



Genetics Education and Training

Report of the
Secretary's Advisory Committee on Genetics, Health, and Society

February 2011

A PDF version of this report is available at
http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_education_report_2011.pdf.



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February 1, 2011

The Honorable Kathleen Sebelius
Secretary of Health and Human Services
200 Independence Avenue, SW
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Dear Secretary Sebelius:

In keeping with its mandate to provide advice on a broad range of policy issues raised by the development and use of genetic technologies, the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) has prepared the report *Genetics Education and Training*. The report describes the genetics education and training needs of point-of-care health professionals, the public health workforce, and patients and consumers and provides six recommendations to address these needs. Implementation of these recommendations would advance access to and use of beneficial genetic information and services by ensuring that clinical and public health professionals and the public have the educational tools and resources to understand, interpret, and appropriately apply such information.

The report is the culmination of 3 years of factfinding, public consultation, analysis, and deliberation by the Committee. In conducting its work, SACGHS used the following methods: (1) a review of published and unpublished research relevant to professional and public education and training in genetics; and (2) surveys of selected health professional organizations and individuals with responsibilities across the continuum of health professional education and public health; and (3) interviews with experts in genetics education and advocacy outreach.

The six recommendations provided in the report urge the Department of Health and Human Services (HHS) to take the following steps to improve genetics education and training, which will help assure the appropriate, effective, and efficient integration of genetic and genomic technologies and services into the health system and equitable access to those technologies and services:

- Convene a task force to identify innovative approaches to prepare health professionals for the genomic age and mechanisms to assure the incorporation of genetic content in

electronic health records, credentialing exams, accreditation of institutions, and continuing education activities

- Evaluate the composition of the public health workforce to identify future education and training needs of those with responsibilities related to genetics and fund the development and implementation of programs that address these needs
- Support programs that increase the diversity and genetic competencies of the health care workforce in underserved communities and ensure that consumer and patient educational materials are culturally and linguistically tailored to the unique needs of the community
- Identify effective communication strategies for translating genetic knowledge into information that consumers and patients can use to make health decisions and develop educational programs that use a wide array of media and community-based learning
- Create and maintain a state-of-the art Internet portal to facilitate access to comprehensive, accessible, and trustworthy web-based genetic information and resources for consumers
- Improve and promote the use of family history tools

Since the SACGHS charter ends February 28, 2011, responsibility for monitoring the implementation of these recommendations should be assumed by relevant HHS agencies such as the Agency for Healthcare Research and Quality, Centers for Disease Control and Prevention, and the National Institutes of Health and steps taken to formulate and track success measures of any programs instituted or funded as a result of these recommendations. The Committee also proposes that the state of genetics education and training be assessed within 5 years to ensure that Federal efforts continue to reflect the diverse and unique needs of health care and public health professionals and the public.

SACGHS has appreciated the opportunity to be of service to you and hopes that its final report *Genetics Education and Training* will prove helpful to you and the Department.

Sincerely,

A handwritten signature in black ink that reads "Steven Teutsch". The signature is written in a cursive, flowing style.

Steven Teutsch, M.D., M.P.H.
Chair, SACGHS

About SACGHS

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) was first chartered in 2002 by the Secretary of Health and Human Services (HHS) as a public forum for deliberation on the broad range of policy issues raised by the development and use of genetic tests and, as warranted, to provide advice on these issues. The charter set out the following specific functions of the Committee:

- Assessing how genetic and genomic technologies are being integrated into health care and public health;
- Studying the clinical, public health, ethical, economic, legal, and societal implications of genetic and genomic technologies and applications;
- Identifying opportunities and gaps in research and in data collection and analysis efforts;
- Examining the impact of current patent policy and licensing practices on access to genetic and genomic technologies;
- Analyzing uses of genetic information in education, employment, insurance, and law; and
- Serving as a public forum for discussion of issues raised by genetic and genomic technologies.

SACGHS held its last meeting in October 2010. Based on the Committee's accomplishments in addressing all the major areas outlined in its charter, SACGHS will sunset February 28, 2011. Over its tenure, the Committee provided advice on a range of complex issues raised by new technological developments in human genetics and produced a body of work that will provide a lasting framework for addressing new developments in the ongoing integration of genetics into clinical practice and public health.

Structurally, SACGHS consisted of up to 17 individuals from around the Nation who have expertise in disciplines relevant to genetics and genetic technologies. These disciplines included biomedical sciences, human genetics, health care delivery, evidence-based practice, public health, bioinformatics, behavioral sciences, social sciences, health services research, health policy, health disparities, ethics, economics, law, health care financing, consumer issues, and other relevant fields. At least two of the members are specifically selected for their knowledge of consumer issues and concerns and the views and perspectives of the general public.

Throughout the course of the Committee's work, representatives of at least 19 Federal departments or agencies also participated in SACGHS in an *ex officio* (nonvoting) capacity. The departments were the Department of Commerce, Department of Defense, Department of Education, Department of Energy, Department of Justice, Department of Labor, Department of Veterans Affairs, Equal Employment Opportunity Commission, Federal Trade Commission, and the following HHS agencies: Administration for Children and Families, Agency for Healthcare Research and Quality, Centers for Disease Control and Prevention, Centers for Medicare &

Medicaid Services, Food and Drug Administration, Health Resources and Services Administration, National Institutes of Health, Office for Civil Rights, Office for Human Research Protections, and Office of Public Health and Science.

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The Committee is also indebted to all of the members of the Task Force for devoting time to the project and sharing their knowledge and expertise on genetics education and training: current and former SACGHS members—Sylvia Mann Au, David Dale, Gwen Darien, James Evans, Barbara Burns McGrath, the late Paul Miller, Joseph Telfair, Marc Williams, and Paul Wise—representatives from federal agencies—Gurvaneet Randhawa (Agency for Healthcare Research and Quality); Muin Khoury and Katherine Kolor (Centers for Disease Control and Prevention); Denise Geolot (Health Resources and Services Administration); Vence Bonham, Kathleen Calzone, W. Gregory Feero, Sarah Harding, Jean Jenkins, and Emma Kurnat-Thoma (National Institutes of Health)—and *ad hoc* members—Judith Benkendorf (American College of Medical Genetics), Joann Boughman (American Society of Human Genetics), Katherine Johansen Taber (American Medical Association), Scott McLean (DNA Direct), Kate Reed (National Coalition for Health Professional Education in Genetics).

The Committee thanks all of the individuals and organizations who responded to the Committee's requests for public comments during the development of this report (see Appendix A). The Committee gave careful consideration to each of the comments. The public's input enhanced the report's analysis and the relevance of the recommendations. SACGHS is also aware of the work of the Secretary's Advisory Committee on Heritable Disorders of Newborns and Children on this topic and appreciated the opportunity to exchange ideas and perspectives with its members.

The Committee also wishes to thank the SACGHS staff for organizing the deliberations of the Task Force and managing the development of the report; Sarah Carr, SACGHS Executive Secretary, for her overall guidance; and the NIH Office of Biotechnology Activities, under the direction of Amy Patterson, for its support and operational management of SACGHS.

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Preface

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) has consistently recognized the importance of professional and public genetics education and 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.¹ SACGHS ranked professional and public education as a high priority in its 2004 priority setting process,² and the Committee issued a formal resolution that was conveyed to the Secretary of Health and Human Services.³ The resolution emphasized the critical importance of appropriate and adequate genetics training and education for health care professionals and the public. In November 2007, SACGHS convened a roundtable to assess the need for a task force on genetics education and training to build on the Committee's earlier work.⁴ In its 2008 priority setting process, the Committee again ranked professional and public genetics education as a high priority.⁵

SACGHS formed the Genetics Education and Training Task Force to identify education and training issues pertinent to (1) point-of-care health professionals with and without expertise in genetics (e.g., primary care professionals such as pediatricians, family practice physicians, obstetrician/gynecologists, and internists, nurses, physician assistants, genetic counselors, and pharmacists), (2) public health providers involved or likely to be involved in providing genetic services, and (3) consumers and patients. Given the rapid proliferation of genetic technologies and the shift toward personalized health care, the Committee determined that the education and training needs of health care professionals working on the front lines of public health and health care delivery were of high priority, as was the need for an informed public. This report's findings and recommendations may also apply to a broader constituency, such as laboratory professionals, health care administrators, payers, policymakers, and lay health providers as well as librarians, judges, law enforcement agents, clergy, science educators, journalists, policy makers, and health care governing bodies.

To conduct its work, the Task Force divided into three workgroups to explore the education needs of these three broad communities (health care professionals, public health providers, and

¹ Secretary's Advisory Committee on Genetics, Health, and Society. SACGHS Documents, Reports, and Correspondence. See http://oba.od.nih.gov/SACGHS/sacghs_documents.html. Accessed on November 24, 2009.

² Secretary's Advisory Committee on Genetics, Health, and Society. (2004). *A Roadmap for the Integration of Genetics and Genomics into Health and Society: The Study Priorities of the Secretary's Advisory Committee on Genetics, Health, and Society*. See <http://oba.od.nih.gov/oba/sacghs/reports/SACGHSPriorities.pdf>. Accessed on November 24, 2009.

³ Secretary's Advisory Committee on Genetics, Health, and Society. (2004). Resolution of the Secretary's Advisory Committee on Genetics, Health, and Society on Genetics Education and Training of Health Professionals. See http://oba.od.nih.gov/SACGHS/sacghs_documents.html. Accessed on February 24, 2010.

⁴ Secretary's Advisory Committee on Genetics, Health, and Society. (2007). Roundtable on Genetics Education and Training of Health Professionals, November 20, 2007. See http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_2007_nov_20.html. Accessed on January 14, 2010.

⁵ Secretary's Advisory Committee on Genetics, Health, and Society. (2009). *The Integration of Genetic Technologies Into Health Care and Public Health: A Progress Report and Future Directions of the Secretary's Advisory Committee on Genetics, Health, and Society*. See http://oba.od.nih.gov/sacghs/sacghs_documents.html. Accessed on November 24, 2009.

consumers and patients). Three methods were used to gather information to inform this report: (1) a review of published and unpublished research relevant to professional and public education and training in genetics; and (2) surveys of selected health professional organizations and individuals with responsibilities across the continuum of health professional education and public health; and (3) interviews with experts in genetics education and advocacy outreach.

At the October 2009 SACGHS meeting, the Committee reviewed the findings of the Task Force's information-gathering efforts and discussed preliminary draft recommendations for the genetics education and training report.⁶ In December 2009, the Task Force held a daylong meeting to revise the recommendations based on the Committee's input and continued its work in developing the report. SACGHS discussed and approved the draft report for public consultation at its February 2010 meeting,⁷ and the report was released for public comment from May 24, 2010, to June 30, 2010.

The public was invited to comment on the draft report through announcements in the Federal Register,⁸ on the SACGHS website, and disseminated through the SACGHS listserv. The Committee received 35 responses to this request from a range of individuals and organizations (see Appendix A for the list of commenters). The comments were carefully reviewed by the Task Force and SACGHS staff. On July 21, 2010, the Task Force held a conference call to discuss how to address each comment. Based on this discussion, the report and recommendations were revised for presentation at the Committee's October 2010 meeting. The revised report reflects the cumulative work of SACGHS, the Task Force, SACGHS staff, and the insightful comments of expert presenters, interviewed stakeholders, and the public.

⁶ Secretary's Advisory Committee on Genetics, Health, and Society. SACGHS Task Force on Genetics Education and Training Findings and Recommendations, October 9, 2010. See <http://oba.od.nih.gov/oba/SACGHS/meetings/October2009/BBMcGrath%20ET%20TF%20session%2010-09-09%20FINAL%2010-7-09.pdf>. Accessed on September 14, 2010.

⁷ Secretary's Advisory Committee on Genetics, Health, and Society. SACGHS Draft Report and Recommendations on Genetics Education and Training, February 4, 2010. See <http://oba.od.nih.gov/oba/SACGHS/meetings/feb2010/BBMcGrath%20ET%20TF%20session%20Feb%204-10%20FINAL.pdf>. Accessed on September 14, 2010.

⁸ Notice of Request for Public Comment. *Federal Register*. May 24, 2010. 75(99): 28809. See <http://edocket.access.gpo.gov/2010/pdf/2010-12450.pdf>. Accessed on September 14, 2010.

Executive Summary

Over the last decade, technological advances and improved knowledge of the genetic underpinnings of disease have led to a dramatic increase in the number of clinically available genetic tests.⁹ Although most of these tests identify mutations in single-gene disorders, those aimed at identifying susceptibility gene mutations for common, chronic diseases and conditions are becoming increasingly available in clinical and commercial settings. The growing integration of genetics and genomics findings into mainstream medicine and the emergence of direct-to-consumer genetic testing amplify the need for understanding risk assessment, multi-gene and genomic diagnostics, genetic-based treatment, and effective strategies in communicating genetic test results to patients and consumers. However, health care professionals, the public health workforce, and patients and consumers are challenged to keep pace with this dynamic and rapidly evolving field. Genetics education and training are critical to realizing the benefits of genetic technologies and guarding against the potential for harm.

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) has consistently recognized the importance of professional and public genetics education and training to assure that genomic research findings benefit the public's health. The purpose of this report is to identify the education and training needs of point-of-care health care professionals, public health providers, and consumers and patients and to provide recommendations that address these needs. Three methods were used to gather information to inform this report: (1) a review of published and unpublished research relevant to professional and public education and training in genetics; (2) surveys of selected organizations, groups, and individuals with responsibilities across the continuum of health professional and public health education and training; and (3) interviews with experts in genetics education and advocacy outreach.

It is important to acknowledge the many genetics education programs and resources developed over several decades by Federal and State Governments as well as efforts in the private sector by health professional and patient advocacy organizations and others. Significant challenges remain, however, in integrating genetics education across learning environments for health care professionals, identifying education needs among the large and varied public health workforce, and meeting the needs of consumers given the diversity of the American public. Thus, a new model for applying genetics to improve health requires a system in which health care professionals, public health providers, and consumers are well informed and able to interact with each other as appropriate. Cooperation and collaboration in processing, interpreting, and applying genetic information will be essential. Without these efforts, society will not benefit from genetic advances, opportunities will be lost for deploying prevention and early detection programs for a wide variety of chronic diseases, and patients and consumers may make poorly informed choices or fail to seek needed professional health services.

SACGHS presents six recommendations that address the identified genetics education and training needs of health care professionals, public health providers, and consumers and patients.

⁹ GeneTests. See <http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/whatsnew/labdirgrowth.shtml>. Accessed on December 13, 2010.

Recommendation 1

Evidence from the United States and abroad suggests inadequate genetics education of health care professionals as a significant factor limiting the integration of genetics into clinical care. Specific inadequacies include the amount and type of genetics content included in undergraduate professional school curricula and the small amount of genetics-related knowledge and skills of physicians, nurses, and other health professionals once they enter clinical practice. Modifications in medical, dental, nursing, public health, and pharmacy school curricula and in medical residency training programs are needed to ensure that health care professionals entering the workforce are well-trained in genetics.

1. Innovative approaches that coordinate the efforts of entities involved in health professional education and training are required to address these gaps. Therefore, HHS should convene a task force of stakeholders to identify:
 - A. Outcomes-based education and training guidelines and models;
 - B. Best practices for enhancing and expanding the content needed to prepare health care professionals for personalized genomic health care;
 - C. Mechanisms to assure the incorporation of up-to-date genetic content in standards, certification, accreditation, electronic health records, and continuing education activities; and
 - D. Funding sources for developing and promoting genetics education for relevant health care professionals.

Recommendation 2

The inherent diversity of the public health workforce makes it difficult to target educational efforts that are relevant across groups. A systematic effort is needed to evaluate the composition of the public health workforce with current job responsibilities related to genetics and genomics and to identify future priorities, such as the potential impact of affordable genomic analysis.

2. HHS and its public health agencies should:
 - A. Assess the public health workforce to determine the number and type of public health providers with responsibilities in genetics and genomics and to ascertain current trends and future education and training needs;
 - B. Identify and engage exemplary public health genomic programs to identify critical workforce information not captured in the assessment; and
 - C. Using the results of these assessments and to address identified gaps, HHS should:
 - Support development of skills, competencies, and leadership in genetics and genomics that specifically address the identified needs; and
 - Based on these skills and competencies, fund the development and implementation of accessible educational programs and continuing education in genetics and genomics for the public health workforce.

Recommendation 3

Findings in the literature and SACGHS surveys indicate that health care professionals and public health providers serving underserved and underrepresented groups and populations face significant challenges.

3. To increase services and access to care in underserved communities, HHS should:
 - A. Identify existing effective educational models for health care professionals and public health providers in underserved communities;
 - B. Identify and support programs to increase the diversity and genetic competencies of the health care workforce serving underserved communities; and
 - C. Incentivize organizations and ensure that consumers and representatives of rural, minority, and underserved communities participate in the process of developing education and training models and materials. Assure that these materials are culturally and linguistically appropriate and tailored to the unique needs of these diverse communities.

Recommendation 4

With the vast increase in scientific knowledge stemming from genetics research, the development of new technologies, and the increase in direct-to-consumer genetic services, educational efforts are needed to translate this information to reach consumers of all literacy levels.

4. HHS should identify effective communication strategies for translating genetics knowledge into information that consumers and patients can use to make health decisions. Specifically, HHS should:
 - A. Support multidisciplinary research that identifies effective methods of patient and consumer communication;
 - B. Based on this research, and to reach diverse people and communities, HHS should develop educational programs that use a wide array of media and community-based learning and provide culturally and linguistically appropriate materials; and
 - C. In collaboration with the Department of Education and the National Science Foundation, support the incorporation of genetics and genomics in K-12 education.

Recommendation 5

A significant amount of genetic-related information directed to consumers and patients exists in a variety of formats and from a number of sources, but the quality of the content is variable. Consumers have consistently expressed the desire for accessible, web-based genetic information that they can trust and consider provision of these resources as a role of the Federal Government.

5. HHS should create and maintain a state-of-the-art Internet portal to facilitate access to comprehensive, accessible, and trustworthy web-based genetic information and resources for consumers.

Recommendation 6

6. Because family health history tools are a potentially powerful asset for consumers and health care professionals to use in risk assessment and health promotion, HHS should:
 - A. Support efforts to educate health care professionals, public health providers, and consumers about the importance of family health history;
 - B. Promote research on how consumers and diverse communities use family history to make health care decisions and incorporate those research findings into consumer educational materials;
 - C. Support the use of family history in clinical care through development of point-of-care educational materials and clinical decision support tools in electronic health records that utilize coded and computable family history, genetic, and genomic information; and
 - D. Promote embedding educational materials in family history collection tools and personal health records directed to consumers and ensure for all by providing these tools in various formats.

Implementation of these recommendations should help advance access to and use of beneficial genetic information and services by ensuring that health care providers, the public health workforce, and patients and consumers have the educational tools and capacities to understand, interpret, and appropriately apply such information. As the advancement of genetic technologies accelerates, so must the education and training efforts.

I. Introduction

A. THE IMPORTANCE OF GENETICS AND GENOMICS IN HEALTH CARE

The Human Genome Project, completed in 2003, resulted in a delineation of a complete human genome sequence, advancing research of the genetic¹⁰ basis of disease. It is estimated that nine out of 10 of the top leading causes of death in the United States have a genetic component.¹¹ Improved genetic knowledge and advanced technologies are leading to new diagnostic approaches for common chronic diseases and conditions.¹² Over the last decade, these technologies have led to a dramatic increase in the number of genetic tests available for screening and diagnostic purposes.⁹ Although most of these tests identify mutations that cause single-gene disorders, tests for genetic variants that indicate the risk of common diseases are becoming increasingly available in the clinical setting, and some are marketed directly to consumers.

Advances in identifying the genetic underpinnings of chronic disease are changing the approaches to disease prevention and treatment.¹³ For example, stratification by genotype or family history provides a means for tailoring screening tests for early detection of certain types of cancer, and this paradigm is likely to be extended to early detection of other conditions.¹⁴ Pharmacogenomics—the study of how genetic differences affect drug response—is being applied to select appropriate drug therapies and guide dosing. Genetic biomarkers can play an important role in identifying responders and nonresponders to avoid toxicity and optimize the efficacy and safety of drug therapies.¹⁵

The field of medical genetics is in a transition period from its role in the health care of a small percentage of people with rare genetic disorders to the delivery of effective health care for everyone.¹⁶ Genetics has and will continue to be applied in the clinical setting in the context of individual, rare, single-gene disorders. However, to realize the full potential of genomics, the complex relationships among genetic variation, the environment, and disease must be considered, which could lead to diagnostics and therapies for complex, common disorders such as cancer, heart disease, diabetes, and mental illness. Realizing this potential will require a

¹⁰ The term “genetics” commonly refers to the actions of single genes, whereas the term “genomics” refers to the complete DNA sequence of an organism. In this report, for ease of reading, the Committee often defaults to the term “genetics” to encompass both genetics and genomics. When the distinctions are critical, the applicable term is used.

¹¹ Center for Disease Control and Prevention. Public Health Genomics. *Genomics and Health*. See <http://www.cdc.gov/genomics/public/index.htm>. Accessed on June 23, 2010.

¹² Wellcome Trust Case Control Consortium. (2007). Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. *Nature*. 447(7145):661-678.

¹³ Feero, W.G., Guttmacher, A.E., and Collins, F.S. (2008). The genome gets personal—almost. *Journal of the American Medical Association*. 299(11):1351-1352.

¹⁴ Khoury, M.J., Gwinn, M., Burke, W., Bowen, S., and Zimmern, R. (2007). Will genomics widen or help heal the schism between medicine and public health? *American Journal of Preventive Medicine*. 33(4):310-317.

¹⁵ Gervasini G., Benitez, J., and Carrillo, J.A. (2010). Pharmacogenetic testing and therapeutic drug monitoring are complementary tools for optimal individualization of drug therapy. *European Journal of Clinical Pharmacology*. 66(8): 755-774.

¹⁶ Guttmacher, A.E. and Collins, F.S. (2002). Genomic medicine: a primer. *New England Journal of Medicine*. 347(19): 1512-1520.

population focus, not only for research, but also in designing strategies to interpret and use genetic and genomic information in community and home-based settings.¹⁷

The emerging understanding of the role of genetics in common disease is increasing the need for knowledge of risk assessment, diagnostics, appropriate treatment approaches, and communication in professional and public education. Patients and consumers, health care professionals, and public health officials are challenged to keep pace with this dynamic and rapidly evolving field. Additionally, the growth of direct-to-consumer (DTC) genetic services highlights the importance of adequate education for consumers to ensure informed decisionmaking. Educating health care professionals, the public health workforce, and the general public is critical to realizing the benefits of genetic technologies and guarding against the potential for harm.

B. TECHNOLOGICAL ADVANCES IN A RAPIDLY EVOLVING FIELD

Research efforts provide increasing opportunities to translate genetic technologies into clinical use to prevent and treat common, chronic diseases. Recent translational research initiatives include the Encyclopedia of DNA Elements (ENCODE)¹⁸—a public research consortium to identify all functional elements in the human genome sequence—the 1000 Genomes Project,¹⁹ the Electronic Medical Records and Genomics (eMERGE) Network,²⁰ and the Cancer Genome Atlas.

Technological advances have also dramatically reduced the cost of sequencing individual genomes. Within the last decade, the price has dropped from \$300 million to less than \$5,000.^{21,22} As the cost continues to decrease and becomes comparable to other medical tests and procedures, the demand for whole-genome sequencing is likely to increase.²³ Presumably, an individual's germline genome would only need to be sequenced once in a lifetime,²⁴ and the information would then be available to assist in all future decisionmaking about medical care.

¹⁷ Khoury, M.J. (2003). Genetics and genomics in practice: the continuum from genetic disease to genetic information in health and disease. *Genetics in Medicine*. 5(4):261-268.

¹⁸ National Human Genome Research Institute. The ENCODE Project: ENCyclopedia Of DNA Elements. See <http://www.genome.gov/10005107>. Accessed on September 14, 2010.

¹⁹ 1000 Genomes Project. See <http://www.1000genomes.org>. Accessed on November 24, 2009.

²⁰ The electronic Medical Records and Genomics (eMERGE) Network. See <https://www.mc.vanderbilt.edu/victr/dcc/projects/acc/index.php/About>. Accessed September 2, 2009.

²¹ Drmanac, R., Sparks, A.B., Callow, M.J., Halpern, A.L., Burns, N.L., Kermani, B.G., Carnevali, P., Nazarenko, I., Nilsen, G.B., Yeung, G., Dahl, F., Fernandez, A., Staker, B., Pant, K.P., Baccash, J., Borcharding, A.P., Brownley, A., Cedeno, R., Chen, L., Chernikoff, D., Cheung, A., Chirita, R., Curson, B., Ebert, J.C., Hacker, C.R., Hartlage, R., Hauser, B., Huang, S., Jiang, Y., Karpinchyk, V., Koenig, M., Kong, C., Landers, T., Le, C., Liu, J., McBride, C.E., Morenzoni, M., Morey, R.E., Mutch, K., Perazich, H., Perry, K., Peters, B.A., Peterson, J., Pethiyagoda, C.L., Pothuraju, K., Richter, C., Rosenbaum, A.M., Roy, S., Shafto, J., Sharanhovich, U., Shannon, K.W., Sheppy, C.G., Sun, M., Thakuria, J.V., Tran, A., Vu, D., Zaranek, A.W., Wu, X., Drmanac, S., Oliphant, A.R., Banyai, W.C., Martin, B., Ballinger, D.G., Church, G.M., and Reid, C.A. (2010). Human genome sequencing using unchained base reads on self-assembling DNA nanoarrays. *Science*. 327(5961): 78-81.

²² "Complete Genomics Gets Gene Sequencing Under \$5,000" November 5, 2009. See <http://www.bloomberg.com/apps/news?pid=20601124&sid=aWutnyE4SoWw>. Accessed on November 24, 2009.

²³ Wolinsky, H. (2007). The thousand-dollar genome: Genetic brinksmanship or personalized medicine? *EMBO Reports*. 8(10):900-903.

²⁴ Shirts, B.H. and Parker, L.S. (2008). Changing interpretations, stable genes: responsibilities of patients, professionals, and policy makers in the clinical interpretation of complex genetic information. *Genetics in Medicine*. 10(11):778-783.

The vast amount of data resulting from whole-genome sequencing will require new capacities for data management, interpretation, and protections. If whole-genome sequencing data are to be used in the clinical setting and in research, bioinformatic tools that link genomic data with electronic health records (EHRs) will be required.²⁵ Consideration must also be given to the complexities related to sequencing methodologies, reinterpretation of genomic data as research reveals new genotype-phenotype associations, appropriate communication to patients about genetic variants of unknown clinical significance, and assurance that health care professionals are properly trained to interpret complex or inconclusive genomic information.^{24,26,27}

C. EMERGING GENETIC TECHNOLOGIES AND HEALTH DISPARITIES

Numerous reports have documented the extent of health disparities in the United States, and the field of genetics is no exception.²⁸ When any new technology emerges it has the potential to exacerbate disparities if patients and providers do not have access to the newly developed resource or relevant information about the use of new technology or, in the case of genetics and genomics, about the meaning of diagnostic test results.

Although studies have documented disparities in access to genetic services, other research suggests that genomics and personalized medicine may help address disparities. For example, the increasingly widespread use of the Internet provides a means of democratizing access to personal genomic information.²⁹ Initial experiences of clients using online genomic services could be used to pilot more effective and direct ways of delivering genetics education and counseling services to much larger numbers of people than are currently seen by genetic counselors or clinical geneticists.²⁹ As whole-genome sequencing becomes more affordable, the technology has the potential to address disparities, especially if used as part of a public health mandate such as newborn screening. Efforts are needed to assure equitable access to this new technology as it is folded into clinical testing and to provide educational resources and services that help patients understand genomic test results.

Genomic medicine can also help close the gap in health disparities by expanding the knowledge of novel alleles related to disease and by using this information to replace imprecise surrogate biomarkers based on race, ethnicity, or ancestry to identify risks for disease or adverse drug response.³⁰ However, genomic information is not sufficient in the personalization of medicine.³¹ As Isaac Kohane notes, personalized medicine also requires a robust understanding of the subpopulation that most resembles the patient and should extend to observable factors such as environmental exposures, cultural practices, and access to medical services.³¹ These factors may

²⁵ Nelson, E.A. and McGuire, A.L. The need for medical education reform: genomics and the changing nature of health information. (2010). *Genomic Medicine*. 2:18-20.

²⁶ Ormond, K.E., Wheeler, M.T., Hudgins, L., Klein, T.E., Butte, At. J., Altman, R.B., Ashley, E.A., and Greely, H.T. (2010). Challenges in the clinical application of whole-genome sequencing. *The Lancet*. 375:1749-1751.

²⁷ Mardis, E.R. (2006). Anticipating the \$1,000 genome. *Genome Biology*. 7(7):112.

²⁸ Hall, M. and Olopade, O.I. (2005). Confronting genetic testing disparities. *Journal of the American Medical Association*. 293(14):1783-1785.

²⁹ Foster, M.W. and Sharp, R.R. (2008). Out of sequence: how consumer genomics could displace clinical genetics. *Nature Review Genetics*. 9(6):419.

³⁰ Gurwitz, D. and Lunshof, J.E. (2009). Ancestry in translational genomic medicine: handle with care. *Genome Medicine*. 25;1(2):24.

³¹ Kohane, I.S. (2009). The twin questions of personalized medicine: who are you and whom do you most resemble? *Genome Medicine*. 20;1(1):4.

be just as important as a shared genetic background for discerning risk. When psychosocial factors are combined with a better understanding of the degree of genetic variation within racial and ethnic groups, genomic studies can move beyond classifying and treating ethnic and racial subpopulations as homogeneous groups.

D. PURPOSE AND SCOPE OF THIS REPORT

The trends discussed above—the growing application of genetic information to health care and personal decisionmaking, the rapidly developing technical capabilities in the field, and the potential to exacerbate or reduce health disparities—call for improved education and training at all levels. The purpose of this report is to identify the education and training needs of point-of-care health care professionals, public health providers, and consumers and patients and to provide recommendations that address these needs.

It is important to acknowledge the many genetics education programs and resources developed over several decades by Federal and State Governments and efforts in the private sector by health professional and patient advocacy organizations and others. Significant challenges remain, however, in integrating genetics education across learning environments for health care professionals, identifying education needs among the large and varied public health workforce, and meeting the needs of consumers given the diversity of the American public.

SACGHS also recognized that there are pertinent issues beyond education and training that influence the use of genetic technologies to improve the public's health. As the clinical utility of genetic tests and services is demonstrated over time, health care professionals will be more likely to see the need to incorporate genetics into their practice. Coverage and reimbursement of genetic tests and services influence the use of such services in clinical practice and therefore may be an important priority for policymakers. New genetic technologies also have the potential to decrease health disparities, but continuing work is needed to assure that underserved populations have access to appropriate genetic services and information.

The challenge to public and private entities will be to develop targeted genetics education and training programs that lead to improved health. Innovative approaches that support health care professionals at the point of care, such as decision support tools imbedded in EHRs, will be increasingly useful mechanisms for professional education. Culturally and linguistically appropriate educational materials will help the public health workforce provide genetic services to the communities they serve. Emerging methods to communicate genetics information to consumers and patients, such as mobile health technologies, will further facilitate dialogue and information sharing. As the advancement of genetic technologies accelerates, so must the education and training efforts.

II. Background

A. GENETICS EDUCATION AND TRAINING LITERATURE REVIEW

To help inform the education and training needs of health care professionals, public health providers, and the public, a literature search was conducted simultaneously in 10 databases via the DIALOG platform for the years 2003 to 2009, covering the fields of medicine, science, education, social science, and psychology. (See Appendix B for details of the search methodology.) Additional literature was reviewed as it became available in 2010, including peer-reviewed studies and nonpeer-reviewed materials such as relevant news items and commentaries.

Health Care Professionals

Health care professionals, particularly those working at the point of care (e.g., physicians, nurses, physician assistants, genetic counselors, and pharmacists) are key to the successful translation of new genetic knowledge into practice by promoting access to, and appropriate use of, genetic technologies. This translation requires a workforce that is adequately trained and educated in genetics. A significant body of literature, however, highlights several factors that contribute to the limited genetics education of health care professionals and the poor integration of genetics into health care.^{32,33,34,35,36,37,38,39,40,41,42,43,44,45,46} These factors include crowded curricula; lack of

³² Guttmacher, A.E., Porteous, M.E., and McInerney, J.D. (2007). Educating health-care professionals about genetics and genomics. *Nature Reviews*. 8:151-157.

³³ Harris, J.N., Bowen, D.J., Kuniyuki, A., McIntosh, L., FitzGerald, L.M., Ostrander, E.A., and Stanford, J. L. (2009). Interest in genetic testing among affected men from hereditary prostate cancer families and their unaffected male relatives. *Genetics in Medicine*. 11(5):1-12.

³⁴ Benjamin, C.M., Anionwu, E.N., Kristoffersson, U., ten Kate, L.P., Plass, A.M., Nippert, I., Julian-Revnier, C., Harris, H.J., Schmidtke, J., Challen, K., Calefato, J.M., Waterman, C., Powell, E., and Harris, R., on behalf of the GenEd Research Group. (2009). Educational priorities and current involvement in genetic practice: a survey of midwives in the Netherlands, UK and Sweden. *Midwifery*. 25(5):483-499.

³⁵ Burke, S., Martyn, M., Thomas, H., and Farndon, P. (2009). The development of core learning outcomes relevant to clinical practice: identifying priority areas for genetics education for non-genetics specialist registrars. *Clinical Medicine*. 9:49-52.

³⁶ Burke, S., Martyn, M., Stone, A., Bennett, C., Thomas, H., and Farndon, P. (2009). Developing a curriculum statement based on clinical practice: genetics in primary care. *British Journal of General Practice*. 59:99-103.

³⁷ Little, J., Potter, B., Allanson, J., Caulfield, J., Carroll, J.D., and Wilson, B. (2009). Canada: public health genomics. *Public Health Genomics*. 12:112-120.

³⁸ Metcalfe, S.A., Bittles, A.H., O'Leary, P., and Emery, J. (2009). Australia: public health genomics. *Public Health Genomics*. 12:121-128.

³⁹ Pestka, E.L., and Williams, J.K. (2005). International collaboration on genomics education for nurses. *Journal of Continuing Education in Nursing*. 36(4):180-184.

⁴⁰ Tomatir, A.G., Sorkun, H.C., Demirhan, H., and Akdaq, B. (2007). Genetics and genetic counseling: practices and opinions of primary care physicians in Turkey. *Genetics in Medicine*. 9(2):130-135.

⁴¹ Kiray, V.B., Tomatir, A.G., Kuzu, K.N., and Taspinar, A. (2009). Nursing students' self-reported knowledge of genetics and genetic education. *Public Health Genomics*. 12(4):225-232.

⁴² McInerney, J.D. (2008). Genetics education for health professionals: a context. *Journal of Genetic Counseling*. 17:145-151.

⁴³ Greb, A.E., Brennan, S., McParlane, L., Rage, R., and Bridge, P.D. (2009). Retention of medical genetics knowledge and skills by medical students. *Genetics in Medicine*. 11(5):1-6.

⁴⁴ Thurston, V.C., Wales, P.S., Bell, M.A., Torbeck, L., and Brokaw, J.J. (2007). The current status of medical genetics instruction in US and Canadian medical schools. *Academic Medicine*. 82(5):441-445.

⁴⁵ Hetteberg, C.G., Prows, C.A., Deets, C., Monsen, R.B., and Kenner, C.A. (1999). National survey of genetics content in basic nursing preparatory programs in the United States. *Nursing Outlook*. 47(4):168-180.

⁴⁶ Murphy, J.E., Green, J.S., Adams, L.A., Squire, R.B., Kuo, G.M., and McKay, A. (2010). Pharmacogenomics in the curricula of colleges and schools of pharmacy in the United States. *American Journal of Pharmaceutical Education*. 74(1):1-10.

knowledgeable faculty; genetics content that is not presented in a way that leads to long-term knowledge retention; failure to incorporate genetics into clinical training; inadequate representation of genetics on certifying exams; lack of evidence-based guidelines in genetics; and misconceptions that genetic medicine is defined by rare, Mendelian disorders, when in fact genetics is increasingly concerned with the common, chronic diseases that are the daily focus for most health professionals.

Innovative and experimental models show promise in improving genetics education. For example, the “Genetics in Primary Care” Faculty Development Initiative is notable for its collaboration across education, genetics, and primary care experts, who designed curricula and case studies to provide a standardized genetics instruction format that also incorporates evidence and assessment skills for newly released scientific findings.⁴⁷ A follow-up evaluation of the program was conducted four years after its initiation to assess the long-term effects on participants’ teaching and clinical practices.⁴⁸ Follow-up data from 19 of the 20 participating institutions indicated that the program promoted long-term behavioral changes and comfort with genetics. All 19 institutes reported changes in teaching practices (e.g., the addition of new genetics material to medical school curriculum and/or residency programs) and formalization of teaching collaborations between geneticists and primary care faculty. Changes in clinical practices among primary care physicians (e.g., changes in genetic services referral patterns and greater importance given to family history) were reported by half of the institutes. Although the initiative was considered successful, participants reported challenges such as convincing other primary care providers of the relevance of genetics in their practice and finding the time and resources to sustain and adapt the program over time.

Attitudes and Working Knowledge Levels of Health Care Professionals Regarding Genetics

The goal of incorporating genetic knowledge into clinical practice is not new.^{49,50} Shortcomings in genetics knowledge and use of genetic tests have been noted for several decades, and the need for integrated genetic instruction across curricula of all health care subspecialties has been advocated for some time.⁵¹ Yet in the United States, health care professionals across a wide range of clinical specialties demonstrate lack of genetics knowledge.^{52,53,54,55,56,57,58} For example,

⁴⁷ Burke, W., Acheson, L., Botkin, J., Bridges, K., Davis, A., Evans, J., Frias, J., Hanson, J., Kahn, N., Kahn, R., Lanier, D., Pinsky, L.E., Press, N., Lloyd-Puryear, M.A., Rich, E., Stevens, N., Thomson, E., Wartman S., and Wilson, M. (2002). Genetics in primary care: a USA faculty development initiative. *Community Genetics*, 5(2): 138-146.

⁴⁸ Laberge, A.M., Fryer-Edwards, K., Kyler, P., Lloyd-Puryear, M.A., and Burke, W. (2009). Long-term outcomes of the “Genetics in Primary Care” Faculty Development Initiative. *Family Medicine*. 41(4):266-70.

⁴⁹ Collins, F.S. (1997). Preparing health professionals for the genetic revolution. *Journal of the American Medical Association*. 278(15):1285-1286.

⁵⁰ Collins, F.S., and Bochm, K. (1999). Avoiding casualties in the gene revolution: the urgent need to educate physicians about genetics. *Academic Medicine*. 74(1):48-49.

⁵¹ Hofman, K.J., Tambor, E.S., Chase, G.A., Geller, G., Faden, R.R., and Holtzman, N.A. (1993). Physicians’ knowledge of genetics and genetic tests. *Academic Medicine*. 68(8):625-632.

⁵² Escher, M., and Sappino, A.P. (2000). Primary care physicians’ knowledge and attitudes towards genetic testing for breast-ovarian cancer predisposition. *Annals of Oncology*. 11(9):1131-1135.

⁵³ Finn, C.T. (2007). Increasing genetic education for psychiatric residents. *Harvard Review of Psychiatry*. 15(1):30-33.

⁵⁴ Kemper, A.R., Uren, R.L., Moseley, K.L., and Clark, S.J. (2006). Primary care physicians’ attitudes regarding follow-up care for children with positive newborn screening results. *Pediatrics*. 118(5):1836-1841.

⁵⁵ Schroy, P.C., Barrison, A.F., Ling, B.S., Wilson, S., and Geller, A.C. (2002). Family history and colorectal cancer screening: a survey of physician knowledge and practice patterns. *American Journal of Gastroenterology*. 97(4):1031-1036.

a random sample of 1,251 licensed primary and specialty care physician members of the American Medical Association was surveyed about hereditary cancers likely to be encountered in their clinical practice. Among the respondents, only 37.5 percent correctly recognized that hereditary breast cancer due to mutations in *BRCA1* and *BRCA2* genes could be transmitted by fathers, only 33.8 percent correctly identified that less than 10 percent of female breast cancer patients carry *BRCA1* or *BRCA2* mutations, and only 13.1 percent knew that the penetrance of mutations for hereditary nonpolyposis colorectal cancer was greater than 50 percent.⁵⁹

Similar findings have emerged from studies of health care professionals other than physicians. A survey of 46 advanced practice nursing students (from nurse practitioner and nurse anesthesia programs) found that 56 percent of respondents had limited or no knowledge of pedigree construction and 54 percent had no knowledge about testing that uses polymerase chain reaction methodologies.⁶⁰ A 2003 study assessed community pharmacists' confidence in their knowledge about the Human Genome Project, genetic testing, and pharmacogenomics⁶¹ and found less than 50 percent with confidence in these topics.

Education is also important to maximize appropriate referrals to genetics specialists and improve the clinician-patient relationship in referral decisions.⁶² For example, a survey of 428 medical students' attitudes toward genetic testing of children for heritable conditions demonstrated that students who had completed a medical school genetics course were less likely to request that a minor be tested for an adult-onset disease suggesting that genetics education leads to more appropriate application of genetic testing in the clinical setting.⁶³ A study of genetic services referral patterns among 284 family physicians revealed similar findings. However, for a clinical scenario that did not warrant referral for genetic counseling and testing per U.S. Preventive Services Task Force guidelines, 92 percent of physicians referred the patient for genetic testing services and 50 percent referred for genetic counseling, believing that refusal to refer would harm their relationship with the patient.⁶²

Health care professionals also are unfamiliar with genetics as related to health policy, legal protection of their patients, and the possible effects of genetic information with regard to health disparities. In 2004, a California-based survey of 191 physicians and 80 nurses (registered nurses and nurse practitioners) found that 58.3 percent were misinformed about the existence of

⁵⁶ Taylor, M.R., Edwards, J.G., and Ku, L. (2006). Lost in transition: challenges in the expanding field of adult genetics. *American Journal of Medical Genetics*. 142C(4):294-303.

⁵⁷ Trinidad, S.B., Fryer-Edwards, K., Crest, A., Kyler, P., Lloyd-Puryear, M.A., and Burke, W. (2008). Educational needs in genetic medicine: primary care perspectives. *Community Genetics*. 11(3):160-165.

⁵⁸ Wilkins-Haug, L., Hill, L.D., Power, M.L., Holzman, G.B., and Schulkin, J. (2000). Gynecologists training, knowledge, and experiences in genetics: a survey. *Obstetrics and Gynecology*. 95(3):421-424.

⁵⁹ Wideroff, L., Vadaparampil, S.T., Greene, M.H., Taplin, S., Olson, L., and Freedman, A.N. (2005). Hereditary breast/ovarian and colorectal cancer genetics knowledge in a national sample of U.S. physicians. *Journal of Medical Genetics*. 42(10):749-755.

⁶⁰ Maradiegue, A., Edwards, O.T., Seibert, D., Macri, C., and Sitzer, L. (2005). Knowledge, perceptions, and attitudes of advanced practice nursing students regarding medical genetics. *Journal of the American Academy of Nurse Practitioners*. 17(11):472-479.

⁶¹ Sansgiry, S.S. and Kulkarni, A.S. (2003). The Human Genome Project: assessing confidence in knowledge and training requirements for community pharmacists. *American Journal of Pharmaceutical Education*. 67(2):291-300.

⁶² White, D.B., Bonham, V.L., Jenkins, J., Stevens, N. and McBride, C.M. (2008). Too many referrals of low-risk women for BRCA1/2 genetic services by family physicians. *Cancer, Epidemiology, Biomarkers & Prevention*. 17(11):2980-2986.

⁶³ Riordan, S.H. and Loescher, L.J. (2006). Medical students' attitudes toward genetic testing of minors. *Genetic Testing*. 10(1):68-73.

protective legislation; more than 50 percent did not know if cases of health insurance genetic discrimination based on cancer genetic testing actually existed; and 13 percent would not refer patients to genetic counseling or for genetic testing even if a strong family history of cancer was present.⁶⁴ More recently, a study of 1,181 physicians and nurse practitioners demonstrated that 96 percent of respondents viewed genetic testing as useful in ascertaining genetic cancer risks, but more than 60 percent were unaware of federal or California laws prohibiting health insurance discrimination.⁶⁵

Genetics and Genomics Clinical Competencies for Health Care Professionals

According to Hundert and Epstein, competency is defined as “the habitual and judicious use of communication, knowledge, technical skills, clinical reasoning, emotions, values, and reflection in daily practice for the benefit of the individual and the community being served.”⁶⁶ The National Coalition of Health Professional Education in Genetics⁶⁷ (NCHPEG) has identified overarching clinical competencies in genetics for all health care professionals, and various professional groups have developed clinical competencies for their individual disciplines (see Appendix C for competencies for physicians, nurses, genetic counselors, and pharmacists). In addition, the National Human Genome Institute (NHGRI) has launched the Genetics/Genomics Competency Center (G2C2),⁶⁸ an online tool to help educate nurses and physician assistants. G2C2 helps match existing resources with educational competencies to encourage sharing and avoid duplicative efforts across health care disciplines.

Pedigree assessment (i.e., family history) is incorporated into many competency recommendations as a basic and minimal competency. Obtaining a family history is widely regarded as a mechanism by which to detect familial transmission of hereditary diseases,^{69,70,71,72,73} and national public health campaigns encourage individuals to bring their family histories to their health care providers.⁷⁴ However, clinicians are hesitant to incorporate

⁶⁴ Blazer, K.R., MacDonald, D.J., Ricker, C., Sand, S., Uman, G.C., and Weitzel, J.N. (2005). Outcome from intensive training in genetic cancer risk counseling for clinicians. *Genetics in Medicine*. 7(1):40-47.

⁶⁵ Lowstuter, K.J., Sand, S., Blazer, K.R., MacDonald, D.J., Banks, K.C., Lee, C.A., Schwerin, B.U., Juarez, M., Uman, G.C., and Weitzel, J.N. (2008). Influence of genetic discrimination perceptions and knowledge on cancer genetics referral practice among clinicians. *Genetics in Medicine*. 10(9):691-698.

⁶⁶ Epstein, R.M., and Hundert, E.M. (2002). Defining and assessing professional competence. *Journal of the American Medical Association*. 287(2):226-235.

⁶⁷ National Coalition of Health Professional Education in Genetics. See <http://www.nchpeg.org/>. Accessed on March 8, 2010.

⁶⁸ Genetics/Genomics Competency Center. See <http://www.g-2-c-2.org>. Accessed on December 9, 2010.

⁶⁹ U.S. Preventive Services Task Force. (2005). Summaries for patients. Genetic risk assessment and BRCA mutation testing for breast and ovarian cancer susceptibility: U.S. Preventive Services Task Force recommendations. *Annals of Internal Medicine*. 143(5):147.

⁷⁰ Centers for Disease Control and Prevention. (2004). Awareness of family health history as a risk factor for disease—United States 2004. *Morbidity and Mortality Weekly Report*. 53(44):1044-1047.

⁷¹ Frezzo, T.M., Rubinstein, W.S., Dunham, D., and Ormond, K.E. (2003). The genetic family history as a risk assessment tool in internal medicine. *Genetics in Medicine*. 5(2):84-91.

⁷² Qin, P., Agerbo, E., and Mortenson, P.B. (2002). Suicide risk in relation to family history of completed suicide and psychiatric disorders: a nested case-control study based on longitudinal registers. *Lancet*. 360(9340):1126-1130.

⁷³ Williams, R.R., Hunt, S.C., Heiss, G., Province, M.A., Benson, J.T., Higgins, M., Chamberlain, R.M., Ware, J., and Hopkins, P.M. (2001). Usefulness of cardiovascular family history data for population-based preventive medicine and medical research (the Health Family Tree Study and the NHLBI Family Heart Study). *The American Journal of Cardiology*. 87(2):129-35.

⁷⁴ U.S. Department of Health and Human Services. Surgeon General’s Family Health History Initiative. See <http://www.hhs.gov/familyhistory/>. Accessed on November 25, 2009.

use of family history assessment due to time constraints, questions about clinical utility, beliefs of unreliability, and absence of meaningful financial reimbursement.^{75,76,77}

Lack of proper education might also be a factor in low use rates of family history information. Greb et al. performed an analysis of medical genetics knowledge and skill retention in 212 medical students following their third year and found that only 36.8 percent correctly asked about presence of family history in a cystic fibrosis case scenario.⁴³ This trend occurs among nursing students as well; only 22 percent of 46 Advanced Practice Nursing students in nurse practitioner programs thought that they could draw a family pedigree.⁶⁰

Despite encouragement to use family history in primary care,⁷⁵ the National Institutes of Health (NIH) State-of-the Science Conference on Family History and Improving Health, held in August 2009, concluded that family history plays an important role in medicine, but more research is needed before a systematically collected family history for common disease will become an evidence-based tool in primary care settings.⁷⁸ This issue highlights some of the challenges of integrating emerging technologies into practice. Family history is a promising clinical aid, but until firm evidence of its clinical utility is available, practitioners will be hesitant to change practice patterns.

Other genetic skills recommended by consensus panels are encountering similar challenges in accurate clinical application and dissemination. For example, a key genetics competency is the ability to counsel patients about genetic concerns and correctly issue referrals for genetic services. In a study of 900 internists, obstetricians, and oncologists regarding BRCA testing for risk of hereditary breast and ovarian cancer, only 13 percent of internists, 21 percent of obstetricians, and 40 percent of oncologists could correctly answer four basic genetics concept questions.⁷⁹ In the same study, although greater genetic knowledge influenced frequency of discussing the BRCA genetic test with patients, 54 percent of oncologists acting on the basis of inaccurate genetics concepts discussed genetic testing with their patients and presumably, made health-related decisions regarding their care.

Establishing competencies will become even more challenging as genetic testing is increasingly applied to common, chronic, multifactorial conditions such as heart disease and diabetes. A recent comprehensive review of the literature shows little data available to health care providers interested in using genetics to manage adult-onset conditions.⁸⁰ This review suggests that until health outcome data on genetic technologies exist, and clear and accessible education mechanisms for current health care professionals and students are available, use of genetics as

⁷⁵ Guttmacher, A.E., Collins, F.S., and Carmona, R.H. (2004). The family history—more important than ever. *The New England Journal of Medicine*. 351(22):2333-2336.

⁷⁶ Rich, E.C., Burke, W., Heaton, C.J., Haga, S., Pinsky, L., Short, M.P., and Acheson, L. (2004). Reconsidering the family history in primary care. *Journal of General Internal Medicine*. 19(3):273-280.

⁷⁷ Suther, S., and Goodson, P. (2003). Barriers to the provision of genetic services by primary care physicians: a systematic review of the literature. *Genetics in Medicine*. 5(2):70-76.

⁷⁸ National Institutes of Health Consensus Development Program: NIH State of the Science Conference, Family History and Improving Health. See <http://consensus.nih.gov/2009/familyhistory.htm>. Accessed on November 16, 2009.

⁷⁹ Doksum, T., Bernhardt, B.A., and Holtzman, N.A. (2003). Does knowledge about the genetics of breast cancer differ between nongeneticist physicians who do or do not discuss or order BRCA testing? *Genetics in Medicine*. 5(2):99-105.

⁸⁰ Scheuner, M.T., Sieverding, P., and Shekelle, P.G. (2008). Delivery of genomic medicine for common chronic adult diseases. *Journal of the American Medical Association*. 299(11):1320-1334.

outlined in competency statements is not likely to reach the bedside without further strategic support.

Academic Preparation, Licensing, Accreditation, and Continuing Education

Educating health care professionals in their respective disciplines includes undergraduate and graduate education, preparation for licensing and certification exams, and continuing education (CE) for practicing professionals. (See Appendix C for genetic and genomic competencies and accreditation and licensing programs for selected health care professionals.)

Studies have documented that health care professionals are generally optimistic about the future utility of genetic tests and are interested in their eventual incorporation into clinical practice.^{52,81} However, many experience feelings of discomfort stemming from lack of confidence in their knowledge of basic genetic concepts, interventions, and management strategies.^{82,83,84} Thus, many resources have been devoted to CE efforts for health care professionals in regional practice settings^{64,85,86,87} and include web-based instruction tutorials, CE seminars, professional workshops, and conference proceedings.^{88,89,90} Access to and participation in CE, however, does not determine proficiency in providing clinical care. An extensive meta-analysis completed through The Cochrane Collaboration demonstrates that improvement in health care provider behaviors (through CE) and ultimately, patient health outcomes, is dependent on the method of instruction and health care providers' access to interactive practice.^{91,92} The critical importance of a well-educated professional workforce and the role of CE in high-quality health care and

⁸¹ Friedman, A.N., Wideroff, L., Olson, L., Davis, W., Klabunde, C., Srinath, K.P., Reeve, B.B., Croyle, R.T., and Ballard-Barbash, R. (2003). U.S. physicians' attitudes toward genetic testing for cancer susceptibility. *American Journal of Medical Genetics*. 120A(1):63-71.

⁸² Edwards, Q.T., Maradiegue, A., Seibert, D., Saunders-Goldson, S., and Humphreys, S. (2009). Breast cancer risk elements and nurse practitioners' knowledge, use, and perceived comfort level of breast cancer risk assessment. *Journal of the American Academy of Nurse Practitioners*. 21(5):270-277.

⁸³ Gramling, R., Nash, J., Siren, K., Eaton, C., and Culpepper, L. (2004). Family physician self-efficacy with screening for inherited cancer risk. *Annals of Family Medicine*. 2(2):130-132.

⁸⁴ Watson, E., Clements, A., Yudkin, P., Rose, P., Bukach, C., Mackay, J., Lucassen, A., and Austoker, A. (2001). Evaluation of the impact of two educational interventions on GP management of familial breast/ovarian cancer cases: a cluster randomized controlled trial. *The British Journal of General Practice*. 51(471):817-821.

⁸⁵ Blazer, K.R., Grant, M., Sand, S.R., MacDonald, D.J., Uman, G.C., and Weitzel, J.N. (2004). Effects of a cancer genetics programme on clinician knowledge and practice. *Journal of Medical Genetics*. 41(7):518-522.

⁸⁶ Clyman, J.C., Nazir, F., Tarolli, S., Black, E., Lombardi, R.Q., and Higgins, J.J. (2007). The impact of a genetics education program on physicians' knowledge and genetic counseling referral patterns. *Medical Teacher*. 29(6):e143-150.

⁸⁷ Prows, C.A., Hetteberg, C., Hopkins, R.J., Latta, K.K., and Powers, S.M. (2004). Development of a web-based genetics institute for a nursing audience. *Journal of Continuing Education in Nursing*. 35(5):223-231.

⁸⁸ Centers for Disease Control and Prevention. Public Health Genomics, Genomic Resources. [See http://www.cdc.gov/genomics/resources/t.htm](http://www.cdc.gov/genomics/resources/t.htm). Accessed on November 25, 2009.

⁸⁹ Harvard Medical School. The Genetic Basis of Adult Medicine. [See http://cme.med.harvard.edu/cmeups/custom/00281416/index.htm](http://cme.med.harvard.edu/cmeups/custom/00281416/index.htm). Accessed on November 25, 2009.

⁹⁰ Dartmouth Medical School. Genetics in Clinical Practice, A Team Approach. [See http://iml.dartmouth.edu/education/cme/Genetics/](http://iml.dartmouth.edu/education/cme/Genetics/). Accessed on November 25, 2009.

⁹¹ Forsetlund, L., Bjørndal, A., Rashidian, A., Jamtvedt, G., O'Brien, M.A., Wolf, F., Davis, D., Odgaard-Jensen, J., and Oxman, A.D. (2009). Continuing education meetings and workshops: effects on professional practice and health care outcomes. *Cochrane Database of Systemic Reviews*. 15(2):CD003030.

⁹² Davis, D., O'Brien, M.A., Freemantle, N., Wolf, F.M., Mazmanian, P., and Taylor-Vaisey, A. (1999). Impact of formal continuing medical education: do conferences, workshops, rounds, and other traditional continuing education activities change physician behavior or health care outcomes? *The Journal of the American Medical Association*. 282(9):867-874.

patient safety were explored in a 2010 report of the Institute of Medicine.⁹³ The report concluded “the absence of a comprehensive and well-integrated system of continuing education in the health professions is an important contributing factor to knowledge and performance deficiencies at the individual and system levels.”

Continuing education must be ongoing to be effective. A recent study examining the impact of a CE program in genetics found that although health care professionals felt more confident applying genetics knowledge after the CE program, 48 percent of respondents applied their new knowledge incorrectly.⁹⁴ Incorrect application of genetics occurred in the misappropriation of risk estimation; approximately half of those receiving the genetics education intervention assigned a high-risk categorization to a low-risk breast cancer presentation. Reinforcement over time of complex content is important to assure appropriate and accurate use of genetic information.

