Case Report

Masquerade Syndrome: Retinoblastoma presenting as Sympathetic Ophthalmia

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Purpose: Masquerade syndromes are a group of ocular diseases that may mimic chronic intraocular inflammation, they may be benign or malignant. We report a case of retinoblastoma presenting as sympathetic ophthalmia: malignant uveitic masquerade syndrome.

Material and Methods: A 6 year old female presented with trauma in the right eye following which she developed uveitis in the left eye two months later. She was diagnosed as a case of sympathetic ophthalmia and enucleation of right eye was done. The histopathology suggested it was a retinoblastoma (RB). Since the child's history was not suggestive of RB and her age was old for RB we requested the laboratory to review the slides again. A board of pathologists reviewed the slides, concluding that it was a RB undoubtedly.

Result: Based on this, she was diagnosed as a case of malignant uveitic masquerade syndrome. Patient was referred to advance oncology unit for further management of RB.

Conclusion: Refractory uveitis in a young child should be thoroughly evaluated for malignant uveitic masquerade syndromes.

A group of diseases mimicking intraocular inflammation and uveitis are termed as masquerade syndrome,[1] which may be classified as benign or malignant. Sympathetic ophthalmia is an autoimmune granulomatous intra-ocular inflammation occurring most commonly within one year of surgery or trauma[2]. The injured eye termed as the excited eye while the fellow eye that develops inflammation without any obvious reason is called as sympathizing eye.

Retinoblastoma is the most common pediatric intraocular tumor occurring classically prior to 5 years of age. It usually presents as leukocoria, strabismus or decreased vision. Only 1 – 3% present of retinoblastomas present as intraocular inflammation[3].

We present as case report of a 6 year old girl in which retinoblastoma presented as sympathetic ophthalmia, thus masquerade syndrome.

CASE REPORT

A six year old female child first presented with chief complaint of painful loss of vision in her right eye (OD) for a few days after trauma with stick. On examination her OD had large hyphema with markedly raised intraocular pressure (IOP) (Fig. 1.A) and left eye (OS) was within normal limits. Paracentesis of OD was done and blood was sent for gram staining and culture, which revealed no growth or micro-organism. Two months later the child presented with complaint of no vision in her OD and painful reduced vision and photophobia in OS. Her general health was good with no history of fever and review of systems was normal. On examination her visual acuity (VA) was no perception of light (NPL) in OD and not recordable in OS due to photophobia. On slit lamp examination her OS had multiple keratic precipitates, cells in anterior chamber (AC) and vitreous, posterior synchiae, fundus view was hazy (Fig. 1.B). Her baseline investigations, x-ray chest and B-scan of OS were normal. Based on these findings patient was provisionally diagnosed as a case of sympathetic ophthalmia, her OD being the excited eye and OS the sympathetic eye which developed uveitis without any obvious etiology other than the trauma to the OD. She was kept on systemic and topical steroids

prior to proceeding for enucleation of OD. Following the enucleation the OS became quiet (Fig. 1.C) and the child was sent home. To our surprise the histopathology report of the enucleated eye concluded it to be a Retinoblastoma (RB). Since the child's history was not suggestive of RB and her age was old for RB we requested the laboratory to review the slides again. A board of pathologists reviewed the slides, concluding that it was a RB indisputably showing well differentiated Homer - Wright rosettes and Flexener - Wisternsteiner rosettes (Fig. 2). On TNM staging patient had a pT_2 Nx Mx with no optic nerve involvement. Patient was referred to an advance oncology unit for further management of RB.

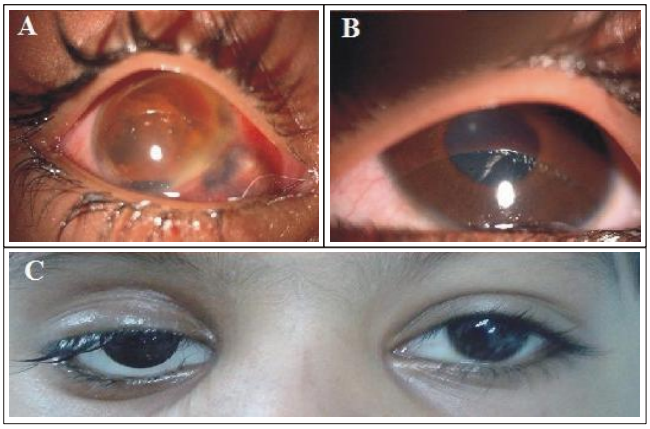

Fig. 1: (A) Right eye on first presentation showing hyphema, anterior staphyloma. (B) Left eye at second visit showing keratic precipitates, posterior synchiae (C) Post enucleation right eye with prosthesis and left eye is normal

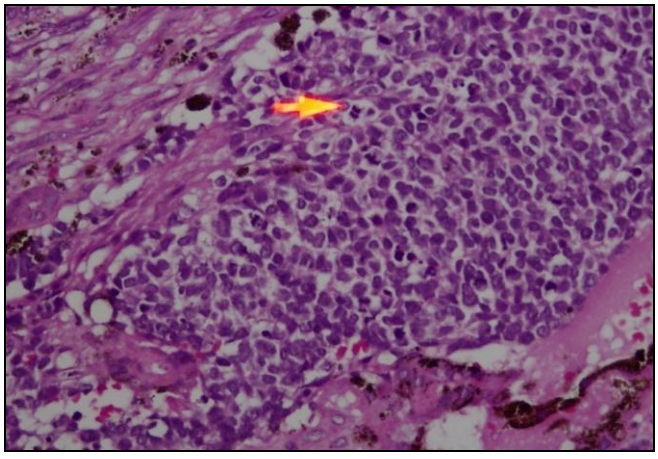

Fig. 2: Histopathological slide of enucleated right eye, arrow showing mitotic figures.

