Case Report

Bilateral Congenital Anophthalmia: A Report of Two Cases and a Case for Increased Anomaly Ultrasound Scans Coverage in Pregnancy in Nigeria

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Abstract

This is a report of two cases of patients with bilateral congenital anophthalmos who presented to a tertiary hospital, in Ibadan Southwest Nigeria, with the aim of highlighting the need for increased implementation of fetal anomaly scans coverage during pregnancy in Nigeria. Information on patients' sociodemographic data, clinical features, and management are described. Both patients presented because of the inability of the parents to view the globes after delivery. Ophthalmic examination and ocular ultrasonography confirmed bilateral absence of the eyeballs in both patients and the parents were counseled on the anomaly including its treatment and prognosis.

Keywords: Anophthalmia, congenital anomalies, Ibadan, prenatal anomaly screening

Introduction

The complete absence of ocular tissue in the orbit in the presence of ocular adnexa such as eyelids, conjunctiva, and lacrimal apparatus is referred to as anophthalmia.[1] This condition may be unilateral or bilateral and it may occur in isolation or as part of a syndrome involving systemic malformations of the cardiac, musculoskeletal, genitourinary, and central nervous system.[1,2]

Diagnosis of anophthalmia is mainly clinical, following a comprehensive ophthalmic examination that reveals the complete absence of the globe.[1] There may be a phenotypic range between anophthalmia and microphthalmia.[3] Ancillary investigations such as imaging and histology, if a postmortem examination is performed, may be useful.

To establish the etiology, a detailed history, thorough physical examination, imaging, and genetic testing are essential. The causes have been identified as chromosomal, monogenic, and environmental factors.[1] The exact pathogenesis has not been fully ascertained however it is reported that infection in pregnancy with influenza or common cold infection as well as paternal systemic diseases such as hypertension and diabetes are specific risk factors for congenital anophthalmia.[3]

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The incidence of anophthalmos in Nigeria is not known. Most data on epidemiology combine data for both microphthalmia and anophthalmia. In addition, it is difficult to compare data as studies use different case definitions and inclusion criteria such as stillbirths, terminations of pregnancy, or living children. In a retrospective cross-sectional study conducted at the University College Hospital, Ibadan, from January 2009 to December 2013 of 248 children with 259 ophthalmic congenital anomalies involving the eye and/or its adnexa, 6.9% were noted to have microphthalmia/anophthalmia.[4] Other cases of anophthalmos reported as part of congenital ocular anomalies studies in Nigeria place the prevalence at 0.8% to 4% even though there is a wide variation in the number of patients, the duration, the demographics of children included, and diagnostic criteria.[5-7]

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The first reports of anophthalmos in literature from Nigeria were of two cases from Lagos in 1977.[8] Recent case reports in Nigeria are from Akwa-Ibom,[9] Benin City,[10,11] Kano,[12] Port Harcourt.[13] Enugu,[14] Benin City,[15] Kano,[5] and Lagos.[6] This indicates a wide geographical spread of cases reported across Nigeria. Syndromic anophthalmos in association with Goldenhar syndrome was also reported at Ile-Ife, Nigeria.[16]

In a study on trends over time in the incidence of some congenital ocular malformations in England from 1999 to 2011, the incidence of congenital anophthalmia was 0.04–0.24/10,000.[17] This figure was based on hospital admissions of live babies.

Prenatal anomaly scans are usually done during the second trimester of pregnancy. This test has been invaluable in the detection of ocular anomalies as well as other congenital defects.[18-20] Although a few centers in Nigeria including the University College Hospital Ibadan offer prenatal anomaly scan, it is not common practice in many centers offering maternal and child health in Nigeria.[21,22] Similarly, there is no policy document to the best of the authors' knowledge on prenatal anomaly scan in Nigeria.

We report 2 cases of female children delivered with bilateral anophthalmia highlighting the need for increased implementation of fetal anomaly scans during pregnancy in Nigeria.

Case Report

Patient 1

A five-week-old female was referred to the Ophthalmology Clinic of the University College Hospital from a pediatric clinic following complaints of the absence of the globes by the parents since birth. The neonate was delivered by spontaneous vertex delivery to a 32-year-old mother following a desired full-term pregnancy. The mother had antenatal care in a private hospital with secondary care facilities. Pregnancy was essentially uneventful. A routine ultrasound scan done in pregnancy did not reveal any abnormalities; however, an anomaly scan was not specifically requested. There was no history of intake of any drug with possible teratogenic effects during pregnancy. In addition, there was no history of fever, abnormal vaginal discharge, body rashes, or history suggestive of infections such as toxoplasma or rubella during pregnancy. She took routine prenatal vitamins but did not accede to use of illicit drugs, alcohol, or tobacco exposure. There was no history of exposure to X-rays, prior use of contraceptives, or desire to terminate pregnancy. The father is a 34-year-old trailer driver with no medical comorbidities such as hypertension or diabetes. The patient has two older sisters who are healthy. There was no family history of anophthalmia/microphthalmia or significant neurological anomalies.

