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Microphthalmos with Cyst

—Case Presentation—

Ho Kyun Cho, M.D.

Department of Ophthalmology, Chung-Ang University Medical School, Seoul, Korea

The author has experienced a case of microphthalmos with large orbital cyst in a 4 months old female, that was found at the time of birth. To facilitate fitting a cosmetic prosthesis, the microphthalmos with cyst was removed surgically.
On serial section I could find an area of discontinuation of the sclera that was suspected to be the defective closure of the embryonic cleft. Some aberrant retinal tissue was found in the wall of the cyst, and markedly disorganized ocular tissue forming a tumor-like mass filled the microphthalmic eyeball. In view of these histopathologic findings I could draw the conclusion that developmental failure of the embryonic eyeball and consequential proliferation of the embryonic neuroepithelial cells occured at an early developmental stage causing the formation of microphthalmos with cyst.

Key Words: *microphthalmos, orbital cyst, embryonic cleft, aberrant retinal tissue*

INTRODUCTION

Microphthalmos with cyst is a rare congenital anomaly of the eyeball. This abnomality is caused by improper fusion of the embryonic fissure between the 7mm to 14mm stages of fetal development. A few cases of orbital cyst resulting from the failure of the fetal fissure to close and proliferation of the neuropithelium through the opening have been reported (Wilson et al., 1985; Lieb et al., 1990). A large orbital cyst may interrupt the growth of the eyeball resulting in microphthalmos.

A case of microphthalmos with large orbital cyst is presented. On microscopic examination an area of suspicious defective closure of the fetal fissure and aberrant retinal tissue in the wall of the cyst were shown in the serial section. To my knowledge, this is the second case report of microphthalmos with cyst in Korea (Hwang et al., 1987).

CASE REPORT

An otherwise healthy 4 months old female presented to Chung-Ang University Hospital with a large protruding mass in her left orbit (Fig. 1). This had been noticed since birth. It was a smooth surfaced, purple colored, soft cystic mass, and transilluminated light easily. The microphtalmic eyeball was displaced medially. No iris, anterior chamber, or lens was detectable on examination under general anesthesia.

The patient was the first baby, and didn't have any eventful parental history. The pregnancy had been uncomplicated, and she was born vaginally at full term weighing 3,100 grams. At the time of visit she showed 50 percentiles of overall growth rate. On physical examination, including respiratory, cardiovascular, abdominal, and genitourinary tract, no remarkable findings were noted.

Skull X-ray and computed tomographic scan showed marked enlargement of the left orbit, and the left orbit was almost filled with the cystic mass displacing a small rudimentary eyeball medially (Fig. 2). An extraocular muscle attaching the microphthalmic eyeball was shown. An area of a suspicious defect in the orbital roof was shown, but on injection of the radiopaque material into the cystic cavity there was no communication between cystic and cerebrospinal fluid. The brain itself showed no significant abnormality by computed tomography.

A frontal craniotomy was designed to remove the

Address for Correspondence: *Ho Kyun Cho, Department of Ophthalmology, Pil-Dong Hospital, Chung-Ang University Medical School, 82-1, Pil-Dong 2-Ga, Chung-Gu, Seoul, 100-272 Korea Phone 02-260-2180*

