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# MACRODYSTROPHIA LIPOMATOSA: CASE REPORT OF UPPER LIMB INVOLVEMENT

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**Abstract:** Macrodystrophia lipomatosa is a rare, non-hereditary congenital condition that manifests as a form of localized gigantism primarily affecting the extremities. This condition is characterized by a permanent and progressive proliferation of the mesenchymal component and disproportionate growth of fibroadipose tissue. We present the case of a patient diagnosed with macrodystrophia lipomatosa, involving the entire left upper limb, with minimal compromise of the thumb and fifth finger. This case report is of particular interest due to the low frequency of this pathology in the upper limb, especially in this instance, which presented with intense pain. We achieved very good symptomatic results after treatment with liposuction, a procedure associated with low morbidity. This report contributes to the limited literature on macrodystrophia lipomatosa and highlights the potential for effective therapeutic strategies for similar cases.

## INTRODUCTION

Macrodystrophia lipomatosa (MDL) is a rare, non-hereditary congenital disorder that manifests as a form of localized gigantism, potentially affecting the fingers, hands, feet or even the entire upper or lower limb. [1-4] It is characterized by a permanent and progressive proliferation of mesenchymal component and disproportionate growth of fibroadipose tissue. [2]

The term MDL was first coined by Feriz in 1925 to describe a case of gigantism located in the lower limb. [3,5-8] Later, Golding expanded the terminology to include cases with upper limb involvement. [5]

We report the case of a patient diagnosed with MDL, with involvement of the entire left upper limb, ipsilateral shoulder girdle, minimally affecting the thumb and fifth finger. We present the clinical presentation, paraclinical evaluation, as well as the treatment implemented and its results.

## CASE REPORT

We present the case of a 38-year-old female patient who is unemployed, a mother of two children, and is self-sufficient and independent in performing activities of daily living. She has a medical history of hypertension, HIV, and is a former smoker, with no other significant pathologies.

Since birth, the patient has exhibited disproportionate growth in the size of the second and third fingers of the left hand, which led to surgery at the age of 2. This surgery consisted in the resection of adipose tissue and the amputation of these digits. Throughout her development, there was a further increase in adipose tissue in the upper limb, resulting in a second intervention at the age of 13. On this occasion, additional soft tissue resection was performed in the palm, arm and the left scapular region. Unfortunately, we do not have records or additional information regarding these procedures

After several years, the patient presented to our clinic motivated by the presence of pain. This pain has been progressive over the months, reaching an intensity of 10, the maximum on the numerical scale, localized in the posterior scapular region and accompanied by a sensation of heaviness throughout the left upper limb. It is continuous, intensifies with movement of the limb, and prevents left lateral decubitus. The discomfort is of such magnitude that it triggers insomnia, suicidal ideation, and request for amputation of the limb, which necessitated hospitalization for comprehensive evaluation and treatment.

During hospitalization, after being assessed by the anesthesiology department, it was decided to place a paravertebral catheter with bupivacaine infusion associated with oral methadone. Additionally, she was evaluated by the medical psychology and psychiatry team, and antidepressant treatment was initiated.

Once the acute episode was resolved, a consultation was held with the plastic surgery team after exhausting all medical treatment options. When we contacted the patient, she emphasized the presence of pain, stating that she does not use the upper limb in daily activities. At the time of the initial evaluation, her symptoms were pharmacologically controlled with tramadol and acetaminophen.

Physical examination of the patient revealed a notable asymmetry of the upper limbs, evidenced by an increase in size of the entire left limb, from the shoulder to the hand, primarily sparing the first and fifth fingers. In the hand, there is an amputation of the second and third rays (Fig.1). Scars corresponding to the surgical approaches in the digital region, as well as in the volar sector of the wrist, were identified. Additionally, scars were present in the ipsilateral scapular region, where tissue resection is presumed to have been performed. No alteration in skin color was observed, and the consistency of the tissue was soft to the touch. The patient has limitation of the range of motion in the shoulder, elbow and wrist, with pain during maneuvers. Sensitivity in the upper limb is preserved. The asymmetry was quantified by circometry, comparing it to the contralateral limb (see Table 1).

