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Predictive genetic risk test in cerebrovascular disorders

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The study of variations in genes involved in the different events that trigger the atherogenic process, such as lipid metabolism (modification of LDL-cholesterol), endothelial function and hypertension, immune response (recruitment of macrophages and foam cell formation) and stability of atherosclerotic plaques (thrombosis), establishes the susceptibility, risk or probability of an individual for suffering arteriopathy and vascular disorders. A total of 2455 cases over 50 years of age were genotyped for a panel of 19 SNPs in 15 genes encoding for proteins involved in the atherogenic process. This study shows the relevance of polymorphisms in APOB (odds ratio (OR), 1.17; 95% confidence interval (95% CI), 0.74-1.85), APOC3 (OR: 1.33; 95%, CI: 0.82-2.17) and APOE (OR: 1.75; 95%, CI: 1.09-2.80) as genetic risk markers for hypercholesterolemia and polymorphisms in ACE (OR: 1.68; 95%, CI: 0.32-8.77) and AGT (OR: 1.74; 95%, CI: 0.97-3.14) for hypertension. Our results also show the transversal importance of pro-inflammatory cytokines in different stages of atherogenesis with special relevance of IL6 (OR: 1.39; 95%, CI: 0.56-3.49) and TNF (OR: 1.40; 95%, CI: 0.92-2.15) related to hypercholesterolemia and hypertension. The set of markers involved in this genetic risk panel are integrated in multigenic panels as development of these diseases is not caused by a single gene but by the interaction of a number of genes. This genetic panel study differentiates genetic polymorphisms or variations in the DNA sequence, which is involved in the development, the prognosis and the evolution of cerebrovascular disorders and represents a key tool in medical practice.

Biography

Juan C Carril is the Director of the Genomics and Pharmacogenomics Department at EuroEspes Biomedical Research Center, Institute of Medical Science and Genomic Medicine, Corunna, Spain. He has received his PhD from Santiago de Compostela University in 2000 with the thesis entitled “Genetic structure and profile of the populations of the Iberian Peninsula by means of markers (STRs and SNPs) of the human Y-chromosome”. He has published more than 40 scientific publications in the fields of population genetics, forensic genetics, genetic epidemiology and pharmacogenetics and over 40 papers at national and international conferences.

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