the syndrome of idiopathic retinal vasculitis, aneurysms, and neuroretinitis (IRVAN)[6] by the multiplicity of aneurysms and other distinguishing retinal features in the latter.

Retinal macroaneurysms are usually easy to diagnose clinically. However, in the context of the optic disc, difficulty may arise if they are associated with the complications of haemorrhage or exudation,[7] since they may easily be confused with other mass lesions of the optic nerve head.[2] They can also be completely asymptomatic, only noted incidentally on routine examination.[3]

Management of optic disc macroaneurysms can be problematic because of the risk of field loss with laser photocoagulation. In the authors' case, a decision was made against any therapeutic intervention other than stopping aspirin, given the absence of any exudative or oedematous changes that would threaten central vision. Furthermore, disc macroaneurysms may involute spontaneously with no sequelae,[3] and aneurysms that haemorrhage into the vitreous have a tendency to allow recovery of full visual function.[1]

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Sir,

New ocular findings in a case of Kabuki syndrome
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Kabuki syndrome was independently described by Niikawa *et al*[1] and Kuroki *et al*[2] in 1981. Patients with this syndrome have typical facial features that resemble the makeup of actors in the Japanese Kabuki theatre. Since its description this syndrome has been identified outside Japan;[3] however, much remains unknown particularly with regard to its inheritance. Previously, these patients were not reviewed by ophthalmologists and this may explain the paucity of reports regarding the ocular findings. We report a case of caruncle lipoma and inferior corneal panus in Kabuki syndrome.

Case report

A boy of 3 months of age presented to the paediatricians with developmental delay and failure to thrive. Peculiar facial features with long palpebral fissures and eversion of the lateral portion of the lower eyelids were noted. A diagnosis of Kabuki syndrome was made by the medical geneticist. Other features present were brachycephaly, low-set deformed ears, micrognathia, and a single palmar crease. Chromosomal studies showed a normal 46XY pattern with no evidence of fragile X syndrome, and molecular and cytogenetic studies revealed no abnormalities. Of note, there was no history of consanguinity or other family history of mental disability or peculiar facies.

He was referred to the ophthalmology department at 2 years of age with epiphoria and marked photophobia. Visual acuities were 6/6 in each eye with a normal orthoptic assessment. There were long palpebral fissures (30 mm bilaterally) with eversion of the lateral parts of the lower lids (Figure 1). Bilateral blepharitis was noticed and the left eye showed a swelling on the caruncle. The patient's mother confirmed that the swelling had been present from birth. Examination under anaesthesia for excision of the caruncular lesion (Figure 2) also showed bilateral lagophthalmos with bilateral inferior corneal pannus. Histopathological examination of the lesion showed it to be a lipoma (Figure 3).

Discussion

The peculiar facial features are highly significant and crucial to the diagnosis of Kabuki syndrome. Five cardinal features in Kabuki syndrome have been described: peculiar faces (100%), skeletal anomalies such as persistent foetal pads, dermatoglyphic abnormalities,

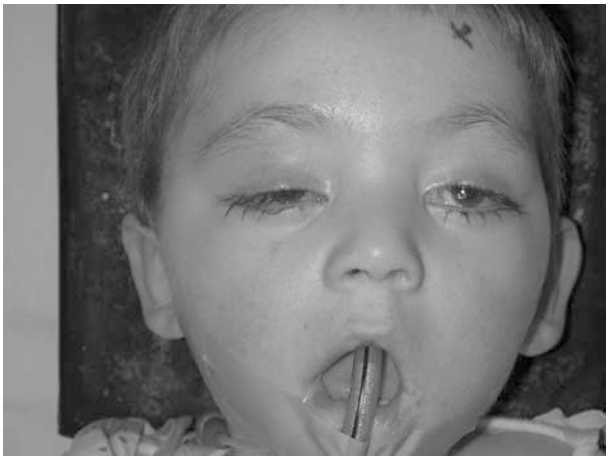

Figure 1 Long palpebral fissures and low-set ears.

Figure 2 Anterior segment appearance showing caruncle lipoma and fluorescein staining of the inferior cornea.

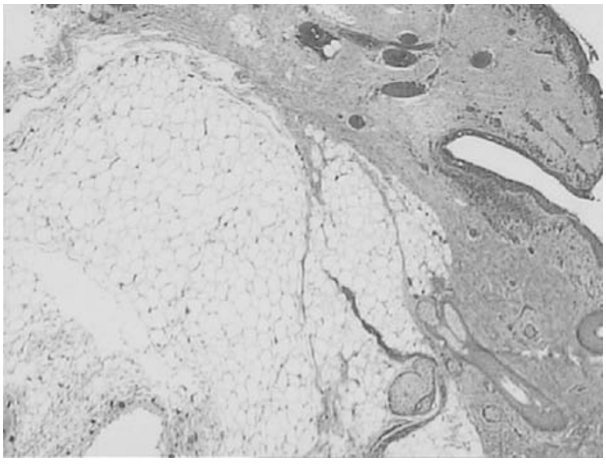

Figure 3 H&E section ($\times 25$) showing the covering inflamed conjunctival epithelium and underlying lipoma.

mild to moderate mental retardation, and postnatal growth deficiency.[3,4]

Kluijt[5] *et al* presented one of the largest papers to quantify ocular manifestations of Kabuki syndrome. They reported six cases with ocular features and reviewed 200 patients from the literature with Kabuki syndrome. Of these, 144 had significant ocular abnormalities in addition to the characteristic external ocular features mentioned above. Amblyopia, refractive errors, strabismus, nystagmus, colobomas, microcornea, corneal opacities, blue sclera, cataracts, and nasolacrimal duct obstruction have all been described. To the best of our knowledge, a caruncular lipoma has not been sited. The corneal pannus may have arisen secondary to lagophthalmos, from blepharitis, or possibly from an anaesthetic cornea. The exact aetiology is not known, as it has not proved possible to test for corneal anaesthesia given his age.

