We examined clinically and radiologically the knees of 46 patients (27 females and 19 males) with diastrophic dysplasia. The age of the patients varied from newborn to 38 years. A total of 18 patients was followed during their growth until adolescence. The knees of two legally aborted fetuses appeared on examination to be macroscopically normal and congruous. Excessive valgus deformity of the tibiofemoral weight-bearing angle with a mean of 14° was noted in infancy. Most of the patients had marked instability of the knees. The range of movement of the knee began to decrease before the age of five years. There were signs of early degeneration and deformation of the bony epiphyses before the age of six years. The patellofemoral joint was abnormal from an early age. A marked patella infera, often associated with a lateral position of the patella with bony fragmentation, was noted. The knee in diastrophic dysplasia is basically unstable, showing early deformation of the subchondral bone and degeneration of the joint.

Among other typical features, such as disproportionate short stature, dysmorphic facies and multiple joint contractures, the knees show pathological changes in infancy.

Delay in epiphyseal maturation and distortion of the femoral epiphyses with degeneration of the joint have been described in earlier reports. In a previous study we reported the ultrasonographic findings of the patellofemoral joint in which it was shown to be laterally rotated with marked patella infera.

Because of the rarity of this disease, detailed studies of the development of the joints are lacking. The clinical and radiological outcome is largely based on studies with a very early degeneration of the joints is a cause of major disability in patients with severe osteochondrodysplasias. Although rare in other countries, diastrophic dysplasia (DD) is the most common skeletal dysplasia in Finland. The development of deformation of the hip with progressive degeneration in DD has been described recently.

Fig. 1
Diagram of the radiological measurements modified from the method of Schlesinger et al (FM, femoral metaphyseal width; FH, femoral epiphyseal height; FE, femoral epiphyseal width; TH, tibial epiphyseal height; TE, tibial epiphyseal width; TM, tibial metaphyseal width).
limited number of patients. We have observed the development of the knee from birth to adolescence.

Patients and Methods

We recorded the clinical and radiological findings in 46 patients, 19 males and 27 females. Their age varied from newborn to 38 years. Longitudinal follow-up of at least eight years was accomplished in 19 patients. Assessment was made before any knee was operated on. Two legally aborted fetuses in which the diagnosis of DD had been confirmed were dissected and their knees photographed. The presence or absence of pain and the range of movement (ROM) in flexion and extension of the knee were noted. The weight-bearing angle of the tibiofemoral joint was measured using a goniometer. Children under the age of 1.5 years were examined in the supine position. The Lachman sign, the anterior drawer sign and mediolateral joint laxity were evaluated. Plain radiographs were taken in anteroposterior and lateral projections with a fixed film focus distance. Radiological measurements were made according to Schlesinger et al.\textsuperscript{11} and are shown in Figure 1. Normal values were measured in healthy age-matched children, 35 boys and 29 girls (see Fig. 6).

Statistical analysis. Graphic displays were used to show the development of the radiological observations as a function of age for the patients with DD and for normal subjects. Scatter plots were produced by Trellis graphics.\textsuperscript{12} A non-parametric, locally weighted regression smoother was applied to describe the development of the variables over the age range from birth up to 15 years.\textsuperscript{13} Alternatively, we applied a regression spline which draws a continuous curve to the data by piecing together polynomials fit to different age categories of the data. The algorithm also yields approximate 95% age-specific confidence intervals.

Results

Nineteen patients were followed for at least eight years. Serial radiographs were taken from 18 patients during growth. The mean follow-up was 11 years.

Clinical findings. The knees of the fetuses showed no notable deformity and the joint compartments were congruous (Fig. 2). During follow-up, pain in the lower limb was confirmed on at least one occasion in 13 out of 41 patients but was not assessed in five. Accurate evaluation of pain was difficult because of multiple joint problems. The range of movement of the knee started to diminish before the age of five years. There was considerable variation especially with regard to loss of extension in the younger age group (Figs 3 and 4). The weight-bearing angle showed a valgus deformity in infancy, in contrast to...
Decrease in the valgus angle was noted during follow-up (Fig. 5). The valgus deformity became more pronounced with simultaneous rotatory subluxation of the tibiofemoral joint in forced passive extension or hyperextension. Instability of the tibiofemoral joint was evident from early infancy. The Lachman sign was positive in 89% of right knees and in 95% of left, while the anterior drawer test was
markedly positive in 85% and 91%, respectively. Valgus instability was noted in 39% of right and 53% of left knees, and varus instability in 38% and 42%, respectively. Spontaneous subluxation of the tibiofemoral joint, during flexion extension, was evident in 60% of the knees. Mediolateral instability decreased after skeletal maturity.

