Combined central retinal vein occlusion and cilioretinal artery occlusion in an MTHFR C677T homozygous 26-year-old patient with hyperhomocysteinemia: case report

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INTRODUCTION

In the present report, we describe the case of a young female with combined non-ischemic central retinal vein occlusion (CRVO) and nasal cilioretinal artery occlusion (CLRAO) associated with hyperhomocysteinemia and homozygosity for the MTHFR C677T mutation. The coexistence of CRVO and CLRAO has been documented in the past, however our case, is one of the very few to report this occurrence along with hypermocysteinemia and the genetic mutation MTHFR C677T.

CASE REPORT

A healthy 26-year-old female presented with sudden, painless visual disturbances in her right eye (RE) which started the evening before and gradually increased in severity.

A complete clinical examination was performed. Best corrected visual acuity (BCVA) was 10/10 bilaterally and anterior segment was normal. She had normal pupillary reaction and no relevant afferent pupillary defect.

Fundus examination: RE revealed signs of a CRVO (hemorrhages, mild venous tortuosity, mild disc swelling and blurred disc margins) & a white ischemic area nasal to the optic nerve. LE normal.

FA: mild attenuation of the arterial filling as well as a mild delay in venous filling corresponding to the nasal area of CRAO. Late frames: mild staining of veins and mild leakage of the optic disc.

OCT-A: mild venous tortuosity. However, when all slabs were projected in a single en-face image, a dark area corresponding to the CRAO was seen, most likely the result of masking due to retinal oedema.

Structural OCT: hyperreflectivity of the inner retinal layers

HVF: RE temporal peripapillary scotoma

Cardiological and hematology evaluation & genetic testing for thrombophilic mutations: increased levels of homocysteine and a positive genetic test for the MTHFR C677T polymorphism.

Prescription of folic and acetylsalicylic acid & follow-up demonstrated gradual improvement on fundoscopy.

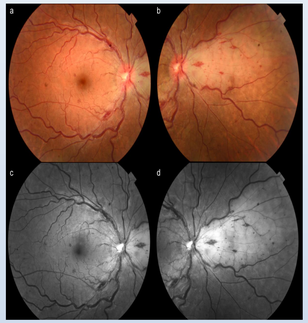

Figure 1. Color (a,b) and red-free (c,d) fundus photographs of the right eye including the macula (a,c) and nasal retina (b,d). Scattered flame-shaped, dot and blot hemorrhages, mild venous tortuosity, optic disc oedema as well as whitening of the retina nasally of the optic disc.

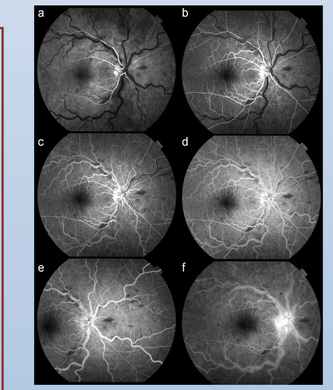

Figure 2. Fluorescein angiography. a-d) Early and mid-frames showing mild attenuation of the arterial filling as well as a mild delay in venous filling corresponding to the nasal area of CRAO. d-f) Late frames showing mild staining of veins and mild leakage of the optic disc.

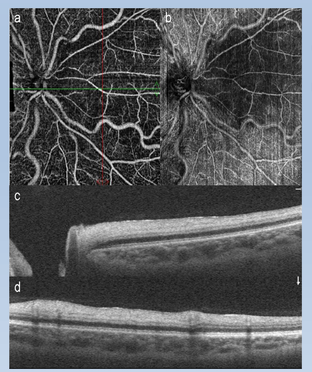

Figure 3. Optical coherence tomography angiography of the nasal ischemic area. a) En-face image (superficial slab) showing mild venous tortuosity b) En-face projection of all slabs showing a dark area corresponding to the CRAO c-d) Cross-sectional scans (corresponding to the green and red lines on a) showing hyperreflectivity of the inner retinal layers.

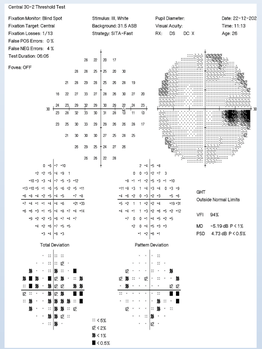

Figure 4. Humphrey visual fields of the right eye showing a temporal peripapillary scotoma corresponding to the area of CRAO.

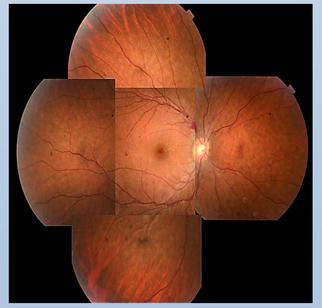

Figure 5. Color fundus photograph montage of the right eye 3 months after presentation, showing less retinal haemorrhages and venous tortuosity. The retinal whitening nasal to the optic disc is resolved and there are some small hard exudates between the fovea and the optic disc.

CONFLICT OF INTEREST

The authors report no conflicts of interest.

DISCUSSION

Cilioretinal arteries (CLRA) originate from the posterior ciliary circulation and are found in approximately 32% of eyes. Only 6.7% out of 2000 eyes investigated in a study by Justice et al, had a nasal cilioretinal artery(1). Their occlusion can be due to atherosclerosis of the carotid artery as well as anterior ischemia in the setting of optic neuritis(2) and it can also be combined with a CRVO.

The underlying pathogenesis of combined CRVO and CLRAO has not been clarified so far. It has been argued that the primary occlusion of the cilioretinal artery might co-exist with CRVO due to obstruction of the posterior ciliary arteries in association with optic neuropathy(3). Furthermore, the theory of “choroidal steal” was introduced, referring to blood redirection from the central retinal artery (CRA) and CLRA into the choroidal bed after a CRVO(4). On the contrary, Hayreh’s hemodynamic block theory(5) suggested that both decreased arterial pressure and increased venous pressure can lead to changes in the intraluminal perfusion pressure. Thus, due to anatomical particularities and the lack of CRA-like autoregulation in the choroidal vascular bed(6), the cilioretinal artery seems to be vulnerable to the transient hemodynamic blockage caused by CRVO.

In our case, homocysteine blood levels were moderately elevated, and genetic testing revealed homozygosity for the polymorphism MTHFR C677T, a cause of hyperhomocysteinemia. While hyperhomocysteinemia is associated with retinal vasvular occlusion (RVO), the exact role of the MTHFR C677T polymorphism remains unclear(7). A vitamin therapy (Vitamins B6, B12 and folic acid) has been suggested, aiming to decrease homocysteine levels(8).

The prognosis of combined CRVO and CLRAO in young adults is generally considered to be good(9) due to the transient nature of the hemodynamic blockage and the establishment of collateral circulation that most often contribute to the restoration of damage(10). Our case showed signs of improvement on follow-up and BCVA remained at 10/10.

The combination of our patient’s young age, negative personal or family medical history as well as the nasal distribution of the occluded cilioretinal artery are all features that have been reported in only a few cases in the past. Further research should investigate in detail the association of the genetic mutation MTHFR C677T with combined CRVO and CLRAO, as well as the therapeutic regimens, including vitamin therapy.

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