

The Medical Genetics Milestone Project

The Milestones provide a framework for assessment of the development of the resident physician in key dimensions of the elements of physician competency in a specialty or subspecialty. The Milestones are designed only for use in evaluation of resident physicians in the context of their participation in Accreditation Council for Graduate Medical Education (ACGME)–accredited residency or fellowship programs. They neither represent the entirety of the dimensions of the 6 domains of physician competency, nor are they designed to be relevant in any other context.

Milestone Reporting

This document presents Milestones designed for programs to use in semiannual review of resident performance and reporting to the ACGME. Milestones are knowledge, skills, attitudes, and other attributes for each of the ACGME competencies organized in a developmental framework from less to more advanced. They are descriptors and targets for resident performance as a resident moves from entry into residency through graduation. In the initial years of implementation, the Review Committee will examine Milestone performance data for each program's residents as 1 element in the Next Accreditation System (NAS) to determine whether residents overall are progressing.

For each period, review and reporting will involve selecting Milestone levels that best describe a resident's current performance and attributes. Milestones are arranged into numbered levels. Tracking from Level 1 to Level 5 is synonymous with moving from novice to expert. These levels do not correspond with postgraduate year of education.

Selection of a level implies that the resident substantially demonstrates the Milestones in that level, as well as those in lower levels (see the **FIGURE**).

Acknowledgements: The Medical Genetics Working Group would like to acknowledge the American College of Medical Genetics and Genomics and their report "Competencies for the Physician Medical Geneticist in the 21st Century." This document was beneficial to the development of the Medical Genetics Milestones.

The Medical Genetics Advisory Group and the ACGME would like to honor Dr. Dave Rimoin for his contribution to the Milestones. He will be greatly missed.

DOI: <http://dx.doi.org/10.4300/JGME-06-0151-32>

Copyright © 2013 Accreditation Council for Graduate Medical Education and American Board of Medical Genetics. All rights reserved. The copyright owners grant third parties the right to use the Medical Genetics Milestones on a nonexclusive basis for educational purposes.

Level 1: The resident demonstrates Milestones expected of an incoming resident.

Level 2: The resident is advancing and demonstrates additional Milestones, but is not yet performing at a midresidency level.

Level 3: The resident continues to advance and demonstrate additional Milestones, consistently including the majority of Milestones targeted for residency.

Level 4: The resident has advanced so that he or she now substantially demonstrates the Milestones targeted for residency. This level is designed as the graduation target.

Level 5: The resident has advanced beyond performance targets set for residency and is demonstrating aspirational goals which might describe the performance of someone who has been in practice for several years. It is expected that only a few exceptional residents will reach this level.

