

MACRODACTYLY

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Macroductyly is a non-hereditary congenital enlargement of a digit. It occurs due to a mutation in the PIK3CA pathway which controls normal cell growth. Barsky defined it as an increase in size of all the elements or structures within a digit or digits¹. Kelikian suggested that it should be called “Nerve territory oriented macroductyly” as the site of the enlargement of the digit coincides with that of the digital nerve².

In the current IFSSH Classification of Congenital Anomalies, the OMT (Oberg, Manske & Tonkin) classification, macroductyly is placed in the general category of *dysplasia*, within the subdivision of ‘hypertrophy’, with further delineation depending on whether the entire upper limb or a part thereof is involved.

PATHOGENESIS

It is an overgrowth condition that occurs due to a gain-of-function mutation in the PIK3CA pathway (Phosphatidylinositol-4,5-Bisphosphate 3-Kinase)^{3,4}. The PI3K/AKT/mTOR signalling pathway plays an essential role in regulating normal cell growth, metabolism and survival. Somatic mutations in this pathway can lead to cancer and a spectrum of overgrowth syndromes known as the PIK3CA-Related Overgrowth Spectrum (PROS). *PIK3CA* variants in PROS cause physiologically inappropriate activation of AKT and mTOR, and lead to

asymmetric overgrowth. This spectrum includes macrodactyly, CLOVES (Congenital Lipomatous Overgrowth, Vascular malformation, Epidermal nevi, Spinal/skeletal anomalies), hemimegalencephaly and others. As these are postzygotic mutations, only some cells carry the mutation while others do not.

CLASSIFICATION

1] Isolated or Syndromic Macrodactyly

Isolated type can be *true macrodactyly* or *pseudomacrodactyly* depending on the involvement of the bone. Pseudomacrodactyly is a soft tissue enlargement from vascular anomalies or tissue edema from constriction band syndrome (*fig 1*).



Figure 1: A case of digital enlargement due to vascular malformation.

Syndromes associated with macrodactyly are Congenital partial gigantism, Neurofibromatosis, Ollier's disease, Maffuci's syndrome, Klippel-Trenaunay-Weber syndrome, Congenital lymphoedema etc.

2] True macrodactyly can be of 2 types [Laurenzi]

i] *Static type/ Macrodactylia simplex congenital* – the enlargement is seen at birth and the enlargement is proportionate to the growth of the uninvolved digits.

ii] *Progressive type/Macrodystrophia lipomatosa progressiva* – it is not always seen at birth, but at about 2 years of age the involved fingers start growing much more rapidly than the normal fingers.

3] Clinically classification in to 4 types:

i] *Type I – Gigantism and Lipofibromatosis*

This nerve related type is the most common. The median nerve accounts for 85% of the enlargements and ulnar nerve for 15%. The involved peripheral nerve is usually normal up to the level of the distal forearm and enlarges distally causing compressive symptoms (fig 2).



Fig 2: Enlarged Median nerve in the palm.

The phalanges also show enlargement with metacarpal affection in severe cases. As the phalanges enlarge, the digit tends to curve. If two adjacent digits are affected, they curve towards each other. In our experience, we have noted that the fingers are deviated from one another due to the overgrowth on one side, probably due to factors associated with the influence of the common digital nerve supplying the fingers. As the child matures, the finger(s) gradually lose function partly due to irregular bony enlargement that affects joint function. Concealment and deliberate lack of use of the affected hand may also contribute.

ii] *Type II – Gigantism and Neurofibromatosis*

Gigantism of the finger, hand or the whole upper limb can occur in Neurofibromatosis. It is characterised by six or more café au lait spots, multiple neurofibromas and pedunculated cutaneous tumours. The affected nerve is large, tortuous and has nodular masses along its length. They differ from type I in that these are usually bilateral, show osteochondral masses arising from the bones of the hand and absent fatty infiltration between the nerve fasciculi.

iii] *Type III – Gigantism and Digital Hyperostosis*

This type is characterised by osteochondral masses from the phalangeal and metacarpal epiphysis (*fig 3*). These are identical to those seen in type I but without nerve involvement. The digital enlargement is asymmetrical.



Figure 3: Digital hyperostosis characterized by osteochondral masses.

iv] Type IV – Gigantism and Hemihypertrophy

Upton added this fourth type and said that it is difficult to categorise this type of hypertrophy. This is usually unilateral and can involve half the body along with the extremities (*fig 4*).



Figure 4: A case of Upper limb hypertrophy.

The hand and digits do not reach the size as seen in the other types. Massive hypertrophy of the thenar, hypothenar areas, forearm and arm can occur in this type.

INHERITANCE

There is no reported familial or chromosomal abnormality.

CLINICAL FEATURES

Macroductyly is one of the least common congenital hand differences; incidence being less than 0.9%⁵. It shows equal sex distribution. Multiple digit involvement occurs in 70% of the cases and the ones involved are always adjacent to each other. About 90% show unilateral involvement with the radial side being affected the most. The index finger is said to be the most frequently involved followed by the middle finger and thumb.

