

The Medical Genetics
Residency Milestones

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BEHALF OF THE MEDICAL GENETICS MILESTONE
WORKING GROUP

Introduction

Medical genetics has been recognized as a distinct medical specialty by the American Board of Medical Specialties since 1991, and as such, medical genetics residency programs are accredited by the Accreditation Council for Graduate Medical Education (ACGME). As a component of the ACGME’s Next Accreditation System (NAS), a process was initiated in 2011 to formulate Milestones for medical genetics residents. As with Milestone projects in other accredited specialties, the intent was to provide a system for evaluation of resident performance over the course of training, which also would serve as an indicator of the educational effectiveness of residency programs.

The project was initiated at a propitious time in the history of medical genetics, as the discipline is experiencing a period of rapid expansion since the sequencing of the human genome. In 2011, the American College of Medical Genetics (ACMG) published a document, “Competencies for the Physician Geneticist in the 21st Century,” that was intended to help prepare the medical genetics community for the new era of genomic medicine.¹ This document served as a template to guide the efforts of the Medical Genetics Milestone Project.

Milestone Development History

The Medical Genetics Milestone Project was jointly sponsored by the ACGME Residency Review Committee (RRC) in Medical Genetics and the American Board of Medical Genetics (ABMG). The project was chaired by the lead author (B.R.K.), and included 2 committees: a working group, which was charged with developing the Milestones, and an advisory group, which was charged with reviewing the Milestones and providing suggestions to the working group. A full list of members of both committees is shown in the BOX. The working group met face-to-face on 3 occasions, and also met several times by teleconference. The advisory group met twice by teleconference, once about midway through the process and again when a draft of the Milestones was completed. Progress

BOX	MILESTONE DEVELOPMENT GROUPS
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reports were given at the annual Association of Professors of Human and Medical Genetics (APHMG) meeting in 2011, 2012, and 2013; this group includes a medical genetics residency directors group. APHMG members were shown a working draft of the document in 2012 and feedback was provided prior to the final meeting of the working group. The final draft was shared with residency directors at the APHMG meeting in 2013. This allowed residency directors and other members of the genetics community to provide feedback to the working group that resulted in a number of changes to the document. Drafts were also presented to the board of directors of the ACMG and ABMG for feedback.

The Medical Genetics Milestone Project was immediately preceded by publication of the ACMG competencies document. The latter document was intended to codify the competencies and learning objectives to guide medical genetics training and curriculum development and provided a ready template to guide the Medical Genetics Milestones. The ACMG competencies are not formulated according to the ACGME 6 competencies, but it was relatively easy to group them according to this framework. Medical genetics residency programs involve 2 years of training, and

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TABLE ORGANIZATION OF THE MEDICAL GENETICS MILESTONES		
Core Competency	Number of Milestones	Milestones
Medical Knowledge	2	- Apply knowledge of anatomy, development, pathophysiology, natural history, clinical history, and inheritance to provide counseling, anticipatory guidance, and longitudinal management to patients with multisystem genetic disorders - Assess and participate in a clinical or translational research study or clinical trial involving patients with or at risk for a genetic disorder
Patient Care	9	- Obtain and interpret medical, social, and family histories, as well as physical examination findings necessary for the evaluation of patients with or at risk for genetic disorders - Incorporate genetic tests into patient management - Incorporate whole genome or exome tests into patient management - Diagnose and manage patients with inborn errors of metabolism - Evaluate infants with abnormal newborn screens in a cost-effective and sensitive manner and educate community providers - Develop proficiency in cancer genetics - Evaluate and manage patients with single malformations, multiple congenital anomalies, developmental disabilities, and growth abnormalities by utilizing knowledge of embryology, teratology, developmental pathways, pathophysiology, and etiologic mechanisms - Develop proficiency in prenatal risk assessment, screening, diagnosis, and counseling - Provide longitudinal management and reproductive counseling in pregnancies with known or suspected genetic conditions in the mother or fetus
Professionalism	3	- Is sensitive and responsive to diverse patient populations with respect to gender, age, culture, race, religion, disabilities, and sexual orientation - Adhere to the ethical principles relevant to the practice of medicine - Demonstrate personal responsibility to maintain emotional, physical, and mental health and accountability to patients, society, and the profession
Interpersonal and Communication Skills	2	- Relationship building, teamwork, and conflict management - Information gathering and sharing
Practice-Based Learning and Improvement	2	- Self-directed learning - Implement a quality improvement project
Systems-Based Practice	2	- Function effectively within the larger context of health care systems, and practice cost-effective medicine - Use technology to accomplish safe health care delivery

Note: This TABLE presents the ACGME core competencies, and the number of Medical Genetics Milestones in each area, and headings of specific Milestones.

residents enter medical genetics training after completion of at least 2 years in an ACGME-accredited residency, and enter medical genetics training with widely varying levels of experience in medical genetics. Several programs also offer joint 5-year programs with pediatrics, internal medicine, or maternal-fetal medicine.

