Innovations in Service Delivery in the Age of Genomics: Workshop Summary

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Erin Hammers, Rapporteur; Roundtable on Translating Genomic-Based Research for Health; Institute of Medicine

INNOVATIONS IN SERVICE DELIVERY IN THE **AGE OF GENOMICS**

WORKSHOP SUMMARY

Erin Hammers, *Rapporteur*

Roundtable on Translating Genomic-Based Research for Health

Board on Health Sciences Policy

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Willing is not enough; we must do."
—Goethe

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This report has been reviewed in draft form by individuals chosen for their diverse perspectives and technical expertise, in accordance with procedures approved by the National Research Council's Report Review Committee. The purpose of this independent review is to provide candid and critical comments that will assist the institution in making its published report as sound as possible and to ensure that the report meets institutional standards for objectivity, evidence, and responsiveness to the study charge. The review comments and draft manuscript remain confidential to protect the integrity of the deliberative process. We wish to thank the following individuals for their review of this report:

David R. Nerenz, Ph.D., Center for Health Services Research, Henry Ford Hospital and Henry Ford Health System Neuroscience Institute

Karen Powell, M.S., C.G.C., Center for Biotechnology, Genomics and Health Research, The University of North Carolina at Greensboro

Sylvia Trujillo, J.D., M.P.P., Division of Legislative Counsel, American Medical Association

David Veenstra, Ph.D., Institute for Public Health Genetics and Department of Pharmacy, University of Washington

Beverly M. Yashar, M.S., Ph.D., Genetic Counseling Graduate Program, Departments of Human Genetics and Ophthalmology and Visual Sciences, Kellogg Eye Center, University of Michigan

Although the reviewers listed above have provided many constructive comments and suggestions, they were not asked to endorse the final draft of the report before its release. The review of this report was overseen by **Dan Blazer, M.D., Ph.D.,** Duke University Medical Center. Appointed by the Institute of Medicine, he was responsible for making certain that an independent examination of this report was carried out in accordance with institutional procedures and that all review comments were carefully considered. Responsibility for the final content of this report rests entirely with the rapporteur and the institution.

Contents

FIGURES

BOX

1

Introduction

New genomic[1] discoveries and technologies are shifting the focus from testing for specific and rare genetic disorders to using genetic testing to predict risk of common diseases and help determine prevention and treatment options. Advances in genetics are contributing to improved understanding of the genetic and molecular basis of disease and are increasingly leading to the development of interventions such as gene-based therapy and pharmacogenomics.

The integration of advances from genetics into the health care system is marked by three major trends. First, information previously handled by medical geneticists and a few specialists is moving into the arena of other medical specialties and primary care. As this transition is made, it will be necessary to deal with associated barriers and challenges. Second, genetic information that has historically been used as an end point will increasingly have the potential to be used to improve health care outcomes. Such a shift requires providers and patients to think about using genetic information in a different way, one more similar to how other health care data are used. Finally, there is a shift from dealing with a limited amount of information to being confronted with a great deal of information. In the past, there may

[1] "Genomics" is the study of the entire human genome—the actions of single genes and the interactions of multiple genes with each other and with the environment, whereas "genetics" is the study of single genes and their functions and effects (IOM, 2003a). However, presenters did not agree in advance to these definitions, so not every presenter refers to these terms in the same way.

have been concern about what was unknown; now the concern is about managing the information that is available.

The current system for delivering genetic services is based on a model of intensive counseling for rare diseases. As the use of genomic technology becomes more prevalent, providers and patients will need new ways of communicating about genetic information and how it may change health care options. Old practice models that rely on extensive education and counseling may not be suitable when patients and payers demand the inclusion of genomic information in making everyday health care decisions. New practice models of service delivery will have to be developed to contend with the rising tide of genomic innovations.

The Roundtable on Translating Genomic-Based Research for Health was established by the Institute of Medicine (IOM) in 2007 to provide a structured opportunity for dialogue and discussion of issues related to the translation of genomic information for use in maintaining and improving health. At its second meeting, following a workshop that focused on the diffusion of genomic innovations, the Roundtable identified the need for a public workshop that would feature presentations and discussion of strategies regarding service delivery in the age of genomics, seeking to understand the current status of service delivery, how needs will change as genomic innovation progresses, and what types of alternative practice models will be needed.

The July 28, 2008, workshop was moderated by Wylie Burke, chair of the Roundtable. Presentations followed by discussion occurred in four areas: the current status of genetic service delivery, strengths and challenges of the current system, new models for service delivery, and a vision for the future. Following these presentations, a panel of workshop speakers and participants brainstormed about a service delivery model for the future. The workshop did not address the ethical or legal considerations surrounding genomic innovations and the various new models of service delivery, including direct-to-consumer marketing of genetic tests. The meeting concluded with a summary of the day's discussions. The complete agenda can be found in Appendix A, and biographical sketches of the speakers are provided in Appendix B.

The following report summarizes speaker presentations and discussions. Any conclusions reported should not be construed as reflecting a group consensus; rather, they are the statements and opinions of the presenters and participants.

2

Genetic Service Delivery: The Current System and Its Strengths and Challenges

CURRENT STATUS OF GENETIC SERVICE DELIVERY

Debra Lochner Doyle, M.S., C.G.C.
Washington State Department of Health

The Washington State Department of Health and the University of Washington entered into a cooperative agreement in 2004 with the Health Resources Services Administration to establish the Genetic Services Policy Project (GSPP). The purposes of this project are to

- characterize how genetic services are currently delivered within the United States,
- explore what kinds of issues in the pipeline will affect the delivery system,
- evaluate potential alternative models for the delivery of genetic services, and
- identify and assess public policies that could better promote cost-effective, accessible, and equitable delivery of services (Doyle and Watts, 2008).

To accurately assess what the future of genetic service delivery will look like, a necessary first step is to collect information about the current system. A major difficulty in collecting such information is that there is no agreed-upon description of what is included in the term *genetic services*.

For purposes of the GSPP, genetic services were defined to include genetic testing, diagnosis of genetic conditions, genetic counseling, and treatments for individuals with, or at risk of, genetic disorders. Genetic testing includes laboratory analysis of DNA, RNA, chromosomes, or gene products, with the exception of genetic analysis of pathogens, "recreational" genetics (e.g., ancestry and dating services), paternity testing, and forensics.

Using this definition, GSPP compiled data on genetic services capacity, socioeconomic and political variables, and relevant legal and regulatory information for all 50 states in the United States. The resulting report, released in 2008, describes what genetic services are delivered, who delivers them, who receives them, and where services are provided. Additionally, the report describes genetic services offered throughout the stages of life, from preconception to prenatal to newborn, pediatric, and adult testing.

Just as the definition of genetic services varies, genetic service providers are an equally indistinct group. Genetic service providers can be categorized into two general groups: those who are formally trained and certified in genetics (e.g., genetic counselors, medical geneticists, genetics nurses) and all other providers. Credentialing organizations make it possible to identify and count those providers with formal genetics education and training. However, it is extremely difficult to determine how many and what type of other health care providers are offering genetic services as part of their practice. For example, most obstetricians and pediatricians are not formally certified in genetics, but it is standard practice for these providers to offer carrier screening to prospective parents or to do chromosome studies on a child who is suspected of having a genetic abnormality such as Down syndrome. Unfortunately there is a huge data gap that hinders attempts to research the current system because there is no way to count, let alone characterize, all health care providers who may offer genetic services.

Furthermore, given that many non-genetics professionals are offering genetic services, there is concern about the quality of care being provided. For example, in a survey of 363 physicians from Mount Sinai Medical Center, 71 percent rated their knowledge of genetics and genetic testing as "fair" to "poor," and almost all said they would refer their patients to a genetic counselor (Menasha et al., 2000). In a study about testing of the adenomatous polyposis coli (APC) gene for familial adenomatous polyposis, 31 percent of physicians interpreted the results of the test incorrectly (Giardiello, 1997). Forty-two percent of pediatricians surveyed in Massachusetts indicated feeling "ill prepared" to talk to families about the results of expanded newborn screening (Gennaccaro et al., 2005). To ensure that patients receive up-to-date, timely, and accurate information, it is crucial that anyone who offers or refers patients to genetic services—formally trained or not—has the knowledge and skills necessary to provide quality services.

In addition to defining genetic services and examining who the providers are, it is important to identify who is receiving these services. Unfortunately, there is no easy way to collect this information. Health insurance claims data have significant limitations. CPT (Current Procedural Terminology, copyrighted by the American Medical Association) codes for cognitive services are the same whether the service provided is a consultation with a medical geneticist or a consultation with any other medical professional. However, it is possible to infer who is receiving services using estimates based on standards of care. For example, nearly all of the 4 million infants born each year receive newborn screening (March of Dimes, 2007), and most pregnant women are offered certain genetic services such as multiple marker maternal serum screening, which is standard practice in obstetrics.

Some areas of the country are collecting data that help identify who receives services. For example, Washington State has compiled service utilization data since 1991 from its 15 regional genetic clinics, finding that, on average, there has been an increase of about 8 percent per year in individuals seeking genetic services, most of them adults (Wang and Watts, 2007). Four states—Michigan, Minnesota, Utah, and Oregon—have added questions about genetic services to their Behavioral Risk Factor Surveillance System (BRFSS) survey. However, there remains a severe data gap in the pursuit of a complete picture of the current genetic service delivery system.

Another important component of understanding the current system for delivery of genetic services is identifying where these services are provided. The GSPP compiled information from professional organizations such as the American College of Medical Genetics, the International Society of Nurses in Genetics, and the National Society of Genetic Counselors. On the basis of professional status survey results and membership data, GSPP determined both the geographic location and the professional setting of genetic service provision. These data are limited, however, because they include only information about formally credentialed genetics professionals, even though many genetic services are provided by non-geneticists.

The vast majority of genetics professionals work in academic medical centers, followed in order of magnitude by public and private hospitals and medical facilities; commercial, diagnostic, and state laboratories; private practice; and the insurance industry. On average in the United States, there are 1 to 1.5 genetics professionals per 100,000 residents. These genetics professionals are concentrated on the West Coast and in the Northeast, a pattern similar to the distribution of all medical doctors per capita.

Despite the availability of information about how many genetics professionals exist, there is another major data gap: no one knows how many genetics professionals are actually needed to ensure access to, and quality of, care. As genetic and genomic technologies advance, and as the public

learns more and demands more, it is likely that there will be an increased need for genetics professionals. Yet no information is available to suggest a number or ratio of genetics-trained professionals that would be sufficient to serve the needs of the public.

As the GSPP gathered data and attempted to characterize the state of the current genetic service delivery system, several data gaps were identified, some of which have been mentioned. Claims data are severely limited in their usefulness because CPT codes are highly variable and not specific enough to identify when genetic services are being provided. Data are often proprietary, making them unavailable or costly. There are few data that demonstrate consumer demand or utilization of retail genetics, whether these services are marketed directly to consumers or to providers. Data suggest low levels of certified genetic service providers nationwide, but there are no data to indicate optimal numbers.

Our current health care system is already fairly fragile. As genomic innovation progresses it is likely that further strains will be placed on systems and providers. It will be imperative, therefore, for researchers to fill these data gaps in order to describe, monitor, and evaluate the provision of genetic services now and in the future.

CHALLENGES OF DISPARITIES AND ACCESS

Alexandra Shields, Ph.D.
Harvard/Massachusetts General Hospital Center on Genomics, Vulnerable Populations & Health Disparities

Health disparities have recently come under greater scrutiny, thanks in part to two Institute of Medicine reports. In *Crossing the Quality Chasm,* the alleviation of health care disparities was, for the first time, noted as one of the six aims for quality improvement in the U.S. health care system: "The availability of care and quality of services should be based on individuals' particular needs and not on personal characteristics unrelated to the patient's condition or to the reason for seeking care. In particular, the quality of care should not differ because of such characteristics as gender, race, age, ethnicity, income, education, disability, sexual orientation, or location of residence" (IOM, 2001, p. 53). *Unequal Treatment* provided exhaustive documentation of racial and ethnic disparities in health care (IOM, 2003b). These disparities were found in a range of health care settings and across disease areas and clinical services, even when controlling for known predictors such as insurance status, socioeconomic status, and access to care.

The Harvard/Massachusetts General Hospital (MGH) Center is applying what is known about health disparities in general to the more specific area of genomic health care by engaging in research that examines where health disparities exist along the trajectory from research to health outcomes. For example, at one end of the trajectory, research practices have implications for health disparities and the health of underserved populations, since the use of race constructs in genome research or the conceptualization and operationalization of measures of environment can influence our understanding of the effect of these variables on the health of different populations. At the other end of the trajectory, as research is translated into clinical practice, there are disparities in provider knowledge and readiness, health system capacity, consumer willingness to undergo genetic testing, and coverage and financing of health care services. The Harvard/MGH Center also monitors the diffusion and impact of genomic services on health outcomes in disparate populations, looking at issues of access, whether genomics affects diagnosis and prognosis, and how genomics ultimately affects health disparities.

