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CASE REPORT

The hidden eye

A case of cryptophthalmos

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ABSTRACT

Objective

To report a case of cryptophthalmos.

Methods

This is a report of a case of cryptophthalmos seen at the University of the Philippines-Philippine General Hospital (UP-PGH). Differential diagnosis and management options are discussed.

Results

A 12-day old boy presented with no right palpebral fissure, eyelashes, or eyebrow. The skin overlying the right orbit was continuous from the forehead to the cheek. Under this skin was a 17 mm x 15 mm soft, round, movable mass anterior to the globe. The left upper lid was colobomatous with no eyebrow and fornix. The left cornea measured 9 mm x 6 mm with exposure keratitis and large ulcer. Ultrasound of the right orbit identified the presence of the right globe with normal posterior segment. Cranial computed tomography (CT) showed a cystic mass anterior to the right globe with absent lens. Visual-evoked response of the left eye established nonspecific severe optic-nerve damage, delayed visual-pathway maturation and visual-pathway affectation.

Conclusion

Management of complete cryptophthalmos is difficult and requires separation of the lids and placement of mucous membrane grafts to allow for fitting of prosthesis. Reconstruction of the lid coloboma is necessary to prevent exposure keratitis. Genetic counseling is also important in the management of cryptophthalmos.

Key words: *Cryptophthalmos, Palpebral fissure, Coloboma, Symblepharon, Congenital*

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CRYPTOPHTHALMOS is a rare congenital eye defect in which the lid folds fail to separate in the embryo, resulting in a continuous sheet of skin from the forehead to the cheeks covering the eyes.[1] The skin over the eye is blended with the cornea, which is usually malformed. Also known as ablepharon or complete congenital symblepharon, cryptophthalmos may be unilateral or bilateral.

Its mode of genetic transmission is usually autosomal recessive, present in 15% of cases particularly in consanguineous marriages. It usually affects siblings, and lacks vertical transmission. It affects both genders and has no known chromosomal abnormality.[2]

In this article we describe the second case of cryptophthalmos seen in the Philippines. The first case was reported in 1978 involving an 8-month old male.[3]

CASE HISTORY

A 12-day old baby boy was referred to the UP-PGH because of absent palpebral fissure. Born full-term (3 kg) via spontaneous vaginal delivery to a 28-year-old G3P2, the infant had a right eye noted by the pediatrician to be "hidden" and a left eye that would not close. Maternal history was negative except for a congenital left upper lid coloboma that was repaired in 1980. The mother's paternal half-sister, paternal first-degree cousin, and maternal aunt had eyelid coloboma. The other siblings have no congenital deformities.

Gross examination of the right eye showed no palpebral fissure, eyelashes, or eyebrow. The skin overlying the right orbit was continuous from the forehead to the cheek, with a 17 mm x 15 mm soft, round, freely movable mass underneath. Examination of the left eye revealed coloboma of the left upper lid with no eyebrow and fornix. The bulbar conjunctiva was hyperemic with yellowish discharge. Cornea was 9 mm x 6 mm vertically with a 6 mm x 4 mm ulcer at the inferior area. The rest of the anterior and posterior segment were not seen due to the corneal ulcer. Dazzle reflex to bright light was present.

Systemic workup was negative. Slide smear of the left eye discharge showed gram-positive cocci confirmed on cultures to be *Staphylococcus epidermidis*. Ocular ultrasound of the right eye showed attached retina, clear vitreous, and no intraocular mass. Visual-evoked response of the left eye showed findings consistent with nonspecific severe optic-nerve damage, delayed visual-pathway maturation and visual-pathway affectation. Cranial computed tomography (CT) showed a 1.6 cm x 5.0 cm x 1.6 cm enhancing cystic mass anterior to the right globe with absent lens (Figure 1).

Emergent tarsorrhaphy was performed for the exposure keratitis in the left eye supplemented with intensive antibiotic eyedrops and ointment. A small incision was made on the skin overlying the right orbit; sample tissues obtained revealed disorganized ocular tissue on histopath.

DISCUSSION

Differential diagnosis of cryptophthalmos are anophthalmos and microphthalmos. Both are also rare conditions, arising from abnormal development of the optic vesicle and better differentiated histologically by orbital sections.

In anophthalmos, ocular tissues are completely absent while in microphthalmos some ocular tissues are present but usually not functional. These disorders are frequently associated with reduced orbital volume and eyelid deformities. In those associated with ankyloblepharon, the lid margins are fused together, producing shortening of palpebral fissure with normal conjunctival fornices and cornea.[4] In anophthalmos, ankyloblepharon may be an expression of developmental arrest due to lack of mechanical stimulus for full differentiation. In normal eyes, it may represent a primary aberration of growth dating from the commencement of development of the lid folds.[1]

In cryptophthalmos, the eyeball is usually present but disorganized. The lid muscles, extraocular muscles, and nerve elements in the brain controlling ocular movement are developed.[5] Findings in this patient consistent with these clinical manifestations were the presence of a normal posterior segment confirmed by ocular ultrasound, a normally attached retina with clear vitreous, and no mass. The CT showed the presence of the right globe and histopath revealed disorganized anterior segment.

