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Retinal ischaemia in type 1 neurofibromatosis

Systemic occlusive arteriopathy associated with type 1 neurofibromatosis (NF-1) has been well described in the aortic and cerebral vasculature¹; however, it is poorly documented in the retinal vasculature with, to our knowledge, only three reported patients whose retinal vasculopathy was diagnosed at a later sequelae stage.^{2–4} We report the case of a young woman with NF-1 who presented with acute macular ischaemia combined with diffuse retinal arterial occlusive disease.

Case report

A 26 year old woman with NF-1 presented with a sudden painless loss of vision in her left eye. Visual acuity was counting fingers left eye and 20/20 right eye. NF-1 had previously been diagnosed on the basis of multiple café au lait spots, plexiform neurofibromas, and a paternal history of NF-1. Left fundus examination revealed confluent cottonwool spots in the perifoveal area, focal dilations and narrowings of retinal venules, and progressive enlargement of retinal arterioles towards the periphery. The right fundus showed only peripapillary myelinated nerve fibres (fig 1).

Fluorescein angiography (FA) of the left eye showed no delayed perfusion in the choriocapillaris or central retinal artery, but a delayed retinal arteriovenous filling time. It revealed occlusion of macular arterioles, retinal arteriovenous communications in the mid-periphery, and a relatively well perfused retinal periphery (fig 2).

Cerebral and cervical magnetic resonance imaging and colour Doppler ultrasonography showed no vascular flow abnormalities. Internal medical examination and laboratory results excluded other diseases associated with retinal microvasculopathies.

One month after the initial examination, all the cottonwool spots disappeared, and sheathing appeared in a few veins. FA showed worsening of the retinal arteriolar occlusions in the mid-periphery (fig 2). A panretinal photocoagulation was subsequently performed. At 2 year follow up, left visual acuity remained unchanged, and the patient did not exhibit any preretinal fibrovascular proliferation.

Comment

To our knowledge, only three cases of retinal vascular occlusion in patients with NF-1 have been previously reported. All cases were diagnosed at a later sequelae stage. Moadel *et al.*² and Kadoi *et al.*³ respectively, reported the cases of a child who presented with an unilateral amblyopia, and a young woman with a history of unilateral poor vision. They both presented with diffuse sheathing of retinal vessels, retinal arteriovenous

Figure 1 Fundus photographs of the left and the right eyes at presentation. (A) Colour fundus photograph of the left eye showing large confluent cottonwool spots in the perifoveal area. (B) Colour fundus photograph of the right eye showing peripapillary myelinated nerve fibres. (C) Red-free fundus photograph of the left eye. Upper periphery showing focal dilations and narrowings of retinal veinules, and capillary rarefaction. Retinal arteriovenous communications are also present (arrows).

Figure 2 Fluorescein retinal angiography of the left eye, 40 seconds after dye injection, at presentation (A) and 1 month later (B). (A) Occlusion of macular arterioles and retinal arteriovenous communications are visible (black arrow). (B) Retinal arteriolar occlusions have worsened in the temporal periphery.

communications, and avascular peripheral retina with fibroglial proliferation. Thölen *et al.*⁴ described a young man whose routine eye examination showed narrowing of a superotemporal retinal arteriole with irregular diameter of the corresponding vein, fibroglial proliferation, and arteriovenous communications. These authors suggested the possibility of either a primary occlusion of a retinal arterial or venous branch or alterations of vessel development. In contrast with other reported cases, our patient presented with acute unilateral occlusion of macular arterioles, combined with diffuse retinal arterial occlusive disease, in adulthood. Panretinal photocoagulation may have prevented fibroglial proliferation which was described in previous reported cases. Recent reports suggest that pathogenesis of NF-1 vasculopathy may result from dysfunction of neurofibromin, the NF-1 gene product, in vessel endothelial and smooth muscle cells.⁵

Our case shows that NF-1 related occlusive retinal arteriopathy may suddenly appear during adulthood and should be considered in patients with NF-1 or in young adults with retinal vascular occlusive disease and unknown NF-1.

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doi: 10.1136/bjo.2005.077701

Accepted for publication 12 August 2005

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Are biometric parameters a risk factor for idiopathic macular hole formation? Results of a matched case-control series

Recognised risk factors for the formation of idiopathic full thickness macular holes (IFTMH) include age, female, sex, and high myopia.^{1,2} However, we noticed that patients with IFTMH within our population, tended to have shorter than normal axial lengths. An age and sex matched case-control study was

therefore performed to test this hypothesis and determine whether biometric factors may be associated with formation of IFTMH.

Methods

Consecutive patients undergoing surgery for IFTMH had age, sex, axial length in millimetres (mm), and corneal keratometry (K) measurements in dioptres (D) recorded.

