Supplemental Guide:
Clinical Biochemical Genetics

ACGME

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Milestones Supplemental Guide

This document provides additional guidance and examples for the Clinical Biochemical Genetics Milestones. This is not designed to indicate any specific requirements for each level, but to provide insight into the thinking of the Milestone Work Group.

Included in this document is the intent of each Milestone and examples of what a Clinical Competency Committee (CCC) might expect to be observed/assessed at each level. Also included are suggested assessment models and tools for each subcompetency, references, and other useful information.

Review this guide with the CCC and faculty members. As the program develops a shared mental model of the Milestones, consider creating an individualized guide (Supplemental Guide Template available) with institution/program-specific examples, assessment tools used by the program, and curricular components.

Additional tools and references, including the Milestones Guidebook, Clinical Competency Committee Guidebook, and Milestones Guidebook for Residents and Fellows, are available on the Resources page of the Milestones section of the ACGME website.

Patient Care 1: Pre-Analytic **Overall Intent:** To ensure the fellow can identify appropriate specimens for testing and methods of collection, storage, and transport	
Milestones	**Examples**
Level 1 *Describes the importance of clinical history for optimal test selection*	● Identifies patient sex/gender, age, family history, and clinical indication as important factors in guiding test selection
Identifies elements of a laboratory test request	● Identifies sample type, collection date, date of birth, referring provider, medical record number, and clinical indication(s) as important elements of a test request form
Level 2 *Gathers pertinent elements of the clinical history to aid in test selection*	● Locates pertinent clinical information required for testing using the electronic health record (EHR)
Recognizes sources of pre-analytic error	● Identifies that test results may be compromised if the specimen is collected in an inappropriate collection container
Describes rationale behind existing routine laboratory workflows	● Recognizes that testing schedules can be modified to accommodate courier delays
Level 3 *Recommends the optimal test option(s) based on clinical or family history, with assistance*	● Recommends urine organic acid testing for a patient with metabolic acidosis and hyperammonemia
Evaluates a specimen for pre-analytic errors, with assistance	● Recognizes that a referring laboratory frequently sends specimens of insufficient quantity and contacts send-out staff to discuss
Suggests modifications to existing laboratory workflows based on clinical need, with assistance	● Proposes a new workflow for short turnaround time (STAT) specimens ● Suggests adding another testing batch to the weekly schedule to accommodate an increase in test volume
Level 4 *Independently recommends the optimal test(s) based on clinical or family history*	● Recommends urine organic acid testing for a patient with metabolic acidosis and hyperammonemia
Independently evaluates a specimen for pre-analytic errors and identifies possible resolutions	● Recognizes that a referring laboratory frequently sends samples of insufficiency quantity and, working with laboratory supervisor, contacts send-out staff to discuss
Independently suggests modifications to existing laboratory workflows based on clinical need	● Suggests adding another testing batch to the weekly schedule to accommodate an increase in test volume

Level 5 *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*	● Creates an EHR pop-up box for providers to consider ordering plasma amino acids for a chief complaint of encephalopathy ● Creates a Laboratory Information System (LIS) alert for specimens received beyond the acceptable time window ● Creates a visual aide for the accessioning staff to help them determine if a specimen was collected in the correct tube ● Creates a new workflow to immediately notify the supervisor when a STAT specimen is received
Assessment Models or Tools	● Direct observation ● LIS audit ● Sample log ● Send-out test audit ● Simulation
Curriculum Mapping	●
Notes or Resources	● American Board of Medical Genetics and Genomics (ABMGG). Learning guides. http://www.abmgg.org/pages/program_learning.shtml. Accessed 2019. ● Clinical and Laboratory Standards Institute (CLSI). CLSI eCLIPSE ultimate access. http://clsi.edaptivedocs.biz/Login.aspx. Accessed 2019. ● College of American Pathologists (CAP). Checklists www.cap.org. Accessed 2019. ● American College of Medical Genetics and Genomics (ACMG). Laboratory Standards and Guidelines. https://www.acmg.net/. Accessed 2019. ● New York Department of Health (NYDOH). Laboratory standards. https://www.wadsworth.org/regulatory/clep/clinical-labs/laboratory-standards. Accessed 2019.

Patient Care 2: Analytic **Overall Intent:** To understand the analytic components of the assays performed in the lab and demonstrate competency in analysis	
Milestones	**Examples**
Level 1 *Describes basic principles for major assays performed in the laboratory* *Describes quality control (QC) in the clinical laboratory*	● Discusses the underlying principles of amino acid analysis ● Identifies the need for appropriate positive and negative controls for organic acid analysis
Level 2 *Performs assays, with substantial assistance* *Identifies QC failures*	● Performs amino acid analysis following standard operating procedures ● Recognizes when quality control results are outside of established limits
Level 3 *Performs assays, with minimal assistance* *Explains possible sources of QC failures*	● Performs organic acid analysis following the standard operating procedures ● Explains how contamination can be a source of QC failures
Level 4 *Independently performs assays* *Investigates QC failures and proposes resolution*	● Demonstrates competency in acylcarnitine analysis ● Identifies mobile phase contamination as a possible source of quality control failure and replaces the reagent
Level 5 *Develops new, alternate, or improved assay* *Identifies and implements a new QC approach for a clinical test*	● Modifies an existing liquid chromatography–mass spectrometry (LC-MS/MS) assay to analyze underivatized compounds ● Incorporates a new qualifier ion for peak identification in an established mass spectrometry-based assay
Assessment Models or Tools	● Direct observation ● Lab-specific competency assessment ● Trend reports
Curriculum Mapping	●
Notes or Resources	● ABMGG Learning Guides ● CAP checklists www.cap.org. Accessed 2019. ● CLSI. http://clsi.edaptivedocs.biz/Login.aspx. Accessed 2019. ● ACMG. Medical Genetics Practice Resources. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Practice_Resources/ACMG/Medical-Genetics-Practice-Resources/Medical-Genetics-Practice-Resources.aspx?hkey=d56a0de8-cfb0-4c6e-bf1e-ffb96e5f86aa. Accessed 2019.

