

Teaching NeuroImages: Distinctive imaging in a paucisymptomatic child with leukodystrophy

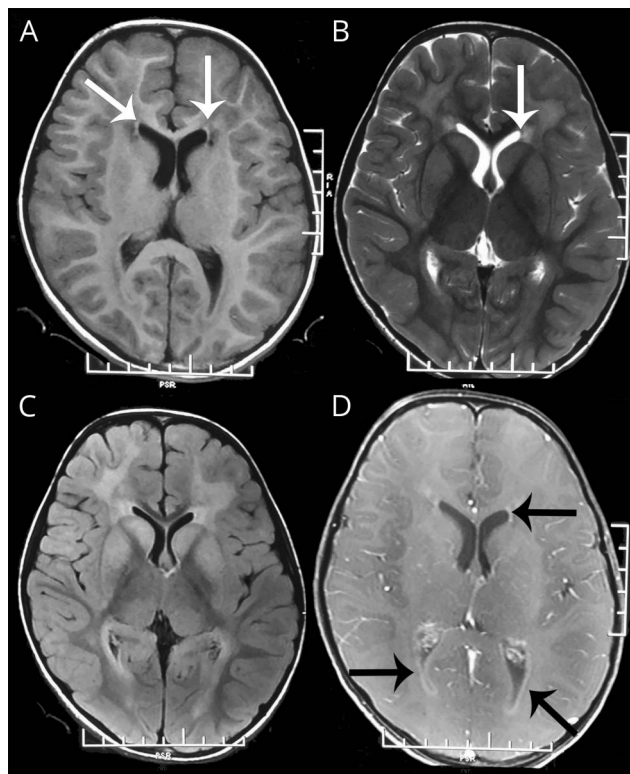
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Figure MRI brain



(A) Axial T1-weighted image shows thin periventricular hyperintense rim (arrows), which was hypointense on (B) T2-weighted image (arrow). (B) Confluent T2-weighted and (C) fluid-attenuated inversion recovery hyperintensities in bilateral periventricular deep and subcortical cerebral white matter (frontal dominant), striatum, and thalami. (D) Postgadolinium axial section shows a thin rim of ependymal enhancement predominantly in trigone and minimally in the frontal horn of lateral ventricles (arrows).

An 8-year-boy, born to nonconsanguineous parents, presented with a history of 2 unprovoked generalized seizures in the preceding 5 years. The examination was unremarkable. An awake EEG showed occasional bifrontal sharps. Based on the MRI brain (figure, A–D) and the detection of a pathogenic variant¹ (c.262C>T[p.Arg88Cys], exon 1) on *GFAP* gene sequencing, a diagnosis of Alexander disease was confirmed. The child remained asymptomatic during a 3-year follow-up period.

Alexander disease is an astrogliaopathy, characterized by megalencephaly, pyramidal signs, progressive psychomotor retardation, ataxia, pseudobulbar signs, and seizures.² However, a rare

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patient may be paucisymptomatic for several years.² The imaging findings are distinctive and allow for directed genetic testing and confirmation.

Author contributions

I.K.S.: patient management, literature review, initial draft manuscript preparation. L.S.: concept and design of the study, analysis of the radiologic data, critical review of manuscript, final approval of the version to be published. A. Kasinathan: patient management, literature review, initial draft manuscript preparation. A. Kaur: patient management, literature review, initial draft manuscript preparation. N.S.: clinician-in-charge, concept and design of the study, critical review of manuscript

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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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