

Case Report :

COVID19 in a case of Jarcho Levin syndrome.

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Abstract

Jarcho–Levin syndrome (JLS) also called spondylothoracic dysostosis is a rare congenital disorder. It is characterized by short stature with multiple abnormalities in the rib cage and vertebrae. The vertebrocoastal anomalies make the thoracic cage incapable of accommodating the growing lung. This in turn leads to thoracic insufficiency, severe cardio-respiratory complications, and frequent chest infections. We present the case of a 1-year-old baby girl who presented with symptoms of acute respiratory tract infection and was diagnosed with The novel Coronavirus Disease-2019 (COVID 19) by a positive rapid antigen test. During workup, she was also diagnosed to be a case of JLS with help of a skeletal survey. The baby died shortly after.

Key Words:

Jarcho–Levin syndrome, spondylothoracic dysostosis, COVID-19, ribs.

Introduction

Spondylothoracic dysostosis or Jarcho Levin syndrome is a rare congenital anomaly. The disease is hypothesized to be related to some genes although the exact gene that may be responsible is unclear. The disease is characterized by a short neck, short trunk, and multiple vertebral and costal deformities resulting in a deformed thoracic cage in turn causing res-

piratory insufficiency.¹ An autosomal recessive inheritance pattern is suggested.¹ Genetic diagnosis is rarely possible due to high costs and paucity of testing labs in the nation making radiology the main tool for diagnosis. Radiological features are distinctive including hemivertebrae, butterfly vertebrae, fused and hypoplastic vertebrae, crab-like fanned out ribs- the crab signs and tram track signs. Costo vertebral abnormalities make the patients

prone to recurrent chest infections resulting which at times are life-threatening². There is no definitive treatment for the condition. Treatment is mainly aimed at aggressive management of respiratory infections to prevent progression to respiratory compromise and subsequent respiratory failure. A titanium prosthesis device was recently approved for the treatment of this condition. COVID-19 is a new highly contagious viral disease. The spectrum of cases varies from asymptomatic to acute respiratory failure requiring mechanical ventilation, septic shock, and multiple organ failure. Our case highlights the role of radiology as a diagnostic tool for this rare condition and also emphasizes the fact that in patients presenting with a respiratory infection and respiratory distress, examination of the thorax becomes very important to rule out this condition as patients suffering from this condition are highly prone to respiratory infection and often deteriorate very quickly to respiratory failure and death

Case Report

A 1-year-old baby girl was brought to the emergency reception (ER) of North Bengal Medical College, Sushruta Nagar, West Bengal, India with complaints of fever with respiratory distress for the last 3 days. On arrival the baby was lethargic and Glas-

gow Coma Scale (GCS) score was calculated as 5/15. She was having an axillary body temperature of 101°F, respiratory rate of 65 per minute, heart rate of 168 per minute, blood pressure of 70/40 mm Hg, and SpO₂ of 90% with room air. Chest indrawing was present along with a wheeze in both lung fields. The baby had a chest wall deformity which was noticed by the attending physician. A chest radiograph was requested. The chest cavity was small and asymmetric. Scoliosis was noted with convexity to right. Ribs were malformed and malaligned fanning out in a “crab-like” manner. A Hemivertebrae was noted at the 5th thoracic vertebrae and there was an absence of the corresponding rib on the right side at that level (Figure 1).



Figure 1 – Multiple radiating crab-like ribs, Hemivertebrae at D5 level with absent corresponding rib on right

A suspicion of Jarcho-Levin syndrome(-JLS) was made. Due to the ongoing pandemic of COVID 19, a Rapid Antigen Test (RAT) for COVID-19 was done at the ER which came positive. The baby was immediately shifted to the dedicated pediatric Intensive care unit(PICU) for management of COVID 19 positive babies. There the baby was put on an oxygen hood with 2 Litre oxygen per minute. A skeletal survey was done with a portable X-ray which came out normal except for the thoracic cage and vertebral anomaly as described previously. Ultrasonography(USG) of the cranium and abdomen was unremarkable. Echocardiography was normal. On taking a detailed history from the father it was found that the family was from a remote tribal area of North Bengal. Due to financial issues, the mother never had any antenatal ultrasound scans. The baby was born at home by normal vaginal delivery with the assistance of a local tribal midwife. The baby was apparent well after birth and the neonatal course was uneventful except for a subtle deformity in the chest which was progressively increasing as the child was growing. There was no delay in developmental milestones. There was no history of similar illnesses in the family. A provisional diagnosis of COVID 19 in a patient of JLS was made. The baby deteriorated on

the following day and was put on mechanical ventilation. Shortly after that, the baby died.

