Concurrent retinoblastoma and morning glory disc anomaly in a 9 month old baby: a case report

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ABSTRACT

Concurrent occurrence of retinoblastoma and other ocular anomalies is rare. Appearance of leukocoria in the other eye usually signals bilateral disease although this is not always the case. This emphasises the need of careful examination always. We present a case of retinoblastoma and morning glory disc anomaly in a baby. Both conditions had leukocoria in each eye, which was noted at different stages of the clinical evaluation. Although our patient did not have other associated features, this scenario requires distinct multi- disciplinary approach for management of each of the conditions and any accompanying clinical disorders.

Key words: Morning glory disc anomaly, Retinoblastoma, Concurrent

INTRODUCTION

Retinoblastoma (RB) is the most common intraocular malignancy in children. It mostly affects children below 5 years[1]. It can be unilateral or bilateral. The presenting signs include leukocoria, strabismus, an inflamed eye, hyphaema, glaucoma and proptosis in advanced cases[2]. Most of the affected persons usually have no other congenital ocular anomalies and this association is rare[3]. We present a rare case of a baby who had concurrent presentation of retinoblastoma and Morning Glory Disc Anomaly (MGDA).

CASE REPORT

A 9 month old girl presented at Kenyatta National Hospital (KNH) with history of a white reflex in the right eye since 2 months of age with no squint, redness or proptosis of the eyes. There was no history of blepharospasms, tearing or photophobia. She had no history of ocular trauma or surgery. Antenatal, birth and developmental history was normal. Family history of retinoblastoma was negative. On examination, she had normal general exam findings, no lymphadenopathy, bone, scalp or abdominal masses. Vision in the right eye was no perception of light, while she could fix and follow light in the left eye. Extraocular muscle motility was free in both eyes. The corneal reflex test (Hirschberg) was central in both eyes. The anterior segment of the right eye had leukocoria while on the left eye it was normal. On indirect fundoscopy in the clinic, a white reflex was seen in the right eye, while in the left eye a large disc, with peripapillary atrophy and abnormal vessels were noted. The white reflex was clearly visible by torch examination as shown in Figure 1. B scan ultrasound of the right eye showed hyperechoic vitreous mass filling more than half of the vitreous cavity that was persistent on reduced gain (Figure 2). A clinical diagnosis of retinoblastoma of the right eye and disc anomaly left eye was made. Examination under anaesthesia (indirect ophthalmoscopy and Retcam imaging) of the right eye showed features consistent with retinoblastoma and the optic disc was inaccessible. In the left eye, there was no tumour but a large excavated disc with peripapillary atrophy and radial distribution of the retinal vessels as shown in Figure 3. The patient underwent myoconjunctival enucleation of the right eye and prosthesis fitting. Histology of the right eye confirmed retinoblastoma with no high risk features (no optic nerve, sclera or choroidal invasion) (Figure 4). The patient was planned for regular examinations under anaesthesia and follow up by the paediatric ophthalmologist. She was referred for evaluation by the paediatrician and neurologist in view of systemic association of MGDA. On follow-up post right eye enucleation, the left eye also revealed a white reflex on uniocular fixation (Figure 5).

