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Case Report

Congenital arhinia: a highly unexpected presentation

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ABSTRACT

Congenital nasal absence (arhinia) is an extremely rare malformation. Arhinia causes severe airway obstruction and poor feeding in the affected neonate. There is an association with other facial anomalies, especially defects of the eyes, ears, palate, and midline defects. In this article, we present a 16-month-old child born with absent nose, microphthalmia and cleft palate. Such patients require multidisciplinary team approach for appropriate care and management.

Keywords: Arrhinia, Multidisciplinary approach, Facial anomalies

INTRODUCTION

Congenital arhinia is a rare defect of embryogenesis.[1-6] The lack of an external nose is generally one part of a complex malformation syndrome1 characterized by the absence of nasal cavities, microphthalmia or coloboma, absence of olfactory bulbs, high arched palate and microtia.[3] In this article we present a 16-month-old female child born with arhinia, cleft palate, microphthalmia and hypertelorism and her complex management process.

CASE REPORT

This child is born in our hospital at 37 weeks, via normal vaginal delivery with a birth weight of 2.6 kg, with congenital absence of nose, low set ears, hypertelorism, cleft palate and microphthalmia (Figure 1 and 3). Antenatal scan reported a tiny nasal bone. Echocardiogram done after birth revealed a large patent ductus arteriosus 3 mm with left to right shunt. She underwent tracheostomy few days after birth and was referred to tertiary centre for multidisciplinary work up. Assessment by the ophthalmologist revealed a suspicious right eye pupil distortion, iris coloboma, cataract and absence of fundal glow in the right eye while it was present in the left. However, she is on regular follow up for assessment of degree of vision in the right eye. The assessment of her swallowing reflex was done by modified barium swallow. It showed poor oral intake but with functional pharyngeal swallow and signs of breathing.

There were signs of incoordination resulting in silent aspiration with large bolus. Functional endoscopic evaluation of swallowing with single feeds revealed mild oral aversion, functional pharyngeal phase, good pharyngeal swallow initiation, adequate laryngeal elevation. Though obvious aspiration was not noted, due to the high probability of silent aspiration, percutaneous endoscopic gastrostomy was advised, as long-term supplemental feeding is to be expected. Ultrasound abdomen was normal.

CT scan head was grossly normal apart from arhinia, absent nasal cavities, hypertelorism, inferior continuation of metopic sutures, cleft lip and cleft palate. MRI brain was also normal (Figure 2 and 4). Currently she is 16 months old (Figure 5), on tracheostomy, with microphthalmia under investigation by ophthalmology

team for any further malformation, and a plan for surgical correction of cleft palate. Her weight is 6.9 kg, made good progress along the curve, with adequate development. The Plastic surgeon suggested nose prosthesis at the age of 5 years with glasses. As per family history, her siblings include 2 elder brothers and a sister, all healthy. There is a history of Cleft palate present on the maternal side. Microarray chromosomal study was reported as normal.

A possibility of Bosmia Arhinia Microphthalmia (BAM) syndrome is to be ruled out. For this, a study to look for mutation in SMCHD1 gene needs to be carried out. As this is likely to be of low occurrence as de novo or autosomal dominant inheritance, and since it will not help the family hence it has not been done so far. Since a case of BAM syndrome may be expected to have hypogonadotropic hypogonadism in future presentation, this evaluation is expected to be obtained by 11 years of age. She is on regular follow up with us for further appropriate management for her age.

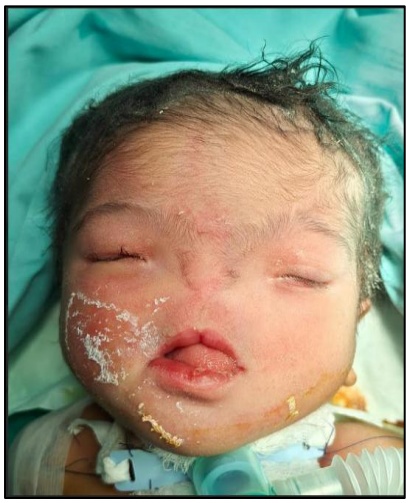

Figure 1: Absent nose.

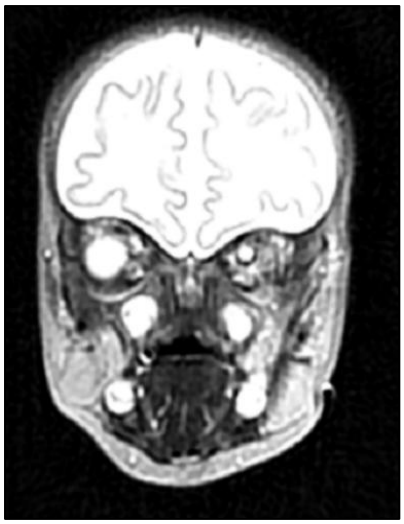

Figure 2: MRI showing absent nasal cavities.