An interactive, case-based, peer education model was shown to increase knowledge and confidence in genetic competencies among physicians delivering genetic services in primary care settings.⁹⁵ Peer education emphasizes the usability of educational materials and concepts and was rated as an effective method for understanding and applying knowledge by most participants in this study.

Challenges and Barriers to Health Care Professional Use of Genetics and Genomics

A recent analysis identified three obstacles that have hindered the adoption of tailored approaches to patient treatment based on genetic testing: (1) lack of scientific evidence supporting the efficacy and utility of genetic testing; (2) economic incentives that are poorly aligned among stakeholders; and (3) resolution of operational issues, such as electronic tracking of diagnostic information and health care provider education and training.⁹⁶ Of the three obstacles, correcting the misalignment of financial incentives among stakeholders—such as payers, health care providers, patients, pharmaceutical and biotechnology companies, and diagnostics research and development companies—was acknowledged as the most challenging. Catalysts that could help correct this misalignment include increasing the pace and predictability of payer coverage for appropriate tests and aligning reimbursement practices to encourage appropriate diagnostic use by physicians (e.g., developing billing codes that are commensurate with the cost and value of each molecular diagnostic test and provide appropriate reimbursement to physicians). SACGHS also identified inadequate coverage and reimbursement as limiting factors in the integration of genetics into medical care and provided recommendations on how to improve access to and utilization of genetic tests and services in a 2006 report.⁹⁷

⁹³ Institute of Medicine. *Redesigning Continuing Education in the Health Professions*. Washington, D.C. National Academies Press, 2010, p.1.

⁹⁴ Bethea, J., Qureshi, N., Drury, N., and Guilbert, P. (2008). The impact of genetic outreach education and support to primary care on practitioner's confidence and competence in dealing with familial cancers. *Community Genetics*. 11(5):289-294.

⁹⁵ Carroll, J.C., Rideout, A.L., Wilson, B.J., Allanson, J., Blaine, S.M., Esplen, M.J., Farrell, S.A., Graham, G.E., MacKenzie, J., Meschino, W., Miller, F., Prakash, P., Shuman, C., Summers, A., and Taylor, S. (2009). Genetic education for primary care providers. *Canadian Family Physician*. 55:e92-99.

⁹⁶ Davis, J.C., Furstenthal, L., Desai, A.A., Norris, T., Sutaria, S., Fleming, E., and Ma, P. (2009). The microeconomics of personalized medicine: today's challenge and tomorrow's promise. *Nature Reviews Drug Discovery*. 8:279-286.

⁹⁷ Secretary's Advisory Committee on Genetics, Health, and Society. (2006). Coverage and Reimbursement of Genetic Tests and Services. See http://oba.od.nih.gov/oba/sacghs/reports/CR_report.pdf. Accessed on November 25, 2009.

The lack of scientific evidence supporting the efficacy and utility of genetic testing applies across all disciplines^{59,98} and influences health professionals' choice of CE offerings, making genetics less of a priority than other topics deemed more relevant. Also, the need for national guidelines based on health care outcome data affects the willingness of clinicians to apply genetics in their practices.^{77,81,99,100,101} Suggested areas for further research include an assessment of the scope of clinical benefits and harms associated with various genetic tests, identification of possible ethical and discriminatory harms related to genetic information, and ascertainment of financial benefits and costs of genetic services.¹⁰²

The Genetic Professional Workforce

Health care professional workforce analyses performed by the Health Resources and Services Administration (HRSA) showed that in the United States there are currently 817,500 physicians (763,200 medical doctors and 54,300 doctors of osteopathy);¹⁰³ 2.9 million registered nurses (376,901 with Master's or Doctorate degrees and 141,209 nurse practitioners);¹⁰⁴ 72,433 physician assistants;¹⁰⁵ and 226,000 pharmacists.¹⁰⁶ In addition, there are 2,789 certified Master's prepared genetic counselors.¹⁰⁷ In 2009, the American Board of Medical Genetics (ABMG) reported that over a 27-year period beginning in 1982, 2,511 individuals had achieved board certification in one or more of the ABMG certification areas.¹⁰⁸ The 1,326 physician geneticists certified between 1982 and 2009 represent less than 0.3 percent of the more than 817,500 U.S. physicians. It is not known how many of these individuals are currently practicing.

A 2005 study concluded that the medical genetics workforce was not sufficient to meet expected patient care needs for clinical genetic services in the next 10 years (2010 to 2020).¹⁰⁹ The mismatch between increased need for genetic services and the size of the genetics workforce is

⁹⁸ Epstein, R.S., Frueh, F.W., Geren, D., Hummer, D., McKibben, S., O'Connor, S., Randhawa, G., and Zelman, B. (2009). Payer perspectives on pharmacogenomics testing and drug development. *Pharmacogenomics*. 10(1):149-151.

⁹⁹ Hindorff, L.A., Burke, W., Laberge, A.M., Rice, K.M., Lumley, T., Leppig, K., Rosendaal, F.R., Larson, E.B., and Psaty, B.M. (2009). Motivating factors for physician ordering of factor V Leiden genetic tests. *Archives of Internal Medicine*. 169(1):68-74.

¹⁰⁰ Suther, S.G., and Goodson, P. (2004). Texas physicians' perceptions of genomic medicine as an innovation. *Clinical Genetics*. 65(5):368-377.

¹⁰¹ Zapka, J.G., Puleo, E., Taplin, S., Solberg, I.E., Mouchawar, J., Somkin, C., Geiger, A.M., and Ulcickas Yood, M. (2005). Breast and cervical cancer screening: clinicians' views on health plan guidelines and implementation efforts. *Journal of the National Cancer Institute Monographs* 2005. (35):46-54.

¹⁰² Rogowski, W.H., Grosse, S.D., and Khoury, M.J. (2009). Challenges of translating genetic tests into clinical and public health practice. *Nature Reviews Genetics*. 10(7):489-495.

¹⁰³ U.S. Department of Health and Human Services, Health Resources and Services Administration, Bureau of Health Professions. The Physician Workforce: Projections and Research into Current Issues Affecting Supply and Demand. December 2008: pps. 4, 13.

¹⁰⁴ U.S. Department of Health and Human Services, Health Resources and Services Administration, Bureau of Health Professions. The Registered Nurse Population: Findings from the March 2004 National Sample Survey of Registered Nurses. 2004; p. 32.

¹⁰⁵ National Physician Assistant Census Report: Results from American Academy of Physician Assistant's 2009 Census. p. 1.

¹⁰⁶ U.S. Department of Health and Human Services, Health Resources and Services Administration, Bureau of Health Professions. The Adequacy of Pharmacist Supply: 2004-2030. December 2008; p. 4.

¹⁰⁷ National Society of Genetic Counselors. <http://www.nsgc.org/> Accessed on September 17, 2010.

¹⁰⁸ American Board of Medical Genetics. *Number of Certified Specialists in Genetics*. See http://www.abmg.org/pages/resources_certspecial.shtml. Accessed on March 31, 2010.

¹⁰⁹ Cooksey, J.A., Forte, G., Benkendorf, J., and Blitzer, M.G. (2005). The state of the medical geneticist workforce: findings of the 2003 survey of ABMG certified geneticists. *Genetics in Medicine*. 7(6):439-443.

exacerbated by data showing that young physicians are not entering the field of genetics.¹¹⁰ Because many States and regions already have too few genetics physicians to meet current demand, the absence of major workforce expansion was projected to leave some patient subgroups with insufficient access to services, particularly patients with inborn errors of metabolism and those living in rural areas.¹¹¹ These deficiencies become even more concerning in light of expanded newborn screening programs, which are expected to detect 10,000 affected infants annually, with many needing chronic disease management. Yet, there are only 200 U.S. physicians specialized in the diagnosis and management of patients with inherited metabolic disease. Physicians who have such expertise are least able to expand services;¹⁰⁹ three-quarters reported that their practices are “nearly full,” and about one-quarter reported that new patients wait more than 3 months for an appointment. Educational efforts that focus on screening procedures and referral practices are critical in maximizing this life-saving public health program.¹¹²

Another concern is that the medical geneticist workforce does not match U.S. racial and ethnic demographics. In 2003, only 13 percent of medical geneticists identified themselves as members of an ethnic or racial minority.¹⁰⁹ Underrepresentation of diverse populations in the health care workforce has been cited as a primary barrier to mitigating health care disparities.^{113,114,115}

In summary, although the need for clinical genetic services continues to increase, the ability of the genetics-specific workforce—which includes medical geneticists, genetic counselors, and other health care workers with specialized training—is not sufficient to meet this need. Greater efforts are required to ensure that adequate numbers of point-of-care health professionals are trained to provide appropriate genetic services and information.^{116,117}

Clinical Decision Support and EHRs

Recent studies assessing genetic content in a variety of commonly used online medical resources identified large gaps in content as well as significant errors in the information that was available to nongeneticist health care professionals seeking information about common genetic conditions.¹¹⁸ As EHRs are increasingly deployed in clinical care, a potential solution to these

¹¹⁰ Brotherton, S.E., and Etzel, S.I. (2009). Graduate medical education, 2008-2009. *Journal of the American Medical Association*. 302(12):1357-1372.

¹¹¹ Cooksey, J.A., Forte, G., Flanagan, P., Benkendorf, J., and Blitzer, M.G. (2006). The medical geneticist workforce: an analysis of clinical subgroups. *Genetics in Medicine*. 8(10): 603-614.

¹¹² Sanford A., Northrup, H., Crandell, S.S., King, T.M., Champaigne, N.L., Yafi, M., Therrell, B.L., and Noblin, S.J. (2009). Expanded newborn screening in Texas: a survey and educational module addressing the knowledge of pediatric residents. *Genetics in Medicine*. 11(3):163-168.

¹¹³ Meghani, S.H., Brooks, J.M., Gipson-Jones, T., Waite, R., Whitfield-Harris, L., and Deatrick, J.A. (2009). Patient-provider race-concordance: does it matter in improving minority patients' health outcomes? *Ethnicity & Health*. 14(1):107-130.

¹¹⁴ Kennedy, E.M. (2005). The role of the federal government in eliminating health disparities. *Health Affairs*. 24(2): 452-458.

¹¹⁵ Institute of Medicine Committee on Understanding and Eliminating Racial and Ethnic Disparities in Health Care. (2003). *Unequal Treatment: Confronting Racial and Ethnic Disparities in Health Care*. See http://www.nap.edu/catalog.php?record_id=10260. Accessed on December 2, 2010.

¹¹⁶ Martin, G.P., Currie, G., and Finn, R. (2009). Reconfiguring or reproducing intra-professional boundaries? Specialist expertise, generalist knowledge and the ‘modernization’ of the medical workforce. *Social Science in Medicine*. 68:1191-1198.

¹¹⁷ Henderson, M. “NHS faces genetic revolution by bringing scientists into clinics.” July 30, 2009. Times Online. See <http://www.timesonline.co.uk/tol/news/uk/health/article6732590.ece>. Accessed on November 23, 2009.

¹¹⁸ Levy, H.P., LoPresti, L., and Siebert, D.C. (2008). Twenty questions in genetic medicine—an assessment of World Wide Web databases for genetics information at the point of care. *Genetics in Medicine*. 10(9):659-657.

issues has emerged—just-in-time education, which provides answers to specific questions at the time practitioners need this information. A study by Trinidad et al. regarding genetics education needs of primary care providers identified a desire to have “just-in-time” resources.⁵⁷ The applicability of active decision support in genetics was discussed in detail in a previous SACGHS report as one means of enhancing patient care.¹¹⁹

The key to just-in-time education is the use of context-sensitive elements embedded in the EHR. This approach involves the EHR “understanding” where the provider is in the patient workflow. When the query is executed, the provider is taken to content that is likely to be relevant to the question the provider is considering. A study by del Fiol et al. demonstrated that answers could be found significantly faster using infobuttons¹²⁰ than traditional electronic search approaches.¹²¹ At Intermountain Healthcare, more than 200 infobuttons related to genetic disorders in the problem list were linked to specific genetic information from GeneTests,¹²² Genetics Home Reference,¹²³ and the Online Mendelian Inheritance in Man (OMIM)¹²⁴ websites. Usage of these genetic-specific infobuttons has continued to increase over time with good provider satisfaction.¹²⁵ Preliminary data from the Intermountain Healthcare System specific to genetic content suggested that providers needed to spend about 45 seconds accessing the resource, but no data are available at this time to address whether specific questions were answered.

The Mayo Clinic also has deployed a just-in-time approach to deliver genomic information to providers.¹²⁶ To date, no rigorous studies have assessed the effectiveness of these types of educational interventions in acquiring and retaining new knowledge that alters practice behavior; however, studies such as one by Maviglia et al. demonstrated that providers found answers to questions about medications 84 percent of the time with an average elapsed time from question to answer of 21 seconds.¹²⁷ Additionally, just-in-time learning tools have been deployed for patient use in a variety of health care settings. Many of these tools involve breast cancer care and include innovative approaches for patients with low literacy¹²⁸ and to aid in genetic testing decisions.¹²⁹ Thus, while gaps have been identified in the availability and accuracy of genetic information needed for clinical care decisionmaking, innovative approaches, such as the use of

¹¹⁹ Secretary’s Advisory Committee on Genetics, Health, and Society. (2008). *U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of Health and Human Services*. See http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf. Accessed on September 3, 2009.

¹²⁰ Infobuttons are icons that appear in certain areas of the EHR (e.g., problem list, medication list, and laboratory results).

¹²¹ Del Fiol, G., Haug, P.J., Cimino, J.J., Narus, S.P., Norlin, C., and Mitchell, J.A. (2008). Effectiveness of topic-specific infobuttons: a randomized controlled trial. *Journal of the American Medical Information Association*. 15(6):752-9.

¹²² GeneTests. See <http://www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests>. Accessed on November 17, 2009.

¹²³ National Library of Medicine Genetics Home Reference. See <http://ghr.nlm.nih.gov/>. Accessed on November 19, 2009.

¹²⁴ Online Mendelian Inheritance in Man. See <http://www.ncbi.nlm.nih.gov/omim/>. Accessed on May 18, 2010.

¹²⁵ Personal communication, Marc S. Williams, M.D., Director, Intermountain Healthcare Clinical Genetics Institute, unpublished data, September 3, 2009.

¹²⁶ Kaihoi, B., Petersen, C., and Bolander, M.E. (2005). Providing “just-in-time” medical genomics information for patient care. *American Medical Information Association Annual Symposium Proceedings*. 1003.

¹²⁷ Maviglia, S.M., Yoon, C.S., Bates, D.W., and Kuperman, G. (2006). KnowledgeLink: impact of context-sensitive information retrieval on clinicians’ information needs. *Journal of the American Medical Information Association*. 13(1):67-73.

¹²⁸ Jibaja-Weiss, M.L., Volk, R.J., Friedman, L.C., Granchi, T.S., Neff, N.E., Spann, S.J., Robinson, E.K., Aoki, N., and Beck, R. (2006). Preliminary testing of a just-in-time, user-defined values clarification exercise to aid lower literate women in making informed breast cancer treatment decisions. *Health Expectations*. 9(3):218-31.

¹²⁹ Green, M.J., Peterson, S.K., Baker, M.W., Harper, G.R., Friedman, L.C., Rubinstein, W.S., and Mauger, D.T. (2004). Effect of a computer-based decision aid on knowledge, perceptions, and intentions about genetic testing for breast cancer susceptibility: a randomized controlled trial. *Journal of the American Medical Association*. 292(4):442-52.

just-in-time learning tools, appears to be a promising method to deliver genetic knowledge to the provider.

Public Health Workforce

In contrast to clinicians, who focus on the needs of individuals, public health practitioners assess the needs of populations to determine the burden of disease and assure that appropriate services are available to individuals, families, and communities. Public health providers work across various sectors, including Federal and State Governments; academia; and professional, community and lay organizations. They work in various population health domains such as epidemiology, biostatistics, environmental health, health promotion, and maternal and child health. The public health community has unique skills and networks to improve general genomic literacy and develop targeted messages about the use of genetic information for disease prevention and health promotion.¹³⁰ In addition, the public health community has a large research infrastructure sorely needed by genomics (e.g., surveillance and data collection systems).

Genetics has been at the center of a number of important public health programs for decades. Most State health departments administer newborn genetic screening and other genetic disease prevention programs focused primarily on diseases related to maternal and child health. Some State health departments employ genetics coordinators and frequently consult with genetics professionals.^{131,132} In general, however, expertise and focus have been limited to the maternal and child health field. In contrast, a more expansive view of public health genomics focuses on the effective and responsible translation of genomics to improve population health.¹³³ Public health genomics is defined by Khoury et al. as using “population-based data on genetic variation and gene-environment interactions to develop, implement, and evaluate evidence-based tools for improving health and preventing disease. It also applies systematic evidence-based assessments of genomic applications in health practice and works to ensure the delivery of validated, useful genomic tools for the benefit of population health.”¹³³

Barriers to Achieving a Genomics Informed Public Health Workforce

The current public health workforce faces challenges assimilating genetic and genomic information. Individualized primary prevention and early detection (often the purview of primary care) intersects with the realm of population health (the purview of public health). Khoury et al. caution that without a more integrated approach between primary care and public health, genomics could easily widen the schism that has long existed between medicine and public health.¹⁴

¹³⁰ Beskow, L.M., Khoury, M.J., Baker, T.G., and Thrasher, JF. (2001). The integration of genomics into public health research, policy and practice in the United States. *Community Genetics*. 4:2-11.

¹³¹ Piper, M.A., Lindenmayer, J.M., Lengerich, E.J., Pass, K.A., Brown, W.G., Crowder, W.B., Khoury, M.J., Baker, T.G., Lloyd-Puryear, M.A., and Bryan, J.L. (2001). The role of state public health agencies in genetics and disease prevention: results of a national survey. *Public Health Reports*. 116:22-31.

¹³² Au, S.M. and Yu, A. Survey of State Genetics Coordinator Survey: Update on Public Health Genetics Over the Past Decade. Presented at the 4th National Conference on Genomics and Public Health, December 9, 2010.

¹³³ Khoury, M.J., Bowen, S., Bradley, L.A., Coates, R., Dowling, N.F., Gwinn, M., Kolor, K., Moore, C.A., St. Pierre, J., Valdez, R., and Yoon, P.W. (2008). A decade of public health genomics in the United States: Centers for Disease Control and Prevention 1997-2007. *Public Health Genomics*. 12:20-29.

The barriers to achieving a more informed public health workforce are multifaceted. First, the public health workforce is diverse and follows many educational and training paths, including a variety of professionals with formal training and certifications, volunteers, and community (lay) health workers. The diversity of settings and service provision and the lack of specific licensure that would otherwise facilitate counting and studying the public health workforce¹³⁴ create an inherent problem in targeting genetic and genomic educational efforts. Second, similar to clinical practitioners, many public health providers in the field today received their formal education before genetics became a critical aspect of medicine and health. Third, attitudes, perceptions, and beliefs shape acceptance and adoption of genetics by the public health community. Thus, a one-size-fits-all approach to genetics education and training is not practical.

A unique feature of the public health workforce is the proportion of nonprofessional, community health workers (CHWs). Defined as “any health worker carrying out functions related to health care delivery; trained in some way in the context of the intervention; [but] having no formal professional or paraprofessional certificated or degreed tertiary education,”¹³⁵ they create an additional challenge to achieving widespread genetic literacy in the public health workforce. Although Texas, Ohio, Indiana, and Alaska require some level of certification for CHWs and several states are considering implementing certification requirements,¹³⁶ most States do not. Until 2007, there were no national standards for certifying or training nonprofessional public health workers.^{136,137,138}

Khoury et al.¹⁴ have noted some of the attitudinal barriers to acceptance of genetics by the public health community. These include a view that genetics is a low-yield investment and low priority because of other more important preventive or modifiable environmental causes of morbidity and mortality. Local issues, national and international pandemics, and environmental causes of morbidity and mortality are viewed as more important priorities than genetics, particularly in the context of limited public health funding. Research also highlights that public health providers do not perceive public health genetics to be part of their job, nor a professional priority. Barriers include not only lack of knowledge regarding the link between genetics and health promotion, but also a lack of current basic genetic knowledge. Future education and training that focuses primarily on basic genomic content will be inadequate; these efforts should address how to apply genomic innovations in health promotion.¹³⁹

To address the place of genetics in public health practice, Chen et al. assessed U.S. public health educators’ attitudes toward genomic competencies, evaluated their awareness of efforts in the

¹³⁴ Health Resources and Services Administration. (2005). *Public Health Workforce*. See <http://bhpr.hrsa.gov/healthworkforce/reports/publichealth/default.htm>. Accessed on December 13, 2009.

¹³⁵ Lewin, S.A., Dick, J., Pond, P., Zwarenstein, M., Aja, G., van Wyk, B., Bosch-Capblanch, X., and Patrick, M. (2005). Lay health workers in primary and community health care. *Cochrane Database Systematic Reviews*. Jan 25;(1):CD004015.

¹³⁶ May, M.L., Kash, B., and Contreras, R. (2005). Southwest Rural Health Research Center: Community Health Worker (CHW) Certification and Training—A National Survey of Regionally and State-based Programs. U.S. Department of Health and Human Services, Health Services and Resources Administration, Office of Rural Health Policy.

¹³⁷ Love, M.B., Gardner, K., and Legion, V. (1997). Community health workers: who they are and what they do. *Health Education and Behavior*. 24(4):510-22.

¹³⁸ Love, M.B., Legion, V., Shim, J.K., Tsai, C., Quijano, V., and Davis, C. (2004). CHWs get credit: a 10-year history of the first college-credit certificate for community health workers in the United States. *Health Promotion and Practice*. 5(4):418-28.

¹³⁹ Chen, L-S., and Goodson, P. (2009). Barriers to adopting genomics into public health education: a mixed methods study. *Genetics in Medicine*. 11(2):104-110.

field to promote and incorporate genomic information and technologies into practice, and attempted to gauge their basic and applied genomic knowledge.¹⁴⁰ While most public health providers agreed with CDC's proposed competencies, incorporating them into public health practice was viewed as important by less than half of the study participants. The study authors concluded that "the simplest and most immediate explanation for such a gap is that the majority of training programs in health education and public health include neither genetics nor genomics in their curricula nor do they require course offerings in these topics for accreditation purposes." Subsequent work by Chen et al.¹⁴¹ found that public health providers are reluctant to adopt genomic competencies into health promotion. Only 35 percent of survey respondents said they were willing to integrate genomic components into community-based education programs¹⁴¹ and only half of basic and applied genomic knowledge questions were answered correctly,¹⁴⁰ suggesting that health educators are not prepared for their professional role in genomics.

Finally, lack of evidence of health benefit might be a significant barrier to public health adoption of genomic competencies. Until such evidence is available, public health providers might be resistant to adoption. Thus, 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, whether they are population screening programs, such as newborn screening, or early case detection and interventions delivered by clinicians."¹⁴

Current Efforts to Improve Proficiencies and Competencies

Recognizing the need for resources applicable to public health, the Partners in Information Access for the Public Health Workforce was launched in 1998. This collaborative effort of 11 U.S. government agencies, public health organizations, and health sciences libraries provides resources on a variety of topics pertaining to public health genetics.¹⁴²

In August 2000, the CDC Office of Genetics and Disease Prevention and representatives from each of the disciplines in public health met to identify and develop specific core competencies necessary for all health professionals to incorporate genomics into public health practice.¹³¹ The competencies were developed as a tool for public health programs and schools of public health to incorporate genomics into existing competencies and program training goals. However, as with any new requirement imposed on an already information-laden curriculum, incorporation of competencies in education and certification or licensure processes takes time. It is possible that various social, organizational, and environmental factors (e.g., certification and licensure requirements) might carry more weight than individuals' attitudes in promoting willingness to adopt genomic competencies.

In addition to convening the working group that developed the core competencies, CDC has made other investments in public health genomics practice and education (see also CDC Federal Activities in Chapter III of this report). It has funded Centers for Genomics and Public Health in

¹⁴⁰ Chen, L-S., and Goodson, P. (2007). Public health genomics knowledge and attitudes: a survey of public health educators in the United States. *Genetics in Medicine*. 9(8):496-503.

¹⁴¹ Chen L-S., Kwok, O.M., and Goodson P. (2008). U.S. health educators' likelihood of adopting genomic competencies into health promotion. *American Journal of Public Health*. 98(9):1651-1657.

¹⁴² Partners in Information Access for the Public Health Workforce. See http://phpartners.org/public_health_genomics.html. Accessed on December 14, 2009.

schools of public health at the University of North Carolina, the University of Michigan, and the University of Washington. These centers provide expertise in translating genomic information into public health knowledge, provide technical assistance to State and community public health agencies, and facilitate integration of genomics into programs and practice.¹³³ CDC also has supported genomics programs in State health departments in Michigan, Minnesota, Oregon, and Utah.

In 2003, the Institute of Medicine (IOM) recommended genetics as one of eight new content areas to be covered by every school of public health.¹⁴³ This recommendation confirms the need for public health provider education and training in genetics previously recommended by several professional groups and CDC.

In 1987, the American Public Health Association (APHA)—which represents more than 50,000 health professionals—published *Genetics and Public Health*.¹⁴⁴ It discussed the need for consensus among a wide variety of institutions and organizations regarding the public health implications of genetics and the need for quality genetic services. In recognition of the broader scope of genomics and its impact on public health and the critical need for public health workforce education, APHA published *The Role of Genomics in Public Health* in 2002.¹⁴⁵ In 2007, APHA established the Genomics Forum to “engage the public health community to promote workforce competency in genomics, including an improved understanding of the relevance and impact of genomics on public health.”¹⁴⁶ The Genomics Forum developed a policy statement on genetic health literacy for health professionals,¹⁴⁷ which was approved by the APHA Governing Council in 2010.¹⁴⁸

Some States have instituted their own initiatives in public health genetics. For example, the Oregon Genetics Program aims to integrate genetics into public health practice, particularly chronic disease program activities. The Oregon Public Health Division received funding to translate genetics into health practice, specifically to develop, implement, and evaluate a surveillance program to monitor awareness, knowledge, and use among health care providers and the public of cancer-related genetic tests and family history. This project also will evaluate disparities associated with accessing cancer-related genetic testing and counseling.¹⁴⁹ Illinois public health officials conducted a needs assessment and published a State genetics plan in

¹⁴³ Institute of Medicine. (2003) *Who will keep the public healthy?* Washington, D.C.: National Academies Press. See <http://www.nap.edu/openbook.php?isbn=030908542X>. Accessed on December 13, 2010.

¹⁴⁴ American Public Health Association. *Genetics and Public Health*. Policy Number 8732PP, January 1, 1987. See <http://www.apha.org/advocacy/policy/policysearch/default.htm?id=1161>. Accessed on January 13, 2010.

¹⁴⁵ American Public Health Association. *The Role of Genomics in Public Health*. Policy Number 2002-1, November 13, 2002. See <http://www.apha.org/advocacy/policy/policysearch/default.htm?id=275>. Accessed on January 13, 2010.

¹⁴⁶ American Public Health Association. Genomic Forum. See <http://www.apha.org/membergroups/forums/>. Accessed on January 13, 2010.

¹⁴⁷ Payne, E., Honore, H., Platt, J., and the American Public Health Association Genomics Forum Policy Committee. (2009). Genetic health literacy. Poster presented at the meeting of the Health Literacy Forum: Building a Community Network to Improve Health Communication, October, 2009. University of Michigan Health Science Libraries, Ann Arbor, MI.

¹⁴⁸ Personal communication, Heather Honoré, Policy Committee Chair, Genomics Forum, American Public Health Association., December 3, 2010.

¹⁴⁹ Oregon Genetics Program. Oregon Cancer Genomics Surveillance Program: Translation of Genomics Application into Healthcare Practices. See http://egov.oregon.gov/DHS/ph/genetics/genomics_grant.shtml. Accessed on December 2, 2010.

2007.¹⁵⁰ The Connecticut Department of Public Health Genomics Office has produced a fact sheet for consumers on DTC personal genetic services.¹⁵¹

Consumers and Patients

The genetics landscape for consumers and patients has changed dramatically in the past 20 years. Today, the term “consumer genomics” refers to the application of genomic technologies by private companies marketing testing services directly to the public via the Internet.²⁹ At this time, it is unknown how direct access to personal genetic information will change the way consumers approach health care and the extent to which they will seek knowledge on their own and bypass their health care providers. The emergence of social networking and sharing genetic information via the Internet raises concerns about confidentiality and the consequences of sharing such information.¹⁵² However, these tools can also be used to improve health through easier access to reliable information. For example, the emergence of mHealth—the provision of health-related services via mobile communications—is being explored as a way to improve health care services, even in remote and resource-poor environments.¹⁵³

As genetic tests become more widely available, the need for public education becomes increasingly important for patients and consumers to make informed decisions about testing. To understand what is known about the genetics knowledge and attitudes of the general public, SACGHS conducted a literature search to identify pertinent studies covering the years 2003 to 2009. In addition, the National Human Genome Research Institute shared with SACGHS a literature review conducted in 2009 by the Academy for Educational Development (AED) that focused on communication and understanding of genetics, genetic risk, DTC genomic services, and personalized medicine among the interested public.¹⁵⁴ Also, Cogent Research provided SACGHS with the findings from its 2008 survey, *Cogent Genomics Attitudes and Trends*.¹⁵⁵ This national, web-based survey consisted of responses from 1,000 adults, representative of the U.S. population by age, socioeconomic status, ethnicity, geographic region, and gender.

Consumer and Patient Knowledge and Awareness of Genetics and Genomics

Studies that have assessed the public’s knowledge of genetic testing generally have found that the public has only a rudimentary knowledge of basic genetic terms,¹⁵⁶ yet has positive attitudes

¹⁵⁰ Illinois Department of Public Health. (2007). Illinois State Genetics Plan. See

http://www.idph.state.il.us/HealthWellness/Genetics_07StatePlan.pdf. Accessed on December 2, 2010.

¹⁵¹ Connecticut Department of Public Health Genomics. *Direct-to-Consumer Personal Genomic Services Information to Consider*. See http://www.ct.gov/dph/lib/dph/state_health_planning/pdf/dtc_10_27_09.pdf. Accessed on December 12, 2009.

¹⁵² Resnik, D.B. (2009). Direct-to-consumer genomics, social networking, and confidentiality. *The American Journal of Bioethics*. 9(6-7):45-58.

¹⁵³ United Nations Foundation. Mhealth for Development. See <http://www.unfoundation.org/global-issues/technology/mhealth-report.html>. Accessed on April 2, 2010.

¹⁵⁴ Academy for Educational Development (AED). Genetic Testing Marketing and Communications: A Review of Literature, 1998 – 2008. June 12, 2009. Submitted to: Trans-NIH Genetics and Common Diseases Communication Program, National Institutes of Health, Rockville, MD.

¹⁵⁵ Cogent Research, LLC. (2008). *Cogent Genomics Attitudes and Trends: 2008*. Provided to SACGHS with permission to cite, April 29, 2009.

¹⁵⁶ Lanie, A.D., Jayaratne, T.E., Sheldon, J.P., Kardia, S.L.R., Anderson, E.S., Fledbaum, M., and Petty, E.M. (2004). Exploring the public understanding of basic genetic concepts. *Journal of Genetic Counseling*. 13(4):305-320.

towards genetics.¹⁵⁷ In general, people seem to be reasonably aware that genetic risk factors contribute to health outcomes.^{156,157,158,159} However, an understanding of genetic risk factors is dependent on education and health literacy, which varies by race, ethnicity, and English language proficiency in the United States.^{160,161,162,163,164,165}

In comparing results from its surveys conducted in 2006 and 2008, Cogent Research found that overall awareness of genetics by the public increased over this time period. In 2008, 79 percent of respondents to the Cogent survey reported that they had heard or read about using individual genetic information to optimize health, and about half of respondents thought that they were informed about their family history.¹⁵⁵ Also, 50 percent of respondents were aware that genes predict the likelihood of developing specific diseases. However, among survey respondents, less than 5 percent understood that genetic information can be used to optimize health.

Smerecnik et al.¹⁵⁹ performed a literature review of studies published between 1990 and 2007 of public knowledge of genetic risk factors of multifactorial genetic diseases. These studies suggest that, on average, 59 percent of individuals surveyed were aware of the existence of genetic risk factors (range, 17.6 to 93.3 percent). Awareness of risk factors varies depending on the disease. For example, 60 percent of the general public was aware of genetic risk in breast cancer, but only 20 percent was aware of genetic factors in cervical cancer. Knowledge beyond awareness, such as processing this information and using it in decisionmaking, was far more limited.

Levels of genetic knowledge have also been found to differ by ethnicity, English language proficiency, and socioeconomic background.^{161,166} Several studies have linked level of education with knowledge of genetic concepts or genetic testing, demonstrating that, as might be expected,

¹⁵⁷ Etchegary, H., Cappelli, M., Potter, B., Vloet, M., Graham, I., Walker, M., and Wilson, B. (2010). Attitude and knowledge about genetics and genetic testing. *Public Health Genomics*. 13:80-88.

¹⁵⁸ Moscarillo, T.J., Holt, H., Perman, M., Goldberg, S., Cortellini, L., Stoler, J.M., DeJong, W., Miles, B.J., Albert, M.S., Go, R.C.P., and Blacker, D. (2007). Knowledge of and attitudes about Alzheimer disease genetics: reports of a pilot survey and two focus groups. *Community Genetics*. 10:97-102.

¹⁵⁹ Smerecnik, C.M.R., Mesters, I., de Vries, N.K., and de Vries, H. (2008). Educating the general public about multifactorial genetic disease: applying a theory-based framework to understand current public knowledge. *Genetics in Medicine*. 10(4):251-258.

¹⁶⁰ Regenstein, M., Huang, J., and West, C. (2009). Hospital Language Services: Quality Improvement and Performance Measures. In: Henriksen K, Battles JB, Keyes MA, Grady ML, editors. *Advances in Patient Safety: New Directions and Alternative Approaches*. Vol. 1. Assessment. AHRQ Publication No. 08-0034-1. Rockville, MD: Agency for Healthcare Research and Quality; August 2008.

¹⁶¹ Catz, D.S., Green, N.S., Tobin, J.N., Lloyd-Puryear, M.A., Kyler, P., Umamoto, A., Cernoch, J., Brown, R., and Wolman, F. (2005). Attitudes about genetics in underserved, culturally diverse populations. *Community Genetics*. 8(3):161-72.

¹⁶² Kennedy, B.R., Mathis, C.C., and Woods, A.K. (2007). African Americans and their distrust of the health care system: healthcare for diverse populations. *Journal of Cultural Diversity*. 14(2):56-60.

¹⁶³ Johnson, V.A., Edwards, K.A., Sherman, S.L., Stephens, L.D., Williams, W., Adair, A., and Deer-Smith, M.H. (2009). Decisions to participate in fragile X and other genomics-related research: Native American and African American voices. *Journal of Cultural Diversity*. 16(3):127-135.

¹⁶⁴ Ku, L., and Waidmann, T. (2003). How race/ethnicity, immigration status and language affect health insurance coverage, access to care and quality of care among the low-income population. Final report. Washington, D.C. Kaiser Family Foundation, Publication No. 4132. See <http://www.kff.org/uninsured/kcmu4132report.cfm>. Accessed on December 14, 2009.

¹⁶⁵ David, R.A., and Rhee, B. (1998). The impact of language as a barrier to effective health care in an underserved urban Hispanic community. *Mount Sinai Journal of Medicine*. 65:393-397.

¹⁶⁶ Kessler, L., Collier, A., and Halbert, C.H. (2007). Knowledge about genetics among African Americans. *Journal of Genetic Counseling*. 16(2):191-200.

genetic knowledge correlated with education level.^{167,168,169,170} In a study that assessed knowledge about genetics and genetic testing among 560 women in Ontario, in which 80 percent had college degrees, only 3 percent reported having no knowledge of genetics, 68 percent thought that their genetic knowledge was about the same as most people, and 21 percent reported knowing more than most people about genetics.¹⁵⁷

It might be expected that people with a family history of a specific genetic-related disorder would be more knowledgeable about genetics in general and their own risk in particular for developing the disorder. However, this has not been found in the literature. Several studies have evaluated genetics literacy among individuals with or at risk for genetic diseases. Marcheco et al. found that first-degree relatives of early-onset familial Alzheimer disease have limited knowledge of their own personal risk of developing the disease;¹⁷¹ similar findings were reported by Moscarillo et al.¹⁷² Basic genetics knowledge was generally low among persons with familial testicular cancer and their family members. Less than half (41 percent) of respondents were able to answer questions correctly regarding testicular cancer and genetics.¹⁷⁰ A study of adults with cystic fibrosis found that they have limited knowledge of the genetics of their disorder.¹⁷³ Furthermore, knowledge of genetics and genetic testing among people with chronic illness has been found to be lacking, particularly among older people and those with less education.¹⁷⁴

Most studies that assess consumers' knowledge or perceived knowledge of genetics do not take into account the confidence that respondents have in their genetics knowledge. Lanie et al.¹⁵⁶ interviewed 62 adults to assess their genetic knowledge and self awareness of their lack of knowledge. The authors found a significant number of individuals who believed they held accurate knowledge but whose responses to question were actually incorrect. Past research suggests that it is easier to educate individuals who realize their current understanding is inadequate than individuals who are unaware of their limitations.¹⁷⁵ In providing genetics education and training for patients and consumers, most resources have been geared towards those who are actively seeking information, while few methods have been proposed for how to educate those who are unaware of their lack of knowledge.

¹⁶⁷ Tambor, E.S., Rimer, B.K., and Strigo, T.S. (1997). Genetic testing for breast cancer susceptibility: awareness and interest among women in the general population. *American Journal of Medical Genetics*. 68:43-49.

¹⁶⁸ Kelly, K., Leventhal, H., Marvin, M., Toppmeyer, D., Baran, J., and Schwalb., M. (2004). Cancer genetics knowledge and beliefs and receipt of results in Ashkenazi Jewish individuals receiving counseling for BRCA1/2 mutations. *Cancer Control*. 11(4):236-244.

¹⁶⁹ Waller, J., McCafferey, K., and Wardle, J. (2004). Beliefs about the risk factors for cervical cancer in a British population sample. *Preventive Medicine*. 38:745-753.

¹⁷⁰ Peters, J. A., Beckjord, E.B., Banda Ryan, D.R., Carr, A.G., Vadaparampil, S.T., Loud, J.T., Korde, L., and Greene, M.H. (2008). Testicular cancer and genetic knowledge among familial testicular cancer family members. *Journal of Genetic Counselors*. 17:351-364.

¹⁷¹ Marcheco, B., Bertoli, A.M., Rojas, I., and Herebero, L. (2003). Attitudes and knowledge about presymptomatic genetic testing among individuals at high risk for familial, early-onset Alzheimer's disease. *Genetic Testing*. 7(1):45-47.

¹⁷² Moscarillo, T.J., Holt, H., Perman, M., Goldberg, S., Cortellini, L., Stoler, J.M., DeJong, W., Miles, B.J., Albert, M.S., Go, R.C.P., and Blacker, D. (2007). Knowledge of an attitude about Alzheimer's disease genetics: reports of a pilot survey and two focus groups. *Community Genetics*. 10:97-102.

¹⁷³ Houser, G.H., Holt, C.L., Clancy, J.P., Leon, K., Rowe, S.M., Gaggar, A., Gutierrez, H.H., Young, K.R., and Robin, N.H. (2008). Genetic and reproductive knowledge among adolescents and adults with cystic fibrosis. *Chest Journal*. 133(6):1533.

¹⁷⁴ Morren, M., Rijken, M., Baanders, A.N., and Bensing, J. (2007). Perceived genetic knowledge, attitudes towards genetic testing, and the relationship between these among patients with a chronic disease. *Patient Education and Counseling*. 65:197-204.

¹⁷⁵ Renner, C.H., and Renner, M. J. (2001). But I thought I knew that: using confidence estimation as a debiasing technique to improve classroom performance. *Applied Cognitive Psychology*. 15:23-32.

Genetic Testing Marketing and Communications: A Review of the Literature by AED

AED conducted a search of published and unpublished literature on the marketing of genetic testing. Its review emphasized DTC genome-wide scans of susceptibility markers for common diseases. The review also revealed information relevant to genetic services and information more generally. The review addressed two questions:

- What is known about current communication and understanding of genetics, genetic risk, DTC genomic services, and personalized medicine among the interested public and health care professionals?
- What are the state-of-the-art research areas or gaps in research regarding current communication and understanding of genetics, genetic risk, DTC genomic services, and personalized medicine among the interested public and health care professionals?

The search yielded 128 relevant articles published between 1998 and 2009. AED concluded from the literature that most consumers have a positive attitude toward genetic testing; however, their understanding of genetic testing is very basic, often misinformed, and does not appear to be improving over time. For example, consumers do not understand that there are many types of genetic tests, and there are many contexts in which these tests are used. Also, consumers do not have ready access to balanced and accurate information or personalized guidance about genetic tests. DTC marketing usually does not fulfill this need. Several government Internet sites provide good information about genetic testing; however, these sites are geared primarily to health care professionals. Although consumers would prefer to learn about genetic tests from their health care professionals, most physicians are not adequately trained in genetics. Physicians recognize the limitations in their knowledge and expertise and are therefore reluctant to order genetic tests and provide genetic counseling.

The AED identified several methods that would improve the public's understanding of genetic testing. Effective communication methods based on succinct, accurate, and unbiased information about genetic tests could be promoted by nonprofit and professional organizations and by government agencies. Education strategies should also consider that limited health literacy constitutes a formidable barrier to the public's understanding of genetic tests. Standardized physician training, including both didactic instruction and supervised experience in the delivery of genetic health care, would allow physicians to better educate the public about genetic tests.

Where the Public Get Its Information

The Internet has become a significant source for consumer and patient knowledge regarding genetics. A 1999 study by Stockdale revealed that even a decade ago people seeking information about the genetics of Alzheimer disease actively searched the Internet for information.¹⁷⁶ More recent studies show that Internet usage by seekers of genetic information has become more sophisticated. Schaffer et al. found that mothers of children with genetic disorders used the Internet to interpret, produce, and circulate genetic information—activities that caused them to

¹⁷⁶ Stockdale, A. (1999). Public understanding of genetics and Alzheimer's disease. *Genetic Testing*. 3(1):139-145.

value their own experiential knowledge.¹⁷⁷ In a study by Roche et al.,¹⁷⁸ 83 percent of families referred to a pediatric genetics clinic obtained information from the Internet regarding their child's diagnosis. Convenience, privacy, and finding information they did not have were cited as advantages to searching the Internet. Two reported barriers to finding relevant, understandable information were difficulties in key word searching methods that produced either too much or too little information and an inability to interpret information that was found.

In a study of perspectives on access to genetic knowledge by families of children with spinal muscular atrophy, most had received some type of genetic counseling, and families who acquired knowledge from the Internet or support groups had roughly the same amount of genetic knowledge as those who received genetic counseling from a health care professional.¹⁷⁹ A general practitioner was the preferred source of genetic information in a Dutch study of patients with chronic disease, followed by information brochures, medical specialists, and special Internet sites.¹⁷⁴ In a focus group study of culturally diverse populations recruited from U.S. community health centers, study participants obtained or wanted to obtain genetic information from television and from someone that they would trust, such as a doctor, suggesting that these would be useful mechanisms to convey genetic information in community health settings.¹⁶¹

The 2008 Cogent Research survey revealed that when participants were asked where they heard about using genetic information to understand and optimize health, 55 percent cited television; 39 percent cited newspaper or magazine stories; 28 percent cited the Internet; and 13 percent cited family members, friends, or co-workers.¹⁵⁵

The Public's Attitudes about Genetics

A number of studies have reported that people who have or think they have an understanding of basic genetics have positive attitudes towards genetic testing.^{174,180,181} Overall, the general public has been supportive of genetic testing to improve disease diagnosis and prevention.^{174,182,183} Etchegary et al. found that 95 percent of survey respondents thought genetic information should be used to improve disease diagnosis and determine why people are more or less likely to develop a disease. Seventy percent thought that genetic information should be used to design individualized drugs for people, and 85 percent believed patients should be able to receive genetic testing even if it conflicted with other family members' decisions not to undergo genetic testing. Further, 43 percent believed that doctors were obligated to share genetic information of importance to other family members, even if it violated the patient's right to privacy.¹⁵⁷ The

¹⁷⁷ Schaffer, R., Kuczynski, K., and Skinner, D. (2008). Producing genetic knowledge and citizenship through the Internet: mothers, pediatric genetics, and cybermedicine. *Sociology of Health & Wellness* 30(1):145-159.

¹⁷⁸ Roche, M.I., and Skinner, D. (2008). How parents search, interpret, and evaluate genetic information obtained from the internet. *Journal of Genetic Counselors*. 18:119-219.

¹⁷⁹ Meldrum, C., Scott, C., and Swoboda, K.J. (2007). Spinal muscular atrophy genetic counseling access and genetic knowledge: parents perspectives. *Journal of Child Neurology*. 22(8):1019-1026.

¹⁸⁰ Reitz, F., Barth, J., and Bengal, J. (2004). Predictive value of breast cancer cognitions and attitudes toward genetic testing on women's interest in genetic testing for breast cancer risk. *Psycho-Social-Medicine*. 1(3):1-13.

¹⁸¹ Jallinoja, P., and Aro, A.R. (2000). Does knowledge make a difference? The association between knowledge about genes and attitudes toward gene tests. *Journal of Health Communication*. 5:29-39.

¹⁸² Henneman, L., Timmermans, D., and van der Wal, G. (2004). Public experiences, knowledge and expectations about medical genetics and the use of genetic information. *Community Genetics*. 7:33-43.

¹⁸³ Baruch, S., Kaufman, D., and Hudson, K. (2007). *U.S. Public Opinion on Uses of Genetic Information and Genetic Discrimination*. See http://www.dnapolicy.org/pub.reports.php?action=detail&report_id=23. Accessed on November 23, 2009.

majority of respondents in this study had not thought about potential negative consequences genetic information might have for insurance coverage or employment discrimination. Regarding attitudes about genetic testing without treatment options, most participants in an Alzheimer disease study believed that testing should not be withheld until better treatment options are available.¹⁸⁴

A 2007 study by the Genetics and Public Policy Center—conducted before the passage of the Genetic Information Nondiscrimination Act of 2008 (GINA)—found that although a majority of Americans “enthusiastically support genetic testing for research and health care,” 92 percent also expressed concern that “results of a genetic test that tells a patient whether he or she is at increased risk for a disease like cancer could be used in ways that are harmful to the person.”¹⁸³ Cogent Research’s 2008 survey was conducted shortly after the passage of GINA. Despite wide media coverage around that time, only 16 percent of respondents to the Cogent survey knew that there were laws that protect the privacy of genetic information, and only one-quarter of those thought that protections were sufficient. Almost half of consumers in the Cogent survey expressed greater concern about having their DNA stored and tested without their permission than having the information be part of their medical record.¹⁵⁵

Selected Education Programs Targeted to the Public

Incorporation of genetic content into K-12 curricula has been underway for some time as a part of a greater effort to improve science literacy. Most States have curriculum content standards that include genetics and related topics.¹⁸⁵ However, 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 genetics education from single-gene, qualitative traits to complex traits.¹⁸⁶ The challenges of improving genetics education at the K-12 level are significant. However, other than acknowledging the importance of K-12 education in enhancing public understanding of genetic and genomics, it is beyond the scope of this report.

Recognizing the need for comprehensive population-based State genetics plans, some States have conducted needs assessments to better understand and define the priorities of the general public, health and human service providers, and educators. For example, the Michigan Department of Community Health—Hereditary Disorders and Newborn Screening Programs conducted a needs assessment in 2000-2002 that gathered input from 1,000 residents to develop a comprehensive State genetics plan.¹⁸⁷ One of its many conclusions was: “There is a tremendous need to educate all sectors of the population especially underrepresented communities about the role of genetics in health and disease, including related ethical, legal, and social issues. A central Michigan-focused source is needed as a portal for the public to obtain reliable information about genetic disorders, resources, and services.” As a result, an online,

¹⁸⁴ Hipps, Y.G., Roberts, S., Farrer, L.A., and Green, R.C. (2003). Differences between African Americans and whites in their attitudes toward genetic testing for Alzheimer’s disease. *Genetic Testing*. 7(1):39-44.

¹⁸⁵ American Society of Human Genetics. Genetics Education State Standards. See http://www.ashg.org/education/k12_statestandards.shtml. Accessed on November 24, 2009.

¹⁸⁶ Dougherty, M.J. (2009). Closing the gap: inverting the genetics curriculum to ensure an informed public. *American Journal of Human Genetics*. 85:6-12.

¹⁸⁷ Beene-Harris, R., and Bach, J.V. (2009). Michigan Genetics Plan: a report on the needs assessment process. *Health Promotion Practice*. 10(2): 201-209.

Michigan-focused genetics resource center providing a central source of information on genetic health care and related topics was developed and a toll-free number established.¹⁸⁸

The importance of a central location for accessible online information for consumers was recognized by the crafters of the Newborn Screening Saves Lives Act of 2008.¹⁸⁹ This legislation earmarked funding and directed the Health and Resources Service Administration (HRSA) to develop a clearinghouse for newborn screening education, family support, and services information that would be interactive, regularly updated, and link to government and nonprofit websites. The Genetic Alliance was awarded the contract to develop this clearinghouse—in partnership with other organizations such as the National Newborn Screening Genetics Resource Center, the Genetics and Newborn Screening Regional Collaborative Groups, March of Dimes, and the Association of Public Health Laboratories—and began work on the project in September 2009.¹⁹⁰

In recent years there has been a concerted effort to increase awareness of the importance of family history and to promote the use of family medical history as an education and screening tool for determining disease risk. The Office of the Surgeon General, NIH, CDC, and HRSA have been particularly active in this public health campaign.

B. HEALTH DISPARITIES

Like other areas in which health disparities exist, disparities in genetic services can occur if there is lack of awareness of, and access to, genetic testing and follow-up services. When the standard of care is to offer a genetic test, there is some evidence that minorities do not participate in genetic testing or are not offered genetic counseling services as often as whites.¹⁹¹ Whether these disparities can be attributed to lack of access to genetic testing versus insufficient knowledge of and/or attitudes about genetic testing is not clear.

An analysis of the use of genetic services (for breast/ovarian/colon cancer, Huntington disease, and sickle cell disease) by U.S. primary care physicians shows that up to two-thirds of those surveyed ordered genetic tests, and more than three-quarters referred patients for genetic counseling.¹⁹² However, there were clear differences in patterns of genetic service referrals, with providers serving minority populations being significantly less likely to order genetic testing or issue referrals.

¹⁸⁸ Michigan's Genetics Resource Center. See <http://www.migeneticsconnection.org/>. Accessed on November 25, 2009.

¹⁸⁹ U.S. Government Printing Office. *Public Law 110-204 – Newborn Screening Saves Lives Act of 2007*. See <http://www.gpo.gov/fdsys/pkg/PLAW-110publ204/html/PLAW-110publ204.htm>. Accessed on February 25, 2010.

¹⁹⁰ Newborn Screening Clearinghouse. See <http://www.nbsclearinghouse.org/about>. Accessed on February 25, 2010.

¹⁹¹ Armstrong, K., Micco, E., Carney, A., Stopfer, J., and Putt, M. (2005). Racial differences in the use of BRCA 1/2 testing among women with a family history of breast or ovarian cancer. *Journal of the American Medical Association*. 293(14):1729-1736.

¹⁹² Sheilds, A.E., Burke, W., and Levy, D.E. (2008). Differential use of available genetic tests among primary care physicians in the United States: results of a national survey. *Genetics in Medicine*. 10(6):404-414.

Several studies comparing knowledge of genetic testing in general¹⁷⁰ and genetic testing specifically for BRCA mutations and cancer risk¹⁹³ have shown lower knowledge levels among African Americans and Latinos as compared to whites. One study suggested that higher levels of acculturation—or the degree to which minorities have adopted the attitudes, values, and behaviors of the majority culture—predict familiarity with genetic testing and perceived benefits among Latinos in New York City.¹⁹⁴

Research regarding differences in attitudes about genetics between African Americans and whites has been equivocal. African Americans and Latinos overall hold a positive view of genetics, but it is not as positive as whites.^{28,184,194,195} Among inner city African Americans and whites, 90 percent of survey participants thought that genetic testing to identify risk of getting a disease was a good idea regardless of race.¹⁹⁵ Nonetheless, concerns about genetic testing leading to racial discrimination are higher in African Americans compared to whites.^{195,196} In two studies exploring attitudes about genetic testing for Alzheimer disease, African Americans expressed less interest in genetic testing but anticipated less negative personal consequences from a positive result compared to whites.^{172,184}

Recent research on the relationship between ethnicity and minority status and socioeconomic status (SES) on awareness and uptake of genetic testing has resulted in inconsistent findings. While Bowen et al.¹⁹⁷ did not find any differences in SES and reactions to a DTC campaign for *BRCA1* and *BRCA2* genetic testing, their study did reveal that, in general, women of lower SES reported less knowledge about genetics and risk, yet more interest in genetic testing. Awareness of genetic testing for cancer susceptibility is lower among racial and ethnic groups compared to whites, but it is important to look more closely at the specific SES factors in addition to race and ethnicity. Education, country of origin, insurance coverage, and parental history of cancer have been found to influence awareness. These factors differ across racial and ethnic groups, suggesting that policy remedies are unlikely to have uniform population effects, and customized strategies using culturally relevant media and native languages are needed among different groups or communities.¹⁹⁸

Another factor to consider in health disparities related to genetic services pertains to literacy and English language proficiency. The U.S. Census Bureau recently reported that 13 percent of Americans had not completed high school in 2008,¹⁹⁹ and from 2000 census data, 21 million

¹⁹³ Mogilner, A., Otten, M., Cunningham, J.D., and Brower, S.T. (1998). Awareness and attitudes concerning BRCA gene testing. *Annals of Surgical Oncology*. 5(7):567-568.

¹⁹⁴ Sussner, K.M., Thompson, H.S., Valdimarsdottir, H.B., Redd, W.H., and Jandorf, L. (2009). Acculturation and familiarity with, attitudes towards and beliefs about genetic testing for cancer risk within Latinas in east Harlem, New York City. *Journal of Genetic Counselors*. 18:60-71.

¹⁹⁵ Zimmerman, R.K., Tabbarah, M., Nowalk, M.P., Raymund, M., Jewell, I.K., Wilson, S.A., and Ricci, E.M. (2006). Racial differences in beliefs about genetic screening among patients at inner-city neighborhood health centers. *Journal of the National Medical Association*. 98(3):370-377.

¹⁹⁶ Peters, N., Rose, A., and Armstrong, K. (2004). The association between race and attitudes about predictive genetic testing. *Cancer Epidemiology, Biomarkers & Prevention*. 13(3):361-365.

¹⁹⁷ Bowen, D.J., Harris, J., Jorgensen, C.M., Myers, M.F., and Kuniyuki, A. (2010). Socioeconomic influences on the effects of a genetic testing direct-to-consumer marketing campaign. *Public Health Genomics*. 13(3):131-42.

¹⁹⁸ Pagan, J.A., Su, D., Li, L., Armstrong, K. (2009). Racial and ethnic disparities in awareness of genetic testing for cancer risk. *American Journal of Preventative Medicine*. 37(6):524-530.

¹⁹⁹ United States Census Bureau. *U.S. Census Bureau News 2009*. See <http://www.census.gov/Press-Release/www/releases/archives/education/013618.html>. Accessed on January 6, 2010.

Americans speak English “less than well.”²⁰⁰ This low level of English language proficiency is more common in minority populations and limits access to medical care, specifically decreased health care visits.²⁰¹ Socioeconomic factors underlie educational level and may account for the increased difficulties disadvantaged individuals will have with health literacy in general, and with specific understanding of the role genetics plays in maintaining health and in defining disease risks.²⁰² One strategy that could address literacy as a barrier in health care is identifying those with lower literacy. A widely used tool to assess general health literacy has been adapted and validated specifically to identify patients with low literacy in genetics.²⁰³

When genetics educational materials are available, they are not always provided in a culturally appropriate fashion, in a language that is used or understood in immigrant or ethnic communities, or provided in formats or through media that disadvantaged communities can access or utilize. Therefore, addressing health disparities through education about genetics will require innovative methods, culturally sensitive translations, and use of locally predominant languages to reach all communities. Research has found that patients who inquire about or request a genetic test serve as an inducement to physician use of genetic services.²⁰⁴ Thus, the use of strategies customized to specific groups and communities may be an effective way to promote the use of emerging genetic technologies, when medically appropriate, and empower a wide variety of consumers to act as their own health care advocates.²⁸ Programs such as the Community Genetics Education Network (CGEN) Project²⁰⁵ reinforce the need to use principles of community-based participatory research to identify effective ways to increase genetic literacy among diverse populations.

