

Analysis of T344C genetic polymorphism of CYP11B2 gene in Malaysian end stage renal disease subjects.

Abstract

The T344C polymorphism of aldosterone synthase CYP11B2 gene at promoter region was vastly studied in various populations with conflicting results in relation to End-stage Renal Disease (ESRD). In this study, researchers aimed to know the association of T344C polymorphism of CYP11B2 gene in Malaysian ESRD subjects. This study involved 165 Malaysian ESRD subjects and 165 healthy individuals as control subjects. Using commercially available kits, genomic DNA was extracted from the subjects using their buccal cells and the blood. The 152 bp product of CYP11B2 gene polymorphism were amplified by Polymerase Chain Reaction (PCR) and digested with Hae III restriction enzyme using Restriction Fragment Length Polymorphism (RFLP) method. The restricted fragments were separated by metaphor agarose gel electrophoresis and showed 152 bp represent TT allele (wild type), 152 and 97 bp represent the TC allele (heterozygous) and 97 and 56 bp represent CC allele (mutant type) as the genotypes for T344C polymorphism. The frequency of TT, TC and CC genotypes of T344C in CYP11B2 gene in ESRD subjects were 98 (59.39), 60 (36.36%) and 7 (4.24%) while 92 (55.76), 70 (42.42) and 3 (1.82%) were found in control subjects, respectively. The genotypic and allelic frequencies of T344C polymorphism of CYP11B2 gene show no significant differences as compared to control subjects ($p>0.05$). The T allele of T344C polymorphism of CYP11B2 gene might not be considered as a possible genetic marker or predisposing risk factor for ESRD in Malaysian subjects. However, this study has to be further continued with more subjects to confirm the association of CYP11B2 gene mutation with ESRD.

Keyword: Polymorphism; T344C; CYP11B2<ESRD; DNA; Malaysia.