Radiology Case Reports 14 (2019) 315–319

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

A rare case of laryngeal cleft in association with VACTERL and malrotation

*Jesse Chen, MD, Jonathan Stern, MD, Jeremy Neuman, MD, June Koshy, MD**

Staten Island University Hospital/Northwell, 475 Seaview Ave. Staten Island, NY 10305, USA

ARTICLE INFO

Article history:
Received 6 August 2018
Revised 1 November 2018
Accepted 3 November 2018
Available online 6 December 2018

Keywords:
Laryngeal cleft
Laryngotracheoesophageal cleft
VACTERL
Malrotation

ABSTRACT

We report a rare case of a neonatal girl who presented with coughing and dyspnea immediately after feeds. At birth, she was noted to have an imperforate anus with a posterior fourchette fistula from which she was stooling. Initial imaging with radiography showed a normal bowel gas pattern; however, lumbar vertebral anomalies were noted. An upper GI series was performed and revealed a laryngeal cleft and malrotation. Ultrasound confirmed malrotation with an abnormal SMA-SMV relationship. Since laryngeal cleft is a rare condition and may not be known to most radiologists, its incidence is likely underestimated. It is important to note the association of laryngeal clefts with VACTERL and malrotation. In addition, it is essential not to confuse a laryngeal cleft with a tracheoesophageal fistula since the management differs.

Introduction

Laryngeal cleft, also known as laryngotracheoesophageal cleft, is a rare entity that is associated with congenital anomalies and rare genetic syndromes. It is likely underdiagnosed due to the nondiagnosis of minor forms and the high mortality rate of more severe forms [1]. It is thus important for radiologists to be aware of this entity. Esophagram is useful in identifying laryngeal clefts; however, it is important not to confuse it with a tracheoesophageal fistula.

Case report

A 4-day term baby girl presented at birth with shortness of breath. On physical exam, she was noted to have an imperforate anus without significant abdominal distention. Initial chest radiography showed clear lungs with a prominent ductus bump. Abdominal radiograph revealed a normal bowel gas pattern; however, vertebral anomalies were detected within the lower lumbar spine at L4 and L5 (Figs. 1a and b). Spine ultrasound confirmed the presence of the lumbar vertebral anomalies without evidence of tethered cord (Figs. 1c and d). The patient began stooling through a posterior fourchette fistula within the first 24 hours of life. She was subsequently started on feeds and immediately was coughing with dyspnea. An upper gastrointestinal (GI) series was performed which showed a large connection between the larynx/upper trachea and the pharynx/upper esophagus from the level of C3 to the level of C5-C6 (Fig. 2). A nasogastric tube was inserted into the stomach under fluoroscopic guidance and the remaining foregut was interrogated. Contrast passed easily into the small bowel; however, examination of the duodenum revealed that the third and fourth parts of the duodenum did not cross the midline and ascend toward the normal position of the ligament of

* Corresponding author.
E-mail address: jkoshy@northwell.edu (J. Koshy).

