Sir,

Uveitis and Fabry disease

In the recent article by Shen *et al*[1] entitled 'Fabry disease manifesting as chronic uveitis-treated with enzyme replacement therapy' the provocative presumption is that uveitis in a patient with Fabry disease was improved by enzyme replacement therapy. We have had experience with chronic uveitis in two patients (0.4% of 527 patients in our clinic) with another lysosomal storage disease, Gaucher disease,[2] but we did not see any improvement with specific replacement therapy, despite concern to the contrary.[3] Both patients have otherwise mild Gaucher disease, but one patient has uveitis well-controlled for 12 years solely by local steroids whereas the other patient has suffered progression of uveitis despite 8 years of steroid drops followed by nearly 5 years of enzyme replacement therapy. The patient described by Shen *et al* arrived with best-corrected visual acuity (BCVA) at 20/400 (OD), and 20/200 (OS) which improved to 20/70 (OD), and 20/100 (OS) with steroid injection but then deteriorated. Enzyme replacement therapy for 24 weeks resulted in some apparent improvement, but required pars plana vitrectomy with resulting corneal opacity that left the patient with BCVA at 20/400 (OD) and 20/800 (OS) despite continued enzyme therapy.

Whereas the authors' hypothesis that specific 'lipid-clearing' therapy should improve the condition if it is Fabry-related, we question whether uveitis in Fabry disease, as in Gaucher disease, is indeed related. In that this is a first report of concordance of uveitis and Fabry disease, heightened awareness of unusual ocular manifestations in Fabry disease is warranted. Nonetheless, despite our lack of optimism *vis a vis* uveitis, we hope that the patient will continue to benefit from enzyme therapy for all the classic disease parameters of Fabry disease, particularly cardiac manifestations[4] which were not mentioned in the report.

References

1 Shen YD, Yang CM, Huang JS. Fabry disease manifesting as chronic uveitis-treated with enzyme replacement therapy. *Eye* 2006, 14 July [E-pub ahead of print].
2 Dweck A, Wollstein G, Elstein D, Zimran A. Uveitis in Gaucher disease: a case report. *Am J Ophthalmol* 2005; **140**: 146–147.
3 Dweck A, Zimran A, Elstein D. Reply. *Am J Ophthalmol* 2006; **141**: 421–422.
4 Altarescu G, Elstein D. Cardiac abnormalities in Fabry disease: natural history in hemizygote males suggests that cardiac pathology is universally present. *Haema* 2004; **8**: 103–108.

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Sir,

Reply to Dweck *et al*

We would like to comment on the letter from Dweck *et al*. First, the presentations in this Fabry case was not typical of a true uveitis because it lacked some common features of a uveitis, such as injection of ocular surface, kerato precipitates of posterior corneal surface, or posterior synechia of iris, despite the anterior chamber cells were very prominent. We believe the change of vascular permeability contributed to the prominent cells in both anterior and posterior chamber and thus consider this case a uveitis-like picture rather than a true uveitis. Second, we agree with the speculation that the occurrence of uveitis in the Fabry case was a coincidence. However, because the clinical condition markedly improved after enzyme replacement therapy, this coincidence was not very likely.

Based on the experience of many vitreoretinal doctors on the use of corticosteroids in diabetic macular oedema,[1,2] we believe the use of steroid in this case was similar to that in diabetic retinopathy: a vascular leakage-decreasing effect rather than a true anti-inflammatory effect. This explained why the effect of steroid was temporary and the enzyme replacement therapy had more long-term effect.

We believe the combined therapy of steroid and enzyme replacement therapy would be a reasonable approach to cases with Fabry disease or Gaucher disease.

References

1 Bakri SJ, Kaiser PK. Posterior subtenon triamcinolone acetonide for refractory diabetic macular edema. *Am J Ophthalmol* 2005; **139**(2): 290–294.
2 Karacorlu M, Ozdemir H, Karacorlu S, Alacali N, Mudun B, Burumcek E. Intravitreal triamcinolone as a primary therapy in diabetic macular oedema. *Eye* 2005; **19**(4): 382–386.