Toledo type brachyolmia

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Abstract

Brachyolmia is a form of spondylodysplasia that has not to the authors' knowledge been described in the UK. It may be a cause of short stature that is currently unrecognised. A case of an 11 year old boy with clinical, radiographic, and eye findings consistent with Toledo type brachyolmia is reported.

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Brachyolmia is a form of spondylodysplasia, resulting in short trunked dwarfism. There is generalised platyspondyly without significant long bone changes. Radiographic findings include narrowing of the intervertebral joint spaces and narrow interpedicular distances, but no generalised metaphyseal, epiphyseal, or diaphyseal changes.¹ One previous report suggests that bone age is normal or advanced in this condition.² Urine shows a qualitative rather than quantitative abnormality of glycosaminoglycans, differentiating the condition from the mucopolysaccharidoses.²⁻⁴ Intelligence is normal. Brachyolmia has been divided into types according to other radiographic changes as well as extra skeletal changes¹ (described later). The Toledo type shares the radiographic findings of the

Hobaek type but in addition peripheral punctate opacities are found in the cornea on slit lamp examination. The inheritance of Toledo type is autosomal recessive.³

Case report

We report the case of an 11 year old boy who presented with anterior knee pain. Clinical and radiological signs were consistent with chondromalacia. However, his appearance was striking in that he had a relatively large head and short trunk (see fig 1). Further assessment, clinical examination, and investigations were therefore undertaken. He had been assessed in a growth clinic at the age of 5 years and his parents given a relatively good prognosis for his final height. His mother's height lay between the 75th and 90th centiles and his father's height lay on the 50th centile. There was no family history of short stature and he had a sister of normal height. He was of normal intelligence.

Examination of his musculoskeletal system showed marked limitations of lateral flexion, no extension, and limited lumbar flexion of his spine. His head circumference was 56 cm (75th-90th centile), weight 43.8 kg (75th-90th centiles), and height 136 cm (3rd-10th centile). His upper to lower body ratio measured to the symphysis pubis was 0.83 (normal for age 0.97). General examination was normal.

Radiography of the spine (see fig 2) showed platyspondyly throughout the length of his spine and reduced intervertebral spaces. There was marked extension of the lateral margins of the vertebrae, with elongation of the vertebral bodies on lateral view. There was no evidence of generalised epiphyseal or metaphyseal abnormality. There was no premature ossification of the costal cartilages. Bone age at a chronological age of 12.5 years was 14 years ± 1 year (Grulich and Pyle).

Ophthalmic examination showed 6/4 acuity in both eyes. Slit lamp examination revealed opacities in the deep stroma of both corneas. Urinary glycosaminoglycans were quantitatively normal.

Analysis of the patient's urine at the University Children's Hospital in Frankfurt showed total mucopolysaccharide (glycosaminoglycan) excretion to be normal, electrophoretic separation of the individual mucopolysaccharide subclasses was also normal, but on digestion with chondroitinase ABC (to degrade chondroitin 4-sulphate, dermatan sulphate, and chondroitin 6-sulphate) an under-sulphated chondroitin sulphate was found.

Discussion

Maroteaux *et al* first used the term brachyolmia for a form of short trunked dwarfism with

Figure 1 Photograph illustrating the relatively short trunk found in brachyolmia.

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Figure 2 Anteroposterior spinal radiograph illustrating the radiographic abnormalities described in the text.

generalised platyspondyly but without significant long bone changes.⁵ It has not to our knowledge been described in the British literature.

Typically birth length is low and short stature becomes progressively more apparent with increasing age. There is shortening of the trunk with low-normal upper-lower body ratio. Where upper-lower body ratio is normal there must also be some metaphyseal shortening. Shortening of the femoral neck is also seen. Shohat *et al* divided brachyolmia into types dependent upon clinical, genetic, and radiographic differences.¹ The Hobaek and Toledo types have the vertebral changes described in our case and are inherited in an autosomal recessive manner. They differ in that peripheral punctate corneal opacities, as seen in our patient, are found only in the Toledo type.³ A third autosomal recessive type is the Maroteaux type where there is rounding of the anterior and posterior vertebral bodies with

less elongation on lateral spinal radiography. A dominant form has also been described with severe vertebral changes. Vitreoretinal dystrophy associated with brachyolmia inherited in an autosomal dominant mode has been reported.⁶ Clinically brachyolmia shares some features with the mucopolysaccharidoses. However, in brachyolmia the urinary glycosaminoglycans are quantitatively normal. The study employing qualitative analysis of glycosaminoglycans undertaken at the University Children's Hospital in Frankfurt showed the presence of undersulphated chondroitin C on enzyme degradation. This is identical to the finding in a patient of theirs previously reported.² This investigation is not routinely available.

Our patient's presentation with chondromalacia patellae may not be of significance as this is a common problem and exacerbated by obesity. Short stature had been a concern of his parents in the past. His present height between the 3rd and 10th centiles is less than would be expected for his midparental height and with his advanced bone age his predicted final height is below the 3rd centile. Advanced bone age has been described in association with brachyolmia.²

We hope that this report will increase awareness of this condition which is probably under reported. Brachyolmia should be considered in short stature when there is relative shortening of the trunk and can be confirmed radiographically with anteroposterior and lateral views of the spine.

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