
Abstract The distinctive radiographic features – megaepiphyses, hypoplastic fibulae, ulnar pseudoepiphyses and brachymesophalangy – in a 14-year-old Japanese boy with mental retardation are reported.

Introduction

We report the case of a boy with a unique cluster of congenital anomalies including megaepiphyses, hypoplastic fibulae, ulnar pseudoepiphyses and brachymesophalangia. Association of these anomalies has not previously been described.

Case report

This boy was the second child of healthy, unrelated parents. The mother was 32 years old and the father 31. The family history was unremarkable. He was delivered at 42 weeks’ gestation after an uneventful pregnancy with a birth weight of 3750 g, length, 49.5 cm and head circumference, 35.6 cm. A subdural effusion was drained when he was 4 weeks old. His motor development was retarded; he started to walk at the age of 20 months.

He was referred for investigation at the age of 21 months because of exaggerated out-toeing. Physical examination showed short stature (75.1 cm, 3 P) and low weight (10.2 kg, 10-25 P). An exaggerated lumbar lordosis was noted at the age of 5 years. His joint laxity persisted. At 8 years he appeared obese and minimal hyperlipoproteinaemia was noted. At 13 years of age extensive endocrine and biochemical examinations all gave normal results.

At the age of 5 years 3 months his height was 96.1 cm (3 P) and weight, 17.3 kg (25–50 P). At 14 years 2 months the height was 141.6 cm (3 P) and weight, 49.1 kg (50 P). An exaggerated lumbar lordosis was noted at the age of 5 years. His joint laxity persisted. At 8 years he appeared obese and minimal hyperlipoproteinaemia was noted. At 13 years of age extensive endocrine and biochemical examinations all gave normal results.

The radiographic examination of the patient at the age of 20 months revealed shortening of the metacarpals and the middle phalanges of the hands. The middle phalanges of the second to fourth fingers showed some coning of the epiphyses, and the middle phalanx of the fifth finger was small and rhomboid in shape. The bone age was delayed. The epiphyses, specifically in the proximal humeri and proximal femora, were large. There was distal shortening of the ulnae with large pseudoepiphyses at the proximal ends. There was wedging of the distal tibial epiphyses. The fibulae were hypoplastic with only the distal fibular epiphyses present. There was wide separation of the ischio-pubic synchondrosis (Fig. 1 a–d). In the ensuing years shortening of the metacarpals became much less marked, with the exception of the third metacarpal which was short and slightly dysplastic. Fusion of the proximal ulnar pseudoepiphysis with the shaft was present at the age of 5 years. The femoral necks became short and the trochanter major prominent (Fig. 1 a–g). CT of the head showed prominence of the left lateral ventricle.
Fig. 1a–d The patient at 20 months. a Short middle phalanges with cone-shaped epiphyses, short metacarpals. The distal radial epiphysis is oval. The carpal bone age is below 3 months. b The epiphysis of the humerus is large, with a large pseudoepiphysis at the proximal end of the ulna. c Widely separated ischio-pubic synchondrosis. d Hypoplastic fibulae with small, round distal epiphyses, large, slightly flattened knee epiphyses and wedge-shaped distal tibial epiphyses

Discussion

The combination of megaepiphyses, hypoplastic fibulae, ulnar pseudoepiphyses and brachyphalangia is unique and is unlikely to be confused with any other disorder. Diagnostic difficulties can only arise if single radiographs are reviewed.

Our patient showed epiphyseal dysplasia but his epiphyses were large, not small. The most unusual finding was of the proximal ulnar pseudoepiphyses. Brachymesophalangia with hyperextensibility of interphalangeal joints, hypoplastic fibulae and short third metacarpals were additional findings. The carpal bone age was retarded.

None of the megaepiphyseal dysplasias show ulnar pseudoepiphyses, hypoplastic fibulae and brachymesophalangia [1–6]. Our patient’s mental retardation is probably coincidental, given that it is not uncommon in the general population. The inheritance of the disorder is uncertain. It may be a new mutation or an autosomal, recessive trait.