CASE REPORT

Madelung's Disease: A Case Report in an Asian Patient.

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Abstract

A middle-aged man presented with a slow-growing circumscribed mass over both upper arms associated with muscle soreness and lethargy. He has a history of chronic alcohol consumption and imaging studies revealed subcutaneous fat tissue hypertrophy. The characteristics of his disease fit a rare symmetrical lipodystrophy disorder known as Madelung's disease (MD). This is a report detailing the description of the case.

Keywords: *Madelung's disease, multiple symmetrical lipomatosis.*

Introduction

Subcutaneous tissue masses or swellings are one of the most common presentations in primary care. Among common causes of subcutaneous tissue masses are lipoma, liposarcoma, angiolipoma, neurofibroma, and neurofibromatosis. In addition to the clinical history and physical examination, imaging studies may be used to narrow the field of possible diagnoses. A growing subcutaneous tissue should be suspected of malignancy until proven otherwise and should be investigated. A rare cause of subcutaneous tissue such as Madelung's disease should be thought of after all other causes have been ruled out, especially if the swelling is symmetrical. Although it is rare among the Asian population, some cases have been reported.

Case Presentation

A 49-year-old Indian man with underlying hypertension presented to the primary care clinic with symmetrical and large swelling over both arms that increased progressively in size over the past four years. The bilateral arm swelling appeared simultaneously. Initially, the swelling is painless but four months ago, he complained of muscle soreness and lethargy if he used his arms for more than half an hour. This has hindered his daily activities and affected his work as a plumber. He denied any constitutional symptoms or history of trauma before this. On further history, he consumed 3-4 bottles of beer per day for over 30 years, however, he stopped consuming alcohol two years ago. He also has a family history of hypertension and diabetes mellitus.

On examination of his right upper limb, there was circumferential diffuse swelling at the upper arm, measuring 20 x 24cm (Figure 1). Examination of his left upper limb showed circumferential diffuse swelling at the arm, measuring 22 x 20cm (Figure 2). The swellings were firm in consistency, non-tender, and had no skin changes. Both shoulders' range of motion was limited due to pain. Otherwise, all of his elbows, wrists, and fingers' range of motion were complete. His power was 5/5, the sensation was intact, distal pulses palpable, and capillary refilling time < 2s, bilaterally.

A complete checkup consisting of fasting blood sugar and fasting lipid profile were all within normal limits. Given the progressively symptomatic nature of the swelling, malignancy needed to be excluded, and the patient was referred urgently to the Orthopaedics team for further investigation. The patient was seen in the Orthopaedics outpatient clinic, and he was arranged for Magnetic Resonance Imaging (MRI) of his bilateral upper limb urgently. The MRI reported increased subcutaneous fatty tissue deposition without Magnetic Resonance (MR) signs of focal lipomatous or infiltration liposarcomatous changes (Figure 4-6). Subcutaneous fatty tissues hypertrophy secondary to focal, metabolic, or nutritional causes needs to be further investigated. After the MRI was reviewed, the patient was given the option of surgical removal, but he preferred to be monitored for the time being for the progression of the disease. He was given future follow-ups for regular review.

Discussion

MD is a rare condition characterized by symmetrical, nonencapsulated fat lumps on the face, neck, and other parts of the body. Brodie was the first to describe the disease in 1846; Madelung, Launois, and Bensaude followed in their footsteps in 1888 and 1898, respectively [1]. MD is also referred to as Launois-Bensaude syndrome, multiple symmetrical lipomatosis, and benign symmetric lipomatosis. Approximately one in every 25,000 men in the Mediterranean and Europe (with a male-to-female ratio of 15-30:1) have this disease, while 58 instances in Asia have also been reported [2]. In view of it being a rare disease, most of us would likely be unable to recognise it, which might lead to delayed diagnosis and treatment for the patient.

60-90% of people with MD exhibit persistent alcoholism, however the relationship between

alcohol and MD remains uncertain. Even though the method for diagnosing MD is uncertain, alcoholism, hepatic dysfunction, poor glucose tolerance, hyperlipoproteinemia, hyperuricemia, neuropathy, renal tubular acidosis, and hypothyroidism have been associated with the disease [3-7].

