Bronchiectasis in the setting of aplasia of the epiglottis

Puwakdandawe Weerasinghe, Rahul Thomas, Brent Masters & Nitin Kapur

Department of Respiratory and Sleep Medicine, Queensland Children’s Hospital, Brisbane, Queensland, Australia.

Keywords
Aplasia, epiglottis, bronchiectasis, aspiration.

Abstract
Aplasia of the epiglottis is a rare airway abnormality requiring airway and feeding interventions. We report a case of bronchiectasis in the setting of congenital aplasia of the epiglottis, secondary to early-life aspiration events. Compensatory mechanisms for airway protection likely develop later in life.

Introduction
The epiglottis has been suggested to provide upper airway stability and protection [1,2]. Abnormalities of the epiglottis can occur, including hypoplasia and aplasia, where the latter is significantly more uncommon, and the exact incidences are unknown. The presentation of aplasia of the epiglottis can vary in nature and timing depending on other laryngeal abnormalities due to common embryological origin. Long-term aspiration is a known cause of chronic pulmonary suppuration and the eventual development of bronchiectasis [3,4]. The case presented here is the first reported case of bronchiectasis as a likely consequence of early-life aspiration events due to aplasia of the epiglottis.

Case Report
A 2-year-old male was referred for investigation of recurrent lower respiratory tract infections. On history, he was a term baby who was admitted to the special care nursery at birth for 12 days for suspected sepsis. His mother had gestational diabetes under good control. He had a complex medical background with dysmorphic features (low set ears, clinodactyly, micrognathia, and multiple ear creases), multiple midline malformations (left soft palate, penoscrotal abnormality, multicystic right testicle, Atrial Septal Defect (ASD) and Ventricular Septal Defect (VSD), everted eyelids), feeding difficulty, abnormal cry, hearing impairment and speech delay. Renal and cranial ultrasounds were normal. Neonatal screening was negative for cystic fibrosis. Neurological assessment at birth and subsequently was normal.

In the first 12 months of life, he had recurrent upper respiratory tract infections, and some of the episodes were associated with wheeze. He also had protracted episodes of wet cough with or without viral or febrile illness. There was no history suggestive of upper airway obstruction, but his cry was noted to be soft. He had feeding difficulty and failure to thrive with weight below the third centile. Feeding difficulty was attributed to the cleft palate. Both VSD and ASD had spontaneous closure in the first year of life. Cleft palate was repaired at 10 months of age without any major complications. Despite the repair, the child continued to have recurrent lower respiratory tract infections and chronic wet cough with bilateral crackles, even during periods of wellness. A chest X-ray at the time of referral showed prominent bronchovascular markings with airspace change in the right middle lobe and left lower lobe. The wet cough persisted despite prolonged courses of oral antibiotics. He was thus further investigated for the cause of this chronic wet cough with a computed tomography (CT) chest, flexible bronchoscopy and immunological investigations.
Immunological and aero-allergen screening tests all demonstrated normal limits. Flexible bronchoscopy indicated the absence of an epiglottis. The appearance was consistent with agenesis or aplasia of epiglottis (Figs. 1–2 and Video S1, Supporting Information). Other findings included widespread bronchitis, mucous plugging of right middle lobe, and branching anomalies. Vocal cord function was normal. Bronchoalveolar lavage fluid (BALF) showed neutrophilia (92% cells neutrophils) and cultured *Pseudomonas aeruginosa, Hemophilus influenzae* and *Streptococcus pneumoniae* in significant amounts. CT chest showed atelectasis in the right middle lobe with mild multi-lobar bronchiectasis, most prominent in the lower lobes bilaterally. He was treated with intravenous anti-Pseudomonal antibiotics and was started on oral azithromycin for 12 weeks. He was also started on airway clearance therapy with chest percussions. Video fluoroscopic swallow studies (VFSS) were performed where the child demonstrated prompt swallow triggers and adequate airway protection and was thus not recommended for a modified diet.

