



Bilateral congenital anophthalmia: A case report.

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Abstract

Introduction: Congenital anophthalmia is a rare ocular malformation; it consists of the absence of the eyeball at birth and can present unilaterally or bilaterally.

Clinical case: Its complex etiology includes environmental and genetic factors. This pathology has an estimated general prevalence of 3 per 100,000 live births in isolation and 30 per 100,000 live births associated with other malformations, representing approximately 4% of the causes of blindness.

Diagnostic workshop: Anophthalmia can be diagnosed both in the prenatal and postnatal periods, and its treatment, complex and prolonged, will depend on the degree of severity of the pathology and the moment of diagnosis, as well as the start of treatment.

Evolution: This malformation contributes significantly to child morbidity and has a significant impact on the child and his family, which is why prenatal diagnosis is essential to be able to initiate prompt intervention by a multidisciplinary team where together with the treatment doctor, emotional and psychological support to the family is emphasized.

Conclusions: Congenital anophthalmia is a severe ocular malformation that can significantly impact the life of the child and his family.

Keywords

MeSH: Anophthalmos; Child; Infant, Newborn, Diseases; Genetic Diseases, Inborn; Protheses and Implants; Case Reports.

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Introduction

Congenital malformations are structural and functional alterations that occur during intrauterine life and can be identified in the prenatal stage, at birth, or later in life. These factors have an essential influence on infant morbidity and mortality [1].

Of the congenital malformations, ocular malformations constitute a broad group of eye organogenesis disorders and occupy one of the first places as a cause of visual impairment or blindness in children, contributing significantly to childhood morbidity [2].

Among the congenital ocular malformations, anophthalmia and microphthalmia are described, defining anophthalmia as the absence of the eyeball with the presence of ocular annexes (eyelids, conjunctiva, and lacrimal apparatus), and microphthalmia is defined as an eye with an axial length two standard deviations below average for age [2, 3].

Anophthalmia was first described by Lyscostenes and Scenck in 1609 and then by Barthelin in 1657. At the beginning of the 19th century, Briggs discovered the hereditary nature of this disorder [3]. This rare malformation can be unilateral or bilateral [4]; the precise pathogenesis of anophthalmia and microphthalmia remains unknown. Mann [8] suggested that anophthalmia has its genesis early in gestation as a result of the failure of development of the anterior neural tube (secondary anophthalmia) or optic fossae to enlarge and form optic vesicles (primary anophthalmia). A third category, consecutive or degenerative anophthalmia, is applied to cases where the optic vesicles degenerate and disappear after forming [5]. Its etiology is complex and includes environmental and genetic factors. The environmental causes may correspond to potentially teratogenic factors such as chemical, biological, or physical agents that interrupt normal ocular development. Within the genetic factors, three types of origin can be recognized: multifactorial, chromosomal, and monogenic, with rates of 18 to 24% of cases that have been associated with mutations of the SOX2, RAX, OTX2,

CHX10, FOXE3, GDF3, and GDF6 genes. CRYBA4 [2, 5, 6] of autosomal recessive or dominant transmission, with diverse phenotypes such as aniridia, foveal hypoplasia, optic nerve hypoplasia or microphthalmia and anophthalmia in the most severe cases.

In congenital bilateral anophthalmia, the eyelids, extraocular muscles, lacrimal system, and orbit are formed independently of the development of the optic vesicle, so in the clinical picture, these structures can be observed to develop with a low degree of compromise, that is, orbits with palpebral indentation and small eyelids, shallow fornices, and tear ducts may or may not be present. This is why there are cases of anophthalmia in which there is low tear production due to a nonexistent stimulation of the eyeball [7].

The diagnosis can be made prenatally or postnatally by clinical findings, imaging studies (ultrasound, computed tomography, and magnetic resonance imaging), and genetic analysis [4, 8].

Treatment aims to stimulate adequate orbit growth in congenital anophthalmia and microphthalmia cases. The degree of globe deformity determines which oculoplastic rehabilitation technique is needed. It will depend on the patient's age and the volume of the orbital content (microphthalmic eye and orbital cyst or only orbital cyst in cases of anophthalmia). For this reason, cavity management in children with microphthalmos and anophthalmos is complicated and prolonged. Among the different therapeutic possibilities, the following stand out: a) ocular prosthesis, b) rigid formers, c) gel expanders with high or low hydrophilicity, d) dermal-fatty graft, e) orbital cyst, f) surgery, and g) combined [9].

The following case of congenital bilateral anophthalmia is presented as a consideration of the rarity of the pathology and the relevant medical, family, and social impact that it entails.

