Published Ahead of Print on August 1, 2023 as 10.1212/WNL.0000000000207546

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The Official Journal of the American Academy of Neurology

AMERICAN ACADEMY OF NEUROLOGY®

Neurology Publish Ahead of Print
DOI: 10.1212/WNL.0000000000207546

Teaching NeuroImage: A New Imaging Finding in a Boy With Salla Disease Caused by a Pathogenic Variant in the *SLC17A5* Gene

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Kshitij Mankad: Drafting/revision of the manuscript for content, including medical writing for content; Analysis or interpretation of data
Utkarsh Kabra: Major role in the acquisition of data; Analysis or interpretation of data
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Meenu Bagarhatta: Major role in the acquisition of data; Analysis or interpretation of data
Ashok Gupta: Major role in the acquisition of data; Analysis or interpretation of data

Figure Count:
1

Table Count:
0

Search Terms:
[91] All Genetics, [120] MRI, [155] Leukodystrophies, [313] Basal ganglia

Acknowledgment:

Study Funding:
The authors report no targeted funding.

Disclosure:
The authors report no relevant disclosures.

Preprint DOI:

Received Date:
2022-12-12
Accepted Date:
2023-05-05

Handling Editor Statement:
Submitted and externally peer reviewed. The handling editor was Resident and Fellow Deputy Editor Ariel Lyons-Warren, MD, PhD.

A 7-year-old boy born to non-consanguineous parents presented with developmental delay, dysmorphism, and an ataxic gait. MRI of the brain demonstrated hypomyelinating leukodystrophy (Figure 1A,B), cerebellar atrophy (Figure 1C,D), and thinning of the corpus callosum (Figure 1E). T1 hyperintensities were also seen in the bilateral deep grey nuclei, brainstem, and cerebellum (Figure 1F-H). Genetic testing confirmed a diagnosis of Salla disease (SD) by revealing a likely pathogenic, homozygous missense variation in the *SLC17A5* gene (chr6:g.73644582C>T). Both parents were found to be carriers consistent with autosomal recessive inheritance. Sialic acid storage disease (SASD), is a neurodegenerative lysosomal storage disorder, which can present as a slowly progressive form (Salla disease), a severe fetal-onset form, or as infantile free sialic acid storage disease (ISSD), however, intermediate forms also exist.[1] The differential for symmetrical T1 hyperintensities includes Kernicterus, hypoxic ischemic injury, neurodegeneration with brain iron accumulation, Fabry disease, other lysosomal storage disorders, and Wilson disease which were considered and excluded for this case. T1 hyperintensities have not previously been described in SD.[1] One plausible explanations is the deposition of sialic acid, which is a paramagnetic substance.[2]

WNL-2023-000365_slides --- http://links.lww.com/WNL/D18

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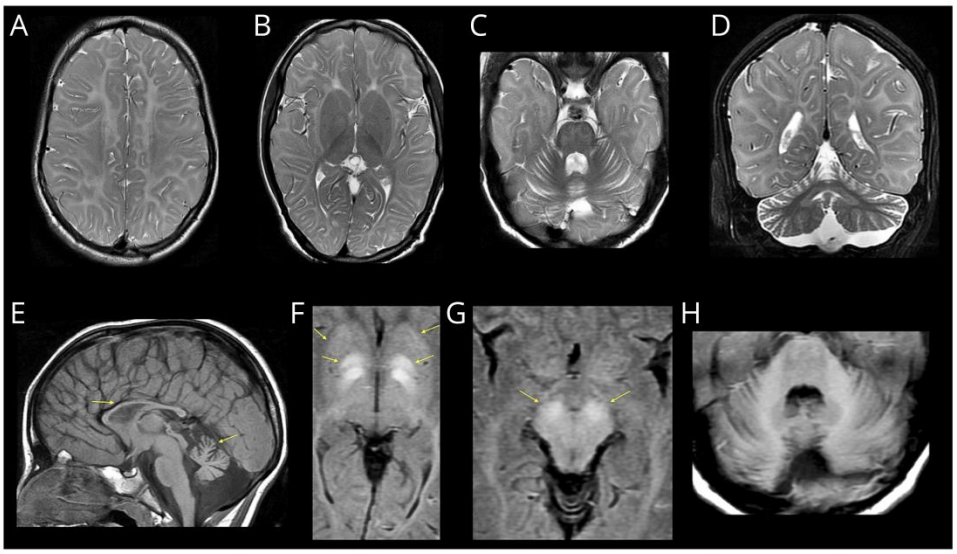
Short Figure Title

MRI Brain Shows Diffuse Hypomyelination with Hyperintensity of Basal Ganglia, Brainstem and Cerebellum

Figure Legend

Figure 1: T2W axial images (A, B) demonstrate diffuse hyperintense hypomyelinating pattern. T2W axial (C), and coronal (D) images demonstrate cerebellar atrophy. T1W sagittal image (E) demonstrates corpus callosum thinning and vermian atrophy (arrows). T1W axial images (F, G, H) demonstrate hyperintensity (arrows) of basal ganglia, brainstem and cerebellum.



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