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LITERATURE REVIEW

Choanal atresia: comprehensive review of diagnosis and management

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ABSTRACT

Choanal atresia is a congenital condition characterized by the obstruction of the posterior nasal apertures. It can present as unilateral or bilateral and is associated with significant morbidity, particularly in neonates. This review provides a comprehensive overview of the diagnosis and management of choanal atresia. We discuss the epidemiology, etiology, and pathogenesis, emphasizing the developmental and genetic factors involved. Clinical presentation varies with age, and diagnosis typically involves a combination of clinical examination and imaging techniques. Management strategies have evolved, with a focus on surgical intervention. Various surgical techniques, including transnasal endoscopic and transpalatal approaches, are explored. Postoperative care and long-term outcomes are critical components of patient management. We also address current controversies in treatment and future directions for research. Understanding choanal atresia's complexities is essential for improving patient outcomes and advancing therapeutic options.

KEYWORDS: choanal atresia, posterior nasal aperture, diagnosis, endoscopic surgery.

INTRODUCTION

Choanal atresia is a congenital malformation characterized by the blockage of the nasal passages due to abnormal bony (70%) or membranous tissue (30%). Very rarely, the blockage is exclusively membranous[1]. This condition, first described in 1755 by Johann George Roederer, is of significant clinical importance due to its potential impact on respiratory function, particularly in neonates who are obligate nasal breathers. It is a well-recognized etiology for congenital nasal airway abnormalities, that could have various clinical presentations ranging from acute airway obstruction to chronic recurrent sinusitis[2]. It may occur as an isolated anomaly or as part of a syndrome, most notably CHARGE syndrome (Coloboma, Heart defects, Choanal Atresia, Growth restriction and developmental delay, Genitourinary abnormalities, Ear abnormalities)[3].

The pathogenesis of choanal atresia involves the failure of the buccopharyngeal membrane to regress during embryonic development, leading to a spectrum of clinical manifestations depending on whether the atresia is unilateral or bilateral. Bilateral choanal atresia is a neonatal emergency due to the risk of airway obstruction, while unilateral atresia often presents later in childhood with symptoms such as unilateral nasal obstruction and discharge. Usually, 65-75% of cases show unilateral atresia, whereas bilateral atresia is less common[4]. Accurate diagnosis and effective management of choanal atresia are critical to preventing complications and ensuring optimal patient outcomes. Advances in imaging techniques and surgical methods have significantly enhanced the prognosis for affected individuals.

This review aims to provide a comprehensive overview of choanal atresia, including its epidemiology, etiology, clinical presentation, diagnostic strategies, and manage-

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ment options, with a focus on recent advancements and future directions in the field.

EPIDEMIOLOGY

Choanal atresia occurs in approximately 1 in 5,000 to 8,000 live births, making it a relatively rare congenital condition[3]. There is a noted predominance in females, with a female-to-male ratio of about 2:1. The condition can present as either unilateral or bilateral, with unilateral cases being more common. Bilateral choanal atresia is an urgent condition, often identified immediately after birth due to severe respiratory distress[5].

While choanal atresia can occur in isolation, it is often associated with syndromic conditions in approximately 50% of cases, most notably CHARGE syndrome, which includes coloboma, heart defects, atresia choanae, growth retardation, genital abnormalities, and ear abnormalities. Other syndromes linked with choanal atresia include Treacher Collins syndrome and Crouzon syndrome. The variability in presentation and association with other anomalies highlights the importance of a thorough clinical and genetic evaluation in affected patients[6,7].

ETIOLOGY AND PATHOGENESIS

The development of choanal atresia is rooted in embryologic malformations. Development of the nasal cavity starts with neural crest cells migration from their origin in the dorsal neural folds at about 3.5 weeks of fetal life. During the subsequent 2 weeks, nasal processes or placodes invaginate to form the nasal pits. The nasal pits burrow deeper into the mesenchyme, while the nasobuccal membrane normally ruptures to create the nasal cavity and the primitive choana. During normal development, the buccopharyngeal membrane, which separates the nasal cavity from the oral cavity, should regress by the sixth week of gestation. One theory regarding the development of choanal atresia is that, this membrane fails to dissolve properly, resulting in a bony, membranous, or mixed obstruction at the posterior nasal aperture[8].

Genetic factors play a significant role in the etiology of choanal atresia. Mutations in the CHD7 gene, which is associated with CHARGE syndrome, have been identified in almost all cases. While the exact mechanisms by which these genetic mutations lead to the anatomical defect are not fully understood, it is clear that choanal atresia is a multifactorial condition influenced by both genetic and environmental factors[9].

Pathologically, choanal atresia can be classified into three types based on the nature of the obstruction: bony, membranous, or mixed. More than half of the patients have a mixed bony-membranous atresia. Understanding these variations is crucial for planning appropriate surgical interventions[10].

