Genetics Education and Training of Health Care Professionals, Public Health Providers, and Consumers

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

Available for Public Comment Until June 30, 2010
A Note to the Public

The importance of professional and public genetics education and training was identified as a priority issue by the Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS or Committee), and recommendations to improve education and genetics and genomics literacy have been included in nearly every SACGHS report issued to date.1 The Genetics Education and Training Task Force composed of SACGHS members, *ex officios*, and *ad hoc* experts from the public and private sectors, was formed in 2007. The Task Force’s charge was to build on the Committee’s earlier work2 and identify the education and training issues pertinent to (1) point-of-care health professionals, (2) public health providers involved or likely to be involved in providing genetic services, and (3) consumers and patients. In focusing on these three groups, SACGHS acknowledges the importance of a wide range of professionals who are experiencing increasing exposure to genetics and genomics or play a role in assuring the proper application of genomic information and technologies to promote health. With the rapid proliferation of genetic technologies and the shift toward personalized health care, the Committee felt that focusing on the education and training needs of health care professionals working on the front lines of public health and health care delivery is of the highest priority, as is recognizing the need for an informed public.

The Committee acknowledges that there are other pertinent issues beyond education and training that influence the use of genetic and genomic 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 and genomics into their practice. Coverage and reimbursement of genetic tests and services, such as family history collection, influences the use of such services in clinical practice and thus may be an important priority for policymakers. New genomic technologies also have the potential to decrease health disparities, but continuing work is needed to assure that appropriate access and utilization of genetic services are made available to underserved populations while not deflecting resources that address basic health care needs.

SACGHS would appreciate input on whether the draft report fully captures the gaps and needs in genetics education and training for health professionals, public health providers, and patients and consumers and whether the draft recommendations target the issues and concerns identified in the report. Comments received by June 30, 2010, will be considered by SACGHS in the preparation of the final report that will be presented to the Secretary of Health and Human Services.

To submit comments to SACGHS, please e-mail them to Kathryn Camp at campkm@od.nih.gov or alternatively, comments can be mailed to Ms. Camp at the NIH office of Biotechnology Activities, 6705 Rockledge Drive, Suite 750, Bethesda, MD, 20892 (20817 when using delivery services other than the U.S. Postal Service) or faxed to 301-496-9839.

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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 sets out the following specific functions of the Committee:

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

Structurally, SACGHS consists of up to 17 individuals from around the Nation who have expertise in disciplines relevant to genetics and genetic technologies. These disciplines include biomedical sciences, human genetics, health care delivery, evidence-based practice, public health, 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.

Representatives of at least 19 Federal departments or agencies may also sit on SACGHS in an ex officio and nonvoting capacity. The departments and agencies are the Department of Commerce, Department of Defense, Department of Energy, Administration for Children and Families (HHS), Agency for Health care Research and Quality (HHS), Centers for Disease Control and Prevention (HHS), Centers for Medicare & Medicaid Services (HHS), Food and Drug Administration (HHS), Health Resources and Services Administration (HHS), National Institutes of Health (HHS), Office for Civil Rights (HHS), Office for Human Research Protections (HHS), Office of Public Health and Science (HHS), Department of Labor, Department of Veterans Affairs, Equal Employment Opportunity Commission, and Federal Trade Commission.
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Preface

The Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS or Committee) has consistently recognized the importance of professional and public genetics education and training. Recommendations to improve education and genetics and genomics literacy have been included in nearly every SACGHS report issued to date. In 2004, the Committee issued a formal resolution that was conveyed to the Secretary of Health and Human Services regarding the critical importance of appropriate and adequate training and education in genetics and genomics for all health care professionals and the public. In its 2004 and 2008 priority-setting processes, SACGHS ranked professional and public education as a high priority. In November 2007, SACGHS convened a roundtable to identify the need for a task force on genetics education and training to build on the Committee’s earlier work. The focus and scope of the task force were discussed at three subsequent SACGHS meetings.

The Genetics Education and Training Task Force was formed, and in consultation with the full Committee, defined its scope and developed a work plan and framework for analysis 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, 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. With regard to consumers and patients, the Task Force focused on identifying their education needs to assist them in informed decisionmaking about the use of genetic and genomic services and to enhance their understanding and use of genetic information with regard to risk identification and management, prevention, diagnosis, and treatment of disease.

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 consumers and patients). With the rapid proliferation of genetic technologies and the shift toward personalized health care, the Task Force felt that focusing on the education and training needs of health care professionals working on the front lines of public health and health care delivery is of the highest priority, as is recognizing the need for an informed public. Two methods were used to gather information to inform this report: (1) a literature review of research relevant to professional and public education and training in genetics and genomics; and (2) surveys of select major organizations, groups, and individuals with

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responsibilities across the continuum of health professional education and public health, and those that
provide advocacy for consumers and patients

In focusing on these three groups, the Task Force and SACGHS acknowledge the importance of a wide
range of professionals who are experiencing increasing exposure to genetics and genomics or play a role
in assuring the proper application of genomic information and technologies to promote health. The
Committee’s future plans call for an assessment of whether this report’s findings and recommendations
may also apply to a broader constituency, such as specialty health care professionals, laboratory workers,
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.

The Committee acknowledges that there are other pertinent issues beyond education and training that
influence the use of genetic and genomic 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 and genomics into their practice. Coverage and
reimbursement of genetic tests and services, such as family history collection, influences the use of such
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genomic technologies have the potential to decrease health disparities, but continuing work is needed to
assure that appropriate access and utilization of genetic services are made available to underserved
populations while not deflecting resources that address basic health care needs. These related issues are
also discussed in this report.
I. Background and Scope

The Human Genome Project was completed in 2003, resulting in a delineation of the complete sequence of the human genome. The sequence data helped advance research into the genetic basis of disease, including common, multifactorial diseases. Expanded genetic and genomic knowledge and technologies are now leading to new approaches to the diagnosis of some common, chronic diseases and conditions. Genomics also forms the basis of the growing field of pharmacogenomics, the study of how individual differences affect drug response. These developments are moving genetics beyond a clinical specialty focused on rare, inherited diseases and chromosomal disorders, yet, many health care and public health professionals lack sufficient knowledge about the application and interpretation of genetics in the clinic or in the community.

Concerns have been raised for nearly four decades about how best to translate, interpret, and deliver complex genetic information to health care professionals and consumers. As the discipline of clinical genetics arose in the 1950s, there was also recognition that nongenetics professionals would eventually also be needed to play a role in providing genetic services to patients. In a 1975 report on the emerging field of genetic screening, the National Academy of Sciences (NAS) 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.

Primary care has been 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 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.”

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11 The term “genetics” commonly refers to the actions of single genes, whereas the term “genomics” often is used to describe the interactions of genes with each other and with the environment. 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.


Two decades later there was considerable debate regarding the role of nongenetics professionals in genetic service provision. Greendale et al.\textsuperscript{19} 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.\textsuperscript{20} argued that implementation of “genomic health care” would necessitate collaboration and cooperation of all health professionals. Increasingly these same concerns have turned toward public health providers, as genomics moves into population-based applications, and toward the public, as consumers gain the ability to purchase their own genetic tests in the marketplace. Recent legislative proposals, starting in 2007, 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 (CE) programs, and promote competencies across clinical, public health, and laboratory disciplines.\textsuperscript{21} However, no bills have been passed that actually provide funding for such programs.

### Technological Advances

In 2010, several trends are moving the use of genomic technologies into the clinic or encouraging the public to access technologies via the marketplace. Thus, improved education at all levels is imperative. For example, as a result of large-scale genome-wide association studies, data is being organized and shared to translate research information into clinical knowledge. In 2003, the National Human Genome Research Institute (NHGRI) launched a public research consortium named EnCODE, the Encyclopedia of DNA Elements.\textsuperscript{22} The goal of the project is to identify all functional elements in the human genome sequence and to determine how genes interact so that preventive and therapeutic strategies can be developed. Other relevant research initiatives are the 1000 Genomes Project, the Electronic Medical Records and Genomics (eMERGE) Network, and the Cancer Genome Atlas. The 1000 Genomes Project is an international effort launched in 2008 to establish a catalogue of human genetic variation based on the sequences of at least 1,000 anonymous participants from a number of different ethnic groups.\textsuperscript{23} The eMERGE Network is a consortium formed to develop, disseminate, and apply approaches to research that combine DNA repositories with electronic medical record systems for large-scale high-throughput genetic research.\textsuperscript{24} The Cancer Genome Atlas is focused on the genetic causes of human cancer.\textsuperscript{25} All of these efforts are accelerating our ability to make clinical sense of genomic data, and ultimately the contribution of genetic variation to common, chronic diseases that burden our health care system.

Aside from the need to understand the clinical importance of genetic findings, the cost of sequencing individual genomes is rapidly decreasing, which could result in greater demand by consumers for this information, placing a greater interpretation burden on health care professionals. Within the last decade, the price of sequencing has dropped from $300 million to $48,000 by early 2009\textsuperscript{26} and $5,000 by late

As next-generation sequencing methodologies become available, costs will decrease further. In addition, NHGRI has funded projects aimed at bringing the cost down to $1,000 per genome. That dollar amount could attract some “customers,” who might equate the cost of whole-genome sequencing as comparable to other medical tests or procedures. With the $1,000 genome, personalized health care moves closer to reality, and the ability to link genomic data with electronic health records (EHRs) raises new possibilities for clinical care as well as for research. However, lower costs alone are not sufficient. Personalized health care will become a reality when alterations in a person’s genome can be causally attributed to an increased risk of developing a chronic disease and something can be done to treat or prevent that disease. Managing that information at the individual, clinical, and population levels requires a greater understanding of genetics and genomics.

**Moving from Genetics to Genomics**

The field of medical genetics is on the brink of a paradigm shift for how genetic tests and genetic information can be applied in clinical practice and disease prevention. Guttmacher and Collins viewed genetics “as the study of single genes and their effects” and genomics as “the study not just of single genes, but of the functions and interaction of all the genes in the genome.” Genetics has and will continue to be applied in the clinical setting in the context of individual, rare, single-gene disorders, which account for the vast majority of genetic tests currently available. However, the greatest potential benefits of applications of genomics will take into consideration the complex relationships among genetic variation, the environment, and disease, providing diagnostics and therapies for complex, common disorders such as cancer, heart disease, diabetes, and mental illness. Realizing this potential will require a 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.

Importantly, it is hoped that advances in genomics will provide new opportunities for prevention, traditionally at the heart of public health, both at the individual level and through population-wide interventions.

“Understanding genetic effects and gene-environment interactions in disease processes could produce recommendations that certain subgroups avoid defined exposures or receive targeted interventions. Stratification by genotype or family history already provides a means for tailoring screening tests for early disease detection (e.g., colorectal cancer screening in genetically susceptible persons), and this paradigm is likely to be extended to early detection of other conditions.”

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

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with disease, interactions with other risk factors), the skills and tools of public health will be increasingly important.

Advances in identifying the genetic underpinnings of chronic disease are rapidly changing the way we think about treating disease and promoting health.\textsuperscript{34} Understanding genetic and genomic influences can affect treatment in a number of ways, for example, through development of targeted medications for specific genetic alterations in an individual that are associated with drug efficacy and/or toxicity (pharmacogenomics); altered needs for specific dietary constituents such as increased folic acid in the presence of mutations in methylenetetrahydrofolate reductase; and increased screening when the presence of specific mutations increase the risk of developing disease such as colorectal cancer. If advances in genetics and genomics are to be effectively applied to improve disease outcome and promote health in the population, research that yields new insights into the pathophysiology of disease must be followed by clinical applications that lead to improved outcomes. These outcomes cannot be achieved without a better educated health care workforce.

The patient and consumer also will play an increasingly critical role in achieving the goals of genetic medicine. While there are gaps and barriers to successful integration of genomics into clinical practice (e.g., clinical utility, privacy, developing an evidence base, developing cost models),\textsuperscript{35} the translation of research discoveries will result in health promotion only if the translation is successfully adopted into clinical practice and leads to individuals adopting health-promoting behaviors. For example, a person who is found to be at increased risk for developing type 2 diabetes, whether this knowledge comes from family history or identification of disease-contributing single nucleotide polymorphisms (SNPs), will not benefit from this information unless he or she is willing to make behavioral changes that minimizes risk.\textsuperscript{36,37} Because knowledge is considered a prerequisite of health behavior, consumers and patients will need knowledge to benefit from advances in genetics and genomics. Additionally, much needed public participation in debates surrounding science and technology, which would include the use of genetic and genomic technologies and services, requires adequate knowledge and understanding.\textsuperscript{38} The availability of genetic and genomic tests that consumers can purchase without the involvement of their health care provider adds urgency to public education efforts.

The Need for a New Model for Delivering Genetics and Genomics Information

With the increased integration of genetics and genomics into a broader health care network, consumers and patients will be using results of genetic technologies increasingly in their own health care decisionmaking. Patients and consumers, health care professionals, and public health officials are challenged to keep pace with this dynamic and rapidly evolving field. The emerging understanding of the role of genetics and genomics in common disease is increasing the need for knowledge and understanding of risk assessment, genetic diagnoses, appropriate treatment approaches, and communication in professional and public education. The accelerated growth of DTC genetic and genomic services highlights the importance of adequate education for consumers to ensure informed decisionmaking. To

realize the benefits of genetic and genomic technologies and guard against the potential for harm, educating health care professionals, the public health workforce, and the general public is critical.

Improving and expanding genetics education for health care professionals, public health providers, and consumers 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, during “teachable moments.” This approach, while continuing to be essential at the individual health level, will not address the much larger fraction of the population with moderately increased risk for various multifactorial diseases with genetic components (e.g., cancer, cardiovascular disease, and diabetes). Effective interventions based on genetic information will rely on the public’s understanding of the meaning and interactions of susceptibility genes of uncertain penetrance with other risk factors. In addition, with the expansion of screening and early detection technologies for many common chronic diseases, the public health workforce will become increasingly integral to both community education and service provision. Moreover, expanded newborn screening increases the need for primary care providers to be educated about the critical nature of a positive result and emphasizes the need for just-in-time resources for referral and patient management. Parents and families also have educational needs related to newborn screening not only if their child has a positive screen and requires follow-up, but also as new issues emerge, such as increasing rates of false positives as more tests are added to the newborn screening panel and the need for parental consent related to the storage and use of residual blood spot specimens.

Thus, a new model for applying genetics to improved health requires a system in which health care professionals, public health providers, and consumers are well informed and able to interact and connect with each other as appropriate. Cooperation and collaboration in processing, applying, and interpreting genetic information will be essential. Without educated health care professionals and consumers, society will not benefit from genetic advances. Without an educated public health workforce, opportunities will be lost for deploying prevention and early detection programs for a wide variety of chronic diseases. And, without an informed public, patients and consumers may make poorly informed choices, or fail to seek needed professional health services.

The Work of the Genetics Education and Training Task Force

To 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 time period 2003-2009. The search included databases from the fields of medicine, science, education, social science, and psychology. It was limited to English-only articles and did not include meeting abstracts. Some unpublished literature was captured by searching the dissertations abstract database. (See Appendix A-1 for data bases searched and search terms used.) Additional literature was reviewed as it became available in 2010.

In addition, SACGHS collected data from Federal agencies and selected organizations with responsibilities across the continuum of health professional education, public health, and consumer and patient advocacy to obtain information regarding their activities in genetics education. The results of surveys and interviews are provided in the following chapters.

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II. The Status of Education and Training of Health Care Professionals

A. Introduction

Health care professionals, particularly those working at the point of care—physicians, nurses, physician assistants, genetic counselors, and pharmacists—must be adequately educated and trained in genetics to promote the effective translation of new genetic knowledge into practice, enhance access to genetic technologies, and ensure that these technologies are appropriately used. Over the past several decades, considerable research has examined levels of knowledge and understanding of genetics in a variety of groups. These studies suggest that health professionals rate their knowledge of genetics as fair to poor; and that a large majority test poorly on knowledge and interpretation of genetic data. Without additional educational efforts, the educational gap will only grow as new applications in genetics and genomics appear across the health care and public health landscape. Although some studies have shown that health professionals’ understanding of genetics has improved over time, more recent research shows that health care professionals still lack the knowledge needed to make optimal use of genetic information.

B. Literature Review

A significant body of literature from the United States and abroad highlights the nature and lack of genetics education of health care professionals as factors limiting integration of genetics into health care. McInerney summarized these contributing factors as:

Crowded curricula. All health-related disciplines face the challenge of including more information in a finite time.

Misconceptions about genetics. Many health care providers still believe that genetic medicine is defined by rare, Mendelian disorders and circumscribed by pediatrics and obstetrics, when in fact genetics is increasingly concerned with the common, chronic diseases that are the daily focus for most health professionals.

Lack of knowledgeable faculty. Many institutions that train health care professionals do not have sufficient faculty with genetic training to teach basic genetics or its applications to patient care.

A disconnect between the basic sciences and clinical experiences during training. Even when students training for health-related careers receive genetics instruction during basic science training, their subsequent clinical training often fails to incorporate genetic perspectives, largely because those responsible for the clinical training do not themselves have substantive education and expertise in genetics.

Failure to integrate genetics across the curriculum. Genetics instruction is poorly integrated into all relevant courses.

Inadequate representation of genetics on certifying exams. Testing often drives curricula, and the certifying exams for most health professionals include little, if any, genetics content.

A dearth of genetics professionals. The low numbers of medical geneticists and genetic counselors in the United States and elsewhere limit the provision of genetic services directly and, further, limit the extent to which other providers have formal and informal access to genetics expertise.

A lack of management and referral guidelines in genetics. The paucity of evidence-based guidelines related to genetic medicine likely hinders the attention genetics receives from providers on a day-to-day basis and raises questions for providers about the clinical utility of genetics.

Lack of knowledge and confidence about genetics among primary care providers. Surveys of health professionals demonstrate a lack of basic knowledge about genetics, and often a lack of confidence to deal with genetics-related issues that arise in the clinical setting.

While the gaps in knowledge and confidence of health care professionals primarily have been identified by genetics or related discipline research, studies of consumers support this premise. A survey of individuals and families with genetic conditions resulted in responses from 5,915 persons, 64 percent of whom reported that they received no genetics education materials from the provider they deemed most important to management of the genetic condition. Overall provider knowledge of the respondents’ genetic conditions was rated poor by an average of 32 percent of consumers.55

The Genetic Professional Workforce

Recent health care professional workforce analyses performed by the Health Resources and Services Administration (HRSA) show that in the United States there are currently 817,000 physicians (763,200 Medical Doctors and 54,300 Doctors of Osteopathy),56 2.9 million Registered Nurses (376,901 with master’s or doctorate degrees and 141,209 Nurse Practitioners),57 66,000 physician assistants,58 226,000

pharmacists, 59 and 2,448 certified genetic counselors. In 2005, a federally funded study concluded that
the medical genetics workforce does not appear sufficient to meet expected patient care needs for clinical
genetic services in the next five to 15 years due to several factors including the mismatch between the
increased need for genetic services and the size of the genetics workforce and data showing that young
physicians are not entering the field of genetics. 60 Because many states and areas of the nation already
have too few genetics physicians to meet current demand, the absence of major workforce expansion may
leave some patient subgroups with new access problems, particularly patients with inborn errors of
metabolism and those living in rural areas. 61

Setting aside the steady entry of genetics into routine care, these deficiencies become even more
concerning in light of expanded newborn screening programs. It is recognized that clinicians generally are
unprepared, and educational efforts that focus on screening procedures and referral practices will be
critical to maximize this life-saving public health program. 62

The 2008 Newborn Screening Saves Lives Act (Pub. L. No. 110-204) 63 recognized and renewed the
national commitment to newborn screening as a critical public health program that saves and improves
children’s lives. Expanded newborn screening programs are expected to detect 10,000 affected infants
annually, with many needing chronic disease management. Yet, there are only 200 physicians specialized
in the diagnosis and management of patients with inherited metabolic disease in the United States, and
some of the conditions detected through newborn screening are so rare that only a handful of experts exist
with experience in their management. Physicians who have such expertise are least able to expand
services 64 and three quarters reported that their practices are “nearly full,” with about one quarter
reporting new patient appointment wait times of more than three months.

Thus, although the need for clinical genetic services has increased, and continues to increase, the ability
of the genetics-specific health care workforce—which includes medical geneticists, genetic counselors,
and other health care workers such as nurses who provide genetic services—is not sufficient to meet this
need. In a survey of graduate medical education conducted in 2008-2009, it was found that only 76
residents were enrolled in a medical genetics subspecialty program. 65 This statistic suggests that the large
and diverse group of health professionals providing services at the point of care must be enlisted to
provide appropriate genetic services and information.

