INTRODUCTION

Neonatal airway obstruction leading to respiratory distress is a fairly common problem in the pediatric ICU setting, and it is important to be able to swiftly delineate the cause of the obstruction to effectively treat it in a timely manner. Bilateral choanal atresia should always be considered in these circumstances because, although it is relatively rare as a cause, it is a highly treatable condition. Even unilateral atresia may cause less severe symptoms and certainly may present later in a child or even adult’s life. If the atresia is associated with other facial and systemic anomalies, one must consider CHARGE and other syndromes during the diagnostic process.

ANATOMY

The word “choana” comes from the Greek word meaning “funnel”. The choana is essentially the posterior nasal aperture by which air flows from the nasal cavity into the nasopharynx. The anatomic borders of the choana are listed below:

- Superior border: inferior surface of the sphenoid body
- Lateral border: lamina of the medial pterygoid plate
- Medial border: the nasal vomer
- Inferior border: horizontal portion of the palatal bone

The following anatomic abnormalities contribute to forming the atretic choana: a narrowed nasal cavity, lateral impingement of the medial pterygoid plate, an abnormally thickened vomer, and a membrane most of mucosa and/or bone extending across the choana. Primary blood supply of the choanal region comes from the sphenopalatine artery (supplies the medial, lateral, and superior
aspects) and the greater palatine artery (primarily feeds the inferior choana though blood supply overlaps the sphenopalatine supply).

It is important to recognize that, in addition to choanal atresia, there are several other potential causes of neonatal nasal obstruction. Fixed anatomic causes may include nasal pyriform aperture stenosis or septal buckling as a result of birth canal trauma. Generally syndromic and non-syndromic children with craniofacial abnormalities of all types may have unfavorable nasal anatomy. Allergy and sinus disease with associated secretions and tissue swelling may contribute as well, especially in children with cystic fibrosis. In addition, nasal masses such as hemangiomas and encephaloceles must be ruled out in any child with nasal obstruction.

EMBRYOLOGY

Between 4-6 weeks of gestation, one can see the formation of primitive nasal structures via neural crest cell migration such as the columella, philtrum, and upper lip. At about the same time, nasal pits form and burrow into the mesenchyme to create the primitive nasal cavities. These nasal pits sit atop the frontal portion of the stomodeum (i.e primitive oral cavity), and the pits are separated from the stomodeum by the nasobuccal membrane. Once the nasobuccal membrane ruptures, the primitive nasal cavity and primitive choana are formed. The primitive choana matures and transforms into the definitive (i.e. secondary) choana. There is also an additional membrane known as the buccopharyngeal membrane which separates the stomodeum (ectoderm) from the primitive pharynx (endoderm).

The following are 4 theories by which choanal atretic plate is thought to exist:

1. An abnormal persistence of the buccopharyngeal membrane (walls of stomodeum from pharynx)
2. A persistence of the nasobuccal membrane (between nasal cavity and stomodeum)
3. Mesoderm forming in the wrong location causing adhesions in the choana
4. Misdirection of neural crest cells to the choanal area

It is important to note that initially theory #2 (persistent nasobuccal membrane) was historically the most accepted, but the majority of scientists believe now that the neural crest theory (#4) is the most plausible. This is in part due to the high correlation with choanal atresia and Treacher Collins Syndrome which is in fact a disorder of aberrant neural crest cell migration.

HISTORY AND EPIDEMIOLOGY

Choanal atresia was first described by a physician named Roederer in 1755. However, the first recorded surgical repair was not until 1854 by Emmert. The technique Emmert used was a blind nasal puncture with a sharp narrow instrument. Blind puncture is not practiced any more due to the unacceptably high rates of serious complications like cerebrospinal fluid leaks, midbrain trauma, and petrous apicitis (similar to Gradenigo syndrome).

In general, unilateral choanal atresia is more common than bilateral (65-75% of patients with unilateral), and females are more likely to have it (2:1 ratio to males). When unilateral, it most
commonly occurs on the right side, and if it is bilateral, there is a high correlation with syndromic illnesses such as CHARGE syndrome, Treacher Collins, and Crouzon’s disease (75% of cases).

Historically teachings of choanal atresia defined it as occurring as bony 90% of the time and membranous only 10% of the time. This was based on a study reported by Fraser in 1910 of 115 patients. More recently, these ratios have been dispelled by Brown et al. (1996) published in Laryngoscope who analyzed data from 63 patients including 47 patient CT scans alone as well as the histology and CT scans of 16 additional patients on whom they operated. They found that 18 patients (29%) had purely bony atretic plates and 45 patients (71%) had a mixed bony-membranous nature. It is crucial to point out that 0 of the patients had a purely membranous-only nature.

