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# A CASE OF BRACHYOLMIA*

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**SUMMARY: Karabıyık N, Oğuz F, Sıdal M, Hekim N, Kayserili H. (Department of Pediatrics, İstanbul University İstanbul Faculty of Medicine, İstanbul, Turkey). A case of brachyolmia. Turk J Pediatr 1997; 39: 415-420.**

**Brachyolmia refers to a form of skeletal dysplasia characterized by general platyspondyly without significant epiphyseal, metaphyseal or diaphyseal changes in long bones. Three, possibly four, types of brachyolmia have been defined: Type I-Hobaeck-Toledo type, Type II-Maroteaux and Type III. We report a patient with brachyolmia and present the clinical and radiological findings. A 15-year-old boy presented to our Outpatient Department because of his short stature. His height, weight, head circumference and arm span were 127 cm (<3rd percentile), (3rd percentile) 39 kg, 55 cm (50th-75th percentile), and 142 cm respectively, and his upper segment/lower segment ratio was 0.91. His neck and trunk were short. He had severe kyphoscoliosis. Slit-lamp examination was normal. Radiologic features included platyspondyly in cervical, thoracic and lumbar vertebrae as well as kyphoscoliosis. Bilateral coxa valga and mild acetabular irregularities were noticed on pelvic radiographies. Levels of chondroitin and heparan sulphate as well as the glycosaminoglycan/creatinine ratio were elevated in the 24-hour urine specimen. The activities of N-acetylgalactosamine-6-sulphatase, β-galactosidase and β-hexosaminosidase were all normal in fibroblast culture. Although the x-ray findings of this patient are consistent with both Types I and III, recessive inheritance and glycosaminoglycan anomalies point to Type I brachyolmia.** *Key words: skeletal dysplasia, brachyolmia, dwarfism.*

The brachyolmia are associated with short-trunk dwarfism, and major radiographic changes are present in the spine. Usually there are no epiphyseal, metaphyseal or diaphyseal changes in the long bones. There are three, possibly four, types of brachyolmia. Type I (Toledo-Hobaek Type), Type II (Maroteaux Type), and Type III (Dominant Type). In this paper, we describe a male patient with the radiological features of brachyolmia.

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## Case Report

The patient, a 15-year-old male, was the third child of first-degree-cousin parents (G6 P6 Ab0). The pedigree is shown in Figure 1. The second, fourth, fifth and sixth children were an 18-year-old girl, a 14-year-old boy, a 13-year-old boy and a 6-year-old girl with heights of 166 cm ($75^{th}$ percentile), 134 cm ($<3^{rd}$ percentile), 116 cm ($<3^{rd}$ percentile), and 98 cm ($<3^{rd}$ percentile), respectively. Except for the second child, all the children were described as having bowed legs (Fig. 2). The father was 178 cm ($75^{th}$ percentile) and the mother was 158 cm tall ($50^{th}$ percentile). Although we could not examine the siblings who lived far away from our hospital, their photographs and heights were available.

The family noticed our patient's short stature when he was seven years old. His birth length was normal, as was his somatic and mental development. When he was 15 years old, his height was 127 cm ($<3^{rd}$ percentile), weight was 39 kg ($3^{rd}$ percentile), head circumference was 55 cm ($50^{th}$-$75^{th}$ percentile), arm

