“Impact of Rare Variants on Left Ventricular Traits in African Americans Using WGS”

Abstract:
Out of four deaths in the United States, one is caused by heart disease. Left Ventricular (LV) hypertrophy and diastolic dysfunction are symptoms of heart disease that could lead to heart failure. Statistics show heart failure is higher in African Americans than any other ethnicity in the United States.

Common variants associated with LV hypertrophy have been identified in several Genome Wide Association studies. However, rare variants using Whole Genome Sequencing (WGS) have not been thoroughly investigated yet, especially in African Americans. Therefore, we used WGS data from ~2000 African American individuals from the family-based Hypertension Genetic Epidemiology Network (HyperGEN) study to find rare variants and genes associated with LV hypertrophy and related echocardiography traits. Eighty-nine million variants went through rigorous quality control (QC) analysis; based on minimum allele frequency (MAF), depth of coverage, genotype quality, missing rate, principle component analysis, and other criteria. Fifty-five million variants passed QC and were used in the final analysis. Variants passing QC were annotated with ANNOVAR. We used the EMMAX software to implement single variant and gene-based tests of nonsynonymous variants at two MAF thresholds (<1% and <5%). We used the Bonferroni correction to adjust for multiple comparisons.

If these associations are confirmed and validated, our results could help identify why African Americans are at higher risk of heart failure and even help develop new treatments and therapies.

Refreshments will be provided

Friday, 1/26/2018 at 10:15am
Shelby Biomedical Research Building, Room 105
1825 University Boulevard
Birmingham, AL 35294

Questions? Please contact Informatics Institute, sominformatics@uabmc.edu