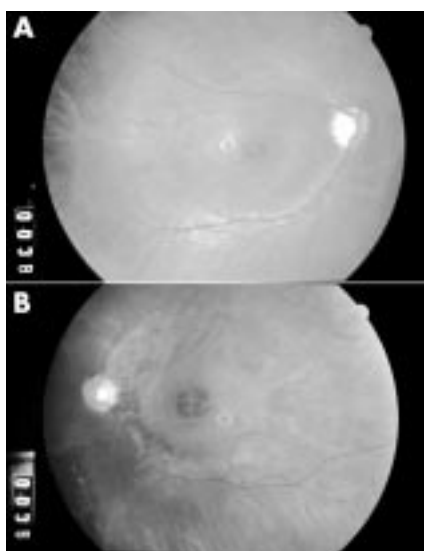


## **Bilateral macular staphylomas in a patient with cone dystrophy**

A posterior staphyloma is characterised by scleral ectasia and is pathognomonic for pathological myopia.<sup>1 2</sup> Posterior staphylomas are classified into five types based on the anatomic location.<sup>1</sup> Type 1 staphylomas extend from the nasal border of the optic nerve into the macular region and are the most frequent staphyloma seen in myopes.<sup>1</sup> Type 2 staphylomas are centred on the macula while type 3 staphylomas are centred on the optic



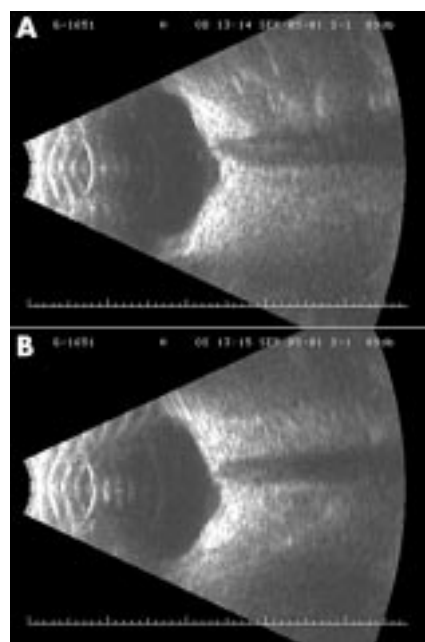
**Figure 1** (A) and (B) Bilateral macular staphylomas in a patient with cone dystrophy. There are macular retinal pigment epithelial changes consistent with cone dystrophy. The retinal vessels in both eyes appear to dive posteriorly into staphylomas that are centred around the macula (type 2 staphyloma).

disc without macular extension.<sup>1</sup> Type 4 staphylomas are located nasal to the optic disc and type 5 staphylomas develop inferior to the optic disc.<sup>1</sup> Staphylomas can be associated with multiple complications including retinal pigment epithelial atrophy, lacquer cracks, retinal and subretinal haemorrhages, and choroidal neovascularisation.<sup>1,3</sup>

This report describes a patient with undiagnosed, bilateral type 2 macular staphylomas compounded by cone dystrophy. To the best of our knowledge, this is a novel association not reported in the literature and with potential therapeutic implications.

### Case report

A 32 year old white woman presented to the Wilmer Ophthalmological Institute, Baltimore, MD, for a second opinion. She reported having progressively worsening vision since childhood and was diagnosed with cone-rod dystrophy at age 18 by an outside ophthalmologist. She experienced photophobia both indoors and outdoors. She denied recent changes in her vision. Past ocular history was



**Figure 3** (A) Horizontal B-scan ultrasound of the right eye. The depth and width of the macular staphyloma is 1.5 mm and 4.6 mm respectively. (B) Horizontal B-scan ultrasound of the left eye. The depth and width of the macular staphyloma is 1.0 mm and 4.2 mm respectively.

otherwise significant for a remote history of corneal abrasion in the right eye. Past medical history and family history were non-contributory.

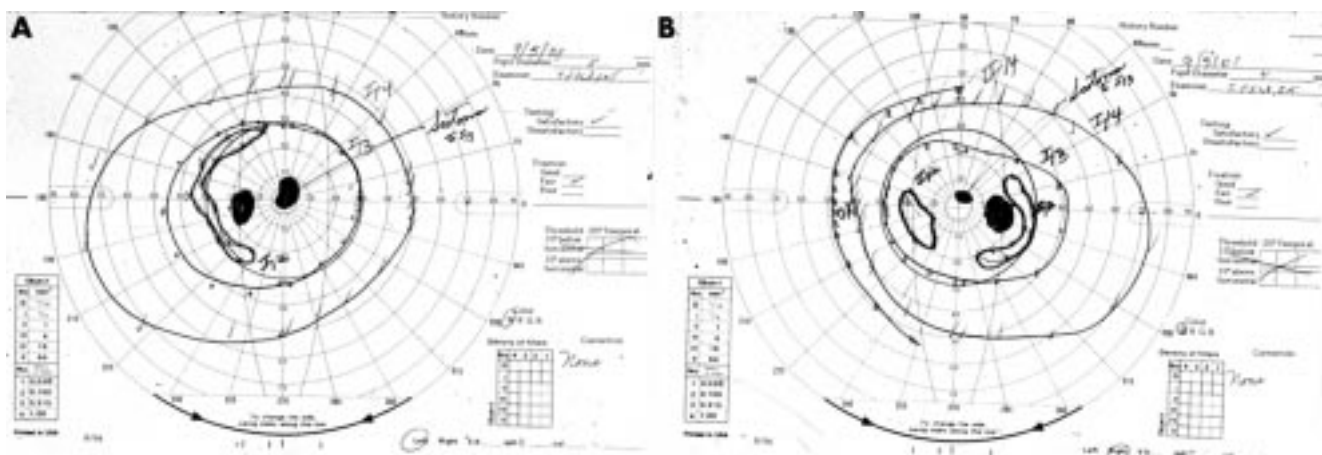
On ophthalmological examination, her uncorrected visual acuity was 20/200-2 in the right eye and 20/200-1 in the left eye. Retinoscopic reflex and refraction were variable and significant for mild myopia. Refraction did not improve her vision. There was no relative afferent pupillary defect and extraocular movements were normal. There was no evidence of nystagmus. Slit lamp biomicroscopy of the anterior segment was unremarkable. Dilated fundus examination showed a tilted optic nerve head in each eye. There were bilateral macular retinal pigment epithelial changes consisting of a ring of hypopigmentation surrounding an area of mildly increased pigmentation centrally (Fig 1). The retinal vessels in each eye appeared to dive into posterior staphylomas (Fig 1). The

