Global Genomic Medicine Collaborative: Genomics Education

Bruce R. Korf, MD, PhD
Department of Genetics
University of Alabama at Birmingham
Evidence from the United States and abroad suggests inadequate genetics education of health care professionals as a significant factor limiting the integration of genetics into clinical care. Specific inadequacies include the amount and type of genetics content included in undergraduate professional school curricula and the small amount of genetics-related knowledge and skills of physicians, nurses, and other health professionals once they enter clinical practice. Modifications in medical, dental, nursing, public health, and pharmacy school curricula and in medical residency training programs are needed to ensure that health care professionals entering the workforce are well-trained in genetics.
Case 1

- 65 year old woman referred to UAB Undiagnosed Diseases Program
- Obesity, chronic fatigue, heat intolerance
- Interested in whole genome sequencing
- Incidental diagnosis of spinocerebellar ataxia 14
- Exam revealed no neurological signs
- No family history of spinocerebellar ataxia
Interpretation
This individual possesses DNA sequence variants of unknown clinical significance in one of the sequenced genes. In addition they possess additional variants of unknown significance or borderline repeat expansion mutations in another gene. It cannot be determined whether this individual is likely to be affected with, or predisposed to developing ataxia associated with the specific borderline repeat(s) or DNA sequence variants of unknown significance. Therefore the interpretation is indeterminate. Please refer to the Comments section for further information and please contact our genetic counselor at 1-800-394-4493 to discuss these results.

Technical Results

<table>
<thead>
<tr>
<th>Test</th>
<th>Results</th>
<th>Alert</th>
<th>Repeats</th>
</tr>
</thead>
<tbody>
<tr>
<td>SCA1</td>
<td>Normal</td>
<td>32 and 30</td>
<td></td>
</tr>
<tr>
<td>SCA2</td>
<td>Normal</td>
<td>22 and 22</td>
<td></td>
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<tr>
<td>SCA3</td>
<td>Normal</td>
<td>28 and 14</td>
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<td>SCA6</td>
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<td>13 and 12</td>
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<td>10 and 10</td>
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<td>SCA8</td>
<td>Normal</td>
<td>24 and 24</td>
<td></td>
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<tr>
<td>SCA10</td>
<td>Normal</td>
<td>18 and 12</td>
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<td>SCA17</td>
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<td>39 and 38</td>
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<tr>
<td>DRPLA</td>
<td>Normal</td>
<td>19 and 18</td>
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<tr>
<td>FRDA1</td>
<td>Normal</td>
<td>4 and 4</td>
<td></td>
</tr>
</tbody>
</table>

SCA14 Result: Variants of Unknown significance
SETX Result: Variants of Unknown significance
POLG1 Result: Normal
SCA5 Result: Normal
SIL1 Result: Normal
TTPA Result: Normal
KCNC3 Result: Normal

SCA14 Variant 1: Transition T > C
Nucleotide position: 1497
Codon position: 499
Amino acid change: None
DNA variant type: Variant of unknown significance

SETX Variant 1: Transition G > A
Nucleotide position: IVS13 +10
Codon position: DNR
Amino acid change: DNR
DNA variant type: Variant of unknown significance, heterozygous

SETX Variant 2: Transition G > A
Nucleotide position: 6507
Codon position: 2169
Amino acid change: None
DNA variant type: Variant of unknown significance, heterozygous

APTX Result: Normal
Outcome

• VUS reclassified as benign variant (7 years later)
• WGS done and no pathogenic variants found to explain her phenotype
Case 2

• 32 year old man seen for genetic counseling
• Family history of breast cancer in his father and paternal aunt
• Paternal aunt positive for pathogenic BRCA2 variant
• Has two siblings without cancer, both also positive
• He was tested and was told result was negative – presented for options for additional testing
<table>
<thead>
<tr>
<th>Collection DT</th>
<th>Specimen</th>
<th>Test Name</th>
<th>Result</th>
<th>Units</th>
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<td>BLOOD</td>
<td>BRCA1 SEQUENCING NEGATIVE</td>
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<td>INTERP SUMX SEE NOTE</td>
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<td>BRCA1 SEQ INTERP SEE NOTE</td>
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<td>BRCA1 DEL/DUP NEGATIVE</td>
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<td>BRCA1 DEL/DUP INTRON NOTE</td>
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<td></td>
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<td>BRCA2 SEQUENCING SEE NOTE</td>
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<td>COMP INTERP SEE NOTE</td>
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<td>ADDITIONAL INFO SEE NOTE</td>
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</table>

