Macrodactyly: Report of three cases

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Introduction

Macrodactyly is a rare congenital anomaly in which there is enlargement of one or several digits of hands or feet. The overgrowth is limited to or predominantly affects the digits and should be distinguished from more extended malformations such as macromelia or hemihypertrophy. Sometimes the words megalodactyly or digital gigantism is used synonymously. Macrodactyly does not appear to be an inherited condition. Although its cause is uncertain, three possible factors are strongly suspected: abnormal nerve supply, abnormal blood supply and abnormal humoral mechanism. Macrodactyly most commonly exists without other conditions but syndactyly is associated with macrodactyly is about 10% of patients.

Case I

A 10-year old boy had history of enlarged left index finger since birth which was progressing in proportion to body growth.

Radiograph showed hypertrophy of soft tissue over the left index finger with enlarged bones of proximal, middle and distal phalanx of index finger. There is also deformity at distal interphalangeal joint of middle finger [Fig. No. 1].

Fig. No. 1 shows hypertrophy of soft tissue over the left index finger with enlarged bones of proximal, middle and distal phalanx of index finger. There is also deformity at distal interphalangeal joint of middle finger.

Fig. No. 2 shows hypertrophy of soft tissue with enlarged bones of proximal, middle and distal phalanx of right middle finger. There was also hypertrophy of soft tissue with mildly enlarged bones of proximal, middle and distal phalanx of right index finger.

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Case II

A 3-year old girl had history of abnormal growth of right middle finger and right index finger. The fingers had grown regularly but for the last one year the growth is static.

Radiograph showed hypertrophy of soft tissue with enlarged bones of proximal, middle and distal phalanx of right middle finger. There was also hypertrophy of soft tissue with mildly enlarged bones of proximal, middle and distal phalanx of right index finger [Fig. No. 2].

Case III

A 10-year girl had history of enlarged second and third toes of right foot since birth. There was gradual increase in size of respective toes.

Radiograph showed hypertrophy of soft tissue and fusion of second and third toes of right foot. Bones of metatarsal, proximal and middle phalanx of second and third toes was also enlarged [Fig. No. 3].

Discussion

In macrodactyly hands and feet are affected with almost equal frequency (1). In both hands and feet, the highest incidence is of second digit involvement, followed by third digit enlargement (1). Involvement of more than one digit is common. Regarding the course of disease, Barsky et al make a distinction between a progressive and a static type (4). In static macrodactyly the deformity is present is infancy. There is usually diffuse enlargement of the digit, however the distal and palmar tissue usually appear more enlarged than the dorsal and proximal tissues. The fingers grow in proportion to normal digital growth. In progressive macrodactyly, the digits may not enlarge during infancy but begin to enlarge rapidly during early childhood, frequently with an annular deformity that makes the finger banana shaped.

The clinical course of overgrowth in macrodactyly is important for prognostic and therapeutic reasons (4). The pathologic substrate of macrodactyly was hypertrophy of bone with hamartomatous overgrowth of predominantly lipomatous or fibrous soft tissue elements (3). The phalanges are always involved and the metacarpals may be enlarged as well. The skin may be thickened and the nails hypertrophied.

Macrodactyly in the majority of patients is an isolated finding without other associated symptoms or systemic involvement (1). There is a range of other, mostly congenital pathologic conditions in which localized overgrowth may mimic the clinical picture of macrodactyly. These include neurofibromatosis, primary lymphatic disorder (Milroy disease) and vascular malformation, for example Klippel-Trenaunay-Weber syndrome. Several rare hereditary syndromes include hamartomatous changes that possibly present as macrodactyly, for example, Proteus syndrome, Bannayan Syndrome, Maffucci Syndrome and Ollier disease.

Diagnostic aids in cases of macrodactyly should include radiography and computed tomography to reveal the extent of bone and tendon involvement. MRI is recommended when the clinical features and plain film findings are indeterminate. Angiography and lymphangiography should be performed when vascular or lymphatic changes, respectively are clinically suspected. Biopsies can provide information on the tissue elements that take part in the formation of the hamartoma.

In our cases, involvement of hands was seen in two patients whereas foot was involved in one patient. Second digit was involved in all the cases and adjoining (third) digit was involved in two patients. Macrodactyly was an isolated finding without any other associated symptoms or systemic involvement.

Fig. No. 3 shows hypertrophy of soft tissue and fusion of second and third toes of right foot. Bones of metatarsal, proximal and middle phalanx of second and third toes was also enlarged.