MACRODACTYLY IN THE FOOT

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Seven patients with macrodactyly in the foot are reported. None showed any stigmata of neurofibromatosis and all were found to have excessive accumulation of fibro-fatty tissue as the most striking pathological feature. It is suggested that this may represent the basic lesion in this condition. The literature is reviewed and attention is drawn to the differences between macrodactyly in the hand and in the foot.

Although generally regarded as a rarity, disproportionate enlargement of one or more fingers or toes is the subject of many reports. Most papers relate chiefly or exclusively to macrodactyly of the hand, which appears to be more common (Timoney 1944; Ben-Bassat, Casper, Kaplan and Laron 1966; Barsky 1967; Ranawat, Arora and Singh 1968; El-Shami 1969). In 1925 Feriz collected a number of cases of macrodactyly in the foot, but there does not appear to be a more recent report dealing entirely with this deformity.

THE PATIENTS

Of the seven patients, two were African, two Indian, two coloured, and one white. There were five females (Table I). Clinical photographs and radiographs were taken of each patient, and five of the excised specimens were examined histologically.

The enlargement began shortly after birth in every patient and was progressive. None gave a family history of any similar deformity and there were no clinical features of neurofibromatosis. Six patients had enlargement of the forefoot. The remaining patient (Case 6) showed only partial hypertrophy of one toe, restricted to the distal end. Two of the patients had macrosyndactyly, and four had hypertrophy of the metatarsal bones.

The increase in size and the appearance were the indications for surgical treatment. The deformities were managed by amputations of varying extent, combined with the removal of as much of the fibro-fatty tissue as possible. The amputation through the interphalangeal joint performed in Case 7 was done at the request of the parents, who refused a more radical procedure.

Although all the patients seemed satisfied with the results of treatment, the final appearance in the more severe deformities was not as good as had been hoped. Delayed wound healing occurred after four operations. Inadequate initial defatting combined with subsequent regrowth of the fibro-fatty tissue were the main reasons for failure, and two patients required a second, more proximal amputation.

TABLE I

Table showing the relevant details of the patients reported

<table>
<thead>
<tr>
<th>Case number</th>
<th>Patient and sex</th>
<th>Toes involved</th>
<th>Forefoot involvement</th>
<th>Metatarsal hypertrophy</th>
<th>Macro-syndactyly</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>White female</td>
<td>Right 1st and 2nd toes</td>
<td>Present</td>
<td>Absent</td>
<td>Absent</td>
</tr>
<tr>
<td>2</td>
<td>Coloured female</td>
<td>Bilateral 1st, 2nd and 3rd toes</td>
<td>Severe</td>
<td>Present in right 2nd and 3rd rays</td>
<td>Left 2nd and 3rd toes</td>
</tr>
<tr>
<td>3</td>
<td>Coloured female</td>
<td>Left 1st and 2nd toes</td>
<td>Present</td>
<td>Present</td>
<td>Absent</td>
</tr>
<tr>
<td>4</td>
<td>African male</td>
<td>Left 2nd and 3rd toes</td>
<td>Present</td>
<td>Present</td>
<td>Present</td>
</tr>
<tr>
<td>5</td>
<td>African female</td>
<td>Left 1st and 2nd toes</td>
<td>Present</td>
<td>Absent</td>
<td>Absent</td>
</tr>
<tr>
<td>6</td>
<td>Indian male</td>
<td>Left 4th toe</td>
<td>Absent</td>
<td>Absent</td>
<td>Absent</td>
</tr>
<tr>
<td>7</td>
<td>Indian female</td>
<td>Right 1st toe</td>
<td>Present</td>
<td>Present</td>
<td>Absent</td>
</tr>
</tbody>
</table>

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DISCUSSION

Macrodactyly is an uncommon malformation characterised by an increase in size of the constituent elements of a digit. Previous authors have stated that the relevant literature is scarce (Moore 1942; Clifford 1959; Tuli, Khanna and Sinha 1969), but it is clear from the bibliography quoted by Kelikian (1974) that this is not so. Most reports have been of isolated cases which, individually, have contributed only slightly to our understanding of the subject. In 1967, Barsky reviewed the literature and stated that there were two varieties of true macrodactyly. The more common static type showed enlargement from birth, and increase in size was proportional with growth. The second was a progressive form, with disproportional growth of the affected digit, which increased in size at a rate faster than could be attributed to the normal growth pattern. This group, in which the enlargement was complicated by the overgrowth of fatty tissue, appeared to be extremely rare, with only seven cases reported. All of our cases appear to belong to this latter category.

