Hyperhomocysteinemia Methylene Tetrahydrofolate Reductase C677T Mutation in a Case of Hemicentral Retinal Vein Occlusion

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Purpose: To report a case of hemicentral retinal vein occlusion with homozygote MTHFR C677T mutation and high plasma homocysteine levels.

Methods: A 35-year-old male presented with a 10 days history of sudden visual impairment on his left eye. Funduscopic examination revealed hemicentral retinal vein occlusion. The patient was also evaluated with optic coherence tomography (OCT), fluorescein angiography (FA), biochemical and genetic tests.

Results: The visual acuity in the left eye was 20/100. The anterior segment was normal on slit-lamp biomicroscopy. Fundus examination showed a superior hemiretinal vein occlusion with venous dilatation and tortuosity, flame-shaped hemorrhages and retinal edema. Severe cystoid macular edema was confirmed by OCT and FA. There was no ischemia on FA. Folic acid level was 5.75 ng/ml (normal range: 5.38-24), vitamin B12 level was 160 pg/ml (normal range: 211-911) and plasma homocysteine level was 16.6 micromol/L (normal range: 5-14). Genetic investigation for thrombophilic conditions showed homozygote MTHFR C677T mutation. The patient received folic acid (5 mg/day) and vitamin B12 (1000 mcg/week) in addition to intravitreal bevacizumab injection for cystoid macular edema.

Conclusion: Hyperhomocysteinemia is modifiable risk factor for thrombotic diseases. Measurement of plasma levels of homocysteine and genetic counseling for thrombophilic disorders, which might benefit from folic acid and vitamin B12, should be done in patients with retinal vein occlusions.