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CASE REPORT

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An unusual presentation of bilateral choanal atresia: How often is the diagnosis missed?

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Abstract: Bilateral choanal atresia is a rare congenital anomaly commonly seen in CHARGE syndrome in which the posterior airway is obstructed between the nose and the pharynx in both nostrils. It constitutes a neonatal emergency. We report the case of bilateral choanal atresia in a late preterm female infant who presented with respiratory distress and cyanosis on the first day of life but whose definitive diagnosis was achieved in the second week due to other antecedent co-morbidities. We highlight the diagnostic challenges and the need to have a high index of suspicion in similar situations.

Keywords: Choanal atresia, breathing difficulty, CHARGE

Introduction

Choanal atresia is a congenital anomaly in which there is an obstruction of the nasal passage posteriorly. It is rare and has a frequency of approximately 1 in 7,000 live births. It can either be unilateral or bilateral. Unilateral defects are more common, with a male: female ratio of approximately 1:2. [1,2] Choanal atresia can be classified into three types: bony, mixed bony-membranous, and membranous. Based on computed tomography scan reports, Brown et al. classified choanal atresia as bony (29%) or mixed bony-membranous (71%), with no pure membranous. [1-3] A few risk factors linked to choanal atresia have been identified: exposure to atrazine herbicide,[,5] coffee consumption, second-hand smoke, high maternal zinc, vitamin B2, and maternal antithyroid medication in the first trimester.[6] Approximately 50–70% of affected infants have other congenital anomalies (CHARGE syndrome Treacher-Collins, Kallmann syndrome, VATER association, Pfeiffer syndrome), with the anomalies occurring more often in bilateral cases. [1,7,8]

Choanal atresia can be suspected if it is impossible to insert a nasal catheter. The catheter length that can be inserted indicates where choanal atresia has occurred. Shorter distances indicate obstruction around the vomer, while longer distances around the posterior choanae.[1] Failure to pass the catheter through the nasal cavity more than 5.5 cm from the alar rim and the lack of movement of a thin wisp of cotton under the nostrils while the mouth is closed are diagnostic, and a CT scan confirms the diagnosis.[9]

We present the case of a female infant with a delayed diagnosis of bilateral choanal atresia to highlight the varied presentation, possible diagnostic challenges, and the need to have a high index of suspicion in similar situations.

Case Report

Baby A, a 3-day old late preterm low birth weight, appropriate for gestational age female neonate, was brought by the father to the Children Emergency Centre following a referral from a peripheral hospital with complaints of respiratory distress noticed from the 14th hour of life. The child was delivered via emergency caesarean section on account of severe pre-eclampsia at 36 weeks gestational age to a 32-year-old mother. The baby cried at birth, and breastfeeding was initiated with the mother's own expressed human milk. At about the 14th hour of life, she was noticed to have choked on feeds while bottle-feeding, developed respiratory distress, and became cyanosed. She was resuscitated and placed on intranasal oxygen and the cyanosis resolved. However, respiratory distress and tachypnoea continued. Chest x-ray (CXR) and echocardiography were requested at the referral center. The CXR showed some patchy opacities on the upper lung zones. A large PDA, small ASD, as well as severe pulmonary hypertension were revealed by the echocardiography. These findings necessitated the referral of the baby on the third day of life.

Upon admission, the baby was crying, and her oxygen saturation was 94% on supplemental oxygen. She had a

receding chin, was dyspnoeic and tachypnoeic, and had fine crepitations over both lung zones. An initial assessment of aspiration pneumonitis to rule out congenital pneumonia was entertained at the emergency center.
At admission into the ward, an additional assessment of congestive cardiac failure was included in the diagnosis, and the patient was commenced on an anti-failure regimen. The result of the complete blood count was not suggestive of sepsis. The baby was tolerating expressed breastmilk feeding via an orogastric tube (the patient was connected to oxygen via nasal prongs). However, respiratory distress, tachypnoea, and noisy breathing with copious nasal discharge continued. After several days on admission, she was observed to be persistently mouth-breathing, and respiratory distress seemed to improve when she dislodged the nasal prongs into her mouth. She was suctioned frequently, and a conscious attempt to pass a nasogastric catheter was met with resistance during one such suctioning. The nasogastric tube could not be inserted in either nasal cavity. A thin wisp of cotton was placed under the nostrils while the mouth was closed, and no air movement was detected in both nares. This prompted a diagnosis of bilateral choanal atresia with a strong suspicion of CHARGE Syndrome.

A multidisciplinary approach was employed in her management subsequently. The ophthalmology examination was normal. The Ear, nose, and throat surgeons were invited, and they reviewed the patient. Due to financial constraints, many of the investigations requested were not carried out on time. A CT scan examination done on the 12th day of life confirmed a bilateral osseous choanal atresia and right external auditory canal atresia (Figure 1). With the identified anomalies in the child, which included arched eyebrows, a receding chin, heart defects, right external auditory canal atresia, as well as choanal atresia, a diagnosis of bilateral choanal atresia with a strong suspicion of CHARGE syndrome was made. The baby's airway was maintained temporarily by using a small-sized orogastric airway while being worked up for surgery.

Intraoperatively, mixed bilateral (bony and membranous) choanal atresia was identified. Bilateral choanoplasty was carried out, and a stent was left in place bilaterally to maintain the airway (Figure 2). Postoperatively, the child was placed on analgesics, some nasal drops, and antibiotics. She was continued on expressed breastmilk and was discharged to the clinic after three days with instructions to introduce direct breastfeeding gradually. A repeat echo at discharge revealed only a tiny patent Foramen Ovale with mild pulmonary hypertension. On the first follow-up visit, the mother reported mild respiratory distress while directly breastfeeding but fed well with a cup. The patency and functionality of the stents were checked and were found intact. She is currently on monthly clinic visits and is stable.