	● CPHG (Wiley publisher). https://currentprotocols.onlinelibrary.wiley.com/journal/19348258. Accessed 2019.

Patient Care 3: Post-Analytic Skills **Overall Intent:** To provide clinically significant interpretation of lab results to effectively guide patient care	
Milestones	**Examples**
Level 1 *Identifies normal results* *Discusses the importance of patient's clinical history to test interpretation*	● Correctly identifies a normal amino acid result ● Recognizes that dietary intake can impact amino acid levels
Level 2 *Interprets simple results, with assistance* *Gathers pertinent elements of the clinical history to aid in interpretation*	● Correctly interprets that a large elevation of phenylalanine could indicate phenylketonuria (PKU) ● Identifies medium-chain triglycerides (MCT) oil supplementation as a possible cause of medium-chain dicarboxylic aciduria
Level 3 *Interprets complex results, with assistance* *Integrates results with the clinical history to develop a final interpretation, with assistance*	● Identifies an abnormal organic acid pattern consistent with propionic acidemia ● Interprets the significance of hypermethioninemia in a patient with liver disease
Level 4 *Independently interprets results* *Independently integrates results with the clinical history to develop a final interpretation*	● Correctly identifies an acylcarnitine pattern consistent with very-long-chain acyl-CoA dehydrogenase (VLCAD) deficiency ● Integrates newborn screening, amino acid, organic acid, and blood chemistry results in a patient with likely cobalamin C disease, with recommendation for molecular confirmation
Level 5 *Develops an improved result interpretation workflow* *Identifies novel correlations between results and clinical history*	● Develops an automated process for organic acid review and interpretation ● Identifies a new biomarker associated with mitochondrial disease
Assessment Models or Tools	● Direct observation ● Multisource feedback ● Medical record (chart) audit ● Report review
Curriculum Mapping	●
Notes or Resources	● ABMGG. Training & Certification Learning Guides. http://www.abmgg.org/pages/program_learning.shtml. Accessed 2019. ● Genetic Databases, e.g., https://www.ncbi.nlm.nih.gov/omim. ● Textbooks

	● ACMG. Technical Standards and Guidelines. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx. Accessed 2019. ● CAP checklists www.cap.org. Accessed 2019.

Patient Care 4: Reports **Overall Intent:** To generate effective clinical genetics reports for both simple and complex cases while using accurate terminology/nomenclature and providing appropriate recommendations	
Milestones	**Examples**
Level 1 *Identifies the elements of a laboratory report* *Identifies that reports can be revised*	● In a report identifies sample type, date of collection, test name, clinical indication, test results, and interpretation as key elements of a report ● Identifies that a typographical error can be corrected after a report is finalized
Level 2 *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 normal plasma amino acids ● Identifies that a wrong date of birth requires a corrected report ● Identifies that a change in variant classification requires an amended report and possible re-contacting of providers ● Identifies that additional test results require an addended report
Level 3 *Drafts a report for complex cases using accurate terminology/ nomenclature, with assistance* *Drafts a revised report, with assistance*	● Drafts a report for findings of elevated valine, isoleucine, and leucine (with normal alloisoleucine) as being suggestive of maple syrup urine disease (MSUD) versus catabolic state ● Drafts amended acylcarnitine profile report with findings of elevated C5OH to clarify its significance based on subsequent organic acid results
Level 4 *Independently generates concise reports for complex cases* *Independently generates a revised report*	● Independently generates an acylcarnitine report for findings consistent with glutaric acidemia type II that clearly identifies glutaric acidemia type II as the ultimate diagnosis ● Independently generates a report for findings of elevated valine, isoleucine, and leucine (with normal alloisoleucine) in a patient with ketosis as being consistent with catabolic state ● Independently generates an addended report to include results of additional testing ● Independently generates an addended acylcarnitine profile report with findings of elevated C5OH to clarify the maternal origin of the abnormality following testing of the mother
Level 5 *Develops a new reporting template for original or revised reports*	● Develops a template to integrate findings of acylcarnitine profile and urine organic acids into a single interpretation ● Creates an automated process for correcting reports
Assessment Models or Tools	● Direct observation ● Lab-specific competency assessment ● Multisource feedback

	● Review of reports at sign-out
Curriculum Mapping	●
Notes or Resources	● ABMGG. Training & Certification Learning Guides. http://www.abmgg.org/pages/program_learning.shtml. Accessed 2019. ● CLSI. ECLIPSE. http://clsi.edaptivedocs.biz/Login.aspx. Accessed 2019. ● CAP checklists www.cap.org. Accessed 2019. ● ACMG. Technical Standards and Guidelines. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx. Accessed 2019.