https://doi.org/10.1016/j.radcr.2018.11.002

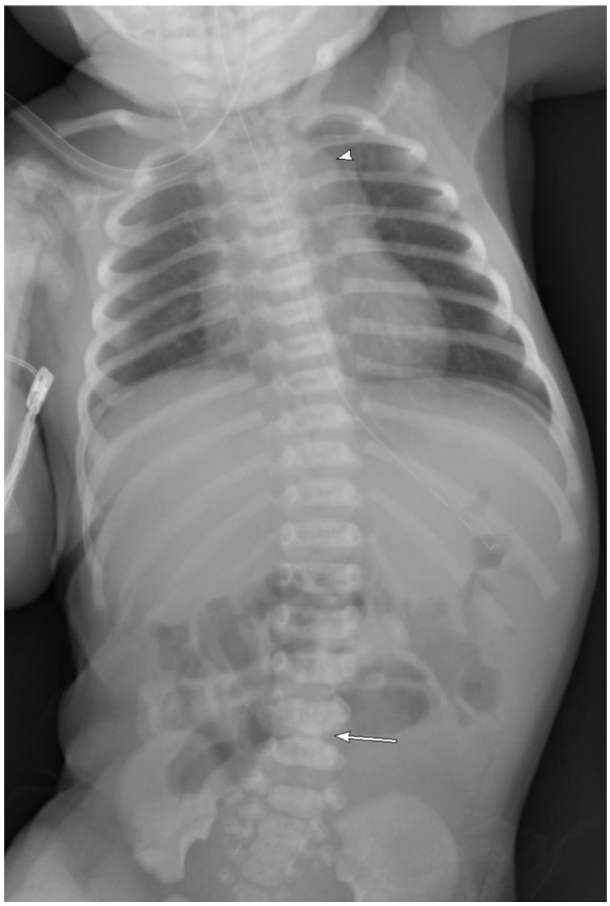

Fig. 1a – Anteroposterior radiograph of the chest and abdomen demonstrating a vertebral anomaly at L4-L5 (white arrow). A normal bowel gas pattern is seen. Incidentally noted is a ductus bump (white arrowhead).

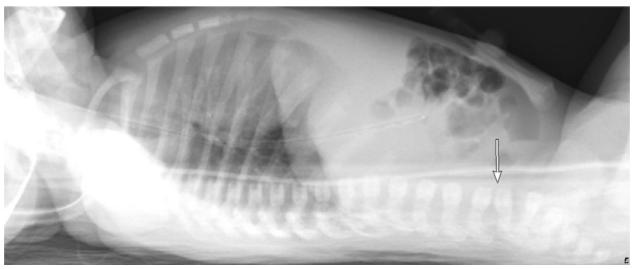

Fig. 1b – Lateral radiograph of the chest and abdomen demonstrating the vertebral anomaly at L4-L5 (white arrow).

Treitz. Instead, the third portion of the duodenum folded back on itself in the right abdomen, suggesting malrotation (Fig. 3). Ultrasound demonstrated an abnormal superior mesenteric artery-superior mesenteric vein (SMA-SMV) relationship with the SMA to the right of the SMV, also compatible with malrotation (Fig. 4). The baby was subsequently transferred to a children's hospital and laryngoscopy was performed which confirmed the presence of a laryngeal cleft (Fig. 5). Laryngoscopy, however, depicted more extensive involvement of the cleft

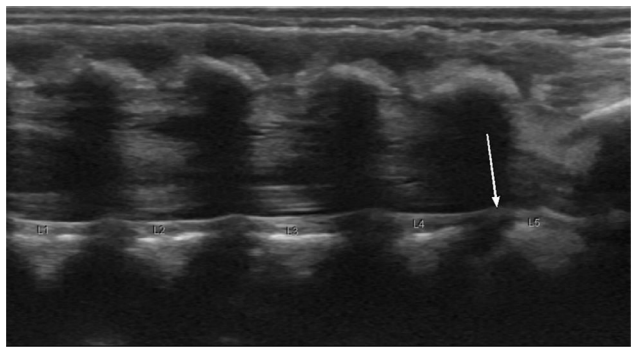

Fig. 1c – Sagittal ultrasound image of the lumbar spine demonstrating the vertebral anomaly at L4-L5 as narrowing of the disc space and tethering of the vertebral bodies (white arrow).

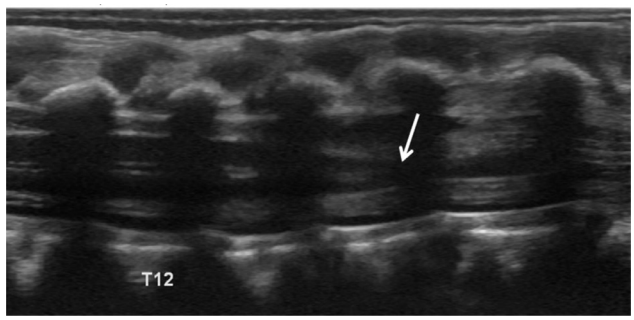

Fig. 1d – Sagittal ultrasound image of the lumbar spine demonstrating a normal shape and position of the conus medullaris at L2 (white arrow).

than originally thought on esophagram with extension near the carina (type IV cleft) therefore, the baby was again transferred to a more specialized children's hospital to undergo surgical intervention.

