

Genetic Literacy and Competency

abstract

Since the sequencing of the human genome was completed, progress toward understanding the genetic contributions to both rare and common disorders has accelerated dramatically. That understanding will lead to new approaches to diagnosis and management, which will be incorporated into day-to-day medical practice. Moreover, the mindset with regard to genetic contributions to health and disease has shifted from 1 gene at a time to genome wide. However, most practicing pediatricians, and even many still in training, are likely to be unfamiliar with the concepts of genetics and genomics and their applications in medical practice. This article addresses the issues of genetic and genomic literacy and competencies for pediatricians and other primary care providers, as they prepare to work with their patients in the emerging world of genomic medicine. *Pediatrics* 2013;132:S224–S230

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KEY WORDS

literacy, genetics, genomics

ABBREVIATIONS

AAMC—Association of American Medical Colleges

ACGME—Accreditation Council for Graduate Medical Education

ACMG—American College of Medical Genetics and Genomics

APHMG—Association of Professors of Human and Medical Genetics

HHMI—Howard Hughes Medical Institute

NCHPEG—National Coalition for Health Professional Education in Genetics

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“Health literacy” is defined as “the degree to which individuals have the capacity to obtain, process, and understand basic health information and services needed to make appropriate health decisions.”¹ The Institute of Medicine² and the Agency for Healthcare Research and Quality³ have undertaken major initiatives to increase health literacy. “Genetic literacy” has been defined as genetics knowledge as it relates to and affects the lives of individuals.⁴ Recent studies have suggested that the general public⁵ and health care providers⁶ have limited genetic literacy. For example, in a large survey of individuals with genetic conditions and their families, 64% of respondents reported having received no genetics education materials from the provider considered most important in the management of their condition.⁶ A recent survey of primary care physicians’ awareness, experience, opinions, and preparedness to answer questions about genetic testing revealed that only 15% of the 382 respondents felt prepared to discuss the topic with patients.⁷ Guttmacher et al⁸ summarized the attitudes of many providers regarding genetics as follows: “The Human Genome Project is interesting, and I know that it will change health care dramatically some day. But unless it will change my practice tomorrow in a concrete way, I really don’t have time to deal with it. What will genetics and genomics do for me now, and how will they improve patient outcomes?” In response to this question, we argue that urging primary care providers to learn numerous facts about genetic and genomic disease determinants and current tests that are marketed to identify them is unproductive. As whole-genome and whole-exome sequencing become more available and affordable, disease-specific tests will likely fall into disuse, and providers will be less likely to need to identify a probable diagnosis

before testing is done. This change will demand a new skill set from providers; either they will be called on to identify when genetic testing (eg, sequencing) is required, or they will need to access the already sequenced genome of a patient and query it for diagnoses related to the patient’s symptoms and other findings. The ability to collaborate with a genetics team and sensitively manage the uncertainty that may arise when risk factors, rather than diagnostic findings, are identified will be important competencies for the primary care provider.

The need to increase genetic literacy was highlighted by a report titled “Genetics Education and Training” published in 2011 by the Secretary’s Advisory Committee on Genetics, Health, and Society.⁹ The first recommendation of the report was as follows:

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.

GENETIC AND GENOMIC COMPETENCIES FOR PEDIATRIC PRIMARY CARE PROVIDERS

Competency-Based Training

Innovations in medical education over the past 2 decades have accelerated a movement toward emphasis on outcomes.¹⁰ Competency-based training, although not new in medical education,¹¹ has had a major impact in recent years on undergraduate, graduate, and continuing medical education

in all fields. The Accreditation Council for Graduate Medical Education (ACGME) and the American Board of Medical Specialties formalized the movement toward competency-based medical education in the late 1990s in the form of the Outcomes Project. This project identified 6 core competencies: patient care, medical knowledge, practice-based learning and improvement, interpersonal and communication skills, professionalism, and systems-based practice.¹² Since then, competency-based education in the health care professions has become a prominent approach to postgraduate training, as well as medical student training and continuing medical education, in the United States, Canada, and many other countries.

According to ten Cate et al,¹³ what matters in competency-based medical education is the outcome. Assessing the outcome, however, presents its own problems. A framework for assessing clinical skills was described by Miller,¹⁴ who envisioned education, and hence assessment of skills, as a pyramid. At the base, the learner first knows some information and then knows how to use that information, shows how to use it, and finally does it: that is, performs the acquired skills in actual clinical practice. Linking this framework to the 6 core competencies is difficult, however, because the competencies themselves are formulated as broad, general attributes of a good doctor.¹⁵ Thus, the competencies need to be placed in clinical context,¹⁵ and to do this, ten Cate et al¹⁵ suggested that important activities be identified for each specialty and that these activities then be linked to the general competencies. These activities encompass all the professional activities that a specific medical specialist can be asked to do, from executing an operational procedure to conveying bad news to chairing an interdisciplinary meeting; ten Cate et al¹⁵ labeled these activities

as “entrustable professional activities.”

