Current concepts

The orthopaedic aspects of multiple epiphyseal dysplasia

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Summary. Five cases of multiple epiphyseal dysplasia (MED) were treated from 1985–1996 at the Orthopaedics and Trauma Department of SSK Izmir Educational Hospital. Four patients were female and one was male. The pedigrees of the first two female patients had the same features of inter-related marriages. The patients have been followed up for 5.5–11 years (average of 7.5 years). Surgical operations were mostly required in the lower limbs. Problems in the hips required adductor myotomy, the Soutter procedure, total hip replacement, and pertrochanteric extension osteotomy. Management of the knees required supracondylar shortening and extension osteotomy of the femur, high tibial extension osteotomy, debridement of the knee joint with removal of osteophytes, osteotomy of the patellar thickness of the knee flexors and posterior capsulotomy. Interphalangeal arthrodesis for hammer toes, extension osteotomy of the head of the first metatarsals, and Kellers operation were carried out in the foot. In the upper limb decompression and anterior transposition of the ulnar nerve, debridement of the elbow joint, extension and valgus osteotomy of the distal radius, and extension osteotomy of the head of the first metacarpal were required.


Introduction

Multiple epiphyseal dysplasia (MED) was so named by Fairbank in 1935 [4, 6, 9, 16–18] and is an autosomal dominant hereditary disorder. MED has lately been divided in two types. In Type I, there is a mutation in the gene coding for cartilage oligomeric matrix protein, as in pseudoachondroplasia. In Type II, there is a mutation in the gene coding for the alpha 2 polypeptide chain of Type IX collagen. Synonyms are dysostosis epiphysealis multiplex, dysplasia polyepiphysaire, hereditary enchondral dysostosis, Fairbank disease, and Ribbing-Müller disease [3, 4, 6, 9, 16–18].

The disease can be diagnosed in early childhood and affects almost all epiphyses. It can cause severe flexion contractures in most joints due to early arthro-
sis. As a result of spinal involvement, spondylarthrosis and moderate scoliosis are seen [16, 17].

Patients complain of a limping during early childhood, a duckling gait, and limb deformities due to degenerative arthritis and contractures [13]. Joint movement may be limited at adolescence and puberty, due to the development of degenerative arthritis, contractures and pain in the hips, knees, and ankles. The muscles are normal and intellectual function is not disturbed. There are no notable abnormalities in the blood or urine. Radiographs demonstrate hypoplastic and irregular epiphyses, but there is no sclerosis. The fingers and toes are short and thick. The tarsal and carpal bones may be hypoplastic. The hip, knee, and ankle joints are most seriously affected. Involvement of the shoulder, elbow, and wrist is moderate and causes relatively few complaints. Tibiotalar inclination is seen in about half of the cases and carries diagnostic importance. In adolescents the vertebral changes may resemble Scheuermann’s disease; later, spondyloarthrosis and, in some cases, a mild degree of scoliosis may develop. The differential diagnosis includes the mucopolysaccharidoses, Perthes disease, osteopetrosis, osteopoikilosis, dysplasia epiphysealis punctata, cretinism, spondyloepiphyseal dysplasia, and hypophyseal dwarfism [1–4, 6, 12, 16, 20, 21].

Conservative treatment may include control of body weight, and underwater vertical retraction with a load has been recommended [4, 18]. Tachdjian [16] has recommended epiphysiodesis for leg length discrepancy, and Volkov [18] has reviewed the surgical management.

Patients and methods

We have treated five patients with MED between 1985 and 1996 at the Orthopaedics and Trauma Department of SSK Izmir Educational Hospital. Four patients were female and one was male. There were four women aged 48, 23, 22, and 21 when first seen, and one man aged 28. The major complaints were of pain in the hips, knees, and ankles, associated with joint stiffness.

Flexion deformities were seen in the hips and knees, which also demonstrated valgus deformity. Cubitus valgus was seen in all the patients. All had an increased lumbar lordosis. Radiological investigation showed severe arthrosis in all joints, especially the hips, knees, and ankles. All laboratory tests were normal.

The grandfather, uncle, and aunt of the first female patient, whose treatment began at the age of 23, had the same disorder (Figs. 1 and 2). The brother of the second, whose treatment began at the age of 48, also had MED, but it was not present in the family history of the other three patients. When first seen two of our patients could not walk. Two others walked with knee and hip in 95° of flexion and the hips in 10° of adduction. Their average standing height was less than 125 cm.

The following operations were carried out in the hips:

(I) Adductor myotomy (8 operations in 4 patients)
(II) Soutter operation (6 operations in 3 patients)
(III) Total hip arthroplasty (6 operations in 3 patients)
(IV) Pertrochanteric extension osteotomy of the femur (4 operations in 2 patients)

In the knees (Fig. 3) we undertook:

(I) Supracondylar shortening and extension osteotomy of the femur (2 operations in 1 patient)
(II) High tibial extension osteotomy (10 operations in 5 patients)
(III) Debridement of the knee joint and removal of osteophytes (8 operations in 4 patients)
(IV) Patellar osteotomy (2 operations in 1 patient)
(V) Lengthening of the knee flexors and posterior capsulotomy (2 operations in 1 patient)

The toes (Fig. 4) required:

(I) Interphalangeal arthrodesis for hammer toes (16 operations in 1 patient)
(II) Extension osteotomy of the head of the first metatarsal (2 operations in 1 patient)
(III) Kellers operation (2 operations in 1 patient)