MULTIDISCIPLINARY APPROACH OF NEONATAL INTENSIVE CARE IN BAM SYNDROME: A RARE CASE REPORT

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pISSN: 2828-4070
Proceedings ICE on IMERI. 2024.
Received: December 1st, 2024
Accepted: December 30th, 2024
Published online: January 17th , 2025

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Abstract

Bosma Arhinia Microphthalmia Syndrome (BAMS) is a very rare condition characterized by eye defects, a complete absence of nose, and hypogonadotropic hypogonadism, which is caused by a genetic mutation in the gene SMCHD1 located in chromosome 18p11. Fewer than 100 cases were reported globally over the previous century, and only a few clinical studies have discussed its occurrence and management in Indonesia. Therefore, there are no clear guidelines about the management of neonates with BAMS due to its rarity. This study reported a BAMS neonatal patient with atresia choana, microphthalmia, pectus excavatum, facial dysmorphic, unspecified hearing loss, and hydronephrosis. An MSCT scan of the patient revealed an absence of cavum nasi, undeveloped paranasal sinus, microcephaly, abnormal size of bulbus oculi sinistra, and the presence of cleft palate. This condition prompted the patient to require intensive medical support early in life due to breathing and feeding difficulties, as well as the need for several examinations on many aspects to determine the extent of the syndrome which has different characteristics for each individual. Therefore, a multidisciplinary approach is necessary to provide the most suitable management for each neonate with BAMS. This case report described the patient's neonatal intensive care and management with a multidisciplinary team which includes a neonatologist, an ophthalmologist, an otorhinolaryngologist, a radiologist, an endocrinologist, and an oral and maxillofacial surgeon. This study aims to improve the knowledge of BAMS patient management in the future since there are no standardized guidelines or treatment protocols, and the case is rarely studied.

Keywords

Bosma arhinia microphthalmia syndrome, case report, multidisciplinary team, neonatal intensive care, rare disease

Introduction

Bosma arhinia microphthalmia syndrome (BAMS) is a rare syndrome described with arhinia, microphthalmia, and hypogonadism. It has only been reported in around 100 cases in the medical literature. Since it was first reported in 1972, there have been no guidelines, and there are still only a few studies discussing the care of neonates with BAMS due to its rarity. The existing medical literature described more about the findings of BAMS itself than the intensive care received by neonates with BAMS. This case report described the care received by neonates with BAMS while in the NICU of Sardjito General Hospital from admission into the NICU until discharge and the collaboration between multiple disciplines in handling this case. This case report was created to provide a new perspective on the care of neonates with BAMS through a multidisciplinary approach in neonatal intensive care, which may improve the understanding of its management.

Case Report

A 19-day-old female baby who was born to a 23-year-old mother P1A0 at the 36th week of gestational age by vaginal delivery was referred from Wonosobo Regional Hospital to Sardjito General Hospital to receive intensive care and tracking regarding the presence of Bosma syndrome. The patient was born without a nose and could not open both eyes. The patient did not cry immediately at birth so resuscitation had to be carried out until the intubation stage. The patient's family reported no pre-natal and ante-natal care history of abnormalities. The patient's birth weight was 2,565 grams, birth length was 43 cm, and head circumference was 29 cm.The patient was then treated in the NICU with a ventilator, until just before being referred.

The patient was treated for 22 days in the NICU at Sardjito General Hospital under the supervision of a pediatric neonatologist. On physical examination, there were findings of facies dysmorphic, choana atresia, microphthalmia, pectus excavatum, and arhinia (**Figure 1**). From the 4th day to the 12th day of treatment, the patient was given a McGovern Nipple. The patient also experienced feeding difficulties so food intake had to be provided through a feeding tube. In treating this patient, the neonatologist collaborates with a radiologist, an oral and maxillofacial surgeon, an otorhinolaryngologist, and an ophthalmologist.

