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CASE REPORT PEER REVIEWED | OPEN ACCESS

Focal laser and intravitreal bevacizumab for choroidal neovascularization secondary to Grönblad–Strandberg syndrome

Olufemi Oderinlo, Toyin Akanbi

ABSTRACT

Introduction: Pseudoxanthoma elasticum is a rare genetic disorder. It has several ocular features including angioid streaks and choroidal neovascularization which can significantly affect vision.

Case Report: A 50-year-old man presented with nine months history of poor vision affecting both distant and near vision. His best corrected visual acuities were 6/18 right eye and 1/60 left eye. His anterior segments were normal. Funduscopy revealed linear hyperpigmented irregular branching peripapillary lesions; angioid streaks both eyes. He had bilateral speckled retina pigment epithelial hyperpigmented lesions interspaced with dot hemorrhages in the macula region suggestive of likely choroidal neovascularization. He had "peau d'orange" skin appearance around his neck. Fundus fluorescein angiography showed features of occult subfoveal choroidal neovascularization. He was referred to a physician and had a skin biopsy which revealed histologic features of Pseudoxanthoma elasticum. He was treated with bilateral focal argon laser. However he had right intravitreal Bevacizumab after a large ipsilateral subfoveal hemorrhage. His visual acuities remained stable afterward.

Conclusion: Patients with Pseudoxanthoma elasticum have to be closely monitored for ocular manifestations. Choroidal neovascularization should be detected early and promptly treated. Antivascular endothelial growth factor injections have become the preferred treatment option for choroidal neovascularization in these patients.

Keywords: Bevacizumab, Choroidal neovascularization, Laser, Pseudoxanthoma elasticum

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INTRODUCTION

Pseudoxanthoma elasticum (PXE) is a genetic metabolic disease with autosomal recessive inheritance caused by mutations in the ABCC6 gene [1]. It affects the skin, eyes, and cardiovascular system, there is progressive calcification and fragmentation of elastic fibers in the affected tissues [2].

The association between retinal angioid streaks and skin features in PXE was reported by Grönblad and by Strandberg in 1929 [3, 4] and PXE is referred to as Grönblad–Strandberg syndrome.

Ocular features of PXE include angioid streaks, peau d'orange, comet lesions, choroidal neovascularization (CNV), chorioretinal atrophy, subretinal fluid independent

from CNV, pattern dystrophy-like changes, debris accumulation under the retinal pigment epithelium, and reticular drusen [5]. Angioid streaks are the most obvious and most observed funduscopic finding in these patients [5]. Choroidal neovascularization is a frequent complication in patients with PXE and it may cause pronounced vision loss [5].

CASE REPORT

A 50-year-old Nigerian man presented with nine months history of poor vision affecting both distant and near vision. He complained of seeing flashes of light in the right eye. He had worn corrective spectacles for 10 years. His mother had been treated for glaucoma, his father also had poor vision, the cause was not known.

Ocular examination revealed best corrected visual acuities 6/18 right eye (OD) and 1/60 left eye (OS). His anterior segments were normal bilaterally (OU). Intraocular pressures were 10 mmHg OU; he had early lens opacities OU. Funduscopy revealed linear hyperpigmented irregular branching peripapillary lesions (angioid streaks) OU. He also had speckled retina pigment epithelial hyperpigmented lesions interspaced with dot hemorrhages in the macula region suggestive of choroidal neovascularization OU (Figure 1). Fundus fluorescein angiography (FFA) showed bilateral peripapillary hyperfluorescent branching patterns as a result of unmasking of choroidal fluorescence and stippled non-homogenous hyperfluorescent macular pattern with irregular edges suggestive of occult subfoveal choroidal neovascularization (Figure 2). He had "peau d'orange" skin appearance around the neck (Figure 3). His cardiovascular system was normal.

He was referred to a physician, his cardiovascular system was normal but his skin biopsy revealed swollen and irregularly clumped altered elastin fibers within the middle and lower thirds of the dermis with mild basophilic staining. There were also foci of accumulation of basophilic acellular materials within the vicinity of the elastin fibers with mild chronic inflammatory cellular infiltrates. He was diagnosed with Grönblad–Strandberg syndrome.

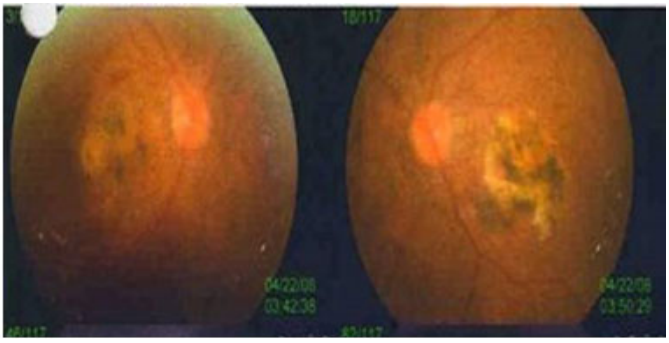

Figure 1: Fundus photographs showing bilateral angioid streaks and speckled retina pigment epithelial hyperpigmented lesions interspaced with dot hemorrhages in the macula.

