Rare disease

Chronic granulomatous disease and neovascularisation causing a vitreous haemorrhage

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SUMMARY

The authors report the case of a 9-year-old boy with chronic granulomatous disease with longstanding inactive chorioretinal lesions developing a spontaneous vitreous haemorrhage. This was due to an undetected branch retinal vein occlusion and was successfully treated with retinal photocoagulation (laser).

BACKGROUND

Chronic granulomatous disease (CGD) is a disorder whereby phagocytes lacking a functional NADPH oxidase cannot generate superoxide anions.¹ Impaired bacteriocidal function leads frequent infections. Histopathology of eyes with CGD has demonstrated multiple foci of granulomatous inflammation of the choroid and sclera.

CASE PRESENTATION

A 9-year-old boy under routine review was noted to have a left vitreous haemorrhage. He had been diagnosed with CGD at the age of 18 months and was initially referred to the ophthalmology service at 4 years of age with 'retinal lesions'. These lesions remained inactive and untreated for 5 years and predated a successful haemopoietic stem cell transplant. He developed left disc swelling which resolved over 9 months with his visual acuity being unaffected. Examination was otherwise unremarkable. Three years later he developed a left sided vitreous haemorrhage with visual acuities of 6/6 right, 6/36 left (figure 1A,B).

INVESTIGATIONS

A B-scan ultrasound confirmed intragel haemorrhage with a flat retina. Examination under anaesthesia revealed an

inferotemporal branch retinal vein occlusion (BRVO) with new vessels elsewhere, which was treated with indirect laser.

OUTCOME AND FOLLOW-UP

We postulate that the ischaemia of the retina secondary to the BRVO resulted in the development of new vessels and subsequent vitreous haemorrhage. The laser treatment caused regression of the new vessels and visual acuity improved as the vitreous haemorrhage cleared.

DISCUSSION

The ophthalmic manifestations of CGD most frequently described are 'punched out' chorioretinal lesions with associated pigment clumping lying along major retinal vessels.² Other ophthalmic manifestations that have been reported include cystoid macula oedema,¹ exudative haemorrhagic retinal detachment,³ neovascular membrane⁴ and a retinal mass.⁵ Peripheral retinal ischaemia has been previously reported⁴ and treated with pan-retinal photocoagulation. This to our knowledge is the first reported case of CGD associated with a BRVO and vitreous haemorrhage.

Figure 1 (A,B) Fundal photographs. (A) Multiple punched out lesions typical of chronic granulomatous disease and (B) vitreous haemorrhage and similar lesions.

Learning points

- ▶ Retinal lesions are seen in 13–35% of patients with chronic granulomatous disease (CGD) and occur in X linked and autosomal CGD.
- ▶ With increasing reports of adverse visual outcomes associated with CGD, long term vigilant follow-up of these patients is recommended.

Competing interests None.

Patient consent Obtained.

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