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

F. Dagbert[a,*], E. Pelascini[a], A. Pasquer[a], R. Gincul[b], F. Mion[b], G. Poncet[a], M. Robert[a]

[a] Service de Chirurgie Digestive, Hôpital Edouard-Herriot, Lyon, France
[b] Service de Gastroentérologie, Hôpital Edouard-Herriot, Lyon, France

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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

* *Corresponding author at: Hôpital Édouard-Herriot, 5 Place d'Arsonval, 69,003 Lyon, France. Tel.: +33 15 148175848.
E-mail address: francois.dagbert@umontreal.ca (F. Dagbert).

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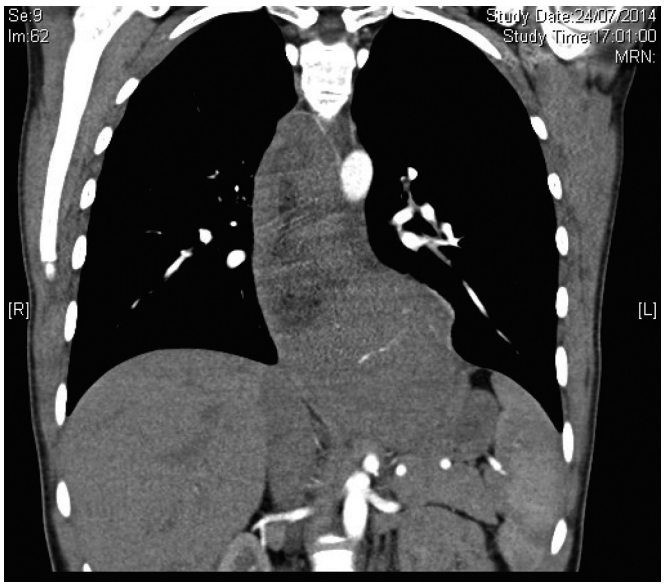

Fig. 1. Preoperative coronal CT-scan showing enlargement of mid and lower esophageal body.

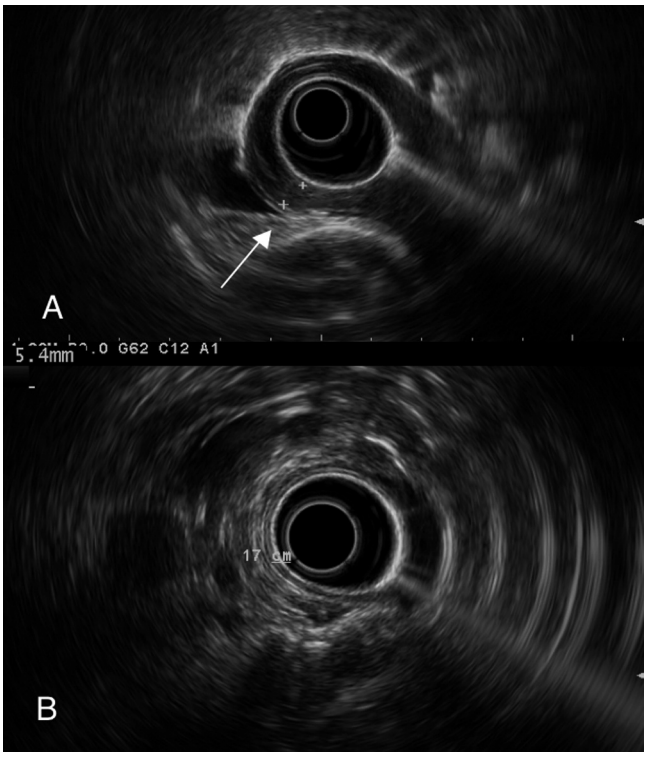

Fig. 2. (A) Endoscopic ultrasound showed a 5.4 mm thickening of the internal circular muscular layer in the lower part of esophagus, at 35 cm from teeth (arrow). (B) Endoscopic ultrasound showed the level of transition with normal muscular layer at 17 cm from teeth (neck level).

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

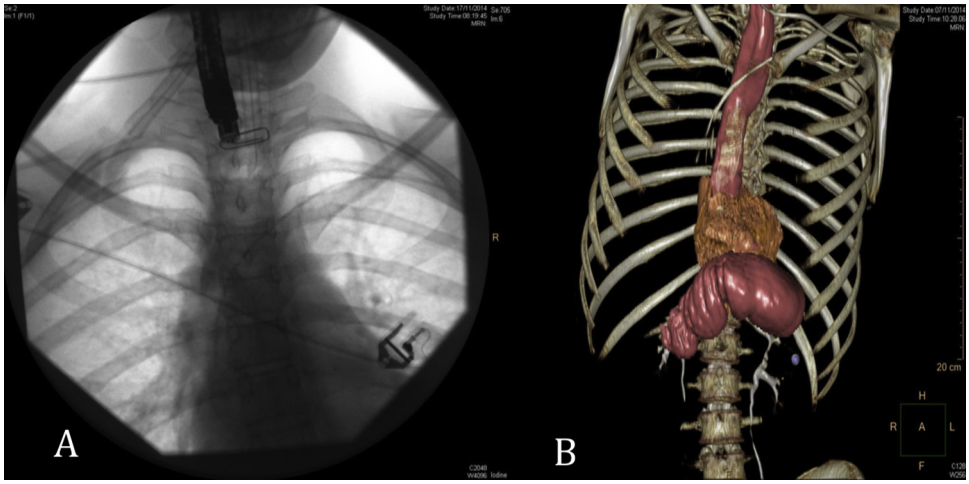

Fig. 3. (A) External placement of metallic clip combined with EUS shows transition point between normal and enlarged circular muscle of esophagus to be in the neck (B) 3D-GCT delineates gastric remnant morphology after previous surgery and shows voluminous leiomyoma of lower esophagus.

