

Medical Biochemical Genetics Milestones

The Accreditation Council for Graduate Medical Education



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Medical Biochemical Genetics Milestones

The Milestones are designed only for use in evaluation of fellows in the context of their participation in ACGME-accredited residency or fellowship programs. The Milestones provide a framework for the assessment of the development of the fellow in key dimensions of the elements of physician competence in a specialty or subspecialty. They neither represent the entirety of the dimensions of the six domains of physician competency, nor are they designed to be relevant in any other context.

Medical Biochemical Genetics Milestones Work Group

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American Board of Medical Genetics and Genomics

Review Committee for Medical Genetics and Genomics

Understanding Milestone Levels and Reporting

This document presents the Milestones, which programs use in a semi-annual review of fellow performance, and then report to the ACGME. Milestones are knowledge, skills, attitudes, and other attributes for each of the ACGME Competencies organized in a developmental framework. The narrative descriptions are targets for resident/fellow performance throughout their educational program.

Milestones are arranged into levels. Tracking from Level 1 to Level 5 is synonymous with moving from novice to expert fellow in the specialty or subspecialty. For each reporting period, the Clinical Competency Committee will review the completed evaluations to select the milestone levels that best describe each learner's current performance, abilities, and attributes for each subcompetency.

These levels *do not* correspond with post-graduate year of education. Depending on previous experience, a junior fellow may achieve higher levels early in his/her educational program just as a senior fellow may be at a lower level later in his/her educational program. There is no predetermined timing for a resident to attain any particular level. Fellows may also regress in achievement of their milestones. This may happen for many reasons, such as over scoring in a previous review, a disjointed experience in a particular procedure, or a significant act by the fellow.

Selection of a level implies the fellow substantially demonstrates the milestones in that level, as well as those in lower levels (see the diagram on page vi).

Additional Notes

Level 4 is designed as a graduation *goal* but *does not* represent a graduation *requirement*. Making decisions about readiness for graduation and unsupervised practice is the purview of the program director. Furthermore, Milestones 2.0 include revisions and changes that preclude using Milestones as a sole assessment in high-stakes decisions (i.e., determination of eligibility for certification or credentialing). Level 5 is designed to represent an expert fellow whose achievements in a subcompetency are greater than the expectation. Milestones are primarily designed for formative, developmental purposes to support continuous quality improvement for individual learners, education programs, and the specialty. The ACGME and its partners will continue to evaluate and perform research on the Milestones to assess their impact and value.

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

A Supplemental Guide is also available to provide the intent of each subcompetency, examples for each level, assessment methods or tools, and other available resources. The Supplemental Guide, like examples contained within the Milestones, is designed only to assist the program director and Clinical Competency Committee, and is not meant to demonstrate any required element or outcome.

Supplemental Guides and other resources are available on the Milestones page of each specialty section of the ACGME website. On www.acgme.org, choose the applicable specialty under the "Specialties" menu, then select the "Milestones" link in the lower navigation bar.

The diagram below presents an example set of milestones for one sub-competency in the same format as the ACGME Report Worksheet. For each reporting period, a fellow's performance on the milestones for each sub-competency will be indicated by selecting the level of milestones that best describes that fellow's performance in relation to those milestones.

Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates organized diagnostic and therapeutic reasoning through notes in the patient record	Concisely reports diagnostic and therapeutic reasoning in the patient record	Communicates clearly, concisely, timely, and in an organized written form, including anticipatory guidance	Provides feedback to junior learners on reporting diagnostic and therapeutic reasoning in the patient record	Models feedback to improve others' written communication
Uses documentation shortcuts accurately, appropriately and in a timely manner	Appropriately selects direct (e.g., telephone, in- person) and indirect (e.g., progress notes, text messages) forms of communication based on context	Achieves written or verbal communication (e.g., patient notes, e-mail) that serves as an example for others to follow	Mentors junior learners in written and verbal communication	Guides departmental or institutional communication around policies and procedures
middle of a milestones	response box in the level implies that in that level and in lower been substantially ed.	between level in lower levels demonstrated	sponse box on the line in s indicates that milestone have been substantially as well as some the higher level(s).	es

