Teaching NeuroImages: A child with macrocephaly and psychomotor development delay

Hajar Rhouda, MD, Latifa Chat, PhD, and Yamna Kriouile, PhD

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Correspondence

Dr. Rhouda
hajar.rhouda@gmail.com

Figure Brain MRI

(A) Parasagittal T1-weighted image shows diffuse cerebral white matter hypointensity with temporal subcortical cyst. (B) Axial T2-weighted image shows increased characteristic diffuse supratentorial white matter signal intensity. (C) Coronal fluid-attenuation inversion recovery image shows bilateral symmetrical anterior temporal lobe subcortical cysts.

A 12-month-old boy, born to consanguineous parents, presented with developmental delay and macrocephaly. MRI revealed diffuse cerebral white matter T2 hyperintensity and temporal subcortical cysts (figure), leading to a diagnosis of megalencephalic leukoencephalopathy with subcortical cysts (MLC). Diffuse white matter changes and temporal subcortical cysts are hallmarks of this rare disorder^{1,2}; these features, in addition to the lack of basal ganglia involvement, distinguish MLC from other leukodystrophies.¹ MLC is caused by *MLC1* or *GLIALCAM* mutations leading to a defect of brain ion and water homeostasis.² Clinical features include macrocephaly, mild developmental delay, and easily controlled seizures.¹

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Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

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Appendix Authors

Name	Location	Role	Contribution
Hajar Rhouda, MD	Unit of Neuropaediatrics and Metabolic Diseases, Rabat Children's Hospital; Mohamed V University, Rabat, Morocco	Author	Acquisition of data, drafting of manuscript, preparation of publication
Latifa Chat, PhD	Radiology Department, Rabat Children's Hospital; Mohamed V University, Rabat, Morocco	Author	Interpretation of data
Yamna Kriouile, PhD	Unit of Neuropaediatrics and Metabolic Diseases, Rabat Children's Hospital; Mohamed V University, Rabat, Morocco	Author	Acquisition of data, critical revision

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