

Significant genotype difference in the CYP2E1 PstI polymorphism of indigenous groups in Sabah, Malaysia with Asian and non-Asian populations

Abstract

CYP2E1 PstI polymorphism G-1259C (rs3813867) genotype distributions vary significantly among different populations and are associated with both diseases, like cancer, and adverse drug effects. To date, there have been limited genotype distributions and allele frequencies of this polymorphism reported in the three major indigenous ethnic groups (KadazanDusun, Bajau, and Rungus) in Sabah, also known as North Borneo. The aim of this study was to investigate the genotype distributions and allele frequencies of the CYP2E1 PstI polymorphism G-1259C in these three major indigenous peoples in Sabah. A total of 640 healthy individuals from the three dominant indigenous groups were recruited for this study. Polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) at G-1259C polymorphic site of CYP2E1 gene was performed using the Pst I restriction enzyme. Fragments were analyzed using agarose gel electrophoresis and confirmed by direct sequencing. Overall, the allele frequencies were 90.3% for c1 allele and 9.7% for c2 allele. The genotype frequencies for c1/c1, c1/c2 and c2/c2 were observed as 80.9%, 18.8%, and 0.3%, respectively. A highly statistical significant difference ($p > 0.001$) was observed in the genotype distributions between indigenous groups in Sabah with all Asian and non-Asian populations. However, among these three indigenous groups, there was no statistical significant difference ($p > 0.001$) in their genotype distributions. The three major indigenous ethnic groups in Sabah show unique genotype distributions when compared with other populations. This finding indicates the importance of establishing the genotype distributions of CYP2E1 PstI polymorphism in the indigenous populations.