Abstract  Multiple epiphyseal dysplasia (MED) is a relatively uncommon inherited disorder of epiphyseal maturation. Affected individuals may have a degree of short-limbed dwarfism, short stubby digits, and stiff or painful joints. We report two families of MED and emphasize the variations of joint involvement. Ten out of 34 members in family A and 13 out of 39 members in family B were suspected of having MED by questionnaire. Radiological examination was done for 3 out of the 10 members in family A and 6 out of the 13 members in family B. In both families, the epiphyseal disturbances in the skeleton were bilaterally symmetric and involved several joints. Apparent dwarfism, short stubby digits and spinal involvement were not observed. The degree and the pattern of affected joints were different in the two families and even among members of the same family. In family A, the knee joint was commonly affected, followed by the ankle joint. The deformity of the joints was mild and caused only slight disability. No apparent hip lesion was present. In family B, the hip joint was predominantly affected, followed by the knee and ankle joints, and the deformity was severer than that in family A. These observations suggest that MED is a group of heterogeneous disorders.

Introduction  Multiple epiphyseal dysplasia (MED) is a genetically determined disorder characterized by dwarfism, stubby digits, and abnormalities in maturing epiphyses. The disorder was always bilateral. It was first described by Fairbank in 1947, who elaborated its clinical and radiographic characteristics [4]. This disorder has been associated with the onset of osteoarthritis (OA) during early adolescence, usually in the hip joints [13].

Fig. 1  Family A pedigree. Ten out of 34 members were suspected of having multiple epiphyseal dysplasia (MED) by questionnaire and/or radiological examination.

Fig. 2  Family B pedigree. Thirteen out of 39 members in family B were suspected of having MED by questionnaire/or radiological examination. All the patients had both hip and knee problems.
The purpose of the report is to describe two families with MED and emphasize the variations of the involvement pattern of this developmental abnormality.

Case reports

Ten out of 34 members in family A (Fig. 1) and 13 out of 39 members in family B (Fig. 2) were suspected of having MED by questionnaire according to the following criteria: short stature, positive family history, and/or early onset of pain of bilateral joints. In family A, 10 members complained of knee pain, and two members had hip pain. In family B, all the patients had both hip and knee pain. Physical and radiological examinations were done for 3 out of the 10 members in family A and 6 out of the 13 members in Family B.

In both families, the epiphyseal disturbances in the skeleton were bilaterally symmetric and involved several joints. Apparent dwarfism, short digits, and spine involvement were not observed. In the two children examined in family A, stiffness in the hips was noted at times, but they attended school and participated normally in games. As for their father, he had had the same complaints in his youth but did not have any difficulties in working or recreational activities when older. In family A, the knee joint was commonly affected (Fig. 3), as well as the ankle joint (Fig. 4). No apparent hip lesion was present. The deformity of the joints was mild and caused only slight disability. In family B, all the patients examined limped and suffered from severe coxalgia since the age of 5 or 6 years, and one of them underwent total hip replacement surgery at the age of 48 years. The hip joint was predominantly affected (Fig. 5), followed by the knee (Fig. 6) and ankle joints (Fig. 7), and the degree of deformity was severer than that in family A.

Discussion

Fairbank first delineated this entity, giving it the name dysplasia epiphysialis multiplex [4]. There is, however, more than one type; that described by Fairbank is the se-