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CASE REPORT

Congenital macrodactyly : A clinical study

Anjum SYED, Rana SHERWANI, Quamar AZAM, Faisal HAQUE, Kafeel AKHTER

From Aligarh Muslim University, Aligarh, India

Congenital macrodactyly is a rare congenital malformation characterised by progressive enlargement of all mesenchymal elements of a digit. The present study is an attempt to draw the attention towards the similarities and differences between macrodactyly of the hand and foot.

Radiographical, operative findings and histopathological examination of five cases are included in the present study. Emphasis was given to know the possible basic lesion. Radiographic findings, which differentiate this entity from other forms of local gigantism, were also analysed.

The most characteristic finding noted was excessive overgrowth of fibro-fatty tissue with unusually large fatty lobules, apparently fixed by a mesh of dense fibrous tissue. Hypertrophy and tortuosity of the digital nerve, a striking feature in macrodactyly of the hand, was notably absent in cases affecting the foot. None of the patients had any other associated congenital anomalies. Neither the patients nor any of their family members had any stigmata of neurofibromatosis. Chromosomal study was normal in all of them.

We conclude that in macrodactyly of the foot, excessive proliferation and accumulation of adipose tissue was the basic lesion, whereas involvement of the nerve might be the fundamental lesion in gigantism of the hand. Furthermore, whatever be the basic lesion, the final pathway must be either the local deficiency of a growth inhibiting factor or local expression of a basic intrinsic factor, leading to excessive growth of all elements of the digit.

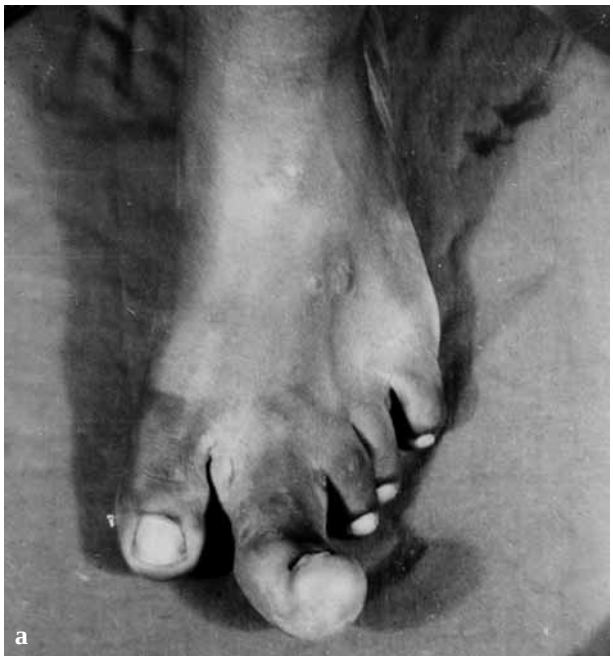
Keywords : macrodactyly ; congenital.

INTRODUCTION

Enlargement of a digit may be due to haemangioma, lymphangioma, lipoma, or tumour mass. In these situations, only a defined element (vessels, subcutaneous fat, bone etc) is affected. Congenital macrodactyly strictly speaking, refers to the rare malformation characterised by enlargement of all structures of a digit or its phalanges, subcutaneous fat, nerve, vessel, skin, nail etc (*1*). Local gigantism has also been described under many names such as megalodactyly, dactylomegaly, macrosomia, macrodystrophia lipomatosa.

Review of literature revealed that macrodactyly of the hand is more common and most studies (*1, 2, 5, 9*) report chiefly or exclusively findings in macro-

■ Anjum Syed, MD, Registrar.
■ Faisal Haque, MD, Lecturer.
Department of Radiodiagnosis, J.N.M.C., A.M.U., Aligarh, India.
■ Rana K Sherwani, MD, Reader.
■ Kafeel Akhter, MD, Lecturer.
Department of Pathology, J.N.M.C., A.M.U., Aligarh, India.
■ Quamar Azam, MS, Registrar.
Department of Orthopaedic Surgery, J.N.M.C., A.M.U., Aligarh, India.
Correspondence : Dr Anjum Syed, Department of Radiodiagnosis, J.N.M.C., A.M.U., Aligarh, India.
E-mail : anjumsyed@yahoo.com.



