Molecular Genetics and Metabolism Reports 1 (2014) 60

Contents lists available at ScienceDirect

Molecular Genetics and Metabolism Reports

journal homepage: http://www.journals.elsevier.com/molecular-genetics-and-metabolism-reports/

Letter to the Editor

The value of neuroimaging in the assessment and follow-up of early-onset methylmalonic aciduria and homocystinuria ☆

Sir,

In their prospective neurodevelopmental evaluation of children with combined methylmalonic acidemia and homocystinuria, cobalamin C type (MMAHCC), Weisfeld-Adams et al. [1] obtained a magnetic resonance imaging and spectroscopy (MRI/MRS) of the brain. While the authors report a range of MRI brain abnormalities similar to previously published series, MR spectra were found to be normal in all subjects. In a 2-month old patient with MMAHCC seen in our Centre, MRS showed a reduction in the main metabolite peaks in frontal white matter, with N-acetylaspartate peak and N-acetylaspartate/creatine ratio reduction. These findings, which indicate diffuse neuronal damage, were confirmed at the MRS imaging follow up, two years later. Abnormal MRS brain imaging in patients with MMAHCC has been reported by other authors and are likely due to remethylation defects [2,3]. Further studies which are explicitly addressed at dynamic evaluation of MRS in children with MMAHCC may better explain the role of impaired methyl group metabolism in disease-related complications.

References

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7 November 2013

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2214-4269/$ – see front matter
http://dx.doi.org/10.1016/j.ymgmr.2013.11.003