

Screening of Genetic Mutations in Early Onset Parkinsonism Patients: A Family Based Study in Tamil Nadu Population

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ABSTRACT Parkinson's disease (PD) is a progressive motor system disorder which distress several parts of the brain, in particular substantia nigral area that controls balance and movement. The intend of the study was to identify the polymorphism in SNCA (α -Synuclein) and Parkin genes using PCR-RFLP and chromosomal analysis by GTG banding in 23 early onset PD patients who are below the age of 50. The results were analyzed and SNCA polymorphism was observed as missense mutation A53T with G-A transition whereas Parkin is also observed with G-A transition and change in amino-acid S167N. The chromosomal abnormality resulted with 22q11.2 deletion which is an increased risk factor in early-onset PD. Therefore, the resulted polymorphism is the foremost report in Tamil Nadu population and the researchers assure that this would be a unique study from the previous researches in India.