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A juvenile case of Parkinson's disease associated with MAPT mutation

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Introduction: Parkinson's disease (PD) is defined primarily as a movement disorder, in addition to the defining dopaminerelated motor symptoms, however, PD is increasingly recognized as a heterogeneous multisystem disorder. Autosomal dominant mutations in the microtubule-associated protein tau (MAPT) gene were found to cause forms of parkinsonism and frontotemporal dementia.

Objective: We describe heterogeneous clinical manifestations in a 51 years old man with PD and carrying a mutation in MAPT gene.

Methods: Subject underwent serial neurological visits, Genetic Analysis, brain magnetic resonance imaging (MRI), brain 123I-FP-CIT SPECT, brain PET-FDG and neuropsychological assessments.

Results: We present the case of a man who was diagnosed with PD at the age of 47 years, beginning with right upper limb bradykinesia. The brain 123I-FP-CIT SPECT confirmed low uptake in the left caudate and putamen while the Brain MRI showed mild ventricular dilation. Therapy with dopamine-agonist, rasagiline and levodopa was started. Interestingly the gene analysis identified a pathogenic variant in exon 10 of MAPT gene (c. 1853C>T; p.Pro618Leu). Further diagnostic investigations highlighted left insular and temporo-parietal hypometabolism through Brain PET-FDG and deficit of attentive, executive e visuospatial skills. In the following years motor symptoms worsened, impulse control disorder occurred (with hyperphagia, hypersexuality, gambling and drug abuse) and the patient presented traits of aggression, requiring psychiatric intervention.

Conclusions: Tauopathies refer to a wide range of phenotypically diverse diseases characterised by the aberrant aggregation of tau in neurons and/or glia, tau dysfunction is sufficient for widespread central nervous system neurodegeneration. In this case we increase the knowledge about the possible role of tau dysfunction in non-motor clinical manifestation of Parkinson disease.