ORIGINAL ARTICLE

# Our experience with macrodactyly: a rare congenital anomaly

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**Abstract**

**Objective:** To present our experience with "Macrodactyly", a rare congenital anomaly and its associated anomalies.

**Background:** Macrodactyly is an increase in the size of one or several fingers or toes. The overgrowth is limited to or predominantly affects the digits. It is characterized by an increase in all mesenchymal elements particularly fibro-adipose tissue. It does not appear to be an inherited condition and is thought to be caused by abnormal nerve supply, abnormal blood supply or abnormal humeral mechanisms. Pathologically, they are benign, soft tissue growths. Macrodactyly is commonly an isolated condition but other congenital anomalies are associated with it. It can be static or a progressive disorder. Soft tissue debulking, phalangectomies, ray resection, ostetomies and arthrodesis of interphalangeal joints are different modes of treatment.

**Design:** Descriptive case series.

**Place and duration of study:** This study was conducted in Plastic Surgery Unit of Hayatabad Medical Complex Peshawar and Orthopaedic unit of Khyber Teaching Hospital, Peshawar from April 2007 to December 2009.

**Patients and methods:** A total of 32 patients were registered during the study period. Patients were admitted through out patient department, written informed consent was obtained from all individuals. Detailed history was taken, every patients was assessed clinically and radiologically. All patients were followed for recurrence. 2 patients were lost in follow up and the study was completed on 30 patients.

**Results:** Mean age of the patients was 13.7 years. Out of 30 patients, 19 were male and 11 were female. Hands were involved in 20 patients and feet in 10 patients. There was no bilateral hands or feet involvement. Eighteen patients had progressive and 12 patients have static macrodactyly. Seventeen patients had isolated macrodactyly while in 13 patients macrodactyly was associated with other congenital anomalies most commonly syndactyly. Most commonly involved digit was index finger in hand and big toe in foot.

**Conclusion:** Macrodactyly is a rare congenital anomaly but cosmetic and functional disability of the patient is significant. Although it is mostly isolated but a significant number of cases were associated with other congenital anomalies which necessitate further research in this field.

**Key Words:** Congenital, Syndactyly, Hemangioma, Neurofibroma

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## Introduction:

Macrodactyly is a rare congenital, non-hereditary malformation presenting as an increased size of one or several fingers or toes[1,2,3]. The overgrowth is limited to or predominantly affects the digits[2]. It is characterized by an increase in all mesenchymal elements particularly fibro-adipose tissue[4,5,6]. Macrodactyly should be distinguished from more extended malformations such as macromelia or hemihypertrophy[1,3,7]. It

has been called as partial acromegaly, dactylomegaly, limited gigantism, macrodactyly and macrodystrophia lipomatosa.[1,8] It has been first described by Feriz in 1925, as partial gigantism of lower extremity.[4,8,9]

Macrodactyly does not appear to be an inherited condition and its exact cause is not known.[1,2,4,8] Three possible factors are strongly suspected in its causation: abnormal nerve supply, abnormal blood supply and abnormal humeral mechanisms[2]. The majority of cases of macrodactyly reported in the literature represent hamartomas comprising varying components of digital tissues[1,5]. Pathologically, they can be defined as benign, soft tissue growths[1,2,8,10].

Macrodactyly most commonly exists without other conditions but syndactyly is associated with macrodactyly in about 10% of cases[2]. In 1967, Barsky classified true macrodactyly into two different types: static and progressive forms[11]. True macrodactyly must be differentiated from tumorus enlargement of a single element, as in hemangioma, lymphangioma or enchondroma[5]. Furthermore, macrodactyly may be isolated, but it can also be associated with several syndromes[5, 6, 9].

Treatment of this condition should be individualized and initiated early in life[12]. Soft tissue debulking, phalangectomies, ray resection, ostetomies and arthrodesis of interphalangeal joints are different modes of treatment.[1, 3, 4, 7, 12, 13]

**Materials and methods:**

This study was conducted at Plastic Surgery Unit of Hayatabad Medical Complex Peshawar and Orthopaedic Unit of Khyber Teaching Hospital, Peshawar from April 2007 to December 2009. A total of 32 patients were registered during the study period. Patient of both isolated macrodacly and macrodactly associated with other congenital deformities were included in the study. Patients of macrodactly operated else where were excluded from the study. Patients were admitted through out patients department, written informed consent was obtained from all individuals. Detailed history was taken, every patient was assessed clinically and radiologically.

