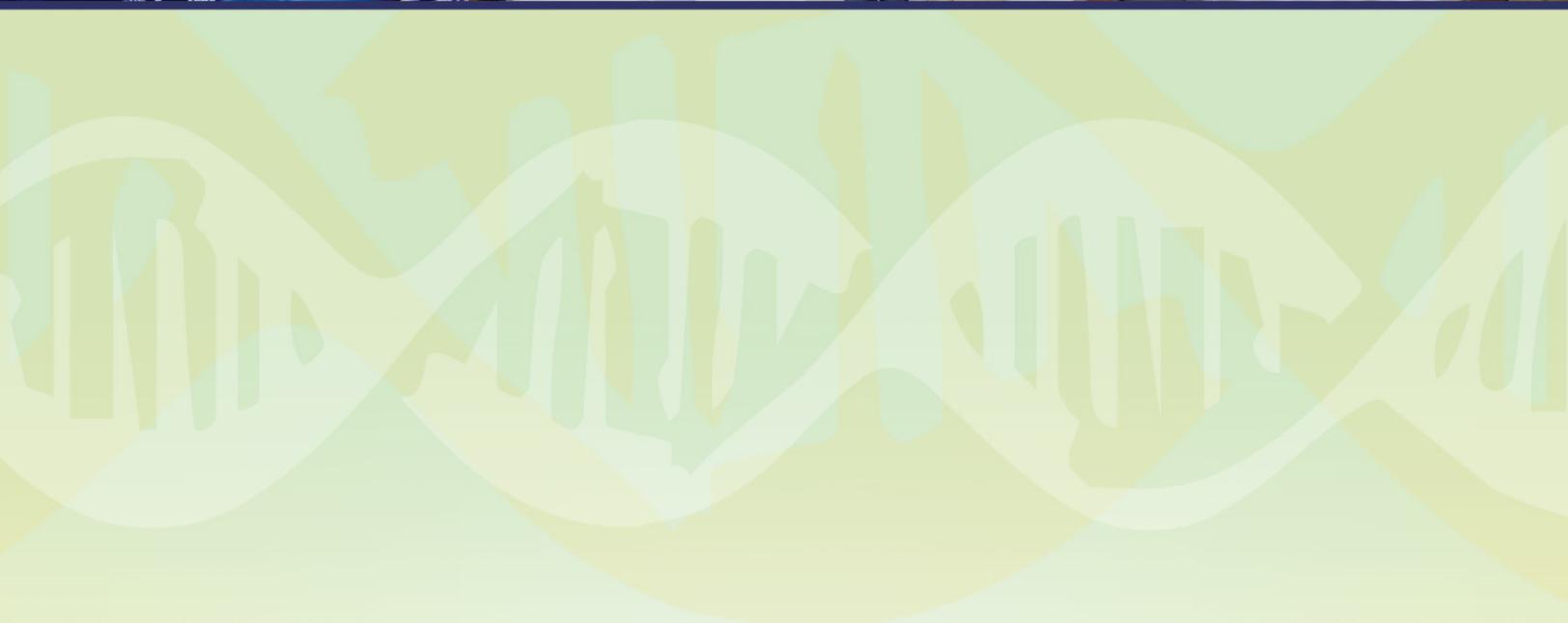


2012–2017

# PRIORITIES FOR PUBLIC HEALTH GENOMICS

STAKEHOLDER CONSULTATION | PRIORITIES CONFERENCE REPORT | SEPTEMBER 2011





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The contents are solely the responsibility of the authors and do not necessarily represent the official views of CDC.

## PREFACE

### Priorities for Public Health Genomics 2012–2017

This report results from an initiative of the Office of Public Health Genomics, Centers for Disease Control and Prevention (CDC/OPHG) to recommend priorities to advance the field of public health genomics during the five year period 2012–2017.

CDC/OPHG initiated three activities to develop these priority recommendations: (1) an extensive consultation of public health genomics stakeholders conducted by the Center for Public Health and Community Genomics at the University of Michigan School of Public Health, including a) analysis of a Request for Information (RFI) issued by the Department of Health and Human Services/ CDC, b) interviews of key informants from the public health system and c) informal discussions with community- and practice-based public health practitioners; (2) interviews of key informants from the non-profit and for-profit sectors conducted by Genetic Alliance; and (3) an all-day meeting held in Bethesda, MD on September 14, 2011, attended by over 70 leaders in the field of public health genomics, based in academic institutions, public health and health care organizations, and community-based organizations.

Part One of this report consists of the Stakeholder Consultation prepared by the Center for Public Health and Community Genomics, focusing on the public health community. The table of recommendations in Part One and the appended report of key informant interviews incorporate inputs gathered by Genetic Alliance. Appendices provide further detail regarding recommendations received as well as a literature review prepared by the Center for Public Health and Community Genomics.

Part Two consists of the report of the September 14 meeting prepared by Genetic Alliance.

Additional materials related to the Stakeholder Consultation can be found in the appendices to this report, and on the website of the APHA Genomics Forum, [www.genomicsforum.org](http://www.genomicsforum.org). The full list of Request for Information (RFI) responses can be found on the following government website: <http://www.regulations.gov/#!docketDetail;dt=FR+PR+N+O+SR+PS;rpp=10;po=0;D=CDC-2011-0008>.

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# **PART ONE**

## **STAKEHOLDER CONSULTATION**





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## II. EXECUTIVE SUMMARY

Several major themes emerged from the review, summarization and categorization of the responses to the Request for Information (hereafter, RFI), interviews of key informants, informal discussions, and relevant literature. Not surprisingly, most of these themes have also been highlighted in earlier reports, workshops, round tables and journal articles addressing the future of public health genomics (see *Appendix 2, Literature Review*), and were also reflected in the recommendations that emerged from the September 14, 2011 conference on “Developing Priorities for Public Health Genomics 2012–2017” (see *Part Two of this report*). While the consistency of many recommendations evidences continued areas of concern, the most recent recommendations demonstrate continued development of the field and its promise for strengthening all areas of public health. This executive summary identifies these major themes, and chapter IV, “Process,” suggests frameworks that can be useful in translating the opinions and recommendations resulting from our Stakeholder Consultation into a comprehensive plan for moving public health genomics forward in the next five years.

### Significant Gains to Date

Our Planning Committee and many respondents stressed the accomplishments that have already been achieved by public health genomics in terms of diseases prevented, human suffering reduced, and lives saved. Past public health successes include effective initiatives (e.g., the establishment and expansion of newborn screening programs) largely focused on heritable disorders, and while the impact has been great, the portion of the population that has benefited has been relatively small. More recently, BRCA gene testing

for breast and ovarian cancer risk, and Lynch syndrome testing for colon cancer risk demonstrate the potential of genomics to reduce mortality and strengthen prevention strategies related to common chronic diseases. We need to make the public—along with health care providers and organizations, third party payers, and others working to improve health within and beyond the sphere of public health genomics—aware of these effective and important accomplishments and opportunities for health impact. Moreover, as new tests for rare and common chronic conditions proliferate, and multiplex screening, microchips, and whole genome sequencing become available at ever-lower cost, the public health system needs a source of credible evidence on the validity and utility of these tests as well as their cost effectiveness. Addressing these issues will require research, a trusted source to evaluate the evidence and report conclusions of the research, and utilization of this evidence by public health and medical practitioners.

### Promising Potential in the Near Future

Genomics research has contributed to the development of tools that hold significant promise for achieving public health goals in the near future. The utilization of family health history and screening for genetic profiles hold great promise for indicating above-average risk for common chronic diseases. Integrating genomic, environmental and behavioral factors into methods of assessing chronic disease risk can have a great impact on population health. The growth of the field will require emphasis on later stages of translation of research into cost-effective, evidence-based applications in public health and medicine, and education of the health professional workforce on genomics

and—as it becomes more understood—the efficacy of genomic tools. Moreover, by using culturally and linguistically appropriate methods, a focus on these applications within communities experiencing an increased burden of disease can help achieve health equity, a major component of the public health mission.

### Integrating Genomic, Social and Environmental Factors

A number of respondents spoke about the potential to reduce disease by integrating genomics with research on the social and environmental factors responsible for disease and health disparities. Epigenetic research was identified as an important means of elucidating the pathways through which social and environmental conditions trigger genetically-based responses responsible for chronic disease and health disparities. Promoting this research will have enormous impact on achieving public health goals by focusing our efforts on the external factors most responsible for disease conditions and disparities. This is a longer-term strategy that will involve (1) developing transdisciplinary research teams linking biology with social science; (2) developing metrics for measuring environmental and social exposures; (3) building repositories combining genetic, social and environmental data, and (4) translating this research into effective prevention strategies, policies and programs.

### Infrastructure, Collaboration and Partnership; Education

Many recommendations spoke to the current fragmentation of efforts to integrate genomics into public health practice. This fragmentation was identified within academe and practice, and at

## DEFINING PUBLIC HEALTH GENOMICS

**“PUBLIC HEALTH GENOMICS IS A MULTIDISCIPLINARY FIELD CONCERNED WITH THE EFFECTIVE AND RESPONSIBLE APPLICATIONS OF GENOME-BASED KNOWLEDGE AND TECHNOLOGIES TO IMPROVE POPULATION HEALTH...IT FOCUSES ON PREVENTION, EVIDENCE-BASED MULTIDISCIPLINARY SCIENCE AND ETHICAL, LEGAL AND SOCIAL IMPLICATIONS, INCLUDING ADDRESSING HEALTH DISPARITIES.”**

—MUIN KHOURY, DIRECTOR OF THE OFFICE OF PUBLIC HEALTH GENOMICS, CDC

federal, state and local levels. Communication and knowledge sharing between those within the public health workforce whose jobs include genomics and those whose work does not is also fragmented. Recommendations called for better collaboration between these various groups, and for shared resources such as data banks and registries. Several recommendations called for significant community engagement through community-based research projects and educational programs, in order to strengthen research, improve the relevance and effectiveness of interventions, and address health disparities. Other recommendations urged the necessity of educating the public health and clinical workforces and the public at large, in order to achieve the level of genomic literacy that will promote the appropriate integration of genomics throughout public health.



### Moving Forward

It became apparent in reviewing the data that much of the problem encountered in securing buy-in for moving public health genomics forward results from a lack of shared understanding and engagement by those who work in all components of the public health system. While many

promising public health genomics interventions are not yet ready for implementation, there is an imbalance between research focusing on pharmacogenomics with potential utilization in medical care and that of furthering evidence-based utilization of genetic tests and family health history in public health programs. The promise of genomics in achieving public health goals needs to be clearly understood throughout public health academe, practice and community, so that all who practice in these areas recognize the relevance of the field to their own work and their own goals. Furthering this shared understanding and broad engagement, and building the collaborations and coalitions that can result, must be a central goal for those currently engaged in public health genomics.

# The Role of Public Health Leadership

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The Planning Committee reflected on the many recommendations calling for collaborations, partnerships, and the breakdown of silos to achieve the promise of public health genomics. Who will exercise the leadership necessary to achieve these results? The CDC, through its Office of Public Health Genomics (OPHG) has been carrying the torch for these efforts for the past 14 years. That leadership included developing model genomics programs and centers, adding significantly to the body of literature on public health genomics, convening conferences to share knowledge, and initiating major programs to develop and provide information on evidence-based genomic applications and to accelerate and streamline the process of research translation.

The Planning Committee believes CDC has the potential to exercise even stronger leadership in the years ahead. It can forge a close, effective, continuing relationship with the diverse stakeholders described in this chapter, utilizing this relationship to inform its own work and to achieve the collective support of the stakeholders in furthering the goals of public health genomics. The three ex officio members of the Planning Committee have spoken of the potential of building this relationship: Toby Citrin, Director of the Center for Public Health and Community Genomics (CPHCG), views the next few years as an opportunity for CDC to align itself with the many and diverse stakeholders making up the field of public health genomics, eliciting their

joint efforts to move the field forward. Dean Hosgood, Chair of the Genomics Forum of the American Public Health Association, has pointed out the role that the Forum and its more than 900 members can play as a critical component of this stakeholders group. Ella Greene-Moton, Community Education Coordinator for CPHCG, speaks of the role that can be played by the National Community Committee network of community-based organizations and community representatives partnering with the nation's 37 Prevention Research Centers (PRC), bringing the voices of diverse communities to support the field.

#### MORE ON THE WEB:

Genomics Forum

<http://genomicsforum.org/>

Prevention Research Centers

<http://www.cdc.gov/prc/>

PRC National Community Committee

[http://www.cdc.gov/prc/newsroom/national\\_community\\_committee.htm](http://www.cdc.gov/prc/newsroom/national_community_committee.htm)

### III. ENVISIONING THE FUTURE OF PUBLIC HEALTH GENOMICS

Even before “public health genomics” was recognized as an emerging field of study and practice, varied stakeholders found common ground at the intersection of genetic science and population health. Today, discussions of public health genomics still bring together a wide range of voices. This chapter is included to capture the vision of the Planning Committee that contributed to this report, which included a wide range of perspectives. The committee consisted of 13 people with extensive experience applying genomics in academe, practice and the community, who advised and provided guidance to the Center for Public Health and Community Genomics in carrying out the stakeholder consultation and preparing this report. Written contributions from Planning Committee members are included as sidebars interspersed throughout the “What We Learned” chapter.

Stakeholders in public health genomics at large include researchers, health professionals, and academics from varied disciplines seeking to understand and treat disease. They include patients, survivors of illness, and members of families at risk for genetic disorders, seeking hope. They include scientists, community leaders and members, and public health advocates seeking to understand the interaction of genetic and environmental factors in health and in health disparities.

To represent these stakeholders in implementing this Stakeholder Consultation on the future of public health genomics, we called to our “roundtable” knights of the academy, of the community, and of health practice. Not unexpectedly, we found in this group no single holy grail. Instead we heard of multiple, sometimes overlapping quests, approaches and strategies that—broadly put—would apply our



growing knowledge base in the field of public health genomics toward improving health outcomes.

Among the 13 Planning Committee members involved in the execution of this consultation were stakeholders from the various spheres that intersect with genomics: health professionals, including nurses and genetic counselors, public health administrators, researchers, community advocates, and academics in disciplines from health to the social sciences. Some came to the discussion sharing past experiences—surviving breast cancer, or being mindful of shared genes with a twin—underscoring the reality that genomics is both public and private, universal and personal, studied and lived.

The Planning Committee noted that, on the one hand, the diversity of stakeholders in the field of public health genomics has led to atomization, or “silo” configurations among academic disciplines and the workforce. But there is also hope that stakeholder diversity will be an asset in the future; a collaborative, multidisciplinary, systems approach to public

health genomics will speed and promote research, translation, and improvements in health outcomes.

The field of public health genomics faces multiple challenges, today, in addition to atomization: under-informed stakeholders, under-developed guidelines, lack of evidence-based applications for young and emerging technologies, and lack of resources to address these challenges adequately.

Still, arising from the kaleidoscopic perspectives of our Planning Committee is a consistent and largely hopeful vision of the future of public health genomics. It is one in which research, education, and communications will create informed health professionals and communities. It is a future in which professional and institutional collaborations paired with the technologies of the information age will bring together valuable data and help identify priorities. And it is one in which stakeholders will remain mindful of the role that environments play in genomics, and engage and keep sight of the people and communities who are served by public health.

## IV. PROCESS

### Methods

The table of recommendations in this report is based on the following sources of input:

- 62 responses to an RFI solicited by the CDC/OPHG, open to the general public from June 30, 2011 through August 1, 2011. The RFI contained the following five questions:
  1. What are the most important activities that should be carried out by the public health system in 2012–2017 to apply genomic knowledge to public health goals?
  2. What outcomes specific to public health might be achieved as a result of carrying out these activities?
  3. What policies are needed in order to achieve these outcomes?
  4. What institutions, organizations and agencies need to participate in achieving these outcomes and what role should they play?
  5. What barriers are anticipated in achieving these outcomes and how might they best be overcome?
- Nine interviews of key informants, selected by the Planning Committee to represent public health practice, academe and the community, were conducted by the Center for Public Health and Community Genomics
- Eight interviews of key informants in the for-profit and non-profit sectors were conducted by Genetic Alliance
- The above inputs were supplemented by three informal discussion groups that were organized to gain additional insight and perspectives from diverse

communities, public health practitioners, and stakeholders at Genetic Alliance's Annual Meeting

### Limitations

The Center for Public Health and Community Genomics and Genetic Alliance were limited to a total of nine key informant interviews each for the Stakeholder Consultation process.

A complete list of RFI respondents, key informants, discussion group participants and Planning Committee members can be found in Appendix One.

Summaries of key informant interviews and informal discussion groups appear in the appendices, and summaries of RFI responses will be posted on the website of the APHA Genomics Forum, [www.genomicsforum.org](http://www.genomicsforum.org).

An initial step in the Stakeholder Consultation process was the selection of a Planning Committee to serve as a multidisciplinary

team providing advice and guidance to the Center for Public Health and Community Genomics. Invitations were sent to leaders in public health genomics from multiple disciplines. Four leaders in academe, four leaders in public health practice, two community leaders and three ex-officio members convened as the project's Planning Committee to assist and consult throughout the Stakeholder Consultation. Five teleconferences were held throughout the months of July, August and September and an in-person meeting was held in Chicago on August 24.

An initial list of major topic areas and sub-themes identified from existing literature on public health and genomics was later refined following a review of RFI responses, key informant interviews, and discussion groups. Themes that were present in the literature but not found in the data were discarded, while themes that appeared within the data and not in the literature were added to the final list. The resulting major themes were used as



categories for organizing the recommendations and other data resulting from the consultation.

After the data were reviewed and organized into themes, sub-themes, and summaries, the Planning Committee convened for a one-day, in-person meeting to discuss the report inputs and the framework for the report. The Planning Committee came to a consensus that data should be presented by major themes in the final report, and additionally organized into a table of recommendations based on the *Public Health in America* framework that includes action steps and key players, along with excerpts from the qualitative data gathered. The presentation by major themes is contained in the “What We Learned” chapter of this report. The table of recommendations is described under the “Frameworks” heading on this page and is located in Chapter VI of this report.

Following are the specific methods used in reviewing each type of input:

#### Request for Information

Each of the 62 responses was analyzed for key themes and sub-themes, and summarized. Frequencies of major themes were tallied by two separate individuals and compared to sift out consistent topic areas. Themes that were tallied 15 or more times were identified as major themes. The table of recommendations utilizing the *Public Health in America* framework includes representative quotations, recommended action steps and key actors identified from RFI responses and used in

the analysis of all inputs and the development of the recommendations.

#### Key Informant Interviews

A total of nine key informants were interviewed by the Center for Public Health and Community Genomics—four from academia, four from public health practice, and one from the community at large. Each informant was asked about his/her background, views on the current state of public health genomics, vision for the future, and priorities to advance the field. Interviews were conducted via telephone and each lasted between 30–60 minutes. All interviews were audio recorded following permission from the interviewee. Upon completion of the nine key informant interviews, summaries were developed and key themes identified. Themes that appeared in at least five of the interviews were identified as major themes for the final report. Supporting quotations, action steps and key actors were pulled from the interviews, entered into the table of recommendations, and used in the analysis throughout this report. An additional eight key informants from the non-profit and for-profit sectors were interviewed by Genetic Alliance, and recommendations were incorporated in the table of recommendations appearing as Chapter VI of this report.

#### Discussion Groups

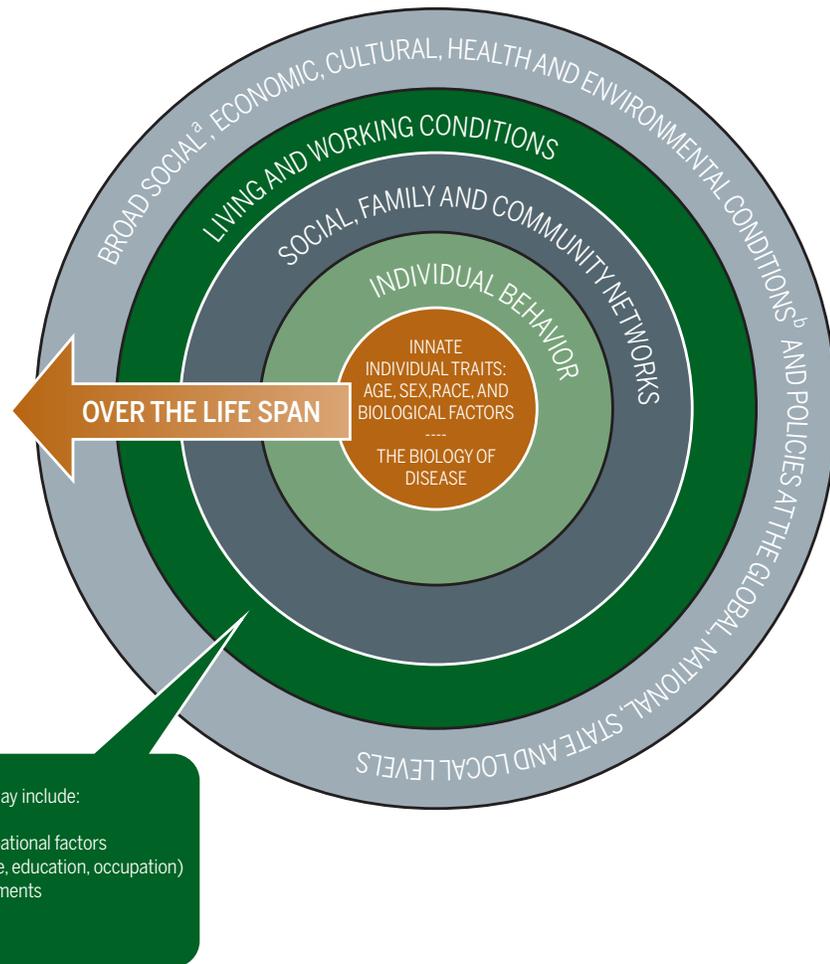
Three discussion groups were held: An in-person discussion group at Genetic Alliance’s annual conference; a teleconference with the National Community Committee’s Special Interest Group on Genomics

(hereafter, NCC SPIG) and a teleconference with a select group of public health practitioners including several genetic counselors working in the public health setting. Themes that appeared in all three discussions (i.e. health outcomes, genomics/health literacy, and family history) were identified and are integrated into this report. Paraphrases from the discussions that seemed fitting appear in this report and in the table of recommendations.

#### Frameworks

The Planning Committee noted the parallel between the fragmentation of public health genomics pointed out by respondents and the “disarray” of the U.S. public health system noted in the 1988 Institute of Medicine report, *The Future of Public Health*. That report began a process of developing frameworks useful in recognizing relationships among various sectors of public health, between research and practice, and among various types of public health services. The Planning Committee concluded that it would be useful to view the recommendations that emerged from the Stakeholder Consultation against these frameworks to foster understanding of their relationships and the way they connect with the public health system. The committee felt that the two most useful frameworks, developed from the 1988 *Future* report and a subsequent 2003 IOM report, *The Future of the Public’s Health in the 21st Century*, are the ecological framework and the framework of public health core functions and essential services.<sup>1</sup>

<sup>1</sup>Committee for the Study of the Future of Public Health; Division of Health Care Services, Institute of Medicine. *The Future of Public Health*. The National Academies Press, 1988. Committee on Assuring the Health of the Public in the 21st Century; Institute of Medicine. *The Future of the Public’s Health in the 21st Century*. The National Academies Press, 2003.



### Ecological Framework

The Ecological Framework, depicted in the second *Future* report by the graphic on this page, relates the biological factors responsible for disease to successive levels of behavioral, social, family, community, workplace, environmental and structural factors, with each level affecting all levels within it.

Successive versions of this framework have incorporated genomics by including the various biological pathways involving genes, proteins and cells, reflecting a molecular perspective on disease causation.

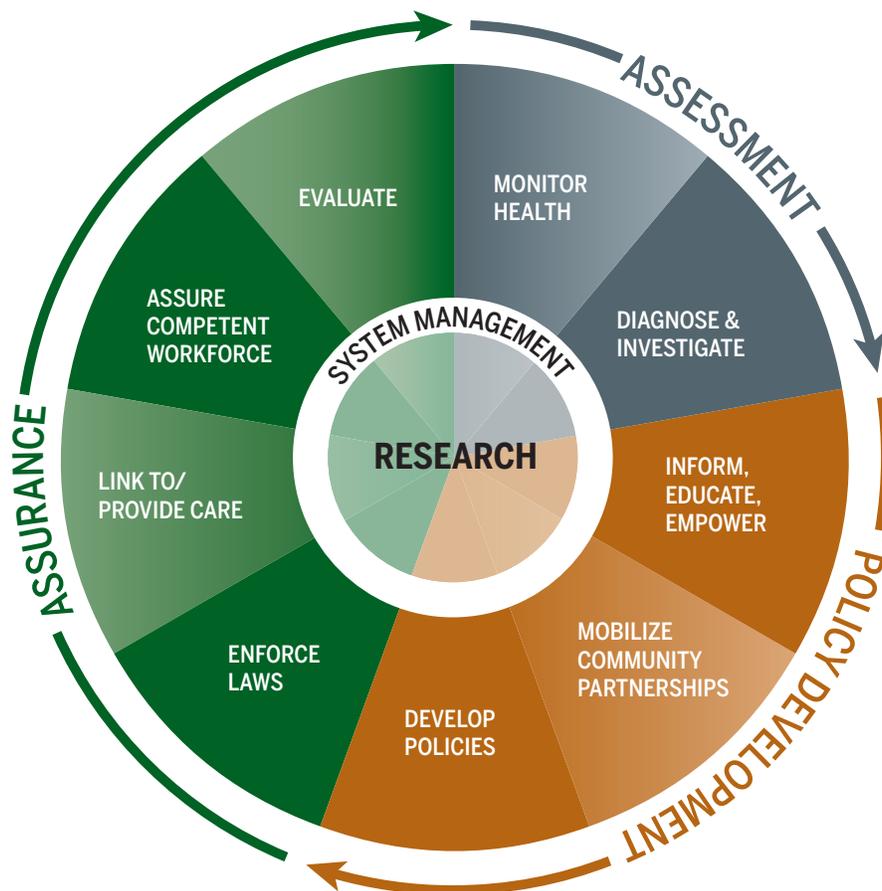
The ecological framework was identified by the Institute of Medicine in its *Future*

*of the Public's Health in the 21st Century* report and a parallel report on the teaching of public health, *Who Will Keep the Public Healthy?*<sup>2</sup> as the framework that should be taught to and used by all public health practitioners and students. It can assist those engaged in public health genomics to better convey the relevance of their field to all public health researchers, practitioners and to the public at large. It can help gain support for the kind of transdisciplinary research and its applications that are noted above. It can also help avoid the “zero sum game” mentality that has often inhibited collaborative research between social scientists and geneticists, each feeling that increased emphasis on one area of research or practice inevitably reduces the emphasis on the other area.

### Public Health in America

The 1988 *Future of Public Health* report developed the framework identifying three core functions of public health—assessment, policy development and assurance—each connecting to the two other core functions. In 1994, the *Public Health in America* project issued its framework dividing the three core functions into 10 essential public health services, each related to research and systems management<sup>3</sup> (see page 15).

In 2001 the Association of State and Territorial Health Officials (ASTHO) issued its *Framework for Public Health Genetics Policies and Practices in State and Local Public Health Agencies* utilizing the *Public Health in America* framework as a



guide to health departments integrating genetics into their programs.

The Planning Committee felt that this framework could be a useful tool for organizing the various recommendations received from the Stakeholder Consultation process. The advantages of utilizing this framework included: (1) assisting all sectors of the public health workforce to recognize how genomics relates to their work; (2) maximizing the “reach” of genomics in strengthening programs

throughout public health practice; (3) leveraging funding for genomics under labels often not utilizing the terms “genetics” or “genomics”; (4) focusing research on practices and goals of public health, and (5) building alliances and securing advocates for genomics throughout the public health system.

To facilitate the utilization of the *Public Health in America* framework we have organized the various recommendations emerging from our Stakeholder

Consultation and the interviews conducted by Genetic Alliance into a table based on this framework. It appears in Chapter VI of this report.

<sup>2</sup>Committee on Educating Public Health Professionals for the 21st Century; Board on Health Promotion and Disease Prevention, Institute of Medicine. *Who Will Keep The Public Healthy? Educating Public Health Professionals for the 21st Century*. The National Academies Press, 2003.

<sup>3</sup>Public Health Functions Steering Committee, *Public Health in America* (1994). See <http://www.health.gov/phfunctions/public.htm>.

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**“I BELIEVE WE ARE STILL A HEALTH ILLITERATE NATION AS IT RELATES TO GENETICS AND GENOMICS....”**

—FORMER SURGEON GENERAL RICHARD CARMONA, EMAIL #1<sup>4</sup>

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## QUICK REFERENCE



For quick reference, see the “COLLAB” (collaboration) and “HD/COM” (health disparities/ community engagement) icons throughout this chapter. A summary of recommendations that also identifies key actors who might execute the recommendations contained in this report are included in the table that follows this chapter.

## V. WHAT WE LEARNED

This chapter summarizes recommendations provided by all stakeholders. It is divided into six thematic sections: education; research; assurance; policy development; health applications, and infrastructure and capacity. Two additional crosscutting themes, collaboration and health disparities/community engagement, are highlighted throughout the following six sections.

### EDUCATION: PROFESSIONAL AND PUBLIC LITERACY

Education provides the foundation to engage individuals, families and communities in informed decision-making and participation in improved health. Culturally competent, relevant and appropriate health education can serve as a conduit to healthier living. As genomic technologies emerge and groundbreaking research is translated into a better understanding of health and disease, genomics literacy and education for the public is imperative. In addition to an informed public, health care professionals must be prepared and positioned to provide accurate information to patients and communities. A well-informed public and competent healthcare workforce will promote a deeper and broader collective understanding of how genomics can and will impact population health.

#### Recommendations

Assuring our health professionals and the public are knowledgeable about genomics

to allow for informed decisions based on appropriate uses of genetic and genomic information is a priority. This can be accomplished by preparing our health professional workforce; engaging communities using social marketing and health communication strategies; implementing education policies at multiple levels; and creating infrastructure that rewards collaboration across academia, public health practice, educational systems and organizations, non-profit and private sectors and the community-at-large.

#### 1. Integrate genomics into health professional programs of study (e.g., schools of public health, medicine, nursing, dentistry, pharmacy, etc.).

Stakeholders stressed the necessity of developing curricula and integrating them into programs of study such as schools of

into primary care education as a common thread throughout coursework and not simply as a specialized focus (Leonard Levy, comment #17); training nurses to collect three-generation pedigrees and perform family history assessments, identify major risk factors, and refer to genetic services appropriately (Laurie Badzek, mail #1); and, finally, weaving genomics into the public health culture so that individuals in all areas of public health (practice, academia and research) will either have the knowledge requirements within their specific area or be able to recognize and identify collaborative partners (Sylvia Au, comment #60). In addition to increasing health professionals trained in genomics, there is need to “create a diverse genomics workforce, including people from traditionally underrepresented and

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**“CURRICULAR CHANGE AND MODIFICATION COULD CREATE AN AWARENESS AMONG THE MEDICAL DOCTORS, EDUCATORS, AND OTHER HEALTH PROFESSIONALS ABOUT THE IMPACT OF NON-MODIFIABLE RISKS AND FASHION PRIMARY PREVENTIVE PROGRAMS ABOUT WAYS TO PREEMPT THEM.”**

—WILLIAM EBOMOVI, CHICAGO STATE UNIVERSITY, COMMENT #27

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medicine, schools of public health and schools of nursing to build a prepared workforce of genomically informed health care professionals. Concrete suggestions included: integrating genomics

underserved populations” (Kristi Zonno, comment #39).

These recommendations are not unlike those issued by the CDC’s OPHG and provided in the Report of the Secretary’s

<sup>4</sup>Comments and emails cited in this chapter were numbered in order of date received. The numbering system corresponds with the one used in the “Summaries of Responses to Request for Information” document on the website of the APHA Genomics Forum, [www.genomicsforum.org](http://www.genomicsforum.org).

Advisory Committee on Genetics Education and Training. In 2001, the OPHG and the Office of Surveillance, Epidemiology, and Laboratory Services developed competencies for public health professionals to ensure that public health workers maintain a basic understanding of genomics and its role in disease, and can appropriately refer individuals to genomic resources. Different competencies were created to meet the needs of public health professionals in different areas of the workforce—including leaders and administrators, clinicians, epidemiologists, health educators, laboratorians, and environmental health workers.<sup>5</sup> A more recent report from the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) has consistently recognized the importance of professional and public genetics education and

well informed in public health genomics. In 2007, the National Coalition for Health Professional Education in Genetics developed a list of core competencies surrounding provider knowledge, skills, and attitudes in genetics and genomics (National Coalition for Health Professional Education in Genetics, 2007). Stakeholder Laura Senier noted the lack of research to explore and evaluate those competencies and how, if at all, they have been integrated into training programs. Furthermore, there is a need to “develop a series of model curricula that will show educational institutions and professional membership organizations how to integrate the genomics competencies into existing educational and training programs” (Laura Senier, comment #16). Certification and accreditation should also be integrated into continuing

### 3. Integrate genomics into K-12 education (e.g., high school biology)

Well before students enter into undergraduate and graduate programs of study, there is a need to incorporate genomics into K-12 education by examining and adjusting science and health curricula. Curricula may aim to expand genetically and focus on concepts such as carrier screening and risk assessment of disease (Tricia Page, comment #21). The SACGHS Genetics Education and Training report states that “there have been persistent calls for improving science curricula overall and genetics content in particular, with emphasis on the need to shift the focus of genetic education from single-gene, qualitative traits to complex traits” (SACGHS, 2011). Revamping school science education will build a foundation of knowledge and empowerment by engaging students in learning the role genes play in common chronic diseases such as cancer, heart disease, and diabetes, and how external and internal factors affect health. Educating interested students early on will help to foster the development of a genomically informed public.

## “MULTIFACETED EDUCATIONAL ACTIVITIES ARE BADLY NEEDED AND MUST TARGET THE PUBLIC HEALTH WORKFORCE, PUBLIC AT LARGE AND PROVIDERS, USING PILOTED TOOLS, PRACTICE GUIDELINES, AND BEST-PRACTICE STANDARDS.”

—MICHAEL WATSON, AMERICAN COLLEGE OF MEDICAL GENETICS, COMMENT #31

training to assure that genomic research findings benefit the public's health. Recommendations to improve education and literacy with regard to genomics and genetics have been included in nearly every SACGHS report issued to date.<sup>6</sup>

### 2. Train the existing health professional workforce

There was consensus among stakeholders that health care professionals are not

education and training programs (James Madara, comment #46). Practicing health care professionals will require learning modules that are dynamic and easily incorporated into daily routines, as genomics information is still relatively new information for providers. Several stakeholders pointed to the use of electronic medical records to coordinate patient data and integrate guidelines for genomics-based applications, as well as using piloted tools and assuring best-practice standards.

### 4. Engage communities in genomics education



Community engagement is an essential process to enhance the genetic and genomic literacy of the public, and the community must be a strong voice in the genomics movement (NCC SPIG, informal discussion group). Community engagement efforts with collaborative ties to academics and public health practitioners, said key informant Wylie Burke, will be necessary to bridge communication gaps and further genomics education. Stakeholders noted that the most effective ways to engage and empower the community are

<sup>5</sup>Genomics Workforce Competencies 2001. <http://www.cdc.gov/genomics/translation/competencies/>

<sup>6</sup>Secretary's Advisory Committee on Genetics, Health, and Society. (February 2011). *Genetics Training and Education: Report of the Secretary's Advisory Committee on Genetics, Health, and Society*. See [http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS\\_education\\_report\\_2011.pdf](http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_education_report_2011.pdf)

<sup>7</sup>See, for example, the University of Utah Genetics Science Learning Center. Tour of the Basics. <http://learn.genetics.utah.edu/content/begin/tour/>

# Education in the ‘New Genetic Era’



**BARBARA BURNS MCGRATH**, RESEARCH ASSOCIATE PROFESSOR, SCHOOL OF NURSING,  
UNIVERSITY OF WASHINGTON; STAKEHOLDER CONSULTATION PLANNING COMMITTEE

Genomics entered my radar screen during the Human Genome Project era when I was asked to write about some of the potential implications for society when, as it was referred to then, the Book of Life revealed its secrets.

Ten years later few of us have been directly impacted by discoveries made in the new genetic era. That doesn't mean we haven't been changed in some ways. I think there have been shifts in the ways we think about our bodies and illness. While searching for cure, the clinical gaze now looks deeper and deeper into our cells and molecules. That is a new view of the body and behavior. We acknowledge the role environmental and social determinants play in disease, and research continues, but the discovery of genes receives greater attention by funders, the media, and the public. At a time when information seeking in general is a popular pastime, consumers and patients are exerting their right to knowledge that was once held and protected by specialists. Direct to consumer marketing of personal genomic testing, advocacy group biobanks, freely accessible genomic databases, and return of research results to study participants are trends that have an impact beyond genetics. In the midst of these changes, the field of public health is finding a new voice and created a new specialty, public health genomics.

The toll that chronic non-communicable diseases takes on global populations is recognized by private and public health organizations, governments, and by a new generation of bright young students entering the field of global health. The contributions of public health are obvious in this context, with the field of public health genomics positioned to play a key role. Practitioners speak the language of genetic science and bio technology, and at the same time are comfortable in the

knowledge that there are other contributions to disease beyond biology. Competencies within the public health workforce are relevant here, such as the integration of medical discoveries into public policy, application of behavioral science theory for health promotion, and consumer advocacy, to list just a few.

