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Laryngopharyngeal Congenital Anomalies Diagnosed in Adulthood: Hypervigilance in Childhood Needed to Prevent Permanent Physical and Psychosocial Sequelae

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Abstract

While often diagnosed in childhood during evaluation for cough, dysphonia and/or dysphagia, congenital laryngopharyngeal anomalies may not become clinically significant until adulthood. The clinical presentation of two adults with dysphagia resulting from significant congenital laryngopharyngeal anomalies and psychological and physical sequalae of delayed diagnosis are described.

Keywords: Congenital anomalies, Dysphonia, Dysphagia

Abbreviations

MBSS: Modified Barium Swallow Study; **CaHA:** Calcium Hydroxylapatite; **VHI:**Voice Handicap Index

Introduction

Congenital anomalies of the larynx and pharynx typically manifest with stridor, dysphagia and/or dysphonia early in infants leading to prompt evaluation and possible intervention. Well-described anomalies include laryngomalacia, subglottic stenosis or hemangioma, laryngeal cleft, laryngeal web, vocal fold paralysis and laryngeal or saccular cysts [1-4].

The development of head and neck anatomy in human embryogenesis is quite complex, and aberrant development may lead to either isolated anomalies or craniofacial syndromes, such as Treacher Collins or DiGeorge Syndrome. As a brief summary, head and neck structures begin to develop around 4-5 weeks gestation with the formation of the branchial arches, which are outpouchings of mesoderm and neural crest cells lined by ectoderm. Each branchial arch is associated with a cranial nerve and artery, and the mesoderm primarily forms structures such as muscle, cartilage, or other connective tissue. In contrast, pharyngeal pouches contain endoderm and develop from the embryonic foregut; therefore, derivatives of pharyngeal pouches tend to be more closely related to the digestive tract and/or glandular structures. As an example, the second pharyngeal pouch involves the development of the tonsillar fossa and palatine tonsils [5].

Although some infants display severe symptoms related to congenital laryngopharyngeal anomalies, such as stridor, cyanosis and respiratory distress, anomalies may also present with more subtle symptoms. It is easy in such cases to leave congenital anomalies off of the differential, however it is important to remain hypervigilant in order to get the appropriate workup and care for the pediatric patient. In this report, two cases of adults with delayed diagnosis of congenital laryngopharyngeal anomalies are discussed, as well as the physical and psychosocial sequalae from treatment delay.

Case 1: The CRNA with Low O_2

A 40-year-old slender, athletic, white male CRNA presented to clinic with a 10-year history of progressive cough and dysphagia, particularly to powdery and spicy foods. As a child, his symptoms were mistaken for allergies and his parents treated him mostly with decongestants. He sought

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evaluation and treatment at this time after experiencing vasovagal syncope after a bout of coughing. His only previous workup was a modified barium swallow study (MBSS) at an outside hospital, which showed a flash subglottic penetration without aspiration. He was never diagnosed with aspiration pneumonia and denied any changes or concerns with his voice.

A rigid laryngoscopy with stroboscopy was performed at his initial clinic visit showing good vocal fold motion; there was, however, a slight asymmetry noted on vocal fold abduction and he appeared to lack cartilage in his post cricoid region although overlying mucosa was intact (Figure 1A). The decision was made to obtain further imaging, including a repeat modified barium swallow, before proceeding with any surgical intervention.

A CT soft tissue neck with contrast was obtained showing soft tissue in the region of cricoid lamina, depicting either an incomplete mineralization or congenital absence (Figure 1B). MBSS revealed a tight pharyngoesophageal segment, tongue base weakness, trace aspiration with nectar-thick liquids secondary to delay and some regurgitation (Figure 1C). It was believed that the deficiency of the patient's cricoid lamina led to soft tissue collapse and a tight pharyngoesophageal segment, resulting in delayed swallow with regurgitation and penetration.

