

“CHOANAL ATRESIA” EMBRYOLOGICAL BASIS AND ITS CLINICAL SIGNIFICANCE

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ARTICLE INFO**Article history:**

Received: 12 October 2016;

Received in revised form:

13 November 2016;

Accepted: 23 November 2016;

Keywords

Anterior nasal aperture,
Posterior nasal aperture,
Lateral wall of the nose,
Nasal septum,
Choane,
Vestibule.

ABSTRACT

Choanal atresia is a rare congenital condition caused by persistence of the buccopharyngeal membrane during the development of embryo. It is characterized by the closure of posterior nares in the nasal cavity. The condition is well managed with an oropharyngeal airway. A 120 degree nasal endoscope and CT are used in the diagnosis of choanal atresia. The definitive treatment for this upper airway obstruction is surgery. The less invasive approach is the transnasal one which is the first-line approach currently. If this approach is failed transpalatal approach is proceeded which is highly invasive.

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Introduction

Choanal atresia is a malformation of the posterior nasal aperture that interferes with airflow from the nose to the rhino pharynx. Johann George Roederer was the first person to describe choanal atresia in 1755. This term was later renamed in 1854 as an anatomical abnormality of palatine bone by Adolf Otto. Carl Emmert was the first surgeon who has done a successful choanal atresia repair in 1854 [1]. It is a well-recognized etiology for congenital nasal airway abnormality that has various clinical manifestations ranging from acute obstruction to chronic sinusitis [2].

This is a very rare condition occurring in 1 out of 10000 live births. This condition is more common in females the ratio being 2:1 and is frequently unilateral and right sided than bilateral. About 50% of these patients have other congenital deformities associated with this disease [3]. The more typical congenital abnormality related with choanal atresia is CHARGE syndrome. CHARGE stands for coloboma, heart defect, atresia choanae (also known as choanal atresia), retarded growth and development, genital abnormality, and ear abnormality. Following the rupture of the vertical epithelial fold between the olfactory groove and the roof of the primary oral cavity the nasal choancho develops [4]. Failure of this development during the third and seventh weeks of embryonic life leads to choanal atresia [5].

The cause of this anomaly is unknown but thought to be both genetic and environmental. Although in 2008, Barbero et al reported the link between choanal atresia and maternal use of methimazole it is not practically accepted because of lack of evidence [6]. A decrease in maternal retinoic acid is thought to disrupt the neural crest cells. Newborns breathe through the nose because of the elevated position of the larynx during the first few weeks of their life, unlike adults [7]. Manifestation of newborns with choanal atresia varies from acute obstructions to chronic intermittent sinusitis based on choanal atresia types [unilateral, bilateral] or co-existing with other airway

deformities. The diagnosis of choanal atresia is made by physical examination and by monitoring the associated signs [8]. A new technique called acoustic rhinometry is also helpful in diagnosing the anomaly. In this technique, nasal obstruction is evaluated by analyzing the reflections of a sonic pulse introduced via the nostrils [9]. A computed tomography with intranasal contrast is used to confirm the diagnosis. Unilateral choanal atresia is less life threatening to a neonate because the nasal passage is only blocked on one side. This allows a newborn to breathe partially normal but the mucous secretion is noticeable on the affected side of the nose. But, in the case of bilateral choanal atresia, if the airway is not maintained respiratory distress may occur and this will lead to cyanosis and death of the infant [10]. Insertion of an oral airway through an OGT is the immediate medical management in neonates with bilateral choanal atresia. Further management involves surgical repair by means of either transnasal or transpalatal approach of this congenital anomaly [11].

Incidence

The estimated incidence of choanal atresia is approximately 0.0082% which is 1 in 5000-7000 live births. About half of the patients with choanal atresia have bilateral choanal atresia with slightly increased risk among twins [12]. Maternal age and parity does not affect the frequency of this congenital anomaly. Mutation in the Chromodomain-helicase-DNA-binding protein 7 genes is found to be a genetic attributing factor for this condition. A survey of 26 researches with 247 total CHD7-mutation positive CHARGE syndrome cases predicted that 95 [38%] cases presented with choanal atresia [13].