At presentation in our clinic, the patient weighed 5.3 kg. Ophthalmic examination revealed deeply set orbits, small palpebral fissures, presence of eyelashes, absence of an eyeball on palpation of both orbits, and no cystic swelling. There were no associated dysmorphic facies and the rest of the systemic examination was normal. The diagnosis was confirmed with an ocular ultrasound scan that showed the absence of ocular tissue within the orbit [Figure 1].

Parents were counseled on the diagnosis and options of multi-staged reconstructive and prosthetic surgeries however the parents are yet to return for definitive care of the patient.

Patient 2

A one-day-old female was referred to the Ophthalmology Clinic of the University College Hospital, Ibadan by a midwife following complaints of inability of the parents to see the globes after her birth. The neonate was delivered by spontaneous vertex delivery to a 27-year-old mother following a full-term pregnancy. The mother had antenatal care in a general hospital with secondary care facilities. Pregnancy was essentially uneventful. Routine ultrasound scan done in pregnancy did not reveal any abnormalities, although an anomaly scan was not specifically requested. There was no intake of any drugs with possible teratogenic effects during pregnancy. No history of fever, abnormal vaginal discharge, body rashes, or history suggestive of infections with toxoplasma or rubella. She also took routine prenatal vitamins but denied the use of illicit drugs, alcohol, or tobacco exposure. There was no history of exposure to X-rays, prior use of contraceptives or desire to terminate pregnancy. The father is a 35-year-old teacher with no medical comorbidities such as hypertension or diabetes. The patient has an older brother who is healthy. There was no family history of anophthalmia/microphthalmia or significant neurological anomalies.

At presentation in our clinic, the patient weighed 2.5 kg. Ophthalmic examination revealed deeply set orbits, small palpebral fissures, presence of eyelashes, absence of an eyeball on palpation of both orbits, and no cystic swelling. There were no associated dysmorphic facies and the rest of the systemic examination was normal. The diagnosis was confirmed with

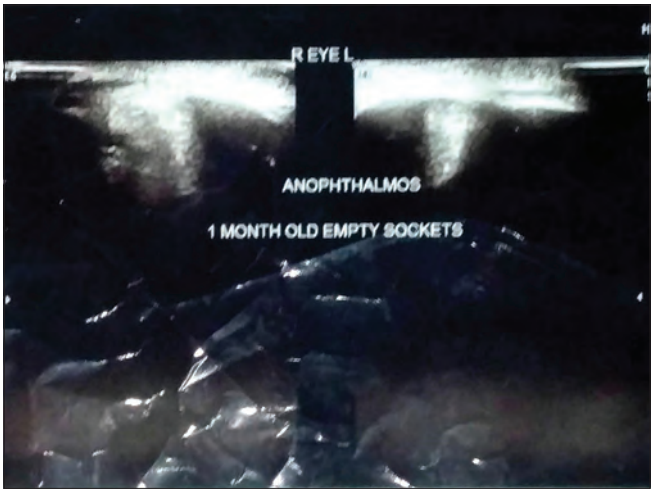

Figure 1: B mode ultrasound scan of the orbit in patient 1 showing the absence of both globes behind the lids

an ocular ultrasound scan that showed the absence of ocular tissue within the orbit [Figure 2].

Parents were counseled on the diagnosis and options of multi-staged reconstructive and prosthetic surgeries. The parents are yet to return for definitive care.

Discussion

We report two female babies with bilateral anophthalmia without any systemic malformations, diagnosed clinically and confirmed with ultrasonography. Bilateral anophthalmia is an uncommon cause of childhood blindness that is not treatable and may not be preventable. Even though blindness is the major problem with this condition, no management options are currently available to restore sight in a patient with anophthalmia. Early rehabilitation is encouraged to maximize potentials of the blind child and reduce mortality as childhood blindness has been associated with under-5 mortality in low-and middle-income countries.[23]

In addition, the absence of the globes does not only pose the problem of childhood blindness, it also has implication for facial morphology as the loss of volume can lead to hypoplasia of the orbit and face, with attendant aesthetic and psychosocial challenges.[24] Furthermore, there are social issues of stigma and parental distress of having a child with malformations.

Anophthalmia is usually bilateral and this was the presentation seen in our patients. This is similar to other cases reported in Lagos,[8] Akwa-Ibom,[9] Benin City,[10,11] Kano,[12] and in Port Harcourt[13] all in Nigeria. Although anophthalmia is closely linked to genetic causes, none of our patients had a family history of a similar condition. Similarly, none of the case reports from Nigeria had a positive family history. This may be an indication of unreliable verbal reports of family history from the parents as they were quite distressed at the time of consultation.

