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SPONDYLOCOSTAL DYSPLASIA AND CARDIAC ANOMALIES IN ONE DIZYGOTIC TWIN*

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SUMMARY: Özkınay F, Akısü M,Oral R, Tansuğ N, Özyürek R, Kültürsay N. (Department of Pediatrics, Ege University Faculty of Medicine, Bornova-İzmir, Turkey). Spondylocostal dysplasia and cardiac anomalies in one dizygotic twin. Turk J Pediatr 1986; 38: 381-384.

A 13-day-old, preterm, male infant was referred for respiratory distress syndrome (RDS) and jaundice. His twin sister had died of RDS on the second day of life in another hospital. The patient had typical features of spondylocostal dysplasia. Ventricular septal defect (VSD) and patent ductus arteriosus (PDA) were also diagnosed by echocardiographic evaluation. Parental consanguinity was not reported. There were no other similar cases in the family, and his twin sister and five-year-old living sister were free of deformities. Therefore, autosomal-recessive transmission may be considered first; however, because the patient was the only affected individual in this family, second denovo autosomal-dominant mutation should also be considered. This is the first reported case of spondylocostal dysplasia with VSD and PDA to our knowledge. *Key words: bones, dysostoses.*

Spondylocostal dysplasia was first described by Jarcho and Levin[1] in two brothers. The disease consists of an unusual association of multiple costal and vertebral deformities, including block vertebrae, hemivertebrae, anterior and posterior clefts of the vertebral column, and bizarre costal features such as variations in the size and thickness of the ribs and partial fusion or bifurcation. In the majority of cases there is a reduction in the number of ribs.

Spondylocostal dysplasia is a good example of genetic heterogeneity since there are two clinically similar forms, one dominant[2,3] and the other recessive[4,5]. We report here a case of spondylocostal dysplasia with cardiac anomalies.

Case Report

A 13-day-old male infant of a 23-year-old gravida 2, para 1 mother had been born following a 34-week twin gestation. No gestational problems, such as maternal disease, bleeding or known teratogenic exposure, were reported.

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The patient's twin sister had died of respiratory distress syndrome. There was no parental consanguinity or family history of congenital abnormalities. His twin sister and five-year-old sister were normal.

The patient's weight and length were 1450 g (3rd percentile) and 42 cm (10th percentile), respectively. Physical examination revealed a markedly short neck and flail chest due to absent ribs. Severe respiratory distress and cyanosis were observed. On cardiac auscultation a systolic murmur was heard.

Radiological findings included multiple vertebral deformities with hemivertebrae, crushed and fused vertebrae and scoliosis with convexity to the left in the dorsal column. The ribs showed marked deformities especially near their vertebral attachments. There were only nine ribs on the right side and ten on the left as well as irregularity in the spacing of the ribs on the right side. In addition, chest radiogram showed external herniation of the right lung and increased pulmonary circulation (Fig. 1). The karyotype was 46 XY.