As a result of these variations in training and prior experience with medical genetics, the working group chose not to specify time-dependent Milestone levels. Level 1 corresponds with an entry-level resident, for whom basic background competency in medicine is expected. Levels 2 and 3 represent an evolution from a resident who requires substantial supervision to one who can be expected to complete most tasks with minimal supervision. A resident is expected to have achieved Level 4 competencies upon

graduation. Level 5 competency represents a very high level of achievement that some residents might attain in some specific area, although it is unlikely that a resident would achieve level 5 competency in many areas and extremely unlikely that any resident would achieve this in all areas of medical genetics.

General Features of the Specialty Milestones

The TABLE in this Introduction provides a list of the 20 Medical Genetics Milestones distributed among the 6 ACGME competencies. Milestones in medical knowledge and patient care are more or less unique to medical genetics and constitute the core of medical practice in genetics. The ACMG competencies document was used as a starting point for formulation of the 11 Milestones in these areas. Domains of professionalism, interpersonal and communi-

cation skills, practice-based learning and improvement, and systems-based practice include some areas that are unique to medical genetics and others that are similar to other disciplines. Most of the Milestones in these areas were adopted from suggested language used in other disciplines, but some were customized to specific issues in medical genetics. For example, within interpersonal and communication skills, the information gathering and sharing Milestone includes recognition of the role of other health professionals, including genetic counselors, and the need to be sensitive to cultural issues in communication of genetic information.

Establishing Milestone Validity, Utility, and Practicality

The Medical Genetics Milestones represent a starting point for a process of objective measurement of validity and utility and future adjustments to ensure the goal of training medical geneticists who are competent to practice in the modern and ever-changing field of genomic medicine. Using the construct for assessment of validity of psychometric instruments proposed by Cook and Beckman, which encompasses the following dimensions.²

- **Content Validity:** The Medical Genetics Milestones were based largely on the ACMG competencies document, which was formulated by consensus among practitioners in the discipline. Further assessment of the scope of practice of medical geneticists based on surveys and field analysis of actual practice would improve the relevance of Milestones to real-world practice.
- **Response Process:** It will be important for residency program directors to be familiar with the use of the Milestones and to be able to use them consistently and reliably. Pilot programs have been selected to begin implementation and training was provided at the 2013 APMG residency program directors group meeting. Ongoing training and assessment of use of the Milestones will be necessary as residency program directors change and as information is acquired as to which Milestones tend to be most difficult to achieve.
- **Internal Structure:** The Medical Genetics Competency Committee will have the opportunity to view meta-data from individual programs over time as well as from multiple programs over time. These data should inform the committee regarding Milestones that may be ambiguously worded or inconsistently achieved so that improvements can be made.
- **Relations to Variables:** There are 2 major opportunities to assess the Milestones in relation to existing assessments: the medical genetics residency

in-service examination,³ and the ABMG medical genetics certification examination. Both are administered by the National Board of Medical Examiners, and, as such, are monitored for performance and validity. Comparison of program outcomes with these measures will be important information for the Medical Genetics Competency Committee and the RRC to consider.

- **Consequences:** It will be important to monitor outcomes for residents who do not achieve Milestones (ie, whether appropriate remediation is undertaken), and also for programs that have multiple residents requiring remediation or that do not consistently use the Milestones, to ensure that the RRC uses these data in the accreditation process.

Planned efforts to validate the Medical Genetics Milestones include pilot testing of the Milestones in actual residencies to generate added information on their usability, validity, and value.

Envisioned Practical Use in Evaluating Residents

The working group hopes that incorporation of the Milestones into medical genetics residencies will be useful in guiding residents to understand what is expected of them; in identifying residents who are having difficulties and in instituting remediation; and in documenting competency in medical geneticists ready to enter unsupervised practice. There are also “reach” goals that will help to motivate residents to achieve high-level competency and contribute to the future development of the discipline.

The Milestones will provide a consistent metric for the medical genetics RRC to assess the success of medical genetics residency programs in training and identify programs that need improvement or have achieved distinction.

Recommendations for Competency Committee Composition and Functioning

Medical genetics is one of the fastest changing of medical disciplines. This is particularly evident in the rapid adoption of whole exome or genome sequencing into clinical practice, which was just beginning as the Milestones were formulated. New opportunities for treatment of genetic disorders are also rapidly emerging. It will therefore be necessary to continually revisit and update the competencies to ensure that they reflect current practice.

A Medical Genetics Competency Committee that is similar in size and composition to the working group, that includes representation of major constituencies, such as prenatal diagnosis, pediatric genetics, and adult genetics, and includes representation of residents, will be needed to

accomplish this goal. The committee can also monitor overall success of residents in programs in meeting the Milestones to identify areas in which goals have been set either too high or too low.

Conclusion

The Medical Genetics Milestones were formulated with the goals of providing more objective assessment of medical genetics training to serve residents, program directors, the RRC, and the public. These should be viewed as a starting point to ensure the quality of medical genetics training and

will undoubtedly evolve with additional information on implementation and also on expected further advances in medical genetics.

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