[2]Central Clinical School, University of Sydney, Sydney, NSW, Australia
[3]Graduate School of Medicine, University of Wollongong, Wollongong, NSW, Australia
[4]Drummoyne Day Surgery Centre, Drummoyne, Sydney, NSW, Australia
[5]Laverty Pathology, Sydney, NSW, Australia
[6]Department of Oculoplastics, Sydney Eye Hospital, Sydney, NSW, Australia
E-mail: neilsss@hotmail.com

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Sir,

Choroidal new vessels in type 1 myotonic dystrophy-related macular dystrophy respond to anti-VEGF therapy

Mytonic dystrophy type 1 (DM1) is an autosomal dominant disorder caused by mutation in the dystrophia myotonica protein kinase (*DMPK*) gene, located on chromosome 19 (19q13.2–q13.3). Ocular findings reported to date include colored iridescent cataract, strabismus, and limitation of extraocular muscle movement, ptosis, and rarely macular dystrophy.[1]

Case report

A 21-year-old male with DM1 presented with a 4-week history of deteriorating vision in his left eye, assumed to be cataract. Clinical examination, however, showed bilateral pattern dystrophy (Figure 1a) with best corrected visual acuity (VA) recorded at 6/6 in the right eye and 6/15 in the left eye, which had choroidal new vessels (CNVs) (Figures 1b and 2aI). Treatment followed consent of the off-label nature of intravitreal bevacizumab (IVB). After 4 weeks, symptoms had improved but due to persisting intraretinal fluid (Figure 2aII) a second IVB injection was given. After 4 weeks, VA had improved to 6/7.5 with both OCT and angiography confirming no leakage (Figures 2aIII and b). No recurrence was seen at most recent follow-up 14 months later. His father who had DM1 was also examined, but did not have macular dystrophy.

Comment

To our knowledge this is the first report of CNVs complicating DM1. The cause of the macular phenotype is still unknown. The expansion of CTG triplet repeats in DM1 arise within the 3′ untranslated region of the *DMPK* gene,[2,3] but this region also overlaps downstream with the promoter of the homeobox gene *SIX5* on chromosome 19q13.3.[4] As expression of this gene has been localized to the lens, retina, and choroid, it has been

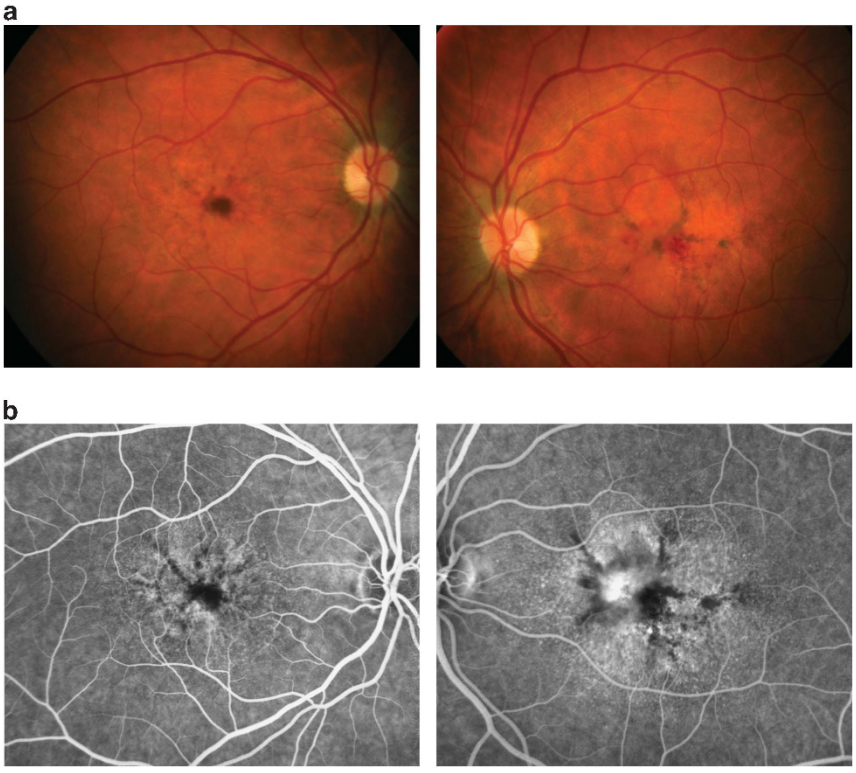

Figure 1 (a) Color images of right and left fundus with pattern retinal pigment epithelium changes. Subretinal hemorrhages are evident in the center of the left macula. (b) Late fundus fluorescein angiogram images before treatment shows RPE changes and, in the left eye, central leakage.

