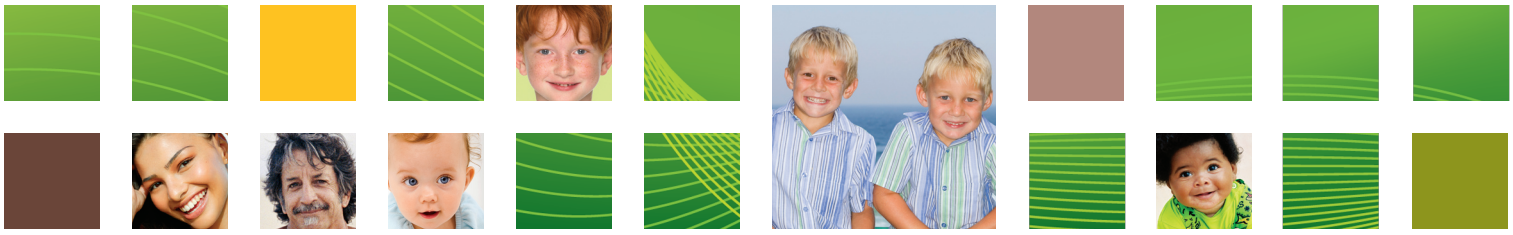


GENOMICS

SUCCESSES AND OPPORTUNITIES FOR POPULATION-BASED RESEARCH AND PRACTICE

AT A GLANCE

2009

NATIONAL CENTER FOR CHRONIC DISEASE PREVENTION AND HEALTH PROMOTION

IMPROVING HEALTH AND QUALITY OF LIFE FOR ALL PEOPLE

DEPARTMENT OF HEALTH & HUMAN SERVICES · USA

CDC

CENTERS FOR DISEASE CONTROL AND PREVENTION

What is Genomics?

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.

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.

> *"Translating the knowledge we are gaining from gene discoveries into practical clinical and public health applications will be critical for realizing the potential of personalized health care and improving the health of the nation."*
>
> **Muin J. Khoury, MD, PhD**
> Director, Office of Public Health Genomics, CDC

CDC's Response

CDC works to integrate human genome knowledge into public health research, policy, and programs. Through the Office of Public Health Genomics (OPHG), CDC strives to improve interventions designed to prevent disease and improve the public's health by

- Conducting population-based genomics research.
- Assessing the role of family health history.
- Supporting the evaluation of genetic tests.
- Translating genomic knowledge.

Conducting Population-Based Genomics Research

Population-based genomics research examines the role that genetic differences play on disease risk and helps to identify associations between genes and factors such as diet and environmental exposures. These studies may help us understand which populations are more susceptible to disease, as well as help us improve their health and treatment outcomes. Examples of OPHG research include

- In 2008, OPHG reported genetic prevalence estimates by age, sex, and race/ethnicity among non-Hispanic whites, non-Hispanic blacks, and Mexican Americans in a nationally representative sample of the U.S. population collected from participants in the Third National Health and Nutrition Examination Survey (NHANES III). These estimates provide the foundation for a comprehensive data bank of human genetic variation, which can be used as a reference for future population-based genomic studies.
- OPHG provides funding to other CDC programs to integrate genomics into their public health studies. New projects funded in 2008 will examine the role of genetics and genomics in autism, maternal and child health, hearing loss, and chronic beryllium disease (a disease primarily affecting the lungs, caused by exposure to beryllium, a metal found in coal, oil, certain rock minerals, and soil).

OPHG tools for researchers include

- **HuGE Navigator**, an up-to-date knowledge base in human genome epidemiology, with information on population prevalence of genetic variants, gene-disease associations, gene-gene and gene-environment interactions, and evaluation of genetic tests. The HuGE Navigator contains more than 30,000 scientific articles on human genome epidemiology (http://www.hugenavigator.net).
- **HuGE Reviews**, systematic reviews of research findings on particular gene-disease associations, which are published in partnership with 10 scientific journals. These reviews typically point to gaps in existing epidemiologic and clinical


CDC's Response (continued)

knowledge, thus stimulating further research in these areas. In 2008, four HuGE reviews were published. Thirteen HuGE reviews are anticipated in 2009 on genetic risk factors and pregnancy and childbirth, mental disorders and epilepsy, lung cancer, and general cancer risk.