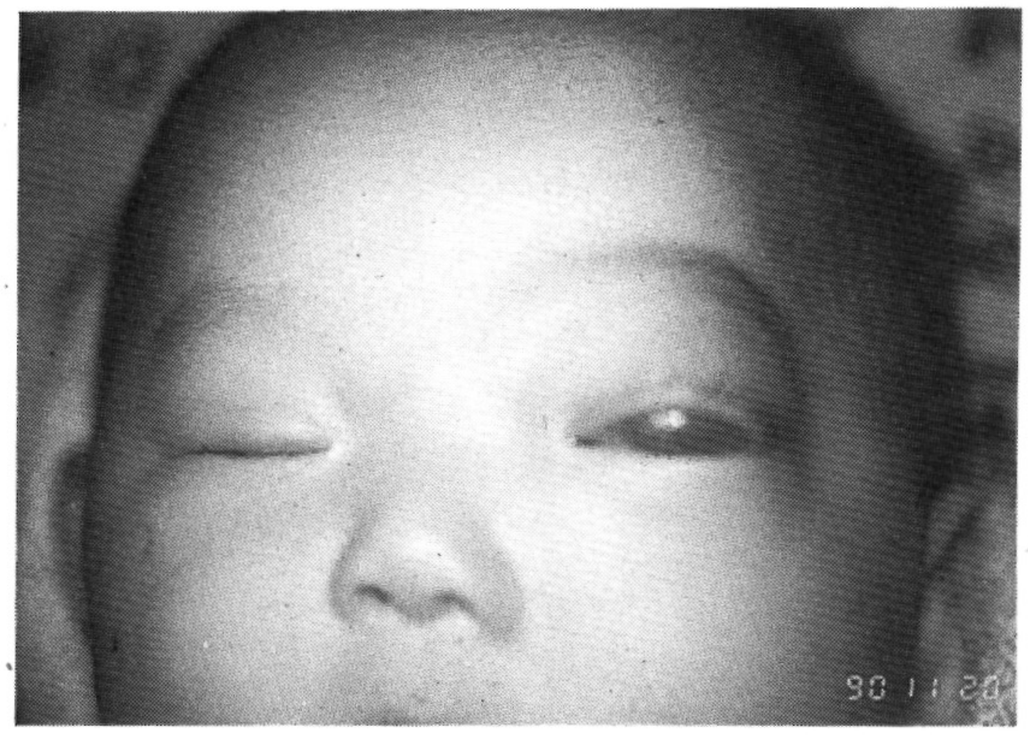

Fig. 1 Four months old female showing purple colored large cystic mass protruding through palpebral fissure in her left orbit.

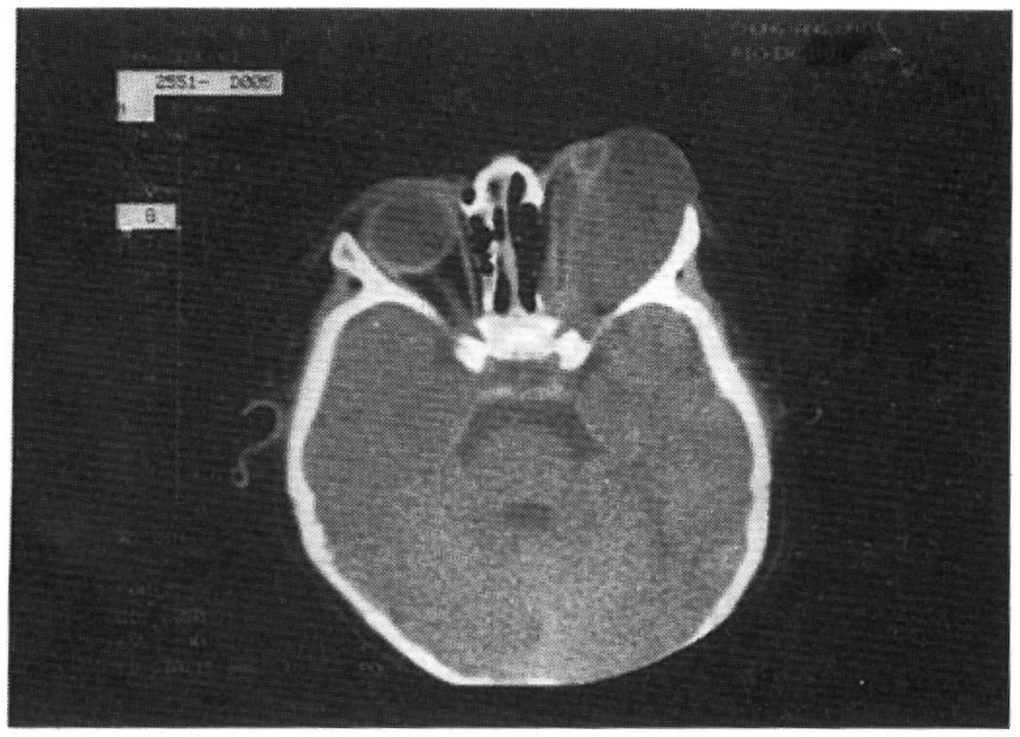

Fig. 2 Computed tomogram of the orbit shows a large cyst with medially displaced microphthalmic eyeball occupying the entire left orbit. An extraocular muscle attaches to the microphthalmic eyeball medially.