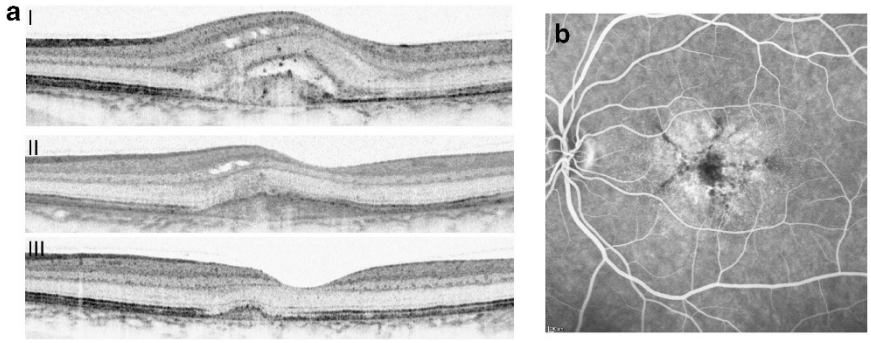

Figure 2 (a) OCT images of the left eye. (I) Left OCT shows central retinal edema, subretinal fluid, loss of foveal contour, and intense sub-RPE hyperreflectivity pigment suggesting occult CNV at initial presentation (I), 4 weeks after the first IVB (II) and 4 weeks after the second IVB injection (III). (b) Late-phase fluorescein angiography confirms complete resolution of central leakage in the left eye.

proposed that SIX5 and not DMPK is responsible for the ocular phenotype of DM.[5] The non-penetrant inheritance of the macular dystrophy, might either indicate a digenic pattern where transcriptional silencing of the *SIX5* gene through expansion of CTG repeats occurs from one generation to the next through anticipation or it might indicate variable expressivity. Beyond the genetics, our observations show that deterioration of VA in patients with DM can be due to macular dystrophy and, rarely, CNVs. The rapid response to IVB provides evidence that VEGF has a role in this disease.

Conflict of interest

The authors declare no conflict of interest.

References

1 Hayasaka S, Kiyosawa M, Katsumata S, Honda M, Takase S, Mizuno K. Ciliary and retinal changes in myotonic dystrophy. *Arch Ophthalmol* 1984; **102**: 88–93.
2 Brook JD, McCurrach ME, Harley HG, Buckler AJ, Church D, Aburatani H *et al*. Molecular basis of myotonic dystrophy: expansion of a trinucleotide (CTG) repeat at the 3′ end of a transcript encoding a protein kinase family member. *Cell* 1992; **68**: 799–808.
3 Mahadevan M, Tsilfidis C, Sabourin L, Shutler G, Amemiya C, Jansen G *et al*. Myotonic dystrophy mutation: an unstable CTG repeat in the 3′ untranslated region of the gene. *Science* 1992; **255**: 1253–1255.
4 Thornton CA, Wymer JP, Simmons Z, McClain C, Moxley RT, 3rd. Expansion of the myotonic dystrophy CTG repeat reduces expression of the flanking DMAHP gene. *Nat Genet* 1997; **16**: 407–409.
5 Winchester CL, Ferrier RK, Sermoni A, Clark BJ, Johnson KJ. Characterization of the expression of DMPK and SIX5 in the human eye and implications for pathogenesis in myotonic dystrophy. *Hum Mol Genet* 1999; **8**: 481–492.

MS Zinkernagel[1], SJ Hornby[1] and RE MacLaren[1,2]

[1]Oxford Eye Hospital and Nuffield Laboratory of Ophthalmology, University of Oxford NIHR Biomedical Research Centre, John Radcliffe Hospital, Oxford, UK
[2]Moorfields Eye Hospital Foundation Trust, London, UK
E-mail: m.zinkernagel@gmail.com

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Sir,

Radiolabelled somatostatin analogue treatment in a case of carcinoid tumour with choroidal metastases

We describe a case of choroidal carcinoid metastases in a young patient and briefly address the considerations and controversy involved in selection of appropriate therapy.

Case report

A systemically well, 22-year-old female presented with blurred vision OS. Visual acuities (VA) were 6/5 OD and 6/6 OS. Anterior segment evaluation was normal OU.

Posterior segment examination confirmed a single, large, amelanotic, choroidal mass OS (6.8 mm ultrasound thickness) (Figure 1a) and two amelanotic choroidal lesions OD measuring 1.8 and 1.2 mm thickness. Systemic work-up revealed a solid mediastinal mass (Figure 2).

Fine needle biopsy of the choroidal lesion OS provided insufficient aspirate for diagnosis. Biopsy of the mediastinal lesion demonstrated typical bronchial carcinoid of low proliferative index (Ki-67 $<$ 2%). Octreotide scintigraphy was performed 1 month following initial assessment. A functioning neuroendocrine tumour (NET) at the site of the right hilar mass (Figure 3a) in addition to a further site of active disease in the left sphenoid bone (Figure 3b) were demonstrated. Management decisions were made in conjunction with oncology and endocrinology services.

Intraretinal fluid accumulation at the left macula caused a reduction in VA to 6/18 OS 8 months following presentation. Neither intravitreal bevacizumab (1.25 mg in 0.05 ml) nor subsequent systemic treatment reduced this fluid.