Sir,

Reversal of chronic ocular ischaemia with good visual recovery in giant cell arteritis

Giant cell arteritis (GCA) is a systemic inflammatory vasculitis of unknown aetiology, affecting medium and large calibre arteries. It can result in profound visual disability that usually occurs from occlusion of the posterior ciliary arteries leading to anterior ischaemic optic neuropathy (AION). Visual loss can also result from occlusion of any other arteries supplying the visual pathway. We report a case of gross, chronic, posterior segment ocular ischaemia with excellent visual recovery following treatment with systemic steroids.

Case report

An 83-year-old man presented with a 5-month history of decreased visual acuity (VA) in the right eye.

On examination, Snellen acuities were 6/60 (OD) and 6/6- (OS). There was no anterior segment neovascularisation. The right fundus was pale with multiple cotton wool spots. There were a small number of cotton wool spots on the left fundus. Significant venous tortuosity was absent and there were no haemorrhages in either side.

Fluorescein angiography (FFA) was performed and demonstrated grossly delayed choroidal filling and areas of absent choroidal filling even in the late phase. There were also dilated retinal veins without tortuosity and delayed venous filling (Figure 1 top). Biochemical investigation revealed raised inflammatory markers (c-reactive protein (CRP) of 137 mg/l and plasma viscosity of 2.02 mPA s). On further enquiry, the patient admitted a 6-month history of myalgia, temporal headache, jaw claudication, and weight loss. It was noted that a diagnosis of chronic confusion, sepsis, and cerebral ischaemia had been made by the care of the elderly physicians following a collapse 5 months earlier, and that the CRP was elevated to 103 when the patient was admitted on that occasion. These findings were attributed to a lower respiratory tract infection. On discharge from elderly care, the diagnosis was 'collapse and chronic confusion'; CRP was 74.

A diagnosis of chronic GCA (later biopsy proven) and choroidal ischaemia was made by us and the patient was admitted and treated with intravenous metyhlprednisolone and oral prednisolone. Temporal artery biopsy revealed granulomatous cell infiltration, proliferation of the intima, and disruption of the internal elastic lamina typical of GCA.

Vision improved to 6/9 (OD), 6/6 (OS) within 2 days and inflammatory markers normalised. The patient was discharged to outpatients on oral prednisolone.

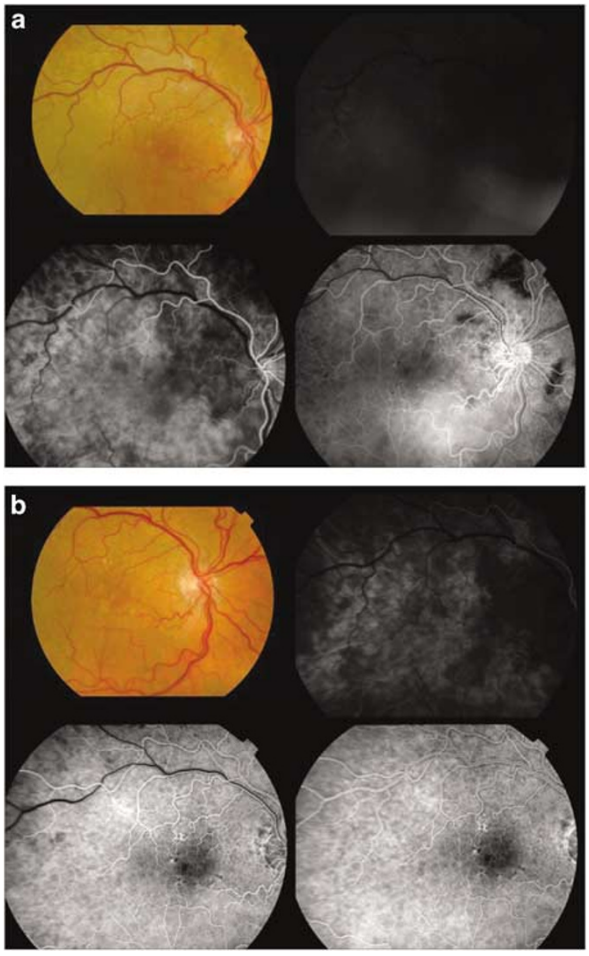

Figure 1 Top: Pretreatment FFA. In clockwise order from top right 32, 36, and 53 s. Bottom: post-treatment FFA. In clockwise order from top right 19, 25, and 34 s.