Currently, at 4 years of age, he is progressing well with resolution of his wet cough. He is maintained on prophylactic macrolides and chest physiotherapy and remains free of chronic cough and is regularly assessed by speech therapists and respiratory physicians. He continues to have mild speech delay but is fully orally fed and is otherwise neurologically normal. He has a small deficiency in the posterior part of the soft palate resulting in intermittent nasal regurgitation. He is observed to have early digital clubbing.

**Discussion**

Congenital absence of the epiglottis is a rare condition. It is more commonly described in the literature as part of a syndrome, such as Nager syndrome, short polydactyly syndrome of the Majewski type, Pierre Robin sequence, or other non-specific syndromic features [5]. Isolated congenital aplasia of the epiglottis is less commonly described in the literature (Table 1).

Development of the epiglottis begins around day 33 of life and is complete by day 48 of life, concurrent with the development of limbs and digits. Disruptions to organogenesis during this period result in abnormalities found in oro-faciodigital syndromes in addition to abnormalities of the epiglottis [1,5].

The condition may be an incidental finding or it may present as aspiration events with associated pneumonitis, recurrent respiratory tract infection, or upper airways obstruction. Most cases of aplasia of the epiglottis in the literature have been managed with tracheostomy and feeding interventions. Our child certainly had significant feeding difficulty early in life, which was attributed to his cleft palate; it is unclear if the absence of epiglottis contributed to this feeding and swallowing difficulty. It is likely that the high frequency of lower respiratory tract infections despite the palatal surgery were consequent to persistent aspiration due to an absent epiglottis.

Recurrent aspiration secondary to structural causes, such as trachea–oesophageal fistula, or neurological impairment, such as cerebral palsy, are known reported causes of...
### Table 1. Summary of case reports on aplasia of epiglottis.

| Study | Presentation | Interventions | Outcome |
|-------|--------------|---------------|---------|
| Constantinides and Cywes [6] | 6-day-old female presented with episodes of choking and cyanosis in the context of complete median cleft of the mandible and suspected Pierre Robin Sequence | Tracheostomy | Tracheostomy obstruction led to death few weeks later |
| Reyes et al. [2] | 3-month-old female presented with stridor and subsequent obstructive sleep apnoea with no aspiration on repeated swallow studies | No interventions | At 8 years of age, she was being advised for weight loss for OSA |
| Koempel and Hollinger [1] | 3-month-old female presented with work of breathing and stridor | Fundoplication and gastrostomy feeds | At 17 months, she was not attempting vocalization or swallowing |
| Bonilla et al. [7] | Newborn with stridor, work of breathing and aspiration pneumonitis on a background of dysmorphic features | Tracheostomy | Successful decannulation at 7 years of age |
| Dritsoula and Thevasagayam [8] | Newborn with retrognathia and presented with stridor. Issues with initiation and propagation of swallow were demonstrated on swallow studies | Fundoplication and PEG insertion at 7 weeks. Glycopyrrolate for secretions | Well at 11 months but continued total gastrostomy feeds |
| Tay et al. [5] | 3-year-old female with abnormal swallow test on the background of Nager syndrome | Tracheostomy and nasogastric feeds | Not detailed |
| Tay et al. [5] | 18-month-old female with VFSS demonstrated aspiration on the background of Nager syndrome | PEG feeds | Not detailed |
| Shahi and Singh [9] | Incidental finding in 30-year-old female with no symptomology at elective procedure | None | Not detailed |
| Kim et al. [10] | Incidental finding in 33-year-old female presenting with acute tonsillitis | None | Acute tonsillitis resolved. Observed to have compensatory action by hypertrophied lingual tonsil on video fluoroscopic barium swallow test |
| Prasad [11] | Six cases of babies 1–3 days old with acute severe respiratory distress, cyanosis and bradycardia in the context of isolated agenesis of epiglottis | Supraglottic closure with temporary tracheostomy and NG feeding. Glottis restored 3–6 months postoperatively when satisfied with pharyngoesophageal coordination | Five of six cases well and thriving at the 6-month follow up. One case passed before intervention could take place |
recurrent lower respiratory tract infections and bronchiectasis in children [13]. To our knowledge, bronchiectasis has never been reported with absent epiglottis (Table 1).

