The Clinical Biochemical Genetics Milestone Project

An Initiative of

The Accreditation Council for Graduate Medical Education

April 2020

Clinical Biochemical Genetics 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 competence 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.

Clinical Biochemical Genetics Milestones

Work Group

Miriam Blitzer, PhD

Keri Bosfield, MBBS

Tina Cowan, PhD

Kristina Cusmano-Ozog, MD

Laura Edgar, EdD, CAE

Sarah Elsea, PhD

Rebecca Ganetzky, MD

Alejandro Iglesias, MD

Marzia Pasquali, PhD

Daniel Sharer, PhD

The ACGME would like to thank the following organizations for their continued support in the development of the Milestones:

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.

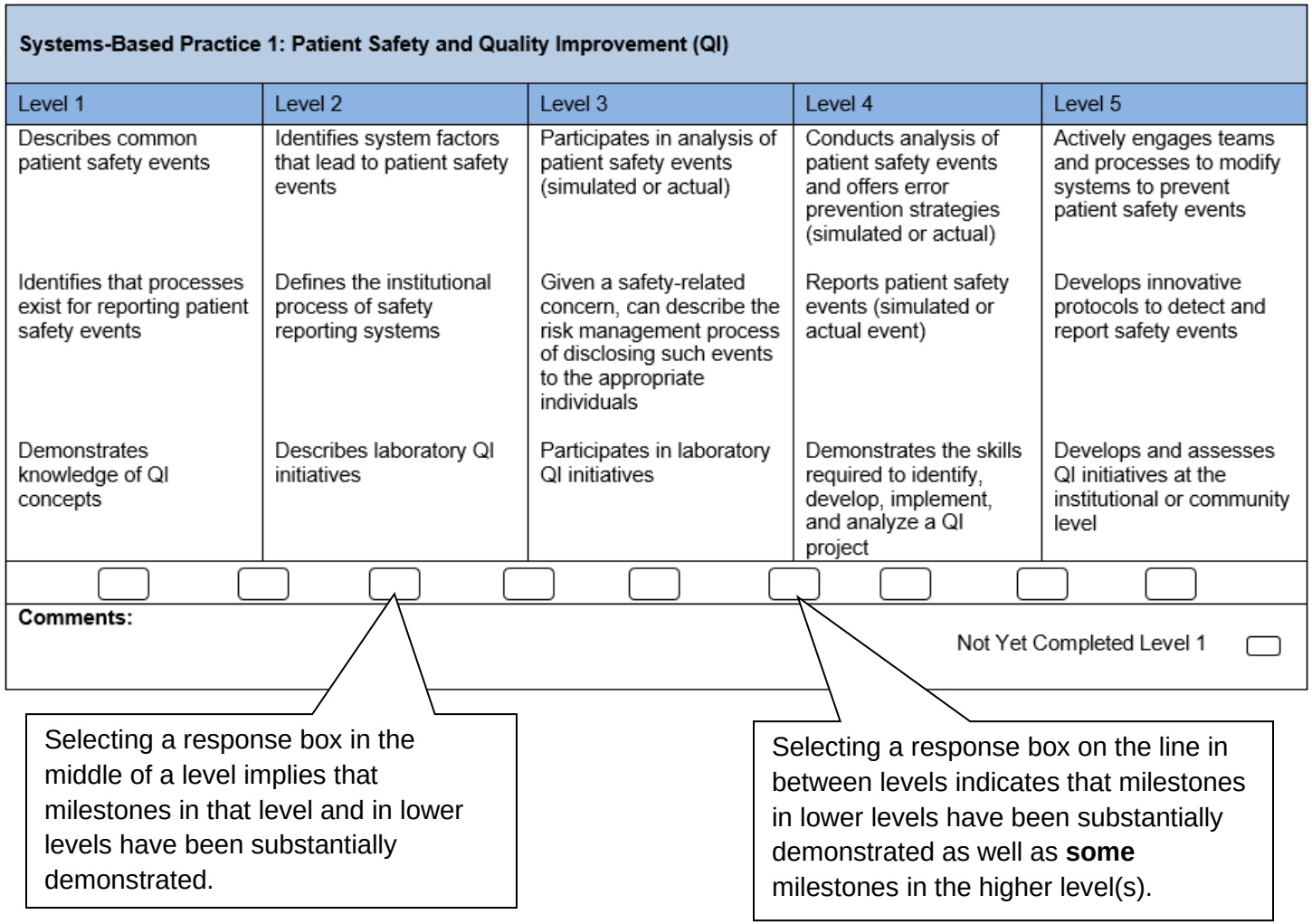
Examples are provided for some milestones within this document. Please note: the examples are not the required element or outcome; they are provided as a way to share the intent of the element.

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.

Additional resources are available in the Milestones section of the ACGME website. Follow the links under “What We Do” at www.acgme.org.

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

Comments:

Not Yet Completed Level 1

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 Identifies elements of a laboratory test request	Gathers pertinent elements of the clinical history to aid in test selection Recognizes sources of pre-analytic error Describes rationale behind existing routine laboratory workflows	Recommends the optimal test option(s) based on clinical or family history, with assistance Evaluates a specimen for pre-analytic errors, with assistance Suggests modifications to existing laboratory workflows based on clinical need, with assistance	Independently recommends the optimal test(s) based on clinical or family history Independently evaluates a specimen for pre-analytic errors and identifies possible resolutions Independently suggests modifications to existing laboratory workflows based on clinical need	Creates an algorithm for test selection based on clinical history Creates a new protocol to assist the laboratory in the appraisal of specimen issues Independently develops a new laboratory workflow
☐	☐	☐	☐	☐ ☐ ☐ ☐
Comments:				
Not Yet Completed Level 1 ☐ Not Yet Assessable ☐				

