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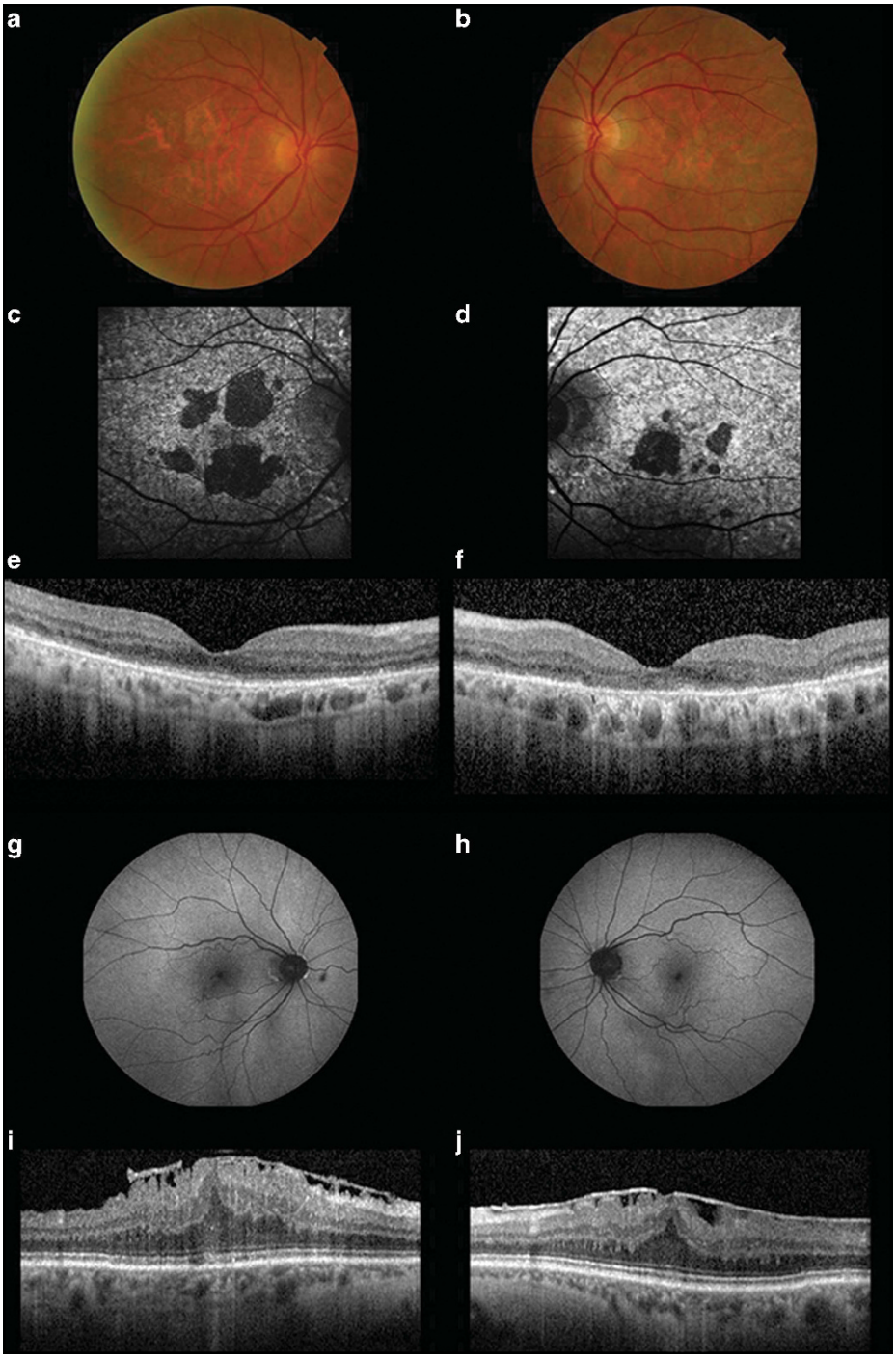
CORRESPONDENCE

Sir,

Macular dystrophy presenting in one of two siblings with limb-girdle muscular dystrophy type 2L due to mutation of *ANO5*

The anoctamin (ANO) family consists of 10 members, many of which have been found to be calcium-activated chloride channels (CaCC).[1–3] Recessive mutations in anoctamin 5 (*ANO5*) result in a proximal limb-girdle muscular dystrophy (LGMD2L).[2,3] *ANO5* is mostly expressed in the skeletal tissue in humans,[2,4] but transcripts have been identified in the retinal pigment epithelium (RPE)/choroid and fetal eye.[4]

The present report describes an association between LGMD2L consequent upon mutation in *ANO5* and macular dystrophy in one affected person.



