Sir,

**Arterial macroaneurysm on the optic disc**
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Retinal arterial macroaneurysms are well recognized.[1] Macroaneurysms at the optic disc, however, are unusual, and few have been reported previously.[1–3] The authors report a single arterial disc macroaneurysm associated with a vitreous haemorrhage.

### *Case report*

A 79-year-old gentleman was referred with increasing floaters and gradually reducing vision of the right eye over a 1-year period. He was being treated for hypertension and dyslipidaemia, and was on daily aspirin following a myocardial infarct a year earlier. Other than myopia, there was no other past ophthalmic history of note. Of possible relevance in the family history, however, was that the patient's brother had had a bleed from an intracarnial aneurysm.

At presentation, his visual acuities were 6/24 and 6/12 of the right and left eye, respectively. The intraocular pressure was 15 mmHg in each eye and slit-lamp examination was unremarkable with mild to moderate lens opacities noted in both eyes. Fundus examination revealed a large arterial macroaneurysm occupying almost the entirety of the right optic disc, associated with a small vitreous haemorrhage. No further arterial aneurysms in the right eye were seen, with no evidence of retinal haemorrhage, exudates or thickening. Examination of the left fundus was unremarkable.

Given the presence of a vitreous haemorrhage, daily aspirin was discontinued, resulting in a slow but sustained resolution of his haemorrhage with improved vision. At 11 months following initial presentation, vision in the right eye had improved to 6/9, with repeat funduscopy revealing partial involution of the macroaneurysm. Fluorescein angiography demonstrated thrombosis of the aneurysm (Figure 1). The patient declined MRI scanning to exclude intracranial aneurysm.

### Discussion

Arterial macroaneurysms on the optic disc occur rarely; in one series they comprised only 8% of retinal arterial macroaneurysms examined.[1] Their aetiology is likely to be the same as that of retinal arterial macroaneurysms elsewhere, that is, arteriosclerotic vasculopathy, with increased prevalence of associated hypertension, and central retinal vein occlusion, as well as female sex.[4] Embolic arterial disease may have also been implicated.[5] The condition is easily distinguished from

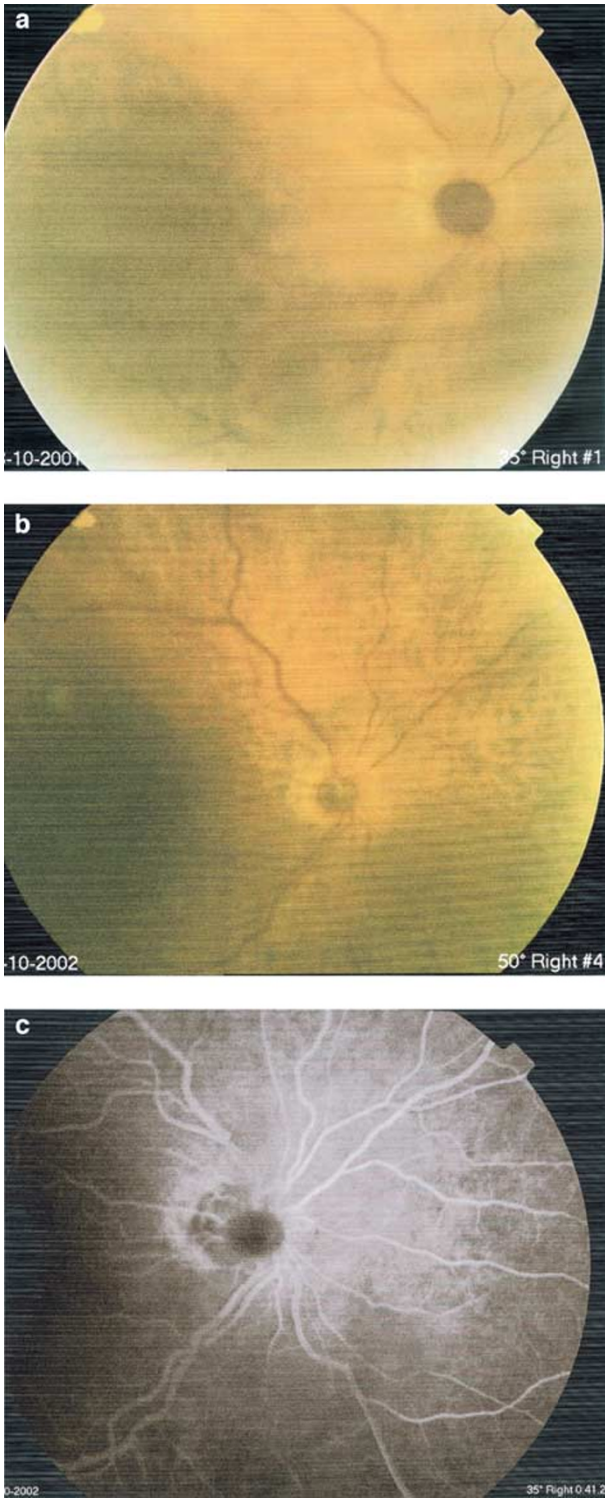


**Figure 1** Macroaneurysm at presentation (a) and at 1 year follow-up (b) (colour), (c) fluorescein angiogram.

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,