

# FREQUENCY OF MONOGENIC PARKINSON'S DISEASE AND PARKINSONISM IN LATIN AMERICA: SYSTEMATIC REVIEW

SAFFIE AWAD Paula<sup>1,2</sup>, TEXEIRA Daniel<sup>3</sup>, CHANA Pedro<sup>1,4</sup>, SCHUMACHER-SCHUH Artur<sup>3,5</sup>

<sup>1</sup> Centro de Trastornos del Movimiento (CETRAM), Santiago, Chile

<sup>2</sup> Clínica Santa María

<sup>3</sup> Serviço de Neurologia, Hospital de Clínicas de Porto Alegre, Porto Alegre, Brazil.

<sup>4</sup> Facultad de Ciencias Médicas, Universidad de Santiago de Chile, Santiago, Chile.

<sup>5</sup> Departamento de Farmacologia, Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil

Corresponding author: Paula Saffie, [psaffie@gmail.com](mailto:psaffie@gmail.com), +56957834785

**Introduction:** Parkinson's Disease (PD) is a complex and heterogeneous neurodegenerative movement disorder. In a minority of cases (3-5%) it is explained by a single pathogenic variant. Most studies have assessed the prevalence of monogenic PD in European or North American cohorts, thus information regarding Latin America's countries is scarce. **Objectives:** To assess the frequency and distribution of monogenic forms of Parkinsonism in Latin America. **Methods:** We performed a systematic literature search using a structured search mechanism in Pubmed, EMBASE, WEB of Science and LILACS until August 2022. We included original articles that reported monogenic forms of parkinsonism in Latin American population, written in English, Spanish or Portuguese. Independent reviewers extracted the data, and discrepancies were assessed by another investigator. **Results:** 74 studies were included from 16 South American countries, with Brazil the one with more publications (50%). 54% were mutational screening studies and 46% case series. A total of 7603 patients were screened for monogenic parkinsonism and 343 of them had a pathogenic mutation (4,5%), being *LRRK2* the most frequent for dominant forms of classical parkinsonism (1,82%) and *PARKIN* for the recessive one (7,31%). Only one patient with SNCA mutation was reported among all studies, and no *VPS35*, *CHCHD2* nor *DJ1* was found. The most frequent atypical parkinsonism gene reported was *SLC20A2*, and *GBA* the one among other diseases with parkinsonism (4,54%). **Conclusion:** *LRRK2* and *PARKIN* are the most common monogenic forms of parkinsonism in Latin America. More robust epidemiologic studies are needed to describe better this underrepresented population.

Keywords: PARKINSON'S DISEASE, MONOGENIC DISEASE, LATIN AMERICA