

# Virtual Mentor

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## MEDICAL EDUCATION

### Genetic and Genomic Competency in Medical Practice

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The principles of Mendelian and molecular genetics have long had a place in the preclinical medical curriculum, but clinical applications of genetics have been barely visible in the clinical training of medical students, residents, and postgraduates. Undoubtedly this absence reflected a perception that the focus of medical genetics is rare disorders, so most medical professionals could get by with minimal exposure to the discipline. Since completion of the sequencing of the human genome, the power of the genetic—and now the genomic—approach has increased enormously, providing new tools to diagnose and even treat both rare and common genetic conditions. Those at all levels of training must now gain competency in a complex and continually evolving area. I will try to set forth some principles that may be helpful in navigating this new area.

#### Principles

*1. Focus on competencies, not knowledge.* The genome can be a source of endless fascination—how is it possible to encode all of the information necessary for a human to develop in three billion bits of information that can be folded into a microscopic structure?—and the technology is dazzling—how is it possible to decode this information in a matter of days (soon, hours)? The practicing physician, however, is not going to be sequencing the genome or interpreting the raw data any more than he or she now analyzes raw data from blood work or MRI scans.

This leads us to the competencies—what should the physician be able to do using the genetic and genomic approach? Physicians need to be able to respond to an abnormal newborn screening result; to know when and how to arrange genetic testing and consultation to help establish a diagnosis; to obtain and interpret family histories so they can inform patients about risks and arrange for genetic testing or consultation to clarify that risk; to use pharmacogenetic testing to customize drug choice and dosage to an individual's physiological needs; and to interpret the results of genome-wide testing for risk of common disease.

Competencies in these areas have been defined for physicians at many levels. The Association of American Medical Colleges (AAMC) and Howard Hughes Medical Institute have tackled premedical and medical genetics education with broad competencies that leave a lot of room for faculty to add detail [1, 2]. The AAMC Medical School Objectives Project provided more detailed objectives in genetics [3], and the Association of Professors of Human and Medical Genetics objectives went into even finer detail [4]. The National Coalition of Health Professional Education in

Genetics [5] has developed core competencies in genetics for all health professionals and is developing a genetics curriculum. Detailed competencies have been written for the medical geneticist by the American College of Medical Genetics [6] (yes, it is possible to do a residency in medical genetics, recognized by the American Board of Medical Specialties and accredited by the Accreditation Council of Graduate Medical Education), guidelines have been proposed for internal medicine residency education [7], and the American Academy of Pediatrics has launched a Genetics in Primary Care Institute aimed at the continuum from residency to independent practice. All of these efforts, of course, must take account of the fact that genomics is quintessentially a moving target; most of the competency projects mentioned above predated the era of whole-exome/whole-genome sequencing, which has only been possible on a clinical basis for the past year or two yet is likely to become the mainstay of testing in the next few years.

Competencies are not acquired by attending lectures or reading books. These modes of instruction can provide a foundation, but competency is achieved by doing. To some extent, the road to competency may be paved by experiences in problem-based learning or simulation, but increasingly genetics and genomics will be incorporated into day-to-day teaching on inpatient and outpatient rotations for students and residents and postgraduate experiences for those in practice. There may be a need for immersion courses for practicing physicians to help them quickly acquire the basic skills necessary for incorporating genetics and genomics into their practices.

## *2. Learn to use point-of-care sources of information and decision support tools.*

Genetics and genomics deal fundamentally with information—indeed, the genome is the biological store of information necessary to build a functioning organism. With more than 20,000 genes and even more regulatory sequences, all of which interact in networks, the genetic data exceeds human processing capability. Just as it is impossible to fly a modern jet airplane without computer assistance, it is becoming impossible to practice medicine without the same. This is not to devalue the human interaction, which always has been and always will be the core of the medical encounter between physician and patient. Rather, it enriches that encounter by giving the physician an unprecedented store of information and tools to improve outcomes.

There are several online sources of crucial genetic information. Some are intended for use by medical geneticists, but others are useful to all practitioners. Online Mendelian Inheritance in Man [8] is the authoritative catalog of human genetic variants, including the clinical characteristics of associated disorders. GeneReviews [9] is an online compendium of indispensable peer-reviewed summaries of a wide variety of rare and common genetic disorders. Its parent site GeneTests [10] is a database of genetic testing laboratories. The new NIH-run Genetic Test Registry [11] is another database that provides information on laboratories that offer genetic tests.

Pharmacogenetic testing will increasingly be used to customize both drug choice and dosage [12]. Most likely, interpretations of test results will be embedded in electronic prescribing systems; physicians will understand that drug dosage may

need to be modified according to genotype, but the calculations are likely to occur behind the scenes. The role of genomic testing to determine risk of common diseases remains uncertain at present; so far, most genetic markers are only modestly predictive of disease risk. Nevertheless, some individuals are being tested for a million single nucleotide polymorphisms at a time (i.e., variations at a million DNA loci), in some cases on a direct-to-consumer basis [13]. Physicians must be able to respond to the results of these tests and help patients use the information wisely.

*3. Counteract misinformation about genetics and genomics.* Most people have at least an intuition about genetics—it's widely recognized that children take after their parents—yet are likely to have misconceptions. One is the notion of genetic determinism—that your destiny, at least regarding your health, is written in your genes. This may be more or less true for some conditions, such as cystic fibrosis or Huntington disease, but as we turn attention to more common multifactorial disorders, gene-environment interactions become more important and the ability to predict disease based on genetic testing less powerful. Moreover, even rare genetic conditions may be subject to modification by changes in lifestyle, environment, surgery, or medication. Medical therapies are being developed for a growing number of genetic disorders previously thought to be untreatable, and genetic testing is playing a major role in assessing familial risk of cancer [14] to provide approaches to risk reduction.

Accordingly, there is a second misconception that genetic information is inherently more sensitive than other types of medical information and requires correspondingly greater protection. This may be fueled by the notion that genetic testing can diagnose risk of disease in healthy people and that risks may apply to family members as well as to those being tested. Avoidance of genetic testing for fear of misuse of the information will deprive individuals of major potential benefits, which was the rationale for passing state and federal laws [15] to protect people from discrimination in employment or eligibility for health insurance based on their genetic information. At the same time, other kinds of test results, including those for risk factors such as cholesterol or infections that can be transmitted to close contacts, may be just as sensitive as genetic test results.

A third misconception is that genetic testing is always expensive and not covered by insurance. In fact, genetic testing varies widely in price and often may provide a shortcut in an otherwise very costly diagnostic odyssey. As with any medical procedure, it is always wise to check on a patient's specific insurance coverage, but many forms of diagnostic and predictive tests are covered with appropriate clinical indications.

### **Conclusions**

Some have complained that the benefits of sequencing the human genome were oversold and that medical applications have been slow to develop. The complexity of translating genetics and genomics information into usable knowledge should not be underestimated, but the pace of change in genomic medicine is accelerating. It cost

more than \$1 billion to sequence the first human genome in 2003 [16]; now a human genome can be sequenced for a few thousand dollars, and the cost is still falling. Clinical genome sequencing has already begun and is likely to be a mainstay of diagnostic testing within the next few years [17]. The era of genomic medicine has begun—our patients expect us to be competent in using this powerful approach to their benefit, and we must work now to insure that our trainees and professional colleagues are prepared for what is here now and what is to come.

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