



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

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**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.<sup>1</sup> 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 work<sup>2</sup> 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](mailto: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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<sup>1</sup> Secretary's Advisory Committee on Genetics, Health, and Society. SACGHS Documents, Reports, and Correspondence. See [http://oba.od.nih.gov/SACGHS/sacghs\\_documents.html](http://oba.od.nih.gov/SACGHS/sacghs_documents.html). Accessed on November 24, 2009.

<sup>2</sup> Secretary's Advisory Committee on Genetics, Health, and Society. (2007). Roundtable on Genetics Education and Training of Health Professionals Session, November 20, 2007. See [http://oba.od.nih.gov/SACGHS/sacghs\\_past\\_meeting\\_2007\\_nov\\_20.html](http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_2007_nov_20.html). Accessed on January 14, 2010.

## 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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1 **Preface**

2  
3 The Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS or Committee) has  
4 consistently recognized the importance of professional and public genetics education and training.  
5 Recommendations to improve education and genetics and genomics literacy have been included in nearly  
6 every SACGHS report issued to date.<sup>3</sup> In 2004, the Committee issued a formal resolution that was  
7 conveyed to the Secretary of Health and Human Services regarding the critical importance of appropriate  
8 and adequate training and education in genetics and genomics for all health care professionals and the  
9 public.<sup>4</sup> In its 2004 and 2008 priority-setting processes, SACGHS ranked professional and public  
10 education as a high priority.<sup>5,6</sup> In November 2007, SACGHS convened a roundtable to identify the need  
11 for a task force on genetics education and training to build on the Committee’s earlier work.<sup>7</sup> The focus  
12 and scope of the task force were discussed at three subsequent SACGHS meetings.<sup>8,9,10</sup>

13  
14 The Genetics Education and Training Task Force was formed, and in consultation with the full  
15 Committee, defined its scope and developed a work plan and framework for analysis to identify education  
16 and training issues pertinent to (1) point-of-care health professionals with and without expertise in  
17 genetics (e.g., primary care professionals such as pediatricians, obstetrician/gynecologists, and internists,  
18 nurses, physician assistants, genetic counselors, and pharmacists), (2) public health providers involved or  
19 likely to be involved in providing genetic services, and (3) consumers and patients. With regard to  
20 consumers and patients, the Task Force focused on identifying their education needs to assist them in  
21 informed decisionmaking about the use of genetic and genomic services and to enhance their  
22 understanding and use of genetic information with regard to risk identification and management,  
23 prevention, diagnosis, and treatment of disease.

24  
25 To conduct its work, the Task Force divided into three workgroups to explore the education needs of  
26 these three broad communities (health care professionals, public health providers, and consumers and  
27 patients). With the rapid proliferation of genetic technologies and the shift toward personalized health  
28 care, the Task Force felt that focusing on the education and training needs of health care professionals  
29 working on the front lines of public health and health care delivery is of the highest priority, as is  
30 recognizing the need for an informed public. Two methods were used to gather information to inform this  
31 report: (1) a literature review of research relevant to professional and public education and training in  
32 genetics and genomics; and (2) surveys of select major organizations, groups, and individuals with

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<sup>3</sup> Secretary’s Advisory Committee on Genetics, Health, and Society. SACGHS Documents, Reports, and Correspondence. See [http://oba.od.nih.gov/SACGHS/sacghs\\_documents.html](http://oba.od.nih.gov/SACGHS/sacghs_documents.html). Accessed on November 24, 2009.

<sup>4</sup> Secretary’s Advisory Committee on Genetics, Health, and Society. (2004). Resolution of the Secretary’s Advisory Committee on Genetics, Health, and Society on Genetics Education and Training of Health Professionals. See [http://oba.od.nih.gov/SACGHS/sacghs\\_documents.html](http://oba.od.nih.gov/SACGHS/sacghs_documents.html). Accessed on February 24, 2010.

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

<sup>6</sup> Secretary’s Advisory Committee on Genetics, Health, and Society. (2004). *A Roadmap for the Integration of Genetics and genomics into Health and Society: The Study Priorities of the Secretary’s Advisory Committee on Genetics, Health, and Society*. See <http://oba.od.nih.gov/oba/sacghs/reports/SACGHSPriorities.pdf>. Accessed on November 24, 2009.

<sup>7</sup> Secretary’s Advisory Committee on Genetics, Health, and Society. (2007). Roundtable on Genetics Education and Training of Health Professionals, November 20, 2007. See [http://oba.od.nih.gov/SACGHS/sacghs\\_past\\_meeting\\_2007\\_nov\\_20.html](http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_2007_nov_20.html). Accessed on January 14, 2010.

<sup>8</sup> Secretary’s Advisory Committee on Genetics, Health, and Society. (2008). Agenda of the fifteenth meeting, February 13, 2008. See [http://oba.od.nih.gov/SACGHS/sacghs\\_past\\_meeting\\_documents.html#feb2008](http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_documents.html#feb2008). Accessed on November 24, 2009.

<sup>9</sup> Secretary’s Advisory Committee on Genetics, Health, and Society. (2008). Agenda of the sixteenth meeting, July 16, 2008. See [http://oba.od.nih.gov/SACGHS/sacghs\\_past\\_meeting\\_documents.html#jul2008](http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_documents.html#jul2008). Accessed on November 24, 2009.

<sup>10</sup> Secretary’s Advisory Committee on Genetics, Health, and Society. (2008). Agenda of the seventeenth meeting, December 17, 2008. See [http://oba.od.nih.gov/SACGHS/sacghs\\_past\\_meeting\\_documents.html#dec2008](http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_documents.html#dec2008). Accessed on November 24, 2009.

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33 responsibilities across the continuum of health professional education and public health, and those that  
34 provide advocacy for consumers and patients  
35

36 In focusing on these three groups, the Task Force and SACGHS acknowledge the importance of a wide  
37 range of professionals who are experiencing increasing exposure to genetics and genomics or play a role  
38 in assuring the proper application of genomic information and technologies to promote health. The  
39 Committee's future plans call for an assessment of whether this report's findings and recommendations  
40 may also apply to a broader constituency, such as specialty health care professionals, laboratory workers,  
41 health care administrators, payers, policymakers, and lay health providers as well as librarians, judges,  
42 law enforcement agents, clergy, science educators, journalists, policy makers, and health care governing  
43 bodies.  
44

45 The Committee acknowledges that there are other pertinent issues beyond education and training that  
46 influence the use of genetic and genomic technologies to improve the public's health. As the clinical  
47 utility of genetic tests and services is demonstrated over time, health care professionals will be more  
48 likely to see the need to incorporate genetics and genomics into their practice. Coverage and  
49 reimbursement of genetic tests and services, such as family history collection, influences the use of such  
50 services in clinical practice and thus may be an important priority for policymakers. In addition, new  
51 genomic technologies have the potential to decrease health disparities, but continuing work is needed to  
52 assure that appropriate access and utilization of genetic services are made available to underserved  
53 populations while not deflecting resources that address basic health care needs. These related issues are  
54 also discussed in this report.

55 **I. Background and Scope**  
56

57 The Human Genome Project was completed in 2003, resulting in a delineation of the complete sequence  
58 of the human genome. The sequence data helped advance research into the genetic basis of disease,  
59 including common, multifactorial diseases.<sup>11</sup> Expanded genetic and genomic knowledge and  
60 technologies are now leading to new approaches to the diagnosis of some common, chronic diseases and  
61 conditions.<sup>12</sup> Genomics also forms the basis of the growing field of pharmacogenomics, the study of how  
62 individual differences affect drug response.<sup>13</sup> These developments are moving genetics beyond a clinical  
63 specialty focused on rare, inherited diseases and chromosomal disorders, yet, many health care and public  
64 health professionals lack sufficient knowledge about the application and interpretation of genetics in the  
65 clinic or in the community.<sup>14</sup>  
66

67 Concerns have been raised for nearly four decades about how best to translate, interpret, and deliver  
68 complex genetic information to health care professionals and consumers. As the discipline of clinical  
69 genetics arose in the 1950s,<sup>15</sup> there was also recognition that nongenetics professionals would eventually  
70 also be needed to play a role in providing genetic services to patients.<sup>16</sup> In a 1975 report on the emerging  
71 field of genetic screening, the National Academy of Sciences (NAS) anticipated the movement of  
72 genetics from the specialized clinic toward point of care and signaled an early concern about the need for  
73 an educated workforce in the application of genetics.<sup>17</sup>  
74

75 Primary care has been the center of much of the focus on professional education needs in genetics. Thirty  
76 years ago Hsia, contemplating the transition of genetics into primary care, raised the following questions  
77 that remain today:  
78

79 “How much genetic knowledge should primary physicians have? Should they be able to  
80 diagnose, treat, and counsel about all genetic diseases? Will it suffice for them to check  
81 the literature or consult a geneticist whenever a genetic problem arises? Optimal  
82 knowledge must lie between these extremes, because a primary physician must have  
83 enough knowledge to recognize a problem as genetic and should have enough familiarity  
84 with genetic principles to be able to use the literature wisely, or to consult with a  
85 geneticist intelligently.”<sup>18</sup>  
86

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<sup>11</sup> 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.

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

<sup>13</sup> Pharmacogenomics is the study of how individual genetic differences affect drug response. See SACGHS’s May 2008 report, *Realizing the Potential of Pharmacogenomics: Opportunities and Challenges*. See [http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS\\_PGx\\_report.pdf](http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_PGx_report.pdf). Accessed on January 5, 2010.

<sup>14</sup> Shirts, B.H., and Parker, L.S. (2008). Changing interpretations, stable genes: responsibility of patients, professionals, and policy makers in the clinical interpretation of complex genetic information. *Genetics in Medicine*. 10(11):778-783.

<sup>15</sup> Greendale, K. and Pyeritz, R.E. (2001). Empowering primary care health professionals in medical genetics: How soon? How fast? How far? *American Journal of Medical Genetics*. 106:223-232.

<sup>16</sup> McNerney, J.D. Genetics education for health professionals: a context. (2008). *Journal of Genetic Counseling*. 17:145-151.

<sup>17</sup> National Academy of Sciences and National Research Council. *Genetic Screening: Programs, Principles and Research*. Washington: National Academy of Sciences, 1975.

<sup>18</sup> Hsia, Y.E., Bucholz, K.E., and Austein, C.A. Genetic knowledge of pediatricians and obstetricians (Connecticut, 1975, 1977): Implications for continuing education. In: Porter, I.H., and Hook, E.B. eds. *Service and Education in Medical Genetics*. Academic Press, 1979. P. 378.

87 Two decades later there was considerable debate regarding the role of nongenetics professionals in  
 88 genetic service provision. Greendale et al.<sup>19</sup> suggested potential problems with empowering primary care  
 89 providers to assume prominent roles in genetic service delivery, citing their lack of knowledge and  
 90 disinterest in the field, while Guttmacher et al.<sup>20</sup> argued that implementation of “genomic health care”  
 91 would necessitate collaboration and cooperation of all health professionals. Increasingly these same  
 92 concerns have turned toward public health providers, as genomics moves into population-based  
 93 applications, and toward the public, as consumers gain the ability to purchase their own genetic tests in  
 94 the marketplace. Recent legislative proposals, starting in 2007, have recognized needs in this area, calling  
 95 for increased funding of programs to develop and disseminate model training programs, ensure adequate  
 96 focus on genetics in certification and accreditation programs, enhance continuing education (CE)  
 97 programs, and promote competencies across clinical, public health, and laboratory disciplines.<sup>21</sup>  
 98 However, no bills have been passed that actually provide funding for such programs.  
 99

### 100 **Technological Advances**

101  
 102 In 2010, several trends are moving the use of genomic technologies into the clinic or encouraging the  
 103 public to access technologies via the marketplace. Thus, improved education at all levels is imperative.  
 104 For example, as a result of large-scale genome-wide association studies, data is being organized and  
 105 shared to translate research information into clinical knowledge. In 2003, the National Human Genome  
 106 Research Institute (NHGRI) launched a public research consortium named EnCODE, the Encyclopedia of  
 107 DNA Elements.<sup>22</sup> The goal of the project is to identify all functional elements in the human genome  
 108 sequence and to determine how genes interact so that preventive and therapeutic strategies can be  
 109 developed. Other relevant research initiatives are the 1000 Genomes Project, the Electronic Medical  
 110 Records and Genomics (eMERGE) Network, and the Cancer Genome Atlas. The 1000 Genomes Project  
 111 is an international effort launched in 2008 to establish a catalogue of human genetic variation based on  
 112 the sequences of at least 1,000 anonymous participants from a number of different ethnic groups.<sup>23</sup> The  
 113 eMERGE Network is a consortium formed to develop, disseminate, and apply approaches to research that  
 114 combine DNA repositories with electronic medical record systems for large-scale high-throughput genetic  
 115 research.<sup>24</sup> The Cancer Genome Atlas is focused on the genetic causes of human cancer.<sup>25</sup> All of these  
 116 efforts are accelerating our ability to make clinical sense of genomic data, and ultimately the contribution  
 117 of genetic variation to common, chronic diseases that burden our health care system.  
 118

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

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<sup>19</sup> Greendale, K., and Pyeritz, R.E. (2001). Empowering primary care health professionals in medical genetics: How soon? How fast? How far? *American Journal of Medical Genetics*. 106:223-232.

<sup>20</sup> Guttmacher, A.E., Jenkins, J., and Uhlmann, W.R. (2001). Genomic medicine: who will practice it? A call to open arms. *American Journal of Medical Genetics*. 106(3):216-222.

<sup>21</sup> See, for example, the Genomics and Personalized Medicine Act of 2007 (S.976). See [http://olpa.od.nih.gov/tracking/110/senate\\_bills/session1/s-976.asp](http://olpa.od.nih.gov/tracking/110/senate_bills/session1/s-976.asp). Accessed on February 25, 2010.

<sup>22</sup> National Human Genome Research Institute. *The ENCODE Project: ENCyclopedia Of DNA Elements*. See <http://www.genome.gov/10005107>. Accessed on November 27, 2009.

<sup>23</sup> 1000 Genomes Project. See <http://www.1000genomes.org>. Accessed on November 24, 2009.

<sup>24</sup> The electronic Medical Records and Genomics (eMERGE) Network. See <https://www.mc.vanderbilt.edu/victr/dcc/projects/acc/index.php/About>. Accessed September 2, 2009.

<sup>25</sup> National Cancer Institute. The Cancer Genome Atlas. See <http://cancergenome.nih.gov/>. Accessed on November 24, 2009.

<sup>26</sup> “Illumina Delivers First Genome Under Personal Genome-Sequencing Service” September 1, 2009. See <http://www.genomeweb.com/sequencing/illumina-delivers-first-genome-under-personal-genome-sequencing-service>. Accessed on November 24, 2009.

2009.<sup>27</sup> 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.<sup>28</sup> That dollar amount could attract some “customers,” who might equate the cost of whole-genome sequencing as comparable to other medical tests or procedures.<sup>29</sup> 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.<sup>30</sup> 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.”<sup>31</sup> 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.<sup>32</sup>

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.”<sup>33</sup>

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

<sup>27</sup> “Complete Genomics Gets Gene Sequencing Under \$5,000” November 5, 2009. See

<http://www.bloomberg.com/apps/news?pid=20601124&sid=aWutnyE4SoWw>. Accessed on November 24, 2009.

<sup>28</sup> National Institutes of Health, Office of Extramural Research. Revolutionary Genome Sequencing Technologies, RFA-HG-08-009. See <http://grants1.nih.gov/grants/guide/rfa-files/RFA-HG-08-009.html>. Accessed on November 24, 2009.

<sup>29</sup> Wolinsky, H. (2007). The thousand-dollar genome: Genetic brinksmanship or personalized medicine? *EMBO Reports*. 8(10):900-903.

<sup>30</sup> Mardis, E.R. (2006). Anticipating the \$1,000 genome. *Genome Biology*. 7(7):112.

<sup>31</sup> Guttmacher, A.E., and Collins, F.S. (2002). Genomic medicine: a primer. *New England Journal of Medicine*. 347(19):1512-1520.

<sup>32</sup> Khoury, M.J. (2003). Genetics and genomics in practice: the continuum from genetic disease to genetic information in health and disease. *Genetics in Medicine*. 5(4):261-268.

<sup>33</sup> Khoury, M.J., Gwinn, M., Burke, W., Bowen, S., and Zimmern, R. (2007). Will genomics widen or help heal the schism between medicine and public health? *American Journal of Preventive Medicine*. 33(4):310-317. P. 313.

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

164  
165 Advances in identifying the genetic underpinnings of chronic disease are rapidly changing the way we  
166 think about treating disease and promoting health.<sup>34</sup> Understanding genetic and genomic influences can  
167 affect treatment in a number of ways, for example, through development of targeted medications for  
168 specific genetic alterations in an individual that are associated with drug efficacy and/or toxicity  
169 (pharmacogenomics); altered needs for specific dietary constituents such as increased folic acid in the  
170 presence of mutations in methylenetetrahydrofolate reductase; and increased screening when the presence  
171 of specific mutations increase the risk of developing disease such as colorectal cancer. If advances in  
172 genetics and genomics are to be effectively applied to improve disease outcome and promote health in the  
173 population, research that yields new insights into the pathophysiology of disease must be followed by  
174 clinical applications that lead to improved outcomes. These outcomes cannot be achieved without a better  
175 educated health care workforce.

176  
177 The patient and consumer also will play an increasingly critical role in achieving the goals of genetic  
178 medicine. While there are gaps and barriers to successful integration of genomics into clinical practice  
179 (e.g., clinical utility, privacy, developing an evidence base, developing cost models),<sup>35</sup> the translation of  
180 research discoveries will result in health promotion only if the translation is successfully adopted into  
181 clinical practice and leads to individuals adopting health-promoting behaviors. For example, a person who  
182 is found to be at increased risk for developing type 2 diabetes, whether this knowledge comes from family  
183 history or identification of disease-contributing single nucleotide polymorphisms (SNPs), will not benefit  
184 from this information unless he or she is willing to make behavioral changes that minimizes risk.<sup>36,37</sup>  
185 Because knowledge is considered a prerequisite of health behavior, consumers and patients will need  
186 knowledge to benefit from advances in genetics and genomics. Additionally, much needed public  
187 participation in debates surrounding science and technology, which would include the use of genetic and  
188 genomic technologies and services, requires adequate knowledge and understanding.<sup>38</sup> The availability of  
189 genetic and genomic tests that consumers can purchase without the involvement of their health care  
190 provider adds urgency to public education efforts.

191  
192 **The Need for a New Model for Delivering Genetics and Genomics Information**

193  
194 With the increased integration of genetics and genomics into a broader health care network, consumers  
195 and patients will be using results of genetic technologies increasingly in their own health care  
196 decisionmaking. Patients and consumers, health care professionals, and public health officials are  
197 challenged to keep pace with this dynamic and rapidly evolving field. The emerging understanding of the  
198 role of genetics and genomics in common disease is increasing the need for knowledge and understanding  
199 of risk assessment, genetic diagnoses, appropriate treatment approaches, and communication in  
200 professional and public education. The accelerated growth of DTC genetic and genomic services  
201 highlights the importance of adequate education for consumers to ensure informed decisionmaking. To

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<sup>34</sup> Feero, W.G., Guttmacher, A.E., and Collins, F.S. (2008). The genome gets personal—almost. *Journal of the American Medical Association*. 299(11):1351-1352.

<sup>35</sup> Scheuner, M.T., Sieverding, P., and Shekelle, P.G. (2008). Delivery of genomic medicine for common chronic adult diseases. *Journal of the American Medical Association*. 299(11):1320-1334.

<sup>36</sup> Sanderson, S.C., Wardle, J., and Michie, S. (2005). The effects of a genetic information leaflet on public attitudes towards genetic testing. *Public Understanding of Science*. 14:213-224.

<sup>37</sup> Zlot, A.I., Bland, M.P., Silvey, K., Epstein, B., Mielke, B., and Leman, R.F. (2009). Influence of family history of diabetes on health care provider practice and patient behavior among nondiabetic Oregonians. *Preventing Chronic Disease Public Health Research, Practice and Policy*. 6(1):1-11.

<sup>38</sup> Etchegary, H., Cappelli, M., Potter, B., Vloet, M., Graham, I., Walker, M., and Wilson, B. (2010). Attitude and knowledge about genetics and genetic testing. *Public Health Genomics*. 13:80-88.

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

204  
205 Improving and expanding genetics education for health care professionals, public health providers, and  
206 consumers will require a comprehensive and coordinated effort. Genomics will challenge the traditional  
207 model of genetic services, in which the use and communication of genetic information occurs in the  
208 clinical setting, during “teachable moments.”<sup>39</sup> This approach, while continuing to be essential at the  
209 individual health level, will not address the much larger fraction of the population with moderately  
210 increased risk for various multifactorial diseases with genetic components (e.g., cancer, cardiovascular  
211 disease, and diabetes). Effective interventions based on genetic information will rely on the public’s  
212 understanding of the meaning and interactions of susceptibility genes of uncertain penetrance with other  
213 risk factors.<sup>40</sup> In addition, with the expansion of screening and early detection technologies for many  
214 common chronic diseases, the public health workforce will become increasingly integral to both  
215 community education and service provision. Moreover, expanded newborn screening increases the need  
216 for primary care providers to be educated about the critical nature of a positive result and emphasizes the  
217 need for just-in-time resources for referral and patient management. Parents and families also have  
218 educational needs related to newborn screening not only if their child has a positive screen and requires  
219 follow-up, but also as new issues emerge, such as increasing rates of false positives as more tests are  
220 added to the newborn screening panel and the need for parental consent related to the storage and use of  
221 residual blood spot specimens.

222  
223 Thus, a new model for applying genetics to improved health requires a system in which health care  
224 professionals, public health providers, and consumers are well informed and able to interact and connect  
225 with each other as appropriate. Cooperation and collaboration in processing, applying, and interpreting  
226 genetic information will be essential. Without educated health care professionals and consumers, society  
227 will not benefit from genetic advances. Without an educated public health workforce, opportunities will  
228 be lost for deploying prevention and early detection programs for a wide variety of chronic diseases. And,  
229 without an informed public, patients and consumers may make poorly informed choices, or fail to seek  
230 needed professional health services.

231  
232 **The Work of the Genetics Education and Training Task Force**

233  
234 To inform the education and training needs of health care professionals, public health providers, and the  
235 public, a literature search was conducted simultaneously in 10 databases via the DIALOG platform for the  
236 time period 2003-2009. The search included databases from the fields of medicine, science, education,  
237 social science, and psychology. It was limited to English-only articles and did not include meeting  
238 abstracts. Some unpublished literature was captured by searching the dissertations abstract database. (See  
239 Appendix A-1 for data bases searched and search terms used.) Additional literature was reviewed as it  
240 became available in 2010.

241  
242 In addition, SACGHS collected data from Federal agencies and selected organizations with  
243 responsibilities across the continuum of health professional education, public health, and consumer and  
244 patient advocacy to obtain information regarding their activities in genetics education. The results of  
245 surveys and interviews are provided in the following chapters.

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<sup>39</sup> Lawson, P.J., and Flocke, S.A. (2009). Teachable moments for health behavior change: a concept analysis. *Patient Education and Counseling*. 76:25-30.

<sup>40</sup> Wilde, A., Meiser, B., Mitchell, P.B., and Schofield, P.R. (2010). Public interest in predictive genetic testing, including direct-to-consumer testing, for susceptibility to major depression: preliminary findings. *European Journal of Human Genetics*. 18(1):47-51.

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

247

### 248 A. Introduction

249

250 Health care professionals, particularly those working at the point of care—physicians, nurses, physician  
 251 assistants, genetic counselors, and pharmacists—must be adequately educated and trained in genetics to  
 252 promote the effective translation of new genetic knowledge into practice, enhance access to genetic  
 253 technologies, and ensure that these technologies are appropriately used. Over the past several decades,  
 254 considerable research has examined levels of knowledge and understanding of genetics in a variety of  
 255 groups. These studies suggest that health professionals rate their knowledge of genetics as fair to poor;<sup>41</sup>  
 256 and that a large majority test poorly on knowledge and interpretation of genetic data.<sup>42,43</sup> Without  
 257 additional educational efforts, the educational gap will only grow as new applications in genetics and  
 258 genomics appear across the health care and public health landscape. Although some studies have shown  
 259 that health professionals’ understanding of genetics has improved over time, more recent research shows  
 260 that health care professionals still lack the knowledge needed to make optimal use of genetic information.  
 261

### 262 B. Literature Review

263

264 A significant body of literature from the United States and abroad highlights the nature and lack of  
 265 genetics education of health care professionals as factors limiting integration of genetics into health  
 266 care.<sup>44,45,46,47,48,49,50,51,52,53</sup> McInerney<sup>54</sup> summarized these contributing factors as:

267

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<sup>41</sup> Menasha, J.D., Schechter, C., and Williams, J. (2000). Genetic testing: a physician’s perspective. *The Mount Sinai Journal of Medicine*. 67(2):144-51.

<sup>42</sup> Hunter, A., Wright, P., Cappelli, M., Kasaboski, A., and Surh, L. (1998). Physician knowledge and attitudes toward molecular genetic (DNA) testing of their patients. *Clinical Genetics*. 53:447-55.

<sup>43</sup> Giardello, F.M., Brensinger, J.D., Peterson, G.M., Luce, M.C., Hylind, L.M., Bacon, J.A., Booker, S.V., Parker, R.D., and Hamilton, S.R. (1997). The use and interpretation of commercial APC gene testing for familial adenomatous polyposis. *New England Journal of Medicine*. 336:823-27.

<sup>44</sup> Guttmacher, A.E., Porteous, M.E., and McInerney, J.D. (2007). Educating health-care professionals about genetics and genomics. *Nature Reviews*. 8:151-157.

<sup>45</sup> Harris, J.N., Bowen, D.J., Kuniyuki, A., McIntosh, L., FitzGerald, L.M., Ostrander, E.A., and Stanford, J. L. (2009). Interest in genetic testing among affected men from hereditary prostate cancer families and their unaffected male relatives. *Genetics in Medicine*. 11(5):1-12.

<sup>46</sup> Benjamin, C.M., Anionwu, E.N., Kristoffersson, U., ten Kate, L.P., Plass, A.M., Nippert, I., Julian-Revnier, C., Harris, H.J., Schmidtke, J., Challen, K., Calefato, J.M., Waterman, C., Powell, E., and Harris, R., on behalf of the GenEd Research Group. (2009). Educational priorities and current involvement in genetic practice: a survey of midwives in the Netherlands, UK and Sweden. *Midwifery*. 25(5):483-499.

<sup>47</sup> Burke, S., Martyn, M., Thomas, H., and Farndon, P. (2009). The development of core learning outcomes relevant to clinical practice: identifying priority areas for genetics education for non-genetics specialist registrars. *Clinical Medicine*. 9:49-52.

<sup>48</sup> Burke, S., Martyn, M., Stone, A., Bennett, C., Thomas, H., and Farndon, P. (2009). Developing a curriculum statement based on clinical practice: genetics in primary care. *British Journal of General Practice*. 59:99-103.

<sup>49</sup> Little, J., Potter, B., Allanson, J., Caulfield, J., Carroll, J.D., and Wilson, B. (2009). Canada: public health genomics. *Public Health Genomics*. 12:112-120.

<sup>50</sup> Metcalfe, S.A., Bittles, A.H., O’Leary, P., and Emery, J. (2009). Australia: public health genomics. *Public Health Genomics*. 12:121-128.

<sup>51</sup> Pestka, E.L., and Williams, J.K. (2005). International collaboration on genomics education for nurses. *Journal of Continuing Education in Nursing*. 36(4):180-184.

<sup>52</sup> Tomatir, A.G., Sorkun, H.C., Demirhan, H., and Akdaq, B. (2007). Genetics and genetic counseling: practices and opinions of primary care physicians in Turkey. *Genetics in Medicine*. 9(2):130-135.

<sup>53</sup> Kiray, V.B., Tomatir, A.G., Kuzu, K.N., and Taspinar, A. (2009). Nursing students’ self-reported knowledge of genetics and genetic education. *Public Health Genomics*. 12(4):225-232.

<sup>54</sup> McInerney, J.D. (2008). Genetics education for health professionals: a context. *Journal of Genetic Counseling*. 17:145-151.

- 268 • *Crowded curricula.* All health-related disciplines face the challenge of including more  
269 information in a finite time.
- 270 • *Misconceptions about genetics.* Many health care providers still believe that genetic medicine is  
271 defined by rare, Mendelian disorders and circumscribed by pediatrics and obstetrics, when in fact  
272 genetics is increasingly concerned with the common, chronic diseases that are the daily focus for  
273 most health professionals.
- 274 • *Lack of knowledgeable faculty.* Many institutions that train health care professionals do not have  
275 sufficient faculty with genetic training to teach basic genetics or its applications to patient care.
- 276 • *A disconnect between the basic sciences and clinical experiences during training.* Even when  
277 students training for health-related careers receive genetics instruction during basic science  
278 training, their subsequent clinical training often fails to incorporate genetic perspectives, largely  
279 because those responsible for the clinical training do not themselves have substantive education  
280 and expertise in genetics.
- 281 • *Failure to integrate genetics across the curriculum.* Genetics instruction is poorly integrated into  
282 all relevant courses.
- 283 • *Inadequate representation of genetics on certifying exams.* Testing often drives curricula, and the  
284 certifying exams for most health professionals include little, if any, genetics content.
- 285 • *A dearth of genetics professionals.* The low numbers of medical geneticists and genetic  
286 counselors in the United States and elsewhere limit the provision of genetic services directly and,  
287 further, limit the extent to which other providers have formal and informal access to genetics  
288 expertise.
- 289 • *Lack of management and referral guidelines in genetics.* The paucity of evidence-based  
290 guidelines related to genetic medicine likely hinders the attention genetics receives from  
291 providers on a day-to-day basis and raises questions for providers about the clinical utility of  
292 genetics.
- 293 • *Lack of knowledge and confidence about genetics among primary care providers.* Surveys of  
294 health professionals demonstrate a lack of basic knowledge about genetics, and often a lack of  
295 confidence to deal with genetics-related issues that arise in the clinical setting.

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

### 304 **The Genetic Professional Workforce**

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

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<sup>55</sup> Harvey, E.K., Fogel, C.E., Peyrot, M., Christensen, K.D., Terry, S.F., and McInerney, J.D. (2007). Providers' knowledge of genetics: a survey of 5915 individuals and families with genetic conditions. *Genetics in Medicine*. 9(5):259-267.

<sup>56</sup> Bureau of Health Professions. (2008). *The Physician Workforce: Projections and Research into Current Issues Affecting Supply and Demand*. Health Resources Service Administration. See <http://bhpr.hrsa.gov/healthworkforce/>. Accessed on November 24, 2009.

<sup>57</sup> Health Resources Services Administration. (2004). *The Registered Nurse Population: Findings from the 2004 National Sample Survey of Registered Nurses*. See <http://bhpr.hrsa.gov/healthworkforce/pharmacy/>. Accessed on December 4, 2009.

<sup>58</sup> Bureau of Labor Statistics. *Occupational Outlook Handbook, 2008-2009*. See [http://www.bls.gov/oco/ocos081.htm#projections\\_data](http://www.bls.gov/oco/ocos081.htm#projections_data). Accessed on December 4, 2009.

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

318  
319 Setting aside the steady entry of genetics into routine care, these deficiencies become even more  
320 concerning in light of expanded newborn screening programs. It is recognized that clinicians generally are  
321 unprepared, and educational efforts that focus on screening procedures and referral practices will be  
322 critical to maximize this life-saving public health program.<sup>62</sup>

323  
324 The 2008 Newborn Screening Saves Lives Act (Pub. L. No. 110-204)<sup>63</sup> recognized and renewed the  
325 national commitment to newborn screening as a critical public health program that saves and improves  
326 children's lives. Expanded newborn screening programs are expected to detect 10,000 affected infants  
327 annually, with many needing chronic disease management. Yet, there are only 200 physicians specialized  
328 in the diagnosis and management of patients with inherited metabolic disease in the United States, and  
329 some of the conditions detected through newborn screening are so rare that only a handful of experts exist  
330 with experience in their management. Physicians who have such expertise are least able to expand  
331 services<sup>64</sup> and three quarters reported that their practices are "nearly full," with about one quarter  
332 reporting new patient appointment wait times of more than three months.

333  
334 Thus, although the need for clinical genetic services has increased, and continues to increase, the ability  
335 of the genetics-specific health care workforce—which includes medical geneticists, genetic counselors,  
336 and other health care workers such as nurses who provide genetic services—is not sufficient to meet this  
337 need. In a survey of graduate medical education conducted in 2008-2009, it was found that only 76  
338 residents were enrolled in a medical genetics subspecialty program.<sup>65</sup> This statistic suggests that the large  
339 and diverse group of health professionals providing services at the point of care must be enlisted to  
340 provide appropriate genetic services and information.

### 341 342 **The Critical Shortage of Medical Geneticists**

343  
344 In 2009, the American Board of Medical Genetics (ABMG) reported that over a 27-year period beginning  
345 in 1982, 2,511 individuals had achieved board certification in one or more of the ABMG certification

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<sup>59</sup> Health Resources and Services Administration. *The Adequacy of Pharmacist Supply: 2004 to 2030*. See <http://bhpr.hrsa.gov/healthworkforce/pharmacy/>. Accessed on December 4, 2009.

<sup>60</sup> Cooksey, J.A., Forte, G., Benkendorf, J., and Blitzer, M.G. (2005). The state of the medical geneticist workforce: findings of the 2003 survey of ABMG certified geneticists. *Genetics in Medicine*. 7(6):439-443.

<sup>61</sup> Cooksey, J.A., Forte, G., Flanagan, P., Benkendorf, J., and Blitzer, M.G. (2006). The medical geneticist workforce: an analysis of clinical subgroups. *Genetics in Medicine*. 8(10): 603-614.

<sup>62</sup> Sanford A., Northrup, H., Crandell, S.S., King, T.M., Champaigne, N.L., Yafi, M., Therrell, B.L., and Noblin, S.J. (2009). Expanded newborn screening in Texas: a survey and educational module addressing the knowledge of pediatric residents. *Genetics in Medicine*. 11(3):163-168.

<sup>63</sup> U.S. Government Printing Office. *Public Law 110-204 – Newborn Screening Saves Lives Act of 2007*. See <http://www.gpo.gov/fdsys/pkg/PLAW-110publ204/html/PLAW-110publ204.htm>. Accessed on November 27, 2009.

<sup>64</sup> Cooksey, J.A., Forte, G., Benkendorf, J., and Blitzer, M.G. (2005). The state of the medical geneticist workforce: findings of the 2003 survey of ABMG certified geneticists. *Genetics in Medicine*. 7(6):39-443.

<sup>65</sup> Brotherton, S.E., and Etzel, S.I. (2009). Graduate medical education, 2008-2009. *Journal of the American Medical Association*. 302(12):1357-1372.

346 areas.<sup>66</sup> Genetics professionals include physician geneticists, those with PhDs, genetic counselors, and  
 347 genetics nurses and are collectively referred to as “medical geneticists.”<sup>67</sup> The 1,326 physician  
 348 geneticists certified between 1982 and 2009 represent less than 0.3 percent of the more than 817,000  
 349 physicians in the United States. It is not known how many of these individuals are currently practicing.  
 350 With the exception of genetics counselors, the numbers of medical geneticists achieving certification and  
 351 entering the workforce has, at best, remained flat for the past 15 years.<sup>68</sup> Although workforce planners are  
 352 reluctant to estimate an adequate or an ideal number of medical geneticists needed to provide quality care,  
 353 it is worth noting that historically, physician geneticists in the United States have devoted only 50 percent  
 354 of their time to direct patient care; most are trained in pediatrics; practice trends favor increasing  
 355 specialization as opposed to a general genetics practice or one that would accommodate more than  
 356 patients with rare diseases; nearly three-quarters of practices are full and unable to increase patient load;  
 357 and current practice paradigms are inefficient in comparison with other medical specialty models, with  
 358 physician geneticists reporting on average that they are able to see only seven new and six follow-up  
 359 patients per week.<sup>69</sup>

360  
 361 In 2004, the Royal College of Physicians in London estimated a need for four full-time medical  
 362 geneticists per one million people.<sup>70</sup> Based on a current U.S. population of roughly 307,919,500, the  
 363 United States needs approximately 1,232 full time equivalents (FTE) medical geneticists. According to  
 364 ACMG data, there are currently 540 FTE medical geneticists in the U.S. workforce, 44 percent of the  
 365 number needed. However, the United Kingdom estimate does not take into account the burgeoning  
 366 demand for genetic evaluation and genetic testing as the field experiences a revolutionary acceleration in  
 367 the delineation of rare and ultra rare diseases and as the demand for adult genetics services and testing in  
 368 various sectors, including oncology, cardiology, neurology, and pharmacogenetics, increases. The U.K.’s  
 369 National Health Service, meanwhile, has apparently recognized this impending crisis and has dedicated  
 370 resources to fill the gap. Select general practitioners are encouraged to train under consultant geneticists  
 371 in order to increase the provision of genetic services,<sup>71</sup> and U.K. laboratory geneticists are being re-  
 372 trained to assume new clinical roles as genetic test advisors for general practitioners.<sup>72</sup> In Canada, there is  
 373 one clinical geneticist per 375,000 individuals, a figure that is nearly identical to that in the United  
 374 States,<sup>73,74</sup> and yet waiting times for routine referrals for genetic services range from several weeks to two  
 375 years, with the consequence that referring physicians often do not seek consultations.<sup>75</sup>

376  
 377 Furthermore, the medical geneticist workforce in the United States does not match racial and ethnic  
 378 demographics. In 2003, only 13 percent of medical geneticists identified themselves as members of an

<sup>66</sup> American Board of Medical Genetics. *Number of Certified Specialists in Genetics*. See [http://www.abmg.org/pages/resources\\_certspecial.shtml](http://www.abmg.org/pages/resources_certspecial.shtml). Accessed on March 31, 2010.

<sup>67</sup> Korf, B.R., Ledbetter, D., and Murray, M.F. (2008). Report of the Banbury Summit Meeting on the evolving role of the medical geneticist, February 12-14, 2006. *Genetics in Medicine*. 10(7):502-507.

<sup>68</sup> Personal communication, Judith Benkendorf, M.S., CGC, Special Assistant to the Executive Director, American College of Medical Genetics, April 1, 2010.

<sup>69</sup> Pletcher, B.A., Jewett, E.A.B., Cull, W.L., Brotherton, S.E., Hoyme, H.E., Pan, R.J., and Mulvey, H.J. (2002). The practice of clinical genetics: a survey of practitioners. *Genetics in Medicine*. 4(3):142-149.

<sup>70</sup> Royal College of Physicians. (2004). *Consultant Physicians Working with Patients—the Duties, Responsibilities and Practice for Physicians*, 3<sup>rd</sup> ed. London: Royal College of Physicians.

<sup>71</sup> Martin, G.P., Currie, G., and Finn, R. (2009). Reconfiguring or reproducing intra-professional boundaries? Specialist expertise, generalist knowledge and the ‘modernization’ of the medical workforce. *Social Science in Medicine*. 68:1191-1198.

<sup>72</sup> Henderson, M. “NHS faces genetic revolution by bringing scientists into clinics.” July 30, 2009. Times Online. See <http://www.timesonline.co.uk/tol/news/uk/health/article6732590.ece>. Accessed on November 23, 2009.

<sup>73</sup> Pletcher, B.A., Jewett, E.A.B., Cull, W.L., Brotherton, S.E., Hoyme, H.E., Pan, R.J., and Mulvey, H.J. (2002). The practice of clinical genetics: A survey of practitioners. *Genetics in Medicine*. 4(3):142-149.

<sup>74</sup> Cooksey, J.A., Forte, G., Flanagan, P., Benkendorf, J., Blitzer, M.G. (2006) The Medical Geneticist Workforce: an analysis of clinical subgroups. *Genetics in Medicine*. 8(10): 603-614.

<sup>75</sup> Silversides, A. (2007). The wide gap between genetic research and clinical needs. *Canadian Medical Association Journal*. 176(3):315-316.

379 ethnic or racial minority.<sup>76</sup> Under-representation of diverse populations in the health care workforce has  
380 been cited as a primary barrier to mitigating health care disparities. In summary, the medical genetics  
381 workforce in the United States appears to be several orders of magnitude short of adequate at the dawn of  
382 the genomic age of medicine.

383  
384 **Clinical Translation of Genetics**  
385

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

- 389
- 390 1. What are the attitudes and working knowledge levels of health care professionals regarding  
391 genetics?
  - 392 2. What essential skills and knowledge in genetics are required for competent clinical practice?
  - 393 3. What academic preparation, licensing, and accreditation processes are appropriate for health care  
394 professionals concerning genetics? What continuing education (CE) mechanisms are needed, and  
395 are genetics content required to maintain active licensing or certification? What evidence exists  
396 about the effects of targeted CE efforts?
  - 397 4. Are health care professionals' genetics practice standards and clinical competencies reflected in  
398 current clinical practices? What are the challenges and barriers to health care professional use of  
399 genetics?
- 400

401 **Attitudes and Working Knowledge Levels of Health Care Professionals Regarding Genetics**  
402

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

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<sup>76</sup> Cooksey, J.A., Forte, G., Benkendorf, J., and Blitzer, M.G. (2005) The state of the medical geneticist workforce: findings of the 2003 survey of ABMG certified geneticists. *Genetics in Medicine*. 7(6):439-443.

<sup>77</sup> Collins, F.S. (1997). Preparing health professionals for the genetic revolution. *Journal of the American Medical Association*. 278(15):1285-1286.

<sup>78</sup> Collins, F.S., and Boehm, K. (1999). Avoiding casualties in the gene revolution: the urgent need to educate physicians about genetics. *Academic Medicine*. 74(1):48-49.

<sup>79</sup> Hofman, K.J., Tambor, E.S., Chase, G.A., Geller, G., Faden, R.R., and Holtzman, N.A. (1993). Physicians' knowledge of genetics and genetic tests. *Academic Medicine*. 68(8):625-632.

<sup>80</sup> Escher, M., and Sappino, A.P. (2000). Primary care physicians' knowledge and attitudes towards genetic testing for breast-ovarian cancer predisposition. *Annals of Oncology*. 11(9):1131-1135.

<sup>81</sup> Finn, C.T. (2007). Increasing genetic education for psychiatric residents. *Harvard Review of Psychiatry*. 15(1):30-33.

<sup>82</sup> Kemper, A.R., Uren, R.L., Moseley, K.L., and Clark, S.J. (2006). Primary care physicians' attitudes regarding follow-up care for children with positive newborn screening results. *Pediatrics*. 118(5):1836-1841.

<sup>83</sup> Schroy, P.C., Barrison, A.F., Ling, B.S., Wilson, S., and Geller, A.C. (2002). Family history and colorectal cancer screening: a survey of physician knowledge and practice patterns. *American Journal of Gastroenterology*. 97(4):1031-1036.

<sup>84</sup> Taylor, M.R., Edwards, J.G., and Ku, L. (2006). Lost in transition: challenges in the expanding field of adult genetics. *American Journal of Medical Genetics*. 142C(4):294-303.

<sup>85</sup> Trinidad, S.B., Fryer-Edwards, K., Crest, A., Kyler, P., Lloyd-Puryear, M.A., and Burke, W. (2008). Educational needs in genetic medicine: primary care perspectives. *Community Genetics*. 11(3):160-165.

<sup>86</sup> Wilkins-Haug, L., Hill, L.D., Power, M.L., Holtzman, G.B., and Schulkin, J. (2000). Gynecologists training, knowledge, and experiences in genetics: a survey. *Obstetrics and Gynecology*. 95(3):421-424.

410 protection of their patients, and the accurate role genetics and genomics may play in health disparities.  
411 Discussed below are several notable studies outlining the extent and scope of these challenges.

412  
413 Lack of genetic knowledge among physicians exists not only with complex, multifactorial conditions, but  
414 also with traditional and well-documented Mendelian conditions such as autosomal dominant hereditary  
415 cancer syndromes. An assessment of U.S. physicians regarding hereditary breast, ovarian, and colorectal  
416 cancer genetics identified limited knowledge about key genetic concepts.<sup>87</sup> In one study, a random sample  
417 of 1,251 licensed physician members of the American Medical Association was surveyed across four  
418 groups of primary care (internal medicine, general practice, family practice, and obstetrics) and specialty  
419 care providers (oncology, general surgery, urology, and gastroenterology) about hereditary cancers likely  
420 to be encountered in their clinical practice. Among the findings were that (1) only 37.5 percent of  
421 respondents correctly recognized that hereditary breast cancer due to mutations in *BRCA1* and *BRCA2*  
422 genes could be transmitted through fathers, (2) only 33.8 percent of respondents correctly identified that  
423 less than 10 percent of female breast cancer patients carry *BRCA1* or *BRCA2* mutations, and (3) only 13.1  
424 percent of respondents knew that penetrance of hereditary nonpolyposis colorectal cancer is more than 50  
425 percent in mutation carriers.

426  
427 A survey of psychiatrists' working knowledge, opinions, and practice patterns found that of 352  
428 psychiatrists, 83 percent felt it was their job to discuss genetics and genomics with patients, and while 58  
429 percent discussed genetics with patients, less than 25 percent felt able to do so competently.<sup>88</sup> From the  
430 same study, few psychiatrists could correctly answer more than half of the questions concerning basic  
431 genetics knowledge, and only 15 percent felt that medical training adequately prepared them to address  
432 genetics questions from patients. Many psychiatrists underestimated the contribution of genetic factors to  
433 common, multifactorial diseases regularly seen in their clinical practice. Finn et al.'s 2005 analyses found  
434 a disconnect between a practitioner's understanding of genetic contributors and actual scientific evidence,  
435 for example, estimating that genetic factors account for 30 percent of cases of schizophrenia, when the  
436 scientific literature estimates such a contribution in 70 to 86 percent of cases.

437  
438 White et al. examined the genetic service referral patterns of 284 family physicians. For a clinical  
439 scenario not warranting referral for genetic counseling and testing per U.S. Preventive Services Task  
440 Force guidelines, 92 percent of participants were referred for genetic testing services and 50 percent were  
441 referred for genetic counseling anyway.<sup>89</sup> Education was recommended to maximize appropriate referrals  
442 and improve the role of clinician-patient relationships in referral decisions.

443  
444 These patterns affect nurses as well. A survey of 46 advanced practice nursing students (from nurse  
445 practitioner (N.P.) and nurse anesthesia programs) found that 56 percent of respondents had minimal or  
446 no knowledge of pedigree construction and less than 10 percent indicated a high level of knowledge of six  
447 basic genetic terms such as meiosis and DNA structure and function.<sup>90</sup>

448  
449 The patient perspective confirms these health care provider self-reports and objective knowledge  
450 assessments. A web-based survey of 5,915 patients receiving health care services from approximately 25

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<sup>87</sup> Wideroff, L., Vadaparampil, S.T., Greene, M.H., Taplin, S., Olson, L., and Freedman, A.N. (2005). Hereditary breast/ovarian and colorectal cancer genetics knowledge in a national sample of US physicians. *Journal of Medical Genetics*. 42(10):749-755.

<sup>88</sup> Finn, C.T., Wilcox, M.A., Korf, B.R., Blacker, D., Racette, S.R., Sklar, P., and Smoller, J.W. (2005). Psychiatric Genetics: a survey of psychiatric knowledge, opinions, and practice patterns. *Journal of Clinical Psychiatry*. 66:821-830.

<sup>89</sup> White, D.B., Bonham, V.L., Jenkins, J., Stevens, N. and McBride, C.M. (2008). Too many referrals of low-risk women for BRCA1/2 genetic services by family physicians. *Cancer, Epidemiology, Biomarkers & Prevention*. 17(11):2980-2986.

<sup>90</sup> Maradiegue, A., Edwards, O.T., Seibert, D., Macri, C., and Sitzler, L. (2005). Knowledge, perceptions, and attitudes of advanced practice nursing students regarding medical genetics. *Journal of the American Academy of Nurse Practitioners*. 17(11):472-479.

451 types of health care providers found that 64 percent received no genetics education materials.<sup>91</sup>  
 452 Approximately 50 percent of respondents either had genetic conditions themselves, or had family  
 453 members with genetic conditions and were aware of their risk for genetic diseases through membership in  
 454 genetic advocacy organizations. Health care providers identified as being most often involved in the  
 455 management of the survey respondent's health conditions included family physician and primary care  
 456 providers (54 percent), pediatricians (42.7 percent), cardiologists (35 percent), neurologists (31.8  
 457 percent), ophthalmologists (34.8 percent), and physical therapists (33 percent).

