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CASE REPORT

Congenital esophageal stenosis: the differential diagnosis and management

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Abstract Congenital esophageal stenosis (CES) is a rare congenital abnormality that is difficult to diagnose and often masquerades as other types of structural esophageal disease. We report three cases of CES with different presenting symptoms. We advocate for balloon dilation as the preferred first approach to therapeutic intervention. CES is an important clinical entity in the evaluation of pediatric esophageal disorders and should be suspected in young infants with dysphagia.

Keywords Congenital esophageal stenosis · Achalasia · Tracheo-esophageal fistula

Introduction

Congenital esophageal stenosis (CES) is a rare condition thought to occur in 1 per 25,000–50,000 live births [1]. CES describes a discrete segmental stenosis of the esophagus and can be classified based on the histologic type of the stenosis: (1) ectopic tracheobronchial remnants (TBR); (2) fibromuscular thickening (FM); and (3) membranous diaphragm (MD) [2]. Most cases of CES are diagnosed within the first year of life when solid foods are introduced and regurgitation of food becomes a prominent clinical feature. Dilation of these stenoses is becoming recognized as a safe and effective initial treatment. We present three cases of congenital esophageal stenosis, each presenting differently and undiagnosed until the time of endoscopy.

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Case 1

A 5-month-old girl born at 37 weeks gestation presented for management of failure to thrive. Her birthweight was 2.9 kg (8th percentile). By one month of age, she was noted to have poor weight gain and was hospitalized twice for workup, but no diagnosis had been made. She was subsequently placed on 90 ml of 26 kcal/oz formula every 3–4 h without improvement. At 5 months of age, her weight was 3.7 kg (<1st percentile). Of note, she never had any significant emesis or other abdominal symptoms. Physical examination was otherwise notable for generalized hypotonia. An esophagogram showed a markedly dilated esophagus with severe narrowing of the gastroesophageal junction (Fig. 1a). Flexible esophagoscopy demonstrated a smooth, concentric, high-grade narrowing at the gastroesophageal junction. The stenosis was serially dilated under fluoroscopic guidance with balloon and Hurst-Maloney dilators up to a 34F without difficulty (Fig. 1b). Post-operatively, she was able to tolerate 180 ml formula every 3–4 h. Three months later, a repeat esophagogram showed mild recurrent distal esophageal narrowing. The esophagus was then dilated up to a 48F without difficulty. At 10 months of age, repeat esophagogram continues to show only mild distal esophageal stenosis without gastroesophageal reflux. She remains asymptomatic with steady weight gain.