The role of the epiglottis in airway protection is unclear. Surgical epiglottectomy in adults has not been associated with swallowing difficulty or increased aspiration risk [14]. However, swallowing impairment and aspiration are present in many cases of absent epiglottis described here, particularly at a young age [5–8]. Whether its link to bronchiectasis in our case was causal remains conjectural as we did not find any other cause for his underlying bronchiectasis. It is also postulated that compensatory airway-protective mechanisms may be acquired with growth in otherwise neurologically intact children with normal vocal cord function, even in the absence of epiglottis. This may explain the absence of significant aspiration in his swallow study at 3 years of age. This would also be supported by the absence of swallowing impairment in the two adult cases of congenital epiglottic aplasia. Kilinc [12] describes an objective improvement in aspiration, with swallow rehabilitation further demonstrating learned compensation in the absence of epiglottis. Regardless, aspiration prevention measures have been undertaken in a majority of the cases described in the literature in the management of these patients. In the absence of these measures, as in our case, the first presentation of aplasia of the epiglottis may be the sequela of aspiration, such as recurrent respiratory tract infections and bronchiectasis.

Disclosure Statement

Appropriate written informed consent was obtained for publication of this case report and accompanying images.

References

1. Koempel JA, and Holinger LD. 1998. Congenital absence of the epiglottis. Int. J. Pediatr. Otorhinolaryngol. 45:237–241.
2. Reyes BG, Arnold JE, and Brooks LJ. 1994. Congenital absence of the epiglottis and its potential role in obstructive sleep apnea. Int. J. Pediatr. Otorhinolaryngol. 30:223–226.
3. Piccione JC, and Boesch RP. 2009. Bronchiectasis due to chronic pulmonary aspiration in children. Chest 136(4):8S.
4. Simon M, and Collins M. 2013. The pediatric lung and aspiration. Perspect. Swallowing Swallowing Disord. 22:142–154.
5. Tay SY, Loh WS, and Lim TC. 2017. A case report of absent epiglottis in children with Nager syndrome: its impact on swallowing. Cleft Palate Craniofac. J. 54:754–757.
6. Constantinides CG, and Cywes S. 1983. Complete median cleft of the mandible and aplasia of the epiglottis. A Case Report. S. Afr. Med. J. 64:293–294.
7. Bonilla JA, Pizzuto MP, and Brodsky LS. 1998. Aplasia of the epiglottis: a rare congenital anomaly. Ear Nose Throat J. 77:51–55.
8. Dritsoula AK, and Thevasagayam MS. 2015. Congenital aplasia/hypoplasia of the epiglottis—a case report and a review of the literature. Int. J. Pediatr. Otorhinolaryngol. 79:1609–1612.
9. Shahi V, and Singh DK. 2015. Asymptomatic absent epiglottis: a case report. Int. J. Anat. Res. 3:945–946.
10. Kim YJ, Myung NS, Lee HJ, et al. 2014. A case of congenital absence of the epiglottis in an adult. Am. J. Otolaryngol. 35:673–675.
11. Prasad RG. 2015. Congenital isolated agenesis of epiglottis presenting with acute life-threatening event successfully managed by temporary Supraglottic closure and tracheostomy. Int. J. Phonosurg. Laryngol. 5:1–3.
12. Kilinc HE, Arslan SS, Demir N, et al. 2018. Swallowing therapy for a case of congenital absence of the epiglottis. Clin. Exp. Health Sci. 8:70–72.
13. Kapur N, Masters IB, Morris PS, et al. 2012. Defining pulmonary exacerbation in children with non-cystic fibrosis bronchiectasis. Pediatr. Pulmonol. 47:68–75.
14. Leder SB, Burrell MI, and Van Dale D. 2010. Epiglottis is not essential for successful swallowing in humans. Ann. Otol. Rhinol. Laryngol. 119:795–798.

Supporting Information

Additional Supporting Information may be found in the online version of this article at the publisher’s web-site: http://onlinelibrary.wiley.com/doi/supinfo.

Video S1. Bronchoscopic video of aplasia of the epiglottis.