Patients were followed fortnightly during the initial stages of treatment, and then on monthly basis for 3 months and then every 3 month for three year. Two patients were lost in follow up and the study was completed on 30 patients.

**Results:**

In this study mean age of the patients was 13.7 years with age range from 1 year to 32 years.

Out of 30 patients 19 (63.3%) were male and 11 (36.7%) were female. Hands were involved in 20 (66.7%) patients with left hand in 14 and right hand in 6 patients, and feet were in involved in 10 (33.3%) patients with left feet in 8 and right feet in 2 patients. There was no bilateral hand or feet involvement. Eighteen (60.0%) patients had progressive and 12 (40.0%) patients had static macrodactyly. Seventeen (56.6%) patients had isolated macrodactyly (Figure 1), while in 13 (43.4%) patients macrodactyly was associated with other congenital anomalies most commonly syndactyly (Figure 2), hemangioma (Figure

Figure 1: *Isolated Macrodactyly*

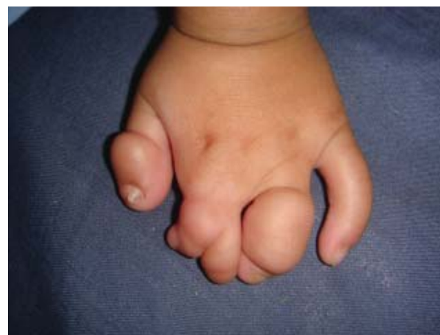

Figure 2: *Macrodactyly associated with Syndactyly*

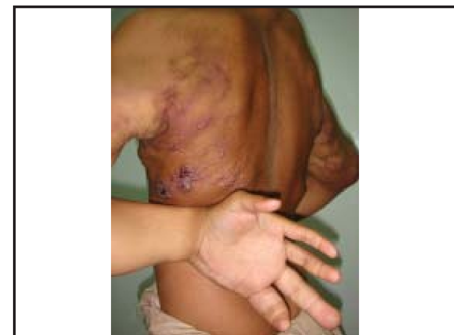

Figure 3: *Macrodactyly associated with Hemangioma*

Table 1: *Overall results of our study*

| Total number of patients | n=30 | |
|---|---|---|
| Mean Age | 13.7 years (range 1-32 years) | |
| Male | 19 (63.3%) | |
| Female | 11 (36.7%) | |
| Hands | 20 (66.7%) | |
| Left hand | | 14 (70.0%) |
| Right hand | | 6 (30.0%) |
| Feet | 10 (33.3%) | |
| Left foot | | 8 (80.0%) |
| Right foot | | 2 (20.0%) |
| Static Macrodactyly | 12 (40.0%) | |
| Progressive Macrodactyly | 18 (60.0%) | |
| Isolated Macrodactyly | 17 (56.6%) | |
| Macrodactyly associated with other anomalies | 13 (43.4%) | |

3), neurofibroma and hemartoma. Overall results are given in Table 1, digital involvement is given in Table 2 and associated congenital anomalies are given in Figure 4.

**Discussion:**

Macrodactyly is an uncommon malformation characterized by an increase in size of constituent elements of a digit[4]. It is present at birth or recognized in early infancy and is progressive during the period of normal skeletal maturation.[10,12] The reported overall sex ratio suggests a slight male predominance[15]. Hands and feet are affected with an almost equal frequency.[1,2] In both hands and feet, second and third digits are involved most frequently.[11,15] In our study male to female ratio was 1.7:1. The ratio of involvement of hands and feet was 2:1. In both hands and feet, the deformity was more common on the left side. Most frequently involved digits were index and thumb followed by ring and middle fingers. In the feet, big toe was most commonly affected followed by 4th and 5th toes.