The lack of awareness and understanding about genetics in clinical practice and public health also plays out in research settings. The promise of genetics may not benefit those who do not participate in genetics research. Studies have found that minorities in the United States are less likely to participate in research, including genetics research.^{28,184} Without the participation of all segments of the population, it will be difficult to tailor treatments and preventive measures for specific subpopulations or for individuals. For example, limited participation in research by minorities becomes problematic as pharmacogenomic research uncovers variance in the efficacy of treatments and drug development and increasingly focuses on products tailored to individual risk.

²⁰⁰ United States Census Bureau. *America Speaks: A Demographic Profile of Foreign-Language Speakers for the United States: 2000*. Table 1a. See <http://www.census.gov/population/socdemo/hh-fam/AmSpks/01%20US%20per.xls>. Accessed on January 6, 2010.

²⁰¹ Shi, L., Lebrum, L., and Tsai, J. (2009). The influence of English proficiency on access to care. *Ethnicity and Health*. 14(6):625-642.

²⁰² Murray, E., Lo, B., Pollack, L., Donelan, K., and Lee, K. (2004). Direct-to-consumer advertising: public perceptions of its effects on health behaviors, health care, and the doctor-patient relationship. *Journal of the American Board of Family Practice*. 17(1):6-18.

²⁰³ Erby, L.H., Roter, D., Larson, S., and Cho, J. (2008). The rapid estimate of adult literacy in genetics (REAL-G): a means to assess literacy deficits in the context of genetics. *American Journal of Medical Genetics*. 146A:174-181.

²⁰⁴ Wideroff, L., Freeman, A., Olson, L., Klabunde, C.N., Davis, D., Srinath, K.P., Croyle, R.T., and Ballard-Barbash, R. (2003). Physician use of genetic testing for cancer susceptibility: results of a National survey. *Cancer Epidemiology, Biomarkers and Prevention*. 12:295-303.

²⁰⁵ Community Genetics Education Network Project. See <http://www.nchpeg.org/slides/2009/Kyler.ppt#271.2>. Accessed on March 18, 2010.

Racial and ethnic health disparities may be exacerbated if researchers assume that the basis of health disparities is solely due to genetics and conduct research in a way that seems to affirm a genetic basis for racial differences in disease prevalence.^{206,207} This assumption arises when researchers overemphasize the genetic contributions to disease and health without consideration of social contributions to health.²⁰⁶ Recommended educational efforts for both the public and the scientific community should therefore focus on ameliorating attitudes about the purposes of genetic research and increasing the understanding of the complex interrelation of genes and the environment, including social contributions to health. Adequate knowledge is also required for research participants to provide meaningful informed consent in complex genetic studies. The recognition that health disparities are heavily rooted in social structure requires that educational efforts acknowledge the broader context of socioeconomics, cultural attitudes, educational level, literacy, gender, and English-language proficiency in order to educate both the scientific community and the public.²⁰⁸

²⁰⁶ Bonham, V.L., Citrin, T., Modell, S.M., Franklin, T.H., Bleicher, E.W.B., and Fleck, L.M. (2009). Community-based dialogue: engaging communities of color in the United States' genetics policy conversation. *Journal of Health Politics, Policy and Law*. 34(3):325-359.

²⁰⁷ Sankar, P., Cho, M.K., Condit, C.M., Hunt, L.M., Koenig, B., Marshall, P., Lee, L.S-J., and Spicer, P. (2004). Genetic research and health disparities. *Journal of the American Medical Association*. 291(24):2985-2989.

²⁰⁸ Ramos, E. and Rotimi, C. (2009). The A's, G's, C's, and T's of health disparities. *BMC Medical Genomics*. 2:29.

III. SACGHS Surveys

SACGHS collected data from selected organizations with missions related to health professional education, public health, and consumer and patient advocacy to obtain information regarding their activities in genetics education. SACGHS also surveyed selected Federal agencies to learn about their efforts that support genetics education and training.

A. SACGHS SURVEY OF HEALTH CARE PROFESSIONAL ORGANIZATIONS

Methodologies

In 2008, SACGHS surveyed selected organizations with responsibilities across the continuum of health professional education to obtain information regarding their activities in genetics education. Key staff members in 60 targeted organizations were contacted via e-mail to respond to a survey developed by SACGHS (see Appendix D-1 for a listing of these organizations).²⁰⁹ The survey explored several major themes including the organizations' perceived role in, and priority ascribed to, genetics education; barriers that impede their role in genetics education; and a description of their past, present, and planned efforts concerning genetics education. Appendix D-2 provides additional information on the methodologies, and the survey instrument is provided in Appendix D-3.

Selection criteria for inclusion in the survey included the organization's role in training professionals destined to provide primary care services and the diversity of levels of training within the organization. Organizations that play a central role in training nurses and primary care physicians, as well as organizations representing genetic professionals with a key role in supporting nongenetics health professionals, were specifically targeted. In addition, three federal advisory committees relevant to genetics education were invited to complete the survey.

Limitations

This survey has several important limitations, including that the sampling of organizations was nonrandom and relied on self-reported, qualitative data, which do not allow generalization across health care professional organizations. Survey data revealed that, as would be expected, many of the larger organizations not focused on genetics have no formal policy or organizational structure for genetics education, thus responses to the survey questions may represent the opinion of the respondent rather than the consensus or policy of the organization. Organizations representing professionals from racial and ethnic minority groups were disproportionately represented among nonresponders. Additionally, organizations that engage in genetics education and training may have been more likely to respond to a survey regarding this topic.

²⁰⁹ The survey was determined to be exempt from the need for Institutional Review Board review and approval by the NIH Office of Human Subjects Research.

Survey Findings

Organizations' Roles and Responsibilities and Barriers to Providing Genetics Education

Survey responses were received from 36 organizations, a 60 percent response rate. (See Appendix D-4 for a list of the responding organizations and their reported number of members or constituents.) Nearly 70 percent of responding organizations considered genetics education and training to be a role or responsibility of their organization (see Appendix D-5, Table 1), although the size and importance of that role varied according to organizational mission and focus (see Appendix D-5, Table 2). Most of these organizations reported they are able to fulfill this role or responsibility (see Appendix D-5, Table 3). Strategies to promote education and training included providing increased funding, evaluating current activities, and generating greater interest with institutional leaders and through publications and annual conferences (see Appendix D-5, Table 4). Only 36 percent of nongenetic-specific organizations reported that they have an established committee, workgroup, or dedicated staff that deals specifically with topics in genetics relevant to their organization's mission. In contrast, two-thirds of genetics-specific organizations reported this activity. (See Appendix D-5, Table 5.)

In response to a series of Likert-scale questions, 30 of 36 organizations considered the development and promotion of general, health-related educational activities to be very important, but fewer organizations (17 of 35) considered genetics-specific educational activities to be very important. In addition, less than one-third of respondents indicated that genetics education has a high priority relative to the organization's overall priorities, and only seven respondents noted that their membership was extremely satisfied with their organization's emphasis on genetics education. About half of respondents considered their organization's leadership to have moderate or strong expertise in genetics education. (See Appendix D-5, Tables 6-10].

The survey also asked participants about barriers to their organizations' ability to provide genetics and genomics education and to rank these barriers from most to least important. Among respondents who provided rankings, the most important barrier was competing priorities within the organization. Other significant barriers were that genetics was not emphasized in certifying exams and credentialing standards, which diminishes incentives for offering education programs, and the lack of accessible educational resources. Five organizations responded there were no barriers, and four indicated that genetics and genomics education was not applicable to the organization's mission. (See Appendix D-5, Tables 11-12.)

Membership Needs, Priorities, and Engagement in Genetics Education

More than half of the respondents have either directly surveyed or received indirect input from their membership about genetics education needs or priorities. Genetic-specific organizations survey routinely, while other organizations obtain input on a more *ad hoc* basis, such as from educational meeting evaluations and/or general needs assessments. The organizations' membership surveys revealed several needs in genetics education such as information about specific genetic disorders, the availability of genetic tests and services, how to interpret genetic test results and family history, and the changing roles of genetic counselors as pharmacogenomic

testing and whole-genome sequencing become more widely available. (See Appendix D-5, Table 13.)

Organizations articulated common themes regarding programs or resources that could enhance the engagement of their organization's members in genetics education if federal funding were available. These included educational grants for faculty training and program development; development of point-of-care tools and tool kits; research and dissemination of evidence-based guidelines; and increased integration of genetics into clinical decision support, electronic medical records, and performance standards. In addition, a registry of genetic tests would facilitate the evaluation of clinical validity and utility and thus, inform genetic test usage in the clinical setting. (See Appendix D-5, Table 14.)

Survey participants were asked to describe genetics initiatives and programs that their organization has implemented in the last 5 years or plan to implement in the near future. The majority of these initiatives included competencies, curricula, publications, and conferences. (See Appendix D-5, Tables 15-16 for additional details.)

Organizations' Mission-Specific Responses

Organizations were asked to answer open-ended questions in one of four categories most relevant to their mission (three organizations answered questions in more than one category). Of the 34 organizations responding, 18 chose education and training of health professionals as most relevant; 12 selected advocacy and support of practicing professionals; six cited certification of health professionals, and five chose accreditation or certification of institutions. Selected comments for each category are provided below.

Education and Training of Health Professionals

Eighteen respondents identified education and training of health professionals as a relevant mission of their organization (see Appendix D-5, Table 17). Fourteen of these respondents thought that integrating genetics into the curriculum and training health professionals are critical needs; however, some organizations indicated that these efforts were not a high priority, and one suggested an uncertain clinical benefit of this approach (See Appendix D-5, Table 18). Several organizations have implemented integrated curricula or offer optional genetics curriculum components (see Appendix D-5, Table 19).

In response to an open-ended question about gaps in genetics education, respondents identified the following needs in genetics education: faculty capable of teaching genetics, the inclusion of genetics in educational materials, effective approaches to educate health professionals about the relevance of genetics, and an understanding of the impact of educational programs on clinical practice. Strategies to address these gaps included additional funding, providing faculty with genetics expertise, supporting curricula development, involving educators with diverse backgrounds, improving access to online genetic educational activities, and demonstrating the clinical relevance of genetics. (See Appendix D-5, Table 20.)

Cultural competency related to genetics education of health professionals was identified as an urgent need by health professional organizations through a roundtable discussion and a survey conducted by SACGHS in 2004.^{210,211} In the 2008 survey, the 18 organizations identifying education and training as part of their mission were asked whether steps were taken to incorporate cultural competency into curricula. Thirteen of the 14 organizations responding to this question stated that cultural competency is part of the curricula or is an accreditation requirement. (See Appendix D-5, Table 21.)

Asked to look ahead 5 to 10 years and anticipate needs in genetics education, organizations provided varied responses ranging from the need to be able to interpret genetic test results and know when to refer patients, to more general statements about the need for health care providers to be knowledgeable about genetics. The need to understand risks for complex diseases was mentioned, as was the ability to assess risks using multiple tools, change patient management based on risk, and communicate risk effectively. (See Appendix D-5, Table 22.)

Advocacy and Support of Practicing Health Professionals

Twelve organizations identified their primary mission as advocacy and support of practicing health professionals. (See Appendix D-5, Table 23 for a description of organization missions.) Several respondents indicated that their organization's membership needs more information about genetics. Suggested topics included information about common genetic diseases, pharmacogenomics, the importance of family history, validity and utility of genetic tests, how to integrate genetics into practice and effectively triage genomic services, and the latest technologies and practices in genomics-based preventive medicine (see Appendix D-5, Table 24.) Six organizations suggested approaches to promote improved knowledge of genetics among their constituencies including the need for more funding for educational opportunities, such as continuing education, resource portals, training of clinical educators and internship supervisors, and certification of professionals. (See Appendix D-5, Table 25.)

Certification of Health Professionals

Organizations were asked if current credentialing exams include questions on genetics. Four genetic-specific organizations (American Board of Genetic Counseling (ABGC), American Board of Medical Genetics, American College of Medical Genetics, and Genetic Nursing Credentialing Commission) reported that all or most of their credentialing exams focused on genetic content. One nursing certification organization (Oncology Nursing Certification Corporation) reported genetic content but at less than 5 percent of total content, and one general professional organization (the Society of General Internal Medicine) reported genetic content at less than 1 percent of total content. (See Appendix D-5, Table 26.)

²¹⁰ Secretary's Advisory Committee on Genetics, Health, and Society. Genetics Workforce, Education, and Training, October 23, 2003. See http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_2003_oct_23.html. Accessed on February 17, 2011.

²¹¹ Secretary's Advisory Committee on Genetics, Health, and Society. Report of the SACGHS Task Force on Genetics Education and Training, June 14, 2004. See http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_2004_jun_14.html. Accessed on February 22, 2010.

Accreditation or Certification of Institutions

Five organizations considered accreditation or certification of institutions as their primary role. These organizations view integration of genetics into the curriculum and training of health professionals as important, and four of the five organizations regularly update curriculum requirements. (See Appendix D-5, Table 27.)

SACGHS Surveys of Health Professional Organizations: Comparison of 2004 and 2008 Surveys

In 2004, 26 organizations were invited via e-mail to respond to a survey that consisted of seven open-ended questions. These organizations were divided into three groups based on their primary role as either genetics specific, professional education, or general professional organizations. Survey results were reported to SACGHS on June 14, 2004.²¹¹ Thirteen responses were received (50 percent). (See Appendix D-6 for a list of the organizations that responded.)

In an effort to assess progress made in the intervening years, SACGHS administered a follow-up survey in 2008. The 2008 survey was formatted differently than the 2004 survey and additional questions were included; however, there were some areas where comparisons could be made. Due to the small number of responses to specific questions in 2004, generalizing more broadly beyond the specific organizations is not possible.

Integration of Genetics into the Curriculum and Training of Health Professionals

Both surveys asked organizations that identify education of professionals as their primary mission to characterize the need for integrating genetics into the curriculum and training of health professionals. In 2004, eight organizations responded that while the need for integration varies, health professionals must be able to address patients' questions, which requires a solid, basic knowledge of genetics and a lifelong commitment to learning. By 2008, 15 of the 18 organizations responded similarly to this question, and several have actually implemented a genetics curriculum. However, some organizations thought that this effort was not a high priority, and one suggested an uncertain clinical benefit for integrating genetics into the curriculum and training of health professionals.

Barriers to Providing Genetics Educational Activities

In the 2004 survey, organizations reported the lack of case models or evidence for clinical application of genetics and competing priorities as some of the top barriers to providing genetics education activities. Based on the 2008 survey responses, the top barriers were competing priorities and that genetics content is not emphasized on certifying exams or in credentialing standards. Comparison of the two surveys indicates that competing priorities remain a significant barrier to providing genetics education activities, but the lack of evidence supporting clinical effectiveness was much less of a barrier by 2008.

Future Directions in Genetics Education of Health Care Professionals

Survey respondents in 2004 and 2008 identified the following factors critical to future directions in educating health professionals:

- The government has a role in supporting genetics education programs.
- Genetics education must be provided across the entire continuum of medical education.
- Funding should target educational programs that are known to change clinician behavior and should include interactive learning with case studies that emphasize clinical application of genetics.
- Education and training should address the importance of obtaining family history.
- There is a need to expand cultural diversity within the health professional workforce and to improve the cultural competency of health professionals in genetics and genomics.

B. SACGHS SURVEY OF PUBLIC HEALTH PROVIDERS

Methodologies

To assess the genetics education and training needs of public health providers, SACGHS developed 12 competencies based on competencies established by public health organizations and institutions (see Box 1). The SACGHS-developed competencies were then used in an online survey²¹² (based on the work of Kirk et al.²¹³ and modified by SACGHS) with the intent of assessing public health providers' opinions on the importance of each competency, their confidence in demonstrating each competency, and the frequency with which they apply each competency. The survey was distributed to approximately 500 public health providers with varying degrees of genetics responsibilities. (See Appendix E-1 for details of the survey methodology, participant recruitment; Appendix E-2 for screen shots of the online survey instrument; and Appendix E-3 for an explanation of survey reliability results.)

Box 1: Twelve Competencies Used in the SACGHS Survey

The following 12 competencies were used in the survey. They are based on skills and knowledge thought to be critical for practicing providers of public health, whether at the local, state, or national level:

1. Maintain up-to-date knowledge on the development of genomic science and technologies within his or her professional field and program to apply genomics as a tool for achieving public health goals.
2. Demonstrate basic knowledge of the role that genetics and genomics plays in the development of disease, and in screening and interventions for programs of disease prevention and health promotion.
3. Describe the importance of family history in assessing predisposition to disease.

²¹² The survey was determined to be exempt from the need for Institutional Review Board review and approval by the NIH Office of Human Subjects Research.

²¹³ Kirk, M., Tolkin, E., and Birmingham, K. (2007). Working with publishers: a novel approach to ascertaining practitioners' needs in genetics education. *Journal of Nursing Research*. 12:597-615.

4. Identify opportunities and integrate genetic and genomic issues into public health practice, policies or programs effectively.
5. Maintain up-to-date knowledge of genetics and genomics-related policies, legislation, statutes, and regulations.
6. Describe the potential physical and psychological benefits, limitations, and risks of genetic and genomic information for individuals, family members, and communities.
7. Collaborate with existing and emerging health agencies and organizations, academic, research, private and commercial enterprises, and community partnerships to apply genetics and genomics knowledge and tools to address public health problems.
8. Identify the resources available to assist clients seeking genetic and genomic information or services, including the types of genetics professionals available.
9. Conduct outcomes evaluation of available genetic and genomic programs and services to determine their effectiveness.
10. Identify the political, legal, social, ethical, and economic issues associated with integrating genomics into public health.
11. Use information technology (IT) to obtain credible, current information about genetics; utilize IT skills to share data and participate in research, program planning, evaluation, and policy development for health promotion and disease prevention.
12. Identify appropriate and relevant genetics research findings that can be translated into public health policies or practices.

Limitations

This survey has a number of limitations that affect the ability to generalize the findings. The sample was one of convenience and relied on snowball sampling²¹⁴ to increase the number and scope of participants. Given the need to keep the survey anonymous, it was not possible to obtain informative data about the survey participants.

By targeting dissemination of the survey to individuals more likely to incorporate genetics into their daily practice (e.g., State genetic coordinators), the data are unlikely to be representative of the opinions and activities of the entire public health workforce. Based on the responsibilities of the individuals to whom the survey was sent directly, the data are more likely to represent the “best case scenario,” meaning that the responses are more strongly supportive of the importance of genetics and the relevance of the competencies than might be expected from the public health workforce as a whole. Because of the snowball sampling methodology of dissemination, it was not possible to determine if the sample was representative of public health workers. Even if it were possible to disseminate a survey to all public health workers, individuals using genetics in their jobs would be more likely to participate.

The competencies that formed the basis of this assessment of education and training needs were derived from existing sources through an expert opinion process and were not independently validated. The data and their interpretation are also limited by the self-assessment nature of this survey. There is no objective measure that can be used to determine the accuracy of the self-assessment.

²¹⁴ van Meter, K.M. (1990). Methodological and design issues: techniques for assessing the representatives of snowball samples. *NIDA Research Monograph*. 98:31-43.

Survey Findings

SACGHS received and analyzed 140 survey responses. It is not possible to calculate response rate because the total number of individuals who eventually received the survey is not known.

Competencies

Survey participants were asked to rank the competencies based on importance, how confident they are in demonstrating the competency, and how frequently they apply the competency.

The majority of individuals responded that all 12 competencies are important; however, the three competencies that were ranked as the most important to public health providers were: (1) demonstrating basic knowledge of the role of genetics in disease development; (2) describing the importance of family history in assessing predisposition to disease; and (3) identifying opportunities and effectively integrating genetic issues into public health practice, policies, or programs (see Appendix E-4, Table 1 for summary data).

Responses varied with regard to the level of confidence in demonstrating the competencies. Two competencies that were ranked as the most important were also those in which respondents indicated the most confidence—the importance of family history and basic knowledge of the role of genetics in disease. The competency that ranked lowest in the level of confidence was the ability to conduct outcomes evaluation of available genetic and genomic programs and services to determine their effectiveness.

Responses also varied in the frequency that the competencies were applied. It appears that there are instances where public health providers never or rarely apply a specific competency, or conversely they very frequently apply a specific competency. Demonstrating basic knowledge of the role of genetics in disease and maintaining up-to-date knowledge on the development of genetic science and technologies within one's professional field were reported to be most frequently applied by public health providers (monthly or weekly). The competency that was ranked lowest and applied rarely (1 or 2 times per year) was conducting outcome evaluation of available genetic programs and services to determine effectiveness. Overall, there appeared to be no competency that was unimportant or irrelevant to these survey respondents.

The Importance of Genetics to Institution Leadership

Sixty percent of survey respondents reported that their leadership believes that genetics knowledge is important or very important to the respondent's job responsibility, while 21 percent responded that their leadership places little or no importance on such knowledge. Thirty-five percent of respondents indicated that their senior administration believes that genetics knowledge is important or very important for the administration's job responsibilities, and 35 percent reported that it was of little or no importance. (See Appendix E-4, Table 2 for summary data.)

One-third of respondents reported that they feel they have adequate to very adequate resources for implementing genetic competencies in their work or role, while two-thirds reported that the

resources they have are not adequate or are only somewhat adequate. (See Appendix E-4, Table 3 for summary data.)

Work Settings and Responsibilities

The survey queried the work settings of respondents (e.g., government, academia, community-based organization). Most respondents work at the state level (41 percent), followed by academia (30 percent), federal level (13 percent), private, nonprofit organizations (9 percent), community-based organizations (4 percent), other institutions such as commercial laboratories, medical center community programs, or nonprofit health organizations (2 percent), and international positions (1 percent). (See Appendix E-4, Table 4.) The survey also asked about time spent on tasks in the following areas: administration, program planning, direct consumer care, research, policy, assessment and evaluation, and education and training. Few respondents dedicated most or all of their time to a specific area; most divided their time across several areas (see Appendix E-4, Table 5).

Delivery of Genetic Services to Underserved or Vulnerable Populations

Using an open-ended question format, respondents were asked to describe organizational efforts to ensure that genetic services or information are available for vulnerable or underserved populations and to recommend specific strategies (70 responses were received). Responses included: provision of educational materials and development of websites; encouraging community involvement, training and education of public health providers; and provision of genetic services. Increased funding and development of federal policies were also suggested as ways to enhance educational efforts. (See Appendix E-5, Table 6 for more detailed responses.)

Other Topics

The last survey item offered an opportunity for respondents to provide additional comments on genetics or genetics education (54 individuals chose to submit comments). The comments included points regarding the need for funding to develop, implement, and evaluate genetics education and training programs; the importance of collaboration across local, State, and Federal programs; the need for training to identify and use best practices in genetics education; and the importance of building leadership support for educational activities. (See Appendix E, Table 7 for details.)

C. SACGHS SURVEY OF CONSUMERS AND PATIENTS

Methodologies

To elucidate the genetics education needs of patients and consumers, here defined as members of the public who seek genetic information, SACGHS collected qualitative and quantitative data using semi-structured interviews with professionals working in consumer and patient health education. In addition, SACGHS conducted a web-based survey of the health advocacy community.

Semi-Structured Interviews

A list of individuals widely regarded as leaders in genetics education and advocacy for consumers and patients was generated by SACGHS, as well as by attendees of an annual NCHPEG meeting. Between December 2008 and February 2009, SACGHS conducted semi-structured interviews²¹⁵ with 11 experts in the fields of disease and disability advocacy, genetics services for patients, health education and communication, for-profit DTC genetics services, and science and genetics education of the public. (See Appendix F-1 for a list of interviewees and interview guide.) The purpose of the interviews was to collect data on current and emerging needs for knowledge of genetics among consumers and patients and to inform the development of a survey.

Web-Based Survey

Based on the interviews, SACGHS developed a 12-item online survey instrument (see Appendix F-2) to collect data from the genetics and health advocacy communities regarding their opinions on the education needs of patients and the general public. During April and May 2009, the survey was distributed to representatives of health advocacy groups, community-based health-focused organizations, and communities specializing in genetics education for the public (see Appendix F-3.). In addition, a special arrangement was made for the Genetic Alliance,²¹⁶ a nonprofit health advocacy organization, to distribute the survey to its membership.

Data Limitations

Qualitative research is ideal for exploring complex themes such as those presented in this report. However, there are limitations to qualitative data including the potential for selection bias and social desirability in responses. There are also limitations to the SACGHS online quantitative survey. A random sampling strategy was not used and stakeholders and the public who responded are not necessarily representative of the general public and may have had unique interests or experiences that led to their participation in the survey. Another potential limitation is the possibility of response bias.

SACGHS sought to minimize any limitations in the data used for this report by using multiple data collection methods. The qualitative approach allowed for in-depth discussion and exploration of themes, and the online survey included opinions from those who are or have been seekers of genetic information. Even with the limitations noted above, this process did provide a snapshot of the needs of consumer and patients who have varying degrees of involvement in genetics.

²¹⁵ The survey was determined to be exempt from the need for Institutional Review Board review and approval by the NIH Office of Human Subjects Research.

²¹⁶ Genetic Alliance. See <http://www.geneticalliance.org>. Accessed on August 18, 2009.

Survey Findings

Semi-Structured Interviews

The interview data revealed common themes related to the educational needs of consumers and patients, successful educational models, and recommended actions the government can take to improve the public's understanding of genetics. The first set of themes relate to perceptions about consumers' understanding of genetics and genomics. Specifically, consumers are finding it difficult to understand new advances in genetic technologies and the potential benefits and risks of these technologies, how genes and behaviors relate to each other, and that a single condition may involve multiple risk factors. Interview findings also suggest that consumers frequently misunderstand the concept of genetic predisposition as well as current limitations in knowledge about test validity and utility. These misunderstandings are compounded by the difficulty consumers have in finding accurate information.

The interviews also explored various approaches to genetics education. Suggestions included the need to improve genetics education for health care providers and to recognize that collaborative projects between public and private organizations can facilitate the identification of specific educational needs. Respondents suggested that an important first step in developing programs is to assess and understand the needs of specific communities. They also suggested that the Internet could be used effectively as a source of balanced, accurate information.

The third set of themes relates to the role of government in educating the public. Respondents suggested that consumers believe that the Federal Government is a more unbiased source of information than commercial sources and that it should have a central role in genetics education of the public. Consumers also think that government should monitor the societal effects of genetic testing and services, clarify the extent to which laboratory tests are regulated, support formal genetics education in schools, and have some influence over educational standards. In addition, those interviewed suggested that government should fund more programs to improve genetic literacy. (See Appendix F-4, Table 1, for additional details of key findings from semi-structured interviews.)

Web-Based Survey Results

There were 337 complete or partial responses to the survey. Based on 256 responses to the question about geographic location, survey participants were located in 39 states plus the District of Columbia. California, Maryland, New York, and the District of Columbia had the largest number of responses. (The geographic distribution is shown in Appendix F-5, Figure 1.)

Respondents represented a wide variety of organizations, including health care organizations, advocacy groups, academic institutions, private industry, public health organizations, community-based health organizations, nonprofit organizations and support groups, and private hospitals and health care practices. (This distribution is presented as a pie chart in Appendix F-5, Figure 2.) Based on 269 responses, 75 percent of respondents considered genetics important or very important to their organization's mission (see Appendix F-5, Table 2), and more than half

of respondents reported planning or implementing genetics education programs for seekers of genetic information (see Appendix F-5, Table 3).

Participants were asked to rank a set of five concepts that individuals most need to know about genetics and genomics to be informed seekers of genetic information as it relates to health (see Appendix F-5, Table 4). Among the respondents answering this question, the most important concept was that “family history is an important tool for understanding health and disease.” In a free-text box, 59 additional responses were provided and included the following concepts: (1) there is a difference between disease risk and disease diagnosis; (2) understanding probabilities and terms like “common” and “rare” are essential to interpreting the results of genetic tests; and (3) genetic tests should be interpreted by knowledgeable people. (See Appendix F-5, 4.1.)

Survey participants were asked to rank a set of four topics that “may have special relevance for seekers of genetic information as it relates to health.” (See Appendix F-5, Table 5.) The topic selected most among the respondents was “where to find reliable genetic and genomic information,” suggesting that the ability to direct consumers to such resources may fill an important and widely recognized gap. (See Appendix F-5, 5.1 for free-text responses on this topic.)

Survey participants also were asked to rank the genetics education and services needs of underserved and vulnerable populations. (See Appendix F-5, Table 6.) The need for basic and relevant genetic health information was ranked highest, and education about access to genetic services was ranked lowest. Skills to make informed health decisions and culturally appropriate genetic health information were ranked second and third, respectively (See Appendix F-5, 6.1 for free-text responses on this topic.)

Respondents were asked if they were part of an organization and, if so, to report whether their organization had created educational programs to address the challenges in underserved and vulnerable populations. Fifty-six percent of respondents (189 of 337) answered this question, reporting development of education programs to address at least one of these challenges. (See Appendix F-5, Table 7.) The most important educational need identified—basic and relevant genetic health information—also was reported as the most common topic for educational programs. Education about access to genetic services was the second most frequent response. A common theme in the free-text responses was the importance of genetics education aimed at disease-specific support groups. (See Appendix F-5, 7.1 for free-text responses on this topic.)

Eighty-three percent of respondents ranked a set of five “barriers to genetics and genomics education efforts for seekers of genetic information as it relates to health.” The two highest ranked barriers were lack of health professionals’ understanding of genetics and lack of individual health literacy in genetics. The lower ranked barriers were DTC marketing of genetic tests before there is evidence of utility and lack of access to genetic services for consumers and patients (see Appendix F-5, Table 8). Twenty-nine free-text responses to this item identified additional important barriers including fear of genetic discrimination and loss of job or insurance based on genetic test results; and lack of cultural competency, whether in terms of spoken language or in the complexity of the terminology used to educate consumers on genetics and genomics. (See Appendix F-5, 8.1 for free-text responses on this topic.)

When asked about the government role in genetics education (at all levels), funding of genetics education programs was ranked as the highest priority. Education about anti-discrimination laws was a high priority for all three levels of government, echoing the sentiments from previous survey items that the public has concerns about the potential for discrimination based on genetic information. Another key role for the Federal Government was to serve as a clearinghouse for educational information. This role, however, was ranked among the lowest priorities for State and local governments. A very low priority at all three levels of government was education about the licensing of genetic health care providers. (See Appendix F-5, Table 9.)

Among the 21 free-text responses regarding the role of local government, 13 indicated that there is no role for local government in genetics education. The remaining responses suggested that local governments could educate the public as to where locally available resources could be found and could require genetics education in public schools. (See Appendix F-5, 9.1 for free-text responses on this topic.)

Suggested Priorities for the Department of Health and Human Services

Respondents were asked about the role that HHS should play to improve public genetics education. Nearly 200 responses were received. (See Appendix F-5, 10.) The following major themes emerged:

- HHS should serve as a clearinghouse of quality educational information, materials, and programs (e.g., web-based, radio, television, printed pamphlets). Respondents stated that the need for government to exert some quality control in information materials applies not only to materials for the public, but also to materials provided to clinicians/providers of health care and State/local health agencies.
- HHS should provide funding. While many respondents did not always specify what programs or initiatives they thought should be funded, others suggested that funding was needed for State and local health agencies, as well as funding to help train physicians, nurses, and genetic counselors.
- HHS should play a role in evaluating genetic tests and services, ensuring validity and utility of genetic testing, as well as ensuring that the public has access to appropriate tests and services.

D. SACGHS SURVEY OF SELECTED FEDERAL AGENCIES

Methodologies

In August 2003, a survey was distributed to SACGHS *ex officio* agencies to obtain information about Federal activities related to the education of professionals in genetics. The agencies were asked to provide information on (1) their overall efforts to assess genetics workforce needs and address genetics education and training of professionals and (2) specific activities the agency

funded in this area for the preceding year, including the nature of the activity, its target audience, and funding information.²¹⁷

In 2008, due to the rapid expansion in relevant genomics information over the intervening five years, SACGHS elected to re-survey Federal agencies with *ex officio* status on the Committee as well as the Indian Health Service, Substance Abuse and Mental Health Services Administration, and National Science Foundation. The 2008 federal survey consisted of a mix of closed- and open-ended, narrative-type response questions. In addition to the questions asked in 2003, new themes were explored such as the perceived role of the responding agency in genomics education; the perceived ability of the agency to fulfill this role; partnerships established to facilitate genomics educational activities; and a brief description of past, present, and planned educational activities (see Appendix G-1 for the survey instrument). The federal survey was distributed in late 2008 and early 2009. Nonresponders were contacted by e-mail or by telephone to prompt completion of the survey. (See Appendix G-1, Table 1 for a list of agencies surveyed in 2003 and 2008.)

Data Limitations

The surveys conducted in 2003 and 2008 yielded numerous examples of genetics education activities in Federal agencies. However, there are important limitations that affect interpretation of the data. Although six agencies responded to both surveys, which provided information on programmatic growth and changes in educational priorities, four agencies participated only in the 2008 survey. Precise comparisons between the 2003 and 2008 surveys were difficult as the level of detail varied across responses. For example, some agencies provided information about web-based materials, program funding amounts, relation of activities to agency mission, and specific numbers of individuals trained, and other agencies provided only the name of a program or project without additional details. Funding information for specific activities and programs was provided by several of the respondents; however, incomplete funding data and inconsistencies in defining health professional education limit interpretation of this information.

Survey Findings

Of the 21 agencies surveyed in 2008, 16 provided a response (see Appendix G-1, Table 1). Among the responding agencies, eight reported activities related to genetics education and training, and five of these agencies specifically noted that genetics education and training was a role or responsibility of their agency. In response to a question about how their agency could meet this role or responsibility more effectively, two of the five responders reported needing additional funding and resources. (See Appendix G-2 for further details.) Six Federal agencies provided information about projected priorities for future initiatives in genetics education, and all but one noted that genetics education for medical as well as nonmedical professionals is a top priority. (See Appendix G-3.)

²¹⁷ Secretary's Advisory Committee on Genetics, Health, and Society. Presentation by Sam Shekar, M.D., M.P.H., on Federal Efforts to Address Genetics Workforce, Education, and Training Issues, October 23, 2003. See http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_2003_oct_23.html. Accessed on February 17, 2011.

A brief overview of the agencies' reported genetics and genomics activities is provided below. For agencies that responded to both SACGHS surveys (in 2003 and 2008), information is provided that compares the reported activities. (See Appendix G-4 for additional information provided in the surveys.)

Centers for Disease Control and Prevention (CDC)

CDC's mission is to collaborate with partners across the Nation to create the expertise, information, and tools that people and communities need to protect their health—through health promotion; prevention of disease, injury and disability; and preparedness for new health threats.

In 2003, CDC's activities in genetics education focused primarily on educating the current and future public health workforce, with the goals of improved population health and decreased disease incidence. CDC developed partnerships with national, State, and local public health organizations to assess the need for genomic educational efforts. It brought together public health leaders, health care clinicians, insurers, and other stakeholders to develop programs and educational tools on genetics targeted to the public health workforce and/or the clinical health care workforce. Additional activities were focused on appropriately utilizing genetic and genomic technologies and ensuring high-quality genetic testing. In all, CDC reported 28 activities in its 2003 survey response.

In 2008, CDC reported that as genetics becomes more integral to public health research and practice, the need for genetics expertise in public health has become even greater than previously reported. CDC expanded its target audiences for genetics education activities to include public health administrators, faculty and students in medical school and residency training programs, primary care and specialty physicians, epidemiologists, health educators, laboratorians, and environmental health workers. CDC's role in genetics education and training of professionals is to promote the effective and responsible application of genomics knowledge and tools to promote population health as it applies to chronic disease, environmental health, occupational health, and infectious disease. The agency reported on 16 existing genetics education programs, however, it is not able to assess fully educational needs among professionals and develop and disseminate training tools and curricula in collaboration with partners.

Currently, education and training activities in genetics cut across several CDC divisions and offices. Although the Office of Public Health Genomics initiates many training activities, others have been conducted by the Division of Laboratory Systems; Division of Birth Defects and Developmental Disabilities; the Division of Nutrition, Physical Activity and Obesity; the Division of Partnerships and Strategic Alliances; and the Office of Workforce and Career Development.

CDC's projected priorities for future initiatives in genetics education and training center on empowering providers with the knowledge and skills to apply genetics knowledge and tools to early detection, disease prevention, and health promotion in populations.

Centers for Medicare & Medicaid Services (CMS)

CMS regulates laboratory testing (except research) performed on humans in the United States through the Clinical Laboratory Improvement Amendments (CLIA), enacted by Congress to ensure the accuracy and reliability of all laboratory testing. CLIA established three categories of laboratory tests: waived tests, moderate-complexity tests, and high-complexity tests. Moderate- and high-complexity testing, which includes genetic tests, is subject to regulations that set minimum qualifications for all persons performing or supervising these tests and require laboratories to participate in approved proficiency testing programs, which provide an external evaluation of the accuracy of the laboratory's test results.

The Division of Laboratory Services (DLS), under the Center for Medicaid and State Operations, has the responsibility for implementing the CLIA Program. CMS responded to the 2008 survey (but did not respond in 2003), and DLS is the only division within CMS that has reported activities in genetics education and training. This training is geared to the surveyors overseeing genetic testing and CLIA compliance at laboratories nationwide. (See Appendix G-CMS for additional information.)

Department of Commerce (DOC)

Of the agencies that comprise DOC, only the National Institute of Standards and Technology (NIST) reported ongoing projects in genetics education and training. These projects are in adherence with NIST's mission to develop and promote measurement, standards, and technology to enhance productivity, facilitate trade, and improve the quality of life.

In 2003, NIST's activities in genetics education focused primarily on cancer genetics, forensic applications, and the education and training of practicing professionals. Although the medical and cancer genetics program predominantly served health-related professionals, the forensic applications involved both health-related and nonhealth-related professionals (e.g., lawyers, judges, and law enforcement professionals). Specific needs addressed through these efforts include the development of standards for measurement technologies of genetic information and the education of professionals in the use of these standards.

By 2008, NIST had expanded its activities in genetics education to include students in training and practicing professionals, educational websites and online resources targeted to students and professionals, forensic laboratory site visits as a component of continuing education, assessments of professional knowledge about genetics, and analyses and evaluations of the genetics workforce training and educational efforts. During the period from 2003-2008, NIST built and currently maintains the world's most widely used web-based database on forensic DNA genetic typing (STRBase), held more than 30 training workshops in forensic laboratories and at major scientific conferences to teach genetic principles to scientists and lawyers, and established the NIST Human Identity Project that educates students and professionals about genetics and is funded by the Department of Justice. (See Appendix G-DOC-NIST for details about these projects.)

DOC's projected priorities for future initiatives include the continuation of the NIST Human Identity Project, ongoing workshops and conferences, and continued efforts to evaluate professional knowledge about genetics and assess laboratory performance in forensic analysis.

Department of Defense (DOD)

The DOD medical care system seeks to enhance our Nation's security by providing health support for the full range of military operations and by sustaining the health of all those entrusted to its care. Genetics education and training is integral to the functioning of the military medical care system, and DOD has focused significant efforts to educate staff in genetics and bioethics.

In 2003, learning needs in the evolving fields of genetics and genetic technologies were identified in order to ensure the integration of new services and technologies throughout the medical treatment facilities. These plans included staff education, policy developments such as operating instructions and guidelines, evidence-based practices, and competency-based evaluation.

The Uniformed Services University of the Health Sciences has taken a leadership role in incorporating genetics content into the curricula of both the School of Medicine and the Graduate School of Nursing. Continuing education programs for clinical specialties such as pediatrics, oncology, and obstetrics and gynecology also include genetics content. The impact and effectiveness of these programs and curricula are being evaluated.

By 2008, DOD articulated a dual health care mission—readiness and benefits. The readiness mission is supported through provision of medical services to the Armed Forces during military operations and the benefits mission through health care to more than 9 million eligible beneficiaries worldwide. DOD continues to recognize the need for professional education and training in genetics for the readiness mission and to provide excellent health care to its beneficiaries.

DOD's current capabilities in genetics include a genetics workforce, laboratory facilities, and educational programs, which are described in more detail in Appendix G-DOD.

Department of Energy (DOE)

DOE's commitment to education in genetics is consistent with its view of science and support of interdisciplinary research. DOE's Office of Science provides ongoing support for research in molecular genetics, genome sequencing and microbiology, and in emerging disciplines such as bioinformatics and structural biology.

In 2003, the DOE survey response focused on the new capabilities emerging in genetics and the mapping of the human genome and emphasized the essential need for genetics education in order to make the best use of these capabilities. DOE listed 26 primarily educational activities that targeted a variety of audiences, including underserved populations, the judiciary, and academia.

By 2008, DOE had established two training programs for professionals at the DOE Joint Genome Institute (JGI). One of the JGI programs provides a system for incorporating genetics research into undergraduate courses. The second program is a joint effort of the American Society of Microbiology and DOE-JGI that introduces basic bioinformatics to undergraduate faculty.

DOE has numerous educational websites related to genetics, which are aimed at practicing professionals, K-12 teachers and students, and graduate students. These and other educational resources can be found at the JGI website.²¹⁸

DOE has been evaluating the impact of its education programs in collaboration with the Oak Ridge Institute for Science and Education (ORISE). Surveys conducted and analyzed by ORISE indicate that JGI programs are addressing an unmet need for research opportunities for undergraduates and faculty development, and allow faculty and students to contribute new knowledge to DOE science. DOE has plans to expand its programs to include building similar tools for metagenome and eukaryotic genome analyses so that students and faculty can participate in the full range of DOE mission-related genomics research. (See Appendix G-DOE for details of additional projects.)

Equal Opportunity Employment Commission (EEOC)

EEOC is responsible for enforcing Federal laws that make it illegal for employers to discriminate against a job applicant or an employee because of the person's race, color, national origin, sex, age, religion, or disability. With the passage of the Genetic Information Nondiscrimination Act (GINA) in 2008, discrimination protections now include discrimination against individuals because of genetic information.

EEOC provided a response to the 2008 survey but not in 2003. EEOC genetics education and online resources include detailed information about Title II of GINA, and training sessions on the legal prohibitions against employment discrimination on the basis of genetic information. (See Appendix G-EEOC for a listing of these trainings.) Once the regulations implementing Title II of GINA become final, EEOC plans to conduct additional training sessions on the legal requirements of Title II for lawyers, human resource professionals, small business owners, and other interested parties.

Federal Trade Commission (FTC)

FTC is the only Federal agency with jurisdiction over consumer protection. Among its many activities, FTC advances consumers' interests and creates practical and plain-language educational programs for consumers and businesses in a global marketplace with constantly changing technologies.

FTC responded to the 2008 survey but not in 2003. As part of its mission to regulate unfair and deceptive practices, FTC cooperated with FDA and CDC in 2006 to develop a fact sheet for consumers to educate them about the limitations of DTC genetic tests. The fact sheet, *At-Home*

²¹⁸ Department of Energy Joint Genome Institute. See <http://www.jgi.doe.gov/education/>. Accessed on December 6, 2010.

Genetic Tests: A Healthy Dose of Skepticism May be the Best Prescription,²¹⁹ provides consumers with clear information to make well-informed decisions when considering whether to purchase DTC genetic tests and answers questions about the usefulness of such tests. FTC will continue to evaluate the need for consumer education about DTC genetic tests and will also monitor consumer-directed advertising of genetic tests and take action, where necessary, to prevent consumer deception. (See Appendix G-FTC for additional information.)

Health Resources and Services Administration (HRSA)

HRSA's mission is to improve and expand access to quality health care for all through the adequate provision of primary care services. To comply with this core mission, HRSA supports ongoing genetics education and training activities for health care professionals with the goal of decreasing health disparities by improving access to quality health care.

In 2003, HRSA reported 64 genetics educational activities. Several of HRSA activities have been co-funded with other HHS agencies including NIH, CDC, and AHRQ. HRSA and NIH activities primarily are geared to addressing issues related to the education and training of practicing health care professionals, graduate students, residents, and fellows. For example, HRSA has awarded ongoing funding for Area Health Education Centers to provide community-based CE programs to health professionals that include a component with genetics content.

The criteria that HRSA used to determine which genetics training and education activities to undertake included a focus on emerging areas of public health significance, such as genetics and bioterrorism; an interdisciplinary focus on the translation of genetic knowledge into practice and research; the applicability of genetics across disciplines; and the need to educate the public about genetic services and genetic testing.

In 2008, the HRSA survey response noted an expanded number of activities in genetics education and training and listed several divisions within HRSA that have a role or responsibility for such programs. These programs aim to educate professionals or trainees about genetics and include programs in the Maternal and Child Health Bureau and the Bureau of Health Professions. (See Appendix G-HRSA for details of these programs.)

From 2003 to 2009, HRSA developed targeted educational products that include web-based materials, newsletters, workshops, and printed materials about genetics to be used specifically by primary care providers, State newborn screening programs, the general public, dietitians, physician assistants, nurses, patients, speech pathologists, and dentists. HRSA has also developed products for all audiences on family history and core competencies in genetics, genetics and common diseases, and genetics, race, and health care.

HRSA participated in several projects between 2003 to 2006 evaluating and assessing professional knowledge about genetics and analyzing the genetics workforce.^{109,111} HRSA has also conducted a more recent genetic workforce analysis, *Assessing Genetic Services and the*

²¹⁹ Federal Trade Commission. *Facts for Consumers, At-Home Genetic Tests: A Healthy Dose of Skepticism May Be the Best Prescription*, July 2006. See www.ftc.gov/bcp/edu/pubs/consumer/health/hea02.shtm. Accessed on November 19, 2009.

Health Workforce,²²⁰ to aid in identifying and planning for supply and demand needs for 2010 and beyond. This analysis enhanced understanding of clinical genetics services, factors affecting demand for genetic services, and the roles of health professionals providing these services. Additional activities reported in 2008 include providing reviews of journal articles related to genetics and genomics and participating in advisory and editorial boards (see Appendix G-HRSA).

Through the 2008 Newborn Screening Saves Lives Act,¹⁸⁹ HRSA was charged, in consultation with NIH and CDC, to establish and maintain a central clearinghouse of educational information, family support and services information, resources, research, and data on newborn screening. The Act authorized funding, and the project is being developed by the Genetic Alliance in partnership with the National Newborn Screening and Genetics Research Center, Genetics and Newborn Screening Regional Collaborative Groups, the March of Dimes, and the Association of Public Health Laboratories.¹⁹⁰

National Institutes of Health (NIH)

NIH is the steward of medical and behavioral research for the Nation. Its mission is to pursue fundamental knowledge and apply that knowledge to improve health and reduce illness and disability through funding basic research and training for scientists. Training health professionals in the area of genetics is essential to ensure that research findings in the rapidly expanding field of genetics are translated into health practice. NIH training activities in genetics focus on improving basic and clinical genetics research to benefit the general public and improve health. Some of the institutes and centers at NIH also provide training in the area of clinical genetics and are described below.

NIH reported in its 2003 survey response that it had funded a number of different workshops and had developed educational tools geared to helping clinicians learn more about the impact of genetics on their practice. To support genetics training of health professionals and to address the translational aspects of genomics, NIH, along with the American Medical Association and American Nurses Association, helped form the NCHPEG.²²¹ Several NIH and HRSA collaborative genetics workforce assessment activities and research are described in Appendix G-NIH.

By 2008, NIH's genetics training and educational activities included trans-NIH programs administered by the Office of Strategic Coordination. Individual institutes at NIH also have developed genetics training and education programs. (See Appendix G-NIH for additional details.)

²²⁰ University of Maryland School of Medicine. *Assessing Genetic Services and the Health Workforce*. See http://medschool.umaryland.edu/ghsrc/research_assessing.asp. Accessed on November 30, 2009.

²²¹ National Coalition for Health Professional Education in Genetics. See www.nchpeg.org. Accessed on November 19, 2009.

Programs of Individual NIH Institutes, Centers, and Offices

National Cancer Institute (NCI)

In its 2008 survey response, NCI reported three programs aimed at educating professionals and trainees about genetics.²²² The Genetics Related Market Research was conducted in conjunction with the Trans-NIH Communications Group on Genetics and Common Diseases to help understand public perceptions about genetic testing and the rapidly growing area of DTC genetic testing. NCI also has developed a wide range of web-based resources focused on genetics specifically designed for health professionals. These tools can be accessed from the Cancer Genetics website and include cancer risk assessments and a link to the HHS Family History page.²²³ (See Appendix G-NCI for additional details.)

National Human Genome Research Institute (NHGRI)

NHGRI listed several activities in its 2008 survey related to genetics education and training. These activities include the development of educational resources to promote nursing and physician assistant education; the Genomic Health Care Commons, a web-based interactive education resource to support groups engaged in trans-disciplinary resource development within the nursing and physician assistant communities; and the organization of meetings. (See Appendix G-NHGRI for details about these programs and meetings.)

National Institute on Aging (NIA)

Education and training of biomedical researchers and dissemination of scientific information to diverse audiences, including health professionals and the general public, is a priority for NIA. (See Appendix G-NIA for additional information.)

National Institute on Deafness and Other Communication Disorders (NIDCD)

NIDCD, in conjunction with NHGRI, co-funded a Summer Program in Genetics for Audiology Faculty in 2006. This program was designed to improve training of future audiologists in the clinical, technical, ethical, social, and legal issues surrounding the provision of genetic services and molecular testing for hereditary types of hearing loss. This program also included a comprehensive evaluation component to determine its effectiveness. The results of the program were used as a model for development of a continuing education online course on genetics and hearing loss that is currently taught through Gallaudet University. (See Appendix G-NIDCD for details about this program.)

²²² R25CA066061: “Advanced Cancer Risk Counseling Training for Nurses,” R25CA075131: “Clinical Cancer Genetics Education,” and R25CA093426: “Genetics Short Course for Cancer Nurses.”

²²³ National Cancer Institute. *Cancer Genetics*. See www.cancer.gov/cancertopics/prevention-genetics-causes/genetics. Accessed on November 19, 2009.

National Institute of Dental and Craniofacial Research (NIDCR)

NIDCR has been conducting assessments of professional knowledge about genetics or genomics since 2001. The assessments have examined core competencies in genetics and the status of genetics education in U.S. dental schools and included focus group research with dental professionals that also evaluated the genetics workforce in dentistry. NIDCR reported a number of conferences, presentations, workforce assessments, and publications relating to genetics and dentistry. NIDCR also has developed online resources and educational websites, and provides outreach and education in dental genetics to the dental practice and dental education communities on an ongoing basis. (See Appendix G-NIDCR for details about these programs.)

National Institute on Drug Abuse (NIDA)

NIDA activities include a *Research Education Grant for Statistical Training in the Genetics of Addiction* and support for a number of meetings aimed to educate professionals and trainees about genetics. Genetics research has improved the understanding of biological processes and the mechanisms underlying addiction. However, the sudden expansion of information has created a critical need for interdisciplinary research education in statistical genetics and computational methods. NIDA's Research Education Grant was intended to address this need by training pre- and post-doctoral students in the genetics of substance use and abuse, and by encouraging development of new, useful, and innovative statistical methods to analyze the increasing body of genetic data. The final phase of the project involves disseminating the course materials through workshops, webcasts, and web pods and developing software user guides to the wider community of substance abuse researchers. (See Appendix G-NIDA for additional information.)

National Library of Medicine (NLM)

NLM supports three genetics training and education programs at the National Center for Biotechnology Information (NCBI); the Lister Hill National Center for Biomedical Communications (LHNCBC); and through an NLM extramural, university-based program. (See Appendix G-NLM for additional information.)

Office of the Director (OD)

The OD Office of Strategic Coordination—within the Division of Program Coordination, Planning, and Strategic Initiatives—provided information about genetics education and training programs and activities supported through the Common Fund. The NIH Reform Act of 2006 established the Common Fund to support trans-NIH programs that are cross-cutting and are expected to have exceptionally high impact on the scientific community. Several Common Fund programs support genetics education and training, such as the National Centers for Biomedical Computing (NCBCs) and the Interdisciplinary Research (IR) program. Additionally, the NIH Clinical Center offers courses that include genetics and genomics content. See Appendix G-OD for additional information.)

National Science Foundation (NSF)

NSF is an independent Federal agency created by Congress in 1950 to promote the progress of science; to advance national health, prosperity, and welfare; and to secure national defense. The agency is tasked with keeping the United States at the leading edge of scientific discovery. The agency's mission is to ensure that the research it supports is fully integrated with education so that today's revolutionary work will also be training tomorrow's top scientists and engineers.

In 2008, the agency reported that it administers approximately 50 active awards that directly or indirectly promote genetics education for K-12, undergraduate, or graduate students, or for the general public. Although NSF has no programs that specifically target genetics education, it recognizes that genetics is vital to an understanding of general biology, as well as workforce issues such as biotechnology training. Current awards thus include projects that indirectly address genetics while targeting a broad range of topics in biology such as molecular/cellular biology, evolution, biodiversity, and ecology.

Many of NSF's education awards have a core objective relating to genetics. Examples of projects funded by NSF in 2008 and beyond include *Literature-Based Scientific Learning in Genetics*, *The Community College Genomics Research Initiative*, *Proteomics and Functional Genomics Scholarship Program*, and *Pre-doctoral Training in Functional Genomics of Model Organisms*. (See Appendix G-NSF for a detailed listing of programs, programmatic goals and individual NSF funded projects with core objectives relating to genetics and genomics).

IV. Discussion

Findings from SACGHS' information-gathering activities supported what has been known for a number of years regarding the needs and gaps in genetic and genomic education and training of health professionals and the public. Improving genetics education for these groups will require a comprehensive and coordinated effort. Genomics will challenge the traditional model of genetic services, in which the use and communication of genetic information occurs in the clinical setting, offered by health care professionals during “teachable moments,” following a diagnosis.²²⁴ This traditional model is not well equipped to serve the much larger segment of the population with moderately increased risk for various multifactorial diseases with genetic components (e.g., cancer, cardiovascular disease, and diabetes). In some of these cases, individuals may be presymptomatic; thus, understanding future probabilities and opportunities for prevention or early detection will require a well trained health care workforce as well as informed consumers and patients. Effective interventions based on genetic information will require consumer understanding of the meaning and interactions of susceptibility genes of uncertain penetrance with other risk factors.²²⁵ In addition, with the expansion of screening and early-detection technologies for many common chronic diseases, the public health workforce, with its population-based focus, will become increasingly integral to both community education and service provision.

Thus, a new model for applying genetics to improve health requires a system in which health care professionals, public health providers, and consumers are well informed and able to interact with each other as appropriate. Cooperation and collaboration in processing, applying, and interpreting genetic information will be essential. Without these efforts, society will not benefit from genetic advances, opportunities will be lost for deploying prevention and early detection programs for a wide variety of chronic diseases, and patients and consumers may make poorly informed choices or fail to seek needed professional health services. Incorporating genomics into health care will be expedited by building on the significant amount of work that has already been done by Federal and State Governments and private sector organizations such as health professional societies and patient advocacy groups. Ultimately, however, innovative strategies that maximize stakeholder participation and consider the needs of health professionals and consumers alike will be required.

A. SYNTHESIS OF FINDINGS FROM LITERATURE AND SURVEYS

Health Care Professionals

Based on a literature review and its survey findings, SACGHS found evidence that suggests inadequate education of health care professionals is a significant factor limiting the appropriate integration of genetics into clinical care. While much work has been done to develop genetics educational curricula and programs at the undergraduate, graduate, and continuing education

²²⁴ Lawson, P.J., and Flocke, S.A. (2009). Teachable moments for health behavior change: a concept analysis. *Patient Education and Counseling*. 76:25-30.

²²⁵ Wilde, A., Meiser, B., Mitchell, P.B., and Schofield, P.R. (2010). Public interest in predictive genetic testing, including direct-to-consumer testing, for susceptibility to major depression: preliminary findings. *European Journal of Human Genetics*. 18(1):47-51.

level, SACGHS also found that these efforts often exist in isolation; do not take into account multidisciplinary approaches to care; and are not always linked to accreditation, certification, and licensure programs that can be critical drivers of education content.

There are many factors affecting the timely incorporation of genetics into patient care. These include the failure to update education curricula to reflect scientific advancements in genetics and genomics, limited application of genetic concepts in clinical training, competing priorities across the continuum of education, lack of funding to support genetics education programs, and lack of evidence supporting clinical effectiveness of genetic testing. Compounding these findings is an insufficient number of M.D. and Ph.D. geneticists available to provide genetic services in clinical care and genetics education to health care professionals.

Many health care professionals lack genetic knowledge about complex, multifactorial conditions as well as traditional and well-documented Mendelian conditions. Analyses of genetic content in formal medical and health care curricula (with the exception of genetic counseling programs) find wide variability in the content and quantity of coursework in genetics. The same variability and levels of insufficiency can be found in licensing and accreditation requirements.

The 2008 SACGHS survey of health professional organizations provided data that support findings from the literature review and revealed insights into how professional organizations are currently approaching the need to educate their members and constituencies in genetics and genomics. Overall, 70 percent of health professional organizations responding to the survey viewed genetics education and training as part of their role or responsibility and reported that developing and promoting genetics educational activities is important. However, these activities are not a high priority relative to the overall priorities facing the organizations. The most commonly mentioned barrier to improving genetic literacy of health care professionals was competing priorities in already crowded curricula.

