Congenital Cystic Eye: A Clinicopathological Review

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SUMMARY

Congenital cystic eye is an extremely rare congenital anomaly that results from failure of the invagination of the primary optic vesicle between the 2mm and 7mm stage of fetal development in which the globe is replaced by a cyst.

Management is multidisciplinary involving but not limited to the

Ophthalmologist, Paediatrician, Radiologist, Histopathologist, an ocularist and a counsellor. Surgical intervention is usually carried out for cosmetic reasons and to confirm the diagnosis. We present the case of a seven-month female infant who presented with right orbital mass since birth and a normal left eye. She had excisional biopsy done and histology confirmed the diagnosis.

Keywords: Congenital, cystic eye, excisional biopsy

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Introduction

Congenital cystic eye also known as anophthalmos with cyst is a condition in which the globe is replaced by a cyst. It is an extremely rare congenital anomaly that results from failure of the invagination of the primary optic vesicle between the 2mm and 7mm stages of fetal development[1]. The cyst represents the primitive optic vesicle that failed to undergo differentiation into its adult components. We present the case of a seven-month old female infant with congenital cystic eye who underwent excisional biopsy for cosmetic reason and confirmation of diagnosis.

Case Presentation

We report the case of a seven-month old female infant who presented to the eye clinic of the University of Maiduguri Teaching Hospital in October 2017 with right orbital swelling since birth. The swelling was said to have gradually increased to its present size. There was no history of swelling in any other part of the body. There was no associated history of irritability or body weakness. The mother attended antenatal care at General Hospital Askira from the seventh month of pregnancy and had an uneventful spontaneous vaginal delivery. The baby had normal developmental milestones for age and is fully immunized for age. The baby is the first child of both parents.

Examination showed a healthy looking child with an obvious right orbital mass protruding between the lids. The mass was round, cystic, non-tender, non-reducible, non-pulsatile and covered with skin. It was pedunculated and making a notch on the upper lid (figure 1).

figure 1: pedunculated cystic mass

It measures about 5x5cmx6cm with excoriations of the skin that was in contact with the right cheek. No eyeball was seen in the orbit. The left eye was normal. The child was also reviewed by the paediatric neurology team and had normal neurological findings. A diagnosis of right congenital cystic eye was made. Parents were counselled and patient was prepared for excisional biopsy.

Her haematological and biochemical parameters were normal. Skull X-ray requested was not done. Other imaging modalities like Magnetic Resonance Imaging (MRI) was not available at that point in time as the machine was undergoing maintainance. Excision was performed under general anaesthesia. The mass was excised from the base of the stump after securing haemostasis. The specimen was immediately sent to the laboratory unfixed for histopathological analysis. Postoperative period was uneventful.

Histopathologically, the findings on macroscopy was a nodular firm to cystic mass covered by negroid skin measuring 5x3.5x3cm, cut surface appear solid grey white with tiny cystic and focal brown areas. Microscopy showed a partly cystic tissue composed of haphazardly arranged intraocular tissue that is surrounded by adipose tissue. The cyst wall is lined by pigmented epithelial cells (figure 2).

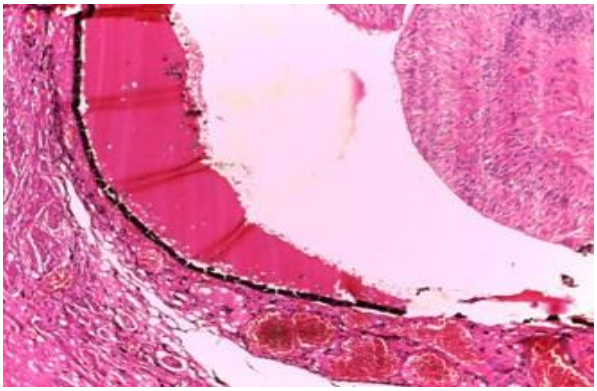

figure 2: histological section of the cyst wall lined by pigmental epithelial cells (H and E, X100)

The tissue elements include epithelial cells with extracellular melanin pigments, neural tissue, nerve trunks and dilated and congested vascular channels surrounded by fibro-collagenous stroma and adipose tissue (figure 3).

