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Spontaneous resolution of retinoschisis and consequent development of retinal detachment in highly myopic eye

Optical coherence tomography (OCT) has indicated that myopic foveoschisis (MF) is not uncommon in highly myopic eyes.¹ MF is often accompanied by a macular hole (MH),^{2,3} while a MH often complicates a retinal detachment (MHRD) in highly myopic eyes. MHRD is believed to result from MF at least in some cases, however, the detail of the process is unknown. We have documented the process of spontaneous resolution of MF and development of RD over a few months.

Case 1

A 65 year old man with high myopia was referred to our clinic with bilateral visual loss and metamorphopsia of 3 months' duration. OCT revealed MF without MH in his right eye and MF with MH in his left eye (fig 1A and C). His right eye underwent vitrectomy, but the left eye was observed. The initial best corrected visual acuity (BCVA) in his left eye was 20/50 and the axial length was 28.42 mm.

Six months later, OCT showed MHMF (fig 1D) and the BCVA decreased to 20/100 in his left eye. Eight months later, MF was resolved and developed MHRD instead (fig 1B and E), although the involved area seemed to be similar. The BCVA was 20/70. However, he refused surgery because he was asymptomatic. At the last visit (16 months after the

Figure 1 Case 1. A fundus photograph (A) and optical coherence tomography (OCT) image (C) at the first patient visit show myopic foveoschisis with a macular hole (MHMF) in the left eye. The best corrected visual acuity (BCVA) at the time was 20/50. An OCT image 6 months after the first visit (D) shows MHMF. A fundus photograph (B) and OCT image 8 months after the first visit (E) show MH and retinal detachment. The BCVA at the time has decreased to 20/70, but the patient is asymptomatic.

Figure 2 Case 2. A fundus photograph (A) and optical coherence tomography (OCT) image at the first patient visit (C) show myopic foveoschisis with a macular hole (MHMF) in the left eye. The best corrected visual acuity (BCVA) at the time was 20/70. One month later, OCT (D) shows a MH and retinal detachment (MHRD). The BCVA has decreased to 20/100. Two months after the first visit, the fundus photograph (B) and OCT (E) show MHRD. The extent of the RD has increased, and the patient opted to undergo vitrectomy.

first visit) the MHRD had not resolved and the BCVA remained at 20/50.

Case 2

A 73 year old woman was referred to our clinic with metamorphopsia in her left eye of 3 months' duration. The BCVA was 20/70 and the axial length was 29.39 mm. OCT revealed MHMF (fig 2A and C).

One month later, she reported visual loss in her left eye, and OCT showed the resolution of MF and consequent development of MHRD (fig 2D). The BCVA decreased to 20/100. Two months after the first visit, the fundus photograph and OCT still showed MHRD (fig 2B and E). Because the RD has extended and the BCVA remained 20/100, she chose vitrectomy with internal limiting membrane peeling and gas tamponade. After vitrectomy, the MHRD resolved, but the MH did not close. At the last visit (20 months after the first visit), the MH remained open and the BCVA was 20/200.

Comment

The pathogenesis of MHRD remains unclear, but the tangential traction of the vitreous cortex, the presence of an epiretinal membrane and posterior staphyloma are thought to have an important role in the development of MHRD,⁴ while inflexibility of the retinal vessels and the internal limiting membrane is believed to be a cause of MF.⁵ However, there is little information regarding this association. In this case series, we reported two cases of MHMF in which retinoschisis spontaneously was resolved and developed into RD.

The actual mechanism of the manner in which MF is resolved and develops into MHRD is unknown. The photoreceptor cells (the outer layer of MF) do not have a pump effect, which is one of the most important functions of the retinal pigment epithelial (RPE) cells. We hypothesise, therefore, that the pump effect of the RPE increased the osmolality of the subretinal fluid and that the intraretinal fluid moved into the subretinal space according to the incline of the osmotic pressure, resulting in the development of MHRD. Another possibility is that the photoreceptors are freed from the RPE according to RD, and MF may resolve because of the restitutive force of the intraretinal columns. In conclusion, spontaneous resolution of retinoschisis and consequent development of RD is one of the most important steps in causing MHRD, at least in some cases.

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Congenital stationary night blindness associated with mutations in GRM6 encoding glutamate receptor MGLuR6

Congenital stationary night blindness (CSNB) is a non-progressive retinal disorder characterised by defective night vision from birth¹ that is clinically and genetically heterogeneous. It is most commonly inherited as an X linked disorder, but autosomal dominant (OMIM #163500) and recessive (OMIM#258100) forms have been described. A recent report describes mutations of the *GRM6* gene in three patients with autosomal recessive CSNB.² *GRM6* encodes the glutamate receptor MGLuR6, which is expressed by the rod and cone ON bipolar cells.³ Pharmacological block of this receptor in primates⁴ leads to a similar CSNB phenotype as that described in subjects with *GRM6* mutations.² We have previously identified mutations in *CACNA1F* and *NYX* genes in males with X linked CSNB,^{5,7} and have encountered several female patients with a similar phenotype. We screened these affected females for mutations in *RHO* which can cause autosomal dominant CSNB,⁸⁻¹⁰ but failed to identify mutations. Recently, following the publication of *GRM6* mutations in CSNB² we have identified nonsense mutations in one female with CSNB.

Case report

The patient was 7 years of age at the time of clinical examination. Her parents thought

that she had always had poor night vision and had noticed horizontal nystagmus since early infancy. She had worn spectacle correction for myopic astigmatism since the age of 3 (right eye: -2.50DS/-3.5DC×85, left eye: -2.50DS/-4.00DC×100). She was otherwise fit and well, and had no family history of ocular disorders. Her distance visual acuity was logMAR +0.3 (6/12, 20/40) in each eye and she was able to read N5 print. Colour vision was full (Ishihara and City University plates). She had no nystagmus in primary position but a small amplitude horizontal nystagmus was noted in lateral gaze. She had a small angle esophoria and 500 seconds of arc stereoacuity. Funduscopy showed inferotemporally tilted discs with mild hypopigmentation in the inferotemporal fundus. There was no ophthalmoscopic evidence of foveal hypoplasia. The patient underwent ISCEV standardised electroretinographic testing using contact gold foil electrodes (protocol previously described)⁷ (fig 1). Subjects with AR or XLCSNB characteristically have a "negative wave" electroretinogram (ERG) with grossly subnormal scotopic (rod photoreceptor pathway) b-wave amplitudes. The patient's ERG waveform was more similar to that in XLCSNB1 ("complete") rather than XLCSNB2 ("incomplete"): the differences being the absence of recordable scotopic oscillatory potentials and the presence of a 30 Hz flicker response in this and the XLCSNB1 subjects. Blood was taken, with informed consent, from the patient and DNA extracted using standard techniques. Using polymerase chain reaction (PCR) we amplified each of her *GRM6* gene exons² and sequenced the coding region and intron-exon boundaries. We identified two nonsense mutations: R238X and Y409X, in exons 2 and 4 respectively (fig 2). These mutations specific have not been previously reported in CSNB but other nonsense mutations are described.²

Figure 1 Comparison of ERG waveforms. The absence of oscillatory potentials and the presence of a 30 Hz flicker response make the ERG in this case more similar to that seen in subjects with the X linked CSNB1 genotype than those with CSNB2.