

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

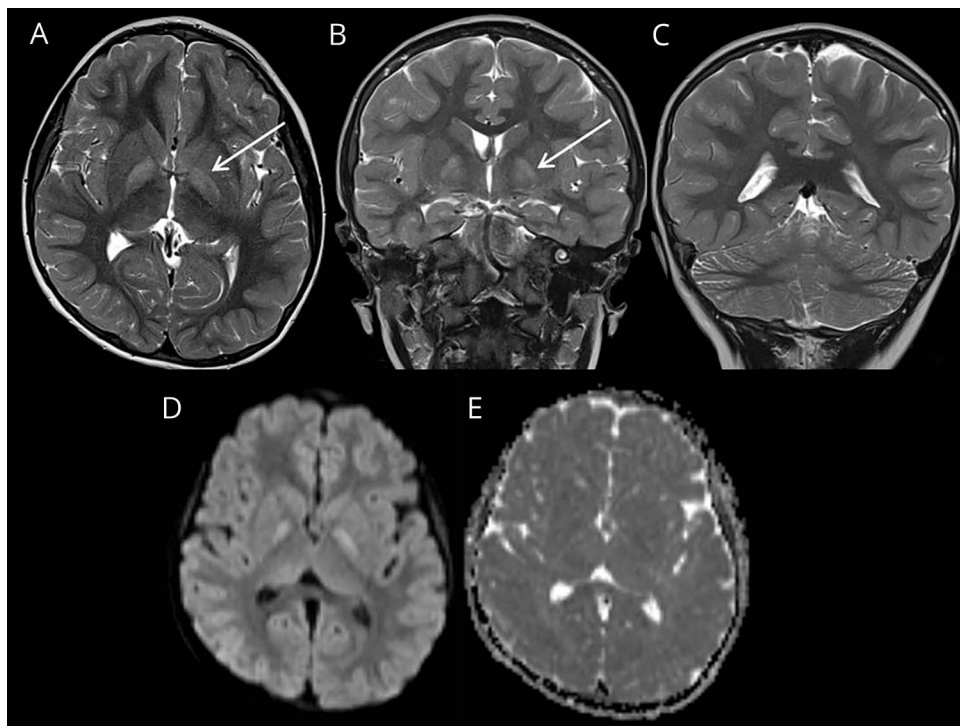
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Figure Globi pallidi T2 symmetrical hyperintensity



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 $\mu\text{g}/\text{mg}$ 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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Disclosure

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Name	Location	Contribution
Silvia Esposito, MD, PhD	Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy	Study design and manuscript writing, care of the patient
Marco Moscatelli, MD	Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy	Interpretation of radiologic images, manuscript revision for intellectual content
Claudio Caccia, PhD	Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy	Manuscript revision for intellectual content, interpretation of metabolic and genetic findings

Appendix (continued)

Name	Location	Contribution
Elisa Granocchio, MD	Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy	Manuscript revision for intellectual content, interpretation of neuropsychological assessment
Chiara Pantaleoni, MD	Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy	Manuscript revision for intellectual content
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Cinzia Gellera, PhD	Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy	Manuscript revision for intellectual content, interpretation of metabolic and genetic findings
Laura Farina, MD	Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan; Fondazione IRCCS Fondazione Santa Lucia, Rome, Italy	Interpretation of radiologic images, manuscript revision for intellectual content

References

1. Barkovich AJ. An approach to MRI of metabolic disorders in children. *J Neuroradiol* 2007;34:75–88.
2. Zuccoli G, Yannes MP, Nardone R, Bailey A, Goldstein A. Bilateral symmetrical basal ganglia and thalamic lesions in children: an update (2015). *Neuroradiology* 2015;57:973–989.

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