

Abstract

The human beta-2 adrenergic receptor (beta2AR) is responsible for the binding of endogenous catecholamines and their exogenously administered agonists and antagonists. Three functional polymorphisms in codons 16, 27 and 164 have been described which have clinical importance for several diseases, including asthma, hypertension, heart failure, cystic fibrosis and obesity, as well as response to beta-agonist therapy. These were evaluated in 726 individuals from 8 distinct ethnic populations (Chinese, Filipino, Southwest Asian, Saudi, Ghanaian, Kenyan, Sudanese, and European from Scotland). The results show that most haplotypes are shared among all populations, yet there are marked differences in their frequency distributions geographically. The genetic distance tree is different from standard human population distance trees, implying a different mode of evolution for this locus than that for human population gene-flow history. The multilocus frequency differences between the observed clusters of populations correspond to historical haplotype groupings that have been found to be functionally different with respect to multiple medically related phenotypes. Further studies are needed to see if functional relationships are the same across populations.