Additional Notes

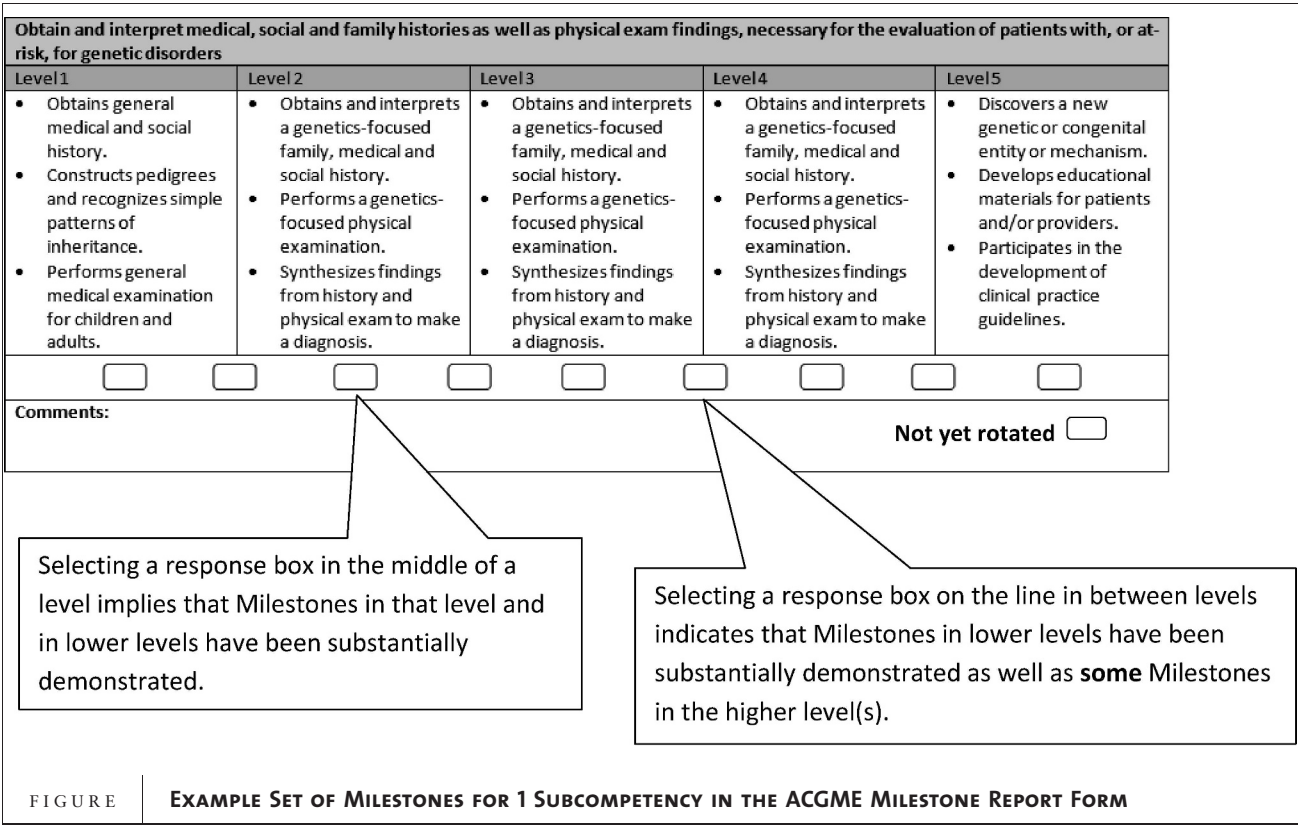
Level 4 is designed as the graduation *target* and *does not* represent a graduation *requirement*. Making decisions about readiness for graduation is the purview of the residency program director. Study of Milestone performance data will be required before the ACGME and its partners will be able to determine whether Milestones in the first 4 levels appropriately represent the developmental framework, and whether Milestone data are of sufficient quality to be used for high-stakes decisions.

Some Milestone descriptions include statements about performing independently. These activities must occur in conformity to ACGME supervision guidelines, as well as institutional and program policies. For example, a resident who performs a procedure independently must, at a minimum, be supervised through oversight.

To aid in scoring the Milestones, a listing of assessment tools by competency is available. These assessment tools are not required.

ACGME Milestone Report Form

The **FIGURE** presents an example set of Milestones for 1 subcompetency in the same format as the Milestone Report Form. For each reporting period, a resident's performance on the Milestones for each subcompetency will be indicated by:



- selecting the level of Milestones that best describes that resident’s performance in relation to the Milestones, or
- for Patient Care and Medical Knowledge Milestones, selecting the option that says the resident has “Not yet rotated,” or
- for Interpersonal and Communication Skills, Practice-Based Learning and Improvement, Professionalism, and Systems-Based Practice Milestones, selecting the option that says the resident has “Not yet achieved Level 1.”

MEDICAL GENETICS MILESTONES

PATIENT CARE					
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—Patient Care 1					
Level 1	Level 2	Level 3	Level 4	Level 5	
- Obtains general medical and social history - Constructs pedigrees and recognizes simple patterns of inheritance - Performs general medical examination for children and adults	- Obtains and interprets a genetics-focused family, medical, and social history with substantial guidance - Performs a genetics-focused physical examination with substantial guidance - Synthesizes findings from history and physical examination to make a diagnosis with substantial guidance	- Obtains and interprets a genetics-focused family, medical, and social history with minimal guidance - Performs a genetics-focused physical examination with minimal guidance - Synthesizes findings from history and physical examination to make a diagnosis with minimal guidance	- Independently obtains and interprets a genetics-focused family, medical, and social history - Independently performs a genetics-focused physical examination - Independently synthesizes findings from history and physical examination to make a diagnosis	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines	
Incorporate genetic tests into patient management—Patient Care 2					
Level 1	Level 2	Level 3	Level 4	Level 5	
- Distinguishes different types of genetic tests - Recognizes different types of genetic variants - Recognizes the distinction between analytical validity, clinical validity, and clinical utility - Recognizes the ethical, legal, social, and economic issues associated with genetic tests	- Selects and prioritizes tests with substantial guidance - Describes analytical techniques used in genetic testing with substantial guidance - Interprets laboratory reports with substantial guidance - Utilizes tools to assess variant of uncertain significance (VUS) with substantial guidance - Provides pretest and posttest counseling with substantial guidance	- Selects and prioritizes tests with minimal guidance - Describes analytical techniques used in genetic testing with minimal guidance - Interprets laboratory reports with minimal guidance - Utilizes tools to assess VUS with minimal guidance - Provides pretest and posttest counseling with minimal guidance	- Independently selects and prioritizes tests - Independently describes analytical techniques used in genetic testing - Independently interprets laboratory reports - Independently utilizes tools to assess VUS - Independently provides pretest and posttest counseling	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines	

