Retinal Angiopathy in Familial Amyloidotic Polyneuropathy with Transthyretin V30M Mutation: a Case Report

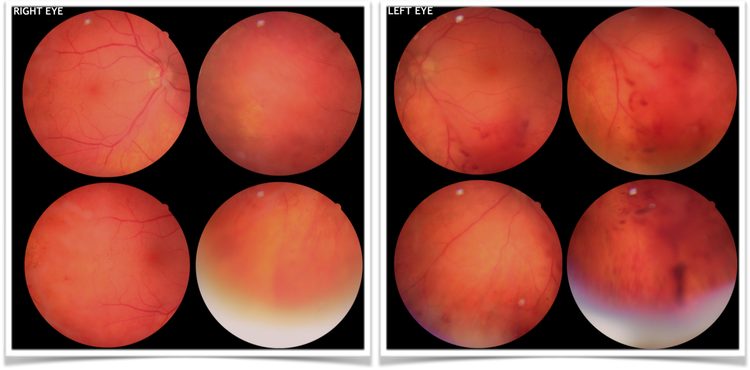
Annelise Nicotti Gonçalves, Leandro Jerez Chaves, Ricardo Okada Nakaghi, Renata Moreto, Marcelo Menegatti Esperandio, Rodrigo Jorge
Hospital das Clínicas da Faculdade de Medicina de Ribeirão Preto. Universidade de São Paulo, Ribeirão Preto - SP

INTRODUCTION

Familial amyloidotic polyneuropathy (FAP) is a group of rare autosomal dominant conditions that result from extracellular amyloid deposition in several target organs, including the eye. We describe a patient with FAP and retinal manifestations.

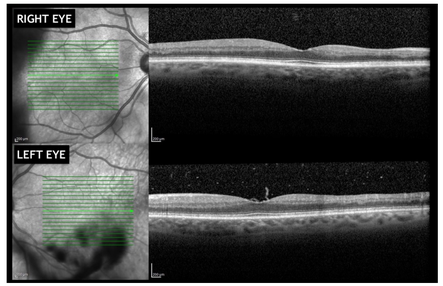
CASE DESCRIPTION

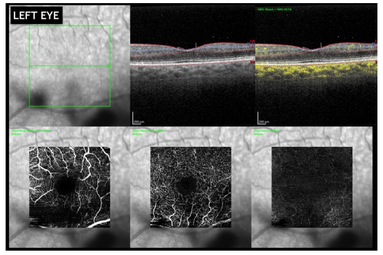
A 77-year-old man was referred for evaluation of left eye floaters for eight months. His medical history was positive for systemic arterial hypertension, psoriatic arthritis, FAP and prostate cancer, for which he received a prostatectomy 12 years before. He had an ophthalmological history of cataract surgery in both eyes. At presentation, his best-corrected visual acuity was 20/20 in his right eye and 20/25 in his left eye. Slit-lamp biomicroscopy was unremarkable, with no evidence of abnormal amyloid deposition nor neovascularization. Posterior segment examination revealed retinal angiopathy with multiple microaneurysms, dot hemorrhages and vitreous opacities in both eyes. Left eye fundus examination revealed vitreous hemorrage but no neovascularitazion was noted.

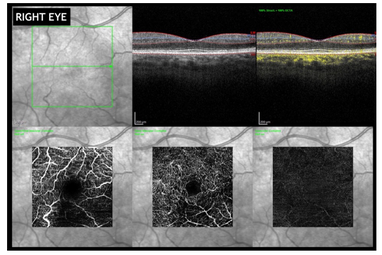


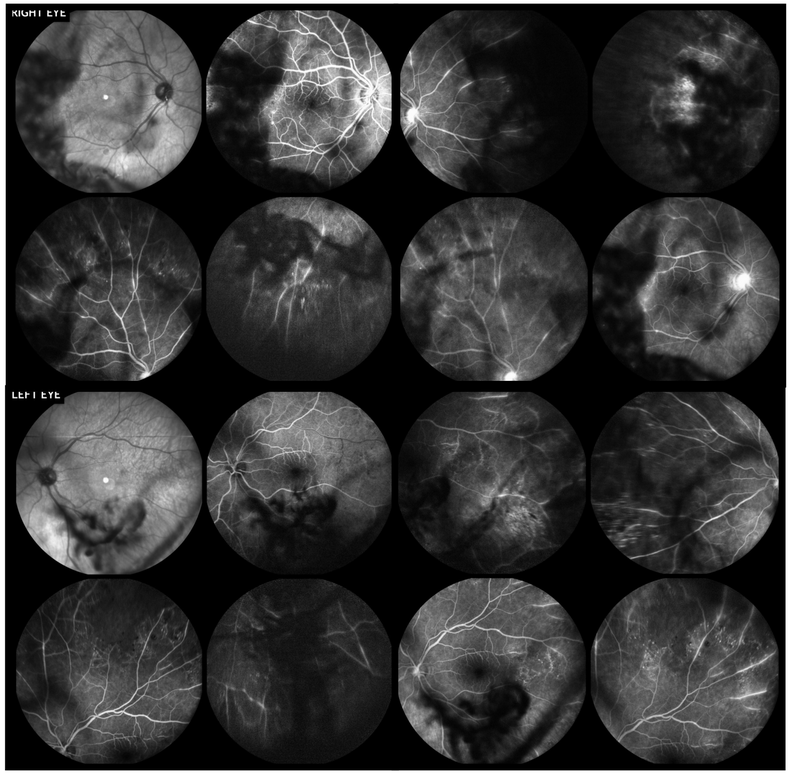
CASE DESCRIPTION

Fluorescein angiography showed large areas of non perfusion in periphery with multiple microaneurysms, capillary telangiectasias and hypofluorescence from blockage from vitreous opacities. A diagnostic work-up for diabetes mellitus, and infectious conditions was negative. Because of family history of amyloidosis, 15 years previously, before he had any clinical manifestation, he underwent genetic study that confirmed the presence of amyloidogenic V30M mutation. He was already receiving treatment with tafamidis and systemic immunosuppression with adalimumab for 2 years because of the diagnosis of PAF. The diagnosis of amyloidosis retinal angiopathy was presumed and, after obtaining the written consent, patient was treated by panretinal laser photocoagulation in both eyes.









DISCUSSION

We describe a rare case of retinal angiopathy in a patient with confirmed transthyretin V30M form of familial amyloidotic polyneuropathy, the least common ocular manifestation of this disease.