Case Report

Late Presentation of Bilateral Choanal Atresia in a 10-Year-Old Child with Moyamoya Disease

Ashley C. Mays, Kristin K. Marcum, and Daniel J. Kirse

Department of Otolaryngology, Wake Forest School of Medicine, Winston-Salem, NC 27157, USA
Address correspondence to Ashley C. Mays, abiddy@wakehealth.edu

Received 7 March 2013; Accepted 19 April 2013

Copyright © 2013 Ashley C. Mays et al. This is an open access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

Abstract

Bilateral choanal atresia is a rare congenital abnormality that almost always is diagnosed shortly after birth and requires prompt intervention to limit respiratory distress and facilitate proper growth. We present a case of a child diagnosed at age 10, despite years of upper airway symptoms, who had a concurrent diagnosis of Moyamoya disease.

Keywords

bilateral choanal atresia; Moyamoya; nasal obstruction

1. Introduction

Congenital choanal atresia is an uncommon anomaly defined by obstruction of the posterior nasal aperture. When the congenital atresia is unilateral, infants rarely experience severe respiratory distress and may go undiagnosed until rhinorrhea and nasal obstruction precipitate a visit to the otolaryngologist. Bilateral choanal atresia, however, presents with respiratory distress requiring immediate assistance as infants are obligate nasal breathers. As such, the infant with bilateral choanal atresia is identified in the early perinatal period in nearly all cases. We report an unusual case of a 10-year-old female with Moyamoya disease who remained undiagnosed until the age of 10 years, with bilateral choanal atresia despite severe symptoms for years. Although she had extensive medical care early in life due to Moyamoya disease, her bilateral choanal atresia was not diagnosed until nasal endoscopy in the operating room. The choanal atresia was repaired endoscopically, and she is followed closely in the pediatric otolaryngology clinic.

2. Case presentation

SP is currently a 13-year-old twin female with a history of Moyamoya disease and bilateral choanal atresia. She was diagnosed with Moyamoya disease at birth and underwent external carotid-internal carotid artery bypass at 6 weeks of age at another institution. She required follow-up early for migraines that ultimately abated with age. Her family history is significant for a twin sister who does not suffer from Moyamoya disease and two older siblings with symptomatic Moyamoya disease. SP was originally referred to our otolaryngology clinic at age 9 due to allergy symptoms, hearing loss, nasal obstruction, and rhinorrhea. Childhood history obtained from the mother noted that at birth the child had difficulty breast feeding and had difficulty breathing through her nose during feeds and sleep. Multiple trips to the pediatrician and other specialists repeatedly reassured the mother that her symptoms were most likely due to her history of Moyamoya disease. At her first otolaryngology visit, her examination was remarkable for bilateral mucoid middle ear effusions and copious clear mucus in bilateral nasal cavities. She was started on nasal saline irrigation and a nasal steroid spray for her rhinorrhea, and was scheduled for a return visit in one month to examine for clinical resolution. The child was lost to follow-up for one year and upon return visit continued to have rhinorrhea, bilateral effusions, and flat tympanograms. Therefore, the plan was made to take the child to the operating room where bilateral tympanotomy tubes were placed. Rigid nasal endoscopy was performed intraoperatively to assess the size of the adenoid pad. To our surprise, the patient was found to have bilateral choanal atresia. A CT scan confirmed the diagnosis of bilateral membranous atresia (Figure 1).

Following an in-depth discussion with the mother about choanal atresia and theories for missing the diagnosis thus far, the child was taken to the operating room for endoscopic repair. Examination of the nasopharyngeal side of the choanal atresia looked very atypical in that there was a moderate amount of scarring of the nasopharyngeal surface of the soft palate to the lateral nasopharyngeal wall. The adenoid bed appeared as a thick scar, and no identifiable openings to either eustachian tube or torus tubarius were identified (Figures 2 and 3). Endoscopic repair of the choanal atresia was performed in standard fashion with take-down of the vomer and atretic plate. Topical Mitomycin C was applied, and stents were not placed.
Figure 1: Soft tissue occlusion of the bilateral posterior choana can be viewed in this axial CT image, confirming bilateral membranous choanal atresia.

SP is now 4 years status post repair of her choanal atresia. Her clinical course has been unusual in that the area of her posterior choana has remained widely patent. However, in the area of her eustachian tubes, she has developed circumferential nasopharyngeal stenosis which has required debridement and balloon dilation to maintain patency. She continues to require ventilation tube placement secondary to persistent eustachian tube dysfunction. She has had nearly constant mucoid drainage from her left middle ear space requiring frequent aural toilet and medical therapy.

