

A Case Study

A case of Jarcho-Levin syndrome with neural tube defect

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Abstract

Jarcho–Levin syndrome is a rare congenital disorder inherited in an autosomal recessive pattern, primarily identified through clinical assessment and radio-logical imaging. It is characterized by malformations of the spine and ribs, producing a distinctive short-trunk stature. Posterior rib fusion with anterior flaring creates the classic crab-like appearance, while the resulting thoracic constriction limits lung growth and compromises intrathoracic organ function. This restriction leads to thoracic insufficiency, manifesting as recurrent respiratory infections, progressive cardiac strain, and severe pulmonary complications. Additional anomalies involving the central nervous system, genitourinary tract, and heart have also been described. The present case illustrates the life-threatening potential of this syndrome and underscores the importance of early recognition to optimize respiratory support and guide genetic counseling and family planning.

Introduction

Jarcho–Levin syndrome, first described by Saul Jarcho and Paul Levin in 1938 (1), is a rare congenital disorder characterized by complex vertebral and rib anomalies. Inheritance may follow autosomal recessive or dominant patterns, with mutations identified in genes such as *DLL3*, *MESP2*, *LFNG*, *HES7*, and *TBX6*. Here we describe a newborn diagnosed with Jarcho–Levin syndrome based on clinical and radiological findings.

Case

Our case is a one month male neonate born to a 30 year old para 2 sometimes abbreviated as G2 P2 mother at 39 weeks of gestation, with APGAR scores of 8 and 9. The neonate was referred to Yekatit 12 Hospital Medical College from Abebech Gobena Primary Hospital for myelomeningocele repair—a severe neural tube defect resulting from incomplete closure of the spinal cord during early fetal development, usually within the first month of gestation. At Abebech Gobena Primary Hospital, the patient had been treated for neonatal sepsis after developing fast breathing and fever one day after delivery, along with a swelling over the back noted since birth. Initial laboratory values showed WBC $19.3 \times 10^9/L$ and platelets $317 \times 10^9/L$. Trans-fontanelle ultrasound was normal. The neonate was treated with ampicillin and gentamicin, and after completion of therapy was referred to Yekatit 12 Hospital Medical College for neurological intervention.

At Yekatit 12 Hospital Medical College, the patient was seen at the pediatric outpatient department. On examination, head circumference was 32 cm, weight 3 kg, and height 47 cm. The infant appeared well and was not sick-looking, with no signs of respiratory distress. Vital signs were within expected ranges: heart rate 150 beats per minute, respiratory rate 58 breaths per minute, axillary temperature 36.5 °C, and oxygen saturation 96%. Chest examination revealed asymmetrical expansion, but bilateral air entry was not decreased. Examination of the lower back revealed a well-defined, mobile, cystic mass measuring 4 × 3 cm. As part of the pre-anesthesia workup, chest X-ray and abdominal ultrasound were performed. The abdominal ultrasound was unremarkable, while the chest X-ray showed a right hemithorax abnormality with T4 and T5 hemivertebrae.

The mother first became aware of her pregnancy at approximately two months' gestation and initiated antenatal care at that time. Her follow-up visits were limited, with the next documented ANC occurring at nine months of pregnancy. During her care, she received daily supplementation with iron and folic acid. She does not recall receiving any vaccinations during pregnancy. An obstetric ultrasound performed during the first trimester at a private facility was unremarkable, and no other ultrasound was performed afterward. There was no significant family history of congenital anomalies, nor evidence of consanguineous marriage.

The family was counseled that the infant required surgery for the neural tube defect and informed about the additional abnormalities seen. They were also advised about possible intraoperative and postoperative complications. After counseling, the family deferred surgical management.



Figure 1. A 4x3cm cystic mobile mass over lower back

Discussion

Jarcho and Levin first described JLS in 1938 while studying cases of thoracic insufficiency due to vertebral or ribs abnormalities (1). It is considered to be a congenital condition that is inherited in an autosomal recessive pattern (2). The most common gene mutation is DLL3 gene mapped to the 19q13.1-q13.3 regions. However there are other genes presumed to be responsible (3). It is a very rare disease and the global prevalence is not well known (4).

JLS has two distinct types; spondylocostal dysostosis (SCD) and spondylothoracic dysplasia (STD). Spondylothoracic dysplasia is characterized by defect in segmentation of the vertebral bones and fusion of the associated ribs. In addition in spondylothoracic dysplasia there is shortening of the thoracic vertebrae. In contrast, in spondylocostal dysostosis there is an intrinsic rib anomaly such as bifurcation, broadening and fusion independent of the vertebral anomaly (5).

JLS is diagnosed clinical and radiological findings. Clinically, it presents as a short trunk dwarfism and short neck and the limbs seem to be disproportionately long. Radiologically, in utero there might be irregular short spine with vertebral defect usually seen during second trimester (3). At birth, the patient will have kyphoscoliosis and at least ten contiguous thoracic spines might have a defect in segmentation. On the chest, the ribs might be bifid, crowded, malaligned and fused posteriorly at the costovertebral joints. Anteriorly the ribs get flared and give the appearance of a crab thus the term “crab like’ chest. In our case there is right hemi thorax crab like with T4 AND T5 hemivertebrae.