Disparities in access to and utilization of genomic technologies do exist (Levy et al., unpublished; Shields et al., 2008; Wideroff et al., 2003). Possible reasons for differential utilization of available genetic tests among demographic groups include but are not limited to

- confidence in the efficacy of tests;
- provider knowledge, training, and capacity;
- practice linkages with specialty care;
- patient awareness and willingness to undergo genetic testing;
- the low priority of testing for patient populations with high comorbidity loads and complex health issues;
- differential coverage, prior approval requirements, and copayments;
- racism and discrimination; and
- geographic variation.

There are various strategies for identifying disparities in utilization and access among different populations, as illustrated by the following discussion.

One approach to identifying disparities is to examine data about patient or provider knowledge of genetic testing as a contributing factor to low or differential uptake of genomic medicine. In 2000, the National Health Interview Survey (NHIS), which surveys the U.S. population on issues of health and health care, included a supplement that asked the question: "Have you ever heard of genetic testing to determine if a person is at greater risk of developing cancer?" (CDC, 2000). The results were striking. After adjusting for age, sex, region of the country, health insurance status, health care utilization, cancer history, and propensity toward preventive

health care, there still existed a strong gradient of difference by race and education level. Blacks and Hispanics were less likely than whites to have heard of genetic testing, and as education level increased from less than high school to college graduate, knowledge of genetic testing increased as well (Wideroff et al., 2003).

NHIS asked another question in the 2000 supplement: "Have you ever discussed the possibility of getting a genetic test for cancer risk with a doctor or other health professional?" (CDC, 2000). A recent analysis (Levy et al., unpublished) used these NHIS data, in conjunction with the 1999 National Comprehensive Cancer Network guideline criteria, to identify about 35,000 women with no personal history of breast or ovarian cancer and to stratify them into high risk or average risk for hereditary breast or ovarian cancer. Of these, 42 percent of the average-risk women had heard of genetic testing for cancer risk, compared to 55.2 percent of the high-risk group. Only 2.2 percent of the average-risk women had discussed testing with their doctor, and 10.7 percent of high-risk women had discussed it. It is encouraging that higher-risk women are more likely to discuss testing with their physician, but the rates are still quite low in comparison with other kinds of practice guidelines, where compliance rates may be over 50 percent (McGlynn et al., 2003).

Another strategy for studying health disparities is to identify high-volume providers, that is, the subset of providers who care for the majority of a certain subpopulation. A survey of primary care physicians was conducted in 2002 that, in addition to questions about incorporating genomic medicine into practice, included questions about patient characteristics: What proportion are minority, on Medicaid, uninsured, or have a primary language other than English? The physicians were ranked according to the percentage of their patients with the characteristics of interest, and the top 20 percent of physicians in each category were defined as "high minority," "high Medicaid," "high uninsured," and "high non-English" serving. The physicians were asked whether they had ever ordered a genetic test for breast cancer, colon cancer, sickle cell disease, Huntington's disease, or any other genetic test. Those physicians who were high minority-serving were less than half as likely to have ever ordered a genetic test for breast cancer, colon cancer, or Huntington's disease (Shields et al., 2008).

These physicians were also asked whether they had ever referred a patient to a genetic counselor, a specialist, a clinical trial, or any other site of care for a genetic test. Again, high minority-serving physicians were half as likely to have referred a patient to a clinical trial or any site of care. Physicians serving a large percentage of Medicaid patients were half as likely to have referred a patient to genetic counselors or any site of care. The other associations studied were not statistically significant, but it is apparent that not all patients have equal access to genetic testing.

Many poor and minority Americans receive their health care from "safety net" sites such as community health centers (CHCs), which are defined as "providers that organize and deliver a significant level of health care and other related services to uninsured, Medicaid, and other vulnerable patients" (IOM, 2000). More than 75 percent of community health center patients are either uninsured or covered by Medicaid (Rosenbaum et al., 2004), which means that the centers themselves often lack the capital to invest in new technologies and infrastructure (Fiscella and Geiger, 2006). A collaboration between the National Association of Community Health Centers and the National Research Center for Health Information Technology, using data collected by the Health Resources and Services Administration, surveyed 627 health centers and found that 4.3 percent of CHCs provided genetic counseling (either by providing it directly or by referring and paying for the service) (Shields, unpublished). The percentage that provided any genetic testing—outside of prenatal—was 11.7 percent, with about 5 percent for breast and colorectal cancer and 3 percent for Huntington's disease. When these data were analyzed to identify characteristics of CHCs that predicted provision of genetic testing, it was found that those centers with the highest specialist-to-patient ratios, those with the most black patients, and those with the most Latino patients were about twice as likely to provide genetic testing as those centers with lower ratios and fewer minority patients. Interestingly, in this case, the main predictor of genetic service provision was size: centers that served more than 10,000 patients were six times more likely to offer services than smaller centers.

Examining the areas of reimbursement and the ability to pay for genetic services is another approach to studying disparities in genetic testing among different populations. Clearly, access to and ability to pay for genetic testing strongly affect whether one will receive these services. However, there seem to be disparities in the utilization of genetic tests even among persons who have identical insurance that covers testing. The Harvard/MGH Center recently examined claims data from 2004 to 2008 for about 15 million commercially insured individuals to determine who had undergone genetic testing for cancer. Specific data examined included data on BRCA1 and BRCA2 tests, which are generally considered clinically valuable in the screening, diagnosis, and treatment of breast cancer and are covered by the commercial insurer. Data were also examined regarding the MLH1 and MSH2 genetic test, which has been shown, in conjunction with appropriate surveillance, to result in a significant decrease in colorectal cancer incidence and overall mortality (Jarvinen et al., 2000). Overall, 10.98 percent of this population received the BRCA1 and BRCA2 test, and 1.42 percent received the MLH1/MSH2 test. Utilization varied significantly by race: whites and Hispanics were almost twice as likely to have received the tests. The difference in utilization by household income was striking; a clear gradient

showed that the highest-income patients were more than four times as likely to have received the BRCA1 and BRCA2 test than the lowest-income patients. This gradient existed despite the fact that all patients were in the same health plan and had the same coverage. Further research is needed to understand this apparent discrepancy: Is the disparity due to patient knowledge or attitudes, provider willingness, or other unknown factors?

It is evident from these early data that health care disparities exist throughout the health care system, including in the area of genetics. As genomic medicine progresses, research is essential to assess the effect of genomic medicine on underserved populations. Critical questions for a research agenda include the following:

- To what extent do genomic applications improve health outcomes?
- Are there important allele frequencies that differ across populations?
- Are there disparities in coverage across plans (e.g., Medicaid versus commercial insurance)?
- Are there disparities in access to genomic medicine? Are these disparities different or the same as other documented health care disparities?
- What is the origin of these disparities—providers, patients, policies, culture, or racism?
- How do disparities in access and utilization affect the gap in clinical outcomes?
- What data infrastructure is needed to answer these questions?

The Harvard/MGH Center on Genomics, Vulnerable Populations & Health Disparities is working to help answer these questions, expanding methods for capturing genomic medicine in administrative data, conducting patient surveys, and exploring the idea of using electronic health records to collect data for use in studying health disparities. One of the key measures against which the investment in genomics research should be judged is its ultimate impact on the health of underserved populations. It is essential that researchers use every tool at their disposal to be able to measure, assess, and understand the factors that cause these disparities to persist.

PATIENT EDUCATION AND COMMUNICATION

Vivian Ota Wang, Ph.D., F.A.C.M.G., C.G.C.

Genetic information and genetic services have become increasingly complex, with multiplex testing, predictive risk testing, and clinical util-

ity and validity evidence that is ambiguous and constantly changing. Providers—whether genetic counselors, physicians, nurses, pharmacists, or specialists—are frequently confronted with the issue of how to convey genetic information effectively to their patients. According to Ota Wang, translating genetic information into something that people can comprehend requires understanding four crucial elements of cognitive psychology: how information is categorized, where attention is focused, how information is processed, and what is culturally responsive communication.

First, in terms of categorization, when humans absorb new information, they use five factors to categorize what they are processing: similarity, simplification, proximity, continuity, and perception. People generally group things that are most *similar* together. They *simplify* information into its easiest form. People group things that are located near each other using the law of *proximity*. They also tend to follow the law of *continuity*; that is, they see something as following a smooth, logical path, rather than breaking it up into parts. Finally, objects that are grouped together tend to be *perceived* as a whole.

Second, when looking at a picture, people focus on different parts of it. Some pay attention to the foreground; others see the background. A classic example is the picture in Figure 2-1, in which one can see a vase or two faces, depending on where attention is focused.

Third, research in cognitive psychology shows that when people are confronted with a large amount of information, some focus on the details that matter, while others are distracted by the excess information. Some think in cognitively complex ways—they can process large amounts of information that is abstract, ambiguous, and uncertain; others think in cognitively simple ways—they can process only limited amounts of concrete information.

Finally, not only do people from different cultures speak different languages, they also communicate in different ways. In languages that are considered "low context," information is conveyed primarily through direct verbal and written communication (e.g., Danish, German, English). Other languages are "high context." In these languages the surroundings and the context are far more important than the literal meaning of the words (e.g., Japanese, Chinese, Vietnamese, French, Spanish, Greek). In low-context communication, background information is made explicit, whereas in high-context communication, the full message must be interpreted by the listener through nonverbal cues and indirect references.

It is imperative for providers to understand that their patients process information in ways that are different from the provider and different from each other. Information can be grouped and categorized in various ways, it can be presented abstractly or concretely, and it can be tailored to different cultural communication systems. Providers often assume that more information is better information, whether it is on a consent form, about a

FIGURE 2-1 Attention focusing: Vase or two faces.
SOURCE: Adapted from Ota Wang, 2008.

test result, or in a discussion about disease. To help patients fully comprehend complex genetic and genomic information and take an active role in their own health care, it behooves providers to learn from the research on cognitive psychology in order to design communication strategies to meet each patient's needs.

EDUCATIONAL PIPELINE AND WORKFORCE

Catherine A. Wicklund, M.S., C.G.C.
Northwestern University

Many different types of professionals provide genetic services to patients, including genetic counselors, medical geneticists, and genetics nurses. As genetic and genomic technologies are incorporated into mainstream medicine and patients learn about and request these services, it will be essential to have an adequate workforce to meet the demand. Before one can discuss the need for future providers, however, it is necessary to understand the current status of those who provide genetic services: who they are, what they do, how they are trained, how many there are, and the challenges that their professions face.

Genetic Counselors

Genetic counselors are health care professionals who work as members of a team to provide information on genetic issues to patients and providers. The following are some of their duties:

- identify and introduce the possibility of genetic risk;
- determine a patient's knowledge and motivations;
- ascertain personal and family medical histories via targeted pedigrees;
- provide risk assessment for patient and family;
- educate about condition, inheritance pattern, risk, management, and prevention;
- facilitate informed decision making;
- obtain informed consent for genetic testing;
- counsel to assess psychosocial impact and provide support;
- identify resources for patients; and
- follow up with the patient, including guidance about informing key relatives.

Genetic counselors are trained through a 2-year accredited master's program that consists of three main elements: (1) coursework in diverse topics such as counseling, molecular biology, genetics, ethics, health care, and research methods; (2) clinical training in a variety of clinical settings such as prenatal, pediatric, cancer, neurogenetic, and cardiovascular; and (3) a research component. The American Board of Genetic Counselors (ABGC) sets the academic standards for institutions and provides accreditation to graduate programs in genetic counseling. Since 1993, the number of accredited programs has almost doubled, from 18 to 32. Together, these programs accept about 30 percent of applicants and graduate about 225 students each year.

In addition to accreditation, the ABGC certifies genetic counselors, requiring recertification every 10 years through examination or continuing education. Unlike some other health care professionals, genetic counselors are not typically licensed. Seven states have passed licensure bills, but only two—Illinois and Utah—currently have active licensure.

There are about 2,500 certified genetic counselors in the United States, which equates to about 1 counselor per 123,000 individuals. Geographically, the counselors are distributed roughly according to population, with the majority on the East Coast, in the Midwest, and on the West Coast.

There are several challenges facing the profession of genetic counseling. One is diversity: 91 percent of genetic counselors identify as Caucasian, and most are women under 40 (Parrot and DelVecchio, 2007). Another

is the issue of reimbursement; genetic counselors cannot bill insurers for direct reimbursement, which results in limited access to counseling services, fewer genetic counselors, and lessened integration into the health care system. Two problems—limited funding for graduate programs and a limited number of genetic counselors available to supervise clinical internships—negatively affect the education and preparation of a new generation of genetic counselors. The National Society of Genetic Counselors has developed a strategic plan to address these challenges that includes

- pursuit of federal legislation that recognizes genetic counselors as health care providers,
- support for states in the effort to license genetic counselors,
- development and growth of relationships with third-party payers,
- exploration of alternative service delivery models,
- monitoring trends in health care to determine how genetic counseling can fit in,
- formation of partnerships with other providers to support integration efforts,
- working with the ABGC and the Association of Genetic Counseling Program Directors to address workforce issues, and
- identification of the needs for continuing education.

In addition, the ABGC will begin to offer the genetic counselor board exam annually in an effort to support licensure efforts and increase the availability of clinical training sites.

