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Esophageal leiomyoma incidentally recognized during an acute attack of hereditary angioneurotic edema

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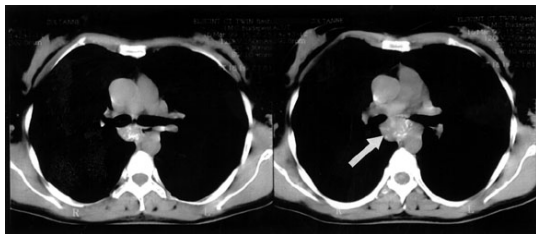
Hereditary angioneurotic edema (HAE)—an autosomal dominant disorder—results from deficiency or dysfunction of the C1-esterase inhibitor (C1-INH). It is characterized by recurrent attacks of edema in the subcutaneous and submucosal tissues of the upper airways and the gastrointestinal tract in particular.¹ Esophageal leiomyomatosis is a rare, benign condition characterized by proliferation of esophageal smooth muscle. More than 50% of patients with esophageal leiomyoma are symptom free.² This condition can occur either sporadically or as an autosomal dominant trait. It is commonly associated with other hereditary diseases (eg, Alport syndrome).³ In the case presented here, however, esophageal leiomyoma coincided with another hereditary disorder, HAE. Of interest, an acute edematous attack of the latter allowed diagnosis of the otherwise subclinical esophageal lesion.

A 41-year-old white woman had had recurrent attacks of subcutaneous and submucosal edema since the age of 10 years. Laryngeal edema occurred after dental and ENT procedures. Type I HAE was diagnosed in 1980 and identified in several first-degree

relatives as well. Long-term prophylaxis with antifibrinolytic agents was ineffective, and treatment with danazol 100 mg/d was elected. Subsequently, the patient had been virtually symptom free for 20 years.

In March 1999, she presented with globus sensation, dyspnea with laryngeal edema, and substernal tightness. C1-INH concentrate (500 U) was administered to alleviate the presumed HAE attack. Her symptoms resolved except for persistent chest tightness, which lasted for 2 more days. Diagnostic imaging (chest x-ray, double air-contrast esophagography, and computed tomography of the chest) was performed and depicted a mass in the posterior mediastinum (Fig 1).

Fig. 1. Computed tomography scans of the esophageal leiomyoma (*arrow*).



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Bronchoscopy revealed external compression of the left main bronchus. Upper gastrointestinal endoscopy findings were negative.

Right-sided thoracotomy was performed, and a submucosal lesion was removed from the midportion of the esophagus—a leiomyoma by histology.

Preoperative preparation included short-term prophylaxis with C1-INH concentrate. The intraoperative course and postoperative recovery were uneventful; the patient has been permanently asymptomatic since then.

DISCUSSION

Esophageal leiomyoma can be manifested by dysphagia, substernal chest pain, vomiting, regurgitation, epigastric pain, and weight loss. Occasionally, it can mimic achalasia or occur with cardiac dysrhythmia. Nevertheless, it is asymptomatic in a large proportion of patients and can be recognized only accidentally. In this case it was diagnosed during an acute edematous attack of HAE. It could be hypothesized that HAE-associated submucosal edema resulted in swelling of the lesion. Alternatively, enlargement of the tumor to a critical size caused compression of surrounding tissues and led to complement activation, which resulted in edema formation. Irrespective of the mechanism, however, the persistence of

residual symptoms should always prompt further investigation to rule out an organic cause.

In this case short-term prophylaxis with C1-INH concentrate was justified because (1) the patient had a history of recurrent life-threatening laryngeal edema, (2) prolonged intubation of the airway increases the risk, and (3) nasogastric intubation precluded administration of oral danazol during the postoperative period. The success of this strategy confirms the acknowledged efficacy¹ of short-term prophylaxis with C1-INH concentrate.

Leiomyomatosis can occur in association with familial disorders—autosomal dominant conditions in particular—in as many as 50% of patients.⁴ This suggests that esophageal leiomyomatosis may be related to a genetic abnormality. Although an association between HAE and leiomyomatosis has not yet been reported, this case might suggest a potential relationship worthy of further research.

Neoplasia—benign or malignant—has been reported in several cases during long-term danazol prophylaxis.⁵ Because hormonal effects are thought to have a role in the cause of leiomyomatosis, continuous treatment with danazol for 20 years might be considered a pathogenetic factor.

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