Medical Knowledge 1: Foundations of Medical Genetics and Genomics **Overall Intent:** To progressively incorporate basic science knowledge into patient care	
Milestones	**Examples**
Level 1 *Defines relevant medical terminology* *Describes basic principles of medical biochemistry and cell biology*	● Demonstrates understanding of common signs and symptoms of genetic conditions ● Describes how the urea cycle detoxifies ammonia ● Describes the role of the Krebs cycle in energy metabolism
Level 2 *Describes genetic conditions using accurate medical terminology* *Describes normal metabolic processes*	● Explains that hypoglycemia, metabolic acidosis, lethargy, and encephalopathy can be signs and symptoms of organic acidemias ● Describes the process of fatty acid beta-oxidation
Level 3 *With assistance, assimilates and integrates medical information to generate a differential diagnosis* *Describes abnormal metabolic processes and clinical phenotypes of associated diseases*	● Recognizes that the clinical presentation can differentiate glycine encephalopathy from propionic acidemia for a patient with elevated plasma glycine ● Describes the biochemical and clinical findings of pyridoxine dependent epilepsy
Level 4 *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*	● Recognizes that the clinical presentation can help to differentiate glycine encephalopathy from propionic acidemia for a patient with elevated plasma glycine ● Describes the etiology and clinical consequences of hyperammonemia in a neonate with methylmalonic acidemia
Level 5 *Mentors others in the process of integration of clinical and laboratory findings to generate a differential diagnosis*	● Compiles a collection of training cases to facilitate teaching how to integrate clinical and laboratory findings
Assessment Models or Tools	● Didactic courses exams (if applicable) ● Direct observation ● Faculty and staff member evaluations ● In-training exam
Curriculum Mapping	●
Notes or Resources	● Gene Reviews. https://www.ncbi.nlm.nih.gov/books/NBK1116/. Accessed 2019. ● ABMGG. Training & Certification Learning Guides. http://www.abmgg.org/pages/program_learning.shtml. Accessed 2019.

Medical Knowledge 2: Testing **Overall Intent:** To understand how to critically evaluate test methodologies and the steps needed to design, assess, and validate a new laboratory test	
Milestones	**Examples**
Level 1 *Defines the basic components of a test validation and verification* *Describes various methodologies used in a clinical biochemical genetics laboratory*	● Defines sensitivity, specificity, positive/negative predictive values, and reproducibility ● Describes the basic principles of chromatography
Level 2 *Describes methods and data used in establishing test performance characteristics for validation and verification* *Describes the strengths and limitations of a biochemical laboratory test*	● Identifies the need for appropriate known normal and abnormal specimens for determining the test reference range or precision ● Explains why acylcarnitine profiling is highly sensitive to elevations, but cannot distinguish between isomers
Level 3 *Determines test performance characteristics, with assistance* *Determines the optimal technologies and platforms for biochemical tests, with assistance*	● Calculates sensitivity, specificity, positive/negative predictive value, and reproducibility ● Identifies that a chromatography step is needed to differentiate between leucine, isoleucine, and alloisoleucine prior to detection and quantification by mass spectrometry
Level 4 *Independently calculates test performance characteristics* *Independently determines the optimal technologies and platforms for biochemical tests*	● Calculates sensitivity, specificity, positive/negative predictive value and reproducibility ● Selects a column for chromatography to differentiate between leucine, isoleucine, and alloisoleucine prior to detection and quantification by mass spectrometry
Level 5 *Designs a test validation/verification and establishes QC metrics* *Independently designs a new test for a genetic condition*	● Designs and validates a tandem mass spectrometry-based method for mucopolysaccharides ● Designs a test validation strategy for plasma to be an acceptable specimen for a laboratory test that is currently only being performed on fibroblasts ● Designs a test to analyze a newly developed biomarker
Assessment Models or Tools	● Direct observation ● In-training exam and/or in-house exam ● Portfolio
Curriculum Mapping	●

Notes or Resources	● ACMG. Technical Standards and Guidelines. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx. Accessed 2019. ● ACMG. Laboratory standards and guidelines: clinical biochemical genetics. https://www.acmg.net/PDFLibrary/Standards-Guidelines-Clinical-Biochemical-Genetics.pdf 2019. ● CDC. Morbidity and Mortality Weekly Report. Good laboratory practices for biochemical genetic testing and newborn screening for inherited metabolic disorders. https://www.cdc.gov/mmwr/pdf/rr/rr6102.pdf 2019.

Systems-based Practice 1: Patient Safety and Quality Improvement (QI) **Overall Intent:** To engage in the analysis and management of patient safety events, including relevant communication with patients, families, and health care professionals/clients; to conduct a QI project	
Milestones	**Examples**
Level 1 *Describes common patient safety events*	● Lists patient misidentification and compromised specimens as common patient safety events
Identifies that processes exist for reporting patient safety events	● Describes how to report, using the institutional reporting systems, that a patient's sample was compromised
Demonstrates knowledge of QI concepts	● Describes fishbone tool, Plan-Do-Study-Act (PDSA) cycle, measures of change (process, outcome), run charts, root cause analysis
Level 2 *Identifies system factors that lead to patient safety events*	● Recognizes that institutional courier delays may compromise specimen quality
Defines the institutional process of safety reporting systems	● Using the institutional reporting system for patient safety events, reports an event that patient's sample was compromised
Describes laboratory QI initiatives	● Examines existing processes/standard operating procedures to identify areas that can be improved to prevent mishandling of patient samples
Level 3 *Participates in analysis of patient safety events (simulated or actual)*	● Participates in the preparation of a laboratory non-conformance report that includes patients' risk assessment
Given a safety-related concern, can describe the risk management process of disclosing such events to the appropriate individuals	● Through simulation, communicates with provider/client about a misplaced sample and recommends remedy options
Participates in laboratory QI initiatives	● Participates in root cause analysis of a compromised patient sample
Level 4 *Conducts analysis of patient safety events and offers error prevention strategies (simulated or actual)*	● Collaborates with quality specialists or supervisor to conduct an analysis of a non-conformance event resulting in a compromised specimen, proposes a solution, and effectively communicates with provider/client about such event
Reports patient safety events (simulated or actual event)	● Participates in a QI project to decrease specimen mishandling occurrence within the institution