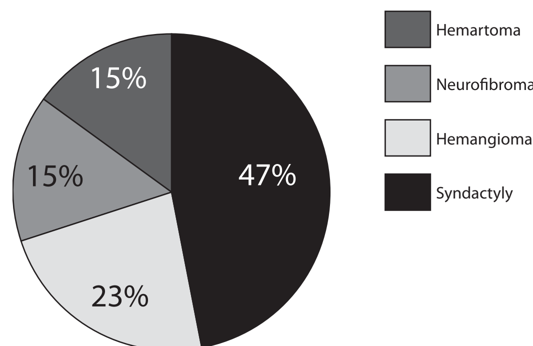


Figure 4: *Associated anomalies with macrodactyly*

Table 2: *Involvement of the individual digits*

| Hand digits | Frequency | Foot digits | Frequency |
|---|---|---|---|
| Thumb | 6 | Big toe | 6 |
| Index finger | 9 | Second toe | 0 |
| Middle finger | 3 | Third toe | 0 |
| Ring finger | 2 | Fourth toe | 2 |
| Little finger | 0 | Fifth toe | 2 |

The term macrodactyly lipomatosa was first introduced by Feriz[4] in 1925. In 1967, Barsky[11] reviewed the literature and stated that there are two varieties of macrodactyly. The more common is static one, showing enlargement from birth and increase in size is proportional to the growth. The second type is the progressive one showing disproportionate growth of the affected digits and increase in size is faster than could be attributed to the normal growth spectrum.[2,4,11] Bailey[20] et al in 1997 made this diagnosis in three patients with macrodactyly who presented with enlargement of the planter aspect of the forefoot due to an overabundance of fibro fatty tissues.

The pathologic substrate of macrodactyly is hypertrophy of the bone with hamartomatous over growth of predominantly lipomatous or fibrous tissues.[2] Histopathologic findings suggest that, in macrodactyly of the foot, excessive proliferation of adipose tissue is the basic etiology, whereas in the hand, hypertrophy and tortuosity of the digital nerves are the predominant findings.[4,5,16] Lipomatous degeneration and disturbances of fetal circulation or local growth promoting / inhibiting factors have also been postulated as common causes of macrodactyly.[16,17]

Macrodactyly in the majority of patients is an isolated finding without other associated symptoms or systemic involvement.[1,2] There is a range of other pathologic conditions in which localized overgrowth may mimic the clinical pictures of macrodactyly.[2,21] These include neurofibromatosis, primary lymphatic disorders and vascular malformations like Klipple-Trenauny-Weber syndrome.[22-24] Several rare hereditary syndromes that possibly present as macrodactyly include Proteus syndrome, Bannayan syndrome, Maf-

fuci syndrome and Ollier disease.[1,2] In our study 13 patients were having associated anomalies of hemangioma, syndactyly, neurofibromas and hemartomas.

The clinical course of overgrowth in macrodactyly is important for prognostic and therapeutic reasons. In the attempt to make a diagnosis in cases of macrodactyly, one should be aware of the difficulty in classifying hamartomas with contribution from varying tissue elements. Enzinger[18] and Weiss emphasize conditions with prominent nerve involvement labeling them as fibrolipomatous hamartomas of nerve or, in cases with overgrowth of bone, neural fibrolipoma with macrodactyly. Boren[19] et al prefer the clinical designation nerve-territory-oriented macrodactyly. Kalen[15] et al stated that nerve enlargement was demonstrated in more than 90% of the hand cases and in 12% of the foot cases.

Diagnostic aids in cases of macrodactyly should include radiographs and computed tomography to reveal the extent of bone involvement.[23,25] Magnetic resonance imaging is recommended when the clinical features and plain films findings are indeterminate.[21] Angiography and lymphangiography should be performed if vascular or lymphatic changes respectively are clinically suspected.[1]

Timing and extent of therapeutic measures have to be considered very carefully to ensure the best functional outcome. The main surgical principle in treating this condition is to improve cosmetic appearance and preserve neurological function as far as possible.[7] Procedures may vary from simple excision of the mass to amputation.[22] Ray resection if possible, seems to give the best results.[10] Judicious and planned use of multiple debulking procedures, epiphyiodesis and ostetomies are advised to achieve the best functional and cosmetic results.[26] A localized recurrence rate of 33%-60% makes the management more demanding.[27]

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