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Mitochondrial disorders and Parkinson's disease: the clinical case of a patient with concomitant diagnosis of Parkinson disease and Leber's hereditary optic neuropathy

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Introduction: Mitochondrial diseases (MIDs) are a heterogeneous group of disorders caused by mitochondrial or nuclear DNA mutations, resulting in multiorgan disorders including the central and peripheral nervous system. Parkinsonism has been described in some cases of MIDs and some studies show a greater sensitivity of substantia nigra to mitochondrial dysfunction that represents a key event in the pathogenesis of Parkinson's disease (PD).

Objective: We present the case of a 70-year-old man with concomitant diagnosis of PD and Leber's hereditary optic neuropathy (LHON).

Method: The patient underwent neurological examinations, radiological tests (brain RMN, 123Ioflupane SPECT), electrophysiological and genetic analysis.

Outcome: Patient arrived at our attention at the age of 67 years, he suffered for PD diagnosed 7 years before and began with bilateral tremor in the upper limbs, greater on the left side. Brain NMR was normal and 123Ioflupane SPECT showed a severe reduction in the putamen uptake, more pronounced on the right side. Moreover, patient was diagnosed for LHON (mutation G11778A) and the same diagnosis was made for a brother and two maternal uncles. Family history was negative for PD, while the father suffered from Alzheimer's disease.

Conclusions: It is known that many genes involved in hereditary forms of PD encode for proteins with mitochondrial functions (Parkin, PINK1 and DJ1). Similarly extrapyramidal syndromes can occur in MIDs, in particular in patients carrying A8344G mutation or mutations in POLG1 and PEO1 genes. LHON is reported to be associated to some extrapyramidal symptoms, especially dystonia, chorea and tics. In this case we found an association of LHNO and PD, of which only very few cases have been reported, enriching the knowledge on the correlation between MIDs and movement disorders. Further studies are needed to understand the pathogenic role of mitochondrial dysfunction on PD.