Ehlers in 1966 reported that one out of five cases of cryptophthalmos was associated with microphthalmos and colobomatous cyst. These presented clinically as bilobular masses beneath the cryptophthalmic skin. The large cyst

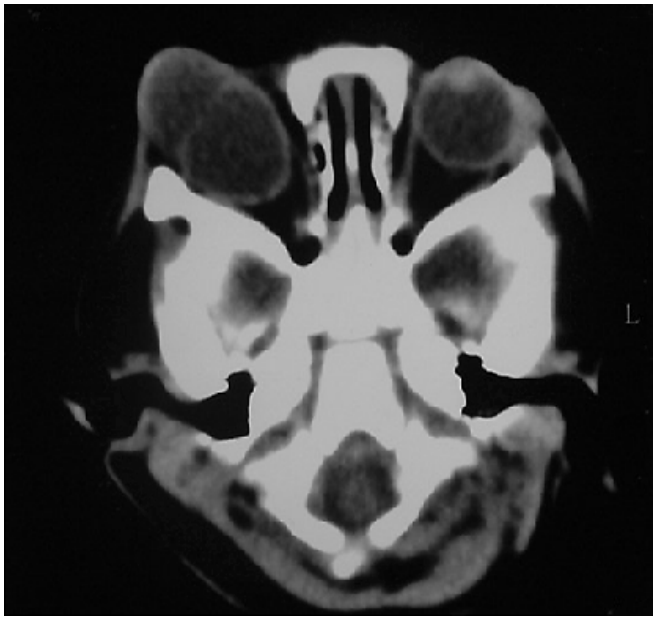

Figure 1. Cranial CT (axial cut) showing a cystic mass anterior to the right globe with no lens. The rest of the structures was normal.

may arise from the small globe, the optic nerve or a noncystic colobomatous malformation of the retina and optic nerve,[6] which was present in the first reported case in the Philippines. When the eyeball was exposed, the thin-walled, fluctuant, and opaque cyst was composed of uveal tissue.[3] In this patient, coronal and axial views of cranial CT showed a 1.6 cm x 5.0 cm x 1.6 cm peripherally enhancing hypodense mass anterior but separate from the right globe, suggesting the presence of a cystic mass.

Some studies have noted through histologic examination that the skin over the globe represented metaplastic change from corneal epithelium rather than a true skin where accessory skin appendages are present. The orbicularis oculi and levator palpebrae superioris muscles are generally present but the other eyelid structures including tarsal plates are rarely evident. The anterior segment of the eye is disorganized,[7] findings consistent with the biopsy of the right orbit of this patient.

In unilateral cryptophthalmos, associated anomalies have been described, including syndactyly of the fingers and toes, genital anomalies, dyscephalies, and ocular anomalies in the other eye such as dermoids, coloboma of upper lids, and microphthalmia.[8] When associated with multiple organ system anomalies, the condition is known as Fraser syndrome.[9] This syndrome should be considered in the differential diagnosis if prenatal echographic examination revealed oligohydramnios with contrastingly voluminous, hyperechogenic lungs.[10] However, ancillary examinations in this patient revealed no other anomalies.

Congenital coloboma of the eyelids can be associated with cryptophthalmos. A defect of the lid margin, one or all four lids may be involved. The defect may vary from a small indentation of the lid border to complete absence of the lid. The entire thickness is usually absent and the edges of the defect are rounded and covered with conjunctiva that unites the lid to bulbar conjunctiva. With large defect, such as in the left eye of this patient, corneal exposure and ulceration can occur. The coloboma is thought to be the result of trauma from amniotic bands or of a localized failure of fusion of the processes of embryonic lid folds.[4] Heredity played little role.

Three theories have been proposed to explain the pathogenesis of cryptophthalmos:

- Failure of mesodermal and ectodermal differentiation resulting in the absence of eyelid folds.
- Intrauterine inflammation resulting in the fusion of the eyelids to the globe.
- Amniotic bands exerting pressure on developing eyelids, causing colobomas with maldifferentiation of the conjunctiva resulting in symblepharon.[11]

Duke-Elder and Ida Mann described two types of this condition: one arising from an initial failure of the lids to form, and the other from a subsequent destruction or absorption of the conjunctivae.[12] Other authors explained the condition on the basis of ankyloblepharon by ankylosis. Currently, many believed that the primary cause is failure of the lids to form or an arrest of development.[13]

Three different forms of cryptophthalmos have been described:

- Complete, as seen in this patient, is the most common. The skin replaces the eyelids, covers the orbit, and connects to the globe so that ocular movement can be seen beneath it.[11]
- Partial or hemicryptophthalmos is seen in about 20% of cases. The lateral portion of the eyelid is normal with fusion of the facial skin to the cornea medially.[11]
- Congenital symblepharon, where the upper eyelid fuses with the globe and the superior cornea is covered by keratinized stratified squamous epithelium.[11]

Management of complete cryptophthalmos is difficult. Extensive reconstructive surgery is involved, including separation of the lids and placement of mucous membrane grafts to allow a conformer or prosthesis to be fitted. Reconstruction of the left eyelid coloboma involves several stages and the use of ear cartilage graft under the skin membrane over the globe to increase mass and act as tarsal plate or the use of mucosal graft to cover the raw surface of the inner side of the upper lid and globe area.

In this patient, the tarsorrhaphy failed on the third week. Repair of the upper lid coloboma by pedicle rotation graft and separation of the skin of the right orbit with placement of mucous membrane grafts were considered.

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