Demonstrates the skills required to identify, develop, implement, and analyze a QI project	
Level 5 *Actively engages teams and processes to modify systems to prevent patient safety events* *Develops innovative protocols to detect and report safety events* *Develops and assesses QI initiatives at the institutional or community level*	● Assumes a leadership role at the departmental or institutional level for patient safety ● Conducts a simulation or internal mock challenge for early identification of patient safety risk ● Initiates and completes a QI project to decrease the ordering of unnecessary tests in order to decrease health care costs
Assessment Models or Tools	● Direct observation ● Medical record (chart) audit ● Multisource feedback ● Portfolio ● Self-reflection ● Simulation
Curriculum Mapping	●
Notes or Resources	● Institute of Healthcare Improvement http://www.ihi.org/Pages/default.aspx. Accessed 2019. ● CMS. How to Use the Fishbone Tool for Root Cause Analysis. https://www.cms.gov/medicare/provider-enrollment-and-certification/qapi/downloads/fishbonerevised.pdf. Accessed 2019.

Systems-Based Practice 2: System Navigation for Patient-Centered Care **Overall Intent:** To effectively navigate the health care system, including the interdisciplinary team and other care providers, to adapt care to a specific patient population to ensure high-quality patient outcomes	
Milestones	**Examples**
Level 1 *Identifies the importance of coordinating care* *Explains the importance of effective transitions of care and hand-offs*	● Understands flow of lab information and responsibilities of specific staff members ● Understands the laboratory and clinical service integration in order to effectively communicate client needs ● Understands that when preparing to go on vacation, it is important to communicate details of pending cases to the covering laboratory fellow/staff and laboratory director
Level 2 *Describes effective care coordination with the clinical and laboratory team* *Describes examples of safe and effective transitions of care/hand-offs*	● Documents communication regarding ongoing cases when handing off pagers or ending a rotation ● Describes the communication for a pending critical specimen/case
Level 3 *Coordinates care with the clinical and laboratory team, with assistance* *Performs safe and effective transitions of care/hand-offs*	● Communicates critical results directly to providers ● Emails successor when rotating off service about an instrument problem and the steps taken to resolve
Level 4 *Coordinates care with the clinical and laboratory team* *Models and advocates for safe and effective transitions of care/hand-offs*	● Communicates critical results directly to providers ● Coordinates multiple test requests on a single specimen ● Prior to going on vacation, proactively prepares a plan and informs the covering fellow/resident/staff members about pending tests for critical patients
Level 5 *Improves quality of transitions of care within and across health care delivery systems to optimize patient outcomes*	● Develops a protocol for transitioning specimen/case status among laboratory fellows/residents/staff members
Assessment Models or Tools	● Direct observation ● Multisource feedback ● Review of sign-out tools, use and review of checklists ● Self-reflection
Curriculum Mapping	●
Notes or Resources	● Laboratory standard operating procedures. ● CAP checklists. www.cap.org. Accessed 2019. ● Kaplan KJ. In pursuit of patient-centered care. 2016. http://tissuepathology.com/2016/03/29/in-pursuit-of-patient-centered-care/#axzz5e7nSsAns. Accessed 2019.

Systems-Based Practice 3: Laboratory Geneticist's Role in Health Care System **Overall Intent:** To understand the resident's role in the complex health care system and how to optimize the system to improve patient care and the health system's performance	
Milestones	**Examples**
Level 1 *Identifies key components of the health care system* *Identifies basic types of medical reimbursement*	● Identifies systems and providers involved in test ordering and payment ● Recognizes that samples collected in the intensive care unit (ICU) versus outpatient clinic may have different priorities ● Demonstrates basic knowledge of how a laboratory is reimbursed for testing
Level 2 *Describes how components of a health care system are interrelated, and how this impacts patient care* *Identifies testing documentation related to billing and reimbursement* *Describes the financial components related to the laboratory operation*	● Understands the impact of health plans on testing workflow and reimbursement ● Identifies the importance of international classification of diseases (ICD)/common procedural technology (CPT) code for insurance billing ● Explains the types of direct and indirect costs
Level 3 *Collaborates with the other members of the health care system, with assistance* *Identifies opportunities for cost-effective patient care* *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)*	● Contacts ordering provider when requisition is missing information or inappropriate tests are ordered ● Reviews worksheets to identify cases of duplicate testing ● Gathers vendor quotes for a new lab purchase to minimize test reagent needs and to reduce unnecessary expenses
Level 4 *Independently collaborates with the other members of the health care system* *Practices cost-effective patient care* *Independently drafts the assessment of a laboratory fiscal metric for director review*	● Contacts ordering provider to suggest alternate test for optimal patient care ● Meets with other members of the health care team to improve testing algorithms for specific clinical indications ● Gives an in-service presentation to the clinical team describing best test-ordering practices to optimize cost-effective care ● Meet with vendors to determine if volume purchasing can result in a price discount ● Evaluates trends in monthly test volumes

Level 5 *Advocates for or leads systems change that enhances high-value, efficient, and effective patient care* *Participates in a local or national committee related to fiscal issues in genetic testing*	● Works with EHR staff to create a pop-up box to confirm an order that may be inappropriate based on patient's age or time of last collection ● Serves as a member of the state newborn screening oversight committee ● Serves as a member of the laboratory utilization committee
Assessment Models or Tools	● Direct observation ● Medical record (chart) audit
Curriculum Mapping	●
Notes or Resources	● The Kaiser Family Foundation: Topic: health reform https://www.kff.org/topic/health-reform/. Accessed 2019. ● Dzau VJ, McClellan M, Burke S, et al. Vital directions for health and health care: priorities from a National Academy of Medicine Initiative. March 2016. https://nam.edu/vital-directions-for-health-health-care-priorities-from-a-national-academy-of-medicine-initiative/. Accessed 2019. ● American Board of Internal Medicine. QI/PI activities. http://www.abim.org/maintenance-of-certification/earning-points/practice-assessment.aspx. Accessed 2019. ● PLUGS ● PharmGKB. https://www.pharmgkb.org/. Accessed 2019. ● CPT and/or ICD Coding manuals

