Choanal Atresia—A Cryptic Congenital Anomaly

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Introduction

Choanal atresia is a cryptic congenital anomaly that may result in significant physiological and anatomical alterations of the dentofacial complex. It is a congenital obstruction of one or both posterior nares. The obstruction may be due to varying amounts of bone or soft tissue. This condition which is of major concern to the Pediatrician and Otolaryngologist has received little attention in the cranio-facial literature.

ANATOMY. The posterior choanae connect the nasal chamber anteriorly with the nasopharynx posteriorly. Each is a four sided opening; its height being greater than its width. The borders of each choana are as follows: medially, the vomer; inferiorly, the horizontal process of the palatine bone; laterally, the medial pterygoid plate of the sphenoid; and superiorly, the ala of the vomer articulating with the vaginal process of the pterygoid plate (*1*).

EMBRYOLOGY. At about five weeks in utero, specialized ectodermal thickenings appear on either side of the frontal prominence. This specialized ectoderm, the nasal placodes, becomes surrounded by horseshoe shaped elevations. These elevations, called the medial and lateral nasal processes grow forward leaving the placodes to line the depressions the processes have created. These depressions are the beginning of nasal cavities and are called the nasal pits or fossae (Figure 1A). As the face continues to grow forward, the floor of the nasal fossae also becomes the roof of the oral cavity. The most anterior part of this separation grows and becomes the primitive palate. The tissue behind the primitive palate thins out to form the nasobuccal membrane; the mesenchyme between the oral and nasal epithelium disappears leaving the epithelial membrane. This membrane degenerates during the seventh week in utero establishing a communication between nasal and oral cavities, the primitive choanae (Figure 1B). The choanae are initially separated from each other in the midline by the frontonasal process and later by the medial processes that fuse in the midline. Deep to the surface, the

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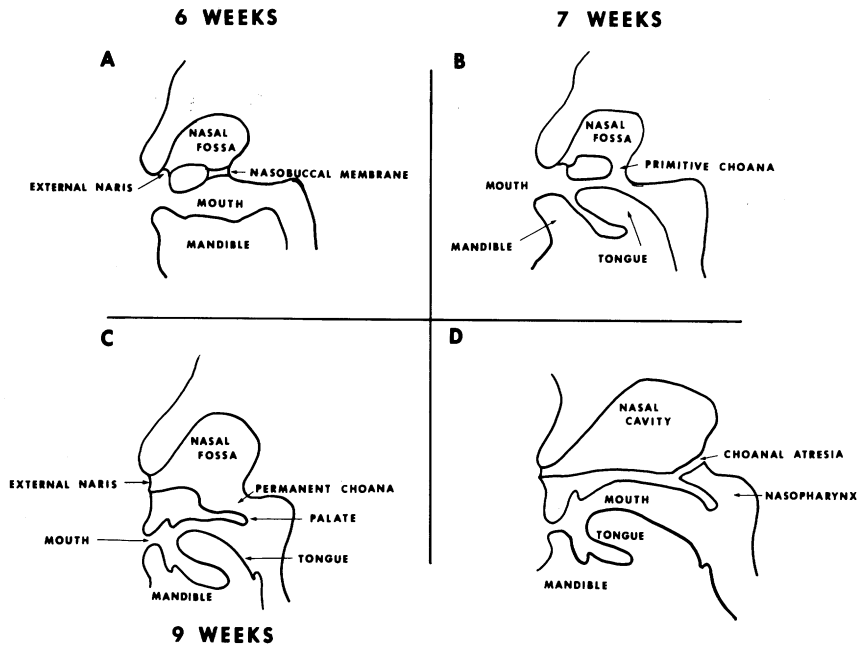

FIGURE 1. This is a schematic diagram of a sagittal section of the developing head. The sections are off the midline through the right nasal fossa. Figures of 6, 7, and 9 weeks are discussed in the text. The diagram to the lower right demonstrates where the atresia will be found after the face has completed its growth in the fetal period. (Drawings adapted from Benton N. Colver, Annals of Otology, Rhinology, and Laryngology, Volume 46, Pages 358–375, 1937).

frontonasal process grows backward in the midline forming the nasal septum. By the end of three months in the utero, the palatal process of the maxilla have fused together and with the nasal septum to increase the dimensions of the nasal cavity at the expense of the oral cavity. Joining of the palatal shelves and the nasal septum creates the permanent choanae in a more posterior position (Figure 1 C) (*2, 3*).

PATHOGENESIS. The etiology of this anomaly is unknown but there are several theories to explain its occurrence.

Failure of the nasobuccal membrane to rupture, with the persistant tissue being carried posteriorly and vertically as the face develops, is considered to be the most likely explanation (Figure 1 D). Since 90% of atresias are bony, the formation of the nasobuccal membrane is thought to be altered before mesoderm has completely disappeared between the oral and nasal epithelium (*4*).

