CASE REPORT

Familial exudative vitreoretinopathy presenting with unilateral rhegmatogenous retinal detachment in a Malay teenager

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SUMMARY

To describe a case of familial exudative vitreoretinopathy presenting with unilateral rhegmatogenous retinal detachment in a Malay teenager.

BACKGROUND

We wish to share with our colleagues a case of familial exudative vitreoretinopathy (FEVR) which was almost undetected as the patient presented with left rhegmatogenous retinal detachment. This case reminds us to constantly challenge our diagnosis in what seems to be a straightforward case of total retinal detachment with proliferative vitreoretinopathy (PVR) where there was a history of mild blunt trauma. As familial exudative vitreoretinopathy is frequently underdiagnosed, a high index of suspicion is needed and a careful examination of the fellow eye in all patients is mandatory. Accurate diagnosis will warrant lifelong monitoring as the condition may progress at any age. Furthermore, screening of family members is warranted.

CASE PRESENTATION

A 14-year-old Malay boy presented to us with left blurring of vision for 9 months. He vaguely remembered a history of blunt trauma to his left eye prior to the blurring of vision and presented to an ophthalmologist 2 months later. He was then referred to our vitreoretinal (VR) service, but he only came 6 months later.

He was a known case of high myopia in both eyes with the refractive error of $-9D$. He started wearing glasses at the age of 10 years. He was a full-term baby with no history of previous hospital admissions and was otherwise well. He was the sixth out of 10 siblings with no family history of blindness.

On examination, his right vision was best corrected to 6/18 with a refraction of $-9.00/-1.00 \times 10^\circ$ with a near vision of N6. His left vision was hand movement. Dilated fundal examination of the left eye showed total retinal detachment, vitreous haemorrhage and PVR grade C with a break temporal to the macula. His right fundus showed peripheral ghost vessels at the temporal region with subtle macular dragging and straightening of vessels (figures 1 and 2).

INVESTIGATIONS

- He was screened for a uveitic cause to the peripheral ghost vessels which was negative (erythrocyte sedimentation rate, full blood

count, C3/C4, C reactive protein, rheumatoid factor, antinuclear antibody, venereal disease research laboratory, Mantoux test, chest x-ray).

- Fundus fluorescein angiogram (FFA) revealed a diagnosis of FEVR in the right eye as evidenced by extensive peripheral avascular zone in all quadrants with extensive branching and anastomosis of the retinal vessels. Angiographically, leaking new vessels were seen at the temporal region (figures 3 and 4).

DIFFERENTIAL DIAGNOSIS

- Rhegmatogenous retinal detachment with underlying high myopia
- Rhegmatogenous retinal detachment secondary to trauma
- Rhegmatogenous retinal detachment secondary to uveitic causes.

TREATMENT

He had undergone pars planar vitrectomy (PPV)/lensectomy/sclera buckling/retinectomy/endolaser/silicone oil for the left eye. He developed redetachment inferiorly which was subsequently treated with laser. His left eye was complicated with the development of rubeosis and secondary glaucoma. A second surgery of removal of silicone oil (ROSO)/retinectomy/siluron 5000 insertion was then performed for the redetachment (figure 5).

Indirect laser photocoagulation was planned for his right leaking peripheral vessels.

Screening of his siblings was done with FFA, which revealed silent FEVR in four of his siblings.

Figure 1 Fundus photograph of the right eye showing a myopic fundus with tilted disc, subtle macular dragging and straightening of vessels.

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Figure 2 Fundus photograph of the periphery of the right eye with ghost vessels marking the demarcation between vascular and avascular zone.

OUTCOME AND FOLLOW-UP

Vision of the left eye remained poor at hand movement with no light projection nasally as retinectomy was done temporally. Although he had silicone oil in the anterior chamber with band keratopathy postoperatively, his intraocular pressure was not raised. The patient then failed to follow-up.

DISCUSSION

FEVR is rare and often underdiagnosed due to its protean manifestations which could be easily mistaken for other ocular disorders. Its clinical features range from macular dragging, temporal radial retinal folds, retinal neovascularisation, vitreous haemorrhage, tractional retinal detachment and subretinal exudation.¹ In this patient, he had mild macular dragging with straightening of vessels and peripheral ghost vessels, which suggest that previous peripheral retinal neovascularisation has taken place and resolved.

Eyes with severe degree of FEVR present at an earlier age with tractional and exudative retinal detachment (RD), but in

Figure 4 Fundus fluorescein angiogram showing leaking periphery vessels and avascular zone at the inferotemporal periphery of the right eye.

less severe cases, it presents later with rhegmatogenous retinal detachment (RRD) due to globe enlargement frequently from myopia and vitreous liquefaction, both occur later in life.² This is especially true for our patient who developed complication at an older age. Juvenile-onset and male sex are the main characteristics of rhegmatogenous retinal detachment associated with FEVR.²

Retinal surgeries in FEVR eyes are never easy and they are usually complicated with non-attachment and PVR compromising the visual outcome.³ However, early detection and prompt treatment for RRD in FEVR do offer a better outcome with current techniques.⁴ However, the outcome of the surgery is guarded in this patient as he presented late with PVR complications.

FEVR is a progressive disease, and hence its detection is of utmost importance. However, it has an unpredictable timeline of progression which necessitates frequent and lifelong

Figure 3 Fundus fluorescein angiogram showing leaking periphery vessels and avascular zone at the superotemporal periphery of the right eye.

Figure 5 Photograph of the left fundus showing laser scars at periphery with retinectomy done temporally. The globe is filled with silicone oil.

monitoring. It is usually a bilateral disease though the presentation may be asymmetric. As it is a bilateral disease, patients with complication of FEVR need an even further vigilance to preserve the vision in the remaining eye. Family screening and education with timely prophylactic treatment if necessary may prevent the devastating complication suffered by this patient.

Learning points

- ▶ Familial exudative vitreoretinopathy (FEVR) can present with rhegmatogenous retinal detachment in a young patient.
- ▶ FEVR can be missed if the fellow eye of patients are not examined for subtle symptoms such as macular dragging and peripheral regressed new vessels in a patient with no history of prematurity.
- ▶ FEVR presenting with complications (RRD) in a young patient should prompt a screening of first-degree relatives.

Contributors Both authors managed the patient and contributed to the writing up of this report.

Competing interests None.

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REFERENCES

- 1 Ranchod TM, Ho LY, Drenser KA, *et al.* Clinical presentation of familial exudative vitreoretinopathy. *Ophthalmology* 2011;118:2070–5.
- 2 Chen SN, Hwang JF, Lin CJ. Clinical characteristics and surgical management of familial exudative vitreoretinopathy-associated rhegmatogenous retinal detachment. *Retina* 2012;32:220–5.
- 3 van Nouhuys CE. Signs, complications, and platelet aggregation in familial exudative vitreoretinopathy. *Am J Ophthalmol* 1991;111:34–41.
- 4 Ikeda T, *et al.* Vitrectomy for rhegmatogenous or tractional retinal detachment with familial exudative vitreoretinopathy. *Ophthalmology* 1999;106:1081–5.

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