

Management of complicated proliferative diabetic retinopathy in a patient with oculocutaneous albinism

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ABSTRACT

Purpose: To describe the management and outcome of a patient with oculocutaneous albinism and complicated proliferative diabetic retinopathy, as well as to discuss treatment challenges and strategies in this patient population.

Observation: A 52-year-old patient with oculocutaneous albinism and diabetes presented with light perception vision in her right eye and 20/300 vision in her left eye. Examination showed a diabetic tractional retinal detachment in the right eye and high-risk proliferative diabetic retinopathy (PDR) in the left eye. In the right eye, the patient underwent pars plana vitrectomy, membrane delamination, endolaser therapy, and silicone oil tamponade, with follow-up evaluations showing a flat retina under silicone oil with regressed retinopathy in this eye. In her left eye, pan-retinal photocoagulation was attempted without success, with persistent PDR and absence of laser marks in this eye. Subsequently, the patient underwent six intravitreal anti-VEGF injections, after which she developed a tractional retinal detachment necessitating treatment cessation and a planned surgical intervention.

Conclusion and Importance: This case highlights the difficulty of using standard medical and surgical treatment strategies when caring for patients with oculocutaneous albinism and complicated proliferative diabetic retinopathy. If medical treatments fail in these patients, surgical approaches should be planned with extreme care due to the risks and challenges posed by hypopigmented fundi.

Method: Interventional case report.

1. Background

Oculocutaneous albinism (OCA) is an autosomal recessive disease characterized by a partial or complete reduction in melanin biosynthesis within melanocytes, leading to hypopigmentation of the eye, hair, and skin. Seven types of oculocutaneous albinism have been reported to date, ranging in severity from a complete lack of melanin production throughout life (OCA1A) to milder forms with variable accumulation of pigment (OCA1B, OCA2-7). Ophthalmic features are similar among the different disease variants and include decreased vision, photophobia, refractive errors, nystagmus, iris transillumination, hypopigmented fundi, and foveal hypoplasia.¹

Here, we present a case of a patient with oculocutaneous albinism and diabetes who presented with decreased vision and was found to

have complicated proliferative diabetic retinopathy. The unique challenges of providing appropriate therapy to this patient are discussed, as well as important treatment strategies for clinicians to consider.

2. Case report

A 52-year-old patient with oculocutaneous albinism presented to our tertiary-care eye hospital with a complaint of gradual loss of vision in both eyes. She had no prior ocular history of trauma, inflammation, or medical interventions. She had a history of diabetes mellitus for 19 years, managed with insulin therapy with fair control, as well as hypertension and dyslipidemia.

On physical examination, her visual acuity was light perception in the right eye and 20/300 in the left eye. Orthoptic evaluation showed a

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moderate degree of esotropia in the right eye and low-amplitude horizontal jerky nystagmus in both eyes. Anterior segment biomicroscopy revealed clear corneas, early nuclear sclerosis, and light-pigmented irides in both eyes. Fundus examination of the right eye showed a hypopigmented fundus with fibrovascular proliferation and a tractional detachment of the macula (Fig. 1). Fundus examination of the left eye showed hypopigmentation with tufts of neovascularization at the posterior pole (Fig. 2). A surgical intervention was planned for the right eye, with pan-retinal photocoagulation (PRP) planned for the left eye.

Pars plana vitrectomy of the right eye was performed in May of 2018. The surgery started with a core vitrectomy and dissection of the fibrovascular membranes associated with the small retinal break. Endolaser therapy was then applied in a pan-retinal fashion with faint retinal bleaching around the break. Finally, silicone oil tamponade was injected. Postoperatively, vision improved in this eye from light perception to hand motion. Follow-up evaluations up to two years after the intervention showed a flat retina with a sealed break and regressed neovascular activity (Fig. 3).

At first presentation, PRP was attempted in the left eye without success, as demonstrated by an absence of laser uptake on ophthalmoscopy (power output ranged from 300 to 700 mW, at which point the patient felt significant pain). Consequently, the patient was scheduled for monthly intravitreal anti-VEGF injections in the left eye, with a total of six ranibizumab injections administered over a 6-month period. At the last follow-up evaluation, the patient showed some regression of the active new vessels but had developed a tractional retinal detachment involving the macula (Fig. 4). The patient was counseled accordingly and offered a surgical repair for this macular detachment with pars plana vitrectomy and membrane peeling, but she didn't agree to proceed for surgery in this eye.

3. Discussion

This case highlights the complexity of managing patients with ocular albinism who develop proliferative diabetic retinopathy. To our best knowledge, only one case has been reported of a patient with albinism who developed proliferative diabetic retinopathy,² with a few other reports describing patients with albinism who developed retinal detachments due to other causes.³⁻⁶ In these previous patients, major reported challenges included control of diabetic neovascular proliferation, identification of retinal breaks, and lack of laser uptake. In this case, our challenges were similar.

During the right-sided vitrectomy, we found that dissection of the fibrovascular membrane was difficult due to a lack of contrast with the



Fig. 2. Fundus photo of the left eye.

