

Long-term follow-up of a case of choroidal neovascularization secondary to reticular pigmentary retinal dystrophy

Seguimento de longo-prazo de um caso de neovascularização de coróide secundária à distrofia reticular pigmentar da retina

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ABSTRACT

Reticular pigmentary retinal dystrophy, also known as Sjögren's reticular dystrophy, is a rare condition characterized by macular lesions with a reticular pattern, which are best seen on fluorescein angiogram. Choroidal neovascularization secondary to this type of dystrophy is even less common. This report describes a case of reticular pigmentary retinal dystrophy with vision loss due to neovascular membrane, which responded well to treatment with anti-vascular endothelial growth factor.

RESUMO

A distrofia reticular pigmentar da retina, também conhecida como distrofia reticular de Sjögren, é uma doença rara, caracterizada por lesões maculares com um padrão reticular, que são mais bem visualizadas na angiografia com fluoresceína. A neovascularização de coróide secundária a este tipo de distrofia é ainda menos comum. Este relato descreve um caso de distrofia reticular pigmentar da retina, com perda de visão devido à membrana neovascular, que respondeu bem ao tratamento com fator de crescimento endotelial antivascular.

INTRODUCTION

Reticular pigmentary retinal dystrophy was first described in 1950 in eight out of 13 children of a Swedish family (Sjögren et al.). Fundusoscopic findings consist of bilateral asymmetric hyperpigmented lesions, which form a reticular pattern. These lesions are also known as “fishing net with knots” and are seen primarily in the posterior pole.^(1,2) Autosomal dominant and recessive inheritance patterns have been described. While some affected individuals may develop central visual impairment, others may be asymptomatic.⁽³⁾

Also known as Sjögren’s dystrophy, this condition is part of the spectrum of dystrophies of the retinal pigment epithelium (RPE), a group of disorders comprising five types of dystrophies with variable inheritance patterns: adult-onset vitelliform macular dystrophy, butterfly-shaped pattern dystrophy, multifocal pattern dystrophy mimicking *fundus flavimaculatus*, *fundus pulverulentus* and reticular pigmentary retinal dystrophy (Figure 1). Pattern dystrophies affect primarily the macula and manifest as bilateral asymmetric lesions, which are often chronic and characterized by slow progression.^(3,4)

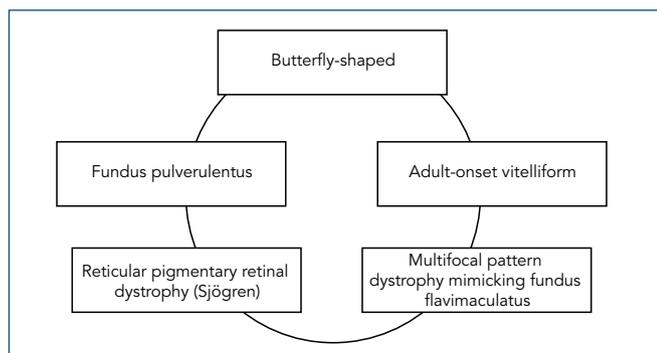


Figure 1. Retinal pigment epithelium dystrophies.

In most cases of reticular pigmentary retinal dystrophy, visual function tends to be preserved, especially in the early stages of the disease. Associations between this type of dystrophy, choroidal neovascularization (CNV) and/or geographic atrophy have seldom been reported, but may explain cases with poor vision and unfavorable prognosis.^(2,4) Spontaneous reduction of subretinal fluid collection in response to CNV has been reported in some studies, whereas others have shown good therapeutic response to intravitreal injection of anti-vascular endothelial growth factor, with anatomical as well as functional improvement.^(4,5)

This study describes a case of reticular pigmentary retinal dystrophy with visual loss due to CNV, which responded well to intravitreal treatment with ranibizumab.

CASE REPORT

A healthy 71-year-old white male patient with controlled hypertension and no other systemic or ocular comorbidities is described. There was no history of consanguineous marriage in his family and no other relevant background.

The patient reported progressive bilateral visual loss, particularly in the right eye (OD) for 2 months. He denied changes in color vision and had no other ocular complaint. Best-corrected visual acuity was 20/800 and 20/200 in the right and the left eye (OS) respectively. Ultrasound biomicroscopy of the anterior segment of the left and right eyes (OS and OD) was unremarkable.

Fundus examination of the OD revealed diffuse rarefaction of the RPE and macular atrophy, seen as an hypopigmented area measuring approximately 1.5 times the disc diameter (1.5 units of disc area) and associated with superficial pigment accumulation. Left eye fundus examination revealed diffuse rarefaction of the RPE and a disciform scar measuring approximately one disc diameter (1 unit of disc area), which was associated with adjacent macular hemorrhage.

Optical coherence tomography (OCT) (Rtvue, Optovue, Fremont, CA, USA) showed a diffuse decrease in retinal thickness suggestive of atrophy in the OD. In the OS, fibrovascular RPE detachment with subretinal fluid was seen, suggesting active subretinal neovascular membrane (Figure 2).

