Teaching *NeuroImages*: Resolution of MRI abnormalities in megalencephalic leukoencephalopathy with subcortical cysts

Scott Otallah, MD
Julie A. Matsumoto, MD
Howard P. Goodkin,
MD, PhD

Correspondence to
Dr. Otallah:
sio2m@hscmail.mcc.virginia.edu

Figure Resolution of MRI abnormalities in remitting megalencephalic leukoencephalopathy with subcortical cysts between ages 10 months and 6 years

(A-C) Age 10 months. (D-F) Age 6 years. (A) T2: Diffuse confluent cerebral hemispheric white matter T2 hyperintensity with associated white matter swelling is consistent with white matter edema. (B) Fluid-attenuated inversion recovery (FLAIR): Bilateral anterior temporal subcortical cystic changes. (C) T1: Profound cerebral white matter hypointensity including the subcortical U-fibers. (D-F) T2, FLAIR, T1: Nearly complete resolution of previously severe white matter abnormalities.

A boy was diagnosed at age 10 months with megalencephalic leukoencephalopathy with subcortical cysts (MLC) based on progressive macrocephaly and characteristic MRI findings (figure, A–C).¹ Over subsequent years, initial motor delays resolved and neurobehavioral difficulties were mild. On repeat MRI at 6 years (figure, D–F), only small areas of frontal and temporal white matter signal alteration remained, consistent with remitting MLC (MLC2b). MLC2b is associated with heterozygous mutations in *HEPACAM* with autosomal dominant inheritance. Often, one parent has macrocephaly.¹ Unlike the more common phenotype seen with *MLC1* mutations, MLC2b patients demonstrate remarkable MRI improvement and have a better clinical prognosis.²

AUTHOR CONTRIBUTIONS

Dr. Otallah is the corresponding author. He wrote the draft and completed final editing. Dr. Matsumoto contributed to neuroradiologic

aspects of this article including formatting of the figure and other professional comments. Dr. Goodkin assisted in drafting and preparing the manuscript for publication.

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From the Department of Neurology (S.O.), University of Virginia Health System, Charlottesville; and the Departments of Radiology and Medical Imaging (J.A.M.) and Neurology and Pediatrics (H.P.G.), University of Virginia, Charlottesville.

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