

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

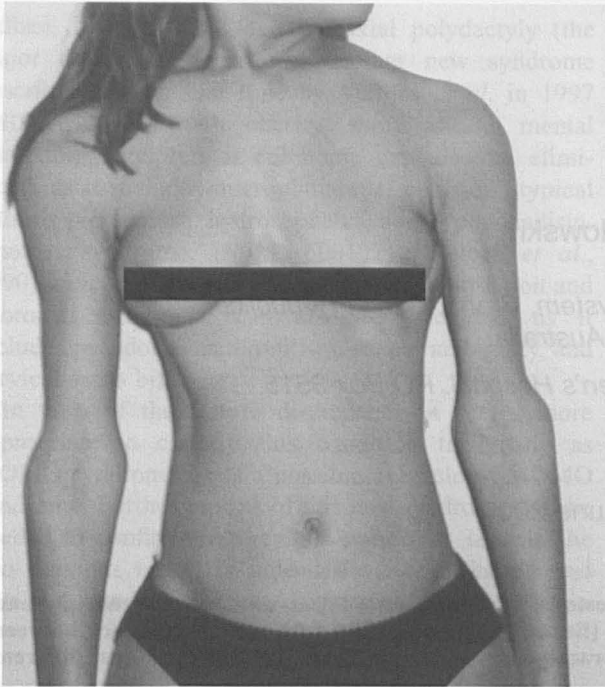


FIGURE 1. 18 year old. Normal face. Deformed chest, narrowed in its lower part

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 dyspla-

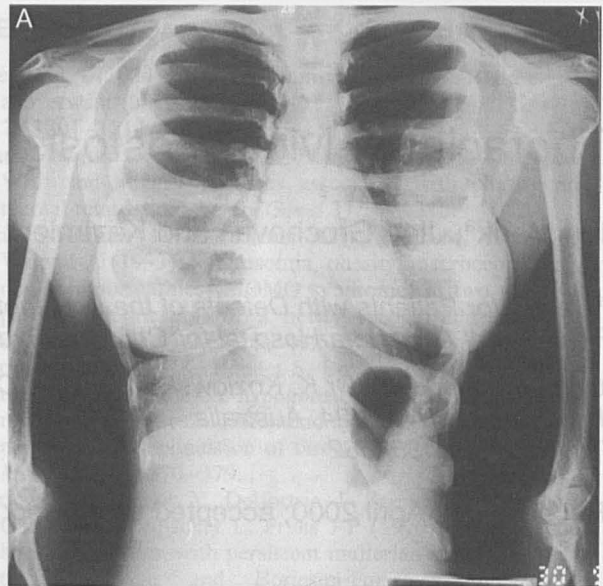


FIGURE 2 (A and B) 18 year old. (A) The chest is elongated and narrowed in its inferior part. The 1–9 ribs are in their posterior part horizontal in position, whereas 10–12 ribs are oblique in position and narrowed posteriorly. Note mild dysplastic changes of the bones of the shoulder region. (B) High 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

sia (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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A case of Varadi-Papp syndrome in a 3-year-old boy whose case has been followed from 8 months of age is described. *Clin Dysmorphol* **9**: 289-290 © 2000 Lippincott Williams & Wilkins

Keywords: oral-facial-digital syndromes, polydactyly, Y-shaped metacarpel, Dandy-Walker malformation.

The oral-facial-digital syndromes (OFDS) are a heterogeneous group of disorders characterized by facial anomalies, oral malformations and digital abnormalities. At least three types of OFDS have been reported. Vandyke syndrome or OFD type V differs from the other two because of cerebral dysgenesis and cerebral atrophy of the brain (Toriihara, 1997).

We report a 3-year-old boy who presents many clinical features of Vandyke syndrome. The patient has been followed since the age of 8 months. He is the product of a 42-year-old woman complicated by threatened miscarriage. The placenta was circumferential and breech. There was a possibility for any birth defects. The baby was born by emergency section because of fetal distress. Birthweight was 1400 g. Birthlength was 50 cm. At birth examination showed that the right hand had central and postaxial polydactyly type B with partial syndactyly between 3rd and 5th fingers; the left hand showed central polydactyly. The feet had bifid talons. A large skin nevus was present. At the age of 4 months he underwent surgery for removal of the extra metacarpal digit of the right hand. He came to our attention for the first time at the age of 1 year. Weight was 9.2 kg (90th centile), length was 68 cm (70th centile), OAR was 45 cm (90th centile). He had brachycephaly with a large anterior fontanel (3.5 × 2.5 cm), broad flat occiput, downslanting palpebral fissures, epicanthic folds, a small nose with a broad nasal bridge and a long philtrum. The mouth was large with a thin upper lip, the tongue was lobulated with a hamman-ricus mass. A high arched palate and retrognathia were

present. In the right hand central polydactyly and a scar on ulnar side due to previous surgical excision were present, on the left hand central polydactyly was present (Figure 1a). The feet showed a bilateral bifid talon and as above mentioned, bilateral syndactyly between 1st and 2nd toes (Figure 1b). On respiratory examination wheezing was present. There was a small umbilical hernia and a nevus flammeus in the sacral region. Neurological examination revealed mild psychomotor retardation and generalized hypotonia. Oral examination with mandibular opening was present. Hand radiographs revealed a bifurcated right third metacarpal with an extra, fully formed ventral digit. Both feet had broad first metatarsals and duplication of the phalanx of the hallux. On laryngoscopy a larynx malformation consisting of a multilobulate epiglottic edge was evident. Brain-MRI scanning showed a hypoplastic cerebellar vermis and a minor Dandy-Walker anomaly. At the age of 2 years the patient manifested an episode of afebrile seizure. At the age of 3 years he began to stand up without walking.

This patient has many of the clinical features of OFD type VI (Varadi-Papp syndrome) (Varadi *et al.*, 1980). Syndactyly, brachydactyly, clinodactyly and occasionally preaxial and/or postaxial polydactyly have been mentioned in patients with OFD type VI and the most frequent hand anomaly is axial or central polydactyly that is defined as a fully developed extra digit arising from an additional central metacarpal or from a bifid or Y-shaped, usually, third metacarpal. The facial anomalies are midline or median and lip and/or highly arched or cleft