“An Imaging Clue for Treatable Early Childhood-Onset Dystonia - Manganism”

Teaching Neuro*Images*

Neurology

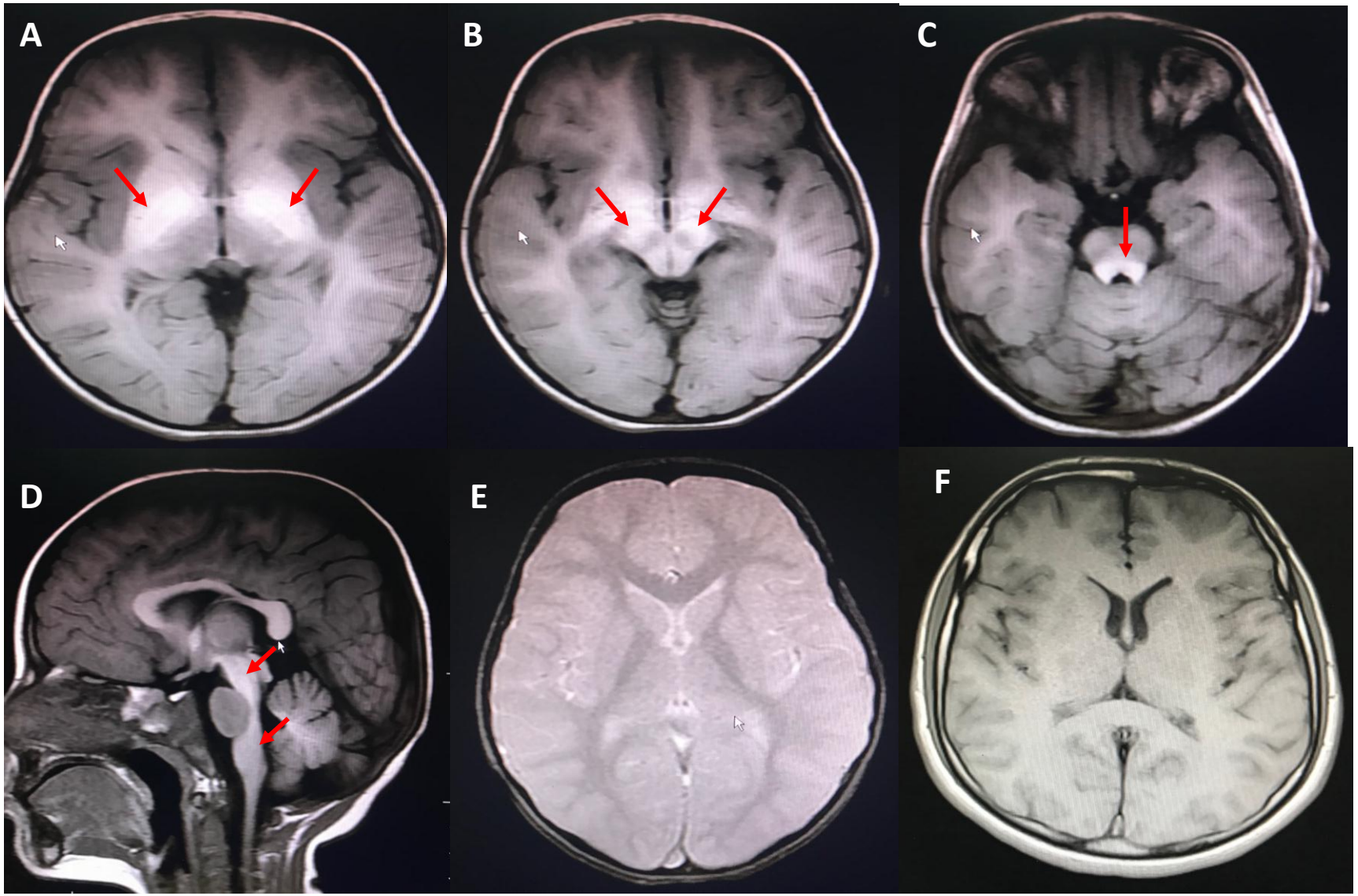
Resident and Fellow Section

AMERICAN ACADEMY OF NEUROLOGY®

Vignette

- **A 3-year-old boy, first born of third-degree consanguineous parentage had presented with recurrent falls and toe-walking from 2-years.**
- **On examination, he had microcephaly(45cm; <-3Z score, WHO), dysarthria and bilateral foot dystonia.**
- **Neuroimaging revealed multiple areas of T1-weighted hyper-intensities including in the basal ganglia and dorsal brainstem(Figure 1)**
- **Serum-manganese was elevated 186 microgram/l(5-15) and hemoglobin increased gradually from 11g/dl to 14.5g/dl(11.5-15.5g/dl).**
- **Next generation sequencing revealed previously unreported homozygous single base pair insertion c.18_19insT(p.Lys7Ter) in exon-1 of *SLC30A10* gene.**

Imaging



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- **Inherited hypermanganesemia are autosomal recessively inherited due to mutations in *SLC30A10, SLC39A14* and *SLC39A8*[1].**
- ***SLC30A10* gene is a cell surface localized manganese efflux transporter and loss of function mutations leads to accumulation of manganese predominantly in liver and brain[1].**
- **Homozygous mutations in *SLC30A10* gene manifests in childhood(2-15 years) with four-limb dystonia, dysarthria, polycythemia, hepatic cirrhosis and characteristic neuroimaging[2].**
- **Chelation with EDTA and iron supplementation might be beneficial[2].**

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