

Characterization of genetic variation and natural selection at the arylamine *N*-acetyltransferase genes in global human populations

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Abstract

Functional variability at the arylamine *N*-acetyltransferase genes is associated with drug response in humans and may have been adaptive in the past owing to selection pressure from diet and exposure to toxins during human evolution. **Aims:** We have characterized nucleotide variation at the NAT1 and NAT2 genes, and at the NATP1 pseudogene in global human populations, including many previously under-represented African populations, in order to identify potential functional variants and to understand the role that natural selection has played in shaping variation at these loci in globally diverse populations. **Materials & methods:** We have resequenced approximately 2800 bp for each of the NAT1 and NAT2 gene regions, as well as the pseudogene NATP1, in 197 African and 132 non-African individuals. **Results & conclusion:** We observe a signature of balancing selection maintaining variation in the 3'-UTR of NAT1, suggesting that these variants may play a functional role that is currently undefined. In addition, we observed high levels of nonsynonymous functional variation at the NAT2 locus that differs amongst ethnically diverse populations.