Congenital Arhinia

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ABSTRACT

Congenital arhinia is a rare craniofacial malformation . The involved baby is usually delivered with respiratory difficulty that may be severe enough to need urgent surgical intervention. As in many congenital anomalies, arhinia may be presented with other congenital anomalies like eye, midline and olfactory nerve defect, in addition to distant anomalies like heart, renal and central nervous system involvement.

Here we are reporting a case of congenital arhinia presented at age of 6 months to our otolaryngology department at Al-Shaheed Ghazi Al-Hariri Hospital, Medical City in Baghdad. The patient was on mouth piece airway to maintain and orongastric tube for feeding since birth. Both tubes were removed and the patient could breath without difficulties because didnot have cleft palate or other oral abnormalities. The corrective surgery was deferred till preschool age to give a time for more facial maturation.

Key words: Congenital abnormalities, Arhinia, respiratory distress.

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INTRODUCTION

Congenital Arhinia is one of the rare craniofacial malformations with only 45 cases reported since 1931.[1] It may cause severe respiratory distress at birth due to upper airway obstruction.

Other congenital anomalies such as eyes, midline defects and central nervous system anomalies may also be associated. The aetiology largely remains unknown.

CASE PRESENTATION

A female full-term infant baby was delivered to a 22-year-old lady and 24-year-old husband who were cousins. This was their fourth pregnancy, the previous three were aborted no definitive cause for these abortions was reported. The parents reported no family history of significant congenital abnormalities.

This was the mother's 4th pregnancy, her previous pregnancies were aborted for unknown cause. The mother reported that she was visiting antenatal care regularly; nothing was of significance apart from a history of pyrexia suspected as being typhoid fever (the diagnosis was based on clinical assumption without laboratory prove) and treated by injectable antibiotics. The pregnancy was ended at term by caesarian section due to failure of progression of the labour.

On delivery, the caring physician has noticed that the baby has no nose and she was in shorts of breath. Baby was admitted to a neonatal care unit for 15 days. Airway piece was inserted to facilitate breathing and an orogastric tube was placed for feeding (see **figure 1**). The child was discharged home and kept on oral airway mouth piece and orogastic feeding.

At age of 6 months the family sought medical help and visited otolaryngologist to wean the child from the oral airway and orogastric

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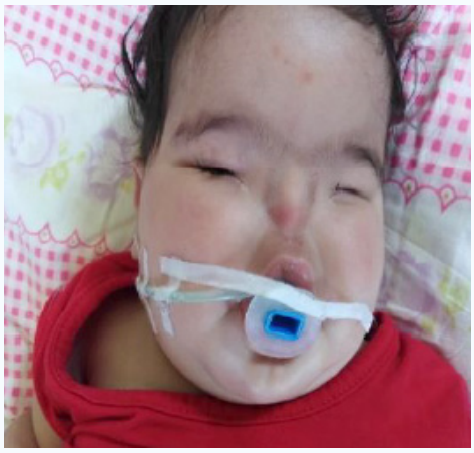

Figure 1: Picture showed absence of the nose airway mouth piece and oronasal tube are in place.

tube. Otorhinologological assessment shows a 6-months-old child weighting about 8 KG with normal developmental milestones. Nasal examination showed a complete absence of the nose, nasal roots and pits. Examination of the eyes showed hypertelorism and microphthalmia bilaterally. The palate was high arched. Echocardiography showed large size aneurysmal secondum atrial septal defect. While abdominal ultrasound revealed normal kidneys.

Radiological assessment showed absence of the nasal cavity, poorly developed nasal septum, bilateral small maxillary antrums, non-fusion of both zygomatic bones, presence of both eyeballs and absence of cribriform plate. **Figrue 2**.

Our primary decision was to wean the child from the airway piece and oro-gastric tube which was successfully done and the child start breathing normally on room air and on oral feeding (**figure 3**). A chance given to her to grow older before any surgical intervention.