Ontogenesis for the normal development of Posterior nasal aperture or Choane

The development of nasal cavities starts at about fourth weeks of fetal life when the precursor neural crest cells migrate from the embryonic ectoderm [14].

The increase in nasal swellings and penetration of the placodes into the mesenchyme during the subsequent two weeks results in the formation of nasal pits followed by nasal sacs. Simultaneously when the sacs develop the buccopharyngeal or oronasal membrane normally ruptures to create a posterior primitive aperture called "Choane" (Fig-1 & Fig-2) [15].

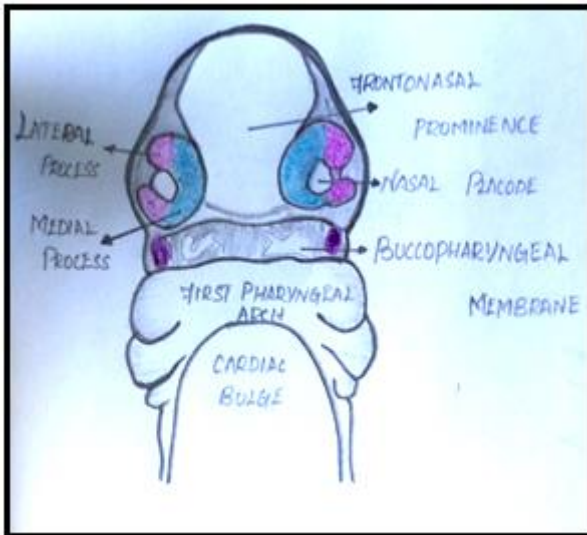


Fig 1. Schematic representation shows the embryological development of choanae.

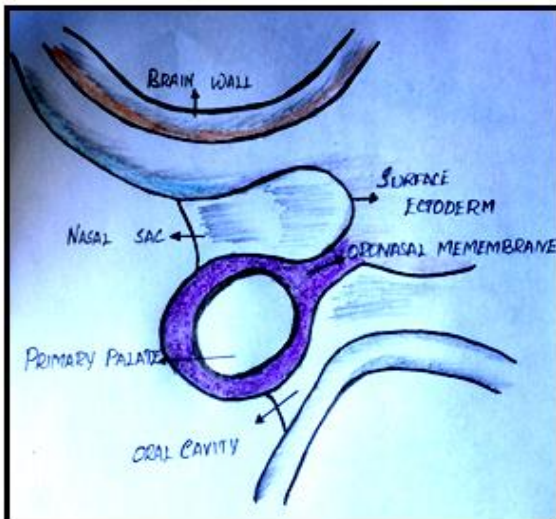


Fig 2. Schematic representation shows the embryological Buccopharyngeal and oropharyngeal membrane ruptures to forms choane.

Ontogenesis for the anomalous Choanal atresia

If this rupture in the buccopharyngeal or oronasal membrane fails to occur, the communication between the nasal cavities and the nasopharynx is lost. Failure of this membrane rupture leads to the development of a congenital anomaly called "Choanal atresia" (Fig-2 & Fig-3) [16, 17]. The following are the four theories that suggest the development of choanal atresia. Out of which, the first and the fourth are the widely accepted ones currently:

- (1) Persistence of the buccopharyngeal membrane from the foregut.
- (2) Persistence of mesodermal adhesions in the nasochoanal region.
- (3) Persistence of the nasobuccal membrane of Hochstetter.
- (4) Diversion of neural crest cell migration [18].

The cause was first suggested as the use of methimazole an antithyroid drug by Barbero et al (2008).

Methimazole crosses the placenta and enters the fetal circulation which in turn reduces the T4 levels of the fetus [19]. Therefore, infants born with choanal atresia have shown a marked decrease in their T4 levels. Also, maternal intake of more than 3 cups of coffee per day increases the risk of choanal atresia by interrupting the membrane rupture[20]. This leads to an outgrowth of palate towards the medial side which blocks the communication between the nasal cavities and the pharynx[21]. The communication between the mouth and the pharynx is patent allowing the infant to breathe only when they cry. Therefore, an infantile colic helps in the diagnosis of choanal atresia [22] (Fig-3).

