Teaching Video NeuroImage: Tremor and Cerebellar Ataxia in a Patient With Fragile X-Associated Tremor/Ataxia Syndrome

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A 63-year-old male presented with a 3-year history of tremors. Neurological examination revealed a head tremor, a lower limb resting tremor, an irregular postural and intention tremor of the distal upper limbs, and cerebellar ataxia (Video 1, http://links.lww.com/WNL/B721). T2-weighted brain MRI showed typical bilateral symmetric hyperintensities in the middle cerebellar peduncles (Figure 1). Genetic testing demonstrated a premutation expansion (90 CGG repeats) of the fragile X mental retardation 1 (FMR1) gene, confirming the diagnosis of fragile X-associated tremor/ataxia syndrome (FXTAS). FXTAS is an X-linked, late-onset, progressive neurodegenerative disease. The characteristic MRI pattern should prompt confirmation of the diagnosis with genetic testing for FMR1 premutations (55-200 CGG repeats). Fragile X syndrome, associated with FMR1 full mutations (>200 CGG repeats), should be differentiated.
## Appendix 1. Authors

<table>
<thead>
<tr>
<th>Name</th>
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<tbody>
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Figure Legends:

**Figure 1. Brain MRI.** (A-B) Axial brain MRI showed bilateral symmetric hyperintensities of the middle cerebellar peduncles (arrows) on T2-weighted fluid-attenuated inversion recovery images and (C-D) bilateral symmetric hypointensities of the middle cerebellar peduncles (arrows) on T1-weighted images, which are typical imaging findings of fragile X-associated tremor/ataxia syndrome.

**Video 1.** Neurologic examination demonstrated a head tremor, a lower limb resting tremor, an irregular postural and intention tremor of the distal upper limbs, ataxic dysarthria, and difficulty with tandem gait.
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