A Family With Ataxia and a Wide Range of Movement Disorders

Teaching Video NeuroImages

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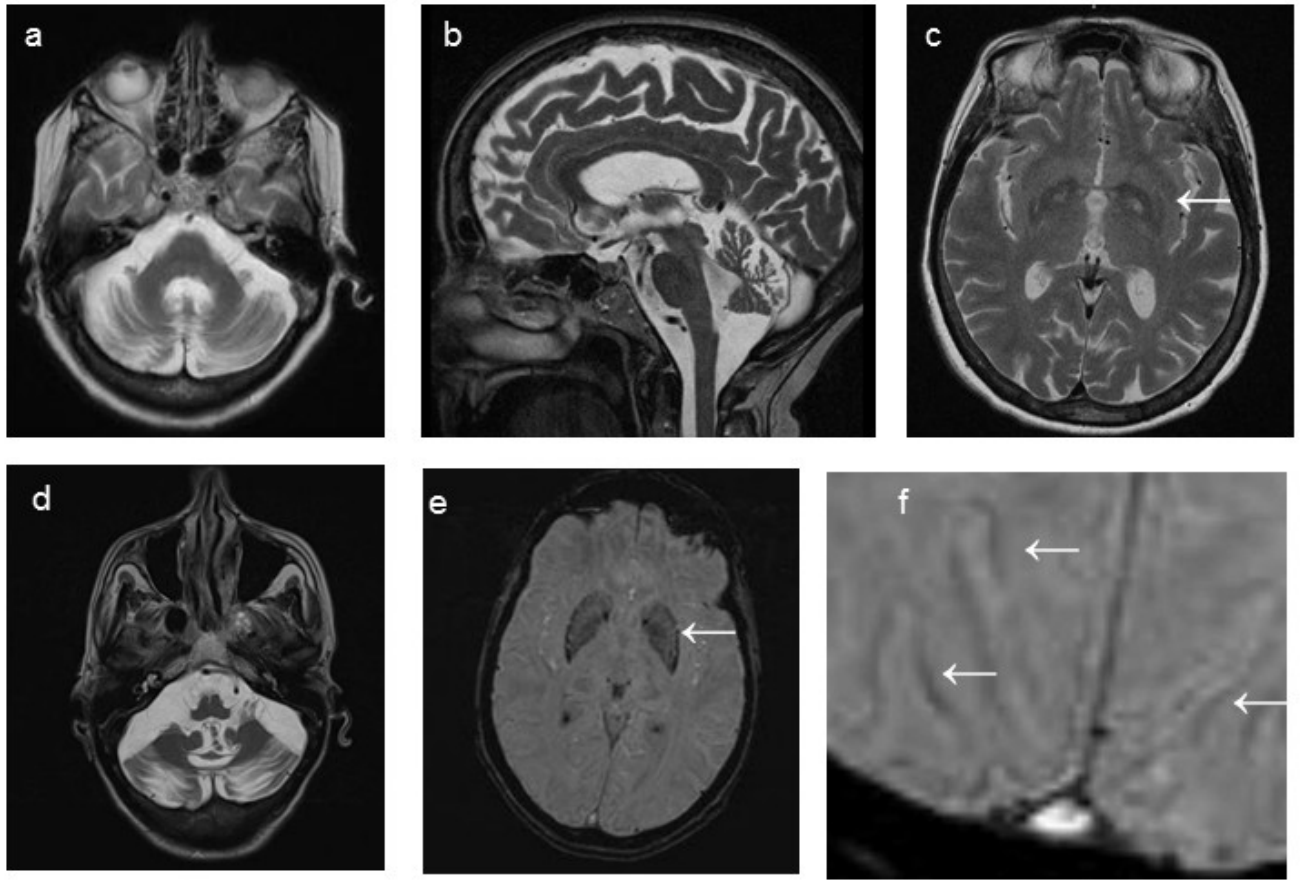
Vignette

- 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, depression, cachexia and cognitive impairment.
- The grandmother suffered from epilepsy and mother from superficial siderosis.
- The son presented with depression, left sided myoclonus, ataxia, and tremor resembling essential tremor which began in childhood without alcohol sensitivity.

