Spondylocostal dysostosis

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SUMMARY A female child with multiple vertebral and rib abnormalities is described.

Skeletal abnormalities localised to the spine and ribs have been recorded under various headings which include 'spondylocostal dysplasia',¹ 'spondylothoracic dysplasia',² 'spondylocostal dysostosis',³ and 'costovertebral dysplasia'.⁴ Individually these conditions are rare and await full delineation. In this report a relatively mild form is described in the 5-year-old daughter of consanguineous parents. No previous report of this condition in the United Kingdom has been traced.

Case report

The patient is the first child of healthy Caucasian parents who are first cousins. She has one normal younger brother and there is no family history of

spinal abnormality. She was born by forceps delivery following an uneventful drug-free pregnancy with birth weight 3.2 kg. Her development has been normal. She has had no serious illnesses, nor has she had symptoms associated with her spinal abnormality.

On examination at 5½ years she presented as a very attractive little girl with weight and head circumferences on the 10th centile, while her height of 97.7 cm fell 3 cm below the 3rd centile. Her upper to lower segment ratio was 0.92 and her span was 107 cm. She had a short webbed neck with a low hair line, an asymmetrical thorax, and a mild scoliosis (fig 1). She showed no other evidence of skeletal abnormality and had no neurological deficit.

Chromosome analysis revealed a normal female karyotype. Radiographs showed multiple abnormalities of the vertebral bodies with hemivertebrae, fused

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FIG 1 The patient aged 5 years. Note her short webbed neck and low posterior hair line.

FIG 2 Oblique view of the chest at the age of 3½ years showing widespread errors of segmentation and fusion in the thoracic vertebrae with associated rib anomalies.

FIG 3 Anteroposterior view of the spine at the age of 3½ years.

vertebrae, and fusion and bifurcation of the ribs (fig 2). These errors of segmentation and fusion extended throughout the vertebral column (fig 3). There were 11 ribs on the right and nine on the left.

Spinal x-rays of the parents were normal.

Discussion

On clinical and genetic grounds it is possible to distinguish at least three different forms of spondylocostal dysostosis. In the autosomal dominant form, multiple errors of segmentation in the vertebrae are associated with aplasia, hypoplasia, and fusion of the ribs.^{1,5} Affected subjects are short but otherwise the disorder appears to run a benign course. In contrast, patients with the severe autosomal recessive form of this condition tend to succumb in infancy or early childhood to pneumonia.^{6,7} These children have a very short neck and trunk, a prominent thorax, and a reduced number of ribs which fuse posteriorly to give a characteristic crab-like appearance on chest x-ray. Many of the reported cases are in Puerto Rican families.

In the third form, inheritance is autosomal recessive

and clinical problems are few.^{2,8-10} The radiological findings are almost identical to those seen in the autosomal dominant type. The clinical, genetic, and x-ray features in the child described here suggest that she is likely to have this relatively mild third form of spondylocostal dysostosis. Despite the bizarre appearance of the spine in these subjects, the integrity of the spinal cord does not seem to be compromised. However, it would seem prudent to monitor neurological function carefully in these patients.

The radiological abnormalities in this group of disorders differ from those in the much more common neural tube defects, in that they extend throughout the spinal column. This case is presented not only to add to the very sparse literature on this subject, but also to stress the importance of distinguishing between spondylocostal dysostosis and a neural tube defect if correct genetic advice is to be given.

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