Other causes might be the persistance of the cephalic portion of the buccopharyngeal membrane and overgrowth or deformity of the vomer, palatine bone, body of the sphenoid or medial pterygoid plate (*4*).

CLINICAL FEATURES. Clinical features will depend on whether the atresia is bilateral or unilateral.

Newborn infants are obligate nose breathers (*5*) and may require as long as several months before learning to breath through the mouth. Because of this,

bilateral atresia is manifest immediately after birth and becomes a neonatal emergency. Clinical signs are dyspnea with chest retractions and cyanosis. The dyspnea and cyanosis may be cyclic, relief occurring when the baby cries which permits air exchange through the mouth. It is necessary to establish an artificial oral airway before serious anoxia occurs.

Oral feedings will be difficult because of the inability to coordinate breathing and swallowing.

Exceptions to this pattern exist in babies who do not show respiratory distress at birth but have feeding difficulties. Most of these cases are diagnosed in the neonatal period, but not all. There are case reports of older children and adults who were found to have bilateral choanal atresia (*6*, *7*).

Unilateral atresia may not be detected until some months or years later when the child is seen by a physician for a persistant mucoid nasal discharge from the affected nostril.

DIAGNOSIS. Choanal atresia should be suspected in any infant or child who has respiratory difficulty or is a persistant mouth breather. These signs and symptoms may also exist in abnormally narrow or stenotic choanae severe enough to interfere with function, although not completely occluded. Diagnosis can be made by 1) attempting to pass a small catheter or probe into the nasopharynx (Figure 2); 2) filling the nasal cavity with a radiopaque substance and studying the area radiographically (Figure 3); and 3) filling the nasal cavity with methylene blue and observing for the blue color in the nasopharynx.

ASSOCIATED ABNORMALITIES. Most associated congenital abnormalities are in the craniofacial region or in the cardiovascular system. Where the atresia occurs only on one side, dentofacial abnormalities are not as frequent as in bilateral atresia and oral structures usually develop normally. Bilateral atresia is often accompanied by major oral abnormalities including palatal deformities and

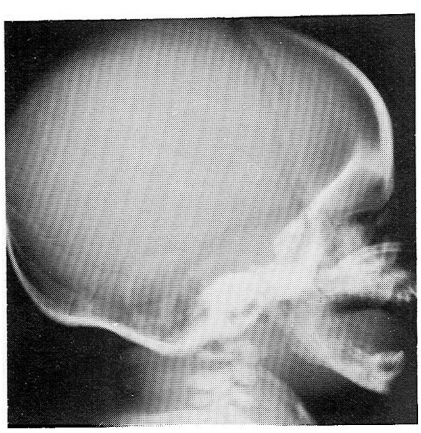

FIGURE 2. A lateral radiograph of a newborn infant demonstrates a catheter inserted to the anterior border of the atresia.

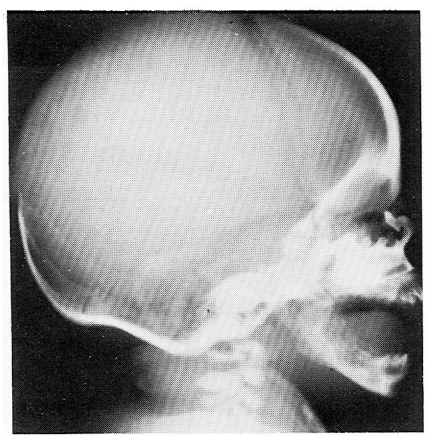

FIGURE 3. The right nasal cavity in this lateral skull film is filled with lipiodol; the study demonstrates the nasal cavity and the location of the atresia. No contrast material is seen in the pharynx.

TABLE 1

Choanal Atresia	bilateral	unilateral
	20	4
Choanal Atresia	female	male
	16	8

TABLE 2

Unilateral Choanal Atresia	Right	Left
	3	1

TABLE 3

type of atresia	*number*
bone	6
membrane	1
1 side bone	
1 side membrane	1
1 side partially of bone	
1 side bone	1
total number reported	9

TABLE 4. Anomalies Associated with Bilateral Choanal Atresia

patients	*anomaly*
1	Craniofacial Dysostosis
2	hypoplasia of the frontal bone
	imperforate anus
	coarctation of the aorta
	tracheo-esophageal fistula
	ureto-pelvic stricture
	hydronephrosis
	patent ductus arteriosus
	esophageal atresia with tracheoesophageal fistula
3	deformed ear-lobes
4	high arched palate
	bifid uvula
	microstomia
	micrognathia
	hemangioma of the superficial scalp overlying the occipital bone
	strawberry hemangioma of the abdomen
	brachydactylia of hands
5	hypotelorism
	micropthalmia and blepharophimosis of the left eye
	anopthalmia of the right eye
	abnormal external nose
	two ridges of bone extending the length of the hard palate in the midline
	microcephaly
6	bilateral colobomas of the optic nerve
	high arched palate
	central nervous system retardation
7	microcephaly
8	craniostenosis (oxycephaly)
	syndactylism of the hands and feet
	high arched palate
	hypoplastic maxilla
9	submucous cleft of the hard palate
10	tricuspid valve atresia
	hypoplastic right ventricle
	ventricular septal defect

clefts, malocclusion, micrognathism, mandibulofacial dysostosis, and craniofacial dysostosis.

