Familial exudative vitreoretinopathy complicated with full thickness macular hole

A case report

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Abstract

Rationale: To report a case of familial exudative vitreoretinopathy (FEVR) complicated with full-thickness macular hole (FTMH).

Patient concerns: A 39-year-old male presented after becoming aware of metamorphopsia in his left eye.

Diagnoses: Fundus examination showed a retinal avascular area, retinal vascular abnormality, and yellow exudation at the peripheral retina in both eyes. Optical coherence tomography (OCT) findings revealed impending MH (IMH) due to posterior vitreous detachment (PVD) in his left eye. Despite of the occurrence of spontaneous complete PVD, an FTMH developed at 4 months after the onset of IMH.

Interventions: To treat the FTMH, vitreous surgery was performed. Intraoperative findings revealed that the thick posterior vitreous membrane (PVM) had no adhesions with the edge of the FTMH. However, a thin epiretinal membrane (ERM) was observed around the MH.

Outcomes: Postoperatively, the FTMH was closed, and the patient’s corrected visual acuity improved from 0.4 to 0.8.

Lessons: In this present case, an IMH developed via traction by a thick PVM, characteristic of FEVR, with an FTMH then developing via traction by a thin ERM. Our findings reveal that it is vital to fully understand these anatomical features before performing vitreous surgery for FTMH complicated with FEVR.

Abbreviations: ERM = epiretinal membrane, FEVR = familial exudative vitreoretinopathy, FTMH = full-thickness MH, ILM = inner limiting membrane, IMH = impending MH, MH = macular hole, OCT = optical coherence tomography, OD = oculus dexter, OS = oculus sinister, PVD = posterior vitreous detachment, PVM = posterior vitreous membrane, RD = retinal detachment, ROP = retinopathy of prematurity, VTMS = vitreo-macular traction syndrome.

Keywords: epiretinal membrane, familial exudative vitreoretinopathy, macular hole, posterior vitreous detachment, vitrectomy

1. Introduction

Familial exudative vitreoretinopathy (FEVR) is somewhat frequently observed in cases of vitreoretinal dystrophy, and its pathology is similar to that of retinopathy of prematurity (ROP), thus resulting in vitreous degeneration and retinal vascular dysplasia.[1–3] Generally, in FEVR cases, the retinal avascular area is mainly formed around the temporal peripheral side, and retinal vessels are linearized and polytomous. An abnormal blood vessel anastomosis is often observed at the boundary with the avascular area, and exudative change occurs around the retinal vessels. Frequently, ocular complications such as a dragged retina, rhegmatogenous and/or tractional retinal detachment (RD), and epiretinal membrane (ERM), etc., can occur. However, and to the best of our knowledge, there have only been 2 previous reports on concomitant macular holes (MHs) and FEVR.[4,5] Here we report a case of a young male in whom an impending MH (IMH) developed via traction by a thick posterior vitreous membrane (PVM), characteristic of FEVR, with a full-thickness macular hole (FTMH) then developing via traction by a thin ERM.

2. Case presentation

This study involved a 39-year-old male who had been undergoing followed-up observations at a nearby clinic due to being diagnosed with FEVR. Around October 2016, he became aware of metamorphopsia in his left eye, and was diagnosed as IMH based on the findings of an optical coherence tomography (OCT) examination performed on November 21, 2016 (Fig. 1A). Although it seemed to indicate the need for vitreous surgery, only follow-up observation was performed due to the development of complete posterior vitreous detachment (PVD). Thereafter, the IMH showed the tendency of remission on December 19, 2016.
However, it subsequently advanced to an FTMH, and the patient was referred to the Department of Ophthalmology, Osaka Medical College, Takatsuki City, Japan to undergo vitreous surgery on March 29, 2017. Although the patient had no history of ocular trauma or ROP, however, his mother had been diagnosed with FEVR. This study was approved by the Ethics Committee of Osaka Medical College, and was performed in accordance with the tenets set forth in the Declaration of Helsinki. Informed written consent was obtained from the patient.

