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Jarcho-Levin Syndrome

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The Jarcho-Levin syndrome (JLS) is a clinico-radiological entity characterized by short-neck, short-trunk, normal sized limbs and multiple vertebral and rib defects on skeletal survey. This syndrome was first described by Jarcho and Levin in 1938(1). About 65 cases have been reported in the literature(2,3). Recently, this syndrome has been divided into two major subtypes: spondylothoracic dysostosis and spondylocostal dysostosis(3,4).

We describe two cases of the Jarcho-Levin syndrome, one of each subtype (spondylothoracic dysostosis and spondylocostal dysostosis). The cases illustrate the typical findings of the syndrome and highlight the differences between the two subtypes of this syndrome.

Case Reports

Case 1: A 2-month-old boy was brought to the Pediatric Out-Patient with a 1.5 months history of fever and respiratory distress. He was born by Cesarean section at term, to a 28-year-old father and 25-year-old mother. His birth weight was 2,500 g and he cried 30 minutes after birth. However, he was noted to have severe respiratory distress at birth, with a respiratory rate of 150 per minute. He was the second child of non-consanguineous parents. His sister had died at four days of age due to cyanotic congenital heart disease.

On examination his weight was 3.75 kg (10-25th centile), length 49 cm (<5th centile) and head circumference 36 cm (10-25th centile). The upper segment to lower segment ratio was 1.33. There was severe respiratory distress with a respiratory rate of

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80 per minute. The neck and chest were extremely short and there was thoracic kyphoscoliosis *(Fig. 1)*. Abdomen was protuberant and there were bilateral congenital hydrocele.

Skeletal survey showed multiple thoracic vertebral anomalies and a "crab-like" configuration of the ribs (*Fig.* 2). There were no intrinsic rib anomalies such as absence, broadening or bifurcation, but some of the ribs on both sides showed posterior fusion. Cranial ultrasound was normal. Abdominal sonography showed that both kidneys were enlarged with distended pelvicalyceal systems. The ureters were not visualized suggesting bilteral partial pelviureteric junction obstruction.

Blood biochemistry showed normal renal function and normal blood gases. *Acinetobacter* was cultured from blood. The child died at 80 days of age with septicemia. Consent for autopsy was refused by the parents.

Case 2: A 1-month-old girl, was brought to the Genetics Clinic with complaints of chest deformity. She was born at home, after full-term gestation, by spontaneous vaginal delivery. There was no birth asphyxia and the birth weight was not recorded. She was the third child of non-consanguineous parents. The father was 35 years old and the mother 32 years of age. Her two siblings were normal.

On examination her weight was 3 kg (<5th centile), length 49 cm (<5th centile),

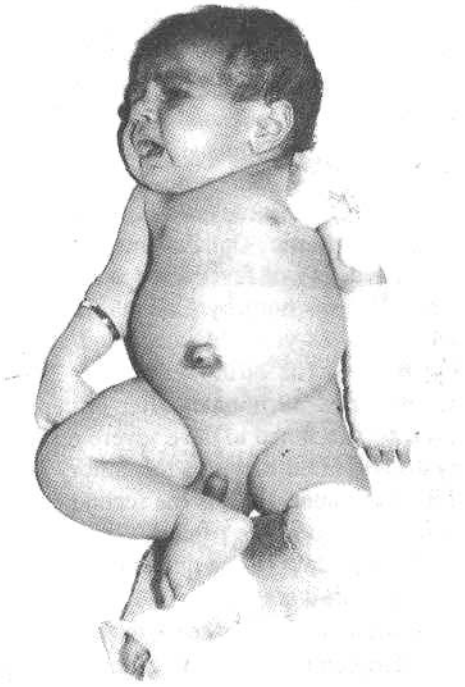

Fig. 1. Case 1 with Jarcho-Levin syndrome (spondylothoracic dysostosis) showing short-neck, short-trunk, protuberant abdomen and normal limbs.

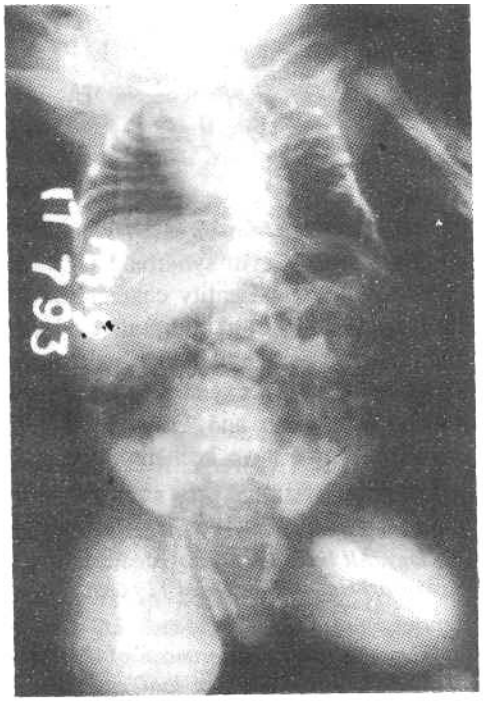

Fig. 2. Infantogram of child in Fig. 1 showing multiple thoracic vertebral defects and "crab-like" configuration of rib-cage due to posterior fusion of some of the ribs.

arm-span 61 cm and upper segment to lower segment ratio 1.13. Her ears were low set and she had a high-arched palate. Both the neck and thorax were extremely short and there was marked thoracic kyphoscoliosis. There was a high thoracic meningocele, the overlying skin of which was very thin and membranous with a tuft of long hair. There was no respiratory distress or neurological deficit.

