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

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].
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.

**REFERENCES**