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Sir,

New ocular findings in a case of Kabuki syndrome
Eye (2004) 18, 322–324. doi:10.1038/sj.eye.6700649

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.

mild to moderate mental retardation, and postnatal growth deficiency.^{3,4}

Kluijt⁵ *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



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

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.

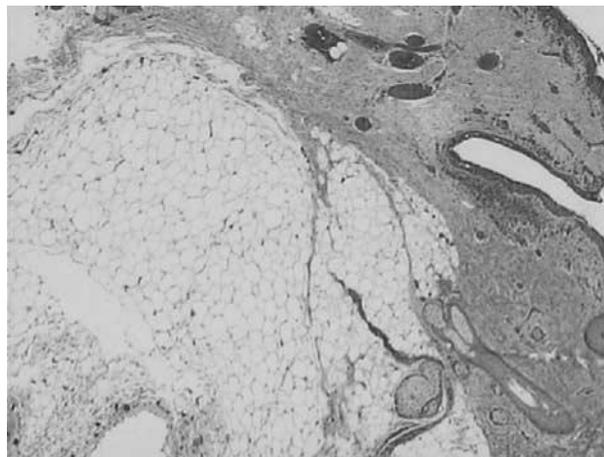


Figure 3 H&E section (x25) showing the covering inflamed conjunctival epithelium and underlying lipoma.

References

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