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Case Report

# Lipodystrophic Hamartoma of a Nerve Associated with Macrodystrophia Lipomatosa: A Rare Case Report

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## Abstract

Macrodystrophia lipomatosa is a rare congenital anomaly characterized by abnormal proliferation of fibrofatty tissue within a nerve territory, most commonly associated with a lipofibromatous hamartoma of the median nerve. We report the case of a 16-year-old patient who had surgery in childhood for macrodactyly of the first three fingers and now presents with recurrence. MRI revealed fatty hypertrophy infiltrating the soft tissues of the hand and enlargement of the median nerve with a "spaghetti-like" appearance, suggestive of a lipofibromatous hamartoma. Although benign, this entity presents a therapeutic challenge. Treatment is often symptomatic, and aesthetic amputation may be considered in severe cases.

***Keywords:*** *Median Nerve; Peripheral Nerve Tumor; MRI; Macrodactyly; Peripheral Nerve Tumor; Imaging Findings; Surgical Management*

## Abbreviations

MDL: Macrodystrophia Lipomatosa; LFH: Lipofibromatous Hamartoma; MRI: Magnetic Resonance Imaging

## Introduction

Macrodystrophia lipomatosa is a rare congenital condition characterized by focal overgrowth of mesenchymal elements, primarily fibroadipose tissue [1]. Lipofibromatous hamartoma of the nerve is a benign peripheral nerve tumor resulting from abnormal proliferation of fibroadipose tissue within the epineurium, leading to nerve enlargement and fascicular separation [2]. The association between these two entities is common, occurring in approximately 27% to 66% of cases [3]. There is a predilection for the upper limb, particularly the median nerve [4]. Magnetic Resonance Imaging (MRI) is the diagnostic modality of choice, allowing both confirmation and exclusion of differential diagnoses.

## Case Report

A 16-year-old patient with a history of congenital macrodactyly of the first three fingers, previously operated on for phalangeal amputation and soft tissue debulking, presented with a recurrence. the patient reported hand paresthesia associated with macrodactyly and aesthetic discomfort. Physical examination revealed no motor deficit. Standard radiography showed enlargement of the first three

fingers with heterogeneous bone texture, areas of cortical thinning, and amputation stigmata of the phalanges in the first two digits. MRI of the hand revealed hypertrophy of fatty tissue in the palmar aspect, infiltrating around the musculoskeletal and neural structures. This tissue appeared hyperintense on T1- and T2-weighted sequences, suppressed on fat-saturation sequences, and showed no enhancement after contrast administration. The median nerve appeared enlarged, with a serpentine configuration of its fascicles giving a characteristic "spaghetti-like" appearance, leading to the diagnosis of macrodystrophia lipomatosa associated with a lipofibromatous hamartoma of the median nerve.

The patient was referred to the plastic surgery department to discuss surgical management in view of the aesthetic discomfort reported by the patient.

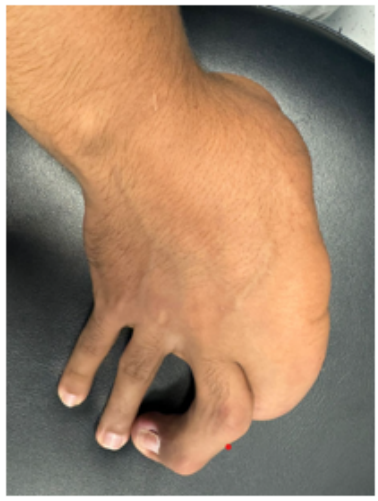

***Figure 1:*** *Clinical image showing macrodactyly with a deformed appearance of the first three fingers.*

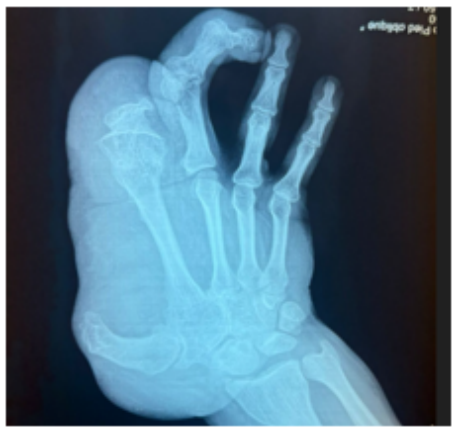

***Figure 2:*** *Standard frontal X-ray showing widening of the first three fingers with heterogeneous bone texture, cortical thinning in some areas, and signs of amputation of the phalanges of the first two fingers.*

