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# Unraveling a Congenital Thoracic Deformity: CT Diagnosis of Spondylocostal Dysostosis in a Newborn

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## ABSTRACT

Spondylocostal dysostosis is a rare congenital disorder characterized by multiple vertebral segmentation anomalies and rib malformations, often resulting in thoracic deformity and scoliosis. We report the case of a newborn presenting with thoracic deformity, investigated by thoracic computed tomography (CT). Imaging revealed bilateral costotransverse synostosis involving multiple ribs, associated with vertebral malformations including butterfly vertebrae at D4 and D5, a single right hemivertebra at D2, and dorsal scoliosis with right convexity. No associated pulmonary, mediastinal, abdominal, or suspicious osseous lesions were identified. CT imaging allowed comprehensive assessment of the costovertebral anomalies and exclusion of associated complications. This case highlights the key radiological features of spondylocostal dysostosis and emphasizes the crucial role of CT in diagnosis, anatomical mapping, and early management planning.

## KEYWORDS

Spondylocostal dysostosis, Costovertebral malformations, Newborn, Thoracic deformity, CT imaging

# MAIN ARTICLE

## INTRODUCTION

Spondylocostal dysostosis is a rare congenital skeletal dysplasia characterized by abnormal vertebral segmentation and rib anomalies. It belongs to the spectrum of vertebral segmentation defects and is often diagnosed in the neonatal period due to thoracic deformity or respiratory compromise. Early and accurate imaging is essential to characterize the extent of anomalies, guide clinical management, and evaluate potential associated abnormalities [1,2].

## CLINICAL INFORMATION

A newborn was referred for imaging evaluation due to an abnormal thoracic contour noted at birth. No signs of respiratory distress were observed, and the clinical examination was otherwise unremarkable.

An initial **plain chest radiograph** demonstrated abnormal rib alignment and suspected vertebral anomalies; however, the findings were inconclusive, and precise characterization of the costovertebral abnormalities was not possible. Given the diagnostic uncertainty, a **thoracic CT scan** was performed for further evaluation.

## IMAGING FINDINGS

Thoracic computed tomography demonstrated the following findings:

Presence of twelve ribs bilaterally.

Multiple osseous bridges were identified between the ribs and the costotransverse joints, consistent with **bilateral costotransverse synostosis** (Figure 1) :

- **Right side:** posterior rib arches from K1 to K5, K6 to K9, and K10 to K11 (Figure 2).
- **Left side:** posterior and middle rib arches from K1 to K8, and posterior arches of K9 and K10 (Figure 2).

Associated vertebral anomalies included (Figure 3) :

- Butterfly vertebrae at D4 and D5.
- A single right hemivertebra at D2.
- Dorsal scoliosis with right convexity.

## DISCUSSION

Spondylocostal dysostosis represents a spectrum of congenital vertebral and rib segmentation defects resulting from abnormal embryologic development of the axial skeleton. Unlike spondylothoracic dysostosis, which is characterized by posterior rib fusion and a “crab-like” thorax, spondylocostal dysostosis typically presents with asymmetric rib anomalies and variable vertebral malformations [2,3,4].

Clinically, affected newborns often present with visible thoracic deformity. The degree of respiratory involvement varies and depends largely on the severity and extent of rib fusion and thoracic cage restriction. In the present case, the absence of respiratory compromise suggests a milder phenotype, although long-term respiratory and orthopedic follow-up remains essential.

From an imaging perspective, plain radiography is usually the first diagnostic modality but may be limited in assessing complex costovertebral anatomy, particularly in neonates. CT offers superior spatial resolution and allows precise evaluation of rib number, morphology, costotransverse relationships, vertebral segmentation anomalies, and spinal alignment. CT is therefore the imaging modality of choice for confirming the diagnosis and mapping the full extent of skeletal involvement. MRI may be considered in selected cases to evaluate the spinal cord or detect associated intraspinal abnormalities [2,4].

