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Extensive preoperative workup in diffuse esophageal leiomyomatosis associated with Alport syndrome influences surgical treatment: A case report

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ABSTRACT

INTRODUCTION: Diffuse esophageal leiomyomatosis is a rare disease. Misdiagnosis is frequent and previous surgeries can complicate surgical management. The only treatment described for severe symptomatic cases is esophagectomy.
PRESENTATION OF CASE: We describe a case of diffuse esophageal leiomyomatosis associated with Alport syndrome in a 21 year-old female where endoscopic ultrasonography (EUS) with concomitant fluoroscopy and 3D-gastric computed tomography (3D-GCT) modified surgical management.
DISCUSSION: The diagnosis of diffuse esophageal leiomyomatosis is difficult but can be greatly facilitated by extensive endoscopic and radiologic workup. Esophagectomy should only be entertained after complete anatomic mapping of the lesions, especially after previous surgeries.
CONCLUSION: EUS and 3D-GCT should strongly be considered as part of routine preoperative workup in these patients.

1. Introduction

Diffuse leiomyomatosis of the esophagus is a rare disease. It is associated with Alport syndrome in two third of cases and involvement of the bronchus, vulva and rectum may occur [1]. It is usually linked to deletion of the COL4A5/COL4A6 genes. When severe dysphagia impairs quality of life, esophagectomy is the only available treatment [2]. The level of proximal transection is not always well defined in this disease diffusely involving smooth muscle of the esophagus. The authors report a clinical case where additional preoperative imaging ultimately changed the surgical decision.

2. Clinical report

A 21 year-old female patient was referred to our institution with clinical complaint of severe progressive dysphagia to both solids and liquids associated with disabling epigastric pain. Her family history was unremarkable.

Her past medical history was significant for symptoms of dysphagia beginning at the age of 7. She then underwent diagnostic imaging with barium esophagogram that showed a dilated esophagus and narrowing of the gastroesophageal junction (GEJ). A diagnosis of achalasia was suspected and medical treatment was initiated for 3 months but showed only modest improvement. A surgery was elected and the patient underwent a laparotomy that showed a massively enlarged esophagus and thickened GEJ. The operative decision was a Thal's procedure which consists in a longitudinal full thickness opening of the lower esophagus and proximal stomach through the mucosae with an anterior fundic serosal patch. The montage was then fixed to the diaphragmatic hiatus with non-absorbable sutures.

The patient was able to resume diet even thought she had recurrent bouts of dysphagia in the following years. At the age of 18, she complained of progressive severe constipation with dilation of left colon. She underwent rectal biopsy which showed a paucity of sympathetic ganglionic cells with hypertrophy of muscularis propria and muscularis mucosae related to diffuse leiomyomatosis. During the following year, she was hospitalized numerous times with severe abdominal pain and fecal impaction. She underwent laparoscopic diverting ileostomy with colonic lavage at the age of 19. She also had resection of a clitoral nodule which proved to be a benign leiomyoma.

With progression of painful dysphagia and weight loss, radiological examination was performed and showed a lower esophageal nodular thickening reaching 8 cm at the GEJ with dilation of the esophagus as well as circular thickening of the entire esophageal length. Pelvic MRI showed a hypertrophic rectal muscularis and an

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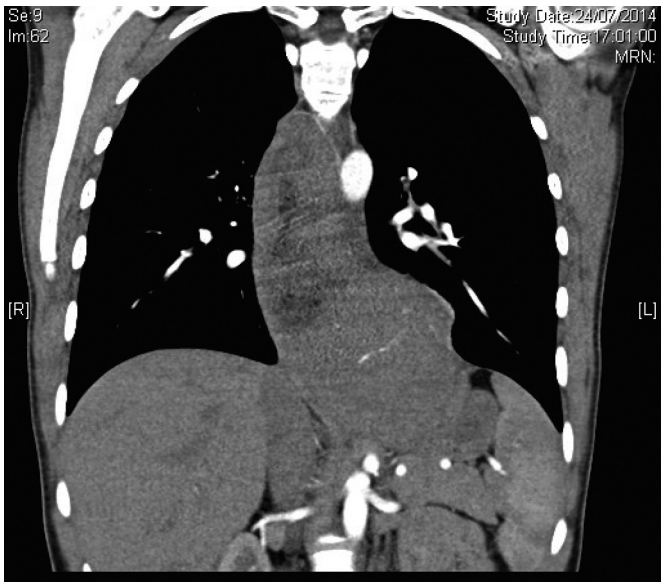

Fig. 1. Preoperative coronal CT-scan showing enlargement of mid and lower esophageal body.

