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Case Report

A non–alcohol-related case of Madelung's disease: Challenging patient with progressive jugular vein distension ☆,☆☆

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ABSTRACT

Madelung's disease or multiple symmetric lipomatosis (MSL) is a rare benign disease characterized by abnormal, multiple and symmetric fat depositions in the subcutaneous layer, involving head, neck, back, trunk and also upper and lower limbs.

MSL may be related to alcohol abuse or metabolic disorders; it may be both silent or clinically manifest.

We describe a case of a 48-yo man with β-thalassemia admitted to medicine department for neck swelling without fever or respiratory symptoms. Patient denied a history of alcoholism and laboratory exam excluded metabolic disorders.

Doppler ultrasound, contrast Enhanced-CT and Magnetic Resonance Imaging exams of the neck showed a symmetric, non-encapsulated fat deposition causing extrinsic compression of the right jugular vein without thrombosis. Once excluded the possibility of malignancy, patient's history, clinical, and radiological findings suggest the diagnosis of non–alcohol-related MSL disease.

Knowing MSL imaging findings and its degree is crucial to guide towards the right management.

Our patient did not require surgical treatment and an US follow-up is needed in order to detect any possible evolution.

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Introduction

Madelung's disease, also known as "Multiple Symmetric Lipomatosis" (MSL), is a rare benign disease characterized by abnormal, multiple and symmetric fat depositions in the subcutaneous layer; it usually involves head, neck, back, trunk and nerve roots of the upper and lower limbs [1–4]. MSL may be related to alcohol abuse or to metabolic disorders (ie, glucose intolerance, dyslipidemia, liver disease, chronic renal failure, or hypothyroidism [5–7]. Recently several authors report a mitochondrial genetic dysfunction associated to MSL [8]. MSL may be asymptomatic or clinically manifest, with symptoms resulting in compression of vascular, nervous, and respiratory tract structures [3,6,9]. This condition has been associated with obstructive sleep apnea syndrome (OSAS) [10]. Rarely, spontaneous transformation of MSL into a liposarcoma has been described in literature [11,12]. Patient's clinical history, clinical and radiological findings play a crucial role to diagnosis the MSL [6]. Madelung's disease treatment ranges from reduction of alcohol consumption – in case of alcohol-related syndrome – to surgical resection [3,6,13].

We describe an intruing case of non–alcohol-related Madelung disease in a patient with neck swelling.

Case Description

A 48-yo man was admitted to medicine department complaining of neck swelling without fever or respiratory symptoms. Patient denied a history of alcoholism or metabolic disorders. He had a brain stroke in 2003 with residual dysarthria. His medical history was remarkable for β-thalassemia intermedia since he was four, which required surgical intervention of splenectomy. Patient suffered the onset of autoimmune hemolytic anemia – and consequently he was assigned to blood transfusion therapy.

At clinical evaluation, the patient showed a bilateral turgor of jugular veins, most evident in supraclavicular regions of the neck, with nonpitting edema. We have also investigated other regions of the body, and no other edema regions were depicted.

Laboratory tests was unrevealing. In detail, basic metabolic panel were normal except for an increase of total bilirubin value of 2.5 mg/dL (reference range [rr.] 0.3-1.2 mg/dL) with a bilirubin direct value normal. These latter values may be correlated to hemolytic icterus due to his blood disorder. In fact the hemocromocitometric parameters (Hct 28.8 % [rr. 41-51%]; Hb 8.8 g/dl [rr. 14-17 g/dl]; RBC 3.65 $\times$ $10^6/\mu$l [rr. 4,6-6,2 $\times$ $10^6/\mu$l]; MCV 79 fl [rr. 80-98 fl]; MCH 24.2 pg [rr. 26-32 pg]; MCHC 30% [rr. 32-36%]; RDW 23.2 % [rr. 11,6-14,6 %]; PST 787 $\times$ $10^3/\mu$l [rr. 150-350 $\times$ $10^3/\mu$l]) reflect the hematological disease of thalassemia.

