Case Report

Bilateral Congenital Anophthalmia and Hydrocephalus in Calabar, Nigeria: A Case Report

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Abstract

Congenital anophthalmia is a rare deformity with a complex aetiology. It can co-exist with disorders of central nervous system. The aim of this report is to present the socio-demographic, clinical data and management challenges of an abandoned HIV-exposed neonate with bilateral congenital anophthalmia and hydrocephalus, in Calabar, Nigeria.

A 6-day-old female term neonate presented to the Newborn Intensive Care Unit of a tertiary hospital in Calabar, Nigeria, with a history of inability of mother to see the baby's eyeballs since birth. There was no family history of a similar eye defect, consanguinity, or other associated histories. Ophthalmic examination findings included: mucopurulent discharge, shortened eyelids and palpebral fissure and absent eyeballs. A Transfontanelle ultrasound scan of the large head showed an obstructive hydrocephalus. Mother abandoned the baby in the hospital.

Management of bilateral congenital anophthalmia is multidisciplinary. There may be an association with HIV infection. Abandonment of malformed babies is of psychosocial importance.

Keywords: Calabar, congenital anophthalmia, HIV-exposed, hydrocephalus

Introduction

Anophthalmia can be defined as the absence of an eye ball.[1] It is an irreversibly blinding condition and may be congenital or acquired, clinical or true, unilateral or bilateral, primary, secondary, or degenerative.[1-3] Though incidence in Nigeria is unknown, this condition is reportedly rare and has a global birth prevalence of 3 per 100,000 population.[1-6]

The process of eye development is complex and intricately initiated by the homeobox gene. Blood vessels, part of the cornea, extraocular muscles and sclera develop from the extracellular mesenchyme. Ciliary body, iris optic nerves and retina from neuroepithelium while corneal epithelium, eyelids and crystalline lens develop from the surface ectodermal tissue.[1,3] Eye development begins during the 3rd week of intrauterine life with the formation of the optic vesicles from the optic grooves from the forebrain. As the optic vesicles make contact with the surface ectoderm, they invaginate and develop into the layered optic cups, the lens placode (which becomes the lens vesicle and eventually crystalline lens) forming from the thickened surface ectoderm.[1,3]

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Congenital anophthalmia results from multiple aetiologies and possibly a complex interplay of genetic and environmental factors.[1,3] The insult to the developing eyes is typically intrauterine, during the first 8 weeks of life.[1]

A clinical diagnosis can be made with radiologic evaluation via ocular ultrasound scans, computed tomography scan (CT scan) and magnetic resonance Imaging (MRI) of the orbit and brain or histopathologic sections, demonstrating the absence of ocular tissues in the orbit and thereby confirming the diagnosis.[1,6,7] While up to 50% of bilateral anophthalmia occurs in association with systemic abnormalities, it may also present as part of a syndrome (Patau's syndrome, CHARGE

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syndrome, Focal Dermal Hypoplasia syndrome, etc.) or following chromosomal abnormalities, for example, SOX2, OTX2 and PAX6 gene deletions etc.[1,3,6]

Preferred management protocols include a detailed history, general and ophthalmic examinations, as well as radiologic and genetic evaluations (including parental screening), anchored on a multidisciplinary team approach as appropriate.[1,3,6,7] The standard of care for patients with anophthalmos includes the surgical use of orbital expanders and ocular prosthesis.[7] Congenital anophthalmia can cause significant psychosocial problems and parental distress.[6] Consequent on this, prenatal detection can be beneficial for planning the child's care pathway and psychosocial support.[6,7]

Bilateral congenital anophthalmos has not been reported in Calabar, Nigeria, to the best of our knowledge. The aim of this report is to present the socio-demographic, clinical data, and management challenges of an abandoned HIV-exposed (HIV-E) neonate with bilateral congenital anophthalmia and hydrocephalus, in Calabar, Nigeria.

