Jarcho-Levin Syndrome with Splenic Herniation: A Rare Presentation

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Conflict of interest: None declared

Patient: Female, 5
Final Diagnosis: Jarco-Levine syndrome
Symptoms: Respiratory distress
Medication: —
Clinical Procedure: Supportive management
Specialty: Pediatrics and Neonatology

Objective: Congenital defects/diseases
Background: Jarcho-Levin syndrome, also known as spondylothoracic dysplasia and spondylocostal dysplasia, is characterized by varieties of vertebrae and rib anomalies. Jarcho-Levin syndrome is a clinical-radiological diagnosis with clinical evidence of short neck, short trunk, normal-sized limbs, or increased arm span, and vertebral and rib defects on the skeletal survey.

Case report: About 400 cases have been reported in world literature and 18 in our Indian literature. We report the case of a one-day-old female baby with a short trunk, short neck, low hairline, apparently long limbs, protuberant abdomen, mild midfacial dysmorphism, low-set ears, and a high-arched palate. There was one cystic swelling over the lateral side of the left hypochondrium sized about 3×3 centimeters, non-pusatile; the skin over the swelling was normal color and free. Radiological findings showed crowding of ribs with pebble-like appearance of the vertebrae and diastematomyelia of the spinal cord (type 2). We report here the first case of Jarcho-Levin syndrome with splenic herniation. To the best of our knowledge there have been no case reports of Jarcho-Levin syndrome with splenic herniation in the literature.

Conclusions: Jarcho-Levin syndrome can be easily diagnosed by clinical-radiological findings in newborns, with short trunk having a high index of suspicion. Prenatal diagnosis using level 2 ultrasonography can make it easier to manage the baby after delivery. Management should be from the basic neonatal care to prevention and immediate treatment of recurrent respiratory infections. Spinal surgical intervention to improve the thoracic volume and hence decrease the pulmonary restriction has been tried.

MeSH Keywords: Dysostoses • Spinal Dysraphism • Wandering Spleen
Abbreviations: STD – spondylothoracic dysostosis; SCD – spondylocostal dysostosis; USG – ultrasonography; HRCT – high-resolution computed tomography
Background

Jarcho-Levin syndrome is a clinical-radiological entity characterized by a short neck, short trunk, normal-sized limbs, and multiple vertebral and rib defects on the skeletal survey. This syndrome was first described by Jarcho and Levin in 1938 [1] and has been divided into two major subtypes: spondylolothoracic dysostosis (STD) and spondylocostal dysostosis (SCD). The purpose of reporting this case is to bring clinical understanding to this rare disorder.

Case Report

A full-term, apparently healthy female baby of a nonconsanguineous marriage was delivered by caesarean section. She cried immediately after birth, with an APGAR score of 9/10 in the first minute and 10/10 in the fifth minute (birth weight 2700 grams). On routine examination a short trunk, short neck, low hairline, apparently long limbs, protuberant abdomen, mild midfacial dysmorphism, low-set ears, and high-arched palate were seen (Figure 1). The baby had two umbilical arteries and one vein. There was one cystic swelling over the lateral side of the left hypochondrium sized about 3×3 centimeters, nonpusatile; the skin over the swelling was normal color and free (Figure 2). The mother breastfed the baby after 1 hour and the baby fed well. But after 2 hours, the baby was brought to the nursery with complaints of gradually developing fast breathing. The baby was pink, with heart rate 132/minute, respiratory rate 88/min with nasal flaring, saturation 97%, and capillary refill time <3 seconds. On auscultation bilateral fine crepts were heard. The baby was kept on warm and moist oxygen FiO2 40%, and very soon settled to a respiratory rate of 68/minute with no nasal flaring and saturation 99%. On day two of life, the baby’s length was 45 centimeters, head circumference was 34 centimeters, upper/lower segment ratio was 1:27, and arm span was 58 centimeters. An infantogram showed classical radiological features of SCD: pebble-beach appearance of the vertebrae, crowding of ribs with a crab-like appearance (left side > right side), and scoliosis (Figure 3). An ultrasonogram (USG) showed diastemetomyelia of the spinal cord, and the cystic swelling on the left lateral hypochondrium was found to be herniation of the spleen (diaphragm intact) due to crowding of ribs. Abdominal, pelvic, and cranial USG showed normal results. Blood biochemistry showed normal renal function and normal blood gases. No microorganism was cultured from blood. Echocardiography showed situs solitarius; high-resolution computed tomography (HRCT) of the thorax showed diastemetomyelia of the spinal cord of type 2 involving D3 to D8 without any tethering of the cord, no other associated central nervous system anomalies, crowding of ribs, and mild hypoplasia of the left lung with splenic herniation. Magnetic resonance imaging (MRI) of the
spine was refused by the parents. The baby was discharged on day 8 of life after routine immunizations. The infant has been followed up for the last 9 months. She is doing well and has had an adequate weight gain.