TFTs, lipid panel and coagulation tests were both normal. Patients Vitamin D 25-H and Uric Acid were also normal. Finally a nonspecific increase of LDH value of 397 mU/ml (rr. 80-300 mU/ml).

The neurological examination excluded the compression of neurovascular structures or other neurological disorders.

He underwent a cardiac examination, including ECG and echo-cardio, which were negative for cardiovascular pathology. Doppler US examination showed mild compression of right jugular vein without jugular, axillary, or subclavian veins thrombosis.

Since cardiovascular diseases have been excluded, other examinations were performed in order to investigate the presence of neck masses.

Potential diseases – such as extramedullary erythropoiesis, solid mass or soft tissue edema – were considered by clinicians.

The chest radiograph revealed normal appearance of superior mediastinal region (Fig. 1); to better analyze vascular structures and neck enlargement, he was scheduled for a contrast-enhanced-CT (CE-CT) scan of the neck and chest.

CE-CT scan showed a symmetric and nonencapsulated excess of fat, with soft tissue density, mainly deposited in the anterior and posterior subcutaneous tissue of the neck – with extrinsic compression of the right jugular vein (Fig. 2).

Magnetic Resonance Imaging (MRI) exam, performed with a 1.5 Tesla scanner, confirmed the presence of superficial well-defined nonencapsulated subcutaneous fat tissue, with bilateral and symmetric distribution, showing normal hyperintense signal in both T1 and T2 weighted sequences and causing an extrinsic compression of the right jugular vein (Fig. 3). Both CT and MRI exams did not show findings suspected for malignancy; therefore, no contrast administration was required performing examinations.

Discussion

MSL is a rare benign disease with unexplained etiology and unclear pathogenesis [6]. It is characterized by multiple symmetric abnormal fat depositions in the subcutaneous layers most frequently localized around the neck [1–4,6].

Based on location, Enzi et al has proposed a classification into two types of lipomatosis: MSL *type 1*, also known as "Madelung's collar" with fat depositions mainly deposited at the upper back, neck and arms leading the patient to a pseudo-athletic look; MSL *type 2*, having fat mainly deposited at hips and thighs like in simple obesity [14]. Once the MLS diagnosis is formulated our patient can be classified as a neck limited form of *type 1*.

MSL is often related to metabolic disorders with or without history of alcohol abuse [6,7]. In literature, cases related to glucose intolerance, dyslipidemia, liver disease, chronic renal insufficiency or hypothyroidism have been reported [5–7].

Recently several authors report a mitochondrial genetic dysfunction associated to MSL [8].

Since there's no evidence for familial disposition in our patient, no MSL inheritance was suspected.

In our patient, all laboratory exams have excluded these mentioned pathological associations. MSL may be clinically silent, or may provoke compression of aero-digestive organs – leading to symptoms such dysphasia, dyspnea or OSAS

