Integrating Genomics into Public Health Research

Genomics promises to provide new insight into the interactions of genetic susceptibility with environment and lifestyle that maintain health or cause disease. Understanding the relationships among these factors at the population level may provide new opportunities for prevention and intervention.

Three major efforts are under way to translate genomics into usable information for improving population health:

- Integrating genomics into public health investigations (PHIs),
- Analyzing genetic variation among specimens in the National Health and Nutrition Examination Survey (NHANES) III DNA Bank,
- Expanding the Human Genome Epidemiology Network (HuGENet™).

Public Health Investigations

CDC is recognized around the world for conducting public health investigations in response to health emergencies and, by doing so, improving people's daily lives. Collecting and analyzing human genomic data in public health investigations has the potential to enhance our ability to understand variation in disease outcomes, characterize environmental exposures more accurately, and refine public health interventions such as vaccination, chemoprophylaxis, exposure reduction, and health promotion.

In 2006, CDC continued building the foundation for integrating human genomics into public health investigations in the following areas:

- Collecting and summarizing information about research protocols approved by CDC's Institutional Review Board as a basis for developing standard language and best practices for obtaining informed consent for DNA sample collection, storage, and testing.
- Working with individual investigators to develop protocols for specimen collection, transport, and banking.
- Developing the Influenza Public Health Genomics Initiative.

NHANES III DNA Bank

The National Health and Nutrition Examination Survey (NHANES) is a nationally representative survey of the U.S. population conducted by CDC's National Center for Health Statistics (NCHS). During the second phase of NHANES III (1991–1994), white blood cells were frozen and cell lines were immortalized to create a DNA bank.

A CDC-wide working group of epidemiologists and laboratory scientists developed a collaborative proposal for determining the prevalence of selected genotypes of public health importance using the NHANES III DNA Bank. The criteria used to select genes

for the proposal included the following:

- Known or hypothesized association with diseases of public health importance,
- Role in pathways affecting multiple diseases,
- Identified functional variants,
- Relatively common variants (prevalence >2.0%),
- Previously described gene–environment or gene–gene interactions,
- Relevant phenotypic data available in NHANES data sets, and
- No current use for clinical risk assessment or intervention.

The final approved proposal included 87 variants of 57 genes known to be important in at least six major pathways: nutrient metabolism; immune and inflammatory responses; activation and detoxification pathways; DNA repair pathways; hemostasis and renin/angiotension pathways; and developmental pathways. An additional proposal to analyze the genotype-phenotype data from NHANES was also approved.

Genotyping has been completed by the National Cancer Institute (NCI) Core Genotyping Facility. A committee composed of CDC's National Office of Public Health Genomics (NOPHG), NCHS, and NCI representatives provides oversight for the laboratory and analytic aspects of the study.

Data from the NHANES database will provide a basis for future analysis of gene–disease associations and gene–environment interactions. Establishing the prevalence of gene variants known to interact with specific environmental factors will provide a foundation for developing and assessing the potential impact of environmental interventions.

HuGENet™

The Human Genome Epidemiology Network (HuGENet™) is a global collaboration of individuals and organizations committed to assessing the role of human genome variation in population health and the potential of genomics to improve human health and prevent disease.

HuGENet™ seeks to advance our global genomics knowledge by determining the population prevalence of various human genetic variations, identifying associations between genetic variants and human diseases, measuring gene–environment interactions, and evaluating genetic tests for screening and prevention. Each week, NOPHG's Weekly Update publishes links to abstracts for HuGE articles newly entered into PubMed; these abstracts become part of HuGENet's curated database, which is freely available online. In collaboration with ten scientific journals, the Human Genome Epidemiology Network (HuGENet) also promotes collaborative systematic reviews and meta-analyses of genetic associations.

For more information, please visit CDC's National Office of Public Health Genomics website at www.cdc.gov/genomics.