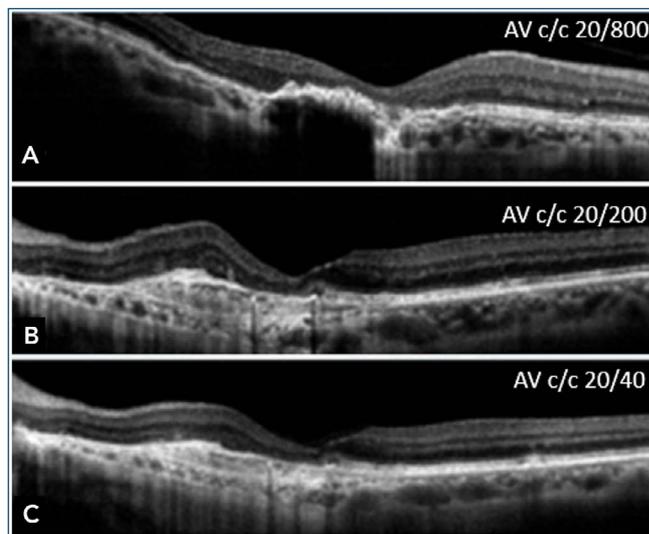


Figure 2. (A) Right eye: Optical coherence tomography image showing macular atrophy. (B) Left eye: Optical coherence tomography image consistent with choroidal neovascularization prior to anti-vascular endothelial growth factor injection. Note fibrovascular retinal pigment epithelium detachment and presence of subretinal fluid. (C) Left eye: Optical coherence tomography image acquired after anti-vascular endothelial growth factor injection. Note reestablishment of the foveal depression and absence of subretinal fluid.

Fluorescein angiography (TRC 50x, Topcon, Tokyo, Japan) revealed hyperfluorescence leakage in the OS macula, suggesting CNV. A window defect corresponding to other areas of atrophy was also seen. The asymmetric hypofluorescent network formed a mild reticular pattern in the posterior pole of both eyes.

After being diagnosed with age-related macular degeneration (AMD) with active neovascular membrane in the OS and macular atrophy in the OD, the patients received 12 intravitreal injections of 0.5 mg of ranibizumab (Lucentis, Genetech Inc., San Francisco, CA, USA) in the OS over a period of 8 years, in a *pro re nata* (PRN) regimen. Good therapeutic response was achieved, with best-corrected visual acuity of 20/40 in this eye. His only complaint was slow vision recovery after intense light exposure.

The patient was reexamined after 8 years. At this time, larger atrophic areas were seen in both eyes, with new macular bleeding the OS. Areas of mild accumulation of yellowish pigment were also detected in the posterior pole of both eyes (Figure 3).

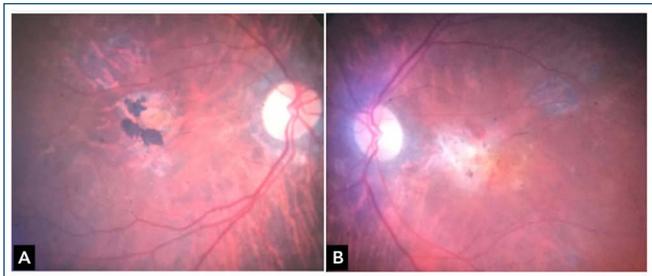


Figure 3. Fundus photographs. (A) Right eye: macular atrophy with pigment accumulation on the macular surface. (B) Left eye: disciform scar associated with adjacent macular hemorrhage (TRC 50x, Topcon, Tokyo, Japan).

Fluorescein angiography was repeated and revealed increased hypofluorescence due to a reticular pattern of pigment, suggesting a diagnosis of pigmentary retinal dystrophy in this patient (Figure 4).



Figure 4. Fluorescein angiography at the 8-year follow-up: Increased hypofluorescence due to blocking caused by a reticular pattern of pigment. Late hyperfluorescence leakage in the left eye, suggesting active subretinal neovascular membrane.

The patient was kept on a PRN treatment regimen consisting of intravitreal anti-VEGF injection in the OS, with good anatomical and functional responses. Right eye macular atrophy remained relatively stable the treatment period.

DISCUSSION

Reticular pigmentary retinal dystrophy has been described in the 1950s. However, there has been a recent increase in the number of cases reported, especially after the advent of OCT and the improvement of autofluorescence imaging.⁽⁶⁾ Central vision is usually preserved in Sjögren dystrophy, except in a few cases characterized by geographic atrophy and/or CNV.^(7,8) In the case reported, the patient already had these complications in the first examination, which revealed concurrent atrophy in the OD and an active neovascular membrane in the OS, with consequent visual impairment.

Choroidal neovascularization may be seen in diseases with RPE involvement. Pigment accumulation and RPE rarefaction may occur and lead to choroidal new vessel formation. Changes in Bruch's membrane may also play a role in the pathophysiology of this type of neovascularization. It has been postulated that newly formed vessels in the subretinal membrane grow towards the retina through a tear in Bruch's membrane caused by multiple factors, which ultimately leads to degeneration and tissue traction, just like in other neovascular membranes, such as those associated with angioid streaks, choroiditis and age-related macular degeneration.⁽⁹⁾

Since 1982, the role of pigmentary alteration in the recruitment of new vessels to the subretinal space in macular dystrophies has been demonstrated in experimental studies.⁽¹⁰⁾ In the case reported, this pigment migration was seen on fluorescence angiography.

The reticular pattern may not readily detected, contributing to reticular dystrophy underdiagnosis and making the distinction from other diseases associated with CNV and macular atrophy difficult. In the case reported, the patient received a first diagnosis of AMD due to late manifestation of the macular pattern typical of Sjögren's dystrophy. However, there were no major consequences, since both diseases require the same treatment.

In most cases of reticular pigmentary retinal dystrophy reported in the literature, treatment was not required. In cases with visual impairment due to active CNV, spontaneous regression of subretinal fluid has been described, with subsequent improvement of visual acuity.⁽²⁾ However, in this case intravitreal anti-VEGF

(ranibizumab) injection was selected to treat the neovascular membrane for the sake of appropriate treatment.

The literature describing the use of intravitreal anti-VEGF injection for treatment of CNV secondary to Sjögren dystrophy is scarce. This report describes the 8-year follow-up of a patient with CNV secondary to RPE dystrophy, who was successfully treated with intravitreal anti-VEGF injection, suggesting a good long-term prognosis.

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