Various theories have been postulated as to the inheritance pattern of this syndrome. Its occurrence is largely sporadic, but there are 10 reports of familial Kabuki syndrome leading to the possibility that in these families Kabuki syndrome could be inherited as a dominant trait.[5] There have been various chromosomal abnormalities detected in children with Kabuki syndrome such as partial 6q monosomy, but none of these have been consistently associated with it.[4]

We report this case to bring attention to the ocular findings associated with Kabuki syndrome and describe two possible new ocular abnormalities. An ophthalmic evaluation of all children with Kabuki syndrome is advised.

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Sir,

A novel temporary treatment remedy for blepharospasm

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Blepharospasm is an involuntary spasmodic bilateral eyelid closure that may be accompanied by dystonia of the facial, oromandibular, and cervical muscles.[1] It is associated in 7% of cases with eyelid apraxia,[1] a nonparalytic motor abnormality characterised by difficulty in initiating the act of elevation in the absence of sensory loss or ataxia.[2,3]

Blepharospasm can be a debilitating condition that may render some patients functionally blind. We present a case of blepharospasm associated with apraxia of eyelid opening and describe a new treatment approach that proved to be an excellent temporising measure resulting in significant improvement in visual function.

Case report

A 45-year-old man was referred to the eye department with a 3-year history of episodic weakness and muscle pain in both eyelids. This had become much worse 3 months prior to presentation. His eyelids would undergo spasmodic closure, but after the spasm subsided, he would still be unable to spontaneously open them (Figure 1a). He could, however, prise them open with his fingers. He also reported that if he held his eyelids open with his fingers, they would remain open for a few minutes during which spontaneous closure was not possible. These symptoms were typically worse in the morning and got worse during the day.

Magnetic resonance imaging of the brain and muscle biopsy was normal, and investigations for myasthenia gravis were negative. A diagnosis of blepharospasm with an apraxia of eyelid opening was made.

Initial treatment with 70 U of Botulinum Toxin type-A (BTA) (Botox) injected into the eyelids (60 U laterally and 10 U medially) had a positive therapeutic effect but lasted only 3 weeks. Another injection of Botox using 80 U was carried out but was ineffective. This was followed by 200 U of Dysport, which still made little difference to his symptoms. All injections were given by the usual subcutaneous approach and no preseptal injections were administered.

At this time, it was felt that any further increase in dosage would only lead to more side effects, as he was a poor responder. In an attempt to improve his eyelid opening, he was referred to the contact lens clinic within the next 3 weeks where he was fitted with scleral contact lenses with ptosis props (Figure 1b). This resulted in marked improvement of his symptoms and he is now able to maintain binocular visual function consistently. Although, he was given another injection of Dysport within the next 3 months of the contact lens fitting, he did not report any unusual benefit following which further BTA therapy was withheld. His improvement was to such a degree that he has resumed his full-time occupation as an engineer. This treatment has been used as a short-term measure while the patient has been referred to a specialist oculoplastic service to be considered for an eyelid myectomy procedure. His follow-up period after the contact lens fitting was 10 months and is still being followed up at Hull Royal Infirmary.

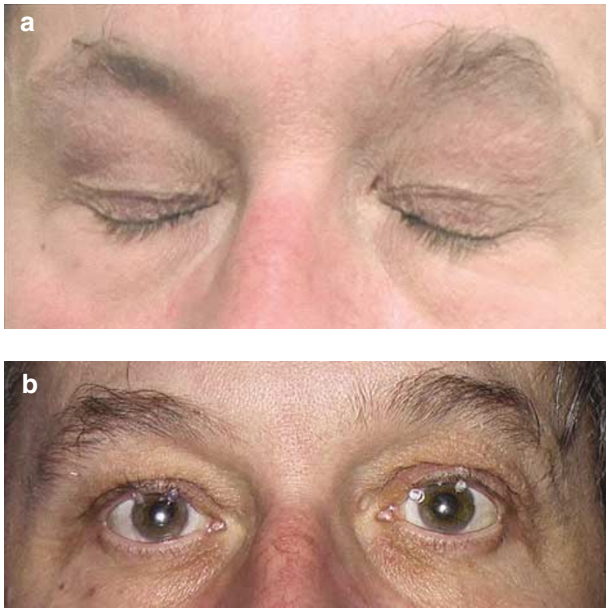

Figure 1 (a) Photograph showing inability to spontaneously open both eyes after an episode of spasmodic closure. (b) Eyelid appearance with scleral lens and ptosis props *in situ*. The pegs on the front surface prevent spontaneous eye closure and facilitate lens handling.