LETTERS TO THE EDITOR

Vogt–Koyanagi–Harada disease masquerading anterior ischaemic optic neuropathy

EDITOR,—Vogt–Koyanagi–Harada (VKH) disease is a chronic panuveitis associated with poliosis, alopecia, vitiligo, dysacusia, and meningal signs.1 In VKH, optic disc involvement is not unusual and disc oedema is one of the hallmarks of the disease.2 However, severe visual loss, atypical visual field defect, and sector filling defect on fluorescein angiography that suggest anterior ischaemic optic neuropathy (AION) are not usual presentations. We describe a case of VKH masquerading AION.

CASE REPORT

The patient was a 68 year old man who had decreased right visual acuity. He had systemic hypertension and multiple small brain infarctions. On initial examination, the visual acuity was 20/50 in the right eye and 20/30 in the left. The optic discs were swollen and sector filling defect on fluorescein angiography was seen. Slit lamp examination showed no signs of ocular inflammation but fundus examination revealed swelling of both optic discs. Eight days later, keratic precipitates and cells in the anterior chamber were found. Multiple serous retinal detachments in the both eyes occurred on the 14th day. Enlargement of the Marriotte blind spot and central scotoma were demonstrated by Goldmann visual field examination. Pleocytosis of the cerebrospinal fluid and human leucocyte antigen (HLA) type DR4 were found. Computed tomography showed small infarctions in the bilateral basal ganglia and the pons.

Fluorescein angiography (FA) showed a wedge-shaped filling defect in the optic disc of the left eye in the early phase and later leakage of the dye from both optic discs (Fig 1A). On the day 8, bilateral hypofluorescence in the posterior pole typical of VKH was observed. Indocyanine green (ICG) videoangiography showed filling delay of choroidal circulation.3 Choroidal vessels were indistinct and speckled hypofluorescence on the diffuse background fluorescence was seen in the early to late phase (Fig 1B). Three months later, the fundus was unchanged with decreased depth of optic disc of VKH. The visual function of the right eye showed some recovery but visual acuity in the left eye was 15/200 and no recovery in the visual field defect was observed.

COMMENT

In this case, at the early stage of disease, signs and symptoms suggestive of AION were found. Typical features of VKH emerged on later examinations. Although disc oedema is a common finding in VKH,2 severe visual loss, sector filling defect, and visual field loss are unusual in VKH. Sector filling defect in the disc on FA and altitudinal visual field defect are more compatible with AION. Whether AION was caused independently of VKH or inflammatory cells in the choroid caused circulatory disturbances is hard to determine because the patient was at high risk of circulatory disorders. But disturbed choroidal circulation by inflammatory cells in VKH may support the latter idea. Indeed, Perry and Font4 reported pathology of severe cases of VKH. In their cases, infiltration of the leptomeninges by lymphocytes and mild to moderate arachnoidal proliferation were found. Severe visual loss from optic nerve involvement in VKH is an important but not well documented complication of VKH.

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Acute retinal necrosis after neonatal herpes encephalitis

EDITOR,—We read with interest the letter of Pavesio et al reporting acute retinal necrosis (ARN) in a 17 year old who had had neonatal herpes simplex virus (HSV) encephalitis.5 We would like to report a patient who also had neonatal HSV encephalitis who had been treated with parenteral aciclovir and developed ARN 16 years later.

CASE REPORT

A 16 year old white woman with a history of mental retardation and seizures as a result of neonatal HSV encephalitis presented with a 5 day history of her left eye being red and apparently uncomfortable. On presentation she was found to have moderate anterior chamber and vitreous inflammation and peripheral retinal necrosis in her left eye for 8 clock hours. There were no choriorretinal scars or optic atrophy indicating earlier infection. The right eye was normal. The diagnosis of ARN was made and she was treated with intravenous aciclovir. There was no history of any recurrent HSV and she was otherwise in good health. Her seizure disorder remained well controlled on her usual dose of carbamazepine.

