Congenital Arhinia

Hasan Alansari, MB, BCh, BAO MRCS (ENT)* Ahmed Fawzi, MSc**
Raneem Alshaikh, MD, BSc*** Noora Althawadi, MB, BCh, BAO***
Waleed Janahi, FRCS (Ed.), MD, MSc**** Isa Alsaad, MBBS, FAAP, FRCP(I), MSc*****

Congenital arhinia is a rare condition described as the absence of the nose at birth. It is a life-threatening condition if not managed properly. Due to its low incidence, not much is understood about the condition.

We report a case of congenital arhinia diagnosed antenatally and managed in the Neonatal Intensive Care Unit (NICU). The case was diagnosed at 27 weeks of gestation when fetal ultrasound revealed absence of nasal bones. At birth, the baby was in respiratory distress and required oral intubation followed by elective tracheostomy.

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Congenital arhinia is described as the absence of the nose at birth. It is a rare condition that has been documented less than 50 times since it was first seen in 1931. Due to neonates being obligatory nasal breathers, congenital arhinia will lead to severe upper airway obstruction and could potentially be life-threatening to the child if not managed properly. The pathogenesis is not well understood due to the scarcity of cases; however, there is an association with other craniofacial malformations[1,2,3].

The aim of this study is to report a case of congenital arhinia diagnosed antenatally and managed in the Neonatal Intensive Care Unit (NICU).

THE CASE

A baby female was born at 38+5 weeks of gestation by emergency Cesarean section due to fetal distress. There was a history of maternal gestational diabetes mellitus and polyhydramnios during pregnancy. Ultrasound performed at 27 weeks of gestation revealed absent nasal bones. At birth, the neonate had an APGAR score of 5 and 8 at 1 and 5 minutes, respectively. Birth weight was 2.66 kg with normal length and head circumference.

Soon after birth, the baby was intubated due to severe respiratory distress and was admitted to the NICU. She was initially kept nil per mouth, and the feeding was started on the second day of life via an orogastric tube.

Skull CT was performed, which reported the absence of the midline nasal skeleton, including the septum, perpendicular plates of ethmoids and the absence of nasal cavity and paranasal sinuses, see figures 1 and 2. In addition, subluxed lens in the right orbit was reported. Additional workup included echocardiograph, abdominal ultrasound, and karyotyping, which were normal. Craniofacial and ophthalmology consultations were sought; a tracheostomy and examination of the eyes under general anesthesia were advised.

The patient had tracheostomy with size 3.5 non-cuffed Shiley tube. Eye examination revealed non-ophthalmic on the right

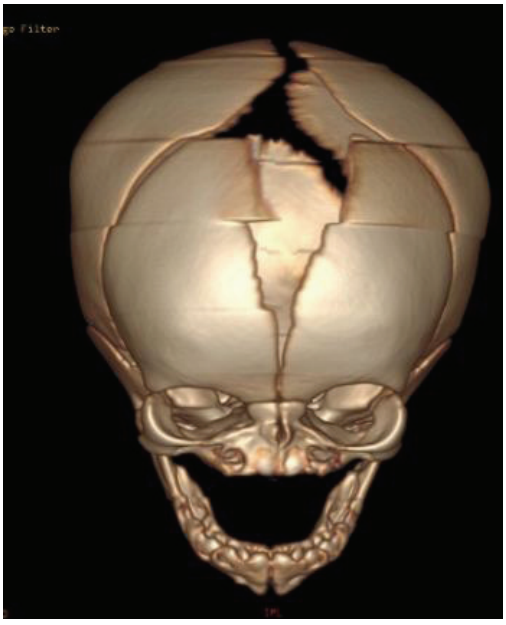

Figure 1: 3D Reconstruction of Facial Bones

* Registrar
Department of ENT
** Registrar
Department of Pediatrics
*** Senior House Officer
Department of ENT
**** Consultant
Department of ENT
***** Consultant Paediatrician, Neonatologist and Head of NICU
King Hamad University Hospital
Kingdom of Bahrain
E-mail: hassan.alansari21@gmail.com, hassan.alansari@khuh.org.bh

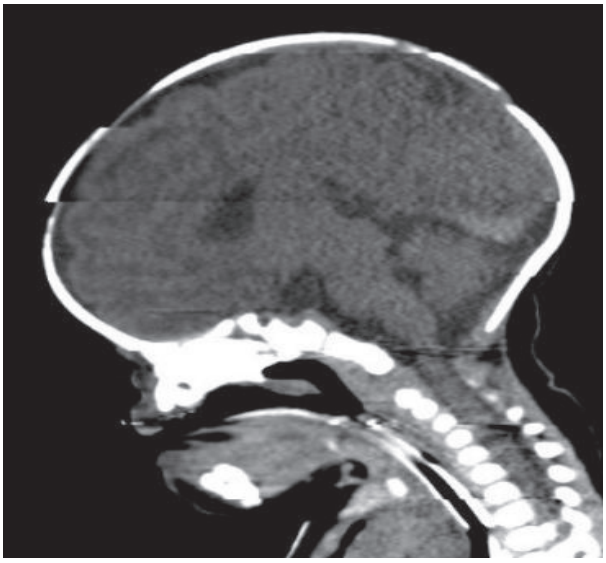

Figure 2: Sagittal Cut of Paranasal Sinus

with a corneal diameter of 5 mm and intraocular pressure of 9 mmHg. Choroidal coloboma with a retinal detachment was found. No postoperative complications were encountered. The baby was weaned off ventilation one day postoperatively and was started on oral feeding.