Case History

V.V.E. was a 6-day-old female born at 39 weeks gestation who presented to the neonatal intensive care unit of the University of Calabar Teaching Hospital (UCTH) with bilateral absence of the eyeballs. Mother was a 41-year-old divorced, gravida 4 para 4 (4 alive), petty trader who is HIV positive (tested by DNA PCR tests) but has yet to commence antiretroviral therapy (ARV). The pregnancy was unplanned but spontaneously conceived. Mother booked for antenatal care (ANC) at a secondary hospital in Calabar Metropolis, although she was inconsistent with her ANC clinic schedules. The antenatal period was eventful, with chronic hypertension diagnosed in the first trimester and managed with Nifedipine tablets. Mother was delivered at will in a church via spontaneous vaginal delivery. The child cried spontaneously after birth and was not breastfed. Father was a 47-year-old traffic officer, and there was no consanguinity; however, other details about father were unknown at the time of this report as attempts to locate him proved futile. There was no positive family history of a similar eye defect.

Anthropometric measurements were: birth weight – 2.15 kg; length – 48cm; occipital frontal circumference (OFC) 34 cm. Ophthalmic examination on admission revealed absence of eye balls in the sockets on palpation, deep-set shortened eyelids, narrow and shortened palpebral fissures and matted inward-turning eyelashes with mucopurulent discharge [Plate 1]. Orbital B-mode ultrasound scans showed the absence of eyeballs behind both eyelids and no discernible optic nerves [Plate 2]. Neonates tolerated oral feeds of volumes of infant formula appropriate for age and opened their bowel as needed. She had a normal temperature, heart rate and other vital signs. She also did not manifest symptoms associated with acutely raised intracranial pressure in infancy, including high-pitched cry, poor feeding, vomiting, irritability or lethargy and bulging of the fontanelle. A diagnosis of low-birth-weight term HIV-E (Mothers HIV status was determined by DNA PCR test) infant with bilateral congenital anophthalmia and neonatal conjunctivitis was made.

In summary, the management plan included conjunctival swabs for gram staining, microscopy, culture and sensitivity and Gutt Tobramycin 12 hourly OU for 2 weeks. Intravenous Ceftriaxone 50 mg/kg daily for 5 days, Syrup Zidovudine 1 mg/kg daily in two divided doses. Syrup Nevirapine 1 mL daily for 2 weeks. Infant formula via cup and spoon, 35 mL 2 hourly. Oculoplastic review was done as patient could receive cosmetic benefit from future planned

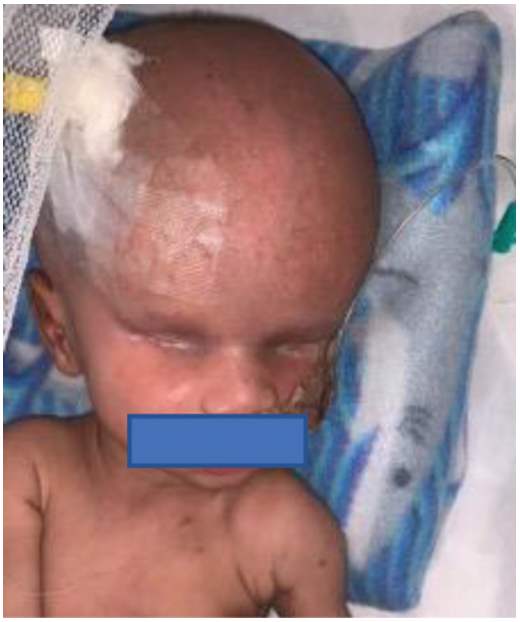

Plate 1: Sixteen-day-old female with bilateral congenital anophthalmia, neonatal conjunctivitis and hydrocephalus.

