

Perampanel as a novel treatment for myoclonus in myoclonus-dystonia syndrome

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Background: Myoclonus-dystonia (MD) is a heterogeneous genetic group of disorders characterized by subcortical myoclonus and mild to moderate dystonia [1,2]. The main causative gene is the epsilon sarcoglycan gene (SGCE) but genetic background can be heterogeneous [3]. Response to medications is variable, with poor tolerability limiting their use [4].

Case presentation: We present the case of a patient with involuntary movements since childhood. Symptoms progressed gradually over the years, involving neck, shoulders and upper limbs and interfering with daily activities. She referred to a neurologist at the age of 46y, and myoclonus-dystonia was diagnosed. Genetic testing identified a novel mutation in SGCE gene (c.907delC) in heterozygosis. Her parents were dead, thus no genetic analysis was possible. However, none of her relatives reported neurological disturbances. Clinically, she presented brief myoclonic jerks predominating in the upper limbs and neck, mild at rest and elicited by action, posture and tactile stimulus. Myoclonus was accompanied by mild neck and right arm dystonia. Over time she assumed a large variety of antiepileptics without beneficial effect on myoclonus and low tolerability. Treatment with Perampanel at 4 mg/day was started, with a beneficial effect on myoclonus. No adverse events were reported.

Discussion: Perampanel is the first selective non-competitive AMPA receptor antagonist approved in add-on for focal and generalized tonic-clonic seizures. It has been used to treat myoclonus in a few patients with Progressive myoclonic epilepsies and Lance Adams syndrome with beneficial effects [5,6] but no reports are available in literature in MD. To our knowledge this is the first trial of perampanel in MD.

Conclusions: We presented the case of a patient with MD due to SGCE mutation who was treated with Perampanel with beneficial effects. We propose Perampanel as a novel treatment for myoclonus in MD.

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