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Jarcho-Levin Syndrome (Spondylocostal Dysostosis) and Hydrocephalia: Case report

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ÖZET

Jarcho-Levin sendromu (spondilokostal dizostozis) ve hidrosefali: Olgu sunumu

Jarcho-Levin sendromu çoklu vertebra ve kosta anomalileri ile karakterize nadir bir genetik bozukluktur. Biz konjenital hidrosefalisi ve Jarcho-Levin sendromu olan 10 günlük bir erkek yenidoğanı rapor ettik.

Anahtar kelimeler: Jarcho-Levin sendromu, kostaların yokluğu, hidrosefali

ABSTRACT

Jarcho-Levin syndrome (spondylocostal dysostosis) and hydrocephalia: case report

Jarcho-Levin syndrome is a rare genetic disorder characterized by multiple vertebral and rib anomalies. Hrein we reported a 10-day-old male newborn with Jarcho-Levin syndrome and congenital hydrocephaly.

Key words: Jarcho-Levin syndrome, absence of rib, hydrocephalia

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INTRODUCTION

There are two types of Jarcho-Levin syndrome, spondylothoracic dysplasia and spondylocostal dysostosis. Spondylothoracic dysplasia has a grimmer prognosis than spondylocostal dysostosis. Many of the infants born with this disorder succumb to respiratory failure. With new advances in medical care, even the more severely affected children have an improved chance of survival (1). This is a case presentation of one neonate with spondylocostal dysostosis.

CASE REPORT

The proband, a male, was born to a 25 year old woman after a 38 week, uncomplicated pregnancy. Apgar scores were 4 and 7 at one and five minutes, respectively. Shortly after birth, the infant had considerable respiratory difficulty requiring intubation in the delivery room and subsequently neonatal intensive care. The thorax was grossly deformed with shortening of the trunk and neck and a protuberant abdomen. On examination at birth, he was 3.030 gr and 46 cm in length. The occipitofrontal head circumference was 37.5 cm.

The spine was short and exhibited marked thoracolumbar lordosis and dextroconvex kyphoscoliosis (Figure 1). There was an imperforate anus. The genitalia were normal except his the left foot which had pes equinovarus. Computerised tomography (CT) scan of the head showed moderate hydrocephalus owing to aqueductal stenosis. An abdominal ultrasonography showed normal structures. X ray of the spine revealed rib and vertebral anomalies. He had only three ribs at right part of thorax. He had "crab-like'' rib anomalies (Figure 2). Extensive and severe developmental anomalies of the vertebral bodies and widely open and spread neural arches were most marked in the thoracolumbar area. Hemivertebrae and block vertebrae were present, while other vertebral bodies were partially fused and irregularly deformed. His echocardiography was normal. Chromosome studies using high resolution banding gave normal results (46,XY).

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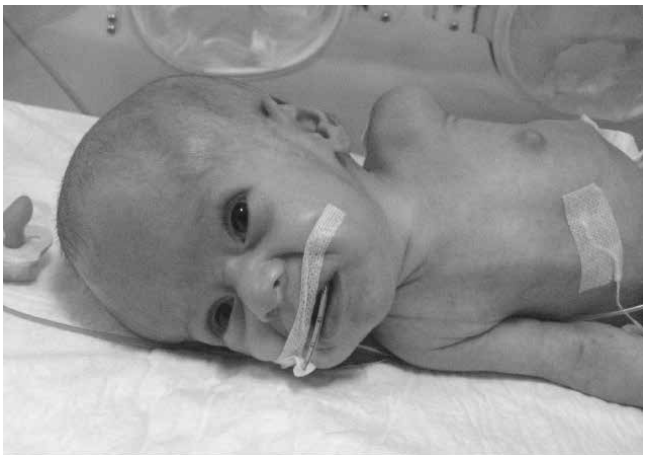

