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Bilateral Anophthalmia: Literature, Review and Case Report

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Authors' contributions

This work was carried out in collaboration among all authors. Author LEYI wrote the first draft of the manuscript. Authors BAAH and ARN reviewed it. Author ARN contributed to literature search. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

Introduction: Anophthalmia which is the congenital absence of the eyes could be bilateral or congenital, it occurs globally although it is rare. It is usually in association with other systemic congenital abnormalities.
Case Report: A case of a female baby delivered at a primary health center in Port Harcourt, Nigeria who had bilateral anophthalmia is reported here along with review of literature Magnetic Resonant Imaging (MRI) of brain and orbit was used to confirm the diagnosis which showed absence of both globes, with hypoplasia of the orbits as well as the extraocular muscles, the optic nerves could not be differentiated from the visualized intra-orbital structures. All other systems were essentially normal. This is an exceptional case in Port Harcourt, Nigeria, as literature search shows that no other case has been reported in Port Harcourt. She is currently being followed up at the paediatric, ophthalmology and ENT clinics of the University of Port Harcourt teaching hospital.

Keywords: Absence of both eyes; anophthalmia; bilateral; blind child; congenital.

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1. INTRODUCTION

Anophthalmia, (Greek: ανόφθαλμος, "without eye"), is the medical term for the absence of one or both eyes [1]. Both the globe (human eye) and the ocular tissue are missing from the orbit [2] Anophthalmia therefore is defined as the absence of ocular tissue in the orbit, it can affect one or both eyes. It can occur alone or in association with other abnormalities constituting a syndrome. More often than not it occurs in company of malformations in other systems of the body especially involving the cardiac, central nervous and the musculoskeletal system [3]. Generally Diagnosis of anophthalmia is clinical, after a comprehensive ophthalmologic examination. The comprehensive ophthalmo-logical examination demonstrates the absence of the whole globe [4]. Aetiology of Anophthalmia/ microphthalmia is multifaceted, it could be monogenic, chromosomal and environmental. Chromosomal abnormalities could be translocation, deletions or duplications. Of the monogenic causes only SOX2 has been recognized as a main causative gene [3] others are RBP4, OTX2, CHX10, RAX and BMP4 [2]. Anophthalmia and microphthalmia prevalence at birth has been commonly predicted to be 3 per 100,000 population [5] while in Scotland microphthalmia anophthalmia and coloboma gave a birth prevalence of 1.9/10000 based on total live births during the study period [6]. The capture- recapture analysis suggested that the true birth prevalence lies between 2.4/10000-3.5/10000 [6]. In Northeast Italy, over a period of 8 years, 22 cases were reported amongst three hundred and sixty-eight thousand two hundred and fifty-six deliveries, giving a birth prevalence of 0.60 per 10,000 [7]. A prevalence of 1.0 per 10000 has been reported in United Kingdom [8]. A case of bilateral anophthalmia was reported in Tuzla, Bosnia [4]. In Benin city, Nigeria two cases were reported over a period of 20years giving a prevalence of 0.2-0.4 per 10,000 live births [9]. In Enugu South eastern Nigeria, over a period of 8 years anophthalmia was said to cause 9.3% of congenital eye anomalies [10]. In Kano state Northern Nigeria there has been a report of one case [11]. Anophthalmia often times occur in association with microphthalmia [2]. Anophthalmia in combination with micro-phthalmia was reported to have an incidence of 1 in 10,000 births [2].

In the United States every year 780 children are born with anophthalmia/microphthalmia [12]. Recurrence rate is 1:8 when Parents already have a child who has the condition that is, there is a 1 in 8 chance of having another child born with it experts say it can be as high as 1: 4 if the parents are from middle east [13]. Bilateral congenital anophthalmia is extremely rare [14,15], has not been reported in this hospital or in Port Harcourt, Nigeria. Hence, the need for this case report and review of literature, to highlight the challenges associated with the management of this case. We are reporting a case of a female child who was delivered with bilateral anophthalmia at a primary health center, and referred to the University of Port Harcourt Teaching hospital from Rivers state University Teaching Hospital. This child's bilateral anophthalmia occurred alone without any associated defects or syndrome. After a comprehensive examination there was no central nervous system, cardiac or musculoskeletal deformation'.

