Madelung’s disease as a rare cause of obstructive sleep apnea*., **

Doença de Madelung como causa rara de apneia obstrutiva do sono

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Abstract

Madelung’s disease, or multiple symmetric lipomatosis, is a rare disease, characterized by accumulation of unencapsulated fat, generally located symmetrically around the neck and shoulders. Here, we present the case of a patient with diffuse lipomatosis accompanied by obstructive sleep apnea due to cervical involvement and facial deformity, which made it necessary to use nasal pillows for ventilation. The patient was hospitalized with a diagnosis of pneumonia and required noninvasive ventilation due to severe hypercapnia. A brief review of the literature was made, and we describe and discuss the investigation of this rare clinical case.

Keywords: Lipomatosis, multiple symmetrical; Sleep apnea, obstructive; Continuous positive airway pressure; Masks.

Resumo

A doença de Madelung ou lipomatose simétrica múltipla é uma doença rara, caracterizada pelo acúmulo de tecido adiposo não-encapsulado, localizado simetricamente ao redor do pescoço e na região escapular. Os autores apresentam um caso de associação entre lipomatose difusa e apneia obstrutiva do sono, devido ao envolvimento cervical e à deformação facial que exigiu a utilização de almofadas nasais para a ventilação. O doente foi admitido com o diagnóstico de pneumonia com necessidade de ventilação não-invasiva devido à hipercapnia grave. É apresentada uma breve revisão da literatura, e descrevemos e discutimos a investigação clínica deste raro caso clínico.

Descritores: Lipomatose simétrica múltipla; Apneia do sono tipo obstrutiva; Pressão positiva contínua nas vias aéreas; Máscaras.

Introduction

Madelung’s disease or multiple symmetric lipomatosis is a rare disease, which is characterized by accumulation of unencapsulated fat, generally located symmetrically around the neck and shoulders. The disease was first documented in 1846 by Brodie, who was followed by Madelung in 1888 and by Launois & Bensaude in 1898.1,2 It has been referred to by other names, such as lipoma anulare colli, morbus Launois-Bensaude, horse collar, multiple symmetric lipomatosis and benign symmetric lipomatosis. In patients with masses increasing the neck diameter, the disease gradually assumes the appearance of a horse collar.2 Although the etiology is unknown, it is often seen in patients with a history of chronic alcoholism, and it is usually accompanied by hyperuricemia, dyslipidemia, macrocytic anemia, peripheral neuropathy, glucose intolerance, renal tubular acidosis and alcohol use.1-5 These fatty masses cause facial disfigurement and are accompanied by digestive and respiratory symptoms. Most patients with Madelung’s disease present reduced neck mobility, limited opening of the mouth and compression of vital structures, causing serious problems with respiration and orotracheal intubation. Symptomatic
untimely deaths. Pulmonologists, maintaining their tradition of combating dreaded diseases, such as tuberculosis, as well as those that are insidious, such as smoking, should be challenged to transform OSAS from a still unknown disease into a treatable cause of morbidity and mortality for patients and bystanders.”

Here, we present a case of diffuse lipomatosis associated with obstructive sleep apnea...
due to cervical and tracheal involvement, and with facial deformity causing problems in the patient’s adaptation to noninvasive ventilation.

Case report

A 62-year-old white male, admitted to the ER with complaints of cough, sputum production and progressive dyspnea for 30 days. He had been diagnosed with Madelung’s disease at the age of 30, having undergone plastic surgery for lipoma removal on numerous occasions. He also had a history of diabetes, alcohol abuse, chronic liver disease and esophageal varices. The patient had been submitted to elective splenectomy five years prior and required invasive mechanical ventilation for 2 days in the ICU. The patient was tachypneic and cyanotic at admission, with an SpO₂ of 84%. A chest X-ray revealed a diffuse bilateral infiltrate without evident consolidation.

The blood gas analysis showed PaCO₂ of 64 mmHg, PaO₂ of 49 mmHg and normal pH. The patient also presented with high acute inflammatory parameters, leukocytosis and high C-reactive protein levels, suggestive of a respiratory infection.

Admitted to the respiratory ward, the patient started treatment with antibiotics and was submitted to continuous bilevel positive air pressure (BiPAP) ventilation, demanding high level pressures to maintain SpO₂ > 90% and to normalize PaCO₂—inspiratory positive air pressure (IPAP): 24 cmH₂O; expiratory positive air pressure (EPAP): 8 cmH₂O.

The clinical status of the patient progressively improved, with subsequent reduction in BiPAP pressures and in the use of the ventilator, which was interrupted on post-admission day 6 due to the appearance of facial ulcerations caused by the nasal mask. During the first night without noninvasive ventilation, there were long periods of oxygen desaturation (SpO₂ < 88%) and snoring. The patient was then submitted to cervicofacial CT (Figures 1 and 2) and a cardiorespiratory sleep study.

The sleep study lasted approximately 8 h, revealing severe obstructive sleep apnea (apnea-hypopnea index: 37 events/h; obstructive apnea index: 29 events/h; median oxygen saturation during sleep: 88%; oxygen desaturation index: 51%/h; and minimum SpO₂: 51%). The CT scan revealed various areas of fat deposition in the mediastinum, causing a reduction in the trachea caliber of 1 cm transversally and 4 cm antero-posteriorly. Continuous positive airway pressure (CPAP) titration was carried out on the second night, and the patient was discharged. At home, the patient remained on BiPAP ventilation (EPAP: 8 cmH₂O; IPAP: 16 cmH₂O), delivered via nasal pillows (Figure 3).

The cardiorespiratory sleep study was repeated four weeks later, and the results confirmed the diagnosis of severe OSAS, with values similar to those obtained in the first sleep study.

Discussion

In the present case, the lipomas caused an enlargement of the neck circumference and a symptomatic reduction in the trachea diameter with airway impairment, especially during the night, causing several obstructive sleep apnea events. Since this patient had a history of plastic surgery with recurrence of the lipomas, a surgical solution was not the first choice of treatments, the use of CPAP being preferred.

The facial deformity, which impaired the use of nasal or facial masks, presented a considerable challenge. During the first days on the ward using a nasal mask, the patient presented multiple facial lesions and skin ulceration. The
solution was to use nasal pillows, which increase comfort with a lower risk of leaks, promoting adherence of the patient to the treatment.

In our review of the literature (Medline), we found only a few case reports of patients with Madelung’s disease and OSAS,[20-23] confirming the rarity of this clinical situation and alerting us to the need for sleep screening in patients with Madelung’s disease due to the risk of OSAS.

References


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