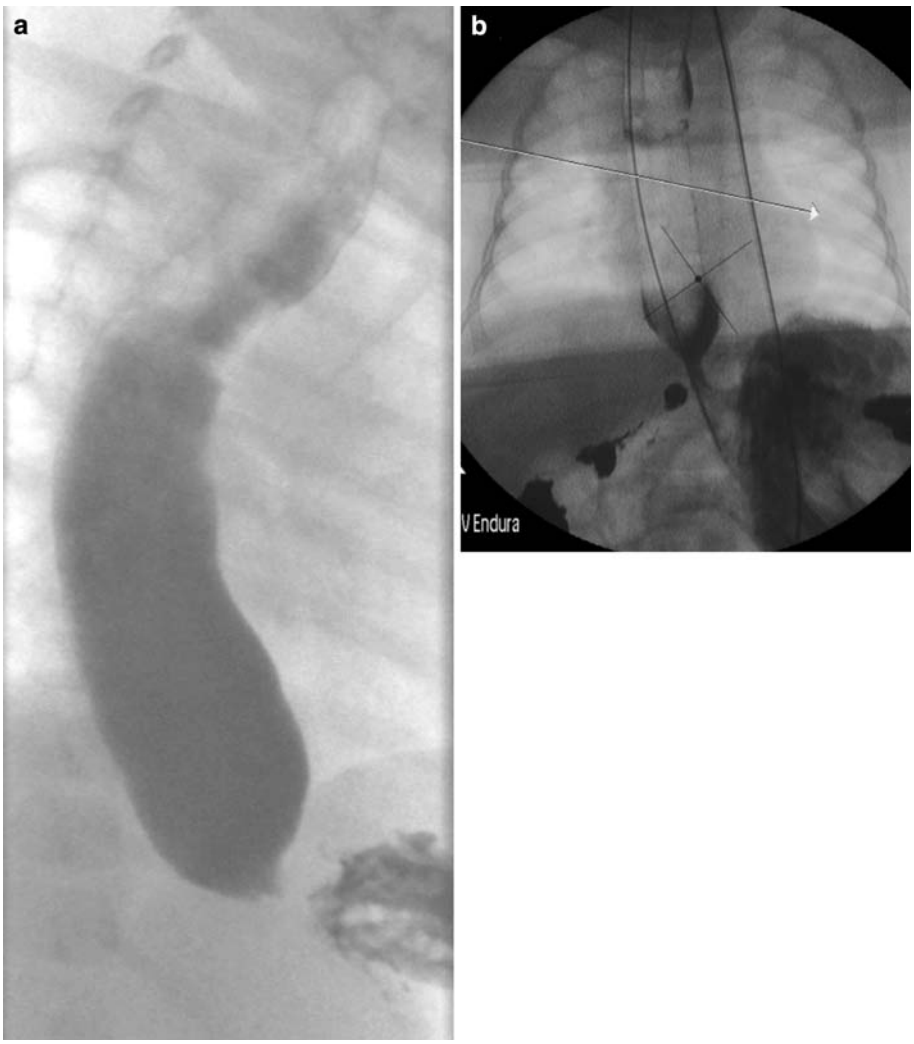

Fig. 1 **a** Case 1: initial esophagogram showing a markedly dilated esophagus with severe narrowing at the gastroesophageal junction. **b** Case 1: fluoroscopic guided balloon dilation of CES

Case 2

A 5½-year-old born at 37 weeks gestational age began to have episodes of vomiting and choking at 9 months of age. At 18 months of age, she was evaluated by a gastroenterologist who diagnosed gastroesophageal reflux. Over the next 4 years, she continued to vomit 4–6 times per day. By 5½ years of age, the patient was at <5th percentile for height and weight. An esophagogram showed marked narrowing of the distal esophagus at the gastroesophageal junction, a bird's beak configuration of visualized contrast material, and dilation of the proximal esophagus (Fig. 2a). This was thought to be consistent with achalasia. Esophageal manometry was obtained which did not yield results typical of achalasia. CT of the chest revealed diffuse circumferential mural wall thickening suggestive of an infiltrative process, possibly suggestive of diffuse esophageal leiomyomatosis (Fig. 2b). An MRI of the chest showed similar findings and also failed to visualize any discrete stenosis. Flexible esophagoscopy was performed to better delineate the patient's esophageal anatomy, revealing a discrete circumferential band-like thickening of the lower esophagus. Sequential dilation was performed using mercury-weighted bougies beginning with 26F and proceeding to 34F. A post-operative esophagogram was obtained which revealed contrast extravasation posterior and inferior to the stenosis. The patient was made NPO, treated with broad-spectrum intravenous antibiotics and monitored in the pediatric intensive care unit. Over the next 10 days, the patient showed no signs of infection. An esophagogram was repeated showing resolution of the contrast leak and free passage of contrast into the stomach. She was discharged on a soft diet without any episodes of emesis post-dilation. She had no residual symptoms at 4 months post-operatively. At that time, she underwent fluoroscopically guided hydrostatic balloon dilation of her residual stenosis. Approximately 1 year following her initial dilation, she is tolerating a regular diet of solid foods.