Comment: Interpretation summary
Comment: POSITIVE FOR A KNOWN PATHOGENIC MUTATION
Comment: BRCA1 Seq Interp
Comment: NO MUTATION DETECTED
Comment: BRCA1 Del/Dup Intexp
Comment: NO MUTATION DETECTED
Comment: BRCA2 sequencing
Comment: c.1755_1759del (p.Lys585Asnfs*3)
Comment: BRCA2 Seq Interp
Comment: KNOWN PATHOGENIC
Comment: BRCA2 Del/Dup Interp
Comment: NO MUTATION DETECTED
Comment: Comprehensive Interp
Comment: This test has identified one copy of the
Comment: c.1755_1759del mutation (also known as
Comment: c.1755_1759delCTGAAA) in exon 10 of the BRCA2 gene.
Comment: This frameshift mutation causes the premature
Comment: termination of BRCA2 protein synthesis.
Comment: (p.Lys585Asnfs*3), and is described in multiple
Comment: BRCA online databases as being pathogenic (see the
Comment: Breast Cancer Information core
Comment: https://research.nbirnih.gov/projects/bic/,
Comment: clinvar http://www.ncbi.nlm.nih.gov/clinvar/, and
Comment: AKUP for BRCA2
Comment: http://arup.utah.edu/database/BRCA/Variants/BRCA2)
Comment: Therefore, this individual is at increased risk

(continued)

Comment: of developing BRCA related cancers. Genetic
Comment: counseling and DNA testing for at-risk family
Comment: members are recommended. Laboratory results and
Competencies

- Recognize indications for testing
- Select appropriate family member to test first
- Discuss issues of payment/risks/benefits
- Select a laboratory
- Interpret report – recognize limitations
- Genomic sequencing – recognize potential for secondary findings
- Refer to specialist as needed
- Discuss results with family
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

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The premise of genomic medicine to maintain health, facilitate diagnosis, and cure or mitigate disease is dependent on the skillful translation of genomic science into meaningful actions at the bedside and in the clinic. Surveys of both primary-care and specialist physicians, often by their professional societies, reveal a desire and even a willingness, to care for genomic data.3,13 The use of genomics in caring for patients with certain cancers and for some pediatric patients is increasing in routine diagnosis and treatment, and this trend is likely to expand to other areas of medical practice in the coming years.4

Nearly half of practicing clinicians in the United States are more than 50 years of age, medical school and residency training for these physicians occurred before the completion of the Human Genome Project and the breakthroughs in genomic medicine.3 Current trainees are faced with a rate of progress in genomics that renders much of what they have learned out of date by the time they enter practice. Considering the rapid rate of change, substantial redesign in the cost of genome sequencing, and the increasing relevance of genomic information to the practice of medicine, the barriers to implementing genomic discoveries within medical practice have to be overcome. Moreover, misuse of genomics by untrained health-care providers may incur cost without advantage and may result in harm to patients based on inaccurate diagnosis or use of unnecessary or incorrect tests.

The National Human Genome Research Institute, together with 23 professional societies, 15 other institutions at the National Institutes of Health, and other organizations interested in physician education, developed the Inter-Society Coordinating Committee for Physician Education in Genomics (ISCC) in the spring of 2013 (see Supplementary Data S1 online). ISCC member organizations focus on physician training, starting with medical school matriculation and continuing through residency and fellowship for active clinicians. The ISCC seeks to improve genomic literacy of physicians and other practitioners and to enhance the practice of genomic medicine through sharing of educational approaches and joint identification of educational needs. The ISCC developed four working groups: Genomic Medicine Competencies, Educational Providers, Use Cases, and Specialty Boards (see Supplementary Data S1 online).

