Thoracic-pelvic dysostosis: a 'new' autosomal dominant form

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SUMMARY A form of thoracic and pelvic dysostosis is reported in a mother and her son. The short ribs caused respiratory distress in the baby and raised the possibility of asphyxiating thoracic dystrophy (ATD). The radiological features, however, distinguish this benign condition from ATD and other described skeletal dysplasias.

A wide variety of conditions may result in respiratory distress in the neonatal period. Respiratory distress may occur in skeletal disorders as a result of a collapsible larynx (campomelic dysplasia), or a collapsible chest wall (hypophosphatasia and osteogenesis imperfecta), or because of very short ribs. The short rib syndromes can be divided into three groups.

One group comprises generalised skeletal dysplasias with marked shortening of all bones including the ribs. These conditions have been extensively reviewed.¹ Respiratory distress is a constant feature in some of them, for example, thanatophoric dysplasia, achondrogenesis, and the short rib-polydactyly syndromes. In others it is an occasional problem in severe cases (for example, achondroplasia, diastrophic dysplasia). These conditions can be distinguished on the basis of specific radiological findings of the skull, spine, and limbs.^{1 2}

There are isolated reports of short ribs in other named generalised skeletal dysplasias, such as metaphyseal chondrodysplasia,³ and isolated cases not fitting into any named category, such as the case of narrow thorax, mesomelic shortening, and hexadactyly.⁴

Conditions in the second group show variable limb reduction and short ribs with a normal skull and spine and include asphyxiating thoracic dysplasia (Jeune) McKusick 20850 (ATD) and chondroectodermal dysplasia. These have similar radiological features, but can be distinguished clinically.^{5 6}

The third group contains cases of isolated thoracic dysostosis which do not fit into either of the other categories.^{4 7 8}

We report another short rib syndrome showing autosomal dominant inheritance and a good

prognosis. Thoracic and pelvic dysostosis was present in a mother and her son.

Case report

This male infant was the second child of unrelated parents. Delivery was at 39 weeks' gestation by elective lower uterine caesarian section because of a small pelvic inlet. Apgar scores were 9 at 1 minute and 10 at 5 minutes. Birth weight was 2800 g. He had a small chest and a protuberant abdomen. The liver and spleen were palpable 3 cm and 2 cm below the costal margins respectively. The limbs appeared normal. There was no evidence of dental, gum, or cardiac anomalies.

He developed mild respiratory distress which required supplemental oxygen for 12 hours. On day 5, following an episode of aspiration, there was further respiratory distress; the respiratory rate was 70 breaths/minute. Chest circumference was 28 cm (3rd centile) compared to the head circumference of 36.5 cm (55th centile). The respiratory distress required observation in hospital for 6 weeks and tachypnoea persisted until 18 months. Despite this he thrived and developed normally. His growth followed the 3rd centile.

At 4 years of age he developed mild episodic asthma and still had a narrow pigeon chest deformity. At 5 years of age his chest circumference equalled his head circumference. When reviewed at 8 years of age he was of normal intelligence and had no residual respiratory problems. His vital capacity was 1.4 l with an FEV1 of 70%, indicating a combination of mild obstructive airways disease and a reduced lung volume. His blood pressure was 110/65 and there was no evidence of renal disease. He was short (10th centile) and he had normal body proportions (upper segment : lower segment = 1.1:1). Chest circumference was 60 cm and head circumference

FIG 1 Radiographs of the chest on day 5 showing high clavicles and short ribs.

small and shallow sciatic notch and poor development of the acetabulum. The acetabular rim was poorly defined, in striking contrast to the acetabular spurs which give the trident appearance seen in ATD. At 7½ years of age his chest was normal in size, but still bell-shaped (fig 2). The upper chest was relatively narrow and there was a mild pigeon chest deformity. The acetabulum was normal, but the ilium remained small, rounded, and irregular with small sciatic notches (fig 3). The hands, spine, and skull were normal.

The mother's chest was bell-shaped like her son's, with a narrow upper chest (fig 4). Her pelvic x-rays were more unusual than her son's (fig 5). The ilium was short and had a tapered neck above the acetabulum. Sciatic notches were small resulting in a

FIG 2 Chest radiograph at 5 years of age showing the bell-shaped configuration. The chest was normal in size.

