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Retinal Vasoproliferative Tumor in Ocular Albinism

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Study Design A
Data Collection B
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Data Interpretation D
Manuscript Preparation E
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Conflict of interest: None declared

Patient: Male, 42-year-old
Final Diagnosis: Ocular albinism
Symptoms: Vision loss
Medication: —
Clinical Procedure: —
Specialty: Ophthalmology

Objective: Rare disease

Background: Retinal vasoproliferative tumor (VPT) is a type of ocular vascular tumor that commonly occurs idiopathically and can be associated with secondary ocular diseases. Ocular albinism is an X-linked inherited disease and distinguished from oculocutaneous albinism by less hair and skin involvement.

Case Report: A 42-year-old man with ocular albinism and moderate myopia presented with a history of insidious decrease in vision in both eyes over a period of months. On examination, the horizontal pendular nystagmus was present and diffuse iris transillumination defects were observed bilaterally. A fundus examination revealed a depigmented fundus with visible choroidal vessels, foveal hypoplasia, and a unilateral, elevated, vascular lesion in the superotemporal aspect of the retinal periphery. Optical coherence tomography of the lesion confirmed the retinal location and fluorescein fundus angiography indicated its vascular nature. B-scan ultrasonography was performed to measure the dimensions of the lesion.

Conclusions: Rare retinal VPT has been reported with systemic and ocular associations, but it has never been reported in the literature in association with ocular albinism. Multiple treatment modalities have been described for the tumor, but observation can be considered in the absence of secondary consequences of the VPT. Retinal VPT should be included in the differential diagnosis of any retinal vascular abnormalities in patients with ocular albinism.

MeSH Keywords: Albinism, Ocular • Retinal Artery • Retinal Neoplasms

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Background

Garrod first described albinism in 1908 under the spectrum of an inborn error of metabolism [1]. This inherited disorder manifests with ocular and systemic features due to reduced synthesis or distribution of melanin and is classified as oculocutaneous albinism, or solely with ocular manifestations as in ocular albinism [2]. The inheritance of ocular albinism is either X-linked or autosomal recessive, and patients with it present with variable features, including reduced visual function with anterior segment and posterior segment characteristic signs in addition to abnormal decussation of the optic chiasm to the contralateral hemisphere [2–5].

Retinal vasoproliferative tumors (VPT) are rare, benign, vascular masses that were considered "presumed acquired nonfamilial retinal hemangioma" before the current terminology was adopted in 1995 [6,7]. Despite their benign nature, the effects on visual acuity can be significant, depending on the complications of the tumors, such as macular edema, exudates, epiretinal membrane, retinal detachment, and neovascular glaucoma in the advanced stages of this pathology [8–11]. VPT has been classified as either a primary tumor, which is not associated with prior ocular pathology, or less commonly as a secondary tumor associated with a probable causative etiology [11].

We report a case of VPT associated with ocular albinism, which to our knowledge, has not been previously reported in the literature.

Case Report

A 42-year-old Saudi man with ocular albinism presented to the retina clinic at King Khaled Eye Specialist Hospital (KKESH), Riyadh, Saudi Arabia, with a complaint of small scotoma in the inferonasal part of the visual field. The patient wore eyeglasses for moderate myopia and denied a history of eye trauma or previous ophthalmic surgery. Systematic review was not significant and the patient's past medical history was unremarkable except for internal fixation with a titanium plate of a bone fracture of the left humerus.

Externally, the patient's eyebrows and eyelashes were black in color with no poliosis, and his skin texture was classified as Type 3 on the Fitzpatrick scale. The best-corrected visual acuity was 20/100 in both eyes and the intraocular pressure was 21 mmHg in the right eye and 18 mmHg in the left eye, measured by the pneumotonometer. The extraocular motility was normal in both eyes and the patient's pupils were equal, reactive to light and near reflex, with no relative afferent pupillary defect. The eyes looked straight in the primary gaze with no deviation; however, a horizontal pendular nystagmus was observed in all directions of gaze.

