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Case Report

Spondylocostal Dysostosis Associated with Type I Split Cord Malformation and Double Nipple on One Side: A Case Report

Tip 1 Split Kord Malformasyonu ve Bir Tarafta Çift Meme Başının Eşlik Ettiği Spondilokostal Dizostozis: Bir Olgu Sunumu

Muhammet Bahadır YILMAZ[1], Aysegul KAYMAK[2], Gokhan KURT[1], Ferda Emriye PERCIN[2], Kemali BAYKANER[1]

[1]Gazi University, Faculty of Medicine, Division of Pediatric Neurosurgery, Department of Neurosurgery, Ankara, Turkey
[2]Gazi University, Faculty of Medicine, Department of Medical Genetics, Ankara, Turkey

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Correspondence address: Muhammet Bahadır YILMAZ / E-mail: bahadiryilmaz@yahoo.com.tr

ABSTRACT

Jarcho Levin syndrome is a rare genetic disorder characterized by multipl vertebral and costal anomalies at birth. Jarcho Levin syndrome includes two phenotypic groups: spondylothoracic dysostosis and spondylocostal dysostosis. The prognosis of spondylothoracic dysostosis has worse than spondylocostal dysostosis, because of respiratory complications. Associated malformations include those of the congenital heart disease, urogenital malformation, skeletal anomalies and neural tube defects. We present a patient with spondylocostal dysostosis, who also had type I split cord malformation, tethered cord, scoliosis and double nipple on the right. Although the association of spondylocostal dysostosis and type I split cord malformation is very rare, double nipples on one side is no previously reported.

KEYWORDS: Spondylocostal dysostosis, Spondylothoracic dysostosis, Jarcho Levin syndrome, Congenital scoliosis, Accessory nipple, Type I split cord malformation

ÖZ

Jarcho Levin Sendromu doğumda multipl vertebral ve kostal anomalileri ile karakterize,nadir bir genetik hastalıktır. Jarcho Levin Sendromu iki fenotipik grup içerir: spondilotorasik dizostozis ve spondilokostal dizostozis. Solunumsal komplikasyonlar nedeniyle spondilotorasik dizostozisin prognozu spondilokostal dizostozisden daha kötüdür. Konjenital kalp hastalıkları, ürogenital malformasyonlar, iskelet anomalileri ve nöral tüp defektleri eşlik eden anomalilerdir. Biz aynı zamanda tip 1 split kord malformasyon, gergin kord, skolyoz ve sağ tarafta çift meme başına sahip olan spondilokostal dizostozisli hasta sunuyoruz. Spondilokostal dizostozis ve tip 1 split kord birlikteliği çok nadir olmakla birlikte, bir tarafta çift meme başının eşlik etmesi daha önce rapor edilmemiştir.

ANAHTAR SÖZCÜKLER: Spondilokostal dizostozis, Spondilotorasik dizostozis, Jarcho Levin sendromu, Konjenital skolyoz, Aksesuar meme başı, Tip 1 split kord malformasyonu

INTRODUCTION

Jarcho Levin syndrome (JLS) is a rare genetic disorder characterized by multiple vertebral and costal anomalies at birth. This syndrome is usually diagnosed in newborns. Jarcho and Levin first described cases in 1938 (8). Solomon et al. classified cases of JLS into two phenotypic groups: spondylothoracic dysostosis (STD) and spondylocostal dysostosis (SCD) (4, 15). The prognosis of STD is grimmer than SCD, because of restrictive lung disease, pulmonary hypertension, congestive heart failure and respiratory infections (4, 10,14, 17). In addition, congenital heart defects, abdominal wall malformations, urogenital and anal abnormalities, multiple skeletal anomalies, upper limb anomalies, spina bifida, inguinal, umblical and diaphragmatic hernias have been described in JLS (5, 6, 12, 13, 17).

We present a patient with SCD, who also had type I split cord malformation, tethered cord, scoliosis and double nipple on the right without respiratory problems. Although the association of SCD and type I split cord malformation is very rare, double nipples on one side is not previously reported.

