Brain & Development 42 (2020) 626

BRAIN & DEVELOPMENT
Official Journal of the Japanese Society of Child Neurology

www.elsevier.com/locate/braindev

Letter to the Editor

Multiple cerebral cysts are another possible feature of Jacobsen syndrome

Dear Editor:

We have read with great interest the case report published by Fujino et al., "White matter abnormality in Jacobsen syndrome assessed by serial MRI" [1], which used serial magnetic resonance imaging (MRI) data of a child with Jacobsen syndrome to report that white matter abnormality (WMA) and myelination improved with age. We would like to share an additional feature of such cases based on our experience in our hospital.

A Japanese boy was born at 41 gestational weeks without asphyxia. He weighed 2644 g and measured 47.5 cm long. His occipitofrontal circumference measured 34.5 cm. He had dysmorphological features and thrombocytopenia, both of which suggested Jacobsen syndrome. He had mitral and aortic valve stenosis and left ventricular hypoplasia. His cerebral MRI at 10 months of age revealed WMA and myelination delay, hypoplasia of the corpus callosum, increased cortical sulci at the frontotemporal lobes, and widening of frontal horns of the lateral ventricles (Supplemental Fig. 1). In addition, the putamina contained heterogeneous nodular-cystic lesions. At 4 years and 8 months of age, the myelinization was terminated but bilateral, nodular-cystic lesions remained in the putamina. Additionally, periventricular multiple cysts appeared. The Enoji Developmental Test showed that his developmental quotient was 22 at the age of 4.5 years [2]. His karyotype was 46,XY,del(11)(q23.3q25),arr[hg19]11q23.3q25 (119,484,933–134,938,470)×1 by G-banding karyotyping, fluorescence in situ hybridisation (FISH) analysis and chromosomal microarray analysis (CytoScan HD array, Affymetrix®).

Fujino et al. speculated that WMA in their patient with Jacobsen syndrome resulted from a deletion in the 11q terminal region of *HEPACAM/GlialCAM*, a known causative gene for megalencephalic leukoencephalopathy with subcortical cysts type 2B (MLC2B).

Although there is no report on the presence of multiple cerebral cysts in Jacobsen syndrome, we speculate they could be an additional feature associated with the disease [1,3].

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.braindev.2020.06.019.

References

[1] Fujino S, Yoshihashi H, Takeda R, Ihara S, Miyama S. White matter abnormality in Jacobsen syndrome assessed by serial MRI. Brain Dev 2020;42:621–5.
[2] Enjoji M, Yanai N. Analytic test for development in infancy and childhood. Pediatr Int 1961;4:2–6.
[3] Yamamoto T, Shimada S, Shimojima K, Sangu N, Ninomiya S, Kubota M. Leukoencephalopathy associated with 11q24 deletion involving the gene encoding hepatic and glial cell adhesion molecule in two patients. Eur J Med Genet 2015;58:492–6.

Yuri Dowa *
Takashi Shiihara
Department of Neurology, Gunma Children's Medical Center, Gunma, Japan
* Address: Department of Neurology, Gunma Children's Medical Center, 779 Shimohakoda, Hokkitsu-machi, Shibukawa, Gunma 377-8577, Japan.
E-mail address: lily-rabbit@umin.ac.jp

https://doi.org/10.1016/j.braindev.2020.06.019