

# Macrodystrophia Lipomatosa of the Foot: A Rare Case

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**Abstract:** *Macrodystrophia Lipomatosa is a rare type of non hereditary congenital localized or generalized gigantism of limb or digits. It may manifest from infancy to late adulthood. There is progressive enlargement due to hypertrophy of mesenchymal elements giving rise to disproportionate increase in amount of fibrofatty tissue. These patients come to hospital usually because of cosmetic reasons or symptoms due to secondary degenerative arthropathies or neurovascular compression caused by large osteophytes. Here we present a case of 18 years male affected by this rare congenital anomaly involving left second toe.*

**Keywords:** Macrodystrophia lipomatosa, second toe, non hereditary

## 1. Introduction

Macrodystrophia Lipomatosa is a sporadic congenital developmental anomaly which is non-hereditary. It is characterized by progressive enlargement of fibroadipose tissue due to disproportionate mesenchymal proliferation. It can be localised or generalized, commonly involving distal limbs or digits.<sup>(1-4)</sup> Lower limbs are affected more as compared to upper limbs. Also, second and third digits are most commonly involved. This condition usually presents at birth or during neonatal period.<sup>(2)</sup> There is slight male preponderance<sup>(1,2)</sup>. The incidence of this condition is yet to be known due to its rarity. Lipomatous degeneration and neurofibromatosis have been hypothesized to be involved in the etiology of Macrodystrophia Lipomatosa<sup>(1)</sup>.

Here we aim to describe this rare condition with its characteristic histopathological features.

## 2. Case Report

An 18 year old male patient presented to the hospital with complaints of swelling of 2<sup>nd</sup> toe of left foot since birth.(Fig. 1)



**Figure 1:** Disproportionately enlarged 2<sup>nd</sup> toe of left foot

The swelling increased in size gradually with no other associated complaints. Rest all other systemic examination was within normal limits. Radio imaging studies were done and USG revealed a heterogenous hypoechoic soft tissue component with few ill defined hypoechoic areas showing internal echoes within. The soft tissue component showed no abnormal vascularity.

Histopathological features:

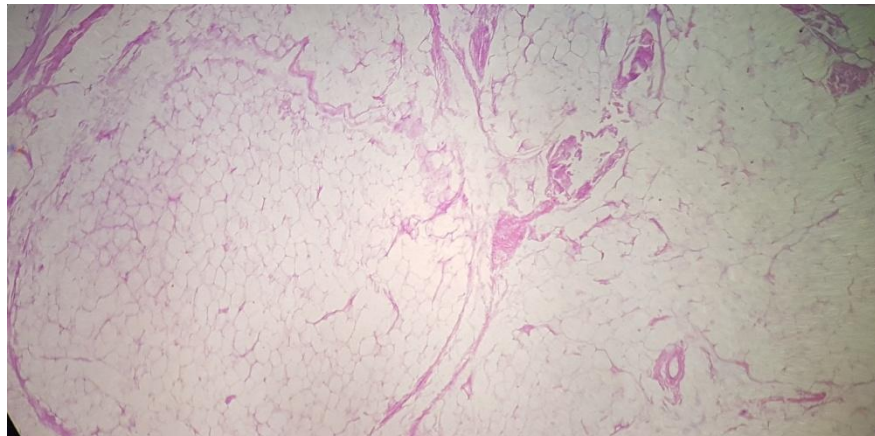
The partial amputation specimen of the second toe of left foot was received in multiple pieces. The toe appeared enlarged measuring 6 x 3.5 x 3.4 cm. The nail and covering skin of the toe appeared unremarkable.(Fig. 2). On serial cut sectioning ventral enlarged part of the toe showed homogenous greyish yellow appearance.



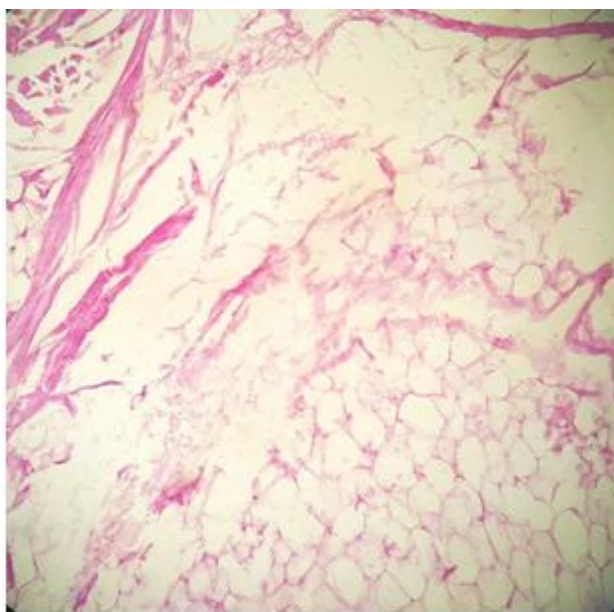
**Figure 2:** Partial amputation specimen of enlarged 2<sup>nd</sup> toe received in pieces

Microscopic examination of this part revealed increased mesenchymal elements, particularly adipose tissue arranged in nest, lobules, fibrous tissue and few proliferating vascular channels. (Fig.3, 4 & 5) So, diagnosis of Macrodystrophia

