Madelung’s disease is rare in the Chinese community. It usually happens in patients with alcoholism. The etiology is unknown and probably relates to mitochondrial disease. We report a 46-year-old male who complaint of submental and neck swelling for many years. Computed tomography (CT) shows prominent fat at the anterior neck region extended from bilateral buccal area to the suprasternal notch, and posteriorly at the occipital to upper back region. Surface shaded model demonstrates grotesque symmetrical enlargement of the soft tissue around buccal to anterior neck region with typical “horse collar” and “hamster cheek” appearance which is diagnostic for Madelung’s disease. Preoperative CT and surface shaded model play a major role in the treatment planning of Madelung’s disease.

Key words: Computed tomography (CT), Image processing; Madelung’s disease

Madelung’s disease, also termed as benign symmetric lipomatosis, multiple symmetric lipomatosis and Launois-Bensaude disease, is a rare disease. It is characterized by multiple symmetrical uncapsulated fatty accumulation diffusely involving the head and neck region and upper trunk. Most reported cases come from countries around the Mediterranean Sea. It seems to be relatively rare in Chinese [1,2]. We report a typical case of Madelung’s disease and the preoperative evaluation by computed tomography (CT) and surface shaded model.

CASE REPORT

A 46-year-old male with a history of alcoholism, smoking and betel nut chewing, complained of neck swelling since seven years ago. The neck mass enlarged in recent years and make him uncomfortable, but without dysphagia, odynophagia, or dyspnea. Axial and coronal neck CT was performed by an 8-slice scanner (LightSpeed Plus, General Electric, WI, USA) with slice thickness of 5 mm. It showed prominent fat at the anterior neck region extended from bilateral buccal regions to the suprasternal notch and posteriorly at the occipital to upper back region (Fig 1-2). Surface shaded model was reformatted based on the raw data from axial sections on AW4.0 work station (GE, Milwaukee, WI), and it demonstrated grotesque symmetrical enlargement of the soft tissue around buccal to anterior neck region with “horse collar” and “hamster cheek” appearance (Fig 3). No involvement of fascia or deep structures was seen. His family history was not related to the same disease. The patient underwent surgical excision of profound lymphoareolar tissue surrounding the anterior neck and anterolateral neck. About 420gm of fatty tissue was removed. Histological examination of the adipose tissue showed normal appearance of mature fat cells (Fig 4). He was discharged after ten days. Patient was well for more than two months with stable condition.
DISCUSSION

Cervical lipomatosis was first documented by Brodie [3] in 1846 and Madelung [4] reported a series of 33 patients in 1888. In 1898, Launois and Bensaude [5] described the main characteristics of the syndrome as multiple symmetrical unencapsulated fatty accumulations diffusely involving the head and neck region and upper trunk. The diagnosis is usually made by clinical appearance and history, and confirmed by sonography, computed tomography and fine needle aspiration cytology [6].

Madelung’s disease is usually sporadic. Only a few familiar cases of multiple lipomatosis with autosomal dominant mode of inheritance have been described. It occurs mostly in men between 30 to 60 years old. The male to female ratio is as high as 15 [7]. In most cases, there is a distinct correlation with heavy alcohol use. Most reported cases come from countries around the Mediterranean Sea [8].

The etiology is still indistinct. Mitochondrial respiratory enzyme dysfunction inducing a defect in catecholamine-stimulated lipolysis at membrane level was reported [9]. Molecular genetic analysis revealed multiple deletion of mitochondrial DNA in muscle and adipose cell [10]. Hyperuricemia, gout, polyneuropathy, diabetes, liver disease, glucose intolerance, renal tubular acidosis, hypertension, hypothyroidism and hyperlipidemia have occasionally been identified in these patients. Although these metabolic abnormalities may be related to excessive fat deposits, many are thought to be secondary to alcoholism [7].

Fat deposition is most predominant in the subcutaneous tissue of the neck posteriorly, and deep to the sternomastoid and trapezius muscles. It was also commonly seen in the supraclavicular fossa, around the paraspinal muscles, in the anterior neck above and below the level of the thyroid. However, the site of maximum deposition has been reported in the soft tissue at lateral aspect of neck [11].

Ultrasoundography usually revealed diffuse heterogeneous echogenic lesion that blended with the subcutaneous fat layer with normal anatomical structures such as vessels and lymph nodes [1]. Sonography combine with fine-needle biopsy provide clinicians some informations, but it is unable to define the degree of infiltration.

CT examination illustrates prominent, symmetrical masses at submental, periauricular, suboccipital, supraclavicular, and axillary areas, composed of symmetrically arranged adipose tissue, which was not enclosed within a membranous capsule with very distinct boundaries, as in this case. CT scan can detect compression of trachea and relationship of fat and all major vessels in the neck and mediastinum [12,13]. The case we reported here was the first case demonstrated by surface shaded model.

The MR findings of Madelung’s disease is much like CT findings. Fat is exquisitely demonstrated on MR imaging because of its high signal intensity on short-repetition-time short-echo-time-weight image [14]. Fibrous component is more prominent on MRI, but CT could confirm the diagnosis and the extent of the disease [1].

Preoperative imaging plays an important role in defining the extent and distribution of fat, in locating the major head and neck vessels, in depicting tracheal narrowing and deformity, and in detecting incidental lesions in the neck. Imaging is also useful to confirm the diagnosis in clinically doubtful cases and to assess progression of disease [11].
Because of the benign nature, most patients are treated for cosmetic reasons, but symptoms of respiratory distress, hoarseness and limited neck motion should be evaluated. The effect of medical treatment such as beta2 agonist is uncertain [15], and abstinence from alcohol does not cause spontaneous regression. Because of the infiltrative nature of the uncapsulated fatty deposits and the frequent recurrence after surgical treatment, debulking of tumors or liposuction is recommended. However, complete surgical resection of infiltrated fat is difficult and not essential [16,17,18].

In conclusion, preoperative CT and surface shaded model may play a major role in the treatment planning of Madelung disease, which is rare in Chinese.

**REFERENCES**

梅德倫疾病之電腦斷層及表面重組表徵：病例報告

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梅德倫疾病是一種罕見的疾病，在東方人中更少見。患者多有酒癮，其病理機制尚未明確，可能與粒腺體的基因缺陷有關。我們報告一名四十六歲男性病患，因多年頸部及下顎腫大來求診，電腦斷層顯現患部呈現均勻脂肪組織浸潤，並未侵犯深層器官，經表面重組後可見兩頰、下顎及頸部對稱性腫大，為典型梅德倫疾病症狀。除了傳統電腦斷層掃描外，表面重組亦可提供三度空間的術前評估。

關鍵詞：電腦斷層，表面重組；梅德倫疾病