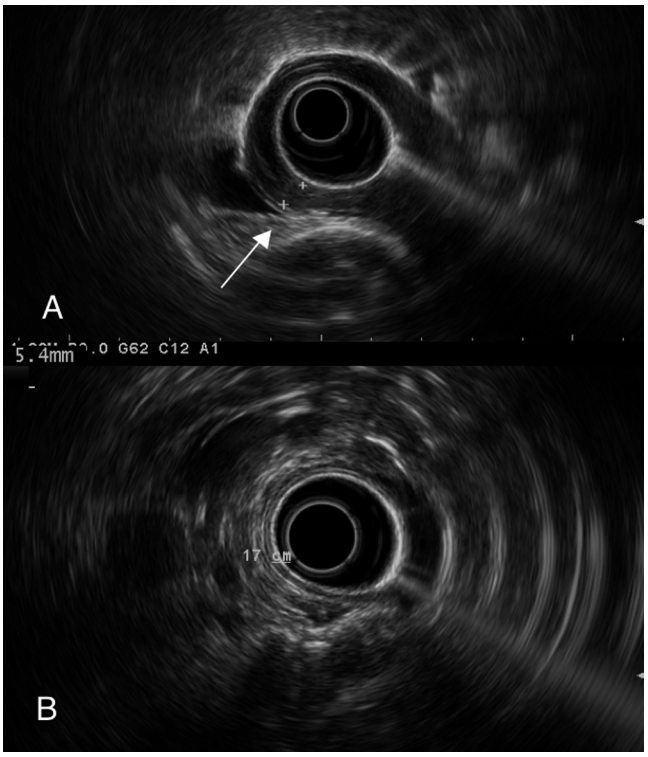

Fig. 2. (A) Endoscopic ultrasound showed a 5.4 mm thickening of the internal circular muscular layer in the lower part of esophagus, at 35 cm from teeth (arrow). (B) Endoscopic ultrasound showed the level of transition with normal muscular layer at 17 cm from teeth (neck level).

internal anal sphincter of 16 mm of thickness with a dilated proximal colon. Genetic testing confirmed deletion of COL4A5/COL4A6 gene. The diagnosis of diffuse leiomyomatosis associated with an Alport syndrome was made with extensive involvement of the esophagus, rectum and clitoris Fig. 1.

Because of severe painful dysphagia, total esophagectomy was elected with formation of a gastric conduit.. Due to the previous surgery on the stomach, possible difficulties with gastric conduit length were evoked. Additional diagnostic procedures with endoscopic ultrasound (EUS) combined with fluoroscopy as well as 3D gastric computed tomography with air (3D-GCT) using multidetector CT scanner after absorption of effervescent salt diluted in 10 ml of water and IV injection of butylscopolamine were realized. The EUS indicated that the level of transition between normal and thickened muscular layers was at the level of the neck, just below Killian's triangle Fig. 2. The 3D-GCT allowed to measure the volume and possible dilation of the residual stomach and showed that a gastric conduit could probably reach the neck Fig. 3.

A three way approach (McKeown operation) for total esophagectomy was preferred with the realization of a neck anastomosis so that all pathologic tissue would be included in the resection. Initially, a high thoracic anastomosis (Ivor–Lewis–Santy operation) was intended. Pathological specimen was 30 cm in length and showed circumferential thickening of both muscular layers to a maximum of 1.0 cm of thickness with prominence of the internal circular muscle. There was also a benign leiomyoma of 6.0 × 8.0 cm of the distal esophagus. Microscopy revealed homogenous cellularity without atypia of the smooth muscular cells or figure of mitosis. Proximal margin had a muscular layer of normal thickness even though it had a slightly thickened muscularis mucosae. The postoperative period was free of major surgical complications. The patient was discharge home 3 weeks after the

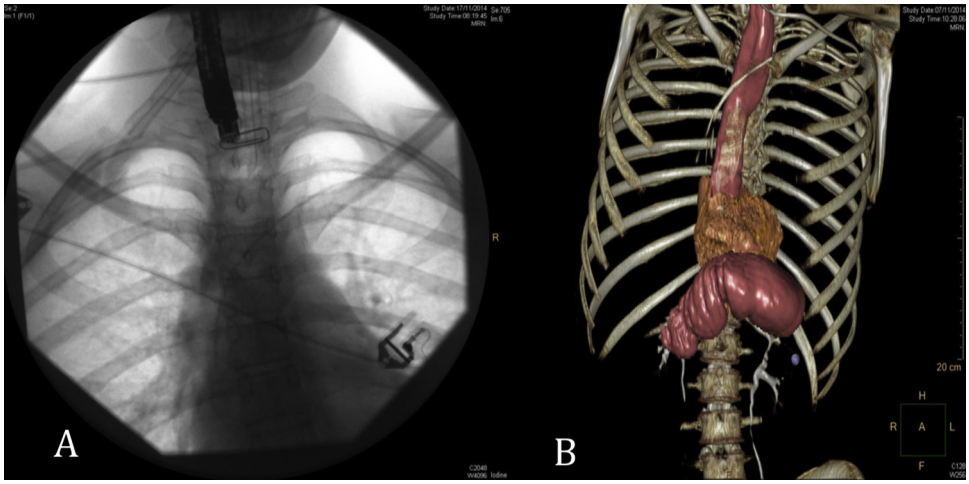

Fig. 3. (A) External placement of metallic clip combined with EUS shows transition point between normal and enlarged circular muscle of esophagus to be in the neck (B) 3D-GCT delineates gastric remnant morphology after previous surgery and shows voluminous leiomyoma of lower esophagus.