The skeletal survey showed several anomalies of the thoracic vertebrae and the ribs on both sides. The thorax had a "crab-like" configuration and the rib anomalies were more pronounced on the right side and included absence, posterior fusion and bifurcation of ribs*(Fig. 3)*. There was also severe thoracic scoliosis. Cranial ultrasound was normal and abdominal ultrasound showed normal kidneys.

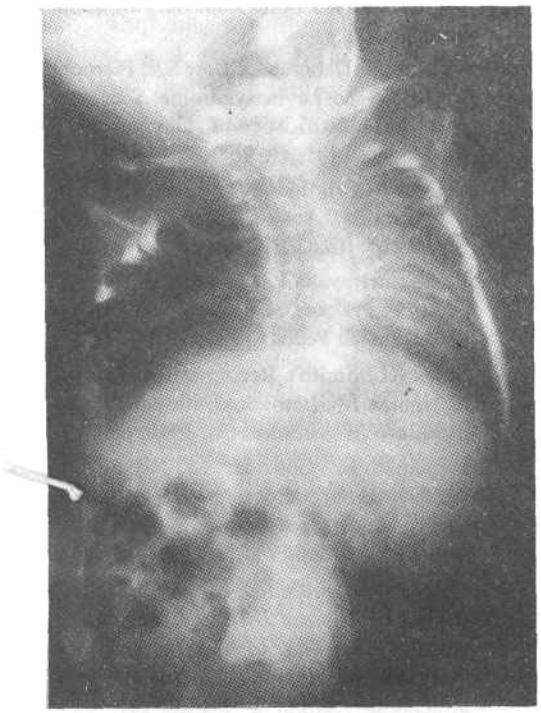

Fig. 3. Infantogram of Case 2 showing multiple thoracic vertebral anomalies and severe rib defects (absence and posterior fusion) more marked on right side.

The child is being followed up and is being managed by the pediatric and orthopedic surgeons for her neural tube defect and thoracic kyphoscoliosis, respectively.

Discussion

Jarcho-Levin syndrome (JLS) is a rare disorder with one possible case report from India(5). This syndrome is underdiagnosed, although it has distinctive clinical and radiological features. In a patient with multiple congenital vertebral and rib anomalies the presence of a short-neck, short-trunk, normal length limbs and certain associated anomalies suggests a diagnosis of JLS. Associated anomalies described in this syndrome include hernias, neural tube defects and anomalies of the anal opening, urinary tract, external genitalia, uterus and lower limbs(3,6). Recently, airway abnormalities have been described in two patients with the Jarcho-Levin syndrome(7). The authors feel that this may be an important factor contributing to the respiratory failure often seen in affected infants(7).

The Jarcho-Levin syndrome has been divided into two major subtypes, namely spondylothoracic dysostosis and spondylocostal dysostosis. These subtypes have different survival rates, associated anomalies, and inheritance patterns(3,4). Patients with spondylothoracic dysostosis (STD) have vertebral anomalies with a "fan-like" or "crab-like" rib configuration. Intrinsic rib abnormalities in this subtype are limited to a decrease in number and posterior fusion. They have a higher incidence of neural tube defects and a higher mortality rate and inherit the disorder in an autosomal recessive manner. Our first patient *(Case 1)* had this

subtype of the Jarcho-Levin syndrome. He had multiple vertebral anomalies with a "crab-like" configuration of the ribs with posterior tethering of the ribs. As he was the only affected child of normal parents an autosomal recessive pattern of inheritance is likely. Although, he died of septicemia he also had respiratory difficulty which is a major cause of death in these patients.

The second subtype of JLS is spondylocostal dysostosis (SCD). Patients with this subtype have vertebral anomalies associated with marked intrinsic rib abnormalities such as absent ribs, abnormal orientation, irregularity of shape and size, bifurcation, broadening, fusion, etc.(3,4). The configuration of the chest is usually not "crab-like" or "fan-like". They are more often short statured than patients with spondylothoracic dysostosis and have twice their survival rate. They are also less likely to have associated neural tube defects and inherit the disorder in either an autosomal recessive or autosomal dominant manner. Our second patient *(Case 2)* falls into this category, due to the presence of severe intrinsic rib defects, even though her thorax had a "crab-like" configuration. This patient also had a high thoracic neural tube defect, an abnormality more frequently seen in patients with spondylothoracic dysostosis. Since neural tube defects are also seen in patients with spondylocostal dysostosis, albeit less frequently, the presence of this defect does not, in itself, alter the diagnosis of SCD in this patient. An autosomal recessive pattern of inheritance was also possible in this patient, but occurrence of a new mutation of an autosomal dominant disorder (a pattern of inheritance also described in SCD but not in STD) cannot be ruled out.

Our aim in presenting these two cases of Jarcho-Levin syndrome is to make pediatricians aware of this disorder, its associated anomalies, its two subtypes and the prognosis based on classification into subtypes. These disorders can be diagnosed antenatally by high-resolution fetal scans at 18-23 weeks of gestation(5,8,9). A correct diagnosis made in a previously affected child can help in genetic counselling of the couple at risk and in establishing the diagnosis prenatally in a subsequent pregnancy.

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