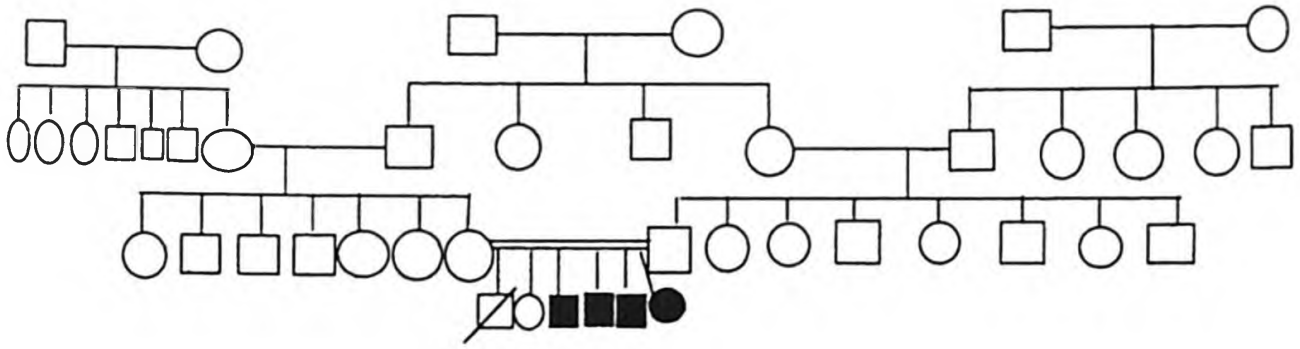

Fig. 1: Our patient's pedigree.

Fig. 2: Our patient's three siblings with short stature and bowed legs.

span was 142 cm and the upper segment/lower segment ratio was 0.91. His facial features appeared somewhat coarse with a wide nasal bridge. He had broad hands, a short neck, a short trunk and kyphoscoliosis but no organomegaly (Fig. 3). Slit-lamp examination did not reveal corneal clouding or punctations.

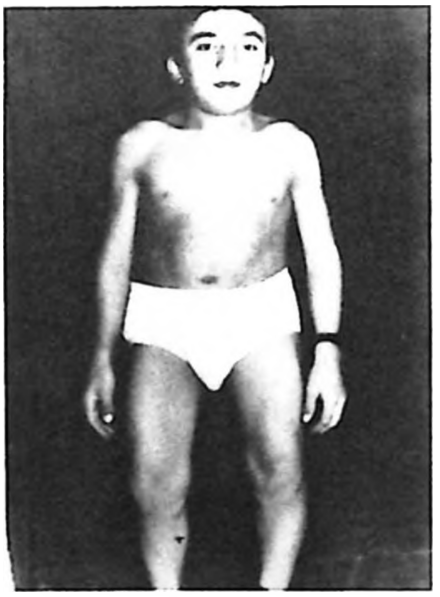

Fig. 3: Our patient: short-trunk dwarfism, coarse face and short neck are evident.

*Radiological Features*

His bone age was 14 years (Fig. 4). The thoracic and lumbar vertebrae showed definite signs of platyspondyly and severe kyphoscoliosis (Figs. 5a and 5b). The intervertebral spaces were markedly narrowed. Cervical vertebrae also showed platyspondyly (Fig. 5c). The skull was normal. Bilateral coxa valga and

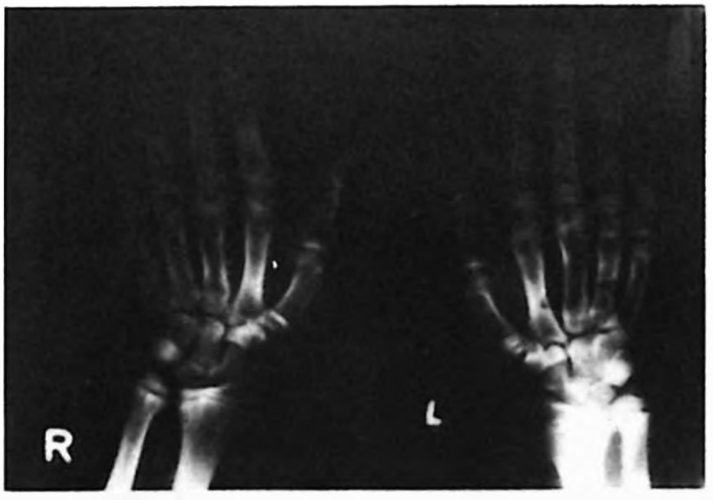


Fig. 4: Our patient's radiograph of hand and wrist.

mild irregularity of the acetabular roof without any epiphyseal or metaphyseal changes were noticed (Fig. 6). There was no precocious calcification of the falx cerebri or costal cartilage.

