Fatty acid 2-hydroxylase deficiency
Clinical features and brain iron accumulation

A 21-year-old woman presented with progressive spastic paraplegia, dysarthria, and strabismus since 7 years of age (video on the Neurology® Web site at Neurology.org). Brain MRI disclosed white matter changes and iron accumulation (figure). Whole exome sequencing detected in fatty acid 2-hydroxylase (FA2H) gene 2 variants never reported: c.169_170insGCGGGCCAGG (p.Asp57Glyfs*66), leading, if translated, to a truncated protein, and c.117C>A (p.Phe39Leu), predicted by computational algorithms to be deleterious.

FA2H deficiency is responsible for SPG35, a rare autosomal recessive complicated hereditary spastic paraplegia.1,2 Strabismus, dysarthria, and spastic paraplegia with brain MRI showing iron accumulation and white matter changes are common in SPG35 and may suggest the diagnosis.1,2 Molecular analysis is necessary to confirm this unusual condition.

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