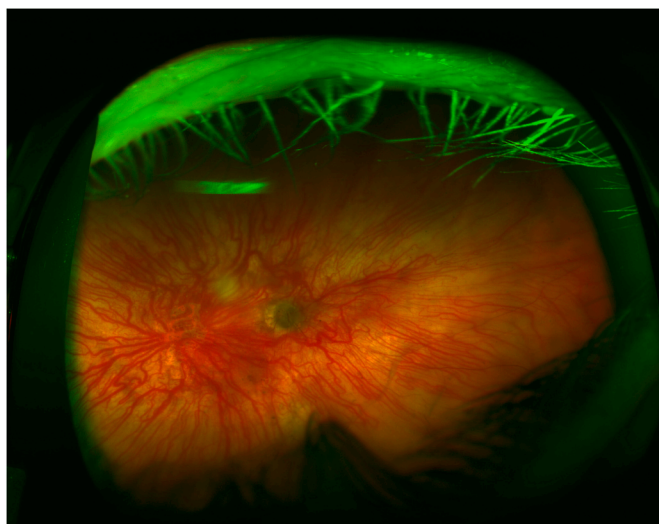


Fig. 3. Fundus photo of the right eye after surgery.

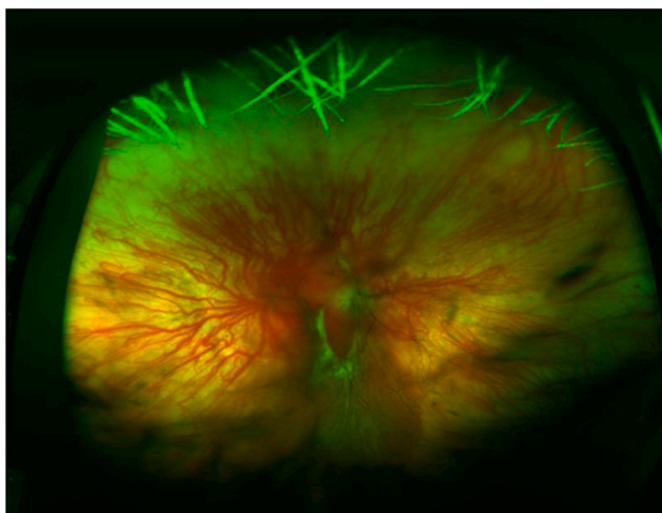


Fig. 1. Fundus photo of the right eye.

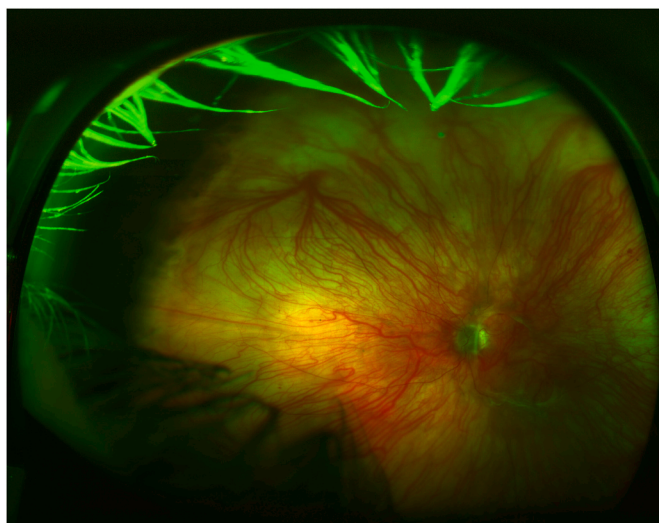


Fig. 4. Fundus photo of the left eye after intravitreal Anti-VEGF injection course.

hypopigmented fundus. In addition, endolaser treatment of the dissection-related break was difficult, and only a faint reaction was observed. Accordingly, silicone oil was chosen for the tamponade because its long-lasting property was considered desirable to support this doubtful laser uptake. In this case, we will likely keep the silicone oil in place for life, unless complications ensue.

Sinha et al. reported a case series of patients with albinism who underwent retinal detachment surgeries, describing an inability to achieve visible laser marks in all the three operated cases.⁴ In addition, Mansour et al. reported partially successful laser therapy in two out of five cases.⁵ Cryopexy has been used in the reported cases as a primary treatment or after the failure of laser to give the adequate support, and it has been shown to be more useful and effective in retinal reattachment.^{4–6} In our case, we speculated that the total absence of fundus pigmentation would have likely made it impossible to obtain a proper chorioretinal adhesion, even with cryotherapy. In addition, the lack of pigmentation would likely have made power titration of both the laser therapy and the cryotherapy very challenging and may have resulted in overtreatment of the retinal tissue, with consequent tissue necrosis and enlargement of the break.

In the left eye, PRP with an argon laser was attempted with no visible reaction, as previously reported in a similar case.² Monthly intravitreal anti-VEGF injections, which has been shown to be noninferior to PRP for management of PDR,^{7,8} was therefore administered to this patient. After six ranibizumab injections, we noted development of a tractional retinal detachment with macular involvement, as well as incomplete resolution of vascular activity. The development or progression of a tractional retinal detachment during anti-VEGF treatment is a known adverse effect of this medication. Arevalo et al.⁹ reported that this complication affected 5.2% of eyes following bevacizumab injections. The development of this complication can potentially be explained by the rapid neovascular involution and membrane contraction which follows a shift in balance between VEGF and connective tissue growth factor (CTGF) levels.¹⁰

Patient consent

Verbal consent to publish this case has been obtained. This report does not contain any personal identifying information.

Summary statement

52-year-old albino patient presented with decreased vision. Examination showed proliferative diabetic retinopathy in both eyes. Basic medical and surgical treatment modalities were applied with major

difficulties encountered due to lack of fundus pigmentation.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Declaration of competing interest

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