Discussion

Jarcho Levin syndrome is a very rare disease in the world of medical literature. In world medical literature there are around 400 reported cases of JLS and around 5 of them are from India.³ To the best of our knowledge, this is probably the first case reporting COVID-19 infection in a patient of JLS

The hallmark feature is abnormal vertebral segmentation and rib abnormalities resulting in a deformed thoracic cavity with ribs fanning out in a “crab-like” fashion. Other associated anomalies seen to occur with this condition include a short neck and chest, a protuberant abdomen multiple site hernias, facial abnormalities, and congenital anomalies of the genitourinary tract.^{1,2} JLS includes two distinct genetic disorders, spondylocostal dysostosis, and spondylothoracic dysplasia.³

Imaging features of this condition are very characteristics. The primary modality of choice is Plain radiographs or CT scans with 3D reconstruction. The pathognomonic features include H-shaped vertebrae, segmentation anomalies, scoliosis, and crab-like fanning out of ribs from the costovertebral base. Another finding is

referred to as the tramline sign resulting from prominent vertebral pedicles and irregularly shaped vertebral bodies.^{1,2}

These alterations cause severe thoracic restriction in most newborns, resulting in respiratory distress syndrome. Due to the extensive rib fusion, the intercostal muscles are unable to expand the thorax. A pulmonary hypoplasia-like syndrome occurs due to the reduced size of the thorax; however, no intrinsic pulmonary alteration has been described yet in literature. Due to respiratory complications, newborns have a mortality rate of up to 40%.^{1,2,3,4}

Although studies have shown an association of a few genes with JLS like DLL3, MESP2, LFNG, HES7, and TBX6, many patients don't have any abnormalities in these genes. Hence is the hypothesis that there is a yet unknown gene responsible for this disease that needs further investigation.⁵

Genetic testing can detect mutations of JLS, but suffer from the lack of widespread availability and high cost of study. Radiology and clinical findings together serve as a cheaper alternative to diagnosis in a resource-poor setting like India.

In the absence of genome testing for JLS, reliance for diagnosis is placed on history, physical appearance, clinical examination, and imaging findings.

Primary treatment is primarily targeted at

the management of respiratory infection to avoid respiratory compromise. Definitive treatment for this condition is still under research. A device called vertical expandable prosthetic titanium rib (VEPTR) was approved in 2004 by USFDA. It straightens the spine and separates the ribs consequently providing room for the lungs to expand as the child grows.^{5,6,7}

COVID-19 is a highly contagious viral illness caused by severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2). The first cases of this illness were first reported in Wuhan, Hubei Province, China, in December 2019. Since then the disease has rapidly spread across the world in a short period. The World Health Organization (WHO) declared it a global pandemic on March 11, 2020. The disease is caused by Coronaviruses which are positive-stranded RNA viruses with a crown-like appearance under an electron microscope. The clinical spectrum of COVID-19 is varied. It varies from asymptomatic forms to clinical illness characterized by acute respiratory failure requiring mechanical ventilation, septic shock, and multiple organ failure. The vast majority of symptomatic patients commonly present with fever, cough, and shortness of breath. Diagnosis is by real-time polymerase chain reaction (RT-PCR) or rapid antigen testing. There is no definitive treatment for this condition. Sev-

eral drugs are currently being investigated for this purpose. Vaccines are available for both adults and children.⁸

Conclusion

This case report highlights a rare case of JLS presenting with COVID 19 pneumonia. In patients presenting with respiratory distress syndrome thoracic, vertebral assessment becomes important to exclude diseases like JLS. These patients have a deformed thoracic cage that in turn, predisposes them to the risk of recurrent chest infection and respiratory insufficiency. Death is mainly due to respiratory failure.

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