The enlargement may be confined to the digit or may be seen extended to the palm, forearm or arm. Angulation deformity of the digit may be noted. At times limitation of motion at the distal interphalangeal joint is seen. In severe cases, two point discrimination will reveal sensory disturbance. Motor deficit is rarely seen. Carpal tunnel syndrome can occur due to the enlarged median nerve. Associated anomalies and features of related syndromes should be looked for. The most common association is syndactyly seen in around 10% cases of macroductyly; other associations include polyductyly, atrial septal defect, lipoma, haemangioma, Goldenhar's syndrome and connective tissue naevus.

Radiographically the bone growth can be evaluated. In mild cases, the phalangeal bones are seen involved. In advanced cases, metacarpal bones are seen enlarged. The joints may be surrounded by osteophytes and in hyperostotic type, by osteocartilaginous masses. These cause restriction of joint motion.

Angiographic studies have shown that the enlarged digit has only one digital artery⁶. The macrodactylous tissue is usually hypoperfused and for this reason macrodactyly patients have a higher incidence of flap and wound healing problems.

PATHOLOGY

The proliferative adipose tissue resembles adult subcutaneous fat. The fat lobules are large, dark and difficult to remove. The digital nerve, median and ulnar nerves are bigger in size and the cross section shows increase in fat and fibrous tissue around the nerve. The digital artery wall is normal with normal thickness. The blood vessels, tendons and their sheaths are not usually involved. Hence, as the hand grows, the blood supply does not keep pace and vascular insufficiency is frequent. The phalangeal medullary canals are always enlarged and fatty marrow is seen. The metacarpals can get involved rarely.

PERI- OPERATIVE COUNSELLING

Parents desire five normal digits in the affected hand after surgery. We should stress the fact that the operative digits will not function like their counterparts or appear normal and that with surgery we hope to improve the overall function of the hand. In patients with progressive type of macrodactyly, it is important to inform the parents that the child needs multiple stages of debulking and, possibly, amputation. Maintaining a good rapport with the parents and children improves the patient compliance and regular follow up and subsequent surgeries become possible. If amputation is planned, a psychology team should be involved. It is important to counsel the stakeholders that even after amputation, the adjacent parts may continue to overgrow and may require surgery. At some centres, photo editing software has been employed to counsel children and parents about how the hand might

look like after amputation. Showing pictures of prior operated children or setting up parental and children support groups can also help in convincing parents about the surgical plan and manage their expectations.

TREATMENT

Patients seek help for the functional impairment, psychosocial effects, cosmesis or features of carpal tunnel syndrome.

The growth rate of the affected area should be observed for a few years. If it is of progressive type, the child needs multiple procedures and growth inhibition by epiphysiodesis or epiphyseal resection when the length of the digit becomes the same as the corresponding digit in the same sex parent. In older children and adults, debulking should be done until the finger is as normal looking as possible.

The operative procedures available are debulking, nerve resection, epiphysiodesis, angulation osteotomy, arthrodesis and amputation. Ezaki et al have devised a treatment algorithm for macrodactyly⁷. If the finger is wider but shorter than the parent's digit, soft tissue debulking is advised. If the macrodactylous digit approaches the length of the adult digit or becomes more than the parent size, debulking and epiphysiodesis to arrest the longitudinal growth is done. If the overgrown digit is non-functional, amputation is advised.

1) DEBULKING:

For hypertrophy limited to the volar aspect of the digits, the soft tissue is pinched to assess the ability to close after tissue reduction. An elliptical volar midline incision can help in reducing the bulk, both skin and soft tissue (*fig 5*). This approach preserves the digital nerve.



Fig 5: Debulking and removal of lipomatous tissue.

For global overgrowth, debulking with excision of the digital nerve is done via a midlateral zigzag incision made on the more involved side of the digit. The digital artery should be preserved at any cost and hence debulking on the other side is not done simultaneously for fear of losing the viability and sensibility of the digit. Debulking is usually done in multiple stages and the interval should be at least 3 months between sessions.

Excision of the involved digital nerve may slow down the overgrowth as believed by Tsuge. The sensory deficit is not disabling because of two reasons; one, the overlying skin is also being excised and two, neuroplasticity in children allows the digital nerve on the other side of the digit to innervate the affected side. Decompression of the nerve may alleviate some of the symptoms. Hence release of the Carpal tunnel and Guyon's canal are recommended. While debulking, a sliver of the nail bed and nail plate along with a part of the distal phalanx can

be excised by elevating the nail fold, to give a better aesthetic appearance.

2) EPIPHYSIODESIS:

To arrest the longitudinal growth epiphysiodesis can be done via a midlateral incision. The epiphysis can be destroyed by dental picks, dental elevators, small drill bits, 18-gauge needles, curettes, electrocautery, stapling or excision. This can be combined with soft tissue debulking.

