FOOT DEFORMITIES IN DIASTROPHIC DYSPLASIA

AN ANALYSIS OF 102 PATIENTS

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The exceptionally high prevalence of diastrophic dysplasia in Finland has enabled us to analyse the foot deformities of 102 patients at their first orthopaedic evaluation and classify 204 feet into five categories.

The most common finding (43%) was a foot with tarsal valgus deformity and metatarsus adductus; 37% showed either equinovarus adductus (29%) or equinus (8%) deformities. At the first examination 13% showed metatarsus adductus deformity alone, and 7% were clinically normal.

The expression 'club foot', generally used for the foot deformity in diastrophic dysplasia is a misnomer. There is a wide spectrum of deformities, some of them specific for the condition.

The Greek-derived word diastrophic, meaning twisted, tortuous or crooked was first used by Lamy and Maroteaux (1960) for a type of dwarfism which is now generally classified as an autosomal recessive hereditary skeletal dysplasia. The biochemical defect in diastrophic dysplasia (DD) is unknown, but the gene mutation has recently been mapped to chromosome 5q (Hästbacka et al 1990).

The diagnosis is usually made at birth, from the disproportionate, short-limbed, dwarfism (Fig. 1). Dysmorphic ear lobes and cleft palate are common, and patients have generalised limitation of joint movement and different kinds of deformities of the skeleton. The lower limbs are invariably described as showing clubfoot deformity (Lamy and Maroteaux 1960; Amuso 1968; Wilson, Chrispin and Carter 1969; Bailey 1973; Horton et al 1978; Rimoin and Lachman 1990). It is suggested that there is a marked variation in clinical expressivity (Horton et al 1978; Kaitila et al 1989) and the term 'diastrophic variant' has caused much confusion. This is best considered as mild DD (Horton et al 1978; Lachman et al 1981). Pseudodiastrophic dysplasia, however, is definitely a separate disorder (Eteson et al 1986).

Fig. 1

A new-born child with the typical appearance of diastrophic dysplasia.
Diastrophic dysplasia has been observed in most Caucasian populations. The prevalence is exceptionally high among the Finns, at 1:30 000 (Kaitila et al 1989). We have performed a comprehensive clinical and radiological evaluation and follow-up of 105 of the known 160 patients in Finland, obtaining much new and detailed information. We now report and discuss the types of foot deformity in DD and classify them.

**PATIENTS AND METHODS**

The patients had been referred to the Children’s Hospital and to the Orthopaedic Hospital of the Invalid Foundation from the whole of Finland from 1947 to 1987. Diagnosis was based on established features of diastrophic dysplasia (Lamy and Maroteaux 1960; Bailey 1973; Kaitila et al 1989). Of the 105 patients reviewed, three were excluded because of insufficient information leaving 102, of whom 44 were male and 58 female. The age range at review was from new-born to 79 years. The state of the feet at the first examination was obtained from hospital records and from photographs; most patients had also had radiographs at that time. All this information was used for the classification.

**RESULTS**

The age of the patients at the time of the first examination is shown in Figure 2; about one-third had been first examined before normal walking age.

The type of foot deformity could be decided in 102 patients, or 204 feet. Five groups were defined:

1. **Clinically normal.** The appearance and function of the foot was within the normal variation for the age of the patient.
2. **Metatarsus adductus** (Fig. 3). There was abnormal adduction of the forefoot, but the foot could be placed in a plantigrade position.

3. **Tarsal valgus combined with metatarsus adductus** (Figs 4, 5). These feet showed significant valgus deformity at the ankle, distinguishing them from the former group. The foot was plantigrade.
4. **Equinus** (Figs 6, 7). The foot was in fixed equinus, but with no significant varus deformity either in the heel or in the forefoot. Nearly all these feet also had metatarsus adductus. Weight-bearing was on the metatarsal heads.

**Fig. 4**

Tarsal valgus deformity combined with metatarsus adductus in a 10-year-old patient.
5. Equinovarus adductus (Figs 1, 8). The foot was fixed in equinus and varus and the forefoot adducted. The weight-bearing area was on the lateral side of the forefoot.

The number and proportion of feet in each of the five categories are shown in Figure 9. Equinovarus adductus deformity was seen only in 29%; and even adding the equinus feet gives only 37% with 'club foot'. The commonest deformity was tarsal valgus with metatarsus adductus (43%). Almost two-thirds of the feet (63%) were plantigrade.

DISCUSSION

Because of the genetic nature of diastrophic dysplasia the deformities are not strictly comparable to those in other affections. We chose criteria which would enable a relatively easy and reliable classification, though this gave some difficulties with borderline cases of plantigrade feet and between some equinus and equinovarus adductus feet. The variable severity of the disease also caused difficulties in classification. Because of the variation in age at the first examination we can give only a cross-section of the spectrum of the deformities.

Scattered observations in patients who had been followed up gave some information about the natural
history of the feet. The problems were different in equinus and plantigrade feet. Most of the equinus and equinovarus adductus feet had been treated operatively (55 of 76 feet). All of the others which were followed up remained in equinus position, although in some cases there had been a change from pure equinus to equinovarus. There was recurrence of deformity in over 80% of the feet treated by tenotomy, or lengthening of the calcaneal tendon, or posteromedial release. This tends to confirm that untreated feet will retain their original equinus or equinovarus deformity.

Almost all the plantigrade feet had been left untreated, but during follow-up some originally normal-looking feet developed a significant metatarsus adductus deformity which became worse with increasing age. Most feet with metatarsus adductus tended to develop progressive tarsal valgus deformity during follow-up; some of the feet in these categories would have been classified differently if they had been examined earlier or later.

Several circumstances may contribute to progression of the deformity during growth, though genetic factors are mainly responsible for the final appearance of the individual, and have a strong influence throughout development. It is extremely difficult to modify this development by treatment.

Another important cause is the loading of the skeleton by mechanical forces. A significant pathological feature of diastrophic dysplasia is the abnormality of cartilage (Horton et al. 1979). This abnormality makes the cartilage softer than normal, and the cartilaginous parts of the skeleton tend to become deformed under load. This influence is also seen in the feet, although this is not as striking as in the hips and knees (Ryöppy et al 1990).

The other element influencing foot deformity may be ligamentous laxity. It is a paradox that while the range of motion is restricted in most joints, some may show abnormal laxity from the beginning. This may help to explain why most of the originally plantigrade feet develop valgus deformity at the ankle when weight-bearing begins.

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REFERENCES