Image



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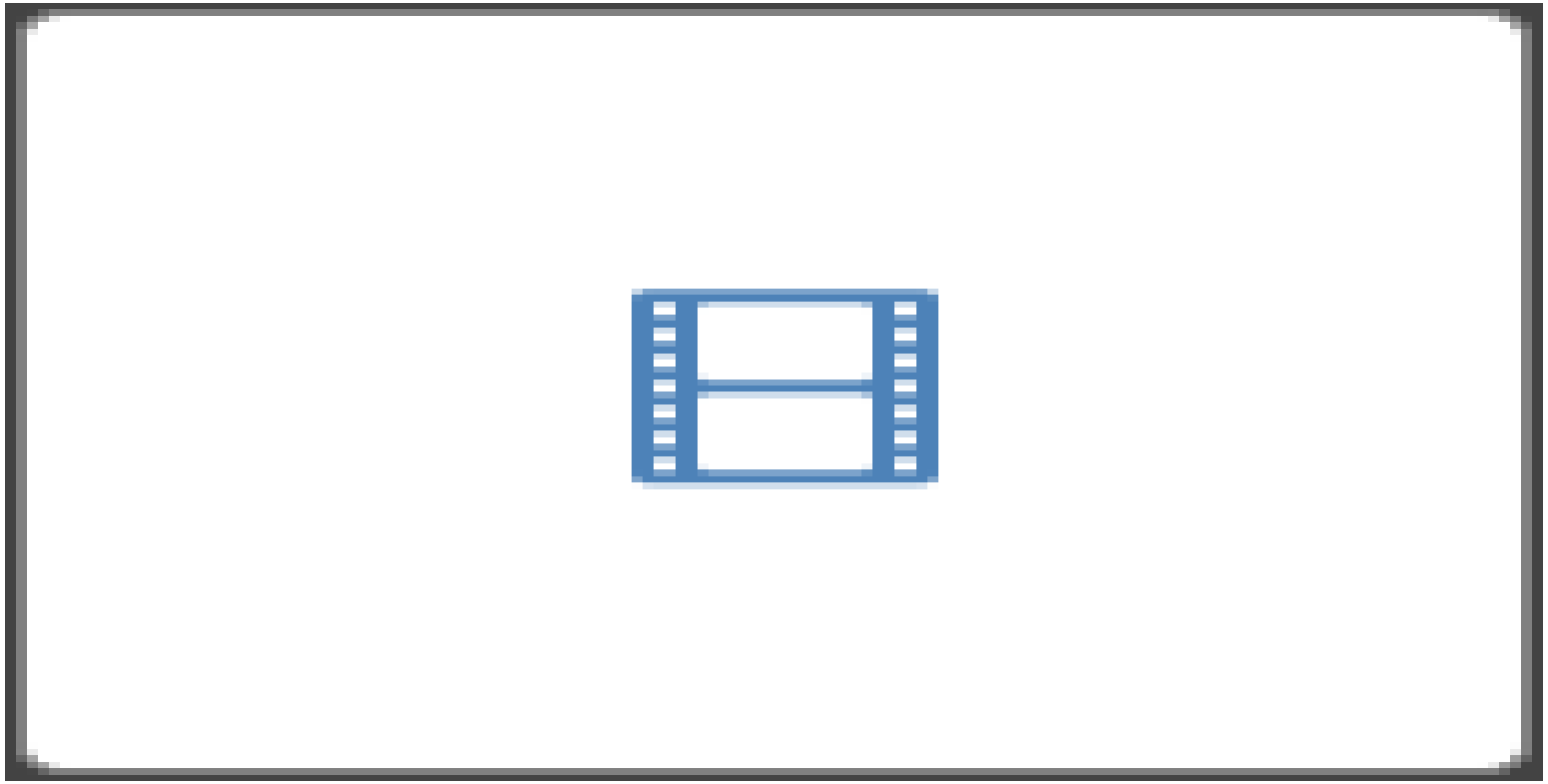
Saft *et al.*

Video



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

- Family history suggests an autosomal dominant inherited disease
- MRI showed cerebellar atrophy, basal ganglia hypointensity and SWI artifacts. NIBA might be discussed.
- Huntington's disease, SCA 1-3, 6, 7, 12, 17, DRPLA, FXTAS and C9orf72 mutation should be excluded
- *STUB1* mutation (SCA48) might caused chorea, tremor, dystonia, myoclonus, ataxia, depression, cognitive impairment or epilepsy
- In this family a new heterozygous splice variant in *STUB1* was identified