Upon initial examination at our clinic, the patient’s visual acuity was (1.0/C2S/C0 4.50 D = C/C0 2.25 D Ax 165°) OD and (0.4/C2S/C0 5.25 D = C/C0 3.00 D Ax 180°) OS, and his intraocular pressure was 13mm Hg OD and 12mm Hg OS. Both eyes showed a retinal avascular area, retinal vascular abnormality, and yellow exudation at the peripheral retina (Fig. 2A and B). Vitreous liquefaction and degeneration were observed in both eyes, and a shrunken PVM was floating in the lower vitreous cavity of the right eye. In the left eye, OCT findings revealed stage 4 FTMH (Fig. 2C) and a thick PVM located on the anterior surface of the retina (Fig. 2D).

Vitreous surgery was performed for the FTMH in the patient’s left eye. During the surgery, the core vitreous gel was first resected, and the thick PVM was excised from the posterior pole toward the periphery. Although the thick PVM had no adhesions with the edge of the FTMH (Fig. 3A), a thin ERM was observed around the MH. After staining the inner limiting membranes (ILM) with brilliant blue G, we peeled off the ERM, along with the ILM (Fig. 3B). The ERM was particularly strongly adhered to the edge of MH at the temporal side, and the MH presented as a somewhat elliptical shape. Artificial PVD was stopped at the mid-periphery due to the firm adhesion between the thick PVM and the retina. Then, fluid-air exchange and a 20% SF6 gas tamponade were performed, thus completing the operation. Postoperatively, the MH was closed, and the patient’s corrected visual acuity improved from (0.4) to (0.8) at 3 months after surgery (Fig. 4A and B).

3. Discussion

In FEVR cases, vitreous liquefaction and degeneration usually becomes more intense compared to normal healthy subjects of the same age, and a thick PVM often tightly adheres to the retina from the posterior pole to the periphery.[1–3] It is widely known that in FEVR cases, ocular complications, such as rhegmatogenous and/or tractional RD and ERM, occur due to the abnormal vitreous structure. It has been reported that in FEVR patients, localized PVD sometimes causes vitreo-macular traction syndrome (VMTS).[6] Shimouchi et al reported 2 cases of FEVR combined with macular disorders, one of which being
a 22-year-old female and the other being a 14-year-old male. In both cases, there was narrow vitreo-retinal adhesion in the fovea accompanied with perifoveal PVD.[7] Normally, it is rare for young people (i.e., aged 10–20 years) to develop MH and/or VMTS with such perifoveal PVD.[8,9] However, many FEVR cases are complicated with thick PVM which causes strong vitreo-retinal traction to the macula.

In addition, it has been reported that PVD is promoted by intraocular inflammation.[10] In FEVR cases, prolonged intraocular inflammation due to exudative lesions in the peripheral avascular area can cause contraction of the vitreous gel. In this present case, the findings of the OCT imaging performed at the nearby clinic revealed more extensive perifoveal PVD and thick PVM, which generated the IMH. It should be noted that complete PVD usually accelerates a spontaneous closure of the IMH. However, in this present case, the FTMH developed long after the spontaneous complete PVD. It appeared that a thin ERM had formed beneath the thick PVM, and that FTMH was generated by the tangential traction to the retina.

In recent years, there have been reports of FTMH after vitreous surgery, and many of those reports observed the ERM around the MH at the time of reoperation.[11,12] Many preexisting diseases are prone to vulnerability in the macular region, such as ERM, rhegmatogenous RD, and retinoschisis complicated with high myopia. In this present case, due to FEVR, the ischemic and inflammatory disease, as well as due to the traction by the thick PVM, the macula region might have become fragile. We theorize that the traction by the thin ERM was possibly responsible for causing the complete FTMH after complete PVD.[13]