Medical Geneticists

A clinical geneticist (also known as an *MD geneticist* or *physician geneticist*) "holds a U.S. or Canadian earned or the equivalent of an earned M.D. or D.O. degree, has had 2 years in an ACGME[1]-accredited clinical residency program in another medical specialty, 2 years in an ACGME-accredited residency in clinical genetics (or 4 years in an accredited clinical genetics residency program), a valid medical license, and demonstrates competence to provide comprehensive genetic diagnostic, management, therapeutic, and counseling services" (ABMG, 2008a). The clinical geneticist's scope of practice is broad, given that genetic issues apply to all organ systems, and the role that clinical geneticists can play varies from condition to condition. The following are some of the clinical geneticist's key functions (ABMG, 2008b):

[1] ACGME is the Accreditation Council for Graduate Medical Education.

- diagnose a wide range of genetic disorders;
- elicit and interpret individual and family histories;
- integrate clinical and genetic information and understand the uses, limitations, interpretation, and significance of specialized laboratory and clinical procedures;
- perform risk assessment;
- interview patients or families to gather the information necessary to reach appropriate conclusions;
- help families and individuals recognize and cope with their emotional and psychological needs;
- recognize situations requiring psychiatric referral;
- transmit pertinent information in a way that is comprehensible; and
- provide appropriate referral or support.

The American Board of Medical Genetics (ABMG) is responsible for certifying physicians and accrediting training programs. There are 1,253 ABMG-certified clinical geneticists in the United States, which is about 0.18 percent of physicians (ABMG, 2008c). The 1,100 clinical geneticists who are currently practicing spend about 45 percent of their time seeing patients, which translates to 495 full-time equivalent physicians.[2] Extrapolating from the work of the Royal College of Physicians (2004), given the size of the U.S. population, the ideal number of full-time equivalent physician geneticists would be 1,200, more than twice the current number.

As genomic technologies expand, this shortage will be exacerbated. An illustration of this looming scarcity can be found in the case of metabolic specialists. There are only about 200 metabolic physicians in the United States, of whom 75 percent describe their practices as "nearly full" and 20 percent expect to retire in the next 5 years (Cooksey et al., 2006). At a time when states want to expand their newborn screening panels to include newer tests, some are unable to do so owing to the lack of metabolic geneticists.

Clearly there are some challenges in the physician geneticist profession. There is a serious mismatch between the expansion of knowledge and the workforce size. The current workforce is not expected to meet patient care needs in the next 5 to 15 years, and young physicians are not entering the field. The American College of Medical Geneticists hosted two conferences (called the Banbury Conferences), one in 2004 and one in 2006, to discuss these challenges and how the field can position itself for the future. The Banbury Conferences developed principles and recommendations for the profession, including the following (Korf et al., 2005, 2008):

[2] J. Benkendorf. Personal communication. July 15, 2008.

- Medical geneticists should work with a team of health care professionals.
- Medical geneticists should provide leadership in the responsible introduction of new technologies, their integration into medical care, and the monitoring of outcomes.
- The medical genetics workforce must be increased to meet current and future needs.
- Training and continuing education programs should include substantial exposure to molecular and population genetics, epidemiology, and bioinformatics.
- The pool of trainees entering the field must be increased and broadened, and training pathways and the certification process must be aligned with this goal.
- Training must be realigned to reflect emphasis on common traits and genetic health care over the lifespan.

Genetics Nurses

Genetics nurses may perform one or more of the following duties, depending on their specialties:

- obtain a detailed family history and prepare a pedigree,[3]
- assess and analyze disease risk factors,
- identify potential genetic conditions or genetic predispositions to disease,
- provide genetic information and psychosocial support to individuals and families,
- provide nursing care for patients and families at risk for or affected by diseases with a genetic component,
- provide genetic counseling, and
- facilitate genetic testing and interpret genetic test results and laboratory reports.

Nurses with a bachelor's degree from an accredited nursing program can become certified genetics clinical nurses by submitting a portfolio to the Genetic Nursing Credentialing Commission (a subsidiary of the International Society of Nurses in Genetics [ISONG]) with 5 years of genetic nursing practice, a log of 50 genetics cases, four written case studies, 45 hours of genetic content in academic courses or continuing education, and evidence of patient, family, or client teaching. To become certified as an

[3] A pedigree is a diagram of family relationships that uses symbols to represent people and lines to represent genetic relationships.

advanced practice nurse in genetics, one must also have a master's-level degree in nursing, 300 hours of genetic practicum experience, and 50 hours of genetic content in academic courses or continuing education in the past 5 years.

Currently, there are 10 genetics clinical nurses and 28 advanced practice nurses in genetics. However, this number is misleading because many nurses work with genetic information but have chosen not to pursue certification. ISONG, which is open to "any registered nurse who has an interest in genetics," has 300 members—a number that probably reflects more accurately how many nurses are delivering genetic health services. ISONG has launched an initiative designed to prepare the entire nursing workforce—not just those nurses who specialize in genetics—to deliver competent genetic health care. It has sought to define and implement nursing competencies and curriculum guidelines for genetics and to survey baseline nursing knowledge, attitudes, and competencies in order to address some of the challenges of recruiting, training, and supporting genetically competent nurses.

DISCUSSION

Wylie Burke, M.D., Ph.D., Moderator

One participant asked the panel if any data were available about how many patients actually want genetic testing and are willing and able to receive the results. Sharon Terry reported that the Genetic Alliance surveyed about 6,000 individuals with genetic conditions and found that overall patient satisfaction with information and services provided by physicians or other genetic service providers was low. Patients reported that they obtained better information from websites and support groups than from their providers. Lochner Doyle said that the National Survey of Children with Special Health Care Needs found that 19 percent of those who wanted genetic services were unable to get them, usually owing to interrupted or no health insurance (HHS, 2004).

Catherine DesRoches noted that the general public has little knowledge of genetic testing, and the levels of uptake of testing are even lower. She suggested that this is due to privacy issues. Shields pointed out that although the data she presented show low uptake of BRCA testing among minority populations, it is unclear whether this is due to patient willingness or interest or to differential provider patterns in offering the test.

One participant asked the panel about the readiness and willingness of service providers—nurses, counselors, MD geneticists—to step out of the box of genetic disease medicine and toward the future of using genetic information for common diseases in everyday practice. Wicklund responded that genetic counselors were ready and able to move into these newer areas because they are already adept at dealing with issues of ambiguity and risk communication. She noted, however, that there is still a question of how many providers will be needed to fulfill the demand in this new era of genomics.

Bruce Korf, of the American College of Medical Genetics, noted that there are two sea changes happening in the field of medical genetics. One is the ability to offer interventions and treatments that were not previously possible. The second is the transition from genetic disease to genetic predisposition toward common disorders. The medical geneticist community, despite a perception that its members are interested only in rare disorders, congenital anomalies, and biochemical genetic disorders, has been thinking hard about how to educate and train current and future practitioners. The ACMG has created a genomic-era curriculum for medical genetics that begins in medical school, continues on through genetics training, and then through continuing education.

One participant relayed a story about ordering an item on Amazon.com, emphasizing how easily the website tailors its offerings to her specific profile and history. She questioned whether genetic testing would ever reach this level of personalization or whether providers were too reluctant to move away from a one-size-fits-all paradigm. Lochner Doyle said that on the basis of her experience in Washington State, patients were moving ahead with genetic testing, with or without their providers, and the health care community needs to begin thinking about how to move away from the traditional model as these market shifts occur. Shields concurred, adding that providers will have to start pooling resources, developing new referral patterns, and building capacity. She mentioned that half of all primary care physicians in the United States are in solo or partnered practices and argued that a continuing education course in "Genetics 101" on a CD-ROM would not be sufficient to enable these providers to incorporate novel applications into their practice. Wicklund commented that genetic counselors are undergoing a culture shift and will have to challenge themselves not to discount something immediately because it is not traditional genetic counseling. She noted that some genetic counselors are upset about the advent of direct-to-consumer marketing, and she remarked that whether counselors approve of this or not, it is a reality, and they will have to adapt in order to meet these challenges.

One participant mentioned that the "elephant in the room" is the economic model behind this shift toward genomic medicine. There is a low

level of reimbursement for genetic testing, and there is a shrinking or static workforce with genetics training programs not filled to capacity. He asked how these advances will be sustainable economically. What will motivate young medical students to enter the field if there is not a realistic professional reimbursement structure in place? Korf said that this question may reveal a fundamental structural problem in the U.S. health care system. If medical students were simply to follow the economics, they would all become cosmetic dermatologists. He argued that to take advantage of the new opportunities that genomics presents, the health care reimbursement system will have to be restructured in order to align incentives with the value of the medical care provided. Money is plentiful in medicine; however, it is not distributed in ways that make sense in terms of prevention and maintenance of health.

Finally, one participant inquired about the level of genetic knowledge provided for "everyday practitioners" in medical school, osteopathic school, or nursing school. Lochner Doyle reported that the literature shows that not only do practitioners feel ill equipped or uncomfortable in their own delivery of genetic services (Gennaccaro et al., 2005; Giardiello, 1997; Menasha et al., 2000), they also may not have an interest in gaining the necessary knowledge. Rather, they want information "on demand"—they want to be able to call a number or check a website in order to get information about a specific genetic condition or test result as needed.[4]

[4] D. Lochner Doyle. Personal communication. February 19, 2009.

3

New Models for Service Delivery

INFORMED MEDICAL DECISIONS, INC.

Heather Shappell, M.S., C.G.C.

The Institute of Medicine report *Assessing Genetic Risks: Implications for Health and Social Policy* (IOM, 1994, p. 148) states:

> As genetic testing expands with the growth of new genetic tests, genetic counseling and education will need to adapt to new modes and settings for the delivery of genetics services, without sacrificing quality.

Unfortunately for patients, health care professionals, and payers in need of skilled genetic health care providers, these same issues are being discussed today, without much change. The scientific community makes great strides, bringing hundreds of new genetic tests to market each year. The rate of discovery, commercialization, and marketing of these tests is accelerating every day. Yet the health care delivery system has not always kept pace with incorporating these new innovations into everyday practice.

A group of former surgeons general recently published a national call to action on cancer prevention and survivorship. The report states, "A shortage of board-certified genetic counselors has made access to reliable information challenging for patients and health care professionals" (Cabe and Springer, 2008, p. 9). The report goes on to offer the telephone genetic counseling service model of Informed Medical Decisions (INFORMED) as a potential solution to this problem.

The idea for INFORMED grew out of Shappell's observations as a genetic counselor responding to telephone and e-mail questions from both patients and health care providers. Questions posed by both patients and providers, before and after they had received test results, clearly demonstrated the dangerous misinformation and lack of information about genetic tests. Attempts to find local genetic experts and refer patients to them were often complicated because of the lack of such experts in many areas of the country. In too many instances, the closest genetic counselor or expert was hundreds of miles away. Traditional genetic counseling services, when locally available, were often seen as inconvenient in terms of wait times for appointments as well as the time commitment required by patients (most centers required at least two in-person appointments) to access the services.

Furthermore, attempts to locate local Spanish-speaking experts were nearly always futile. Shappell observed that many physicians were unwilling to refer their patients to a genetic counselor, even when they felt genetic counseling was important, because the counselor practiced at an organization that was viewed as a competitor. Direct-to-physician and direct-to-consumer marketing of tests was increasing, and providers and patients were being directed away from appropriately trained and unbiased experts. In addition to these patient-centered challenges, insurers faced difficulties as well. They recognized the importance of genetic counselors and preferred to pay for pre- and posttest counseling rather than paying for indiscriminate testing. However, many insurers did not realize that genetic counselors could not bill directly for their services.

Acting on lessons learned, INFORMED developed a telephone and Internet protocol for cancer genetic counseling service delivery that focuses on access to high-quality unbiased genetic counseling services that are in accordance with national guidelines. Counseling is required before testing can be considered, and the primary care physician is engaged throughout the process. INFORMED worked with a health literacy expert from the Centers for Disease Control and Prevention (CDC) to develop web content and patient materials, with the goal of presenting the necessary information in a way that patients could readily understand. INFORMED provides genetic counseling in both English and Spanish 7 days a week and has a capacity of 5,000 new genetic consultations per year with a network of contracted counselors that allows this number to more than double on demand.

When a patient first visits the INFORMED website, he or she completes an online risk screening tool, which assesses whether he or she would benefit from genetic counseling. Next, the patient fills out a personal and family medical history questionnaire and makes an appointment to speak with a

genetic counselor by telephone. Anyone who does not have access to the Internet can use the telephone to provide his or her family medical history and schedule an appointment. During the appointment, patient and counselor discuss family history, cancer risk, and testing options, and the patient decides whether or not to go forward with a genetic test. INFORMED charges for this initial counseling session only to ensure that the goal of the call is to make an informed decision, rather than to encourage testing.

If a patient decides to obtain a test, INFORMED sends a precompleted test kit to the patient and contacts the personal physician to ask him or her to act as the ordering physician. After test results are received by INFORMED, a result disclosure follow-up call is scheduled with the patient in which the genetic counselor talks about results, risk, prevention, and screening options to discuss with the patient's physician, and what the results mean for members of a patient's family. Finally, a thorough personal genetic counseling summary report is sent to the patient and the physician. INFORMED's genetic counselors are available at no additional cost at any point during this process to answer questions from the patient or the provider.