Most of the organizations reported that they were able to fulfill their role or responsibility to educate their membership. However, despite this interest in genetics education, less than half reported that their organizations seek input from their membership regarding educational needs and priorities, have dedicated staff specifically focused on genetic topics, or have published position statements or practice competencies. Organizations cited lack of sufficient resources, financial and otherwise, as a barrier to developing or accessing appropriate education and training opportunities for members. Respondents cited funding, program evaluation, and increasing interest within the organization's leadership as factors that would help them meet their genetics education role or responsibility more effectively. Additionally, federal support of research and dissemination of evidence-based guidelines would help engage their members' interest in additional genetic topics.

Thus, the need for educational efforts to increase the use of genetic information in clinical care is widely recognized and acknowledged. The SACGHS survey highlighted the challenges facing many organizations attempting to meet this need against a backdrop of competing demands and limited resources.

Public Health Providers

The literature review provided evidence that the current public health workforce is not well prepared to receive and assimilate genetic and genomic information. It also revealed that the barriers to achieving a more genomics-informed public health workforce are multifaceted. First, the public health workforce is diverse and follows many educational and training paths, including a variety of professionals with formal training and certifications, volunteers, and community (lay) health workers. Thus, a one-size-fits-all approach to education and training is not practical. Second, many professionals in the field today received their formal education before genomics became a component of population-based screening or early detection for common conditions. Third, attitudes, perceptions, and beliefs shape the acceptance and adoption of genetics and genomics by the public health community. Khoury et al.¹⁴ and Chen^{139,140,141} noted that one of the attitudinal barriers to acceptance of genetics and genomics by the public health community is skepticism about genomics and genomics research, which is considered as a low-yield investment and low priority because of other more important preventive or modifiable environmental causes of disease. Other barriers include competing priorities; many public health providers consider local issues, national and international pandemics, and environmental causes of morbidity and mortality as more important priorities than genetics and genomics, particularly in the context of limited public health funding.

The literature also revealed that public health providers do not perceive public health genomics to be part of their job, nor a professional priority.¹⁴ Until the evidence of public health benefits of genetic testing can be demonstrated, public health providers might be resistant to embracing genetics given the other demands of public health practice.

The SACGHS survey of public health providers identified genetic competencies that are considered important and are frequently and confidently applied. The most important and most frequently applied competency was demonstrating a basic knowledge of the role that genetics plays in the development of disease. However, respondents indicated they were most confident in describing the importance of family history, which may lead to greater uptake among public health providers of this key element for identifying predisposition to genetic conditions. Conducting outcomes evaluation of available genetic services ranked the lowest in importance, in frequency of application, and in confidence in demonstrating this competency. Only one-third of respondents indicated that resources for implementing genetic and genomic competencies were adequate or very adequate.

The survey provided important information on the delivery of genetic services to underserved or vulnerable populations. Respondents described organizational efforts to create culturally and linguistically appropriate educational materials, conduct community-based participatory research, train entities within local communities to foster outreach, provide genetic counseling either in person or via teleconference calls, and conduct research to understand barriers to community access to genetic services. Strategies and recommendations were also identified to target vulnerable or underserved populations. These included the need for increased funding to enhance genetic services and outreach, and, to foster mechanisms for increased community input from vulnerable or underserved communities; development of websites as part of outreach tools;

and the need for policies to enhance genetic services, raise awareness, and increase education of local community members.

Many of the concerns and barriers highlighted in the SACGHS survey of public health providers were the same as those reported in the literature. Overall, survey respondents had a positive attitude toward genetics, which may be attributed to the nature of the survey dissemination that targeted individuals more likely to incorporate genetics into their daily practice.

Consumers and Patients

There is an underlying need for improved genetic literacy beginning in the formative years and continuing throughout the lifespan, as evidenced by the review of current literature, findings from a SACGHS survey, and interviews exploring consumer attitudes and beliefs about genetics and gaps in genetics knowledge. SACGHS' data-gathering activities found that consumers understand that there is a relationship between genetics and health outcomes, but they generally do not understand complex traits and the contribution of genetics to common diseases, nor do they understand how to use genetic information to optimize health. Levels of genetic knowledge also have been found to differ by race, ethnicity, and socioeconomic background.

In general, the literature review found that although consumers have a limited understanding of genetic testing, they have been supportive of it when used for improving disease diagnosis and prevention. Despite the availability of DTC testing, consumers would prefer to learn about genetic tests from their health care providers. This desire on the part of consumers underscores the deficiencies of most primary care providers in their general genetic knowledge and their specific lack of comfort in selecting, ordering, and interpreting genetic tests and in providing appropriate genetic counseling. There are indications that the Internet and other forms of media have become a substantial source for consumer and patient knowledge regarding genetics.

A majority of the SACGHS consumer survey respondents agreed about the need for basic and relevant genetic health information. This information was defined as knowledge of specific terminology such as “probabilities” and concepts such as “variability” and “common conditions” as opposed to “rare variants.” For consumers to understand genetic testing, they must appreciate the distinction between the risk for a disease and its diagnosis. They also agreed that genetics education should focus on multifactorial disorders, the value and limitations of genetic testing and DTC genetic services, and personalized guidance about genetic tests. Even though much of the data that inform this report were collected shortly after passage of GINA, concerns persist about confidentiality and disclosure of genetic information that might lead to loss of a job or insurance. The fear of DNA being collected without consent was also expressed by survey respondents.

Federal Agency Activity

Findings from the SACGHS surveys of selected Federal agencies conducted in 2003 and 2008 suggest that Federal genetics educational programs and resources for professionals and consumers increased over the five-year period. Of particular note were the increases in resources for consumers and nongeneticist professionals.

CDC has been very active in genetics education efforts and reported funding a number of activities since 2003.

DOD reported activities related to health care professional education through its medical training entities in 2003; however, by 2008, educational activities had broadened significantly with personalized medicine programs and a DOD-wide newborn screening program that includes education of health care professionals and parents.

In 2003, DOE had already been heavily involved in genetic and genomic education activities as a result of its participation in the Human Genome Project. By 2008, DOE had established the Joint Genome Institute (JGI) to increase the incorporation of genomic research at all educational levels and to develop websites aimed at practicing professionals, K-12 teachers and students, and graduate students.

HRSA reported 64 educational activities in 2003, but they were primarily targeted to practicing health care professionals, graduate students, residents, and fellows. HRSA has expanded its focus over the intervening years to include the general public and a wider range of health care professionals. It also developed products for all audiences on family history, newborn screening, and the genetics of common diseases.

NIH reported 41 genetics education and training activities in 2003, including funding to support NCHPEG. By 2008, NIH had numerous activities within individual Institutes and through some of its trans-NIH programs.

In 2003, CMS reported having no activities relevant to the SACGHS survey. In response to the 2008 survey, CMS reported that it conducts activities in genetics education and training for surveyors who conduct laboratory inspections under the Clinical Laboratory Improvement Amendments.

FTC has been working with FDA and CDC on consumer education for DTC genetic testing. FTC will continue to evaluate the need for consumer education about DTC genetic tests and will also monitor consumer-directed advertising of genetic tests and take action, where necessary, to prevent consumer deception.

EEOC is responsible for Title II of GINA and provides education and online resources on prohibitions against employment discrimination on the basis of genetic information. EEOC provides training on the legal prohibitions against employment discrimination on the basis of genetic information.

NSF awards grants to promote genetics education for K-12, undergraduate and graduate students, and the general public. In response to the 2008 SACGHS survey, the agency reported that it administered approximately 50 active awards that directly or indirectly promote genetics or genomics education.

B. THE GROWING NEED FOR EDUCATION AND TRAINING ACROSS MULTIPLE DISCIPLINES

The issue of how best to translate, interpret, and deliver complex genetic information to health care professionals and consumers has been examined for several decades. The discipline of clinical genetics arose in the 1950s,²²⁶ and soon thereafter came the recognition that nongenetics professionals also play a role in providing genetic services to patients.⁴² In a 1975 report on the emerging field of genetic screening, the National Academy of Sciences anticipated the movement of genetics from the specialized clinic toward point of care and signaled an early concern about the need for an educated workforce in the application of genetics.²²⁷

As a result of these early efforts, primary care became the center of much of the focus on professional education needs in genetics. Thirty years ago, Hsia—contemplating the transition of genetics into primary care—raised the following questions that remain relevant today: “How much genetic knowledge should primary physicians have? Should they be able to diagnose, treat, and counsel about all genetic diseases? Will it suffice for them to check the literature or consult a geneticist whenever a genetic problem arises? Optimal knowledge must lie between these extremes, because a primary physician must have enough knowledge to recognize a problem as genetic and should have enough familiarity with genetic principles to be able to use the literature wisely, or to consult with a geneticist intelligently.”²²⁸

The provision of genetic services by nongenetics professionals is not without challenges. Greendale et al.²²⁶ suggested potential problems with empowering primary care providers to assume prominent roles in genetic service delivery, citing their lack of knowledge and disinterest in the field, while Guttmacher et al.²²⁹ argued that implementation of “genomic health care” would necessitate collaboration and cooperation of all health professionals.

Increasingly these same concerns are occurring in the public health arena, as genomics moves into population-based applications. The public health perspective will be crucial not only in application of genetic and genomic knowledge but also in assessing its validity and utility. Because the clinical validity of genetic information is highly dependent on population characteristics (i.e., prevalence of the genetic variant, strength of its association with disease, interactions with other risk factors), the skills of the public health workforce, as well as tools and resources, will be increasingly important.

In 2005, a federally funded study concluded that the medical genetics workforce was not sufficient to meet expected patient care needs for clinical genetic services in the next five to 15 years due to several factors including the increased need for genetic services and data showing that young physicians are not entering the field of genetics.¹⁰⁹ These workforce deficiencies have

²²⁶ Greendale, K. and Pyeritz, R.E. (2001). Empowering primary care health professionals in medical genetics: How soon? How fast? How far? *American Journal of Medical Genetics*. 106:223-232.

²²⁷ National Academy of Sciences and National Research Council. *Genetic Screening: Programs, Principles and Research*. Washington: National Academy of Sciences, 1975.

²²⁸ Hsia, Y.E., Bucholz, K.E., and Austein, C.A. Genetic knowledge of pediatricians and obstetricians (Connecticut, 1975, 1977): Implications for continuing education. In: Porter, I.H., and Hook, E.B. eds. *Service and Education in Medical Genetics*. Academic Press, 1979. P. 378.

²²⁹ Guttmacher, A.E., Jenkins, J., and Uhlmann, W.R. (2001). Genomic medicine: who will practice it? A call to open arms. *American Journal of Medical Genetics*. 106(3):216-222.

not been addressed, and in fact, are being exacerbated as new genetic technologies become available and demand for genetic services increases.

Legislative proposals have recognized needs in this area, calling for increased funding of programs to develop and disseminate model training programs, ensure adequate focus on genetics in certification and accreditation programs, enhance continuing education programs, and promote competencies across clinical, public health, and laboratory disciplines.²³⁰ However, no bills have been passed that actually provide funding for such programs.

C. ROLE OF THE FEDERAL GOVERNMENT IN GENETICS EDUCATION

Organizations responding to the SACGHS survey cited lack of resources as a barrier to advancing genetics education. They suggested that Federal funding, if available, could support educational grants for faculty training, program development and program evaluation, development of point-of-care tools and tool kits, research and dissemination of evidence-based guidelines, increased integration of genetics into clinical decision support, integration of genetics information with electronic medical records, and development of performance standards. In addition, establishing a registry of genetic tests would facilitate the evaluation of clinical validity and utility and thus inform genetic test usage in the clinical setting.

Experts interviewed about consumer needs suggested that consumers believe that the Federal Government is a more unbiased source of information than commercial sources and that it should have a central role in public genetics education and literacy efforts. Consumers also think that government should monitor the societal effects of genetic and genomic testing and services, clarify the extent to which laboratory tests are regulated, determine who is qualified to provide genetic services, support formal genetics education in schools, and exert some influence over educational standards.

Federal and State Governments are viewed as having important roles in educating consumers and health care providers alike. The Federal Government is seen as the logical repository for educational information, and many believe it should serve as a clearinghouse for this information.

The complexity and rapid evolution of knowledge and technology related to genetics and the varying learning needs of communities and individual consumers will require that educational efforts and resources directed to consumers be appropriately translated and tailored to specific segments of the population. Federal funding and additional program development may be necessary to address disparities in access to consumer educational resources and to provide educational materials that are appropriately targeted and effectively delivered to various segments of the population.

Experts who were interviewed about consumer needs considered education about antidiscrimination laws to be a high priority. Given the public's concern about genetic

²³⁰ For example, the Genomics and Personalized Medicine Act of 2007 (S.976). See http://olpa.od.nih.gov/tracking/110/senate_bills/session1/s-976.asp. Accessed on February 25, 2010.

discrimination and lack of awareness of current protections, interviewees thought that efforts were needed at all levels of government.

Experts interviewed also provided a number of suggestions for the specific role the HHS should play to improve public genetics education and several major themes emerged. These roles included serving as a clearinghouse of quality educational information, materials, and programs geared to the public, health care providers, and State and local health agencies; providing funding for State and local health agencies, as well as funding to train physicians, nurses, and genetic counselors; and evaluating genetic tests and services to ensure the validity and utility of tests and facilitate broad public access to appropriate tests and services.

V. Conclusions and Recommendations

SACGHS examined the genetics education and training needs of health care professionals, public health providers, and consumers and patients through surveys, environmental scans, and literature reviews. Since its last consideration of this area in 2004, SACGHS found that genetics education and training efforts in the private and public sectors have increased, and a number of strategies to incorporate genetics into health care have been adopted. However, these efforts have not kept pace with the emerging understanding of the human genome and rapid evolution of genetic technologies.

For point-of-care health care professionals and public health providers across all levels of training, the literature speaks to course materials that have not kept up with the rapid advancements in genetics and/or have not been effectively incorporated in curricula. Additionally, genetics education and training efforts are not sufficiently linked to requirements for accreditation, certification, and licensure. Educational approaches based on genetic competencies targeted to the training needs of the multiple professional roles within clinical practice and public health will be required for the workforce to address health needs effectively. Without additional educational efforts, the gap in knowledge will only widen and continue to challenge the integration of genetics across the health care and public health landscapes.

Another critical step in promoting increased knowledge of genetics among health care professionals is ensuring adequate reimbursement for genetic services; particularly, given the reality that health care professionals—and the professional societies representing them—are unlikely to invest significant resources in genetics education and training to support services that are not reimbursable or only partially reimbursable. These services include reimbursement for the time that health care professionals spend in direct patient care delivering genetic services, such as providing counseling, conducting informed consent, interpreting genetic tests, and collecting family history. Such reimbursements would be applicable to all members of interdisciplinary teams providing genetic services, for distance consultations, and for telemedicine services that are used in underserved regions. SACGHS made recommendations to the Secretary regarding the need for reimbursement in its 2006 report *Coverage and Reimbursement of Genetic Tests and Services*.⁹⁷ The Committee urges the Secretary to act on these previous recommendations as a key element for promoting health care and public health provider interest and proficiency in genetics.

Another concern—highlighted in numerous workforce analyses—is the shortage of health care professionals and public health providers trained in genetics. In addition to M.D. and Ph.D. geneticists, others trained in genetics—such as genetic counselors, pharmacists with pharmacogenomic training, and nurse geneticists—should be encouraged to step into educator roles. Genetics education programs that use trained peer educators have been successful and well accepted. Also, enhancing the use of clinical decision support tools will provide just-in-time education and support the optimal use of genetics and genomics in health care.

Patients and consumers face many challenges in seeking, understanding, and using genetic information for health care decisionmaking. Studies point to a lack of educational materials that

are culturally appropriate and tailored to the specific needs of communities and that have been validated using certified health educational standards to ensure comprehension by the target audience. Given the wide range of educational levels and motivations among individuals seeking genetic information, a variety of strategies are needed to enhance learning. These strategies include expansion of Internet resources, toll-free hot lines, printed materials, and community-specific radio and television programs that may be more accessible to individuals with lower literacy or who are nonEnglish speaking. Efforts to improve the quality and accessibility of web-based resources will be important to provide information in a manner preferred by consumers.

SACGHS surveys of selected Federal agencies conducted in 2003 and 2008 highlighted many programs and a number of government-sponsored websites that support educational efforts in genetics. An effort to publicize these resources and maintain a centralized entry point for their access would facilitate dissemination of accessible, credible genetic information to health professionals and the public.

SACGHS presents six recommendations that address the identified genetics education and training needs of health care professionals, public health providers, and consumers and patients.

Recommendation 1

Evidence from the United States and abroad suggests inadequate genetics education of health care professionals as a significant factor limiting the integration of genetics into clinical care. Specific inadequacies include the amount and type of genetics content included in undergraduate professional school curricula and the small amount of genetics-related knowledge and skills of physicians, nurses, and other health professionals once they enter clinical practice. Modifications in medical, dental, nursing, public health, and pharmacy school curricula and in medical residency training programs are needed to ensure that health care professionals entering the workforce are well-trained in genetics.

1. Innovative approaches that coordinate the efforts of entities involved in health professional education and training are required to address these gaps. Therefore, HHS should convene a task force of stakeholders to identify:
 - A. Outcomes-based education and training guidelines and models;
 - B. Best practices for enhancing and expanding the content needed to prepare health care professionals for personalized genomic health care;
 - C. Mechanisms to assure the incorporation of up-to-date genetic content in standards, certification, accreditation, electronic health records, and continuing education activities; and
 - D. Funding sources for developing and promoting genetics education for relevant health care professionals.

Recommendation 2

The inherent diversity of the public health workforce makes it difficult to target educational efforts that are relevant across groups. A systematic effort is needed to evaluate the composition

of the public health workforce with current job responsibilities related to genetics and genomics and to identify future priorities, such as the potential impact of affordable genomic analysis.

2. HHS and its public health agencies should:

- A. Assess the public health workforce to determine the number and type of public health providers with responsibilities in genetics and genomics and to ascertain current trends and future education and training needs;
- B. Identify and engage exemplary public health genomic programs to identify critical workforce information not captured in the assessment; and
- C. Using the results of these assessments and to address identified gaps, HHS should:
 - Support development of skills, competencies, and leadership in genetics and genomics that specifically address the identified needs; and
 - Based on these skills and competencies, fund the development and implementation of accessible educational programs and continuing education in genetics and genomics for the public health workforce.

Recommendation 3

Findings in the literature and SACGHS surveys indicate that health care professionals and public health providers serving underserved and underrepresented groups and populations face significant challenges.

3. To increase services and access to care in underserved communities, HHS should:

- A. Identify existing effective educational models for health care professionals and public health providers in underserved communities;
- B. Identify and support programs to increase the diversity and genetic competencies of the health care workforce serving underserved communities; and
- C. Incentivize organizations and ensure that consumers and representatives of rural, minority, and underserved communities participate in the process of developing education and training models and materials. Assure that these materials are culturally and linguistically appropriate and tailored to the unique needs of these diverse communities.

Recommendation 4

With the vast increase in scientific knowledge stemming from genetics research, the development of new technologies, and the increase in direct-to-consumer genetic services, educational efforts are needed to translate this information to reach consumers of all literacy levels.

4. HHS should identify effective communication strategies for translating genetics knowledge into information that consumers and patients can use to make health decisions. Specifically, HHS should:

- A. Support multidisciplinary research that identifies effective methods of patient and consumer communication;
- B. Based on this research, and to reach diverse people and communities, HHS should develop educational programs that use a wide array of media and community-based learning and provide culturally and linguistically appropriate materials; and
- C. In collaboration with the Department of Education and the National Science Foundation, support the incorporation of genetics and genomics in K-12 education.

Recommendation 5

A significant amount of genetic-related information directed to consumers and patients exists in a variety of formats and from a number of sources, but the quality of the content is variable. Consumers have consistently expressed the desire for accessible, web-based genetic information that they can trust and consider provision of these resources as a role of the Federal Government.

- 5. HHS should create and maintain a state-of-the-art Internet portal to facilitate access to comprehensive, accessible, and trustworthy web-based genetic information and resources for consumers.

Recommendation 6

- 6. Because family health history tools are a potentially powerful asset for consumers and health care professionals to use in risk assessment and health promotion, HHS should:
 - A. Support efforts to educate health care professionals, public health providers, and consumers about the importance of family health history;
 - B. Promote research on how consumers and diverse communities use family history to make health care decisions and incorporate those research findings into consumer educational materials;
 - C. Support the use of family history in clinical care through development of point-of-care educational materials and clinical decision support tools in electronic health records that utilize coded and computable family history, genetic, and genomic information; and
 - D. Promote embedding educational materials in family history collection tools and personal health records directed to consumers and ensure for all by providing these tools in various formats.

APPENDIX A

Public Commenters

The following individuals and organizations responded to a May 24, 2010, request for public comment on an earlier version of this report.

America's Health Insurance Plans
American Academy of Nursing
American Association for Dental Research (Joint comment with American Dental Education Association)
American College of Medical Geneticists
American College of Preventative Medicine
American Dental Education Association (Joint Comment with American Association for Dental Research)
American Medical Association
American Nurses Association
American Pharmacists Association
Association of Genetic Technologists
Association of Molecular Pathology
Association of Professors of Human and Medical Genetics
Bombard, Yvonne, Ph.D.
Campos-Outcalt, Doug
Chen, Frederick M., M.D., M.P.H.
Chicago State University
Cleveland Clinic
Connecticut Department of Public Health
DNAdirect
Drake University School of Pharmacy & Health Sciences
Ehrle, Lynn Howard, M.Ed.
Life Technologies
MedBiquitous
Mount Sinai School of Medicine
National Association of Pediatric Nurse Practitioners
National Society of Genetic Counselors
Navigenics, Inc.
Northwest Association Biomedical Research
Oncology Nursing Society
Quest Diagnostics
Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
Shepherd University
Society of General Internal Medicine
University of Tennessee Health Science Center
Washington State Department of Health
Winston Salem Sate University

APPENDIX B

Literature Review Methodologies

DATABASES SEARCHED

The following databases were searched via DIALOG platform for the time period: 2003-2009. MEDLINE, ERIC (Education Resources Information Center/DOE), Social Science Citation Index, PsycINF, Dissertation Abstracts, Social Sciences Abstracts, Education Abstracts, Biosis Previews, Science Citation Index; EMBASE were accessed.

SEARCH TERMS

Specific words and phrases used in the literature search can be grouped into several categories, recognizing that there is overlap. These categories include educational terminology, scientific terms, social scientific terms and concepts, and terms that identify stakeholders in genetics and genomics education.

Educational terminology used:

EDUCATION, TRAINING, TEACHING, INSTRUCTION, CONTINUING EDUCATION
LITERACY, KNOWLEDGE, COMPETENCE, LEARNING
EDUCATION MODEL
HEALTH EDUCATION
UNIVERSITY PROGRAM, COURSE, CLASSES
SYLLABUS, CURRICULUM
INNOVATE
METHOD

Scientific terms used:

GENOMIC, GENETIC, HUMAN GENOME
PHARMACOGENOMIC, PHARMACOGENETIC
TOXICOGENOMIC, TOXICOGENETIC
FORENSIC
EVOLUTIONARY, EVOLUTION
MOLECULAR
POPULATION GENETICS
EPIDEMIOLOGY

Social scientific terms and concepts used:

ATTITUDE
BELIEF

Stakeholders in genetics and genomics were identified using terms such as:

HEALTHCARE, HEALTHCARE PROVIDER, PRIMARY CARE
PROFESSIONAL, MEDICAL, PHARMACEUTICAL
HEALTH SCHOOL, SCHOOL, COLLEGE, UNIVERSITY
PUBLIC HEALTH
STUDENT, PUPIL
PATIENT
GENERAL PUBLIC, CONSUMER
PHYSICIAN ASSISTANT

APPENDIX C

Genetics and Genomics Competencies, Accreditation, and Licensing of Selected Health Care Providers

HEALTH CARE PROFESSIONALS

Competencies for All Health Care Professionals

Competencies developed for all health care professionals by the National Coalition for Health Professional Education in Genetics include:

At a minimum, each health care professional should be able to:

- Examine one's competence of practice on a regular basis, identifying areas of strength and areas where professional development related to genetics and genomics would be beneficial
- Understand that health-related genetic information can have important social and psychological implications for individuals and families
- Know how and when to make a referral to a genetics professional

In the knowledge domain, all health professionals should understand:

- Basic human genetics terminology,
- Basic patterns of biological inheritance and variation, both within families and within populations,
- How identification of disease-associated genetic variations facilitate development of prevention, diagnosis, and treatment options,
- The importance of family history (minimum three generations) in assessing predisposition to disease,
- The interaction of genetic, environmental, and behavioral factors in predisposition to disease, onset of disease, response to treatment, and maintenance of health,
- The difference between clinical diagnosis of disease and identification of genetic predisposition to disease (genetic variation is not strictly correlated with disease manifestation),
- Various factors that influence the client's ability to use genetic information and services, for example, ethnicity, culture, related health beliefs, ability to pay, and health literacy,
- The potential physical and/or psychosocial benefits, limitations, and risks of genetic information for individuals, family members, and communities,
- Resources available to assist clients seeking genetic information or services, including the types of genetics professionals available and their diverse responsibilities,
- The ethical, legal and social issues related to genetic testing and recording of genetic information (e.g., privacy, the potential for genetic discrimination in health insurance and employment), and
- One's professional role in the referral to or provision of genetics services, and in follow-up of those services.

In the skills domain, all health professionals should be able to:

- Gather genetic family history information, including at minimum a three-generation history,

- Identify and refer clients who might benefit from genetic services or from consultation with other professionals for management of issues related to a genetic diagnosis,
- Explain effectively the reasons for and benefits of genetic services,
- Use information technology to obtain credible, current information about genetics, and
- Assure that the informed-consent process for genetic testing includes appropriate information about the potential risks, benefits, and limitations of the test in question.

In the attitudes domain, all health professionals should:

- Appreciate the sensitivity of genetic information and the need for privacy and confidentiality, and
- Seek coordination and collaboration with an interdisciplinary team of health professionals.

Examples of the eighteen critical “minimums” in the three content areas spanning knowledge, skills, and attitude domains:

Basic requirements, such as understanding:

- Basic genetic terminology,
- Patterns of inheritance,
- Differences between genetic inheritance and risk predisposition,
- The importance of family history,
- The role of the environment in gene-environment interactions,
- Cultural and psychosocial factors,
- How to initiate and follow-through on referral for genetic services,
- Recognition of available resources for patients and families,
- Risks/benefits of genetic testing, and
- Ethical, legal, and social implications in provision of genetics services.

Skill-specific competencies include the ability to:

- Accurately elicit a patient’s three-generation family history,
- Identify and refer clients to relevant professionals given a genetic diagnosis,
- Effectively communicate why a patient would want to consider utilizing genetic services,
- Use technology to obtain accurate information about genetics, and
- Ensure any informed consent process in the genetic testing process includes accurate review of risks, benefits and limits of test being considered.

Attitude-specific requirements outline that health care professionals should be able to:

- Appreciate the need for privacy and confidentiality when working with a patient about their genetic information, and
- Preemptively seek interdisciplinary collaboration with other health care professionals when providing, discussing, or initiating genetic services for a client.

PHYSICIANS

Competencies for Physicians

As part of the Association of American Medical Colleges' (AAMC) 2004 Medical School Objectives Project, 21 learning objectives in genetics were established across attitude, knowledge, and skill domains.²³¹ In January 2010, the AAMC and the Association of Professors of Human and Medical Genetics jointly developed Core Competencies for Medical School Genetics Education providing recommendations on the fundamental genetics principles that should be demonstrated by all medical school graduates.²³² This updated set of competencies conforms to requirements of the Liaison Committee on Medical Education (LCME) that it be mapped to educational objects set forth by the Accreditation Council for Graduate Medical Education and broadly categorize as:

- Organization of the genome and regulation of gene expression as it relates to medical genetic diagnosis;
- Genetic variation and the implications for diversity of normal variation and disease;
- Principles of inheritance patterns;
- Clinical, ethical and social implications for diagnosis, family health, prediction, and personalized medicine;
- Importance of genetic testing including cytogenetics, molecular genetics, genome sequencing, and biochemical genetics;
- Unique features of the genetics for cancer and prenatal diagnosis; and
- Treatment of genetic conditions including family counseling.

In 2009, AAMC collaborated with the Howard Hughes Medical Institute and released a report on updated expected competencies for graduating physicians and pre-medical program students.²³³ Medical school competencies span eight domains; those specific to genetics include knowledge and competent application of “individual and population-based genetics and genomics to guide medical care decisions.” Many subcomponent competencies have genetic and genomic elements such as pharmacogenomics and pharmacogenetics, and the analytical validity, clinical validity, and clinical utility of genetic tests.

From 2000 to 2008, ACMG published numerous condition-specific medical practice and diagnostic evaluation guidelines specific to single-gene disorders, including guidelines for genetic susceptibility to breast and ovarian cancer, carrier screening for spinal muscular atrophy, carrier screening for Ashkenazi Jewish individuals, genetic testing for colon cancer, and many others.²³⁴

These clinical guidelines and practice standards have helped shape practice uniformity with respect to work-ups for common genetic conditions across primary care, pediatrics, oncology, obstetrics, and psychiatric clinical settings. Many of these clinical guidelines were released jointly with the American Society of Human Genetics (ASHG).

²³¹ Association of American Medical Colleges. (2004). *Report VI Contemporary Issues in Medicine: Genetics Education*. See www.ttuhsu.edu/som/curriculum/documents/msop_genetics.pdf. Accessed on November 24, 2009.

²³² Association of Professors of Human and Medical Genetics. *APHMG Medical School Core Curriculum in Genetics*. See <http://www.aphmg.org/>. Accessed on March 31, 2010.

²³³ Howard Hughes Medical Institute-Association of American Medical Colleges. (2009). *Scientific Foundations for Future Physicians: Report of the HHMI-AAMC Committee*. See www.hhmi.org/grants/pdf/08-209_AAMC-HHMI_report.pdf. Accessed on November 24, 2009.

²³⁴ American College of Medical Genetics. *Practice Guidelines*. See www.acmg.net/AM/Template.cfm?Section=Practice_Guidelines&Template=/CM/HTMLDisplay.cfm&ContentID=3701. Accessed on November 24, 2009.

Reflecting the scientific progress beyond single-gene disorders, in January 2007, ASHG released policy recommendations concerning DTC genetic testing technologies.²³⁵ The scope of this policy statement pertained to health-related DTC testing, but the overall policy outlined specific issues that health care providers should be mindful of when interacting with patients who use DTC genetic tests for complex disease susceptibility determinations (e.g., diabetes, heart disease, depression, and cancer). ASHG's primary recommendation concerning health care professionals indicated that professional societies would need to assume a greater level of responsibility in educating their members about this type of genetic testing.

Many professional societies have released or revised practice competency standards or policies focused on genetics and genomics. For example:

- In 2008 the American Academy of Family Physicians released a medical genetics core competency guideline document for residency training.²³⁶ Minimal standards include being able to (1) identify patients at risk for genetic conditions through accurate collection of personal and family histories, (2) effectively ascertain both environmental and behavioral genetic risk factors from a patient interview, (3) appreciate ethical and social implications of any genetic testing efforts, and (4) recognize limitations in personal genetics knowledge and practice capacity by seeking further multi-disciplinary counsel if uncertain about how to help a patient.
- The American Medical Association (AMA) has adopted policies that encourage physicians to become more knowledgeable about genetic testing for complex diseases such as hereditary cancer. The policy encourages patients interested in genetic testing to contact a health care provider and directs the AMA to assist educating physicians about genetics-related clinical practice issues.²³⁷
- The American Academy of Pediatrics' Committee on Genetics has authored numerous policy and professional practice statements on various heritable and complex health conditions.²³⁸
- Updated annually, the American Society of Clinical Oncology develops evidence-based clinical practice guidelines outlining appropriate methods and standards of cancer care related to clinical diagnoses and management of conditions. Included are reviews of current genetic technologies in cancer management settings, and recommendations on use of approved medical procedures and tests.²³⁹

In June 2009, NIH, the Centers for Disease Control and Prevention (CDC), and HRSA convened a workshop that included participants from health professional organizations representing primary care providers.²⁴⁰ The workshop focused on incorporation of genetics and genomic medicine into maternal and

²³⁵ Hudson, K., Javitt, G., Burke, W., and Byers, P., along with the American Society of Human Genetics Social Issues Committee. (2007). ASHG Statement on direct-to-consumer testing in the United States. *The American Journal of Human Genetics*. 81:635-637.

²³⁶ American Academy of Family Physicians. *Recommended Curriculum Guidelines for Family Medicine Residents, Medical Genetics*. See www.aafp.org/online/etc/medialib/aafp_org/documents/about/rap/curriculum/medical_genetics.Par.0001.File.tmp/medicalgenetics.pdf. Accessed on November 16, 2009.

²³⁷ American Medical Association Policy Database. See <http://www.ama-assn.org/ama/pub/advocacy/policy-finder.shtml>. Accessed on January 14, 2010.

²³⁸ American Academy of Pediatrics, Committee on Genetics. See www.aap.org/visit/cmte18.htm#statements. Accessed on November 16, 2009.

²³⁹ American Society of Clinical Oncology. *Guidelines*. See www.asco.org/ASCOv2/Practice+&+Guidelines/Guidelines. Accessed on November 16, 2009.

²⁴⁰ Kemper, A.R., Trotter, T.L., Loyd-Puryear, M.A., Kyler, P., Feero, W.G., and Howell, R.R. (2010). A blueprint for maternal and child health primary care physician education in medical genetics and genomic medicine: recommendations of the United States Secretary for Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children. *Genetics in Medicine*. 12(2):77-80.

child health care. A list of knowledge areas for maternal and child health primary care providers was developed based on the ongoing work of NCHPEG and the recognition that primary care providers underestimate the degree to which genetics and genomic medicine play in the health of their patients.

- Genetics and genomic medicine literacy, including understanding of basic terminology, types of mutations, and how genes and the environment can interact to affect health;
- The interpretation of clinical utility of genetic tests;
- The role of primary care providers in newborn screening;
- How to collect, document, and act on a family health history across the lifespan of a woman and her family;
- Sources for guidelines and clinical recommendations for genetics and genomic medicine in primary care;
- Methods of informing families about genetic testing and obtaining consent;
- How to communicate information about risk of conditions to women before pregnancy and when pregnant; and
- When and how to refer families to a genetic counselor or geneticist.

Workshop participants identified the lack of time as the most important barrier to educating primary care providers in genomic medicine for both those in training and those in practice. Lack of geneticists to provide education, mentoring, and curricular oversight in residency programs and lack of enthusiasm about genetics and genomic medicine by trainees and those in practice limit effective educational efforts.

To address the issues identified during the workshop, the recommendations summarized below, were made and subsequently adopted by the Advisory Committee on Heritable Disorders in Newborns and Children:

- Develop a case-based genetics and genomic medicine educational curriculum that could be incorporated into residency training programs that presents common genetic concepts using scenarios.
- Ensure that board certification exams assess knowledge related to core educational goals and basic literacy in genetics and genomic medicine.
- Make available continuing medical education (CME) at meetings and through the Internet that focuses on practical aspects of incorporating genetics and genomic medicine into primary care, focusing on useful skills such as obtaining family history and identifying red flags for referral for genetic counseling.
- Promote participation in these educational activities through the maintenance of board certification process.
- Create a website that would include clinical recommendations and practical office tools to facilitate incorporation of genetic and genomic medicine into routine practice.

The workshop endorsed the development of the Genetics in Primary Care Training Institute (GPCTI) based on the concept of a “learning collaborative”²⁴¹ that would pair primary care providers with experts in genetic and genomic medicine. These learning collaboratives would develop a 1-year project that includes an outcomes component, and the training institute would then formally evaluate these projects to inform the process of broader dissemination. The Advisory Committee on Heritable Disorders in Newborns and Children approved the learning collaborative concept and recommended that HRSA provide funding for the project. HRSA is implementing this recommendation through the formation of

²⁴¹ Young, P.C., Glade, G.B., Stoddard, G.J., and Norlin, C. (2006). Evaluation of a learning collaborative to improve the delivery of preventive services by pediatric practices. *Pediatrics*. 117(5):1469-1476.

GPCTI and funding the initiative as a Special Project of Regional and National Significance by the Maternal and Child Health Bureau at HRSA.

Education and Licensure of Physicians and Accreditation of Medical Schools

In the United States there are 131 accredited medical schools granting M.D. degrees and 25 colleges of osteopathic medicine granting D.O. degrees.²⁴² In 2001, the Association of Professors of Human and Medical Genetics and ASHG released a report, “Medical School Core Curriculum in Genetics,” outlining critical education elements to be required in medical preparation programs.²⁴³ Building on these efforts in 2004, the AAMC (representing all medical schools, approximately 400 teaching hospitals, 68 Veterans Affairs departments, and 90 professional societies), reported that greater genetics training was a critical requirement and provided competencies.²⁴⁴ Driving this need is a significant shortage of medical genetics experts prepared to address the onslaught of implications stemming from genetic science.^{44,244} Subsequent analyses of issues identified in these reports confirmed that medical students’ genetic knowledge and competence demonstrated a need for medical schools to integrate additional training and education.^{245,246}

A recent analysis of genetic content in graduate medical curriculums found that 77 percent of programs taught medical genetics only in the first year of medical school and that 47 percent failed to incorporate any genetic content in third and fourth year instruction.⁴⁴ Furthermore, only 11 percent provided practical clinical applications of genetics. In addition, 46 percent reported stand-alone courses only, with the remaining respondents offering medical genetic content built into another course. A key recommendation from several organizations to obtain a genetically competent physician workforce is to reorient undergraduate scientific foundations and integrate genetic and genomic science concepts into, and across, all medical education requirements.^{247,248}

To obtain an M.D. professional license, students must successfully pass the United States Medical Licensing Examination (USMLE), a three-step examination administered by the independent medical licensing authority, the National Board of Medical Examiners.²⁴⁹ Genetic content includes DNA and RNA concepts related to biochemistry and molecular biology coursework; congenital human development; Hardy-Weinberg principles; pharmacogenetics; and standard heritable conditions (e.g., single-gene disorders, chromosomal aberrations) and skills related to their clinical management. Genetic content is similarly incorporated in the final examination.²⁵⁰

²⁴² Association of American Medical Colleges. *Medical Schools*. See www.aamc.org/medicalschoools.htm and American Association of Colleges of Osteopathic Medicine. See <http://www.aacom.org/people/colleges/Pages/default.aspx>. Accessed on November 24, 2009.

²⁴³ Association of Professors of Human and Medical Genetics/American Society of Human Genetics. *Medical School Core Curriculum in Genetics*. See www.ashg.org/pdf/Medical%20School%20Core%20Curriculum%20in%20Genetics.pdf. Accessed on November 16, 2009.

²⁴⁴ Korf, B.R. (2005). Genetics training in the genomic era. *Current Opinion in Pediatrics*. 17(6):747-750.

²⁴⁵ Telner, D.E., Carroll, J.C., and Talbot, Y. (2008). Genetics education in medical school: a qualitative study exploring educational experiences and needs. *Medical Teacher*. 30(2):192-198.

²⁴⁶ Baars, M.J., Scherpier, A.J., Schuwirth, L.W., Henneman, L., Beemer, F.A., Cobben, J.M., Hennekam, R.C., Verweij, M.M., Cornel, M.C., and ten Kate, L.P. (2005). Deficient knowledge of genetics relevant for daily practice among medical students nearing graduation. *Genetics in Medicine*. 7(5):295-301.

²⁴⁷ Howard Hughes Medical Institute-Association of American Medical Colleges. 2009. *Scientific Foundations for Future Physicians: Report of the HHMI-AAMC Committee*. www.hhmi.org/grants/pdf/08-209_AAMC-HHMI_report.pdf. Accessed on November 24, 2009.

²⁴⁸ Spencer, A.L., Brosenitsch, T., Levine, A.S., and Kanter, S.L. (2008). Back to the basic sciences: an innovative approach to teaching senior medical students how best to integrate basic science and clinical medicine. *Academic Medicine*. 83(7):662-669.

²⁴⁹ United States Medical Licensing Examination. *About USMLE*. See www.usmle.org/General_Information/general_information_about.html. Accessed on November 16, 2009.

²⁵⁰ United States Medical Licensing Examination. *Content Outline*. See <http://www.usmle.org/Examinations/step1/content/principles.html>. Accessed on November 16, 2009.

To obtain a D.O. license, students must successfully pass the Comprehensive Osteopathic Medical Licensing Examination (COMLEX-USA), also a three-step examination process administered by an independent medical licensing authority—the National Board of Osteopathic Medical Examiners. Genetic content for COMLEX-USA is similar to USMLE, but differing approaches between M.D. and D.O. programs result in variability of the approach to health and illness management on the examinations. Moreover, recently emerging concepts of genomics resulting in dynamic probabilistic contexts for chronic disease in individual patients is usually not included on these examinations.²⁵¹

For all physicians, state medical licensing boards require evidence of CME each year for license re-registration, which needs to be submitted at one to four year intervals depending on the state. Great variability exists across state medical board requirements, with some boards requiring evidence for as little as 12 hours per year, to as many as 50 hours per year.²⁵² Although physicians may obtain genetic education and training through pre-approved sponsored activities such as seminars, conferences, self-learning opportunities, and other professional development activities, requirements are not tied to minimal completion of genetic content. Genetic content across certificates is not well tracked and presumably contains great variability in amount and type of information provided.

Accreditation of U.S. medical school programs is provided through the Liaison Committee on Medical Education (LCME) or the American Osteopathic Association (AOA). Published LCME accreditation standards require basic science instruction and include mention of genetics, but the standards do not outline either amounts or presence of genetics topic requirements before accreditation is issued to a graduate medical education program.²⁵³ AOA similarly addresses genetics in its accreditation processes—presence of genetics is required under the umbrella of basic science requirements and the care of hereditary conditions.²⁵⁴

NURSES

Competencies for Nurses

In 2005, genetics competencies for all practicing RNs were developed by consensus and endorsed by 49 professional organizations, encompassing four areas of clinical action: (1) correctly applying/integrating genetic and genomic knowledge when assessing patients; (2) accurately identifying patient genetic/genomic needs and issues; (3) conducting appropriate patient referrals; and (4) providing competent education, clinical care and psychosocial support to patients and families.^{255,256,257,258}

²⁵¹ United States Medical Licensing Examination. 2009 *USMLE Bulletin* – Overview. See www.usmle.org/General_Information/bulletin/2009/overview.html. Accessed November 24, 2009.

²⁵² State Medical Licensure Requirements and Statistics, 2006. Continuing Medical Education for Licensure Re-registration. See www.ama-assn.org/ama1/pub/upload/mm/455/licensurerereg-06.pdf. Accessed on November 16, 2009.

²⁵³ Liaison Committee on Medical Education. *Functions and Structure of a Medical School, Standards for Accreditation of Medical Education Programs Leading to the M.D. Degree*, June 2008. See www.lcme.org/functions2008jun.pdf.

²⁵⁴ Liaison Committee on Medical Education. *Standards for Accreditation of Medical Education Programs Leading to the M.D. Degree* (2008), See www.osteopathic.org/pdf/SB03-Standards%20of%20Accreditation%20July%202009.pdf. Accessed on November 24, 2009.

²⁵⁵ Jenkins, K., and Calzone, K.A. (2007). Establishing the essential nursing competencies for genetics and genomics. *Journal of Nursing Scholarship*. 39(1):10-16.

²⁵⁶ Calzone, K.A., Cashion, A., Feetham, S., Jenkins, J., Prows, C., Williams, J.K., and Wung, S.F. (2010). Nurses transforming health care using genetics and genomics. *Nursing Outlook* 58(1), 26-35.

²⁵⁷ Consensus Panel on Genetic/Genomic Nursing Competencies (2006). The essentials of baccalaureate education for professional nursing practice. See <http://www.aacn.nche.edu/Education/pdf/baccessentials08.pdf>. Accessed on November 24, 2009.

Correctly integrating genetic and genomic knowledge encompasses the nurse's ability to:

- Appreciate genetics and genomics in prevention, screening, diagnostics, treatment selection, monitoring, and clinical outcome evaluation processes
- Collect a complete family health history
- Accurately construct a multi-generational pedigree
- Collect patient health histories that include genetic/genomic health information
- Perform physical assessments that include genetic/genomic risk factors
- Assess patient understanding of genetic/genomic information
- Competently construct plans of health care that incorporate genetics and genomics

Patient identification skills expected of professional nurses encompasses their ability to:

- Ascertain who could benefit from genetic/genomic information or services
- Recognize accurate sources of genetic/genomic information for patients based upon their unique health needs
- Appreciate relevant ethical, legal, and social implications related to genetic information and genomic technologies
- Define issues acting against a patient's ability to autonomously and voluntarily gather relevant genetic information and act upon findings

Genetics and genomics health care services that all nurses are expected to provide include:

- Accurately interpret genetic/genomic health information (e.g., diagnostic tests, health histories)
- Appropriately collect and review genetic/genomic health information from reliable information sources to facilitate a patient's decisionmaking
- Correctly apply genetics and genomics information into health promotion counseling for patients
- Correctly use genetic/genomic health interventions to improve patient health outcomes
- Work with other members of the multi-disciplinary clinical team, including allied health providers and insurance companies, to provide genetics and genomics clinical care
- Correctly use interventions and treatments that are tailored to patients' genetic/genomic health needs
- Correctly evaluate patient health outcomes following use of genetic/genomic health intervention or treatment, and facilitate redirection of health care planning as necessary

Education and Licensure of Nurses and Accreditation of Nursing Schools

As of 2004, there were more than 2.9 million nurses, of which 45.6 percent graduated from nursing school before 1984.²⁵⁹ Nursing contains great academic and professional heterogeneity stemming from multiple academic pathways to becoming a registered nurse (RN); two accrediting bodies for academic curricula with varying requirements; presence of nursing education programs that lack accreditation; and

²⁵⁸ Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Measures; 2nd edition. (2008). See www.genome.gov/Pages/Careers/HealthProfessionalEducation/geneticscompetency.pdf. Accessed on November 24, 2009.

²⁵⁹ U.S. Department of Health and Human Services. 2005. *The Registered Nurse Population: Preliminary Findings from the National Sample Survey of Registered Nurses, March 2004*. HRSA Bureau of Health Professions, Division of Nursing.

numerous specialty advanced practice pathways with variable routes to certification (e.g., family nurse practitioner (NP), pediatric NP, geriatric NP). Genetic content is required by the American Association of Colleges of Nursing Baccalaureate Essentials, which serve as the basis for Commission on Collegiate Nursing Education (CCNE) accreditation. However, very little data exist to ascertain extent of genetics integration in U.S. nursing curricula. There are more than 1,600 tracked accredited nursing programs; however, there are more schools of nursing that are not accredited and are difficult to monitor.

Entry-level professional RNs may pursue one of four possible academic paths: (1) a four-year baccalaureate in nursing offered by colleges or universities; (2) a two- to three-year associate degree in nursing offered by community and junior colleges; (3) a three-year hospital-based diploma program; or (4) as a Clinical Nurse Leader, that is, an individual who is entry-level with a B.S. in another field but enter nursing with a master's preparation. The current trend within the nursing field; however, has been to pair associate/diploma programs with baccalaureate institutions to increase numbers of nurses with baccalaureate preparation. In 2006, there were 709 organizations offering bachelor's degrees, 850 organizations offering associate degrees, and 70 programs offering hospital diplomas.²⁶⁰

Advanced Practice Nurses (e.g., NPs, clinical nurse specialists, certified nurse midwives, certified registered nurse anesthetists) are RNs who obtain a master's degree from one of the country's 448 accredited nursing programs. Eventually expected to replace master's prepared Advanced Practice Nurses, Doctors of Nursing Practice (D.N.P.) are RNs who obtain a practice-based doctoral degree from one of the country's 92 accredited D.N.P. programs. Available since 2005, the D.N.P. represents a new movement in nursing to incorporate greater foundations of scientific knowledge, as the D.N.P. is equivalent to other health professional doctorates. An additional 100 schools of nursing are expected to implement D.N.P. programs at their institutions in the near future.²⁶¹

The need for education of nurses in genetics is well documented.²⁶² Available figures from a subset of the country's accredited schools of nursing published in 1999 indicated less than 10 median hours of total genetics instruction across programs; 30 percent contained none at all.⁴⁵ A recent follow-up evaluation of a small sub-sample of these schools suggests that not much progress has been made in integrating genetics instruction hours in accredited baccalaureate, accelerated, diploma, and associate degree programs.²⁶³

A 2005 nursing faculty survey conducted by Prows, et al. found that 29 percent of schools reported no genomic curriculum content (no change since similar data were collected in 1996), citing an already overloaded curriculum and lack of knowledge among faculty about genetics.²⁶³ The vast majority of programs responding to the survey offered five hours or less on genetic content.

Individual state boards of nursing manage and issue professional RN licenses; however, some states have chosen to be part of a broader effort to streamline requirements and are members of the National Council of State Boards of Nursing. Individuals completing an approved nursing program by state nursing boards from baccalaureate, associate, or diploma programs must successfully complete the National Council Licensure Examination (NCLEX) to obtain the RN professional license. Little genetic content is contained in NCLEX and certification examinations, and at the master's level, there is significant

²⁶⁰ Bureau of Labor Statistics. *Occupational Outlook Handbook*, 2008-2009 Edition. See www.bls.gov/oco/ocos083.htm. Accessed on November 16, 2009.

²⁶¹ American Association of Colleges of Nursing. *Doctor of Nursing Practice Talking Points*. See <http://www.aacn.nche.edu/DNP/talkingpoints.htm>. Accessed on December 12, 2009.

²⁶² Prows, C.A., Glass, M., Nicol, M.J., Skiton, H., and Williams, J. (2005). Genomics in nursing education. *Journal of Nursing Scholarship*. 37(3):196-202.

²⁶³ Prows, C., Calzone, K., and Jenkins, J. (2006). *Genetics content in nursing curriculum*. Paper presented at the National Coalition Health Professional Education in Genetics, Bethesda, MD.

variability in exam criteria across the certifying organizations. The Genetic Nursing Credentialing Commission is recognized by the American Nurses Association and offers two clinical genetics specialty certifications, one for baccalaureate RNs. (Genetics Clinical Nurse) and one for master's prepared nurses (Advanced Practice Nurse in Genetics). At the time this report was written, there were 40 individuals certified as Advanced Practice Nurse in Genetics and 11 individuals certified as Genetics Clinical Nurse.²⁶⁴ Nurses in genetic practice settings with direct patient, family, client, and colleague in-service teaching responsibilities can obtain these credentials to enhance their professional portfolios.

There are two bodies that accredit educational institutions and curricula for the nursing profession: the National League for Nursing Accrediting Commission (NLNAC) and the CCNE arm of the American Association of Colleges of Nursing. NLNAC accredits all levels of nursing academic programs from diploma and associate degrees (as well as licensed practical nurse programs) to advanced practice and D.P.N.s; the CCNE accredits only baccalaureate and graduate nursing academic programs. The two organizations have very different assessment criteria, and consequently some schools carry accreditation from both. NLNAC and CCNE now require objective evidence of genetic content or instruction in nursing curriculums.²⁶⁵ For programs renewing during the next accreditation cycle in 2010, CCNE will begin to assess if schools are moving toward incorporation of genetic content.

CE for RNs is extremely heterogeneous and in some states is monitored per the requirements of state boards of nursing. Presently, 19 states have no CE requirements for renewal of active RN licenses.²⁶⁶ The remaining states have widely varying requirements, extending from as little as 5 hours of CE per year to as many as 15 hours per year. No state board of nursing has a genetics and genomics requirement for maintenance of an active RN professional license.

PHYSICIAN ASSISTANTS

Competencies for Physician Assistants

Four physician assistant (PA) organizations represent more than 84,000 PAs, 40 percent of whom work in primary care. A 2008 survey by these PA organizations among members found that 85 percent of respondents had gathered family history in the past six months and 70 percent indicated that they had used that information in decisionmaking. Yet only 22 percent reported feeling that their supervising physician was knowledgeable about genetics.

A survey of PA training programs found that 81 percent perceive a need to enhance their genetics curriculum despite an already overloaded curriculum and lack of time to develop resources.²⁶⁷ In response, the Physician Assistance Education Association is creating faculty development opportunities, monitoring and reporting innovations in genetics education, developing curricula resources for best practices, developing assessment tools for students and faculty, and developing a database to track genetics activities and outcomes in PA education. These professional organizational efforts use traditional methods of dissemination—newsletters, annual conferences, journals, and web-based continuing education activities—to educate members in genetics. Recently, an ad hoc group of clinical leaders

²⁶⁴ Personal communication, Jeanine Seguin Santelli, Ph.D., A.N.P.-B.C./G.N.P.-B.C. GNCC, Executive Director. Keuka Park, N.Y. December 14, 2009.

²⁶⁵ National League for Nursing Accrediting Commission, Inc. *NLNAC 2008 Standards and Criteria*. See www.nlnac.org/manuals/SC2008.htm. Accessed on November 24, 2009.

²⁶⁶ NurseWeek. *Nursing Continuing Education Requirements by State*. See www.nurse.com/ce/Requirements.html; <http://www.nursingcenter.com/pdf.asp?AID=636579>. Accessed on November 16, 2009.

²⁶⁷ Presented by M. Rackover at “Developing a Blueprint for Primary Care Physician Education in Genomic Medicine,” June 8-9, 2009. National Institutes of Health, Bethesda, MD.

established the *Essential Physician Assistant Guidelines for Genetics and Genomics*.²⁶⁸ Similar to other professional efforts, their proposed competencies are focused on three core concepts—knowledge, skills, and attitudes.

Knowledge requirements include understanding genetics terminology, inheritance patterns, diagnostics, family history assessment, screening, and making appropriate referrals, among other issues. PAs are expected to have the skills to elicit family history, identify the need for referrals, provide patient education (including providing credible sources of information), and assess the benefits and limits of genetic tests. They are also expected to understand the sensitivity of genetic information, appreciate psychosocial and cultural factors, and be knowledgeable about social, legal, and ethical concerns.

Education and Licensure of Physician Assistants and Accreditation of PA Programs

Academic paths to becoming a PA include baccalaureate study prior to acceptance into a Surgical or Physician Assistant graduate program. There are presently 136 accredited PA programs in the United States; they average 26 months in duration and comprise one year of didactic and one year of clinical training. Recent survey results of 100 accredited PA programs indicated two-thirds of them devote 7 to 20 hours to genetics content in their curricula, and many plan to incorporate further genetic content in the near future.²⁶⁹ Recognizing the importance that genetics is garnering for future clinical practice, recent foundational curriculum guidelines were issued, and cover content ranging from classic medical genetics to Human Genome Project implications and polymorphisms as genetic health markers.²⁷⁰ Following completion of an accredited program, the National Commission on Certification of Physician Assistants (NCCPA) certifies PA candidates. For individuals to receive the Physician Assistant-Credentialed (PA-C) credential, they must meet professional knowledge and skill standards as measured by successful performance on the Physician Assistant National Certifying Exam (PANCE).²⁷¹ Although covering single-gene disorders and other hereditary conditions, the PANCE does not include a genetics section or genomics content.

Accreditation of physician assistant programs is granted via the Accreditation Review Commission on Education for the Physician Assistant. The current standards were last reviewed in 2006 and include requirements for instruction of molecular concepts as related to health and disease, including genetics.²⁷² However, similar to other disciplines, these standards are largely restricted to biologic scientific principles and limited clinical application contexts, such as single-gene disorders.

The American Academy of Physician Assistants is the primary professional organization representing the clinical, educational, and research interests of the PA community and offers discipline-specific CE. To maintain active certification status, certified PAs must complete 100 CE hours every two years. At least half of all CE units (50 hours) must come from attending seminars or conference sessions from pre-approved sponsor sources. The remaining 50 hours of CE can come from elective sources (e.g., journal reviews, practice-related activities, self-learning modules, independent studies), for which genetics and genomics content is covered only as a function of individual interest. Re-certification is required every six

²⁶⁸ Rackover, M., Goldgar, C., Wolpert, C., Healy, K., Feiger, J., and Jenkins, J. (2007). Establishing essential physician assistant clinical competencies guidelines for genetics and genomics. *Journal of Physician Assistant Education*. 18(2):47-48.

²⁶⁹ National Institutes of Health. Physician Assistant Competencies for Genomic Medicine: Where We Are Today and How to Prepare for the Future. Meeting Summary, March 29-30, 2007. See www.genome.gov/Pages/About/OD/ReportsPublications/PAMeetingSummaryMarch2007.pdf. Accessed on November 16, 2009.

²⁷⁰ Goldgar, C.M., and Rackover, M. (2008). Recommendations for a Physician Assistant medical genetics curriculum. *The Journal of Physician Assistant Education*. 19(2):30-36.

²⁷¹ National Commission on Certification of Physician Assistants. See www.nccpa.net/. Accessed on November 24, 2009.