Case report

A 71-year-old man was referred after persistent left eye distortion after uncomplicated cataract surgery. He was known to be affected by LGMD2L, having presented with proximal muscle weakness in his early 40 s. There was no history of retinotoxic medication. He had a 5-year history of type-2 diabetes and was a moderate smoker (20 cigarettes/day). One older brother out of nine siblings was similarly affected with symptoms and signs consistent with adult LGMD, which presented at the age of 44 years. He had no ophthalmic complaints and was an ex-smoker of 6 years. No other relatives were known to have either muscular dystrophy or retinal disease.

Visual acuities (VAs) were 6/9 right, 6/12 left. Ishihara colour test was 5/17 right, 1/17 left. Imaging revealed

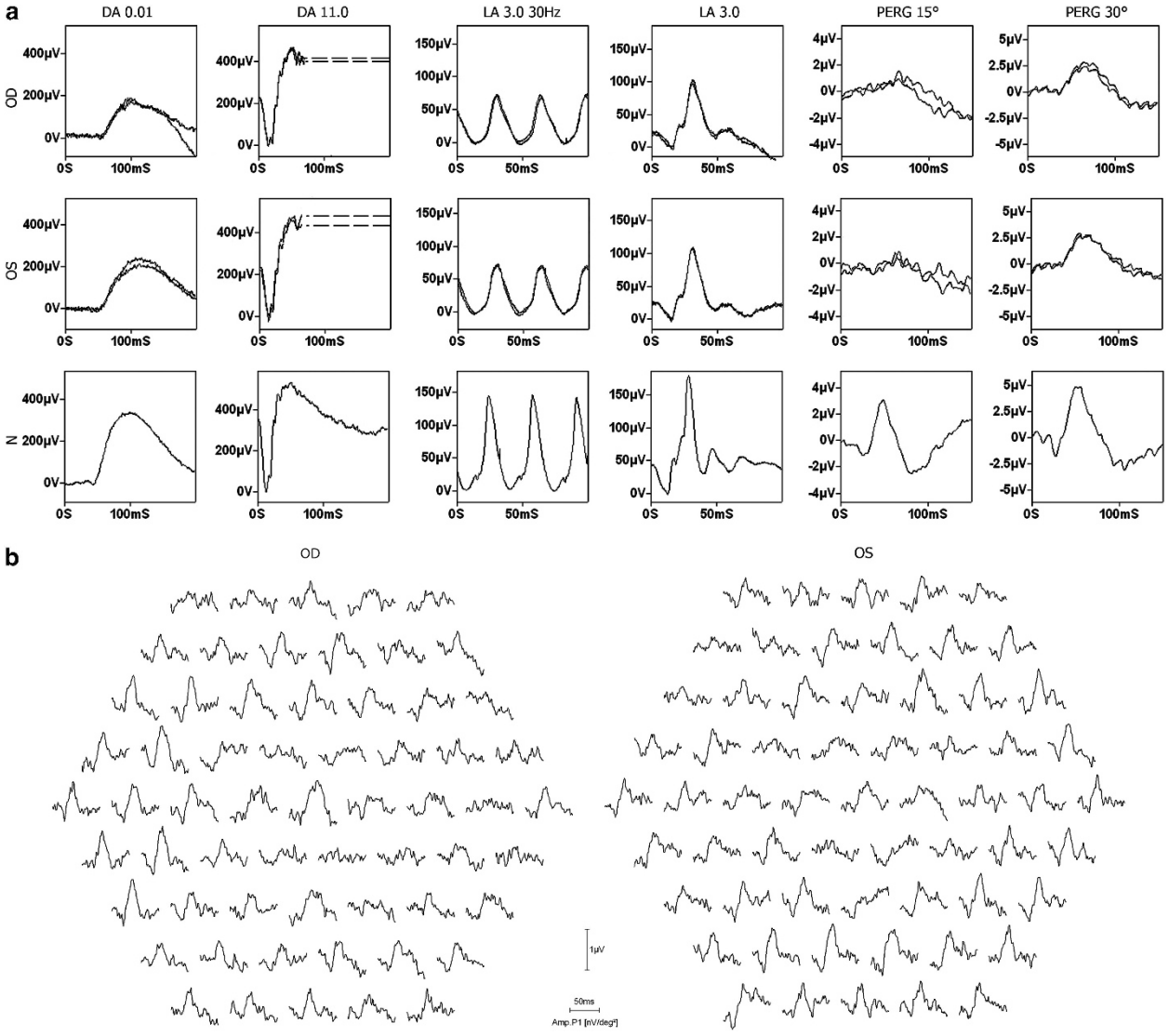


Figure 2 Electrophysiology of affected individual. (a) ISCEV standard full-field ERGs show no significant abnormalities for age. Pattern ERGs (PERGs) are bilaterally subnormal and delayed, but with reasonable amplitude increase with a larger field (30°). (b) Multifocal ERGs (mfERG) show relative preservation of the response to the central foveal hexagon with significant loss of amplitude in immediate parafoveal responses, right eye worse than left (DA, dark adapted; LA, light-adapted; PERG, pattern electroretinogram. 0.01, 11.0. 3.0 are light intensities measured in cd.s.m^{-2}).

Figure 1 Colour fundus photographs of the index case show bilateral macular symmetrical atrophic chorioretinal patches, OD (a) and OS (b) with no apparent flecks or drusen; which are hypoautofluorescent, OD (c) and OS (d) and correspond to areas of retinal atrophy and ellipsoid layer disruption on SD-OCT, OD (e) and OS (f). His sibling's autofluorescence shows no abnormality, OD (g) and OS (h); SD-OCT shows bilateral epiretinal membranes with diffuse retinal thickening, OD (i) and OS (j).

parafoveal atrophic hypoautofluorescent patches in both maculae (Figures 1c and d). Spectral domain optical coherence tomography (SD-OCT) appears in Figures 1e and f. Pattern electroretinograms (ERGs) were abnormal and multifocal ERG revealed parafoveal dysfunction (Figure 2b). Full-field ERGs (Figure 2a) and electrooculography were normal. The *PRPH2*/*RDS* gene showed a normal coding sequence.

Sanger sequencing of exons 1–22 of the *ANO5* gene showed homozygosity for c.191dupA, p.(N64KfsX15) in exon 5, which, if expressed would lead to a peptide missing the carboxyl 850 of its 913 amino acids.
The 78-year-old affected brother had VAs of 6/9 right, 6/6 left, with bilateral epiretinal membranes (Figures 1i and j). Autofluorescence was normal (Figures 1g and h).

