INFANTILE THORACIC DYSTROPHY*

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Infantile thoracic dystrophy is a rare generalized skeletal abnormality, familial in character, which leads to severe chest wall deformity and early onset of respiratory complications. This disease, of which 28 cases only have been reported, has features typical enough to merit separate classification in the spectrum of the osteochondrodystrophies.

A further observation is presented here together with a brief discussion.

CASE REPORT

V.J., a 2-month-old Coloured boy, was admitted to hospital with increasing respiratory difficulty and intermittent cyanosis of a few days' duration. Past history disclosed that while under medical care because of mild icterus in the first week of life a chest wall anomaly had been noticed, in addition to easily palpable abdominal viscera.

On examination, the baby was pyrexial, in moderately severe respiratory distress, tachypnoeic and slightly cyanosed, with marked subcostal retraction but minimal thoracic movements. Air entry was decreased over the right lung, and râles were heard bilaterally. A general oddity about his body proportions retained one's attention (Fig. 1). His chest was narrow, elongated and very small relative to the size of the head and abdomen. The costochondral junctions were thickened and formed a well-marked rosary in the anterior axillary line, a position much more lateral than usual. Despite the severe distress the thorax was almost immobile and the respirations were purely diaphragmatic. The abdomen appeared distended, the edges of the liver and the spleen were 3 cm. below the costal margins, and both kidneys were easily palpable. The extremities were short relative to the body length and the skull was flattened on the left side.

Routine blood cell counts, urinalysis and serum calcium, phosphorus and alkaline phosphatase values were normal. No mucopolysaccharides or phospho-ethanolamine were demonstrable in the urine.

X-ray studies revealed a narrow and elongated thorax, scattered blurry pulmonary opacities and consolidation of the right upper lobe (Fig. 2). The clavicles were situated abnormally high. The ribs were broad and short, the upper ones projecting almost horizontally. Their distal ends were widened and club-shaped and did not extend beyond the anterior axillary line (Fig. 3). The pelvis was small, the iliac wings appearing hypoplastic and squared.

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The acetabular roofs were flattened and had a saw-tooth appearance (Fig. 4). The femurs were slightly shortened, with widened metaphyses. The upper extremities also showed some shortening of the long bones, and the skull and spine were within normal limits.

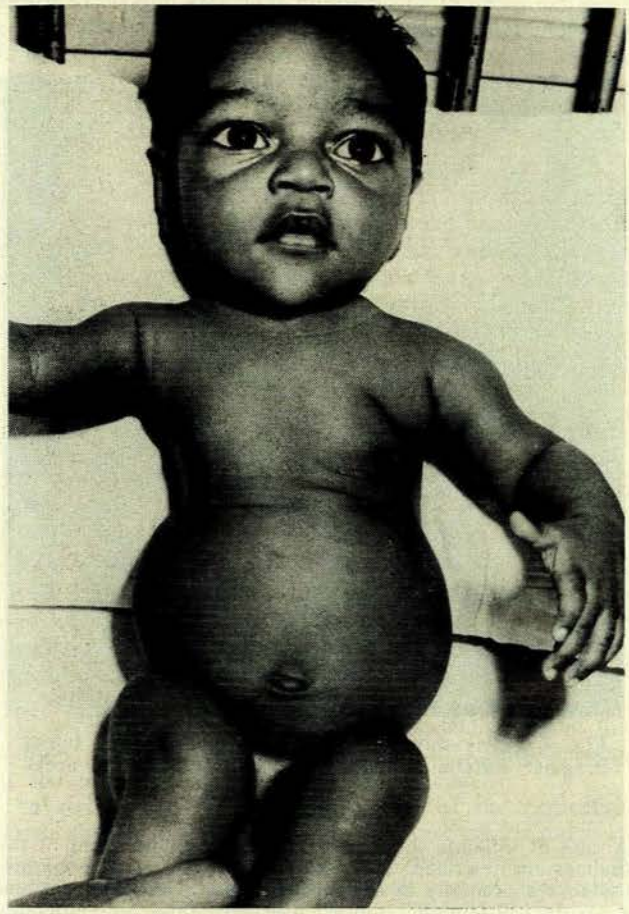

Fig. 1. A view of the patient sitting up, emphasizing the marked thoracic deformity.

