AUTONOMIC DYSFUNCTION IN HEREDITARY SPASTIC PARAPLEGIA *SPG4*

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Introduction: *SPG4* gene mutations are the most common cause of hereditary spastic paraplegia (HSP-SPG4) and characterized by progressive weakness, spasticity and hyperreflexia in lower limbs. There are few studies about non-motor manifestations in this disease and none about the autonomic system involvement. The aim of this study was to determine the frequency and pattern of autonomic complaints in patients with HSP-SPG4, as well as the clinical relevance and the possible factors associated with these manifestations.

Materials and Methods: Clinical and electrophysiological evaluations were performed in 32 patients with HSP-SPG4 confirmed by molecular tests and 38 healthy controls. The Scales for Outcomes in Parkinson's Disease: Autonomic Questionnaire (SCOPA-AUT) was applied to quantify the severity of autonomic symptoms. Electrophysiological tests included Sympathetic skin response (SSR) recorded in hands and feet; and Quantitative Sudomotor Axonal Response Test (QSART) in the typical recording sites: the medial forearm, the proximal leg, the distal leg and the proximal foot. The groups were compared with Mann Whitney and Chi-Square test, p values < 0.05 were considered significant.

Results: There were 20 men, with a mean age of 45.4 ± 15.3 years. The median SCOPA-AUT score was 13 ($p = 0.003$). Urinary and cardiovascular domains subscore were significant (6 vs 1 $p = 0.002$; 0 vs 2 $p = 0.028$ respectively). Absent SSR in the feet were more frequent among patients (59.4% vs 2.6%, $p < 0.01$). QSART responses were also smaller in the HSP-SPG4 group at the forearm (0.44 µL vs 1.03 $p = 0.008$), at distal leg (0.55 vs 1.15 $p = 0.028$) and the feet (0.26 vs 0.7 $p = 0.004$).

Discussion: These results indicates that patients have sudomotor dysautonomia. The abnormalities found in QSART test indicated damage to small postganglionic cholinergic fibers.

Conclusion: Patients with HSP-SPG4 frequently present symptoms of dysautonomia. Electrophysiological tests showed autonomic sudomotor dysfunction.

References:

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