459 Few studies have evaluated genetic education and training needs of pharmacists. A 2003 study assessed  
 460 community pharmacists' confidence in their knowledge about the Human Genome Project, genetic  
 461 testing, and pharmacogenomics<sup>92</sup> and found less than 50 percent with confidence in these topics. In 2004,  
 462 a survey questionnaire administered to pharmacists assessing attitudes relative to genetic testing revealed  
 463 that half agreed that drug development will be faster due to genetic testing and 60 percent either disagreed  
 464 or were neutral about the need for patients' genetic information to be available to improve drug  
 465 dispensing.<sup>93</sup>

467 Genetics knowledge deficiency in health care professionals also extends to policy and legal matters. In  
 468 2004, a California State-based survey of 191 physicians and 80 nurses (registered nurses (R.N.s and  
 469 N.P.s) found that 58.3 percent were misinformed about the existence of protective legislation; more than  
 470 50 percent did not know if cases of health insurance genetic discrimination based on cancer genetic  
 471 testing actually existed; and 13 percent would not refer patients to genetic counseling or for genetic  
 472 testing even if a strong family history of cancer was present.<sup>94</sup> More recently, a study of 1,181 physicians  
 473 and nurse practitioners demonstrated that although 96 percent of respondents viewed genetic testing as  
 474 useful in ascertaining genetic cancer risks, more than 60 percent were unaware of the newly passed  
 475 protective genetic anti-discrimination law.<sup>95</sup>

477 A survey of 428 medical students' attitudes toward genetic testing of children for heritable conditions  
 478 demonstrated that personal understanding and use of genetics was dependent on previous education.<sup>96</sup>  
 479 Even if access to rigorous genetic education and CE exists, health care professionals' personal attitudes  
 480 influence clinical actions. For example, a recent study evaluated 1,121 primary care physicians regarding  
 481 their willingness to use pharmacogenetic testing to tailor smoking treatments.<sup>97</sup> Despite strong evidence  
 482 of the role of genetics in smoking cessation treatment, surveyed physicians purposely avoided use of the  
 483 term "genetic" testing. If the smoking treatment was described in nongenetic terms (i.e., serum protein  
 484 detection), there was greater enthusiasm and interest in using the smoking test.

486 **Adoption of Genetics and Genomics Clinical Competencies for Health Care Professionals**

487

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<sup>91</sup> Harvey, E.K., Fogel, C.E., Peyrot, M., Christensen, K.D., Terry, S.F., and McInerney, J.D. (2007). Providers' knowledge of genetics: a survey of 5915 individuals and families with genetic conditions. *Genetics in Medicine*. 9(5):259-267.

<sup>92</sup> Sangsiry, S.S., and Kulkarni, A.S. (2003). The Human Genome Project: assessing confidence in knowledge and training requirements for community pharmacists. *American Journal of Pharmaceutical Education*. 67(2):291-300.

<sup>93</sup> Sangsiry, S.S., and Kulkarni, A.S. (2004). Genetic testing: the community pharmacist's perspective. *Journal of the American Pharmacists Association*. 44(3):399-402.

<sup>94</sup> Blazer, K.R., MacDonald, D.J., Ricker, C., Sand, S., Uman, G.C., and Weitzel, J.N. (2005). Outcome from intensive training in genetic cancer risk counseling for clinicians. *Genetics in Medicine*. 7(1):40-47.

<sup>95</sup> Lowstuter, K.J., Sand, S., Blazer, K.R., MacDonald, D.J., Banks, K.C., Lee, C.A., Schwerin, B.U., Juarez, M., Uman, G.C., and Weitzel, J.N. (2008). Influence of genetic discrimination perceptions and knowledge on cancer genetics referral practice among clinicians. *Genetics in Medicine*. 10(9):691-698.

<sup>96</sup> Riordan, S.H., and Loescher, L.J. (2006). Medical students' attitudes toward genetic testing of minors. *Genetic Testing*. 10(1):68-73.

<sup>97</sup> Sheilds, A.E., Levy, D.E., Blumenthal D., Currivan, D., McGinn-Shapiro, M., Weiss, K.B., Yucel, R., and Lerman, C. (2007). Primary care physicians' willingness to offer a new genetic test to tailor smoking treatment, according to test characteristics. *Nicotine & Tobacco Research*. 10(6):1037-1045.

488 The National Coalition of Health Professional Education in Genetics<sup>98</sup> (NCHPEG) identified overarching  
 489 clinical competencies for all health care professionals and various professional groups have developed  
 490 clinical competencies for their individual disciplines. For example, pedigree assessment is incorporated  
 491 into many competency recommendations. When properly conducted, family history is widely regarded as  
 492 a mechanism by which to detect familial transmission of hereditary diseases, such as familial cancer  
 493 syndromes and common multifactorial diseases.<sup>99,100,101,102,103</sup> Its role is recognized as being so important  
 494 that public service announcements have been created, aimed at engaging the U.S. public in understanding  
 495 how their family history can be used to guide health care decisionmaking.<sup>104</sup>

496  
 497 While national public health campaigns are encouraging individuals to bring their family histories to their  
 498 health care providers, family history proficiency is not a competency required in order to graduate from a  
 499 medical education program. Clinicians are hesitant to incorporate use of family history assessment due to  
 500 time constraints, questions about clinical utility, beliefs of unreliability, and absence of meaningful  
 501 financial reimbursement.<sup>105,106,107</sup> Greb et al. performed an analysis of medical genetics knowledge and  
 502 skill retention in 212 medical students following their third year and found that only 36.8 percent  
 503 correctly asked about presence of family history in a cystic fibrosis case scenario.<sup>108</sup> This trend is present  
 504 in nursing students as well; only 22 percent of 46 Advanced Practice Nursing students in N.P. programs  
 505 felt they could draw a family pedigree.<sup>109</sup> Despite encouragement to use family history in primary care,<sup>110</sup>  
 506 the National Institutes of Health (NIH) State-of-the Science Conference on Family History and Improving  
 507 Health, held in August of 2009, concluded that, while family history plays an important role in medicine,  
 508 more research is needed before a systematically collected family history for common disease will become  
 509 an evidence-based tool in primary care settings.<sup>111</sup>

510 Other genetics skills recommended by consensus panels are encountering similar challenges in accurate  
 511 clinical uptake and dissemination. For example, a key genetics competency is the ability to counsel

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<sup>98</sup> National Coalition of Health Professional Education in Genetics website. See <http://www.nchpeg.org/>. Accessed on March 8, 2010.

<sup>99</sup> U.S. Preventive Services Task Force. (2005). Summaries for patients. Genetic risk assessment and BRCA mutation testing for breast and ovarian cancer susceptibility: U.S. Preventive Services Task Force recommendations. *Annals of Internal Medicine*. 143(5):147.

<sup>100</sup> Centers for Disease Control and Prevention. (2004). Awareness of family health history as a risk factor for disease—United States 2004. *Morbidity and Mortality Weekly Report*. 53(44):1044-1047.

<sup>101</sup> Frezzo, T.M., Rubinstein, W.S., Dunham, D., and Ormond, K.E. (2003). The genetic family history as a risk assessment tool in internal medicine. *Genetics in Medicine*. 5(2):84-91.

<sup>102</sup> Qin, P., Agerbo, E., and Mortenson, P.B. (2002). Suicide risk in relation to family history of completed suicide and psychiatric disorders: a nested case-control study based on longitudinal registers. *Lancet*. 360(9340):1126-1130.

<sup>103</sup> Williams, R.R., Hunt, S.C., Heiss, G., Province, M.A., Benson, J.T., Higgins, M., Chamberlain, R.M., Ware, J., and Hopkins, P.M. (2001). Usefulness of cardiovascular family history data for population-based preventive medicine and medical research (the Health Family Tree Study and the NHLBI Family Heart Study). *The American Journal of Cardiology*. 87(2):129-35.

<sup>104</sup> U.S. Department of Health and Human Services. Surgeon General's Family Health History Initiative. See <http://www.hhs.gov/familyhistory/>. Accessed on November 25, 2009.

<sup>105</sup> Guttmacher, A.E., Collins, F.S., and Carmona, R.H. (2004). The family history—more important than ever. *The New England Journal of Medicine*. 351(22):2333-2336.

<sup>106</sup> Rich, E.C., Burke, W., Heaton, C.J., Haga, S., Pinsky, L., Short, M.P., and Acheson, L. (2004). Reconsidering the family history in primary care. *Journal of General Internal Medicine*. 19(3):273-280.

<sup>107</sup> Suther, S., and Goodson, P. (2003). Barriers to the provision of genetic services by primary care physicians: a systematic review of the literature. *Genetics in Medicine*. 5(2):70-76.

<sup>108</sup> Greb, A.E., Brennan, S., McParlane, L., Rage, R., and Bridge, P.D. (2009). Retention of medical genetics knowledge and skills by medical students. *Genetics in Medicine*. 11(5):1-6.

<sup>109</sup> Maradiegue, A., Edwards, O.T., Seibert, D., Macri, C., and Sitzler, L. (2005). Knowledge, perceptions, and attitudes of advanced practice nursing students regarding medical genetics. *Journal of the American Academy of Nurse Practitioners*. 17(11):472-479.

<sup>110</sup> Guttmacher, A.E., Collins, F.S., and Carmona, R.H. (2004). The family history—more important than ever. *The New England Journal of Medicine*. 351(22):2333-2336.

<sup>111</sup> National Institutes of Health Consensus Development Program: NIH State of the Science Conference, Family History and Improving Health. See <http://consensus.nih.gov/2009/familyhistory.htm>. Accessed on November 16, 2009.

512 patients about genetic concerns and correctly issue referrals for genetic services. In a study of 900  
513 internists, obstetricians, and oncologists regarding breast cancer (i.e., *BRCA1* and *BRCA2*) mutation  
514 testing, only 13 percent of internists, 21 percent of obstetricians, and 40 percent of oncologists could  
515 correctly answer four basic genetics concept questions.<sup>112</sup> In the same study, although greater genetic  
516 knowledge influenced frequency of discussing the *BRCA* genetic test with patients, 54 percent of  
517 oncologists operating on inaccurate genetics concepts discussed genetic testing with their patients and  
518 presumably, made health-related decisions regarding their care.

519  
520 More recently, an analysis of the use of genetic services (for breast/ovarian/colon cancer, Huntington  
521 disease, and sickle cell disease) by U.S. primary care physicians shows that up to two-thirds of those  
522 surveyed ordered genetic tests, and more than three-quarters referred patients for genetic counseling.<sup>113</sup>  
523 However, there were clear differences in patterns of genetic service referrals, with providers serving  
524 minority populations being significantly less likely to order testing or issue referrals.

525  
526 Extending beyond well-validated applications, complexities will be considerably greater for management  
527 of chronic, multifactorial diseases. A recent comprehensive review of the literature shows little data  
528 available to health care providers interested in using genetics to manage adult-onset conditions.<sup>114</sup> These  
529 reviews suggest that until health outcome data on genetic technologies exists and there are clear and  
530 accessible education mechanisms for current health care providers and students, use of genetics as  
531 outlined in competency statements is not likely to reach the bedside without further strategic support.

### 532 533 **Essential Skills and Knowledge in Genetics and Genomics Required for Competent Clinical** 534 **Practice**

535  
536 Although several disciplines have overarching clinical practice competency standards, numerous  
537 professional societies and organizations spanning Federal, academic, private, and public domains have  
538 developed recommendations according to overarching practice requirements and clinical subspecialty.  
539 Professional competencies are discussed according to Hundert and Epstein's 2002 definition:  
540 "Competency is the habitual and judicious use of communication, knowledge, technical skills, clinical  
541 reasoning, emotions, values, and reflection in daily practice for the benefit of the individual and the  
542 community being served."<sup>115</sup>

543  
544 Setting the stage for genetic competency in clinical practice, NCHPEG first developed a list of necessary  
545 competencies for health care professionals in 2001 in expectation of the completion of the Human  
546 Genome Project.<sup>116</sup> Initially, the list included 44 competencies but was revised in 2005 and again in 2007,  
547 to its current 18 competencies (see Appendix A-2 for complete wording) reflecting the rapidly changing  
548 biomedical landscape and recognition of the need to focus competencies on measureable outcomes. Three  
549 minimum expectations for health care professionals across all clinical practice settings advise them to  
550 identify when professional development related to genetics and genomics would be beneficial, understand  
551 the social and psychological implications of health-related genetic information on patients and families,

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<sup>112</sup> Doksum, T., Bernhardt, B.A., and Hotlzman, N.A. (2003). Does knowledge about the genetics of breast cancer differ between nongeneticist physicians who do or do not discuss or order BRCA testing? *Genetics in Medicine*. 5(2):99-105.

<sup>113</sup> Sheilds, A.E., Burke, W., and Levy, D.E. (2008). Differential use of available genetic tests among primary care physicians in the United States: results of a national survey. *Genetics in Medicine*. 10(6):404-414.

<sup>114</sup> Scheuner, M.T., Sieverding, P., and Shekelle, P.G. (2008). Delivery of genomic medicine for common chronic adult diseases. *Journal of the American Medical Association*. 299(11):1320-1334.

<sup>115</sup> Epstein, R.M., and Hundert, E.M. (2002). Defining and assessing professional competence. *Journal of the American Medical Association*. 287(2):226-235.

<sup>116</sup> National Coalition for Health Professional Education in Genetics. (2007). Core competencies in genetics for health professionals. 3<sup>rd</sup> edition. See <http://www.nchpeg.org/>. Accessed on November 16, 2009.

552 and know how and when to make a genetics referral. More specific recommendations within these three  
 553 content areas are provided in Appendix A-2.

554  
 555 Similar to NCHPEG's overarching competencies, various practice recommendations and/or genetics-  
 556 specific clinical competencies have emerged. These practice recommendations largely fall into two broad  
 557 categories and are health professional discipline-specific or issued according to clinical subspecialties in  
 558 medical, nursing, physician assistant, and genetic counseling communities. See Appendix A-2 for a  
 559 discussion and listing of competencies for physicians, nurses, genetic counselors, and pharmacists.

560  
 561 **Academic Preparation, Licensing, and Continuing Education**

562  
 563 The dearth of genetics and genomics content in pre-professional, health professional curricula, and CE is  
 564 widely recognized across all disciplines.<sup>117</sup> In addition, larger, more global issues affecting all disciplines  
 565 are well recognized and include outdated models of scientific instruction, shortage of adequately trained  
 566 academic faculty, limited purview of genetics as single-gene disorders only, and inability to commit to  
 567 genetics CE given many other competing needs and priorities.<sup>118</sup> A review of basic training and  
 568 education, licensure, post graduate education of health care professionals and accreditation of professional  
 569 schools is provided in Appendix A-3.

570  
 571 **Continuing Education in Genetics and Genomics**

572  
 573 Health care professionals generally are optimistic about the future utility of genetic tests and are  
 574 interested in their eventual incorporation into clinical practice.<sup>119,120</sup> But many experience feelings of  
 575 discomfort stemming from lack of confidence in their knowledge of basic genetic concepts, interventions,  
 576 and management strategies.<sup>121,122,123</sup> Thus, many focused resources have been devoted to CE efforts for  
 577 health care professionals in regional practice settings.<sup>124,125,126,127</sup> Standard instructional methods used to  
 578 convey genetic and genomic content in these endeavors include web-based instruction tutorials, CE  
 579 seminars, professional workshops, and conference proceedings.<sup>128,129,130,131</sup>

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<sup>117</sup> McInerney, J.D. (2007). Genetics education for health professionals: a context. *Journal of Genetic Counselors*. 17:145-151.

<sup>118</sup> Guttmacher, A.E., Porteous, M.E., and McInerney, J.D. (2007). Educating health-care professionals about genetics and genomics. *Nature Reviews*. 8:151-157.

<sup>119</sup> Escher, M., and Sappino, A.P. (2000). Primary care physicians' knowledge and attitudes towards genetic testing for breast-ovarian cancer predisposition. *Annals of Oncology*. 11(9):1131-1135.

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

<sup>121</sup> Edwards, Q.T., Maradiegue, A., Seibert, D., Saunders-Goldson, S., and Humphreys, S. (2009). Breast cancer risk elements and nurse practitioners' knowledge, use, and perceived comfort level of breast cancer risk assessment. *Journal of the American Academy of Nurse Practitioners*. 21(5):270-277.

<sup>122</sup> Gramling, R., Nash, J., Siren, K., Eaton, C., and Culpepper, L. (2004). Family physician self-efficacy with screening for inherited cancer risk. *Annals of Family Medicine*. 2(2):130-132.

<sup>123</sup> Watson, E., Clements, A., Yudkin, P., Rose, P., Bukach, C., Mackay, J., Lucassen, A., and Austoker, A. (2001). Evaluation of the impact of two educational interventions on GP management of familial breast/ovarian cancer cases: a cluster randomized controlled trial. *The British Journal of General Practice*. 51(471):817-821.

<sup>124</sup> Blazer, K.R., MacDonald, D.J., Ricker, C., Sand, S., Uman, G.C., and Weitzel, J.N. (2005). Outcome from intensive training in genetic cancer risk counseling for clinicians. *Genetics in Medicine*. 7(1):40-47.

<sup>125</sup> Blazer, K.R., Grant, M., Sand, S.R., MacDonald, D.J., Uman, G.C., and Weitzel, J.N. (2004). Effects of a cancer genetics programme on clinician knowledge and practice. *Journal of Medical Genetics*. 41(7):518-522.

<sup>126</sup> Clyman, J.C., Nazir, F., Tarolli, S., Black, E., Lombardi, R.Q., and Higgins, J.J. (2007). The impact of a genetics education program on physicians' knowledge and genetic counseling referral patterns. *Medical Teacher*. 29(6):e143-150.

<sup>127</sup> Prows, C.A., Hetteberg, C., Hopkins, R.J., Latta, K.K., and Powers, S.M. (2004). Development of a web-based genetics institute for a nursing audience. *Journal of Continuing Education in Nursing*. 35(5):223-231.

<sup>128</sup> Centers for Disease Control and Prevention. Public Health Genomics, Genomic Resources. See <http://www.cdc.gov/genomics/resources/t.htm>. Accessed on November 25, 2009.

580 However, access to and participation in CE does not determine proficiency in providing clinical care. An  
 581 extensive meta-analysis completed through The Cochrane Collaboration demonstrates that improvement  
 582 in health care provider behaviors (through CE) and ultimately, patient health outcomes, is dependent on  
 583 the complexity of the learned material, method of instruction, and health care providers' access to  
 584 interactive practice.<sup>132,133</sup> This finding has significant implications for effective clinical translation of  
 585 genetics and genomics into diffusible health practices.

587 A recent study examining the impact of a genetic outreach education initiative found that although health  
 588 care professionals felt more confident in using genetics after the CE intervention, 48 percent of  
 589 respondents applied their new knowledge incorrectly.<sup>134</sup> Incorrect application of genetics occurred in the  
 590 misappropriation of risk estimation; approximately half of those receiving the genetics education  
 591 intervention assigned a high-risk categorization to a low-risk breast cancer presentation. Reinforcement of  
 592 complex content is important to assure appropriate and accurate use of genetic information. A genetics  
 593 education program that increased knowledge and confidence in genetic competencies among physicians  
 594 delivering genetic services in primary care settings utilized an interactive, case-based, peer education  
 595 model.<sup>135</sup> Peer education emphasizes the usability of the educational materials and concepts and was  
 596 rated as an effective method by most participants in this study.

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

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

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<sup>129</sup> Centers for Disease Control and Prevention. Public Health Genomics, Genomic Resources. Educational Materials. See <http://www.cdc.gov/genomics/resources/e.htm>. Accessed on November 25, 2009.

<sup>130</sup> Harvard Medical School. The Genetic Basis of Adult Medicine. See <http://cme.med.harvard.edu/cmeups/custom/00281416/index.htm>; Accessed on November 25, 2009.

<sup>131</sup> Dartmouth Medical School. Genetics in Clinical Practice, A Team Approach. See <http://iml.dartmouth.edu/education/cme/Genetics/>. Accessed on November 25, 2009.

<sup>132</sup> Forsetlund, L., Bjørndal, A., Rashidian, A., Jamtvedt, G., O'Brien, M.A., Wolf, F., Davis, D., Odgaard-Jensen, J., and Oxman, A.D. (2009). Continuing education meetings and workshops: effects on professional practice and health care outcomes. *Cochrane Database of Systemic Reviews*. 15(2):CD003030.

<sup>133</sup> Davis, D., O'Brien, M.A., Freemantle, N., Wolf, F.M., Mazmanian, P., and Taylor-Vaisey, A. (1999). Impact of formal continuing medical education: do conferences, workshops, rounds, and other traditional continuing education activities change physician behavior or health care outcomes? *The Journal of the American Medical Association*. 282(9):867-874.

<sup>134</sup> Bethea, J., Qureshi, N., Drury, N., and Guilbert, P. (2008). The impact of genetic outreach education and support to primary care on practitioner's confidence and competence in dealing with familial cancers. *Community Genetics*. 11(5):289-294.

<sup>135</sup> Carroll, J.C., Rideout, A.L., Wilson, B.J., Allanson, J., Blaine, S.M., Esplen, M.J., Farrell, S.A., Graham, G.E., MacKenzie, J., Meschino, W., Miller, F., Prakash, P., Shuman, C., Summers, A., and Taylor, S. (2009). Geneti education for primary care providers. *Canadian Family Physician*. 55:e92-99.

<sup>136</sup> Davis, J.C., Furstenthal, L., Desai, A.A., Norris, T., Sutaria, S., Fleming, E., and Ma, P. (2009). The microeconomics of personalized medicine: today's challenge and tomorrow's promise. *Nature Reviews Drug Discovery*. 8:279-286.

<sup>137</sup> Wideroff, L., Vadaparampil, S.T., Greene, M.H., Taplin, S., Olson, L., and Freedman, A.N. (2005). Hereditary breast/ovarian and colorectal cancer genetics knowledge in a national sample of US physicians. *Journal of Medical Genetics*. 42(10):749-755.

<sup>138</sup> Epstein, R.S., Frueh, F.W., Geren, D., Hummer, D., McKibben, S., O'Connor, S., Randhawa, G., and Zelman, B. (2009). Payer perspectives on pharmacogenomics testing and drug development. *Pharmacogenomics*. 10(1):149-151.

609 ascertainment of financial benefits and costs.<sup>139</sup> Also, the need for national guidelines stemming from  
 610 health care outcome data is a common theme across health care provider reports of willingness to use  
 611 genetics.<sup>140,141,142,143,144</sup>

612  
 613 Another barrier to integrating genetics in education, training, and practice relates to economic challenges  
 614 unique to the U.S. health care delivery system. Misalignment of financial incentives between payers,  
 615 health care providers, patients, pharmaceutical and biotechnology companies, and diagnostics research  
 616 and development companies, are well documented.<sup>145,146</sup> The competing priorities among these  
 617 stakeholders have significant implications as to whether a useful genetic test or genomic technology will  
 618 be incorporated into health care delivery practices. For example, some analysts have written that  
 619 establishing high scientific thresholds of evidence for financial reimbursement (payers) can greatly  
 620 influence whether a health care professional uses what they have been taught concerning genetics and  
 621 genomics.<sup>147</sup> Synchronization of stakeholder interests between the U.S. health care and clinical research  
 622 systems has been identified as a key priority in meeting upcoming and current U.S. genetics data needs.<sup>148</sup>

623  
 624 Coverage and reimbursement of genetic tests and services has been identified as limiting their  
 625 accessibility and integration into the health care system. The current state of coverage and reimbursement  
 626 of genetic tests and services and recommendations on how to improve mechanisms for coverage and  
 627 reimbursement are covered in detail in the SACGHS report on the coverage and reimbursement of genetic  
 628 tests and services.<sup>149</sup>

629  
 630 **Health Care Professional Faculty Development in Genetics and Genomics**

631  
 632 As noted throughout the previous sections, genetics content of health sciences curricula is variable and  
 633 tends to focus on single-gene disorders. It is often not presented in a way that leads to long-term  
 634 knowledge retention for clinical application.<sup>150,151,152</sup> Strategies identified to address these deficiencies

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<sup>139</sup> Rogowski, W.H., Grosse, S.D., and Khoury, M.J. (2009). Challenges of translating genetic tests into clinical and public health practice. *Nature Reviews Genetics*. 10(7):489-495.

<sup>140</sup> Freedman, A.N., Wideroff, L., Olson, L., Davis, W., Klabunde, C., Srinath, K.P., Reeve, B.B., Croyle, R.T., and Ballard-Barbash, R. (2003). US physicians' attitudes toward genetic testing for cancer susceptibility. *American Journal of Medical Genetics*. 120A(1):63-71.

<sup>141</sup> Hindorff, L.A., Burke, W., Laberge, A.M., Rice, K.M., Lumley, T., Leppig, K., Rosendaal, F.R., Larson, E.B., and Psaty, B.M. (2009). Motivating factors for physician ordering of factor V Leiden genetic tests. *Archives of Internal Medicine*. 169(1):68-74.

<sup>142</sup> Suther, S.G., and Goodson, P. (2004). Texas physicians' perceptions of genomic medicine as an innovation. *Clinical Genetics*. 65(5):368-377.

<sup>143</sup> Suther, S., and Goodson, P. (2003). Barriers to the provision of genetic services by primary care physicians: a systematic review of the literature. *Genetics in Medicine*. 5(2):70-76.

<sup>144</sup> Zapka, J.G., Puleo, E., Taplin, S., Solberg, I.E., Mouchawar, J., Somkin, C., Geiger, A.M., and Ulcickas Yood, M. (2005). Breast and cervical cancer screening: clinicians' views on health plan guidelines and implementation efforts. *Journal of the National Cancer Institute Monographs* 2005. (35):46-54.

<sup>145</sup> Secretary's Advisory Committee on Genetics, Health, and Society. (2006). Coverage and Reimbursement of Genetic Tests and Services. See [http://oba.od.nih.gov/oba/sacghs/reports/CR\\_report.pdf](http://oba.od.nih.gov/oba/sacghs/reports/CR_report.pdf). Accessed on November 25, 2009.

<sup>146</sup> Secretary's Advisory Committee on Genetics, Health, and Society. (2008). U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of Health and Human Services. See [http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS\\_oversight\\_report.pdf](http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf). Accessed on November 25, 2009.

<sup>147</sup> Khoury, M.J., Berg, A., Coates, R., Evans, J., Teutsch, S.M., and Bradley, L.A. (2008). The evidence dilemma in genomic medicine. *Health Affairs*. 27(6):1600-1611.

<sup>148</sup> Califf, R.M. (2004). Defining the balance of risk and benefit in the era of genomics and proteomics. *Health Affairs*. 23(1):77-87.

<sup>149</sup> Secretary's Advisory Committee on Genetics, Health, and Society. (2006). Coverage and Reimbursement of Genetic Tests and Services. See [http://oba.od.nih.gov/oba/sacghs/reports/CR\\_report.pdf](http://oba.od.nih.gov/oba/sacghs/reports/CR_report.pdf). Accessed on November 25, 2009.

<sup>150</sup> Greb, A.E., Brennan, S., McParlane, L., Rage, R., and Bridge, P.D. (2009). Retention of medical genetics knowledge and skills by medical students. *Genetics in Medicine*. 11(5):1-6.

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

637  
 638 Given the dearth of medical genetics experts, faculty development is a key concern that was identified  
 639 across many health care education programs.<sup>155</sup> For example, a random convenience survey of N.P.  
 640 faculty ascertained that although 95 percent of faculty identified genetics as being important only 10  
 641 percent reported their academic institution as offering a genetics course as part of the N.P. curriculum,  
 642 and 20 percent reported instruction of genetics as limited to Mendelian content.<sup>156</sup> A study published in  
 643 2010 assessed the level of faculty development in pharmacogenomics by surveying U.S. pharmacy  
 644 schools. It found that most of the 75 schools responding to the survey included pharmacogenomics  
 645 content in their curricula, however, more than half of these schools had no plans for faculty development  
 646 in this area.<sup>157</sup>

647  
 648 More promising are the long-term results from the “Genetics in Primary Care” Faculty Development  
 649 Initiative, where follow-up data indicated permanent changes in teaching (100 percent at three years) and  
 650 in clinical practice habits (82 percent at three years).<sup>158</sup> This experimental model is notable for its  
 651 collaboration across education, genetics, and primary care experts, who designed curricula and case  
 652 studies to provide a standardized genetics instruction format that also incorporates evidence and  
 653 assessment skills for newly released scientific findings. Although this model was able to promote long-  
 654 term behavioral changes and comfort with genetics, there remain significant challenges. Only 9 percent of  
 655 the faculty respondents reported teaching their medical students and residents how and when to refer a  
 656 patient for genetic counseling; 18 percent reported incorporation of formal genetics teaching for their  
 657 primary care residents; and 36 percent increased the amount of genetics in medical school curriculums.

658  
 659 **Clinical Decision Support and Electronic Health Records**

660  
 661 Recent studies assessing genetic content in a variety of commonly used online medical resources  
 662 identified large gaps in content as well as significant errors in the information that was available.<sup>159</sup>

663  
 664 As electronic health records are increasingly deployed in clinical care, a potential solution to these issues  
 665 has emerged—just-in-time education. Just-in-time education provides specific answers to specific  
 666 provider questions at the time the provider asks a question. A study by Trinidad et al. regarding genetic

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<sup>151</sup> Thurston, V.C., Wales, P.S., Bell, M.A., Torbeck, L., and Brokaw, J.J. (2007). The current status of medical genetics instruction in US and Canadian medical schools. *Academic Medicine*. 82(5):441-445.  
<sup>152</sup> Hetteberg, C.G., Prows, C.A., Deets, C., Monsen, R.B., and Kenner, C.A. (1999). National survey of genetics content in basic nursing preparatory programs in the United States. *Nursing Outlook*. 47(4):168-180.  
<sup>153</sup> Howard Hughes Medical Institute-Association of American Medical Colleges. (2009). *Scientific Foundations for Future Physicians: Report of the HHMI-AAMC Committee*. See [www.hhmi.org/grants/pdf/08-209\\_AAMC-HHMI\\_report.pdf](http://www.hhmi.org/grants/pdf/08-209_AAMC-HHMI_report.pdf). Accessed on November 24, 2009.  
<sup>154</sup> Prows, C.A., Glass, M., Nicol, M.J., Skiton, H., and Williams, J. (2005). Genomics in nursing education. *Journal of Nursing Scholarship*. 37(3):196-202.  
<sup>155</sup> Guttmacher, A.E., Porteous, M.E., and McInerney, J.D. (2007). Educating health-care professionals about genetics and genomics. *Nature Reviews*. 8:151-157.  
<sup>156</sup> Edwards, Q.T., Maradiegue, A., Siebert, D., Macri, C., and Sitzer, L. (2006). Faculty members' perceptions of medical genetics and its integration into nurse practitioner curricula. *The Journal of Nursing Education*. 45(3):124-130.  
<sup>157</sup> Murphy, J.E., Green, J.S., Adams, L.A., Squire, R.B., Kuo, G.M., and McKay, A. (2010). Pharmacogenomics in the curricula of colleges and schools of pharmacy in the United States. *American Journal of Pharmaceutical Education*. 74(1):1-10.  
<sup>158</sup> Laberge, A.M., Fryer-Edwards, K., Kyler, P., Lloyd-Puryear, M.A., and Burke, W. (2009). Long-term outcomes of the “Genetics in Primary Care” Faculty Development Initiative. *Family Medicine*. 41(4):266-70.  
<sup>159</sup> Levy, H.P., LoPresti, L., and Siebert, D.C. (2008). Twenty questions in genetic medicine—an assessment of World Wide Web databases for genetics information at the point of care. *Genetics in Medicine*. 10(9):659-657.

667 education needs of primary care providers identified a desire to have “just-in-time” resources available.<sup>160</sup>  
 668 The applicability of active decision support in genetics was discussed in detail in a previous SACGHS  
 669 report.<sup>161</sup>  
 670  
 671 The key to the provision of a specific answer provided in response to a question involves the use of  
 672 context-sensitive elements embedded in the EHR. This approach involves the EHR “understanding”  
 673 where the provider is in the patient workflow so that when the query is executed the provider is taken to  
 674 content that is highly likely to be relevant to the question the provider is considering. A study by del Fiol  
 675 et al. demonstrated that answers could be found significantly faster using infobuttons<sup>162</sup> than traditional  
 676 electronic search approaches.<sup>163</sup> At Intermountain Healthcare, more than 200 infobuttons relating to  
 677 genetic disorders in the problem list were linked to specific genetic information contained at GeneTests<sup>164</sup>  
 678 and Genetics Home Reference<sup>165</sup> in addition to traditional sources such as Online Mendelian Inheritance  
 679 in Man (OMIM).<sup>166</sup> Following implementation, analysis of the usage of these genetic-specific infobuttons  
 680 has continued to increase over time with good provider satisfaction.<sup>167</sup>  
 681  
 682 The Mayo clinic has also deployed a just-in-time approach to deliver genomic information to  
 683 providers.<sup>168</sup> To date, no rigorous studies have assessed the effectiveness of these types of educational  
 684 interventions in acquiring and retaining new knowledge that alters practice behavior; however, studies  
 685 such as one by Maviglia et al. demonstrated that providers found answers to questions about medications  
 686 84 percent of the time with an average elapsed time from question to answer of 21 seconds.<sup>169</sup>  
 687 Preliminary data from the Intermountain Healthcare System specific to genetic content suggested that  
 688 providers needed to spend longer amounts of time (~45 seconds) accessing the resource, but no data are  
 689 available at this time to address whether specific questions were answered. Just-in-time learning has also  
 690 been deployed to patients in a variety of health care settings. Many of these involve breast cancer care  
 691 including innovative approaches in patients with low literacy<sup>170</sup> and to aid in genetic testing decisions.<sup>171</sup>  
 692 Thus, this approach appears to be a promising way to deliver genetic knowledge to the provider.  
 693

<sup>160</sup> Trinidad, S.B., Fryer-Edwards, K., Crest, A., Kyler, P., Lloyd-Puryear, M.A., and Burke, W. (2008). Educational needs in genetic medicine: primary care perspectives. *Community Genetics*. 11(3):160-5

<sup>161</sup> Secretary’s Advisory Committee on Genetics, Health, and Society. (2008). *U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of Health and Human Services*. See [http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS\\_oversight\\_report.pdf](http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf). Accessed on September 3, 2009.

<sup>162</sup> Infobuttons are icons that appear in certain areas of the EHR (e.g., problem list, medication list, and laboratory results).

<sup>163</sup> Del Fiol, G., Haug, P.J., Cimino, J.J., Narus, S.P., Norlin, C., and Mitchell, J.A. (2008). Effectiveness of topic-specific infobuttons: a randomized controlled trial. *Journal of the American Medical Information Association*. 15(6):752-9.

<sup>164</sup> GeneTests. See <http://www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests>. Accessed on November 17, 2009.

<sup>165</sup> National Library of Medicine Genetics Home Reference. See <http://ghr.nlm.nih.gov/>. Accessed on November 19, 2009.

<sup>166</sup> Online Mendelian Inheritance in Man. See <http://www.ncbi.nlm.nih.gov/omim/>. Accessed on May 18, 2010.

<sup>167</sup> Personal communication, Marc S. Williams, M.D., Director, Intermountain Healthcare Clinical Genetics Institute, unpublished data, September 3, 2009.

<sup>168</sup> Kaihoi, B., Petersen, C., and Bolander, M.E. (2005). Providing "just-in-time" medical genomics information for patient care. *American Medical Information Association Annual Symposium Proceedings*. 1003.

<sup>169</sup> Maviglia, S.M., Yoon, C.S., Bates, D.W., and Kuperman, G. (2006). KnowledgeLink: impact of context-sensitive information retrieval on clinicians' information needs. *Journal of the American Medical Information Association*. 13(1):67-73.

<sup>170</sup> Jibaja-Weiss, M.L., Volk, R.J., Friedman, L.C., Granchi, T.S., Neff, N.E., Spann, S.J., Robinson, E.K., Aoki, N., and Beck, R. (2006). Preliminary testing of a just-in-time, user-defined values clarification exercise to aid lower literate women in making informed breast cancer treatment decisions. *Health Expectations*. 9(3):218-31.

<sup>171</sup> Green, M.J., Peterson, S.K., Baker, M.W., Harper, G.R., Friedman, L.C., Rubinstein, W.S., and Mauger, D.T. (2004). Effect of a computer-based decision aid on knowledge, perceptions, and intentions about genetic testing for breast cancer susceptibility: a randomized controlled trial. *Journal of the American Medical Association*. 292(4):442-52.

694 **C. SACGHS Surveys of Health Professional Organizations**

695

696 **Methodology**

697

698 In 2008, SACGHS surveyed selected organizations with responsibilities across the continuum of health  
699 professional education to obtain information regarding their activities in genetics education. Key staff  
700 members in 60 targeted organizations (see Appendix B-1 for a listing of these organizations) were  
701 contacted via e-mail to respond to the survey, which consisted of 15 open- and close-ended questions  
702 developed by SACGHS. The survey explored several major themes including the organizations'  
703 perceived role in, and priority ascribed to genomics education; barriers to enhancing their role in  
704 genomics education; and a description of their past, present, and planned efforts around genomics  
705 education.

706

707 The survey was determined to be exempt from the need for Institutional Review Board review and  
708 approval by the NIH Office of Human Subjects Research. See Appendix B-2 for more information on the  
709 methodologies and Appendix B-3 for the survey instrument. Thirty-six responses were received (60  
710 percent). See Appendix B-4 for a list of the responding organizations and their reported number of  
711 members or constituency.

712

713 The survey population was determined by consensus among SACGHS members and staff. Selection  
714 criteria for inclusion included the diversity of levels of training within the organization and the  
715 organization's role in training professionals destined to provide primary care services. Organizations that  
716 play a central role in training nurses and primary care physicians, as well as organizations representing  
717 genetic professionals with a key role in supporting nongenetics health professionals were specifically  
718 targeted. In addition, three federal advisory committees relevant to genomics education were invited to  
719 complete the survey.

720

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

730

731 **Survey Findings**

732

733 ***Roles and Responsibility for Genetics Education and Training***

734

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

738

739 In response to a question asking if the organization was able to fulfill its role or responsibility in genetics  
740 education and training, 21 of 25 (84 percent) responding organizations fulfilled their role, one did not, and  
741 two stated that they were able to fulfill this role partially. Two of the responses delineate the barriers  
742 organizations face in fulfilling their responsibility for genetic education and training:

743

744 American College of Physicians: “We are able to develop programs and products related to  
745 genetics education. The difficulty is getting members to be interested in them.”  
746

747 American Academy of Pediatrics (AAP): “The AAP is currently and actively engaged in this  
748 activity. It seems implausible to suggest that any single organization could “fulfill” the role of  
749 educating 60,000 pediatricians on the topic of genetics.”  
750

751 Several themes emerged from the survey about how organizations can meet their role or responsibility  
752 more effectively. These strategies included increasing funding, evaluating current activities, and  
753 generating greater interest with institutional leaders and through publications and annual conferences.  
754

755 ***Importance of Genetics and Genomics to the Organization, and Leadership Proficiency in Genetics***  
756 ***Education***  
757

758 In general, the priority placed on genetics education varied depending on the focus of the organization  
759 with genetics-specific organizations indicating the strongest interest. Although other professional  
760 organizations assigned moderate priority to genetics education, they also reported that such activities are  
761 very important. As expected, high levels of proficiency and comfort in genetics education were found  
762 only in genetics-specific organizations.  
763

764 ***Entities within Organizations Dedicated to Genetics Education***  
765

766 Organizations were asked if they have an established committee, workgroup, or dedicated staff that deals  
767 specifically with topics in genetics relevant to their organization’s mission. Thirty-six percent of  
768 nongenetic-specific professional organizations indicated that they had such entities, with more than twice  
769 the activity occurring in genetics-specific organizations. See Appendix B-5, Table 2 for a breakdown  
770 among organization categories.  
771

772 ***Barriers to Providing Genetics Educational Activities***  
773

774 From a list of seven barriers, more than half of all organizations indicated competing priorities and one-  
775 third indicated that genetics and genomics was not emphasized in certifying exams and credentialing  
776 standards as barriers that impeded their ability to provide genetics education. See Appendix B-5, Table 3  
777 for percentages of organizations that selected each barrier.  
778

779 ***Membership Needs and Priorities for Genetics Education***  
780

781 More than half of organizations either have directly surveyed or received indirect input from their  
782 membership about genetics education needs or priorities. Genetic-specific organizations survey routinely,  
783 while other organizations obtain input on a more ad hoc basis, such as from educational meeting  
784 evaluations and/or general needs assessments. NCHPEG noted that although there are efforts within some  
785 of its member organizations (i.e., nurses and PAs) to focus attention and educational efforts on genetics  
786 through competencies, this emphasis is lacking in some of its other member organizations. As noted  
787 earlier in this chapter, NCHPEG recently revised its Core Competencies for All Health Care  
788 Professionals, streamlining them based on the experiences of its membership (see Appendix A-2 for a  
789 listing of these competencies). It also is poised to release Core Competencies in Family History.  
790 Professional organizations have, in the past, used these resources to structure their own specific  
791 competencies (e.g., public health professionals and nurses).  
792

793 ***How to Engage Members in Genetics Education***

794  
795 Organizations were asked what types of programs or resources could enhance the engagement of their  
796 organization's members in genetics education and if there are programmatic needs that could be  
797 addressed by the Federal Government.

798  
799 The need for funding in the form of educational grants for faculty training and program development, and  
800 development of point-of-care tools and tool kits were common themes. Respondents suggested that  
801 Federal support of research and dissemination of evidence-based guidelines would help engage their  
802 members' interest in additional genetics topics. Respondents also reported that increased integration of  
803 genetics into clinical decision support, electronic medical records, and performance standards would  
804 improve member engagement. In addition, a registry of genetic tests would facilitate the evaluation of  
805 clinical validity and utility and thus inform genetic test usage in the clinical setting.

806  
807 **Information Relevant to Organizations' Missions**

808  
809 Organizations were asked to answer open-ended questions in one of four categories most relevant to their  
810 mission. Of the thirty-three organizations responding, 18 indicated their most relevant mission as  
811 education and training of health professionals; 10 as advocacy and support of practicing professionals;  
812 two as certification of health professionals, and three as accreditation or certification of institutions.  
813 Selected comments for each category are provided below.

814  
815 **Education and Training of Health Professionals**

816  
817 ***Integration of Genetics into the Curriculum and Training of Health Professionals***

818  
819 Organizations were asked to characterize the need for integrating genetics into the curriculum and training  
820 of health professionals. Most organizations felt that this is a critical need and several have already  
821 implemented curricula nationally. Several organizations indicated, however, that this effort was not a high  
822 priority and one suggested uncertain clinical benefit of this approach. The following quote from  
823 NCHPEG articulates the need and challenges faced by organizations.

824  
825 "The sheer volume of new information now at the disposal of biomedical researchers and health  
826 care providers is transforming our understanding of disease processes – including those of  
827 common, chronic diseases such as cancer, diabetes, and mental illness – and is changing the  
828 delivery of health care. Increasingly, health care providers – regardless of specialty, role, or  
829 practice setting – will face questions about the implications of genetics and genomics for their  
830 patients. And yet, the rapid pace of the science and the relative paucity of professional training in  
831 genetics continue to leave many clinicians without satisfactory answers to genetic questions from  
832 their patients. A prime example is the large number of genome-wide association studies that are  
833 finding genetic associations with a vast array of phenotypes. Some of this information is making  
834 its way into clinical care through direct-to-consumer marketing. Many health care professionals  
835 will be at a loss to interpret this information correctly, let alone determine whether management  
836 should be approached differently. While there are a number of ongoing and proposed efforts to  
837 help facilitate the appropriate translation of genomic information into the clinic, currently  
838 practicing health professionals would benefit from a greater understanding of the benefits and  
839 limitations of genetic information in the context of complex diseases."

840

841 ***Development of Curricular Components***

842  
843 Organizations whose mission is to educate and train health professionals were asked whether they assist  
844 member organizations in developing curriculum components related to genetics. Although some  
845 organizations do not, others assisted with curriculum components used nationally. Individual responses  
846 can be found in Appendix B-4, Table 4.

847  
848 ***Cultural Competency Incorporated into Curricula***

849  
850 Cultural competency related to genetics education of health professionals was identified as an urgent need  
851 by health professional organizations through a roundtable discussion and a survey conducted by  
852 SACGHS in 2004.<sup>172</sup> To ascertain whether steps were taken by professional organizations to incorporate  
853 cultural competency into curricula, this question was asked of 18 organizations that delineate education  
854 and training of health professionals as their primary mission. Twelve of 13 responding organizations  
855 stated that cultural competency is part of the curricula or is an accreditation requirement.

856  
857 ***Future Needs in Genetics Education***

858  
859 Organizations that identified education and training of health professionals as most relevant to their  
860 mission were asked to look ahead 5 to 10 years and indicate anticipated needs in genetics education.  
861 Responses were varied and ranged from the need to be able to interpret genetic test results and know  
862 when to refer patients, to more general statements about the need for health care providers to be  
863 knowledgeable about genetic topics. The need to understand risks for complex diseases was mentioned as  
864 was the need to be able to assess risks using multiple tools, change management based on risk, and  
865 communicate risk effectively. One organization felt that there is no end in sight to the need for education  
866 in genetics, while another stated that genetics will be a part of mainstream education and clinical practice  
867 in 5 to 10 years.

868  
869 ***Advocacy and Support of Practicing Health Professionals***

870  
871 ***Continuing Education Programs and Activities Related to Genetics***

872  
873 Among the 10 organizations who identified their primary role as advocacy and support of practicing  
874 health professionals, the American Academy of Family Physicians responded that it surveyed members in  
875 2003 regarding CE programs and found that the top two CE genetics topics requested by members were  
876 common genetic diseases and genetic testing and counseling. This organization has a subcommittee on  
877 genomics that has also identified that members would benefit from more information regarding  
878 pharmacogenomics.

879  
880 ***Promoting Greater Knowledge of Genetics***

881  
882 When asked what would help to promote a greater knowledge of genetics, the following responses were  
883 provided from six organizations that identified their mission as advocacy and support of practicing  
884 professionals:

- 885  
886
  - Additional CE opportunities, learning communities, and resource portals.

---

<sup>172</sup> Secretary's Advisory Committee on Genetics, Health, and Society. (2004). *Resolution of the Secretary's Advisory Committee on Genetics, Health, and Society on Genetics Education and Training of Health Professionals*. See [http://oba.od.nih.gov/SACGHS/sacghs\\_documents.html](http://oba.od.nih.gov/SACGHS/sacghs_documents.html). Accessed on December 9, 2009.

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

903

#### 904 **Certification of Health Professionals**

905

#### 906 *Credentialing Exams*

907

908 Organizations were asked if current credentialing exams include questions on genetics. Four genetic-  
909 specific organizations (American Board of Genetic Counseling (ABGC), American Board of Medical  
910 Genetics, American College of Medical Genetics, and Genetic Nursing Credentialing Commission)  
911 reported that all or most of their credentialing exams were on genetic content. One nursing certification  
912 organization reported genetic content but at less than 1 percent of total content and one general  
913 professional organization reported genetic content at less than 5 percent of total content.

914

#### 915 **Accreditation or Certification of Institutions**

916

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

921

#### 922 **SACGHS Surveys of Health Professional Organizations: Comparison of 2004 and 2008 Surveys**

923

924 In 2004, 26 organizations were invited via e-mail to respond to a survey that consisted of seven open-  
925 ended questions. These organizations were divided into three groups based on their primary role as either  
926 genetic specific, professional education, or general professional organizations. The results of the survey  
927 were reported to SACGHS on June 14, 2004.<sup>173</sup> Thirteen responses were received (50 percent). See  
928 Appendix A-5 for a list of the organizations who responded.

929

930 The 2004 survey was considerably shorter than the 2008 survey; however, there are several areas where  
931 comparisons can be made. Due to the small number of responses to specific questions in 2004,  
932 generalizing more broadly beyond the specific organizations is not possible.

933

---

<sup>173</sup> Secretary's Advisory Committee on Genetics, Health, and Society. *Presentation on Information Gathered on Efforts in Genetics Education and Training*. See [http://oba.od.nih.gov/SACGHS/sacghs\\_past\\_meeting\\_2004\\_jun\\_14.html](http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_2004_jun_14.html). Accessed on February 22, 2010.