Patient Care 1: Initial or	Acute Management			
Level 1	Level 2	Level 3	Level 4	Level 5
With guidance, recognizes signs and symptoms of inborn errors of metabolism and identifies when further testing is needed	Independently recognizes signs and symptoms of inborn errors of metabolism and identifies when further testing is needed	Synthesizes the clinical context and pattern of laboratory results to identify the likelihood of a common inborn error of metabolism	Synthesizes the clinical context and pattern of laboratory results to identify the likelihood of a complex or rare inborn error of metabolism	Leads development of institutional protocols for diagnosis and management of inborn errors of metabolism
Selects and orders diagnostic tests and develops a rudimentary management plan for initial evaluation or implements the existing management plan	Develops and implements an initial management plan for common patient presentations of inborn errors of metabolism (e.g., organic aciduria, urea cycle)	Develops and implements an initial management plan for complex or rare patient presentations of inborn errors of metabolism (e.g., pyruvate dehydrogenase complex deficiency, carnitine-acylcarnitine translocase deficiency)	Anticipates and creates contingency plan for an acute response and develops an acute secondary plan	
Comments: Not Yet Completed Level 1 Not Yet Assessable				

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Patient Care 2: Chronic Management				
Level 1	Level 2	Level 3	Level 4	Level 5
Takes a comprehensive genetic history for a patient with pertinent positive and negative findings; integrates the history with other data to develop a differential diagnosis	Takes an inborn error of metabolism focused history for a patient with an established common inborn error of metabolism (e.g., PKU)	Takes a disease-specific history for a patient with a complex inborn error of metabolism with multisystemic manifestations (e.g., Hunter syndrome)	Efficiently takes a nuanced disease-specific history for a patient with an inborn error of metabolism	
Implements dietary and medical therapy for patients with a common inborn error of metabolism (e.g., carnitine uptake defect, medium-chain acyl-CoA dehydrogenase deficiency, dietary therapy for phenylketonuria (PKU))	Synthesizes clinical and laboratory data to optimize complex dietary and medical therapy (e.g., urea cycle disorders)	Identifies a patient who, based on natural history, is a candidate for disease modifying therapy (e.g., solid organ transplant, enzyme replacement therapy, cell-based therapy)	Guides a patient through shared decision making on novel or high-risk therapies; coordinates implementation of these therapies	Implements a clinical trial to improve management of patients with an inborn error of metabolism
Provides symptomatic and supportive care to patients	Provides routine disease- specific surveillance	Provides ongoing comprehensive surveillance of disease progression and care coordination	Anticipates and creates contingency plans for highly pleotropic inborn errors of metabolism (e.g., mucopolysaccharidosis I)	Implements a multi- disciplinary clinic for care of patients with an inborn error of metabolism
Comments: Not Yet Completed Level 1 Not Yet Assessable				

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Patient Care 3: Newborn Screening				
Level 1	Level 2	Level 3	Level 4	Level 5
Applies published (state or national) protocols to obtain follow-up testing based on initial newborn screen results	Generates a differential diagnosis based on newborn screen results that includes diagnoses other than inherited metabolic diseases	Applies published protocols for diagnosis with modifications to account for patient specific factors	Integrates data from multiple sources to arrive at an accurate diagnosis for the patient	Participates in state, regional, or national newborn screening program policy development or evaluation projects
Applies published (state or national) protocols for initial management based on the initial newborn screen result	Develops a continuing management plan when initial testing is inconclusive	Makes appropriate decisions about urgency of implementation of management based on initial clinical history	Develops a plan for management of a patient with incidental findings or results that indicate concern for a disease that is not intended to be identified with newborn screening	
Comments:			Not Yet Co	ompleted Level 1
			Not Yet As	ssessable

Medical Knowledge 1: Molecular and Metabolic Mechanisms				
Level 1	Level 2	Level 3	Level 4	Level 5
Describes biochemical pathways and principles underpinning diagnosis and therapy for categories of inborn errors of metabolism (e.g., protein restriction, cofactor therapy, pathway modifying medications)	Describes how manipulation of biochemical pathways through various therapeutic modalities (diet, medications, etc.) can be employed for management of specific inborn errors of metabolism (e.g., ornithine transcarbamylase deficiency versus argininosuccinate lyase deficiency; cystathionine beta-synthase deficiency versus cobalamin C disease)	Demonstrates knowledge of the interconnectedness of biochemical pathways and employs these concepts in diagnosis and management	Integrates knowledge of the complex nature of biochemical pathways to prioritize and employ the range of therapeutic options for a patient, based upon an individual's clinical and biochemical response to therapy	Expands understanding of underlying biochemical basis of inborn errors of metabolism through publication of new knowledge that impacts the diagnosis or treatment of a disease or category of inborn errors of metabolism
Comments: Not Yet Completed Level 1 Not Yet Assessable				

