ANALYSIS OF LRRK2 MUTATIONS IN COLOMBIAN PARKINSON’S DISEASE PATIENTS

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Introduction: Mutations in LRRK2 are a common cause of autosomal dominant and sporadic Parkinson’s disease (PD). In several Latin American populations the frequency of the LRRK2 p.G2019S mutation fluctuates among 2-3% in sporadic cases, whereas the LRRK2 p.R1441C/G/H mutations have been reported only in Mexico.

Aims: To determine the frequency of the LRRK2 pG2019S and R1441C/G/H mutations in Colombian PD patients.

Methods: We analysed the frequency of the LRRK2 p.G2019S and p.R1441C/G/H mutations in 154 colombian PD patients (24 familial and 130 sporadic cases; 24 early and 130 late-onset cases) and 162 normal controls. The mutations were genotyped by PCR and restriction endonuclease digestion with the SfcI enzyme for the p.G2019S mutation and with the BstUI enzyme for the p.R1441C/G/H mutations. Fragments were resolved using agarose gel electrophoresis and visualized with ethidium bromide.

Results: We found the LRRK2 p.G2019S mutation in two late onset PD cases (1.3%; a male and a female) with classical PD signs, good response to L-Dopa, no cognitive decline, no behavioral alterations. One of them had family history of PD. Additionally, we found one asymptomatic control positive for the mutation (0.6%) without family history of PD (OR=2.118, p= 0.965). LRRK2 mutations p.R1441C/G/H were not found.

Conclusions: The LRRK2 p.G2019S and p.R1441C/G/H mutations are not an important causal factor in Colombian PD patients, having similar frequencies to those reported in other Latin American populations. However, a larger sample must be analysed to clarify these observations. Funded by DIB-Unal.