Principal Investigator: Christine Seidman

Institution Affiliation: Harvard Medical School

Ancillary Study Title: Analyses of the Allelic Spectrum of CVD Genes in the Jackson Heart Study Cohort

Project Overview

This study will seek common and rare sequence variants in genes that have been found to contribute to monogenic forms of cardiac hypertrophy, hypertension, dyslipidemia, diabetes, and cardiac arrhythmias. Relationships will be assessed between the allelic forms of these genes and the disorders of interest and their intermediate phenotypes. Exons and flanking regions of >250 selected candidate genes will be sequenced in DNA of approximately 2,500 consenting JHS participants. Because samples are to be phenotypically unselected, we will evaluate association of individual variants with quantitative traits across the cohort. Note: Because of the rapid and ongoing identification of new candidate genes, it is likely that subsequent requests will be made to extend the current project for resequencing of these additional candidate genes. Analyses will first detect from raw sequence traces the full complement of allelic variants in each candidate gene in the JHS cohort. The frequency and distribution of rare and common sequence variants in candidate genes will then be assessed in their relationship to specific cardiovascular risk factors. Finding risk alleles for cardiovascular disease may lead to the identification of new therapeutic targets or the development of tests that could contribute to individualized prevention and therapy.