Clinical case

A 25-year-old female patient with a 39-week pregnancy received prenatal care throughout the pregnancy, in

addition to private ultrasound controls where no anomalies were reported. Prenatal routine, he said, average. Chromosomal echo reported with Doppler abnormality of the uterine arteries. Morphological echo, reported without irregularities. Glucose tolerance curve within normal parameters. No reported congenital or urinary infections during pregnancy, nor consumption of teratogenic drugs; only reports prenatal vitamins and acetylsalicylic acid 100 mg once a day up to 24 hours before delivery; denies exposure to other medications, use of alcohol, tobacco, or other drugs. There was no history of radiation exposure. It is the first pregnancy of healthy parents with no significant personal or family history. During the first trimester, he reported SARS-CoV-2 infection, which occurred with an unquantified thermal rise that did not require hospitalization or the need for supplemental oxygen.

It was decided to terminate the pregnancy by cesarean section due to cephalopelvic disproportion without complications. A female newborn receives cephalic presentation with a circular cord in the neck, APGAR 9 - 9. Clear amniotic fluid with lumps. Anthropometric data were adequate for gestational age: weight 2870 grams, height 47.5 centimeters, head circumference 33.5 centimeters, and arm circumference 10 centimeters. The immediate physical examination revealed an empty bilateral eye orbit and fused eyelids, and the bilateral eyeball was not palpable.

The audiometric and motor exams were standard. An ophthalmic ultrasound was assessed by ophthalmology with results of an ocular cyst of 7.68 mm in the right eye and the absence of an eyeball in the left eye. The diagnosis was confirmed by an MRI of the simple skull with results of right microphthalmia and left anophthalmia, without pathological changes at the brain level. Complementary tests are performed to rule out accompanying malformations, including an echocardiogram that reports situs solitus and levocardia.

Figure 1. Patient with anophthalmia on physical examination.



Figure 2. Echocardiogram.

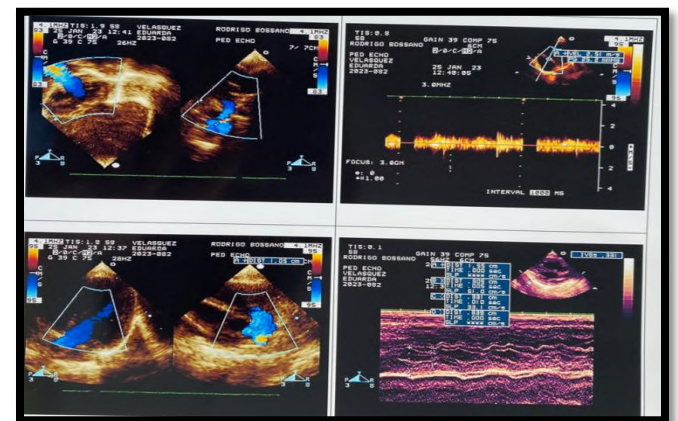


Figure 3. Ophthalmic ultrasound of the right eye: ocular cyst of 7.68 mm.

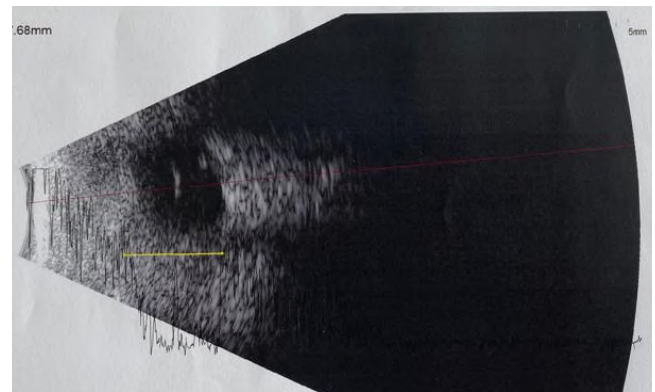
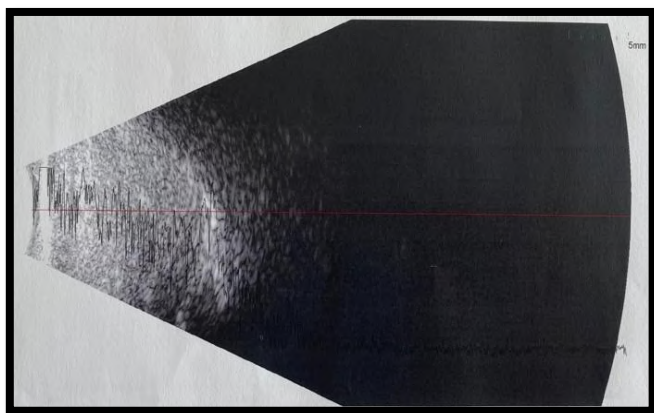


Figure 4. Ophthalmic ultrasound: left eye, absence of eyeball.