CASE REPORT

A 2-year-old female child presented with thoracolumbar scoliosis, and skin lesion, hypertrichosis and red discolouration at thoracolumbar junction (Figure 1). We found double nipples on the right side during physical examination of the patient (Figure 1). Skin lesion on the back of the patient showed similarity of radiological image of split cord (Figure 1). The neurological examination was normal. There was no respiratory problem. Our patient's chromosomal analysis was

normal. Radiological examination showed that the conus was tethered, type I split cord malformation consists of two hemicords at the level of L2 vertebra, and multiple vertebral deformities at the level of T9 between L1 (Figure 2). The first four ribs were absent on right side. There was an irregular appearance at the fifth with sixth ribs, and seventh with eighth ribs (Figure 2). In addition, X-ray demonstrated that the median dense area at the level of L2 (Figure 2). Her parents had no vertebral anomaly. She was operated and the bone septum was removed (Figure 3). Two hemicords within two separate dural sheaths were placed into same dura. There was no neurological deficit after the operation. She was followed in terms of tethered cord and respiratory complications.

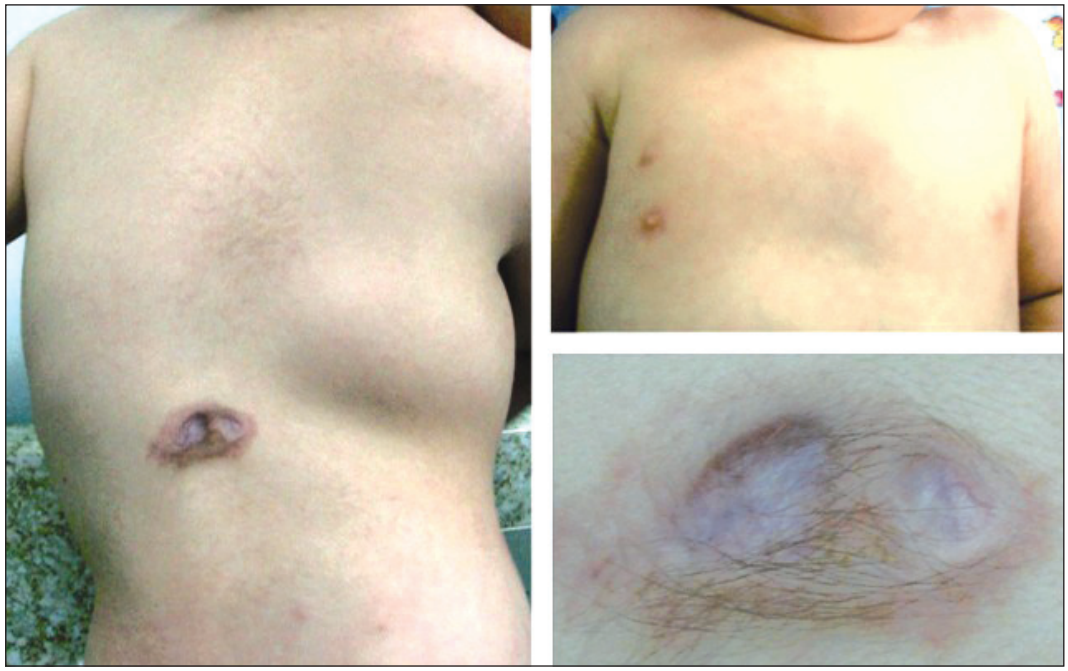


Figure 1: Picture showing a scoliosis, hypertrichosis, red discolouration at thoracolumbar junction, and double nipples on the right side. Skin lesion on the back of the patient showing similarity of radiological image of split cord.

