



CASE REPORT

Madelung's disease: clinical findings and patient management

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Abstract

Multiple Symmetric Lipomatosis (MSL), also known as Madelung's Disease, is a rare condition characterized by the presence of diffuse, symmetrical, unencapsulated lipomas commonly located on the face, neck, and in the proximal region of the upper limbs. Lesions are generally asymptomatic, grow slowly and progressively, but can lead to cervical mobility limitation and involve deep mediastinal structures. The disease is more prevalent in men of Mediterranean origin aged 30-60 years and is associated with chronic alcoholism. Diagnosis is based on clinical findings and complementary imaging tests. Generally, the most accepted treatment approach is surgical excision of the lesions, although several alternative treatments have been described. This study reports the clinical case of an MSL patient treated with multiple surgical resections. It describes the diagnosis and compares the clinical findings and treatment of the patient with the main correlated comorbidities in the literature.

Keywords: Madelung's disease; Launois-Bensaude Syndrome; Multiple Symmetrical Lipomatosis.

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Introduction

Multiple Symmetric Lipomatosis (MSL), Madelung's Disease, or Launois-Bensaude Syndrome is characterized by the presence of multiple unencapsulated lipomas commonly located in the neck, chest, and roots of the upper limbs. Lesions usually develop slowly and asymptotically, but there may be limited cervical mobility and involvement of deep mediastinal structures, leading to respiratory and sleep disturbances and difficulties in orotracheal intubation¹.

The etiology of MSL is still unknown, but a correlation with catecholamine-induced abnormal lipid metabolism has been reported². Diagnosis is reached through clinical investigation and complementary imaging tests, which aim to differentiate it from tumors that affect the same region, assess disease extent, and provide for good preoperative planning³.

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This study reports the clinical case of an MSL patient treated surgically. It describes the diagnosis and compares the clinical findings of the patient with the main data in the literature.

Case report

This case report was approved by the Ethics Committee of the aforementioned Institution under protocol (CAAE) no. 22869719.8.0000.0096. A 57-year-old male patient with non-contiguous circumferential masses in the cervical, left pectoral and presternal regions, with progressive growth over the course of 10 years (Figure 1). An important limitation to cervical mobility, with no local pain, dyspnea or dysphagia, was observed. Alcoholic patient with smoking exposure of 15 pack-years. Alcohol consumption ceased after medical advice. History of comorbidities included chronic venous insufficiency and an episode of deep venous thrombosis. No family history of the disease. Normal routine laboratory test results.

Magnetic resonance imaging (MRI) of the cervical region showed multiple subcutaneous masses, with the largest (23 x 15 cm) located in the left lateral region. The masses insinuated in the posterior cervical spaces and compressed the parotid and submandibular glands. The larynx and trachea were displaced to the left (Figure 2).



Figure 1. Cervical lipomatous masses before the last surgical resection performed in 2019. **A** - Anterior view. **B** - Posterior view.

