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Association study between late onset Alzheimer's disease and genes implicated in the Aβ metabolism in Mexican patients

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Background

Alzheimer Disease (AD) is clinically characterized by global, progressive and irreversible loss of mental faculties. Neuropathologically is characterized by neurofibrillary tangles and neuritic plaques; composed mainly by Amyloid β-peptide (Aβ). Generation, aggregation and degradation of Aβ represent three important steps to be considered in the study of the pathological mechanisms implicated in AD. Several genes have been suggested as implicated in each of these processes: Beta-site amyloid-precursor protein cleaving enzyme (BACE) in generating, Apolipoprotein E (APOE) in aggregation, and urokinase-type plasminogen activator (PLAU), involved in degradation; have been exhaustively documented [1]. This is the first study in Mexican population that analyzes genetic risk factors related to AD.

Materials and methods

A case-control study was design to evaluate the possible association between candidate genes involved in these three processes with AD. Data collection was performed from 49 patients with AD and 50 controls. We analyzed alleles and genotype distributions for APOE (ε2/ε3/ε4), 2 APOE promoter polymorphisms –219 G/T and –491 T/A, 1SNP located in exon 5 of the BACE-1 gene (G/C), and one (C/T) polymorphism in exon 6 of the PLAU gene.

Results

We found different allele and genotype frequencies for all SNPs analyzed between cases and controls with exception for –491 T/A. Association was found for the APOE ε4 allele (OR =2.42), –219 TT genotype (OR =1.77), CC genotype of BACE-1 (OR =1.88) and TT genotype of PLAU (OR =2.10).

Conclusions

These data suggest a genetic association between APOE (ε4),–219TT, BACE-1 (CC), and PLAU (TT) genotypes with AD in Mexican population.

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References

1. Tanzi RE, Bertram L: **Twenty years of the Alzheimer's Disease Amyloid Hypothesis: A Genetic Perspective.** *Cell* 2005, **120(3):**545-55.
2. Wang JC, Kwon JM, Shah P, Morris JC, Goate A: **Effect of APOE genotype and promoter polymorphism on risk of Alzheimer's disease.** *Neurology* 2000, **55:**1644-1649.
3. Gold G, Blouin JL, Herrmann FR, Michon A, Mulligan R, Duriaux Sail G, Bouras C, Giannakopoulos P, Antonarakis SE: **Specific BACE1 Genotypes Provide Additional Risk for Late-Onset Alzheimer disease in APOE ε4 Carriers.** *Am J Med Genet B Neuropsychiatr Genet* 2003, **119:**44-47.
4. Ertekin-Taner N, Ronald J, Feuk L, Prince J, Tucker M, Younkin L, Hella M, Jain S, Hackett A, Scalin L, *et al.*: **Elevated amyloid beta protein (Aβ42) and late onset Alzheimer's disease are associated with single nucleotide polymorphisms in the uroki-**

nase-type plasminogen activator gene. *Hum Mol Genet* 2005, **14:**447-460.