

# The Laboratory Genetics and Genomics Milestone Project

*An Initiative of*

The Accreditation Council for Graduate Medical Education



ACGME

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# Laboratory Genetics and Genomics Milestones

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

## Laboratory Genetics and Genomics Milestones

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American Board of Medical Genetics and Genomics  
Association of Professors of Human and Medical Genetics  
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 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 fellow 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.

Examples are provided for some milestones within this document. Please note: the examples are not the required element or outcome; they are provided as a way to share the intent of the element.

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](http://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.

Systems-Based Practice 1: Patient Safety and Quality Improvement (QI)				
Level 1	Level 2	Level 3	Level 4	Level 5
Describes 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
Identifies that processes exist for reporting patient safety events	Defines the institutional process of safety reporting systems	Given a safety-related concern, can describe the risk management process of disclosing such events to the appropriate individuals	Reports patient safety events (simulated or actual event)	Develops innovative protocols to detect and report safety events
Demonstrates knowledge of QI concepts	Describes laboratory QI initiatives	Participates in laboratory QI initiatives	Demonstrates the skills required to identify, develop, implement, and analyze a QI project	Develops and assesses QI initiatives at the institutional or community level
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <span style="float: right;">Not Yet Completed Level 1 <input type="checkbox"/></span>				

Selecting a response box in the middle of a level implies that milestones in that level and in lower levels have been substantially demonstrated.

Selecting a response box on the line in between levels indicates that milestones in lower levels have been substantially demonstrated as well as **some** milestones in the higher level(s).

Patient Care 1: Pre-Analytic				
Level 1	Level 2	Level 3	Level 4	Level 5
Describes the importance of clinical history for optimal test selection	Gathers pertinent elements of the clinical history to aid in test selection	Recommends the optimal test option(s) based on clinical or family history, with assistance	Independently recommends the optimal test(s) based on clinical or family history	Creates an algorithm for test selection based on clinical history
Identifies elements of a laboratory test request	Recognizes sources of pre-analytic error	Evaluates a specimen for pre-analytic errors, with assistance	Independently evaluates a specimen for pre-analytic errors and identifies possible resolutions	Creates a new protocol to assist the laboratory in the appraisal of specimen issues
	Describes rationale behind existing routine laboratory workflows	Suggests modifications to existing laboratory workflows based on clinical need, with assistance	Independently suggests modifications to existing laboratory workflows based on clinical need	Independently develops a new laboratory workflow
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">                     Not Yet Completed Level 1 <input type="checkbox"/>                      Not Yet Assessable <input type="checkbox"/> </div>				

Patient Care 2: Analytic				
Level 1	Level 2	Level 3	Level 4	Level 5
Describes basic principles for major assays performed in the laboratory	Performs assays, with substantial assistance	Performs assays, with minimal assistance	Independently performs assays	Develops new, alternate, or improved assay
Describes quality control (QC) in the clinical laboratory	Identifies QC failures	Explains possible sources of QC failures	Investigates QC failures and proposes resolution	Identifies and implements a new QC approach for a clinical test
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">                     Not Yet Completed Level 1 <input type="checkbox"/>                      Not Yet Assessable <input type="checkbox"/> </div>				



Patient Care 3: Post-Analytic Skills				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies normal results	Interprets simple results, with assistance	Interprets complex results, with assistance	Independently interprets results	Develops an improved result interpretation workflow
Discusses the importance of patient's clinical history to test interpretation	Gathers pertinent elements of the clinical history to aid in interpretation	Integrates results with the clinical history to develop a final interpretation, with assistance	Independently integrates results with the clinical history to develop a final interpretation	Identifies novel correlations between results and clinical history
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></div>				

Patient Care 4: Reports				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies the elements of a laboratory report	Drafts a report for simple cases using accurate terminology/ nomenclature, with assistance	Drafts a report for complex cases using accurate terminology/ nomenclature, with assistance	Independently generates concise reports for complex cases	Develops a new reporting template for original or revised reports
Identifies that reports can be revised	Identifies when to correct, amend, or addend a report based on the type of alteration required	Drafts a revised report, with assistance	Independently generates a revised report	
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">                     Not Yet Completed Level 1 <input type="checkbox"/>                      Not Yet Assessable <input type="checkbox"/> </div>				