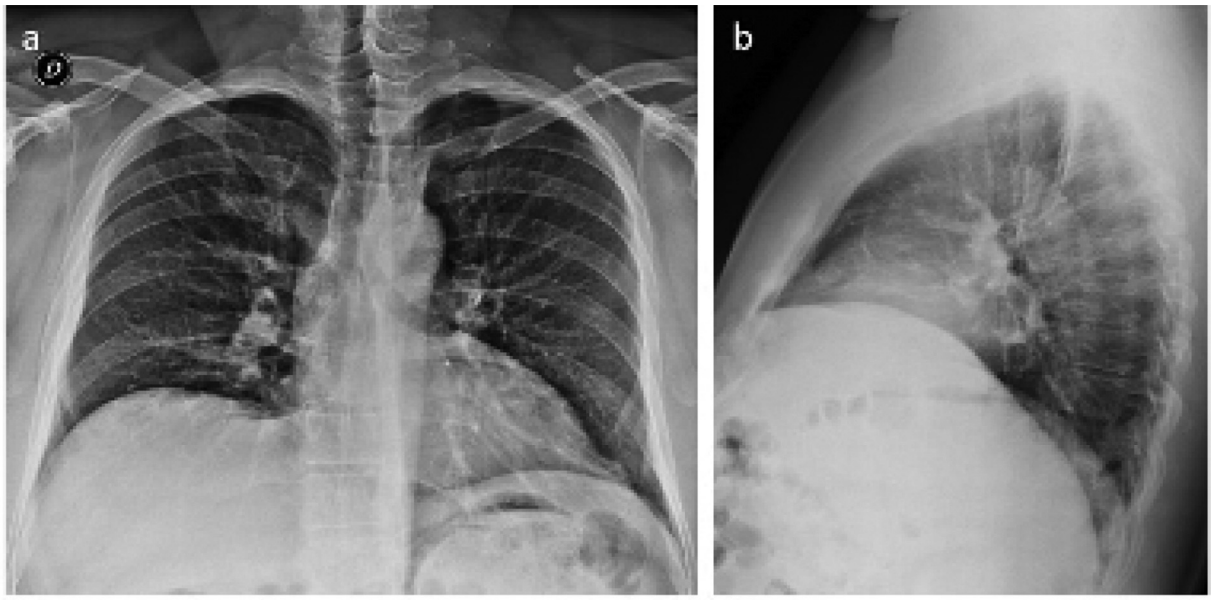

Fig. 1 – Posteroanterior (PA) (a) and lateral views (b) of chest radiograph show normal appearance of superior mediastinal region.