There are many hypotheses regarding the pathogenesis of MD. One hypothesis has indicated that the excess fat may be caused by an anomaly in the production and release of the intracellular adenosine monophosphate (cAMP). Beta-adrenergic receptor levels seem to be reduced in those who engage in heavy drinking to an unhealthy degree. Possible secondary reasons include changes in mitochondrial DNA or sympathetic denervation of brown fat adipocytes. High oxidative mitochondrial metabolism and lipolysis are both hallmarks of brown fat. Dysfunctional adenosine triphosphate production may compromise lipoprotein storage, which might therefore lead to oxidative dysfunction [8]. Multiple symmetrical fat deposits in the neck and upper back, with just a little amount of fat deposition elsewhere, are key to determining if the patient has MD. Ultrasound, computerized tomography (CT) scans, and magnetic resonance imaging (MRI) are imaging procedures that enable physicians to determine the degree of fat accumulation in afflicted areas, the compression of underlying tissues, and the existence of blood vessels in fatty tumours [5,9]. In addition, imaging investigations are utilized to rule out other diagnoses and offer information for preoperative surgical planning. In MD, fatty deposits are not encapsulated and are distributed along vascular and muscle planes, whereas they are encapsulated in lipoma. Other disorders, such as angiolipoma, neurofibroma, liposarcoma and lipoblastomatosis, lipodystrophy, lymphoma, neurofibromatosis, and diseases of the salivary glands, should be investigated, because removing these from the list of diagnoses will enable successful treatment [5].

. According to Enzi's classification system, MD is divided into two categories depending on the anatomic fat distribution. When it comes to type 1, fat is symmetrically distributed and concentrated in the neck and shoulders as well as in the supraclavicular triangle and upper arms. While the distribution of fat in type 2 is lopsided and concentrated in the belly and thighs, it is sometimes mistaken for normal obesity [10]. According to Donhauser et al. (1991), the Enzi type 1 is distinguished by the way the neck is distributed; type 2 has a faux athletic aspect; and type 3 has a gynecoid appearance [11]. However, it is worth noting that some persons may have many distribution types. In our patient, no other body parts, such as the neck or upper torso, were affected, as was the case in the previous research. We can most likely classify this patient as Donhauser type 2.

Peripheral neuropathy is widespread in Madelung's disease, particularly as the patient ages, as indicated by histology tests that reveal a loss of more myelinated cells without demyelination or axonal degeneration several years after the appearance of fat lumps. The histology studies revealed that after a few years, the fat lumps result in myelinated cell depletion (no demyelination or axonal degeneration). It can trigger myopathy (loss of muscle power) in the proximal muscles of the arms and legs [12]. Depending on the degree of the condition, fatty tumours in the neck can compress the airway (trachea), voice box (larynx), oesophagus, and carotid blood arteries, producing symptoms such as difficulties breathing (dyspnoea), swallowing (dysphagia), or speaking (dysphonia) [1].

As the illness progresses, the patient's neck mobility may decrease, leading to sleep apnoea. It is crucial to keep in mind that the physical changes in a patient's body produced by excessive fat deposition may have a detrimental effect on their mental health and lead to depressive disorders. The patient has also claimed social loss as a result of reduced mobility and other issues, such as difficulties in performing or retaining employment. In our case, the patient complained of muscle discomfort and fatigue, which limited his movement and prevented him from working.

According to other research, sixteen out of twenty individuals exhibited comparable symptoms [10]. No particular therapy is indicated. Although abstinence from alcohol and weight loss are advantageous, they do not reverse or prevent the disease's progression [5]. This issue is pertinent to this instance since the oedema continues to expand in size even though the patient has stopped drinking. When symptoms appear, surgical resection or liposuction, as well as injectable lipolysis, or both, are indicated for the palliative elimination of fatty tissue [9]. The total recurrence rate of surgical treatments for Madelung disease was 18.3% [2].

Conclusion

The case presented completely fits into the rare symmetrical lipodystrophy known as Madelung's disease. Although it is a rare disease especially among Asians, it is useful for medical practitioners to be knowledgeable of this disease as one of the differential diagnoses of fatty growth. Correct identification will lead to early diagnosis and correct management that would improve patients' morbidity.

Acknowledgement

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Conflict of interest

The authors declare no conflict of interest.

Statement of informed consent

Informed consent was obtained from the patient.