Medical Knowledge 1: Foundations of Medical Genetics and Genomics				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>Defines relevant medical terminology</p> <p>Explains basic structure and function of the genome</p> <p>Describes the basic mechanisms of germline and somatic disease development</p>	<p>Describes genetic conditions using accurate medical terminology</p> <p>Identifies the basic genomic alterations underpinning abnormal lab results</p> <p>Recognizes the pathogenic mechanisms for well-described conditions</p>	<p>With assistance, assimilates and integrates medical information to generate a differential diagnosis</p> <p>Correlates genomic alterations and phenotypic information to guide laboratory testing and interpretation, with assistance</p> <p>Applies understanding of pathogenic mechanisms to interpretation of test results, with assistance</p>	<p>Independently assimilates and integrates medical information to generate a differential diagnosis</p> <p>Independently correlates genomic alterations and phenotypic information to guide laboratory testing and interpretation</p> <p>Independently applies understanding of pathogenic mechanisms to interpretation of test results</p>	<p>Mentors others in the process of integration of clinical and laboratory findings to generate a differential diagnosis</p> <p>Presents a new clinical genomic/phenotypic correlation</p>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<p><b>Comments:</b></p> <p style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></p> <p style="text-align: right;">Not Yet Assessable <input type="checkbox"/></p>				

Medical Knowledge 2: Testing				
Level 1	Level 2	Level 3	Level 4	Level 5
Defines the basic components of a test validation and verification	Describes methods and data used in establishing test performance characteristics for validation and verification	Determines test performance characteristics, with assistance	Independently calculates test performance characteristics	Designs a test validation/verification and establishes QC metrics
Describes various methodologies used in a clinical genetics laboratory	Describes the strengths and limitations of a genetic laboratory test	Determines the optimal technologies and platforms for genetic tests, with assistance	Independently determines the optimal technologies and platforms for genetic tests	Independently designs a new test for a genetic condition
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="float: right;">                     Not Yet Completed Level 1 <input type="checkbox"/>                      Not Yet Assessable <input type="checkbox"/> </div>				

Systems-Based Practice 1: Patient Safety and Quality Improvement (QI)				
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Describes 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
Identifies that processes exist for reporting patient safety events	Defines the institutional process of safety reporting systems	Given a safety-related concern, can describe the risk management process of disclosing such events to the appropriate individuals	Reports patient safety events (simulated or actual event)	Develops innovative protocols to detect and report safety events
Demonstrates knowledge of QI concepts	Describes laboratory QI initiatives	Participates in laboratory QI initiatives	Demonstrates the skills required to identify, develop, implement, and analyze a QI project	Develops and assesses QI initiatives at the institutional or community level
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></div>				

Systems-Based Practice 2: Systems Navigation for Patient-Centered Care				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies the importance of coordinating care	Describes effective care coordination with the clinical and laboratory team	Coordinates care with the clinical and laboratory team, with assistance	Coordinates care with the clinical and laboratory team	
Explains the importance of effective transitions of care and hand-offs	Describes examples of safe and effective transitions of care/hand-offs	Performs safe and effective transitions of care/hand-offs	Models and advocates for safe and effective transitions of care/hand-offs	Improves quality of transitions of care within and across health care delivery systems to optimize patient outcomes
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></div>				

Systems-Based Practice 3: Laboratory Geneticist's Role in Health Care System				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies key components of the health care system	Describes how components of a health care system are interrelated, and how this impacts patient care	Collaborates with the other members of the health care system, with assistance	Independently collaborates with the other members of the health care system	Advocates for or leads systems change that enhances high-value, efficient, and effective patient care
Identifies basic types of medical reimbursement	Identifies testing documentation related to billing and reimbursement	Identifies opportunities for cost-effective patient care	Practices cost-effective patient care	
	Describes the financial components related to the laboratory operation	Identifies inter-relationship between fiscal responsibility and quality metrics in a lab (e.g., balancing staffing needs, test reagent needs, cost containment, and billing efficiency)	Independently drafts the assessment of a laboratory fiscal metric for director review	Participates in a local or national committee related to fiscal issues in genetic testing
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></div>				

