**INTRODUCTION**

The normal angle between the neck and shaft of the femur varies between 135 and 145 degrees in children. A decrease in this angle below 110 degrees constitutes coxa vara. Infantile (congenital) coxa vara may be part of some skeletal dysplasias, such as achondroplasia, multiple epiphyseal dysplasia, spondyloepiphyseal dysplasia, and cleidocranial dysplasia, and occurs especially in those dysplasias where there is a delay in the ossification of the capital femoral epiphysis. Congenital coxa vara is commonly associated with abnormalities of development of the femur such as congenital short femur, congenital bowed femur and proximal focal femoral deficiency.1-4 Metaphyseal chondrodysplasias is a heterogenous group of intrinsic dysplasias characterized by radiological changes in the metaphysis (irregular patchy sclerosis of the metaphyses). These dysplasias present in infancy or in childhood, when the patient, usually dwarfed, may be proportionate, so that some forms may be confused with rickets or other lesions. Radiological abnormalities mainly affect the metaphysis of the shortened limb bones, bilaterally and symmetrically.5,6

We describe an unusual form of metaphyseal dysostosis not consistent with any recognized form. Interestingly, there was father-to-son transmission, raises the possibility of autosomal dominant pattern of inheritance.

**CASE REPORT**

A 6-year-old boy was referred to the Paediatric Orthopaedic Department because of significant limp on the left side. He was a product of uncomplicated gestation born full term. At birth, his length and weight was around the 10th percentile and the occipito-frontal-circumference was around the 25th percentile.

The mother was a 29-year-old primipara married to a 31-year-old-unrelated man. No serious illnesses were noted in child’s first year of life, but his subsequent course of motor development were delayed. He began to walk at the age of 15 months albeit with difficulties. Despite parents concerns no dramatic measures were taken. In his early childhood, gait abnormalities predominated his motor activity followed by development of a progressive limp on the left side. At the time of presentation, he had left proximal varus deformity with an angle of 80°. Limb length inequality was about -2.5 cm. Measurements used to define the degree of coxa vara were the head-shaft angle and the neck shaft angle respectively. The angle between the neck and the shaft of the femur on the affected side was 80°.

Child’s physical examination at the age of 6 years revealed height of 105 cm (-3 SD) i.e. proportionate short stature and weight of 17 kg (-2 SD). There were no particular cranio-facial dysmorphic features to suggest a dysmorphic clinical entity. Musculoskeletal examination showed a positive Trendelenburg sign, abduction limited to 30°, and internal rotation limited to 10° were present on the left hip. The right hip could be abducted to 60° and internally rotated to 35°. Neither ligamentous hyperlaxity nor stiffness was noted. Upper limbs showed no abnormalities (no hand/limb shortenings). Discrete spinal stiffness was encountered, but no associated spine maldevelopment was notable.

Neurological examination was normal. Hearing, vision and intelligence were normal. Examination of hair, teeth, nails and skin were normal. Laboratory studies showed normal white and red blood cells normal platelet counts, normal serum calcium, phosphorus, and alkaline phosphatase levels. Urine amino-acids and mucopolysaccharides were also normal, and he had a normal karyotype. Further blood analysis showed normal serum total protein, and albumin-globulin ratio was also normal.

On the basis of skeletal survey, there was unilateral coxa vara on the left side, severe shortness of the femoral necks associated with vertical radiolucent and a triangular bone fragment in the inferior / medial aspect of
the left femoral neck associated. The angle between the neck and the shaft of the femur was 80°, the femoral head displaced further distally relative to the shaft, ultimately reaching the level of the lesser trochanter (Figure 1). Significant irregular patchy sclerosis and medial corner fracture was evident in the inferior distal femoral metaphysis and the proximal tibial metaphysis as well associated with metaphyseal striations (Figure 2). Anteroposterior foot radiograph in which the metatarsals and the phalanges showed variable degrees of metaphyseal changes (Figure 3). We intervened by performing proximal femoral intertrochanteric valgus osteotomy in accordance with Pauwels technique. The caput-collum-diaphyseal (CCD) angle was corrected to normal (130°). The osteotomy was fixed using an AO-plate to enhance the bone synthesis process. In addition to achieving a stable osteosynthesis, 3 Kirschner wires were inserted. Simultaneously, an epiphysiodesis of the greater trochanter epiphysis was done to avoid overgrowth (Figure 4).

