EPIDEMIOLOGICAL PROFILE OF THE PATIENTS WITH VAPB MUTATION AND MOTOR NEURON DISEASE AS WELL AS THEIR IMMEDIATE RELATIVES - A STUDY OF FUNCTIONAL IMPAIRMENT AND AUTONOMIC DYSFUNCTION

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Introduction: Motor neuron diseases comprise conditions that are associated with progressive degeneration of both or either one of the motor neurons. First described in the late 19th century by Charcot, amyotrophic lateral sclerosis (ALS) is sporadic in almost 90% of the cases and is traditionally understood as a disease restricted to the motor neurons. This concept, however, is changing and nowadays it is more frequently understood as a multisystemic disorder. In a previous study, we observed that VAPB-associated disease may have an underestimated important autonomic component.

Objectives: Our main goal in this study is to disclose the prevalence of dysautonomia in a population of VAPB patients.

Methods: We established remote contact with patients and applied both ALS-FRS-R and COMPASS 31 scales. Whilst the former evaluated function impairment related to the disease, the latter searched for autonomic dysfunction by assessing orthostatic intolerance, vasomotor, secretomotor, gastrointestinal, bladder, and pupillomotor domains.

Results: The mean age of our patients was 50 years. 55% were female and 50% had other chronic diseases, among which hypertension, diabetes, and hypothyroidism. The mean ALSFS-R score was 34 and COMPASS 31 score was 22,05. 55% of the patients reported sexual dysfunction. Regarding dysautonomia, gastrointestinal manifestations were the most common (90%), followed by affected secretomotor (70%) and bladder (65%) domains.

Conclusion: VAPB mutation disorders seem to be associated with important autonomic manifestations. Detailing these manifestations will bring more understanding of the disease and better patient care.

Keywords: Motor neuron disease. VAPB gene. Dysautonomia. Spinal muscular atrophy. Amyotrophic lateral sclerosis.