A Case Report:

CONGENITAL CYSTIC EYE – A CASE REPORT

Dr.SujataPriyambada
HI-TECH MEDICAL COLLEGE AND HOSPITAL, ROURKELA

ABSTRACT

Congenital cystic eye (Anophthalmia with cyst) is an extremely rare congenital ocular anomaly where a cystic mass replaces normal eyeball secondary to partial or complete failure of invagination of the optic vesicle during foetal development. Here we describe the case of cystic eye in a 20 year old girl associated with microphthalmos in the other eye.

KEYWORDS

Anophthalmia, Congenital Cystic Eye, Microphthalmos

CASE REPORT

A 20 year old girl presented with a mass left eye and diminished vision in right eye. The mass was present since birth and had not increased significantly in size.

Examination revealed a erythmatous soft tissue mass protruding from left eye with absence of normal ocular structures. The mass caused stretching of lower eyelids which was otherwise normal. The mass was non-tender, non pulsatile, non-reducible and cystic in consistency; the size did not change on coughing. The posterior limits of mass could not be ascertained.

Examination of right eye revealed microphthalmos.Systemic evaluation was otherwise normal.There was no history of any such abnormality in the family. Routine haemogram, urine analysis and bioche-mical tests were normal.

Radiographs of the skull revealed a soft tissue mass of the left orbitwith expansion and remodelling of the inferior and lateral bony walls of the orbit. No abnormalities of the chest or abdomen were noted. US of the left orbit demonstrated a mainly cystic lesion with a maximumdiameter of about 5 cm. with few septa within the cyst. No ocular structure was identified in the left orbit.

MRI of the orbits and the head demonstrated the lesion.The lesion was mainly cystic. Thewall of the cyst, the septa, and the solidcomponent demonstrated contrast enhancement after intravenous administrationof gadolinium. The intraconalpart of the optic nerve was not clearlydepicted by MRI, whereas the extraconalpart of the optic nerve and the opticchiasm were demonstrated to havenormal morphology by MRI. Extra ocularmuscles were not recognizable.

The patient was enucleated for cosmetic reasons. At surgerythe orbital mass was excised, and norecognizable eye structure was identified.An orbital prosthesis was placed inthe orbit.

Tissue sent for histopathological examination revealed multiloculated fluid-filled cystic cavity lined by dense fibrous connective tissue.Microscopically, the cyst wall was composed of glial and fibrous tissue. Other eye structures, including thecornea, sclera, lens, vitreous chamber, retina and choroid were absent.

DISCUSSION

Congenital cystic eye, known also as anophthalmos with cyst, is an extremely rare congenital anomaly, first described by Mann in1939. [8]

Pathogenetically, it results from an arrest in the invagination of the primary optic vesicle between 2-mm and 7-mm stages of foetal developmentwhen neuroectodermal elements are not able to develop into future eye structures.[1]

Congenital cystic eye is discovered at the time of birth as a cystic lesion filling the orbit, located behind the upper or lower eyelid without any evidence of an eyeball. The late presentation in this case reveals the lack of concern for the girl child. It is a non-hereditary, unilateral disorder of unknown origin with no gender preponderance. There is no evidence of chromosomal abnormality, although 13q deletion syndrome, known as Orbeli syndrome may be associated.[1,5]The predilection of the congenital cystic eyeball and its associated anomalies for the left side has been highlighted in a number of studies.

Children with this anomaly may be healthy or have associated abnormalities including facial cleft, cleft lip and palate, skin tags, notch, and periocular dermal appendages on the ipsilateral side, eyelidcoloboma on opposite side, choanal atresia, saddle nose, malformation of the nostril, multiple punched-out lesions of the scalp and face, EEG abnormalaties, microphallus with hydrocele, tetralogy of Fallot, flat fingernails on short stubby fingers, and bifid thumb. Associated intracranial anomalies include malformation of the sphenoid bone, agenesis of the corpus callosum, holoprosencephaly, basal encephalocele, microcephaly, midbrain deformities and anomalies of the central visual pathways.[5,6,9,11]

The contralateral eye is normal in most cases, but high myopia, microphthalmos with cyst, non-persistent hyperplastic primary vitreous of the contralateral eye have been reported. Although the lesion does not move like a normal eye, the extraocular muscles may be attached to the mass in a normal or anomalous pattern

On microscopic examination, congenital cystic eye is usually lined by a dense fibrous connective tissue resembling sclera, to which skeletal muscle and adipose tissue are attached. Immature retinal tissue usually lines the inner aspect of the cyst. Because of a developmental failure of the lens placode, the lens is always absent. Posteriorly, an optic nerve-like structure isfound, consisting of fibrous astrocytes without neurons.[1, 5]

Surgical removal of the lesion with placement of a ball implant is usually performed for cosmetic reasons

Differential diagnosis from othercystic lesions of the orbit should bebased mainly on the absence of anyrecognizable eye structure within theorbit, which is filled with and expandedby a cystic lesion.Differential diagnosis includes microphthalmos with cyst (colobomatous cyst), teratoma, lymphangioma, encephalocele, ectopic brain tissue, and optic nerve meningocele.