The Critical Shortage of Medical Geneticists

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

59 Health Resources and Services Administration. The Adequacy of Pharmacist Supply: 2004 to 2030. See
60 Cooksey, J.A., Forte, G., Benkendorf, J., and Blitzer, M.G. (2005). The state of the medical geneticist workforce: findings of
Expanded newborn screening in Texas: a survey and educational module addressing the knowledge of pediatric residents.
64 Cooksey, J.A., Forte, G., Benkendorf, J., and Blitzer, M.G. (2005). The state of the medical geneticist workforce: findings of
302(12):1357-1372.
areas. Genetics professionals include physician geneticists, those with PhDs, genetic counselors, and genetics nurses and are collectively referred to as “medical geneticists.” The 1,326 physician geneticists certified between 1982 and 2009 represent less than 0.3 percent of the more than 817,000 physicians in the United States. It is not known how many of these individuals are currently practicing. With the exception of genetics counselors, the numbers of medical geneticists achieving certification and entering the workforce has, at best, remained flat for the past 15 years. Although workforce planners are reluctant to estimate an adequate or an ideal number of medical geneticists needed to provide quality care, it is worth noting that historically, physician geneticists in the United States have devoted only 50 percent of their time to direct patient care; most are trained in pediatrics; practice trends favor increasing specialization as opposed to a general genetics practice or one that would accommodate more than patients with rare diseases; nearly three-quarters of practices are full and unable to increase patient load; and current practice paradigms are inefficient in comparison with other medical specialty models, with physician geneticists reporting on average that they are able to see only seven new and six follow-up patients per week.

In 2004, the Royal College of Physicians in London estimated a need for four full-time medical geneticists per one million people. Based on a current U.S. population of roughly 307,919,500, the United States needs approximately 1,232 full time equivalents (FTE) medical geneticists. According to ACMG data, there are currently 540 FTE medical geneticists in the U.S. workforce, 44 percent of the number needed. However, the United Kingdom estimate does not take into account the burgeoning demand for genetic evaluation and genetic testing as the field experiences a revolutionary acceleration in the delineation of rare and ultra rare diseases and as the demand for adult genetics services and testing in various sectors, including oncology, cardiology, neurology, and pharmacogenetics, increases. The U.K.’s National Health Service, meanwhile, has apparently recognized this impending crisis and has dedicated resources to fill the gap. Select general practitioners are encouraged to train under consultant geneticists in order to increase the provision of genetic services, and U.K. laboratory geneticists are being re-trained to assume new clinical roles as genetic test advisors for general practitioners. In Canada, there is one clinical geneticist per 375,000 individuals, a figure that is nearly identical to that in the United States and yet waiting times for routine referrals for genetic services range from several weeks to two years, with the consequence that referring physicians often do not seek consultations.

Furthermore, the medical geneticist workforce in the United States does not match racial and ethnic demographics. In 2003, only 13 percent of medical geneticists identified themselves as members of an

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68 Personal communication, Judith Benkendorf, M.S., CGC, Special Assistant to the Executive Director, American College of Medical Genetics, April 1, 2010.
ethnic or racial minority.\textsuperscript{76} Under-representation of diverse populations in the health care workforce has been cited as a primary barrier to mitigating health care disparities. In summary, the medical genetics workforce in the United States appears to be several orders of magnitude short of adequate at the dawn of the genomic age of medicine.

Clinical Translation of Genetics

To provide background for this report, SACGHS searched the literature in an effort to answer the following questions related to the need to prepare the health care workforce for clinical translation of genetics:

1. What are the attitudes and working knowledge levels of health care professionals regarding genetics?
2. What essential skills and knowledge in genetics are required for competent clinical practice?
3. What academic preparation, licensing, and accreditation processes are appropriate for health care professionals concerning genetics? What continuing education (CE) mechanisms are needed, and are genetics content required to maintain active licensing or certification? What evidence exists about the effects of targeted CE efforts?
4. Are health care professionals’ genetics practice standards and clinical competencies reflected in current clinical practices? What are the challenges and barriers to health care professional use of genetics?

Attitudes and Working Knowledge Levels of Health Care Professionals Regarding Genetics

The goal of incorporating genetic knowledge into clinical practice is not new.\textsuperscript{77,78} Shortcomings have been noted for nearly two decades in health care professional knowledge of genetics and use of genetic tests, and the need for integrated genetic instruction across curricula of all health care subspecialties has been advocated for some time.\textsuperscript{79} In the United States, health care providers across a wide range of clinical specialties demonstrate lack of genetics knowledge and recognize the need to incorporate genetics into their practice.\textsuperscript{80,81,82,83,84,85,86} Health care professionals not only lack understanding of genetics as necessary for direct patient care but also are not familiar with genetics as related to health policy, legal

Lack of genetic knowledge among physicians exists not only with complex, multifactorial conditions, but also with traditional and well-documented Mendelian conditions such as autosomal dominant hereditary cancer syndromes. An assessment of U.S. physicians regarding hereditary breast, ovarian, and colorectal cancer genetics identified limited knowledge about key genetic concepts. In one study, a random sample of 1,251 licensed physician members of the American Medical Association was surveyed across four groups of primary care (internal medicine, general practice, family practice, and obstetrics) and specialty care providers (oncology, general surgery, urology, and gastroenterology) about hereditary cancers likely to be encountered in their clinical practice. Among the findings were that (1) only 37.5 percent of respondents correctly recognized that hereditary breast cancer due to mutations in $BRCA1$ and $BRCA2$ genes could be transmitted through fathers, (2) only 33.8 percent of respondents correctly identified that less than 10 percent of female breast cancer patients carry $BRCA1$ or $BRCA2$ mutations, and (3) only 13.1 percent of respondents knew that penetrance of hereditary nonpolyposis colorectal cancer is more than 50 percent in mutation carriers.

A survey of psychiatrists’ working knowledge, opinions, and practice patterns found that of 352 psychiatrists, 83 percent felt it was their job to discuss genetics and genomics with patients, and while 58 percent discussed genetics with patients, less than 25 percent felt able to do so competently. From the same study, few psychiatrists could correctly answer more than half of the questions concerning basic genetics knowledge, and only 15 percent felt that medical training adequately prepared them to address genetics questions from patients. Many psychiatrists underestimated the contribution of genetic factors to common, multifactorial diseases regularly seen in their clinical practice. Finn et al.’s 2005 analyses found a disconnect between a practitioner’s understanding of genetic contributors and actual scientific evidence, for example, estimating that genetic factors account for 30 percent of cases of schizophrenia, when the scientific literature estimates such a contribution in 70 to 86 percent of cases.

White et al. examined the genetic service referral patterns of 284 family physicians. For a clinical scenario not warranting referral for genetic counseling and testing per U.S. Preventive Services Task Force guidelines, 92 percent of participants were referred for genetic testing services and 50 percent were referred for genetic counseling anyway. Education was recommended to maximize appropriate referrals and improve the role of clinician-patient relationships in referral decisions.

These patterns affect nurses as well. A survey of 46 advanced practice nursing students (from nurse practitioner (N.P.) and nurse anesthesia programs) found that 56 percent of respondents had minimal or no knowledge of pedigree construction and less than 10 percent indicated a high level of knowledge of six basic genetic terms such as meiosis and DNA structure and function.

The patient perspective confirms these health care provider self-reports and objective knowledge assessments. A web-based survey of 5,915 patients receiving health care services from approximately 25

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types of health care providers found that 64 percent received no genetics education materials.\textsuperscript{91}

Approximately 50 percent of respondents either had genetic conditions themselves, or had family

members with genetic conditions and were aware of their risk for genetic diseases through membership in

genetic advocacy organizations. Health care providers identified as being most often involved in the

management of the survey respondent’s health conditions included family physician and primary care

providers (54 percent), pediatricians (42.7 percent), cardiologists (35 percent), neurologists (31.8

percent), ophthalmologists (34.8 percent), and physical therapists (33 percent).

Few studies have evaluated genetic education and training needs of pharmacists. A 2003 study assessed

community pharmacists’ confidence in their knowledge about the Human Genome Project, genetic
testing, and pharmacogenomics\textsuperscript{92} and found less than 50 percent with confidence in these topics. In 2004,
a survey questionnaire administered to pharmacists assessing attitudes relative to genetic testing revealed

that half agreed that drug development will be faster due to genetic testing and 60 percent either disagreed

or were neutral about the need for patients’ genetic information to be available to improve drug

dispensing.\textsuperscript{93}

Genetics knowledge deficiency in health care professionals also extends to policy and legal matters. In

2004, a California State-based survey of 191 physicians and 80 nurses (registered nurses (R.N.s and

N.P.s) found that 58.3 percent were misinformed about the existence of 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.\textsuperscript{94} More recently, a study of 1,181 physicians

and nurse practitioners demonstrated that although 96 percent of respondents viewed genetic testing as

useful in ascertaining genetic cancer risks, more than 60 percent were unaware of the newly passed

protective genetic anti-discrimination law.\textsuperscript{95}

A survey of 428 medical students’ attitudes toward genetic testing of children for heritable conditions
demonstrated that personal understanding and use of genetics was dependent on previous education.\textsuperscript{96}

Even if access to rigorous genetic education and CE exists, health care professionals’ personal attitudes
influence clinical actions. For example, a recent study evaluated 1,121 primary care physicians regarding

their willingness to use pharmacogenetic testing to tailor smoking treatments.\textsuperscript{97} Despite strong evidence

of the role of genetics in smoking cessation treatment, surveyed physicians purposely avoided use of the

term “genetic” testing. If the smoking treatment was described in nongenetic terms (i.e., serum protein
detection), there was greater enthusiasm and interest in using the smoking test.

Adoption of Genetics and Genomics Clinical Competencies for Health Care Professionals

\textsuperscript{91} Harvey, E.K., Fogel, C.E., Peyrot, M., Christensen, K.D., Terry, S.F., and McInerney, J.D. (2007). Providers’ knowledge of


Pharmacists Association}. 44(3):399-402.

\textsuperscript{94} Blazer, K.R., MacDonald, D.J., Ricker, C., Sand, S., Uman, G.C., and Weitzel, J.N. (2005). Outcome from intensive training in

\textsuperscript{95} 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.,


among clinicians. \textit{Genetics in Medicine}. 10(9):691-698.


Primary care physicians’ willingness to offer a new genetic test to tailor smoking treatment, according to test characteristics.
The National Coalition of Health Professional Education in Genetics (NCHPEG) identified overarching clinical competencies for all health care professionals and various professional groups have developed clinical competencies for their individual disciplines. For example, pedigree assessment is incorporated into many competency recommendations. When properly conducted, family history is widely regarded as a mechanism by which to detect familial transmission of hereditary diseases, such as familial cancer syndromes and common multifactorial diseases. Its role is recognized as being so important that public service announcements have been created, aimed at engaging the U.S. public in understanding how their family history can be used to guide health care decisionmaking.

While national public health campaigns are encouraging individuals to bring their family histories to their health care providers, family history proficiency is not a competency required in order to graduate from a medical education program. Clinicians are hesitant to incorporate use of family history assessment due to time constraints, questions about clinical utility, beliefs of unreliability, and absence of meaningful financial reimbursement. 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. This trend is present in nursing students as well; only 22 percent of 46 Advanced Practice Nursing students in N.P. programs felt they could draw a family pedigree. Despite encouragement to use family history in primary care, the National Institutes of Health (NIH) State-of-the Science Conference on Family History and Improving Health, held in August of 2009, concluded that, while family history plays an important role in medicine, more research is needed before a systematically collected family history for common disease will become an evidence-based tool in primary care settings. Other genetics skills recommended by consensus panels are encountering similar challenges in accurate clinical uptake 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 breast cancer (i.e., BRCA1 and BRCA2) mutation
testing, only 13 percent of internists, 21 percent of obstetricians, and 40 percent of oncologists could
correctly answer four basic genetics concept questions. In the same study, although greater genetic
knowledge influenced frequency of discussing the BRCA genetic test with patients, 54 percent of
oncologists operating on inaccurate genetics concepts discussed genetic testing with their patients and
presumably, made health-related decisions regarding their care.

More recently, 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.
However, there were clear differences in patterns of genetic service referrals, with providers serving
minority populations being significantly less likely to order testing or issue referrals.

Extending beyond well-validated applications, complexities will be considerably greater for management
of chronic, multifactorial diseases. A recent comprehensive review of the literature shows little data
available to health care providers interested in using genetics to manage adult-onset conditions. These
reviews suggest that until health outcome data on genetic technologies exists and there are clear and
accessible education mechanisms for current health care providers and students, use of genetics as
outlined in competency statements is not likely to reach the bedside without further strategic support.

**Essential Skills and Knowledge in Genetics and Genomics Required for Competent Clinical
Practice**

Although several disciplines have overarching clinical practice competency standards, numerous
professional societies and organizations spanning Federal, academic, private, and public domains have
developed recommendations according to overarching practice requirements and clinical subspecialty.
Professional competencies are discussed according to Hundert and Epstein’s 2002 definition:
“Competency is 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.”

Setting the stage for genetic competency in clinical practice, NCHPEG first developed a list of necessary
competencies for health care professionals in 2001 in expectation of the completion of the Human
Genome Project. Initially, the list included 44 competencies but was revised in 2005 and again in 2007,
to its current 18 competencies (see Appendix A-2 for complete wording) reflecting the rapidly changing
biomedical landscape and recognition of the need to focus competencies on measurable outcomes. Three
minimum expectations for health care professionals across all clinical practice settings advise them to
identify when professional development related to genetics and genomics would be beneficial, understand
the social and psychological implications of health-related genetic information on patients and families,

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nongeneticist physicians who do or do not discuss or order BRCA testing? Genetics in Medicine. 5(2):99-105.
116 National Coalition for Health Professional Education in Genetics. (2007). Core competencies in genetics for health
and know how and when to make a genetics referral. More specific recommendations within these three content areas are provided in Appendix A-2.

Similar to NCHPEG’s overarching competencies, various practice recommendations and/or genetics-specific clinical competencies have emerged. These practice recommendations largely fall into two broad categories and are health professional discipline-specific or issued according to clinical subspecialties in medical, nursing, physician assistant, and genetic counseling communities. See Appendix A-2 for a discussion and listing of competencies for physicians, nurses, genetic counselors, and pharmacists.

Academic Preparation, Licensing, and Continuing Education

The dearth of genetics and genomics content in pre-professional, health professional curricula, and CE is widely recognized across all disciplines. In addition, larger, more global issues affecting all disciplines are well recognized and include outdated models of scientific instruction, shortage of adequately trained academic faculty, limited purview of genetics as single-gene disorders only, and inability to commit to genetics CE given many other competing needs and priorities. A review of basic training and education, licensure, post graduate education of health care professionals and accreditation of professional schools is provided in Appendix A-3.

Continuing Education in Genetics and Genomics

Health care professionals generally are optimistic about the future utility of genetic tests and are interested in their eventual incorporation into clinical practice. But many experience feelings of discomfort stemming from lack of confidence in their knowledge of basic genetic concepts, interventions, and management strategies. Thus, many focused resources have been devoted to CE efforts for health care professionals in regional practice settings. Standard instructional methods used to convey genetic and genomic content in these endeavors include web-based instruction tutorials, CE seminars, professional workshops, and conference proceedings.

However, access to and participation in CE 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 complexity of the learned material, method of instruction, and health care providers’ access to interactive practice.\(^{132,133}\) This finding has significant implications for effective clinical translation of genetics and genomics into diffusible health practices.

A recent study examining the impact of a genetic outreach education initiative found that although health care professionals felt more confident in using genetics after the CE intervention, 48 percent of respondents applied their new knowledge incorrectly.\(^{134}\) 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 of complex content is important to assure appropriate and accurate use of genetic information. A genetics education program that increased knowledge and confidence in genetic competencies among physicians delivering genetic services in primary care settings utilized an interactive, case-based, peer education model.\(^{135}\) Peer education emphasizes the usability of the educational materials and concepts and was rated as an effective method by most participants in this study.

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

A recent analysis of the hurdles for the United States in adopting genetics for health care delivery identified three overarching areas: (1) need for scientific evidence; (2) need for economic incentive alignment; and (3) resolution of operational issues such as electronic tracking of diagnostic information and health care provider education and training.\(^{136}\) The need for scientific evidence regarding efficacy and utility applies across all disciplines in realizing the benefits for genetics in health care applications.\(^{137,138}\) The lack of scientific evidence for incorporating genetics into clinical care also influences health professionals’ choice of CE offerings, making it less of a priority than other topics deemed more relevant. Suggested areas for further research include assessment of the scope of clinical benefits and harms involved with various genetic tests, identification of possible ethical and discriminatory harms, and

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Another barrier to integrating genetics in education, training, and practice relates to economic challenges unique to the U.S. health care delivery system. Misalignment of financial incentives between payers, health care providers, patients, pharmaceutical and biotechnology companies, and diagnostics research and development companies, are well documented. The competing priorities among these stakeholders have significant implications as to whether a useful genetic test or genomic technology will be incorporated into health care delivery practices. For example, some analysts have written that establishing high scientific thresholds of evidence for financial reimbursement (payers) can greatly influence whether a health care professional uses what they have been taught concerning genetics and genomics. Synchronization of stakeholder interests between the U.S. health care and clinical research systems has been identified as a key priority in meeting upcoming and current U.S. genetics data needs.

Coverage and reimbursement of genetic tests and services has been identified as limiting their accessibility and integration into the health care system. The current state of coverage and reimbursement of genetic tests and services and recommendations on how to improve mechanisms for coverage and reimbursement are covered in detail in the SACGHS report on the coverage and reimbursement of genetic tests and services.

Health Care Professional Faculty Development in Genetics and Genomics

As noted throughout the previous sections, genetics content of health sciences curricula is variable and tends to focus on single-gene disorders. It is often not presented in a way that leads to long-term knowledge retention for clinical application. Strategies identified to address these deficiencies include:

often focus on enhancing science foundations prior to entrance into health education programs, and integrating genetics content across curriculum requirements.153,154

Given the dearth of medical genetics experts, faculty development is a key concern that was identified across many health care education programs.155 For example, a random convenience survey of N.P. faculty ascertained that although 95 percent of faculty identified genetics as being important only 10 percent reported their academic institution as offering a genetics course as part of the N.P. curriculum, and 20 percent reported instruction of genetics as limited to Mendelian content.156 A study published in 2010 assessed the level of faculty development in pharmacogenomics by surveying U.S. pharmacy schools. It found that most of the 75 schools responding to the survey included pharmacogenomics content in their curricula, however, more than half of these schools had no plans for faculty development in this area.157

More promising are the long-term results from the “Genetics in Primary Care” Faculty Development Initiative, where follow-up data indicated permanent changes in teaching (100 percent at three years) and in clinical practice habits (82 percent at three years).158 This experimental model 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. Although this model was able to promote long-term behavioral changes and comfort with genetics, there remain significant challenges. Only 9 percent of the faculty respondents reported teaching their medical students and residents how and when to refer a patient for genetic counseling; 18 percent reported incorporation of formal genetics teaching for their primary care residents; and 36 percent increased the amount of genetics in medical school curriculums.

Clinical Decision Support and Electronic Health Records

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.159

As electronic health records are increasingly deployed in clinical care, a potential solution to these issues has emerged—just-in-time education. Just-in-time education provides specific answers to specific provider questions at the time the provider asks a question. A study by Trinidad et al. regarding genetic

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education needs of primary care providers identified a desire to have “just-in-time” resources available.\textsuperscript{160} The applicability of active decision support in genetics was discussed in detail in a previous SACGHS report.\textsuperscript{161}

The key to the provision of a specific answer provided in response to a question involves the use of context-sensitive elements embedded in the EHR. This approach involves the EHR “understanding” where the provider is in the patient workflow so that when the query is executed the provider is taken to content that is highly 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 infobuttons\textsuperscript{162} than traditional electronic search approaches.\textsuperscript{163} At Intermountain Healthcare, more than 200 infobuttons relating to genetic disorders in the problem list were linked to specific genetic information contained at GeneTests\textsuperscript{164} and Genetics Home Reference\textsuperscript{165} in addition to traditional sources such as Online Mendelian Inheritance in Man (OMIM).\textsuperscript{166} Following implementation, analysis of the usage of these genetic-specific infobuttons has continued to increase over time with good provider satisfaction.\textsuperscript{167}

The Mayo clinic has also deployed a just-in-time approach to deliver genomic information to providers.\textsuperscript{168} 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.\textsuperscript{169}

Preliminary data from the Intermountain Healthcare System specific to genetic content suggested that providers needed to spend longer amounts of time (~45 seconds) accessing the resource, but no data are available at this time to address whether specific questions were answered. Just-in-time learning has also been deployed to patients in a variety of health care settings. Many of these involve breast cancer care including innovative approaches in patients with low literacy\textsuperscript{170} and to aid in genetic testing decisions.\textsuperscript{171}

Thus, this approach appears to be a promising way to deliver genetic knowledge to the provider.


\textsuperscript{162} Infobuttons are icons that appear in certain areas of the EHR (e.g., problem list, medication list, and laboratory results).


\textsuperscript{167} Personal communication, Marc S. Williams, M.D., Director, Intermountain Healthcare Clinical Genetics Institute, unpublished data, September 3, 2009.


C. SACGHS Surveys of Health Professional Organizations

Methodology

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 (see Appendix B-1 for a listing of these organizations) were contacted via e-mail to respond to the survey, which consisted of 15 open- and close-ended questions developed by SACGHS. The survey explored several major themes including the organizations’ perceived role in, and priority ascribed to genomics education; barriers to enhancing their role in genomics education; and a description of their past, present, and planned efforts around genomics education.

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. See Appendix B-2 for more information on the methodologies and Appendix B-3 for the survey instrument. Thirty-six responses were received (60 percent). See Appendix B-4 for a list of the responding organizations and their reported number of members or constituency.

The survey population was determined by consensus among SACGHS members and staff. Selection criteria for inclusion included the diversity of levels of training within the organization and the organization’s role in training professionals destined to provide primary care services. 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 genomics education were invited to complete the survey.

