CHOANAL ATRESIA IN CHARGE SYNDROME: FOR THE PHYSICIAN

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ANOMALIES SEEN IN CHARGE

A little more than 50% of children with CHARGE have some form of choanal atresia. The range is complete - from bilateral bony choanal atresia to unilateral choanal stenosis. About half of the patients with choanal atresia have bilateral choanal atresia. In the general population, the incidence of choanal atresia is approximately 1 in 5000-7000 live births. A large percentage of these probably represent children with CHARGE.

DIAGNOSTIC TESTS

Physical Exam

Once the diagnosis of choanal atresia has been made, it can be confirmed initially on physical exam by failure to pass a # 6 to 8 French plastic catheter through the nares into the pharynx. (a typical solid feeling will be encountered at the level of the posterior choanal approx. 3-3.5 cm from the alar rim). Passage of soft metal probes has also been proposed.

Imaging Studies

Plain films and tomograms of the skull with radiopaque dye instilled into the nasal cavities can confirm choanal atresia. An axial noncontrast high resolution CT scan with thin sections (2-5mm) has become the single radiographic study of choice. The CT scan has proven invaluable in the accurate assessment of both the normal and abnormal anatomy of the nasal cavity, posterior nasal choanae and nasopharynx. The knowledge obtained from the CT scan is valuable in the preoperative planning of the method and design of the repair.

When planning a CT scan or MRI in a child with suspected CHARGE, consult with otolaryngology: often a few more cuts can yield important information about the inner ear abnormalities seen in CHARGE.

CONSEQUENCES OF CHOANAL ATRESIA IN CHARGE

Bilateral choanal atresia

Bilateral choanal atresia causes complete nasal obstruction - immediate respiratory distress and even potential death due to asphyxia (because newborns are obligate nose breathers until approximately 4 to 6 weeks at which time mouth breathing is learned). The respiratory obstruction is cyclic - as the child falls asleep the mouth closes and a progressive obstruction starting with stridor followed by increased respiratory effort and cyanosis. Either the observer opens the child's mouth or the child cries and the obstruction is cleared.

Initial feeding is often the alerting event - as the child starts with inability to eat and breath at the same time, there is a progressive obstruction of the airway and subsequent cyanosis and choking due to aspiration of the milk. This can mimic a tracheoesophageal fistula (TEF). TEF and/or esophageal atresia is common in CHARGE.

Unilateral choanal atresia

Unilateral choanal atresia rarely causes any acute respiratory distress. The most common finding is a unilateral mucoid discharge. Unilateral choanal atresia does not require any immediate surgical attention, but may eventually require treatment because children with CHARGE have numerous other potential respiratory complications, which can be exacerbated by UL choanal atresia.

MEDICAL MANAGEMENT

An oral airway of some sort must be implemented very early on in the treatment of newborns with choanal atresia. A typical anesthesia oral airway is often sufficient, however if not then orogastric tube may be considered. A large nipple can be modified by having its end cut off and then ties are attached to the nipple and placed around the occiput. This type of airway is called a McGovern nipple and provides an airway through which the baby can breathe. A very small feeding tube can then be passed either through another hole in the nipple or along side the nipple for gavage feeding. This is the preferred method of establishing an oral airway.

Tracheotomy and caveats for CHARGE

This is a controversial issue and many physicians conclude that there is never a need for this drastic a step in the initial management of infants with choanal atresia. Patients with CHARGE have a high propensity of airway instability. These children's early repair of their choanal atresia is rarely successful, primarily due to their abnormal anatomy of their nasopharynx and upper aerodigestive tract. Many children with CHARGE have at least one cardiopulmonary arrest prior to their definitive procedure. Therefore, some investigators have proposed that children who have CHARGE should have early tracheotomy to protect their brain from anoxic injury and delay the repair of their choanal atresia until they are at lease 2 years of age. This is still a very controversial subject.