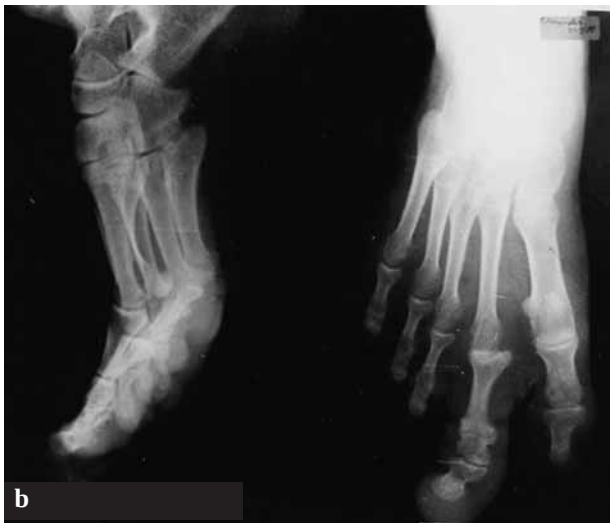


Fig. 1. — **a)** Clinical photograph of case 1 showing macrodactyly of the second toe with dorsal and lateral curvature ; **b)** Radiograph of the same patient showing both soft tissue and phalanges involvement of the second toe.

dactyly of the hand. Feriz (*6*) in 1925 and Dennyson (*4*) in 1977 collected a number of cases of macrodactyly of the foot.

The present study is an attempt to draw the attention towards the similarities and differences between macrodactyly in the hand and the foot. It is also discussed how radiographic and sonographic features can help in differential diagnosis.

MATERIAL AND METHOD

We had an exceptional opportunity to study three cases of local gigantism of the foot and two cases of macrodactyly of the hand over a period of 4.5 years. Although parents noted the abnormalities shortly after birth, none of the patients presented before five years of age. Increasing difficulty in wearing shoes and cosmetic worries were the main cause for seeking medical advice. The presenting age ranged from 7 to 17 years. Three of these were male while two were female.

A detailed history revealed that the enlargement began shortly after birth in all the cases and the affected digit grew at faster rate. Clinical examination of the affected finger or toe revealed thickened pale, glossy and non-tender skin. Consistency was firm in some areas and soft in others. Dorsal and lateral curvature of the affected digit was seen in two cases (fig 1), while plantar flexion was seen in one case.

None of the patients had any other associated congenital anomalies. No area of pigmentation was noted anywhere. Chromosomal studies performed demonstrated no abnormalities in any patient. None gave a family history of any such deformity. Neither the patients nor their families had nodules in the skin or subcutaneous tissue.

Clinical photographs and radiographs were taken in every case. In three of the five cases de-fatting surgery was initially undertaken, but due to recurrence of the deformity, amputation of varying degree was required in all the cases. Inability to remove all the fibro-fatty tissues combined with subsequent re-growth was the main reason of failure. All the excised specimens were examined histologically. Table I shows the relevant details of all cases.

Ultrasound (Gray scale and Doppler) examination of the digits revealed diffuse soft tissue thickening. No evidence of increased blood flow was found in the affected region in any of the cases.

RESULTS

In all cases of macrodactyly of the foot, there was abundance of fatty tissue ; the fat lobules were unusually large and difficult to remove (fig 2c), apparently fixed by a mesh of dense fibrous tissue. Enlargement of the digital nerve was conspicuous

Table I. — Relevant details of all cases

Case number	Age (years)/Sex	Location of anomaly	Digit involved	Syndactyly	Neural involvement
1	13 / male	Foot	Second toe	Absent	Absent
2	17 / female	Foot	Fourth toe	Absent	Absent
3	11 / male	Foot	First & Second toe	Absent	Absent
4	16 / female	Hand	Middle finger	Absent	Present
5	7 / male	Hand	Index & Middle finger	Present	Present

by its absence in all three cases of macrodactyly of the foot. However, remarkable enlargement of the digital nerve (fig 3c) was noted in both cases of macrodactyly of the hand.

The indication for surgery was mainly cosmetic in cases of macrodactyly of the hand, whereas inability to wear shoes was the reason in dactylomegaly of the foot. Delayed wound healing occurred in two cases at the foot. The final appearance in both cases was not as good as expected, however both of them were able to comfortably wear shoes and seemed satisfied.

