at full-term through an elective cesarean section through an uneventful pregnancy and delivery. There was no significant family history of consanguinity among the parents. The parents reported rapid breathing and retractions in the first hours of life. The thorax appeared asymmetrical and showed limited chest expansion during inspiration, with evident retractions of the left hemithorax with each breath and increased respiratory effort [Figure 1]. The child's vital signs were within the normal range, except for tachypnea. On auscultation, reduced breath sounds were noted over the left hemithorax, suggesting hypoventilation. Palpation of the thorax revealed mild scoliosis and rib abnormalities. These clinical features raised suspicion for congenital skeletal dysplasia. The child was sent to the Department of Radiology for further evaluation.

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Radiological investigations were performed to assess the skeletal anomalies. Antero-posterior and lateral radiographs of the thorax and spine were obtained. The X-rays revealed abnormalities of the ribs and thoracic spine and mild thoracic dextroscoliosis. There was an absence of the left 4th–6th ribs. Furthermore, there were segmentation defects of the thoracic vertebrae in the form of hemivertebrae [Figure 2]. The vertebral and rib anomalies were consistent with the diagnosis of SCD. In addition, there was dextroposition of the heart. The rest of the skeletal survey revealed no abnormalities.

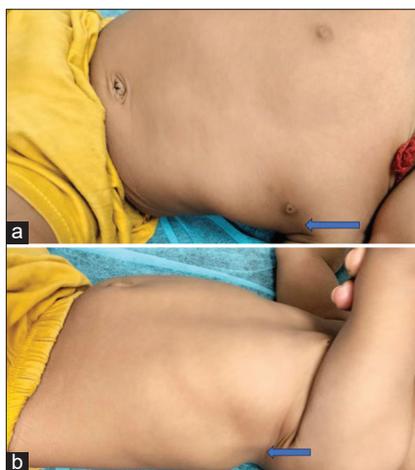


Figure 1: (a and b) Clinical pictures of the child show an asymmetrical small thorax with a clinically palpable defect in the left-sided chest wall (blue arrow).



Figure 2: Anteroposterior X-rays of the thorax and spine show abnormalities of the ribs and thoracic spine with mild thoracic dextroscoliosis. Note the absence of the left 4th–6th ribs and segmentation defects of the thoracic vertebrae in the form of hemivertebrae.

A computed tomography (CT) scan of the thorax and spine was performed to obtain more detailed imaging of the skeletal and, if any, associated cardiac abnormalities. The CT images confirmed thoracic dextroscoliosis and the presence of hemivertebrae with asymmetrically fused vertebral bodies. However, no bony spinal canal stenosis was noted. There was an absence of the left 4th–6th ribs with abnormally short and fused ribs, contributing to the thoracic cage deformities observed on clinical examination. In addition, there was a dextroposition of the heart; however, no congenital cardiac anomalies were seen [Figure 3].

MRI of the brain and spine was done to rule out associated congenital anomalies. It showed a low lying and tethered cord with filum terminale lipoma. There was associated dorsolumbar short segment syrinx and sacral spinal meningeal cysts type III (arachnoid cysts) [Figure 4].

DISCUSSION

SCD is characterized by abnormal segmentation and formation of the spine and rib. There is resultant mild congenital scoliosis with moderate thoracic insufficiency, and most of the affected children survive till adulthood. It usually shows autosomal recessive inheritance and is associated with DLL3 gene mutation. The main clinical and radiological differential diagnosis is spondylothoracic dysostosis (Lavy–Moseley syndrome).^[3] This can be differentiated with the help of certain imaging features [Table 1].

SCD and its association with neural tube defects have been frequently reported in the literature. It has been postulated that the vertebral anomalies observed in this syndrome are secondary to a developmental disturbance between the 4th and 8th weeks of gestation, as multiple centers of chondrification form at that time around the notochord to form a complete cartilaginous pro-vertebra. Failure of these chondrification centers to develop or unite can result in hemivertebrae or butterfly vertebrae with anomalous rib development secondary to anomalous vertebral development.^[4]

There are multiple surgical treatments described for SCD, such as vertical expandable prosthetic titanium ribs (VEPTR) and chest wall reconstruction with latissimus dorsi flap transfers for thoracic insufficiency. The treatment depends on the degree of thoracic insufficiency, concurrent pulmonary complications, and chest wall defects aimed at improving quality of life. In patients with chest wall defects such as herniation of thoracic contents or paradoxical chest wall movements during respiration, the use of a latissimus dorsi flap improves respiratory function and decreases the rate of infections. VEPTR manages the issue of thoracic insufficiency better than the latissimus dorsi flap. The goal of VEPTR is to improve scoliotic curves and expand thoracic volume, hence helping to create space for lung development. In VEPTR, one or more vertically