This survey has several important limitations, including that the sampling of organizations was non-random and relied on qualitative data, which do not allow generalization to health care professional organizations as a whole. Survey data revealed that many of the larger nongenetic organizations have no formal policy or organizational structure that focuses on genetics and genomics education of their constituency so responses to the survey questions may represent the opinion of an individual member rather than consensus of the organization. Additionally, organizations that engage in genetic education and training may be more likely to respond to a survey regarding this topic thus leaving the impression that genetics education and training is more important to health care professional organizations than it actually is.

Survey Findings

Roles and Responsibility for Genetics Education and Training

Twenty-five organizations (70 percent) consider genetics education and training to be a role or responsibility of their organization, although the size and importance of that role varied according to organizational mission and focus (see Appendix B-5, Table 1).

In response to a question asking if the organization was able to fulfill its role or responsibility in genetics education and training, 21 of 25 (84 percent) responding organizations fulfilled their role, one did not, and two stated that they were able to fulfill this role partially. Two of the responses delineate the barriers organizations face in fulfilling their responsibility for genetic education and training:
American College of Physicians: “We are able to develop programs and products related to genetics education. The difficulty is getting members to be interested in them.”

American Academy of Pediatrics (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.”

Several themes emerged from the survey about how organizations can meet their role or responsibility more effectively. These strategies included increasing funding, evaluating current activities, and generating greater interest with institutional leaders and through publications and annual conferences.

**Importance of Genetics and Genomics to the Organization, and Leadership Proficiency in Genetics Education**

In general, the priority placed on genetics education varied depending on the focus of the organization with genetics-specific organizations indicating the strongest interest. Although other professional organizations assigned moderate priority to genetics education, they also reported that such activities are very important. As expected, high levels of proficiency and comfort in genetics education were found only in genetics-specific organizations.

**Entities within Organizations Dedicated to Genetics Education**

Organizations were asked if they have an established committee, workgroup, or dedicated staff that deals specifically with topics in genetics relevant to their organization’s mission. Thirty-six percent of nongenetic-specific professional organizations indicated that they had such entities, with more than twice the activity occurring in genetics-specific organizations. See Appendix B-5, Table 2 for a breakdown among organization categories.

**Barriers to Providing Genetics Educational Activities**

From a list of seven barriers, more than half of all organizations indicated competing priorities and one-third indicated that genetics and genomics was not emphasized in certifying exams and credentialing standards as barriers that impeded their ability to provide genetics education. See Appendix B-5, Table 3 for percentages of organizations that selected each barrier.

**Membership Needs and Priorities for Genetics Education**

More than half of organizations either have 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. NCHPEG noted that although there are efforts within some of its member organizations (i.e., nurses and PAs) to focus attention and educational efforts on genetics through competencies, this emphasis is lacking in some of its other member organizations. As noted earlier in this chapter, NCHPEG recently revised its Core Competencies for All Health Care Professionals, streamlining them based on the experiences of its membership (see Appendix A-2 for a listing of these competencies). It also is poised to release Core Competencies in Family History. Professional organizations have, in the past, used these resources to structure their own specific competencies (e.g., public health professionals and nurses).
How to Engage Members in Genetics Education

Organizations were asked what types of programs or resources could enhance the engagement of their organization’s members in genetics education and if there are programmatic needs that could be addressed by the Federal Government.

The need for funding in the form of educational grants for faculty training and program development, and development of point-of-care tools and tool kits were common themes. Respondents suggested that Federal support of research and dissemination of evidence-based guidelines would help engage their members’ interest in additional genetics topics. Respondents also reported that increased integration of genetics into clinical decision support, electronic medical records, and performance standards would improve member engagement. 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.

Information Relevant to Organizations’ Missions

Organizations were asked to answer open-ended questions in one of four categories most relevant to their mission. Of the thirty-three organizations responding, 18 indicated their most relevant mission as education and training of health professionals; 10 as advocacy and support of practicing professionals; two as certification of health professionals, and three as accreditation or certification of institutions. Selected comments for each category are provided below.

Education and Training of Health Professionals

Integration of Genetics into the Curriculum and Training of Health Professionals

Organizations were asked to characterize the need for integrating genetics into the curriculum and training of health professionals. Most organizations felt that this is a critical need and several have already implemented curricula nationally. Several organizations indicated, however, that this effort was not a high priority and one suggested uncertain clinical benefit of this approach. The following quote from NCHPEG articulates the need and challenges faced by organizations.

“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 diseases.”
Development of Curricular Components

Organizations whose mission is to educate and train health professionals were asked whether they assist member organizations in developing curriculum components related to genetics. Although some organizations do not, others assisted with curriculum components used nationally. Individual responses can be found in Appendix B-4, Table 4.

Cultural Competency Incorporated into Curricula

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. To ascertain whether steps were taken by professional organizations to incorporate cultural competency into curricula, this question was asked of 18 organizations that delineate education and training of health professionals as their primary mission. Twelve of 13 responding organizations stated that cultural competency is part of the curricula or is an accreditation requirement.

Future Needs in Genetics Education

Organizations that identified education and training of health professionals as most relevant to their mission were asked to look ahead 5 to 10 years and indicate anticipated needs in genetics education. Responses were varied and ranged 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 genetic topics. The need to understand risks for complex diseases was mentioned as was the need to be able to assess risks using multiple tools, change management based on risk, and communicate risk effectively. One organization felt that there is no end in sight to the need for education in genetics, while another stated that genetics will be a part of mainstream education and clinical practice in 5 to 10 years.

Advocacy and Support of Practicing Health Professionals

Continuing Education Programs and Activities Related to Genetics

Among the 10 organizations who identified their primary role as advocacy and support of practicing health professionals, the American Academy of Family Physicians responded that it surveyed members in 2003 regarding CE programs and found that the top two CE genetics topics requested by members were common genetic diseases and genetic testing and counseling. This organization has a subcommittee on genomics that has also identified that members would benefit from more information regarding pharmacogenomics.

Promoting Greater Knowledge of Genetics

When asked what would help to promote a greater knowledge of genetics, the following responses were provided from six organizations that identified their mission as advocacy and support of practicing professionals:

- Additional CE opportunities, learning communities, and resource portals.

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• Medical education, continuing education, certification of professionals.
• More funding to allow for more genetics educational opportunities for members. While there are many grants available for genetic research, there are fewer opportunities to obtain funding for education and training.
• Until utility of genetic testing is shown, genetics and genomics will probably not have great uptake. For physicians, the primary desire will be positive clinical outcome studies. Until these studies have been undertaken, education of health care professionals about current technologies and surrounding issues is needed.
• In the next 10 years, potentially every person seeking health care services will have had at least one genomic test and possibly full-genome sequencing. The results of these tests will become an important component of every aspect of medical decisionmaking, from assessing the significance of a cholesterol result, to prescribing a medication, to determining whether a cancer patient needs more aggressive treatment than indicated based on histology alone. We need to identify ways to train clinical educators/internship supervisors to recognize genetics issues in clinics so that trainees are able to observe and then take part in identifying and managing these issues in their clinical rotations.

Certification of Health Professionals

Credentialing Exams

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 were on genetic content. One nursing certification organization reported genetic content but at less than 1 percent of total content and one general professional organization reported genetic content at less than 5 percent of total content.

Accreditation or Certification of Institutions

Two organizations, ABGC and Accreditation Council for Graduate Medical Education, considered accreditation or certification of institutions as their primary role. Both organizations view integration of genetics into the curriculum and training of health professionals as important and regularly update these curriculum requirements.

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 genetic specific, professional education, or general professional organizations. The results of the survey were reported to SACGHS on June 14, 2004. Thirteen responses were received (50 percent). See Appendix A-5 for a list of the organizations who responded.

The 2004 survey was considerably shorter than the 2008 survey; however, there are several areas where comparisons can 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 patient questions and thus require a solid, basic knowledge of genetics with a lifelong commitment to learning. By 2008, 15 of the 17 organizations responding to this question felt similarly and several have actually implemented genetic curriculum nationally. However, in 2008, several organizations felt that this effort was not a high priority, and one suggested uncertain clinical benefit of integrating genetics into the curriculum and training of health professionals.

Barriers to Providing Genetics Educational Activities

In 2008, 64 percent of organizations cited competing priorities as a barrier to providing genetics educational activities, followed by 28 percent citing genetics and genomics not emphasized on certifying exams or credentialing standards. Only 8 percent cited lack of evidence supporting clinical effectiveness of care based on genetic or genomic information. Again, the numbers of responding organizations in 2004 were small; however, similar barriers were noted: 57 percent reported competing priorities; 43 percent reported lack of evidence for clinical application of genetics; and 43 percent identified lack of prepared faculty. While competing priorities remains a significant barrier to providing genetic and genomic educational activities, organizations were much less likely to report lack of evidence supporting clinical effectiveness in 2008.

Themes Common to Both Surveys Regarding Future Directions in Genetic Education of Health Care Professionals

- The government has a role in supporting genetics education programs.
- Genetics education must be represented throughout 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.

D. Summary

A significant body of literature from the United States and abroad highlights the nature and lack of genetics education of health care professionals as factors limiting integration of genetics into health care. Genetics content is often minimal in health education programs, focused primarily on single-gene disorders, and not associated with long-term knowledge retention for clinical application.

A recent study examining the impact of a genetic outreach education initiative found that although health care providers felt more confident in using genetics after a CE intervention, many applied their new knowledge incorrectly. Incorrect application of genetics occurred in the misappropriation of risk estimation; approximately half of those receiving the genetics education intervention issued a high-risk categorization to a low-risk breast cancer presentation.

Lack of genetic knowledge among health care providers exists not only with complex, multifactorial conditions—but also with traditional and well-documented Mendelian conditions such as autosomal...
dominant hereditary cancer syndromes. Analyses of genetic content in formal medical and health care
curricula (with the exception of genetic counseling programs) find great 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. To address some these deficiencies, several professional groups,
particularly those representing PAs and nurses, have included genetics knowledge in their competencies
and guidelines and offer CE in genetic content.

The 2008 SACGHS survey data found that overall, 70 percent of health professional organizations
responding to the survey view genetics education and training as part of their role or responsibility. Most
of these organizations reported that they were able to fulfill this role or responsibility; however, funding,
program evaluation, and increasing interest within the organization’s leadership would allow them to
meet this 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.

Only half of survey respondents reported that they had received input from their membership regarding
educational needs and priorities in genetics, but those that did were able to provide numerous examples of
how they obtained this feedback. Open-ended survey questions yielded information on ways to engage
members, integrate genetics into curriculum and training, develop curricular components, incorporate
cultural competency into curricula, advocate for practicing health professionals, and develop CE
programs and activities. Despite this relative interest in genetics education, only nine organizations
reported having published position statements or practice competencies regarding genetics.

Although health professional organizations across the board reported that developing and promoting
educational activities related to genetics is important, these topics are not a high priority relative to the
overall priorities facing the organizations. Nongenetic-specific organizations reported only moderate
proficiency in and comfort of their leadership in genetics education, and less than half of these
organizations have dedicated entities specifically focused on genetic topics relevant to their mission. The
majority of all organizations surveyed identified competing priorities as a barrier to their ability to
provide genetics education.

The 2008 SACGHS survey of health professional organizations provided data that supports findings from
the literature review yet revealed new insights into how professional organizations are currently
approaching the need to educate their members and constituencies in genetics and genomics. Competing
priorities in an already crowded curriculum was the most commonly mentioned barrier to improving
genetic literacy of health care professionals. In addition, respondents cited lack of sufficient resources,
financial and otherwise, as a barrier to developing or accessing appropriate education and training
opportunities for members. In response to the growing need for proficiency and competency,
organizations offer tailored CE programs or include innovative programs that reflect emerging genetic
content in their annual meeting agendas. Thus, the need for educational efforts to increase the use of
genetic and genomic information in clinical care is widely recognized and acknowledged. The SACGHS
survey, however, highlighted the challenges facing many organizations attempting to fill the gap for their
constituencies against a backdrop of competing demands and limited resources.

Based on a literature review and its survey findings, SACGHS found evidence that suggests inadequate
education of health care professionals is a significant factor that limits the integration of genetics into
clinical care. Enhancing the use of clinical decision support tools, promoting the importance of family
history, and ensuring adequate reimbursement for genetic services are among the approaches that would
support the optimal use of genetics and genomics in health care.
III. The Status of Education and Training of Public Health Providers

A. Introduction

Genetics has been at the center of a number of 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 include genetics coordinators and frequently consult genetics professionals. However, expertise and focus has, in general, 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. Public health genomics is defined by Khoury et al. as seeking “to use 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.”

This chapter briefly describes the role of the public health workforce in population health, the range of pathways to a public health career, accreditation of schools of public health, training opportunities, and reviews what is available in the literature regarding the readiness of the public health workforce to engage in genetics and genomics. In addition, SACGHS developed 12 competencies, derived from competencies developed by public health organizations and institutions, to use in an online survey instrument assessing public health providers’ genetic and genomic training and education needs. The results of that survey are reported here.

The 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, develop policies, and assure that appropriate services are available to individuals, families, and communities. A landmark 1988 report by the Institute of Medicine (IOM) defined public health as the collection of society’s efforts to achieve conditions in which people can be healthy. The IOM report further defined a public health professional as any professional who approaches health from a population lens. Public health providers can work across various sectors, including Federal and state government, academia, professional, community and lay organizations, and the private sector. 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 the unique skills and networks potentially to raise the level of general genomic literacy and develop targeted messages about the use of genetic information for disease prevention and health promotion.
addition, the public health community has a large research infrastructure sorely needed by genomics, for example its surveillance and data collection systems.

The diversity of settings and service provision and the lack of specific licensure that would otherwise facilitate counting and studying the public health workforce create an inherent problem in targeting genetic and genomic educational efforts. In 2000, the public health workforce was estimated to consist of 448,254 persons in salaried positions, supplemented by 2.9 million volunteers. In this estimate, 44 percent of workers were identified as professionals (e.g., physicians, nurses, laboratorians, dentists, health educators) but 24 percent of the workers could not be categorized. It was estimated that over half of all public health workers have at least a college education. Since 2000, numerous reports have attempted to count and categorize various levels of the public health workforce, which is considered to be a critical step to assuring that the workforce is sufficiently large and skilled to deliver essential public health services to the U.S. population. However, availability of this data and sufficient resources to support research of the public health workforce are lacking.

Schools of Public Health

The Council on Education for Public Health (CEPH) is an independent agency recognized by the U.S. Department of Education to accredit schools of public health and community health and preventative medicine programs. These schools and programs prepare students for entry into public health careers and offer Master of Public Health (M.P.H.), Doctor of Public Health (Dr.P.H.), and Master of Health Care Administration (M.H.A.) degrees. The Association of Schools of Public Health (ASPH) represents the 43 CEPH-accredited schools of public health and the eight Associate Member Schools that are working on accreditation. However, other programs exist which are not members of ASPH and, moreover are not necessarily CEPH-accredited. ASPH also supports graduate internships and fellowships to provide practice experiences. The National Public Health Training Centers Network, funded through the Health Resources Services Administration, has partnered with schools of public health, related academic institutions, and public health agencies and organizations to assess learning needs and provide training to meet those needs. Their distance education center lists several genetics and genomics courses offered by partner organizations.

B. Literature Review

Barriers to Achieving a Genomics Informed Public Health Workforce

The current public health workforce faces challenges in receiving and 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. has expressed concern 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.190

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 is not feasible. Second, many providers in the field today received their formal education before genomics became a critical aspect of medicine and health. Third, attitudes, perceptions, and beliefs shape acceptance and adoption of genomics by the public health community.

Khoury et al.191 have noted some of the attitudinal barriers to acceptance of genetics and genomics by the public health community to include skepticism about genomics and genomics research being seen as a low-yield investment and low priority because of other more important preventative or modifiable environmental causes of morbidity and mortality. For many public health providers, local issues, national and international pandemics, and environmental causes of morbidity and mortality are more important priorities than genetics and genomics, particularly in the context of limited public health funding. Research also highlights that public health providers do not perceive public health genomics to be part of their job, nor a professional priority.

Public health educators perceived barriers include not only lack of knowledge regarding the link between genomics and health promotion, but also lack of current basic genomic knowledge. Thus, future education and training of public health providers focusing primarily on basic genomic content will be inadequate.192 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 field to promote and incorporate genomics, and attempted to gauge their basic and applied genomic knowledge. While most public health providers agreed with the Centers for Disease Control and Prevention (CDC)-proposed competencies, incorporating them into public health practice was viewed as important by less than half of the study participants.193 Subsequent work by Chen et al. 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 genomic education programs, suggesting that health educators are not ready for their professional role in genomics,194 and only half of basic and applied genomic knowledge questions were answered correctly. 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 curriculum nor do they require course offerings in these topics for accreditation purposes.”

Finally, lack of evidence might be a significant barrier to public health adoption of genomic competencies. Until evidence of health benefit can be shown (e.g., population screening for \textit{BRCA1} and \textit{BRCA2} mutations and hereditary hemochromatosis), public health providers might be resistant to adoption. Thus, public health genomics 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.”

Thus, one approach to educating the public health workforce is to have clear examples of beneficial applications of genomics at the population level, which can be built on as new evidence arises.

Further confounding educational efforts for public health providers is the proportion of non-professional, community health workers in the public health workforce. The use of lay health providers creates an additional barrier to achieving widespread genetic and genomic literacy in public health programs.

Community health workers (CHWs) are, by definition, “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.” Although Texas, Ohio, Indiana, and Alaska require some level of certification for CHWs and several states are considering implementing certification requirements, most states do not, and there were until 2007, no national standards for certifying or training non-professional public health workers.

The Center for Sustainable Health Outreach (CSHO) issued a report in 2002 that listed CHW programs that offer credit, certificates, or degrees at institutions of higher education. At that time, there were 15 programs in 10 states that offered courses, certificates, and/or degrees for generalist CHWs. The majority of the programs were offered at community or junior colleges and led to certificates, rather than Baccalaureate degrees. Even where programs exist for formal training of CHWs, the emphasis is on communication skills (including bilinguality), service coordination skills, advocacy skills and “a knowledge base on specific health issues.”

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202 Center for Sustainable Health Outreach. (2002). \textit{A Report of College and University Programs Awarding Credit, Certificates, and/or Degrees in the Community Health Worker Field}. See \url{http://www.usm.edu/csho/report.htm}. Accessed on December 12, 2009.

One of the most rigorous trainings for CHWs is in Ohio. There, certification training programs are operated under the authority of the Nursing Practices Act and require at least 100 hours of didactic instruction by certified medical professionals and 130 hours of clinical instruction. The three educational programs currently certified in Ohio require coursework in basic anatomy and the physiology of major body systems, medical terminology, health education related to child-bearing, and competencies in the areas of immunization and appropriate referrals to health care facilities and practitioners. Although not ostensibly related to genetics or genomics, the approved curricula could be expanded to require competency and knowledge about genetics and genomics as part of the biology coursework or health education competencies related to childbearing and immunization.

Current Efforts to Improve Proficiencies and Competencies

Several professional groups and CDC have turned their attention to the need for public health provider education and training in genomics. The IOM report recommended genomics as one of eight new content areas to be covered by every school of public health. The American Public Health Association (APHA), representing more than 50,000 health professionals, has published policy statements related to genetics and genomics and the public health workforce. Genetics and Public Health, published in 1987, 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. The need for professional education on advances in genetics was outlined in the objectives and implementation methods that were proposed to achieve these objectives. Recognizing the need for an information infrastructure 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 genomics. In recognition of the broader scope of genomics and its impact on public health and the critical need for public health workforce education in genomics, APHA published The Role of Genomics in Public Health in 2002. In 2007, The Genomics Forum was established within APHA 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”. The Genomics Forum has developed a policy statement on genetic health literacy for health professionals to be submitted to APHA for review and publication in 2010.

In August 2000, the CDC Office of Genetics and Disease Prevention and representatives from each of the disciplines in public health met to identify the core competencies necessary for all health professionals to incorporate genetics into public health practice. The group developed specific

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210 Personal communication, Heather Honore, Policy Committee Chair, Genomics Forum, American Public Health Association., January 13, 2010.

genomic competencies for various public health providers. 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. Encouraging, rather than requiring, that such competencies be demonstrated can further slow their adoption. It is possible that various social, organizational, and environmental factors (e.g., certification and licensure requirements) would 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 V). It has funded Centers for Genomics and Public Health in 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. CDC also has supported genomics programs in four state health departments (Michigan, Minnesota, Oregon, and Utah).

Other states have instituted their own initiatives in public health genomics. For example, in 2009, Washington sponsored the Summer Institute in Public Health Genomics: Translating Genomics into Policy and Practice. The Oregon Genetics Program aims to integrate genomics into public health practice, particularly chronic disease program activities. The Oregon Public Health Division received funding to translate genomics applications 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 genomic tests and family history. This project will also evaluate disparities associated with accessing cancer-related genetic testing and counseling. Illinois public health officials conducted a needs assessment and created a state genetics plan in 2006. The Connecticut Department of Public Health Genomics Office has produced a fact sheet for consumers on DTC personal genomic services, highlighting the types of activities that can be accomplished in the public health arena.

C. SACGHS Survey of Public Health Providers

Methodology

To assess the genetics education needs of public health providers, SACGHS developed 12 competencies that were derived from competencies found in public health organizations and institutions. The 12 competencies were used in an online survey instrument assessing public health providers’ genetic and genomic training and education needs. The online survey was based on the work of Kirk et al. and modified by SACGHS members and staff. 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. The survey was distributed to around 500 public health providers with varying degrees of genetics responsibilities and 140 responses were received and analyzed. See Appendix C-1 for details of the survey methodology and participant recruitment. Appendix C-2 provides screen shots of the online survey instrument and Appendix C-3 explains survey reliability results.


