Branch retinal vein occlusion in a case of Best vitelliform maculopathy

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DESCRIPTION

A 35-year-old woman presented to us with a sudden onset diminution of vision in the right eye. She had reduced vision in both eyes since childhood. At presentation, her best corrected visual acuity (BCVA) was 20/120 OD and 20/40 OS. Anterior segment examination was within the normal limits. Dilated fundus examination showed yellow subretinal deposits under the macula in both eyes. OD showed dense flame-shaped intraretinal haemorrhages along the superotemporal (ST) arcade extending to the fovea with dilated and tortuous ST veins. Optical coherence tomography revealed foveal thinning with cystoid macular oedema and minimal subretinal fluid OD and OS showed subretinal hyperreflective deposits with subretinal

fluid ([figure 1](#)). A diagnosis of Best vitelliform maculopathy with right superotemporal branch retinal vein occlusion (BRVO) was made. Intravitreal ranibizumab (0.5 mg/0.05 mL) was injected in the right eye and vision improved to 20/60 at 1 month follow-up. The patient later developed retinal neovascularisation in the ST quadrant with minimal dispersed vitreous haemorrhage. Sectoral laser photocoagulation was performed and the retina was stable for more than a year without vitreous haemorrhage or macular oedema and BCVA of 20/60. However, after 14 months, the patient developed sudden onset of loss of vision preceded by floaters OD. On examination, vitreous haemorrhage obscuring a major part of the retina was noted. Patient was advised observation with a propped up position. The haemorrhage did not clear for >4 weeks and pars plana vitrectomy with additional laser photocoagulation was performed. Vision improved to 20/40 and has remained stable for more than a year till last follow-up ([figure 2](#)).

Best vitelliform macular dystrophy or Best disease is a hereditary macular dystrophy which comes under the spectrum of bestrophinopathies which involves mutation of the gene BEST1 present in the long arm of chromosome 11 (11q12).¹ The disease usually presents itself in childhood and young adults and is characterised by typical egg yolk appearance in the macula. As the disease progresses, it takes the appearance of pseudohypopyon, scrambled egg, atrophic and may even lead to the appearance of choroidal neovascularisation

Figure 1 (A) Right eye fundus showing yellow subretinal lesions under the macula and flame-shaped haemorrhages at the superotemporal quadrant. (B) Left eye fundus showing yellow subretinal lesions with subretinal fluid. (C) Fluorescein angiography of the right eye showing blocked fluorescence at the superotemporal quadrant with altered hypofluorescence and hyperfluorescence at the macula. (D) Fluorescein angiography of the left eye showing altered hypofluorescence and hyperfluorescence with pooling at the macula. (E) OCT of the right eye showing cystoid macular oedema and subretinal hyperreflective deposits with minimal subretinal fluid. (F) OCT of the left eye showing subretinal hyperreflective deposits with subretinal fluid. OCT, optical coherence tomography.

Figure 2 (A) Right eye fundus showing clearance of flame-shaped haemorrhages with sclerosed vein and neovascularisation elsewhere (NVE) (black arrow). (B) Right eye fundus showing persistent vitreous haemorrhage. (C) Right eye fundus following vitrectomy showing sectoral laser marks and stable retina. (D) OCT of the right eye after vitrectomy showing foveal thinning with subretinal hyperreflective deposits. OCT, optical coherence tomography.

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(CNV).² The vitelliform material is known to be accumulation of lipofuscin material of degenerated retinal pigment epithelium (RPE).³ The hallmark of Best disease is reduced Arden's ratio (<1.5) on electrooculography. Best disease has been associated with CNV at later stages, subretinal fibrosis, macular hole and retinal detachment.⁴ This is the first case report in the literature in which Best disease and BRVO have been observed in the same patient. In our report, the patient had been diagnosed with bilateral macular lesion elsewhere since the age of 20 years with BCVA of 6/12 OU and first presented to us when she developed

BRVO in the right eye. The cause of the BRVO was investigated as the patient did not have any systemic comorbidities. Coagulation profile, protein C and S, serum homocysteine, lupus anticoagulant and other investigations for infectious and inflammatory aetiologies were normal. The aetiology of BRVO was not known in our case. However, with this case report, we do not suggest any causal relationship between the two diseases.

Patient's perspective

I have been having subnormal vision since childhood. But I am scared of having repeated loss of vision in the right eye which is affecting my daily life.

Learning points

- Best disease should be considered in patients with symmetrical or asymmetrical yellowish dots under the macula in young adults.
- There is no causal relationship between Best disease and branch retinal vein occlusion.

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