Abstract

Parkinson’s is a common disease (PD) caused by degeneration of dopaminergic neurons in the substantia nigra and other brain areas. Several genes and mutations have been implicated in its pathogenesis, the latter have been identified mainly in the PARK2 gene. We report the evaluation of this gene and of its flanking region in a large family from the southwestern part of Colombia. The parents are first cousins and four out of their ten children were affected at juvenile age. Molecular evaluation included typing of microsatellites (SSTRs) and direct sequencing of the exons of the gene. Our findings showed the presence, in a homozygous manner, of the mutation c.255delA, at exon 2 of PARK2. In addition, it was possible to identify a haplotype carried by both parents, and present in a homozygous manner in the affected children. A high rate of recombinants was observed in the analysed chromosomal region. Mutation c.255delA in PARK2 had been previously reported in families from both France and Spain. Our findings reconfirm the role of the PARK2 gene in the etiology of Parkinson’s disease, in particular of its juvenile form. Furthermore, taking into account that the identified mutation had been previously found in European populations, it is likely that it came into Colombia from that continent. Alternatively, this mutation might have occurred in a recurrent manner in a close ancestor of the studied family. In order to verify both possibilities it would be necessary to test flanking markers of the mutation in both European and Colombian chromosomes carrying it. Such markers could be either STRs, as reported in this study, or SNPs.

Keywords

Genetics of Parkinson’s Disease, Juvenile Parkinson’s Disease, Mutation c.255delA, Park2 Gene.