Incorporate whole genome or exome tests into patient management—Patient Care 3				
Level 1	Level 2	Level 3	Level 4	Level 5
Explains differences between genetic and genomic analysis	- Identifies clinical situations where genomic testing is appropriate with substantial guidance - Provides pretest and posttest counseling with substantial guidance - Interprets and applies the information obtained from genomic testing with substantial guidance	- Identifies clinical situations where genomic testing is appropriate with minimal guidance - Provides pretest and posttest counseling with minimal guidance - Interprets and applies the information obtained from genomic testing with minimal guidance	- Independently identifies clinical situations where genomic testing is appropriate - Independently provides pretest and posttest counseling - Independently interprets and applies the information obtained from genomic testing	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines
Diagnose and manage patients with inborn errors of metabolism—Patient Care 4				
Level 1	Level 2	Level 3	Level 4	Level 5
- Demonstrates ability to manage critically ill patients - Recognizes basic abnormalities seen on metabolic testing	- Identifies signs and symptoms of inborn errors of metabolism with substantial guidance - Formulates a differential diagnosis and orders appropriate tests with substantial guidance - Recognizes normal variants and complex patterns of metabolites with substantial guidance - Manages acute metabolic crises with substantial guidance - Provides chronic management with substantial guidance	- Identifies signs and symptoms of inborn errors of metabolism with minimal guidance - Formulates a differential diagnosis and orders appropriate tests with minimal guidance - Recognizes normal variants and complex patterns of metabolites with minimal guidance - Manages acute metabolic crises with minimal guidance - Provides chronic management with minimal guidance	- Independently identifies signs and symptoms of inborn errors of metabolism - Independently formulates a differential diagnosis and orders appropriate tests - Independently recognizes normal variants and complex patterns of metabolites - Independently manages acute metabolic crises - Independently provides chronic management	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines

Evaluate infants with abnormal newborn screens in a cost-effective and sensitive manner and educate community providers—Patient Care 5				
Level 1	Level 2	Level 3	Level 4	Level 5
Describes goals and principles underlying newborn screening (NBS)	- With substantial guidance interprets NBS results - With substantial guidance collaborates with the primary provider to act upon results in a timely manner - With substantial guidance develops a differential diagnosis and orders appropriate confirmatory testing - With substantial guidance communicates results to families	- With minimal guidance interprets NBS results - With minimal guidance collaborates with the primary provider to act upon results in a timely manner - With minimal guidance develops a differential diagnosis and orders appropriate confirmatory testing - With minimal guidance communicates results to families	- Independently interprets NBS results - Independently collaborates with the primary provider to act upon results in a timely manner - Independently develops a differential diagnosis and orders appropriate confirmatory testing - Independently communicates results to families	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines
Develop proficiency in cancer genetics—Patient Care 6				
Level 1	Level 2	Level 3	Level 4	Level 5
- Recognizes that cancer is the accumulation of genetic changes, some of which can be inherited - Recognizes the use of germ line and somatic genetic changes in risk assessment and management of cancer - Identifies potential high-risk genetic pedigrees for common malignancies	- With substantial guidance assesses and manages genetic risk of common cancers - With substantial guidance diagnoses and manages inherited cancer susceptibility syndromes - With substantial guidance manages developmental disorders associated with increased risk of cancers - With substantial guidance evaluates the results of somatic genetic and genomic testing, and incorporates them into the management plan	- With minimal guidance assesses and manages genetic risk of common cancers - With minimal guidance diagnoses and manages inherited cancer susceptibility syndromes - With minimal guidance manages developmental disorders associated with increased risk of cancers - With minimal guidance evaluates the results of somatic genetic and genomic testing, and incorporates them into the management plan	- Independently assesses and manages genetic risk of common cancers - Independently diagnoses and manages inherited cancer susceptibility syndromes - Independently manages developmental disorders associated with increased risk of cancers - Independently evaluates the results of somatic genetic and genomic testing, and incorporates them into the management plan	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines

