Identification of Cardiovascular Disease Associated Genetic Variants in a Population with African Ancestry

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Cardiovascular disease (CVD) is the leading cause of death in the United States, regardless of ethnicity and gender. Coronary heart disease (CHD), the most prevalent form of CVD, kills 370,000 people each year. A wide variety of CVD conditions including CHD are heritable, meaning DNA variation can influence disease risk. Identifying specific variants and genes that underlie the inheritance of CHD has the possibility to reveal insights into human biology and disease pathophysiology. Many known genetic variants associated with CVD only focus on a population of European ancestry. Those with African ancestry have disproportionately high susceptibility to CVD, and thus, it is crucial to identify specific CVD-related variants for this population.

In order to do so, differences in p-values between European and African CVD markers at known loci were analyzed. Regions with non-significant p-values in one dataset and statistically significant (p<1x10^-5) in the other were specifically targeted. Number of alleles and allele frequencies of these variants between populations of African ancestry and those of European ancestry were compared, and the functions of each variant were identified.

Such study of genetic variants will assist in more efficient methods and tools such as genome wide association studies for identifying CVD-related genes that are specific to those of African ancestry. This information will also help generate more advanced medical applications for diagnosing and treating the disease. Finally, it will further enhance genetic risk scoring for CVD in individuals of non-European ancestry.