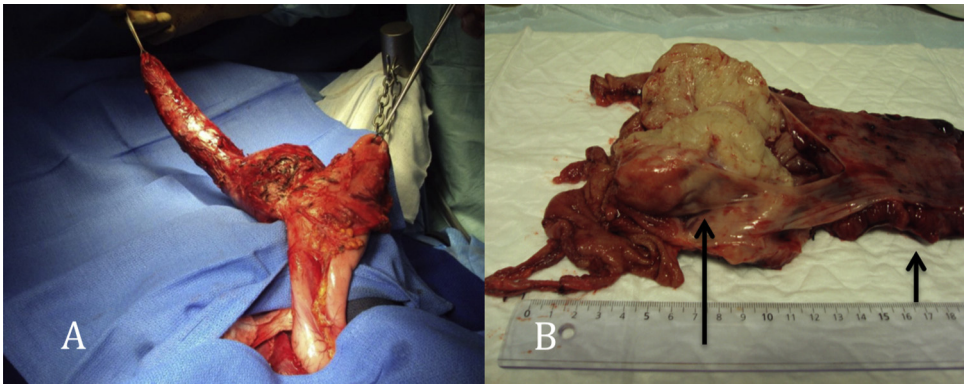

Fig. 4. (A) Operative specimen showing completely thickened esophagus (B) Voluminous leiomyoma of the lower esophagus (long arrow) and enlarged circular muscle fibers of the mid and upper esophagus (short arrow).

operation with persistent epigastric and abdominal pain related to food intake but no dysphagia. Because of the intensity of the abdominal pain, she had to be discharged with home parenteral nutrition Fig. 4.

3. Discussion

Diffuse esophageal leiomyomatosis associated with X-linked Alport syndrome has been well described and may imply a contiguous gene deletion involving the 5′exons of COL4A5 and COL4A6 genes [2–4]. Diffuse leiomyomatosis is characterized by circumferential thickening of smooth muscle layers of the esophagus and should be distinguish from leiomyoma which are well circumscribed benign tumor, usually single and rarely multiple [5]. The combination of both pathologies in the same patient as in our patient is unusual but has been described in few cases [6].

The presenting symptoms of dysphagia and odynophagia in young patients often leads to investigations and to the erroneous diagnosis of achalasia and megaesophagus in more than 50% of cases [7]. EUS seems to be the single best diagnostic study to confirm leiomyomatosis and could potentially avoid unnecessary and potentially harmful surgery [5]. Even high-resolution manometry can be interpreted as classic type II achalasia [8]. Treatment with myotomy or dilation invariably leads to recurrence of symptoms since the cause is not treated. Unfortunately, such previous surgery can complicate furthermore surgical treatment in these patients, especially when part of the gastric fundus is used in the covering of the myotomy and when the esophagus is fixed to the diaphragmatic hiatus.

In cases where previous surgery might interfere with normal anatomy, additional workup is needed to determine the best operative approach for reconstruction after esophagectomy. Blanchet et al. have demonstrated the usefulness of 3D-GCT with air dilation of the stomach in defining anatomy for reoperative bariatric procedures [9]. In our case, this test was helpful in determining the stomach volume and the possibility of using a gastric conduit for reconstruction. Esophagocoloplasty was ruled out in our patient because of the previous diverting ileostomy as the excluded colon would be too fragile for such a surgery.

Esophageal leiomyomatosis more commonly affects the middle and distal third of the esophagus but can involve the entire length in up to 35% of cases [2]. Preoperative standard imaging is rarely effective in delineating the extent of proximal involvement. EUS allows defining the precise limit between thickened muscularis propria and normal thickness esophagus [5]. With the addition of fluoroscopy during the procedure, external landmarks can easily be visualized and surgical planning between high thoracic or cervical anastomosis determined. These few centimeters can be of vital importance especially in the setting of a gastric conduit of limited length.

Surgical goal in these patients should be complete resection of the thickened smooth muscle to limit the risk of recurrence. Another important consideration is quality of life after esophagectomy as life expectancy is long in young patients with benign disease. Gastric reconstruction after esophagectomy confers good functional results even after long term follow-up and should be the preferred option in these young patients [10]. This technique is used more commonly now, even in pediatric surgery, and shows good long term results and less reoperation for conduit redundancy than esophagocoloplasty [11–13].

Long term follow-up is crucial in young patients with diffuse esophageal leiomyomatosis associated with Alport syndrome since the risk of recurrence and new extra-esophageal lesion is unknown. Familial screening is also necessary for siblings and children as familial involvement is well described [6]. Extensive preoperative workup including EUS and in some cases 3D-GCT is mandatory for good surgical management of these complex patients.

Conflicts of interest

All authors declare no conflict of interest.

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Ethical approval

This work has the approval of the ethical committee of the Hopital Edouard-Herriot where the patient was treated.

Consent

Oral and written consent was obtained from the patient for the manuscript and the pictures.

Author contribution

F. Dagbert : study concept, data collection and analysis, writing of the manuscript, review of the paper.

E. Pelascini : study design, data collection, critical review of the paper.

A. Pasquer : data collection and analysis, critical review of the paper.

R. Gincul : data collection and analysis, critical review of the paper.

F. Mion : data collection and analysis, critical review of the paper.

G. Poncet : study design, data collection and analysis, critical review of the paper.

M. Robert : study design, data collection and analysis, critical review of the paper.

Guarantor

Francois Dagbert is the guarantor of the study.

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