CONCLUSION

Congenital cystic eye is an extremely rare congenital ocular anomaly where a cystic mass or neuroglial tissue replaces normal eyeball secondary to failure of invagination of the optic vesicle.Although congenital cystic eye is usually discovered at birth, diagnosis is established during surgery demonstrating complete absence of a recognizable eye structure. Imaging modalities, especially CT and MRI,are crucial for the preoperative planning of these lesions and can identify other associated intracranial anomalies. Management is surgical removal of cyst followed by implantation of prosthesis.

REFERENCES

1. Ioannis Tsitouridis, Michael Michaelides, Christos Tsantiridis, Styliani Spyridi, Mary Arvanity.Congenital cystic eye with multiple dermal appendages and

intracranial congenital anomalies.Diagn Interv Radiol 2010; 16:116–121

2. Rice NS, Minwalla SP, Wania JH. Case of congenital cystic eye and accessory limb of the lower eyelid. Br J Ophthalmol 1966;50:409–413.

3. Pillai AM, Rema, Sambasivan M. Congenital cystic eye—a case report with CT scan. Indian J Ophthalmol 1987; 35:88–91.

4. Pasquale LR, Romayananda N, Kubacki J,Johnson MH, Chan GH. Congenital cysticeye with multiple ocular and intracranialanomalies. Arch Ophthalmol 1991;109:985–987.

5. Chaudhry IA, Shamsi FA, Elzaridi E, Arat YO, Riley FC. Congenital cystic eye with intracranial anomalies: a clinicopathologic study. IntOphthalmol 2007; 27:223–233.

6. Gupta R, Seith A, Guglani B, Jain TP. Congenital cystic eye: features on MRI. Br J Radiol 2007; 80:e137–140.

7. Gupta P, Malik KP, Goel R. Congenital cystic eye with multiple dermal appendages: a case report. BMC Ophthalmol 2003; 3:7.

8. Mann I. A case of congenital cystic eye.TransOphthalmolSocAust 1939; 1:120–124

9. Waring GO 3rd, Roth AM, Rodrigues MM. Clinicopathologic correlation of microphthalmos with cyst. Am J Ophthalmol 1976; 82:714–721.

10. Guthoff R, Klein R, Lieb WE. Congenital cystic eye. Graefes Arch ClinExpOphthalmol 2004; 242:268–271.

11. Hayashi N, Repka MX, Ueno H, IliffNT,Green WR. Congenital cystic eye: report of two cases and review of the literature. SurvOphthalmol 1999; 44:173–179.

12. Mridula Mehta, NeelamPushker, SeemaSen, Sanjay Sharma, Kamlesh Gupta, Mandeep S. Bajaj, and SupriyoGhose.**Congenital Cystic Eye: A ClinicopathologicStudy.**Journal of Pediatric Ophthalmology and Strabismus Vol. 47 e1–4

13. Sen R, Pratap A, Agrawal A, Adhikary S, Pandey S R Isolated accessory limb of lower eyelid with multiple dermal appendages Singapore Med Case Report J 2008; 49(4) : e93

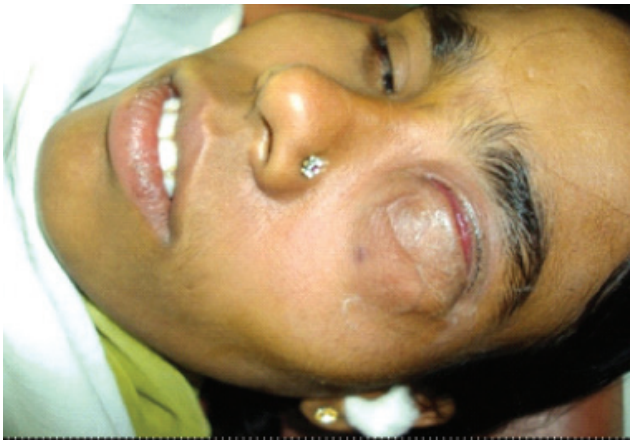

FIG 1 : RE MICRO-OPHTHALMIC WHILE LE PRESENTS AS CYST

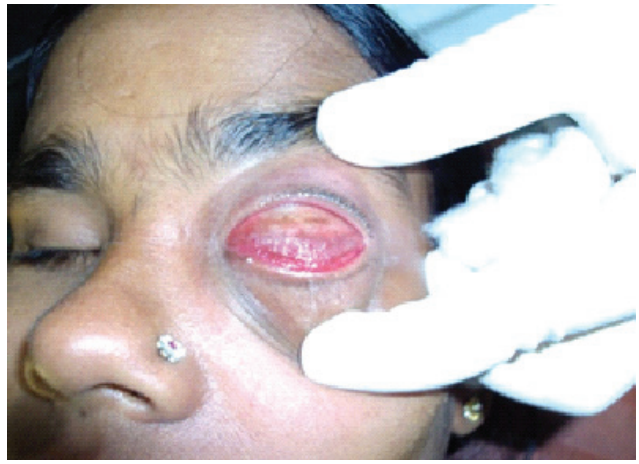

FIG 2 : CYST IN RE REPLACING NORMAL OCULAR STRUCTURE