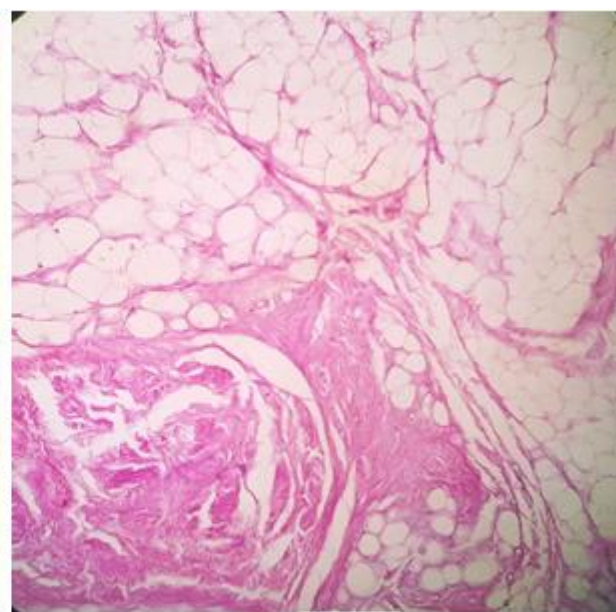
Lipomatosa was rendered. Post operative follow up of the patient is uneventful.



(3)



(4)



(5)

**Figure 3, 4, 5:** Microphotograph showing increased mesenchymal elements, particularly adipose tissue arranged in nest, lobules, fibrous tissue and few proliferating vascular channels. (Fig 3 -100x, Fig 4,5- 400x)

### 3. Discussion

Macrodistrophia Lipomatosa is hamartomatous enlargement of the soft tissue components leading to congenital gigantism of a limb, either localised or generalized. This non-hereditary condition is characterized by progressive hypertrophy of mesenchymal elements with a disproportionate increase in amount of adipose and fibrous tissue<sup>(6)</sup>. Sometimes there may be bilateral involvement of the limbs<sup>(7)</sup>. Other than limbs, abdominal wall involvement has also been reported<sup>(7,8)</sup>. The involvement is found to develop in a specific sclerotome region of the body<sup>(8)</sup>.

The commonly affected sites are lateral aspect of upper limb along the median nerve distribution and medial aspect of lower limb along plantar nerve distribution. Distal limb is more usually affected<sup>(8,9)</sup>. The affected region progressively grows till puberty and then the growth reaches a plateau<sup>(10)</sup>.

Feriz coined the term Macrodistrophia Lipomatosa in 1925. Barsky described local gigantism in detail and differentiated two forms of it as static and progressive. Progressive form correlates with what was described by Feriz<sup>(11)</sup>. Various other congenital anomalies like syndactyly, brachydactyly, polydactyly and clinodactyly may be associated with Macrodistrophia Lipomatosa<sup>(6)</sup>. However our case did not reveal such anomalies. Progressive macrodactyly is more common than the static type where growth of the affected part is faster than the rest of the body. Patients present because of functional problems like difficulty in gait or grasping or in wearing shoes. Surgery is usually done for cosmetic reasons rather than for mechanical problems. One case presented as progressive form of Macrodistrophia Lipomatosa of the foot.

Various radioimaging modalities such as plain radiographs, ultrasonography, MRI and CT scan have definite role in the evaluation of Macrodistrophia Lipomatosa. Soft tissue and skeletal muscle hypertrophy with translucent fat and soft tissue are seen on plain radiograph. The phalanges appear like mushroom because of splaying and endosteal and periosteal bone deposition more commonly in distal phalanx. There can be secondary osteoarthritic changes like subchondral bone cyst, decreased joint space and osteophytes during second decade of life<sup>(12)</sup>. Proliferation of fat along the nerve territory is seen on CT and Ultrasonography<sup>(13,14)</sup>. In our case proliferation of fat was observed on ultrasonography. MRI demonstrates excess fibrofatty tissue in subcutaneous plane without any obvious capsule<sup>(15)</sup>. Soler et. al. proposed that MRI should be used as the diagnostic method of choice for Macrodistrophia Lipomatosa<sup>(16)</sup>.

Microscopically, significant increase in all mesenchymal elements is seen<sup>(15)</sup>. Large amount of adipocytes with interspersed fibrous tissue are usually found<sup>(17)</sup>. The etiopathogenesis is not clear, but proposed hypotheses include fatty degeneration, altered fetal circulation, segmentation problems and changes in growth inhibiting factors<sup>(18)</sup>.

Certain conditions presenting with localised limb hypertrophy should be considered in differential diagnosis of

Macrodistrophia Lipomatosa. These differentials include neurofibromatosis type 1, fibrolipomatous hamartoma (FLH), hemangiomas, lymphangiomas, Mafucci syndrome, Proteus syndrome and Ollier disease. All of these differential diagnoses have a positive family history and have characteristic cutaneous, neurological and systemic manifestations.

The mainstay of treatment is surgical excision which improves cosmetic appearance and helps in maintaining neurological function. Partial amputation and function preserving debulking surgeries give superior results.

### 4. Conclusion

Macrodistrophia Lipomatosa is a progressive non-hereditary hamartomatous condition with proliferation of mesenchymal elements, predominantly adipose and fibrous tissue causing local gigantism. Detailed clinical examination, radioimaging studies and characteristic histological features are crucial for definitive diagnosis.

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