Association analysis of exonic variants of the gene encoding the GABAB receptor and idiopathic generalized epilepsy

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The gene encoding the GABAB receptor (GABABR1) maps close to the HLA-F locus on chromosome 6p21.3 in the same region to which a major susceptibility locus for common subtypes of idiopathic generalized epilepsy (IGE), designated as EJM1, has been localized. Moreover, animal models suggest that the GABAB receptor plays a critical role in the epileptogenesis of absence seizures. Accordingly, the present association study tested the candidate gene hypothesis that genetic variants of the human GABABR1 gene confer susceptibility to common subtypes of IGE. Three DNA sequence variants in exons 1a1, 7, and 11 of the GABABR1 gene were assessed by PCR-based restriction fragment length polymorphisms in 248 unrelated probands of German descent, comprising 72 patients with juvenile myoclonic epilepsy (JME), 46 patients with idiopathic absence epilepsy (IAE), and 130 control subjects without a history of epileptic seizures and lack of generalized spike-wave discharges in their electroencephalogram. The results revealed no evidence for an allelic association of any of the GABABR1 sequence variants with either JME or IAE (P >0.18). Thus, we failed to demonstrate that any of the three exonic GABABR1 variants themselves, or other so-far unidentified mutations, which are in strong linkage disequilibrium with the investigated variants, are involved in the pathogenesis of common IGE subtypes.

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