Competencies for the physician medical geneticist in the 21st century

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ertification for physician medical geneticists has been offered by the American Board of Medical Genetics since 1982 and has been recognized by the American Board of Medical Specialties since 1991. Since this time, major advances have been made in genetics and genomics that greatly increase the impact of the discipline in all areas of medicine. New opportunities include treatment of both rare and common genetic disorders, expanded newborn screening and carrier screening programs, testing for risk of cancer, and other adult-onset disorders, pharmacogenetic testing, and genomic analysis, including whole exome or genome sequencing. Despite these advances, the number of physicians who complete medical genetics training and achieve American Board of Medical Genetics certification has remained flat in recent years (Fig. 1). In part, this may be due to national trends, fueled by the high debt of graduating medical students and a consequent tendency of medical students to train in high-paying specialties. In part, it may be due to a relative lack of visibility of medical genetics to students at a time when they are making career decisions.

The medical genetics community has organized a set of meetings to discuss ways to increase the "pipeline" of medical genetics trainees. The first, held at the Banbury Conference Center in 2004,¹ proposed expansion of the scope of medical genetics training to encompass new areas of practice. The second, held at the Banbury Center in 2006,² considered the scope of practice of medical genetics given new advances in genetics and genomics. The latter meeting concluded with the suggestion that the medical genetics curriculum for training and maintenance of certification should be updated.

In response to that suggestion, a meeting was held on February 19–22, 2010, at Stone Mountain Conference Center, Atlanta, GA. Participants were asked to prepare a brief document outlining major areas of knowledge expected of the medical geneticist in a variety of topic areas and the 10 most common clinical presentations that might be seen by a medical geneticist. The group then used this material to compile the competencies and learning objectives, including overarching competencies and those specific to a given topic area.

The decision to focus on competencies rather than to produce a detailed list of knowledge, skills, and attitudes was made for several reasons. First, a comprehensive listing would be unwieldy and inevitably would be incomplete. Second, such a list would likely go out of date quickly. Third, it would seem prescriptive and might limit learning decisions made by faculty

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and trainees. Finally, formulation of competencies and learning objectives makes it clear not only what the medical geneticist should know but also what he or she should be able to do. This can be critical in a discipline that spans all of medicine and in which medical geneticists must interact with colleagues in other medical specialties.

The product of this meeting can be accessed at http:// www.acmg.net/StaticContent/Misc/ACMG_Competencies.pdf. The format of this document differs from the six core competencies used by the American Council of Graduate Medical Education to guide residency training (patient care, medical knowledge, practice-based learning and improvement, interpersonal communication skills, professionalism, and systemsbased practice). It was believed that the competencies expected of a medical geneticist would be clearer if organized according to the work activities specific to medical genetics. The competencies in this document can be mapped to the Accreditation Council of Graduate Medical Education (ACGME) six core competencies, which would be helpful for medical genetics residency program directors.

The document is divided into two sections: overarching competencies and discipline-specific competencies. Medical genetics encompasses essentially all of medicine; the overarching competencies define the history taking, risk assessment, physical examination, communication skills, etc., that span all areas of medical genetics. The discipline-specific competencies are organized by body system for the most part and identify skills that are specific to that area. This is not to imply that a trainee in medical genetics only needs to acquire competency in specialty areas of interest. A medical geneticist is expected to acquire all these competencies; organization into disciplinespecific areas provides an expedient way to organize a large volume of information.

Most of the discipline-specific sections include three types of competencies. First are things a medical geneticist should be able to do when functioning as a consultant. This includes things such as providing a differential diagnosis of genetic disorders and organizing and interpreting genetic testing. It is presumed that, in most cases, the geneticist will not have primary responsibility for the care of the patient but instead is helping to guide the genetic evaluation along with other providers. The second competency addresses disorders where the medical geneticist is likely to play a role in providing longitudinal management. The third competency addresses the interpretation of genomic tests for common disorders that fit within the discipline.

Two important areas that deserve special emphasis are genomic medicine and clinical trials. Whole exome sequencing, whole genome sequencing, array analysis, and genomic testing to assess risk of common disease are new to medicine and comprise critical areas of competency in modern medical genetics. Clinical trials also are a relatively new but vitally important area. The number of genetic disorders amenable to treatment is increasing rapidly as the pathophysiological mechanisms come to light. Medical geneticists have the understanding of natural history to drive the testing of new approaches to

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Fig. 1. Average number of new American Board of Medical Genetics (ABMG) diplomates in clinical genetics by year; the dip from 1996 to 2002 is attributable to the change in residency accreditation from ABMG to ACGME, with a temporary decline in the number of accredited programs.

therapy, but this requires expertise in conducting clinical trials. Not all residencies will be able to involve trainees in clinical trials, but all should be in the position to train residents in the basics of clinical research and at least in the principles of conducting clinical trials.

It is hoped that this document will help in the formulation of curricula for training of medical geneticists in ACGME-accredited residencies. It will be used as a template for the ACGME Medical Genetics Milestone Project, which is intended to identify points in the course of medical genetics training where specific competencies should be achieved. By virtue of the continuing rapid pace of advance in medical genetics, the competencies document will always be a work in progress. We welcome feedback, additions, and corrections, which can be addressed to acmg@acmg.net. Finally, addressing competencies for the physician medical geneticist is but the first step along a pathway to reexamine the training for medical genetics in general. A similar effort will soon be launched for laboratory geneticists, including biochemical, cytogenetics, and molecular genetics. Medical genetics is experiencing a period of exceedingly rapid change and is at the cutting edge of advances in medicine in general. This will require corresponding evolution of our approach to training that will permit our specialty to rise to new opportunities and challenges.

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