EDUCATION

How to know when physicians are ready for genomic medicine

Jason L. Vassy,[1,2,3*] Bruce R. Korf,[4] Robert C. Green[3,5]

Despite perceptions to the contrary, physicians are as prepared for genomic medicine as they are for other medical innovations; educational initiatives and support from genetics specialists can enhance clinical practice.

With the tremendous investment in the genomic sciences over the last two decades, the biomedical community is eager to apply new genomic knowledge to patient care. Genomic testing, including whole-genome and exome sequencing, has demonstrated clinical utility in certain contexts (*1*). However, the workforce of fewer than 2000 board-certified medical geneticists is insufficient to meet the demand created by the greater prominence of genomics in clinical medicine (*2*). As a result, physicians without specialized genetics training will be increasingly called upon to order genomic testing and use the results in the care of their patients. At the same time, several studies have found that physicians report "unpreparedness" and low confidence in their ability to apply genomic data to patient care (*3*). Here we discuss how to assess whether physicians are ready for the genomic revolution and whether previous medical innovations have been held to the same standards.

POTENTIAL PERILS OF UNPREPAREDNESS

Despite recent standardization efforts, most molecular laboratories do not currently use a uniform approach to the clinical interpretation of the large number of genetic variants revealed by genome sequencing, most of which have unknown clinical significance. Even for variants known to be pathogenic, often insufficient data exist to predict the likelihood of disease in an individual who is currently asymptomatic. Communicating this uncertainty to the clinician remains a challenge for the clinical laboratory. For example, if genomic sequencing for a 60-year-old patient with no signs, symptoms, or family history of heart disease uncovers a rare genetic variant in a known cardiomyopathy gene, the treating clinician has no data to guide her in interpreting the significance of that finding for her patient. The complexity of clinical sequencing reports might be prone to misinterpretation by nongeneticist physicians, leading to over- or underestimation of the disease risk associated with a given variant. Such results might prompt physicians to order an expensive cascade of follow-up diagnostic tests, each with its own potential complications, risks to the patient, and costs (*4*).

The concerns voiced above reflect an appropriate respect for key principles in medicine, including the avoidance of patient harm and good stewardship of limited health care resources. But at what point do they create an artificial crisis that distracts from more productive questions that might point the way forward for a new field? If physicians are not ready to use genomic medicine now, how will we know when they are? A useful thought experiment is to ask the same of other medical innovations. When did the biomedical community declare nonradiologist physicians ready to order computerized tomography (CT) scans for their patients and make clinical decisions based on their findings? Introduced in the 1970s, CT carries risks associated with radiation, intravenous contrast agents, and the discovery of radiographic lesions incidental to the test's primary indication. However, with guidance from their radiologist colleagues, physicians routinely use CT for the standard management of conditions ranging from abdominal pain and headaches to cancer and stroke.

On what grounds would we hold genomic technology to different standards? Similar to the radiologist, the genomics-laboratory director holds primary responsibility for interpreting sequencing results using available data and generating reports that physicians can understand. The receiving physicians may choose to communicate back with the specialist for further guidance in medical decision-making. As genomic medicine grows in prominence, nongeneticist physicians might also seek greater support from genetic counselors than they now do in practice.

Genetic exceptionalism is the idea that genetic technology and information are inherently different from other routine processes in medical care and, by extension, should be handled more cautiously (*5*). Additional caution might be appropriate for genetic information that is potentially stigmatizing or anxiety provoking. But like any other medical test, most genetic testing is used for diagnosis, prognosis, and therapeutics. Similarly, the degree of physician preparedness for genomic medicine is not exceptional among other complex medical innovations such as myriad types of imaging, microscopic pathology assessment, or targeted therapies.

DEFINING PREPAREDNESS

The concerns about physician preparedness for genomic medicine are also problematic because no universal definition of preparedness exists. To a large extent, these concerns stem from surveys in which physicians have reported little experience with genomic medicine in their practices and low perceived confidence in their ability to order genetic tests and manage the results appropriately (*3*). But self-reported attitudes and perceptions do not necessarily correlate with skills and behavior. Even objective genomics knowledge assessment might not adequately determine whether a physician can use genomic medicine in his or her practice.