A post treatment FFA (Figure 1 bottom) was performed and revealed dramatic improvement in perfusion at all levels.

Comment

Choroidal ischaemia is an uncommon consequence of GCA,[1] with an incidence of one in 170 in one case series. [2] The posterior ciliary arteries are most commonly affected. It is notable that in this case AION did not occur despite severe choroidal ischaemia. Choroidal ischaemia is much more common in cases of carotid artery insufficiency due to atherosclerotic disease. Despite carotid endarterectomy and resumption of blood flow in these cases, significant visual recovery is rare.[3]

This case demonstrates that, contrary to popular opinion, GCA-induced ischaemia can be successfully

reversed with treatment. As long as irreversible end-organ damage has not occurred (choroidal ischaemia *vs* choroidal infarction, or more commonly AION), treatment can restore perfusion and end-organ function.

References

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N Saha and SU Rehman

Calderdale Royal Hospital, Salterhebble, Halifax, UK

Correspondence: N Saha, Ophthalmology, Calderdale Royal Hospital, Salterhebble, Halifax, Yorkshire HX3 0PW, UK.
Tel: +01422 222504;
Fax: +01422 222544.
E-mail: docsaha@yahoo.co.uk

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Sir,

Novel mutation in exon 2 of *COL2A1* gene in Japanese family with Stickler Syndrome type I

Stickler Syndrome (STL) is an autosomal dominant disorder characterized by degeneration of the vitreous and retina, and is frequently associated with myopia.[1] It is also accompanied by nonocular signs, such as orofacial anomalies, deafness, and arthritis. There are no widely accepted clinical diagnostic criteria for STL in ophthalmology.[2] Based on locus heterogeneity, a subclassification of STL has been proposed; *COL2A1* mutation associated STL type I with a congenital 'membranous' vitreous anomaly; *COL11A1* mutations associated with STL type II showing a 'beaded' phenotype; and *COL11A2* mutations associated with non-ocular STL type III (OMIM 120140, 120280, and 120290).[3] A subgroup of STL type I patients has been identified who are characterized by predominantly ocular disorders without systemic involvement.[4,5] It has been suggested that molecular genetics and scrutiny of the phenotype will provide evidence that clinicians require for accurate diagnosis.[2] However, several cases of STL with different degrees of severity and manifestations, and genetic background, have been reported mainly in the Western world.

Case report

We report on a 25-year-old Japanese woman who was referred to our clinic with a diagnosis of rhegmatogenous retinal detachment of the right eye. Family history revealed that her mother had undergone retinal detachment surgery in her forties. On the initial examination, her best-corrected visual acuity was 20/20 OU, and her refraction was −9.0 diopter sphere (DS) OD eye and −7.5 DS OS. Anterior-segment examination was unremarkable with clear lenses. Vitreous examination confirmed the presence of a type I membranous vitreous anomaly (Figure 1a and b). Ophthalmoscopy showed a horseshoe tear surrounded by a retinal detachment in the right peripheral retina, and circumferentially oriented lattice degenerations in both eyes. Atrophy of the retinal pigment epithelium, choriocapillaris and radial perivascular degeneration were not seen. No systemic abnormalities were found. We performed scleral buckling on the right eye and the detached retina was reattached.

Although we had tentatively diagnosed the proband with predominantly ocular STL type I based on her ocular features, we could not completely exclude other possibilities because of the unknown genetic a etiology of STL in the Eastern world. In addition, the absence of systemic involvement indicated that the patient had not met the criteria for the diagnosis of STL proposed by Snead.[1]

After obtaining informed consent, we performed direct sequencing of all coding regions of the *COL2A1* gene and found a heterozygous deletion of a G at position 237, which predicts a downstream premature stop codon in exon 5 of the *COL2A1* gene (accession number: NM001844) (Figure 2). Her mother, who declined ophthalmic examination, carried the same mutation in the *COL2A1* gene in the heterozygous state. This deletion was not detected in her father and 45 healthy controls.

Comments

Our study adds a novel mutation of the *COL2A1* gene to the existing mutations that causes STL type I. Based on the mutational analyses, we counseled our patient that her future children should undergo ophthalmic examinations and molecular analysis for earlier diagnosis or exclusion of STL. Our observations further supported the idea that, irrespective of race, mutations involving exon 2 of the *COL2A1* gene are characterized by a predominantly ocular STL phenotype.[4,5]