Teaching NeuroImages: Rapidly progressive leukoencephalopathy in mitochondrial complex I deficiency

At age 8 months, an infant girl displayed rapid developmental regression. Family history, birth, and initial development were unremarkable. After hospital admission, cerebral MRI showed bilateral cystic lesions in the centrum semiovale. Follow-up imaging after 3 months demonstrated a dramatic progression in these alterations with demyelination of the supratentorial white matter (figure). Biochemical and genetic analyses confirmed isolated mitochondrial complex I deficiency due to an *NDUFS1* mutation (encoding NADH-dehydrogenase-ubiquinone Fe-S protein 1; see also reference 1, patient 1). Of note, leukoencephalopathy is uncommon in mitochondrial complex I mutations but may be a feature of *NDUFS1* defects.

**AUTHOR CONTRIBUTIONS**

Fabian Baertling: drafting and revising the manuscript for intellectual content. Jörg Schaper: drafting and revising the manuscript for intellectual content. Ertan Mayatepek: drafting and revising the manuscript for intellectual content. Felix Distelmaier: drafting and revising the manuscript for intellectual content.

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REFERENCES


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