

Teaching Video NeuroImage: New *STUB1* Variant Causes Chorea, Tremor, Dystonia, Myoclonus, Ataxia, Depression, Cognitive Impairment, Epilepsy, and Superficial Siderosis

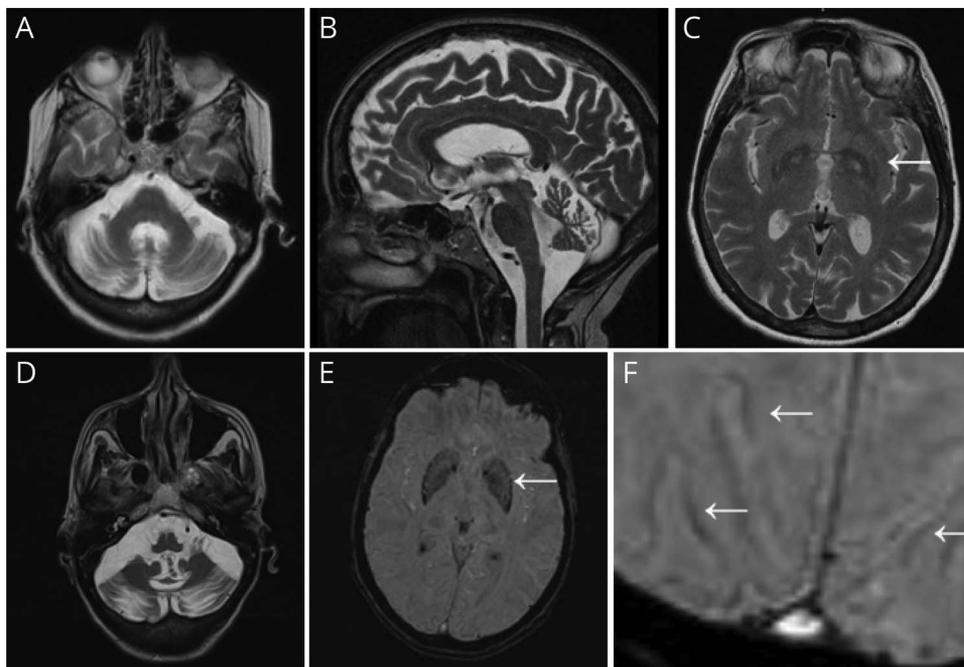
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Figure Imaging Results



(A–C) T2-weighted MRI of the grandmother shows cerebellar atrophy (A, B) and basal ganglia hypointensity (with no correlate in cerebral CT; C; see arrow). (D–F) T2-weighted and susceptibility-weighted imaging (SWI)-weighted MRI of the mother shows cerebellar atrophy (D) and basal ganglia SWI artifacts (E; see arrow) and cortical SWI artifacts highly suggestive of superficial siderosis (F; see arrows).

We present a family with a wide range of movement disorders carrying a new heterozygous splice variant in *STUB1* (NM_005861:c.787-2_787del), probably causing skipping of exon 7, as relevant differential diagnosis to Huntington disease.^{1,2} The grandmother and mother (improved with olanzapine) presented with chorea, dystonia (also platysma), head tremor, orofacial dyskinesias (prior to neuroleptics), saccadic eye movements, ataxia with cerebellar atrophy (Figure), depression, cachexia, and cognitive impairment. The grandmother had epilepsy and the mother had superficial siderosis (Figure). The son presented with depression, left-sided myoclonus and ataxia, and tremor resembling essential tremor, which began in childhood, without alcohol sensitivity. *STUB1* mutation in this family caused chorea, tremor, dystonia,

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myoclonus, ataxia, depression, cognitive impairment, epilepsy, and superficial siderosis (Video 1).

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Carsten Saft, MD	Department of Neurology, Huntington-Centre NRW, St. Josef Hospital	Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Study concept or design; Analysis or interpretation of data
Sabine Skodda, MD	Department of Neurology, Knappschafts Krankenhaus	Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data

Appendix (continued)

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