²⁷² Accreditation Review Commission on Education for the Physician Assistant, Inc. *Accreditation Standards for Physician Assistant Education*. 3rd edition (October, 2007). See http://www.arc-pa.org/acc_standards/. Accessed on November 24, 2009.

years by NCCPA via the Physician Assistant National Recertifying Exam, with genetic examination content similar to the PANCE.

GENETIC COUNSELORS

Competencies for Genetic Counselors

Practiced-based competencies were issued by the American Board of Genetic Counseling in 2008.²⁷³ They focus on the need for all genetic counselors to demonstrate competency spanning four skill-based content domains: (1) communication; (2) critical thinking; (3) interpersonal counseling and psychosocial assessment; and (4) professional ethics and values.

Professional ethics and values expected of genetics counselors include the ability to serve their profession by maintaining expected ethical, legal and philosophical approaches valued by the genetic counseling community; advocating for clients and families; presenting and exploring research options with clients; accurately identifying self limitations in knowledge and practice capacities; and continually developing professionally.

Communication skills encompass the genetic counselor's need to:

- Establish a mutually agreeable counseling plan with clients
- Comprehensively elicit family history information
- Accurately obtain client medical histories in a variety of clinical settings
- Ascertain complete social/psychosocial histories
- Accurately convey technical medical and genomic information to clients
- Accurately communicate reproductive options
- Communicate all information to clients and families with cultural competence, and
- Plan and organize professional education programs on genetics and counseling issues

Critical thinking skills for genetic counselors include the ability to:

- Identify and calculate genetic and teratogenic predictive risks
- Evaluate a client's social/psychosocial history
- Integrate the entirety of a client's medical information to guide client/family counseling needs
- Demonstrate ability to manage case portfolio needs
- Assess a client's capacity and ability to understand genetic information and redirect care plans accordingly, and
- Identify and access local, regional, and national clinical genetics resources for clients and families

Interpersonal counseling and psychosocial assessment involve the genetic counselor's need to:

- Provide accurate response to client/family concerns that may emerge unexpectedly or over time
- Correctly ascertain and interpret a client's communication and behavioral cues
- Correctly use a wide variety of interviewing methods
- Provide necessary psychological support for a client's short term needs

²⁷³ American Board of Genetic Counseling. *Practice Based Competencies*. See [http://abgc.iasonline.com/CMFiles/Practice Based Competencies Aug 2006 10-29-0951KFH-10292008-6844.pdf](http://abgc.iasonline.com/CMFiles/Practice%20Based%20Competencies%20Aug%202006%2010-29-0951KFH-10292008-6844.pdf). Accessed on November 16, 2009.

- Assist clients to make their own personal health decisions in an unbiased, noncoercive, and nonjudgmental way, and
- Demonstrate capacity for professionalism in multi-disciplinary health care teams

Education and Certification of Genetic Counselors and Accreditation of Genetic Counseling Programs

Academic paths to becoming a genetic counselor include baccalaureate study prior to acceptance into one of the country's 30 accredited graduate genetic counselor programs. Following completion of an accredited program, candidates are eligible for certification from the American Board of Genetic Counseling (ABGC) to obtain the Certified Genetic Counselor credential, which remains active for a period of 10 years. In 2010, this 10-year period will be halved, and certification will be granted in 5-year increments. As of January 2008, six states—California, Illinois, Massachusetts, Oklahoma, Tennessee, and Utah—require a professional license in addition to certification.²⁷⁴

The ABGC accredits genetic counselor programs. Revised in March of 2009, the expanded genetic and genomic content requirements are built into accreditation standards.²⁷⁵ Included in the accreditation requirements are the expected molecular concepts such as inheritance patterns, population genetics, human genetic variation and related susceptibilities, family history analysis, and human development and reproduction. Also included are laboratory and research experiences, as related to capacity for competent clinical practice.

Current pathways for recertification are successful re-examination or through accumulation of CE credits. CE for genetic counselors are issued and monitored by the ABGC, which has specific Professional Activity Credit requirements that may be fulfilled through a wide range of professional development paths.²⁷⁶ The primary professional society representing genetic counselors, the National Society of Genetic Counselors, provides CE units per pre-approved criteria and sponsor initiated activities.²⁷⁷ The ABGC CE program currently is being restructured to meet the 5-year recertification cycle going into effect in 2010.

PHARMACISTS

Competencies for Pharmacists

Pharmacists are recognized as medication experts who improve overall patient care through partnering with physicians.²⁷⁸ In defining the role of pharmacists in the emerging field of pharmacogenomics, Brock stated “the ability to use genetic information as part of individualized patient care complements the

²⁷⁴ National Conference of State Legislators. *Genetic Counselor Licensing*. See www.ncsl.org/default.aspx?tabid=14282. Accessed on November 24, 2009.

²⁷⁵ American Board of Genetic Counseling, Inc., Required Criteria for Graduate Programs in Genetic Counseling Seeking Accreditation by The American Board of Genetic Counseling. See www.abgc.net/CMFiles/REQUIRED_CRITERIA_revised_Mar_25_200951KIH-432009-1159.pdf. Accessed on November 25, 2009.

²⁷⁶ American Board of Genetic Counseling. Professional Activity Credits. See www.abgc.net/english/view.asp?x=1659. Accessed on November 25, 2009.

²⁷⁷ National Society of Genetic Counselors. Continuing Education Information. See www.nsgc.org/CEU/ApprovedPrograms1.cfm. Accessed on November 25, 2009.

²⁷⁸ Leape, L.L., Cullen, D.J., Dempsey Clapp, M., Burdick, E., Demonaco, H.J., Erickson, J.I., and Bates, D.W. (1999). Pharmacist participation on physician rounds and adverse drug events in the intensive care unit. *Journal of the American Medical Association*. 282:267-270.

professional role of pharmacists.”²⁷⁹ Brock identified three specific roles: (1) researcher or discoverer; (2) educator or faculty scientist; and (3) clinician or practitioner. More recent literature has addressed the need for the pharmacy profession to embrace new roles while recognizing that there is little empirical evidence about services and outcomes.²⁸⁰ Gaps persist between knowledge in pharmacogenomics and clinical application but potential roles for pharmacists include developing research methodologies to evaluate the link between genetics and drug response establishing the value of pharmacogenetic testing in clinical practice, and implementing pharmacogenetics in the clinical setting.²⁸¹

Although recognition of the inherited differences in drug effects was documented as early as 1931,²⁸² it was not until 2002 that the American Association of Colleges of Pharmacy’s (AACP) Academic Affairs Committee made specific recommendations regarding the need to develop a requisite knowledge base for pharmacists in the emerging areas of pharmacogenomics and pharmacogenetics. Guided by the NCHPEG recommendations on health professional core competencies in genetics, the AACP Academic Affairs Committee presented a draft set of competencies for pharmacists.²⁸³ These included specific competencies within three broad categories: (1) knowledge, skills, and attitudes relative to the genetic basis of disease; (2) knowledge and skills relative to drug discovery and disposition/drug targets; and (3) ethical applications and social and economic implications.

Accreditation of Pharmacists in Genetics and Genomics

In 2002, Brock et al. sent surveys to the curriculum committee chairpersons at the 82 accredited pharmacy schools in the United States, asking how many lecture hours were devoted to genomic topics.²⁸⁴ Of the 50 responses, 64 percent reported 0 to 1 hour devoted to ethical considerations, and 30 percent reported 0 to 1 hour for practical applications. By 2005, 78 percent of pharmacy schools surveyed provided some instruction in pharmacogenomics. However, the average pharmacy school that included instruction related to pharmacogenomics addressed only half of the AACP Academic Affairs Committee 2002 recommendations regarding the need for pharmacogenomics and pharmacogenetics knowledge.²⁸⁵

The AACP House of Delegates passed policy resolutions in 2008 stating that pharmacy curricula must adequately address contemporary issues associated with biotechnology advances in personalized medicine, including competencies in genetics and genomics and preparing faculty to contribute to education and research related to genetics and genomics.²⁸⁶

²⁷⁹ Brock, T.P., Valgus, J.M., Smith, S.R., and Summers, K.M. (2003). Pharmacogenomics: implications and considerations for pharmacists. *Pharmacogenomics*. 4(3):321-330.

²⁸⁰ Clemerson, J.P., Payne, K., Bissell, P., and Anderson, C. (2006). Pharmacogenetics, the next challenge for pharmacy? *Pharmacy World and Science*. 28:126-130.

²⁸¹ El-Ibiary, S., Cheng, C., and Alldredge, B. (2008). Potential roles for pharmacists in pharmacogenetics. *Journal of the American Pharmacology Association*. 48:e21-e32.

²⁸² Garrod, A.E. (1931). *The Inborn Factors in Disease: An Essay*. Oxford: Oxford University Press.

²⁸³ Johnson J.A., Bootman, J.L., Evans, W.E., Hudson, R.A., Knoell, D., Simmons, L., Straubinger, R.M., and Meyer, S.M. (2002). Pharmacogenomics: A scientific revolution in pharmaceutical sciences and pharmacy practice. Report of the 2001-2002 Academic Affairs Committee. *American Journal of Pharmaceutical Education*. 2002;66(Winter Supplement):12S-15S.

²⁸⁴ Brock, T.P., Faulkner, C.M., Williams, D.M., and Smith, S.R. (2002). Continuing education programs in pharmacogenomics for pharmacists. *American Journal of Health Systems Pharmacology*. 59:722-725.

²⁸⁵ Latif, D.A., and McKay, A. (2005). Pharmacogenetics and pharmacogenomics instruction in colleges and schools of pharmacy in the United States. *American Journal of Pharmaceutical Education*. 69(2):Article 23.

²⁸⁶ American Association of Colleges of Pharmacy. Minutes of the House of Delegates Sessions, July 20-23, 2008. See www.aacp.org/governance/HOD/Documents/HOD_Minutes08.pdf. Accessed on November 25, 2009.

In 2009, Murphy et al. conducted a follow-up survey to Brock's 2002 survey. Results indicate that 92 percent of colleges of pharmacy reported teaching pharmacogenomics within their programs, up from 78 percent of programs surveyed in 2005.²⁸⁷

To meet the pharmacogenomic educational needs of U.S. Colleges of Pharmacy, the Pharmacogenomics Education Program: Bridging the Gap between Science and Practice (*PharmGenEd*TM),²⁸⁸ was developed. Funded by CDC, it is an evidence-based pharmacogenomics education program designed for pharmacists and physicians, pharmacy and medical students, and other health care professionals. The program team at University of California, San Diego Skaggs School of Pharmacy and Pharmaceutical Sciences is collaborating with national pharmacy, medical, and health care organizations to deliver *PharmGenEd*TM materials to more than 100,000 pharmacists, physicians, and health care professionals. Program directors have conducted ongoing surveys and collected evaluation data from resulting *PharmGenEd*TM educational programs. Highlights of pre- and post-program survey results were provided at the 2009 American Pharmacists Association's annual meeting, showing, for example, increased knowledge of adverse drug reactions related to HLA-B*5701 variation and increased overall ability to address pharmacogenomic testing with patients. As a result of the program, pharmacists indicated they would be more likely to:

- Explain the rationale to patients for pharmacogenomic testing (69 percent)
- Discuss risks and benefits of pharmacogenomic testing with patients (67 percent)
- Find credible and current literature related to pharmacogenomic testing (63 percent)
- Recommend or refer patients for pharmacogenomic testing, if applicable (61 percent)
- Recommend the *PharmGenEd*TM CE/CME program to colleagues (84 percent)
- Agree that the pharmacy profession should be more active in educating patients and other health care professionals about pharmacogenomic testing (88 percent)
- Understand that issues related to ethical, social, legal, and economic aspects of genetics are important in translating pharmacogenomics evidence into practice (96 percent).

²⁸⁷ Murphy, J.E., Green, J.S., Adams, L.A., Squire, R.B., Kuo, G.M., and McKay, A. (2010). Pharmacogenomics in the curricula of colleges and schools of pharmacy in the United States. *American Journal of Pharmaceutical Education*. 74(1):1-10.

²⁸⁸ University of California, San Diego Scaggs School of Pharmacy and Pharmaceutical Sciences. Pharmacogenomics Educational Program PharmGenEd. See <http://pharmacogenomics.ucsd.edu>. Accessed on September 8, 2009.

APPENDIX D

SACGHS Survey of Health Care Professional Organizations

1. HEALTH CARE PROFESSIONAL ORGANIZATIONS SURVEYED

A total of 60 organizations were invited to participate in the survey. They were broken into three groups for analysis: genetic-specific organizations, nongenetic organizations, and Federal advisory committees.

Genetic-Specific Organizations (9)

American Board of Genetic Counseling (ABGC)
American Board of Medical Genetics (ABMG)
American College of Medical Genetics (ACMG)
American Society of Human Genetics (ASHG)
Association of Professors of Human and Medical Genetics (APHMG)
Genetic Nursing Credentialing Commission (GNCC)
International Society of Nurses in Genetics (ISONG)
National Coalition for Health Professional Education in Genetics (NCHPEG)
National Society of Genetic Counselors (NSGC)

Nongenetic Organizations (48)

Accreditation Council for Graduate Medical Education (ACGME)
Accreditation Review Commission on Education for the Physician Assistant (ARC-PA)
Alliance of Academic Internal Medicine (AAIM)
American Academy of Family Physicians (AAFP)
American Academy of Nursing (AAN)
American Academy of Pediatrics (AAP)
American Academy of Physician Assistants (AAPA)
American Association of Colleges of Nursing (AACN)
American Association of Colleges of Osteopathic Medicine (AACOM)
American Association of Colleges of Pharmacy (AACCP)
American College of Clinical Pharmacology (ACCP)
American College of Obstetricians and Gynecologists (ACOG)
American College of Physicians (ACP)
American College of Preventive Medicine (ACPM)
American Dental Education Association (ADEA)
American Medical Association (AMA)
American Nurses Association (ANA)
American Osteopathic Association (AOA)
American Residency Coordinators in Obstetrics and Gynecology (ARCOG)
American Society for Clinical Oncology (ASCO)
Association of American Indian Physicians (AAIP)
Association of American Medical Colleges (AAMC)
Association of Black Women Physicians (ABWP)
Association of Family Medicine Program Directors (AFMPD)
Association of Pediatric Program Directors (APPD)
Association of Professors of Gynecology and Obstetrics (APGO)

Association of Schools of Allied Health Professions (ASAHP)
Association of Schools of Public Health (ASPH)
Association of Women’s Health, Obstetric and Neonatal Nurses (AWHONN)
Council on Medical Student Education in Pediatrics (COMSEP)
Council on Resident Education in Obstetrics and Gynecology (CREOG)
Liaison Committee on Medical Education (LCME)
National Association of Pediatric Nurse Practitioners (NAPNP)
National Black Nurses Association (NBNA)
National Board of Medical Examiners (NBME)
National Coalition of Ethnic Minority Nurses Association (NCEMNA)
National Hispanic Medical Association (NHMA)
National League of Nursing (NLN)
National Medical Association (NMA)
National Organization of Nurse Practitioner Faculties (NONPF)
Network of Ethnic Physician Organizations (NEPO)
Oncology Nursing Certification Corporation (ONCC)
Oncology Nursing Society (ONS)
Physician Assistant Education Association (PAEA)
Robert Graham Center
Sigma Theta Tau International (STTI)
Society of General Internal Medicine (SGIM)
Society of Teachers of Family Medicine (STFM)

Federal Advisory Committees (3)

Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)
Advisory Committee on Training in Primary Care Medicine and Dentistry (ACTPCMD)
Council on Graduate Medical Education (COGME)

2. HEALTH CARE PROFESSIONAL ORGANIZATIONS’ SURVEY METHODOLOGY

The main body of the survey instrument consisted of 17 questions developed by SACGHS. Close-ended questions were in multiple-choice or Likert scale formats. Organizations were also asked to complete a narrative description of ongoing genomics-related projects. A supplemental question asked for specialized information based on the mission of the organization. The draft instrument was piloted with board members of the NCHPEG, refined, and subsequently reviewed by a survey methodologist to maximize survey validity.

In December 2008, the survey was sent via e-mail to key staff in 60 targeted organizations. All nonresponders were contacted by e-mail and/or telephone by SACGHS staff to maximize response rates. Thirty-six responses were received (60 percent).

Survey data were compiled and analyzed by SACGHS staff. The organizations were divided into three major divisions: genetic-specific organizations, nongenetic organizations, and Federal advisory committees, and analyses were conducted according to those divisions. Responses were extracted from returned surveys and manually entered into a FileMaker Pro 10 database. Once complete, the derived data were exported as an Excel spreadsheet for further analyses.

3. HEALTH CARE PROFESSIONAL ORGANIZATIONS' SURVEY INSTRUMENT

- 1) Name of organization:
- 2) What is your title and primary role in the organization?
- 3) What is the size of your organization's constituency or membership?
- 4) Please identify which of the following most closely describes your organization's mission. **Circle or underline one answer.**
 - A) Advocacy for and support of practicing health professionals
 - B) Education and training of health professionals
 - C) Certification of health professionals
 - D) Accreditation or certification of institutions
 - C) Other (please describe):
- 5) Is genetics education and training part of the role or responsibility of your organization?
If no, please proceed to question 6. If yes,
 - A) Please briefly describe this role or responsibility.
 - B) Is your organization currently able to fulfill this role or responsibility?
 - C) Are there ways in which your organization could meet this role or responsibility more effectively? If yes, please describe how.

For questions 6-10, please circle or underline the most appropriate number; circle or underline NA if not applicable to your organization.

- 6) What importance does your organization place on the development and promotion of educational activities (including continuing education) in the health area generally?
Not at all important **1** **2** **3** **4** **5** **NA** Very important
- 7) What importance does your organization place on the development and promotion of educational activities (including continuing education) specifically related to genetics and genomics?
Not at all important **1** **2** **3** **4** **5** **NA** Very important
- 8) Where does genetics and genomics education fall relative to the overall priorities facing your organization?
Low priority **1** **2** **3** **4** **5** **NA** High priority
- 9) To what extent is your organization's membership satisfied with the organization's current emphasis on genetics and genomics education?
Not at all satisfied **1** **2** **3** **4** **5** **NA** Extremely satisfied

10) How proficient and comfortable would you say your organization's leadership is with genetics and genomics education?

Low expertise/comfort **1** **2** **3** **4** **5** **NA** High expertise/comfort

11) Does your organization have an established committee, workgroup, or dedicated staff that deals specifically with topics in genetics or genomics relevant to your organization's mission? **Please circle or underline one answer.**

- A) Yes
- B) No
- C) Not sure (please explain):

12) Which of the following do you consider to be barriers to your organization's ability to provide genetics and genomics education? **Please circle or underline all that apply.**

- A. Genetics and genomics education is not applicable to the organization's mission
- B. The organization's leadership lacks knowledge of genetics and genomics
- C. The organization has competing priorities
- D. There is a lack of accessible educational resources for genetics and genomics
- E. Genetics and genomics is not emphasized in certifying examinations/credentialing standards
- F. The organization believes there is a lack of evidence supporting clinical effectiveness of care based on genetic or genomic information
- G. Other (please list):
- H. From our organization's perspective, there are no barriers

13) In the space below, please rank the items selected in question 12 from the most important to least important barrier (e.g., E, D, C).

14) Please fill out the table below to describe any **completed** initiatives/programs your organization has implemented in the last five years for educating its membership on genetics and genomics topics. Please expand the table as needed for each section or to include additional programs.

Program #1	
Brief description	
Outcome measures used to evaluate program's success	
External collaborators (if applicable)	
URLs for web-based resources related to the program	
Publication citations (if any) related to the program	

Program #2	
Brief description	
Outcome measures used to evaluate program's success	
External collaborators (if applicable)	
URLs for web-based resources related to the program	
Publication citations (if any) related to the program	

15) Please fill out the table below to describe any **ongoing or planned** initiatives/programs of your organization for educating its membership on genetics and genomics topics. Please expand the table as needed for each section or to include additional programs.

Program #1	
Brief description	
Outcome measures used to evaluate program's success	
External collaborators (if applicable)	
URLs for web-based resources related to the program	
Publication citations (if any) related to the program	
Program #2	
Brief description	
Outcome measures used to evaluate program's success	
External collaborators (if applicable)	
URLs for web-based resources related to the program	
Publications citations (if any) related to the program	

16) Has your organization surveyed or received input from your membership about genetics and genomics education needs or priorities?
If yes, please briefly summarize the responses or the input.

17) What types of programs or resources could enhance the engagement of your organization's members in genetics and genomics education? Are there programmatic needs that could be addressed by the Federal government?

Specialized Information

Please answer the questions in only one category below. Select the category that is **most relevant** to the mission of your organization (i.e., education, practice advocacy, certification of professionals, accreditation of institutions). If needed, please use additional space to answer the questions. If your organization does not fall into one of these categories, please state that none of the categories apply.

Category 1: Education and training of health professionals

- 1) What is the role of your organization in health professional education?
- 2) From the perspective of your organization, please characterize the need for the integration of genetics and genomics into the curriculum and training of health professionals.
- 3) Briefly describe required and optional curriculum components related to genetics and genomics.
- 4) Is cultural competency incorporated into curricula?
If yes, is it incorporated in a required or optional component of the curriculum?
- 5) Does your organization provide assistance or guidance in developing genetics and genomics curriculum to your membership?
If yes, what type of assistance/guidance?
- 6) Are there gaps in genetics and genomics education?
If yes, please describe briefly. How could these gaps be addressed?
- 7) Looking ahead 5 to 10 years, what needs do you anticipate in genetics and genomics education?

Category 2: Advocacy for and support of practicing health professionals

- 1) What is the role of your organization in education, training, and assessment of the professional workforce?
- 2) Do you offer continuing education programs/activities?
If yes, are any specific to genetics or genomics?
- 3) Has your organization published any position statements or practice competencies regarding genetics? (Please circle or underline your answer)
A) Yes
B) No
C) In progress
D) Not sure (please explain):
- 4) Do you think your members need more information about genetics and genomics?
If yes, on what topics?
- 5) What would help to promote a greater knowledge of genetics and genomics?

Category 3: Certification of Health Professionals

- 1) Do current credentialing exams include questions on genetics and genomics?
If yes, approximately what percentage of the questions is on genetics and genomics?
- 2) How frequently are the questions updated?
- 3) Would your organization like help in developing questions on genetics and genomics?

Category 4: Accreditation or Certification of Institutions

- 1) Are there minimum curriculum requirements in genetics or genomics?
If yes, please provide a brief description.
- 2) If there are minimum curriculum requirements in genetics or genomics, how often are they updated?
- 3) From the perspective of your organization, please characterize the need for the integration of genetics and genomics into the curriculum and training of health professionals.

4. NAME AND MEMBERSHIP SIZE OF RESPONDING HEALTH CARE PROFESSIONAL ORGANIZATIONS

The table below lists the organizations that responded to the survey with their reported constituency or membership numbers. Organizations that represent a profession as a whole have some overlap in numbers with smaller subgroups (e.g., the American Nursing Association and the Oncology Nursing Society). Thus, the membership or constituency numbers cannot be added together, and the total number of unique health professionals represented by these organizations is not known.

Abbreviation	Organization Name	Membership/ Constituency
	Genetic-Specific (9 of 9 returned = 100 percent)	
ABGC	American Board of Genetic Counseling	2,488
ABMG	American Board of Medical Genetics	2,000
ACMG	American College of Medical Genetics and ACMG Foundation	1,500
ASHG	American Society of Human Genetics	7,500
APHMG	Association for Professors of Human and Medical Genetics	100
GNCC	Genetic Nurses Credentialing Commission	47
ISONG	International Society of Nurses in Genetics	415
NCHPEG	National Coalition for Health Professional Education in Genetics	65
NSGC	National Society of Genetic Counselors	2,400
	Nongenetic Organizations (25 of 48 returned = 54 percent)	
ACGME	Accreditation Council for Graduate Medical Education	9,200
ARC-PA	Accreditation Review Commission on Education for the Physician Assistant	163
AAIM	Alliance for Academic Internal Medicine	6,500
AAFP	American Academy of Family Physicians	94,600
AAP	American Academy of Pediatrics	60,000
AAPA	American Academy of Physician Assistants	75,000
AACN	American Association of Colleges of Nursing	625
AACP	American Association of Colleges of Pharmacy	2,910
ACCP	American College of Clinical Pharmacology	2,910
ACOG	American College of Obstetricians and Gynecologists	54,000
ACP	American College of Physicians	126,000
ACPM	American College of Preventive Medicine	2,500
ADEA	American Dental Education Association	17,000
AMA	American Medical Association	250,000

ANA	American Nurses Association	2,900,000
AOA	American Osteopathic Association	64,000
ARCOG	American Residency Coordinators in Obstetrics and Gynecology	225
AWHONN	Association of Women's Health, Obstetric and Neonatal Nurses	23,000
COMSEP	Council on Medical Student Education in Pediatrics	300
NAPNAP	National Association of Pediatric Nurse Practitioners	7,000
ONCC	Oncology Nursing Certification Corporation	27,000
ONS	Oncology Nursing Society	35,000
PAEA	Physician Assistant Education Association	75,000
STTI	Sigma Theta Tau International	130,000
SGIM	Society of General Internal Medicine	2,500
	Federal Advisory Committees (2 of 3 returned = 67 percent)	
ACHDNC	Advisory Committee on Heritable Disorders in Newborns and Children	N/A
CGME	Council on Graduate Medical Education	N/A

5. RESPONSES TO THE HEALTH CARE PROFESSIONAL ORGANIZATION SURVEY

Table 1 Genetics Education and Training as Part of the Role or Responsibility of the Organization (Survey Question 5)

	Number of Responses
Yes	25
No	5
No Answer	6

Table 2 Organizations' Role or Responsibility in Genetics Education and Training (Survey Question 5a)

Organization Abbreviation	Education and Training Role Description
AACN	<p>AACN has partnered with NHGRI and NCI on several initiatives:</p> <ol style="list-style-type: none"> 1. Assisting with creating and endorsing the Essential Competencies and Curricula Guidelines for Genetics & Genomics (2005). 2. Assisting with creating a tool kit for faculty development. 3. Assisting with creating a tool kit repository.
AAFP	<ul style="list-style-type: none"> • Educates family medicine residents and, through CME, educates its physician members. • Regarding resident education, AAFP participates in the review committee for family medicine program requirements related to the Accreditation Council for Graduate Medical Education (ACGME). AAFP has devised curriculum guidelines for family medicine residents on medical genetics, based on the ACGME recommendations for educational competencies. • There is no specific requirement for genetics in its CME, however, AAFP incorporates genetic/genomic components into CME programs as relevant. Currently, presenters of CME may get a faculty pre/post checklist prompting them to include any relevant areas related to their presentation, of which genetics is one. CME presenters may also be given a needs assessment that includes genetics and genomics as necessary.

	<ul style="list-style-type: none"> • AAFP is a member of NCHPEG.
AAP	The AAP is concerned about all aspects of pediatric care, including genetics.
APA	A responsibility of our organization is to provide opportunities for continuing medical education on topics of relevance to physician assistant practice. We identified genetics as an important area and provide CME through our annual conference, journal articles and partnerships with other organizations, like NCHPEG, to create CME programs for PAs.
ABGC	<p>Yes. While we do not provide education ourselves, we accredit the genetic counseling training programs. In this way, we influence the curriculum used in the education of genetic counselors. In addition, we provide certification and recertification for practicing genetic counselors which ensures their competence.</p> <p>Competencies (PBCs) were originally developed in 1996 (Fine BA et al. JGC 1996; S: 113-121) as the basis for the beginning of ABGC accreditation of genetic counseling training programs. They were reviewed by smaller workgroups of current and former ABGC Board members in attendance at the Chicago retreat in 2005 and minor revisions of the language were made. In addition, in 2008 ABGC undertook its first practice analysis of genetic counselors to develop a detailed content outline for our certification examination beginning with the 2009 exam. The certification examination items each map directly to a component of the detailed content outline. Since this is skill-based, the examinee has to have mastered the background genetic counseling knowledge in order to pass the exam. It is important that there are numerous opportunities for our diplomats to obtain continuing education units through conferences on genetics and genomics.</p>
ABMG	ABMG accredits training programs in clinical cytogenetics, biochemical genetics, and molecular genetics. Educational standards are designed by the ABMG for implementation by the training programs.
ACCP	Pharmacogenetics is a component of clinical pharmacology; therefore it may be included in the symposia that we sponsor. We belong to NCHPEG, and provide information and web links regarding their genetics teaching resources to our membership via e-mail notices.
ACHDNC	<p>The grant program established under Section 1109 of our authorizing legislation specifies these activities.</p> <ol style="list-style-type: none"> 1. Assist in providing health care professionals and laboratory personnel education and training in newborn screening. 2. Provide educational programs to parents, families and patient advocacy groups.
ACMG	As a membership organization representing medical geneticists, it is inherent in our responsibilities. Our members direct training programs for medical geneticists and are directly involved in teaching and training of others in academic medical centers.
ACPM	ACPM is currently developing a CME program for its membership and broader community of primary care physicians.
ACP	We incorporate genetics education into our live courses and publish materials that include genetics education.
ACOG	Develop clinical guidelines and patient and professional resources.
AMA	The AMA mission is to support physicians by working on important health issues. The AMA Program in Genetics and Molecular Medicine aims to identify genetics issues relevant to physicians and provide educational support to physicians as they integrate genetic technologies into clinical practice.
ANA	Provide online materials or links to CE opportunities.
APHMG	We represent professors of genetics in all areas of genetics, and are involved in resident, fellow, medical student, and graduate student education.
ASHG	Support of trainees in presenting research, travel to meetings. Our director of education and Committee help with education in K-12 to open the pipeline early.

AWHONN	Genetics information is integrated in other specialty specific content for our educational resources.
COMSEP	Set national curricula.
ISONG	ISONG is a global nursing specialty organization dedicated to fostering the scientific and professional growth of nurses in human genetics and genomics worldwide. It provides a forum for education and support for nurses providing genetic health care.
NAPNAP	NAPNAP seeks to educate its members through our national conference, bi-monthly journals and local chapters. Genetics education and training has become an important part of these educational efforts.
NCHPEG	NCHPEG’s mission is to promote health professional education and access to information about advances in human genetics to improve the health care of the nation. NCHPEG fulfills this mission by: <ul style="list-style-type: none"> • integrating genetics content into the knowledge base of health professionals and students of the health professions, • developing educational tools and information resources to facilitate the integration of genetics into health professional practice, and strengthening and expanding the Coalition's interdisciplinary community of organizations and individuals committed to coordinated genetics education for health professionals.
NSGC	<ol style="list-style-type: none"> 1. NSGC provides continuing education for Genetic Counselors through a variety of educational activities including conferences and online educational offerings. NSGC is also the provider of continuing education credits through the IACET for genetic counselors. 2. NSGC maintains a speakers’ bureau to facilitate connections between genetic counselors and the public, including other healthcare providers, for the purpose of promoting genetic education. 3. NSGC has recently created a healthcare providers section of their website with the goal that it will contain helpful educational resources for a variety of providers. 4. NSGC develops brochures for patient and provider education on specific genetics topics. 5. The NSGC president attends the NHGRI sponsored “Physician Assistant and Genomic Medicine” meeting annually for the purpose of providing input and assistance to PA (and now nursing) training programs as the move towards integrating genetics/genomics competencies into their curriculum. Past President Angela Trepanier is a member of an advisory committee that has developed out of this meeting to create a web-based repository of educational resources for PA and nursing programs. 6. NSGC is a member of NCHPEG and regularly sends a representative to the national meeting.
ONS	One of the topics in the ONS Strategic Plan for 2009-2012, is biology and cancer and emerging trends in diagnosis and treatment. Genetics is a big force in these two areas and needs.
PAEA	Helping to provide teaching resources to faculty of PA programs is one role of our organization. Advocacy of certain educational issues is another responsibility—and genetics has been seen as an area of importance.
STTI	Planning for free online repository available to health professionals for content, tool kits, etc., related to genetics through the Virginia Henderson International Library.

Table 3 Organization Currently Able to Fulfill Role or Responsibility (Survey Question 5b)

Organization	Response
ACHDNC	At present no appropriations for this section of its authorizing legislation.
ANA	Partially
ACP	We are able to develop programs and products related to genetics education. The difficulty is getting members to be interested in them.
AAFP	Yes: AAFP is able to provide guidelines for residency training and incorporate

	genetics/genomics into ongoing CME activities.
ONS	Currently we have an online genetics course that covers genetics in great depth. We also discuss genetics and its impact on diseases in some of our other projects as well. For example: hereditary breast cancer and pharmacogenomics
STTI	Discussions in progress with Dr. Jean Jenkins (NIH/NHGRI and ISONG) regarding content. STTI has the resources to implement.
ACCP	Yes, we have an online course in Pharmacogenomics that is free from our website, www.ACCP1.org .
AAP	The AAP is currently and actively engaged in this activity. It seems implausible to suggest that any single organization could “fulfill” the role of educating 60,000 pediatricians on the topic of genetics.
NAPNAP	Somewhat – there is such a large amount of information in all areas of pediatrics that it is difficult to spend sufficient time on each area.
NCHPEG	Yes, genetics education is all we do.
ASHG	Yes, and we are expanding. Currently have 2 professionals on staff and plan to expand. Also writing grants to support activities.
ACMG	To a somewhat limited degree we are able to fulfill some of this. We offer annual meetings and other educational opportunities to our members. However, this is a limit to how much we are able to do through practice guidelines and clinical decision support tool development due to the breadth of conditions with significant genetic components and the very large number of rare disease genes in which we currently provide testing and service. These are numbered in the thousands.
NSGC	NSGC has been very successful in providing educational opportunities for genetic counselors and in participating with NHCPEG and the PA group we are still enhancing our ability to provide resources on our website to other healthcare providers and to market our speaker’s bureau. This is a major initiative for NSGC in 2009.
AACN	Yes; AACN recently revised the Essentials of Baccalaureate Education for Professionals Nursing Practice, which contains competencies and content related to genetics and genomics. Conference programming for faculty development related to genetics and genomics.
PAEA	Their willingness and commitment (financially) to sponsor a representative to NCHPEG from an organizational standpoint helps PAEA fulfill this responsibility.
AMA	We are able to fulfill this role in a limited way. There is only one staff member working on genetics full time, so we are at about the maximum in terms of genetics resources/education programs/policy involvement that can be achieved without additional staff.
AAPA, ACOG, ACPM, APHMG, AWHONW, ABGC, ABMG, ISONG, answered only “yes” to this question	

Table 4 Strategies for Organizations to Fulfill their Role in Genetics Education More Effectively (Survey Question 5c)

Organization	Response
PAEA	There is somewhat of a sense that one person with some expertise in genetics is supported and reports back to the organization, but the dissemination of genetics education information is not taken much beyond that at this point.
AAFP	AAFP would be able to provide our members with more opportunities to learn about genetics and genomics with more funding dedicated to that specific purpose. Though there are many grants that have become available for genetic research, there are fewer opportunities available for obtaining funding for education and training purposes. A cooperative agreement related specifically to genetic/genomic education could be one way in which this could happen. For instance, AAFP has a cooperative agreement with the CDC related to immunizations with the

	<p>goal of increasing immunizations in private practice.</p> <p>Another way in which AAFP might be able to more effectively educate our members regarding genetics is through partnership with groups such as the Evaluation of Genomics in Prevention and Practice (EGAPP) Working Group. It might be helpful to create a partners group that works with EGAPP to learn their process for making recommendations in order to best communicate these recommendations to our members.</p>
ACCP	Grant money provided to do so would help
ACP	Yes, if we could generate greater interest.
ACPM	No. ACPM has a long history of developing high quality professional education and has brought together leaders in the field to guide this effort as it has done with other successful efforts in the past.
AMA	Additional staff is needed to provide additional programs and resources. This may require obtaining grants or contracts.
ANA	With more funds we could partner with other organizations or work independently to provide education, specifically the dissemination and utilization of the Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics (2006).
ACMG	Different problems are apparent at each level of education in which we are involved, including medical students and residents in other specialties limited in their exposure to those who practice medical genetics. Further, trained medical geneticists are not available in about 25% of medical schools. Funding to support the development of educational materials including case based learning modules and others educational modules would be most helpful. Funding to support development of practice guidelines and clinical decision support tools would significantly improve our ability to address these issues for practicing physicians and would build the underlying infrastructure to integrate these materials into the evolving electronic health system world.
ASHG	We are working on engaging our broad membership more effectively, as in the Genetics Educator Outreach Network (GEON) now including several hundred geneticists ready to engage in community activities.
APHMG	If professor groups had more support from medical schools for education, there would be more resources for education.
ISONG	Additional funding would enhance development of genetic teaching materials and educational programs.
NCHPEG	Because we are a coalition of organizations representing practical health professionals, one of our aims is to be responsive to needs in an effective and efficient manner. We would benefit from funding streams that have enough flexibility to allow us to respond to the educational needs of our members proactively rather than waiting to respond to RFPs. Specifically, we see needs in determining the best approaches to educating about complex topics and evaluating the effectiveness of educational programs using clinical outcomes or their proxies. At this point NCHPEG and other groups engaged in genetics education would benefit from stepping back from development to critically assessing our approach and effectiveness of the programs. With better understanding of the educational process in genetics, we can tackle the complex educational topics more effectively.
NSGC	With more resources we could utilize the skills of an instructional designer to enhance the resources currently available on our website. It would also be helpful to have access to data from a needs assessment for various healthcare providers (as well as the public) to determine what resources we should prioritize developing. Such a needs assessment would also help us determine how to most effectively market our speaker's bureau.
NAPNAP	Perhaps more focus on genetics at annual conferences and in a journal.
ONS	We continue to look for ways to increase genetics knowledge for nurses as this is not a subject matter covered in any great detail in most colleges and universities.

STTI	Verifying that the key players are involved in the process.
ABGC	None recognized at this time.
ABMG	Evaluation is of training programs is by web based forms as on-site evaluation is too expensive and complex for a small organization.
AACN	Additional grant money to assist with faculty development related to genetics and genomics.
COMSEP	We meet yearly and update the curriculum every few years.
AAPA	We are always looking for partnerships with other organizations to bring quality education materials to physician assistants.
ACHDNC	Non-applicable

Table 5 Committees, Workgroups, or Dedicated Staff for Genetics or Genomics Education (Survey Question 11)

	All Organizations		Genetic-Specific Organizations		Nongenetic Organizations		Federal Advisory Committees	
	%	#	%	#	%	#	%	#
Yes	47	17	78	7	36	9	50	1
No	47	17	22	2	56	14	50	1
Not sure	6	2	0	0	8	2	0	0
Total	36		9		25		2	

Table 6 Importance the Organization Places on the Development and Promotion of General Health Education Activities (Survey Question 6)

Organization Type	Not at all Important				Very Important		Total Responses
	1	2	3	4	5	NA	
Federal Advisory Committee	-	-	1	-	1	-	2
General Professional Organization	-	-	-	1	17	-	18
Genetic-Specific Organization	-	1	1	-	6	-	8
Professional Education Organization	-	-	-	-	6	2	8
All Organizations	-	1	2	1	30	2	36

Table 7 Importance the Organization Places on Development and Promotion of Genetic Health Education Activities (Survey Question 7)

Organization Type	Not at all Important				Very Important		Total Responses
	1	2	3	4	5	NA	
Federal Advisory Committee	-	-	1	-	1	-	2
General Professional Organization	1	1	4	5	4	2	17*

Genetic-Organization	-	-	-	-	8	-	8
Professional Education Organization	-	1	1	-	4	2	8
All Organizations	1	2	6	5	17	4	35*

*One General Professional Organization did not respond to this question.

Table 8 Priority Level of Genetics Education within Organization (Survey Question 8)

Organization Type	Low Priority			High Priority			Total Responses
	1	2	3	4	5	NA	
Federal Advisory Committee	1	-	-	-	1	-	2
General Professional Organization	1	3	6	3	2	2	17*
Genetic-Specific Organization	-	-	-	1	6	1	8
Professional Education Organization	1	1	2	1	1	2	8
All Organizations	3	4	8	5	10	5	35*

*One General Professional Organization did not respond to this question.

Table 9 Extent to Which Organization's Membership is Satisfied with Current Emphasis on Genetics and Genomics Education (Survey Question 9)

Organization Type	Not at all Satisfied				Extremely Satisfied			Total Responses
	1	2	3	4	5	NA		
Federal Advisory Committee	-	-	-	-	-	2	2	
General Professional Organization	-	-	4	6	3	4	17*	
Genetic-Specific Organization	-	-	-	3	3	2	8	
Professional Education Organization	-	-	4	1	1	2	8	
All Organizations	-	-	8	10	7	10	35*	

*One General Professional Organization did not respond to this question.

Table 10 Proficiency of Organization’s Leadership with Genetics and Genomics Education (Survey Question 10)

Organization Type	Low Expertise/ Comfort			High Expertise/ Comfort			Total Responses
	1	2	3	4	5	NA	
Federal Advisory Committee	-	-	1	-	1	-	2
General Professional Organization	1	3	2	7	2	2	17*
Genetic-Specific Organization	-	-	-	1	7	-	8
Professional Education Organization	-	2	3	1	-	2	8
All Organizations	1	5	6	9	10	4	35*

*One General Professional Organization did not respond to this question.

Table 11 Barriers to Providing Genetics Educational Activities (Survey Question 12)

Barriers	Number of Organizations Selecting Each Barrier				
	All Organizations	Genetic-Specific Organizations	Professional Education Organizations	General Professional Organizations	Federal Advisory Committees
A. Not Applicable	4	0	2	2	0
B. Organization Lacks Knowledge	4	0	1	3	0
C. Competing Priorities	18	1	4	12	1
D. Lack Educational Resources	8	3	2	2	1
E. No Need for Certification	12	4	1	6	1
F. No Evidence of Effectiveness	4	1	0	2	1
G. Other *	9	4	0	5	0
H. No Barriers	5	1	3	1	0

Table 12 Barriers Ranked in Order of Importance (Survey Question 13)

Barriers	Ranked Order of Importance (Based on 25 Responses)							
	1st	2nd	3rd	4th	5th	6th	7th	8th
A. Not Applicable	3	0	0	1	0	3	0	0
B. Organization Lacks Knowledge	0	0	2	2	4	0	0	0
C. Competing Priorities	10	1	2	1	0	0	0	0
D. Lack Educational Resources	4	5	3	1	0	0	0	0
E. No Need for Certification	1	10	1	1	1	0	0	0

F. No Evidence of Effectiveness	2	2	2	0	0	0	1	0
G. Other *	3	2	0	0	0	0	0	0
H. No Barriers	0	0	1	0	0	0	0	0

*Other Barriers Listed by Respondents:

- Lack of funding
- Lack of staff/corporate support
- Oncology nurses do not see the importance in having genetic knowledge
- Limited pool of pediatric geneticists
- Unable to educate other health care professionals
- Lack of understanding about what health care providers specifically need to know about genetics (which is quite different from what a genetic professional needs to know)

Table 13 Organization Survey Results or Input from Members Regarding Genetics and Genomics Education (Survey Question 16)

Was Membership Surveyed?	Number of Responses
Yes	21
No	12
No Answer	3
Organization	Response
AACN	<p>AACN’s partners from NHGRI and NCI presented talks to three groups of nurses December 2008 and utilized Turning Point Software and voting keycards to gather data utilizing specific questions based on constructs of the transtheoretical model and stages of change (Prochaska, J., Redding, C., & Evers, K., 2002).</p> <p>Group 1: those attending (only faculty) an American Association Colleges of Nursing Meeting in Texas 12/5/08 Session 1 (n= maximum response 57)</p> <p>Group 2: those attending (only faculty) an American Association Colleges of Nursing Meeting in Texas 12/5/08 Session 2 (n= maximum response 56)</p> <p>Group 3: those attending (mixed group-faculty, practicing nurse, educator, researcher, student) a Sigma Theta Tau International (STTI) Regional Meeting in NY 12/7/08 (n= maximum response 54)</p> <p>We collapsed data from the three groups together summarizing total responses to the questions (see Table 1 for complete results; Max n= 161). A brief summary of highlights:</p> <ol style="list-style-type: none"> 1. The majority of attendees had a masters (45%) or doctoral degree (46%) 2. For the STTI meeting only: there were (38%) faculty and (33%) practicing nurses attending. 3. The majority had heard that Genetics/Genomics (G/G) was recommended in the Baccalaureate Essentials to be included in nurse preparation (88%). 4. The majority rated their own personal G/G knowledge as low (48%). 5. The majority felt that preparing nurses to use G/G information was an important role for nurse educators (39% agree; 44% strongly agree). 6. The majority were motivated by healthcare advances to learn more about G/G (40% agree; 42 % strongly agree). 7. Less were motivated by the academic environment to learn more about G/G (33%

	<p>agree; 38 % strongly agree).</p> <ol style="list-style-type: none"> 8. Most were likely to include G/G in nursing courses they teach (45% very likely). 9. Most intended to adopt curriculum/course changes to include G/G within the next six months (60%). 10. Most felt positive (49%) about the Baccalaureate Essentials including G/G in nurse preparation with 32% feeling extremely positive. 11. The majority (68%) strongly agreed it was time to start teaching the next generation of nurses about G/G. 12. Regarding relevancy: 41% strongly disagreed that genomics is unlikely to become relevant for nursing practice in the next 12 months; and 75% strongly disagreed that genomics is unlikely to become relevant for nursing practice in the next 5 years. 13. The majority (36%) strongly disagreed that the promise of G/G for healthcare had been exaggerated. 14. The majority (61%) strongly disagreed with the statement “there are no changes I can make in nursing curriculum/courses to make room for G/G”. 15. The majority (73%) strongly disagreed with the statement “family history with G/G content has little value for patient care”. 16. The majority (71%) strongly agreed that teaching nurses G/G is important to keep them as a central partner in patient/family care. 17. Most (36%) reported they needed web-based toolkit resources to include G/G in the courses they are teaching; and model curricula (34%) was a close second. 18. In terms of priorities of curriculum concepts to include in nurse preparation, G/G (7%) was fourth out of eight items with patient centered care identified most (34%). <p>Conclusions from data: Nursing faculty are aware of the incorporation of G/G into the Baccalaureate Essentials. They currently feel G/G is relevant to nursing practice. Most intend to adopt curriculum/course changes to include G/G within the next six months and would value resources such as web-based toolkit materials and model curricula.</p>
ANA	There were follow on activities and plans from the competency working group.
NCHPEG	From discussions with our membership, we know that there is a need for focused, relevant information that is available at the point of care. We also know from our own experience, as well as the literature, that health care professionals, in general, lack understanding of genetics concepts, such as those involved with taking and interpreting a family history and interpreting genetic test results. This deficit has been linked to a lack of confidence in discussing genetic conditions and risks with patients. Although there are efforts within some of our member organizations (nurses, Pas) to focus attention and educational efforts on genetics and genomics through the creation of genomics competencies, this emphasis is lacking in many of our other member organizations. NCHPEG has recently revised our Core Competencies for All Health Care Professionals, by streamlining them based on the experience of our membership. We are also in the final stages of review before releasing Core Competencies in Family History; the associated slide set that covers core content and case examples is currently available. Professional organizations have, in the past, used these resources to structure their own specific competencies (e.g., public health professionals and nurses).
AMA	Limited input. Following the educational session at the 2006 Interim Meeting, evaluation forms asked whether the participants would like to have more genetics programs offered to them, and the response was overwhelmingly yes. Also, the AMA House of Delegates adopted policy in 2006 that directs the AMA to be involved in the development of educational materials to assist physicians with genetics-related practice issues. (D-460.976, AMA Policy Database). There has not been a broad survey of AMA members about genetics education needs. The AMA did partner with Medco to survey physicians about their knowledge and use

	of pharmacogenomics. Close to 90% of respondents (approximately 10,000 respondents) indicated that they did not have adequate information about genetic test availability and application in therapy.
ACGME	Program requirements for each specialty and subspecialty undergo review/revision every 5 years. As part of the review/revision process, the RRCs seek input from the constituents about priorities for all topics related to the disciplines under review.
AAFP	The most recent feedback from our members regarding genetics education needs comes from our 2008 survey asking members about clinical topic areas in which they need more education. Some topics they rated at near or below the mean for medical knowledge and skill that are related to genetics include: allergies, sickle cell disease, developmental/behavioral disorders, degenerative disorders and Alzheimer's. Based upon this survey, there is an indication for an educational need in these areas. In addition, a prior survey, the 2003 CME Topics Survey, asked about genetic needs specifically. This survey was mailed to 4000 members with a total response rate of 14.3%. 476 respondents' ranked genetics (in general) as a high (18.3%), moderate (43.7%) or low (38%) priority, respectively.
ACMG	The nature of our business is such that our members provide us with information as to the aspects of genetics, genetic diseases and genetics services that they most want included among our educational offerings.
ASHG	Since genetics and genomics are our business, we focus on educating our members about educational principles, methods, and are gathering information on misconceptions that can be dispelled by our members in other forums
ISONG	We are in the process of developing a needs assessment to better meet needs of the membership; we have surveyed the membership for a Genetics Nurse brochure in preparation for revision and translation (Spanish, Portuguese, and Japanese).
ONS	We have not specifically surveyed the membership on genetics. We do still have those members that choose genetics as an area that they want more education.
COMSEP	We have occasionally surveyed the membership about their attitudes toward different content areas including genetics.
ACCP	A needs assessment survey for our upcoming annual meetings. They want education on this topic.
ACOG	Is an important area and routine part of obstetrics and gynecologic care
AACP	See report of 2004 work by Walif and McKay being repeated in 2009
AWHONW	There have been several board of director discussions and it has been included in the strategic plan.
ACPM	We've asked about topic preferences for CME offerings and this was on the list.
NSGC	Conducted member focus groups at the end of 2008 to solicit information on member needs related to the future of genetics and genomics. Much of this input focused on the need for education related to the changing roles of genetic counselors as educators and translators of information as full genome sequencing, pharmacogenomics and other advances in technology lead to an increase in the genetic information available to consumers and healthcare providers. NSGC also requested input regarding education needs and priorities from the attendees of the 2008 NSGC Annual Education Conference and the response was similar to that of the focus groups. NSGC will be distributing a survey to our full membership in early 2009 to further prioritize our strategic and education focus areas for the short and long-term. NSGC is also planning to conduct a membership survey in the summer of 2009 to gain further input from our membership regarding educational needs and priorities.
ABMG	Work with training programs to update training areas to include more modern knowledge
AHDNC	Organizational presentations
AAAA	In 2007 we surveyed 113 PA leaders on several aspects of genetics. We asked them to rate, on a scale of one to ten (least important to most important), the importance of genetics in the

	<p>education of PAs. The mean score was 7.32. More experienced PAs rated it higher in importance. We also asked them to rate the most useful formats for learning about genetics. The most useful methods were electronic, a course with printed materials, printed self study guides and 1-2 day courses. Least useful were lunch conferences, one week courses and informal consultations.</p> <p>In 2008 we surveyed 1800 PAs on their educational needs. When offered a list of nine Topic areas which they might be interested in attending review courses, “Genomics and genetics” was ranked as the least valuable. 28% of PAs rated it as Valuable or Very Valuable and 72% rated it as Less Valuable or Not Valuable.</p>
PAEA	<p>From the abstract:</p> <p>Results: 100 programs responded, for 75% response rate. Eighty-one percent of responding programs expressed the need to enhance the quality and extent of genetic and molecular medicine in their curricula, and 62% planned to make changes in the near future. Genetics is delivered with an assortment of curriculum models and taught with a rich variety of methods. It was surprising to find some salient concepts not being taught in 12-16% of programs, e.g., molecular biology, Mendelian and non-Mendelian inheritance, pedigree structure, and genetic diagnostic testing. Pharmacogenomics is taught in only a quarter of programs. PA program core facility plays a major role in teaching genetics. PA program respondents indicated the following to be particularly helpful for genetics curriculum development: (1) centrally developed resources, (2) genetic case studies, and (3) up-to-date valid resources on the Web for educators and students. Conclusions: PA programs clearly see the importance of genetics in PA education, but have expressed the need for more resources to further develop their curricula. It is hoped that this survey will provide baseline data that will aid in the development of competencies and curricula components in the future.</p>
	15 organizations reported either no survey or did not respond to the question.

Table 14 Programs to Enhance Engagement in Genetic and Genomic Education/ Programmatic Needs to be Addressed by the Federal Government (Survey Question 17)

Organization	Response
AACN	As the data suggests, faculty are aware of the importance of G/G material. However, there is an urgent need to develop faculty knowledge related to this area. Grants and resources to assist with faculty development are needed.
ACOG	Confidentiality issues, lab issues, direct marketing of genetic tests to patients poses issues
AACP	Curricular resources, faculty training.
STTI	Evidence based content to apply in practice and education with some support from federal funding.
ACP	Financial support for additional live programs and development of enduring materials would be useful.
ACCP	Funding in the form of educational grants.
ACMG	Funding that allows us to bring genetics education to other specialty organizations is useful. They often ask us to help them develop programs related to genetics for their own meetings and to identify faculty as well.
ISONG	Funding to develop toolkits to integrate genetics into education, to copy/duplicate a handout book from ISONG pre-conferences.
AAFP	It would be helpful if there was federally supported research and a report that supported good evidence that implementation of genetics information in the clinical setting materially affected outcomes. We could then use this report to ideally engage our members’ interest in more

	genomic topics.
AMA	More opportunities for educational forums that could communicate the pace of genetic technology's entry into the clinical setting. More point-of care tools that could be a quick reference for physicians as they practice. More integration of genetics into clinical decision support, electronic medical records, and performance standards. Clarity around guidelines for use of genetic tests and evaluation tools for genetic tests that are currently available. As suggested by SACGHS in its report on the oversight of genetic testing, federal implementation of an agency that would develop and maintain a registry for genetic tests would be a first step in evaluating clinical validity and utility, which will likely be hurdles that genetic tests must clear before they are widely used in the clinical setting.
ACHDNC	Non-applicable-Secretary already engaged
ONS	No I don't think so
ABMG	None
COMSEP	Not necessarily. The organization understands that an understanding of genetics is important.
AOA	Online educational materials
ADEA	Our members could benefit from any and all forms of curricular and other educational materials related to genetics and genomics education, whether addressed by the Federal government or other entities.
ACGME	Our review/revision process works very well.
NSGC	Resources or programs that could bring together NSGC with other healthcare providers (like the PA Genomic Medicine Meeting) so that genetic counselors could get a better understanding of the informational/educational needs of these providers. Also, a needs assessment of different providers groups would help genetic counselors/NSGC in developing targeted, effective educational resources.
ACPM	The current CME program is a good way to get our members engaged in this area. However, this was made possible by an industry grant, and even though we followed CME rules to a "T", perceptions of bias could be present. Federal funding for more of these types of programs would be welcome.
NCHPEG	The main programmatic need from NCHPEG's perspective is for a genetics/genomics education agenda to inform the development of RFPs and other government initiatives. Bringing the significant players in genomics education together to discuss the potential agenda could help to define aims, identify gaps, reduce redundancy in effort, and promote collaboration to build off existing efforts. A well-informed education agenda could promote further understanding of the best approaches to education and support the effective development of new content while reducing redundancy of effort. The resources needed to support such efforts include those that would allow us to bring together a diverse group of educators and researchers in adult education to provide insights into the most effective adult education strategies for educating about complex topics when the desired outcome is behavior change. This group should include distance education providers, communications experts, evaluators, and commercial organizations to broaden the discussion and provide a broad base of research and experience in adult education. The discussion would need to consider best approaches to education delivery for health professionals given the vast array of teaching mechanisms that are available (e.g., traditional web-based, web 2.0, podcasts, traditional CME, interactive CME, etc). For any future efforts in genomics education, mechanisms for long-term funding are necessary to allow for evaluation the effectiveness of educational efforts by clinical outcomes or their proxies.
ABGC	There are a few federal resources that could benefit the genetic counseling profession by enhancing greater educational efforts, although these would not directly affect our organization. (1) Recognizing that 55% of genetic counselors are actively engaged in genetics/genomics education invest in expanding or developing genetic counselor training

	programs; (2) provide grants such as Special Projects of Regional and National Significance (SPRANS) specifically geared toward providing genetic/genomics education; and (3) require all federally funded healthcare training programs to incorporate genetic/genomics education within their curricula.
SGIM	Uncertain. We've been successful in having either a 1/2 day or full day pre-course and a workshop on genetics at every annual meeting since 1999
ANA	We would like to partner with others to implement use of the Competencies developed in 2006.
AAPA	PAs are required to obtain 100 hours of continuing medical education every two years and recertify by examination every six. This means they are highly motivated to obtain CME which is not only relevant to their practice but also relevant to the generalist recertification exam. Dynamic, interactive and relevant CME programs on genomics are the best way to engage them. We have worked with several Federal agencies to offer sessions at our annual conference. Continued support by those agencies is important to us.
PAEA	The types of resources made available by NCHPEG worked well for our train the trainer workshop as well as the website for PAs that NCHPEG sponsors. Development of modules for various genetic educational aspects of clinical medicine that could be shared on the Internet would be welcomed by faculty and patients alike.
ASHG	We have one NSF grant (STEM grant) and have submitted another, but focus on what the genetics professionals can do to educate other would be helpful.