Radiograph

Radiographs demonstrated enlargement of both soft tissues and phalanges (fig 1b, 2a, 3b). Soft tissues at the volar surface were mostly involved. Mottled lucencies were noted in soft tissues in all three cases at the foot. Bone enlargement was restricted to the phalanges. Phalanges were longer and broader when compared with uninvolved phalanges of the same extremity. Metatarsal involvement was not seen in any of our cases.

Pathology

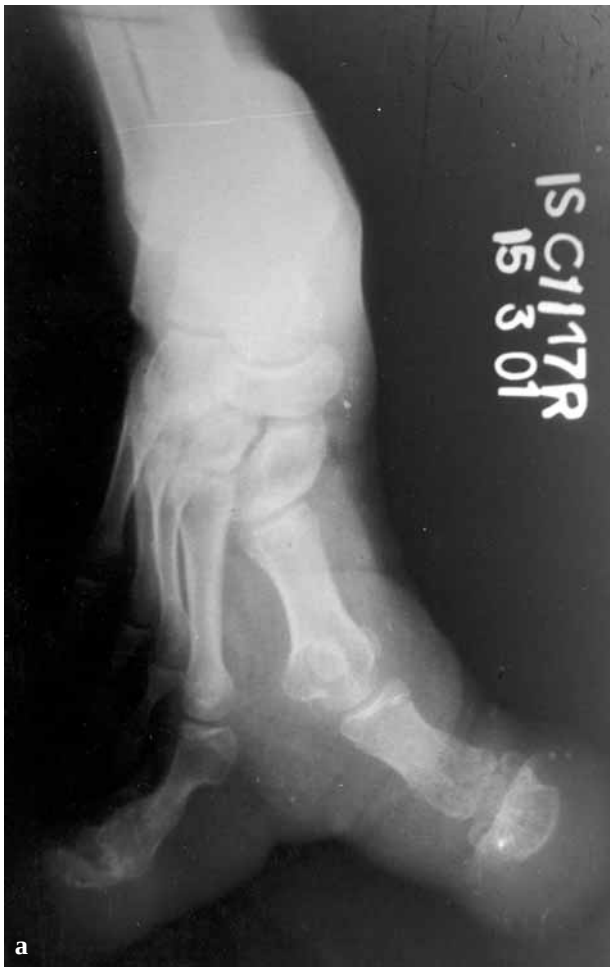
The most striking feature noted microscopically was abundant overgrowth of fibro-fatty tissue (fig 2c, 3c). The hypertrophied adipose tissue with very large lobules of fat pervaded all surrounding tissues. An increase in bone-marrow fat was also seen. Proliferating fusiform cells, interspersed with collagen fibers, were seen between periosteum and the cortical bone.

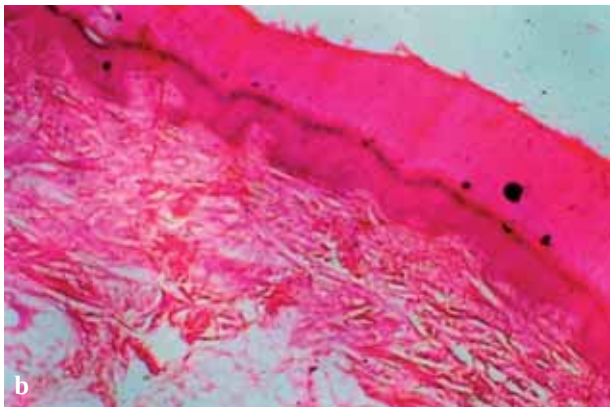
In addition to startling abundance of fatty tissues, significant enlargement of the digital nerve was noted in both cases of macrodactyly of the hand. There was excessive proliferation of epineural and perineural tissues ; however, nerve fascicles were normal and neuromata were not seen in macrodactyly of the hand (fig 3c). The skin showed dermal fibrosis and flattening of the rete-pegs (fig 2b).

