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 $\text{SLC30A10}$ gene.

The $\text{SLC30A10}$ 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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**Figure** Axial T1-weighted image, sagittal T1-weighted image, and T2* gradient-recalled echo (GRE) axial image

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

**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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