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Atypical Presentation of Madelung Disease

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Abstract

We present a unique case of a sixty two year old woman with symmetric lipomatosis of the lower back, similar to cup C female breast. Lipoma is the most common type of soft tissue mesenchymal tumor. Lipoma is commonly found in the upper back, neck, shoulder and abdomen, and rarely in the face, hands or feet. When lipomatosis is symmetrical it is often termed Multiple Symmetrical Lipomatosis (MSL), also known as Madelung disease, a rare syndrome of unknown etiology.

Our patient was referred with large symmetrical subcutaneous masses in her lower back similar to cup C female breast. Prior to her surgery, the patient was referred to ultrasonography of the lower back which demonstrated two large subcutaneous lesions with an average size of 12.5*3*14 cm. Lesions were excised under general anesthesia, with the left lipoma weighing 535 gr', and the right 425 gr', and sent for pathological examination. Patient was discharged following the removal of drains after two days, pathology confirmed clinical diagnosis and upon six months follows up, surgical wounds have healed nicely.

Although this disease remains rare, the deferential diagnosis between simple lipoma, MSL and obesity may contribute to current under estimation of MSL.

The importance of accurate diagnosis lies within thorough patient evaluation for related systemic co morbidities. Plastic surgeons should bear in mind that clinical presentation may vary, as for our patient, and must be familiar with this disease and its management.

Keywords

Madelung disease, Symmetric lipomatosis, Lipoma

Introduction

Lipoma, a benign mesenchymal lesion, is the most common type of soft tissue tumor. It is usually located superficially in the subcutaneous tissue. Less frequently it can be located deeper, under the fascia or within the muscles [1]. Lipoma is mostly asymptomatic but may compress near structures, depending on its size and location. Lipoma is most commonly found in the upper back, neck, shoulder or abdomen, but can be found anywhere in the body [2]. When lipomatosis is symmetrical it is often termed Multiple Symmetrical Lipomatosis (MSL), also known as Madelung disease, benign symmetrical lipomatosis, Launois-Bensaude adenolipomatosis, Brodie syndrome or buschke disease. Its first description was in 1846 by Benjamin Brodie. The classical "horse collar pattern" of cervical lipomas distribution was described by Otto W Madelung in 1888. Launois and Bensaude concluded the description of the syndrome in 1898, naming it as multiple symmetrical adenolipomatosis [3].

MSL is characterized by the presence of large adipose masses, minimally encapsulated, soft, painless, slowly growing and classically located at the cervical or limb area. The disease generally occurs in men aged 30 to 60, women/men ratio ranges between 1/15 - 1/30, and is seen mostly in Mediterranean countries [4]. MSL is associated with increased alcohol intake in up to 95% of cases and patients have also been known to suffer from other comorbidities such as peripheral neuropathy (80% of patients), macrocytic anemia, alcoholic fatty liver or cirrhosis, hyperuricemia, hyperproteinemia, high lipid blood levels, arterial hypertension, COPD, obesity and diabetes mellitus. Differential diagnosis with cushing disease, simple obesity, neck cysts, salivary gland an thyroid gland disease, leukemia and soft tissue sarcoma. The etiology of MSL remains unknown. Although there might be a genetic predisposition, most cases are sporadic with exact risk factors and pathophysiology unknown [5]. As MSL is highly associated with alcohol intake, theories suggests that alcohol may influence enzymatic process in the mitochondria or impair adrenergic lipolysis and lead to uncontrolled emergence of fat deposits in the body [6-8]. Other theories include increased lipoprotein lipase activity, mitochondrial DNA (mtDNA) mutations or sympathetic denervation of brown fat adipocytes leading to hypertrophy [5]. In a case report presented by Pazmatzi et al., a female patient with type I MSL was presented. She did not report increased alcohol intake similar to our patient. The authors suggested that mtDNA mutation may account for the majority of MSL cases without a history of alcoholism. As previously demonstrated by Klopstock *et al.*, mtDNA mutation was identified in 2/3 patients without a history of alcoholism, but in none of the patients with high alcohol intake [9].