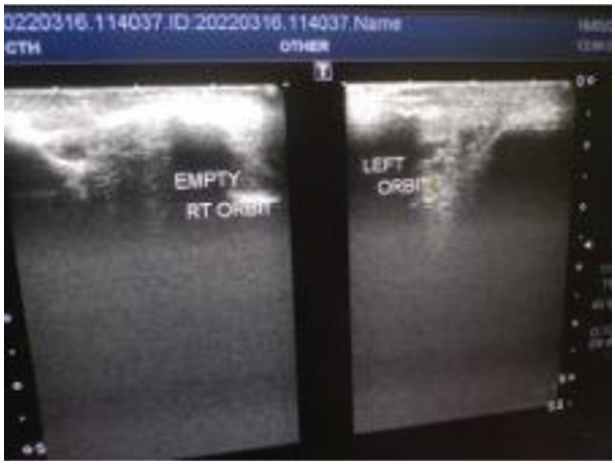

Plate 2: Ocular B mode ultrasound scan of both orbits in index patient confirming absence of globes behind the eyelids.

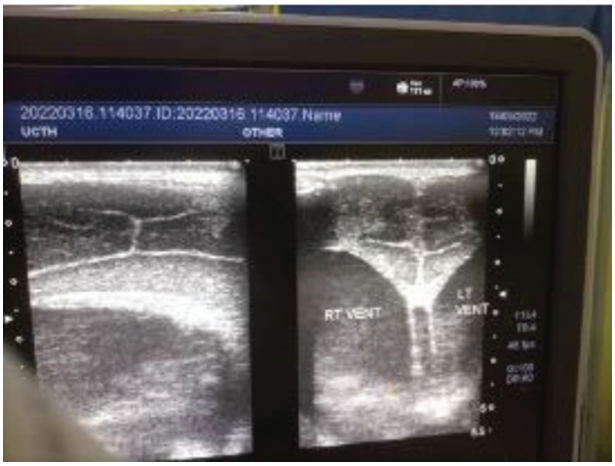

Plate 3: Trans-fontanelle ultrasound scan of hydrocephalus in a newborn infant.

orbital reconstructive surgeries with orbital implant placement and the use of ocular prosthesis. Cardiovascular assessment for cardiac associations was also done.

Furthermore, the occipitofrontal circumference (OFC) rapidly increased in size from 34 cm at presentation to 40.8 cm, with other systemic examinations remaining essentially normal. Trans-fontanelle ultrasound scan showed moderate dilatation of both lateral, third and fourth ventricles, with the frontal horns of the lateral ventricles measuring 14 and 15 mm on the right and left, the third ventricle measuring 12 mm and the fourth ventricle 11 mm. There was an obstructive hydrocephalus at the level of Foramen of Magendie [Plate 3].[13] The diagnoses were subsequently modified to include moderate arrested hydrocephalus. The neurosurgical team commenced conservative management with active surveillance to determine if (by close monitoring of the OFC) or when (i.e., when signs of progressive hydrocephalus are present) to institute medical and surgical management as indicated in active hydrocephalus. A brain MRI scan was also requested.

The mother abandoned her infant after the fourth day of hospital admission and could neither be found by the hospital social services team nor traced to the address or phone numbers given.

The newborn ICU team was unable to do the TORCHES syndrome titer, arrange for orbital surgery and prosthetic treatment, perform the brain MRI scan and do the chromosome makeup studies at the time of this report due to significant financial constraints (in our resource limited setting) and the absence of a health insurance policy cover.

Discussion

We present a report on the management challenges of an abandoned female neonate with bilateral congenital anophthalmia and arrested hydrocephalus. Congenital bilateral anophthalmia with negative family histories has similarly been reported in Nigeria and elsewhere.[2,4-6] Risk factors include antenatal exposure to environmental factors, such as solvents and drug misuse and abuse and exposure to radiation (including X-rays). Others include maternal infections acquired during pregnancy (especially Rubella, congenital rubella syndrome, influenza virus and other viral infections), maternal vitamin A deficiency, as well as paternal diabetes and hypertension.[1,3] Low birth weight, multiple gestations, increased maternal age and exposure to teratogenic medication early in pregnancy have likewise been implicated.[1,2,4] Gender and race predilections are not established.