Individuals and institutions continue to struggle to define how we can measure achievement in the competencies. To take the next steps in advancing outcome assessment, the ACGME and the American Board of Pediatrics defined benchmarks, or milestones of knowledge, skills, and attitudes, to document increasing mastery of the 6 core competencies.¹⁷ Subcompetencies were added to flesh out the activities that were needed to define the work of a pediatrician. Similar work has been done by other specialty organizations, including the American College of Medical Genetics and Genomics (ACMG).¹⁸ The American Board of Pediatrics is exploring the linkage of these subcompetencies, with their associated milestones, to the framework of entrustable professional activities.¹⁹

The education of medical professionals can be viewed as a vector, beginning with primary and secondary school education and continuing through undergraduate education, medical school, residency, and postgraduate education. Competencies in genetics and genomics have been considered for all these levels and can inform efforts to define competencies relevant to pediatrics.

Undergraduate Education and Medical School

Core curricula in genetics were set forth by the Association of Professors of Human and Medical Genetics (APHMG) in 1998²⁰ and by the Association of American Medical Colleges (AAMC) in 2004,²¹ the latter as a component of the Medical School Objectives Project. Neither of these curricula, however, was placed in a competency-based framework, and both predated the advent of genomic approaches, such as comparative genomic hybridization or whole-exome sequencing. Two more-recent efforts, however, do take a

competency-based approach and include genomics as well as genetics.

In 2009, a joint committee of the Howard Hughes Medical Institute (HHMI) and the AAMC issued a report entitled “Scientific Foundations for Future Physicians.”²² The committee approached the competencies from the perspective of an undergraduate premedical student as well as a medical student. Given the breadth of coverage, details regarding any specific area were necessarily high level. For undergraduates, 3 genetics-specific competencies were defined: (1) biochemical processes involved in information transfer in the cell, beginning with DNA; (2) principles of genetics and epigenetics; and (3) principles of evolution. For medical students, 1 major competency dealt with principles of genetic transmission, the human genome, and population genetics, with a goal of permitting students to determine disease risk, formulate a risk-mitigation plan, obtain and interpret a family health history, order genetic tests, and guide therapeutic planning. A major goal of the HHMI-AAMC joint initiative was to encourage matriculating medical students to be better prepared to achieve competency in modern scientific approaches relevant to medical practice, changing the traditional premedical areas of emphasis. Revision of the Medical College Admission Test, a critical component of this process, is currently under way. Similarly, the National Board of Medical Examiners has been exploring avenues to align the medical licensing examinations to this competency-based approach.

Following on the noncompetency-based core curricula described previously, the APHMG recently developed a competency-based core curriculum in genetics²³ intended to guide course directors in medical schools in the United States and Canada. The curriculum is mapped to the 6 ACGME

competencies and is based on a framework of 6 broad areas: (1) genome organization and regulation of gene expression; (2) genetic variation; (3) patterns of inheritance; (4) clinical, ethical, and social implications; (5) cancer and prenatal diagnostic issues; and (6) treatment and counseling.

National Coalition for Health Professional Education in Genetics Core Competencies

The National Coalition for Health Professional Education in Genetics (NCHPEG) has published core competencies in genetics for health professionals.²⁴ The NCHPEG core competencies were last updated in 2007. The competencies are divided into areas of knowledge, skills, and attitudes. Baseline competencies, including the need to continuously re-evaluate individual competency in genetics, recognize the ethical, legal, and social implications of genetics, and know when and how to work with professional genetics colleagues, were included as well. These competencies provide a framework that remains relevant to the needs of primary care providers but may fall short in terms of addressing new genomic approaches, including approaches based on whole-genome sequencing.

ACMG Competencies for the Physician Medical Geneticist

The ACMG has developed a set of competencies and learning objectives aimed at the physician medical geneticist.¹⁸ The approach is divided into 2 sections: overarching competencies and discipline-specific competencies. The former include taking and interpreting a family health history, assessing genetic risks, ordering and interpreting genetic and genomic tests, and counseling patients and families. The latter focus on areas dedicated to specific systems or specialties, such as prenatal diagnosis, biochemical

genetics, neurogenetics, and cardiovascular genetics. The competencies are, necessarily, aimed at the higher level of performance expected of a subspecialist and therefore do not serve as a direct model relevant to pediatric primary care providers, although the competencies do help define what a pediatrician can expect from a medical genetics consultant. The ACGME is now using these competencies as a basis for defining milestones to assess the progress of medical genetics residents in their training.

