

Abstract

As part of a project on environmental disasters in minority populations, this study aimed to evaluate differences in the sequence of *N-acetyltransferase 2* (*NAT2*) as a metabolic susceptibility gene in yet unexplored ethnicities. Eight single-nucleotide polymorphisms (SNPs) in the *NAT2* coding region and a variant in the 3' flanking region were analysed in 290 unrelated Kyrgyz and 140 unrelated Romanians by SNP-specific polymerase chain reaction analysis. The variants 341C, 481T and 803G were less and 857A more prevalent in Kyrgyz ($p < 0.0001$). The variant at site 857 indicates Asian descent. 282C>T and 590G>A showed no significant variation by ethnicity. 364G>A and 411A>T turned out to be monomorphic. Database comparisons of the *NAT2* minor allele frequencies showed that Romanians belong to Caucasians and Kyrgyz are in-between Caucasians and East-Asians. The distributions of predicted haplotypes differed significantly between both ethnicities where the Kyrgyz showed a higher diversity. The haplotype without the mutations under study was more common in Kyrgyz (40.1% in Kyrgyz, 29.3% in Romanians). Accordingly, the imputed slow acetylator phenotype was less prevalent in Kyrgyz (35.2 % versus 51.4 % in Romanians). We found pronounced ethnic differences in *NAT2* haplotypes with yet unknown impact on the health risks for environmental or occupational exposures in minority populations.