

Botulinum neurotoxin in FXTAS-related tremor: a “successful” case report

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Introduction: Fragile-X-associated tremor/ataxia syndrome (FXTAS) is a X-linked neurodegenerative disease due to the premutation in the FMR1 gene [1], manifesting as late-onset ataxia associated with postural-kinetic tremor and cognitive decline. In this condition, tremor commonly represents a disabling and unsatisfactorily-treated symptom.

Objective: We report the case of a FXTAS-affected man with disabling tremor, who experienced a significant benefit from botulinum neurotoxin (BoNT) injection treatment.

Methods: A 59-year-old man came to our attention for a 10-years-lasting hand tremor. The tremor, mainly during voluntary movements, began in the right hand and progressively extended to the left hand and the lower limbs. The neurological examination revealed a moderate, right-prevailing, kinetic tremor of the hands, accompanied by rest and postural components, with subtle right-hand clumsiness. The brain MRI showed a cerebello-pontine atrophy with T2-FLAIR hyperintensity of both the middle cerebellar peduncles and the splenium of corpus callosum. Genetic testing was performed and revealed an expansion of the CGG-trinucleotide FMR1 gene sequence with 106 triplets.

The patient did not tolerate oral propranolol therapy due to bradycardia. Hence, an attempt with BoNT was made with a single session of incobotulinum toxin type A injection (diluted in 2cc of 0.9% sodium chloride solution) in the right flexor digitorum superficialis muscle (30 units) and right extensor digitorum muscle (10 units).

A pre-injection and post-injection evaluations with wrist-worn accelerometry were performed by using a wrist triaxial accelerometer and recording tremor in rest, postural and kinetic conditions.

Results: About two weeks after the injection, the patient reported a subjectively significant reduction of the tremor. The post-injection accelerometric assessment demonstrated a great decrease in the amplitude of the tremor. The beneficial effect lasted about 5 months.

Conclusions: BoNT injection typically constitutes a therapeutic second-line-option in tremor syndromes refractory to pharmacological agents [2-3]. Our case suggests the possibility of its broader application in the symptomatic treatment of FXTAS-related tremor.

References:

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