Case Report

Bilateral Extreme Microphthalmia in a Newborn: A Rare Case Report from Bangladesh

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Abstract

Extreme microphthalmia and anophthalmia are rare congenital ocular anomalies that may occur as isolated defects or in association with systemic abnormalities. We report a rare case of bilateral extreme microphthalmia in a full-term male newborn delivered at President Abdul Hamid Medical College Hospital, Kishoreganj, Bangladesh. The baby presented with sunken, deep-seated orbits and absent palpable globes since birth. There was no history of maternal infection, teratogenic exposure, or familial congenital anomalies. Systemic evaluation including TORCH screening, chest X-ray, abdominal ultrasonography and echocardiography revealed no abnormalities. Computed tomography confirmed poor development of the globes and orbits with a normally developed brain. The patient was referred for further management. This case highlights the importance of early neonatal ocular examination, advanced imaging and the need for improved antenatal diagnostic facilities in resource-limited settings. To our knowledge, this is one of the few documented cases of bilateral extreme microphthalmia reported from Bangladesh.

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Introduction

Extreme microphthalmia or anophthalmia are rare congenital anomalies that may present isolated or with systemic manifestations like central nervous system and musculoskeletal or cardiac abnormalities[1]. It can be classified as true, extreme microphthalmos and clinical anophthalmia. Classification based on causes is primary, where there is no development of the eye; secondary, where development of the eye starts but does not progress later on. It arises from a genetic mutation in the sox[2] gene on either a single gene or an entire chromosome. An entire chromosome may be absent or duplicated or undergo translocation, transferring a chromosome segment to a different chromosome. These may be associated with corneal sclerosis, cataracts, optic disc anomalies, mental retardations, facial dysmorphisms, etc. Anophthalmia diagnosis is primarily clinical, using universal ocular examination and imaging, such as a three-dimensional ultrasonogram and computed tomography (CT)/ magnetic resonance imaging (MRI)[2]. Genetic study is also helpful for diagnosis. The prevalence of anophthalmia is about 3-14 per 100,000 population[3,4]. In the United Kingdom, the prevalence is 1 per 100,000 live births[5].

Case Report

Amale full-term newborn was delivered by normal vaginal delivery at the Department of Obstetrics and Gynecology, President Abdul Hamid Medical College Hospital, Kishoreganj, on 1st December 2021. The baby was referred for ophthalmic evaluation immediately after birth due to the presence of abnormally sunken eyes that had not opened. The mother was a 29-year-old multipara with an otherwise uneventful antenatal period. This was her third pregnancy. She had regular antenatal checkups at a local union sub-center located approximately 20 kilometers from the Upazila health complex. There was no history of maternal fever, rash, teratogenic drug intake, radiation exposure, tobacco use, or alcohol consumption during pregnancy. She received only iron and calcium supplementation. There was no family history of congenital ocular anomalies, neurological disorders, or genetic syndromes. Her first living child was healthy. She had a history of spontaneous abortion six months prior to the index pregnancy following a fall. On examination, the newborn weighed 2600 grams. Both orbits appeared sunken and deep seated. The palpebral fissures

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were extremely small and a tiny cystic mass was observed on the inner surface of both lower eyelids. The eyeballs could not be palpated. No facial dysmorphism, neurological deficit, or motor abnormality was detected.

Investigations

Serological screening for intrauterine infections including rubella, toxoplasmosis and cytomegalovirus were negative. Chest X-ray and whole abdominal ultrasonography revealed no abnormalities. Echocardiography showed a structurally normal heart.scan of the orbitand brain revealed grosslywhile intracranial structures were normal.Magnetic resonance imaging (MRI)could not be performed as the parents declined further investigations.

Management and Referral

After confirmation of the diagnosis of bilateral extreme microphthalmia, the parents were counseled regarding the nature of the disease, prognosis,andlong-term management options.The patient wasreferredto theNational Instituteof Ophthalmologyand Hospital, Dhaka, for further evaluation, visual rehabilitation planning, socket development and possible future prosthetic rehabilitation.

Discussion

Bilateral extreme microphthalmia is a rare congenital ocular anomaly characterized by severely underdeveloped globes in both orbits and is often considered within the anophthalmia-microphthalmia spectrum. The reported global prevalence ranges from 3 to 14 per 100,000 population, while in the United Kingdom it is approximately 1 per 100,000 live birth[3,4,5]. The bilateral form is more frequently associated with systemic abnormalities and genetic mutations compared to unilateral cases. In our case, the newbornpresented with bilateral extreme microphthalmia without any detectable systemic, neurological, or cardiac anomalies. This is noteworthy because bilateral cases commonly show associations with central nervous system defects, facial dysmorphism and chromosomal abnormalities[1]. The absence of systemic involvement in this patient suggests a sporadic isolated presentation, which is less commonly reported in the literature. Genetic mutations, particularly involving the SOX2 gene, have been identified as one of the major etiological factors in bilateral anophthalmia and severe microphthalmia1. However, due to the unavailability of genetic testing in our setting and refusal of further investigations by the parents, genetic confirmation could not be performed. Additionally, the mother had no history of teratogenic drug intake, TORCH infections, irradiation exposure, or significant systemic illness during pregnancy. TORCH screening was also negative, which excludes common infectious causes associated with congenital ocular malformations. Early antenatal diagnosis of microphthalmia is possible with high-resolution ultrasonography and three-dimensional imaging techniques[2]. In resource-limited settings like rural Bangladesh, such advanced diagnostic facilities are often inaccessible, leading to delayed or missed antenatal detection, as seen in this case. Postnatal imaging with CT scan confirmed poor development of the globesand orbits, while the brain was normally developed. Although MRI provides better soft-tissue detail, it could not be performed due to parental refusal. This case highlights the importance of routine neonatal ocular examination by trained ophthalmologists, especially in peripheral hospitals, for early diagnosis of rare congenital anomalies. Early identification allows timely referral, visual rehabilitation, socket reconstruction planning and parental counseling, which are crucial for long-term functional and cosmetic outcomes. To the best of our knowledge, this appears to be one of the very few documented cases of bilateral extreme microphthalmia reported from Bangladesh. Reporting such rare presentations is important to improve local epidemiological data, raise clinical awareness and emphasize the need for strengthening antenatal screening and postnatal ophthalmic evaluation services in low-resource settings.

Conclusion

Bilateral extreme microphthalmia is a rare congenital condition that may present either as an isolated anomaly or in association with systemic defects. Early diagnosis is essential for timely intervention, visual rehabilitation planning and parental counseling. Advanced imaging such as ultrasound, CT and MRI plays a crucial role in confirming the diagnosis. Strengthening antenatal screening programs and ensuring routine

postnatalocular examinations, especially in peripheralhealthcare settings, can significantly improve early detection and management outcomes. Reporting such rare cases contributes to national epidemiological data and increases clinical awareness among healthcare professionals.

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