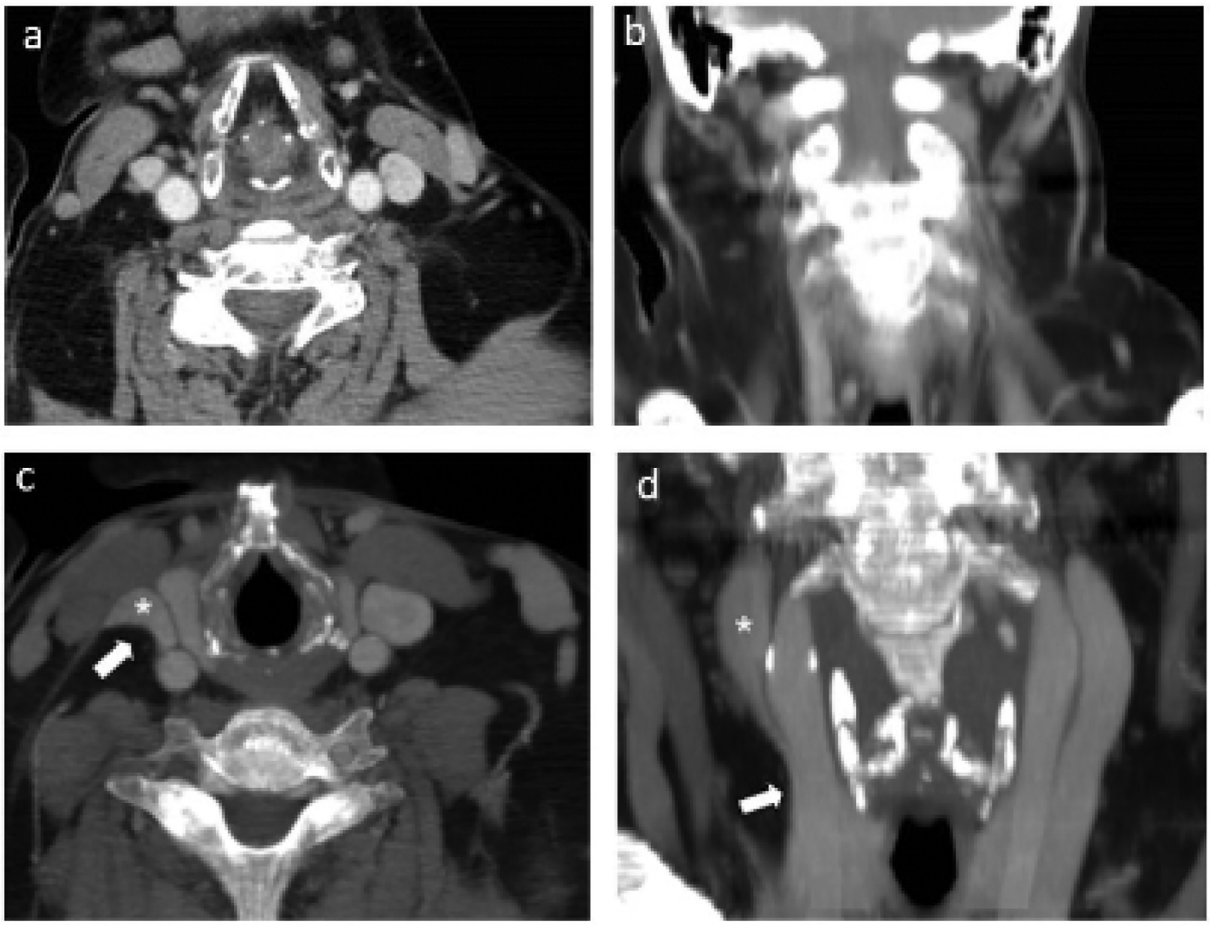

Fig. 2 – Axial (a,c) and coronal (b,d) CT images show bilateral neck lipomatosis with extrinsic compression (arrow) of the right jugular vein (*) for about 4 cm in length.