The patient was taken to the OR for suspension microlaryngoscopy with injection of Restylane (hyaluronic acid gel) into the interarytenoid space. Restalyne® acts a temporary filler and patients are counseled that results typically last about 3 months before this substance is absorbed; this is done initially to ensure patients experience improvement in their symptoms before a more permanent substance is injected. Interestingly, during the procedure the patient was unable to tolerate standard jet ventilation and required intubation. Despite ventilation with 100% FiO_2, his oxygen saturations only reached a maximum of 97-98% and would intermittently drop his SpO_2 into the low 90s. A type II posterior laryngeal cleft was palpated. Restylane was injected directly in the interarytenoid area and medial aspect of the arytenoid processes bilaterally. The patient tolerated the procedure well. The only complication was postoperative pneumonia, which was treated with a 10-day course of doxycycline.

At his 1-month postoperative visit, the patient reported dramatic improvement in his symptoms. He no longer experienced coughing or choking episodes and was very pleased with these results. Approximately 7 months following his initial surgery, however, the patient returned with concerns that his symptoms started to slowly return about 3 months after the Restalyne injection. The patient was counseled on proceeding with a permanent injection, direct suturing of his cleft, or placement of a cartilage implant. Given the risk of cricoarytenoid joint fixation with direct suturing or cartilage implantation, and given his positive results with the temporary injection, the patient decided to proceed with suspension microlaryngoscopy with interarytenoid injection of calcium hydroxylapatite (CaHA).

During his second procedure, he again needed to be intubated to maintain adequate O_2 saturations. A total of 0.3 cc CaHA was placed in the interarytenoid area. At his 1-month postoperative visit, he once again reported resolution of his coughing episodes. Overall, the patient was very pleased with the outcome and was instructed to follow up as needed.

Case 2: The Man with a "Gurgly" Voice

A 37-year-old white male presented to clinic with solid food and pill dysphagia, as well as a gurgly, breathy dysphonia since puberty. Prior to puberty, his voice was slightly high-pitched but otherwise normal. He denied any previous smoking history, neck trauma, or caustic injections. His Voice Handicap Index-10 (VHI-10), a validated survey assessing quality of life in patients with dysphonia, was 24 on presentation; a VHI score above 11 is considered abnormal [6]. He was noted to be a very socially reserved individual who continued to live at home with his parents and whose employment is to construct fiberoptic light cables.

On his initial clinic visit, a flexible laryngoscopy with stroboscopy was performed revealing a deep-seated larynx with an abnormal band of soft tissue extending around the vallecula to the posterior glottis. Although this was the most striking feature, he also exhibited abnormal vibratory movement of the vocal folds and a significant glottic gap from vocal fold atrophy (Figure 2). A CT soft tissue neck with contrast was obtained confirming that his defect was entirely soft tissue as seen on his laryngoscopy. There were no masses or lesions visualized.

MBSS was also performed revealing moderate to severe pharyngeal dysphagia due to limited epiglottic inversion and hyoid movement from the soft tissue band. He aspirated both thin and nectar thick consistencies. These findings were discussed at length with the patient and it was hypothesized that this abnormal band of tissue was a second pharyngeal arch congenital anomaly. Suspension microlaryngoscopy with lysis of his abnormal tissue band with CO_2 laser was recommended.

Intraoperatively, a CO_2 laser was used at 10W to address the tissue extending down the posterior pharyngeal wall. There was a noticeable release

