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Myoclonic dystonia with DYT1 gene mutation

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Introduction: DYT1 gene mutations lead to early-onset dystonia that typically begins with twisting of an arm or leg and subsequent generalization. Anyway, it has emerged that the phenotype associated with the DYT1 mutation can be variable.

Objective: In the present work, we describe the case of a 54 years old man who presented an early onset (7-8 years old) of unilateral tremor of the right hand, spreading to the contralateral one during the following 4-5 years, and progressively worsening over time. Abnormal and painful muscular contractions of both hands were associated to tremor.

Methods: Clinical and neurophysiological evaluations were performed. Previously conducted neuroimaging studies (brain MRI, CT scan) were negative for pathological findings.

Results: The patient had an unremarkable history for perinatal or central nervous system insults. Family history revealed similar symptoms in the patient's father and father's uncle with juvenile onset. At the age of 43 he first came for evaluation at our Institute, where therapy with anticholinergic drugs and botulinum toxin injections was introduced in the suspicion of dystonic tremor, with mild clinical benefit. At a clinical re-evaluation, brief and sudden jerks were observed in the right upper limb; at electromyographic recording of arms muscles, short jerks (duration ranging from 50 to 200 ms) occurring at rest and during posture with pseudo-rhythmic pattern were found in the distal right arm, with co-contraction or alternating contraction of antagonist muscles. Clinical and familiar history, along with findings from neurophysiological examination were suggestive for myoclonic dystonia, and a DYT11 gene mutation could be suspected. Genetic counseling and molecular analysis with NGS panel testing including genes associated with isolated/combined dystonia were performed.

Conclusions: Recurrent deletion c.907_909del (p.Glu303del) of the TOR1A gene was found. Thus, we concluded for an atypical presentation of DYT1 dystonia with segmental dystonia and myoclonus. Clonazepam was initiated with great clinical benefit.