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Thursday

Genetic evaluation of *FMR1* in a person presenting with symptoms of frontotemporal degeneration.

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State of the art: Currently, clinical genetic evaluation for frontotemporal degeneration (FTD) typically includes focused testing for the genes associated with autosomal dominant FTD (*C9orf72*, *MAPT*, *GRN,* and less common genes such as *TBK1*). Given symptom overlap with other dementias and psychiatric conditions, genetic testing is often pursued for diagnostic clarification. However, other conditions can mimic FTD, and the genes that cause those conditions are not typically included in FTD genetic evaluation.

Methodology: A 66-year-old man presented to care for cognitive and behavioral changes, anxiety, and depression, with concern for FTD. He used a walking stick, but there was no ataxia noted. He was referred to genetic counseling. Both family history (balance issues, psychiatric symptoms, infertility, autism) and brain MRI findings (signal abnormality in the middle cerebellar peduncles) put Fragile X Tremor Ataxia Syndrome (FXTAS) on the differential. *FMR1* genetic testing was added to the typical FTD genetic workup.

Results: A premutation of 95 CGG repeats was identified in *FMR1*, confirming a diagnosis of FXTAS. Accurate genetic diagnosis resulted in expedited referral to an ataxia specialist for management and allowed family members to pursue genetic counseling and testing.

Conclusions: FXTAS is a potentially important and underdiagnosed mimic of FTD. *FMR1* analysis should be considered in addition to the typical FTD genetic evaluation, especially if ataxia or MRI findings of white matter hyperintensities in the brainstem or cerebellum are noted. We are in the process of assessing frequency of *FMR1* premutations in our cohort of patients with FTD and FTD phenocopies.

Conflicts of interest

No relevant disclosures of interest for this research.