Teaching NeuroImages: Imaging in metabolic leukoencephalopathy, L-2-hydroxyglutaric aciduria

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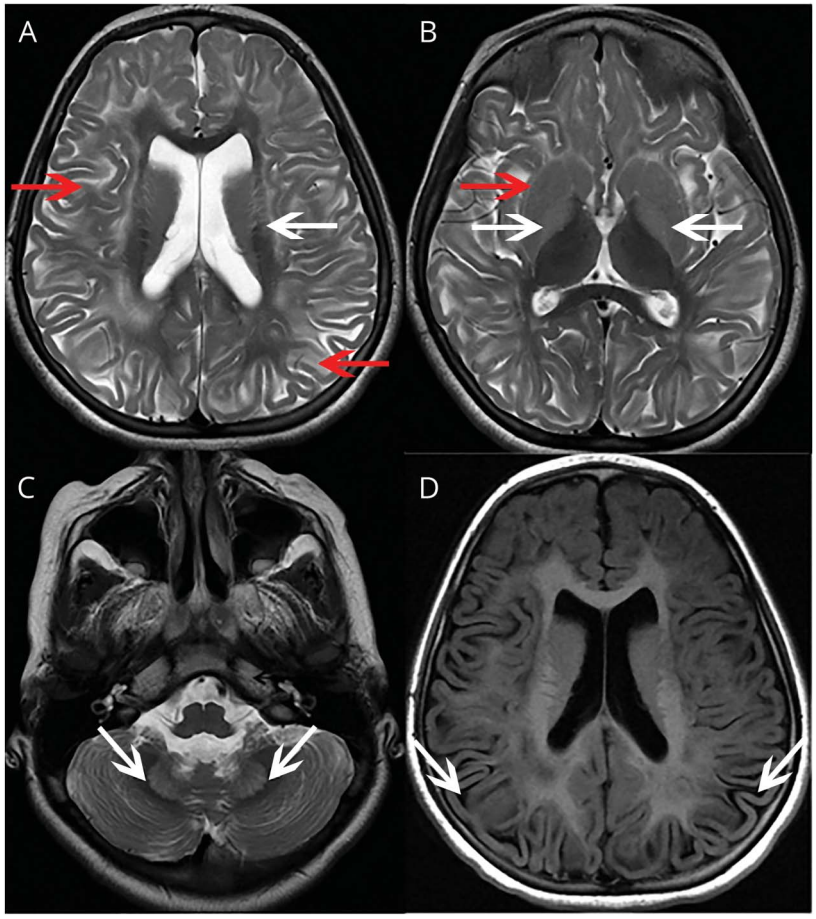
Figure Leukoencephalopathy with basal ganglia hyperintensity and outer rim sign of the putamen



Axial T2-weighted MRIs show (A) bilateral symmetrical white matter (WM) hyperintensity (red arrows) in the centripetal pattern involving subcortical and deep WM, with sparing of periventricular WM (white arrows). (B) Hyperintense basal ganglia (white arrows) with more hyperintensity along the outer rim of the putamen (outer rim sign, red arrow). (C) Hyperintense dentate nucleus (white arrows). (D) The fluid-attenuated inversion recovery image shows rarefaction (white arrows).

A 9-year-old girl presented to us with insidious onset difficulty in walking, recurrent falls, anxiety, and poor scholastic performance from age 5 years. Her MRI findings (figure) were classic of L-2-hydroxyglutaric aciduria (L2-HGA). However, similar findings are also seen in Leigh syndrome, 3-hydroxy-3-methylglutaric aciduria, and succinic semialdehyde dehydrogenase deficiency. Urine analysis showed elevated levels of L-2-hydroxyglutaric acid, which confirmed the diagnosis. L2-HGA is a rare autosomal recessive metabolic disorder due to deficiency of L-2-hydroxyglutarate dehydrogenase.[1] The characteristic MRI findings in this condition are

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bilateral symmetrical basal ganglia and dentate nuclei involvement along with subcortical white matter abnormality.[2]

Author contributions

S. Sundaram: biochemical and clinical workup of the patient. V. S: reporting of MRI and differential diagnosis. S. Sivadasan: manuscript preparation and critical revision of the manuscript.

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Disclosure

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References

1. Kranendijk M, Struys EA, Salomons GS, Van der Knaap MS, Jakobs C. Progress in understanding 2-hydroxyglutaric acidurias. J Inherit Metab Dis 2012;35:571–587.
2. Steenweg ME, Salomons GS, Yapici Z, et al. L-2-hydroxyglutaric aciduria: pattern of MR imaging abnormalities in 56 patients. Radiology 2009;251:856–865.

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