Normal anatomical relationship. Average chamber size and thickness. Normal function of the left ventricle Tapase 1.19. Absence of congenital and acquired malformations. Normal coronary arteries. Expected flows, pulmonary normotension (SBP 30 mm). Normal pericardium.

In addition, pelvic and abdominal ultrasound showed no morphological changes in the liver, gallbladder, pancreas, or kidneys. Transfontanellar ultrasound did not reveal superimposed lesions in the caudothalamic sulci. Hydrocephalus is not observed. Cerebral sulcation of the usual pattern preserved. No alterations in the echogenicity of the white matter were observed.

Adequately developed midline elements. Brainstem and cerebellum without alterations in shape and echogenicity. Symmetrical choroid plexuses of preserved size.

She was referred to the ophthalmologic surgery department for possible placement of an ocular prosthesis.

Discussion

Clinical congenital anophthalmia is the unilateral or bilateral absence of the eyeball, which occurs in isolation or as part of a syndrome [10]; it can present as part of a syndrome with multiple malformations. Goldenhar syndrome and Hallerman Streiff syndrome are the most frequent, causing auricular, mandibular,

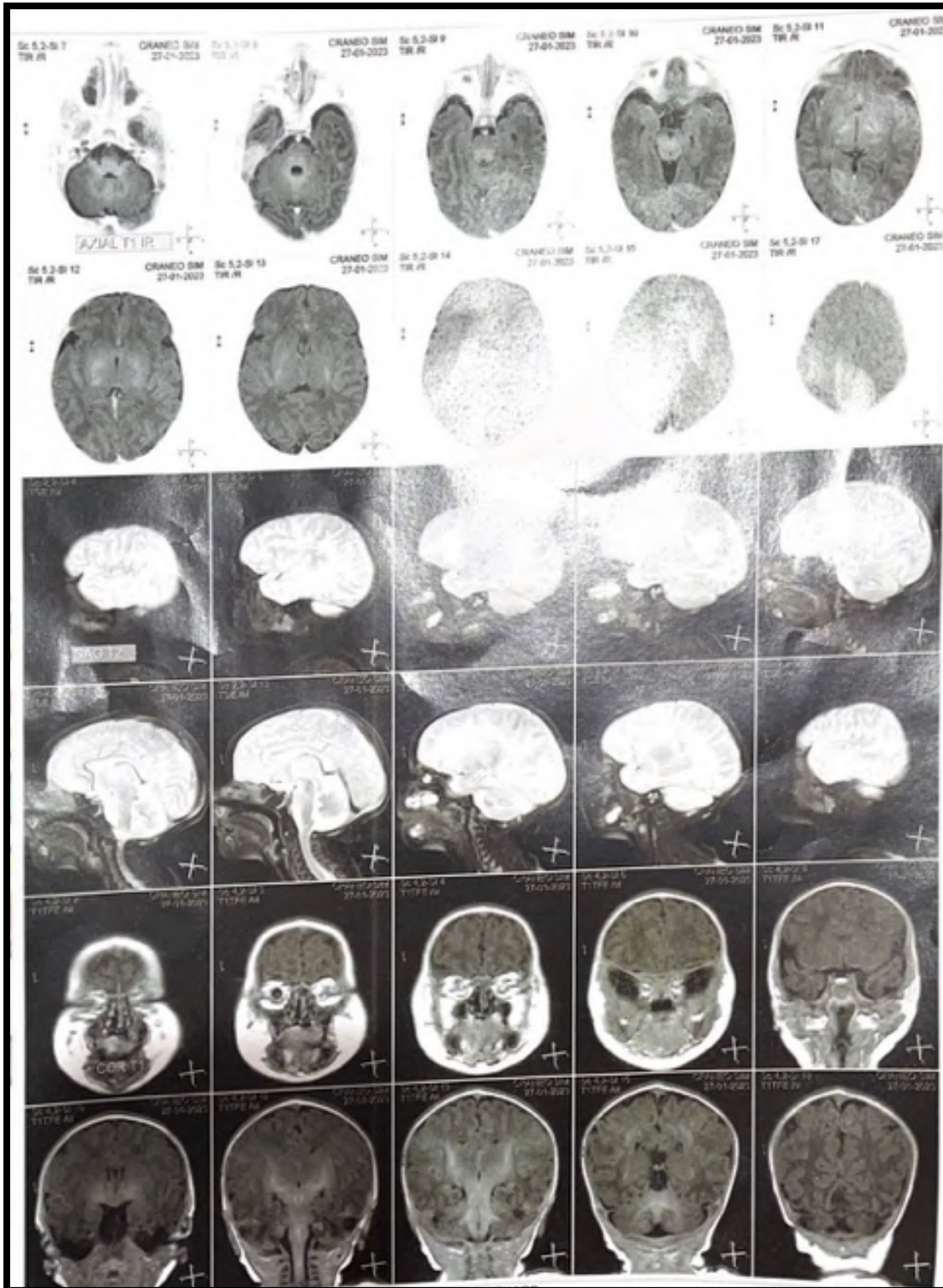
and vertebral malformations and sparse hair, tooth abnormalities, degenerative skin changes, and short stature, respectively [7]. The case presented complies with the concept of anophthalmia, and it has not been associated with any congenital syndrome until this report.