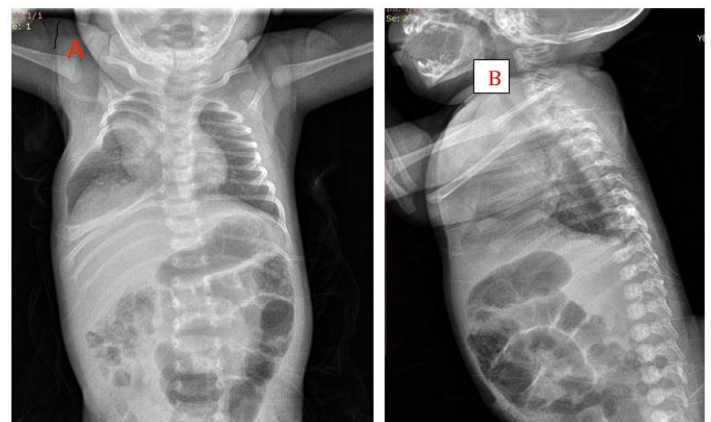


Figure 2. A. Right hemi thorax crab like with T4 AND T5 hemivertebrae. B. Prominent soft tissue density at the lower back

In some patients with Jarcho–Levin syndrome (JLS), concomitant anomalies such as neural tube defects may occur. Spina bifida occulta appears to be a relatively common finding in Jarcho–Levin syndrome. In our case, a cystic structure measuring 4 × 5 cm was identified.

The coexistence of Jarcho–Levin syndrome with neural tube defects has been described in the literature. The vertebral changes are thought to reflect an early developmental error during the fourth to eighth weeks of gestation, when multiple centers of cartilage normally form around the notochord to build the vertebral column. Failure of proper formation or fusion at this stage can result in hemivertebrae or butterfly vertebrae, with rib anomalies occurring as secondary consequences.

Reports from Indian journals have documented similar cases, including neonates with associated anomalies such as hydrocephalus, hydronephrosis, and meningomyelocele, further supporting the spectrum of developmental errors linked to Jarcho–Levin syndrome (6). Genitourinary (renal and urethral abnormalities, hypospadias) and cardiac (Atrial septal defects and other complex congenital heart diseases) have also been reported.

Many individuals with Jarcho–Levin syndrome will have cardiorespiratory complication because of the chest cavity volume restriction. These are recurrent pulmonary infections, pulmonary hypertension and heart failure (7). As a result, patient with this condition die at an early age,

however if milder form of the disease is present the patient might lead a healthy life (8).

Several syndromes can mimic Jarcho–Levin syndrome, including Alagille, VACTERL, Klippel–Feil, campomelic dysplasia, oculo-auriculo-vertebral spectrum, multiple pterygium syndrome, sirenomelia, and Robinow syndrome. These conditions typically present with vertebral anomalies but are distinguished by systemic or craniofacial features such as cholestasis and facies (Alagille), multi-system malformations (VACTERL), cervical fusion (Klippel–Feil), limb bowing and airway issues (campomelic), craniofacial microsomia, pterygia and contractures (multiple pterygium), limb fusion (sirenomelia), or mesomelic shortening and genital hypoplasia (Robinow). In our patient, thoracic hemivertebrae with right hemithorax asymmetry and a co-occurring myelomeningocele were present, while none of the systemic or craniofacial features of these mimics were identified, supporting the diagnosis of Jarcho–Levin syndrome (9).

Management of JLS depends on the severity of the condition particularly the chest deformity. Respiratory support starting at neonatal age might be necessary as well as treatment of lung infection since this is the primary cause of mortality. In our case, the patient has been admitted Neonatal Intensive care unit and put on antibiotics and respiratory support for neonatal sepsis of chest focus. As mentioned above, Jarcho–Levin syndrome can have associated Neural tube defect and other congenital anomalies, which should be managed accordingly. As the

child grows, modern orthopedic procedures can be done to improve chest restriction by placing vertical expandable prosthetic titanium rib (VEPTR). This can increase survival beyond early childhood. However these patients might have progressive scoliosis, neurological dysfunction and paraplegia secondary to spinal cord compression, etc.

Lessons Learned

This case highlights that Jarcho–Levin syndrome (JLS) often involves multisystem anomalies, with central nervous system, genitourinary, and cardiac malformations requiring active screening in affected neonates. Early recognition is essential, as concomitant neural tube defects may obscure the diagnosis if attention is directed only to the spinal mass. Equally important is respiratory vigilance, since thoracic insufficiency remains the leading cause of morbidity and mortality; timely respiratory support and aggressive infection control are therefore critical to improving survival and long-term outcomes.

Conclusion

Jarcho–Levin syndrome is a very rare disease. Defects in the rib and vertebrae render the patient to have cardio respiratory complication that can be fatal during early childhood. Jarcho–Levin syndrome can also present with other concomitant findings such as Neural tube defect, which can be a more striking feature and might obscure its diagnosis. Modern orthopedic and respiratory interventions have shown a significant improvement in survival as well as

quality of life to the patient affected by Jarcho–Levin syndrome.

Data Availability

The data used to support the findings of this study are available from the corresponding author upon reasonable request.

Ethical Approval

The report was conducted as per the Declaration of Helsinki. At Yekatit 12 Hospital medical college, the Institutional Review Board granted ethical clearance, including publishing this patient's case information. The patient's privacy and confidentiality were protected.

Consent

The patient gave written informed consent for this case report and any related images to be published.

Conflicts of Interest

The authors declare that they have no conflicts of interest.

Authors' Contributions

All authors contributed to data analysis, drafting, or revising of the article, have agreed on the journal to which the article will be submitted, gave final approval of the version to be published, and agree to be accountable for all aspects of the work.

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