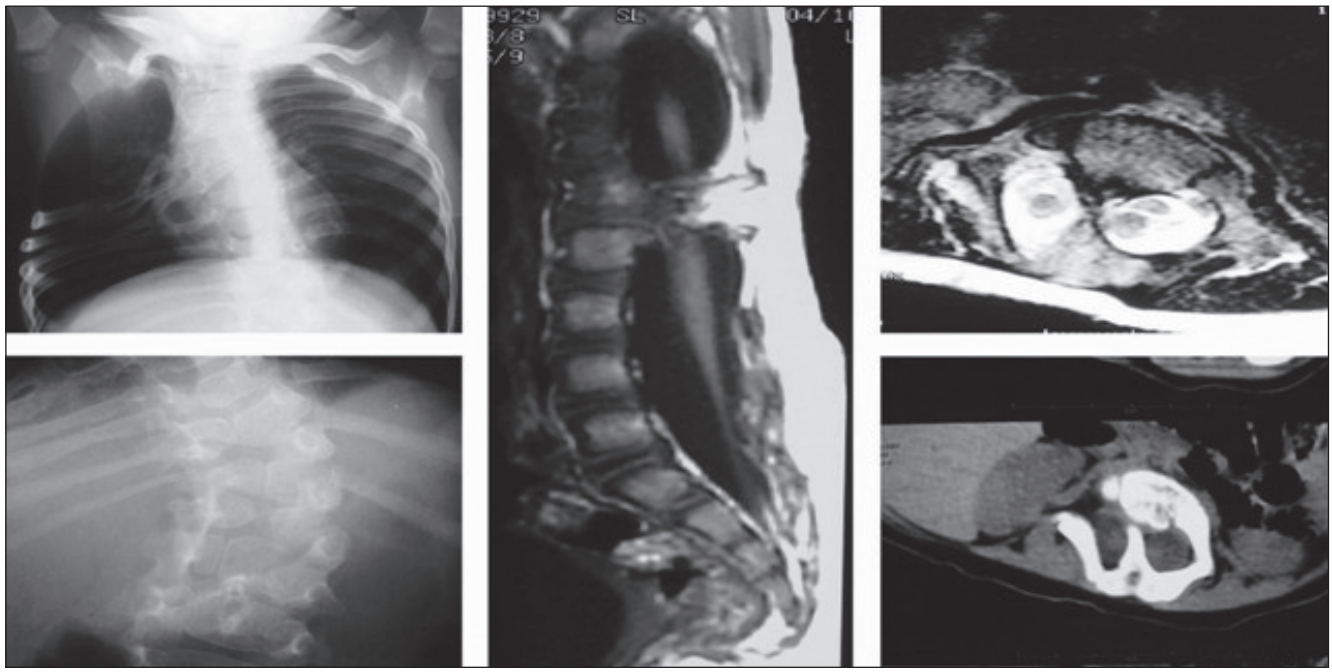
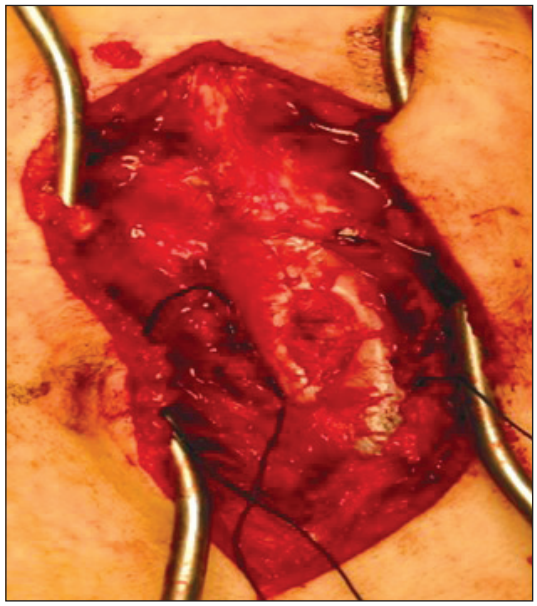


Figure 2: Chest radiograph shows the right first four ribs to be missing, irregular fusion of the ribs (5-8) on the right side. In addition, spine radiograph shows the median dense area at the level of L2, and multiple vertebral deformities at the level of T9 between L1. Magnetic resonance images show tethered cord, type I split cord malformation consists of two hemicords at the level of L2 vertebrae. Computed tomography shows bone septum at the level of L2 vertebrae.