Discussion

Laryngeal clefts are a rare congenital anomaly with an estimated incidence of 1 in 10,000-20,000 live births. They represent approximately 0.2%-0.5% of malformations involving the larynx [2]. Since laryngeal clefts are rare, they may not be easily recognized by clinicians and radiologists. Familiarity with this entity is important because early treatment is imperative. Delay in management can result in complications related to aspiration and gastric reflux [3]. Laryngeal clefts should not be confused with tracheoesophageal fistulas because each entity has a different management algorithm and misdiagnosis may lead to delayed definitive treatment. Tracheoesophageal fistulas may also occasionally occur with laryngeal clefts [4]. Upon identification of a laryngeal cleft, the patient should be evaluated for additional associations since this will also affect the overall prognosis. Laryngeal clefts are associated with vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities (VACTERL), malrotation, and several rare genetic syndromes

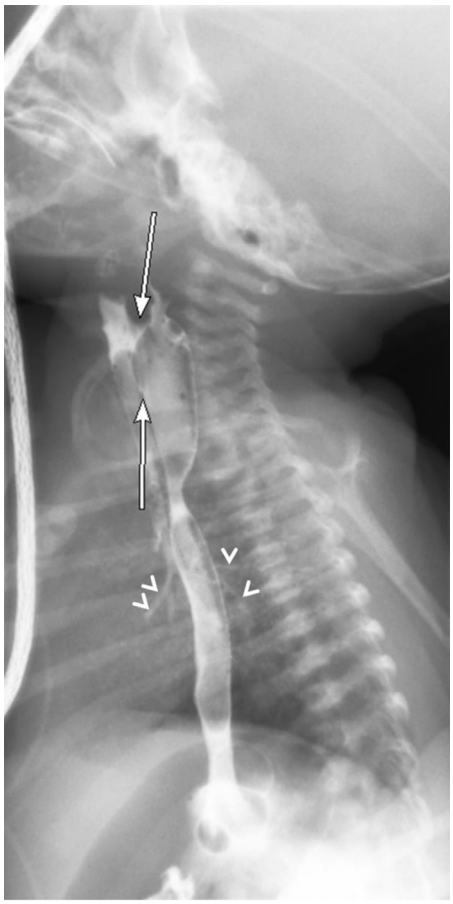

Fig. 2 – Lateral image from an esophagram demonstrating a large connection between the larynx/upper trachea and the pharynx/upper esophagus from the level of C3 to the level of C5-C6 (white arrows). Contrast is seen extending into the bronchial tree (white arrowheads).

(ie, CHARGE syndrome, Opitz GBBB syndrome, Pallister-Hall syndrome) and thus the radiologist will play an important role in evaluation of these patients. Isolated tracheoesophageal fistulas are less likely to be associated with congenital anomalies whereas the laryngeal clefts are often associated with other syndromes [2].

The main distinguishing feature between tracheoesophageal fistulas and laryngeal clefts is the location. Tracheoesophageal fistulas have an abnormal connection between the trachea and esophagus whereas in laryngeal clefts, the abnormal connection is between the larynx and esophagus. Additional involvement of the trachea can be seen in the most severe forms. The Benjamin and Inglis classification system for laryngeal clefts is the most widely accepted and classifies them into 4 types. The mildest form, Type I is supraglottic with extension no further than the level of the true vocal cords. Type II clefts extend below the vocal cords into the cricoid cartilage. Type III extends through the cricoid cartilage with or without extension into the trachea. Type IV clefts extend to the trachea and may extend to the level of the carina [1].