934 ***Integration of Genetics into the Curriculum and Training of Health Professionals***  
935

936 Both surveys asked organizations that identify education of professionals as their primary mission to  
937 characterize the need for integrating genetics into the curriculum and training of health professionals. In  
938 2004, eight organizations responded that while the need for integration varies, health professionals must  
939 be able to address patient questions and thus require a solid, basic knowledge of genetics with a lifelong  
940 commitment to learning. By 2008, 15 of the 17 organizations responding to this question felt similarly  
941 and several have actually implemented genetic curriculum nationally. However, in 2008, several  
942 organizations felt that this effort was not a high priority, and one suggested uncertain clinical benefit of  
943 integrating genetics into the curriculum and training of health professionals.  
944

945 ***Barriers to Providing Genetics Educational Activities***  
946

947 In 2008, 64 percent of organizations cited competing priorities as a barrier to providing genetics  
948 educational activities, followed by 28 percent citing genetics and genomics not emphasized on certifying  
949 exams or credentialing standards. Only 8 percent cited lack of evidence supporting clinical effectiveness  
950 of care based on genetic or genomic information. Again, the numbers of responding organizations in  
951 2004 were small; however, similar barriers were noted: 57 percent reported competing priorities; 43  
952 percent reported lack of evidence for clinical application of genetics; and 43 percent identified lack of  
953 prepared faculty. While competing priorities remains a significant barrier to providing genetic and  
954 genomic educational activities, organizations were much less likely to report lack of evidence supporting  
955 clinical effectiveness in 2008.  
956

957 ***Themes Common to Both Surveys Regarding Future Directions in Genetic Education of Health Care***  
958 ***Professionals***  
959

- 960 • The government has a role in supporting genetics education programs.
- 961 • Genetics education must be represented throughout the entire continuum of medical education.
- 962 • Funding should target educational programs that are known to change clinician behavior and  
963 should include interactive learning with case studies that emphasize clinical application of  
964 genetics.
- 965 • Education and training should address the importance of obtaining family history.
- 966 • There is a need to expand cultural diversity within the health professional workforce and to  
967 improve the cultural competency of health professionals in genetics and genomics.  
968

969 **D. Summary**  
970

971 A significant body of literature from the United States and abroad highlights the nature and lack of  
972 genetics education of health care professionals as factors limiting integration of genetics into health care.  
973 Genetics content is often minimal in health education programs, focused primarily on single-gene  
974 disorders, and not associated with long-term knowledge retention for clinical application.  
975

976 A recent study examining the impact of a genetic outreach education initiative found that although health  
977 care providers felt more confident in using genetics after a CE intervention, many applied their new  
978 knowledge incorrectly. Incorrect application of genetics occurred in the misappropriation of risk  
979 estimation; approximately half of those receiving the genetics education intervention issued a high-risk  
980 categorization to a low-risk breast cancer presentation.  
981

982 Lack of genetic knowledge among health care providers exists not only with complex, multifactorial  
983 conditions—but also with traditional and well-documented Mendelian conditions such as autosomal

984 dominant hereditary cancer syndromes. Analyses of genetic content in formal medical and health care  
985 curricula (with the exception of genetic counseling programs) find great variability in the content and  
986 quantity of coursework in genetics. The same variability and levels of insufficiency can be found in  
987 licensing and accreditation requirements. To address some these deficiencies, several professional groups,  
988 particularly those representing PAs and nurses, have included genetics knowledge in their competencies  
989 and guidelines and offer CE in genetic content.

990  
991 The 2008 SACGHS survey data found that overall, 70 percent of health professional organizations  
992 responding to the survey view genetics education and training as part of their role or responsibility. Most  
993 of these organizations reported that they were able to fulfill this role or responsibility; however, funding,  
994 program evaluation, and increasing interest within the organization's leadership would allow them to  
995 meet this role or responsibility more effectively. Additionally, Federal support of research and  
996 dissemination of evidence-based guidelines would help engage their members' interest in additional  
997 genetic topics.

998  
999 Only half of survey respondents reported that they had received input from their membership regarding  
1000 educational needs and priorities in genetics, but those that did were able to provide numerous examples of  
1001 how they obtained this feedback. Open-ended survey questions yielded information on ways to engage  
1002 members, integrate genetics into curriculum and training, develop curricular components, incorporate  
1003 cultural competency into curricula, advocate for practicing health professionals, and develop CE  
1004 programs and activities. Despite this relative interest in genetics education, only nine organizations  
1005 reported having published position statements or practice competencies regarding genetics.

1006  
1007 Although health professional organizations across the board reported that developing and promoting  
1008 educational activities related to genetics is important, these topics are not a high priority relative to the  
1009 overall priorities facing the organizations. Nongenetic-specific organizations reported only moderate  
1010 proficiency in and comfort of their leadership in genetics education, and less than half of these  
1011 organizations have dedicated entities specifically focused on genetic topics relevant to their mission. The  
1012 majority of all organizations surveyed identified competing priorities as a barrier to their ability to  
1013 provide genetics education.

1014  
1015 The 2008 SACGHS survey of health professional organizations provided data that supports findings from  
1016 the literature review yet revealed new insights into how professional organizations are currently  
1017 approaching the need to educate their members and constituencies in genetics and genomics. Competing  
1018 priorities in an already crowded curriculum was the most commonly mentioned barrier to improving  
1019 genetic literacy of health care professionals. In addition, respondents cited lack of sufficient resources,  
1020 financial and otherwise, as a barrier to developing or accessing appropriate education and training  
1021 opportunities for members. In response to the growing need for proficiency and competency,  
1022 organizations offer tailored CE programs or include innovative programs that reflect emerging genetic  
1023 content in their annual meeting agendas. Thus, the need for educational efforts to increase the use of  
1024 genetic and genomic information in clinical care is widely recognized and acknowledged. The SACGHS  
1025 survey, however, highlighted the challenges facing many organizations attempting to fill the gap for their  
1026 constituencies against a backdrop of competing demands and limited resources.

1027  
1028 Based on a literature review and its survey findings, SACGHS found evidence that suggests inadequate  
1029 education of health care professionals is a significant factor that limits the integration of genetics into  
1030 clinical care. Enhancing the use of clinical decision support tools, promoting the importance of family  
1031 history, and ensuring adequate reimbursement for genetic services are among the approaches that would  
1032 support the optimal use of genetics and genomics in health care.

### 1033 **III. The Status of Education and Training of Public Health Providers**

1034

#### 1035 **A. Introduction**

1036

1037 Genetics has been at the center of a number of public health programs for decades. Most state health  
 1038 departments administer newborn genetic screening and other genetic disease prevention programs focused  
 1039 primarily on diseases related to maternal and child health. Some state health departments include genetics  
 1040 coordinators and frequently consult genetics professionals.<sup>174</sup> However, expertise and focus has, in general,  
 1041 been limited to the maternal and child health field. In contrast, a more expansive view of “public health  
 1042 genomics” focuses on the effective and responsible translation of genomics to improve population  
 1043 health.<sup>175</sup> Public health genomics is defined by Khoury et al. as seeking “to use population-based data on  
 1044 genetic variation and gene-environment interactions to develop, implement, and evaluate evidence-based  
 1045 tools for improving health and preventing disease. It also applies systematic evidence-based assessments  
 1046 of genomic applications in health practice and works to ensure the delivery of validated, useful genomic  
 1047 tools for the benefit of population health.”<sup>176</sup>

1048

1049 This chapter briefly describes the role of the public health workforce in population health, the range of  
 1050 pathways to a public health career, accreditation of schools of public health, training opportunities, and  
 1051 reviews what is available in the literature regarding the readiness of the public health workforce to engage  
 1052 in genetics and genomics. In addition, SACGHS developed 12 competencies, derived from competencies  
 1053 developed by public health organizations and institutions, to use in an online survey instrument assessing  
 1054 public health providers’ genetic and genomic training and education needs. The results of that survey are  
 1055 reported here.

1056

#### 1057 **The Public Health Workforce**

1058

1059 In contrast to clinicians, who focus on the needs of individuals, public health practitioners assess the needs  
 1060 of populations to determine the burden of disease, develop policies, and assure that appropriate services are  
 1061 available to individuals, families, and communities. A landmark 1988 report by the Institute of Medicine  
 1062 (IOM)<sup>177</sup> defined public health as the collection of society’s efforts to achieve conditions in which people  
 1063 can be healthy. The IOM report further defined a public health professional as any professional who  
 1064 approaches health from a population lens.<sup>178</sup> Public health providers can work across various sectors,  
 1065 including Federal and state government, academia, professional, community and lay organizations, and  
 1066 the private sector. They work in various population health domains such as epidemiology, biostatistics,  
 1067 environmental health, health promotion and maternal and child health. The public health community has  
 1068 the unique skills and networks potentially to raise the level of general genomic literacy and develop  
 1069 targeted messages about the use of genetic information for disease prevention and health promotion.<sup>179</sup> In

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<sup>174</sup> Piper, M.A., Lindenmayer, J.M., Lengerich, E.J., Pass, K.A., Brown, W.G., Crowder, W.B., Khoury, M.J., Baker, T.G., Lloyd-Puryear, M.A., and Bryan, J.L. (2001). The role of state public health agencies in genetics and disease prevention: results of a national survey. *Public Health Reports*. 116:22-31.

<sup>175</sup> Khoury, M.J., Bowen, S., Bradley, L.A., Coates, R., Dowling, N.F., Gwinn, M., Kolor, K., Moore, C.A., St. Pierre, J., Valdez, R., and Yoon, P.W. (2008). A decade of public health genomics in the United States: Centers for Disease Control and Prevention 1997-2007. *Public Health Genomics*. 12:20-29.

<sup>176</sup> Khoury, M.J., Bowen, S., Bradley, L.A., Coates, R., Dowling, N.F., Gwinn, M., Kolor, K., Moore, C.A., St. Pierre, J., Valdez, R., and Yoon, P.W. (2008). A decade of public health genomics in the United States: Centers for Disease Control and Prevention 1997-2007. *Public Health Genomics*. 12:20-29.

<sup>177</sup> Institute of Medicine. *The Future of Public Health*. Washington, D.C.: National Academies Press, 1988.

<sup>178</sup> Institute of Medicine. *Who will keep the public healthy?* Washington, D.C.: National Academies Press, 2003.

<sup>179</sup> Beskow, L.M., Khoury, M.J., Baker, T.G., and Thrasher, JF. (2001). The integration of genomics into public health research, policy and practice in the United States. *Community Genetics*. 4:2-11.

1070 addition, the public health community has a large research infrastructure sorely needed by genomics, for  
 1071 example its surveillance and data collection systems.

1072  
 1073 The diversity of settings and service provision and the lack of specific licensure that would otherwise  
 1074 facilitate counting and studying the public health workforce<sup>180</sup> create an inherent problem in targeting  
 1075 genetic and genomic educational efforts. In 2000, the public health workforce was estimated to consist of  
 1076 448,254 persons in salaried positions, supplemented by 2.9 million volunteers.<sup>181</sup> In this estimate, 44  
 1077 percent of workers were identified as professionals (e.g., physicians, nurses, laboratorians, dentists, health  
 1078 educators) but 24 percent of the workers could not be categorized. It was estimated that over half of all  
 1079 public health workers have at least a college education. Since 2000, numerous reports have attempted to  
 1080 count and categorize various levels of the public health workforce,<sup>182,183</sup> which is considered to be a  
 1081 critical step to assuring that the workforce is sufficiently large and skilled to deliver essential public  
 1082 health services to the U.S. population.<sup>184</sup> However, availability of this data and sufficient resources to  
 1083 support research of the public health workforce are lacking.<sup>185</sup>

1084  
 1085 **Schools of Public Health**

1086  
 1087 The Council on Education for Public Health (CEPH) is an independent agency recognized by the U.S.  
 1088 Department of Education to accredit schools of public health and community health and preventative  
 1089 medicine programs. These schools and programs prepare students for entry into public health careers and  
 1090 offer Master of Public Health (M.P.H.), Doctor of Public Health (Dr.P.H.), and Master of Health Care  
 1091 Administration (M.H.A.) degrees.<sup>186</sup> The Association of Schools of Public Health (ASPH)<sup>187</sup> represents  
 1092 the 43 CEPH-accredited schools of public health and the eight Associate Member Schools that are  
 1093 working on accreditation. However, other programs exist which are not members of ASPH and, moreover  
 1094 are not necessarily CEPH-accredited. ASPH also supports graduate internships and fellowships to provide  
 1095 practice experiences. The National Public Health Training Centers Network, funded through the Health  
 1096 Resources Services Administration, has partnered with schools of public health, related academic  
 1097 institutions, and public health agencies and organizations to assess learning needs and provide training to  
 1098 meet those needs.<sup>188</sup> Their distance education center lists several genetics and genomics courses offered  
 1099 by partner organizations.<sup>189</sup>

1100

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<sup>180</sup> Health Resources and Services Administration. (2005). *Public Health Workforce*. See <http://bhpr.hrsa.gov/healthworkforce/reports/publichealth/default.htm>. Accessed on December 13, 2009.

<sup>181</sup> Bureau of Health Professionals National Center for Health Workforce Information and Analysis. (2000). *The Public Health Work Force Enumeration 2000*. See [www.uic.edu/sph/prepare/courses/chsc400/resources/phworkforce2000.pdf](http://www.uic.edu/sph/prepare/courses/chsc400/resources/phworkforce2000.pdf). Accessed on December 13, 2009.

<sup>182</sup> Health Resources and Services Administration (2005). *Public Health Workforce*. See <http://bhpr.hrsa.gov/healthworkforce/reports/publichealth/default.htm>. Accessed on December 13, 2009.

<sup>183</sup> The Center for Health Workforce Studies, School of Public Health, University at Albany. (2008). Enumeration of the Local Public Health Workforce in New York: 2006. See [www.albany.edu/news/pdf\\_files/0802\\_Local\\_Public\\_Health\\_Workfore\\_NY\\_2006.pdf](http://www.albany.edu/news/pdf_files/0802_Local_Public_Health_Workfore_NY_2006.pdf). Accessed on December 13, 2009.

<sup>184</sup> Gebbie, K.M., Raziano, A.R., and Elliot, S. (2009). Public health workforce enumeration. *American Journal of Public Health*. 99(5):786-787.

<sup>185</sup> Moore, J. (2009). Studying an ill-defined workforce: public health workforce research. *Journal of Public Health Management Practice*. 15(6 Suppl):S48-53.

<sup>186</sup> Council on Education for Public Health. See <http://www.ceph.org/i4a/pages/index.cfm?pageid=1>. Accessed on December 14, 2009.

<sup>187</sup> Association of Schools of Public Health. See <http://www.asph.org/document.cfm?page=200>. Accessed on December 14, 2009.

<sup>188</sup> National Public Health Training Centers Network. See <http://www.asph.org/document.cfm?page=780>. Accessed on December 14, 2009.

<sup>189</sup> National Public Health Training Centers Network Distance Programs. See <http://www.asph.org/document.cfm?page=718>. Accessed on December 14, 2009.

1101 **B. Literature Review**

1102  
1103 **Barriers to Achieving a Genomics Informed Public Health Workforce**

1104  
1105 The current public health workforce faces challenges receiving and assimilating genetic and genomic  
1106 information. Individualized primary prevention and early detection (often the purview of primary care)  
1107 intersects with the realm of population health (the purview of public health). Khoury et al. has expressed  
1108 concern that without a more integrated approach between primary care and public health, genomics could  
1109 easily widen the schism that has long existed between medicine and public health.<sup>190</sup>

1110  
1111 The barriers to achieving a more genomics informed public health workforce are multifaceted. First, the  
1112 public health workforce is diverse and follows many educational and training paths, including a variety of  
1113 professionals with formal training and certifications, volunteers, and community (lay) health workers.  
1114 Thus, a one-size-fits-all approach is not feasible. Second, many providers in the field today received their  
1115 formal education before genomics became a critical aspect of medicine and health. Third, attitudes,  
1116 perceptions, and beliefs shape acceptance and adoption of genomics by the public health community.

1117  
1118 Khoury et al.<sup>191</sup> have noted some of the attitudinal barriers to acceptance of genetics and genomics by the  
1119 public health community to include skepticism about genomics and genomics research being seen as a  
1120 low-yield investment and low priority because of other more important preventative or modifiable  
1121 environmental causes of morbidity and mortality. For many public health providers, local issues, national  
1122 and international pandemics, and environmental causes of morbidity and mortality are more important  
1123 priorities than genetics and genomics, particularly in the context of limited public health funding.  
1124 Research also highlights that public health providers do not perceive public health genomics to be part of  
1125 their job, nor a professional priority.

1126  
1127 Public health educators perceived barriers include not only lack of knowledge regarding the link between  
1128 genomics and health promotion, but also lack of current basic genomic knowledge. Thus, future education  
1129 and training of public health providers focusing primarily on basic genomic content will be inadequate.<sup>192</sup>  
1130 To address the place of genetics in public health practice, Chen et al. assessed U.S. public health  
1131 educators’ attitudes toward genomic competencies, evaluated their awareness of efforts in the field to  
1132 promote and incorporate genomics, and attempted to gauge their basic and applied genomic knowledge.  
1133 While most public health providers agreed with the Centers for Disease Control and Prevention (CDC)-  
1134 proposed competencies, incorporating them into public health practice was viewed as important by less than  
1135 half of the study participants.<sup>193</sup> Subsequent work by Chen et al. found that public health providers are  
1136 reluctant to adopt genomic competencies into health promotion—only 35 percent of survey respondents  
1137 said they were willing to integrate genomic components into community-based genomic education  
1138 programs, suggesting that health educators are not ready for their professional role in genomics,<sup>194</sup> and  
1139 only half of basic and applied genomic knowledge questions were answered correctly. The study authors  
1140 concluded that “the simplest and most immediate explanation for such a gap is that the majority of training

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<sup>190</sup> Khoury, M.J., Gwinn, M., Burke, W., Bowen, S., and Zimmern, R. (2007). Will genomics widen or help heal the schism between medicine and public health? *American Journal of Preventive Medicine*. 33(4):310-317.

<sup>191</sup> Khoury, M.J., Gwinn, M., Burke, W., Bowen, S., and Zimmern, R. (2007). Will genomics widen or help heal the schism between medicine and public health? *American Journal of Preventive Medicine*. 33(4):310-317.

<sup>192</sup> Chen, L-S., and Goodson, P. (2009). Barriers to adopting genomics into public health education: a mixed methods study. *Genetics in Medicine*. 11(2):104-110. P. 109.

<sup>193</sup> Chen, L-S., and Goodson, P. (2007). Public health genomics knowledge and attitudes: a survey of public health educators in the United States. *Genetics in Medicine*. 9(8):496-503.

<sup>194</sup> Chen L-S., Kwok, O.M., and Goodson P. (2008). U.S. health educators’ likelihood of adopting genomic competencies into health promotion. *American Journal of Public Health*. 98(9):1651-1657.

1141 programs in health education and public health include neither genetics nor genomics in their curriculum nor  
 1142 do they require course offerings in these topics for accreditation purposes.”<sup>195</sup>

1143  
 1144 Finally, lack of evidence might be a significant barrier to public health adoption of genomic  
 1145 competencies. Until evidence of health benefit can be shown (e.g., population screening for *BRCA1* and  
 1146 *BRCA2* mutations and hereditary hemochromatosis), public health providers might be resistant to  
 1147 adoption. Thus, public health genomics will “hit a translation roadblock if no investments are made in  
 1148 evaluating the best methods for assuring delivery and monitoring safety and effectiveness of gene-based  
 1149 interventions, whether they are population screening programs, such as newborn screening, or early case  
 1150 detection and interventions delivered by clinicians.”<sup>196</sup> Thus, one approach to educating the public health  
 1151 workforce is to have clear examples of beneficial applications of genomics at the population level, which  
 1152 can be built on as new evidence arises.

1153  
 1154 Further confounding educational efforts for public health providers is the proportion of non-professional,  
 1155 community health workers in the public health work force. The use of lay health providers creates an  
 1156 additional barrier to achieving widespread genetic and genomic literacy in public health programs.  
 1157 Community health workers (CHWs) are, by definition, “any health worker carrying out functions related  
 1158 to health care delivery; trained in some way in the context of the intervention; [but] having no formal  
 1159 professional or paraprofessional certificated or degreed tertiary education.”<sup>197</sup> Although Texas, Ohio,  
 1160 Indiana, and Alaska require some level of certification for CHWs and several states are considering  
 1161 implementing certification requirements,<sup>198</sup> most states do not, and there were until 2007, no national  
 1162 standards for certifying or training non-professional public health workers.<sup>199,200,201</sup>

1163  
 1164 The Center for Sustainable Health Outreach (CSHO) issued a report in 2002 that listed CHW programs  
 1165 that offer credit, certificates, or degrees at institutions of higher education. At that time, there were 15  
 1166 programs in 10 states that offered courses, certificates, and/or degrees for generalist CHWs.<sup>202</sup> The  
 1167 majority of the programs were offered at community or junior colleges and led to certificates, rather than  
 1168 Baccalaureate degrees. Even where programs exist for formal training of CHWs, the emphasis is on  
 1169 communication skills (including bilinguality), service coordination skills, advocacy skills and “a  
 1170 knowledge base on specific health issues.”<sup>203</sup>

1171

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<sup>195</sup> Chen L-S., and Goodson P. (2007). Public health genomics knowledge and attitudes: a survey of public health educators in the United States. *Genetics in Medicine*. 9(8):496-503. P. 501.

<sup>196</sup> Khoury, M.J., Gwinn, M., Burke, W., Bowen, S., and Zimmern, R. (2007). Will genomics widen or help heal the schism between medicine and public health? *American Journal of Preventive Medicine*. 33(4):314.

<sup>197</sup> Lewin, S.A., Dick, J., Pond, P., Zwarenstein, M., Aja, G., van Wyk, B., Bosch-Capblanch, X., and Patrick, M. (2005). Lay health workers in primary and community health care. *Cochrane Database Systematic Reviews*. Jan 25;(1):CD004015.

<sup>198</sup> May, M.L., Kash, B., and Contreras, R. (2005). Southwest Rural Health Research Center: Community Health Worker (CHW) Certification and Training - A National Survey of Regionally and State-based Programs. U.S. Department of Health and Human Services, Health Services and Resources Administration, Office of Rural Health Policy.

<sup>199</sup> Love, M.B., Gardner, K., and Legion, V. (1997). Community health workers: who they are and what they do. *Health Education and Behavior*. 24(4):510-22.

<sup>200</sup> Love, M.B., Legion, V., Shim, J.K., Tsai, C., Quijano, V., and Davis, C. (2004). CHWs get credit: a 10-year history of the first college-credit certificate for community health workers in the United States. *Health Promotion and Practice*. 5(4):418-28.

<sup>201</sup> May, M.L., Kash, B., and Contreras, R. (2005). Southwest Rural Health Research Center: Community Health Worker (CHW) Certification and Training - A National Survey of Regionally and State-based Programs. U.S. Department of Health and Human Services, Health Services and Resources Administration, Office of Rural Health Policy.

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

<sup>203</sup> Health Resources and Services Administration. (2007). Community Health Worker National Workforce Study. See <http://bhpr.hrsa.gov/healthworkforce/chw/default.htm#preface>. Accessed on December 12, 2009.

1172 One of the most rigorous trainings for CHWs is in Ohio. There, certification training programs are  
 1173 operated under the authority of the Nursing Practices Act and require at least 100 hours of didactic  
 1174 instruction by certified medical professionals and 130 hours of clinical instruction. The three educational  
 1175 programs currently certified in Ohio require coursework in basic anatomy and the physiology of major  
 1176 body systems, medical terminology, health education related to child-bearing, and competencies in the  
 1177 areas of immunization and appropriate referrals to health care facilities and practitioners. Although not  
 1178 ostensibly related to genetics or genomics, the approved curricula could be expanded to require  
 1179 competency and knowledge about genetics and genomics as part of the biology coursework or health  
 1180 education competencies related to childbearing and immunization.

1181  
 1182 **Current Efforts to Improve Proficiencies and Competencies**  
 1183

1184 Several professional groups and CDC have turned their attention to the need for public health provider  
 1185 education and training in genomics. The IOM report recommended genomics as one of eight new content  
 1186 areas to be covered by every school of public health.<sup>204</sup> The American Public Health Association (APHA),  
 1187 representing more than 50,000 health professionals, has published policy statements related to genetics  
 1188 and genomics and the public health workforce. *Genetics and Public Health*,<sup>205</sup> published in 1987,  
 1189 discussed the need for consensus among a wide variety of institutions and organizations regarding the  
 1190 public health implications of genetics and the need for quality genetic services. The need for professional  
 1191 education on advances in genetics was outlined in the objectives and implementation methods that were  
 1192 proposed to achieve these objectives. Recognizing the need for an information infrastructure for resources  
 1193 applicable to public health, the Partners in Information Access for the Public Health Workforce was  
 1194 launched in 1998. This collaborative effort of 11 U.S. government agencies, public health organizations  
 1195 and health sciences libraries provides resources on a variety of topics pertaining to public health  
 1196 genomics.<sup>206</sup> In recognition of the broader scope of genomics and its impact on public health and the  
 1197 critical need for public health workforce education in genomics, APHA published *The Role of Genomics*  
 1198 *in Public Health* in 2002.<sup>207</sup> In 2007, The Genomics Forum was established within APHA to “engage the  
 1199 public health community to promote workforce competency in genomics, including an improved  
 1200 understanding of the relevance and impact of genomics on public health”.<sup>208</sup> The Genomics Forum has  
 1201 developed a policy statement on genetic health literacy for health professionals to be submitted to APHA  
 1202 for review and publication in 2010.<sup>209,210</sup>

1203  
 1204 In August 2000, the CDC Office of Genetics and Disease Prevention and representatives from each  
 1205 of the disciplines in public health met to identify the core competencies necessary for all health  
 1206 professionals to incorporate genetics into public health practice.<sup>211</sup> The group developed specific

<sup>204</sup> Institute of Medicine. *Who will keep the public healthy?* Washington, D.C.: National Academies Press, 2003.

<sup>205</sup> American Public Health Association. *Genetics and Public Health*. Policy Number 8732PP, January 1, 1987. See <http://www.apha.org/advocacy/policy/policysearch/default.htm?id=1161>. Accessed on January 13, 2010.

<sup>206</sup> Partners in Information Access for the Public Health Workforce. See [http://phpartners.org/public\\_health\\_genomics.html](http://phpartners.org/public_health_genomics.html). Accessed on December 14, 2009.

<sup>207</sup> American Public Health Association. *The Role of Genomics in Public Health*. Policy Number 2002-1, November 13, 2002. See <http://www.apha.org/advocacy/policy/policysearch/default.htm?id=275>. Accessed on January 13, 2010.

<sup>208</sup> American Public Health Association. Genomic Forum. See <http://www.apha.org/membergroups/forums/>. Accessed on January 13, 2010.

<sup>209</sup> Payne, E., Honore, H., Platt, J., and the American Public Health Association Genomics Forum Policy Committee. (2009). Genetic health literacy. Poster presented at the meeting of the Health Literacy Forum: Building a Community Network to Improve Health Communication, October, 2009. University of Michigan Health Science Libraries, Ann Arbor, MI.

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

<sup>211</sup> Piper, M.A., Lindenmayer, J.M., Lengerich, E.J., Pass, K.A., Brown, W.G., Crowder, W.B., Khoury, M.J., Baker, T.G., Lloyd-Puryear, M.A., and Bryan, J.L. (2001). The role of state public health agencies in genetics and disease prevention: results of a national survey. *Public Health Reports*. 116:22-31.

1207 genomic competencies for various public health providers. The competencies were developed as a tool  
1208 for public health programs and schools of public health to incorporate genomics into existing  
1209 competencies and program training goals. However, as with any new requirement imposed on an already  
1210 information-laden curriculum, incorporation of competencies in education and certification or licensure  
1211 processes takes time. Encouraging, rather than requiring, that such competencies be demonstrated can further  
1212 slow their adoption. It is possible that various social, organizational, and environmental factors (e.g.,  
1213 certification and licensure requirements) would carry more weight than individuals' attitudes in promoting  
1214 willingness to adopt genomic competencies.

1215  
1216 In addition to convening the working group that developed the core competencies, CDC has made other  
1217 investments in public health genomics practice and education (see also CDC Federal Activities in Chapter  
1218 V). It has funded Centers for Genomics and Public Health in schools of public health at the University of  
1219 North Carolina, the University of Michigan, and the University of Washington. These centers provide  
1220 expertise in translating genomic information into public health knowledge, provide technical assistance to  
1221 state and community public health agencies, and facilitate integration of genomics into programs and  
1222 practice.<sup>212</sup> CDC also has supported genomics programs in four state health departments (Michigan,  
1223 Minnesota, Oregon, and Utah).

1224  
1225 Other states have instituted their own initiatives in public health genomics. For example, in 2009,  
1226 Washington sponsored the Summer Institute in Public Health Genomics: Translating Genomics into  
1227 Policy and Practice. The Oregon Genetics Program aims to integrate genomics into public health practice,  
1228 particularly chronic disease program activities. The Oregon Public Health Division received funding to  
1229 translate genomics applications into health practice, specifically to develop, implement, and evaluate a  
1230 surveillance program to monitor awareness, knowledge, and use among health care providers and the  
1231 public of cancer-related genomic tests and family history. This project will also evaluate disparities  
1232 associated with accessing cancer-related genetic testing and counseling. Illinois public health officials  
1233 conducted a needs assessment and created a state genetics plan in 2006. The Connecticut Department of  
1234 Public Health Genomics Office has produced a fact sheet for consumers on DTC personal genomic  
1235 services, highlighting the types of activities that can be accomplished in the public health arena.<sup>213</sup>  
1236

## 1237 **C. SACGHS Survey of Public Health Providers**

### 1238 **Methodology**

1239  
1240  
1241 To assess the genetics education needs of public health providers, SACGHS developed 12 competencies  
1242 that were derived from competencies found in public health organizations and institutions. The 12  
1243 competencies were used in an online survey instrument assessing public health providers' genetic and  
1244 genomic training and education needs. The online survey was based on the work of Kirk et al.<sup>214</sup> and  
1245 modified by SACGHS members and staff. The survey was determined to be exempt from the need for  
1246 Institutional Review Board review and approval by the NIH Office of Human Subjects Research. The  
1247 survey was distributed to around 500 public health providers with varying degrees of genetics  
1248 responsibilities and 140 responses were received and analyzed. See Appendix C-1 for details of the  
1249 survey methodology and participant recruitment. Appendix C-2 provides screen shots of the online survey  
1250 instrument and Appendix C-3 explains survey reliability results.

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<sup>212</sup> Khoury, M.J., Bowen, S., Bradley, L.A., Coates, R., Dowling, N.F., Gwinn, M., Kolor, K., Moore, C.A., St. Pierre, J., Valdez, R., and Yoon, P.W. (2008). A decade of public health genomics in the United States: Centers for Disease Control and Prevention 1997-2007. *Public Health Genomics*. 12:20-29.

<sup>213</sup> Connecticut Department of Public Health Genomics. *Direct-to-Consumer Personal Genomic Services Information to Consider*. See [http://www.ct.gov/dph/lib/dph/state\\_health\\_planning/pdf/dtc\\_10\\_27\\_09.pdf](http://www.ct.gov/dph/lib/dph/state_health_planning/pdf/dtc_10_27_09.pdf). Accessed on December 12, 2009.

<sup>214</sup> Kirk, M., Tolkin, E., and Birmingham, K. (2007). Working with publishers: a novel approach to ascertaining practitioners' needs in genetics education. *Journal of Nursing Research*. 12:597-615.

1251 **Limitations**

1252  
1253 This survey has a number of limitations that affect the ability to generalize the findings. The sample was  
1254 one of convenience and relied on snowball sampling<sup>215</sup> to increase the number and scope of participants.  
1255 Given the need to keep the survey anonymous, it was not possible to obtain informative data about the  
1256 survey participants.

1257  
1258 By targeting dissemination of the survey to individuals more likely to incorporate genetics into their daily  
1259 practice (e.g., state genetic coordinators), the data are unlikely to be representative of the opinions and  
1260 activities of the entire public health workforce. Based on the responsibilities of the individuals to whom  
1261 the survey was sent directly, the data are more likely to represent the "best case scenario", meaning that  
1262 the responses are more strongly supportive of the importance of genetics and the relevance of the  
1263 competencies than might be expected from the public health workforce as a whole. Because of the  
1264 snowball sampling methodology of dissemination, it was not possible to determine if the sample was  
1265 representative of public health workers. Even if it were possible to disseminate a survey to all public  
1266 health workers, individuals using genetics in their jobs would be more likely to participate.

1267  
1268 The competencies that formed the basis of this assessment of education and training needs were derived  
1269 from existing sources through an expert opinion process and were not independently validated.

1270  
1271 The data and their interpretation are also limited by the self-assessment nature of this survey. There is no  
1272 objective measure that can be used to determine the accuracy of the self-assessment.

1273  
1274 **Twelve Competencies Used in the SACGHS Survey**

1275  
1276 The following 12 competencies based on skills and knowledge thought to be critical for practicing  
1277 providers of public health, whether at the local, state, or national level, were used in the survey:

- 1278
- 1279 1. Maintain up-to-date knowledge on the development of genomic science and technologies within  
1280 his or her professional field and program to apply genomics as a tool for achieving public health  
1281 goals.
  - 1282 2. Demonstrate basic knowledge of the role that genetics and genomics plays in the development of  
1283 disease and in screening and interventions for programs of disease prevention and health  
1284 promotion.
  - 1285 3. Describe the importance of family history in assessing predisposition to disease.
  - 1286 4. Identify opportunities and integrate genetic and genomic issues into public health practice,  
1287 policies or programs effectively.
  - 1288 5. Maintain up-to-date knowledge of genetics and genomics-related policies, legislation, statutes,  
1289 and regulations.
  - 1290 6. Describe the potential physical and psychological benefits, limitations, and risks of genetic and  
1291 genomic information for individuals, family members, and communities.
  - 1292 7. Collaborate with existing and emerging health agencies and organizations, academic, research,  
1293 private and commercial enterprises, and community partnerships to apply genetics and genomics  
1294 knowledge and tools to address public health problems.
  - 1295 8. Identify the resources available to assist clients seeking genetic and genomic information or  
1296 services, including the types of genetics professionals available.
  - 1297 9. Conduct outcomes evaluation of available genetic and genomic programs and services to  
1298 determine their effectiveness.

---

<sup>215</sup> van Meter, K.M. Methodological and design issues: techniques for assessing the representatives of snowball samples. (1990). *NIDA Research Monograph*. 98:31-43.

- 1299 10. Identify the political, legal, social, ethical, and economic issues associated with integrating  
1300 genomics into public health.  
1301 11. Use information technology (IT) to obtain credible, current information about genetics; to utilize  
1302 IT skills to share data and participate in research, program planning, evaluation, and policy  
1303 development for health promotion and disease prevention.  
1304 12. Identify appropriate and relevant genetics research findings that can be translated into public  
1305 health policies or practices.  
1306

1307 Survey participants were asked to rank the competencies based on the importance of the competency, how  
1308 confident they are in demonstrating the competency, and how frequently they apply the competency.  
1309

## 1310 **Survey Findings**

### 1311 1312 *Perception of the Importance of the Competencies*

1313  
1314 Overall, little variability was found among the responses regarding the importance of each competency.  
1315 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  
1316 Appendix C-4, Table 1 for summary data). The majority of individuals responded that all of the  
1317 competencies are important.  
1318

1319 The three competencies ranked most important to public health providers were: (1) demonstrating basic  
1320 knowledge of the role of genetics and genomics in development of disease; (2) describing the importance  
1321 of family history in assessing predisposition to disease; and (3) identifying opportunities and effectively  
1322 integrating genetic and genomic issues into public health practice, policies or programs.  
1323

1324 No single item was ranked low, thus there is no reason to conclude that any specific competency is not  
1325 important to public health providers.  
1326

### 1327 *Level of Confidence in Demonstrating the Competencies*

1328  
1329 Responses varied with regard to level of confidence in demonstrating the competencies. However, two of  
1330 the competencies that ranked most important were also those in which respondents could demonstrate the  
1331 most confidence, i.e., the importance of family history and basic knowledge of the role of genetics and  
1332 genomics. In addition, respondents felt competent to describe the potential physical and psychological  
1333 benefits, limitations, and risks of genetic and genomic information for individuals, family members, and  
1334 communities.  
1335

1336 The lowest mean ranked competencies were: (1) maintaining up-to-date knowledge of genetics and  
1337 genomics-related policies, legislation, statutes, and regulations; (2) using information technology (IT) to  
1338 obtain credible, current information about genetics; and (3) conducting outcomes evaluation of available  
1339 genetic and genomic services to determine their effectiveness.  
1340

### 1341 *Frequency of Application of Competencies*

1342  
1343 Responses were varied for how frequently the competencies are applied with mean values ranging from  
1344 2.0 to 3.3. It appears that depending on the competency, there are instances where public health providers  
1345 never or rarely apply a specific competency or conversely they very frequently apply a specific  
1346 competency. Demonstrating basic knowledge of the role of genetics and genomics in the development of  
1347 disease and maintaining up-to-date knowledge on the development of genomic science and technologies  
1348 within his or her professional field were reported to be most frequently applied by public health providers.  
1349

1350 The majority of the public health providers apply these two competencies monthly or weekly. The lowest  
1351 mean ranked competency is conducting outcome evaluation of available genetic and genomic programs  
1352 and services to determine their effectiveness. The majority of public health practitioners either never  
1353 apply this competency or they apply it rarely (1-2 times per year). Overall, when considering the  
1354 importance of these competencies to public health providers, there appears to be no competency that  
1355 stands out as unimportant or irrelevant to these survey respondents.

1356  
1357 ***The Importance of Genetics and Genomics to Institution Leadership***  
1358

1359 Respondents were asked how important knowledge and experience in genetics and genomics is to their  
1360 roles and responsibilities from their senior administration's perspective. Sixty percent of survey  
1361 respondents reported that their senior administration feels that genetics and genomics is important or very  
1362 important to their job responsibility, while 21 percent responded that they feel their administration thinks  
1363 it is of little or no importance. See Appendix C-4, Table 2 for summary data.

1364  
1365 In addition, one-third of respondents reported that their senior administrators think that genetics and  
1366 genomics are important to very important to their own job responsibilities; one-third felt it was somewhat  
1367 important; and one-third felt that genetics and genomics are of little or no importance. See Appendix C-4,  
1368 Table 2 for summary data.

1369  
1370 One-third of respondents reported that they feel they have adequate to very adequate resources for  
1371 implementing genetic and genomic competencies in their work or role, while two-thirds reported that the  
1372 resources they have are not or only somewhat adequate. See Appendix C-4, Table 3 for summary data.

1373  
1374 ***Respondents' Role in Public Health***  
1375

1376 Respondents were asked to indicate the level of public health setting in which they work and the amount  
1377 of time spent on genetic or genomic tasks. Most respondents work at the state level (41 percent), followed  
1378 by academia (30 percent), federal level (13 percent), private, nonprofit organizations (9 percent),  
1379 community-based organizations (4 percent), other institutions (e.g., commercial laboratory, medical  
1380 center community programs, nonprofit health organizations) (2 percent), and international positions (1  
1381 percent). No respondents reported working at the local level. The majority of respondents spend less than  
1382 half of their work time on genetic-specific tasks. (Appendix C-4, Table 4 provides a summary of the  
1383 frequencies and percentages of responses to each job level.)

1384  
1385 ***Delivery of Genetic Services to Underserved or Vulnerable Populations***  
1386

1387 Using an open-ended question format, respondents were asked to describe efforts that their organization  
1388 has undertaken to ensure that genetic services or information are available for vulnerable or underserved  
1389 populations and to recommend specific strategies. A total of 71 responses were received in response to  
1390 this question. Reported efforts and strategies included provision of educational materials and development  
1391 of websites; encouraging community involvement, training and education of public health providers, and  
1392 provision of genetic services. Increased funding and development of federal policies were also suggested  
1393 as ways to enhance educational efforts. See Appendix C-5, 1 for more detailed responses.

1394  
1395 The survey closed with an opportunity for individuals to provide additional comments to SACHGS on the  
1396 topic of genetics and genetics education for public health providers. Fifty-four responses were received,  
1397 describing themes around funding, networking and collaboration, best evidence-based practices, and  
1398 education. Details can be found in Appendix C-5, 2.

1399

1400 **D. Summary**

1401  
1402 The literature review provides evidence that the current public health workforce is not well prepared to  
1403 receive and assimilate genetic and genomic information. It also demonstrates that the barriers to achieving  
1404 a more genomics-informed public health workforce are multifaceted. First, the public health workforce is  
1405 diverse and follows many educational and training paths. Thus, a one-size-fits-all approach is not feasible.  
1406 Second, many professionals in the field today received their formal education before genomics became a  
1407 critical aspect of medicine and health. Third, attitudes, perceptions, and beliefs shape the acceptance and  
1408 adoption of genetics and genomics by the public health community.

1409  
1410 Some studies have found that public health educators' perceived barriers included not only lack of basic  
1411 genomic knowledge but also lack of knowledge regarding the link between genomics and health  
1412 promotion. The literature also reveals that public health providers do not perceive public health genomics  
1413 to be part of their job, nor a professional priority. Until evidence of public health benefits of genetic  
1414 testing can be demonstrated (e.g., population screening for *BRCA1* and *BRCA2* and hereditary  
1415 hemochromatosis), public health providers might be resistant to adoption.

1416  
1417 There have been efforts to develop competencies in genetics and genomics for public health providers. In  
1418 August 2000, the CDC Office of Genetics and Disease Prevention and representatives from each of the  
1419 disciplines in public health met to identify the core competencies necessary for all health professionals to  
1420 incorporate genetics into public health practice. The group developed specific genomic competencies for  
1421 various public health providers. ASPH also has highlighted the importance of genomics in the Master's  
1422 Degree in Public Health Core Competency Development Project.

1423  
1424 The SACGHS survey found that respondents believe the 12 competencies developed by SACGHS were  
1425 important. Demonstrating a basic knowledge of the role that genetics and genomics plays in the  
1426 development of disease was considered the most important and most frequently applied competency,  
1427 while confidence in describing the importance of family history ranked highest. Conducting outcomes  
1428 evaluation of available genetic and genomic services ranked the lowest in importance, frequency of  
1429 application, and confidence in demonstrating this competency.

1430  
1431 Sixty percent of survey respondents reported that their senior administration feels that genetics and  
1432 genomics are important to the respondents' job responsibilities. However, the topic was not central to the  
1433 overall administration of the workplace, and only one-third of respondents felt that resources for  
1434 implementing genetic and genomic competencies were adequate or very adequate.

1435  
1436 More than half of respondents provided information on delivery of genetic services to underserved or  
1437 vulnerable populations. These responses included organizational efforts to create culturally and  
1438 linguistically appropriate educational materials, conduct community-based participatory research, train  
1439 entities within local communities to foster outreach, provide genetic counseling either in person or via  
1440 teleconference calls, and conduct research to understand barriers to community access to genetic services.

1441  
1442 Survey respondents also identified strategies and recommendations to target vulnerable or underserved  
1443 populations. These included the need for increased funding to enhance genetic services, outreach, and  
1444 partnerships with vulnerable or underserved populations; development of websites as part of outreach  
1445 tools; and the need for policies to enhance genetic services, raise awareness, and increase education of  
1446 local community members.

1447  
1448 The literature points to a number of factors that impede incorporation of genetics and genomics into  
1449 public health practice and demonstrates that well-defined lines separate the public health workforce

1450 engaged in genetics and genomics, such as in newborn screening programs, and those who do not see  
1451 genetics and genomics as related to their work. While many of the same concerns and barriers were  
1452 highlighted in the SACGHS survey of public health providers, overall, survey respondents had a positive  
1453 attitude toward genetics and genomics. Further, all of the genetic and genomic competencies developed  
1454 for and used in the survey instrument were considered important by respondents. As discussed in the  
1455 survey limitations above, respondents may over represent individuals and organizations that have  
1456 responsibilities in genetics and genomics and thus be biased favorably to the importance of these  
1457 competencies.

1458  
1459 Based on a literature review and its survey findings, SACGHS recognizes that the public health workforce  
1460 is divergent and heterogeneous, which complicates genetic and genomic education and training efforts.  
1461 Educational approaches based on genetic and genomic competencies targeted to the training needs of the  
1462 multiple professional roles within public health will be required for the workforce to effectively address  
1463 public health needs, while recognizing issues of cultural competence, social and economic determinants  
1464 of health, and reduction in health disparities. Identifying effective educational models for public health  
1465 providers serving in underserved communities and identifying the role of family history in population  
1466 health will contribute to improved public health.

## 1467 **IV. The Status of Consumer and Patient Education**

1468

### 1469 **A. Introduction**

1470

1471 Since the inception of the Human Genome Project (HGP), there has been the recognition that the vast  
 1472 amount of information revealed about the human genome would result in ethical, legal, and social issues  
 1473 (ELSI) that affect individuals, families, and communities.<sup>216</sup> The ELSI Program was established in 1990  
 1474 to investigate ELSI issues raised by and as a consequence of the HGP<sup>217</sup> and provide guidance to policy  
 1475 makers and the public on the implications of human genome research.<sup>218</sup> However, it is unlikely that 20  
 1476 years ago we would have expected consumers to be able to order a genetic test directly from the Internet  
 1477 without the participation of a physician.

1478

1479 Today, the term “consumer genomics” refers to the application of genomic technologies by private  
 1480 companies marketing testing services directly to the public via the Internet.<sup>219</sup> How direct access to  
 1481 personal genetic information will change the way consumers approach health care and the extent to which  
 1482 they will seek knowledge on their own and bypass their health care professionals is not known. The  
 1483 emergence of social networking and sharing genetic information via the Internet compounds concerns of  
 1484 confidentiality and the consequences of sharing genetic information in this manner are difficult to  
 1485 predict.<sup>220</sup> However, new technologies also provide innovative solutions to societal problems; for  
 1486 example, the emergence of mHealth—the provision of health related services via mobile  
 1487 communications—is being explored as a way to improve health care services, even in remote and resource-  
 1488 poor environments.<sup>221</sup>

1489

1490 As genetic testing becomes more widely available, the need for education about genetics and the results  
 1491 and implications of testing will grow steadily. However, the number of genetic professionals will not be  
 1492 able to meet the translational and interpretive need,<sup>222</sup> and consumers and patients are likely to seek  
 1493 information on their own. This situation will require genetic literacy. However, genetic literacy is based  
 1494 on having a sufficient educational background (i.e., exposure to, at a minimum, high school scientific  
 1495 coursework) and English language proficiency. In the United States, 21 million individuals speak English  
 1496 “less than very well” and are thus said to be limited English-proficient.<sup>223,224</sup> This chapter summarizes the  
 1497 available literature on consumer and patient knowledge and understanding of genetics and explores how

---

<sup>216</sup> National Human Genome Research Institute. *ELSI Research Program*. See <http://www.genome.gov/>.

Accessed on December 12, 2009.

<sup>217</sup> National Human Genome Research Institute. (1996). Report on The Joint NIH/DOE Committee to Evaluate the Ethical, Legal, and Social Implications Program of the Human Genome Project. See <http://www.genome.gov/10001745>. Accessed on January 14, 2010.

<sup>218</sup> National Human Genome Research Institute. *The Human Genome Project Completion: Frequently Asked Questions*. See <http://www.genome.gov/11006943>. Accessed on February 16, 2010.

<sup>219</sup> Foster, M.W., and Sharp, R.R. (2008). Out of sequence: how consumer genomics could displace clinical genetics. *Nature Review Genetics*. 9(419):AOP, published online 15 April 2008; doi:10.1038/nrg2374.

<sup>220</sup> Resnik, D.B. (2009). Direct-to-consumer genomics, social networking, and confidentiality. *The American Journal of Bioethics*. 9(6-7):45-58.

<sup>221</sup> United Nations Foundation. Mhealth for Development. See <http://www.unfoundation.org/global-issues/technology/mhealth-report.html>. Accessed on April 2, 2010.

<sup>222</sup> Cooksey, J.A., Forte, G., Benkendorf, J., and Blitzer, M.G. (2005). The state of the medical geneticist workforce: findings of the 2003 survey of ABMG certified geneticists. *Genetics in Medicine*. 7(6):439-443.

<sup>223</sup> Regenstien, M., Huang, J., and West, C. (2009). Hospital Language Services: Quality Improvement and Performance Measures. In: Henriksen K, Battles JB, Keyes MA, Grady ML, editors. *Advances in Patient Safety: New Directions and Alternative Approaches*. Vol. 1. Assessment. AHRQ Publication No. 08-0034-1. Rockville, MD: Agency for Healthcare Research and Quality; August 2008.

<sup>224</sup> U.S. Bureau of the Census. *Profile of Selected Social Characteristics: 2000 (Table DP-2)*. See [http://factfinder.census.gov/servlet/ACSSAFFacts?\\_submenuid=factsheet\\_1&\\_sse=on](http://factfinder.census.gov/servlet/ACSSAFFacts?_submenuid=factsheet_1&_sse=on). Accessed on January 14, 2010.

1498 genomics, and personalized medicine may address social, economic, and linguistic disparities that  
1499 genetics research had previously exacerbated.

1500  
1501 To understand what is known about the genetics literacy of the general public, a search was conducted to  
1502 identify pertinent literature covering the years 2003 to 2009 as described in Chapter I (page 8). Additional  
1503 salient documents were collected and reviewed, in particular a literature review conducted in 2009 for the  
1504 National Institutes of Health (NIH) by the Academy for Educational Development (AED).<sup>225</sup> To ensure  
1505 that the opinions of the general public were reviewed for this report, Cogent Research provided SACGHS  
1506 with the findings from its 2008 survey, Cogent Genomics Attitudes and Trends.<sup>226</sup> This national, web-  
1507 based survey consisted of responses from 1,000 adults, representative of the U.S. population by age,  
1508 socioeconomic status, ethnicity, geographic region, and gender.