53.9 cm. There were no dysmorphic features. He wore corrective lenses for hypermetropia with no evidence of retinal dysplasia.

His mother's height was 151 cm (3rd centile). Her body proportions were normal (upper segment: lower segment = 1:1) and she had narrow hips. Her head circumference was 55 cm. She gave no history of respiratory problems in childhood and there was no information about the size of her chest as a child.

The neonatal x-ray of the chest confirmed the presence of short ribs and high clavicles (fig 1). There was very little flaring of rib ends and no cupping. The ilium was rounded and small with a

FIG 3 Radiograph of the pelvis at 5 years of age. The appearance is more normal but the ilium small, rounded, and irregular with shallow sciatic notches.

FIG 4 Radiograph of the mother's chest showing a bell-shaped configuration.

FIG 5 Radiograph of the mother's pelvis showing the small heart-shaped pelvic inlet and short ilium with a narrow neck above the acetabulum.

very small heart-shaped pelvic inlet, the feature which necessitated delivery by lower uterine caesarian section. Her hands, spine, and skull were radiologically normal.

Discussion

We believe that this condition is distinct from

previously described skeletal dysplasias. The abnormality was confined to the ribs and pelvis and there were no cardiac or ectodermal changes. The only named dysplasia requiring serious consideration is ATD (Jeune).⁹

ATD is a heterogeneous syndrome.^{4 9 10-14} There are specific radiological changes in the chest and pelvis (similar to those seen in chondroectodermal dysplasia) and in the hands. In one of the reports of ATD the appearance of the pelvis is not described¹⁵ and in others it is reported as normal.^{4 7 16} It can be argued that cases without pelvic changes should not be classified as ATD. Indeed, one of these cases¹⁶ was subsequently republished as metaphyseal chondrodysplasia.³

Respiratory failure occurred in the majority of reported cases of ATD and the degree of respiratory difficulty has not always been proportional to the degree of narrowness of the chest, raising the possibility of pulmonary hypoplasia.^{12 17} Renal failure has developed in mid-childhood in a number of patients who survived the respiratory insufficiency or never suffered respiratory difficulty.^{11 12 15 18} The renal changes probably progress during childhood; normal kidney histology has been reported in children dying of respiratory complications in the neonatal period.^{10 12} However, renal involvement has been described even in the first few days after birth.¹² The renal pathology is reminiscent of that seen in nephronophthisis and includes glomerular and tubular cysts as well as an increase in 'embryonic' connective tissue.^{12 15 18} Other associated anomalies have included portal fibrosis with bile duct proliferation^{12 18} and retinal aplasia.¹⁹ Two sibs described as ATD with pancreatic insufficiency¹⁶ were subsequently reclassified as metaphyseal chondrodysplasia.³ A few patients have survived into late childhood or adult life^{7 12 20} and some of these patients have shown marked improvement in pulmonary function and radiological appearance of the chest and pelvis.^{2 21} These patients may have a condition which is different from that in the babies who die early in life.

ATD is recessively inherited. There was an isolated claim of dominant inheritance in a mother and her child.⁷ However, the baby in this family had a small larynx. This is probably a different condition.

In the baby we have described the degree of respiratory distress was less than expected considering the shortness of the ribs. The child made a good recovery and appears to have a lung capacity just below normal limits at the age of 8. He has no evidence of retinal or renal disease and the radiological appearance of his hands is normal. The changes in the pelvis were not those of ATD.

This form of thoracic pelvic dysostosis appears to be dominantly inherited and to run a benign course.

The chest and pelvic changes and the finding of normal limbs are sufficiently different to distinguish this form of dysostosis from ATD and from reports of isolated thoracic dysostosis with limb abnormalities.⁴ It is also distinct from the thoracic dysostosis reported by Rabushka *et al*⁸ in a father and four of his children. None had neonatal respiratory problems but they did have chest infections and asthma in childhood. The radiological changes in the chest showed a bell-shaped thorax with short ribs which had an irregular contour posteriorly. The rest of the skeleton was normal.

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