3) RECESSION OSTEOTOMY:

If the growth arrest is not adequate, shortening osteotomies are needed. Though many procedures have been described in literature, the methods devised by Barsky and Tsuge have been extensively followed. Barsky's method is to ablate the distal half of the middle phalanx and proximal half of the distal phalanx and fuse it with K wires after the fingertip is telescoped proximally. Tsuge has modified the technique wherein a part of the distal phalanx and middle phalanx are excised and removed. The redundant skin can be excised after 6 weeks as second stage. However, these procedures lead to stiffness and contractures.

4) ANGULATION OSTEOTOMY:

Corrective osteotomy is done for curved digits. A midlateral incision is made on the convex side of the digit and a wedge of bone from the metaphysis is removed. If further correction is needed, resection at the diaphysis can be done (*fig 6,7*).

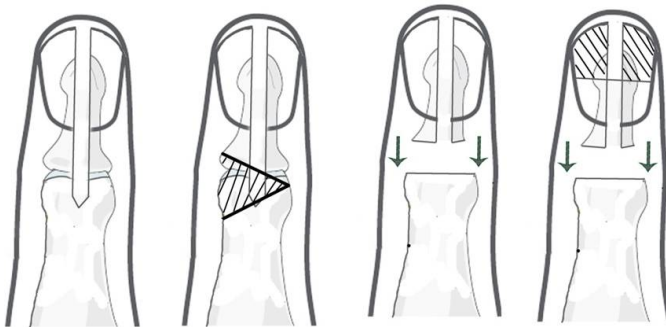


Fig 7: In a case of macrodactyly with enlarged soft tissue as well as phalanx with angulation deformity, a combination of the bony procedures can be done.

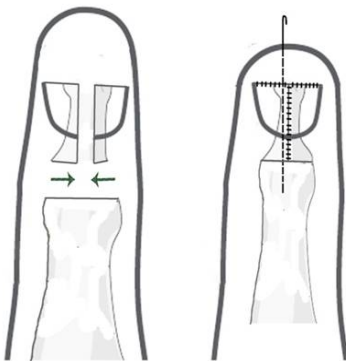


Fig 8: K-wire fixation of the bony fragments

The bone is then fixed with K wire which will be retained for 4-6 weeks depending on the bone union and age of the patient. This can be done along with physseal arrest.

5) AMPUTATION:

The main indications for amputation are overgrown digits with stiffness and limited function or those that interfere with the function of the other normal digits (*fig 8,9*). Ray amputation is advised even in normal metacarpal to reduce the central width of the hand. The

metacarpal can be disarticulated from the carpometacarpal joint (CMC) joint. If the resected ray contains the insertion of one of the wrist tendons, the base is preserved or the insertion is detached and reinserted to an adjacent metacarpal.



Fig 9: Stiff macrodactylous left index and middle fingers.



Fig 10: Ray amputation of the affected fingers.

6) MACRODACTYLY OF THE THUMB:

Ray resection of the affected thumb is not done without microvascular free toe transfer or pollicisation. The other options are multiple debulking, shortening, arthrodesis and epiphysiodesis. Millesi's method of reducing the thumb size can also be attempted. The distal half of the nail and phalanx, middle third of the distal phalanx and proximal phalanx are removed in this method. The remaining bones are fixed with K wires (fig.10).

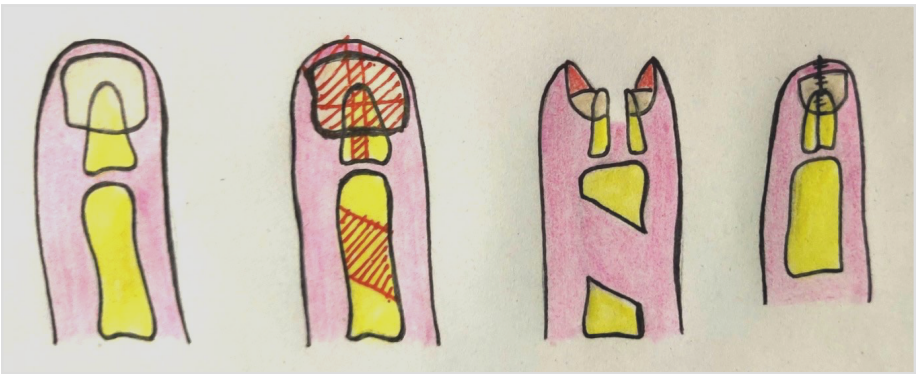


Fig 11: Millesi's method for thumb macrodactyly: Removal of distal half of the nail and phalanx and middle third of distal and middle phalanges with K-wire fixation of the bony fragments.

COPING WITH MACRODACTYLY

Hands are one of the most noticed parts of the body, second only to the face. Over half of the children with congenital hand differences face stress and half of them have stress unrecognised by parents⁸. Children with macrodactyly often fail to understand what they have as we are unable to give a good explanation or treatment. Clinicians can help parents by informing about the potential stressors and screening

the children for anxiety and anger and provide them supportive care. It is rightly said that macrodactyly dwarfs the giants in hand surgery.

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