

RADIOLOGY CASE OF THE MONTH

Multiple Symmetric Lipomatosis: Madelung's Disease

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A 56 year old African-American man presented to the emergency department with dyspnea and dysphagia with drooling. On his initial evaluation, disproportionate obesity of the face, neck and shoulders were noted. The patient's history was significant for obstructive sleep apnea, end-stage renal disease, alcoholic liver disease, pulmonary hypertension and alcoholic cardiomyopathy. He had multi-decade history of heavy alcohol abuse, but quit drinking two years previously.

FIGURE: CT examination post-contrast. (A and B) Axial sections of the upper and lower neck respectively revealed symmetrical non-encapsulated fat deposits with significant compromise of the airways (airway tube in-place). (C) Coronal section of the neck revealed symmetrical non-encapsulated lipomatosis in the posterior cervical spaces (tracheotomy tube in-place).



What is your diagnosis?
Elucidation is on page 61.

INTERPRETATION OF IMAGES: *Computed Tomography (CT) of the soft tissue of the head and neck showed symmetric, non-encapsulated adipose deposits in the buccal, posterior cervical neck, axillary regions and with significant oropharyngeal narrowing (Figure). There was no compression of the trachea. Further discussion with the patient revealed that the masses in his head and neck had been growing slowly over the past three years and demonstrated on previous images. The adipose deposits sparing the distal extremities in this patient were consistent with so-called Multiple Symmetric Lipomatosis (MSL), also known as Madelung's disease.*

DISCUSSION

This condition occurs primarily in men, and is strongly associated with a history of alcoholism.¹ The pathogenesis is not fully understood, but recent evidence suggests that MSL results from defective noradrenergic regulation of mitochondria in brown fat.^{2,3} Notably, the distribution of excessive adipose tissue in MSL mirrors the distribution of brown fat found in infants. The course of the disease is typically slow progressive growth of adipose deposits. MSL patients also commonly suffer from various neuropathies, especially paraesthesias and autonomic neuropathy. These neuropathies do not correlate with alcohol intake, and are believed to be an intrinsic aspect of the disease process.⁴ MSL is also associated hyperuricemia and sleep apnea.^{4,5} The most effective treatments for MSL is surgical: lipectomy, liposuction, or ultrasound-assisted liposuction.⁶ Treatment indications include: sleep apnea, aerodigestive tract compression, neck pain, and/or aesthetics. Alcohol cessation may help stop the progression of lipomatosis, but regression is rare. Even patients who cease alcohol intake or who were never alcoholics may see progression of the disease.^{2,4}

Our patient's presentation with MSL was notable in that he was African-American. Only one case of MSL has previously been described in an African American.⁷ MSL is believed to be most common in males of Mediterranean descent.⁴ Additionally, while our patient's history of alcoholism and liver disease is consistent with past reports on MSL, his clinical presentation was complicated by his end stage renal disease, pulmonary hypertension and cardiomyopathy. His acute dyspnea and dysphagia resulted from airway compression due in part to the size of his facial and anterior cervical adipose deposits. A sleep study was warranted for this patient, as his oropharyngeal compromise was the cause if his sleep apnea, a known complication of MSL.^{4,5} The presence of apnea could have contribute to his pulmonary hypertension.

There is often a long delay before diagnosis of MSL, which is often confused with obesity. However, MSL is a distinct clinical entity from obesity. Diagnosis is made clinically, based on the distribution of adipose tissue and also the patient's sex, age and history of alcohol use. CT or Magnetic Resonance Imaging (MRI) can confirm the presence of symmetrical un-encapsulated fat deposits in pre-surgical stage.⁸

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