Radiography is useful as an initial imaging modality to evaluate for findings related to VACTERL such as vertebral

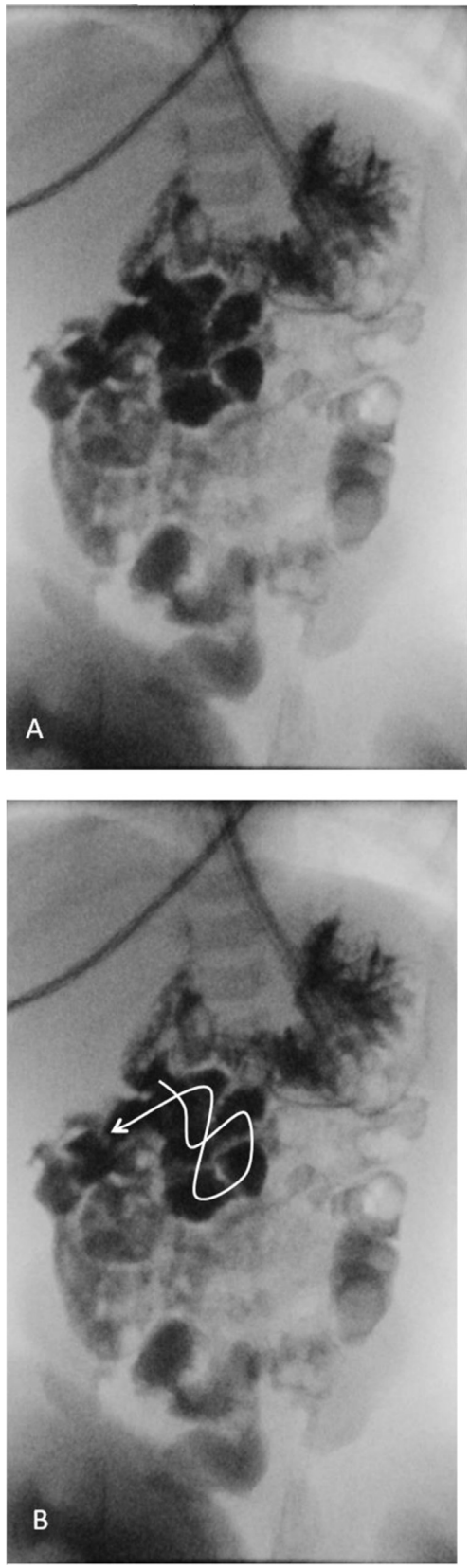

Fig. 3 – (A) Supine upper GI series image. The duodenum does not cross midline and instead descends inferiorly and back to the right side without ascending toward the Ligament of Treitz. (B) The observed path of the duodenum is traced.

