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Case Report

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Jarcho Levin Syndrome with open neural tube defect: A case report

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KEYWORDS

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Meningomyelocele,
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ABSTRACT

Background: Jarcho Levin syndrome is a rare genetic disorder characterized by spine abnormalities and the absence of ribs. It is associated with various congenital malformations and carries significant morbidity and mortality. The simultaneous presentation of this syndrome with neural tube defects is seldom encountered.

Case Presentation: We report a case of a 2-day-old male who presented with a lesion on his back since birth. Clinical and radiological examinations revealed thoracolumbar meningomyelocele with the absence of upper ribs on the right side and a patent foramen ovale. Subsequently, the patient underwent surgery for meningomyelocele, while the rib anomalies were managed conservatively.

Conclusion: Jarcho Levin syndrome is a rare disorder, and its association with meningomyelocele is extremely uncommon. It should be considered in the evaluation of a patient with a neural tube defect and respiratory distress.

INTRODUCTION

Spondylocostal dysostosis (SCD), commonly known as Jarcho Levin syndrome, is a rare genetic disorder [1]. It is characterized by malformed vertebrae and the congenital absence of ribs, resulting in a classical crab-like appearance in a chest radiograph [2]. To date, only a few cases of this syndrome have been reported worldwide. This rare entity is associated with other congenital malformations such as congenital heart disease, renal anomalies, and anorectal malformations. However, the association with neural tube defects has rarely been observed. In this article, we report on one such case that was successfully managed at our center.

CASE REPORT

A 2.5-kg newborn male presented to the emergency department at our institute on the second day of life with complaints of swelling over the back since birth. Antenatally, the mother was not evaluated, and no ultrasound was performed. The baby was born into a non-consanguineous marriage with young parents (father 21 years old, mother 20 years old). The baby was delivered via normal vaginal delivery at a private hospital. The baby did not cry immediately after birth and required oxygen support. Subsequently, the baby was referred to a government hospital, from where it was further referred to our tertiary care center.

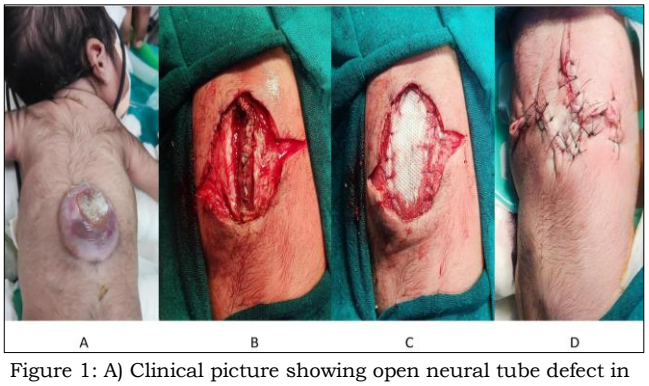

Figure 1: A) Clinical picture showing open neural tube defect in thoracolumbar region; B & C) intraoperative picture showing large defect covered with synthetic dura patch; D) skin coverage with rhomboid flap

On evaluation, there was a cystic swelling of size 7cm (V) x 5cm (H) occupying the thoracolumbar region. The swelling was covered with a thin membrane with some exposed neural elements in its center (Fig. 1A). There was no leakage of any fluid. Bilateral lower limb weakness (1/5) was observed with a lack of flexion at the hip and knee joints when the baby was lifted in

both arms. The anal sphincter was lax, and the bladder was palpable and expressible. The anterior fontanelle was flat, and the head circumference was 38 cm. Ultrasound (USG) cranium with a local region and KUB was performed. The USG cranium showed dilated bilateral lateral ventricles (24mm). The USG local region suggested a sac-like outpouching from the thoracolumbar region with an echogenic nerve root within. There was splaying of the posterior elements of the spine in the thoracolumbar region. USG KUB was reported as normal. In view of respiratory distress, a chest X-ray was performed, revealing the absence of upper ribs on the right side with an abnormal curvature of the spine [crab-like appearance] (Fig. 2A). An echocardiogram was also performed due to multiple anomalies, revealing a tiny patent foramen ovale (PFO) with a left-to-right shunt. To confirm the chest radiograph finding of the absence of ribs, a CT thorax with 3D (three-dimensional) reconstruction was performed, confirming the absence of 3rd to 5th ribs on the right side with fusion of 3rd to 5th ribs posteriorly on the left side and 6th to 8th ribs posteriorly on the right side. Additionally, there was fusion of the posterior elements of the T2-T3 thoracic vertebrae and splaying of posterior elements of the spine in the thoracolumbar region (Fig. 2B).

