

The association of beta-site APP cleaving enzyme (BACE) C786G polymorphism with Alzheimer,s disease

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摘要

Abstract

The deposition of amyloid beta-peptide (Abeta) plays a crucial role in the pathogenesis of Alzheimer's disease (AD). Beta-site APP cleaving enzyme (BACE) is the rate-limiting enzyme in the Abeta formation. BACE mutations/polymorphisms may be associated with AD. We searched the BACE coding region mutations/polymorphisms of cDNA in 25 AD patients and 100 healthy controls by single-strand conformational polymorphism. A polymorphism at BACE coding region was identified and confirmed to be 786C/G polymorphism by nucleotide sequencing. Based on these findings, we investigated the association of this polymorphism with the occurrence of AD by PCR-RFLP. A total of 98 AD patients along with 138 controls were recruited in the present study. The allele frequencies of the 786C/G polymorphism were 0.622 for C and 0.378 for G in AD. In controls, the C and G allele frequencies were 0.691 and 0.309, respectively. No significant association of this polymorphism with the occurrence of AD can be established. Larger sample size may be necessary to identify other potential mutations/polymorphisms among BACE gene.