The Genetic Medicine Competencies Working Group was charged with the development of a framework whereby...

The ISCC seeks to improve genomic literacy of physicians and other practitioners and to enhance the practice of genomic medicine through sharing of educational approaches and joint identification of educational needs. The ISCC developed four working groups: Genomic Medicine Competencies, Educational Providers, Use Cases, and Specialty Boards (see Supplementary Data S1 online).
ISCC Competencies in Genomic Medicine

• Elicit, document, and act on relevant family history pertinent to patient’s clinical status
• Utilize genomic testing appropriately to guide patient management
• Utilize genomic information to inform treatment decisions
• Support the use of genomic information to guide the diagnosis and management of cancer and other disorders involving somatic genetic changes
• Utilize genomic tests that identify microbial contributors to human health and disease, as well as genomic tests that guide therapy of infectious diseases
Global Genomic Medicine Collaborative (G2MC)

- Genomic medicine is global
- Genome ‘hubs’ in most continents
- Explore synergies, redundancies, collaborative opportunities
- Opportunities to advance the genome sciences as a global agenda and to impact global health
Global implementation of genomic medicine: We are not alone


Around the world, innovative genomic medicine programs capitalize on singular capabilities arising from local health care systems, cultural or political milieu, and unusual select risk alleles or disease burdens. Such individual efforts might benefit from the sharing of approaches and lessons learned in other local scales. The US National Human Genome Research Institute and the National Academy of Medicine recently brought together 23 of these groups to compare projects, discuss the current state of implementation and identify near-term capabilities, and to identify opportunities for collaboration that promote the responsible practice of genomic medicine. Efforts to coalesce these groups around concrete but compelling signature projects should accelerate the responsible implementation of genomic medicine in efforts to improve clinical care worldwide.

The growing number of advances in human genomics that are directly relevant to disease diagnosis, treatment, and prevention coupled with the high cost of genomic sequencing has promoted the use of genomic technologies in routine clinical care (1). Among the many challenges to widespread implementation of genomic medicine, what looks least is the lack of evidence to demonstrate improved clinical or economic outcomes (2). Other needs include standardization and quality assurance of genomic data produced by clinical laboratories, a clinical-informatics infrastructure for managing genomic information, education for health professionals and patients in using the information, and policies for data sharing that permit ongoing capture of generalizable clinical experiences in what has been termed “evidence-generating medicine” (3). A host of ongoing efforts exist worldwide to establish national implementation strategies for genomic medicine (table S1) (4), but many such efforts are being conducted in relative isolation. Sharing of strategies, data, and standards could minimize wasteful duplication and speed progress in identifying genomics-based interventions and translating them to the clinic.

To assess the current global state of the art (2), the US National Human Genome Research Institute and the US National Academy of Medicine convened 90 leaders in genomic medicine from the United States and 25 other countries on five continents for a Global Leaders in Genomic Medicine symposium in 2014 (table S1). Although the organizers attempted to identify and invite every nation working on the implementation of genomic medicine, participation was somewhat restricted by the lack of systematic information on such efforts and limited travel funding (see full list of participating countries at www.genome.gov/27550775). Here we summarize efforts described by the participants, with an emphasis on 13 regions with singular capabilities because of the structures of their health care systems, cultural or political readiness for implementation, or unusual disease burdens or risk allele frequencies (5) the current state of implementation in the various countries and capabilities desired over the next 3 to 5 years and (6) opportunities for collaboration to promote the responsible implementation of genomic medicine.

INTERNATIONAL LANDSCAPE

Early efforts at genomic medicine collaborations were initiated by the European Association for Predictive, Preventive, and Personalised Medicine (EAPPM), the European Commission’s Framework and observatory, and the European Federation of the Medicine Alliance (table S1) (5), which have spearheaded promising projects such as the application of genome sequencing in pharmacogenomics and the development of online pharmacogenomic resources. Related efforts include the International Rare Disease Research Consortium (IRDRC), which develops new diagnostic strategies and therapies for rare diseases, the Alliance for Genomics and Precision Health (AGAPP), which promotes responsible sharing of genomic data for research (see supported context [table S1]), which is drafting professional guidelines for diagnostic DNA sequencing.

