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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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A 7-year-old boy born to nonconsanguineous parents presented with developmental delay, dysmorphism, and an ataxic gait. An MRI of the brain demonstrated hypomyelinating leukodystrophy (Figure, A and B), cerebellar atrophy (Figure, C and D), and thinning of the corpus callosum (Figure, E). T1 hyperintensities were also seen in the bilateral deep gray nuclei, brainstem, and cerebellum (Figure, F–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 (SD), a severe fetal-onset form, or as infantile free SASD; however, intermediate forms also exist. 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.

Figure MRI of the Brain Shows Diffuse Hypomyelination With Hyperintensity of the Basal Ganglia, Brainstem, and Cerebellum

T2-weighted axial images (A, B) demonstrate diffuse hyperintense hypomyelinating pattern. T2-weighted axial (C) and coronal (D) images demonstrate cerebellar atrophy. T1-weighted sagittal image (E) demonstrates corpus callosum thinning and vermian atrophy (arrows). T1-weighted axial images (F–H) demonstrate hyperintensity (arrows) of the basal ganglia, brainstem, and cerebellum.

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T1 hyperintensities have not previously been described in SD.\(^1\) One plausible explanation is the deposition of sialic acid, which is a paramagnetic substance.\(^2\)

**Author Contributions**

J. Gupta: drafting/revision of the article for content, including medical writing for content; major role in the acquisition of data; study concept or design; and analysis or interpretation of data. N. Reddy: drafting/revision of the article for content, including medical writing for content; analysis or interpretation of data. K. Mankad: drafting/revision of the article for content, including medical writing for content; analysis or interpretation of data. U. Kabra: major role in the acquisition of data; analysis or interpretation of data. A. Bhandari: major role in the acquisition of data; analysis or interpretation of data. M. Bagarhatta: major role in the acquisition of data; analysis or interpretation of data. A. Gupta: major role in the acquisition of data; analysis or interpretation of data.

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**References**


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