ABSTRACT

Educational Objective: At the conclusion of this presentation, the participants should be able to discuss the presentation and management of Madelung's disease, including the rare case of upper airway obstruction.

Objectives: We present the case of a patient with Madelung's disease presenting with acute upper airway obstruction who was found to have mediastinal and laryngeal involvement of the disease. We also review the literature pertaining to this unusual presentation.

Study Design: Case report and literature review.

Conclusions: Madelung's disease is a rare disorder characterized by the presence of multiple, symmetric, non-encapsulated lipomas involving various parts of the body. The deeper soft tissues of the neck can be involved, which can lead to acute upper airway obstruction. Only 19 cases of laryngeal involvement and 5 cases associated with obstructive sleep apnea have been reported in the literature. To our knowledge, this is the first report with illustrations of endolaryngeal findings.

CASE REPORT

The patient is a 62 y/o man with a history of alcohol dependence and obstructive sleep apnea who was initially managed at another facility for alcohol withdrawal, stridor, and eventually, respiratory failure requiring intubation. The patient was transferred to our facility for further management.

On admission, the patient had a weight of 58.7 kg, a height of 1.57.5 cm, and a BMI of 23.7. Physical exam was notable for bilateral cervical lipomatous masses. MRI (Fig. 1) and CT of the neck demonstrated diffuse lipomatosis extending from the superior mediastinum into the anterior and posterior aspects of the neck and into the retropharyngeal and masticator spaces, consistent with Madelung's disease. MRI was also notable for fat deposits between the thyroid and thyroid cartilage, and surrounding the trachea but the trachea was patent.

A tracheostomy with debulking of cervical fat was performed upon transfer to our facility. Intraoperative findings were notable for fat deposits within the deep fascial planes of the neck and directly overlying the trachea. Direct laryngoscopy revealed fatty submucosal infiltration of the larynx, including the aryepiglottic folds, arytenoids, and false cords, as well as the piriform sinuses (Fig. 2). Pathological analysis of the resected specimens revealed multiple irregular to partly circumscribed yellow-tan fibroadipose tissue.

The post-operative course was unremarkable and the patient remains tracheostomy dependent.

DISCUSSION

Madelung's disease:
- Multiple, symmetrical, non-encapsulated fat deposits around the cheek, neck, shoulder girdle, and trunk.
- There have been about 200 cases reported in the literature with higher prevalence of the disease in the Mediterranean region.
- A history of alcohol use is seen in 60-90% of cases.

Etiology:
- Unknown but several hypotheses:
  - Abnormality in adrenergic-stimulation of lipolysis
  - Mitochondrial dysfunction
  - Defect in the noradrenergic modulation of proliferation and differentiation of brown adipocytes

Presentation:
- Often asymptomatic or patients may present with cosmetic deformity or limited neck range of motion.
- Rarely, patients present later in the course of disease with aerodigestive problems characterized by dysphagia, stridor, dysphonia, and dysphagia due to mediastinal involvement (Table 1).

Diagnosis:
- Clinical diagnosis, but imaging with CT or MRI can be helpful in identifying patients with mediastinal or airway involvement.
- A biopsy is needed to exclude liposarcoma.
- Differential diagnoses include: lipoma, hibernoma, hygroma, branchiogenic clef cyst, liposarcomas, massive cervical lymphadenopathy, trunca obesity, Cushing syndrome, and thyroid goiter.
- Airway obstruction can occur from mediastinal involvement of the disease, external laryngotracheal compression, or from laryngeal involvement either via direct compression or invasion of the recurrent laryngeal nerve.
- Fat deposits in the pharyngeal spaces can be a risk factor for obstructive sleep apnea (OSA) with 5 reported cases of this association in medical search engines.

Management:
- Alcohol abstinence is the only non-surgical management option that is partly effective but does not decrease existing masses.
- The surgical options are debulking lipoectomy or a suction-assisted lipoectomy.
- The lipomas do not spontaneously involute, making airway involvement a constant risk even in patients who have undergone multiple surgical resections.

CONCLUSION

To our knowledge, there have only been 19 reported cases in the English literature of direct laryngeal involvement. Our case adds to this literature and highlights the association of Madelung's disease with OSA.

REFERENCES