- **HuGENet™**, a global collaboration of investigators around the world. Genomics researchers can become involved by undertaking systematic reviews and participating in meetings and workshops.

Assessing the Role of Family Health History

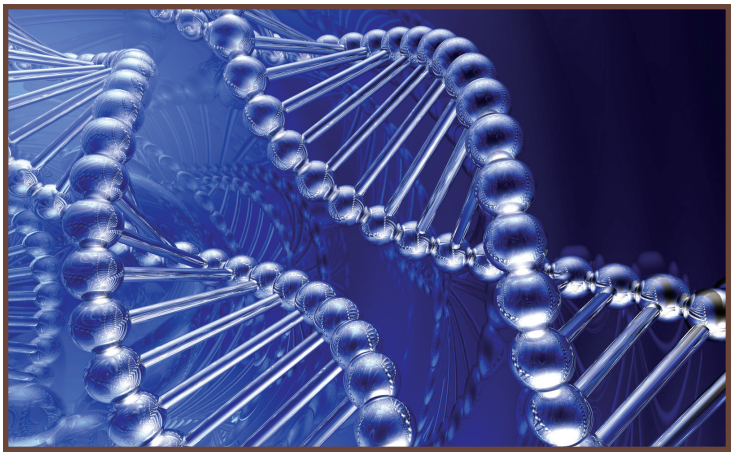
Family history reflects an individual's genes and the behaviors and environmental factors they share with their family members. Family history is a risk factor for many chronic diseases, such as cancer, coronary heart disease, and diabetes, making it an important tool for identifying people at increased risk for these diseases. OPHG research and projects include

- A study using the Family Healthware™ tool, in which researchers examined whether personalized prevention messages tailored to familial risk motivates people to change their behaviors and lifestyle and seek additional medical advice or screening. The methods and results of this study will be published in early 2009.

- A national working group on family history, which is part of the U.S. Department of Health and Human Services' (HHS) Personalized Health Care initiative. OPHG is helping to develop a plan to enable consumers to electronically and securely share their family health history information with family members and health care providers, and ultimately, to allow this information to become part of the consumer's electronic health record.

Supporting the Evaluation of Genetic Tests

Genetic tests for more than 1,600 diseases have been developed, and clinical testing for more than 1,300 diseases is currently available. Most are used for diagnosis of rare genetic disorders, but a growing number of tests may help with screening for and prevention of chronic diseases. Based on reports of the HHS Secretary's Advisory Committee on Genetics Health and Society, reliable, objective information is needed on genetic tests to help health care providers, consumers, and policy makers make decisions on their appropriate use. These reports support



OPHG's systematic and evidence-based process for evaluating genetic tests. OPHG-supported activities include

- In 2008, the independent Evaluation of Genomics Applications in Practice and Prevention (EGAPP) Working Group published their methods for collecting, analyzing, and grading evidence used to evaluate the analytic and clinical validity and clinical utility of genetic and genomic tests.

- In January 2009, the independent EGAPP Working Group released the following three new evidence-based recommendations based on EGAPP-commissioned, CDC-funded evidence reports:

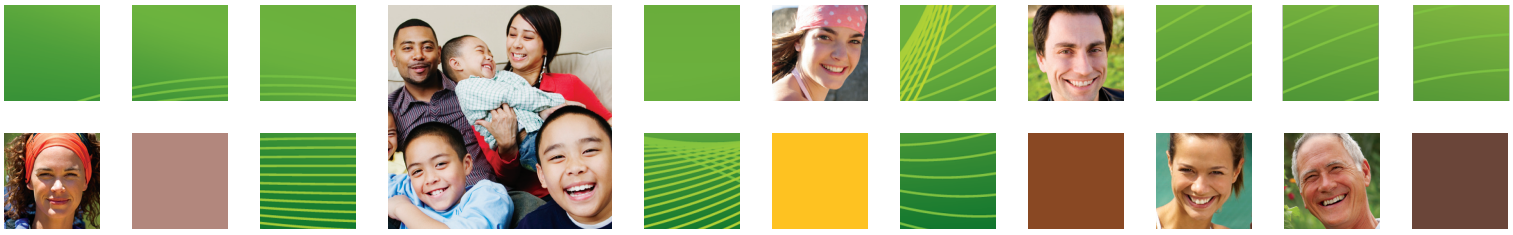
  - *DNA Testing Strategies Aimed at Reducing Morbidity and Mortality from Lynch Syndrome.*

