SCIENTIFIC CORRESPONDENCE

Report of a family with dominantly inherited upper lid entropion

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Abstract

Aim—To report the occurrence of late onset, bilateral, idiopathic upper lid entropion, occurring in three members of the same family, with a known family history.

Methods—Five family members were examined, and a history taken, at Moorfields Eye Hospital. Three patients were treated surgically, and one also had a tarsconjunctival biopsy.

Results—In all cases, no aetiology was found. The family history suggests an autosomal dominant inheritance pattern. All patients were treated with anterior lamellar repositioning, and had optimal results.

Conclusion—The family reported seems to be affected by a familial form of primary acquired upper lid entropion, that shows an autosomal dominant inheritance pattern.

(Br J Ophthalmol 2000;84:1303-1305)

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Accepted for publication
25 April 2000

Upper lid entropion is generally an acquired condition and usually occurs secondary to a cicatrising process, such as trachoma, chronic blepharoconjunctivitis, Stevens-Johnson syndrome, or trauma.¹ Bilateral upper lid entropion, of late onset and without any apparent cause is, however, rare. By comparison, the congenital forms of upper lid entropion are also rare. They are usually secondary to abnormalities of the tarsus or orbicularis muscle, or due to the mechanical effects of epiblepharon,

microphthalmos, or anophthalmos, which cause variable degrees of upper lid inversion.^{2 3}

All the affected members of the family described in this article became symptomatic during their teenage years or early adulthood, and therefore are best classified as "primary acquired", rather than congenital cases.

To date, all reported cases of primary upper lid entropion have been sporadic. This is the first report of upper lid entropion in which an inheritance pattern is apparent.

Materials and methods

The three members of the family affected (Fig 1: IIc; IIIb; IVc) attended the oculoplastic clinic at Moorfields Eye Hospital, and they were examined and treated by the same surgeon.

Apparently, upper lid problems were well known within this family, and further history revealed that two other members (Ib; IIb) had been affected by "this same lid problem". They, however, had not been seen at Moorfields Eye Hospital, and so further information was unavailable.

The siblings of patient IVc (IVa; IVb) were examined and both were found to be normal.

Results**CASE 1**

A 16 year old boy (IVc) was referred to our clinic during May 1998 complaining of watery and gritty eyes and bilateral photophobia of 1 year's duration.

There was no history of trauma or allergy. Ten months before attending our unit he was diagnosed with an IgA type of nephropathy, which was confirmed on renal biopsy. He was taking no medical treatment and was leading the life of a normal teenage boy.

On examination visual acuities were 6/6 in each eye. There was bilateral upper lid entropion with eyelashes in contact with the globe and no associated trichiasis or distichiasis (Fig 2). There was no horizontal tarsal kink. Signs of chronic tarsconjunctival irritation were present in both eyes including upper lid swelling, blepharospasm, bulbar conjunctival erythema, and a significant tarsal papillary reaction, but no associated scarring. No associated epiblepharon or epicanthus was noticed. Both corneas showed scattered punctate epi-

Figure 1 Pedigree of a family with upper lid entropion.

Figure 2 Bilateral upper lid entropion with eyelashes in contact with the globe but no associated trichiasis or distichiasis.

Figure 3 After 3 months there was no sign of entropion or tarsoconjunctival inflammation.

thelial erosions. Conjunctival cultures showed normal comensal organisms only.

Bilateral anterior lamellar repositioning was performed. Macroscopically the tarsal plate appeared normal. Full thickness tarsoconjunctival biopsy was taken at the time of surgery. The histology showed a mixed chronic inflammatory infiltrate with lymphocytes and plasma cells, but no eosinophils. This is evidence against atopic disease or scarring processes in the aetiology.

Three months postoperatively, the patient remains symptom free, with no sign of entropion or tarsoconjunctival inflammation (Fig 3).

CASE 2

In June 1986, at the age of 28, the mother (IIIb) attended our department complaining of photophobia and watering of 8 years' duration.

On examination she had bilateral upper lid entropion with no distichiasis or cicatricial changes. She had, however, developed a mild bilateral superior limbal pannus. Her treatment during this entire period consisted of self epila-

tion of her eyelashes. There was no history of atopy, or other significant ocular or systemic disease.

Similarly, she underwent bilateral anterior lamellar repositioning and has remained symptom free.

CASE 3

The grandmother (IIc) sought our advice in November 1988, at the age of 70. She had experienced the same symptoms for most of her lifetime, in her right eye. This had worsened over several years before her attendance, concurrent with the development of dermatochalasis. She also gave a history of long term self epilation of the upper lashes.

Examination revealed an asymmetrical degree of upper lid inversion with a significant right upper lid entropion, and mild left upper lid inversion. She only required treatment for the symptomatic right eye, and a right anterior lamellar reposition with a grey line split was performed. She was discharged free of symptoms.

Discussion

Primary acquired upper lid entropion in an otherwise healthy young adult, is rare and must be differentiated from entropion due to congenital upper lid abnormalities. Congenital upper lid entropion is also rare, and it occurs sporadically. Hiles and Wilder⁴ reviewed the 14 cases reported up until 1969 with congenital upper lid entropion. Six of these cases had abnormal tarsal anatomy as the cause of the upper lid entropion, and the aetiology of the other eight remaining cases was unclear. No family history was reported. Since then, other isolated cases of congenital upper lid entropion have been published, but they were related to obvious horizontal tarsal kink.⁵⁻¹⁰ None of these report any inheritance pattern.

The three cases in the family described here (IVa; IIIa; IIb) became symptomatic in their youth or early adulthood, had no history of ocular disease, and the tarsal anatomy and histopathology were normal. They were members of three consecutive generations, which suggests a dominant inheritance pattern.

Congenital upper lid entropion has been associated with cardiovascular, musculoskeletal, and central nervous system abnormalities.^{4,5} In our family, case 1 (IVc) did have an IgA nephropathy, but all other family members were unaffected. It is unknown whether this is related to his upper lid entropion.

In view of the normal tarsal anatomy, and the fact that they all improved with simple anterior lamellar repositioning, it is possible that hypertrophy of the orbicularis muscle has a role in the aetiology of our cases. Usher published a pedigree of spastic congenital entropion of the lower lids, as a dominant characteristic, in 13 members of four consecutive generations.¹¹ Ours is the first known report of a family with upper lid entropion with an apparent inheritance pattern.

All three cases were treated with upper lid anterior lamellar repositioning procedures and to date they all remain asymptomatic, with upper lids in a normal position. This confirms

the efficacy of this procedure in the treatment of upper lid entropion where normal tarsal anatomy is present.

We will watch with interest to see whether our patient's children, or either of his younger siblings (IVa, IVb), develop upper lid abnormalities.

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