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Screening for HLA-B*1502 Polymorphism in Febrile Seizure Predicted Lead to Epilepsy

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Mutation in neuronal sodium channel α-1-subunit gene (SCN1A) and neuronal sodium channel β-1-subunit gene (SCN1B) has been linked with forms of generalized epilepsy with febrile seizure plus (GEFS+) and epileptic infantile syndrome like severe myoclonic epilepsy of infancy (SMEI) (Mulley et al., 2005; Scheffer et al., 2007). Since this idiopathic epilepsy typically begins with prolonged febrile seizures (FS) in the first year of life, therefore febrile seizure patient with mutation in SCN1A has a high risk to develop epilepsy on their later life (Dube et al., 2009). Carbamazepine (CBZ) has been known as the most common anti-epileptic drug which can cause Steven-Johnson Syndrome (SJS) and toxic epidermal necrolysis (TEN) in patients with HLA-B*1502 polymorphism. Since the Javanese population have 16,67% of these allele, studying the presence of these allele in patients predicted epilepsy is important.

Furthermore, this study was intended to develop a PCR-based diagnostic protocol to screen HLA-B*1502 polymorphism in epileptic patients to prevent SJS/TEN by carbamazepine. Focusing on epileptic predicted patients, HLA-B*1502 genotyping by sequence specific primer (SSP)-PCR was performed on 31 repeated FS patients with mutation in SCN1A and SCN1A/SCN1B gene.

The result show that the HLA-B*1502 polymorphism was detected in 14 (45,2%) individuals including 8 cases related to mutation SCN1A gene and 6 to SCN1A/SCN1B gene. It illustrates that HLA-B*1502 allele is frequent in these patients. It can thus be suggested that detection of this allele should be done before epilepsy treatment. Later, patients with this allele should avoid CBZ to prevent SJS/TEN during drug administration.

*Keywords: HLA-B*1502; Steven-Johnson syndrome; toxic epidermal necrolysis; epilepsy; febrile seizure; SCN1A mutation; SCN1B mutation.*