Hereditary costovertebral dysplasia with malignant cerebral tumour

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SUMMARY  Costovertebral dysplasia comprises multiple malformations of the vertebrae and ribs, with a characteristic clinical picture of short trunk dwarfism, short neck, scoliosis, and rib cage deformity. We describe two sibs with the syndrome who are presumed to represent the autosomal recessive form of the disorder. One sib died from a malignant cerebral tumour and the association may be more than fortuitous.

Costovertebral dysplasia consists of multiple anomalies of the vertebrae and ribs associated with shortening of the trunk.1-11 The condition is also known as spondylothoracic dysplasia2 4 and spondylcostal dysplasia.1 10 11 The neck is short and rotatory movements are limited. Gross disorganisation of vertebral segmentation is manifested radiographically by a reduced number of vertebrae, fused vertebrae, hemivertebrae, and sagittally cleft vertebrae. The odontoid peg may be hypoplastic,4 an important fact should the need for an anaesthetic arise. There is a reduction in the number of ribs and vertebral pedicles associated with multiple rib fusions. The chest wall is grossly deformed. All other parts of the osseous skeleton are normal in this condition.

The condition is commonly familial. Reports of recessive inheritance1 3 4 7-9 12 clearly outnumber reports of dominant inheritance,6 10 but sporadic cases are well recognised and may be associated with congenital heart disease.5 Predictably, in severe cases there is an association with serious respiratory infections and death in infancy.2 3 7 13

We report a brother and sister with this syndrome. The sister died from a malignant cerebral tumour.

Case reports

Case 1
Case 1 was born on 30.11.75. It was the first pregnancy of healthy parents when the mother was 19 and the father 20 years old. A urinary infection occurred in the second month and was treated with an unspecified antibiotic but the pregnancy was otherwise normal. Normal delivery occurred at 40 weeks' gestation, birth weight 2875 g. Scoliosis, a short neck, a malformed rib cage, and a right inguinal hernia were noted at birth. The right inguinal hernia was repaired at 6 weeks of age. One year later he was thought to have a recurrence of the hernia. At surgical exploration there was no recurrence, but weakness of the inguinal musculature was noted on both sides. At the age of 7 years his height was 103.5 cm (<3rd centile) and weight was 16.5 kg (<3rd centile). He was in good general health apart from eczema and dry ichyotic skin.

Radiographs showed a reduction in the total number of segmental levels of the spine with multiple hemivertebrae in the cervical and lumbar spine, fusion of the high thoracic vertebrae, failure of normal development of thoracic discs, and partial sacral agenesis (figs 1 and 2). The ribs had multiple fused posterior ends with fusion of the shafts of the first and second ribs bilaterally. There was a thoraco-lumbar scoliosis concave to the left. Anterior defects were present in the lumbar spine. The odontoid peg, seen on an early film, looked normal.

Case 2
Case 2 was born on 8.11.80. It was the mother's second pregnancy, which was normal throughout with the exception of vaginal bleeding for 7 days occurring within a few days of the first missed period. No drugs were given. Normal delivery occurred at 40 weeks' gestation, birth weight 3175 g. Scoliosis, a short neck, and a malformed rib cage were noted at birth, the appearances being identical to those of her brother (case 1).
Radiographs showed very similar changes to case 1 in the spine, with a reduction in the total number of segments, multiple hemivertebrae in the lumbar spine, and sacral deficiency and failure of segmentation of the thoracic vertebrae. Anterior defects in the lumbar spine and a thoracolumbar scoliosis were also present. The ribs also had multiple fused posterior ends (fig 3). The odontoid peg was normal.

She was admitted to hospital on 30.8.81 aged 9 months with a 6-week history of vomiting each morning, and her parents noted that she had been pulling at her hair for one week. She was unconscious, a CT scan showed an extensive posterior fossa tumour, and she died 2 days later. At necropsy the tumour histology was that of a malignant epithelial ependymoma. Apart from the skeletal malformations, no other abnormalities were apparent.
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Parents

Radiographs of the whole spine and chest of the mother (born 9.1.57) and father (born 5.2.53) were normal apart from failure of fusion of the neural arch of the father’s first sacral vertebra. There was no consanguinity, no other pregnancies, and no family history of this or any other malformation.

Discussion

Vertebral malformations are aetologically heterogeneous, and costovertebral dysplasia is merely one of the numerous aetiological categories. The clinical and radiological features of the condition are distinctive, though the syndrome is itself heterogeneous, being attributable in some families to recessive genes and in others to dominant genes. In this family we suspect a recessive mode of inheritance.

Some reports suggest a possible aetiological relationship between multiple vertebral abnormalities and spina bifida, but costovertebral dysplasia appears to be a discrete entity unrelated to neural tube defects. The present case is of particular interest because of the increasingly apparent association between congenital malformations and the development of neoplasms. The association more commonly involves hamartoses, and with the exception of the possible link between vertebral malformations and sacrococcygeal teratoma, neither vertebral malformations nor bone dysplasias appear to have any special association with malignancy. Ependymomas are a relatively uncommon cerebral tumour, comprising 12% of intracranial and spinal tumours in the Manchester Children’s Tumour Registry from 1954 to 1968. So few cases of costovertebral dysplasia have been reported that it is difficult to assess the significance of the tumour association in this report, but considering the rarity of both disorders the combination may be more than a coincidence.

It is said that poor muscle tone may be a feature of the condition, leading to protuberance of the abdomen and abdominal wall hernias. Poor muscle tone was certainly not present in our two cases, and indeed the abdomen was strikingly flat rather than protuberant. Nevertheless, it is noteworthy that case 1 had an inguinal hernia and at operation weak inguinal musculature was noted. It may be that there is a spectrum of severity in costovertebral dysplasia, with the more severe cases dying from respiratory failure or infection in infancy. One suggestion is that the respiratory disorder in the severe cases may be related primarily to abnormal lung development, which could be secondary to reduced fetal breathing movements or an abnormal rib cage. If muscular hypotonia is a feature of the disorder in some cases then this would contribute to a predisposition to respiratory infection, and there may also be an element of ventilatory restriction akin to that found in asphyxiating thoracic dystrophy.

It is suspected that costovertebral dysplasia is under-reported and often not recognised as a separate entity. Patients with milder degrees of spondylocostal dysplasia, or with similar radiographic changes, are likely to be seen in scoliosis or growth clinics. Recognition of the disorder is important because of the genetic implications.

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References

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