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RBD in pure autonomic failure

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Introduction

Pure autonomic failure is considered to be a degenerative condition of the autonomic nervous resulting from accumulation of alpha-synuclein in autonomic ganglia [1,2]. Lack of CNS involvement distinguishes this disorder from other synucleopathies with CNS involvement like MSA, DLB and PD. However, long term follow-up of PAF patients has shown that some of these cases convert to one of these disorders most commonly MSA [3]. Also, recent data, albeit limited in nature has shown the association of REM sleep disorder in PAF patients challenging the previous notion that PAF is a disease restricted to autonomic nervous system. This data has led to the discovery of a clinical syndrome with a position in the middle of the spectrum of these disorders with PAF at one end of the spectrum and MSA, DLB and PD at the other end. We hereby present a case report of one such patient with PAF associated with RBD but without any other CNS manifestations.

Case report

A 50-year-old male presented to this hospital in 2010 with complaints of recurrent episodes of orthostatic dizziness, occasionally associated with transient loss of consciousness for less than a minute. These complaints had started 3 years prior to his first admission. Over time, patient noticed to have decreased sweat production on heat exposure. This was followed by loss of early morning erections. All these symptoms occurred over a period of 2 years. Urinary symptoms began to appear in the third year of illness. He started having increased frequency of micturition, hesitancy and intermittent interruption of urinary stream, urgency at times and sense of incomplete bladder evacuation. Early satiety, stomach fullness and constipation were the other symptoms described by the patient. Apart from the symptoms related to autonomic nervous system, patient had history of vivid dreams and enacting out of dreams. These

symptoms however, had started 20 years prior to the onset of his new symptoms. Patient did not have any history of unsteadiness of gait, difficulty in reaching out at objects or any difficulty in performing tasks with his hands. He didn't have slowness or change in posture or gait difficulty. He was not on any medications. He was a non-smoker and did not indulge in drug abuse. He was a salesman by profession. In his first assessment, his cognitive functions were normal with MMSE of 28. Cranial nerve examination, motor, sensory and cerebellar examination were normal. No bradykinesia or rigidity was noted. A postural drop of 40 mmHg in systolic and 20 mm Hg in diastolic BP after 3 minutes of standing was noted. Head-up tilt table test was detected to be abnormal. Valsalva test was abnormal. MRI brain was normal. Serum norepinephrine level in supine position was detected to be 90 pg/ml. Patient was diagnosed as pure autonomic failure and advised non pharmacological measures and oral fludrocortisone.

He had multiple admissions during last 10 years for delirium and was diagnosed with UTI on a couple of occasions and hyponatremia on one occasion. After the treatment of precipitating cause, he would return to his baseline condition. In 2017, he had a fall during syncopal episode that resulted in fracture neck of femur on right side. Considering his diagnosis of Pure autonomic failure, he was not subjected to surgery and since then, patient is restricted to his bed, though he is able to take care of some of his activities including feeding himself, shaving and urination. In Feb 2021, he again got admitted with delirium. E.Coli was grown from his urine culture. He was treated with antibiotics and improved. Detailed cognitive assessment was done, which was normal. No cerebellar signs or parkinsonian features were noted on examination. A postural drop of 100 mmHg from supine to sitting position was noted. Valsalva test showed no variation of heart rate in phase 2 and phase 4. A total lack of sweating was noted on Thermoregulatory sweat test. MRI brain was unremarkable except for insignificant white matter hyperintensities. Nerve conduction study was normal. Supine norepinephrine was 82 pg/ml and it increased marginally to 86 pg/ml in sitting position. Video-polysomnography revealed lack of REM sleep atonia as noted by phasic limb and submental EMG twitching. Episodes were accompanied by limb and trunk movements, myoclonic activity and vocalization. Patient was diagnosed as Pure autonomic failure with REM sleep disorder. This case supports the concept of association of REM sleep disorder with pure autonomic failure.

Discussion

Pure autonomic failure has long been considered to be a degenerative condition of autonomic nervous system without any CNS involvement. The lack of CNS involvement was used to distinguish PAF from MSA, although at pathological level, both are considered to be synucleopathies. However, recently there have been case reports depicting the association of REM sleep disorder with pure autonomic failure. Our case supports the association of RBD with PAF.

Plazzi et al in his case series of 10 patients had concluded that the presence of REM behavioural disorder served as a distinguishing feature between PAF and MSA as he found that 4 patients who progressed to MSA had history of RBD. RBD was not seen in those patients who remained with a diagnosis of PAF [4]. However, Miglis et al reported the presence of RBD in 5 out of 8 patients with a diagnosis of PAF [5]. Weyer et al also reported 3 cases of PAF with RBD [6]. One case series reported the presence of frequent nocturnal vocalization in 9 out of 13 patients diagnosed with PAF [7]. The duration of autonomic symptoms in these studies has been 3 to 11 years and RBD symptoms 3 to 8 years. It has also been reported in these studies that RBD symptoms appeared after the onset of autonomic symptoms. The RBD symptoms in our patient, however, appeared 20 years prior to the onset of autonomic symptoms while as autonomic symptoms have been present for nearly 13 years. No extrapyramidal and cerebellar symptoms have appeared in the patient till now, thereby making the chances of progression to MSA less likely. This is in line with the results shown by Mabuchi et al who concluded that patients with PAF have a very slow disease progression as compared to MSA although the patients with PAF in his study were not reported to have RBD symptoms [8]. Singer et al reported that nearly 10% of initially diagnosed PAF cases ultimately evolve to develop MSA. Factors reported to predict conversion to MSA include subtle motor signs at presentation, supine norepinephrine > 100 pg/ml, preganglionic pattern of sweat loss, severe bladder dysfunction and mild cardiovagal involvement [3]. Our case again highlights the slow disease course of Pure autonomic failure even when associated with RBD, in sharp contrast to the disease course described in MSA. Serum norepinephrine level has been used to distinguish PAF from MSA. Serum norepinephrine levels are usually less than 200 pg/ml and can reach less than 100 pg/ml in advanced stages, while as MSA patients have usually normal or high levels in supine position, but fail to rise in standing position resulting in orthostatic hypotension [9]. Our patient also reflected a markedly low norepinephrine level that did not rise with the sitting position consistent with the diagnosis of PAF.

The explanation for involvement of one particular region in CNS in exclusion to others is still an unsolved puzzle. Although existence of RBD in association with PAF is being cited more often than not, only future research on a large scale is going to unravel whether or not PAF with RBD symptoms should be regarded as a distinct entity.

Statement of conformation to the declaration of Helsinki: All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants involved in the study.

Consent for publication: Consent for publication was taken from the patient and if needed will be produced.

Author contribution: All the listed authors were involved with the management of this case. Shabeer A Paul and C. Patra compiled the manuscript. Sarbajit Das reviewed the literature. K.C. Ghosh, R. Bhattacharyya and G.P. Mondal supervised the manuscript.

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