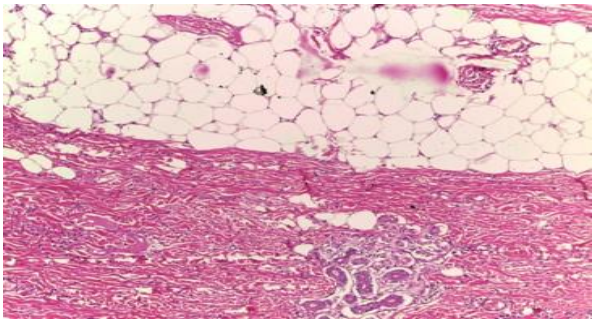

Figure 3: Histological section showing epithelial cells with extracellular melanin pigments, neural tissue, nerve trunks and dilated and congested vascular channels surrounded by fibrocollagenous stroma and adipose tissue (H and E X100)

Other eye structures like the cornea, lens, vitreous and choroid were absent. There was no evidence of malignancy. A diagnosis of right orbital tissue hamartoma was made.

Discussion

Congenital cystic eye is a rare congenital anomaly that results from failure of the invagination of the primary optic vesicle between the 2mm and 7mm stages of fetal development[1]. The orbit therefore contains a cyst that replaces the normal eyeball. The exact aetiology is not known and no hereditary tendencies or abnormalities during pregnancy or perinatal period have been described[2]. Our patient was found to have complete absence of the eyeball in the orbit both clinically and on histopathology, the orbit was replaced with the cystic mass. This confirms the diagnosis of congenital cystic eye. Congenital cystic eye is usually unilateral[1,2] as in our case, although bilateral case have been described by Goldberg et al[3]. A case of unilateral left anophthalmos with orbital mass and a contralateral normal eye reported in Benin, Nigeria[4] is similar to our case. The histology, however was not reported. Failed closure of the fetal fissure during the 7mm-14mm stages of fetal development results in the much more common microphthalmos with cyst[5]. In microphthalmos with cyst (colobomatous cyst) a small but recognizable eye that contains normal eye structures (cornea, iris, ciliary, body, lens, vitreous cavity, retina and choroid) is found[5]. The cyst here arises from failed closure of the fetal fissure, corresponding to the 7mm and 14mm stages of fetal development; it usually enlarges inferiorly, displacing the lower eyelid[5]. Imaging studies like ultrasonography, computed tomography (CT), and magnetic resonance imaging (MRI) are also helpful in making a diagnosis of congenital cystic eyeball and also in excluding intracranial abnormalities. However, diagnosis is only confirmed by histology.

Congenital cystic eye may occur in isolation or with other malformations[1,2,6]. These malformations could be ocular or non-ocular. Our patient was found to have a normal contralateral eye and no other abnormalities of the body. The contralateral eye is normal in most cases, although a case of high myopia[7], a case of microphthalmos with cyst[8], and a case of non-persistent hyperplastic primary vitreous[9] of the contralateral eye have been reported. Non-ocular abnormalities associated with congenital cystic eye include multiple dermal appendages[2], agenesis of the corpus callosum[1] and microcephaly[10].

Management of congenital cystic eye is multidisciplinary and involves but not limited to the Ophthalmologist, Paediatrician, Radiologist, Histopathologist, an ocularist and a counsellor. Surgical intervention is usually carried out for cosmetic reasons and also to confirm diagnosis. Our patient is being followed up with a view to obtaining adequate globe size and the future use of ocular prosthesis to obtain good cosmetic result. We have also advised the parents on the need to have a brain Magnetic Resonance Imaging (MRI) done to exclude any associated intracranial abnormalities even though clinically the patient has no neurological abnormalities.

Conclusion

In conclusion, congenital cystic eye (anophthalmia with cyst) although rare should be suspected in cases where a cystic swelling is present at birth in the orbit with no recognizable eyeball. Diagnosis is confirmed by absence of an eyeball at surgery and histopathology.

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