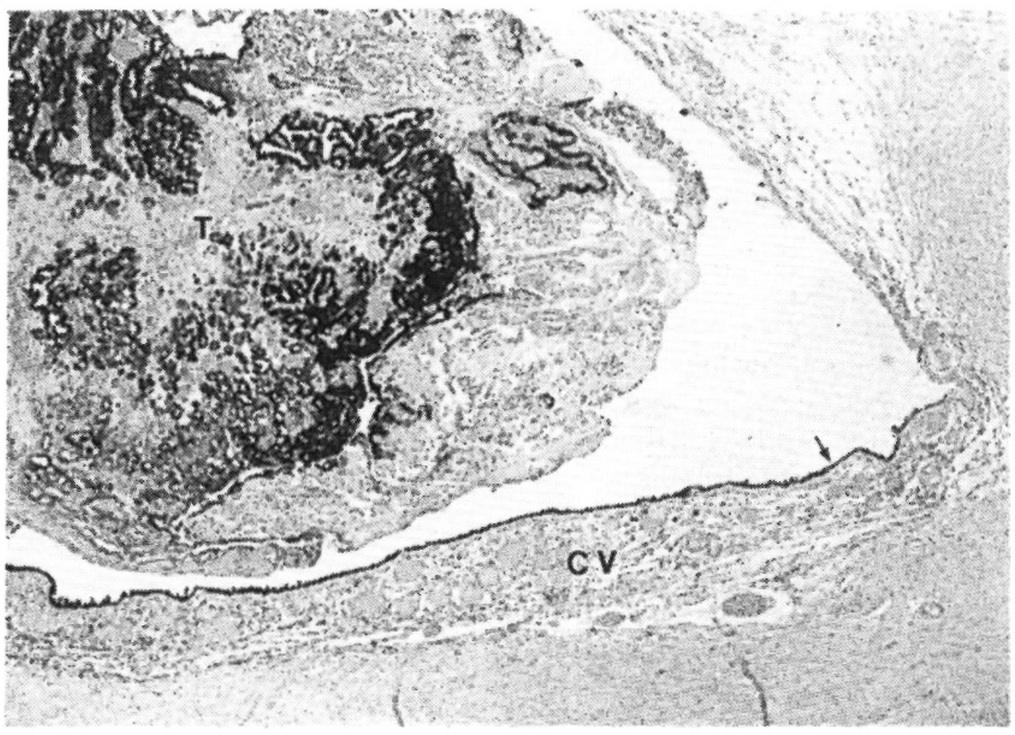

Fig. 3. Markedly disorganized intraocular tissue forming a tumor-like mass in the microphthalmic eyeball (T). A portion of the microphthalmic eyeball shows many choroidal vessels (CV) and one layer of pigmented epithelial cells (arrow) (hematoxylin and eosin stain, ×40).