Medical Knowledge 2: Diagnostic Testing				
Level 1	Level 2	Level 3	Level 4	Level 5
Describes the technology and use of diagnostic testing for inborn errors of metabolism	Identifies possible methods for diagnosis and subsequent laboratory monitoring for inborn errors of metabolism	Identifies best methods for diagnosis and subsequent laboratory monitoring for common inborn errors of metabolism	Identifies best methods for diagnosis and subsequent laboratory monitoring for complex inborn errors of metabolism	Develops polices or practice guidelines for diagnostic testing of inborn errors of metabolism
	Interprets common metabolic testing (e.g., plasma amino acids) for inborn errors of metabolism	Interprets complex metabolic testing (e.g., very long chain fatty acids) for inborn errors of metabolism	Integrates results of metabolic and genomic testing to arrive at a diagnosis	Participates in new assay development or gene discovery
Demonstrates knowledge of the differences between newborn screening and diagnostic testing	Describes causes of false positive/negative rates and how these factors can impact newborn screening test interpretation	Demonstrates knowledge of positive predictive values of newborn screening tests and tools for improved discrimination of positive and negative results	Integrates data from multiple sources to identify how likely an individual newborn screening test is a true positive	Contributes to generalizable medical knowledge of newborn screening tests
Comments: Not Yet Completed Level 1 Not Yet Assessable				

Systems-Based Practice	e 1: Patient Safety and Qua	lity Improvement		
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates knowledge of common patient safety events	Identifies system factors that lead to patient safety events	Participates in analysis of patient safety events (simulated or actual)	Conducts analysis of patient safety events and offers error prevention strategies (simulated or actual)	Actively engages teams and processes to modify systems to prevent patient safety events
Demonstrates knowledge of how to report patient safety events	Reports patient safety events through institutional reporting systems (simulated or actual)	Participates in disclosure of patient safety events to patients and families (simulated or actual)	Independently discloses patient safety events to patients and families (simulated or actual)	Mentors others in the disclosure of patient safety events
Demonstrates knowledge of basic quality improvement methodologies and metrics	Describes local (institutional) quality improvement initiatives	Participates in local (institutional) quality improvement initiatives	Demonstrates the skills required to identify, develop, implement, and analyze a quality improvement project	Creates, implements, and assesses quality improvement initiatives at the institutional or community (state/federal) level
Comments: Not Yet Completed Level 1				

Systems-Based Practice 2: System Navigation for Patient-Centered Care				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates knowledge of care coordination	Coordinates care of patients in routine clinical situations effectively using the roles of the interprofessional teams, including non-physician patient caregivers	Coordinates care of patients in complex clinical situations effectively using the roles of the interprofessional teams	Models effective coordination of patient-centered care among different disciplines and specialties including referrals and testing	Analyzes the process of care coordination and leads in the design and implementation of quality improvements
Identifies key elements for safe and effective transitions of care and hand-offs	Performs safe and effective transitions of care/hand-offs in routine clinical situations	Performs safe and effective transitions of care/hand-offs in complex clinical situations	Models and advocates for safe and effective transitions of care/hand- offs within and across health care delivery systems including outpatient settings, referrals, and testing	Improves quality of transitions of care within and across health care delivery systems to optimize patient outcomes
Demonstrates knowledge of population and community health needs and disparities	Identifies specific population and community health needs and inequities for the local population	Uses local resources effectively to meet the needs of a patient population and community	Participates in changing and adapting practice to provide for the needs of specific populations including advocating for a patient's genetic testing coverage	Leads innovations and advocates for populations and communities with health care inequities at the state or federal level
Comments: Not Yet Completed Level 1				

