Madelung disease: case report of a rare disorder and review of literature

Poster No.: P-0036
Congress: ESSR 2015
Type: Educational Poster
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Keywords: Head and neck, Musculoskeletal soft tissue, Soft tissues / Skin, CT, MR, Ultrasound, Education, Epidemiology
DOI: 10.1594/essr2015/P-0036

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Learning objectives

The learning objectives of this poster is to present a case report on Madelung Disease and review of literature.
Background

77 year old male non-alcoholic presented to the out-patient clinic with a history of slowly growing abnormality in the neck, finding it difficult to close the upper buttons of his shirt, and difficulty in tying the neck tie.

Clinical diagnosis was of a goitre and ultrasound examination was requested. Ultrasound examination revealed a normal thyroid gland. However, a large amount of hyperechoic tissue was noted in the neck. CT Scan of the neck with contrast was performed. CT scan showed extensive fatty tissue in the all compartments of the neck extending up to the sternal notch. No focal mass lesion noted. No adenopathy. The findings were consistent with the diagnosis of Madelung disease.

**Madelung disease** is a rare disorder characterized by benign multiple symmetric lipomas involving the upper trunk and neck. The etiology is unknown - mutations of mitochondrial DNA {DNA (A8344G)} and alcoholism (60-90 %) have been associated with it. It is most commonly seen in Mediterranean population between the third and fifth decade. Male to female ratio is 15:1. Madelung disease features in the rare disease registry in Europe and as other countries

Madelung’s disease was initially described by Sir Benjamin Brodie in 1846. In 1888, Otto Madelung reported the first series of 33 patients with lipomas associated with alcoholism, and in the next year Launois and Bansaude presented a total of 65 patients with similar features. Approximately 200 patients have been reported in the literature.

Besides Madelung’s disease, other names that have been attributed to this condition, including benign symmetric lipomatosis, Launois- Bansaude disease, multiple symmetric lipomatosis, lipomatosis simplex indolens, and symmetric adenolipomatosis
The condition characterized by the multiple, non-encapsulated, infiltrative lipomas located symmetrically on the trunk, neck, and proximal parts of the limbs. The fatty tissue accumulates gradually. Areas of deposition include the anterior/posterior subcutaneous tissue of the neck, deep to sternocleidomastoid and trapezius muscles, posterior cervical triangle and around the salivary glands. There is physical deformity, may lead to pain, decreased mobility of the neck, peripheral neuropathy. The fatty tissue can infiltrate spaces between adjacent structures, leading to decreased mobility of the neck, dysphagia, superior vena cava syndrome. In rare circumstances lipomas may become malignant {2 cases reported} however they are mostly benign.

Enzi et al. described two types of lipomatosis based on the distribution of fat tissue: **TYPE I** is characterized by lipomas located in the nape of the neck, the supraclavicular and deltoid regions (Madelung's collar), while in **TYPE II** lipomatosis, fat tissue diffuses extensively in to the subcutaneous fat layer giving appearance of obesity.

The disease occurs more commonly in middle-aged men. Other features of include polyneuropathy, proximal muscle weakness, and fractures of small bones. Upper airway obstruction due to compression by lipomas and sudden heart death are the most frequent causes of fatality.

Diagnosis is usually clinical as the changes are self evident. Imaging by ultrasound/ CT is simple, the latter showing the extent of the fat deposition. Microscopically, the findings are not different from normal fatty tissue.

There is no effective treatment, though alcohol abstinence and surgical removal are current approaches. The lesion does not regress spontaneously. Ultrasound guided liposuction has been attempted at certain institutions { Ref 10}

**Differential diagnoses** includes--lipoma, hibernoma, hygroma, branchial cyst, liposarcomas, massive cervical lymphadenopathy, truncal obesity, Cushing syndrome, and Goiter, inflammation of the salivary glands, particularly the submandibular glands

Madelung's Disease - Literature Review


**Images in clinical medicine. Madelung's disease**

Ampollini L, Carbognani P

**Neuropathy in multiple symmetric lipomatosis. Madelung’s disease.**

Pollock M1, Nicholson Gl, Nukada H, Cameron S, Frankish P.

Looks into the fact that the neuropathy in Madelung’s disease is generally attributed to alcoholism; In this study, sural nerve biopsies from these patients revealed an absence of acute axonal degeneration, instead the pathology supports the view that the neuropathy of Madelung's disease is not alcohol-induced but that a chronic distal axonopathy is an integral part of the syndrome.