The new normal doesn't seem to be going away with its steady state of new knowledge, rapid change, and endless innovation coming at us at with breathtaking speed. There is consensus that educational programs for health professional have not keep pace. This factor, among others, limits the integration and evaluation of genetics into health care. It is not realistic to assume that traditional methods of presenting accepted scientific truths in textbooks, or at annual continuing education lectures are adequate. New approaches are called for, such as the use of dynamic web-based learning modules, or incorporating decision support tools in electronic health records. Health care and public health policy do not occur in a vacuum, and there is need for a genomically informed public. The emerging trend of the democratization of knowledge provides great opportunities for creative genetics education and communication strategies to be developed that target consumers and patients of all literacy levels.

In February 2011 the Secretary's Advisory Committee on Genetics, Health, and Society released its report, "Genetics Education and Training," which is available at [http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS\\_education\\_report\\_2011.pdf](http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_education_report_2011.pdf). The report describes the genetics education and training needs of point-of-care health professionals, the public health workforce, and patients and consumers. Six recommendations were submitted to the Secretary of HHS.

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**“COMMUNITY ENGAGEMENT EFFORTS, IN COLLABORATION WITH ACADEMICS AND PUBLIC HEALTH PRACTITIONERS, ARE NECESSARY TO BRIDGE COMMUNICATION GAPS AND FURTHER GENOMICS EDUCATION.”**

—KIM KAPHINGST (PARAPHRASED), WASHINGTON UNIVERSITY IN ST. LOUIS, KEY INFORMANT INTERVIEW

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through “genetics 101” education<sup>7</sup> and the collection of family health histories. Other consumers are informed but yearn for reliable sources of up-to-date information. Because communities have varying levels of health literacy and different health needs, tailored, culturally competent education and social marketing strategies are key factors in informing the public.

Stakeholders also expressed the need for minority buy-in. The NCC SPIG group discussed a number of strategies for moving the genomics agenda forward in communities with equity and parity in mind. The group suggested connecting with community leaders, community health workers, faith-based organizations, and community fairs to disseminate education and information, providing an understanding of the role of genetics in

predisposition to diseases. NCC SPIG members additionally stressed the importance of understanding family health history as well as how behaviors and environment can impact health and disease. There was strong emphasis from stakeholders to use education as a means to close knowledge gaps to help reduce health disparities (NCC, informal discussion group).

**5. Build partnerships and collaborations in academia, public health practice and communities to promote genetic and genomic literacy**



A crosscutting theme present throughout all topic areas in the Stakeholder Consultation is the push for collaboration and partnership. While public health has the power to serve as a convener of multiple disciplines, stakeholders stated

constituents in public health genomics continue to operate in silos. This causes disjointed communication and unclear messages about what public health genomics is and what it isn't. This trend persists in education, too. A number of stakeholders spoke specifically about the schism between clinical medicine and public health. As translation moves into practice, an informed public and healthcare workforce will require collaborative efforts among researchers, physicians, nurses, dentists, public health professionals, medical geneticists and genetic counselors, social and behavioral scientists, legislators and communities. There is a need for multidisciplinary teams that can envision a competent healthcare workforce and genomically educated communities.

# Community Representation



**ELLA GREENE-MOTON**, COMMUNITY EDUCATION COORDINATOR, CENTER FOR PUBLIC HEALTH AND COMMUNITY GENOMICS, UNIVERSITY OF MICHIGAN SCHOOL OF PUBLIC HEALTH; FACILITATOR, NCC SPIG; STAKEHOLDER CONSULTATION PLANNING COMMITTEE



As I began to consider how I would approach expressing my thoughts on the future of public health genomics over the next five years, it struck me that the most telling aspect of this whole process, from a community perspective, is to be invited to the table. I was reminded of a comment captured in the CDC-funded Prevention Research Centers' National Community Committee's Collective Voices publication (2009): "...The community voice is woefully quiet." It also reminded me of the constant struggles involved in assuring that the community's voice is even invited to the table and ultimately included in these types of groundbreaking

discussions. One glaring reality though, is if it were not for forward-thinking individuals (and you know who you are) who are always on guard and ready to ask, where would the community (grassroots) representation be? As we tease out the agenda of public health genomics for the next five years (and beyond), I would ask that we continue to seek out that voice, embracing the idea that community lies at the heart of public health and that using an engaged approach will provide the opportunity and space for a more valued understanding, acceptance, and implementation of the recommendations generated from these discussions, based on the input from a diverse group of stakeholders....

# Respecting Communities



**WINONA HOLLINS HAUGE**, WASHINGTON COMMISSION FOR AFRICAN AMERICAN AFFAIRS, UNIVERSITY OF WASHINGTON, HEALTH PROMOTION RESEARCH CENTER; REPRESENTATIVE, NCC SPIG; STAKEHOLDER CONSULTATION PLANNING COMMITTEE



My vision for the role of public health genomics includes [this wisdom from] Elie Wiesel, from *The Nazi Doctors and the Nuremberg Code*: "We must not see (any) person as an abstraction. Instead, we must see in every person a universe with its own secrets, with its own treasures, with its own anguish and with some measure of

triumph." In my vision we would use our collective vision and voice to include the communities that we serve.

...We share [responsibility] as leaders of the [GenoCommunity Think Tank meeting] to work toward a new vision and work that fully embraces inclusion, equity, parity, bioethics, translational science, and community based practice.

## RESEARCH

Research is the vital groundwork that explores and investigates new technologies, interventions, and better strategies for healthier living. The *Public Health in America* framework prioritizes research insights and innovations stating: “Findings must be analyzed through a public health lens...” Over the last 10 years, since the completion of the Human Genome Project, genomics research has focused on the discovery phase of research. As new discoveries emerge, research agendas must focus on translation to reach all populations with the goal of improving health outcomes. Additionally, research must explore built environments and the interaction between environmental and genetic factors, as well as assess ethical, social and legal implications of new information and health interventions.

### Recommendations

“...We must first provide leadership and advocate for a robust research agenda in public health genomics. This research agenda should include a focus on evaluating genes, environmental factors and behaviors, and the related economic,

understood examples of what we do and how genomic information can be successfully integrated into clinical and public health settings” (Karen Edwards, Professor, Department of Epidemiology; Director, Institute for Public Health Genetics, University of Washington).

Genomics still predominantly resides in the laboratory. With some notable exceptions, there are few clinical tools and health interventions to date. Stakeholders consulted for this report repeatedly highlighted research as a priority and expressed the need for translation into clinical uses and guidelines. In the next five years, research agendas should include: building a strong evidence base; focusing on translation into improved health outcomes; creating large databases of phenotype-genotype information and encouraging data sharing; creating multidisciplinary research teams to explore external and internal impacts of the genome, and engaging communities. At the same time, “also needed are transparent discussions around ethical and legal issues, including protection of privacy, data acquisition, storage, use, interpretation and dissemination in the area of human genomics” (James M. Hughes, comment #53).

regarding the clinical utility of currently available genomics interventions in the clinic or in the public health arena. Stakeholders expressed that top priorities in the next five years should be to conduct public health research on the impact of genetic factors on health and disease, the interaction between genetic and social/environmental factors, and economic, ethical, social and legal issues. There is a need to demonstrate cost-effectiveness and net benefits of genomic applications as compared to interventions that ignore genetic and genomic differences among individuals and populations. With a strong evidence base, public health and healthcare professionals can move to the implementation stage, assuring appropriate and well-designed interventions.

Critiques from stakeholders emphasized that, due to a lack of sound evidence, genomics is just not “ready for prime time” at a population health level. Genomics is still in the research phase and, given funding constraints, key informant Steven Teutsch noted, that is (with the exception of newborn screening) where it should remain. Key informant Chris Kuzawa cautioned that the role public health genomics can play is especially unclear for conditions such as obesity that involve multiple pathways. It is too great of a leap for public health genomics to go from large, integrated phenotypes to nucleotides; rather the focus should be on epigenetic mechanisms (Chris Kuzawa, key informant interview). Other stakeholders shared a sense of optimism about the impact that public health genomics can and will have on population health, if the research agenda is advanced with a public health lens.

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**“PUBLIC HEALTH RESEARCH ALLOWS THE NATION TO HAVE A ‘REALITY CHECK’ ON HOW GENETIC INFORMATION IS BEING USED IN PRACTICE AND ENSURES THAT ALL SEGMENTS OF THE POPULATION WILL BENEFIT FROM NEW GENETIC KNOWLEDGE.”**

—MUIN KHOURY, TESTIMONY ON GENOMICS RESEARCH IN THE 21ST CENTURY BEFORE THE HOUSE SUBCOMMITTEE ON HEALTH, COMMITTEE ON ENERGY AND COMMERCE, U.S. HOUSE OF REPRESENTATIVES, MAY 22, 2003

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ethical, legal and social issues. This information should then be used to develop and evaluate appropriate interventions. Secondly, we need to present a balanced view regarding the utility and potential application of genomic information. Third, we need to better define public health genomics and provide easily

#### 1. Develop and implement research that leads to a strong evidence base for public health action

Due to the nature of public health genomics, some health outcomes may take years or generations to accrue, but genomics research has the potential to greatly benefit population health. Yet there is still an absence of a strong evidence base

Genetic testing for cancer susceptibility is already enabling thousands to take steps to significantly reduce their risk. The movement around Sudden Cardiac Death of the Young is another prime example of moving research into implementation. See the “Health Applications” section of this chapter.

# Translation



**SHARON KARDIA**, CO-DIRECTOR, CENTER FOR PUBLIC HEALTH AND COMMUNITY GENOMICS, PROFESSOR AND CHAIR, DEPARTMENT OF EPIDEMIOLOGY, UNIVERSITY OF MICHIGAN SCHOOL OF PUBLIC HEALTH, STAKEHOLDER CONSULTATION PLANNING COMMITTEE

Public health genetics has a very long history in the United States dating back to the turn of the last century. At that time, its emphasis was on the health of the gene pool through methods and messages we now recognize as misguided. However, its presence in the nation's Public Health Service was a testimony to the fact that we are genetic beings. It wasn't until the early 1960's that the first applied molecular testing for inborn errors of metabolism became available and accurate at a population level. With the completion of the Human Genome Project, the list of known rare genetic diseases has grown dramatically and we are finally beginning to understand the genetic contributors to common chronic diseases across the world's populations. The genomic technologies capable of accurately measuring the base-pair sequences, epigenomic landscapes, transcriptomic profiles, and complex metabolic pictures of our bodies are becoming affordable and available for population studies of disease. The key question before us now is how to use the incredible advancements in the ability to measure our human biology to improve the public's health. Will it be by slowly and

surely adding to the already very successful, though admittedly limited Newborn Screening Programs, or is there another whole venue of public health genomics that is ready to emerge? What would it look like? Perhaps it could be a partnership between departments of health (local and state), the many "mini-publics" of local hospitals and healthcare providers (complete with biobanks and electronic medical records), academic researchers, and biomedical industry. What would they do? They would create and ride the health information highways and social networks doing everything from identifying local health issues to studying the relevant local genetic and environmental risk factors and their interactions to sending information to local doctors, industry reps, and communities in culturally relevant ways that maximize understanding, development of new solutions, and intervention. In other words, public health genomics in the 21st century would be neighborly and community-minded so that it could contribute to the public health village in as many ways as it can....

**“THERE IS A CLEAR NEED FOR GREATER RIGOR IN TRANSLATION IN WHICH OPHG HAS SHOWN INTERNATIONAL LEADERSHIP AND IMPACT, AND THE EVIDENCE TAKES TIME TO DEVELOP. LACK OF RECOGNITION THAT STRATEGIC INVESTMENT IN INITIATIVES THAT DO NOT LEAD TO IMMEDIATE HEALTH GAIN, BUT ARE OF ENORMOUS POTENTIAL IMPORTANCE IN THE LONGER TERM, IS A BARRIER THAT NEEDS TO BE OVERCOME.”**

—JULIAN LITTLE, UNIVERSITY OF OTTAWA, COMMENT #42

### 2. Direct research toward translation, shifting focus to health outcomes

Genomics stakeholders to date have put much of their effort into the discovery phase of research. But the stakeholders consulted expressed the need for a shift of focus to prioritize the translation phase, to apply promising advancements in biology to improving the public's health. Several stakeholders agreed that by shifting genomics research from basic research toward translational research, public health will become better equipped to streamline discoveries into clinical practice. By focusing research efforts into translation, innovation can move into health applications, reaching populations and improving health outcomes.

### 3. Create large phenotype-genotype databases and infrastructure for data sharing

Several stakeholders proposed the need for large population databases to catalog phenotypic-genotypic, demographic, socioeconomic, environmental, and behavioral data in order to explore genetic and external environmental contributions to disease. Large databases would store “relevant clinical and transactional information, including genetic contributions to health and outcomes, from health systems around the country...” (Maki Moussavi, comment #28). In addition to creating these large databases, many stakeholders noted the need for data sharing across researchers, states and disciplines. “The research community needs to adopt a new set of behavioral norms to fully exploit genomic data where multiple investigators participate in, and

are appropriately acknowledged and rewarded for, contributing to common, pre-competitive projects” (Jonathan Izant, comment #59). The CDC OPHG could provide leadership to ensure infrastructure for streamlining of data sharing.

Maintaining large databases can potentially allow scientists to examine risk across populations through segmentation, improving an understanding of why some individuals and/or populations develop disease and some remain healthy. Databases may lead to a priority in “research to provide evidence that segmentation of populations improves the effectiveness and efficiency of public health

interventions, especially in the fields of obesity, diabetes, heart disease, stroke, cancer, and neurodegenerative disorders” (Ron Zimmern, email #3).

In order for genomics to reach full potential in translation, there must be “at least one entity that is held responsible for warehousing population genetic/genomic research findings, spearheading the standardization of risk prediction from research findings, assisting with interpretation of risk prediction results, and ushering genetic/genomic findings through the translational research process” (Ann Cashion, Yvette Conley and Lorraine Frazier, comment #34).

## Project Highlight: The Michigan Neonatal Biobank

Michigan has been creative in using genomic information. The Michigan Neonatal Biobank stores residual newborn screening blood spots, dating back to 1984, which can be used for health research. Paired with population data, these samples can be used to tie together genetics and environment and used to understand health disparities. Collaborations with academics will be necessary to translate research that comes out of such endeavors.

See Jean Chabut, Chief Administrative Officer, Michigan Department of Community Health, key informant interview.

Safeguards for confidentiality, legal protections and community engagement will protect the public and promote trust as databases and data-sharing infrastructures grow. “The lack of a diverse research subject population, perhaps because certain communities choose not to participate, can be overcome by an electronic health environment where information is appropriately de-identified and anonymized, as well as by engaging those communities at the grassroots level through traditional public health methodologies...” (Michael Watson, comment #31).

#### 4. Create multidisciplinary research teams and facilitate collaboration



There are a number of reasons why multidisciplinary teams should collaborate in public health genomics research. By definition, genomics calls for the exploration of the interaction between social and physical environments and genetic factors, which implies a need for expertise beyond biological science. There is a need, in fact, to “explicitly [bring] together social and biological models of disease and show

that both are needed for optimal public health outcomes” (Richard Carmona, email #1). Multidisciplinary teams will ideally comprise biomedical researchers, epidemiologists, medical geneticists, genetic counselors, ethicists, and social and behavioral scientists, who will provide area expertise to illuminate the complexities of genomics in varying segments of the population. Understanding the genome outside of a “vacuum” and in the context of complex, external influences will require a level of creativity, which can be achieved through building “dream teams” of researchers and stakeholders across

disciplines. Research that contributes to our understanding of these complex interactions will need to focus on the full range of research strategies addressing the interactions between genetic and non-genetic contributors to health, including improved strategies to define phenotypes and social contributors to health, epigenetics, ecogenetics, and microbiome studies (Wylie Burke, comment #56).

Additionally, to make most effective use of funding, public health genomics must be integrated into multidisciplinary research and preexisting research agendas (e.g., cancer, heart disease, diabetes).

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**“INTEGRATION OF GENOMIC DATASETS FROM MULTIPLE INVESTIGATORS COUPLED WITH COLLECTIVE, ITERATIVE ANALYSIS WILL HELP BUILD ACCURATE AND PREDICTIVE MODELS OF DISEASE THAT INDUSTRY AND ACADEMIA CAN USE TO REDUCE BOTH THE INDIVIDUAL AND THE COMMUNITY BURDEN OF DISEASE.”**

—JONATHAN IZANT, SAGE BIONETWORKS, COMMENT #59

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## Genes and the environment

Key informant Kenneth Olden discussed the need for multidisciplinary research teams to study both the environmental and genetic risk factors that cause disease. In order to identify environmental health risk factors, one needs to understand variations in gene frequencies. Most importantly, he noted, there is a need to integrate genetics with epigenetics. A combination of genetics and the environment together is going to be the risk that is largely responsible for chronic disease. We can use genetics to identify at-risk populations and also identify differences in environmental factors (e.g., East Harlem vs. downtown New York City). There is a need for a concept that recognizes the importance of all determinants (genetics, social behaviors, environment, etc.) because it's the interaction that will be crucial to examine. It's really a combination of the total environment and total genetic makeup.

See Kenneth Olden, Founding Dean, CUNY School of Public Health, key informant interview



genomics must leverage the public health system to assure that genetics services and technologies are safe, effective, and appropriately integrated into the healthcare setting by providing assurance through an evidence base. Providers, payers, and researchers must collaborate to evaluate the clinical validity and clinical utility of genetic tests. The results of such evaluations should be made available to the public to facilitate decision-making for all stakeholders. Researchers must work with providers to develop realistic guidelines for implementing genetic tests and services in the clinic. Finally, public health genomics should assure access to appropriate collection and analysis of family health history information, and to genetic tests with proven clinical utility.

### 5. Engage community in research agendas



Community voices are imperative and must be present in the discussion around ongoing issues in evidence development and evaluation (Genetic Alliance Conference, informal discussion group). Mistakes and abuses in the past have led to mistrust and skepticism surrounding research among specific communities. “Education and outreach to American Indian and Alaska Native communities about genomics research and its public health implications is also critical. There is a great deal of suspicion and misinformation about genetics research in Indian Country” (Jacqueline Johnson Pata, comment #30). Key informant Chikezie Maduka stated that collaborations and partnerships must be made with academics and the community to foster trust. It’s a win-win for researchers and community members, as the researcher can conduct a study and community members benefit from learning about the research topic

(Chikezie Maduka, key informant interview). Additionally, building partnerships will help to facilitate conversation with communities to identify and address their greatest needs and concerns. Research agendas must strive for transparency and allow community members to sit at the table and serve as leaders and decision-makers. Funding and university policy must also work to support incentivizing community-based participatory research.

## ASSURANCE

Assurance is an essential function of public health. As the Institute of Medicine’s 1988 “The Future of Public Health” report describes, “Public health agencies assure their constituents that services necessary to achieve goals are provided, either by encouraging action by other private or public entities, by requiring such action through regulation, or by providing services directly.” Similarly, public health

## Recommendations

In order for public health genomics to fulfill its assurance duties, genetic tests must be evaluated to ensure safety and value, and the results should be shared in a public database. Research results need to be translated into practical guidelines to move tests from the lab to the clinic. So far, limited data are available to confirm the value of genetic testing; however, family health history is an inexpensive and widely accepted tool that could serve as a pilot to test the value of incorporating genomics into healthcare and patient education.

### 1. Evaluate genetic tests for clinical validity and utility

Public health genomics uses analytic validity, clinical validity, and clinical utility as measures for the effectiveness and quality of a genetic test, service, or technology.<sup>8</sup> To facilitate the evaluation of genetic tests for clinical validity and clinical utility, “methods for evaluation of genomic tests can be adapted by other

<sup>8</sup>Analytic validity refers to the accuracy and precision of the laboratory test in detecting a genetic marker. (Hogarth S, Javitt G, Melzer D. 2008. The current landscape for direct-to-consumer genetic testing: legal, ethical, and policy issues. *Annu. Rev. Genomics Hum. Genet.* 9:161–82.) A test’s ability to predict a disease outcome is clinical validity. (Hogarth S, Javitt G, Melzer D, 2008.) Clinical utility describes how well the genetic test prevents or improves a disease outcome. (Hogarth S, Javitt G, Melzer D, 2008.) Currently, analytic validity is evaluated by the Centers for Medicaid and Medicare Services; however, clinical validity and clinical utility are not assessed by a uniform system.

governmental organizations such as the Agency for Healthcare Research and Quality (AHRQ) and the National Institutes of Health (NIH), institutions that develop clinical guidelines, and healthcare payers” (Wylie Burke, comment #56).

By using already existing formulas and methods, time, money, and resources are saved. These methods must include “an approach to [demonstrate] the incremental value of genomics compared to other strategies (impact and cost effectiveness)” (Steven Teutsch, comment #4), as genomics should add value to the healthcare system, and not just increase costs (Julian Little, comment #42). Once methods for evaluation are accepted among stakeholders, funds will be necessary to conduct these evaluations. Stakeholders must work to “promote and fund research into the clinical utility of genetic and genomic tests. The vast majority of genetic and genomic tests are made available in the absence of any evidence that the use of such tests improves health outcomes” (Diane J. Allingham-Hawkins, comment #14).

Some respondents suggested that a priority should be “the continuation of a group such as Evaluation of Genomic Applications in Practice and Prevention (EGAPP) to provide evidence-based, authoritative recommendations on which genetic tests are useful in improving outcomes” (Herbert F. Young, comment #41). Others have stated that the CDC must become more involved in health technology assessments and the development of guidelines for genetic technologies. While it is unclear who should take the responsibility of evaluating genetic tests for clinical validity and utility, it is certain that many stakeholders must be engaged and a rigorous and consistent method must be followed.

## 2. Engage stakeholders to develop clinical guidelines



After clinical validity and clinical utility have been assessed, the results must be translated into guidelines that can be followed

and used by providers. This translation requires work from the bodies that evaluate genetic tests to ensure that the results are disseminated and implemented into practice. For example, one individual suggested “[expanding] the EGAPP Working Group activities, [and] also [developing] a mechanism for integrating EGAPP Working Group Recommendations into a coordinated implementation plan” (James Haddow, comment #2). The evidence generated by evaluations “will provide the foundation to promulgate guidelines for improved targeted screening, disease management, and prevention across the lifespan” (Michael Watson, comment #31) and improve health outcomes.

To ensure acceptance of genetic test guidelines, many stakeholders in addition to CDC must be at the table during their development. “Evidence-based guidance is needed to ensure that genomics is utilized effectively and efficiently. NIH, AHRQ, and professional groups should be at the table for the development of guidelines” (Steven Teutsch, comment #4). Suggested professional groups to involve include the American Society of Human Genetics, National Coalition for Health Professional Education in Genetics, American College of Medical Genetics, American Medical Association, American Association of Pediatrics, and others. In addition to NIH and AHRQ, the Health Resources and Services Administration, the CDC, Food and Drug Administration, and Centers for Medicare and Medicaid should also be involved to ensure that services are implemented and appropriately covered by insurers (Gurvaneeet Randhawa, email #2; Ned Colange, comment #20; Ruth Lynfield, comment #49, Kim Caple, comment #57).

A benefit of engaging professional societies in the development of guidelines is buy-in from the provider community to implement services and improve healthcare. “Providing data on the utility of genomic applications in defined clinical scenarios and increasing the capacity

of providers to apply that information will result in a more appropriate use of genomic applications in public health and healthcare” (Joan Scott, comment #47).

## 3. Create a credible, publicly available database of genomics tools and findings on their validity and utility



As evidence accrues and guidelines are developed, the information should be stored in a publicly available database. This database would benefit researchers, providers, payers, test developers, and patients by providing information on tests, including clinical validity and utility. The database should be managed to “allow the citizen, the patients and the physicians to assess what evidence exists for the validity and utility of different genetic tests available in the marketplace” (Ron Zimmern, email #3). This information will facilitate the development of tests, along with their evaluation and implementation as it can be used to build on and improve already available tests and technologies. Further, the transparency of the database will allow all stakeholders to access the same information and guide their decision-making. “A coordinated approach to evidence generation that incorporates the input of stakeholders throughout the public health and healthcare system and that broadly disseminates findings will help ensure that the best designed studies are implemented to maximize the use of limited resources” (Joan Scott, comment #47). This coordination will reduce waste and promote the translation of research results into usable and practical information, for example in the form of clinical guidelines.

## 4. Evaluate family history to demonstrate value of genetic tests, technologies, services, and tools

While there are many genetic tests available to the public, there are few that have proven their added value to healthcare for patients and the healthcare system. There is wide consensus over the need to evaluate family health history, as it is an

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**“POLICIES ARE NEEDED THAT CREATE AND APPLY TOOLS TO EVALUATE APPROPRIATE APPLICATION OF GENOMICS IN HEALTHCARE IN ORDER TO INCREASE UTILITY, ENSURE EFFECTIVE FOLLOW-UP, AND IMPROVE HEALTH OUTCOMES.”**

—MICHAEL WATSON, AMERICAN COLLEGE OF MEDICAL GENETICS, COMMENT #31

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inexpensive tool that is widely accepted, and can save lives by facilitating decision making about the likely benefit of genetic testing and the appropriateness of preventive interventions. One stakeholder recommended: “Urge CDC to devote resources to answering [effectiveness as a clinical tool] questions about family history. In this era of cost-cutting in health care, effective family history collection and interpretation is a ‘genetic technology’ that is inexpensive, and that can preclude the need for further unnecessary and expensive testing” (James Madara, comment #46). Participants in Genetic Alliance Conference discussion group came to a similar conclusion: Family health history may be one of the genomics tools that can demonstrate its value in addressing critical public health problems (Genetic Alliance Conference, informal discussion group). See the “Health Applications” section of this chapter for a more detailed discussion of family health history.

## POLICY DEVELOPMENT

As the ASTHO Framework for Public Health Genetics Policies and Practices in State and Local Public Health Agencies states, “sound health policy development requires a combination of scientific guidance and analyses of existing policies, regulations, resources, and strategic priorities. Public health policy aims to improve the health of the community while providing necessary individual protections.” Similarly, public health genomics policies are necessary to respond to

“identified problems, barriers, and needs such as genetic screening, diagnosis, treatment, and prevention programs.” Scientists must work with all stakeholders to develop policies at various levels, from federal and state policies to organizational policies to assure safe use of genomics and improved health of communities.

### Recommendations

Public health genomics policies are needed to ensure a competent workforce, a health literate population, effective research practices, safe genetic tests, and standardized clinical guidelines. The development of these policies will require input from all stakeholders, including government, public health practitioners, clinicians, industry, and the community. By incorporating the values of each of these stakeholders, the policies are likely to be implemented effectively and have their intended impact on communities and the field of public health genomics. As policies are implemented, they must be monitored to make certain that the appropriate improvements are achieved, without any unanticipated harms to society.

#### 1. Implement policies to ensure a competent workforce and a health literate nation

Genomics is a relatively young field and new information is generated rapidly each day. Continuing education courses are necessary to keep providers up-to-date, and medical and public health curricula must continue to develop genomics courses to maintain a workforce that is competent in genomics. The scientific community must partner with

professional organizations to develop competencies and implement curricula to ensure that providers can handle emerging genomic technologies and utilize these technologies to improve patient care. “CDC should partner with professional organizations such as the National Coalition for Health Professional Education in Genetics (NCHPEG) to develop a series of model curricula that will show educational institutions and professional membership organizations how to integrate the genomics competencies into existing educational and training programs” (Laura Senier, comment #16). Further, CDC should partner with professional societies and schools of public health to integrate and develop genomic competencies in master’s level programs.

In addition to educating the workforce, policies must be developed that improve the health literacy of the entire population. Genetics and science education curricula should be implemented in primary and secondary education nation-wide. “What [the IOM, AMA, and Healthy People 2020] policies do not address is the need for ensuring that the public has access to resources that provide for an understanding of genetics programs and services or the need for health care providers who can successfully communicate risk assessment and other genomic information to patients, two crucial components of genetic and genomic literacy” (Kristi Zonno, comment #39). By improving the health and genomics literacy of the population, patients will be better equipped to understand and use rapidly emerging genetic technologies.

## 2. Implement policies that further public health genomics research agendas



Stakeholders recommended many types of research policies to address various goals, from reducing health disparities to improving efficiencies in data collection. Some stressed the need to include minorities in research trials to ensure equal representation of all members of the population. Specifically, the National Congress of American Indians suggested that “the CDC have a policy requirement for affiliated researchers (including those receiving grants and contracts) to treat tribes as full partners in studies” (Jacqueline Johnson Pata, comment #30).

Policies must continually be examined to ensure that ethical guidelines and privacy protections are in place (NCC SPIG informal discussion group). “...More could be done to educate lay audiences about the range of ethical issues involved in genetic medicine, and to deliberately elicit their feedback about the scope and direction of public health genetics activities” (Laura Senier, comment #16). Community education about the ethics of public health genomics will benefit both researchers and communities by facilitating community participation in research projects.

Another stakeholder recommended: “Standards for technology need to be in place, databases of samples and data made broadly available, particularly for minorities that typically do not participate in studies in general and genetics in particular” (Steven Galen, comment #19). Others highlighted the need for policies that encourage translational research, as the current funding prioritizes research and ethical, legal and social issues, and not enough money is going towards integration and translation of genomics (Public Health Practice, informal discussion group). To ensure that research dollars are used to develop products that improve clinical care, policies that support research on genomic technologies are essential. Groups such as EGAPP

are needed to make recommendations and develop guidelines to be integrated in public health practice (Kim Kaphingst, key informant interview).



Finally, policies must be implemented that encourage data sharing and collaborations among researchers. Such policies will eliminate waste and speed translation. “...Without commitment to translational research and sharing resources to advance our knowledge, we won’t get where we need to be. A greater focus on effective communication and dissemination of information... would help address the current concerns” (Joan Scott, comment #47). This will require policies to ensure that researchers utilize the data-sharing system and effective communications across the field, as well as policies to maintain the quality of the system.

## 3. Implement policies that ensure proper regulation of genomic technologies

Regulatory policies are needed at all stages of the research trajectory. Before genetic tests become routinely available in clinical practice, the laboratory tests must meet analytical validity standards. Federal and state governments need to “come up with some sort of rational guidelines regarding CLIA certification for genome wide genotyping or sequencing. The current guidelines are not conducive to research or clinical care” (Josh, comment #10). This regulation must be applied to all genetic tests, including those that are direct-to-consumer.

“Again, work with DTC genetic testing companies to establish guidelines for assessing the validity of their tests and interpretations” (Connie Bormans, comment #55). In addition to regulating the quality of genetic tests, policy-makers must develop policies to ensure the proper translation of technologies into clinical care. Further, regulatory agencies must ensure that insurance companies are at the table throughout the regulatory process to make certain that genetic tests are appropriately covered.

## 4. Implement policies that ensure appropriate use of genomic technologies

In addition to policies that regulate genetic tests, policies must be developed to ensure safe and effective use of genetic tests and tools in the clinic. These policies include guidelines for using genetic tools, including family health history, as well as policies to ensure that patients have adequate access to genetics specialists. “Legislation needs to be introduced and passed to require all physicians to comply with the basic standards of care and ask their patients for their family histories...and to keep such information in a confidential folder within the patient’s file and which may not be disclosed to anyone, including all insurance companies” (Maki Mousavi, comment #26). As clinical guidelines are incorporated into the clinical setting, they must be evaluated to ensure improved patient outcomes, and changed when they do not.

“Policies that foster a learning healthcare system (translational medicine) to include point-of-care education and cross-provider education will improve health outcomes and mitigate disparities by addressing access and the maldistribution of genetics providers/services” (Michael Watson, comment #31). Many stakeholders also stressed the need for genetic counselors to be licensed in all 50 states. Currently, only a few states have this requirement as a policy. States also need to work with Medicaid and private insurers to ensure that genetic counseling services are reimbursed, and therefore accessible (Public Health Practice, informal discussion group). The development of these policies will require collaborations from patients, providers, professional societies, federal and state governments, and regulatory agencies.

While implementing all these policies is no simple task, they will provide the structures to improve the quality of genetic tests, tools, technologies, and service, and ensure safe uses that improve patient outcomes.

## HEALTH APPLICATIONS: FAMILY HEALTH HISTORY AND CHRONIC DISEASE

Research, assurance, policy development, and education are all connected in the process of integrating valuable health applications into practice. There is need for processes that focus on the development of health applications that aid in prevention, especially of common chronic diseases. Public health genomics must remain clear about which health applications work well and which technologies are simply not ready for prime time. Streamlining communication about how genomics can apply to the improvement of health outcomes will be crucial to keeping the field of public health genomics alive.

### Recommendations

Healthy People 2020 includes two objectives for public health genomics: (1) Increase the proportion of women with a family history of breast and/or ovarian cancer who receive genetic counseling, and (2) increase the proportion of persons with newly diagnosed colorectal cancer who receive genetic testing to identify Lynch syndrome<sup>9</sup> (or familial colorectal cancer syndromes). Health applications such as tests for BRCA mutations and screening for risk of sudden cardiac death of the young are clear examples of how genomics can lead to prevention. Guidelines for genetic screenings and risk that lead to prevention of morbidities and mortalities align with Healthy People 2020 goals and often already have received federal buy-in. As the public health focus shifts predominantly to chronic diseases, there is a need to provide relevant, reliable, and high impact health applications as they relate to common chronic diseases. Public health must continue to provide concrete examples of what works well in practice and advocate for genomic

strategies that serve and benefit population health. Stakeholders identified prevention, the use of family health history to flag individual risk, and the integration of genomics into common chronic disease applications as priorities for the field.

#### 1. Utilize family health history

Family members share genes, behaviors, lifestyles, environments and experiences. Many families already engage in drawing connections to brothers and sisters, mothers and fathers, aunts and uncles, and grandparents. Family health history provides an opportunity to broaden the way most families already casually speak of shared genes and translate those shared genetic connections into understanding health, behavior and disease. Key informant Maxine Hayes stressed that in this funding environment, public health genomics must be careful in what is prioritized. Family history is an inexpensive tool that is easy for consumers to relate to

of surveillance and preventive measures” (Diane J. Allingham-Hawkins, comment #14).

Some individuals and communities may be hesitant to discuss certain aspects of their family history. Communication strategies promoting the use of family health history need to adopt culturally competent messages. Family health history can be used to facilitate conversation, empower individuals, identify risk, and ultimately tailor preventive strategies for additional family members. Key informant Chikezie Maduka stated that health history is one way of looking at almost all health issues, as the better people can understand their past, the better they can prepare for their future.

#### 2. Shift focus to common chronic disease

Common chronic diseases contribute to the majority of morbidity and mortality across this nation. Public health

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**“I TRULY BELIEVE FAMILY HISTORY COULD BE USED TO ACHIEVE AND SUSTAIN BEHAVIOR CHANGES THAT ARE PLAGUING OUR NATION, LIKE OBESITY, EXERCISE, CATCHING CANCER AT EARLY, MORE TREATABLE STAGES, PREVENTION OR DELAY OF DIABETES OR HEART DISEASE, GIVEN FOCUS, SUPPORT, AND FUNDING FROM CDC FOR PUBLIC HEALTH ORGANIZATIONS TO FIGURE OUT HOW TO USE THIS WITH SOCIAL MARKETING PRINCIPLES.”**

—JENNY JOHNSON, COMMENT #13

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and can be incorporated into many public health programs (Maxine Hayes, key informant interview). “Increased emphasis on family history could yield better understanding of the heritable nature of some conditions and allow better identification of those at risk, which has the potential to lead to earlier implementation

and recent funding streams have largely focused efforts on cancer, heart disease, stroke and diabetes prevention. While genetics has historically focused on rare diseases, gains have been made that improve the understanding of genetic contributions to common chronic diseases. It is now a priority for public health

# Family Health History



**IMOGENE WIGGS**, DEPARTMENT OF HEALTH & SENIOR SERVICES, BUREAU OF HEALTH PROMOTION, STAKEHOLDER CONSULTATION, PLANNING COMMITTEE

As a fraternal twin and a sibling with five other siblings I have always been curious about our lives. Some of the siblings seem so different and others are exactly like our mother or father. The biggest difference was between my twin and myself. These are some of the reasons I have been interested in family health history. It seems like you don't realize the importance of it until you develop a serious family illness, etc. Our parents never did discuss anything with us children and we didn't know anything about our health problems. Insurance was out of the question and we seldom saw a doctor if ever. We did have questions about certain behaviors but it was only in recent years that we were able to untangle some of the answers. This proved to shine a light on some questions.

It is said that family members share genes, behaviors, lifestyles, and environments that together may influence their health and their risk of chronic disease. Most people have a family health history of some chronic diseases (e.g., cancer, coronary heart disease, and diabetes) and health conditions (e.g., high blood pressure and hypercholesterolemia). People who have a close

family member with a chronic disease may have a higher risk of developing that disease than those without such a family member.

Family health history is a written or graphic record of the diseases and health conditions present in your family. A useful family health history shows three generations of your biological relatives, the age at diagnosis, and the age and cause of death of deceased family members. Family health history is a useful tool for understanding health risks and preventing disease in individuals and their close relatives.

