CASE REPORT

Severe dyspnea as atypical presenting symptom of Madelung's disease

Milisavljevic D[1], Zivic M[1], Radovanović Z[2], Stankovic P[1]

[1] Clinic for Otorhinolaryngology, University Clinical Center Nis, Serbia
[2] Institute for Radiology, University Clinical Center Nis, Serbia

Abstract

Madelung's disease (benign symmetrical lipomatosis) is a rare disease of unknown etiology manifesting as symmetric abnormal deposits of adipose tissue in the head, neck and upper trunk. We report a case of a 58-year-old man with a long lasting Madelung's disease in whom progressive fatty tissue accumulation caused a severe inspiratory dyspnea as atypical presenting symptom. The etiopathogenetic, clinico-diagnostic and therapeutic aspects of this rare disease are discussed. Due to its progressive but not so easily predictable enlarging behavior Madelung's disease has not only aesthetic but also functional and sometimes life threatening consequences which need to be treated. Hippokratia 2010; 14 (2): 133-135

Key words: benign symmetrical lipomatosis, Madelung's disease, Launois-Bensaude syndrome

Corresponding author: Milisavljevic D, Clinic for Otolaryngology, Bul. Zorana. Djindjica 52, 18000 Nis, Serbia, tel: +38163444657, fax: +381184531272, e-mail: dusanorl@yahoo.com

Madelung's disease (benign symmetrical lipomatosis) is a rare disease of unknown etiology manifesting with symmetric abnormal deposits of adipose tissue in the head, neck and upper trunk. In 1846, Brodie first described the disorder but in 1888, Madelung collected a series of cases outlining the macroscopic structure and clinical features of this disease[1,2]. The disorder usually occurs in middle-aged men with history of alcoholism[3,4]. Patients suffering from this benign symmetric lipomatosis may complain of difficulties in swallowing, turning of the head, pain in the neck or larynx or symptoms associated with organs in the mediastinum. Sensory, motor and autonomic neuropathies are frequently present. In long lasting cases, enlarging fat tissue can push larynx and trachea causing also dyspnea.

We present a case of a 58-year-old man with a long lasting Madelung's disease in which progressive fatty tissue accumulation caused a severe inspiratory dyspnea and we discuss the etiopathogenetic, clinico-diagnostic and therapeutic aspects of this rare disease.

Case report

A 58-year-old man presented to the department of Otorhinolaryngology with severe dyspnea due to a 7 years history of a gradually progressing Madelung's disease. Painless thickening of the face, neck and shoulders was noticed. He had a history of heavy alcohol comsumption and suffered from altered lipid metabolism (Figure 1,2).

A chest x-ray and a Computed Tomography (CT) scan of the neck and thorax without contrast showed excess of fat predominantly in the posterior part of the neck, deeper to trapezius muscle, in the sternocleidomastoid muscles, in the supraclavicular fossa, in the anterior neck, in the superior mediastinum, and also in the pretracheal space with compression of the trachea. Due to the intense dyspnea a tracheotomy was performed and at the same time a surgical lipectomy was done in order to remove the compression on laryngotracheal region. Com-

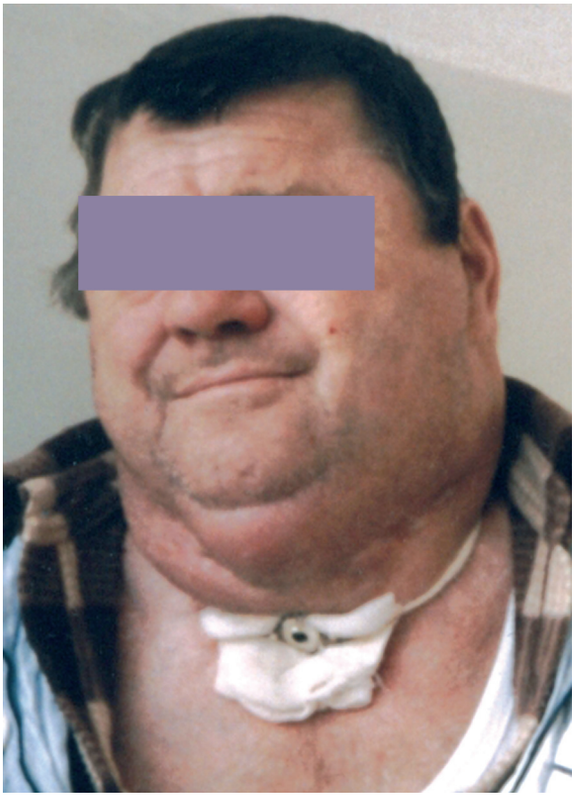

Figure 1: Patient after tracheostomy, head and neck, frontal view.

