P52

Neurophysiological assessment of juvenile parkinsonism due to primary monoamine neurotransmitter disorders

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Background: No studies have investigated voluntary movement abnormalities and their neurophysiological correlates in patients with parkinsonism due to inherited primary monoamine neurotransmitter (NT) disorders.

Methods: Nine NT disorders patients and 16 healthy controls (HCs) were enrolled. Objective measurements of repetitive finger tapping were obtained using a motion analysis system. Primary motor cortex (M1) excitability was assessed by recording the input/output (I/O) curve of motor-evoked potentials (MEPs) and using a conditioning test paradigm for short-interval intracortical inhibition (SICI) assessment. M1 plasticity-like mechanisms were indexed according to MEPs amplitude changes after the paired associative stimulation protocol. Patient values were considered abnormal if they were greater than two standard deviations from the average HCs value.

Results: Patients with aromatic amino acid decarboxylase, tyrosine hydroxylase, and 6-pyruvoyl-tetrahydropterin synthase defects showed markedly reduced velocity (5/5 patients), reduced movement amplitude, and irregular rhythm (4/5 patients). Conversely, only 1 out of 3 patients with autosomal-dominant GTPCH deficiency showed abnormal movement parameters. Interestingly, none of the patients had a progressive reduction in movement amplitude or velocity during the tapping sequence (no sequence effect). Reduced SICI was the most prominent neurophysiological abnormality in patients (5/9 patients). Finally, the I/O curve slope correlated with movement velocity and rhythm in patients.

Conclusions: We provided an objective assessment of finger tapping abnormalities in monoamine NT disorders. We also demonstrated M1 excitability changes possibly related to alterations in motor execution. Our results may contribute to a better understanding of the pathophysiology of juvenile parkinsonism due to dopamine deficiency.