Among the causes associated with anophthalmia are those of environmental origin: exposure to teratogens (chemical, biological or physical agents), infections acquired during pregnancy (rubella, cytomegalovirus, toxoplasmosis, chickenpox, and influenza), and noninfectious causes, such as exposure to ionizing radiation, drug use and vitamin A deficiency in breast milk. Additionally, it has been proven that they are hereditary (autosomal dominant transmission, autosomal recessive, and linked to the X chromosome) [3]. In the case of our patient, association with environmental factors was ruled out. However, a genetic study is required to determine whether it is chromosomal or monogenic.

Prenatal diagnosis can be made by ultrasonography when the eyeballs and lens are absent. Nevertheless, it can often be challenging to differentiate from severe microphthalmia, and in some cases, it can be associated with other brain abnormalities (anencephaly and midline abnormalities). Three-dimensional ultrasonography is superior to two-dimensional ultrasonography for diagnosing these cases [4]. In our patient's case, it was impossible to determine the ocular malformation in the prenatal period, reaching its diagnosis based on clinical findings at birth.

The case presented is classified as isolated congenital bilateral anophthalmia since no other associated anomalies have been detected both in the clinical examination and the imaging studies, and its frequent evaluation is essential to monitor its facial development to initiate timely treatment and of choice for the patient according to their characteristics.

Figure 5. Simple skull magnetic resonance: right microphthalmia and left anophthalmia.



The importance of developing information regarding rare congenital malformations lies in the possibility of

establishing diagnostic and therapeutic protocols to achieve timely intervention, both in the pathology

management and in the impact that this can have on the patient and their family and social environment.

In Ecuador, up to January 2022, 471,205 people with disabilities were registered; of these, approximately 12% had visual impairment, and approximately 24% of this group had prenatal causes [11, 12].

Despite having these epidemiological data, in our country, there is no epidemiological information related to this congenital malformation, and in the search carried out, two cases reported in Ecuador were recorded: one as a case of congenital bilateral anophthalmia [7] and another associated with complex cardiopathy, [11] which leads us to conclude that it is relevant to create a database that makes it possible to naturally determine the rare or infrequent pathologies present in our country to develop health policies that seek to improve the living and health conditions of this affected population.

Conclusions

The case presented leads us to conclude on the importance of prenatal diagnosis to identify probable causes and define patient therapy and emotional and psychological support, both for the child and his family, due to the significant impact of anophthalmia on personal, family and social development.

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Abbreviations

SBP: systolic blood pressure.

Supplementary information

No supplementary materials are declared.

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Does not apply.

Author contributions

Doménica Jijón: Conceptualization, Data conservation, Acquisition of funds, Research, Resources, Software, Writing - original draft.
Tania Carolina Pérez Cárdenas: Conceptualization, Data conservation, Supervision, Fund acquisition, Research, Resources.
Luis Celi Cueva: Conceptualization, Supervision, Fund acquisition, Research, Resources.
Rosario Vaca: Conceptualization, supervision, acquisition of funds, research.
All the authors have read and approved the final version of the manuscript.

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Availability of data and materials

The data sets generated and analyzed during the current study are not publicly available due to clinical case confidentiality.

Statements

Ethics committee approval and consent to participate

Not required for clinical cases.

Publication Consent

The authors have permission for publication from the patient's parents.

Conflicts of interest

The authors declare they have no conflicts of interest.

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