Case Report

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A case report of Jarcho–Levin syndrome

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Abstract

Jarcho–Levin syndrome (JLS) is a rare, congenital skeletal disorder, inherited in an autosomal recessive pattern that represents a spectrum of clinical and radiographic abnormalities of the spine and chest, resulting in varying degrees of thoracic insufficiency, presenting at birth. The association with other organs such as the spinal, brain, and the urinary system has been reported. The paper reports a rare case of Jarcho-Levin syndrome associated with neural tube defects and other congenital malformations. The diagnosis is based on clinical and radiological findings, characteristic physical appearance, symptoms of thoracic insufficiency, family history, consanguineous parents, skeletal survey, or specialized genetic tests for mutations. Milder forms are compatible with life.

Keywords: Jarcho–Levin syndrome, congenital disorder, spondylocostal dysostosis, vertebral deformity, Respiratory distress

Case Report

A two-month-old full-term baby boy presented to our hospital with respiratory distress. He was the first child of a consanguineous parents. He was born full term by C- section delivery. Apgar score was normal. Birth weight was 2.550 gram. The second trimester morphology ultrasound showed a shortened spine with disordered vertebrae and thick skin and there was no family history of inherited skeletal dwarfism. He was noticed to have short neck / trunk and abdominal distension and the limbs though of healthy appearance appeared relatively longer. There was no evidence of any mental or neurologic deficit. He developed respiratory distress soon after birth. The chest cavity appeared small and asymmetric. The ribs were malformed, and a few were fused, bifid, or missing. There was a posterior crowding of the ribs owing to their fusion at the next costovertebral junction and anterior flaring, giving a crab like or fan like appearance to the rib cage. Cranial ultrasound was normal. Medullar ultrasound showed tethered cord syndrome associated to lipomyelocele. Abdominal ultrasound showed a single kidney.

Discussion

Jarcho-Levin syndrome (JLS) has been reported as 1 in 40000 births globally. It occurs with equal frequency in males and females. Jarcho-Levin is caused by a gene mutation inherited as an autosomal recessive trait. It is characterized by abnormalities in the spine with malformed vertebrae and chest with a crab like appearance of rib cage. It was first described in 1938 by Jarcho and Levin in cases of thoracic insufficiency due to vertebral and rib anomalies.

The broad group of JLS is subdivided into two major clinical subtypes spondylothoracic dysplasia (STD) and spondylocostal dysostosis (SCD). The typical small malformed "fan like" or "crab like" rib cage is due to posterior fusion and anterior

Figure 1: *Case with jarcho-levin syndrome (SCD) showing short neck, short trunk, protuberant abdomen and normal limbs.*

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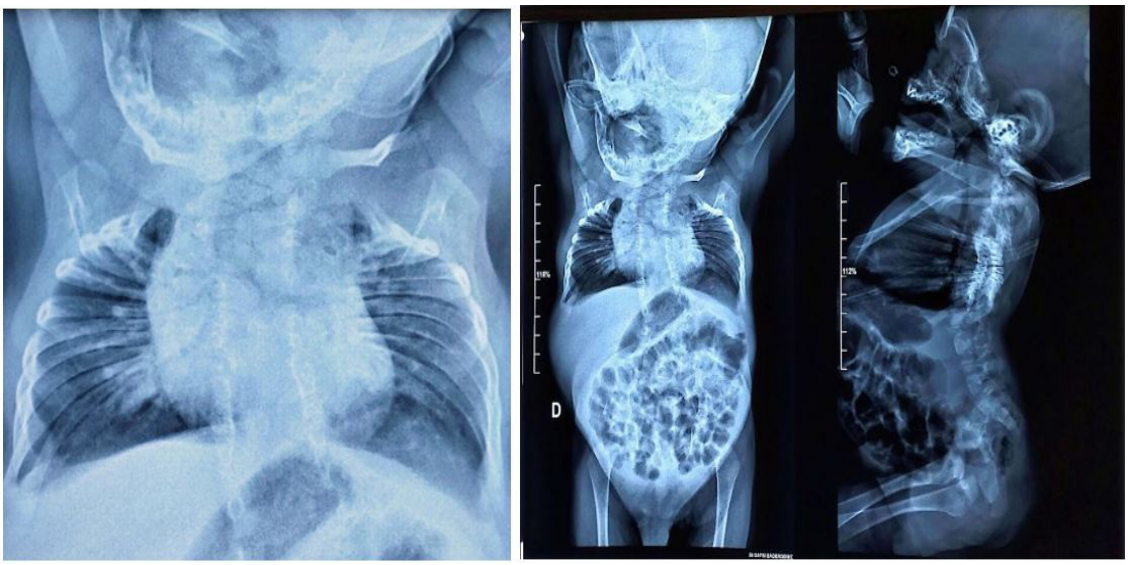

Figure 2: *Chest and thoraco-abdominal radiographs (a and b) and lateral view (c) showing multiple rib anomalies seen as crab like or fan like, associated with vertebral segmentation and fusion anomalies.*

