

# Congenital Anophthalmos in Benin City

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## ABSTRACT

Congenital anophthalmos is a rare clinical condition. This is a report of two cases of clinical anophthalmos seen in Benin City, Nigeria. Two cases of unilateral anophthalmos have been previously reported. Bilateral clinical anophthalmos is a relatively rare condition hence the first reported case in Benin City is presented. The first patient presented with primary anophthalmos while the second case presented with degenerative or consecutive anophthalmos.

**Keywords:** Anophthalmia, anophthalmos, child, clinical, congenital, empty socket

## INTRODUCTION

Anophthalmia refers to the absence of ocular tissue in the orbit.<sup>[1]</sup> It is a rare condition with a birth prevalence of 3 per 100,000 population.<sup>[2]</sup> It may occur in isolation or as part of a syndrome. Chromosomal, monogenic and environmental causes have been identified. Chromosomal causes include chromosomal duplications, deletions and translocations. *SOX2* has been identified as a major causative gene, other genes such as *PAX6*, *OTX2*, *CHX10* and *RAX* have been linked to this condition.<sup>[1]</sup> Environmental factors which have been found to be contributory include gestational-acquired infection, maternal vitamin A deficiency, exposure to X-rays, solvent misuse and thalidomide exposure.<sup>[1]</sup>

True anophthalmos results from failure of development or the complete regression of the optic vesicle. It is rare and can only be diagnosed after serial histological sectioning of the orbit to exclude neuro-ectodermal structures such as the retina or optic nerve. Computed tomography (CT), magnetic resonance imaging (MRI)

and ultrasonography are helpful in making the diagnosis revealing a complete absence of ocular tissue in the orbit.<sup>[3]</sup>

The term clinical anophthalmos is used in the absence of histological diagnosis and denotes the absence of clinically detectable ocular structures.<sup>[3]</sup> It could be primary, secondary or consecutive (degenerative).<sup>[4]</sup>

Cases of bilateral clinical anophthalmos are relatively rare. Some studies<sup>[5,6]</sup> have reported bilateral anophthalmos. In Nigeria, two bilateral cases were first reported in Lagos.<sup>[7]</sup> In Benin City, the first report was of two unilateral cases.<sup>[8]</sup> Another report presented two cases, the first of which had right anophthalmos and left microphthalmos, while the second had a right microphthalmos and a left anophthalmos.<sup>[9]</sup> A recent study on congenital ophthalmic anomalies conducted between January 2005 and December 2009 reported four cases of anophthalmos/microphthalmos.<sup>[10]</sup> To the best of our knowledge, this is the first reported case of bilateral anophthalmos in Benin City, Nigeria

## CASE REPORTS

We describe two cases of clinical anophthalmos, the first being bilateral and the other unilateral, being managed in University of Benin Teaching Hospital, Benin City

### Case 1

E.E is a 9-day-old female child who presented with an inability to open both eyes since birth [Figure 1]. Both

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palpebral fissures were narrow with no visible eyeball in both sockets clinically. She had no other congenital anomalies. She was delivered after a normal full-term pregnancy. There was no history of maternal exposure to X-rays, ingestion of native concoctions or steroids, febrile illness and rash during pregnancy. There was no history of consanguinity. Orbital ultrasound revealed rudimentary and deeply set eyeballs. The axial length of the right eye measured 5.4 mm with no evidence of intraocular tissue while the left eye measured 6.6 mm and appeared as a solid mass. She was booked for examination under anesthesia and possible bilateral canthoplasty but the patient has been lost to follow up.

### Case 2

A.M. is a 1-week-old male child who presented with a mass protruding from the left orbit from birth [Figure 2]. The mass was firm in consistency. He had no other swellings or congenital anomalies. Antenatal period was uneventful. There was no history of maternal ingestion of native concoctions, exposure to X-rays, febrile illness or rash during pregnancy. The patient is the first child in a monogamous setting. Ocular ultrasound revealed an echogenic mass measuring  $14.5 \times 9.5 \text{ mm}^2$  arising from the left orbit with no detectable globe. The right eyeball had an axial length of 18.5 mm. The intraocular and retrobulbar structures of the right eye were normal. The patient had an excisional biopsy of the left orbital mass when he was 14 weeks old. He subsequently developed a contracted socket and atrophy of the medial third of the upper eyelid which precluded the fitting of an orbital prosthesis.

