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Mini-symposium: Upper airway abnormalities

Rare Upper Airway Anomalies

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EDUCATIONAL AIMS

- To discuss the normal embryology of the upper airway and how missteps in these complex developmental pathways give rise to various congenital defects
- To describe the clinical presentation of a select group of rare airway anomalies, including their effect on the functions of breathing, feeding, and phonation
- To review the diagnostic tools used for workup of these disorders and briefly discuss surgical and medical management strategies

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SUMMARY

A broad spectrum of congenital upper airway anomalies can occur as a result of errors during embryologic development. In this review, we will describe the clinical presentation, diagnosis, and management strategies for a few select, rare congenital malformations of this system. The diagnostic tools used in workup of these disorders range from prenatal tests to radiological imaging, swallowing evaluations, indirect or direct laryngoscopy, and rigid bronchoscopy. While these congenital defects can occur in isolation, they are often associated with disorders of other organ systems or may present as part of a syndrome. Therefore workup and treatment planning for patients with these disorders often involves a team of multiple specialists, including paediatricians, otolaryngologists, pulmonologists, speech pathologists, gastroenterologists, and geneticists.

INTRODUCTION

The upper aerodigestive tract extends from the nose and mouth proximally to the trachea and oesophagus distally and is critical in the functions of breathing, feeding, and speech. A variety of congenital anomalies can occur at any subsite in this system, having variable effects on these functions. These anomalies are the result of embryologic missteps during organogenesis. Here, we will briefly review the normal development of the upper airway and then describe several rare congenital anomalies affecting the upper airway.

EMBRYOLOGY OF THE UPPER AIRWAY

Development of the face, neck, and pharynx begins early in the fourth week of embryologic development with the appearance of the pharyngeal arches [1]. These accumulations of tissue on either side of the foregut consist of an outer ectodermal layer, a middle mesenchymal layer, and an inner endodermal layer; the arches are separated externally by pharyngeal clefts and internally by pharyngeal pouches. Development of the face involves the formation and fusion of two pairs of maxillary and mandibular prominences, derivatives of the first pharyngeal arches, and the fronto-nasal prominence, a product of neural crest cell expansion from the midbrain and forebrain. These prominences form the boundaries of the primitive oral cavity. As these structures grow and fuse, they form the structures of the face. Incorrect fusion can result in the various congenital facial clefts. Ultimately, the fronto-nasal prominence forms the forehead and dorsal and apical part of the nose; the lateral nasal prominences go on to form the nasal alae; the medial nasal prominences form the nasal septum,

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ethmoid bone, and cribriform plate; the maxillary prominences form the upper cheek and upper lip; and the mandibular prominences form the chin, lower lip, and lower cheek [2].

Development of the larynx and trachea begins in the fourth week of gestation with a ventral out-pouching of the primordial pharynx called the lung bud, or respiratory diverticulum [3]. This diverticulum grows ventrally and caudally, eventually branching into left and right bronchial buds which are precursors to the lungs. The main trunk of the diverticulum will form the trachea and larynx. At the same time, folds of tissue develop longitudinally in the lung bud and fuse, forming the trachea-oesophageal septum and separating the foregut into the oesophagus and laryngotracheal tube. The endoderm of this tube forms the laryngeal epithelium while the laryngeal cartilages and muscles are derived from mesenchyme from the fourth and sixth pharyngeal arches. Proliferation of mesenchyme forms arytenoid swellings near the opening of the laryngotracheal tube and the hypopharyngeal eminence, laying just superiorly. The hypopharyngeal eminence gives rise to the supraglottic structures, epiglottis, and tongue base. Meanwhile, the laryngeal epithelium proliferates, occluding the lumen of the larynx temporarily. By ten weeks of gestation, the lumen recanalises, creating the normal glottic opening and the laryngeal ventricle.

PYRIFORM APERTURE STENOSIS

Congenital nasal pyriform aperture stenosis (CNPAS) is a rare cause of upper airway obstruction in the newborn, thought to be caused by bony overgrowth of the nasal process of the maxilla in utero resulting in a narrowing of the anterior-most bony nasal airway [4,5]. Patients present with signs and symptoms of nasal obstruction including feeding difficulties, tachypnea, apneic or cyanotic episodes that resolve with crying or mouth-breathing, and difficulty passing a suction catheter via the nose [5,6]. Symptoms may range from mild to life-threatening, depending on the degree of stenosis. Initial evaluation may include anterior rhinoscopy or nasal endoscopy, which reveals narrowed nasal passages and inability to pass an endoscope. Clinical presentation is similar to that of bilateral choanal atresia, and therefore thin-cut axial CT should be obtained for confirmation of the diagnosis and possible surgical planning. (Figure 1) A pyriform aperture width of less than 11 mm in a term infant is considered diagnostic [7]. CNPAS may occur as an isolated defect or in association with other craniofacial anomalies. Most commonly, it has been associated with holoprosencephaly and particularly, a solitary median central maxillary incisor, a midline defect linked to the holoprosencephaly spectrum [6]. A solitary median maxillary incisor has been reported in up to 60% of cases of CNPAS [6]. Abnormalities of the hypothalamic-pituitary axis have also been reported in up to 40% of patients with CNPAS. [8] Brain MRI as well as genetics and endocrine evaluation has therefore been suggested as part of the workup for these patients [5,8,9].