There have been 2 previous reports of FTMH occurring in cases of FEVR. Khwarg et al[4] reported a 9-year-old boy who...
had advanced to FTMH from perifoveal PVD. In that study, laser photocoagulation was first performed on the retinal avascular area, and then vitreous surgery was performed at the time when the tractional RD had progressed. Intraoperative findings indicated ERM around the MH. In a study by Bochicchio et al, the authors reported a 28-year-old female, in whom the findings of her condition were similar to that of Coat disease, who had thick PVM and lamellar MH which had progressed to FTMH after the initial vitreous surgery. She underwent reoperation, thus resulting in closure of the MH. It should be noted that there are some common points between the 2 previous reports described above and our present case. However, and to the best of our knowledge, this is the first report of a case in which IMH had first been generated by the traction of the thick PVM, characteristic of FEVR, with FTMH then developing via the traction of the thin ERM.

When performing vitreous surgery on MH associated with FEVR, special attention must be paid to the fact that the thick PVM is firmly adhered to the retina, thus making it difficult to create an artificial PVD. In conclusion, since FTMH complicated with FEVR has a relatively different pathogenesis from idiopathic FTMH, it is vital to fully understand these anatomical features before performing vitreous surgery.

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References

[1] Criswick VG, Schepens CL. Familial exudative vitreoretinopathy. Am J Ophthalmol 1969;68:578–94.

[2] Nicholson DH, Galvis V. Criswick-Schepens syndrome (familial exudative vitreoretinopathy). Study of a Colombian kindred. Arch Ophthalmol 1984;102:1519–22.

[3] Ranchod TM, Ho LY, Drenser KA, et al. Clinical presentation of familial exudative vitreoretinopathy and macular hole exhibited in same individual. Semin Ophthalmol 2007;22:85–6.

[4] Khwarg JW, Bouria D, Gonzales CA, et al. Familial exudative vitreoretinopathy: spectral-domain optical coherence tomography of the vitreoretinal interface, retina, and choroid. Ophthalmology 2015;122:2270–7.

[5] Bochicchio S, Pellegrini M, Cereda M, et al. Macular hole in a young patient affected by familial exudative vitreoretinopathy. Retin Cases Brief Rep 2017;doi: 10.1097/ICB.0000000000000613. [Epub ahead of print].

[6] Yonekawa Y, Thomas BJ, Drenser KA, et al. Familial exudative vitreoretinopathy: spectral-domain optical coherence tomography of the vitreoretinal interface, retina, and choroid. Ophthalmology 2015;122:2270–7.

[7] Shimosuchi A, Takahashi A, Nagaoka T, et al. Vitreomacular interface in patients with familial exudative vitreoretinopathy. Int Ophthalmol 2013;33:711–5.

[8] Johnson MW. Perifoveal vitreous detachment and its macular complications. Trans Am Ophthalmol Soc 2005;103:537–67.

[9] Duker JS, Kaiser PK, Binder S, et al. The International Vitreomacular Traction Study Group classification of vitreomacular adhesion, traction, and macular hole. Ophthalmology 2013;120:2611–9.

[10] Hogan MJ. Inflammation and its effect on the vitreous. Trans Ophthalmol Soc U K 1975;95:378–81.

[11] Khurana RN, Wykoff CC, Bansal AS, et al. The association of epiretinal membrane with macular hole formation after rhegmatogenous retinal detachment repair. Retina 2017;37:1073–8.

[12] Gao X, Ikuno Y, Fujimoto S, et al. Risk factors for development of full-thickness macular holes after pars plana vitrectomy for myopic foveoschisis. Am J Ophthalmol 2013;155:1021.e1–7.e1.

[13] Tsai CY, Hsieh YT, Yang CM. Epiretinal membrane-induced full-thickness macular holes: the clinical features and surgical outcomes. Retina 2016;36:1679–87.

[14] Ikeda T, Fujikado T, Tano Y, et al. Vitrectomy for rhegmatogenous or tractional retinal detachment with familial exudative vitreoretinopathy. Ophthalmology 1999;106:1081–5.

[15] Yamane T, Yokoi T, Nakayama Y, et al. Surgical outcomes of progressive tractional retinal detachment associated with familial exudative vitreoretinopathy. Am J Ophthalmol 2014;158:1049–55.