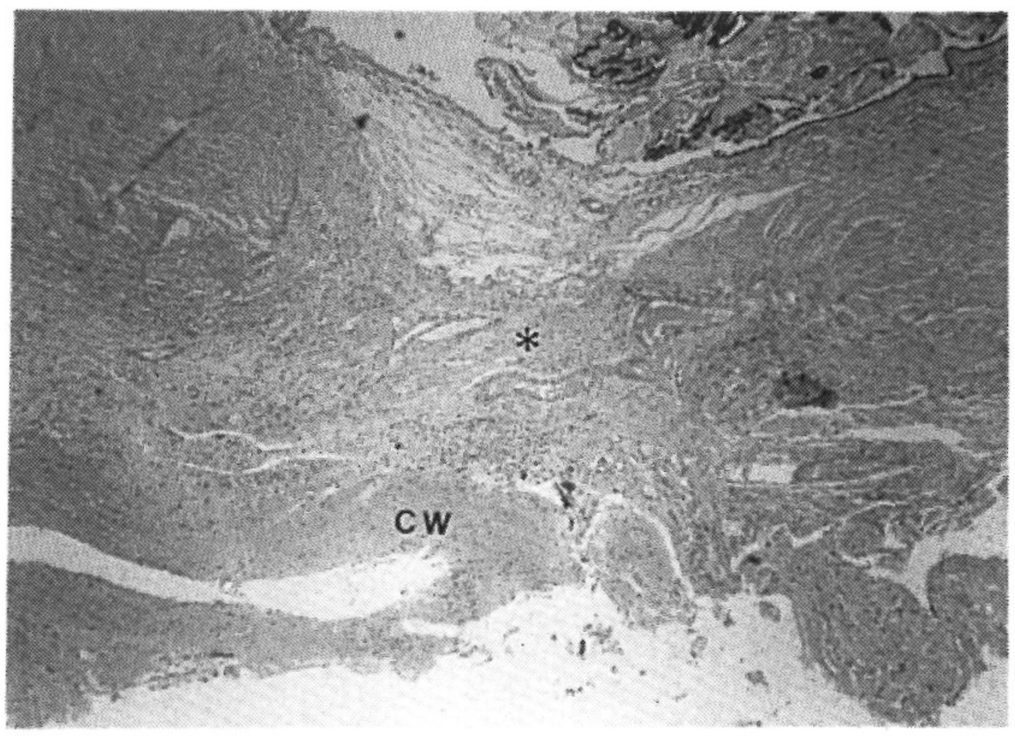

Fig. 4. A suspicious area of the defective sclera (asterisk). An area of the defective sclera composed of collagenous tissue is very similar to that of the wall of the cyst (CW) (hematoxylin and eosin stain, ×100).

orbital cyst by a neurosurgeon. The orbital roof was very thin, but neither bony defect nor intracranial extension of the cyst was noted. The cyst was attached firmly to the orbital periosteum. The cyst was ruptured during removal, and clear fluid flew out. The cystic fluid was aspirated for biochemical study. After complete removal of the ruptured cyst and microphthalmic eyeball the orbital roof was reconstructed with silastic sheet.

On gross examination the excised specimen consisted of a microphthalmic eyeball 6×4mm in size and a collapsed cyst. The specimen was cut serially. The microphthalmic eyeball was almost completely occupied with a large tumor-like mass (Fig. 3). On microscopic examination the tumor-like mass showed many pigments and some areas of well differentiated retinal tissue. Some ciliary processes were seen in the microphthalmic eyeball, but no iris, anterior chamber, or cornea were found. The thick wall of the microphthalmic eyeball was composed of a dense fibrous tissue, and one layer of pigmented epithelium lined the inner wall. The choroid had many blood vessels. An ectopic lens was found in the cystic cavity. The cystic wall was composed of two kinds of tissue; an inner layer consisting of neuroglial cells and an acellular dense fibrous outer layer. In serial section discontinuation of the scleral tissue was found in one portion of the microphthalmic eyeball (Fig. 4). The tissue forming that portion was very similar to that of the wall of the cyst. Some ectopic retinal tissue was found in the wall of the cyst (Fig. 5).

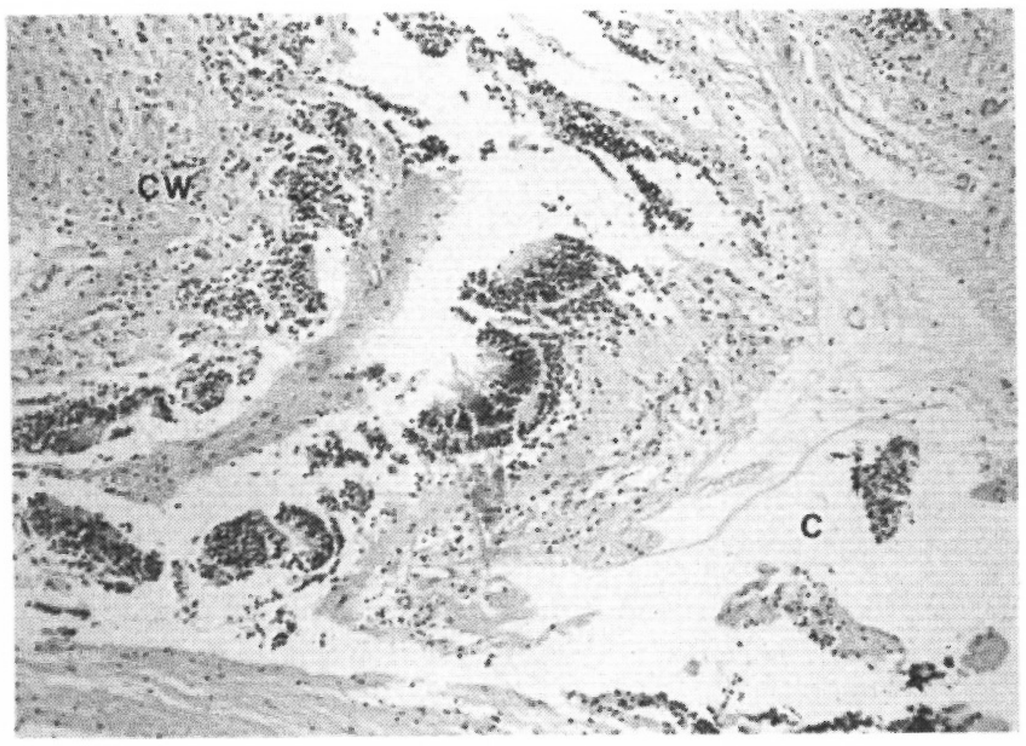

Fig. 5. Aberrant retinal tissue in the wall of the cyst (asterisk). The wall of the cyst (CW) is composed of loose collagenous tissue and primitive neuroepithelial cells. Cystic cavity (C). (hematoxylin and eosin stain, ×100).