SYMPTOMATOLOGY AND DIAGNOSIS

The primary problem and cause for concern in a neonate with choanal atresia (more specifically bilateral) is that neonate are obligate nasal breathers for at least the first 4-6 weeks after birth. Also a neonate’s anatomy includes a relatively large tongue contacting more of the palate, a more superiorly placed larynx, and a large floppy epiglottis. These anatomical entities unique to the newborn contribute their need to be obligate nasal breathing and hence, worsen their obstruction when choanal atresia is present.

Signs and symptoms of choanal atresia usually differ based on a bilateral or unilateral presentation. In bilateral atresia, this often presents as cyanosis in a newborn, creating increased respiratory effort and notable chest retractions. It is worse with feedings, and classically the cyanosis along with oxygen saturation will improve when the child is crying. Bilateral atresia may be diagnosed or suspected on prenatal ultrasound when polyhydramnios is seen. Unilateral choanal atresia typically presents later in life (commonly a few years old) and may be suspected in a child with chronic unilateral, thickened nasal discharge and with other signs of obstruction.

On initial assessment, diagnosis may obtained via several simplistic methods. The physician may note an inability to pass a NG tube (6 French commonly used) or suction catheter through one or both sides of the nose. A mirror or shiny metal object may also be employed to diagnose it by placing it under the nose and seeing if there is fogging or not from the nasal air flow. If suspected, more sophisticated tests such as CT scan and nasal endoscopy should be used. A 120 degree endoscope can be utilized to visualize the atresia from below when inserted into the oral cavity. On CT scan the presence of excess bone and/or membrane in the area of the choana are readily identified.

CHARGE SYNDROME

When discussing choanal atresia, it is prudent to discuss one of its most commonly and classically associated syndromes: CHARGE. Originally termed Hall-Hitner syndrome (1979), the term CHARGE syndrome came to fruition after Pagon et al. identified a non-random association between several anomalies in 1981. CHARGE syndrome is fairly rare with its incidence being somewhere between 0.1 and 1.2 in every 10,000 births. It is thought to occur due to a mutation in the CHD-7 gene which is apart of the chromodomain gene family. This gene codes for the protein chromodomain helicase DNA-binding protein. This gene mutation was identified in 69 of 107 patients in a 2005 study by
Jongmans et al. and similar findings were seen the follow year in 64/110 patients in a 2006 study by Lalani et al. The majority of the mutations are de novo autosomal dominant mutations though there is some association with advanced paternal age. There have been a few case reports of this gene being passed to offspring, but this is unlikely to occur given the significant hormonal and reproductive abnormalities of those affected with CHARGE.

Affecting up to 80% of patients with CHARGE, the “C” stands for the ocular abnormality known as coloboma. A coloboma is essentially a hole in one of the structures of the eye, often given an external appearance of a “cat’s eye”. It can be bilateral or unilateral and most commonly affects the retina but can occur in the iris or both. Vision may normal but most commonly is affected negatively to some degree so these patients should always undergo ophthalmologic testing including visual analysis and electroretinogram.

Heart (“H”) abnormalities are a sensitive but not very specific finding in a CHARGE patient, occurring in 75-80%. 33% of these patient have tetralogy of Fallot but may also have a patent ductus arteriosus (PDA), ventricular septal defects, and atrial septal defects. Diagnosis is with echocardiogram, and treatment is generally cardiothoracic surgery. Yet it is important to administer prostaglandins if the child has a PDA to keep it open until surgery can be performed. Cardiopulmonary issues are commonly to blame in the high rate of mortality in these children perinatally.

Atresia (“A”) of the choana (i.e. choanal atresia) occurs only in approximately 50-60% of kids of CHARGE. The atretic plate is typically bilateral and mainly bony in consistency. Choanal atresia coinciding with cardiac abnormalities creates a highly fatal combination of problems for the newborn.

Children with CHARGE commonly have issues with retardation (“R”) of growth and other developmental problems. Oddly enough, they are usually normal weight and length at birth so these issues do not manifest until later in life. Growth hormone is commonly found to be deficient. Though IQ is variable and occasionally normal, at least 70% of cases show IQ below 70 which qualifies as mental retardation. Behavior problems are common including autism and obsessive compulsive disorder.

Several genitourinary (“G”) abnormalities are found in CHARGE patients. Hypoplasia of the genitals is the most common and easiest to recognize in the male patients. Genital anomalies include microphallus, complete penile agenesis, and hypospadias in males, and for females one often finds atresia of the uterus, cervix or vagina as well hypoplastic labia/clitoris. This commonly extends into the urinary system with bladder, ureter, and kidney pathology such as vesicoureteral reflux.