Regarding the paraclinical studies performed, a plain X-ray of the hand was obtained (Fig.2), and an arterial and venous Doppler ultrasound of the left upper limb was also conducted, which showed no significant alterations, except for an enlarged and tortuous cephalic vein. Furthermore, a magnetic resonance imaging (MRI) of the upper limb, including the shoulder and scapular region, was performed. This study revealed an increase in soft tissue thickness in the scapular region, arm, and forearm, with fatty atrophy of the muscular structures in the scapular region, except for the supraspinatus, infraspinatus and deltoid muscles. In the arm and forearm, muscle atrophy predominated in the posterior compartment. No alterations in bone signal intensity were identified (Fig. 3).

The patient's medical history, which reveals no related family history, along with the physical examination showing asymmetric and disproportionate overgrowth of the upper limb since birth, and the MRI demonstrating an increase soft tissue enlargement with infiltration of adipose tissue in the subcutaneous cellular tissue and muscle structures, lead to the diagnosis of MDL.

Following evaluation by the plastic surgery team, debulking by liposuction was proposed as initial treatment, with the intention of assessing the evolution and considering a potential subsequent tissue resection (Fig. 4).

The procedure was performed without complications under general anesthesia, infiltrating Klein's solution in the areas to be addressed at the level of the shoulder and left arm, followed by traditional suction-assisted liposuction. Cannulas of 4 and 5mm were used, resulting in the removal of 1.600ml. After a brief postoperative monitoring period, the patient was discharged without complications. In the postoperative period, she experienced recovery without significant discomfort, using minor analgesics (acetaminophen) during the first few days, and subsequently did not require regular analgesic consumption, resulting in a significant improvement in her previous symptoms.

Figure 1. Images of the initial clinical assessment, both anterior and posterior views. Note the asymmetry of the entire left upper limb compared to the contralateral side.

| | Left upper limb | Right upper limb |
|---|---|---|
| **Palm of hand** | 29 | 18 |
| **Fist** | 21.5 | 15.5 |
| **Half forearm** | 32.5 | 24 |
| **Elbow** | 44 | 26 |
| **Half arm** | 52 | 30 |
| **Proximal third of arm** | 58.5 | 35.5 |

Table 1. Circumference measurements of both upper limbs, recorded in centimeters.

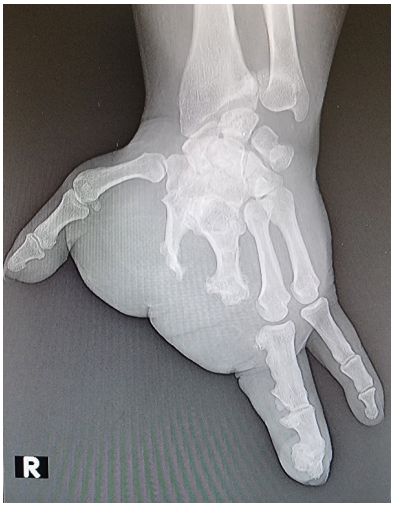


Figure 2. Plain X-ray of the left hand. Note the bony structure of the first and fifth rays without alterations, as well as the fourth metacarpal. In contrast, the fourth digit shows alterations in the 3 phalanges. Additionally, there are alterations observed at the level of the first and second metacarpals, along with amputation of the remaining portions of both rays.

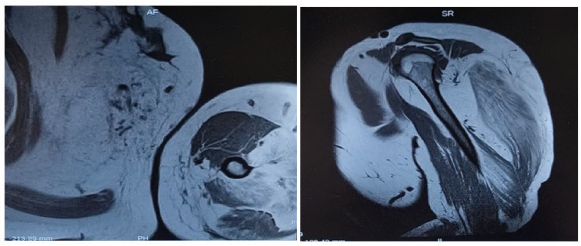

Figure 3. MRI of the shoulder in T1 axial view on the left and right sagittal view on the right. Note the significant increase in soft tissue and fat infiltration.