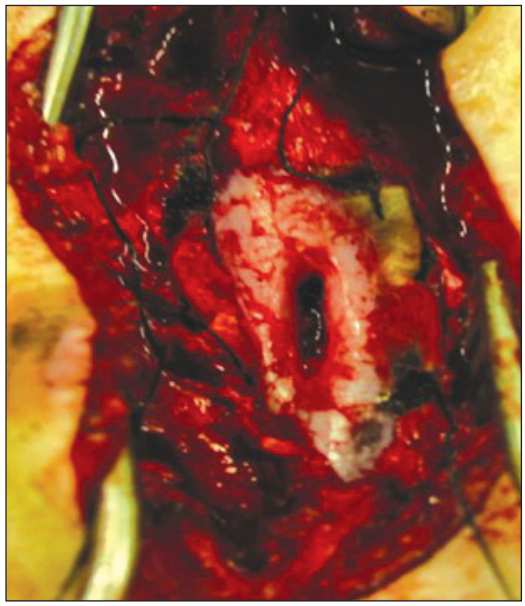


Figure 3: Picture showing bone septum and postoperative appearance.

DISCUSSION

Jarcho and Levin first described cases in 1938 (8). They described two sibs with short neck and short thorax resulting from multiple congenital vertebral and rib cage malformations (2, 17). In 1978, Solomon et al. classified cases of JLS into two phenotypic groups: spondylothoracic dysostosis (STD) and spondylocostal dysostosis (SCD) (4, 15). The incidence is unknown for STD, but SCD has a reported prevalence of 0.2 per 100,000 liveborns (2, 11, 17). Although Campbell reported no gender predilection, Vázquez-López reported that it is more common in females (2, 17).

SCD is also a rare congenital disorder with multiple vertebral and numerical or structural rib abnormalities that resulting in thoracic asymmetry and short stature and neck (7). Rib anomalies include absence, posterior fusion and irregular or bifid ribs (4, 17). STD is a rare congenital disorder with segmentation and formation defects in cervical, thoracic and lumbar spine such as hemivertebrae, block vertebrae, butterfly vertebrae, and unsegmented bars with fusion of all the ribs at the costovertebral junction (10). Hemivertabrae is the most common spinal anomaly and seen mostly in the thorocolumbar region (4, 9) STD is a characterized by the presence of 'crab-like' chest, but there are no intrinsic rib malformations reported (4, 14).

The prognosis of STD is grimmer than SCD, because of restrictive lung disease, pulmonary hypertension, congestive heart failure and respiratory infections (4, 14, 17). Dane et al. reported 45% mortality in neonatal or infancy with STD from restrictive lung disease, pulmonary hypertension, congestive heart failure and respiratory infections (2, 4, 5, 10, 14, 17). Patient with SCD has usually a much lower mortality rate. Most cases of SCD are transmitted in autosomal recessive way, but rarely autosomal dominant transmission was reported (7, 9, 13, 17). STD has autosomal recessive transmission way (9, 17). Patients with SCD have mutations in the delta-like 3 (DLL3) gene on chromosome 19 (1, 7, 10, 16, 17). Children with STD have no mutations in the DLL3 gene (3, 17). Whittock et al. demonstrated a mutation in the MESP 2 gene in 2 sibs affected by a mild variety of SCD (10, 18).

Scoliosis occurs in one-third of the JLS (17). Other associated anomalies include congenital heart defects, abdominal wall malformations, inguinal, umbilical and diaphragmatic hernias, urinary tract and renal malformations, anal atresia, upper limb anomalies, spina bifida (5, 6, 12, 13,17). Less commonly, other skeletal malformations such as winged scapula, irregular clavicle, absent atlas, and hypoplastic humerus can be found. Most patients do not have craniofacial malformations (9, 17).

Although the association of SCD and type I split cord malformation is described, the association between those pathologies and double nipples on one side is not previously reported.

Although prenatal ultrasound diagnosis can be practiced as early as 16. weeks of gestation (10), in some cases could be difficult. Ultrasound characteristically revealed presence of fanned out ribs and skeletal defects.

The aim of management, should be suitable patient care, prevention and early, aggressive treatment of respiratory infections. Preoperative pulmonary function tests are important. The lung capacity of the child younger than age 6 years should be assessed with tomography (2).

Spinal surgery of those cases prevents scoliosis and pulmonary restriction. Hence associated pulmonary and cardiac complications could be decreased (10). Reconstructive surgery to gain more thoracic volume by titanium rib implants has been described in the treatment of that disorder.

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