SNCA POLYMORPHISMS IN NORTHERN SPANISH POPULATION: LOOKING FOR RISK HAPLOTYPES

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Introduction: 1-2% of the global population over 60 years old suffer from Parkinson's Disease (PD). SNCA gene causes Autosomic Dominant PD and several polymorphisms have been associated with PD, especially in the 3'UTR and promoter regions.

Aim: To replicate the association with the disease obtained with several polymorphisms along the SNCA gene in other European populations and GWA's studies and to find risk or protective haplotypes in our population.

Methods: 478 patients and 208 controls were studied for the SNP's rs2619364 (promoter region) and rs6848726 (intron4) and 763 patients and 480 controls for the SNP's rs356165 (3'UTR) and rs11931074 (3'). All people included in the study were Spanish caucasian. To genotype the rs2619364, rs6848726 and rs11931074 we used TaqMan SNP's Genotyping Assays on an ABI7500 Real Time PCR System. The rs356165 was genotyped by the restriction enzyme Tail.

Results: We found 2 LD blocks; haplotypes block1: AC, AT, GC and haplotypes block2: CT, TG, CG. Block2 haplotypes CG and CT are overrepresented in patients. Allele C (rs356165) and T (rs11931075) are associated with risk of suffering PD and allele C is also associated too with risk of EO.

Conclusions: It seems that, in our population alleles C and T of the rs356165 and rs11931074 are associated with risk of PD and allele C with risk of EO too. The haplotypes CT and CG are more frequent in patients than in controls meanwhile the haplotype TG seems to be protective.