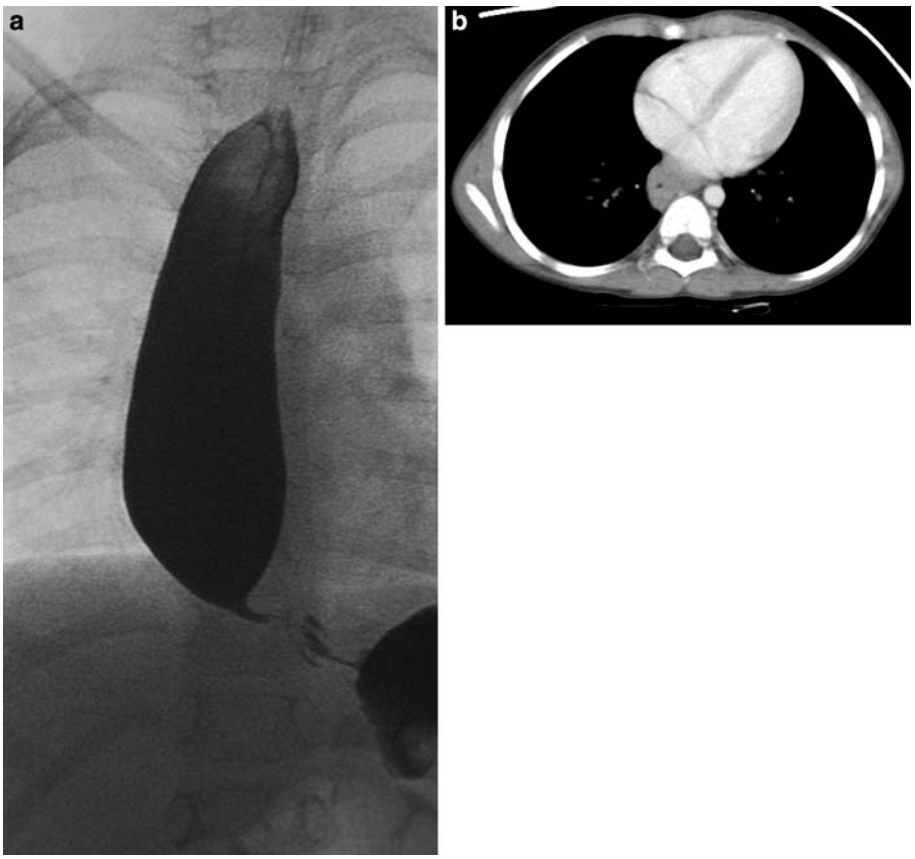

Fig. 2 **a** Case 2: initial esophagogram showing marked narrowing of the distal esophagus, bird's beak configuration of contrast material and dilation of the proximal esophagus. **b** Case 2: image from CT of the chest showing diffuse, circumferential esophageal wall thickening. Esophageal wall thickness measures 6 mm

Case 3

A 9-month-old girl born at 31 weeks gestation with multiple VACTERL anomalies presented for dysphagia following repair of a type C esophageal atresia (EA) and tracheoesophageal fistula (TEF) at an outside hospital. At presentation, she could not tolerate solid foods. An esophagogram showed moderate distal esophageal narrowing with minimal gastroesophageal reflux and no anastomotic stricture (Fig. 3). Flexible esophagoscopy showed a widely patent anastomosis and a smooth, concentric stricture approximately 1 cm above the gastroesophageal junction. The stenosis was serially dilated under fluoroscopic guidance with Hurst-Maloney dilators up to a 28F without difficulty. Post-operatively, she developed a large left pneumothorax requiring placement of a 10F chest tube. Constrast swallow studies performed on the third post-operative day showed a large posterolateral distal esophageal leak into the left pleural space. She was made NPO, but subsequently developed severe mediastinitis requiring broad-spectrum antibiotics and thorascopic decortication. Cultures grew *Streptococcus*, *Enterobacter*, and *Candida*. Her clinical state ultimately improved. Two weeks later, a repeat swallow study demonstrated no evidence of a distal esophageal stenosis or leak. At 13 months of age, she remains asymptomatic with improving oropharyngeal dysphagia.

Discussion

The three cases described above illustrate the variety of ways in which congenital esophageal stenosis can present.

In the first case, the patient had no evidence of food intolerance or vomiting but did have profound failure to thrive. Esophagogram suggested the diagnosis of CES and esophagoscopy confirmed it. Because this lesion was so responsive to dilation, it is likely of the FM type. This case illustrates that CES may be present even in the absence of clinically evident dysphagia and/or vomiting and should be considered in infants with unexplained failure to thrive.

In the second case, the patient was older than most children diagnosed with CES. It is commonly diagnosed when solid foods are introduced into a child's diet and the stenosis causes dysphagia, but she adapted to her condition by taking a mostly liquid diet. Furthermore, all diagnostic workup had indicated the diagnosis of achalasia. She underwent upper endoscopy as a precaution prior to Heller myotomy due to equivocal manometry results. Had the endoscopy not been performed, this

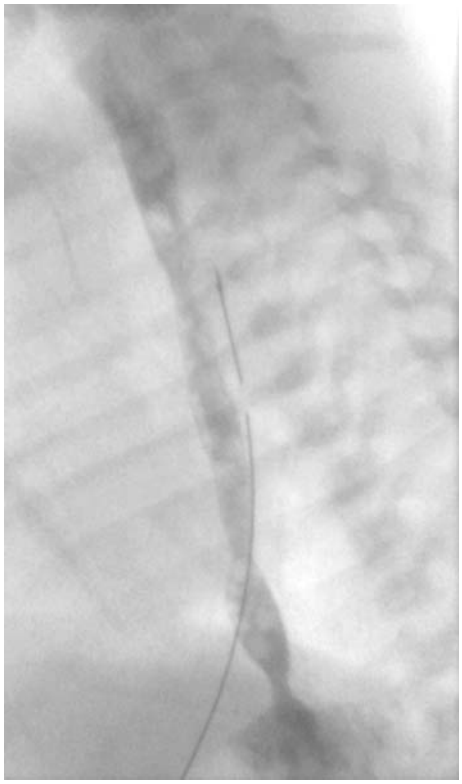

Fig. 3 Case 3: initial esophagogram showing moderate distal esophageal narrowing with minimal gastroesophageal reflux and no anastomotic stricture. This patient had recently undergone repair of EA/TEF

patient would have been subjected to an unnecessary surgical procedure and would certainly have experienced no remittance of symptoms. On the original esophagogram, we believe that the stenosis was slightly proximal to the lower esophageal sphincter and thus gave the appearance of achalasia. Furthermore, the diffuse thickening of the esophagus seen on CT did not represent an infiltrative process, but rather a predictable structural adaptation to the stenosis.

