Letter to Editor

The Cerebrospinal Fluid Level of 5-Methylterahydrofolate in a Japanese Boy with Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum

To the Editor

Mercimek-Mahmutoglu and Stockler-Ipsiroglu (2007) reported the decreased level of 5-methylterahydrofolate (5-MTHF) in the cerebrospinal fluid (CSF) of a patient with hypomyelination with atrophy of the basal ganglia and cerebellum (H-ABC). They suggested the relationship between H-ABC and cerebral folate deficiency (CFD), and showed the efficacy of the treatment with folic acid on their patient.

We have also considered the possibility if our patient with H-ABC (Wakusawa et al. 2006; van der Knaap et al. 2007) had CFD. Since 5-MTHF could not be measured in any laboratory in Japan, we measured 5-MTHF in the CSF of our patient in Zürich, Switzerland. It was revealed that the level of 5-MTHF of our patient was not decreased; the level of 5-MTHF of our patient was 86.2 nmol/l (age matched normal value: 41-117 nmol/l). We have therefore concluded that there is a low possibility of having CFD in our patient.

H-ABC is a syndrome defined by the findings of magnetic resonance imaging: hypomyelination of cerebral cortex, atrophy of the basal ganglia, and atrophy of cerebellum (van der Knaap et al. 2002). However, the pathophysiology of H-ABC is still unknown. Our finding suggests that H-ABC is a symptomatic representation of several diseases including CFD. More patients are needed for understanding the pathophysiology of H-ABC.

Acknowledgments

We thank Dr. Mercimek-Mahmutoglu for his thoughtful suggestions and detailed information about 5MTHF.

Keisuke Wakusawa, Mitsugu Uematsu, Shigeru Tsuchiya
Department of Pediatrics, Tohoku University School of Medicine, Sendai, Japan
Kazuhiro Haginoya
Department of Pediatric Neurology, Takuto Rehabilitation Center for Children, Sendai, Japan
Nenad Blau
Division of Clinical Chemistry and Biochemistry, University Children’s Hospital, Zürich, Switzerland

References


Correspondence: Keisuke Wakusawa, M.D., Department of Pediatrics, Tohoku University School of Medicine, 1-1 Seiryo-machi, Aoba-ku, Sendai 980-8574, Japan.
e-mail: k-wakusawa@nifty.com