SPONDYLOTHORACIC AND SPONDYLOCOSTAL DYSOSTOSIS

HEREDITARY FORMS OF SPINAL DEFORMITY

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Two siblings with spondylothoracic dysostosis, and two siblings and three unrelated children with spondylocostal dysostosis are described. Both conditions are inherited and characterised by malformed thoracic and lumbar vertebrae. Spondylothoracic dysostosis produces "crab-like" deformities of the ribs, and is usually fatal during early infancy due to respiratory failure. Spondylocostal dysostosis causes short-trunked dwarfism but does not usually reduce life expectancy.

These clinical features are distinct from congenital scoliosis, although all three conditions are associated with a particular group of malformations.

Many factors contribute to the aetiology of congenital scoliosis, and although several vertebrae may be affected, the greater part of the spine is normal. Spondylothoracic dysostosis (Solomon, Jimenez and Reiner 1978) and spondylocostal dysostosis (Young and Moore 1984) are hereditary conditions in which most if not all the vertebrae are malformed. Only one case of spondylothoracic dysostosis (Heilbronner and Renshaw 1984) and two cases of spondylocostal dysostosis (Fogarty, Beatty and Dowling 1985) have, as far as we know, been described in the orthopaedic literature.

We report seven more cases, two siblings with spondylothoracic dysostosis, and two siblings and three unrelated children with spondylocostal dysostosis. All had Caucasian parents. The clinical and radiological features of these patients are discussed and compared to those of congenital scoliosis.

CASE REPORTS

Cases 1 and 2. An unrelated couple had had one healthy boy but their second child died soon after birth from respiratory failure. Postmortem radiographs showed extensive rib and vertebral malformations (Fig. 1), and at autopsy there was no diaphragm on the left side and the left kidney was absent. The mother's third pregnancy ended at 18

weeks in the spontaneous abortion of a normal fetus, and her fourth (Case 2) was terminated at 20 weeks because ultrasound of the fetus showed a severely deformed spine and rib cage; postmortem radiographs were similar to those seen in Case 1 and an autopsy found abnormal adrenal glands as well as anal atresia (this sibship has been reported in greater detail elsewhere: Tolmie et al. 1987).

Cases 3 and 4. Two brothers, now in their early twenties, had been observed since childhood; their birth-weights had been 2.1 kg and 2.7 kg. They have normal, unrelated parents and three normal siblings.

Fig. 1

Case 1. Radiograph showing the typical "crab-like" ribs in spondylothoracic dysostosis.

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They were of short stature (Fig. 2) with reduced upper-to-lower segment ratios. Radiographs showed malformations of all the thoracic and lumbar vertebrae and similar rib anomalies (Figs 3 and 4), and both had interbody fusions between two lower cervical vertebrae. Apart from these rib and vertebral malformations (Fig. 5), both boys were normal.

Case 5. An eight-year-old boy was seen when two years old with a thoracolumbar scoliosis of 41° . He had no siblings, his parents were first cousins, and his birth-weight was 2.8 kg. His height has remained consistently just below the third centile. Radiographs showed multiple hemivertebrae: he had 13 thoracic segments of which only the seventh, eighth and ninth were normal, and six lumbar segments, all of which

were abnormal. There were 13 normal ribs on each side. He had no other malformations.

Case 6. A two-year-old girl, the only child of unrelated parents, presented because of short stature. Her birth-weight was 2.1 kg and her height has remained below the third centile. A radiograph (Fig. 6) showed extensive vertebral and rib anomalies but she had no other malformations.

Case 7. A two-year-old girl with two normal siblings, was born to unrelated parents; her birth-weight was 2.8 kg and she had cyanotic heart disease which required surgery shortly after birth. Her height is still below the third centile and radiography showed multiple hemivertebrae and rib fusions.

Fig. 2

Figure 2 - Growth-charts of two brothers, Cases 3 and 4. The mature heights are well below the third centile. Figures 3 and 4 - Radiographs of both boys at the age of 10 years. The malformations are similar with multiple hemivertebrae, butterfly vertebrae and posterior fusion of the right tenth and eleventh ribs. Figure 5 - Clinical photograph of the two boys.

