Case Challenge

A 14-Year-Old Female with Difficulty Swallowing

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A 14-year-old female presented to the emergency department (ED) for the evaluation of difficulty swallowing. She experienced no headache, visual changes, respiratory abnormalities, unusual ingestions, hematemesis, or bilious vomiting. The rest of her review of symptoms was negative except for dizziness upon standing.

Her medical history was significant for seeking medical care at another facility 7 months prior for gradually worsening epigastric pain that began at age 8 or 9 years. The pain was described as a “feeling like food was stuck.” She vomited food often after meals and felt the need to drink large amounts of water with eating in order to “push food down.” As part of the evaluation at the other facility, she underwent an upper gastrointestinal (UGI) series that demonstrated sharp tapering of the esophagus at the gastroesophageal (GE) junction and absence of esophageal peristalsis. Due to complete intolerance of oral intake at this ED visit, she was admitted to the pediatric service for further evaluation and management.

Upon admission, the girl was in no acute distress, was afebrile, and her heart rate was 120 beats per minute. Her weight of 54 kg and height of 1.727 m placed her body mass index at 18.10 kg/m². She appeared thin and anxious but otherwise had a normal physical examination. Unremarkable laboratory studies included a complete blood cell count, metabolic panel, and serum levels of magnesium, phosphorus, amylase, and lipase. Urine pregnancy test was negative. Review of her outside UGI series (Figure 1) confirmed sharp “bird’s beak” tapering of the esophagus at the GE junction, with mild esophageal dilatation and retained debris.

Evaluation by a pediatric gastroenterologist revealed a fluid-filled upper esophageal pouch, which was consistent with a diagnosis of Zenker diverticulum. The patient was taken for an esophagogastroduodenoscopy (EGD) and had a stricture dilated, which improved her symptoms. She was discharged home on a liquid diet and was seen in clinic for follow-up.

Figure 1. Barium swallow revealed a classic “bird’s beak” appearance of the esophagus at the gastroesophageal junction.

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Editor’s note: Each month, this department features a discussion of an unusual diagnosis. A description and images are presented, followed by the diagnosis and an explanation of how the diagnosis was determined. As always, your comments are welcome via email at pedann@Healio.com.
The patient underwent repeat EGD (Figure 2) that confirmed a mildly dilated and debris-filled esophagus, no significant mucosal abnormalities, and the ability to advance the gastroscope into the stomach without difficulty. The ability to pass the gastroscope without difficulty indicated the absence of a fixed or fibrotic stricture or mass. Subsequent esophageal manometry (Table 1) demonstrated a hypertensive lower esophageal sphincter (LES), poor LES relaxation in response to swallows, and absent peristalsis, confirming a diagnosis of achalasia. A nasogastric feeding tube was placed and the patient was discharged for home enteral tube feedings. She was readmitted the following week and underwent an uncomplicated laparoscopic Heller myotomy with Dor fundoplication. After a contrast esophagram on the first postoperative day showed no extravasation, she quickly advanced to a soft diet without vomiting and was discharged on the third postoperative day on full oral feeds. At her 3-week postoperative visit, she continued to do well and had gained nearly 3 kg of weight.

**DISCUSSION**

Esophageal achalasia is a rare condition in children and adolescents, with an estimated incidence of about 0.11 per 100,000.1 Approximately 6% of cases are diagnosed during infancy.2 Achalasia in the pediatric population typically occurs in isolation, and its etiology is idiopathic.3 Described as a neurodegenerative disorder of the esophagus, idiopathic achalasia is associated with the loss of neurons in the myenteric plexus. The LES is unable to relax, which restricts transportation of contents from the esophagus to the stomach and leads to the primary complaint of dysphagia. Rare instances of familial achalasia with a genetic etiology of autosomal recessive inheritance have been described, along with syndromic achalasia. Allgrove’s syndrome, also known as triple-A syndrome, is an autosomal recessive disorder that is characterized by achalasia, alacrima, and adrenal insufficiency.4 Approximately 2% of patients with Trisomy 21 have achalasia.3

