Teaching NeuroImages: Symmetrical abnormalities of the globi pallidi in succinic semialdehyde dehydrogenase deficiency

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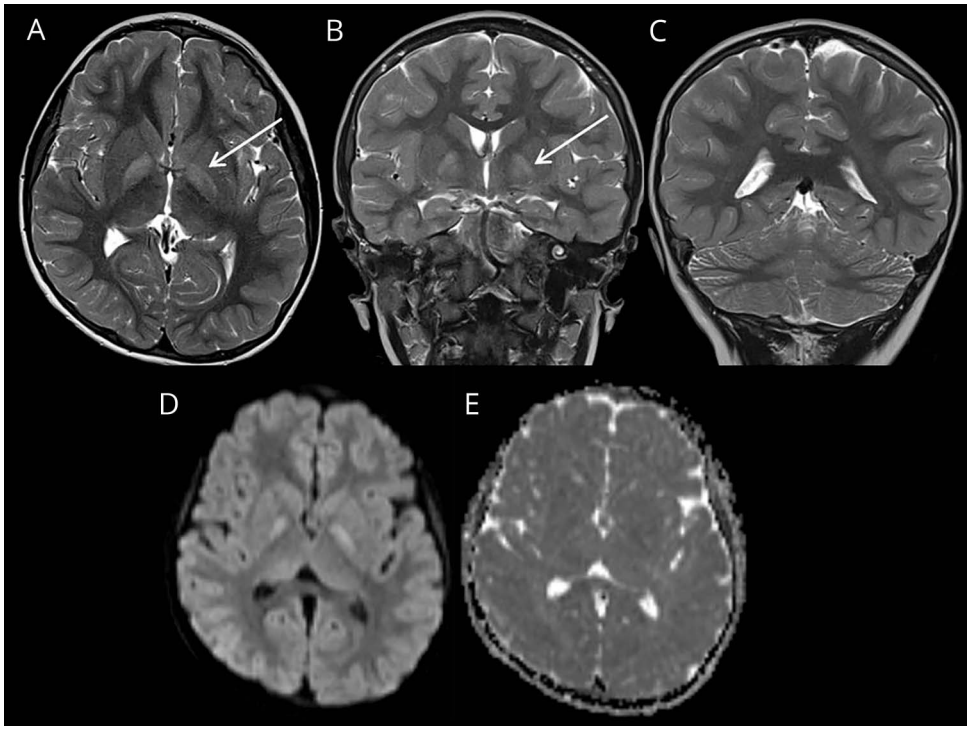
Figure Globi pallidi T2 symmetrical hyperintensity



Axial (A) and coronal T2-weighted imaging (B, C) show symmetrical hyperintensity of the globi pallidi (arrows). Note that both pallidal components (internal and external pallidus) are well discernible. No dentate involvement or cerebellar atrophy is seen. (D) Diffusion-weighted images show pallidal diffusion restriction at high b-values confirmed by apparent diffusion coefficient map (E).

A 4-year-old boy born full term after an uneventful pregnancy and with normal early developmental milestones achievement presented hypotonia, motor coordination disorder, and childhood apraxia of speech. Brain MRI revealed globi pallidi T2 symmetrical signal abnormalities (figure). An extensive diagnostic workup panel was performed to exclude metabolic and acquired conditions.[1,2] Urine organic acid profile showed marked γ-hydroxybutyrate aciduria (222 μg/mg creatinine, normal <5). Genetic analysis revealed 2 compound heterozygous pathogenic mutations in the *ALDH5A1* gene (p.Pro81Ser, maternal origin; p.Arg527Ter, paternal origin) consistent with succinic semialdehyde

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dehydrogenase deficiency suspicion. This rare entity must be suspected in case of T2-weighted MRI symmetrical abnormalities of the globi pallidi.[1,2]

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