Systems-Based Practice 4: Accreditation, Compliance, and Quality Management				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies laboratory accreditation and licensing agencies	Describes the accreditation process, its requirements and necessary documentation	Participates in review of laboratory practice to assure compliance with accreditation requirements	Actively participates in the laboratory self-inspection	Participates in the inspection of an external laboratory
Defines terminology related to laboratory quality	Interprets quality data and charts and trends, including proficiency testing results, with assistance	Independently evaluates quality indicators, including proficiency testing results	Formulates a response for a proficiency test failure (actual or simulated)	Reviews the quality management plan to identify areas for improvements
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></div>				



Systems-Based Practice 5: Informatics				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>Demonstrates familiarity with basic technical concepts of hardware, operating systems, databases, and software for general purpose applications</p> <p>Identifies the various resources including software, tools and databases used for classification of variants</p>	<p>Identifies laboratory specific software, key technical concepts, interfaces, workflow, barcode application, and automation systems (enterprise systems architecture)</p> <p>Uses resources to classify variants in the context of the phenotype, with assistance</p>	<p>Discusses laboratory initiatives based on informatics (system implementation and configuration)</p> <p>Independently uses resources to perform routine variant classification using professional standards and guidelines</p>	<p>Applies informatics tools as needed in laboratory initiatives (e.g., data management and security, computational statistics, information governance)</p> <p>Independently uses resources to reclassify ambiguous variants</p>	<p>Proposes medical informatics improvements for the operation of the laboratory</p> <p>Contributes to the knowledge base for the refinement of ambiguous test results and variant classifications</p>
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<p><b>Comments:</b></p> <p style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></p>				

Practice-Based Learning and Improvement 1: Evidence-Based Practice				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates how to access and select applicable evidence	Identifies and applies the best available evidence and/or clinical laboratory standards/guidelines to guide diagnostic evaluation of simple cases	Identifies and applies the best available evidence and/or clinical laboratory standards/guidelines to guide diagnostic evaluation of complex cases	Critically appraises and applies evidence to guide lab-based recommendations, even in the face of conflicting data	Mentors others to critically appraise and apply evidence for complex cases; and/or participates in the development of laboratory standards/guidelines
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></div>				

Practice-Based Learning and Improvement 2: Reflective Practice and Commitment to Personal Growth				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>Realizes responsibility for personal and professional development by establishing goals</p> <p>Identifies the gap(s) between expectations and actual performance</p> <p>Actively seeks opportunities to improve</p>	<p>Demonstrates willingness to receiving performance data and feedback in order to inform goals</p> <p>Analyzes and reflects on the factors which contribute to gap(s) between expectations and actual performance</p> <p>Designs and implements a learning plan, with assistance</p>	<p>Seeks performance data and feedback with respect</p> <p>Institutes behavioral change(s) to narrow the gap(s) between expectations and actual performance</p> <p>Independently creates and implements a learning plan</p>	<p>Models appropriate and thoughtful seeking and consideration of feedback</p> <p>Critically evaluates the effectiveness of behavioral changes in narrowing the gap(s) between expectations and actual performance</p> <p>Uses performance data to measure the effectiveness of the learning plan and improves it when necessary</p>	<p>Coaches others in personal and professional development</p> <p>Facilitates the design and implementation of learning plans for others</p>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<p><b>Comments:</b></p> <p style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></p>				

