

Leucine-rich repeat kinase 2 (LRRK2) gene mutations are the most common genetic cause of Parkinson's disease (PD). More than 300 rare LRRK2 variants have been described, with approximately 17 having confirmed or probable pathogenic role in PD. The distribution differs across ethnic groups, but no PD-related LRRK2 pathogenic variant has been described in persons of Black African ancestry within or outside Africa. We previously reported the absence of LRRK2 p.Gly2019Ser mutation in 126 PD and 55 controls from Nigeria. Using Kompetitive Allele Specific Polymerase Chain Reaction, we screened a new cohort of 92 Nigerians with PD and 210 ethnically matched controls for 12 rare LRRK2 variants shown to be pathogenic in other ethnic populations, including p.Gly2019Ser, p.Arg1441His, p.Gly2385Arg, p.Ala419Val, p.Arg1628Pro, p.Pro755Leu, p.Ile2020Thr, and Tyr1699Cys. All were absent in PD and controls, endorsing our previous findings and confirming that rare LRRK2 pathogenic variants reported in Caucasians, Asians, and persons of mixed ancestry are absent in West Africans. Future studies applying next generation sequencing are necessary to explore novel LRRK2 variants indigenous to Black Africans.

Keywords: African; Black; Genes; Genetic; LRRK2; Nigeria; Parkinson's disease.