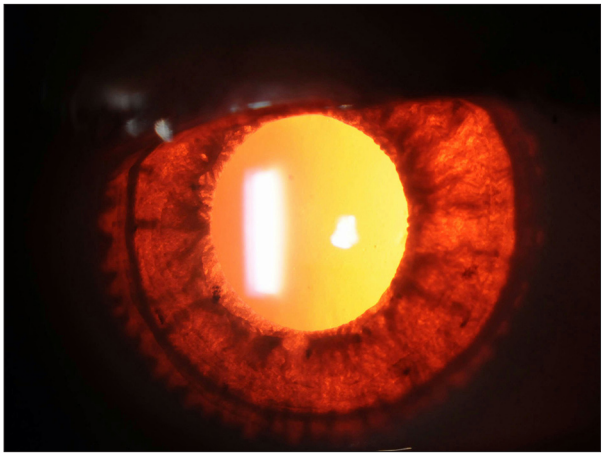

Figure 1. External ocular photo showing marked iris transillumination defect by retroillumination.

Slit lamp biomicroscopy in both eyes showed normal eyelids, quiet conjunctiva, clear cornea, deep and quiet anterior chamber, diffuse iris transillumination defects, and clear crystalline lens (Figure 1). A dilated fundus examination revealed a clear vitreous with posterior vitreous detachment and generalized depigmentation of the fundus with clearly visible large choroidal vessels and vortex vein ampullae. The macular area showed foveal hypoplasia with no exudation, lipid deposits or epiretinal membranes over the macula. In the peripheral superotemporal area of the retina of the right eye, an elevated angiomatous lesion was visible, which had vascular malformation and pronounced telangiectatic changes but no apparent afferent-efferent vessels (Figure 2).

Optical coherence tomography (OCT) of the macula indicated an absence of foveal pits in either eye. An OCT scan over the vascular lesion confirmed its retinal location rather than a choroidal pathology. B-scan ultrasonography was performed to measure the dimensions of the lesion and showed an apical height of 0.95 mm and a basal diameter of 3.19 mm. A-scan ultrasonography was difficult to perform because of the small size of the lesion; however, it showed low internal reflectivity (Figure 2). Fluorescein fundus angiography (FFA) indicated the vascular nature of this lesion, with hyperfluorescence in the early frames and a slight increase in fluorescence in the later frames (Figure 3). Magnetic resonance imaging was not performed because the patient had a metal implant in his left arm. Clinical examination supported by ancillary tests suggested a diagnosis of VPT.