### Genetic Correlation

Spondylocostal dysostosis is genetically heterogeneous and most commonly inherited in an autosomal recessive pattern. It is associated with mutations in genes involved in the Notch signaling pathway, which plays a critical role in somitogenesis. Reported pathogenic variants include *DLL3*, *MESP2*, *HES7*, *LFNG*, and *TBX6*. These mutations lead to abnormal vertebral segmentation and rib development. Although genetic testing was not performed in the present case, recognition of the characteristic radiological phenotype should prompt consideration of

genetic counseling and molecular analysis, particularly in familial cases or when additional anomalies are present. Identification of the genetic etiology may aid in prognostic assessment and recurrence risk evaluation.

The absence of associated visceral or pulmonary anomalies in this case suggests a relatively isolated form of spondylocostal dysostosis. Early diagnosis of spondylocostal dysostosis allows appropriate multidisciplinary management, including pediatric, orthopedic, respiratory, and genetic follow-up, to monitor spinal deformity progression and respiratory function [1,3,4].

## CONCLUSION

This case demonstrates the diagnostic value of thoracic CT in a newborn with thoracic deformity and inconclusive plain radiographic findings. CT imaging confirmed the presence of characteristic costovertebral malformations consistent with spondylocostal dysostosis, including bilateral costotransverse synostosis, butterfly vertebrae, a single hemivertebra, and dorsal scoliosis. Comprehensive imaging assessment is essential for accurate diagnosis, exclusion of associated anomalies, and early clinical and genetic management planning.

## FIGURES:

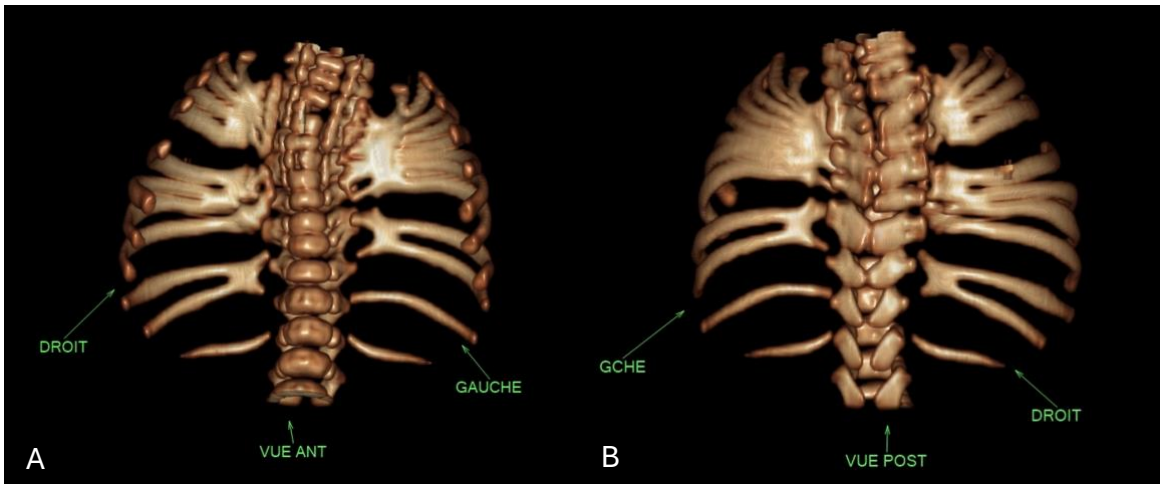


*Figure 1: Coronal CT reconstructions demonstrating bilateral costotransverse synostosis with multiple osseous bridges between the ribs and transverse processes, shown in anterior (A) and posterior (B) views.*

