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Case Report

Horse collar: a rare case report of Madelung disorder with review of literature

Rajendra B. Metgudmath[1], Pallavi Kallur[1]*, Virupaxi V. Hattiholi[2]

[1]Department of ENT and HNS, K.L.E.S. Dr. Prabhakar Kore Hospital and Medical Research Centre, J. N. Medical College, KAHER, Belagavi, Karnataka, India
[2]Department of Radiodiagnosis, K.L.E.S. Dr. Prabhakar Kore Hospital and Medical Research Centre, J. N. Medical College, KAHER, Belagavi, Karnataka, India

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***Correspondence:**
Dr. Pallavi Kallur,
E-mail: pallavikallur95@gmail.com

ABSTRACT

Multiple symmetric lipomatosis is a rare disorder of fat metabolism which is characterized with bilateral symmetric and usually painless masses at various anatomical sites. Generally, the patient with this disease presents with cosmetic deformities. Extensive masses are known to have compressive symptoms like breathing difficulty and dysphagia. Imaging plays a pivotal role diagnosing this condition. Treatment options include both surgical and non-surgical modalities along with management of other metabolic disorders like diabetes and abstaining from alcohol intake. Recurrence is common. Given the uncommon nature of this illness and the debates surrounding its management, we report a case of multiple symmetric lipomatosis.

Keywords: Madelung disease, Multiple symmetric lipomatosis, Liposuction, Intralipotherapy

INTRODUCTION

Multiple symmetric lipomatosis is an inherited disorder that is characterized by progressive, painless, highly vascularized, symmetric unencapsulated lipomas in the interscapular region and subcutaneous tissue of the posterior portion of the neck, but it can also be seen in the maxillofacial region, trunk, limbs and other areas.[1,2] The patient's body may have pseudo-athletic-looking fat deposits over the shoulders, back, and chest; parotideomasseteric regions, which resemble the cheek pouches of hamsters; and the neck and nape.[1,2] It is most commonly seen in males, who are of Mediterranean origin.[1] Although very uncommon in the Indian population, few cases have been reported.

The disease may be associated with alcoholic liver disease, diabetes mellitus, glucose intolerance, hyperlipidemia, and hyperuricemia.[3] As the condition worsens, the patient may experience functional anomalies in addition to aesthetic ones which include tingling brought on by nerve compression, dysphagia, decreased cervical spine range of motion and dyspnea.[2] Surgical lipectomy is the current line of management.

CASE REPORT

A 48-year-old male, presented with bilateral lower neck swelling since one year, which has been slowly progressive. He had a history of tingling sensation over the foot. The patient had occasional history of mild pain in the swelling only while working. There was no history of breathing difficulty or difficulty in swallowing or change in voice. The patient was a known diabetic and hypertensive and was on treatment. Patient was a known chronic alcoholic and betel nut chewer daily for 20 years. Clinical examination revealed soft, non-tender swelling occupying the supraclavicular fossa bilaterally (Figure 1). There were no signs of inflammation over the swelling. Examination of oral cavity, oropharynx, laryngopharynx, nose and nasopharynx were normal. There was no significant cervical lymphadenopathy.

Figure 1: Clinical photograph of a patient showing swelling bilaterally in the supraclavicular area.

Magnetic resonance imaging (MRI) scan of the neck and thorax revealed symmetrical, non-encapsulated, non-enhancing T1 and T2 hyperintense tissue showing suppression on fat-saturation (FATSAT) sequences noted in the bilateral supra and infraclavicular regions, predominantly along the medial one third of the clavicle, lateral to the sternocleidomastoid muscle on both the sides suggestive of lipomatous tissue. The lipomatous tissue is seen extending posteriorly into the supraspinatus regions on both sides below the deltoid muscle. There was no evidence of underlying bony erosion. These features were suggestive of Madelung disease (Figure 2). The treatment options were discussed with the patient. The patient refused to have any major interventions because his only complaint was swelling, however he did indicate that he was willing to give up alcohol, strictly manage his diabetes and to be on regular follow-up.

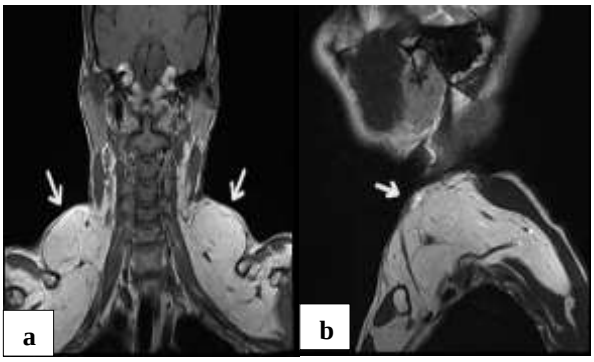

Figure 2 (a and b): MRI of the neck and thorax.

