AT A GLANCE

National Office of Public Health Genomics

Seeking New Ways to Improve Public Health

2007

"Genomics is to the 21st century what infectious disease was to the 20th century . . . [it] should be considered in every facet of public health: infectious disease, chronic disease, occupational health, environmental health, [and] maternal and child health."

Gerard S, Hayes M, Rothstein MA. On the Edge of Tomorrow: Fitting Genomics into Public Health Policy. Journal of Law, Medicine, and Ethics 2002;30(3 Suppl):173–6.

Revised April 2007

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 that influence traits such as hair and eye color. **Genomics** is a newer term that describes 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% may hold important clues about the causes of disease. The study of genomics may 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.

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.

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 new 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.

CDC recognizes the potential of genomic research to improve the public's health. In 1997, the agency responded to this potential by creating the Office of Genomics and Disease Prevention (OGDP). In 2006, the office was renamed the **National Office of Public Health Genomics** (NOPHG). Since the office was created, CDC has provided public health leadership and built partnerships with other federal agencies, public health organizations, professional groups, and the private sector. In fiscal year 2007, Congress allocated $6.9 million for the NOPHG.

CDC defines public health genomics as the study and application of knowledge about the human genome and interactions between genes and their environment as they relate to health and disease in populations. Activities in the area of public health genomics include the following:

- Conducting surveillance for diseases with known or suspected genetic components.
- Performing epidemiological studies to understand the interactions among genes, behaviors, and the environment as they relate to common diseases.
- Using the information collected through surveillance and research to develop evidence-based programs and policies.
- Educating health professionals and the general public about how genomics can be used to prevent disease and improve public health.
- Evaluating and ensuring access to genetic tests and services that can help to prevent disease and improve health for all Americans.

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.

Family health history is a low-cost, low-tech genomic tool that can provide vital clues about people's shared environment and behaviors, as well as their risk for developing a particular disease or condition.

In 2002, CDC, in collaboration with NIH, academia, and health care organizations, began an initiative to develop and evaluate whether family history can be used to assess risk for common diseases and strengthen early disease detection and prevention strategies.

Major activities in 2006 included the following:

- Continued to evaluate the Family Healthware™ Web-based tool, developed in 2005, which collects information about health behaviors, screening tests, and a person's family history for stroke, coronary heart disease, diabetes, and colorectal, breast, and ovarian cancers. This tool is being evaluated for clinical use by the academic medical centers at the University of Michigan, Case Western Reserve University, and Evanston Northwestern Healthcare.
- Continued to collaborate with the U.S. Department of Health and Human Services on the Surgeon General's Family History Initiative, a campaign that marked Thanksgiving as National Family History Day. This project included creation of a Web-based tool called My Family Health Portrait (a simplified version of the Family Healthware™ tool) that organizes family health information into a printed version that people can take to their health care professional to help determine whether they are at higher risk for disease.
- Promoted the importance of knowing your family history to the public by developing a Web site that includes fact sheets, case studies, news articles, and other resources (http://www.cdc.gov/genomics/public/famhisMain.htm).

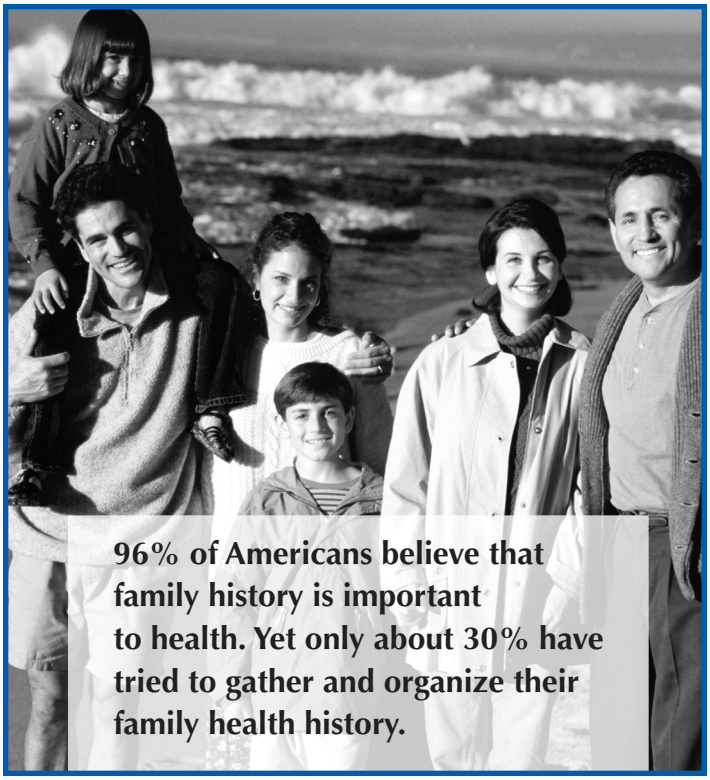
EGAPP Project

The success of the Human Genome Project has led to rapid translation of genomic information into clinical applications. Although most of the more than 1,000 genetic tests currently available for clinical testing are used to diagnose rare, single-gene disorders, a growing number have population-based applications with potential public health impact.

In 2004, CDC launched a model project called Evaluation of Genomic Applications in Practice and Prevention (EGAPP). EGAPP is working to implement and evaluate a coordinated, systematic, and evidence-based process for assessing genetic tests and other applications of genomic technology that are in transition from research to clinical and public health practice.