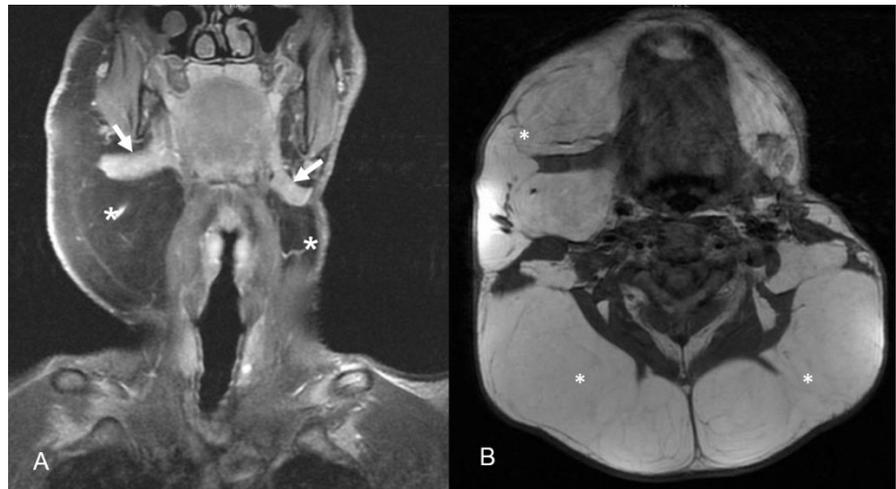


Figure 2. MRI of the cervical region. **A** - Coronal section showing cervical lipomatous masses located subcutaneously (asterisks), with compressive effect and displacement on the parotid and submandibular glands (arrows). Discrete displacement of the larynx and trachea to the left. **B** - Cross section with a lipomatous mass in the anterior region and two masses in the posterior region (asterisks).

The patient underwent six cervical surgical resections and there was need for fiberoptic bronchoscopy for orotracheal intubation in the first surgical procedure. In one of the surgeries, a lipoma contiguous with the carotid sheath, encompassing the marginal mandibular and anterior cervical nerves and part of the parotid, was found, and there were no intraoperative lesions. The anatomopathological analyses were compatible with lipoma, and the total weight of the pieces was 1.748 g.

Discussion

There is comprehensive description in the literature that MSL commonly affects men aged 30-60 years, with no hereditary correlation, and with a history of excessive alcohol intake, which would be associated with an aggravating or predisposing factor in up to 90% of cases. MSL seldom occurs in women and non-alcoholics. In this context, alcohol abstinence can limit the progression of lesions, but it does not lead to their regression. Often, MSL is also associated with systemic changes such as liver disease, glucose intolerance, diabetes mellitus, hyperlipidemia, hypertension, hyperuricemia, polyneuropathies, and hypothyroidism. Moreover, it usually involves the neck, chest, and roots of the upper limbs, and spares the face, hands, and feet⁴. The epidemiological profile of this clinical case corroborates the literature, and it was observed that although the patient ceased alcohol consumption after the third surgery on medical advice, this was not sufficient to prevent lesion progression. In addition, the patient did not have a history of affected family members, once again corroborating the non-hereditary character of this disease described in the literature.

It is proposed that the pathophysiology of MSL is related to mitochondrial DNA mutations, which are associated with changes in intracellular enzymes and membrane receptors that lead to reduction in lipolysis mediated by

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the noradrenergic system. In this context, alcohol consumption seems to induce reduced expression of noradrenergic receptors and a disorder in the mitochondrial DNA of the adipose tissue, participating as an enhancer of the pro-lipogenesis and anti-lipolysis effects and reducing lipid oxidation in genetically susceptible individuals².

The correct management, in addition to clinical and laboratory history for the correct finding of associated comorbidities, involves tests such as MRI, computed tomography (CT), or ultrasonography (US)³. In this clinical case, no noteworthy laboratory abnormalities or associated systemic comorbidities were observed, and MRI was chosen for better preoperative planning, because the lipomatous masses could affect the vascular and nervous structures, which could be damaged during surgery.

There is no specific recommendation in the literature for aggressive lipectomy, with reports on clinical observation or liposuction, due to the predominantly benign behavior of MSL, although surgical management is considered the preferred approach in the management of affected patients^{3,5}. The most frequent surgical indications found in the literature are due to esthetic issues, limited mobility, and local discomfort⁵. In this clinical case, a choice was made for sequential surgical excision, starting from the lesions that caused greater cervical mobility limitation and discomfort. Open surgery enabled better exposure of the operative field, less chance of injury to nervous and vascular structures, good hemostasis, and smaller chances of having residual masses.

Anatomopathological analyses showed no signs of malignant transformation, and the diagnosis was compatible with lipoma. Thus, the importance of continuous follow-up of affected patients is highlighted, not only due to the minimal chance of sarcomatous transformation, but also to the relapsing and progressive nature of this disease, with the possibility of serial surgical approaches to treat its consequences.

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