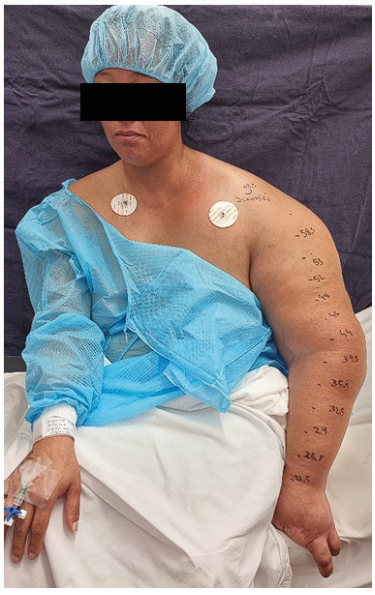

Figure 4. Preoperative marking of the patient and upper limb circometry.

## DISCUSSION

MDL is characterized by progressive growth of mesenchymal tissues, which includes adipose tissue, soft tissue and bone. Although the incidence of this pathology is unknown, historical data indicate that, over a period of 65 years since 1950, only 108 cases have been documented, highlighting its rarity. [2] No significant difference in the frequency of involvement based on sex has been observed. Generally, it presents unilaterally, as in this case, although there is published literature on patients with bilateral involvement. [1,7]

Distal involvement predominates in the limbs, with the lower limb being affected more frequently than the upper limb, in nearly 60%


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of cases. In turn, the combination of second and third fingers affected is the most frequent in both hand and foot. [4] In the case of our patient, the involvement is restricted to the upper limb and shoulder girdle. The history of digital amputation, along with the bone involvement observed in the X-rays, aligns with existing statistics, supporting a more frequent involvement of second and third fingers. Generally, the digital enlargement associated with this pathology ceases at puberty. The overgrowth primarily affects the volar aspect. [1]

In 1967, Barsky distinguished between two forms of localized gigantism based on the growth pattern: static and progressive. [1,6,8] The latter is the less frequent and presents with a disproportionate growth of one area compared to the rest of the body. Despite this difference between the two, it has not been discussed in case reports whether they correspond to the same pathologic entity. [7,8]

The complete etiopathogenesis of MDL remains unknown; however, there are several hypotheses. It has been proposed as a variant of neurofibromatosis, as well as a lipomatous degeneration, fetal circulation alteration, trophic influence of a tumefied nerve, intrauterine alterations such as disturbances in growth-limiting factors or errors in segmentation. [1,8]

Recent research has established an association between certain types of segmental overgrowth and mutations in the PIK3CA gene. However, the absence of detection of this mutation does not rule out the clinical diagnosis in patients with high suspicion of MDL. [7] The difficulty in distinguishing between these entities hinders progress in understanding the underlying etiopathogenesis.

It is important to distinguish MDL from other conditions of localized gigantism. The main differential diagnoses include neurofibromatosis type 1, fibrolipomatous hamartoma, lymphangiomatosis, hemangiomatosis, Klippel-Trenaunay-Weber syndrome, Maffucci syndrome, Ollier's disease and Proteus syndrome. However, unlike MDL, all these conditions have a family history and are characterized by cutaneous or systemic manifestations. [1,3] All these elements are absent in the clinical case presented.

MDL may be associated with other congenital anomalies such as syndactyly, polydactyly, brachydactyly or clinodactyly. [1,7] It may rarely be associated with intestinal or subcutaneous lipomas, pulmonary cysts, calvarial anomalies or pigmented nevi. [3,5,6]

We do not have clinical or paraclinical evidence of the presence of any of these alterations in our patient's case.

