Introduction

- Familial amyloid polynuropathy (FAP) is an inherited disorder with autosomal dominant transmission, characterized by systemic extracellular accumulation of amyloid fibrils. The peripheral nervous system and the heart are the main target organs, but the eye may also be involved.
- Most common type of FAP is related to transthyretin (TTR) and Portugal is an endemic country for FAP TTR Val30Met.
- TTR is mainly produced by the liver, but a few amount is also synthesized in the eye, mainly in retinal pigment epithelium.
- Vitreous amyloid opacities usually occur in advanced stages of the disease or several years after liver transplantation (LT). Nevertheless, vitreous opacities can be the presenting manifestation of the disease in late onset FAP.
- Recurrence of vitreous opacities has been described in the literature. This study reports management of de novo vitreous amyloid opacities after previous pars plana vitrectomy (PPV).

Materials/methods

- Retrospective, observational, consecutive case series of 4 eyes of 3 FAP patients submitted to re-vitrectomy (Ferreira NL), because of de novo vitreous opacities after previous PPV.
- Demographic data, TTR mutation involved, age at beginning of disease, period of evolution of disease, LT, previous ocular surgeries, time between VPP and re-intervention and ocular changes related to FAP were evaluated.
- Surgical procedures of re-intervention included:
  - Phacoemulsification with intra-cular lens implantation in presence of phakic eye
  - Re-vitrectomy as complete as possible
  - Posterior capsulotomy (with vitreous cutter)
  - ILM peeling if presence of wrinkling of internal retinal surface

Results

- All patients were female and TTR Val30Met with late onset disease.
- Mean age of onset was 56 years and average evolution time of disease was 6.5 years.
- Two patients had been submitted to LT 4 and 9 years before and both had lost one eye because of advanced glaucoma. None was on tafamidis medication.
- Two eyes were phakic and one had glaucoma. None had glaucoma surgery history.
- Time between first VPP and surgical re-intervention was longer than 2 years in all cases (24, 34, 26 and 58 months respectively).
- At re-intervention, vitreous opacities were observed behind posterior lens capsule and at vitreous base. ILM peeling was achieved in 3 cases.
- All cases had retinal amyloid microangiopathy (vascular tortuosity and occlusion, peripheral retinal haemorrhages and ischaemia). It was performed Argon lasertherapy in one eye.
- No futher recurrence has been observed after re-intervention (follow-up between 3 and 11 months). Until now, none case had worsening of glaucoma after surgery.

Conclusions

- Amyloid vitreous opacities requiring VPP to restore vision are common in TTR Val30Met patients with late onset disease, even after LT.
- De novo vitreous amyloid opacities may occur several years after PPV. Amyloid deposition in vitreous cavity was observed only in strong vitreous adherence locations (behind posterior lens capsule and at vitreous base).
- Posterior capsulotomy associated with an extensive vitrectomy could be an effective procedure to prevent de novo vitreous amyloid opacities.
- Retinal amyloid microangiopathy was observed in all eyes that needed a re-intervention. Peripheral retina had to be frequently and carefully observed in order to prevent sight-threatening complications because of retinal ischemia.

References