Challenges and opportunities for evidence-based genetics practice

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As someone whose professional responsibilities are primarily concerned with issues outside of the day-to-day practice of genetic medicine, I am profoundly encouraged by the disciplined and responsible intellect that is reflected in the pages of *"Genetics in Medicine."* The challenges in American medicine today are largely concerned with the availability of new clinically relevant scientific knowledge and its appropriate translation into population and individual health and medical care interventions by knowledgeable health professionals and patients. Our nation struggles with the interrelated burdens of escalating health costs; tragic numbers of uninsured people; unacceptable incidence rates of preventable and chronic disease; suboptimal use of existing health care assets; and insufficiently prepared health care consumers. Into this context are delivered the innovations that flower from the genomic revolution. If the health of the nation is to be improved through affordable access to these innovations, successful attention to at least the following clinical evidence-related priorities is required.

Enhancing the research infrastructure for prevention, clinical, and health services research

Although innovation is interesting, it becomes exciting when we understand whether it works; whether it works better than existing interventions; and whether it is more cost-effective and of greater comparative value than other interventions when evaluated from the perspective of total cost of care. Relatedly, many effective tests are often utilized on the wrong target population at a cost of up to \$3000 per intervention. It is of concern that the availability of existing translational knowledge in genetics is insufficient to meet today's needs. And, without urgent action, will be significantly inadequate to meet emerging needs. In addition to statistically robust, well-designed randomized controlled trials; well-designed cohort studies; large single- and multi-site observational studies, we also need new tools, methodologies, researchers, expertise, and enhanced infrastructures. As such, the genetics community should intensify its advocacy for the research budgets not only of National Institutes of Health; but also the Centers for Disease Control, Agency for Healthcare Research and Quality, and Food and Drug Administration (FDA), and specifically call for more

genetics-related research support. The public voice of scientists and clinicians, when properly harnessed, can be very persuasive.

Maturing the oversight and processes for translation of evidence-based genetic knowledge into practice

The successful introduction of new products, techniques, and services into health care requires mature infrastructures and processes for advancing the assessment and evaluation of evidence for key stakeholders including researchers, manufacturers, health professionals, public and private sector health payers, employers, and consumers. Despite years of discussion, there is still concern about important gaps in the federal oversight authority for many genetic tests and the relative responsibilities of the FDA and Health Resources and Services Administration's Clinical Laboratory Improvement Act authority. We need to achieve an appropriate oversight and regulatory infrastructure that provides protections and necessary data while not stifling innovation.

Today, we see the emergence of organizations, such as the Institute for Clinical and Economic Review and the Center for Medical Technology Policy, that provide neutral forums that produce assessments of new interventions; translate findings in support of value-based insurance benefit designs, coverage, and reimbursement policy; and advance studies in areas that are not amenable to the traditional randomized control trials. In genetics, the progress of the Evaluation of Genomic Applications in Practice and Prevention and its Working Group in developing a systematic process for evidence-based assessment is encouraging.

Our challenge is to harness, coordinate, and find sustainable financing for the forums necessary to support stakeholder decisions within the context of escalating costs and health care affordability concerns.

Supporting clinicians and their professional societies in the translation of evidence into clinical guidance

The foundation of American medicine is based on the ethically based expectation that physicians will consistently define, refine, and comply with the standards that inform clinical practice. If we have learned anything about health care delivery, it is the importance of physician leadership and desire for autonomy in providing their peers with clinical guidance. Given the pace of discovery in medicine generally, and innovation in genetics specifically, the resources of professional societies are inadequate to address contemporary and emerging challenges. Without urgent attention to this deficit, it is unreasonable to expect that clinical practice will be sufficiently informed and consistently updated by timely translation of evidence into expert clinical guidance. Given this public role of medical specialty societies and the "common good" that is derived, it is

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appropriate and necessary for new grant support to be made available for this important work.

Expanding the availability and training of the health professional workforce necessary to translate genetic-based evidence into appropriate health care for people

The availability of a competent health professional workforce is of obvious importance to the realization of evidence-based genetic practice. There is reason for concern about the training and continuing professional development of today's workforce. Mastering a lifetime of learning that increasingly includes the translation of new scientific knowledge into *individual* patient-appropriate practice requires a support infrastructure that is currently inadequate to meet the needs of today's clinicians. Although we can be excited about the new opportunities for more precise drug therapy, tracking disease progression through markers, and predicting the interactions between multiple disease processes that all lead to more tailored individual care, there is reason for concern about how well prepared clinicians are to effectively utilize these new capabilities. Of equal concern is the extremely limited supply of trained and "certified" professionals available to counsel and support patients and their families in their use of genetic information in making personally appropriate choices and decisions. We have yet to create the infrastructure to recruit, train, and certify sufficient genetic counselors to meet the needs of patients or physicians.

As such, urgent attention is required not only in the medical school curriculum, but also in the processes of Continuing Medical Education and the assessment of continuing professional development within the requirements of the criteria for Maintenance of Certification by our medical specialty boards. Expanding the availability of trained genetic counselors should be a fundamental priority for our nation's academic health science centers and the genetic counseling professional societies must continue to advance the certification criteria that will facilitate appropriate reimbursement for their services.

Developing the criteria and the data to evaluate the performance of the prevention and delivery system in its use of genetic-based health care assets

A defining characteristic of the contemporary medical care system is its attention to the transparent evaluation of quality and cost-effectiveness in clinical performance. Arising directly from evidence-based science and clinical guidance, performance measures are now available to describe a wide variety of clinical care interventions. Additionally, clinical data infrastructures are becoming more robust and aggregated. At its best, this performance feedback has become a significant tool in advancing the continuing professional development of the clinician, and providing consumers with guidance regarding the best quality and most appropriate health professionals to meet their individual needs. Given the specificity of genetic-based disorders, it is inevitable that there will be an increased demand in the future for transparent differentiation of expertise by health care purchasers, payers, and consumers. As such, it is timely that the measures of performance be advanced by appropriate clinical experts and that mechanisms for accumulating the necessary data to evaluate performance be integrated into the design of existing databases. Enforcement of protections for privacy and confidentiality afforded by the Genetic Information Nondiscrimination Act are essential to advancing this agenda.

Standing outside of the genetics laboratory and the clinical examination room, one cannot help but be excited by the potential for more personally appropriate health and medical care interventions that are promised by the genomic revolution. However, one can also be concerned that the infusion of new information, tools, and products could easily exacerbate the quality, cost efficiency, and accessibility problems that already plague the nation's health system. All stakeholders including researchers, clinicians, payers, purchasers, policy leaders, elected and appointed officials, and patients, have a common stake in advancing the intelligent evidence-based introduction of genetics into clinical practice. We should take every opportunity to accomplish at least these five priorities.