

THE DOPAMINE β -HYDROXYLASE C-1021T POLYMORPHISM IN ALZHEIMER'S DISEASE

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Alzheimer's disease (AD) is a genetically complex neurodegenerative disorder. Degeneration of the locus coeruleus and decreased cortical levels of noradrenaline (NA) were detected in AD. Dopamine β -hydroxylase (DBH) catalyses the conversion of dopamine to NA, and DBH activity were reduced in post-mortem hippocampus and neocortex in AD. The aim of our study was to examine the possible role of the DBH C-1021T polymorphism (rs1611115) and its synergistic effect with the APOE E4 allele in the development of AD.

DNA sample was collected from 246 Hungarian AD probands and from 202 elderly, cognitively intact, ethnically matched controls. The clinical diagnosis of AD fulfilled the criteria for NINCDS-ADRDA. The genetic analyses were performed by PCR-RFLP.

The DBH C/C genotype occurred more frequently in the AD as compared to the HC group, however, the difference did not reach statistical significance ($p=0.086$). Given the relatively low occurrence of the T/T genotype, the statistical analysis was also performed by the presence or absence of the T allele. The C/C genotype carriers had a slightly significantly increased risk for AD (OR=1.547, 95%CI: 1.027-2.329, $p=0.037$) considering the T allele carriers as reference category. Logistic regression analysis revealed no interaction between the DBH and APOE polymorphisms ($p=0.392$).

Our results suggest that the C/C genotype of the DBH rs1611115 polymorphism may confer risk for developing AD. We failed to detect an epistasis between the DBH and the APOE polymorphisms in the development of AD. This work was supported by a grant from TÁMOP-4.2.2A-11/1/KONV-2012-0052.