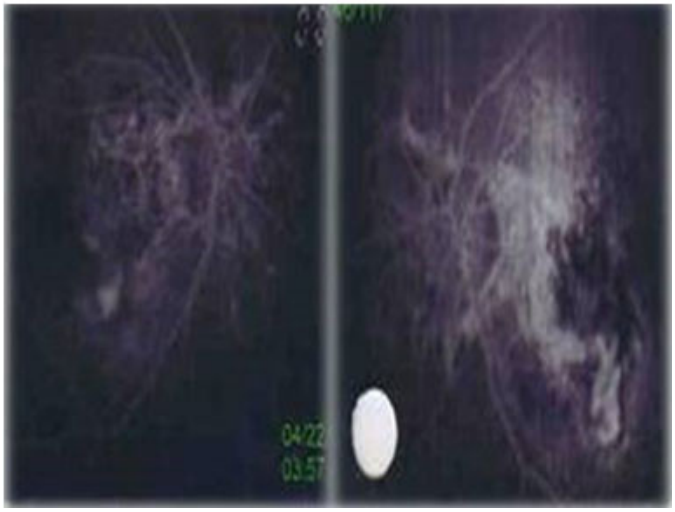

Figure 2: Fundus fluorescein angiography showing bilateral peripapillary hyperfluorescent branching pattern and stippled non-homogenous hyperfluorescent macular pattern with irregular edges suggestive of occult subfoveal choroidal neovascularization.

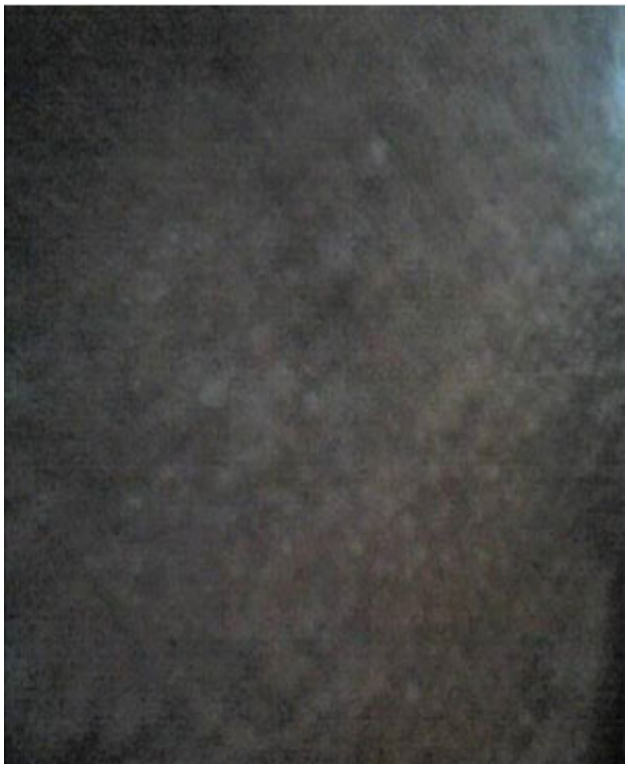

Figure 3: Photograph of patient's neck showing "peau d'orange" skin appearance of the neck.

He was treated with bilateral focal argon laser (250 μm × 0.12 mW × 122 pulses). Five months later he developed a large subfoveal hemorrhage OD and a disciform scar OS. His visual acuity had reduced to hand movement OD and 6/36 in OS. He had one dose of intravitreal Bevacizumab 1.25 ug/0.05 mg OD and more laser photocoagulation OD three months later with complete resolution of choroidal neovascularization and stabilization of visual acuity.

DISCUSSION

There is no cure for PXE and patients should be monitored on a regular basis [1]. The most severe ocular complication of PXE is CNV [6]. Untreated CNV results in subretinal bleeding and exudation, leading to fibrovascular scarring and a progressive loss of vision [5].

Our patient had bilateral CNV; he was treated with bilateral focal laser and intravitreal Bevacizumab OD with stabilization of his visual acuity. Bevacizumab was given OD post focal laser when he developed a huge subfoveal hemorrhage.

Gliem et al. reported that laser photocoagulation yielded comparable results as photodynamic therapy; however, the application of laser was mostly restricted to extrafoveal lesions; it was complicated by frequent recurrences, and led to more retinal damage with subsequent absolute scotomas [7].

The use of intravitreal anti-vascular endothelial growth factors (VEGF) have been documented in several case reports, case series, and clinical trials and they have become the first-choice treatment for CNV secondary to angioid streaks and the ocular manifestations of PXE [8].

Intravitreal Bevacizumab [9–11], Ranibizumab [12], Aflibercept [13], and Conbercept [14] have been used in treating CNV associated with PXE.

Finger et al. reported that intravitreal Bevacizumab therapy demonstrates long-term effectiveness by preserving function in advanced disease and improving function in early disease [11].

CONCLUSION

Pseudoxanthoma elasticum is a rare disease. Patients with PXE have to be closely monitored for ocular manifestations. It may be associated with CNV which can cause significant visual impairment. Choroidal neovascularization should be detected early and promptly treated. Intravitreal anti-VEGF injections have become the mainstay of treatment for CNV in these patients.

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Author Contributions

Olufemi Oderinlo – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Toyin Akanbi – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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Conflict of Interest
Authors declare no conflict of interest.

Data Availability
All relevant data are within the paper and its Supporting Information files.