In late 2007, after an initial pilot program, Aetna decided to cover INFORMED's telephone genetic counseling services for all 16 million Aetna members nationwide. In addition to this direct partnership, INFORMED collaborates in other ways with payers and physicians, such as helping payers assess which genetic tests have proven clinical utility, creating reimbursement or coverage criteria for those tests, and providing individual case consultation as needed, for example, when a patient appeals a denial for genetic testing.

The innovative service delivery model that INFORMED has created has worked to integrate providers, patients, and payers into one efficient delivery system. As genomic innovations progress, systems such as these may serve as an example of how to overcome many barriers to traditional genetic counseling and provide effective, time- and cost-efficient services to patients.

NAVIGENICS

Elissa Levin, M.S., C.G.C.

The field of web-based genetic testing has expanded rapidly in recent years, offering everything from traditional diagnostics to dating matches

based on DNA. People seek genetic testing online for many reasons, including the following:

- In-person genetic testing requires making an appointment, often months in advance, traveling to a clinic, taking time off from work, and perhaps finding child care.
- Provider awareness of genetic testing options is not always optimal.
- A patient may see value in a test while a provider does not.
- Web-based testing can be more cost-effective.
- Some patients have privacy and confidentiality concerns that drive them to seek testing online.
- Patients—fueled by media attention, information on the Internet, or their own investigations—may have a proactive interest in learning about genetic contributions to disease and be unwilling to wait until new tests have been integrated into clinical practice.

Web-based genetic testing varies both in types of testing offered and in the model of service used. Types of testing range from diagnostic (to confirm or rule out monogenic disorders and specific genetic conditions), to predisposition (the risk of developing diseases with a genetic basis before signs or symptoms occur), to recreational (e.g., ancestry, nutrigenomics, dating). Models of service range from direct access testing that has no physician ordering, no context given for results, and no professional support, to a "virtual" clinic, which includes physician involvement, genetic counseling, and ongoing support and education.

Patients and providers need to know many things about the companies that offer genetic testing directly to consumers, including

- the type of genetic testing offered,
- whether the laboratory used has CLIA (Clinical Laboratory Improvement Amendments) certification,
- whether educational information is provided,
- the level of services provided,
- whether genetics professionals are involved in providing the services,
- if interpretation of results is provided and whether this is clear and consumer friendly,
- if the company engages one's physician,
- if costs are clearly stated or whether there are there additional hidden costs, and
- if company privacy policy and standards are available.

Three examples of these new models of web-based testing companies are Kimball Genetics, DNA Direct, and Navigenics. Kimball Genetics is a

consumer-driven testing service. Patients can go to Kimball's website and order a test directly from the company, but they are required to provide physician contact information. A test kit is sent to the patient, who sends it back to Kimball's CLIA-certified laboratory for processing. Results, including interpretive reports, recommendations, and educational information, are sent to the patient's physician. Genetic counselors are available by telephone to patients, families, and providers.

The mission of DNA Direct is to create a virtual setting that mirrors traditional clinical practice. The company uses questionnaires, pretest education, and genetic counseling to address patient concerns about inheritance patterns, risk factors, the limits of testing, the testing process, insurance coverage, and family history. DNA Direct has condition-specific protocols—for example, BRCA testing requires in-depth, pretest genetic counseling services. Once a patient chooses to pursue a genetic test and the results are available, a web-based customized report is provided that includes family and medical history information, a lab report, and letters to the physician and the family. This report helps the patient build an action plan for future steps.

The web-based report is unique in allowing the company to track what information interests its patients most. Patients can follow many different links after seeing their results, from medical guidelines to support resources. By observing which links are used and in what order use occurs, DNA Direct can adapt materials to meet consumer demand. DNA Direct reports that of its clients who took a test, 34 percent test tested positively for the mutation or condition of interest. In contrast, rates in a large reference laboratory are approximately 7 to 10 percent (Phelan et al., 2008).

According to Levin, Navigenics was founded by a human geneticist and a clinical oncologist in April 2008, with the goal to "improve individuals' health across the population by educating, empowering, and motivating people to take action to prevent the onset of disease or lessen its impact." The Navigenics Health Compass uses SNP chips (single nucleotide polymorphism microarrays) to determine genetic predisposition for a variety of common diseases and provides information on how patients can delay or prevent the onset of those diseases.

Membership in the Navigenics Health Compass is $2,500 initially, with a $250 resubscription fee each year thereafter. Members provide Navigenics with a saliva sample that is sent to a CLIA-certified lab for a genome-wide scan that captures data on 1.8 million genetic markers. The results of this scan are uploaded into a private web portal that members can access at any time. This online access details, among other things,

- the patient's risk and the average risk for a variety of diseases,
- in-depth condition reports,

- preventive measures that can be taken, and
- guidance on how to discuss results with one's primary physician.

In addition, members have access to board-certified genetic counselors with specific training in risk assessment for common, multifactorial health conditions. As new research emerges, Navigenics evaluates evidence about the clinical validity and utility of genetic tests and updates member reports with the new information.

To be included in the Navigenics Health Compass risk estimates, each SNP–disease association must meet a series of scientific and clinical criteria:

- The association must be replicated and published in top-tier journals. Studies must use a reasonable sample size to detect weak effects.
- There must be a statistically significant result after correction for multiple testing.
- Evidence of the association must come from a well-designed study with sound laboratory practices.
- Clinically, a condition must affect more than 1 in 1,000 Americans.
- The condition must be clinically relevant and actionable, meaning that early screening, lifestyle, or medication can make a difference; it must affect multiple organ systems; or it must affect other diseases.
- Finally, the risk information must be clinically and socially responsible. For example, Navigenics will not test for IQ, athletic propensity, or HIV susceptibility.

More than 95 percent of studies fail to meet Navigenics criteria, and Navigenics currently tests for 23 conditions (see Box 3-1). Members can opt out of learning about specific conditions.

According to Levin, Navigenics research has shown that its members are capable of handling the complex predictive nature of the information provided. They understand the concept of risk factors and how environmental, behavioral, and genetic factors all play a role in health status. Many members report feeling empowered and informed. In addition, 46 percent of early testers took actions such as lifestyle modification, screening, or treatment interventions in response to their test results. More research is needed, however, to understand the long-term effect of genomic risk information on health behaviors. Navigenics is in the early stages of collaborating with several academic institutions on research studies designed to address this issue.

BOX 3-1
Conditions Covered by Navigenics Genetic Testing in 2008

- Abdominal aneurysm
- Alzheimer's disease
- Atrial fibrillation
- Brain aneurysm
- Breast cancer
- Celiac disease
- Colon cancer
- Crohn's disease
- Diabetes, type 2
- Glaucoma
- Graves' disease
- Heart attack
- Lung cancer
- Lupus
- Macular degeneration
- Multiple sclerosis
- Obesity
- Osteoarthritis
- Prostate cancer
- Psoriasis
- Restless legs syndrome
- Rheumatoid arthritis
- Stomach cancer

According to Levin, companies that are using new and innovative genetic testing models, including Navigenics, Kimball Genetics, DNA Direct, and others, are stretching the bounds of what health care delivery is and what it could be in the future. Whether the models require the participation of a primary physician, market directly to consumers, or blend aspects of new and traditional service delivery, these companies are attempting to address some of the challenges of conventional genetic services and are attempting to shorten the gap between research findings and clinical use and to give consumers more autonomy in managing their health care and well-being.

CINCINNATI CHILDREN'S HOSPITAL MEDICAL CENTER

Cynthia Prows, M.S.N., R.N.

The translation of genomics into clinical care will greatly influence diagnosis and treatment of complex pediatric disorders. Genomic discoveries can help define molecular and cellular mechanisms of disease, can be the basis for new therapies, can identify predispositions to childhood disorders, and can identify susceptibility to unintended responses to available therapies. At Cincinnati Children's Hospital Medical Center, researchers and clinicians are collaborating on two projects aimed at bringing genetics

into the clinical setting: the Genetics Pharmacology Service and a nurse education program.

Cincinnati Children's Hospital focused on pharmacogenetics for several reasons: The influence of genes on drug metabolism has been studied for more than 50 years; medications influenced by CYP2D6, CYP2C19, CYP2C9, or TPMT variants are used in pediatric settings; clinical tests exist for these genes; and clinicians in specialty practices expressed interest in using pharmacogenetics. The Genetics Pharmacology Service received internal start-up funding in 2003, and the service became available in July 2004. The primary developers of the service, Richard Wenstrup, M.D., Tracy Glauser, M.D., and Alexander Vinks, Pharm.D., Ph.D., promoted the full integration of genetic testing and pharmacology by making sure that specialty clinicians were involved in the process and by using experts to develop intuitive ordering systems and report templates.

The goal of the pharmacogenetics project was to improve patient safety by giving providers the tools they need for using genetic information appropriately to prescribe and dose medications. Thirty-six drugs were identified for which there was clear evidence that polymorphisms in CYP2D6, CYP2C19, CYP2C9, or TPMT resulted in variable metabolism. One of the key features of the system is that providers order pharmacogenetic tests by drug, rather than by gene, so there is no need for providers to memorize or keep up-to-date on gene–drug associations.

The developers of the program strove to make the ordering system intuitive, efficient, and simple. In the current system, when a provider chooses to prescribe a drug for a patient, he or she uses a computer ordering system with fields for dose, route, frequency, and other notes. When the provider prescribes one of the 36 selected drugs, an automatic pop-up screen asks the provider if he or she wants to order a pharmacogenetic test that can help predict how the child would metabolize the drug. The provider is required to answer "yes" or "no" before moving on to the next screen. If a patient has already had a genetic test done for the gene in question (but for a different drug), the provider is asked if he or she wants to order an interpretation of the genetic results for the new drug. If a provider is prescribing a drug for which a test has already been completed, the provider can choose to view the results.

Once a test is ordered, results are available within 2 business days. The lab operates days and evenings, Monday through Saturday. Reports include the type of test performed, the predicted phenotype, general dosing recommendations, key enzyme inhibitors and inducers, critical drug–drug interactions, test limitations, and the location of supplemental information. The actual genotype was not originally included in the report because of concerns about insurance discrimination.

Over time, the service has been altered in response to feedback from

providers. Psychiatrists were concerned about ordering a new test each time they shifted from one medication to another while searching for an effective treatment. Therefore, a panel was provided that tested and interpreted both CYP2D6 and CYP2C19. Pediatric gastroenterologists and oncologists at Cincinnati Children's Hospital were already accustomed to ordering TPMT; they wanted to continue ordering by gene and wanted the genotype included on the report. Adult oncologists in the local community requested tamoxifen pharmacogenetic testing, and local cardiologists and thoracic surgeons were interested in using CYP2C9 and VKORC1 tests for warfarin dosing.

Another major component of Cincinnati Children's Hospital's genetics program is the education of nurses and other providers. Before implementing the Genetics Pharmacology Service, providers were educated about how the program would work and how it could assist in delivering patient care. Education was provided through in-service training programs and at faculty–staff meetings, pediatric grand rounds, resident conferences, and unit-based nurse educators meetings. Patient and parent fact sheets were posted online, and education binders were provided for every patient care unit. In addition, Cincinnati Children's Hospital has a Genetics Education Program for Nurses (GEPN) that offers online genetics educational opportunities and resources. There are moderated classes as well as self-paced modules on topics ranging from ethics to environmental genetics. The 18-week Genetics Institute course has been offered since 2002, and 193 nursing faculty and advanced practice nurses have completed the course. There have been 1,189 individuals who have completed one or more training modules, including 196 in the pharmacogenetics module.

DISCUSSION

Wylie Burke, M.D., Ph.D., Moderator

One participant asked presenters to clarify how their organizations define "validated" associations. He noted that, for example, testing for CYP450 for SSRIs (selective serotonin uptake inhibitors) was not recommended by the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) because there are no data on clinical validity and utility or on patient outcomes. He expressed concern that in this confusing, rapidly moving environment, new ways of implementing service delivery may be risky. Shappell responded, stating that INFORMED does not sell genetic testing; it provides access to genetic counseling. When stating that INFORMED

offers only tests for which there is clinically useful, actionable information, this means tests that have national guidelines recommending testing in certain clinical situations. Because INFORMED works directly with payers to help them identify clinically useful tests, counselors talk to patients only about tests that have already been approved by their health insurance plans. If a test that is clinically useful according to national guidelines is warranted and the insurer has not yet created a testing coverage policy, INFORMED works with the health insurer to assist in developing an appropriate policy for coverage.

Prows concurred with the questioner that there are no large, randomized controlled trials that prove the clinical utility of pharmacogenetic testing for psychiatry. She went on to point out that because drugs prescribed in children's psychiatry are only about 50 percent effective, providers want and need as much information as they can get when they are choosing medications.

Another participant directed a two-pronged question to Levin. First, it appears that physicians doubt the clinical value of many of the tests being offered by direct-to-consumer companies. Might this be a sign that the testing is not ready to be put into patients' hands? Second, a virtual clinic requires a virtual patient. How does Navigenics know who its patients are and where the samples are coming from? Levin responded that the physicians who work with Navigenics and are using the service tend to be early adopters who are interested in maximizing the number of tools that they have to tailor health care. Although it is perhaps too early to think about genetic testing being incorporated into a primary care context, there is motivation in the health care community to use genetic tests from those who are actively engaged in thinking about the clinical utility of genetics. Concerning the question of who the patients are, Levin stated that Navigenics does not test minors and that a consent form is required to proceed with testing. It is recognized that some clients use pseudonyms and that it is not always possible to know the actual identity of the individuals tested.

