Case report

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Madelung's disease. Two case reports with pseudoathletic appearance

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Abstract

Madelung's disease is a rare syndrome characterized by the presence of multiple masses of unencapsulated adipose tissue, symmetrically distributed throughout different regions. It predominantly affects middle-aged men of Mediterranean origin with a history of alcoholism. The pathogenesis is still unknown. Diagnosis is essentially established through clinical history and physical examination. We report two cases for their unusual presentation and to emphasize the importance of early diagnosis. ***Clin Ter 2021; 172 (3):190-192. doi: 10.7417/CT.2021.2311***

Key-words: *Madelung's disease, cutaneous lipomatosis, multiple symmetric lipomatosis, Launois-Bensaude syndrome, benign symmetric lipomatosis*

Dear editor,

Madelung's disease (MD; also known as multiple symmetric lipomatosis, Launois-Bensaude syndrome, or benign symmetric lipomatosis) is a rare syndrome (1:25.000) characterized by abnormal fat tissue distribution (1,2).

It predominantly affects middle-aged men of Mediterranean origin with a history of alcoholism. The pathogenesis is still unknown. Nevertheless, several hypothesis such as a defect in the adrenergic stimulated lipolysis, a primary defect within the surface membrane of the adipocyte cell, a neoplastic disease that could originate from brown fat, or a defect in brown fat mitochondrial DNA (both in inherited and acquired defect) have been proposed (3-5).

The typical distribution consists of massive lipomatous deposits around the neck, which gives the classic descriptions of lipoma as anulare colli, "buffalo hump", and "horse collar" (2).

We report two patients with an unusual presentation.

The first patient, a 58-year-old man, showed prominent, symmetric masses of adipose tissue on the bilateral upper arms, shoulders, abdomen, and along her back (Fig. 1).

The familial history was negative for multiple lipomas. The patient reported more than 20 years of alcohol abuse (about 500 g/d) and heavy smoking (about 30 cigarettes per day). Physical examination disclosed a BMI of 28.9. The patient had no evidence of sensory, motor or autonomic neuropathy.

Lab tests showed hypertriglyceridemia (267 IU/ml). Histopathologic examination of adipose tissue removed confirmed adipose tissue without malignant transformation. CT scan (with contrast) of the chest showed diffuse enhancement of soft tissues (fat density) without internal lesions.

In view of the history, clinical finding and the histology report, a diagnosis of MD was made.

The second patient, a 45-year-old woman showed symmetrical, painless tumors, of stiff, smooth, and soft texture, located on her arms, anterior thorax, trapezius, and along her back giving the patient an "pseudo-athtletic" shape (Fig. 2).

The familial history was negative for multiple lipomas. There was alcohol abuse in his history with consumption of more than 1.5 L of wine per day.

Physical examination disclosed a BMI of 30.9. The patient had no evidence of sensory, motor or autonomic neuropathy. An ultrasound examination of cutaneous lesions showed unencapsulated fat deposits. Histological examination showed mature adipocyte depositions in the subcutaneous tissue. Lab results revealed a total cholesterol level of 303 mg/dl and a triglyceride level of 276 mg/dl.

In view of the history, clinical finding and the histology report, a diagnosis of MD was made.

MD is a rare syndrome characterized by the presence of multiple masses of unencapsulated adipose tissue, symmetrically distributed throughout different regions. The main affected areas are the neck, shoulder girdle, dorsal region, and proximal upper and lower limbs, but other locations such as the retroauricular region and submandibular area, tongue, perineum, mammary region, abdomen, or scrotum have also been described.

Diagnosis is essentially established through clinical history and physical examination. CT and MRI imaging have been found to be useful for evaluation of the spread of adipose tissue, tracheal compression, vasculature within the adipose mass, and exclusion of malignancy. Biopsy can further confirm a suspected diagnosis (6).

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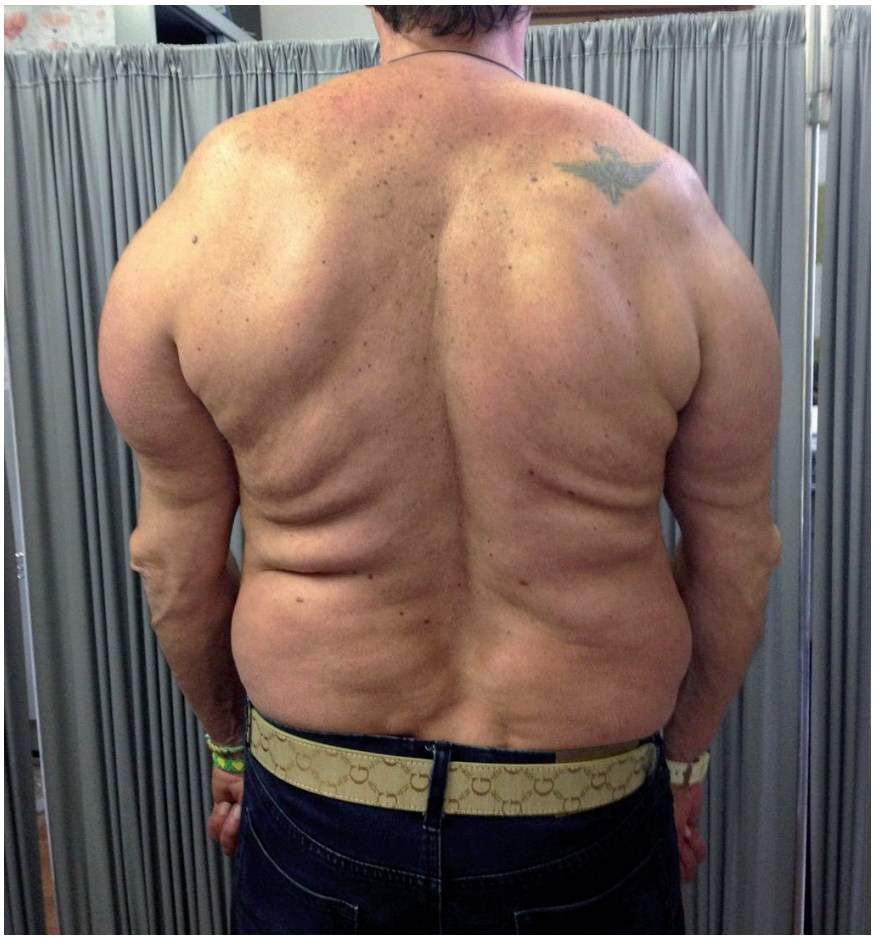