1509  
1510 In addition, to elucidate the genetic education needs of patients and consumers, here defined as members  
1511 of the public who seek genetic information, SACGHS collected qualitative and quantitative data using  
1512 semi-structured interviews with experts in consumer and patient health education and a web-based survey  
1513 of the health advocacy community. The results of that data gathering effort are presented here.

1514

## 1515 **B. Literature Review**

1516

### 1517 **Genetic Testing Marketing and Communications: A Review of Literature by AED**

1518

1519 The AED conducted a search of published and unpublished literature on the marketing of genetic testing.  
1520 Their review emphasized direct-to-consumer (DTC) genome-wide scans of susceptibility markers for  
1521 common diseases. The review also yielded information relevant to genetic services and information more  
1522 generally. The review addressed two questions:

1523

- 1524 • What is known about current communication and understanding of genetics, genetic risk, DTC  
1525 genomic services, and personalized medicine for the interested public and health care  
1526 professionals?
- 1527 • What are the state-of-the-art research areas or gaps in research regarding current communication  
1528 and understanding of genetics, genetic risk, DTC genomic services, and personalized medicine  
1529 for the interested public and health care professionals?

1530

1531 The search yielded 128 relevant articles published between 1998 and 2009. These were reviewed by AED  
1532 to assess what is being communicated to consumers about genetics, genetic risks, genetic services, and  
1533 personalized medicine.

1534

1535 AED concluded from the literature that, while most consumers have a positive attitude toward genetic  
1536 testing, their understanding of genetic testing is very basic, often misinformed, and does not appear to be  
1537 increasing over time. Specifically, the AED literature review identified that:

1538

- 1539 • Consumers do not understand that there are many types of genetic and genomic tests, and there  
1540 are many contexts in which they are used.

---

<sup>225</sup> 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.

<sup>226</sup> Cogent Research, LLC. (2008). Cogent Genomics Attitudes and Trends: 2008. Provided to SACGHS with permission to cite, April 29, 2009.

- 1541 • Consumers do not have ready access to balanced and accurate information or personalized  
1542 guidance about genetic tests. DTC marketing usually does not fulfill this need. Several  
1543 government Internet sites provide good information about genetic testing; however, these sites are  
1544 geared primarily to health care professionals.
- 1545 • Although consumers would prefer to learn about genetic tests from their health care professionals,  
1546 most physicians are not adequately trained in genetics. Physicians recognize the limitations in  
1547 their knowledge and expertise and are therefore reluctant to order genetic tests and provide  
1548 genetic counseling.

1549  
1550 The AED identified the following methods that would improve the public's understanding of genetic  
1551 testing:

- 1552
- 1553 • Effective communication methods based on succinct, accurate, and unbiased information about  
1554 genetic tests could be promoted by nonprofit and professional organizations and by government  
1555 agencies.
  - 1556 • Education strategies should consider that limited health literacy constitutes a formidable barrier to  
1557 the public's understanding of genetic tests.
  - 1558 • Standardized physician training, to include both didactic instruction and supervised experience in  
1559 the delivery of genetic health care, would allow physicians to better educate the public about  
1560 genetic tests.

### 1561 **Knowledge of Consumers and Patients Regarding Genetics and Genomics**

1562  
1563  
1564 Studies that have assessed the public's knowledge of genomics and genetic testing generally have found  
1565 that the public has only a rudimentary knowledge of basic genetic terms<sup>227</sup> yet overall positive attitudes  
1566 towards genetics.<sup>228</sup> In general, people seem to be reasonably aware that genetic risk factors contribute to  
1567 health outcomes.<sup>229,230,231,232</sup> However, understanding of genetic risk factors is dependent on education  
1568 and health literacy, which also varies by race, ethnicity and English language proficiency in the United  
1569 States.<sup>233,234,235,236,237,238</sup> According to Cogent Research, overall awareness of genetics by the public

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<sup>227</sup> Lanie, A.D., Jayaratne, T.E., Sheldon, J.P., Kardia, S.L.R., Anderson, E.S., Fledbaum, M., and Petty, E.M. (2004). Exploring the public understanding of basic genetic concepts. *Journal of Genetic Counseling*. 13(4):305-320.

<sup>228</sup> Etchegary, H., Cappelli, M., Potter, B., Vloet, M., Graham, I., Walker, M., and Wilson, B. (2010). Attitude and knowledge about genetics and genetic testing. *Public Health Genomics*. 13:80-88.

<sup>229</sup> Moscarillo, T.J., Holt, H., Perman, M., Goldberg, S., Cortellini, L., Stoler, J.M., DeJong, W., Miles, B.J., Albert, M.S., Go, R.C.P., and Blacker, D. (2007). Knowledge of and attitudes about Alzheimer disease genetics: reports of a pilot survey and two focus groups. *Community Genetics*. 10:97-102.

<sup>230</sup> Smerecnik, C.M.R., Mesters, I., de Vries, N.K., and de Vries, H. (2008). Educating the general public about multifactorial genetic disease: applying a theory-based framework to understand current public knowledge. *Genetics in Medicine*. 10(4):251-258.

<sup>231</sup> Etchegary, H., Cappelli, M., Potter, B., Vloet, M., Graham, I., Walker, M., and Wilson, B. (2010). Attitude and knowledge about genetics and genetic testing. *Public Health Genomics*. 13:80-88.

<sup>232</sup> Lanie, A.D., Jayaratne, T.E., Sheldon, J.P., Kardia, S.L.R., Anderson, E.S., Fledbaum, M., and Petty, E.M. (2004). Exploring the public understanding of basic genetic concepts. *Journal of Genetic Counseling*. 13(4):305-320.

<sup>233</sup> Regenstein, M., Huang, J., and West, C. (2009). Hospital Language Services: Quality Improvement and Performance Measures. In: Henriksen K, Battles JB, Keyes MA, Grady ML, editors. *Advances in Patient Safety: New Directions and Alternative Approaches*. Vol. 1. Assessment. AHRQ Publication No. 08-0034-1. Rockville, MD: Agency for Healthcare Research and Quality; August 2008.

<sup>234</sup> Catz, D.S., Green, N.S., Tobin, J.N., Lloyd-Puryear, M.A., Kyler, P., Umamoto, A., Cernoch, J., Brown, R., and Wolman, F. (2005). Attitudes about genetics in underserved, culturally diverse populations. *Community Genetics*. 8(3):161-72.

<sup>235</sup> Kennedy, B.R., Mathis, C.C., and Woods, A.K. (2007). African Americans and their distrust of the health care system: healthcare for diverse populations. *Journal of Cultural Diversity*. 14(2):56-60.

<sup>236</sup> Johnson, V.A., Edwards, K.A., Sherman, S.L., Stephens, L.D., Williams, W., Adair, A., and Deer-Smith, M.H. (2009). Decisions to participate in fragile X and other genomics-related research: Native American and African American voices. *Journal of Cultural Diversity*. 16(3):127-135.

1570 increased between 2006 and 2008. In 2008, 79 percent of respondents reported that they had heard or  
 1571 read about using individual genetic information to understand and optimize health and about half of  
 1572 respondents felt informed about their family history.<sup>239</sup>

1573  
 1574 The literature and surveys, however, are not informative about whether members of the public know how  
 1575 to use genetic information to understand their risk of disease or to optimize health. And, because those  
 1576 who participate in studies may not reflect the balance of race and ethnicity in the United States, the  
 1577 findings may not apply to the general U.S. population.

1578  
 1579 Smerecnik et al.<sup>240</sup> performed a literature review of studies published between 1990 and 2007 of public  
 1580 knowledge of genetic risk factors of multifactorial genetic diseases. These studies suggest that, on  
 1581 average, 59 percent of individuals surveyed were aware of the existence of genetic risk factors (range,  
 1582 17.6 to 93.3 percent). Cogent Research found that 50 percent of respondents were aware that genes  
 1583 predict the likelihood of developing specific diseases. However, among Cogent survey respondents, less  
 1584 than 5 percent understood that genetic information can be used to optimize health.<sup>241</sup> Awareness of risk  
 1585 factors also varies depending on the disease; for example, in the studies reviewed by Smerecnik,<sup>242</sup> 60  
 1586 percent of the general public was aware of genetic risk in breast cancer, while only 20 percent was aware  
 1587 of genetic factors in cervical cancer. Knowledge beyond awareness, however, such as how to process  
 1588 such information and use it in decisionmaking, was far more limited.

1589  
 1590 Levels of genetic knowledge have also been found to differ by ethnicity, English language proficiency,  
 1591 and socioeconomic background.<sup>243,244</sup> Several studies have linked level of education with knowledge of  
 1592 genetic concepts or genetic testing, demonstrating that, as might be expected, the higher the education  
 1593 level achieved, the greater the genetic knowledge.<sup>245,246,247,248</sup> In a study that assessed knowledge about  
 1594 genetics and genetic testing among 560 women in Ontario, in which 80 percent had college degrees, only  
 1595 3 percent reported having no knowledge of genetics, 68 percent felt that their genetic knowledge was

<sup>237</sup> Ku, L., and Waidmann, T. (2003). How race/ethnicity, immigration status and language affect health insurance coverage, access to care and quality of care among the low-income population. Final report. Washington, D.C. Kaiser Family Foundation, Publication No. 4132. See <http://www.kff.org/uninsured/kcmu4132report.cfm>. Accessed on December 14, 2009.

<sup>238</sup> David, R.A., and Rhee, B. (1998). The impact of language as a barrier to effective health care in an underserved urban Hispanic community. *Mount Sinai Journal of Medicine*. 65:393-397.

<sup>239</sup> Cogent Research. Cogent Genomics Attitudes and Trends: 2008. Provided to SACGHS with permission to cite, April 29, 2009.

<sup>240</sup> Smerecnik, C.M.R., Mesters, I., de Vries, N.K., and de Vries, H. (2008). Educating the general public about multifactorial genetic disease: applying a theory-based framework to understand current public knowledge. *Genetics in Medicine*. 10(4):251-258.

<sup>241</sup> Cogent Research. Cogent Genomics Attitudes and Trends: 2008. Provided to SACGHS with permission to cite, April 29, 2009.

<sup>242</sup> Smerecnik, C.M.R., Mesters, I., de Vries, N.K., and de Vries, H. (2008). Educating the general public about multifactorial genetic disease: applying a theory-based framework to understand current public knowledge. *Genetics in Medicine*. 10(4):251-258.

<sup>243</sup> Catz, D.S., Green, N.S., Tobin, J.N., Lloyd-Puryear, M.A., Kyler, P., Umamoto, A., Cernoch, J., Brown, R., and Wolman, F. (2005). Attitudes about genetics in underserved, culturally diverse populations. *Community Genetics*. 8:161-172.

<sup>244</sup> Kessler, L., Collier, A., and Halbert, C.H. (2007). Knowledge about genetics among African Americans. *Journal of Genetic Counseling*. 16(2):191-200.

<sup>245</sup> Tambor, E.S., Rimer, B.K., and Strigo, T.S. (1997). Genetic testing for breast cancer susceptibility: awareness and interest among women in the general population. *American Journal of Medical Genetics*. 68:43-49.

<sup>246</sup> Kelly, K., Leventhal, H., Marvin, M., Toppmeyer, D., Baran, J., and Schwalb, M. (2004). Cancer genetics knowledge and beliefs and receipt of results in Ashkenazi Jewish individuals receiving counseling for BRCA1/2 mutations. *Cancer Control*. 11(4):236-244.

<sup>247</sup> Waller, J., McCafferey, K., and Wardle, J. (2004). Beliefs about the risk factors for cervical cancer in a British population sample. *Preventive Medicine*. 38:745-753.

<sup>248</sup> Peters, J. A., Beckjord, E.B., Banda Ryan, D.R., Carr, A.G., Vadaparampil, S.T., Loud, J.T., Korde, L., and Greene, M.H. (2008). Testicular cancer and genetic knowledge among familial testicular cancer family members. *Journal of Genetic Counselors*. 17:351-364.

1596 about the same as most people, and 21 percent reported knowing more than most people about  
 1597 genetics.<sup>249</sup>

1598  
 1599 It might be expected that people with a family history of a specific genetic-related disorder would be  
 1600 more knowledgeable about genetics in general and their own risk in particular for developing the disorder.  
 1601 However, this has not been shown in the literature. Several studies have evaluated genetics literacy  
 1602 among individuals with or at risk for genetic diseases. First-degree relatives of early onset familial  
 1603 Alzheimer disease were found to have limited knowledge of their own personal risk of developing the  
 1604 disease.<sup>250</sup> Similar findings were reported by Moscarillo et al.<sup>251</sup> General genetics knowledge among  
 1605 persons with familial testicular cancer and their family members was found to be generally low, with less  
 1606 than half (41 percent) of respondents able to answer questions correctly regarding testicular cancer and  
 1607 genetics.<sup>252</sup> A study of adults with cystic fibrosis found that they have limited knowledge of the genetics  
 1608 of their disorder.<sup>253</sup> Furthermore, knowledge of genetics and genetic testing among people with chronic  
 1609 illness has been found to be lacking, particularly among older people and those with less education.<sup>254</sup>

1610  
 1611 **Where the Public Get Its Information**

1612  
 1613 The Internet has become a significant source for consumer and patient knowledge regarding genetics. A  
 1614 1999 study by Stockdale found that even a decade ago people seeking information about the genetics of  
 1615 Alzheimer disease actively searched the Internet for information.<sup>255</sup> More recent studies show that  
 1616 Internet usage by seekers of genetic information has become more sophisticated. Schaffer et al. found  
 1617 that mothers of children with genetic disorders used the Internet to interpret, produce, and circulate  
 1618 genetic knowledge—activities that caused them to value their own experiential knowledge.<sup>256</sup> Eighty-  
 1619 three percent of families referred to a pediatric genetics clinic obtained information from the Internet  
 1620 regarding their child’s diagnosis.<sup>257</sup> In this study, convenience, privacy, and finding information they did  
 1621 not otherwise have were cited as advantages to searching the Internet. Two reported barriers to finding  
 1622 relevant, understandable information were difficulties in key word searching methods that produced either  
 1623 too much or too little information, and an inability to interpret information that was found.  
 1624 In a study of perspectives on access to genetic knowledge by families of children with spinal muscular  
 1625 atrophy, most had received some type of genetic counseling, and families who acquired knowledge from  
 1626 the Internet or support groups had roughly the same amount of genetic knowledge as those who received  
 1627 genetic counseling from a health care professional.<sup>258</sup> A general practitioner was the preferred source of

---

<sup>249</sup> Etchegary, H., Cappelli, M., Potter, B., Vloet, M., Graham, I., Walker, M., and Wilson, B. (2010). Attitude and knowledge about genetics and genetic testing. *Public Health Genomics*. 13:80-88.

<sup>250</sup> Marcheco, B., Bertoli, A.M., Rojas, I., and Heredero, L. (2003). Attitudes and knowledge about presymptomatic genetic testing among individuals at high risk for familial, early-onset Alzheimer’s disease. *Genetic Testing*. 7(1):45-47.

<sup>251</sup> Moscarillo, T.J., Holt, H., Perman, M., Goldberg, S., Cortellini, L., Stoler, J.M., DeJong, W., Miles, B.J., Albert, M.S., Go, R.C.P., and Blacker, D. (2007). Knowledge of an attitude about Alzheimer’s disease genetics: reports of a pilot survey and two focus groups. *Community Genetics*. 10:97-102.

<sup>252</sup> Peters, J. A., Beckjord, E.B., Banda Ryan, D.R., Carr, A.G., Vadaparampil, S.T., Loud, J.T., Korde, L., and Greene, M.H. (2008). Testicular cancer and genetic knowledge among familial testicular cancer family members. *Journal of Genetic Counselors*. 17:351-364.

<sup>253</sup> Houser, G.H., Holt, C.L., Clancy, J.P., Leon, K., Rowe, S.M., Gaggar, A., Gutierrez, H.H., Young, K.R., and Robin, N.H. (2008). Genetic and reproductive knowledge among adolescents and adults with cystic fibrosis. *Chest Journal*. 133(6):1533.

<sup>254</sup> Morren, M., Rijken, M., Baanders, A.N., and Bensing, J. (2007). Perceived genetic knowledge, attitudes towards genetic testing, and the relationship between these among patients with a chronic disease. *Patient Education and Counseling*. 65:197-204.

<sup>255</sup> Stockdale, A. (1999). Public understanding of genetics and Alzheimer’s disease. *Genetic Testing*. 3(1):139-145.

<sup>256</sup> Schaffer, R., Kuczynski, K., and Skinner, D. (2008). Producing genetic knowledge and citizenship through the Internet: mothers, pediatric genetics, and cybermedicine. *Sociology of Health & Wellness* 30(1):145-159.

<sup>257</sup> Roche, M.I., and Skinner, D. (2008). How parents search, interpret, and evaluate genetic information obtained from the internet. *Journal of Genetic Counselors*. 18:119-219.

<sup>258</sup> Meldrum, C., Scott, C., and Swoboda, K.J. (2007). Spinal muscular atrophy genetic counseling access and genetic knowledge: parents perspectives. *Journal of Child Neurology*. 22(8):1019-1026.

1628 genetic information in a Dutch study of patients with chronic disease, followed by information brochures,  
1629 medical specialists, and special Internet sites.<sup>259</sup> In a focus group study of culturally diverse populations  
1630 recruited from community health centers, study participants obtained or wanted to obtain genetic  
1631 information from television and from someone that they would trust, such as a doctor, suggesting that  
1632 these would be useful mechanisms to convey genetic information in community health settings.<sup>260</sup>  
1633

1634 The 2008 Cogent Research survey revealed that when participants were asked where they heard about  
1635 using genetic information to understand and optimize health, 55 percent cited television; 39 percent cited  
1636 newspaper or magazine stories; 28 percent cited the Internet; and 13 percent cited family members,  
1637 friends, or co-workers.<sup>261</sup>  
1638

### 1639 **The Public's Confidence in Its Genetic Knowledge**

1640

1641 Most studies that assess consumers' knowledge or perceived knowledge of genetics do not take into  
1642 account the confidence that respondents have in their genetics knowledge. Lanie et al.<sup>262</sup> interviewed 62  
1643 adults to assess their genetic knowledge and self awareness of their lack of knowledge. The authors found  
1644 a significant number of individuals who believed they held accurate knowledge but whose responses to  
1645 question were actually incorrect. Past research suggests that it is easier to educate individuals who realize  
1646 their current understanding is flawed than individuals who are unaware of their limitations.<sup>263</sup> In  
1647 providing genetics education and training for patients and consumers, most resources have been geared  
1648 towards those who are actively seeking information, while few methods have been proposed for how to  
1649 educate those who are unaware of their lack of knowledge.  
1650

### 1651 **The Public's Attitudes about Genetics**

1652

1653 A number of studies have reported that people who have or think they have an understanding of basic  
1654 genetics have positive attitudes towards genetic testing.<sup>264,265,266</sup> Overall, the general public has been  
1655 supportive of genetic testing to improve disease diagnosis and prevention.<sup>267,268,269</sup> Etchegary et al. found  
1656 that 95 percent of survey respondents thought genetic information should be used to improve disease

---

<sup>259</sup> Morren, M., Rijken, M., Baanders, A.N., and Bensing, J. (2007). Perceived genetic knowledge, attitudes towards genetic testing, and the relationship between these among patients with a chronic disease. *Patient Education and Counseling*. 65:197-204.

<sup>260</sup> Catz, D.S., Green, N.S., Tobin, J.N., Lloyd-Puryear, M.A., Kyler, P., Umamoto, A., Cernoch, J., Brown, R., and Wolman, F. (2005). Attitudes about genetics in underserved, culturally diverse populations. *Community Genetics*. 8:161-172.

<sup>261</sup> Cogent Research. Cogent Genomics Attitudes and Trends: 2008. Provided to SACGHS with permission to cite, April 29, 2009.

<sup>262</sup> Lanie, A.D., Jayaratne, T.E., Sheldon, J.P., Kardia, S.L.R., Anderson, E.S., Fledbaum, M., and Petty, E.M. (2004). Exploring the public understanding of basic genetic concepts. *Journal of Genetic Counseling*. 13(4):305-320.

<sup>263</sup> Renner, C.H., and Renner, M. J. (2001). But I thought I knew that: using confidence estimation as a debiasing technique to improve classroom performance. *Applied Cognitive Psychology*. 15:23-32.

<sup>264</sup> Reitz, F., Barth, J., and Bengal, J. (2004). Predictive value of breast cancer cognitions and attitudes toward genetic testing on women's interest in genetic testing for breast cancer risk. *Psycho-Social-Medicine*. 1(3):1-13.

<sup>265</sup> Morren, M., Rijken, M., Baanders, A.N., and Bensing, J. (2007). Perceived genetic knowledge, attitudes towards genetic testing, and the relationship between these among patients with a chronic disease. *Patient Education and Counseling*. 65:197-204.

<sup>266</sup> Jallinoja, P., and Aro, A.R. (2000). Does knowledge make a difference? The association between knowledge about genes and attitudes toward gene tests. *Journal of Health Communication*. 5:29-39.

<sup>267</sup> Morren, M., Rijken, M., Baanders, A.N., and Bensing, J. (2007). Perceived genetic knowledge, attitudes towards genetic testing, and the relationship between these among patients with a chronic disease. *Patient Education and Counseling*. 65:197-204.

<sup>268</sup> Henneman, L., Timmermans, D., and van der Wal, G. (2004). Public experiences, knowledge and expectations about medical genetics and the use of genetic information. *Community Genetics*. 7:33-43.

<sup>269</sup> Baruch, S., Kaufman, D., and Hudson, K. (2007). *U.S. Public Opinion on Uses of Genetic Information and Genetic Discrimination*. See [http://www.dnapolicy.org/pub\\_reports.php?action=detail&report\\_id=23](http://www.dnapolicy.org/pub_reports.php?action=detail&report_id=23). Accessed on November 23, 2009.

1657 diagnosis and determine why people are more or less likely to develop a disease. Seventy percent thought  
 1658 that genetic information should be used to design individualized drugs for people, and 85 percent believed  
 1659 patients should be able to receive genetic testing even if it conflicted with other family members' decision  
 1660 not to undergo genetic testing. Further, 43 percent believed that doctors were obligated to share genetic  
 1661 information of importance to other family members, even if it violated the patient's right to privacy.<sup>270</sup>  
 1662 The majority of respondents in this study had not thought about potential negative consequences genetic  
 1663 information might have for insurance coverage or employment discrimination.

1664  
 1665 A 2007 study by the Genetics and Public Policy Center—conducted before the passage of the Genetic  
 1666 Information Nondiscrimination Act of 2008 (GINA)—found that although a majority of Americans  
 1667 “enthusiastically support genetic testing for research and health care;” 92 percent also expressed concern  
 1668 that “results of a genetic test that tells a patient whether he or she is at increased risk for a disease like  
 1669 cancer could be used in ways that are harmful to the person.”<sup>271</sup> Cogent Research's 2008 survey was  
 1670 conducted shortly after the passage of GINA on May 16, 2008. Despite wide media coverage around that  
 1671 time, only 16 percent of respondents to the Cogent survey knew that there were laws that protect the  
 1672 privacy of genetic information, and only one-quarter of those felt that protections were sufficient. Almost  
 1673 half of consumers in the Cogent survey expressed greater concern about having their DNA stored and  
 1674 tested without their permission than having the information be part of their medical record.<sup>272</sup> Regarding  
 1675 attitudes about genetic testing without treatment options, most participants in an Alzheimer disease study  
 1676 believed that testing should not be withheld until better treatment options are available.<sup>273</sup>

1677  
 1678 **Health Disparities and Cultural Issues Related to Genetics**

1679  
 1680 Numerous reports have documented the extent of health disparities in the United States, and the field of  
 1681 genetics is no exception.<sup>274</sup> When any new technology emerges it has the potential to exacerbate  
 1682 disparities if patients and providers do not have access to appropriate and relevant information. Genetic-  
 1683 related disparities include lack of awareness of and access to genetic counseling and genetic testing.  
 1684 When the standard of care is to offer a genetic test, there is evidence that minorities do not participate in  
 1685 genetic testing or are offered genetic counseling services as often as whites. African American women  
 1686 were found to be much less likely to undergo genetic counseling than white women for *BRCA1* and  
 1687 *BRCA2* genetic testing.<sup>275</sup> How much of these disparities can be attributed to issues of access versus  
 1688 knowledge of and attitudes about genetic testing is not clear. However, several studies have attributed  
 1689 lower genetic knowledge to socioeconomic factors. Those with lower levels of education have been found  
 1690 to be less knowledgeable about basic genetic information, the role of genetics and chronic disease, and  
 1691 genetic testing.<sup>276, 277, 278</sup>

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<sup>270</sup> Etchegary, H., Cappelli, M., Potter, B., Vloet, M., Graham, I., Walker, M., and Wilson, B. (2010). Attitude and knowledge about genetics and genetic testing. *Public Health Genomics*. 13:80-88.

<sup>271</sup> Baruch, S., Kaufman, D., Hudson, K. (2007). *U.S. Public Opinion on Uses of Genetic Information and Genetic Discrimination*. See [http://www.dnapolicy.org/pub\\_reports.php?action=detail&report\\_id=23](http://www.dnapolicy.org/pub_reports.php?action=detail&report_id=23). Accessed November 23, 2009.

<sup>272</sup> Cogent Research. Cogent Genomics Attitudes and Trends: 2008. Provided to SACGHS with permission to cite, April 29, 2009.

<sup>273</sup> Higgs, Y.G., Roberts, S., Farrer, L.A., and Green, R.C. (2003). Differences between African Americans and whites in their attitudes toward genetic testing for Alzheimer's disease. *Genetic Testing*. 7(1):39-44.

<sup>274</sup> Hall, M., and Olopade, O.I. (2005). Confronting genetic testing disparities. *Journal of the American Medical Association*. 293(14):1783-1785.

<sup>275</sup> Armstrong, K., Micco, E., Carney, A., Stopfer, J., and Putt, M. (2005). Racial differences in the use of BRCA 1/2 testing among women with a family history of breast or ovarian cancer. *Journal of the American Medical Association*. 293(14):1729-1736.

<sup>276</sup> Mogilner, A., Otten, M., Cunningham, J.D., and Brower, S.T. (1998). Awareness and attitudes concerning BRCA gene testing. *Annals of Surgical Oncology*. 5(7):567-568.

<sup>277</sup> Chu, K.C., and Lamar, C.A. (2003). Racial disparities in breast carcinoma survival rates. *Cancer*. 97(11):2853-2860.

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1692 Several studies have compared knowledge of genetic testing in general and genetic testing specifically for  
1693 *BRCA* mutations and cancer risk among African Americans and whites. Forty-nine percent of African  
1694 Americans and 72 percent of whites had heard of genetic testing in general.<sup>279</sup> Knowledge has been  
1695 shown to be lower for *BRCA* genetic testing with 19 to 25 percent of African Americans and 35 to 68  
1696 percent of whites reporting knowledge of this test.<sup>280,281</sup> Wideroff et al. found that 49.9 percent of whites  
1697 had heard of genetic testing for cancer risk compared to 32.9 percent of African Americans and only 20.6  
1698 percent of Latinos.<sup>282</sup>  
1699  
1700 Zimmerman et al. found that 90 percent of a survey sample of equal numbers of inner city African  
1701 Americans and Caucasians thought “genetic testing to check for risk of getting a disease was a good idea”  
1702 regardless of race.<sup>283</sup> Other studies have found that attitudes about genetics among African Americans  
1703 and Latinos differ from whites. African Americans and Latinos overall hold a positive view of genetics,  
1704 but it is not as positive as whites.<sup>284,285,286,287</sup> Nonetheless, Zimmerman et al. found that 58 percent of  
1705 African Americans and 34 percent of whites thought genetic testing would lead to racial  
1706 discrimination.<sup>288</sup> In a survey of 170 African Americans and 181 Caucasians in Philadelphia, Peters et al.  
1707 found that the belief that genetic testing would lead to racial discrimination was low but more prominent  
1708 among African Americans than whites,<sup>289</sup> and that African Americans were less likely to endorse the  
1709 health benefits of genetic testing. In two studies exploring attitudes about genetic testing for Alzheimer  
1710 disease, African Americans expressed less interest in genetic testing but anticipated less negative personal  
1711 consequences from a positive result compared to whites.<sup>290,291</sup> In a study among Latinos in New York

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<sup>278</sup> Sussner, K.M., Thompson, H.S., Valdimarsdottir, H.B., Redd, W.H., and Jandorf, L. (2009). Acculturation and familiarity with, attitudes towards and beliefs about genetic testing for cancer risk within Latinas in east Harlem, New York City. *Journal of Genetic Counselors*. 18:60-71.

<sup>279</sup> Peters, J. A., Beckjord, E.B., Banda Ryan, D.R., Carr, A.G., Vadaparampil, S.T., Loud, J.T., Korde, L., and Greene, M.H. (2008). Testicular cancer and genetic knowledge among familial testicular cancer family members. *Journal of Genetic Counselors*. 17:351-364.

<sup>280</sup> Mogilner, A., Otten, M., Cunningham, J.D., and Brower, S.T. (1998). Awareness and attitudes concerning BRCA gene testing. *Annals of Surgical Oncology*. 5(7):567-568.

<sup>281</sup> Peters, J. A., Beckjord, E.B., Banda Ryan, D.R., Carr, A.G., Vadaparampil, S.T., Loud, J.T., Korde, L., and Greene, M.H. (2008). Testicular cancer and genetic knowledge among familial testicular cancer family members. *Journal of Genetic Counselors*. 17:351-364.

<sup>282</sup> Wideroff, L., Vadaparampil, S.T., Breen, N., Croyle, R.T., and Freedman, A.N. (2003). Awareness of genetic testing for increased cancer risk in the year 2000 National Health Interview Survey. *Community Genetics*. 6(3):147-156.

<sup>283</sup> Zimmerman, R.K., Tabbarah, M., Nowalk, M.P., Raymund, M., Jewell, I.K., Wilson, S.A., and Ricci, E.M. (2006). Racial differences in beliefs about genetic screening among patients at inner-city neighborhood health centers. *Journal of the National Medical Association*. 98(3):370-377.

<sup>284</sup> Hips, Y.G., Roberts, S., Farrer, L.A., and Green, R.C. (2003). Differences between African Americans and whites in their attitudes toward genetic testing for Alzheimer’s disease. *Genetic Testing*. 7(1):39-44.

<sup>285</sup> Hall, M., and Olopade, O.I. (2005). Confronting genetic testing disparities. *Journal of the American Medical Association*. 293(14):1783-1785.

<sup>286</sup> Sussner, K.M., Thompson, H.S., Valdimarsdottir, H.B., Redd, W.H., and Jandorf, L. (2009). Acculturation and familiarity with, attitudes towards and beliefs about genetic testing for cancer risk within Latinas in east Harlem, New York City. *Journal of Genetic Counselors*. 18:60-71.

<sup>287</sup> Zimmerman, R.K., Tabbarah, M., Nowalk, M.P., Raymund, M., Jewell, I.K., Wilson, S.A., and Ricci, E.M. (2006). Racial differences in beliefs about genetic screening among patients at inner-city neighborhood health centers. *Journal of the National Medical Association*. 98(3):370-377.

<sup>288</sup> Zimmerman, R.K., Tabbarah, M., Nowalk, M.P., Raymund, M., Jewell, I.K., Wilson, S.A., and Ricci, E.M. (2006). Racial differences in beliefs about genetic screening among patients at inner-city neighborhood health centers. *Journal of the National Medical Association*. 98(3):370-377.

<sup>289</sup> Peters, N., Rose, A., and Armstrong, K. (2004). The association between race and attitudes about predictive genetic testing. *Cancer Epidemiology, Biomarkers & Prevention*. 13(3):361-365.

<sup>290</sup> Hips, Y.G., Roberts, S., Farrer, L.A., and Green, R.C. (2003). Differences between African Americans and whites in their attitudes toward genetic testing for Alzheimer’s disease. *Genetic Testing*. 7(1):39-44.

<sup>291</sup> Moscarillo, T.J., Holt, H., Perman, M., Goldberg, S., Cortellini, L., Stoler, J.M., DeJong, W., Miles, B.J., Albert, M.S., Go, R.C.P., and Blacker, D. (2007). Knowledge of an attitude about Alzheimer’s disease genetics: reports of a pilot survey and two focus groups. *Community Genetics*. 10:97-102.

1712 City, Sussner et al. found that individuals with higher levels of acculturation—or the degree to which they  
 1713 have adopted the attitudes, values, and behaviors of the majority culture—were more likely to be familiar  
 1714 with genetic testing and to perceive its benefits.<sup>292</sup>

1715  
 1716 Recent research on the relationship between ethnicity and minority status and socioeconomic status (SES)  
 1717 on awareness and uptake of genetic testing has resulted in inconsistent findings. While Bowen et al.<sup>293</sup> did  
 1718 not find any differences in SES and reactions to a DTC campaign for *BRCA1* and *BRCA2* genetic testing,  
 1719 their study did reveal that in general, women of lower SES reported less knowledge about genetics and  
 1720 risk, yet more interest in genetic testing. This finding suggests that women of lower SES may be  
 1721 requesting unnecessary genetic tests based on an incomplete understanding of genetic risks. Awareness of  
 1722 genetic testing for cancer susceptibility is lower among racial and ethnic groups compared to whites, but  
 1723 it is important to look more closely at the specific SES factors in addition to race and ethnicity. Education,  
 1724 country of origin, insurance coverage, and parental history of cancer have all been found to have an  
 1725 influence on awareness. These factors differ across racial and ethnic groups, suggesting that policy  
 1726 remedies are unlikely to have uniform population effects, and customized strategies using culturally  
 1727 relevant media and native languages are needed among different groups or communities.<sup>294</sup>

1728  
 1729 Another factor to consider in health disparities relates to literacy and English language proficiency. The  
 1730 U.S. Census Bureau reported that 13 percent of Americans had not completed high school in 2008<sup>295</sup> and  
 1731 from 2000 census data, 21 million Americans speak English “less than well”.<sup>296</sup> This low English  
 1732 language proficiency is more common in minority populations and limits access to medical care,  
 1733 specifically by a decrease in health care visits.<sup>297</sup> Socioeconomic factors underlie educational level and  
 1734 may account for the increased difficulties disadvantaged individuals will have with health literacy in  
 1735 general, and with specific understanding of the role genomics plays in maintaining health and in defining  
 1736 disease risks.<sup>298</sup> One strategy that may begin to address literacy as a barrier in health care is to identify  
 1737 those with lower literacy, and a tool has been developed to identify patients with low literacy in a clinical  
 1738 genetics setting.<sup>299</sup>

1739 When genetic and genomic educational materials are available, they are not always provided in a  
 1740 culturally appropriate fashion, in a language that is used or understood in immigrant or ethnic  
 1741 communities, or provided in formats or through media that disadvantaged communities can access or  
 1742 utilize. Addressing health disparities through education about genetics and genomics may therefore  
 1743 require innovative methods, culturally sensitive translations, and use of locally predominant languages to  
 1744 reach all communities. Research has found that patients who inquire about or request a genetic test serve

<sup>292</sup> Sussner, K.M., Thompson, H.S., Valdimarsdottir, H.B., Redd, W.H., and Jandorf, L. (2009). Acculturation and familiarity with, attitudes towards and beliefs about genetic testing for cancer risk within Latinas in east Harlem, New York City. *Journal of Genetic Counselors*. 18:60-71.

<sup>293</sup> Bowen, D.J., Harris, J., Jorgensen, C.M., Myers, M.F., and Kuniyuki, A. (2010). Socioeconomic influences on the effects of a genetic testing direct-to-consumer marketing campaign. *Public Health Genomics*. 13(3):131-42.

<sup>294</sup> Pagan, J.A., Su, D., Li, L., Armstrong, K. (2009). Racial and ethnic disparities in awareness of genetic testing for cancer risk. *American Journal of Preventative Medicine*. 37(6):524-530.

<sup>295</sup> United States Census Bureau. *U.S. Census Bureau News 2009*. See <http://www.census.gov/Press-Release/www/releases/archives/education/013618.html>. Accessed on January 6, 2010.

<sup>296</sup> United States Census Bureau. *America Speaks: A Demographic Profile of Foreign-Language Speakers for the United States: 2000*. Table 1a. See <http://www.census.gov/population/socdemo/hh-fam/AmSpks/01%20US%20per.xls>. Accessed on January 6, 2010.

<sup>297</sup> Shi, L., Lebrum, L., and Tsai, J. (2009). The influence of English proficiency on access to care. *Ethnicity and Health*. 14(6):625-642.

<sup>298</sup> Murray, E., Lo, B., Pollack, L., Donelan, K., and Lee, K. (2004). Direct-to-consumer advertising: public perceptions of its effects on health behaviors, health care, and the doctor-patient relationship. *Journal of the American Board of Family Practice*. 17(1):6-18.

<sup>299</sup> Erby, L.H., Roter, D., Larson, S., and Cho, J. (2008). The rapid estimate of adult literacy in genetics (REAL-G): a means to assess literacy deficits in the context of genetics. *American Journal of Medical Genetics*. 146A:174-181.

1745 as an inducement to physician use of genetic services;<sup>300</sup> thus, the use of strategies customized to specific  
 1746 groups and communities may be an effective way to promote the use of emerging genetic and genomic  
 1747 technologies, when medically appropriate, and empower a wide variety of consumers to act as their own  
 1748 health care advocates.<sup>301</sup> Programs such as the Community Genetics Education Network (CGEN)  
 1749 Project<sup>302</sup> reinforce the need to use principles of community-based participatory research to identify  
 1750 effective ways to increase genetic literacy among diverse populations.

1751  
 1752 The lack of awareness and understanding about genetics in clinical practice and public health also play  
 1753 out in research settings. The promise of genomics may not benefit those who do not participate in genetic  
 1754 and genomic research. Studies have found that attitudes about genetics among African Americans and  
 1755 Latinos differ from whites, and that minorities in the United States are less likely to participate in  
 1756 research, including genetic and genomic research. Without the participation of all segments of the  
 1757 population, it will be difficult to tailor treatments and preventive measures for specific sub-populations or  
 1758 for individuals. For example, limited participation in research by minorities becomes problematic as  
 1759 pharmacogenomic research uncovers variance in the efficacy of treatments and drug development and  
 1760 increasingly focuses on products tailored to individual risk. However, racial and ethnic health disparities  
 1761 may be exacerbated if researchers assume that the basis of health disparities is solely due to genetics and  
 1762 conduct research in a way that seems to affirm a genetic basis for racial differences in disease  
 1763 prevalence.<sup>303,304</sup> This assumption arises when researchers overemphasize the genetic contributions to  
 1764 disease and health without consideration of social contributions to health.<sup>305</sup> Recommended educational  
 1765 efforts should therefore focus on ameliorating attitudes about the purpose of genomic research and  
 1766 increasing the understanding of the complex interrelation of genes and the environment, including social  
 1767 contributions to health. When the promise of personalized medicine is understood, disparities in research  
 1768 participation and in the provision of appropriately tailored health care can be potentially reduced. The  
 1769 recognition that health disparities are heavily rooted in social structure requires that educational efforts  
 1770 acknowledge the broader context of socioeconomic, cultural attitudes, educational level, literacy, gender,  
 1771 and English-language proficiency in order to educate both the scientific community and the public.<sup>306</sup>

1772  
 1773 Although studies have documented disparities in access to genetic services, other studies suggest that  
 1774 genomics and personalized medicine may help address disparities. Increasingly widespread use of the  
 1775 Internet, for example, may open access to personal genomic information and reach larger numbers of  
 1776 people than are currently seen by genetic counselors or clinical geneticists.<sup>307</sup> Furthermore, should whole-  
 1777 genome sequencing become affordable and accessible in the future, fair representation of all major groups

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<sup>300</sup> Wideroff, L., Freeman, A., Olson, L., Klabunde, C.N., Davis, D., Srinath, K.P., Croyle, R.T., and Ballard-Barbash, R. (2003). Physician use of genetic testing for cancer susceptibility: results of a National survey. *Cancer Epidemiology, Biomarkers and Prevention*. 12:295-303.

<sup>301</sup> Hall, M., and Olopade, O.I. (2005). Confronting genetic testing disparities. *Journal of the American Medical Association*. 293(14):1783-1785.

<sup>302</sup> Community Genetics Education Network Project. See <http://www.nchpeg.org/slides/2009/Kyler.ppt#271.2>. Accessed on March 18, 2010.

<sup>303</sup> Bonham, V.L., Citrin, T., Modell, S.M., Franklin, T.H., Bleicher, E.W.B., and Fleck, L.M. (2009). Community-based dialogue: engaging communities of color in the United States' genetics policy conversation. *Journal of Health Politics, Policy and Law*. 34(3):325-359.

<sup>304</sup> Sankar, P., Cho, M.K., Condit, C.M., Hunt, L.M., Koenig, B., Marshall, P., Lee, L.S-J., and Spicer, P. (2004). Genetic research and health disparities. *Journal of the American Medical Association*. 291(24):2985-2989.

<sup>305</sup> Bonham, V.L., Citrin, T., Modell, S.M., Franklin, T.H., Bleicher, E.W.B., and Fleck, L.M. (2009). Community-based dialogue: engaging communities of color in the United States' genetics policy conversation. *Journal of Health Politics, Policy and Law*. 34(3):325-359.

<sup>306</sup> Ramos, E., and Rotimi, C. (2009). The A's, G's, C's, and T's of health disparities. *BMC Medical Genomics*. 2:29.

<sup>307</sup> Foster, M.W., and Sharp, R.R. (2008). Out of sequence: how consumer genomics could displace clinical genetics. *Nature Review Genetics*. 9(6):419.

1778 will be required to avoid large gaps in understanding the human genome.<sup>308</sup> A shift to whole-genome  
 1779 sequencing may also help resolve the emphasis on research that uses ancestry or ethnicity as an easy  
 1780 shortcut for identifying genomic associations.<sup>309</sup>

1781  
 1782 With whole-genome sequencing, and incorporation of related ancestry and family health history, the “too  
 1783 narrow focus on genetic variation” can be replaced by “personalized information that can and should  
 1784 guide clinical decisionmaking for individuals.”<sup>310</sup> This personalized information would include  
 1785 “observables such as the environment and physiology” that would help clarify genomically similar  
 1786 individuals with different environmental exposures, cultural practices, and access to medical services.<sup>311</sup>  
 1787 These factors, and the individual’s self-identification with a specific family health history (i.e., high blood  
 1788 pressure or heart disease among family members), may be just as important as a shared genetic  
 1789 background for discerning risk. When psychosocial factors are combined with a better understanding of  
 1790 the degree of genetic variation within racial and ethnic groups, genomic studies can move beyond  
 1791 “classifying and subsequently treating [ethnic/racial sub-populations] as one uniform group.” Ramos and  
 1792 Rotimi, for example, explore how studies on the efficacy of beta-blockers among African Americans  
 1793 highlight the need for personalized, rather than racial or ethnic categories for appropriate and effective  
 1794 treatment decisions.<sup>312,313</sup>

1795  
 1796 **Selected Education Programs Targeted to the Public**

1797  
 1798 Incorporation of genetic content into K-12 curricula has been underway for some time as a part of a  
 1799 greater effort to improve science literacy. Most states have curriculum content standards that include  
 1800 genetics and related topics.<sup>314</sup> However, there have been persistent calls for improving science curricula  
 1801 overall and genetics content in particular, with emphasis on the need to shift the focus of genetics  
 1802 education from single-gene, qualitative traits to complex traits and in essence, “invert” the genetics  
 1803 curriculum to teach about complex traits before rare, Mendelian genetic concepts.<sup>315</sup> The challenges of  
 1804 improving genetics education at the K-12 level are significant. However, other than to acknowledge how  
 1805 important K-12 education is in enhancing public understanding of genetic and genomics, it is beyond the  
 1806 scope of this report.

1807  
 1808 Recognizing the need for comprehensive population-based state genetics plans, some states have  
 1809 conducted needs assessments to better understand and define the priorities of the general public, health  
 1810 and human service providers, and educators. For example, the Michigan Department of Community  
 1811 Health–Hereditary Disorders and Newborn Screening Programs conducted a needs assessment in 2000-  
 1812 2002 that gathered input from 1,000 residents to develop a comprehensive state genetics plan.<sup>316</sup> One of

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<sup>308</sup> Need, A.C., and Goldstein, D.B. (2009). Next generation disparities in human genomics: concerns and remedies. *Trends in Genetics*. 25(11):489-94.

<sup>309</sup> Gurwitz, D., and Lunshof, J.E. (2009). Ancestry in translational genomic medicine: handle with care. *Genome Medicine*. 25;1(2):24.

<sup>310</sup> Kohane, I.S. (2009). The twin questions of personalized medicine: who are you and whom do you most resemble? *Genome Medicine*. 20;1(1):4.

<sup>311</sup> Kohane, I.S. (2009). The twin questions of personalized medicine: who are you and whom do you most resemble? *Genome Medicine*. 20;1(1):4.

<sup>312</sup> Ramos, E., and Rotimi, C. (2009). The A's, G's, C's, and T's of health disparities. *BMC Medical Genomics*. 22;2:29.

<sup>313</sup> Liggett, S.B., Cresci, S., Kelly, R.J., Syed, F.M., Matkovich, S.J., Hahn, H.S., Diwan, A., Martini, J.S., Sparks, L., Parekh, R.R., Spertus, J.A., Koch, W.J., Kardia, S.L., Dorn, G.W. (2008). A GRK5 polymorphism that inhibits beta-adrenergic receptor signaling is protective in heart failure. *Nature Medicine*. 14(5):510-7.

<sup>314</sup> American Society of Human Genetics. See [http://www.ashg.org/education/k12\\_statestandards.shtml](http://www.ashg.org/education/k12_statestandards.shtml). Accessed on November 24, 2009.

<sup>315</sup> Dougherty, M.J. (2009). Closing the gap: inverting the genetics curriculum to ensure an informed public. *American Journal of Human Genetics*. 85:6-12.

<sup>316</sup> Beene-Harris, R., and Bach, J.V. (2009). Michigan Genetics Plan: a report on the needs assessment process. *Health Promotion Practice*. 10(2): 201-209.

1813 its many conclusions was: “There is a tremendous need to educate all sectors of the population especially  
1814 underrepresented communities about the role of genetics in health and disease, including related ethical,  
1815 legal, and social issues. A central Michigan-focused source is needed as a portal for the public to obtain  
1816 reliable information about genetic disorders, resources, and services.” As a result, an online, Michigan-  
1817 focused genetics resource center providing a central source of information on genetic health care and  
1818 related topics was developed and a toll-free number established.<sup>317</sup> The importance of a central location  
1819 for accessible online information for consumers was recognized by the crafters of the Newborn Screening  
1820 Saves Lives Act of 2008.<sup>318</sup> This legislation earmarked funding and directed the Health and Resources  
1821 Service Administration (HRSA) to develop a clearinghouse for newborn screening educational and family  
1822 support and services information, materials, resources, research, and data that would be interactive,  
1823 regularly updated, and link to government and nonprofit websites. The Genetic Alliance was awarded the  
1824 contract to develop this clearinghouse and began work on the project in September 2009.<sup>319</sup>

1825  
1826 In recent years there has been a concerted effort to develop tools and public involvement in efforts to  
1827 enhance family history taking. In particular, the Office of the Surgeon General, NIH, the Centers for  
1828 Disease Control and Prevention, and HRSA initiated a public health campaign to increase awareness of  
1829 the importance of family history and to promote the use of family medical history as an education and  
1830 screening tool for determining disease risk. This effort is further addressed in Chapter V, but it is an  
1831 important tool for consideration in public education efforts.  
1832

### 1833 **C. SACGHS Survey of the Genetic and Genomic Information Needs of** 1834 **Consumers and Patients**

#### 1835 **Methodologies**

1836  
1837  
1838 To elucidate the genetic education needs of patients and consumers, here defined as members of the  
1839 public who seek genetic information, SACGHS collected qualitative and quantitative data using semi-  
1840 structured interviews with professionals working in consumer and patient health education and a web-  
1841 based survey of the health advocacy community. These strategies provided the Committee with additional  
1842 data to inform their recommendations.  
1843

1844 **Semi-Structured Interviews.** A list of 30 individuals widely regarded as leaders in genetics advocacy for  
1845 consumers and patients was generated by SACGHS members and staff, as well as by attendees of an  
1846 annual NCHPEG meeting. Based on this list, between December 2008 and February 2009, SACGHS  
1847 conducted semi-structured interviews with 11 experts (see Appendix D-1) in the fields of disease and  
1848 disability advocacy, genetics services for patients, health education and communication, for-profit DTC  
1849 genetics service companies, and science and genetics education of the public. The purpose of the  
1850 interviews was to collect data on current and emerging needs of consumers and patients as their lives are  
1851 increasingly influenced by genomics, and to inform the development of a survey. The interviews were  
1852 conducted by telephone with a lead interviewer from the National Human Genome Research Institute and  
1853 with one or two experts participating in each interview. The lead interviewer used an interview script with  
1854 general themes asked of all experts and specific questions based upon the interviewees’ expertise (see  
1855 Appendix D-1). These themes were used to help construct the web-based survey.  
1856

1857 **Web-Based Survey.** Informed by the interviews, SACGHS developed a 12-item online survey instrument  
1858 (see Appendix D-2) to collect data from the genetics and health advocacy communities regarding their

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<sup>317</sup> Michigan’s Genetics Resource Center. See <http://www.migeneticsconnection.org/>. Accessed on November 25, 2009.