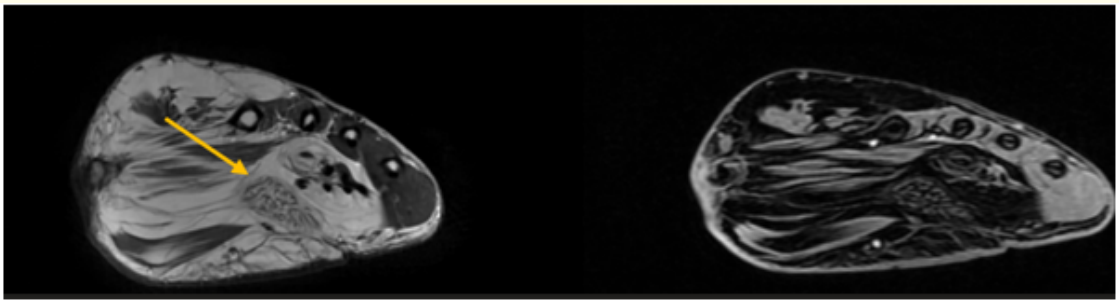

***Figure 3:*** *Axial MRI of the hand in T2 and T2 STIR sequences showing enlargement of the palmar fat pad infiltrating the muscles and nerves, with enlargement of the median nerve presenting serpentine fascicles, producing the characteristic "spaghetti-like" appearance (Yellow arrow).*

## Discussion

Lipofibromatous hamartoma of the nerve, also known as neural fibrolipoma, perineural lipoma, or intraneural lipoma, is a benign tumor characterized by infiltration of the epineurium and perineurium of unknown origin [5]. It predominantly affects the upper limb (78 to 96%), with a preference for the median nerve.

Clinically, the patient presents with a gradually enlarging soft tissue mass, typically located on the palmar side or anterior forearm, usually in individuals under the age of 30.

The diagnosis of lipofibromatous hamartoma of the median nerve is generally based on magnetic resonance imaging (MRI) or ultrasound. The "spaghetti-like" appearance on imaging suggests an enlarged nerve (containing coaxial axonal bundles of 2 to 3 mm in diameter, cable-shaped, surrounded by epineural fibrous tissue), which is pathognomonic [4].

Dystrophic macrodactyly is a congenital, unilateral, and localized macrodactyly. It is characterized by the excessive growth of all mesenchymal tissues within one or more nerve territories. Its etiopathogenesis is unknown, with a preferential involvement of the second or third toe or finger. The deformity appears rapidly after birth and ceases progression at puberty [6].

X-ray may show finger enlargement, as seen in our patient, with cortical thickening and hypertrophy of surrounding soft tissues.

MRI is the key examination for diagnosis. It reveals fat accumulation in the subcutaneous tissues, without a discernible capsule. Fibrous strands may be seen within this fatty accumulation, along with cortical thickening, bony hypertrophy, and bony outgrowths arising from the underlying bone. Neural thickening may also be visualized [7].

There is a frequent association between dystrophic macrodactyly and lipofibromatous hamartoma of the nerve.

The differential diagnoses include neurofibromatosis type 1 (plexiform neurofibroma), vascular malformations (Sometimes associated with Klippel-Trénaunay-Weber syndrome), and Proteus syndrome. In our patient, the absence of a family history, the lack of café-au-lait spots, and the presence of the characteristic "spaghetti-like" sign on MRI allowed the diagnosis to be established without the need for tissue biopsy.

There is no definitive treatment other than nerve resection, which is controversial due to significant sensory and motor sequelae. A symptomatic approach is recommended in cases of carpal tunnel syndrome. Aesthetic amputation may be considered in cases of macrodactyly [8].

Surgical management appears to be more appropriate than non-surgical treatment for MDL. The most commonly performed surgical procedures include amputation followed by debulking or volume reduction, which are usually carried out as single-stage operations. Multi-stage surgical approaches are typically reserved for more complex cases requiring prolonged follow-up and may involve epiphysiodesis, osteotomy, and carpal tunnel release.

Epiphysiodesis and osteotomy are performed with the aim of halting the growth of the abnormal limb and correcting deformities resulting from disproportionate growth. Carpal tunnel release is performed only when the patient complains of pain in the affected region [9].

Reported complications include sensory loss, under correction, and skin necrosis. Sensory loss may result from extensive lesions requiring aggressive surgical intervention. The risk of complications following extensive surgery is estimated to be approximately 30 - 50% for nerve injury and 33 - 60% for recurrence of macrodystrophia lipomatosa (MDL) [10]. In our patient, amputation of the first three phalanges was performed, followed by recurrence during adolescence. Given the aesthetic discomfort reported by the patient, an additional surgical intervention was proposed.

## Conclusion

Lipofibromatous hamartoma of the nerve, often associated with dystrophic macrodactyly, is a rare but distinctive benign condition primarily affecting young individuals. Diagnosis relies heavily on MRI, which reveals characteristic features such as nerve enlargement with fatty infiltration. While conservative management may be appropriate in some cases, treatment options remain limited and controversial, especially due to the functional risks of nerve resection. In severe cases, especially those with macrodactyly or compressive symptoms, surgical intervention-including decompression or aesthetic amputation-may be considered on a case-by-case basis.

## Informed Consent

Written informed consent was obtained from the parents of patients for the publication of this case report.

## Ethical Approval

Our institution does not require ethical approval for reporting individual cases or case series.

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