Activities at CDC

OGDP Accomplishments 2001

Informed consent for population-based public health research involving genetics is necessary to assure that study subjects understand the risks and benefits of participation.

Informed Consent Document Developed for Participants in Population-Based Studies: Research involving genetics must be conducted ethically and include the informed consent of study participants. However, little guidance, has been available for population-based studies of common gene variants with low to moderate individual disease risks. Recommendations developed for family-based research are not well suited for most population-based studies because they generally do not distinguish clinically relevant information from epidemiologic data, which are meaningful only at the population level and involve few physical, psychological, or social risks for individual participants.

In response to this dilemma, CDC worked with a multidisciplinary group of experts and developed content and process guidelines for informed consent for population-based genetic research studies. The guidelines address several important issues. The consent document must include information that outlines why the study is being done, what is involved in the study, how information collected about research subjects will be kept confidential, and what the risks and benefits might be for study participants. The consent document must clearly state what is to be done with any remaining biological samples when the study is completed. In addition, some genetic studies may have the potential to result in a product with commercial value. When this possibility exists, it should be disclosed along with a statement about whether participants would share in any profits.

An article about these guidelines (http://www.cdc.gov/genetics/info/reports/policy/consentarticle.htm) for informed consent for population-based genetics research was published in the November 14, 2001, issue of JAMA. It is hoped that this new informed consent document and supplemental brochure will help people make informed decisions about participating in genetic studies and, in turn, help bridge the gap between gene discovery and the ability to use genetic information to improve health and prevent disease.

Readers will find additional information on Informed Consent: Public Health Perspective at http://www.cdc.gov/genetics/info/perspectives/infmcnst.htm

New Centers for Genomics and Public Health Established: In 2001, CDC awarded funding to three schools of public health, establishing the first Centers for Genomics and Public Health. The University of Michigan, the University of North Carolina, and the University of Washington will each receive approximately $300,000 per year for three years. Through a cooperative agreement with the Association of Schools of Public Health and CDC, each center will develop a regional hub of expertise to use information about gene-environment interactions associated with disease to develop new strategies for improving health.

CDC is collaborating with three schools of public health to develop the knowledge base around chronic disease specific areas and provide genomics training to the public health workforce and provide technical assistance in genomics to state and local public health agencies.

The centers will build on and complement existing programs at the universities (in public health, medicine, genetics, and other disciplines) and will establish relations with local and state health departments. Centers may also draw on other regional resources, such as professional organizations, the clinical community, and industry to develop activities in three areas: contributing to the knowledge base on genomics and public health; providing technical assistance to local, state, and regional public health organizations; and developing and providing training for the current and future public health work force. Although some of these activities now exist at schools of public health, it is believed that establishing the centers will generate a high level of synergy, collaboration, and networking among schools of public health and other public health institutions. With this collaborative approach, CDC hopes to draw attention to gaps in the translation of gene discoveries into disease prevention and to demonstrate-through real examples-a way to begin addressing the gaps.

Readers will find additional information on (CDC) Awards Funds for Genetics Programs at http://www.cdc.gov/genetics/activities/fund2001.htm

Genomics Toolkit Project Initiated: Building capacity in genomics in state public health agencies requires tools that are useful in achieving the aims of health promotion and disease prevention. Success also depends upon a sufficient level of commitment from public health agencies to use these tools in public health programs. To help make these tools readily available and to encourage the commitment to use them, CDC is developing a Genomics Toolkit for state and local public health agencies. The toolkit will be the product of a working group coordinated and convened by the Association of State and Territorial Health Officials (ASTHO) that includes representatives from CDC and other public health organizations with interests in laboratory science, chronic disease, public policy, genetics, maternal and child health, local public health, and epidemiology. The project is also being guided by the results of ongoing needs assessments of state and local public health agencies. The Genomics Toolkit is intended to be an evolving document that will be updated as new resources are identified and as genetic science evolves.

Public Health Impact of Genetic Tests Assessed at the End of the 20th Century: As more genetic tests are considered for population screening, and as associations between genes, environment, and common diseases are discovered, the number of people who might

of the disease or condition being tested. Of the 423 clinical tests, 51 had potentially greater impact on public health because of their use in either statewide newborn screening programs or other population screening programs or of their use in testing for common diseases with a prevalence of more than 1 in 2,000 people. Among the 328 tests performed for research purposes, only 18 met the criteria for potentially greater public health impact.

CDC concluded that at the end of the 20th century, fewer than 10% of genetic tests were relevant to public health. At the present time, the majority of genetic test are used in diagnosis or genetic counseling for rare, single-gene disorders in a limited number of people. However, in reviewing the diseases for which genetic tests are being developed in research settings, CDC foresees that tests for more common conditions are likely to become more prevalent.

Readers will find additional information on the Public Health Impact of Genetic Tests at the End of the 20th Century at
http://www.cdc.gov/genetics/info/reports/files/print/gim3-6-405.pdf

ACCE provides a model system to evaluate data on the analytic validity, clinic validity and clinical utility of emerging genetic tests.

Standard Approach Being Developed to Evaluate Genetic Test Data: In spite of the continued proliferation of genetic tests, only a few have been evaluated for clinical validity and utility. In a cooperative agreement with the Foundation for Blood Research, (http://www.cdc.gov/genetics/activities/fbr.htm) CDC is establishing a standard approach for evaluating data on genetic tests. ACCE, named for the four components of evaluation-analytic validity; clinical validity; clinical utility; and ethical, legal, and social implications - is a model process for evaluating data on emerging genetic tests.