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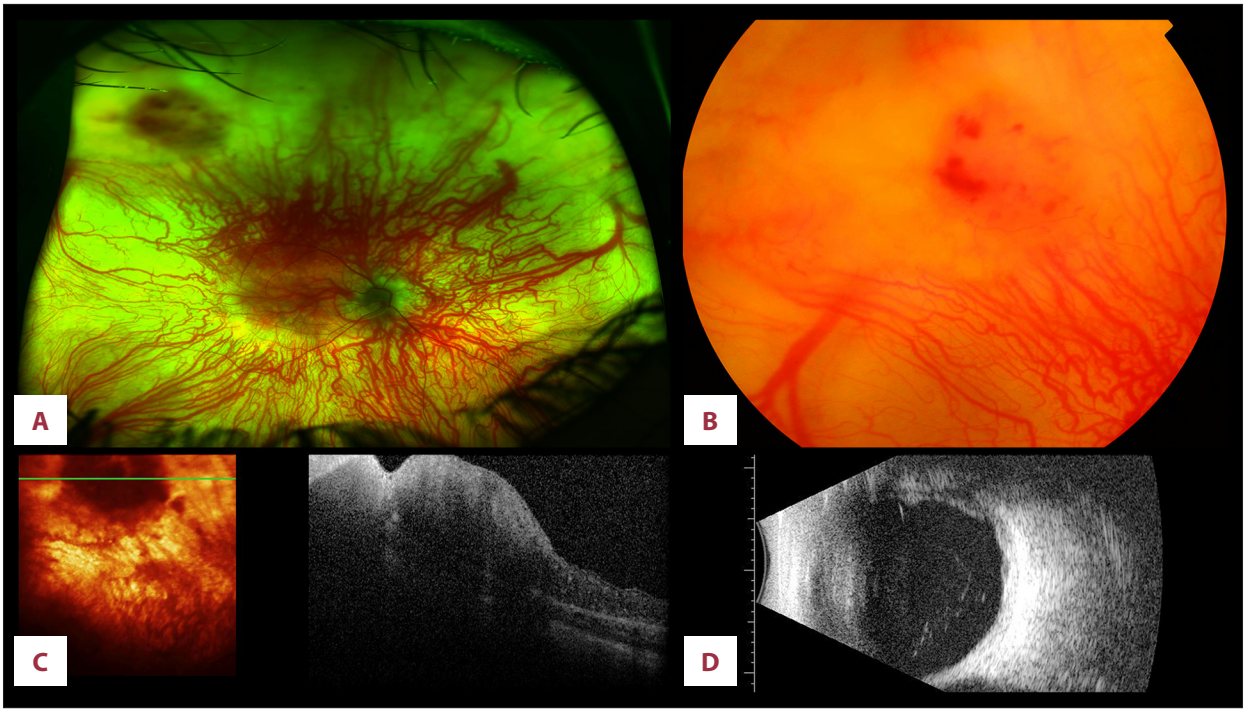

Figure 2. (**A**) Color fundus photo (Optos) showing superotemporal retinal mass near the superior arcade. (**B**) Color fundus photo (Topcon) showing closer image of the mass with angiomatous appearance in the superotemporal quadrant. (**C**) Optical coherence tomography through the lesion showing retinal elevation and thickening with no obvious choroidal abnormality. (**D**) Ultrasound B-scan image showing a retinal mass with apical height of 0.95 mm and a basal diameter of 3.19 mm.