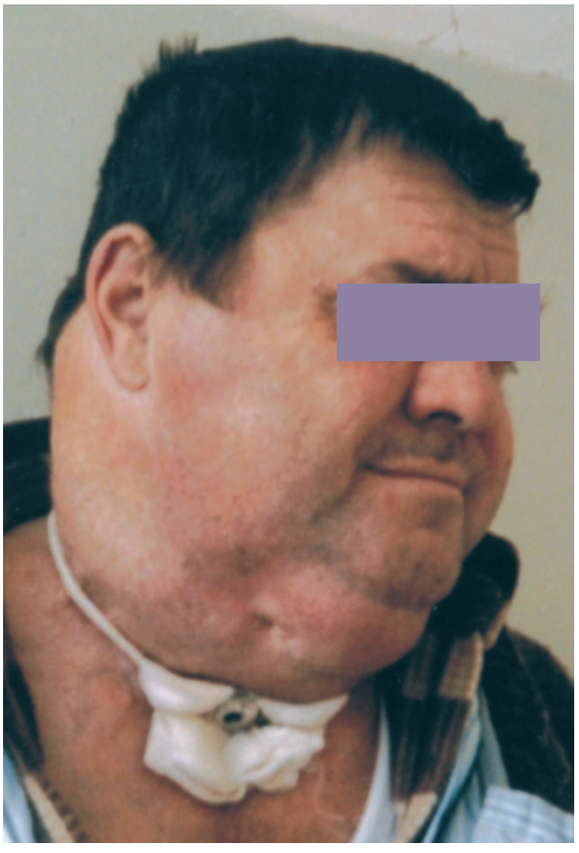

Figure 2: Patient after tracheostomy, head and neck, side view.

plete removal of fat tissue was, however, limited by its infiltrative nature and involvement of the salivary glands and facial nerve. The histological examination revealed mature adipose tissue with an increased fibrocollagenous component. The postoperative course of the patient was uneventful and at the one year follow-up there were no signs of recurrence or functional problems. However due to the partial resection of the adipose tissue a strict long-term follow-up was advised to the patient.

Discussion

Multiple symmetric lipomatosis is a rare disorder of lipid metabolism which is mainly reported in Mediterranean and eastern European populations. The disorder predominantly affects white males (male: female ratio 15: 1). In 60-90% of patients, alcoholism is associated with Madelung's disease[5].

Depending on the anatomic location, according to Donhauser et al, multiple symmetric lipomatosis can be divided into 3 types: a) madelung fatty neck (involving the neck); b) pseudo-athletic type (involving shoulders and back) and; c) gynecoid type (involving the hips and stomach)[6]. Nowadays, 2 clinical phenotypes are recognized: a) type I (Madelung' collar), affecting predominantly males with fat accumulation in the neck, nape of the neck, shoulders, upper arms, and upper back; and b) type II also affecting women although characterized by a superabundant female fat distribution in the upper back, deltoid region, upper arms, hips, and upper thigh region, giving the appearance of simple obesity[7].

The pathogenesis of Madelung's disease 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[8,9]. Recently, it was shown that 80% of patients with HIV-1 infection treated with a protease inhibitor (indinavir or lamivudine) develop this syndrome, probably as a result of the effect of protease inhibitors on sugar and lipid metabolism[10].

Diagnosis is based on the distinctive clinical appearance and imaging. Particularly Magnetic Resonance Imaging (MRI) and CT without contrast administration provide adequate information to the surgeon on the real extent of the disease and on the involvement of vital structures. There are also able to distinguish encapsulated fat (progressive nodular lipomatosis and multiple hereditary lipomas, hibernoma) from Madelung's disease, by means fat suppression and clinical shift imaging[11,12].

Although the fatty accumulation is considered a benign disease, compression of the aerodigestive tract may occur in long lasting cases causing severe respiratory problems. and increased anesthesia related risks[13].

Our patient presented with severe respiratory distress that required an urgent tracheotomy combined with surgical lipectomy. These actions are considered the treatment of choice. However, when it is not possible to completely resect the adipose tissue due to it's accumulation around vital structures or due to the infiltrative behavior of the disease, as it was also observed in our case, recurrences usually occur requiring close long-term follow-up to prevent major complications as are severe dyspnea and dysphagia.

Medical therapy including (β2-agonist - Salbutamol), vitamins and thyroid extracts have been used to control the phenomenon. However, at the moment they seem to have only a auxiliary role next to surgery[8].

In conclusion Madelung's syndrome due to its progressive and occasionally unpredictable behavior has to be considered not only for aesthetic reasons but also for its functional and sometimes life threatening consequences. Type I disease (Madelung collar) staged surgery has to be scheduled in order to avoid serious complications and to obtain substantial improvement in both functional and aesthetic appearance of the affected individual.

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