Medical Xpress

Researchers provide genetic associations from a genome-wide scan for cardiovascular disease traits

September 19 2007

Researchers from Boston University School of Medicine (BUSM), Boston University School of Public Health (BUSPH), and the National Heart, Lung and Blood Institute (NHLBI), have completed analyses of a genome-wide scan on a group of two generations of participants from the landmark Framingham Heart Study (FHS). The analyses, which examine genetic differences that potentially affect the risk for cardiovascular disease and other disorders using data collected from FHS participants, are described in a series of articles published today in the on-line open access issue of *BMC Medical Genetics*.

Known as the Framingham 100K genome-wide scan, the results also are freely available through the database of Genotypes and Phenotypes, or dbGaP (view.ncbi.nlm.nih.gov/dbgap), developed by the National Library of Medicine's National Center for Biotechnology Information (NCBI). The database provides a number of electronic enhancements for viewing and examining the data, such as enabling users to drill down for precise details on all associations and allowing the data to be explored in the context of other NCBI genomic resources.

"We are excited by the possibilities of genome-wide association studies in uncovering genetic components involved in cardiovascular disease and its risk factors," said lead author of the overview paper L. Adrienne Cupples, PhD, a professor of biostatistics at BUSPH.

The Framingham Heart Study is a large, longitudinal study supported by the NHLBI of the National Institutes of Health and conducted in collaboration with Boston University. The study has been the source of key research findings regarding the contribution of high blood pressure, high cholesterol, cigarette smoking and other risk factors to the development of cardiovascular disease.

Cardiovascular diseases are major illnesses among Americans, affecting about one-third of the population and resulting in more than 870,000 deaths annually. Cardiovascular disease and its risk factors have substantial genetic contributors. FHS researchers and others have previously reported that certain traits related to heart disease, blood pressure, lipids, diabetes, and weight can be largely inherited and can be linked to or associated with specific genomic regions.

To evaluate how genetic variations contribute to these characteristics, or phenotypes, the researchers evaluated genotypes from the 100K Affymetrix GeneChip Human Mapping Set using DNA from 1,345 FHS participants in two generations. This genotyping technology tests subjects' DNA at 100,000 sites along the genome where people are known to commonly differ. Researchers then looked for association between the genotypes and 987 clinical "phenotype" measures, collected by the FHS over 59 years of follow up, including cardiovascular risk factors and biomarkers; cardiovascular disease; cancer; longevity and aging; and traits in the areas of lung function, sleep, neurology, and renal and endocrine function.

"It is our hope that the results from the genome-wide association study will lead to a deeper understanding of the role of common genetic variation in the development of cardiovascular disease and its risk factors," said Philip A. Wolf, MD, principal investigator of this study and a professor of neurology, research professor of medicine (epidemiology and preventive medicine) at BUSM. "Although these

results should be considered hypothesis generating and need to be replicated, these papers clearly provide proof of genome-wide associations," he added.

According to the researchers, because NCBI's dbGaP Web site permits investigators to review patterns of statistical significance within the unfiltered genetic association tests across the entire human genome, researchers around the world will be able to rapidly access and potentially replicate or refute findings from the FHS 100K study or develop new hypotheses for further research.

"The NCBI web site has the added advantage of linking into dozens of other databases, thereby providing access to other relevant human and animal data. The free exchange of data will accelerate the discovery of pathways to human disease," said Emelia J. Benjamin, MD, ScM, a professor of medicine at BUSM and one of the contributing authors.

"The Framingham Heart Study 100K project has investigated hundreds of phenotypes and as such it is among the largest genome-wide association studies of its kind, representing the first set of genome-wide association findings from the FHS," notes Christopher J. O'Donnell, MD, MPH, FHS, associate director and senior advisor to the NHLBI director for genome research. "These results set the stage for the launch of a much larger and more powerful NHLBI-funded genome-wide association study in the FHS known as SHARe (SNP Health Association Resource), which will be available in the fall of 2007."

"Together, these projects will provide a number of opportunities for scientists to take advantage of the wealth of data collected by FHS over several decades in an ethically responsible way," added Daniel Levy, MD, FHS director.

"The Framingham Heart Study dataset is another example of the

groundbreaking collaboration of researchers from Boston University Schools of Medicine and Public Health and from the NHLBI," said Karen Antman, MD, dean of BUSM and provost of Boston University Medical Campus. "Our researchers make extraordinary contributions that continuously improve the well-being of people all over the world."

Source: Boston University

Citation: Researchers provide genetic associations from a genome-wide scan for cardiovascular disease traits (2007, September 19) retrieved 4 May 2024 from https://medicalxpress.com/news/2007-09-genetic-associations-genome-wide-scan-cardiovascular.html

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.