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P22- APOE allele frequency in late onset Alzheimer’s disease patients from Iberia

Ricardo González1,2, Sandra Martins1,2, Luis Durães3, Patrícia Sousa3, Manuel Figueruelo4, María Rodríguez4, Carmen Pita4, Miguel Arenas5, Luis Álvarez6, Roberto Hornero7, Carlos Gómez7, Nádia Pinto1,2,8, Alexandra M Lopes1,2, Iva Gomes1,2

1- Instituto de Patologia e Imunologia Molecular da Universidade do Porto (IPATIMUP), Portugal; 2- Instituto de Investigação e Inovação em Saúde (i3s), Universidade do Porto, Portugal; 3- Associação Portuguesa de Familiares e Amigos de Doentes de Alzheimer, Portugal; 4- Asociación de Familiares y Amigos de Enfermos de Alzheimer y otras demencias de Zamora, Spain; 5- Department of Biochemistry, Genetics and Immunology, University of Vigo, Spain; 6- TellmeGen, Valencia, Spain; 7- Biomedical Engineering Group, University of Valladolid, Spain; 8- Centro de Matemática da Universidade do Porto, Portugal

Aims/Context: Alzheimer’s Disease (AD) is a progressive neurodegenerative disease associated with cognitive decline. It is one of the most severe brain disorders affecting the elderly population, being secondary to the increase of life expectancy. Although multi-factorial, the primarily genetic risk factor for late-onset AD is the Apolipoprotein E (APOE) ε4 allele. The APOE gene encodes a 299 amino acid protein that plays a key role in the transport and metabolism of plasma cholesterol and triglycerides, as well as in injury repair in the brain. APOE isoforms differ in the amino acids 112 and 158, which affect its structure, influencing its capability to bind to lipids and receptors, leading to the onset of AD. Due to the preponderant role in AD pathology, screening the APOE gene can facilitate AD diagnostics. Methods: DNA was extracted from saliva samples from 95 patients with clinical diagnostic of AD from Iberia (Northern Portugal and Castile and León). After whole genome amplification, we sequenced the APOE locus by Sanger sequencing to analyze SNPs rs429358 and rs7412, therefore assessing APOE alleles in our cohort of AD patients. Results: We observed 34% of individuals carrying the risk APOE ε4 allele, whereas the more common ε3 allele was present in 59% of the patients. The frequency of ε2 allele (associated to a decreased disease risk) was estimated in our sample set with a frequency of 7%. Conclusions: This work is a preliminary study about the frequency distribution of the APOE polymorphic ε2, ε3 and ε4 alleles in a cohort of late-onset patients of AD from Northern Iberian regions. The genetic characterization of APOE provides a forecast on the landscape of AD risk in these regions based on the haplotype data obtained from APOE alleles at SNPs rs429358 and rs7412.