FAMILIAL EXUDATIVE VITREORETINOPATHY: OUR EXPERIENCE

VITREORRETINOPATÍA EXUDATIVA FAMILIAR: NUESTRA EXPERIENCIA

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ABSTRACT

Objective: To describe our experience in the diagnosis of Familial Exudative Vitreoretinopathy (FEVR) at its different clinical stages. We also report our outcomes in early treatment of this inherited disorder.

Methods: Retrospective interventional and descriptive case series of 11 patients (17 eyes) affected by FEVR evaluated and treated in our hospital.

Results: In our series, 33.3% of patients were classified as stage I, while 16.7% and 50% were classified as stage II and III, respectively. Visual acuity (VA) remained stable after treatment in stages I and II (mean VA was 0.8 and 0.4, respectively) but decreased in stage III.

Conclusions: Early treatment of FERV may improve the visual prognosis and avoid complications such as subfoveal chronic exudation and vitreoretinal peripheral interphase contraction and fibrosis (Arch Soc Esp Oftalmol 2008; 83: 703-708).

Key words: Vitreoretinopathy, retina, exudation, dystrophy, vitrectomy.

RESUMEN

Objetivo: Evidenciar la variabilidad de la presentación inicial de la vitreoretinopatía exudativa familiar (VREF) y la importancia de su diagnóstico y tratamiento precoz. Así como, valorar la respuesta al tratamiento de los diferentes estadios clínicos.

Métodos: Realizamos un estudio retrospectivo, intervencional y descriptivo de 11 pacientes (17 ojos) afectos de vitreoretinopatía exudativa familiar tratados en nuestro centro.

Resultados: De los pacientes afectos de vitreoretinopatía exudativa familiar, un 33,3% se encontraban en el estadio I, un 16,6% en el estadio II y el 50% restante en estadio III.

Conclusiones: El tratamiento en estadios iniciales puede mejorar el pronóstico visual y evitar complicaciones como la exudación subfoveal crónica y la contracción y fibrosis en la interfase retina vitrea periférica que pueden provocar ectopia macular.

Palabras clave: Vitreoretinopatía, retina, exudación, distrofia, vitrectomía.
INTRODUCTION

Familial Exudative Vitreoretinopathy (FEVR) is a disease which belongs to the group of vitreoretinal dystrophies (fig. 1) having a low prevalence and dominant autosomal inheritance (1,2). It occurs more frequently in the first decade of life with bilateral expression, although its development is usually asymmetrical (1).

This disease is mainly characterized by peripheral ischemia (2) and alterations in the vitreoretinal interface, both occurring with greater frequency in the temporal area (1,2).

As the disease develops, patients experience loss of vision which may lead to blindness due to the complications derived from the ischemia and fibrovascular proliferation. For this reason, the main objective is the early diagnostic and treatment of the disease.

Three clinical stages are differentiated (3):

— Stage I: defined as the slight degree of the disease in which patients do not exhibit symptoms, but the vitreoretinal interface exhibits alterations such as white with or without pressure and cystoid degeneration as well as avascular areas in the peripheral retina. Less characteristic are vascular ingurgitation and telangiectasiae, microaneurisms and arterial-venous shunts (figs. 2-4).

— Stage II represents the proliferative and exudative stage of the disease, exhibiting neovascularization, fibrovascular proliferation and sub- and intra-retinal exudation. The fibrovascular lesions can cause complications such as macular ectopia and papillary traction events (figs. 5-7).

— In stage III the scar lesion causes tractional, regmatogenous and exudative retina detachment and falciform folds.
On some occasions, the clinical condition becomes complicated with optical atrophy, cataracts, glaucoma and strip keratopathy (figs. 8-10). The treatment of this disease varies according to each stage. In stage 1, when patients are usually free of symptoms, treatment is focused on prophylaxis by means of photocoagulation (PCG) and cryocoagulation (CCG) (4). In Stage II the treatment must focus on releasing the peripheral retinal tractions by dissecting membranes, placing scleral cerclage and pars plana vitrectomy (PPV).

![Fig. 5: Patient in stage II. The image shows small fibrovascular proliferations in the posterior pole.](image)

![Fig. 7: The same case, showing the rupture of the temporal peripheral vascular network.](image)

![Fig. 6: FAG of the posterior pole of the same case, showing fluorescein leak through telangiectasia-like vascular alterations and partial foveal ischemia (rupture of the peripheral capillary network).](image)

![Fig. 8: Eye in Stage III, showing vascular tortuosity, subretinal exudation and tractional retinal strips.](image)

![Fig. 9: Another stage III diagnosed case showing vascular tortuosity, subretinal exudation and exudative DR.](image)
In Stage III surgical options are combined into the advanced stage of the disease (5).

