GENOMICS

SUCCESSES AND OPPORTUNITIES FOR POPULATION-BASED RESEARCH AND PRACTICE

AT A GLANCE

2010

National Office of Public Health Genomics
Centers for Disease Control and Prevention

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 physical and social 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 public health and clinical practice 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."***
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> **Muin J. Khoury, MD, PhD**
> Director, National Office of Public Health Genomics, CDC

CDC's Response

CDC works to integrate human genomics into public health research, policy, and programs. CDC's National Office of Public Health Genomics (NOPHG) strives to improve interventions designed to prevent disease and improve the public's health by helping to develop and distribute evidence-based knowledge and tools in

- Human genome epidemiology.
- Family health history.
- Genetic and genomic testing.

Human Genome Epidemiology

Human genome epidemiology (HuGE) examines the role of genetics in disease and health and helps to identify links between genes and other factors, such as diet and environmental exposures, in populations. Research in these areas can help us understand which populations might be more likely to develop disease as well as design interventions to improve treatment and health outcomes.

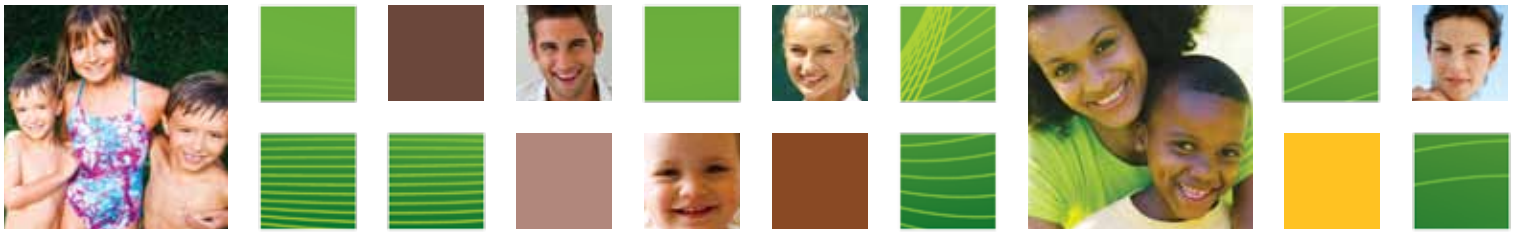
Population-Based Studies

Since 2008, NOPHG and the National Center for Influenza and Respiratory Diseases have funded the Pediatric Acute Lung Injury and Sepsis Investigators (PALISI) Network to conduct a genetic epidemiology study of life-threatening and fatal flu in children and young adults. As of August 2009, PALISI has enrolled more than 130 people who had severe or fatal flu, with 40% presumed to have H1N1 flu. Preliminary findings from the study will be published this year.

In 2009, NOPHG continued to fund five CDC genomics-related projects that focused on genetic aspects of folic acid supplementation before pregnancy, environmental hazards that affect autism spectrum disorder, infant blood lead concentrations and auditory brainstem response, worker susceptibility to noise-induced hearing loss, and chronic beryllium disease. NOPHG also continued to work with other groups at CDC to examine the links between selected genetic variants and disease outcomes in data from the third National Health and Nutrition Examination Survey (NHANES III). Studies published in 2009 focused on associations between selected genetic variants, blood lead levels, and blood pressure and between selected gene variants and lead exposure and cognitive function.

New Tools and Resources for Researchers

Two new tools were developed in the online database HuGE Navigator (http://www.hugenavigator.net) to help researchers search and select data from all published studies that examine genetic variation across the human genome. These tools also provide prevalence estimates for selected genetic variants in the U.S. population. In addition to these new tools, the second edition of *Human Genome Epidemiology: Building the Evidence for Using Genetic Information to Improve Health and Prevent Disease* was published in January 2010.



CDC's Response (continued)

The Human Genome Epidemiology Network (HuGENet™) developed new guidelines that were simultaneously published in eight journals to help researchers standardize results and interpret evidence found in human genetic association studies.

Family Health History

Family history reflects individuals' 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. NOPHG projects in this area include the following:

- In 2009, results of NOPHG's Family Healthware™ evaluation study showed that people perceive their risk for cancer to be significantly higher than for other diseases when they base their risk on family history. Men worried most about getting heart disease, while women worried most about breast cancer, followed by heart disease. Diabetes was perceived to be the least severe condition. Heart disease was perceived to be the most controllable condition compared with cancer, which was perceived to be the least controllable. Compared with men, women had higher levels of perceptions of risk and worry for several diseases.

- As part of a national work group on family health history led by the U.S. Department of Health and Human Services, NOPHG is helping to enhance My Family Health Portrait, an Internet-based tool for collecting family health histories, so that it also assesses disease risk and provides advice based on risk and family history. Other projects are underway with Microsoft HealthVault and My Health*e*Vet, an electronic health system for the Veteran's Administration.

- In 2009 and early 2010, four CDC studies on family history and health were published or accepted for publication. The topics of the studies included family history of asthma, early-onset stroke/transient ischemic attack, and diabetes. One study examined family history of diabetes and the likelihood of developing gestational diabetes, the prevalence of metabolic syndrome, and the ability to detect undiagnosed diabetes in adults.

Genetic and Genomic Testing

Researchers have developed genetic tests for more than 1,800 diseases. Most tests look at single genes and are used to diagnose rare genetic disorders, such as fragile X syndrome and Duchenne muscular dystrophy. However, a growing number of tests look at multiple genes that may increase or decrease a person's risk for common diseases, such as cancer or diabetes. In addition, some genetic tests look at rare genetic variants, such as those responsible for some hereditary breast and ovarian cancers. Such tests have the potential to help prevent common diseases and improve the health of individuals and populations.

