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Risk polymorphisms associated with dementia in the Spanish population

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According to the WHO, cerebrovascular and neurodegenerative disorders affect one billion people around the world. A number of these disorders are characterized by the onset of dementia. Alzheimer's disease (AD) is the major cause of dementia in Western countries, affecting 45-60% of the population, followed by vascular dementia (VD) and mixed dementia (MD) with prevalence of 30-40% and 10-20%, respectively. Population studies evaluate the genetic risk, i.e., the probability of an individual carrying a specific disease-associated polymorphism. Identification of risk polymorphisms is essential for an accurate diagnosis or prognosis of a number of pathologies. The aim of this study was to characterize the influence of risk polymorphisms associated with dementia in a large population of Spanish individuals affected by a variety of brain and vascular disorders as well as metabolic syndrome. We performed a cross-sectional study in 4415 individuals from a widespread regional distribution in Spain (48.15% males and 51.85% females) with mental, neurodegenerative, cerebrovascular and metabolic disorders. We evaluated polymorphisms in genes associated with the accumulation of A β and the cleavage of the A β precursor protein such as A2M and PSEN1, respectively and also with APOE, which is one of the most prevalent risk genes in AD, especially in those individuals harboring the APOE- ϵ 4 allele, whereas the APOE- ϵ 2 variant is protective for dementia. Allele and genotype frequencies of those polymorphisms in our population were compared with representative Spanish and European populations. Risk polymorphisms in PSEN1 and APOE (specially the APOE- ϵ 4 allele) are representative of our population as compared to reference data of Spanish and European individuals. These data indicate that risk polymorphisms in PSEN1 and APOE- ϵ 4 may be the trademark of AD in the Spanish population.

Biography

Oscar Teijido Hermida is the Head of the Medical Epigenetics Department at EuroEspes Biomedical Research Center, Institute of Medical Science and Genomic Medicine, Corunna, Spain. He has received his PhD from the University of Barcelona, Spain in 2007. During his scientific career in University of A Corunna, Spain, University of Barcelona, Spain, New York University, USA and The National Institutes of Health, USA, he achieved more than 20 scientific publications in the molecular genetics, biochemistry and physiology fields and presented his work in more than 25 international conferences.

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