Foreword

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Public Health Professional
A person educated in public health or a related discipline who is employed to improve health through a population focus.

(Who Will Keep the Public Healthy? Educating Public Health Professionals in the 21st Century, IOM, 2003, p. 30)

Welcome to the first CDC report on genomics and population health! In this periodic report, we hope to present a timely and practical collection of vignettes of the status of genomics and population health in the United States. This information is intended for **public health professionals** who are interested in integrating genomics into health promotion, disease prevention and health care. The report includes information on what we know, what we don't know and what is currently relevant in this rapidly evolving field. At the outset, we realize that many terms and concepts may be new and we therefore include a brief introduction to the "lingo" developed by the University of North Carolina at Chapel Hill. For your convenience, additional links to relevant Web sites and other resources are also available throughout the report.

With the completion of the Human Genome Project in 2003, the stage has been set for an accelerated pace of discovery of thousands of genetic variants. Many variants will be studied for association with diseases of major public health importance, including adult chronic diseases, childhood conditions, infectious, environmental and occupational diseases. Applications of genetic information in diagnosis and prevention of various diseases must be driven by evidence on gene functions in normal and disease states as well as by the value of such information to improve health outcomes. In spite of the potential promise and excitement about human gene discoveries, there are still immense gaps in the knowledge needed for a successful translation of new research results into population health benefits. This "translation gap" calls for an important public health leadership role in applied research, policy development and integration of genomics into the practice of 21st century medicine.

In this first report, we present some examples to show how public health is beginning to address three major gaps along the genomics "translation highway":

1. Conducting genomics and population health research,
2. Developing evidence on the value of genomic information, and
3. Integrating genomic information in practice and programs.

1. Conducting genomics and population health research.

Most human disease results from interaction between inherited genetic variations and numerous environmental factors (e.g., diet, infections, lifestyle, chemicals and social factors). With the thousands of genetic variants discovered, there is a real urgency to characterize what genetic variation means for health, to assess the prevalence of genetic variants in different populations, and to examine their contribution to the population burden of disease, death and disability. In this first report, we highlight two current CDC efforts in this area. The first (Chapter 1) is a project to evaluate the prevalence of 57 genes and their variants in a nationally representative sample of the United States population. We provide an overview of the criteria used to select these genes for study and a description of the planned project. The second example (Chapter 2) discusses the potential for examining the role of human genomics in the setting of acute public health investigations, a mainstay of public health efforts to characterize and prevent disease occurrence in communities with acute health problems or a disproportionate burden of disease.

2. Developing evidence on the value of genomic information.

To use genetic information successfully in population-level programs and individual management of patients, we need solid scientific evidence to help guide policy development and guideline recommendations. In this report, we review the public health implications of the evolving asthma genomics research, including pharmacogenomics, the new targeting of drug therapies to specific genotypes (Chapter 3), as well as the evolving evidence and guidelines about genetic testing for breast and ovarian cancer (Chapter 4). We also discuss the example of MCADD to describe an emerging area of newborn screening using the new technology of tandem mass spectrometry, which is increasingly adopted in state public health programs (Chapter 5). We also highlight a CDC initiative to develop and evaluate family history tools for augmenting chronic disease prevention efforts (Chapter 6). We ask critical questions about the role of "genetic profiling" tests that are being promoted for preventing coronary heart disease and determine whether or not such tests are ready for prime time (Chapter 7), and review ethical, legal and social issues (Chapter 8).

3. Integrating genomic information in practice and programs.

Because health professionals are most concerned with practice and programs, this section of our report is the longest. To integrate genomic information into practice and programs, we need a competent workforce, a robust health system, and an informed public. We need careful policy development and planning that recognizes the complexity of genomics issues while building on approaches that have been successful in evaluating other health related technologies. We provide an update on efforts to ensure the quality of genetic testing (Chapter 10). We also provide timely and relevant practice information for two specific conditions: hereditary

hemochromatosis (Chapter 11) and cystic fibrosis (Chapter 9). We cover training issues (Chapter 12), genomic tools for public health (Chapter 13), issues related to state genetic planning (Chapter 14), and provide Internet-based resources (Chapter 15).

We hope that the topics chosen for this report reflect emerging common interests and concerns. We have tried to present population-based data when available, describe potential applications to public health and prevention practice, and offer value-added interpretation. The report is based on a collaboration of many individuals and programs at CDC and other partners. We are indeed very thankful for their efforts.

We would like to invite readers to give us feedback on this first report, to help us improve future editions. Please use the comments card found in this report or visit our Web site: (http://www.cdc.gov/genomics/activities/ogdp/2003.htm). Current information on the application of genomics in public health is still sparse and contains many gaps; however, we hope that increasing experience at the state and community levels will help to fill these gaps over time. We hope that the data and information contained in these reports will prove useful in guiding public health research, policy and practice in order to help reap the benefits of the Human Genome Project for citizens in the 21st century.