Case Report:

Jarcho-Levin syndrome (spondylocostal dysostosis) associated with Sprengel deformity

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ABSTRACT

Jarcho-Levin syndrome (JLS) is a rare heterogeneous disorder characterised by short neck, short trunk and multiple vertebral and rib anomalies. Sprengel shoulder deformity is elevation of shoulder joint due to upward displacement of scapula. We are reporting a case of Jarcho-Levin syndrome associated with Sprengel deformity.

Key words: *Jarcho-Levin syndrome, Sprengel deformity*

Bindu CM, Krishnappa J, Hegde P, Bharath M. Jarcho-Levin syndrome (spondylocostal dysostosis) associated with Sprengel deformity. J Clin Sci Res 2017;6:46-9. DOI: http://dx.doi.org/10.15380/2277-5706.JCSR.16.03.005.

INTRODUCTION

Jarcho-Levin syndrome is a rare genetic malformation of vertebra and ribs. In this syndrome there are multiple segmentation defects of vertebra and multiple rib malformations. Sprengel shoulder deformity is elevation of shoulder joint due to upward displacement of scapula. These children have short neck, short trunk, scoliosis and crab like ribs, including crowding of ribs. This short neck and short trunk deformity is diagnosed by clinicoradiological characteristics. This syndrome was first described in 1938 by Jarcho and Levin.[1]

This syndrome is classified into subtypes i.e., spondylothoracic dysostosis and spondylocostal dysostosis. Infants of Jarcho-Levin syndrome often suffer from repeated respiratory infections.[2,3]

CASE REPORT

A 4-year-old female child presented with fever, cough, breathlessness and chest indrawing of eight days duration. There was history of repeated respiratory tract infections present since birth.This child was the first child of a non consanguinously married couple.

On examination, child was dyspneic,febrile with elevated pulse 100/mm, tachypnoea respiratory rate 60/min, and chest indrawing. Mild pallor was present. There was no cyanosis. Anthropometric measurements revealed protein energy malnutrition (PEM) Grade ll as per Indian Academy of Paediatrics (IAP) classification.[4]

The child had low set ears and wide nasal bridge. Musculoskeletal examination revealed upward displacement of right scapula, scoliosis, and crowding of ribs (Figure 1). Respiratory system examination revealed decreased breath sounds and coarse crepitations over right supraclavicular, right infraclavicular and interscapular regions.

Laboratory investigations revealed haematological parameters to be within normal limits.

Chest radiograph showed upward displacement of scapula, crowding of ribs and malformed ribs

Received: March 22, 2016; Revised manuscript received: August 26, 2016; Accepted: September 03, 2016.

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Online access
http://svimstpt.ap.nic.in/jcsr/jan-mar17_files/4cr.16.03.005.pdf
DOI: http://dx.doi.org/10.15380/2277-5706.JCSR.16.03.005

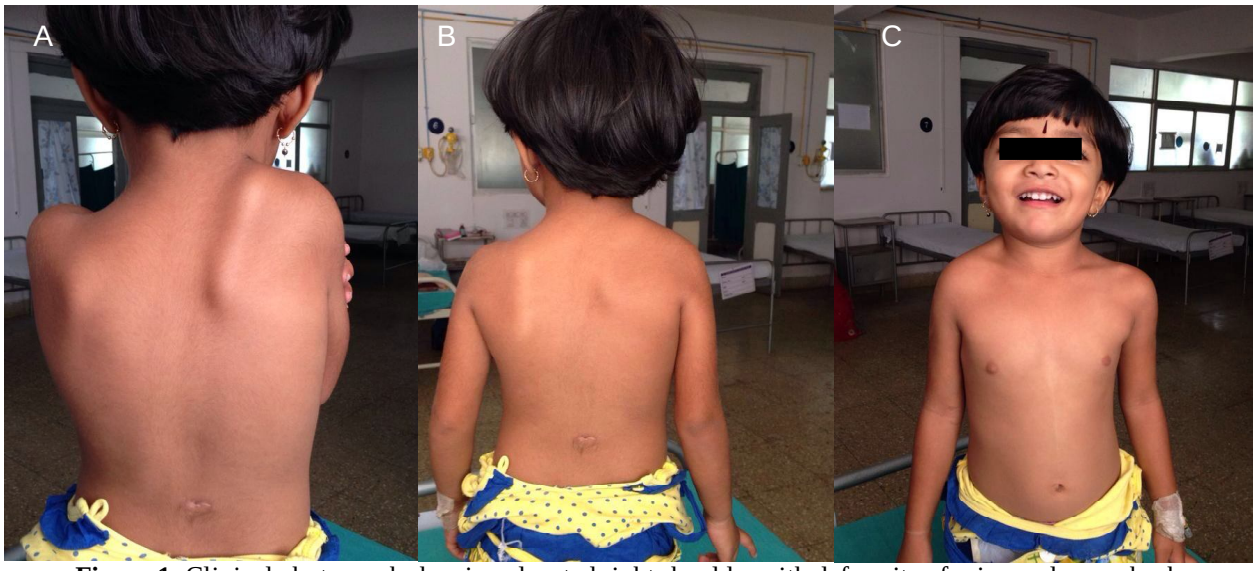

Figure 1: Clinical photograph showing elevated right shoulder with deformity of spine and upper back

on right side with "crab like appearance" (Figure 2).

