Madelung’s Disease: A Report of a Rare Case with an Atypical Presentation

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Abstract
Madelung’s disease is a rare condition characterized by the symmetric deposition of non-encapsulated lipomas throughout the body. This article reports an atypical presentation of Madelung’s disease and provides important epidemiologic, diagnostic, and treatment options that contribute to the ever-growing body of medical literature regarding this uncommon disease.

Introduction
Madelung’s disease, also known as multiple symmetric lipomatosis, is a rare condition of fat metabolism that results in the symmetrical deposition of adipose tissue most commonly around the mental, cervical, supraclavicular and upper-extremity regions. The disease was first reported in 1846 and has since troubled the medical community due to its unknown etiology and pathophysiology. Literature on this rare disease reports that men are most commonly affected, at a ratio of 15:1.3,4 Furthermore, disease manifestation is often seen in those of Mediterranean descent, suggesting a possible genetic inheritance pattern. Up to 90% of patients with Madelung’s disease suffer from chronic alcoholism as well.1,2,5,6 Common comorbid conditions such as hyperlipidemia, hyperuricemia and diabetes have been reported in cases of Madelung’s disease.2,7 As such, the disease process can be associated with both environmental and genetic predispositions, but a definitive etiology has not yet been identified.

As the disease most commonly presents in the cervical and upper-extremity regions, the typical presentation of Madelung’s disease is a male patient in his fourth or fifth decade complaining of progressive dysphagia, dyspnea, myopathy and paresthesia, which are due to mass effect of the adipose deposition on the vascular and anatomic structures in these respective regions.4 The prognosis of Madelung’s disease is based upon infiltration of adipose tissue into vascular and anatomic structures. The most common cause of mortality in these patients is sudden cardiac death due to mass effect of tumors on autonomic nerve fibers coursing through the superior thoracic aperture.8

Case Presentation
The patient was a 59-year-old male of Indian descent who was referred by rheumatology for evaluation of profound adipose deposition in the bilateral arms. The patient reported this first occurred in May 2015, following a three-week hospital stay for Henoch-Schönlein purpura. The patient denied any pain or tenderness in the lesions but did note intermittent swelling localized to the upper extremities. Family history was negative for any similar conditions. Upon further questioning, the patient noted that he was a social alcohol drinker and past smoker.

On physical inspection, symmetrical enlargements of the bilateral upper extremities were observed (Figures 1 and 2). The masses were soft, homogeneous, and non-edematous to palpation, with overlying atrophic epidermis with areas of telangiectasia (Figure 2). The neck and supraclavicular areas were completely spared, suggesting an atypical presentation of this disease. A tissue punch biopsy of the proximal segment of the anterior surface of the patient’s right upper arm was reported as a lipoma with focal lipodystrophic changes (Figures 3 and 4), which, combined with the patient’s presentation, was diagnostic of Madelung’s disease. Due to the lack of symptoms from these adipose deposits over the course of two years, imaging studies were not performed. The patient was referred to plastic surgery for liposuction of the upper extremities.

Discussion
This report describes a highly atypical presentation of Madelung’s disease. First, while Mediterranean descent is most characteristic, our patient was native to India. Cases of Madelung’s have been reported only rarely in patients of Asian descent.9,10 Second, the patient’s physical presentation of this disease was not typical of reported cases. Most often, Madelung’s disease presents first and foremost with adipose deposition around the cervical and supraclavicular regions. Along with infiltrations in these regions, adipose tissue can be found along the upper extremities. The disease is rarely reported to occur with adipose tissue deposits isolated strictly within the upper extremities, with no involvement of the neck region. Third, the most common comorbid condition associated with this disease is a history of chronic alcohol abuse. Our patient had no history of alcohol abuse and described himself as strictly a social drinker. Thus, our case demonstrates that Madelung’s disease can present in patients even in the absence of common associated risk factors.

Diagnosis of Madelung’s disease is made by a thorough history and physical exam, a clinical knowledge of the disease, and clinical suspicion, and it is confirmed by biopsy of the tumors to confirm non-encapsulated lipomas. Imaging studies like CT and MRI are commonly ordered.
to assess tumor infiltration in symptomatic patients, although MRI is preferred because it is more sensitive in delineating soft-tissue masses. In this case, imaging studies were not ordered due to the chronic, two-year, asymptomatic history of the tumors. In instances where patients present with paresthesia, pain, or other sensory changes, imaging should be considered to evaluate for potential nerve-root compression. The diagnosis of Madelung’s disease is often missed because of lack of awareness of the disease and because it can present similarly to many other common disease processes. For example, in patients presenting with symmetric fatty deposits in the neck and upper-extremity region, physicians often attribute the condition to obesity and Cushing’s syndrome. However, the fatty tumors of Madelung’s disease do not decrease in size with exercise and weight loss, which helps differentiate them from the adipose deposits in obesity. Madelung’s tumors often remain even in patients with cachexia. Furthermore, a patient’s history of prescription oral steroid use and serum cortisol levels on laboratory investigation can help to distinguish it from Cushing’s syndrome.

Lack of extensive medical literature regarding Madelung’s disease results in lack of diagnosis of the condition. Most commonly, patients are diagnosed based on their clinical history of a slowly growing, symmetric adipose mass and by tissue biopsy, which is crucial to diagnosis, showing non-encapsulated lipomas. First-line treatment is surgical excision or liposuction, with liposuction non-encapsulated lipomas. First-line treatment is slowly growing, symmetric adipose mass and by deposition and penetration of anatomic and vascular structures, resulting in a potentially benign or life-threatening condition. Hopefully, as more cases are reported in the literature, we can begin to investigate other possible associations with Madelung’s disease, such as Henoch-Schönlein purpura (as seen in our case), and alternative treatment protocols.

Conclusion
This case illustrates the importance of considering Madelung’s disease in any patient presenting with a progressive increase in subcutaneous mass in a symmetrical distribution, with or without associated comorbid conditions. Prognosis is entirely dependent upon the distribution of adipose deposition and penetration of anatomic and vascular structures, resulting in a potentially benign or life-threatening condition. Hopefully, as more cases are reported in the literature, we can begin to investigate other possible associations with Madelung’s disease, such as Henoch-Schönlein purpura (as seen in our case), and alternative treatment protocols.

References

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