hyaloid failing to regress. The Norrie disease gene (*NDP*) maps to Xp11.3⁶ and has two coding exons. Multiple pathogenic mutations have been reported with no strong genotype-phenotype correlation, although there is evidence that mutations at the 3' end of the gene lead to a milder phenotype.⁷ In addition, intrafamilial variability of the ocular phenotype has been described.⁸ The sequence change in the Norrie gene detected in this family led to a non-conservative amino acid change (R41S), but it was not certain that this variant would have a functional effect since it had not been reported previously. Family studies have, however, demonstrated that it is a de novo change, probably arising in the grandparental germline, and is therefore likely to be pathogenic. The proband's mother was 24 weeks' pregnant and the study suggested a low recurrence risk for isolated PFV, but a one in two chance that future sons would have Norrie disease, and a one in two chance that any future daughters would be carriers. Prenatal testing was declined, but this could be offered in future pregnancies. The proband's maternal aunt can be offered carrier testing because although the mutation most likely arose de novo in the proband's mother, the possibility that one of the proband's maternal grandparents might carry the mutation in a mosaic form cannot be excluded.

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Isolated lichen planus of the conjunctiva

Lichen planus is an autoimmune condition of unknown aetiology affecting the skin and mucous membranes. Classic lesions appear as recurrent, small, discrete, polygonal, flat topped, violaceous, pruritic papules distributed symmetrically on the flexor surfaces of the extremities and trunk. These lesions are often accompanied by lesions of the oral, genital, and rarely, ocular mucosa. Conjunctival lichen planus can lead to irreversible damage to the ocular surface and loss of vision from corneal scarring.^{1,2} We report two patients with isolated lichen planus of the conjunctiva. The referring ophthalmologists had attributed these ocular findings to a severe dry eye state. The correct diagnosis was made by conjunctival biopsy. Treatment with topical ciclosporin A resulted in suppression of the inflammation and arrest of the progressive cicatrization.

Case 1

A 57 year old white woman with a diagnosis of secondary Sjögren's syndrome was referred for further management of severe keratoconjunctivitis sicca, which had been refractory to treatment with multiple topical medications, including topical steroids, as well as punctal plugs. The patient had a history of rheumatoid arthritis with crippling deformities in her hands.

On examination, slit lamp biomicroscopy revealed inadequate tear film with debris, severe scar formation of the upper tarsal conjunctiva, moderate bulbar conjunctival hyperaemia, inferior fornix foreshortening, and severe diffuse punctate epithelial keratitis, bilaterally (fig 1). A conjunctival biopsy specimen was harvested from the inferior fornix of the left eye for evaluation. Direct immunofluorescence staining of the specimen revealed a heavy linear fibrinogen deposition along the epithelial basement membrane zone (fig 2). No IgG, IgA, IgM, or C3 deposition was present. A systemic examination of the patient revealed no lesions on the skin, oral, or genital mucosa. A trial of pulsed topical prednisolone acetate 1% was initiated but was quickly tapered and discontinued after a rise in the intraocular pressure. While tapering the topical steroids, the patient was started on ciclosporin A 0.05% eyedrops (Restasis, Allergan Inc) four times daily. The ciclosporin dose was increased to six times daily after the discontinuation of the topical steroids, and within 1 month the inflammation was fully controlled. The ciclosporin was gradually tapered to a maintenance dose of twice daily, on which the patient's cicatrizing conjunctivitis has remained quiescent for 20 months, with no evidence of skin or other mucosal involvement.

Case 2

An 86 year old white man was referred for further management of keratoconjunctivitis

sicca and blepharitis attributed to acne rosacea. The patient's condition had been refractory to treatment with oral tetracyclines combined with topical steroids. His ocular history was remarkable for postoperative endophthalmitis in the left eye following combined cataract extraction and trabeculectomy, necessitating a subsequent penetrating keratoplasty with secondary intraocular lens implantation. The graft failed within a few months of transplantation.

On examination, slit lamp biomicroscopy demonstrated eyelid thickening with irregular margins, marked telangiectasis, and significant meibomian gland dysfunction in both eyes. The bulbar conjunctivae were hyperaemic. Symblepharon formation and foreshortening of lower forniceal conjunctivae were present bilaterally. The corneal epithelium on the right was irregular, with subepithelial and superficial stromal haze. The left eye had a failed corneal graft with 360 degrees of neovascularisation. A conjunctival specimen was harvested from the inferior fornix of the left eye. Direct immunofluorescence staining revealed shaggy linear deposition of fibrin along the epithelial basement membrane zone. No immunoglobulin or complement deposition was present. A detailed medical examination of the patient revealed no lesions on the skin, oral or genital mucosa. A trial of topical ciclosporin A 0.05% drops (Restasis, Allergan Inc) four times daily, combined with prednisolone acetate 1% four times daily, brought the symptoms under control. During the 18 months of follow up, both medications have been successfully tapered, and there has been no progressive cicatrization.