### DISCUSSION

Congenital anophthalmos could be primary, secondary or consecutive.<sup>[4]</sup> In primary anophthalmos, there is a failure of development of the optic vesicle, but no gross anomalies of the medullary tube. This condition is

thought to be bilateral and sporadic.<sup>[5]</sup> In the secondary type, there is a complete suppression or gross anomalous development of the entire anterior part of the neural tube. This results in severe malformations which are probably not compatible with life.<sup>[5]</sup> In consecutive or degenerative anophthalmos, the optic vesicle is formed but subsequently undergoes degeneration.<sup>[5]</sup>

The etiology of anophthalmos has been the subject of speculation.<sup>[5]</sup> Chromosomal, monogenic and environmental causes have been identified.<sup>[1]</sup> Clinical anophthalmos has been associated with genetic diseases such as trisomy 13 and Klinefelter's syndrome.<sup>[3,11]</sup> Most cases appear to occur sporadically, but certain modes of inheritance have been documented including autosomal dominant, autosomal recessive and X-linked transmission.<sup>[3,11]</sup> Other factors implicated in the etiology of anophthalmos include physical agents, such as X-rays, chemical teratogens, such as lithium, selenium, noxious teratogens, intrauterine infections, such as rubella, syphilis and toxoplasmosis.<sup>[11]</sup> In a case of anophthalmos in Edo State, Nigeria, ingestion of traditional medication during pregnancy was presumed to be the teratogenic agent.<sup>[9]</sup> In another case, the mother had native herbs applied to her abdomen during pregnancy though she denied ingesting of traditional medications.<sup>[8]</sup> In the two cases presented, there was no positive family history hence suggesting that they may be sporadic.

Management of anophthalmos is multidisciplinary and involves the pediatric ophthalmologist, pediatrician, counselor for genetic counseling and clinical geneticist. The aim of treatment is to maximize the visual potential of the child. In children with an only eye, the sighted eye should be protected and spectacles prescribed for any significant refractive error.<sup>[12]</sup> Surgical treatment involves socket expansion through socket reconstruction or use of socket expanders which should



Figure 1: Bilateral anophthalmos



Figure 2: Unilateral anophthalmos with a left orbital mass

be instituted within weeks of life to minimize the facial deformity and can be started soon after the birth.<sup>[12]</sup> Socket expanders such as hydrophilic expanders by Acri.Tec® or sequential solid orbital expanders such as acrylic shapes can be used but are not available in our facility. This procedure is best carried out in a specialist unit with a good ocular prosthetic service available.<sup>[12]</sup> The patient with bilateral anophthalmos could have benefited from bilateral expansion of the sockets and fitting of bilateral customized prosthesis. Also such a patient would have needed to be rehabilitated and supported vocationally by attending the school for the blind.

Alternatively if the mass is within the orbit, it serves as a stimulus for orbital growth. However, the mass protruded outside the orbit in the patient would not have contributed to the growth of the orbit hence it requires excision. The orbit was hypoplastic and patient would need socket reconstruction and orbital implantation.

In developed countries, vision support services are available to aid visually impaired children and their parents can access the financial support. Organizations for the visually impaired offer specialist advice and family support for these children and their families.<sup>[12]</sup> These support services are sadly lacking in our environment.

## CONCLUSION

Congenital anophthalmos is a rare anomaly. Attempt at expanding the socket through the socket expanders or socket reconstruction is needful. Early socket expansion and maximization of visual potential where possible is

recommended. Provision of specialized oculoplastic services through the training of oculoplastic surgeons with other support staff and adequate facilities to aid in the support and rehabilitation of these patients is recommended.

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