

Jarcho-Levin Syndrome: A Case Report

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Abstract : Introduction: Spondylothoracic dysostosis, also known as Jarcho-Levin syndrome, is defined by a shortened neck and thorax, a protruding abdomen, inguinal and umbilical hernias, atypical spinal structure and rib fusion, leading to restricted chest movement or difficulty in breathing, along with urinary tract abnormalities and, potentially severe scoliosis. Aim: This is the case of a patient diagnosed with Jarcho-Levin syndrome, aiming to detail the range of abnormalities observed in this syndrome, the observed complications, and the therapeutic approaches employed. Results: A three-month-old male infant, born of a consanguineous marriage, delivered at full term by cesarean section, was admitted to the pediatric department for severe acute bronchiolitis. In his prenatal history, morphological ultrasound revealed macrosomia, a shortened spine, irregular vertebrae with thickened skin, normal fetal cardiac ultrasound, and the absence of the right kidney. His perinatal history included respiratory distress, requiring ventilatory support for five days. Upon physical examination, he had stunted growth, scoliosis, a short neck and trunk, longer upper limbs compared to lower limbs, varus equinus in the right foot, a neural tube defect, a low hairline, and low-set ears. Spondylothoracic dysostosis was suspected, leading to further investigations, including a normal transfontaneous ultrasound, a spinal cord ultrasound revealing a lipomyelocele-type closed dysraphism with a low-attached cord, an abdominal ultrasound indicating a single left kidney, and a cardiac ultrasound identifying Kommerell syndrome. Due to a lack of resources, genetic testing could not be performed, and the diagnosis was based on clinical criteria. Conclusion: Jarcho-Levin syndrome can result in a mortality rate of about 50%, primarily due to respiratory complications associated with thoracic insufficiency syndrome. Other complications, like heart and neural tube defects, can also lead to premature mortality. Therefore, early diagnosis and comprehensive treatment involving various specialists are essential.

Keywords : Jarcho-Levin syndrome, congenital disorder, scoliosis, spondylothoracic dysostosis, neural tube defect

Conference Title : ICP 2024 : International Conference on Pediatrics

Conference Location : Jeddah, Saudi Arabia

Conference Dates : February 19-20, 2024