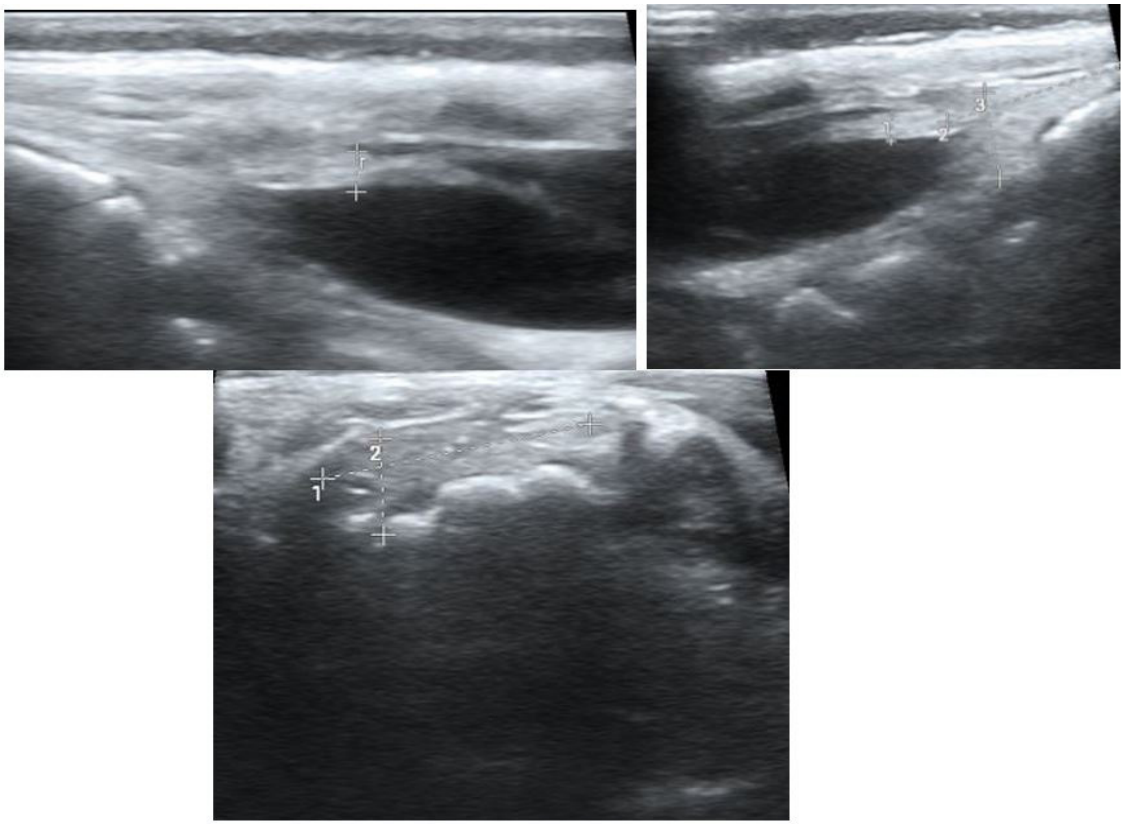

Figure 3: *Medullar ultrasound showing a lipomyelocele associated with a low-lying cord.*

flaring of the ribs. Our case showed deformities in vertebra and ribs unilaterally with fan like malformed ribs, thus being consistent with SCD. The clinical findings are an abnormality in the spine and chest. The patient has a short trunk dwarfism and a short neck with restricted mobility and limbs that appear disproportionately long. The small malformed chest cannot expand for the developing lungs resulting in reduced lung capacity, difficulty in breathing, and repeated infections (pneumonia) of the lungs, which can be mild, moderate, or life threatening.

Other anomalies associated with JLS include inguinal and umbilical hernias, neural tube defects, minor facial dysmorphism, anomalies of the digit and the lower limbs, urogenital and anal malformations, and complex congenital heart disease. Ultrasound could reveal the prenatal diagnosis at 16 weeks of gestation; the diagnostic criteria could be unpaired or poorly formed vertebrae, indistinct or fused posterior ribs, irregular short 'pebble-like' appearance of the spine, short trunk, protuberant abdomen, hernias, normal amniotic fluid, normal limb length and normal bi-parietal diameter. The treatment

of JLS cases depends on the degree of skeletal deformity (especially of the chest) and thoracic insufficiency. Initial management aims at aggressive neonatal care, by providing respiratory support to prevent pulmonary infections and avoid respiratory compromise, which is the primary cause of mortality and morbidity in infancy. It requires collaborative efforts from pediatric, orthopedic, and thoracic surgeons. Although modern technologies have improved the survival of infants with JLS, however, those who survive beyond childhood have progressive scoliosis, neurological dysfunction, and paraplegia secondary to spinal cord compression.

Conclusion

The paper reports a rare case of Jarcho-Levin syndrome associated with neural tube defects and other congenital malformations, presenting with respiratory distress.

Jarcho–Levin syndrome (JLS) or spondylothoracic dysostosis is a genetic skeletal disorder characterized by specific vertebral and rib anomalies, resulting in respiratory compromise.

Radiological tests are very useful in the initial differentiation, but genetic testing is the most important for making an accurate diagnosis. It will also make it easier for the family to make decisions about further procreation.

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