Some people may know a lot about their family health history or only a little. It is helpful to talk with family members about your health history, write this information down, and update it from time to time. This way family members will have organized and accurate information ready to share with their health care provider. Family health history information may help health care providers determine which tests and screenings are recommended to help family members know their health risk.

to “identify chronic disease areas with greatest potential for genomics integration, and gaps where genomics can add value” (Jean Chabut, comment #50). This would include identifying opportunities for genomics to impact Healthy People 2020 objectives. Genomics does not have to be an isolated field, but rather should be integrated into existing programs and research agendas, finding partners in Prevention Research Centers (NCC SPIG,

informal discussion group) and state health departments, among others.

State departments of health have already begun to address chronic diseases through public health genomics programs providing tests for hereditary breast and ovarian cancers (BRCA mutations), colorectal cancer (Lynch syndrome), familial hypercholesterolemia, heart disease and sudden cardiac death of the young. Steps must

continue to be taken to “align all efforts at the local, state and national levels to achieve the Healthy People 2020 goals...” (Jean Chabut, comment #50). To continue to build successful programs, key informant Deborah Klein Walker suggested that state public health departments must convene groups that bring laboratory scientists, epidemiologists, and legal, maternal and child health, and chronic disease experts to the table.

## Family History

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**KAREN GREENDALE**, DIRECTOR, CANCER SURVIVORSHIP INITIATIVES, BUREAU OF CHRONIC DISEASE CONTROL, NEW YORK STATE DEPARTMENT OF HEALTH, STAKEHOLDER CONSULTATION, STAKEHOLDER CONSULTATION PLANNING COMMITTEE

My passionate hope is that all folks with personal and/or family histories indicating an increased risk for cancer—regardless of ability to pay, where they live, how much education they have had or what kind of provider they see—should be able to benefit from high quality genetic counseling and (when appropriate and desired) genetic testing. I am a three-time breast cancer survivor with a BRCA mutation who 23 years after my diagnosis am healthy, strong and looking forward to many more productive years of life. As a genetic counselor and public health professional, I can talk intelligently about the clinical utility of the tests for Hereditary Breast / Ovarian Cancer Syndrome or Hereditary Non-Polyposis Colon Cancer. As a woman who has personally had access to the services I needed, I can vouch first hand for the more ephemeral benefits—for instance, the “empowerment” and quiet satisfaction associated with knowing that I have taken whatever steps were available to me to reduce my risk of dying of breast (or especially) ovarian cancer. It is beyond distressing for me to think of individuals and families who have not had these choices for a variety of reasons, including financial barriers, misinformed providers, lack of proficiency in English, etc. Some of my work has focused on these issues, but even in other arenas, I always bring up family health history, public health genomics, and the need to look at health and health care “through a genetics lens”... these are new concepts for many working in clinical care and public health and there’s so much more to be done!

## Project Highlight

# Too Young to Die:

## Exploring the Causes of Sudden Cardiac Death in Young People



**DEBRA DUQUETTE**, GENOMICS COORDINATOR, MICHIGAN DEPARTMENT OF COMMUNITY HEALTH, STAKEHOLDER CONSULTATION PLANNING COMMITTEE

“I thought we were forgotten, I thought no one cared” are the words of a mother asked to participate in a next-of-kin interview regarding the sudden death of her teenage son. Darryl (name changed to protect privacy) had collapsed and died unexpectedly at age 18 while playing basketball in a recreational league. Bystanders did not know how to perform CPR, and no AED (Automated External Defibrillator) was available. The family never received information about the cause of his death, or whether other relatives could also be at risk. Sudden deaths in young people are especially tragic and often high profile. Not infrequently, they occur in athletes who were thought to be in their prime.

Over the last seven years, the state public health department has been working to uncover answers for families like Darryl’s through creation of a surveillance system for sudden cardiac death of the young (SCDY). The project has used multiple state data sources including death certificates, population health data (Behavioral Risk Factor Surveillance System), medical records, autopsies, next-of-kin interviews, and expert reviews to

better understand the statewide burden of SCDY. As a result, we now know that about 300 sudden cardiac or unexplained deaths occur in people between the ages of 1 and 39 each year, and 6.3% of state residents have a family history of SCDY. There are significant racial disparities. The age-adjusted mortality rate for black males is 15.8 per 100,000, more than double the rate for white males at 6.4 per 100,000.

The causes of SCDY vary, but many have a strong genomic component. Our state’s SCDY surveillance system has provided important data for action and systems changes needed to prevent future deaths. In collaboration with over 150 partners representing the medical community, schools, athletic organizations, and parent advocacy groups, the state Genomics Program staff promote changes in pre-participation sports screening; provider education; public awareness of SCDY risk factors and cardiac symptoms; CPR/AED training; emergency response and medical examiner protocols. Learn more at [www.michigan.gov/genomics](http://www.michigan.gov/genomics).

## INFRASTRUCTURE AND CAPACITY

Infrastructure and capacity for public health genomics are limited for making progress in the areas of translation, community engagement, the development of new population-based services by state health departments, the reduction of health disparities, and the development of collaborative initiatives that could set and implement priorities in this emergent field. Still, stakeholders are hopeful that key areas in public health genomics will have the capacity to move forward in years to come. Some stakeholders suggested prioritizing the least expensive (or most cost-effective) programs in public health genomics, such as integrating family health histories into practice; funding research to determine which are the most useful and cost-effective strategies, and integrating public health genomics within established and well-funded areas such as chronic disease prevention programs, particularly for cancer and diabetes. There is a current expectation that funding of governmental public health programs will be increasingly determined by evidence of lives saved and expected to be saved. Participants in the breakfast discussion held at Genetic Alliance Annual Conference noted that public health practitioners will have to develop metrics to demonstrate the value of programs utilizing genomics in public health practice.

“Recent RFAs from NIH related to genomic translation tend to lack a public health perspective—a worrisome sign about the future of research trajectories,” according to Wylie Burke, Professor and Chair of Bioethics and Humanities at the University of Washington School of Medicine. In her view, it speaks to an artificial divide of genomics into clinical applications, public health, and research. “Researchers and clinicians will be faced with huge amounts of genomic information in the future,” she said. “A population perspective is necessary to ensure a balanced approach to translation.”

## Recommendations

The following ten recommendations speak to the need to expand or increase the described activities in order to enable public health genomics to achieve maximum impact on disease prevention:

### 1. Conduct research to evaluate the utility and cost-effectiveness of genomic applications in personalized medicine and population health

We can and should recognize the limits of genetic technologies. “As public health genomics moves forward,” cautioned Steven Teutsch, Chief Science Officer for the Los Angeles Department of Health, “we need to remain skeptical. As resources are limited for public health, genetic technologies must be carefully evaluated to ensure that there is a significant net benefit before releasing technologies into practice.”

Still, technological advances are making genotyping a growing reality in public health and personalized medicine. With continuation of research into the clinical utility of genetic and genomic tests and technologies, uses will expand and funds can be channeled strategically.

“Individuals with highly penetrant genes can be identified for early treatment and intervention through genetic testing,” wrote one RFI commenter. “This technology will be most valuable for genes that are penetrant, conditions that are treatable, conditions that cannot be identified without the genetic information, and genes that are common. ...Yet, further research on clinical validity and clinical utility is necessary before integration into routine practice” (Jim Evans, email #4).

EGAPP has already been engaged in evaluating genetic tests, but as of yet, has only recommended one test. Several Stakeholder Consultation respondents pointed to the need to maintain the EGAPP Working Group as a “trusted voice for public health genomics” (Elizabeth Balkite, comment #6), and others like it that are needed to make recommendations

and formulate guidelines to be integrated into public health practice.

### 2. Conduct translational research

Many RFI respondents identified the CDC as the institution that should take the lead in facilitating translation (Ann Cashion, Yvette Conley and Lorraine Frazier, comment #33). The Personalized Medicine Coalition suggests the CDC OPHG in particular should develop a committee to promote research and appropriate access to personalized medicine products and services, and to address related issues of comparative effectiveness, reimbursement practice, benefit design and training (Amy Miller, comment #23). The CDC can serve a leadership role in the translation of microbial genetic research and its application to public health, according to respondent James M. Hughes, (comment #53); the Infectious Disease Society of America (IDSA) recommends that CDC focus on questions of public health importance, develop a prioritized research agenda, coordinate activities with NIH, and make a clear commitment to develop a forward path. “There is a clear need for greater rigour in translation in which OPHG has shown international leadership and impact, and the evidence takes time to develop. Lack of recognition that strategic investment in initiatives that do not lead to immediate health gain, but are of enormous potential importance in the longer term, is a barrier that needs to be overcome,” wrote one respondent (Julian Little, comment #42).

### 3. Conduct research focusing on community engagement and health disparities

Grant funds should promote community engagement and public education to create a genomically informed public and establish community trust, particularly among minority communities. NHGRI provides money for community events for National DNA Day, but, participants noted in the Public Health Practice informal discussion group, it can be hard to get people excited.



The challenge calls for dynamic education programs and creative approaches to communication. Also an end in itself, public trust built through community engagement may yield research benefits.

Priorities must include furthering Congress' goal of eliminating health disparities within the [American Indian/Alaskan Native] population. Outcomes of genomic knowledge should focus on elimination of health disparities in a number of areas, especially diabetes. Policies must involve AI/AN communities. Funding is a barrier, and so it is important that grant opportunities, technical assistance and outreach are made available to

Urban Indian Health Programs (D'Shane Barnett, comment #58).

**4. Integrate family histories into health practices and research**

As stakeholders repeatedly suggested (see the "Health Applications" section of this chapter), family history will be relatively easy and inexpensive to incorporate into many public health programs. Integrated into routine primary care and electronic medical records, family histories can improve preventive medicine and impact chronic conditions such as obesity, cancer and heart disease. Additionally, family histories can serve as an empowerment

tool, providing patient education and communicating health risk to individuals.

**5. Promote collaboration**



Stakeholders repeatedly identified the "silo" configuration of public health genomics "insiders" as a major obstacle in the field. Partnerships to link communities and professionals, public health and medical communities, and multiple academic disciplines will promote research and health outcome goals of public health genomics.

# Genetics in Context



**SARA SHOSTAK**, ASSISTANT PROFESSOR, BRANDEIS UNIVERSITY, DEPARTMENT OF SOCIOLOGY, STAKEHOLDER CONSULTATION PLANNING COMMITTEE

Public health genomics challenges us to focus on the irreducibly social, economic, and political factors that interact with genetics in creating health and illness across populations. These include not only access to care (including access to genetic counseling and testing), but education (as a resource for making sense of genetic information), culture (as a lens through which genetic information becomes apprehended and meaningful), and people's lived experiences of their neighborhoods and work places (which shape perceptions of probabilistic risk estimates). More broadly, public health genomics highlights the importance of addressing inequalities in environmental exposures (in the absence of which, many genetic risks are inconsequential) and opportunities to act upon the "lifestyle prescriptions" which many envision as an important outcome of genetic counseling and testing (e.g., access to nutritious foods, venues for exercise, minimizing workplace exposures, using assisted reproductive technologies, etc.). That is, even as it calls attention to genetic variations, public health genomics demands that we understand—and address—how unequally distributed exposures, resources, and other factors outside of the body enter into molecular processes to shape health and illness within and across populations.

Health departments need to develop partnerships across the public health system. In order to sustain genomics in public health, partnerships must be created with academics and the workforce. Grants are one way to encourage these collaborations (Maxine Hayes, key informant interview).

Partnerships will also extend the reach of limited grants. Since the NIH provides substantial funding to academics, for example, public health departments should partner with schools of public health to help integrate research initiatives into practice (Public Health Practice, informal discussion group).

### 6. Expand professional education

The need for an educated workforce, discussed in detail under this chapter's "Education" heading, can partially be addressed through funding initiatives, including grant programs to promote continuing education and stipends to encourage genetics experts to study public health. Respondent Laurie Badzek stressed the role that the nursing workforce will play in taking family histories, developing three-generation pedigrees, and appropriately referring individuals at risk to genetic services: "Policies in education and practice including funding that supports a practice environment that values the integration of genomics in nursing practice are needed to promote a culture where the link between genomic discovery and patient outcomes [is] clearly understood and valued" (Laurie Badzek, mail #1).

### 7. Focus on gene/environment research

Various research strategies should be supported to address the complex interactions between genetic and non-genetic contributors to health, including improved strategies to define phenotypes and social contributors to health, epigenetics, ecogenetics and microbiome studies (Wylie Burke, comment #56). (See this chapter's research section for additional detail on this topic.)

### 8. Create databases to aggregate research findings (See research and assurance sections of this chapter for a full discussion)

A priority for public health genomics should be gathering and storing genomic and genomic-related information that will be useful in genomics research, whether or not such research is carried out by the public health system or within the next five years (Genetic Alliance Conference, informal discussion group). Large datasets will be important in translational studies that will influence human health by identifying markers that are predictive of disease (Kim Caple, comment #57). Policies to integrate collection of specimens for genetic and genomic susceptibility testing into ongoing studies evaluating epidemiologic risk factors could also minimize costs (Infectious Diseases Human Genomics Working Group CDC, comment #52).

### 9. Focus on chronic diseases, and, in particular, HBOC and Lynch syndrome

The Healthy People 2020 goals include actions aiming to reduce morbidity and mortality due to hereditary breast/ovarian cancer syndrome and Lynch syndrome, and with existing federal buy-in, these could be priorities among stakeholders. Additionally, many states have received support for colorectal cancer screening and cancer screening in general (Public Health Practice, informal discussion group). "Lynch syndrome, a condition with high heritability, is a noteworthy condition for public health genomics [because of the life-saving potential that Lynch syndrome screening has for at-risk individuals]" (Linda Bruzzone, comment #26).

### 10. Expand state-level utilization of genomics programs

Given the growth of genetic technologies and increased focus on complex gene disorders, state health departments need funds for genomics specialists to participate in disease prevention and control programs. If health departments do not

stay on top of fast-changing genomic technologies, public health will fall behind (Kimberly Kaphingst, key informant interview).



"A very important outcome should be that individuals at all levels of public health in practice, academia and research will either have the genomics knowledge needed for their area of public health or be able to identify and collaborate with a person who does have the knowledge to appropriately use genomic knowledge in relevant aspects of his/her practice" (Sylvia Au, comment #60). State health departments may achieve this by hiring full-time state genetics coordinators to cover public health genetics programs beyond newborn screening; currently, newborn screening coordinators serve as both (Joyce Hooker, comment #11).

# Program Highlight

## Collaboration and the Hawaii Department of Health



**SYLVIA AU**, STATE GENETICS COORDINATOR, HAWAII DEPARTMENT OF HEALTH; STAKEHOLDER CONSULTATION PLANNING COMMITTEE



I made the leap from clinical practice as a genetic counselor to public health genetics as a state genetics coordinator in the Department of Health almost two decades ago. From the beginning, the foundation for the state genetics program's success has been the partnerships and friendships that have been built over the many years. The program has grown tremendously in the years since I was working as the only person in the program. We now have four full-time genetic counselors, assorted part-time and consultant staff, and technical and clerical personnel. In addition, we have robust newborn screening and birth defects programs. The staff works hard to cultivate relationships within and outside the health department. We provide education, technical assistance and consultation to a broad range of public and private

sector programs, organizations, educational institutions and community groups. In addition to the in-state activities, we have also developed partnerships with other states including heading the Western States Genetic Services Collaborative. Our secret to sustainability and growth is to build on our relationships. Our partners value our expertise and ability to “play nicely with others” so they want to include our program in their activities. In turn, we are able to reach out to our partners to help with our genetics program activities. Given that many state genetics programs have disappeared over the last decade, I firmly believe that partnerships and collaborations are the only solutions to sustainability that will drive the use of genomic information and technologies to improve the public's health.

## VI. TABLE OF RECOMMENDATIONS UTILIZING THE *PUBLIC HEALTH IN AMERICA* FRAMEWORK

Essential Public Health Services	Action Steps	Supporting Quotations	Key Actors
<b>Assessment</b>			
Monitor health status to identify and solve community health problems.	<ul style="list-style-type: none"> <li>• Use databases to monitor health of the population</li> <li>• Study gene-environment interactions</li> </ul>	<ul style="list-style-type: none"> <li>• “Understanding the population prevalence of the thousands of genetic variants in different population subgroups and geographic locations and their associations with health and disease is crucial for planning screening programs and guiding future research” (CDC Strategic Plan, including 2007 Vision Statement).</li> <li>• “Make available data warehouses with relevant clinical and transactional information, including genetic contributions to health and outcomes, from health systems around the country to simplify and enable translation research” (Maki Moussavi, comment #28).</li> <li>• “Collecting information on the environmental component to genomics, with consideration of differing levels of complexity and genetic attribution to common/chronic disease” (Michael Watson, comment #31).</li> <li>• There is a need to integrate genetics with epigenetics, and understanding of variations in gene frequencies being the first part. We need to recognize the importance of all determinants (genetics, social behaviors, environment, etc.) because it’s the interaction that will be crucial to examine (Kenneth Olden, key informant interview).</li> <li>• Continue to monitor detection and the number of individuals who are being diagnosed during the early phases of the disease (R4, Genetic Alliance, key informant interview).</li> </ul>	<ul style="list-style-type: none"> <li>• Academe</li> <li>• Government</li> <li>• Practice</li> </ul>
Diagnose and investigate health problems and health hazards in the community.	<ul style="list-style-type: none"> <li>• Utilize family history to identify at-risk individuals</li> <li>• Integrate electronic health records to improve coordination of care</li> </ul>	<ul style="list-style-type: none"> <li>• “Increased emphasis on family history could yield...better identification of those at risk, which has the potential to lead to earlier implementation of surveillance and preventive measures” (Diane J. Allingham-Hawkins, comment #14).</li> <li>• “I truly believe family history could be used to achieve and sustain behavior changes that are plaguing our nation, like obesity, exercise, catching cancer at early, more treatable stages, prevention or delay of diabetes or heart disease, given focus, support, and funding from CDC for public health organizations to figure out how to use this with social marketing principles” (Jenny Johnson, comment #13).</li> <li>• “Leverage electronic healthcare infrastructure to achieve several goals: outcomes research, quality improvement, decision support” (Maki Moussavi, comment #28).</li> <li>• “We suggest that CDC focus its efforts on applying genomics to common disease, supporting the generation of evidence that demonstrates the utility of genomic-based screening and interventions, and delivery models that will test broad application of such technologies” (James Madara, comment #46).</li> </ul>	<ul style="list-style-type: none"> <li>• Government</li> <li>• Practice</li> </ul>

Essential Public Health Services	Action Steps	Supporting Quotations	Key Actors
<b>Assessment</b>			
		<ul style="list-style-type: none"> <li>• "...understanding population genetic variation may contribute to a better understanding of population health disparities, especially among racial and ethnically defined populations. Conversely, failing to incorporate knowledge of the environmental and social determinants of health in population genetics research may result in inappropriate conclusions about genetic contributions to health disparities" (Wylie Burke, comment #56).</li> </ul>	
<b>Policy Development</b>			
<p>Inform, educate, and empower people about health issues.</p>	<ul style="list-style-type: none"> <li>• Improve genomic literacy of the public</li> <li>• Develop high school curricula for genomics</li> <li>• Use social marketing to teach the community about genomics in understandable language</li> </ul>	<ul style="list-style-type: none"> <li>• "...the most important aspect of genomics [is] first improving the health literacy of the public (to increase knowledge and demand for this essential information and concomitantly improving the health literacy of health professionals in order that they are prepared to answer questions and efficiently and effectively utilize this rapidly emerging and evolving science" (Richard Carmona, email #1).</li> <li>• "Genomics especially needs to be translated into understandable terms for the general public, with an emphasis on general issues of concern such as poverty, racism, and the appearance of disease" (Terri Combs-Orme, comment #3).</li> <li>• "...increasing integration of genetics education in high school biology curricula, especially focusing on the concepts of carrier screening and risk assessment for disease" (Tricia Page, comment #21).</li> <li>• As a long term approach to improving the genetic and genomic literacy of the general public, strong arguments can be made for the need to revamp K-12 education from the perspectives of both science and health to incorporate genomic approaches to common health issues (Kristi Zonno, comment #39).</li> <li>• Educate faith-based organizations so they can disseminate knowledge to communities. Visual tools must be developed and shared with community leaders/gatekeepers. These tools must be in genomics 101 language (NCC SPIG, informal discussion group).</li> </ul>	<ul style="list-style-type: none"> <li>• Academia</li> <li>• Community</li> <li>• Government</li> <li>• Practice</li> </ul>
<p>Mobilize community partnerships and action to identify and solve health problems.</p>	<ul style="list-style-type: none"> <li>• Community engagement</li> </ul>	<ul style="list-style-type: none"> <li>• "State health departments are important actors in the research translation network in part because they set policy for their states, but also because they provide care to medically underserved populations" (Laura Senier, comment #16).</li> </ul>	<ul style="list-style-type: none"> <li>• Community</li> <li>• Government</li> <li>• Practice</li> </ul>

Essential Public Health Services	Action Steps	Supporting Quotations	Key Actors
<b>Policy Development</b>			
<p>Mobilize community partnerships and action to identify and solve health problems.</p>	<ul style="list-style-type: none"> <li>• Community engagement</li> </ul>	<ul style="list-style-type: none"> <li>• “Engage community clinics and community-based organizations to get minority community buy-in into programs. Barriers to overcome: community mistrust, lack of sustained funding, difficulty working in the community” (Steven Galen, comment #19).</li> <li>• “Stakeholders from all levels...including community organizations, local public health, providers and health professional organizations, health plans, hospitals, private research organizations-provider and public education [to] translate into practice” (Jean Chabut, comment #50).</li> <li>• “Education and outreach to American Indian and Alaska Native communities about genomics research and its public health implications are also critical. There is a great deal of suspicion and misinformation about genetics research in Indian Country” (Jacqueline Johnson Pata, comment #30).</li> <li>• Community partners, prevention practitioners from state health departments, genetic counselors, and communications researchers need to be at the table to push community based participatory research agendas forward. (Kim Kaphingst, key informant interview).</li> <li>• Develop programs that will allow individuals to store their own health data and control who has access to it. (R2, Genetic Alliance, key informant interview).</li> </ul>	<ul style="list-style-type: none"> <li>• Community</li> <li>• Government</li> <li>• Practice</li> </ul>
<p>Develop policies and plans that support individual and community health efforts</p>	<ul style="list-style-type: none"> <li>• Implement policies to:</li> <li>• Promote accessibility of genomic technology</li> <li>• Focus on community education and the use of family history</li> </ul>	<ul style="list-style-type: none"> <li>• “Implement policy at all levels (legislation, programmatic, etc.) to appropriately use family history and other genetic based recommendations that may come in the future” (Jenny Johnson, comment #13).</li> <li>• “By using both public values and expert knowledge in an intentional, collaborative, open, and transparent environment, genomic scientists, the public, and policy makers can reasonable hope to create effective genomics policy in the future” (Gregory Fowler, comment #38).</li> <li>• Community engagement efforts in collaboration with academics and public health practitioners are necessary to bridge communication gaps and further genomics educations (Kim Kaphingst, key informant interview).</li> <li>• “What [the IOM, AMA, and Healthy People 2010] policies do not address is the need for ensuring that the public has access to resources that provide for an understanding of genetics programs and services or the need for health care providers who can successfully communicate risk assessment and other genomic information to patients, two crucial components of genetic and genomic literacy” (Kristi Zonno, comment #39).</li> </ul>	<ul style="list-style-type: none"> <li>• Community</li> <li>• Government</li> <li>• Practice</li> </ul>

Essential Public Health Services	Action Steps	Supporting Quotations	Key Actors
<b>Policy Development</b>			
<p>Enforce laws and regulations that protect health and ensure safety.</p>	<p>Implement:</p> <ul style="list-style-type: none"> <li>• Regulatory policies and guidelines for genomic applications</li> <li>• Insurance coverage for high risk individuals</li> </ul>	<ul style="list-style-type: none"> <li>• Groups such as EGAPP are needed to make recommendations and develop guidelines to be integrated in public health practice. Also, direct-to-consumer genetic tests must be regulated to ensure that consumers are getting accurate information about their genetic susceptibilities (Kim Kaphingst, key informant interview).</li> <li>• “Policies are needed that create and apply tools to evaluate appropriate application of genomics in healthcare in order to increase utility, ensure effective follow-up, and improve health outcomes” (Michael Watson, comment #31).</li> <li>• “Policies to improve health insurance coverage and other support services to help high risk people” (Jean Chabut, comment #50).</li> <li>• “Monitor and recommend public policy to safeguard the public from detrimental use of genomic information” (Cornelia Van Duijn, comment #36).</li> <li>• Establish policies for dealing with technologies that do not meet the current evidentiary thresholds and strategies for incremental evidence development tied to reimbursement (R6, Genetic Alliance, key informant interview).</li> </ul>	<ul style="list-style-type: none"> <li>• Government</li> <li>• Practice</li> </ul>
<b>Assurance</b>			
<p>Link people to needed personal health services and assure the provision of health care when otherwise unavailable.</p>	<ul style="list-style-type: none"> <li>• Ensure accessibility to genomic applications and services</li> </ul>	<ul style="list-style-type: none"> <li>• “More genetic counselors and genetic professionals need to be trained” (Jill Hagenkord, comment #15).</li> <li>• “Federal and state agencies should work with professional organizations, payers, test developers, the pharmaceutical industry, patients, and consumer advocacy groups [to] ensure access, low costs, and benefits. Barriers must be overcome to share resources and information among groups with different interests (Gurvaneet Randhawa, email #2).</li> <li>• “Need to engage community clinics and community-based organizations to get minority community buy-in” (Steven Galen, comment #19).</li> <li>• “Policies that foster a learning healthcare system (translational medicine) to include point-of-care education and cross-provider education will improve health outcomes and mitigate disparities by addressing access and the maldistribution of genetics providers/services” (Michael Watson, comment #31).</li> <li>• “Call on public health and health care service agencies and related organizations to ensure access to culturally competent, accurate, and complete genetic and genomic information and resources for conditions affecting our diverse populations” (Kristi Zonno, comment #39).</li> </ul>	<ul style="list-style-type: none"> <li>• Community</li> <li>• Government</li> <li>• Practice</li> </ul>

Essential Public Health Services	Action Steps	Supporting Quotations	Key Actors
<b>Assurance</b>			
<p>Assure competent public and personal health care workforce.</p>	<ul style="list-style-type: none"> <li>• Incorporate genomics into the curricula of medical schools, nursing schools, and schools of public health</li> <li>• Provide opportunities for continuing education around genomics</li> </ul>	<ul style="list-style-type: none"> <li>• There needs to be “a greater understanding by public health professionals of what genome-based knowledge can bring to public health practice” (Ron Zimmern, email #3).</li> <li>• “Encourage collaborations between schools of public health and public health programs, government agencies, professional and community-based organizations, and genetics advocacy groups to disseminate genetic information on topics relevant to community members, and use public input on ways to enhance genetic and genomics literacy at the community level (Kristi Zonno, comment #39).</li> <li>• Curricula must be built to ensure genomic competencies for the public health workforce (Jean Chabut, key informant interview).</li> <li>• “Genetics/genomics needs to be integrated into every Public Health program/study and training. Genetics/genomics needs to be a part of the Public Health culture; Public Health professionals need to think genetically” (Elizabeth Balkite, comment #6).</li> <li>• “Fund the development and implementation of accessible educational programs and continuing education in genetics and genomics for the public health workforce” (SACGHS Report on Genetics Education and Training).</li> <li>• “Professional organizations, such as ACMG, NSGC, ASCO, AMA, etc. must provide educational support and knowledge resources to improve the understanding of the impact of genomics on the specialty areas of these organizations to prime providers in those areas to quickly adapt to changes needed to integrate genomics into practice” (Maki Moussavi, comment #28).</li> <li>• Funding would provide an effective education format that allows the nurse to: 1) perform family history assessment; construct a three-generation pedigree; identify family history red flags indicative of an inherited predisposition; and refer at risk individuals to genetic services as indicated; 2) utilize appropriately genomic information and technology in the delivery of healthcare; 3) provide an evaluation of the educational program(s) for efficacy that includes: knowledge and skill measures, as well as measuring the amount of practice application (Laurie Badzek, mail #1).</li> <li>• Ensure health care providers have the education and training needed to interpret the results of complex genomic tests (R5, Genetic Alliance, key informant interview).</li> </ul>	<p>Academia</p> <ul style="list-style-type: none"> <li>• Government</li> <li>• Practice</li> </ul>
<p>Evaluate effectiveness, accessibility, and quality of personal and population-based health services.</p>	<ul style="list-style-type: none"> <li>• Evaluate genomic tests to ensure efficacy, safety, and ethicalness</li> <li>• Continue efforts of evaluation groups</li> </ul>	<ul style="list-style-type: none"> <li>• “Promote and fund research into the clinical utility of genetic and genomic tests. The vast majority of genetic and genomic tests are made available in the absence of any evidence of such tests improves health outcomes...unproven tests [should be] removed from the marketplace” (Diane J. Allingham-Hawkins, comment #14).</li> </ul>	<ul style="list-style-type: none"> <li>• Academia</li> <li>• Government</li> <li>• Practice</li> </ul>

Essential Public Health Services	Action Steps	Supporting Quotations	Key Actors
<b>Assurance</b> (continued)			
Evaluate effectiveness, accessibility, and quality of personal and population-based health services.	<ul style="list-style-type: none"> <li>Evaluate genomic tests to ensure efficacy, safety, and ethicalness</li> <li>Continue efforts of evaluation groups</li> </ul>	<ul style="list-style-type: none"> <li>"[A priority should be] continuation of a group such as EGAPP to provide evidence-based, authoritative recommendations on which genetics tests are useful in improving outcomes" (Herbert F. Young, comment #41).</li> <li>"Research to provide evidence that segmentation of populations improves the effectiveness and efficiency of public health interventions, especially in the fields of obesity, diabetes, heart disease, stroke, cancer, and neurodegenerative disorders" (Ron Zimmern, email #3).</li> <li>"Engage health care organizations to study true patient and economic benefits of genomic testing-start with low hanging fruit of valid markers that need pragmatic trial testing" (Brian, comment #5).</li> <li>Use health economics to understand how much money improved diagnostics can save by preventing ineffective treatments from being administered (R8, Genetic Alliance, key informant interview).</li> </ul>	<ul style="list-style-type: none"> <li>Academia</li> <li>Government</li> <li>Practice</li> </ul>
<b>System Management</b>			
System Management	<ul style="list-style-type: none"> <li>Coordination between all sectors of the public health system</li> <li>Capitalize on the social science aspect of public health and the hard science aspect of genomics</li> </ul>	<ul style="list-style-type: none"> <li>"Improved coordination of related and overlapping federal and non-federal activities in health information technology, outcomes research, validation of gene-based tests, regulation and payment of gene-based tests" (Gurvaneet Randhawa, email #2).</li> <li>"Public and private funding should be made available to implement and expand public health genomics activities at the state and local levels. As part of implementation or expansion of successful public health genomics activities, the emphasis on transitioning from temporary grant funding to sustainable activities needs to be a priority and even a requirement to receive the funding" (Sylvia Au, comment #60).</li> <li>"Explicitly bringing together social and biological models of disease and showing that both are needed for optimal public health outcomes" (Ron Zimmern, email #3).</li> <li>"...I think that the vision being developed by more strongly in the environmental health sciences—in which genomics is used to inform public policies which aim to protect the health of populations- deserves great consideration in the context of public health genomics, especially insofar as reducing health disparities is a central goal" (Sara Shostak, comment# 51).</li> <li>"The stakeholders in genomic research and clinical translation are well known. However, each tends to work independently to meet their own missions. Incentives for coordination and leadership committed to sharing information and resources would go a long way in providing a more holistic approach to ensure we capitalize on the investment and advances already made in genomic science" (Joan Scott, comment #47).</li> </ul>	<ul style="list-style-type: none"> <li>Academia</li> <li>Community</li> <li>Government</li> <li>Practice</li> </ul>

Essential Public Health Services	Action Steps	Supporting Quotations	Key Actors
<b>System Management</b>			
System Management	<ul style="list-style-type: none"> <li>• Coordination between all sectors of the public health system</li> <li>• Capitalize on the social science aspect of public health and the hard science aspect of genomics</li> </ul>	<ul style="list-style-type: none"> <li>• “Filling the ‘gap’ between what is currently being extensively done (evidence reviews, meta-analyses) and areas needing more attention (translation to end users—physicians, public health practitioners, policy makers, and consumers) was viewed as a major priority” (The Genomic Applications in Practice and Prevention Network [GAPPNet]).</li> </ul>	<ul style="list-style-type: none"> <li>• Academia</li> <li>• Community</li> <li>• Government</li> <li>• Practice</li> </ul>
<b>Research</b>			
Research for new insights and innovative solutions to health problems	<ul style="list-style-type: none"> <li>• Fund research</li> <li>• Focus efforts on translational research</li> <li>• Develop transdisciplinary research agendas</li> <li>• Engage community members</li> </ul>	<ul style="list-style-type: none"> <li>• There is a need for research incentives to facilitate [transdisciplinary] partnerships and for grants to include community participation (Kenneth Olden, key informant interview).</li> <li>• We need to rethink our current method of selecting pilot projects—rather than rewarding the best grant writers, we should be selecting pilot sites that present the best opportunity for integration (R2, Genetic Alliance, key informant interview).</li> <li>• “...two areas that are critically important to achieving the goal of translating genome-based science and technology into improvements in population health: 1) The upstream, two-directional communication and engagement with the public, scientific experts, and other relevant stakeholders; and, armed with those tools; 2) Shaping well-reasoned public policy” (Gregory Fowler, comment #38).</li> <li>• Currently, the most fundamental part of genomics, translation, is underfunded. There continues to be very robust funding in the discovery phase of the research cycle. Very little research money goes towards delivery and outcomes (Wylie Burke, key informant interview).</li> <li>• Funding is one barrier to furthering the integration of genomics into public health priorities, as it is difficult for academics to find public health genomics research grants. There is a tension between the public health folks and those in clinical medicine as to where the research dollars belong (Kim Kaphingst, key informant interview).</li> <li>• Collaborations and partnerships must be made with academics and the community to foster trust. This is a win-win for researchers and community members, as the researcher can conduct a study and community members learn about the research topic (Chikezie Maduka, key informant interview).</li> </ul>	<ul style="list-style-type: none"> <li>• Academia</li> <li>• Community</li> <li>• Government</li> <li>• Practice</li> </ul>

## VII. CONCLUSION

The stakeholder consultation involved input from more than 100 individuals representing the many sectors of public health academe, practice and community. Their insights, which formed the foundation of this report, will help guide leaders in public health genomics in prioritizing actions over the next five years. This report aims to encourage all engaged in public health genomics to learn from each other, find ways to share an understanding of the field with those who still need

to be engaged, and develop the collaborations necessary to realize the maximum potential of the field for achieving public health goals. CDC, having initiated this stakeholder consultation process, can utilize the results to continue to provide effective leadership for the field. This consultation demonstrated that the human capital available to provide support is substantial and already engaged. Significant work is needed to further education, research, assurance, policy development,

and health applications. By maintaining current partnerships while developing new and innovative collaborations, public health genomics promises to positively impact public health and healthcare. This report can assist both in the prioritization of actions over the next five years as well as in the identification of the many individuals and organizations whose combined efforts can address these actions and achieve the desired results to improve health in the United States.



# **PART TWO**

## **PRIORITIES CONFERENCE REPORT**





## I. INTRODUCTION

It has been more than a decade since the Human Genome Project was completed. Recently, the National Human Genome Research Institute (NHGRI) released a new strategic plan, “Charting a course for genomic medicine from base pairs to bedside,” to accelerate the translation of genomic discoveries into clinical applications. The plan covers five domains (genome structure, genome biology, disease biology, medical science, and effective healthcare) and shows that, until now, most accomplishments in genomics have occurred in the first two domains, while in the next ten years, progress is expected mostly in the second and third domains. The plan predicts that beyond 2020 we should expect to see major accomplishments in medical science and healthcare. Thus, although the potential for genomics to impact population health remains strong, the immediate implications are less clear.

Since 1997, the CDC Office of Public Health Genomics (OPHG) has worked to develop the public health genomics enterprise, engaging many partners to anticipate, effect, and evaluate the translation of genome discoveries into products and practices that impact public health. From the beginning, this strategy has comprised two complementary approaches: bringing a population perspective to genomic research, and translating genomic research findings for public health benefit. During the last decade, epidemiologic studies and methodology have become more prominent in genomic research, and the demonstration of an evidence-based approach to evaluating genetic tests has helped establish a societal perspective on their rational integration into healthcare and disease prevention.

With a plan in place to accelerate clinical applications of genomic discoveries comes a charge to the public health community: identify opportunities and challenges for using genomics to impact population health. This includes addressing important public health issues and improving the effectiveness and efficiency of public health programs. These developments call for stronger and novel partnerships among stakeholders across and beyond the public health arena.

The OPHG hosted a daylong meeting to help reevaluate and prioritize near- and longer-term objectives in public health genomics. At this meeting, the OPHG engaged stakeholders from federal, state, academic, industry, consumer, and professional organizations in a facilitated discussion of how the changing environment will affect priorities, goals, and strategies for public health genomics in the next five years and how enhanced partnership can help to advance the field. The product of this meeting is a list of short-term actions and coordinated approaches by various stakeholders to guide the use of genomics to improve population health outcomes. In the following report we summarize the proceedings, including the rationale for the methodology employed to engage stakeholders in action-oriented discussions. We also describe the priority outcomes and action items that were identified by participants during the course of the day’s discussion.