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 sampling\(^\text{215}\) 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.

Twelve Competencies Used in the SACGHS Survey

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

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.
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; to 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.

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

**Survey Findings**

**Perception of the Importance of the Competencies**

Overall, little variability was found among the responses regarding the importance of each competency. On a scale of 1 to 4 (with 1 not important and 4 very important) mean values range from 3.6 to 3.8 (see Appendix C-4, Table 1 for summary data). The majority of individuals responded that all of the competencies are important.

The three competencies ranked most important to public health providers were: (1) demonstrating basic knowledge of the role of genetics and genomics in development of disease; (2) describing the importance of family history in assessing predisposition to disease; and (3) identifying opportunities and effectively integrating genetic and genomic issues into public health practice, policies or programs.

No single item was ranked low, thus there is no reason to conclude that any specific competency is not important to public health providers.

**Level of Confidence in Demonstrating the Competencies**

Responses varied with regard to level of confidence in demonstrating the competencies. However, two of the competencies that ranked most important were also those in which respondents could demonstrate the most confidence, i.e., the importance of family history and basic knowledge of the role of genetics and genomics. In addition, respondents felt competent to describe the potential physical and psychological benefits, limitations, and risks of genetic and genomic information for individuals, family members, and communities.

The lowest mean ranked competencies were: (1) maintaining up-to-date knowledge of genetics and genomics-related policies, legislation, statutes, and regulations; (2) using information technology (IT) to obtain credible, current information about genetics; and (3) conducting outcomes evaluation of available genetic and genomic services to determine their effectiveness.

**Frequency of Application of Competencies**

Responses were varied for how frequently the competencies are applied with mean values ranging from 2.0 to 3.3. It appears that depending on the competency, 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 and genomics in the development of disease and maintaining up-to-date knowledge on the development of genomic science and technologies within his or her professional field were reported to be most frequently applied by public health providers.
The majority of the public health providers apply these two competencies monthly or weekly. The lowest mean ranked competency is conducting outcome evaluation of available genetic and genomic programs and services to determine their effectiveness. The majority of public health practitioners either never apply this competency or they apply it rarely (1-2 times per year). Overall, when considering the importance of these competencies to public health providers, there appears to be no competency that stands out as unimportant or irrelevant to these survey respondents.

**The Importance of Genetics and Genomics to Institution Leadership**

Respondents were asked how important knowledge and experience in genetics and genomics is to their roles and responsibilities from their senior administration’s perspective. Sixty percent of survey respondents reported that their senior administration feels that genetics and genomics is important or very important to their job responsibility, while 21 percent responded that they feel their administration thinks it is of little or no importance. See Appendix C-4, Table 2 for summary data.

In addition, one-third of respondents reported that their senior administrators think that genetics and genomics are important to very important to their own job responsibilities; one-third felt it was somewhat important; and one-third felt that genetics and genomics are of little or no importance. See Appendix C-4, Table 2 for summary data.

One-third of respondents reported that they feel they have adequate to very adequate resources for implementing genetic and genomic competencies in their work or role, while two-thirds reported that the resources they have are not or only somewhat adequate. See Appendix C-4, Table 3 for summary data.

**Respondents’ Role in Public Health**

Respondents were asked to indicate the level of public health setting in which they work and the amount of time spent on genetic or genomic tasks. 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 (e.g., commercial laboratory, medical center community programs, nonprofit health organizations) (2 percent), and international positions (1 percent). No respondents reported working at the local level. The majority of respondents spend less than half of their work time on genetic-specific tasks. (Appendix C-4, Table 4 provides a summary of the frequencies and percentages of responses to each job level.)

**Delivery of Genetic Services to Underserved or Vulnerable Populations**

Using an open-ended question format, respondents were asked to describe efforts that their organization has undertaken to ensure that genetic services or information are available for vulnerable or underserved populations and to recommend specific strategies. A total of 71 responses were received in response to this question. Reported efforts and strategies 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 C-5, 1 for more detailed responses.

The survey closed with an opportunity for individuals to provide additional comments to SACHGS on the topic of genetics and genetics education for public health providers. Fifty-four responses were received, describing themes around funding, networking and collaboration, best evidence-based practices, and education. Details can be found in Appendix C-5, 2.
D. Summary

The literature review provides evidence that the current public health workforce is not well prepared to receive and assimilate genetic and genomic information. It also demonstrates 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. Thus, a one-size-fits-all approach is not feasible. Second, many professionals in the field today received their formal education before genomics became a critical aspect of medicine and health. Third, attitudes, perceptions, and beliefs shape the acceptance and adoption of genetics and genomics by the public health community.

Some studies have found that public health educators’ perceived barriers included not only lack of basic genomic knowledge but also lack of knowledge regarding the link between genomics and health promotion. The literature also reveals that public health providers do not perceive public health genomics to be part of their job, nor a professional priority. Until evidence of public health benefits of genetic testing can be demonstrated (e.g., population screening for \textit{BRCA1} and \textit{BRCA2} and hereditary hemochromatosis), public health providers might be resistant to adoption.

There have been efforts to develop competencies in genetics and genomics for public health providers. In August 2000, the CDC Office of Genetics and Disease Prevention and representatives from each of the disciplines in public health met to identify the core competencies necessary for all health professionals to incorporate genetics into public health practice. The group developed specific genomic competencies for various public health providers. ASPH also has highlighted the importance of genomics in the Master’s Degree in Public Health Core Competency Development Project.

The SACGHS survey found that respondents believe the 12 competencies developed by SACGHS were important. Demonstrating a basic knowledge of the role that genetics and genomics plays in the development of disease was considered the most important and most frequently applied competency, while confidence in describing the importance of family history ranked highest. Conducting outcomes evaluation of available genetic and genomic services ranked the lowest in importance, frequency of application, and confidence in demonstrating this competency.

Sixty percent of survey respondents reported that their senior administration feels that genetics and genomics are important to the respondents’ job responsibilities. However, the topic was not central to the overall administration of the workplace, and only one-third of respondents felt that resources for implementing genetic and genomic competencies were adequate or very adequate.

More than half of respondents provided information on delivery of genetic services to underserved or vulnerable populations. These responses included 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.

Survey respondents also identified strategies and recommendations to target vulnerable or underserved populations. These included the need for increased funding to enhance genetic services, outreach, and partnerships with vulnerable or underserved populations; 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.

The literature points to a number of factors that impede incorporation of genetics and genomics into public health practice and demonstrates that well-defined lines separate the public health workforce.
engaged in genetics and genomics, such as in newborn screening programs, and those who do not see

genetics and genomics as related to their work. While many of the same concerns and barriers were
highlighted in the SACGHS survey of public health providers, overall, survey respondents had a positive
attitude toward genetics and genomics. Further, all of the genetic and genomic competencies developed
for and used in the survey instrument were considered important by respondents. As discussed in the
survey limitations above, respondents may over represent individuals and organizations that have
responsibilities in genetics and genomics and thus be biased favorably to the importance of these
competencies.

Based on a literature review and its survey findings, SACGHS recognizes that the public health workforce
is divergent and heterogeneous, which complicates genetic and genomic education and training efforts.
Educational approaches based on genetic and genomic competencies targeted to the training needs of the
multiple professional roles within public health will be required for the workforce to effectively address
public health needs, while recognizing issues of cultural competence, social and economic determinants
of health, and reduction in health disparities. Identifying effective educational models for public health
providers serving in underserved communities and identifying the role of family history in population
health will contribute to improved public health.
IV. The Status of Consumer and Patient Education

A. Introduction

Since the inception of the Human Genome Project (HGP), there has been the recognition that the vast amount of information revealed about the human genome would result in ethical, legal, and social issues (ELSI) that affect individuals, families, and communities.\(^{216}\) The ELSI Program was established in 1990 to investigate ELSI issues raised by and as a consequence of the HGP\(^ {217}\) and provide guidance to policy makers and the public on the implications of human genome research.\(^ {218}\) However, it is unlikely that 20 years ago we would have expected consumers to be able to order a genetic test directly from the Internet without the participation of a physician.

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.\(^ {219}\) 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 professionals is not known. The emergence of social networking and sharing genetic information via the Internet compounds concerns of confidentiality and the consequences of sharing genetic information in this manner are difficult to predict.\(^ {220}\) However, new technologies also provide innovative solutions to societal problems; 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.\(^ {221}\)

As genetic testing becomes more widely available, the need for education about genetics and the results and implications of testing will grow steadily. However, the number of genetic professionals will not be able to meet the translational and interpretive need,\(^ {222}\) and consumers and patients are likely to seek information on their own. This situation will require genetic literacy. However, genetic literacy is based on having a sufficient educational background (i.e., exposure to, at a minimum, high school scientific coursework) and English language proficiency. In the United States, 21 million individuals speak English “less than very well” and are thus said to be limited English-proficient.\(^ {223,224}\) This chapter summarizes the available literature on consumer and patient knowledge and understanding of genetics and explores how


To understand what is known about the genetics literacy of the general public, a search was conducted to identify pertinent literature covering the years 2003 to 2009 as described in Chapter I (page 8). Additional salient documents were collected and reviewed, in particular a literature review conducted in 2009 for the National Institutes of Health (NIH) by the Academy for Educational Development (AED).\footnote{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.} To ensure that the opinions of the general public were reviewed for this report, Cogent Research provided SACGHS with the findings from its 2008 survey, Cogent Genomics Attitudes and Trends.\footnote{Cogent Research, LLC. (2008). Cogent Genomics Attitudes and Trends: 2008. Provided to SACGHS with permission to cite, April 29, 2009.} 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.

In addition, to elucidate the genetic 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 experts in consumer and patient health education and a web-based survey of the health advocacy community. The results of that data gathering effort are presented here.

**B. Literature Review**

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

The AED conducted a search of published and unpublished literature on the marketing of genetic testing. Their review emphasized direct-to-consumer (DTC) genome-wide scans of susceptibility markers for common diseases. The review also yielded 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 for 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 for the interested public and health care professionals?

The search yielded 128 relevant articles published between 1998 and 2009. These were reviewed by AED to assess what is being communicated to consumers about genetics, genetic risks, genetic services, and personalized medicine.

AED concluded from the literature that, while most consumers have a positive attitude toward genetic testing, their understanding of genetic testing is very basic, often misinformed, and does not appear to be increasing over time. Specifically, the AED literature review identified that:

- Consumers do not understand that there are many types of genetic and genomic tests, and there are many contexts in which they are used.
 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 the following 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 consider that limited health literacy constitutes a formidable barrier to the public’s understanding of genetic tests.
- Standardized physician training, to include both didactic instruction and supervised experience in the delivery of genetic health care, would allow physicians to better educate the public about genetic tests.

Knowledge of Consumers and Patients Regarding Genetics and Genomics

Studies that have assessed the public’s knowledge of genomics and genetic testing generally have found that the public has only a rudimentary knowledge of basic genetic terms yet overall positive attitudes towards genetics. In general, people seem to be reasonably aware that genetic risk factors contribute to health outcomes. However, understanding of genetic risk factors is dependent on education and health literacy, which also varies by race, ethnicity and English language proficiency in the United States. According to Cogent Research, overall awareness of genetics by the public

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increased between 2006 and 2008. In 2008, 79 percent of respondents reported that they had heard or read about using individual genetic information to understand and optimize health and about half of respondents felt informed about their family history.239

The literature and surveys, however, are not informative about whether members of the public know how to use genetic information to understand their risk of disease or to optimize health. And, because those who participate in studies may not reflect the balance of race and ethnicity in the United States, the findings may not apply to the general U.S. population.

Smerecnik et al.240 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). Cogent Research found that 50 percent of respondents were aware that genes predict the likelihood of developing specific diseases. However, among Cogent survey respondents, less than 5 percent understood that genetic information can be used to optimize health.241 Awareness of risk factors also varies depending on the disease; for example, in the studies reviewed by Smerecnik,242 60 percent of the general public was aware of genetic risk in breast cancer, while only 20 percent was aware of genetic factors in cervical cancer. Knowledge beyond awareness, however, such as how to process such information and use 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.243,244 Several studies have linked level of education with knowledge of genetic concepts or genetic testing, demonstrating that, as might be expected, the higher the education level achieved, the greater the genetic knowledge.245,246,247,248 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 felt that their genetic knowledge was

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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 shown in the literature. Several studies have evaluated genetics literacy among individuals with or at risk for genetic diseases. First-degree relatives of early onset familial Alzheimer disease were found to have limited knowledge of their own personal risk of developing the disease. Similar findings were reported by Moscarillo et al. General genetics knowledge among persons with familial testicular cancer and their family members was found to be generally low, with less than half (41 percent) of respondents able to answer questions correctly regarding testicular cancer and genetics. A study of adults with cystic fibrosis found that they have limited knowledge of the genetics of their disorder. 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.

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 found that even a decade ago people seeking information about the genetics of Alzheimer disease actively searched the Internet for information. 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 knowledge—activities that caused them to value their own experiential knowledge. Eighty-three percent of families referred to a pediatric genetics clinic obtained information from the Internet regarding their child’s diagnosis. In this study, convenience, privacy, and finding information they did not otherwise 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. A general practitioner was the preferred source of

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genetic information in a Dutch study of patients with chronic disease, followed by information brochures, medical specialists, and special Internet sites. In a focus group study of culturally diverse populations recruited from 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.

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.

The Public’s Confidence in Its Genetic Knowledge

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. 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 flawed than individuals who are unaware of their limitations. 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.

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. Overall, the general public has been supportive of genetic testing to improve disease diagnosis and prevention. Etchegary et al. found that 95 percent of survey respondents thought genetic information should be used to improve disease

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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’ decision 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.270 The majority of respondents in this study had not thought about potential negative consequences genetic information might have for insurance coverage or employment discrimination.

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.”271 Cogent Research’s 2008 survey was conducted shortly after the passage of GINA on May 16, 2008. 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 felt 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.272 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.273

Health Disparities and Cultural Issues Related to Genetics

Numerous reports have documented the extent of health disparities in the United States, and the field of genetics is no exception.274 When any new technology emerges it has the potential to exacerbate disparities if patients and providers do not have access to appropriate and relevant information. Genetic-related disparities include lack of awareness of and access to genetic counseling and genetic testing. When the standard of care is to offer a genetic test, there is evidence that minorities do not participate in genetic testing or are offered genetic counseling services as often as whites. African American women were found to be much less likely to undergo genetic counseling than white women for BRCA1 and BRCA2 genetic testing.275 How much of these disparities can be attributed to issues of access versus knowledge of and attitudes about genetic testing is not clear. However, several studies have attributed lower genetic knowledge to socioeconomic factors. Those with lower levels of education have been found to be less knowledgeable about basic genetic information, the role of genetics and chronic disease, and genetic testing.276, 277, 278

Several studies have compared knowledge of genetic testing in general and genetic testing specifically for \textit{BRCA} mutations and cancer risk among African Americans and whites. Forty-nine percent of African Americans and 72 percent of whites had heard of genetic testing in general.\textsuperscript{279} Knowledge has been shown to be lower for \textit{BRCA} genetic testing with 19 to 25 percent of African Americans and 35 to 68 percent of whites reporting knowledge of this test.\textsuperscript{280,281} Wideroff et al. found that 49.9 percent of whites had heard of genetic testing for cancer risk compared to 32.9 percent of African Americans and only 20.6 percent of Latinos.\textsuperscript{282}

Zimmerman et al. found that 90 percent of a survey sample of equal numbers of inner city African Americans and Caucasians thought “genetic testing to check for risk of getting a disease was a good idea” regardless of race.\textsuperscript{283} Other studies have found that attitudes about genetics among African Americans and Latinos differ from whites. African Americans and Latinos overall hold a positive view of genetics, but it is not as positive as whites.\textsuperscript{284,285,286,287} Nonetheless, Zimmerman et al. found that 58 percent of African Americans and 34 percent of whites thought genetic testing would lead to racial discrimination.\textsuperscript{288} In a survey of 170 African Americans and 181 Caucasians in Philadelphia, Peters et al. found that the belief that genetic testing would lead to racial discrimination was low but more prominent among African Americans than whites,\textsuperscript{289} and that African Americans were less likely to endorse the health benefits of genetic testing. 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.\textsuperscript{290,291} In a study among Latinos in New York City, 74 percent of Latinos and 72 percent of whites had heard of gen etic testing in general.\textsuperscript{282}

Knowledge has been shown to be lower for \textit{BRCA} genetic testing with 19 to 25 percent of African Americans and 35 to 68 percent of whites reporting knowledge of this test.\textsuperscript{280,281} Wideroff et al. found that 49.9 percent of whites had heard of genetic testing for cancer risk compared to 32.9 percent of African Americans and only 20.6 percent of Latinos.\textsuperscript{282}


City, Sussner et al. found that individuals with higher levels of acculturation—or the degree to which they have adopted the attitudes, values, and behaviors of the majority culture—were more likely to be familiar with genetic testing and to perceive its benefits.  

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. 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. This finding suggests that women of lower SES may be requesting unnecessary genetic tests based on an incomplete understanding of genetic risks. 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 all been found to have an influence on 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.

Another factor to consider in health disparities relates to literacy and English language proficiency. The U.S. Census Bureau reported that 13 percent of Americans had not completed high school in 2008 and from 2000 census data, 21 million Americans speak English “less than well”. This low English language proficiency is more common in minority populations and limits access to medical care, specifically by a decrease in health care visits. 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 genomics plays in maintaining health and in defining disease risks. One strategy that may begin to address literacy as a barrier in health care is to identify those with lower literacy, and a tool has been developed to identify patients with low literacy in a clinical genetics setting.

When genetic and genomic 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. Addressing health disparities through education about genetics and genomics may therefore 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

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as an inducement to physician use of genetic services,\textsuperscript{300} thus, the use of strategies customized to specific
groups and communities may be an effective way to promote the use of emerging genetic and genomic
technologies, when medically appropriate, and empower a wide variety of consumers to act as their own
health care advocates.\textsuperscript{301} Programs such as the Community Genetics Education Network (CGEN)
Project\textsuperscript{302} 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 play
out in research settings. The promise of genomics may not benefit those who do not participate in genetic
and genomic research. Studies have found that attitudes about genetics among African Americans and
Latinos differ from whites, and that minorities in the United States are less likely to participate in
research, including genetic and genomic research. Without the participation of all segments of the
population, it will be difficult to tailor treatments and preventive measures for specific sub-populations 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. However, 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.\textsuperscript{303,304} This assumption arises when researchers overemphasize the genetic contributions to
disease and health without consideration of social contributions to health.\textsuperscript{305} Recommended educational
efforts should therefore focus on ameliorating attitudes about the purpose of genomic research and
increasing the understanding of the complex interrelation of genes and the environment, including social
contributions to health. When the promise of personalized medicine is understood, disparities in research
participation and in the provision of appropriately tailored health care can be potentially reduced. 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.\textsuperscript{306}

Although studies have documented disparities in access to genetic services, other studies suggest that
genomics and personalized medicine may help address disparities. Increasingly widespread use of the
Internet, for example, may open access to personal genomic information and reach larger numbers of
people than are currently seen by genetic counselors or clinical geneticists.\textsuperscript{307} Furthermore, should whole-
genome sequencing become affordable and accessible in the future, fair representation of all major groups


will be required to avoid large gaps in understanding the human genome. A shift to whole-genome sequencing may also help resolve the emphasis on research that uses ancestry or ethnicity as an easy shortcut for identifying genomic associations.

With whole-genome sequencing, and incorporation of related ancestry and family health history, the “too narrow focus on genetic variation” can be replaced by “personalized information that can and should guide clinical decisionmaking for individuals.” This personalized information would include “observables such as the environment and physiology” that would help clarify genomically similar individuals with different environmental exposures, cultural practices, and access to medical services. These factors, and the individual’s self-identification with a specific family health history (i.e., high blood pressure or heart disease among family members), may 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 subsequently treating [ethnic/racial sub-populations] as one uniform group.” Ramos and Rotimi, for example, explore how studies on the efficacy of beta-blockers among African Americans highlight the need for personalized, rather than racial or ethnic categories for appropriate and effective treatment decisions.

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. 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 and in essence, “invert” the genetics curriculum to teach about complex traits before rare, Mendelian genetic concepts. The challenges of improving genetics education at the K-12 level are significant. However, other than to acknowledge how important K-12 education is 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. One of

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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, 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.317 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 2008.318 This legislation earmarked funding and directed the Heath and Resources
Service Administration (HRSA) to develop a clearinghouse for newborn screening educational and family
support and services information, materials, resources, research, and data that would be interactive,
regularly updated, and link to government and nonprofit websites. The Genetic Alliance was awarded the
contract to develop this clearinghouse and began work on the project in September 2009.319

In recent years there has been a concerted effort to develop tools and public involvement in efforts to
enhance family history taking. In particular, the Office of the Surgeon General, NIH, the Centers for
Disease Control and Prevention, and HRSA initiated a public health campaign 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. This effort is further addressed in Chapter V, but it is an
important tool for consideration in public education efforts.

C.  SACGHS Survey of the Genetic and Genomic Information Needs of
Consumers and Patients

Methodologies

To elucidate the genetic 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 and a web-
based survey of the health advocacy community. These strategies provided the Committee with additional
data to inform their recommendations.

