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Unilateral choanal atresia in child with recurrent infections

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To the Editor

Unilateral choanal atresia is the obliteration of the posterior nasal aperture and commonly presents later in life with mild symptoms. We report a case of unilateral choanal atresia in which the patient was initially suspected to have an immunodeficiency due to recurrent rhinosinusitis and respiratory failure during a viral pneumonia in late childhood.

A 10-year-old female with a history of recurrent rhinosinusitis presented to the emergency department with one week of barking cough, fatigue and low-grade fevers. Her past history included trachea-esophageal fistula repair as an infant and speech delay. She had a history of rhinosinusitis 2–4 times per year since age 3 years, diagnosed clinically and treated with antibiotics. She had few episodes of acute otitis media; no prior pneumonias, asthma, eczema, autoimmune disorders or infections. Birth history was significant for 34.5 weeks gestation. No history of carbimazole use in pregnancy, which has been reported to cause choanal and esophageal atresias in exposed infant.

On the admission physical exam, she had diffuse wheezing and decreased aeration in the bases of the lungs. Due to increasing need for respiratory support, she was intubated and ventilated with high-frequency oscillatory ventilator. Her chest x-ray progressed to show consolidation in the left lower lobe. A flexible bronchoscopy showed absent apical segment of right upper lobe (RUL) and thick mucous plugs from left lower lobe distal airways. Bronchoalveolar lavage revealed acute inflammatory exudate with 4% macrophages, 94% neutrophils, 2% lymphocytes and human metapneumovirus (hMPV) as measured by PCR. She was slowly weaned from the ventilator to room air. Allergy/Immunology was consulted for concerns for an immunodeficiency. Her immunology work-up, including serum

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immunoglobulins, tetanus and diphtheria titers, and complete blood count with differential were normal.

Two weeks later in Allergy clinic, the patient's respiratory concerns were resolved. The mother reported persistent daily green and clear rhinorrhea, worse on the left since infancy, which was currently exacerbated. On exam, the external auditory canals were both stenotic, the right turbinate appeared pale and boggy, and the left turbinate was erythematous with thick purulent rhinorrhea obstructing full view. Her serum specific IgE to *alternaria alternata*, *helminthosporium halodes*, maple, birch, white ash, white oak, timothy grass, English plantain, cat, dog, American cockroach, and dust mite were negative. Pulmonary function tests were normal. A sinus CT showed left bony stenosis with membranous choanal atresia and subtotal maxillary and ethmoid opacification consistent with acute rhinosinusitis (Figure 1). Rhinoscopy showed a mixed bony and membranous atretic plate on the left. This was repaired via transnasal endoscopy and a nasal trumpet stent was left in place. She required a second repair due to recurrence of the left atretic plate and a third repair revision with bilateral inferior turbinate reduction.

Discussion

Choanal atresia is a relatively uncommon condition with unilateral atresia occurring in 1:8000 births and bilateral atresia occurring half as frequently.¹ Unilateral choanal atresia often presents later in life with mild unilateral chronic rhinorrhea to rarely feeding difficulties or airway symptoms², whereas bilateral choanal atresia presents at birth with cyanosis and airway obstruction because neonates are obligate nose breathers.³ The atretic plate is often mixed membranous and bony (70–90%) and less often pure bony (10–30%).² A mixed defect was found in our patient which required three surgical repairs; there is no definitive evidence to support any specific surgical technique. The mode of development of the atretic plate is unclear; it has been hypothesized that choanal atresia is caused by the persistence of the buccopharyngeal membrane, the medial outgrowth of the processes of the palatine bone, or a defect in neural crest migration.¹

Choanal atresia can be an isolated anomaly or associated with a syndrome (e.g. CHARGE, VATER, Treacher-Collins, Pfeiffer, DiGeorge). Approximately 20% of healthy infants have one minor anomaly (ex: absent apical RUL segment) and these infants have a 3% risk of associated major malformation (ex: trachea-esophageal fistula, choanal atresia); this risk increases with multiple minor anomalies.⁴ Choanal atresia is most frequently (25% of cases) associated with CHARGE syndrome, which is a constellation of **C**oloboma, **H**ear defects, choanal **A**tresia, growth/mental **R**etardation, **G**enito-urinary anomalies, and **E**ar anomalies.¹ Other symptoms include trachea-esophageal fistula, facial dysmorphology, and palatal defects.¹ Considering this patient's history, she underwent genetic testing for chromodomain helicase DNA-binding protein 7 (*CD7*) gene and was negative for the mutation associated with CHARGE syndrome. Genetics consultation excluded Treacher-Collins and Pfeiffer syndrome given the lack of characteristic craniofacial features. VATER is diagnosed by presence of three of the following malformations: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities; our patient did not meet this clinical diagnosis.⁵ DiGeorge syndrome can present with choanal atresia and

recurrent infections; however, patients have cardiac anomalies, hypoparathyroidism causing hypocalcemia, and/or T cell dysfunction due to thymic hypoplasia. The suspicion for this diagnosis was low in our patient and was ruled out clinically.

This case illustrates that because her chronic unilateral rhinorrhea was missed thus her recurrent rhinosinusitis and hMPV severe pneumonia prompted the investigation for a possible immunodeficiency. Choanal atresia predisposes patients to rhinosinusitis due to the mechanical obstruction preventing sinus drainage despite the maxillary sinus size being normal.^{2,6,7} Cases of unilateral choanal atresia presenting with chronic sinusitis have been reported, but more often presentations involve unilateral chronic rhinorrhea.^{2,7}

hMPV causes symptoms similar to hRSV (wheezing, fever, cough, rhinitis)⁸. However, hMPV infection is more severe, causing pneumonia and even respiratory failure in immunocompromised children. Therefore, the suspicion of immunodeficiency was higher in this case with recurrent rhinosinusitis and hMPV induced respiratory failure. Primary immunodeficiencies are rare diseases with overall prevalence of 1:10,000, thus the differential diagnosis for recurrent rhinosinusitis should include more common causes such as allergic rhinitis, vasomotor rhinitis, nasal polyposis, and cystic fibrosis. Other anatomic defects, such as Haller's cells, paradoxical curvature of the middle turbinate, and concha bullosa deformity have been reported to increase incidence of sinusitis; however, there is conflicting data.⁹ Baseline sinus CT Scan is highly recommended by experts to evaluate for anatomic abnormalities in patients with chronic and recurrent sinusitis.⁹ Clinicians should have an index of suspicion for unilateral choanal atresia in children with recurrent rhinosinusitis and chronic rhinorrhea.

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Clinical Implications

Anatomical defects, such as unilateral choanal atresia, should be considered in children with recurrent rhinosinusitis. Children with recurrent sinopulmonary infections should be evaluated for primary immunodeficiencies and allergic diseases; however, other causes such as anatomic defects should not be overlooked.

A **B** **Figure 1.**

Computed tomography scan of the face of a ten year old female with history of recurrent sinusitis. **A**, Axial view showing left choanal atresia (arrow) which is made up of a mixed bony and membranous atretic plate. **B**, Coronal view showing subtotal sinus opacification of maxillary sinuses (arrow) and right ethmoid opacification.