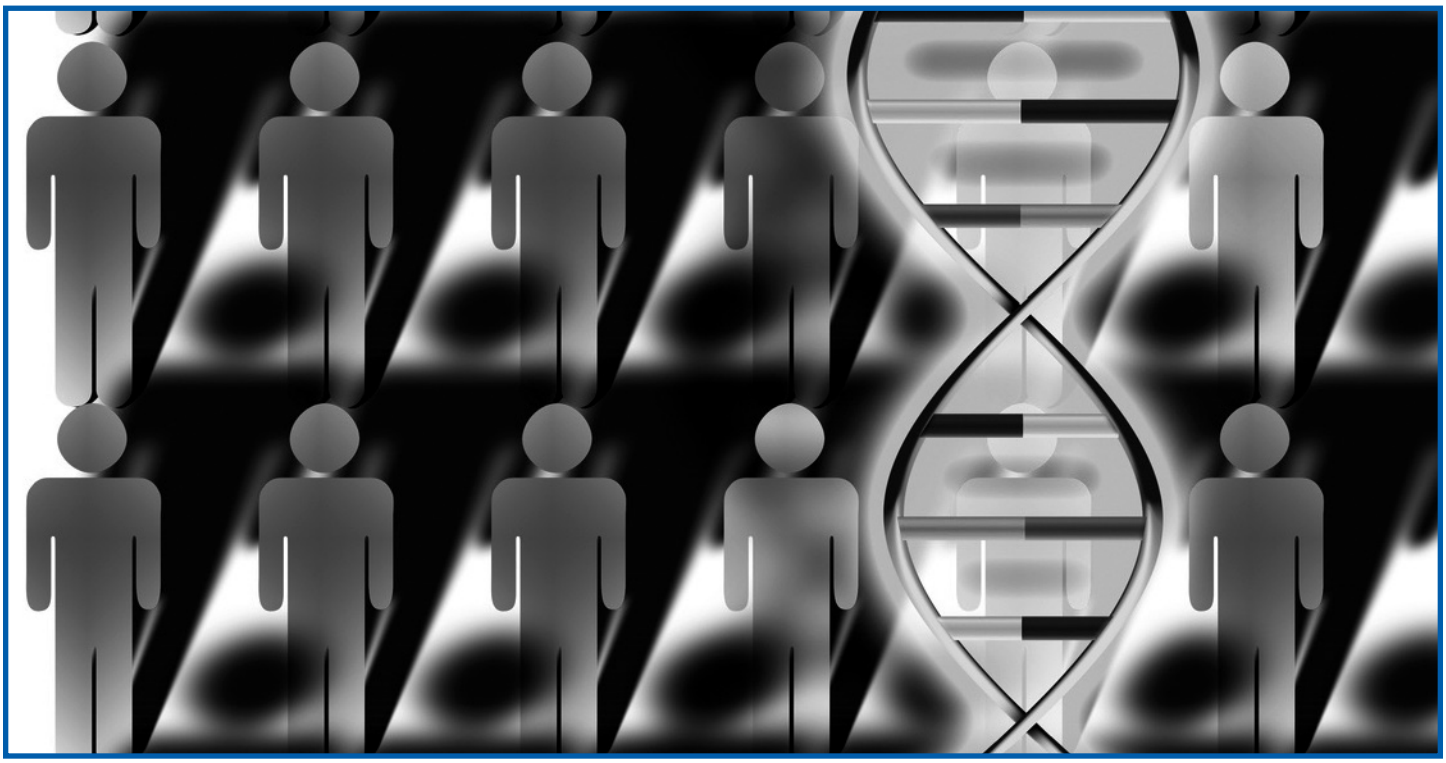
AT A GLANCE

National Office of Public Health Genomics

Translating Genomic Discoveries into Population Health Benefits 2008



"The new discipline of public health genomics represents the best approach for translating exciting genomic discoveries into appropriate actions for preventing disease and promoting health for individuals, families, and communities."

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U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
CENTERS FOR DISEASE CONTROL AND PREVENTION
COORDINATING CENTER FOR HEALTH PROMOTION

The Importance of Genomics

What is Genomics?

