

Clinical Article

Spondylocostal Dysostosis: Presentation of Two Cases

Karakurt C, MD; Kara C, MD; Akgün D, MD; Sipahi T, MD

Abstract

Spondylocostal dysostosis is an uncommon inherited disease with multiple chondral and vertebral deformities (including cleft vertebrae, hemivertebrae and butterfly vertebrae) and bizarre costal features, such as variations in the size and thickness of the ribs and partial rib fusion or missing ribs. There are three major subtypes with different inheritance patterns. We present two cases of spondylothoracic dysostosis. *Int Pediatr.* 2001;16(3):173-175.

Key words: spondylocostal dysostosis

Introduction

Jarcho and Levin first reported malformations and abnormal fusion of several thoracic vertebrae and ribs associated with a short trunk and respiratory insufficiency in 1938. Spondylocostal dysostosis has been known by various names, including Jarcho-Levin syndrome, hereditary malformations of the vertebral bodies, syndrome of bizarre vertebral anomalies, costovertebral dysplasia, and occipitofacial-cervicothoracic-abdominodigital dysplasia, spondylocostal dysostosis.¹ The purpose of this paper is to present two cases of spondylothoracic dysostosis, a rarely seen heritable disease.

Case 1

BD, a 9-month-old girl, the product of a normal term delivery weighed 2750 g at birth, was admitted because of respiratory problems, cyanosis and vomiting. The patient was the first child of healthy parents who were first cousins. At the time of admission, the mother and father were 18 and 25 years old, respectively. There was no family history of congenital abnormalities. She has been admitted to different hospitals previously because of respiratory problems.

On admission, her weight was 4990 g (<3 percentile), length was 54 cm (<3 percentile) and head circumference was 44 cm (50-75 percentile).

Physical examination revealed: mild cyanosis, tachypnea, short trunk, retractions and rales, a protruding abdomen, and bilateral inguinal hernia. Radiological findings included vertebral anomalies, hemivertebrae, butterfly vertebrae, and absence of some of the lumbar vertebrae, respectively. Eight ribs were counted on the right and nine on the left side. The eighth rib on the right was enlarged (Fig 1). Abdominal ultrasonography, biochemical studies and chromosomal analysis were normal. Serum concentrations of amino acids and urinary excretion of mucopolysaccharides were normal.

Case 2

DV, a boy was delivered by a 25-year-old gravida 4, para 4, healthy women with three living children after an uneventful and appropriate prenatal care. Birth weight was 2500 g and length 44 cm. The father was a healthy 27-year-old man. Parents were first cousins. There was no family history of spinal abnormality. The patient has been admitted to different hospitals for repeated episodes of bronchopneumonia previously.

On admission, he was two years old. His weight was 7700 g (<3 percentile), length was 77 cm (<3 percentile) and head circumference was 49 cm (50th percentile).

Clinical examination showed a short neck and flail chest due to absent ribs. Radiographic examination showed multiple vertebral deformities, with hemivertebrae and marked costal anomalies (agenesis of the seventh, eighth and ninth rib) (Fig 2).

Abdominal ultrasonography, biochemical and chromosomal studies and echocardiography were normal. Radiographs of the spines of both parents were interpreted as normal. Serum concentrations of amino acids and urinary excretion of mucopolysaccharides were normal.

Discussion

Spondylocostal dysostosis is a clearly defined disorder because of clinical and radiological characteristics. Affected individuals have short trunk dwarfism of prenatal onset.² The patients show dwarfism because of cervico-

From Dr Sami Ulus Children's Hospital (Dr Karakurt, Dr Kara, Dr Akgün, Dr Sipahi T) Eryaman /Ankara/Turkey.

Address reprint request to İçtaş blokları B 2 Blok No: 22, Eryaman /Ankara/Turkey (Dr Karakurt).

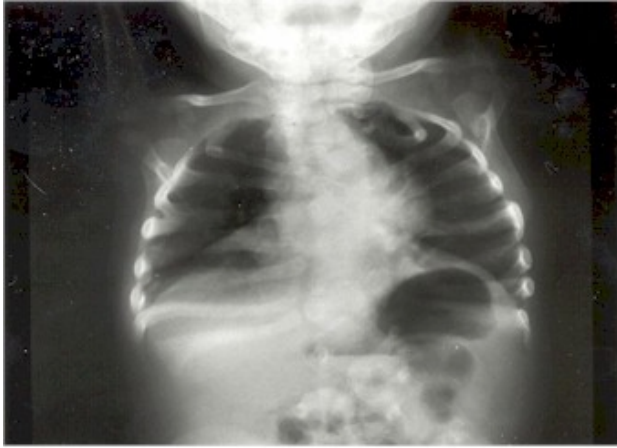


Fig 1 - Chest anteroposterior roentgenogram of Case 1.



Fig 2 - Chest anteroposterior roentgenogram of Case 2.

dorsal scoliosis or kyphoscoliosis, pigeon chest and /or flail chest, and reduction in the length of the thorax.³

From a radiological perspective, the disease is characterized by a series of structural changes that affect the vertebral body and ribs, and is associated with significant morphological and functional derangements in the verte-

bral column and thorax. The vertebral malformations include hemivertebrae, block vertebrae, fused vertebrae and spina bifida. The deformities of the ribs include symmetrically or asymmetrically absent ribs, and bifid or fused ribs especially near vertebral attachments.^{4,5} In addition to the characteristic skeletal malformations, other reported features of this syndrome include malformations of the urogenital system, cerebral malformations, cardiac malformation and neural tube defect.^{6,7,8,9}

The pathogenesis of the spondylocostal dysostosis remains unknown. Vertebral deformities may be attributed to defective segmentation of the somite at about the fourth or fifth week of intrauterine life. The costal abnormalities are probably secondary to the vertebral anomalies.¹⁰

On clinical and genetic grounds it is possible to distinguish at least three forms of spondylocostal dysostosis. In the first form, the inheritance is autosomal dominant and there is an abnormal life span without history of respiratory problems. The vertebral anomalies are mild. Patients with the severe autosomal recessive form are characterized by a symmetric crab-like chest and vertebral segmentation defects. Death may occur from respiratory infections before the age of two years.^{5,11} Benign autosomal recessive form is the most frequently described entity within the problems that are milder than the other forms.¹⁰

In our cases, autosomal recessive mode of inheritance may be considered because of the presence of one-degree parental consanguinity, and the lack of previous anomalies in the families. We suggest that these cases are examples of autosomal recessive benign forms of spondylocostal dysostosis.

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