Satisfactory response occurred to therapy with antibiotics and oxygen. When transferred to a convalescent home, the patient's chest was clear but subcostal recession persisted. Both parents and 5 siblings were examined and showed no clinical or radiographic abnormalities.

DISCUSSION

Infantile thoracic dystrophy was first described by Jeune *et al.*[1] in 1954 and was termed 'asphyxiating thoracic dystrophy'. Further publications followed in the European[2-5] and American[6,7] literature, bringing the total number of reported cases to 28.

In this disorder all sections of bone may be involved, and the degree of involvement of different parts of the skeleton is quite variable. Most severely affected are the cartilaginous elements of the ribs, resulting in marked thoracic deformity. This constant finding gives the patients an undoubted individuality. The short and hypoplastic ribs with their swollen distal ends result in a small, immobile thorax and a rachitic type of rosary situated far back in the anterior or medial axillary line. Minimal chest movement occurs and the respirations are mainly abdominal. Because of this impairment of respiratory function, these patients are prone to recurrent pulmonary infections leading to severe respiratory distress, asphyxia and death. The pelvis is small, the iliac wings appear squared, and saw-toothing of the flattened acetabular roofs may be present. The long bones tend to be short and wide, with splayed metaphyses. The skull and spine are normal.

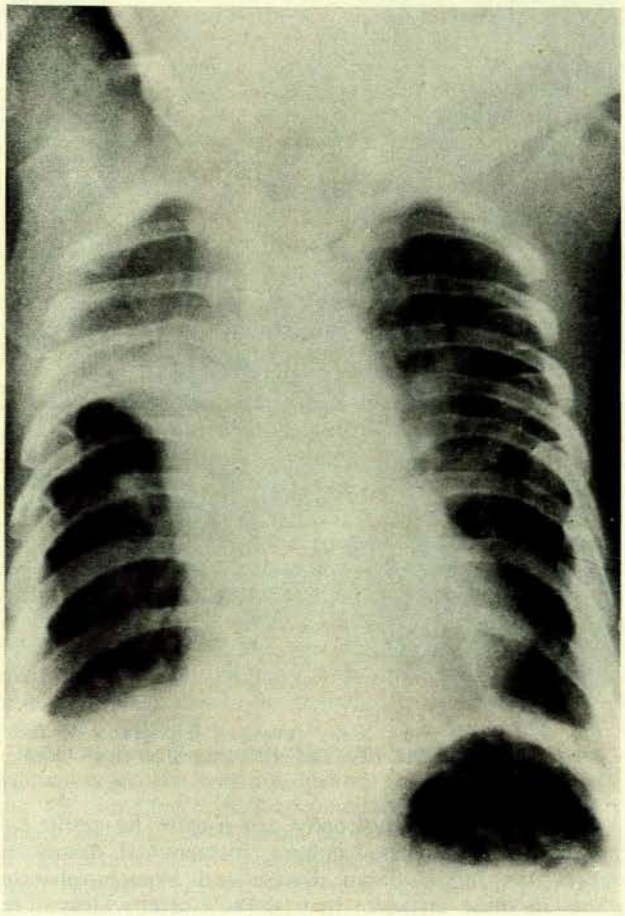

Fig. 2. Chest X-ray showing, in addition to pulmonary pathology, the characteristic narrow thorax and abnormally high clavicles.

The skeletal deformities may be gross and obvious at birth, or they may be minimal and detected only on X-ray examination. In its severe form, the progress of the disease is rapid. Respiratory difficulties occur almost continuously and most patients die during their first year of life. Less affected patients may have minimal respiratory problems which tend to decrease with age. Survivors either show negligible growth of the thoracic cage, or marked improvement occurs over the years, the ribs becoming essentially normal in length and texture. The familial character of the disorder is evidenced by a number of publications reporting several affected siblings.

The only available histological study[3] showed disordered endochondral ossification at the costochondral junctions, with a limited and irregularly vascularized zone of provisional calcification. The continuity between the cartilage columns and bone was disturbed, and a proliferation of cartilage and an abnormal fibrocollagenous tissue was responsible for the widened appearance of the distal costal metaphyses.