Semi-Structured Interviews. A list of 30 individuals widely regarded as leaders in genetics advocacy for
consumers and patients was generated by SACGHS members and staff, as well as by attendees of an
annual NCHPEG meeting. Based on this list, between December 2008 and February 2009, SACGHS
conducted semi-structured interviews with 11 experts (see Appendix D-1) in the fields of disease and
disability advocacy, genetics services for patients, health education and communication, for-profit DTC
genetics service companies, and science and genetics education of the public. The purpose of the
interviews was to collect data on current and emerging needs of consumers and patients as their lives are
increasingly influenced by genomics, and to inform the development of a survey. The interviews were
conducted by telephone with a lead interviewer from the National Human Genome Research Institute and
with one or two experts participating in each interview. The lead interviewer used an interview script with
general themes asked of all experts and specific questions based upon the interviewees’ expertise (see
Appendix D-1). These themes were used to help construct the web-based survey.

Web-Based Survey. Informed by the interviews, SACGHS developed a 12-item online survey instrument
(see Appendix D-2) to collect data from the genetics and health advocacy communities regarding their

opinions on the genetic and genomic 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 genetic education for the public. The survey also was distributed by the Genetic Alliance, a nonprofit health advocacy organization committed to transforming health through genetics. 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. The survey took about 10 minutes to complete and participants could opt out of answering any of the questions. An invitation to participate in the survey, with a hyperlink to the survey instrument was sent to approximately 1,100 individuals. The survey system received 337 whole or partial responses.

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 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 approaches used were designed to gather data in different formats from different constituencies. 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 addressed above, this process provides a snapshot of the needs of consumer and patients who have varying degrees of involvement in genetics.

Survey Findings

Semi-Structured Interviews

An analysis of the qualitative interview transcripts identified 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 and genomics. 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, complex traits, and how 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 our knowledge of test validity and utility. These misunderstandings are compounded by the difficulty consumers have in finding accurate information about genetics and genomics.

The interviews also explored various approaches to genetics education. Suggestions included the need to improve genetic and genomic education among 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 about genetics and genomics. 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 and genomic testing and services, clarify the extent to which laboratory tests are regulated, and should 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. These themes, aggregated as key findings, are explored in greater detail in Appendix D-1, Table 1.

Web-Based Survey Results

Two hundred and fifty-eight individuals responded to a question asking in what state they work, providing information on the geographical distribution of service provision. Respondents work in 39 states plus the District of Columbia. This distribution, shown in Appendix D-3, Figure 1, demonstrates that 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.

The respondents represent a wide variety of organization types, including health care organizations (47) advocacy groups (53), academic institutions (66), private industry (29), public health organizations (14), and other (60). The “other” category, which required a free-text response, included community-based health organizations, nonprofit organizations and support groups, and private hospitals and private health care practices. This distribution is presented as a pie chart in Appendix D-3, Figure 2.

Respondents were asked to rate how important genetics is to their organization. Sixty percent felt that genetics was important or very important to their organization, while only 1 percent stated that it was not at all important (see Appendix D-3, Table 2). About 55 percent of respondents reported that they had been involved with their organization in planning or implementing a genetics education program for seekers of genetic information.

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 D-3, Table 3). Eighty-nine percent of respondents answered this question. The most important concept was that “family history is an important tool for understanding health and disease.”

Participants also were given the opportunity to suggest more important items in a free-text response, resulting in 60 additional responses. Themes that emerged from these responses were: (1) the concept that there is a difference between disease risk and disease diagnosis; (2) understanding probabilities, as well as terms like “common” and “rare,” is essential to interpreting the results of genetic tests; and (3) genetic tests should be interpreted by people knowledgeable in genetics and genomics.

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 D-3, Table 4). Eighty-five percent of respondents answered this question. The most favored topic was where consumers would find reliable information on genetics and genomics, indicating that the ability to direct consumers to such resources may present a significant gap in available resources or awareness of existing resources. Thirty-six free-text responses suggested other important topics. Among these, the challenges of cost, insurance reimbursement, and malpractice insurance requiring practitioners to give “worst case scenarios” rather than balanced risk assessments emerged.
Respondents were asked to rank the following list of genetic education and services needs of underserved and vulnerable populations.

- Basic and relevant genetic health information
- Skills to make informed health decisions
- Culturally appropriate genetic health information
- Education about access to genetic services

First, however, they were given the option to state whether there were more pressing needs above genetics education to which 7 percent (22 of 315 responses) responded in the affirmative. There was poor discrimination among the rankings but the need for basic and relevant genetic health information was ranked highest and education about access to genetic services was ranked lowest (see Appendix D-3, Table 5).

Respondents were asked that if they were part of an organization, to report whether their organization had created educational programs to address the challenges in underserved and vulnerable populations. Fifty-three percent of respondents (189 of 337) answered this question, reporting development of educational programs to address at least one of these challenges.

The most important educational need identified (i.e., 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, even though this challenge was the lowest priority identified in the previous question. In the free-text responses to this item, a common theme was genetic education aimed at disease-specific support groups.

Eighty-three percent of participants responded to a request to rank 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 direct-to-consumer marketing of genetic tests before there is evidence of utility and lack of access to genetic services for consumers and patients (see Appendix D-3, Table 6).

Among the 29 free-text responses to this item, additional important barriers included 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 language used to educate consumers on genetics and genomics.

In a series of questions, the respondents were asked to rank a set of six potential roles for three levels of government: Federal, state, and local. Appendix D-3, Table 7 shows these rankings.

At all levels of government, funding was ranked as the key role. The key secondary 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. Another very low priority at all three levels of government was education about the licensing of genetic health care providers.

Education about anti-discrimination laws was determined to be of high priority for all three levels of government, echoing the sentiments from previous survey items that the public has a fear of being discriminated against based on genetic tests.
Among the 21 free-text responses regarding the role of local government, 13 indicated that there is no role in genetics and genomics education for local government. 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.

**Suggested Priorities for the Department of Health and Human Services**

In a free-text box, respondents were asked for their opinion about the role that the U.S. Department of Health and Human Services (HHS) should play to improve genetics education for those seeking information about genetics as it relates to health. Nearly 200 responses were received. 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. Summary**

In general, the literature review found that the public has been supportive of genetic testing when it is used for improving disease diagnosis and prevention. The literature review conducted in 2009 by AED found that although consumers have a limited understanding of genetic testing, they have positive attitudes about genetic testing and are generally motivated to seek information and undergo testing. However, it is not clear from the literature that the public understands how to use genetic information to optimize health. Knowledge beyond awareness, such as how to process such information and use it in decisionmaking, is limited. Levels of genetic knowledge also have been found to differ by race, ethnicity, and socioeconomic background.

The literature highlights that genetic tests are not all alike, and the particular disease risk being tested influences awareness, attitudes, and understanding. Consumers would benefit from an increased understanding that there are many types of genetic and genomic tests, and there are many contexts in which they are used. Consumers would prefer to learn about genetic tests from their health care providers, but most physicians are not trained in genetics and recognize the limitations in their knowledge and expertise and are therefore reluctant to order genetic tests and provide 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.

Consistent themes emerged from the SACGHS survey data. Consumers get information about genetics and genomics from the media and their health care providers. Consumers understand that genes and behaviors are related to health outcomes, but knowledge of complex traits and the multifactorial basis of disease are not well understood. Survey respondents approached consensus regarding consumers’ 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.” An important concept is the understanding that using genetic information can
optimize health. For consumers to understand genetic testing, they must appreciate the distinction between the risk for a disease and its diagnosis. There is consensus 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.

Despite the availability of DTC testing, consumers still prefer to have genetic tests done in their doctor’s office. 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.

Even though much of the data that informs this report was collected shortly after passage of GINA, concern about confidentiality and disclosure of genetic information that might lead to loss of a job or insurance persists. The fear of DNA being collected without consent was also expressed.

Health literacy in genetics for health professionals and consumers is considered a gap, and an underpopulated genetic workforce is a barrier to rectifying this problem. State and Federal governments are viewed as having important roles in educating consumers and health care providers alike. There also is a belief that the Federal government should regulate and evaluate genetic tests and determine who is qualified to provide genetic services. The Federal government is seen as the logical repository for educational information and should serve as a clearinghouse for this information. Providing funding for educational programs is considered a primary role of government.

Review of current literature, findings from a SACGHS survey, and interviews exploring consumer attitudes and beliefs about genetics and gaps in genetics and genomic education, point to an underlying need for improved genetic literacy beginning in the formative years and continuing throughout the lifespan. The complexity and rapid evolution of knowledge and technology related to genetics and genomics 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.
V. Activities of Selected Federal Agencies

In August 2003, a survey was distributed to 16 SACGHS ex officio agencies to obtain information about Federal activities related to the education of professionals in genetics. The agencies were asked to provide: (1) information on their overall efforts to assess genetics workforce needs and to address genetics education and training of professionals in both health and nonhealth-related fields; (2) a list of specific activities the agency funded in this area for the preceding year; and (3) specific information about the nature and purpose of the activity, its target audience, and funding information.

Seven ex officio agencies—the Department of Commerce (DOC), the Department of Defense (DOD), the Department of Energy (DOE), the Department of Justice (DOJ), and three Department of Health and Human Services (HHS) agencies—the Centers for Disease Control and Prevention (CDC), the Health Resources and Services Administration (HRSA), and the National Institutes of Health (NIH)—submitted information about their ongoing activities in response to the request. Eight ex officio agencies—the Department of Labor; the Equal Employment Opportunity Commission (EEOC); and six HHS agencies (the Administration for Children and Families (ACF), the Agency for Healthcare Research and Quality (AHRQ), the Centers for Medicare & Medicaid Services (CMS), the Food and Drug Administration (FDA), the Office for Civil Rights (OCR), and the Office for Human Research Protections (OHRP)—reported that they were not performing or funding any activities relevant to the SACGHS request but do engage in outreach and educational activities in other areas. Results of this survey were presented to SACGHS on October 23, 2003. 

With the rapid expansion in relevant genomics information over the intervening five years, SACGHS elected to repeat the prior survey of Federal agencies with those that currently have ex officio representation on the Committee. The final version of the 2008 survey was shortened from the original survey, which had required agencies to enumerate specific projects with great granularity.

The 2008 Federal survey consisted of a mix of closed- and open-ended, narrative-type response questions. These questions explored themes 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.

The survey was distributed to ex officio agency representatives to SACGHS in late 2008 and early 2009. Nonresponders were contacted by e-mail or telephone to prompt completion of the survey.

Ten ex officio agencies—DOC, DOD, DOE, the Federal Trade Commission (FTC), EEOC, the National Science Foundation (NSF), and four HHS agencies—CDC, CMS, HRSA, and NIH—submitted information about their ongoing activities in response to the request. Six ex officio agencies—the Department of Education, and five HHS agencies (ACF, AHRQ, OCR, OHRP, and the Substance Abuse and Mental Health Services Administration)—reported that they were not performing or funding any activities relevant to the SACGHS request but do engage in outreach and educational activities in other areas. The Department of Veterans Affairs reported that it conducts activities but was not able to complete the survey due to a change in personnel.

The discussion below provides a brief overview of the agencies reported genetics and genomics activities, the criteria used to determine what types of educational activities to pursue and how these activities relate

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to the agency’s mission, the target audience for the educational activities, and the identification of future needs in genetics and genomic education and training.

For agencies that responded to both SACGHS surveys (in 2003 and 2008), information is provided that compares and/or contrasts the reported material.

**Data Limitations**

The surveys conducted in 2003 and 2008 yielded numerous examples of genetics and genomics educational activities in federal agencies. However, there are several important limitations that affect interpretation of the data including that the agencies that participated in 2003 and 2008 were not the same making precise comparisons difficult. Although six agencies responded to both surveys, which provided information on the growth of programs or changes in educational priorities, four agencies participated only in the 2008 survey. In addition, the level of detail varied across responses. Some agencies provided URLs for 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, there are limitations in interpreting this information both among organizations and across time due to vagaries in defining health professional education and incomplete data on the funding of intramural or nonprogrammatic activities (e.g., website architecture and content, agency staff time and effort). Furthermore, given the range of genetic and genomic educational activities and training programs conducted by federal agencies since the first survey in 2003, the individuals who responded to the surveys may not have been aware of all the programs, Internet resources, printed publications, or targeted training programs initiated by specific divisions within their agency.

**Agencies Responding to the 2003 and 2008 Surveys**

**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 on genetics and genomics. These activities were viewed as essential to realizing 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 others to develop programs and educational tools on genetics and genomics targeted to the public health workforce and/or the clinical health care workforce. Additional activities reported at that time were focused on appropriately utilizing genetic and genomic technologies and ensuring high-quality genetic testing. In all, CDC reported 28 activities in their 2003 survey response.

In 2008, CDC reported that as genetics and genomics become more integral to public health research and practice, the need for genetics expertise in public health has become even greater than previously reported. CDC’s target audiences for genetics education activities include public health administrators, medical school and residency training programs to include faculty and students, primary care and specialty physicians, epidemiologists, health educators, laboratorians, and environmental health workers; and encompassing both the internal CDC workforce and external clinical and public health providers. The survey response noted that CDC’s role in genetic education and training of professionals is to promote the effective and responsible application of genomics knowledge and tools to promote population health that
spans multiple applications, including chronic disease, environmental health, occupational health, and infectious disease. Although, the agency reported on 16 existing genetics education programs, due to limited available resources to assess educational needs among professionals, and to develop and disseminate training tools and curricula in collaboration with their partners, CDC reported that they are not currently able to develop this area fully and respond to emerging developments in genomics.

Currently, education and training activities in genomics 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 genomics knowledge and tools for early detection, disease prevention, and health promotion in populations.

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 (i.e., lawyers, judges, and law enforcement professionals). Specific needs addressed through the genetic education and training efforts at NIST included 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 genetic 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 (CE); 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 DOJ. (See Appendix E-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.

DOD

The DOD health 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. DOD considers genetics education and training as integral to the functioning of the military health care system
and has focused significant efforts to ensure that genetics is appropriately integrated and that staff is
deductively educated in genetics and ethics.

In 2003, learning needs in the evolving fields of genetics and genetic technologies were identified through
assessment and consideration of applicable practice standards. This effort ensured that new services and
technologies were integrated with organized implementation plans 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.
Genetics also has been a component of CE programs for clinical specialties such as pediatrics, oncology,
and obstetrics and gynecology. These programs and curricula are evaluated using academic, professional,
and CE association methods to determine the impact and effectiveness of these activities.

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. The 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. Currently, the U. S. military is the most experienced practitioner of
pharmacogenomic screening on a large, population-based scale. In addition to ongoing genetic testing
programs, DOD has developed a comprehensive DOD-wide newborn screening laboratory program and
has plans to create a general genetics division under the supervision of an Air Force geneticist. (See
Appendix E-DOD for details.)

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). In addition, DOD has multiple inter-departmental relationships engaged in
personalized medicine programs and EHR standardization efforts pertaining to genomics. 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.

DOE

In 2003, the DOE survey response focused on the new capabilities emerging in genetics and the mapping
of the human genome as the context for its activities. At that time, DOE noted that in order to make the
best use of new capabilities in science, education in genetics and genomics was essential. DOE also
focused on some of the social implications of the mapping of the genome and, along with NIH, has
developed 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.
DOE’s commitment to education in genetics and genomics is consistent with its view of science and support of interdisciplinary research. The 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.

The agency’s original survey response listed 26 primarily educational activities that targeted a variety of audiences, including underserved populations, the judiciary, and academia. Among the highlights of 13 years of DOE educational efforts was 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.

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 genomics 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 genomics, which are aimed at practicing professionals, K-12 teachers and students, and graduate students. These and other educational resources about genomics can be found at the JGI website, www.jgi.doe.gov/education.

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 E-DOE for details of additional projects.)

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 relating to the education and training of practicing health care professionals, graduate students, residents, and fellows. For example, HRSA has provided ongoing funding for Area Health Education Centers (AHEC) 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 genomics, and include programs in the Maternal and Child Health Bureau and the Bureau of Health Professions. (See Appendix E-HRSA for details of these programs.)

HRSA developed targeted educational products from 2003 to 2009 that include web-based materials, newsletters, workshops, and printed materials about genetics geared specifically to 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 genomics and analyzing the genetics workforce. HRSA has also conducted more recent genetic workforce analysis, Assessing Genetic Services and the Health Workforce, 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 E-HRSA).

Through the 2008 Newborn Screening Saves Lives Act (Pub. L. No. 110-204), 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, partnering with the National Newborn Screening and Genetics Research Center, Genetics and Newborn Screening Regional Collaborative Groups, March of Dimes, and the Association of Public Health Laboratories.

NIH

NIH is the steward of medical and behavioral research for the Nation. Its mission is science in pursuit of fundamental knowledge about the nature and behavior of living systems, and the application of that knowledge to extend healthy life and reduce the burdens of illness and disability. NIH accomplishes this mission by 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 and genomics are translated into health practice.

Most 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, including the National Cancer Institute (NCI), National Human Genome

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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. An example of this effort was the development of a series of articles on genomics medicine published in the New England Journal of Medicine from 2002 to 2004.\(^{327}\) This series included articles about population screening and the ethical, legal, and social implications of genomics and genomic medicine. Workforce assessment activities were also carried out at NIH, 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.

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 National Coalition for Health Professional Education in Genetics (NCHPEG).\(^{328}\) 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.

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 E-NIH for a listing of trans-NIH programs and individual institutes involved in these training programs.)

The trans-NIH programs were developed after passage of the NIH Reform Act of 2006 that established a Common Fund (CF) to support programs that might not otherwise be funded by a single institute or center due to their cross-cutting and potentially risky nature, but whose outcomes are expected to have exceptionally high impact on the scientific community. Several Common Fund programs support activities involving training and education in genetics and genomics. Two such programs, the National Centers for Biomedical Computing (NCBCs) and the Interdisciplinary Research (IR) program, support a number of extramural activities relating to genetics and genomics.

### NIH Individual Institute Programs

**NCI**: NCI reported three programs aimed at educating professionals and trainees about genetics or genomics in their 2008 survey response.\(^{329}\) One of the projects supported by NCI, 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 direct-to-consumer genetic testing. NCI also has developed a wide range of web-based resources focused on genetics and genomics 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 DHHS Family History page.\(^{330}\)

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NHGRI: NHGRI listed several activities related to genetics education and training in the 2008 survey. 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 E-NHGRI for details about these programs and meetings.)

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 CE online course on genetics and hearing loss that is currently taught through Gallaudet University. (See Appendix E-NIDCD for details about this program.)

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. For example, along with the Josiah Macy Jr. Foundation and the American Dental Education Association, NIDCR provided conference funding for a major study initiative, “New Models of Dental Education.” This initiative convened a panel in February 2007 that examined the implications of genetics in clinical dental practice and education. 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 E-NIDCR for details about these programs.)

National Institute on Drug Abuse (NIDA): NIDA identified the Division of Basic Neuroscience and Behavioral Research and the Office of Science Policy and Communications as having primary responsibility for genetics education and training. Activities at NIDA 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 or genomics. (Details of meeting support can be found in Appendix E-NIDA.)

Genetics research has tremendously increased 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. The 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 vast and ever 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.

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 as articulated in its Strategic Directions. Between 2003 and 2009, NIA supported one institutional training grant award entitled, Neurobehavior, Neuroendocrinology and Genetics of Alzheimer Disease. This project—whose goals were to increase the understanding of the pathogenesis of
Alzheimer disease and foster development of new therapeutic approaches—provided post-doctoral training in clinical research.

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.

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.

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. 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. (See Appendix E-NLM for additional information on NLM programs.)

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Agencies Responding Only to the 2008 Survey

Several ex officio agencies that did not respond to the 2003 survey did provide a response to the 2008 survey. These agencies include CMS, FTC, EEOC, and NSF. Summaries of their reported genetics education and training activities follow.

CMS

CMS regulates all 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, under the Center for Medicaid and State Operations, has the responsibility for implementing the CLIA Program and 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.

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.

FTC

FTC deals with issues that touch the economic lives of Americans and 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.

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 direct-to-consumer genetic tests. The fact sheet, *At-Home Genetic Tests: A Healthy Dose of Skepticism May be the Best Prescription*, 332 provides consumers with clear information to make well-informed decisions when considering whether to purchase direct-to-consumer (DTC) genetic tests and answers questions about the usefulness of such tests. More than 16,000 copies of the print version of the consumer fact sheet have been distributed since July 2006. The fact sheet is also available on the FTC website where it has been accessed more than 18,000 times since 2006. 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.

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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 genetics education and online resources include detailed information about Title II of GINA, and this agency provides training on the legal prohibitions against employment discrimination on the basis of genetic information. (See Appendix E-EEOC for a listing of these trainings.)

Once the regulations implementing Title II of GINA become final, EEOC plans on conducting additional training sessions on the legal requirements of Title II for lawyers, human resource professionals, small business owners, and other interested parties.

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. Therefore, in addition to funding research in the traditional academic areas, the agency also supports "high-risk, high pay-off" ideas, novel collaborations, and numerous projects. 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 the Directorate of Education and Human Resources, in collaboration with the Directorate for Biological Sciences, administers approximately 50 active awards that directly or indirectly promote genetics or genomics education for K-12, undergraduate, or graduate students, or for the general public. Although NSF has no programs that specifically target genetics education, there is the recognition 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 and genomics while targeting a broad range of topics in biology such as molecular/cellular biology, evolution, biodiversity, and ecology.