Computed tomography (CT) of thorax showed unsegmented vertebra (block vertebra) involving T1 to T5 vertebra, hemivertebrae involving T1 to T3, crowding and deformity of second, third and fourth ribs on right side with high position of right scapula. (Figures 3,4,5 and 6).

Ultrasonography of abdomen and echo-cardiogram were normal. Karyotyping study was normal. This child was treated for respiratory infection and surgical correction at a later date was advised.

DISCUSSION

Jarcho-Levin syndrome is a rare genetic disorder with autosomal dominant (AD) or autosomal recessive (AR) inheritance. The gene involved in this syndrome is delta-like protein 3 (DDL3) on chromosome 19 at 19q13,[5] and mesoderm posterior protein 2 (MESP2) gene located on chromosome 15 at 15q26.1.[6] The small size of the thorax in newborns frequently leads to respiratory compromise and death in infancy. Some rare variants with survival into adulthood have also been described.

This syndrome is found more frequently in people in Spanish origin.[7] Prenatal diagnosis by ultrasonography is helpful in early detection. Type 1 (spondylothoracic dysostosis) is the severe form characterised by severe spine involvement and respiratory failure and it is AR. Type ll (spodylocostal dysostosis) is a mild form, has AD inheritance and may not be diagnosed in utero. They have near normal longitivity. Males and females are equally affected in both types.[7]

Babies with this syndrome are usually born of a consanguinous marriage, but this was not so in our case. Affected children usually present with chest deformity or severe lung infections. This patient presented with repeated respiratory

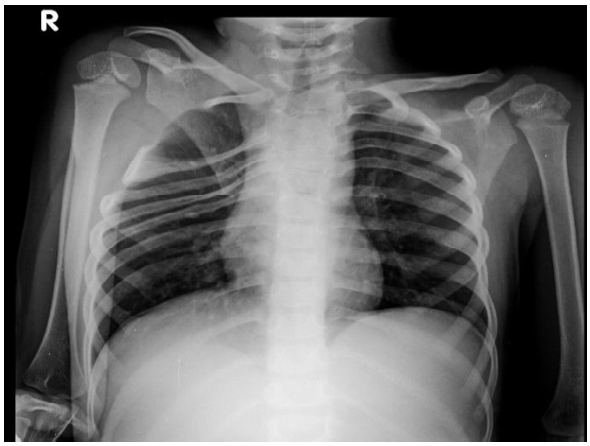

Figure 2: Chest radiograph (antero-posterior view) shows segmentation anomaly involving upper thoracic vertebrae. Right 1st rib appears hypoplastic with crab like deformity of the 2nd, 3rd, 4th and 5th ribs on right side.The right scapula is at an elevated level compared to the left

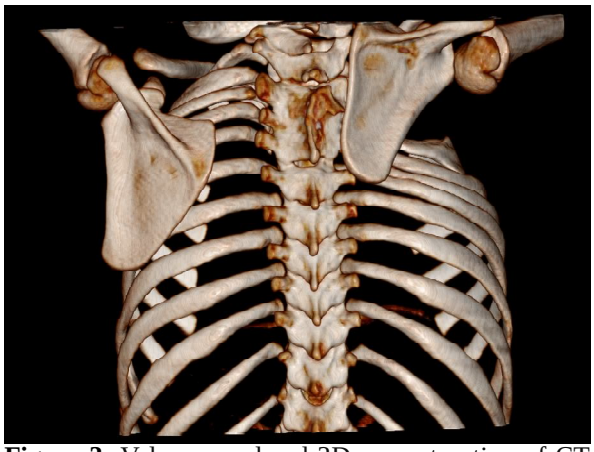

Figure 3: Volume rendered 3D reconstruction of CT images showing fusion of lamina of T1-T5 vertebrae and elevation of right scapula
CT = computed tomography

infections and Sprengel deformity of right side. External features of Jarcho-Levin syndrome are dysmorphic facies such as low set ears,wide nasal bridge and long philtrum. Our patient had two of these three features.

Jarcho-Levin syndrome is primarily characterized by short neck, short trunk and a narrow thorax due to multiple rib (crab-like or fan-like radiation of the ribs) and vertebral anomalies including butterfly vertebrae, hemivertebrae, fused vertebrae, hypoplastic vertebrae.