DISCUSSION

Macrodactyly appears to be more common in the hand than in the foot (*1, 2, 5*). The literature indicates a slight male preponderance (*1, 11, 12*). The male to female ratio in this series was 3:2. None of our patients had any family history of similar deformities. This is consistent with the previous literature, which states that heredity does not play a role (*1, 6, 11*).

Our operative findings and microscopic examination showed that in all three cases of macrodactyly of the foot, there was excessive proliferation of adipose tissue coupled with absence of any neural involvement. In sharp contrast to these findings, microscopic examination of both cases of hand macrodactyly clearly demonstrated involvement of the digital nerve. Based on these observations, we agree with Dennyson *et al* (*4*) who studied seven cases in the foot and claimed that hypertrophy and tortuosity of the digital nerve, a notable finding in macrodactyly of the hand, is rarely seen in the foot. We believe that congenital macrodactyly is an independent pathological process and this is consistent with Barsky (*1*), who postulated that, “macrodactyly is the result of interaction of two or more intrinsic and extrinsic agents (genetic and/or environmental) each unable to produce the deformity itself”. He further conceptualised that during





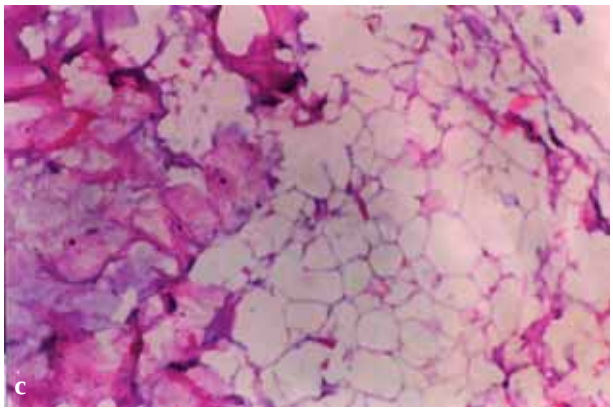


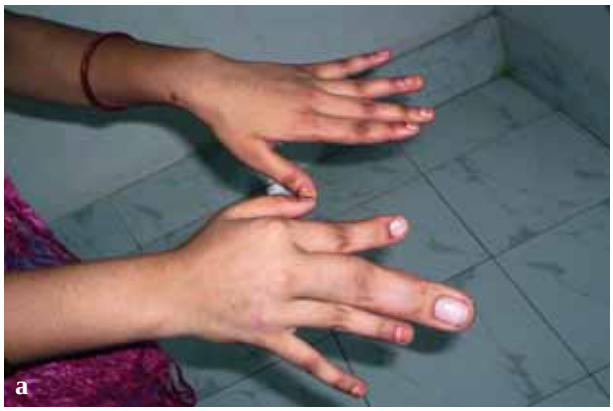
Fig. 2. — **a)** Radiograph of the foot of case 3 showing involvement of first and second toe ; **b)** Section of involved skin of macrodactyly of the same patient showing excessive deposition of connective tissue and flattening of rete pegs due to excessive deposition of connective tissue. H&E Stain 10X (X2.5X) ; **c)** Histological section of the same patient showing hypertrophied adipose tissue interspersed with collagen fibers. Lobules of fat were noted to be unusually large. H&E Stain 20X (X2.5X).

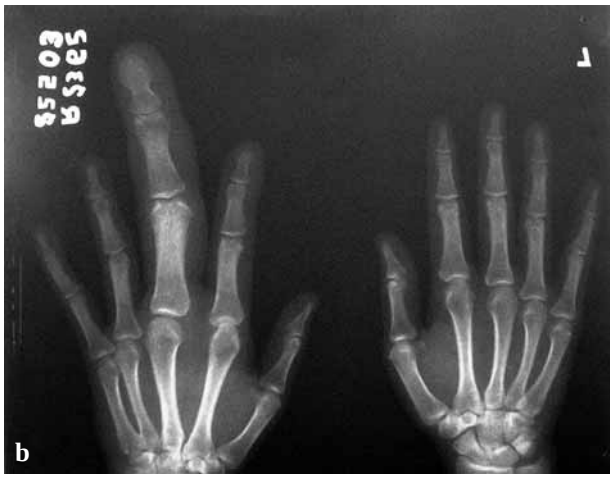
foetal development, some disturbance of growth limiting factor occurs in the affected local area, which continues to increase in size.