Fig 1: CT scan showing the Bilateral Osseous obstruction (Choanal Atresia).

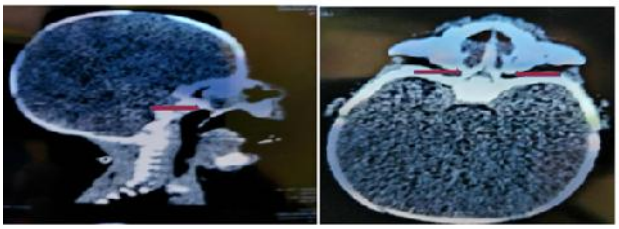

Fig 2: Baby with the stents after the surgery

Discussion

Newborns are traditionally considered to be obligate nose breathers, with mouth breathing established by approximately 4–6 weeks of life.[1] If the nasal obstruction is unilateral, the newborn can breathe using the opposite side. However, if bilateral, the newborn breathes while crying; hence, the reason cyanosis improves during the cry. While bilateral choanal atresia may be easily detected, it may also be missed in babies who can adapt to mouth breathing. Research has shown that newborn infants have variable abilities to breathe through their mouths, so nasal obstruction does not produce the same symptoms in every infant.[1] Infants are not obligate nose breathers.[10,11] Rodenstein et al demonstrated that in acute nasal obstruction, infants could initiate oral breathing by detaching the soft palate from the tongue, thereby opening the oropharyngeal isthmus to airflow.[10] The index patient, was diagnosed after 12 days because she adapted to mouth breathing. The underlying cardiac defect and the persistent pulmonary hypertension were initially focused on as the reason for the respiratory distress.

Infants with bilateral choanal atresia who have difficulty with mouth breathing make vigorous attempts to inspire, often suck on their lips, and develop cyanosis. Those who can breathe through their mouths have difficulty coordinating sucking and swallowing with breathing; hence, they may aspirate and become cyanotic during feeds.[1] The index patient was said to have choked on feeds, became cyanosed, resuscitated, and was placed on nil by mouth until she was referred. Bilateral choanal atresia is an emergency that is initially best treated by inserting an oropharyngeal airway to break the seal formed by the tongue with the palate so that the airway

can be kept patent temporarily till elective surgery is undertaken. This was done for the index patient with marked resolution of respiratory distress. In the absence of an oral airway, the McGovern nipple or intubation could be an alternative method of securing the airway. [12]
The index patient has at least three of the major distinguishing characteristics of CHARGE syndrome - atresia choanae, heart defects, and ear abnormalities and several minor characteristics such as micrognathia, dysmorphic features, and respiratory distress. Harris et al., [13] in an analysis of choanal atresia-associated malformations, indicated a weak association between the malformations of CHARGE and concluded that the diagnosis be restricted to infants with multiple malformations and choanal atresia and/or coloboma combined with other cardinal malformations (heart, ear, and genitals). Several other researchers also identified coloboma, choanal atresia, and ear abnormalities as major diagnostic features,[13-15] while characteristics like growth retardation are typically noticed in late infancy. The lack of this major characteristic finding may have contributed to the delayed diagnosis seen in index patient with the apparent low index of suspicion we exhibited for the syndrome. The role of health insurance for newborns to enable prompt diagnosis and earlier interventions cannot be over-emphasized. This played out in the delay in reaching a definitive diagnosis in our patient as most of the associated anomalies identified required imaging studies done on the 12th day due to out-of-pocket payment. The use of imaging studies to assess multiple congenital anomalies is very important as CT scans and Echo studies clinched the diagnosis in our patient. CT scan was diagnostic of osseous bilateral choanal atresia, one of the classical types of choanal atresia.[1,8,9] It also detected the right external auditory canal atresia. CT Scan has been demonstrated to be the imaging of choice in the diagnosis of choanal atresia. It can be used to characterize the type and degree of atresia as well as identifying and differentiating other causes of nasal obstructions in the newborns.[16,17]

Counselling caregivers is a major feature in managing children with congenital anomalies to ensure optimal care.[18] Counselling our index patient's parents was done severally with good psychosocial support by the hospital team. Both parents were involved in making management decisions and showed enthusiasm for attaining the best care possible for the baby.
Management of bilateral choanal atresia as an association with CHARGE syndrome is multidisciplinary and depends on the severity of the presentation.[1] The surgical treatment for bilateral choanal atresia is geared toward establishing nasal patency through the transnasal puncture, transpalatal resection, and endoscopic resection, which can be supported with or without stents.[8] Our patient had a transnasal puncture, after which stents were put in place. She had minimal complications following the procedure and was discharged after three days.

Conclusion

Bilateral choanal atresia is a rare clinical condition whose diagnosis can be challenging, especially in babies who adapt to oral breathing and have no other visible malformations. A newborn that mouth-breathes should raise a very high index of suspicion for the condition. A good clinical evaluation with appropriate imaging, such as computed tomography, is indispensable for diagnosis and appropriate intervention.

Author Contributions
MAA and BNE conceived the idea and designed the study. MAA, ANA, and ERB contributed to data acquisition, while all contributed to the interpretation of data. MAA, ANA, and BNE drafted the manuscript. BNE, IBF, and VCE revised it critically for important intellectual content. All gave the final approval for the version to be published.
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