DEVELOPING GENETIC AND GENOMIC COMPETENCES FOR PEDIATRICS

Applicability of the NCHPEG Core Competencies

How useful are these lists of competencies in defining what the pediatrician should know and do about genetics and genomics? We agree with Miller¹⁴ that the apex of the educational period is in performing the acquired skill in clinical practice. Therefore genetic and genomic competencies should emphasize the clinical context, rather than knowledge for its own sake, and should direct the pediatrician to actions that will benefit patients and families.

It is not surprising that the core competencies and subcompetencies of pediatricians and geneticists have some elements in common, as well as differences. It is also no surprise that the genetic competencies for all health care providers, as defined by NCHPEG, overlap to a degree with these competencies for pediatricians and geneticists. The overlap with competencies and subcompetencies for pediatricians and geneticists as defined by their respective professional organizations provides some confidence that the NCHPEG competencies are in line with the thinking of the leadership of these

important specialty groups. For example, the NCHPEG list of core competencies in genetics states that all health professionals should understand the basic terminology of human genetics as well as basic patterns of biological inheritance and variation. The Pediatrics Milestone Project of the American Board of Pediatrics lists as a subcompetency of medical knowledge that pediatricians should “demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics.”¹⁷ These documents underscore the necessity for a basic understanding of the principles and terminology of human genetics.

The NCHPEG competencies include the following: “All health professionals should understand...the potential physical and/or psychosocial benefits, limitations, and risks of genetic information for individuals, family members, and communities.”²⁴ As a subcompetency of Personal and Professional Development, the Pediatric Milestones Project indicates that pediatricians should “recognize that ambiguity is part of clinical medicine and respond by utilizing appropriate resources in dealing with uncertainty.”

The NCHPEG competencies include the skill of gathering genetic family health history information, including a 3-generation history. This skill is not specifically mentioned in the Pediatric Milestones Project, although a subcompetency under patient care emphasizes the importance of family-unit correlates of disease in completion of the medical interview. If the pediatric primary care provider is to identify patients who would benefit from genome analysis or other diagnostic studies, skill at obtaining a family health history is a requirement.

An NCHPEG competency states that health care providers should be able to “effectively use new information technologies to obtain current information

about genetics.”²⁴ Although genetics and genomics are not specifically cited as important areas of knowledge in the Pediatric Milestones Project, the project includes subcompetencies involving the ability to critically evaluate and apply current medical information and scientific evidence for patient care, and the ability to use information technology to optimize learning and care delivery. Pediatricians will undoubtedly need to use information technology to access their patient's genomic data and to query it as patient findings, family history, or both change over time. Similarly, pediatricians will need to use information technology to keep abreast of new preventive and treatment strategies as they become available.

Comparison of these 3 lists of competencies reveals that only the pediatric competencies take into account a central challenge in genetic medicine as noted by McInerney in 2002²⁵: the management of uncertainty. As genomic data continue to accumulate, uncertainty about the number of genes involved in any given disease, about the roles and interactions of their protein products, and about the interactions of those products with the environment continues to increase. What is the nature of the susceptibility to disease conferred by these genes? What is the predictive power of genetic and genomic findings? McInerney²⁵ asks a fundamental question: What knowledge and skills does the average person require to manage this uncertainty and to participate as a full partner in a prevention-based health care system that is informed by genetic and genomic knowledge? We ask a related question: What special competencies are required by primary care providers to allow them to work with their patients and families to identify risk factors, order appropriate tests, make referrals, and participate in the

management of uncertainty that is likely to arise?

A Conceptual Structure for Core Competences in Genetics for Pediatricians

Efforts to develop core competences in genetics for any medical specialty must take account of the rapid advance of knowledge and technology in genetics and genomics. At a high level, however, 3 major domains may provide a conceptual framework to guide thinking about competency-based education in genetics for the future. These are applications of genetics in prevention, diagnosis, and management.

