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Case Report

A Rare Case Report of Made Lung Disease

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Abstract:

Introduction: Madelung's disease, or multiple symmetric lipomatosis, is a rare disorder of fat metabolism characterized by non-encapsulated, symmetrically distributed adipose deposits, predominantly involving the neck, shoulders, and upper trunk. With fewer than 500 cases globally, it remains uncommon in Asia and underreported in India. Though typically benign, the disease can produce significant cosmetic disfigurement, compressive symptoms, and functional limitations. Chronic alcohol intake is the most frequently associated risk factor, while metabolic and endocrine abnormalities may contribute to disease progression.

Methodology: We report the case of a 46-year-old male with a long history of alcohol consumption who presented with progressive, bilateral cervical swellings over one year. Clinical assessment was supported by ultrasonography, computed tomography (CT), and magnetic resonance imaging (MRI), all of which revealed diffuse, symmetric, non-encapsulated adipose proliferation in cervico-facial and parapharyngeal regions. Fine-needle aspiration cytology (FNAC) yielded fatty aspirates, consistent with benign lipomatous pathology, and findings were integrated with radiological features to confirm Madelung's disease.

Results: Imaging demonstrated extensive adipose tissue infiltration in parapharyngeal, retropharyngeal, and pre-vertebral spaces, displacing bilateral parotid glands. MRI confirmed diffuse lipomatous proliferation consistent with symmetric neck lipomatosis. FNAC showed mature adipocytes without atypia, ruling out malignancy. The clinical, radiological, and pathological findings converged toward the diagnosis of Madelung's disease.

Conclusion: This case underscores the diagnostic importance of correlating clinical presentation with radiological and cytological findings in rare fat metabolism disorders. Madelung's disease, though benign, can lead to functional and esthetic complications. Early recognition is paramount, particularly in populations where the disease is rarely reported, as in India. Long- term alcohol consumption remains a significant etiological factor, highlighting the need for counselling on lifestyle modification and close monitoring for progression or systemic associations.

Introduction

Madelung's disease, also known as benign symmetric lipomatosis, Launois-Bensaude syndrome, Brodie syndrome, Buschke disease, or multiple symmetric lipomatosis, is a rare acquired disorder of fat metabolism [1]. First described by Sir Benjamin Brodie in 1846, it was later clinically defined by Otto Wilhelm Madelung, who published detailed observations on 33 patients in 1898 [1, 5].

Subsequently, Launois and Bensaude broadened its clinical characterization by presenting 65 additional cases in the same year [6, 7].

Enzi later classified the disorder into two morphological types based on the distribution of adipose deposits. Type I is distinguished by discrete, symmetrical, non-encapsulated fatty masses that protrude prominently from the body surface, resulting in the characteristic 'pseudo-athletic' appearance. Type II, by contrast, demonstrates diffuse subcutaneous fat deposition, often resembling generalized obesity [8, 9].

Clinically, the condition is marked by multiple, symmetrically distributed lipomas, most often involving the neck, shoulder, upper trunk, and parotid regions. Distinctive topographic patterns have been described: 'hamster cheeks' in the parotid area, 'horse collar' involvement around the cervical region, and 'buffalo hump' formation over the posterior neck. Collectively, these features produce the classical pseudo-athletic body habitus [1, 10].

The disease predominantly affects middle-aged men between the ages of 30 and 60 years [1], with the highest prevalence reported in the Mediterranean region, while cases remain uncommon in Asia [6, 11]. Its etiology is strongly linked to chronic alcohol consumption, although associations with metabolic, endocrine, and hepatic dysfunctions are increasingly

recognized. Clinical manifestations vary and may include dyspnea, dysphagia, polyneuropathy, and progressive muscle weakness. While cosmetic disfigurement often represents the primary clinical concern [1, 4], more than 400 cases have been reported worldwide. Reports from the Indian population remain scarce, providing the basis for the present study.

