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A Case Report

A Rare case of Congenital Bilateral Anophthalmia in Irrua: A Case Report.

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Abstract:

Introduction: Anophthalmia is a rare congenital malformation characterized by absence of the eye globes which may be either unilateral or bilateral. It may occur in isolation or in association with other Congenital anomalies.
Although its diagnosis can be made clinically, imaging can play a crucial role in confirming this condition and also screen for associated anomalies.
Objectives: To report a case of congenital bilateral anophthalmia in a one-week-old neonate.
To highlight the role of diagnostic imaging, in particular ultrasonography in evaluating a patient with anophthalmia.
To describe the notable ultrasound findings and review relevant existing literature on this rare condition.

Case Report: The case of a one week old female neonate who was delivered at term in a private medical facility by a Para-3 middle aged petty trader and referred by the attendant physician for imaging at Irrua Specialist Teaching Hospital, after being suspicious of an ocular malformation due to persistent closure of the neonate's eye lids is reported and the findings on sonography are also described. Sonography revealed bilateral absence of the eye with hypoplasia of the orbital cavity. No other anomaly was demonstrable.

Conclusion: Anophthalmia is a rare Congenital ocular malformation. The role of imaging is crucial. Particularly in resource poor settings where advanced imaging modalities like magnetic resonance imaging are unavailable, ultrasonography can be used for confirmation of the anomaly and also to screen for associations.
Keywords: Anophthalmia, ultrasonography, Magnetic Resonance imaging, Irrua.

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Introduction

Anophthalmia is a severe form of ocular malformation characterized by complete absence of the eye globes.1,2,3,4 It is a rare entity which can occur either in isolation or in association with other Congenital malformations especially involving the central nervous system, cardiac, musculoskeletal and renal system.1

The aetiology of anophthalmia is complex and not fully understood. However, an interplay between genetic and environmental factors has been postulated.

Some environmental factors have been implicated in the aetiology. Examples include cytomegalovirus infection, toxoplasmosis, rubella and parvovirus B19 infection etc. Nutritional deficiency such as vitamin A, and exposure to toxic agents like alcohol, thalidomide and warfarin have also been implicated.1,3,4

Mutation in certain genes such as SOX2, OTX2, PAX6, STRA6, etc, have been implicated in the pathogenesis of anophthalmia.5,6
The eye is formed via the differentiation of tissue structures derived from neuroectoderm, neural crest cells, surface ectoderm and mesoderm. It is also important to note that embryologic development of the optic tissues typically begins at about the 4th week of development with the closure of the rostral neuropore of the neural tube. Failure or any alteration in the stages of differentiation results in ocular malformation.7

Anophthalmia is a rare ocular malformation whose global prevalence is reported to be about 3 per 100,000 population. This is far less common when compared to microphthalmia whose global prevalence has also been reported to be about 14 per 100,000.[8,9] In another earlier study done in south-Eastern Nigeria by Chukaokosa *etal*,[10] over a period of eight years, the prevalence was observed to be about 9.3% of all congenital ocular malformations.

Case Report

We report a case of a one week old female neonate who was delivered at 39 weeks gestational age by a Para-3 middle aged petty trader in a private medical facility, and was referred by the attending physician to the radiology department of Irrua Specialist Teaching Hospital for imaging, after the neonate's persistently apposed eye lids and inability of the physician to palpate either globe underneath the eyelids which raised his suspicion of a congenital ocular anomaly. The mother had attended her antenatal clinic regularly and she also claimed to have been compliant with her routine prenatal multivitamins. There is no family history of congenital ocular diseases or similar presentation in prior siblings. The mother had an antenatal scan done at the second trimester which showed no obvious fetal anomaly. The antenatal period was essentially uneventful. The neonate was delivered via the vaginal route and was reported to have cried immediately after birth. The physician's observation during examination of the neonate necessitated their referral for ocular sonography.

Both orbits were scanned using a high frequency (7-12MHz) transducer placed over each orbit in turn, after application of a thin film of ultrasonic gel.

Sonography revealed empty and hypoplastic (small sized) orbital cavities with bilateral absence of the globes. A further peep into the cranium via the anterior trans-fontanelle route revealed normal cerebral cortical convolutions and parenchymal echogenicity. The corpus callosum showed normal size and outline, with no demonstrable evidence of agenesis. Both lateral, third and fourth ventricles were within normal limits.

An impression of congenital bilateral anophthalmia was made. The neonate's abdomen was also scanned for abnormalities, and the viscerals were normal. The parents were then referred to the ophthalmology department for specialist care.

Most of the few reported cases in developed countries of the world have employed a spectrum of hitech imaging modalities for confirmation of the diagnosis of anophthalmia. In our own environment (Sub-Saharan Africa) wherethis case is being reported from, there is paucity of hitech modalities like magnetic resonance imaging, as opposed to

ultrasonography which is the primarily available modality. Ultrasonography is also cheap and non-ionizing, with good soft tissue resolution, hence it was used primarily in imaging the neonate's orbit, cranium and abdomen.