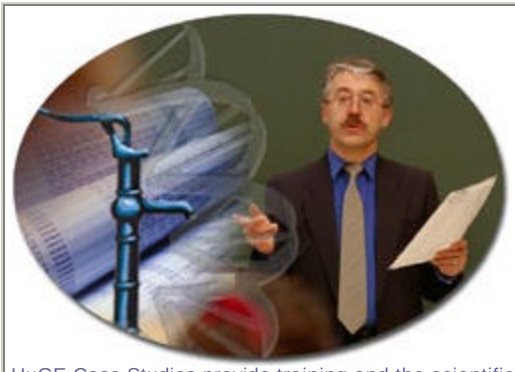
The process includes collecting, evaluating, interpreting, and reporting data about DNA and related testing for disorders with a genetic component, then putting the data in a format that allows policy makers to have access to up-to-date and reliable information for decision making. An important part of this process is the identification of gaps in knowledge about the validity of genetic tests. Conditions to be examined in the first year of this effort will include cystic fibrosis, hereditary hemochromatosis, and factor V Leiden. Cancer- related genetic tests will be assessed in the second year. The long-term goal of this project is to develop a process that can be used by others to critically review genetic tests.

Readers will find additional information on ACCE: A CDC-Sponsored Project Carried Out by the Foundation for Blood Research at
http://www.cdc.gov/genetics/info/perspectives/files/testACCE.htm

Human Genome Epidemiology Network Expanded The Human Genome Epidemiology Network (HuGENet™), (http://www.cdc.gov/genetics/hugenet/default.htm) a global collaboration of individuals and organizations who develop and share epidemiologic information about the human genome, has expanded its Web site with the inclusion of HuGE Case Studies, the E-Journal Club, and the Published Literature Database.

The purpose of HuGE Case Studies is to train health professionals in the practical application of human genome epidemiology and to help readers acquire conceptual and practical tools for critically evaluating the growing scientific literature in specific disease areas. The knowledge gained from Case Studies will inform readers of diverse backgrounds on how epidemiologic data can provide a scientific basis for using genetic information to improve health and prevent disease.

Readers will find additional information on HuGE Case Studies at
http://www.cdc.gov/genetics/hugenet/casestudies.htm



HuGE Case Studies provide training and the scientific evaluation of using genetic information to improve health and prevent disease.

The HuGE e-Journal Club summarizes the findings and discusses the public health implications of published genetic epidemiologic research in an interactive forum.

The HuGE e-Journal Club is an electronic discussion forum where new human genome epidemiologic findings, published in the scientific literature and included in CDC's Office of Genetics and Disease Prevention's Weekly Update, are abstracted, summarized, presented, and discussed via a newly created HuGENet™ listserv. The E-Journal Club presents epidemiologic information in a standardized format and promotes discussion among HuGENet™ members and other interested public health and clinical professionals on selected articles from the current published literature.

Readers will find a menu on HuGE e-Journal Club at
http://www.cdc.gov/genetics/hugenet/ejournal.htm

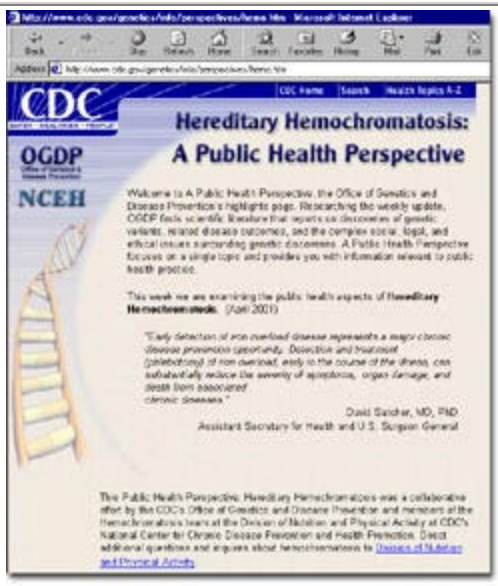
The HuGE Published Literature Database provides users access to a database of indexed epidemiologic literature on gene variant frequencies, gene-disease associations, gene-gene and gene-environment interactions, and genetic test assessments. Database users are able to obtain a list of relevant articles by querying the database using selected search terms. In addition, key information from each article is provided, along with a direct link to PubMed's abstract of the research article.

Readers may search on HuGE Literature Database Online Search at
http://www2.cdc.gov/nceh/genetics/hugenet/frmSearchMenu.asp

Researchers can query the HuGE Published Literature Database to find published reports on genetic research relevant to public health.

These additional features offer a new dimension to the HuGE Net Web site and provide opportunities for discussion among professionals from clinical, public health, and other diverse backgrounds about human genome epidemiology and about education for decision-making involving the use of genetic tests and services.

Public Health Perspective Series Presents Complex Information to a Diversified Web Audience: Recent advances in human genetics emanating from the Human Genome Project present interesting challenges and opportunities for both health communicators and educators. Information about genes, diseases, and environmental risk factors is complex and must be communicated in a way that is meaningful for professionals in public health and related health professions. CDC has developed a communication tool designed to present scientific information in a clear and factual manner to a diversified audience.



Public Health Perspectives provide clear, factual information to a diversified audience on specific topics related to the use of genetics in improving health and preventing disease.

Each Public Health Perspective is essentially a web site that contains information and commentary on a single topic. Complex topics such as hereditary hemochromatosis, informed consent, pharmacogenomics, and genetic testing are examined from a public health perspective. Readers will find both scientific information as well as popular press news stories, video, and commentary geared to public health professionals. This “something for everyone” approach allows readers to examine and understand discoveries of genetic variants; related disease outcomes; and complex social, legal, and ethical issues surrounding genetic discoveries. Perspectives pieces are published by OGDP in collaboration with other CDC offices and centers and are featured in the Weekly Update every four to six weeks. The entire series of Public Health Perspectives can be found on the OGDP Web site at http://www.cdc.gov/genetics/info/perspective.htm .