

Frequency Variant rs1051740 T337C in Carbamazepine Carbide Enzyme Enzyme (EPHX1) Genes on Healthy Volunteers of Javanese Women in Yogyakarta

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ABSTRACT

Carbamazepine (CBZ) is a first-line antiepileptic drug belonging to a class of drugs with a narrow therapeutic index. Women are having hormonal factors that can increase seizure seizures. Gen Epoxide Hydrolase 1 (EPHX1) is a microsomal epoxide hydrolase (mEH) enzyme coding gene for CBZ metabolism which is reported to have variations in rs1051740 with a Minor Allele Frequency (MAF) value of 0.3133. This study aims to examine the variance frequency of rs1051740 T337C gene EPHX1 encoding CBZ metabolism enzyme in healthy female volunteers of Javanese Tribe in Yogyakarta Special Province province. Methods of research conducted DNA isolation on blood samples of healthy volunteers women Javanese as much as 50 subjects test by silica gel method, then do DNA amplification by using Polymerase Chain Reaction (PCR) method. Primer used forward-5'GATAAGTTCCGTTTCACCC-3 'and reverse 5'TGTTCTGCCTAGCTCTAA-3'. PCR conditions were initial denaturation of 94 ° C for 3 minutes followed by 32 cycles of denaturation at 94 ° C for 30 seconds, the annealing temperature of 55 ° C for 25 seconds and extension of 72 ° C for 1 minute and the final extension at 72 ° C for 5 minutes. PCR 368bp product was optimized for Length Polymorphism Restriction Fragment (RFLP) by using an EcoRV restriction enzyme, then screening all PCR products to ensure that the enzyme was not able to cut the target sequence. Continued confirmation using the Sequencing method in three randomly selected samples. The results of the study showed that there were no gene variations when done using the RFLP method. There are two out of three samples of nucleotide sequence confirmed by Sequencing DNA method is heterozygous (CT). It is recommended to do research with Real-Time PCR method.

Keywords: *EPHX1*, rs1051740, polymorphism, Carbamazepine, DNA, Javanese.