Abstract

Objective: To assess nonparoxysmal movement disorders in *ATP1A3* mutation-positive patients with alternating hemiplegia of childhood (AHC).

Methods: Twenty-eight patients underwent neurologic examination with particular focus on movement phenomenology by a specialist in movement disorders. Video recordings were reviewed by another movement disorders specialist and data were correlated with patients' characteristics.

Results: Ten patients were diagnosed with chorea, 16 with dystonia (nonparoxysmal), 4 with myoclonus, and 2 with ataxia. Nine patients had more than one movement disorder and 8 patients had none. The degree of movement disorder was moderate to severe in 12/28 patients. At inclusion, dystonic patients (n = 16) were older (p = 0.007) than nondystonic patients. Moreover, patients (n = 18) with dystonia or chorea, or both, had earlier disease onset (p = 0.042) and more severe neurologic impairment (p = 0.012), but this did not correlate with genotype. All patients presented with hypotonia, which was characterized as moderate or severe in 16/28. Patients with dystonia or chorea (n = 18) had more pronounced hypotonia (p = 0.011). Bradykinesia (n = 16) was associated with an early age at assessment (p < 0.01). Significant dysarthria was diagnosed in 11/25 cases. A history of acute neurologic deterioration and further regression of motor function, typically after a stressful event, was reported in 7 patients.

Conclusions: Despite the relatively limited number of patients and the cross-sectional nature of the study, this detailed categorization of movement disorders in patients with AHC offers valuable insight into their precise characterization. Further longitudinal studies on this topic are needed.