Feeding

Gavage feeding is usually required until the child has learned to mouth breath. Then once the child learns the mouth breath, oral feeding can be attempted. A majority of children with CHARGE have significant feeding problems, possibly related to abnormalities of cranial nerves IX an X. Many of these children require gastrostomy feeding for a number of years. See sections on Swallowing and Growth.

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CHOANAL ATRESIA: PARENT INFORMATION

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NORMAL STRUCTURE AND FUNCTION

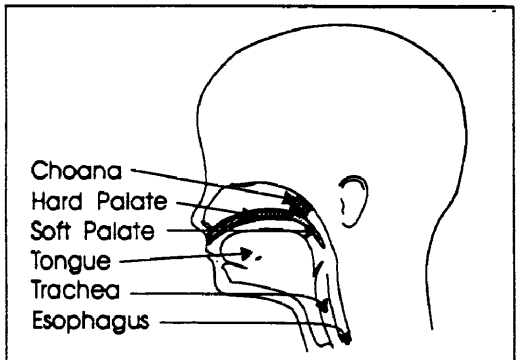

Fig. 1 Normal anatomy showing cut-away view of funnel-shaped open choana at rear of right nasal passage.

The choanae are funnel-shaped openings at the back of the nasal passages which connect the nose with the throat. They are located just above the roof of the mouth, where the hard and soft palate meet (figure 1).

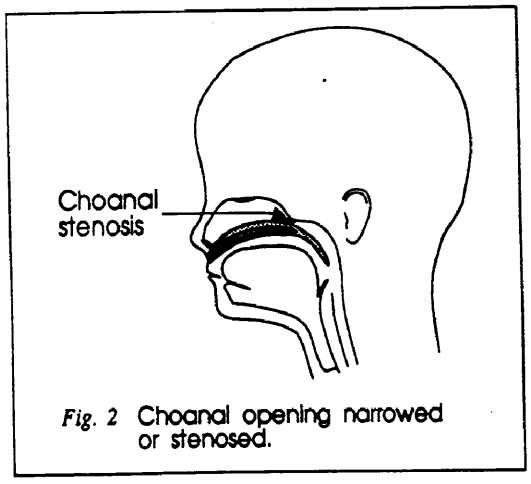

Fig. 2 Choanal opening narrowed or stenosed.

In normal fetal development, the choanae open when a membrane breaks down at about the seventh week of pregnancy. If this membrane does not break down, choanal atresia results. If the blockage is only partial, the term choanal stenosis (meaning tight or narrow) is used (figure 2). Choanal atresia is total blockage between the nose and the throat (figure 3).

The choanae are critical in the newborn because newborn babies are obligate nose-breathers. If both nasal passages are totally closed off (bilateral choanal atresia), the newborn child almost always has trouble breathing. Babies cannot instinctively switch from nose breathing to mouth breathing. This can lead to lack or air (asphyxia) with lack of oxygen (anoxia), which can result in respiratory distress, brain damage, or death if not recognized and treated quickly.

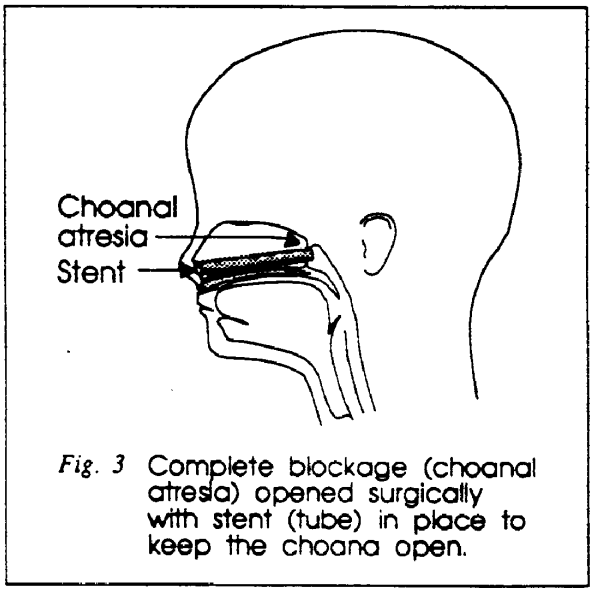

Fig. 3 Complete blockage (choanal atresia) opened surgically with stent (tube) in place to keep the choana open.

