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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 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 explanation is the deposition of sialic acid, which is a paramagnetic substance.²

References

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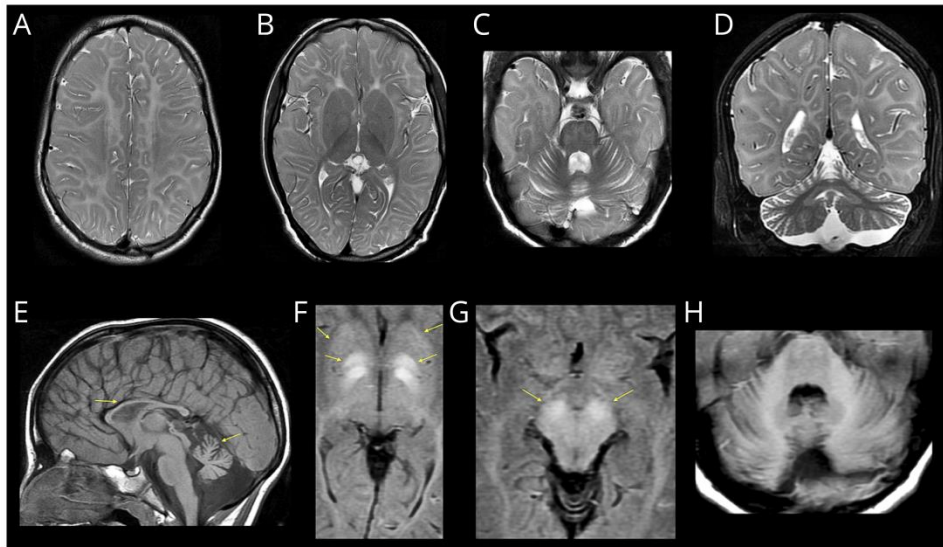
Short Figure Title

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

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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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