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VITREORETINAL COMPLICATIONS SECONDARY TO FAMILIAL AMYLOID POLYNEUROPATHY (FAP): DIAGNOSIS AND MANAGEMENT

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Introduction:

Familial Amyloid Polyneuropathy is a hereditary disorder (autosomal dominant) of systemic extracellular fibrillar deposits due to a protein misfolding. Transthyretin (TTR) is the mutated protein and is produced predominantly in the liver. FAP is endemic in Portugal, Sweden and Japan with different mutations. The main manifestations are polyneuropathy and cardiomyopathy thus a delayed diagnosis can be life threatening. Ocular involvement is reported in 10% of the patients, sometimes it is present before the systemic symptoms. The incidence of vitreous opacities varies from 5-35%. It has also been proven that these opacities can progress faster after liver transplantation. Perivascular deposits are present in some cases leading to microvascular permeability alterations. The aim of this study was to present two cases of FAP with specific features and treatment, one patient developed vitreous opacities 1 year after liver transplantation and the other (novel mutation) presented extensive peripheral occlusions.

Materials and methods:

We included two non-related patients with vitreous opacities secondary to FAP confirmed by genetic testing. Demographics, ocular and systemic alterations were reported, pathology result, vitrectomy findings and treatment were also reported.

Results:

Case 1: 55-year-old female with a known TTR gene mutation (p.S50R,T>A) and a progressive loss of vision of 1 year that started after liver transplantation, as part of the systemic treatment. Best corrected visual acuity (BCVA) of hand movement, normal intraocular pressure (IOP) and no inflammation signs in anterior chamber were found in both eyes. Glass wool-like appearance-vitreous opacities were found in the vitreous cavity, the periphery of the retina was normal. A vitrectomy with vitreous sample was performed in both eyes, during surgery we found firm hyaloid adhesions to the vessels and the opacities were like waxy paper. The pathology report confirmed the presence of amyloid fibrils with Congo red staining. She recovered a vision 20/20 in both eyes with no further complications after 2 years follow-up.
Case 2: 45-year-old male, with pacemaker for cardiac arrhythmia. Referred to have a progressive loss of vision of 3 years, he presented with hand movement BCVA secondary to total dense whitish vitreous opacities in both eyes, normal anterior chamber and IOP. The B-ultrasound showed hyperechoic opacities with posterior vitreous attachment and attached retina in both eyes. A vitrectomy with vitreous sample was performed in both eyes, same characteristics of the vitreous were seen during vitrectomy, diffuse white deposits were found along the arteries and peripheric ischemic vessels were visible in both eyes. The pathology analysis reported amyloid fibrils confirmed with positive Congo red staining and green birefringence under polarized light. A novel TTR gene mutation was found in this patient (c.251T>A). Autofluorescence showed marked hyperautofluorescent deposits along the arteries and in the fovea. Fluorescein angiography showed extensive peripheric areas with

no perfusion in both eyes without neovascularization, vascular disfunction and microaneurysms were found. After 3 months of each surgery, he started with moderate vitreous hemorrhage and was treated with peripheral laser photocoagulation. Finally, the vision was stabilized achieving 20/40 in both eyes.

Conclusions:

Ocular amyloidosis with vitreoretinal complications are common in patients with FAP , they can present after several years in the course of the disease or they can be accelerated after liver transplantation. The ocular involvement can be the first evident manifestation and can lead to a prompt diagnosis as neurologic or cardiac symptoms can be ambiguous in most of the patients. A wide spectrum of vitreoretinal alterations can be found going from moderate vitreous opacities with excellent functional results after vitrectomy to ischemic vascular abnormalities that may lead to neovascularization as we demonstrated in the case 2 with a novel mutation. Also special considerations should be given during vitrectomy. A multidisciplinary approach should be done as these patients as they can have severe systemic complications.

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