Sir,

Eye changes in a patient with lamellar ichthyosis and toe pseudoainhum

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Autosomal recessive primary ichthyosis (ARPI) is a rare, genetically heterogeneous skin disease.[1] Several loci of responsible genes have been identified, including 14q11, which controls transglutaminase-1 expression, and 2q33–35.[2–4] Clinical, biochemical, and histological evidence exists separating ARPI into two distinct diseases: lamellar ichthyosis (LI; the severe phenotype) and congenital ichthyosiform erythroderma (CIE, the milder phenotype).[1] The estimated incidence is 1 : 200 000 to 300 000 live births. Affected babies are called 'collodion babies,' because they are embedded in an inelastic membrane that dries soon after birth. Within 2 weeks the collodion membrane spontaneously sheds and the skin develops plate-like scales covering the entire body. In LI the scales are dark, large, polygonal, and tightly adherent and give a typical lifelong disfiguring appearance. Histopathologically, there is marked hyperkeratosis and a prominent granular layer. LI is a retention disorder, with normal epidermal turnover. Pseudoainhum is an affection characterized by the appearance of a constricting band around a digit, which may lead to spontaneous amputation (dactylolysis). This lesion has been observed in vascular, neurological, and skin disorders. Pseudoainhum is very rarely found in patients with LI.[5] We report herein the eye changes in a patient with LI and toe pseudoainhum.

Case report

A 41-year-old man with LI and pseudoainhum of three toes was referred to us for progressive visual loss in both eyes. Skin transglutaminase activity, assayed by measuring the incorporation of [1,4(*n*)-3H]-putrescine into dimethylcasein, was greatly reduced. The patient had used oral retinoids and ophthalmic tear lubricants. Best-corrected visual acuity was 6/12 bilaterally. Eyelid examination showed bilateral lower ectropion, cicatricial lagophthalmos, and severe chronic blepharitis with scales on the eyelashes and meibomian gland dysfunction (Figure 1). Upward eye rotation during forceful closure of the palpebral fissure (Bell's phenomenon) was positive. Slit-lamp examination revealed bilateral conjunctival involvement with thickening, hyperemia, and papillae. The corneal epithelium was normal in both eyes, but a small, round, peripheral, stromal scar was detected at the 7 o'clock position in the left cornea. Nuclear cataract was found bilaterally. Intraocular pressure and fundus examination were normal in both eyes.

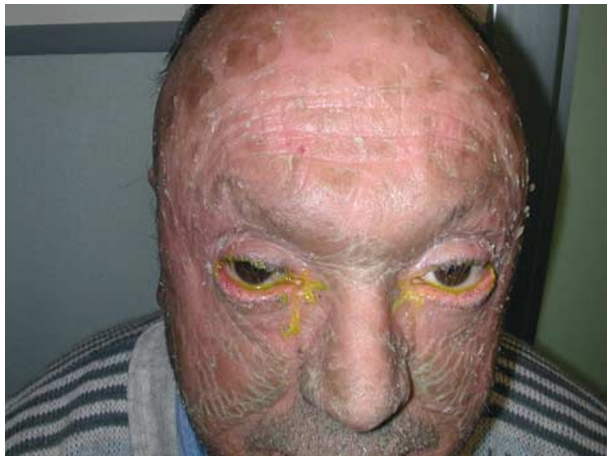

Figure 1 Lower eyelid ectropion, cicatricial lagophthalmos, and severe chronic blepharitis in lamellar ichthyosis with toe pseudoainhum.

Comment

Cicatricial ectropion is the most common eye abnormality of ARPI.[6–8] Ectropion of the lower eyelid was assumed to cause exposure keratopathy in severe cases.[7] Conversely, Cruz *et al*[8] postulated that corneal damage is not directly linked to lower ectropion. These authors suggested that the interaction between eyelid malposition and corneal disease is complex and corneal damage depends on several factors, including lower ectropion, upper eyelid restriction, lack of Bell's phenomenon, and eyelash retraction. Despite moderate lower ectropion, our patient showed no signs of exposure keratopathy. The presence of a significant amount of upper eyelid movement and normal Bell's phenomenon may explain the good condition of the ocular surface in our patient. Symptomless scales on the eyelashes are relatively common in ARPI;[6] however, severe chronic blepharitis has been observed in CIE but not in LI.[8]

To our knowledge, nuclear cataract has never been reported in relatively young patients with LI. In one series, senile lens changes were seen in six (10.3%) patients with ichthyosis vulgaris or recessive X-linked ichthyosis, aged 54 years or older.[6] Our patient had severe LI with pseudoainhum, an extremely rare condition. We know of only one other reported case, but the patient, a 30-year-old woman, had no documented lens changes.[5] The reason why our patient developed early nuclear cataract is unclear. The greatly reduced transglutaminase activity could possibly affect the normal ageing process of the lens fibres. Indeed, a progressive decrease in transglutaminase activity has been observed in the human lens during the progression of nuclear cataract.[9]

In conclusion, because the eyelid abnormalities in LI are lifelong, careful serial ophthalmic examination for corneal exposure is warranted. Patients with LI and pseudoainhum with greatly reduced transglutaminase activity may be at high risk of developing early nuclear cataract.

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Sir,

Viagra or What?

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Viagra, or sildenafil citrate, was an eagerly awaited drug a few years ago almost as though it were a panacea to all impotence-related problems in middle-aged males. However, increasing use of the drug has revealed a spectrum of side -effects, some of these being fairly severe. We present a case of visual loss that took place within 12 h of intake of the drug.

Case report

A 61-year-old taxi driver presented to the eye casualty with a 12-h history of loss of vision in his right eye following consumption of double his normal dose of Viagra (100 mg). He was a high hypermetrope with an amblyopic left eye. He also complained of severe frontal headaches of at least a week's duration, worse in the two days before presentation. His general practitioner commenced him on oral penicillin for suspected sinusitis and upper respiratory tract infection, which in his opinion accounted for his headaches.

He was a chronic smoker, smoking about 30–40 cigarettes a day ever since he was a teenager. Past ophthalmic and medical history was otherwise unremarkable.

Examination revealed Snellen acuity of counting fingers in the right eye and 6/12 in his amblyopic left eye. The anterior segments were normal, but there appeared to be a subtle relative afferent pupillary defect with red desaturation; dilated fundoscopy revealed a very faint focal cloudy swelling of the retinal nerve fibre layer along the inferotemporal retinal artery and its branches as well as one straddling the disc; these later evolved into full-fledged cotton wool spots (Figure 1, left in composite). The nasal disc margin was blurred; however, frank and global optic disc swelling was never seen.

His superficial temporal arteries were palpable and pulsatile, but tender to touch with the presence of scalp tenderness.

The haematology results were normal apart from a Westergren erythrocyte sedimentation rate (ESR) of 30 mm/h and a C-reactive protein (CRP) of 96 mg/l.

It was quite unclear at this stage whether we were actually dealing with a case of branch retinal artery occlusion (BRAO) and/or anterior ischaemic optic neuropathy (AION), arteritic or nonarteritic, associated with Viagra. Since this was the patient's better eye, the consensus of opinion was to treat him with a high dose of oral corticosteroids. Temporal artery biopsy (TAB), performed within 5 days of commencement of steroids, showed no evidence of giant cell arteritis (GCA).

It was encouraging to see his visual acuity improve steadily with resolution of headaches. His fundus appearance had improved with cotton-wool spots present only inferiorly (Figure 1, centre in composite). As his CRP declined steadily, oral prednisolone was tapered.