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 tract 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, while inflexibility of the retinal vessels and the internal limiting membrane is believed to be a cause of MF. 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 osmolarity 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 the incline of the osmotic pressure, resulting in the development of MHRD. Further investigation is required to understand the mechanism of MHRD formation.

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**References**
suggest that nyctalopin (encoded by NYX) plays an important part in signalling through normal. But the 30 Hz flicker response is grossly subnormal. The striking similarities between GRM6 CSNB and CSNB1 caused by NYX mutations suggest that nyctalopin (encoded by NYX) plays an important part in signalling through the "on" bipolar pathway. Since little is known about the function of nyctalopin these clinical observations may help to guide further studies of its function and any interaction with mGluR6 in bipolar cell function.

Comment

Mutations in GRM6 cause some cases of ARCSNB. In darkness, glutamate is tonically released from the photoreceptor presynaptic terminal while light induced photoreceptor hyperpolarisation diminishes its release. The reduced stimulation of the "on" bipolar cell in light conditions results in its depolarisation. Absence of the MgluR6 receptor as a result of GRM6 gene mutation causes the "on" bipolar cells to be relatively depolarised (light adapted) in darkness, causing the symptom of night blindness. Both this form of ARCSNB and XLCNSNB1 appear to profoundly affect the "on" bipolar cell retinal pathway only and therefore have similar waveforms: specifically grossly subnormal, often absent scotopic b-wave response, an absence of scotopic oscillatory potentials but a relatively well preserved 30 Hz flicker response. In subjects with a GRM6 mutation, a normal b-wave and oscillatory potentials are usually recordable but the 30 Hz flicker response is grossly subnormal.

Metastatic choriocarcinoma causing cavernous sinus syndrome

Choriocarcinoma is a rare trophoblastic tumour. We describe a case of metastatic choriocarcinoma to the cavernous sinus causing complete ophthalmoplegia and ptosis, following an uneventful pregnancy.

Case report

A 28 year old woman was referred to the oculoplastic and orbital service with a 1 week history of right sided ptosis and 6 weeks of right facial pain. Six weeks earlier she underwent evacuation of retained products of conception, following spontaneous vaginal delivery. Facial pain immediately ensued and her dentist extracted a right upper tooth, but the pain continued to worsen. The visual acuity was 6/9 right eye, 6/5 left eye, with complete right sided ptosis, total ophthalmoplegia, and a dilated unreactive pupil (fig 1A, B). Reduced sensation in the distribution of right trigeminal branches V1 (including cornea and V2 was noted. There was no relative afferent pupillary defect, visual field or colour vision defects, or proptosis...

Magnetic resonance imaging (MRI) scans of brain and orbits (fig 1C, D), revealed a right cavernous sinus mass extending to right temporal lobe. Image guided biopsy of the temporal lobe revealed a dural encased highly vascular choriocarcinoma. Subsequent computed tomography (CT) imaging revealed a grossly enlarged uterus and multiple large pulmonary metastases. Serum β-HCG levels were markedly elevated (13.9 x 10^9 IU/L). Consistent with choriocarcinoma. She was commenced on systemic and intrathecal chemotherapy. Five months later, β-HCG levels returned to normal, her facial pain and sensation improved, and her ptosis resolved.

MRI scans at 6 months revealed a reduced tumour size (fig 1E and F). She remains clinically stable 18 months after presentation with a useful field of binocular single vision.

Comment

This patient had a potentially fatal cavernous sinus and intracranial metastasis from chorionicarcinoma. Patients can become symptomatic long after an uneventful pregnancy.1

References


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Figure 1

(E, F) MRI scans 6 months after chemotherapy, showing reduction in tumour size. Reproduced with permission.

Figure 2 Mutations in GRM6 gene.

Comment

Mutations in GRM6 cause some cases of ARCSNB. In darkness, glutamate is tonically released from the photoreceptor presynaptic terminal while light induced photoreceptor hyperpolarisation diminishes its release. The reduced stimulation of the “on” bipolar cell in light conditions results in its depolarisation. Absence of the MgluR6 receptor as a result of GRM6 gene mutation causes the “on” bipolar cells to be relatively depolarised (light adapted) in darkness, causing the symptom of night blindness. Both this form of ARCSNB and XLCNSNB1 appear to profoundly affect the “on” bipolar cell retinal pathway only and therefore have similar waveforms: specifically grossly subnormal, often absent scotopic b-wave response, an absence of scotopic oscillatory potentials but a relatively well preserved 30 Hz flicker response. In subjects with a GRM6 mutation, a normal b-wave and oscillatory potentials are usually recordable but the 30 Hz flicker response is grossly subnormal.

The striking similarities between GRM6 CSNB and CSNB1 caused by NYX mutations suggest that nyctalopin (encoded by NYX) plays an important part in signalling through the “on” bipolar pathway. Since little is known about the function of nyctalopin these clinical observations may help to guide further studies of its function and any interaction with mGluR6 in bipolar cell function.

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Figure 2 (A, B) Right sided complete ptosis, fixed dilated pupil, and total ophthalmoplegia. (C, D) Axial and coronal post gadolinium T1 MRI brain scans, showing enhancing right cavernous sinus mass extending to right temporal lobe surface. Arrows illustrate tumour location. (E, F) MRI scans 6 months after chemotherapy, showing reduction in tumour size. Reproduced with permission.
Congenital stationary night blindness associated with mutations in GRM6 encoding glutamate receptor MGlur6
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