An independent, nonfederal working group was created to select topics for review and to develop systematic review methods. The 13-member, multidisciplinary EGAPP Working Group has written protocols, developed criteria for topic selection, and commissioned five evidence reviews (two more reviews are planned for 2007).

The working group considers the evidence and then prepares conclusions and recommendations for publication. Other important EGAPP activities include engaging stakeholders to help develop and disseminate targeted messages based on the evidence and recommendations, surveying stakeholders to assess EGAPP's value and impact, and exploring ways to develop a sustainable EGAPP-like process.



Population Research

Integrating Genomics into Public Health Investigations

Public health investigations are essential to CDC's mission to improve public health. By collecting human genomic data, investigators could help identify additional risk factors for disease susceptibility, severity, and transmission.

To create a foundation for genomic surveillance, CDC is

- Assessing and developing public health infrastructure and capacity to study genomics.
- Building a science base for genomics and its relationship to human exposure to diseases and environmental factors.
- Developing standard language for informed consent for DNA sample collection, storage, and testing in public health research.
- Developing standard guidelines and tools for specimen collection, processing, transport, and storage.

Genes of Public Health Importance

In 2004, CDC and NIH's National Cancer Institute began a collaboration to measure population variation in selected genes using stored DNA samples collected during the third National Health and Nutrition Examination Survey (NHANES III). The goal of the project is to develop genotype prevalence estimates for the top 100 genetic variants of public health significance for a nationally representative sample of the U.S. population.

Human Genome Epidemiology Network

CDC established the Human Genome Epidemiology Network (HuGENet™) in 1998 to help translate genetic research findings into opportunities for preventive medicine and public health. HuGENet™ is an international collaboration of individuals and organizations committed to the integration and dissemination of population-based data on the role of human genetic variation in health and disease. Recent partners include coordinating centers in the United Kingdom, Canada, and Greece. In 2006, HuGENet™ published the first edition of an online handbook for systematic reviews (http://www.cdc.gov/genomics/hugenet/reviews/guidelines.htm), which are peer-reviewed and published in partnership with 10 scientific journals. The network's free online resources include a weekly summary of new scientific articles on human genome epidemiology, a searchable database (HuGE Pub Lit), case studies for training, and information on workshops and publications.

Integrating Genomics into State Programs

Centers for Genomics and Public Health

CDC funds Centers for Genomics and Public Health at Schools of Public Health at the University of Michigan and the University of Washington. These centers serve as regional hubs of expertise in genomics and public health with a focus on translating genomic information into practical public health knowledge, providing technical assistance to state and local public health agencies, and integrating genomics into programs and practice.

For example, the centers collaborated with CDC to provide two Web-based training programs for public health professionals. The first is a 45-minute introductory presentation called Genomics for Public Health Practitioners. It describes the application of genomics to public health, dispels myths, and identifies challenges in public health genomics. A second, more in-depth program is called Six Weeks to Genomics Awareness and includes six presentations designed to help public health professionals understand how advances in genomics are relevant to public health. In 2005, CDC funded the centers in Washington and Michigan to continue their work and to develop new collaborations on key projects, such as the Family History Public Health Initiative and the EGAPP Project. These collaborations continued in 2006.

State Capacity Grants

In July 2003, CDC established cooperative agreements with state health departments in Michigan, Minnesota, Oregon, and Utah. Since 2004, CDC has helped these states develop and expand their capacity to integrate genomics tools and knowledge into state public health programs and external organizations. State activities focus on building infrastructure and partnerships, training the public health workforce, educating the public, using surveillance surveys such as the Behavioral Risk Factor Surveillance System to assess genomics integration, and promoting genomics screening tools. Highlights of these programs can be found in *Genomics and Population Health 2005* (http://www.cdc.gov/genomics/activities/ogdp/2005/chap10.htm).

State Programs in Action

With support from CDC, states are working to educate their public health professionals about how genomics can be used in the fight against chronic diseases. For example, **Utah** developed Genomics 101 presentations for public health professionals to increase their knowledge and interest in the topic. **Michigan** presented a workshop series called Cancer Genomics for Public Health for public health professionals working in the area of cancer control. **Minnesota** included genomics as a topic for regional chronic disease workshops held across the state, and it held the state's first Genomics and Public Health Conference in 2004.

In addition, states are looking for ways to incorporate genomics into existing chronic disease programs and activities. For example, **Michigan, Minnesota, Oregon,** and **Utah** have integrated genomics into their state comprehensive cancer control plans. **Michigan** and **Minnesota** are working with local WISEWOMAN (Well-Integrated Screening and Evaluation for Women Across the Nation) projects to evaluate and modify existing family history questions to identify women at increased risk for cardiovascular disease.

Future Directions

CDC and its partners will continue to translate advances in genomics into public health practice and to integrate genomics into research, policy, and programs. CDC also is working to create an infrastructure that will build on the achievements of the Human Genome Project to prevent disease and improve health in the 21st century.

For more information, please contact the Centers for Disease Control and Prevention
National Center for Chronic Disease Prevention and Health Promotion
4770 Buford Highway NE, Mail Stop K–89, Atlanta, GA 30341-3717
Telephone: 770-488-8510 • Fax: 770-488-8336
E-mail: genetics@cdc.gov • Web: http://www.cdc.gov/genomics

CS110431