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Original Research Paper

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# A CASE OF BOSMA ARHINIA MICROPHTHALMIA SYNDROME FOR NASAL RECONSTRUCTION

**Dr. Arathy. P** — Junior Resident, Department of Anaesthesiology and critical Care, KEM Hospital

**Dr. Yogita Patil** — Additional Professor, Department of Anaesthesiology and Critical Care, KEM Hospital

**ABSTRACT**

Bosma arhinia microphthalmia syndrome is a rare disorder, the incidence of which is thought to be less than 50 (1). It is associated with missense mutations in epigenetic regulator SMCHD1 gene (2). Due to the facial structural anomalies, the patients can often present with difficulty in feeding and breathing. Our patient presented with hypoplastic left eye, absent left nostril, also diagnosed with autism disorder. There were no other complaints or systemic ailments. He was posted for nasal reconstruction, with forehead flap and had presented for preanaesthetic evaluation. All lab investigations were found to be normal. Keeping in mind the risk of encountering difficulty in securing the airway, the case was induced under general anaesthesia, with the patient being intubated and maintained on inhalational anaesthetic during the course of surgery. Post operatively, after ensuring adequate respiratory efforts, he was extubated. Post-operative recovery period was uneventful.

**KEYWORDS :**

## INTRODUCTION

Bosma arhinia microphthalmia syndrome is a rare condition characterized by

1) Congenital arhinia
2) Ocular malformation
3) Sexual maturation dysfunction (3)

There have been less than a hundred patients reported world wide. The most common etiology is the spontaneous mutation of SMCHD1 gene in either the egg or the sperm.

There are various degrees of manifestation of the syndrome, which could range from anosmia, to complete arhinia. These children may require tracheostomy in early childhood to enable breathing during feeding. They also may require multiple surgical interventions in life.

## CASE PRESENTATION

A 4year old male child, a known case of Bosma arhinia microphthalmia syndrome, with autism spectrum disorder, presented to preoperative anaesthesia assessment outpatient clinic. He was posted for nasal reconstruction with forehead flap.

The mother stated that the child was born at 39 weeks of gestation via Lower Segment Caesarean Section. He was noted to have a partially absent nose, with small, widely set eyes. He was kept in Neonatal ICU admission for 5 days for observation. He did not have any difficulty feeding or breathing. He was not found to have any other systemic illnesses. The mother did not suffer from any chronic illness, nor did she give history of any significant illness during pregnancy. She had received appropriate prenatal care and was unaware of the condition of the foetus during pregnancy. The mother was a housewife, and father, an engineer. There was no significant family history or history of similar illness in the family. The patient had delayed development of milestones, but all milestones had currently been achieved for age. He was fully immunized for age.

There was history of left dacryocystorhinostomy, done at 1 year of age, details of which were unavailable.

On examination, the child was active and playful. Facial asymmetry was noted. He was found to have wide set eyes. Left eye was significantly smaller and poorly developed when compared to right. Right nostril was absent. Bilateral ears were prominent. Micropenis was noted, with absent right testis.

Mouth opening was more than 3 fingers, with normal palate and dentition. Mallampatti score was 1. Neck extension was not restricted.

He was vitally stable, with normal systemic examination. All lab reports were found to be within normal range

Perioperative management

We report a case of BAM syndrome in a 4year old boy, with left arhinia, left microphthalmia, and in the autism spectrum for nasal reconstruction with forehead flap.

On the morning of the surgery, anaesthesia machine was checked, and difficult airway cart was kept ready, comprising of endotracheal tubes of various sizes, oral airways, masks of different sizes, direct laryngoscope, video laryngoscope, and laryngeal mask airways.

After ensuring adequate starvation, the child was wheeled into the operation theatre, where standard ASA monitors were attached. He was adequately preoxygenated for 3 minutes. Induction was done with Inj. Fentanyl 30mcg, Inj. Propofol 30mg, Inj rocuronium 15mgIV.

Intubation was done with mac 2 blade under direct laryngoscopy, and 5mm cuffed South Pole tube was inserted. Anaesthesia was maintained with sevoflurane, maintaining a target mac of 1.0. Post procedure, after ensuring adequate respiratory effort, he was extubated. Immediately post extubation, he had an episode of breath holding, which was managed with mask holding with positive pressure ventilation with 100% oxygen. Postoperative recovery was uneventful.

## DISCUSSION

Bosma Arrhinia Microphthalmia syndrome is characterized by spontaneous mutation in the SMCHD gene, either paternal or maternal in origin.

Depending on the severity of the condition, there can be complete lack of external nares. But neonates are obligate nasal breathers. Besides this, they will be unable to breathe while feeding. In such situations, these children are often tracheostomized in early life.

Craniofacial Dysfunction, with hypoplastic maxilla, high arched palate can all lead to difficult intubation. Difficult

airway cart is to be kept ready, with oral airways, videolaryngoscope, bougie, fibreopticbronchoscope, supraglottic airway devices. Emergency surgical interventions should also be kept ready for cricothyrotomy or tracheostomy

Antisialogogue medication should be administered and child should be anaesthetized maintaining spontaneous ventilation Intubation can be performed under deep inhalational anaesthesia, or with intravenous induction agents, taking care to preserve spontaneous ventilation. It is safer to avoid skeletal muscle relaxants at induction as it may result in cant ventilate can't intubate situation, necessitating establishment of surgical airway rapidly

## CONCLUSION

Bam syndrome is characterized by facial deformities that can lead to difficult airway. As the airway is shared, there is risk of kinking of tube or tube disconnection. Thus, good discussion and planning is imperative to a successful outcome.

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