Commentary

Genetic Epidemiology: the Value of Population Differences

Maria Elena Martinez, Senior Editor

In this issue of the journal, Rabstein et al. (1) report data on the differences in prevalence of nine single nucleotide polymorphisms in the *N-acetyltransferase* (*NAT2*) gene between two populations, one of European descent and the other from Central Asia. Investigators also provide predicted haplotypes for these groups. Consequently, *NAT2* acetylator phenotype prevalence differences were also shown between the two populations. These data suggest that ethnic variations of polymorphisms should be evaluated in detail and differences should be incorporated into investigations of these susceptibility variants.

Because disease genes may be geographically restricted, knowledge of individual ancestry is an important consideration in research studies. In trying to develop a detailed understanding of heritable variation in the human genome, it is necessary to characterize genetic variation among different population groups. These groups may be defined by racial, ethnic, or geographic characteristics. Because race and ethnicity are poorly defined and frequently serve as surrogates for genetic, cultural, social, and environmental factors (2), it is important to understand variability within and between these groups when assessing disease risk. Although it has been shown that >95% of genetic variation is shared across populations and that <10% is specific to a single population (3), it is also well known that prevalence of the allele variants differs across racial/ethnic groups. It is this phenomenon that adds considerable importance to studies that comprise a diverse group of racial/ethnic groups or populations with different ancestry. For example, the study of genetic variability among various subgroups might help us better understand disease susceptibility and pharmacologic response to drugs. Furthermore, as has been noted in the literature (4), comparison of the various allele patterns in different populations might help us understand the evolutionary influences of that specific population that might have influenced diseases of importance today.

Although *CEBP* does not tend to publish descriptive papers of this nature, we did so to highlight the importance of the need to consider population differences in genetic variants. However, we strongly encourage the submission of manuscripts of studies that link genetic variants to function, phenotypes, and disease end points. In addition, linking these to disease risk and assessing effect modification by environmental, behavioral, and other factors will allow for a more comprehensive study of potential mechanisms involved in cancer etiology.

References

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