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

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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REFERENCES

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