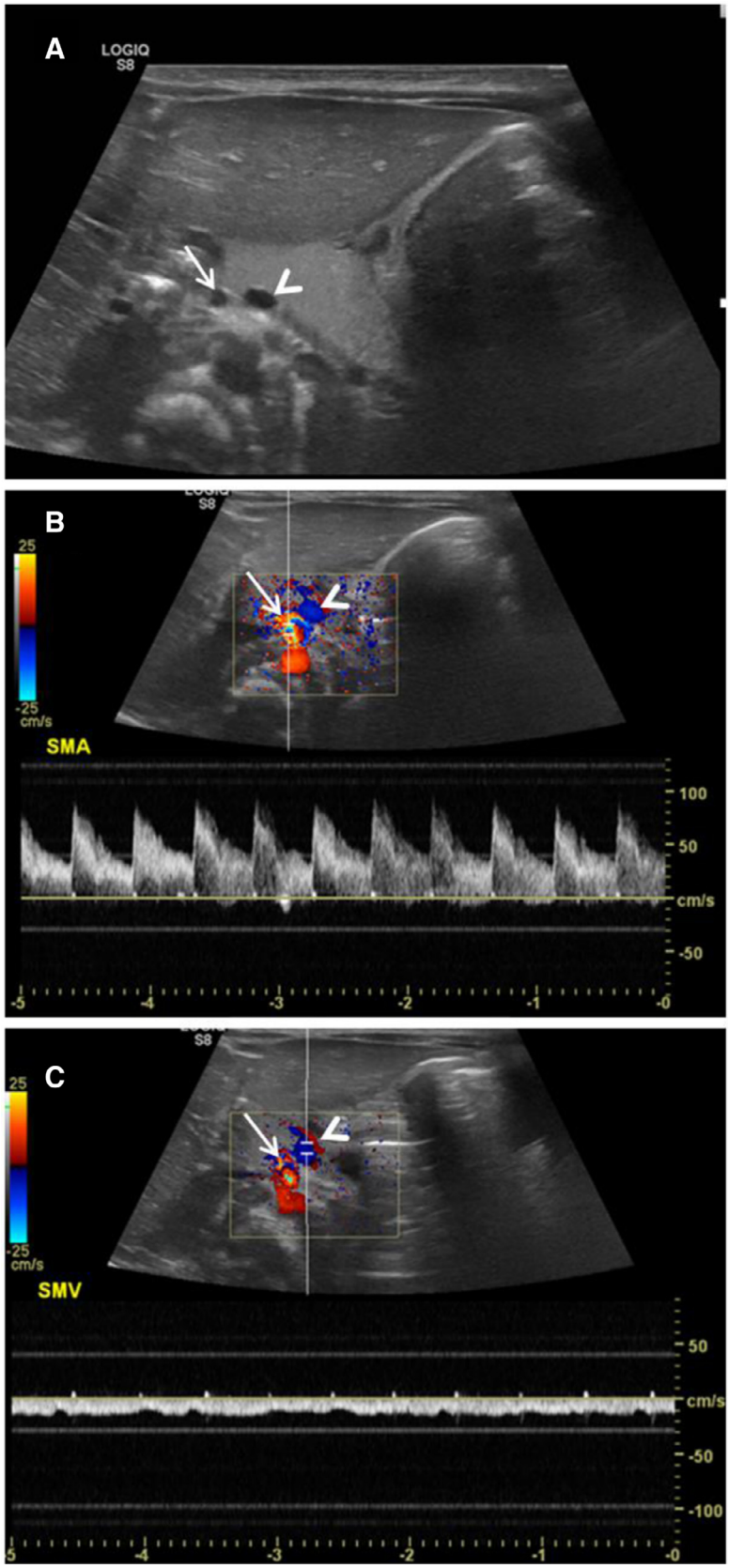

Fig. 4 – Abdominal ultrasound demonstrating the reversed relationship of the SMA (arrow) and SMV (arrowhead). The SMA typically sits on the left side of the SMV. (A) SMA and SMV identified. (B) Color Doppler and spectral waveforms of the vessel on the anatomic right demonstrate arterial flow, representing the SMA. (C) Venous waveforms are identified on the anatomic left, representing the SMV.

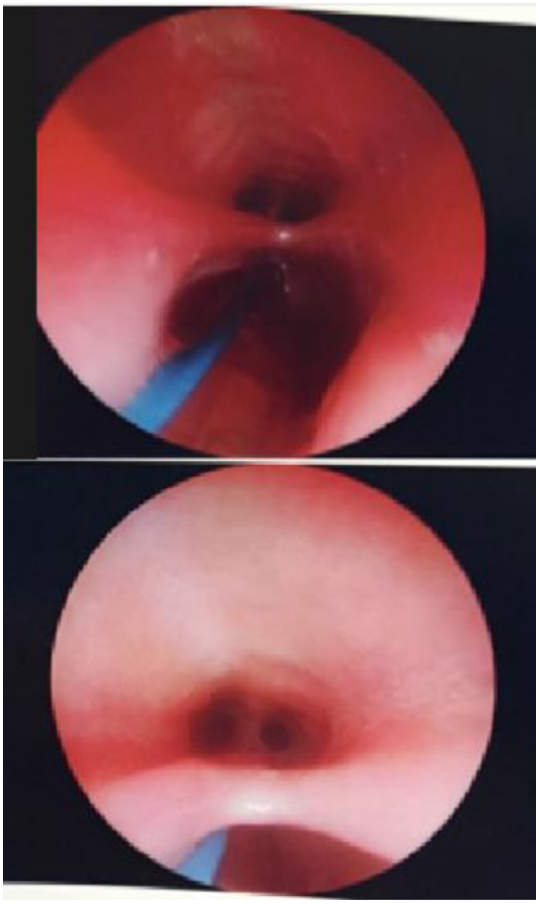

Fig. 5 – Laryngoscopic view of the aerodigestive tract, with nasogastric tube (blue stripe) inserted into the esophagus, demonstrating a large defect of the posterior tracheal wall extending within 1 cm from the carina.

body anomalies, cardiac enlargement, radial ray anomalies, and esophageal atresia. In this case, the combination of anorectal malformation and vertebral body anomalies led to the suspected diagnosis of VACTERL.