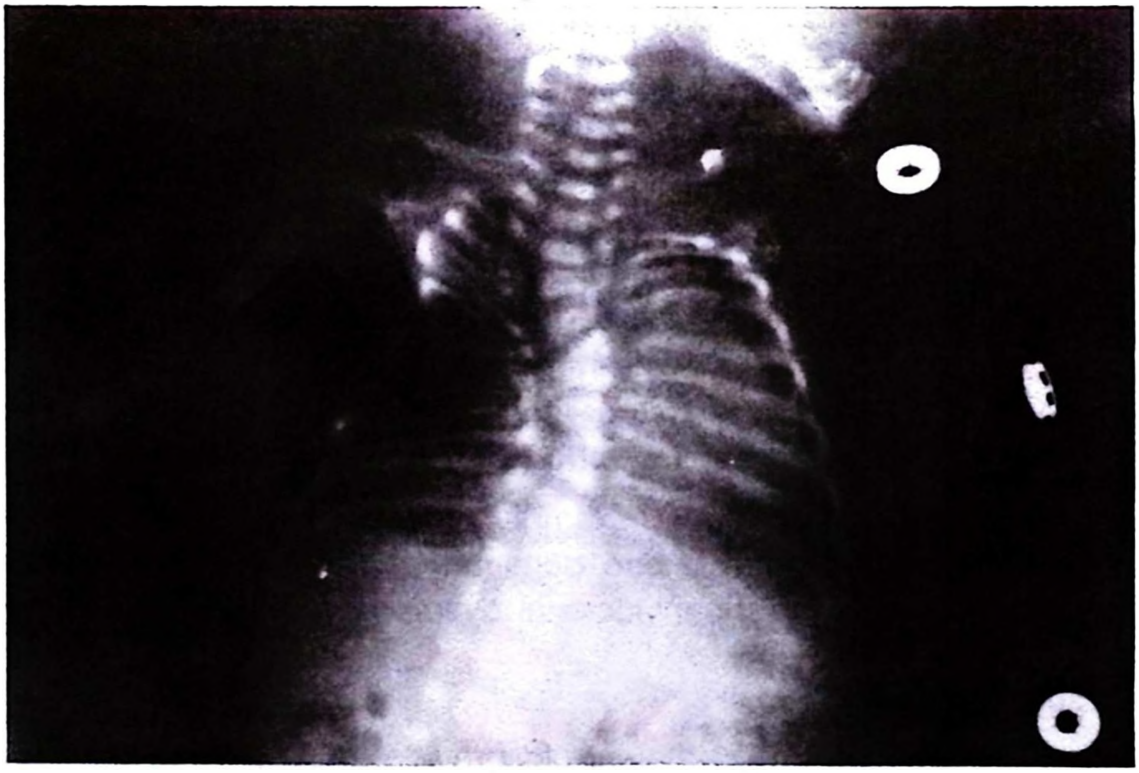

Fig. 1: X-ray of the spine and thorax showing malformed vertebral bodies (hemivertebrae, vertebral fusions), scoliosis and marked costal anomalies with absent ribs.

Echocardiography revealed a patent ductus arteriosus (PDA) of 2 mm, ventricular septal defect (VSD) of 3 mm and pulmonary hypertension.

Severe congestive heart failure developed on the 15th day of life, for which the patient was put on diuretic and digitalis treatment. This therapy was not satisfactory since the patient had pulmonary hypertension due to increased

pulmonary circulation. Three doses of indomethacin, a cyclo-oxigenase inhibitor, were administered perorally at a dose of 0.2 mg/kg in order to close the ductus arteriosus. Ductal closure was achieved after 36 hours, and the clinical findings of congestive heart failure subsequently disappeared.

Discussion

Spondylocostal dysplasia is a disorder with clearly defined clinical and radiological characteristics. The clinical picture is always apparent at birth. The patients show dwarfism because of cervico-dorsal scoliosis or kyphoscoliosis; torticollis, with a general decrease in the motility of the neck; pigeon chest and/or flail chest; and a reduction in the length of the thorax[4, 5].

From the radiological point of view, the disease is characterised by a series of structural changes that affect the vertebral body and ribs, and is associated with significant morphological and functional derangements in the vertebral column and thorax[5, 6]. Several types of malformations may be found in the vertebrae and ribs. The vertebral column may be affected from the atlas to the coccyx. The vertebral malformations include hemivertebrae, block vertebrae, fused vertebrae and spina bifida. The deformities of the ribs are closely related to those found in the spine and seem to be secondary to them. These deformities include symmetrically or asymmetrically absent ribs, and bifid or fused ribs especially near vertebral attachments[5, 6]. A primary failure in the development of chondrification centers takes place between the third and sixth weeks of gestation, subsequently leading to the aforesaid vertebral and rib abnormalities[6].

Both autosomal dominant and recessive inheritance have been reported in this disorder. Dominant inheritance has been demonstrated in the cases of Van der Sar[2] Peralta et al[7] and Caffey[8], and these patients were less severely affected than the patients who had autosomal-recessive inheritance[1, 9, 10]. Surviving children show severe respiratory complications in infancy, such as pulmonary atelectasis, upper respiratory tract infections, bronchitis and bronchopneumonia, which may be lethal[5, 6].

Both autosomal dominant and recessive transmission are possible in our patient. Autosomal recessive transmission may be considered first because of normal parents. However, denovo autosomal dominant mutation should also be considered because the patient is the only affected individidual in this family.

Only Franceschini et al.[5] have described an associated cardiac abnormality in the form of Fallot's tetralogy in a patient with spondylocostal dysplasia. This patient died of bronchopneumonia at the age of six years. A case of dizygotic twins with only one twin affected with spondylocostal dysplasia has not been reported before to the best of our knowledge; neither has the presence of these specific cardiac anomalies.

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