Genetic study of copy number variants in Greek patients with early-onset Parkinson's disease

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Background: Parkinson's disease (PD) is a multifactorial progressive neurodegenerative disorder characterized by motor and non-motor symptoms. PD is the second most prevalent neurodegenerative disease. Monogenic forms account for about 10-15% of the PD cases and often are characterized by early onset (<50 years). Single nucleotide variants (SNVs), insertion or deletion (Indels) and copy number variations (CNVs) in PD-associated genes have been detected across many populations and ethnic groups. This study aimed to estimate the frequency of CNVs in Greek patients with early-onset PD (EOPD).

Patients and Methods: The study cohort consisted of 75 Greek index EOPD patients. All patients had been tested negative for the pathogenic variants p.A30G and p.A53T of the α-synuclein gene (*SNCA*), which represent common causes of PD in the Greek population. Multiplex ligation-dependent probe amplification (MLPA) technique was used for the molecular investigation of CNVs.

Results: MLPA indicated that 1 patient harbored a heterozygous deletion in exon 2 of the *PARK2* gene. It is a sporadic case with age at onset of 30 years and mild disease course. Thus, the frequency of CNVs in Greek patients with EOPD is estimated at 1.3%.

Conclusion

The detection rate of CNVs in the present study is in accordance with other studies. Our findings may contribute in developing the appropriate diagnostic algorithm for genetic testing of Greek patients with EOPD providing timely and accurate diagnosis.

ADDITIONAL INFORMATION

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