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Eye (2007) **21,** 430–431. doi:10.1038/sj.eye.6702562;
published online 29 September 2006

Sir,

Fabry disease manifesting as chronic uveitis—treated with enzyme replacement therapy

Fabry disease, a metabolic disease, is caused by α-galactosidase A deficiency.[1] Its ophthalmic manifestations include vortex keratopathy, cataract, and retinal vessel tortuosity.[2] We report a case of Fabry disease presenting with chronic uveitis, cystoid macular oedema, and marked visual impairment. It responded only temporarily to periocular steroid injections and was finally stabilized under enzyme replacement therapy (ERT). To our knowledge, these unusual manifestations and the treatment have never been reported.

Case report

A 22-year-old man visited us in May 2002 and complained of progressive decreased vision (OU) for 4 years, along with acroparesthesia, hypohidrosis, and heat intolerance. Best-corrected visual acuity (BCVA) was 20/400 (OD) and 20/200 (OS). Ophthalmological exams showed bilateral vortex keratopathy, anterior chamber reaction, anterior subcapsular cataract, vitreous haze, periarterial infiltrate, cystoid macular oedema (Figure 1a and b), and diffuse vascular leakage (Figure 1c and d). Optical coherence tomography (OCT) showed severe macular oedema (Figure 1e and f). Fabry disease was confirmed by enzymatic analysis. Investigations looking for other causes of uveitis were all negative.

Because the clinical condition progressed, a therapeutic trial with posterior subtenon steroid injection (triamcinolone acetonide 20 mg, OU) was performed. Two weeks later, BCVA improved to 20/70 (OD) and 20/100 (OS). The vitreous haze and macular oedema partially resolved (Figure 1g and h). However, the inflammation waxed and waned despite subsequent periocular steroid injections. BCVA decreased to counting fingers at 40 cm (OU) owing to macular oedema, cataract, and severe vitreous haze. He received ERT (agalsidase alfa, 10.5 mg every 2 weeks from September 2002 to November 2003; agalsidase beta, 70 mg every 2 weeks since December 2003) and cataract extraction (OD, November 2002; OS, May 2003). After 24 weeks of ERT, acroparesthesia, hypohidrosis, and heat intolerance relieved and the uveitis-like picture gradually subsided. Subsequently, he underwent *pars plana* vitrectomy (OD) for persistent severe vitreous opacity. Postoperatively, the macula showed mottling change without oedema (Figure 2a). Although no inflammatory signs such as vitreous cells were noted in the left eye, severe vitreous opacity still obscured the fundus and probably affected the vision (Figure 2b). The latest BCVA was 20/400 (OD) and 20/800 (OS).

Comment

The most common retinal manifestation of Fabry disease is large vessel tortuosity.[2] In contrast, this case presented with uveitis, vitreous haze, vascular leakage, cystoid

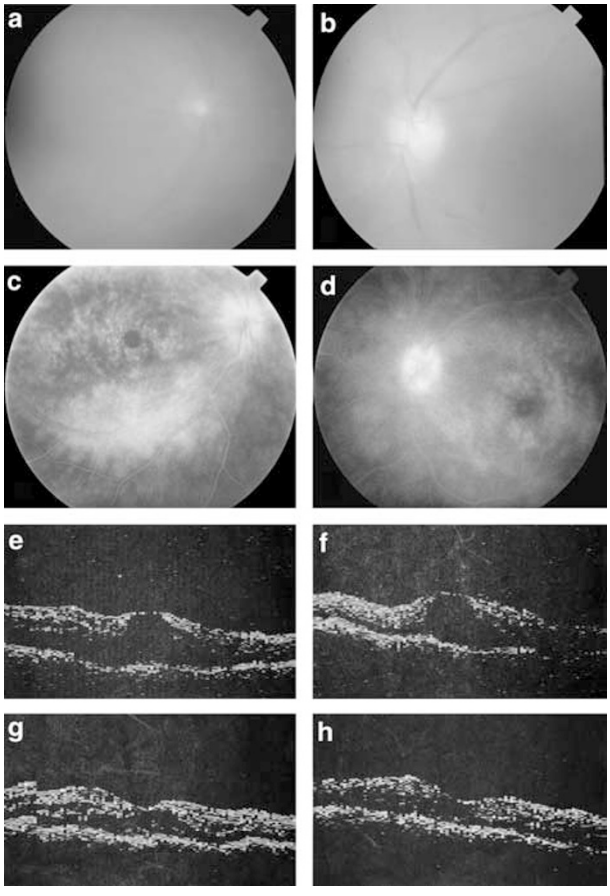

Figure 1 (a, b) Fundus photography showed the vitreous haze obscuring the retina. (c, d) Fluorescein angiography showed vascular leakage, leakage from the optic disc, and cystoid macular oedema. (e, f) OCT before posterior subtenon steroid injection showed thickening of macula (e, OD; f, OS). (g, h) OCT showed significant decrease in macular thickness after posterior subtenon steroid injection (g, OD; h, OS).

