PostScript

LETTERS

Keratoconus associated with CSNB1

A 35-year-old man was referred with progressive impairment of vision. He was previously diagnosed with anisometropic amblyopia and registered partially sighted. He had lifelong problems of night vision and described a family pedigree suggestive of X-linked inheritance, with two generations of male members affected with nyctalopia and high myopia. No male-to-male transmission occurred and female relatives were unaffected. Family members could not be contacted.

Refraction showed distorted retinoscopy reflexes, with best corrected visual acuities of $-18.00/+10.00 \times 180$ (6/60) OD and $-10.00/+8.00 \times 5$ (6/36) OS. Bilateral steep central corneal thinning, paracentral ectasia and Vogt's striae were present. His fundus appeared normal. Corneal topography disclosed 7.4 dioptres of irregular astigmatism in the central 3 mm with thinning ($335 \mu\text{m}$) characteristic of keratoconus (fig 1). There were no medical or environmental influences for his keratoconus. Electroretinography (ERG) showed no

response to the dim flash in the dark, a "negative" ERG to the standard flash and attenuated tests in photopic conditions with near absence of B-wave (fig 2).

Comment

Keratoconus has not been previously linked with CSNB1 either as a chance or true association though both show genetic predisposition. Keratoconus is characterised by progressive irregular myopic astigmatism, central corneal thinning and ectasia¹ and associated with retinal diseases including retinitis pigmentosa, macular coloboma, Leber's congenital amaurosis, retrolental fibroplasia and cone dystrophy.² The genetic bases for keratoconus is poorly defined³ although one form is associated with mutations in the retinal transcription factor VSX1 and abnormal retinal function.^{3,4}

X-linked CSNB is characterised by impaired night vision, myopia, nystagmus and reduced visual acuity.⁵ It can be subdivided into complete (CSNB1) and incomplete (CSNB2) phenotypes distinguished by ERG responses.⁶ In CSNB1 there is complete absence of response to a dim flash in the dark, a "negative" response to the standard flash and an altered response to the light adapted standard flash. An isolated gene in

Xp11.4 (NYX, nyctalopin) is mutated in patients with CSNB1,⁷⁻⁹ which may influence the assembly of collagen fibrils.¹⁰

Although the occurrence of keratoconus and CSNB in the above patient may represent a chance association, further reports may disclose a possible pathogenetic link. It would therefore be of interest to investigate the corneal topography of patients with CSNB1 to determine if there is a possible association.

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Figure 1 Corneal topography of the right eye showing a steep central and inferior thin cornea characteristic of keratoconus.

Figure 2 Top row shows a representative normal trace with L.ocus dexter and L.ocus sinister responses of subject in rows 2 and 3.

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Combination treatment of intravenous immunoglobulin and cultivated oral mucosal epithelial transplantation for ocular cicatricial pemphigoid

Ocular cicatricial pemphigoid (OCP) is a progressive cicatrizing autoimmune disease affecting the skin and mucous membranes, including ocular surface epithelia.¹ Although systemic immunosuppression is advocated, both progression of ocular surface inflammation and systemic side effects remain a problem.² Intravenous administration of immunoglobulin (IVIg) is an immunomodulating treatment, and encouraging results have been reported for acute-phase OCP.³ However, corneal scarring and neovascularisation have also been reported. Cultivated oral mucosal epithelial transplantation (COMET) is a recently developed surgical method for ocular surface transplantation.⁴ Autologous oral epithelial cells were cultivated on preserved amniotic membranes, and the sheets were transplanted after fibrotic tissue excision. As autologous cells are used, immunological rejection is avoided, thereby eliminating complications associated with immunosuppression.⁵ In this report, we describe a patient with OCP successfully treated by a combination of IVIg and COMET.

Case report

A 55-year-old man with bilateral conjunctival inflammation, photophobia and bulla formation in oral mucosa was first seen in May 2003 with a 2-month history of systemic corticosteroids without improvements. The best-corrected visual acuity (BCVA) was hand motion in his right eye and 20/1000 in his left. Slit-lamp examination disclosed subconjunctival fibrosis, partial conjunctivalisation, foreshortening of fornices, and large corneal and conjunctival epithelial defects in both eyes (fig 1A). Immunohistology of the conjunctival and oral mucosal biopsy samples showed immunoglobulin (Ig)G deposition at the basement membrane zone, confirming the diagnosis of OCP.

Intensive treatments including ciclosporin, 1% methylprednisolone, preservative-free 0.3% hyaluronic acid, levofloxacin for topical treatments and prednisolone starting from 60 mg/day and cyclophosphamide 100 mg/day for systemic treatment were started. Amniotic membrane transplantation was also performed. Although some improvements were appreciated, recurrent epithelial defects and progress of fibrosis were noted. We started IVIg with the protocol of receiving immunoglobulin 20 g/day for 5 days, repeated as one cycle every alternate week, then tapered; after the 9th cycle, the applied dose was decreased gradually.³

Soon after starting of IVIg, the photophobia and pain improved. Repeated ulceration in the skin and oral mucosa, and nasal bleeding were also resolved. However, symblepharon and conjunctivalisation remained, the Schirmer