After 6 days of treatment her retinitis was responding to parenteral aciclovir but she was found to have a small peripheral retinal detachment and a viretectomy was performed. Polymerase chain reaction (PCR) was performed at the National Eye Institute on the vitreous biopsy and revealed the presence of HSV DNA.

COMMENT

Both our patient and the patient reported by Pavesio et al5 developed ARN at about the same age, but it will take a larger series to know whether this is significant. Thompson et al6 reported two patients with neonatal and infantile central nervous system (CNS) HSV infections who developed ARN at ages 10 and 4, respectively. A 30 year old woman with ARN who had had recurrent HSV disease, although small lesions in the peripheries may have been obscured by the active retinitis. Whether such findings are indicative of the need for long term prophylaxis with aciclovir in children who had herpetic encephalitis as suggested by Pavesio et al,1 is an important issue. Prophylaxis could be considered after HSV encephalitis to prevent ARN or after ARN to prevent second eye involvement. Aciclovir used as prophylaxis for recurrent genital HSV infection in adults for 5 or more years has been associated with minimal toxicity and the selection of resistant strains has not been demonstrated,1 but there is little experience with the duration of prophylaxis that would have been necessary to prevent ARN in our patients. The presence or absence of earlier ocular involvement may not be a good indication of the need for long term prophylaxis to prevent ARN after HSV encephalitis as has been suggested.1 One child reported by Thompson et al had recurrent presumed cutaneous herpes and the other had choriorretinal scars,5 and the patient reported by Pavesio et al had optic atrophy before the diagnosis of ARN.1 Our patient had no apparent previous ocular findings nor has she had any other recurrent HSV disease, although small lesions in the periphery may have been obscured by the active retinitis. Whether such findings are indicative of an increased risk of the development of ARN cannot be determined from isolated case reports.

Second eye involvement in ARN is not uncommon.5 Although Palay et al7 reported that treatment of ARN with aciclovir does decrease second eye involvement in the short term in an adult population with most cases likely to be from varicella zoster virus, we do not recommend long term prophylaxis.5 Like

Figure 1 (A) Fluorescein angiography of the left eye. A wedge-shaped filling defect was found in the optic disc (arrow). (B) Indocyanine green angiography of the left eye. Indistinct choroidal vessels and speckled hypofluorescence on the diffuse background fluorescence were found.

Figure 1 (A) Fluorescein angiography of the left eye. A wedge-shaped filling defect was found in the optic disc (arrow). (B) Indocyanine green angiography of the left eye. Indistinct choroidal vessels and speckled hypofluorescence on the diffuse background fluorescence were found.
Pavesio et al. have chosen to continue oral aciclovir as long term prophylaxis against second eye involvement, but more data are needed to guide us in the appropriate management of these patients.

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CASE REPORT

Disc detachments and optic pit vortices are often complicated with sensory macular retinal detachment, but their symptoms rarely become significant before the age of 20. Based on a review of 15 patients with optic nerve pits and maculopathy, Lincof and associates suggested that the retinal elevation is most often due to communication between the optic nerve pit and a schisis-like separation of the inner and outer retinal layers that may occur secondarily and in association with an outer lamellar macular hole. This finding was recently confirmed by optical coherence tomography (OCT). In addition, OCT suggests that the formation of an outer lamellar macular hole may be secondary to chronic cystoid macular edema. To our knowledge, however, this mechanism has not been identified in eyes with optic disc coloboma. Lincof et al. suggested that in eyes with a maculopathy associated with optic disc pits, the fluid from the pits entering the disc elevates the nerve fibre layer, causing a schisis-like separation of the inner retinal layers. Even though FA of the present case did not show the origin of subretinal fluid, we were able to confirm our diagnosis of retinoschisis and lamellar macular hole based on fundus examinations including SLO and FA, which clearly revealed an elevated inner layer connected to the disc coloboma. These findings are very similar to those of Lincof et al’s cases demonstrating an irregular and partial thickness macular hole and schisis with an outer layer detachment which doe not extend to the optic disc. Non-rhegmatogenous retinal detachment may occur in association with disc colobomas. We believe that the pathogenesis of the schisis-like separation identified in optic nerve pits and optic disc colobomas may be similar.

