CASE REPORT

Bilateral consecutive optic neuropathy in a patient with thrombophilia

Nurgül Örnek, Zafer Onaran, Kemal Örnek, Nesrin Büyüktortop

Department of Ophthalmology, Kırıkkale University, Kırıkkale, Turkey

Correspondence to
Dr Kemal Örnek,
kemalornek@hotmail.com

SUMMARY

A 39-year-old man was admitted with a sudden visual loss in the left eye. Visual acuities were 10/10 on the right and 1/10 on the left. Fundus examination did not show any abnormalities. Visual acuity improved to 10/10 and visual field defect regressed in the following 2 weeks. Three years later, the patient returned with acute visual loss in the right eye. Visual acuities were 2/10 on the right and 10/10 on the left. Right optic disc had blurred margins with mild oedema. The tests revealed methylenetetrahydrofolate reductase A1298C mutation with positive lupus anticoagulant and hyperhomocysteinaemia. Enoxaparin was initialised with vitamin B_{12} supplementation. Complete visual recovery occurred in the following 3 weeks in both eyes. Thrombophilic screening seems to be important in the treatment and prevention of an attack in the second eye of patients with non-arteritic anterior ischaemic optic neuropathy.

BACKGROUND

Non-arteritic anterior ischaemic optic neuropathy (NAION) is a common cause of vision loss in the middle-aged and elderly. Several risk factors for NAION have been identified including nocturnal hypotension, sleep apnoea syndrome, hypertension, diabetes mellitus, arteriosclerosis, hypercholesterolaemia and prolonged surgical procedures. A significant association of NAION with crowded optic disks, markedly raised intraocular pressure, optic disk drusen and cataract extraction has been reported.[1] In addition, reports of thrombotic tendencies in patients with NAION suggested a possible causative relationship between thrombophilia and NAION.[2–4]

Here, we report the first case of bilateral consecutive NAION in a young patient associated with lupus anticoagulant, hyperhomocysteinaemia and heterozygosity for methylenetetrahydrofolate reductase (MTHFR) A1298C mutation with complete visual recovery.

CASE PRESENTATION

Three years ago, a 39-year-old man was admitted with an acute painless visual loss in his left eye. He had a history of transient cerebral ischaemic attack. Ocular examination showed best corrected visual acuity of 10/10 on the right and 1/10 on the left. The right visual field was normal, a visual field defect involving the superior field was found in the left eye. He had also a left afferent pupillary defect (APD) and congenital dyschromatopsia. Both optic discs did not show any abnormalities (figure 1A,B).

The patient did not have symptoms like headache, jaw claudication, scalp tenderness or other systemic discomfort. Complete blood count, biochemical investigations, coagulation profile and chest radiograph were in normal limits apart from low platelet levels. Erythrocyte sedimentation rate and C reactive protein were within normal ranges. He had no previous episodes and did not have symptoms suggestive of multiple sclerosis. He was afebrile. Cardiological and rheumatological examinations did not reveal any pathologies. The patient was started clopidogrel 75 mg once a day owing to the ischaemic lesions on the brain MRI. Visual acuity improved to 10/10 and visual field defect regressed in the following 2 weeks. He lost to follow-up after complete resolution of the symptoms.

Three years later, the patient returned with acute visual loss in the right eye. He had stopped using clopidogrel. On examination, best corrected visual acuities were 2/10 on the right and 10/10 on the left. Perimetry showed an inferior altitudinal visual field defect in the right eye, left visual field was normal. He had a right APD with dyschromatopsia on both sides. Right optic disc was hyperaemic with blurred margins and mild oedema and the left optic disc was normal (figure 2A,B).

INVESTIGATIONS

Neurological and routine laboratory and radiological investigations were normal. The patient was screened for inherited thrombophilic defects. The tests revealed a heterozygous MTHFR A1298C mutation with positive lupus anticoagulant. Homocystein level was high (15 µmol/L) and vitamin B_{12} level was low (187 pg/mL).