Patient Care 2: Analytic				
Level 1	Level 2	Level 3	Level 4	Level 5
Describes basic principles for major assays performed in the laboratory Describes quality control (QC) in the clinical laboratory	Performs assays, with substantial assistance Identifies QC failures	Performs assays, with minimal assistance Explains possible sources of QC failures	Independently performs assays Investigates QC failures and proposes resolution	Develops new, alternate, or improved assay Identifies and implements a new QC approach for a clinical test
☐	☐	☐	☐	☐
Comments: Not Yet Completed Level 1 ☐ Not Yet Assessable ☐				

Patient Care 3: Post-Analytic Skills				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies normal results Discusses the importance of patient's clinical history to test interpretation	Interprets simple results, with assistance Gathers pertinent elements of the clinical history to aid in interpretation	Interprets complex results, with assistance Integrates results with the clinical history to develop a final interpretation, with assistance	Independently interprets results Independently integrates results with the clinical history to develop a final interpretation	Develops an improved result interpretation workflow Identifies novel correlations between results and clinical history
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments: Not Yet Completed Level 1 ☐				

Patient Care 4: Reports				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies the elements of a laboratory report Identifies that reports can be revised	Drafts a report for simple cases using accurate terminology/ nomenclature, with assistance Identifies when to correct, amend, or addend a report based on the type of alteration required	Drafts a report for complex cases using accurate terminology/ nomenclature, with assistance Drafts a revised report, with assistance	Independently generates concise reports for complex cases Independently generates a revised report	Develops a new reporting template for original or revised reports
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments:				
				Not Yet Completed Level 1 ☐ Not Yet Assessable ☐

Medical Knowledge 1: Foundations of Medical Genetics and Genomics				
Level 1	Level 2	Level 3	Level 4	Level 5
Defines relevant medical terminology Describes basic principles of medical biochemistry and cell biology	Describes genetic conditions using accurate medical terminology Describes normal metabolic processes	With assistance, assimilates and integrates medical information to generate a differential diagnosis Describes abnormal metabolic processes and clinical phenotypes of associated diseases	Independently assimilates and integrates medical information to generate a differential diagnosis Describes the primary and secondary metabolic effects and clinical phenotypes of various inborn errors of metabolism	Mentors others in the process of integration of clinical and laboratory findings to generate a differential diagnosis
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments:				
Not Yet Completed Level 1 ☐ Not Yet Assessable ☐				

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 various methodologies used in a clinical biochemical genetics laboratory	Describes methods and data used in establishing test performance characteristics for validation and verification Describes the strengths and limitations of a biochemical laboratory test	Determines test performance characteristics, with assistance Determines the optimal technologies and platforms for biochemical tests, with assistance	Independently calculates test performance characteristics Independently determines the optimal technologies and platforms for biochemical tests	Designs a test validation/verification and establishes QC metrics Independently designs a new test for a genetic condition
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments:				
Not Yet Completed Level 1 ☐ Not Yet Assessable ☐				

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
☐	☐	☐	☐	☐
Comments:				
				Not Yet Completed Level 1 ☐

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
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments:				
Not Yet Completed Level 1 ☐				

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
☐	☐	☐	☐	☐

Comments:

Not Yet Completed Level 1 ☐

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 Defines terminology related to laboratory quality	Describes the accreditation process, its requirements, and necessary documentation Interprets quality data and charts and trends, including proficiency testing results, with assistance	Participates in review of laboratory practice to assure compliance with accreditation requirements Independently evaluates quality indicators, including proficiency testing results	Actively participates in the laboratory self-inspection Formulates a response for a proficiency test failure (actual or simulated)	Participates in the inspection of an external laboratory Reviews the quality management plan to identify areas for improvements
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments: Not Yet Completed Level 1 ☐				

Systems-Based Practice 5: Informatics				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates familiarity with basic technical concepts of hardware, operating systems, databases, and software for general purpose applications	Identifies laboratory specific software, key technical concepts, interfaces, workflow, barcode application, and automation systems (enterprise systems architecture)	Discusses laboratory initiatives based on informatics (system implementation and configuration)	Applies informatics tools as needed in laboratory initiatives (e.g., data management and security, computational statistics, information governance)	Proposes medical informatics improvements for the operation of the laboratory
☐	☐	☐	☐	☐
Comments:				
Not Yet Completed Level 1 ☐				

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
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments:				
Not Yet Completed Level 1 ☐				

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

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

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
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments:				Not Yet Completed Level 1 ☐

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
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments:				
Not Yet Completed Level 1 ☐				

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 the role of the laboratory in communicating results to patients	Identifies complex barriers to effective communication 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	With prompting, reflects on personal biases and can identify barriers in written language that may challenge understanding by patients and families Uses nonverbal behavior to demonstrate respect and establish rapport while observing a complex result disclosure by a clinician	Communicates the content of a report or educational materials in a way that is accessible to patients and families Orally communicates simple results to families in a concise manner, under the supervision of an attending clinician	Develops systems to communicate laboratory information in a way that is accessible to patients and families Independently orally communicates laboratory results with relevant information to providers
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments: Not Yet Completed Level 1 ☐				

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

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 Identifies institutional and departmental procedures for communication of issues	Selects content, recipient, and communication methods based on context and clinical urgency, with guidance Uses institutional structure to effectively communicate clear and constructive suggestions, with assistance	Effectively and securely communicates clinical information, with guidance Uses institutional structure to effectively communicate clear and constructive suggestions	Independently communicates clinical information Initiates conversations on difficult subjects with appropriate stakeholders to improve the system	Models effective communication of clinical information Facilitates dialogue regarding systems issues among larger community stakeholders
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments: Not Yet Completed Level 1 ☐				