2. CASE REPORT

The mother observed that her baby failed to open her eyes from birth as shown in Fig 1 below and immediately notified the midwife at the primary health center where the baby was born who reassured her and told her to wait for at least five days after which if baby still does not open her eyes she should return to the clinic where the baby was born. After five days parents represented to the primary health centre with same complaint but in addition child was having jerky movements of upper and lower limbs. They were referred to the Rivers state University teaching hospital. Here, child was seen in the ophthalmology department and ophthalmic examination revealed depressed and closed eye sockets and child was further referred to the consultant Ophthalmologist at the University of Port Harcourt teaching hospital. As the protocol of the University of Port Harcourt teaching hospital requires that every child seeking medical care from the hospital most pass through the department of paediatrics, child was seen and admitted in to the children emergency ward. Baby was delivered via spontaneous vaginal delivery to a 21 year old unmarried mother after a term pregnancy. Pregnancy was supervised at Pott Johnson primary health center complicated by a febrile illness but no rash at 3months of gestation, with slight drainage of liquor, which was treated by a traditional birth attendant, with some herbal concoctions in form of leaves inserted into the vagina daily for 3 days, she also received vitamin k injection from the midwife at the primary health center. She had vaginal

discharge at 6 months for which a high vaginal swab was done which yielded candida. She was then treated for candidiasis. There was no history suggestive of congenital infections like toxoplasmosis or rubella but however we cannot say if the herbal leaves inserted into the vagina at three months gestation were teratogenic. She had her routine antenatal drugs, was not exposed to x-rays, alcohol or tobacco. No family history of microphthalmia/anophthalmia or any neurologic abnormality of note. Examination revealed a normal weight baby 4 kg, ophthalmological examination revealed bilateral absent globes, no dysmorphic features or other neurologic sequalae. The motor and auditory system were intact. Diagnosis of congenital bilateral anophthalmia was confirmed with a Magnetic Resonance Imaging of brain and orbit (as shown in Fig. 2) which showed absence of both globes, with hypoplasia of the orbits as well as the extraocular muscles, the optic nerves could not be differentiated from the visualized intraorbital structures. Normal cerebral and cerebellar hemispheres. No foci of altered attenuation, sulci and gyri showed normal configuration. Grey white matter interface was preserved. No intra axial nor extra axial mass lesion or collection. Normal ventricular system. Brainstem, cerebellopontine angles, Sella and pituitary are normal. Chest x-ray and abdominal ultrasound scan done were normal. Clinically our patient had no murmur to suggest a congenital heart disease, however an echocardiography was done for the patient which also confirmed the absence of a congenital heart defect. Antibodies to Toxoplasma, Rubella and Cytomegalovirus were not done. Parents had a lot of financial constraints. The managing team supported by raising funds for the few investigations that were done. Parents could not afford to pay for any. The child is currently being followed up at the paediatric, ophthalmology and Ear Nose Throat (ENT) clinics. She would benefit from a conformer while waiting for a prosthetic eye which she will eventually need.