In a study done by Evans and Maclachlan (*8*) of 65 patients with choanal atresia, 28 (43%) had an associated congenital anomaly. Of these 28, 19 (68%) had an associated craniofacial abnormality.

REVIEW OF CASE HISTORIES. A review of the records at Children's Hospital of Philadelphia for the past twenty years reveal 24 cases that met the diagnostic criteria for choanal atresia. Twenty were bilateral and four were unilateral (Table 1, 2, and 3).

Diagnosis. All twenty of the patients with bilateral atresia presented with respiratory distress and difficulty feeding in the neonatal period. Three of the unilateral atresias were diagnosed at three to five years of age, the fourth being detected as a neonate because of periodic respiratory distress and difficulty in feeding.

Family history. It is interesting to note that in three cases, there is a family history of relative dying in the neonatal period. The maternal uncle of one infant had died at the age of two days and was diagnosed as having pheumonia. The maternal aunt of an infant with choanal atresia died of asphyxia at two days of age and in another instance, two maternal aunts died in the neonatal period of asphyxia, the causes of which are unknown.

Choanal atresia is recognized as having a familial tendency. Cases have been reported of choanal atresia in two successive generations and in two sisters (*10*). The genetic aspects of choanal atresia are discussed in a paper by Grahne and Kaltiokellio (*11*). There are also reported cases of choanal atresia with a family history of death in the newborn period of asphyxia or unknown causes (*10, 12*). It is possible that some of these could have been due to choanal atresia.

Associated Abnormalities. In the four cases of unilateral atresia, there were no other abnormalities reported. Of the twenty bilateral atresias, ten had an associated congenital anomaly and nine of these were in the craniofacial region (Table 4).

CASE HISTORIES. The following three examples illustrate the variations in patients with this condition.

Case #1. H. C. was a 4½ year old girl referred to the Otolaryngology Department of Children's Hospital of Philadelphia because of a persistant nasal discharge from the right nostril. At the age of three, her tonsils and adenoids were removed but the discharge persisted. The diagnosis of right choanal atresia was made by failure to pass a catheter into the nasopharynx and x-ray contrast studied. A panoramic radiograph (Figure 4), taken at age 5, shows a deviated septum to the atretic side and increased radiopacity of the right nasal fossa. At surgery, the atresia was found to be bony.

Case #2. (Figure 5) L. S., a nine year old female, was first seen at 2½ hours old because of respiratory distress and cyanosis that improved when she cried.

The family history included two maternal aunts who died in the newborn period of asphyxia, the causes of which are unknown.

At two days of age, the bony atresia was removed at surgery. Also noted at surgery was a widened vomer bone. She has subsequently required four

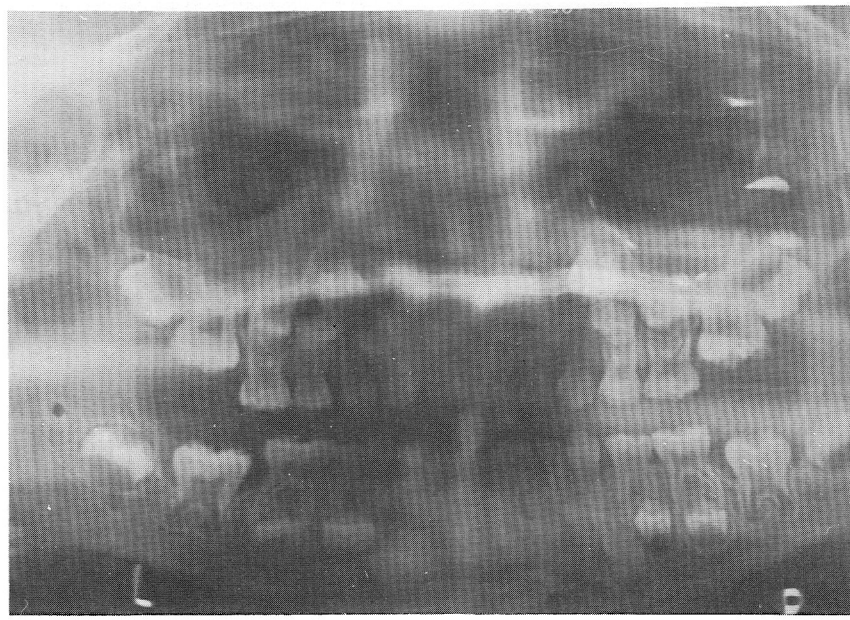

FIGURE 4. Case #1. Unilateral right choanal atresia. Radiograph taken at the age of 5 shows a deviated nasal septum and increased radiopacity in the right nasal fossa.

