D J Med Sci 2024;10(2):71-73
doi: 10.5606/fng.btd.2024.155

Case Report

A rare case of Jarcho-Levin syndrome in the emergency room

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ABSTRACT

Jarcho-Levin syndrome is a rare genetic disorder in which abnormal costovertebral segmentation is seen and may be accompanied by neural tube defects and some anomalies involving the urinary and renal systems. In most cases of Jarcho-Levin syndrome, breathing difficulties occur due to rib deformity. Patients die due to respiratory insufficiency. In this case report, a 39-year-old patient presented to our emergency department for the first time at eight weeks of gestation with complaints of spotting. Due to its rarity, a case of a newborn diagnosed with spondylothoracic type Jarcho-Levin syndrome, accompanied by meningomyelocele and hydrocephalus, is presented.

Keywords: Hydrocephalus, Jarcho-Levin syndrome, meningomyelocele.

Jarcho-Levin syndrome was first identified by Saul Jarcho and Paul M. Levin in 1938. It is commonly inherited in an autosomal recessive pattern and has a prevalence of 2.5 per 100,000 individuals.[1]

It consists of two subgroups: spondylothoracic dysostosis (STD) and spondylocostal dysostosis (SCD). Spondylothoracic dysostosis is characterized by vertebral deformities and is more frequently associated with other anomalies, whereas in SCD, rib anomalies are more prominent.[2]

Jarcho-Levin syndrome, also known as spondylocostal dysostosis, describes a variety of short-trunk skeletal dysplasias characterized by rib anomalies and multiple segmentation defects of the vertebrae.[3]

CASE REPORT

A 39-year-old patient, gravida 3 para 2, presented to our emergency department at eight weeks of gestation for the first time with complaints of spotting. The patient had no known additional illnesses in her medical history. She had a history of two previous cesarean sections. Ultrasound (USG) revealed a pregnancy with a crown rump length: 8W+1, with positive fetal cardiac activity. The patient was prescribed progesterone, discharged, and advised to attend follow-up visits at the obstetrics clinic. However, the patient reported that she did not attend any follow-up visits until the 35th week of pregnancy. Upon her visit to the clinic, the USG examination revealed the presence of hydrocephalus, preventing visualization of the brain tissue, and the biparietal diameter was measured at 129 mm. A 43×48 mm anechoic cystic lesion was observed in the neck. Thoracolumbar spina bifida was observed, while the abdominal circumference and extremities were consistent with the gestational age. The patient was scheduled for an elective cesarean section at 37 weeks of gestation. However, she missed her appointment and returned to the emergency department at 39 weeks of gestation with complaints of contractions. Due to the patient's 3 cm dilation, she was taken for an emergency cesarean section. A 4090 g male baby with anomalies, including hydrocephalus (Figure 1a) and meningomyelocele, shown in Figure 1b, was delivered via Kerr + Tshot incision

Received: July 02, 2024
Accepted: July 19, 2024
Published online: October 01, 2024

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Cite this article as:
Pınar Cengiz Z, Yener C, Terzioğlu T, Ilhan C. A rare case of Jarcho-Levin syndrome in the emergency room. D J Med Sci 2024;10(2):71-73. doi: 10.5606/fng.btd.2024.155.