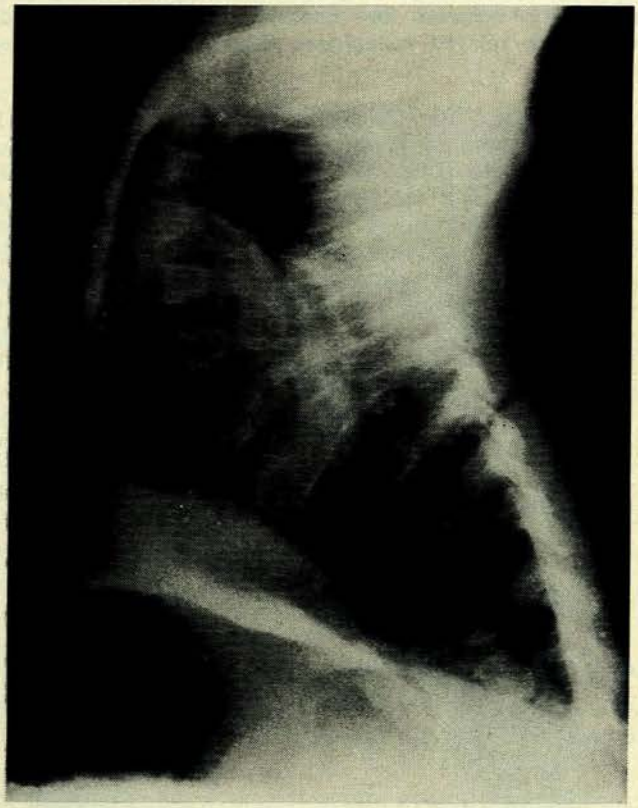

Fig. 3. Lateral chest X-ray revealing hypoplasia of the anterior part of the ribs and club-shaped costochondral junctions.

Infantile thoracic dystrophy can usually be easily differentiated from achondroplasia, metaphyseal dysostosis, rickets, Werdnig-Hoffman disease and hypophosphatasia which in some instances may show a certain distant resemblance. The existence of dental anomalies and polydactylism in a few patients with infantile thoracic dystrophy[4] suggests a possible link with the Ellis-Van Creveld syndrome. However, the rarity of thoracic abnormalities in the Ellis-Van Creveld syndrome and the very common occurrence of a cardiac malformation suggest that these two diseases are quite distinct.

Careful observation and early treatment of the respiratory complications, including the use of assisted ventilation, may help to tide patients with infantile thoracic dystrophy over the critical first 2 years of life. There is increasing evidence that in later childhood respiratory distress is less severe and that the chondrodystrophic changes may even be reversible. It has therefore been suggested[7] that the original term 'asphyxiating' be deleted from the name of this disorder, as minor regressive forms with long-term survival are being reported.

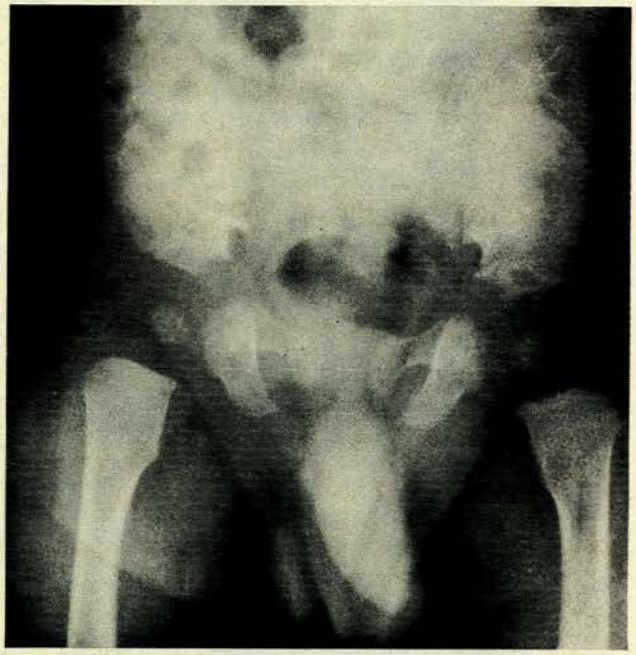

Fig. 4. Pelvis showing hypoplastic squared iliac bones, irregular acetabular margins and widened femoral necks.

SUMMARY

A case of infantile thoracic dystrophy is reported and X-ray findings are described. This rare generalized skeletal abnormality results primarily in an inadequate costal development and a narrow and immobile thorax. Affected infants are vulnerable to repeated respiratory infections, which often prove fatal. Although the disorder is still difficult to classify amidst the osteochondrodystrophies, it seems justified, because of its typical features, to regard it as a distinct entity.

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