Teaching NeuroImages:

Distinct neuroimaging features of fucosidosis

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Figure Classic MRI findings in fucosidosis

T1-weighted axial (A), T2-weighted axial (B), and coronal (C) MRI of the brain. Bilateral globi pallidi (arrows) show hyperintensity on T1 and marked hypointensity on T2-weighted images. In addition, there is diffuse symmetric hyperintensity of bilateral subcortical and deep white matter on T2-weighted images with normal appearance on T1-weighted image consistent with hypomyelination.

A 5-year-old girl, the product of nonconsanguineous marriage, presented with early-onset intellectual disability and autistic features. There was no regression, seizures, or vision or hearing impairment. She had normal head circumference, coarse facies, angiokeratomas, and lower limb rigidity. She had no cherry-red spot or hepatosplenomegaly. Her 2-year-old brother had similar features with right hemidystonia. The MRI brain findings (figure), positive urine oligosaccharides, and absent α -L-fucosidase activity in plasma and leukocytes confirmed the diagnosis of fucosidosis, an autosomal recessive lysosomal storage disorder with poor cognitive outcome.^{1,2} Mucopolysaccharidosis-like phenotype, angiok-

eratomas, oligosacchariduria, and classic MRI findings suggest fucosidosis.

AUTHOR CONTRIBUTIONS

Puneet Jain, Konanki Ramesh, and Akbar Mohamed provided clinical care to the patient and drafted the manuscript. Atin Kumar provided radiologic input. Sheffali Gulati critically reviewed the manuscript.

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