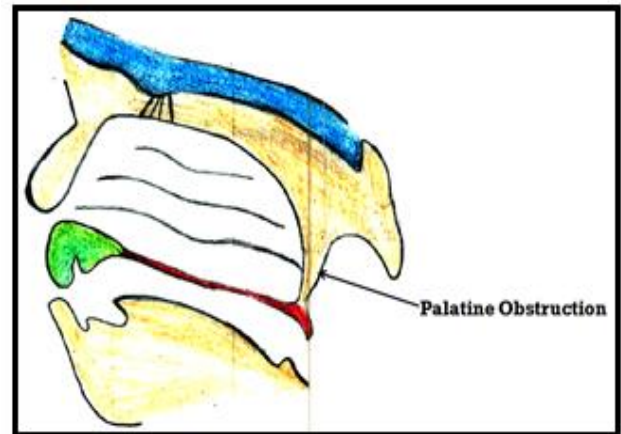


Fig 3. Schematic representation shows the Obstruction of Palatine process causes the choanal atresia.

Discussion

The concern for neonates with choanal atresia is increased due to the fact that neonates are mandatory nose breathers for the first 4-6 weeks of life [23]. Also on looking at the newborn's anatomy, they have a large tongue in communication with most of the palate, a larynx that is placed more superiorly, and a colossal epiglottis as compared to adults(fig3&4). These anatomical modifications exacerbate their obstruction when choanal atresia exists [24].

The manifestations of choanal atresia usually differ based on unilateral or bilateral obstruction. Bilateral choanal atresia is suspected if prenatal polyhydramnios is present [25]. Unilateral choanal atresia presents typically in the later stage of life and is suspected if unilateral nasal discharge and other indications of nasal obstruction are present. Inability to pass a naso-gastric tube or a suction tube through one or both nostrils also indicates nasal obstruction. An endoscope via the oral cavity to visualize the nasal obstruction from below is used in the diagnosis [26]. The presence of bony or membranous obstruction in the area of choana is easily identified on a CT scan [27].

The most commonly associated syndrome of choanal atresia is CHARGE syndrome. Also known as Hall- Hitner syndrome is a rare congenital anomaly with its incidence of 0.1-1.2 in every 10,000 births [28]. The CD-7 gene of chromodomain gene family that codes for protein chromodomain helicase DNA-binding protein is mutated in this syndrome. Choanal atresia develops in 50-60% of neonates of CHARGE syndrome [29]. The 3C triad by Verloes for the diagnosis of CHARGE syndrome includes choanal atresia, colobama, and semicircular canal hypoplasia. The atresia seen in CHARGE is mostly bilateral and bony in texture. Choanal atresia coexisting with cardiac abnormalities results in fatal outcome.

A study by Jongmans et al. in 2005 revealed that 69 of 107 patients with choanal atresia presented with CHARGE syndrome [30].

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It seems to be no difference between right- or left-side unilateral choanal atresia. According to Teissier et al, there is a high prevalence on right sided choanal atresia, as our study showed [32].

The Composition of this congenital anomaly can be differentiated into two: osseous-membranous (two-thirds of cases) or purely osseous. In our regimens, 77.8% of the cases are mixed type[33]. No membranous case was discovered, authenticating the idea that choanal atresia is not only a membrane in the posterior nasal aperture but actually a medialization of the pterygoid processes and the whole lateral nasal wall [34].

Early diagnosis is trouble free in bilateral cases as newborn nasal difficulties lead to show severe clinical indications. However, unilateral cases are often diagnosed later in life, when patients seek medical treatment because of long-standing unilateral nasal obstruction, anosmia, and rhinorrhea [35]. This difference in clinical manifestations can be concluded from the statistically significant difference between the median age at surgery and between the times from diagnosis to surgery, which is lesser in the bilateral group [36].

Conclusion

On encountering a newborn or a toddler with signs of upper airway obstruction one must rule in the possibilities of choanal atresia. If choanal atresia is recognised it is obligatory to check other anomalies of CHARGE syndrome. If bilateral choanal atresia is found stabilizing the patient should be the first line before considering surgical intervention as it is considered an upper airway emergency. The most effective surgical approach is handled to give the patient the best possible outcome.

