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 İzmir 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, ogleotomy of the patellar lengthening 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.

Résumé. Cinq cas de dysplasie multi-épiphysaire ont été traités pendant la période 1985–1996 dans le département d'Orthopédie et Traumatologie de l'Hôpital Universitaire SSK İzmir. Il y avait 5 femmes et un homme. L'arbre généalogique des deux premiers patients montre des particularités tel que le mariage entre proches parents. Les patients ont été observés pendant un délai de 5,5 à 11 années (7,5 en moyenne). La plupart des interventions chirurgicales

ont été faites sur les membres inférieurs. Au niveau de la hanche ont été pratiquées: la ténatomie des adducteurs, l'opération de Soutter, arthroplastie totale, et l'ostéotomie d'extension pertrochantérienne. Au niveau des genoux, ont été faits: ostéotomie supracondylienne de récurvatum et raccourcissement, ostéotomie d'extension tibiale, débridement articulaire avec ablation des ostéophytes, amincissement rotulien, allongement des fléchisseurs du genou et capsulotomie postérieure. Au niveau des pieds, ont été réalisées l'arthrodèse inter-phalangienne pour orteils en marteau, l'ostéotomie d'extension de la tête du premier métatarsien et l'opération de Keller. Au niveau du membre supérieur ont été réalisés: transposition antérieure du nerf cubital, débridement du coude, ostéotomie de valgus extension du radius distal et ostéotomie d'extension de la tête du premier métacarpien.

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, dysplasie polyépiphysaire, 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-

Fig. 1. Patient Ş.G.: extension osteotomy at the trochanteric area for flexion contracture of the hip

Fig. 2. Patient Ş.G.: spondyloarthrotic changes at the lumbar area

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 İzmir Edu-

cational 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)

Fig. 6. **A** Patient Ş.Ö.: Fracture of the acetabular cup by an accident 10 years after the hip replacement. **B** revision arthroplasty of the acetabular cup by using an acetabular reinforcement ring. The femoral component was not changed by a new one

One patient (Fig. 5) required decompression and anterior transposition of the ulnar nerve, and another debridement of the elbow joint. One patient had an extension and valgus osteotomy of the distal radius and another extension osteotomy of the head of the first metacarpal.

The patients were subsequently followed-up for between 5.5 and 11 years (average 7.5 years).

In one patient who required a total hip replacement, a custom made miniature straight stemmed femoral component was needed. We carried out pertrochanteric osteotomy in all the hip replacements, although in patients who do not have MED it has been suggested that pertrochanteric osteotomy should not be carried out in patients under the age of 40 [14]. In another patient the architecture of the hip was such that replacement arthroplasty was not possible. In this patient valgus and extension osteotomies of the hips were carried out.

High tibial extension osteotomy was undertaken as described by Lök et al. [10, 11] and Çelebi et al. [5]. The osteotomies were fixed by staples, but in another patient an external fixator was used.

On several occasions difficulty in endotracheal intubation was encountered due to cervical arthrosis but the problems were overcome by an experienced anaesthetist.

Results

A residual flexion deformity of 10–15° remained in 6 hips in 3 patients with total hip arthroplasty. Ectopic ossification was observed about the prosthesis in the oldest patient, and this restricted movement. Patients

with prostheses were pain free and were able to walk upright. A flexion contracture of 5–15° remained in the 4 knees of 2 patients, but the range of movement had been increased by 10–15°. Despite major debridement in the knee joint no effusion or further complaints were encountered. One hip prosthesis dislocated while the patient was being brought back from the operating room, but this was reduced and remained stable. One other patient with a total hip arthroplasty had a fracture of the cup after an accident 10 years after operation. The femoral part of the prosthesis remained stable. The fractured component was changed and an acetabular ring used for reinforcement (Fig. 6).

Discussion

Multiple epiphyseal dysplasia is a rare disorder of autosomal dominant inheritance [3, 4, 6, 9, 16–18], and may result in short stature [3, 4, 9, 16–18]. Four of our five patients were less than 125 cm tall, and the 5th was 152 cm in height. MED is not accompanied by mental retardation, and the levels of intelligence of our patients were normal.

Cotta advised splints, corsets, traction and exercises for maintenance of posture in childhood and early adult life [4]. Tachdjian emphasised the need to differentiate this disorder from Perthes' disease and advised against the use of orthopaedic devices unnecessarily [16]. Volkov proposed physiotherapy and mud baths [18]. For surgical management, release of the contractures and correction of the axial deviation [4], epiphysiodesis for leg length discrepancy [16], and supracondylar femoral osteotomy for varus and valgus deformities in the knee [15] are recommended. For established osteoarthritis, Tachdjian proposed primarily symptomatic treatment and then, if this failed, appropriate surgical intervention [16].

Canale pointed out the importance of maintaining the range of movement in the hips in order to be able to carry out daily activities and advised that arthrodesis of the hip should not be undertaken in these patients [3]. Woolson and Harris used miniature components in one patient with MED requiring total hip arthroplasty [19].

Ingram [8] described involvement of the elbow joint in 50 patients with MED. On clinical grounds, he identified three groups, normal, asymptomatic with flexion contractures and symptomatic with flexion contractures. Our patients fell into all three groups.

Herring [7] reported rapidly developing scoliosis in one patient. A mild degree of scoliosis is common in MED and was present in two of our five patients.

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