To assess residency education across the spectrum of medical specialties, the Accreditation Council for Graduate Medical Education (ACGME) has identified six core competencies that training programs should target: patient care, knowledge for practice, practice-based learning and improvement, interpersonal and communication skills, professionalism, and systems-based practice; the Association of American Medical Colleges (AAMC) has added two others: interprofessional collaboration and personal and professional development (*6*). Although these discrete competencies facilitate the evaluation of trainees, they oversimplify what it means to be an effective physician. To better describe the roles a physician plays, medical specialties are now defining entrustable professional

[1]Section of General Internal Medicine, VA Boston Healthcare System, Boston, MA 02130, USA. [2]Division of General Medicine and Primary Care, Department of Medicine, Brigham and Women's Hospital, Boston, MA 02115, USA. [3]Department of Medicine, Harvard Medical School, Boston, MA 02115, USA. [4]Department of Genetics, University of Alabama at Birmingham, Birmingham, AL 35233, USA. [5]Division of Genetics, Department of Medicine, Brigham and Women's Hospital, Boston, MA 02115, USA.
*Corresponding author. E-mail: jvassy@partners.org

	Family history	Genomic testing	Patient treatment based on genomic results
EPAs:	**Elicit, document, and act on relevant, pertinent family history**	**Use genomic testing appropriately to guide patient management**	**Use genomic information to make treatment decisions**
Competency			
Patient care	Assemble family history Use standard pedigree symbols Recognize patterns of Mendelian inheritance and calculate risks	Discuss implications for genomic testing (benefits, risks, and alternatives) Order, interpret, and communicate results of appropriate genomic tests Provide referral to appropriate specialist	Identify medical conditions and drug responses that have a strong genetic component Discern potential clinical impact of genetic variation on risk stratification and individualized treatment
Knowledge for practice	Describe basic patterns of Mendelian inheritance	Explain how genomic changes may cause different phenotypes Explain concepts of analytical/clinical validity and utility	Identify single-gene disorders that may be amenable to targeted pharmacologic therapy Recognize genomic test results that may guide choice of therapy
Practice-based learning and improvement	Incorporate family history into health record	Incorporate genomic findings into health record and care plan	Use evidence-based recommendations
Interpersonal and communication skills	Explain and document findings from family history to patient, including implications for other family members	Ensure joint decision-making between physician and patient	Discuss benefits, risks, and alternatives of preventative and therapeutic approaches driven by genomic findings
Professionalism	Respect privacy of patient and family in assembling family history	Be aware of and respond to patient's concerns about genetic discrimination	Respect and guard privacy of patient and family members
Systems-based practice	Facilitate patient's desire to communicate family history to health providers and family members	Facilitate access to relevant clinical studies and trials based on testing	Be aware of patient's needs as an individual who has a genetic disease or pharmacogenomic variation
Interprofessional collaboration	Make appropriate referrals for specialty evaluation	Talk with clinical laboratory to ensure that test(s) are ordered and interpreted appropriately	Recognize potential involvement of multiple organ systems in genetic disorders and seek appropriate consultation
Personal and professional development	Identify sources of information on genetic disorders	Engage in CME regarding advances in genomic medicine and changing indications for and interpretation of genomic testing	Be familiar with available databases and resources relevant to genetic variation

PHOTO CREDIT: A. GAULT/*GETTY IMAGES*

Fig. 1. Get ready for genomic medicine. Shown are three EPAs for physicians and examples of the competencies each requires. Not shown are EPAs for somatic and microbial genomics. CME, continuing medical education.

activities (EPAs), the "professional activities that together constitute the mass of critical elements that operationally define a profession" (*7*). Each EPA (for example, triaging ill patients to an appropriate level of medical care) can map to several competencies (for example, the medical knowledge, procedural skills, and professionalism required to triage a patient appropriately).

Efforts are under way to apply this educational framework to genomic medicine for the nongeneticist. In anticipation of the more widespread diffusion of genomics into clinical medicine, genomics education has become a priority for the National Human Genome Research Institute (NHGRI) of the U.S. National Institutes of Health. It has developed a Genetics/Genomics Competency Center for Education (G2C2) to curate high-quality curricula and other resources for physicians and other health care providers, searchable by genomic medicine competencies. Through its Inter-Society Coordinating Committee on Practitioner Education in Genomics (ISCC-PEG), NHGRI is partnering with more than 30 professional organizations representing generalists and specialists, such as the American Medical Association, the American Heart Association, and the American Board of Ophthalmology (*8*). The ICSS-PEG has developed five genomics EPAs in the areas of (i) family history, (ii) genomic testing, (iii) treatment based on genomic results, (iv) somatic genomics, and (v) microbial genomics, each of which can be mapped to specific genomic medicine competencies (Fig. 1) (*3*). The ICSS-PEG recognizes that "preparedness" for genomic medicine will have different definitions for different types of physicians; thus the committee addresses common educational needs while supporting individual professional societies and specialty boards in developing their own genomics EPAs for their members. For example, for the primary care physician, the EPA of eliciting, documenting, and acting on relevant family history pertinent to a patient's clinical status might include the recognition of a potential hereditary colorectal cancer syndrome such as Lynch syndrome. For the ophthalmologist, this EPA would include using a family history assessment in a patient with retinitis pigmentosa to identify the potential implications of its inheritance pattern for family members. It is unknown whether the application of an EPA framework to genomic medicine will improve clinical practice, but defining genomic medicine competencies and EPAs is a first step toward evaluating the degree to which medical learners at all stages of training and practice are meeting them.

