Abortive Cryptophthalmos: A Case Report and a Review of Cryptophthalmos

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ABSTRACT
Cryptophthalmos is a rare, probably autosomal recessive condition 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. Cryptophthalmos has many variants and is usually associated with multiple other congenital anomalies.

This article reports on an isolated case of "abortive cryptophthalmos" with multiple systemic anomalies. The surgical treatment is described and the general features of the cryptophthalmos syndrome and its variants are reviewed.

INTRODUCTION
The term cryptophthalmos was first coined in 1872 by Zehender and Manz to describe a 4-month-old infant with skin passing continuously from the forehead to the cheeks covering rudimentary eyes. However, the earliest description of what is now called cryptophthalmos was actually in 1832 by Saint Hilaire. Since these first reports there have been a total of 86 cases of cryptophthalmos reported in the world's literature.

CASE REPORT
A male infant was born at term by spontaneous vaginal delivery of an uneventful pregnancy, APGARs were 8 and 9 at 1 and 5 minutes respectively. Birth weight was 3666 g and length was 49.5 cm, both appropriate for gestational age. The parents are unrelated and this was their first child.

Ophthalmic examination at 11 days of age revealed a normal right eye. Visual acuity of the left eye was assessed as grossly normal by a crisp blink reflex to bright light. The pupils were equal, round and reactive to light. No relative afferent pupillary defect was present. External examination demonstrated telecanthus. There was a colobomatous defect of the nasal one-fourth of the left upper eyelid (Figs 1-2). A fibrovascular tuft extended from the margins of this defect onto the superior nasal one-third of the cornea, partially occluding the visual axis. The superior punctum was absent and the inferior punctum appeared hypoplastic and stenotic. Anterior segment examination was remarkable for a shortened superior fornix and opaque cornea in area of involvement with the fibrovascular membrane. Temporal cornea was clear. The anterior chamber was deep and well-formed with a normal iris and lens.

The general physical exam was remarkable for poorly formed cartilage in upper helices of both ears, a wide nasal bridge with notching of the left nares, 180° malrotation of the penis, and anterior displacement of the anus.

Diagnostic evaluation included a normal CT scan of the head, normal renal ultrasound, normal karyotype, normal blood chemistry, normal complete blood count and urinalysis.

An exam under anesthesia was performed in conjunction with surgery at 3 weeks. Streak retinoscopy OS was not possible because of partial pupillary occlusion by the fibrovascular tuft on the cornea. A cycloplegic refraction OD was -1.00 sphere. IOPs were 10 to 12 mm/Hg OD and 6 to 7 mm/Hg OS. Corneal diameters were 10.5 mm OU.

The area of cornea covered by the fibrovascular tuft measured 3.5 mm horizontally and 6.0 mm vertically in the superior nasal quadrant. Portable slit-lamp exam confirmed our earlier findings of a normal anterior segment with the exception of involved corneas. Funduscopy exam was normal OU.

Surgery was planned to restore lid structure and function, and to clear the visual axis. Surgery consisted of sharp dissection of the fibrovascular tuft from the cornea, which was deeply embedded within the corneal stroma.
 Due to this corneal thinness, complete dissection of all membrane remnants from the cornea was not possible. The colobomatous defect of the left upper lid was repaired by performing a lateral canthotomy and cantholysis which extended superior temporally in a curvilinear fashion. Skin superior to this incision was freed from underlying tissue by blunt dissection. The margins of the coloboma were incised to expose fresh tissue. A free conjunctival graft from the inferior fornix was used to deepen the shortened
superior fornix. The temporal upper lid was then advanced nasally to close the colobomatous lid defect (Fig 3).

Postoperatively, the child has done well with good cosmesis and good upper lid closure (Figs 4-5). There is some residual corneal scarring and 2 D of "with the rule" astigmatism induced by dissection of the fibrovascular tuft from the cornea. The patient has developed esotropia and amblyopia of the left eye. The child also has chronic epiphora and recurrent conjunctivitis secondary to the defective lacrimal drainage system.