Level 1	Level 2	Level 3	Level 4	Level 5
Identifies key components of the complex health care system (e.g., hospital, skilled nursing facility, finance, personnel, technology)	Describes how components of a complex health care system are interrelated, and how this impacts patient care	Discusses how individual practice affects the broader system (e.g., access to genetic testing and treatments, testing advocacy)	Manages various components of the complex health care system to provide efficient and effective patient care and transition of care	Advocates for or leads systems change that enhances high-value, efficient, and effective patient care and transition of care
Describes basic health payment systems (e.g., government, private, public, uninsured care) and practice models	Delivers care with consideration of each patient's payment model (e.g., insurance type) and access to genetic testing or formula	Engages with patients in shared decision making, often informed by each patient's payment models	Advocates for patient care needs (e.g., community resources, patient assistance resources) with consideration of the limitations of each patient's payment model, including genetic testing through research	Participates in health policy advocacy activities
Identifies basic knowledge for effective transition to practice (e.g., information technology, legal, billing and coding, financial, personnel)	Demonstrates use of information technology required for medical practice (e.g., electronic health record, documentation required for billing and coding)	Describes core administrative knowledge needed for transition to practice (e.g., contract negotiations, malpractice insurance, government regulation, compliance)	Analyzes individual practice patterns and professional requirements in preparation for practice	Educates others to prepare them for transition to practice

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Practice-Based Learning and Improvement 1: Evidence-Based and Informed Practice				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates how to access and use available evidence, and incorporate patient preferences and values to take care of a routine patient	Articulates clinical questions and elicits patient preferences and values to guide evidence-based care	Locates and applies the best available evidence, integrated with patient preference, to the care of complex patients	Critically appraises and applies evidence even in the face of uncertainty and conflicting evidence to guide care, tailored to the individual patient	Mentors others to critically appraise and apply evidence for complex patients; and/or participates in the development of guidelines
Comments: Not Yet Completed Level 1				

Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates openness to performance data (feedback and other input) to inform goals	Seeks performance data episodically, with adaptability and humility	Seeks performance data consistently with adaptability and humility	Models seeking performance data with adaptability and humility	Develops evaluations or education resources/tools for learners
Analyzes and reflects on the factors which contribute to gap(s) between expectations and actual performance	Analyzes, reflects on, and institutes behavioral change(s) to narrow the gap(s) between expectations and actual performance	Challenges assumptions and considers alternatives in narrowing the gap(s) between expectations and actual performance	Mentors others on reflective practice	Participates in the development of courses for the education of students or other physicians
Designs and implements a learning plan, with prompting	Independently creates and implements a learning plan	Uses performance data to measure the effectiveness of the learning plan and when necessary, improves it	Facilitates the design and implementing learning plans for others	

Professionalism 1: Profe	essional Behavior and Ethic	cal Principles		
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates compassion, sensitivity, honesty, integrity, and identifies potential triggers for professionalism lapses	Demonstrates compassion, sensitivity, honesty, integrity, and takes responsibility for own professionalism lapses	Demonstrates compassion, sensitivity, honesty, and integrity in complex/stressful situations	Demonstrates compassion, sensitivity, honesty, integrity, and serves as a role model to others	Coaches others when their behavior fails to meet professional expectations
Demonstrates knowledge of the ethical principles underlying patient care	Analyzes straightforward situations using ethical principles	Recognizes need to seek help in managing and resolving complex ethical situations	Recognizes and uses appropriate resources for managing and resolving ethical dilemmas as needed	Identifies and seeks to address system-level factors that induce or exacerbate ethical problems or impede their resolution
Demonstrates basic knowledge of conflict of interest	Identifies different types of conflicts of interest, knows guidelines for interactions with vendors	Identifies resources for managing and resolving conflicts of interest	Demonstrates consistently professional behavior regarding conflicts of interest relevant to presentations, publishing, consulting, and service	
Comments: Not Yet Completed Level 1				

Level 1	Level 2	Level 3	Level 4	Level 5
Takes responsibility for failure to complete tasks and responsibilities, identifies potential contributing factors, and describes strategies for ensuring timely task completion in the future	Performs tasks and responsibilities in a timely manner with appropriate attention to detail in routine situations	Performs tasks and responsibilities in a timely manner with appropriate attention to detail in complex or stressful situations	Recognizes and addresses situations that may impact others' ability to complete tasks and responsibilities in a	Volunteers to improve and takes ownership of system outcomes
Responds promptly to requests or reminders to complete tasks and responsibilities	Recognizes situations that may impact his/her own ability to complete tasks and responsibilities in a timely manner	Proactively implements strategies to ensure that the needs of patients, teams, and systems are met	timely manner	
Recognizes the role of appearance, daily demeanor and conduct in the role of a professional	Demonstrates a professional appearance, daily demeanor, and conduct	Sets a standard for appearance, daily demeanor, and conduct as a professional	Promotes professional appearance, demeanor, and conduct in their peers and associates	