Evaluation of Genetic Tests

In 2009, the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group established by NOPHG released three new evidence-based recommendations that address genetic testing for various clinical scenarios involving breast and colorectal cancer. The EGAPP Working Group recommended genetic testing for Lynch syndrome (a type of inherited colorectal cancer) for patients with newly diagnosed colorectal cancer to help relatives of these patients determine their disease risk. However, not enough evidence was available for the EGAPP Working Group to recommend either for or against the use of gene expression profiling for tumors to improve health outcomes in specific populations of women with breast cancer or for the routine use of *UGT1A1* genotyping to determine the use of irinotecan in chemotherapy for patients with metastatic colorectal cancer.

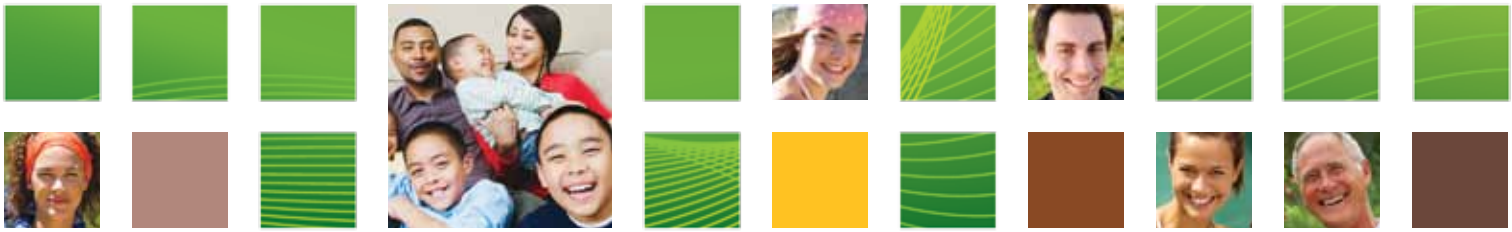
In 2010, the EGAPP Working Group will release recommendations on genetic testing in adults with a history of venous thromboembolism and on the use of genomic profiling to assess risk for cardiovascular disease.

Translation of Genomic Applications into Practice

In 2009, NOPHG, the National Institutes of Health, and other stakeholders established the Genomic Applications in Practice and Prevention Network (GAPPNet™) to effectively and responsibly translate evidence-based genetic and genomic tests into practice. GAPPNet™ is made up of more than 100 stakeholders. In 2009, genomics translations projects funded by NOPHG achieved the following:

- Developed a Web-based tool for clinicians and policy makers to assess the potential benefits and harms of new genetic tests.

- Developed a pharmacogenomics program to educate health professionals and students about the validity, usefulness, and



CDC's Response (continued)

potential benefits and harms of pharmacogenomic tests.

- Planned a program to educate health care providers about the benefits of documenting patients' family histories, identifying family history factors considered high risk, referring patients to genetic consultation, and using genetic testing.
- Promoted and helped to increase health insurance coverage for genetic testing of *BRCA1/2* mutations associated with hereditary breast and ovarian cancer in 3 of the 24 health plans in Michigan.
- Set up a genomics surveillance program in Oregon that uses data from the Behavioral Risk Factor Surveillance System, Medicaid claims, the Oregon State Cancer Registry, and genetics counseling and testing services to monitor awareness and use of genomic tests for cancer and documentation of family history by health care providers and the public.

In 2008, NOPHG used two surveys to learn more about U.S. health care providers' and consumers' awareness and use of personal genome tests. These tests are developed on the basis of limited scientific information and may not provide valid or useful results for people who are tested. Of the 5,399 consumers surveyed, 22% were aware of the tests and 0.3% had used them. Of the 1,880 health care providers surveyed, 42% were aware of the tests, with dermatologists and pediatricians most aware (50%) and obstetrician/gynecologists least aware (36%).

Among health care providers who were aware of the tests, 42% had at least one patient who had asked questions in the past year about having such a test, and 15% had at least one patient who had brought the results of a personal genome test to them for discussion. Among this 15% (mainly internists and family doctors), 75% said the test results changed some aspect of the patient's care, such as screening tests offered, medications or dosages prescribed, lifestyle changes recommended, frequency of follow-up appointments planned, or diagnoses made.

In 2009, the Healthy People 2020 Federal Interagency Workgroup approved two 10-year national objectives to (1) increase the proportion of people with newly diagnosed colorectal cancer who receive genetic testing to identify Lynch syndrome or other familial colorectal cancer syndromes and (2) increase the proportion of women with a family history of breast or ovarian cancer who receive genetic counseling.

Future Directions

In 2010, NOPHG will enhance national efforts to translate evidence-based genomics applications—such as genetic tests, technologies, and family history—into better, more targeted interventions for preventing disease, improving treatments, and reducing health disparities. To achieve these objectives, NOPHG is

- Developing tools for researchers and health care providers that provide evidence summaries on the validity and usefulness of specific genomic applications and the potential harms and benefits of these applications.
- Starting a new initiative called Genetics for Early Disease Detection and Intervention to educate health care providers and public health practitioners on the use of clinical, genetic, and family history information for early diagnosis of rare genetic disorders and common diseases with the goal of improving health outcomes.
- Enhancing its efforts to integrate genomics into national and state surveillance systems and surveys and using NHANES to generate and combine data on the distribution of genetic risk factors in the U.S. population and its subgroups.
- Increasing efforts to integrate genomics into public health investigations. For little additional cost, public health practitioners can collect genetic information to learn more about why some people develop disease.

For more information, please contact the Centers for Disease Control and Prevention
1600 Clifton Road NE, Mail Stop E-61, Atlanta, GA 30333
Telephone: 404-498-0001 • Fax: 404-498-0140
E-mail: genetics@cdc.gov • Web: http://www.cdc.gov/genomics