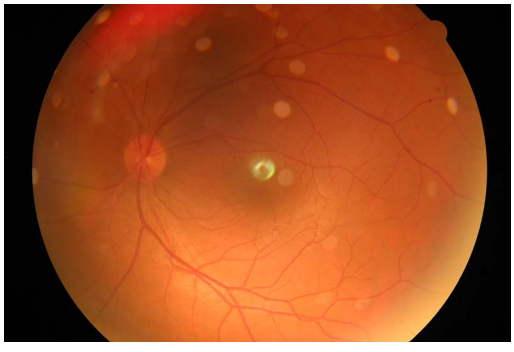

Figure 1 Normal optic nerve head in the left eye.

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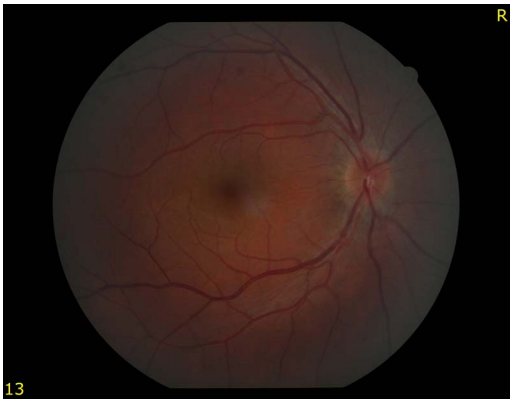

Figure 2 Right optic disc with blurred margin and mild oedema.

TREATMENT

The diagnosis of NAION was made owing to the clinical presentations, characteristic visual field loss and thrombophilic defects. Enoxaparin twice daily was initialised with vitamin B_{12} supplementation both for the treatment and prevention.

OUTCOME AND FOLLOW-UP

Visual acuity improved in the following week and visual field defect regressed.

DISCUSSION

NAION is a multifactorial lesion of the optic nerve head, which may be either unilateral or affect the other eye at a later time. It occurs owing to acute ischaemia of the optic nerve head supplied by the posterior ciliary arteries causing a sudden and painless visual loss. Besides various local and systemic risk factors, thrombophilic disorders seem to play a role in the pathogenesis of NAION.

Thrombophilia is a disorder of haemostasis in which there is a predilection for thrombosis to occur. These disorders associated with NAION include resistance to activated protein C, homocysteinaemia, the antiphospholipid antibody syndrome etc.[2–4] Our patient had lupus anticoagulant and hyperhomocysteinaemia as previously reported, but the presence of heterozygosity for MTHFR A1298C mutation is the first in the current literature in a young patient with NAION.

The contribution of thrombophilic defects to the pathogenesis of NAION has still some controversies. Glueck *et al*[5] reported that patients with NAION were more likely to demonstrate thrombophilic defects. Srinivasan *et al*[6] successfully treated NAION in a patient with primary antiphospholipid syndrome and factor V Leiden mutation using anticoagulants. On the other hand, Salomon *et al*[7] did not find a relationship between NAION and a wide range of thrombophilic risk factors.

In this case, NAION seems to be associated with inherited thrombophilia, heterozygosity for the A1298C mutation of the MTHFR gene combined with hyperhomocysteinaemia and lupus anticoagulant. The patient had no known cardiovascular or autoimmune disease. He had an NAION in the left eye followed by a complete visual recovery, and subsequently had a recurrence in the left eye. We suppose that, a reversible thrombosis may have led to transient decompensation of the blood supply to the optic disc, in the presence of thrombophilic defects as in this case or these defects might be a part of the multifactorial aetiology of ocular vascular diseases, and worked concurrently with other risk factors for the disease.

Learning points

- Thrombophilia with or without vascular damage may have a role in the development of non-arteritic anterior ischaemic optic neuropathy (NAION).
- Thrombophilic screening may be important in the treatment and prevention of an attack in the second eye of patient with NAION.
- Identifying inherited coagulation disorders, especially in young patients, should also be of value in the recognition and prevention of occlusions in other vascular beds of the body.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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