<sup>318</sup> U.S. Government Printing Office. *Public Law 110-204 – Newborn Screening Saves Lives Act of 2007*. See <http://www.gpo.gov/fdsys/pkg/PLAW-110publ204/html/PLAW-110publ204.htm>. Accessed on February 25, 2010.

<sup>319</sup> Newborn Screening Clearinghouse. See <http://www.nbsclearinghouse.org/about>. Accessed on February 25, 2010.

1859 opinions on the genetic and genomic education needs of patients and the general public. During April and  
1860 May 2009, the survey was distributed to representatives of health advocacy groups, community-based  
1861 health-focused organizations, and communities specializing in genetic education for the public. The  
1862 survey also was distributed by the Genetic Alliance,<sup>320</sup> a nonprofit health advocacy organization  
1863 committed to transforming health through genetics. The survey was determined to be exempt from the  
1864 need for Institutional Review Board review and approval by the NIH Office of Human Subjects Research.  
1865

1866 The survey took about 10 minutes to complete and participants could opt out of answering any of the  
1867 questions. An invitation to participate in the survey, with a hyperlink to the survey instrument was sent to  
1868 approximately 1,100 individuals. The survey system received 337 whole or partial responses.  
1869

### 1870 ***Data Limitations***

1871  
1872 Qualitative research is ideal for exploring complex themes such as those presented in this report.  
1873 However, there are limitations to qualitative data including the potential for selection bias and social  
1874 desirability in responses. There are also limitations to the SACGHS online quantitative survey. A random  
1875 sampling strategy was not used and stakeholders and the public who responded are not necessarily  
1876 representative of the public and may have had unique interests or experiences that led to their  
1877 participation in the survey. Another potential limitation is the possibility of response bias.  
1878

1879 SACGHS sought to minimize any limitations in the data used for this report by using multiple data  
1880 collection methods. The approaches used were designed to gather data in different formats from different  
1881 constituencies. The qualitative approach allowed for in-depth discussion and exploration of themes, and  
1882 the online survey included opinions from those who are or have been seekers of genetic  
1883 information. Even with the limitations addressed above, this process provides a snapshot of the needs of  
1884 consumer and patients who have varying degrees of involvement in genetics.  
1885

### 1886 **Survey Findings**

#### 1888 ***Semi-Structured Interviews***

1889  
1890 An analysis of the qualitative interview transcripts identified common themes related to the  
1891 educational needs of consumers and patients, successful educational models, and recommended actions  
1892 the government can take to improve the public's understanding of genetics and genomics. The first set of  
1893 themes relate to perceptions about consumers' understanding of genetics and genomics. Specifically,  
1894 consumers are finding it difficult to understand new advances in genetic technologies and the potential  
1895 benefits and risks of these technologies, how genes and behaviors relate to each other, complex traits, and  
1896 how a single condition may involve multiple risk factors. Interview findings also suggest that consumers  
1897 frequently misunderstand the concept of genetic predisposition as well as current limitations in our  
1898 knowledge of test validity and utility. These misunderstandings are compounded by the difficulty  
1899 consumers have in finding accurate information about genetics and genomics.  
1900

1901 The interviews also explored various approaches to genetics education. Suggestions included the need to  
1902 improve genetic and genomic education among health care providers and to recognize that collaborative  
1903 projects between public and private organizations can facilitate the identification of specific educational  
1904 needs. Respondents suggested that an important first step in developing programs is to assess and  
1905 understand the needs of specific communities. They also suggested that the Internet could be used  
1906 effectively as a source of balanced, accurate information.  
1907

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<sup>320</sup> Genetic Alliance. See <http://www.geneticalliance.org>. Accessed on August 18, 2009.

1908 The third set of themes relates to the role of government in educating the public about genetics and  
1909 genomics. Respondents suggested that consumers believe that the Federal government is a more unbiased  
1910 source of information than commercial sources and that it should have a central role in genetics education  
1911 of the public. Consumers also think that government should monitor the societal effects of genetic and  
1912 genomic testing and services, clarify the extent to which laboratory tests are regulated, and should support  
1913 formal genetics education in schools and have some influence over educational standards. In addition,  
1914 those interviewed suggested that government should fund more programs to improve genetic literacy.  
1915 These themes, aggregated as key findings, are explored in greater detail in Appendix D-1, Table 1.

1916  
1917 ***Web-Based Survey Results***

1918  
1919 Two hundred and fifty-eight individuals responded to a question asking in what state they work,  
1920 providing information on the geographical distribution of service provision. Respondents work in 39  
1921 states plus the District of Columbia. This distribution, shown in Appendix D-3, Figure 1, demonstrates  
1922 that the largest number of responses (> 17) came from California, Maryland, New York, and the District  
1923 of Columbia, with a strong showing (>10 responses) from Massachusetts, North Carolina, Georgia,  
1924 Florida, Texas, Michigan, and Illinois. No responses were received from Alaska, Hawaii, Idaho,  
1925 Wyoming, North Dakota, South Dakota, Mississippi, West Virginia, Vermont, New Hampshire, and  
1926 Rhode Island.

1927  
1928 The respondents represent a wide variety of organization types, including health care organizations (47)  
1929 advocacy groups (53), academic institutions (66), private industry (29), public health organizations (14),  
1930 and other (60). The “other” category, which required a free-text response, included community-based  
1931 health organizations, nonprofit organizations and support groups, and private hospitals and private health  
1932 care practices. This distribution is presented as a pie chart in Appendix D-3, Figure 2.

1933  
1934 Respondents were asked to rate how important genetics is to their organization. Sixty percent felt that  
1935 genetics was important or very important to their organization, while only 1 percent stated that it was not  
1936 at all important (see Appendix D-3, Table 2). About 55 percent of respondents reported that they had been  
1937 involved with their organization in planning or implementing a genetics education program for seekers of  
1938 genetic information.

1939  
1940 Participants were asked to rank a set of five concepts that “individuals most need to know about genetics  
1941 and genomics to be informed seekers of genetic information as it relates to health” (see Appendix D-3,  
1942 Table 3). Eighty-nine percent of respondents answered this question. The most important concept was that  
1943 “family history is an important tool for understanding health and disease.”

1944  
1945 Participants also were given the opportunity to suggest more important items in a free-text response,  
1946 resulting in 60 additional responses. Themes that emerged from these responses were: (1) the concept that  
1947 there is a difference between disease risk and disease diagnosis; (2) understanding probabilities, as well as  
1948 terms like “common” and “rare,” is essential to interpreting the results of genetic tests; and (3) genetic  
1949 tests should be interpreted by people knowledgeable in genetics and genomics.

1950  
1951 Survey participants were asked to rank a set of four topics that “may have special relevance for seekers of  
1952 genetic information as it relates to health” (see Appendix D-3, Table 4). Eighty-five percent of  
1953 respondents answered this question. The most favored topic was where consumers would find reliable  
1954 information on genetics and genomics, indicating that the ability to direct consumers to such resources  
1955 may present a significant gap in available resources or awareness of existing resources. Thirty-six free-  
1956 text responses suggested other important topics. Among these, the challenges of cost, insurance  
1957 reimbursement, and malpractice insurance requiring practitioners to give “worst case scenarios” rather  
1958 than balanced risk assessments emerged.

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1959 Respondents were asked to rank the following list of genetic education and services needs of underserved  
1960 and vulnerable populations.

1961

- 1962 • Basic and relevant genetic health information
- 1963 • Skills to make informed health decisions
- 1964 • Culturally appropriate genetic health information
- 1965 • Education about access to genetic services

1966

1967 First, however, they were given the option to state whether there were more pressing needs above genetics  
1968 education to which 7 percent (22 of 315 responses) responded in the affirmative. There was poor  
1969 discrimination among the rankings but the need for basic and relevant genetic health information was  
1970 ranked highest and education about access to genetic services was ranked lowest (see Appendix D-3,  
1971 Table 5.)

1972

1973 Respondents were asked that if they were part of an organization, to report whether their organization had  
1974 created educational programs to address the challenges in underserved and vulnerable populations. Fifty-  
1975 three percent of respondents (189 of 337) answered this question, reporting development of education  
1976 programs to address at least one of these challenges.

1977

1978 The most important educational need identified (i.e., basic and relevant genetic health information) also  
1979 was reported as the most common topic for educational programs. Education about access to genetic  
1980 services was the second most frequent response, even though this challenge was the lowest priority  
1981 identified in the previous question. In the free-text responses to this item, a common theme was genetic  
1982 education aimed at disease-specific support groups.

1983

1984 Eighty-three percent of participants responded to a request to rank a set of five “barriers to genetics and  
1985 genomics education efforts for seekers of genetic information as it relates to health.” The two highest  
1986 ranked barriers were lack of health professionals’ understanding of genetics and lack of individual health  
1987 literacy in genetics. The lower ranked barriers were direct-to-consumer marketing of genetic tests before  
1988 there is evidence of utility and lack of access to genetic services for consumers and patients (see  
1989 Appendix D-3, Table 6).

1990

1991 Among the 29 free-text responses to this item, additional important barriers included fear of genetic  
1992 discrimination and loss of job or insurance based on genetic test results; and lack of cultural competency,  
1993 whether in terms of spoken language or in the complexity of the language used to educate consumers on  
1994 genetics and genomics.

1995

1996 In a series of questions, the respondents were asked to rank a set of six potential roles for three levels of  
1997 government: Federal, state, and local. Appendix D-3, Table 7 shows these rankings.

1998

1999 At all levels of government, funding was ranked as the key role. The key secondary role for the Federal  
2000 government was to serve as a clearinghouse for educational information. This role, however, was ranked  
2001 among the lowest priorities for state and local governments. Another very low priority at all three levels  
2002 of government was education about the licensing of genetic health care providers.

2003

2004 Education about anti-discrimination laws was determined to be of high priority for all three levels of  
2005 government, echoing the sentiments from previous survey items that the public has a fear of being  
2006 discriminated against based on genetic tests.

2007

2008 Among the 21 free-text responses regarding the role of local government, 13 indicated that there is no role  
2009 in genetics and genomics education for local government. The remaining responses suggested that local  
2010 governments could educate the public as to where locally available resources could be found and could  
2011 require genetics education in public schools.

2012  
2013  
2014

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

2015 In a free-text box, respondents were asked for their opinion about the role that the U.S. Department of  
2016 Health and Human Services (HHS) should play to improve genetics education for those seeking  
2017 information about genetics as it relates to health. Nearly 200 responses were received. The following  
2018 major themes emerged:

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2030

- 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.

2031  
2032

**D. Summary**

2033 In general, the literature review found that the public has been supportive of genetic testing when it is  
2034 used for improving disease diagnosis and prevention. The literature review conducted in 2009 by AED  
2035 found that although consumers have a limited understanding of genetic testing, they have positive  
2036 attitudes about genetic testing and are generally motivated to seek information and undergo testing.  
2037 However, it is not clear from the literature that the public understands how to use genetic information to  
2038 optimize health. Knowledge beyond awareness, such as how to process such information and use it in  
2039 decisionmaking, is limited. Levels of genetic knowledge also have been found to differ by race, ethnicity,  
2040 and socioeconomic background.

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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.

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2057

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

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2058 optimize health. For consumers to understand genetic testing, they must appreciate the distinction  
2059 between the risk for a disease and its diagnosis. There is consensus that genetics education should focus  
2060 on multifactorial disorders, the value and limitations of genetic testing and DTC genetic services, and  
2061 personalized guidance about genetic tests.  
2062

2063 Despite the availability of DTC testing, consumers still prefer to have genetic tests done in their doctor's  
2064 office. This desire on the part of consumers underscores the deficiencies of most primary care providers  
2065 in their general genetic knowledge and their specific lack of comfort in selecting, ordering, and  
2066 interpreting genetic tests and in providing appropriate genetic counseling.  
2067

2068 Even though much of the data that informs this report was collected shortly after passage of GINA,  
2069 concern about confidentiality and disclosure of genetic information that might lead to loss of a job or  
2070 insurance persists. The fear of DNA being collected without consent was also expressed.  
2071

2072 Health literacy in genetics for health professionals and consumers is considered a gap, and an  
2073 underpopulated genetic workforce is a barrier to rectifying this problem. State and Federal governments  
2074 are viewed as having important roles in educating consumers and health care providers alike. There also  
2075 is a belief that the Federal government should regulate and evaluate genetic tests and determine who is  
2076 qualified to provide genetic services. The Federal government is seen as the logical repository for  
2077 educational information and should serve as a clearinghouse for this information. Providing funding for  
2078 educational programs is considered a primary role of government.  
2079

2080 Review of current literature, findings from a SACGHS survey, and interviews exploring consumer  
2081 attitudes and beliefs about genetics and gaps in genetics and genomic education, point to an underlying  
2082 need for improved genetic literacy beginning in the formative years and continuing throughout the  
2083 lifespan. The complexity and rapid evolution of knowledge and technology related to genetics and  
2084 genomics and the varying learning needs of communities and individual consumers will require that  
2085 educational efforts and resources directed to consumers be appropriately translated and tailored to specific  
2086 segments of the population.

2087 **V. Activities of Selected Federal Agencies**  
 2088

2089 In August 2003, a survey was distributed to 16 SACGHS *ex officio* agencies to obtain information about  
 2090 Federal activities related to the education of professionals in genetics. The agencies were asked to  
 2091 provide: (1) information on their overall efforts to assess genetics workforce needs and to address  
 2092 genetics education and training of professionals in both health and nonhealth-related fields; (2) a list of  
 2093 specific activities the agency funded in this area for the preceding year; and (3) specific information about  
 2094 the nature and purpose of the activity, its target audience, and funding information.  
 2095

2096 Seven *ex officio* agencies—the Department of Commerce (DOC), the Department of Defense (DOD), the  
 2097 Department of Energy (DOE), the Department of Justice (DOJ), and three Department of Health and  
 2098 Human Services (HHS) agencies—the Centers for Disease Control and Prevention (CDC), the Health  
 2099 Resources and Services Administration (HRSA), and the National Institutes of Health (NIH)—submitted  
 2100 information about their ongoing activities in response to the request. Eight *ex officio* agencies—the  
 2101 Department of Labor; the Equal Employment Opportunity Commission (EEOC); and six HHS agencies  
 2102 (the Administration for Children and Families (ACF), the Agency for Healthcare Research and Quality  
 2103 (AHRQ), the Centers for Medicare & Medicaid Services (CMS), the Food and Drug Administration  
 2104 (FDA), the Office for Civil Rights (OCR), and the Office for Human Research Protections (OHRP)—  
 2105 reported that they were not performing or funding any activities relevant to the SACGHS request but do  
 2106 engage in outreach and educational activities in other areas. Results of this survey were presented to  
 2107 SACGHS on October 23, 2003.<sup>321</sup>  
 2108

2109 With the rapid expansion in relevant genomics information over the intervening five years, SACGHS  
 2110 elected to repeat the prior survey of Federal agencies with those that currently have *ex officio*  
 2111 representation on the Committee. The final version of the 2008 survey was shortened from the original  
 2112 survey, which had required agencies to enumerate specific projects with great granularity.  
 2113

2114 The 2008 Federal survey consisted of a mix of closed- and open-ended, narrative-type response questions.  
 2115 These questions explored themes such as the perceived role of the responding agency in genomics  
 2116 education; the perceived ability of the agency to fulfill this role; partnerships established to facilitate  
 2117 genomics educational activities; and a brief description of past, present, and planned educational  
 2118 activities.  
 2119

2120 The survey was distributed to *ex officio* agency representatives to SACGHS in late 2008 and early 2009.  
 2121 Nonresponders were contacted by e-mail or telephone to prompt completion of the survey.  
 2122

2123 Ten *ex officio* agencies—DOC, DOD, DOE, the Federal Trade Commission (FTC), EEOC, the National  
 2124 Science Foundation (NSF), and four HHS agencies—CDC, CMS, HRSA, and NIH—submitted  
 2125 information about their ongoing activities in response to the request. Six *ex officio* agencies—the  
 2126 Department of Education, and five HHS agencies (ACF, AHRQ, OCR, OHRP, and the Substance Abuse  
 2127 and Mental Health Services Administration)—reported that they were not performing or funding any  
 2128 activities relevant to the SACGHS request but do engage in outreach and educational activities in other  
 2129 areas. The Department of Veterans Affairs reported that it conducts activities but was not able to  
 2130 complete the survey due to a change in personnel.  
 2131

2132 The discussion below provides a brief overview of the agencies reported genetics and genomics activities,  
 2133 the criteria used to determine what types of educational activities to pursue and how these activities relate

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<sup>321</sup> Secretary’s Advisory Committee on Genetics, Health, and Society Website. *Federal Efforts in Genetics Education and Training of Professionals, October 23, 2003*. See [http://oba.od.nih.gov/SACGHS/sacghs\\_past\\_meeting\\_2003\\_oct\\_23.html](http://oba.od.nih.gov/SACGHS/sacghs_past_meeting_2003_oct_23.html). Accessed on August 31, 2009.

2134 to the agency's mission, the target audience for the educational activities, and the identification of future  
2135 needs in genetics and genomic education and training.

2136  
2137 For agencies that responded to both SACGHS surveys (in 2003 and 2008), information is provided that  
2138 compares and/or contrasts the reported material.  
2139

2140 ***Data Limitations***

2141  
2142 The surveys conducted in 2003 and 2008 yielded numerous examples of genetics and genomics  
2143 educational activities in federal agencies. However, there are several important limitations that affect  
2144 interpretation of the data including that the agencies that participated in 2003 and 2008 were not the same  
2145 making precise comparisons difficult. Although six agencies responded to both surveys, which provided  
2146 information on the growth of programs or changes in educational priorities, four agencies participated  
2147 only in the 2008 survey. In addition, the level of detail varied across responses. Some agencies provided  
2148 URLs for web-based materials, program funding amounts, relation of activities to agency mission, and  
2149 specific numbers of individuals trained, and other agencies provided only the name of a program or  
2150 project without additional details. Funding information for specific activities and programs was provided  
2151 by several of the respondents; however, there are limitations in interpreting this information both among  
2152 organizations and across time due to vagaries in defining health professional education and incomplete  
2153 data on the funding of intramural or nonprogrammatic activities (e.g., website architecture and content,  
2154 agency staff time and effort). Furthermore, given the range of genetic and genomic educational activities  
2155 and training programs conducted by federal agencies since the first survey in 2003, the individuals who  
2156 responded to the surveys may not have been aware of all the programs, Internet resources, printed  
2157 publications, or targeted training programs initiated by specific divisions within their agency.  
2158

2159 **Agencies Responding to the 2003 and 2008 Surveys**

2160  
2161 **CDC**

2162  
2163 CDC's mission is to collaborate with partners across the Nation to create the expertise, information, and  
2164 tools that people and communities need to protect their health—through health promotion, prevention of  
2165 disease, injury and disability, and preparedness for new health threats.  
2166

2167 In 2003, CDC's activities in genetics education focused primarily on educating the current and future  
2168 public health workforce on genetics and genomics. These activities were viewed as essential to realizing  
2169 the goals of improved population health and decreased disease incidence. CDC developed partnerships  
2170 with national, state, and local public health organizations to assess the need for genomic educational  
2171 efforts. It brought together public health leaders, health care clinicians, insurers, and others to develop  
2172 programs and educational tools on genetics and genomics targeted to the public health workforce and/or  
2173 the clinical health care workforce. Additional activities reported at that time were focused on  
2174 appropriately utilizing genetic and genomic technologies and ensuring high-quality genetic testing. In all,  
2175 CDC reported 28 activities in their 2003 survey response.  
2176

2177 In 2008, CDC reported that as genetics and genomics become more integral to public health research and  
2178 practice, the need for genetics expertise in public health has become even greater than previously  
2179 reported. CDC's target audiences for genetics education activities include public health administrators,  
2180 medical school and residency training programs to include faculty and students, primary care and  
2181 specialty physicians, epidemiologists, health educators, laboratorians, and environmental health workers;  
2182 and encompassing both the internal CDC workforce and external clinical and public health providers. The  
2183 survey response noted that CDC's role in genetic education and training of professionals is to promote the  
2184 effective and responsible application of genomics knowledge and tools to promote population health that

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2185 spans multiple applications, including chronic disease, environmental health, occupational health, and  
2186 infectious disease. Although, the agency reported on 16 existing genetics education programs, due to  
2187 limited available resources to assess educational needs among professionals, and to develop and  
2188 disseminate training tools and curricula in collaboration with their partners, CDC reported that they are  
2189 not currently able to develop this area fully and respond to emerging developments in genomics.  
2190

2191 Currently, education and training activities in genomics cut across several CDC divisions and offices.  
2192 Although the Office of Public Health Genomics initiates many training activities, others have been  
2193 conducted by the Division of Laboratory Systems; Division of Birth Defects and Developmental  
2194 Disabilities; the Division of Nutrition, Physical Activity and Obesity; the Division of Partnerships and  
2195 Strategic Alliances; and the Office of Workforce and Career Development.  
2196

2197 CDC's projected priorities for future initiatives in genetics education and training center on empowering  
2198 providers with the knowledge and skills to apply genomics knowledge and tools for early detection,  
2199 disease prevention, and health promotion in populations.  
2200

### 2201 **DOC**

2202  
2203 Of the agencies that comprise DOC, only the National Institute of Standards and Technology (NIST)  
2204 reported ongoing projects in genetics education and training. These projects are in adherence with NIST's  
2205 mission to develop and promote measurement, standards, and technology to enhance productivity,  
2206 facilitate trade, and improve the quality of life.  
2207

2208 In 2003, NIST's activities in genetics education focused primarily on cancer genetics, forensic  
2209 applications, and the education and training of practicing professionals. Although the medical and cancer  
2210 genetics program predominantly served health-related professionals, the forensic applications involved  
2211 both health-related and nonhealth-related professionals (i.e., lawyers, judges, and law enforcement  
2212 professionals). Specific needs addressed through the genetic education and training efforts at NIST  
2213 included the development of standards for measurement technologies of genetic information and the  
2214 education of professionals in the use of these standards.  
2215

2216 By 2008, NIST had expanded its activities in genetic education to include students in training and  
2217 practicing professionals; educational websites and online resources targeted to students and professionals;  
2218 forensic laboratory site visits as a component of continuing education (CE); assessments of professional  
2219 knowledge about genetics; and analyses and evaluations of the genetics' workforce training and  
2220 educational efforts. During the period from 2003-2008, NIST built and currently maintains the world's  
2221 most widely used web-based database on forensic DNA genetic typing (STRBase); held more than 30  
2222 training workshops in forensic laboratories and at major scientific conferences to teach genetic principles  
2223 to scientists and lawyers; and established the NIST Human Identity Project that educates students and  
2224 professionals about genetics and is funded by DOJ. (See Appendix E-DOC-NIST for details about these  
2225 projects.)  
2226

2227 DOC's projected priorities for future initiatives include the continuation of the NIST Human Identity  
2228 Project, ongoing workshops and conferences, and continued efforts to evaluate professional knowledge  
2229 about genetics and assess laboratory performance in forensic analysis.  
2230

### 2231 **DOD**

2232  
2233 The DOD health care system seeks to enhance our Nation's security by providing health support for the  
2234 full range of military operations and by sustaining the health of all those entrusted to its care. DOD  
2235 considers genetics education and training as integral to the functioning of the military health care system

2236 and has focused significant efforts to ensure that genetics is appropriately integrated and that staff is  
2237 adequately educated in genetics and ethics.

2238  
2239 In 2003, learning needs in the evolving fields of genetics and genetic technologies were identified through  
2240 assessment and consideration of applicable practice standards. This effort ensured that new services and  
2241 technologies were integrated with organized implementation plans throughout the medical treatment  
2242 facilities. These plans included staff education, policy developments such as operating instructions and  
2243 guidelines, evidence-based practices, and competency-based evaluation.

2244  
2245 The Uniformed Services University of the Health Sciences has taken a leadership role in incorporating  
2246 genetics content into the curricula of both the School of Medicine and the Graduate School of Nursing.  
2247 Genetics also has been a component of CE programs for clinical specialties such as pediatrics, oncology,  
2248 and obstetrics and gynecology. These programs and curricula are evaluated using academic, professional,  
2249 and CE association methods to determine the impact and effectiveness of these activities.

2250  
2251 By 2008, DOD articulated a dual health care mission—readiness and benefits. The readiness mission is  
2252 supported through provision of medical services to the Armed Forces during military operations and the  
2253 benefits mission through health care to more than 9 million eligible beneficiaries worldwide. DOD  
2254 continues to recognize the need for professional education and training in genetics for the readiness  
2255 mission and to provide excellent health care to its beneficiaries.

2256  
2257 DOD's current capabilities in genetics include a genetics workforce, laboratory facilities, and educational  
2258 programs. The genetics workforce consists of physicians with training in clinical genetics, genetic  
2259 counselors, and pathologists with certification in molecular genetics. Facilities focused on genetics  
2260 include a dedicated molecular genetics and cytogenetic laboratory, the Armed Forces Institute of  
2261 Pathology, which performs clinical molecular genetics testing, and plans for a reference molecular  
2262 genetics laboratory. Currently, the U. S. military is the most experienced practitioner of  
2263 pharmacogenomic screening on a large, population-based scale. In addition to ongoing genetic testing  
2264 programs, DOD has developed a comprehensive DOD-wide newborn screening laboratory program and  
2265 has plans to create a general genetics division under the supervision of an Air Force geneticist. (See  
2266 Appendix E-DOD for details.)

2267  
2268 Educational activities include fellowship training in genetics and ongoing efforts to update curriculum  
2269 and clinical training to meet accreditation requirements of the Accreditation Council on Graduate Medical  
2270 Education (ACGME). In addition, DOD has multiple inter-departmental relationships engaged in  
2271 personalized medicine programs and EHR standardization efforts pertaining to genomics. Future DOD  
2272 activities in genetics education and training include support for additional genetics fellowships. DOD will  
2273 also maintain ACGME certification for its CE curricula in genetics, expand its workforce of geneticists  
2274 and genetic counselors, create new laboratory capabilities, and increase its understanding of the gene-  
2275 environmental impacts associated with military operations.

2276  
2277 **DOE**

2278  
2279 In 2003, the DOE survey response focused on the new capabilities emerging in genetics and the mapping  
2280 of the human genome as the context for its activities. At that time, DOE noted that in order to make the  
2281 best use of new capabilities in science, education in genetics and genomics was essential. DOE also  
2282 focused on some of the social implications of the mapping of the genome and, along with NIH, has  
2283 devoted 3 to 5 percent of its annual Human Genome Project budget to studying the ethical, legal, and  
2284 social issues related to the availability of genetic information.

2285

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2286 DOE's commitment to education in genetics and genomics is consistent with its view of science and  
2287 support of interdisciplinary research. The DOE's Office of Science provides ongoing support for research  
2288 in molecular genetics, genome sequencing and microbiology, and in emerging disciplines such as  
2289 bioinformatics and structural biology.

2290  
2291 The agency's original survey response listed 26 primarily educational activities that targeted a variety of  
2292 audiences, including underserved populations, the judiciary, and academia. Among the highlights of 13  
2293 years of DOE educational efforts was a series of 38 workshops geared to the judiciary. At the workshops,  
2294 judges explored the fundamentals of genetics and discussed some of the expected ethical, legal, and social  
2295 challenges that were anticipated to lead to court cases, policy and rule making, or new legislation related  
2296 to genomics. In addition to the workshops geared to judges, the DOE also supported many programs that  
2297 provided outreach to communities and to schools.

2298  
2299 By 2008, DOE had established two training programs for professionals at the DOE Joint Genome  
2300 Institute (JGI). One of the JGI programs provides a system for incorporating genomics research into  
2301 undergraduate courses. The second program is a joint effort of the American Society of Microbiology and  
2302 DOE-JGI that introduces basic bioinformatics to undergraduate faculty.

2303  
2304 DOE has numerous educational websites related to genomics, which are aimed at practicing  
2305 professionals, K-12 teachers and students, and graduate students. These and other educational resources  
2306 about genomics can be found at the JGI website, [www.jgi.doe.gov/education](http://www.jgi.doe.gov/education).

2307  
2308 DOE has been evaluating the impact of its education programs in collaboration with the Oak Ridge  
2309 Institute for Science and Education (ORISE). Surveys conducted and analyzed by ORISE indicate that  
2310 JGI programs are addressing an unmet need for research opportunities for undergraduates and faculty  
2311 development, and allow faculty and students to contribute new knowledge to DOE science. DOE has  
2312 plans to expand its programs to include building similar tools for metagenome and eukaryotic genome  
2313 analyses so that students and faculty can participate in the full range of DOE mission-related genomics  
2314 research. (See Appendix E-DOE for details of additional projects.)

### 2315 2316 **HRSA**

2317  
2318 HRSA's mission is to improve and expand access to quality health care for all through the adequate  
2319 provision of primary care services. To comply with this core mission, HRSA supports ongoing genetics  
2320 education and training activities for health care professionals with the goal of decreasing health disparities  
2321 by improving access to quality health care.

2322  
2323 In 2003, HRSA reported 64 genetics educational activities. Several of HRSA activities have been co-  
2324 funded with other HHS agencies including NIH, CDC, and AHRQ. HRSA and NIH activities primarily  
2325 are geared to addressing issues relating to the education and training of practicing health care  
2326 professionals, graduate students, residents, and fellows. For example, HRSA has provided ongoing  
2327 funding for Area Health Education Centers (AHEC) to provide community-based CE programs to health  
2328 professionals that include a component with genetics content.

2329  
2330 The criteria that HRSA used to determine which genetics training and education activities to undertake  
2331 included a focus on emerging areas of public health significance, such as genetics and bioterrorism; an  
2332 interdisciplinary focus on the translation of genetic knowledge into practice and research; the applicability  
2333 of genetics across disciplines; and the need to educate the public about genetic services and genetic  
2334 testing.

2335

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2336 In 2008, the HRSA survey response noted an expanded number of activities in genetics education and  
2337 training and listed several divisions within HRSA that have a role or responsibility for such programs.  
2338 These programs aim to educate professionals or trainees about genetics and genomics, and include  
2339 programs in the Maternal and Child Health Bureau and the Bureau of Health Professions. (See Appendix  
2340 E-HRSA for details of these programs.)

2341  
2342 HRSA developed targeted educational products from 2003 to 2009 that include web-based materials,  
2343 newsletters, workshops, and printed materials about genetics geared specifically to primary care  
2344 providers, state newborn screening programs, the general public, dietitians, physician assistants, nurses,  
2345 patients, speech pathologists, and dentists. HRSA has also developed products for all audiences on  
2346 family history, and core competencies in genetics, genetics and common diseases, and genetics, race, and  
2347 health care.

2348  
2349 HRSA participated in several projects between 2003 to 2006 evaluating and assessing professional  
2350 knowledge about genetics and genomics and analyzing the genetics workforce.<sup>322 323</sup> HRSA has also  
2351 conducted more recent genetic workforce analysis, *Assessing Genetic Services and the Health*  
2352 *Workforce*,<sup>324</sup> to aid in identifying and planning for supply and demand needs for 2010 and beyond. This  
2353 analysis enhanced understanding of clinical genetics services, factors affecting demand for genetic  
2354 services, and the roles of health professionals providing these services. Additional activities reported in  
2355 2008 include providing reviews of journal articles related to genetics and genomics and participating in  
2356 advisory and editorial boards (see Appendix E-HRSA).

2357  
2358 Through the 2008 Newborn Screening Saves Lives Act (Pub. L. No. 110-204),<sup>325</sup> HRSA was charged, in  
2359 consultation with NIH and CDC, to establish and maintain a central clearinghouse of educational  
2360 information, family support and services information, resources, research, and data on newborn screening.  
2361 The Act authorized funding and the project is being developed by the Genetic Alliance,<sup>326</sup> partnering with  
2362 the National Newborn Screening and Genetics Research Center, Genetics and Newborn Screening  
2363 Regional Collaborative Groups, March of Dimes, and the Association of Public Health Laboratories.

### 2364 **NIH**

2365  
2366  
2367 NIH is the steward of medical and behavioral research for the Nation. Its mission is science in pursuit of  
2368 fundamental knowledge about the nature and behavior of living systems, and the application of that  
2369 knowledge to extend healthy life and reduce the burdens of illness and disability. NIH accomplishes this  
2370 mission by funding basic research and training for scientists. Training health professionals in the area of  
2371 genetics is essential to ensure that research findings in the rapidly expanding field of genetics and  
2372 genomics are translated into health practice.

2373  
2374 Most NIH training activities in genetics focus on improving basic and clinical genetics research to benefit  
2375 the general public and improve health. Some of the institutes and centers at NIH also provide training in  
2376 the area of clinical genetics, including the National Cancer Institute (NCI), National Human Genome

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<sup>322</sup> Cooksey, J.A., Forte, G., Benkendorf, J., and Blitzer, M.G. (2005). The state of the medical geneticist workforce: Findings of the 2003 survey of ABMG certified geneticists. *Genetics in Medicine*. 7(6): 439-443.

<sup>323</sup> Cooksey, J.A., Forte, G., Flanagan, P., Benkendorf, J., and Blitzer, M.G. (2006). The Medical Geneticist Workforce: An Analysis of Clinical Subgroups. *Genetics in Medicine*. 8(10): 603-614.

<sup>324</sup> University of Maryland School of Medicine. *Assessing Genetic Services and the Health Workforce*. See [http://medschool.umaryland.edu/ghsrc/research\\_assessing.asp](http://medschool.umaryland.edu/ghsrc/research_assessing.asp). Accessed on November 30, 2009.

<sup>325</sup> U.S. Government Printing Office. *Public Law 110-204 – Newborn Screening Saves Lives Act of 2007*. See <http://www.gpo.gov/fdsys/pkg/PLAW-110publ204/html/PLAW-110publ204.htm>. Accessed on February 23, 2010.

<sup>326</sup> The Genetic Alliance website. See <http://www.geneticalliance.org/about>. Accessed on February 23, 2010.

2377 Research Institute (NHGRI), and the National Institute for Child Health and Human Development  
2378 (NICHD).

2379  
2380 NIH reported in its 2003 survey response that it had funded a number of different workshops and had  
2381 developed educational tools geared to helping clinicians learn more about the impact of genetics on their  
2382 practice. An example of this effort was the development of a series of articles on genomics medicine  
2383 published in the *New England Journal of Medicine* from 2002 to 2004.<sup>327</sup> This series included articles  
2384 about population screening and the ethical, legal, and social implications of genomics and genomic  
2385 medicine. Workforce assessment activities were also carried out at NIH, including the HRSA/NIH co-  
2386 funded study, *Assessing Genetic Services and the Health Workforce*, which was conducted by HRSA's  
2387 National Center for Health Workforce Analysis.

2388  
2389 To support genetics training of health professionals and to address the translational aspects of genomics,  
2390 NIH, along with the American Medical Association and American Nurses Association, helped form the  
2391 National Coalition for Health Professional Education in Genetics (NCHPEG).<sup>328</sup> In addition, NIH and  
2392 HRSA funded a national study of the delivery of genetics services, and the roles of geneticists and other  
2393 health professionals in service delivery. This study described the existing and emerging health care  
2394 models for providing genetics services, the genetics specialist workforce, the role of primary care  
2395 physicians and other clinicians in genetic services, and factors influencing the supply and demand for  
2396 genetic services across the country.

2397  
2398 By 2008, NIH's genetics training and educational activities included trans-NIH programs administered by  
2399 the Office of Strategic Coordination. Individual institutes at NIH also have developed genetics training  
2400 and education programs. (See Appendix E-NIH for a listing of trans-NIH programs and individual  
2401 institutes involved in these training programs.)

2402  
2403 The trans-NIH programs were developed after passage of the NIH Reform Act of 2006 that established a  
2404 Common Fund (CF) to support programs that might not otherwise be funded by a single institute or center  
2405 due to their cross-cutting and potentially risky nature, but whose outcomes are expected to have  
2406 exceptionally high impact on the scientific community. Several Common Fund programs support  
2407 activities involving training and education in genetics and genomics. Two such programs, the National  
2408 Centers for Biomedical Computing (NCBCs) and the Interdisciplinary Research (IR) program, support a  
2409 number of extramural activities relating to genetics and genomics.

#### 2410 2411 **NIH Individual Institute Programs**

2412  
2413 **NCI:** NCI reported three programs aimed at educating professionals and trainees about genetics or  
2414 genomics in their 2008 survey response.<sup>329</sup> One of the projects supported by NCI, Genetics Related  
2415 Market Research, was conducted in conjunction with the Trans-NIH Communications Group on Genetics  
2416 and Common Diseases to help understand public perceptions about genetic testing and the rapidly  
2417 growing area of direct-to-consumer genetic testing. NCI also has developed a wide range of web-based  
2418 resources focused on genetics and genomics specifically designed for health professionals. These tools  
2419 can be accessed from the Cancer Genetics website and include cancer risk assessments and a link to the  
2420 DHHS Family History page.<sup>330</sup>

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<sup>327</sup> Guttmacher A.E., and Collins, F.S. eds (2002-2003). Genomic Medicine. *The New England Journal of Medicine*. See <http://content.nejm.org/misc/genmed.shtml>. Accessed on November 30, 2009.

<sup>328</sup> National Coalition for Health Professional Education in Genetics. See [www.nchpeg.org](http://www.nchpeg.org). Accessed on November 19, 2009.

<sup>329</sup> R25CA066061: "Advanced Cancer Risk Counseling Training for Nurses"; R25CA075131: "Clinical Cancer Genetics Education"; and R25CA093426: "Genetics Short Course for Cancer Nurses".

<sup>330</sup> National Cancer Institute. *Cancer Genetics*. See [www.cancer.gov/cancertopics/prevention-genetics-causes/genetics](http://www.cancer.gov/cancertopics/prevention-genetics-causes/genetics). Accessed on November 19, 2009.

2421 **NHGRI:** NHGRI listed several activities related to genetics education and training in the 2008 survey.  
2422 These activities include the development of educational resources to promote nursing and physician  
2423 assistant education; the Genomic Health Care Commons, a web-based interactive education resource to  
2424 support groups engaged in trans-disciplinary resource development within the nursing and physician  
2425 assistant communities; and the organization of meetings. (See Appendix E-NHGRI for details about these  
2426 programs and meetings.)

2427  
2428 **National Institute on Deafness and Other Communication Disorders (NIDCD):** NIDCD, in  
2429 conjunction with NHGRI, co-funded a Summer Program in Genetics for Audiology Faculty in 2006. This  
2430 program was designed to improve training of future audiologists in the clinical, technical, ethical, social,  
2431 and legal issues surrounding the provision of genetic services and molecular testing for hereditary types  
2432 of hearing loss. This program also included a comprehensive evaluation component to determine its  
2433 effectiveness. The results of the program were used as a model for development of a CE online course on  
2434 genetics and hearing loss that is currently taught through Gallaudet University. (See Appendix E-NIDCD  
2435 for details about this program.)

2436  
2437 **National Institute of Dental and Craniofacial Research (NIDCR):** NIDCR has been conducting  
2438 assessments of professional knowledge about genetics or genomics since 2001. The assessments have  
2439 examined core competencies in genetics and the status of genetics education in U.S. dental schools, and  
2440 included focus group research with dental professionals that also evaluated the genetics workforce in  
2441 dentistry. NIDCR reported a number of conferences, presentations, workforce assessments, and  
2442 publications relating to genetics and dentistry. For example, along with the Josiah Macy Jr. Foundation  
2443 and the American Dental Education Association, NIDCR provided conference funding for a major study  
2444 initiative, “New Models of Dental Education.” This initiative convened a panel in February 2007 that  
2445 examined the implications of genetics in clinical dental practice and education. NIDCR also has  
2446 developed online resources and educational websites, and provides outreach and education in dental  
2447 genetics to the dental practice and dental education communities on an ongoing basis. (See Appendix E-  
2448 NIDCR for details about these programs.)

2449  
2450 **National Institute on Drug Abuse (NIDA):** NIDA identified the Division of Basic Neuroscience and  
2451 Behavioral Research and the Office of Science Policy and Communications as having primary  
2452 responsibility for genetics education and training. Activities at NIDA include a *Research Education Grant*  
2453 *for Statistical Training in the Genetics of Addiction* and support for a number of meetings aimed to  
2454 educate professionals and trainees about genetics or genomics. (Details of meeting support can be found  
2455 in Appendix E-NIDA.)

2456  
2457 Genetics research has tremendously increased understanding of biological processes and the mechanisms  
2458 underlying addiction. However, the sudden expansion of information has created a critical need for  
2459 interdisciplinary research education in statistical genetics and computational methods. The Research  
2460 Education Grant was intended to address this need by training pre- and post-doctoral students in the  
2461 genetics of substance use and abuse, and by encouraging development of new, useful, and innovative  
2462 statistical methods to analyze the vast and ever increasing body of genetic data. The final phase of the  
2463 project involves disseminating the course materials through workshops, webcasts, and web pods, and  
2464 developing software user guides to the wider community of substance abuse researchers.

2465  
2466 **National Institute on Aging (NIA):** Education and training of biomedical researchers and dissemination  
2467 of scientific information to diverse audiences, including health professionals and the general public, is a  
2468 priority for NIA as articulated in its Strategic Directions. Between 2003 and 2009, NIA supported one  
2469 institutional training grant award entitled, *Neurobehavior, Neuroendocrinology and Genetics of*  
2470 *Alzheimer Disease*. This project—whose goals were to increase the understanding of the pathogenesis of

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2471 Alzheimer disease and foster development of new therapeutic approaches—provided post-doctoral  
2472 training in clinical research.

2473  
2474 **National Library of Medicine (NLM):** NLM supports three genetics training and education programs at  
2475 the National Center for Biotechnology Information (NCBI), the Lister Hill National Center for  
2476 Biomedical Communications (LHNCBC), and through an NLM extramural, university-based program.

2477  
2478 **NCBI:** More than one million users access NCBI daily, thousands of whom make use of NCBI's  
2479 genomics or biomedical literature databases. The NCBI program, *Training and Support of NCBI*  
2480 *Sequence and Genomic Information Resources*, provides training so that users can effectively and  
2481 efficiently utilize NLM's online molecular biology and genomic resources. In addition, specific  
2482 training courses at NIH, and periodically at sites across the country, have been offered to  
2483 familiarize users with the range of genomics-related data at NCBI and train researchers in the  
2484 operation and application of the analysis tools to molecular biology research. Interest in the  
2485 courses offered nearly always exceeded the manpower available for teaching and, in each year of  
2486 the program, from 2002 through 2007, approximately 6,000 participants registered for  
2487 approximately 150 courses.

2488  
2489 Although NCBI reports a 10-percent increase in use of its data resources, it notes that future  
2490 needs include providing specialized training on advanced tools (e.g., use of programming  
2491 languages for large-scale data analyses) and more sophisticated tracking through web log analysis  
2492 of how NCBI data resources are used. This analysis would help determine actual use of resources  
2493 and how changes in web page presentation affect usage patterns.

2494  
2495 **Lister Hill National Center for Biomedical Communications (LHNCBC):** Because rapid  
2496 advances in genetics research are impacting the health and medical needs of the public, the  
2497 nonexpert citizen has an increasing need for information written in nontechnical terms.  
2498 Recognizing this need, LHNCBC began development of the Genetics Home Reference website in  
2499 2001.<sup>331</sup> This website addresses NLM's goal of advancing scientific knowledge in molecular  
2500 biology by providing information about hereditary conditions and their underlying genetic causes  
2501 in a consumer friendly format. Usage statistics for the website show a continuous increase in  
2502 users over the five years since it was launched in 2003, with more than 2.7 million users in 2008.  
2503 LHNCBC continues to investigate a variety of ways to make the results of the Human Genome  
2504 Project more readily available to the public through the Genetics Home Reference website and  
2505 will continue to add new content and new features. Existing materials are reviewed and updated  
2506 on a regular basis.

2507  
2508 **NLM Extramural Program:** Since 1972, NLM has provided ongoing funding for *NLM*  
2509 *University-based Biomedical Informatics Research Training Programs*. These training programs,  
2510 conducted at various universities nationwide, address the need for training informatics researchers  
2511 and practitioners in the representation, management, and delivery of biomedical knowledge.  
2512 Genomics training is a small component of the informatics training, but a more prominent  
2513 component in four programs that focus on bioinformatics. An assessment of the NLM training  
2514 programs was completed in 2008 and is now under analysis by the program director. (See  
2515 Appendix E-NLM for additional information on NLM programs.)  
2516

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<sup>331</sup> National Library of Medicine. Genetics Home Reference: See <http://ghr.nlm.nih.gov/>. Accessed on November 19, 2009.

2517 Agencies Responding Only to the 2008 Survey

2518

2519 Several *ex officio* agencies that did not respond to the 2003 survey did provide a response to the 2008  
2520 survey. These agencies include CMS, FTC, EEOC, and NSF. Summaries of their reported genetics  
2521 education and training activities follow.

2522

2523 **CMS**

2524

2525 CMS regulates all laboratory testing (except research) performed on humans in the United States through  
2526 the Clinical Laboratory Improvement Amendments (CLIA), enacted by Congress to ensure the accuracy  
2527 and reliability of all laboratory testing. CLIA established three categories of laboratory tests: waived tests,  
2528 moderate-complexity tests, and high-complexity tests. Moderate- and high-complexity testing, which  
2529 includes genetic tests, is subject to regulations that set minimum qualifications for all persons performing  
2530 or supervising these tests and require laboratories to participate in approved proficiency testing programs,  
2531 which provide an external evaluation of the accuracy of the laboratory's test results.

2532

2533 The Division of Laboratory Services, under the Center for Medicaid and State Operations, has the  
2534 responsibility for implementing the CLIA Program and is the only division within CMS that has reported  
2535 activities in genetics education and training. This training is geared to the surveyors overseeing genetic  
2536 testing and CLIA compliance at laboratories nationwide.

2537

2538 From October to November 2007, a Basic Surveyors Training program was provided for new and current  
2539 State Agency and Regional Office surveyors. The purpose of the week-long program was to provide CMS  
2540 surveyors the proper materials and training needed to assess a genetic testing laboratory for CLIA  
2541 compliance. The surveyor training included two sessions that addressed current genetic testing  
2542 technologies and the CMS survey process for genetic testing laboratories. Evaluations of these sessions  
2543 were highly favorable and were used to determine the next basic training agenda and to plan for  
2544 additional training programs.

2545

2546 **FTC**

2547

2548 FTC deals with issues that touch the economic lives of Americans and is the only Federal agency with  
2549 jurisdiction over consumer protection. Among its many activities, FTC advances consumers' interests and  
2550 creates practical and plain-language educational programs for consumers and businesses in a global  
2551 marketplace with constantly changing technologies.

2552

2553 As part of its mission to regulate unfair and deceptive practices, FTC cooperated with FDA and CDC in  
2554 2006 to develop a fact sheet for consumers to educate them about the limitations of direct-to-consumer  
2555 genetic tests. The fact sheet, *At-Home Genetic Tests: A Healthy Dose of Skepticism May be the Best  
2556 Prescription*,<sup>332</sup> provides consumers with clear information to make well-informed decisions when  
2557 considering whether to purchase direct-to-consumer (DTC) genetic tests and answers questions about the  
2558 usefulness of such tests. More than 16,000 copies of the print version of the consumer fact sheet have  
2559 been distributed since July 2006. The fact sheet is also available on the FTC website where it has been  
2560 accessed more than 18,000 times since 2006. FTC will continue to evaluate the need for consumer  
2561 education about DTC genetic tests and will also monitor consumer-directed advertising of genetic tests  
2562 and take action, where necessary, to prevent consumer deception.

2563

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<sup>332</sup> Federal Trade Commission. *Facts for Consumers, At-Home Genetic Tests: A Healthy Dose of Skepticism May Be the Best Prescription*, July 2006. See [www.ftc.gov/bcp/edu/pubs/consumer/health/hea02.shtm](http://www.ftc.gov/bcp/edu/pubs/consumer/health/hea02.shtm). Accessed on November 19, 2009.