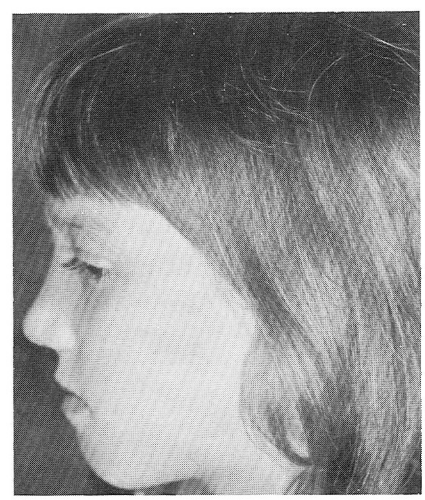

FIGURE 5. Case #2. Bilateral choanal atresia, occeous, age of 9. (photograph courtesy of Dr. Raymond Werther)

operations to maintain patency of the choanae. Her dental development reveals a hypoplastic constricted maxilla in bilateral anterior crossbite. She has been a chronic mouth breather most of her life in spite of repeated attempts to establish a patent nasal airway. She has a high arched palate (Figure 6).

Case #3. R. S. is a five month old white female referred for repair of bilateral bony choanal atresia. She is the only child of a 22 year old female born after 34 weeks gestation and weighed 4 pounds at birth. The pregnancy was

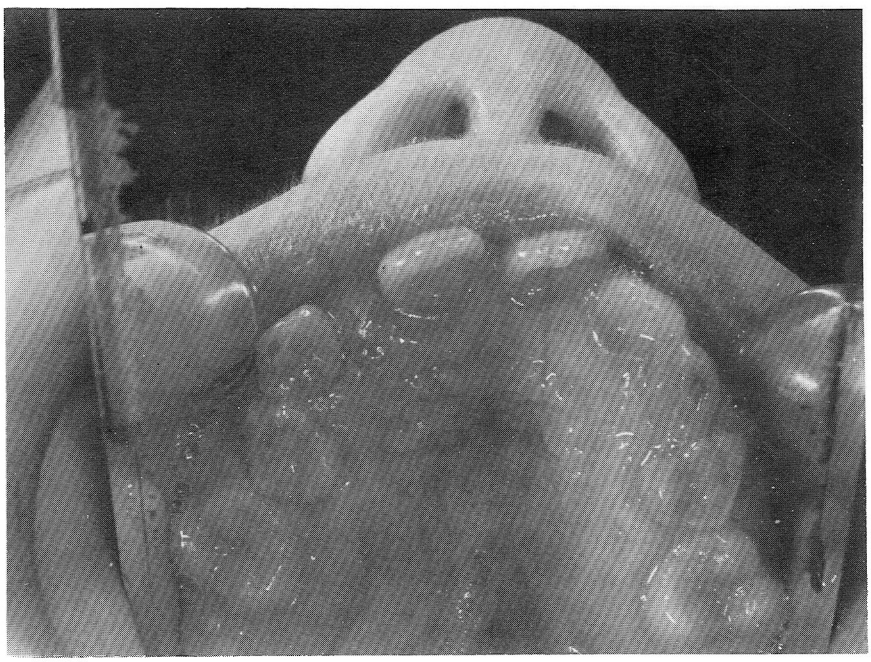

FIGURE 6. Case #2. High arched palate, age of 9. (photograph courtesy of Dr. Raymond Werther)

complicated by vomiting throughout the gestation and by edema of the lower extremities in the third trimester. The edema was treated with diuretics. The mother has a cleft of the hard palate that was repaired by surgery.

At birth, the baby was noted to have marked respiratory distress with retractions and cyanosis. She was intubated and resuscitated with oxygen. The diagnosis of bilateral choanal atresia was made and at 2½ months of age surgery was performed to remove the atresia. At five months, the choanae became occluded and another operation was necessary.

Because of the number of associated abnormalities noted at birth, karyotyping was done and found to be normal. The associated congenital anomalies noted were superficial hemangioma of the scalp over the occipital bone, strawberry hemangioma of the abdomen, hypoplastic maxilla and mandible, microstomia, bifid uvula, high arched palate and brachydactyly.

Conclusion

Choanal atresia is an uncommon but significant congenital anomaly because of its ability to interfere with normal respiratory function and a high incidence of associated congenital abnormalities. It should be suspected in cases of airway obstruction, persistant mouth breathing, and continuous nasal discharge.

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