In an informal poll of participants prior
2015: Global Attendance

>100 International Genomic Medicine Leaders

> 25 Countries
Working Groups

- IT/bioinformatics
- Education/workforce
- Pharmacogenomics
- Evidence
- Policy
- Sequencing
Education
[Bruce Korf, Vajira Dissanayake]

• Genomic Medicine Workforce
  • What are the kinds of health professionals who practice genomic medicine in your country?
  • What is the training required for these professionals?
  • What is their scope of practice?
  • How do other health professionals interact with genomic medicine professionals, if they exist?
  • Are genomic medicine training programs in consideration if they do not currently exist?

• Method
  • Develop survey to share with members and other international contacts
  • Report and store information on central web site
Education, continued

• Case Based Teaching
  • Develop examples of cases used for teaching genomic medicine
  • Place in open-source format for use by G2MC members to use or modify
  • Expect that those who modify a case will post the modification
  • Attention to use of copyrighted material or patient photos

• Grand Rounds
  • WebEx webinars hosted by different members of G2MC
    • Monthly meetings
    • Some participate in real time, others by viewing recording
  • Content
    • Country-specific projects
    • Case conferences
  • Q&A
    • Real time for those who can participate
    • Maintain online videos and discussions
Grand Rounds
03 August 2016 - PDT 06.00 am; CDT 08.00 am; EDT 09.00 am; London 2.00 pm; Dubai 05.00 pm; New Delhi 06.30 pm; Manila 09.00 pm; Brisbane 11.00 pm

Regeneron / Geisinger DiscovEHR Programme

David Carey, PhD
Director
Weis Center for Research
Geisinger Health System
USA

Visit bit.ly/g2mcaugust to register
G2MC Grand Rounds

May: UAB Undiagnosed diseases programme – Dr. Bruce Korf

June: Pharmacogenomic Initiatives in Thailand – Dr. Wasun Chantratita, Mahidol University, Thailand

July: The BEST Network as a online tool for sharing and delivery of online genetic resources in genetics – Nicholas Hawkins, University of Queensland, Australia

August: Regeneron/Geisinger: DiscovEHR Programme – David Cary PhD, Geisinger Health System

YouTube Channel Link
https://www.youtube.com/channel/UCOMoopNfFVht04ukMMmHXKg

To subscribe to the email list for notifications

Please send a blank email to subscribe-g2mc@hgucolombo.org
Case 4.1

Alex is a four-year old boy with developmental delays and dysmorphic features. He has a small VSD and short stature. He is an only child and referral for genetics evaluation has not revealed a diagnosis.

The Diagnostic Odyssey
Case 4.2

He is seen by a geneticist, who recommends exome sequencing. This is done, and he is found to have a mutation in the MLL2 gene, indicative of Kabuki syndrome.
Case 4.3

When Alex’s parents speak with the geneticist they learn something else unexpected: Alex carries a known pathogenic mutation in the *BRCA1* gene. There is no known family history of breast or ovarian cancer, but Alex’s father was adopted.
Incidental Findings

American College of Medical Genetics and Genomics

ACMG Recommendations for Reporting of Incidental Findings in
Clinical Exome and Genome Sequencing

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S. Kalia, ScM, CGC1, Bruce R. Korf, MD, PhD7, Christa L. Martin, PhD, FACMG8, Amy McGuire,
JD, PhD9, Robert L. Nussbaum, MD10, Julianne M. O’Daniel, MS, CGC11, Kelly E. Ormond, MS,
CGC12, Heidi L. Rehm, PhD, FACMG12–13, Michael S. Watson, MS, PhD, FACMG14, Marc S.
Williams, MD, FACMG15, Leslie G. Biesecker, MD16

Genet Med. 2013 Jul;15(7):565-74. doi:
10.1038/gim.2013.73. Epub 2013 Jun 20.
Recommendations