The cystic fluid contained protein, glucose, and chloride 428 mg/dl, 64 mg/dl, and 654 mg/dl respectively.

DISCUSSION

A developmental failure of the fetal fissure and resulting proliferation of the neuroepithelial tissue through the defect may cause the formation of orbital cyst (Duke-Elder, 1963). If the developmental failure occurs early in the embryonic stage before or just after the fourth week, congenital cystic eye may develop. Microphthalmos with cyst may occur, if the developmental failure occurs before the sixth week of the embryonic stage (Markley et al., 1969). The size of the resultant cyst varies from small to large enough to occupy the entire orbital cavity. An enlarging cyst may interrupt the growth of the eyeball resulting in a microphthalmic eyeball. In this case I could easily suspect that a huge orbital cyst might interrupt the growth of the eyeball.

The cause of the developmental failure of the fetal fissure is still obscure. Microphthalmos with cyst may associate with systemic and ocular anomalies (Foxman et al., 1984). Most cases occur sporadically, and some have chromosomal abnormalities (Weiss et al., 1987; Guterman et al., 1990). No systemic abnormality was found in this case. And also I could'nt find any familial history or maternal drug use during pregnancy.

On microscopic examination the microphthalmic eyeball was filled with a tumor-like mass. It was composed of markedly disorganized intraocular tissues. The cyst wall was composed of collagenous connective tissue resembling sclera and often interdigitating neuroectodermal element containing retinal photoreceptor cells. This finding was very similar to that of other case (Waring et al., 1976). In serial section there was a suspicious area of defective closure of an embryonic cleft in the wall of the microphthalmic eyeball. This consisted of relatively loose collagenous tissue similar to that of the cyst wall, and that was quite different from adjacent scleral tissue. We can suspect that intraocular neuroepithelial tissue may grow through the site of a defective closure of the fetal fissure and forms aberrant intraocular tissue in the wall of the cyst. In this case relatively well differentiated retinal tissue was found in the tumor-like mass, and also some aberrant retinal tissue was found in the wall of the cyst.

The lens was found inside the cyst. It was difficult to describe the cause of the ectopic location of the lens, but we can suspect that the growing intraocular tumor-like mass might have pushed the lens into the cystic cavity through the defective closure.

The optic nerve fiber is composed of the axons of the ganglion cells. Nerve fibers appear in 19mm human embryo (Ozanics et al., 1982). The cavity of the optic stalk is closed by developing nerve fibers which grow toward the brain from the retinal ganglion cells. A large cyst resulted from proliferation of the neuroectodermal tissue through the defective embryonic fissure might arrest the growth of the embryonic eyeball. And also the developmental disturbance of the inner layer of the neuroectoderm including retinal photoreceptor cells and ganglion cells caused the formation of a tumor-like mass. I can draw the conclusion that an absence of the optic nerve in this case might be due to the developmental disturbance of the retinal cells and ganglion cells. The content of the cystic fluid was similar to that of cerebrospinal fluid except for a high protein concentration, and that was similar to other case (Wilson et al., 1985).

The diagnosis of orbital cyst is not so difficult. A positive transillumination of light provides easy diagnosis of the cyst. Ultrasonography and computerized tomographic findings are also helpful to differentiate a cyst from solid tumor masses (Weiss et al., 1985). An injection of radiopaque material into the cyst gives information about intercommunication between cerebrospinal fluid and the orbital cystic mass.

A non-growing small orbital cyst doesn't need any treatment. But a large cyst causing poor cosmetic configuration or mechanical effect to the eyeball needs surgical removal. Aspiration of cystic fluid may collapse the cyst, but it refills soon after (Weiss et al., 1985; Waring et al., 1976). Total removal of the cyst including the microphthalmic eyeball is needed for a good cosmetic result.

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