Rbd and other sleep disorders in a cohort of p.a53t snca mutation carriers

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REM sleep behavior disorder (RBD), defined as REM sleep without atonia (RWA) plus either dream enactment behavior, sleep related injuries, potentially injurious or disruptive behaviors, documented by medical history, or polysomnography (PSG), may occur in association with neurodegenerative diseases, mainly α-synucleinopathies. In idiopathic Parkinson's Disease (PD) RBD may precede the motor manifestations of the disease. There is a general question whether PD due to defined genetic causes, transmitted through Mendelian inheritance, is similar to idiopathic PD. In this regard, it is especially interesting to assess whether RBD and other sleep abnormalities occur in carriers of the p.A53T alpha-synuclein gene (SNCA) mutation, the prototypical genetic synucleinopathy. Such a systematic study has not been performed previously. We have accordingly assessed 10 p.A53T carriers with PSG, Epworth Sleepiness Scale, RBD Screening Questionnaire (RBDSQ), UPDRSIII and MOCA. Three of the p.A53T carriers were asymptomatic, had no evidence of RBD in PSG and scored ≤5 in RBDSQ. All 7 symptomatic p.A53T carriers had evidence of sleep disorder in PSG. Three were diagnosed with RBD in PSG, however 2 of them were treated with antidepressants and only 1 of them scored >5 in RBDSQ. In three others, PSG showed RWA, but only 1 of them scored >5 in RBDSQ. The last one was diagnosed with PLM in PSG, was not treated with drugs and scored <5 in RBDSQ. We conclude that RBD or RWA occur in the majority of PD p.A53T manifesting carriers, possibly at a higher percentage compared to idiopathic PD.