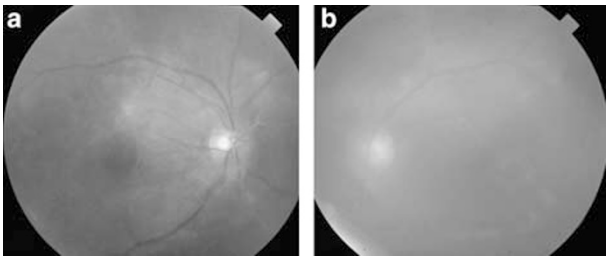

Figure 2 (a) Fundus photography showed macular mottling without oedema in the right eye after *pars plana* vitrectomy. (b) Vitreous haze still obscured retina in the left eye.

macular oedema, and a markedly compromised vision, suggesting diffuse involvement of the intraocular vessels.

Pathology has demonstrated lipid deposition in the vascular smooth muscle cells, the perithelial cells, and the endothelial cells.[3] We believe this may disrupt the blood–ocular barrier, leading to a picture of chronic uveitis. Triamcinolone acetonide has been shown to reduce the breakdown of blood–retinal barrier,[4] introducing the rationale for corticosteroid use in diabetic macular oedema[5] and this case as well.

As the effect of periocular steroid was temporary and the ocular manifestations were well controlled after a prolonged course of ERT alone, the concurrence of Fabry disease and other uveitis seems unlikely. ERT has been shown to be effective in clearing lipid deposition in renal microvascular tissue.[1] In this case, ERT may have reduced the lipid deposition in ocular vascular tissue, leading to clinical improvement.

In conclusion, Fabry disease may significantly alter the vascular permeability and present as chronic uveitis. Periocular steroid injection may temporarily reduce vascular permeability and relieve the ocular manifestations. Long-term ERT could relieve the ocular manifestations.

References

1 Desnick RJ, Brady R, Barranger J, Collins AJ, Germain DP, Goldman M. Fabry disease, an under-recognized multisystemic disorder: expert recommendations for diagnosis, management, and enzyme replacement therapy. *Ann Intern Med* 2003; **138**: 338–346.
2 Sher NA, Letson RD, Desnick RJ. The ocular manifestations in Fabry's disease. *Arch Ophthalmol* 1979; **97**: 671–676.
3 Riegel EM, Pokorny KS, Friedman AH, Suhan J, Ritch RH, Desnick RJ. Ocular pathology of Fabry's disease in a hemizygous male following renal transplantation. *Surv Ophthalmol* 1982; **26**: 247–252.
4 Wilson CA, Berkowitz BA, Sato Y, Ando N, Handa JT, de Juan E Jr. Treatment with intravitreal steroid reduces blood–retinal barrier breakdown due to retinal photocoagulation. *Arch Ophthalmol* 1992; **110**: 1155–1159.
5 Martidis A, Duker JS, Greenberg PB, Rogers AH, Puliafito CA, Reichel E. Intravitreal triamcinolone for refractory diabetic macular edema. *Ophthalmology* 2002; **109**: 920–927.

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The authors have no proprietary interest and receive no research funding

Eye (2007) **21,** 431–432. doi:10.1038/sj.eye.6702517; published online 14 July 2006

Sir,

A Hazard of undiagnosed diabetes with benign prostatic hyperplasia: bilateral endogenous bacterial endophthalmitis

Case report

A 61-year-old male presented with a history of floaters, blurred vision and discomfort for 1 week in his left eye and 2 days in his right. He had recently been polyuric and had lost 10 kg in weight. He was treated for a urinary tract infection 2 weeks prior to presentation.

Examination found best-corrected visual acuity (BCVA) of 6/60 in each eye. There was bilateral panuveitis, and intraocular pressures were 4 mmHg bilaterally. Retinal haemorrhages were present in the right fundus. The left fundus was not visible; a B-scan showed an attached retina. Blood tests showed raised blood glucose (43.5 mmol/l), and marked neutrophilia (29.8×10^9/l).

A diagnosis of bilateral endogenous bacterial endophthalmitis (EBE) was made. The patient was admitted and intravenous insulin started. Bilateral anterior chamber and vitreous biopsies were performed