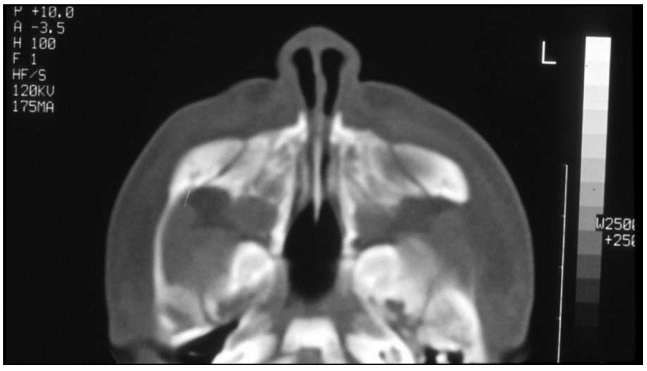

Figure 1. CT of piriform aperture stenosis.

Patients with CNPAS can often be managed conservatively with placement of an oral airway, nasal decongestants, frequent suctioning, intranasal steroid drops, and humidification [10]. As they grow, their stenosis may cease to cause symptomatic airway obstruction. However, failure of these conservative measures, persistent need for invasive ventilation, or failure to thrive are indications for surgical repair [10]. In one series of 10 patients with CNPAS, 50% of patients ultimately required surgical repair after failing medical managements [6]. The authors did not find pyriform aperture width to be a statistically significant factor in determining whether patients were managed medically or surgically, however they noted that all of the patients in their series with pyriform aperture width <5 mm ultimately underwent surgical repair [6]. Surgical correction is most often performed using a sublabial approach to remove the bony overgrowth causing the stenosis, with placement of post-operative nasal stents to maintain patency of the nasal airway [5].

LARYNGEAL CLEFT

Laryngeal clefts are congenital posterior airway defects in which there is an abnormal connection between the larynx and hypopharynx or oesophagus caused by failure of fusion of the interarytenoid tissues, cricoid cartilage, or trachea-oesophageal septum [11,12]. The Benjamin and Inglis classification system is commonly used to describe laryngeal clefts based on their depth [11]. Type 1 clefts involve the interarytenoid region above the level of the vocal cords. Type II clefts extend partially, but not completely, through the cricoid lamina. Type III clefts involve the entire cricoid cartilage, and may extend to involve the cervical esophagus. Type IV clefts extend into the intrathoracic trachea.

Clinical presentation varies according to the type of cleft as well as any additional cardiopulmonary co-morbidities the patient may have. A patient with a type I cleft may be asymptomatic or have mild feeding difficulties while those with deeper clefts may present with signs of frank aspiration, recurrent pneumonia, chronic cough, stridor after feeding, cyanosis, and respiratory distress [11,13]. Some patients with type I clefts may not obtain a diagnosis until adulthood, while those with III and IV clefts will usually present in the neonatal period with significant respiratory symptoms [14].

Laryngeal clefts can be difficult to diagnose. Given their relative rarity, the patient should also be assessed for other conditions which can present similarly, including gastro-oesophageal reflux disease (GERD), laryngomalacia, neuromuscular dysphagia, and asthma. These conditions may be present in conjunction with laryngeal cleft, and the diagnosis of laryngeal cleft may be suggested if the patient does not improve despite aggressive treatment for these medical problems. A multidisciplinary team of specialists is often involved in the workup and care of these patients, including pulmonologists, gastroenterologists, neurologists, and speech pathologists in addition to the otolaryngologist. Workup can include i) chest x-ray or chest CT, which may shows signs of chronic aspiration or pneumonia; ii) modified barium swallow, which will show aspiration in the vast majority of patients with laryngeal clefts; iii) functional endoscopic evaluation of swallowing, which may also show aspiration; and iv) direct laryngoscopy with direct probing of the interarytenoid area, which is the most reliable method of diagnosis [13]. Among patients who do undergo direct laryngoscopy for recurrent respiratory symptoms, the incidence of laryngeal cleft has been reported to range from 0.2 to 7.6% [13].

Associated anomalies have been described in over half of patients with laryngeal clefts [13]. Most commonly these include