

Full text open access online (Since 2001)

©  Kamla-Raj IJHG 2023

PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330

Int J Hum Genet, 23(2-3): 140-146 (2023)

DOI: 10.31901/24566322.2023/23.2-3.857

Mutational Analysis in *SNCA* and Chromosomal Aberration in Parkinson's Disease (PD) Patients of Tamil Nadu Population

Neethu Raj¹, Harysh Winster Suresh Babu¹, Kiruthika Balasubramani¹, Ajay Elangovan¹, Priyanka Selvaraj¹, Dhivya Venkatesan, Vimalraj Pachaiyappan¹, Thahoora Aafreen S¹, Bupesh Giridharan², Arul Narayanaswamy³ and Balachandar Vellingiri^{1,4*}

¹*Human Molecular Cytogenetics and Stem Cell Laboratory,
Department of Human Genetics and Molecular Biology,
Bharathiar University, Coimbatore 641 046, Tamil Nadu, India*

²*Natural Products and its Compound Research Laboratory, Department of Forest Science,
Nagaland University, Zunheboto 798 627, Nagaland, India*

³*Disease Proteomics Laboratory, Department of Human Genetics and Molecular Biology,
Bharathiar University, Coimbatore 641 046, Tamil Nadu, India*

⁴*Stem cell and Regenerative Medicine, Translational Research, Department of Zoology,
School of Basic Sciences, Central University of Punjab, Bathinda 151401, Punjab, India*

KEYWORDS Chromosomal Aberration. Parkinson's Disease (PD). Rating Scales. *SNCA*. 22q11.2

ABSTRACT Parkinson's disease (PD) is an age-related disorder which deteriorates dopaminergic neurons that control balance and movement. The study aimed at identifying the chromosomal alterations and *SNCA* mutation in $n=126$ PD subjects with equivalent number of control subjects. The subjects were characterized as late-onset ($n=92$), early-onset ($n=22$) and juvenile ($n=12$). In this study primarily, severity and stages of PD were analysed using the Unified Parkinson's Disease Rating Scale (UPDRS) and Hoehn and Yahr (HY) scale. The UPDRS shows significant values in all the three age groups whereas HY scaling showed significance in late-onset alone. Cytogenetic analysis with 22q11.2 deletion was observed in late-onset subject with higher significance and point mutation in *SNCA* with A53T and A30P was significant in late-onset and early-onset subjects. Therefore, the researchers conclude that genetic alterations have strong correlation with PD and it is necessary for therapeutic researches in PD.