Fig. 3

Fig. 4

Fig. 5

Fig. 6

Case 6. Radiographs showing malformations of all the thoracic and lumbar vertebrae; the patient also had multiple rib deformities.

DISCUSSION

There are at least two types of congenital spinal deformity characterised by multiple vertebral and rib malformations. The first, spondylothoracic dysostosis (Cases 1 and 2), is an autosomal-recessive deformity and causes death in early infancy from respiratory failure. It is also known as the Jarcho-Levin syndrome (Jarcho and Levin 1938) or as spondylothoracic dysplasia (Pochaczewsky et al. 1971). The second variety, spondylocostal dysostosis, may be inherited as a recessive or a dominant trait, and patients usually have a normal life expectancy; Cases 3 and 4 are examples of recessive inheritance and, in view of the parental consanguinity, so probably is Case 5. Cases 6 and 7 could either be recessive or dominant. Other names for spondylocostal dysostosis are Jarcho-Levin Type II (Heilbronner and Renshaw 1984) or spondylocostal dysplasia (Rimoin, Fletcher and McKusick 1968).

The radiological features are distinctive: in both types of deformity there are multiple alternating hemivertebrae which, unlike congenital scoliosis, affect all or almost all segments of the thoracic and lumbar spine. The ossification centres seldom cross the midline

(Solomon et al. 1978). The cervical spine may be normal, may have hemivertebrae or may have interbody fusions. In Case 6 and in the twins reported by Fogarty et al. (1985) the number of vertebral segments was reduced but this is unusual; indeed, in one of our patients (Case 5) there were extra segments. In spondylothoracic dysostosis the rib deformities are severe and multiple posterior fusions produce a small thorax with a "crab-like" radiographic appearance. By contrast, in spondylocostal dysostosis the ribs are only slightly affected. Although Casamassima et al. (1981) described a child who required spinal fusion, scoliosis is seldom severe; the worst seen in our series (Case 5) now measures 45°.

Children with spondylocostal dysostosis have low birth-weights and are of short stature, with reduced upper-to-lower-segment ratios. Standing height remains just below the third centile until the onset of puberty but, because vertebral growth is diminished, there is virtually no adolescent growth-spurt; thus height at maturity is much reduced, usually below 155 cm (Fig. 2). Although congenital scoliotics may be small, such severe disproportionate dwarfism is most unusual.

One-third of children with spondylothoracic dysostosis and a quarter of those with spondylocostal dysostosis have other malformations (Casamassima et al. 1981): in both groups the most common are congenital heart disease and renal anomalies but urogenital abnormalities, polydactyly, tracheo-oesophageal fistulae and anal atresia have also been reported. In many of the infants who died, there was no autopsy so the real incidence may be greater. These malformations occur in the same non-random fashion as in congenital scoliosis and follow the so-called VACTERL pattern, that is vertebral, ano-rectal, cardiac, tracheal, esophageal, radial, renal, and lower limb deformities (Khoury et al. 1983). This is not surprising because they all arise from abnormal development of paraxial mesoderm.

In congenital scoliosis the risk to siblings for closed malformations is 3% (Wynne-Davies 1975), but this figure does not apply to hereditary conditions. For normal parents who have a child with the dominant form of spondylocostal dysostosis, the risk to subsequent children is negligible, but the affected infant's offspring carry a risk of 1 in 2. In recessive inheritance the risk to siblings of an affected child is 1 in 4 but the risk of their own children being affected is negligible.

When attempting to determine whether a child may have a hereditary spinal deformity, considerations should include family history, consanguinity, malformations of all or almost all the vertebrae, multiple posterior rib fusions and severe shortening of the trunk. Parents who have a child with spondylothoracic or spondylocostal dysostosis should be given genetic counselling; subsequent pregnancies should be monitored by ultrasound to detect the lethal spondylothoracic form in time to consider termination (Tolmie et al. 1987). The relationship of these conditions to neural-tube defects is

not clear, but because there has been one case report of a child with spondylothoracic dysostosis and spina bifida (Eller and Morton 1970), this also should be borne in mind during ultrasound examination.

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