Fig. 5a: Lateral radiograph of the spine showing marked platyspondyly, kyphosis and rectangular vertebral bodies.

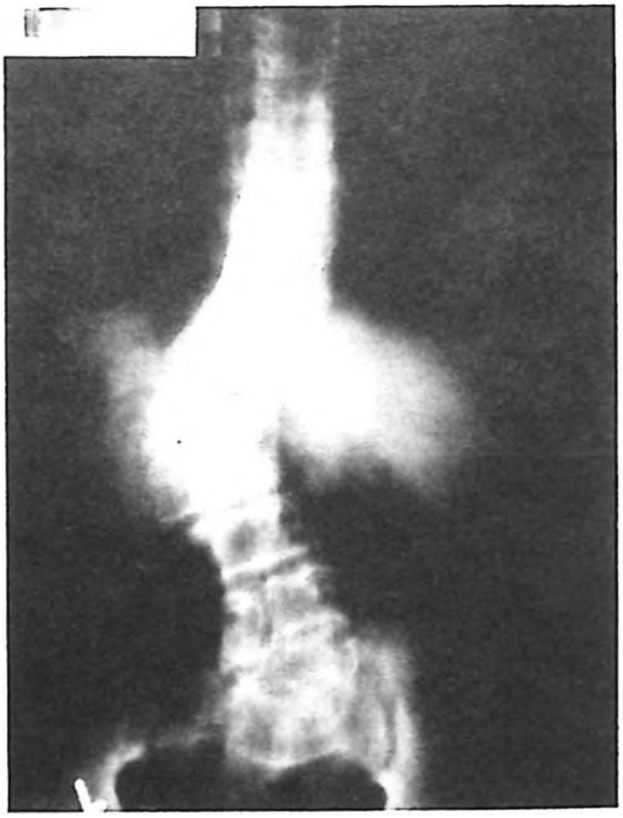

Fig. 5b: Anteroposterior raoliograph of the spine shawing scoliosis, platyspondyly and narrow entervertebral spaces.

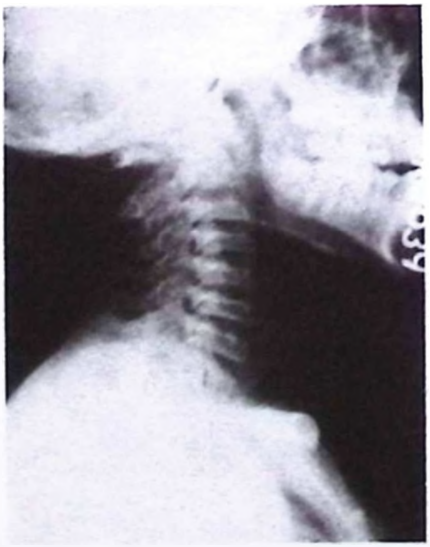

Fig. 5c: Lateral radiograph of the cervical spine showing severe flattening of the vertebral bodies.