Professionalism 3: Self-Awareness and Help-Seeking				
Level 1	Level 2	Level 3	Level 4	Level 5
Recognizes status of personal and professional well-being, with assistance	Independently recognizes status of personal and professional well-being	With assistance, proposes a plan to optimize personal and professional well-being	Independently develops a plan to optimize personal and professional well-being	Coaches others when emotional responses or limitations in knowledge/skills do not meet professional expectations
Recognizes limits in the knowledge/skills of self or team, with assistance	Independently recognizes limits in the knowledge/ skills of self or team and demonstrates appropriate help-seeking behaviors	With assistance, proposes a plan to remediate or improve limits in the knowledge/skills of self or team	Independently develops a plan to remediate or improve limits in the knowledge/skills of self or team	
Comments:				ompleted Level 1

This subcompetency is not intended to evaluate a fellow's well-being, but to ensure each fellow has the fundamental knowledge of factors that impact well-being, the mechanisms by which those factors impact well-being, and available resources and tools to improve well-being.

Level 1	Level 2	Level 3	Level 4	Level 5
Uses language and nonverbal behavior to demonstrate respect and establish rapport	Establishes a therapeutic relationship in straightforward encounters using active listening and clear language	Establishes a therapeutic relationship in challenging patient encounters	Establishes therapeutic relationships, with attention to patient/family concerns and context, regardless of complexity	Mentors others in situational awareness and critical self-reflection to consistently develop positive therapeutic relationships
Identifies common barriers to effective communication while accurately communicating own role within the health care system	Identifies complex barriers to effective communication	When prompted, reflects on personal biases while attempting to minimize communication barriers	Recognizes personal biases while attempting to proactively minimize communication barriers	Models self-awareness practice while identifying teaching a contextual approach to minimize communication barriers
Identifies the need to adjust communication strategies based on assessment of patient/family expectations and understanding of their health status and treatment options	Organizes and initiates communication with patient/family by introducing stakeholders, setting the agenda, clarifying expectations, and verifying understanding of the clinical situation	With guidance, sensitively and compassionately delivers medical information, elicits patient/family values, goals, and preferences, and acknowledges uncertainty and conflict	Uses shared decision making to align patient/family values, goals, and preferences with treatment options to make a personalized care plan	Models shared decision making in patient/family communication including those with a high degree of uncertainty/conflict

Level 1	Level 2	Level 3	Level 4	Level 5
Clearly and concisely requests a consultation	Checks own understanding of consultant recommendations	Coordinates recommendations from different members of the health care team to optimize patient care	Leads a metabolic team of diverse members to optimize patient care	Models flexible communication strategies that value input from all health care team members, resolving conflict when needed
Clearly and concisely responds to a consultation request	Checks requestor's understanding of recommendations when providing consultation	Provides information to the primary care team regarding rationale for recommendations	Leads interactions between the primary care and metabolic team regarding rationale for recommendations	
Communicates information effectively with all health care team members	Uses active listening to adapt communication style to fit team needs	Models active listening to other health care team members	Provides constructive feedback on active listening to health care team members	
Comments:				

Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates organized diagnostic and therapeutic reasoning through notes in the patient record	Concisely reports diagnostic and therapeutic reasoning in the patient record	Communicates clearly, concisely, timely, and in an organized written form, including anticipatory guidance	Provides feedback to more junior learners on reporting diagnostic and therapeutic reasoning in the patient record	Models feedback to improve others' written communication
Uses documentation shortcuts accurately, appropriately and in a timely manner	Appropriately selects direct (e.g., telephone, inperson) and indirect (e.g., progress notes, text messages) forms of communication based on context	Achieves written or verbal communication (e.g., patient notes, email) that serves as an example for others to follow	Mentors more junior learners in written and verbal communication	Guides departmental or institutional communication around policies and procedures