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Juvenile Parkinson disease and mutation in RFC1 replication factor

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Introduction: The biallelic repeat expansion (AAGGG)exp in the replication factor C subunit 1 gene (RFC1) is a frequent cause of cerebellar ataxia, neuropathy and vestibular areflexia syndrome (CANVAS) as well as late-onset ataxia [1]. The clinical spectrum from the outset has been shown to be diverse and non-classical phenotypes are often observed.

Objective: We present the case of a 61-year-old man with concomitant diagnosis of Parkinson Disease (PD) and CANVAS.

Methods: The patient underwent neurological examinations, radiological tests (brain MRI, Ioflupane-SPET), electrophysiological and genetic analysis (AAGGG intronic expansion of the RFC1 gene, on two alleles).

Outcome: The patient presented in 2020 with difficulty in fine movements of the left hand. But since 2013 he had decreased sensitivity/paresthesias in the lower limbs starting from the foot and then radiating to the knee, followed by slight paresthesias in the hands, greater on the left. EMG (2020): polyneuropathy, predominantly sensory and axonal type, motor almost exclusively at lower limbs, mixed and distal type. Brain MRI reperto incidentale of small aneurysmal formation partially thrombosed. Ioflupane-SPET (2021) showed reduction of DAT sites in the putamen, prevalent on the right. He had recurring cough and rhinorrhea. Not familiarity for PD, but family history of disability (2 maternal cousins wheelchair bound at age 6-7, unknown diagnosis).

Conclusions: Our results suggest that (AAGGG)exp in RFC1 is a rare cause of early-onset PD. The present case shows an association with PD, recently reported rare cases in the literature. Certainly, a greater number of cases of juvenile PD should be studied to find an effective correlation between the

References:

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