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Thoracic–pelvic dysostosis

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We report a third patient, a female, with thoraco-pelvic dysostosis. This rare disorder is similar in phenotypic and radiographic appearances to thoraco-laryngo-pelvic dysplasia (Barnes syndrome). The only major difference between these two diseases is absence of laryngeal involvement in thoraco-pelvic dysplasia. They may represent two different entities or a contiguous gene syndrome.
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INTRODUCTION

In 1969 Barnes *et al.* reported a case of bone dysplasia characterized by thoracic dystrophy, laryngeal stenosis and a small pelvis, which they named thoracolaryngo-pelvic dysplasia (TLPD). Reassessment of the Barnes family in which a mother and two children – including the original Barnes case – were affected, was published by Burn *et al.* in 1986. In 1983 Bankier and Danks described two patients with a phenotype similar to the Barnes case, under the title of thoracic-pelvic dysostosis (TPD). The radiographic appearances of the chest and pelvis showed a close resemblance. The significant difference was the absence of a laryngeal abnormality in the Bankier and Danks patients. We report a third patient with thoracic-pelvic dysostosis (TPD).

CASE REPORT

This female was born at 39 weeks of gestation to a 32-year-old, para 5-2-3, healthy mother. The first two pregnancies were spontaneous abortions of unknown cause. The oldest sister was operated on because of a thoracic deformity resulting in constrictive pulmonary disease. The older sister was normal. The father was healthy but died suddenly of an unknown cause at the age of 38 years.

At the age of 18 years she was referred to the Centre for Patients with Defects of the Locomotor System in Prague from Department of Medical Genetics in Brno with the diagnosis Asphyxiating Thoracic Dystrophy. Her karyotype was 46,XX,13ps+.

At the age of 18 years her height was 146 cm, weight 33 kg (Figure 1). There were no dysmorphic facial features. She never had any serious respiratory problems. Her voice was normal. She has been treated with a Milwaukee brace for 1 year. Her proportions were normal. There was a scoliosis convex to the right in the thoracic and upper lumbar spine. The thorax was flat and narrow in transverse diameter, particularly in its lower part. The manubrium sterni and the clavicles were prominent. There was some anterior tilt of the pelvis. The elbow and knee joints were in a 10° flexion position. There was increased internal and decreased external rotation of the hips consistent with increased anteversion of the femoral necks. The lungs, the heart and the abdomen showed no abnormality.

The biochemical examinations were normal. Radiographic studies documented diagnostic findings in the chest and pelvis. The chest was deformed – narrow, particularly in its lower part. The posterior parts of the 1–9 ribs were in a horizontal position, whereas the

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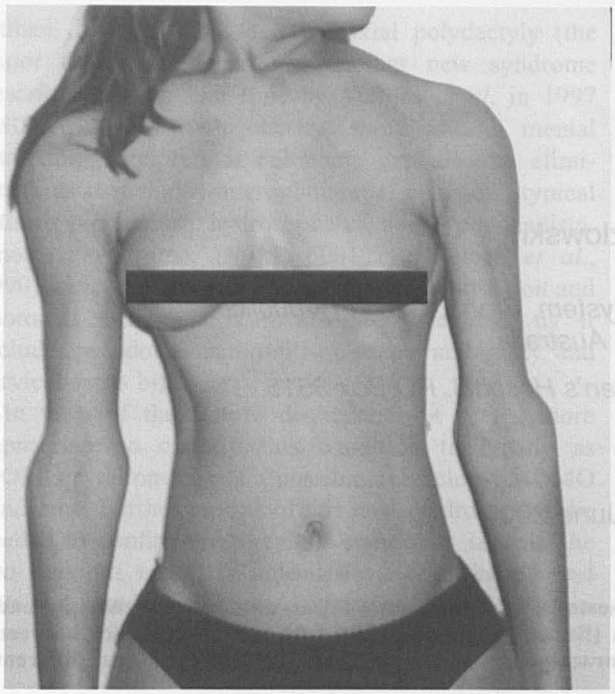