Prevention has long played a key role in pediatric genetics through the application of newborn screening, which for the most part involves early diagnosis and treatment of genetic disease. Recently, newborn screening has expanded beyond the diagnosis of a few inborn errors of metabolism, such as phenylketonuria, to the diagnosis of more than 2 dozen conditions.²⁶ Although it is important to diagnose such conditions early because they are treatable, many of them are extremely rare, and most pediatricians are unfamiliar with their management. The ACMG has developed a set of ACTION Sheets that are available online and provide basic information to guide the next steps of management upon receipt of notification of an abnormal newborn screen.²⁷ This approach may provide a model for instilling “just-in-time” competency for other rare conditions. The scope of prevention in pediatric genetics will likely expand in the coming years, as genomic tests enable early detection of variants that predict future disease.²⁸ We are still a long way from having established the clinical utility and predictive value of such testing, but at the least, pediatricians will need to be able to answer parents’ questions about tests, some of

which are marketed directly to the consumer. As tests mature and become available for routine clinical use, the pediatrician will need to appreciate the uses and limitations of genetics-based risk assessment.

Genetic diagnosis may be familiar in principle to pediatricians, but what may not be apparent is the degree to which testing technologies are evolving. The familiar cytogenetic testing has been replaced by cytogenomic microarray testing as a first line of evaluation for a child with intellectual disability, autism spectrum disorder, or congenital anomalies.²⁹ It is especially important for pediatricians to recognize that patients who had normal chromosomal analysis in the past might benefit from microarray testing now, because the detection rate for gene deletions and duplications is much higher for microarray testing than for cytogenetic analysis. Pediatricians also need to recognize the possibility of finding variants of unknown significance, as well as how to work with genetics professionals to further evaluate these variants and explain them to patients and their families. The power of molecular genetic testing is likewise increasing rapidly; the list of single-gene disorders amenable to molecular genetic testing has expanded greatly in recent years, but the clinical validity and clinical utility of these tests must be viewed critically.³⁰ As noted already, sequencing of the entire exome or genome is increasingly available clinically³¹; these approaches not only have the power to identify genetic mutations without previous knowledge of the gene in question but also have an increased likelihood of finding variants of unknown significance or mutations that predict future disease unrelated to the original indications for testing. Here, too, pediatricians will need to work with genetics

professionals to interpret such tests and counsel families.

Treatment of genetic disease is also evolving rapidly. For a long time, the only treatable genetic conditions were biochemical disorders, including those tested in newborn screening. Enzyme replacement therapy for lysosomal storage disorders has been available for some conditions for 2 decades, and the list of conditions treatable by this approach continues to grow.³² More recently, understanding of the molecular pathogenesis of many genetic conditions has opened the door to the development of targeted treatments. Although such approaches are in their early days, they are likely to proliferate in the years to come, permitting treatment of conditions traditionally viewed as untreatable. The pediatrician will need to recognize the potential of new treatments for patients whom they follow, referring them to specialists to initiate treatment and working as a partner in supporting the patient once treatment has begun. Improvements in management also mean an increasing likelihood of long-term survival, necessitating the eventual transition from pediatric to adult care.

These 3 domains address the continuum of pediatric care across the age spectrum and from rare to common conditions. Given the large number of specific disorders, no pediatrician is likely to gain competency in all these areas. More global competency in recognizing situations in which a child may benefit from genetic testing or treatment of a genetic disorder, and to work in partnership with genetics professionals to support the patient and family is, however, a realistic goal. Instilling competencies in these areas will require use of multiple educational modes at all levels of training, and especially the incorporation of point-of-care, just-in-time aids, such as ACTION

Sheets, perhaps embedded into electronic health records.

NEXT STEPS

Although we have no special platform from which to propose new competencies in genetics and genomics for any group of providers, we believe that delineation of genetics and genomics competencies for pediatricians should be considered by appropriate groups in the future. Using the genetic competencies that have already been defined by the NCHPEG, APHMG, and HHMI-AAMC, we propose a new and cooperative effort to reach a common understanding of the competencies that would allow providers to fulfill the needs and

expectations of patients and their families as the fields of genetics and genomics assume increasing importance in the general practice of medicine.

The rapid pace of discovery of new genetic and genomic knowledge makes it inevitable that some of today's facts will populate the wastebaskets of the future. Thus, the trend toward emphasizing lifelong learning in medical education is highly relevant to all providers as they grapple with the information revolution that is medicine in general, and genetics and genomics in particular. As Guttmacher et al⁸ noted in 2007, "If ever there were an area of medicine that is appropriate

for lifelong learning, it is genomics." Given the youth and rapid evolution of the field, it is not possible to identify either the knowledge or the clinical applications that will be commonplace when today's trainees and young clinicians are in mid-career. Therefore, Guttmacher et al⁸ argue that, in addition to providing tools that can be applied today, clinicians need to appreciate the future clinical importance of genomics, so that they are motivated to continue learning throughout their careers. They need to believe that genomic medicine will change outcomes for their patients, and they need to keep up with the new tools and concepts that are inevitable in a rapidly evolving field.

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