

**Detection On Genetic Variaton In Gene Encoding Valproic Acid
Metabolizing Enzyme *UGT1A6 rs2070959 A>G* Healthy Volunteers of
Javanese People In Indonesia**

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ABSTRACT

Epilepsy is a seizure disease which has a risk of death three times higher in patients of epilepsy compared to normal people and also has the risk *Sudden Unexpected Death in Epilepsy* (SUDEP), which causing sudden death due to epilepsy. As many as 70% of people with epilepsy can be controlled with an *Antiepileptic drugs* (OAE). One of the first line OAE agents in epilepsy therapy is Valproic Acid (VA). Valproate monitoring is indispensable for its variability occurrence as the result of drug level taken and drug dosing between each individual; also monitoring for variability occurrence happens by gender, age, and their genetics. The occurrence of polymorphism genetic in metabolizing gene of Valproic Acid (*UGT1A6*) as a gene encoding the glucoronidase enzyme is resulting different drug level in plasma which may effect therapeutic results. This study aimed to determine the frequency of genetic variations in the *UGT1A6 rs2070959 A>G* in Javanese population. The sample used in this study were stored as DNA specimen from 100 healthy respondent who included the inclusion criteria. The analysis was using by *Polymerase Chain Reaction-Restriction Fragment Length Polymorphism* (PCR-RFLP) method with *forward primary* 5'-CTT TAA GGA GAG CAA GTT TGA TG-3' and *reverse primary* 5'-CCA CTC GTT GGG AAA AAG TC-3'. The DNA from PCR amplification digested by *NsiI* enzyme which cut on ATGCA|T sequences from the DNA template resulted *wild type* AA (128 and 87 bp) of genotypic variant. Frequency variant of *UGT1A6* gene *rs2070959 A>G* can not be shown in this research. Some trials had been executed for optimizing the cutting by increasing amount of enzyme, time and temperature of incubation, also DNA concentration. The failure of digestion process in optimizing PCR-RFLP was confirmed by *Direct Sequencing* method on two random samples. Sequencing result showed that there were samples that had genotype variants of AG (heterozygous mutants) which had the results of cuts, 128, 87 and 215 bp. Therefore, further research is needed related to the detection of variations of the *UGT1A6* gene *rs2070959 A>G* using restriction enzymes other then *NsiI* in Javanese population.

Kata kunci : *Valproic Acid, Polymorphisms, UGT1A6 rs2070959, PCR-RFLP, Javanese population.*