Cerebellar T2 hyperintensities in a patient with tremor

Figure MR images

(A) T2-weighted MR image showing moderate cerebellar volume loss. (B) T2-weighted MR image showing marked hyperintensity in the middle cerebellar peduncles extending into the posterior white matter of the cerebellum. (C) Axial FLAIR MR image through the level of the lateral ventricles showing cerebral periventricular hyperintensities.

A 58-year-old man noticed mild hand tremor for 2 years. As the tremor increased, he was referred to brain CT and consequently to MRI showing cerebellar and cerebral hyperintense areas on T2-weighted images (figure). Neurologic examination was normal except for mild postural and intentional tremor, more pronounced on the left hand. Because of the typical,^{1,2} though not specific,³ radiologic features, diagnosis of fragile X premutation tremor/ataxia syndrome (FXTAS) was subsequently confirmed by finding a CGG repeat number of 100 on genetic study. This case illustrates the wide spectrum of clinical features of FXTAS and the necessity for high suspicion even in monosymptomatic adults.

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