Comment

The index case maculopathy is unlike that seen in age-related macular disease, or in known mendelian dystrophies and probably represents a novel late-onset macular dystrophy. *ANO5* is expressed in the human RPE/choroid[4] and although its exact function remains unknown, it is thought to encode a CaCC.[1,2] CaCCs have a physiological role in RPE, important for fluid and ion transport across the RPE.[1,5,6]

To the best of our knowledge this is the first report regarding this association.

Although the possibility of the coincidence of two rare disorders in the proband cannot be excluded, the data are consistent with the occurrence of retinal macular disease in *ANO5*-mediated muscular dystrophy in some, but not all, mutation carriers.

Conflict of interest

The authors declare no conflict of interest.

Acknowledgements

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S Vaz-Pereira[1,2], K Dansingani[1], GE Holder[1] and AR Webster[1,3]

[1]Moorfields Eye Hospital, NHS Foundation Trust, London, UK
[2]Department of Ophthalmology, Hospital de Santa Maria, Lisbon, Portugal
[3]Institute of Ophthalmology, University College London, London, UK
E-mail: Andrew.Webster@ucl.ac.uk

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Sir,
Secondary glaucoma due to chronic scleritis: trabeculectomy in scleromalacia: a case report

Secondary open-angle glaucoma is frequently refractory and difficult to manage.[1]

Glaucoma is uncommon in scleritis, but may develop due to permanent damage to the trabecular meshwork even in quiescent scleritis.[2] Posterior scleritis has been reported to cause angle-closure glaucoma.[3–6] Other mechanisms include secondary steroid-induced glaucoma, peripheral anterior synechiae, and angle neovascularisation.

Scleritis has been reported following surgical trabeculectomy.[7]

Case report

A 68-year-old Caucasian patient had been followed up for 14 years with recurrent bilateral sectoral anterior non-necrotising scleritis in the absence of underlying systemic disease.

The intraocular pressure (IOP) had been controlled with topical g.Timolol 0.5% BD for the previous 5 years. Five months prior to surgery, IOP increased to 30 mm Hg. Despite maximal medical treatment (g.Bimatoprost/Timolol (Ganfort), g.Brinzolamide (Azopt), and Acetazolamide SR 250 mg p.o. BD) her IOP remained at 39 mm Hg. This was associated with a cup-disc ratio of 0.85 and predominantly nasal superior and inferior visual field defects (Figure 1).

This patient was surgically challenging due to the presence of diffuse scleromalacia (Figures 2 and 3