Figure 1 Patient 1. Significant foreshortening and subepithelial scar formation of the right lower forniceal conjunctiva.

Figure 2 Direct immunofluorescence staining of the lower bulbar conjunctival biopsy specimen from the same patient, demonstrating a heavy, linear deposition of fibrinogen along the basement membrane.

Comment

Lichen planus is a very common skin disease and the overwhelming majority of the patients have oral lesions as well. However, lichen planus isolated to the conjunctiva is very rare. The cause of lichen planus is unknown. Available evidence points toward a T cell mediated immunological response to an induced antigenic change in the basal membrane zone of mucosa or skin.³ Conjunctival involvement, albeit rare, may be severe, causing cicatrix formation that is clinically indistinguishable from other forms of cicatrizing conjunctivitis.^{1,2} A conjunctival biopsy is the key to the correct diagnosis of the underlying aetiology. An irregular, coarse band of fibrin or fibrinogen deposition in the basement membrane, in the absence of immunoreactants demonstrated with direct immunofluorescein staining, strongly supports a diagnosis of lichen planus.²

Multiple therapeutic options including local and systemic corticosteroids, psoralens with ultraviolet A light, retinoids, ciclosporin, and cyclophosphamide have been recommended.⁴ Ciclosporin A is an immunomodulator that specifically inhibits T lymphocyte proliferation via inhibition of interleukin 2 receptor expression.⁵ Several clinical studies demonstrated favourable results with topical ciclosporin in the treatment of ulcerative or erosive oral lichen planus. Successful treatment of patients with isolated conjunctival lichen planus using topical ciclosporin A 2% in olive oil has also been reported.^{1,2}

We recommend that lichen planus be routinely included in the differential diagnosis of cicatrizing conjunctivitis, as this has important therapeutic and prognostic implications. Topical ciclosporin A may be of benefit in the treatment of selected patients with isolated conjunctival lichen planus.

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Bilateral peripapillary staphyloma, a clinicoradiological report

Peripapillary staphyloma is a rare non-hereditary congenital anomaly characterised by a relatively normal appearing optic nerve head located at the base of a staphylomatous excavation.^{1–3} It is generally not associated with other congenital defects or systemic diseases.¹

Peripapillary staphyloma is generally unilateral with bilateral cases being extremely rare.^{4–6} To the best of our knowledge, there is no previous report of neuroimaging findings in peripapillary staphyloma. We report a case of bilateral peripapillary staphyloma with the magnetic resonance imaging (MRI) findings.

Case report

An 11 month old boy was referred to the neuro-ophthalmology clinic by an ophthalmologist because of a suspected optic nerve mass noted on MRI. The patient was born to non-consanguineous parents at 39 weeks of

gestational age with a birth weight of 2800 g. The neonatal period was unremarkable. Development, other than ocular findings was normal. The boy was the first child of his parents and there was no history of significant ocular disease in his family. His parents' ocular examinations were normal.

Systemic physical examination including weight, length, and head circumference were normal for his age. Neurological examination was also normal except for vision.

He could only fix with his right eye. The eyes were orthotropic by Hirschberg examination. Cycloplegic refraction was –4.00 RE and –4.00 –2.5×180 LE. Slit lamp evaluation of both eyes was normal. Funduscopy examination (fig 1) revealed a deep posterior excavation with a normal appearing optic nerve head at its base in both eyes. Retinal vasculature was normal in both eyes. There was marked retinal pigment epithelium alteration surrounding the staphyloma especially in the left eye. No retinochoroidal coloboma was present.

MRI of the orbits (fig 2) disclosed a deep excavation of posterior wall of the globe with

Figure 1 Fundus photograph of right (A) and left (B) eyes, showing peripapillary staphyloma with surrounding mottled pigment epithelium and choroids. Normal appearing optic disc is located at the base of staphyloma. Retinal vessels are normal in number and distribution, emerging from the centre of the disc.

Figure 2 T1 weighted axial without contrast (top left), T2 weighted axial (top right), T1 weighted axial with contrast (bottom left), and T1 weighted sagittal view (bottom right) of MRI show a posterior excavation of the globe connected to the vitreous cavity with sharp borders and no enhancement on both eyes, which are compatible with a bilateral peripapillary staphyloma.