

Chondrodysplasia punctata mimicking Blount's disease

A case report

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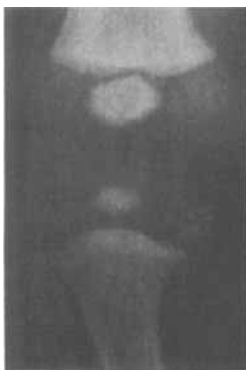
Case report

A 10-month-old Japanese girl with a bowed right leg that had been evident from birth was referred to our department. The girl was the first child of a 28-year-old woman. The birth weight was 2,840 g and the length 45 cm. The family history was negative for bone diseases, short stature, skin, and eye anomalies, except that the mother was 144 cm tall and had bilateral genu valgum. Her skeletal survey, however, showed no abnormality. The parents were not consanguineous. The patient had a normal younger brother.

The patient was 68 cm tall (-1.5 SD of Japanese standard for her age) with a normal facies and body proportions. No cataracts, skin lesions, scoliosis, or contracture were found. There was a varus deformity of the right leg, which was 1.5 cm shorter than the left. The right foot was also shorter than the left by 1 cm. Laboratory studies were normal.

Radiographs at the age of 4 months taken locally showed that the right proximal tibial epiphysis was smaller than the left, and that there was punctate stippling medial to the epiphysis with mild varus angulation at the proximal tibial metaphysis (Figure 1). Stippling was also present in the distal epiphysis of the right tibia and in the right medial cuneiform (Figure 2). At the age of 10 months, the varus deformity had progressed with a radiolucent area in the metaphysis and irregularity of the medial half of the physis in addition to the punctation.

The punctation decreased with age: it had disappeared by the age of 4 years in both the distal tibia and the medial cuneiform. In the proximal tibia, stippling was only faintly visible at the age of 2 years and 3 months, and the radiographic changes were similar to those of Blount's disease: there was beaking of the medial metaphysis with translucency, and the medial side of the epiphyseal plate was widened and irregular (Figure 1).



Aged 4 months.



Aged 10 months.



Aged 2 years and 3 months.



Aged 6 years and 5 months.

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Figure 1. The right proximal tibia.

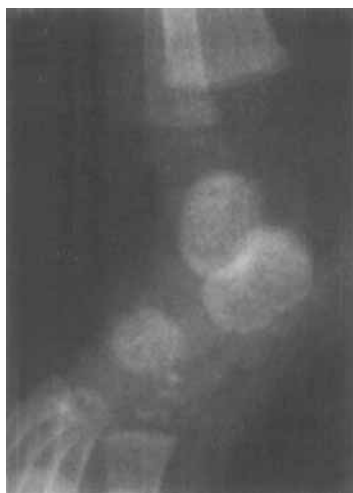


Figure 2. The right foot at the age of 4 months. Stippling of the distal tibial epiphysis and medial cuneiform.

Corrective osteotomy of the right tibia was performed at the age of 2 years and 5 months. There was a loss of correction in the early postoperative period, and 5° of varus deformity remained. However, no progression of the deformity was observed after 3 years of age. Radiographs at the age of 6 years and 5 months revealed only slight beaking and sclerosis of the medial metaphysis (Figure 1).

The leg-length discrepancy increased during the preoperative period (10 mm at the age of 5 months; 18 mm at the age of 2 years and 5 months) and remained constant at 1 cm after the operation.

Discussion

In our case the radiographic appearance of the knee gradually became indistinguishable from that of Blount's disease. To our knowledge, no case of chondrodysplasia punctata mimicking Blount's disease has previously been reported. However, this is partly because most cases of the disorder are fatal, and reports of serial radiographic changes are few. Moreover, punctation tends to subside with age: it disappears usually within the first year and at the latest by 3 or 4 years of age (Mosekilde 1968, Tasker et al. 1970). Therefore, some cases diagnosed as infantile Blount's disease may actually be chondrodysplasia punctata after stippling has disappeared.

Acknowledgement

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References

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- Tasker W G, Mastri A R, Gold A P. Chondrodystrophia calcificans congenita (dysplasia epiphysalis punctata). Recognition of the clinical picture. *Am J Dis Child* 1970; 119 (2): 122-7.