Another participant asked Prows about the voluntary nature of the pharmacogenetic testing program. If the testing is beneficial, why not require that physicians use it rather than making it optional? Prows responded that providers need to feel that the testing will assist them in their decision making; some people rapidly adopt new technologies, whereas others are more reticent and should not be forced to use them. Another participant said that because genetic testing is not yet the standard of care, a physician still needs to have the option of deciding whether or not to use it. It is assumed that when genetic testing does become the standard of care, it will no longer be optional, the participant continued, noting that many new technologies over the years have gone through these phases from optional to standard.

A final questioner asked if Cincinnati Children's Hospital had gathered data to indicate whether the program is having an effect on clinical utility. Prows clarified that the program was designed as a clinical service, not as a research program, so plans were not put in place to collect outcomes data. As such, clinicians did not consistently document in the charts how pharmacogenetic testing affected their clinical decision making, hindering attempts to assess the impact of the tests by implementing a retrospective research study. Additionally, clinical services do not systematically collect outcome data. Prows noted that Cincinnati Children's Hospital is currently developing different study designs (e.g., registry) to address the clinical utility of pharmacogenetic testing.

4

Vision of the Future

Wylie Burke, M.D., Ph.D.
University of Washington

Extraordinary promises about the future of genomics in health include individualized health care based on testing for inherited risk, improved clinical management based on molecular characterization of disease, and new therapeutics. Although genomic technology is still nascent, there are already compelling examples of technologies that fulfill these promises. It is becoming standard practice to test for mutations in the BRCA or MLH1 and MSH2 genes to identify genetic susceptibility to breast cancer and colon cancer or to test for genetically based hypersensitivity to certain drugs (e.g., HLA-B*5701 and abacovir). Gene expression profiling of tumor tissue has given providers new information to manage disease and make treatment decisions. New therapeutics (e.g., fomiversen, imatinib, trastuzumab) have also been developed based on a genetic or molecular understanding of disease.

Despite these important advances in genomic technology that have affected health care and health outcomes, many uncertainties have yet to be fully addressed. How does one match the potential promise of genomics with particular health conditions? What strategy will yield the greatest benefit for a given condition? What is the scope of harm from the application of genomic technologies? How many good ideas will fail during the development process? What will be the effect on the costs of health care, and how will genomic technologies strain the system? Strains to the system are already apparent. There is a tremendous need for rapid assessment of emerging technology, but it is difficult to gather adequate study populations, secure funding, and determine the most appropriate way in which to assess comparative effectiveness. There is lack of trustworthy processes

that use rigorous and transparent methodology to create guidelines and produce the educational tools to ensure their implementation. Access to genomic technologies is unequally distributed by factors such as geography, socioeconomic status, race, and education level.

Genomic screening for colorectal cancer is one possible application of technology that could serve as an example of the potential promises and pitfalls of genomics. Colorectal cancer is the fourth most common cancer in the United States (NLM, 2009), and family history has long been known as a risk factor. Having a first-degree relative with a history of colon cancer almost doubles an individual's risk, and it shifts the risk to an earlier age. For these reasons, colorectal cancer screening is recommended at age 40 for people with a family history, instead of the generally recommended age of 50.

The assessment of family history is not as clear-cut as it may seem, however. There is a continuum of family history. A history of colorectal cancer in the family could be anything from a grandfather diagnosed at age 80, to a grandmother diagnosed at age 65 and a mother with a polyp at age 52, to colorectal cancer at a young age in every generation. Some family histories indicate an increased risk of colorectal cancer, and some do not. About 7 to 10 percent of the population has a family history indicating a moderately increased risk of colorectal cancer, while fewer than 1 percent have "high-risk" family histories (such as those with early-onset cancer occurring sequentially in multiple generations) indicating the presence of rare genetic conditions such as Lynch syndrome and familial adenomatous polyposis (FAP). Assessing family history information can, therefore, be time-consuming. Compounding the difficulty is that family histories are often imperfect because people may not know or may fail to recall the medical problems of relatives. Another potential problem is the case of the "vanishing family history" (Figure 4-1). Thanks to improved screening and treatment, instead of a diagnosis of colorectal cancer at the age of 60, a family member may have a polyp in his or her 50s, get screened and treated early, and avert cancer. Other family members may never know that the individual had an adenomatous polyp (a non-cancerous growth that can progress to cancer); the risk information previously provided by the family history of colorectal cancer is, for positive reasons, no longer available.

There are several ways to address the problems involved in relying on the collection of family history to estimate an individual's risk of colorectal cancer. Information technology could be used to obtain self-administered data, process them, interpret them, and deliver them to the patient and the provider. Electronic medical records from multiple family members could be integrated to provide a more accurate and thorough picture of family history. Alternatively, one might ultimately be able to do away with family history collection and rely solely on genetic testing.

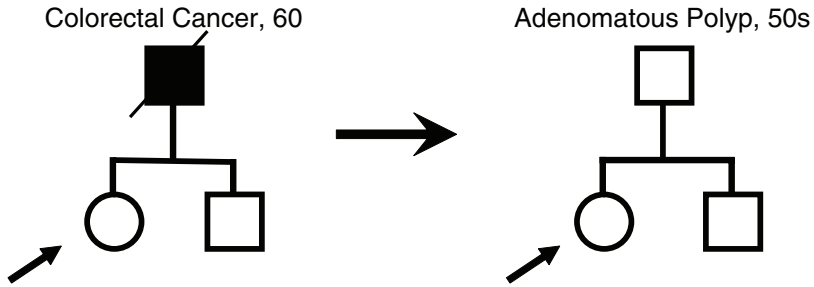

FIGURE 4-1 Vanishing family history of colorectal cancer.
NOTE: Top boxes represent father; circles and squares beneath represent female and male children, respectively. Slash through box indicates that father is deceased.
SOURCE: Burke, 2008.

As genome-wide association studies identify all of the genetic contributors to colon cancer risk, including common gene variants with modest effects on risk, it will likely be possible to create a genomic test to identify the 7 to 10 percent of people with moderately increased risk, rather than relying on traditional assessment of family history. A test of this kind could have several benefits: It wouldn't depend on imperfect recall or knowledge of family history, it might result in reduced misclassification, the blood test could be less costly than spending the time to collect family history, and screening of patients could potentially be tailored better than with family history if gene variants in the test panel were associated with polyp dwell time (the time a non-cancerous growth takes to evolve into cancer) or age of onset.

There are also pitfalls to using genetic testing. If a provider is performing a genomic test to identify people at moderately increased risk, it is likely that he or she would also want to test for highly penetrant single-gene diseases that are also associated with colorectal cancer, such as FAP or Lynch syndrome, and this could raise problems. FAP has a frequency of about 1 in 32,000 in the population and results in a nearly 100 percent lifetime risk of developing colorectal cancer. If it is assumed that the test for FAP performs very well, with a sensitivity, specificity, and negative predictive value of 99.9 percent, .001 percent of those tested will get a false positive result simply because the low prevalence of the condition affects the positive predictive value of the test. In other words, 32 individuals out of every 32,000 tested would be told that they have an FAP mutation when they do not, resulting in further expensive, time-consuming, and uncomfortable testing and medical procedures. If a genomic screen for colorectal cancer

of this type were vastly expanded and offered as a standard of care to all approximately 300 million people in United States, there would be 300,000 false positives for FAP and 9,365 people correctly diagnosed with FAP. This may be an acceptable ratio, but there might be better ways to identify high-risk individuals, such as careful family-based detection after a diagnosis is made. This example indicates that genomic profiling for risk of colorectal cancer is a complex issue, and the benefits and drawbacks must be weighed and considered. In the process, emerging, potentially disruptive technologies would also have to be considered: effective virtual colonoscopy or therapy to inhibit polyp formation could change the risk–benefit analysis for genomic risk profiling.

In addition to the advances that create the possibility of genomic profiling for disease risk, there are rapidly expanding opportunities for disease characterization and new therapeutics. Important developments have occurred in gene expression profiling of tumor tissue, and there is strong reason to believe that the development of proteomics and metabolomics will expand the opportunities to use molecular tools for disease characterization (Gowda et al., 2008; Latterich et al., 2008).

Recent genome-wide association studies have exponentially increased the ability to identify genes associated with disease and biological pathways involved in disease processes. This new information is likely to lead to insights into potential therapeutics and drug targets that are necessary for drug development.

As these rapid advancements in scientific knowledge arise, it is crucial to ask the question, how much health improvement and at what cost? There is an urgent need for rapid assessment of these emerging technologies in order to obtain comparative effectiveness data as well as to study questions about the cost, access, and harm that are likely to increase with research success.

One particular harm that is likely to accompany advances in genomic technologies is the "cascade effect." The cascade effect, as described by Deyo (2002), occurs when an unnecessary test is performed or a false positive result is obtained from a test that was not clinically necessary. Further testing and treatment follow, and avoidable adverse effects—or even morbidity or mortality resulting from intervention—occur. Currently, the best example of the cascade effect is in the field of radiological imaging. As imaging becomes more sensitive, health care providers are discovering unanticipated incidental findings of uncertain clinical significance. The medical follow-up from these incidental findings may cause greater problems than the problem originally intended to be addressed with imaging. Deyo lists common "triggers" for the cascade effect, several of which are particularly applicable to genomics, including

- shotgun testing, such as large-scale genome profiling or sequencing;
- underestimation of the likelihood of false positives;
- screening inappropriately simply because tests are available;
- errors in interpretation of highly complex information;
- patient demand—that is, as potential benefits of genomic tests are publicized, patient demand will be stimulated; and
- low tolerance of ambiguity by patients and providers, which leads to further testing.

Substantial harms may potentially result from an expansion of genetic risk assessment, whether in the form of testing for inherited risk or characterizing a disease process. For example, there is the potential for unwanted information, that is, information that will not help solve the problem at hand and may trigger the cascade effect. In addition to false positive results, there is the possibility of finding variants of unknown clinical significance. In BRCA testing, for example, approximately 6 percent of women of European descent with a mutation will have a variant of unknown significance, and a greater percentage of positive results in minority patients will be of unknown clinical significance. Patients are undergoing these tests to answer important questions, and findings of unknown significance do nothing but confuse the situation. Some of the incidental or clinically insignificant findings may not only lead to further testing and treatment but actually lead to downstream adverse effects. For example, while testing as part of a smoking cessation program, a physician may find that a patient has genetic variants associated with an increased likelihood of substance abuse. In the future, that physician may be reluctant to provide the patient with adequate pain treatment when needed.

The advances in genomic technology, along with the potential harms and benefits, have generated an intensified discussion about the scope and coordination of oversight. The Secretary's Advisory Committee on Genetics, Health, and Society (2008) recently released a report on oversight of genetic testing. The direct-to-consumer movement has resulted in new calls for more regulation. There are interesting developments and continued discussion about accreditation and licensure. There is an increased need for high-quality health technology assessment. All of these trends are likely to continue and illustrate the need for careful assessment of emerging genomic tests and technologies.

Eisenberg (1999) wrote, "Technology is rarely inherently good or bad, always or never useful. The challenge is to evaluate when . . . it is effective, for whom it will enhance outcomes, and how it should be implemented or interpreted." Lessons learned from the health technology assessment process described by Eisenberg can be applied to emerging genomic technologies. For example, innovation and flexibility are needed when con-

ducting genomic technology assessment. Furthermore, assessment must not be limited to randomized controlled clinical trials but rather must be an ongoing process that uses a variety of study methods. Assessment must also examine various outcome measures such as quality of life. The process must incorporate information from the real world of health care and avoid redundancy by encouraging sharing of knowledge. In addition, it is critical that those who are assessing genomic technologies engage with both providers and the public.

The methods used in developing guidelines for decision making about when and how genomics enters the health care system must be independent, transparent, and trustworthy. The resulting guidelines and associated educational material need to be made available in direct and simple language that is readily accessible by the public and health care providers.

DISCUSSION

Wylie Burke, M.D., Ph.D., Moderator

One participant asked that Burke envision the year 2020 and describe her idea of the role of a genetics professional. Burke first noted that many of the ideas discussed earlier in the workshop were quite relevant to this question, for instance, delivering testing via the web or directly to consumers. She added that, ideally, this type of delivery system would be linked to high-quality, assessment-driven guidelines. It is also crucial that there be appropriate follow-up using a system that is convenient for patients and providers. Burke referred to a previous statement made by Levin about the amount of time it can take a patient simply to get to a clinic and agreed that receiving genetic results via the web would sometimes be more efficient. She stated that the pharmacogenetics ordering system at Cincinnati Children's Hospital was a very interesting model, and as more and more tests are developed, this kind of assistance will become evermore important for providers. Finally, Burke stated that reimbursement schemes will have to be redesigned to permit genetic counselors to be reimbursed for giving advice to primary care providers. With the small number of geneticists and the increasing amount of genomic technology, there needs to be a systems-level change to allow reimbursement for this type of expert consultation.