Systems-Based Practice 4: Accreditation, Compliance, and Quality Management **Overall Intent:** To gain in-depth knowledge of the components of laboratory accreditation, regulatory compliance, and quality management	
Milestones	**Examples**
Level 1 *Identifies laboratory accreditation and licensing agencies* *Defines terminology related to laboratory quality*	● Describes the roles of College of American Pathologists (CAP) and Clinical Laboratory Improvement Amendments (CLIA) in clinical testing ● Defines the terms quality management, quality assurance, quality control, quality improvement, and proficiency testing
Level 2 *Describes the accreditation process, its requirements, and necessary documentation* *Interprets quality data and charts and trends, including proficiency testing results, with assistance*	● Describes regulatory requirements and compliance ● Evaluates Levey-Jennings charts for plasma amino acids ● Evaluates daily instrument quality control and proficiency testing result summaries
Level 3 *Participates in review of laboratory practice to assure compliance with accreditation requirements* *Independently evaluates quality indicators, including proficiency testing results*	● Participates in departmental quality assurance/quality control meetings, conferences, and accreditation/regulatory summation meetings ● Evaluates Levey-Jennings charts for plasma amino acids ● Evaluates monthly and daily instrument quality control and proficiency testing reports
Level 4 *Actively participates in the laboratory self-inspection* *Formulates a response for a proficiency test failure (actual or simulated)*	● Performs mock or self-inspections using a CAP checklist ● Assists in developing a strategy for handling quality control or proficiency testing failures
Level 5 *Participates in the inspection of an external laboratory* *Reviews the quality management plan to identify areas for improvements*	● Participates an inspection of another laboratory ● Serves on a committee for a regional or national accreditation agency ● Identifies alternate assessment options for laboratory proficiency testing
Assessment Models or Tools	● Audit of proficiency testing response draft ● Audit of quality control reviews ● Direct observation ● Documentation of inspector training and participation in fellow portfolio ● Planning and completion of quality improvement projects ● Presentation reviews

	● Multisource feedback
Curriculum Mapping	●
Notes or Resources	● ABMGG. Training & Certification Learning Guides. http://www.abmgg.org/pages/program_learning.shtml. Accessed 2019. ● CLSI. http://clsi.edaptivedocs.biz/Login.aspx. Accessed 2019. ● CAP checklists. www.cap.org. Accessed 2019. ● ACMG. Technical Standards and Guidelines. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx. Accessed 2019. ● CMS. CLIA. https://www.cms.gov/Regulations-and-Guidance/Legislation/CLIA/index.html?redirect=/CLIA/05_CLIA_Brochures.asp. Accessed 2019. ● CDC. Good Laboratory Practices for Biochemical Genetic Testing and Newborn Screening for Inherited Metabolic Disorders. https://www.cdc.gov/mmwr/pdf/rr/rr6102.pdf. Accessed 2019. ● CAP. Inspector Training. https://www.cap.org/laboratory-improvement/accreditation/inspector-training. Accessed 2019.

Systems-Based Practice 5: Informatics **Overall Intent:** To be able to collect, manage, use, and share data and information to support the delivery of accurate, high-quality health care and promote optimal patient outcomes	
Milestones	**Examples**
Level 1 *Demonstrates familiarity with basic technical concepts of hardware, operating systems, databases, and software for general purpose applications*	● Logs into institutional systems
Level 2 *Identifies laboratory specific software, key technical concepts, interfaces, workflow, barcode application, and automation systems (enterprise systems architecture)*	● Describes laboratory information systems and other interfaced systems and their roles in laboratory operations and health care delivery
Level 3 *Discusses laboratory initiatives based on informatics (system implementation and configuration)*	● Explains the role and responsibility of laboratory geneticists with regard to selection, oversight, and use of informatics systems in the laboratory
Level 4 *Applies informatics tools as needed in laboratory initiatives (e.g., data management and security, computational statistics, information governance)*	● Uses computational statistics to identify turnaround time outliers ● Retrospectively reviews large data set to confirm/establish reference ranges
Level 5 *Proposes medical informatics improvements for the operation of the laboratory*	● Identifies and resolves issues, potential problems, and challenges in EHR handling of genetic test results
Assessment Models or Tools	● Direct observation: how residents reflect their knowledge of laboratory information systems components in the health care system in the care of patients ● Portfolio of completed projects ● Publication and presentation record ● Training on clinical genetic data analysis software systems
Curriculum Mapping	●
Notes or Resources	● ACMG. Technical Standards and Guidelines. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx. Accessed 2019. Institutional training for site-specific policies and procedures

