Bilateral Congenital Cystic Eye Posterior to the Lower Eyelid: Case Report

Alt Göz Kapağı Arkasında Bilateral Konjenital Kistik Göz

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ABSTRACT One day after delivery, an infant boy was referred to our clinic with the complaint of bilateral inferior eyelid bluish discoloration and absence of globe in orbital space. External ocular examination showed no globe in orbital space but a mild bulging of the right and left lower eyelid with an area of bluish discoloration. There were no other ocular or non-ocular abnormalities. Systemic evaluation was normal. Magnetic resonance imaging (MRI) confirmed presence of bilateral cystic eye and absence of ocular structures in orbital space. MRI at age of 14 months showed enlargement of the left cystic eye. A clear fluid (5 cc) was aspirated from the cyst to restore the globe size within the lower eyelid. Biochemical analysis of cystic fluid revealed no abnormalities.

Key Words: Congenital, hereditary, and neonatal diseases and abnormalities; eye abnormalities

ÖZET Bir erkek bebek, doğumundan bir gün sonra her iki alt göz kapağında mavimsi renk değişikliği ve göz kavitesinde gözlerinin olmaması şikayetleriyle kliniğimize yönlendirildi. Dış göz muayenesinde, her iki göz boşluğunda gözlerin olmadığı ancak her iki alt göz kapağında mavimsi renk değişikliğinin olduğu bir bölgenin eşlik ettiği hafif bombeleşme tespit edildi. Bunların dışında göz ve komşu yapılarda bir bulguya rastlanmadı. Sistemik muayene normal bulundu. Yapılan magnetik rezonans görüntüleme (MRG), göz boşluğunda iki taraflı kistik göz ve oküler yapıların yokluğunu doğruladı. On dördüncü ayda yapılan MRG'de, sol kistik göz yapısında genişleme tespit edildi. Alt göz kapağı içindeki göz büyüklüğünü düzeltmek üzere kist içeriğinden 5 cc aspire edildi. Kist sıvısının biyokimyasal incelemesinde normal bulundu.

Anahtar Kelimeler: Doğumsal, kalıtsal, yenidoğan hastalıkları ve anomalileri; göz anomalileri

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Congenital cystic eye results from invagination failure of optic vesicle between the second and fourth weeks of gestation.[1,2] It is an extremely rare condition. The clinical appearance suggests an absent eye that is replaced by a thin-walled cyst of varying size. The cyst may have a bluish appearance and is usually centrally placed within the orbit. Associated ocular abnormalities include periocular skin appendages and contralateral microphthalmos with cyst.[3,4] Congenital cystic eye must be differentiated from microphthalmos with cyst. Differentiation may be achieved with computerized tomography (CT), magnetic resonance imaging (MRI), or both.[4,5] Congenital cystic eye, microphthalmos with cyst, and

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microphthalmos with cystic teratoma should be suspected in patients with a small or unrecognizable eye when an orbital cystic mass that is detected by palpation or visualization. However, definitive diagnosis is accomplished with histopathologic examination of the orbit.

CASE REPORT

An infant boy with a gestational age of 38 weeks and a birth weight of 3450 g was delivered vaginally. There were no complications during the pregnancy or the delivery. The parents were healthy and there was no history of serious illness or drug use during pregnancy, except for consanguinity of the parents. There were no developmental malformations in the parents in their family history. The orbits appeared norma, yet no globes were identified in bilateral orbits. There was noinferior cul-de-sacs in the orbital cavities. Infant had only soft masses behind the both lower eyelids. External appearance showed a mild bulging of the right and left lower eyelids with an area of bluish discoloration and translucency (Figure 1). There were no other ocular or non-ocular abnormalities. T2 weighted MRI revealed presence of bilateral cystic eyes and absence of ocular structures (Figure 2A). No relationship was detected between the cyst and the intracranial space. Systemic examination was unremarkable and the karyotype analysis was normal (46, XY).

When the child was 14 months old, the left cyst began to enlarge, causing the lower eyelid to protrude, everting the lower tarsus into the interpalpebral space. On T1 weighted MRI at age of 14 months, there was an enlargement of the left cystic eye (Figure 2B). Five milliliters of clear fluid was aspirated from the cyst in order to restore the globe size within the lower eyelid. Cystic fluid contained Na: 148 mmol/L; K: 4,1 mmol/L; Cl: 142 mmol/L; and Mg: 148 mg/dl; with osmolality of 299 mOsm/kg. Final appearance of the case at age 18 months is shown in Figure 3.