Many of the Directorate of Education and Human Resources’ active awards have a core objective relating to genetics and/or genomics. 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 E-NSF for a detailed listing of programs, programmatic goals and individual NSF funded projects with core objectives relating to genetics and genomics).

D. Summary

To obtain information about Federal activities relating to genetic education and training of health professionals and consumers in genetics and genomics, surveys were distributed to SACGHS ex officio agencies in 2003 and again in 2008. Four agencies reported activities only in 2008; five agencies replied to both survey requests reporting no activities relevant to the survey questions; and six agencies responded to both surveys, allowing comparative analysis of the growth of activities in these agencies in the intervening years. Not surprisingly, there was a significant expansion of activities related to genetic
and genomic education and an increased emphasis on activities directed to the public. A brief comparison
of responses from the 2003 and 2008 surveys for the agencies that responded to both surveys follows:

- CDC has been very active in genetics education efforts and funded a number of activities in 2003. Although it has continued to expand its role in genetics and genomics education and collaborate with a number of entities, it reported in 2008 that due to limited available resources the agency is unable to develop this area fully.

- DOC’s reported activities related to genetics and genomics reside within the National Institute of Standards and Technology (NIST) and have expanded beyond cancer genetics and forensic applications targeted to practicing professionals to now include health professionals in training. NIST has also broadened its educational methods to include websites and online resources.

- 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 to incorporate genomic research into undergraduate courses and websites aimed at practicing professionals, K-12 teachers and students, and graduate students.

- Although HRSA reported 64 educational activities in 2003, 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 other health care professionals and the general public and to produce 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 support for the National Coalition of Health Professional Education in Genetics. By 2008, NIH had numerous activities within individual Institutes and through some of its trans-NIH programs. NCI, NHGRI, NIDCD, NIDCR, NIDA, NIA, and NLM all reported significant programs and educational resources for health care professionals and consumers.

The activities of the four agencies that responded only to the 2008 survey are summarized as follows:

- CMS 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 direct-to-consumer genetic testing.

- 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.

- NSF awards grants to promote genetics education for K-12, undergraduate and graduate students, and the general public.

Based on SACGHS surveys of Federal agencies conducted in 2003 and 2008, a considerable number of activities in genetics and genomics have lead to development of educational programs, materials, and resources for a variety of professional disciplines and for the public. A number of government-sponsored websites house and/or link to government and nongovernment resources. An effort to evaluate and consolidate these materials and maintain an entry point for their access would facilitate dissemination of accessible, credible genetic and genomic information to health professionals and the public.
VI. Conclusions and Recommendations

Conclusions

The Secretary’s Advisory Committee on Genetics, Health, and Society (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. However, these efforts have not kept pace with the emerging understanding of the human genome and rapid evolution of genomic technologies. The following discussion summarizes SACGHS’ findings and its recommendations that address the needs of health care professionals, public health providers, and consumers and patients.

A. Health Care Professionals

A review of the literature and findings from SACGHS surveys of health professional organizations revealed that much work has been done to develop genetics educational curricula and programs at the undergraduate, graduate, and continuing education level. However, SACGHS also found that these efforts often exist in isolation and are not always linked to accreditation, certification, and licensure programs. Recent collaborative efforts by professional societies to align genetic competencies with educational objectives are a promising step forward but need to be replicated and extended if progress is to continue.

SACGHS found that several barriers impede incorporation of genetics and genomics 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. SACGHS survey respondents indicated that competing curriculum priorities is the most significant barrier. Incorporating genetics across disciplines and topic areas is an important way to overcome this barrier.

Compounding the barriers discussed above, there is an insufficient number of M.D. and Ph.D. geneticists available to provide genetics education to health care professionals. To address this shortage, others trained in genetics, such as genetic counselors, pharmacists with pharmacogenomic training, and nurse geneticists should be encouraged to step into educator roles. Genetic education programs that use trained peer educators have been successful and well accepted by health care professionals. Also, enhancing the use of clinical decision support tools, promoting the importance of family history, and ensuring adequate reimbursement for genetic services are among other approaches that would support the optimal use of genetics and genomics in health care.

B. Public Health Providers

To assess the genetics education needs of public health providers, SACGHS reviewed findings from the literature and conducted a survey of public health providers. The survey used 12 core competencies developed by SACGHS. Although survey respondents identified all 12 competencies as important, they were the most confident in utilizing family history to assess predisposition to disease. This finding suggests that some genetic-related information is accepted by public health providers and that they agree that promoting the role of family history in population health will contribute to improved public health.

The literature review and SACGHS survey revealed several barriers that limit the uptake and appropriate use of genetic and genomic services by public health providers. These barriers include a workforce with...
diverse education and training needs, a significant number of public health workers trained before the
genomic era, and lack of an understanding of the need for genetics and genomics expertise in public
health practice.
To address the diversity of the public health workforce, educational approaches should target the unique
training needs, and the range of expertise and genetic literacy, of each type of public health professional.
These educational approaches should include curricula on cultural competence, social and economic
determinants of health, and ways to address and reduce health disparities. Survey respondents identified
strategies to ensure that genetic services and information are available to vulnerable and underserved
populations. These strategies ranged from local-level community engagement to policy development at
the federal level. Identifying effective educational models for public health providers who serve these
communities will also help ensure that appropriate genetic services are provided to vulnerable and
underserved populations.

C. Consumers and Patients
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.
The federal government and private-sector organizations have developed family history tools as one
means for individuals and families to gain health literacy and to take a more active role in preventing and
managing disease, particularly inherited conditions. These tools can help both consumers and health care
professionals in risk assessment, but for optimal use of this tool, electronic health records (EHRs) must be
capable of accepting family health history data that have been provided by a consumer (e.g., My Family
Health Portrait).
Consumers obtain information about genetics and genetic testing from a variety of sources including their
doctors and the media, particularly the Internet. Efforts to improve the quality and accessibility of web-
based resources will be important to provide information in a manner preferred by consumers.
Additionally, improved genetic and genomic knowledge among health care professionals will be needed
as consumers rely on them as trusted sources of information.
Given the wide range of educational levels and motivations among individuals seeking genetic and
 genomic 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. In addition, educational resources should be culturally appropriate and tailored to the specific
needs of communities and validated using certified health educational standards to ensure comprehension
by the target audience.

D. Selected Federal Activities
To obtain information about Federal activities related to genetics and genomics education and training of
health professionals and consumers, surveys were distributed to SACGHS ex officio agencies in 2008 and
compared to 2003 survey results. Some agencies participated in both surveys, while several others
participated only in 2003 or 2008.
The survey findings suggest that the number of genetics educational programs and resources established
by Federal agencies for professionals and consumers have increased over the past five years and are
helping to increase genetic knowledge and address part of the educational needs. However, as previously
discussed, the data from the literature and other SACGHS surveys suggest that these efforts are not
sufficient and in particular there is a lack of attention to health disparities. For example, 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. Federal efforts in professional and consumer education
should be continued as a way of improving competency in the rapidly evolving fields of genetics and
genomics.

**Recommendations**

Seven recommendations are presented 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. Innovative
approaches that coordinate the efforts of entities controlling health professional education and training are
needed.

1. HHS should convene a workshop to identify innovative education and training approaches that will
promote integration of genetics and genomics into clinical care. The workshop would include
representatives of HHS agencies and other federal departments with established programs in genetic
and genomic professional education; representatives of health professional organizations engaged in
accreditation, certification, and continuing education efforts; and private sector entities that provide
genetics education. The workshop goals are to:

   A. identify successful education and training guidelines and models that are outcomes based;
   B. identify current funding streams for developing and promoting genetic and genomic education as
      well as gaps in funding;
   C. recommend mechanisms for expanding and enhancing the content needed to prepare health care
      professionals for personalized genomic health care;
   D. recommend mechanisms for evolving standards, certification, accreditation, and continuing
      education activities to incorporate genomic content;
   E. determine the need, and if appropriate, appoint an ongoing advisory panel to facilitate
      implementation of the approaches identified during the workshop; and
   F. publish findings and recommendations and develop a plan to monitor the outcome of these
      efforts.

**Recommendation 2**

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.

2. HHS should promote the development and implementation of targeted genetic and genomic education
and training models for health care professionals and public health providers serving underserved and
underrepresented groups and populations. Specifically, HHS should:

   A. direct research funding to identify effective educational models for health care professionals and
      public health providers in underserved communities;
   B. identify and support programs to increase the diversity of the health care workforce in general
      and the genetic-specific workforce; and
C. ensure that consumers and representatives of rural, minority, and underserved communities participate in the process of developing education and training models to assure that they are culturally and linguistically appropriate and tailored to the unique needs of these diverse communities.

Recommendation 3

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.

3. Tapping the expertise of its agencies with relevant missions in public health (e.g., HRSA, CDC, the Indian Health Service, and NIH), HHS should assess the workforce to determine the number of public health providers with responsibilities in genetics and genomics to ascertain current trends and future needs, to identify education and training needs, and to promote leadership development in the field. Based on this assessment, HHS should:

A. support and encourage the incorporation of basic genetic and genomic core competencies in the knowledge base of federal and nonfederal public health providers, and specific competencies for those whose responsibilities require specialized genetic knowledge, such as environmental interactions and risk assessment for population-based genomics; and

B. fund educational programs based on these competencies.

Recommendation 4

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.

4. HHS should endorse, fund, and maintain an Internet entry point or portal to a vetted collection of comprehensive, accessible, and trustworthy web-based genetic information and resources for consumers. This portal should utilize existing governmental resources (such as those developed by NIH and CDC) in addition to new materials. The portal should have the ability to be customized by the consumer in order to access desired information easily. HHS should assure that:

A. these resources include scientifically validated information and/or links to credible information regarding topics such as genetic contributions to health and disease, gene-environmental interactions, genetic testing, and legal protections against genetic discrimination;

B. these resources include links to information that are not web-based such as television and radio programs and print materials; and

C. the availability of this portal be promoted using a wide range of strategies from collaborating with developers of Internet search engines to working with community leaders at the local level. Mechanisms to alert interested persons to updates and new information should be developed.

Recommendation 5

With the vast increase in scientific knowledge stemming from genetic and genomic research and 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.
5. HHS should support research that identifies methods that are effective for translating genetic and genomic knowledge into information that consumers and patients can use to make health decisions. Specifically, HHS should:

A. support 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 (e.g., radio, television, print, and mobile phones) and provide for translation of materials into locally predominant languages; and
C. support the dissemination of these educational programs and materials into science and/or health education initiatives through collaboration with other relevant departments and agencies such as the Department of Education and the National Science Foundation.

**Recommendation 6**

Family health history tools (e.g., My Family Health Portrait) are a powerful asset for consumers and health care professionals to use in risk assessment and health promotion.

6. HHS should support continued efforts to educate health care professionals, public health providers, and consumers about the importance of family health history.

A. For health care professionals, HHS should support the use of family history in clinical care through development of clinical decision support tools and mechanisms to integrate pedigrees into electronic health records.
B. For public health providers, HHS should promote research identifying the role of family history in population health.
C. For consumers, HHS should:
   1. promote research on how consumers use family history to make health care decisions;
   2. assess the effects of gathering family histories within diverse cultures and communities and among individuals where family histories are unavailable;
   3. expand public health awareness programs and patient information materials on the importance of sharing family history information with primary care providers; and
   4. promote the embedding of educational materials in family history collection tools directed to consumers and ensure access for all by providing these tools in various formats.

**Recommendation 7**

Given the reality that health care professionals and the professional societies representing them are unlikely to invest significant resources in education and training in content areas for which services are only partially or not reimbursable, a critical step in promoting increased knowledge of genetics and genomics among health care professionals is ensuring adequate reimbursement.

7. In order to increase incentives and encourage investment by public and private organizations in education and training in genetics and genomics, and to increase the willingness of health care professionals to participate in educational programs, the Secretary should:

A. ensure adequate reimbursement for health care professional time spent in direct patient care delivering genetic and genomic services such as interpretation of genetic tests and collecting family history;
B. ensure adequate reimbursement for all members of interdisciplinary teams providing genetic services and for distance consultation and telemedicine services that are used in underserved regions; and
C. act on the recommendations in the 2006 SACGHS report *Coverage and Reimbursement of Genetic Tests and Services.*
APPENDICES

Appendix A: Literature Review

1. Literature 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, PsycINFO, 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
2. Genetics and Genomics Competencies for Selected Health Care Providers

National Coalition for Health Professional Education in Genetics

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

1. 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.
2. Understand that health-related genetic information can have important social and psychological implications for individuals and families.
3. Know how and when to make a referral to a genetics professional.

In the knowledge domain, all health professionals should understand:

1. basic human genetics terminology,
2. the basic patterns of biological inheritance and variation, both within families and within populations,
3. how identification of disease-associated genetic variations facilitate development of prevention, diagnosis, and treatment options,
4. the importance of family history (minimum three generations) in assessing predisposition to disease,
5. the interaction of genetic, environmental, and behavioral factors in predisposition to disease, onset of disease, response to treatment, and maintenance of health,
6. the difference between clinical diagnosis of disease and identification of genetic predisposition to disease (genetic variation is not strictly correlated with disease manifestation),
7. 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,
8. the potential physical and/or psychosocial benefits, limitations, and risks of genetic information for individuals, family members, and communities,
9. the resources available to assist clients seeking genetic information or services, including the types of genetics professionals available and their diverse responsibilities,
10. 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
11. 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:

1. gather genetic family history information, including at minimum a three-generation history,
2. 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,
3. explain effectively the reasons for and benefits of genetic services,
4. use information technology to obtain credible, current information about genetics, and
5. 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:

1. appreciate the sensitivity of genetic information and the need for privacy and confidentiality, and
2. 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.

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. 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. This updated set of competencies conforms to requirements of the Liaison

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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.\textsuperscript{335} 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.\textsuperscript{336}

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).

Reflecting the scientific progress beyond single-gene disorders, in January 2007, ASHG released policy recommendations concerning DTC genetic testing technologies.\textsuperscript{337} 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. 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.

The American Academy of Pediatrics’ Committee on Genetics has authored numerous policy and professional practice statements on various heritable and complex health conditions.

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.

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. The workshop focused on incorporation of genetics and genomic medicine into maternal and 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;

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

Competencies for Nurses

In 2005, genetics competencies for all practicing R.N.s 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.344

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

Competencies for Physician Assistants

Four 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. In response, the Physician Assistance Education Association is creating faculty development opportunities, monitoring and reporting innovations in genetics education, developing curricula resources for best

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345 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.
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 established the Essential Physician Assistant Guidelines for Genetics and Genomics.\textsuperscript{346} 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.

**Competencies for Genetic Counselors**

Practiced-based competencies were issued by the American Board of Genetic Counseling in 2008.\textsuperscript{347} 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
- 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

Competencies for Pharmacists

Pharmacists are recognized as medication experts who improve overall patient care through partnering with physicians. 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 professional role of pharmacists.” 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. 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.

Although recognition of the inherited differences in drug effects was documented as early as 1931, 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. 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.

3. Academic Preparation, Licensure, and Accreditation of Professional Schools

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. 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. 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. Driving this need is a significant shortage of medical genetics experts prepared to address the onslaught of implications stemming from genetic science. 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.

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. 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.

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. 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.,

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single-gene disorders, chromosomal aberrations) and skills related to their clinical management. Genetic content is similarly incorporated in the final examination.\textsuperscript{365} 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.\textsuperscript{366}

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.\textsuperscript{367} 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.\textsuperscript{368} 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.\textsuperscript{369}

**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.\textsuperscript{370} Nursing contains great academic and professional heterogeneity stemming from multiple academic pathways to becoming a R.N.; two accrediting bodies for academic curricula with varying requirements; presence of nursing education programs that lack accreditation; and numerous specialty advanced practice pathways with variable routes to certification (e.g., Family N.P., Pediatric N.P., Geriatric N.P.). 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.


\textsuperscript{368} 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.


Entry-level professional R.N.s 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. Advanced Practice Nurses (N.P.s, clinical nurse specialists, certified nurse midwives, certified registered nursing anesthetists, etc) are R.N.s 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 R.N.s 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.

The need for education of nurses in genetics is well documented. 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. 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.

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. 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 R.N. 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 R.N. professional license. Little genetic content is contained in NCLEX and certification examinations, and at the master’s level, there is significant 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 R.N.s. (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.\(^{377}\) 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.\(^{378}\) 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 R.N.s 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 R.N. licenses.\(^{379}\)
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 R.N. professional license.

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.\(^{380}\) 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.\(^{381}\) 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).\(^{382}\) Although covering
single-gene disorders and other hereditary conditions, the PANCE does not include a genetics section or
genomics content.

\(^{377}\) 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.  
\(^{378}\) National League for Nursing Accrediting Commission, Inc. NLNAC 2008 Standards and Criteria. See
\(^{379}\) NurseWeek. Nursing Continuing Education Requirements by State. See www.nurse.com/ce/Requirements.html;
\(^{380}\) National Institutes of Health. Physician Assistant Competencies for Genomic Medicine: Where We Are Today and How to
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. 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 years by NCCPA via the Physician Assistant National Recertifying Exam, with genetic examination content similar to the PANCE.

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.

The ABGC accredits genetic counselor programs. Revised in March of 2009, the expanded genetic and genomic content requirements are built into accreditation standards. 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. The primary professional society representing genetic counselors, the National Society of Genetic Counselors, provides CE units per pre-approved criteria and sponsor initiated activities. The

ABGC CE program currently is being restructured to meet the 5-year recertification cycle going into effect in 2010.

**Education 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. 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.

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.

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.

To meet the pharmacogenomic educational needs of U.S. Colleges of Pharmacy, the Pharmacogenomics Education Program: Bridging the Gap between Science and Practice (*PharmGenEd™*), 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™* 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™* 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™* 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).
Appendix B: 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 (AACP)
- American College of Clinical Pharmacology
- 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)
2. Health Care Professional Organizations’ Survey Methodology

The main body of the survey instrument consisted of 15 open- and close-ended questions developed by SACGHS (see Appendix B-3). Close-ended questions were in both multiple-choice and Likert scale formats. Organizations were also asked to complete a narrative description of ongoing genomics-related projects. The draft instrument was piloted with board members of the NCHPEG, refined, and subsequently reviewed by a survey methodologist to maximize survey validity. The instrument explored several major themes including the organizations’ perceived role in, and priority ascribed to genomics education; barriers to enhancing their role in genomics education; and a description of their past, present, and planned efforts around genomics education.

The survey was sent via e-mail to key staff in the 60 targeted organizations in January 2009. All nonresponders were contacted by e-mail and/or phone by SACGHS to maximize response rates. Thirty-six responses were received (60 percent).

Survey data were complied by SACGHS staff and entered into Excel spreadsheets. The organizations were divided into three major divisions: genetic-specific organizations; nongenetic organizations; and Federal advisory committees, and analyses 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.**

a. 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

b. 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

c. 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

d. 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

e. How proficient and comfortable would you say your organization’s leadership is with genetics and genomics education?
f. 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):

g. 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.

1. Genetics and genomics education is not applicable to the organization’s mission  
2. The organization’s leadership lacks knowledge of genetics and genomics  
3. The organization has competing priorities  
4. There is a lack of accessible educational resources for genetics and genomics  
5. Genetics and genomics is not emphasized in certifying examinations/credentialing standards  
6. The organization believes there is a lack of evidence supporting clinical effectiveness of care based on genetic or genomic information  
7. Other (please list):

8. 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) | | | | | |
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.

<table>
<thead>
<tr>
<th>Program #1</th>
<th>Program #2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Brief description</td>
<td>Brief description</td>
</tr>
<tr>
<td>Outcome measures used to evaluate program’s success</td>
<td>Outcome measures used to evaluate program’s success</td>
</tr>
<tr>
<td>External collaborators (if applicable)</td>
<td>External collaborators (if applicable)</td>
</tr>
<tr>
<td>URLs for web-based resources related to the program</td>
<td>URLs for web-based resources related to the program</td>
</tr>
<tr>
<td>Publication citations (if any) related to the program</td>
<td>Publication citations (if any) related to the program</td>
</tr>
</tbody>
</table>
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. Health Care Professional Organization’s Survey Names and Constituency or Membership of Responding Organizations

The table below lists the organizations that responded to the survey with their reported constituency or membership numbers noted. Because organizations were asked to indicate the size of their constituency or membership, those 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.