  - *Can UGT1A1 Genotyping Reduce Morbidity and Mortality in Patients with Metastatic Colorectal Cancer Treated with Irinotecan?*

  - *Can Tumor Gene Expression Profiling Improve Outcomes in Patients with Breast Cancer?*

Translating Genomic Knowledge

Translation of genomic knowledge into health practice involves a multidisciplinary approach. Understanding the validity, utility, and use of genomic information and applications will help guide the development of health interventions that maximize health benefits and minimize harm to individuals and populations. OPHG-supported activities include



CDC's Response (continued)

- State health departments in Michigan, Minnesota, Oregon, and Utah completed their 5-year genomics cooperative agreements in July 2008. The states integrated genomics goals and activities into cancer control plans and helped establish family history assessments as part of disease prevention efforts for cancer, cardiovascular disease, and diabetes. Through population-based surveys, the states gained knowledge about consumer awareness and use of family history in disease prevention and of direct-to-consumer (DTC) genetic tests. For example, the results of a 2006 Behavioral Risk Factor Surveillance System (BRFSS) survey in Michigan, Oregon, and Utah published in 2008 showed that 24.4% of Oregon consumers, 19.7% of Utah consumers, and 7.6% of Michigan consumers were aware of DTC nutrigenomic tests. Older individuals up to 65 years and those with more education and higher income were more likely to be aware of these tests.
- In 2008, OPHG funded five new projects that translate promising genomic applications, such as genetic tests and family history, into medical and public health practice. These projects will assess the impact of these applications on population health. These projects include
  - Family history education to improve genetic risk assessment for cancer.
  - Promotion of best practices for cancer genomics through surveillance, education, and policy.
  - Pharmacogenomics education to bridge the gap between science and practice.
  - Translation of genomics applications into health practice in Oregon.
  - Development and evaluation of a risk-benefit framework for new genetic tests to educate clinicians, policy makers, and other key decision makers about the potential benefits and harms of genetic testing.
- In 2009, the OPHG-sponsored EGAPP Stakeholders Group will help to translate and disseminate the new EGAPP recommendations for use in clinical and public health practice. This group also is helping to increase awareness of the EGAPP initiative and its methods and processes.
- Genomics centers at the Universities of Michigan and Washington continue to provide expertise and technical assistance for national and state efforts to help translate genomic information and applications, including the EGAPP recommendations, into public heath research, policy, and programs.
- In 2008, OPHG used two national surveys to assess U.S. consumer awareness and use of direct-to-consumer personal genome scans (HealthStyles) and knowledge of and experiences with these scans among U.S. physicians (DocStyles). Preliminary analyses of these data indicate that one in five consumers surveyed were aware of these tests, but very few had actually used them. OPHG also worked with three state health departments to assess U.S. consumer awareness and use of these scans using the 2009 BRFSS.

Additional projects on cancer genomics have been funded by the National Cancer Institute (NCI) in 2009.

Future Directions

In 2009, OPHG will continue to strengthen its genomics translation initiative by establishing a network of government agencies, academic and research institutions, health care plans, consumer advocacy groups, biotechnology industries, and other organizations around common goals for using genomics in preventing disease, improving treatments, and reducing health disparities. OPHG is also more effectively using public-private partnerships in evaluating genetic tests. Additionally, OPHG will support more extensive genotyping, which will expand the human genetic variation data bank and make these data more accessible to the scientific research community.

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