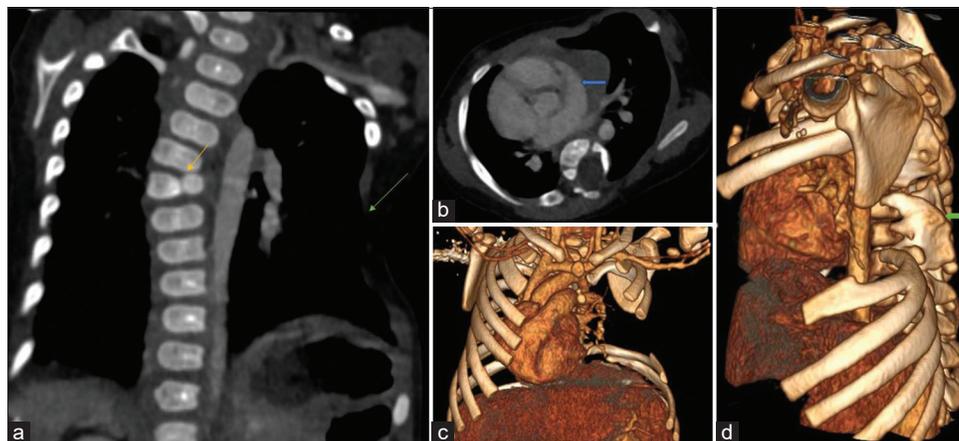


Figure 3: Computed tomography (CT) scan of the thorax and spine. (a) Showing the absence of the left 4th–6th ribs leading to the thoracic cage deformity (green arrow) and vertebral anomalies (yellow arrow). (b) Showing dextroposition of the heart. (c and d) 3D CT images confirming multiple rib and vertebral anomalies (green arrow).

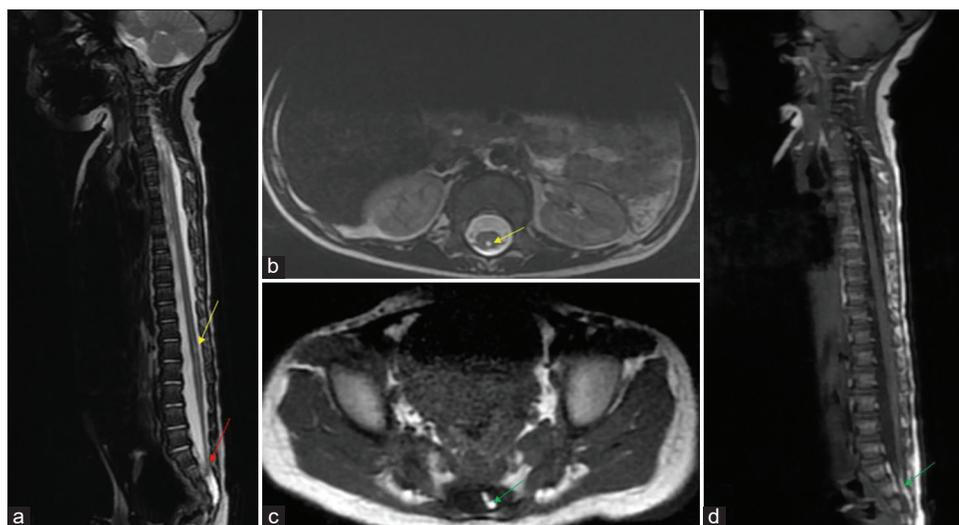


Figure 4: Magnetic resonance imaging of brain and spine. (a and b) T2W sagittal and axial images showing low-lying and tethered cord (red arrow) with associated dorso-lumbar short segment syrinx (yellow arrow), (c and d) T1W axial and sagittal images showing filum terminale lipoma (green arrow).

Table 1: Differential diagnosis.

	Spondylocostal dysostosis	Spondylothoracic dysostosis
Rib anomalies	Fused, missing, or overgrown ribs	Posterior fusion of ribs (Shows crab-like configuration)
Spine anomalies	Fusion, hemivertebrae, or block vertebrae	Cervicothoracic fusion or fusion of occiput and C1
Associated congenital anomalies	Rare; however, neural tube defects can be seen	More common, including neural tube defects, anorectal malformations
Distribution	Worldwide	50% from Puerto Rico
Clinical course	Most survive into adulthood	High infant mortality rate

oriented distracting devices are attached superiorly to the ribs and inferiorly to the ribs, spine, or pelvis and are

serially lengthened as the child grows. However, close clinical follow-up with these patients is recommended,

particularly early on in life, considering the high rates of infant mortality observed. The ideal time to operate should maximize growth potential and minimize respiratory morbidity. This time frame remains undetermined. If a significant decline in function and respiratory status or failure to thrive is observed, then surgical intervention with VEPTR should be considered.^[5]

CONCLUSION

This case report highlights the importance of considering SCD in a child with small thorax and respiratory distress. Cross-sectional imaging plays a crucial role in better delineating the osseous and other associated craniospinal anomalies. Restrictive lung disease is the key factor determining the quality of life and span, thus requiring apt management.

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Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

REFERENCES

1. Jarcho S, Levin PM. Hereditary malformations of the vertebral bodies. *Bull Johns Hopkins Hospital*. 1938;62:216-26.
2. Prabhakar N, Patel B. Jarcho-Levin syndrome: A rare case of thoracic insufficiency syndrome in a newborn. *Indian Pediatr Case Rep* 2023;3:171-3.
3. Berdon WE, Lampl BS, Cornier AS, Ramirez N, Turnpenny PD, Vitale MG, *et al.* The clinical and radiological distinction between Spondylothoracic dysostosis (Lavy-Moseley Syndrome) and Spondylocostal dysostosis (Jarcho-Levin syndrome). *Pediatr Radiol* 2010;41:384-8.
4. Muthukumar N. Spondylocostal dysostosis (Jarcho-Levine syndrome) associated with occult spinal dysraphism: Report of two cases. *J Pediatr Neurosci* 2015;10:127-32.
5. Southam BR, Schumaier AP, Crawford AH. Spondylocostal dysostosis: A literature review and case report with long-term follow-up of a conservatively managed patient. *Case Rep Orthop* 2018;2018:1795083.

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