Relevant research between rs2126152 polymorphism in SCN1A gene and epilepsy with febrile seizures plus

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View from specialist: It is creative, and of certain scientific and educational value.

[ABSTRACT] Objective: To screen the SCN1A in Chinese patients diagnosed as generalized epilepsy with febrile seizures plus (GEFS⁺) and to explore the possible relationship between SCN1A and GEFS⁺.

Methods: After collection of blood samples from 60 patients with GEFS⁺ and 104 normal control subjects, all 26 coding exons and introns relevant to mRNA splice of SCN1A were amplified with PCR technology and then sequence analysis was performed. Results: No SCN1A mutation was detected but a single nucleotide polymorphism (SNP) was found; EXON14-37A>C (rs2126152). Genotypes and allelic frequencies for the SNP in both groups were not significantly different (P > 0.05). Conclusion: Our results indicate that the SNP EXON14-37A>C of SCN1A gene might not be one of the susceptibility factors for GEFS⁺.

[KEY WORDS] Epilepsy; SCN1A; Mutation; Single nucleotide polymorphism