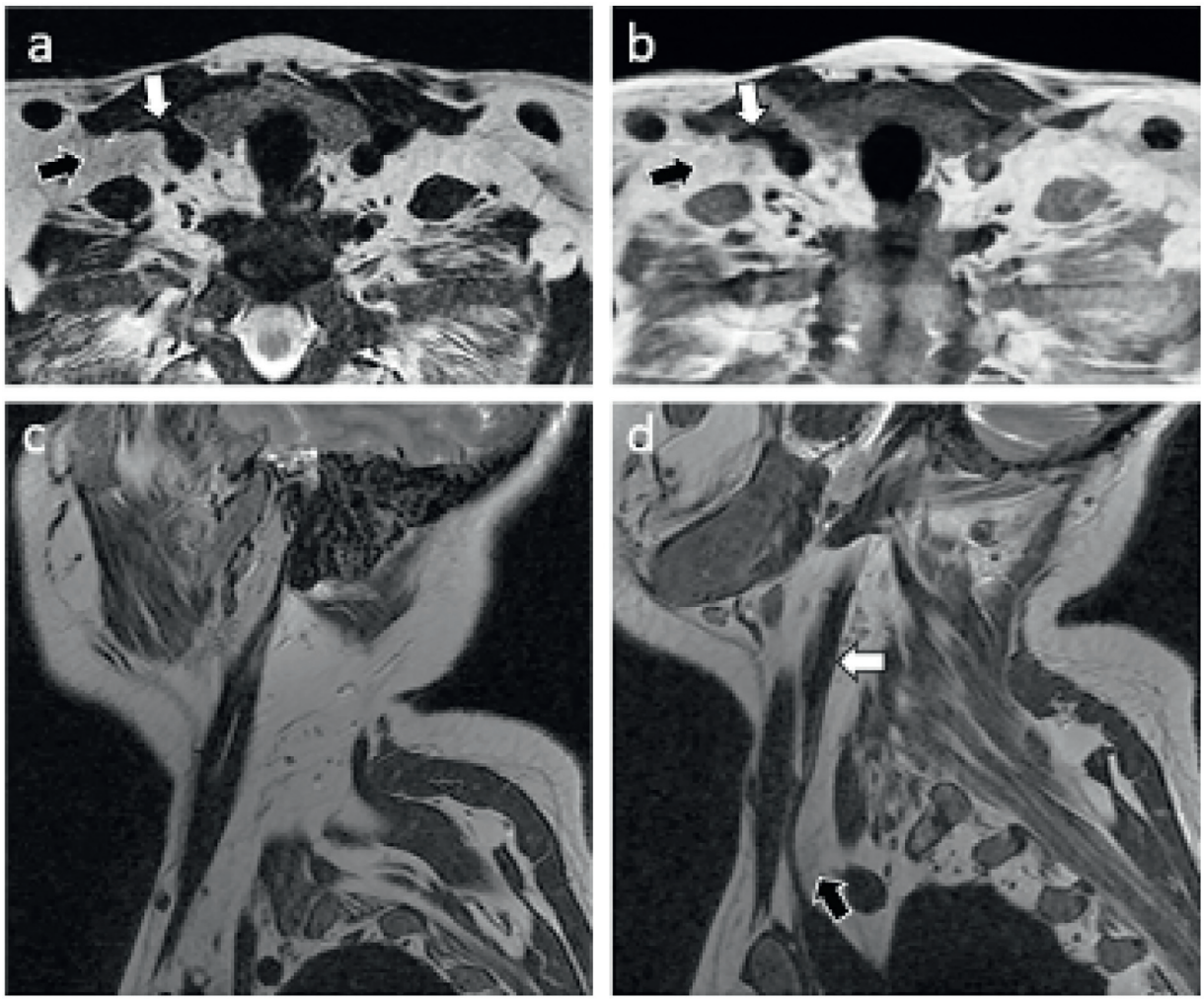

Fig. 3 – Axial T2 (a), axial T1 (b) and sagittal T2 MRI images show bilateral neck lipomatosis with extrinsic compression (black arrow) of the right jugular vein (white arrow).

[3,6,10]. Additionally, compression of neurovascular structures has been also reported [9].

In our patient, the neck limited form without any neurological symptoms excluded the possibility of myasthenia gravis or muscular dystrophies, which have been described as associated conditions in literature [15].

Doppler ultrasound (US) examination is now the first-line to achieve diagnosis of MSL; usually, it provides useful information regarding major vessels of the neck. In our case, we have revealed a lipomatous tissue in the inferior part of the neck – showing only mild compression of right jugular vein – without signs of vascular thrombosis. Doppler US is also useful to exclude the presence of benign or malignant lymphadenopathy.

Cross-sectional imaging procedures play a crucial role in the evaluation of the fat deposition; it allows also to obtain important findings regarding relationships with the surrounding structures, and the possibility to depict signs of malignant degeneration [2,10,12]. Therefore we have decided to perform CT and MRI in our patient, just to better define its location and relationship with vessels and muscles.

CE-CT scan has confirmed the presence of a symmetric and nonencapsulated deposition of fat at subcutaneous tissue of the neck, with mild extrinsic compression of the right jugular vein – which was previously depicted by US. At MRI examination, the nonencapsulated deposition of fat tissue at subcutaneous layers has been demonstrated as soft-tissue swelling having well-defined margins, and without any involvement of surrounding muscles. Both CT and MRI exams confirmed the absence of findings suspected for malignancy; in this regard, it's important to keep in mind that some descriptions of spontaneous transformation into a liposarcoma in literature have been reported. In these cases, a differential diagnosis could be relatively difficult [10,12].

In conclusion, considering the benign nature of neck swelling at radiological examination - with homogenous appearance of fat, well-defined margins, not-infiltrating pattern, and without involvement of surrounding structures - we finally formulated the hypothesis of Madelung'disease.

Madelung's disease treatment is palliative and includes surgical resection of the adipose tissue, liposuction, or injection lipolysis [3,6]. In case of alcohol-related syndrome, reduc-

ing its consumption is also recommended [13]. For our patient we performed a follow-up based on laboratory tests and US examination – in order to monitor the eventual evolution toward a case that requires surgery which was excluded.

Contributions of Authors

CG and FG: drafting the article, acquisition and interpretation of data. SP and SSS: conception and design of study. AB: final approval of manuscript.

Ethical approval

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Informed consent

Informed consent was obtained from the individual included in the case report.

Consent for publication

Consent for publication was obtained for every individual person's data included in the study.

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