Table 15 Completed Initiatives/Programs Implemented by Organization (Survey Question 14)

Organization	Program or Initiative
NCHPEG	<ul style="list-style-type: none"> • Competencies in Family History (2008) • Core Competencies for All Health Care Professionals (2007) • Genetics in the Physician Assistant's Practice (2007) • Genetics in the Practice of Speech-Language Pathology and Audiology (2006) • Principles of Genetics in the Context of Common Disease (2006) • Genetics and Common Disorders: Implications for Primary Care and Public Health Providers (2005) • Core Principles in Genetics (2004) • Genetics, Disease, and Dentistry (2004) • Genetics and Major Psychiatric Disorders: A Program for Genetic Counselors (2002) • Genetics Is Relevant Now: Nurses' views and patients' stories • Race, Genetics, and Healthcare • Survey of Genetic Consumers • Newsletters
AACN	<ul style="list-style-type: none"> • Partnered with the National Human Genome Research Institute (NHGRI) and National Cancer Institute (NCI) on several initiatives <ul style="list-style-type: none"> i. Assisting with creating and endorsing the <i>Essential Competencies and Curricula Guidelines for Genetics & Genomics</i> (2005) ii. Assisting with creating a tool kit for faculty development iii. Assisting with creating a tool kit repository • Recently revised the <i>Essentials of Baccalaureate Education for Professional Nursing Practice</i>, which contains competencies and content related to genetics & genomics • Conference programming for faculty development related to genetics and genomics

AAP	<ul style="list-style-type: none"> • The AAP’s annual National Conference and Exhibition (NCE) is the Academy’s premier educational event. CME-bearing presentations on genetics topics have been included each year. • AAP regional educational programs, as well as programs sponsored by the independently-chartered state-based AAP chapters, have also included genetics educational programming. • A special, multi-disciplinary, ad-hoc writing committee was formed to develop a new AAP Clinical Report entitled, “Newborn Screening Expands: Recommendations for Pediatricians and Medical Homes—Implications for the System.” The resultant document can be accessed here: http://aappolicy.aappublications.org/cgi/content/full/pediatrics;121/1/192
ACMG	<ul style="list-style-type: none"> • ACMG Annual Meeting (CME approved) • ACMG Basics: Genetics for Providers– A CD Rom-based Educational Program • Review of USMLE Steps 1-3 (see National Coordinating Center) • Clinical decision support tools for primary care providers for newborn screening (ACMG ACT Sheets)
STTI	<ul style="list-style-type: none"> • <i>Nurse’s Advantage</i> book collection on genomics in nursing, 2007. • 16 Scholarly Publications • 6 Continuing Education Offerings • 5 presentations at major conferences • 4 Online Publications
AMA	<ul style="list-style-type: none"> • Brochure on warfarin pharmacogenomics • Pharmacogenomics and Personalized Medicine CME • Survey of physicians about knowledge and use of pharmacogenomics • Arranged for 2008 NCHPEG annual meeting to be CME eligible • Educational Session for AMA Interim Meeting, 2006 • Future Perfect: Conversations on the Meaning of the Genetics Revolution • CME monograph on Risk Assessment for Hereditary Cancers • Family Medical History in Disease and Prevention • AMA Council/Board reports on genetics subject
ACOG	<ul style="list-style-type: none"> • Develop clinical guidelines and patient resources • Continuing education
ACP	<ul style="list-style-type: none"> • Published a book entitled <u>Case Studies in Genes and Disease: A Primer for Clinicians</u> • Co-sponsored an annual genetics course with the Genetics Division at Brigham and Women’s Hospital.
NSGC	<ul style="list-style-type: none"> • NSGC Twenty-Seventh Annual Education Conference • NSGC 2008 JGC CEU Program • NSGC 2008 Online Course
ISONG	<ul style="list-style-type: none"> • Genetics and Genomics Nursing: Scope and Standards of Practice • Survey: What is a Genetics Nurse? • ISONG collaborated with the development of <i>The Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics</i> and a number of ISONG members served in key roles in the development of this document, including the primary individuals involved. • 2008 ISONG Conference: Nursing Revolution in Genomics: Uniting Research, Education and Practice • 2007 ISONG Conference: Global Issues in Genomic Healthcare • 2006 ISONG Conference: Genomic Healthcare: The Future is Now

ACGME	<ul style="list-style-type: none"> • The ACGME Program Requirements for Graduate Medical Education in Medical Genetics and the ACGME Program Requirements for Graduate Medical Education in Molecular Genetic Pathology underwent major revisions in 2007. • In 2007, the ACGME approved that the RRC for Medical Genetics could begin accrediting programs in the fellowship of Medical Biochemical Genetics. The RRC developed ACGME Program Requirements for Graduate Medical Education in this subspecialty.
ACPM	<ul style="list-style-type: none"> • Sessions on Genetics at Preventive Medicine Annual Meetings • Symposium: Filling the Gap in Preventive Patient Care: Genomics in Practice
ONS	<ul style="list-style-type: none"> • Genetics Short Course Grant • Genetics Online Education Series
AAFP	<ul style="list-style-type: none"> • Annual Clinical Focus (ACF) 2005: Genomics • Articles and commentaries in AAFP-related publications
ANA	<ul style="list-style-type: none"> • <i>Genetics and Ethics in Health Care: New Questions in the Age of Genomic Health</i>. Rita Black Monsen, DSN, MPH, RN, FAAN. 2008. • ANA partnered to start NCHPEG and is an active member of ISONG • Links to educational programs and resources posted on our website
NAPNAP	<ul style="list-style-type: none"> • At least one session at each annual conference on genetics • Journal articles fairly often on genetics topics
AAPA	<ul style="list-style-type: none"> • Daily Dose of DNA • PA-specific website for case-based genetics education
ACCP	<ul style="list-style-type: none"> • Free, non-credit, Online Pharmacogenomics Course consisting of 13 modules • Pre-meeting workshop, and two symposia with the 36th annual meeting, September 16-19, 2006, on topics of genomics
PAEA	<ul style="list-style-type: none"> • National Survey of PA programs regarding genetics curricula • Proposed genetic/genomic competencies for PAs • Ideas to help faculty members organize genetics curricula according to the aforementioned competencies • Train the trainers for PA faculty
SGIM	<ul style="list-style-type: none"> • Pre-courses on genetics as part of annual meeting • Workshops on genetics as part of annual meeting
AACP	<ul style="list-style-type: none"> • Argus Commission: Science in 2029 • Miscellaneous educational sessions at AACP meetings
ABMG	<ul style="list-style-type: none"> • Maintenance of Certification

Table 16 Ongoing or Planned Initiatives/Programs Implemented by Organization (Survey Question 15)

Organization	Program or Initiative
NCHPEG	<ul style="list-style-type: none"> • Genetics for Social and Behavioral Researchers • Genetics and Colorectal Cancer • Access to Credible Genetics Resources Network • Genetics Education: Resources for Health Professionals
AACN	<ul style="list-style-type: none"> • Continued work on tool kit and tool kit repository
AAP	<ul style="list-style-type: none"> • CME-bearing genetics-related programming will continue to be scheduled during the AAP's annual NCE, regional events, and chapter-based activities. • The AAP's Division of Children with Special Needs, working in cooperation and

	<p>under contract with the American College of Medical Genetics, recently completed the pilot year for its Visiting Professorship Program. The program's goal are to: 1) increase knowledge of genetics in the medical home at the point of care, 2) support pediatricians in addressing their patients' genetics-related questions, and 3) facilitate linkages between the AAP Chapters and the regional genetics and newborn screening service collaborative.</p> <ul style="list-style-type: none"> • The AAP National Center for Medical Home Initiatives includes educational materials and tools related to genetics and newborn screening. http://www.medicalhomeinfo.org/screening/newborn.html
ACMG	<ul style="list-style-type: none"> • Revision of Medical Genetics Board training curriculum • Expansion of ACT Sheets to include genetic testing and family history tools for conditions likely to be referred by primary care.
APHMG	<ul style="list-style-type: none"> • Program Directors Group • Medical School Course Directors Group • Question Bank: bank for questions appropriate for medical student courses, as well as in service exams for residents.
STTI	<ul style="list-style-type: none"> • Plan to continue CE offerings, publications and conference meeting discussions related to genetics.
AMA	<ul style="list-style-type: none"> • Genetics of Colorectal Cancer CME • Education module for medical students on genetics of hereditary cancers • Brochure on pharmacogenomics • Educational piece for physicians on Direct-to-consumer genetic testing
ACOG	<ul style="list-style-type: none"> • Develop clinical guidelines and patient resources • Continuing education
ACP	<ul style="list-style-type: none"> • Co-publishing a new genetics text for clinicians with McGraw-Hill • Became a member of the National Coalition for Health Professional Education in Genetics (NCHPEG)
NSGC	<ul style="list-style-type: none"> • NSGC Twenty-Eighth Annual Education Conference • NSGC 2009 JGC CEU Program • NSGC 2009 Online Course • NSGC 2009 Regional Meetings (Chicago and Baltimore/Washington DC) • Genetic Counseling Foundation
ACPM	<ul style="list-style-type: none"> • CME program on Genetic Screening • Symposium: Filling in the Gap in Preventive Patient Care: Genomics in Practice
ISONG	<ul style="list-style-type: none"> • Needs Assessment of Membership • <i>Genetics and Ethics in Health Care: New Questions in the Age of Genomic Health</i>. Co-published by the American Nurses Association and ISONG • Collaboration with the National Society of Genetic Counselors (NSCG)
ONS	<ul style="list-style-type: none"> • The Genetics Online Education Series (as above) will continue
AAFP	<ul style="list-style-type: none"> • Communication to members about EGAPP recommendations • Ongoing CME activities
ANA	<ul style="list-style-type: none"> • Grant: "Development of Nursing New Media Education using Cancer Family History and BRCA1/2 Testing as a Paradigm"
AAPA	<ul style="list-style-type: none"> • National PA Organizations and NHGRI • Institutionalization of genomics in PA literature
ACCP	<ul style="list-style-type: none"> • Free, non-credit, Online Pharmacogenomics Course consisting of 13 modules • Several within the various slides provided
PAEA	<ul style="list-style-type: none"> • Though not initiated by PAEA directly, the genetics workgroup of PA educators is

	working collaboratively with nursing to produce a website for faculty and providers regarding genetics educational resources called Genetic/Genomic Resource Tool for Interdisciplinary Education (GERTIE)
SGIM	<ul style="list-style-type: none"> Genetics in Primary Care Internet Group
AACP	<ul style="list-style-type: none"> UCSD project on genomics curriculum

Table 17 Organizations Role in Health Professional Education (Category 1, Question 1)

Organization	Response
CGME	Recommend changes in the types of GME programs designed to assure an adequate supply of physicians to serve U.S. healthcare needs
ACP	We are the major national organization responsible for the lifelong education of internal medicine specialists and subspecialists. Our involvement spans the spectrum that includes medical students' education, graduate medical education, and continuing medical education.
ONS	The mission of the Oncology Nursing Society is to promote excellence in oncology nursing and quality cancer care.
ACCP	ACCP is an accredited sponsor of Continuing Medical Education and Continuing Pharmacy Education.
NAPNAP	NAPNAP is the main national organization for pediatric nurse practitioners. There are many specific initiatives for the membership to utilize in clinical practice – examples -initiative on obesity, mental health. Local state chapters also provide educational programs for the membership.
AOA	The AOA accredits CME, approves osteopathic intern and residency training programs, and provides other educational activities
AWHONW	Provision of standards and evidence based guidelines for the specialty of women's health and infants
SGIM	Most members serve as faculty in divisions of general medicine, or division of, residency program directors, dept. chair of internal medicine
NCHPEG	NCHPEG is a non-profit organization with the mission of promoting health professional education and access to information about advances in human genetics to improve the health care of the nation.
ASHG	Teaching and mentoring of basic and clinical genetics.
ACMG	We provide educational content for medical geneticists, primary care providers and other physicians.
ISONG	Provide a forum for education and support for nurses providing genetic/genomic healthcare.
AACN	AACN establishes the Essentials or standards for baccalaureate and higher degree programs in nursing education
ADEA	The American Dental Education Association represents all 57 dental schools in the United States in addition to 714 dental residency training programs and 577 allied dental programs, as well as the more than 17,000 faculty staff and students in these institutions. It is at these academic dental institutions that future practitioners and researchers gain their knowledge; where the majority of dental research is conducted; and, where significant dental care is provided.
COMSEP	We are primarily involved in the education of medical students, most specifically students in the clinical years but all aspects of undergraduate education remain important.
AACP	We are the association representing all colleges of pharmacy
ARCOG	Facilitate implementation of the goals and objectives of the American College of OB-GYN in residency training programs

PAEA	<p>PAEA’s mission is to pursue excellence, foster faculty development, advance the body of knowledge that defines quality education and patient-centered care, and promote diversity in all aspects of physician assistant education. To accomplish this mission, PAEA:</p> <ul style="list-style-type: none"> • Encourages and assists programs to educate competent and compassionate PAs • Enhances programs’ capability to recruit, select, and retain well qualified PA students • Supports programs in the recruitment, selection, development, and retention of well-qualified faculty • Facilitates the pursuit and dissemination of research and scholarly work • Educates PAs who will practice evidence-based, patient centered medicine • Serves as the defining voice on matters related to entry level PA education, nationally and internationally • Fosters professionalism and innovation in health professions education • Promotes inter-professional education and practice • Forges linkages with other organizations to advance its mission
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Table 18 Need for the Integration of Genetics and Genomics into the Training of Health Professionals (Category 1, Question 2)

Organization	Response
CGME	Because of competing priorities, the subject has not been taken up by our Council
ACP	We believe that trainees and physicians at all levels need to be better educated about this continually evolving field.
ONS	ONS has a position statement regarding the nurse’s role in cancer genetics. You can view this position statement by visiting the following link: http://www.ons.org/publications/positions/CancerGeneticCounseling.shtml
ACCP	It is a priority, but not necessary the top one.
NAPNAP	The importance of genetics/genomics has been recognized for a many years – NAPNAP has had a representative at NCHPEG for a number of years.
AOA	Important, but a low priority
AWHONW	Integration is the key word and the organization realizes the need.
SGIM	Not high because uncertain clinical benefit.
NCHPEG	The sheer volume of new information now at the disposal of biomedical researchers and health care providers is transforming our understanding of disease processes – including those of common, chronic diseases such as cancer, diabetes, and mental illness – and is changing the delivery of health care. Increasingly, health care providers – regardless of specialty, role, or practice setting – will face questions about the implications of genetics and genomics for their patients. And yet, the rapid pace of the science and the relative paucity of professional training in genetics continue to leave many clinicians without satisfactory answers to genetic questions from their patients. A prime example is the large number of genome-wide association studies that are finding genetic associations with a vast array of phenotypes. Some of this information is making its way into clinical care through direct-to-consumer marketing. Many health care professionals will be at a loss to interpret this information correctly, let alone determine whether management should be approached differently. While there are a number of ongoing and proposed efforts to help facilitate the appropriate translation of genomic information into the clinic, currently practicing health professionals would benefit from a greater understanding of the benefits and limitations of genetic information in the context of complex disease.

ASHG	We have become more integrated, but we don't want the central portions of genetics and genomics to become too "watered down" or so disseminated that genetics is unrecognizable except as a technology rather than an approach to medicine and science
ACMG	Genetics education needs to be widely distributed throughout health care with attention to the scope of practice of those being educated.
ISONG	We believe that the need for integration of genetics and genomics into the curriculum and training of health care professionals is a critical need. Although there are ongoing efforts to help prepare nursing faculty to integrate genetics and genomics into both undergraduate and graduate faculty (e.g., the Genetics Education Program for Nurses
AACN	AACN supports the integration of genetics/genomics into the curriculum of health professionals.
ADEA	There is a compelling need for the integration of genetics and genomics into the curriculum, and into the education and training of dental professionals.
COMSEP	Genetics is a major component of the core curriculum.
AACP	Considered essential and focus of regular programming
ARCOG	Each of the 225+ program implements knowledge through didactic lectures, journal clubs, and other scholarly activities and research
PAEA	Recognizing that genetics is playing a larger role in clinical practice, the challenge for PA educators is to prepare our students with an appropriate foundation of knowledge, skills and attitudes. A knowledge base of fundamental genetic principles and approaches is essential, as are the skills that will allow students to apply these principles in the clinic and to exhibit professional attitudes related to genetic information and diagnosis. This knowledge and skill set will be required for common clinical tasks, for example, when assessing familial risk for common diseases as well as ordering, interpreting, and explaining genetic test results to a patient. PA programs need to develop appropriate curricula to meet this dynamic challenge.

Table 19 Development of Curricular Components (Category 1, Question 3)

Organization	Response
CGME	Because of competing priorities, the subject has not been taken up by our Council.
ACP	All of our curricular offerings are optional – lives courses, content embedded within broader courses, and enduring materials that we develop.
ONS	Please see above for the current options in genetics that ONS provides. We also have a Genetics Clinical Resource Area on our website: http://www.ons.org/clinical/prevention/genetics/index.shtml
NAPNAP	No standardized genetics components but NAPNAP is a professional organization and not a professional nursing school so the members do receive curricular content of genetics in their educational programs.
AOA	Not required
AWHONW	Optional curriculum related to diseases in women, genetic testing for women and infants
SGIM	None by SGIM. The genetics in primary care faculty development curriculum or genetics through a primary care lens is used by educators.
NCHPEG	Please find the NCHPEG core competencies submitted and included in Appendix C-2.
ASHG	Individualized for PhD and undergraduate institutions. MD training falls under ACMG.
ISONG	These are clearly articulated in The Essential Nursing Competencies and Curricula Guidelines for Genetics and genomics, which are available at http://www.genome.gov/17517146 .
AACN	The revised Baccalaureate Essentials (2008) incorporates competencies and content related to

	genetics and genomics.
ADEA	Requirements and optional components of any area are at the discretion of our member institutions. We do not set curricular requirements.
COMSEP	<p>The organization wrote a national curriculum. Individual schools or directors may use the curriculum as they see fit. Here is the chapter on Genetics:</p> <p>Rationale</p> <p>A physician should be able to distinguish between congenital disorders (disorders present at birth) that are genetic from those that are nongenetic, as well as recognize common genetic diseases presenting later in childhood. Genetic abnormalities may produce congenital malformations, metabolic disturbances, specific organ dysfunction, abnormal growth patterns, and abnormalities of sexual differentiation. New technology and knowledge of genetics have raised ethical questions that physicians and society will need to address.</p> <p>Prerequisites</p> <ul style="list-style-type: none"> • Knowledge of gene structure, regulation and function • Basic knowledge of the Human Genome Project and the role of genetic inheritance in multifactorial diseases, such as cancer, heart disease and diabetes • Basic mechanisms of Mendelian inheritance, multifactorial inheritance, the “carrier” state, incomplete penetrance, variable expression, and spontaneous mutations • Basic embryology and teratology • Introductory history taking and physical examination skills <p>Competencies</p> <p>Knowledge</p> <ol style="list-style-type: none"> 1. Describe the genetic basis and clinical manifestations of the following syndromes, malformations, and associations: <ul style="list-style-type: none"> • Common chromosomal abnormalities, (e.g. Trisomy 21 (CP), Turner syndrome (CP), Klinefelter syndrome (M)) • Syndromes due to teratogens (e.g. fetal alcohol syndrome) (CP) • Other common genetic disorders (e.g. cystic fibrosis, sickle cell disease, hemophilia) (CP) • Single malformations with multifactorial etiology (e.g. spina bifida, congenital heart disease, cleft lip and palate) (M) 2. List common medical and metabolic disorders (e.g. hearing loss, hypothyroidism, PKU, hemoglobinopathies) detected through newborn screening programs (CP) 3. Discuss the effects of maternal health and potentially teratogenic agents on the fetus and child, including maternal diabetes and age (CP), alcohol use (CP) illicit drug use (CP), and prescribed medications such as phenytoin, valproate, and retinoic acid (M) 4. List common prenatal diagnostic assessments (e.g. maternal serum screening, amniocentesis, and ultrasonography) and understand their use (M) 5. Describe the use of chromosome studies in the diagnosis of genetic disorders (M) 6. Discuss the role of genetics in common multifactorial conditions (e.g. inflammatory bowel disease, pyloric stenosis, congenital heart disease, cleft lip, diabetes and cancer) (M) <p>Skills</p> <ol style="list-style-type: none"> 7. Use a family history to construct a pedigree (e.g., for the evaluation of a possible genetic disorder) (CP)
AACP	Not available info across all US colleges of Pharmacy, new survey in field
ARCOG	There is the Unit 7 Genomics produced by ACOG that we encourage to be incorporated into the program’s curriculum as a part of the overall ACOG curriculum.
PAEA	Genetics should be intergraded across the curriculum – and if organ systems are taught,

	should be emphasized where appropriate in each organ system. The “language” of genetics may need to be taught separately as a stand-alone course to complement the integration. A wide variety of teaching and evaluation methods, including didactic lectures, small group sessions with cases. Web-assisted interactive learning, and clinical experience, are likely helpful for adult learners. Teaching should be related to clinical cases and/or experiences whenever possible.
ACCP	Non-applicable

Table 20 Gaps in Genetics and Genomics Education (Category 1, Question 6)

Organization	Response
CGME	Because of competing priorities, the subject has not been taken up by our Council
ACP	Yes, The main problem is that clinicians have not yet broadly embraced their need to understand genetics/genomics and the application of these disciplines to the care of their patients.
ACCP	Unknown
NAPNAP	NAPNAP could be more structured in its inclusion of genetics in the educational materials and programs it provides.
NCHPEG	Although there are gaps in educational content, especially as genetics relates to common disease and with content geared appropriately for different health professions, there are greater deficits in our understanding of the most effective approaches to educating health professionals about genetics and the impact of educational programs on clinical practice. To address the gap related to educational approach, we need resources to bring together a diverse group of educators and those interested in adult education efforts. This group would provide insights into the most effective adult education strategies and delivery systems for educating about complex topics with a desired outcome of behavior change. Measuring the clinical impact of educational programs requires long term funding and innovative approaches to evaluation. NCHPEG is beginning to apply some different evaluation approaches, including simulated patients, but more work is needed in this area.
ASHG	There are gaps in every curriculum, and these are best addressed by specific professional groups.
ACMG	Funding is the greatest limitation to developing educational content for providers
ISONG	Yes, there continue to be gaps in genetics/genomics education. Many nursing programs do not have faculty with expertise in genetics/genomics. Increased access to online educational activities in genetics/genomics would help to decrease the gaps. Also, increased funding for genetics education would help.
AACN	Yes gaps exist and AACN is providing faculty development as part of its strategic plan.
ADEA	Probably
AACP	Yes- information lag of science translation to practice
PAEA	Yes – there are always gaps! Making genetics as clinically relevant as possible is critical – need more stories.

Table 21 Cultural Competency Incorporated into Curricula (Category 1, Question 4)

Organization	Response
ACP	Variably. We are trying to include more relating to cultural competency in the programming of our annual meeting.
NAPNAP	NAPNAP provides no formal curriculum to its members

AOA	Yes, it is required for postdoctoral education and certification
AWHONW	It is incorporated
SGIM	Yes
NCHPEG	One of the NCHPEG core competencies is the following: [health care professionals should know the various factors that influence the client’s ability to use genetic information and services, for example, ethnicity, culture, related health beliefs, ability to pay, and health literacy.
ASHG	We do address this on a regular basis
ACMG	Yes – required
ISONG	Yes, cultural competency is incorporated into the Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics. It is a required component
AACN	Yes, cultural competency is incorporated at all levels of nursing education and is required.
ADEA	Usually
AACP	Yes, per accreditation requirements, and it is required
ARCOG	It is supposed to be
PAEA	Yes, by our accreditation standards, cultural competency is a required component of PA curricula.
ACCP	Non-applicable

Table 22 Anticipated Future Needs in Genetics and Genomics Education (Category 1, Question 7)

Organization	Response
ACP	Because this is such a rapidly evolving field, the issue will be assuring that physicians have continually updated information and education re the application of genetics and genomics to patient care.
ONS	With oncology nursing education, we just need to keep up with genetics and how it relates to oncology. There is more and more each day on pharmacogenomics and how it can be utilized in oncology.
ACCP	More funding
NAPNAP	Will become more important for all pediatric health care providers to be knowledgeable about these topics
SGIM	Test interpretation, implication of genetic disorders for prevention and screening
NCHPEG	Need for increased evidence base to inform educational efforts and increase provider interest Proven strategies to increase uptake of practice recommendations Communicating risk effectively Interpreting genetic test results Knowing when, which patients, and to whom to refer Determining appropriate testing Understanding genetic risk for complex disease, including susceptibility testing Assessing risk using multiple tools (testing, family history, environment) Changing management based on risk New delivery models, incorporation into EHRs
ASHG	By being everywhere, we don’t want it to be nowhere!!!
ACMG	No end to need for education in genetics in sight. Expect expansion of electronic health system based clinical decision support tools.
ISONG	We expect the need for genetics/genomics education will continue to increase because of the escalating rate of genetic/genomic discoveries.
AACN	Continued tool kit development, conference programming, and faculty development
ADEA	That the need for incorporating genetics and genomics education into the curricula of dental

	professions will increase substantially.
AACP	We believe it will be mainstream education and clinical practice by then.
PAEA	More epigenetics, how the genomes of other entities interact with the human genome, increased pharmacogenomics. We'll see if that GWAS stuff pans out...

Table 23 Role of the Organization in Education, Training and Assessment of the Professional Workforce (Category 2, Question 1)

Organization	Response
AAPA	We assess educational needs and provide educational opportunities for physician assistants through our annual conference and other activities. We work with a variety of groups to advocate for the PA role and ensure PAs have the information they need to provide the highest quality of care to patients.
ANA	ANA has a broad role in each of these areas. The ANA is the only full-service professional organization representing the interests of the nation's 2.9 million registered nurses through its 53 constituent member nurses associations, its 23 organizational affiliates serving 330,000 members of national nursing specialty organizations, and its workforce advocacy affiliate, the Center for American Nurses. The ANA advances the nursing profession by fostering high standards of nursing practice, promoting the rights of nurses in the workplace, projecting a positive and realistic view of nursing, and by lobbying the Congress and regulatory agencies on health care issues affecting nurses and the public. Learning needs assessment, continuing education, publications and collaboration with other organizations directly contribute to fostering high standards of nursing practice.
ACPM	We provide support, advocacy and resources for professionals in both their graduate training and their Preventive Medicine practice stages of their careers.
ACOG	Develop practice guidelines and professional resources
AAFP	The AAFP helps to educate and train residents as well as physicians. This is done through providing residency program recommendations for residents. For physicians, the AAFP offers numerous CME opportunities.
AMA	AMA mission is to provide support to practicing physicians as they deal with important issues in medicine and health care. Some of that support is in the form of educational resources (other is in the form of lobbying and political advocacy). Educational resources are varied – a few genetics resources, but lots of other subjects as well. Ethics, financing your practice, approaches to treating different diseases/conditions, etc.
STTI	We educate members and nurses globally to improve health at point of care.
AAP	With regard specifically to genetics: The AAP Committee on Genetics (COG) studies and makes recommendations to the Board of Directors on recent advances in genetics and provides support to the chapters on state legislative issues as they related to genetics. The COG develops AAP Policy Statements, Clinical Reports, and Technical Reports on genetics related issues. A sample of topics includes: The Newborn Screening Fact Sheets; Clinical Genetic Evaluation of the Child with Mental Retardation or Developmental Delay, Prenatal Screening and Diagnosis for Pediatricians and Health Supervision Guidelines for NF, Down Syndrome, Fragile X, and other disorders. The Committee is in the early stages of developing a new manual on genetics for pediatricians. The Section on Genetics and Birth Defects (SOGBD) is focused on the education of primary care pediatricians; development of genetics-related educational programming for the annual AAP NCE; and supporting genetics-related advocacy efforts at the federal, state, and local level. The SOGBD also sponsors an annual Young Investigator Research Grant Award, which

	<p>offers a grant-in-aid of \$18,000 to encourage young investigators to explore opportunities in the realm of genetics and birth defects.</p> <p>Academy-wide: The AAP Department of Education (DOE) is responsible for the development, implementation, and evaluation of AAP Continuing Medical Education (CME) activities. These activities include CME courses (subspecialty/section courses, Practical Pediatrics and PREP The Course), the Future of Pediatrics Conference, and the National Conference & Exhibition (www.AAPexperieNCE.org).</p> <p>The DOE's Division of Workforce and Medical Education Policy guides the Academy in the formation of health policy related to the education, supply, requirements, demographics, and geographic distribution of general and subspecialty pediatricians. This involves managing a wide range of policy and regulatory issues for the Academy, including requirements for core and subspecialty residency education, the funding of graduate medical education, the efficacy of state-level incentive programs, the regulation of the pediatrician pipeline through federal legislation and appropriations, physician retirement patterns, and physician reentry into practice.</p> <p>The DOE's Division of E-Learning provides dynamic online learning resources for pediatric professional's growth. This mission is supported through the development of web-based, custom-tailored educational resource providing CME opportunities and services.</p> <p>AAP educational publications include: Pediatrics, the premier scientific publication in pediatric medicine; AAP News, the AAP's official monthly newsmagazine; AAP Grand Rounds, a monthly synopsis of the most pertinent articles to practicing pediatricians found in nearly 100 medical journals; NeoReviews; and Pediatrics in Review.</p>
APHMG	<p>Development of groups for Program Directors, Medical Course Directors, and the Question Bank are examples of our initiatives, As a Professors group for Human and Medical Genetics, our reason for being is to help fellow professors in all aspects of their work, of which teaching and education is a major part.</p>
NSGC	<p>The mission of the NSGC, as the professional organization of genetic counselors, is “to advance the various roles of genetic counselors in health care by fostering education, research, and public policy to ensure the availability of quality genetic services”. With regard to the educational objective specifically, the NSGC strives to keep abreast of changes in genetic-genomics, medical genetics/genomics, technology, and policy for the purpose of providing up-to-date, relevant continuing education for genetic counselors. Individual genetic counselors have historically been involved in providing medical genetics education to other healthcare professionals. In fact, our 2006 Professional Status Survey showed that over 60% of genetic counselors participate in educational outreach. The NSGC recognizes the importance of these efforts and has begun to develop organizational approaches to facilitating and promoting them (e.g., speaker's bureau, healthcare providers section of the website, public relations efforts, articles in Community Oncology about clinical cancer genetics). One of the NSGC's strategic initiatives is to “position genetic counselors as key players in the integration of genomics across the healthcare spectrum,” and we recognize that providing effective, targeted education to other healthcare providers is an important and necessary component of achieving this goal.</p>
ACMG	<p>We provide education to medical geneticists and others interested in medical genetics. We have active programs in work force in medical genetics assessments.</p>
SGIM	<p>Strong advocacy for primary care physicians workforce</p>

Table 24 Additional Genetics Information Needed by Members (Category 2, Question 4)

Organization	Response
ANA	Education of faculty and those in practice, integration of genetics into the certification process, interdisciplinary learning so all move ahead at the same pace.
ACPM	Yes, the latest technologies and practices in genomics-based preventive medicine, the evidence supporting their effectiveness, and applications in clinical and population-based practice.
ACOG	Yes, emerging issues
AAFP	Yes -According to the 2008 AAFP survey on clinical topics, members need information on allergies, sickle cell disease, developmental/behavioral disorders, degenerative disorders and Alzheimer’s. According to the 2003 CME survey, the top two areas in which they were interested in learning more regarding genomics were common genetic diseases and genetic testing and counseling. The Subcommittee on Genomics which is hosted by the AAFP Commission on Health of the Public and Science (CHPS) also believes that it would benefit members to have more information regarding pharmacogenomics.
AMA	Yes – the Genetics and Molecular Medicine program is working to provide physicians with more information and resources about genetics. However, senior management is faced with competing priorities that many times outweigh genetics. Physicians could use more information on pharmacogenomics (which is an area that seems more ready for prime-time than many other genetic tests), direct-to-consumer genetic tests, how to evaluate the validity and utility of genetic tests (including a physician-friendly resource such as GeneTests to help them in that evaluation), the importance of a family history, information on whether insurance companies will pay for tests, etc.
AAP	There is recognition of the need to raise the “genetic literacy” of pediatricians. As previously noted, discussions around this issue are currently occurring at high levels within the AAP.
APHMG	No, since we are a genetics organization, but we always have up-to-date scientific sessions as part of our annual meeting.
NSGC	Yes. Given how rapidly genomic medicine is evolving, our members need to have continually updated information about genomics (and genetics). Genetics/genomics has changed substantially since the first genetic counselors graduated from a graduate program in 1971 and genetic counselors, with the assistance of organizations like NSGC, have been successful in determining how to incorporate new information and new technology into practice fairly rapidly. However, genetic counselors could benefit from topics on how to integrate genomics into practice, at a practical level, and how to effectively triage genomics services.
AAPA	For most practicing clinicians the genetics test they use most often is family history. We need to continue to emphasize the importance of family history to PAs. A selected few other tests are available (BRCA, etc.) but they actually have limited usefulness in the day to day practice of most PAs. I think our challenge is to not overplay the importance of genetic testing and for many clinicians the promise of genomics is over-hyped and remains just that; a promise yet unfulfilled. PAs and other clinicians need more realistic information about the role of genetics in their day to day practice. And, they also need good guidelines on when a genetic test adds value and when family history or other less costly tools will suffice. In some ways we need to wait for the evidence of effectiveness to catch up with the science of genomics. Education on those issues is probably more important than the last GWAS association.
SGIM	Yes; test interpretation; implication of genetic disorders for prevention and screening.
ACMG	Yes; our members want more practiced based short courses.

Table 25 Approaches to Promote Genetics Education among Health Care Professionals (Category 2, Question 5)

Organization	Response
ACPM	Additional CME opportunities, learning communities and resource portals would greatly help; as well as evidence-based consumer education
ACOG	Medical education, continuing education, certification, certification of professionals
AAFP	<p>As stated under number 5C earlier in the survey, AAFP would be able to provide our members with more opportunities to learn about genetics and genomics with more funding dedicated to that specific purpose. Though there are many grants that have become available for genetic research, there are fewer opportunities available for obtaining funding for education and training purposes. A cooperative agreement related specifically to genetic/genomic education could be one way in which this could happen. For instance, AAFP has a cooperative agreement with the Centers for Disease control and Prevention (CDC) related to immunizations with the goal of increasing immunizations in private practices.</p> <p>Another way in which AAFP might be able to more effectively educate our members regarding genetics is through partnerships with groups such as the Evaluation of Genomics in Prevention and Practice (EGAPP) Working Group. It might be helpful to create a partners group that works with EGAPP to learn their processes for making recommendations in order to best communicate these recommendations to our members.</p>
AMA	<p>Genetics and genomics will probably not have great uptake in the clinic until utility of genetic testing is shown. For physicians, the primary desire will be positive clinical outcome studies. Thus, results of clinical trials showing utility are needed. However, that is going to be slow in coming. In the meantime, education of health care professionals about current technologies and surrounding issues is needed. Clear demonstrations of how utilizing genetic technology can help the physician and patient (and also how NOT using it could harm the patient) should help. There are several education programs underway designed to make genetics easier for physicians – one of them is the RAND project to come up with a standardized genetic lab report, others are CMEs about how to use genetic testing in different clinical scenarios. There should also be increased genetics content in medical school curriculum, and more genetics exposure in residencies – this is an efficient way to accelerate the entry of genetics knowledge into the physician workforce.</p>
NSGC	<p>In the next 10 years, potentially every person seeking healthcare services will have had at least one genomic test, possibly full genome sequencing, and probably over many different periods of their lives. The results of these tests will become an important component of every aspect of medical decision making from assessing the significance of a cholesterol result to prescribing a medication to determining whether a cancer patients needs more aggressive treatment than indicated based on histology alone. Therefore, every provider involved with patient care will have to have some familiarity with genetics/ genomics. These topics are covered thoroughly in genetic counseling programs. However, currently, these topics are not adequately covered in other healthcare professional training programs/medical schools. When they are covered, they still tend to focus on single gene disorders and/or they are taught in a single course after which trainees do not have the opportunity to apply the didactic information to the clinical setting. Since genomics will be a part of all medicine, topics related to genomics need to be integrated in all subjects in all courses. We also need to identify ways to train clinical educators/internship supervisors to recognize genetics/genomics issues in clinics so that trainees are able to observe and then take part in identifying and managing these issues in their clinical rotations.</p>
AAPA	<p>See above, and PAs need to learn the language of genetics/genomics first, in a practical way. Then we can build on the language with new interventions as they become available. I think also the ELSI issues are particularly compelling, and having more education in those areas will lead</p>

	to better understanding and desire for knowledge.
SGIM	Stronger RRC requirements for residency education.
ACMG	The power seems to be in the hands of those who control educational content. Hence, means of raising their awareness of how those they train are performing with regard to genetics sub-score in licensing examinations is critical.

Table 26 Certification of Health Professionals (Category 3, Questions 1-3)

Organization	Questions Dedicated to Genetics	Frequency Questions are Updated	Help Developing Questions
ABGC	100%	Continuously	Yes
ABMG	100%	Annually	No
ACMG	100%	Annually	No
GNCC	100%	-	-
ONCC	<5%	Annually	Yes
SGIM	<1%	Annually	Stronger RRC requirements for residency education

Table 27 Accreditation or Certification of Institutions (Category 4, Question 1-3)

Organization	Minimum Curriculum Requirements in Genetics	Frequency Curriculum Requirements are Updated	Need for Integration of Genetics into Curriculum
ABGC	For a full list of the minimum curriculum requirements please visit our website (http://www.abnc.net/en~lish/view.asp?x1=642&mid=110#A). Several required curriculum content areas related to genetics or genomics include: principles of human genetics, applicability of related sciences to medical genetics (e.g., cytogenetics, biochemical genetics, molecular genetics, developmental genetics, cancer genetics), principles and practice of clinical medical genetics, and social, ethical, and legal issues pertaining to the delivery of genetic services.	They are regularly evaluated on an annual basis	We view this as an important need.
ACGME	The full sets of all program requirements are available at www.acgme.org Review Committees.	The program must document formal, systematic evaluation of curriculum at least annually.	These topics are well intergraded into the curricula.
ARC-PA	From the PA Educational Standards	The	Important

	<p>Instruction in the professional phase of the program must include instruction in the following basic medical sciences:</p> <ul style="list-style-type: none"> a)Anatomy b)Physiology c)Pathology d)Pharmacology and pharmacotherapeutics e)The genetic and molecular mechanisms of health and disease 	responsibility of the program	component in the education of PA students and practicing professionals
SGIM	Yes; required but not clear to examinees.	Uncertain	The need is far greater than the leadership appreciates because the focus remains on other determinants of illness and disease.
ACMG	Yes; they are accredited by the ACGME	Continual	This must be done, though with a focus on the role of the provider in health care delivery and the scope of practice of the providers being educated.

6. SACGHS 2004 Health Professional Organization Survey Respondents

Genetic-Specific Organizations

American Society of Human Genetics
International Society of Nurses in Genetics
National Society of Genetic Counselors
National Coalition for Health Professional Education in Genetics

Professional Education Organizations

American Association of Medical Colleges
American Association of Colleges of Nursing
American Association of Colleges of Pharmacy
American Dental Education Association
Association of Schools of Allied Health Professionals
National Organization of Nurse Practitioner Faculties

General Professional Organizations

American Medical Association
American Nursing Association
American College of Physicians

APPENDIX E

SACGHS Survey of Public Health Providers

1. SURVEY METHODOLOGY

Using the Delphi technique,^{289,290} SACGHS developed 12 competencies in genetics of relevance to the public health workforce. Many of the competencies were derived from existing sources, including the National Coalition of Health Professional Education in Genetics (NCHPEG),²⁹¹ the Centers for Disease Control and Prevention (CDC),²⁹² the Association of State Territorial Health Officers,²⁹³ Training Finder Real-time Affiliate Integrated Network,²⁹⁴ and the University of Washington.²⁹⁵ These competencies were translated into an online survey instrument with the intent of assessing public health providers' opinions on the importance of each competency, their confidence in demonstrating each competency, and the frequency with which they apply each competency. The conceptualization and formatting of the competencies into an online survey was based on work by Kirk, et al., who sought to implement a novel approach to ascertain practitioners' needs in genetics education.²⁹⁶ The questionnaire was reviewed by SACGHS members and staff, and additional items were added to assess the importance of genetics and genomics to the respondent's leadership and their own role in public health. The final online survey was a mixed-format assessment tool that included demographic questions.

To achieve a broad representation of public health providers who work in a variety of settings, recruitment utilized multiple strategies that included (1) using a list of state public health and genetic professionals, (2) partnering with the American Public Health Association Genomics Forum, and (3) partnering with the National Society of Genetic Counselors. An e-mail invitation to participate in the survey was then distributed to approximately 500 public health professionals. Some respondents forwarded the survey to others they felt were appropriate. Online survey participants reflected a diversity of public health providers with varying degrees of genetics responsibilities. For some it is their primary job, for others genetics is just one aspect of their position. A total of 140 responses were received and analyzed. It is not possible to calculate response rate because the total number of individuals who eventually received the survey is unknown.

Survey data for the public health providers in genetic and genomic competencies were initially entered into Microsoft Excel and subsequently converted into the Statistical Package for the Social Sciences (SPSS). For the two open-ended qualitative questions, responses were downloaded and entered into

²⁸⁹ The Delphi technique is a commonly used qualitative method that involves the use of experts to develop, review and refine documents, programs, forms, and other formats for programmatic and research efforts. The process involves the initial development of the document or form by moderator(s) and a subsequent request for input from the experts. This interactive request-input back-and-forth, called 'rounds', continues until the appropriate level of completion is generally agreed on by all. There are generally up to three rounds in the process. As used here, SACGHS served as the content experts and three rounds were carried out to arrive at the final list of competencies.

²⁹⁰ Bernard, H. R., (2000). *Social Research Methods: Qualitative and Quantitative Approaches.*, Thousand Oaks, CA: Sage Publications. p. 247.

²⁹¹ National Coalition of Health Professional Education in Genetics. (2004). Core Principles in Genetics. See http://www.nchpeg.org/index.php?option=com_content&view=article&id=95&Itemid=105. Accessed on December 30, 2010.

²⁹² Centers for Disease Control and Prevention. (2001). Genomic Workforce Competencies. See <http://www.cdc.gov/genomics/translation/competencies/>. Accessed on December 30, 2010.

²⁹³ Association of State Territorial Health Officers. (2001). Framework for Public Health Genetics Policies and Practices in State and Local Public Health Agencies. See <http://www.ct.gov/dph/LIB/dph/Genomics/astho.pdf>. Accessed on December 30, 2010.

²⁹⁴ Training Finder Real-time Affiliate Integrated Network. (2001). Core Competencies for Public Health Professionals. See <https://www.train.org/Competencies/corecompetencies.aspx?tabid=94>. Accessed on December 30, 2010.

²⁹⁵ University of Washington. (2001). Competencies in Public Health Genetics. See <http://depts.washington.edu/phgen/about/Competencies.shtml>. Accessed on December 30, 2010.

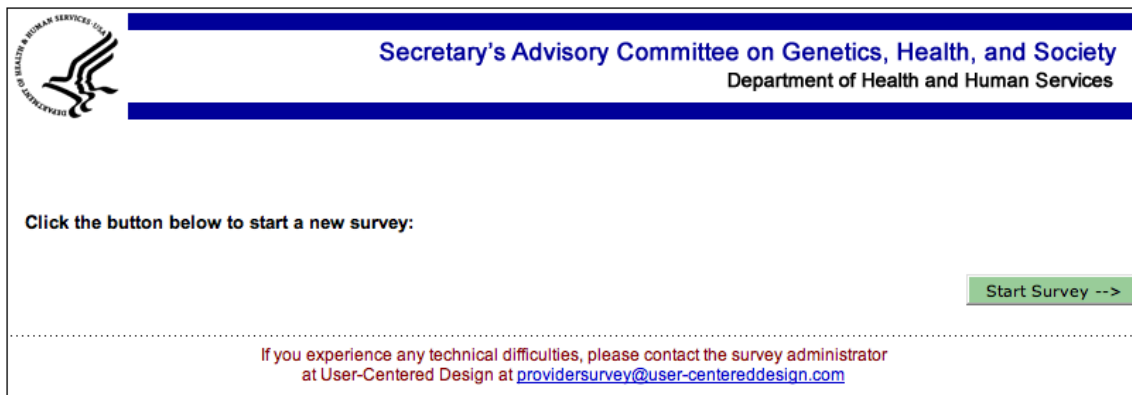
²⁹⁶ Kirk, M., Tolkin, E., Birmingham, K. (2007). Working with Publishers: A novel approach to ascertaining practitioners' needs in genetics education. *Journal of Nursing Research*. 12;597-615.

qualitative analytical software, Atlas TI. The responses were analyzed for commonalities among the responses.

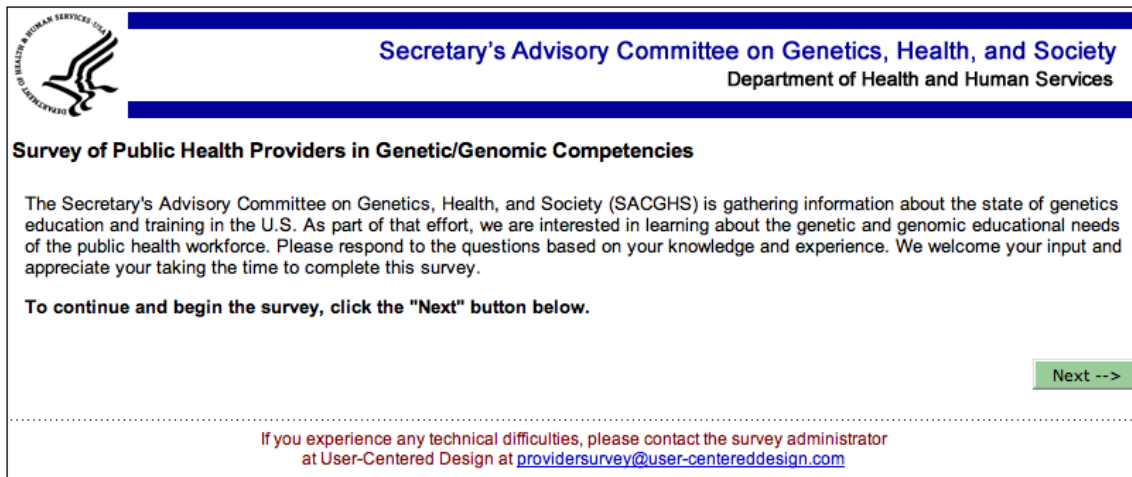
Sample Size and Missing Data

The total sample size used in the analysis was 140 participants. There were instances in which the total number of responses was below 140 because data were missing for specific questions within each competency. Missing data values on Likert scale questions were recoded to equal “no answer.” As a result of the recode, the means were computed based on subtracting the “no answer” responses from the computation and using the 140 participants as the common denominator. The response rate to each question for the 12 competencies appears to be relatively high indicating that minimal data were missing.

2. PUBLIC HEALTH PROVIDERS’ SURVEY INSTRUMENT



Note: the page above was added when requirement for a survey validation ID was removed.





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Privacy Statement

Your participation in this survey is completely voluntary. Please be assured that your participation in the survey will be kept confidential and your responses will never be linked or associated with you. You may skip any questions that you prefer not to answer. You are also free to stop participating at any point during the survey and have your responses deleted by clicking the "Opt out of survey" box at the bottom of each survey page.

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Part I: Your Practices of 12 Competencies

% Completed

Reflecting on your current role, please check one answer from the questions below that best describes your practice for each competency statement.

A public health professional is able to:

1. Maintain up-to-date knowledge on the development of genomic science and technologies within his or her professional field and program to apply genomics as a tool for achieving public health goals.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

2. Demonstrate basic knowledge of the role that genetics/genomics plays in the development of disease and in screening and interventions for programs of disease prevention and health promotion.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

3. Describe the importance of family history in assessing predisposition to disease.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

4. Identify opportunities and integrate genetic/genomic issues into public health practice, policies or programs effectively.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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Part I (cont'd)

% Completed

Reflecting on your current role, please check one answer from the questions below that best describes your practice for each competency statement.

A public health professional is able to:

5. Maintain up-to-date knowledge of genetics/genomics-related policies, legislation, statutes, and regulations.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

6. Describe the potential physical and psychological benefits, limitations, and risks of genetic/genomic information for individuals, family members, and communities.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

7. Collaborate with existing and emerging health agencies and organizations, academic, research, private and commercial enterprises, and community partnerships to apply genetics/genomics knowledge and tools to address public health problems.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

8. Identify the resources available to assist clients seeking genetic/genomic information or services, including the types of genetics professionals available.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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Part I (cont'd)

% Completed

Reflecting on your current role, please check one answer from the questions below that best describes your practice for each competency statement.

A public health professional is able to:

9. Conduct outcomes evaluation of available genetic/genomic programs and services to determine their effectiveness.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

10. Identify the political, legal, social, ethical, and economic issues associated with integrating genomics into public health.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

11. Use information technology (IT) to obtain credible, current information about genetics; to utilize IT skills to share data and participate in research, program planning, evaluation, and policy development for health promotion and disease prevention.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

12. Identify appropriate and relevant genetics research findings that can be translated into public health policies or practices.

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Not at All	Not Very	Somewhat	Very
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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Part II: Importance of genetics/genomics to your institution's leadership

% Completed

1. Does your senior administration think that genetics/genomics is important to your job responsibilities?

- Not at all important
- Of little importance
- Somewhat important
- Important
- Very Important

2. Does your senior administration think that genetics/genomics is important to their job responsibilities?

- Not at all important
- Of little importance
- Somewhat important
- Important
- Very Important

3. How adequate are your resources for implementing genetic/genomic competencies into your work/role?

- Not at all adequate
- Somewhat adequate
- Adequate
- Very adequate

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Part III: Your role in Public Health

[Progress Bar] % Completed

1. At what level of public health do you work?

- Federal
- State
- Local
- Academic
- Private, non-profit organization
- Community-based Organization
- International
- Other (specify):

2. What is your job title?

3. What percent of your time do you spend doing the following?

Role	< 25%	25-50%	50-75%	> 75%
Administrative	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Program Planning	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Direct Consumer Care	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Policy/Legislative	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Research	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Assessment/Evaluation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Education/Training	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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Part III (cont'd)


[Progress Bar] % Completed

4. Please describe any efforts that you or your organization have undertaken to ensure that genetic services or information are available for vulnerable or underserved populations. Are there particular strategies you would recommend? (Limit 200 words).

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Part IV: Additional Information % Completed

1. Please provide any additional information that you would like to share with SACGHS on the topic of genetics and genetics education for Public Health Providers. *(Limit 200 words).*

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3. RELIABILITY RESULTS AND DISCUSSION

A total of 140 respondents were entered into the dataset. For the reliability analysis, the valid sample size was 132 participants due to missing data that were automatically excluded from the analysis. The number of total items in the overall reliability analysis was 36. These items consisted of three of the same questions for each of the 12 competencies. Additionally, three separate reliability analyses were conducted for each of the three questions that were asked for all 12 competencies. In each of these three analyses the total number of items in the analysis was 12.

Reliability for Overall Instrument (12 Competencies each with 3 Questions Totaling 36 Items)

The overall Cronbach's Alpha for the instrument is 0.980, indicating excellent overall reliability of the survey instrument. The corrected item-total correlations show that the correlations between each item and the total score from the instrument are well correlated (correlation values greater 0.3), and as a result items from the overall instrument should not be dropped. The correlation values range from 0.651 to 0.842 for the 36 items.

Alpha values for each item, if the item is dropped from the analysis, are close to the overall Cronbach's Alpha. In every instance the alpha value for each item, if dropped, is slightly under 0.980. Once again, deletion of items from the overall instrument is not necessary. In other words, none of the items would statistically influence reliability if dropped from the analysis. In fact, deleting any item from the analysis would actually lower the overall reliability from 0.980 to 0.979.

The overall instrument for all competencies appears to have good internal consistency with a Cronbach's Alpha of 0.980. All items were acceptable for retention. All items correlate to the overall instrument with an acceptable degree with correlations above the comparison threshold of $r = 0.30$.