Another participant cautioned that one of the main risks of new innovations is premature belief in their effectiveness. The health care community must take a very critical look at what the data tell us before making recommendations about the use of new technologies. Frederick Chen brought up

the example of the full-body CT (computed tomography) scan, noting that it was a technology that was marketable because it made practical sense and had intuitive value to the average consumer. Because clinical utility did not exist, however, neither the medical community nor the insuring community supported the use of this technology. The result was that full-body scans were not nearly as successful as business models suggested they would be. Full-body CT scans are just one example in a long line of failed assumptions in medicine. The medical community, noted Chen, has two options for assessment: Either wait until these new technologies are in practice or collect evidence prior to implementing the new technology.

One participant asked Burke whether genetics is different from other types of highly cognitive areas of practice. Burke responded that at present two things are particularly important about genetics and genomics. One is the dramatic pace of discovery. The other has to do with risk assessment. Although most genetic risks are similar to other kinds of risks (e.g., cholesterol level, blood pressure), there are two ways in which genomic risk assessment may pose particular problems: the breadth of the proposed risk assessment process and the fact that some data suggest that people respond differently to risk assessments delivered by a DNA test versus a family history. A participant added to this discussion by noting that genetics is unique from other forms of medicine because there is an opportunity for interventionary genetic therapies and genetic manipulation, which are new and potentially dangerous uses of scientific knowledge.

One participant stated that the American Health Information Community is attempting to gather and make evidence available by centralizing and standardizing decision support systems. He also mentioned the efforts of Evaluation of Genomic Applications in Practice and Prevention (EGAPP), which looks at and attempts to answer questions such as: What is the level of certainty? How large are the impacts? How do they compare to various alternatives? The participant emphasized that information needs to be presented in a systematic manner in order to help make decisions.

Shields commented that reimbursement policy can be used as a very effective lever to incentivize the provision of genetic services. For example, many years ago in Massachusetts it was extremely difficult to provide health care for the homeless. Once Medicaid allowed for reimbursement of health care provided in a homeless shelter, that situation changed. Data that are being collected on genetic services are generally used for reimbursement rather than to track the clinical effect of genetic medicine on health outcomes. The coding used for reimbursement simply notes that a genetic test was conducted, not the specific type. Shields suggested that advocates, private payers, and the Centers for Medicare and Medicaid Services (CMS) work together to add routine collection of clinical information that could be used for long-term understanding of clinical efficacy.

Naomi Aronson, a representative of Blue Cross/Blue Shield and a member of the Roundtable, responded that payers are also concerned about coding and are open to the idea of building other coding systems with more room for clinical information. Representatives of testing laboratories noted that attempts had been made in the past to use modifier codes, but disagreement between the laboratories and the payers resulted in discontinuing those efforts.

One participant stated that while it is possible to put enough SNPs together to obtain a risk ratio for several diseases (e.g., prostate cancer, type 2 diabetes, breast cancer), that ratio is not enough to establish clinical utility. Unless a result changes the course of prevention or treatment, genetic testing does not provide any new information for either the provider or the patient. Another participant said that the standard provider reaction to a genetic test is to give the patient the same preventive message given to all patients, even when the patient's genes signal that there is something different about his or her biology and that standard preventive measures may not have the same effect for that patient. Another participant added that taking action on very low relative risks could be quite dangerous and that only higher relative risks of 9 or 10 should be used. It is also necessary to ensure that the data are correct. Finally, another participant said that there is money in the system for finding gene variants but little money or other resources for the much more expensive task of determining whether or not these variants have any clinical utility.

In a final comment, one participant noted that despite all of the research and the committees that are developing new data, medical practice ultimately comes down to the physician, who must sort through a variety of information on many factors, filter out that which is not useful, and make decisions for each patient. He said that physicians are generally hesitant about new innovations.

5

Brainstorming on a Service Delivery Model for the Future

Sharon Kardia, Ph.D., Moderator
Panelists: Debra Lochner Doyle, M.S., C.G.C.; Alexandra Shields, Ph.D.; Vivian Ota Wang, Ph.D., F.A.C.M.G., C.G.C.; Catherine Wicklund, M.S., C.G.C.; Frederick Chen, M.D., M.P.H.; Catherine DesRoches, Dr.P.H.; Bruce Korf, M.D., Ph.D.; and Sharon Terry

The focus of the following discussion was to explore what new and future models of genetic service delivery might look like, including who will provide the services, what those providers need to know, how the providers will inform patients, where the services will be delivered, and what, if any, burden might be placed on providers using these new models.

HEALTH CARE DELIVERY SYSTEM

Lochner Doyle began the discussion by saying that there is no one-size-fits-all service delivery system for genetics and genomics. Different models must be tailored and modified over time. Entrepreneurs outside the medical system may help to develop new ways of meeting the needs of the provider and patient markets. Primary care physicians, according to Chen, are undertaking a renovation of the medical home. Elements such as information management, information technology, and the use of electronic health records are driving major change in primary care practice. Reimbursement for primary care, however, has not kept pace with other areas of medicine. Shields concurred that reimbursement must be addressed if the medical community is to realize the potential of genomics. The full continuum of services that are needed to deliver genomic medicine adequately and appropriately must be reimbursed and available for both commercially and publicly insured populations. If medicine has value, society must ensure that there is a floor beneath which citizens cannot fall; there must be funding for public hospitals, community health centers, and providers that serve uninsured patients. Primary care and prevention must be adequately reimbursed;

the promise of genomics in everyday medicine could perhaps be used as a leverage point with which to incentivize the reformation of primary care.

Chen raised the question of how genomics will fit into a system that is already overloaded. There are workforce studies that claim a massive shortage of physicians. In addition to looking at physician-to-population ratios, one must also examine the distribution of physicians across the population. Currently there is a maldistribution of providers, and this is likely to present difficulties for the practice of genomics as well.

Korf used Figure 5-1 to illustrate the stratification of the world of genetics and its place in medicine. At the top of the triangle are rare monogenic conditions, and it is in this space that medical geneticists have historically practiced. These disorders, such as inborn errors of metabolism and congenital anomalies, are relatively rare, single-gene conditions. Despite their rare occurrence, an increasing number of diagnoses are possible, and new interventions are being developed for individuals with these kinds of conditions. Newborn screening, in particular, is having a significant effect on the incorporation of genetics into primary care.

The middle tier, referred to here as common monogenic, represents

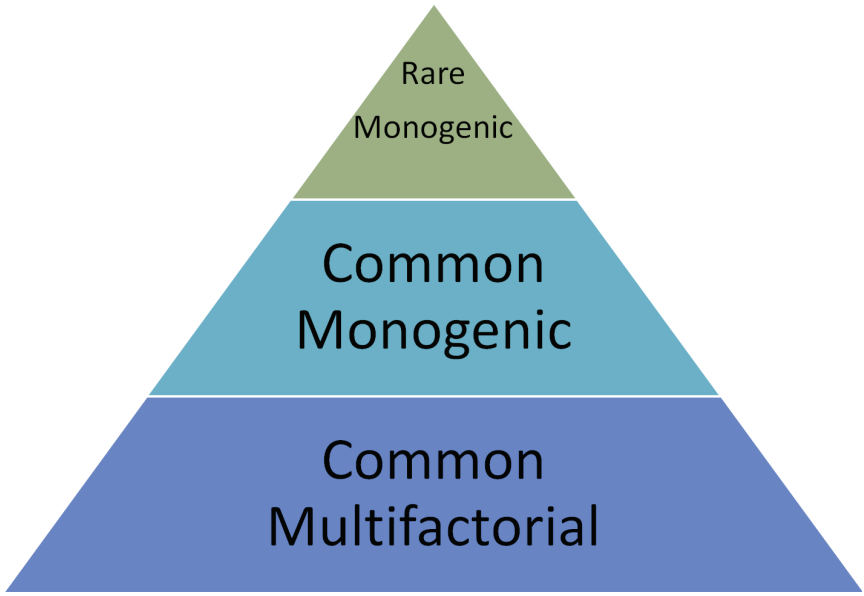

FIGURE 5-1 Stratification of genetics in medicine.
SOURCE: Korf, 2008.

conditions such as breast and ovarian cancer and hereditary nonpolyposis colon cancer, which are not necessarily monogenic but are fairly common with well-established genetic associations. Individuals with these predispositions do not look any different from other patients. The only way to identify that they are at risk is to conduct a family history, which is a lengthy and complex process, or to perform genetic testing, which has its own benefits and pitfalls.

The base of the triangle, continued Korf, is the area in which data are least sound. Given the lack of data and understanding, medical prediction will likely be about as accurate as weather prediction, that is, it will be correct to some degree, but far from exact. Enormous amounts of data and statistical modeling go into a prediction, and stochastic factors can affect the outcome. As data continue to be developed, it will be critical that partnerships within the professional genetics community guide the rest of the medical community through the disruptive technologies and new service models that arise.

Terry pointed out that although there is much discussion about how providers and other sectors of the medical and research community must be integrated, it is crucial that the consumer community also be involved. There are many examples of consumer demand driving rapid change in other industries, such as computer technology innovation over the last 30 years. Similarly, companies such as Amazon and Flickr have used technology to make it possible to connect people, and their physical and digital media, all over the world. Consumers are forming communities online, demanding and receiving information, and determining for themselves what is important or useful to them. The genetics and medical communities should be thinking about how they can help to augment these consumer-driven innovations, rather than fearing or delaying inevitable changes.

Wicklund added that providers of health care and consumers of health care may not have the same definition of an ideal system of service delivery. While both certainly want quality of care, consumers may wish for more flexibility in their care, including the ability to seek the information they want in the form they want. They may wish to take their health care into their own hands.

Wang asked, what if, after exhaustive research on gene associations and gene–environment interactions, it turns out that the message is that patients need to eat better, sleep 8 hours, and exercise? Health communication messages aren't always about complex diseases and treatments; the importance of helping people do very basic, yet difficult, things cannot be overlooked.

Levin noted that research, data collection, and helping providers use new technologies are all important components of moving genomics into health care. However, it is also critical to have an educated patient base,

across socioeconomic groups, that is prepared to receive this information and understand the advantages, disadvantages, and limitations of genetic testing.

HEALTH INFORMATION TECHNOLOGY

DesRoches reported on her recently published study (DesRoches et al., 2008) that attempted to establish a baseline estimate for the number of physicians who are using a fully functional electronic medical record (EMR), one that can integrate family history into a medical record and provide physicians with clinical decision support, e.g., suggestions to order a specific test or offer a specific screening. The baseline number was 4 percent. Even in the largest practices in the nation, only a small minority of physicians use this technology. When asked specifically about clinical decision support functions, 21 percent of physicians reported that they had a computerized system for reminders and guidelines. Only 17 percent of them use it, which translates into about 2 percent of physicians nationwide using this technology. Among the 4 percent of physicians who have a system that prompts them to order a genetic test when appropriate, only 8 percent have ever ordered a genetic test because of the prompt.

DesRoches noted that both Google and Microsoft have offered web-based tools that consumers can use to store their health information. This could potentially save time and energy by allowing providers to simply upload a patient's information into their system to populate a medical record. Currently, two hospitals in the country have this capacity. It is often assumed that genomics can be incorporated into a physician's practice through the use of these types of health information technology. The current low utilization of available technology, however, suggests that it may be a very long time before genomics is fully integrated into everyday practice.

Shields added that when health information technology is put in place, the most critical element needed is a method for tracking the use of genomic medicine and collecting adequate clinical detail to assess its effect on health, both in real time and over time. EMRs can and must be made more clinically rich to be useful in genomic medicine, structured so that they can be used for research, medical practice, and public health. Shields asked the audience to imagine an EMR that includes information such as race, ethnicity, education level, housing situation, diet, levels of stress, and allergies that could both be informative for individual risk assessment and also provide data for future research to find patterns among the EMRs of millions of people.

One workshop participant expressed her opinion that EMRs and personal medical records are a false messiah. She said that EMRs are pro-

grammed for billing, not for patient care, which makes it very difficult to capture the subtleties, nuances, and innovations in medicine that exist in the standard medical record. Similarly, the personal EMR, if it mimics what patients currently bring to the office, is a hodgepodge of previous opinions and test results that are of very little use to the physician. The thing primary care providers and their patients need is more time for contemplation. Unfortunately, under the current billing system, providers are rewarded only for actions that are billable, not for the time it takes to interact with patients and consider the appropriate course of action. From the provider's point of view, when a patient comes into the office, collecting data for an EMR research project is a very low priority. The top priority is helping the patient with his or her health concerns. Asking a physician to collect information about demographics and environment is a waste of the provider's time.

DesRoches responded that the vast majority of physicians agree that these health information technology tools are not very easy to use and that they take more time than they save. This is because the financial incentives for this technology are completely misaligned. The savings that one could garner from using the system accrue to payers, not providers; therefore, there is no incentive for providers to use the system. As providers are inundated with more and more genomic information and innovation, there will have to be a technological solution that helps them filter the information and decide on a course of action. This system must be created to benefit the provider, however, or it will not be used in practice.

One participant observed that residents-in-training are, like their predecessors, extremely intelligent. However, they have lost some skills (e.g., math, spelling, clear handwriting) because new technologies enable them to function without those skills. If a robust decision support system was fully integrated into clinical care, he asked, is it possible that physicians would lose their ability to think and become technicians who simply input information and await the response?

