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CASE REPORT

Spondilocostal disostosis associated with neural tube defect

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Abstract

Case report of a newborn with respiratory distress in the first hours of life due to a malformation of the costal framework. The present case becomes relevant because it raises an unusual cause of early respiratory distress in newborns and also, because there is an associated neural tube defect.

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INTRODUCTION

Spondylocostal dysostosis is a syndrome characterized by multiple segmentation defects of the vertebrae and ribs.¹ Associated defects, such as neural tube malformations, may be present.^{1,2} Individuals with this syndrome show respiratory impairment and may have a short stature. Treatment includes multidisciplinary follow-up.³

CASE DESCRIPTION

The patient is a female newborn at term weighing 3,100 g, who had Apgar scores of 9 and 10 as well as uneventful antenatal care. She was referred to our service after presenting respiratory distress in the first hours of life and requiring oxygen inhalation therapy.

The patient was admitted in good general health conditions, acting and reacting. Respiratory auscultation was normal, with a respiratory rate of 47 breaths/min and oxygen saturation of 97% at an oxygen flow rate of 0.5 L/min. She had a marked retraction of the left hemithorax, where, upon palpation, a partial absence of the thoracic cage could be observed. Cardiac auscultation was normal, with a heart rate of 138 beats/min. No changes were noted in the abdomen, back, urogenital region, and limbs.

Chest radiography showed multiple bilateral vertebral and rib malformations, most evident on the left, with a normal heart area.

The patient underwent abdominal echocardiography and ultrasonography, with normal results. Reports of thoracic spine computed tomography scan and magnetic resonance imaging revealed discreet dextroconvex scoliosis; multiple vertebral malformations with cervical and thoracic hemivertebrae; apparent anterolateral meningocele at the level of the T7–T8 segment, accompanied by syringomyelia; and agenesis of several left ribs, associated with hypoplasia and fusion of the other ribs at several levels. (Figures 1 and 2).

The patient's family history showed no similar cases or reports of parental consanguinity.

The patient remained hemodynamically stable and required oxygen inhalation therapy at only 0.5 L/min. She was discharged after a week of hospitalization, with exclusive breastfeeding and intermittent oxygen therapy, and was referred for multidisciplinary outpatient follow-up.

DISCUSSION

The first case of spondylocostal dysostosis was published in 1938 by Saul Jarcho and Paul M. Levin, who reported a case of two siblings with multiple vertebral and rib defects.²

The term "spondylocostal dysostosis" refers to a radiological phenotype characterized by multiple defects of the vertebrae and associated with rib malformations, such as poor alignment and fusion, and/or a small number of ribs.^{1,3} It is a defect in the embryonic cartilage segmentation of the axial skeleton due to autosomal recessive inheritance.

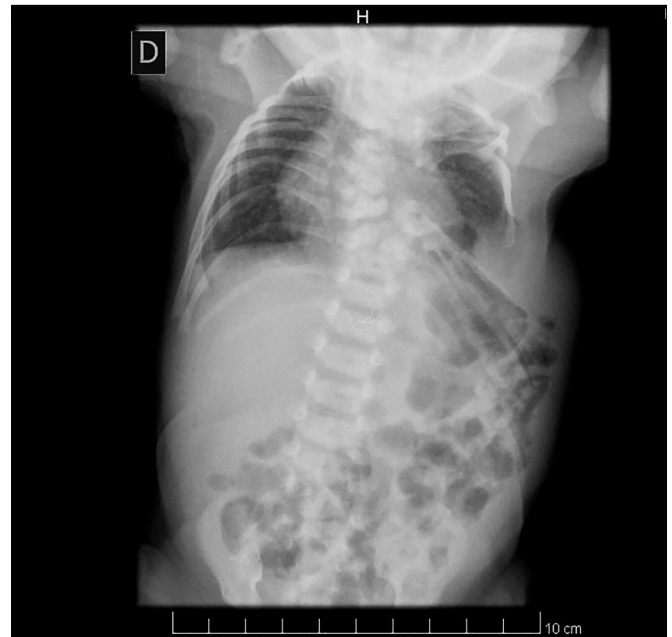


Figure 1. X-ray performed upon admission showing multiple hemivertebrae and left rib malformations.



Figure 2. Nuclear magnetic resonance showing anterior meningocele at the level of T7-T8, accompanied by syringomyelia.

Affected individuals have the following clinical findings: short neck, thorax shortened in relation to the rest of the body, short stature (most frequent), and mild to moderate non-progressive scoliosis.¹ A frequent occurrence of inguinal hernia appears to be related to the syndrome.^{1,3} Cases of spondylocostal dysostosis associated with neural tube defects, such as this one, are described in the literature, and it is believed that there might be some etiological connection between both conditions.^{2,3}

Among the clinical aspects, the respiratory complications resulting from poor thoracic expansibility have received the most attention.² Such complications can vary from mild respiratory distress, as in this case, to serious infections leading to respiratory failure, the latter being the leading cause of death in these individuals.³

The radiological presentation includes i) abnormal segmentation of at least 10 contiguous vertebral segments; ii) moderate scoliosis; iii) rib malformation, including fusion and/or reduced number; and, in general, iv) symmetrical shape of the thorax or slight asymmetry.¹

There are four different clinical and radiological subtypes, according to the genes involved:¹

- Type I - DLL3 gene: in addition to the four previous criteria, this type includes an ovoid-shaped ossification pattern of the vertebral bodies;
- Type II - MESP2 gene: all the vertebral bodies feature at least one type of malformation, with little involvement of the lumbar vertebrae, in comparison to the thoracic ones;
- Type III - LFNG gene: more severe shortening of the vertebral column in comparison to subtypes I and II, owing to more malformations of the vertebral bodies;
- Type IV - HES7 gene: vertebral malformation pattern that resembles both spondylothoracic dysostosis and type-I spondylocostal dysostosis. This type appears to have a higher association with neural tube malformations.¹

To confirm that a patient presenting multiple vertebral defects meets the diagnostic criteria for spondylocostal dysostosis, it is recommended to request radiological examinations of the entire skeleton in order to detect other abnormalities; to perform a complete physical examination; and to request ultrasound images of the abdomen, heart, kidneys, and urinary tract in order to exclude findings consistent with the differential diagnoses.¹⁻³ Family history should also be thoroughly searched for affected siblings or parental consanguinity.²

Differential diagnoses include Klippel-Feil syndrome and Alagille syndrome, among others.¹⁻³

The diagnosis of this syndrome is defined by clinical and radiological findings.¹ The subtype can be determined by a multigenic panel, which is still not commonly available for us.

Therapeutics in these cases mainly comprise the management of respiratory complications (respiratory distress and infections) with ventilatory support when necessary and physical therapy to improve lung function.⁴ It is extremely important to remain updated with the vaccination schedule, including respiratory syncytial virus immunization, given that respiratory infections are the leading cause of death before the first year of life among these patients.²

Selected cases can benefit from vertical expandable prosthetic titanium rib implants. This is an expandable metal prosthesis that simulates the function of ribs.⁵

Genetic counseling should also be provided to parents and patients in order to clarify the possibility of new cases in the family.¹

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