DISCUSSION

Congenital arhinia is the absence of external nose, nasal cavity and olfactory apparatus. The pathogenesis of congenital arhinia is not well understood. The nose forms between 3rd and 10th weeks of gestation.[2]

In arhinia, there is failure of development of nasal placode on one or both sides leading to failure of growth of medial and lateral nasal prominences. This also secondarily leads to failure of development of olfactory bulb and tract. Failure of absorption of nasal epithelial plug during 13–15th weeks of gestation,[3] premature fusion of medial nasal prominences and migration defect of neural crest cells have also been proposed in the pathogenesis of arhinia.[4]

Arhinia may be associated with other anomalies such as absence of olfactory bulbs and nerves, absence of paranasal sinuses, high arched or cleft palate, eye anomalies, low set ears and central nervous system abnormalities.[5] In our case local developmental abnormalities of the nose and sinuses were found. In addition to atrial septal defect. No other distant congenital anomalies were found.

The problem with Arhinia is that it may lead to airway obstruction and feeding difficulty; our case has no cleft lip or palate and she can breath and eat normally after removal of the airway mouth piece and oronasal tube.

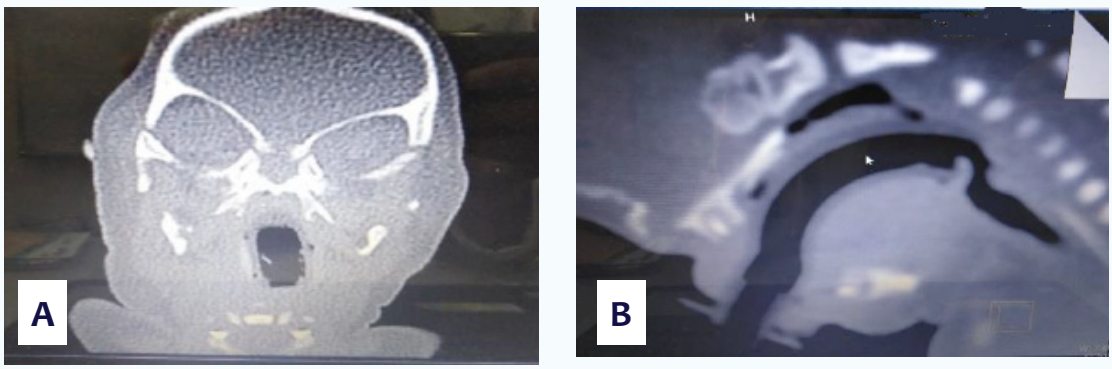

Figure 2: CT scan of the face, coronal reconstruction (A) and sagittal reconstruction (B) shows absence of the nasal cavity, poorly developed nasal septum, bilateral small maxillary antrums, non-fusion of both zygomatic bones, presence of both eyeballs and absence of cribriform plate.

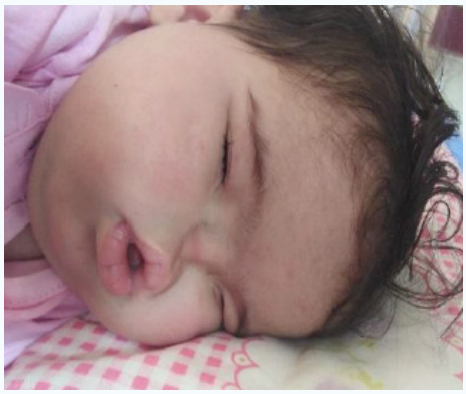

Figure 3: The child at age of 6 month breathing normally and eating orally..

Management of congenital arhinia mainly involves management of airways and feeding difficulty. Neonates are obligate nasal breathers. Immediate management includes oral airway, tracheostomy in case of life-threatening respiratory distress in neonatal period. Fortunately we are in no need for such intervention in our case.

Surgical reconstruction needs multidisciplinary approach involving neonatologist, paediatric, plastic, and craniofacial surgeons. Surgical reconstruction is preferably delayed until preschool age because facial development is almost complete till then.[6] Psychological support of the parents is also essential in the management of such cases.

CONCLUSION

Congenital arhinia is a rare anomaly and needs multidisciplinary approach. Airway and feeding management is the mainstay of treatment. Reconstructive surgery can be delayed until preschool age.

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