Little is known of the aetiology. Most reports indicate a slight male preponderance (Jones 1963; Barsky 1967; Tsuge 1967), though five of our patients were female. Heredity does not appear to play a role (Feriz 1925; Rechnagel 1967; Kelikian 1974) and none of our patients gave a family history of any similar deformity. Barsky (1967) reported normal chromosome studies, although abnormalities have been described in association with congenital asymmetry (Ferrier, Ferrier, Stalder, Bühler, Bamatter and Klein 1964). A chromosome study on one of our patients was normal.

Equally undecided is the pathogenesis. There has been a tendency to over-simplify the problem by attempting to define a single process to account for all types of digital enlargement. At present there seem to be two major concepts, the more popular relating the condition to a manifestation of neurofibromatosis (Moore 1942; McCarroll 1950; Allende 1967). Brooks and Lehman (1924) believed that the development of neurofibromata in the perioseal nerves caused bony destruction and regeneration, and claimed neurofibromatosis to be the only possible cause of the rapid localised overgrowth. Inglis (1950) suggested that the enlargement was influenced by the neuro-intrinsic factor of neurofibromatosis acting locally. However, Thorne, Posch, and Mladick (1968), reporting thirteen cases of macrodactyly, did not find any clinical or histological evidence of neurofibromatosis. Kelikian (1974) was unable to correlate the two conditions, while conceding that macrodactyly could represent a "forme fruste" of neurofibromatosis. In addition to the absence of a genetic factor, we failed to find any features of this disease in the patients examined.

The second concept is that the basic process is one of lipomatous degeneration. Since the accumulation of fat appeared the most striking feature of pedal macrodactyly, Feriz (1925) derived the term macrodystrophia lipomatosa progressiva, which has been suggested as the basic lesion in reports of the manual variety (Golding 1960; Mikhail 1964; Ranawat et al. 1968). Though both varieties show this overgrowth of fibro-fatty tissue, more notable findings in macrodactyly of the hand are hypertrophy and tortuosity of the palmar nerves and their digital branches, features which are rarely seen in the foot.

This implies a different aetiology; yet a common neurological pathogenesis cannot be easily dismissed since the digits involved in both varieties are predominantly of a pre-axial distribution. The possibility of autonomic nerve dysfunction has been considered (Moore 1941), resulting in localised abnormalities of fat deposition and growth. We agree with Thorne et al. (1968) that such a theory is attractive, though at present purely conjectural.

Clinical features—The deformity appears soon after birth and comprises an enlarged but otherwise normal-looking toe. The entire digit may not be uniformly involved, and examples of partial hypertrophy are reported, with the enlargement restricted to the distal end (Tsuge 1967). The patient in our Case 6 demonstrates this (Fig. 1). Some combination of first, second or third toe involvement is almost invariable, with the second toe most commonly affected (Charters 1957; Barsky 1967; Tuli et al. 1969). Enlargement of the little toe appears to be rare, the only case being one of Moore's (1942), with macrodactyly of the left three outer toes and right two outer toes. Most of the abnormal bulk of the toe is due to the excessive fibro-fatty tissue, especially abundant on the sides and on the plantar aspect. This asymmetrical hypertrophy causes the affected digits to curl dorsally. The skin becomes markedly thickened and the digits have a soft, rubbery consistency. Macrosyndactyly, said to be commoner in the foot, was present in two of the patients in this series. Though the digital enlargement is the most immediately obvious feature, involvement of the forefoot is often overlooked. The fibro-fatty tissue extends from the toe into the forefoot, causing it to expand laterally.

Hypertrophy of the soft tissues on the dorsum is said to be rare (Tsuge 1967), the more impressive enlargement being present on the plantar surface (Fig. 2). Most cases show this proximal extension (Tsuge 1967; Thorne et al. 1968; Tuli et al. 1969) and in all but one of our patients, this was a notable feature.