Jarcho-Levin syndrome is very rare. Our patient had both Jarcho-Levin syndrome and Sprengel

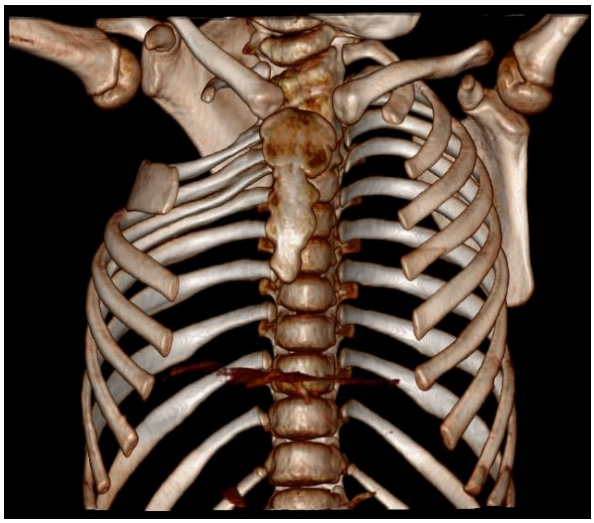

Figure 4: Volume rendered 3D reconstruction of CT images showing hypoplastic right 1st rib with crab like deformity of the 2nd, 3rd, 4th and 5th ribs on right side. The right scapula is elevated
CT = computed tomography

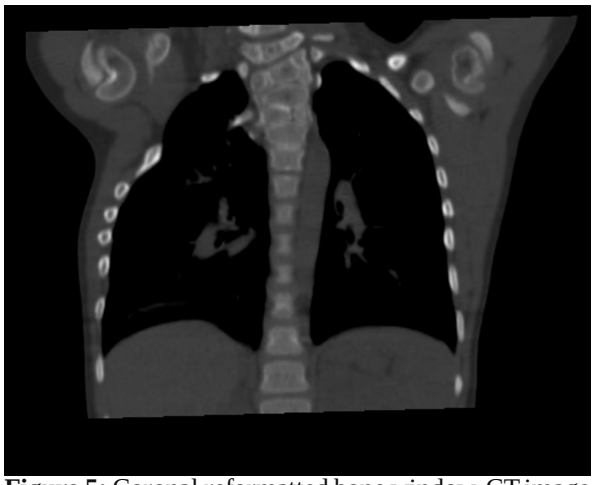

Figure 5: Coronal reformatted bone window CT image showing fusion of T1-T5 vertebral bodies and segmentation anomaly
CT = computed tomography

deformity.Very few such cases are reported in the literature.These cases can be diagnosed prenatally by three-dimensional ultrasonography and measurement of nuchal thickness.[8]

Radiological features such as multiple vertebral deformities, abnormal ribs and elevated scapula are recognised by radiographs and CT. Most cases reported in the literature have Springel deformity on the left side. Our patient had upward displacement of right scapula, which is rare.

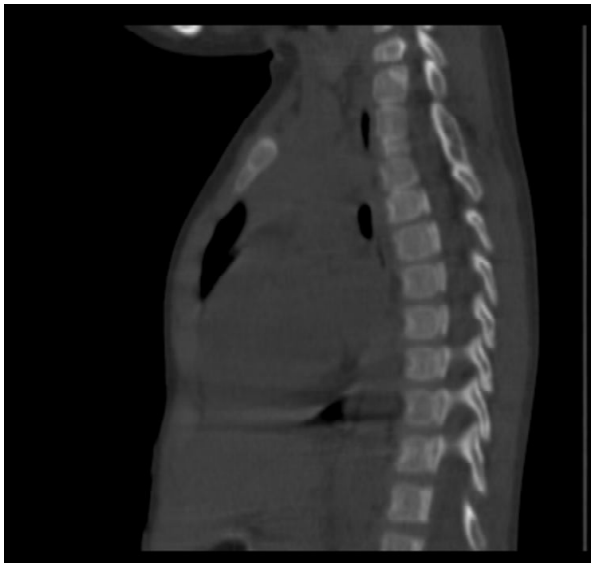

Figure 6: Sagittal reformatted bone window CT image showing segmentation anomaly in upper thoracic vertebrae
CT = computed tomography

Jarcho-Levin syndrome may be associated with hydrocephalus, renal anomalies, facial dysmorphism, complex congenital heart disease,[3] limb and digit anomalies,[9] hernias,neural tube defects and anomalies of anal opening, urinary tract and uterus.[10] Of these additional features, our patient had only dysmorphic facial features.

This case of Jarcho-Levin with Sprengel deformity is being reported because of the rarity of this combination. Paediatricians should be aware of this disorder. The present case is atypical in that the Sprengel deformity is located on the right side.

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