Clinically, at the upper limb it generally presents as a progressive growing mass in the fingers and wrist, potentially extending to the forearm. [2] Many times, the initial consultation is motivated by aesthetic reasons; in other cases, mechanical problems may arise secondary to joint degeneration or neurovascular compressions due to large osteophytes. [1,3] Individuals are usually asymptomatic except for deformity and its aesthetic impact. [5] It is not common for clinical presentations to include pain of severe intensity, as in the case we report in this work.

In MDL, the involvement of areas can be observed according to the distribution of a peripheral nerve. In the hand, the most common is the compromise of the median nerve territory, while in the foot it is the involvement of the plantar nerve. [3]

Nerve involvement often influences clinical presentation; frequent involvement of the median nerve in the upper limb may manifest as carpal tunnel syndrome. In contrast, distribution affecting the ulnar nerve territory is sporadic. [9] In the case of our patient, scars are observed on the volar aspect of the wrist, suggesting that she may have undergone surgical release of the median nerve in childhood.

Among the most useful paraclinical studies are plain X-rays (Rx), Doppler ultrasound, MRI and biopsy with histological analysis. X-ray may show elongation and widening of phalanges with soft tissue hypertrophy particularly in the volar sector. Doppler ultrasound can reveal an increase in fat in the subcutaneous tissue and throughout the entire nerve territory. [3] MRI typically shows a proliferation of unencapsulated fibroadipose tissue around the affected fingers, muscles, subcutaneous cellular tissue and nerve sheath. Bone hypertrophy may be observed with cortical thickening in the affected area which can lead to exostosis, as well as widening at the ends of the bones with the classic mushroom appearance. [1,6]

While the gold standard for diagnosis is biopsy for histopathologic study, many authors agree that the clinical presentation, lack of family history, and MRI often lead to an accurate diagnosis without the need for invasive techniques. [4,7]

In cases where a biopsy is performed, the most notable histopathological finding is the increase of adipose tissue dispersed in a fine network of fibrous tissue, involving bone marrow, periosteum, muscles, nerve sheaths and subcutaneous tissues. [1]

In the presented case, the X-ray reveals characteristic bone alterations in the hand, while the MRI shows an increase in soft tissue with abundant adipose tissue in the scapular region, arm and forearm with muscular infiltration by adipose tissue. Given the clear clinical presentation, supported by imaging, a biopsy was not considered necessary, although further histopathological study is not ruled out in case a second surgical procedure is required.

The treatment of this condition should be individualized according to the form of presentation and the patient's expectations. [4] In cases with localized disease, volume reduction procedures or even amputation of the affected ray may be more appropriate to achieve aesthetic improvement without functional detriment. [1] Multiple volume reduction procedures, epiphysiodesis and even osteotomies have been described in severe cases. Among the volume reduction procedures, we have liposuction, the technique we employ in our patient, while another option is tissue resections. Generally, these procedures do not typically cause nerve damage and are considered the treatment of choice; however, they are associated with a high recurrence rate of 33 to 60% following surgical treatment. [1,3,5,8]

The proposed treatment through liposuction has the advantages of being a minimally invasive procedure and allows for the reduction of part of the excess pathological adipose component that is so characteristic of this condition. Additionally, whether due to the reduction in volume, albeit limited, or mechanisms that cannot be established in this study, it has resulted in a remarkable symptomatic improvement for the patient. Therefore, we consider it may be useful as a first surgical approach in similar cases.

## CONCLUSIONS

Even though 100 years have passed since the first report on MDL, numerous questions about this pathology persist. Given its infrequency, each new case can contribute to its understanding and the search for better therapeutic options for patients.

This work is of particular interest, as it affects almost the entire upper limb, a highly unusual occurrence, as well as its presentation with associated intense pain.

It is important to highlight that, although the volume reduction achieved through liposuction treatment was limited in relation to the degree of limb deformity, excellent results have been observed in symptom control. Therefore, we consider it a valuable therapeutic tool to be considered in analogous situations.

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