Family history revealed that the father of the proband had a similar clinical history to that of his son, typical phenotype and short stature (-3 SD). Unfortunately, no previous radiographs were found in the archive. Surgery at the institution was carried out to repair the defect of a previous implant. The latter was performed in a different orthopaedic institute. At time of admission in our hospital, the implant became friable and loose, and therefore, was removed including plates and cerclage material. After an extensive synovectomy a revision cup (Allofit) with screw-fixation and a new stamp were inserted. It was possible to restore the limb length discrepancy and the abductor lever arm. However, due to the many previous operations, a weakness of the abductor muscle had to be accepted (Figure 5).
Congenital coxa vara is a developmental abnormality characterized by a primary cartilaginous defect in the femoral neck with an abnormal decrease in the femoral neck-shaft angle, shortening of the femoral neck, relative overgrowth of the greater trochanter, and shortening of the affected lower limb. Typically the deformity either is not present at birth or is sufficiently subtle as not recognized at that time. Affected patients almost invariably present after walking age, and sometimes as late as adolescence, with a limp Trendelenburg or short-leg gait and in unilateral cases, relatively mild limb length inequality. Acquired forms of coxa vara might be a complication of transcervical injury (fractures of the neck of the femur). The greatest incidence is in children aged between 11-12 years, and three times as many boys are affected as girls. Transcervical fractures is, however, a different orthopaedic entity and have been considered as the most common type of femoral neck fractures causing a shaft angle of about 120 degrees is commonly encountered in patients treated by closed reduction and abduction spica plaster cast. Such coxa vara is likely to persist or even worsen for a long period after seemingly adequate union, particularly in cervico-trochanteric fractures, and leads to a poor result with shortening, an abductor lurch and ultimately, degenerative changes in the hip. Thus, sub-trochanteric valgus osteotomy needs to be made.

Congenital coxa vara is a frequent finding in skeletally dysplastic patients with known genetic causes, such as metaphyseal dysostosis, Jansen metaphyseal dysplasia, spondylometaphyseal dysplasia, especially Kozlowski type, all of which are autosomal dominant disorders. There are reports of the condition in families.

Oh et al. studied 46 patients with coxa vara. Spondyloepiphysyeal dysplasia congenita or spondyloepimeta-physyeal dysplasias were the forms of osteochondrodysplasias reported in connection with congenita coxa vara. They concluded that the lack of epiphyseal ossification was the most challenging element.

The exact cause of congenital coxa vara (CCV) remains unknown. Many hypotheses have been proposed, including: the mechanical intrauterine stresses affecting hip development; avascular necrosis involving selected areas of the proximal femoral physis/thead and neck; and metabolic abnormalities causing deficient production of, or a delay in the normal ossification process of the proximal end of the femur.

Pylkkanen proposed the most widely accepted theory regarding the cause of CCV. He postulated that the proximal femoral deformity is the result of a primary ossification defect in the inferior femoral neck, on which physiologic shearing stresses (applied during weight bearing) cause fatigue of the local dystrophic bone, resulting in progressive varus deformity.

Chung and Riser, described the postmortem gross, microscopic, and vascular findings of the hip in a 5-year-old child who died 2 years after valgus osteotomy for developmental coxa vara. They noted that the acetabular volume and femoral head were smaller, the affected side than on the normal contralateral side. Both the number of blood vessels on the metaphyseal side of the physis and the number of medial ascending cervical arteries were decreased. The bony trabecular network providing support of the medial neck was absent in the epiphysis and metaphysis of both hips in this report, a difference they found striking compared with specimens of normal hips from like-aged children.

It is to be stressed that classifying bone dysplasias in terms of impediments to bone modelling is still far from daily practice and still difficult to classify systemic bone diseases according to pathogenesis.

REFERENCES


