Allele Frequency Distributions of the Drug Transporter Genes Carbamazepine : *ABCB1* rs1045642 C>T on Healthy Javanese Men in Special Region of Yogyakarta

Sandra Vitriyani
Department of Pharmacy

ABSTRACT

Carbamazepine (CBZ) is the main choice of epilepsy treatment, especially in the type of symptomatic epilepsy focal (partial) experienced by many males. The presence of variation in patient response to CBZ use may be due to genetic variation in the main CBZ transporter P-glycoprotein encoded by the *ABCB1* gene. *Single Nucleotide Polymorphisms* (SNP) *ABCB1* rs1045642 C> T as the cause of excessive expression of P-glycoprotein in the blood brain barrier that contributes to CBZ resistance. Polymorphism with MAF value of 0.3952 at rs1045642 may be possible to affect varieties of CBZ pharmacokinetics. Research on the frequency of *ABCB1* gene rs1045642 has not been done in the population of the Javanese tribe with the largest population in Indonesia. This study aims to study the frequency of *ABCB1* gene rs1045642 C> T in healthy male Javanese volunteers in Special Region of Yogyakarta. The study was conducted on 50 healthy male volunteers who met the inclusion criteria. The genetic variation on the target SNP was analyzed by PCR-RFLP method using *forward* primer 5'-TCG TGA AG-TGG TGA TGA TGA TGA TGA TGA 3 AEG-3 ', and *reverse* primer 5'-ACA TTA GGC AGT GAC TCG ATG AAG GCA-3', and the *Mbo*I restriction enzyme that will bypass the |GATC sequence with a *wild-type* target (Genotype CC) at 192 bp. The data were analyzed using univariate test and presented in percentage proportion for each genetic variant. The allele frequencies C and T obtained in this study were 57% and 43%. The allele frequency obtained is almost similar to the results of research in Japan and Turkey. Further studies are recommended to examine the correlation between these polymorphisms with the variability of pharmacokinetic profiles and Carbamazepine clinical response.

Key words: Carbamazepine, P-glycoprotein, PCR-RFLP, rs1045642.