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—Patient Care 7				
Level 1	Level 2	Level 3	Level 4	Level 5
- Recognizes normal physical findings and patterns of growth and development - Accurately defines categories of birth defects utilizing principles of embryology	- Performs assessment incorporating physical findings and patterns of growth and development to establish diagnoses with substantial guidance - Implements a management plan with substantial guidance	- Performs assessment incorporating physical findings and patterns of growth and development to establish diagnoses with minimal guidance - Implements a management plan with minimal guidance	- Independently performs assessment incorporating physical findings and patterns of growth and development to establish diagnoses - Independently implements a management plan	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines
Develop proficiency in prenatal risk assessment, screening, diagnosis, and counseling—Patient Care 8				
Level 1	Level 2	Level 3	Level 4	Level 5
- Demonstrates knowledge of normal embryologic development - Recognizes the importance of family history, ethnicity, parental age, aneuploidy screening, teratogen exposure, and ultrasound findings in the assessment of fetal risk - Describes goals and principles underlying prenatal screening	- Performs risk assessment and provides pretest counseling with substantial guidance - Interprets prenatal screening results with substantial guidance - Collaborates with obstetric provider to act upon results in a timely manner with substantial guidance - Develops a differential diagnosis and recommends a diagnostic approach with substantial guidance - Provides posttest counseling with substantial guidance	- Performs risk assessment and provides pretest counseling with minimal guidance - Interprets prenatal screening results with minimal guidance - Collaborates with obstetric provider to act upon results in a timely manner with minimal guidance - Develops a differential diagnosis and recommends a diagnostic approach with minimal guidance - Provides posttest counseling with minimal guidance	- Independently performs risk assessment and provides pretest counseling - Independently interprets prenatal screening results - Independently collaborates with obstetric provider to act upon results in a timely manner - Independently develops a differential diagnosis and recommends a diagnostic approach - Independently provides posttest counseling	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines

Provide longitudinal management and reproductive counseling in pregnancies with known or suspected genetic conditions in the mother or fetus—Patient Care 9				
Level 1	Level 2	Level 3	Level 4	Level 5
- Recognizes the importance of counseling during the transition of care from prenatal to postnatal life - Recognizes the importance of a postpregnancy management plan upon establishment of a diagnosis - Recognizes the implications of maternal genetic disease for pregnancy management	- Collaborates with the obstetric provider to develop a management plan for pregnant women with genetic disorders with substantial guidance - Collaborates with the multidisciplinary team to develop a management plan for pregnant women with fetuses with genetic disorders with substantial guidance - Provides postpregnancy counseling and discussion of future reproductive options with substantial guidance	- Collaborates with the obstetric provider to develop a management plan for pregnant women with genetic disorders with minimal guidance - Collaborates with the multidisciplinary team to develop a management plan for pregnant women with fetuses with genetic disorders with minimal guidance - Provides postpregnancy counseling and discussion of future reproductive options with minimal guidance	- Independently collaborates with the obstetric provider to develop a management plan for pregnant women with genetic disorders - Independently collaborates with the multidisciplinary team to develop a management plan for pregnant women with fetuses with genetic disorders - Independently provides postpregnancy counseling and discussion of future reproductive options	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines
MEDICAL KNOWLEDGE				
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—Medical Knowledge 1				
Level 1	Level 2	Level 3	Level 4	Level 5
- Formulates a care plan that is incomplete - Recognizes the uses and limitations of practice guidelines	- Formulates and prioritizes a care plan with substantial guidance - Applies practice guidelines based upon patient's needs, genotype, and family history with substantial guidance	- Formulates and prioritizes a care plan with minimal guidance - Applies practice guidelines based upon patient's needs, genotype, and family history with minimal guidance	- Independently formulates and prioritizes a care plan - Independently applies practice guidelines based upon patient's needs, genotype, and family history	- Makes a nationally recognized contribution by discovering a new genetic or congenital entity or mechanism - Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines

