Association between genetic polymorphisms of human MDR1 Exon 21 and 26 in Korean and Vietnamese populations

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The frequencies of two closely linked polymorphisms in MDR1 gene, C3435T and G2677T/A, were evaluated in 528 healthy Koreans and compared with those of 102 Vietnamese. There was no significant difference in frequency of C3435T mutation (p=0.3) between Koreans (T allele frequency, 39.5%) and Vietnamese (T allele frequency, 34.1%). Interestingly, however, G2677T/A mutations revealed significant ethnic difference between Korean (T allele frequency, 42.4%; A allele frequency, 14.6%) and Vietnamese (T allele frequency, 36.8%; A allele frequency, 7.4%) population at p<0.05. While the frequency of G2677T/A variations in Vietnamese was comparable to those of Chinese, Malay and European Americans, the frequency of Koreans was significantly different from these ethnic groups. The overall SNP distribution in a Korean population was similar to that of Japanese. We also analyzed the association between C3435T mutation and G2677T or A mutation. While G2677T mutation was found to be closely associated with C3435T mutation, G2677A mutation was not linked to C3435T mutation. In good agreement with this finding, the ethnic variation of G2677A mutation was different from that of G2677T mutation. These results suggest that the introduction of G2677A conversion into MDR1 gene seems to be independent of G2677T and C3435T mutations and this mutation adds more complexity in addition to two closely linked mutations, C3435T and G2677T, to predict the clinical relevance of MDR1 genotypes and drug disposition.