CONGENITAL MACRODACTYLY
A Case Report with a Three-year Follow-up

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The literature pertaining to macrodactyly is sparse (Fèvre and Bricage 1936, Clifford 1959, Laurenzi 1962) and a clear concept of its pathogenesis does not yet exist. The most popular theory relates the condition to neurofibromatosis (Moore 1942, Inglis 1950, Mouly and Debeyre 1961). Among the cases reported some were in children (Moore 1942, Inglis 1950) but, so far as is known, studies in bone maturation and development of the affected finger have not been reported.

CASE REPORT

In April 1960, after a normal first pregnancy and labour, a boy weighing 3.3 kilograms was born to young Yemenite Jewish parents. Macrodactyly, involving the middle finger of the left hand only, was present at birth. No other abnormality was found. The physical and mental development for the first three years was normal. Body length increased along the tenth percentile and weight along the twenty-fifth percentile.

The affected finger, as shown in Figure 1, showed great uniform enlargement, thickening of soft tissues, nail and bones with limitation of movement in the interphalangeal joints, which had ulnar deviation. Radiographs of the right hand at the age of six months showed that the skeletal age (Greulich and Pyle 1950) was compatible with the chronological age. The left hand, with the exception of the enlarged finger, showed the same degree of maturation. The

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<th>Chronological age (years)</th>
<th>Length of middle finger (centimetres)</th>
<th>Bone age (years)</th>
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<tr>
<td></td>
<td>Normal</td>
<td>Abnormal</td>
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enlarged finger showed, apart from gigantism, epiphyseal growth centres in the phalanges which were compatible with a bone age of approximately two years. In the normal fingers no epiphyseal centres of ossification were demonstrable radiologically. The difference in bone maturation between the macrodactyle and the normal finger is seen even better at the age of twenty months (Fig. 2). Table I shows a comparison between the length of the normal and the abnormal fingers and their bone ages until the age of three years.
Because of the loss of function caused by the gigantism and because psychological disturbances were already manifest, amputation at the metacarpo-phalangeal joint was done when the child was three years old. Angiography before operation showed no vascular abnormality (Fig. 3).

**PATHOLOGICAL FINDINGS**

**Macroscopic examination**—The finger measured nine by four centimetres. A transverse section through the distal one-third (Figs. 4 and 5) and a longitudinal section through the proximal two-thirds of the digit showed that the hypertrophy involved both soft tissue and bone. In the
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Figs. 3 and 4

Figure 3—Angiograph at 3 years of age just before amputation. Figure 4—A transverse section through distal phalanx at the level of the base of the nail, showing the relationship between bone and soft tissue and tumour-like proliferation of the adipose tissue.

Fig. 5

Microphotograph of the distal phalanx, as in Figure 4. (Haematoxylin and eosin, 65.)
distal third the enlargement was mostly caused by soft tissue. The distal phalanx was tubular in appearance, one centimetre in diameter and with a thin wall and relatively wide lumen. It was surrounded by a broad ring of fatty tissue one and a half centimetres in width.

In the proximal two-thirds of the finger the enlargement was caused by both hypertrophy of bone and soft tissue. The two proximal phalanges were approximately twice the size of normal phalanges of a child of comparable age (Fig. 6). These were also surrounded by adipose tissue but to a lesser extent.

Microscopic examination—Histological abnormalities were found in the soft tissues as well as in the bone. In the abundant fatty tissue small nerve fibres were detected with proliferation of perineural as well as endoneural connective tissue. No pathological changes were observed in the blood vessels. The phalanges showed an unusual abnormality in the form of a band consisting of fusiform cells interspersed with collagen fibres which was found between the periosteum and the cortical bone (Fig. 7). This band was of variable thickness widening towards the end of the phalanges (Fig. 8). The fusiform cells appeared to be either fibroblasts or osteoblasts. Above the cortical bone a layer of osteoid tissue was present and the surface showed Howship’s lacunae containing osteoclasts (Fig. 9). No cartilage cells were seen. The areas of endochondral ossification and the centres of ossification were undisturbed. The bone marrow of the phalanges was adipose with a hollow and abnormally wide marrow cavity.
FIG. 7
The band of proliferating fusiform cells, interspersed with collagen fibres, can be seen between the periosteum and the cortical bone. (Haematoxylin and eosin, ×28.)

FIG. 8
Figure 8—The periosteal segment shown in Figure 7 at a higher magnification. (Haematoxylin and eosin, ×96.)

FIG. 9
Figure 9—Osteoid tissue is shown and there are Howship’s lacunae containing osteoclasts on the border between the cortical bone and proliferating periosteal cells. (Haematoxylin and eosin, ×96.)
DISCUSSION

Proliferation of the adipose tissue and fibrosis in the peripheral nerves have been described in previously reported cases of macrodactyly. On the basis of these findings most authors (Moore 1942, Inglis 1950, Mouly and Debye 1961) believe that macrodactyly is related to a neurogenic disorder and that it constitutes a component of neurofibromatosis. Inglis (1950) suggested that "the increased growth of the enlarged digits in macrodactyly is influenced by the neural intrinsic factor of neurofibromatosis acting locally." Moore (1942) described five cases of macrodactyly in all of which pathological changes in the peripheral nerves were found; in one there was a neurofibroma and, in three of the others, there were clinical signs of neurofibromatosis—café-au-lait spots, lipoma and naevi. Inglis (1950) described the appearance of dense connective tissue in contact with the phalanx containing cartilaginous cells. He interpreted this as an early tumorous state, or osteochondromata, which is also associated with neurofibromatosis.

In the case reported here no tumorous growth was demonstrable in the finger or elsewhere, so that we are not justified in regarding it as related to neurofibromatosis. The outstanding pathological finding in this case, which has as yet not been described in other cases of macrodactyly, was the proliferation of the fibroblastic tissue between the cortex and the periostum which was responsible on the one hand for laying down of osteoid tissue and on the other for destruction of bone, with the resulting thickening of the cortex and gigantism of the phalangeal bone.

SUMMARY

1. A patient with macrodactyly of the middle finger of the left hand was followed up from birth until the finger was amputated at the age of three.
2. The affected finger, besides showing gigantism at birth, grew at a faster rate than the normal fingers. The degree of bone maturation (as judged from the appearance and size of the phalangeal ossification centres) proceeded at a faster rate than the normal fingers. No vascular abnormality which could account for the gigantism was detected either radiologically or microscopically. The affected finger showed histological abnormalities of both bone and soft tissues.

REFERENCES