Writer's cramp and gait dystonia in SGCE-myoclonus dystonia syndrome: describing patterns and exploring rating tools

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Introduction and objectives: Although less prominent than myoclonus, patients with Myoclonus-dystonia syndrome (MDS) are often significantly impaired by dystonia. Previous studies by our group found that the Burke-Fahn-Marsden Dystonia Rating Scale did not satisfy criteria for validity in MDS patients. For this reason, we aimed to explore other scales to rate limb dystonia in a large cohort of SGCE mutated patients.

Methods: Video-analysis of 53 patients with SGCE-MDS was performed by three experts in movement disorders. Modified versions of the Writer's Cramp Rating Scale (WCRS) including shoulder and trunk movements were used to rate writing dystonia. Gait dystonia was rated with a new proposed scale analogous to WCRS, named Walking and Running Dystonia Rating Scales (WDRS and RDRS).

Results: 49/53 patients were included (if sufficient quality video-recording), mean age was 15,2 years (2,5-51). Overall, 45 (92%) exhibited dystonia. A minority had dystonia at rest (1/49, 2%) and during upper limbs posture (12/47, 26%). On the contrary, writer's cramp was observed in 35/42 (83%) patients, with some particular proximal/distal patterns described. In 2 patients with writer's cramp, WCRS was 0, unlike the modified versions. Action-induced lower limb dystonia was observed on walking (12/46, 26%) and running (13/30, 43%). The most frequent dystonic position/movement was ankle plantar flexion on walking and knee extension on running. Mean RDRS scores were significantly higher than WDRS scores. Modified WCRS, WDRS and RDRS scores correlated significantly with writing/walking/running speed, patient-perceived handwriting/walking difficulties and functional tests.

Conclusions: Action-induced dystonia was present in almost all patients with SGCE-MDS. The proposed rating tools (which are limb and action-specific scales) allowed to assess the severity of writer's cramp and gait/running

dystonia in our series and correlated significantly with functional impairment and disability.