Genetics is the study of inheritance, or the way traits are passed down from one generation to another. Genes carry the instructions for making proteins, which direct the activities of cells and functions of the body and influence human traits (e.g., hair and eye color) and susceptibility to diseases. **Genomics** is the study of all the genes in a person, as well as the interactions of those genes with each other and a person's environment. All people are 99.9% identical in genetic makeup, but differences in the remaining 0.1% hold important clues about health and disease.

The study of genomics is likely to help doctors and other health care professionals understand why some people get sick from certain infections, environmental factors, and behaviors while others do not. This information could lead to new and better ways to improve health and prevent diseases for individuals and populations.

Human Genome Research

The human genome is a person's complete set of DNA. In 2003, researchers completed the 13-year Human Genome Project, which identified approximately 25,000 genes in human DNA. Much of this information is being transferred to the private sector to foster development of new medical applications. The project was coordinated by the U.S. Department of Energy and the National Institutes of Health (NIH), with support from international partners.

Now that the thousands of variations in human genes have been identified, researchers are studying these variations for associations with diseases of major public health importance, including chronic diseases such as heart disease, diabetes, stroke, and cancer, as well as infectious, environmental, and occupational diseases. Taking into account all factors that determine disease, it can be said that 100% of human disease results from the interactions between genetics and environment.

CDC's National Leadership

CDC is committed to ensuring that all people, especially those at greater risk for health disparities, will achieve their optimal lifespan with the best possible quality of health in every stage of life. With agency-wide health protection goals that support healthy people in healthy places across all life stages, CDC is setting the agenda to enable people to enjoy a healthy life by delaying death and the onset of illness and disability by accelerating improvements in public health.

Through the National Office of Public Health Genomics (NOPHG), CDC provides national and international leadership in public health genomics while building partnerships with other federal agencies, state health departments, public health organizations, professional groups, and the private sector. In fiscal year 2008, Congress allocated $12 million to support NOPHG's goals and initiatives.

Public health genomics is a multidisciplinary field focused on the effective and responsible translation of genome-based knowledge and technologies into health care practices to improve population health. It uses population data on genetic variation and gene-environment interactions to develop evidence-based tools for improving health and preventing disease. NOPHG's mission is to integrate genomics into public health research, policy, and programs. Doing so could improve interventions designed to prevent and control the country's leading chronic, infectious, environmental, and occupational diseases. Our efforts include conducting population-based genomic research, assessing the role of family history in determining risk and preventing disease, and evaluating genetic tests.

Genomics in Practice

Family History Public Health Initiative

People who have close relatives with common diseases such as heart disease, diabetes, and cancer are more likely to develop those diseases themselves. According to the 2004 Healthstyles survey, 96% of Americans believe that knowing their family history is important to their health. Yet only about 30% say they have ever tried to gather and organize this information. In 2002, NOPHG started the Family History Public Health Initiative to increase awareness of family history as a major risk factor for common chronic diseases. The initiative also promotes use of family history in programs designed to reduce the chronic disease burden in the United States.

In 2007, NOPHG analyzed data from the National Health and Nutrition Examination Survey (NHANES) to assess the risk for common chronic diseases and conditions in the U.S. population that can be attributed to family health history. NOPHG found that people with a moderate or high risk for diabetes because of family history were 2.3 and 5.5 times, respectively, more likely to have diabetes than people with average risk. This risk was independent of a person's sex, race/ethnicity, age, body mass index, blood pressure, income, or education.

NOPHG also found that 19.8% of U.S. women aged 20 years or older had a family history of osteoporosis. Women aged 35 years or older with a family history of osteoporosis were 2.3 times more likely to have the disease. The association grew stronger (to 8.4 times) when two or more close relatives had a family history of osteoporosis.

Evaluation of Genomics Applications in Practice and Prevention (EGAPP)

The EGAPP initiative was launched by NOPHG to establish and test a systematic, evidence-based process for evaluating genetic tests and other applications of genomic technology that are in transition from research to clinical and public health practice.

