

Teaching NeuroImages: An imaging clue for treatable early childhood-onset dystonia: Manganism

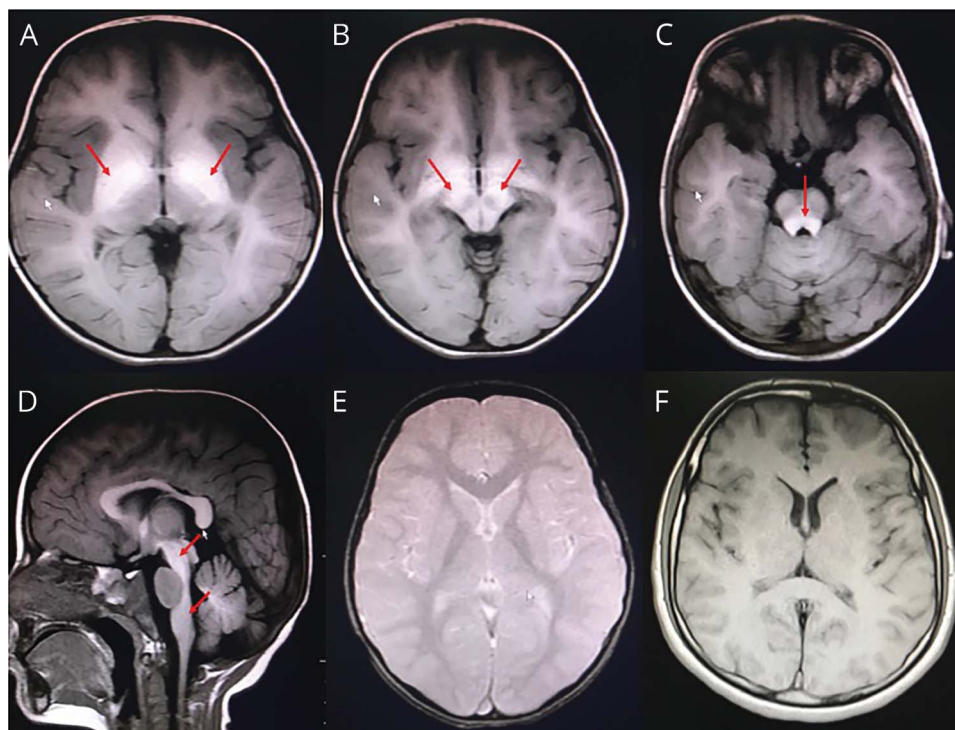
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Figure Axial T1-weighted image, sagittal T1-weighted image, and T2* gradient-recalled echo (GRE) axial image



Axial T1-weighted image shows bright signal intensity (T1 shortening) in the (A) Globus pallidus, (B) Substantia nigra, and (C) Tectum of pons. (D) Sagittal T1-weighted image shows bright signal intensity in the dorsal brainstem. (E) T2* GRE axial image at the level of basal ganglia shows no evidence of blooming (unlike iron or calcium deposition, manganese deposition will not cause blooming on T2* GRE or susceptibility-weighted imaging). (F) Normal axial T1-weighted image for comparison at the level of basal ganglia shows no signal changes.

A 3-year-old boy, born of consanguineous parentage, presented with recurrent falls and toe-walking from 2 years. Examination showed microcephaly (45 cm; <-3 Z score, WHO), dysarthria, and bilateral foot dystonia. Neuroimaging revealed multiple areas of T1-weighted hyperintensities, including in the basal ganglia and dorsal brainstem (figure). Serum manganese was elevated at 186 mg/L (normal 5–15 mg/L) with hemoglobin of 14.5 g/dL (normal 11.5–15.5 g/dL). Next-generation sequencing revealed novel homozygous single base pair insertion c.18_19insT (p.Lys7Ter) in exon 1 of the *SLC30A10* gene.

The *SLC30A10* gene is a cell surface localized manganese efflux transporter and loss of function mutations lead to accumulation of manganese in liver and brain.¹ It is inherited autosomal recessively and manifests in childhood (2–15 years) with 4-limb dystonia, dysarthria, polycythemia,

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hepatic cirrhosis, and characteristic neuroimaging.² EDTA chelation and iron supplementation might be beneficial.²

Author contributions

Hansashree Padmanabha: study concept and design, first draft, literature review, critical review of manuscript for intellectual content. Savita Krishnamurthy: study concept and design, first draft, literature review. Sharath Kumar GG: study concept and design, neuroimaging discussion. Indumathi Chikkanayakana: study concept and design, first draft, literature review. Aruna Sethuraman: study concept and design, first draft, literature review. Thomas Mathew: study concept and design, first draft, literature review.

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