

Brachyolmia

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[Abstract](#)

[Keywords](#)

[Disease name](#)

[Synonyms](#)

[Definition, Diagnostic Criteria, Clinical Description](#)

[Differential Diagnosis](#)

[Etiology](#)

[Genetic Counseling](#)

[Management](#)

[References](#)

Abstract

All types of brachyolmia are characterized by childhood onset short trunk-short stature and generalized platyspondyly without significant epiphyseal or metaphyseal changes in the long bones.

There are four main types:

The Hobaek type: short stature commencing in late childhood or early teens. The shortness is mainly limited to the trunk.

The Toledo type: probably identical to the Hobaek type with regard to the skeletal and vertebral changes, but patients also have abnormal excretion of glycosaminoglycans and peripheral punctate corneal opacities.

The Maroteaux type: similar phenotype of childhood onset, but with irregular and reduced intervertebral spaces and marked extension of the lateral margins of the vertebrae. Rounding of the anterior and posterior vertebral borders is present.

The Autosomal dominant type: symptoms somewhat less than in the other types. Most cases have scoliosis or kyphosis. This type follows autosomal dominant inheritance, while all other types are autosomal recessive

Besides these four types there are individual cases that can not be classified at present.

Based on the inheritance pattern of each type of brachyolmia, the appropriate genetic counseling can be given. Prenatal diagnosis is not available.

There is no specific treatment for any of the types of brachyolmia.

Keywords

Short stature, short trunk short stature, platyspondyly, vertebral changes

Disease name

Brachyolmia is derived from the Greek word meaning "short trunk".

Synonyms

Brachyolmia Hobaek type
Brachyolmia Toledo type (spondyloepiphysel dysplasia tarda Toledo type)
Brachyolmia Maroteaux type
Brachyolmia Autosomal dominant type

Definition, Diagnostic Criteria, Clinical Description

Hobaek type

The Hobaek type is characterized by short stature commencing in late childhood or the early teens. Final height is between 125 cm and 159 cm. A complication reported by Mukamel *et al.* (2003), was spinal stenosis. Psychomotor development is usually normal.

Radiological examination reveals platyspondyly of cervical, thoracic and lumbar vertebrae, and in addition may show mild scoliosis, short iliac bones and slightly short tubular bones. The intervertebral spaces are irregular and reduced and there is marked extension of the lateral margins of the vertebrae. Rectangular and elongated vertebral bodies, which become more pronounced with age, are seen on lateral views of the spine (Shohat *et al.*, 1989).

In some patients radiological studies also show a horizontal acetabular roof, short femoral neck, and vertical mixed lucent and dense striation pattern in the metaphyses of large long bones (Hoo and Oliphant, 2003). The disorder follows an autosomal recessive inheritance.

Toledo type

The Toledo type is radiographically similar to the Hobaek type. However, additional abnormalities allow distinction from the Hobaek type: peripheral punctate corneal opacities on slit lamp examinations, precocious ossification of costal cartilage, and a qualitative abnormal excretion of glycosaminoglycans (GAG) (Toledo *et al.*, 1978).

Sewell *et al.* (1991) described a girl with brachyolmia and a pathological urinary glycosaminoglycan excretion pattern – the amount of urinary glycosaminoglycans was normal, but there was an increased amount of an undersulphated chondroitin sulphate molecule. They suggested that this was a case of brachyolmia with a possible defect in chondroitin sulphate-sulphotransferase.

In another case with the typical changes of brachyolmia (Karabiyik *et al.*, 1997), slit-lamp ophthalmologic examination gave normal results, but the levels of chondroitin and heparan sulphate, as well as the glycosaminoglycan/creatinine ratio were elevated in a 24-hour urine specimen. This suggested that the distinction between the Hobaek and Toledo type may actually be artificial. The entity follows an autosomal recessive inheritance.

Maroteaux type

The Maroteaux type is clinically similar to the Hobaek. The patients have short trunk - short stature of childhood onset, and radiological examination shows universal platyspondyly, irregular and reduced intervertebral spaces and marked extension of the lateral margins of

the vertebrae. Psychomotor development is usually normal. The type is distinguished from the Hobaek and Toledo types by rounding of the anterior and posterior vertebral borders with less elongation on lateral view and less lateral extension on antero-posterior view, and may be associated with precocious calcification of the falx cerebri and minor facial anomalies (Shohat *et al.*, 1989).

