Journal of Neonatal Surgery
ISSN(Online): 2226-0439
Vol. 14, Issue 11s (2025)
https://www.jneonatalsurg.com

OPEN ACCESS

Jarcho-Levin Syndrome Associated With Anorectal Malformation: A Rare Case Report

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Cite this paper as: Illona Okvita Wiyogo, IGB Adria Hariastawa, (2025) Jarcho-Levin Syndrome Associated With Anorectal Malformation: A Rare Case Report. *Journal of Neonatal Surgery*, 14 (11s), 27-30.

ABSTRACT

Introduction: Jarcho-Levin syndrome is a congenital disorder characterized most commonly by the presence of rib and vertebral defects at birth. The association with other organs such as the spinal, brain, and the urinary system has been reported. The paper reports a rare case of Jarcho-Levin syndrome associated anorectal malformation.

Case presentation: The case is a full-term boy which had a short neck with an undefined chest deformity and protuberant abdomen. He did not pass any stool since birth and had no anal opening. Early identification and management is crucial for optimal outcomes.

Conclusion: The paper reports a rare case of Jarcho-Levin syndrome associated with anorectal malformation. Early screening and diagnosis is important to determine further management.

Keywords: *Anorectal Malformation, Jarcho-Levin syndrome, infant, newborn*

1. INTRODUCTION

Jarcho-Levin syndrome is a congenital disorder characterized most commonly by the presence of rib and vertebral defects at birth. This syndrome most commonly presents in infancy and is characterized by identification of a short neck, short trunk, normal-sized limbs, and multiple vertebral and rib defects on the skeletal survey. Skeletal survey characterizes multiple vertebral anomalies at different levels of the spine, including "butterfly vertebrae," hemivertebrae, and fused hypoplastic vertebrae. This syndrome was first described by Jarcho and Levin in 1938 [1] and has been divided into two major subtypes: spondylothoracic dysostosis (STD) and spondylocostal dysostosis (SCD). Other abnormalities have also been described in Jarcho–Levin syndrome, including neural tube defects, Arnold–Chiari malformation, renal/urinary tract abnormalities, hydrocephalus, and hydroureteronephrosis [2]. The purpose of reporting this case is to bring clinical understanding to this rare disorder. In this report, we describe a 13-hour-old baby with JLS associated with anorectal malformation.

CASE REPORT

A male infant was born at 38-39 weeks of gestation and delivered by caesarean section due to prolonged labor. He cried immediately after birth, with an APGAR score of 6-7, birth length and weight are 48 centimeters and 3050 grams respectively. The baby did not look cyanotic and the amniotic fluid was clear. The infant had a short neck, protuberant abdomen, and no anal opening since birth. He was the third offspring of nonconsanguineous parents, her siblings were normal and there were no history of similar abnormalities in any of his family members. The mother, aged 37, did not find any abnormalities during prenatal examination.

Figure 1. Clinical presentation of the baby

The baby was reported to have an increasingly distended stomach since 10 hours after birth, he had an orogastric tube (OGT) placed with 2 ml output retention and been fasting ever since. The infant did not pass stool since birth and there was no history of meconium stains on diapers, however, clear yellow urine stains were visible equivalent to 2 ml/kg/hour. The baby looked icteric, with heart rate 150/minute, respiratory rate 48/minute, saturation 98% in room air, body temperature 36.8◦ C, and capillary refill time < 3 seconds.

Physical examination revealed an anal dimple with no visible opening for defecation around the genital. No fistula was found in the perineum. The external urethral orifice can be seen at the tip of the glans penis. Baby had normal cardiovascular and respiratory examination. Abdominal examination revealed distended abdomen with venectation, no bowel contour or movement is visible and normal umbilical stump. Laboratory findings showed normal blood count, with hemoglobin count 12.8 g/dL, leukocyte count 22.030 u/L, platelet count 242.000 u/L, prothrombin time 13.4 seconds, aPTT 35.1 seconds.