Assess and participate in a clinical or translational research study or clinical trial involving patients with or at risk for a genetic disorder—Medical Knowledge 2				
Level 1	Level 2	Level 3	Level 4	Level 5
▪ Completes training in protection of human subjects ▪ Demonstrates knowledge of the principles of research studies and clinical trials	▪ Identifies research studies or clinical trials appropriate for management with substantial guidance ▪ Facilitates enrollment of patients in research studies with substantial guidance ▪ Addresses the ethical issues that are specific to genetic and genomic studies and clinical trials with substantial guidance	▪ Identifies research studies or clinical trials appropriate for management with minimal guidance ▪ Facilitates enrollment of patients in research studies with minimal guidance ▪ Addresses the ethical issues that are specific to genetic and genomic studies and clinical trials with minimal guidance	▪ Independently identifies research studies or clinical trials appropriate for management ▪ Independently facilitates enrollment of patients in research studies ▪ Independently addresses the ethical issues that are specific to genetic and genomic studies and clinical trials	▪ Makes a nationally recognized contribution by conducting a clinical research study or clinical trial
SYSTEMS-BASED PRACTICE				
Function effectively within the larger context of health care systems, and practice cost-effective medicine—Systems-Based Practice 1				
Level 1	Level 2	Level 3	Level 4	Level 5
▪ Recognizes how health care systems influence individual practice and patient care ▪ Sensitive to cost-effectiveness of care ▪ Recognizes that diagnosis and management have implications for care at home and in the community across the lifespan of the patient	▪ Functions effectively within different systems with substantial guidance ▪ Incorporates and advocates for genetics services to enhance cost-effectiveness of care with substantial guidance ▪ Recognizes and manages the variation in access to genetic testing with substantial guidance ▪ Facilitates management and transitions of care teams as the patient ages with substantial guidance ▪ Participates in identifying system errors and implementing potential system solutions with substantial guidance	▪ Functions effectively within different systems with minimal guidance ▪ Incorporates and advocates for genetics services to enhance cost-effectiveness of care with minimal guidance ▪ Recognizes and manages the variation in access to genetic testing with minimal guidance ▪ Facilitates management and transitions of care teams as the patient ages with minimal guidance ▪ Participates in identifying system errors and implementing potential system solutions with minimal guidance	▪ Independently functions effectively within different systems ▪ Independently incorporates and advocates for genetics services to enhance cost-effectiveness of care ▪ Independently recognizes and manages the variation in access to genetic testing ▪ Independently facilitates management and transitions of care teams as the patient ages ▪ Independently participates in identifying system errors and implementing potential system solutions	▪ Leads systems change

Use technology to accomplish safe health care delivery—Systems-Based Practice 2				
Level 1	Level 2	Level 3	Level 4	Level 5
- Utilizes the electronic health record and computerized physician order entry in prevention of medical errors - Recognizes the risk added by copy/paste strategies to create notes	- Documents essential elements of genetics encounters to enhance the transfer of information and patient safety with substantial guidance - Utilizes decision support tools with substantial guidance	- Documents essential elements of genetics encounters to enhance the transfer of information and patient safety with minimal guidance - Utilizes decision support tools with substantial guidance	- Independently documents essential elements of genetics encounters to enhance the transfer of information and patient safety - Independently utilizes decision support tools	Develops new technologies to improve health care in genetics
PRACTICE-BASED LEARNING AND IMPROVEMENT				
Self-directed learning—Practice-Based Learning and Improvement 1				
1. Identify strengths, deficiencies, and limits in one's knowledge and expertise 2. Set learning and improvement goals by seeking and incorporating feedback 3. Identify and perform appropriate learning activities 4. Use information technology to optimize learning 5. Critically read and interpret scientific literature				
Level 1	Level 2	Level 3	Level 4	Level 5
- Acknowledges gaps in personal knowledge and expertise - Demonstrates computer literacy and basic computer skills	- Assesses self-performance and develops a learning plan with substantial guidance - Critically evaluates and utilizes information from diverse sources with substantial guidance	- Assesses self-performance and develops a learning plan with minimal guidance - Critically evaluates and utilizes information from diverse sources with minimal guidance	- Independently assesses self-performance and develops a learning plan - Independently critically evaluates and utilizes information from diverse sources	Develops nationally recognized materials and approaches for education of others
Implement a quality improvement project—Practice-Based Learning and Improvement 2				
Level 1	Level 2	Level 3	Level 4	Level 5
Recognizes the value of quality improvement	Describes elements of a quality improvement project	Identifies areas for improvement in individual practice	Participates in a quality improvement project	Designs and leads quality improvement project
PROFESSIONALISM				
Is sensitive and responsive to diverse patient populations with respect to gender, age, culture, race, religion, disabilities, and sexual orientation—Professionalism 1				
Level 1	Level 2	Level 3	Level 4	Level 5
Understands the importance of caring, honest, and sensitive behavior	Demonstrates caring, honest, and sensitive behavior in common and uncomplicated situations	Demonstrates caring, honest, and sensitive behavior in complex situations	Demonstrates caring, honest, and sensitive behavior in all relationships and situations	Demonstrates leadership and mentoring regarding these principles

