

Case Reports

Anophthalmos

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Abstract

A 2 yr old girl came to our out patient department with sunken eye balls since birth. We did a ultrasound B scan which suggested absence of ocular tissue in both the eyes. Systemic check up showed no abnormality. Our diagnosis is true anophthalmos.

Introduction

Background

T rue or primary anophthalmos is very rare. Only when there is a complete absence of the ocular tissue within the orbit can a diagnosis of true anophthalmos be made. Extreme microphthalmos is seen more commonly. In this condition, a very small globe is present within the orbital soft tissue, which is not visible on initial examination.

Anophthalmia and microphthalmia may occur secondary to the arrest of development of the eye at various stages of growth of the optic vesicle. It is important to recognize microphthalmia because the development of the orbital region, as well as the lids and the fornices, is dependent on the presence of a normal-sized eye in utero.

Anophthalmia may lead to serious problems in a child due to not only the absence of a seeing eye but also the secondary disfigurement of the orbit, the lids, and the eye socket. Early treatment with various expanders or surgery, when necessary, will help decrease the orbital asymmetry and cosmetic deformities in these children.

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Pathophysiology

Anophthalmia occurs when the neuroectoderm of the primary optic vesicle fails to develop properly from the anterior neural plate of the neural tube during embryological development. The more commonly seen microphthalmia can result from a problem in development of the globe at any stage of growth of the optic vesicle.

Proper growth of the orbital region is dependent on the presence of an eye, which stimulates growth of the orbit and proper formation of the lids and the ocular fornices. Commonly, a child born with anophthalmia has a small orbit with narrow palpebral fissure and shrunken fornices.

Case Report

A 2yr old girl came to our out patient department



Fig. 1

with sunken eye balls since birth. We did a ultrasound B scan which suggested absence of ocular tissue in both the eyes. Systemic check up showed no abnormality. Our diagnosis is true anophthalmos (Fig. 1).

Frequency

Congenital anophthalmos is a very rare condition that has a reported prevalence rate of 0.18 per 10,000 births.¹

Mortality/Morbidity

- Growth and development of the bony orbit is directly dependent on outgrowth of the globe.
- Lack of an eye or a microphthalmic eye causes improper development of the orbit.
- A small bony orbit results in hemifacial hypoplasia and will not allow a prosthesis to be fit. The cosmetic deformity can be quite significant.

Race

Racial predilection for this condition has not been reported.

Sex

Sexual predilection for congenital anophthalmos has not been reported.

Age

Anophthalmos occurs in utero and is a congenital anomaly that is present at birth.

Clinical

History

- The defect occurs in utero and is congenital.
- Children with anophthalmos are born with a unilaterally small orbit and no visible ocular tissue within the orbit.

Physical

- Orbital findings
 - Small orbital rim and entrance

- Reduced size of the bony orbital cavity
- Extraocular muscles are usually absent.
- Lacrimal gland and ducts may be absent.
- Small and maldeveloped optic foramen

- Eyelid findings

- Foreshortening of the lids in all directions
- Absent or decreased levator function with decreased lid folds
- Contraction of orbicularis oculi muscle
- Shallow conjunctival fornix, especially inferiorly

- Globe findings

- Globe is completely absent in primary anophthalmos.
- Extremely small and malformed globe is seen in microphthalmos.

Causes

- Idiopathic/sporadic
- Inherited as dominant, recessive, or sex linked
- Chromosome deletion in band 14q22-23 with associated polydactyly
- Trisomy 13-15
- Genetic deletions involving *SOX2*, *SIX6*, and *STRA6*
- Maternal infections during pregnancy (i.e., rubella, toxoplasmosis)
- Often associated with syndromes with craniofacial malformation (i.e., Goldenhar syndrome, Hallermann-Streiff syndrome)

Workup

Imaging Studies

- CT scan or MRI of the head and the orbits
- To assess the presence of an extremely microphthalmic globe
- Bilateral anophthalmos may have an associated absence of the optic chiasma, a diminished size of the posterior pathways, as well as agenesis or

dysgenesis of the corpus callosum.²

- Patients with unilateral anophthalmos may have severe craniofacial anomalies that need to be evaluated by scanning.
- Ultrasound imaging
 - B-scan ultrasound will show a complete absence of ocular tissue in anophthalmos.
 - A-scan ultrasound will show a decreased axial length in microphthalmos.
 - Transvaginal ultrasound can detect eye malformations after 22 weeks' gestation; however, its sensitivity for use in the detection of anophthalmia is not known.

Histologic Findings

Primary anophthalmos is characterized by a complete absence of ocular tissue within the orbit. Extreme microphthalmos is seen more commonly. Histopathological evaluation of orbital contents reveals an extremely small or malformed globe with only rudimentary ocular contents. Overall, extraocular muscles often are absent or markedly decreased in anophthalmia.