Case Report: A 46-year male hailing from Barmer presented with swelling around neck. Bilateral symmetrical swelling was present around neck behind ear and angle of mandible for one year. The swelling has increased more in last 6 months. Swelling are firm in consistency and non-tender. No lymphadenopathy seen. Had history of alcohol intake for last 30 years. No history of smoking There is no history of hypertension and diabetes. Other general and systemic examination were unremarkable. This case was referred from the ENT dept of MDM for FNAC to department of Pathology, MDM.

Figure 1: Photographs of the patient showing the swelling

Routine laboratory investigation like Lipid profile and Thyroid were within normal limit.

On USG, diffused fatty proliferation with symmetric lobulated sub cutaneous fatty deposition in a centripetal distribution. Swelling region are non-tender on ultra-sound suggestive of Madelung disease.

On CT, there is excessive fat deposition in deep spaces of neck with extensive adipose tissues seen in parapharyngeal, retropharyngeal parotid and pre-vertebral spaces, both parotids are displaced anteriorly by non-encapsulated fatty masses. These findings are suggestive of symmetric lobulated sub cutaneous fatty deposition in a centripetal distribution in favour of Madelung disease also known as Madelung Launois-Bensaude syndrome.

Magnetic resonance imaging (MRI) revealed a nearly symmetric, non-encapsulated, non-mass-like hypertrophic proliferation of superficial adipose tissue involving the neck and upper trunk, consistent with symmetric neck lipomatosis (Madelung's disease). Subsequently, the patient underwent fine-needle aspiration cytology (FNAC) in our department.

FNAC performed from the palpated mass present at the nape of the neck and angle of mandible which is firm in consistent and non-tender. Mode of aspiration was direct. Yield was fatty aspirate. Under the microscope, Giemsa-stained smears show mature fibro fatty fragments, adipocytes and fat droplets. Impression features suggestive of benign lipomatous lesion likely lipoma with clinical history suggestive of Madelung disease. He was further suggested for histopathological examination.

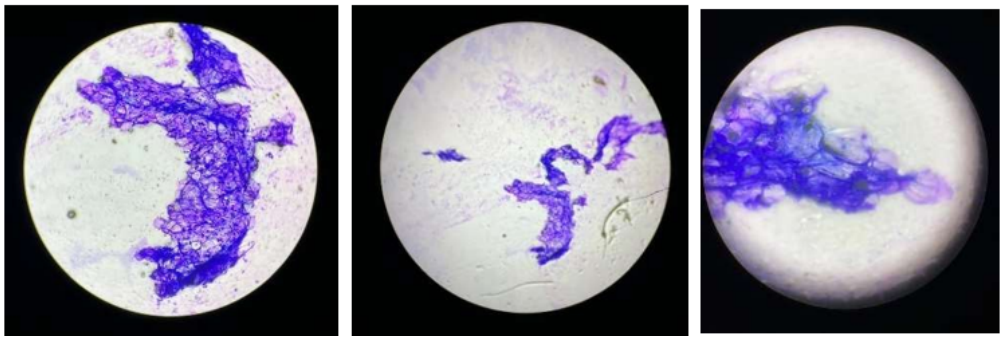
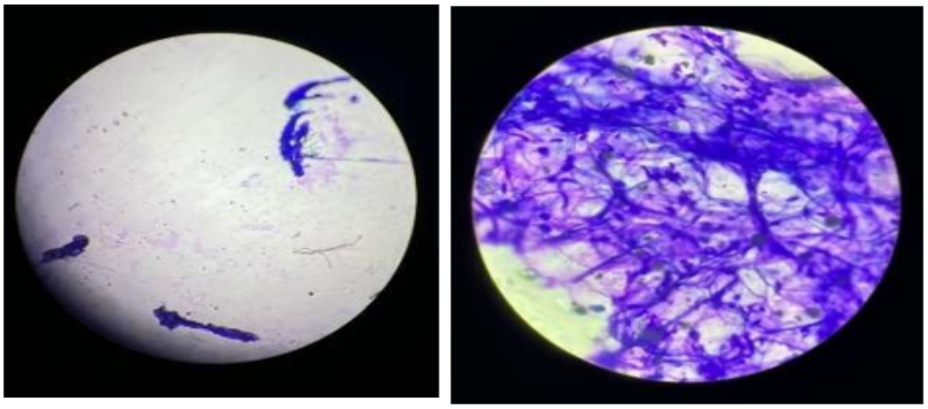