Adhere to the ethical principles relevant to the practice of medicine—Professionalism 2					
Level 1	Level 2	Level 3	Level 4	Level 5	
- Is aware of basic bioethical principles and their importance in patient care - Demonstrates respect for patient privacy and autonomy	- Effectively manages personal beliefs to avoid any negative impact on patient care with substantial guidance - Recognizes and manages ethical issues in genetics practice with substantial guidance	- Effectively manages personal beliefs to avoid any negative impact on patient care with minimal guidance - Recognizes and manages ethical issues in genetics practice with minimal guidance	- Independently effectively manages personal beliefs to avoid any negative impact on patient care - Independently recognizes and manages ethical issues in genetics practice	- Makes a nationally recognized contribution by developing educational materials for patients and/or providers - Makes a nationally recognized contribution by participating in the development of clinical practice guidelines	
Demonstrate personal responsibility to maintain emotional, physical, and mental health and accountability to patients, society, and the profession—Professionalism 3					
Level 1	Level 2	Level 3	Level 4	Level 5	
- Recognizes the importance of maintaining personal emotional, physical, and mental health - Maintains professional appearance - Is aware of the issues related to fatigue and sleep deprivation	- Demonstrates adequate management of personal, emotional, physical, and mental health - Effectively manages the issues related to fatigue and sleep deprivation - Recognizes limits of knowledge and asks for assistance - Completes professional responsibilities in a timely manner	Identifies and manages situations in which maintaining personal emotional, physical, and mental health is challenged by common and typical clinical care situations	- Identifies and manages situations in which maintaining personal emotional, physical, and mental health is challenged by complicated and less frequent clinical care situations - Recognizes signs of physician impairment and demonstrates appropriate steps to address impairment in colleagues	Develops institutional and organizational strategies to improve physician performance	

INTERPERSONAL AND COMMUNICATION SKILLS					
Relationship building, teamwork, and conflict management—Interpersonal and Communication Skills 1					
Level 1	Level 2	Level 3	Level 4	Level 5	
- Develops a positive relationship based on mutual trust with patients/families in uncomplicated situations - Recognizes conflicts, especially those that pertain to patient care	- Develops and sustains relationships with families and health care team members with substantial guidance - Manages conflicts with patients/families and within the health care team with substantial guidance - Participates in team-based care with substantial guidance - Joins professional organizations	- Develops and sustains relationships with families and health care team members with minimal guidance - Manages conflicts with patients/families and within the health care team with minimal guidance - Participates in team-based care with minimal guidance - Participates in regional and national organizations	- Independently develops and sustains relationships with families and health care team members - Independently manages conflicts with patients/families and within the health care team - Independently participates in team-based care	- Develops models/approaches to managing difficult communications - Assumes leadership roles within professional organizations	
Information gathering and sharing—Interpersonal and Communication Skills 2					
Level 1	Level 2	Level 3	Level 4	Level 5	
- Actively seeks and understands the patient's/family's perspective - Is aware of factors that affect communication	- Communicates general and sensitive information with awareness of the social context with substantial guidance - Demonstrates knowledge of and sensitivity to cultural values in communicating genetic information with substantial guidance - Provides information and resources with substantial guidance - Utilizes multiple sources and genetic counselors to obtain information with substantial guidance	- Communicates general and sensitive information with awareness of the social context with minimal guidance - Demonstrates knowledge of and sensitivity to cultural values in communicating genetic information with minimal guidance - Provides information and resources with minimal guidance	- Independently communicates general and sensitive information with awareness of the social context - Independently demonstrates knowledge of and sensitivity to cultural values in communicating genetic information - Independently provides information and resources	Makes a nationally recognized contribution by developing educational materials for patients and/or providers	