

## SPOT DIAGNOSIS (IMAGE GALLERY)

**CRAB LIKE RIBS**

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A 2 years old boy born to a non consanguineous couple presented with febrile status epilepticus. The child was treated according to our PICU protocol. On stabilizing, the child was evaluated for the gross morphological abnormalities. He was short with height less than 3rd centile. Upper: lower segment ratio was 0.93. He

had a short webbed neck, low posterior hairline, asymmetrical thorax and scoliosis. He had no other skeletal abnormality or neurological deficit. Chromosome analysis revealed a normal male karyotype. Radiographs showed multiple abnormalities of vertebral bodies with hemivertebrae, fusion and bifurcation of ribs with sun ray like appearance. (Fig. 1) MRI of spine confirmed the above findings. MRI of brain was normal. Metabolic work up, lumbar puncture, blood counts were within normal limits.

**What is the genetic disorder?**

Spondylocostal dysostosis (Jarcho-Levine syndrome). It comprises of multiple malformations of the vertebrae and ribs coupled with a characteristic clinical picture of short neck, scoliosis, short trunk and deformities of the rib cage. (1,2) In the autosomal dominant form, affected children are short but otherwise the disorder runs a benign course. In contrast the autosomal recessive form succumbs to pneumonia in infancy or early childhood. Despite the abnormalities of the spine, the integrity of the spinal cord is not altered and the neurological function remains normal. The main clinical features are plagiocephaly (3) facial asymmetry, a short rigid neck and scoliosis. Radiographically, hemi- and fused vertebrae give rise to a short trunk, in severe cases. The spinal defects can be associated with absent or fused ribs. Turnpenny et al. (4) reported the clinical, radiographic and molecular findings in 10 families with autosomal recessive SCD, abnormal vertebral segmentation and the notch signalling pathway. Abnormal segmentation throughout the entire spine has been described as a predominating and a sole pathology. In recent years, genetic mapping and candidate gene sequencing approaches have identified causative mutations in four genes, all components of the Notch signalling pathway. SCDO1 (OMIM 277300) represents the majority of cases, as the present one and in which the underlying genetic cause has been identified and is due to mutation of the DLL3 gene with 27 distinct causative mutations identified to date. The closest differential diagnoses include neural tube defects which need to be evaluated. Eventually the prognosis has been considered directly related to respiratory complications. (5) The treatment includes surgical correction of the deformities by a team approach and regular follow-ups.

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