## II. METHODOLOGY

### Morning Introductions

Although extensive background material was provided to attendees in advance of the meeting, the first two hours of the meeting were allocated for contextual presentations. These presentations ensured that all participants were equipped with the necessary information and framework for the subsequent breakout group discussions devoted to four topic areas. The morning introductions included presentations from CDC's OPHG, NIH's National Human Genome Research Institute, and HRSA's Genetic Services Branch as well as two content presentations on the stakeholder consultation process conducted by the Center for Public Health and Community Genomics at University of Michigan and Genetic Alliance. Ms. Sharon Terry ended the morning session with a charge for the topic area teams. A more thorough description of the background materials provided to participants as well as the highlights from the morning talks can be found in Section III, Setting the Stage.

### Breakout Sessions

The goal of this meeting was to identify near- and long-term priority outcomes for the field of public health genomics and to develop and prioritize the concrete, actionable activities needed to achieve these outcomes. Accordingly, the majority of the meeting time was allocated to breakout groups, each consisting of approximately 20 participants, organized around the following four topic areas: Prevention, Detection, Development & Evaluation, and Pathways & Interactions. Participants were pre-assigned to topic areas in order to ensure that each group had representation from stakeholders with a diversity of perspectives



and expertise. The breakout sessions were organized into two two-hour sessions. Each breakout session was assigned two co-facilitators to help energize the group and keep participants focused on the objectives. Co-facilitators were selected on the basis of their content knowledge and leadership skills.

### Morning Breakout Session

During the morning session each breakout group was asked to discuss and reach a general consensus on priority outcomes in their topic area for the next five years. Towards this end, participants were asked to consider the following five questions, which had also been used during the stakeholder consultation processes done in preparation for this meeting:

1. What are the most important activities that should be carried out by the public health system in 2012–2017 to apply

genomic knowledge to public health goals?

2. What outcomes specific to public health might be achieved as a result of carrying out these activities?
3. What policies are needed in order to achieve these outcomes?
4. What institutions, organizations, and agencies need to participate in achieving these outcomes and what role should they play?
5. What barriers are anticipated in achieving these outcomes and how might they be best overcome?

By the end of the morning session each group had developed a small number of higher-level outcomes and a list of potential barriers that could then be used to draft concrete action items in the afternoon session.

### Laying the Foundation for the Future of Public Health Genomics

In order to maximize the number of concrete, actionable items produced through this meeting, we used the lunch break to engage participants in an activity to help lay the foundation for the future of public health genomics. The meeting attendees included many leaders in the field of public health genomics and representation from diverse stakeholder groups. As such, the meeting presented a unique opportunity to begin forging the collaborative efforts needed to advance the field of public health genomics. To capitalize on this opportunity, participants were asked to use lunch and other breaks to network with fellow meeting attendees and identify collaborative projects that will advance public health genomics. Blank “bricks” were prepared, on which participants were asked to write descriptions of their proposed collaborations. The bricks were assembled on a wall so

that participants could view the commitments made by others. Participants were encouraged to identify at least one “brick” or area for collaborative commitment.

### Afternoon Breakout Session

The goal of the afternoon breakout sessions was to produce recommendations for concrete activities needed to achieve the outcomes identified during the morning session. Each group was asked to consider a number of cross cutting issues in their discussion, including education, workforce, community engagement, infrastructure, integration, resources, dissemination, and ethical, legal, and social implications. Breakout groups were instructed to record the identified action-items on notecards along with their funding requirements, stakeholder groups, and milestones. The groups were provided with three different colors of notecards so that the recommended action items could be categorized as low,

medium, or high priority. At the end of the session, each group was asked to transfer their action items to a large five-year timeline hanging in the main meeting area.

### Reporting

Following the afternoon breakout sessions, the co-facilitators were asked to give a 10-minute presentation summarizing the outcomes and high-priority action items that their group identified. To expedite the presentations, co-facilitators were provided with a PowerPoint template to report on the outcomes and action items, including the associated funding needs, stakeholder needs, time frame, and metrics. A summary of the outcomes and high-priority action items identified by each of the four breakout groups can be found in Section IV, “Breakout Groups.”

## III. SETTING THE STAGE

### Background Materials

Two weeks prior to the meeting, participants were provided with access to a shared drive containing the background material for the meeting. The background material included perspective pieces and white papers from the OPHG, strategic

plans from related agencies such as the NHGRI, summaries from related meetings and workshops, and relevant news articles. A full listing of the material that was provided is shown in Table I. Our purpose in providing this material was to help ensure that all participants were

aware of both the history and current state of the field of public health genomics. This helped to ensure that participants would not rehash topics that have already been extensively discussed elsewhere and to empower participants to cover new ground during the meeting.

**Table I: Meeting Background Material**

Title	Author	Link
10 Years of Public Health Genomics at CDC 1997–2007	Centers for Disease Control and Prevention, National Office of Public Health Genomics	<a href="http://www.cdc.gov/genomics/about/reports/2007/">http://www.cdc.gov/genomics/about/reports/2007/</a>
Genomics 2010 At a Glance: Successes and Opportunities for Population-based Research and Practice	Centers for Disease Control and Prevention, National Office of Public Health Genomics	<a href="http://www.cdc.gov/genomics/about/AAG/index.htm">http://www.cdc.gov/genomics/about/AAG/index.htm</a>
Genome Medicine: Past, Present and Future	Auffray et al.	<a href="http://genomemedicine.com/content/3/1/6">http://genomemedicine.com/content/3/1/6</a>
Current Priorities for Public Health Practice in Addressing the Role of Human Genomics in Improving Population Health	Khoury et al.	<a href="http://www.ncbi.nlm.nih.gov/pubmed/21406285">http://www.ncbi.nlm.nih.gov/pubmed/21406285</a>
Future Health Applications of Genomics: Priorities for Communication, Behavioral, and Social Sciences Research	McBride et al.	<a href="http://www.ncbi.nlm.nih.gov/pubmed/20409503">http://www.ncbi.nlm.nih.gov/pubmed/20409503</a>
OPHG Strategic Plan 2010–2015	Office of Population Health Genomics	<a href="http://www.genomics.health.wa.gov.au/wagc/index.cfm">http://www.genomics.health.wa.gov.au/wagc/index.cfm</a>
Challenges of Translating Genetic Tests into Clinical and Public Health Practice	Rogowski et al.	<a href="http://www.ncbi.nlm.nih.gov/pubmed/19506575">http://www.ncbi.nlm.nih.gov/pubmed/19506575</a>
Genome-based knowledge and public health: the vision of tomorrow and the challenge of today	TS Baumen	<a href="http://www.ncbi.nlm.nih.gov/pubmed/21247867">http://www.ncbi.nlm.nih.gov/pubmed/21247867</a>

**Table I: Meeting Background Material**

Title	Author	Link
The Emergence of Translational Epidemiology: From Scientific Discovery to Population Health Impact	Khoury et al.	<a href="http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2927741/?tool=pubmed">http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2927741/?tool=pubmed</a>
Genomics and Public Health in the United States: Signposts on the Translation Highway	Gwinn et al.	<a href="http://www.ncbi.nlm.nih.gov/pubmed/16490955">http://www.ncbi.nlm.nih.gov/pubmed/16490955</a>
Invited Commentary: Genes, Environment, and Hybrid Vigor	Gwinn et al.	<a href="http://www.ncbi.nlm.nih.gov/pubmed/19671836">http://www.ncbi.nlm.nih.gov/pubmed/19671836</a>
What Role for Public Health in Genetics and Vice Versa?	Khoury et al.	<a href="http://www.ncbi.nlm.nih.gov/pubmed/17003539">http://www.ncbi.nlm.nih.gov/pubmed/17003539</a>
The Future of the Public's Health in the 21st Century	Committee on Assuring the Health of the Public in the 21st Century	<a href="http://www.iom.edu/Reports/2002/The-Future-of-the-Publics-Health-in-the-21st-Century.aspx">http://www.iom.edu/Reports/2002/The-Future-of-the-Publics-Health-in-the-21st-Century.aspx</a>
Charting a Course for Genomic Medicine From Base Pairs to Bedside	Green et al.	<a href="http://www.ncbi.nlm.nih.gov/pubmed/21307933">http://www.ncbi.nlm.nih.gov/pubmed/21307933</a>
Framework for Public Health Genetics Policies and Practices in State and Local Public Health Agencies	ASTHO	<a href="http://www.ct.gov/dph/LIB/dph/Genomics/astho.pdf">www.ct.gov/dph/LIB/dph/Genomics/astho.pdf</a>
CDC Wants Near-term Public Health Genomics Plans	M Jones	<a href="http://www.genomeweb.com/cdc-wants-near-term-public-health-genomics-plans">http://www.genomeweb.com/cdc-wants-near-term-public-health-genomics-plans</a>
Massive Project to Study the Link between Genetics and Health	E Singer	<a href="http://www.technologyreview.com/biomedicine/38133/">http://www.technologyreview.com/biomedicine/38133/</a>
National Prevention Strategy: America's Plan for Better Health and Wellness	National Prevention, Health Promotion and Public Health Council	<a href="http://www.healthcare.gov/prevention/nphpphc/strategy/report.pdf">www.healthcare.gov/prevention/nphpphc/strategy/report.pdf</a>
NHGRI to Dive into Ethical, Legal, Social Issues	M Jones	<a href="http://www.genomeweb.com/nhgri-dive-ethical-legal-social-issues">http://www.genomeweb.com/nhgri-dive-ethical-legal-social-issues</a>

## Introductory Presentations

Several speakers presented briefly at the start of the meeting to provide the context for the day. Dr. Muin Khoury, Director of the Office of Public Health Genomics, CDC, began the day with an overview of public health and a brief history of the integration of genomics into public health programs at the CDC. Dr. Khoury emphasized the need to think about two forces: a push to integrate genomics into medicine and public health and a pull to include key stakeholders in the decisions around when and how the integration should take place. Dr. Khoury ended his talk with a charge to participants to go beyond a generic research agenda to develop collaborative, measurable recommendations with near-term population health impact.

Following Dr. Khoury, Dr. Eric Green, Director of the National Human Genome Research Institute, NIH, summarized the near- and long-term potential for genomics to revolutionize the practice of medicine. Dr. Green outlined some of the technological advancements likely to impact the integration of genomics into medicine, such as the decreasing cost of sequencing, while pointing out that technological advances are milestones, not destinations. Dr. Green ended his talk by emphasizing that the NIH is only one piece of a much larger puzzle, including

many international efforts to advance genomic medicine.

Next, Dr. Sara Copeland, Acting Chief of the Genetics Services Branch, Maternal and Child Health Bureau, HRSA, described HRSA's mission around integrating genetics into healthcare and some of the challenges and barriers they face. Dr. Copeland discussed HRSA's priorities, including making healthcare safer by reducing harm caused in the delivery of care, promoting effective communication and coordination of care, and making quality healthcare affordable. In addition, she highlighted the need to ensure that genetics is a part of both the President's and Secretary's healthcare priorities. Following Dr. Copeland's presentation, Ms. Sharon Terry, President and CEO of Genetic Alliance, briefly reviewed the design of the day and charged participants to look beyond their own work and consider the field of public health genomics as a whole. Ms. Terry emphasized the need to think practically throughout the entire day.

Finally, Mr. Toby Citrin, Director of the Center for Public Health and Community Genomics at the University of Michigan, and Mr. James O'Leary, Chief Innovation Officer at Genetic Alliance presented results from the stakeholder engagement process that occurred in

preparation for the meeting. Mr. Citrin reported on the responses received to the Request for Information that the CDC published in the Federal Register on June 21, 2011 and on the results of stakeholder interviews, focus groups, and literature reviews. Mr. Citrin's team at the University of Michigan focused on understanding the perspectives of stakeholders in academia and the public health sector, while Mr. O'Leary's team at Genetic Alliance focused on gathering the nonprofit sector and private industry perspectives. The results of this extensive stakeholder engagement process emphasized distinct but complementary issues. From the academic and public health community, emphasis was placed on issues like the importance of workforce education, funding to support the integration of genomics into public health, the need for community engagement to reduce health disparities, translational research to impact health outcomes, and the need to leverage the electronic healthcare infrastructure to develop the evidence base. Similarly, members of the nonprofit community and for-profit sector highlighted the need for improving the evidence base and advancing translational research. They also noted that a key priority for advancing translational research is the creation of open-access databases that will enable widespread sharing of genetic datasets.

## IV. BREAKOUT GROUPS

### DETECTION GROUP

#### Introduction:

The Detection group focused on the outcome, “detect diseases early and intervene effectively.” The morning discussion largely focused on the balance of roles between clinicians and traditional public health stakeholders in providing information, education, and services. With a large number of tests already on the market and whole genome sequencing on the horizon, the group felt that a flexible, dynamic public health system was needed to bridge the gap between successful, established screening programs (such as newborn screening) and the scattered application of other testing and screening. In addition, it was clear that the application of family health history remains a high priority for the field.

Overall, participants felt that the biggest shift in priorities for the field centered on the need to use data effectively in the areas of electronic health records, decision support, and tracking. Group members felt that it is the responsibility of the public health system to ensure that patients and providers have access to information on genetic tests and services and to ensure that evidence of effectiveness (or lack thereof) is available and understandable. In addition, a clear focus on data infrastructure and policies emerged, with a specific concern that genetics and genomics issues are not being taken into account. Finally, in many ways, the group saw the public health genomics community as the ideal group of stakeholders to inform and drive high-priority changes in areas such as the use of patient data and consent.

#### Outcomes:

The working group objective, detect diseases early and intervene effectively, led to the identification of a number of important outcomes, including: (1) consistent health messaging, (2) the creation of resources and tools that offer genetic testing decision support for patients and providers, (3) the inclusion of public health genomics needs in national data standards and health IT priorities, (4)

the development of an active surveillance system at the intersection of clinical care and public health, (5) expansion of public health screening programs to include cascade and life course screening, (6) better identification and quantification of high risk populations, and (7) identification of gaps in available testing and screening at the services level and transfer that information to the research community.

#### Priority Action Items:

##### OUTCOME 1: Consistent health messaging

- I. Activities:
  - a. Develop a small number of specific health messages for public health genomics.
    - i. Review relevant literature around health communications.
    - ii. Hold a planning meeting to determine the topics for messaging (e.g. counseling, family history), assess the capabilities of states to launch pilot programs, assess the audience, and assess platforms for effective dissemination.
    - iii. Pilot test messaging in states using multiple communications platforms.
- II. Funding:
  - a. Approximately \$50,000 per state plus the cost of meeting planning
- III. Stakeholder Needs:
  - a. State and local health departments
  - b. Providers
  - c. Alternative medicine
  - d. Academia
  - e. Communities and disease-specific organizations
  - f. Laboratories
  - g. Health plans
  - h. Government
- IV. Timeline:
  - a. 1–3 years
- V. Metrics:
  - a. The number of web hits and phone calls

**OUTCOME 2: Create resources and tools that offer genetic testing decision support for patients and providers**

- I. Activities:
  - a. Create a companion guide to the Genetic Testing Registry.
    - i. Convene a development group.
    - ii. Establish a rubric to help providers “trust or trash” specific genetic tests using available information.
  - b. Create summaries of recommended actions, similar to ACMG Act Sheets, for genetic tests that have been reviewed by Evaluation of Genomic Applications in Practice and Prevention (EGAPP) or similar bodies.
    - i. Identify organizations or associations to lead the process.
    - ii. Convene a volunteer group of professionals to develop sheets.
    - iii. Develop action sheets on a rolling basis. Begin with sheets for professionals and develop follow-up lay versions of each sheet.
- II. Funding:
  - a. \$50,000 for a companion guide or \$500,000 a year for development of supplemental analysis
  - b. \$10,000 per sheet
- III. Stakeholder Needs:
  - a. State and local health departments
  - b. Providers
  - c. Health communication specialists
  - d. Community-based and disease-specific organizations
  - e. Laboratories
  - f. Health plans
  - g. Government agencies
- IV. Timeline:
  - a. 6 months to convene a group
  - b. 18 months to develop guide
- V. Metrics:
  - a. The percentage of tests in the Genetic Testing Registry that are included in the guide
  - b. The number of times guides are accessed

**OUTCOME 3: Inclusion of public health genomics needs in national data standards and health IT priorities**

- I. Activities:
  - a. Coordinate public health response to changes in the Common Rule.
    - i. Submit a response to the request for comments.
    - ii. Recruit public health stakeholders to be involved in national committees.
    - iii. Solicit feedback from the public health community.
  - b. Create a strategy for engaging identified groups.
    - i. List volunteer individuals with the requisite knowledge and skills.
    - ii. Match expertise to identified organizations.
  - c. Raise awareness of public health genomics issues through knowledge dissemination.
    - i. This will be achieved through engagement, such as through advisory capacities or inclusion of individuals in organizations.
- II. Funding:
  - a. None initially
- III. Stakeholder Needs:
  - a. Academia
  - b. State and local health departments
  - c. Providers
  - d. Health communications specialists
  - e. Community-based and disease-specific organizations
  - f. Laboratories
  - g. Health plans
  - h. Government agencies
- IV. Timeline:
  - a. Approximately one year for the Common Rule
- V. Metrics:
  - a. Changes to the Common Rule based on our response

**OUTCOME 4: Development of an active surveillance system at the intersection of clinical care and public health**

- I. Activities:
  - a. Integrate genetics/genomics fields into national health information exchange (HIE) architecture.
    - i. Assess current state-based HIE projects
    - ii. Convene a planning group
    - iii. Identify states with existing genetics staff, both adult and newborn screening focused, and sufficient HIE experience
    - iv. Conduct pilot projects in a few states
  - b. Partner with the Electronic Medical Records for Genetic Research Consortium (eMERGE) to address issues of consent and data collection from public health systems.
    - i. Reach out to eMERGE
    - ii. Engage stakeholder programs such as PRCs, National Children’s Study, HRSA-funded health centers
    - iii. Expand eMERGE research questions
- II. Funding:
  - a. Minimal cost for convening and information gathering
  - b. \$400,00 per state per year for the HIE pilot projects
  - c. Minimal costs for engagement
  - d. \$100,000 to expand existing study
- III. Stakeholder Needs:
  - a. State and local health departments
  - b. Chronic disease experts
  - c. HIE personnel
  - d. Academia
  - e. Community-based and disease-specific organizations
  - f. Government agencies
  - g. Additional eMERGE stakeholders
- IV. Timeline:
  - a. 6 months to convene
  - b. 5 years to execute

- V. Metrics:
  - a. The number of HIEs with slots for genomic data
  - b. The percentage of data that makes it into HIE
  - c. eMERGE metrics

**OUTCOME 5: Expansion of public health screening programs to include cascade and life-course screening**

- I. Activities:
  - a. Further the goals of the Genetics for Early Disease Detection and Intervention (GEDDI) project.
    - i. Have the GEDDI working group members review draft publication and recommendations.
    - ii. Disseminate the publication to attendees and public health stakeholders.
- II. Funding:
  - a. None
- III. Stakeholder Needs:
  - a. Academia
  - b. State and local health departments
  - c. Providers
  - d. Community-based and disease-specific organizations
  - e. Laboratories
  - f. Health plans
- IV. Timeline:
  - a. Approximately one year
- V. Metrics:
  - a. The number of reviewers
  - b. Distribution of publications

## DEVELOPMENT & EVALUATION GROUP

### Introduction:

The Development & Evaluation group was charged with focusing on advancing technology development and evidence generation. Two overarching principles guided the group's work. The first was the issue of contextualization: the development of genomic applications and the evaluation of evidence for their use is context-dependent. The second principle is an extension of the first: all development and evaluation should be in response and accountable to the emergent and critical clinical questions important to patients and their families.

The group discussed the continuum from precision medicine to population health and whether genomics is a value-based addition to healthcare. There is a tension between whether genomics applications

will simply add cost to the system and the possible benefit they might impart on patient care and outcomes. Should the focus be on screening for highly penetrant conditions or on the benefit in equipoise between therapy A or B? The abundance of information that is available because of next generation sequencing does not necessarily increase knowledge. Current mechanisms for aggregating variation, significance, and disseminating evidence are not adequate. The group felt that information systems that rapidly and seamlessly disseminate data and knowledge would be most effective. Lastly, the group discussed the barriers and challenges to information sharing, including the fact that current policies to protect patient privacy may hinder essential elements in accelerating translational research. Ultimately the

group asked: is genetics relevant in the public health context? If the answer is yes, then determining the clinical questions and, in response, developing the necessary technology and evaluating its clinical utility would be most productive.

### Outcomes:

The Development & Evaluations group identified three outcomes that they felt will be critical to advancing technology development and evidence generation including: 1) embedding a knowledgeable genomics presence into organizations and activities that impact healthcare decision making; 2) creating a framework for sharing knowledge; and 3) making the key clinical questions for public and individual health be the drivers for innovation in genomics.

### Priority Action Items:

#### OUTCOME 1: Embed genomics knowledge presence in all organizations and activities involved in healthcare decision-making

- I. Activities:
  - a. Create an inventory of relevant organizations.
    - i. Determine whether genetics/genomics expertise is present.
    - ii. Prioritize engagement with organizations that match the priorities for public health genomics.
  - b. Create a strategy for engaging identified groups.
    - i. List volunteer individuals with the requisite knowledge and skills.
    - ii. Match expertise to identified organizations.
  - c. Raise awareness of public health genomics issues through knowledge dissemination.

- i. Achieve through engagement, such as through advisory capacities or inclusion of individuals in organizations.
- II. Funding:
  - a. Approximately \$20,000 for coordination.
  - b. Can also use volunteers and cooperation.
- III. Stakeholder Needs:
  - a. Everyone
- IV. Timeline:
  - a. Approximately one year
- V. Metrics:
  - a. Inventory completed and publicly available
  - b. Priorities targeted
  - c. Determination of messaging

**OUTCOME 2: Create a framework for open access sharing, aggregation, and dissemination of knowledge**

- I. Activities:
  - a. Knowledge needs to be developed so that there is a continuum from the individual to the population and from research to service. Knowledge needs to flow from the individual to the population and from the population to the individual for decision-making at each level.
  - b. Convene a group that will create a compendium of knowledge developers.
    - i. Identify infrastructure needs to ensure data sharing, standards for interoperability of information, nomenclature, methodologies, etc.
    - ii. Convene developers working in this space to create a vision / principles, determine the gaps, and establish collaborations.
    - iii. Reconvene a subgroup in one year to assess progress.
    - iv. Plan and host a workshop on the commoditization of the genome.
- II. Funding:
  - a. \$50,000 for a staff person to coordinate the process.
- III. Stakeholder Needs:
  - a. Everyone
- IV. Timeline:
  - a. Approximately one year
- V. Metrics:
  - a. Completion of compendium
  - b. Identification of gaps
  - c. Collaborative pilot project developed
  - d. Dissemination of knowledge from workshop.

**OUTCOME 3: Position the key questions for public and individual healthcare as the driver of innovation in genomics**

- I. Activities:
  - a. Change the current paradigm from technology driving discovery to individual/population health benefit as the driver.
    - i. Have clinical questions be the seeds for implementation and innovation.
  - b. Enhance public/private collaborations to answer questions.
    - i. Define benefits of collaboration.
  - c. Identify 3–5 key questions where genomics has a role so that it can help build knowledge and data (generated by multiple groups).
  - d. Matchmaking between groups with clinical questions and researchers.
    - i. Need to identify synergies to increase incentives toward this end.
- II. Funding:
  - a. \$75,000 to staff, \$50,000 to analyze systems for questions
  - b. For the “matchmaking” activity, \$75,000 to staff and \$100,000 for an intelligent informatics system.
- III. Stakeholder Needs:
  - a. Everyone
- IV. Timeline:
  - a. One year to disseminate
  - b. A second year to analyze and decide
- V. Metrics:
  - a. Completion of compendium
  - b. Identification of gaps
  - c. Collaborative pilot project developed
  - d. Dissemination of knowledge from workshop.

## PATHWAYS & INTERACTIONS GROUP

### Introduction:

The pathways group was charged with understanding “how pathways and gene-environment interactions impact population health.” The group began by defining pathways and interactions as the fundamental way genetics is used to understand physiology. This includes the genetic basis of disease and health, as well as pathways that lead to disease or treatments. The group identified priority outcomes, for both research and practical implementation. Common themes that emerged included the importance of community engagement, the need for better measures of the environment (including social, behavioral, chemical and physical exposures) and the integration of these measures into research on human health and illness, and the need for education of the public and the workforce surrounding genetics and health. The group also acknowledged the importance of data sharing (including sharing data on environmental exposures, for which there is currently no central repository for data), leveraging resources that currently exist, and identifying and building from successful models. Barriers were also discussed and included the lack of coordination, lack of infrastructure, public distrust of the government, public skepticism regarding science, and current funding climate. Once all group members had the opportunity to provide input on their priority outcomes, the group listed a variety of organizations and projects that could be used as successful models for the themes discussed. The group also emphasized the contribution of genetics, epigenetics, and environments (again, social and physical) on genes and pathways and discussed the importance of looking at all three together, in order to understand common, complex diseases that are a major focus of public health. It is important to note that a cornerstone of the discussion was community engagement and the need to do it well.

### Outcomes:

To develop outcomes, the group reviewed the key areas discussed above and then selected two areas of focus for further discussion. The group split into two, worked on one area of focus, and then switched to the other area of focus. The group approached each area of focus with a comprehensive plan.

Area of focus: 1) Community engagement: Building trust, understanding and support of science (e.g. genes and environment) and its application to research and practice and 2) Creating a public health village: Through a process of

community engagement, develop a set of model systems that integrate partners of the public health system to use gene-environment interactions to solve and study a sexy problem that is of concern to the community.

Four general activities were discussed across both areas of focus: a) engage stakeholders in the community in genomics b) identify appropriate communication vehicles to reach the community to inform, educate and learn from the community, c) increase health/ science literacy, and 4) educate, train and recruit a diverse work force.

### Priority Action Items:

#### OUTCOME 1: Community engagement

- I. Activities:
  - a. Stakeholder engagement activity in research and practice
    - i. Define stakeholders through Community Based Participatory Research approach with diversity as a requirement
    - ii. Conduct landscape analysis using surveys and focus groups
    - iii. Learn from community and disseminate information to community
    - iv. Conduct community engagement activities
  - b. Use an RFP process to develop models for community engagement around genes and environment.
    - i. Test communication vehicles such as social media, YouTube, and television
    - ii. Develop champions
    - iii. Employ strategies to improve health literacy
    - iv. Utilize on the ground resources
  - c. Convene a national forum to bring best practices for dissemination to a larger stage.

d. Continue the process from this meeting in local corners.

II. Funding:

a. Funding will be needed through private / public partnerships and/or interagency funding.

III. Stakeholder Needs:

a. Broad, but a focus on the local level

IV. Timeline:

a. RFP process for 2012 with project timeline for 2013–2015

b. Dissemination at a national forum in 2016

V. Metrics:

a. Meaningful metrics are needed to improve public trust

b. Suggested metrics could include pre- and post- evaluation, the number of collaborations, uptake of genetics curriculum, graduate education, jobs created, etc.

c. Develop and use partnerships to begin to identify needs, opportunities, challenges, and media explosion opportunities.

i. Leverage current funding, programs, infrastructure, and encourage sharing

d. Create short-term health goals and business opportunities in these communities.

i. ROIs could include save lives, reduce health disparities, reduce health costs, reduce exposure, reduce hospital visits, trips to ER, etc.

e. Create ties to federal and local agencies

i. Open access to information, software, sharing, algorithms, informatics, resources, publications, etc.

f. Increase health and genomics literacy across all partners.

g. Conduct assessment and capture lessons learned

h. Project examples could include cancer in superfund sites, adverse drug events, asthma in children, or childhood obesity and diabetes.

II. Funding:

a. Funding will be needed through a combination of foundations, federal agencies, and philanthropists

III. Stakeholder Needs:

a. Broad, but a focus on the local level

IV. Timeline:

a. 5 year process

i. Gathering partners and identifying projects in year 1

ii. Create short term goals in years 2–3

iii. Develop ties with federal agencies in year 3

iv. Assess lessons learned in year 5.

b. Working to increase health and genomics literacy will occur across entire project.

V. Metrics:

a. Meaningful metrics to be developed based on specific projects

## OUTCOME 2: Create a public health village

I. Activities:

a. Gather partners

i. Partners could include departments of health, community groups, academia, health care providers, government, business and labor, celebrities and local heroes, media, and funders such as Robert Wood Johnson, Kellogg, Federal agencies, or philanthropists

b. Identify at least 4 major geographically diverse city projects in the country to serve as model.

i. Ensure connection and perhaps competition to raise sense of group mission to improve health of community

## PREVENTION GROUP

### Introduction:

The Prevention Group was charged with “identifying individuals, families, and communities at risk.” One of the first and most essential issues brought up by the group was the necessity of ending the theoretical and practical separation between genomics research and public health. The group highlighted the enhanced awareness of the importance of genetics and genomic factors in the etiology of chronic disease and increasing connections found between chronic diseases and genomics, and advocated that rhetoric and policy reflect this relationship. The necessary integration of genomics into public health, the group decided, should be accomplished through existing programs and institutions, utilizing tools already available. Furthermore, there should be constant surveillance to ensure that these integration activities reduce rather than widen disparities.

The Prevention Group also attempted to address the challenges inherent in education and messaging programs concerning public health genomics. One of the major barriers is public health’s interest in broadcasting simple, straightforward, one-size-fits-all messages that do not recognize genetic variability. On the other hand, members of the group acknowledged that often your zip code means more than your genetic code, making it difficult to communicate the vital role of genomics in health and disease. Though these obstacles may be overcome through efforts like a focus on family health history and/or partnerships with influential organizations such as the American Heart Association, meaningful education and engagement of individuals at the community level should be a component of all public health genomics initiatives. As one member of the group expressed —“genomics can’t remain the domain of the few.”

### Outcomes:

Multiple outcomes were identified, including: (1) genomic thinking integrated into healthy community initiatives at all levels and via all existing infrastructures (city, county, state, tribal, national), (2) family health history as a primary prevention tool integrated into clinical care

and public health practice, (3) genomics data incorporated into and utilized by public health surveillance systems such as cancer registries, (4) equitable distribution of pharmacogenetic and cancer genomic tools, and an (5) evidence base that reflects diverse ancestral populations.

### Priority Action Items:

#### **OUTCOME 1: Integrate genomic thinking into healthy community initiatives at all levels and utilize all existing infrastructures including city, country, state, tribal, and national**

- I. Activities:
  - a. Build strategic partnerships.
  - b. Raise awareness and educate through marketing campaign / strategic messaging
  - c. Other priority activities
    - i. Create more of a presence of genetic programs in Chronic Disease.
  - d. Create more interactions between Maternal and Child Health and the Chronic Disease Genetics program.
- II. Funding:
  - a. Encourage new funding mechanisms while at the same time ‘mining’ existing funding streams.
- III. Stakeholders Needs:
  - a. Everyone!
  - b. Genetic Alliance as champion
  - c. Health Resources and Services Administration gives imperatives
  - d. The ‘doers’ will be genetics organizations (public and private), state health departments, common chronic disease groups (such as American Cancer Society, American Heart Association, and American Diabetes Association), Prevention Research Centers, etc.

**OUTCOME 2: Standardize and integrate family health history as the primary prevention tool**

- I. Activities:
  - a. Incorporate family history, genetic test results, etc. into EMRs
  - b. Evaluate family health history as primary prevention tool (e.g., Lynch syndrome)
  - c. Evaluate efficacy of generic intervention in subgroups at high risk due to family health history (e.g., 70% of people in Diabetes Prevention Program have family health history of diabetes)
  - d. Other priorities:
    - i. Create more protective genomics privacy legislation, beyond GINA

**OUTCOME 3: Incorporate genomics data into public health surveillance infrastructure, such as cancer registries**

- I. Activities:
  - a. Review current registries to evaluate consistency and identify gaps.
  - b. Craft additional protective genomics privacy legislation, beyond GINA.
- II. Stakeholder Needs:
  - a. States
  - b. Surveillance, Epidemiology, and End Results at National Cancer Institute
  - c. National Association of Cancer Registries
  - d. American Cancer Society

**OUTCOME 4: Ensure the equitable distribution of pharmacogenomic and cancer genomic tools**

- I. Activities:
  - a. Surveillance activities to evaluate whether new cancer genomics and pharmacogenomic tests are being differentially accessed according to social demographics.
- II. Stakeholder Needs:
  - a. Agency for Healthcare Research and Quality
  - b. Patient Centered Outcomes Research Institute
  - c. Centers for Disease Control and Prevention
  - d. American Academy of Cancer Research
  - e. Professional societies such as the National Society of Genetic Counselors
  - f. Office of Minority Health
  - g. Office of Public Health Genomics

**OUTCOME 5: Develop an evidence base that reflects diverse ancestral populations**

- I. Activities:
  - a. Build trust with and obtain data from diverse populations.
- II. Stakeholder Needs:
  - a. National Human Genome Research Institute

## V. RESULTS

A careful review of the results presented by each of the breakout groups has led to the identification of several overarching priority objectives for the field of public health genomics:

- Public education about genetics
- Evidence development
- Rethinking the approach to technology development
- Embedding genetics in all aspects of healthcare
- Expanding public health screening programs that utilize genetic information

Each of these areas was identified as a priority by at least two groups and in most instances three or four. These priorities have been synthesized into the following five recommendations. Though the recommendations are broad, examples of specific activities and programs that the groups provided have been included. The recommendations are not comprehensive, but rather serve as a starting point for further discussion. In fact, several of the meeting attendees expressed an interest in continuing the discussion of these recommendations within their community to help identify local programs and activities that could contribute to overarching priorities. Development of mechanisms

for capturing and disseminating these discussions would be a valuable follow-up activity.

### Recommendations

#### Improve public education about genetics and related issues through community engagement

Several of the breakout groups made recommendations about activities and programs that could be used to improve public education about genetics and related topics through community engagement. The Detection group recommended the creation of a companion guide to the Genetic Testing Registry (GTR) currently being developed by the National

“I, \_\_\_\_\_, with \_\_\_\_\_ will...”

- “...convene to consider “Best Practices” (broadly defined) in Community Engagement about Translational Genomics Priorities & Practices.”
- “...exchange resources and data to promote genomics education at PH front lines in health centers and incorporate lessons learned into nursing education for future PH professionals.”
- “...educate the public of genetic research and the use of genetic information in healthcare to remedy misconceptions and positive receptiveness.”
- “...determine how postdocs could play a role in public education [relating] genomics to prevention.”
- “...work on public health campaigns on healthy food/eating and raise awareness of gene-environment impact on health.”
- “...inform community about Genomics in all Wash. Healthy Communities; work on healthy campaigns in Wash. St.; assist with identifying community partners.”

Institutes of Health. The group suggested the formation of a working group, including individuals with expertise in health communications, to produce a guide to help consumers evaluate the quality of information related to genetic testing.

The Pathways and Interaction groups identified the need to launch a public outreach and awareness campaign to increase health and science literacy. This program would include community engagement to learn about the health issues that are most important or of greatest interest to the public. The group recommended testing a variety of communication vehicles, including social media, YouTube, and television.

The Prevention group also felt that there was a need for a public education and outreach campaign to help raise awareness among the public of the vital role that genomics plays in personal health. We should not create an entirely new program; rather, it was recommended that the campaign be integrated with existing outreach initiatives. Health advocacy organizations, including common chronic disease groups like the American Heart Association, as well as government agencies such as HRSA were identified as key stakeholders for such a campaign.

Finally, several of the meeting participants made informal collaborative commitments related to public education and outreach. A few of these commitments are listed below.

**Continue to work on issues related to evidence development**

The working groups made several recommendations related to developing evidence for emerging technologies. The Development & Evaluation group felt

“I, \_\_\_\_\_, with \_\_\_\_\_ will...”

- “...work to increase clinical lab data submission into public databases.”
- “...develop criteria for addressing level of evidence need for implementation of a genetic / genomic test in patient care...with clinical molecular lab colleagues.”

that knowledge should flow from the individual to the population and from the population to the individual to promote effective decision-making at each level. To help achieve this, they recommended creating a framework for open access sharing, aggregation, and dissemination of knowledge. The Prevention group also indicated that there is a need to increase the diversity of individuals included in studies that form the evidence base. To increase the diversity of individuals included in research, the Prevention group recommended that the NHGRI focus on building trust with and obtaining data from diverse populations.

In addition to the recommendations issued by the working groups, several participants made informal collaborative commitments related to the topic of evidence development:

**Take a bottom-up approach to technology development**

Two of the working groups recommended that the public health genomics community rethink its approach to technology development. The Development

& Evaluation group recommended that we change the current paradigm from technology driving discovery to one in which clinical needs are the driver of the development process. Towards this end, the group recommended that the public health genomics community come together to identify three to five key areas where genomics has a role. Similarly, the Detection group recommended that the public health genomics community come together to identify the gaps in available testing and screening and transfer this information to the research community.

In addition to the recommendations issued by the working groups, one participant made an informal collaborative commitment related to the recommendation that we reengineer the technology development process:

**Embed genetics into all aspects of healthcare**

Every one of the working groups made the recommendation that genetics needs to be better integrated into healthcare. The recommended activities for achieving this outcome were many and diverse. The Detection group recommended that

public health genomics be incorporated into national data standards and health IT priorities. Towards this end, they recommended that public health leaders be involved in national committees and coordinate public health feedback to relevant policies, such as the advanced notice of proposed rule making recently issued by the Department of Health and Human Services with regard to the Common Rule. Such activities would not have

funding needs but would require participation from a diversity of stakeholders including academia, health departments, providers, health communications specialists, disease-specific organizations, laboratories, health insurers, and government agencies.