Anophthalmia with systemic associations (polydactyl) has been reported in Lagos.[8] The study was a case report on two patients, one of the patients presented with bilateral anophthalmos and associated polydactyly of both hands. In Kano, there were associated cardiac septal defects.[5] In the Ibadan study, the association between microphthlamia/anophthalmia and the occurrence of multiple systemic anomalies was significantly higher at 37.5% when compared to the association of systemic anomalies with other ophthalmic congenital anomalies such as congenital cataracts (22.0%).[4] Our patients did not return for follow-up and further evaluation, hence, we may have missed non clinically obvious associations. Unilateral congenital anophthalmos has also been reported in Nigeria, precisely, in two patients from Benin City.[15]

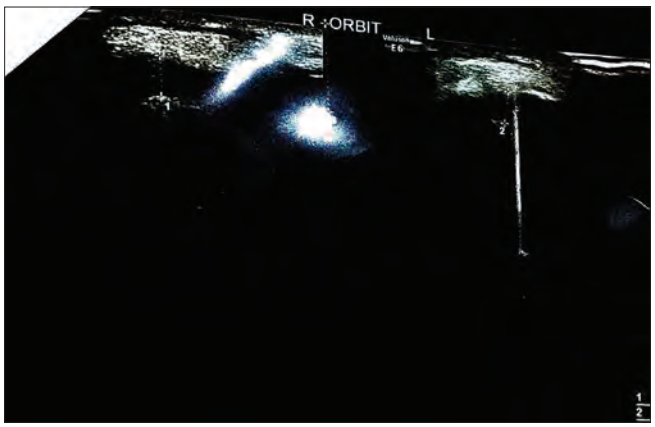

Figure 2: B mode ultrasound scan of the orbit in patient 2 showing the absence of both globes behind the lids

Other etiological factors presumably implicated in the diagnosis of anophthalmia include oral ingestion of maternal drugs (especially herbal medications) during pregnancy,[9,15] and per vaginum insertion of herbal concoctions.[13]

Antenatal ultrasound scans during pregnancy did not detect the anophthalmia in both patients. It is not known if specific anomaly scans were performed. Routine antenatal ultrasound scans are not fully able to detect all anomalies as reported in Port Harcourt, Côte d'Ivoire as well as in Bosnia and Herzegovina.[13,25,26] This highlights the limitations of the current radiology-based antenatal diagnosis of pathologies in Nigeria which may be due to lack of specialized ultrasound equipment or low coverage of skilled radiologists. This report also highlights the importance of fetal anomaly screening during pregnancy and the importance of the ultrasonography as a diagnostic tool. Reports of prenatal diagnosis of anophthalmia during second-trimester anomaly screening using ultrasound are documented in the literature.[20] This impacts greatly on parental counseling, other support measures, and tailored treatment options as well as timing of treatment subsequently.[27] In our patients, the diagnosis was at birth and the parents especially the mothers were trying to cope with the stress of nursing a baby and receiving the distressing news of congenital anomaly in their baby at the same time.

Management of anophthalmos is challenging, prognosis is varied and patients usually require the support of a comprehensive multidisciplinary team made up of the ophthalmologist, ocularist, pediatrician, radiologist, clinical geneticist, caregiver, and counselor. This is necessary for mobility rehabilitation, plastic surgery, and psychosocial support.[1,24,28] Ocular prostheses, multiple surgeries, close follow-up, and prolonged recovery are considerations but even in the presence of facilities, counseling the family on realistic outcome of such therapy cannot be overempazised.[24] In the management of the anophthalmic socket, a conformer can be worn with or without an implant.[24] Surgical management is usually multi-staged and may involve the insertion of an implant, use of orbit tissue expanders, conjunctival socket expansion, and orbitocranial advancement surgery in different combinations.[24] The functional and aesthetic results of surgical intervention may sometimes be disappointing.

Other challenges with management that may be peculiar to resource-poor countries include poverty and lack of health

insurance to cover cost of multiple surgical interventions. Both patients did not have health insurance and explaining the cost of care to the parents without any assurance of restoration of vision may be a significant barrier to uptake of care.

Furthermore, both children may have difficulty integrating into the society because of their blindness and facial malformation as very few facilities exist to cater for the blind in our locality. In addition, psychosocial needs of the patients and parents are important for acceptance and developing coping mechanisms.

A limitation was the inability to obtain magnetic resonance imaging scan images of the brain to confirm the absence of associated intracranial abnormalities. Milder abnormalities not immediately evident on clinical examination may have been missed. Furthermore, genetic/chromosomal analysis could not be carried out due to financial constraints.

We recommend increased adoption of anomaly scans as part of antenatal care for pregnant women in Nigeria. Although rare, a diagnosis of anophthalmos in the antenatal period can set up psychological support and counseling even before delivery and this may equip the parents to cope better when the child is delivered.

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Conflicts of interest

There are no conflicts of interest.

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