Case Report

Spondylocostal dysplasia – Jarcho–Levin syndrome: A case report

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ABSTRACT

Jarcho–Levin syndrome (JLS) refers to a rare congenital disorder, inherited in an autosomal recessive pattern, which presents as a spectrum of clinical and radiographic abnormalities of the spine and chest. The precise genetic basis of JLS is not clear, hence diagnosis is usually made by typical clinical features such as malformed ribs which are malaligned, crowded, fused, and bifid with a posterior symmetric fusion of all the ribs at the costovertebral joints and flared anteriorly giving "crab-like" or "fan-like" appearance to the chest. Here, we report the case of a baby boy who presented with respiratory distress and was noted to have the characteristic skeletal abnormalities, which include congenital scoliosis with vertebral anomalies, multiple fused ribs with congenital heart disease large 7-mm apical ventricular septal defect (VSD), multiple mid-muscular VSD with increased pulmonary blood flow, 5 mm ostium secundum atrium septal defect, and left ventricular (LV) dilatation with LV failure. The diagnosis of JLS is made if there is a characteristic physical appearance with radiological findings in the clinical setting of thoracic insufficiency. In this case report, we intend to bring forth the learning point that, when a baby is presented with respiratory distress and vertebral, as well as, thoracic cage malformations, the diagnosis of JLS should be kept in mind, if left untreated there is a chance of respiratory failure and thereby making it potentially fatal.

Key words: Jarcho–Levine syndrome, Spondylocostal dysplasia, Thoracic deficiency, Vertebral anomalies

Jarcho–Levin syndrome (JLS) is an autosomal recessive disorder, which presents as a spectrum of clinical and radiographic abnormalities of the spine and chest associated with respiratory insufficiency [1]. It was first described in 1938 by Jarcho and Levin in cases of thoracic insufficiency due to vertebral and rib anomalies [2,3]. It is characterized by a short neck, short trunk, normal-sized limbs, abnormalities of vertebrae, and ribs on the skeletal survey [4].

CASE REPORT

A full-term baby boy was born to a 30-year-old primi gravida mother with protected serologies and an uneventful antenatal period by emergency cesarean section in view of non-progression of labor. His birth weight was 3.35 kg. The APGAR scores were 8/10 and 9/10 at 1 min and 5 min, respectively. Antenatal scans were normal other than a suspected scoliosis in the baby. He was the first-born offspring of non-consanguineous parents and there was no family history of similar abnormalities. The baby was shifted to the neonatal intensive care unit in view of respiratory distress.

On examination, the baby was in respiratory distress and had thoracolumbar scoliosis. The baby required respiratory support in the form of non-invasive positive pressure ventilation initially which was gradually weaned off to room air. A skeletal survey showed multiple butterfly vertebrae and hemivertebrae noted from T3 to T12, scoliosis with convexity to the right side and multiple fused ribs (4^{th}, 5^{th}, 6^{th} ribs) on the left side (Fig. 1). Neurological examination was essentially normal. Eye examination showed extensive areas of subretinal pigmentary changes in both eyes. Echocardiogram showed a large 7 mm apical ventricular septal defect (VSD), multiple mid-muscular VSD with increased pulmonary blood flow, 5 mm ostium secundum atrium septal defect, and left ventricular (LV) dilatation with LV failure.

The baby required continuous positive airway pressure support initially, which was tapered to high-flow humidified oxygen support, and was maintaining saturation in room air by day 28 of age. During the course of the hospital stay, he also received intravenous fluid and electrolyte supplementation and by the time of discharge, the baby was on oral feeds and was gaining weight. His LV dysfunction was managed with diuretics and fluid restriction.

DISCUSSION

JLS is a rare, congenital disorder, inherited in an autosomal recessive pattern that represents a spectrum of clinical and radiographic abnormalities of the spine and chest [4].

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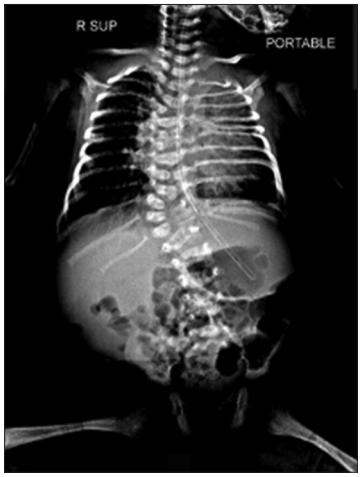

Figure 1: Baby gram showed multiple butterfly vertebrae and hemivertebrae noted from T3 to T12. Scoliosis noted with convexity to the right side, and multiple fused ribs 4th, 5th, and 6th ribs on the left side

Diagnosis can be made by typical clinical features. Chest X-ray shows malformed ribs such as mal-aligned, crowded, fused, and bifid with a posterior symmetric fusion of the ribs at the costovertebral joints, which are flared anteriorly giving a "crab-like" or "fan-like" appearance to the chest [1]. The precise genetic basis of JLS is not clear but it has been attributed to a mutation in one of at least five different genes, specifically DLL3, MESP2, LFNG, HES7, and TBX6 [5,6]. Due to the absence of clear genetic markers for JLS, diagnosis is made by physical appearance, characteristic symptoms, a detailed patient and family history, and a thorough clinical examination and imaging [7]. It can be picked up on antenatal fetal ultrasound (anomaly scan) due to some defects associated with the spine and chest, such as irregular short "pebble-like" spine with poorly formed vertebrae, normal amniotic fluid, standard limb length, and biparietal diameter [4]. The differential diagnosis of spondylocostal dysplasia includes campomelic dysplasia, Klippel–Feil syndrome, VACTERL anomaly, etc.

The estimated incidence of JLS is 1 in 40,000 births globally; however, the exact prevalence is unknown due to its rarity and misdiagnosis [8]. Asghar *et al*. reviewed multiple cases of JLS presented with multiple other concurrent anomalies including neural tube defects, caudal regression, congenital heart defects, abnormal airways, diaphragmatic hernias, and renal anomalies [8,9]. In our case, the only association was congenital heart disease.

CONCLUSION

The diagnosis is based on clinical and radiological findings with characteristic physical appearance and symptoms of thoracic insufficiency. In a neonate with respiratory distress syndrome, costovertebral assessment becomes the most important, with the intention of excluding syndromes associated with defects in the costovertebral segmentation, such as JLS, which causes respiratory impairment that can even lead to death by respiratory failure.

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