Discussion

Jarcho-Levin syndrome is characterized by congenital rib and vertebral anomalies leading invariably to short trunk. Only 18 cases of Jarcho-Levin syndrome have been reported in the Indian literature. It can be inherited as autosomal dominant or recessive. It does not show any relationship with consanguinity of marriage. Radiologically the vertebral anomalies range from absent vertebrae to splitting of the cord [2–4]. Ribs show also a vivid presentation; they might be absent, fused, or bifurcated. The pebble-beach appearance of vertebrae and crab-like or fan-like appearance of the thoracic cavity are very specific radiological findings associated with Jarcho-Levin syndrome.

The case reported here had clinical-radiological features of Jarcho-Levin syndrome. A nonspecific unusual splenic herniation found in our case was due to the abnormal orientation of the left lower lateral ribs. The anomalies associated with Jarcho-Levin syndrome are minor facial dysmorphism and diastematomyelia of the spinal cord. This patient with Jarcho-Levin syndrome did not show other system involvement like genitourinary abnormalities, anorectal malformations, congenital cardiac disease, or any limb malformations that can be associated with the condition.

Jarcho-Levin syndrome patients can have restrictive diseases of the lungs because the chief abnormality lies in the thoracic cavity and can be associated with lung hypoplasia. Pulmonary function restriction leads to recurrent chest infections.

Jarcho-Levin syndrome can be classified into two subtypes: (1) STD and (2) SCD. These classifications are based on skeletal anomalies.

STD shows autosomal recessive inheritance. Posteriorly it shows normal and symmetrical costovertebral joints. Anteriorly vertebrae show fragmentation over the length of the spine. The ribs are normal in number, shape, and joint formation both anteriorly and posteriorly. In STD, death is seen in the neonatal age, mostly due to restrictive lung functions and pulmonary hypertension.

SCD shows both autosomal recessive and dominant inheritance. This variety is characterized by rib abnormalities, i.e., crowding of ribs either due to absence or abnormal position, or fusion or bifurcation in addition to vertebral anomalies [5,6]. Prognosis is better in this variety, especially if patients cross six months of age [7].

Recently, after identification of 4 genes for SCD (DLL3, MESP2, LFNG, and HES7) SCD was classified into four types with specific characteristics: SCDO1 (DLL3-associated SCDO), SCDO2 (MESP2-associated SCDO), SCDO3 (LFNG-associated SCDO), and SCDO4 (HES7-associated SCDO) [8].

Our case of Jarcho-Levin syndrome was of the SCD type, with both vertebrae and rib anomalies. Molecular genetic testing was not done so we were unable to subcategorize the SCD. Ultrasound can be done near 16 weeks of pregnancy for prenatal diagnosis. Ultrasound criteria for diagnosis are unpaired or poorly formed spine, absent or fused ribs, pebble-like appearance of the spine, short trunk, protuberant abdomen, small thoracic cavity, and increased nuchal translucency [6].

Counseling the affected family is not a simple task because of the varied presentation and striking intrafamilial variability. The exact clinical-radiological diagnosis for molecular diagnosis is essential.

The prognosis is poor, but not consistently lethal. For the better survival of Jarcho-Levin syndrome patients, preventive or early diagnostic methods should be used for pulmonary complications (recurrent pneumonia), congestive heart failure, and pulmonary hypertension [9].
Conclusions

Jarcho-Levine syndrome is underdiagnosed, although it has distinctive clinical and radiological features. Prognosis of Jarcho-Levin syndrome is poor; about 81% patients die in infancy due to respiratory insufficiency. However, it is not an invariably lethal condition. There is 56% survival among the patients prospectively evaluated for intelligence, chest infections, congestive heart failure, and pulmonary hypertension. Jarcho-Levin syndrome patients need sympathetic and supportive treatment. Genetic counseling may be of benefit in subsequent pregnancies.

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