She is currently in the NICU, maintaining oxygen saturation on room air and tolerating full oral feed, see figure 3. Consent for publication was obtained from the patient's parents.

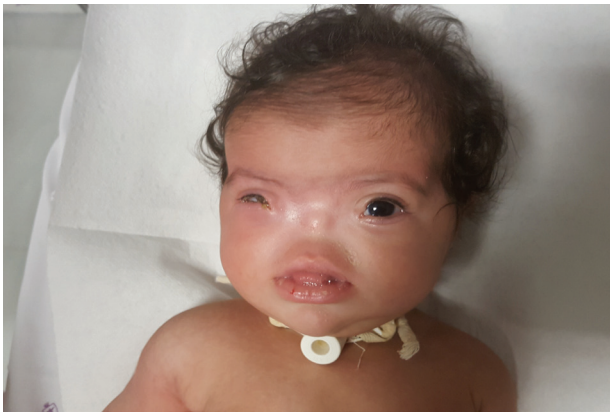

Figure 3: Post-Tracheostomy

DISCUSSION

The embryological development of the face and nose starts at 3 weeks of gestation and continues to the tenth week[4]. Early in the embryological development, the face is made up of a superior frontal process, paired bilateral maxillary processes and a pair of mandibular processes caudally[5]. Two ridge-like structures known as the nasal placodes develop on the 4th week of gestation. These placodes eventually form the lateral and medial nasal swellings. The nasal placodes invaginate on the underlying mesenchyme to form nasal pits around the 5th week of gestation and the medial nasal swellings fuse to form the nasal septum. During the 6th week, the maxillary and frontal processes fuse to form the primitive palatal shelve, and the cells from the nasal pit migrate posteriorly to form the primitive nasal cavity. Merging of the said processes occurs in week 7 and week 8, however, in cases of congenital arhinia the lateral processes and maxillary processes fail to fuse which results in the anomaly[6].

There is no gene mutation contributing to arhinia, and less than a handful of cases had an abnormal chromosomal analysis[2]. However, congenital arhinia is said to be associated with other malformations such as cleft palate, hypertelorism, umbilical hernia, neurological anomalies and absence of the paranasal sinuses[7].

Antenatal ultrasound can be useful in detecting congenital arhinia; only one case was detected in the second trimester[8]. Most of the patients with this anomaly were diagnosed at birth except for 6 reported cases including ours. Early diagnosis allows for better education of the parents and allows the medical team to be better prepared for delivery and its complications. Initial management involves protecting the airway which is usually achieved through endotracheal intubation at the time of birth, which is critical as neonates with arhinia can quickly develop respiratory distress. Prompt identification of the condition at the time of birth and proper insertion of an airway can prevent possible hypoxia-related complications, including death. Placement of an endotracheal tube prevents the neonate from feeding orally and is not ideal for long-term management[9]. A tracheostomy tube can be placed once the patient is stable and fit for surgery. This will facilitate oral feeding and contribute to the neonate's overall health and avoiding prolonged intubation. The tracheostomy tube can be removed once the patient is able to maintain oxygen saturation through mouth breathing while sleeping and feeding.

Reconstruction of the nose is another dilemma. Due to the various degrees of nasal deformity or absence, each case has to be studied carefully to reach an individualized surgical care plan. Repair of the nose can be divided into two steps; the first step involves creating a nasal cavity, which could be performed by simply drilling two separate nasal cavities and leaving silicone tubes in the new passageways so they can remain patent[10]. Another method involves the use of maxillary osteotomies to create new epithelium-lined nasal cavities. The second step in the repair process involves constructing an external nose to achieve functional and aesthetic results. This can be achieved with the placement of skin expanders on the forehead, an external nose can then be created with the use of the expanded forehead flap with local peri-nasal flaps and costochondral grafts[11].

The age at which the patient can undergo such surgery is debatable. Some surgeons prefer waiting until the patient reaches adolescence or young adulthood, whilst others prefer intervening early to lessen the psychological impact on the patient[12].

CONCLUSION

Congenital arhinia is a rare condition that is not seen in every day practice, not much is known about the cause and no gene mutation has been implicated. Airway management is vital for long-term neonatal care; this is usually achieved via intubation followed by a tracheostomy.

Nasal reconstruction is usually complex and is performed in 2 stages, the timing of the reconstruction is still being debated amongst surgeons.

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