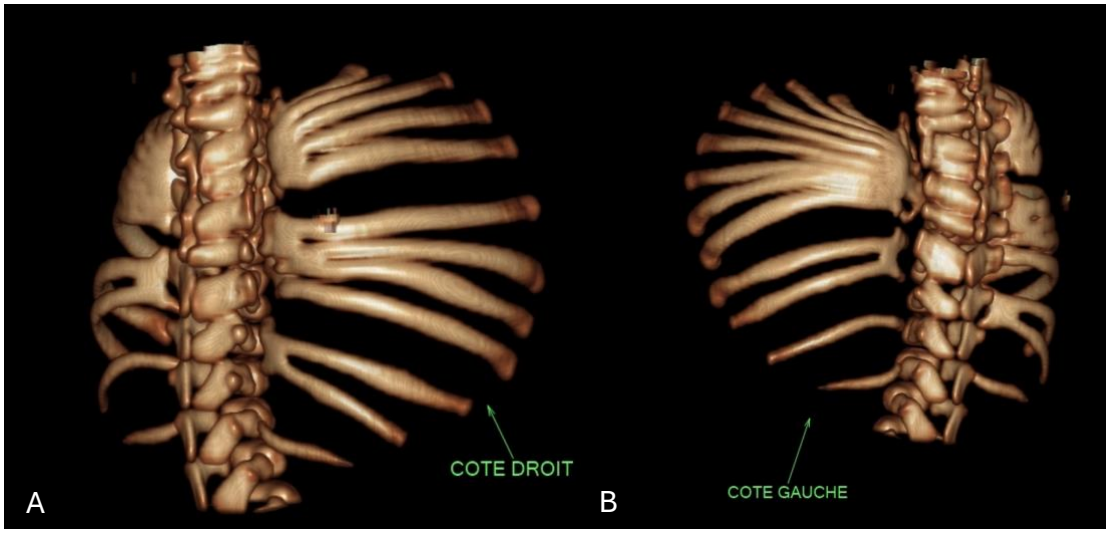


*Figure 2 : Sagittal CT reconstructions illustrating bilateral costotransverse synostosis with multiple osseous bridges between the ribs and transverse processes on the right (A) and left (B) sides.*

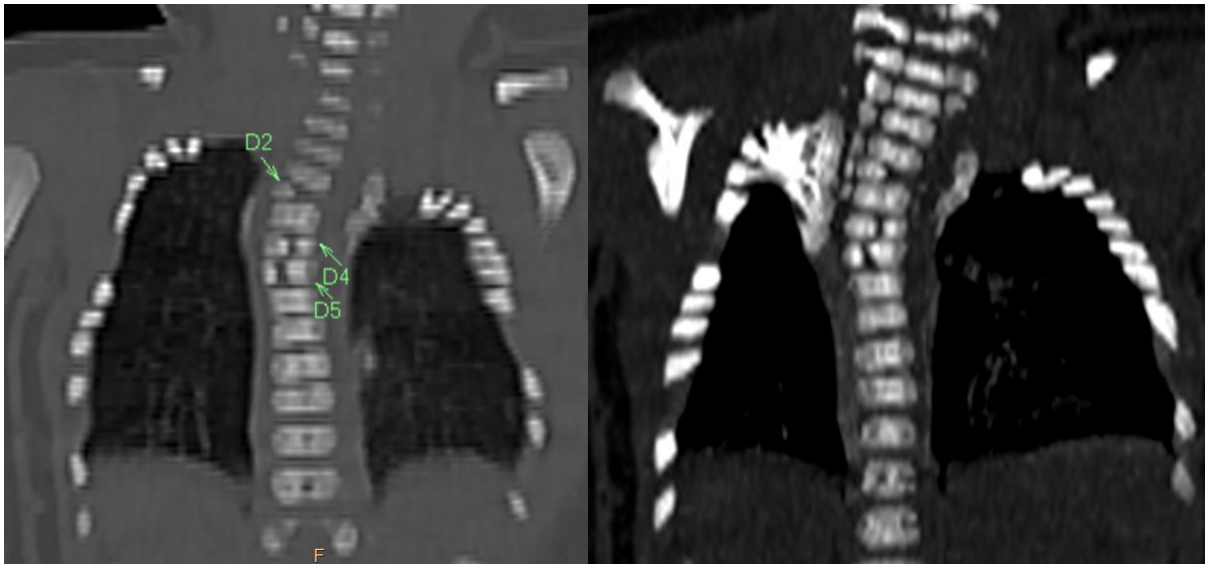


*Figure 3 : Coronal thoracic CT image showing butterfly vertebrae at D4 and D5, a right-sided hemivertebra at D2, and dorsal scoliosis with right convexity.*

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