Reference

[1] Ajit Kumar, E.Ganesh, T.Malarvani, Manish Kr. Singh. Bilateral supernumerary heads of biceps brachii. *Int J Anat Res.* 2014; 2(4):650-52.
 [2] Ajit Kumar, Ganesh Elumalai, Malarvani Thangamani, Nirmala Palayathan, Manish Kr Singh. A Rare Variation in Facial Artery and Its Implications in Facial Surgery: Case Report. *Journal of Surgery.*2014; 2(5): 68-71.
 [3] Ganesh Elumalai, Sushma Chodisetty. Anomalous "Mutilated Common Trunk" Aortic Arch Embryological Basis and its Clinical Significance. *Texila International Journal of Basic Medical Science.* 2016; 1(1): 1-9.
 [4] Ganesh Elumalai, Emad Abdulrahim Ezzeddin. "The sudden soul reaper" - hypertrophic cardiomyopathy – its embryological basis. *Elixir Embryology.* 2016; 99: 43284-43288.
 [5] Ganesh Elumalai, Muziwandile Bayede Mdletshe. "Arteria lusoria"- aberrant right subclavian artery embryological basis and its clinical significance. *Elixir Embryology.* 2016; 99: 43289-43292.

[6] Ganesh Elumalai, Sushma Chodisetty, Pavan Kumar D.2016. Ganesh Elumalai et al Classification of Type - I and Type - II "Branching Patterns of the Left Arch Aorta". *Imperial Journal of Interdisciplinary Research.* 2(9): 161-181.
 [7] Ganesh E, Sushma C. The deer horn aortic arches" embryological basis and surgical implications. *Anatomy Journal of Africa.*2016; 5(2): 746 – 759.
 [8] Ganesh Elumalai, Sushma Chodisetty. Teratological Effects of High Dose Progesterone on Neural Tube Development in Chick Embryos. *Elixir Gynaecology.* 2016; 97: 42085-42089.
 [9] Ganesh Elumalai, Sushma Chodisetty. "The True Silent Killers" - Bovine and Truncus Bicaroticus Aortic Arches its Embryological Basis and Surgical Implications. *Elixir Physio. & Anatomy.* 2016; 97: 42246-42252.
 [10] Ganesh Elumalai, Sushma Chodisetty, Bridget Omo Usen and Rozminabanu Daud Patel. "Patent Ductus Caroticus" - Embryological Basis and its Clinical significance. *Elixir Physio. & Anatomy.* 2016; 98: 42439-42442.
 [11] Ganesh Elumalai, Sushma Chodisetty, Eliza Arineta Oudith and Rozminabanu Daud Patel. Common anomalies origin of left vertebral artery and its embryological basis. *Elixir Embryology.* 2016; 99: 43225-43229.
 [12] Ganesh Elumalai, Sushma Chodisetty, Sanjoy Sanyal. Common Nasal Anomalies and Its Implications on Intubation in Head and Neck Surgeries. *Journal of Surgery.* 2016; 4 (4): 81-84.
 [13] Ganesh Elumalai, Malarvani Thangamani, Sanjoy Sanyal, Palani Kanagarajan. Deficient sacral hiatus cause mechanical low back pain: a radiological study. *Int J Anat Res.* 2016; 4(1):1758-64.
 [14] Mark E Gerber, MD Clinical Associate Professor of Surgery, University of Chicago Pritzker School of Medicine, Laryngeal Stenosis.2016 <http://emedicine.medscape.com/article/867177-overview>.
 [15] John E McClay, MD Associate Professor of Pediatric Otolaryngology, Department of Otolaryngology-Head and Neck Surgery, Children's Hospital of Dallas, University of Texas Southwestern Medical Center. 2016 <http://emedicine.medscape.com/article/995328-overview>.
 [16] Ted L Tewfik, MD Professor of Otolaryngology-Head and Neck Surgery, Professor of Pediatric Surgery, McGill University Faculty of Medicine. <http://emedicine.medscape.com/article/837630>.
 [17] The-myer-cotton-staging-system, 2016 https://www.researchgate.net/figure/12245392_fig1_Figure-1-Figure-The-Myer-Cotton-classification-for-subglottic-stenosis
 [18] Medical embryology book by Ben Panksy, Ph.D., M.D. 2016 <http://discovery.lifemapsc.com/library/review-of-medical-embryology/chapter-58-development-of-the-lower-respiratory-system-larynx-and-trachea>.
 [19] John E McClay, MD Associate Professor of Pediatric Otolaryngology, Department of Otolaryngology-Head and Neck Surgery, Children's Hospital of Dallas, University of Texas Southwestern Medical Center. 2016 <http://emedicine.medscape.com/article/864208-overview>
 [20] John E McClay, MD Associate Professor of Pediatric Otolaryngology, Department of Otolaryngology-Head and Neck Surgery, Children's Hospital of Dallas, University of Texas Southwestern Medical Center. 2016 http://tracheostomy.com/resources/articles/subglottic_stenosis.
 [21] Jonathan B. Ida, MD, MA, J. Lindhe Guarisco, MD, Tulane University School of Medicine, Department of Otolaryngology-Head and Neck Surgery, New Orleans, LA