DISCUSSION

Multiple symmetric lipomatosis (MLS) also known by the names Madelung disease/Launois-Bensuade disease is a rare disorder of fat metabolism. The head, neck, and upper torso are the primary areas affected symmetrically by the subcutaneous unencapsulated fatty tissue expansion, which is painless and gradually spreads to the upper extremities.[1] In contrast to typical lipomas, MLS lacks distinct boundaries and a membranous capsule.[4] It is commonly seen in middle-aged males with a male-to-female ratio of 15:1 to 30:1 (Mediterranean population).[1]

High alcohol intake is a contributing factor. It may also be associated with glucose intolerance, dyslipidemia and hyperuricemia.[4] Exact etiopathogenesis is unknown, but it may be attributed to a defect in the metabolism of brown fat cells.[3] The aberrant synthesis of intracellular cyclic adenosine monophosphate (cAMP) caused by noradrenaline activation is the cause of these masses.

The autonomy of fat cells in MSL is caused by a deficiency in adrenergic-stimulation lipolysis.[5] As the catalytic unit of adenyl cyclase is the primary problem, alcoholism appears to cause a disruption in the mitochondrial DNA in the adipose tissue, peripheral nerve, muscle, and central nervous system in addition to a decrease in beta-adrenergic receptors.[5] Pathogenicity at the cellular level has been found in the mitochondria. According to the study done by Plummer et al "the normal lipolysis of brown fat is impeded by impaired mitochondrial respiration, which intensifies the development of midline lipomas".[5]

Enzi classified lipomatosis into two types. one where the fatty tumors remain as bounded masses that protrude from the body surface, with a marked atrophy of the unaffected adipose tissue over the course of the disease; and another where the lipomatous tissue diffuses, affecting the subcutaneous fat layer extensively and giving the patient the external appearance of simple obesity.[6] In 1991, Donhauser categorized the lipomatosis into four types - type 1: horse collar, type 2: pseudo athletic, type 3: gynecoid type and type 4: abdominal type.[7] In the newest classification proposed in 2018, Schiltz et al classified MLS into 3 types which showed type 1 as involving upper body, which was in turn divided into 3 subtypes. Type 1a if only neck is involved, type 1b if neck, shoulder girdle and upper arms are involved and type 1c if neck, shoulder girdle, upper arms and trunk are involved. Type 2 showed involvement of hips, buttocks, upper legs. Type 3 was a general distribution without the involvement of head, forearms, and lower legs.[7] Our patient was classified according to Donhauser classification as type 1c since only neck, upper chest, back, and shoulder girdle were affected.

The assessment of the extent of the adipose tissue deposition; the tracheal compression; the existence of blood vessels within the adipose mass; and the malignant transformation can be achieved with the use of imaging exams, such as computed tomography and magnetic resonance imaging.[1] In addition to the imaging the metabolic profile of the patient must be assessed including but not limited to diabetic profile, and renal parameters like urea, creatinine, and uric acid. Lipid profile and nerve conduction studies may be done to study the progression of neuropathy. [2,3,4]

The mainstay of treatment is surgical management which may be either liposuction or lipectomy.[8] The various methods followed for liposuction are ultrasound-assisted,

tunneling techniques, cervical dissection, powered-assisted, and rhytidectomy approaches. Intralipotherapy has also been suggested by a few authors. It is defined as the procedure in which the lipoid masses are injected with phosphatidylcholine/deoxycholate which helps in limiting the growth of adipose tissue. Various studies have been done to prove the efficacy of the injection. One such study done by Scevola et al confirmed the efficacy of the treatment. Patients who have received injection lipolysis might develop adhesion and thus can pose a threat while operating. Along with surgical management, alcohol abstinence helps in fewer chances of recurrence.[8] Even though wide varieties of surgical techniques are available, surgery should be performed only after careful consideration of risks. The primary criteria for a patient to be eligible for surgery should be functional abnormalities of the digestive and respiratory systems, together with difficulties with day-to-day functioning.[9,10] It is done to prevent premature and radical surgery.

CONCLUSION

Given the uncommon nature of this illness and the debates surrounding its management, it might be worthwhile to discuss our patient's situation.

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