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

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

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### Appendix Authors

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### References


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