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Pure autonomic failure: which markers of phenoconversion to central alpha-synucleinopathies? A clinical case

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Introduction: Pure autonomic failure (PAF) is a neurodegenerative disorder of the autonomic nervous system characterized by peripheral deposition of a-synuclein in autonomic ganglia and nerves. However, patients with PAF may progress into other synucleinopathies with CNS involvement.

Clinical case: A 48 years-old women developed hortostatic hypotension (OH) with frequent syncope. She was seen at our clinic, at the age of 58. Autonomic testing showed neurogenic OH and anhidrosis. Supine plasma norephinephrine levels were <25pg/ml and HR was 68bpm. Constipation, urinary urgency, loss of smell and a history of RBD were referred. Neurological examination and [123I]-FP-CIT
were normal, whereas 123-I MIBG demonstrated a cardiac denervation. A diagnosis of PAF was made. The patient underwent to follow-up visits every 4 months. Five years later, she developed mild generalized bradykinesia, hypomimia and difficulty performing fine movements mainly on left hand. A [123I]-FP-CIT was repeated demonstrating presynaptic dopaminergic deficit in both putamen and in both caudate nuclei (right>left). Levodopa and rasagiline were started with good response on motor symptoms (UPDRS 11vs23).

D*iscussion:* The age of the onset of autonomic failure in our patient was 48 years and the time of phenoconversion to diffuse synucleinopathy was 15 years. It has been suggested that the early onset of autonomic failure (before the age of 50) makes phenoconversion to MSA more likely than PD/DLB (mid-60s) and that the time of phenoconversion to MSA is about 5y compared to PD/DLB, which is expected to be 9.5y. The combination of olfactory dysfunction, RBD and cardiac denervation is considered a predictor of phenoconversion to PD. Furthermore, plasma norepinephrine levels<110 pg/ml and HR<70 bpm are more associated with patients maintaining the PAF phenotype [1]. Our patient presented with a combination of these signs and symptoms. The coexistence of different prognostic markers should suggest careful clinical follow-up of patients with PAF.

References:

[1] Horacio Kaufmann at al. The Natural History of Pure Autonomic Failure: a U.S. Prospective Cohort 2017 February; 81(2): 287–297. doi:10.1002/ana.24877.