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Organization Name</th>
<th>Membership/Constituency</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td><strong>Genetic Specific (9 of 9 returned = 100 percent)</strong></td>
<td></td>
</tr>
<tr>
<td>ABGC</td>
<td>American Board of Genetic Counseling</td>
<td>2,488</td>
</tr>
<tr>
<td>ABMG</td>
<td>American Board of Medical Genetics</td>
<td>2,000</td>
</tr>
<tr>
<td>ACMG</td>
<td>American College of Medical Genetics and ACMG Foundation</td>
<td>1,500</td>
</tr>
<tr>
<td>ASHG</td>
<td>American Society of Human Genetics</td>
<td>7,500</td>
</tr>
<tr>
<td>APHMG</td>
<td>Association for Professors of Human and Medical Genetics</td>
<td>100</td>
</tr>
<tr>
<td>GNCC</td>
<td>Genetic Nurses Credentialing Commission</td>
<td>47</td>
</tr>
<tr>
<td>ISONG</td>
<td>International Society of Nurses in Genetics</td>
<td>415</td>
</tr>
<tr>
<td>NCHPEG</td>
<td>National Coalition for Health Professional Education in Genetics</td>
<td>65</td>
</tr>
<tr>
<td>NSGC</td>
<td>National Society of Genetic Counselors</td>
<td>2,400</td>
</tr>
<tr>
<td></td>
<td><strong>Federal Advisory Committees (2 of 3 returned = 67 percent)</strong></td>
<td></td>
</tr>
<tr>
<td>ACHDNC</td>
<td>Advisory Committee on Heritable Disorders in Newborns and Children</td>
<td>N/A</td>
</tr>
<tr>
<td>CGME</td>
<td>Council on Graduate Medical Education</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td><strong>Other Professional Organizations (25 of 48 returned = 54 percent)</strong></td>
<td></td>
</tr>
<tr>
<td>ACGME</td>
<td>Accreditation Council for Graduate Medical Education</td>
<td>9,200</td>
</tr>
<tr>
<td>ARC-PA</td>
<td>Accreditation Review Commission on Education for the Physician Assistant</td>
<td>163</td>
</tr>
<tr>
<td>AAIM</td>
<td>Alliance for Academic Internal Medicine</td>
<td>6,500</td>
</tr>
<tr>
<td>AAFP</td>
<td>American Academy of Family Physicians</td>
<td>94,600</td>
</tr>
<tr>
<td>AAP</td>
<td>American Academy of Pediatrics</td>
<td>60,000</td>
</tr>
<tr>
<td>AAPA</td>
<td>American Academy of Physician Assistants</td>
<td>75,000</td>
</tr>
<tr>
<td>AACN</td>
<td>American Association of Colleges of Nursing</td>
<td>625</td>
</tr>
<tr>
<td>AACP</td>
<td>American Association of Colleges of Pharmacy</td>
<td>2,910</td>
</tr>
<tr>
<td>ACCP</td>
<td>American College of Clinical Pharmacology</td>
<td>2,910</td>
</tr>
<tr>
<td>ACOG</td>
<td>American College of Obstetricians and Gynecologists</td>
<td>54,000</td>
</tr>
<tr>
<td>ACP</td>
<td>American College of Physicians</td>
<td>126,000</td>
</tr>
<tr>
<td>ACPM</td>
<td>American College of Preventive Medicine</td>
<td>2,500</td>
</tr>
<tr>
<td>ADEA</td>
<td>American Dental Education Association</td>
<td>17,000</td>
</tr>
<tr>
<td>AMA</td>
<td>American Medical Association</td>
<td>250,000</td>
</tr>
<tr>
<td>ANA</td>
<td>American Nurses Association</td>
<td>2,900,000</td>
</tr>
<tr>
<td>AOA</td>
<td>American Osteopathic Association</td>
<td>64,000</td>
</tr>
<tr>
<td>ARCOG</td>
<td>American Residency Coordinators in Obstetrics and Gynecology</td>
<td>225</td>
</tr>
<tr>
<td>AWHONN</td>
<td>Association of Women's Health, Obstetric and Neonatal Nurses</td>
<td>23,000</td>
</tr>
<tr>
<td>COMSEP</td>
<td>Council on Medical Student Education in Pediatrics</td>
<td>300</td>
</tr>
<tr>
<td>NAPNP</td>
<td>National Association of Pediatric Nurse Practitioners</td>
<td>7,000</td>
</tr>
<tr>
<td>ONCC</td>
<td>Oncology Nursing Certification Corporation</td>
<td>27,000</td>
</tr>
<tr>
<td>ONS</td>
<td>Oncology Nursing Society</td>
<td>35,000</td>
</tr>
<tr>
<td>PAEA</td>
<td>Physician Assistant Education Association</td>
<td>75,000</td>
</tr>
<tr>
<td>STTI</td>
<td>Sigma Theta Tau International</td>
<td>130,000</td>
</tr>
<tr>
<td>SGIM</td>
<td>Society of General Internal Medicine</td>
<td>2,500</td>
</tr>
</tbody>
</table>
Table 1. Organizations’ Role or Responsibility in Genetics Education and Training

<table>
<thead>
<tr>
<th>Organization Abbreviation</th>
<th>Education and Training Role Description</th>
</tr>
</thead>
</table>
| AACN                      | AACN has partnered with NHGRI and NCI on several initiatives:  
2. Assisting with creating a tool kit for faculty development.  
3. Assisting with creating a tool kit repository. |
| AAFP                      | • 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.  
• AAFP is a member of NCHPEG. |
| AAP                       | The AAP is concerned about all aspects of pediatric care, including genetics. |
| AAPA                      | 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                      | 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.  
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. This is available on our website at http://abbrc.iamonline.com/CMFiles/ABGDC CO Final5 1K |
OM- 10292008-1 06 1 .pdf. 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.

<table>
<thead>
<tr>
<th>Organization</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>ABMG</td>
<td>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.</td>
</tr>
<tr>
<td>ACCP</td>
<td>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.</td>
</tr>
<tr>
<td>ACHDNC</td>
<td>The grant program established under Section 1109 of our authorizing legislation specifies these activities. 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.</td>
</tr>
<tr>
<td>ACMG</td>
<td>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.</td>
</tr>
<tr>
<td>ACPM</td>
<td>ACPM is currently developing a CME program for its membership and broader community of primary care physicians.</td>
</tr>
<tr>
<td>ACP</td>
<td>We incorporate genetics education into our live courses and publish materials that include genetics education.</td>
</tr>
<tr>
<td>ACOG</td>
<td>Develop clinical guidelines and patient and professional resources.</td>
</tr>
<tr>
<td>AMA</td>
<td>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.</td>
</tr>
<tr>
<td>APHMG</td>
<td>We represent professors of genetics in all areas of genetics, and are involved in resident, fellow, medical student, and graduate student education.</td>
</tr>
<tr>
<td>ASHG</td>
<td>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.</td>
</tr>
<tr>
<td>AWHONN</td>
<td>Genetics information is integrated in other specialty specific content for our educational resources.</td>
</tr>
<tr>
<td>COMSEP</td>
<td>Set national curricula.</td>
</tr>
<tr>
<td>ISONG</td>
<td>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.</td>
</tr>
<tr>
<td>NAPNP</td>
<td>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.</td>
</tr>
</tbody>
</table>
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:
- 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.

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.

Planning for free online repository available to health professionals for content, tool kits, etc., related to genetics through the Virginia Henderson International Library.

**Table 2. Committees, Workgroups, or Dedicated Staff for Genetics or Genomics Education.**

<table>
<thead>
<tr>
<th></th>
<th>All Organizations</th>
<th>Genetics Specific Organizations</th>
<th>Federal Advisory Committees</th>
<th>Other Organizations</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>%</td>
<td>#</td>
<td>%</td>
<td>#</td>
</tr>
<tr>
<td>Yes</td>
<td>47%</td>
<td>17</td>
<td>78%</td>
<td>7</td>
</tr>
<tr>
<td>No</td>
<td>47%</td>
<td>17</td>
<td>22%</td>
<td>2</td>
</tr>
<tr>
<td>Not Sure</td>
<td>6%</td>
<td>2</td>
<td>0%</td>
<td>0%</td>
</tr>
<tr>
<td>Total Answers</td>
<td>36</td>
<td>9</td>
<td>2</td>
<td>25</td>
</tr>
</tbody>
</table>

**Table 3. Barriers to Providing Genetics Educational Activities**

<table>
<thead>
<tr>
<th>Barriers</th>
<th>Percent of all organizations</th>
<th>Percent of genetic-specific organizations</th>
<th>Percent of other organizations</th>
<th>Percent of Federal Advisory Committees</th>
</tr>
</thead>
<tbody>
<tr>
<td>The organization has competing priorities</td>
<td>53</td>
<td>22</td>
<td>64</td>
<td>50</td>
</tr>
<tr>
<td>Genetics and genomics is not emphasized in certifying exams/credentialing standards</td>
<td>33</td>
<td>44</td>
<td>28</td>
<td>50</td>
</tr>
<tr>
<td>There is a lack of accessible educational resources for genetics and genomics</td>
<td>22</td>
<td>33</td>
<td>16</td>
<td>50</td>
</tr>
<tr>
<td>From our organization’s perspective, there are no barriers</td>
<td>14</td>
<td>11</td>
<td>16</td>
<td>0</td>
</tr>
</tbody>
</table>
Genetics and genomics is not applicable to the organization’s mission  

| The organization believes there is lack of evidence supporting clinical effectiveness of care based on genetic or genomic information | 11 | 11 | 8 | 50 |
| The organization’s leadership lacks knowledge of genetics and genomics | 11 | 0 | 16 | 0 |

Table 4. Development of Curricular Components Responses

Because of competing priorities, the subject has not been taken up by our Council

All of our curricular offerings are optional – lives courses, content embedded within broader courses, and enduring materials that we develop.

Please see above for the current options in genetics that ONS provides. We also have a Genetics Clinical Resource Area on our website. Click here for the link:

http://www.ons.org/clinical/prevention/genetics/index.shtml

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.

optional curriculum related to diseases in women, genetic testing for women and infants

None by SGIM. The genetics in primary care faculty development curriculum or genetics through a primary care ed. is used by educators.

Please find the NCHPEG core competencies submitted and included in Appendix A-2

Individualized for PhD and undergraduate institutions. MD training falls under ACMG.

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.

The revised Baccalaureate Essentials (2008) incorporates competencies and content related to genetics and genomics.

Requirements and optional components of any area are at the discretion of our member institutions. We do not set curricular requirements.

The organization wrote a national curriculum. Individual schools or directors may use the curriculum as they see fit. Here is the chapter on Genetics:

Rationale
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.

Prerequisites
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
Table 4. Development of Curricular Components Responses

<table>
<thead>
<tr>
<th>Competencies</th>
<th>Knowledge</th>
</tr>
</thead>
<tbody>
<tr>
<td>penbreance, variable expression, and spontaneous mutations</td>
<td>1. Describe the genetic basis and clinical manifestations of the following syndromes, malformations, and associations:</td>
</tr>
<tr>
<td>Basic embryology and teratology</td>
<td>Common chromosomal abnormalities, (e.g. Trisomy 21 (CP), Turner syndrome (CP), Klinefelter syndrome (M))</td>
</tr>
<tr>
<td>Introductory history taking and physical examination skills</td>
<td>Syndromes due to teratogens (e.g. fetal alcohol syndrome) (CP)</td>
</tr>
<tr>
<td>Competencies</td>
<td>Other common genetic disorders (e.g. cystic fibrosis, sickle cell disease, hemophilia) (CP)</td>
</tr>
<tr>
<td>Knowledge</td>
<td>Single malformations with multifactorial etiology (e.g. spina bifida, congenital heart disease, cleft lip and palate) (M)</td>
</tr>
<tr>
<td>2. List common medical and metabolic disorders (e.g. hearing loss, hypothyroidism, PKU, hemoglobinopathies) detected through newborn screening programs (CP)</td>
<td>2. Use a family history to construct a pedigree (e.g., for the evaluation of a possible genetic disorder) (CP)</td>
</tr>
<tr>
<td>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)</td>
<td>3. List common prenatal diagnostic assessments (e.g. maternal serum screening, amniocentesis, and ultrasonography) and understand their use (M)</td>
</tr>
<tr>
<td>4. List common prenatal diagnostic assessments (e.g. maternal serum screening, amniocentesis, and ultrasonography) and understand their use (M)</td>
<td>4. 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)</td>
</tr>
<tr>
<td>5. Describe the use of chromosome studies in the diagnosis of genetic disorders (M)</td>
<td>Skills</td>
</tr>
<tr>
<td>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)</td>
<td>1. Use a family history to construct a pedigree (e.g., for the evaluation of a possible genetic disorder) (CP)</td>
</tr>
<tr>
<td>Not available info across all US colleges of Pharmacy, new survey in field</td>
<td></td>
</tr>
</tbody>
</table>

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

5. 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 C: SACGHS Survey of Public Health Providers

1. Survey Methodology

Using the Delphi technique, 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), the Centers for Disease Control and Prevention (CDC), the Association of State Territorial Health Officers, Training Finder Real-time Affiliate Integrated Network, and the University of Washington. 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. 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 38-item assessment tool that included demographic questions (see Appendix C-2).

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). See Appendix C-3 for a discussion of reliability analysis.

The survey included two open-ended qualitative questions. All responses were downloaded and entered into qualitative analytical software, Atlas TI. The responses were analyzed for commonalities among the responses. The results provided below highlight the most common themes that emerged.

Sample Size and Missing Data

The total sample size used in the analysis was 140 participants. There were instances in which data were missing for specific questions within each competency where the response rate was below 140. In

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393 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.


situations such as this, missing data values were recoded to equal “no answer” on the Likert scale. 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 are missing.

2. Public Health Providers’ Survey Instrument

Note: the page above was added when requirement for a survey validation ID was removed.
### Part I: Your Practices of 12 Competencies

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:

<table>
<thead>
<tr>
<th>Competency</th>
<th>Not at All</th>
<th>Not Very</th>
<th>Somewhat</th>
<th>Very</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Maintain up-to-date knowledge on the development of genomic science and</td>
<td></td>
<td></td>
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<tr>
<td>technologies within his or her professional field and programs to apply</td>
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<tr>
<td>genomics as a tool for achieving public health goals.</td>
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<tr>
<td>How important is the competency?</td>
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<tr>
<td>How confident are you in demonstrating this competency?</td>
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<td></td>
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<tr>
<td>How frequently do you apply this competency?</td>
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<tr>
<td>2. Demonstrate basic knowledge of the role that genetics/genomics plays</td>
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<tr>
<td>in the development of disease and in screening and interventions for</td>
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<tr>
<td>programs of disease prevention and health promotion.</td>
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<td>How important is the competency?</td>
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<td>How confident are you in demonstrating this competency?</td>
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<tr>
<td>How frequently do you apply this competency?</td>
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<tr>
<td>3. Describe the importance of family history in assessing predisposition</td>
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<td>to disease.</td>
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<td>How important is the competency?</td>
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<td>How confident are you in demonstrating this competency?</td>
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<tr>
<td>How frequently do you apply this competency?</td>
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<td></td>
<td></td>
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<tr>
<td>4. Identify opportunities and integrate genetic/genomic issues into public</td>
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<td>health practice, policies or programs effectively.</td>
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<td>How important is the competency?</td>
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<td>How confident are you in demonstrating this competency?</td>
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<td>How frequently do you apply this competency?</td>
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</tbody>
</table>
Part I (cont’d)

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.

How important is the competency? Not at All Not Very Somewhat Very

How confident are you in demonstrating this competency? Never 1-2 Per Year Monthly Weekly

How frequently do you apply this competency? Never 1-2 Per Year Monthly Weekly

6. Describe the potential physical and psychological benefits, limitations, and risks of genetic/genomic information for individuals, family members, and communities.

How important is the competency? Not at All Not Very Somewhat Very

How confident are you in demonstrating this competency? Never 1-2 Per Year Monthly Weekly

How frequently do you apply this competency? Never 1-2 Per Year Monthly Weekly

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.

How important is the competency? Not at All Not Very Somewhat Very

How confident are you in demonstrating this competency? Never 1-2 Per Year Monthly Weekly

How frequently do you apply this competency? Never 1-2 Per Year Monthly Weekly

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? Not at All Not Very Somewhat Very

How confident are you in demonstrating this competency? Never 1-2 Per Year Monthly Weekly

How frequently do you apply this competency? Never 1-2 Per Year Monthly Weekly
9. Conduct outcomes evaluation of available genetic/genomic programs and services to determine their effectiveness.

<table>
<thead>
<tr>
<th>How important is the competency?</th>
<th>Not at All</th>
<th>Not Very</th>
<th>Somewhat</th>
<th>Very</th>
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<tr>
<th>How frequently do you apply this competency?</th>
<th>Never</th>
<th>1-2 Per Year</th>
<th>Monthly</th>
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</table>

10. Identify the political, legal, social, ethical, and economic issues associated with integrating genomics into public health.

<table>
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<tr>
<th>How important is the competency?</th>
<th>Not at All</th>
<th>Not Very</th>
<th>Somewhat</th>
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<tr>
<th>How frequently do you apply this competency?</th>
<th>Never</th>
<th>1-2 Per Year</th>
<th>Monthly</th>
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</table>

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.

<table>
<thead>
<tr>
<th>How important is the competency?</th>
<th>Not at All</th>
<th>Not Very</th>
<th>Somewhat</th>
<th>Very</th>
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<th>How frequently do you apply this competency?</th>
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<th>1-2 Per Year</th>
<th>Monthly</th>
<th>Weekly</th>
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</tbody>
</table>

12. Identify appropriate and relevant genetics research findings that can be translated into public health policies or practices.

<table>
<thead>
<tr>
<th>How important is the competency?</th>
<th>Not at All</th>
<th>Not Very</th>
<th>Somewhat</th>
<th>Very</th>
</tr>
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<tbody>
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<tr>
<th>How confident are you in demonstrating this competency?</th>
<th>Not at All</th>
<th>Not Very</th>
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<th>Very</th>
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<tr>
<th>How frequently do you apply this competency?</th>
<th>Never</th>
<th>1-2 Per Year</th>
<th>Monthly</th>
<th>Weekly</th>
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<tbody>
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</tbody>
</table>
Part II: Importance of genetics/genomics to your institution’s leadership

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
Part III: Your role in Public Health

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?

<table>
<thead>
<tr>
<th>Role</th>
<th>&lt; 25%</th>
<th>25-50%</th>
<th>50-75%</th>
<th>&gt; 75%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Administrative</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Program Planning</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Direct Consumer Care</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Policy/Legislative</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Research</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Assessment/Evaluation</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Education/Training</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
</tbody>
</table>

Opt out of survey

If you experience any technical difficulties, please contact the survey administrator at User-Centered Design at providersurvey@user-centereddesign.com

Part III (cont'd)

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).

Opt out of survey

If you experience any technical difficulties, please contact the survey administrator at User-Centered Design at providersurvey@user-centereddesign.com
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. The overall reliability for the survey instrument is excellent. 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. Public Health Providers’ Survey Tables and Summary Data