Practice-Based Learning and Improvement 1: Evidence-Based Practice **Overall Intent:** To incorporate evidence into clinical practice	
Milestones	**Examples**
Level 1 *Demonstrates how to access and select applicable evidence*	● Accesses ACMG Laboratory Standards and Guidelines ● Performs a search on PubMed to address a clinical question
Level 2 *Identifies and applies the best available evidence and/or clinical laboratory standards/guidelines to guide diagnostic evaluation of simple cases*	● Applies ACMG Laboratory Standards and Guidelines to the interpretation of a biotinidase test result
Level 3 *Identifies and applies the best available evidence and/or clinical laboratory standards/guidelines to guide diagnostic evaluation of complex cases*	● Uses published tables, textbooks or Human Metabolome Database to help interpret an elevated organic acid that may be dietary in nature
Level 4 *Critically appraises and applies evidence to guide lab-based recommendations, even in the face of conflicting data*	● Assesses the primary literature when evaluating the significance of decreased alpha-iduronidase activity in an apparently healthy patient
Level 5 *Mentors others to critically appraise and apply evidence for complex cases; and/or participates in the development of laboratory standards/guidelines*	● As part of a team, develops a diagnostic protocol for the confirmatory approach to a newly added newborn screening marker based on available evidence
Assessment Models or Tools	● Direct observation ● Oral or written examinations ● Presentation evaluation ● Review of drafted reports ● Scholarly portfolio
Curriculum Mapping	●
Notes or Resources	● U.S. National Library of Medicine. PubMed Tutorial. https://www.nlm.nih.gov/bsd/disted/pubmedtutorial/cover.html. Accessed 2019. ● ClinGen. https://clinicalgenome.org/. Accessed 2019. ● ACMG. Technical Standards and Guidelines. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx. Accessed 2019.

Practice-Based Learning and Improvement 2: Reflective Practice and Commitment to Personal Growth **Overall Intent:** To seek clinical performance information with the intent to improve care; to reflect on all domains of practice, personal interactions, and behaviors, and their impact on colleagues and patients (reflective mindfulness); to develop clear objectives and goals for improvement in some form of a learning plan	
Milestones	**Examples**
Level 1 *Realizes responsibility for personal and professional development by establishing goals* *Identifies the gap(s) between expectations and actual performance* *Actively seeks opportunities to improve*	● Adopts the ACGME Milestones as personal study guide to perform periodic self-assessment in one or multiple areas ● Uses ABMGG Learning Guides to identify gaps in knowledge ● Identifies mentors for personal and career development ● Asks for input from program director
Level 2 *Demonstrates willingness to receiving performance data and feedback in order to inform goals* *Analyzes and reflects on the factors which contribute to gap(s) between expectations and actual performance* *Designs and implements a learning plan, with assistance*	● Identifies areas for improvement using the ACGME Milestones when performing periodic self-assessment in one or multiple areas ● Assesses time management skills to achieve competence in a laboratory process ● Works with mentors to create or adapt a career development plan and to seek additional professional and personal growth opportunities ● When prompted, develops individual learning plan to improve skills in evaluating urine organic acid chromatograms
Level 3 *Seeks performance data and feedback with respect* *Institutes behavioral change(s) to narrow the gap(s) between expectations and actual performance* *Independently creates and implements a learning plan*	● Performs a monthly review of learner's case interpretations with the program director ● Executes plans for improvement in weak areas/gaps identified when using the ACGME Milestones or ABMGG Learning Guides to perform periodic self-assessment ● Executes their career development plan with mentor(s) and self-monitors progress periodically
Level 4 *Models appropriate and thoughtful seeking and consideration of feedback*	● Establishes a monthly review of learner's case interpretations with the program director and faculty members, and encourages others to do the same

Critically evaluates the effectiveness of behavioral changes in narrowing the gap(s) between expectations and actual performance *Uses performance data to measure the effectiveness of the learning plan and improves it when necessary*	● Seeks additional rotation(s) in weak areas/gaps identified when using the ACGME Milestones and ABMGG Learning Guides ● Uses in-training exam scores and monthly case reviews to ensure readiness for independent practice
Level 5 *Coaches others in personal and professional development* *Facilitates the design and implementation of learning plans for others*	● Coaches first-year fellows in how to use the ABMGG Learning Guides ● Assists first-year fellows in developing their individualized learning plans
Assessment Models or Tools	● Direct observation ● Multisource feedback ● Review of learning plan ● Self-reflection
Curriculum Mapping	●
Notes or Resources	● Hojat M, Veloski JJ, Gonnella JS. Measurement and correlates of physicians' lifelong learning. *Acad Med.* 2009;84(8):1066-74. *Contains a validated questionnaire about physician lifelong learning.* ● Burke AE, Benson B, Englander R, Carraccio C, Hicks PJ. Domain of competence: practice-based learning and improvement. *Acad Pediatr.* 2014;14:S38-S54. ● American Board of Medical Genetics and Genomics (ABMGG). Learning guides. http://www.abmgg.org/pages/program_learning.shtml. Accessed 2019.

Professionalism 1: Professional Behavior and Ethical Principles **Overall Intent:** To recognize and address lapses in ethical and professional behavior, demonstrate ethical and professional behaviors, and use appropriate resources for managing ethical and professional dilemmas	
Milestones	**Examples**
Level 1 *Demonstrates knowledge of the ethical principles underlying laboratory testing* *Describes how to report professionalism lapses, including strategies for addressing common barriers*	● Understands that being tired can contribute to lapses in professionalism ● Understands that being late can have an adverse effect on patient care and on professional relationships ● Articulates how the principle of "do no harm" applies to laboratory testing
Level 2 *Analyzes straightforward situations using ethical principles* *Demonstrates insight into professional behavior in routine situations; takes responsibility for own professionalism lapses*	● Refrains from discussing a case when in public places ● Notifies appropriate supervisor when another trainee appears to be impaired
Level 3 *Recognizes the need for and uses appropriate resources to seek solutions in managing and resolving complex ethical situations* *Demonstrates professional behavior in complex or stressful situations*	● After noticing a colleague's inappropriate social media post, reviews policies related to posting of content and seeks guidance ● Remains calm when confronted by a provider who is upset or frustrated
Level 4 *Manages complex ethical situations* *Recognizes situations that may trigger professionalism lapses and intervenes to prevent lapses in self and others*	● Models respect for patients and promotes the same from colleagues when lab errors are identified and required correction ● When observing a faculty member being aggressive towards learners, identifies institutional resources for reporting and intervenes on the learner's behalf
Level 5 *Identifies and seeks to address system-level factors that introduce or exacerbate ethical problems or impede their resolution* *Coaches others when their behavior fails to meet professional expectations*	● Coaches others when their behavior fails to meet professional expectations and creates a performance improvement plan to prevent recurrence ● Engages laboratory staff to address delayed turnaround time to decrease patient and provider frustrations ● Creates a mini-course for laboratory staff members to address customer service concerns

