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Case Report

Bilateral Retinoblastoma Presenting Atypically with Atrophic Bulbi and Contralateral Buphthalmos

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Abstract

Patients with retinoblastoma commonly present with leucocoria (56%) and strabismus (20%). Presentation in the form of phthisis bulbi or atrophic bulbi and buphthalmos in a single patient is unusual and highly suggestive of retinoblastoma and is observed commonly in older children where it delays the diagnosis and management. This case highlights the unusual signs of this malignant tumor in a 2 year old boy who presented with atrophic bulbi in right eye and buphthalmos with scleral metastasis in the left eye. Computed tomography scan demonstrated calcified atrophic right globe with a buphthalmic left eye with intralesional calcification and a scleral metastatic nodule. The patient was planned for further investigations to rule out distant metastasis however was lost to follow-up.

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Keywords: *Bilateral retinoblastoma, Atrophic bulbi, Buphthalmos, Scleral metastasis, Phthisis bulbi, Calcification, Enucleation*

A two year old boy presented with gradually worsening inability to walk independently for last nine months. Parents noticed whitish opacity in the black of both eyes (OU) for eight months along-with reduced size of right eye (OD) since last seven months (Figure 1A, 1B, 1C). A rapidly increasing painless reddish swelling was observed over supero-temporal aspect of the left eye (OS) 15 days prior to presentation to us (Figure 1C). There was no associated history of trauma, protrusion or deviation of either eye, significant weight loss or any previous ocular surgery. On examination, the child was neither fixing nor following light OU. Episcleral vessels were prominent OU (Figure 1B). A well-defined scleral nodule was observed supero-temporally OS measuring 10 mm x 8 mm (Figure 1D). Examination under anesthesia revealed opaque normal sized corneas with leucomatous opacities OU along-with accompanying vascularisation of the opacity OS. There were near total posterior synechiae OD. A partially absorbed calcified lenticular opacity along-with neovascularization of iris was observed OS through a small clear area in the visual axis. Fundal glow was absent OU. Intraocular pressure measured using schiotz tonometer was 17.3 mmHg OD and 40 mmHg OS. Anterior chamber had no hypopyon or hyphaema. Hertel's exophthalmometry confirmed absence of proptosis. The scleral nodule increased in size over seven days of hospital stay to 15 x 12 mm. Ultrasound B scan OU revealed characteristic V Y pattern along-with calcification (Figure 2A, 2B). Contrast enhanced computed tomography demonstrated soft tissue mass in posterior chamber with intralesional calcifications OU (Figure 2C) which was reaching up to the optic disc with slight deformity at optic nerve insertion and lens was dislocated superiorly OD. Lens could not be identified OS however optic nerve appeared grossly normal. Scleral metastasis was noted with evidence of calcification in the solitary nodule. Axial length was 17 mm OD and 24 mm OS (Figure 2C). No intracranial, extra-ocular muscle or bony involvement was noted OU. A diagnosis of bilateral retinoblastoma with atrophic bulbi OD and buphthalmos OS was presumed. Systemic evaluation for distant metastasis was planned however the child was lost to follow up.

Phthisis (incidence of 2%) and simultaneous buphthalmos in a single patient is extremely rare however strongly indicates bilateral retinoblastoma as emphasized by Boniuk and Zimmerman's and is commonly observed in older children where it delays the diagnosis and management.[1,2] Rapid proliferation rate and high mitotic index of the tumor results in spontaneous infarction and necrosis with subsequent calcification and the eye undergoes phthisis.[2,3] Such eyes need subsequent imaging and enucleation followed by histopathological confirmation of the presence of tumor and also to exclude extra-ocular and optic nerve extension. Imaging features may be altered as a result of tumor necrosis. Under 10 similar cases have been reported until now.[4,5] This case is unique and very rare combination of these two presenting signs in different eyes of the same child adds further to the existing scant literature pertaining to the possibility of harbouring retinoblastoma in these eyes and hence its prompt management.

Figure 1(A-D): *Sequential patient photographs - A 2 year male child presented with bilateral opaque corneas with atrophic right globe and a buphthalmic left eye (1A) with prominent episcleral vessels in both eyes (1B). A well-defined progressively increasing scleral nodule was noticed in the supero-temporal quadrant of the left eye that measured 15mm x 12 mm (1C,1D).*

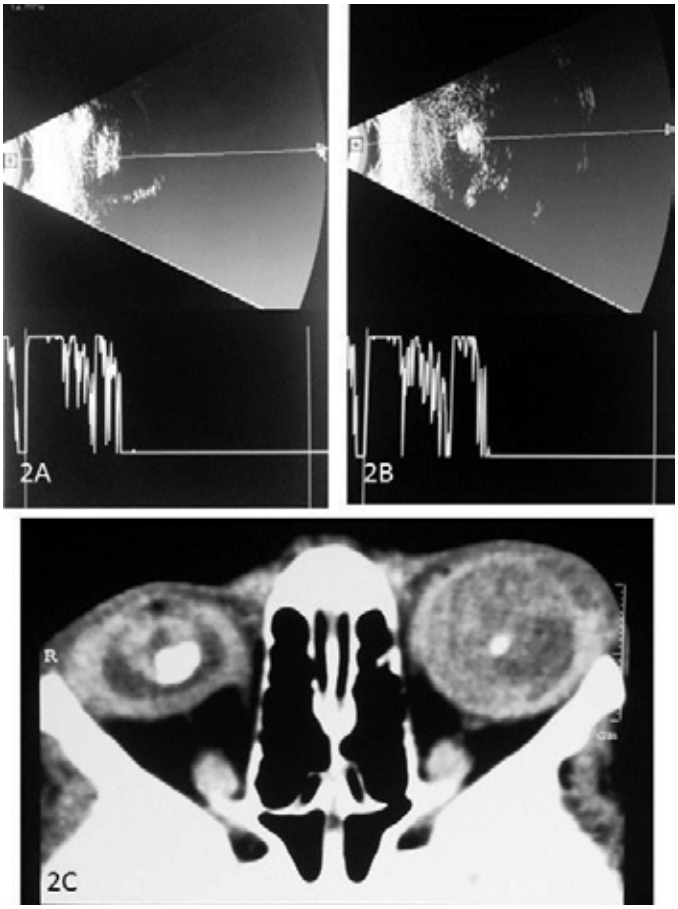

Figure 2(A-C): *Ultrasound and computed tomography images - Ultrasound B scan of both eyes revealed characteristic V Y pattern along-with high intensity spikes suggestive of calcification (2A, 2B). Contrast enhanced computed tomography (CT) scan of the right globe demonstrated heterogenously enhancing soft tissue mass measuring 10 x 10 x 9 mm in posterior chamber with calcifications within the mass (Figure 2C). Antero-posterior diameter of right globe was 17 mm suggestive of atrophic right globe. Left eye had a heterogenously enhancing soft tissue mass measuring 23 x 19 x 17 mm diffusely involving posterior chamber with calcifications within it (2C). Scleral metastasis was noted with evidence of calcification in this solitary nodule and the left globe measured 24 mm anteroposteriorly (2C). No extra-ocular muscle or bony involvement was noted in either eye.*

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