EGAPP activities are focused around the independent, nonfederal EGAPP Working Group established in May 2005. These activities include optimization of methods and processes for evidence review to deal with complex and rapidly emerging technologies; identification, prioritization, and selection of topics; participation on technical expert panels that guide conduct of evidence reports; and development of recommendation statements for clinicians based on the evidence.

In 2007, four CDC-funded evidence reports were completed for the EGAPP Working Group by Evidence-Based Practice Centers (EPCs), through an interagency agreement with the Agency for Healthcare Research and Quality (AHRQ). These reports are *Genomic Tests for Ovarian Cancer Detection and Management*, *Testing for Cytochrome P450 (CYP450) Polymorphisms in Adults With Non-Psychotic Depression Treated With Selective Serotonin Reuptake Inhibitors (SSRIs)*, *Hereditary Nonpolyposis Colorectal Cancer: Diagnostic Strategies and Their Implications*, and *Impact of Gene Expression Profiling Tests on Breast Cancer Outcomes*.

In December 2007, the first in a series of EGAPP Working Group recommendation statements, on CYP450 testing in patients with depression treated with SSRIs, was accepted for publication in *Genetics in Medicine.* Other EGAPP accomplishments for 2007 include the establishment of the EGAPP Stakeholders Group and creation of an independent EGAPP Web site (http://www.egappreviews.org), which hosts products of the EGAPP Working Group.

Population Research

NHANES Collaborative Genomics Project

In 2002, CDC and NIH's National Cancer Institute (NCI) began a collaboration to measure population variation in selected genes using stored DNA samples collected during the third NHANES. In 2007, CDC and NCI laboratories completed the genotyping of 90 variants in 50 genes and successfully deposited the results at CDC's National Center for Health Statistics (NCHS).

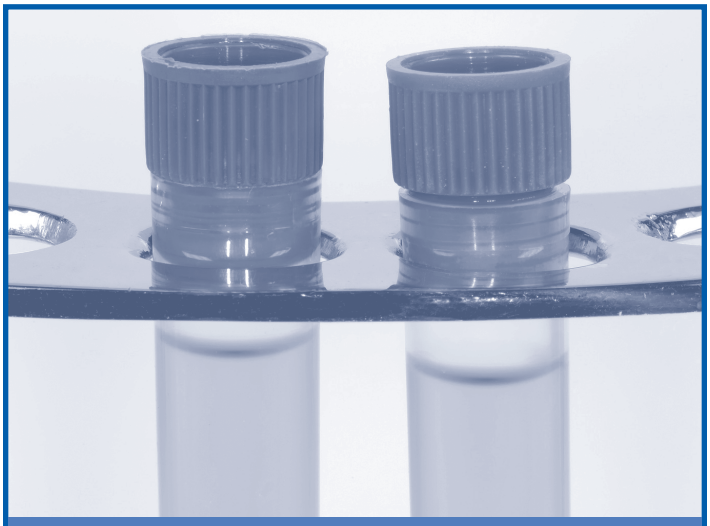
Researchers at NCHS are now conducting statistical analysis for about 35 genotype-phenotype correlation studies. The first publication of this study, "Prevalence in the United States of Variants in Genes of Public Health Importance: Third National Health and Nutrition Examination Survey (NHANES III), 1991–1994," is anticipated for 2008.

Public Health Investigations

In 2006, NOPHG provided seed funding for 11 innovative CDC projects to integrate genomics into their research and programs. Project topics included infectious disease, chronic disease, birth defects, pharmacogenomics, and environmental exposures. CDC extended funding for nine of these projects in 2007, and researchers expect to finish genotyping and data analysis in 2008.

Human Genome Epidemiology Network (HuGENet)

NOPHG established HuGENet in 1998 to help translate genetic research findings into practice for preventive medicine and public health. The network is intended to help advance the synthesis, interpretation, and dissemination of population data on human genetic variation in health and disease.



