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IMAGES IN PAEDIATRICS

Short stature and scoliosis: revealing signs of ultrarare skeletal dysplasia

Talla baja y escoliosis: datos reveladores de displasia esquelética ultrarara

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A girl age 7 years was referred to our unit for evaluation of scoliosis without a relevant family history. She had a history of hemivertebra, detected prenatally.

The findings of the physical examination were: height, 115.3 cm (2nd percentile; z-score, −2.14); weight, 20.4 kg (10th percentile; z-score, −1.29). The actual height differed from the expected height. The upper/lower segment ratio was 0.9; the arm span, 114 cm; the sitting/standing height ratio was 0.497 (21st percentile; z-score, −0.81). No dysmorphia. The patient had a short and wide thorax, pectus excavatum and prominent ribs. Tanner 1 stage of pubertal development. The blood panel and karyotyping results were normal.

The plain radiograph (Fig. 1) and magnetic resonance imaging (Fig. 2) of the spine evinced abnormalities, and the next generation sequencing skeletal dysplasia panel (463 genes) identified a compound heterozygous variant, NM_016941.3:c.[988 G >T];[1312 T >C], p.[(Gly330Cys)];[(Cys438Arg)], in the *DLL3* gene. Each parent carried one of the alternate alleles. Both were classified as variants of uncertain significance by the American College of Medical Geneticists.

Jarcho-Levin syndrome, also known as spondylocostal dysostosis (OMIM #277300) is an autosomal recessive genetic disease characterized by segmentation defects of the vertebrae.[1,2] It should be suspected in individuals presenting with short stature, short neck and trunk asymmetries and is confirmed in imaging tests by the presence of anomalies such as hemivertebrae, block fusion or rib deformities. The results of genetic testing suggested that the identified changes in *DLL3* could be the cause of dysplasia, but did not allow confirmation of the diagnosis, as they were variants of uncertain significance. The management consists of supportive care, and infection is the most frequent complication.[3]

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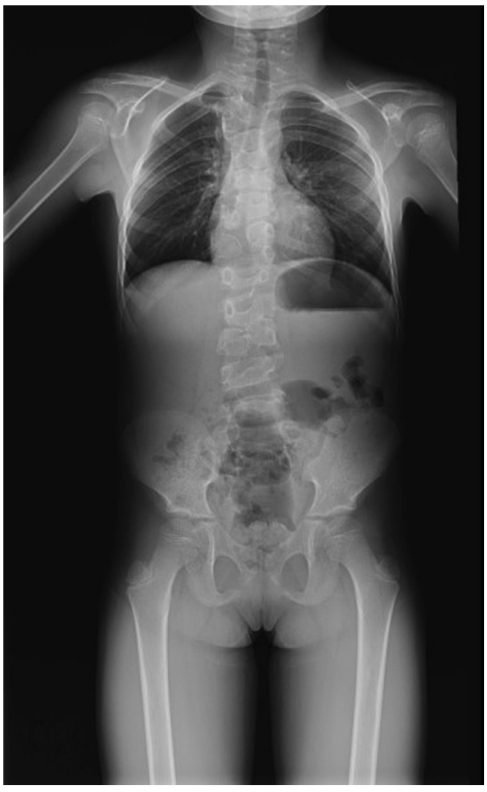

Figure 1 Complete X-ray of the spine (7 years and 7 months): multiple vertebral and rib malformations. Scoliosis with high dorsal curve and mild convexity to the right.

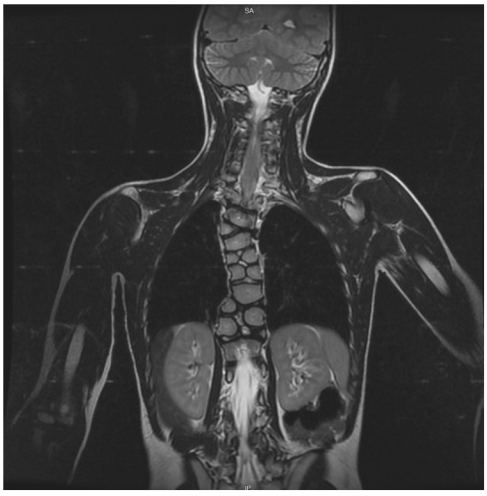

Figure 2 MRI of the spine, coronal and sagittal views (7 year and 10 months). Cervical segment: loss of physiological cervical lordosis. Dorsal segment: 9 rib pairs. Right-sided hemivertebrae at D3 and D5-D6. Two hemivertebrae at D8-D9. Rudimentary hemivertebra at D10. Absence of D11 and D12.

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Conflicts of interest

The authors have no conflicts of interest to declare.

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