Fig. 1. Multiple lipomas located on arms, shoulders, trapezius, and along back in a man with Madelung's disease (pseudo-athtletic" shape).

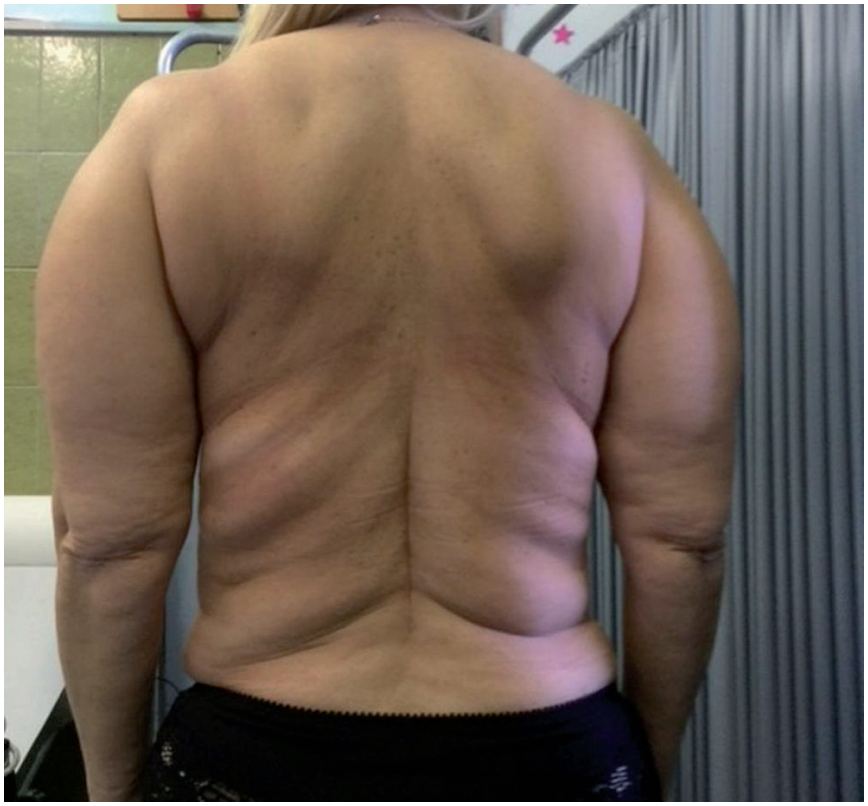

Fig. 2. Multiple lipomas of the trunk in a woman with Madelung's disease (pseudo-athtletic" shape).

The differential diagnosis is made with morbid obesity, Dercum's disease, familial multiple lipomatosis, iatrogenic cutaneous lipomatosis, Cushing syndrome, angiolipomatosis, encapsulated lipomas, neurofibromatosis, myxoid liposarcoma, lymphoma, salivary gland disease, Frölich syndrome and lipomatosis syndrome in patients infected with HIV (7-18). Lipomas can sometimes, though rare, be associated with certain disorders such as Ruvalcaba syndrome, Cowden syndrome and Neurofibromatosis (19-22).

There are principally 3 different classifications used in the literature.

Enzi's classification, which is the most widely accepted, proposes two types of diseases according to the anatomic distribution of fatty deposits. In MD type 1, the location is symmetrical and mainly distributed in the neck (Madelung's collar or "horse collar"), shoulders, supraclavicular fossa, and proximal regions of the upper limbs. In severe cases, this type can cause mediastinal extension and tracheal obstruction. In type 2, the neck area and upper trunk are not affected; deposits occur in abdomen and thighs and may be confused with normal obesity (23).

Donhauser et al. subdivided Enzi's type 1 and distinguishes three types of MD: type 1 (neck distribution), type 2 (pseudoathletic appearance), type 3 (gynecoid). In some patients, more than one type of distribution may be present (24).

Schiltz divided phenotypes of MD into 5 types: type Ia (neck), type Ib (neck + shoulder girdle + upper arms), type Ic (neck + shoulder girdle + upper arms + trunk), type II (hips, bottom upper legs), type III (general distribution skipping head, fore-arms, and lower legs) (25).

Over 400 cases were described in literature, but only a few patients had a pseudoathletic appearance. In the classic form of MD lipomas occur along the neck, upper aspect of the trunk, and proximal upper extremities, with sparing of the lower half of trunk and abdomen.

We report these cases for their unusual presentation and to emphasize the importance of early diagnosis. Patients with MD can develop functional symptoms including dysphagia, odynophagia, or hoarseness as a result of fatty deposits compressing the cervical region.

Madelung's disease, and in particular the pseudoatletic appearance, is less well known and can go undiagnosed for years. A careful clinical history is very important.

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