Sir,

A case of epiphora associated with Urbach–Wiethe syndrome

Lipoid proteinosis or Urbach–Wiethe syndrome is a rare autosomal recessive disorder associated with hyaline-like deposits in the skin and pharyngolaryngeal mucous membranes causing dysphonia and hoarseness.[1] Ocular manifestations include deposits along the free edges of the eyelids, conjunctiva, cornea trabeculum and Bruch's membrane.[2] We present bilateral punctal stenosis and epiphora in a 6-year-old girl who is the sister of a 9-year-old boy with dry eye syndrome associated with Urbach–Wiethe syndrome reported previously.[3] To the best of our knowledge this presentation has not previously been reported.

Case report

A 6-year-old girl who was known to have hoarseness from infancy was admitted to the ophthalmology department of Hacettepe University for evaluation of bilateral epiphora of 6 months' duration. Despite a negative family history of Urbach–Wiethe syndrome the patient and her brother suffered from this disorder. She was born to first-degree consanguineous parents. Her brother had presented with dry eye syndrome and yellowish pearly papules along the free margin of the eyelids, hoarseness, thickening of the vocal cords, papules in the oral mucosa and hyperkeratosis on the extensor surfaces of the hands and elbows.[3]

The girl also suffered from hoarseness caused by thickening of the vocal cords. White papules on the lips and oral mucosa were present. Ophthalmic examination revealed numerous yellowish papular lesions along the lid margin of the upper and lower lids (Fig. 1). The lacrimal puncta of both eyes were covered by cystic structures. Her best-corrected visual acuity was 6/6 in both eyes. Slit-lamp examination did not reveal any lesions in the conjunctiva and cornea. Fundus examination did not disclose any posterior segment abnormalities. Haematological investigation was normal and biochemical analysis did not yield any high levels of serum proteins and lipids. Blood glucose level was normal.

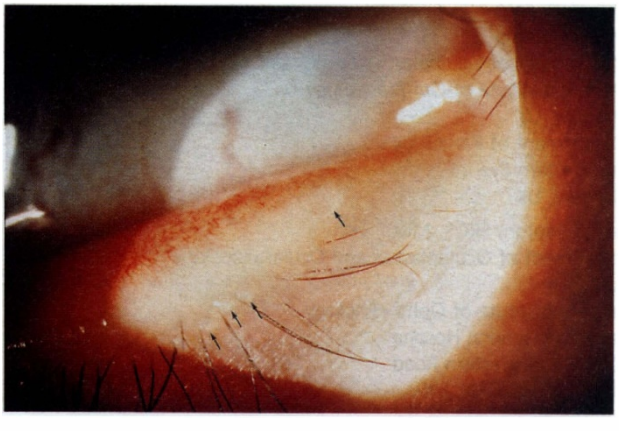

Fig. 1. *Yellow-white papular lesions were present on the lid margins of the lower and upper eyelids and the lacrimal punctum (arrows).*

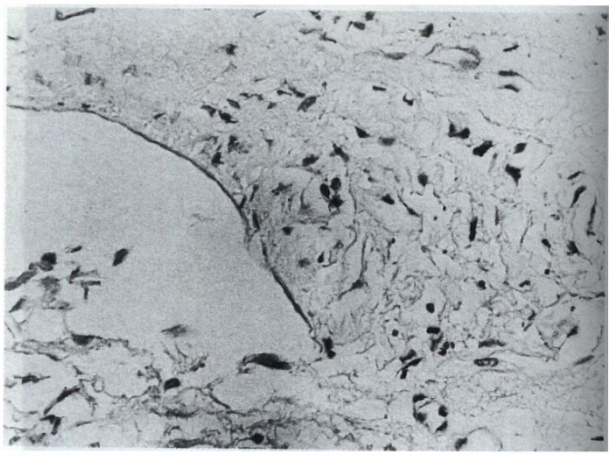

Fig. 2. *Eyelid biopsy revealed a simple cystic structure composed of fibrous tissue. The lining epithelium was not detected (HE, ×40).*

Excision of the cystic structures and bilateral dilatation of the superior and inferior lacrimal puncta were performed under general anaesthesia. Light microscopic examination of an excised cyst revealed a simple cystic structure composed of a fibrous wall with hemosiderin accumulation (Fig. 2). The lining epithelium of the cyst was not observed. The lesion was periodic acid–Schiff negative. The light microscopic findings did not reveal any glycoprotein accumulation in the wall of the cyst.

