Teaching NeuroImages: Early imaging of sulfite oxidase deficiency mimics severe hypoxic ischemic encephalopathy

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Figure MRI and magnetic resonance spectroscopy (MRS) in a 4-day-old term infant

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(A) Axial T2 shows cortical swelling with decreased gray-white differentiation. (B) Axial diffusion-weighted imaging shows abnormal increased signal in the cortex, basal ganglia, thalami, and corpus callosum with (C) associated decreased signal on apparent diffusion coefficient. (D) MRS, obtained over the left basal ganglia, demonstrates an elevated lactate peak.

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A term baby, with uncomplicated pregnancy and delivery, developed intractable multifocal seizures on day 3 of life. EEG showed frequent multifocal electrographic and electroclinical seizures. MRI brain demonstrated diffuse diffusion restriction (figure). Metabolic investigations revealed elevated urine sulfo-cysteine, reduced plasma cysteine and homocysteine, and normal plasma uric acid. CSF lactate was elevated. The diagnosis was consistent with isolated sulfite oxidase deficiency, with normal plasma uric acid differentiating this from molybdenum cofactor deficiency. The patient underwent confirmatory genetic testing. Sulfite oxidase deficiency is an autosomal recessive inherited inborn error of metabolism without treatment.^{1,2} Initially, it may mimic profound hypoxic ischemic encephalopathy.

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Appendix Authors

Name	Location	Contribution
Carly Scramstad, MD	University of Manitoba, Winnipeg	Preparation of case report, drafting and revision for intellectual content
Hayley Moffatt, MD	University of Manitoba, Winnipeg	Preparation of figure, drafting and revision for intellectual content
Mubeen Rafay, MBBS	University of Manitoba, Winnipeg	Conceptualization, drafting and revision for intellectual content

References

1. Claerhout H, Witters P, Regal L, et al. Isolated sulfite oxidase deficiency. *J Inherit Metab Dis* 2018;41:101–108.
2. Relinque B, Bardallo L, Granero M, Jimenez PJ, Luna S. Isolated sulfite oxidase deficiency. *J Neonatal Perinatal Med* Epub 2015 Mar 10.

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