Figure 1: White reflex (Leukocoria) in the right eye

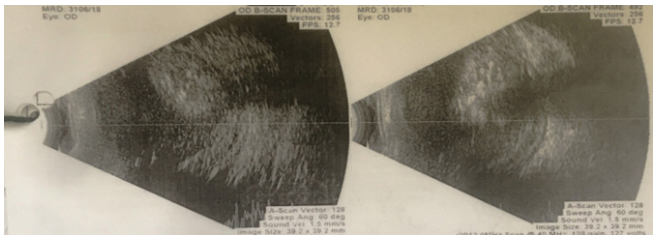

Figure 2: B scan ultrasound of the right eye

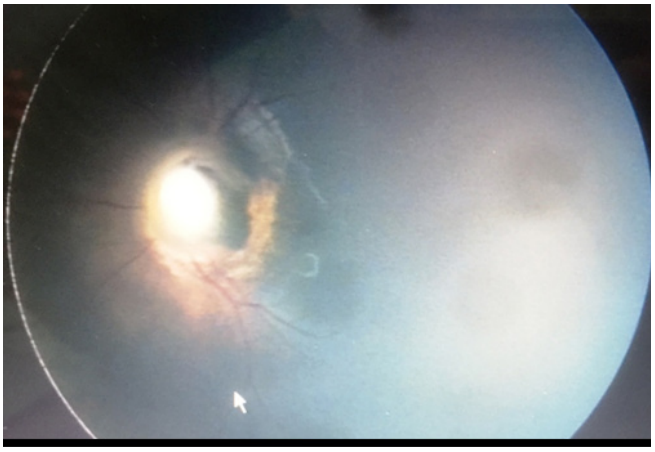

Figure 3: Retcam images of the left eye showing radial pattern of vessels around an excavated disc and peripapillary atrophy

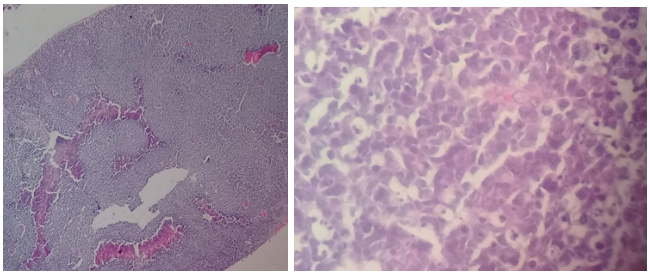

Figure 4: Histology slides of the right eye showing features consistent with retinoblastoma

Figure 5: Post RE enucleation follow-up; Left eye shows white reflex due to the disc anomaly

DISCUSSION

Retinoblastoma normally presents in isolation. Rarely, it may be associated with other ocular anomalies such as persistent foetal vasculature and retinopathy of prematurity[4,5]. In our search, we found only one case report in literature of simultaneous presentation of retinoblastoma with MGDA[6].

Though a rare occurrence, the combination of these two conditions poses a peculiar scenario in the management of the child, which we wish to highlight. Firstly, in the absence of genetic testing for retinoblastoma in our setup, there is a 15% likelihood that the child may have germline mutation with the risk of developing tumours in the left eye, second cancers later in life and passing the mutation to her offsprings. This calls for life-long eye examination and follow-up for herself and her offsprings[7].

On the other hand, MGDA which is usually unilateral but can be bilateral, has both ocular and systemic associations. It was first described in 1970 by Kindler, having a characteristic resemblance to the morning glory flower with features of an enlarged excavation of the optic disc, abnormal retinal vascular pattern, annular pigmentation surrounding the nerve head, and a characteristic glial tuft[8]. It is thought to result from sporadic embryological maldevelopment of lamina cribrosa and the posterior sclera[9]. In the eye, there is risk of associated strabismus, microphthalmos, afferent pupillary defects, cataract, coloboma of the crystalline lens, serous retinal detachment in 30% of the cases and choroidal neovascularization. Vision is generally poor with only a third of the patients attaining normal vision[10]. Facial abnormalities such as hypertelorism, cleft lip and palate have also been reported. Systemic association include central nervous system malformations such as abnormal narrowing of cerebral arteries (Moyamoya disease), encephalocoeles and corpus callosum agenesis[11,12]. Others include abnormalities of the endocrine, respiratory and renal systems[11].

Although our patient did not have obvious ocular or systemic anomalies at the time of initial management, it is important to emphasise the need for close follow-up and comprehensive evaluation by a multidisciplinary team of experts for early mitigation of any untoward events related to either the retinoblastoma or the MGDA. In addition, more than one ocular condition may coexist hence the need for complete and bilateral eye examination.

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