**Radiological findings.** The normal distribution of the radiological features in healthy children according to age are shown in Figure 6. The values in DD are shown in Figure 7. The femoral epiphyseal height-to-width ratio (FEHW) in DD showed slightly lower values than that in the control group. The femoral epiphyseal height to metaphyseal width ratio (FEHMW) of the patients with DD displayed less increase with age than that in the control group indicating the relative flattening of the bony epiphysis of the distal femur. The tibial epiphyseal height-to-width ratio (TEHW) did not differ between the patients and the control group, although lower values were noted in DD in infancy. The development of the tibial epiphyseal height to metaphyseal width ratio (TEHMW) in DD was clearly different from that of the controls. The increase of the TEHMW with age was less in DD, although at or near skeletal maturity the values were quite similar.

We were unable to observe the radiological appearance of the bony epiphysis under the age of six months in the distal femur in six patients and in the proximal tibia in 11.

Degenerative changes such as joint incongruity, unevenness of the subchondral bone and flattening of the bony epiphysis were noted from as early as three years and increased with age (Fig. 8). In many cases, physeal intrusions into the metaphysis were seen. In some knees, subchondral calcification of the distal femur appeared (Fig. 9). The shape of the proximal tibia was better preserved than that of the distal femur, although sloping of the tibial plateau was often seen (Fig. 8c). The proximal fibula was hypoplastic and short in relation to the proximal tibia in almost all the patients.

The time of radiological appearance of the bony epiphysis of the patella was delayed in DD compared with the normal children. In the four-to-six-year-old age group, the patella was not visible in the radiographs of 23 out of 36 patients. In four patients the patella was still not visible radiologically between eight and ten years. Patella infera was present in every patient, and in many cases the patella was at the level of or even below the tibiofemoral joint.

Fragmentation of the bony patella was also a common finding. It was seen in most knees between the age of four
and six years. After skeletal maturity, however, the patella appeared solid, although deformed, with evidence of early arthritis (Fig. 8c).

Discussion

The clinical manifestations of DD have been described by Amuso, Kopits, Ryöppy et al, Poussa et al and Peltonen et al. According to Kopits, Wilson et al and Hollister and Lachman flexion contracture with an excessive valgus of the knee is commonly seen. Most of the earlier studies are based on small clinical series and case reports. We found a gross anteroposterior instability in over 85% of knees and mediolateral instability in 40%. The high incidence (60%) of spontaneous tibiofemoral subluxation suggests that there is probably a congenital defect of the knee ligaments, and the disease does not simply affect the articular cartilage. Nevertheless, lack of extension was not...
Radiograph showing subchondral calcification in the distal femur of a 3.5-year-old child with DD.

A common finding. There was marked variation in the flexion contracture, especially in infancy. A decrease in ROM was evident towards skeletal maturity, mainly due to secondary deformation of the joint and, probably, to the extreme inferior position of the patella. The hips and knees behave differently in DD. Rigid flexion contracture develops early in the hip.\(^2,3\) The stability of the tibiofemoral joint is highly dependent on the intra-articular ligaments which may, in DD, have a different phenotype compared with the extra-articular structures which are responsible for the stability of the hip.

An excessive valgus position on weight-bearing was noted in childhood. This may be partly due to overgrowth of the medial femoral condyle and lateral rotation of the patellofemoral complex\(^1,10\) (Fig. 8). In our study, pain was recorded in one-third of the patients. This is probably due to the degenerative changes.

Schlesinger et al\(^11\) have described the radiological parameters for the distal femoral epiphysis in bone dysplasias. In our study the values for the distal femur and the FEHMW showed less increase with age than those of the control group, but the flattening of the epiphysis was as described in other skeletal dysplasias.\(^11\) The development of the proximal tibial epiphysis followed a similar pattern, although the deformation and degeneration were less pronounced than in the distal femur. Physeal irregularity and metaphyseal intrusions were also more severe in the distal femur, which may be due to the different anatomy of these growth areas.

Hypoplasia of the fibula in DD has been reported by Vazquez and Lee,\(^8\) Horton et al\(^21\) and Walker et al.\(^1\) This was a common finding in our study. The mechanism of fibular hypoplasia in DD is unknown, although it has been described in other inherited skeletal disorders.\(^22\) The delay in the radiological appearance of the patellar epiphysis and the extreme patella infera were typical findings. The ultrasonographic findings of the patellofemoral joint have been reported in our earlier study,\(^10\) and these may be associated with the progressive sloping of the tibial plateau with increasing age.


The final outcome is early arthritis in all the compartments of the joint with gross deformity of the knee.

This study has been supported by grants from the Ulla Hjelt Fund of the Foundation for Paediatric Research and The Sivija Kosti Fund.

The authors want to thank Dr Ilkka Kaitila for his advice in clinical genetics and Senior Researcher Markku Nurminen for statistical expertise.

No benefits in any form have been received or will be received from a commercial party related directly or indirectly to the subject of this article.

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