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Corneal endothelial changes and trinucleotide repeat expansion of DRPLA gene

EDITOR.—Dentatorubral and pallidolysian atrophy (DRPLA) is an autosomal dominant disorder that manifests in a combination of chorea, myoclonus, seizure, ataxia, and dementia. It is caused by the unstable expansion of a CAG trinucleotide repeat coding for glutamine in the DRPLA gene. Several other genes with an unstable trinucleotide repeat expansion of CAG were cloned in some types of spinocerebellar degeneration (SCD). Several reports have also suggested an association between ocular changes and SCD. We report here the association of ocular changes in patients with an expanded allele of the trinucleotide repeat of the DRPLA gene.

CASE REPORT

A 46 year old woman (IV-2 in Fig 1) noticed gait disturbance and truncal ataxia at age 36 years. When we visited her, her general condition was very severe, and visual acuities were not examined. Pupils, ocular media, and fundus examination showed normal findings. Corneal endothelial cell density was 762 cells/mm² right eye and 540 cells/mm² left eye by specular microscopy (Fig 2). DNA

Figure 1  Fundus photograph of the left eye showing dome-shaped macular detachment (*) with an outer lamellar macular hole with irregular margins ($) surrounded by a schisis-like separation between the inner and outer retina (>). Note the optic and adjacent choroidal coloboma.

Figure 2  Fluorescein angiogram of the left eye showing early (top) and late (bottom) hyperfluorescence of the disc and choroidal coloboma. Note the lack of hyperfluorescence in the macular region.
analysis from peripheral lymphocytes revealed that she had an expanded allele of the DRPLA gene with a normal allele (expanded/normal allele; 67/20), which was estimated by a fluorescein isothiocyanate (FITC) labelled primer used for polymerase chain reaction (PCR) amplification, according to our previous methods. A 39 year old woman (IV-4 in Fig 1) noticed truncal ataxia at age 36 years, and the symptoms have gradually increased. When she consulted us her visual acuity was 1.0 in both eyes. Pupillary reactions, intraocular pressures, ocular media, and fundus examination findings were normal. Corneal endothelial cell density was 951 cells/mm² and were free of other ocular changes. Although we will continue to follow these patients for other ocular symptoms, our patients IV-2 has had a long history of the disease (10 years), and we believe that the findings are specific to the mutation of the DRPLA gene. We will also examine other families with the mutation of the DRPLA gene to exclude the possibility of corneal endothelial dystrophy.

COMMENT

Neither patients had a history of trauma, inflammation, or surgery of the eye. The difference in corneal endothelial cell density was statistically significant, compared with that in healthy members of the family; in patients with Machado–Joseph disease, who have a trinucleotide repeat expansion allele with a normal repeat allele of the Machado–Joseph disease gene; and in age matched normal healthy controls in our clinic (p=0.004, unpaired t test) (data not shown). The enlargement of the cell may be generated by a cytopathological condition of the cell due to an impropriety function of the DRPLA protein (termed atrophin-1), which may be expressed in corneal endothelial cells or adjacent cells. Previously, decreased corneal endothelial cell density, optic atrophy, and mild attenuation of oscillatory potentials observed on ERG in patients with spinocerebellar degeneration and trinucleotide expansion allele with a normal repeat allele of the Machado–Joseph disease gene have been reported. However, our patients showed only decreased corneal endothelial cell density and were free of other ocular changes. Although we will continue to follow these patients for other ocular symptoms, our patient IV-2 has had a long history of the disease, and we believe that the findings are specific to the mutation of the DRPLA gene. We will also examine other families with the mutation of the DRPLA gene to exclude the possibility of corneal endothelial dystrophy.