

FACULTAD CIENCIAS BÁSICAS Y BIOMÉDICAS

MAESTRÍA EN GENÉTICA

Análisis de asociación genética caso–control entre variantes de los genes *APOE*, *ABCA7*, *CLU* y *TREM2* y el deterioro cognitivo leve en población del Atlántico.

Línea de investigación:

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Resumen

El Deterioro Cognitivo Leve (DCL) es una condición intermedia entre el envejecimiento normal y la demencia, de origen multifactorial. El objetivo fue identificar variantes genéticas en los genes *CLU*, *ABCA7*, *APOE* y *TREM2* en pacientes con DCL y controles sanos del Departamento del Atlántico. Se realizó un estudio de casos y controles en 45 participantes (26 casos y 19 controles) mediante secuenciación genómica completa (DNBSEQ™) y análisis con BWA, GATK, ANNOVAR y PLINK. El PCA mostró una estructura genética mestiza con predominio de ancestría latinoamericana. El SNP rs7748513 en *TREM2* se asoció con DCL (OR=8,81; p=0,017). En *ABCA7*, el indel rs3833881 mostró efecto protector (OR=0,4; p=0,0365). En *CLU*, cuatro SNPs se asociaron con menor riesgo. El alelo *APOE* ε4 fue más frecuente en casos con puntajes bajos de MoCA. Los resultados respaldan la contribución genética al DCL en esta población.

Palabras clave: Deterioro cognitivo leve, genética, *APOE*, *TREM2*, *ABCA7*, *CLU*, bioinformática.

Abstract

Mild Cognitive Impairment (MCI) is an intermediate condition between normal aging and dementia with multifactorial etiology. This study aimed to identify genetic variants in *CLU*, *ABCA7*, *APOE*, and *TREM2* in patients with MCI and healthy controls from the Department of Atlántico, Colombia. A case–control study was conducted with 45 participants (26 cases and 19 controls) using whole-genome sequencing (DNBSEQ™) and bioinformatic analysis with BWA, GATK, ANNOVAR, and PLINK. PCA showed a mixed genetic structure with predominant Latin American ancestry. The *TREM2* rs7748513 variant was significantly associated with MCI (OR=8.81; p=0.017). In *ABCA7*, the rs3833881 indel showed a protective effect (OR=0.4; p=0.0365). Four SNPs in *CLU* were associated with reduced risk. The *APOE* ε4 allele was more frequent in cases with lower MoCA scores. These findings support the genetic contribution to MCI and the relevance of genomic approaches for early detection.

Keywords: Mild Cognitive Impairment, genetics, *APOE*, *TREM2*, *ABCA7*, *CLU*, bioinformatics.

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