Assessment Models or Tools	● Direct observation ● Global evaluation ● Multisource feedback ● Oral or written self-reflection ● Simulation
Curriculum Mapping	●
Notes or Resources	● American Medical Association Code of Ethics. https://www.ama-assn.org/delivering-care/ama-code-medical-ethics. Accessed 2019. ● Byyny RL, Papadakis MA, Paauw DS, Pfiel S, Alpha Omega Alpha. *Medical Professionalism Best Practices*. Menlo Park, CA: Alpha Omega Alpha Honor Medical Society; 2015. https://alphaomegaalpha.org/pdfs/2015MedicalProfessionalism.pdf. Accessed 2019. ● Levinson W, Ginsburg S, Hafferty FW, Lucey CR. *Understanding Medical Professionalism*. 1st ed. New York, NY: McGraw-Hill Education; 2014. https://accessmedicine.mhmedical.com/book.aspx?bookID=1058. Accessed 2019. ● Domen RE, Johnson K, Conran RM, et al. Professionalism in pathology: a case-based approach as a potential education tool. *Arch Pathol Lab Med.* 2017;141:215-219. https://doi.org/10.5858/arpa.2016-2017-CP. Accessed 2019. ● Bynny RL, Paauw DS, Papadakis MA, Pfeil S. *Medical Professionalism Best Practices: Professionalism in the Modern Era*. Menlo Park, CA: Alpha Omega Alpha Honor Medical Society; 2017. http://alphaomegaalpha.org/pdfs/Monograph2018.pdf. Accessed 2019.

Professionalism 2: Accountability and Conscientiousness **Overall Intent:** To take responsibility for one's own actions and the impact on patients and other members of the health care team	
Milestones	**Examples**
Level 1 *Responds promptly to instructions, requests, or reminders to complete tasks and responsibilities*	● Has timely attendance at laboratory meetings, grand rounds and clinical conferences ● Completes administrative tasks, safety training documentation, and procedure review by specified due date
Level 2 *Takes ownership and performs tasks and responsibilities in a timely manner*	● Completes evaluations by specified due date ● Completes assigned tasks before taking scheduled leave
Level 3 *Anticipates situations that may impact own ability to meet responsibilities and describes the impact on team*	● Notifies director of errors, testing delays, complex results that require more time ● Asks for assistance from director, lab staff members, or faculty members as needed ● Arranges coverage for assigned tasks and notifies appropriate individuals in preparation for scheduled leave
Level 4 *Shares responsibility for system outcomes as a member of the team*	● Takes responsibility for inadvertently omitting key diagnostic information from a report
Level 5 *Designs new strategies to ensure that the needs of patients, teams, and systems are met*	● After soliciting input from providers, designs a testing algorithm for a specific disease
Assessment Models or Tools	● Compliance with deadlines and timelines ● Direct observation ● Global evaluations ● Multisource feedback including co-workers, supervisors, and other health providers ● Self-evaluations ● Self-reflection
Curriculum Mapping	●
Notes or Resources	● ABMGG Professionalism Guidelines ● ACMG Code of Conduct ● Code of conduct from fellow/resident institutional manual ● Expectations of fellowship program regarding accountability and professionalism

Professionalism 3: Personal and Professional Well-Being **Overall Intent:** To identify, use, manage, improve, and seek help for personal and professional well-being for self and others	
Milestones	**Examples**
Level 1 *Describes common indicators of personal or professional well-being*	● With assistance, acknowledges own response to patient's fatal genetic diagnosis ● Recognizes the need for time away from work for personal care
Level 2 *Independently recognizes status of personal and professional well-being and seeks help when needed*	● Independently identifies and communicates impact of a personal family tragedy on one's ability to be productive ● Recognizes a pattern of agitation and works to handle appropriately
Level 3 *Proposes a plan to optimize personal and professional well-being*	● With a mentor, develops a reflective response to deal with personal impact of difficult cases and disclosures of abnormal results ● Proposes to start a book club for members of the laboratory team
Level 4 *Implements a plan to optimize personal and professional well-being*	● Independently identifies and implements ways to manage personal stress ● Starts a book club for members of the laboratory team
Level 5 *Coaches others to develop and implement plans to optimize personal and professional well-being*	● Assists in organizational efforts to address laboratorian well-being after an adverse patient outcome resulting from a laboratory error ● Helps junior fellows organize and implement a stress-relieving activity
Assessment Models or Tools	● Direct observation ● Group interview or discussions for team activities ● Individual interview ● Institutional online training modules ● Self-assessment and personal learning plan
Curriculum Mapping	●
Notes or Resources	● Local resources, including Employee Assistance, HR ● ACGME. Tools and Resources. https://www.acgme.org/What-We-Do/Initiatives/Physician-Well-Being/Resources. Accessed 2019. ● AAMC. Wellness. https://www.aamc.org/news-insights/wellbeing/faculty. Accessed 2019.

