MONOGENIC PARKINSON'S DISEASE IN A CHILEAN COHORT

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Introduction: About 3–5% of Parkinson's disease (PD) patients have a monogenic form of the disease. *LRRK2 and SNCA* are the most prevalent in autosomal dominant forms, and *PRKN* and *PINK1* in recessive ones. Information on Chile's epidemiology is scarce: only 5 studies have assessed pathogenic mutations on Chilean patients, mainly case series. **Objectives:** To describe the minimum prevalence and distribution of monogenic forms of PD in a Chilean cohort. **Methods:** We consecutively assessed PD patients older than 18 years old that consulted at CETRAM movement disorders center from April 2021 to November 2022. Those that fulfilled the United Kingdom Parkinson's Disease Brain Bank Society criteria and agreed to participate were included and evaluated according a structured template. DNA from blood was extracted according to standard protocols and genotyping with Illumina Multi-Ethnic Genotyping Array (MEGA) assay. **Results: We included** 116 PD patients with a mean age of onset 55.2 ± 12.1 years, and 54% were men. Around 35% began before 50 years of age and 36% had positive family history. In 10% of the patients, a pathogenic mutation was found; 4 in *GBA* (p.N370S) and 8 in *LRRK2* (p.G2019S). Twenty five percent of the *GBA* patients and 66% of *LRRK2* had a positive family history, and their mean age of onset was 57.5 ±13.1 for the former and 61.1 ± 7.5 for the latter.

No mutation on *SNCA*, *DJ1*, *VPS35*, *PARKIN* nor *PINK1* were found. **Conclusion:** The minimum prevalence of monogenic PD was 10% with *LRRK2* being the most prevalent one.

Keywords: PARKINSON'S DISEASE, GENETICS, CHILE