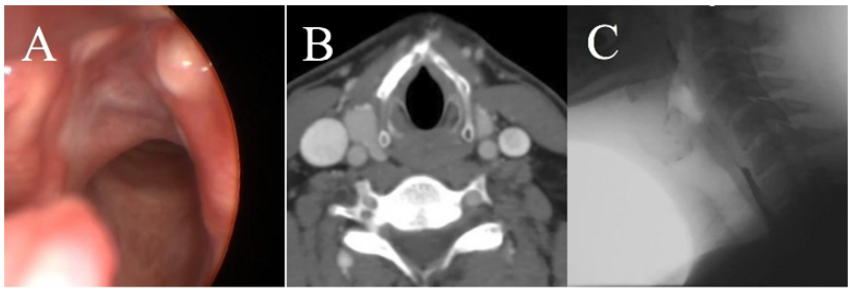

Figure 1: Pre-operative laryngoscopy and imaging for case 1. Panel **A-** shows the patient's pre-operative laryngoscopy with interarytenoid mucosa intact, but no evidence of cartilage support. Panel **B-** depicts lack of mineralization of or incomplete cricoid lamina. Panel **C-** shows trace aspiration of nectar-thick consistencies of barium.

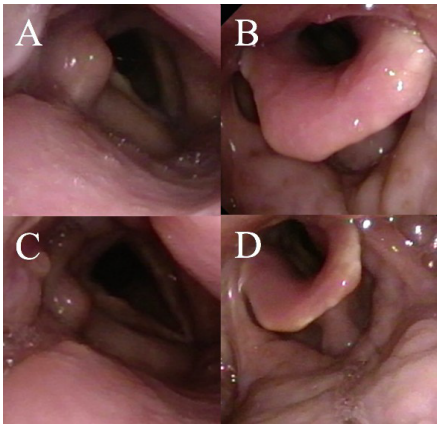

Figure 2: Pre- and post-operative laryngoscopy for case 2. Panels **A** and **B**- show the patient's initial clinic laryngoscopy. He exhibits R > L vocal fold atrophy, a deep-seated larynx, and an abnormal band of soft tissue extending from lingual tonsil to around the posterior glottis. His post-operative images following his most recent procedure are shown in panels **C** and **D**. Improved medicalization of the right vocal fold after temporary gel injection and an elevated and open position of the larynx are appreciated.

of a white scar band, with greater scar burden seen on the right compared to the left. In order to prevent a scar band forming between the epiglottis and the lateral pharyngeal wall on the right, a conservative scar lysis was performed with the knowledge that additional surgeries may be indicated. Topical mitomycin was then applied to the treated areas for 3 minutes to prevent scar reformation. A biopsy of the soft tissue was obtained, which revealed "dense fibroconnective tissue, consistent with scar."

The patient followed up in 2 weeks and reported significant improvement in his dysphagia, although no change in his voice. On in-clinic laryngoscopy, the position of his larynx was noted to be slightly more elevated after his scar band release. A second-stage procedure was recommended to release additional scar bands and to perform a Restalyne® injection to temporarily augment the patient's vocal folds and improve dysphonia.

During the second suspension microlaryngoscopy, only minimal release of the additional left scar band using the CO_2 laser was completed due to bleeding. Bilateral vocal fold Restalyne® injections were performed without complication. The patient followed up 3 weeks postoperatively. His voice was noted to be stronger and easier to understand per his family. His VHI dropped to 13, a marked improvement compared to his baseline of 24. There were no reported changes in his swallowing compared to the first surgery.

He was then enrolled in an intensive voice remediation program with Speech Language Pathology. A repeat MBSS was obtained, showing improvement in his epiglottic inversion although still incomplete due to some persistent scar bands. Throughout this time, the patient noted a slight decline in his voice as the temporary gel injection in his vocal folds began to absorb.

He was taken back to the OR for suspension microlaryngoscopy, repeat scar lysis and repeat Restalyne® injection. Due to the bleeding previously encountered on the left, the CO_2 laser was used to address additional scar band remaining on the right, taking great care to avoid the aryepiglottic fold. After this, vocal fold augmentation was performed with injection of additional Restalyne® bilaterally.

The patient presented for a 1-month postoperative visit with dramatic improvement in his voice. His pitch was lower and overall more appropriate, and his voice demonstrated less of a "gurgly" quality. His dysphagia remained stable. Figure 2 displays his most recent laryngoscopy. At this time, the patient continues to participate in voice therapy. At his next visit, a permanent vocal fold injection will be discussed given the patient's dramatic improvement in his dysphonia with previous temporary injections, as well as additional slow and controlled releases of his remaining scar bands in order to improve the patient's hyolaryngeal excursion during swallow.