Treatment

Medical Care

- Ocular/orbital
- - A solid conformer may be placed in the orbit to stimulate bony orbital growth and to enlarge the orbital cavity in an attempt to attain normal proportions.
- - Progressively increasing the size of conformers will often help to increase the size of the orbit.
 - An ocular prosthesis may be fitted over the conformer to improve the appearance.
- - In patients with unilateral anophthalmos, they and their families should be aware that the target proportions of a reconstructed orbit are not planned to exactly mirror that of a healthy orbit. In all likelihood, a perfectly

normal-looking orbit will not be achieved.

Surgical Care

Inflatable expander

- If conformers are not tolerated or are unsuccessful, an inflatable expander may be placed surgically.
- The expander works best if placed relatively early in life, especially within the first year.
- The inflatable silicone expander is placed surgically deep into the orbit and is accessible by a tube placed in the lateral orbital rim.
- The expander can be gradually filled with liquid (e.g., saline) on a weekly or biweekly basis.
- The advantage of an inflatable expander is that it may allow more rapid and extensive orbital tissue expansion as compared with solid conformers.
- Self-expanding hydrophilic, osmotic expanders³
 - A new possible treatment is the use of self-inflating expanders.
 - Hydrophilic expanders are placed in their dry, contracted state. The expanders then expand gradually to their full size via osmotic absorption of surrounding tissue fluid.
 - This method offers the benefit of controllable self-expansion, without the necessity of repeated fittings of solid conformers or surgical placement of external tubing required for inflatable expanders.
 - Long-term biocompatibility studies have not been completed, but early results are promising.
- Eyelid surgery
 - The increase in the size of a conformer is often limited by shortening of the eyelids in the palpebral fissure, which do

not permit passage of a large conformer. The horizontal length of the palpebral fissure may be increased surgically by performing a lateral canthotomy or cantholysis.

- An additional method to lengthen the eyelids can be accomplished by a combination of skin, mucosal, or cartilage grafts.

- Orbital surgery
 - If conformers and expanders are unsuccessful, the bony orbit may be expanded surgically. This method is preferred in cases of late referral or insufficient orbital volume.
 - The orbit can be expanded in 3 different directions, as follows: laterally, inferiorly, and superiorly.
 - Surgical expansion of the orbit can be accomplished by dividing the bony orbital rim into 3 parts in a stepwise fashion.
 - Cranial bone grafts may be used to augment deficient orbital contours.
 - Lastly, a bicoronal approach through the scalp may be necessary when the orbital roof has to be elevated.

Follow-up

Complications

- Significant cosmetic deformities are often seen if the anophthalmic orbit is not treated early.
- Even after proper treatment using conformers, expanders, or surgical treatment, results are often cosmetically disappointing.
- Fitted prostheses are completely immobile.
- The eyelids often show significant malformations and are shortened and immobile.

Prognosis

- True anophthalmos is considered a

paediatric ocular emergency.

- As the growth and development of the bony orbit are dependent on the growth of the globe, the absence of an eye or an extremely microphthalmic eye impedes the proper development of the orbit.
- The small bony cavity not only is a cosmetic deformity but also does not allow proper fitting of a prosthesis.
- Even with proper early treatment, results are often disappointing in patients with anophthalmos.

Patient Education

- Inform the child with anophthalmos as well as the family that the treatment of this condition could be a long and complicated one.
- Multiple conformers, expanders, and surgical treatments may be necessary to create an adequate-sized orbital cavity for placement of a proper-fitting prosthesis. In addition, multiple eyelid and conjunctival surgeries may be necessary throughout the child's life.

Medical/Legal Pitfalls

- The practitioner taking care of a child with primary anophthalmos needs to recognize the problem immediately and to institute treatment as early as possible to avoid the complications associated with a maldeveloped or small orbit, as well as potential eyelid problems associated with this condition.
- Along with the institution of early treatment of this condition, it is important to educate the child's family regarding the long and complicated process involved in the progressive enlargement of the orbital cavity as well as any potential eyelid surgery.
- Proper education of the parents regarding the complex nature of the treatment of

this condition will help to decrease the chance of legal problems due to the issues that arise from a cosmetic deformity and difficulty with the fitting of a prosthesis as the child grows.

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OH, WHAT A YEAR IT WAS: A LOOK BACK AT MEDICINE IN 2008

The most important advances reported in 2008, was good news for patients with ischaemic stroke they can effectively receive intravenous thrombolysis with alteplase for a longer window of time than had been previously thought. Stem-cell research took a leap forward in two studies. We published a report of the first organ transplant, a trachea, grown from a patient's own stem cells.

For the jetlagged who are reading this at some crazy hour, now you can go back to bed. A new melatonin agonist, tasimelteon, could come to your rescue.

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