

exists about its metabolism and effects on development.³ Given the rarity of its use in children, performing a randomised control trial is unrealistic, but an alternative method would be to establish a central database that allows clinicians who use anti-vascular endothelial growth factor therapy in children to report results and complications.

Conflict of interest

The authors declare no conflict of interest.

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Sir,
Methylmalonic aciduria and homocystinuria-associated maculopathy

Methylmalonic aciduria and homocystinuria is a rare inherited disorder resulting from impaired conversion of dietary vitamin B12 to its metabolically active forms, and has been associated with retinal findings. To the best of our knowledge, we report the first fluorescein angiographic study of this rare infantile maculopathy.

Case report

A 2-year-old boy was reported to develop nystagmus at 5 months of age. He was born without complication at 40 weeks gestation and weighed 6 pounds 11 ounces at birth. At the age of 1 month, he was evaluated for failure to thrive and found to have elevated urine levels of methylmalonic acid and homocysteine, and was diagnosed with methylmalonic aciduria and homocystinuria. He has since received regular intramuscular injections of hydroxycobalamin and betaine, and been maintained on a special diet.

Ophthalmic examination revealed the ability to fix and follow objects, and the presence of horizontal jerk nystagmus. Fundoscopic examination showed well-circumscribed, round, and relatively flat yellow lesions in both maculae, which were vitelliform in appearance. The lesion in the right eye appeared to be larger than the one in the left eye, which was ring-like, suggesting an early stage of development (Figure 1). Fluorescein angiography was performed under anaesthesia using the RetCam (Clarity Medical Systems, Pleasanton, CA, USA), and demonstrated normal vascular filling without evidence of leakage or staining of the vessels. The macular lesions were hyperfluorescent initially and stained in the late frames (Figure 2).

Comment

Methylmalonic aciduria with homocystinuria may be associated with ocular and other systemic findings.^{1–5} Treatment involving increased levels of methionine may restore normal rod photoreceptor sensitivity; however, this has not been shown to reverse the maculopathy or fully rescue retinal responses.⁴

Ophthalmological referral is critical for these patients to ensure that their maximal visual potential is achieved. The maculopathy, as in this case, often bears resemblance to early stages of Best vitelliform macular dystrophy on fundoscopic examination. It appears from our experience in this case that the fluorescein angiographic findings are similar as well. It may be speculated that, similar to Best disease, the yellow material represents lipofuscin accumulation, in this case secondary to photoreceptor disruption from low methionine levels with accompanying retinal pigment epithelium degeneration. Although photoreceptor damage from methionine deficiency has not been established, it is known that taurine deficiency (of which methionine is a precursor) results in photoreceptor degradation.⁶ In cases that present early to the ophthalmologist, it is important that methylmalonic

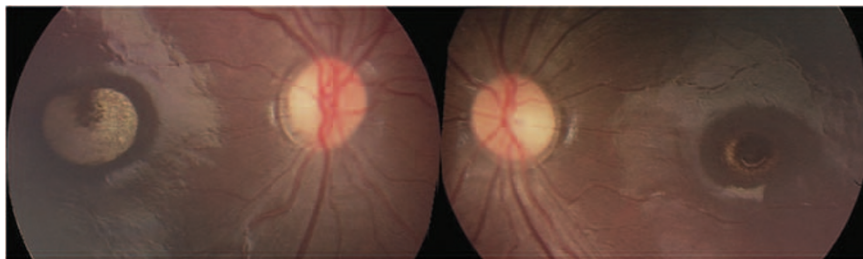


Figure 1 Maculopathy of the right eye (left panel) and left eye (right panel).

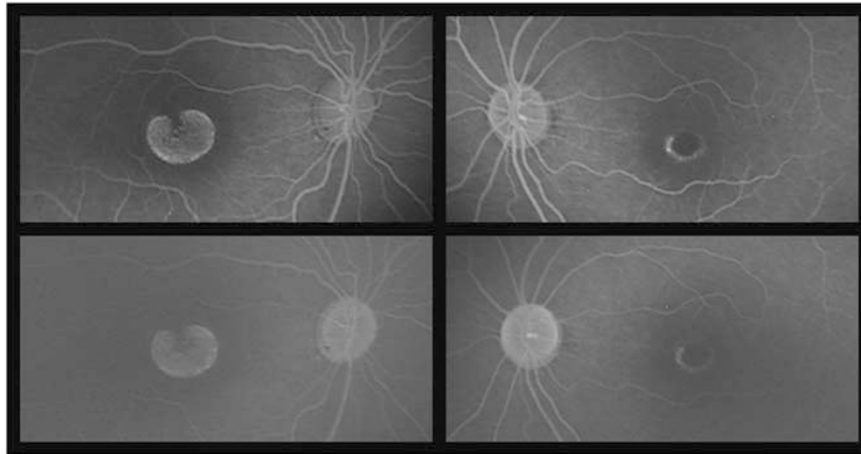


Figure 2 Fundus fluorescein angiogram of the right eye (upper left panel) and left eye (upper right panel) demonstrating early hyperfluorescence of macula lesion, and late staining of lesions in the right (lower left panel) and left (lower right panel) eyes.

aciduria and homocystinuria be remembered in the differential diagnosis of infantile maculopathy.

Conflict of interest

Dr Rosen is a member of the scientific advisor board of Clarity Medical Systems. The remaining authors declare no conflict of interest.

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Sir,
Morning glory with serous macular detachment responds to oral acetazolamide

We report a case of morning glory optic disc with serous macular detachment in the left eye. The patient was treated with oral acetazolamide and followed up for a period of 3 years.

Case report

A 27-year-old lady presented in 2006 with onset of distortion in her left eye. Her visual acuity was 6/36 with a morning glory disc anomaly (Figure 1) and serous macular detachment (Figure 3). The central foveal thickness (CFT) was 928 μm in the left eye at baseline. With 1 month of oral slow-release acetazolamide 250 mg (Diamox) twice a day, her visual acuity improved to 6/18 and CFT was reduced to 553 μm . The dosage of acetazolamide was gradually tapered to 62.5 mg once a day over a period of 7 months. Her visual acuity improved to 6/9 in the left eye and CFT reduced

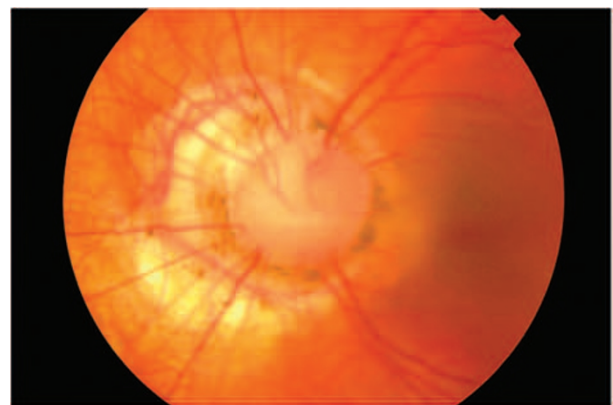


Figure 1 Left eye morning glory optic disc anomaly.