MICROPHTHALMOS WITH AN ORBITAL CYST: A CASE REPORT

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Abstract: To present a case of bilateral microphthalmos with a unilateral intraorbital cyst with calcification on B scan and CT scan.

Keywords: Microphthalmos, Intraorbital Cyst, Retinoblastoma.

Introduction

Microphthalmos is a rare condition that involves the overgrowth of the inner layer of the optic cup [1]. As a result, the eye is smaller than usual with recognizable eye elements such as retina and lens [1]. In addition, defective closure of embryonic fissure during 6-7 week of gestation leads to microphthalmos associated with an orbital cyst [2].

The prevalence of microphthalmos is 1.4-3.5/10000 births, and cysts are seen in 0.3-0.6/10000 cases [3]. The cysts associated with microphthalmos are usually unilateral and seen within the first few months after birth [3,4].

There are three categories of cysts associated with microphthalmos described by Duke Elder 1) a small cyst not apparent clinically with a relatively normal eye 2) a deformed eye with a prominent cyst 3) a larger cyst that is big enough to push the globe backwards making the globe not visible [5].

Sometimes microphthalmos is associated with other systemic abnormalities, including cardiac and renal defects [1,3].

Case Presentation

A 3-year-old girl presented with right eye swelling since birth (Figure 1). Initially, the swelling was minimal and involved the inferior part of the eye. It slowly progressed to involve the entire eye.

On examination, there was marked right eye proptosis (> 4mm). However, the mass was covering the entire globe and details of anterior and posterior segment of the eye were not visible with no perception of light in the right eye.

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The left eye was microphthalmic with counting fingers close vision. Anterior segment examination showed iris and lens colobomas.

There was no associated fever, lymphadenopathy, or other signs of infection or inflammation on systemic examination and labs. Both her parents were first cousins.

The patient had been seen by multiple ophthalmologists within the past month and ultrasound of the eye (A and B scan), CT orbit and an MRI of the orbit and brain had been performed. The A and B scans showed a mass occupying both anterior and posterior segments with a small eye and some calcification in the posterior segment. The left eye on Bscan was noticed to be small with 2 small cysts (Figure 2).

MRI right orbit showed an eye with a deformed structure and a mass measuring 3.2 x 4.1x 2.9 cm in size with a hemorrhage inside the mass (Figure 3a and b). Both CT and MRI showed a suspicion of retinoblastoma in the right eye. There was no intracranial involvement on either side. The patient had been advised exenteration previously.

Based on our clinical examination and investigations we did not suspect a retinoblastoma. There was slow growth of the mass and the contralateral microphthalmic eye gave a clue that the affected eye may also be microphthalmic with an intraorbital cyst. All lab tests were done, and a pediatric opinion was taken for any systemic signs and symptoms, which turned out to be normal.

We planned surgical exploration and removal of the mass. During surgery, a large cyst was seen surrounding a small rudimentary microphthalmic eye that was completely adherent to the cyst. There was oozing of clear fluid from the cyst. The eye was eviscerated, and ball implant with scleral shell was placed.

The large orbital cyst in this patient has played an essential role in socket expansion thus, the socket was big enough to hold a conformer.

Discussion

There are no concrete guidelines for the management of cysts associated with microphthalmic eyes [1]. Treatment depends on

the cyst's size, eye's visual potential, age of presentation, orbital volume and the growth pattern of the cyst [4,6]. In cases of small cysts, they can be aspirated and observed with a high chance of recurrence [4]. In slightly larger cysts, they can be excised completely. In severe cases, cyst removal can involve enucleation and replacement of orbital volume [4]. In our case, the cyst was very large, occupying the entire orbit, and progressively increasing in size in a blind eye. The cyst ruptured with oozing of clear fluid, and microphthalmic eye was completely enclosed by the cyst that required evisceration. It is the procedure of choice in eyes with severe microphthalmia with no visual potential and the presence of a large cyst[4].
Another factor that made the surgery urgent was the sudden increase in the size of the mass and calcification on CT, making it more suspicious of malignancy.

