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

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Kundian Guo: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Analysis or interpretation of data

Yue Liu: Major role in the acquisition of data

Zhen Hong: Drafting/revision of the manuscript for content, including medical writing for content; Study concept or design

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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 (*FMRI*) 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 *FMRI* premutations (55-200 CGG repeats).¹ Fragile X syndrome, associated with *FMRI* full mutations (>200 CGG repeats), should be differentiated.¹

Appendix 1. Authors

Name	Location	Contribution
Kundian Guo, MD,	West China Hospital, Sichuan University, Sichuan, China,	Major role in the acquisition of clinical information; drafted the manuscript
Yue Liu, MD,	West China Hospital, Sichuan University, Sichuan, China,	Collected clinical information
Zhen Hong, MD, PhD,	West China Hospital, Sichuan University, Sichuan, China,	Design and conceptualized study; revised the manuscript for intellectual content

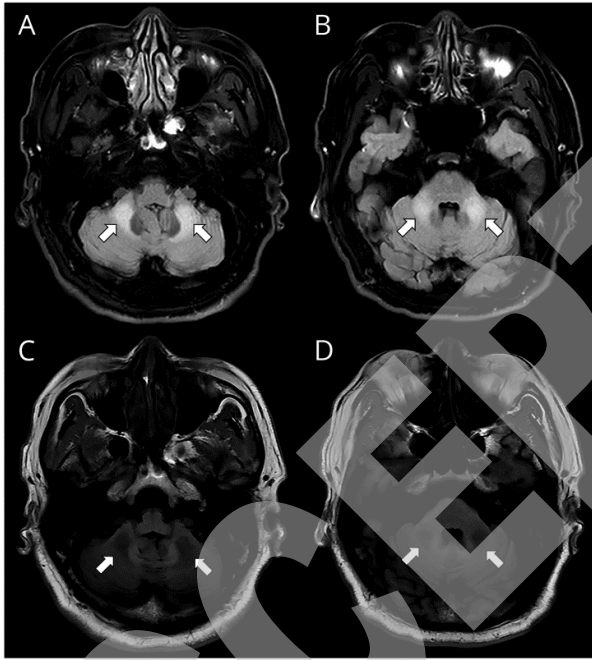
[AZ 12.7.2021] 177271 Teaching Slides-<http://links.lww.com/WNL/B720>

[AZ 12.7.2021] 177271 Video 1-<http://links.lww.com/WNL/B721>

1. Jacquemont S, Hagerman RJ, Leehey MA, et al. Penetrance of the fragile X-associated tremor/ataxia syndrome in a premutation carrier population. JAMA 2004;291:460-469.

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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