

A tremor in the family

Tommaso Carrer, G. Bonato, M. Carecchio

Parkinson and Movement Disorders Unit, Department of Neuroscience, University of Padua, Padua Italy

Introduction: We describe the case of a 66 year-old woman with onset at age 54 of head and upper limb tremor, worsened by stressful events, not invalidating but slightly progressive over time. Through the years she also developed a subjective sensation of dizziness and disequilibrium. The neurological examination showed a normal based gait with reduced arm swing and some difficulties in tandem gait; positive pull test and Romberg test with marked retropulsion; head tremor with cervical dystonia and mild vocal tremor; at the upper limbs mild rest and postural tremor, worsened by specific positions, not responsive to propranolol and levodopa. DAT-SPECT demonstrated no significant uptake reduction of the radiotracer, whereas a brain MRI showed a mild cerebellar atrophy. The patient has two children affected by X-fragile syndrome and she reported to have a sibling and an aunt with tremor and extrapyramidal symptoms. She is carrier of a premutation of the gene FMR1 (104 CGG repeats) with unbalanced inactivation and functional exclusion by methylation of the normal FMR1 allele. She was diagnosed with FXTAS (Fragile X–Associated Tremor/Ataxia Syndrome). Her medical history was relevant for depression and premature ovarian insufficiency, both linked to FMR1 premutation.