For each case three age and sex matched controls were randomly identified from a cataract surgery database (Medisoft). Biometric measurements were made in all cases and controls using the IOL-Master (Zeiss, Jena). Mean corneal curvature was calculated by averaging the two K readings.

Descriptive statistics and conditional logistic regression method for analysis of matched pair data were used to infer estimates.

Results

Thirty five consecutive cases and 105 controls were identified with a mean age of 67.3 years (35–77 years) and a female to male ratio of 24:11. The mean (SD) axial length in the cases was 22.94 (0.96) mm whereas in controls it was 23.48 (1.44) mm. The axial lengths of the cases also fell within a narrower range than those of the controls (fig 1). Mean K readings in cases was 44.04 D (range 41.87–46) and 43.44 D in controls. (range 40.56–47.92).

Evidence suggests that the longer the axial length the less the odds of being a case; odds ratio (OR) 0.67 ($p = 0.036$, CI 0.47 to 0.77) and the higher the dioptre power of the curvature the higher the odds of being a case as opposed to a control; OR 1.50 ($p = 0.02$, CI 1.07 to 2.01).

Comment

As in previous studies our population sample demonstrates an increased risk of IFTMH in females with most cases clustering in the 60–70 year age group.^{3,4} The mean axial length in controls was also found to be similar to results in the published literature.^{5,6}

Mean spherical equivalence (MSE) was measured by the Eye Disease Case Control Study Group⁷ and although their findings were statistically insignificant, their regression analysis found an increased likelihood of hypermetropia in the IFTMH cases. Our cases clustered in a relatively narrow range of axial

lengths (21 mm–25.31 mm in the cases compared to 20.64 mm–29.48 mm in the controls) with the maximum axial length in the cases being 25.31 mm (fig 1). In Japanese literature, high myopia is a prominent, recognised risk factor and in one study the overall mean MSE was -0.66 D.² From our study it appears myopic macular holes are encountered much less frequently in a predominantly white population.

We believe the significant corneal dioptric differences found may be a result of the natural process of emmetropisation of a shorter eye, or alternatively be an independent risk factor.

IFTMH are thought to arise from retinal surface traction which in turn arises from an abnormal and incomplete posterior vitreous detachment.⁸ Our results suggest that eyes with shorter than average axial lengths and therefore lower volume vitreous cavity are either at increased risk of developing such incomplete and abnormal vitreous detachments or, in the presence of such an incomplete vitreous detachment, of developing a macular hole. We are not aware that shorter axial length has previously been shown to be significantly associated with IFTMH.

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doi: 10.1136/bjo.2005.077008

Accepted for publication 21 August 2005

RLJ has a commercial and proprietary interest in the cataract database from which control data were derived.

Figure 1 Axial lengths (mm) of patients with macular holes compared with age and sex matched controls.

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Choroiditis and exudative macular detachments in a post transplant leukaemic patient: an unusual presentation of *Pneumocystis jiroveci* infection

Pneumocystis jiroveci is a commensal found in the respiratory tract of healthy individuals. It can cause infections of almost any organ in immunocompromised hosts. *P. jiroveci* choroiditis, featuring multifocal choroidal lesions, has been reported in immunocompromised patients despite prophylactic use of inhaled pentamidine. We report a post-transplant leukaemic patient whose posterior segment findings demonstrate an atypical picture of recurrent *P. jiroveci* choroiditis.

Case report

A 20 year old woman presented with a 1 week history of bilateral blurred vision, dyspnoea, and dry cough. Visual acuity was reduced to 6/36 bilaterally with posterior subcapsular cataracts. Funduscopy disclosed multifocal, creamy-yellow choroidal lesions at both posterior poles with exudative retinal detachments at the maculae (fig 1). Optic discs and retinal vessels were normal. Fluorescein angiography showed early patchy hypofluorescence (corresponding to the choroidal lesions) and late hyperfluorescence with pooling beneath the detachments.

Three years earlier she had undergone a successful allogeneic bone marrow transplant for chronic myeloid leukaemia. Her maintenance therapy included inhaled pentamidine as prophylaxis against *P. jiroveci* infection. However, she admitted to not taking her inhaled pentamidine. Molecular genetic and radiological investigations excluded leukaemic recurrence. Chest x ray showed left lung shadowing and bronchoalveolar lavage isolated *P. jiroveci*. *P. jiroveci* pneumonia with *P. jiroveci* choroiditis was diagnosed and high dose oral co-trimoxazole 7.2 g/day (120 mg/kg/day) and prednisolone 60 mg/day commenced. Within 2 weeks her respiratory and ocular symptoms improved. Visual acuities returned to 6/9 and the fundal appearances resolved. She was discharged on daily oral co-trimoxazole 960 mg, prednisolone 20 mg, and inhaled pentamidine.