4. RESPONSES TO THE PUBLIC HEALTH PROVIDERS' SURVEY: SUMMARY DATA

Table 1 Perception of the Importance of the Competencies

Competency	Question	Number of respondents ranking concept as priority					Resp. Rate	Mean
		0	1	2	3	4		
1. Maintain up-to-date knowledge on the development of genomic science and technologies within his or her professional field and program to apply genomics as a tool for achieving public health goals.	How important is the competency?	9	0	4	35	92	94 %	3.7
	How confident are you in demonstrating the competency?	9	3	21	61	46	94%	3.1
	How frequently do you apply this competency?	10	6	25	32	67	93%	3.2
2. Demonstrate basic knowledge of the role that genetics and genomics plays in the development of disease and in screening and interventions for programs of disease prevention and health promotion.	How important is the competency?	10	0	2	17	111	93%	3.8
	How confident are you in demonstrating the competency?	13	1	17	43	66	91%	3.4
	How frequently do you apply this competency?	12	6	20	35	67	91%	3.3
3. Describe the importance of family history in assessing predisposition to disease.	How important is the competency?	12	1	1	22	104	91%	3.8
	How confident are you in demonstrating the competency?	11	5	11	39	74	92%	3.4
	How frequently do you apply this competency?	12	11	27	43	47	91%	3.0
4. Identify opportunities and integrate genetic/genomic issues into public health practice, policies or programs effectively.	How important is the competency?	10	0	1	28	101	93%	3.8
	How confident are you in demonstrating the competency?	11	2	23	56	48	92%	3.2
	How frequently do you apply this competency?	12	8	37	36	47	91%	3.0
5. Maintain up-to-date knowledge of genetics and genomics-related policies, legislation, statutes, and regulations.	How important is the competency?	14	0	4	42	80	90%	3.6
	How confident are you in demonstrating the competency?	15	4	34	55	32	89 %	2.9
	How frequently do you apply this competency?	15	13	43	44	25	89%	2.6
6. Describe the potential physical and psychological	How important is the competency?	14	0	2	33	91	90%	3.7

benefits, limitations, and risks of genetic/genomic information for individuals, family members, and communities.	How confident are you in demonstrating the competency?	15	2	20	49	54	89%	3.2
	How frequently do you apply this competency?	18	10	39	32	41	87%	2.9
7. Collaborate with existing and emerging health agencies and organizations, academic, research, private and commercial enterprises, and community partnerships to apply genetics and genomics knowledge and tools to address public health problems.	How important is the competency?	14	1	1	27	97	90%	3.7
	How confident are you in demonstrating the competency?	16	6	26	54	38	89%	3.0
	How frequently do you apply this competency?	15	13	46	34	32	89%	2.7
8. Identify the resources available to assist clients seeking genetic/genomic information or services, including the types of genetics professionals available.	How important is the competency?	13	0	6	25	96	91%	3.7
	How confident are you in demonstrating the competency?	14	7	29	37	53	90%	3.1
	How frequently do you apply this competency?	18	24	34	30	34	87%	2.6
9. Conduct outcomes evaluation of available genetic/genomic programs and services to determine their effectiveness.	How important is the competency?	16	1	8	34	81	89%	3.6
	How confident are you in demonstrating the competency?	16	19	29	49	27	89%	2.7
	How frequently do you apply this competency?	16	44	48	19	13	89%	2.0
10. Identify the political, legal, social, ethical, and economic issues associated with integrating genomics into public health.	How important is the competency?	16	0	4	26	94	89%	3.7
	How confident are you in demonstrating the competency?	17	9	23	44	47	88%	3.0
	How frequently do you apply this competency?	18	17	41	33	31	87%	2.6
11. Use information technology (IT) to obtain credible, current information about genetics; to utilize IT skills to share data and participate in research, program planning, evaluation, and policy development for health promotion and disease prevention.	How important is the competency?	16	1	4	38	81	89%	3.6
	How confident are you in demonstrating the competency?	16	11	25	54	34	89%	2.9
	How frequently do you apply this competency?	17	22	36	30	35	88%	2.6

12. Identify appropriate and relevant genetics research findings that can be translated into public health policies or practices.	How important is the competency?	16	0	5	24	95	89%	3.7
	How confident are you in demonstrating the competency?	17	5	22	51	45	88%	3.1
	How frequently do you apply this competency?	19	12	47	33	29	86%	2.7

Table 2 Importance of Genetics and Genomics to Job Responsibility (Part II, Question 1 & 2)

No Answer	Not at All Important	Of Little Importance	Somewhat Important	Important	Very Important
Does your senior administration think that genetics and genomics is important to your job responsibilities? (123 total responses; 88%)					
17	6	20	24	22	51
Does your senior administration think that genetics and genomics is important to their job responsibilities? (121 total responses; 86%)					
19	10	32	37	15	27

Table 3 Adequacy of Resources to Implement Genetic and Genomic Competencies (Part II, Question 3)

No Answer	Not at all Adequate	Somewhat Adequate	Adequate	Very Adequate
How adequate are your resources for implementing genetic/genomic competencies into your work/role? (123 total responses; 88%)				
17	28	51	27	17

Table 4 Level of Job in Public Health Industry (Part III, Question 1)

Level	Number of Responses	Percent Responding
Federal	16	13%
State	51	41%
Local	0	0
Academic	38	30%
Private, nonprofit organization	11	9%
Community-based organization	5	4%
International	1	1%
Other (commercial laboratory, medical center community programming, nonprofit health organization)	3	2%
Total respondents	125	100%

Table 5 Percentage of Time on Specific Tasks (Part III, Question 3)

Specific Task	No Answer	1	2	3	4	Percent Responding
		< 25%	25-50%	50-75%	> 75%	
Administrative	27	54	39	14	6	81%
Program Planning	30	49	55	4	2	79%
Direct Consumer Care	43	77	15	5	0	69%
Policy/Legislative	39	70	25	4	2	72%
Research	35	48	24	19	14	75%
Assessment/Evaluation	33	61	42	3	1	76%
Education/Training	26	57	41	10	6	81%

Table 6 Efforts Individual or Organization has Taken to Ensure Genetic Services or Information are Available for Vulnerable or Underserved Populations (Part III, Question 4)

We are currently looking at disparities in access to genetic services by utilizing multiple data sources: cancer registry, genetic services data, Medicaid, and Insurance company data.
In my role, this is not discussed; however, I believe that the organization is making efforts to do this. I think that more funding needs to be given to interdisciplinary, graduate-level training programs, especially for terminal degree programs. The shortage of trained professionals in this field and the lack of funding support available for these professionals is dismal, especially given the broad public health implications of such technologies. Training in ethics is also in short supply, as the implications for vulnerable populations, including children and those with disabilities is of particular importance.
Our organization has developed experience in community-based participatory approaches to developing resources and materials for underserved, underrepresented populations. We hope to build on this and engage other communities as we develop our dissemination strategies for existing information and create new resources.
Formative research on needs of these populations as it pertains to genomics. Identifying ways to overcome barriers to genetic literacy - including modifying literacy of complicate family history tools, language barriers, and access barriers.
Designated Regional Genetic Centers with a contractual relationship with the program to provide outreach and community education as well as education of other health professionals. All local health departments aid in locating infants and their parents and linking them to services.
Patients with single gene diseases receive counseling from me about which clinical procedures they can participate in or not before visiting their hematologists for life threatening diseases. Both graduate and undergraduate students receive genetic epidemiology from me about clinical computation and clinical implications about specificity, sensitivity, PV+, PV-, and efficiency and the ELSI of genetic testing. There are numerous strategies I have developed, which are being published (in press) but embargoed.
No efforts that I am aware of for underserved populations.
We are using mixed methods strategies to assess patients', and particularly members of minority groups', willingness to participate in genetic services/technologies and the types of information they need to make informed decisions concerning participation in genetic research. In public health, we need to push for increased health education at the K-12th levels to insure that the general population is adequately prepared for utilizing genetic services/technologies beyond prenatal and newborn screening. We have National Health Education Standards that address knowledge and skills related to family health history and health risk assessment, but it's unclear whether these are universally adopted and used in the K-12 setting. Advocating and enforcing NHES-based assessment in this population would be a good step

towards ensuring some measure of proficiency (genetic health literacy) in the wider public.
Provide access to genetic counseling for all for whom it is indicated. Recommend working on access to counseling (location and payment structures) for target populations.
Comment on this survey: you will not get an accurate picture of practice, when open-ended replies are not possible. There is a gap between doing something 1-2 times/yr vs. on a monthly basis. Also, collaborations question re: public agencies, nonprofit sector, etc., should not include industry in same category. We are selective about with whom we collaborate. We need to be able to get more information about the various industry-backed campaigns that specifically are targeting underserved populations with piths based on "pop genomics" , such as those now being promoted by biopharma industry to discredit "evidence based medicine" as not being in best interests of minority patients, and to promote use of brand-name Rx drugs over generics. And for the general public something needs to be done to strictly regulate the private cord blood industry & its disinformation campaigns. During the Bush Admin, HHS sat on its hands, while allowing the PUBLIC cord blood banking program to become more & more enmeshed with industry, seemingly in a move to privatize it. Meanwhile, no ready source of accurate, independent information is available.
We contract with genetic consultants who have developed educational materials for parents.
My colleagues in our School (Toby Citrin & Sharon Kardia) have developed various strategies for community engagement to disseminate genomic information to diverse audiences. They have used a variety of techniques, including community forums, tailored websites, and developing curricula for high school science teachers.
NYS provides funds for comprehensive, non-categorical prenatal and clinical services throughout the state. All qualified centers receive funds which are targeted mainly to genetic counseling and support services. NYMAC has developed Genetics and Your Life Brochures which will be distributed region-wide. Emergency cards have been developed to ensure that children diagnosed through newborn screening are always able to access appropriate care. More work needs to be done to raise the understanding about the impact genetics has on health; more needs to be done to ensure the availability of genetics professionals through increased recruitment and training, using distance strategies and collaborations with primary care to eliminate barriers due to distance, insurance, literacy and culture.
Hold work-group meetings with representatives of under-served or under-participating communities to engage them in planning, implementation and evaluation of newborn screening programs.
Developed fact sheets and other communication products for consumer audiences Applied for grants to integrate family history screening into primary care settings Collaborated in pilot projects to determine how family history is being collected and used in primary care Have offered workshops on family history collection and interpretation to lay public audiences Have developed courses, conferences and other educational events for public health professionals and general public
We have undertaken initiatives to do research with underserved communities. CBPR would be ideal, but funding doesn't usually support such involvement and long term commitment.
I would not recommend our current strategy, which is to contract out genetic services with very little accountability.
More Spanish-language materials are needed online (and on paper). Public libraries should not be overlooked as a place where people go to look for info.
I would recommend community engagement using community based participatory research methodologies and adult learner format
I am in the process of developing a public health genomics certificate program for our Institute here at the university (HBCU). The mission of our public health program is to improve the health status of the poor and underserved through graduate training, research, and service. Therefore, the purpose of our public health genomics program will be to supply our minority graduates with the knowledge and skills to assume a role in making sure the disparities gap is not widened when it comes to who has access to genetic technology, information or services.

Genetics day at the Capitol since 2000. Very effective in educating legislators. Having a State Genetics Plan and also an advisory board.
Worked on study for state health department. Recommendations re device redistribution, increasing reimbursement for prep/counseling/insurance negotiation time unheeded by professional organizations and by legislature/payers. Federal policy has to push state and local policy.
Research projects focused on genetic literacy
My organization is conducting research aimed at determining whether there are differences in uptake of genetic testing between Caucasian Americans and African Americans. Furthermore, we have conducted several studies that aim to clarify what the barriers are to access to genetic testing at the population level.
Genetic services are available to all members; however, no effort has been made to target underserved individuals at this time.
We educate/train public health professionals about communication of risk, genetic literacy levels, and how to target health messages to different audiences. We test the messages and materials with the target audience to get their input before distributing them. We teach the importance of engaging the community and involving them in the development of any materials or messages to get their input. This increases the chances the product is appropriate for the target audience. We also train them to become messagers within their community.
Have included none in this position in the short time I've been here - though I have made preliminary contact with the state health department genetics program to see how our organization could collaborate with them in this area. I believe we could be helpful in providing professional education opportunities.
Study comparing consumer genetic testing services for companion animals as compared to those for humans. Provide articles on genetics for those interested. Teach future veterinarians and scientists how to access available information on bioinformatics and clinical genetics.
I am an expat American working in Australia. I work on GxE studies for a common childhood condition at a tertiary hospital clinic. As Australia has a universal single-payer health system, all populations are eligible to attend the clinic. As the condition I study is very common and the clinic is very small, wait times to attend the clinic are longer than for other conditions.
Before funding was cut for the Utah Department of Health Chronic Disease Genomics Program, I worked with a university to develop a Spanish Family Health History Toolkit (see http://health.utah.gov/genomics/familyhistory/toolkit.html). It was tested with ESL classes. We also worked with the Genetic Science Learning Center to develop genetics curriculum in Spanish and Tongan for students, their families, and teachers. I also developed a Senior-friendly Family Health History Toolkit with a senior center. All materials are available for free online and used extensively by numerous agencies, health care settings, public health professionals, and the general public.
Community meetings, developed educational materials
Research related to family history as a health promotion tool among the medically underserved
A requirement for all of our programs that we fund. In addition, we have formed partnerships with a wide variety of underserved or vulnerable groups to develop specific public education materials and conduct activities to reach out to these populations
Developed a genetics crosswalk across the department to share genetic information and advances. Worked across the department to promote family health history. Programs in place to contract with genetic tertiary centers statewide to provide services to vulnerable populations.
Several fact sheets on genetic component of chronic diseases, in English and Spanish, low literacy level, on website and paper.
I haven't noticed any efforts by my organization to ensure that genetics services or information are available to vulnerable/underserved populations.
To work with organization such as the Genetic Alliance to develop materials for specific/targeted populations and provide resources for education and outreach.
Develop educational materials on sickle cell disease and related conditions for the general public and for parents of children with these conditions.

We have reading material in English (6th grade level) and Spanish currently. We are working towards serving more underserved populations as well as emerging populations through our outreach program and with our partners. Would recommend more media outlets (radio, TV, billboard, twitter, etc).
Recommend to increase funding and research availability to perform basic science and clinical research in Sickle cell disease and sickle cell trait
New Center for Health Policy
Website has undergone extensive review and revision to reach out to general public. My particular program does not address vulnerable populations directly, but other programs have more interaction through provider offices or parenting programs.
We provide this service daily. Materials are poor. Update # affected, accuracy and completeness of information (esp. re hemoglobin disorders), inform physicians and nurses.
Not enough time to complete but a major effort is funding by state of a Hemoglobinopathy Coordinator in our institution to provide genetic education to patients
Provide outreach genetics services and education via in-person and teleconferencing sessions. Have website with resources. Program listed in phone book and other state resources.
Have improved access to genetic services in the rural areas through telemedicine; conducted Neighbor Island Genetic Clinics; provided translators for Micronesian and other immigrant clients; referred clients who need close genetic follow-up services to community public health nurses; admitted vulnerable and underserved populations to Children with Special Health Needs Branch for service coordination and financial assistance for genetic services.
Extensive work re: PCP education, consumer awareness, health plan coverage of specialized services and community based participatory research focusing on environments conducive to full engagement (physical and social community participation) for people with disabilities - particularly mobility and/or cognitive disabilities.
So far we have developed a two-year program to educate the public, particularly Hispanics. We focused in the importance of newborn screening program and family health history awareness. The program has created bilingual resources like a website: easylearn genetics.net , posters and brochures, and mass media campaigns. Working in partnership with other organizations and professionals was a useful strategy for us.
I work at Boston Medical Center for an organization dedicated to supporting families of children with autism and conducting genetic studies to advance science and understanding of it. The organization employs a Resource Specialist and Research Coordinator to do the former and latter, respectively, at 5 hospitals in the Boston area, and communication and collaboration are encouraged among all sites.
Attend local health dept agencies to provide information about SCD as well as other local agencies to dispense information.
We had a genomics program for five years that was recently discontinued.
We are a public health association so therefore do not do any direct care and very little legislation due to pur NP status. Our planning and assessments have never shown genetics to be a priority, even though we realize they play a large part in healthy people after lifestyles. We have very limited resources to do only what is a TOP priority in our state, and often run short in those areas.
Assumption is that "vulnerable or underserved" means rural, low income has a disability, racial/ethnic minority, etc. We provide information in alternative formats (e.g., Braille, large font, translated language, etc); reflect diverse populations in health promotion messages; serve as a neutral party to discuss statewide access to genetic services, billing/reimbursement issues, etc.; provide monetary assistance for cytogenetic testing among some low income patients.
HRSA grant to collaborate with community-based organizations on developing genetics education resources for underrepresented communities
Our agency contracts with genetics clinics in our state (especially in rural areas) to help improve access - these clinics must accept all patients regardless of ability to pay. We also provide financial assistance on a sliding scale for cytogenetics testing to patients who meet income eligibility criteria. Working with Medicaid, our office created and maintains a credentialing process for genetic counselors so that they may bill Medicaid

for prenatal diagnosis genetic counseling services.
Our parent education materials for newborn screening are translated into Spanish. Some newborn screening materials are also translated into Hmong and Somali. Some are adapted for Amish populations by removing pictures of people.
Developed a State Genetics Plan Administer Genetic contracts Support 8 DPH Genetic Counseling positions: 5 of which are regional
We have translated our patient education materials into 10 languages most commonly spoken here. We cover payment for metabolic formula and pay for up to \$2000 in metabolic foods for every patient identified through newborn screening. Currently coverage is not dependent on income level, and is available to every resident without age limit.
Genetic/genomic services may help vulnerable populations or hurt them, depending on whether the services are based on sound evidence. Very few genetic/genomic services are ready for clinical or public health practice for vulnerable populations. Our office helps populations (vulnerable or otherwise) identify services that are actually useful.
The Tracking program advocates for continued and increased funding for Treatment Centers so that the centers may provide care to the underserved and vulnerable populations.
Outreach clinic one time a month or 3 times a year (depends on location)
State Public Health Genetics Web Page that links to services, conducting GIS analysis of availability genetic services, working on Medicaid reimbursement policy
A seminar series on Public Health Genomics that informs population science. There may be funding opportunities targeted to reducing health disparities among vulnerable populations.
Genetic clinics are set up in public health units that are generally frequented by underserved populations.
Our state funds genetic centers to provide services (counselors) to all who need them. Additionally the Child Health Plus Managed Care Program plus the Prenatal Care Assistance Program provides services to those without insurance. We are working on an education program for physicians on genetic testing so that they are able to better serve all populations.
1. We recently increased our kit fees which fund the Utah Newborn Screening Program, a state mandated but not state funded program for all newborns in Utah. We added Cystic Fibrosis to our screening in 1/09 which includes a DNA panel. 2. I developed a task force to look at Medicaid coverage for needed genetic testing for our genetic and developmental clinics, both of which serve underserved and/or vulnerable children.
Worked to "spread the word" about family health history, how and where to locate resources, have produced informational documents on hereditary cancers (breast, ovarian and colorectal) for PH agency's website.
Three respondents replied with "none" or "not applicable" to this question.

Table 7 Additional Comments to SACGHS on the Topic of Genetics and Genetics Education for Public Health Providers (Part IV)

Genetics education needs to incorporate funding for public health education that not only target public health professionals, but also public health students. As a recent graduate from an ASPH, I had no training in public health genetics. This competency should be included throughout public health education, in addition to programs targeting practicing public health providers. More funding should be provided to institutions that do provide such training.
We have been reaccredited to provide CME for our online genetics education modules, Genetics & Your Practice online (www.marchofdimes.com/gyponline). While the primary audience is health care providers, the modules would provide the public health practitioner insight. We also have a suite of education materials and projects that can be reviewed at www.marchofdimes.com/genetics
Highly important to involve the genetics community itself, particularly genetic counselors, in the education of

Public Health Providers. Increase involvement of clinical genetics professionals in local, state, and national public health offices.
NCHPEG is developing a web-course on genomics for social and behavioral scientists. Likely will be relevant for public health providers.
Newborn Screening Follow Up programs need money and people to perform long term follow and track outcomes of confirmed cases. It will not happen without federal assistance. Federal law prevents the Department of Education sharing information with other state agencies. The Department of Education tracks children with developmental delays and behavioral problems. Information such as that would be invaluable to Newborn Screening and Hearing Screening programs. The cost benefit cannot be truly ascertained until the outcomes are documented.
The public health community must become expert in accurately describing absolute risk associated with a possibly mutation. This is especially important dealing with reports from commercial vendors who offer SNP testing for the lay public (without the support of health professionals)).
At the next APHA conference, my colleagues and I will present papers on valid curriculum to train physicians worldwide on genetics in medicine, public health, nursing and pharmaceutical sciences. Genetics and genetic education for public health is now a monumental academic issue for public health providers because we have opened a Pandora's box with genomics because of the recent international accomplishment with full-fledged sequencing of the human genome, I do not mean the fragmented SNP screening being marketed by 23andme from California.
The provider must have a great deal of self-motivation to navigate the numerous self-learning portals and gain critical competencies.
We need a nationwide study on adoption and willingness to adopt genomic competencies across public health fields. Further, we need dedicated funds for curriculum development and training programs.
NCHPEG is leading development of a genetics education module for social and behavioral researchers so there might be some synergies across the two groups.
Most Public Health Providers have limited knowledge and involvement in genetics. The demands of their current roles limit their ability to learn about how genetics impacts other facets of life and health. In general few people consider genetics unless and until it impacts them, either personally or professionally.
A significant gap in educating PH providers (and other providers, as well) is the lack of ready-to-use tools and information that is ready to use in practice today. There needs to be more emphasis paid to supporting genomics capacity building in state and local health departments and in funding translation research that produces the necessary evidence for application of genetics /genomics in practice.
I feel that this area is extremely tangential for most practitioners. They don't see the relevance to their duties and the promise of personalized medicine is still a ways off, so it may be difficult to compel busy professionals to give this area due attention - especially amidst an economic crisis and the latest public health outbreak du jour.
I would like to see funding available (again) for states to prepare or update genetics plans with an emphasis on integrating genetics education into public health programs that target specific ages or conditions (not JUST newborn screening!)
Education for public health providers should introduce population-level (epidemiologic) concepts for interpreting GWAS and other widely publicized research findings. The "classical" genetics of heritable diseases is a poor frame of reference for interpreting these studies. Too often, "genetics professionals" are doing the training and their focus is naturally on the latter domain. But this is only a tiny slice of modern genomics.
Knowing what you don't know is more important than knowing what you do know. Finding a way to help those who don't know understand what is missing is most important
In this age of premature attempts at personalized medicine (e.g., direct-to-consumer genetic testing offered by companies such as Navigenics, 23andMe, deCODEme, etc), the genetics and public health communities need to be more vocal about the limitations of rushing to commercialize research findings that have no proven clinically validity or utility. Understanding these limitations requires more education from a genetics and

genetic epidemiology standpoint that most people could benefit from.
I feel that genetics education for public health should not get so wrapped up in the "medical genetics" aspect of new genomic technology that it no longer has meaning for the communities or populations that we serve. PH Providers need to be stocked with adequate genetic knowledge and skills to assure the well-being of the public (utility, validity, ELSI, etc) but not to take on the role of genetic diagnosing, counseling, and referring...that is a different field.
Widely re-publicize the availability of Six Weeks to Genomic Awareness module; provide CEU's not only to RN's for genomics education; integrate genomics issues into all MPH/DrPH policy curricula (for real, not pro-forma); incentivize LHD folks to take module; provide model, non-specialized bibliographic suggestions for LHD journal clubs...
It needs to be made a priority for senior level management and for new funding if it is to advance.
Survey did a good and comprehensive job identifying critical issues. Results should be widely disseminated.
From my point of view, it is key that health professionals understand basic concepts in genetics that will allow them to integrate and communicate about genetic technologies as they develop. At this point, the most important use of genetic information for the general public is analysis of family health histories. It is also key that health professionals be able to distinguish (or find resources that distinguish) useful genetic tests from ones that have little health benefit.
Primary understanding of how consumers and doctors currently understand genetics and may want to use this information is key. The second most important issue is program evaluation. Developing programs without evaluation components or before understanding the need is not a good use of resources and can lead to confusion among consumers.
It is very important and they should be thinking genetically about every program, adding it in as appropriate. But to do this at the state and local level, they need the basic science and an advisor/mentor who can help them incorporate genetics into their daily work. Public health professionals work in a "silo" not a matrix. This limits their networking/collaborating with other organizations and holds them back. Also, they are constantly reinventing the wheel because they are not aware at many levels of what the genetics community has already done in key areas.
PH providers will need to thoroughly understand sensitivity/specificity and population attributable risk to evaluate whether genetic screenings are of value under a PH framework. Understanding proteomics/metabolomics/etc should also be part of a genetic risk educational resource.
Please don't think genomics education is just research and clinical medicine! Most public health professionals are scared of genomics and need basic education. I really think that they don't need all the genetics knowledge like a genetic researcher or genetic counselor to effectively apply tools like family health history into practice and make it meaningful for the public. Most of the time they are doing something "genomically" but they don't realize it. Also, family health history is the most likely tool to be applied in practice and should be the emphasis behind any education. This education must be done at state health departments, local levels, and in community-based organizations.
It would be valuable if we could share information with legislators on the importance of evidence-based information on genetics before passing legislations to screen for tests that are not population-based.
It is important that community physicians understand genetics and not minimize carrier states like sickle cell trait. In feedback from mothers with trait it seems that the pediatricians and other PCPs tell parents that "there is nothing to worry about" if the child carries sickle cell trait. As a result, some parents do not pass this piece of family health history on to the child as they get older, or other family members for that matter.
Ethical and legal issues of genetics are only beginning to be realized. Public Health Providers have a great responsibility to be educated in these issues so that they can protect the public from discrimination and exploitation based on data acquired through testing for genetic conditions.
Please have national certification programs. For example in CA, you must be a certified counselor with the state- this helps ensure everyone has the same information.
Please keep in mind that genetics is only a part of the picture and needs to be presented in context with other environmental factors. And by environment, I mean everything from actual drug/toxin interaction to the

public policies, disparities in economic and housing opportunities, and racism and sexism that overlays the whole thing. It's a multi-leveled approach to environment.
Provide accurate, up to date, complete information.
There is a need for more genetics education for public health providers such as public health nurses; primary health care centers; outpatient clinics; midwives; childbirth educators; nurse practitioners; nutritionists; physicians; laboratorians; maternal child health programs; chronic disease programs.
This survey asks about public health providers in general - hence some of my answers. While I do believe genomics crosses all public health subspecialties, I'm not sure if all public health professionals need the same level of expertise in genetics/genomics - at a minimum there should be this level of expertise within an agency AND that staff are aware and encouraged to seek this expertise. That currently happens in some areas but not across all programs in my state.
More funding for public health genomics research
More funding for genetic education programs should be allocated to health departments and local organizations interested in this area. Engaging community organizations with trainings and disseminating materials gave us wonderful results, however we need to fund their educational activities for program sustainability.
PHPs provide a unique opportunity to build programs for the public based on the outcomes of genetics research at the molecular and clinical level, yet there are too few programs in practice and being developed. Communication is key, and there just isn't enough of it going on to create a huge effect.
There absolutely must be leadership support for educational activities to be successful in Public Health organizations. The Leadership must value and sanction time for genomics activities.
Providers need to be able to effectively public health and epidemiology concepts to the lay population and their community. Genetics is just one small piece of this, albeit an important one
It's not on people's radar.
Would be good to have some continuing education on this topic.
Since we are not a direct care provider my responses may not even apply to your survey. In order for us to get involved with genetics education (which is where we could assist) we would have to have a source of funding for at least a part time person.
The standards will likely vary, depending on the role of the public health professional. For instance, newborn screening and genetic services programs will need a high degree of competency in the areas listed. However, other programs in public health (e.g., drinking water, chronic disease programs, quality assurance programs, etc) may not need high levels of competency in these areas. It may be that instead, they need to be able to identify people internal/external to their agencies that can provide support at the time that it's needed. Also, some of the competencies listed may be easier to comprehend, but more difficult to implement because of political/financial context or the nature of an agency's structure.
I think if public health providers have a basic knowledge and understanding of genetics, where to find up-to-date, accurate information, and know how to approach thinking about genetics issues, this will help lay the foundation for the other competencies. Although many professionals do not even have genetics on their radar screen, I also see a problem at the opposite end of the spectrum of professionals being overly eager to incorporate unproven genetic tests or inaccurate genetic information into their program activities. (This can especially be true for hot topics like autism.) I appreciate their enthusiasm, but it's important to understand what the test results can and can't tell us, and to think through the potential implications for families pursuing genetic information. When educating public health professionals, we should be careful not to overhype the potential benefits, to emphasize the need to know where to find accurate information, and to get them thinking about ELSI issues. Otherwise, they're likely to get their information about genetics from the media and marketing messages - not the best sources.
I have recently published a textbook entitled "Public Health Genomics: The Essentials" (Jossey-Bass/Wiley, 2008), which addresses the public health genomics competencies devised by the CDC. It serves as an excellent resource for public health students and professionals who seek an overview of core concepts in the field, and is a great tool to use in graduate and/or post-graduate (continuing education) settings.

<p>I think it would be helpful to have guidance documents (such as OHRP's document for investigators and IRBs on genetics and GINA). It keeps everyone on the same page and up-to-date with changes in policy, legislation, etc. Before people in our institution used to think that anything having to do with genetics was high risk, but now we evaluate the context of that research taking place so that we can more appropriately inform people of reasonably foreseeable risks as well as offering genetic counseling services if applicable. Another change is that we evaluate genetic activities in terms of their potential broader impact on populations and communities. What we would like to do is to find a good way to education the local community about genetics so they feel more empowered to make decisions relating to genetic testing or understanding genetic risk and associations.</p>
<p>Those who work in newborn screening are acutely aware of genetics issues. In states that don't have a full time state genetics coordinator, efforts to promote that need to public health leadership in the states would be appreciated. It should be demonstrated to them; the many ways a state genetic coordinator could work with multiple public health programs and collaborate with schools of public health, med schools and private health care providers. Of course funding of those positions would be a big help.</p>
<p>Since 95% plus of research in genomics is in gene discovery and little practical, valid, beneficially research is coming from genetics/genomics research and at the same time genomics is being hyped by marketers, it is very difficult to sort out what genomic services are valid, beneficial, practical and ready for use. So, the main competency professionals need is ability to sort out the hype from the evidence and to identify credible sources of information on what can be applied. Most don't need to know genomics science, just as all don't need to know all the carcinogens in tobacco. They need to know what works and how to get services that work provided to the population. Since only 50% of people get recommended services, the challenge is in getting services provided, when science shows they actually benefit more people than they harm.</p>
<p>Most of our Genetics education is to the Birthing Facility Staff on the policies and procedures for newborn screening. Our education to the general population is general because of the lack of funding for media resources. We do take advantage of the Newborn Screening Month (September) and try to provide as much education on a more widespread geographical area as possible. The State Department of Public Health in CT is committed to genomics/genetics education. Collaborative efforts are making impacts in the Sickle Cell Disease community and Cystic Fibrosis.</p>
<p>The Institute for Public Health Genetics at the University of Washington, Seattle offers interdisciplinary MPH and PhD degrees in Public Health Genetics. This program provides training in all of the competences listed in this survey, and there are more than 50 graduates of the program. The training is provided by 20 faculty members from many different departments, schools, and research institutions in Seattle. The Ph.D. program was just approved for "continuing status" as an established degree at the UW, and was described as a model interdisciplinary program by the review committee.</p>
<p>See previous note about physician education on genetic testing.</p>
<p>Although I do not oversee this effort, I know that our federal grant for our genomics effort within the UDOH was not re-funded. There is currently NO state funding or other federal funding for the efforts in this area in Utah. I feel strongly that public health should be aware of all the potential genetic tests that are available to consumers. We should be able to offer training on family history taking. We should be a resource for the public. Without funding, this is not possible.</p>
<p>It is greatly needed and underfunded; there is concern that the larger, nationally-seen efforts aren't getting into the hands of providers at the local and community level. More needed in that area.</p>
<p>One respondent replied with "not applicable" to this question.</p>

APPENDIX F

SACGHS Study of Consumer and Patients

1. SEMI STRUCTURED INTERVIEWS

Experts Participating in Semi-Structured Interviews

Health communications and genetics education:

Kimberly Kaphingst, Sc.D. Investigator, Social and Behavioral Research Branch
NHGRI/NIH

Celeste Condit, Ph.D. Professor, University of Georgia

Molecular genetics:

Louisa Stark, Ph.D., Director, Genetic Science Learning Center at the University of Utah

David Micklos, Executive Director, Dolan DNA Learning Center

Clinicians:

Mimi Blitzer, Ph.D., Professor, University of Maryland

Cindy Prows, M.S.N., RN, Cincinnati Children's Hospital Medical Center

National lay advocacy outreach:

Sue Friedman, Executive Director, FORCE

Andy Imparato, President, CEO, American Association of People with Disabilities

Industry:

Erin Cline Davis, Ph.D., 23andMe

Trish Brown, M.S., CGC, DNA Direct


Policy:

Kathy Hudson, Ph.D., Director, Genetics and Public Policy Center, Johns Hopkins School of Public Health (Dr. Hudson held this position at the time of the interviews)

Interview Guide

- Background and expertise of individuals or the organization they represent
- Involvement of the individual or organization in projects related to genetics education for consumers or patients
- The general public's current need for knowledge of genetics
- Genetic information that needs translation to consumers and patients
- Recommendations to provide genetics information to the public, includes major topic areas and potential methods
- The role of the federal government and state and local government in genetics education of the public

2. CONSUMERS' SURVEY INSTRUMENT



Secretary's Advisory Committee on Genetics, Health, and Society
Department of Health and Human Services


SACGHS Survey of Genetic and Genomic Education for Seekers of Genetic Information

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) is gathering information about the state of genetics education and training in the U.S. As part of that effort, we are interested in learning about the genetic and genomic educational needs of patients and general public. Please respond to the questions based on your knowledge and experience. We welcome your input and appreciate your taking the time to complete this survey.

In the survey that follows, the phrase "seekers of genetic information" is intended to represent both consumers and patients seeking out genetics information for themselves or family members.

To continue and begin the survey, click the "Next" button below.

If you experience any technical difficulties, please contact the survey administrator at User-Centered Design at consumersurvey@user-centereddesign.com



Secretary's Advisory Committee on Genetics, Health, and Society
Department of Health and Human Services

Privacy Statement

Your participation in this survey is completely voluntary. Please be assured that your participation in the survey will be kept confidential and your responses will never be linked or associated with you. You may skip any questions that you prefer not to answer. You are also free to stop participating at any point during the survey and have your responses deleted by clicking the "Opt out of survey" box at the bottom of each survey page.

If you experience any technical difficulties, please contact the survey administrator at User-Centered Design at consumersurvey@user-centereddesign.com



Secretary's Advisory Committee on Genetics, Health, and Society
Department of Health and Human Services

Page 1

% Completed

1. Have you been involved with planning or implementing a genetics education program for seekers of genetic information?

- Yes
 No

2. Based on your opinion, please rank the concepts individuals most need to know about genetics and genomics to be informed seekers of genetic information as it relates to health. (Rank 1-5, 1 being the highest priority. Enter integers only.)

Rank
1-5

- Basic genetic and genomic concepts and terminology (i.e. inheritance, what is a gene; what is a genome)
 Common diseases are caused by complex genetic and environmental factors
 Genetics is relevant to everyone's health
 Family history is an important tool for understanding your health and disease
 Understanding an individual's genetic makeup by itself will not solve all health problems

If there are more important items not listed above, please specify:

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% Completed

3. Please rank the importance of the following topics that may have special relevance for seekers of genetic information as it relates to health. (Rank 1-4, 1 being the highest priority. Enter integers only.)

Rank

1-4

- How to access genetic tests
- How to interpret and evaluate the credentials of a genetics professional
- How to interpret results of a genetic test
- Where to find reliable genetic and genomic information

If there are more important items not listed above, please specify:

4. Please rank the following barriers to genetics and genomics education efforts for seekers of genetic information as it relates to health. (Rank 1-5, 1 being the most important. Enter integers only.)

Rank

1-5

- Lack of health professionals' understanding of genetics
- Lack of individual health literacy in genetics
- Lack of access to genetic services for consumers/patients
- Direct-to-consumer marketing of genetic tests before there is evidence of their utility or benefit
- Lack of patient understanding of genetic testing implications for themselves or their family (i.e. whether to share results with family members)

If there are more important items not listed above, please specify:

Opt out of survey

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% Completed

5(a). Please rank the potential roles in genetics and genomics education of the public for the federal government. (Rank 1-6, 1 being the highest priority. Enter integers only.)

	Federal Government (rank 1-6)
Funding genetics education programs	<input type="checkbox"/>
Education about the regulation of genetic services	<input type="checkbox"/>
Education about the regulation of genetic tests	<input type="checkbox"/>
Education about the licensing of genetic health care providers	<input type="checkbox"/>
Education about genetic anti-discrimination laws	<input type="checkbox"/>
Serving as a clearinghouse of educational information	<input type="checkbox"/>

If there are more important items not listed above, please specify:

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% Completed

5(b). Please rank the potential roles in genetics and genomics education of the public for state governments. (Rank 1-6, 1 being the highest priority. Enter integers only.)

	State Government (rank 1-6)
Funding genetics education programs	<input type="checkbox"/>
Education about the regulation of genetic services	<input type="checkbox"/>
Education about the regulation of genetic tests	<input type="checkbox"/>
Education about the licensing of genetic health care providers	<input type="checkbox"/>
Education about genetic anti-discrimination laws	<input type="checkbox"/>
Serving as a clearinghouse of educational information	<input type="checkbox"/>

If there are more important items not listed above, please specify:

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% Completed

5(c). Please rank the potential roles in genetics and genomics education of the public for local governments.
(Rank 1-6, 1 being the highest priority. Enter integers only.)

	Local Government (rank 1-6)
Funding genetics education programs	<input type="checkbox"/>
Education about the regulation of genetic services	<input type="checkbox"/>
Education about the regulation of genetic tests	<input type="checkbox"/>
Education about the licensing of genetic health care providers	<input type="checkbox"/>
Education about genetic anti-discrimination laws	<input type="checkbox"/>
Serving as a clearinghouse of educational information	<input type="checkbox"/>

If there are more important items not listed above, please specify:

Opt out of survey

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6. Please rank the genetic education and services needs of underserved and vulnerable communities and patient populations. (Rank 1-4, 1 being the highest priority. Enter integers only.)

If you believe that there are no genetic education and service needs due to more pressing health education concerns for this population, please check this box and move to question 7.

Rank
1-4

- Education about access to genetic services
- Basic and relevant genetic health information
- Culturally appropriate genetic health information
- Skills to make informed health decisions

If there are more important items not listed above, please specify:

7. If you are part of an organization, to your knowledge has it created any education programs to address the challenges listed in question 6? (Please check all that apply.)

- Education about access to genetic services
- Basic and relevant genetic health information
- Culturally appropriate genetic health information
- Skills to make informed health decisions
- Other (specify):

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8. In your opinion, what role do you think the U.S. Department of Health and Human Service should take to improve genetics education for those seeking information about genetics as it relates to health? (Limit 50 words)

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The following questions ask for general demographic information about you and your work.

9. In what state do you work?

10. How would you best characterize your organization?

- Health Care Organization
- Advocacy Group
- Public Health Organization
- Academic Institution
- Private Industry
- Other (specify):

11. How important would you say genetics is to the mission of your organization?

- Extremely important
- Important
- Somewhat important
- Not very important
- Not at all important

Opt out of survey

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12. Please provide any additional information that you would like to share with the SACGHS on the topic of genetics and genomics education for patients and the general public (*Limit 50 words*).

Opt out of survey

Submit Survey

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Survey Complete

% Completed

Thank you for completing this survey!

Thank you for your time and contribution to this effort of the SACGHS. Your input is valuable.

If you experience any technical difficulties, please contact the survey administrator
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3. CONSUMER ORGANIZATIONS PARTICIPATING IN WEB-BASED SURVEY

23AndMe

American Association of People with Disabilities (AAPD)

Arrowhead Orthopedics Physician Assistant Residency Program (OSPAR)

Asian & Pacific Islander American Health Forum (APIAHF)

Asian American/Pacific Islander Nurses Association, Inc. (AAPINA)

Association for Community Affiliated Plans (ACAP)

Association of American Indian Physicians (AAIP)

Association of Asian Pacific Community Health Organizations (AAPCHO)

Association of Hispanic Healthcare Executives (AHHE)

Association of Hispanic Mental Health Professionals (AHMHP)

Association of Minority Health Professions Schools, Inc. (AMPHS)

California Pan-Ethnic Health Network (CPEHN)

City of Hope

Colorectal Cancer Coalition (C3)

DeCode

DNA Direct

FORCE: Facing Our Risk of Cancer Empowered

Genetic Alliance

Hispanic Dental Association (HDA)

Latino Health Communications (LHC)

Latinos & Hispanics in Dietetics and Nutrition (LAHIDAN)

League of United Latin American Citizens (LULAC)

National Alliance for Hispanic Health

National Asian Pacific Center on Aging (NAPCA)

National Asian Women's Health Organization (NAWHO)

National Association for the Advancement of Colored People (NAACP)

National Association of Community Health Centers (NACHC)

National Association of Hispanic Nurses (NAHN)

National Association of Public Hospitals and Health Systems (NAPH)

National Coalition of Ethnic Minority Nurse Associations (NCEMNA)

National Council of La Raza (NCLR)

National Council of Urban Indian Health (NCUIH)

National Forum for Latino Healthcare Executives (NFLHE)

National Hispanic Medical Association (NHMA)

National Hispanic Chamber of Commerce on Health (NHCCH)

National Indian Education Association (NIEA)

National Indian Health Board (NIHB)

National Latino Behavioral Health Association (NLBHA)

National Medical Association (NMA)

Navigenics

Philippine Nurses Association of America, Inc. (PNAA)

Racial and Ethnic Health Disparities

Summit Health Institute for Research and Education, Inc. (SHIRE)

The Hispanic-American Allergy, Asthma and Immunology Association (HAAMA)

The Latino Caucus of the American Public Health Association

Translational Genomics Research Institute (TGen)

Urban Appalachian Council (UAC)

We Act for Environmental Justice

4. SEMI-STRUCTURED INTERVIEWS

Table 1 Key Finding from Semi-Structured Interviews

<p>Perceptions about consumers' understanding of genetics and genomics</p> <ul style="list-style-type: none"> • Segments of the general public are struggling to stay abreast of rapidly advancing genetic technologies and the potential benefits and risks of these technologies. • The public understands that genes and behaviors are related to health outcomes but they have less understanding of how genes and behaviors relate to each other. • Segments of the public have a common misconception that genetic predisposition is deterministic. • Segments of the public do not understand complex traits and that there are multiple risk factors for a single health condition.
<p>Challenges consumers face in obtaining information about genetics and genomics</p> <ul style="list-style-type: none"> • Finding accurate information about genetics and genomics is difficult. • The public includes many diverse cultures and languages that have different concepts and words to describe inheritance.
<p>Where people get information</p> <ul style="list-style-type: none"> • From a variety of sources including the news, television, Internet, local and religious communities.
<p>Successful and suggested models for genetics education</p> <ul style="list-style-type: none"> • When developing programs, organizations must assess and understand the needs of the specific community. • Improve genetic and genomic education among health providers because many consumers and patients prefer to get their health information from their primary health care provider. • Enhance the communication skills of researchers so scientific concepts and the importance of research and public participation can be fostered among consumers and patients. • Collaborative projects between nonprofit organizations and academic institutions or agencies like CDC or NIH excel at identifying immediate educational priorities and can act quickly to implement strategies to fill a specific need. • The Internet is an important and growing source for genetic and genomic information and could be used effectively to provide balanced, accurate information and help counter existing exaggerated claims and miscommunication.
<p>The role of government in activities related to genetics education of the public</p> <ul style="list-style-type: none"> • The Federal government is seen as a more unbiased source of information than a commercial company or corporate source and thus has an important role to play in educating the public in genetics and genomics. • The government should clarify the issue of regulation of laboratory tests and genetics in general. There is the assumption that all genetic tests have gone through FDA approval or some other rigorous review by a Federal agency. • On a societal level, it was felt that the government should play a monitoring role. • The government can influence education and support formal genetics education in schools and update the National Science Education Standards. • All of the interviewees agreed the government should fund more programs to improve genetic literacy.

5. RESPONSES TO THE CONSUMER SURVEY

Figure 1 Geographic Distribution of Responses (Survey Question 9)

Respondents were asked “In which state do you work?” Responses to this question were received from 256 individuals in 39 states plus the District of Columbia. These respondents are shown in the map below. Numbers refer to the number of responses from each state. The color of each state and the District of Columbia is proportional to the number of responses (darker colors indicate more responses than lighter colors). The largest number of responses (> 17) came from California, Maryland, New York, and the District of Columbia, with a strong showing (>10 responses) from Massachusetts, North Carolina, Georgia, Florida, Texas, Michigan, and Illinois. No responses were received from Alaska, Hawaii, Idaho, Wyoming, North Dakota, South Dakota, Mississippi, West Virginia, Vermont, New Hampshire, and Rhode Island.

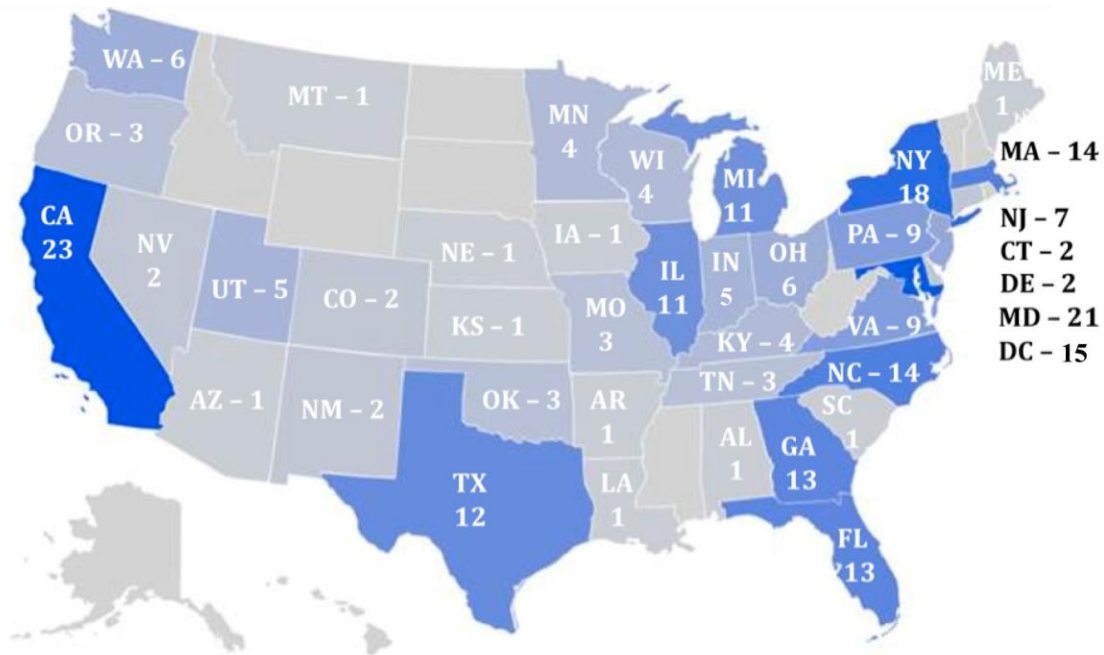


Figure 2 **Distribution of Organization Types (Survey Question 10)**

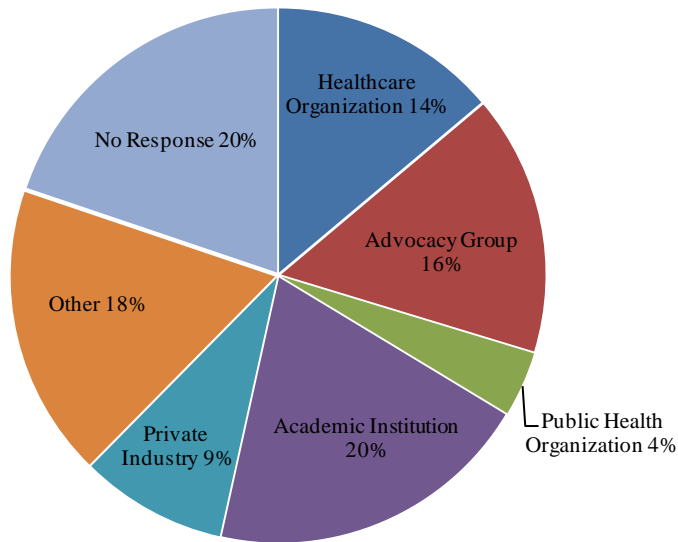


Table 2 **Importance to Genetics to Organizational Mission (Survey Question 11)**

	Number of respondents
Extremely Important	126
Important	75
Somewhat Important	44
Not Very Important	19
Not at All Important	5
No Answer	68

Table 3 **Involvement with Planning or Implementing Genetics Education Program for Seekers of Genetics Information (Survey Question 1)**

	Number of respondents	Percent of those responding
Yes	168	55%
No	138	45%
No Answer	31	n/a

Table 4 Concepts for Informed Seekers of Genetic Information (Survey Question 2)

Item Name	Number of respondents ranking concept as a priority*					No Response
	1	2	3	4	5	
Terminology	92	34	56	45	72	38
Cause Factors	42	65	84	65	42	39
Relevance	69	59	44	65	59	41
Family History	66	101	57	50	26	37
Genetic Makeup	30	40	55	70	98	44

* Rank 1-5, 1 being highest priority

4.1 Free-Text Responses to Concepts for Informed Seekers of Genetic Information

1. A basic sense that different versions of genes make different versions of body systems which then do different tasks more or less efficiently, etc.
2. A better understanding of the difference between what is possible in research vs. what is clinically available or interpretable.
3. All genetic tests do not provide the same type of information
4. Although the focus of this survey is genetic information, a higher level of knowledge about health issues in general would help empower the average individual to advocate for themselves.
5. Basic biological education in American schools is lacking because of so called "Intelligent Design" nonsense.
6. Basic concepts relating to risk, probability.
7. Certain therapies are genetically-based so it is important to understand your genetic makeup
8. Clinical lab testing for genetic disorders is, in concept and practice, no different than any other kind of clinical lab testing.
9. Comment: the first category (basic genetic and genomic concepts) needs to be taught AS PART OF the other four categories, rather than alone. Think sidebars rather than articles.
10. Difference between genes that confer risk versus definitive diagnosis
11. Don't place blame for a genetic disorder on yourself or others
12. Educating children regarding (family) genetics is also vital
13. ELSI issues related to genetic screening and testing; GINA; differences between genetic screening and testing; financing /reimbursement of genetic tests etc.
14. Ethics and informed consent
15. Even with a great deal of genetic information, environment can modify gene function (epigenetics)
16. Family history and genetic testing (markers, mutations) do not guarantee development of disease.
17. For many health problems, behavioral and environmental changes are more efficacious and cost effective than attempts at genetic interventions.
18. Gene diagnosis should be reserved for special situations; there are many less expensive diagnostic tools.
19. Genetic factors result in positive diversity.
20. Genetic information protection from GINA laws and others
21. Genetic makeup is an important factor in assessing RISK for many disorders.
22. Genetic test results should be interpreted with caution by someone who is familiar with the test.
23. Genetic testing is not right for everyone.
24. Genetics cannot and will not ever be a useful predictor of most common diseases.
25. Genetics in the context of other measureable risk factors.
26. Genetics information is still primitive and undeveloped.

27. Genetics is not more complicated than other medical information.
28. How to distinguish between a high and low quality genetics laboratory service
29. Impact of genetic data on insurability.
30. Knowing your genetic susceptibility to the environment can prevent premature morbidity and mortality.
31. Knowledge of genetics and family history are important components of a broader initiative for individuals to become more knowledgeable and proactive about health, healthy lifestyle, and informed personal health management.
32. Limitations and applications of genetic testing
33. Many things in genetics are not 100%. For example, negative testing is often not fully informative, and sometimes a positive result may have wide variability.
34. May have important relevance to disparities when combined with environmental triggers
35. Mechanisms of action for targeted therapies
36. Medical specialists in this area exist (Medical Geneticists and Genetic Counselors).
37. Most diseases and disorders do not need to be weeded out by genetics aimed at selective abortion of possibly "defective" fetuses.
38. Need to understand risk as probabilities, genetic variation,
39. Not to feel guilty about your families genetic history
40. Primary care providers lack information on rare conditions, patients need to know how to get information to their PCPs, PCPs need information on rare conditions (e.g. Huntington's Disease)
41. Prognosis for parent and child
42. Rare diseases (not just common diseases) can also have both complex genetic and environmental factors.
43. Recognizing that having a genetic predisposition to a disease is not the same as having the disease.
44. Role of genomics in personalizing treatment based on individual biology.
45. Taking action regarding a genetic trait could prolong an individual's life
46. Test result may be relevant for close family
47. That each genetic "breakthrough" is not an instant solution making people complacent that nothing more needs to be done.
48. That genetic test results should be interpreted with the assistance of a genetic counselor.
49. That the genome controls normal function and development, and that the genetic inputs into disease are the least common activities in any person's genome (though the impact in terms of chronic disease or death is great)
50. The availability of genetic support groups to individuals & families
51. The importance of seeking out information from qualified health care experts with specific training in genetics!
52. There are examples of genomics bringing better treatments to people
53. Translation takes time: science knows more than doctors can offer
54. Understanding probability and what it means to an individual versus a population
55. Understanding the genetic disorder preventable rather than treatable
56. We must be aware that with genetic research come eugenic applications.
57. What can my genetic information do for my disease?
58. What genetics and genomes might be used for other than improving health
59. Who and how to access care and education of relevant genetic healthcare information AND implications of genetic biobanks

Table 5 Topics of Special Relevance for Informed Seekers of Genetic Information (Survey Question 3)

Number of respondents ranking topic as a priority*					
Item Name	1	2	3	4	No Response
Genetic Test Access	36	83	90	69	59
Evaluate Professionals Credentials	19	82	82	98	56
Interpret Test Results	41	68	73	98	57
Find Reliable Information	189	49	32	15	52

* Rank 1-4, 1 being highest priority

5.1 Free-Text Responses to Topics of Special Relevance for Informed Seekers of Genetic Information

1. Before we look at the above issues we must develop a better understanding of genetics and genetic tests among the nongenetic professionals in health care. We must figure out who is going to provide the massive amount of genetic information and interpretation.
2. Concerns about "liability" that forces medical practitioners to give "worst case scenarios" during genetic counseling of potential parents.
3. Current research on treatment of rare genetic conditions, training primary care providers in rare genetic conditions, working with insurance companies to obtain funding for specialty genetic services
4. Finding medical professionals
5. Genetic information must be culturally sensitive and health literacy and age appropriate.
6. How does this information directly impact my healthcare plan?
7. How to access genetic counselors
8. How to afford it, can it be reimbursed, how does someone find out?
9. How to communicate with a genetics professional
10. How to contact a genetics specialist
11. How to determine whether testing is the right course to take.
12. How to evaluate if a non-genetics health professional really understands genetics!
13. How to find a genetics professional
14. How to find and speak with others who have the disorder/syndrome
15. How to find experts in clinical genetics
16. How to get your doctor/insurance to order the tests
17. How to incorporate genetic information, particularly information related to normal function, into their lives in practical, meaningful ways.
18. How to pay for molecular genetic tests; reimbursement issues related to decision making by third party payers.
19. How to protect the privacy and confidentiality of individual in a culturally respectful manner
20. How to talk to your doctor about genetics/testing
21. How understanding your genetic makeup can impact your insurability.
22. Interpreting the results should be done by the professional not the individual
23. Know your family members' health histories
24. Learn how to deal with the genetic disorder
25. Pitfalls of genotyping
26. That most people do not need genetic test given a negative family history
27. The definition and variety of genetic professionals based on disease expertise

28. The emotional toll of testing
29. The importance of seeking pre and post genetic testing counseling ranks higher than how to get a test. Seeing a genetics professional will assure that the correct test is ordered and properly interpreted.
30. These should be done with a genetic counseling
31. Weaknesses (caveats) of genetic testing
32. What follow up actions could be and what they would cost
33. What health adjustments do I need to make, given my genetic data?
34. What you will NOT learn ever from genetic tests - the concept of risk factor as different from cause!
35. When to seek genetic counseling and testing
36. Which genetic test is appropriate to take

Table 6 Genetic Education and Services Needs of Underserved and Vulnerable Populations (Survey Question 6)

Item Name	Number of respondents ranking topic as a priority*				
	1	2	3	4	No Response
Access to Services	40	41	64	98	94
Relevant Health Information	77	77	57	34	92
Culturally Appropriate Information	54	59	69	63	92
Informed Decision Making Skills	78	69	53	48	89

* Rank 1-4, 1 being highest priority

6.1 Free-Text Responses to Genetic Education and Services Needs of Underserved and Vulnerable Populations

1. Educating physicians and health care providers about the importance of genetics
2. Education about specimen collection, storage, and use
3. Education about the basic concepts of risk vs. causality!
4. Education and services should be equal to all that need
5. Ethnic predispositions
6. Financial assistant for genetic counseling and testing!!!! # 1 need!
7. Genetic education materials in other languages besides English, and reassurance of pt. privacy with testing and results.
8. Good basic primary health care
9. Health care access in general is more important than genetic information for underserved and vulnerable populations.
10. How to participate in patient advocacy organizations e.g. Genetic Alliance member groups
11. Importance of genetics for their health
12. Information about how their genes and genome, viewed as part of a system that includes their environment, acts to shape their daily lives.
13. Motivating primary care providers to seek information about rare genetic conditions
14. Must always be health literacy and age appropriate and spend time on informed consent and the complex concept of 'risk'

15. Professionals with the knowledge to explain to their patients re: genetic disorders and direction to services
16. Require Health Insurance to Cover Testing
17. The importance of participation in clinical trials. Most projects are underfunded and take longer to complete due to lack of participation
18. There is a tremendous need to provide genetic education for underserved communities!
19. What is culturally appropriate in problematic
20. What is your definition of genetic health information?

Table 7 Genetic Education and Services Created to Address Needs of Underserved and Vulnerable Populations (Survey Question 7)

Item Name	Number of Times Item was Selected
Access to Services	114
Basic Health Information	138
Culturally Appropriate Information	80
Informed Decision Making Skills	86
Other	33
Total individuals responding to at least one topic	189

7.1 Free-Text Responses to Genetic Education and Services Created to Address Needs of Underserved and Vulnerable Populations

1. Basic knowledge about genes and the environmental interaction, mechanisms of heritability
2. Connecting families with similar disorders
3. Disease specific education re: sickle cell disease
4. Educating the professional
5. Education about specimen collection, storage, and use
6. Genetic research availability
7. Information about the interplay of genetics and environment
8. Lack of funding
9. My organization deals with only one genetic disease
10. On-line ID of resources for the specific disease
11. Online list of resources, and we are working on having our CAPS guidebook and also the website in other languages, esp. Spanish.
12. Position statements of genetic healthcare issues through professional organizations, research into perceptions of individuals regarding biobank contribution
13. Public advocacy about the ethical concerns of genetic research
14. Research
15. Skills to give in informed consent for genetics research participation
16. We at the ATF try to direct people to testing facilities for TMAU.
17. We have not yet created a formal education program but we provide input on concerns about genetic research.
18. Worked with Children's Hospital
19. Yahoo support group for hereditary Spherocytosis

Table 8 Barriers Preventing Education in Genetics and Genomics (Survey Question 4)

Item Name	Number of respondents ranking topic as priority*					No Response
	1	2	3	4	5	
Health Professionals Understanding	83	80	39	35	39	2.5
Genetic Health Literacy	74	72	63	44	24	2.5
Access to Services	42	37	50	80	64	3.3
Marketing of DTC Tests	33	33	45	44	120	3.7
Implications of Test Results	47	56	78	69	29	2.9

* Rank 1-5, 1 being highest priority

8.1 Free-Text Responses to Barriers Preventing Education in Genetics and Genomics

1. "Gene patents" restricting research and development of better tests
2. An overwhelming preoccupation with how genes relate to disease, which ignores and downplays the role genes play in normal development and adult function.
3. Appropriate use of genetic screening by individuals & doctors.
4. Bringing awareness to community
5. Easy on line access to genetic info for knowledgeable patients
6. Fear of discrimination (employment, insurance, socially) based on results.
7. Fear of genetic discrimination
8. Fear of loss of insurance or jobs if they are tested
9. Financing professional health care information transmission to individuals by their health care providers.
10. Health insurance coverage for services and privacy issues
11. I do not see these as barriers to genetics education efforts as much as I see, the two topics I have ranked, as problems with the existing system of DTC testing. With regard to health professionals' understanding of genetics and individual health literacy
12. I do not understand why DTC in this list is a barrier to education!
13. I think that direct to consumer marketing IS a benefit, not an obstacle--this is very leading, so I will not rank it. To me it is wrong. All of is DREVEN by this availability.
14. Impact of genetic knowledge to insurance companies
15. Lack of culturally competent genetic providers
16. lack of genetics education at the high school and college level
17. Lack of health professionals' understanding of the importance of genetics
18. Lack of insurance coverage for counseling and testing
19. Lack of professional understanding of the benefits & availability of self-help support groups
20. Lack of understanding of providers and consumers of biobank implications
21. Language, access to professionals who speak their language
22. Misrepresentation of the "benefits" of genetic tests by commercial companies
23. Need to provide continuing education for all health professionals and you need to provide genomics education and family history education to every high school student, start in grade school with concepts. Provide community education and empowerment forums.
24. Previous lack of governmental funding and support for equal access to genetic specialty services, challenges to medicine keeping pace with research and technology, barrier of lack of government funding for care

25. Really none of these except health professionals' lack of understanding is a REAL issue. These are pseudo-questions manufactured by those who presume that people should know a lot and care a lot. The real barriers are that people have short term time f
26. Related to the DTC issue is the issue that biotech companies that are CLIA approved can market tests with absolutely no oversight. Even if there is utility and benefit companies are not providing balanced information.
27. The desire to have the genetic test not matter what the outcome
28. Too much belief in science as fact...
29. Willingness to accept that there is a genetic disorder

Table 9 Roles for Government in Public Education of Genetics and Genomics

Number of respondents ranking topic as priority*							
Federal Government (Question 5a)							
Item Name	1	2	3	4	5	6	No Response
Funding	147	55	25	13	16	20	61
Regulation of Services	9	36	54	64	63	39	72
Regulation of Tests	16	32	56	73	56	35	69
Licensing of Health Care Professional	7	26	29	71	64	70	70
Anti-discrimination Laws	32	63	76	23	47	32	64
Clearinghouse	69	63	31	24	20	66	64
Funding	123	35	26	12	24	30	87
Regulation of Services	13	43	62	50	51	27	91
Regulation of Tests	14	29	55	77	44	26	92
Licensing of Health Care Professional	26	29	44	56	54	35	93
Anti-discrimination Laws	33	65	37	36	41	35	90
Clearinghouse	47	48	22	12	28	89	91
Funding	101	30	17	10	27	34	118
Regulation of Services	22	42	57	39	38	18	121
Regulation of Tests	9	31	51	60	41	23	122
Licensing of Health Care Professional	20	23	36	58	38	39	123
Anti-discrimination Laws	35	56	39	31	40	18	118
Clearinghouse	41	38	16	16	28	79	119

* Rank 1-6, 1 being highest priority

9.1 Free-Text Responses to Roles for Government in Public Education in Genetics and Genomics

Federal Government

1. Items 3-6 are not appropriate roles of federal government.
2. Again, this is biased. It focuses on regulation and I do not think that is the major problem. I am an experienced researcher and find this to be prejudicial and focused only on "get the" commercial sector who dared to bring this to the fore.
3. Cultural issues of genetic testing and research among medically underserved populations
4. Educating doctors about new genetic-linked conditions, esp. rare disorders and the resources available to help these patients, such as the NIH
5. Education about the eugenic concerns of using genetics to illuminate an entire population of people against their will, i.e. autistic people, deaf people, people with Down Syndrome.
6. Funding state genetics programs
7. Government financial assistance for persons requiring testing, treatment, etc.
8. I don't think any of these are high priority for the federal govt.
9. Improving the general education in the public schools so that children in primary school have at least a basic understanding of genetics.
10. Money would be better spent on non-genetics related public health endeavors.
11. Must not be strictly Federal government
12. Number one priority of federal government is assuring health care services, long term care services, and disability services for people with or at risk for genetic conditions.
13. Personally, I don't think that the public needs to know about the regulation of genetic services or tests or about the licensing of genetic health care providers. As a rule, adult learners are only interested in information that relates directly to their
14. Programs must be professionals and the public. Professional licensure must demonstrate competencies.

