

Background:

Fragile X-associated tremor/ataxia syndrome (FXTAS) is a late-onset neurodegenerative disorder linked to premutation alleles (55–200 CGG repeats) in the FMR1 gene. It predominantly affects males and is characterized by cerebellar gait ataxia, intention tremor, and frontal-executive dysfunction.

Objectives:

To describe a challenging case of late-onset FXTAS and its symptomatic management.

Discussion and Conclusions:

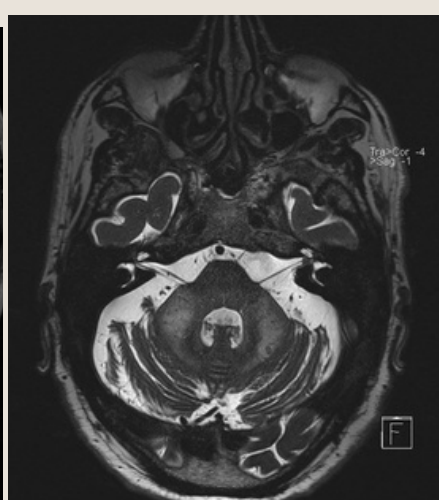
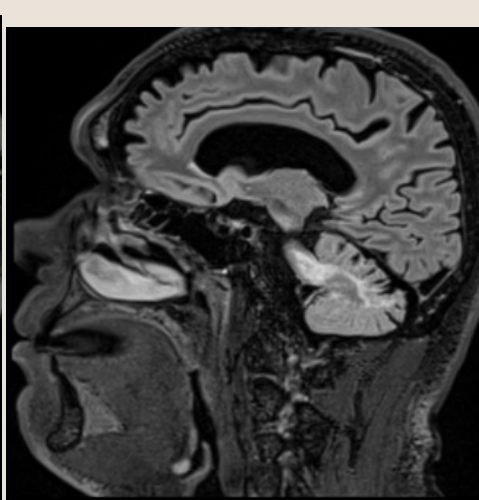
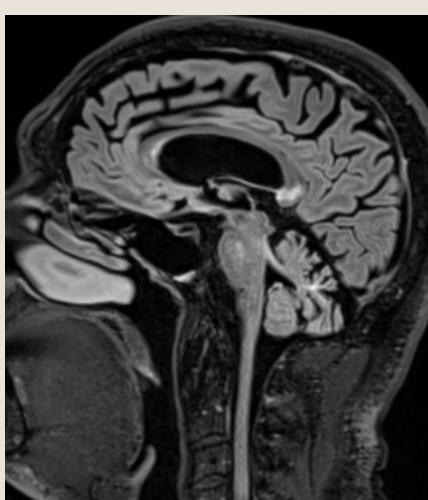
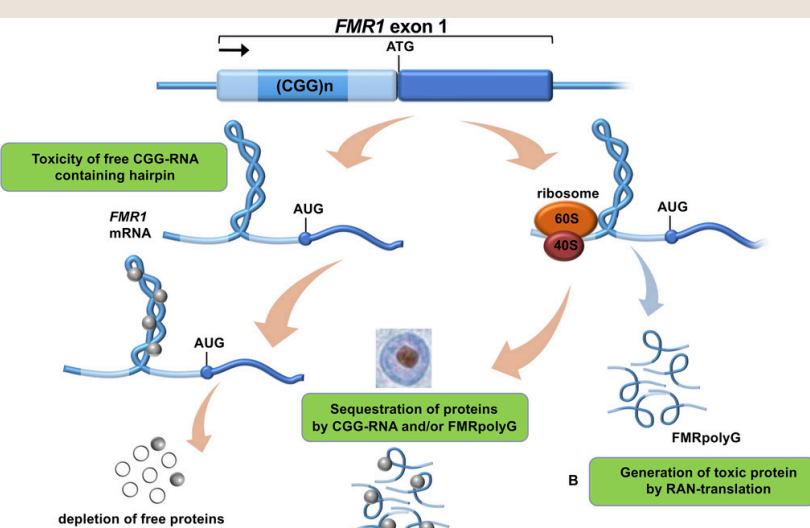
FXTAS should be considered in the differential diagnosis of late-onset tremor and ataxia, particularly in males with a suggestive family history. The clinical presentation may overlap with other inherited ataxias, such as spinocerebellar ataxias, making diagnosis challenging. Neuroimaging can be decisive, as T2/FLAIR hyperintensity of the middle cerebellar peduncles is a hallmark of FXTAS. Propranolol may offer symptomatic relief for tremor in selected cases.

Materials and Methods:

We present the case of a 65 year old man with a ten year history of progressively disabling action tremor and balance impairment. His family history included unspecified adult onset neuro degenerative diseases. An extensive diagnostic workup was performed, including neurological evaluations, brain MRI, and genetic testing.

Results:

Neurological examination revealed wide-based gait, cerebellar dysarthria, limb dysmetria, coarse irregular postural and kinetic tremor of the upper limbs, jaw tremor, and mild cognitive slowing with apathy. Serial brain MRI showed progressive cerebellar atrophy and T2/FLAIR hyperintensities in the middle cerebellar peduncles, pontine white matter, and corpus callosum. Genetic analysis confirmed a premutation allele with 97 CGG repeats in the FMR1 gene, supporting the diagnosis of FXTAS. Propranolol led to improvement of tremor.



References

Leehey MA. Fragile X-associated tremor/ataxia syndrome: clinical phenotype, diagnosis, and treatment. J Investig Med. 2009 57(8) 830-6



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