DesRoches responded that the unintended consequences of the use of health information technology are unknown at this point, because use is minimal. As it becomes more widespread, researchers will have to keep a close eye on these types of consequences, which are a serious concern.

Chen added that although it may be true that students and residents no longer know how to listen to heart sounds, physicians also no longer taste and smell urine to arrive at diagnoses. Disruptive technologies are called disruptive for a reason, and they encounter much well-meaning opposition before they are finally adopted. Companies such as Microsoft and Google are capable of grasping the complexity of health care, which some health information technology vendors tend to underestimate.

DATA COLLECTION

Several participants noted that one emerging theme seemed to be how to collect data from clinical settings, not only from claims data, but from creative new systems that do not burden providers.

Korf pointed out that one of the challenges to such a data collection system is developing a systematic method for capturing phenotypic data that can be compared and aggregated on a large scale. Despite the fact that human phenotypes have been studied for millennia, and genotypes only for decades, it is far easier to classify and categorize genotypes.

In response, one participant reported on a study that examined phenotype data (Roses et al., 2005). Experts on 17 diseases contributed their opinions, and the researchers were successful in classifying the phenotypes of diseases within disease categories. However, an enormous amount of money and time was required. The sheer amount of data necessary to classify a patient as having a disease would prohibit large-scale data collection in a clinical setting.

Another participant added that even if it is possible to collect excellent genotypic and phenotypic data, still missing from the equation are the molecular data gleaned from samples of blood, tissue, tumors, etc. There are systems in place, such as those at the Department of Veterans Affairs (VA), that collect data on millions of patients per day, yet neglect to collect samples for molecular profiles. There is a focus on building a system for archiving clinical information, but molecular data must be included if the system is to be robust.

Another participant noted that there is a major disadvantage to collecting data through clinical practice, which is that because medical practice is fairly standardized, there would be no standard "control" group. Rather than answering an experimental question, the data would simply reflect current medical practice. Having all the data may be interesting, but it would not necessarily translate into better clinical care. She added that some of the discussion had focused on how to fit genomics into the current delivery system. It is quite likely, however, that the future system will be vastly different. The cost of sequencing a genome is decreasing rapidly, and at some time in the future everyone may have their genome on "Google Genomics." An individual will be able to look at recent studies of gene associations and use that information in conjunction with his or her health history to make decisions about prevention and care. Perhaps, instead of newborn screening, every baby will leave the hospital with its genome on a CD. While randomized controlled clinical trials will still exist, the way in which data are translated into health will be very different than in our current system.

Another participant argued that although evidence thresholds of when to move from research to practice will vary for different genes and conditions, the fundamental problem is balancing the need for evidence of clinical validity and utility with a rapidly changing world in which consumers are demanding information and industry is continually developing new technologies. Genomic technologies can serve as a wake-up call for the medical system as a whole. The system is already strained under misaligned reimbursements and maldistribution of resources. The rapid growth of consumer demand for genomic technologies may soon break the system completely. The challenge is to collect quality data and establish evidence thresholds that could be used to determine coverage and reimbursement in practice.

However, that participant continued, relying on traditional research methods is not feasible for genomic innovations. A large cohort study on a million people for 20 years would result in data in 40 years and a trillion dollars spent. In the meantime, the medical community would have to live with uncertainty and assess each new technology as it is developed. Innovations that meet a certain standard of evidence can be moved into practice and studied in a postmarketing environment. Large health care providers such as the VA are capable of performing this sort of research and practice along with informed consent. There must be a technology assessment system in place that is robust enough to allow promising technologies into clinical care and then provide data on the clinical utility of these applications.

Another participant asserted that technological innovation in genomics, like every other innovation in medicine, is likely to go charging forward with tremendous waste, major errors, disenchantment, reenchantment, and finally a stable foundation on which to build the future. It will take a long time to translate the knowledge that is gained through research into the wisdom to know what to do with it.

Panelists were asked to list their top priority area for service delivery, and then additional audience participation was encouraged. Chen placed priority on developing the clinical utility of genetic tests. DesRoches favored realigning reimbursements for primary care physicians. Wicklund also placed a priority on reimbursement, emphasizing the need for reimbursement for services other than just procedures. Korf said that his priority was developing a well-educated group of health care providers. Lochner Doyle emphasized the need to address the lack of data. Wang stated that priority should be given to focusing on people. Shields stated that her priority was to obtain better clinical information to evaluate the impact of genetics. Terry favored creation of a coordinated system with participation from all stakeholders.

DISCUSSION

Sharon Kardia, Ph.D., Moderator

One participant stated that perhaps the people sitting around the table are the wrong people to be brainstorming about innovation. The members of the Roundtable are present because they are experts, she said; they have learned and are invested in the current system. True innovation may have to come from a table of outsiders with disruptive minds who can grapple with these issues with unbounded thinking. There are many innovative models in which "outsider" genetic service providers are taking risks and struggling with the issues that have been discussed here. Perhaps the "insider" community should collaborate with these other providers to move innovation along.

Another participant responded that although the idea of collaborating with innovators sounds appealing, there is conflict when innovators are also trying to turn a profit. Innovation can be done creatively, but its evaluation should follow the rules of research and take place in a relatively controlled environment. Innovation without research and assessment could result in more harm than good on a population level. Collaboration between the private sector and the government in setting parameters for innovation would help to ensure that new technologies are beneficial rather than harmful.

A participant reported that at a recent American Medical Association meeting, there was discussion about the idea of a government institute, separate from the National Institutes of Health, that would study the delivery of health care. Examples of what might be achieved by such an institute can be found by examining health care improvements that were made because of battlefield observations. Statisticians who were working on military battlefield health identified and solved problems with innovative health *care,* rather than medical solutions. Eye injuries were a major problem. The statisticians interviewed soldiers and discovered that they were not wearing their goggles because they looked ugly. Eye injuries decreased significantly when new, more attractive goggles were designed. Catheter infections were reduced to nearly zero with the implementation of a checklist for health care providers. The checklist was incorporated into care in the State of Michigan, with similar results (although investigators were castigated for breaking all the institutional review board [IRB] rules).

Rather than attempt to change reimbursement policies, the participant continued, perhaps a "National Institute of Health Care Delivery" could tackle many of the problems in the system today, with physicians, ethicists, statisticians, and others taking a systems approach to health care. Another participant pointed out that three government agencies are charged with

studying and improving health care: the Health Resources and Services Administration (HRSA), the Agency for Healthcare Research and Quality (AHRQ), and the Centers for Disease Control and Prevention (CDC). The agencies have not, however, had great success in meeting that charge.

Another participant observed that much of the discussion has focused on money, that is, on reimbursement policy, laws about Medicaid and Medicare, and money for research. He suggested that universal health care is a way to save money, keep people moving out of the intensive care unit and toward preventive care, and increase the amount of time and energy that could be focused on making health care better. Additionally, primary care physicians will be entering the field of genomics whether they want to or not. Basic genomics education of these providers is crucial, and when providers begin to use the new technologies it is essential that they be given information that will help them understand the genetic test and the results.

One participant said that newborn screening is by far the most widely used example of genetic testing. Nearly every baby is screened for 30 different diseases, regardless of the parents' ability to pay. Although this testing is different from the genomic tests that have been discussed at this workshop, there are lessons to be learned. For example, when a baby is diagnosed with a rare metabolic disease, the doctor is unlikely to know much about the condition. One-page sheets have been developed to accompany the lab report and give the doctor a quick overview of what needs to be known and done.

Shields said it had been mentioned that genomic medicine could possibly be used as a leverage point to address larger problems within the medical system as a whole. Similarly, perhaps genomics could be leveraged to reduce health disparities. If a health information technology system were implemented to collect clinical data on patients, data about environment and demographics could be collected as well. Very little is known about the experiences and exposures of poor people in this country, and data collection on this scale could help explain some of the health disparities. In addition, disparities in access to and utilization of genomic technologies suggest that there needs to be an effort to communicate and deliver information in ways that are meaningful to all people. There are opportunities all along the trajectory from research to care to address racial, ethnic, and socioeconomic disparities.

One participant said that there remains a huge amount of work ahead in the integration of genomics into health care. The health care system in this country is complex and difficult to navigate, with or without money. Physicians must be given well-researched, straightforward tools that improve the time they spend interacting with patients, rather than tools that consume more time than they are worth.

6

Concluding Remarks

Wylie Burke, M.D., Ph.D.
Catherine Wicklund, M.S., C.G.C.

Burke and Wicklund were asked to list the main themes they had heard throughout the day's discussions.

Burke remarked that she noted five main themes:

1. The issues for genomic translation are embedded in issues for translation and health in general in our system. The problems that exist in the system are inescapably part of genomic translation.
2. Innovation should be supported, and technology can help reduce cost and increase patient convenience, but it should never entirely replace the opportunity for doctors and patients to interact.
3. There is a lack of robust methods for collecting and assessing data on innovative services in order to determine which ones are beneficial and which are not.
4. There is a potential need for a public–private partnership to generate data, particularly data from health systems with large populations in electronic databases (e.g., Veterans Administration).
5. Health technology assessments need to be deliberative processes that are prospective in nature.

Wicklund observed that discussion seemed to center around five different tensions:

1. "First, do no harm" versus encouraging innovation
2. Gathering data versus moving forward with "enough" data
3. Information technology versus the human element of medicine

4. Providing services that are appropriate versus providing services that are reimbursable
5. Providing access to genomic services to those who can afford them now versus the potential to exacerbate disparities

As a final point, one participant suggested that rather than trying to collect data on patients during their physician visits, genomics researchers might try to find a Framingham of their own to study an entire population over many years. She posited that many people in this nation would volunteer for such an endeavor. She noted that "the perfect is the enemy of the good" and cautioned against doing nothing for fear of not fixing the system entirely. She encouraged the Roundtable members to do what they can do, rather than what they should do—at least they will have done something.

The meeting was adjourned.

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Appendix A

Workshop Agenda

Innovations in Service Delivery in the Age of Genomics Workshop
July 28, 2008

Keck Building
Room 100
500 Fifth Street, NW
Washington, DC 20001

Workshop Goals:

1. To understand the current status of genetic and genomic service delivery
2. To explore how needs will change as genomic innovation progresses
3. To examine what types of alternative practice models will be needed

8:30 a.m. **Welcome and Introductions**
Wylie Burke, University of Washington
Chair, Roundtable on Translating Genomic-Based Research for Health

8:45 a.m. **Session 1: Current Status of Genetic Service Delivery**
Debra Lochner Doyle, Washington State Department of Health

9:05 a.m. **Session 2: The Strengths and Challenges of the Current System**
Disparities and access
Alexandra Shields, Harvard/Massachusetts General Hospital Center on Genomics, Vulnerable Populations & Health Disparities
Patient education and communication
Vivian Ota Wang, Genetic Counselor

Educational pipeline and workforce
Catherine Wicklund, National Society of Genetic Counselors

10:05 a.m. **Discussion**

10:30 a.m. **Break**

10:45 a.m. **Session 3: New Models for Service Delivery**
Heather Shappell, Informed Medical Decisions
Elissa Levin, Navigenics
Cynthia Prows, Cincinnati Children's Hospital

11:30 a.m. **Discussion**

12:15 p.m. **Lunch**

1:15 p.m. **Session 4: Vision of the Future**
Wylie Burke, University of Washington

1:45 p.m. **Discussion**

2:30 p.m. **Break**

2:45 p.m. **Session 5: Brainstorming on a Service Delivery Model for the Future**
Sharon Kardia, University of Michigan, *Moderator*
Frederick Chen, University of Washington
Catherine DesRoches, Institute for Health Policy, Massachusetts General Hospital
Bruce Korf, American College of Medical Genetics
Debra Lochner Doyle, Washington State Department of Health
Alexandra Shields, Harvard/Massachusetts General Hospital Center on Genomics, Vulnerable Populations & Health Disparities
Sharon Terry, Genetic Alliance
Vivian Ota Wang, Genetic Counselor
Catherine Wicklund, National Society of Genetic Counselors

4:00 p.m. **Discussion**

5:00 p.m. **Summing Up: Lessons Learned**
Wylie Burke, University of Washington
Catherine Wicklund, National Society of Genetic Counselors

5:30 p.m. **Workshop Adjourns**

Appendix B

Speaker Biosketches

Wylie Burke, M.D., Ph.D., is professor and chair of the Department of Medical History and Ethics at the University of Washington. She received a Ph.D. in genetics and an M.D. from the University of Washington and completed a residency in internal medicine at the University of Washington. She was a medical genetics fellow at the University of Washington from 1981 to 1982. Dr. Burke was a member of the Department of Medicine at the University of Washington from 1983 to 2000, where she served as associate director of the Internal Medicine Residency Program from 1988 to 1994 and as founding director of the University of Washington's Women's Health Care Center from 1994 to 1999. She was appointed chair of the Department of Medical History in October 2000. She is also an adjunct professor of medicine and epidemiology and an associate member of the Fred Hutchinson Cancer Research Center. She was a visiting scientist at the Centers for Disease Control and Prevention in 1998 and is a fellow of the American College of Physicians. She has served on the NIH National Advisory Council for Human Genome Research and the Secretary's Advisory Committee on Genetic Testing. Dr. Burke's research addresses the social, ethical, and policy implications of genetic information, including genetic test evaluation, the development of practice standards for genetically based services, and genetics education for health professionals. She is also the director of the University of Washington Center for Genomics and Healthcare Equality, a Center of Excellence in Ethical, Legal, and Social Implications (ELSI) research funded by the National Human Genome Research Institute (NHGRI).