The Development & Evaluation group issued the recommendation that genomic knowledge and expertise be embedded

into all organizations and activities involved in healthcare decision-making. The group recommended that genomics professionals could serve in a volunteer advisory capacity to organizations in need of genomics expertise. To achieve this outcome, the group recommended matching professionals with the appropriate background in genomics to organizations in need of genetics expertise.

“I, \_\_\_\_\_, with \_\_\_\_\_ will...”

- “...develop a genomics/host factors project that interested sites will participate in...with the CDC Emerging Infections Program.”
- “...work to integrate family health history in existing community level programs in Flint & Genesee County”
- “...work together to strengthen collaborative genomics related efforts at the State and local health department levels.”
- “... reach out to the Health Information Exchange in my home state to find out what is happening regarding the exchange and public health monitoring of genetic / genomic information.”
- “...collaborate on integrating genetics programs with MCH Project at University of Maryland Prevention Research Center, Seat-Pleasant-UMD Health Partnership and Prince George’s County Hospital.”
- “...reinvigorate HO’s inclusion of genomics in all Health Community Initiatives (leadership role) Prevention and Chronic Disease.”
- “...write a white paper on the last IOM workshop re: integration of large amounts of genetic info into clinical practice.”
- “...work with community members to develop a family history / historical health project.”
- “...continue to assure community family history experiences are shared with the nursing community, PA, etc & that resources from those disciplines are shared with V. Ct. Project groups.”

The Pathways & Interactions and Prevention groups also recommended activities for improving the integration of genetics into healthcare. The Pathways & Interactions group recommended focusing on recruiting, educating, and training a diverse workforce, while the Prevention group recommended that genomic thinking be better integrated into healthy community initiatives. Participants made a number of informal collaborative commitments related to improving the integration of genetics into healthcare:

**Expand the public health screening programs that utilize genetic information**

The Detection and Prevention groups both recommended that the number of public health screening programs utilizing genetic information be expanded. The Detection group recommended that a process be put in place to expand public health screening programs to include cascade and life stage screening where appropriate. A few members of the group committed to working with the Genetics

for Early Disease Detection and Invention (GEDDI) working group in reviewing and drafting a publication with models and recommendations for integrating cascade and life stage screening programs.

The Prevention group recommended that family health history become the primary prevention tool. To achieve this outcome, there would need to be a push to standardize the information collected in electronic medical records. Additional studies are also needed that evaluate family health history as the primary prevention tool for specific diseases, such as Lynch syndrome. The group also recommended that studies be initiated to evaluate intervention that results from family health history. Lastly, the group recommended that there be additional protective legislation around genomics beyond the Genetic Information Nondiscrimination Act. This additional protective legislation will be needed as genetic information becomes more ubiquitous in the public health system and healthcare.

Several of the participants made informal collaborative commitments pertaining to expanding public health screening programs using genetic information and family health history.

“I, \_\_\_\_\_, with \_\_\_\_\_ will...”

- “...consider what genomic info should be collected by MN Birth Defects Information System.”
- “...compare implementation specifics on tumor-based Lynch syndrome Screening.”
- “...utilize successes of MI’s early cardiac death project for possible implementation in CT / N.E.”

## VI. CONCLUSIONS

This meeting provided a unique opportunity to convene a diverse group of stakeholders to discuss the field public health genomics over the next five years. The meeting agenda was designed to provide participants with the necessary context and maximum amount of time to produce concrete, actionable outcomes and activities. Even so, it was clear from the outset that the goal was ambitious for a one-day meeting. Because genomics has such a wide grasp, touching countless areas of public health, medicine,

research, and even individual identity, the development of clear, community-wide goals and objectives has always been a challenge. What is clear from the current state of funding for public health infrastructure and implementation is that the development of these goals is critical. Although the breakout groups were able to produce an impressive array of priority outcomes and activities, these recommendations should be viewed as a starting point for future discussions rather than the final result. Taking the high level

priorities identified at this meeting back to the local level will be essential for crafting a more comprehensive plan and ensuring that there is buy-in from the individuals who will be responsible for implementation. Moreover, with limited budgets and resources, the integration of genomics into public health cannot be taken for granted. Desire alone will not create that reality; a coordinated effort is essential. Those we serve call us to action; their health is at stake.

# APPENDICES



APPENDICES TO PART ONE

STAKEHOLDER CONSULTATION APPENDIX 1: PARTICIPANT LIST

Participant	Affiliation	RFI	KII	IDG	PC
Sylvia Au	Hawaii Department of Health	●			●
Diane J. Allingham-Hawkins	Hayes, Inc.	●			
Laurie Badzek	West Virginia University School of Nursing	●			
Elizabeth Balkite	ABQ Associates Inc	●			
D'Shane Barnett	National Council of Urban Indian Health	●			
Vence Bonham	National Human Genome Research Institute			●	
Connie Bormans		●			
Jo Boughman	American Society of Human Genetics			●	
James Bowery	Diogenes Institute	●			
Brian		●			
Frieda Brown	Prevention Research Center at Tulane University			●	
Linda Bruzzone	Lynch Syndrome International	●			
Wylie Burke	University of Washington Center for Genomics and Healthcare Equality	●	●		
Kim Caple	Life Technologies	●			
Richard Carmona*	17th Surgeon General of the United States	●			
Ned Calonge	The Colorado Trust	●			
Ann Cashion	The University of Tennessee Health Science Center	●			
Jean Chabut	Michigan Department of Community Health	●	●		
Toby Citrin	Center for Public Health and Community Genomics University of Michigan School of Public Health			●	●
David Collins	Morehouse School of Medicine Prevention Research Center			●	
Terri Combs-Orme	The University of Tennessee College of Social Work	●			
Yvette Conley	University of Pittsburgh	●			
Suzanne Cupal	Genesee County Health Department				●
Cornelia van Duijn	Erasmus MC	●			
Debra Duquette	Michigan Department of Community Health			●	●
William Ebomoyi	Chicago State University	●			
Karen Edwards	Center for Genomics and Public Health at the University of Washington				●
Jimmy Efird	The Brody School of Medicine	●			
Alex Ellerbeck	Genetic Alliance			●	
Jim Evans*	The University of North Carolina at Chapel Hill	●			
Greg Feero	National Human Genome Research Institute			●	
Suzanne Feetham	University of Wisconsin-Milwaukee	●			
Cindy Fogle		●			
Gregory Fowler	Oregon Health & Science University	●			
Lorraine Frazier	University of Texas at Houston	●			
Steven Galen	Primary Care Coalition	●			
Aaron Goldenberg	Case Western Reserve University			●	
Frieda Gonzales	New Mexico Department of Health			●	
Karen Greendale	New York State Department of Health			●	●
Ella Greene-Moton	Center for Public Health and Community Genomics University of Michigan School of Public Health NCC SPIG			●	●
James Haddow	Women & Infants Hospital of Rhode Island	●			
Jill Hagenkord	Creighton University	●			
Maxine Hayes	Washington State Department of Health		●		
Catherine Haywood	Prevention Research Center at Tulane University			●	
Winona Hollins-Hauge	University of Washington Health Promotion Research Center NCC SPIG			●	●
Joyce Hooker	Mountain States Genetics Regional Collaborative Center	●			

\*RFI response submitted via email    \*\*Name not listed    Note: Participant names and affiliations appear as reported on original RFI responses

Key:    RFI = Request for Information    KII = Key Informant Interview    IDG = Informal Discussion Group    PC = Planning Committee

Participant	Affiliation	RFI	KII	IDG	PC
Dean Hosgood	National Cancer Institute				●
James M. Hughes	Infectious Diseases Society of America	●			
Jonathan Izant	Sage Bionetworks	●			
Jenny Johnson	Utah Department of Health	●			
Josh		●			
Sharon Kardia	University of Michigan School of Public Health				●
Kim Kaphingst	Washington University in St. Louis School of Medicine		●		
Michelle Kempf-Weibel	Wisconsin Division of Public Health			●	
Barbara Koenig	Mayo Clinic, University of California San Francisco			●	
Chris Kuzawa	Northwestern University		●		
Dale Lea	Maine Genetics Program			●	
Leonard Levy	Nova Southeastern University College of Osteopathic Medicine	●			
Julian Little	University of Ottawa	●			
Donald Lyman	National Association of Chronic Disease Directors	●			
Ruth Lynfield		●			
James Madara	American Medical Association	●			
Chikezie Maduka	The University of Maryland Prevention Research Center		●	●	
Gail Marcus	North Carolina Division of Public Health			●	
Barbara Burns McGrath	University of Washington School of Nursing				●
Amy Miller	Personalized Medicine Coalition	●			
Amber Mills	Genetic Alliance			●	
Claudia Mikail	American Public Health Association Genomics Forum	●			
Maki Moussavi	Cerner Corporation	●			
Kenneth Olden	CUNY School of Public Health at Hunter College		●		
Timothy J. O'Leary	Association for Molecular Pathology	●			
Tricia Page	Emory University	●			
Jacqueline Johnson Pata	National Congress of American Indians	●			
Elizabeth Prom-Wormley	Virginia Commonwealth University			●	
Jean Public		●			
Michael Rackover	National Human Genome Research Institute American Academy of Physician Assistants			●	
J. James Rohack	Scott & White Center for Healthcare Policy	●			
Nuanonong Seal	University of Wisconsin-Milwaukee	●			
Joan Scott	National Coalition for Health Professional Education in Genetics	●			
Laura Senier	University of Wisconsin-Madison	●			
Sharon Shad	University of Massachusetts Medical School Worcester County Prevention Research Center			●	
Sara Shostak	Brandeis University	●			●
Cara Tenenbaum	Ovarian Cancer National Alliance	●			
Steven Teutsch	Los Angeles County Department of Public Health	●	●		
Jillian Tietjen	University of Michigan			●	
Philip Zazove	University of Michigan	●			
Ron Zimmern*	PHG Foundation	●			
Kristi Zonno	American Public Health Association Genomics Forum Policy Committee	●			
Deborah Klein Walker	Abt Associates Former President of American Public Health Association		●		
Michael Watson	American College of Medical Genetics	●			
Imogene Wiggs	Prevention Research Center in St. Louis Missouri Department of Health & Senior Services NCC SPIG			●	●
Marc Williams	Intermountain Healthcare			●	
Herbert F. Young	American Academy of Family Physicians	●			
Amy Zlot	Oregon Health Services			●	
**	Infectious Diseases Human Genomics Working Group	●			
**	Oregon Health Authority	●			

\*RFI response submitted via email    \*\*Name not listed    Note: Participant names and affiliations appear as reported on original RFI responses

**Key:**    RFI = Request for Information    KII = Key Informant Interview    IDG = Informal Discussion Group    PC = Planning Committee

APPENDICES TO PART ONE

STAKEHOLDER CONSULTATION  
APPENDIX 2: LITERATURE REVIEW

CURRENT AND FUTURE TRENDS IN PUBLIC  
HEALTH GENOMICS:  
A MULTIFACETED LITERATURE REVIEW

Stephen M. Modell, MD, MS\*, Nora Isack, MPH, Megan Knaus, MPH  
September 29, 2011

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A. INTRODUCTION AND METHODOLOGY

Public health genomics departs from classical genetics in its emphasis on the full assembly of genes within the human being, not just single genes, and its consideration of the genome’s interplay with the person’s surrounding physical and social environment. Individuals, families, and communities stand to benefit from validated discoveries emerging in the field having the potential to guide public health and clinical practice. Practitioners also have a clear call to direct new developments toward the solution of larger dilemmas, such as healthcare disparities and environmental challenges impacting schools and neighborhoods. These levels of research and action were held in mind as this literature review was developed.

Themes for this review were based on Planning Committee recommendations from inspections of public consultation Request-for-Information responses and a frequency analysis of themes in the responses, as well as interpretation of those themes appearing most frequently in the public health genomics literature. Inclusion and exclusion of articles was based on centrality of the article to the themes being explored. Literature reviewed came from three main sources: an active literature search, suggestions made by the expert Planning Committee, and literature submitted for the September 14, 2011 “Developing Priorities for Public Health Genomics 2012–2017” meeting convened in Bethesda, Maryland by the Centers for Disease Control and Prevention Office of Public Health Genomics. A GoogleScholar search was begun prior to the public launch of a CDC Request-for-Information (RFI) on the most important steps in research, policy, and practice for the field of public health genomics in the next five years. Combinations of the following key terms were used: Public, public health, community, consultation, participation, engagement, wiki, web, Internet, genetics, genomics, and typology. The search yielded the following sets of articles (reviewed / unreviewed): genetics- and genomics-related (11/0); other health-related (6/6); participatory methods and non-health-related (7/1); wiki- and web-related (10 + 1 book/6+ 1 book). Separate general searches were conducted to find examples of participatory wikis and government commission requests for public commentary (Secretary’s Advisory Committee on Genetics Health, and Society (SACGHS), the Secretary’s Advisory Committee on Genetic Testing (SACGT), and the National Bioethics Advisory Commission (NBAC)). These papers informed the process of data collection and analysis of the stakeholder consultation RFI responses and are reviewed in section J. below. Genetic Alliance maintained an electronic drop box of public health genomics literature for the September Bethesda meeting; 12 documents were downloaded and reviewed from this drop box; several other major papers from organizations listed in the drop box were independently downloaded. A third major source of document suggestions that were assiduously pursued and downloaded was the Stakeholder Consultation Planning Committee

convened by the Center for Public Health and Community Genomics. These suggestions substantially fill the several pages of bibliography found at the end of this literature review. The last two sources also provided author and organizational names and themes that suggested further acquisitions.

## B. POLICY CONTEXT

Essentially all the topic discussions below end with recommendations for policy creation; for that reason the review will avoid a single, stand-alone section on policymaking. Policy development is one of the three core functions of public health genomics, the other two being assessment and assurance (ASTHO 2001, pp. 2–3; Beskow et al. 2001, pp. 4–5). Policy development may be defined as the formulation of standards and guidelines by a collaboration of stakeholders that promotes the appropriate and effective use of genomic information and associated genetic tests and services (ASTHO 2010, p. 9). Conferences to develop screening guidelines for conditions like cystic fibrosis, hereditary hemochromatosis (iron overload disorder), and Lynch syndrome (hereditary nonpolyposis colorectal cancer) have been populated by medical and public health experts alike. A 2007 article examining the relationship between medicine and public health genomics argues for a partnership between them, in which information from healthcare providers and healthcare records can be merged with public health (epidemiologic, cost-benefit, death certificate) records and data to produce population-relevant guidelines (Khoury et al. 2007, p. 316).

Public health has a real opportunity to set the precedent and pace in policy formation. Agencies like the Centers for Disease Control and Prevention (CDC) can help initiate policymaking in areas where genomic applications are emerging by holding workshops with multiple sectors (including consumers and industry) and policymaking groups at the table, then pulling together data from different directions that reveal the population risk associated with genetically-associated conditions and validity/utility of testing technology. Though biologic information on text-book Mendelian conditions is important, public health is in a unique position to gather information and formulate policy for common, complex conditions that conditionally manifest (e.g., having a low penetrance), for asymptomatic carriers (often at-risk family members), and for the myriad of conditions where a test result will have a potential psychosocial impact (Ibid., pp. 315–6). Public health officials are also uniquely aware of the PHELSEI (public health ethical, legal, and social issues) laden with the new technologies, including the debate about genetic exceptionalism (“Should this technology merit protections beyond the conventional tools of public health?”), privacy and confidentiality (HIPAA requirements vs. the needs of public health researchers and consumers), and informed consent (e.g., with family studies and biobanking) (Burke et al. 2010, p. 786;

Office of Public Health Genomics, Institute for Public Health Genetics, Public Health Genetics Unit 2005, pp. 9–10).

## C. T1/2 TRANSLATION RESEARCH

Journals like the *International Journal of Epidemiology*, *Epidemiologic Reviews*, and *Genetics in Medicine* publish systematic reviews of genetically-related conditions of public health significance. These reviews help guide whether a genetic test should hit the mainstream, a provider should consider a genetic test for a patient or family member, or a patient should undergo a genetic vs. more conventional test. Public health is in a position to act as an “honest broker” to inform providers, the public, and policymakers whether a genetic test for a specific condition or set of tests for a family of conditions (such as familial colorectal cancer or heritable arrhythmias) is ready for “prime time” (Khoury et al. 2011a, p. 488). The honest broker role can be performed at multiple levels, such as by a federal agency or state health departments. It involves a trickle down of information, but also a shared motivation to address stakeholders’ questions in a language that will best serve them.

The issue of progress in translation research appears frequently in the literature. A contrast is built between the public health vision and the National Institutes of Health (NIH) Roadmap, which defines the translation framework in terms of drug discovery. Medical and public health approaches are distinct in the former’s emphasis on clinical trials leading to curative therapies, and the latter’s emphasis on early prevention and long-term outcomes (Khoury et al. 2007., p. 311; Zerhouni 2003, p. 72). These days both depend on the results of molecular genetics, especially genome-wide association (GWA) studies that can lead to a set of testable polymorphisms (Green and Guyer 2011; Khoury et al. 2011b). Public health genomics tempers its zeal by a standard that requires consideration of the full flush of translational steps from primary research about a disease condition or genetic test to assurance of effective service delivery (Khoury et al. 2007, p. 314). This translation highway is commonly described by T1/2 and T3/4 (Arar et al. 2011, pp. 194–5). T1/2 relates to the initial translation from a gene discovery to clinical guidelines, while T3/4 focuses on the use of new genetic tools in practice and the evaluation of their impact. There is also a certain amount of desperation voiced about the translational research enterprise as it attempts to channel benefits from an onrushing cascade of new genetic tests: “It is becoming increasingly difficult for evidence-based, independent review panels to evaluate quickly and thoroughly the proposed health benefits and harms of the fast-growing number of genetic tests and family health history tools” (ODPHP 2011, p. 2).

The existence of a centralized body able to judge the evidence is instrumental in moving the translation research enterprise along. The U.S. Preventive Services Task Force (USPSTF), now

in its third iteration, has generated evidence reports in a large number of chronic disease and mental health areas (Harris et al. 2001, p. 24). The Agency for Healthcare Research and Quality (AHRQ) and the National Human Genome Research Institute (NHGRI) have produced technology assessments for the use of family health history in the clinical and community settings. The Evaluation of Genomic Applications in Practice and Prevention (EGAPP) working group led by CDC produced eight genomic evidence reports between 2006 and 2010, or one to two evidence reports per year. Several, such as the reports on Lynch syndrome genetic testing strategies and on testing for cytochrome P450 polymorphisms in adults with nonpsychotic depression, have provided arguments for either speeding up or regulating the marketing of tests, and have been widely cited (Grossman et al. 2010, pp. 597–617; Teutsch et al. 2009). In the case of Lynch syndrome, valid screening interventions could reduce the risk of colorectal cancer by 60 percent (ODPHP 2011, p. 1). The production of evidence is valuable if you are the consumer getting a genetic test; the important question is one of pace.

The gathering of primary science (T1) evidentiary data in the public health genetics context has sprouted two strategies: one cumulative—the assessment on a study-by-study basis of whether a condition is ready for population testing, and the more wholesale accumulation of data via population-wide means. With respect to the latter, the 2001 ASTHO framework for public health genetics policies and practices recommends the development of a health data collection system with the capacity to “monitor genetic factors that affect health status and identify health problems within the community” (ASTHO 2001, p. A-3). Fulfilling this objective, in its “Vision for the Next 10 Years of Public Health Genomics at CDC,” the CDC Office of Public Health Genomics (OPHG) announces the launch of a new initiative, Beyond Gene Discovery (BGD), to assess population genetic variation across the United States in relation to health and disease, and ultimately develop strategies to impact health and eliminate disparities between population groups (OPHG 2007a, p. 3). BGD’s initial focus was to cull the U.S. National Health and Nutrition Examination Survey (NHANES) and NHANES III for genetic data from a representative sample of 15,000 persons. This project would meet OPHG’s goal of addressing disparities since it “oversamples the two largest race/ethnic minority groups, non-Hispanic blacks and Mexican Americans, along with other subgroups of the population” (Ibid.).

Healthy People 2020, however, cautions against overspecialized sampling. “As the number of recommended tests increases, valid and reliable national data are needed to establish baseline measures and track progress toward targets. Many tests are recommended for use in small subpopulations, making it difficult for most national health information systems, such as the National Health Interview Survey, to monitor progress” (ODPHP 2011, p. 2). BGD actually seems to have two missions. On the one

hand lies the ambition to develop the first “Genome Profile of the United States,” which would survey the most common genetic variants in the country, including for racial and ethnic population groups. This goal seems practical and to fill a need not yet addressed. On the other lies the desire to develop a searchable, online information system of human genome variation (allele, genotype, and haplotype frequencies at individual and multiple genetic loci) readily accessible to researchers. This second goal resembles more the compilations of mutational data that fill scientific databases and provide the groundwork for many of OPHG’s HuGE Reviews. Countless rare mutations and low frequency polymorphisms fill the ranks of these databases, hardly a reflection of the attempt to address common illness in major segments of the population. The real distinction seems to be between placing emphasis on a scientific investigative agenda vs. squarely addressing the causes of health disparities.

Major projects such as that of forming a genomic profile of the population would likely carry on a tradition of collaboration formed between federal agencies and investigative teams. For example, the National Cancer Institute (NCI) has funded epidemiologic consortia that have designed GWAS leading to the uncovering and risk characterization of numerous low penetrance cancer susceptibility genes (Khoury et al. 2011b, p. 7). Such collaborative studies represent T1 level basic science research identifying alleles for further translational projects, perhaps BGD. The medical research community has utilized major discoveries such as the role of the *HER-2/neu* oncogene in breast cancer genesis and mutations in the *KRAS* gene in colorectal cancer growth for the development of therapies and prognostic markers for secondary prevention. It is also possible that T1 knowledge of genetic variants can be carried on into larger, population-oriented projects to determine extent of risk and more fully map-out which population segments might benefit from greater attention (Khoury et al. 2011a, p. 492). This effort corresponds to public health’s assessment role. However, to avoid the problem of over-division decried by Healthy People 2020, mutations and polymorphisms that are mapped could be judiciously chosen according to their public health relevance rather than replicating the type of primary investigations that observational and experimental mutational studies undertake. The original idea behind a population-based effort was actually to provide a platform for measuring “the prevalence of genotypes *with potential public health importance* [italics added]” (Gwinn and Khoury 2006, p. 22).

Research at the next, T2 level concerns the development of evidence-based recommendations applicable in both medical and public health contexts. At least two steps seem to be needed to yield recommendations that can either initiate or influence guidelines, though these steps are not neatly distinguished in the literature. First, a multidisciplinary process of consensus-building must take place, involving a variety of experts (Office

of Public Health Genomics, Institute for Public Health Genetics, Public Health Genetics Unit 2005, p. 12). Second, what data passes muster is determined by a broader information synthesis. At this stage (one could almost call it T2B), public health has filled a genuine gap in evaluating what genetic tests might be developed further which would have an impact on the greater population. The T2B information synthesis process, because it depends on numerous sources of information and coordinated teamwork, takes time. For example, between 2001 and 2006 the United States Preventive Services Task Force (USPTSF) developed only two evidence-based guidelines related to genomic applications (OPHG 2007a, p. 2).

When information synthesis occurs in overlapping medical and public health fields, opportunities exist both for collaboration and further delineation of mission. The Institute of Medicine (IOM) report on comparative effectiveness research has outlined a goal of comparing the effectiveness of genetic and biomarker testing, and usual care in preventing and treating a variety of cancers, from lung to prostate (Khoury et al. 2011b, p. 25). The National Cancer Institute (NCI) working group has recommended forming a knowledge synthesis study group to assess genomic and nongenomic factors affecting treatment response and adverse events and potential pharmacogenomic applications (Ibid., p. 26). These bodies share an interest in the identification of cancer mutations and cancer prevention with the CDC divisions, but the above goals do not move in the direction of developing scientific standards for personal genomic tests or enhancing population research and wellbeing (Ibid., p. 25). Information synthesis would proceed on a quicker footing if the various agencies each carve out their particular niche and avoid overlap. The rule is “Divide and conquer.”

An expert group convened in 2008 by the NHGRI addressed clinical and public health research translation in equal measure and arrived at a practical interim solution to the moderate pace of T2 translation. The group suggested a bidirectional exchange of knowledge between basic discovery and translational research according to an “inverse planning” model (McBride et al. 2010, p. 558). The kernel idea is that translational research could be conducted using early prototypes “such as a representative set of genetic susceptibility tests that embody important characteristics of future tests—not necessarily the full set of markers” under development. The prototypes could be evaluated in comparison with other existing interventions to assess providers’ interest and practice utilization (Ibid.). This proposal might be one means of collecting the necessary clinical validity and utility data quicker, enabling a speedier synthesis process.

#### D. T3/T4 TRANSLATION RESEARCH

The next steps in translation research involve implementation and dissemination to move discoveries into practice and control

programs (T3), and assessment of interventional effectiveness, cost effectiveness, and outcomes at the population level (T4) (Khoury et al. 2011b, p. 5). The data outlook is rather sad, for lack of a better term, when it comes to product movement through these latter stages. An inspection of PubMed and HuGE Navigator queries reveals that the number of publications reporting results of human genome discoveries far exceeds (by more than a 10:1 ratio) the number shuffling these discoveries into workable clinical applications (McBride et al. 2010, p. 557). Moreover, a recent analysis of NCI-funded genome research shows that less than 2% is post “bench to bedside,” and less than 1% is post bedside (Khoury et al. 2011b, p. 9). The causes of moderately paced adoption of new tests and interventions go beyond behavioral aversions to change to a more macroscopic set of factors, including slow adoption of revised professional society standards (an outcome of T2 research), public health and health system past precedent, the financial cost of change at all levels within the system, health plan coverage of a new test, administrator and provider willingness, and consumer preferences, attitudes, and perceptions.

Approaches to addressing hindering forces at the T3/T4 translation research level occupy at least three categories: active, advisory, and investigational. One active approach would be the attempt to change policy at the governmental level, as is being done with professional society and advocacy group leverage on the FDA to regulate Direct-to-Consumer (DTC) genetic testing. It is conceivable that similar leverage could be imposed on the right bodies to modify the rate at which CLIA approval of laboratory genetic tests is taking place, what has been likened to a rebalancing of the regulatory environment (Burke et al. 2010, p. 787). It is unclear how effective this approach would be in the genomics domain as DTC companies seem to have at least temporarily outstripped government in release of new genetic tests and test panels. The market itself is exerting pressure and compelling the industry to establish linkages with practitioners and health plans.

Another active approach that side steps government fences is to go ahead and disseminate information about new tests, interventions, and validity/utility information directly to end-users. This approach, to its credit, has been used by the NCI in the formation of the Global Tobacco Research Network (Leischow et al. 2008) and by CDC-OPHG in the formation of the Genomic Applications in Practice and Prevention Network (GAPPNet) (Khoury et al. 2009). Both agencies have formed virtual networks of scientists, professionals, and end-users to exchange leading-edge information on the Internet in hopes of easing pathways taken by the translation process.

Public health genomics has used the advisory approach for constructive gain. GAPPNet convened committees to brainstorm methods to accelerate and streamline the effective integration of validated genomic knowledge into medical and public health

practice (Khoury et al. 2009, p. 490). The committee roles, devoted to knowledge synthesis and translation research and programs, were designed to facilitate this process. One outstanding aspect of GAPPNet, which serves as a model, is the capacity to gather together different sectors—professional, industry, non-profit, and academe. This kind of human interlocking promotes the exploration of solutions. Another example of translational advisory work is the convening of a multi-disciplinary working group by CDC and NCI to tackle issues in the implementation of Lynch syndrome screening (Khoury et al. 2011b, p. 9). The group identified challenges at the individual, family, system, and policy levels leading to clinical and population strategies.

The genomic applications investigational route is in its early stages. GAPPNet has begun a process of inquiring about methods and approaches key informants use in overcoming genetics translation hurdles. This effort takes the form of surveys, and actually has been conceived in several forms by GAPPNet committees. The assessment can be purely empirical (“grounded”) or the investigators can have a preliminary working model in mind beforehand. It can be directed at collecting research translation approaches in general, or at examining a particular aspect (e.g., the incorporation of electronic health record systems). Another investigational route is to examine translational research case studies. A working group put together by the Knowledge Synthesis and Dissemination Committee considered facilitating and impeding factors within professional societies, the Veterans Administration and the Kaiser Health System in an examination of cancer diagnostic test kit uptake (Arar et al. 2011). The case studies approach can be used to form projections of the pace of technology adaptation. These types of research can take place at the committee or working group level, independent of the meeting schedule of the entire organizational membership. The T3/4 translational research gap can be approached by multiple routes. The gap continues in the same way that other public health dilemmas dealing with complex systems, such as drug abuse or racial-ethnic disparities, persist through time. Like them, the translational research gap will loom as a continuing challenge, rather than an impassible hurdle, over the decade to come.

## E. GENE-ENVIRONMENT RELATIONSHIP

While contemplating the pace of technology adaptation, public health investigators and practitioners need to bear in mind that “generally, addressing social determinants of health (reduced poverty, increased education) can have the largest impact on population health” (Khoury et al. 2011a, p. 487). The circumstances that envelope an urban dwelling family—industrial factories and highways, reduced income and the inability to renovate a home, long distances to obtaining fresh food—will have a health effect. These impacts are public health concerns and need to be taken into account when formulating genetics policy

(McBride 2010, p. 558). At the same time, a certain amount of selectivity is needed in applying the tools of public health. House mites and dust in inner city dwellings can be responsible for the increased prevalence of asthma in urban minority home dwellers, and this knowledge can very well aid public health authorities, but not every family shows a predisposition to asthma. Public health genomics makes possible the avoidance of a “one-size-fits-all” approach.

Omenn likens the increasing ability to bring statistical, epidemiologic, environmental, and genomic tools to bear on eco-genetic health problems as the “Golden Age” for the public health sciences, albeit one that we are just now entering (Omenn 2010, p. 2; 2001, p. 32). In the case of asthma, a study of the genotypes of affected children in Mexico found that simple supplementation with antioxidant vitamins C and E improved lung function in youngsters with a common glutathione transferase polymorphism exposed to ozone (Khoury et al. 2007, p. 313). GWA studies have revealed a large number of possible associations with small effects. Increases in incidence and differential rates of transmission from mother and father suggest the influence of environmental factors and gene-environment interactions with imprinting. Omenn suggests we are entering an era where epigenetics could help transform asthma therapy from palliation to prevention (Omenn 2010, p. 4). The overall story shows that eco-genetics can foreseeably have a gradual effect, with general measures preceding insights that lead to more specific solutions.

Smoking is a major risk factor for chronic bronchitis and emphysema across the globe. Studies have yielded results from both ends, e.g., an association between the *CYP2A6* polymorphism and tobacco dependence (Khoury et al. 2011b, p. 8), and one between the *bOGGI Cys/Cys* genotype and adenocarcinoma of the lung in heavy smokers (Gwinn et al. 2009, p. 704). The argument has been made that individuals who believe they are not genetically susceptible will be emboldened to smoke, though this need not be the case. Further, second-hand smoke is a cause of lung cancer in family members of those who smoke. Knowledge of predisposition to lung cancer could be helpful to whole families. Gwinn et al. remind the reader of the heterogeneity of results in GWA cancer susceptibility studies, and argue that the investigators of the future must collect and analyze “candidate exposures” just like “candidate genes” (Ibid.).

Lead exposure is another prime example—fetal exposure is associated with adverse pregnancy outcomes and developmental and cognitive deficits. Public health campaigns led to the removal of lead from gasoline, resulting in a dramatic drop in serum lead levels in the United States (Khoury et al. 2011a, p. 491). In some segments of the U.S. and in many developing countries, however, excess lead exposure still represents a public health problem. A study of 103 umbilical cord blood samples from mothers in Mexico found that greater levels of lower extremity bone lead

levels were associated with diminished cord blood methylation levels (Pilsner et al. 2009, p. 13). The authors note that the fetal epigenome may bear the brunt of the intergenerational transmission of lead burden, which could influence long-term epigenetic programming and affect disease susceptibility. Untoward influence on a young person's development is a fairly serious outcome. It would be quite useful to have a handle on predicting outcomes in groups sensitive to lead levels.

In looking to the future, a 2004 IOM conference examining the implications of recent developments for genomics and public health concluded “progress is being made to identify and prevent gene-environment interactions,” but that public health researchers and practitioners need to persist in uncovering the environmental factors impacting at-risk groups (Hodge 2004, pp. 23–4). If one considers that drugs are external agents taken in from the person's environment, then the combination of therapy plus genetic diagnostic kit represents one wave of the eco-genetic era already taking place. Toxicogenomics and nutrigenomics represent two other crests in the speeding wave front (Schulte 2011, p. 4). In considering the ecologic factors underlying disease, it is also worth remembering that social conditions, looking from the inward vector of the gene outward to surrounding circumstances, are often a root cause of ill health (Khoury and Gwinn 2006, p. 282; Shostak 2003, p. 2338). Family history, reasons Omenn, is one way of assessing environmental, behavioral, and cultural factors while taking genetic predisposition into account (Omenn 2010, p. 6). In the final analysis, though, health policy-makers should avoid emphasizing innovative technologies to the exclusion of environmental and grassroots social solutions.

## F. FAMILY HISTORY

A host of common complex diseases are still in search of exact genetic identities. For example, three years ago the FUSION (Finland-United States Investigation of NIDDM Genetics; <http://fusion.sph.umich.edu>) Study yielded three likely gene candidates for type-2 diabetes; combined GWA studies have now expanded that number to 30. While large-scale studies of this magnitude will hopefully bring to fruition the promise of personalized genomics, in the immediate term multifactorial conditions such as diabetes are best served by recourse to the “first genetic test”—family health history. A review of NHANES data showed that addition of family history to the analysis of the undiagnosed population in the U.S. could yield more than 600,000 new cases (Khoury et al. 2011a, p. 491). This example underscores the Healthy People 2020 statement that family health history is an important risk factor for common diseases, and that it has the potential to improve health by finding people who are at risk for future disease or who are already sick but have not been diagnosed (ODPHP 2011, p. 2).

The U.S. Surgeon General's “Family Health Portrait” tool has been out since 2004, and the CDC and major universities have conducted pilot studies to assess family history tools of use in both public health and the primary care setting (Valdez et al. 2010). More specific work is yet to be done. The use of family history has been validated more rigorously for some disease categories than others, e.g., heart disease and type II diabetes more so than stroke and asthma. In some instances risk assessment tools are being used that have not yet been validated, so that disease specific work needs to continue (Ibid.). The “Family Health Portrait” needs to be developed to the point where it yields risk scores that can provide definitive advice to caregivers and tailored recommendations to family members (OPHG 2010a, p. 3). Further, family history information can be used in community-based screening, and provide motivation for participation and adherence (Omenn 2010, p. 6).

Family health history is uniquely adaptable to both healthcare and public health. Healthcare systems are currently undergoing a heavy push to incorporate health records into electronic systems that yield point-of-contact information. Family health history should be incorporated into electronic health record systems and, indeed, this process is now happening in a range of for-profit and nonprofit healthcare systems including the Veteran's Administration (Ibid.). From 2003 to 2008 the health departments in four states (Michigan, Minnesota, Oregon, and Utah) established programs to integrate family history assessment tools and surveillance findings into state and local chronic disease prevention and health promotion strategies (ASTHO 2010, p. 12). Other chronic disease programs, such as WISEWOMAN, also utilize family history risk assessment and educational activities. These efforts need to continue and to expand into other states, and very much depend on the availability of funding from the federal level.

An external review panel of OPHG's activities recommended “Expand the family history project by adding a translational research dimension” (OPHG 2007b, p. 1). Experts are not the only ones to advocate for this type of translation, however. Participants in a National GenoCommunity Think Tank (<http://genocommunity.org>) comprised 54% of community organization members (38% from academia; 6% from public health practice), marked family health history as the top-most (59/74 respondents) thematic area they would include in their future activities, followed closely by attention to health disparities, then addressing chronic disease prevention. The intersection between chronic disease and family health history may be one reason community members expressed such a vigorous interest (Modell 2010, pp. 3–4).

A large proportion of participants in the Think Tank represented community-based organizations (CBOs) and Prevention Research Centers (PRCs) assisting communities with significant

non-white racial-ethnic and lower to lower middle SES distributions. CBOs often incorporate chronic disease prevention into their health-related activities; family history needs to be made an accessible tool for community action. Efforts to enable diverse communities to take advantage of family health history in culturally relevant contexts should be promoted. Validated resources exist that require transfer from the piloting state to commonplace usage (Genetic Alliance 2010).

## G. RACIAL-ETHNIC DIVERSITY

Chronic diseases of major public health significance such as coronary heart disease, stroke, asthma, diabetes, and multiple cancers (gastrointestinal, prostate, cervical) display racial-ethnic disparities (National Prevention Council 2011a, p. 25). Public health practitioners appreciate the role of behavioral and environmental risk factors in health and illness, including tobacco use and adverse diet in African Americans and excess salt intake in the diet of Japanese citizens. In addition, African American men and women tend to be diagnosed at a later stage of cancer, which has led to higher case-fatality rates. The Institute of Medicine 2003 report on racial and ethnic disparities in healthcare showed that inequalities remain even when socioeconomic and access-related factors are controlled, suggesting a variety of factors, including systemic or organizational patterns, behavioral practices, and biological factors (OMH 2010, p. 57957). A lasting approach to health disparities cannot be singular. The National Prevention Strategy framework outlines four pillars for action to improve health and quality of life: (1) healthy communities; (2) preventive clinical and community efforts; (3) empowering individuals; and (4) eliminating health disparities, but (4) can itself be addressed by attending to the first three pillars (National Prevention Council 2011b, p. 10).