3. Discussion
Congenital choanal atresia was first reported by Roderer in 1755 and was anatomically defined by Otto in 1830 in a child with choanal atresia associated with deformed palatal bones [6,7]. Proposed etiologies of the congenital malformation include persistence of the buccopharyngeal membrane from the foregut, nasobuccal membrane of Hochstetter, abnormal persistence or location of mesoderm forming adhesions in the nasochoanal region or misdirection of neural crest cell migration [5,6,9].

In most reported series, choanal atresia has an incidence of 1 in 5,000 to 1 in 8000 births, with a slight female predominance. Bilateral congenital choanal atresia presents in the newborn period with life-threatening respiratory distress and cyanosis relieved by crying [2,8]. Rarely an infant may compensate by breathing rapidly through the mouth, and the diagnosis may escape detection for months or years [2,8]. Temporizing measures such as the use of an oral airway, McGovern nipple, or intubation is required prior to definitive surgical treatment [9]. Unilateral atresia presents with unilateral rhinorhea and airway symptoms might be minimal, which might include stertor resulting in some mild feeding difficulties or interrupted sleep if the patent nasal airway is compromised by a deviated nasal septum or other inflammatory condition. Unilateral atresia is more common than bilateral atresia, and 10–50% of children with choanal have associated anomalies, most common

Figure 2: Multiple membranous bands in the area of the adenoid bed and lateral wall of the nasopharynx can be viewed in this endoscopic view from the oropharynx into the nasopharynx using the 70-degree rigid telescope. The soft palate is labeled with an asterisk.

Figure 3: Complete obstruction of the posterior choanae is seen in this endoscopic view of the right posterior nasal cavity using a 0-degree rigid telescope. There is an asterisk on the nasal septum and an arrow on the posterior end of middle turbinate.
being the CHARGE association [2,3,6]. Anatomically, choanal atresia may be classified as membranous, bony, or mixed. CT definitively diagnoses choanal atresia, identifying bony abnormalities, including narrowing of posterior nasal cavity with medial bowing and thickening of the lateral wall of the nasal cavity with impingement at the level of the anterior aspect of the pterygoid plates and enlargement of the posterior portion of the vomer, with or without a central membranous connection.

If the child is not in respiratory distress, the original treatment for unilateral choanal atresia involves supportive treatment with special attention to proper feeding and weight gain for approximately 1 year to allow for some growth of the child and a less technically challenging endoscopic repair [6,9]. However, in cases of bilateral choanal atresia, a surgical plan must be entertained in the neonatal period, which might include endoscopic repair or tracheotomy tube placement. This choice is dependent on the experience of the surgeon, size of the infant, associated medical or anatomic factors, and preference of the family. Patients with CHARGE association, however, present a special challenge. At our institution, these patients are more likely to be managed with a tracheotomy tube as an initial surgical intervention since they tend to have poor muscle tone, reflux, and dysphagia, which more often leads to poorer surgical outcomes with early endoscopic repair.

Moyamoya disease is a cerebrovascular disorder characterized by an idiopathic, nonatherosclerotic narrowing or occlusion of major intracranial blood vessels. Chronic ischemia leads to the development of extensive collateral vessels, resulting in the characteristic Moyamoya (literally Japanese for “puff of smoke”) appearance at the base of the brain. Moyamoya has a peak presentation in childhood and is usually a consequence of the primary disease process, and current treatments are aimed at improving blood flow to prevent strokes either medically or more commonly by surgical intervention.

4. Conclusion

We are not the first to report a child with bilateral choanal atresia presenting at a later age. Candan reported a 16-year-old girl initially diagnosed with sinusitis, and Voelgels and Llorens report on a 13-year-old female and 17-year-old patient who both even underwent an adenoidectomy before diagnosis [2,11]. Although choanal atresia is often associated with other congenital anomalies, a connection with Moyamoya could not be identified in the literature. Although we would not suggest that bilateral choanal atresia should be high on the differential diagnosis of a 10-year-old child with rhinorrhea, it is interesting to note that the etiology of the nasal obstruction was delayed for many years. It is possible that the child’s history of Moyamoya, a disease not often seen in a general practice, could have misled or clouded the symptoms in this child’s perinatal period and thereafter. Nevertheless, it is a reminder that in a child with a complicated medical history, significant difficulty with feeding should continue to prompt the search for the most common causes.

References