SUBJECTS, MATERIAL AND METHODS

This paper presents a retrospective, intervention- al and descriptive study of patients affected by familial exudative vitreoretinopathy treated in our centre.

The objective of this study is to evidence the variability of the initial presentation of familial exudative vitreoretinopathy and the importance of its early diagnostic and treatment. In addition, it aims at assessing responses to treatments in the different clinical stages of the disease.

In all, 17 eyes corresponding to 11 patients were studied.

RESULTS

The age of patients ranged between 6 and 24, with a male/female ratio of 3:1.

The distribution of patients on the basis of the stage was as follows:

- 33.3% of patients were in stage I,
- 16.6% were diagnosed in stage II and the remaining 50% were in Stage III.

The treatment applied to our cases was as follows.

- 15.3% of the patients in Stage I received a PCG session.
- The remaining 18% received one PCG session and one CCG session in the temporal area, which had to be completed subsequently to 360°.
- The visual acuity of patients remained stable, in a mean value of 0.8.

In Stage II, all the patients received one PCG session and in time were also submitted to the vitrectomy (PPV) as well as scleral cerclage. In these cases the VA also remained stable with a mean value of 0.4.

The cases diagnosed as belonging to Stage III (50%) received several PCG and CCG sessions. In addition, PPV, dissection of posterior hyaloids and membranes were carried out as well as placement of scleral cerclage. The final VA of these cases deteriorated (mean VA of 0.05).

DISCUSSION

FEVR was described in 1969 by Criswick and Schepens (6), who analyzed six patients with clinical conditions similar to those exhibited by premature retinopathy (PR) but without personal history of prematurity or oxygen supplementation.

This paper describes a retrospective, interventional and descriptive study of 17 eyes belonging to 11 patients affected by FEVR. As with PR, this pathology is characterized by the presence of ischemia areas in the periphery of the retina, which are an essential characteristic for the diagnostic.

In our case, that diagnostic was suspected on the basis of clinical findings and confirmed by fluorescein angiography carried out in all patients. One third (33.3%) of eyes in our study only exhibited peripheral retinal ischemia and were classified in stage I. The patients classified as Stage II (16.6%) also exhibited fibrovascular proliferation, neovascularization and subretinal exudation. The remaining 50% of eyes exhibited a cicatricial component associated to a tractional, regmatogenous and exudative retinal detachment. Accordingly, it is important to determine the variability of clinical presentation and carry out a differential diagnostic with other pathologies such as Eales disease, premature retinopathy and others which develop peripheral ischemia.

FEVR is a genetically heterogeneous disease because it can exhibit different hereditary patterns (1). The most frequently described pattern is dominant autosomal (DA). The literature also describes cases of recessive autosomal inheritance (RA),
recessive inheritance and linked to chromosome X as well as some sporadic cases. In addition, we also found in literature different genetic mutations responsible for the different forms of inheritance of FEVR.

The alteration of FZD4, the gene which encodes receptor Wnt Frizzled-4 and which occupies locus EVR1, is located in chromosome 11 locus 11q 13-23 and is responsible for the DA inheritance forms.

The second locus or EVR2 corresponds to the alteration of the same gene which is responsible for Norrie’s disease. Its mutation has been described in FEVR cases linked to chromosome X.

A further genetic alteration, locus EVR3, is that of the gene located in chromosome 11 locus 11p 12-13, the characteristics of which are as yet unknown.

A fourth gene present in locus EVR4 is gene LRP5 which encodes a low density lipoproteic receptor which is located in chromosome 11 locus 11q13. This gene is also considered responsible for some DA forms of the disease.

Finally, the RA forms have also been described although the genetic defect which accounts for these forms still remain unknown.

The management of these patients is a matter of controversy. Not all authors defend prophylaxis with photocoagulation (PCG) or cryocoagulation (CCG) even though the angiography evidences ischemic areas in the peripheral retina. According to the majority of researchers, the main indication for prophylactic treatment is peripheral neovascularization and exudative retinal detachment, although the results of this approach have not been conclusive (7,8). 15.3% of the eyes diagnosed in Stage I of our series received one session of PCG while the rest also received 1 CCG session. In Stages II and III, according to the degree of evolution of the disease, the therapeutic options ranged from PCG and CCG to PPV and scleral cerclage, as well as peeling of membranes and dissection of fibrovascular proliferations.

Therefore, we believe that a diagnostic in the early stages is important to improve the visual prognosis, as well as early treatment to avoid complications such as chronic subfoveal exudation and contraction and the process in the peripheral retina-vitreous interface which could cause macular ectopia.

REFERENCES