- [22] Kimsey H. Rodriguez, MD, and Ronald G. Amedee, MD, FACS, Ochsner Clinic Foundation, Department of Otolaryngology-Head and Neck Surgery, New Orleans, LA
- [23] Bainton CR. Complications of managing the airway. In: Benumof JL, ed. *Airway Management: Principles and Practice*. St Louis: Mosby, 1995; 888.
- [24] Scamman FL, Babin R. W. An unusual complication of nasotracheal intubation. *Anesthesiology* 1983; 59: 352-3.
- [25] Wilkinson JA, Mathis RD, Dire Turbinate destruction: a rare complication of nasotracheal intubation. *J Emerg Med* 1986; 4: 209.
- [26] Cooper R. Bloodless turbinectomy following blind nasal intubation. *Anesthesiology* 1989; 71: 469.
- [27] Kuo MJ, Reid AP, Smith JE. Unilateral nasal obstruction: an unusual presentation of a complication of nasotracheal intubation. *J Laryngol Otol* 1994; 108: 991-2.
- [28] Bandy DP, Theberge DM, Richardson DD. Obstruction of nasotracheal tube by inferior turbinate. *Anesth Prog* 1991; 38:27-8.
- [29] Smith JE, Reid AP. Asymptomatic intranasal abnormalities influencing the choice of nostril for nasotracheal intubation. *B J Anaesth*. 1999; 83: 882-886.
- [30] Ahmed Nusrath A, Tong JL, Smith JE. Pathways through the nose for nasal intubation: a comparison of three endotracheal tubes. *B J Anaesth*. 2007; 100: 269-274.
- [31] Prakash S, Rapsang AG, Mahajan S, Bhattacharjee S, Sing R, Gogia AR. Comparative Evaluation of the Sniffing Position with Simple Head Extension for Laryngoscopic View and Intubation Difficulty in Adults Undergoing Elective Surgery. *Anesthesiol Res Pract*. 2011.
- [32] Samsoon GL, Young JR. Difficult tracheal intubation: A retrospective study. *Anaesthesia*. 1987;42: 487-90.
- [33] Wilson ME, Spiegelhalter D, Robertson JA, Lesser P. Predicting difficult intubation. *Br J Anaesth*. 1988;61:211-6.
- [34] Ramadhani SA, Mohamed LA, Rocke DA, Gouws E. Sternomental distance as the sole predictor of difficult laryngoscopy in obstetric anaesthesia. *Br J Anaesth*. 1996;77:312-6.
- [35] Tintinalli, Judith E., Claffey, James. Complications of nasotracheal intubation. *Annals of Emergency Medicine*, 1981; 10 (3): 142-144.
- [36] Watt S, Pickhardt D, Lerman J, Armstrong J, Creighton PR, Feldman L. Telescoping tracheal tubes into catheters minimizes epistaxis during nasotracheal intubation in children. *Anesthesiology*. 2007; 106 (2): 238-42.
- [37] Prior S, Heaton J, Jatana KR, Rashid RG. Parker flex-tip and standard-tip endotracheal tubes: A comparison during nasotracheal intubation. *Anesthesia Progress*. 2010; 57 (1):18-24.