**Journal Impact Factor: 10.226**

3) *RSNA Case 143: Madelung disease. Radiology.*

Mark S. Landis, MD, MSc, Roya Etemad-Rezai, MD, FRCPC, Karan Shetty, MD, FRCPC, Mark Goldszmidt, MD, FRCPC

Case Report and Imaging review of Madelung’s Syndrome

**Journal Impact factor: 6.214**


**Madelung’s disease involving the tongue.**

Vargas-Díez E1, Daudén E, Jones-Caballero M, García-Díez A.

Reports the third case of Madelung's syndrome with involvement of the tongue in the form of macroglossia

**Journal Impact Factor: 5.004**

Head and neck cancers associated with Madelung's disease.

Chan ES1, Ahuja AT, King AD, Lau WY

Two of the eight patients with Madelung's Syndrome reviewed developed aerodigestive symptoms and were subsequently found to have head and neck cancers. These two patients are described. A mechanism possible in this group of patients is the synergistic effect of smoking and alcohol abuse as risk factors for both Madelung's disease and malignant tumors of the airway.

Impact Factor: 3.943


Madelung disease: distribution of cervical fat and preoperative findings at sonography, MR, and CT.

Ahuja AT1, King AD, Chan ES, Kew J, Lam WW, Sun PM, King W, Metreweli C.

Documents the distribution of excess fat in the neck and to determine the preoperative role of sonography, CT, and MR imaging in patients with Madelung disease

Journal Impact Factor: 3.675


Borriello M1, Lucidi A, Carbone A, Iannone V, Ferrandina G.

Reports a case of liposarcomatous transformation of Madelung's disease in a 59-year-old Italian woman with a coincidental diagnosis of breast cancer

Journal Impact Factor: 2.411


Madelung's disease: case report and discussion of treatment options.
Adamo C1, Vescio G, Battaglia M, Gallelli G, Musella S.

An outline from a Plastic Surgery point of view focusing on treatment options - describes surgical debulking, liposuction for smaller lesions, how using a ‘facelift pattern’ for skin incision and removal can lead to optimal cosmetic results.

Journal Impact Factor: 1.458

Madelung's disease associated the proximal myopathy in a young non-alcoholic male was reported by CJ S Chandran et al { Ref: 8}

19 cases of Madelung's disease with upper Airway obstruction were reported according to B Ojo et al who presented such case at Mount Sinai School of Medicine, New York

A Tufan et al. { Ref 11}describe an unusual case of Madelung's disease with multiple atypical fractures in a middle aged man with a long standing history of ethanol abuse.

The patient's initial presentation was mostly related to the neurological manifestations. He presented with paresthesia, apparent proximal muscle weakness and palpitations, diarrhea, and flushing. He had no complaints related to the lipomatosi, probably due to their noncritical locations. The most striking feature of the patient was multiple atypical spontaneous fractures. He denied any previous major trauma or falls, as well as symptoms related to the fractures. Anaesthesia secondary to neuropathy or analgesic effect of ethanol use by itself might have contributed to the lack of fracture symptoms. Such widespread fractures might have been facilitated by the long-term use of topical potent corticosteroid and nutritional deficiencies secondary to chronic alcoholism.

The first case of Madelung's disease accompanied by Klinefelter's syndrome, but they cannot state whether there is a shared genetic or metabolic basis for the two diseases or whether they randomly co-occurred. Klinefelter's syndrome is a sex chromosome abnormality.{ Ref 9}

C Shetty et al report this rare case in a 8 year old girl{ only the second case reported in children}{ Ref 12}
Fig. 1: Axial CT image: Arrows pointing to the extensive fatty tissue in the neck

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**Fig. 2:** Axial images showing the fatty tissue in the compartments of the neck

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Fig. 3: Coronal recon showing the extent of fatty tissue

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Fig. 4: Sagittal recon showing the anterior & posterior extension

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Fig. 5: Further details seen on coronal recon image

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Fig. 6: 3D and Volume rendering images showing the neck swelling

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Fig. 7: Neck swelling seen on 3D volume rendered image

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Conclusion

Madelung disease is a rare disease, around 200 cases reported worldwide and is even rarer in children (only two cases reported).

Madelung disease should be included in the differential diagnosis of the cause of neck masses.
References

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