Comment

The aetiology of Urbach–Wiethe syndrome is unknown. Some authors believe that it is caused by a systemic disorder of increased serum levels of lipids and proteins or an anomaly of carbohydrate metabolism. In contrast, others assume that it is a primary connective tissue disorder.[4,5]

The ophthalmic manifestations of this disease are quite characteristic. The eyelids are frequently involved. Hyaline-like deposits are located along the free margins of the palpebra, the Zeis, Moll and Meibomian glands. In the brother of our case the deposits presumably affected the Meibomian glands and caused a dry eye syndrome. In our case, the presence of cysts on the lacrimal puncta caused epiphora.

The family's pedigree indicated autosomal recessive inheritance to be the most likely mode of transmission of the disease. This is in accordance with the finding of other investigators.[6] Though there was parental consanguinity, the parents were normal and they had affected offspring of which one was a male and the other female.

One of the most intriguing questions in diseases with recessive inheritance is whether one can identify the carrier. We suggest that all the family members be

evaluated carefully if there is a member showing the characteristic clinical findings of the disease, especially in consanguineous marriages.

References

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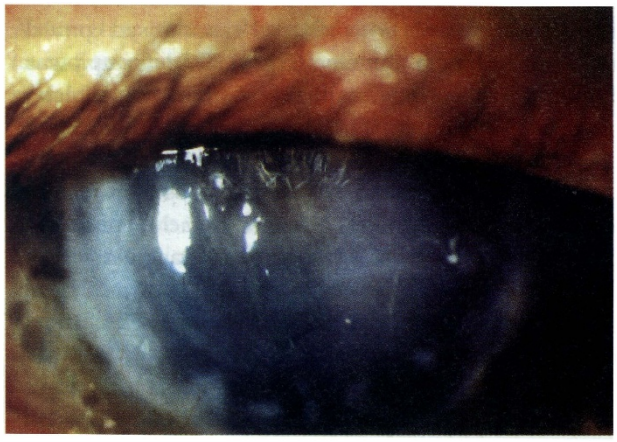
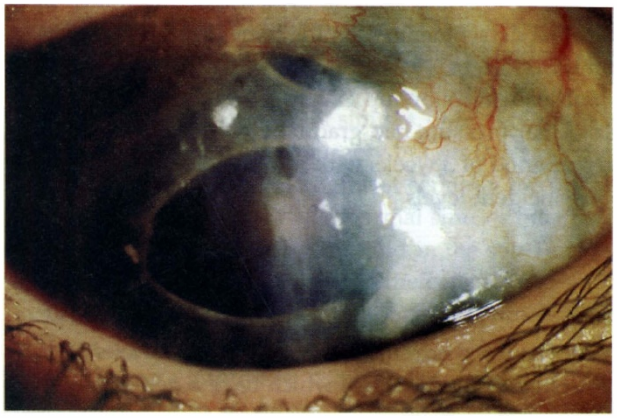


(a)



(b)

Fig. 1. *(a), (b) Anterior segment photographs showing bilateral thin vascularised cornea.*

Sir,

Spontaneous perforation of the globe in Ehlers Danlos syndrome

Ehlers Danlos syndrome is a congenital disease associated with abnormal collagen formation. We present a case of spontaneous rupture of the globe in this condition.

Case report

A 50-year-old Caucasian man with a history of Ehlers Danlos syndrome presented to eye casualty with a 1 day history of blurring of vision in his left eye and noticing that his left eyeball felt very soft while washing his face that morning. There was no history of trauma. His past ocular history included two penetrating keratoplasties in each eye for keratoconus and a right retinal detachment 17 years previously. The last graft performed in the left eye was 5 years ago. His ocular medications included 4% flourometholone eye drops twice a day to both his eyes and saline drops to the left eye.

Eye examination revealed a visual acuity of hand movements close to face in the right eye and counting finger 1/2 m in the left eye. There was marked scleral

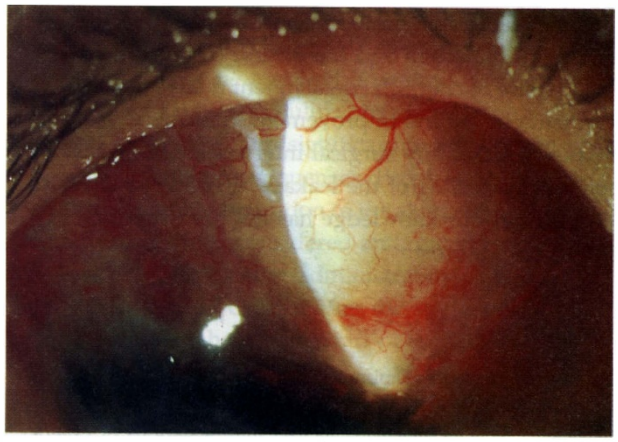


Fig. 2. *Anterior segment photograph showing a superior diffuse conjunctival bleb.*