Genomic public health efforts touch on a number of ways to reduce health disparities, such as the promotion of family health history in diverse communities, offering or referring at-risk persons for genetic screening where indicated, e.g., fecal genetic testing for colorectal cancer, and promotion of behavior change in at-risk groups. These efforts lie more in the area of assurance than assessment. Both the National Prevention Strategy and the HRSA Strategic Plan have improvement of health equity as a goal, with use of testing and “harness(ing) technology” as vehicles (National Prevention Council 2011b, p. 6; HRSA 2010, p. 2). The future of public health genomics must take account of these national objectives, particularly since public health has a concern with groups that have to the present suffered marginalization. The situation in the United States differs somewhat from those European countries offering universal health care such that all groups may equally benefit from public health assessment and intervention efforts. The report of an expert workshop on genome-based research and population health (Bellagio Report)

argues that public health genomics has as its focus population health; and that nuanced approaches along the targeted intervention-“one size fits all” spectrum should incorporate differences in individual susceptibility (OPHG, Institute for Public Health Genetics, Public Health Genetics Unit 2005, pp. 7–8). Combine these two thoughts and the outcome is that public health genomics should pay special attention to population groups of differing susceptibility or risk.

The Office of Minority Health (OMH) of the U.S. Department of Health and Human Services has developed guiding standards for culturally and linguistically appropriate services (CLAS standards) that apply just as much to genetic services as they do to other conventional forms of testing and treatment (Graham 2008; OPHS 2001). These standards recently underwent public comment under an OMH enhancement initiative (OMH 2010). The first of the twelve standards urges that health care be delivered in a manner compatible with patients’ and consumers’ cultural health beliefs and preferred languages, principles that are consonant with public health research and practice (OMH 2001, p. 7). Standard 11, “Health care organizations should maintain a current demographic cultural, and epidemiological profile of the community as well as needs assessment,” would be taken to the next level were any type of population-wide mapping of genetic variants to take place. State level public health departments as well as the March of Dimes (under its Genetics Education Needs Evaluation (GENE) Project) have undertaken genetics needs assessments, efforts which deserve replication in different locales. Standard 12, recommending “participatory, collaborative partnerships with communities” to design and implement CLAS-related activities, has been applied in community-based participatory research on asthma, breast cancer, diabetes, high blood pressure, and environmental pollution, and has made strides in the genetics arena (e.g., with family history, genetics policy, and genetics education) involving communities of color. The standards go a long way towards establishing the preconditions for health while mitigating the fear that genetic advancements could further stratify communities and spawn injustices (Hodge 2004, p. 23).

## H. PROVISION OF GENETIC SERVICES

Public health genomics’ assurance function may be defined as “the development of public health genetic programs, evaluation of prevention effectiveness, and quality assurance” (Khoury and the Genetics Working Group 1996, p. 1718). Assurance applies to all groups, as evidenced by universal screening for sickle cell disease for all newborns in the United States (Ibid.; Wang and Watts 2007, p. 622). A 2007 survey of nineteen state genetics plans yielded a list of ten essential public health services, the three most programmatically common being birth defects surveillance and prevention, newborn screening (NBS), and clinical

genetics-related (Wang and Watts 2007, pp. 621–2). In the first category are activities like sponsoring / partnering in folic acid campaigns and sponsoring teratogen information services. The second involves sickle cell disease outreach efforts, partnering with universities and advocacy groups to provide services and referrals, and translation to community health care. The third category, newborn screening, includes the processing of specimens in state labs, often with the use of tandem mass spectrometry (TMS), expanding NBS rules and regulations, and providing assistance with dietary formulas. All of these duties will continue to be a part of public health practice regardless of technologic era.

The expansion of newborn screening options and attempt to build uniformity among states have posed a challenge ever since the advent of TMS on the newborn scene. The Secretary's Advisory Committee on Heritable Disorders in Newborn Children and individual state program reviews continue to take account of new evidence and drive towards evidentiary reform of existing panels as well as general convergence towards a uniform panel (ASTHO 2010, p. 48). The Newborn Screening Saves Lives Act of 2007 and state mandates promote follow-up care, including the provision of medically necessary foods for disorders identified through NBS. Long-term follow-up is needed to ensure quality care of individuals diagnosed with newborn screening conditions (Khoury 2011c, p. 207). A number of states (California, Connecticut, and Michigan) are connecting or have begun to connect neonatal and prenatal collections with the research enterprise, the public health analogue to larger international biobanks (Gwinn and Khoury 2006, pp. 22–4). These efforts call for public input to ensure buy-in from those individuals and groups who will eventually be impacted. The Michigan Neonatal Biobank, for example, has engaged the public through focus groups and ongoing community dialogues. Departments of public health and community health and the committees overseeing these biobanks will need to monitor them both for ethical compliance and evaluation of the types of users accessing biobank material.

Gaps in newborn screening services reflect those existing in other areas. Cost barriers to genetic testing can sometimes be solved by state-level action. Some states have mandated coverage of certain genetic tests, such as colorectal cancer screening for high-risk individuals (ASTHO 2010, p. 57). The problem of lack of appropriate billing and diagnostic codes for genetic tests and certain types of healthcare providers, such as genetic counselors, is perennial. Collaboration between health departments, genetic advocacy organizations, and policymakers is needed. Other problems, such as insufficient funding for programs or lack of leadership support occur at multiple levels, and must be addressed as such. Public health genetic services provision is known for its complexity. Solutions to shortages in well-trained providers of genetic services will neither be simple nor quick and

inexpensive. Public health departments will need to continue to identify funding sources and form constructive relationships with knowledgeable providers to ensure that qualified personnel and facilities are available to the public (ASTHO 2001, p. 7).

The cultural component is a very important part of genetic services. In public health, guaranteeing culturally appropriate services will often depend on active inclusion of money in grant programs for needed interpreters, or partnering with community-based organizations that offer needed capabilities. In Illinois, the Midwest Latino Health Research, Training and Policy Center has staff able to service both English- and Spanish-speaking clients, offers culturally competent genetics training to clinic and health department staff, and engages community health workers in its programs (ASTHO 2010, p. 61). Interestingly, the Midwest Latino Center partners with the Chicago Center for Jewish Genetic Disorders to promote family history awareness through DNA Day. This partnership is a splendid example of one non-profit organization linking with another NPO in a shared, non-conference-related activity.

A strong sense exists among both European and North American authorities that the healthcare landscape is about to change as a result of new genomic technologies moving in the direction of personalized medicine (Auffray et al. 2011; Brand et al. 2008). A team with the German Center for Public Health Genomics writes:

*On a conceptual level, public health and medicine seem to converge as the assessment and stratification of risks, which becomes essential for the individual and society at large. ... There is a potential for much more target-oriented and stratified prevention strategies finally replacing 'a one size fits all' approach." (Brand et al. 2008, pp. 8, 10)*

Some organizations, such as the Institute of Medicine Roundtable on Translating Genomic-Based Research for Health, are preparing for the advent of whole genome sequencing into clinical care (IOM 2010, p. 5). Public health provides a critical perspective on the pace of progress. Genome-based research and the introduction of predictive and diagnostic panels seem to be progressing rapidly in the area of cancer and infectious diseases (i.e., pathogen genomics), at an intermediate clip for cardiovascular diseases, and slowly for neurodegenerative disorders (Schulte 2011, p. 4). The release of genomic profiles has so far been at the behest of biotech corporations and health care plans reacting positively to their release. Public health will continue to have a responsibility to interpret to providers and the public which genomic profiles are ready for the mainstream.

Personalized genomic services and risk stratification will not come without a price. The German paper reminds readers of the potential for social inequalities if steps are not taken to avoid partial coverage of personalized technologies within two-tier

healthcare systems (Brand et al. 2008, p. 9). The World Health Organization report *Community Genetic Services* openly states that numerous unaddressed needs exist in low- and middle-income countries, including infectious diseases, antenatal care, labor and delivery, and newborn care (WHO 2010, p. 3). The flavor of the report departs from the vision of the first world countries. Policymakers need to keep context in mind when they contemplate the applicability of horizon technologies. At the same time, the evidence base being accumulated on genetic variants and tests needs to be comprehensive enough to pertain to the health circumstances of less well-to-do nations. The unique aspect of public health is that once more efficient means of risk stratification become available, population-based approaches would advocate for tailored prevention strategies based on vitamins, diet and lifestyle changes, which are within the reach of all (Khoury et al. 2007, p. 313; Gwinn and Khoury 2006, p. 24). Organizations like WHO and international networks such as the Genome-based Research and Population Health International Network (GRAPH Int.) and the Public Health Genomics European Network, and governments themselves need to continue to move forward in a spirit of collaboration (Burke et al. 2010, p. 789; Brand et al. 2008, pp. 6, 10; Office of Public Health Genomics, Institute for Public Health Genetics, Public Health Genetics Unit 2005, pp. 17–19).

Monitoring health and population surveillance, which fall under the category of “assessment,” have always been central to the public health mission. Following the consideration of cystic fibrosis as a candidate for population-wide screening (now integrated into many state newborn screening panels and offered in the form of carrier testing as part of obstetric care), attention turned to hereditary hemochromatosis as a possibility for population surveillance (Beskow et al. 2001, p. 6). The pluses and minuses of mass population screening using genetic testing to detect HFE mutations were hotly contested (Burke et al. 2000). The next evolution in surveillance was the possible merging of data from multiple across-the-board sources: vital records, census data, hospital discharge data, and other health-related information sources (Beskow et al. 2001, p. 6). This idea became a reality with the sudden cardiac death surveillance program in Michigan. Started in 2005, the program combines death records, autopsy results, medical records review, next of kin interviews, and investigation of the circumstances around individual deaths with expert panel review to suggest actions to prevent SCD of the young in future cases (ASTHO 2010, p. 21). The program is a good example of integrated surveillance with sufficient commitment from all involved to continue beyond the limited funding interval (CDC through 2008).

Bodies like the IOM Roundtable and GAPPNet have taken the concept of surveillance two steps further in advocating harnessing the power of bioinformatics. Both see it as a tool to make available to the clinical genetics workforce (IOM 2010, p. 4;

Khoury et al. 2009, p. 492). Several biorepositories in the U.S. (e.g., Mayo Clinic, Marshfield Clinic, Vanderbilt BioVU) are already engaged in the process of synthesizing medical record and biologic sample data into genotypic information that might be transmitted along with management recommendations to providers having patients fitting a certain phenotypic profile. The Scottish Health Informatics Program (SHIP) collects, manages, and disseminates clinical information on the country’s general population, and is engaged in pharmacovigilance using electronic patient records. It recently began research on biological samples from 10,000 patients with type 1 diabetes over age 16 to try to get at the genetic causes of the condition.

The Centers for Disease Control and Prevention’s BGD initiative would transcend the single disease limitation. OPHG envisions “a searchable, online information system of human genome variation ... that is readily accessible to researchers, health care providers and policy makers” (OPHG 2007, p. 4). For the BGD to achieve full applicability, it would need to overcome the strictly clinical emphasis of existent repositories to service the greater public health community. The difficulty inherent in such a project, or even the gradual accumulation of systematic data and reviews on categories of genetic variations, is infrastructure. “The absence of a comprehensive infrastructure raises substantive questions about how such primary data should be generated and collected, and further issues as to who should evaluate and analyze the data, and against which criteria or standards the utility of a test might be measure” (Burke et al. 2010, p. 786). The current public health infrastructure is arranged to handle newborn screening and disease surveillance, but not the envisioned onslaught of genetic variation information. Countries involved in the Bellagio Conference and beyond—the United States, Canada, and the European nations—stand in the same position with respect to the upcoming opportunities and challenges, and can exchange incremental solutions as systems evolve.

## I. EDUCATION AND TRAINING

Public health action falls along a continuum from mandates to awareness-raising. Education holds an important position in this spectrum. Ostensibly it can be divided into categories like professional education, workforce training, and public education. A 2003 Institute of Medicine committee identified genomics as one of the eight cross-cutting priority areas for graduate M.P.H. programs (IOM 2003, p. 17). The University of Michigan and University of Washington have for more than a decade maintained curricula in public health genetics, covering scientific-technical, ethical, and legal areas. The field continues to grow as Sarah Lawrence College started a certificate program in Public Health Genetics/Genomics in 2006. The mission with these programs does not end with fixed curricula, however. Two future directions are (Caumartin et al. 2001, p. 575):

- Incorporat(ing) genetic education into the curricula of all schools of public health, either as an elective or special program, or as a component of all departments.
- Educat(ing) the faculty of schools of public health about the implications of genetics for the disciplines in which they teach and carry out research.

Only a small portion of public health employees have benefited from these genetics graduate programs. A national survey of public health educators by Chen and Goodson indicated adequate understanding of applied topics, such as folic acid's role in preventing neural tube defects, but shortcomings in basic knowledge related to genetic testing and risk assessment (Chen and Goodson 2007). The CDC devised genomic workforce competencies in 2001 to address such deficits and orient public health workers towards including basic genetics knowledge and skills in their specific areas of practice (OPHG 2010b). The survey by Wang and Watts shows how the core competencies play out in actual duties public health practitioners undertake (Wang and Watts 2007; Goltz et al. 2010):

- Training health care personnel and community health workers to increase their awareness of genetics and genetic services.
- Providing information and sponsoring lectures for health professional and lay communities.
- Communicating the role of genetics and genomics in public health to policymakers and community stakeholders.
- Providing genetic and genomic information resources to hospitals, physicians' offices, and laboratories.

These activities constitute a subset of the areas outlined in the CDC competencies, which also indicate the importance to public health professionals of awareness of ethical, legal, social, and financial issues related to genetic testing.

Practitioners in a given area of public health practice will naturally depend more on one set of competencies than another. Some overlap exists, however. Among 140 responses by public health providers to a survey of twelve genomics competencies, the Secretary's Advisory Committee on Genetics Health, and Society found that providers marked conducting a family history, and utilizing a basic knowledge of the role of genetics in disease as their two areas of greatest confidence (SACGHS 2011, p. 40). The importance they placed on and confidence they expressed in use of family history is consonant with the value placed on it by public health. The basic knowledge ranking would be laudable except for Chen and Goodson's finding of shortcomings in basic knowledge related to genetic testing. Disparities of this type can be a cause for concern since they indicate possible overconfidence on the part of practitioners, and the need for continuing genetics education. State departments and schools of public health can collaborate in

training the public health workforce in genetics, either through real-time courses and workshops, or web-based courses.

The SACGHS report also dealt with barriers towards incorporation of genomics in professionals' practice. It indicated that education and training that focuses on basic genomic content, though needed, will not be enough. Training efforts should address how to apply genomic innovations in health promotion (Ibid., p. 20). It surmised that many public health professionals would need to see evidence of health benefit to feel compelled to incorporate genomics into their routine practice. In a sentence that strikingly connects translation research with education, the report stated: "Public health genetics will 'hit a translation roadblock if no investments are made in evaluating the best methods for assuring delivery and monitoring safety and effectiveness of gene-based interventions ...'" (Ibid., p. 21).

The community (lay) health worker (CHW) represents another category of public health practitioner. CHWs have been trained as a mainstay of public health programs targeting asthma, heart disease, and other chronic diseases within the community. Although probably just the basics of genetics need be conveyed, the fact that a number of states (Texas, Ohio, Indiana, and Alaska) require some level of certification means a certain standard of knowledge can be maintained (Ibid., p. 20).

Lay audiences have and will continue to benefit from public health practitioners' expertise in explaining down to earth aspects of genetic screening, dietary and lifestyle modification, and family history. Surveys have found the public to have limited knowledge of genetic risk factors. Studies have also reported that the public has generally low levels of genetic knowledge but positive attitudes towards genetic information, especially when it can be used to identify personal and familial disease risks (Goltz et al. 2010, p. 3). Public health practitioners and healthcare workers can address such needs.

The literature also describes newly arising areas which will compel both providers and consumers to become more genetically savvy: (1) the increased attention to common complex conditions as opposed to rare Mendelian disorders; (2) the rise of the Internet as a vehicle for patient and consumer learning; and (3) the existence of Direct-to-Consumer genetic testing and the potentials and challenges it poses for consumer-provider relationships (Bonham and Terry 2010, pp. 2–3; McBride et al. 2010, pp. 558–9).

K-12 genetics education appears in state genetics plans and is one of the recommended areas of continued attention in the SACGHS report. The report suggests collaboration between secondary schools, the Department of Education, and the National Science Foundation (SACGHS 2010, p. 3). Schools of public health can serve as the agent by which fundamental molecular ideas of genetics and disease, in paired down form, can be brought into the schools.

## J. PUBLIC ENGAGEMENT

Community genetics education often occupies some place in the consultation or dialogue process. Participants engaged in community consultations can also provide fresh perspectives on genetics-related programs that experts might not otherwise have considered. A WHO expert advisory group observed that geneticists "... have much to learn from support and advocacy groups representing those with genetic disorders" (Avard et al. 2008, p. 5). Given the opportunity to participate and make a difference, participants feel more trust in the decisions that result. International organizations recognize the benefits of involving the public in genetic policymaking. For example, in the context of pharmacogenomic research, the Human Genome Organization (HUGO) publicizes that researchers have an obligation to engage citizens "to earn the trust of the community" (Ibid., p. 3). A participant in the Quebec CARTaGENE biobank consultation project affirmed public concerns about confidentiality and misuse of biobank contents, and the possibility of public backlash if steps were not taken by the government to establish trust (Godard et al. 2007, p. 150; 2004, pp. 466–8).

The NIH defines community consultation as "a vehicle to listen to the community's interests and concerns, to address ethical issues, and to communicate information about the research to the community" (Godard et al. 2004, p. 459). Recurring characteristics of consultation in the literature include: (1) building trust and public confidence; (2) demonstrating transparency and openness; and (3) socio-cultural features, such as consultation leading to culturally appropriate policies, promoting solidarity and collaboration, and engendering a democratic spirit and equity (Avard et al. 2008, p. 3). Various typologies exist to classify the types of participatory activities that have been tried in the social and health arenas. Avard et al. proffer a relatively simple 2-part scheme based on (1) education, and (2) direct (two-way, e.g., dialogue, deliberative consensus conferences) and indirect (one-way, e.g., using a consultation or a survey) involvement (Ibid., p. 5). Burton et al. (2009, p. 17) and Rowe and Frewer (2005, p. 263) differentiate between exercises that merely inform, which they place at the bottom of the ladder, community consultation methodologies in the middle, and user-led or -owned processes at the top of the ladder. The solicitations for comment circulated by the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) and its forerunner, the SACGT, represent the middle category. The July 2000 report "Public Consultation on Oversight of Genetic Tests" is exemplary of the quality output that can be produced. Heavy public involvement in decision-making and the development of public health initiatives fall in the top rung ([http://oba.od.nih.gov/oba/sacgt/reports/oversight\\_report.pdf](http://oba.od.nih.gov/oba/sacgt/reports/oversight_report.pdf) and <http://oba.od.nih.gov/oba/sacgt/appendixB.pdf>). Two more extensive schemes in the public participation literature form a continuum from information provision; to consultation (unitary or segmented public

audience) and partnership (advisory boards, community dialogues); and finally delegation (commissions, public hearings) and control by the public (referenda) (Bishop and Davis 2002, p. 20; Lowndes et al. 2001, p. 207). Many variants exist, thus participatory genomic projects frequently start-out with focus groups on the way to more participatory engagement, and in Scotland, members of the public held a Citizens' Jury on Genetic Test Results and Life Insurance (Burton et al. 2009, p. 17).

The literature on public engagement in genomic policymaking has really picked up over the last decade. Here is a smattering of what has been done:

- **Participatory Policy Project on Prenatal Genetic Testing, the Netherlands** (Buning et al. 2008)
  - Participants: invited public, 8 affected by a condition, majority women 25–35 years
  - Methodologies: opinion polls, surveys, focus groups, forums, consensus conferences, citizen panels
- **Review of Stakeholder Involvement in Newborn Blood Spot Screening, United Kingdom, United States, Australia, and Canada** (Potter et al. 2008)
  - Participants: parents, members of patient associations, physician association members
  - Methodologies: workshops, organizational meetings, consultations, public forums, working groups
- **Asthma Genomics Community Consultation, Seattle, Washington** (Harrison et al. 2005)
  - Participants: asthma-related community organization members, researchers, healthcare providers, public health practitioners
  - Methodologies: group meetings, telephone and in-person key informant interviews
- **Biobanks Public Deliberation, British Columbia, Canada** (MacLean and Burgess 2010)
  - Participants: general population sample selected for diversity
  - Methodologies: small and large group deliberations

Deliberative processes in public health have in the past also devoted attention to working with particular racial-ethnic communities and faith groups, as well as community-based organizations (Bonham et al. 2009). Research in ethnically-identified communities can be preceded by community consultation (Licinio 2001, p. 85).

Community-based collaborations can themselves be evaluated. One review supported by the Health Resources and Services Administration (HRSA) argued that collaborative mechanisms

are not routinely feasible for program implementation purposes and to influence systems change (Kreuter et al. 2000, p. 61). It did reservedly note benefits in the areas of community awareness, participation, empowerment, and increased capacity to address health issues. While the theme of community empowerment is echoed in other literature (Bonham et al. 2009, p. 348), clear examples exist to counter the skepticism. The multi-state Communities of Color and Genetics Policy project led to further consultation of several of its members at national-level meetings and engendered a “Genetics Education Needs Evaluation” in two Michigan cities (Ibid., p. 349). The Michigan Governor’s Commission on Genetics Privacy and Progress held cross-state public forums that resulted in the inclusion of genetics and modification of existent language in an array of state legislation. In a completely different governmental example, Collabforge in Melbourne, Australia is conducting an online community consultation to connect the public with decision-makers for the design of “Future Melbourne” city initiatives (Koop 2010). The important point is that people feel they are being listened to and making a constructive difference, which increases public buy-in of projects as well as the quality of project outcomes.

“Future Melbourne” has one of the most user-informative and user-friendly wikis on the Internet ([www.futuremelbourne.com.au/wiki](http://www.futuremelbourne.com.au/wiki)). Some authors claim that web interaction and wikis represent the future of deliberative democracy (Noveck 2009). Example genomics-related wikis include:

- The Personal Genome Project on the Creative Commons

[http://wiki.creativecommons.org/Case\\_Studies/Personal\\_Genome\\_Project](http://wiki.creativecommons.org/Case_Studies/Personal_Genome_Project)  
<http://www.personalgenomes.org>

- The Science & Justice Working Group

<http://research.pbsci.ucsc.edu/scienceandjustice/blog/areas-of-inquiry>

- WikiAdvocacy created by the Genetic Alliance

<http://wikiadvocacy.org>

These wikis emulate the information knols belonging to GAPPNet, but are more freeform in their use. Solicitations for public comment and community consultations represent varieties of public participation that have allowed researchers and policymakers to gather the comments of and respond to a more heterogeneous public than organizational meetings and focus groups. They will continue to stock the armamentarium of important investigational and policymaking approaches. The shiny spot is that in this electronic era, the web can be used just as productively by health agencies and organizations as it can by DTC genetic testing companies to reach the public and move the genomics agenda forward.

## K. CONCLUSION

The field of public health genomics is multi-faceted in a number of ways: the genetically-related conditions its researchers study and its practitioners hope to prevent, the dual emphasis on scientific data collection and workable outcomes, and the focus on human welfare and broader concerns. Different parties will naturally have different priorities, but they may be convergent rather than divergent, and mesh together if the various stakeholders talk. The goal: “Thirty years from now, we will look back and be amazed that we can regenerate lost or injured body parts, that we can tailor health outcomes with individualized prescriptions, that we can eliminate preventable deaths from lung diseases caused by smoking, that we can prevent Alzheimer’s” (NIH 2010). Cross-cutting collaboration between agencies, departments, and sectors (public and private) is the key. If it serves its purpose, this literature review will provide a platform for discussion as the parties sharing in consultation envision and invent the future of public health genomics.

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## APPENDICES TO PART ONE

# STAKEHOLDER CONSULTATION APPENDIX 3: SUMMARIES OF KEY INFORMANT INTERVIEWS

## PUBLIC HEALTH SYSTEM

### Academia

#### Wylie Burke, MD, PhD

Professor and Chair

Department of Bioethics and Humanities

#### Summary

Public health genomics can be defined as the focus on genetics from a broad population health perspective. It is difficult to define public health genomics, here in the United States, when there is such a divide between public health and clinical medicine. Health is a continuum and genomics has a role in both public health and clinical medicine. Certain aspects of genomics fit nicely into the conventional model of public health, but public health genomics also has a role in looking at the evidence-based uses of genomics in clinical medicine.

Currently, the most fundamental part of genomics, translation, is underfunded. There continues to be very robust funding in the discovery phase of the research cycle. Very little research money goes towards delivery and outcomes. There continues to be an assumption that after discovery, development will follow, but we often do not understand how to translate innovation; more money is needed for technology assessments. For example, we do not study the outcomes of newborn screening. We do not know how false positives affect people. The recent cut in funding to the Office of Public Health Genomics is a bad sign about the future of research trajectories. It really speaks to the divide of genomics into clinical applications, public health, and research. NIH will be faced with huge amounts of genomic information in the future; in the past, CDC played the role of filling the conceptual gaps needed for translation. The National Center for Advancing Translation Sciences at NIH is focusing on drug discovery. This supports the development of products and does not focus on improving health. Drugs are not going to solve health disparities, and we are missing huge opportunities to think creatively about the use of genomics.

As public health genomics moves forward, no discipline can be off the table; we must cut through silos and address issues from many angles. The OPHG has a lot of convening power, and that should be taken advantage of. The discussions do not have to revolve around what we need funding for, but rather, how to

use the knowledge we have to improve health outcomes. Social determinants of health need to be discussed, as health disparities are not addressed enough in public health genomics. There seems to be a disconnect in the definition of public health genomics between genomics and epigenetics. The community needs to be engaged as solutions to health problems are discussed. For example, discussions can revolve around genomics and diabetes and the relationship between family history and environment. From this discussion we can begin to move research into practice and focus on health outcomes. Public health genomics has a real role in understanding outcomes. This is a challenge for public health genomics because genetic susceptibility does not predict disease outcomes, it merely predicts susceptibility.

The OPHG also has a role in informing policy through the use of surveillance data. There is also a lot of buzz around genome and exome sequencing. The OPHG can be involved in health technology assessments and the development of guidelines for genetic technologies. Additionally, the office can make suggestions for the overall research agenda. Questions about what molecular tools have a place in public health have to be asked. For example, genetic susceptibility may help us define environmental exposures and set guidelines for occupational exposures. From there we can begin to think about how to develop standards based on groups of people and their susceptibilities. This will require partnerships and collaborations between individuals and both hard and soft sciences.

#### Priorities

1. Application of evolving technologies in newborn screening
2. Application of genetic technologies in healthcare; ensure responsible evaluation and guidelines
3. Use convening power of CDC to promote conversations about translation, paying careful attention to those topics that are transdisciplinary

#### Kimberly Kaphingst, Sc.D.

Assistant Professor

Washington University in St. Louis

#### Summary

Public health genomics is the use of genomic information to advance the health of individuals and communities. Public health genomics distinguishes itself from clinical genetics with its focus on underserved and disadvantaged communities. The term “public health genomics” is relatively new to those in public health practice; however, there is a definite increase in individuals entering the field of public health genomics, especially from cancer research. Still, public health does not fully recognize

genomics as a relevant part of public health, or how quickly the field is growing.

Funding is one barrier to furthering the integration of genomics into public health priorities, as it is difficult for academics to find public health genomics research grants. There is a tension between the public health folks and those in clinical medicine as to where research dollars belong. Genomics does not have to be completely separate; it can be integrated into chronic disease prevention programs, particularly for cancer and diabetes. Genomics competencies must be integrated into schools of public health so that academics and practitioners know how to deal with this information. Family history is one way to integrate genomics into other areas of public health, although more research is needed to determine how people understand and deal with genetic susceptibility information. There is a perception that genetic susceptibility information does not change behavior; however, there is little evidence to support this.

Community engagement efforts, in collaboration with academics and public health practitioners, are necessary to bridge communication gaps and further genomics education. Funding is a barrier here, and grant money is necessary to fund such initiatives. Community partners, prevention practitioners from state health departments, genetic counselors, and communications researchers need to be at the table to push community based participatory research agendas forward. This will bridge the different areas of public health and further the integration of genomics into population health programs.

Policies that support research on genomic technologies are essential. Groups, such as EGAPP, are needed to make recommendations and develop guidelines to be integrated into public health practice. Also, direct-to-consumer genetic tests must be regulated to ensure that consumers are getting accurate information about their genetic susceptibilities. Finally, state health departments need the funds for individuals who specialize in genomics to participate in disease prevention and control programs. If health departments do not stay on top of the fast-paced growth in genomic technologies, public health will fall behind.

#### Priorities:

1. Stay ahead of genomic technology advancements
2. Integrate family history with disease prevention and control programs
3. Research gene-environment interactions
4. Make genomic information understandable and acceptable for communities

#### Chris Kuzawa, PhD

Associate Professor  
Department of Anthropology

Northwestern University

#### Summary

Public health genomics is using genetic information to impact population health, health disparities, and policy. Genomics has been focusing on genetic variation, such as SNPs, but this has not been very productive. The Human Genome Project provided us with a blueprint of human biology. However, just knowing a nucleotide sequence does not tell much about complex disease; nucleotide sequences only provide a clear link between genotype and phenotype for Mendelian diseases. Conditions like obesity do not have this clear genotype-phenotype relationship. In the case of obesity, gene-environment interactions become very important. We have already invested in GWAS and we can only explain about 1–2% of the variance. Complex conditions involve multiple pathways, and it is uncertain what role public health genomics can play in affecting these complex pathways.

While schools of public health should be teaching genetics, we need to be realistic and proceed with caution. The complexity of genomics must be taught, and we must stop looking at big integrated phenotypes, and instead look at problems that involve one specific pathway. For example, cholesterol is something where genetics can play a role. In addition, it is a simple phenotype and drugs work to lower cholesterol. Obesity is a phenotype that is too far removed from a simple molecule.

Genomics should focus on those areas with greatest marginal utility. To determine where the greatest effects will be, we need justifications. Anthropology truly connects soft science with hard science. Schools of public health can use the many different approaches to strengthen evidence for public health genomics.

Dr. Kuzawa is a part of a longitudinal study in the Philippines that examines fetal development. This study allows for a retrospective analysis of the mother's exposures throughout her life. We do not know how to predict birth weight in the short term, as prenatal supplements have shown a minimal effect on birth weight, but examining the environment allows a look at the long term predictors of birth weight. The research team is comprised of individuals who are experts in health disparities, social epidemiology, medical anthropology, and epigenetics.

Public health genomics must shift from an isolated field to a field that is integrated into all public health disciplines. It is too great of a leap to go from public health to the nucleotides; all the social determinants of health that happen in between genetics and health outcomes need to be further examined.

**Priorities**

1. Shift the focus from molecular biology to phenotypes and phenotypic responses to environment (across generations)
2. Focus on epigenetic mechanisms

**Kenneth Olden, PhD**

Dean of CUNY School of Public Health  
Hunter College  
August 4, 2011

**Summary**

The definition of public health genomics represents an effort to improve human health by developing more effective personalized medicine based on one's genetic profile. This perspective is only a part of the story, however. The scope of public health genomics is too narrow; this approach alone will not have a huge impact on population health largely because Americans will not be able to afford a personalized medicine approach. There is a need for a population-based approach to genomics.

Environment alone isn't the answer either. In order to identify environmental health risk factors, one really needs to understand variations in gene frequencies. Most importantly, there is a need to integrate genetics with epigenetics. A combination of genetics and the environment together is going to be the risk that is largely responsible for chronic disease. We can use genetics to identify at-risk populations and also identify differences in environmental factors (i.e. East Harlem vs. downtown New York City). There is a need for a concept that recognizes the importance of all determinants (genetics, social behaviors, environment, etc.) because it's the interaction that will be crucial to examine. It's really a combination of the total environment and total genetic makeup.

An important step to explore all determinants of gene-environment interactions would be to develop a database cataloguing genetics, physical and social environment, and determinants of health (diet, exercise, etc.). A transdisciplinary team built of scientists, public health professionals, and social and behavioral scientists will be needed in order to carry out this sort of database. Currently, every field is in its own silo (environmental health, social sciences, genetics, epigenetics); all of which are conducting isolated research. This is difficult because these groups of researchers often don't speak the same language. The community also needs to be a stakeholder. There is a need for research incentives to facilitate these partnerships and for grants to include community participation. Original research with bold models is what will move this field forward.

**Priorities**

1. Shift research focus to study epigenetics/gene-environment interactions; Use genetics to identify high risk populations and then examine the environment and exposures.
2. Build a transdisciplinary approach exploring all determinants of disease (environmental, genetics, social and behavioral, epigenetics, etc.) and develop database cataloguing all of these determinants.

**Community****Chikezie Maduka**

University of Maryland, Prevention Research Center

**Summary**

Genomics is a field that touches on all aspects of public health, especially when relating chronic disease and family history. Health history is one way of looking at almost all health issues, as the better people can understand their past, the better than can prepare for their future.

Communities must be engaged in public health genomics, and the first step is to provide "genomics 101." Collaborations and partnerships must be made with academics and the community to foster trust in the communication. This is a win-win for researchers and community members, as the researcher can conduct a study and community members learn about the research topic. Genomics should be integrated into all the projects at Prevention Research Centers; it does not matter if the focus is explicitly genetics. Family history is a great way to encourage the integration of genomics into public health research. This is particularly important for teenagers, as they have the chance to ask their parents and their grandparents about their health histories. This information can be given to a physician and used to prevent disease onset or progression. Some communities will be resistant to discussing health history; however, any progress is progress. This process is worth the investment, as trust must be built as genomics moves forward.

Beyond communities, politicians need to be educated about genomics so that funding and ethical issues can be addressed at the national level.

## Public Health Practice

### Jean Chabut, MPH

Chief Administrative Officer

Michigan Department of Community Health

#### Summary

Public health genomics is hard to define. It was thought that the Human Genome Project would bring miracle cures and public health applications, but that has not happened yet. Public health genomics should be centered on the interactions between genes and environments. Because there is still so much unknown, public health needs to stay on top of the latest in genomics so that it does not fall behind. Chronic disease folk and the newborn screening folk of state and local health departments need to collaborate and keep each other informed. States need to work with each other to share successful tools so that public health genomics will move forward and not fall behind.

Michigan has done a lot of work to bridge the different areas of the public health department. Janice Bach has been a key player in ensuring that chronic disease people sit down and educate the newborn screening and laboratory people and vice versa, even without the funding to do so. This keeps genomics on the mind, and facilitates the integration into various public health programs. For example, family history was added to the WISE-woman program on breast and ovarian cancer, and it is looked at to prevent sudden cardiac death. All areas of public health should be engaged in genomics. The CDC and [CDC Director] Dr. Thomas Frieden must do a better job of addressing public health genomics.

The CDC Office of Public Health Genomics should look at the Healthy People 2020 goals and see what opportunities there are for genomics. Cancer is a genetic disease, and often people seek genetic counseling. However, there are barriers to accessing genetic counselors as they are not licensed in most states and many insurance companies do not cover their services.

Michigan has been creative in using genomic information. The biobank of stored blood spots, dating back to 1984, can be used to produce DNA and RNA samples to be used for research. Birth certificates provide information about the mother's address and the child's place of birth. This information can be used to tie together genetics and environment and used to understand health disparities. Databases can be created that keep individuals anonymous but apply information to public health issues. Collaborations with academics will be necessary to translate the research that comes out of such endeavors.

Universities have a role in public health genomics. Genomics education across the multiple disciplines in public health is necessary and curricula must be built to ensure genomic competencies for the public health workforce. Universities must also

help public health practice translate research findings into usable tools. Cancer centers are an easy place to begin these discussions about genomics, but they often think singularly about genomics. Genomics needs to be discussed and integrated across schools and centers within schools of public health. Discussion groups should be formed that cut across the public health silos. People from chronic disease prevention, maternal and child health, epidemiology and labs, and genetics need to determine what is and what is not ready for integration into public health practice. The Prevention Research Centers, placed within academic institutions, are a great place to develop genomics discussion groups.

Genetic interests have historically been tied to maternal child health in dealing with single gene disorders. As public health genomics moves forward, chronic disease experts must be engaged. An understanding of gene and environment interactions will help bridge the divide between clinical medicine and public health as we begin to tackle the chronic disease epidemic in the United States. Funding must shift back into public health genomics to make these goals realities.

#### Priorities

1. Get the attention of CDC and Dr. Frieden and prioritize genomics
2. Provide funding for demonstration projects
3. Focus on low hanging fruit, including the use of family health history to motivate people to be healthier
4. Use surveillance and BRFSS to get information, and integrate genomic information

### Dr. Maxine Hayes

State Health Officer

Washington State Department of Public Health

#### Summary

Public health genomics is using the information we gained from the Human Genome Project to improve population health and healthcare. A lot was learned about human biology through the Human Genome Project; however public health genomics has made little progress with that information. We need to reach a consensus, across states, as how to integrate genomics into public health programs.