2564 **EEOC**

2565  
2566 EEOC is responsible for enforcing Federal laws that make it illegal for employers to discriminate against  
2567 a job applicant or an employee because of the person's race, color, national origin, sex, age, religion, or  
2568 disability. With the passage of the Genetic Information Nondiscrimination Act (GINA) in 2008,  
2569 discrimination protections now include discrimination against individuals because of genetic information.

2570  
2571 EEOC genetics education and online resources include detailed information about Title II of GINA, and  
2572 this agency provides training on the legal prohibitions against employment discrimination on the basis of  
2573 genetic information. (See Appendix E-EEOC for a listing of these trainings.)  
2574

2575 Once the regulations implementing Title II of GINA become final, EEOC plans on conducting additional  
2576 training sessions on the legal requirements of Title II for lawyers, human resource professionals, small  
2577 business owners, and other interested parties.  
2578

2579 **NSF**

2580  
2581 NSF is an independent Federal agency created by Congress in 1950 to promote the progress of science; to  
2582 advance national health, prosperity, and welfare; and to secure national defense. The agency is tasked  
2583 with keeping the United States at the leading edge of scientific discovery. Therefore, in addition to  
2584 funding research in the traditional academic areas, the agency also supports "high-risk, high pay-off"  
2585 ideas, novel collaborations, and numerous projects. The agency's mission is to ensure that the research it  
2586 supports is fully integrated with education so that today's revolutionary work will also be training  
2587 tomorrow's top scientists and engineers.  
2588

2589 In 2008, the agency reported that the Directorate of Education and Human Resources, in collaboration  
2590 with the Directorate for Biological Sciences, administers approximately 50 active awards that directly or  
2591 indirectly promote genetics or genomics education for K-12, undergraduate, or graduate students, or for  
2592 the general public. Although NSF has no programs that specifically target genetics education, there is the  
2593 recognition that genetics is vital to an understanding of general biology, as well as workforce issues such  
2594 as biotechnology training. Current awards thus include projects that indirectly address genetics and  
2595 genomics while targeting a broad range of topics in biology such as molecular/cellular biology, evolution,  
2596 biodiversity, and ecology.  
2597

2598 Many of the Directorate of Education and Human Resources' active awards have a core objective relating  
2599 to genetics and/or genomics. Examples of projects funded by NSF in 2008 and beyond include *Literature-*  
2600 *Based Scientific Learning in Genetics*, *The Community College Genomics Research Initiative*, *Proteomics*  
2601 *and Functional Genomics Scholarship Program*, and *Pre-doctoral Training in Functional Genomics of*  
2602 *Model Organisms* (see Appendix E-NSF for a detailed listing of programs, programmatic goals and  
2603 individual NSF funded projects with core objectives relating to genetics and genomics).  
2604

2605 **D. Summary**

2606  
2607 To obtain information about Federal activities relating to genetic education and training of health  
2608 professionals and consumers in genetics and genomics, surveys were distributed to SACGHS *ex officio*  
2609 agencies in 2003 and again in 2008. Four agencies reported activities only in 2008; five agencies replied  
2610 to both survey requests reporting no activities relevant to the survey questions; and six agencies  
2611 responded to both surveys, allowing comparative analysis of the growth of activities in these agencies in  
2612 the intervening years. Not surprisingly, there was a significant expansion of activities related to genetic

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2613 and genomic education and an increased emphasis on activities directed to the public. A brief comparison  
2614 of responses from the 2003 and 2008 surveys for the agencies that responded to both surveys follows:  
2615

- 2616 • CDC has been very active in genetics education efforts and funded a number of activities in  
2617 2003. Although it has continued to expand its role in genetics and genomics education and  
2618 collaborate with a number of entities, it reported in 2008 that due to limited available resources  
2619 the agency is unable to develop this area fully.
- 2620 • DOC's reported activities related to genetics and genomics reside within the National Institute of  
2621 Standards and Technology (NIST) and have expanded beyond cancer genetics and forensic  
2622 applications targeted to practicing professionals to now include health professionals in training.  
2623 NIST has also broadened its educational methods to include websites and online resources.
- 2624 • DOD reported activities related to health care professional education through its medical training  
2625 entities in 2003; however, by 2008, educational activities had broadened significantly with  
2626 personalized medicine programs and a DOD-wide newborn screening program that includes  
2627 education of health care professionals and parents.
- 2628 • In 2003, DOE had already been heavily involved in genetic and genomic education activities as a  
2629 result of its participation in the Human Genome Project. By 2008, DOE had established the Joint  
2630 Genome Institute to incorporate genomic research into undergraduate courses and websites  
2631 aimed at practicing professionals, K-12 teachers and students, and graduate students.
- 2632 • Although HRSA reported 64 educational activities in 2003, they were primarily targeted to  
2633 practicing health care professionals, graduate students, residents, and fellows. HRSA has  
2634 expanded its focus over the intervening years to include other health care professionals and the  
2635 general public and to produce products for all audiences on family history, newborn screening,  
2636 and the genetics of common diseases.
- 2637 • NIH reported 41 genetics education and training activities in 2003, including funding support for  
2638 the National Coalition of Health Professional Education in Genetics. By 2008, NIH had  
2639 numerous activities within individual Institutes and through some of its trans-NIH programs.  
2640 NCI, NHGRI, NIDCD, NIDCR, NIDA, NIA, and NLM all reported significant programs and  
2641 educational resources for health care professionals and consumers.

2642  
2643 The activities of the four agencies that responded only to the 2008 survey are summarized as follows:  
2644

- 2645 • CMS conducts activities in genetics education and training for surveyors who conduct laboratory  
2646 inspections under the Clinical Laboratory Improvement Amendments.
- 2647 • FTC has been working with FDA and CDC on consumer education for direct-to-consumer  
2648 genetic testing.
- 2649 • EEOC is responsible for Title II of GINA and provides education and online resources on  
2650 prohibitions against employment discrimination on the basis of genetic information.
- 2651 • NSF awards grants to promote genetics education for K-12, undergraduate and graduate students,  
2652 and the general public.

2653  
2654 Based on SACGHS surveys of Federal agencies conducted in 2003 and 2008, a considerable number of  
2655 activities in genetics and genomics have lead to development of educational programs, materials, and  
2656 resources for a variety of professional disciplines and for the public. A number of government-sponsored  
2657 websites house and/or link to government and nongovernment resources. An effort to evaluate and  
2658 consolidate these materials and maintain an entry point for their access would facilitate dissemination of  
2659 accessible, credible genetic and genomic information to health professionals and the public.

2660 **VI. Conclusions and Recommendations**

2661  
2662 **Conclusions**

2663  
2664 The Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS) examined the genetics  
2665 education and training needs of health care professionals, public health providers, and consumers and  
2666 patients through surveys, environmental scans, and literature reviews. Since its last consideration of this  
2667 area in 2004, SACGHS found that genetics education and training efforts in the private and public sectors  
2668 have increased. However, these efforts have not kept pace with the emerging understanding of the human  
2669 genome and rapid evolution of genomic technologies. The following discussion summarizes SACGHS’  
2670 findings and its recommendations that address the needs of health care professionals, public health  
2671 providers, and consumers and patients.  
2672

2673 **A. Health Care Professionals**

2674  
2675 A review of the literature and findings from SACGHS surveys of health professional organizations  
2676 revealed that much work has been done to develop genetics educational curricula and programs at the  
2677 undergraduate, graduate, and continuing education level. However, SACHGS also found that these efforts  
2678 often exist in isolation and are not always linked to accreditation, certification, and licensure programs.  
2679 Recent collaborative efforts by professional societies to align genetic competencies with educational  
2680 objectives are a promising step forward but need to be replicated and extended if progress is to continue.  
2681

2682 SACGHS found that several barriers impede incorporation of genetics and genomics into patient care.  
2683 These include the failure to update education curricula to reflect scientific advancements in genetics and  
2684 genomics, limited application of genetic concepts in clinical training, competing priorities across the  
2685 continuum of education, lack of funding to support genetics education programs, and lack of evidence  
2686 supporting clinical effectiveness. SACGHS survey respondents indicated that competing curriculum  
2687 priorities is the most significant barrier. Incorporating genetics across disciplines and topic areas is an  
2688 important way to overcome this barrier.  
2689

2690 Compounding the barriers discussed above, there is an insufficient number of M.D. and Ph.D. geneticists  
2691 available to provide genetics education to health care professionals. To address this shortage, others  
2692 trained in genetics, such as genetic counselors, pharmacists with pharmacogenomic training, and nurse  
2693 geneticists should be encouraged to step into educator roles. Genetic education programs that use trained  
2694 peer educators have been successful and well accepted by health care professionals. Also, enhancing the  
2695 use of clinical decision support tools, promoting the importance of family history, and ensuring adequate  
2696 reimbursement for genetic services are among other approaches that would support the optimal use of  
2697 genetics and genomics in health care.  
2698

2699 **B. Public Health Providers**

2700  
2701 To assess the genetics education needs of public health providers, SACGHS reviewed findings from the  
2702 literature and conducted a survey of public health providers. The survey used 12 core competencies  
2703 developed by SACGHS. Although survey respondents identified all 12 competencies as important, they  
2704 were the most confident in utilizing family history to assess predisposition to disease. This finding  
2705 suggests that some genetic-related information is accepted by public health providers and that they agree  
2706 that promoting the role of family history in population health will contribute to improved public health.  
2707

2708 The literature review and SACGHS survey revealed several barriers that limit the uptake and appropriate  
2709 use of genetic and genomic services by public health providers. These barriers include a workforce with

2710 diverse education and training needs, a significant number of public health workers trained before the  
2711 genomic era, and lack of an understanding of the need for genetics and genomics expertise in public  
2712 health practice.

2713  
2714 To address the diversity of the public health workforce, educational approaches should target the unique  
2715 training needs, and the range of expertise and genetic literacy, of each type of public health professional.  
2716 These educational approaches should include curricula on cultural competence, social and economic  
2717 determinants of health, and ways to address and reduce health disparities. Survey respondents identified  
2718 strategies to ensure that genetic services and information are available to vulnerable and underserved  
2719 populations. These strategies ranged from local-level community engagement to policy development at  
2720 the federal level. Identifying effective educational models for public health providers who serve these  
2721 communities will also help ensure that appropriate genetic services are provided to vulnerable and  
2722 underserved populations.

2723 .

### 2724 **C. Consumers and Patients**

2725

2726 SACGHS' data gathering activities found that consumers understand that there is a relationship between  
2727 genetics and health outcomes, but they generally do not understand complex traits and the contribution of  
2728 genetics to common diseases, nor do they understand how to use genetic information to optimize health.  
2729 The federal government and private-sector organizations have developed family history tools as one  
2730 means for individuals and families to gain health literacy and to take a more active role in preventing and  
2731 managing disease, particularly inherited conditions. These tools can help both consumers and health care  
2732 professionals in risk assessment, but for optimal use of this tool, electronic health records (EHRs) must be  
2733 capable of accepting family health history data that have been provided by a consumer (e.g., My Family  
2734 Health Portrait).

2735

2736 Consumers obtain information about genetics and genetic testing from a variety of sources including their  
2737 doctors and the media, particularly the Internet. Efforts to improve the quality and accessibility of web-  
2738 based resources will be important to provide information in a manner preferred by consumers.  
2739 Additionally, improved genetic and genomic knowledge among health care professionals will be needed  
2740 as consumers rely on them as trusted sources of information.

2741

2742 Given the wide range of educational levels and motivations among individuals seeking genetic and  
2743 genomic information, a variety of strategies are needed to enhance learning. These strategies include  
2744 expansion of Internet resources, toll-free hot lines, printed materials, and community-specific radio and  
2745 television programs that may be more accessible to individuals with lower literacy or who are nonEnglish  
2746 speaking. In addition, educational resources should be culturally appropriate and tailored to the specific  
2747 needs of communities and validated using certified health educational standards to ensure comprehension  
2748 by the target audience.

2749

### 2750 **D. Selected Federal Activities**

2751

2752 To obtain information about Federal activities related to genetics and genomics education and training of  
2753 health professionals and consumers, surveys were distributed to SACGHS *ex officio* agencies in 2008 and  
2754 compared to 2003 survey results. Some agencies participated in both surveys, while several others  
2755 participated only in 2003 or 2008.

2756

2757 The survey findings suggest that the number of genetics educational programs and resources established  
2758 by Federal agencies for professionals and consumers have increased over the past five years and are  
2759 helping to increase genetic knowledge and address part of the educational needs. However, as previously

2760 discussed, the data from the literature and other SACGHS surveys suggest that these efforts are not  
2761 sufficient and in particular there is a lack of attention to health disparities. For example, funding and  
2762 additional program development may be necessary to address disparities in access to consumer  
2763 educational resources and to provide educational materials that are appropriately targeted and effectively  
2764 delivered to various segments of the population. Federal efforts in professional and consumer education  
2765 should be continued as a way of improving competency in the rapidly evolving fields of genetics and  
2766 genomics.  
2767

## 2768 **Recommendations**

2769  
2770 Seven recommendations are presented that address the identified genetics education and training needs of  
2771 health care professionals, public health providers, and consumers and patients.  
2772

### 2773 **Recommendation 1**

2774  
2775 Evidence from the United States and abroad suggests inadequate genetics education of health care  
2776 professionals as a significant factor limiting the integration of genetics into clinical care. Innovative  
2777 approaches that coordinate the efforts of entities controlling health professional education and training are  
2778 needed.  
2779

- 2780 1. HHS should convene a workshop to identify innovative education and training approaches that will  
2781 promote integration of genetics and genomics into clinical care. The workshop would include  
2782 representatives of HHS agencies and other federal departments with established programs in genetic  
2783 and genomic professional education; representatives of health professional organizations engaged in  
2784 accreditation, certification, and continuing education efforts; and private sector entities that provide  
2785 genetics education. The workshop goals are to:
  - 2786 A. identify successful education and training guidelines and models that are outcomes based;
  - 2787 B. identify current funding streams for developing and promoting genetic and genomic education as  
2788 well as gaps in funding;
  - 2789 C. recommend mechanisms for expanding and enhancing the content needed to prepare health care  
2790 professionals for personalized genomic health care;
  - 2791 D. recommend mechanisms for evolving standards, certification, accreditation, and continuing  
2792 education activities to incorporate genomic content;
  - 2793 E. determine the need, and if appropriate, appoint an ongoing advisory panel to facilitate  
2794 implementation of the approaches identified during the workshop; and
  - 2795 F. publish findings and recommendations and develop a plan to monitor the outcome of these  
2796 efforts.

### 2797 **Recommendation 2**

2798  
2800 Findings in the literature and SACGHS surveys indicate that health care professionals and public health  
2801 providers serving underserved and underrepresented groups and populations face significant challenges.  
2802

- 2803 2. HHS should promote the development and implementation of targeted genetic and genomic education  
2804 and training models for health care professionals and public health providers serving underserved and  
2805 underrepresented groups and populations. Specifically, HHS should:
  - 2806 A. direct research funding to identify effective educational models for health care professionals and  
2807 public health providers in underserved communities;
  - 2808 B. identify and support programs to increase the diversity of the health care workforce in general  
2809 and the genetic-specific workforce; and

- 2810 C. ensure that consumers and representatives of rural, minority, and underserved communities  
2811 participate in the process of developing education and training models to assure that they are  
2812 culturally and linguistically appropriate and tailored to the unique needs of these diverse  
2813 communities.  
2814

2815 **Recommendation 3**  
2816

2817 The inherent diversity of the public health workforce makes it difficult to target educational efforts that  
2818 are relevant across groups. A systematic effort is needed to evaluate the composition of the public health  
2819 workforce with current job responsibilities related to genetics and genomics and to identify future  
2820 priorities, such as the potential impact of affordable genomic analysis.  
2821

- 2822 3. Tapping the expertise of its agencies with relevant missions in public health (e.g., HRSA, CDC, the  
2823 Indian Health Service, and NIH), HHS should assess the workforce to determine the number of public  
2824 health providers with responsibilities in genetics and genomics to ascertain current trends and future  
2825 needs, to identify education and training needs, and to promote leadership development in the field.  
2826 Based on this assessment, HHS should:
- 2827 A. support and encourage the incorporation of basic genetic and genomic core competencies in the  
2828 knowledge base of federal and nonfederal public health providers, and specific competencies for  
2829 those whose responsibilities require specialized genetic knowledge, such as environmental  
2830 interactions and risk assessment for population-based genomics; and
  - 2831 B. fund educational programs based on these competencies.  
2832

2833 **Recommendation 4**  
2834

2835 A significant amount of genetic-related information directed to consumers and patients exists in a variety  
2836 of formats and from a number of sources, but the quality of the content is variable. Consumers have  
2837 consistently expressed the desire for accessible, web-based genetic information that they can trust and  
2838 consider provision of these resources as a role of the federal government.  
2839

- 2840 4. HHS should endorse, fund, and maintain an Internet entry point or portal to a vetted collection of  
2841 comprehensive, accessible, and trustworthy web-based genetic information and resources for  
2842 consumers. This portal should utilize existing governmental resources (such as those developed by  
2843 NIH and CDC) in addition to new materials. The portal should have the ability to be customized by  
2844 the consumer in order to access desired information easily. HHS should assure that:
- 2845 A. these resources include scientifically validated information and/or links to credible information  
2846 regarding topics such as genetic contributions to health and disease, gene-environmental  
2847 interactions, genetic testing, and legal protections against genetic discrimination;
  - 2848 B. these resources include links to information that are not web-based such as television and radio  
2849 programs and print materials; and
  - 2850 C. the availability of this portal be promoted using a wide range of strategies from collaborating with  
2851 developers of Internet search engines to working with community leaders at the local level.  
2852 Mechanisms to alert interested persons to updates and new information should be developed.  
2853

2854 **Recommendation 5**  
2855

2856 With the vast increase in scientific knowledge stemming from genetic and genomic research and new  
2857 technologies and the increase in direct-to-consumer genetic services, educational efforts are needed to  
2858 translate this information to reach consumers of all literacy levels.  
2859

- 2860 5. HHS should support research that identifies methods that are effective for translating genetic and  
2861 genomic knowledge into information that consumers and patients can use to make health decisions.  
2862 Specifically, HHS should:  
2863 A. support research that identifies effective methods of patient and consumer communication;  
2864 B. based on this research, and to reach diverse people and communities, HHS should develop  
2865 educational programs that use a wide array of media (e.g., radio, television, print, and mobile  
2866 phones) and provide for translation of materials into locally predominant languages; and  
2867 C. support the dissemination of these educational programs and materials into science and/or health  
2868 education initiatives through collaboration with other relevant departments and agencies such as  
2869 the Department of Education and the National Science Foundation.  
2870

2871 **Recommendation 6**

2872  
2873 Family health history tools (e.g., My Family Health Portrait) are a powerful asset for consumers and  
2874 health care professionals to use in risk assessment and health promotion.  
2875

- 2876 6. HHS should support continued efforts to educate health care professionals, public health providers,  
2877 and consumers about the importance of family health history.  
2878 A. For health care professionals, HHS should support the use of family history in clinical care  
2879 through development of clinical decision support tools and mechanisms to integrate pedigrees  
2880 into electronic health records.  
2881 B. For public health providers, HHS should promote research identifying the role of family history  
2882 in population health.  
2883 C. For consumers, HHS should:  
2884 1. promote research on how consumers use family history to make health care decisions;  
2885 2. assess the effects of gathering family histories within diverse cultures and communities and  
2886 among individuals where family histories are unavailable;  
2887 3. expand public health awareness programs and patient information materials on the  
2888 importance of sharing family history information with primary care providers; and  
2889 4. promote the embedding of educational materials in family history collection tools directed to  
2890 consumers and ensure access for all by providing these tools in various formats.  
2891

2892 **Recommendation 7**

2893  
2894 Given the reality that health care professionals and the professional societies representing them are  
2895 unlikely to invest significant resources in education and training in content areas for which services are  
2896 only partially or not reimbursable, a critical step in promoting increased knowledge of genetics and  
2897 genomics among health care professionals is ensuring adequate reimbursement.  
2898

- 2899 7. In order to increase incentives and encourage investment by public and private organizations in  
2900 education and training in genetics and genomics, and to increase the willingness of health care  
2901 professionals to participate in educational programs, the Secretary should:  
2902 A. ensure adequate reimbursement for health care professional time spent in direct patient care  
2903 delivering genetic and genomic services such as interpretation of genetic tests and collecting  
2904 family history;  
2905 B. ensure adequate reimbursement for all members of interdisciplinary teams providing genetic  
2906 services and for distance consultation and telemedicine services that are used in underserved  
2907 regions; and  
2908 C. act on the recommendations in the 2006 SACGHS report *Coverage and Reimbursement of*  
2909 *Genetic Tests and Services*.

2910 **APPENDICES**

2911

2912 **Appendix A: Literature Review**

2913

2914 **1. Literature Methodologies**

2915

2916 **Databases Searched**

2917

2918 The following databases were searched via DIALOG platform for the time period: 2003-2009.  
2919 MEDLINE, ERIC (Education Resources Information Center/DOE), Social Science Citation Index,  
2920 PsycINF, Dissertation Abstracts, Social Sciences Abstracts, Education Abstracts, Biosis Previews,  
2921 Science Citation Index; EMBASE were accessed.

2922

2923 **Search Terms**

2924

2925 Specific words and phrases used in the literature search can be grouped into several categories,  
2926 recognizing that there is overlap. These categories include educational terminology, scientific terms,  
2927 social scientific terms and concepts, and terms that identify stakeholders in genetics and genomics  
2928 education.

2929

2930 Educational terminology used:

2931 EDUCATION, TRAINING, TEACHING, INSTRUCTION, CONTINUING EDUCATION

2932 LITERACY, KNOWLEDGE, COMPETENCE, LEARNING

2933 EDUCATION MODEL

2934 HEALTH EDUCATION

2935 UNIVERSITY PROGRAM, COURSE, CLASSES

2936 SYLLABUS, CURRICULUM

2937 INNOVATE

2938 METHOD

2939

2940 Scientific terms used:

2941 GENOMIC, GENETIC, HUMAN GENOME

2942 PHARMACOGENOMIC, PHARMACOGENETIC

2943 TOXICOGENOMIC, TOXICOGENETIC

2944 FORENSIC

2945 EVOLUTIONARY, EVOLUTION

2946 MOLECULAR

2947 POPULATION GENETICS

2948 EPIDEMIOLOGY

2949

2950 Social scientific terms and concepts used:

2951 ATTITUDE

2952 BELIEF

2953

2954 Stakeholders in genetics and genomics were identified using terms such as:

2955 HEALTHCARE, HEALTHCARE PROVIDER, PRIMARY CARE

2956 PROFESSIONAL, MEDICAL, PHARMACEUTICAL

2957 HEALTH SCHOOL, SCHOOL, COLLEGE, UNIVERSITY

2958 PUBLIC HEALTH

2959 STUDENT, PUPIL

2960 PATIENT  
2961 GENERAL PUBLIC, CONSUMER  
2962 PHYSICIAN ASSISTANT

2963  
2964 **2. Genetics and Genomics Competencies for Selected Health Care Providers**

2965  
2966 **National Coalition for Health Professional Education in Genetics**

2967  
2968 *At a minimum, each health care professional should be able to:*

- 2969  
2970 1. Examine one's competence of practice on a regular basis, identifying areas of strength and areas  
2971 where professional development related to genetics and genomics would be beneficial.  
2972 2. Understand that health-related genetic information can have important social and psychological  
2973 implications for individuals and families.  
2974 3. Know how and when to make a referral to a genetics professional.

2975  
2976 *In the knowledge domain, all health professionals should understand:*

- 2977  
2978 1. basic human genetics terminology,  
2979 2. the basic patterns of biological inheritance and variation, both within families and within  
2980 populations,  
2981 3. how identification of disease-associated genetic variations facilitate development of prevention,  
2982 diagnosis, and treatment options,  
2983 4. the importance of family history (minimum three generations) in assessing predisposition to  
2984 disease,  
2985 5. the interaction of genetic, environmental, and behavioral factors in predisposition to disease,  
2986 onset of disease, response to treatment, and maintenance of health,  
2987 6. the difference between clinical diagnosis of disease and identification of genetic predisposition to  
2988 disease (genetic variation is not strictly correlated with disease manifestation),  
2989 7. the various factors that influence the client's ability to use genetic information and services, for  
2990 example, ethnicity, culture, related health beliefs, ability to pay, and health literacy,  
2991 8. the potential physical and/or psychosocial benefits, limitations, and risks of genetic information  
2992 for individuals, family members, and communities,  
2993 9. the resources available to assist clients seeking genetic information or services, including the  
2994 types of genetics professionals available and their diverse responsibilities,  
2995 10. the ethical, legal and social issues related to genetic testing and recording of genetic information  
2996 (e.g., privacy, the potential for genetic discrimination in health insurance and employment), and  
2997 11. one's professional role in the referral to or provision of genetics services, and in follow-up of  
2998 those services.

2999  
3000 *In the skills domain, all health professionals should be able to:*

- 3001  
3002 1. gather genetic family history information, including at minimum a three-generation history,  
3003 2. identify and refer clients who might benefit from genetic services or from consultation with other  
3004 professionals for management of issues related to a genetic diagnosis,  
3005 3. explain effectively the reasons for and benefits of genetic services,  
3006 4. use information technology to obtain credible, current information about genetics, and  
3007 5. assure that the informed-consent process for genetic testing includes appropriate information  
3008 about the potential risks, benefits, and limitations of the test in question.  
3009

3010 ***In the attitudes domain, all health professionals should:***  
3011

- 3012 1. appreciate the sensitivity of genetic information and the need for privacy and confidentiality, and  
3013 2. seek coordination and collaboration with an interdisciplinary team of health professionals.  
3014

3015 ***Examples of the eighteen critical “minimums” in the three content areas spanning knowledge, skills,***  
3016 ***and attitude domains:***  
3017

3018 ***Basic requirements, such as understanding:***  
3019

- 3020 • basic genetic terminology,  
3021 • patterns of inheritance,  
3022 • differences between genetic inheritance and risk predisposition,  
3023 • the importance of family history,  
3024 • the role of the environment in gene-environment interactions,  
3025 • cultural and psychosocial factors,  
3026 • how to initiate and follow-through on referral for genetic services,  
3027 • recognition of available resources for patients and families,  
3028 • risks/benefits of genetic testing, and  
3029 • ethical, legal, and social implications in provision of genetics services.  
3030

3031 ***Skill-specific competencies include the ability to:***  
3032

- 3033 • accurately elicit a patient’s three-generation family history,  
3034 • identify and refer clients to relevant professionals given a genetic diagnosis,  
3035 • effectively communicate why a patient would want to consider utilizing genetic services,  
3036 • use technology to obtain accurate information about genetics, and  
3037 • ensure any informed consent process in the genetic testing process includes accurate review of  
3038 risks, benefits and limits of test being considered.  
3039

3040 ***Attitude-specific requirements outline that health care professionals should be able to:***  
3041

- 3042 • appreciate the need for privacy and confidentiality when working with a patient about their  
3043 genetic information, and  
3044 • preemptively seek interdisciplinary collaboration with other health care professionals when  
3045 providing, discussing, or initiating genetic services for a client.  
3046

### 3047 **Competencies for Physicians**

3048

3049 As part of the Association of American Medical Colleges’ (AAMC) 2004 Medical School Objectives  
3050 Project, 21 learning objectives in genetics were established across attitude, knowledge, and skill  
3051 domains.<sup>333</sup> In January 2010, the AAMC and the Association of Professors of Human and Medical  
3052 Genetics jointly developed Core Competencies for Medical School Genetics Education providing  
3053 recommendations on the fundamental genetics principles that should be demonstrated by all medical  
3054 school graduates.<sup>334</sup> This updated set of competencies conforms to requirements of the Liaison

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<sup>333</sup> Association of American Medical Colleges. (2004). *Report VI Contemporary Issues in Medicine: Genetics Education*. See [www.ttuhscc.edu/som/curriculum/documents/msop\\_genetics.pdf](http://www.ttuhscc.edu/som/curriculum/documents/msop_genetics.pdf). Accessed on November 24, 2009.

<sup>334</sup> Association of Professors of Human and Medical Genetics. *APHMG Medical School Core Curriculum in Genetics*. See <http://www.aphmg.org/>. Accessed on March 31, 2010.

3055 Committee on Medical Education (LCME) that it be mapped to educational objects set forth by the  
3056 Accreditation Council for Graduate Medical Education and broadly categorize as:

- 3057
- 3058 • Organization of the genome and regulation of gene expression as it relates to medical genetic  
3059 diagnosis;
  - 3060 • Genetic variation and the implications for diversity of normal variation and disease;
  - 3061 • Principles of inheritance patterns;
  - 3062 • Clinical, ethical and social implications for diagnosis, family health, prediction, and personalized  
3063 medicine;
  - 3064 • Importance of genetic testing including cytogenetics, molecular genetics, genome sequencing,  
3065 and biochemical genetics;
  - 3066 • Unique features of the genetics for cancer and prenatal diagnosis; and
  - 3067 • Treatment of genetic conditions including family counseling.
- 3068

3069 In 2009, AAMC collaborated with the Howard Hughes Medical Institute and released a report on updated  
3070 expected competencies for graduating physicians and pre-medical program students.<sup>335</sup> Medical school  
3071 competencies span eight domains; those specific to genetics include knowledge and competent  
3072 application of “individual and population-based genetics and genomics to guide medical care decisions.”  
3073 Many subcomponent competencies have genetic and genomic elements such as pharmacogenomics and  
3074 pharmacogenetics, and the analytical validity, clinical validity, and clinical utility of genetic tests.

3075

3076 From 2000 to 2008, ACMG published numerous condition-specific medical practice and diagnostic  
3077 evaluation guidelines specific to single-gene disorders, including guidelines for genetic susceptibility to  
3078 breast and ovarian cancer, carrier screening for spinal muscular atrophy, carrier screening for Ashkenazi  
3079 Jewish individuals, genetic testing for colon cancer, and many others.<sup>336</sup>

3080

3081 These clinical guidelines and practice standards have helped shape practice uniformity with respect to  
3082 work-ups for common genetic conditions across primary care, pediatrics, oncology, obstetrics, and  
3083 psychiatric clinical settings. Many of these clinical guidelines were released jointly with the American  
3084 Society of Human Genetics (ASHG).

3085

3086 Reflecting the scientific progress beyond single-gene disorders, in January 2007, ASHG released policy  
3087 recommendations concerning DTC genetic testing technologies.<sup>337</sup> The scope of this policy statement  
3088 pertained to health-related DTC testing, but the overall policy outlined specific issues that health care  
3089 providers should be mindful of when interacting with patients who use DTC genetic tests for complex  
3090 disease susceptibility determinations (e.g., diabetes, heart disease, depression, and cancer). ASHG’s  
3091 primary recommendation concerning health care professionals indicated that professional societies would  
3092 need to assume a greater level of responsibility in educating their members about this type of genetic  
3093 testing.

3094

3095 Many professional societies have released or revised practice competency standards or policies focused  
3096 on genetics and genomics. For example:

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<sup>335</sup> Howard Hughes Medical Institute-Association of American Medical Colleges. (2009). *Scientific Foundations for Future Physicians: Report of the HHMI-AAMC Committee*. See [www.hhmi.org/grants/pdf/08-209\\_AAMC-HHMI\\_report.pdf](http://www.hhmi.org/grants/pdf/08-209_AAMC-HHMI_report.pdf). Accessed on November 24, 2009.

<sup>336</sup> American College of Medical Genetics. *Practice Guidelines*. See [www.acmg.net/AM/Template.cfm?Section=Practice\\_Guidelines&Template=/CM/HTMLDisplay.cfm&ContentID=3701](http://www.acmg.net/AM/Template.cfm?Section=Practice_Guidelines&Template=/CM/HTMLDisplay.cfm&ContentID=3701). Accessed on November 24, 2009.

<sup>337</sup> Hudson, K., Javitt, G., Burke, W., and Byers, P., along with the American Society of Human Genetics Social Issues Committee. (2007). ASHG Statement on direct-to-consumer testing in the United States. *The American Journal of Human Genetics*. 81:635-637.

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

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- In 2008 the American Academy of Family Physicians released a medical genetics core competency guideline document for residency training.<sup>338</sup> 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.<sup>339</sup>
  - The American Academy of Pediatrics' Committee on Genetics has authored numerous policy and professional practice statements on various heritable and complex health conditions.<sup>340</sup>
  - 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.<sup>341</sup>
- 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.<sup>342</sup> 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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<sup>338</sup> American Academy of Family Physicians. *Recommended Curriculum Guidelines for Family Medicine Residents, Medical Genetics*. See [www.aafp.org/online/etc/medialib/aafp\\_org/documents/about/rap/curriculum/medical\\_genetics.Par.0001.File.tmp/medicalgenetics.pdf](http://www.aafp.org/online/etc/medialib/aafp_org/documents/about/rap/curriculum/medical_genetics.Par.0001.File.tmp/medicalgenetics.pdf). Accessed on November 16, 2009.

<sup>339</sup> American Medical Association Policy Database. See <http://www.ama-assn.org/ama/pub/advocacy/policy-finder.shtml>. Accessed on January 14, 2010.

<sup>340</sup> American Academy of Pediatrics, Committee on Genetics. See [www.aap.org/visit/cmtel18.htm#statements](http://www.aap.org/visit/cmtel18.htm#statements). Accessed on November 16, 2009.

<sup>341</sup> American Society of Clinical Oncology. *Guidelines*. See [www.asco.org/ASCOv2/Practice+&+Guidelines/Guidelines](http://www.asco.org/ASCOv2/Practice+&+Guidelines/Guidelines). Accessed on November 16, 2009.

<sup>342</sup> Kemper, A.R., Trotter, T.L., Loyd-Puryear, M.A., Kyler, P., Feero, W.G., and Howell, R.R. (2010). A blueprint for maternal and child health primary care physician education in medical genetics and genomic medicine: recommendations of the United States Secretary for Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children. *Genetics in Medicine*. 12(2):77-80.

- 3133           • How to communicate information about risk of conditions to women before pregnancy and when  
3134           pregnant; and  
3135           • When and how to refer families to a genetic counselor or geneticist.  
3136

3137 Workshop participants identified the lack of time as the most important barrier to educating primary care  
3138 providers in genomic medicine for both those in training and those in practice. Lack of geneticists to  
3139 provide education, mentoring, and curricular oversight in residency programs and lack of enthusiasm  
3140 about genetics and genomic medicine by trainees and those in practice limit effective educational efforts.  
3141

3142 To address the issues identified during the workshop, the recommendations summarized below, were  
3143 made and subsequently adopted by the Advisory Committee on Heritable Disorders in Newborns and  
3144 Children:  
3145

- 3146           • Develop a case-based genetics and genomic medicine educational curriculum that could be  
3147           incorporated into residency training programs that presents common genetic concepts using  
3148           scenarios.  
3149           • Ensure that board certification exams assess knowledge related to core educational goals and  
3150           basic literacy in genetics and genomic medicine.  
3151           • Make available continuing medical education (CME) at meetings and through the Internet that  
3152           focuses on practical aspects of incorporating genetics and genomic medicine into primary care,  
3153           focusing on useful skills such as obtaining family history and identifying red flags for referral for  
3154           genetic counseling.  
3155           • Promote participation in these educational activities through the maintenance of board  
3156           certification process.  
3157           • Create a website that would include clinical recommendations and practical office tools to  
3158           facilitate incorporation of genetic and genomic medicine into routine practice.  
3159

3160 The workshop endorsed the development of the Genetics in Primary Care Training Institute (GPCTI)  
3161 based on the concept of a “learning collaborative”<sup>343</sup> that would pair primary care providers with experts  
3162 in genetic and genomic medicine. These learning collaboratives would develop a 1-year project that  
3163 includes an outcomes component, and the training institute would then formally evaluate these projects to  
3164 inform the process of broader dissemination. The Advisory Committee on Heritable Disorders in  
3165 Newborns and Children approved the learning collaborative concept and recommended that HRSA  
3166 provide funding for the project. HRSA is implementing this recommendation through the formation of  
3167 GPCTI and funding the initiative as a Special Project of Regional and National Significance by the  
3168 Maternal and Child Health Bureau at HRSA.  
3169

### 3170 **Competencies for Nurses**

3171  
3172 In 2005, genetics competencies for all practicing R.N.s were developed by consensus and endorsed by 49  
3173 professional organizations, encompassing four areas of clinical action: (1) correctly applying/integrating  
3174 genetic and genomic knowledge when assessing patients; (2) accurately identifying patient  
3175 genetic/genomic needs and issues; (3) conducting appropriate patient referrals; and (4) providing  
3176 competent education, clinical care and psychosocial support to patients and families.<sup>344</sup>

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<sup>343</sup> Young, P.C., Glade, G.B., Stoddard, G.J., and Norlin, C. (2006). Evaluation of a learning collaborative to improve the delivery of preventive services by pediatric practices. *Pediatrics*. 117(5):1469-1476.

<sup>344</sup> These *Essential Competencies* are currently in the 2nd edition published by the American Nurses Association in February 2009. Also, see Jenkins, K., and Calzone, K.A. (2007). Establishing the essential nursing competencies for genetics and genomics. *Journal of Nursing Scholarship*. 39(1):10-16. See [www.genome.gov/Pages/Careers/HealthProfessionalEducation/geneticscompetency.pdf](http://www.genome.gov/Pages/Careers/HealthProfessionalEducation/geneticscompetency.pdf). Accessed on November 24, 2009.

3177 ***Correctly integrating genetic and genomic knowledge encompasses the nurse's ability to:***  
3178

- 3179 • Appreciate genetics and genomics in prevention, screening, diagnostics, treatment selection,  
3180 monitoring, and clinical outcome evaluation processes
- 3181 • Collect a complete family health history
- 3182 • Accurately construct a multi-generational pedigree
- 3183 • Collect patient health histories that include genetic/genomic health information
- 3184 • Perform physical assessments that include genetic/genomic risk factors
- 3185 • Assess patient understanding of genetic/genomic information
- 3186 • Competently construct plans of health care that incorporate genetics and genomics

3187  
3188 ***Patient identification skills expected of professional nurses encompasses their ability to:***  
3189

- 3190 • Ascertain who could benefit from genetic/genomic information or services
- 3191 • Recognize accurate sources of genetic/genomic information for patients based upon their unique  
3192 health needs
- 3193 • Appreciate relevant ethical, legal, and social implications related to genetic information and  
3194 genomic technologies
- 3195 • Define issues acting against a patient's ability to autonomously and voluntarily gather relevant  
3196 genetic information and act upon findings

3197  
3198 ***Genetics and genomics health care services that all nurses are expected to provide include:***  
3199

- 3200 • Accurately interpret genetic/genomic health information (e.g., diagnostic tests, health histories)
- 3201 • Appropriately collect and review genetic/genomic health information from reliable information  
3202 sources to facilitate a patient's decisionmaking
- 3203 • Correctly apply genetics and genomics information into health promotion counseling for patients
- 3204 • Correctly use genetic/genomic health interventions to improve patient health outcomes
- 3205 • Work with other members of the multi-disciplinary clinical team, including allied health  
3206 providers and insurance companies, to provide genetics and genomics clinical care
- 3207 • Correctly use interventions and treatments that are tailored to patients' genetic/genomic health  
3208 needs
- 3209 • Correctly evaluate patient health outcomes following use of genetic/genomic health intervention  
3210 or treatment, and facilitate redirection of health care planning as necessary

3211  
3212 **Competencies for Physician Assistants**  
3213

3214 Four PA organizations represent more than 84,000 PAs, 40 percent of whom work in primary care. A  
3215 2008 survey by these PA organizations among members found that 85 percent of respondents had  
3216 gathered family history in the past six months and 70 percent indicated that they had used that  
3217 information in decisionmaking. Yet only 22 percent reported feeling that their supervising physician was  
3218 knowledgeable about genetics.

3219  
3220 A survey of PA training programs found that 81 percent perceive a need to enhance their genetics  
3221 curriculum despite an already overloaded curriculum and lack of time to develop resources.<sup>345</sup> In  
3222 response, the Physician Assistance Education Association is creating faculty development opportunities,  
3223 monitoring and reporting innovations in genetics education, developing curricula resources for best

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<sup>345</sup> 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.

3224 practices, developing assessment tools for students and faculty, and developing a database to track  
3225 genetics activities and outcomes in PA education. These professional organizational efforts use traditional  
3226 methods of dissemination—newsletters, annual conferences, journals, and web-based continuing  
3227 education activities—to educate members in genetics. Recently, an ad hoc group of clinical leaders  
3228 established the *Essential Physician Assistant Guidelines for Genetics and Genomics*.<sup>346</sup> Similar to other  
3229 professional efforts, their proposed competencies are focused on three core concepts—knowledge, skills,  
3230 and attitudes.

3231  
3232 Knowledge requirements include understanding genetics terminology, inheritance patterns, diagnostics,  
3233 family history assessment, screening, and making appropriate referrals, among other issues. PAs are  
3234 expected to have the skills to elicit family history, identify the need for referrals, provide patient  
3235 education (including providing credible sources of information), and assess the benefits and limits of  
3236 genetic tests. They are also expected to understand the sensitivity of genetic information, appreciate  
3237 psychosocial and cultural factors, and be knowledgeable about social, legal, and ethical concerns.

### 3238 3239 **Competencies for Genetic Counselors**

3240  
3241 Practiced-based competencies were issued by the American Board of Genetic Counseling in 2008.<sup>347</sup>  
3242 They focus on the need for all genetic counselors to demonstrate competency spanning four skill-based  
3243 content domains: (1) communication; (2) critical thinking; (3) interpersonal counseling and psychosocial  
3244 assessment; and (4) professional ethics and values.

3245  
3246 Professional ethics and values expected of genetics counselors include the ability to serve their profession  
3247 by maintaining expected ethical, legal and philosophical approaches valued by the genetic counseling  
3248 community; advocating for clients and families; presenting and exploring research options with clients;  
3249 accurately identifying self limitations in knowledge and practice capacities; and continually developing  
3250 professionally.

#### 3251 3252 ***Communication skills encompass the genetic counselor's need to:***

- 3253
- 3254 • establish a mutually agreeable counseling plan with clients
  - 3255 • comprehensively elicit family history information
  - 3256 • accurately obtain client medical histories in a variety of clinical settings
  - 3257 • ascertain complete social/psychosocial histories
  - 3258 • accurately convey technical medical and genomic information to clients
  - 3259 • accurately communicate reproductive options
  - 3260 • communicate all information to clients and families with cultural competence, and
  - 3261 • plan and organize professional education programs on genetics and counseling issues

#### 3262 3263 ***Critical thinking skills for genetic counselors include the ability to:***

- 3264
- 3265 • identify and calculate genetic and teratogenic predictive risks
  - 3266 • evaluate a client's social/psychosocial history
  - 3267 • integrate the entirety of a client's medical information to guide client/family counseling needs
  - 3268 • demonstrate ability to manage case portfolio needs

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<sup>346</sup> Rackover, M., Goldgar, C., Wolpert, C., Healy, K., Feiger, J., and Jenkins, J. (2007). Establishing essential physician assistant clinical competencies guidelines for genetics and genomics. *Journal of Physician Assistant Education*. 18(2):47-48.

<sup>347</sup> American Board of Genetic Counseling. *Practice Based Competencies*. See [http://abgc.iasonline.com/CMFiles/Practice\\_Based\\_Competencies\\_Aug\\_2006\\_10-29-0951KFH-10292008-6844.pdf](http://abgc.iasonline.com/CMFiles/Practice_Based_Competencies_Aug_2006_10-29-0951KFH-10292008-6844.pdf). Accessed on November 16, 2009.

- 3269
- assess a client’s capacity and ability to understand genetic information and redirect care plans accordingly, and
- 3270
- identify and access local, regional, and national clinical genetics resources for clients and families
- 3271
- 3272
- 3273

3274 ***Interpersonal counseling and psychosocial assessment involve the genetic counselor’s need to:***

3275

- 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
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- 3283

3284 **Competencies for Pharmacists**

3285

3286 Pharmacists are recognized as medication experts who improve overall patient care through partnering with physicians.<sup>348</sup> 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.”<sup>349</sup> 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.<sup>350</sup> 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.<sup>351</sup>

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3297 Although recognition of the inherited differences in drug effects was documented as early as 1931,<sup>352</sup> 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.<sup>353</sup> 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.

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<sup>348</sup> Leape, L.L., Cullen, D.J., Dempsey Clapp, M., Burdick, E., Demonaco, H.J., Erickson, J.I., and Bates, D.W. (1999). Pharmacist participation on physician rounds and adverse drug events in the intensive care unit. *Journal of the American Medical Association*. 282:267-270.

<sup>349</sup> Brock, T.P., Valgus, J.M., Smith, S.R., and Summers, K.M. (2003). Pharmacogenomics: implications and considerations for pharmacists. *Pharmacogenomics*. 4(3):321-330.

<sup>350</sup> Clemerson, J.P., Payne, K., Bissell, P., and Anderson, C. (2006). Pharmacogenetics, the next challenge for pharmacy? *Pharmacy World and Science*. 28:126-130.

<sup>351</sup> El-Ibiary, S., Cheng, C., and Alldredge, B. (2008). Potential roles for pharmacists in pharmacogenetics. *Journal of the American Pharmacology Association*. 48:e21-e32.

<sup>352</sup> Garrod, A.E. (1931). *The Inborn Factors in Disease: An Essay*. Oxford: Oxford University Press.

<sup>353</sup> Johnson J.A., Bootman, J.L., Evans, W.E., Hudson, R.A., Knoell, D., Simmons, L., Straubinger, R.M., and Meyer, S.M. (2002). Pharmacogenomics: A scientific revolution in pharmaceutical sciences and pharmacy practice. Report of the 2001-2002 Academic Affairs Committee. *American Journal of Pharmaceutical Education*. 2002;66(Winter Supplement):12S-15S.

3307 **3. Academic Preparation, Licensure, and Accreditation of Professional Schools**

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3309 **Education and Licensure of Physicians and Accreditation of Medical Schools**

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3311 In the United States there are 131 accredited medical schools granting M.D. degrees and 25 colleges of  
 3312 osteopathic medicine granting D.O. degrees.<sup>354</sup> In 2001, the Association of Professors of Human and  
 3313 Medical Genetics and ASHG released a report, “Medical School Core Curriculum in Genetics,” outlining  
 3314 critical education elements to be required in medical preparation programs.<sup>355</sup> Building on these efforts in  
 3315 2004, the AAMC (representing all medical schools, approximately 400 teaching hospitals, 68 Veterans  
 3316 Affairs departments, and 90 professional societies), reported that greater genetics training was a critical  
 3317 requirement and provided competencies.<sup>356</sup> Driving this need is a significant shortage of medical genetics  
 3318 experts prepared to address the onslaught of implications stemming from genetic science.<sup>357,358</sup>  
 3319 Subsequent analyses of issues identified in these reports confirmed that medical students’ genetic  
 3320 knowledge and competence demonstrated a need for medical schools to integrate additional training and  
 3321 education.<sup>359,360</sup>

3322

3323 A recent analysis of genetic content in graduate medical curriculums found that 77 percent of programs  
 3324 taught medical genetics only in the first year of medical school and that 47 percent failed to incorporate  
 3325 any genetic content in third and fourth year instruction.<sup>361</sup> Furthermore, only 11 percent provided  
 3326 practical clinical applications of genetics. In addition, 46 percent reported stand-alone courses only, with  
 3327 the remaining respondents offering medical genetic content built into another course. A key  
 3328 recommendation from several organizations to obtain a genetically competent physician workforce is to  
 3329 reorient undergraduate scientific foundations and integrate genetic and genomic science concepts into,  
 3330 and across, all medical education requirements.<sup>362,363</sup>

3331

3332 To obtain an M.D. professional license, students must successfully pass the United States Medical  
 3333 Licensing Examination (USMLE), a three-step examination administered by the independent medical  
 3334 licensing authority, the National Board of Medical Examiners.<sup>364</sup> Genetic content includes DNA and  
 3335 RNA concepts related to biochemistry and molecular biology coursework; congenital human  
 3336 development; Hardy-Weinberg principles; pharmacogenetics; and standard heritable conditions (e.g.,

<sup>354</sup> Association of American Medical Colleges. *Medical Schools*. See [www.aamc.org/medicalschoools.htm](http://www.aamc.org/medicalschoools.htm) and American Association of Colleges of Osteopathic Medicine. See <http://www.aacom.org/people/colleges/Pages/default.aspx>. Accessed on November 24, 2009.

<sup>355</sup> Association of Professors of Human and Medical Genetics/American Society of Human Genetics. *Medical School Core Curriculum in Genetics*. See [www.ashg.org/pdf/Medical%20School%20Core%20Curriculum%20in%20Genetics.pdf](http://www.ashg.org/pdf/Medical%20School%20Core%20Curriculum%20in%20Genetics.pdf). Accessed on November 16, 2009.

<sup>356</sup> Korf, B.R. (2005). Genetics training in the genomic era. *Current Opinion in Pediatrics*. 17(6):747-750.