Figure 1: Appearance at birth showing hydrocephalia

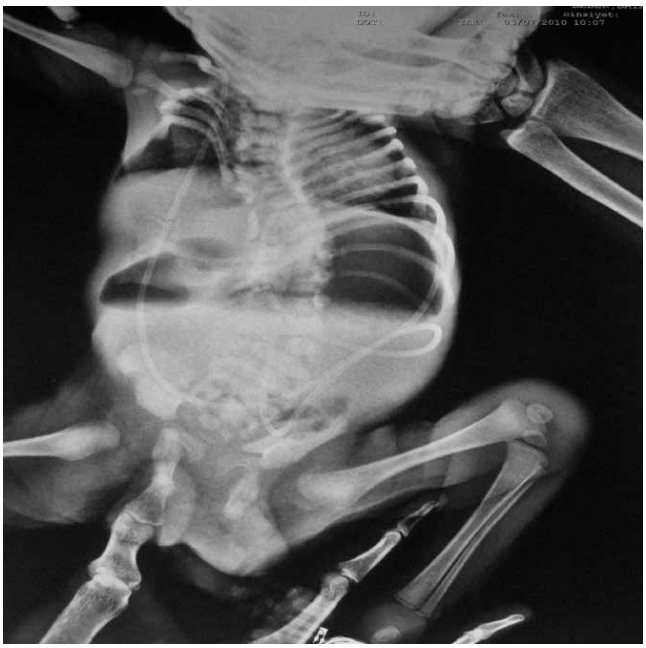

Figure 2: Anteroposterior radiograph showing shortening of the spine and vertebral and rib abnormalities

The infant has remained dependent on a respirator and had an episode of pneumonia. He underwent repair of the meningomyelocele and placement of a ventriculoperitoneal shunt. The infant's parents were non-consanguineous. There was no history of teratogen exposure during the pregnancy. The father was 30 years old. The 8 and 6 year old sibs were normal.

DISCUSSION

Jarcho-Levin syndrome is a rare genetic disorder characterized by multiple vertebral and rib anomalies (1). Jarcho and Levin described this disorder in 1938 (2). It is now clear that Jarcho-Levin Syndrome represents one of at least three disorders characterized by multiple vertebral segmentation defects. Most reported cases of Jarcho-Levin syndrome have occurred in Puerto Rican individuals. This disorder has an autosomal recessive inheritance pattern.

This syndrome has short trunk dwarfism of prenatal onset, prominent occiput; tendency to have broad forehead, wide nasal bridge, anteverted nares, and upsland to palpebral fissures. Short thorax with "crab-like'' rib cage associated with multiple vertebral segmentation defects and ribs that flare in a fan-like pattern; posterior fusion and absence of ribs; short neck and low posterior hairline, pectus carinatum, increased anterioposterior chest diameter; lordosis; kyphoscoliosis. Normal with impression of being long (2).

Occasional abnormalities of Jarcho-Levin syndrome is cleft palate, cryptorchidism, hernias, hydronephrosis with ureteral obstruction, bilobed bladder, absent external genitalia, and anal and uretheral atresia, uterus didelphys, cerebral polygria, neural tube defects, single umblical artery (2). The vast majority of affacted individuals die in early infancy as a result of recurrent pulmorary infection and respiratory insufficiency secondary to the small thoracic volume (2,3). In most recent studies, some authors reported two neonates with the lethal variety of this syndrome. One neonate had associated anomalies like hydrocephalus, hydroureteronephrosis and meningomyelocoele while the other had no additional anomalies. Also it is reviewed in the literature regarding this less understood disorder focusing on the applied clinical aspects that have stemmed out from the recent molecular research (4). In several studies, hitherto, a plethora of associated anomalies have been described in several reports. The prognosis is directly related to respiratory complications. Reported findings associated with Jarcho-Levin syndrome include congenital heart defects, abdominal wall malformations, genitourinary malformations, upper limb anomalies, and neural tube defects (3,5,6). The small size of the thorax in newborns frequently leads to respiratory compromise and death in infancy (7). In a recent report, the authors describe a case of Jarcho-Levin syndrome who has Wilms tumor and bilateral cystic renal disease (8). Our patient had short trunk and hydrocephaly, meningomyelocele, vertebral anomalies, absence of rib,

pes equinovarus and anal atresia, with respiratory problems.

There are two types of Jarcho-Levin syndrome, spondylothoracic dysplasia and spondylocostal dysostosis. Spondylothoracic dysplasia has a grimmer prognosis than spondylocostal dysostosis. Many of the infants born with this disorder succumb to respiratory failure. With new advances in medical care, even the more severely affected children have an improved chance of survival (1). Our case was spondylocostal dysostosis.

Based on clinical and radiologic evaluation of infants with multiple vertebral segmentation defects, tree distinct entities have been identified: Jarcho-Levin syndrome is characterized by symmetric "crab-like'' thoracic spine and ribs with short-trunk short stature and radiographic features of multiple hemivertebrae and rib fusion, and early lethality in virtually all cases. Spondylothoracic dysostosis is also an autosomal recessive disorder. Intrafamilial variability in clinical severity is striking. Despite multiple vertebral segmentation defects, a "crab-like'' thoracic spine is not evident. Spondylocostal dysostosis is an autosomal dominant disorder that usually presents after infancy with problems related to kyphoscoliosis, low back pain, or decreased mobility of the spine (2). Prenatal diagnosis of spondylocostal dysostosis is important to provide appropriate genetic counseling and to have an adequate setting for the delivery of the fetus (5,9).

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