### Table 1. Perception of the Importance of the Competencies

<table>
<thead>
<tr>
<th>Competency</th>
<th>Question</th>
<th>0</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>Response Rate</th>
<th>Mean</th>
</tr>
</thead>
<tbody>
<tr>
<td>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.</td>
<td>How important is the competency?</td>
<td>9</td>
<td>0</td>
<td>4</td>
<td>35</td>
<td>92</td>
<td>94 percent</td>
<td>3.7</td>
</tr>
<tr>
<td></td>
<td>How confident are you in demonstrating the competency?</td>
<td>9</td>
<td>3</td>
<td>21</td>
<td>61</td>
<td>46</td>
<td>94 percent</td>
<td>3.1</td>
</tr>
<tr>
<td></td>
<td>How frequently do you apply this competency?</td>
<td>10</td>
<td>6</td>
<td>25</td>
<td>32</td>
<td>67</td>
<td>93 percent</td>
<td>3.2</td>
</tr>
<tr>
<td>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.</td>
<td>How important is the competency?</td>
<td>10</td>
<td>0</td>
<td>2</td>
<td>17</td>
<td>11</td>
<td>93 percent</td>
<td>3.8</td>
</tr>
<tr>
<td></td>
<td>How confident are you in demonstrating the competency?</td>
<td>13</td>
<td>1</td>
<td>17</td>
<td>43</td>
<td>66</td>
<td>91 percent</td>
<td>3.4</td>
</tr>
<tr>
<td></td>
<td>How frequently do you apply this competency?</td>
<td>12</td>
<td>6</td>
<td>20</td>
<td>35</td>
<td>67</td>
<td>91 percent</td>
<td>3.3</td>
</tr>
<tr>
<td>3 Describe the importance of family history in assessing predisposition to disease.</td>
<td>How important is the competency?</td>
<td>12</td>
<td>1</td>
<td>1</td>
<td>22</td>
<td>10</td>
<td>91 percent</td>
<td>3.8</td>
</tr>
<tr>
<td></td>
<td>How confident are you in demonstrating the competency?</td>
<td>11</td>
<td>5</td>
<td>11</td>
<td>39</td>
<td>74</td>
<td>92 percent</td>
<td>3.4</td>
</tr>
<tr>
<td></td>
<td>How frequently do you apply this competency?</td>
<td>12</td>
<td>11</td>
<td>27</td>
<td>43</td>
<td>47</td>
<td>91 percent</td>
<td>3.0</td>
</tr>
<tr>
<td>4 Identify opportunities and integrate genetic/genomic issues into public health practice, policies or programs effectively.</td>
<td>How important is the competency?</td>
<td>10</td>
<td>0</td>
<td>1</td>
<td>28</td>
<td>10</td>
<td>93 percent</td>
<td>3.8</td>
</tr>
<tr>
<td></td>
<td>How confident are you in demonstrating the competency?</td>
<td>11</td>
<td>2</td>
<td>23</td>
<td>56</td>
<td>48</td>
<td>92 percent</td>
<td>3.2</td>
</tr>
<tr>
<td></td>
<td>How frequently do you apply this competency?</td>
<td>12</td>
<td>8</td>
<td>37</td>
<td>36</td>
<td>47</td>
<td>91 percent</td>
<td>3.0</td>
</tr>
<tr>
<td>5 Maintain up-to-date knowledge of genetics and genomics-related policies, legislation, statutes, and regulations.</td>
<td>How important is the competency?</td>
<td>14</td>
<td>0</td>
<td>4</td>
<td>42</td>
<td>80</td>
<td>90 percent</td>
<td>3.6</td>
</tr>
<tr>
<td></td>
<td>How confident are you in demonstrating the competency?</td>
<td>15</td>
<td>4</td>
<td>34</td>
<td>55</td>
<td>32</td>
<td>89 percent</td>
<td>2.9</td>
</tr>
<tr>
<td></td>
<td>How frequently do you apply this competency?</td>
<td>15</td>
<td>13</td>
<td>43</td>
<td>44</td>
<td>25</td>
<td>89 percent</td>
<td>2.6</td>
</tr>
<tr>
<td>6 Describe the potential physical and psychological benefits, limitations, and risks of genetic/genomic information for individuals, family members, and communities.</td>
<td>How important is the competency?</td>
<td>14</td>
<td>0</td>
<td>2</td>
<td>33</td>
<td>91</td>
<td>90 percent</td>
<td>3.7</td>
</tr>
<tr>
<td></td>
<td>How confident are you in demonstrating the competency?</td>
<td>15</td>
<td>2</td>
<td>20</td>
<td>49</td>
<td>54</td>
<td>89 percent</td>
<td>3.2</td>
</tr>
<tr>
<td></td>
<td>How frequently do you apply this competency?</td>
<td>18</td>
<td>10</td>
<td>39</td>
<td>32</td>
<td>41</td>
<td>87 percent</td>
<td>2.9</td>
</tr>
<tr>
<td></td>
<td>How important is the competency?</td>
<td>How confident are you in demonstrating the competency?</td>
<td>How frequently do you apply this competency?</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>---</td>
<td>---------------------------------</td>
<td>--------------------------------------------------------</td>
<td>---------------------------------------------</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>7</td>
<td>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.</td>
<td>14 1 1 27 97</td>
<td>90 percent 3.7</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>16 6 26 54 38</td>
<td>89 percent 3.0</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>15 13 46 34 32</td>
<td>89 percent 2.7</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>8</td>
<td>Identify the resources available to assist clients seeking genetic/genomic information or services, including the types of genetics professionals available.</td>
<td>13 0 6 25 96</td>
<td>91 percent 3.7</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>14 7 29 37 53</td>
<td>90 percent 3.1</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>18 24 34 30 34</td>
<td>87 percent 2.6</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>9</td>
<td>Conduct outcomes evaluation of available genetic/genomic programs and services to determine their effectiveness.</td>
<td>16 1 8 34 81</td>
<td>89 percent 3.6</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>16 19 29 49 27</td>
<td>89 percent 2.7</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>16 44 48 19 13</td>
<td>89 percent 2.0</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>10</td>
<td>Identify the political, legal, social, ethical, and economic issues associated with integrating genomics into public health.</td>
<td>16 0 4 26 94</td>
<td>89 percent 3.7</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>17 9 23 44 47</td>
<td>88 percent 3.0</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>18 17 41 33 31</td>
<td>87 percent 2.6</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>11</td>
<td>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.</td>
<td>16 1 4 38 81</td>
<td>89 percent 3.6</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>16 11 25 54 34</td>
<td>89 percent 2.9</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>17 22 36 30 35</td>
<td>88 percent 2.6</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>12</td>
<td>Identify appropriate and relevant genetics research findings that can be translated into public health policies or practices.</td>
<td>16 0 5 24 95</td>
<td>89 percent 3.7</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>17 5 22 51 45</td>
<td>88 percent 3.1</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>19 12 47 33 29</td>
<td>86 percent 2.7</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Table 2. Importance of Genetics and Genomics to Job Responsibility

<table>
<thead>
<tr>
<th>No Answer</th>
<th>Not at all important</th>
<th>Of little importance</th>
<th>Somewhat important</th>
<th>Important</th>
<th>Very important</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
</tr>
</tbody>
</table>

Does your senior administration think that genetics and genomics is important to your job responsibilities? (123 total responses; 88 percent)

|           | 0 | 6 | 20 | 24 | 22 | 51 |

Does your senior administration think that genetics and genomics is important to their job responsibilities? (121 total responses; 86 percent)

|           | 19 | 10 | 32 | 37 | 15 | 27 |

Table 3. Adequacy of Resources to Implement Genetic and Genomic Competencies

<table>
<thead>
<tr>
<th>No Answer</th>
<th>Not at all adequate</th>
<th>Somewhat adequate</th>
<th>Adequate</th>
<th>Very adequate</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>3</td>
</tr>
</tbody>
</table>

How adequate are your resources for implementing genetic/genomic competencies into your work/role? (123 total responses; 88 percent)

|           | 17 | 28 | 51 | 27 | 17 |

Table 4. Level of Job In Public Health Industry

<table>
<thead>
<tr>
<th>Level</th>
<th>Number</th>
<th>Percent Total Responding</th>
</tr>
</thead>
<tbody>
<tr>
<td>Federal</td>
<td>16</td>
<td>13 percent</td>
</tr>
<tr>
<td>State</td>
<td>51</td>
<td>41 percent</td>
</tr>
<tr>
<td>Local</td>
<td>0</td>
<td>0 percent</td>
</tr>
<tr>
<td>Academic</td>
<td>38</td>
<td>30 percent</td>
</tr>
<tr>
<td>Private, nonprofit organization</td>
<td>11</td>
<td>9 percent</td>
</tr>
<tr>
<td>Community-based organization</td>
<td>5</td>
<td>4 percent</td>
</tr>
<tr>
<td>International</td>
<td>1</td>
<td>1 percent</td>
</tr>
<tr>
<td>Other (commercial laboratory, medical center community programming, nonprofit health organization)</td>
<td>3</td>
<td>2 percent</td>
</tr>
<tr>
<td>No answer</td>
<td>15</td>
<td>n/a</td>
</tr>
<tr>
<td>Total</td>
<td>140</td>
<td>n/a</td>
</tr>
</tbody>
</table>

5. Summary Responses to Questions:

1. Please describe any efforts that you or your organization has undertaken to ensure that genetic services or information are available for vulnerable or underserved populations. Are there particular strategies you would recommend?

- **Educational Materials**: Organizations are involved in either creating new or updating existing educational materials that are culturally and linguistically competent. These educational materials are available in different languages and are disseminated to vulnerable and underserved populations.

- **Community Involvement**: A majority of organizations focus on the principles of community-based participatory research and involve vulnerable or underserved communities in developing, planning,
and evaluating resources and materials. Furthermore, these organizations have involved these communities in the dissemination of resources and materials.

- **Training and Education:** Organizations are actively involved in training and educating local public health providers, undergraduate and graduate students, teachers, lay health advisors, and parents within their communities to foster outreach and community education. Training includes a wide array of strategies including curricula development for high school science teachers, training in genetic epidemiology targeting students, collaboration with local health departments to train and educate health professionals, and funding research and outreach efforts involving genetic services and community engagement via presentations at health fairs and conferences. Most training and education sessions involve topics such as communicating risk, genetic literacy levels, and how to target health messages to different audiences, particularly vulnerable and/or underserved populations.

- **Genetic Services:** Respondents indicated that some organizations are involved in engaging communities through the provision of genetic services, particularly genetic counseling, either through in-person sessions or teleconference calls. Another important type of genetic service is provision of genetic testing or provision of monetary assistance to organizations that provide genetic testing to all populations including vulnerable and/or underserved populations.

- **Research:** Some organizations are involved in funding and conducting research to understand barriers to genetic services and community involvement in accessing these services. Appropriate measures and efforts are undertaken to overcome and resolve those barriers in order to better facilitate and engage vulnerable and/or underserved populations.

- **Funding:** Most respondents viewed the limited availability of funds as a potential barrier to outreach and community engagement involving vulnerable and/or underserved populations. They recommended that funding should be increased and appropriately allotted to enhance genetic services, outreach, and partnerships with vulnerable or underserved populations.

- **Websites:** Some respondents recommended that websites should be a part of outreach tools that can be easily accessed by clients. Materials should be readily available to view or download.

- **Policy:** Other respondents recommended that federal policy needs to facilitate state and local policy by involving legislators, local community leaders, and community members to enhance genetic services, raise awareness, and increase education of local community members about their efforts within the vulnerable and/or underserved communities.

2. The survey closed with an opportunity for individuals to provide additional comments to SACHGS on the topic of genetics and genetics education for public health providers. Fifty-four responses were received, describing the following themes:

- **Funding:** Funding should be provided to develop and implement genetic curricula and training programs, integrate genetics education into public health programs such as newborn screening, develop ready-to-use tools and resources for local organizations and communities, and provide genetic services to all affected families within a community.

- **Networking and Collaboration:** It is vital that organizations within and across states are encouraged to share ideas and information concerning the success of programmatic and outreach efforts. The lack of networking and collaboration across local, state, and federal level leads to reinvention of programs.

- **Best Evidence-Based Practices:** Public health providers should be trained and educated to identify the best practices of genetics and genetics education and incorporate these practices into their services and programs.

- **Education:** Some respondents reported that they do not view genetics and/or genetics education as an important facet of their profession. Others felt that it is very important and should be integrated into their training. Recommended educational topics should include population-level epidemiology, review of widely publicized research findings, understanding the concept of risk associated with single nucleotide polymorphisms, clinical validity and utility, analysis of family health histories, and
the ELSI issues. In addition, many respondents recommended that basic education about genetics, genomics, and its related competencies should be provided to public health providers that include but are not limited to nurse practitioners, midwives, primary health care centers, outpatient clinics, nutritionists, physicians and childbirth educators.
Appendix D: 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., R.N., 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., C.G.C., 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)

General Interview Guide Theme Areas

- 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

Table 1. Key Findings from Semi-Structured Interviews

<table>
<thead>
<tr>
<th>Perceptions about consumers’ understanding of genetics and genomics</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Segments of the general public are struggling to stay abreast of rapidly advancing genetic technologies and the potential benefits and risks of these technologies.</td>
</tr>
<tr>
<td>• 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.</td>
</tr>
<tr>
<td>• Segments of the public have a common misconception that genetic predisposition is deterministic.</td>
</tr>
<tr>
<td>• Segments of the public do not understand complex traits and that there are multiple risk factors for a single health condition.</td>
</tr>
</tbody>
</table>
### Challenges consumers face in obtaining information about genetics and genomics

- 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.

### Where people get information

- From a variety of sources including the news, television, Internet, local and religious communities.

### Successful and suggested models for genetics education

- 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.

### The role of government in activities related to genetics education of the public

- 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.
2. Consumers’ Survey Instrument

SACGHS Draft Report on Genetics Education and Training 5-19-2010

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

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
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.)
   - 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: [ ]

If you experience any technical difficulties, please contact the survey administrator at User-Centered Design at consumersurvey@user-centereddesign.com
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: __________________________
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.)

<table>
<thead>
<tr>
<th>Federal Government (rank 1-6)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Funding genetics education programs</td>
</tr>
<tr>
<td>Education about the regulation of genetic services</td>
</tr>
<tr>
<td>Education about the regulation of genetic tests</td>
</tr>
<tr>
<td>Education about the licensing of genetic health care providers</td>
</tr>
<tr>
<td>Education about genetic anti-discrimination laws</td>
</tr>
<tr>
<td>Serving as a clearinghouse of educational information</td>
</tr>
</tbody>
</table>

If there are more important items not listed above, please specify: ____________________________

Opt out of survey

If you experience any technical difficulties, please contact the survey administrator at User-Centered Design at mcsumer.survey@user-centered-design.com

---

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.)

<table>
<thead>
<tr>
<th>State Government (rank 1-6)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Funding genetics education programs</td>
</tr>
<tr>
<td>Education about the regulation of genetic services</td>
</tr>
<tr>
<td>Education about the regulation of genetic tests</td>
</tr>
<tr>
<td>Education about the licensing of genetic health care providers</td>
</tr>
<tr>
<td>Education about genetic anti-discrimination laws</td>
</tr>
<tr>
<td>Serving as a clearinghouse of educational information</td>
</tr>
</tbody>
</table>

If there are more important items not listed above, please specify: ____________________________

Opt out of survey

If you experience any technical difficulties, please contact the survey administrator at User-Centered Design at mcsumer.survey@user-centered-design.com
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.)

<table>
<thead>
<tr>
<th>Role</th>
<th>Local Government (rank 1-6)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Funding genetics education programs</td>
<td></td>
</tr>
<tr>
<td>Education about the regulation of genetic services</td>
<td></td>
</tr>
<tr>
<td>Education about the regulation of genetic tests</td>
<td></td>
</tr>
<tr>
<td>Education about the licensing of genetic health care providers</td>
<td></td>
</tr>
<tr>
<td>Education about genetic anti-discrimination laws</td>
<td></td>
</tr>
<tr>
<td>Serving as a clearinghouse of educational information</td>
<td></td>
</tr>
</tbody>
</table>

If there are more important items not listed above, please specify: [ ]
8. 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**

- [ ] 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 8? (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): 

If you experience any technical difficulties, please contact the survey administrator at User-Centered Design at consumers.survey@user-centereddesign.com

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)
The following questions ask for general demographic information about you and your work.

9. In what state do you work?
   -- Select --

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

---

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).

---
3. Consumer Survey Figures and Tables

Figure 1. Geographic Distribution of Responses. Respondents were asked “In which state do you work?” Responses were received from 258 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).
Figure 2. Distribution of Organization Types.

Table 2. Importance of Genetics to Organizational Mission

<table>
<thead>
<tr>
<th>Importance</th>
<th>#</th>
<th>percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Extremely important</td>
<td>126</td>
<td>37</td>
</tr>
<tr>
<td>Important</td>
<td>75</td>
<td>22</td>
</tr>
<tr>
<td>Somewhat important</td>
<td>44</td>
<td>13</td>
</tr>
<tr>
<td>Not very important</td>
<td>19</td>
<td>6</td>
</tr>
<tr>
<td>Not at all important</td>
<td>5</td>
<td>1</td>
</tr>
<tr>
<td>No response</td>
<td>68</td>
<td>20</td>
</tr>
</tbody>
</table>

Table 3. Concepts for Informed Seekers of Genetic Information

<table>
<thead>
<tr>
<th>Rank</th>
<th>Concepts</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Family history is an important tool for understanding your health and disease</td>
</tr>
<tr>
<td>2</td>
<td>Basic genetic and genomic concepts and terminology (e.g., inheritance, gene, genome)</td>
</tr>
<tr>
<td>2</td>
<td>Common diseases are caused by complex genetic and environmental factors</td>
</tr>
<tr>
<td>2</td>
<td>Genetics is relevant to everyone's health</td>
</tr>
<tr>
<td>5</td>
<td>Understanding an individual's genetic makeup by itself will not solve all health problems</td>
</tr>
</tbody>
</table>

Table 4. Topics of Special Relevance for Informed Seekers of Genetic Information

<table>
<thead>
<tr>
<th>Rank</th>
<th>Topics</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Where to find reliable genetic and genomic information</td>
</tr>
<tr>
<td>2</td>
<td>How to access genetic tests</td>
</tr>
<tr>
<td>2</td>
<td>How to interpret results of a genetic test</td>
</tr>
<tr>
<td>2</td>
<td>How to interpret and evaluate the credentials of a genetics professional</td>
</tr>
</tbody>
</table>
### Table 5. Genetic Education and Services Needs of Underserved and Vulnerable Populations

<table>
<thead>
<tr>
<th>Rank</th>
<th>Educational service needs</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Basic and relevant genetic health information</td>
</tr>
<tr>
<td>2</td>
<td>Skills to make informed health decisions</td>
</tr>
<tr>
<td>3</td>
<td>Culturally appropriate genetic health information</td>
</tr>
<tr>
<td>4</td>
<td>Education about access to genetic services</td>
</tr>
</tbody>
</table>

### Table 6. Barriers Preventing Education in Genetics and Genomics

<table>
<thead>
<tr>
<th>Rank</th>
<th>Barriers</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Lack of health professionals' understanding of genetics</td>
</tr>
<tr>
<td>1</td>
<td>Lack of individual health literacy in genetics</td>
</tr>
<tr>
<td></td>
<td>Lack of patient understanding of genetic testing implications for</td>
</tr>
<tr>
<td></td>
<td>themselves or their family (i.e., whether to share results with family</td>
</tr>
<tr>
<td></td>
<td>members)</td>
</tr>
<tr>
<td>4</td>
<td>Lack of access to genetic services for consumers/patients</td>
</tr>
<tr>
<td>5</td>
<td>Direct-to-consumer marketing of genetic tests before there is evidence of</td>
</tr>
<tr>
<td></td>
<td>their utility or benefit</td>
</tr>
</tbody>
</table>

### Table 7. Roles for Governments in Public Education in Genetics and Genomics

#### Federal government (82 percent response rate)

<table>
<thead>
<tr>
<th>Rank</th>
<th>Barriers</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Funding genetics education programs</td>
</tr>
<tr>
<td>2</td>
<td>Serving as a clearinghouse of educational information</td>
</tr>
<tr>
<td>3</td>
<td>Education about genetic anti-discrimination laws</td>
</tr>
<tr>
<td>4</td>
<td>Education about the regulation of genetic tests</td>
</tr>
<tr>
<td>5</td>
<td>Education about the regulation of genetic services</td>
</tr>
<tr>
<td>6</td>
<td>Education about the licensing of genetic health care providers</td>
</tr>
</tbody>
</table>

#### State governments (74 percent response rate)

<table>
<thead>
<tr>
<th>Rank</th>
<th>Barriers</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Funding genetics education programs</td>
</tr>
<tr>
<td>2</td>
<td>Education about genetic anti-discrimination laws</td>
</tr>
<tr>
<td>3</td>
<td>Education about the regulation of genetic services</td>
</tr>
<tr>
<td>4</td>
<td>Serving as a clearinghouse of educational information</td>
</tr>
<tr>
<td>4</td>
<td>Education about the regulation of genetic tests</td>
</tr>
<tr>
<td>4</td>
<td>Education about the licensing of genetic health care providers</td>
</tr>
</tbody>
</table>

#### Local governments (65 percent response rate)

<table>
<thead>
<tr>
<th>Rank</th>
<th>Barriers</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
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</tr>
<tr>
<td>4</td>
<td>Education about the regulation of genetic tests</td>
</tr>
<tr>
<td>5</td>
<td>Serving as a clearinghouse of educational information</td>
</tr>
<tr>
<td>5</td>
<td>Education about the licensing of genetic health care providers</td>
</tr>
</tbody>
</table>
Appendix E: SACGHS Surveys of Federal Agency Activities 2003-2009

Additional Programs and Activities

**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](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](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**
- Pharmacogenomic Screening: All service members undergo G6PD testing, sickle cell screening, and color vision screening, with subsequent environmental and pharmacologic management designed to prevent disease.
- Newborn Screening 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.
- Fellowships: 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.

**DOE**
- 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).

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 Neurodevelopmental 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/](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 access 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**

- **Trans-NIH projects are administered by the Office of Strategic Operations through the National Institute on Drug Abuse, the National Institute of Research Resources, the National Institute on Mental Health, the National Institute of General Medical Sciences, and the National Institute of Diabetes and Digestive and Kidney Disorders. These Common Fund programs include:**
  - Clinical Center (CC) Grand Rounds devoted to genetics and genomics
  - A certificate program in Integrative Biomedical Informatics
  - Development of a curriculum to foster a basic understanding of the correlations between genetic and molecular findings and systems biology, health and disease
  - A post-doctoral program in neuro-developmental toxicology that includes a gene-environmental interaction component
  - A training program in models and technologies for defining phenotypes
  - Post-doctoral training in biobehavioral interventions in developmental disabilities
  - Training programs in pharmacoinformatics
  - Training program in genetics and complex diseases

- **Genetics education and training programs at individual institutes include programs at the National Cancer Institute, National Human Genome Research Institute, National Institute on Deafness and Other Communication Disorders, National Institute of Dental and Craniofacial Research, National Institute on Drug Abuse, National Institute on Aging, National Center for Biotechnology Information, and the National Library of Medicine.**

  - **NCI Programs:**
    - 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](http://www.cancer.gov/cancertopics/prevention-genetics-causes/genetics)).
• NHGRI Programs:
  o Educational materials: educational web casts and interactive web-based learning tools were developed that fulfill recently adopted nursing competencies in genetics education.
  o 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.

• NIDCD Summer Program in Genetics for Audiology Faculty included:
  o A needs assessment survey of existing graduate level training programs in audiology that incorporate genetics into their curriculum
  o The establishment of an Advisory Board to guide development of an educational program in genetics
  o 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
  o The establishment of a comprehensive evaluation component to determine the effectiveness of the educational program

• NIDCR Programs:
  o 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.
  o 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.
  o Publications resulting from NIDCR genetics/genomic educational activities include:

• NIDA scientific meeting support included:
  o Travel fellowships to the Jacksonville Short Course in Medical and Experimental Genetics
  o 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.
  o Participation at the Community Anti-Drug Coalitions of America mid-year training institute conferences.
**NLM Programs include:**

- **NCBI:** *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.

- **The NLM University-based Biomedical Informatics Research Training Programs.** Training grants are provided to universities nation wide, 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, 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).

**EEOC**

- **Trainings for professionals on genetic discrimination and about GINA, Title II** were presented at the following conferences or to the following organizations:
  - SACGHS (June 2005)
  - Annual EXCEL Conference for federal agency EEO and HR professionals and federal agency counsel (August 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)
  - 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)

**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.