<sup>357</sup> Korf, B.R. (2005). Genetics training in the genomic era. *Current Opinion in Pediatrics*. 17(6):747-750.

<sup>358</sup> Thurston, V.C., Wales, P.S., Bell, M.A., Torbeck, L., and Brokaw, J.J. (2007). The current status of medical genetics instruction in US and Canadian medical schools. *Academic Medicine*. 82(5):441-445.

<sup>359</sup> Telner, D.E., Carroll, J.C., and Talbot, Y. (2008). Genetics education in medical school: a qualitative study exploring educational experiences and needs. *Medical Teacher*. 30(2):192-198.

<sup>360</sup> Baars, M.J., Scherpier, A.J., Schuwirth, L.W., Henneman, L., Beemer, F.A., Cobben, J.M., Hennekam, R.C., Verweij, M.M., Cornel, M.C., and ten Kate, L.P. (2005). Deficient knowledge of genetics relevant for daily practice among medical students nearing graduation. *Genetics in Medicine*. 7(5):295-301.

<sup>361</sup> Thurston, V.C., Wales, P.S., Bell, M.A., Torbeck, L., and Brokaw, J.J. (2007). The current status of medical genetics instruction in US and Canadian medical schools. *Academic Medicine*. 82(5):441-445.

<sup>362</sup> Howard Hughes Medical Institute-Association of American Medical Colleges. 2009. *Scientific Foundations for Future Physicians: Report of the HHMI-AAMC Committee*. [www.hhmi.org/grants/pdf/08-209\\_AAMC-HHMI\\_report.pdf](http://www.hhmi.org/grants/pdf/08-209_AAMC-HHMI_report.pdf). Accessed on November 24, 2009.

<sup>363</sup> Spencer, A.L., Brosenitsch, T., Levine, A.S., and Kanter, S.L. (2008). Back to the basic sciences: an innovative approach to teaching senior medical students how best to integrate basic science and clinical medicine. *Academic Medicine*. 83(7):662-669.

<sup>364</sup> United States Medical Licensing Examination. *About USMLE*. See [www.usmle.org/General\\_Information/general\\_information\\_about.html](http://www.usmle.org/General_Information/general_information_about.html). Accessed on November 16, 2009.

3337 single-gene disorders, chromosomal aberrations) and skills related to their clinical management. Genetic  
3338 content is similarly incorporated in the final examination.<sup>365</sup>

3339  
3340 To obtain a D.O. license, students must successfully pass the Comprehensive Osteopathic Medical  
3341 Licensing Examination (COMLEX-USA), also a three-step examination process administered by an  
3342 independent medical licensing authority—the National Board of Osteopathic Medical Examiners. Genetic  
3343 content for COMLEX-USA is similar to USMLE, but differing approaches between M.D. and D.O.  
3344 programs result in variability of the approach to health and illness management on the examinations.  
3345 Moreover, recently emerging concepts of genomics resulting in dynamic probabilistic contexts for  
3346 chronic disease in individual patients is usually not included on these examinations.<sup>366</sup>

3347  
3348 For all physicians, state medical licensing boards require evidence of CME each year for license re-  
3349 registration, which needs to be submitted at one to four year intervals depending on the state. Great  
3350 variability exists across state medical board requirements, with some boards requiring evidence for as  
3351 little as 12 hours per year, to as many as 50 hours per year.<sup>367</sup> Although physicians may obtain genetic  
3352 education and training through pre-approved sponsored activities such as seminars, conferences, self-  
3353 learning opportunities, and other professional development activities, requirements are not tied to minimal  
3354 completion of genetic content. Genetic content across certificates is not well tracked and presumably  
3355 contains great variability in amount and type of information provided.

3356  
3357 Accreditation of U.S. medical school programs is provided through the Liaison Committee on Medical  
3358 Education (LCME) or the American Osteopathic Association (AOA). Published LCME accreditation  
3359 standards require basic science instruction and include mention of genetics, but the standards do not  
3360 outline either amounts or presence of genetics topic requirements before accreditation is issued to a  
3361 graduate medical education program.<sup>368</sup> AOA similarly addresses genetics in its accreditation processes—  
3362 presence of genetics is required under the umbrella of basic science requirements and the care of  
3363 hereditary conditions.<sup>369</sup>

### 3364 3365 **Education and Licensure of Nurses and Accreditation of Nursing Schools**

3366  
3367 As of 2004, there were more than 2.9 million nurses, of which 45.6 percent graduated from nursing  
3368 school before 1984.<sup>370</sup> Nursing contains great academic and professional heterogeneity stemming from  
3369 multiple academic pathways to becoming a R.N.; two accrediting bodies for academic curricula with  
3370 varying requirements; presence of nursing education programs that lack accreditation; and numerous  
3371 specialty advanced practice pathways with variable routes to certification (e.g., Family N.P., Pediatric  
3372 N.P., Geriatric N.P.). Genetic content is required by the American Association of Colleges of Nursing  
3373 Baccalaureate Essentials, which serve as the basis for Commission on Collegiate Nursing Education  
3374 (CCNE) accreditation. However, very little data exist to ascertain extent of genetics integration in U.S.

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<sup>365</sup> United States Medical Licensing Examination. *Content Outline*. See <http://www.usmle.org/Examinations/step1/content/principles.html>. Accessed on November 16, 2009.

<sup>366</sup> United States Medical Licensing Examination. 2009 *USMLE Bulletin* – Overview. See [www.usmle.org/General\\_Information/bulletin/2009/overview.html](http://www.usmle.org/General_Information/bulletin/2009/overview.html). Accessed November 24, 2009.

<sup>367</sup> State Medical Licensure Requirements and Statistics, 2006. Continuing Medical Education for Licensure Re-registration. See [www.ama-assn.org/ama/pub/upload/mm/455/licensurere-reg-06.pdf](http://www.ama-assn.org/ama/pub/upload/mm/455/licensurere-reg-06.pdf). Accessed on November 16, 2009.

<sup>368</sup> 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](http://www.lcme.org/functions2008jun.pdf).

<sup>369</sup> Liaison Committee on Medical Education. *Standards for Accreditation of Medical Education Programs Leading to the M.D. Degree* (2008), See [www.osteopathic.org/pdf/SB03-Standards%20of%20Accreditation%20July%202009.pdf](http://www.osteopathic.org/pdf/SB03-Standards%20of%20Accreditation%20July%202009.pdf). Accessed on November 24, 2009.

<sup>370</sup> U.S. DHHS. 2005. *The Registered Nurse Population: Preliminary Findings from the National Sample Survey of Registered Nurses, March 2004*. HRSA Bureau of Health Professions, Division of Nursing.

3375 nursing curricula. There are more than 1,600 tracked accredited nursing programs; however, there are  
3376 more schools of nursing that are not accredited and are difficult to monitor.

3377  
3378 Entry-level professional R.N.s may pursue one of four possible academic paths: (1) a four-year  
3379 baccalaureate in nursing offered by colleges or universities; (2) a two- to three-year associate degree in  
3380 nursing offered by community and junior colleges; (3) a three-year hospital-based diploma program; or  
3381 (4) as a Clinical Nurse Leader, that is, an individual who is entry-level with a B.S. in another field but  
3382 enter nursing with a master's preparation. The current trend within the nursing field; however, has been to  
3383 pair associate/diploma programs with baccalaureate institutions to increase numbers of nurses with  
3384 baccalaureate preparation. In 2006, there were 709 organizations offering bachelor's degrees, 850  
3385 organizations offering associate degrees, and 70 programs offering hospital diplomas.<sup>371</sup>

3386  
3387 Advanced Practice Nurses (N.P.s, clinical nurse specialists, certified nurse midwives, certified registered  
3388 nurse anesthetists, etc) are R.N.s who obtain a master's degree from one of the country's 448 accredited  
3389 nursing programs. Eventually expected to replace master's prepared Advanced Practice Nurses, Doctors  
3390 of Nursing Practice (D.N.P.) are R.N.s who obtain a practice-based doctoral degree from one of the  
3391 country's 92 accredited D.N.P. programs. Available since 2005, the D.N.P. represents a new movement in  
3392 nursing to incorporate greater foundations of scientific knowledge, as the D.N.P. is equivalent to other  
3393 health professional doctorates. An additional 100 schools of nursing are expected to implement D.N.P.  
3394 programs at their institutions in the near future.<sup>372</sup>

3395  
3396 The need for education of nurses in genetics is well documented.<sup>373</sup> Available figures from a subset of the  
3397 country's accredited schools of nursing published in 1999 indicated less than 10 median hours of total  
3398 genetics instruction across programs; 30 percent contained none at all.<sup>374</sup> A recent follow-up evaluation of  
3399 a small sub-sample of these schools suggests that not much progress has been made in integrating  
3400 genetics instruction hours in accredited baccalaureate, accelerated, diploma, and associate degree  
3401 programs.<sup>375</sup>

3402  
3403 A 2005 nursing faculty survey conducted by Prows, et al. found that 29 percent of schools reported no  
3404 genomic curriculum content (no change since similar data were collected in 1996), citing an already  
3405 overloaded curriculum and lack of knowledge among faculty about genetics.<sup>376</sup> The vast majority of  
3406 programs responding to the survey offered five hours or less on genetic content.

3407  
3408 Individual state boards of nursing manage and issue professional R.N. licenses; however, some states  
3409 have chosen to be part of a broader effort to streamline requirements and are members of the National  
3410 Council of State Boards of Nursing. Individuals completing an approved nursing program by state nursing  
3411 boards from baccalaureate, associate, or diploma programs must successfully complete the National  
3412 Council Licensure Examination (NCLEX) to obtain the R.N. professional license. Little genetic content is  
3413 contained in NCLEX and certification examinations, and at the master's level, there is significant  
3414 variability in exam criteria across the certifying organizations. The Genetic Nursing Credentialing

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<sup>371</sup> Bureau of Labor Statistics. *Occupational Outlook Handbook*, 2008-2009 Edition. See [www.bls.gov/oco/ocos083.htm](http://www.bls.gov/oco/ocos083.htm). Accessed on November 16, 2009.

<sup>372</sup> American Association of Colleges of Nursing. *Doctor of Nursing Practice Talking Points*. See <http://www.aacn.nche.edu/DNP/talkingpoints.htm>. Accessed on December 12, 2009.

<sup>373</sup> Prows, C.A., Glass, M., Nicol, M.J., Skiton, H., and Williams, J. (2005). Genomics in nursing education. *Journal of Nursing Scholarship*. 37(3):196-202.

<sup>374</sup> Hetteberg, C.G., Prows, C.A., Deets, C., Monsen, R.B., and Kenner, C.A. (1999). National survey of genetics content in basic nursing preparatory programs in the United States. *Nursing Outlook*. 47(4):168-180.

<sup>375</sup> Prows, C., Calzone, K., and Jenkins, J. (2006). *Genetics content in nursing curriculum*. Paper presented at the National Coalition Health Professional Education in Genetics, Bethesda, MD.

<sup>376</sup> Prows, C., Calzone, K., and Jenkins, J. (2006). *Genetics content in nursing curriculum*. Paper presented at the National Coalition Health Professional Education in Genetics, Bethesda, MD.

3415 Commission is recognized by the American Nurses Association and offers two clinical genetics specialty  
3416 certifications, one for baccalaureate R.N.s. (Genetics Clinical Nurse) and one for master's prepared nurses  
3417 (Advanced Practice Nurse in Genetics). At the time this report was written, there were 40 individuals  
3418 certified as Advanced Practice Nurse in Genetics and 11 individuals certified as Genetics Clinical  
3419 Nurse.<sup>377</sup> Nurses in genetic practice settings with direct patient, family, client, and colleague in-service  
3420 teaching responsibilities can obtain these credentials to enhance their professional portfolios.

3421  
3422 There are two bodies that accredit educational institutions and curricula for the nursing profession: the  
3423 National League for Nursing Accrediting Commission (NLNAC) and the CCNE arm of the American  
3424 Association of Colleges of Nursing. NLNAC accredits all levels of nursing academic programs from  
3425 diploma and associate degrees (as well as licensed practical nurse programs) to advanced practice and  
3426 D.P.N.s; the CCNE accredits only baccalaureate and graduate nursing academic programs. The two  
3427 organizations have very different assessment criteria, and consequently some schools carry accreditation  
3428 from both. NLNAC and CCNE now require objective evidence of genetic content or instruction in  
3429 nursing curriculums.<sup>378</sup> For programs renewing during the next accreditation cycle in 2010, CCNE will  
3430 begin to assess if schools are moving toward incorporation of genetic content.

3431  
3432 CE for R.N.s is extremely heterogeneous and in some states is monitored per the requirements of state  
3433 boards of nursing. Presently, 19 states have no CE requirements for renewal of active R.N. licenses.<sup>379</sup>  
3434 The remaining states have widely varying requirements, extending from as little as 5 hours of CE per year  
3435 to as many as 15 hours per year. No state board of nursing has a genetics and genomics requirement for  
3436 maintenance of an active R.N. professional license.

### 3437 3438 **Education and Licensure of Physician Assistants and Accreditation of PA Programs**

3439  
3440 Academic paths to becoming a PA include baccalaureate study prior to acceptance into a Surgical or  
3441 Physician Assistant graduate program. There are presently 136 accredited PA programs in the United  
3442 States; they average 26 months in duration and comprise one year of didactic and one year of clinical  
3443 training. Recent survey results of 100 accredited PA programs indicated two-thirds of them devote 7 to 20  
3444 hours to genetics content in their curricula, and many plan to incorporate further genetic content in the  
3445 near future.<sup>380</sup> Recognizing the importance that genetics is garnering for future clinical practice, recent  
3446 foundational curriculum guidelines were issued, and cover content ranging from classic medical genetics  
3447 to Human Genome Project implications and polymorphisms as genetic health markers.<sup>381</sup> Following  
3448 completion of an accredited program, the National Commission on Certification of Physician Assistants  
3449 (NCCPA) certifies PA candidates. For individuals to receive the Physician Assistant-Credentialed (PA-C)  
3450 credential, they must meet professional knowledge and skill standards as measured by successful  
3451 performance on the Physician Assistant National Certifying Exam (PANCE).<sup>382</sup> Although covering  
3452 single-gene disorders and other hereditary conditions, the PANCE does not include a genetics section or  
3453 genomics content.

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<sup>377</sup> 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.

<sup>378</sup> National League for Nursing Accrediting Commission, Inc. *NLNAC 2008 Standards and Criteria*. See [www.nlnac.org/manuals/SC2008.htm](http://www.nlnac.org/manuals/SC2008.htm). Accessed on November 24, 2009.

<sup>379</sup> NurseWeek. *Nursing Continuing Education Requirements by State*. See [www.nurse.com/ce/Requirements.html](http://www.nurse.com/ce/Requirements.html); <http://www.nursingcenter.com/pdf.asp?AID=636579>. Accessed on November 16, 2009.

<sup>380</sup> National Institutes of Health. *Physician Assistant Competencies for Genomic Medicine: Where We Are Today and How to Prepare for the Future*. Meeting Summary, March 29-30, 2007. See [www.genome.gov/Pages/About/OD/Reports/Publications/PAMeetingSummaryMarch2007.pdf](http://www.genome.gov/Pages/About/OD/Reports/Publications/PAMeetingSummaryMarch2007.pdf). Accessed on November 16, 2009.

<sup>381</sup> Goldgar, C.M., and Rackover, M. (2008). Recommendations for a Physician Assistant medical genetics curriculum. *The Journal of Physician Assistant Education*. 19(2):30-36.

<sup>382</sup> National Commission on Certification of Physician Assistants. See [www.nccpa.net/](http://www.nccpa.net/). Accessed on November 24, 2009.

3455 Accreditation of physician assistant programs is granted via the Accreditation Review Commission on  
3456 Education for the Physician Assistant. The current standards were last reviewed in 2006 and include  
3457 requirements for instruction of molecular concepts as related to health and disease, including genetics.<sup>383</sup>  
3458 However, similar to other disciplines, these standards are largely restricted to biologic scientific principles  
3459 and limited clinical application contexts, such as single-gene disorders.

3460  
3461 The American Academy of Physician Assistants is the primary professional organization representing the  
3462 clinical, educational, and research interests of the PA community and offers discipline-specific CE. To  
3463 maintain active certification status, certified PAs must complete 100 CE hours every two years. At least  
3464 half of all CE units (50 hours) must come from attending seminars or conference sessions from pre-  
3465 approved sponsor sources. The remaining 50 hours of CE can come from elective sources (e.g., journal  
3466 reviews, practice-related activities, self-learning modules, independent studies), for which genetics and  
3467 genomics content is covered only as a function of individual interest. Re-certification is required every six  
3468 years by NCCPA via the Physician Assistant National Recertifying Exam, with genetic examination  
3469 content similar to the PANACE.

3470

### 3471 **Education and Certification of Genetic Counselors and Accreditation of Genetic Counseling** 3472 **Programs**

3473

3474 Academic paths to becoming a genetic counselor include baccalaureate study prior to acceptance into one  
3475 of the country's 30 accredited graduate genetic counselor programs. Following completion of an  
3476 accredited program, candidates are eligible for certification from the American Board of Genetic  
3477 Counseling (ABGC) to obtain the Certified Genetic Counselor credential, which remains active for a  
3478 period of 10 years. In 2010, this 10-year period will be halved, and certification will be granted in 5-year  
3479 increments. As of January 2008, six states—California, Illinois, Massachusetts, Oklahoma, Tennessee, and  
3480 Utah—require a professional license in addition to certification.<sup>384</sup>

3481

3482 The ABGC accredits genetic counselor programs. Revised in March of 2009, the expanded genetic and  
3483 genomic content requirements are built into accreditation standards.<sup>385</sup> Included in the accreditation  
3484 requirements are the expected molecular concepts such as inheritance patterns, population genetics,  
3485 human genetic variation and related susceptibilities, family history analysis, and human development and  
3486 reproduction. Also included are laboratory and research experiences, as related to capacity for competent  
3487 clinical practice.

3488

3489 Current pathways for recertification are successful re-examination or through accumulation of CE credits.  
3490 CE for genetic counselors are issued and monitored by the ABGC, which has specific Professional  
3491 Activity Credit requirements that may be fulfilled through a wide range of professional development  
3492 paths.<sup>386</sup> The primary professional society representing genetic counselors, the National Society of  
3493 Genetic Counselors, provides CE units per pre-approved criteria and sponsor initiated activities.<sup>387</sup> The

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<sup>383</sup> Accreditation Review Commission on Education for the Physician Assistant, Inc. *Accreditation Standards for Physician Assistant Education*. 3rd edition (October, 2007). See [http://www.arc-pa.org/acc\\_standards/](http://www.arc-pa.org/acc_standards/). Accessed on November 24, 2009.

<sup>384</sup> National Conference of State Legislators. *Genetic Counselor Licensing*. See [www.ncsl.org/default.aspx?tabid=14282](http://www.ncsl.org/default.aspx?tabid=14282). Accessed on November 24, 2009.

<sup>385</sup> American Board of Genetic Counseling, Inc., Required Criteria for Graduate Programs in Genetic Counseling Seeking Accreditation by The American Board of Genetic Counseling. See [www.abgc.net/CMFiles/REQUIRED\\_CRITERIA\\_revised\\_Mar\\_25\\_200951KIH-432009-1159.pdf](http://www.abgc.net/CMFiles/REQUIRED_CRITERIA_revised_Mar_25_200951KIH-432009-1159.pdf). Accessed on November 25, 2009.

<sup>386</sup> American Board of Genetic Counseling. Professional Activity Credits. See [www.abgc.net/english/view.asp?x=1659](http://www.abgc.net/english/view.asp?x=1659). Accessed on November 25, 2009.

<sup>387</sup> National Society of Genetic Counselors. Continuing Education Information. See [www.nsgc.org/CEU/ApprovedPrograms1.cfm](http://www.nsgc.org/CEU/ApprovedPrograms1.cfm). Accessed on November 25, 2009.

3494 ABGC CE program currently is being restructured to meet the 5-year recertification cycle going into  
 3495 effect in 2010.

3496  
 3497 **Education of Pharmacists in Genetics and Genomics**  
 3498

3499 In 2002, Brock et al. sent surveys to the curriculum committee chairpersons at the 82 accredited pharmacy  
 3500 schools in the United States, asking how many lecture hours were devoted to genomic topics.<sup>388</sup> Of the 50  
 3501 responses, 64 percent reported 0 to 1 hour devoted to ethical considerations, and 30 percent reported 0 to  
 3502 1 hour for practical applications. By 2005, 78 percent of pharmacy schools surveyed provided some  
 3503 instruction in pharmacogenomics. However, the average pharmacy school that included instruction  
 3504 related to pharmacogenomics addressed only half of the AACP Academic Affairs Committee 2002  
 3505 recommendations regarding the need for pharmacogenomics and pharmacogenetics knowledge.<sup>389</sup>  
 3506

3507 The AACP House of Delegates passed policy resolutions in 2008 stating that pharmacy curricula must  
 3508 adequately address contemporary issues associated with biotechnology advances in personalized  
 3509 medicine, including competencies in genetics and genomics and preparing faculty to contribute to  
 3510 education and research related to genetics and genomics.<sup>390</sup>  
 3511

3512 In 2009, Murphy et al. conducted a follow-up survey to Brock's 2002 survey. Results indicate that 92  
 3513 percent of colleges of pharmacy reported teaching pharmacogenomics within their programs, up from 78  
 3514 percent of programs surveyed in 2005.<sup>391</sup>  
 3515

3516 To meet the pharmacogenomic educational needs of U.S. Colleges of Pharmacy, the Pharmacogenomics  
 3517 Education Program: Bridging the Gap between Science and Practice (*PharmGenEd*<sup>TM</sup>),<sup>392</sup> was developed.  
 3518 Funded by CDC, it is an evidence-based pharmacogenomics education program designed for pharmacists  
 3519 and physicians, pharmacy and medical students, and other health care professionals. The program team at  
 3520 University of California, San Diego Skaggs School of Pharmacy and Pharmaceutical Sciences is  
 3521 collaborating with national pharmacy, medical, and health care organizations to deliver *PharmGenEd*<sup>TM</sup>  
 3522 materials to more than 100,000 pharmacists, physicians, and health care professionals. Program directors  
 3523 have conducted ongoing surveys and collected evaluation data from resulting *PharmGenEd*<sup>TM</sup>  
 3524 educational programs. Highlights of pre- and post-program survey results were provided at the 2009  
 3525 American Pharmacists Association's annual meeting, showing, for example, increased knowledge of  
 3526 adverse drug reactions related to HLA-B\*5701 variation and increased overall ability to address  
 3527 pharmacogenomic testing with patients. As a result of the program, pharmacists indicated they would be  
 3528 more likely to:  
 3529

- 3530 • Explain the rationale to patients for pharmacogenomic testing (69 percent)
- 3531 • Discuss risks and benefits of pharmacogenomic testing with patients (67 percent)
- 3532 • Find credible and current literature related to pharmacogenomic testing (63 percent)
- 3533 • Recommend or refer patients for pharmacogenomic testing, if applicable (61 percent)
- 3534 • Recommend the *PharmGenEd*<sup>TM</sup> CE/CME program to colleagues (84 percent)

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<sup>388</sup> Brock, T.P., Faulkner, C.M., Williams, D.M., and Smith, S.R. (2002). Continuing education programs in pharmacogenomics for pharmacists. *American Journal of Health Systems Pharmacology*. 59:722-725.

<sup>389</sup> Latif, D.A., and McKay, A. (2005). Pharmacogenetics and pharmacogenomics instruction in colleges and schools of pharmacy in the United States. *American Journal of Pharmaceutical Education*. 69(2):Article 23.

<sup>390</sup> American Association of Colleges of Pharmacy. Minutes of the House of Delegates Sessions, July 20-23, 2008. See [www.aacp.org/governance/HOD/Documents/HOD\\_Minutes08.pdf](http://www.aacp.org/governance/HOD/Documents/HOD_Minutes08.pdf). Accessed on November 25, 2009.

<sup>391</sup> Murphy, J.E., Green, J.S., Adams, L.A., Squire, R.B., Kuo, G.M., and McKay, A. (2010). Pharmacogenomics in the curricula of colleges and schools of pharmacy in the United States. *American Journal of Pharmaceutical Education*. 74(1):1-10.

<sup>392</sup> University of California, San Diego Scaggs School of Pharmacy and Pharmaceutical Sciences. Pharmacogenomics Educational Program PharmGenEd. See <http://pharmacogenomics.ucsd.edu>. Accessed on September 8, 2009.

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- 3535 • Agree that the pharmacy profession should be more active in educating patients and other health  
3536 care professionals about pharmacogenomic testing (88 percent)
- 3537 • Understand that issues related to ethical, social, legal, and economic aspects of genetics are  
3538 important in translating pharmacogenomics evidence into practice (96 percent).

3539 **Appendix B: SACGHS Survey of Health Care Professional Organizations**

3540

3541 **1. Health Care Professional Organizations Surveyed**

3542

3543 A total of 60 organizations were invited to participate in the survey. They were broken into three groups  
3544 for analysis: genetic-specific organizations, nongenetic organizations, and Federal advisory committees.  
3545

3546

3546 **Genetic-Specific Organizations (9)**

3547 American Board of Genetic Counseling (ABGC)

3548 American Board of Medical Genetics (ABMG)

3549 American College of Medical Genetics (ACMG)

3550 American Society of Human Genetics (ASHG)

3551 Association of Professors of Human and Medical Genetics (APHMG)

3552 Genetic Nursing Credentialing Commission (GNCC)

3553 International Society of Nurses in Genetics (ISONG)

3554 National Coalition for Health Professional Education in Genetics (NCHPEG)

3555 National Society of Genetic Counselors (NSGC)

3556

3557 **Nongenetic Organizations (48)**

3558

3559 Accreditation Council for Graduate Medical Education (ACGME)

3560 Accreditation Review Commission on Education for the Physician Assistant (ARC-PA)

3561 Alliance of Academic Internal Medicine (AAIM)

3562 American Academy of Family Physicians (AAFP)

3563 American Academy of Nursing (AAN)

3564 American Academy of Pediatrics (AAP)

3565 American Academy of Physician Assistants (AAPA)

3566 American Association of Colleges of Nursing (AACN)

3567 American Association of Colleges of Osteopathic Medicine (AACOM)

3568 American Association of Colleges of Pharmacy (AACP)

3569 American College of Clinical Pharmacology

3570 American College of Obstetricians and Gynecologists (ACOG)

3571 American College of Physicians (ACP)

3572 American College of Preventive Medicine (ACPM)

3573 American Dental Education Association (ADEA)

3574 American Medical Association (AMA)

3575 American Nurses Association (ANA)

3576 American Osteopathic Association (AOA)

3577 American Residency Coordinators in Obstetrics and Gynecology (ARCOG)

3578 American Society for Clinical Oncology (ASCO)

3579 Association of American Indian Physicians (AAIP)

3580 Association of American Medical Colleges (AAMC)

3581 Association of Black Women Physicians (ABWP)

3582 Association of Family Medicine Program Directors (AFMPD)

3583 Association of Pediatric Program Directors (APPD)

3584 Association of Professors of Gynecology and Obstetrics (APGO)

3585 Association of Schools of Allied Health Professions (ASAHP)

3586 Association of Schools of Public Health (ASPH)

3587 Association of Women's Health, Obstetric and Neonatal Nurses (AWHONN)

3588 Council on Medical Student Education in Pediatrics (COMSEP)

- 3589 Council on Resident Education in Obstetrics and Gynecology (CREOG)
- 3590 Liaison Committee on Medical Education (LCME)
- 3591 National Association of Pediatric Nurse Practitioners (NAPNP)
- 3592 National Black Nurses Association (NBNA)
- 3593 National Board of Medical Examiners (NBME)
- 3594 National Coalition of Ethnic Minority Nurses Association (NCEMNA)
- 3595 National Hispanic Medical Association (NHMA)
- 3596 National League of Nursing (NLN)
- 3597 National Medical Association (NMA)
- 3598 National Organization of Nurse Practitioner Faculties (NONPF)
- 3599 Network of Ethnic Physician Organizations (NEPO)
- 3600 Oncology Nursing Certification Corporation (ONCC)
- 3601 Oncology Nursing Society (ONS)
- 3602 Physician Assistant Education Association (PAEA)
- 3603 Robert Graham Center
- 3604 Sigma Theta Tau International (STTI)
- 3605 Society of General Internal Medicine (SGIM)
- 3606 Society of Teachers of Family Medicine (STFM)

3607

3608 **Federal Advisory Committees (3)**

3609

- 3610 Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)
- 3611 Advisory Committee on Training in Primary Care Medicine and Dentistry (ACTPCMD)
- 3612 Council on Graduate Medical Education (COGME)

3613

3614 **2. Health Care Professional Organizations' Survey Methodology**

3615

3616 The main body of the survey instrument consisted of 15 open- and close-ended questions developed by  
3617 SACGHS (see Appendix B-3). Close-ended questions were in both multiple-choice and Likert scale  
3618 formats. Organizations were also asked to complete a narrative description of ongoing genomics-related  
3619 projects. The draft instrument was piloted with board members of the NCHPEG, refined, and  
3620 subsequently reviewed by a survey methodologist to maximize survey validity. The instrument explored  
3621 several major themes including the organizations' perceived role in, and priority ascribed to genomics  
3622 education; barriers to enhancing their role in genomics education; and a description of their past, present,  
3623 and planned efforts around genomics education.

3624

3625 The survey was sent via e-mail to key staff in the 60 targeted organizations in January 2009. All  
3626 nonresponders were contacted by e-mail and/or phone by SACGHS to maximize response rates. Thirty-  
3627 six responses were received (60 percent).

3628

3629 Survey data were compiled by SACGHS staff and entered into Excel spreadsheets. The organizations  
3630 were divided into three major divisions: genetic-specific organizations; nongenetic organizations; and  
3631 Federal advisory committees, and analyses conducted according to those divisions. Responses were  
3632 extracted from returned surveys and manually entered into a FileMaker Pro 10 database. Once complete,  
3633 the derived data were exported as an Excel spreadsheet for further analyses.

3634

3635 **3. Health Care Professional Organizations' Survey Instrument**

3636

- 3637 1) Name of organization:
- 3638
- 3639 2) What is your title and primary role in the organization?

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- 3640  
 3641 3) What is the size of your organization’s constituency or membership?  
 3642  
 3643 4) Please identify which of the following most closely describes your organization’s mission. **Circle or**  
 3644 **underline one answer.**  
 3645  
 3646 A) Advocacy for and support of practicing health professionals  
 3647 B) Education and training of health professionals  
 3648 C) Certification of health professionals  
 3649 D) Accreditation or certification of institutions  
 3650 C) Other (please describe):  
 3651

- 3652 5) Is genetics education and training part of the role or responsibility of your organization?  
 3653 If no, please proceed to question 6. If yes,  
 3654  
 3655 A) Please briefly describe this role or responsibility.  
 3656  
 3657 B) Is your organization currently able to fulfill this role or responsibility?  
 3658  
 3659 C) Are there ways in which your organization could meet this role or responsibility more  
 3660 effectively? If yes, please describe how.  
 3661

3662 **For questions 6-10, please circle or underline the most appropriate number; circle or underline NA**  
 3663 **if not applicable to your organization.**  
 3664

- 3665 a. What importance does your organization place on the development and promotion of educational  
 3666 activities (including continuing education) in the health area generally?  
 3667

3668 Not at all important    **1**    **2**    **3**    **4**    **5**    **NA**    Very important  
 3669

- 3670  
 3671 b. What importance does your organization place on the development and promotion of educational  
 3672 activities (including continuing education) specifically related to genetics and genomics?  
 3673

3674 Not at all important    **1**    **2**    **3**    **4**    **5**    **NA**    Very important  
 3675  
 3676

- 3677 c. Where does genetics and genomics education fall relative to the overall priorities facing your  
 3678 organization?  
 3679

3680 Low priority    **1**    **2**    **3**    **4**    **5**    **NA**    High priority  
 3681  
 3682

- 3683 d. To what extent is your organization’s membership satisfied with the organization’s current emphasis  
 3684 on genetics and genomics education?  
 3685

3686 Not at all satisfied    **1**    **2**    **3**    **4**    **5**    **NA**    Extremely satisfied  
 3687  
 3688

- 3689 e. How proficient and comfortable would you say your organization’s leadership is with genetics and  
 3690 genomics education?

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3691  
 3692 Low expertise/comfort 1 2 3 4 5 NA High expertise/comfort  
 3693

3694  
 3695 f. Does your organization have an established committee, workgroup, or dedicated staff that deals  
 3696 specifically with topics in genetics or genomics relevant to your organization’s mission? **Please**  
 3697 **circle or underline one answer.**  
 3698

- 3699 A) Yes  
 3700 B) No  
 3701 C) Not sure (please explain):  
 3702  
 3703

3704 g. Which of the following do you consider to be barriers to your organization’s ability to provide  
 3705 genetics and genomics education? **Please circle or underline all that apply.**  
 3706

- 3707 1. Genetics and genomics education is not applicable to the organization’s mission  
 3708  
 3709 2. The organization’s leadership lacks knowledge of genetics and genomics  
 3710  
 3711 3. The organization has competing priorities  
 3712  
 3713 4. There is a lack of accessible educational resources for genetics and genomics  
 3714  
 3715 5. Genetics and genomics is not emphasized in certifying examinations/credentialing standards  
 3716  
 3717 6. The organization believes there is a lack of evidence supporting clinical effectiveness of care based  
 3718 on genetic or genomic information  
 3719  
 3720 7. Other (please list):  
 3721  
 3722 8. From our organization’s perspective, there are no barriers  
 3723

3724 13) In the space below, please rank the items selected in question 12 from the most important to least  
 3725 important barrier (e.g., E, D, C).  
 3726  
 3727

3728 14) Please fill out the table below to describe any **completed** initiatives/programs your organization has  
 3729 implemented in the last five years for educating its membership on genetics and genomics topics.  
 3730 Please expand the table as needed for each section or to include additional programs.  
 3731

<b>Program #1</b>	
Brief description	
Outcome measures used to evaluate program’s success	
External collaborators (if applicable)	

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

3732  
3733  
3734  
3735  
3736  
3737  
3738

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.

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

collaborators (if applicable)	
URLs for web-based resources related to the program	
Publications citations (if any) related to the program	

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3780

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?

3781  
3782 7) Looking ahead 5 to 10 years, what needs do you anticipate in genetics and genomics education?  
3783

3784  
3785 **Category 2: Advocacy for and support of practicing health professionals**  
3786

3787 1) What is the role of your organization in education, training, and assessment of the professional  
3788 workforce?

3789  
3790 2) Do you offer continuing education programs/activities?  
3791 If yes, are any specific to genetics or genomics?

3792  
3793 3) Has your organization published any position statements or practice competencies regarding  
3794 genetics? (Please circle or underline your answer)

3795 A) Yes

3796 B) No

3797 C) In progress

3798 D) Not sure (please explain):  
3799

3800 4) Do you think your members need more information about genetics and genomics?  
3801 If yes, on what topics?

3802  
3803 5) What would help to promote a greater knowledge of genetics and genomics?  
3804  
3805

3806 **Category 3: Certification of Health Professionals**  
3807

3808 1) Do current credentialing exams include questions on genetics and genomics?  
3809 If yes, approximately what percentage of the questions is on genetics and genomics?  
3810

3811 2) How frequently are the questions updated?  
3812

3813 3) Would your organization like help in developing questions on genetics and genomics?  
3814

3815 **Category 4: Accreditation or Certification of Institutions**  
3816

3817 1) Are there minimum curriculum requirements in genetics or genomics?  
3818 If yes, please provide a brief description.  
3819

3820 2) If there are minimum curriculum requirements in genetics or genomics, how often are they  
3821 updated?  
3822

3823 3) From the perspective of your organization, please characterize the need for the integration of  
3824 genetics and genomics into the curriculum and training of health professionals.  
3825

3826 **4. Health Care Professional Organization's Survey Names and Constituency or Membership**  
3827 **of Responding Organizations**  
3828

3829 The table below lists the organizations that responded to the survey with their reported constituency or  
3830 membership numbers noted. Because organizations were asked to indicate the size of their constituency  
3831 or membership, those that represent a profession as a whole have some overlap in numbers with smaller

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3832 subgroups (e.g., the American Nursing Association and the Oncology Nursing Society). Thus, the  
 3833 membership or constituency numbers cannot be added together, and the total number of unique health  
 3834 professionals represented by these organizations is not known.  
 3835

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

3836

3837 **5. Health Care Professional Organization Survey Tables**  
 3838  
 3839

**Table 1. Organizations' Role or Responsibility in Genetics Education and Training**

<b>Organization Abbreviation</b>	<b>Education and Training Role Description</b>
AACN	AACN has partnered with NHGRI and NCI on several initiatives: 1. Assisting with creating and endorsing the Essential Competencies and Curricula Guidelines for Genetics & Genomics (2005). 2. Assisting with creating a tool kit for faculty development. 3. Assisting with creating a tool kit repository.
AAFP	<ul style="list-style-type: none"> <li>• Educates family medicine residents and, through CME, educates its physician members.</li> <li>• 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.</li> <li>• 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.</li> <li>• AAFP is a member of NCHPEG.</li> </ul>
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 <a href="http://abrcc.iamonline.com/CMFiles/ABGDC_CO_Final5_1K">http://abrcc.iamonline.com/CMFiles/ABGDC_CO_Final5_1K</a>

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	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.
ABMG	ABMG accredits training programs in clinical cytogenetics, biochemical genetics, and molecular genetics. Educational standards are designed by the ABMG for implementation by the training programs.
ACCP	Pharmacogenetics is a component of clinical pharmacology, therefore it may be included in the symposia that we sponsor. We belong to NCHPEG, and provide information and web links regarding their genetics teaching resources to our membership via e-mail notices.
ACHDNC	The grant program established under Section 1109 of our authorizing legislation specifies these activities. <ol style="list-style-type: none"> <li>1. Assist in providing health care professionals and laboratory personnel education and training in newborn screening.</li> <li>2. Provide educational programs to parents, families and patient advocacy groups.</li> </ol>
ACMG	As a membership organization representing medical geneticists, it is inherent in our responsibilities. Our members direct training programs for medical geneticists and are directly involved in teaching and training of others in academic medical centers.
ACPM	ACPM is currently developing a CME program for its membership and broader community of primary care physicians.
ACP	We incorporate genetics education into our live courses and publish materials that include genetics education.
ACOG	Develop clinical guidelines and patient and professional resources.
AMA	The AMA mission is to support physicians by working on important health issues. The AMA Program in Genetics and Molecular Medicine aims to identify genetics issues relevant to physicians and provide educational support to physicians as they integrate genetic technologies into clinical practice.
APHMG	We represent professors of genetics in all areas of genetics, and are involved in resident, fellow, medical student, and graduate student education.
ASHG	Support of trainees in presenting research, travel to meetings. Our director of education and Committee help with education in K-12 to open the pipeline early.
AWHONN	Genetics information is integrated in other specialty specific content for our educational resources.
COMSEP	Set national curricula.
ISONG	ISONG is a global nursing specialty organization dedicated to fostering the scientific and professional growth of nurses in human genetics and genomics worldwide. It provides a forum for education and support for nurses providing genetic health care.
NAPNP	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.

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NCHPEG	NCHPEG’s mission is to promote health professional education and access to information about advances in human genetics to improve the health care of the nation. NCHPEG fulfills this mission by: <ul style="list-style-type: none"> <li>• integrating genetics content into the knowledge base of health professionals and students of the health professions,</li> <li>• 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.</li> </ul>
ONS	One of the topics in the ONS Strategic Plan for 2009-2012, is biology and cancer and emerging trends in diagnosis and treatment. Genetics is a big force in these two areas and needs.
STTI	Planning for free online repository available to health professionals for content, tool kits, etc., related to genetics through the Virginia Henderson International Library.

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

	All Organizations		By Organization Type					
			Genetics Specific Organizations		Federal Advisory Committees		Other Organizations	
	%	#	%	#	%	#	%	#
<b>Yes</b>	47%	17	78%	7	50%	1	36%	9
<b>No</b>	47%	17	22%	2	50%	1	56%	14
<b>Not Sure</b>	6%	2	0%		0%		8%	2
<b>Total Answers</b>		<b>36</b>		<b>9</b>		<b>2</b>		<b>25</b>

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

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

Genetics and genomics is not applicable to the organization's mission	11	11	12	0
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

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**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.

not required

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**

penetrance, variable expression, and spontaneous mutations
<input type="checkbox"/> Basic embryology and teratology
<input type="checkbox"/> Introductory history taking and physical examination skills
Competencies
Knowledge
1. Describe the genetic basis and clinical manifestations of the following syndromes, malformations, and associations:
<input type="checkbox"/> Common chromosomal abnormalities, (e.g. Trisomy 21 (CP), Turner syndrome (CP), Klinefelter syndrome (M))
<input type="checkbox"/> Syndromes due to teratogens (e.g. fetal alcohol syndrome) (CP)
<input type="checkbox"/> Other common genetic disorders (e.g. cystic fibrosis, sickle cell disease, hemophilia) (CP)
<input type="checkbox"/> Single malformations with multifactorial etiology (e.g. spina bifida, congenital heart disease, cleft lip and palate) (M)
2. List common medical and metabolic disorders (e.g. hearing loss, hypothyroidism, PKU, hemoglobinopathies) detected through newborn screening programs (CP)
3. Discuss the effects of maternal health and potentially teratogenic agents on the fetus and child, including maternal diabetes and age (CP), alcohol use (CP) illicit drug use (CP), and prescribed medications such as phenytoin, valproate, and retinoic acid (M)
4. List common prenatal diagnostic assessments (e.g. maternal serum screening, amniocentesis, and ultrasonography) and understand their use (M)
5. Describe the use of chromosome studies in the diagnosis of genetic disorders (M)
6. Discuss the role of genetics in common multifactorial conditions (e.g. inflammatory bowel disease, pyloric stenosis, congenital heart disease, cleft lip, diabetes and cancer) (M)
Skills
1. Use a family history to construct a pedigree (e.g., for the evaluation of a possible genetic disorder) (CP)
Not available info across all US colleges of Pharmacy, new survey in field
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

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**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

## 3869 **Appendix C: SACGHS Survey of Public Health Providers**

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### 3871 **1. Survey Methodology**

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3873 Using the Delphi technique,<sup>393,394</sup> SACGHS developed 12 competencies in genetics of relevance to the  
 3874 public health workforce. Many of the competencies were derived from existing sources, including the  
 3875 National Coalition of Health Professional Education in Genetics (NCHPEG), the Centers for Disease  
 3876 Control and Prevention (CDC), the Association of State Territorial Health Officers, Training Finder Real-  
 3877 time Affiliate Integrated Network, and the University of Washington. These competencies were translated  
 3878 into an online survey instrument with the intent of assessing public health providers' opinions on the  
 3879 importance of each competency, their confidence in demonstrating each competency, and the frequency  
 3880 with which they apply each competency. The conceptualization and formatting of the competencies into  
 3881 an online survey was based on work by Kirk, et al., who sought to implement a novel approach to  
 3882 ascertain practitioners' needs in genetics education.<sup>395</sup> The questionnaire was reviewed by SACGHS  
 3883 members and staff, and additional items were added to assess the importance of genetics and genomics to  
 3884 the respondent's leadership and their own role in public health. The final online survey was a mixed-  
 3885 format 38-item assessment tool that included demographic questions (see Appendix C-2).

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3887 To achieve a broad representation of public health providers who work in a variety of settings,  
 3888 recruitment utilized multiple strategies that included (1) using a list of state public health and genetic  
 3889 professionals, (2) partnering with the American Public Health Association Genomics Forum, and, (3)  
 3890 partnering with the National Society of Genetic Counselors. An e-mail invitation to participate in the  
 3891 survey was then distributed to approximately 500 public health professionals. Some respondents  
 3892 forwarded the survey to others they felt were appropriate. Online survey participants reflected a diversity  
 3893 of public health providers with varying degrees of genetics responsibilities. For some it is their primary  
 3894 job, for others genetics is just one aspect of their position. A total of 140 responses were received and  
 3895 analyzed. It is not possible to calculate response rate because the total number of individuals who  
 3896 eventually received the survey is unknown.

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3898 Survey data for the public health providers in genetic and genomic competencies were initially entered  
 3899 into Microsoft Excel and subsequently converted into the Statistical Package for the Social Sciences  
 3900 (SPSS). See Appendix C-3 for a discussion of reliability analysis.

3901

3902 The survey included two open-ended qualitative questions. All responses were downloaded and entered  
 3903 into qualitative analytical software, Atlas TI. The responses were analyzed for commonalities among the  
 3904 responses. The results provided below highlight the most common themes that emerged.

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### 3906 **Sample Size and Missing Data**

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3908 The total sample size used in the analysis was 140 participants. There were instances in which data were  
 3909 missing for specific questions within each competency where the response rate was below 140. In

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<sup>393</sup> 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.

<sup>394</sup> Bernard, H. R., (2000). *Social Research Methods: Qualitative and Quantitative Approaches.*, Thousand Oaks, CA: Sage Publications. p. 247.

<sup>395</sup> Kirk, M., Tolkin, E., Birmingham, K. (2007). Working with Publishers: A novel approach to ascertaining practitioners' needs in genetics education. *Journal of Nursing Research.* 12;597-615.

3910 situations such as this, missing data values were recoded to equal “no answer” on the Likert scale. As a  
3911 result of the recode, the means were computed based on subtracting the “no answer” responses from the  
3912 computation and using the 140 participants as the common denominator. The response rate to each  
3913 question for the 12 competencies appears to be relatively high indicating that minimal data are missing.

3914 **2. Public Health Providers’ Survey Instrument**  
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Click the button below to start a new survey:

Start Survey -->

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**Survey of Public Health Providers in Genetic/Genomic Competencies**

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 the public health workforce. 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.

To continue and begin the survey, click the "Next" button below.

Next -->

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**Privacy Statement**

Your participation in this survey is completely voluntary. Please be assured that your participation in the survey will be kept confidential and your responses will never be linked or associated with you. You may skip any questions that you prefer not to answer. You are also free to stop participating at any point during the survey and have your responses deleted by clicking the "Opt out of survey" box at the bottom of each survey page.

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**Part I: Your Practices of 12 Competencies** % Completed

Reflecting on your current role, please check one answer from the questions below that best describes your practice for each competency statement.

**A public health professional is able to:**

---

**1. Maintain up-to-date knowledge on the development of genomic science and technologies within his or her professional field and program to apply genomics as a tool for achieving public health goals.**

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

---

**2. Demonstrate basic knowledge of the role that genetics/genomics plays in the development of disease and in screening and interventions for programs of disease prevention and health promotion.**

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

---

**3. Describe the importance of family history in assessing predisposition to disease.**

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

---

**4. Identify opportunities and integrate genetic/genomic issues into public health practice, policies or programs effectively.**

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Never	1-2 Per Year	Monthly	Weekly
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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**Part I (cont'd)** % Completed

Reflecting on your current role, please check one answer from the questions below that best describes your practice for each competency statement.

**A public health professional is able to:**

---

**5. Maintain up-to-date knowledge of genetics/genomics-related policies, legislation, statutes, and regulations.**

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Not at All	Not Very	Somewhat	Very	
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Never	1-2 Per Year	Monthly	Weekly	
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Not at All	Not Very	Somewhat	Very	
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Never	1-2 Per Year	Monthly	Weekly	
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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**7. Collaborate with existing and emerging health agencies and organizations, academic, research, private and commercial enterprises, and community partnerships to apply genetics/genomics knowledge and tools to address public health problems.**

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Not at All	Not Very	Somewhat	Very	
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Never	1-2 Per Year	Monthly	Weekly	
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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**8. Identify the resources available to assist clients seeking genetic/genomic information or services, including the types of genetics professionals available.**

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Not at All	Not Very	Somewhat	Very	
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Never	1-2 Per Year	Monthly	Weekly	
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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**Part I (cont'd)** % Completed

Reflecting on your current role, please check one answer from the questions below that best describes your practice for each competency statement.

**A public health professional is able to:**

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**9. Conduct outcomes evaluation of available genetic/genomic programs and services to determine their effectiveness.**

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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**11. Use information technology (IT) to obtain credible, current information about genetics; to utilize IT skills to share data and participate in research, program planning, evaluation, and policy development for health promotion and disease prevention.**

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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

	Not at All	Not Very	Somewhat	Very
How important is the competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How confident are you in demonstrating this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
How frequently do you apply this competency?	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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**Part II: Importance of genetics/genomics to your institution's leadership**

% Completed

1. Does your senior administration think that genetics/genomics is important to your job responsibilities?

- Not at all important
- Of little importance
- Somewhat important
- Important
- Very Important

2. Does your senior administration think that genetics/genomics is important to their job responsibilities?

- Not at all important
- Of little importance
- Somewhat important
- Important
- Very Important

3. How adequate are your resources for implementing genetic/genomic competencies into your work/role?

- Not at all adequate
- Somewhat adequate
- Adequate
- Very adequate

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**Part III: Your role in Public Health** % Completed

1. At what level of public health do you work?

- Federal
- State
- Local
- Academic
- Private, non-profit organization
- Community-based Organization
- International
- Other (specify):

2. What is your job title?

3. What percent of your time do you spend doing the following?

Role	< 25%	25-50%	50-75%	> 75%
Administrative	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Program Planning	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Direct Consumer Care	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Policy/Legislative	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Research	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Assessment/Evaluation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Education/Training	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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**Part III (cont'd)** % Completed

4. Please describe any efforts that you or your organization have undertaken to ensure that genetic services or information are available for vulnerable or underserved populations. Are there particular strategies you would recommend? (Limit 200 words).