A diagnosis of achalasia should be suspected with the presence of one or more of the following clinical symptoms: progressive dysphagia of both solids and liquids, vomiting of undigested food, failure to thrive with weight loss, and recurrent choking. Whereas inflammatory or stricturing lesions of the esophagus typically present with dysphagia primarily for solid foods, the presence of dysphagia for liquids should alert the clinician to the possibility of a motility disorder such as achalasia. Other symptoms may include chest pain, heartburn, and respiratory symptoms such as nocturnal cough and a history of recurrent pneumonia.5

The evaluation of the patient begins with an extensive history taking, includ-
ing obtaining an adequate family history to help identify a potential genetic component as a causative factor of achalasia. The physical examination is usually unremarkable but may indicate failure to thrive and signs of dehydration. Diagnostic tests include barium swallow, EGD, and the current gold standard, esophageal manometry. Characteristic signs of achalasia demonstrated by esophageal manometry include absence of peristalsis in the lower two-thirds of the esophagus, elevated LES pressure (>30 mm Hg) with increased resting tone, and absent or incomplete relaxation of the LES.

The adult criteria established for diagnosing achalasia through manometry may also be applied to children. Manometry alone can determine the diagnosis in up to 90% of cases of achalasia. Furthermore, the severity of the disease can be determined by the pressure curve shown on manometry, which can further assist in developing an appropriate treatment plan for the patient. Typical findings of achalasia on a barium swallow are dilation of the body of the esophagus with tapering at the GE junction in a classic “bird’s beak” appearance (Figure 1), and the extent of these findings can depend on the severity of the disease. Obtaining an EGD is necessary in the evaluation of achalasia to differentiate primary versus secondary achalasia due to another esophageal disorder as a potential cause of the disease. Although rare in childhood, secondary causes, such as esophagitis, stricture and tumors, may present with similar manometric findings as achalasia and thus need to be ruled out by endoscopy. EGD findings with advanced achalasia can include dilation of the esophagus with or without food particles present.

As in this case, because achalasia is due to failure of relaxation of the LES rather than a fixed mechanical obstruction, the endoscopist usually encounters no difficulty in advancing the gastroscope into the stomach.

The primary goals of treatment of achalasia are to reduce the LES pressure and relieve the functional obstruction. Patients may be treated nonsurgically with pneumatic dilatation or surgically with esophagomyotomy. Pneumatic dilatation of the esophagogastric junction was the nonsurgical treatment of choice in the 1970s for both adult and pediatric achalasia and is still performed today. The procedure is performed by inflating a balloon in the area of constriction, which weakens the LES by tearing its muscle fibers. The most common complication is esophageal perforation. The procedure may have a short effective period and may require re-treatment. Furthermore, very few studies have reported the effectiveness of pneumatic dilatation in children. In a study performed by Hussain et al., 2 of 33 children with achalasia underwent pneumatic dilatations, with both patients requiring re-treatments within the same year. Therefore, this procedure is not currently the treatment modality of choice.

Heller myotomy had been the surgical approach to treatment of achalasia since the 1920s. However, with recent technological advances, a minimally invasive laparoscopic myotomy is now the standard approach, along with simultaneous esophagoscopy to assist with visualization of the esophagus by providing light and insufflations. Possible complications of the surgery include esophageal perforation and acid reflux. In the study conducted by Hussain et al., 17 of the 33 pediatric cases of achalasia underwent Heller myotomy for treatment. Initially, the patients were deemed as asymptomatic; however, the majority experienced postoperative GER reflux. Thus, laparoscopic Heller esophagomyotomy with fundoplication is the current surgical treatment of choice for pediatric achalasia.

CONCLUSION

Although achalasia is a rare medical condition in childhood, dysphagia is a common complaint. In order to adequately evaluate the child who complains of dysphagia, a complete differential diagnosis must be considered, so that an achalasia diagnosis would not be overlooked.

REFERENCES