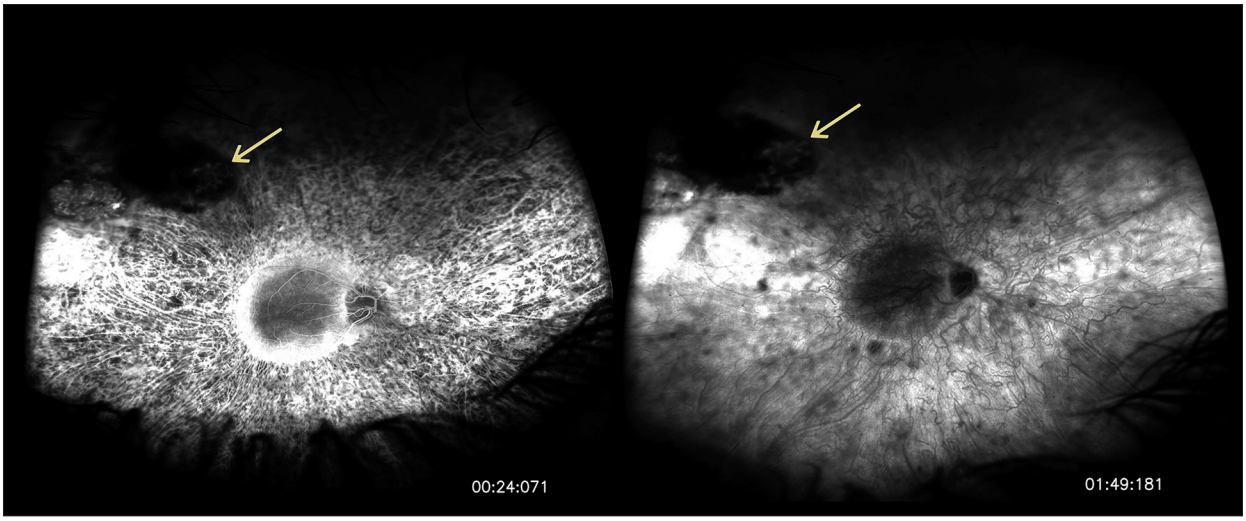

Figure 3. Early and late frames from fluorescein angiography. Mild hyperfluorescence corresponding to telangiectasia with no retinal neovascularization is shown in the early frame, adjacent to an area of hypofluorescence, which represents ischemia. In the late frame, a mild increase in hyperfluorescence in the lesions is visible.

Discussion

The pathophysiology of VPT is not well understood. Vasoproliferation is the main characteristic of the tumor, as indicated by the terminology. Histologically, however, it arises from primary glial cell proliferation, which leads to secondary vasoproliferation [12]. This observation correlates with the finding of retinal thickening and shadowing noted on the OCT images of the lesion in our case. VPT has been linked to many ocular conditions, including retinitis pigmentosa, pars planitis, post-retinal detachment surgery, and other rare etiologies. However primary (idiopathic) VPT is much more common [11].

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Shields et al. [11] conducted the largest study and reported the similarities and differences between primary and secondary VPT. The main characteristics shared by the majority of subgroups were that the majority of the tumors were located inferotemporally and anterior to the equator; however, in secondary VPT, the prevalence of bilaterality, multifocality, and post-equatorial location was much higher compared with that in primary VPT [11]. One distinctive feature of retinal VPT is feeder and draining vessels, but they do not manifest in all of the lesions. A report by Shields et al. [11] showed that only 56–57% of VPTs have these vessels. The tumors may present with normal retinal vessel gauge and generally do not have dilated tortuous feeder vessels, as in capillary hemangioma.

To the best of our knowledge, a secondary association of VPT with ocular albinism has not been reported in the literature. In addition, the superotemporal quadrant is not the typical site for the tumor and the absence of remote macular manifestations, including macular edema or exudates, explains our patient's equal vision in both eyes. Ancillary testing including FFA may be helpful in confirming the diagnosis of VPT. FFA can be used to exclude other retinal vascular lesions by revealing early or late hyperfluorescence associated with leakage from the tumor, especially if the VPT is not covered with hemorrhages or extensive membranes that might block the full extent of the activity [11,13]. It also can be used to assess tumor activity after treatment.

The best explanation for the association between ocular albinism and secondary VPT is impairment in the integrity of the retinal pigment epithelial layer, which is known to occur in patients with albinism [14]. Rundle et al. [15] reported a case with an association between Waardenburg syndrome and secondary VPT, a link that was attributed to the defective retinal pigment epithelium seen with the syndrome, which also was evident in our case. Multiple treatment modalities with variable results have been used to minimize or eliminate the consequences to visual function of VPT, including cryotherapy, laser photocoagulation, intravitreal anti–vascular endothelial growth factor, photodynamic therapy, and even vitreoretinal surgery, especially if the tumor is associated with vitreous bleeding or retinal detachment [8,13]. In our case, VPT was not affecting the patient's visual acuity or macular area, so rather than risk a decrease in his vision, we did not use any of these modalities.

Conclusions

In summary, ocular albinism can be associated with VPT and should be suspected in patients with albinism who have retinal vascular lesions. The management of the tumors is variable, but observation can be considered in the absence of any macular manifestations that can reduce visual function. To the best of our knowledge, the occurrence of retinal VPT in patients with ocular albinism has not been reported previously in the literature. This case may shed light on the possible association between ocular albinism and VPT as a secondary etiology similar to the previously mentioned ocular associations mentioned in the literature. Abnormal retinal pigment epithelium integrity, as seen with albinism, may be the best explanation for development of the association between the condition and retinal VPT.

Conflicts of interest

None.

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