staphylomas were centred around the macula in each eye. The peripheral retina in each eye was otherwise normal.

Fluorescein angiography demonstrated mottled hyperfluorescence without leakage corresponding to the retinal pigment epithelium (RPE) changes (data not shown). Goldmann visual fields were remarkable for central scotomas in both eyes with peripheral isoptres full to II-4 stimulus in the right eye and I-4 stimulus in the left eye (Fig 2). A B-scan showed bilateral staphylomas with macular involvement (Fig 3). On electroretinography, photopic responses were markedly reduced. The dim scotopic responses were normal. The mixed scotopic responses were 90% of normal in the right eye and 97% of normal in the left eye. There were markedly reduced photopic flash and flicker responses, with a questionable response of 10% of the normal amplitude. Pelli-Robson contrast sensitivity testing was depressed at 1.2 log units in a dim environment (normal = 1.65). D15 colour testing detected four major and three minor errors in the right eye, and five major and two minor errors in the left eye. A therapeutic red tinted contact lens was prescribed to eliminate the photophobia and aversion to light due to cone dystrophy, and thereby to reduce the level of visual dysfunction.<sup>4,5</sup> After 1 month of wear, the patient reported being a lot more comfortable in bright surroundings. She did not have to squint as much as before using these lenses, was able to sustain prolonged eye contact with other individuals, had improved face recognition and demonstrated improved posture. Visual acuity was 20/125 in each eye tested separately and 20/80-2 when both eyes were tested together.

### Comment

In summary, we have described a patient whose findings are consistent with a diagnosis of cone dystrophy compounded by bilateral macular staphylomas. We believe that this does not represent congenital achromatopsia given the absence of nystagmus and the history of progressively worsening vision. Although there is a report of familial cone dystrophy with bilateral macular colobomata,<sup>6</sup> we are unaware of a case of bilateral macular staphylomas associated with cone dystrophy. To our knowledge, this case represents a previously unreported association of cone dystrophy with macular staphylomas. Awareness of this association will hopefully contribute to proper diagnosis as this finding had presumably been missed in previous ophthalmological examinations.



**Figure 2** (A) and (B) Goldmann visual fields in both eyes demonstrate central scotomas with peripheral isoptres full to II-4 stimulus in the right eye (bottom right) and I-4 stimulus in the left eye (bottom left).

Given the significant association of macular staphylomas with numerous complications listed above, especially the risk for choroidal neovascularisation and haemorrhage, such patients should receive counselling regarding its symptoms and receive periodic comprehensive ophthalmological examinations.

Financial interests: None.

Financial support: None.

**R S Apte, J S Sunness**

The Retinal Vascular Center, The Wilmer Ophthalmological Institute, The Johns Hopkins University School of Medicine, Baltimore, MD, USA

**R S Apte, B G Goldstein**

The Vitreoretinal Division

**W L Park, J S Sunness**

Lions Vision Center

**R Z Raden, M J Elman**

Elman Retina Group, PA, Baltimore, MD, USA

Correspondence to: Janet S Sunness, MD, The Wilmer Ophthalmological Institute, The Johns Hopkins University School of Medicine, 550 N Broadway, 6th Floor, Baltimore, MD 21205, USA; [jsunness@jhmi.edu](mailto:jsunness@jhmi.edu)

Accepted for publication 2 January 2003

## References

- 1 **Quaranta M**, Brindeau C, Coscas G, *et al*. Multiple choroidal neovascularizations at the border of a myopic posterior macular staphyloma. *Graefes Arch Clin Exp Ophthalmol* 2000;**238**:101–3.
- 2 **Curtin BJ**, Carlin DB. Axial length measurements and fundus changes in the myopic eye. *Am J Ophthalmol* 1971;**71**:42–53.
- 3 **Steidl SM**, Pruett RC. Macular complications associated with posterior staphyloma. *Am J Ophthalmol* 1997;**123**:181–7.
- 4 **Young RSL**, Krefman RA, Fishman GA. Visual improvements with red-tinted glasses in a patient with cone dystrophy. *Arch Ophthalmol* 1982;**100**:268–71.
- 5 **Zisman F**, Harris MG. Therapeutically tinted contact lenses. In: Harris MG, London R, eds. *Contact lenses: treatment options for ocular disease*. St Louis: Mosby, 1996:105–22.
- 6 **Miller SA**, Bresnick G. Familial bilateral macular colobomata. *Br J Ophthalmol* 1978;**62**:261–4.