Treatment—Ablation, combined with the removal of as much fibro-fatty tissue as possible, is the method of treatment generally accepted for pedal macrodactyly. Destruction of the epiphysis (Clifford 1959; Jones 1963), and shortening of the digit by a plastic procedure (Tsuge 1967), are more appropriate to the hand, where amputa-
tion is to be avoided. Because many of the deformities have reached grotesque proportions before presenting (Fig. 3), even radical surgery may prove cosmetically disappointing. Amputation gives no guarantee against overgrowth of the adipose tissue proximal to the site of ablation, and secondary procedures may be necessary. Early complications after operation, mostly from delayed wound healing, are also common because of the interference with the blood supply during excision of the fibro-fatty tissue and to the marked surrounding fibrosis. Four of our incisions failed to heal primarily.

Radiology—Increase in the length and breadth of the phalanges of the affected digit is an integral feature of macrodactyly, but metacarpal or metatarsal bone involvement is not so clearly defined. Barsky (1967) stated that they are not involved, as did Tuli et al. (1969) and Poznanski (1974), while Tsuge (1967) and Thorne et al. (1968) found their enlargement uncommon. In the hyperostotic variety of manual macrodactyly, Kelikian (1974) reported that both the metacarpal bones and phalanges may overgrow, and because osteoplastic changes are more common in the foot, it is surprising that metatarsal hypertrophy has not been noted more often. In this series four patients showed this feature (Figs. 4 and 5).

Studies in bone maturation have been reported by Ben-Bassat et al. (1966) and El-Shami (1969). They noted that the bone age, as denoted by the epiphysial centres in the phalanges of the affected digits, was increased when compared to the normal side. The assessment of bone age from radiographs of the feet is not standardised, and we therefore compared the radiological appearance of the epiphysial centres of the affected toes with those of the normal side; the bilateral case was excluded. Two patients demonstrated advanced bone maturation when assessed in this way, and this is shown in Figure 5 in which the metatarsal capital epiphysis and the proximal phalangeal basal epiphysis are well formed and larger than their normal counterparts.

Figure 4. Case 7—A radiograph showing hypertrophy of the phalanges and the first metatarsal bone. Figure 5. Case 4—A radiograph showing advanced bone maturation of the affected toes.
Pathology — The most striking and constant feature is the overgrowth of fibro-fatty tissue. Barsky (1967) described the tissue as resembling adult subcutaneous fat rather than children's fat, with large lobules that are difficult to extrude by pressure. The resemblance to a neoplastic proliferation was a striking feature and has already been noted (Feriz 1925; Ben-Bassat et al. 1966; Ranawat et al. 1968), the hypertrophied adipose tissue pervading all surrounding tissues including muscle and nerve. An increase in bone marrow fat reported by Oosthuizen and Barnetson (1947) was also found. Feriz (1925) noted fibrous bands radiating outwards from the periosteum, and Minkowitz and Minkowitz (1965) described increased fibroblastic activity near the periosteum, the bands enmeshing the nerves and blood vessels. Ben-Bassat et al. (1966) felt that this proliferation of fibroblastic tissue between the cortex and the periosteum was the most outstanding pathological finding and accounted for the cortical thickening and gigantism of the phalanges in the affected digits. Figure 6 shows this feature, and further examination under higher magnification suggested that the cells are either proliferating periosteal cells or fibroblasts (Fig. 7).

Because many authorities believe macrodactyly to be related to a neurogenic disorder, considerable care was taken in the search for abnormalities in the peripheral nerves, which curiously are uncommon in pedal macrodactyly. Our specimens had nerves more prominent than usual, with much proliferation of the epineural and perineural tissues (Fig. 8).

The nerve fascicles were normal and neurofibromata were not seen. The histology of the blood vessels showed no evidence of endarteritis, which Moore (1941) had suggested as being indicative of dysfunction of the autonomic system. The skin showed dermal fibrosis and flattening of the rete pegs, a feature noted by Tuli et al. (1969). The microscopical changes were, in general, unremarkable and we could not find any important differences from previous reports.

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