In addition to the above findings some patients with the Maroteaux type also have a pectus excavatum, wide-spaced nipples, clinodactyly, hypoplastic nails, and hyperlaxity of joints (Shohat *et al.*, 1989). The entity is transmitted as an autosomal recessive trait.

Autosomal dominant type

The Autosomal dominant type is characterized by childhood onset short trunk - short stature, usually somewhat less severe than the autosomal recessive types. Final height is between 160 and 168 cm. Clinically the patients often have scoliosis with or without kyphosis. Radiological examination shows generalized platyspondyly. Irregular and reduced intervertebral spaces and marked extensions of the lateral margins of the vertebrae are also found. This type has the most severe vertebral changes with flattening and irregularities of the cervical vertebrae (Shohat *et al.*, 1989; Gardner and Beighton, 1994). A few cases with this type have been reported with metaphyseal irregularities in the femur and humerus (Brown and MacDonald, 1933; Lomas and Boyle, 1959). As a result, Kozlowski *et al.* (1982) concluded that pure brachyolmia does not exist and that metaphyseal involvement may be minimal and scattered but is always present. Ikegawa *et al.* (1995) described a patient with probably the dominant type who had atlantoaxial instability and failure of ossification of the dens. Psychomotor development is usually normal. As the name indicates the entity is autosomal dominantly inherited)

Differential Diagnosis

This depends on demonstrating the combination of the specific characteristic radiological and clinical features. The differential diagnosis is mainly with other skeletal dysplasias that show a short trunk dwarfism, but show in addition epiphyseal or metaphyseal dysplasia. Different forms of mucopolysaccharidoses should be excluded as well. The autosomal dominant type should

be differentiated from severe Scheuermann disease.

Etiology

Heterogeneous. Three types are transmitted by autosomal recessive inheritance and one by autosomal dominant. There is no biochemical or molecular diagnostic tool, except for the abnormal excretion of glycosaminoglycans in the Toledo type. All types of brachyolmia are infrequently described, and are seemingly rare.

Genetic Counseling

Based on the inheritance pattern of each type of brachyolmia, the appropriate genetic counseling can be given. Prenatal diagnosis is not available.

Management

There is no specific treatment for any of the types of brachyolmia.

References

Brown DO, MacDonald C. Three cases of familial osseous dystrophy. *Aust NZ J Surg* 1933;3:78-88.

Gardner J, Beighton P. Brachyolmia: an autosomal dominant form. *Am J Med Genet* 1994;49:308-312.

Hoo JJ, Oliphant M. Two sibs with brachyolmia type Hobaek: five year follow-up through puberty. *Am J Med Genet* 2003;116A:80-84.

Ikegawa S, Nakamura K, Nakamura S, Nagano A. Brachyolmia: a report of two cases. *J Pediatr Orthop* 1995;15:105-107.

Karabiyik N, Oguz F, Sidal M, Hekim N, Kayserili H. A case of brachyolmia. *Turk J Pediatr* 1997;39:415-420.

Kozlowski K, Beemer FA, Bens G, Dijkstra PF, Iannaccone G, Emons D, Lopez-Ruiz P, Masel J, van Nieuwenhuizen O, Rodriguez-Barrionuevo C. Spondylo-metaphyseal dysplasia: Report of 7 cases and essay of classification. In Papadatos CJ, Bartsocas CS (eds): "Skeletal Dysplasias." New York: Alan R. Liss, Inc. 1982:89-101.

Lomas JJP, Boyle AC. Osteochondrodystrophy (Morquio's disease) in three generations. *Lancet* 1959;2:430-432.

Mukamel M, Karmazyn B, de Vries L, Horev G, Shohat M. Brachyolmia and spinal stenosis. *Am J Med Genet* 2003;120A:272-275.

Sewell AC, Wern C, Pontz BF. Brachyolmia: a skeletal dysplasia with an altered mucopolysaccharide excretion. *Clin Genet* 1991;40:312-317.

Shohat M, Lachman R, Gruber HE, Rimoin DL. Brachyolmia: radiographic and genetic evidence of heterogeneity. *Am J Med Genet* 1989;33:209-219.

Toledo SPA, Mauraö PAS, Lamego C, Alves CAR, Dietrich CP, Assis LM, Mattar E. Recessively inherited, late onset spondylar dysplasia and peripheral corneal opacity with anomalies in urinary mucopolysaccharides: a possible error of chondroitin-6-sulfate synthesis. *Am J Med Genet* 1978;2:385-395.