The aetiopathogenesis of this condition is still obscure. Two concepts have been put forward : one is association with neurofibromatosis, and the second states that the basic process is lipomatous degeneration.

Brooks and Lehman (*3*) believed that neurofibromatosis of periosteal nerves is the main cause of rapid localised overgrowth. They stated : "we know of no other condition in which there is a spontaneous excessive growth in length of a single bone other than Von Recklinghausen's disease". Moore (*8*) forwarded the strongest argument in favour of the view that neurofibroma is the cause of the hypertrophy after his observation that there is a definitive segmental relationship between the affected nerve and the overgrowth. Inglis (*7*) suggested that the enlargement was influenced by the neuro-intrinsic factor of neurofibromatosis acting locally. He demonstrated a strikingly similar pathologic picture between the neural enlargement of macrodactyly and section of terminal branches of proved cases of neurofibromatosis.

However, Thorne *et al* (*10*) in their study of thirteen cases did not find any evidence of neurofibromatosis. Feriz (*6*) coined the term macrodystrophia lipomatosa progressiva after his observation of excessive proliferation of fat in pedal macrodactyly.





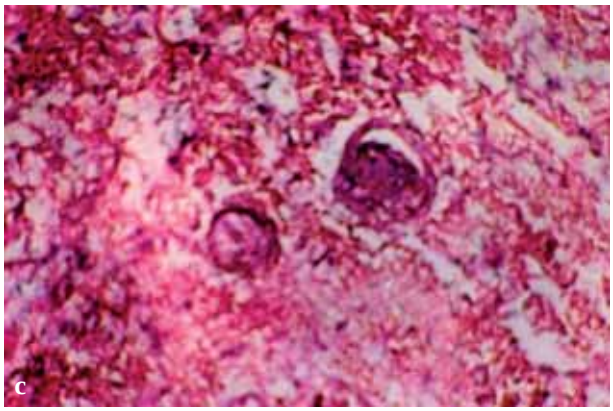


Fig. 3. — **a)** Clinical photograph of case 4 showing enlargement of middle finger ; **b)** Radiograph of the same patient showing enlargement of both soft tissue and phalanges of middle finger ; **c)** Photomicrograph of the same patient showing marked proliferation of fatty tissue along with involvement of digital nerve with excessive epineural and perineural tissues.

Table II. — Differential diagnosis of local gigantism

Increase in all mesenchymal elements	True macrodactyly
Tumorous overgrowth of a single element	Hemangioma Lymphangioma Ollier's disease Plexiform neurofibromatosis Klippel-Trenaunay syndrome

Authors like Ben-Bassat *et al* (*2*), El-Shami (*5*) and Dennyson *et al* (*4*) demonstrated advanced bone maturation of the affected digit, in comparison to the radiological appearance of the epiphyseal center of the unaffected toes. Barsky (*1*) and Tuli (*11*) stated that metacarpals or metatarsals are not involved, while Thorne *et al* (*10*) stated that they are rarely involved.

We attempted to study how plain radiographic and sonographic examination might be useful in narrowing down the differential diagnosis (table II). In haemangiomas and lymphangiomas, plain radiographs showed hypertrophy of soft tissue without involvement of phalanges. No evidence of enchondromas in plain roentgenograms further eliminates the possibility of Ollier's disease. Color Doppler sonography further helps in ruling out the possibility of Klippel-Trenaunay syndrome by demonstrating absence of increased blood flow.

CONCLUSION

Considering the rarity of macrodactyly and in absence of any genetic involvement, no theory can be easily formed or dismissed. Our histopathologic findings suggest that in pedal macrodactyly excessive proliferation and accumulation of fat is the basic lesion, whereas in macrodactyly of the hand involvement of nerves might be the fundamental lesion.

In addition to our study, following careful review of the literature, we noted that the selectivity of the disease is such that only a single digit is affected, or when more than one digit is affected, usually these digits are adjacent. Furthermore, we observed overgrowth of the surrounding tissues even after ablative surgeries, we thereby concluded, "whatever the fundamental lesion, the final pathway has

to be either local deficiency of growth inhibiting factor or excessive local expression of basic intrinsic factor, causing excessive growth of all elements of the digit".

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