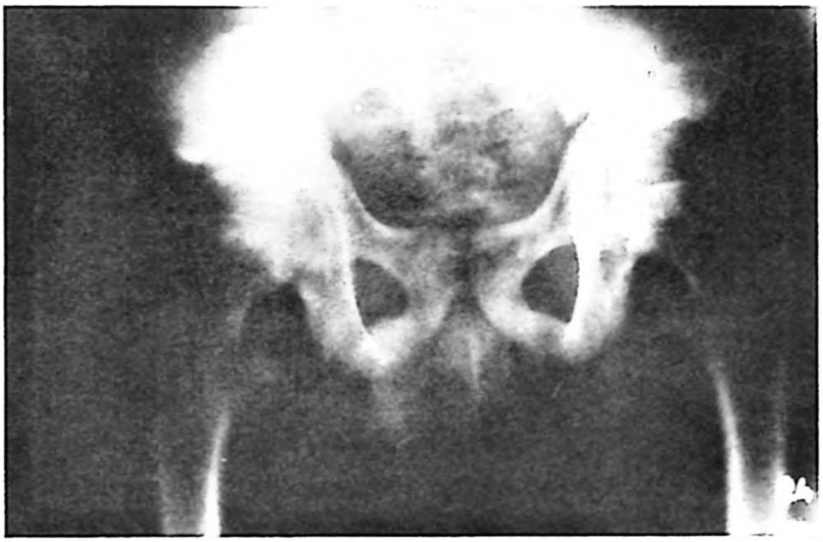

Fig. 6: Anteroposterior radiograph of pelvis and hips showing coxa valga and mild acetabular irregularities.

*Laboratory Findings*

Calcium, phosphorus and alkaline phosphatase levels were normal. A 24-hour urine glycosaminoglycan electrophoresis revealed increased levels of chondroitin sulphate and heparan sulphate with an elevated urine glycosaminoglycan/ creatinine ratio of 8 (London Hospital for Sick Children). N-acetylgalactosamine-6-sulphatase, β-galactosidase and β-hexoaminidase activities were normal in fibroblast culture.

## Discussion

The term "brachyolmia" comes from the Greek word for short trunk. In 1969 Maroteaux[1] used the term brachyolmia to describe short-trunk dwarfism and generalized platyspondyly. Shohat et al[2] have defined at least three distinct types of brachyolmia. Type I (Hobaek Type) is inherited as an autosomal-recessive trait and is associated with squared-off platyspondyly, narrow intervertebral spaces, marked endplate irregularity and close-set pedicles on anteroposterior view. Elongated rectangular vertebral bodies are seen on lateral radiographic views. The greatest flattening is in the thoracic region. The ilia may be somewhat shortened, but the acetabular and femoral heads are normal. The femoral neck may be short and broad. Scoliosis may also be present, although it is not as common or severe as in other types. The Hobaek type seems to be most common type of brachyolmia[2,3]. In 1978 Toledo et al[4] described a family with similar radiographic changes and qualitative glycosaminoglycans (GAG) anomalies, mostly involving chondroitin-6-sulphate in the urine, but the affected patients

also had corneal opacities and precocious anterior rib calcification. Later, Sewell et al[5] also reported qualitative glycosaminoglycans anomalies in one case with Type I brachyolmia.

Type II (Maroteaux Type) cases differ from Type I in having rounded vertebral edges, mild endplate irregularities, and normal or broadened intervertebral disc spaces. Mild facial anomalies have been described. Precocious falx cerebri calcification can be seen. This condition is transmitted as an autosomal-recessive trait[2].

Type III has been described in a mother and son in which there was severe involvement of the vertebrae similar to that in Type II, as well as severe cervical platyspondyly. Type III was described in this family, suggesting autosomal-dominant inheritance. These patients also had the most severe scoliosis and demonstrated marked cervical vertebral flattening and irregularity[2]. A report from South Africa in 1994[6] described a mother and her son as having autosomal-dominant type brachyolmia, but they differed from previously documented autosomal-dominant cases of brachyolmia in terms of the severity of their platyspondyly and the shape of their vertebral bodies.

There is some controversy concerning the classification of this disorder. In our case, a first-degree-cousin marriage, body habitus similar to siblings and some of the radiological findings suggest Hobaek-type brachyolmia, which is an autosomal-recessive disorder. However, the severe kyphoscoliosis and cervical vertebrae findings suggest Type III brachyolmia, which is an autosomal-dominant disorder. These radiological and genetic differences support genetic heterogenity of brachyolmia. As a result, our patient may be an example of this heterogenity. We conclude that further investigation is needed to classify brachyolmia.

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