Dysmorphology report

Unknown syndrome: proportionate short stature, mandibular prognathism, and short femoral necks

P J Morrison, N C Nevin, R W Kendrick

Abstract
We describe a young woman with proportionate short stature, short femoral necks, limitation of wrist flexion, and a prominent mandible.

Clinical history
Prenatal and postnatal history was normal and there was good general health, normal intelligence, and normal sexual development. She was the first child of healthy, unrelated parents. Her mother was aged 31 years and her father 35 years at birth. There was one younger normal male sib and no family history of congenital abnormalities or of genetic disease.

Clinical examination
Aged 14·4 years, there was striking facial dysmorphism with deep set eyes, full nasal bridge, small, normally situated ears and mouth, normal palate and dentition, and marked mandibular prognathism (figure). She had limitation of wrist flexion and finger extension and syndactyly of the left second and third toes. There was proportionate short stature (standing height 139·2 cm) and weight of 38·8 kg (both less than the 3rd centile). Sitting height was 77·8 cm. The feet were small. At 17 years her height was 1·40 m.

Investigations
Skeletal survey showed normal vertebral alignment with normally shaped vertebral bodies. All epiphyses of the long bones are closed and the bone age is 14·5 years. The femoral necks are short but with normal angulation. Skull x-ray shows marked mandibular prognathism and bridging of the sella turcica. High resolution G banding showed a chromosome karyotype 46,XX. Biochemistry profile was normal.

Discussion
The patient has facial dysmorphism with deep set eyes, a prominent, pointed chin, marked prognathism,
and proportionate short stature. The short femoral necks and the limitation of full extension of the elbow joints were suggestive of hypochondroplasia, but there were no other confirming features. Short femoral necks and short stature are features of several skeletal dysplasias, including multiple epiphyseal dysplasia, metaphyseal chondrodysplasia Schmid type, pseudoachondroplasia, and diaphyseal aclasis. These skeletal dysplasias can be excluded on clinical and radiological grounds. A prominent mandible, short stature, and short femoral necks may be features of the trichorhinophalangeal syndrome but our patient has normal hair. An extensive search of published and personal databases was unsuccessful in suggesting a possible diagnosis. The mode of inheritance is unknown, but the condition in our patient could represent a new dominant mutation, although autosomal recessive inheritance cannot be excluded. It is planned to undertake facial reconstructive surgery.

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