CASE REPORT

Congenital arhinia – First published case in Malaysia

Ng Rui Lun, MPAEDS, Kokila Rajapathy, MPAEDS, Zainab Ishak, MPAEDS

Department of Paediatrics, Hospital Kulim, Kedah Darul Aman, Malaysia

SUMMARY

Congenital arhinia is one of the rare craniofacial malformation that may cause severe respiratory distress at birth due to upper airway obstruction. Our patient, whose abnormalities were only detected after delivery in our centre, is the first reported case of congenital arhinia in Malaysia. Contrary to popular belief that neonates are obligate nasal breather, our patient adapted well to breathing through mouth before an elective tracheostomy was performed on day four of life.

KEY WORDS:
Arhinia, Nose, Congenital

INTRODUCTION

There are not many reported cases of congenital arhinia worldwide and at present no case of congenital arhinia from Malaysia is found in literature. The most worrying aspect of managing patient with congenital arhinia is severe upper airway obstruction. We hope to use this case as an illustration that neonate may also breathe via mouth and patient with congenital arhinia can tolerate well on room air until definitive airway is planned.

CASE REPORT

A female term infant was delivered via spontaneous vertex delivery by a 31-year-old mother. This was her second marriage, fifth pregnancy and third living child. Both parents were non-consanguineous and there was no family history of congenital malformation. During antenatal check-up, the mother had gestational diabetes but her blood sugar was well controlled with diet restriction. During the third trimester, she had polyhydramnios with Amniotic Fluid Index (AFI) of 24cm. A detailed ultrasound scan performed by a maternal foetal specialist showed a normal female foetus with no obvious structural abnormality. However, the maternal foetal specialist was unable to visualise the foetus face at that instance as the foetus position prevented adequate views of the face.

Paediatric team was asked to review patient after she was born. On physical examination, the baby had no external nostrils or nasal pyramid. However, she had a small longitudinal ridge-like protuberance in the midface. Besides, she also had micropthalmia and microcornea, low set ears, hypertelorism, and overlapping fingers. Other systemic examinations were unremarkable. She did not show any signs of respiratory distress despite having absence nasal passage. Patient was admitted to Neonatal Intensive Care Unit (NICU) for close observation and further management. In the ward, she could maintain normal saturation under room air without any signs of breathing difficulties. She was initially kept nil by mouth in anticipation of airway management, but feeding was started on second day of life when she adapted well to mouth breathing. We had decided to commence feeding via orogastric route instead of oral feeding in fear of aspiration and respiratory distress during feeding. On following days, patient was comfortable under room air and tolerated feeding well.

After discussion among parents, paediatrician and otorhinolaryngologist, she had undergone an elective tracheostomy on day four of life. Computed tomography (CT) of the face and neck showed narrowing of the nasal cavity with absence of the nasal septum and stenosis of the pyriform aperture and bilateral choanal. The nasal cavity is filled with soft tissue density. We had arranged for few other tests to look for associated structural abnormalities for this female infant, i.e., echocardiography which showed a fenestrated atrial septal defect and abdominal ultrasound which showed splaying of left pelvicalyceal system. Chromosomal Analysis showed a normal female karyotype of 46XX and other routine blood investigations were reported as normal results.

DISCUSSION

Congenital arhinia is one of the rare defect that occurred during early embryogenesis. Worldwide, there are less than 40 published cases of congenital arhinia and our patient is the first reported case in Malaysia.

The development of nose and nasal cavity occurs between third to tenth weeks of gestation. Rapid differentiation and division occurred 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 1) failure of medial and lateral nasal processes to grow, 2) premature fusion of the nasal medial processes, 3) lack of resorption of the nasal epithelial plugs.

From literature review, congenital arhinia is not associated with any specific genetic mutation. Most of the patients with congenital arhinia have normal chromosomal analysis which is similar in our patient, except for few with abnormal karyotyping.[2] 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, syndactyly, and hypospadias.[3] For patient with multiple congenital

This article was accepted: 14 May 2017
Corresponding Author: Ng Rui Lun
Email: rolandnrl@yahoo.com

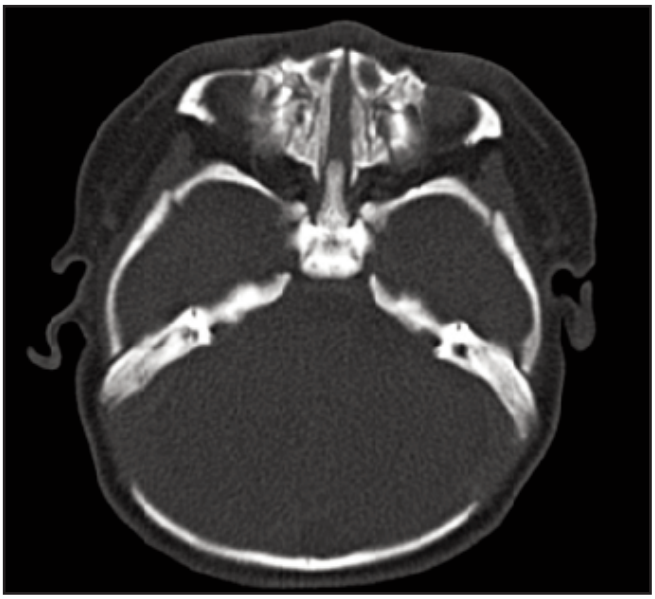

Fig. 1: Axial CT image showing narrowed nasal cavity with absence of the nasal septum and stenosis of the pyriform aperture and bilateral choanal.