FIGURE 1. 18 year old. Normal face. Deformed chest, narrowed in its lower part

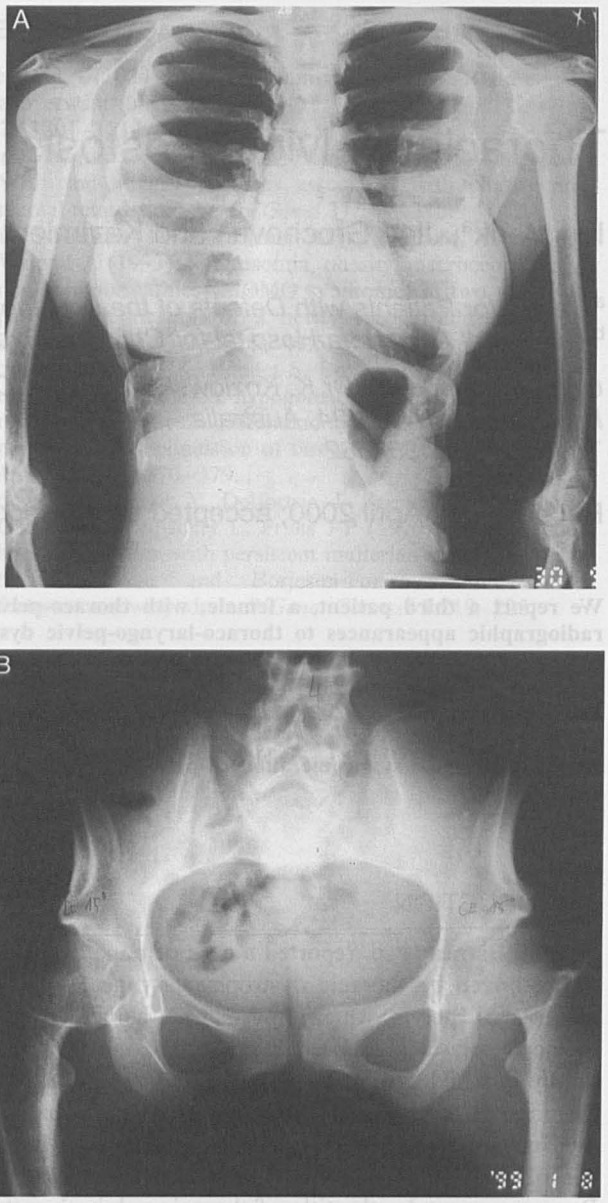

FIGURE 2 (A and B) 18 year old. (A) The chest is elongated and narrowed in its inferior part. The 1–9 ribs are in their posterior part horizontal in position, whereas 10–12 ribs are oblique in position and narrowed posteriorly. Note mild dysplastic changes of the bones of the shoulder region. (B) High iliac angles. Hypoplastic bodies of the iliac bones. Shallow, slightly irregular acetabular cavities. Short femoral necks. Oval pelvic inlet. The lumbar canal was rectangular in shape with constant interpedicular distances L1-L5

remaining 10–12 ribs were in the oblique position and narrowed in the posterior part. There was right sided scoliosis in the thoracic and upper lumbar spine. The vertebral bodies were slightly bi-concave. The upper airways showed no abnormality (Figure 2A). In the pelvis both the bodies and the wings of the iliac bones were hypoplastic. Both the acetabular and particularly the iliac angles were high. The pelvic inlet was oval shaped. The femoral necks were short (Figure 2B).

Non-diagnostic bone changes included thin fibulae with proximal shortening and distal shortening of the ulnae. There was some bowing of the humeri and the forearm bones. The scapulae were small and the clavicles thin and slightly bowed superiorly.

DISCUSSION

There is little doubt that our patient – although slightly more severely affected – has the same disorder as that reported by Bankier and Danks in 1983 and designated as TPD. TPD is easily recognizable as the diagnostic findings are localized to the chest and pelvis with minor or borderline findings in the remaining skeleton. The only disorder with a similar phenotype and similar radiographic findings is thoraco-laryngo-pelvic dysplasia (TPLD) (Barnes *et al.*, 1969; Burn *et al.*, 1986). The main difference is laryngeal stenosis and proneness to respiratory tract infections, features absent in TPD.

Asphyxiating thoracic dystrophy (ATD) can be easily

differentiated. The shape of the thoracic cage in early, severe ATD is more rectangular. In the benign forms of ATD the deformity of the chest is less marked but the triradiate acetabulum of ATD cannot cause confusion. Finally the cone-shaped epiphyses, a common major sign of ATD are absent in TPD and in TPLD. In thoracic dysostosis only the thorax is affected (Rabushka *et al.*, 1973). Other bone dysplasias which severely affect the chest such as metatropic dysplasia should cause no confusion.

Both TPD and TPLD are very rare diseases. Taybi and Lachman (1996) regard them as the same disorder. It is possible that they represent different expressivity of the same disease or are a contiguous gene syndrome. However, until patients with TPD and TPLD in the same family are reported, or gene mapping of both disorders is successful, the relationship between TPD and TPRD remains uncertain.

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