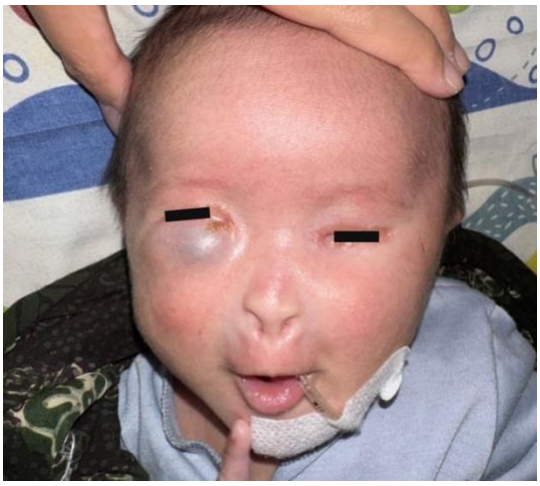

Figure 1. Facies dysmorphic, atresia choana, microphthalmia, and arhinia

The head MSCT scan revealed a cleft palate, absence of nasal cavity, and undeveloped paranasal sinuses. Bilateral bulbus occuli were seen, with the left bulbus occuli being smaller than the right bulbus occuli. Bilateral intraocular lenses were absent. Dilation of the subarachnoid space in the left frontotemporal region was noted, with microcalcification in bilateral hemispheres. In the abdominal ultrasonography; pelviectasis was found in both kidneys, but the urinary bladder was normal (**Figure 2**). Leukocyte esterase and bacteria were detected in the routine urine examination, indicating urinary tract infection and the patient received antibiotic therapy for 14 days. The peripheral blood smear test detected an inflammation or infection and hypersensitivity reaction.

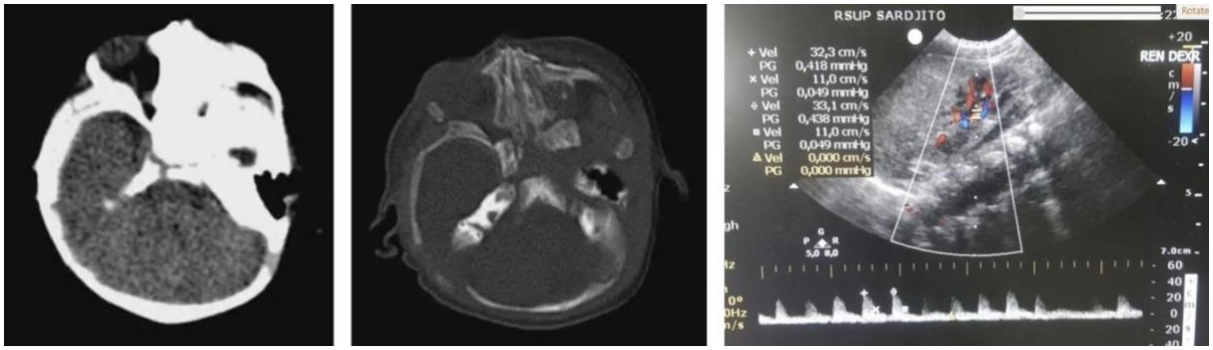

Figure 2. Head MSCT scan without contrast (transverse section) and USG results

Since this patient experienced breathing difficulties, the oral and maxillofacial surgeons suggested using the McGovern Nipple to maintain airway patency. On the ninth-day follow-up, the patient was given a trial to breathe without McGovern Nipple aid and was breathing fine with her mouth. The patient's oral intake was good and the patient's body weight increased.

The patient also underwent an indirect fundoscopic examination. The clinical findings were that both eyes had no light perception, produced secretions, and did not have lenses. On the lower palpebral of the right eye, there's a purplish rubbery mass with a distinct border. The ophthalmologist diagnosed ODS microphthalmia, conjunctivitis ophthalmia neonatorum, and OD suspected hemangioma. The patient was given antibiotic eye drops and warm water compresses on the right palpebral mass.

This patient also underwent a transnasal endoscopic examination which showed hypoplasia of the nasal alae, atresia of the nares, and palatoschisis. During the hearing test, it was found that the patient had unspecified hearing loss in both ears. Regarding the condition of the undeveloped nasal cavity and paranasal sinuses, the otolaryngologist recommends conservative treatment until the patient is three months old, after which reconstructive surgery can be considered.

Discussion

Neonates with BAMS have several abnormalities that need to be taken care of comprehensively. Therefore, a comprehensive evaluation conducted by a multidisciplinary team approach consisting of multiple specialists related to their symptoms and phenotypic traits is needed to handle such cases.

