



ORIGINAL RESEARCH PAPER

Paediatrics

A CASE STUDY OF JARCHO-LEVIN SYNDROME IN TERM BABY

KEY WORDS:

Spondylocostal dysostosis, Jarcho-Levine Syndrome, Skeletal dysplasia, Rib anomaly, Clinical-Radiological findings.

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ABSTRACT

Jarcho-Levin Syndrome or spondylocostal dysostosis, is a rare axial skeleton anomaly due to mutation in genes involved in somatic segmentation during embryogenesis. We describe a term newborn baby with mid thoracic lordotic deformity with skeletal features of multiple rib anomalies and vertebral defects, but with normal limb lengths. The baby was noted to have poor respiratory effort at birth with meconium aspiration. This syndrome is under diagnosed and we wish to highlight the simple process involved in making a clinico-radiological diagnosis.

INTRODUCTION

Jarcho-Levin Syndrome, also known as Spondylocostal dysostosis is an autosomal recessive condition first described by Jarcho-Levin in 1938.⁽⁶⁾ Jarcho-Levin Syndrome or Spondylocostal dysostosis is a rare axial skeleton anomaly due to mutation in genes involved in somatic segmentation during embryogenesis. It is a rare congenital disorder, inherited in autosomal recessive pattern which represents a spectrum of clinical and radiographic abnormalities of spine and chest. Jarcho-Levin Syndrome is characterised by short thorax and neck, protruding abdomen, abnormal vertebral segmentation and fusion of posterior costal vertebral segment which results in thoracic restriction or respiratory failure and scoliosis. This structural abnormality can result in significant respiratory problems due to restricted space for lung development and expansion. This syndrome presents in early infancy, often with respiratory distress as a main feature. The severity of symptoms varies with some patients requiring intensive medical support.

Case Study

A female infant was delivered at term to primigravida mother with protected serologies via emergency cesarean section in view of non progression of labor. The baby was born with a birth weight of 3.1 kg. The APGAR Scores were 8/10 and 9/10 at 1 min and 5 min respectively. She was first born offspring of non consanguineous parents and there was no family history of similar abnormalities. The baby was noted to have poor respiratory efforts at birth with thick meconium stained amniotic fluid present.

At birth, the neonate exhibited signs of severe respiratory distress including grunting and cyanosis. The infant had a short neck, disproportionately shortened neck, protuberant abdomen, polythelia, prominent left lower rib and thoracic asymmetry. No limb length discrepancy was noted at birth. The baby required respiratory support in the form of high flow humidified oxygen support via nasal prongs which was gradually weaned off to room air during the course of hospital stay. She also received intravenous fluid and electrolytes supplementation.

A chest x-ray revealed diffused patchy infiltrates consistent with meconium aspiration syndrome. Additionally the chest x ray also showed multiple vertebral anomalies including fused and hemivertebrae along with abnormal rib morphology consistent with Jarcho-Levin syndrome. Echocardiography ruled out congenital heart disease and USG Abdomen and Cranium was normal.

The above findings were consistent with clinico-radiological diagnosis of Jarcho-Levin Syndrome. Parents were offered genetic counselling and orthopaedic consult for the baby. Baby was discharged and under regular follow up.



Figure 1: Neonate with left sided polythelia. Short neck, short trunk and protuberant abdomen are also noted.



Figure 2: Radiograph of the neonate showed multiple vertebral anomalies including fused and hemivertebrae along with abnormal rib morphology.

DISCUSSION:

Congenital vertebral segment anomalies presents with wide variety of well characterised disorders but many times with poorly understood phenotypes.⁽²⁾ Any associated anomaly of thorax with spine processes is unique clinical and surgical challenge. Jarcho, a pathologist and Levin, a neurologist in 1938 described two children from puerto rico with short neck, abnormal vertebral segmentation and irregularly aligned ribs with normal skull and long bones. Since then Jarcho-Levin Syndrome has been used for any form of costovertebral malformation.⁽⁶⁾ The inheritance is autosomal recessive with several genes associated with NOTCH signaling pathway implicated, DLL3(most common), MESP2, LFNG or HES7 genes but rarely by autosomal dominance due to mutation in TBX6 gene.1 It is a rare skeletal dysplasia affecting the axial skeleton with worldwide incidence of 1 in 40000 births.⁽¹⁾

Clinically, diagnosis of spondylocostal dysostosis is suggested by a short webbed neck, short trunk, protuberant abdomen and scoliosis. Brachecephaly, wide nasal bridge, upslant palpebral fissures, low posterior hairline , pectus carinatum, lordosis, inguinal or umbilical hernial may also be present in some cases.⁽⁴⁾ Skeletal malformations predominantly affect the chest wall and spine, sparing the extremities. Multiple segmentation defects of the vertebrae in combination with abnormalities of ribs which are maligned, brodened, bifid with variable number of irregular intercostal rib fusions and sometimes with a reduction in rib number, giving it like a crab like configuration.⁽¹⁾⁽⁴⁾

As per Asghar Zameer criteria, our infant fits into definite clinical diagnosis of JLS.

Major criteria
1. Vertebral anomalies including hemivertebrae, vertebral agenesis, butterfly vertebrae, hypoplastic vertebrae, costovertebral fusion defects.
2. Rib anomalies including malalignment, crowding, fanning-out, fusion, absence.
3. Genetic evaluation showing mutation in <i>MESP2</i> , <i>DLL3</i> , <i>IFNG</i> , <i>HES7</i> , or <i>PSEN1</i> genes.
4. Prenatal sonographic features including costovertebral segmentation, indistinct or fused posterior ribs, irregularly shortened spine, short trunk, and short neck.
Minor criteria
1. Child born of a consanguineous marriage.
2. Having a sibling diagnosed with Jarcho-Lavin syndrome.
A definite diagnosis of JLS is made on the presence of two or more major criteria.
A diagnosis is probable in the presence of one major and two minor criteria.

The major cause of morbidity and mortality in spondylocostal dysostosis is thoracic insufficiency leading to decrease lung volume and underdevelopment leading to recuurent lung infections.⁽⁶⁾ Treatment depends upon the degree of thoracic insufficiency, concurrent pulmonary complications, presence of chest wall defects and quality of life. Non severe cases may survive to adulthood, without any requirement of surgical interventions.

CONCLUSIONS

JLS is a unique skeletal dysplasia involving only the axial skeleton and routine survey may miss the crucial external findings. The characteristic features of asymmetrical ribs and vertebral anomalies helps in clinico-radiological identification of this syndrome and needs to be differentiated from it's ominous close differential diagnosis spondylothoracic dysostosis which has similar fan like configuration of ribs, but with a higher degree of respiratory embarrassment and presence of cervicothoracic fusion with

rigid neck point. These infants need life long follow up due to their higher predilection for pulmonary infections.

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