Lastly, considered to be one of the most specific findings of CHARGE syndrome, a variety of ear (“E”) defects are often seen (80-100% of cases). Externally, the classic finding is a “cup” or “lop” shaped pinna caused by malformed cartilage. In the middle ear, ossicular defects are commonly found including absent stapedius, hypoplastic incus/stapes, or ossicular fixation. Absence of oval or round windows may be present, and many of these children have problems with chronic otitis media, likely related in part to eustachian tube dysfunction from choanal atresia. In the inner ear, one may find a Mondini dysplasia (decreased number of turns of the cochlea), yet one of the most specific findings in CHARGE syndrome as a whole is the absence of semicircular canals. Given these abnormalities, it is
prudent to acquire audiologic testing in these children in the form of audiogram or auditory brainstem response. Since they may already be getting a CT sinus scan for choanal atresia evaluation, one should consider at the same time requesting a CT of the temporal bones to evaluate the ears.

Though the above-listed findings are typical of CHARGE syndrome, this syndrome is not limited to them. Multiple cranial nerve defects are found in CHARGE. Facial nerve palsy with associated facial asymmetry is seen in 50-90% of cases. Approximately 80% of these children have feeding and swallowing issues (likely multi-factorial but IX and X cranial nerve palsies contribute), so it is highly important to involve a feeding team as aspiration and failure to thrive are common in these children. Other features include gastroesophageal reflux disease, tracheoesophageal fistulas, laryngomalacia and vocal paralysis, cleft lip/palate, and arhinencephaly (specifically hypoplastic olfactory bulb).

Previous major and minor diagnostic criteria schemes were established by Pagon in 1981 and then followed up by Blake in 1996. However, the most current criteria were set forth by Verloes et al. in 2005. Verloes focused on 3 major findings as diagnostic for CHARGE including ocular coloboma, choanal atresia, and hypoplasia of semicircular canals (known as the 3C triad). For minor criteria they emphasized the specific findings of rhombencephalic dysfunction (such as cranial nerve palsies) and arhinencephaly with associated anosmia.

**TREATMENT OF CHOANAL ATRESIA**

There are many considerations to make when undergoing treatment of choanal atresia such as the various surgical approaches and whether or not to use stenting or Mitomycin C. However the most crucial initial interventions in a child with bilateral atresia should focus on stabilizing the patient. In achieving this, the goal should be finding a way to oxygenate the patient despite the crippling upper airway obstruction. For this purpose one may use a special nipple called a McGovern nipple which has a large hole in its center for air passage or a plastic oral airway piece. More invasive measures include endotracheal intubation and tracheostomy. In many circumstances tracheostomy may be avoided, but it may be needed if primary surgical repair must be delayed for various reasons such as cardiac issues.

There exists several methods of surgical repair of the atresia including transnasal (including blind puncture, microscopic, and endoscopic approaches), transpalatal, transseptal, and sublabial approaches. Though many of these are of historic significance only, the transnasal and transeptal approaches are the methods commonly used in current practice.

The transnasal approach is by far the most common and successful method used today for repair of choanal atresia. This is primarily accomplished via an endoscopic approach given the safety and convenience of direct visualization with endoscopy with 0 and 120 degree endoscopes. Puncture of the atretic plate may be accomplished using a curved/straight urethral sound, or powered instruments such as a microdebrider or diamond burr drill may be used to open the plate. Below listed are proposed steps of the procedure:

1. Decongest the nose topically (oxymetazoline) and local anesthesia injection with 1% lidocaine with epinephrine into the borders of the atretic plate
2. Expose the palate and nasopharynx (may use mouth gag) and visualize the atresia with a 120 degree endoscope from below
3. Directing it inferomedially, use a spinal needle to puncture through the plate and visualize it from below to confirm proper position
   a. Note: inferomedial placement is crucial to avoid complications such as brain trauma
4. Create laterally based anterior mucosal flaps with sickle knife over plate
5. Mechanically open the atretic plate (drill or microdebrider) and remove the thickened posterior vomer with a drill or backbiter
6. Enlarge the new choana to the size of a 16 French cather/urethral sound
7. +/- stenting and/or Mitomycin C placement

To emphasize again, it is important to direct the dissection in an inferomedial direction on each side of the nasal cavity and also always try to be cognizant of the location of the eustachian tube opening. If injured, permanent eustachian tube dysfunction may ensue.