Discussion

The pediatrician and parents must remain hypervigilant in cases where patients have persistent cough, dysphonia, dysphagia, or breathing difficulties after ruling out common etiologies. As demonstrated in this report, treatment delays for patients with congenital laryngopharyngeal anomalies can result in significant physical and psychosocial sequalae for the patient and their families.

Laryngeal clefts result from the incomplete fusion of the cricoid lamina, as was depicted in Case 1. Although there are different classifications for laryngeal clefts, the Benjamin-Inglis system is most commonly used to distinguish clefts by their location: Type I (interarytenoid), Type II (partial or complete cricoid), Type III (cervical trachea) and Type IV (intrathoracic or to carina) [7]. Patients most commonly present with chronic cough, as well as aspiration and recurrent pneumonia. Unless the patient has a pre-existing condition that exacerbates mild symptoms (i.e. cardiac disease), Type I and Type II clefts are often misdiagnosed due to delayed referral to an Otolaryngologist for microlaryngoscopy and bronchoscopy [8]. For this reason, the exact prevalence of laryngeal cleft remains unclear, but a prospective longitudinal study by Chien et al. found the incidence of Type I laryngeal cleft to be 7.6% [8-9]. In Case 1, the patient did not exhibit any recurrent respiratory infections that may have alerted parents and providers to the need for further workup; however, his chronic cough should have undergone further evaluation when allergy treatment was ineffective. This patient was an otherwise healthy 40-year old male with low O_2 reserve from years of undiagnosed microaspiration events. It is very uncommon for a healthy patient with a normal weight to not tolerate jet ventilation; however, in his case, he could only achieve a maximum of 98% SpO_2 while intubated with 100% FiO_2. If his chronic cough had been recognized earlier and not discarded for allergies, then this physical sequalae of chronic aspiration could have been prevented.

The second case demonstrates an individual with a rarer anomaly when compared to laryngeal cleft. There are two case reports of "pharyngolaryngeal bands" with abnormal soft tissue extending from base of tongue to the lateral pharyngeal walls, though to be a 2nd pharyngeal pouch anomaly. In both of these case reports, the diagnosis of a congenital anomaly was made during infancy or childhood [10,11]. Fayoux et al. reported a newborn with severe respiratory distress at birth requiring intubation at birth and was referred to an ENT for continued severe apneas at 1 month of age [10]. Prescott described a child who presented with this anomaly in the setting of interstitial pulmonary fibrosis [11]. The case described in this report is unique in that the patient had a noticeably abnormal voice, yet referral to an Otolaryngologist and subsequent diagnosis was delayed until adulthood. This delay led to decreased social interaction. Although he does have a voice, his ability to communicate effectively was diminished by his pathology and this led to more isolation and a reserved personality, as well as the selection of a job that does not require verbal communication. If he had been referred to an Otolaryngologist after symptoms became prominent around puberty, perhaps interventions to provide a more functional voice during adolescence would have decreased the psychosocial sequalae of this patient's congenital anomaly.

Although the anomalies discussed are rare and may be easy to misdiagnose, there should be a low threshold to investigate potential congenital anomalies as a source of the patient's symptoms when common etiologies are excluded. In cases such as these, an in-clinic laryngoscopy performed by a Pediatric Otolaryngologist or Laryngologist can be an effective tool to see if structural laryngopharyngeal anomalies exist.

Conclusion

Persistent cough, dysphagia, or dysphonia can be minimized by parents and physicians if a child does not become acutely ill and routine etiologies are excluded. The possibility of congenital laryngopharyngeal anomalies should be heightened in these cases to prevent severe physical and psychosocial sequelae of undiagnosed congenital anomalies.

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