Our patient had bilateral microphthalmic eyes and a unilateral cyst. Unilateral cysts are much more common than bilateral, and one case series showed that out of 150 cases, only one-third were bilateral [2]. Specks of calcification have been reported in other cases [2].In our patient, the globe was surrounded by the cyst making the eye invisible, which has been documented in other studies, whereas in other cases, the globe may be visible [3,7].
It may not be easy to identify the intraorbital cyst clinically, and this was the scenario in our case [3]. However, ultrasound, CT and MRI have all been used to help with the diagnosis, especially in cases where clinical signs are not very suggestive[3].
Ultrasound A and B scans may prove to be diagnostic, but, in our case, similar to some other cases, they did not yield any information [6].
Since there were no other systemic associations in the child, the diagnosis was more difficult as it was an isolated malformation without systemic association. Such cases have been reported in the literature that could not be diagnosed with ultrasound or a CT scan [6]. In our patient, having a microphthamic contralateral eye on an ocular exam gave us a clue, and surgery proved us correct.

Conclusion

Microphthalmos with an intraorbital cyst is not a very rare condition and anything with calcification on scans doesn't necessarily point to a retinoblastoma. Careful history and examination are essential to exclude malignant causes.
Genetic disorders may be associated with these cysts, and genetic counselling is crucial. In addition, regular pediatric examination and ophthalmological exams for any recurrence and follow-up for the other eye are critical.

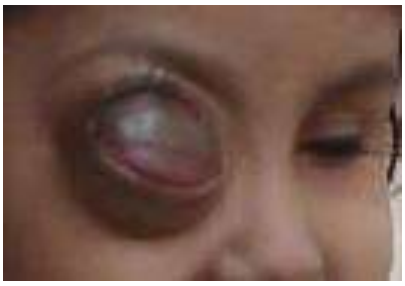

Figure 1: 3-year-old presenting with right eye proptosis

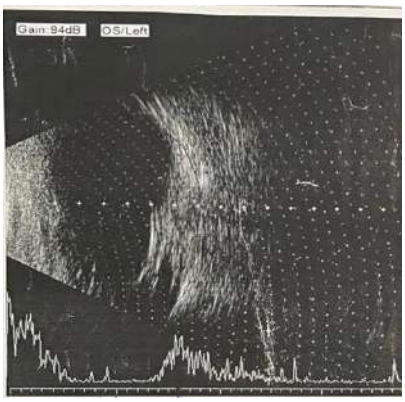

Figure 2: Left eye B scan shows a small eye

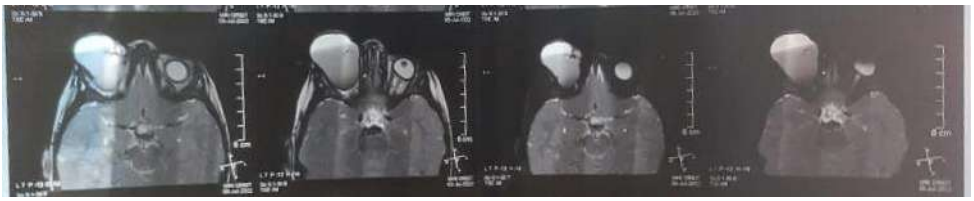

Figure 3a: MRI scan showing the mass in the right orbit

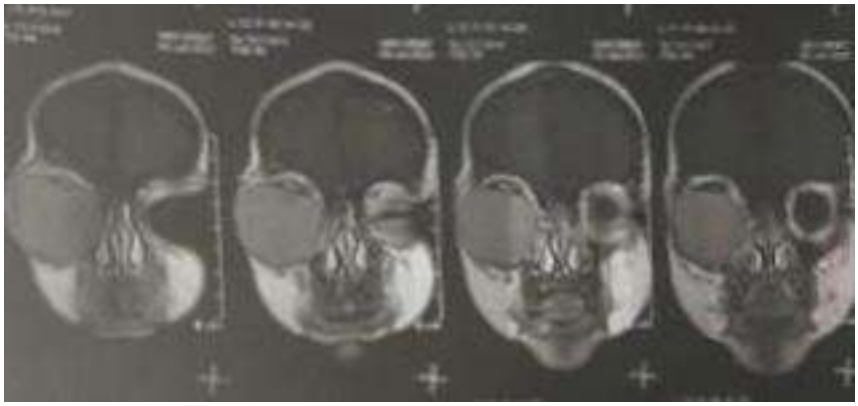

Figure 3b: MRI scan showing the mass in the right orbit

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