Frederick M. Chen, M.D., M.P.H., acting assistant professor of family medicine, is an investigator in the WWAMI Rural Health Research Center in the Department of Family Medicine at the University of Washington and associate medical director of the Washington State Uniform Medical Plan, an employer-sponsored health insurance plan for state of Washington employees. He currently chairs the American Academy of Family Physicians' subcommittee on genomics. His research has focused on medical education, rural health, and health policy. Before returning to the University of Washington, he was the Kerr White Visiting Scholar at the Agency for Healthcare Research and Quality and an Atlantic Fellow in Public Policy at University College London.

Catherine DesRoches, Dr.PH., is an assistant in health policy at the Institute for Health Policy at Massachusetts General Hospital and an instructor in the Department of Medicine (Health Policy) at Harvard Medical School. She currently directs several large national survey projects examining the adoption of key health information technologies by providers and hospitals. She has played a key role in both national and international surveys of physicians and the general public. Her research interests include measurement and survey research, the adoption and effective use of health information technology, public knowledge of disparities in health care and health outcomes, health workforce issues, and physician professionalism.

Sharon Kardia, Ph.D., is an associate professor of epidemiology at the University of Michigan. She is director of the Public Health Genetics Program, codirector of the Michigan Center for Genomics and Public Health, and codirector of the Life Sciences & Society Program housed in the University of Michigan School of Public Health. Dr. Kardia received her doctoral degree in human genetics from the University of Michigan, was a postdoctoral fellow in the Department of Microbiology and Immunology, and continued postdoctoral work in the Department of Human Genetics. Her main research interests are in the genomic epidemiology of cardiovascular disease and its risk factors. She is particularly interested in gene–environment, gene–gene interactions and in modeling complex relationships among genetic variation, environmental variation, and risk of common chronic diseases. Her work also includes using gene expression and proteomic profiles for molecular classification of tumors and survival analysis in lung and ovarian cancers. As a part of her Michigan Center activity, Dr. Kardia is also actively working on moving genetics into chronic disease programs in state departments of health. She has been a member of three National Academy of Sciences committees (Genomics and the Public's Health in the 21st Century; Assessing Interactions Among Social, Behavioral, and Genetic Factors and Health; and Applications of Toxicogenomics Technologies to Predictive Toxicology).

Bruce R. Korf, M.D., Ph.D., received his M.D. from Cornell University Medical College and his Ph.D. in genetics and cell biology from Rockefeller University. He then completed training in pediatrics, pediatric neurology, and genetics at Children's Hospital, Boston. He served as clinical director in the Division of Genetics at Children's Hospital from 1986 to 1999 and as the medical director of the Harvard-Partners Center for Genetics and Genomics from 1999 to 2002. Currently he is the Wayne H. and Sara Crews Finley Professor of Genetics and chairman, Department of Genetics at University of Alabama, Birmingham. In his previous appointment at Harvard Medical School, he served as codirector of the course Genetics, Developmental and Reproductive Biology, taught to all first-year students at Harvard Medical School. His book based on this course, *Human Genetics: A Problem-Based Approach*, published by Blackwell Science, is currently in its third edition. He is also coauthor of *Medical Genetics at a Glance*, coeditor of the fifth edition of Emery and Rimoin's *Principles and Practice of Medical Genetics*, and coeditor of *Current Protocols in Human Genetics*. Dr. Korf is president-elect of the American College of Medical Genetics. He has completed terms as president of the Association of Professors of Human and Medical Genetics, member of the boards of directors of the American College of Medical Genetics and the American Society of Human Genetics, and member of the Liaison Committee on Medical Education. He currently serves on the National Cancer Institute Board of Scientific Counselors.

Elissa Levin, M.S., C.G.C., is a board-certified genetic counselor whose experience ranges from the research lab to medical centers to the Internet. Levin is currently the director of genetic counseling at Navigenics, where she developed the first comprehensive genetic counseling program to support genomic risk assessment for common health conditions. Prior to Navigenics, Levin was the director of clinical services at DNA Direct, where she helped pioneer one of the first direct-to-consumer genetic testing programs, providing testing, education, and counseling services for specific medical conditions. Levin began her career focused on the genetics of congenital heart disease, providing genetic counseling and education to families and staff in the cardiology division of the Children's Hospital of Philadelphia. At the University of California, San Francisco, Medical Center, she provided clinical services for clients of all ages about general and metabolic genetics and coordinated clinical trials for enzyme replacement therapy. Her efforts to increase awareness and access to genetic services and her dedication to setting high standards for providing reliable, professional services through the Internet have led to nationwide lectures, workshops, and training sessions for a variety of audiences, from consumers to health care providers.

Debra Lochner Doyle, M.S., C.G.C., is the designated state genetics coordinator for the Washington State Department of Health. She has a B.S. in genetics from the University of Washington and an M.S. in human genetics and genetic counseling from Sarah Lawrence College. She is board certified by the American Board of Medical Genetics and the American Board of Genetic Counseling. Before joining the Department of Health, she served as a cytogenetic technologist with Memorial Sloan-Kettering Cancer Research Center and as the senior genetic counselor for the Jones Institute for Reproductive Medicine, the Children's Hospital of the King's Daughters in Virginia, and Women and Infants Hospital in Rhode Island. She is a national leader, serving as a member of the Health Care Professional Advisory Committee and Current Procedural Terminology adviser for the American Medical Association, a founding member of the Economics of Genetic Services Committee of the American College of Medical Genetics, past president of the National Society of Genetic Counselors, and a founding member and past president of the Coalition of State Genetic Coordinators.

Vivian Ota Wang, Ph.D., C.G.C., F.A.C.M.G., is a National Institutes of Health (NIH) agency representative of the National Science and Technology Council of the Executive Office of the President to the National Nanotechnology Coordination Office and program director of the Ethical, Legal, and Social Implications Research Program of NHGRI. Previously she was a senior adviser to the director of the Office of Behavioral and Social Sciences Research at NIH. Prior to joining NIH, she held tenure-track faculty positions at Rutgers, Arizona State, and Vanderbilt universities, where her research program focused on information processing related to race and racial identity in genetics, education, and community outreach. She received the National Society of Genetic Counselors Special Projects Award to develop a multicultural genetic counseling curriculum. She is currently working on policy and risk communication issues related to the scientific, ethical, legal, and social implications of nanoscience and nanotechnology. Dr. Ota Wang's accomplishments have been recognized by numerous awards and honors. To name a few, she has been awarded a U.S. Department of Health and Human Services Secretary's Award for Distinguished Service, the Colorado College's Louis T. Benezet Award, and the Asian American Psychological Association's Distinguished Contributions Award. Dr. Ota Wang received a B.A. in biology from Colorado College, an M.S. in genetic counseling from the University of Colorado, and an M.Phil. and Ph.D. in counseling psychology from Columbia University. She is a fellow of the American College of Medical Genetics, a diplomate of the American Board of Genetic Counseling, a clinical laboratory specialist in cytogenetics, and a licensed psychologist.

Cynthia A. Prows, R.N., is a clinical nurse specialist in genetics at the Cincinnati Children's Hospital Medical Center (CCHMC). Ms. Prows directs the Genetics Education Program for Nurses (GEPN). This program started as the Genetics Program for Nursing Faculty in 1996 with funding from NHGRI's ELSI extramural research program. In 2003, web-based genetics education offerings were developed to meet the needs of the broader nursing community. Again, these were possible with funding from NHGRI's ELSI program as well as Health Resources and Services Administration's Division of Nursing. Various GEPN web-based offerings have been sustained through registration fees and have provided genetics education to more than 1,000 nurses. Ms. Prows has been a core member of the CCHMC Genetic Pharmacology Service since its inception and has published articles about pharmacogenetics in the nursing literature. Ms. Prows is a past president and an active member of the International Society of Nurses in Genetics, a fellow of the American Academy of Nursing, and a cochair of the Content and Instruction Work Group of the National Coalition for Health Professionals Education in Genetics. She has served on NIH scientific review panels and HRSA's National Advisory Council on Nurse Education and Practice.

Heather L. Shappell, M.S., C.G.C., is a board-certified genetic counselor who completed her graduate training at the University of Pittsburgh and has focused on hereditary cancer genetics throughout her professional career. She is a member of the Scientific Advisory Board of the hereditary cancer patient advocacy organization FORCE (Facing Our Risk of Cancer Empowered). In addition to her face-to-face clinical work at several National Cancer Institute–designated comprehensive cancer centers, Ms. Shappell was employed by Myriad Genetic Laboratories, Inc., where she provided hereditary cancer consultation and education to patients, physicians, and Myriad employees about hereditary cancer and genetic testing by telephone. Through her extensive contact with patients and health care providers throughout the United States, she recognized that limited access to genetic counseling expertise was compromising patient care. Her concern about patient access to quality care combined with her expertise in conveying detailed and life-saving information by telephone led her to create the first national company to address the problem of limited access to genetic counseling experts. As founder of Informed Medical Decisions, Ms. Shappell's combined experience in the field of hereditary disease, genetic testing, and telephone genetic counseling has enabled her to create new avenues of access to genetic counselors, empowering patients and their doctors to make the most informed decisions regarding genetic testing, disease screening, and prevention.

Alexandra E. Shields, Ph.D., is director of the newly formed Harvard/Massachusetts General Hospital (MGH) Center on Genomics, Vulnerable Populations & Health Disparities. She holds faculty appointments in medicine (health policy) at Harvard Medical School and MGH. Prior to joining the Harvard faculty, Dr. Shields was associate research professor in public policy at Georgetown University. She received her Ph.D. in social policy from the Heller School, Brandeis University, where she was a Pew Health Policy Scholar and an Agency for Health Care Policy and Research fellow. While at Brandeis, she also served as staff researcher for the Council on the Economic Impact of Health System Change. Prior to her doctoral work, Dr. Shields held several senior positions in state government, including director of the Bureau of Ambulatory Care for the Massachusetts Rate Setting Commission (now the Division for Health Care Finance and Policy), where she set reimbursement policy for all publicly purchased ambulatory services in the state. She also holds a B.A., summa cum laude, Phi Beta Kappa, in sociology and theology, and a master's degree, with distinction, in systematic theology from Boston College, where she was the Bernard J. Lonergan Scholar in Theology.

Sharon Terry is president and CEO of Genetic Alliance, a coalition of more than 600 disease-specific advocacy organizations working to increase capacity in advocacy organizations and to leverage the voices of the millions of individuals and families affected by genetic conditions. She is the founding executive director of PXE International, a research advocacy organization for the genetic condition pseudoxanthoma elasticum (PXE). She is at the forefront of consumer participation in genetics research, services, and policy and serves as a member of many of the major governmental advisory committees on medical research, including the Food and Drug Administration's Cellular, Tissue and Gene Therapies Advisory Committee and the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. She is a member of the board of directors of the Biotechnology Institute and on the advisory board of the Johns Hopkins Genetics and Public Policy Center funded by the Pew Charitable Trusts. She is the chair of the Coalition for Genetic Fairness, composed of advocates, health care providers, and industry working to enact effective federal policy to prohibit genetic information discrimination. She is also chair of the Social Issues Committee of the American Society of Human Genetics. In 2005, she received an honorary doctorate from Iona College for her work in community engagement and haplotype mapping. Ms. Terry is a cofounder of the Genetic Alliance Biobank and serves as president of its board. It is a centralized biological and data (consent/clinical/environmental) repository catalyzing translational genomic research on rare genetic diseases. The Biobank works in partnership with academic and industrial collaborators

to develop novel diagnostics and therapeutics to better understand and treat these diseases. Along with the other coinventors of the gene associated with PXE (ABCC6), she holds the patent for the invention. She codirects a 19-lab research consortium and manages 52 offices worldwide for PXE International.

Catherine A. Wicklund, M.S., C.G.C., is the director of the Graduate Program in Genetic Counseling at Northwestern University and an assistant professor in the Department of Obstetrics and Gynecology. She received her master's in genetic counseling from the University of Texas Graduate School of Biomedical Sciences. She has 15 years' experience in clinical genetic counseling and has provided prenatal and pediatric genetic services. Before joining Northwestern, she codirected the Graduate Program in Genetic Counseling at the University of Texas. While she was at the University of Texas, she was also the director of Genetic Counseling Services in the Department of Obstetrics, Gynecology, and Reproductive Medicine. She serves on the board of directors of the National Society of Genetic Counselors (NSGC) and is currently the immediate past president. As a leader in NSGC, she has represented the organization on several national committees, including the Secretary's Advisory Committee on Genetics, Health, and Society, and she is a member of the Institute of Medicine Roundtable on Translating Genomic-Based Research for Health. She is also active on a state level and is working with the Illinois Department of Public Health on genetics education and finance and reimbursement issues and is on the Genetic and Metabolic Diseases Advisory Committee.