CLINICAL PRESENTATION

The clinical presentation of choanal atresia varies significantly depending on whether the condition is unilateral or bilateral. Bilateral choanal atresia is typically identified at birth due to the newborn's inability to breathe through the nose, which leads to cyanosis, difficulty breathing, and feeding problems. These symptoms improve when the infant cries, as crying allows air to enter the lungs through the mouth[10]. Unilateral choanal atresia may go undiagnosed for several years, but it is usually discovered between 5 months and 2 years of age. It often presents with chronic unilateral nasal discharge, nasal obstruction, and recurrent sinus infections. The diagnosis is frequently delayed because the symptoms can be subtle and gradually progressive. In addition to respiratory symptoms, children with choanal atresia may experience feeding difficulties, failure to thrive, and sleep disturbances due to obstructed breathing. The impact on quality of life can be significant, necessitating timely and effective management to prevent long-term complications[11].

In a multicentre case series involving six tertiary care pediatric hospitals across Canada, Paradis et al.[12] found that the most common presenting symptom was rhinorrhea, with the majority of unilateral cases occurring on the right side. Other symptoms included respiratory distress and feeding difficulties.

DIAGNOSTIC WORKUP

Diagnosing choanal atresia involves a combination of clinical examination and imaging studies. Initial suspicion often arises from physical examination findings and the presence of characteristic symptoms. In neonates with suspected bilateral choanal atresia, a simple test involves attempting to pass a small catheter through each nostril into the nasopharynx. Failure to pass the catheter indicates obstruction. The distance of encountered resistance can provide insights into the etiology of nasal obstruction. An obstruction approximately 1–2 cm from the alar rim in neonates most likely suggests deflection of the nasal septum or inferior turbinate, while 3–3.5 cm from the alar rim indicates obstruction at the level of the posterior choanae[8].

Imaging studies are essential for confirming the diagnosis and planning the surgical intervention. Computed tomography (CT) scans with fine-cut axial, coronal and sagittal images provide detailed views of the nasal and choanal structures, allowing for precise identification of

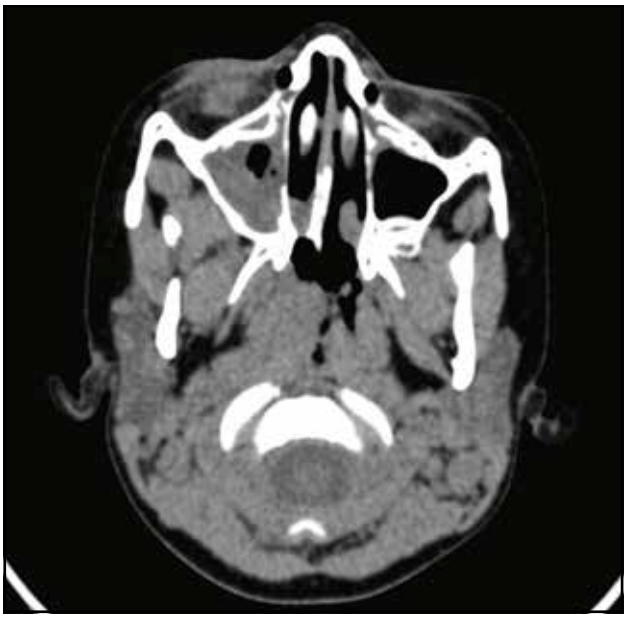

Figure 1. Cranio-facial CT, axial slice – right nasal fossa choanal atresia.

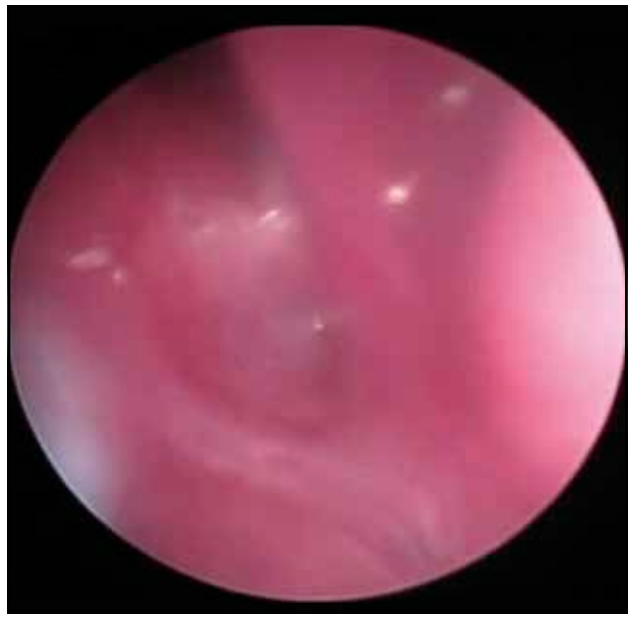

Figure 2. Endoscopic examination of the left nasal fossa choanal atresia.

the location and type of atresia, as well as associated cranio-facial malformations (Figure 1). Magnetic resonance imaging (MRI) is indicated only for selected cases and can be useful in assessing soft tissue structures and associated anomalies.

Endoscopic examination is also valuable, providing direct visualization of the nasal cavity and choanal area (Figure 2). This can be particularly helpful in differentiating between membranous and bony atresia and in planning the surgical approach[1,5].