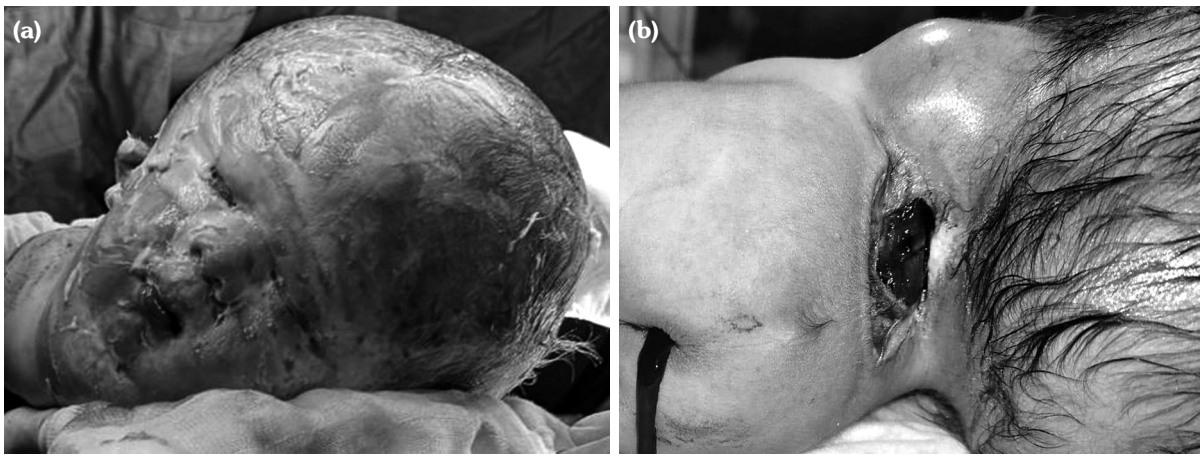

Figure 1. The baby with **(a)** hydrocephalus and **(b)** meningomyelocele anomalies.

in the uterus. The newborn had an Apgar score of 3-5. A written informed consent was obtained from the patient.

DISCUSSION

It was first described by Jarcho and Levin in 1938.[4] Jarcho-Levin syndrome is a condition associated with abnormal development of the notochordal vertebral cartilage during Weeks 4-8 of intrauterine life, leading to rib deformities, and vertebral deformities due to inadequate segmentation of somites during Weeks 4-5. This syndrome can also be accompanied by various anomalies affecting the nervous, cardiac, urinary, and gastrointestinal systems.[5]

In genetic etiology, mutations in the delta-like 3 (DLL3) gene on chromosome 19 have been identified in some patients with SCD. The DLL3 encodes a ligand in the Notch signaling pathway, which is important for normal somitogenesis.[5] Consequently, a genetic etiology has not yet been identified for many patients, suggesting that the condition may involve significant heterogeneity.[6]

Prenatal USG in cases of Jarcho-Levin syndrome can reveal a range of anomalies. In our cases, although meningomyelocele and hydrocephalus were detected, the diagnosis of Jarcho-Levin syndrome was not made prenatally. Anomalies that may accompany Jarcho-Levin syndrome include hydrocephalus, meningomyelocele, spina bifida, atrial septal defect, ventricular septal defect, renal agenesis, renal hypoplasia, hydronephrosis, polycystic kidney, horseshoe kidney, bladder duplication, diaphragmatic hernias, anal atresia, imperforate anus, and Meckel's diverticulum.[7]

With advancements in medical technology and improvements in neonatal intensive care, the survival rates of patients with Jarcho-Levin syndrome who have severe anomalies have increased.[8]

In conclusion, patients with Jarcho-Levin syndrome presenting with meningomyelocele and hydrocephalus are rare. Both heterozygous and homozygous gene mutations have been identified in its development. When diagnosed intrauterine, it is important to inform the family about the progression of the condition, and genetic counseling should be recommended before planning future pregnancies. Early diagnosis during the prenatal and neonatal periods facilitates appropriate genetic counseling and treatment.

Data Sharing Statement: The data that support the findings of this study are available from the corresponding author upon reasonable request.

Author Contributions: Idea/concept, other: Z.P.C.,C.Y.; Design, literature review, writing the article, references and fundings: Z.P.C.; Control/supervision, critical review, materials, data collection and/or processing: Z.P.C., T.T. Analysis and/or interpretation: Z.P.C., C.Y., C.I.

Conflict of Interest: The authors declared no conflicts of interest with respect to the authorship and/or publication of this article.

Funding: The authors received no financial support for the research and/or authorship of this article.

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