Direct-to-Consumer Nutrigenomic Tests

Direct-to-consumer genetic tests are increasingly available and may improve confidentiality, convenience, and accessibility. Amid ethical concerns and an uncertain regulatory landscape, the future of this mode of delivery is unclear. One class of products, nutrigenomic tests, is used to analyze DNA and lifestyle habits to assess health risks. In 2006, NOPHG used two national surveys, HealthStyles and DocStyles, to collect data on awareness and use of such tests among U.S. consumers and doctors. Results showed that 14% of consumers were aware of these tests, and 0.6% reported using them. Awareness was higher (44%) among doctors. Of these doctors, 41% had never had a patient ask about the tests, and 74% had never discussed the results of these tests with a patient. These data will help researchers understand and track public demand for these tests, as well as their potential for harm, and will provide a historical reference of trends in awareness and use over time.

HuGENet achievements in 2007 include the following:

- A HuGENet short course was held in the United Kingdom, and a workshop on the Assessment of Cumulative Evidence on Genetic Associations was held in Italy.
- HuGENet collaborators published 14 articles on genomic research topics in scientific journals.
- Eleven HuGE reviews were published by authors in the United States and five other countries, reflecting the network's global reach. Reviews are available for free at http://www.cdc.gov/genomics/hugenet/reviews.htm.
- NOPHG launched HuGE Navigator, an up-to-date, online knowledge base on human genome epidemiology. HuGE Navigator is primarily for researchers in public health genomics and other related disciplines. It is available at http://www.hugenavigator.net.

Building Public Health Genomics Capacity

State Genomics Programs

Since 2003, NOPHG has supported genomics programs in health departments in Michigan, Minnesota, Oregon, and Utah to integrate genomics knowledge (e.g., genetic risk factors) and tools (e.g., family history assessments) into chronic disease prevention programs and core public health functions. Program achievements in 2007 include the following:

- The Michigan Department of Community Health collaborated with Michigan State University to launch a pilot mortality review system to assess sudden cardiac death among young people aged 1–29 years in Michigan. This project is designed to raise awareness and increase prevention among people at high risk by identifying health care system changes and family-based interventions.
- The Minnesota Department of Health worked with University of Minnesota's School of Public Health to organize three new courses and a roundtable session on public health genomics as part of the annual Summer Public Health Institute. These activities build genomics capacity by educating the public health workforce on the importance and application of genomics principles in research, practice, and policy development.

Centers for Genomics and Public Health

Since 2005, NOPHG has funded Centers for Genomics and Public Health at the University of Michigan and the University of Washington. These centers provide expertise in translating genomic information into usable public health knowledge and technical assistance to state and community public health agencies. They also help states integrate genomics into programs and practice.

Center achievements in 2007 include the following:

- The Michigan center led development of the new Genomics Forum of the American Public Health Association to promote workforce competency in genomics; increase awareness and knowledge of genetic services; and participate in policy development, advocacy, and networking. More information is available at http://aphagenomicsforum.org.
- The Washington center launched a newsletter to educate and update public health practitioners, doctors, and the public on topics in genomics. The newsletter is distributed to libraries and medical clinics statewide.

Public Health Genomics Seminar Series

During 2007, NOPHG organized a seminar series called Public Health Genomics: Closing the Gap Between Human Genome Discoveries and Population Health. The series was presented in partnership with the NCI, the National Human Genome Research Institute, the National Institute for Child Health and Development, and the Office of Behavioral and Social Sciences Research.

The goal of the series was to educate health researchers and practitioners about public health genomics. Nine seminars were conducted throughout the year at NIH and broadcast live at CDC. Videocasts, presentations, and selected articles are available online at http://www.cdc.gov/genomics/events/special1.htm.

Future Directions

NOPHG's vision for public health genomics at CDC in the next 10 years is to accelerate the evaluation of new research findings and applications in genomics that can be used to improve health and prevent disease in the U.S. population. NOPHG will enhance collaborations with other CDC programs to integrate this knowledge appropriately into their goals and plans. We will strengthen our efforts to engage and educate health care providers and consumers about genomics, genetics, and family history to help them make decisions about health and health care. We also will expand national and international partnerships to further integrate genomics into policy, research, and programs.

For more information, please contact the Centers for Disease Control and Prevention
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