Two variants are described by the Enzi classification most widely used: Type I MSL where lipomatosis may be profound and is distributed mostly around the neck (nuchal, subclavian and deltoid regions) forming the "Madelung collar", and type II where lipomatosis is located more caudally, without affecting the neck. In type II MSL patients, lipomatosis is not profound and they lack lipodystrophy of the arms and legs [7].

Diagnosis of MSL is clinical, based on symmetrical distribution of fatty mass [4]. Imaging may assist diagnosis. Chest radiographs may show abnormal symmetrical accumulation. Ultrasound and CT may evaluate disease extent however MRI is the best diagnostic tool for evaluating the spread of adipose tissue, presence of tracheal

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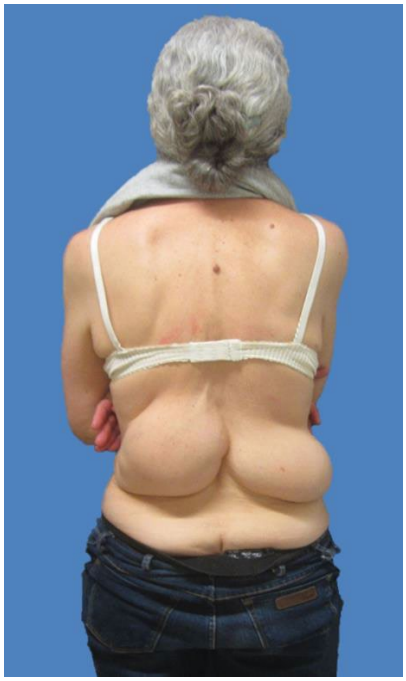

Figure 1: Clinical appearance before surgery with bilateral symmetric lipomatosis of lower back.

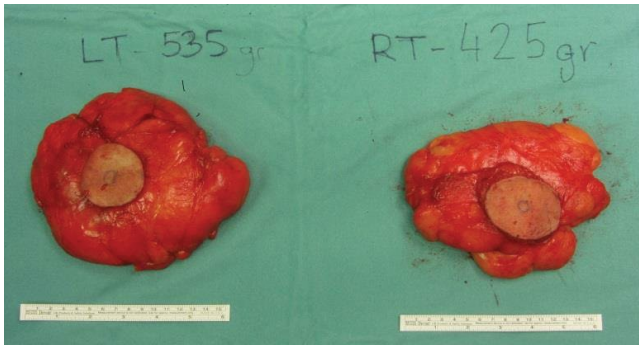

Figure 2: Complete surgical removal of the lesions with skin excess. Notice that lipomas lack a distinct membranous capsule.

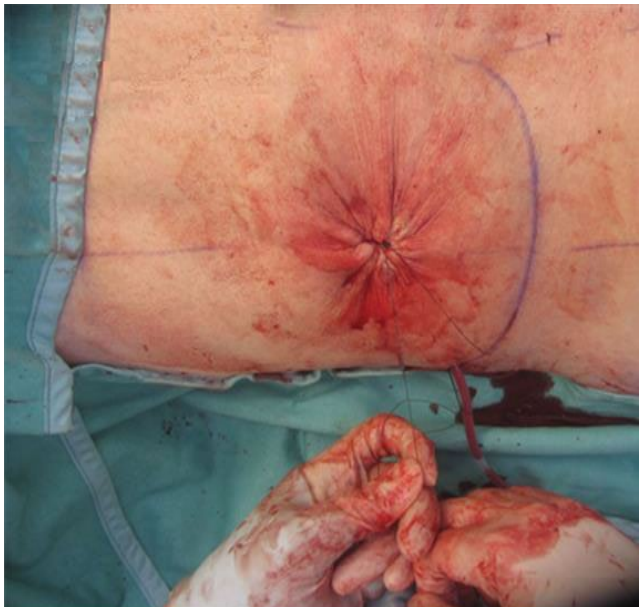

Figure 3: After lipoma removal, skin reduction and wound closure were first addressed with a "Periareolar skin pattern reduction" and purse- string suture. However, due to high tension closure, a "mastectomy type" horizontal scar was made that better addressed skin excess.

