OCULAR INVOLVIMENT IN A TRANSTHYRETIN FAMILIAL AMYLOIDOSIS: A CASE REPORT

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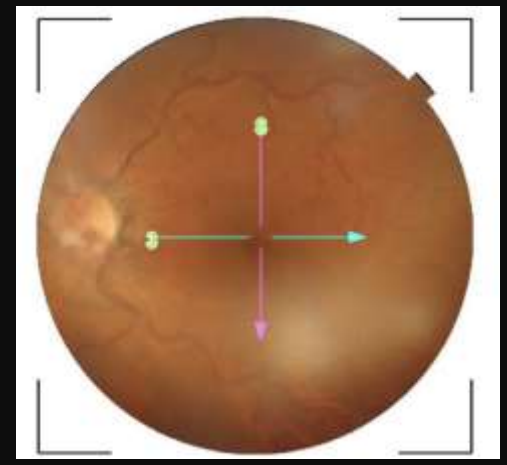
Case Presentation

M. J. S. M, 68 years old, female, from São Luís-MA, Brazil, was admitted to the ophthalmology service of the UDI Hospital being referred by the dermatologist due to visual loss in left eye and a slightly pigmented, non-erythematous skin lesion on the right arm with no change in sensitivity over the past 3 months. The laboratory tests investigation were negative to rheumatological diseases and the CT scan of the Chest showed no alterations suggestive of tuberculosis or other granulomatous diseases. At presentation, her best corrected visual acuity was 20/200 in left eye and 20/30 in right eye and intraocular pressure was 20 bilaterally. Anterior segment examination showed bilateral initial cataract, but no abnormalities were seen in the conjunctiva, cornea, iris, pupil, or trabecular meshwork.

Case Presentation

- Dilated fundoscopy in the left eye revealed vascular tortuosity, crowding cupping of the optic nerve and peripapillary atrophy in addition to yellowwish-white glass-wool vitreous opacitiy and in the right eye was normal. Retinography shows yellowwish-white glass-wool vitreous opacity in LE (image 1). Optical coherence tomography reveals subtle thickening of the outer layers of the retina, a foveal cyst and microaneurysms in the outer plexiform layer (Image 2). Optical coherence tomography angiography showed choriocapillaris ischemia (Image 3). Thus, the hypothesis of amyloidosis was suggested and a skin biopsy of the lesion was performed, which revealed findings suggestive of amyloidosis and vitrectomy was required

Image 1



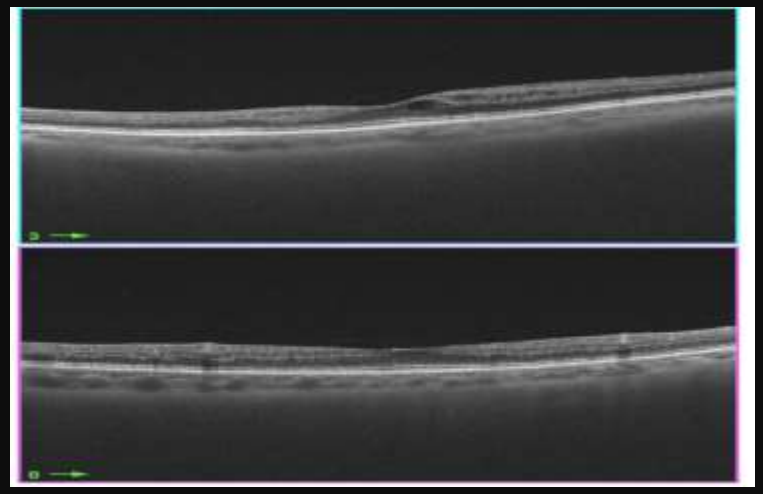
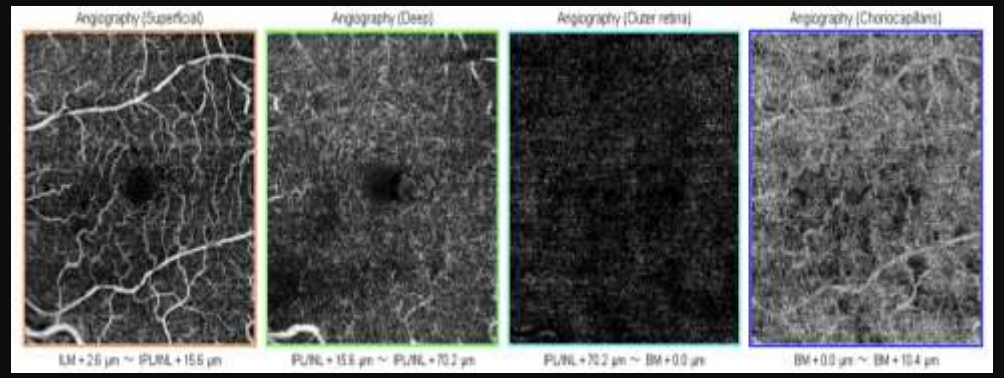


Image 2

Image 3



Discussion

- Ocular manifestations of TTR amyloidosis are believed to be secondary to amyloid deposition within different structures of the eye. The most common ocular manifestation of TTR amyloidosis was vitreous involvement. The symptoms of vitreous opacity are floating objects in front of the eyes and progressive vision loss. Ocular findings in patients with familial TTR amyloidosis may be secondary to either the systemic production of dysfunctional TTR or production by the RPE. Vascular abnormalities, for instance, may be secondary to systemic TTR production. Vitreous amyloid is postulated to be secondary to RPE production of TTR. Another important ocular manifestation is secondary glaucoma. The deposition of amyloid protein around blood vessels in the conjunctiva and sclera can lead to an increase in scleral venous pressure and intraocular pressure; ocular amyloid deposition blocks the trabecular meshwork and Schlemm canal, and the discharge of aqueous humor is blocked, resulting in glaucoma.

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