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Homozygosity mapping in an Iranian pedigree affected with early onset Parkinson's disease (EOPD) and linkage to chromosome 6

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Parkinson's disease (PD) is a common neurodegenerative disease. Its prevalence is estimated 2% among individuals older than 65 years. Both environmental and genetic factors contribute to the etiology of PD. Five (5) principal PD causing genes were identified: *α-synuclein* (PARK1), *LRRK2* (Leucine-rich repeat kinase 2; PARK8), *PRKN* (parkin; PARK2), *PINK1* (PTEN-induced putative kinase 1; PARK5), and *DJ-1* (PARK7). Mutations in these genes account for disease in a few percent of the patient, suggesting other PD causing genes remain to be identified. The combination of homozygosity mapping and exome sequencing is a powerful and efficient gene finding method applicable to recessive disorders in inbred populations, and the finding of new genes will enhance understandings of diseases pathogenesis. In our study, genome-wide single nucleotide polymorphisms (SNP) genotyping was carried out in an Iranian EOPD family using high density SNP chips. Two (2) affected siblings, 2 unaffected siblings and unaffected parents were genotyped. Homozygous regions common to all affected individuals and absent in non-affected individuals were sought. Disease status in the family is linked to a large homozygous region of 15Mb on chromosome 6. The linked region included 130 genes. Mutation screening of these genes is difficult and costly, so exome sequencing on 2 affected and 2 unaffected siblings was performed and candidate genes in the linked region were analyzed then. Age at onset of symptoms was in the second decade of life, and the mode of inheritance was autosomal recessive. Bradykinesia and rigidity, tremor, eye movement abnormalities including supranuclear gaze palsy, dystonia and bulbar anomalies were reported in 2 affected siblings.

Biography

Afagh Alavi received his BSc in Biology from Shahid Chamran University of Ahvaz, Iran (1998) and MSc and PhD in Molecular Biology from the University of Tehran, Iran (respectively, 2007 & 2013). He spent more than 15 years as a teacher of Biology (1998-2013), and then moved to University of Social Welfare and Rehabilitation Sciences, Tehran, Iran (2013). Since 2013, he serves as an Assistant Professor in the Genetics Research Center in this university. His main areas of research interest lie in the area of genetics and molecular analysis of neuromuscular and muscular disorders.

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