Diffuse esophageal leiomyomatosis was a possible diagnosis for the patient in case 2 based on CT results prior to endoscopy. Diffuse esophageal leiomyomatosis is a non-neoplastic process of diffuse smooth muscle thickening in the esophagus described in approximately 60 cases [3, 4]. Diffuse esophageal leimyomatosis is characterized by circumferential smooth muscle thickening that may reach 4 cm in thickness and involving the entire esophagus in 35% of cases [3]. As in our patient, dysphagia and vomiting are the most common presenting complaints and are attributable to encroachment of the thickened esophageal wall on the lumen [3, 4]. Dysphagia is usually progressive and severe and eventually leads to surgical treatment with esophagectomy when weight loss becomes prominent. Extension into the cardia and fundus of the stomach occurs in 80% of cases and may necessitate partial gastrectomy [3].

In the final case, the stenosis was diagnosed only after the repair of an esophageal atresia and tracheo-esophageal fistula. Its recognition as distinct esophageal pathology in the post-operative setting is a challenge. CES in the setting of EA is well-described [5] and should be considered whenever an infant has dysphagia following EA repair. Routine post-operative esophagogram after EA repair may show CES if the index of suspicion is high.

The primary diagnostic modality for CES is the esophagogram which may show an abrupt or tapered stenosis. Some investigators have argued that the abrupt stenosis on esophagogram correlates with the TBR type of CES and the tapered stenosis correlates with the FM type. However, these relationships are not consistent and, in fact, the results of an esophagogram may bear no relation to histologic type [6]. Due to its varied clinical presentations and poor specificity of imaging techniques, treatment is often undertaken before the type of stenosis can be determined.

In the past, segmental esophageal resection with primary anastomosis was the treatment of choice for the FM and TBR types of CES. The MD type has typically been managed by endoscopic dilation or excision [7, 8]. In recent years, however, dilation has proven to be an effective treatment for the FM type as well [9]. Dilation has become such a mainstay of treatment in the FM that in many series, if a membranous diaphragm is not visualized on endoscopy and if the CES is treated effectively by dilation, then it is presumed that the CES is the FM type [5]. The TBR CES is the most common type of CES and also the most resistant to dilation, eventually requiring surgical intervention in up to 70% of cases [6].

Observations about the responsiveness of each histologic type to dilation are made retrospectively, because only the MD type can be diagnosed at the time of endoscopy. Dilation for all types of CES has been accepted by some groups as an appropriate first-line treatment for this reason, and because it is effective, minimally invasive and safe [6]. In their series, Amae et al. [6] found that 8 of 11 children who underwent initial dilation developed a recurrence. In those patients who developed a single recurrence, they then proceeded to esophageal resection. In our own experience, two of our three patients required a repeat dilation; however, this second dilation proved successful. This suggests that taking an approach of repetitive dilations may well avoid surgical resection. Clearly, if a CES lesion proves to be resistant to a series of dilations, then it can be tentatively diagnosed as the TBR type and surgical resection can be planned with the knowledge that less invasive treatment options have been attempted first.

Perforation is a well-known complication of dilation [5]. It incurs extended hospitalization and may require a

thoracotomy for surgical correction. However, the risk of perforation should not preclude the use of endoscopic dilation as first-line treatment. Performing esophageal resection on patients with lesions potentially amenable to dilation would unnecessarily commit those patients to a shortened esophagus, gastroesophageal reflux and, in some, the need for an eventual antireflux procedure. Furthermore, though resection may eliminate the CES lesion, a proportion of patients will develop post-resection stricture and will experience similar symptomatology despite treatment [5].

We believe that all CES lesions should be initially treated with dilation. To minimize the risk of perforation, endoscopically guided or fluoroscopically guided balloon dilation may be performed instead of bougie dilation. These methods have the mechanical advantage of applying a circumferential rather than an axial shearing force on the stenosis.

Conclusion

Congenital esophageal stenosis is a difficult diagnosis to make, but vigilance in the workup of esophageal pathology may spare patients unnecessary interventions. When CES is diagnosed, patients should be treated with endoscopic or fluoroscopically-guided dilation of the lesion. Only if the lesion fails to respond to a series of dilations should surgical resection be considered.

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