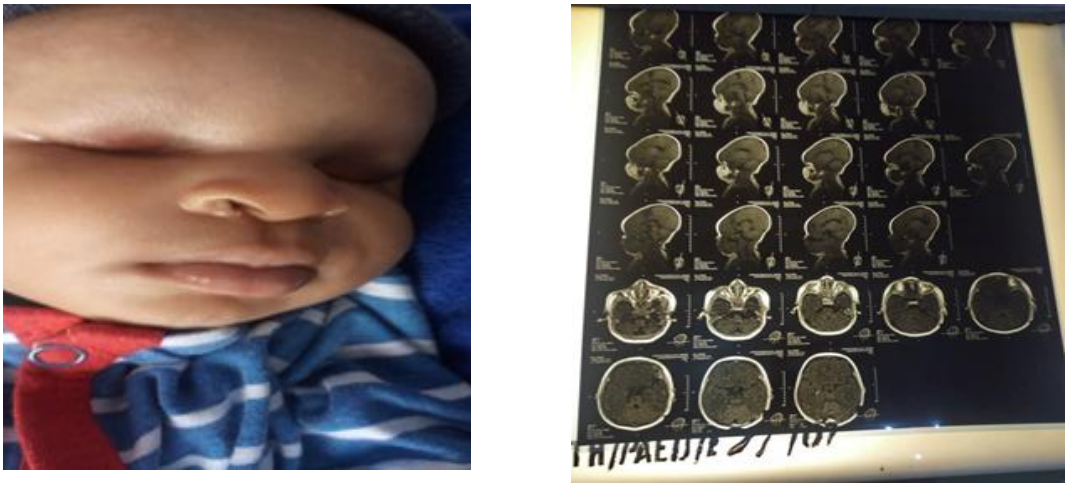

Fig 1. Photograph of the patient with bilateral anophthalmia

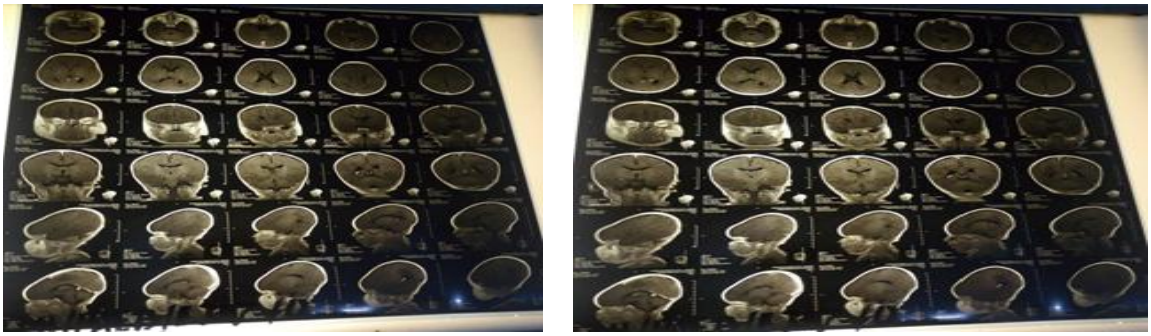

Fig 2. Magnetic resonance imaging of the patient's brain and orbit

3. DISCUSSION

Our patient had bilateral anophthalmia. Anophthalmia is frequently bilateral, similar to what was reported in Tulza in Bosnia and Kano in Nigeria [4,11]. Respectively but however, it is in contrast to what was reported in Benin city Nigeria where the two cases reported were unilateral. [8] Reviewing literature, we found out that a case of bilateral anophthalmia had been reported in Kano state [11] in Nigeria, two in Benin City Nigeria [9] while anophthalmia constituted 9.3% of the congenital eye anormalies in Enugu state southern Nigeria [10]. Though anophthalmia is associated with genetic and familiar causes [3]. We could not establish history of any family member with such a condition in our patient. The diagnosis of bilateral anophthalmia in our patient was made after birth, although she had five antenatal ultrasonography, none of them was able to pick up our patient's condition. This is similar to what was observed in Tulza in Bosnia and Kano Nigeria [4,11]. Respectively where patients with anophthalmia were diagnosed postnatally although the patient had done antenatal ultrasound scan which failed to pick up the condition. Although ultrasound scans is very useful equipment in prenatal diagnosis of congenital abnormalities, particularly after the enhancement of the quality of the pictures. The diagnosis of anophthalmia can be made by using two-dimensional ultrasonography, in the absence of eye ball and lens, but the three-dimensional ultrasound is more informational [3,15] particularly when the head of the foetus is not properly positioned [3,15]. In contrast to our report there has been antenatal diagnosis of two cases of anophthalmia in New Zealand using the reverse face view of three-dimensional (3D) ultrasound scan which is very helpful in providing useful information about the orbits and the eye balls for prenatal diagnosis and assessment of anophthalmia [15]. So it is important for the operator of the ultrasound scan to be proficient to avoid missing such rare cases as happened in this situation. Our patients mother called the attention of the midwife to the fact that her baby was not opening her eyes but was told to go home and observe her baby for five days and come back, this is very saddening and should not happen in any environment where babies are being delivered. Every new born delivered in a health facility should have a thorough physical examination by a skilled health worker before being discharged home to avoid missing such anomalies like imperforate anus, anophthalmia, hearing impairment etc. Computerized tomography scan and Magnetic resonance Imaging are very helpful in diagnosis of anophthalmia as both are able to demonstrate the nonappearance of the globe inside the orbit. However, both are very expensive in our environment, it may cost as much as 80,000 naira in Nigerian currency which amounts to over 200 dollars to have a brain/orbital Magnetic resonance imaging or Computerized tomography scan. Our patient's parents could not afford to pay for Computerized Tomography scan /Magnetic resonance Imaging; the managing team had to raise funds to support them in other to confirm the diagnosis of anophthalmia. Similar to what has been previously reported in literature [4] our patient did not have any congenital heart disease. However, in contrast to this report, the case of anophthalmia reported in Kano state, Nigeria had associated trivial tricuspid atresia, Atrial septal defect and Ventricular Septal Defect [11]. The maternal age of 21 years detected in our study is similar to 21 years and 20 years reported in Bosnia and kano [4,11] Respectively but it however, contrast with advance maternal age in another report [3] Our patients parents are faced with the ordeal of caring for a blind child for life in a resource limited setting like ours. Presently our patient will benefit from conformers while waiting for a more definitive eye prosthetics, her parents are sourcing for the necessary funds for this while she is being followed up at paediatrics, ophthalmology and ENT clinics.

4. CONCLUSION

We have reported a very rare condition of condition of congenital bilateral anophthalmia and reviewed the literature. We have also highlighted some of the challenges currently being faced by our patient. There is need for further research into the aetiology of this very rare condition with the hope of preventing it.

5. LIMITATIONS

There was a lot of financial burden on both the managing team and the parents of the child and as such some of the viral studies could not be done.

CONSENT

Authors declare that the consent of the parents was obtained.

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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