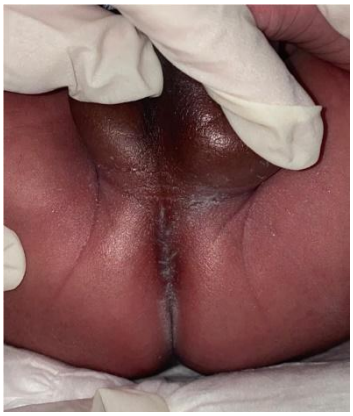

Figure 2. Genitalia examination shows the presence of anal dimple with no anal opening

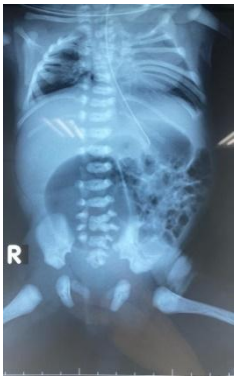

Figure 3. Babygram

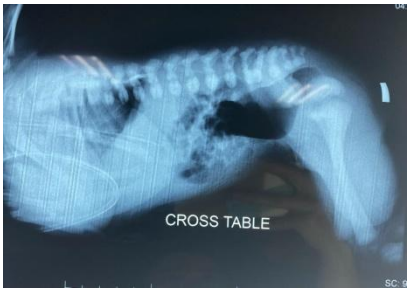

Figure 4. Cross table lateral to confirm position

Surgical consultation was taken. Baby underwent sigmoidostomy. Post operatively, hemodynamics were stable. Further treatment for Jarcho-Levin syndrome was not yet taken since there is no evidence of thoracic insufficiency. Caretakers were taught colostomy care and plan is to follow up with pediatric surgeon.

DISCUSSION

Jarcho-Levin syndrome is a rare genetic disorder characterized by malformation of bones in the spinal column and the ribs, leading invariably to short trunk. It can be inherited as autosomal dominant or recessive. Spondylothoracic dysostosis (STD) and spondylocostal dysostosis (SCD) are known as two different types of Jarcho-Levin syndrome. Due to either a spondylothoracic dysostosis or spondylocostal dysostosis variant, Jarcho-Levin syndrome results in a thoracic volume depletion deformity caused by shortness of the thoracic cavity. However, for years, the term Jarcho-Levin syndrome has been inaccurately applied to individuals affected with either SCD or STD. Jarcho-Levin syndrome (JLS) is now applied to the clinical phenotype of spondylocostal dysostosis (SCD), while spondylothoracic dysplasia/dysostosis (STD) wears a name of Lavy-Moseley syndrome (LMS) [4]. JLS has a more benign clinical course with a higher survival rate than STD. Typical clinical presentations of a patient with JLS are intrinsic rib malformations (broadening, fusion, missing ribs, abnormal orientation, bifurcation, irregularity of shape and size). The vertebral defects in both JLS and STD include a decreased number of vertebrae with segmentation and formation defects like block vertebrae, hemivertebrae, butterfly vertebrae, missing vertebra, and fused vertebrae. 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. Other associated anomalies with SCD include: Hernias, neural tube defects and anomalies of the anal opening, urinary tract, external genitalia, uterus and lower limbs may be associated [3, 7]. On this case, the patient has a mild form of JLS with the association of anomalies of the anal opening. The early management of a newborn with an anorectal anomaly is crucial, which can associate with abdominal distension, dehydration and sepsis. Initial resuscitation with intravenous fluid and broad spectrum antibiotics holds the key for the final outcome in such cases. After assessment of associated anomalies the child can be taken for a protective colostomy, followed by delayed repair later or a single staged definitive procedure can also be performed in selected cases. The clinical course and vertebral malformations are, also, more severe in patients with STD compared to SCD. Infants affected by this disorder have respiratory insufficiency, and other abnormalities such as recurrent respiratory infections are expected. In milder cases, surgery including the vertical expandable titanium rib (VEPTR) is a possibility, but in severe cases, prognosis is poor. Since this disorder can be an autosomal recessive or dominant trait, parents in this situation need genetic counseling to be aware of how this could affect future pregnancies [5]. 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. Thorough screening and other examinations could reduce their morbidity as more sophisticated imaging may be done to better visualize the anatomical malformation [6].

CONCLUSION

The paper reports a rare case of Jarcho-Levin syndrome associated with anorectal malformations. It is recommendable to confirm the type of anorectal malformation before deciding any operative intervention. Screening out for CHARGE and VACTERL association is needed. The management should be carefully customized. Jarcho-Levin syndrome patients and family need supportive treatment. Genetic counseling may be of benefit in subsequent pregnancies.

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