Current management consists of occlusion therapy and a soft contact lens to mask corneal astigmatism. Future considerations will be a punctalplasty, nasal lacrimal duct probing and possible penetrating keratoplasty if amblyopia fails to improve with patching and soft contact lens wear.

DISCUSSION

Since its original description over 100 years ago, there have been sporadic case reports of cryptophthalmos and its many variants. Francois et al. was the first to categorize these variants into three groups:

- complete cryptophthalmos,
- incomplete or partial cryptophthalmos, and
- abortive cryptophthalmos (also described by some authors as congenital symblepharon).5,7,8

Complete cryptophthalmos can involve any number of anomalies of the eye and adnexae.1-20 These anomalies can be the absence of eyelids, or continuous skin from forehead to cheek covering a microphthalmic disorganized eye. The anterior segment is typically very dysplastic with thin cornea fused to overlying skin, a shallow anterior chamber with angle structure abnormalities, aphakia and or absence of the iris. The posterior segment in most cases has been found to be relatively intact. The eyebrows are frequently absent and on occasion hair extends from the temporal region of the scalp into the area of the brow.

Eyelashes, glandular structures of the lids and puncta also are typically not found. The conjunctival fornices are absent.

Incomplete cryptophthalmos represents a milder form of cryptophthalmos with fusion of the lids nasally but presence of a small palpebral fissure temporally. Microphthalmia and disorganization of the anterior segment are common.1,2,5,6,9,14

Abortive cryptophthalmos is the least severe of the three types of cryptophthalmos.4,6 It is characterized by a coloboma of upper eyelid nasally with skin from this defect extending over and adherent to the superior cornea. The upper punctum is frequently absent while the lower eyelid and punctum are usually normal. This differs somewhat from our case in which the lower punctum was hypoplastic and stenotic. Rarely microphthalmos has been reported, but most often the eye, with exception of involved cornea, is found to be normal. There is one case in the literature of a post mortem on a child with abortive cryptophthalmos in which the intraocular contents and structure of the eye, by histopathology, were found to be entirely normal. Our case most closely fits the latter description of abortive cryptophthalmos.

Many theories on pathogenesis of cryptophthalmos have been suggested.6,20 The most legitimate explanation is some degree of failure of lid fold development during embryogenesis. Other theories have included: intrauterine inflammation, amniochorionic bands, defective differentiation of conjunctiva, abnormalities of maternal vitamin A metabolism, and a defect in the process of controlled necrosis in fused structures in embryo such as the digits and the lids.

Cryptophthalmos has been reported as an isolated entity but most often it is described in conjunction with multiple other congenital malformations. Francois, in a review of 46 cases described the cryptophthalmos syn-
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drome consisting most frequently of cryptophthalmos, dysencephaly, syndactyly and urogenital anomalies. Consanguinity is reported in 15% of cases and an autosomal recessive pattern of inheritance is apparent in the majority of cases.

More recently, Thomas et al, on a thorough review of the 86 cases in the world’s literature, developed major and minor criteria for the diagnosis of the cryptophthalmos syndrome (Table). The major criteria are cryptophthalmos, syndactyly, abnormal genitalia and a history of a sibling with the cryptophthalmos syndrome. Minor criteria involve any number of congenital malformations of the nose, ears, larynx, cleft lip or palate, skeletal defects, umbilical hernias, renal agenesis, and mental retardation.

There have been rare reports of other anomalies such as congenital heart defects, basal cephaloceleces, imperforate anus and ascitis.

The diagnosis of cryptophthalmos syndrome can be made on the basis of two or more major criteria and one minor criterion; or one major criterion and four or more minor criteria. Our case meets these criteria for the diagnosis of the cryptophthalmos syndrome based on the presence of two major criteria (abortive cryptophthalmos and abnormal genitalia) and two minor criteria (anomalies of the ears and nose).

REFERENCES