Professionalism 1: Professional Behavior and Ethical Principles				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>Demonstrates knowledge of the ethical principles underlying laboratory testing</p> <p>Describes how to report professionalism lapses, including strategies for addressing common barriers</p>	<p>Analyzes straightforward situations using ethical principles</p> <p>Demonstrates insight into professional behavior in routine situations; takes responsibility for own professionalism lapses</p>	<p>Recognizes the need for and uses appropriate resources to seek solutions in managing and resolving complex ethical situations</p> <p>Demonstrates professional behavior in complex or stressful situations</p>	<p>Manages complex ethical situations</p> <p>Recognizes situations that may trigger professionalism lapses and intervenes to prevent lapses in self and others</p>	<p>Identifies and seeks to address system-level factors that introduce or exacerbate ethical problems or impede their resolution</p> <p>Coaches others when their behavior fails to meet professional expectations</p>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<p><b>Comments:</b></p> <p style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></p>				

Professionalism 2: Accountability and Conscientiousness				
Level 1	Level 2	Level 3	Level 4	Level 5
Responds promptly to instructions, requests, or reminders to complete tasks and responsibilities	Takes ownership and performs tasks and responsibilities in a timely manner	Anticipates situations that may impact own ability to meet responsibilities and describes the impact on team	Shares responsibility for system outcomes as a member of the team	Designs new strategies to ensure that the needs of patients, teams, and systems are met
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></div>				

Professionalism 3: Personal and Professional Well-Being				
Level 1	Level 2	Level 3	Level 4	Level 5
Describes common indicators of personal or professional well-being	Independently recognizes status of personal and professional well-being and seeks help when needed	Proposes a plan to optimize personal and professional well-being	Implements a plan to optimize personal and professional well-being	Coaches others to develop and implement plans to optimize personal and professional well-being
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></div>				

Interpersonal and Communication Skills 1: Patient and Family-Centered Communication				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies common barriers to effective communication	Identifies complex barriers to effective communication	With prompting, reflects on personal biases and can identify barriers in written language that may challenge understanding by patients and families	Communicates the content of a report or educational materials in a way that is accessible to patients and families	Develops systems to communicate laboratory information in a way that is accessible to patients and families
Identifies the role of the laboratory in communicating results to patients	Uses nonverbal behavior to demonstrate respect and establish rapport while observing in the clinical setting and can identify factors that could make a clinical situation psychosocially complex	Uses nonverbal behavior to demonstrate respect and establish rapport while observing a complex result disclosure by a clinician	Orally communicates simple results to families in a concise manner, under the supervision of an attending clinician	Independently orally communicates laboratory results with relevant information to providers
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b>				Not Yet Completed Level 1 <input type="checkbox"/>

Interpersonal and Communication Skills 2: Interprofessional and Team Communication				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>Uses language that values all members of the health care team, including clinical and laboratory professionals</p> <p>Describes the utility of constructive feedback</p>	<p>Adapts communication style to fit team needs</p> <p>Solicits feedback on personal performance as a member of the laboratory team</p>	<p>Communicates information effectively with all health care team members, including clinical and laboratory professionals, with assistance</p> <p>Integrates feedback from team members to improve own communication and other skills</p>	<p>Independently communicates information effectively with all health care team members, including clinical and laboratory professionals</p> <p>Facilitates regular health care team-based communications and feedback in complex situations</p>	<p>Models flexible communication strategies that value input from all health care team members, resolving conflict when needed</p>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b>				Not Yet Completed Level 1 <input type="checkbox"/>



Interpersonal and Communication Skills 3: Communication within Health Care Systems				
Level 1	Level 2	Level 3	Level 4	Level 5
Protects patient personal health information by following institutional policies	Selects content, recipient, and communication methods based on context and clinical urgency, with guidance	Effectively and securely communicates clinical information, with guidance	Independently communicates clinical information	Models effective communication of clinical information
Identifies institutional and departmental procedures for communication of issues	Uses institutional structure to effectively communicate clear and constructive suggestions, with assistance	Uses institutional structure to effectively communicate clear and constructive suggestions	Initiates conversations on difficult subjects with appropriate stakeholders to improve the system	Facilitates dialogue regarding systems issues among larger community stakeholders
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Comments:</b> <div style="text-align: right;">Not Yet Completed Level 1 <input type="checkbox"/></div>				