In the state of Washington, the health department is working on several genetics projects. The health department contracts with genetics clinics to ensure that those who need to see genetic counselors have access to such services. The state works with Medicaid to ensure that genetic services are adequately covered. Genetic counselors receive licensure in the state of Washington,

which also increases access to genetic services. Family history is being incorporated into chronic disease prevention programs, including many cancer programs, as family history is a low-cost tool that individuals can relate to.

The funding environment limits further initiatives in Washington and across the country. Collaboration and partnerships within and across health departments are essential for integrating genomics into public health. Newborn screening has been so successful because we are always willing to fund maternal and child health programs, and, in addition, all states are doing newborn screening. There are no population based services like newborn screening that all states are doing. Family history is something that is easy to relate to and can be incorporated into many public health programs. In this environment, we have to be careful what we prioritize, and family history is an inexpensive tool to implement, and it has benefits. Specifically, family history should be integrated into chronic disease prevention, and electronic medical records. Health departments have to stay on top of technology developments; Texts4Baby, a twitter feed, is an example of using technology to provide health information.

In order to sustain genomics in public health, partnerships must be created with academics and the workforce. Grants are one way to encourage these collaborations. Health departments, and the Office of Public Health Genomics, need money to fund individuals to work at developing partnerships across the public health system. Partnerships require work and time. Further, genomics must be integrated into all subject areas in schools of public health and medical schools. The public health workforce needs to understand the big picture of what genomics can accomplish, along with the details. Providers need a thorough understanding of genomics and genetic risk factors so that they can appropriately refer patients for genetic services. With the expansion of coverage through the Affordable Care Act, we need to make sure that providers are using genetic technologies wisely. To achieve this we need groups to evaluate genetic tools, providers to be educated, and genetic services to be covered by insurance.

#### Priorities

1. Consensus definition of public health genomics
2. Adequate capacity to support genomics activities
3. Genomics needs to be integrated with all other areas of public health
4. Genomics research needs to be translated into guidelines for practice
5. Family health history must be incorporated into chronic disease prevention

#### Steven Teutsch, MD, MPH

Chief Science Officer

Los Angeles Department of Health

#### Summary

Public health genomics is the application of genomics to improve population health. The county of Los Angeles is doing very little in public health genomics; in fact, beyond newborn screening there is not much else going on. Public health genomics is looking for issues to solve in public health, but the technologies are not ready for prime time. There is a lot of research on bacteria and virus genomics that is more helpful for understanding epidemics than human genomics research is.

Genomics has been receiving a lot of hype; however, there have been few real wins in the field. There is not significant evidence that investing in genomics is valuable to public health. The focus of public health needs to be on social and environmental factors. There are so many determinants of health, including education, jobs, and the built environment, and little to support that focusing on genomics will improve health.

Genomics is still in the research phase and not ready for practice. There is not enough evidence for using genomics to predict chronic disease. Even for the small number of people that genetic testing does benefit, for example in the case of Lynch syndrome, there are not enough cost savings to implement testing at the population level. There is a lot of interesting work being done on family history. Still, the consensus is that more research and evidence is needed before policies can be put into public health practice.

The CDC Office of Public Health Genomics has a place in this field; however, given the funding environment, its role must shift to pushing the appropriate agendas. Most of the genomics information is not ready for CDC yet, and genomics needs further investigation at the NIH level. Groups such as EGAPP at the CDC had very limited success. People do not listen to EGAPP, they listen to USPSTF. EGAPP did not find technologies that were worth evaluating, and questions could not be answered quickly enough and were rather complicated. EGAPP could not provide simple recommendations given the harms of false positives and false negatives from genetic tests. Beyond these challenges, EGAPP was successful in getting all stakeholders at the table and willing to share the stories that need to be told.

As public health genomics moves forward, we need to remain skeptical. As resources are limited for public health, genetic technologies must be carefully evaluated to ensure that there is a significant net benefit before releasing technologies into practice.

**Priorities**

1. Honest stories about the state of genomics for improving health
2. Evaluation of genetic technologies for the incremental cost-effectives

**Deborah Klein Walker, EdD**

Vice President, Abt Associates

Former President, APHA

**Summary**

The Association of State and Territorial Health Officials (ASTHO) has a good definition of public health genomics. ASTHO defines public health genomics as “a multidisciplinary field focused on the effective and responsible translation of human genome-based information and applications 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.” Public health genomics takes a population level approach; however many people still think of public health genomics in clinical terms.

Public health genomics must embrace the ten essential functions of public health (monitor, diagnose and investigate, inform, educate, and empower, mobilize community partnerships, develop policies and plans, enforce laws and regulations, link people to health services, assure competent workforce, evaluate, and research). The public must be educated about public health genomics, and this can start with family health history. The legal issues are beginning to be resolved through the implementation of the Genetic Information Nondiscrimination Act, but more work is to be done. Additionally, sustainable funds must be provided to state to develop the infrastructures needed to incorporate genomics into health departments.

Schools of public health can be leaders in the field, as their multidisciplinary researchers can approach genomics from a wide range of perspectives. Schools of public health should also work with their state health departments to address gaps in public health genomics. A lot of schools focus on international health, as it is popular among students; however, even the WHO is looking at genomics to address the burden of chronic disease.

Muin Khoury is a great leader who encourages this multidisciplinary approach and we must make sure that academia and practitioners do not fall back into their silos. A systems approach is necessary to improve health, and genomics can be applied to the core principles of public health. The CDC Office of Public Health Genomics needs to use its convening power to bring together diverse stakeholders to discuss future goals and priorities for the field. States that have been successful in maintaining public health genomics programs have effective communication between the maternal and child health and the chronic disease individuals. State public health departments must convene groups that include the laboratory/epidemiology, legal, maternal and child health, and chronic disease experts to integrate public health genomics into existing programs. Funding is barrier to such efforts.

Public health genomics truly needs a group of advocates to fight for what progress has been made and ensure that further budget cuts do not happen. The APHA Genomics Forum may be one group of interdisciplinary individuals who can really push the agenda at the federal level to keep public health genomics moving forward. While, genomics has not lived entirely up to its promise, we must continue to articulate where it can go and how it can improve population health going forward.

**Priorities**

1. Integrated national approach executed at the state level
2. Public health genomics must remain on the radar
3. Fight to keep what we have now

## APPENDICES TO PART ONE

# STAKEHOLDER CONSULTATION APPENDIX 4: REPORT OF KEY INFORMANT INTERVIEWS

## NONPROFIT AND FOR-PROFIT ORGANIZATIONS

### Introduction

Since 1997, the CDC Office of Public Health Genomics (OPHG) has worked to develop the public health genomics enterprise, engaging many partners to anticipate, effect, and evaluate the translation of genome discoveries for public health impact. From the beginning, this strategy has been comprised of two complementary approaches: bringing a population perspective to genomic research, and translating genomic research findings for public health benefit. Certainly, epidemiological studies and methodology have become more prominent in genomic research during the last decade. At the same time, demonstrating an evidence-based approach to evaluating genetic tests has helped establish a societal perspective on their rational integration into health care and disease prevention.

At a time when the entire public health community is faced with shifting funding and changing focus, the OPHG has planned a meeting to help reevaluate and prioritize near-term and longer-term objectives in public health genomics. At this meeting, the OPHG plans to engage stakeholders from federal, state, academic, industry, consumer, and professional organizations in a facilitated discussion of how the changing environment will affect priorities, goals, and strategies for public health genomics in the next five years and how enhanced partnership can help to advance the field. The expected outcome of this meeting is a list of short-term actions and coordinated approaches by various stakeholders to leverage and guide the use of genomics to improve population health outcomes.

In addition to performing an internal needs assessment, the OPHG has commissioned the Center for Public Health and Community Genomics at the University of Michigan and Genetic Alliance to gather perspectives from experts and leaders in many external stakeholder communities regarding the future of public health genomics and the role that CDC might play in supporting effective translation. University of Michigan has engaged key informants in the academic and public health sector, while Genetic Alliance has gathered input through a series of interviews with individuals from the private and non-profit sectors, with a focus on leaders of public-private partnerships.

In the following report, we detail the findings from the for-profit and non-profit sector, including a profile of our eight key

informants. In addition, we identify the overarching themes that emerged in the four topic areas that will be discussed at the meeting: Prevention, Detection, Development & Evaluation, and Pathways & Interactions.

### Summary of Results

The for-profit and non-profit sectors provide an interesting viewpoint into the public health system. While many of our informants viewed themselves as working on public health, they shied away from traditional definitions. In line with the IOM's 2002 report entitled *The Future of the Public's Health in the 21st Century*, key informants frequently cited intersectorial partnerships, evidence-based decision-making, using multiple determinants of health, and better information sharing and communication in the health system as components of a modern public health system.

Overall, two priorities for the field of public health genomics emerged from Genetic Alliance's stakeholder engagement interviews. First, our key informants identified a need to improve our understanding of the molecular drivers of common, complex disorders as a top priority for the field of public health genomics in the next five years. Informants expressed the opinion that enhancing our understanding of the molecular basis of disease can significantly improve public health by enhancing our ability to identify at-risk individuals, predict the natural course of disease, and stratify subgroups of individual most likely to benefit from intervention. Secondly, our key informants consistently identified the need to improve the evidence base for emerging genomic applications, including both our ability to develop evidence in a cost-effective manner and to increase our capacity to perform evidentiary reviews.

To improve our understanding of the molecular drivers underlying common, complex diseases, our informants identified a number of issues that need to be addressed in the coming five years, including infrastructure, data sharing, community engagement, education, and integration. In particular, they identified data sharing as a paramount issue in advancing our understanding of the molecular basis of disease. Creating large, widely accessible genotype-phenotype datasets that include information on a diverse population, building the infrastructure needed to house and facilitate the seamless distribution of large genomic datasets, and increasing consumer awareness of the importance of data sharing are three issues that our informants felt would be important to address to promote broader data sharing.

Informants also identified a number of priority issues related to improving evidence base for emerging genomic applications. Several informants expressed the opinion that the discussion around what constitutes an acceptable level of evidence for moving new genomic applications into a clinical setting was one that needs to continue. Informants also felt that there is a need

to continue to discuss and disseminate models for incremental evidence development and integration strategies for dealing with genomic applications with less than comprehensive evidence.

In addition to priority issues, informants also recommended metrics to track progress over the next five years. To measure health outcomes, informants felt that it will be important to continue to develop surrogate endpoints and factors outside the traditional sphere of clinical outcomes, such as personal utility or improved adherence to treatment. In terms of evidence development, informants highlighted the need for a method of capturing incremental improvement, in addition to more standard measures related to the number of tests with sufficient evidence.

Lastly, our informants felt that it is important to track progress in two additional areas: 1) The number of new diagnostic devices brought to market and the economic ramifications of these devices; and 2) Workforce education and training in genetics.

While participants expressed concerns regarding the fate of existing programs such as EGAPP and GAPPNet, there was an overall sense of optimism that the next five years will bring tremendous growth in our understanding of genetics and an increasing host of clinical applications that utilize this knowledge to improve public health.

## PROFILE OF KEY INFORMANTS

Respondent	Sector	Organization	Position
R1	Non-profit	Health education society	CEO
R2	Non-profit	Disease-specific advocacy organization	CEO
R3	Non-profit	Bioinformatics research organization	President
R4	Non-profit	Disease-specific advocacy organization	VP of Research Programs
R5	For-profit	Health benefits company	Senior medical director
R6	For-profit	Diagnostic company	Technical Director, Molecular Genetics
R7	For-profit	Health benefits company, technology evaluation center	Executive Director
R8	For-profit	Molecular diagnostic company	Executive Chairman

## SUMMARY OF KEY INFORMANTS RESPONSES

### Q1. What are the most important public health priorities are for the next five years?

Respondent	Summary of Response
R1	<ul style="list-style-type: none"> <li>• Tackle diseases with the widest prevalence in the United States• Implement mechanisms to tackle these diseases at the systems level</li> <li>• Identify where interventions can be targeted to individuals most likely to benefit from treatment</li> <li>• Maintain existing public health infrastructure in a time of restricted funding</li> </ul>
R2	<ul style="list-style-type: none"> <li>• Build an infrastructure capable of collecting and storing genomic data</li> <li>• Collect data from existing infrastructures such as electronic medical records</li> <li>• Institute new policies that examine how data is collected and accessed</li> </ul>
R3	<ul style="list-style-type: none"> <li>• Decode the common diseases that have the greatest impact on public health</li> <li>• Spend more time and attention understanding the molecular drivers of disease</li> <li>• Improve our ability to accurately predict response to treatment</li> </ul>
R4	<ul style="list-style-type: none"> <li>• Promote collaboration and data sharing</li> <li>• Enhance existing data sharing policies to reduce the lag phase between when the data is generated and when it is shared after publication</li> <li>• Collect data from broader populations, not just the narrowly defined populations used in clinical trials</li> <li>• Improve the accessibility of large datasets</li> <li>• Develop predictive models of disease progression using genomic information</li> <li>• Better educate consumers</li> </ul>
R5	<ul style="list-style-type: none"> <li>• The public health priorities for the coming five years will not be different from the public health priorities for the past five years</li> <li>• Evaluate the evidence on technologies coming into clinical practice and establish a basis for what constitutes 'adequate' evidence on emerging technologies</li> <li>• Ensure health care providers have the education and training needed to interpret the results of complex genomic tests</li> <li>• Evaluate the cost impact of emerging technologies and the value that they bring to the health care system</li> </ul>
R6	<ul style="list-style-type: none"> <li>• Interpret the results of whole genome sequencing, a technology that will be increasingly accessible in the next five years</li> <li>• Identify subpopulations of individuals that will benefit from therapeutic intervention</li> <li>• Improve detection of individuals at-risk for a diseases and those who are beginning to show early stage clinical manifestations</li> </ul>
R7	<ul style="list-style-type: none"> <li>• Focus on common, complex disorders: cardiovascular disease, diabetes, obesity, and oncology</li> <li>• Learn more about how to use genomic information to assess risk and what individuals who are identified as 'at-risk' can do towards prevention</li> <li>• Figure out how we can use whole genome sequencing information to improve health and what actionable steps we can take with this information</li> </ul>
R8	<ul style="list-style-type: none"> <li>• Shift our investments from strictly therapeutic based research to include more research on diagnostics</li> <li>• Address the problem of overutilization of therapies that are not working in a large segment of affected individuals</li> <li>• Invest in pharmacogenomics</li> </ul>

**Q2. What are programs and activities that address these priorities?**

Respondent	Summary of Response
R1	<ul style="list-style-type: none"> <li>• Establish the evidence base for emerging technologies</li> <li>• Fund programs for complex, evidence-based research on the effectiveness of different genomic applications within health care or public health system</li> <li>• Support research beyond the basic science discovery phase</li> </ul>
R2	<ul style="list-style-type: none"> <li>• Enlarge the precompetitive space and the number of individuals who can access genomic databases</li> <li>• Write legislation to help enable data sharing</li> <li>• Develop programs that will allow individuals to store their own health data and control who has access to it</li> <li>• Promote federal activity and grass roots efforts to address the needed activities and programs</li> </ul>
R3	<ul style="list-style-type: none"> <li>• Activate citizen scientists through national programs and local activities</li> <li>• Develop national programs to make the issue of citizen control of data a prominent issue and institute regulations that will help, rather than hinder, data generation</li> <li>• Create mechanisms for patients to control where their health data lives and provides them with the ability to share it with relevant studies</li> </ul>
R4	<ul style="list-style-type: none"> <li>• Implement programs and policies that promote broader data sharing, particularly from agencies that fund genomics research</li> <li>• Broaden interagency collaboration within the federal government</li> <li>• Increase government leadership in establishing large collaborative studies</li> </ul>
R5	<ul style="list-style-type: none"> <li>• Increase our capacity to evaluate emerging genetic technologies by better engaging a broader range of stakeholder groups, such as professional societies, and putting existing evaluation systems into 'overdrive'</li> <li>• Develop new business models for bringing diagnostics to market in a cost-effective manner</li> <li>• Evaluate the value of new technologies using traditional methods for evaluating cost effectiveness</li> </ul>
R6	<ul style="list-style-type: none"> <li>• Address the rigidity in our current system around what constitutes sufficient evidence for reimbursement</li> <li>• Establish policies for dealing with technologies that do not meet the current evidentiary thresholds and strategies for incremental evidence development tied to reimbursement</li> <li>• Decrease our focus on infectious disease and increasing the investment in genetics</li> </ul>
R7	<ul style="list-style-type: none"> <li>• Implement broad public education about genetics in general and whole genome sequencing in particular</li> <li>• Support studies that demonstrate utility of existing tests in a real world context, not randomized control trials</li> <li>• Move away from randomized control trials to assess effectiveness and value of emerging technologies because they are ill-suited to the task of measuring what will be impactful in a real world setting</li> </ul>
R8	<ul style="list-style-type: none"> <li>• Increase the larger community's recognition of the value of accurate diagnostics</li> <li>• Invest more money and time in developing diagnostic devices</li> <li>• Reduce overutilization of ineffective treatments</li> </ul>

**Q3. What are the measures of health impact used to track progress in addressing public health priorities?**

Respondent	Summary of Response
R1	<ul style="list-style-type: none"> <li>• Develop short term, surrogate outcomes in addition to traditional outcome measures such as reduction of morbidity and mortality</li> <li>• Create metrics that take into account less traditional outcome measures, such as personal utility and adherence to intervention</li> </ul>
R2	<ul style="list-style-type: none"> <li>• Measure the number of individuals that can store and control access to their electronic health information</li> <li>• Track the number of integrated datasets that are available</li> </ul>
R3	<ul style="list-style-type: none"> <li>• Measure our ability to collect information about how individual variation affects response to therapy</li> <li>• Measure the number of mechanisms available for collecting patient-reported outcomes in addition to physician-entered data</li> </ul>
R4	<ul style="list-style-type: none"> <li>• Establish a baseline of the proportion of individuals in the general public that are aware of the diseases they are at-risk for or beginning to show symptoms of the early stages</li> <li>• Continue to monitor detection and the number of individuals who are being diagnosed during the early phases of the disease</li> <li>• Establish metrics around the amount of data that is being shared</li> </ul>
R5	<ul style="list-style-type: none"> <li>• Count the number of new evidence reviews and the number of agencies performing evidentiary reviews</li> <li>• Measure the number of medical schools with robust genetics curriculums</li> <li>• Use self-rating tests to quantify practicing physician’s knowledge and competency in genetics</li> <li>• Measure the consistency of how well-validated tests are used in a clinical setting</li> <li>• Use health economics to evaluate the return-on-investment for novel diagnostics, including data on cost savings from reduced medical complications</li> </ul>
R6	<ul style="list-style-type: none"> <li>• Measure incremental improvements in evidence development and be prepared to deal with situations where there is less than full evidence</li> </ul>
R7	<ul style="list-style-type: none"> <li>• Make sure that we measure things that are meaningful and impactful to the individuals rather than more traditional metrics that are used in academic settings</li> </ul>
R8	<ul style="list-style-type: none"> <li>• Measure the number of new prognostic and predictive devices that come to market</li> <li>• Use health economics to understand how much money improved diagnostics can save by preventing ineffective treatments from being administered</li> </ul>

**Q4. What are the most important advances in genomics that can potentially impact public health priorities?**

Respondent	Summary of Response
R1	<ul style="list-style-type: none"> <li>• Genomics as another tool for identifying at-risk individuals and subgroups of affected individuals that will benefit from intervention</li> </ul>
R2	<ul style="list-style-type: none"> <li>• Genotype – phenotype correlation studies</li> <li>• Our ability to identify genetic modifiers, individuals at risk, and make predictions about response to treatment based on genomic information</li> </ul>
R3	<ul style="list-style-type: none"> <li>• Integrating genomics into public health will be critical to move us from a system-based characterization of disease to a molecular-based classification of disease</li> <li>• Genomics will be critical for identifying sub-groups of affected individuals that will benefit from treatment from those who will not benefit</li> </ul>
R4	<ul style="list-style-type: none"> <li>• Our ability to identify and understand extreme phenotypes and make the findings more generalizable to larger populations</li> <li>• Systems biology approach and our ability to integrate genomic data with metabolomics and proteomic data</li> </ul>
R5	<ul style="list-style-type: none"> <li>• Decreasing cost and increasing efficacy of gene chips</li> <li>• The rise of commercial groups that will evaluate and make recommendations on new genomic technologies</li> </ul>
R6	<ul style="list-style-type: none"> <li>• Our rapidly growing understanding of the human genome</li> </ul>
R7	<ul style="list-style-type: none"> <li>• Whole genome sequencing and the potential to have all consumers sequenced as the cost of the technology continues to decline</li> <li>• Advances in proteomics and the decreasing cost of performing proteomic analyses</li> </ul>
R8	<ul style="list-style-type: none"> <li>• The decreasing cost of sequencing</li> </ul>

**Q5. Is genomics and family history currently integrated into programs and activities that address public health priorities?**

Respondent	Summary of Response
R1	<p>No, we need to:</p> <ul style="list-style-type: none"> <li>• Better disseminate intervention strategies with proven success</li> <li>• Implement more demonstration projects to determine the best integration strategies</li> <li>• Create 'plug and play' programs that public health officials can easily integrate into existing infrastructure</li> <li>• Fund the clinical training needed for implementation of genomics and family history</li> </ul>
R2	<p>No, not even in rare, Mendelian inherited disorders. We need to:</p> <ul style="list-style-type: none"> <li>• Rethink our current method of selecting pilot projects—rather than rewarding the best grant writers, we should be selecting pilot sites that present the best opportunity for integration</li> </ul>

Respondent	Summary of Response
R3	No, there currently is not good integration
R4	No and there is a gap in integrating genetic information with family history
R5	No—currently physicians are operating under the false assumption that they can intuit genetic risk with a limited amount of phenotypic information. We need to: <ul style="list-style-type: none"> <li>• Improve integration of family history through the emergence of genomic applications that will require in depth family history before a provider can receive reimbursement for ordering the test, such as the BRCA test</li> </ul>
R6	No, health care providers are currently not integrating genomics and family history into public health areas well enough. We need to: <ul style="list-style-type: none"> <li>• Improve integration</li> <li>• Develop a reimbursement mechanism for providers to get paid for doing in depth family health histories</li> </ul>
R7	Not really. It is clearly very important but the fact that they are not aware of how well or poorly it has been integrated probably speaks to the fact that there hasn't been enough integration into existing programs
R8	No, not integrated well enough. We need to: <ul style="list-style-type: none"> <li>• Create more interaction between industry and clinicians as a large amount of continuing education to practicing physicians, at least around therapies, comes from industry.</li> <li>• Improve our genetics curriculum in medical schools</li> </ul>

**Q6. How are progress and health impact being tracked in integrating genomics in public health priorities?**

Respondent	Summary of Response
R1	<ul style="list-style-type: none"> <li>• Difficult to perform good analyses when individuals are collecting different types of data or using different metrics</li> <li>• Need to provide standards about what type of data should be collected and how it should be measured</li> </ul>
R2	<ul style="list-style-type: none"> <li>• We need to make sure that we establish good metrics around the data collection process as we are establishing large, public health programs</li> </ul>
R3	<ul style="list-style-type: none"> <li>• Currently, genomics is being used too narrowly in public health, mainly to track infectious agents and it is regrettable that we have little to no data on the host.</li> <li>• We need to begin collecting information in genomic variation in the public and should be measuring the number and diversity of individuals that we have genomic data on</li> </ul>
R4	—
R5	<ul style="list-style-type: none"> <li>• Evaluating hospitals and physicians on how consistently they are integrating well-validated genetic tests</li> <li>• Evaluating how committed medical colleges are to teaching genetics in their curriculum</li> </ul>

Respondent	Summary of Response
R6	<ul style="list-style-type: none"> <li>• Developing better carrots / sticks to affect physician behavior</li> <li>• Issuing clinical care guidelines alone will not be sufficient to change provider practice</li> </ul>
R7	<ul style="list-style-type: none"> <li>• We need to build the infrastructure for tracking outcomes in individuals that receive an intervention and perform longitudinal studies</li> <li>• By looking at clinical outcomes</li> </ul>
R8	<ul style="list-style-type: none"> <li>• Using health economics</li> <li>• Assessing whether there is an improvement in the effectiveness of intervention by sub-stratifying the patient populations most likely to benefit from treatment</li> <li>• Evaluating the number of new tests that are coming to market</li> </ul>

**Q7. Given the current economic realities in public health, what are the most important near-term priorities for action?**

Respondent	Summary of Response
R1	<ul style="list-style-type: none"> <li>• Maintain support for programs, such as EGAPP and GAPPNet, that provide us with models for evidentiary reviews</li> <li>• Continue support for communicating and disseminating evidentiary reviews</li> <li>• Fund large projects that look at both building the evidence base and implementation strategies, as opposed to fragmented projects</li> </ul>
R2	<ul style="list-style-type: none"> <li>• Ensure that existing programs do not get dismantled in this time of restricted funding because it is a huge waste of resources and investment</li> <li>• Ensure that we are not crashing existing broad programs to focus in on a specific disease area because it is a loss for the community</li> <li>• Perform an audit of the best pilot project opportunities as opposed to putting out grant applications</li> </ul>
R3	<ul style="list-style-type: none"> <li>• Correct the problem whereby patients give up the right to have any of the information generated on their samples in clinical trials where they served as participants</li> <li>• Bring about a shift in our thinking about whether patients have the right to access and share their own genomic data with the relevant research studies</li> <li>• Change the incentive structure for researchers to promote broader data sharing</li> </ul>
R4	<ul style="list-style-type: none"> <li>• Translate basic research findings into clinical practice</li> <li>• Improve provider education and raise awareness of new genetic information as it becomes available within the clinical community</li> <li>• Take a registry approach to collect genetic data on a larger, broader group of individuals</li> <li>• Improve the breadth and diversity of populations represented in genomic datasets</li> </ul>
R5	<ul style="list-style-type: none"> <li>• Conduct evidence based reviews on emerging genetic applications</li> <li>• Establish mechanisms for clinicians to make sense of emerging genomic knowledge and applications. Professional societies would be an excellent way to provide continuing education to physicians</li> </ul>

Respondent	Summary of Response
R6	<ul style="list-style-type: none"> <li>• Determine how we will translate the rapid improvements in our basic understanding of the genome into clinically actionable items</li> <li>• Continue the discussion around what level of evidence is needed to integrate new technologies into a clinical setting</li> <li>• Design trials to help us achieve incremental evidence development</li> </ul>
R7	<ul style="list-style-type: none"> <li>• Connect genetic information with lifestyle (environmental) variables</li> <li>• Fix Medicare and Medicaid</li> </ul>
R8	<ul style="list-style-type: none"> <li>• Encourage clinical investment in genomic diagnostics</li> <li>• Improve our ability to diagnose and predict disease progression</li> </ul>

### Emerging Themes by Working Group Topic Area

For the purposes of reevaluating and prioritizing near-term and long-term objectives in public health genomics, the OPHG is organizing discussions around four topic areas:

- 1) Prevention: Identify individuals, families, and communities at risk
- 2) Detection: Detect diseases early and intervene effectively
- 3) Development & Evaluation: Advance technology development, evaluation, and evidence generation

- 4) Pathways & Interactions: Understanding how pathways and gene-environment interactions impact population health

Each of these areas will need to assess a number of crosscutting issues, including education, workforce training, community engagement, infrastructure, integration, funding, data sharing, and ethical, legal and social implications (ELSI). In the following section we break down the comments of our key informants by topic areas and crosscutting issues.

### Prevention

Topic	Comments
Integration	<ul style="list-style-type: none"> <li>• Improve integration of family health history and genetic information into clinical practice to help identify at-risk individuals</li> </ul>
Funding	<ul style="list-style-type: none"> <li>• Create a reimbursement mechanism to compensate physicians for taking in depth family health history</li> </ul>

### Detection

Topic	Comments
Funding	<ul style="list-style-type: none"> <li>• Increase the investment in diagnostics and pharmacogenomics</li> <li>• Perform more health economic studies to demonstrate the value new diagnostic devices bring to the health care system</li> <li>• Tie reimbursement of diagnostics to value rather than costs</li> </ul>
Workforce Training	<ul style="list-style-type: none"> <li>• Ensure practicing physicians receive the continuing education need to appropriately utilize and interpret complex genomic-based applications</li> <li>• Use physician self-reported data on competencies and comfort with genetics as a metric for tracking progress</li> </ul>

## Development & Evaluation

Topic	Comments
Community Engagement	<ul style="list-style-type: none"> <li>• Continue to engage the community in a discussion around ongoing issues in evidence development and evaluation</li> <li>• Continue to discuss the level of evidence needed to move emerging genetic applications into a clinical setting</li> <li>• Continue to discuss models for evidence generation and implementation strategies</li> <li>• Engage a greater number of stakeholder groups, such as professional societies, in producing evidence reviews</li> </ul>
Infrastructure	<ul style="list-style-type: none"> <li>• Maintain existing evidence review programs such as GAPPNet and EGAPP in the face of funding restrictions because dismantling the existing infrastructure will halt progress and result in a loss on the investment that has already been made in these programs</li> </ul>
Integration	<ul style="list-style-type: none"> <li>• Discuss strategies for integrating emerging technologies in the absence of perfect evidence</li> <li>• Continue to discuss models for incremental evidence development</li> <li>• Improve the dissemination of evidence-based reviews to the appropriate professional</li> </ul>
Funding	<ul style="list-style-type: none"> <li>• Maintain or increase the funding stream for evidence reviews</li> <li>• Increase our investment in developing diagnostic products</li> <li>• Fund holistic projects that both collect evidence on effectiveness and develop implementation strategies</li> </ul>

## Pathways & Interactions

Topic	Comments
Infrastructure	<ul style="list-style-type: none"> <li>• Create data repositories capable of housing large, widely accessible phenotype-genotype datasets</li> <li>• Create mechanisms to systematically and electronically pull phenotype information from existing sources such as medical records</li> <li>• Build the infrastructure needed for individuals to access, store, and control the sharing of their clinical information electronically</li> <li>• Develop a system for collecting and tracking characteristics of responders and non-responders</li> </ul>
Data Sharing	<ul style="list-style-type: none"> <li>• Enact new policies that will help, not hinder, data generation</li> <li>• Enhance existing data sharing policies to reduce the lag phase between when the data is generated and when it is shared after publication</li> </ul>
Education	<ul style="list-style-type: none"> <li>• Educate consumers, particularly those involved in clinical trials, on the importance of data sharing in biomedical research</li> </ul>
Integration	<ul style="list-style-type: none"> <li>• Develop a plan for integrating the “tsunami” of basic research findings on the human genome into translational research projects</li> <li>• Improve interagency collaboration among federal organizations</li> <li>• Improve federal leadership in creating large, collaborative research studies</li> </ul>
Community Engagement	<ul style="list-style-type: none"> <li>• Engage consumers to become more vocal in demanding that the locus of control on clinical and genetic data generated through clinical research reside with the individual</li> <li>• Encourage consumers to demand that they be allowed to control the secondary reuse of their data</li> </ul>

## APPENDICES TO PART ONE

# STAKEHOLDER CONSULTATION APPENDIX 5: SUMMARIES OF INFORMAL DISCUSSIONS

DEVELOPING PRIORITIES FOR PUBLIC HEALTH GENOMICS  
2012–2017

Stakeholder Engagement of the Public Health Community  
(Academe; Practice; Community)

## I. BREAKFAST DISCUSSION—GENETIC ALLIANCE CONFERENCE—JUNE 25, 2011

### Summary of Discussion

#### Present:

Toby Citrin, Center for Public Health and Community Genomics,  
Facilitator

Vence Bonham, National Human Genome Research Institute

Jo Boughman, American Society for Human Genetics

Alex Ellerbeck, Genetic Alliance

Greg Feero, National Human Genome Research Institute

Aaron Goldenberg, Case Western Reserve University

Barbara Koenig, Mayo Clinic/University of California San Francisco  
(soon)

Chikezie Maduka, University of Maryland Prevention Research Center

Amber Mills, Genetic Alliance

Elizabeth Prom-Wormley, Virginia Commonwealth University

Michael Rackover, National Human Genome Research Institute/American  
Academy of Physician Assistants

Jillian Tietjen, University of Michigan

Marc Williams, Intermountain Healthcare

### Introduction

Toby Citrin introduced the breakfast discussion with background on the Stakeholder Consultation (see accompanying summary sheet). He described this session as a “discussion” sharing ideas about the future of public health genomics, to be incorporated with other sources of input including key informant interviews, a Wiki developed by the APHA Genomics Forum, and responses to a Request for Information expected to be issued by CDC’s Office of Public Health Genomics. Toby suggested that we consider the public health system broadly, utilizing the IOM’s definition embracing governmental public health, the healthcare delivery system, academe, business/labor, media and the community at large. Toby emphasized that this is to be an informal discussion sharing ideas, as distinguished from

the more formal Request for Information that will take place in the future.

### Key Ideas from the Discussion

- There is a current expectation that funding of governmental public health programs will be increasingly determined by evidence of lives saved and expected to be saved. While these are medical questions rather than public health questions, we will have to develop metrics to demonstrate the value of programs utilizing genomics in public health practice.
- At least some components of newborn screening programs can and should demonstrate their cost-effectiveness.
- One area where we should be able to justify cost effectiveness is in the evaluation of genetic tests. EGAPP has been doing this, but has yet only recommended one test. We need an evaluation process defining outcomes, prioritizing interventions, and implementing them to see if they work. We need to clarify what makes a good study and how to know when an intervention is valuable enough to be implemented. We need a clearinghouse providing access to information on these interventions. CDC’s GAPPNet project and the CDC-Genetic Alliance’s GEDDI project have been working in these areas.
- Genomics needs to be incorporated in multiple sectors of public health rather than being in its own compartment. We can think in terms of a matrix with priority public health challenges on one axis and genomics as a cross-cutting axis applying what we know about genomics to addressing each of these problems. We need to assemble and disseminate this knowledge across the public health system. Healthy People 2020 can be used in this process. Genomics need to become “mainstream” rather than being treated exceptionally.
- In applying the above matrix framework, people responsible for specific areas of public health (e.g., environmental health) need to sit at the table with genomics experts to identify ways in which genomics can enhance the effectiveness of public health programs. Comparative Effectiveness Research should be applied in this process.
- A priority for public health genomics should be gathering and storing genomic and genomic-related information that will be useful in genomics research, whether or not such research is carried out by the public health system or within the next five years.
- Public, K-12 and provider education should be an agenda for public health genomics. Public health should partner with science education groups in carrying this out.
- We need to develop and utilize standards indicating the value that can be achieved by applying genomics to various public

health problem areas. Family health history may be one of the genomic tools that can demonstrate its value in addressing critical public health problems. Education should be used to disseminate knowledge of effectiveness of genomic approaches facilitating their utilization.

- We need to recognize that genomics is still a “half-way technology” with respect to common complex diseases. At present, however, non-genetic interventions (e.g., diet and exercise) are much more effective in disease prevention.

## II. SPIG TELECONFERENCE—JULY 18, 2011

### On Call

#### SPIG Members:

Ella Greene-Moton, University of Michigan, NCC SPIG Facilitator  
 Winona Hollins-Hauge, University of Washington, HPRC/NCC  
 Catherine Haywood, Tulane University Prevention Research Center  
 Frieda Brown, Tulane University Prevention Research Center  
 Chikezie Maduka, University of Maryland Prevention Research Center  
 Sharon Shad, University of Massachusetts Medical School Prevention Research Center  
 David Collins, Morehouse School of Medicine Prevention Research Center  
 Imogene Wiggs, Missouri Department of Health and Senior Services  
 Frieda Gonzales, New Mexico Department of Health

#### CPHCG Staff:

Sally Meyer  
 Megan Knaus  
 Nora Isack  
 Toby Citrin

### Introduction

The teleconference began with an on-line viewing of genomics 101 materials from the Utah Genetics Science Learning Center ([learn.genetics.utah.edu](http://learn.genetics.utah.edu)). Ella Greene-Moton stated how happy the SPIG is to be the voice of the community for the stakeholder consultation process. Ella described the CDC Office of Public Health Genomics’ initiative to develop a strategic plan for public health over the next five years in response to the Office’s significant downsizing. The group was encouraged to share their thoughts and ideas on the future of public health genomics from the community perspective.