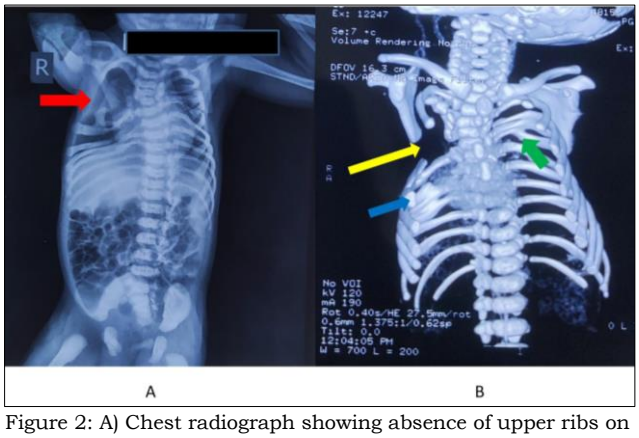

Figure 2: A) Chest radiograph showing absence of upper ribs on right side [red arrow]; B) CT thorax with 3D reconstruction showing absence of 3rd to 5th ribs on right side [yellow arrow] with fusion of thoracic vertebrae T2-T3 and fusion of 3rd to 5th ribs posteriorly on left side [green arrow] and 6th to 8th ribs posteriorly on right side [blue arrow]

With these findings, parents were informed about the baby's prognosis in terms of morbidity and mortality. After obtaining written and informed consent from the parents, the patient underwent excision and repair of the meningomyelocele. Intraoperatively, the sac membrane was excised, preserving the neural elements. The neural tube was created with a continuous interlocking absorbable suture. The dura defect measured 4 cm (V) x 2 cm (H) and was covered with a synthetic dura patch (Fig. 1B, C, & D). The baby started spoon feeding on postoperative day (POD) 1 and breastfeeding on POD 2. Gradually, oxygen support was weaned off, and the baby was successfully discharged with a plan for regular follow-up for hydrocephalus and respiratory problems.

DISCUSSION

Jarcho Levin syndrome (JLS) is a rare genetic disorder with autosomal recessive inheritance [3]. Initially described by Jarcho and Levin in 1938 [1], JLS, also known as spondylocostal dysostosis (SCD), is characterized by spine abnormalities, the absence of ribs, and malformed vertebrae, resulting in a crab-like appearance on a chest radiograph [4]. Due to its rarity, the exact prevalence of this genetic disorder remains unknown. To date, only a limited number of cases have been reported globally, with 400 case reports worldwide and 18 cases documented in Indian literature [5].

JLS is known to be associated with other congenital malformations, including congenital heart disease, renal anomalies, anorectal malformations, and neural tube defects, although such associations are observed in a small number of cases. Unfortunately, most affected infants face a grim prognosis, succumbing to recurrent pulmonary infections and respiratory insufficiency during infancy. The underdeveloped chests of these infants restrict lung expansion, leading to reduced lung capacity, breathing difficulties, and recurrent lung infections [6]. While the association of JLS with neural tube defects is infrequently reported, Dane B. et al. documented four cases of JLS with neural tube defects, and Rai N et al. reported a case of spondylocostal dysostosis associated with right-sided polythelia, right-sided rib deformity, and meningocele [7, 8].

Successful management of meningomyelocele in this case underscores the need for varied treatment approaches depending on the degree of skeletal deformity associated with JLS. Initial treatment focuses on respiratory support, while later stages may involve orthopedic procedures, including the use of prosthetics such as the vertical expandable prosthetic titanium rib (VEPTR), aiming to improve thoracic function [9].

The case history of the index baby sheds light on the challenges faced by young parents from rural backgrounds who often lack access to basic antenatal facilities, as evident in the absence of obstetric consultations and ultrasounds during pregnancy. A comprehensive approach involving education on anomalies and their link to nutritional deficiencies in females of reproductive age, coupled with enhanced antenatal health checkups and facilities like antenatal ultrasonography in rural areas, is crucial to address these healthcare gaps.

In conclusion, Jarcho Levin syndrome remains a rare disorder, and its association with meningomyelocele

is exceptionally rare. Awareness of this syndrome should be heightened during the evaluation of patients presenting with neural tube defects and respiratory distress. Additionally, addressing systemic issues, such as the lack of basic antenatal facilities in rural areas, is paramount for early detection and comprehensive management of congenital anomalies.

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