State Government

1. Funding for providing genetic services
2. 3-6 are not appropriate roles of government
3. Assuring Medicaid and third party insurance reimbursement for clinical genetic services, long term care, and disability for people with or at risk for genetic conditions.
4. Education about resources in the public/state for genetic conditions to improve access to care and services.
5. Education about newborn screening as a preventive approach to genetic diseases
6. For states, I see very little role.
7. Funding programs for genetic counselors
8. If the federal government is going to serve as a clearing house, then this is not an activity that the states would have to take on. Also, the only reason I think that it is necessary to deal with the topic of anti- genetic discrimination laws is because
9. Licensure of professionals must demonstrate competencies. State DOH must have a Div of genomics, websites must have tools for professionals and the community including teachers
10. Must not be strictly state government
11. None of the above
12. Requiring genetics education for licensure of health care professionals
13. Cultural issues of genetic testing and research

Local Government

1. 3-6 are not appropriate roles of government
2. Almost no role for local govt. They can't even get TB testing right.
3. Education about purpose, risks and benefits of population biobanks
4. Education about where to go for genetic testing/counseling.
5. Education to the public about locally-available resources regarding genetics testing, care and esp. funding and care of special education children's needs that have genetic conditions
6. I do not think local governments have the capacity to be trying to do this. Better done at a higher level correctly and not repeated (possibly with variation and confusion) at multiple levels
7. I don't see a role for local government
8. I don't see any of this as a local government role.
9. I don't see local government as having a role in this
10. Information on how to find the information needed
11. It will be a long time before local governments will be broadly qualified to help citizens with these matters.
12. Must work with state and federal government
13. New requirements for greater science training across academic institutions
14. No role
15. No role for local governments
16. None
17. None of the above
18. Not a local government issue
19. Not sure of a need for local government in this issue.
20. Requirement for genetics education in public schools

10 Role HHS Should Take in Genetics Education (Survey Question 8)

Consolidated Responses

1. Provide funding
 - To other organizations for educational programs (21 responses)
 - To genetic counselors and genetics programs (6 responses)
 - To universities and medical schools for genetics programs (2 responses)
 - For testing and insurance coverage (1 response)
 - For private/university programs and oversight (1 response)
2. Promote genetics education for
 - The general public (40 responses)
 - Healthcare professionals (24 responses)
 - Medical students (7 responses)
 - K12 and undergraduates (4 responses)
 - HHS and all other government agencies (2 responses)
3. Promote genetics education by
 - Creating national campaigns/public services announcements (13 responses)
 - Creating an informational website (12 responses)
 - Utilizing culturally appropriate information (7 responses)
 - Presenting unbiased/non-religious information (8 responses)
 - Publicly supporting established genetic organizations and programs (5 responses)
 - Distributing printed materials in doctors' offices, hospitals, clinic, etc. (5 responses)

- Using the media such TV and radio (3 responses)
 - Implementing services in the workplace (1 response)
 - Creating guidelines for all published educational material (2 responses)
4. Act as a clearinghouse. (17 responses)
 5. Investigate and enforce the validity of the genetics information made available to the public (7 responses)
 6. Provide education on the use and availability of genetic tests (7 responses)
 7. Provide universal access to genetic services (6 responses)
 8. Regulate of genetic testing (3 responses)
 9. Facilitate and encourage collaboration among health care professionals, support groups, the government, etc (3 responses)
 10. Specifically target and promote education and awareness of Sickle Cell Anemia (3 responses)
 11. Publish information about all known genetic related diseases (3 responses)
 12. Mandate billing/reimbursement for genetic counselors (2 responses)

Additional Responses

13. Keep public informed about updates in genetics medicine
14. The U.S. Dept of Health should gather lists of people affected by rare genetic diseases so that it is known how many are, in fact, impacted by each particular disease.
15. Support scholarships for health educators, genetic counselors and genetic specialists
16. Refine the meaning of transmission and tradition in family values.
17. Promote the availability of genetics professionals (genetic counselors, medical geneticists, genetic nurses, etc) and provide support to these professionals and funding to training programs
18. Expand Orphan drug act to cover drugs that have now been found to help genetic disorders
19. Build up genetic consultations in most health care facilities
20. Establish (genetics) journals for the general population
21. Implement Prenatal and Postnatal Diagnosed Conditions Awareness Act: with disability groups and reproductive rights organizations
22. Facilitate the collection of information from experts, create policies and programs to advance the recommendations of the expert community
23. Create an advisory group to review and grade health and genetic information internet sites.
24. Have more information for medical professionals and patients about APBD.
25. Increase number of evidence based reviews to help providers know what is legit and what is not.
26. Publish abstracts of research projects about genetics and health problems.
27. Inform public about timelines involved in breakthrough findings and public benefit; each breakthrough is not the complete puzzle, just a small piece of the big picture.
28. Promote a realistic expectation of what genetics can accomplish and in what time frame
29. Explain the practical relevance of dollars spent on genetic research to potential therapies and improved health care for millions of Americans
30. Develop and promulgate the use of electronic family medical histories for primary physicians use
31. Determine who will be responsible for oversight of genetic testing.
32. Delineate a national strategy and priorities that others can then help to meet.
33. Conduct storage and processing of information on a regular basis about current state of education and educational needs among patients and their family members.
34. Please be sure the patient's privacy is respected
35. The most active group should be at the Federal level.
36. Should not be used for testing for gay gene or the "I want a blue eyed, blonde hair child"
37. Impartial role without suggesting that certain populations such as autistics, be eliminated from the gene pool.

38. The first step should be when infants are born with medical complications. The second should be when children are diagnosed as special needs.

APPENDIX G

SACGHS Surveys of Federal Agency Activities 2003-2008

1. 2008 FEDERAL AGENCY SURVEY INSTRUMENT

SECTION 1: Department/Agency Information

1. Name of Department/Agency:
2. Is the education or training of professionals in genetics part of the role or responsibility of your Department/Agency? If no, please proceed to Section 2. If yes,
 - A. Please briefly describe this role or responsibility.
 - B. Is your Department/Agency currently able to fulfill this role or responsibility? Are there ways in which your Department/Agency could meet this role or responsibility more effectively?
 - C. How many divisions within your Department/Agency have a role/responsibility for genetics education or training? Please provide the names and directors of those divisions.

SECTION 2: Activities Relevant to Genetics Education and Training

Please list the specific projects your agency funded in genetics education and training of professionals for FY 2003 – FY 2009. You may also highlight any other activities that were supported before FY 2003 or projected for FY 2009 or beyond. Please do not include education and training activities that do not pertain to genetics or genomics.

1. Programs aimed to educate professionals or trainees about genetics or genomics
2. Meetings or conferences aimed to educate professionals and/or trainees about genetics or genomics
3. Genetics educational websites or online resources
4. Activities in which genetics or genomics education may be an important but secondary goal
5. Assessments of professional knowledge about genetics or genomics
6. Analyses or evaluations of the genetics workforce (i.e., capacity, preparedness)
If data are available, can they be shared with SACGHS?
7. Other

SECTION 3: Summary of Each Activity

For each project listed in Section 2, please answer items 1-15 for each project.

1. Title of activity:

2. Amount of award:

3. Funding mechanism: e.g., grant, contract, cooperative agreement, intramural effort

4. Project timeline (start and end dates):

5. Name of funding recipient:

6. Type of funding recipient:

- State/territory government department/agency
- Local government department/agency
- Tribal communities
- Academic institutions
- Community-based organizations
- Professional associations/organizations
- Foundations
- Private industry
- Other (please specify):
- Not Applicable

7. Type of activity:

- Genetics education & training of students/residents entering into professional practice
- Genetics education & training of professionals already in practice
- Genetics workforce analysis
- Other (please specify):

8. Purpose/Goals of activity: What need does this activity address?

9. Focus of activity: Was genetics education and training or genetics workforce analysis a primary goal of the activity? Or a secondary goal?

10. Please provide a brief summary of project/activity:

11. Target audience (check all that apply): Who is this activity trying to reach?

CATEGORY	TARGET AUDIENCE
Students/Residents	
Undergraduate health/science majors	
Graduate or professional school students/residents Specify program type:	

Practicing Professionals	
Academicians: graduate or professional school faculty/deans/curriculum administrators Specify program type:	
Clinical psychologists	
Dentists Specify specialty, if applicable:	
Genetic counselors	
Health plan administrators	
Judges	
Laboratorians	
Law enforcement	
Lawyers	
Nurses Specify specialty, if applicable:	
Advanced practice nurses Specify specialty:	
Other allied health professionals (e.g., speech pathologists, audiologists, nutritionists, occupational or physical therapists) Specify type(s):	
Pharmacists	
Physician Assistants	
Primary care physicians Specify specialty:	
Physician specialists Specify specialty:	
Public health	
Other Specify type(s):	

12. Federal partnerships (check all that apply): What other Federal Departments/Agencies, if any, co-funded/co-sponsored this activity with your Department/Agency?

- Department of Commerce
- Department of Defense
- Department of Education
- Department of Energy
- Department of Health and Human Services
 - Administration for Children and Families
 - Agency for Healthcare Research and Quality
 - Centers for Disease Control and Prevention
 - Centers for Medicare and Medicaid Services
 - Food and Drug Administration
 - Health Research and Services Administration
 - National Institutes of Health
 - Office for Civil Rights
 - Office for Human Research Protections
 - Substance Abuse and Mental Health Services Administration
- Department of Justice
- Department of Labor

- Environmental Protection Agency
- Equal Employment Opportunity Commission
- National Science Foundation
- Veterans Affairs
- Other (please specify)
- Not applicable

13. Non-Federal partnerships (check all that apply): What other groups, if any, co-funded/co-sponsored this activity with your Department/Agency? Note: funding recipients are not considered partners.

- State/territory government department/agency
- Local government department/agency
- Tribal communities
- Academic institutions
- Service/provider organizations
- Community-based organizations
- Professional associations/organizations
- Foundations
- Private industry
- Media
- Other (please specify)
- Not Applicable

14. Impact

- A. What is the need this project is attempting to address?
- B. What is the magnitude of this need?
- C. How do you perceive the urgency of this need?
- D. How many individuals did the project reach?

15. Evaluation

- A. What have been the results of the activity?
- B. Are the results available? If so, where?
- C. Have the results been use? If so, how?
- D. Were the goals of the activity achieved?
- E. What, if any, additional needs or next steps were identified?

SECTION 4: Other Relevant Information

Has there been an evaluation of your Department's/Agency's initiatives or overall efforts in genetics education and training? If so, please summarize the findings.

What are your Department's/Agency's projected priorities for future initiatives in genetics education and training?

Under what legislative authority does your Department/Agency fund genetics education and training projects?

Please provide any additional information you think the SACGHS should know about your Department's/Agency's activities in genetics education and training.

Table 1 Federal Agencies Surveyed in 2003 and 2008

Agency	2003	2008
Administration for Children and Families (ACF)	Responded, no activity	Responded, no activity
Agency for Healthcare Research and Quality (AHRQ)	Responded, no activity	Responded, no activity
Centers for Disease Control and Prevention (CDC)	Responded, provided genetic-related activities	Responded, provided genetic-related activities
Centers for Medicare & Medicaid Services (CMS)	Responded, no activity	Responded, no activity
Department of Commerce (DOC)	Responded, provided genetic-related activities	No response
Department of Defense (DOD)	Responded, provided genetic-related activities	Responded, provided genetic-related activities
Department of Energy (DOE)	Responded, provided genetic-related activities	Responded, provided genetic-related activities
Department of Justice (DOJ)	Responded, provided genetic-related activities	No response
Department of Labor (DOL)	Responded, no activity	No response
Department of Education (ED)	Not included in survey	Responded, no activity
Equal Employment Opportunity Commission (EEOC)	Responded, no activity	Responded, provided genetic-related activities
Federal Trade Commission (FTC)	Not included in survey	Responded, provided genetic-related activities
Food and Drug Administration (FDA)	Responded, no activity	No response
Health Resources and Services Administration (HRSA)	Responded, provided genetic-related activities	Responded, provided genetic-related activities
Indian Health Service (IHS)	Not included in survey	No response
National Institutes of Health (NIH)	Responded, provided genetic-related activities	Responded, provided genetic-related activities
National Science Foundation (NSF)	Not included in survey	Responded, provided genetic-related activities
Office for Civil Rights (OCR)	Responded, no activity	Responded, no activity
Office for Human Research Protections (OHRP)	Responded, no activity	Responded, no activity
Substance Abuse and Mental Health Services Administration (SAMHSA)	Not included in survey	Responded, no activity
Department of Veterans Affairs (VA)	Not included in survey	Responded, no activity

2. RESPONSES TO SECTION 1 OF THE SURVEY OF SELECTED FEDERAL AGENCIES

Genetics Education Role or Responsibility of Federal Agency (Section 1, Question 2a and 2b)

Federal Agency	Is Genetics Education Part of Agency's Role or Responsibility?	Ways Agency Could Meet Role or Responsibility More Effectively
ACF	Did not respond to question	Did not respond to question
AHRQ	Did not respond to question	Did not respond to question
CDC	Yes	While CDC has contributed to genomics education and training of professionals, the agency is not currently able to fully develop this area and respond to emerging developments in genomics, due to limited available resources to assess educational needs among professionals, and to develop and disseminate training tools and curricula, in collaboration with our partners.
CMS	No	Did not respond to question
DOC	Yes	NIST has ongoing projects in genetics education and training of professionals in health-related and nonhealth-related fields. The other agencies of the DOC do not have projects in this area.
DOD	Did not respond to question	Did not respond to question
DOE	No	Did not respond to question
DOJ	Did not respond to survey	Did not respond to survey
DOL	Did not respond to survey	Did not respond to survey
ED	Did not respond to question	Did not respond to question
EEOC	Yes	No. The majority of this training has not yet occurred, as we are waiting for the regulations implementing Title II of GINA to be finalized, likely in the 3rd or 4th quarter of FY 2009.
FTC	No	Did not respond to question.
FDA	Did not respond to survey	Did not respond to survey
HRSA	Yes	Yes, the HRSA is able to fulfill this role and its' responsibilities effectively.
IHS	Did not respond to survey.	Did not respond to survey.
NIH	Yes	Training and education in genetics/genomics are a key component of several CF programs and could be considered a priority area that could be expanded with additional funding.
NSF	Did not respond to question	Did not respond to question
OCR	Did not respond to question	Did not respond to question
OHRP	Did not respond to question	Did not respond to question
SAMHSA	Did not respond to question	Did not respond to question
VA	Did not respond to question	Did not respond to question

3. RESPONSES TO SECTION 4 OF THE SURVEY OF SELECTED FEDERAL AGENCIES

Federal Agencies Projected Priorities for Future Initiatives in Genetics Education and Training (Section 4, Question 2)

Federal Agency	Projected Priorities
ACF	Did not respond to question
AHRQ	Did not respond to question
CDC	Empowering providers with the knowledge and skills to apply genomics knowledge and tools for early disease detection, disease prevention, and health promotion in populations.
CMS	Continue to provide GT information in future CMS basic surveyor trainings and other CMS activities.
DOC	Did not respond to question
DOD	Did not respond to question
DOE	Did not respond to question
DOJ	Did not respond to survey
DOL	Did not respond to survey
ED	Did not respond to question
EEOC	Once the regulations implementing GINA Title II become final, we plan on conducting a number of training sessions for lawyers, HR professionals, small business owners and other interested parties on the legal requirements of Title II.
FTC	The FTC does not engage in professional training, but we will continue to evaluate the need for consumer education about at-home genetic tests and consumer-directed advertising of such tests. We will also monitor consumer-directed advertising of genetic tests and may take action where necessary to prevent consumer deception.
FDA	Did not respond to survey
HRSA	HRSA's Bureau of Health Professions will continue to collaborate with NIH to prepare faculty, students, and currently practicing nurses in application of genetics and genomics science to their nursing practice. Additionally, the Maternal and Child Health Bureau has a new grant initiative for Family Health History for Prenatal Care Providers.
IHS	Did not respond to survey
NIH	Planning and implementation of the Roadmap are highly dynamic processes that are intended to afford NIH the flexibility to quickly respond to new ideas, challenges, gaps, and advances in biomedical research. Nonetheless, decisions regarding use of the Common Fund are based on strategic planning processes involving multiple sources of external and internal input. Common Fund programs currently support activities involving training and education in genetics/genomics. It is possible that, through the strategic planning process, additional funding may be identified as a critical need.
NSF	Did not respond to question
OCR	Did not respond to question
OHRP	Did not respond to question
SAMHSA	Did not respond to question
VA	Did not respond to question

4. RESPONSES TO SECTIONS 2 AND 3 OF THE SURVEY OF SELECTED FEDERAL AGENCIES

Federal Activities Relevant to Genetics Education and Training (Summation of responses from most of the questions in sections 2 and 3)

CDC

The CDC reported the following programs and activities:

- Genomics translation education, surveillance, and policy interventions
 - Family History Education to Improve Genetic Risk Assessment for Cancer
 - Pharmacogenomics Education Program: Bridging the Gap between Science and Practice
 - Promoting Cancer Genomics Best Practices through Surveillance, Education, and Policy
 - Oregon Genomics Surveillance Program: Translation of Genomics Applications into Health Practice
- Academic Centers for Genomics and Public Health
- Genomics educational and training opportunities for public health professionals through the following programs:
 - Prevention Effectiveness
 - Association of Public Health Laboratories
 - Association of Teachers of Preventive Medicine
 - Oak Ridge Institute for Science Education.
- American Society of Human Genetics (ASHG) Public Health Genetics Fellowship
- Public Health Genomics Capacity Building at State Health Departments
- Education materials based on genomics for early disease detection and intervention
 - Primary Immune Deficiency Resource Center (<http://www.info4pi.org/>)
 - Hemochromatosis for Health Professionals (<http://www.cdc.gov/ncbddd/hemochromatosis/training/index.htm>)
 - Make Early Diagnosis to Prevent Early Deaths (MEDPED) Familial Hypercholesterolemia
- Genomics training workshops for CDC epidemiologists
 - Developing Protocols for Genetic Research Studies—Kaiser Permanente, Oregon (March 2009)
 - Conducting, Analyzing and Interpreting Gene-Environment Interaction Studies –Harvard University (May 2009)
 - Overview of Statistical Methods for Using Genetics in Prediction of Disease Risk –Erasmus University, The Netherlands (September 2009)
 - Human Genome Variation Data Analysis and Association Studies Workshop, Atlanta, GA (February 2009)
 - Genetic Epidemiology Short Course, Atlanta, GA(October 2007)

The CDC's Division of Laboratory System (DLS) reported the following programs and activities:

- Meetings and Conferences
 - Northeast CLIA Consortium meeting, Philadelphia, PA. Lecture title: Genetic Testing Quality Assessment (March 2007)
 - Western CLIA Consortium meeting (May 2007)
 - Annual CLIA Surveyor Training Course. Dallas, TX. Lecture title: Genetic Testing Quality Assessment (October-November 2007)

- 14th National CLIA Program Conference of the Department of Veterans Affairs. Charleston, SC. Lecture title: Genetic Testing Quality Assessment (July 2008)
- Improving the Quality of Genetic Testing and Assuring Its Appropriate Integration into Clinical and Public Health Practice
- Reporting DNA-Based Genetic Test Results Applicable to Heritable Conditions and/or Markers of Drug Metabolism: The Clinical Laboratory Report as a Decision Support Tool
- GeT-EQUIP—placed online in 2008: <http://wwwn.cdc.gov/dls/genetics/getequip/portal.aspx>

CMS

From October to November 2007, a Basic Surveyors Training program was provided for new and current State Agency and Regional Office surveyors. The purpose of the week-long program was to provide CMS surveyors the proper materials and training needed to assess a genetic testing laboratory for CLIA compliance. The surveyor training included two sessions that addressed current genetic testing technologies and the CMS survey process for genetic testing laboratories. Evaluations of these sessions were highly favorable and were used to determine the next basic training agenda and to plan for additional training programs.

DOC-NIST

- NIST has built and maintains the world’s most widely used, web-based database on forensic DNA genetic typing, the STRBase. (<http://www.cstl.nist.gov/biotech/strbase/NIJ/STRBase.htm>).
- NIST has also held more than 30 training workshops in forensic laboratories and at major scientific conferences to teach genetic principles to scientists and lawyers. (<http://www.cstl.nist.gov/biotech/strbase/training.htm>).
- NIST Human Identity Project is an ongoing program, begun in 2003, that educates students and professionals about genetics and is funded by the Department of Justice.

DOD

- The DOD genetics workforce consists of physicians with training in clinical genetics, genetic counselors, and pathologists with certification in molecular genetics. Facilities focused on genetics include a dedicated molecular genetics and cytogenetic laboratory, the Armed Forces Institute of Pathology, which performs clinical molecular genetics testing, and plans for a reference molecular genetics laboratory. The DOD has plans to create a general genetics division under the supervision of an Air Force geneticist.
- Currently, the U. S. military is the most experienced practitioner of pharmacogenomic screening on a large, population-based scale. All service members undergo G6PD testing, sickle cell screening, and color vision screening, with subsequent environmental and pharmacologic management designed to prevent disease. In addition to ongoing genetic testing programs, DOD has developed a comprehensive DOD-wide newborn screening laboratory program. The Assistant Secretary of Defense, Health Affairs, has charged the Newborn Screening Integrated Project Team with creating policy and a comprehensive military newborn screening program that would include a comprehensive educational program, a DOD newborn screening website, an EHR-based newborn screening registry, and a comprehensive statement of work for a global newborn screening laboratory contract that would be potentially available for 50,000 annual births to active duty and retired DOD personnel.
- Educational activities include fellowship training in genetics and ongoing efforts to update curriculum and clinical training to meet accreditation requirements of the Accreditation Council on Graduate Medical Education (ACGME). From 2009-2011, the DOD will support the “steady production of one

geneticist per year” in the Army, as well as two-year genetics fellowships followed by a one-year molecular genetics fellowship among Air Force personnel. Future DOD activities in genetics education and training include support for additional genetics fellowships. DOD will also maintain ACGME certification for its CE curricula in genetics, expand its workforce of geneticists and genetic counselors, create new laboratory capabilities, and increase its understanding of the gene-environmental impacts associated with military operations.

- DOD has multiple inter-departmental relationships engaged in personalized medicine programs and EHR standardization efforts pertaining to genomics.

DOE

- In 2003, the DOE survey response focused on some of the social implications of the mapping of the genome and, along with NIH, has devoted 3 to 5 percent of its annual Human Genome Project budget to studying the ethical, legal, and social issues related to the availability of genetic information.
- Since 2003, DOE’s educational efforts included a series of 38 workshops geared to the judiciary. At the workshops, judges explored the fundamentals of genetics and discussed some of the expected ethical, legal, and social challenges that were anticipated to lead to court cases, policy and rule making, or new legislation related to genomics. In addition to the workshops geared to judges, the DOE also supported many programs that provided outreach to communities and to schools.
- Supported the translation of a high school curriculum unit about genomic science into Spanish.
- Sponsored a series of workshops for communities of color in coordination with the Zeta Phi Beta sorority organization. More than 1,000 African-American citizens had attended these workshops by 2003, where they learned about genomic science and about some of the many clinical, ethical, legal, and social implications of genetics research.
- JGI program trains faculty to annotate microbial genomes in the context of the undergraduate curriculum, and for undergraduate research using tools developed by the JGI. Since many faculty need to develop research opportunities for their students, the program gives them the tools and the data so that students can carry out bioinformatics research. In the first year and a half of the program 55 faculty members and approximately 700 students were trained.
- American Society of Microbiology/DOE-JGI Program: a Bioinformatics Institute held twice yearly that introduce basic bioinformatics to undergraduate faculty. Dr. Kerfeld, JGI, co-organizes the pedagogy for the DOE-JGI/ASM workshops with Professor Brad Goodner, Hiram College and, along with additional experts they recruit, they teach this 3-day intensive hands-on workshop. From 2004 to 2008 the workshops were attended by approximately 100 faculty members and, through them, reached thousands of students with timely and relevant information on bioinformatics.
- JGI Presentations: Past and upcoming invited presentations include American Society for Microbiology Council on Undergraduate Education Meetings in 2007 and 2008; American Society for Biochemistry and Molecular Biology Meeting, 2009; Annual International Meeting on Microbial Genomics, 2006 and 2008; and the Meeting of the Australian Microarray and Associated Technologies Association Meeting 2009.
- Educational websites: Includes the IMG/EDU developed by JGI Genome Biology group in collaboration with JGI’s Education Program, and the IMG/ACT website developed by JGI. (www.jgi.doe.gov/education).

EEOC

Trainings for professionals on genetic discrimination and about GINA, Title II were presented at the following conferences or to the following organizations:

- Blind Lawyers Association, Washington Seminar (January 2005)
- SACGHS (June 2005)

- Annual EXCEL Conference for federal agency EEO and HR professionals and federal agency counsel (August 2007)
- Defense Contract Audit Agency (DCAA) Annual EEO Conference (November 2007)
- ABA Labor and Employment Section meeting (March 2008)
- Upper Midwest Employment Conference (May 2008)
- Technical Assistance Program Seminars (TAPS) in Denver and Albuquerque (June 2008)
- American Law Institute-American Bar Association Webcast (July 2008)
- WEB Employee Benefits Luncheon (July 2008)
- West Legalworks Webcase (August 2008)
- TAPS presentation in Richmond VA (August 2008)
- Department of Labor/National Association of State Workforce Agencies 19th Annual National Equal Opportunity Professional Development Forum (August 2008) New York City Practicing Law Institute (October 2008)
- ABA/Joint Committee on Employee Benefits Meeting (October 2008)
- National Association of ADA Coordinators National Conference in Las Vegas (October 2008)
- TAPS presentation for Trenton/NYC area (October 2008)

FTC

In its response to the 2008 survey, FTC indicated that genetics education and training were neither primary nor secondary goals of the agency. However, in cooperation with the FDA and CDC, FTC developed a fact sheet for consumers to educate them about the limitations of direct-to-consumer genetic tests. As of 2008, more than 16,000 copies of the print version of the consumer fact sheet have been distributed, and it has been accessed more than 18,000 times from the FTC website. The FTC stated the goals of this project were successful as “the fact sheet provided consumers with clear information to help them make well-informed decisions when considering whether to purchase an at-home genetic test. The FTC will continue to evaluate the need for consumer education about at-home genetic tests and will also monitor consumer-directed advertising of genetic tests with the goal of preventing consumer deception

HRSA

- Supports Area Health Education Centers (AHECs) that address health care workforce issues by exposing students to health care career opportunities that they otherwise would not have encountered, establishing community-based training sites for students in service-learning and clinical capacities, providing continuing education programs for health care professionals, and evaluating the needs of underserved communities. In 2003, the AHEC program was providing community-based continuing education programs to health professionals that included a component with genetics content to 9 of 46 participating U.S. medical schools.
- Maternal and Child Health Bureau programs:
 - Leadership Education in Neurodevelopment and related Disabilities (LEND)
 - Heritable Disorders Program, Regional Genetic & Newborn Screening Services (7 regional screening collaborative centers across the United States and the National Coordinating Center)
 - Consumer Initiatives for Genetics Resources and Services (CIGRS)
 - National Newborn Screening and Genetic Resources Center
- Bureau of Health Professions programs: A contract was awarded to the National Coalition of Health Professional Education in Genetics (NCHPEG) by an IAA among the NHGRI and ORD/NIH, CDC, and HRSA to promote health professional education and access to information about advances in human genetics. An additional IAA between HRSA and the NIH/NCI was for the development of Curricula in Genetics and genomics for Nurse Faculty Development.

- Presentations: Representatives of the Maternal and Child Health Bureau have presented at meetings of the American College of Medical Genetics, American Society of Human Genetic, the Association of Public Health Laboratories, the Genetic Alliance, and the National Coalition for Health Professional Education in Genetics. Staff of NCHPEG have presented at universities in Maryland, Michigan, Utah, South Carolina and Louisiana, and to organizations such as the American Institute of Biological Sciences, Office of Veteran Affairs, National Society of Genetic Counselors, Centers for Disease Control and Prevention Office of Public Health Genomics, the International Congress of Human Genetics, and the American Public Health Association.
- The Bureau of Health Professions has held meetings since 2000 on genetics, including an expert panel on Genetics and Nursing, 2000, an invitational meeting co-organized with the NHGRI in 2008 on The Genetics and Genomics Toolkit for Faculty, and additional meetings from 2003 to 2008 on pharmacogenomics, family history, risk assessment and communications of risk, genetics and religion, and genetics and common disease.
- Websites:
 - A portion of the Genetics/Genomic Toolkit for Faculty may be found at www.genome.gov/17517037, along with other resources, curricula, books and online courses on genomics and genetics for health professionals.
 - The IAA with NCHPEG has produced a website (www.nchpeg.org) that has steadily grown and improved as the number of educational offerings has increased. This website is also used to facilitate information sharing, host online surveys, and provide access to archived information and slide sets.
 - The Maternal and Child Health Bureau websites include the Genetics Services Branch website, regional genetics and newborn screening collaborative websites, the Sickle Cell Disease and Newborn Screening Program, GeneTests-GeneClinics, Community Centered Family Health History, March of Dimes Perinatal Data Center, and the National Newborn Screening and Genetics Resource Center website, among others. All these resources can be accessed at <http://mchb.hrsa.gov/>.
- Evaluation and Assessment projects:
 - The Division of Medicine and Dentistry contract allowed NGHPEG to collaborate with the Genetic Alliance on a survey of consumers of genetic services to assess their perceptions of the genetic competence of their providers.
 - HRSA's Division of Nursing participated with NIH/NCI and NHGRI to determine needs for nursing education in genetics and genomics.
- HRSA staff provide reviews of articles with genetics content for publications such as the Journal of Genetic Counseling, Genetics in Medicine, American Journal of Medical Genetics, and Quarterly Review of Biology, among others.
- NCHPEG staff participates in advisory boards and editorial boards with international, national and regional impact such as the Board of Directors/Personalized Medicine Coalition, CDC Advisory Committee on the Use of Family History in Pediatrics, Information and Education Committee/American Society of Human Genetics, and the editorial boards of the journals Community Genetics and Quarterly Review of Biology.

NIH

NIH collaborates with other agencies, such as HRSA, on programs and activities related to genetics education and training. HRSA/NIH workforce assessment activities were reported in the 2003 survey, including the HRSA/NIH co-funded study, *Assessing Genetic Services and the Health Workforce*, which was conducted by HRSA's National Center for Health Workforce Analysis. In addition, NIH and HRSA funded a national study of the delivery of genetics services, and the roles of geneticists and other health professionals in service delivery. This study described the existing and emerging health care models for

providing genetics services, the genetics specialist workforce, the role of primary care physicians and other clinicians in genetic services, and factors influencing the supply and demand for genetic services across the country.

Genetics education and training programs supported by individual NIH Institutes, Centers, and Offices include programs at the National Cancer Institute (NCI), National Human Genome Research Institute (NHGRI), National Institute on Aging (NIA), National Institute on Deafness and Other Communication Disorders (NIDCD), National Institute of Dental and Craniofacial Research (NIDCR), National Institute on Drug Abuse (NIDA), the National Library of Medicine (NLM), and the Office of the Director (OD).

NCI

- Advanced Cancer Risk Counseling Training for Nurses
- Clinical Cancer Genetics Education
- Genetics Short Course for Cancer Nurses
- A Cancer Genetics website that includes a cancer genetics overview, cancer genetics risk assessment and counseling, and information about the genetics of breast and ovarian cancer, colorectal cancer, medullary thyroid cancer, and prostate cancer. At this website, one can access links to materials developed and regularly updated by the PDQ Cancer Genetics Editorial Board specifically designed for health professionals. (www.cancer.gov/cancertopics/prevention-genetics-causes/genetics).

NHGRI

- Educational materials: educational web casts and interactive web-based learning tools were developed that fulfill recently adopted nursing competencies in genetics education
- Meetings: a Nursing Champions Meeting and a Primary Care Genetics Summit were held in 2009. The nursing meeting focused on development of a toolkit of genetics educational resources for nurse educators, and the identification of a suitable network of nursing “champions” with expertise in the translation of genetics into health care. The Primary Care Genetics Summit brought together key representatives of primary care physician organizations, such as the American Academy of Family Physicians, to discuss novel approaches to genetics education.

NIA

- Between 2003 and 2009 NIA supported one institutional training grant award aimed at training researchers and/or health professionals in topics related to genetics or genomics (5T32AG000258-10, Neurobehavior, Neuroendocrinology, and Genetics of Alzheimer’s Disease (AD)). This program provides postdoctoral training in clinical research regarding the neurobehavior, neuroendocrinology, and neurogenetics of AD and related dementias. In particular, the program focuses on training clinical researchers capable of translating critical findings from basic science into hypotheses regarding the etiology, pathophysiology, and treatment of AD. Participants also receive specialized training in two areas of study, neuroendocrinology and neurogenetics, which hold promise for increasing the understanding of the pathogenesis of AD and for developing new therapeutic approaches. The PI of the project is the only investigator with an entire research program focused on the interactions between insulin and AD, an emerging and increasingly important area of study. This area will become increasingly important in light of the obesity epidemic grows and as more and more Americans who are obese and diabetic grow older. By the end of the first funding cycle, 6 M.D.s and 6

Ph.D.s had received training through this program. Seven additional individuals are currently undergoing training.

- NIA's future initiatives in genetics education and training are a priority for the Institute, as articulated in the NIA Strategic Directions document, as well as the dissemination of scientific information to diverse audiences, including health professionals and the general public (see www.nia.nih.gov/AboutNIA/StrategicDirections/goal_f.htm).

NIDA

- Travel fellowships to the Jacksonville Short Course in Medical and Experimental Genetics
- An American Society of Human Genetics satellite meeting on Addiction Genetics Workforce Development and Collaboration. Presentations from the satellite session can be found at www.sei2003.com/nida/1014039/index.htm.
- Development of a NIDA Short Course on Genetics and Epigenetics of Addiction, presentations can be found at <http://drugabuse.gov/about/organization/Genetics/geneticsepigenetics/index.html>.
- Participation at the Community Anti-Drug Coalitions of America mid-year training institute conferences.

NIDCD

- Summer Program in Genetics for Audiology Faculty that included:
 - A needs assessment survey of existing graduate level training programs in audiology that incorporate genetics into their curriculum
 - The establishment of an Advisory Board to guide development of an educational program in genetics
 - The organization of three consecutive 7-day summer workshops targeted to faculty of audiology training programs and the development of an educational notebook for participants in the workshops to assist them in integrating genetics information into their own curricula
 - The establishment of a comprehensive evaluation component to determine the effectiveness of the educational program

NIDCR

- New Models of Dental Education initiative convened several panels – Genetics and Its Implications for Clinical Dental Practice and Education, held in 2007, and Practical Strategies for Genetics Education in Dentistry, held in 2005.
- Websites developed include the Genetics in Dentistry Case Simulator (www.dent.umich.edu/health/index.php), and the Genetics, Disease and Dentistry website, www.nchpeg.org/dental.
- Publications resulting from NIDCR genetics/genomic educational activities include:
 - Johnson, L., Genco, R.J., Damsky, C., Haden, N.K., Hart, S., Shuler, C.F., Tabak, L.A., and Tedesco, L.A. (2008). Genetics and its implications for clinical dental practice and education: report of panel 3 of the Macy study. *Journal of Dental Education*.72 (2 Suppl):86-94.
 - Dudliceck, L.L., Gettig, E.A., Etzel, K.R., and Hart, T.C. (2004). Status of genetics education in U.S. dental schools. *Journal of Dental Education*. 68(8):809-818.
 - Collins, F., and Tabak, L. (2004). A call for increased education in genetics for dental health professionals. *Journal of Dental Education*. 68(8):807-808.

NLM

- NCBI: More than one million users access NCBI daily, thousands of whom make use of NCBI's genomics or biomedical literature databases. The NCBI program, *Training and Support of NCBI Sequence and Genomic Information Resources*, provides training so that users can effectively and efficiently utilize NLM's online molecular biology and genomic resources. In addition, specific training courses at NIH, and periodically at sites across the country, have been offered to familiarize users with the range of genomics-related data at NCBI and train researchers in the operation and application of the analysis tools to molecular biology research. Interest in the courses offered nearly always exceeded the manpower available for teaching and, in each year of the program, from 2002 through 2007, approximately 6,000 participants registered for approximately 150 courses.
- Although NCBI reports a 10-percent increase in use of its data resources, it notes that future needs include providing specialized training on advanced tools (e.g., use of programming languages for large-scale data analyses) and more sophisticated tracking through web log analysis of how NCBI data resources are used. This analysis would help determine actual use of resources and how changes in web page presentation affect usage patterns.
- *Training and Support of NCBI Sequence and Genomic Information Resources*. This program addresses the continuing need for genomics education, especially as informatics becomes an increasingly greater component of molecular biology research. In addition to on-site training and support, NCBI manned exhibits and provided workshops at 20 to 25 scientific meetings per year. The program has been very successful – training not only approximately 30,000 university students and researchers, but also establishing a “train-the-trainers” program of approximately 50 specialists, primarily in medical libraries, who have established their own local programs.
- Lister Hill National Center for Biomedical Communications (LHNCBC): Because rapid advances in genetics research are impacting the health and medical needs of the public, the nonexpert citizen has an increasing need for information written in nontechnical terms. Recognizing this need, LHNCBC began development of the Genetics Home Reference website in 2001.²⁹⁷ This website addresses NLM's goal of advancing scientific knowledge in molecular biology by providing information about hereditary conditions and their underlying genetic causes in a consumer friendly format. Usage statistics for the website show a continuous increase in users over the five years since it was launched in 2003, with more than 2.7 million users in 2008. LHNCBC continues to investigate a variety of ways to make the results of the Human Genome Project more readily available to the public through the Genetics Home Reference website and will continue to add new content and new features. Existing materials are reviewed and updated on a regular basis.
- NLM Extramural Program: Since 1972, NLM has provided ongoing funding for *NLM University-based Biomedical Informatics Research Training Programs*. These training programs, conducted at various universities nationwide, address the need for training informatics researchers and practitioners in the representation, management, and delivery of biomedical knowledge. Genomics training is a small component of the informatics training, but a more prominent component in four programs that focus on bioinformatics. An assessment of the NLM training programs was completed in 2008 and is now under analysis by the program director.
- *The NLM University-based Biomedical Informatics Research Training Programs*. Training grants are provided to universities nationwide, however, specific institutions may change at each 5-year recompetition of the program. In 2008, 18 universities were receiving funding through this program including Columbia University, Harvard, Johns Hopkins, Oregon Health and Sciences,

²⁹⁷ National Library of Medicine. Genetics Home Reference: See <http://ghr.nlm.nih.gov/>. Accessed on November 19, 2009.

Rice, Stanford, Yale, Vanderbilt, and Indiana University, among others. An assessment of this program was conducted in 2008 in terms of basic goals (e.g., ability to obtain qualified trainees, ability of institutions to provide adequate resources and faculty, and career and publication outcomes of trainees).

OD

Programs funded by the Common Fund are known collectively as the NIH Roadmap, and administrative oversight of Roadmap programs is the responsibility of the OD Office of Strategic Coordination, within the Division of Program Coordination, Planning, and Strategic Initiatives. Two Roadmap programs that support genetics education and training are the National Centers for Biomedical Computing (NCBC)—led by NIDA and the National Center for Research Resources—and the Interdisciplinary Research (IR) program—led by the National Institute on Mental Health, National Institute of General Medical Sciences, National Institute of Diabetes and Digestive and Kidney Disorders, and NIDA. The following activities related to genetics and genomics are supported by these programs:

- National Center for Integrative Biomedical Informatics (NCIBI): This program establishes innovative education and training programs to educate NIH researchers on the use of NCIBI systems and tools to ensure best practices in use of experimental data and data analysis, and to facilitate data sharing and software dissemination.
- Centers for Computational Biology: This project provides interdisciplinary training for all levels of personnel by providing an integrated curriculum to foster a basic understanding of the correlations between genetic and molecular findings and systems biology, health and disease.
- Training—Neurodevelopmental Toxicology: This activity supports a new interdisciplinary post-doctoral program in neurodevelopmental toxicology in the Department of Environmental Health at the Harvard School of Public Health. The program provides a unique opportunity for students to receive training in the integrated disciplines of exposure assessment (for chemical, nutritional, social environmental factors), epidemiology, and risk assessment and to apply this training to the study gene-environment interaction in neurodevelopmental diseases. Participants receive cross-training in five required core training tracks, one of which is in genetics.
- Models and Technologies for Defining Phenotype: A 15-day training program that targets both graduate and post-graduate investigators in Genomics and Bioengineering Sciences at Wake Forest University. The program includes a significant component of didactic and laboratory training that addresses fundamental issues in genomic, physical, and imaging science research related to research design, core techniques, data interpretation, and strategies for successful integration of these types of research. Also, a program at Baylor School of Medicine is designed to train students in the following areas: (1) data acquisition—knowledge of the methods of genomics, proteomics and imaging; (2) computation—knowledge of mathematical and statistical algorithms, implementation of effective computer codes as well as an emphasis on methods of data warehousing in relational, deductive and other databases; and (3) data integration.
- Biobehavioral Intervention in Developmental Disabilities (BIDD): This activity supports a new interdisciplinary post-doctoral training program in BIDD at Vanderbilt University. The goal of BIDD is to provide postdoctoral trainees with an understanding of the relationships between behavioral phenotypes and biological markers of specific developmental disabilities, and to define the predictive value of these relationships for eventually developing and applying successful interventions. This program integrates knowledge in human behavior, genetics, and developmental neurobiology.
- Interdisciplinary Obesity Training: A post-doctoral program at the University of North Carolina at Chapel Hill that provides an innovative approach to education and training related to obesity and draws from multiple but interrelated disciplines of nutrition, epidemiology, physiology, health behavior and genetics.

- **Interdisciplinary Training for Autism Researchers:** A formal training program at the University of California-Davis that provides training in core competencies areas relevant to autism research including epidemiology, genetics-genomics, immunology, animal behavior, human behavior, human development, and neurochemistry-pharmacology.
- **Training in Pharmacoinformatics:** This activity at the University of Texas provides a comprehensive training program in the interdisciplinary area of pharmacoinformatics that capitalized on advances in bioinformatics, genomics, computing and other fields.
- **Training—Genetics and Complex Disease:** The goal of this program at the Harvard School of Public Health is to develop a cadre of young scientists who can participate at the intersection of molecular biology, epidemiology, and biostatistics and who become leaders in integrative and team approaches to understanding genetics and complex diseases in the public health arena.

In addition to Roadmap programs, genetics and genomics topics are included in the following courses and weekly Grand Rounds provided by the NIH Clinical Center:

Courses

- One lecture, annually, in the “Introduction to the Principles and Practice of Clinical Research” course is given by Christopher Austin, M.D., Senior Translational Research Advisor to the Director, NHGRI entitled, “Human Genome Project and Clinical Research”
- One lecture in the “Principles of Clinical Pharmacology” course given by David A. Flockhart, M.D., Ph.D., Chief, Division of Clinical Pharmacology; Professor of Medicine, Genetics and Pharmacology, Indiana University School of Medicine entitled, “Clinical Pharmacogenomics.”

Grand Rounds

- 1/28/09 “The Emerging Paradigm of Clinical Genomics: Technologic Developments and Clinical Implications,”-Eric Green, M.D., Ph.D, NHGRI Scientific Director and Leslie G. Biesecker, M.D., Chief, Genetic Disease Research Branch, NHGRI
- 05/28/08 “Menkes Disease” Steven Kaler, M.D., Clinical Director, NICHD
- 2/27/08 “Huntington Gilford Progeria”-Francis Collins, M.D., Ph.D., former NHGRI director and Wendy Inrone, M.D., NHGRI
- 1/16/08 “Genetics and Prevention Strategies in Type 2 Diabetes Mellitus”-William Knowler, M.D., Dr. PH, NIDDK and Jose Floresz, M.D., Massachusetts General Hospital
- 2/28/07 “Gaucher Disease and Small Molecular Screening”-Ellen Sidransky, M.D., NHGRI and Christopher Austin, M.D., NHGRI
- 11/15/06 “Genetics of Renal Cell Carcinoma”-Len Neckers, Ph.D., NCI and Marston Linehan, M.d., NCI
- 6/22/05 “Turner Syndrome in the Genomic Era”-Carolyn Bondy, M.D., NICHD
- 5/18/05 “Identification of Genes Underlying Parkinson Disease: Altering Clinical Practice and Understanding”-Andrew Singleton, M.D., NIA
- 10/29/03 “Clinical Research on Pediatric Genetic Disorders: Nephropathic Cystinosis and Osteogenesis Imperfecta,” William Gahl, M.D., Ph.D. and Joan Marini, M.D., Ph.D., respectively

NSF

Discovery Research Program projects include:

- *Developing the Next Generation of Middle School Science Materials – Investigating and Questioning our World through Science and Technology.* The primary objective of this project is the development of a comprehensive 6-8th grade curriculum which encompasses physics, Earth science, biology, and chemistry and that will lead to reading literacy in these topics. The project emphasizes professional development that supports teachers as learners, especially in terms of

learning scientific content and pedagogical tools and techniques. The efficacy of this project will be examined by comparing the performance, on standards-based assessments, of 8th grade students who participated in the 3-year curriculum to those who come from a comparable classroom with alternate materials.

- *The GENIQUEST (GENomics Inquiry through Quantitative Trait Loci Exploration with SAIL Technology): Bringing STEM Data to High School Classrooms.* GENIQUEST seeks to develop and test software which will put authentic biological data, along with powerful analysis tools, at the disposal of high school teachers and students. This software assists the framing of testable questions based on this data, at a level appropriate to the students' intellectual capacity, thereby increasing the knowledge of biology, data analysis, the nature of science, and computational biology.

Math and Science Partnership Program projects include:

- *The Geneticist-Educator Network of Alliances (GENA) Project.* A collaboration of the American Society of Human Genetics, the Genetics Society of America, the National Science Resources Center and the National Association of Biology Teachers, GENA provides tools to instruct, facilitate, and measure meaningful engagement of secondary STEM faculty through the outreach of geneticists at any level. The project seeks to develop a network of master Geneticist-Educator alliances to design strategies to maximize the effective and meaningful interaction between the geneticists and students. This project will serve as a model which may be adapted to other disciplinary scientific societies.
- *Baltimore Research and Innovations for New-STEM Partnerships.* The MSP-Start "BRAIN-STEM" project is a partnership between Morgan State University and Baltimore City Public School System which seeks to integrate mathematical and biological concepts suitable for high school courses, beginning with discrete mathematics and genomics. The project addresses the content and pedagogical needs of Baltimore school teachers, based on a needs analysis.

Course, Curriculum and Laboratory Improvement Program projects include:

- *Literature-Based Scientific Learning in Genetics.* Using constructivist learning and a collection of literature-based case studies, the project strives to promote scientific thinking, conceptual understanding and scientific information competence. The results for this experiential scientific learning project will be developed into an interactive, inquiry-based electronic textbook. The project may serve as a model for other disciplines and is expected to impact the training of future science teachers by involving graduate and undergraduate student assistants.
- *The New Genetics: Electronic Tools for Educational Innovation.* This project aims to create and evaluate an innovative set of educational materials. Using an interactive CD-ROM courseware, the project combines genetic and genomic science, technological concepts, environmental, agricultural and biomedical applications, and societal and ethical issues, thereby engaging student interest in the cutting edge of science. This project also expects to create informed citizens who understand science, are excited about the fruits of scientific research, and advocate for public support of scientific research and education. The model will be evaluated in several courses offered in numerous community colleges, a state university and a private university in California, providing a balanced evaluation under widely varying classroom conditions.
- *Pathways for New Laboratory Modules in Undergraduate Genetics and Cell Physiology Education: Characterization of Puerto Rican Cassava.* By introducing community-relevant research-based plant specific laboratory activities into upper division Genetics and Cell Physiology courses, the University of Puerto Rico seeks to expose approximately 700 Hispanic undergraduate students per year to modern molecular and cellular technologies. This project not only provides students with the confidence to trust in their abilities to learn, understand and implement techniques in modern science, but also leads to the sustainable management of cassava

Puerto Rican genetic resources, a real world application of the science students learn in a more traditional setting.

- *Project Laboratory in Genetics and Genomics*. By creating a new laboratory course, Brandeis University will provide “a myriad” of new experiences for its undergraduate biology students. Students will look at transposon mutation in *E. coli*, for example, and then integrate their findings with public domain genomic information resources to develop a web page for each gene investigated. The project provides students with greater access to a real research laboratory experience, as well as integrating the expertise of both research and teaching faculty who do not now collaborate on course design. Students are assessed before and after the course, for their level of mastery of basic cellular and molecular processes and for their attitudes towards, and understanding of, scientific research. In addition, students evaluate the value of various aspects of the course, to aid in its future refinements.
- *ComGen: The Community College Genomics Research Initiative*. This project exposes community college students to real-world research experiences in genomics. This reversal of normal research hierarchy will strengthen the pipeline of students engaged in scientific discovery and excited about STEM careers by including students before they have made a major commitment to a STEM field. This effort will be evaluated for its potential for replication at community colleges nationwide.

Advanced Technological Education Program project:

- *Innovating Biotechnology Education: Incorporating Novel Genomics Research in the Development of a True 2+2+2 Educational Pathway*. In response to a shortage in research-skilled laboratory technicians, Mesa Community College proposes a 2+2+2 program. This program is unique because it uses genomics research to prepare high school science instructors with skills and curriculum to prepare their students for the rigors of post-secondary degrees in biotechnology related fields. If successful, this model can easily be integrated into other biotechnology programs around the country.

NSF Scholarships in STEM projects include:

- *Proteomics and Functional Genomics Scholarship Program*. This scholarship program is designed for talented but financially needy students. The project aims to support more than 20 students who will eventually attend graduate school or obtain jobs in proteomics and functional genomics or related fields.
- *BHSU Integrative Genomics Transition Scholarship Program*. This program will provide support to 20 Master’s degree students in the emerging area of Integrative Genomics, as well as 10 scholarships for undergraduate biology majors with an interest in pursuing the Master’s degree in this area. Furthermore, the project is creating a pipeline to the Integrative Genomics program for Native American Indian students which should increase overall the number of Native American Indian STEM graduates pursuing advanced degrees.

Historically Black Colleges and Universities-Undergraduate Program project:

- *Targeted Infusion Project: Integration of Plant Genomics into the Undergraduate Curriculum*. This project will incorporate plant genomics into the undergraduate curriculum of the Plant Science and Biology departments. A Plant Genomics senior level course will be developed and newly designed genomics modules will be incorporated into several existing courses, thus preparing students in these courses for various careers in the biological sciences, and the burgeoning fields of genomics and bioinformatics. The teaching materials developed at one university will be widely disseminated through a variety of media.

Interdisciplinary Training for Undergraduates in Biological and Mathematical (UBM) Sciences project:

- *Undergraduate Training and Research in Applied Mathematics and Biological Sciences.* This project builds on an existing undergraduate major in Applied Mathematics-Biology. Student teams work on joint projects in physiology and genomics with faculty advisors and alongside graduate students and post-doctoral associates. This project provides students with a background in mathematics and biological science that will prepare them for future interdisciplinary graduate level programs.

Centers of Research Excellence in Science and Technology project:

- *CREST Center in Tropical Ecology and Evolution of Marine and Terrestrial Environments.* The goal of this program is to become a highly collaborative research center in tropical conservation biology and environmental sciences in Hawaii. The program consists of three interconnected subprojects: Evolutionary Genomics and Ecology of Local Adaptation and Speciation, Terrestrial Ecology, and Coral Reef Ecosystem. The NSF CREST Program will build on the current strengths of the center, especially an integrated research and education program that is building the STEM pipeline for students in Hawaii from K-12 through to undergraduate and graduate programs.

Integrative Graduate Education and Research Traineeship Program projects:

- *IGERT in Chemical Genomics: Forging Complementation at the Interface of Chemistry, Engineering, Computational Sciences and Cell Biology.* Chemical genomics uses small molecules to probe protein function in complex cellular systems. This approach offers a strategy which may fill in some crucial gaps in the study of functional genomics in plants by addressing the issues of overlapping gene function in gene families, lethal loci, and control of dosage and tissue/development specific application. The program will prepare graduates with skills for multidisciplinary research, acute awareness of the potential for their discoveries to address global food, health and environmental problems, of the ethical implications of their research, and with exposure to a variety of research environments in academia and industry.
- *IGERT: Predoctoral Training in Functional Genomics of Model Organisms.* The objective of this project is to initiate an interdisciplinary, inter-institutional degree program in Functional Genomics of Model Organisms. It is a collaboration of the University of Maine, the Jackson Laboratory, and the Maine Medical Center Research Institute. As it becomes clear that genome projects, regardless of the organism, will rely increasingly on the physical and computational sciences, interdisciplinary work and thinking becomes increasingly important. This program introduces a new educational paradigm, developed to train students to move freely among the disciplines needed to investigate genome function.

Informal Science Education Program projects:

- *Indonesian Origins: Genes, Languages and Culture video programs.* This “Communicating Research to Public Audiences” project will produce a quality television program that will showcase an interdisciplinary approach to the history of the peopling of the Indonesian archipelago, combining genetics, archaeology, historical linguistics and ethnography. The primary intended audience is American viewers of scientific documentary television programs, although it possibly could be shown in secondary schools and colleges.
- *The DNA Files III.* SoundVision Productions proposes to develop 5 one-hour radio documentaries, 5 five-minute features, and a website to inform a diverse public about important advances in genomics and related sciences. The project will offer audiences an awareness of the societal benefits of research and the intellectual tools to join in legal and social policy debates. A comprehensive outreach strategy will be implemented by 20 local public radio stations around the country in partnership with community organizations.