Computed tomography imaging may be useful in visualizing a direct communication between the trachea and esophagus; however, an esophagram is most useful in evaluating laryngeal clefts and distinguishing them from tracheoesophageal fistulas [2,5]. Isolated tracheoesophageal fistulas without esophageal atresia, also known has H- or N-type fistulas, have a narrow connection between the trachea and esophagus that is sometimes only seen while the patient is in the prone position. Endoscopy is not 100% sensitive, which may also lead to delayed diagnosis [6]. Type II-IV laryngeal clefts, on the other hand, have a larger defect that is easily seen on lateral imaging [5]. Type I laryngeal clefts are usually only visible on laryngoscopy [1]. Identifying involvement of the larynx is important in differentiating between the two entities. Although it may be difficult to distinguish between the exact type of cleft on imaging, it is important to note how far the defect extends on imaging and whether the trachea is involved. Laryngoscopy is gold standard for diagnosis and classification [2,3].

Treatment for laryngeal clefts varies depending on the severity. Minor clefts can sometimes be managed conservatively; however, most laryngeal clefts require repair. The extent of surgery depends on the type with low-grade clefts often repaired endoscopically and high-grade clefts treated with open surgical intervention [3]. Although surgical intervention is often required for both laryngeal clefts and tracheoesophageal fistulas, the approach differs and highly specialized care is needed for the severe type laryngeal clefts to avoid complications.

Laryngeal clefts have been previously reported; however, since they are rare, most radiologists may not be familiar with the diagnosis. The incidence of laryngeal cleft may be higher than previously reported and thus radiologists should be aware of this condition. This case stresses the importance of accurate diagnosis of laryngeal clefts and a thorough work up to evaluate for associated syndromes and anomalies.

Conclusion

Laryngeal clefts may be difficult to diagnose due to lack of familiarity with this entity. Esophagram is useful in distinguishing the laryngeal clefts from tracheoesophageal fistulas. Patients should undergo laryngoscopy and subsequent surgical treatment at a dedicated pediatric center in order to avoid misdiagnosis and mistreatment. Identification of associated anomalies is also important and the radiologist plays an important role in the evaluation of these patients.

Conflict of Interest

None.

REFERENCES

[1] Johnston DR, Watters K, Ferrari LR, Rahbar R. Laryngeal cleft: evaluation and management. Int J Pediatr Otorhinolaryngol 2014;78:905–11. doi:10.1016/j.ijporl.2014.03.015.
[2] Leboulanger N, Garabedian EN. Laryngo-tracheo-oesophageal clefts. Orphanet J Rare Diseases 2011;6:81. doi:10.1186/1750-1172-6-81.
[3] Rahbar R, Rouillon I, Roger G, Lin A, Nuss RC, Denoyelle F, et al. The presentation and management of laryngeal cleft: a 10-year experience. Arch Otolaryngol Head Neck Surg 2006;132:1335–41. doi:10.1001/archotol.132.12.1335.
[4] Fraga JC, Adil EA, Kacprowicz A, Skinner ML, Jennings R, Lillehei C10.1002/lary.24804. The association between laryngeal cleft and tracheoesophageal fistula: myth or reality? Laryngoscope 2014;125(2):469–74. doi:10.1002/lary.24804.
[5] Tao TY, Menias CO, Herman TE, McAlister WH, Balfe DM. Easier to swallow: pictorial review of structural findings of the pharynx at barium pharyngography. Radiographics 2013;33(7):e189–208. doi:10.1148/rg.337125153.
[6] Fallon SC, Langer JC, St Peter SD, Tsao K, Kellagher CM, Lal DR, et al. Congenital H-type tracheoesophageal fistula: a multicenter review of outcomes in a rare disease. J Pediatr Surg 2017. doi:10.1016/j.jpedsurg.2017.05.002. epub ahead of print.