Images acquired are shown in figs. 1, 2 and 3 below.

Fig 1 is a photograph of the neonate showing the closed eyelids bilaterally.

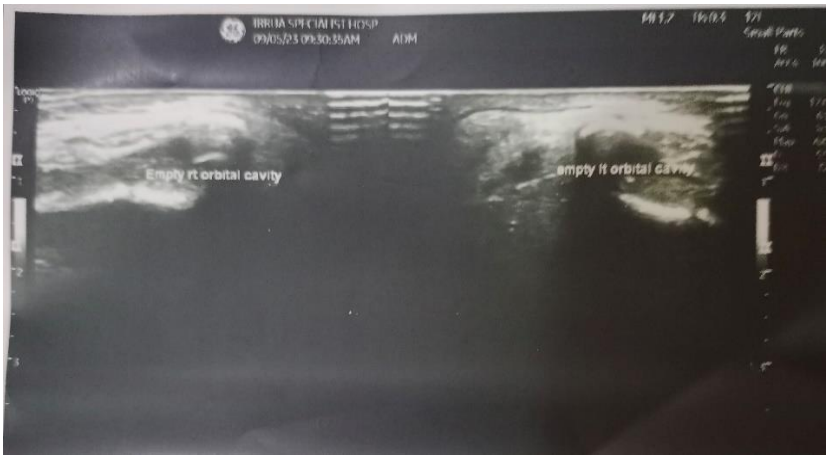

Fig 2 is a transverse sonogram of both orbits showing absence of the eye globes in each orbital cavity. Hypoplasia of the cavities is also noted.

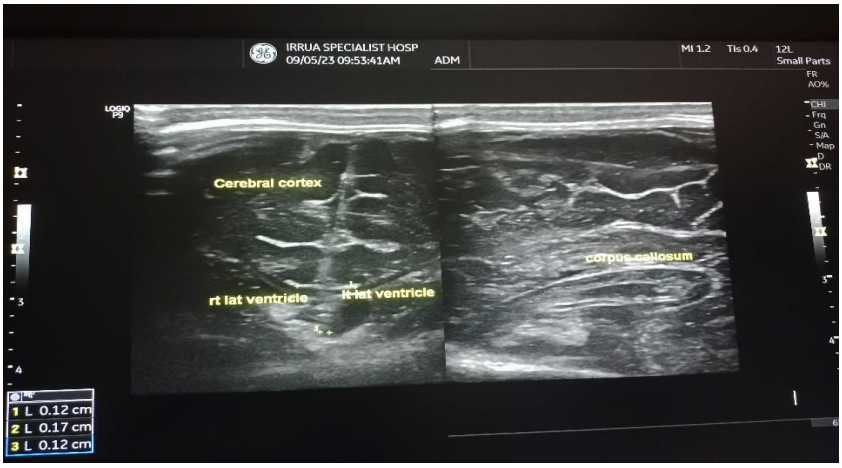

Fig 3 depicts sonograms of the neonatal brain in the coronal and sagittal plane. The brain cortical mantle, cerebral convolutions, corpus callosum and ventricles are within normal limits.

Discussion

Although anophthalmia is rare condition which might present either as a unilateral or bilateral defect, oftentimes, it is bilateral. Some authors have reported bilateral anophthalmia in the past.[1,2] This currently reported case closely mirrors that earlier reported by Meliha *etal*[2] in Bosnia, in that the newborn are both females, born at term to middle aged parents via the vaginal route. Both mothers had uneventful antenatal periods, and without history of the use of illicit drugs, alcohol or tobacco.

Anophthalmia is rarely detected by prenatal ultrasonography, most especially in the absence of other coexisting congenital anomalies.[11] This reported case was detected after delivery of the fetus, and there were no other coexisting anomalies. Ultrasonography being an operator-dependent modality which relies principally on the skill of the user, may partly account for the reason why the defect was not detected intrapartum, as it was revealed that the only antenatal scan which the patient did, wasn't done by a radiologist. The absence of history of ocular diseases in prior siblings, other first degree relatives, as well as the absence of obvious congenital anomalies in large and major organs of the body, may reduce the suspicion of a malformation in smaller fetal organs like the eye balls. However, it is practical knowledge that congenital fetal anomalies may occur in associations, thus, it is advisable to search for other anomalies during imaging.

Anophthalmia may be associated with variable degrees of septo-optic dysplasias (combination of optic nerve dysplasia and absence of septum pellucidum), as has previously been reported by Manisha *etal*.[12] In this case, ocular sonogram revealed absence (agenesis) of both optic nerves, as well as hypoplasia of the orbital cavity. The visualized intracranial structures were sonographically normal.

Conclusion

Anophthalmia is a rare congenital ocular malformation. The role of imaging is crucial for its assessment. In resource-poor settings where advanced imaging modalities like magnetic resonance imaging are unavailable, ultrasonography can be used for confirmation of the anomaly, and also to screen for associations.

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