

An Interesting Form of Osteochondrodystrophy –A Case Report of a Family

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Abstract

A 12-year-old boy presented with progressive increasing deformity of both knee joints since last 10 years. The radiograph of femur, tibia and phalanges showed different dysplastic changes of epiphysis. It showed a different skeletal dysplastic nature to multiple epiphysal dysplasias. Silfverskiöld described similar types of skeletal dysplasia.

Key words : Multiple epiphysal dysplasia (MED), osteochondrodysplasia, epiphysis.

Introduction:

Multiple epiphysal dysplasia (MED), common type of osteochondrodysplasia, is an uncommon inherited condition resulting in the formation of abnormal epiphyses.

The term chondro-osteodystrophy was first given by Brailsford in 1929 to a disorder of the skeleton which manifested itself during the first three years of life. In the same year Morquio described “a form of familial osseous dystrophy” in four members of a family of five¹. The disorder was manifested as multiple areas of abnormal growth and ossification of the epiphysis. It affected predominantly hips, knees, ankles, and wrists. Although present at birth, symptoms of MED do not develop until years later². The radiologic finding of multiple abnormal epiphysis ossification centres is diagnostic. The affected epiphyses are small, irregular,

mottled and/or fragmented, irregularly mineralised, late ossifying, and usually found in the long bones of the lower limbs. The femur are irregular symmetric, and commonly have early acetabular changes and mild metaphyseal flaring.

Case Report:

A 12 years boy came to our outpatient department with chief complaint of genuvalgum. There was history of similar deformity of his two male siblings. Among his three elder siblings, eldest one died at the age of 4 years due to unknown disease. The second elder sibling was normal and not suffering with similar type of deformity. But the third elder and younger siblings affected with the similar deformity (Fig 1). Parents noticed the progressive deformity in the entire above mentioned child after 2 years when they start walking.

On examination we found large head, base of nose

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Figure 1



Fig 2

flattened, broad and short phalanges, hip flexion deformity, laterally displaced patella, genu valgum with externally rotated tibia, broad and shortened toe, 4th toe overriding 3rd and 5th toes and arm was comparatively shorter than forearm (Fig 2).

On biochemical analysis : Blood group B +ve, TC-8150/cmm, DC-W-54, L-40, E-6, Hb%-10.4, ESR- 26/hour, BT-0'46", CT-5'12", calcium-10.2mg/dl, phosphorus-4.4mg/dl, alkaline phosphatase-192IU/l, T4-105ng/ml, TSH-0.7 μ IU/ml, urine-RE/ME -normal.

On radiological analysis there were dysplastic changes seen in epiphysis of both ends of femur, both ends of tibia, both wrist joints. All phalanges were short (Figs 3-5). There was fish mouth appearance of spine seen in lateral view (Fig 6). In orthoscanogram there was multiplanar deformity seen in both femur and tibia (Fig 7).

Discussion:

In 1925 and 1926 Silfverskiöld³ described a total of four patients among five had different skeletal dysplasia. As the disease pictures vary greatly, it was impossible to give any useable definition. Silfverskiöld's patient was an eleven-year-old boy in whom the symptoms included disproportionate dwarfism with short legs, large head, flattened nose, broad chest and large trochanters. Hans Fredrik Helweg-Larsen (1917-1969) and Mørch later reported the syndrome in a family.

Apart from the description of Silfverskiöld and Hans Fredrik Helweg-Larsen, we noticed fourth toe overriding 3rd and 5th toes (Fig 8), Arm was shorter compared to forearm, laterally displaced patella and 4th metatarsal and phalanx were shorter than others in our cases.

Murphy *et al* (1973)⁴ observed that roentgenograms showed well recognised typical features of the disease:



Fig 3



Fig 4



Fig 5



Fig 6



Fig 7

short metacarpals, widening of the distal radial metaphysis, the flattened humeral head with shallow glenoid fossa, and oblique acetabulum with flattened femoral head. In addition, a frequent feature was a slanting of the ankle joint mortise found in ten of twelve patients.

Treble *et al* (1990)⁵ reported that the development of ossific nucleus of the femoral head was abnormal. The nucleus was small, misshapened and showed abnormal pattern of ossification. In our case the acetabular components of the hip joint were comparatively normal and the femoral head misshapened and flattened symmetrically.

Conclusion:

These cases were presented to us as bilateral symmetrical involvement of all the epiphysis along with above findings. These cases were unusual form of osteochondrodystrophy with skeletal defects with hereditary involvement.

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Fig 8

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