• Constitutional mutations on minimal list should be reported regardless of age of patient
• Laboratories should seek and report specific types of mutations on list
• Ordering clinician responsible for pre- and post-test counseling
• Patients may opt out of learning about incidental findings
<table>
<thead>
<tr>
<th>Type</th>
<th>Genes</th>
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<tbody>
<tr>
<td>Tumor Predisposition (Breast/ovarian, Li-Fraumeni, Peutz-Jeghers, Lynch, FAP, Polyposis, Von Hippel-Lindau, MEN1/2, Medullary thyroid ca, PTEN hamartoma, retinoblastoma, Paraganglioma/Pheo, TSC, WT1-related Wilms’, NF2)</td>
<td>BRCA1, BRCA2, TP53, STK11, MLH1, MSH2, MSH6, PMS2, APC, MUTYH, VHL, MEN1, RET, NTRK1, PTEN, RB1, SDHD, SDHAF2, SDHC, SDHB, TSC1, TSC2, WT1, NF2</td>
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<td>Connective Tissue Dysplasia (EDS vascular type, Marfan, Loeys-Dietz, Familial thoracic and aortic aneurysms/dissections)</td>
<td>COL3A1, FBN1, TGFB1, TGFB2, SMAD3, ACTA2, MYLK, MYH11</td>
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<tr>
<td>Cardiomyopathy (Hypertropic, dilated)</td>
<td>MYBPC3, MYH7, TNNT2, TNNT3, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA</td>
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<tr>
<td>Arrhythmia (Arrhythmogenic RVCM, Romano-Ward, Brugada)</td>
<td>RYR2, PKP2, DSP, DSC2, TMEM43, DSG2, KCNQ1, DCNH2, SCN5A</td>
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<tr>
<td>Hypercholesterolemia</td>
<td>LDLR, APOB, PCSK9</td>
</tr>
<tr>
<td>Malignant hyperthermia</td>
<td>RYR1, CACNA1S</td>
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</tbody>
</table>
Revolutionising Biomedical Education

BEST - the Biomedical Education Skills and Training Network is a digitally-powered community of experts who share a vision: that every student and teacher will have access to the best biomedical education.

WATCH THE VIDEO

DISCIPLINARY LOOPS
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Search, annotate and collaborate on premium medical images with the world’s most advanced cloud-based biomedical image bank.

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JOIN NOW
MENDELIAN INHERITANCE

Resource Visibility: Public

Chapter 1: Basic Patterns of Mendelian Inheritance

Description

AUTHOR
Bruce Korf MD, PhD

DESCRIPTON
Learn about the patterns of single-gene inheritance.

Last reviewed: March 08, 2014.
Topics: Genetics
License Information: All Rights Reserved.
• Swipe badge in computer
• Assignment to
  • Hemochromatosis
  • Breast cancer
  • MEN 2
• Make counseling appointment
You are 30 years old and have been recently diagnosed with medullary thyroid carcinoma. Your medical history is otherwise unremarkable. Your endocrinologist tells you he would like to refer you to a genetic counselor because this particular type of thyroid cancer can run in families. You meet with a genetic counselor who gathers family history information.

The only potentially significant finding is some thyroid problem your mother had as a child, but records are not available. You have a healthy 2-year-old son and a 23-year-old brother. A geneticist does a physical exam, which she says is unremarkable. After a lengthy discussion, you decide to pursue testing of the RET gene and have your blood drawn. You arrange to return in four weeks to get the test results.
Compe**tency M3**

*Use the principles of genetic transmission, molecular biology of the human genome, and population genetics to infer and calculate risk of disease, to institute an action plan to mitigate this risk, to obtain and interpret family history and ancestry data, to order genetic tests, to guide therapeutic decision making, and to assess patient risk.*
ACMGF
Summer
Scholars
Program
How to know when physicians are ready for genomic medicine

Jason L. Vassy,1,2,3 Bruce B. Korf,4 Robert C. Green1,4

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 results might be prone to misinterpretation by nongeneticists 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 equal or greater 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 nonradiologic physicians ready to order computerized tomography (CT) scans for their patients and make clinical decisions based on their findings? As 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 genomic 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 (3). 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 diagnoses, prognosis, and therapies. 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: interdisciplinary 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
What will it take to integrate genomics into clinical workflow?

- Economics
- Quality and Outcomes
- Workforce