

Achalasia in a 16-year-old: diagnostic dilemma

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Introduction

- ❖ Achalasia is a disorder, particularly rare in children, with an incidence of 0.11/100000 in the pediatric population.
- ❖ It is a motility disorder of the esophagus characterized by inadequate esophageal peristalsis and failure of the esophageal sphincter to relax.
- ❖ Symptoms include but are not limited to dysphagia, vomiting, and weight loss.
- ❖ Barium esophagogram and manometry are the tools used for diagnosis. Treatment options among children include laparoscopic Heller myotomy and peroral endoscopic myotomy.

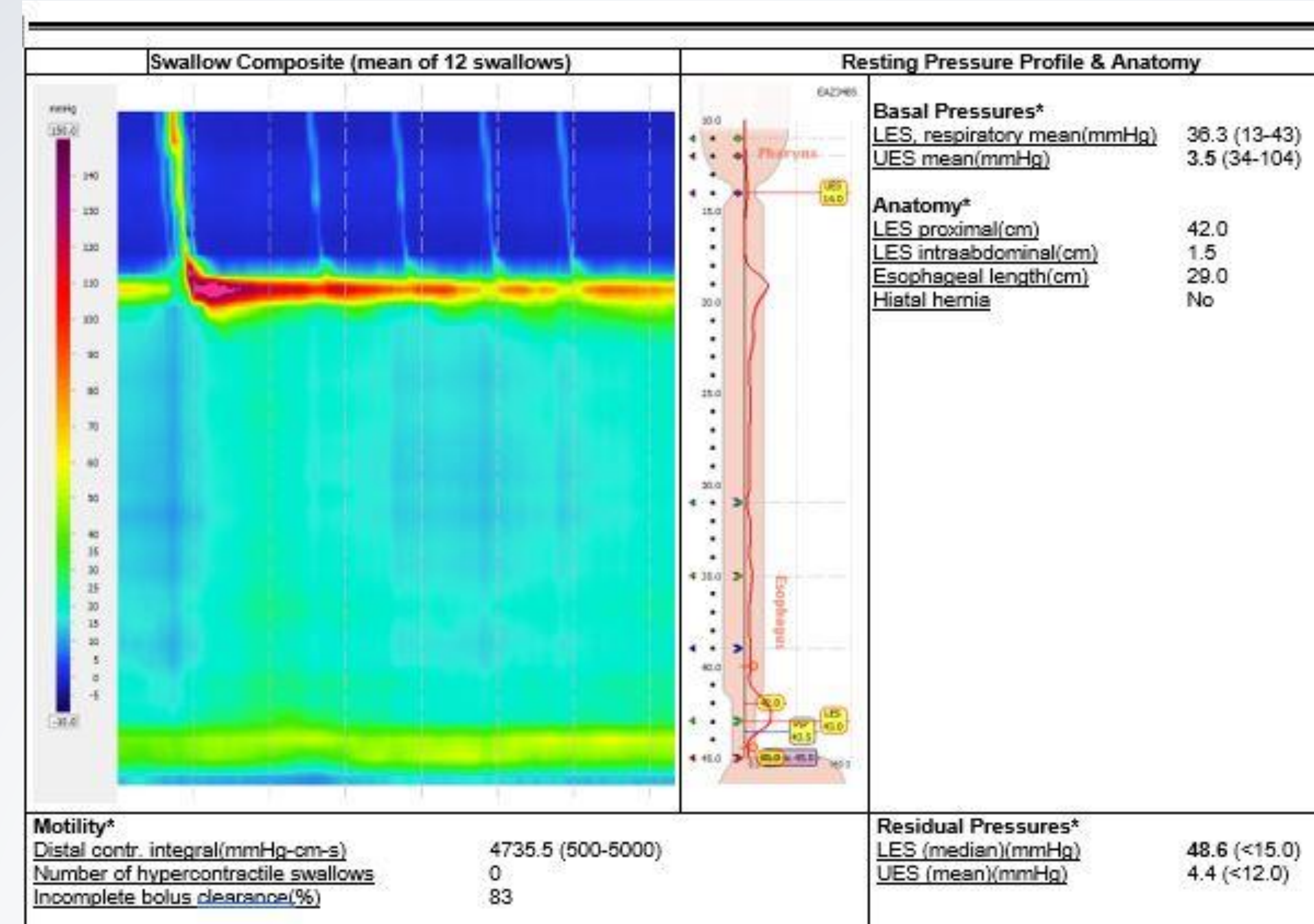


Figure 1: Manometry

Discussion

- ❖ Achalasia is a rare disorder that usually presents between the age of 25 and 40.
- ❖ Establishing an early diagnosis in children can be challenging since more than half of the cases are diagnosed at an older age creating a diagnostic dilemma for pediatricians and gastroenterologists.
- ❖ Nonspecific symptoms can lead to a misdiagnosis posing significant health risks to the patient if left untreated.
- ❖ Our patient had 50 pounds of weight loss and was started on TPN for nutrition even before the diagnosis.
- ❖ Given the rarity of the case, it helps increase awareness among pediatricians and gastroenterologists to consider motility disorders early on, with esophageal manometry considered the gold standard for diagnosis.

Case Presentation

- ❖ Our patient was a 16-year-old child who presented with persistent vomiting and weight loss for six to eight months. The patient underwent an EGD at an outside hospital which was inconclusive.
- ❖ Initially, there were concerns about an eating disorder, but it was ruled out. The patient had episodes of hypokalemia and syncope, requiring admissions to the hospitals on numerous occasions. The patient had to be started on TPN for nutrition.
- ❖ During our hospital admission, the patient underwent an EGD, which showed dilated esophagus suspected of a motility disorder. Esophagram showed findings consistent with an early stage of achalasia. Subsequently patient had endoscopy-guided placement of a manometry probe, and she was diagnosed with Achalasia type 1.
- ❖ The patient underwent POEM, which resulted in significant improvement of her symptoms. Her electrolyte abnormalities resolved as she started to tolerate a diet without emesis.



Figure 2: Esophagram



Figure 3: POEM