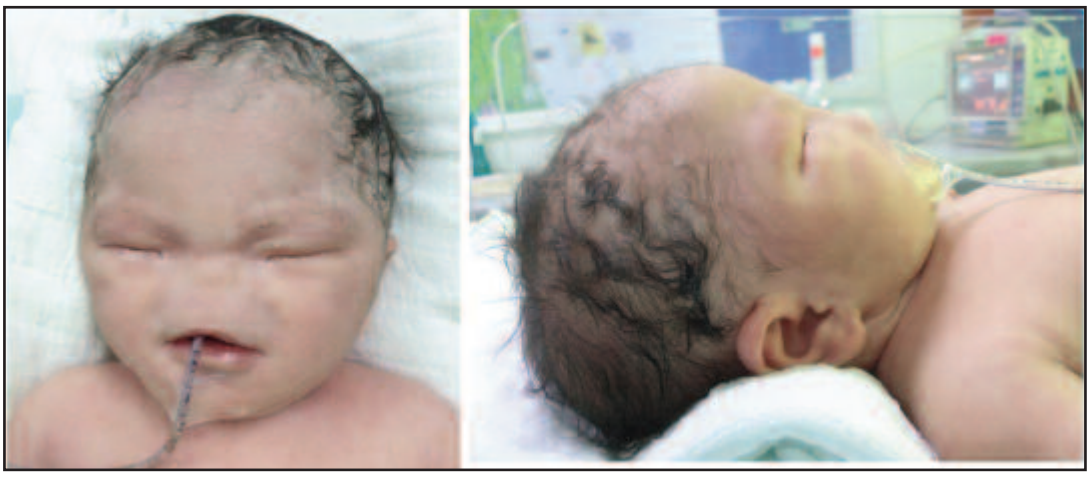

Fig. 2: Clinical photographs of patient showing small longitudinal ridge-like protuberance in midface with absence external nostrils.

anomalies, a multi-disciplinary team involvement is needed in the care of the patient. It is important to involve clinical geneticist to give opinion if patient with congenital arhinia has association with any clinical syndrome. This information is vital when counselling parents for recurrence risk of similar defect in future child-bearing.

The most worried immediate consequence when dealing with patients with congenital arhinia is severe airway obstruction. However, there was reported case of patient with congenital arhinia who did not require breathing support after birth and managed to adapt to mouth breathing.[4] This is similar in our patient where she did not require any breathing assistance immediately after birth and she could maintain normal saturation under room air. Tracheostomy was performed electively in our patient as our hospital is a district hospital with only a visiting paediatric otorhinolaryngologist in service. Feeding for patient with congenital arhinia is also a huge challenge for the treating paediatrician as both the breathing and feeding share the common route. However, feeding can be given via orogastric route which bypasses the shared pathway until definitive airway is performed.

Most of the patients with congenital arhinia were diagnosed only after delivery except for five reported cases of isolated arhinia which were detected during antenatal scan. All the cases were detected in second trimester, ranging from 23 to 29 weeks.[5] Among the features seen during scan which are suggestive of arhinia includes midfacial hypoplasia and absence of midfacial opening or nares. Systematic approach of first trimester scan may help in early recognition of arhinia. Prenatal diagnosis of arhinia is crucial for parents and clinicians to plan the delivery and care afterwards. However, chances of detecting arhinia during antenatal scan would largely depend on ultrasonographers who are skilful in detecting the ultrasound features and sonography machines that can provide good images, i.e., 3D or HD images.

Long term prognosis for our patient is uncertain as there are limited local experience in managing patient with congenital arhinia. Literature review showed that patient with arhinia can have successful facial reconstructive surgery and later lead normal life after surgery. The timing for reconstruction of nose and nasal cavities is usually by pre-school age when the facial development is complete. Prior to corrective surgery, patient with arhinia will benefit from multi-disciplinary management which include continuous growth and developmental assessment, ophthalmology review, hearing assessment and geneticist review. In the immediate future, her caregiver will have to master the skill of caring for tracheostomy and feeding via orogastric tube.

CONCLUSION

It may not be easy to detect arhinia during antenatal scan as likelihood of detecting arhinia depends on many factors. However, we hope to use this case to highlight that patients with congenital arhinia may adapt to breathing through mouth and thus bypass intubation or tracheostomy in acute setting despite popular belief that newborns are obligate nasal breather.

REFERENCES

1. Albernaz VS, Castillo M, Mukherji SK, Ihmeidan IH. Congenital arhinia. AJNR Am J Neuroradiol 1996; 17: 1312-4.
2. Olsen QE, Gjelland K, Reigstad H, Rosendahl K. Congenital absence of the nose: a case report and literature review. Pediatr Radiol 2001; 31: 225-32.
3. Biswas KD, Dutta S, Ghatak S, Sinha R. Congenital complete arhinia: Report of a rare case. IJSS Case Reports & Reviews 2014; 1(6): 8-10.
4. Cole RR, Myer CM, Bratcher GO. Congenital absence of the nose: a case report. J Pediatr Otorhinolaryngol 1989; 17: 171-7.
5. Leroy D, Slachmuylder E, Popijn M, Cassart M, Massez A, D'Haene N et al. Antenatal diagnosis of isolated total arhinia in the second trimester of pregnancy. Open J Obstet Gynecol 2016; 6: 419-23.