Opt out of survey
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**Secretary's Advisory Committee on Genetics, Health, and Society**  
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**Part IV: Additional Information** % Completed

1. Please provide any additional information that you would like to share with SACGHS on the topic of genetics and genetics education for Public Health Providers. *(Limit 200 words).*

If you experience any technical difficulties, please contact the survey administrator at User-Centered Design at [providersurvey@user-centereddesign.com](mailto:providersurvey@user-centereddesign.com)

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**3. Reliability Results and Discussion**

A total of 140 respondents were entered into the dataset. For the reliability analysis, the valid sample size was 132 participants due to missing data that were automatically excluded from the analysis. The number of total items in the overall reliability analysis was 36. These items consisted of three of the same questions for each of the 12 competencies. Additionally, three separate reliability analyses were conducted for each of the three questions that were asked for all 12 competencies. In each of these three analyses the total number of items in the analysis was 12.

**Reliability for Overall Instrument (12 Competencies each with 3 Questions Totaling 36 Items)**

The overall Cronbach's Alpha for the instrument is 0.980. 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  $r = 0.30$ .

3960 **4. Public Health Providers' Survey Tables and Summary Data**

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3962 **Table 1. Perception of the Importance of the Competencies**

Competency		Question	0	1	2	3	4	Response Rate	Mean
1	Maintain up-to-date knowledge on the development of genomic science and technologies within his or her professional field and program to apply genomics as a tool for achieving public health goals.	How important is the competency?	9	0	4	35	92	94 percent	3.7
		How confident are you in demonstrating the competency?	9	3	21	61	46	94 percent	3.1
		How frequently do you apply this competency?	10	6	25	32	67	93 percent	3.2
2	Demonstrate basic knowledge of the role that genetics and genomics plays in the development of disease and in screening and interventions for programs of disease prevention and health promotion.	How important is the competency?	10	0	2	17	11 1	93 percent	3.8
		How confident are you in demonstrating the competency?	13	1	17	43	66	91 percent	3.4
		How frequently do you apply this competency?	12	6	20	35	67	91 percent	3.3
3	Describe the importance of family history in assessing predisposition to disease.	How important is the competency?	12	1	1	22	10 4	91 percent	3.8
		How confident are you in demonstrating the competency?	11	5	11	39	74	92 percent	3.4
		How frequently do you apply this competency?	12	11	27	43	47	91 percent	3.0
4	Identify opportunities and integrate genetic/genomic issues into public health practice, policies or programs effectively.	How important is the competency?	10	0	1	28	10 1	93 percent	3.8
		How confident are you in demonstrating the competency?	11	2	23	56	48	92 percent	3.2
		How frequently do you apply this competency?	12	8	37	36	47	91 percent	3.0
5	Maintain up-to-date knowledge of genetics and genomics-related policies, legislation, statutes, and regulations.	How important is the competency?	14	0	4	42	80	90 percent	3.6
		How confident are you in demonstrating the competency?	15	4	34	55	32	89 percent	2.9
		How frequently do you apply this competency?	15	13	43	44	25	89 percent	2.6
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?	14	0	2	33	91	90 percent	3.7
		How confident are you in demonstrating the competency?	15	2	20	49	54	89 percent	3.2
		How frequently do you apply this competency?	18	10	39	32	41	87 percent	2.9

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7	Collaborate with existing and emerging health agencies and organizations, academic, research, private and commercial enterprises, and community partnerships to apply genetics and genomics knowledge and tools to address public health problems.	How important is the competency?	14	1	1	27	97	90 percent	3.7
		How confident are you in demonstrating the competency?	16	6	26	54	38	89 percent	3.0
		How frequently do you apply this competency?	15	13	46	34	32	89 percent	2.7
8	Identify the resources available to assist clients seeking genetic/genomic information or services, including the types of genetics professionals available.	How important is the competency?	13	0	6	25	96	91 percent	3.7
		How confident are you in demonstrating the competency?	14	7	29	37	53	90 percent	3.1
		How frequently do you apply this competency?	18	24	34	30	34	87 percent	2.6
9	Conduct outcomes evaluation of available genetic/genomic programs and services to determine their effectiveness.	How important is the competency?	16	1	8	34	81	89 percent	3.6
		How confident are you in demonstrating the competency?	16	19	29	49	27	89 percent	2.7
		How frequently do you apply this competency?	16	44	48	19	13	89 percent	2.0
10	Identify the political, legal, social, ethical, and economic issues associated with integrating genomics into public health.	How important is the competency?	16	0	4	26	94	89 percent	3.7
		How confident are you in demonstrating the competency?	17	9	23	44	47	88 percent	3.0
		How frequently do you apply this competency?	18	17	41	33	31	87 percent	2.6
11	Use information technology (IT) to obtain credible, current information about genetics; to utilize IT skills to share data and participate in research, program planning, evaluation, and policy development for health promotion and disease prevention.	How important is the competency?	16	1	4	38	81	89 percent	3.6
		How confident are you in demonstrating the competency?	16	11	25	54	34	89 percent	2.9
		How frequently do you apply this competency?	17	22	36	30	35	88 percent	2.6
12	Identify appropriate and relevant genetics research findings that can be translated into public health policies or practices.	How important is the competency?	16	0	5	24	95	89 percent	3.7
		How confident are you in demonstrating the competency?	17	5	22	51	45	88 percent	3.1
		How frequently do you apply this competency?	19	12	47	33	29	86 percent	2.7

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No Answer	Not at all important	Of little importance	Somewhat important	important	Very important
0	1	2	3	4	5
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

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No Answer	Not at all adequate	Somewhat adequate	Adequate	Very adequate
0	1	2	3	4
How adequate are your resources for implementing genetic/genomic competencies into your work/role? (123 total responses; 88 percent)				
17	28	51	27	17

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

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**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,

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- 3982 and evaluating resources and materials. Furthermore, these organizations have involved these  
3983 communities in the dissemination of resources and materials.
- 3984 ● **Training and Education:** Organizations are actively involved in training and educating local public  
3985 health providers, undergraduate and graduate students, teachers, lay health advisors, and parents  
3986 within their communities to foster outreach and community education. Training includes a wide array  
3987 of strategies including curricula development for high school science teachers, training in genetic  
3988 epidemiology targeting students, collaboration with local health departments to train and educate  
3989 health professionals, and funding research and outreach efforts involving genetic services and  
3990 community engagement via presentations at health fairs and conferences. Most training and education  
3991 sessions involve topics such as communicating risk, genetic literacy levels, and how to target health  
3992 messages to different audiences, particularly vulnerable and/or underserved populations.
  - 3993 ● **Genetic Services:** Respondents indicated that some organizations are involved in engaging  
3994 communities through the provision of genetic services, particularly genetic counseling, either through  
3995 in-person sessions or teleconference calls. Another important type of genetic service is provision of  
3996 genetic testing or provision of monetary assistance to organizations that provide genetic testing to all  
3997 populations including vulnerable and/or underserved populations.
  - 3998 ● **Research:** Some organizations are involved in funding and conducting research to understand  
3999 barriers to genetic services and community involvement in accessing these services. Appropriate  
4000 measures and efforts are undertaken to overcome and resolve those barriers in order to better facilitate  
4001 and engage vulnerable and/or underserved populations.
  - 4002 ● **Funding:** Most respondents viewed the limited availability of funds as a potential barrier to outreach  
4003 and community engagement involving vulnerable and/or underserved populations. They  
4004 recommended that funding should be increased and appropriately allotted to enhance genetic services,  
4005 outreach, and partnerships with vulnerable or underserved populations.
  - 4006 ● **Websites:** Some respondents recommended that websites should be a part of outreach tools that can  
4007 be easily accessed by clients. Materials should be readily available to view or download.
  - 4008 ● **Policy:** Other respondents recommended that federal policy needs to facilitate state and local policy  
4009 by involving legislators, local community leaders, and community members to enhance genetic  
4010 services, raise awareness, and increase education of local community members about their efforts  
4011 within the vulnerable and/or underserved communities.

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4013 2. The survey closed with an opportunity for individuals to provide additional comments to  
4014 SACHGS on the topic of genetics and genetics education for public health providers. Fifty-four  
4015 responses were received, describing the following themes:

- 4017 ● **Funding:** Funding should be provided to develop and implement genetic curricula and training  
4018 programs, integrate genetics education into public health programs such as newborn screening,  
4019 develop ready-to-use tools and resources for local organizations and communities, and provide  
4020 genetic services to all affected families within a community.
- 4021 ● **Networking and Collaboration:** It is vital that organizations within and across states are encouraged  
4022 to share ideas and information concerning the success of programmatic and outreach efforts. The lack  
4023 of networking and collaboration across local, state, and federal level leads to reinvention of programs.
- 4024 ● **Best Evidence-Based Practices:** Public health providers should be trained and educated to identify  
4025 the best practices of genetics and genetics education and incorporate these practices into their services  
4026 and programs.
- 4027 ● **Education:** Some respondents reported that they do not view genetics and/or genetics education as an  
4028 important facet of their profession. Others felt that it is very important and should be integrated into  
4029 their training. Recommended educational topics should include population-level epidemiology,  
4030 review of widely publicized research findings, understanding the concept of risk associated with  
4031 single nucleotide polymorphisms, clinical validity and utility, analysis of family health histories, and

4032 the ELSI issues. In addition, many respondents recommended that basic education about genetics,  
4033 genomics, and its related competencies should be provided to public health providers that include but  
4034 are not limited to nurse practitioners, midwives, primary health care centers, outpatient clinics,  
4035 nutritionists, physicians and childbirth educators.

4036 **Appendix D: SACGHS Study of Consumer and Patients**

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**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**

<b>Perceptions about consumers’ understanding of genetics and genomics</b>
<ul style="list-style-type: none"> <li>• Segments of the general public are struggling to stay abreast of rapidly advancing genetic technologies and the potential benefits and risks of these technologies.</li> <li>• 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.</li> <li>• Segments of the public have a common misconception that genetic predisposition is deterministic.</li> <li>• Segments of the public do not understand complex traits and that there are multiple risk factors for a single health condition.</li> </ul>

<b>Challenges consumers face in obtaining information about genetics and genomics</b>
<ul style="list-style-type: none"> <li>• Finding accurate information about genetics and genomics is difficult.</li> <li>• The public includes many diverse cultures and languages that have different concepts and words to describe inheritance.</li> </ul>
<b>Where people get information</b>
<ul style="list-style-type: none"> <li>• From a variety of sources including the news, television, Internet, local and religious communities.</li> </ul>
<b>Successful and suggested models for genetics education</b>
<ul style="list-style-type: none"> <li>• When developing programs, organizations must assess and understand the needs of the specific community.</li> <li>• 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.</li> <li>• Enhance the communication skills of researchers so scientific concepts and the importance of research and public participation can be fostered among consumers and patients.</li> <li>• 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.</li> <li>• 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.</li> </ul>
<b>The role of government in activities related to genetics education of the public</b>
<ul style="list-style-type: none"> <li>• 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.</li> <li>• 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.</li> <li>• On a societal level, it was felt that the government should play a monitoring role.</li> <li>• The government can influence education and support formal genetics education in schools and update the National Science Education Standards.</li> <li>• All of the interviewees agreed the government should fund more programs to improve genetic literacy.</li> </ul>

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4076 2. Consumers' Survey Instrument  
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**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.**

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

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Department of Health and Human Services

**Privacy Statement**

Your participation in this survey is completely voluntary. Please be assured that your participation in the survey will be kept confidential and your responses will never be linked or associated with you. You may skip any questions that you prefer not to answer. You are also free to stop participating at any point during the survey and have your responses deleted by clicking the "Opt out of survey" box at the bottom of each survey page.

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**Page 1** % Completed

1. Have you been involved with planning or implementing a genetics education program for seekers of genetic information?

Yes  
 No

2. Based on your opinion, please rank the concepts individuals most need to know about genetics and genomics to be informed seekers of genetic information as it relates to health. *(Rank 1-5, 1 being the highest priority. Enter integers only.)*

**Rank 1-5**

- Basic genetic and genomic concepts and terminology (i.e. inheritance, what is a gene; what is a genome)
- Common diseases are caused by complex genetic and environmental factors
- Genetics is relevant to everyone's health
- Family history is an important tool for understanding your health and disease
- Understanding an individual's genetic makeup by itself will not solve all health problems

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

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**Page 2** % Completed

3. Please rank the importance of the following topics that may have special relevance for seekers of genetic information as it relates to health. *(Rank 1-4, 1 being the highest priority. Enter integers only.)*

**Rank 1-4**

- How to access genetic tests
- How to interpret and evaluate the credentials of a genetics professional
- How to interpret results of a genetic test
- Where to find reliable genetic and genomic information

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

4. Please rank the following barriers to genetics and genomics education efforts for seekers of genetic information as it relates to health. *(Rank 1-5, 1 being the most important. Enter integers only.)*

**Rank 1-5**

- Lack of health professionals' understanding of genetics
- Lack of individual health literacy in genetics
- Lack of access to genetic services for consumers/patients
- Direct-to-consumer marketing of genetic tests before there is evidence of their utility or benefit
- Lack of patient understanding of genetic testing implications for themselves or their family (i.e. whether to share results with family members)

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

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Department of Health and Human Services

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**Page 3** % Completed

5(a). Please rank the potential roles in genetics and genomics education of the public for the federal government.  
(Rank 1-6, 1 being the highest priority. Enter integers only.)

	Federal Government (rank 1-6)
Funding genetics education programs	<input type="checkbox"/>
Education about the regulation of genetic services	<input type="checkbox"/>
Education about the regulation of genetic tests	<input type="checkbox"/>
Education about the licensing of genetic health care providers	<input type="checkbox"/>
Education about genetic anti-discrimination laws	<input type="checkbox"/>
Serving as a clearinghouse of educational information	<input type="checkbox"/>

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

Opt out of survey
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Department of Health and Human Services

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**Page 4** % Completed

5(b). Please rank the potential roles in genetics and genomics education of the public for state governments.  
(Rank 1-6, 1 being the highest priority. Enter integers only.)

	State Government (rank 1-6)
Funding genetics education programs	<input type="checkbox"/>
Education about the regulation of genetic services	<input type="checkbox"/>
Education about the regulation of genetic tests	<input type="checkbox"/>
Education about the licensing of genetic health care providers	<input type="checkbox"/>
Education about genetic anti-discrimination laws	<input type="checkbox"/>
Serving as a clearinghouse of educational information	<input type="checkbox"/>

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

Opt out of survey
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 Department of Health and Human Services

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**Page 5** % Completed

5(c). Please rank the potential roles in genetics and genomics education of the public for local governments.  
*(Rank 1-6, 1 being the highest priority. Enter integers only.)*

	Local Government (rank 1-6)
Funding genetics education programs	<input type="checkbox"/>
Education about the regulation of genetic services	<input type="checkbox"/>
Education about the regulation of genetic tests	<input type="checkbox"/>
Education about the licensing of genetic health care providers	<input type="checkbox"/>
Education about genetic anti-discrimination laws	<input type="checkbox"/>
Serving as a clearinghouse of educational information	<input type="checkbox"/>

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

Opt out of survey
Next -->

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Department of Health and Human Services

Page 6 % Completed

6. Please rank the genetic education and services needs of underserved and vulnerable communities and patient populations. (Rank 1-4, 1 being the highest priority. Enter integers only.)

If you believe that there are no genetic education and service needs due to more pressing health education concerns for this population, please check this box and move to question 7.

**Rank**  
**1-4**

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

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

7. If you are part of an organization, to your knowledge has it created any education programs to address the challenges listed in question 6? (Please check all that apply.)

Education about access to genetic services  
 Basic and relevant genetic health information  
 Culturally appropriate genetic health information  
 Skills to make informed health decisions  
 Other (specify):

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Department of Health and Human Services

Page 7 % Completed

8. In your opinion, what role do you think the U.S. Department of Health and Human Service should take to improve genetics education for those seeking information about genetics as it relates to health? (Limit 50 words)

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Department of Health and Human Services

Page 8 % Completed

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

9. In what state do you work?

10. How would you best characterize your organization?  
 Health Care Organization  
 Advocacy Group  
 Public Health Organization  
 Academic Institution  
 Private Industry  
 Other (specify):

11. How important would you say genetics is to the mission of your organization?  
 Extremely important  
 Important  
 Somewhat important  
 Not very important  
 Not at all important

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

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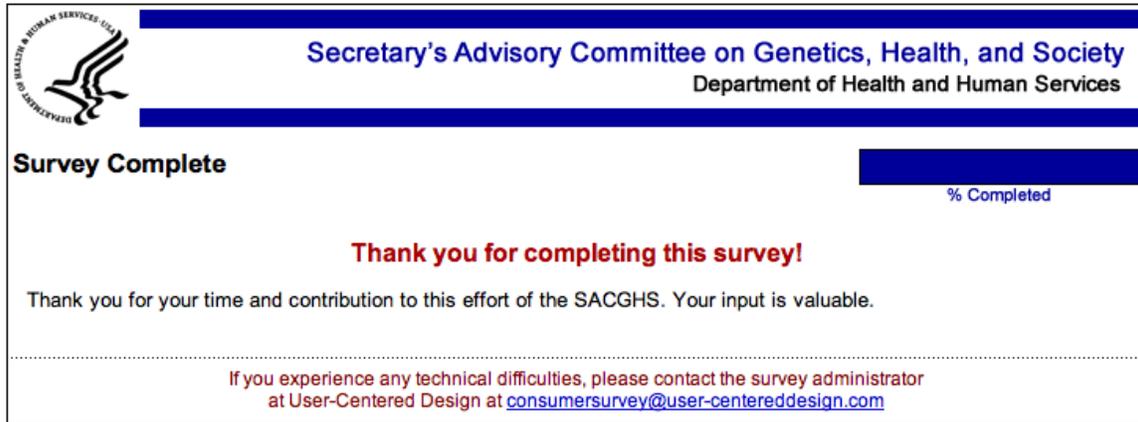
 Secretary's Advisory Committee on Genetics, Health, and Society  
Department of Health and Human Services

Page 9 % Completed

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*).

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

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**Survey Complete** % Completed

**Thank you for completing this survey!**

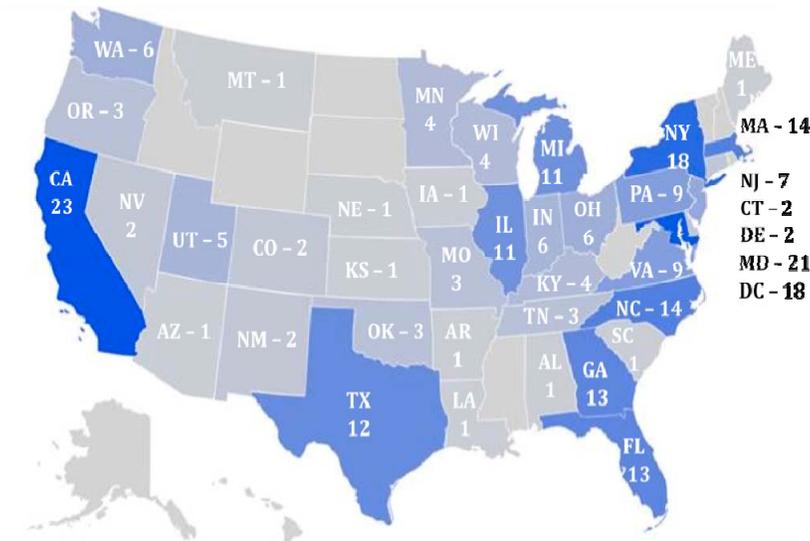
Thank you for your time and contribution to this effort of the SACGHS. Your input is valuable.

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

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**3. Consumer Survey Figures and Tables**

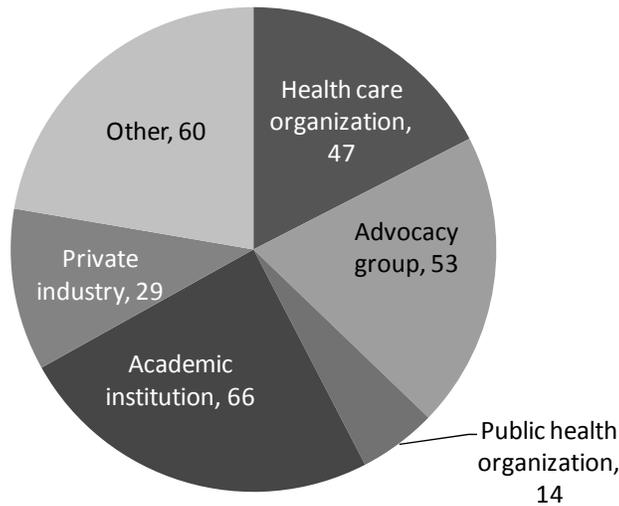
**Figure 1. Geographic Distribution of Responses.** Respondents were asked “In which state do you work?” Responses were received from 258 individuals in 39 states plus the District of Columbia. These respondents are shown in the map below. Numbers refer to the number of responses from each state. The color of each state and the District of Columbia is proportional to the number of responses (darker colors indicate more responses than lighter colors).



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**Figure 2. Distribution of Organization Types.**



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**Table 2. Importance of Genetics to Organizational Mission**

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	#	percent
Extremely important	126	37
Important	75	22
Somewhat important	44	13
Not very important	19	6
Not at all important	5	1
No response	68	20

**Table 3. Concepts for Informed Seekers of Genetic Information**

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

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

Rank	Topics
1	Where to find reliable genetic and genomic information
2	How to access genetic tests
2	How to interpret results of a genetic test
2	How to interpret and evaluate the credentials of a genetics professional

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**Table 5. Genetic Education and Services Needs of Underserved and Vulnerable Populations**

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

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

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

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

<b>Rank</b>	<b>Federal government (82 percent response rate)</b>
1	Funding genetics education programs
2	Serving as a clearinghouse of educational information
3	Education about genetic anti-discrimination laws
4	Education about the regulation of genetic tests
5	Education about the regulation of genetic services
6	Education about the licensing of genetic health care providers
<b>State governments (74 percent response rate)</b>	
1	Funding genetics education programs
2	Education about genetic anti-discrimination laws
3	Education about the regulation of genetic services
4	Serving as a clearinghouse of educational information
4	Education about the regulation of genetic tests
4	Education about the licensing of genetic health care providers
<b>Local governments (65 percent response rate)</b>	
1	Funding genetics education programs
2	Education about genetic anti-discrimination laws
3	Education about the regulation of genetic services
4	Education about the regulation of genetic tests
5	Serving as a clearinghouse of educational information
5	Education about the licensing of genetic health care providers

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4131 **Appendix E: SACGHS Surveys of Federal Agency Activities 2003-2009**  
4132 **Additional Programs and Activities**

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4134 **DOC-NIST**

- 4135 • NIST has built and maintains the world's most widely used, web-based database on forensic  
4136 DNA genetic typing, the STRBase. (<http://www.cstl.nist.gov/biotech/strbase/NIJ/STRBase.htm>).
- 4137 • NIST has also held more than 30 training workshops in forensic laboratories and at major  
4138 scientific conferences to teach genetic principles to scientists and lawyers.  
4139 (<http://www.cstl.nist.gov/biotech/strbase/training.htm>).
- 4140 • NIST Human Identity Project is an ongoing program, begun in 2003, that educates students and  
4141 professionals about genetics and is funded by the Department of Justice.

4142  
4143 **DOD**

- 4144 • Pharmacogenomic Screening: All service members undergo G6PD testing, sickle cell screening,  
4145 and color vision screening, with subsequent environmental and pharmacologic management  
4146 designed to prevent disease.
- 4147 • Newborn Screening Program: The Assistant Secretary of Defense, Health Affairs, has charged the  
4148 Newborn Screening Integrated Project Team with creating policy and a comprehensive military  
4149 newborn screening program that would include a comprehensive educational program, a DOD  
4150 newborn screening website, an EHR-based newborn screening registry, and a comprehensive  
4151 statement of work for a global newborn screening laboratory contract that would be potentially  
4152 available for 50,000 annual births to active duty and retired DOD personnel.
- 4153 • Fellowships: From 2009-2011, the DOD will support the “steady production of one geneticist per  
4154 year” in the Army, as well as two-year genetics fellowships followed by a one-year molecular  
4155 genetics fellowship among Air Force personnel.

4156  
4157 **DOE**

- 4158 • Supported the translation of a high school curriculum unit about genomic science into Spanish.
- 4159 • Sponsored a series of workshops for communities of color in coordination with the Zeta Phi Beta  
4160 sorority organization. More than 1,000 African-American citizens had attended these workshops  
4161 by 2003, where they learned about genomic science and about some of the many clinical, ethical,  
4162 legal, and social implications of genetics research.
- 4163 • JGI program trains faculty to annotate microbial genomes in the context of the undergraduate  
4164 curriculum, and for undergraduate research using tools developed by the JGI. Since many faculty  
4165 need to develop research opportunities for their students, the program gives them the tools and the  
4166 data so that students can carry out bioinformatics research. In the first year and a half of the  
4167 program 55 faculty members and approximately 700 students were trained.
- 4168 • American Society of Microbiology/DOE-JGI Program: a Bioinformatics Institute held twice  
4169 yearly that introduce basic bioinformatics to undergraduate faculty. Dr. Kerfeld, JGI, co-  
4170 organizes the pedagogy for the DOE-JGI/ASM workshops with Professor Brad Goodner, Hiram  
4171 College and, along with additional experts they recruit, they teach this 3-day intensive hands-on  
4172 workshop. From 2004 to 2008 the workshops were attended by approximately 100 faculty  
4173 members and, through them, reached thousands of students with timely and relevant information  
4174 on bioinformatics.
- 4175 • JGI Presentations: Past and upcoming invited presentations include American Society for  
4176 Microbiology Council on Undergraduate Education Meetings in 2007 and 2008; American  
4177 Society for Biochemistry and Molecular Biology Meeting, 2009; Annual International Meeting  
4178 on Microbial Genomics, 2006 and 2008; and the Meeting of the Australian Microarray and  
4179 Associated Technologies Association Meeting 2009.

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- 4180 • Educational websites: Includes the IMG/EDU developed by JGI Genome Biology group in  
4181 collaboration with JGI's Education Program, and the IMG/ACT website developed by JGI.  
4182 ([www.jgi.doe.gov/education](http://www.jgi.doe.gov/education)).

### 4183 HRSA

- 4184
- 4185 • Supports Area Health Education Centers (AHECs) that address health care workforce issues by  
4186 exposing students to health care career opportunities that they otherwise would not have  
4187 encountered, establishing community-based training sites for students in service-learning and  
4188 clinical capacities, providing continuing education programs for health care professionals, and  
4189 evaluating the needs of underserved communities. In 2003, the AHEC program was providing  
4190 community-based continuing education programs to health professionals that included a  
4191 component with genetics content to 9 of 46 participating U.S. medical schools.
  - 4192 • Maternal and Child Health Bureau programs:
    - 4193 ○ Leadership Education in Neurodevelopmental and related Disabilities (LEND)
    - 4194 ○ Heritable Disorders Program, Regional Genetic & Newborn Screening Services (7  
4195 regional screening collaborative centers across the United States and the National  
4196 Coordinating Center)
    - 4197 ○ Consumer Initiatives for Genetics Resources and Services (CIGRS)
    - 4198 ○ National Newborn Screening and Genetic Resources Center
  - 4199 • Bureau of Health Professions programs: A contract was awarded to the National Coalition of  
4200 Health Professional Education in Genetics (NCHPEG) by an IAA among the NHGRI and  
4201 ORD/NIH, CDC, and HRSA to promote health professional education and access to information  
4202 about advances in human genetics. An additional IAA between HRSA and the NIH/NCI was for  
4203 the development of Curricula in Genetics and genomics for Nurse Faculty Development.
  - 4204 • Presentations: Representatives of the Maternal and Child Health Bureau have presented at  
4205 meetings of the American College of Medical Genetics, American Society of Human Genetic, the  
4206 Association of Public Health Laboratories, the Genetic Alliance, and the National Coalition for  
4207 Health Professional Education in Genetics. Staff of NCHPEG have presented at universities in  
4208 Maryland, Michigan, Utah, South Carolina and Louisiana, and to organizations such as the  
4209 American Institute of Biological Sciences, Office of Veteran Affairs, National Society of Genetic  
4210 Counselors, Centers for Disease Control and Prevention Office of Public Health Genomics, the  
4211 International Congress of Human Genetics, and the American Public Health Association.
  - 4212 • The Bureau of Health Professions has held meetings since 2000 on genetics, including an expert  
4213 panel on Genetics and Nursing, 2000, an invitational meeting co-organized with the NHGRI in  
4214 2008 on The Genetics and Genomics Toolkit for Faculty, and additional meetings from 2003 to  
4215 2008 on pharmacogenomics, family history, risk assessment and communications of risk, genetics  
4216 and religion, and genetics and common disease.
  - 4217 • Websites:
    - 4218 ○ A portion of the Genetics/Genomic Toolkit for Faculty may be found at  
4219 [www.genome.gov/17517037](http://www.genome.gov/17517037), along with other resources, curricula, books and online  
4220 courses on genomics and genetics for health professionals.
    - 4221 ○ The IAA with NCHPEG has produced a website ([www.nchpeg.org](http://www.nchpeg.org)) that has steadily  
4222 grown and improved as the number of educational offerings has increased. This website  
4223 is also used to facilitate information sharing, host online surveys, and provide access to  
4224 archived information and slide sets.
    - 4225 ○ The Maternal and Child Health Bureau websites include the Genetics Services Branch  
4226 website, regional genetics and newborn screening collaborative websites, the Sickle Cell  
4227 Disease and Newborn Screening Program, GeneTests-GeneClinics, Community Centered  
4228 Family Health History, March of Dimes Perinatal Data Center, and the National Newborn

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- 4229 Screening and Genetics Resource Center website, among others. All these resources can  
4230 be accessed at <http://mchb.hrsa.gov/>.
- 4231 • Evaluation and Assessment projects:
    - 4232 ○ The Division of Medicine and Dentistry contract allowed NGHPEG to collaborate with
4233 the Genetic Alliance on a survey of consumers of genetic services to access their
4234 perceptions of the genetic competence of their providers.  - 4235 ○ HRSA's Division of Nursing participated with NIH/NCI and NHGRI to determine needs
4236 for nursing education in genetics and genomics.
- 4237 • HRSA staff provide reviews of articles with genetics content for publications such as the Journal
- 4238 of Genetic Counseling, Genetics in Medicine, American Journal of Medical Genetics, and4239 Quarterly Review of Biology, among others.
- 4240 • NCHPEG staff participates in advisory boards and editorial boards with international, national
- 4241 and regional impact such as the Board of Directors/Personalized Medicine Coalition, CDC4242 Advisory Committee on the Use of Family History in Pediatrics, Information and Education4243 Committee/American Society of Human Genetics, and the editorial boards of the journals4244 Community Genetics and Quarterly Review of Biology.
- 4245
- 4246 **NIH**
- 4247 • Trans-NIH projects are administered by the Office of Strategic Operations through the National
4248 Institute on Drug Abuse, the National Institute of Research Resources, the National Institute on4249 Mental Health, the National Institute of General Medical Sciences, and the National Institute of4250 Diabetes and Digestive and Kidney Disorders. These Common Fund programs include:
    - 4251 ○ Clinical Center (CC) Grand Rounds devoted to genetics and genomics
4252 ○ A certificate program in Integrative Biomedical Informatics4253 ○ Development of a curriculum to foster a basic understanding of the correlations4254 between genetic and molecular findings and systems biology, health and disease4255 ○ A post-doctoral program in neuro-developmental toxicology that includes a gene-4256 environmental interaction component4257 ○ A training program in models and technologies for defining phenotypes4258 ○ Post-doctoral training in biobehavioral interventions in developmental disabilities4259 ○ Training programs in pharmacoinformatics4260 ○ Training program in genetics and complex diseases  - 4261 • Genetics education and training programs at individual institutes include programs at the National
4262 Cancer Institute, National Human Genome Research Institute, National Institute on Deafness and4263 Other Communication Disorders, National Institute of Dental and Craniofacial Research,4264 National Institute on Drug Abuse, National Institute on Aging, National Center for Biotechnology4265 Information, and the National Library of Medicine.
    - 4266 • NCI Programs:
      - 4267 ○ Advanced Cancer Risk Counseling Training for Nurses
4268 ○ Clinical Cancer Genetics Education
4269 ○ Genetics Short Course for Cancer Nurses4270 ○ A Cancer Genetics website that includes a cancer genetics overview, cancer genetics4271 risk assessment and counseling, and information about the genetics of breast and4272 ovarian cancer, colorectal cancer, medullary thyroid cancer, and prostate cancer. At4273 this website, one can access links to materials developed and regularly updated by the4274 PDQ Cancer Genetics Editorial Board specifically designed for health professionals.4275 ([www.cancer.gov/cancertopics/prevention-genetics-causes/genetics](http://www.cancer.gov/cancertopics/prevention-genetics-causes/genetics)).

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- NHGRI Programs:
    - Educational materials: educational web casts and interactive web-based learning tools were developed that fulfill recently adopted nursing competencies in genetics education
    - Meetings: a Nursing Champions Meeting and a Primary Care Genetics Summit were held in 2009. The nursing meeting focused on development of a toolkit of genetics educational resources for nurse educators, and the identification of a suitable network of nursing “champions” with expertise in the translation of genetics into health care. The Primary Care Genetics Summit brought together key representatives of primary care physician organizations, such as the American Academy of Family Physicians, to discuss novel approaches to genetics education.
  - NIDCD Summer Program in Genetics for Audiology Faculty included:
    - A needs assessment survey of existing graduate level training programs in audiology that incorporate genetics into their curriculum
    - The establishment of an Advisory Board to guide development of an educational program in genetics
    - The organization of three consecutive 7-day summer workshops targeted to faculty of audiology training programs and the development of an educational notebook for participants in the workshops to assist them in integrating genetics information into their own curricula
    - The establishment of a comprehensive evaluation component to determine the effectiveness of the educational program
  - NIDCR Programs:
    - New Models of Dental Education initiative convened several panels – Genetics and Its Implications for Clinical Dental Practice and Education, held in 2007, and Practical Strategies for Genetics Education in Dentistry, held in 2005.
    - Websites developed include the Genetics in Dentistry Case Simulator ([www.dent.umich.edu/health/index.php](http://www.dent.umich.edu/health/index.php)), and the Genetics, Disease and Dentistry website, [www.nchpeg.org/dental](http://www.nchpeg.org/dental).
    - Publications resulting from NIDCR genetics/genomic educational activities include:
      - Johnson, L., Genco, R.J., Damsky, C., Haden, N.K., Hart, S., Shuler, C.F., Tabak, L.A., and Tedesco, L.A. (2008). Genetics and its implications for clinical dental practice and education: report of panel 3 of the Macy study. *Journal of Dental Education*. 72(2 Suppl):86-94.
      - Dudlicek, L.L., Gettig, E.A., Etzel, K.R., and Hart, T.C. (2004). Status of genetics education in U.S. dental schools. *Journal of Dental Education*. 68(8):809-818.
      - Collins, F., and Tabak, L. (2004). A call for increased education in genetics for dental health professionals. *Journal of Dental Education*. 68(8):807-808.
  - NIDA scientific meeting support included:
    - Travel fellowships to the Jacksonville Short Course in Medical and Experimental Genetics
    - An American Society of Human Genetics satellite meeting on Addiction Genetics Workforce Development and Collaboration. Presentations from the satellite session can be found at [www.sei2003.com/nida/1014039/index.htm](http://www.sei2003.com/nida/1014039/index.htm).
    - Development of a NIDA Short Course on Genetics and Epigenetics of Addiction, presentations can be found at <http://drugabuse.gov/about/organization/Genetics/geneticsepigenetics/index.html>.
    - Participation at the Community Anti-Drug Coalitions of America mid-year training institute conferences.

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- 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

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- Trainings for professionals on genetic discrimination and about GINA, Title II were presented at the following conferences or to the following organizations:
    - Blind Lawyers Association, Washington Seminar (January 2005)
    - SACGHS (June 2005)
    - Annual EXCEL Conference for federal agency EEO and HR professionals and federal agency counsel (August 2007)
    - Defense Contract Audit Agency (DCAA) Annual EEO Conference (November 2007)
    - ABA Labor and Employment Section meeting (March 2008)
    - Upper Midwest Employment Conference (May 2008)
    - Technical Assistance Program Seminars (TAPS) in Denver and Albuquerque (June 2008)
    - American Law Institute-American Bar Association Webcast (July 2008)
    - WEB Employee Benefits Luncheon (July 2008)
    - West Legalworks Webcase (August 2008)
    - TAPS presentation in Richmond VA (August 2008)
    - Department of Labor/National Association of State Workforce Agencies 19<sup>th</sup> Annual National Equal Opportunity Professional Development Forum (August 2008) New York City Practicing Law Institute (October 2008)
    - ABA/Joint Committee on Employee Benefits Meeting (October 2008)
    - National Association of ADA Coordinators National Conference in Las Vegas (October 2008)
    - TAPS presentation for Trenton/NYC area (October 2008)

### NSF

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- 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 6-8<sup>th</sup> 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

- 4377 that supports teachers as learners, especially in terms of learning scientific content  
 4378 and pedagogical tools and techniques. The efficacy of this project will be examined  
 4379 by comparing the performance, on standards-based assessments, of 8<sup>th</sup> grade students  
 4380 who participated in the 3-year curriculum to those who come from a comparable  
 4381 classroom with alternate materials.
- 4382 ○ *The GENIQUEST (GENomics Inquiry through Quantitative Trait Loci Exploration*  
 4383 *with SAIL Technology): Bringing STEM Data to High School Classrooms.*  
 4384 GENIQUEST seeks to develop and test software which will put authentic biological  
 4385 data, along with powerful analysis tools, at the disposal of high school teachers and  
 4386 students. This software assists the framing of testable questions based on this data, at  
 4387 a level appropriate to the students' intellectual capacity, thereby increasing the  
 4388 knowledge of biology, data analysis, the nature of science, and computational  
 4389 biology.
  - 4390 ● Math and Science Partnership Program projects include:
    - 4391 ○ *The Geneticist-Educator Network of Alliances (GENA) Project.* A collaboration of  
 4392 the American Society of Human Genetics, the Genetics Society of America, the  
 4393 National Science Resources Center and the National Association of Biology  
 4394 Teachers, GENA provides tools to instruct, facilitate, and measure meaningful  
 4395 engagement of secondary STEM faculty through the outreach of geneticists at any  
 4396 level. The project seeks to develop a network of master Geneticist-Educator alliances  
 4397 to design strategies to maximize the effective and meaningful interaction between the  
 4398 geneticists and students. This project will serve as a model which may be adapted to  
 4399 other disciplinary scientific societies.
    - 4400 ○ *Baltimore Research and Innovations for New-STEM Partnerships.* The MSP-Start  
 4401 "BRAIN-STEM" project is a partnership between Morgan State University and  
 4402 Baltimore City Public School System which seeks to integrate mathematical and  
 4403 biological concepts suitable for high school courses, beginning with discrete  
 4404 mathematics and genomics. The project addresses the content and pedagogical needs  
 4405 of Baltimore school teachers, based on a needs analysis.
  - 4406 ● Course, Curriculum and Laboratory Improvement Program projects include:
    - 4407 ○ *Literature-Based Scientific Learning in Genetics.* Using constructivist learning and a  
 4408 collection of literature-based case studies, the project strives to promote scientific  
 4409 thinking, conceptual understanding and scientific information competence. The  
 4410 results for this experiential scientific learning project will be developed into an  
 4411 interactive, inquiry-based electronic textbook. The project may serve as a model for  
 4412 other disciplines and is expected to impact the training of future science teachers by  
 4413 involving graduate and undergraduate student assistants.
    - 4414 ○ *The New Genetics: Electronic Tools for Educational Innovation.* This project aims to  
 4415 create and evaluate an innovative set of educational materials. Using an interactive  
 4416 CD-ROM courseware, the project combines genetic and genomic science,  
 4417 technological concepts, environmental, agricultural and biomedical applications, and  
 4418 societal and ethical issues, thereby engaging student interest in the cutting edge of  
 4419 science. This project also expects to create informed citizens who understand science,  
 4420 are excited about the fruits of scientific research, and advocate for public support of  
 4421 scientific research and education. The model will be evaluated in several courses  
 4422 offered in numerous community colleges, a state university and a private university  
 4423 in California, providing a balanced evaluation under widely varying classroom  
 4424 conditions.
    - 4425 ○ *Pathways for New Laboratory Modules in Undergraduate Genetics and Cell*  
 4426 *Physiology Education: Characterization of Puerto Rican Cassava.* By introducing  
 4427 community-relevant research-based plant specific laboratory activities into upper

- 4428 division Genetics and Cell Physiology courses, the University of Puerto Rico seeks to  
 4429 expose approximately 700 Hispanic undergraduate students per year to modern  
 4430 molecular and cellular technologies. This project not only provides students with the  
 4431 confidence to trust in their abilities to learn, understand and implement techniques in  
 4432 modern science, but also leads to the sustainable management of cassava Puerto  
 4433 Rican genetic resources, a real world application of the science students learn in a  
 4434 more traditional setting.
- 4435 ○ *Project Laboratory in Genetics and Genomics*. By creating a new laboratory course,  
 4436 Brandeis University will provide “a myriad” of new experiences for its undergraduate  
 4437 biology students. Students will look at transposon mutation in *E. coli*, for example,  
 4438 and then integrate their findings with public domain genomic information resources  
 4439 to develop a web page for each gene investigated. The project provides students with  
 4440 greater access to a real research laboratory experience, as well as integrating the  
 4441 expertise of both research and teaching faculty who do not now collaborate on course  
 4442 design. Students are assessed before and after the course, for their level of mastery of  
 4443 basic cellular and molecular processes and for their attitudes towards, and  
 4444 understanding of, scientific research. In addition, students evaluate the value of  
 4445 various aspects of the course, to aid in its future refinements.
  - 4446 ○ *ComGen: The Community College Genomics Research Initiative*. This project  
 4447 exposes community college students to real-world research experiences in genomics.  
 4448 This reversal of normal research hierarchy will strengthen the pipeline of students  
 4449 engaged in scientific discovery and excited about STEM careers by including  
 4450 students before they have made a major commitment to a STEM field. This effort  
 4451 will be evaluated for its potential for replication at community colleges nationwide.
  - 4452 ● Advanced Technological Education Program project:
    - 4453 ○ *Innovating Biotechnology Education: Incorporating Novel Genomics Research in the*  
 4454 *Development of a True 2+2+2 Educational Pathway*. In response to a shortage in  
 4455 research-skilled laboratory technicians, Mesa Community College proposes a 2+2+2  
 4456 program. This program is unique because it uses genomics research to prepare high  
 4457 school science instructors with skills and curriculum to prepare their students for the  
 4458 rigors of post-secondary degrees in biotechnology related fields. If successful, this  
 4459 model can easily be integrated into other biotechnology programs around the country.
  - 4460 ● NSF Scholarships in STEM projects include:
    - 4461 ○ *Proteomics and Functional Genomics Scholarship Program*. This scholarship  
 4462 program is designed for talented but financially needy students. The project aims to  
 4463 support more than 20 students who will eventually attend graduate school or obtain  
 4464 jobs in proteomics and functional genomics or related fields.
    - 4465 ○ *BHSU Integrative Genomics Transition Scholarship Program*. This program will  
 4466 provide support to 20 Master’s degree students in the emerging area of Integrative  
 4467 Genomics, as well as 10 scholarships for undergraduate biology majors with an  
 4468 interest in pursuing the Master’s degree in this area. Furthermore, the project is  
 4469 creating a pipeline to the Integrative Genomics program for Native American Indian  
 4470 students which should increase overall the number of Native American Indian STEM  
 4471 graduates pursuing advanced degrees.
  - 4472 ● Historically Black Colleges and Universities-Undergraduate Program project:
    - 4473 ○ *Targeted Infusion Project: Integration of Plant Genomics into the Undergraduate*  
 4474 *Curriculum*. This project will incorporate plant genomics into the undergraduate  
 4475 curriculum of the Plant Science and Biology departments. A Plant Genomics senior  
 4476 level course will be developed and newly designed genomics modules will be  
 4477 incorporated into several existing courses, thus preparing students in these courses  
 4478 for various careers in the biological sciences, and the burgeoning fields of genomics

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- 4479 and bioinformatics. The teaching materials developed at one university will be  
4480 widely disseminated through a variety of media.
- 4481 • Interdisciplinary Training for Undergraduates in Biological and Mathematical (UBM)  
4482 Sciences project:
    - 4483 ○ *Undergraduate Training and Research in Applied Mathematics and Biological*  
4484 *Sciences*. This project builds on an existing undergraduate major in Applied  
4485 Mathematics-Biology. Student teams work on joint projects in physiology and  
4486 genomics with faculty advisors and alongside graduate students and post-doctoral  
4487 associates. This project provides students with a background in mathematics and  
4488 biological science that will prepare them for future interdisciplinary graduate level  
4489 programs.
  - 4490 • Centers of Research Excellence in Science and Technology project:
    - 4491 ○ *CREST Center in Tropical Ecology and Evolution of Marine and Terrestrial*  
4492 *Environments*. The goal of this program is to become a highly collaborative research  
4493 center in tropical conservation biology and environmental sciences in Hawaii. The  
4494 program consists of three interconnected subprojects: Evolutionary Genomics and  
4495 Ecology of Local Adaptation and Speciation, Terrestrial Ecology, and Coral Reef  
4496 Ecosystem. The NSF CREST Program will build on the current strengths of the  
4497 center, especially an integrated research and education program that is building the  
4498 STEM pipeline for students in Hawaii from K-12 through to undergraduate and  
4499 graduate programs.
  - 4500 • Integrative Graduate Education and Research Traineeship Program projects:
    - 4501 ○ *IGERT in Chemical Genomics: Forging Complementation at the Interface of*  
4502 *Chemistry, Engineering, Computational Sciences and Cell Biology*. Chemical  
4503 genomics uses small molecules to probe protein function in complex cellular  
4504 systems. This approach offers a strategy which may fill in some crucial gaps in the  
4505 study of functional genomics in plants by addressing the issues of overlapping gene  
4506 function in gene families, lethal loci, and control of dosage and tissue/development  
4507 specific application. The program will prepare graduates with skills for  
4508 multidisciplinary research, acute awareness of the potential for their discoveries to  
4509 address global food, health and environmental problems, of the ethical implications  
4510 of their research, and with exposure to a variety of research environments in  
4511 academia and industry.
    - 4512 ○ *IGERT: Predoctoral Training in Functional Genomics of Model Organisms*. The  
4513 objective of this project is to initiate an interdisciplinary, inter-institutional degree  
4514 program in Functional Genomics of Model Organisms. It is a collaboration of the  
4515 University of Maine, the Jackson Laboratory, and the Maine Medical Center  
4516 Research Institute. As it becomes clear that genome projects, regardless of the  
4517 organism, will rely increasingly on the physical and computational sciences,  
4518 interdisciplinary work and thinking becomes increasingly important. This program  
4519 introduces a new educational paradigm, developed to train students to move freely  
4520 among the disciplines needed to investigate genome function.
  - 4521 • Informal Science Education Program projects:
    - 4522 ○ *Indonesian Origins: Genes, Languages and Culture video programs*. This  
4523 “Communicating Research to Public Audiences” project will produce a quality  
4524 television program that will showcase an interdisciplinary approach to the history of  
4525 the peopling of the Indonesian archipelago, combining genetics, archaeology,  
4526 historical linguistics and ethnography. The primary intended audience is American  
4527 viewers of scientific documentary television programs, although it possibly could be  
4528 shown in secondary schools and colleges.

- 4529                   ○ *The DNA Files III*. SoundVision Productions proposes to develop 5 one-hour radio
- 4530                   documentaries, 5 five-minute features, and a website to inform a diverse public about
- 4531                   important advances in genomics and related sciences. The project will offer
- 4532                   audiences an awareness of the societal benefits of research and the intellectual tools
- 4533                   to join in legal and social policy debates. A comprehensive outreach strategy will be
- 4534                   implemented by 20 local public radio stations around the country in partnership with
- 4535                   community organizations.

4536  
4537                   **THE END**