Figure 2: Microscopic examination showing mature fibro fatty fragments, adipocytes and fat droplets



Discussion

Madelung's disease, also referred to as multiple symmetric lipomatosis, is a rare disorder characterized by multiple, symmetric, non-encapsulated proliferations of adipose tissue distributed mainly in the head, neck, upper trunk, shoulders, and proximal arms [1, 2]. These features distinguish it from encapsulated lipomas, which may be either symmetric or asymmetric, and from lipodystrophy syndromes, which typically demonstrate a combination of lipohypertrophy and lipoatrophy [8, 12].

Although Madelung's disease is generally benign, reports of malignant transformation are exceedingly rare.

Chronic alcohol consumption remains the most frequently reported associated factor, though other comorbid conditions such as endocrine disorders, liver disease, upper aerodigestive tract malignancies [6, 15], and metabolic syndromes (including hyperlipidemia, hyperuricemia, type 2 diabetes mellitus, and hypothyroidism) have also been described. While the precise pathogenesis remains unclear, proposed mechanisms include mitochondrial dysfunction in adipose tissue, reduced cytochrome c oxidase activity, catecholamine-mediated fat deposition, impaired adrenergic-stimulated lipolysis, and decreased inducible nitric oxide synthase (iNOS) levels [6, 17]. Histological and molecular studies further suggest that the lipomatous tissue resembles brown adipose tissue (BAT) [6,18], with proliferation of adipocytes rather than hypertrophy of existing cells. Recent molecular analyses have shown hyperactivation of signaling pathways such as AKT, CK2, and ERK1/2 in affected adipose tissue, supporting a proliferative pathophysiology.

Long-term heavy alcohol intake is considered the most significant risk factor, potentially leading to mitochondrial DNA mutations, oxidative injury, and impaired lipid metabolism [6, 20]. Alcohol- induced reduction in β-adrenergic receptor density and activity, along with increased triglyceride synthesis, contributes to abnormal fat accumulation. When combined with high dietary fat intake, this effect is further amplified [6, 21]. Familial cases with possible autosomal dominant inheritance have also been reported, suggesting both acquired and genetic susceptibilities [1, 8].

Clinically, the disease manifests as multiple, painless, slowly enlarging, symmetrical fatty masses, most prominently involving the face and neck due to the complex maxillofacial anatomy. Progressive enlargement can restrict cervical mobility, compromise upper airway patency, and produce compressive symptoms such as dysphagia, sleep-disordered breathing, and, in rare cases, laryngo- tracheal stenosis. Involvement of the parotid and lingual regions may result in facial deformity, restricted tongue mobility, and macroglossia [6, 23]. Neurological manifestations, typically in the form of peripheral neuropathy and

limb weakness, are recognized, while central nervous system involvement remains rare.

Four morphologic subtypes have been described based on fat distribution [8, 22]:

1. Type I: “Madelung’s collar” localized to the cervico-occipital region.
2. Type II: Pseudoathletic type involving the shoulder girdle, deltoids, upper arms, and torso.
3. Type III: Gynecoid type predominantly affecting the pelvic girdle.
4. Type IV: Abdominal type.

The condition is often accompanied by systemic comorbidities, including hepatic dysfunction, hypertension, dyslipidemia, diabetes mellitus, hyperuricemia, hypothyroidism, renal impairment, and obstructive sleep apnea–hypopnea syndrome [6, 8, 24].

Differential diagnoses encompass liposarcoma, multiple familial lipomatosis, Dercum’s disease, neurofibromatosis, drug-induced lipomatosis (secondary to steroids or antiretroviral therapy), angiolipoma, and hibernoma [1, 5]. Accurate diagnosis is made clinically and confirmed through imaging modalities such as computed tomography (CT), magnetic resonance imaging (MRI), and ultrasonography. Fine-needle aspiration cytology (FNAC) or histopathology may be useful in excluding malignant lesions. Typically, growth is indolent and progressive over years, although in some cases, including ours, rapid enlargement may occur within months.

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