The mother did not have exposure to X-rays or known teratogenic drug use, which agrees with some reports.[2-8] There was no history of febrile illness or rash early in pregnancy, use of peri-conception folic acid, illicit drugs, alcohol, or substance abuse. She did not attempt an abortion of the pregnancy.[3,7] However, she had an increased maternal age and had been HIV positive for an undetermined duration. Globally, more than a million HIV-E infants are born to HIV infected mothers, with the burden in poorer economies.[9] There is a higher prevalence of birth defects in children born to mothers with HIV infection than in the general population.[10] Nevertheless, it is unclear whether in-utero HIV exposure and infection can lead to characteristic organ developmental defects and congenital malformations, including anophthalmia and hydrocephalus, via HIV embropathy.[9-11]

Associations between in utero, first trimester exposure to ARVs, especially a potential teratogen- efavirenz (a non-nucleoside analogue), or Lopinavir and birth defects (including neural tube defects, unilateral microphthalmia and anophthalmia) have been found.[1,9] However, the mother of our index patient was yet to commence ARVs. The HIV status of our patient could not be ascertained at the time of this report (6 months after birth) as a result of a national lack of HIV PCR testing kits though blood samples were collected. Our inability to obtain HIV test results in a reasonable time is a significant hindrance to instituting timely and appropriate clinical management.

Our neonates' mother had chronic hypertension (commonly associated with pregnancy) and was placed on Nifedipine (considered safe during pregnancy) for this in her first trimester of pregnancy. This drug is not a major teratogen, and a newborn whose mother received it has similar risks of developing congenital malformations (especially cardiovascular) as those whose mothers have hypertension but do not receive Nifedipine.[12]

Congenital anophthalmia has a high rate of CNS associations,[3,7] and although hydrocephalus has a multifactorial aetiology,[13] the association of bilateral anophthalmia with hydrocephalus is yet to be reported in Nigeria to the best of our knowledge. However, hydrocephalus has been reported with ocular abnormalities, where it was seen with congenital anophthalmia in Ireland.[8]

The majority of these children had Patau's syndrome (Trisomy 13). Our patient also did not have congenital heart disease, as reported elsewhere in Nigeria.[4-6]

Our patient had neonatal conjunctivitis, was exposed to some risk factors for this including an inconsistent ANC schedule and delivery in unusual places. However, mother did not have a history of premature rupture of membranes, vaginal discharge or peri-partum pyrexia.[14] Importantly, what has been presented as an infected socket in the newborn, may be equated to neonatal conjunctivitis.[8]Sadly, our patient was also abandoned and this posed a significant management challenge. While it is not always clear what makes mothers abandon their children,[15] poverty or financial hardship, maternal illness including HIV or poor mental health, social exclusion and childhood illness (especially in women with children suffering from disabilities and deformities) are some of the established reasons for child abandonment.[15] Others include having more children than they feel they can afford and being unmarried.[15] Some of the reasons for child abandonment reported in literature were found in our case.

Better access to and improved ANC (including an antenatal abnormality scan), as well as social intervention strategies against reasons for child abandonment, may better prepare parents to accept a newborn with congenital malformations or deformit.[6] The management of congenital anophthalmia is complex. Our patient is unique because she has a negative family history, is HIV-E, had neonatal conjunctivitis and hydrocephalus, and has been abandoned.

Conclusion

We present the socio-demographic and clinical data of a neonate with bilateral congenital anophthalmia and a negative family history, in Calabar, Nigeria. Congenital anophthalmia may be associated with neonatal conjunctivitis and hydrocephalus as was seen in our patient. An environmental association may exist with HIV-E and HIV infection. Ultrasound scans of the orbit and trans-fontanelle scans are useful to confirm the diagnosis. The management challenges of our malformed neonate included a complex, medical care plan, inadequate psychosocial support and abandonment by her mother. There is a need to strengthen social services as part of the health system strengthening for ophthalmic medical care in our environment.

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Conflicts of interest

There are no conflicts of interest.

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