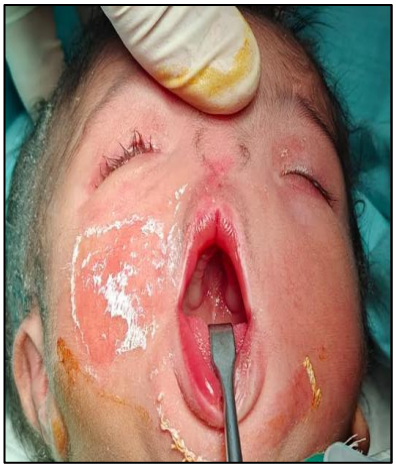

Figure 3: Cleft palate.

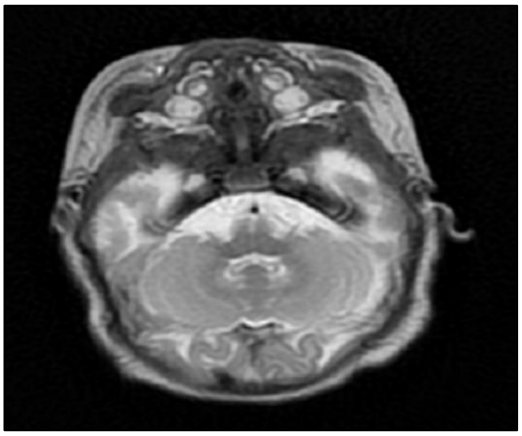

Figure 4: MRI showing palatal defect.

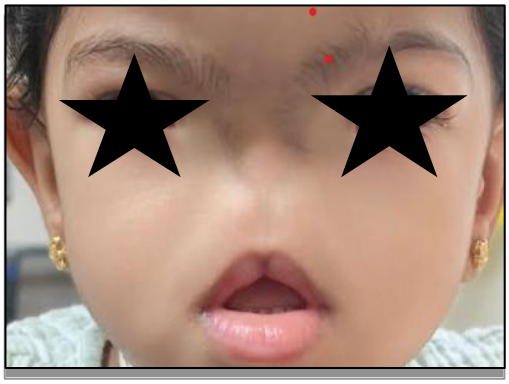

Figure 5: 16-month-old.

DISCUSSION

Congenital arhinia is one of the rare defects that occurs during early embryogenesis. Worldwide, there are less than 100 cases of congenital arhinia reported as per literature. The development of nose and nasal cavity occurs between third to tenth weeks of gestation. Rapid differentiation and division occur during this time where the nasal bridge, nasal septum, nostrils, soft and hard

palate are formed. Pathogenesis of arhinia is still not clearly understood. There were few postulations of how arhinia may happen, which include failure of medial and lateral nasal processes to grow; premature fusion of the nasal medial processes; lack of resorption of the nasal epithelial plugs.[6] From the review of literature, congenital arhinia is not associated with any specific genetic mutation.[4] Most of the patients with congenital arhinia have normal chromosomal analysis which is similar to that of our patient. Many of the patients with congenital arhinia have other associated deformities such as absence of the paranasal sinuses, cleft palate, hypertelorism, central nervous system anomalies, umbilical hernia, and syndactyly. Arhinia is associated with the following conditions and syndromes: Arhinia with choanal atresia and microphthalmia syndrome (BAM syndrome); Holoprosencephaly 1; Holoprosencephaly 13, X-linked; Holoprosencephaly-radial heart renal anomalies syndrome.

BAM syndrome is an extremely rare genetic disorder that has been reported in fewer than 100 patients worldwide in the past century. It is defined by three major features: complete absence of the nose; eye defects; absent sexual maturation. The only known genetic cause of BAM syndrome is a change (variant) in the SMCHD1 gene. In most patients, this change occurs spontaneously in the egg or sperm cell and is not inherited from the parents.[8]

The most immediate consequence of patients with congenital arhinia is airway obstruction and feeding difficulty. Our patient was intubated soon after birth and she required minimal ventilatory support and further on, tracheostomy was done to maintain her airway. Her issues with swallowing were dealt with percutaneous endoscopic gastrostomy tube insertion.

CONCLUSION

Congenital arhinia is one of the rare craniofacial malformations that can cause severe respiratory distress at birth due to upper airway obstruction. It needs a multidisciplinary team work. It represents a challenge to paediatricians, maxillofacial surgeons, otolaryngologists and plastic surgeons. Construction of the nose is usually delayed till preschool years when facial development is nearly completed. This was the first case reported from South Sharqiyah governorate, Sultanate of Oman.

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Ethical approval: Not required

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