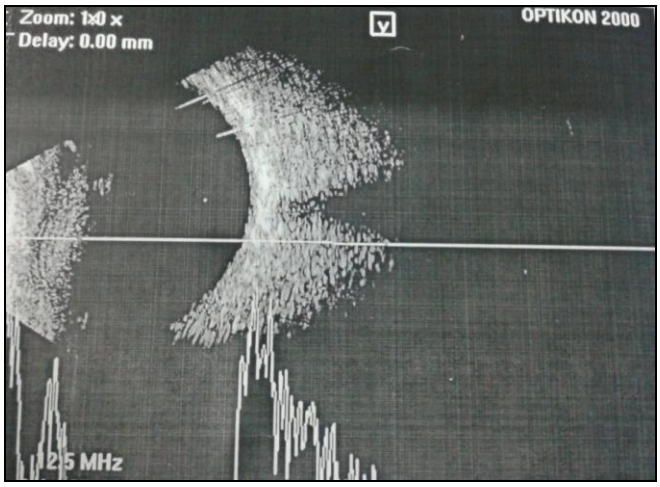

Fig. 3: Normal ultrasound B - Scan of left eye after enucleation of right eye

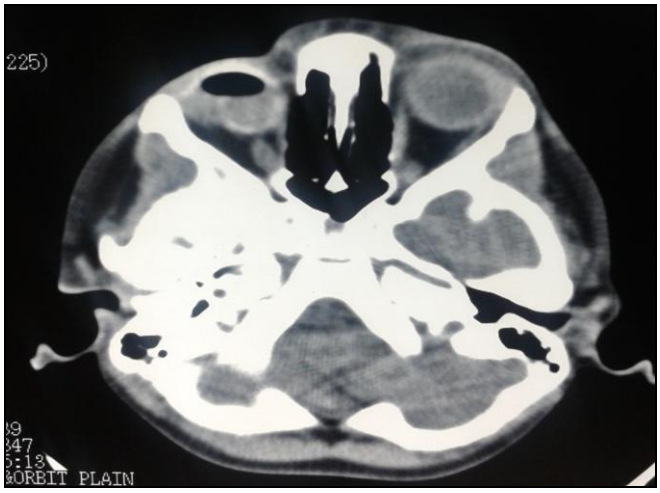

Fig. 4: CT Scan of brain and orbit done after enucleation, showing implant in right eye and normal left eye.

DISCUSSION

Masquerade syndrome is a rare entity as it presents an unusual presentation of relatively rare conditions; therefore mostly they are either undiagnosed or under diagnosed. They may be benign or malignant; however the latter are more common. Some of the malignant masquerading conditions include primary intraocular lymphoma, uveal melanoma, retinoblastoma, leukemia, metastatic lesions and paraneoplastic syndromes.[1] Uveitis in the pediatric population is less common than adults. A British study reported the incidence of pediatric uveitis to be 3.15 per 100,000 children up to 5 years of age[4].

The literature does not give the actual incidence of sympathetic ophthalmia (SO), as it is a rare condition but most studies concur that it is 0.2% to 0.5%

following ocular trauma and comprises only 0.3% of all uveitis[2]. The treatment protocol is mainly medical comprising of corticosteroids or immuno-suppressive therapy. Enucleation remains a debatable topic in SO: studies suggest primary enucleation done within 10 days of injury, is beneficial for reducing the inflammation in the sympathetic eye. But it is quite difficult to give a definite time frame for enucleation especially if the patient is a child. In our case we performed a secondary enucleation as the excited eye was a painful blind eye[5] and the fellow eye developed refractory uveitis, which resolved following the surgery.

Less than 10% of retinoblastomas (RB) present as intraocular inflammation and 50% of these present as orbital cellulitis[6]. Any other presentation makes the already difficult diagnosis next to impossible, accounting for the majority of misdiagnosed RBs. Our case did not have a typical presentation of RB, she had no relevant family history and she had passed the age of usual RB which is mostly diagnosed prior to 5 years of age[6]. The definite diagnosis can only be reached after a histopathological evidence as was in our case.

CONCLUSION

Our case highlight a rare and unique presentation of retinoblastoma as sympathetic ophthalmia, thus masquerade syndrome. Any refractory uveitis particularly in a child should raise a high index of suspicion of a masquerade syndrome and should be evaluated thoroughly.

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