

# LETTERS

## Homocystinuria and Marfan's Syndrome

### To the editor:

In the "Clinical Challenge" article, "Two Neuro-Ophthalmic Episodes Separated in Time and Space," (Cullom RD Jr, Cullom ME: *Surv Ophthalmol* 40:217-224, [November-December], 1995), the authors present a very interesting case report, with comments by Randy Kardon and Kathleen Digre, about a 46-year-old woman with a prior history of a spontaneously resolving right central retinal artery occlusion.<sup>2</sup> The patient presented with an acute right third order Horner's syndrome and vague ocular pain. Magnetic resonance angiography demonstrated a right internal carotid artery dissection. The dissection improved on anticoagulation therapy. Marfan's syndrome or Ehlers-Danlos syndrome was suspected because of tall stature, long digits and pectus excavatum, but ruled out by clinical criteria and skin biopsy, respectively. A common cause was not found for the two neuro-ophthalmic episodes.

When I was reading this case report, I wondered why homocystinuria was not excluded. Homocystinuria is one of the causes for premature, multifocal and recurrent vascular complications, including central retinal artery occlusion and carotid artery obstruction.<sup>3-6</sup> Osteoporosis, mentioned in the past history of the patient, is another major sign of homocystinuria.<sup>3</sup> The marfanoid features of the patient are a known cause for misdiagnosis of homocystinuria as Marfan's disease.<sup>1</sup> I believe that homocystinuria should be excluded in this patient by measuring the metabolites of methionine metabolism in serum and the enzyme activity of cystathionine synthase in cultured fibroblasts, because confirmation of that diagnosis has consequences for treatment.<sup>1,3</sup>

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### Authors' response:

We thank Dr. Cruysberg for his interesting comments on our paper. His point is well taken that Marfan's syndrome and homocystinuria can often have similar clinical presentations. This can be a source of confusion for the clinician.

Indeed, homocystinuria has been identified as an etiology for central retinal artery occlusion.<sup>5,7</sup> It is also clearly documented that homocystinuria predisposes to premature atherosclerosis and thromboembolic phenomenon.<sup>1,2,4</sup> We are unaware that any association between homocystinuria and carotid artery dissection has been reported in the medical literature. Specifically, in our case, the magnetic resonance angiogram did not demonstrate any aneurysmal dilation of the carotid artery suggestive of homocystinuria; therefore, at that point, testing for this condition was not indicated.<sup>3,6</sup> The association between connective tissue disorders and arterial dissections is well recognized and, in our patient, attention was focused on identifying a known etiology for her current carotid dissection once that diagnosis was established.

We do agree with Dr. Cruysberg that consideration must be given to homocystinuria as a possible cause for recurrent vascular events in both pediatric and elderly populations.

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