Fig. 3
Complete blockage (choanal atresia) opened surgically with stent (tube) in place to keep the choanal open.

FORMS OF CHOANAL ATRESIA IN CHARGE

About half of all children with CHARGE have some form of choanal atresia. It is very rare in other conditions, and therefore very helpful in making a diagnosis of CHARGE. Children with CHARGE can have choanal atresia (blockage) or choanal stenosis (narrowing). It can affect both sides (bilateral, BL) or only one side (unilateral, UL). Choanal atresia can be bony (bone or cartilage blocking the passageway) or membranous (soft tissue membrane blocking the passageway).

Choanal atresia can be present even if there is a cleft palate. It may be more difficult to diagnose, as air can move through the cleft. Unilateral choanal atresia and choanal stenosis can be very difficult to recognize.

DIAGNOSIS OF CHOANAL ATRESIA

Polyhydramnios (excess amniotic fluid) during pregnancy is often the first sign of choanal atresia. Normally, the fetus breathes amniotic fluid during pregnancy. If the nose is blocked, and the fetus can't "nose breathe," the amniotic fluid is not "recycled" by fetal breathing motions and excess fluid may build up in the uterus.

A tube should be passed through each nostril and nasal passage of any infant who has unexplained breathing difficulties in the first month of life. This will establish if the nasal passages are open (patent). X-rays can be performed using dye in the nasal passages to verify they are not open. Sometimes other imaging (MRI, CT) is used to confirm the diagnosis and determine the extent of the problem.

TREATMENT

Bilateral choanal atresia

Bilateral choanal atresia is life threatening in the newborn period, and treatment is urgent. Emergency treatment usually consists of placing a plastic airway (tube) into the mouth to keep the mouth open. This allows the baby to mouth-breathe. Other newborns may require intubation: passing a breathing tube through the mouth and down into the windpipe (trachea) so oxygen goes directly into the lungs. Occasionally, it is necessary to do a tracheotomy: surgically putting a breathing tube directly into the trachea through a hole in the lower front of the neck.

The above procedures are all temporary. Soon after, surgery will be performed to open the bone or membrane covering the nasal passage. A stent (plastic tube) is placed in each nasal passage to keep it open and guarantee adequate air entry (figure 3). The stents may have to stay in for a few weeks or even months. Children with CHARGE are more likely to have complications following choanal atresia repair than other children with choanal atresia. A few require repeat surgery later because the choanae close up again when the stents are removed.

Unilateral choanal atresia and/or choanal stenosis

Lesser problems usually occur when one nasal passage is totally obstructed (UL atresia) or when one or both nasal passages are narrowed (stenosis). Air can pass in varying amounts through the nose into the lungs. The child with UL atresia or stenosis often does not have obvious respiratory symptoms, but he or she may have a constant runny nose on the side that is blocked or narrow. Recognizing choanal stenosis or unilateral atresia can be important in the treatment of these respiratory complications.

OUTCOME

A major long-term complication of choanal atresia or stenosis is recurrent ear infections, which can lead to a conductive hearing loss. Because children with CHARGE also have a high risk for sensorineural hearing loss, these ear infections should be treated aggressively.

Children with CHARGE may require more than one surgery before the choanal atresia is permanently repaired. Often, difficult decisions must be made about surgery - should the heart be fixed first or the choanal atresia? Is the baby strong enough for surgery, given the heart problems, etc? Any newborn with CHARGE should have the choanae evaluated before any surgery is done because of possible complications. Choanal atresia and the resultant lack of oxygen increases the risk for mental retardation due to brain damage.