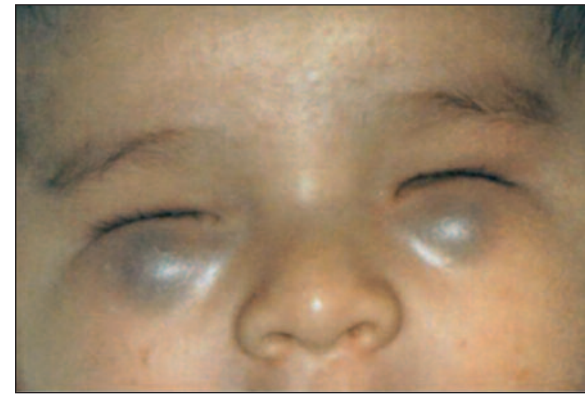

FIGURE 1: Infant had only soft masses beneath the lower eyelids. External appearance was showing a mild bulging of the right and left lower eyelids with an area of bluish discoloration.
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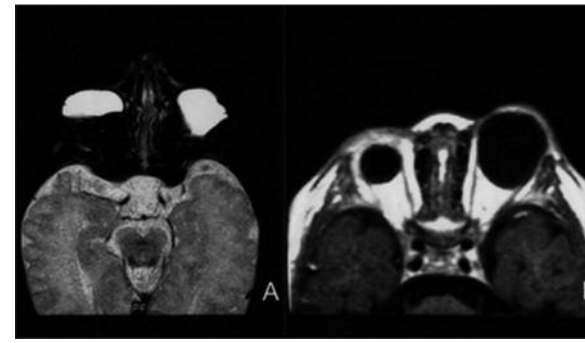

FIGURE 2: Presence of bilateral cystic eyes with irregular borders on T2 weighted magnetic resonance imaging at the age of 3 days **(A)**. Enlargement of the cystic eye on T1 weighted images at the age of 14 months **(B)**.

FIGURE 3: Final external appearance of the case at the age of 18 months.
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DISCUSSION

Developmental abnormilities of the globe include anophthalmia, congenital cystic eye, microphthalmos with cyst, synophthalmia and microphthalmos with cystic teratoma. Cystic lesions of orbit, which account for approximately 10 to 30 % of non-thy-

roid orbital lesions are a heterogeneous group of conditions with various etiologies, presentations and pathologic appearances.[6] They can be divided into two groups as the ones with and without epithelial lining. Epithelial-lined cysts are dermoid cysts and acquired cysts, such as mucoceles. Cysts without an epithelial lining are neurogenic and associated with the primary developmental abnormalities of the globe, such as congenital cystic eye, microphthalmos with cyst, and microphthalmos with teratoma.

Complete or partial failure of invagination of primary optic vesicle gives rise to a congenital cystic eye lined by neuroglial tissue with no evidence of normal ocular structures, such as lens, ciliary body, choroids or retina. Failure of closure of the fetal fissure results in a spectrum of ocular abnormalities, such as choroidal colobomas and microphthalmos with cyst. Some detrimental influences between the 2 mm and 7 mm stages of embryonic development leads to congenital cystic eye, and between 7 mm and 14 mm stages leads to microphthalmos with cyst.[1,2] The time proximity of these two abnormalities is supported by the occurrence of both conditions in the same individual.[7]

Congenital cystic eye is histopathologically similar to the cystic portion of microphthalmos with cyst.[8] There are a few differentiating features between these malformations. In most congenital cystic eyes, the cyst is centrally placed in the orbit or may bulge toward the upper eyelid more. In microphthalmos with cyst, the cyst bulges towards the lower eyelid because the cyst is attached to the inferior portion of globe. Bulging of the upper eyelid with microphthalmos with cyst has also been reported.[9] In our case, bilateral cysts had placed and bulged to the lower eyelids. Histopathologic examination was not performed because we were not permitted to remove the cystic eye.

Congenital cystic eye is rare condition. Duke-Elder reviewed the ophthalmic literature from 1880 to 1960, and found only 16 cases with congenital cystic eyes.[1] Congenital cystic eye can occur with or without other ocular and non-ocular abnormalities. Associated eyelid abnormalities are accessory limb, skin tags, a notch, and periocular dermal appendages on the same side and colobomatous eyelid defect on the opposite side of cystic eye.[3,8,10,11] Reported non-ocular abnormalities are facial clefting, saddle nose malformation of nose, choanal atresia, and malformed sphenoid bone, multiple punched-out lesions of scalp and face agenesis of the corpus callosum, basal cephalocele, electroencephalographic abnormal signs in region of Rolandic area, midbrain deformity, microphallus with hydrocele, hypoconvex fingernails on short stubby fingers, bifid thumb and ventroperitoneal shunts. Non-ocular abnormalities are more common in bilateral involvement,[12,13] Although bilateral congenital cystic eye was present in our case, ocular and non-ocular abnormalities were not detected.

The size of the cyst may vary from 18x10x5 mm to 50x45x45 mm.[3,4] There is no relationship between its size and age. The size of the cyst is believed to the related to patency of the optic stalk.[1] However, Helveston et al. found a non-patent posterior stalk.[10] The sizes of globes were 17x21x23 mm in the right eye and 17x29x21 mm in the left eye at 3 days of age in our case. The left eye was enlarged to 30x45x30 mm at 14 monts of age. Then, aspiration (5 cc) was performed from left eye and eye returned to a normal size. The right eye size did not change during the follow up period.

Although the cause of congenital cystic eye not known definitely, no hereditary tendency has been established yet. It is commonly accepted that environmental factors play a role in the pathogenesis of cystic eye. However, consanguinity detected in the parents of our case of might have been one of the factors contributing to the development of congenital cystic eye.

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