For the transpalatal approach, first a mouth gag is inserted and then local anesthesia applied into the palate. The greater palatine artery is the main supply for this operative area. Next, typically a U-shaped palatal mucosal flap based on this artery is made (must preserve). Once the mucosal flap elevated, the involved bone creating the atresia and the palatal bone anterior to the vessels is resected using a drill or bone rongeur. Lastly, stents are put in place, and the palatal flap is put back into place which is used to resurface the mucosa.

There exists some notable advantages to the transpalatal approach including the ability to directly visualize the surgical field without need for endoscopy, the creation of a new mucosa lined cavity, and typically a decreased needed duration of stenting. Yet, this approach is often not first-line largely owing to its many complications. The most common and debilitating complication is its tendency to cause a lasting crossbite deformity (52% of cases) as well as generally stunting palatal growth. Since most palatal growth is complete by age 5, it is recommended to wait until after this age to attempt this approach. There is a high probability of flap necrosis is blood supply from the greater palatine arteries is compromised.

RESTENOSIS AND TREATMENT ADJUNCTS

One of the major complications of choanal atresia is restenosis of the atretic plate. Significant debate exists in the literature on whether placing stents in the operative site after the procedure provides improved outcomes. Stents are usually made out some type of plastic tubes. Portex and manipulated endotracheal tubes are often used. As a proponent of stenting, Friedman et al. (2002) reviewed 46 cases of choanal atresia and cited a favorable prognosis (defined as less operations needed) with children who had stents in for greater than 12 weeks. Later in 2004, Gujrathi et al. performed a study of 52 patients who has stents in place for a median of 12 weeks and on 2 of the 52 patients required a revision operation.

Despite these studies, several opponents makes claims against the efficacy of stenting, often citing how there is a significant foreign body tissue reaction with the stent in place leading to more granulation tissue and scarring down of the tissues. Schoem et al (2004) demonstrated this with a study of 13 patients who did not have stents placed, with 4 of the subjects requiring no further operations and 9 of them requiring only one additional operation. Llorente et al. (2013) showed similar outcomes without stents, having 100% patency in their test subjects after 27 months of following them.
In addition to stents, the anti-neoplastic agent Mitomycin C has been used as a treatment adjunct to try to decrease stenosis. Mitomycin works by inhibiting fibroblast growth and proliferation, which may help keep granulation tissue at bay. This drug is already commonly used in other areas of head and neck surgery including during subglottic stenosis treatment and with laryngotracheal resection. It is typically applied as a topical agent on a cotton pledget for a few minutes in the nose to achieve the desired effect. A study by Holland et al (2001) showed promising outcomes with the use of the drug. The study has 8 test subjects and 15 controls. The outcome measured the number of dilations required after the initial surgery and they found that the study group patients required a significantly less number of dilations than the control group without the Mitomycin C application (0.375 dilations in test versus 3.667 dilations in control).

TREATMENT COMPLICATIONS

Some of the major complications have been discussed previously and some that have not need to be addressed. There are several potential complications with the use of stents including foreign body tissue reaction, nasal alar erosion, and columella pressure necrosis. The transpalatal approach complications as shown prior include flap necrosis and crossbite deformities. In general, mucosal flap death is possibly for the transpalatal and transnasal approaches. In either approach, the patient may sustain damage to the tori tubarius and eustachian tube openings given their close proximity to the operative field. Similarly, there is a chance of central nervous system trauma (especially with blind puncture) if the skull base is violated by sharp instruments or too much torque/force on the bone. And lastly, restenosis is one of the most common complications of the surgeries and usually requires additional procedures as well as consideration of treatment adjuncts like stents and Mitomycin.

In regards to patient outcomes, some work as been done by Friedman et al. (2000) in establishing predictive factors for successful repair. Their retrospective study analyzed data from 46 children (28 bilateral and 18 unilateral) with 40% having other major anomalies like CHARGE syndrome. All of these patients underwent a transnasal drillout procedure with placement of Portex endotracheal tube stents. The findings they reported mainly were significant for the bilateral atresia patients. Some of the predictors of favorable outcome in these children with bilateral atresia which they found included weight of >2.3 kg at time of initial surgery, a stent size > 3.5 mm used, stenting used for > 12 weeks, and the patient not having associated facial anomalies. For the children with unilateral atresia, they found no change in outcome based on the duration of stenting nor the presence of facial anomalies.

CONCLUSION

When faced with a neonate or other aged child with signs of nasal/upper airway obstruction or respiratory distress, one must consider the possible of a unilateral or bilateral choanal atresia. If choanal atresia is identified, it is necessary for the physician to investigate other possible associated anomalies such as those of CHARGE syndrome (3C’s of coloboma, choanal atresia, semicircular canals absence). If bilateral atresia is found, this is considered an upper airway emergency and the proper measures should be taken to stabilize the patient first before considering surgical intervention. When undergoing surgical treatment, one must consider the most effective approach (transnasal
endoscopic versus transpalatal) and possible treatment adjuncts like Mitomycin C and stenting to give the patient the best outcome possible.