THE NONEXCEPTIONALISM OF GENOMIC PREPAREDNESS

Beyond physician education, there remain several challenges to the clinical integration of genomic medicine. For many clinical contexts, the impact of genomic testing on patient outcomes, such as improved survival or quality of life, remains to be demonstrated (*9*). The electronic health records of most health systems are not currently prepared to incorporate the complexity of genomic data into clinical care, despite the tremendous opportunities for integrated clinical decision support to help busy physicians use genomic data in medical decision-making (*10*). However, the preparedness of physicians themselves for genomic medicine is not unique among other diagnostic and therapeutic innovations. The fundamental principles of medicine in genomics are the same as elsewhere in medicine—modest training and support from specialists and health systems will prepare nongeneticist physicians to use genomics in the care of their patients.

REFERENCES AND NOTES

1. L. G. Biesecker, R. C. Green, Diagnostic clinical genome and exome sequencing. *N. Engl. J. Med.* **370**, 2418–2425 (2014).
2. B. R. Korf, Genomic medicine: Educational challenges. *Mol. Genet. Genomic Med.* **1**, 119–122 (2013).
3. B. R. Korf, A. B. Berry, M. Limson, A. J. Marian, M. F. Murray, P. P. O'Rourke, E. R. Passamani, M. V. Relling, J. Tooker, G. J. Tsongalis, L. L. Rodriguez, Framework for development of physician competencies in genomic medicine: Report of the Competencies Working Group of the Inter-Society Coordinating Committee for Physician Education in Genomics. *Genet. Med.* **16**, 804–809 (2014).
4. A. L. McGuire, W. Burke, An unwelcome side effect of direct-to-consumer personal genome testing: Raiding the medical commons. *JAMA* **300**, 2669–2671 (2008).
5. M. J. Green, J. R. Botkin, "Genetic exceptionalism" in medicine: Clarifying the differences between genetic and nongenetic tests. *Ann. Intern. Med.* **138**, 571–575 (2003).
6. R. Englander, T. Cameron, A. J. Ballard, J. Dodge, J. Bull, C. A. Aschenbrener, Toward a common taxonomy of competency domains for the health professions and competencies for physicians. *Acad. Med.* **88**, 1088–1094 (2013).
7. O. ten Cate, F. Scheele, Competency-based postgraduate training: Can we bridge the gap between theory and clinical practice? *Acad. Med.* **82**, 542–547 (2007).
8. T. A. Manolio, M. F. Murray, Inter-Society Coordinating Committee for Practitioner Education in Genomics, The growing role of professional societies in educating clinicians in genomics. *Genet. Med.* **16**, 571–572 (2014).
9. W. G. Feero, T. A. Manolio, M. J. Khoury, Translational research is a key to nongeneticist physicians' genomics education. *Genet. Med.* **16**, 871–873 (2014).
10. C. L. Overby, I. Kohane, J. L. Kannry, M. S. Williams, J. Starren, E. Bottinger, O. Gottesman, J. C. Denny, C. Weng, P. Tarczy-Hornoch, G. Hripcsak, Opportunities for genomic clinical decision support interventions. *Genet. Med.* **15**, 817–823 (2013).

Acknowledgments: The authors are supported by the U.S. National Institutes of Health (grant numbers KL2-TR001100, L30-DK089597, R01-HG005092, U01-HG006500, and U19-HD077671).

10.1126/scitranslmed.aaa2401

Citation: J. L. Vassy, B. R. Korf, R. C. Green, How to know when physicians are ready for genomic medicine *Sci. Transl. Med.* **7**, 287fs19 (2015).

Science Translational Medicine

How to know when physicians are ready for genomic medicine

Jason L. Vassy, Bruce R. Korf and Robert C. Green

Sci Transl Med **7**, 287fs19287fs19.
DOI: 10.1126/scitranslmed.aaa2401