Lethal Progressive Thoracic Insufficiency in a Neonate Due to Jarcho Levin Syndrome

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ABSTRACT

A rare case of Jarcho Levin syndrome (JLS) presenting as a lethal progressive respiratory insufficiency in early neonatal period is reported. The neonate had classical features of this syndrome including vertebral segmentation defects, typical costo-vertebral fusion defects and scoliosis resulting in small thoracic volume and limited chest expansion; all consistent with a clinical diagnosis of JLS with thoracic insufficiency. In addition, our case had a rare association of dextrocardia and acyanotic congenital heart disease.

Key words:
Dextrocardia, Jarcho Levin syndrome, respiratory insufficiency, vertebral segmentation, ventricular septal defect

INTRODUCTION

Extensive deformities of the thoracic cage may affect the function and growth of lungs leading to its inability to support normal respiration. Jarcho Levin syndrome (JLS) is a rare lethal cause of such respiratory insufficiency. It is characterized by vertebral segmentation defects leading to congenital scoliosis and fusion of ribs. It was first reported by Jarcho and Levin in 1938. We present one such typical case of JLS with associated cardiac defects presenting in early neonatal period.

CASE REPORT

This was a case report of a 2.2 kg girl born at 34 weeks to an unbooked primi mother out of a non-consanguineous marriage presented on 2nd day of life with severe respiratory distress and limited visible chest expansion requiring mechanical ventilation. Baby had short trunk with scoliosis to the right. There was no apparent dysmorphism. There was no history of similar bony defects in any member of the family. Her weight and head circumference were 2.2 kg and 31 cm, both corresponding to 50th centile while the length of 31 cm was less than 3rd centile. Upper to lower segment ratio was 1.3:1 (normal is 1.7:1) suggestive of short trunk dwarfism. Skeletal survey revealed a small volume rib cage with crab like appearance [Figure 1]. Four ribs were absent on the left side and 3 on the right side. There was bilateral asymmetric costo-vertebral fusion of ribs with scoliosis to the right and hemivertebrae in cervical and thoracic spine [Figure 2]. There were no intrinsic abnormalities of ribs such as bifid, fused or broadened ribs. Vertebral segmentation defects were seen throughout the spine. Cardiac examination and echocardiography revealed dextrocardia and 8 mm ostium secundum atrial septal defect and a 2.5 mm patent ductus arteriosus, both with a left to right shunt. Ultrasound abdomen, magnetic resonance imaging brain, screen for bacterial/intrauterine infections and karyotyping were all normal. Baby succumbed due to progressive respiratory failure over next 2 days. A clinical diagnosis of JLS with lethal progressive respiratory insufficiency, dextrocardia and an acyanotic congenital heart disease was made.

DISCUSSION

Thoracic skeletal anomalies present as a rare cause of progressive respiratory insufficiency. Extensive vertebral and rib anomalies like JLS, Juene syndrome and Ellis Van Crevald syndrome affect thoracic function and growth, which in turn affects lung growth. Shortening of the thoracic cage due to these anomalies leads to thoracic insufficiency. Our case was befitting into the diagnosis of a severe lethal form of JLS with thoracic insufficiency due to the presence of vertebral segment defects throughout the spine, asymmetric bilateral costo-vertebral fusion of ribs and scoliosis with limited thoracic volume and expansion leading to progressive respiratory failure and death in the early neonatal period.
JLS was first reported in 1938 in Puerto Rican origins by Jarcho and Levin.\(^{[2]}\) They described a syndrome with a spectrum of vertebral segmentation defects and rib anomalies. Later in an article in 1996 Mortier et al. classified congenital vertebral segmentation defects into three subtypes - JLS, spondylothoracic dysostosis (STD) and spondylolocostal dysostosis (SCD).\(^{[3]}\) Individuals with STD have an autosomal dominant inheritance pattern. Vertebral segment anomalies in STD spare the sacrococcygeal region and they do not have intrinsic rib anomalies. SCD is a benign form and follows autosomal recessive inheritance. It is characterized by involvement of > 10 contiguous vertebral segments, pebble beach appearance of vertebrae on X-ray, involvement of sacrococcygeal region with intrinsic rib anomalies.\(^{[4,5]}\) JLS is a lethal subtype which presents with vertebral segmentation defects like hemivertebrae or block vertebrae throughout the spine with fusion of ribs at costovertebral junction bilaterally asymmetrically leading to crab like appearance on X-ray. There is the presence of scoliosis and absence of ribs without intrinsic rib anomalies such as bifid, broadened or fused ribs. Diagnosis is essentially clinical. They usually succumb to respiratory insufficiency due to restrictive lung disease. Usually death due to respiratory insufficiency in the severe form of this condition commonly occurs within the first 2 years of life.\(^{[6]}\) However, our case presented and succumbed early in the neonatal period itself.

JLS has been reported to be associated with cardiac, urogenital, digital and neural tube defects.\(^{[7-10]}\) In a review, Hatakeyama et al.\(^{[11]}\) reported association of congenital heart disease in 9 out of 87 cases of JLS. Most common among these were heterotaxia. Our case had dextrocardia, atrial septal defect and patent ductus arteriosus.

JLS is an important cause of progressive thoracic insufficiency often with lethal outcome. Diagnosis of this syndrome is purely clinical based on short trunked dwarfism associated with typical vertebral segmentation and costo-vertebral rib fusion defects. Association with cardiac defects is also known.

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**How to cite this article:** Bhutia E, Maria A, Verma A, Sethi SK. Lethal progressive thoracic insufficiency in a neonate due to jarcho levine syndrome. J Clin Neonatal 2014;3:49-50.

**Source of Support:** Nil, **Conflict of Interest:** None declared.