FACULTY DISCUSSION:

**Shraddha Mukerji, M.D.**

I just had a few questions: mainly about stenting or non-stenting- Did they look at patients who were syndromic or those with bilateral atresia without syndromes? In my experience I feel that patients who have CHARGE syndrome who are syndromic and have bilateral choanal atresia may benefit from tracheostomy earlier on because they have so many other issues going on and for some reason I think the nasal bone or vomer or everything is just hard or difficult to do surgery on. It might be that even stenting these patients may definitely be required for about two months. Usually I would do it for about four to six weeks depending on the initial surgery. These are the patients that I definitely think would benefit from stenting.

Children who have bilateral choanal atresia who’s cardiopulmonary status is otherwise normal, you might want to give them a chance of initial stenting without a tracheostomy, but children who have multiple issues going on I think it would be worthwhile doing a tracheostomy, putting in a pet tube and allowing them to grow but doing the surgery and taking care of the other issues as well.

And when we do surgery the other thing is a small baby is especially difficult in a syndromic kid. And making the flaps and everything sounds very good in theory but practically it’s very, very difficult. So that does not mean that you should not try to save mucosa as much as possible but remember that it may not always be possible to do that and you don’t want to compromise the opening that you have made just because you wanted to preserve that mucosa. So I would say that whenever you think that the opening is adequate and you still have some bone to remove go ahead and remove that bone because there will be at least 20 to 30% restenosis and that’s a lot where the stenting also helps.

So that was the thing. Also I feel that using Ciprudex on these patients- it’s an antibiotic combination with steroids helps and is not toxic and doesn’t do anything different if you have not used Mitomycin C.

**Harold Pine, M.D.**

That was good, Dr. Yantis. For the resident who have not had any sort of lecture on choanal atresia, we all realize that in the NICU setting any time they can’t pass a catheter the first thing they think of is, “Oh, this baby has choanal atresia.” And it’s hardly ever choanal atresia. That automatically generates an ENT consult. And because we have the tools readily available once we get called you gotta bring the scope up there and just rule it out. Sometimes we can pass the catheter but in the end if we’re being asked to rule it out. The best way to do it is with the scope. Now if you look and think there is a choanal atresia either unilateral or bilateral, and you’re going to be sending the kid to the scanner -- one of the tricks they taught me at Great Ormond Street is to get a better
scan, Afrinize the nose first, and then suck out the nose right before they go to the scanner. That way you’re going to get rid of all the mucus and crud that’s going to build up in the nose and let the radiologist get a better sense of what’s really down there and not be confused by secretions that could have been suctioned away. One of the things that Dr. Yantis pointed out, that I think is a great thing to consider, is if you’re going to be sending the child for a CT scan of the sinuses or of the head maybe it’s best nowadays to get that temporal bone scan and really look for the semicircular canals instead of having to send them back down with these new criteria. That’s something I wouldn’t have thought of in the past, but it’s a good new recommendation.

I did a fair number of these while at Great Ormond Street and I think I’ve done maybe one since being here and I didn’t stent and I have to tell you it’s a pain in the neck putting in the stents and it’s a bigger pain taking care of the stents. You are basically having these kids walk around with foreign bodies in their noses. They get clogged. If you use a stitch to sew them in and you almost always have to and if you aren’t careful where you put the knot in the stitch if you’re not careful you’ll end up putting the knot in the darned stents and it’s blocked with mucus and it’s a real pain to keep these stents open. Going forward I’m always looking for an opportunity for not putting the stent because it’s really difficult to keep them open.

And then a final point: I have a particular friend who doing her fellowship at Cincinnati -- put one of these sounds right up into the brain and it ruined her fellowship because she was no longer trusted and I’ll tell you that when I talked to her about it, she did first look with the scope. So here’s my recommendation to you since we have it available look with the scope like we usually do in sinus surgery and get the lay of the land before you actually puncture through have someone looking from behind with the 120 degree scope. You will guarantee yourself since you’re going to see the probe knocking before it pushes through so you’ll know where your probe or your sound is going to come out into the opening and you can get fooled by just looking in the nose and end up going up higher than you thing you’re going. It’s easy to do – that’s why we have that 120 degree scope there and that’s occasionally why during an adenoidectomy I will show you guys how it works for that one particular thing.

BIBLIOGRAPHY