

Schmid Type Metaphyseal Chondrodysplasia

Schmid Type is the commonest Metaphyseal chondrodysplasia. The incidence is about 3 to 6 cases per million of population. Though genetic studies have confirmed 'Autosomal dominant mode of inheritance', many cases are sporadic. Bone maturation is always normal. The clinical pattern varies widely(1).

A 13-year-old high school going female child, born of non consanguineous marriage was brought for short stature. It was a full term normal vaginal delivery. Short stature and waddling gait was obvious by 2years. She was 'Average' at school. At the age of 13years her height was 118 cm (US:LS :: 1.11:1) and weight was 19 Kg. Legs showed lateral bowing with knees wide apart. The wrists and ankles appeared thickened. No dysmorphic facial features were noted.

Her vision and hearing were both within normal limits. She was investigated with the clinical impression of Schmid Type Metaphyseal chondrodysplasia. Radiology showed normal skeletal maturation. Limb Epiphyses were normal. As seen in the figure (Fig. 1) lower femoral metaphyses appeared



Fig. 1. Schmid type metaphyseal chondrodysplasia.

expanded, irregular and cupped. Joint spaces were normal.

Most authorities agree that the Schmid type is a milder form of Jansen type Metaphyseal chondrodysplasia. Significant radiologic features include an enlarged capital femoral epiphysis in early childhood, coxa vara, greater involvement of the distal femoral metaphysis than the proximal, anterior rib changes and a normal spine. Chondroosseous morphology is not specific. Radiological signs in the metaphyses disappear after epiphyseal fusion. Presentation in nonfamilial cases is no earlier than the second year of life(2). Schmid metaphyseal chondrodysplasia is due to heterogeneous mutations of the gene encoding type X collagen. Radiologic changes are confined to the metaphyses(3).

Histologically there is disorganization of cartilage in the growth plate and retardation of endochondral bone formation(4). Severe Coxa Vara may need surgical correction.

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