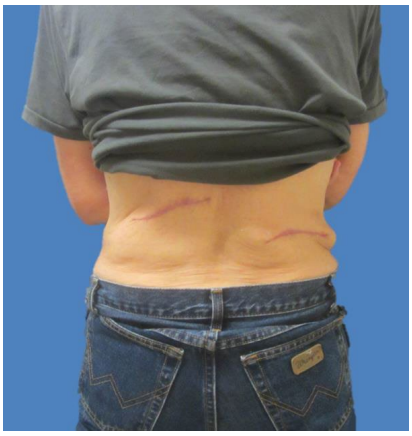

Figure 4: Postoperative view upon 6 months follow up with immature scars.

compression and vascular topography within the fat mass. Patients initially complain of aesthetic changes but without treatment reduced neck mobility or compression of respiratory structures may appear [10]. Indications for treatment include breathing difficulties, dysphagia, decreased head movements and aesthetic reasons. Possible treatment options for patients with aesthetic or functional deformities include dietary changes, surgical excision and liposuction. Surgical excision remains the most common and effective treatment modality, however recurrence may occur for all treatment modalities. Possible surgical complications are hematoma, seroma, recurrence or neuropraxia. Dietary treatment is ineffective as lipomas remain even with cachexia [5,8,11,12].

Case Report

A sixty two year old healthy Caucasian woman, married with two children, was referred to our department with large symmetrical subcutaneous masses in her lower back, similar to cup C female breast (Figure 1). She doesn't take medication on a regular basis, doesn't smoke or drink alcohol. She has no known allergies or family history of lipomatosis. Lesions were asymptomatic and grew slowly over a period of twenty years, but recently started to accelerate their growth. Physical examination showed two soft symmetrical subcutaneous masses in the lumbar area, on each side of the vertebral spine. Ultrasonography of the lower back was performed preoperatively, which revealed subcutaneous lesions with an average size of 12.5*3*14 cm and consistent with lipoma. Lesions were excised under general anesthesia, with the left lipoma weighing 535 g and the right 425 g (Figure 2), and sent for pathological examination. After lipoma removal, skin reduction and wound closure were first addressed with a "Periareolar skin pattern reduction" and purse-string suture (Figure 3). However, at the end, a "mastectomy type" horizontal scar was made that better addressed skin excess. Surgical drains were removed after two days and the patient was discharged. Pathology report confirmed clinical diagnosis of lipoma and upon six months follow up, surgical wounds have healed nicely (Figure 4). Finally, there were neither complications nor evidence of recurrence up to 18 months follow up.

Discussion

Madelung's disease, known as multiple symmetric lipomatosis, is a rare syndrome of unknown etiology characterized by indolent growth of symmetrical asymptomatic fatty masses.

Two variants of the disease have been described. Type I MSL, which is more common and characterized by symmetrical growth of fatty masses distributed around the face, neck, upper trunk, and proximal parts of limbs. Type II MSL where symmetrical distribution appears elsewhere. The disease generally occurs in middle aged men, women/ men ratio ranges between 1/15- 1/30, and is seen mostly in Mediterranean countries [4,10]. The disease usually has biphasic course, an initial rapid growth that is followed by a slow progressive phase. Currently spontaneous regression has not been reported.

Unlike lipomas, these lesions lack a distinct membranous capsule. Although this disease remains rare, the deferential diagnosis between simple lipoma, MSL and obesity may contribute to current underestimation of MSL. Ergo, they are often considered as simple adipose tissue due to obesity [4,13]. Other possible reasons for misdiagnosis are the lack of strict inclusion criteria as to the localization and dimensions of lipomas in MSL due to scarce reports in the literature and slow progression with delay in diagnosis. The treatment of MSL is unsatisfactory as there is a high recurrence rate. Weight loss and alcohol consumption cessation have no effect on lipoma growth, while liposuction and surgical excision are often associated with recurrence [10]. The importance of accurate diagnosis making lies within thorough patient evaluation for the above mentioned systemic comorbidities. Furthermore, as 90% of MSL cases are related to high alcohol intake, alcohol abuse must be ruled out and if present, patients must be referred for appropriate care. Surgeons should bear in mind that clinical presentation may vary, as for our patient, and must be familiar with this disease and its management.

Disclosure and Statement of Ethical Standards

None of the authors has a financial interest in any of the products, devices, or drugs mentioned in this manuscript. Maisel Lotan A, Retchkiman M, Gronovich Y declares that they have no conflict of interest.

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