

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

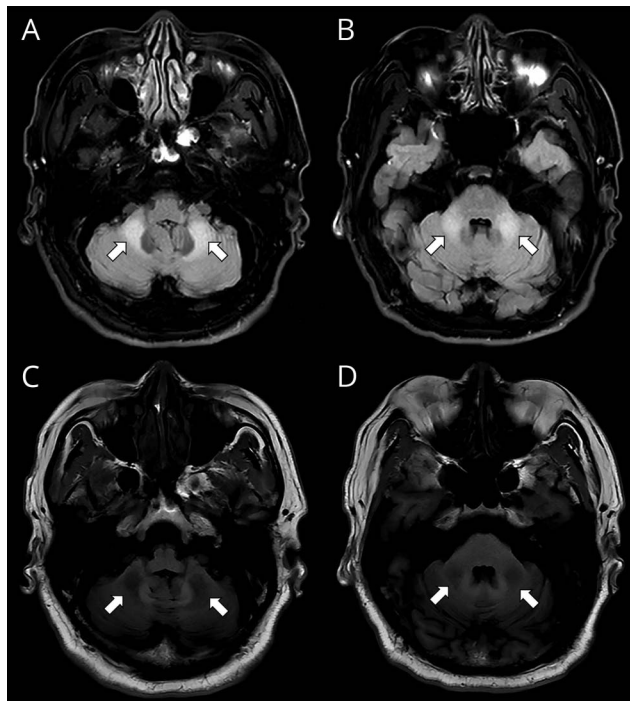
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Figure Brain MRI



(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.

A 63-year-old man presented with a 3-year history of tremors. Neurologic 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). T2-weighted brain MRI showed typical bilateral symmetric hyperintensities in the middle cerebellar peduncles (Figure). 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.¹ The characteristic MRI pattern should prompt confirmation of the diagnosis with genetic testing for *FMR1* premutations (55–200 CGG repeats).¹ Fragile X syndrome, associated with *FMR1* full mutations (> 200 CGG repeats), should be differentiated.¹

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Video

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Disclosure

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Appendix Authors

Name	Location	Contribution
Kundian Guo, MD	West China Hospital, Sichuan University, Sichuan, China	Major role in the acquisition of clinical information and drafted the manuscript
Yue Liu, MD	West China Hospital, Sichuan University, Sichuan, China	Collected clinical information

Appendix (continued)

Name	Location	Contribution
Zhen Hong, MD, PhD	West China Hospital, Sichuan University, Sichuan, China	Design and conceptualized study and revised the manuscript for intellectual content

Reference

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