Two single nucleotide polymorphisms in SCN1A of Chinese GEFS+ patients

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Objective: Recent studies have provided evidence that mutations in SCN1A represent the most frequent cause of generalised epilepsy with febrile seizures plus (GEFS+). Whether the variation of SCN1A single nucleotide polymorphisms (SNPs) in people represented sensitivity to GEFS+ had not been known. This study aimed to evaluate the correlation of SCN1A SNPs and GEFS+ in China.

Methods: A total of 32 PCR amplicons including the whole exon and their flanking intronic splice site have been screened by denaturing high performance liquid chromatography (DHPLC). For those with abnormal elution peak, direct sequence analysis was performed. Exons 7 -21 C>T and c.3199G>A; T10 67A of SCN1A were analysed in 168 Chinese with GEFS+ and 127 normal control subjects. SPSS 14.0 was used to determine the difference between the two groups.

Results: The results showed that genotypes and allelic frequencies for these two SCN1A gene polymorphisms in both groups were not significantly different. (Figure 1)

Conclusion: Our study suggests that the two SNPs might not be one of the susceptibility factors for GEFS+. The impact of other polymorphisms in SCN1A on the development of GEFS+ merits further research.

Figure 1. Segregation analysis of the Exons 7 -21 C>T and c.3199G>A; T 1067A mutation by DHPLC and direct sequencing.

A: DHPLC profiles of the 1A7-PD125 and 1A16-PD127 had abnormal profiles.
B: DNA sequences of the 1A7-PD125 and 1A16-PD127. 1A7-PD125 had the SNP C>T and 1A16-PD127 had the SNP A>G.