BAM Syndrome is an extremely rare syndrome caused by a genetic mutation in the gene SMCHD1 located in chromosome 18p11.[1] BAMS causes a manifestation of malformations during frontonasal processes associated with anomalies of the skull, face, and brain.[1] Patients with BAM syndrome are reported to have three cardinal signs such as arhinia, microphthalmia, and hypogonadism.[2] In this case, the patient had distinguishable BAMS features: choanal atresia, microphthalmia, and a cleft palate.

Ocular malformations in BAMS can manifest with or without coloboma, cataract formation, anophthalmia or microphthalmia, and nasolacrimal duct obstruction.[3] Anophthalmia or microphthalmia in BAMS could cause the patient to have a decrease up

to complete vision loss. These ocular manifestations could occur due to crystalline lens and ocular lens structural defects.[4] Diagnostic imaging such as CT or MRI may reveal features like asymmetric microphthalmia or coloboma, which may be evident in the iris, retina, choroid, or optic disc. In this case, the patient's head CT scan showed asymmetric microphthalmia and the absence of a lens in both eyes, confirmed by indirect fundoscopy. In managing BAMS, an ophthalmologist plays a role in the early detection of the patient's eye abnormalities and subsequent treatment. If microphthalmia is managed early, it can be treated by early socket expansion which is essential in reducing facial deformity.[5]

Arhinia in BAMS could range from hypoplastic nose to complete arhinia which may lead to feeding problems and respiratory distress since neonates are nasal breathers by default and only learn the reflex of mouth breathing by six months old.[1] Therefore, immediate airway patency management and feeding tubes are required in neonates with BAMS. In this case, McGovern nipple is the preferred breathing aid considering the patient's condition of bilateral choanal atresia and was used for eight days. McGovern nipple is a technique that is done by cutting off the end of an intraoral bottle nipple, thus leaving its end with a large opening. To help with the feeding problem, a feeding tube could be placed through the McGovern nipple or beside the McGovern nipple.[6] In this case, the feeding tube is placed next to the McGovern nipple. Prenatal diagnosis of arhinia is possible to screen with the aid of fetal MRI or ultrasound as a flat fetal profile as early as the 23rd gestational week.

BAMS is also significantly associated with hypogonadotropic hypogonadism which may cause delayed puberty. Abnormal reproductive system development or major developmental defects are more likely to cause psychological problems in the future. Affected males in BAMS may have underdeveloped reproductive tissues, cryptorchidism, and inguinal hernias. However, in females, the problem with the reproductive system usually becomes apparent in their adolescent years.[7] The patient in this case had normal genitalia and endocrinology laboratory workup results. Hence, the patient needs regular follow-up with an endocrinologist up to puberty to detect any problems early.

Surgical reconstruction is still an issue in studies. However, reconstruction is preferably delayed until the age of pre-school, when facial development is nearly completed.[8] There are three suggested times for nasal reconstruction: early reconstruction (neonatal); preschool or school-age; and adolescence. The indication in

infants born with facial dysmorphic and arhinia to undergo earlier surgery was respiratory distress and feeding issues. Hence, immediate management such as oral airway, tracheostomy, or surgical creation of nasal airway for life-threatening respiratory distress was recommended in the neonatal period.[9] In this case, the otorhinolaryngologist suggested conservative management and reconstructive surgery to be done after the patient is three months old.

BAMS is an extremely rare syndrome with cardinal manifestations including microphthalmia, arhinia, and hypogonadism. These manifestations of BAMS could compromise the patient's life and reduce their quality of life in the future which can be alleviated by comprehensive early management. In this case report, the patient is presented with cardinal signs of BAMS. This patient is managed for 22 days in NICU with a multidisciplinary approach which leads to the most appropriate clinical decision that progressively improves the condition of the patient until discharge. This study shows that a standardized guideline which includes multidisciplinary approaches in managing neonates with BAMS need to be established in order to provide a systemized algorithmic approach.

Competing Interests

The authors have no conflict of interest to declare.

Acknowledgments

This article was presented at the 9th International Conference and Exhibition on Indonesian Medical Education and Research Institute 2024, Faculty of Medicine, Universitas Indonesia. We are grateful for the outstanding assistance provided by the committee of The 9th ICE on IMERI 2024 throughout the process.

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