OUTCOMES AND PROGNOSIS

The prognosis for patients with choanal atresia has improved significantly with advances in surgical techniques and postoperative care. Most patients achieve good functional outcomes with appropriate treatment. However, the prognosis can vary based on the presence of associated anomalies, the type of atresia, and the timing of intervention. Short-term outcomes generally include relief from nasal obstruction and improved breathing and feeding. Long-term outcomes focus on maintaining airway patency and preventing complications.

The recurrence rate of choanal atresia can vary, with some studies reporting rates as high as 20-30%. Factors influencing recurrence include the initial surgical technique used, the presence of associated syndromes, and the meticulousness of postoperative care. In order to avoid restenosis, it is recommended to place nasal stents at the end of the intervention. However, even with this procedure, Achour et al. recorded six cases of recurrence (23.08%) after a mean period of 7.5 months[5].

TREATMENT OF CHOANAL ATRESIA

Management of choanal atresia depends on the severity of the symptoms and the type of atresia. Bilateral choanal atresia in neonates is a medical emergency requiring immediate intervention to secure the airway. Initial management may include the use of oral airway devices or the placement of an oropharyngeal airway to ensure adequate ventilation until definitive surgical repair can be performed[1].

Surgical intervention is the mainstay of treatment for choanal atresia. Several surgical approaches are available, including the transnasal endoscopic, the transpalatal, and the sublabial approaches. The choice of technique depends on the type of atresia, the patient's age, and the surgeon's expertise. Although transpalatal and transseptal approaches have been used in the past, the overwhelming majority of repairs are now performed transnasally using endoscopes to visualize the atresia plate. Endoscopic surgical tools, powered drills/microdebriders, and lasers are used to remove soft tissue and bone[7].

The transnasal endoscopic approach has gained popularity due to its minimally invasive nature and shorter recovery time. It involves using endoscopic instruments to remove the atretic tissue and create a patent airway (Figure 3). The transpalatal approach, while more invasive, provides excellent exposure and is often used for complex cases or when endoscopic techniques are not feasible. However, despite the low incidence of restenosis, the complications associated with this type of approach are significant. These include malocclusion, palate necrosis, oronasal fistula, soft palate muscle dysfunction, and velopharyngeal insufficiency. As a result, this method is not

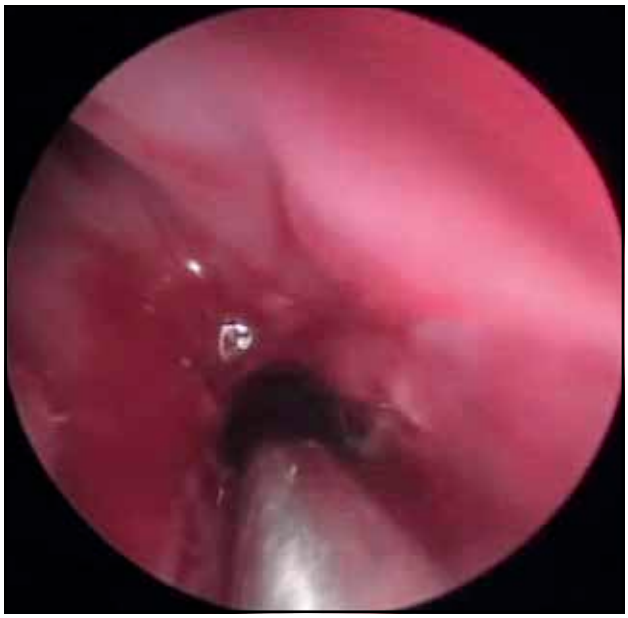

Figure 3. Transnasal endoscopic approach of a choanal atresia.

recommended for children under six[1].

Postoperative care is crucial for ensuring successful outcomes. This includes regular follow-up visits, endoscopic evaluations and, in some cases, the use of stents to prevent restenosis. Complications such as restenosis, infection, and granulation tissue formation can occur, necessitating careful monitoring and management[5,9].

DISCUSSIONS

The management of choanal atresia remains a topic of ongoing debate, particularly regarding the best surgical approach and the use of stents postoperatively. Definitive surgical management can be achieved with either endoscopic transnasal or transpalatal approaches. Some studies advocate for the routine use of stents to maintain airway patency, while others suggest that stents may increase the risk of infection and granulation tissue formation. Comparative studies and long-term follow-up are needed to establish best practices[13].

Future research should focus on identifying the genetic and molecular mechanisms underlying choanal atresia, which could lead to targeted therapies and improved outcomes. Advances in imaging and surgical technology also hold promise for enhancing the precision and efficacy of interventions.

CONCLUSIONS

Choanal atresia is a complex congenital condition with significant implications for the respiratory function, particularly in neonates. Accurate diagnosis and timely surgical intervention are essential for preventing complications and ensuring optimal outcomes. While current management strategies have improved prognosis, ongoing research and advancements in surgical techniques are crucial for further enhancing patient care. Understanding the multifactorial nature of choanal atresia and the associated challenges is key to improving clinical practice and outcomes.

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