### Key Ideas from the Discussion

#### Why Important that Community Voice is Heard

- Community is the heart of public health
- Community is often left behind as public health moves forward

#### Framework/Issues for Discussion

- Importance of public health genomics in communities
  - People are still unaware; public health genomics is new to most. Education is needed in communities.
  - Communities need to understand the role of genetics in predisposition to diseases and behaviors.
  - Epigenetics is an important aspect of public health genomics. Behaviors and the environment can increase/reduce risk for certain individuals. How do we deal with this information?
  - Public health genomics should be used to reduce health disparities.
- Engaging the community in genomics
  - The SPIG should play a role in educating PRC boards and getting genomics on their agendas.
  - Educational tools are needed.
  - The importance of family health history needs to be realized by community members.
  - There are genetic components to common diseases. We need to teach the community and incorporate genomics into current PRC research agendas.
- Actions that need to be taken
  - Educate faith-based organizations so they can disseminate knowledge to communities.
  - Visual tools must be developed and shared with community leaders/gatekeepers. These tools must be in genomics 101 language.
- Engaging public health agencies
  - Community health workers need an early understanding of genomics. They have good connections with the community, especially those who are sick. If community health workers have training they can disseminate information.
  - People need to know what resources are available in their county health departments.
  - Hands on activities are the best way to engage community members. Such activities should be shared at back-to-school nights and community health fairs.
- Community needs to further public health genomics

- What is going to be the emphasis of public health genomics over the next five years? Should money be spent on personalized medicine or for population health and prevention? We want to find ways that genomics can be used to help public health and reduce health disparities. Money should be spent on population health.
  - Communities need education. Closing the education gap will help reduce health disparities. Education is key.
- Helpful policies to guide public health genomics
    - Policies must be in place to protect privacy, prevent discrimination, especially from insurance companies. Communities need transparency.
    - The NCC SPIG will add looking at gaps in policies to its agenda.
    - Ethics around genomics should be investigated by communities.
    - People should understand genomics and feel empowered to use genetic information to improve their health outcomes.
  - Anticipated barriers and how to overcome them
    - There will be barriers in disseminating information
    - Information has the potential for misuse, and could result in discrimination. Processes need to be explained to the public.

### III. PUBLIC HEALTH PRACTICE DISCUSSION GROUP—AUGUST 19, 2011

#### Summary of Discussion

##### Facilitators

Karen Greendale, Director, Cancer Survivorship Initiatives, New York State Department of Health

Deb Duquette, Genomics Coordinator, Michigan Department of Community Health

##### On Call

Dale Lea, Public Health Consultant, Maine State Genetic Programs

Amy Zlot, Oregon Genetics Program, Oregon Health Service

Michelle Kempf-Weibel, State Genetic Coordinator, Wisconsin Division of Public Health

Gail Marcus, Public Health Genetics, North Carolina Department of Public Health

##### CPHCG Staff

Nora Isack

##### Defining public health genomics

- The definition of public health genomics should be revisited to stress the relationships between genes, the environment, and personal choices and behaviors. Public health genomics must also focus on increasing access to genetics services and reducing health disparities.
- Public health genomics must be defined in terms that are understood by and exciting to the public health community and the community-at-large.

##### Anticipated barriers and how to overcome them

- Not enough providers are educated about genomics and how to use genomic tools, including family health history
- Reaching out to nursing schools and integrating genomics competencies into their curricula is one way to impact workforce education
- Educational tools must be evaluated to ensure that we are making a difference and getting the health outcomes that we want. We need data on the impact of educational efforts.
- The current funding prioritizes research and ELSI, and not enough money is going towards integration and translation of genomics

##### Engaging the community in genomics

- Public health departments have a role in educating the public. Efforts have focused on provider education, but if people do not have access to a provider, they may not have a way of getting the information they need.
- NHGRI provides money for community events for National DNA Day, but it's hard to get people excited.
- The CDC has a community guide, and there is no mention of genomics within that. This is a place to focus on genomics within communities.

##### Partnerships with academia

- NIH provides a lot of funding to academics, and public health departments should partner with schools of public health to help integrate research initiatives into practice.
- Funds should be put towards stipends to encourage genetics experts to study public health, and there are not enough people who do public health and genetics.

**Helpful policies to guide public health genomics**

- The Healthy People 2020 goals include actions for HBOC and Lynch syndrome, and given the funding climate, these are things we should focus on. There is already buy-in from every federal agency with these, and these are already priorities. We should focus on these conditions and use them to guide future programs. Additionally, many states have received CDC money for colorectal cancer screening, and every state has funding for breast and cervical cancer screening.
- Genetic counselors should be licensed, and right now only a few states have this as a policy. States also need to work with Medicaid and private insurers to ensure that genetic counseling services are reimbursed.
- There are not enough genetic counselors in many states, creating geographical disparities. This is a result of a lack of recognition of genetic counseling services and reimbursement.
- Information gathering, such as family health history, should be standardized and integrated into electronic medical records to facilitate surveillance.

**Priorities for public health genomics over the next five years**

- Revisit the definition of public health genomics to make it more exciting and include a lifespan approach
- Develop a national plan for integrating genomics competencies into nursing education that includes an outcomes evaluation
- Leverage genomics in the Healthy People 2020 goals by developing materials and fact sheets, and focusing on HBOC and Lynch syndrome within cancer screening programs.
- Develop policies for genetic counselor licensure and insurance coverage.
- Engage communities in public health genomics education
- Utilize surveillance as a tool to track genetic diagnosis, prevention, intervention across populations.

## APPENDICES TO PART ONE

## STAKEHOLDER CONSULTATION APPENDIX 6: REQUEST FOR INFORMATION (RFI) SUMMARY

Comment #	Name	Respondent Category	Summary	Major Themes
1	Cindy Fogle	Practice, Non-traditional	Incorporation of massage therapists into neoadjuvant practices and research studies related to cancer, specifically related to hypoxia and breast cancer. Policies and scope of practice must be examined to incorporate massage therapy into treatment.	<ul style="list-style-type: none"> <li>• Research (translational)</li> <li>• Non-traditional techniques</li> <li>• Cancer</li> </ul>
2	James Haddow	Practice, Medical	The EGAPP working group needs revisiting to streamline processes, networks, and implementation strategies. Time and efficiency is a great concern of the public; however, there is no process for those tests that have positive recommendations. Funding and dedicated staff from the CDC is essential for sustainability of a group like the EGAPP working group.	<ul style="list-style-type: none"> <li>• Evidence-based genomics</li> <li>• Knowledge management</li> <li>• Funding</li> <li>• Evaluation</li> <li>• Research, translational</li> <li>• Policy development</li> <li>• Past successes of OPHG</li> <li>• Lynch syndrome</li> </ul>
3	Terri Combs-Orme	Academic, other institutions	Genomics needs to be translated into understandable terms for the general public and used for prevention, ultimately reducing health disparities, by focusing on the understanding of epigenetics.	<ul style="list-style-type: none"> <li>• Prevention</li> <li>• Health disparities</li> <li>• Health/genomics literacy</li> <li>• Gene-environment interaction/epigenetics</li> </ul>
Email #1	Richard Carmona	Government, federal	Improving health literacy, for both the healthcare workforce and the general public, is a necessary first step to further the integration of genomics in public health practice. HHS, CDC, White House, and Congress need to push these issues to create demand and continue improving the nation's health.	<ul style="list-style-type: none"> <li>• Professional training</li> <li>• Health genomics/literacy</li> <li>• Research-translational</li> <li>• Knowledge management</li> </ul>
4	Steven Teutsch	Government, local ; Practice, public health	Evidence-based guidance is needed to ensure that genomics is utilized effectively and efficiently. NIH, AHRQ, and professional groups should be at the table for the development of guidelines.	<ul style="list-style-type: none"> <li>• Clinical criteria</li> <li>• Evidence-based genomics</li> <li>• Efficacy</li> <li>• Immediate tasks/short-term steps</li> <li>• Research</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
5	Brian	None provided	Academic institutions and healthcare providers must partner to develop physician guidelines, clinical utility, and cost-effectiveness data for genetic tests. Infrastructure for IRBs and databases will facilitate the sharing of information to facilitate the evidence finding process.	<ul style="list-style-type: none"> <li>•Clinical criteria</li> <li>•Professional training</li> <li>•Health/genomics literacy</li> <li>•Knowledge management</li> <li>•Databases</li> <li>•Collaborations/partnerships</li> </ul>
6	Elizabeth Balkite	Non-profit organization, health-related	Public health genomics needs to identify champions and pursue them to ensure that genomics remains an important component of public health. Education must be included in professional degree curricula, and public health needs to incorporate genomics into its activities. Groups like EGAPP must be provided with adequate funding and support, in the long-term, to be a trusted voice for public health genomics.	<ul style="list-style-type: none"> <li>•Professional training</li> <li>•Health/genomics literacy</li> <li>•Message tailoring</li> <li>•Evidence-based genomics</li> <li>•Defining public health genomics</li> </ul>
7		None provided	Funds need to be provided for randomized control studies of medical interventions for individuals with specific genotypes. Scientific evidence will enable targeted medical interventions based on genotype. Barriers will include the time and cost of such studies.	<ul style="list-style-type: none"> <li>•Research</li> <li>•Evidence-based genomics</li> <li>•Pharmacogenomics</li> <li>•Efficacy</li> <li>•Funding</li> </ul>
8	Sarah Copeland	Government, federal	Public health genomics needs to be better defined to maximize strategies to realize its full public health potential.	<ul style="list-style-type: none"> <li>•Health/genomics literacy</li> <li>•Defining public health genomics</li> </ul>
Email #2	AHRQ Gurvaneet Randhawa	Government, Federal	Much needs to happen for genomics to integrate into public health. More gene-based tests need to be developed, and the social outcomes, as well as the incremental benefit of these tests. This will facilitate the development of evidence-based guidelines and clinical decision support for genetic tests. The public health system should improve coordination of genomics research and activities through electronic infrastructures and improve public-private partnerships. Federal and state agencies should work with professional organizations, payers, test developers, the pharmaceutical industry, patients, and consumer advocacy groups ensure access, low costs, and benefits. Barriers must be overcome to share resources and information among groups with different interests. Past efforts, such as GAPPNet, EGAPP, and Effective Health Care, should be expanded upon.	<ul style="list-style-type: none"> <li>•Genetic services</li> <li>•Efficacy</li> <li>•Evidence-based genomics</li> <li>•Knowledge management</li> <li>•Accessibility</li> <li>•Funding</li> <li>•Collaborations/Partnerships</li> <li>•Utilization</li> <li>•Health/genomics literacy</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
10	Josh	None given	Research should be done to improve outcomes for complex diseases through the use of genetic tests. Improve regulatory guidelines will ensure quality research and clinical care.	<ul style="list-style-type: none"> <li>•Genetic services</li> <li>•Clinical criteria</li> <li>•Policy development</li> </ul>
11	Joyce Hooker	Non-profit organization, health-related	State health departments need to have full time state genetics coordinators to cover public health genetics programs beyond newborn screening. Currently, newborn screening coordinators serve as both.	<ul style="list-style-type: none"> <li>•Funding</li> <li>•Immediate tasks/short-term steps</li> <li>•Professional training</li> </ul>
12	Philip Zazove	Practice, medical	Physicians need to incorporate family health history into routine primary care. The development of tools, through electronic medical records, will help facilitate use of family history to improve preventive medicine.	<ul style="list-style-type: none"> <li>•Family history</li> <li>•Knowledge management</li> <li>•Electronic medical records</li> <li>•Professional training</li> <li>•Funding</li> </ul>
13	Jenny Johnson	Government, state ; Practice, public health	Public health genomics should focus on the most practical public health programs, such as family health history. State and local health departments are not equipped for high cost research initiatives. Family health history is a true public health program that is simple for communities and practitioners to engage in, and family health history can impact chronic conditions such as obesity, cancer, and heart disease.	<ul style="list-style-type: none"> <li>•Family history</li> <li>•Prevention</li> <li>•Chronic diseases</li> <li>•Health/genomics literacy</li> <li>•Professional training</li> <li>•Health communications/ social marketing</li> </ul>
14	Diane J. Allingham-Hawkins	For-profit organization, consulting	Public health genomics stakeholders require education about genomics and genetic tests, and the applications, costs, and benefits of such tests.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Evidence-based genomics</li> <li>• Funding</li> <li>• Policy development</li> <li>• Health/genomics literacy</li> <li>• Family history</li> <li>• Collaborations/ partnerships</li> </ul>
15	Jill Hagenkord	Academic, Medical schools	More education and policies related to provider education in genomics and clinical guidelines will help the medical professionals and patients realize cost-savings and preventive measure for those with particular genotypes.	<ul style="list-style-type: none"> <li>•Clinical criteria</li> <li>•Professional training</li> <li>•Efficacy</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
16	Laura Senier	Academic, Medical Schools	CDC needs to lead the development and integration of genetic education competencies for both the workforce and medical/public health students. Learning outcomes should be evaluated to ensure genomic competencies are integrated into healthcare. Vetted competencies are a step towards adoption of tools such as family health history, as well as continued and improved integration of these tools into healthcare. State health departments are key players in this process, especially in their role of providing services to minority and underserved communities.	<ul style="list-style-type: none"> <li>• Policy development</li> <li>• Health/genomic literacy</li> <li>• Professional training</li> <li>• Ethicalness</li> <li>• Family history</li> <li>• Genetic testing</li> <li>• Chronic disease</li> </ul>
Email #3	Ron Zimmern	Non-profit organization, health-related	The potential of genomics in public health has yet to be realized by healthcare agencies and leaders in the United State. Funding, education, and partnerships with industry are essential to bringing genomics to the forefront of public health to tackle chronic conditions.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Health communications/ social marketing</li> <li>• Health applications</li> <li>• Evidence-based genomics</li> <li>• Collaborations/ partnerships</li> <li>• Immediate tasks/short-term steps</li> </ul>
17	Leonard Levy	Academic, Medical Schools	Genetics and genomics should be integrated into medical education, supplemented by efforts in public and private health professional agencies and organizations, to further the role of genomics in improving public health.	<ul style="list-style-type: none"> <li>• Professional training</li> </ul>
Email #4	Jim Evans	Academic, Medical Schools	Due to technology advances, genotyping is becoming more and more of a public health reality. Individuals with highly penetrant genes can be identified for early treatment and intervention through genetic testing. This technology will be most valuable for genes that are penetrant, conditions that are treatable, conditions that cannot be identified without the genetic information, and genes that are common. Barriers include costs, ethical concerns, and education	<ul style="list-style-type: none"> <li>• Research</li> <li>• Clinical criteria</li> <li>• Health/genomics literacy</li> <li>• Professional training</li> <li>• Accessibility</li> <li>• Population screening</li> <li>• Ethicalness</li> <li>• Health disparities</li> </ul>
18	Association for Molecular Pathology Timothy J. O'Leary	Non-profit, health related	Advances in genetic technology are contributing to diagnosis and the advancement of personalized medicine. Yet, further research on clinical validity and clinical utility is necessary before integration into routine practice.	<ul style="list-style-type: none"> <li>• Clinical criteria</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
19	Steven Galen	Non-profit organization, community-based organization	Education is essential to advancing genetic technologies in public health. Communities, especially minority communities, must be engaged in the process to ensure trust and buy-in.	<ul style="list-style-type: none"> <li>• Policy development</li> <li>• Professional training</li> <li>• Health/genomics literacy</li> <li>• Databases</li> </ul>
20	Ned Calonge	Non-profit organization, health related	More research is needed to develop the evidence needed for integration of genomics into public health and medicine. However, genetic exceptionalism remains a barrier, and the public health system must find ways to gain public trust and access genetic information at a population level. This will help practitioners identify genomic trends and the most effective interventions, without wasting time, money, and/or resources or putting patients at risk. An evidence-based approach requires collaborations between NIH, AHRQ, CDC, and CMS to conduct research and develop clinical and coverage guidelines. Evidence is a key part of education and the policymaking that will move genomics.	<ul style="list-style-type: none"> <li>• Regulation</li> <li>• Clinical Criteria</li> <li>• Policymaking</li> <li>• Health Outcomes</li> <li>• Funding</li> <li>• Research</li> <li>• Professional training</li> <li>• Genetic exceptionalism</li> </ul>
21	Tricia Page	Academic, Medical Schools	Public health genomics should take on young adult genetic screening as a prevention strategy. Education, system management, further research, and ethical considerations are priorities for young adult screening.	<ul style="list-style-type: none"> <li>• Population screening</li> <li>• Health/genomics literacy</li> <li>• Knowledge management</li> </ul>
22	Donald Lyman	Non-profit organization, health-related	The integration of genomics into public health will require evidence-based reviews of applications, the development of protocols, innovative ways to integrate data sets and registries, and the help of federal agencies to manage research, policy, and translation.	<ul style="list-style-type: none"> <li>• Clinical criteria</li> <li>• Evidence-based genomics</li> <li>• Knowledge management</li> </ul>
23	Amy Miller	Non-profit organization, health-related	The Personalized Medicine Coalition believes that the CDC Office of Public Health Genomics should develop a committee that can tackle the funding, evaluation, programs, and education necessary for personalized medicine.	<ul style="list-style-type: none"> <li>• Clinical criteria</li> <li>• Policy development</li> <li>• Professional training</li> <li>• Efficacy</li> <li>• Pharmacogenomics</li> </ul>
24	Sheri Schully	Government, federal	Pharmacogenomics should a priority for public health genomics, as it can reduce morbidity and mortality, as well as health care costs. Collaboration across agencies and funding will be necessary.	<ul style="list-style-type: none"> <li>• Pharmacogenomics</li> <li>• Funding</li> <li>• Collaborations/ partnerships</li> </ul>
25	James Bowery	Non-profit organization, health-related	Genetic information should be included in surveillance data for use in research and information gathering.	<ul style="list-style-type: none"> <li>• Population screening</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
26	Linda Bruzzone	Non-profit organizations, health-related	Lynch syndrome is a condition with high heritability, and therefore is an exemplary condition for public health genomics. Provider education about family history taking, family risk for cancer, and Lynch syndrome will facilitate preventive measures to save lives and reduce costs. Partnerships across the healthcare system are essential to ensure that interventions reach everyone, especially those without health insurance.	<ul style="list-style-type: none"> <li>• Clinical criteria</li> <li>• Policymaking</li> <li>• Policy development</li> <li>• Professional training</li> <li>• Prevention</li> <li>• Lynch syndrome</li> <li>• Family history</li> </ul>
27	William Ebomoyi	Academic, Schools of Public Health	Providers need more education about genomics and how to integrate genomics into everyday practice. Insurance companies also need to be educated to understand the value of genomic tools.	<ul style="list-style-type: none"> <li>• Professional training</li> <li>• Insurance coverage</li> </ul>
28	Maki Moussavi	For-profit organizations, Consulting	Genomics needs to be integrated into clinical practices. This can be achieved through the creation of data warehouse, clinical decision support tools, and the translation of genomic knowledge. Provider education, along with further research, will ultimately improve disease management and further the integration of genomics into practice.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Policy development</li> <li>• Professional training</li> <li>• Collaborations/partnerships</li> </ul>
30	Jacqueline Johnson Pata	Non-profit organizations, community-based organization	Before public health genomics can impact health in the American Indian and Alaska Native communities, researchers and the government must work with tribal leaders to establish priorities, protocols, and an understanding of benefits to gain trust among community members. If this trust is achieved, American Indian and Alaska Native communities will gain education about genomics as well as improved health outcomes based on research.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Health disparities</li> <li>• Health/genomics literacy</li> <li>• Collaborations/partnerships</li> <li>• Ethicalness</li> </ul>
31	Michael Watson	Non-profit organizations, health-related	Data collection is necessary to improve the evidence base for genomics in multiple populations, as well as incorporate information about environment into data sets. Workforce education and collaborations with federal agencies, professional societies, state health departments, and regulatory agencies are necessary to facilitate the translation of genomics into public health practice. Caution must be taken to ensure that genomic technologies are not integrated into the healthcare system without proper evidence and education.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Professional training</li> <li>• Collaborations/partnerships</li> <li>• Genetic services</li> <li>• Knowledge management</li> <li>• Gene-environment interactions/epigenetics</li> <li>• Evidence-based genomics</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
32	Jimmy Efirid	Brody School of Medicine	Examine the interaction of pesticide exposure and various gene variants including PON1,33, se4242382, rs783732846, EGFR, RAS, NF1, PTEN, PI3K, TP53, MDM2, MDM4, p14ARF, p16, RB1, IDH1, IDH2 on risk for brain cancer.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Cancer</li> </ul>
33	Oregon Health Authority	Government, state ; practice, public health	CDC must play a role in the translation of genomics research as its reach is nationwide. The public health system must work with all stakeholders to ensure that genomics technologies are evidence-based and accessible to the entire public. Partnerships and adequate funding will ensure translation of genomics to improve health.	<ul style="list-style-type: none"> <li>• Gene-environment interaction/epigenetics</li> <li>• Funding</li> <li>• Knowledge management</li> <li>• Research</li> </ul>
34	Ann Cashion	Academic, Other Institutions	CDC must play a role in the translation of genomics research as its reach is nationwide. The public health system must work with all stakeholders to ensure that genomics technologies are evidence-based and accessible to the entire public. Partnerships and adequate funding will ensure translation of genomics to improve health.	<ul style="list-style-type: none"> <li>• Gene-environment interaction/epigenetics</li> <li>• Funding</li> <li>• Knowledge management</li> <li>• Research</li> </ul>
35	Cara Tenenbaum	Non-profit organizations, health-related	More research is needed to address the genetic susceptibility to ovarian cancer, particularly the genes BRCA1/2 and the genes for HNPCC.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Cancer</li> </ul>
36	Cornelia Van Duijn	Academic, Schools of Medicine	Public health genomics can utilize screening technology to identify susceptible populations and engage these populations in prevention.	<ul style="list-style-type: none"> <li>• Policy development</li> <li>• Population screening</li> </ul>
37	Nuananong Seal	Academic, Other Institutions	Environment is a necessary component for addressing obesity and health outcomes in underserved populations.	<ul style="list-style-type: none"> <li>• Gene-environment interactions/epigenetics</li> </ul>
38	Gregory Fowler	Oregon Health and Science University	Community engagement is vital to policy development, particularly in the evaluation of values, where scientific evidence cannot speak.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Policymaking</li> <li>• Policy development</li> <li>• Collaboration/ Partnerships</li> <li>• Ethicalness</li> <li>• Community engagement</li> </ul>
39	Kristi Zonno	Non-profit organizations, health-related	Genomic literacy is essential for both the public and workforce to apply genetic technologies to health goals. This will require an interdisciplinary approach and the integration of genomics competencies at all levels of schooling and professional training. Grant programs should be developed to fund these initiatives.	<ul style="list-style-type: none"> <li>• Health/genomics literacy</li> <li>• Professional training</li> <li>• Health communications/ social marketing</li> <li>• School curriculum</li> <li>• Funding</li> <li>• Accessibility</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
40	Suzanne Feetham	Academic, Other Institutions	Education for the public and the workforce is an essential step for the integration of genomics into public health. The CDC OPHG must collaborate with the other agencies and professional societies to gather the evidence that will secure the role of genomics in improving population health.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Clinical criteria</li> <li>• Funding</li> <li>• Professional training</li> <li>• Policy development</li> <li>• Health/genomics literacy</li> <li>• Family history</li> <li>• Collaborations/partnerships</li> </ul>
41	Herbert F. Young	Non-profit organization, health related	Genomic technologies need to be evaluated for evidence of validity and utility.	<ul style="list-style-type: none"> <li>• Research</li> <li>• Evidence-based genomics</li> <li>• Clinical criteria</li> <li>• Funding</li> <li>• Professional training</li> <li>• Health/genomics literacy</li> </ul>
42	Julian Little	Academic, Schools of Public Health	Genomic technologies need to be rigorously evaluated, both for their safety and cost-effectiveness, in a timely manner to be integrated with healthcare. The cutting of funding to NOPHG will severely hinder this necessary step in the translation of genomic technologies.	<ul style="list-style-type: none"> <li>• Clinical criteria</li> <li>• Risk assessment</li> <li>• Evidence-based genomics</li> <li>• Policy development</li> <li>• Efficacy</li> </ul>
44	None provided	None provided	Genomic tests need to be evaluated and the public needs to be educated on what the tests can and cannot do.	<ul style="list-style-type: none"> <li>• Clinical criteria</li> <li>• Evidence-based genomics</li> <li>• Health/genomics literacy</li> <li>• Regulation</li> <li>• Knowledge management</li> </ul>
45	J. James Rohack	Non-profit organization, general health	The genomics of infectious disease, health applications, and drug effectiveness should be studied as well as behavioral outcomes related to genetic testing. Standardization of data collection is crucial. Policies to coordinate agencies must be put in place and furthermore institutions must collaborate. Payment protocol for genetics laboratories must be examined.	<ul style="list-style-type: none"> <li>• Health applications (infectious disease, chronic disease)</li> <li>• Genetic technologies (screening)</li> <li>• Databases</li> <li>• Collaboration/partnerships</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
46	James Madara	Non-profit organizations, health-related	CDC should focus efforts in genomics on common disease and genomic-based screening and interventions that will test broad application. Family health history should be a priority to evaluate in its applications to health. Evidence-based techniques (and EGAPP's work) must be supported and funded. A genomic literate healthcare workforce is crucial.	<ul style="list-style-type: none"> <li>Evidence-based genomics</li> <li>Health applications (chronic disease)</li> <li>Evaluation (genetic technology)</li> <li>Professional training/education</li> <li>Genomics/health literacy</li> </ul>
47	Joan Scott	Non-profit organization, Health-related (professional society)	Evidence that supports the use of specific genomic applications in defined populations is important. There is a need for an increase in public health and provider skills in genomics. Collaboration and partnership of the medical community and public health community must happen; incentives might help coordinate this process.	<ul style="list-style-type: none"> <li>Evidence-based genomics</li> <li>Clinical criteria (Clinical utility)</li> <li>Professional training/education</li> <li>Silos</li> <li>Collaboration/partnerships</li> </ul>
49	Ruth Lynfield	None provided	Genomics can advance public health in the area of infectious disease by exploring microbes and host responses. Advances in microbial genetics can lead to targeted therapies and vaccines. Privacy and security of data and materials, consent issues and policies for high standards are important. There needs to be collaboration and coordination between CDC, NIH, academic institutions and professional societies.	<ul style="list-style-type: none"> <li>Microbial genetics</li> <li>Ethicalness</li> <li>Collaboration/partnerships</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
50	Jean Chabut	Government, state ; Practice, Public Health	Identify how genomics can fit into comprehensive national initiatives, collaborate with chronic disease and develop a multidisciplinary approach, and eliminate "silos". Identify evidence-based and cost-effective genomics applications. Additionally evaluate and regulate genomics technologies. Develop coordinated plan on how to best implement genomics with health applications that will improve overall health outcomes.	<ul style="list-style-type: none"> <li>• Research (translational)</li> <li>• Funding</li> <li>• Policymaking</li> <li>• Health outcomes</li> <li>• Health disparities</li> <li>• Collaboration/partnerships (state and federal levels)</li> <li>• Silos</li> <li>• Health applications (chronic disease)</li> <li>• Evidence-based genomics</li> <li>• Cost</li> <li>• Professional training</li> <li>• Genomics/health literacy</li> <li>• Regulation (genomic technology, gene patent)</li> <li>• Risk assessment</li> <li>• Evaluation</li> <li>• Efficacy (cost-benefit, cost-effectiveness)</li> <li>• Leadership</li> <li>• Defining public health genomics</li> <li>• Future of public health genomics</li> <li>• Genetic counselor licensure</li> </ul>
51	Sara Shostak	Academia, Other institutions	Explore the interaction of the public's access to care, education, culture, and lived experiences within their environments within the scope of genomics, particularly within the lens of environmental health sciences. Understand and address that unequal exposures, resources and factors outside of the body contribute to the molecular processes that shape health and illness.	<ul style="list-style-type: none"> <li>• Future of public health genomics</li> <li>• Gene-environment interactions/epigenetics</li> <li>• Health outcomes</li> <li>• Health disparities</li> <li>• Ethicalness</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
52	Infectious Diseases Human Genomics Working Group	Government, Federal (CDC Working group)	Research needed to define genetic/genomic epidemiologic risk factors such as susceptibility to infection, progression to disease, response to therapy, drug metabolism, treatment outcomes, and vaccine efficacy for infectious diseases. Policies to integrate collection of specimens for genetic/genomic susceptibility testing into ongoing studies evaluating epidemiologic risk factors should be explored, which could also minimize costs. NIH and CDC should form a collaboration/partnership.	<ul style="list-style-type: none"> <li>• Microbial genetics</li> <li>• Genetics/genomic risk factors</li> <li>• Health outcomes</li> <li>• Collaboration/partnerships</li> <li>• Research (translational)</li> </ul>
53	Infectious Diseases Society of America James M. Hughes	Non-profit organization, Health-related	There is a need to understand why some people are at higher risk for severe disease when infected due to differences in the way they respond to pathogens. CDC can serve a leadership role in the translation of microbial genetic research and its application to population health. IDSA recommends that CDC focus on questions of public health importance, develop a prioritized research agenda, coordinate activities with NIH, develop incentives and infrastructure for support of creative collaborations, think creatively about connecting pieces of emerging genomics data and make a clear commitment to develop a forward path.	<ul style="list-style-type: none"> <li>• Microbial genomics</li> <li>• Health applications</li> <li>• Future of OPHG</li> <li>• Future of public health genomics</li> <li>• Long term steps</li> <li>• Research (translational)</li> <li>• Health outcomes</li> <li>• Health disparities</li> <li>• Funding</li> <li>• Collaborations/partnerships (CDC/NIH)</li> <li>• Evaluation</li> <li>• Research incentives</li> </ul>
54	Joseph Capella	Academic, Other institutions	The use of data about individuals' beliefs and behavior will help guide the message tailoring for participation in genetic testing and genetic services.	<ul style="list-style-type: none"> <li>• Message tailoring</li> <li>• Genetic testing</li> </ul>
55	Connie Bormans	Non-profit organization, Health-related	The public health system should work with DTC genetic testing companies instead of against them to bring their laboratories and methods up to acceptable standards. Current policies should be streamlined to provide one uniform body of standards. Current regulatory channels have no approval process and no way of evaluating genetic testing.	<ul style="list-style-type: none"> <li>• Regulation (genetic testing)</li> <li>• Genetic Testing</li> <li>• Prevention</li> <li>• Regulatory policies</li> <li>• Professional training</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
56	Wylie Burke	Academic, Schools of Public Health	There are three priority areas: 1) Safe and effective uses of genomic information in clinical and public health; 2) Promoting appropriate use of genomics in addressing health disparities; and 3) Promoting research essential to the future of public health. Population genetic variation may lead to a better understanding of population health disparities. The CDC has convening power to develop a multidisciplinary team. Public health genomics research needs to focus on the full range of research strategies addressing the complex interactions between genetic and non-genetic contributors to health, including improved strategies to define phenotypes and social contributors to health; epigenetics; ecogenetics; and microbiome studies.	<ul style="list-style-type: none"> <li>• Evaluation (genomic tests)</li> <li>• Evidence-based genomics</li> <li>• Collaborations/partnerships</li> <li>• Gene-environment interactions/epigenetics</li> <li>• Health disparities</li> <li>• Leadership</li> </ul>
57	Kim Caple	For-profit organization, Biotechnology	The CDC should prioritize the use of next-generation sequencing (NGS) for surveillance and investigation of pathogenic microorganism outbreaks and discuss applications of NGS to understand the dynamics of the human microbiome and influences on human physiology and disease. The CDC, FDA, USDA and state and local public health agencies must work together on these objectives. Large datasets are important in moving to the next step of being able to take findings into translational studies that will influence human health by identifying markers that are predictive of disease.	<ul style="list-style-type: none"> <li>• Genetic technologies</li> <li>• Health applications</li> <li>• Databases</li> <li>• Funding (barrier)</li> </ul>
58	D'Shane Barnett	Non-profit organization, Community-based organization	The most important priorities should be to further Congress' goal of eliminating health disparities within the AI/AN population. Outcomes of genomic knowledge should focus on elimination of health disparities in a number of areas, especially diabetes. Policies must involve AI/AN communities. Funding is a barrier, and so it is important that grant opportunities, technical assistance and outreach are made available to UIHPs.	<ul style="list-style-type: none"> <li>• Health outcomes</li> <li>• Health disparities</li> <li>• Health applications (diabetes)</li> <li>• Funding (barrier)</li> </ul>
59	Jonathan Izant	Non-profit organization, Biotechnology	Public health research needs to adopt new and more cooperative approaches to sharing data and results in order to translate genomic information into public health advances. Integration of genomic datasets to help build accurate and predictive models of disease that can be used to reduce individual and community burden of disease. Research leaders, department chairs, institutional administrators, research funding organizations, government regulatory agencies and publishers need to collaborate.	<ul style="list-style-type: none"> <li>• Databases</li> <li>• Research (translational)</li> <li>• Health outcomes</li> <li>• Collaboration/partnerships</li> <li>• Policymaking (governmental, regulatory agencies, academic)</li> </ul>

Comment #	Name	Respondent Category	Summary	Major Themes
60	Sylvia Au	Government, State; Practice, Public Health	Public health leaders and educators must recognize and embrace importance of genomic knowledge and its appropriate use and population impact for each area of public health (maternal and child health, chronic disease, environmental health, infectious disease). Immediate translation of activities must be ready for public health implementation. National, state and local public and private organizations and agencies, health care providers, policy makers, educators and researches must work together, instead of in silos.	<ul style="list-style-type: none"> <li>• Research (translational)</li> <li>• Utilization</li> <li>• Partnerships/ collaboration</li> <li>• Education (professional)</li> <li>• Funding</li> <li>• Policymaking (governmental)</li> <li>• Silos</li> <li>• Leadership</li> </ul>
61	Claudia Mikail	Non-profit, Health-related	Emphasis should be on the use of pharmacogenomics for treatment and management of chronic diseases, especially for disadvantaged populations. Public health should collaborate with the medical field to build large-scale databases. Implementation of electronic medical records could further facilitate the integration of databases. Health departments, hospitals, academic medical centers, HMOs, community physicians, pharmaceutical companies all need to work together.	<ul style="list-style-type: none"> <li>• Interventions (pharmacogenomics advances)</li> <li>• Collaborations/Partnerships (medicine and public health)</li> <li>• Education (professional)</li> <li>• Interventions (Electronic medical records)</li> </ul>
Mail	Laurie Badzek	Academia, Other Institutions (Nursing)	The most important activities include assessment, education, and evaluation of genomic education for the nursing workforce. Prepare nurses to be able to take family history, develop 3-generation pedigrees, and to know when to refer at risk individuals to genetic services. CDC should engage and fund nursing organizations to develop comprehensive educational programs, which must also be evaluated.	<ul style="list-style-type: none"> <li>• Assessment</li> <li>• Funding</li> <li>• Evaluation</li> <li>• Education (professional)</li> <li>• Professional training</li> <li>• Interventions</li> <li>• Health applications</li> </ul>

## APPENDIX TO PART TWO

## APPENDIX 1: MEETING AGENDA

BETHESDA NORTH MARRIOTT HOTEL AND CONFERENCE CENTER  
SEPTEMBER 14TH, 2011

## Meeting Agenda

- 7:30 Registration and breakfast**
- 8:15 Welcome and Charge**  
Muin J. Khoury, Director of Office of Public Health Genomics, CDC
- 8:35 Where is genomics going in the next decade?**  
Eric Green, Director of NHGRI, NIH
- 8:50 Genetics Services Branch and HRSA: Setting the Context for the Next 10 Years**  
Sara Copeland, Acting Chief, Genetic Services Branch, MCHB, HRSA
- 9:05 Design of the Day**  
Sharon Terry, President & CEO, Genetic Alliance

## What We Learned from the Stakeholder Consultation Process

- 9:10 The public health perspective: Toby Citrin, Director of the Center for Public Health and Community Genomics, University of Michigan
- 9:45 Non-profit & for-profit sector perspective: James O'Leary, Chief Innovative Officer, Genetic Alliance

- 10:00 Charge for Topic Area Teams**  
Sharon Terry, President & CEO, Genetic Alliance

10:15 Break

**10:30 Topic Area Team Sessions Breakout with Facilitators/Animators**

**Prevention:** Identifying individuals, families, and communities at risk

*Co-Facilitators:* Karen Greendale (Invited)  
Ella Green-Moton

**Detection:** Detect diseases early and intervene effectively

*Co-Facilitators:* Deb Duquette  
James O'Leary

**Development & Evaluation:** Advance technology development and evidence generation

*Co-Facilitators:* Sharon Terry  
Adam Berger (Invited)

**Pathways & Interactions:** Understand how pathways and gene-environment interactions impact population health

*Co-Facilitators:* Sara Shostak  
Sharon Kardia

Each group will use the following six questions to guide the discussion:

1. What are the most important activities that should be carried out by the public health system in 2012–2017 to apply genomic knowledge to public health goals?
2. What outcomes specific to public health might be achieved as the result of carrying out these activities?
3. What policies are needed in order to achieve these outcomes?
4. What institutions, organizations and agencies need to participate in achieving these outcomes and what roles should they play?
5. What barriers are anticipated in achieving these outcomes and how might they best be overcome?

**12:30 Networking Lunch**

Facilitators to meet with organizers

**1:30 Topic Area Teams Develop Action Items and Timeline**

Each group will be asked to develop a timetable for the recommended activities and outcomes discussed in the morning session with designated milestones. All teams will consider the following:

- Education
- Workforce
- Community Engagement
- Infrastructure
- Integration
- Funding
- Data Sharing & Dissemination
- Ethical, Legal, and Social Implications

**3:30 Break**

**3:45 Team Presentations and Facilitated Discussion**

**4:45 Concluding Remarks**

Muin J. Khoury, Director of Office of Public Health Genomics, CDC

**5:00 Meeting Adjourn**



**The Regents of the University of Michigan:**

Julia Donovan Darlow, Ann Arbor; Laurence B. Deitch, Bingham Farms; Denise Ilitch, Bingham Farms; Olivia P. Maynard, Goodrich; Andrea Fischer Newman, Ann Arbor; Andrew C. Richner, Grosse Pointe Park; S. Martin Taylor, Grosse Pointe Farms; Katherine E. White, Ann Arbor; Mary Sue Coleman, ex officio

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