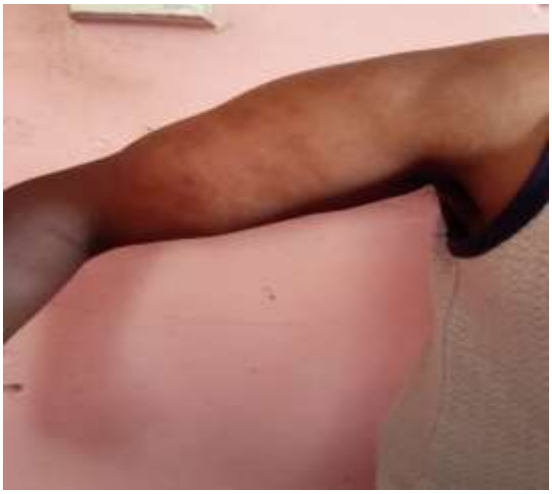

Figure 1. Patient's right arm

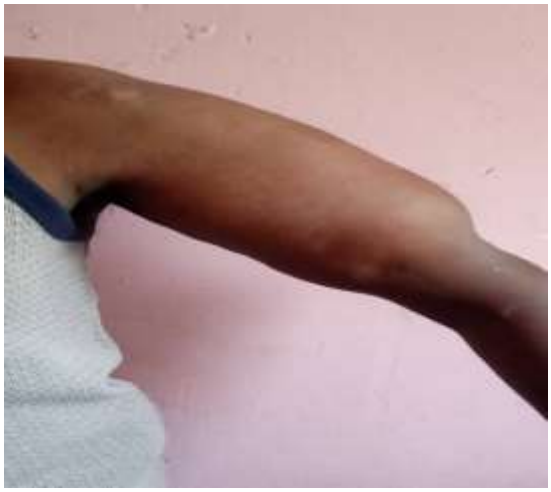

Figure 2. Patient's left arm

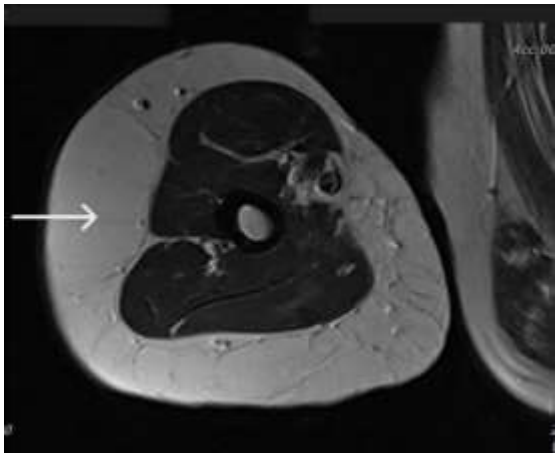

Figure 3. Magnetic resonance of right humerus from axial plane

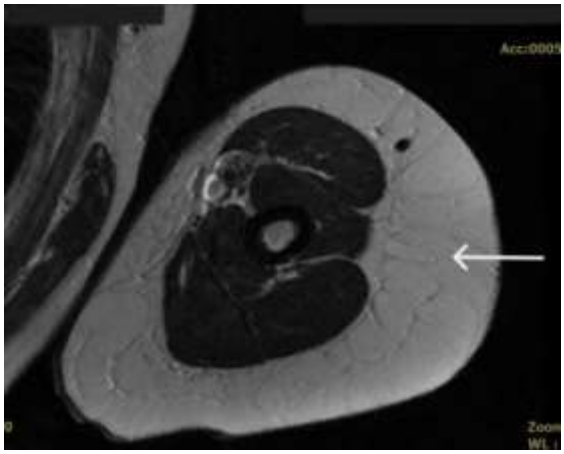

Figure 4. Magnetic resonance of left humerus from axial plane

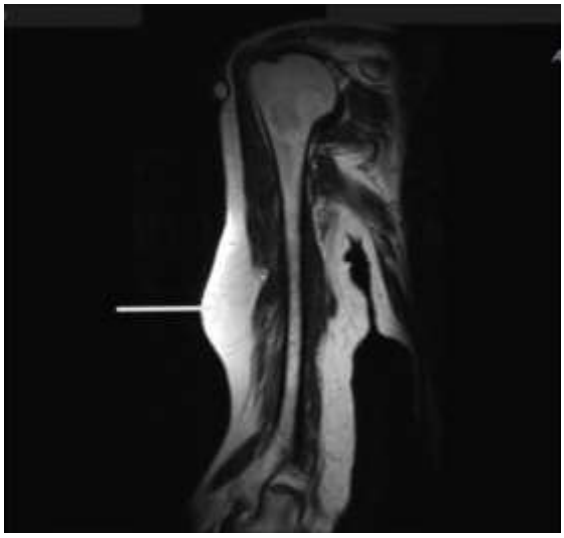

Figure 5. Magnetic resonance of right humerus from coronal plane

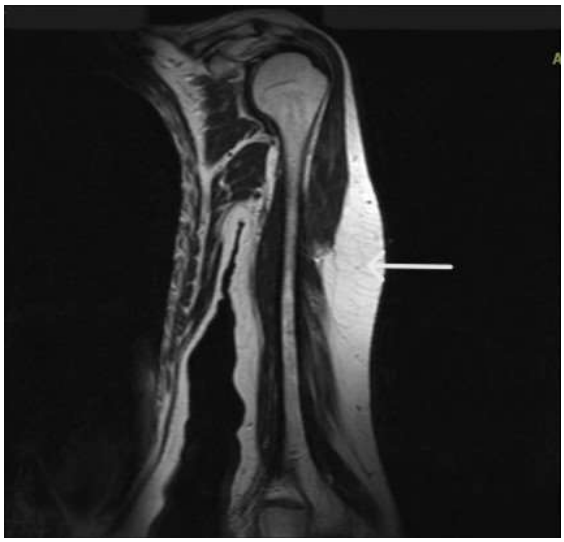

Figure 6. Magnetic resonance of left humerus from coronal plane

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