Interpersonal and Communication Skills 1: Patient- and Family-Centered Communication **Overall Intent:** To effectively communicate with the patient and family, to identify communication barriers including personal biases, and to organize and lead communication around shared decision making	
Milestones	**Examples**
Level 1 *Identifies common barriers to effective communication* *Identifies the role of the laboratory in communicating results to patients*	● Identifies that language (verbal and nonverbal cues) can be a barrier to effective communication ● Understands that laboratory reports may be viewed by patients ● Describes institutional policies and procedures for laboratory communication to patients
Level 2 *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*	● Identifies genetic literacy of patients as a barrier to communication ● Recognizes the benefit of diagrams and pictures to communicate information ● Remains attentive during a clinical encounter rather than focusing on electronic devices ● Arrives on time and remains present for the entire patient encounter
Level 3 *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*	● Reflects on one's personal bias against pregnancy termination and describes its potential influence on communicating prenatal results to patients and families ● Maintains eye contact and speaks directly to patients with intellectual disabilities
Level 4 *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*	● Creates an educational brochure to explain the need for ongoing testing in the management of PKU ● Explains urine organic acid results to a family of an infant diagnosed with isovaleric acidemia, under the supervision of an attending clinician

Level 5 *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*	● Serves on an institutional ethics committee ● Collaborates on enhancements to the patient portal to provide educational resources on laboratory test interpretation
Assessment Models or Tools	● Direct observation ● Kalamazoo Essential Elements Communication Checklist (Adapted) ● OSCE ● Self-assessment including self-reflection exercises ● Skills needed to set the state, Elicit information, Give information, Understand the patient, and End the encounter (SEGUE) ● Standardized patients
Curriculum Mapping	●
Notes or Resources	● Laidlaw A, Hart J. Communication skills: an essential component of medical curricula. Part I: Assessment of clinical communication: AMEE Guide No. 51. *Med Teach*. 2011;33(1):6-8. ● Makoul G. Essential elements of communication in medical encounters: The Kalamazoo consensus statement. *Acad Med*. 2001;76:390-393. ● Makoul G. The SEGUE Framework for teaching and assessing communication skills. *Patient Educ Couns*. 2001;45(1):23-34. ● Symons AB, Swanson A, McGuigan D, Orrange S, Akl EA. A tool for self-assessment of communication skills and professionalism in fellows. *BMC Med Educ*. 2009;9:1. ● Skotko BG, Capone GT, Kishnani PS, Postnatal Diagnosis of Down Syndrome: Synthesis of the Evidence on How Best to Deliver the News. *Pediatrics*. 2009;124(4):e751-8. doi:10.1542/peds.2009-0480. ● Skotko BG, Kishnani PS, Capone GT, Prenatal diagnosis of Down syndrome: how best to deliver the news. *Am J Med Genet A*. 2009;149A(11):2361-7.

Interpersonal and Communication Skills 2: Interprofessional and Team Communication **Overall Intent:** To effectively communicate with the health care and clinical laboratory team in both straightforward and complex situations	
Milestones	**Examples**
Level 1 *Uses language that values all members of the health care team, including clinical and laboratory professionals* *Describes the utility of constructive feedback*	● Acknowledges the contribution of each member of the laboratory team ● Identifies that constructive feedback benefits the entire team
Level 2 *Adapts communication style to fit team needs* *Solicits feedback on personal performance as a member of the laboratory team*	● Communicates diagnostic evaluation results clearly and concisely in an organized and timely manner ● Asks the laboratory team how they can improve explanation of test results ● Asks senior fellow how to improve timeliness for completion of assay
Level 3 *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*	● After a test has been interpreted, communicates with the primary care team to verify they have received and understand the results ● Shares information gained regarding the request for a STAT result with the lab team and confirms reporting of that result in a rapid manner ● Shares concerns that an instrument is not working properly with the laboratory supervisor
Level 4 *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*	● Asks other members of the health care team to repeat back recommendations to ensure understanding ● Participates in multidisciplinary meetings to discuss prioritization of testing in neonates requiring multiple biochemical tests when limited blood volume is available
Level 5 *Models flexible communication strategies that value input from all health care team members, resolving conflict when needed*	● Mediates a conflict resolution between different members of the laboratory team
Assessment Models or Tools	● Direct observation ● Multisource feedback ● Self-reflection ● Simulation

Curriculum Mapping	●
Notes or Resources	● Roth CG, Eldin KW, Padmanabhan V, Freidman EM. Twelve tips for the introduction of emotional intelligence in medical education. *Med Teach*. 2018;21:1-4. https://doi.org/10.1080/0142159X.2018.1481499. Accessed 2019. ● Green M, Parrott T, Cook G. Improving your communication skills. *BMJ*. 2012;344:e357. https://doi.org/10.1136/bmj.e357. Accessed 2019. ● Henry SG, Holmboe ES, Frankel RM. Evidence-based competencies for improving communication skills in graduate medical education: a review with suggestions for implementation. *Med Teach*. 2013;35(5):395-403. https://doi.org/10.3109/0142159X.2013.769677. Accessed 2019. ● Dehon E, Simpson K, Fowler D, Jones A. Development of the faculty 360. *MedEdPORTAL*. 2015;11:10174. http://doi.org/10.15766/mep_2374-8265.10174. Accessed 2019. ● Lane JL, Gottlieb RP. Structured clinical observations: a method to teach clinical skills with limited time and financial resources. *Pediatrics*. 2000;105:973-7. https://pediatrics.aappublications.org/content/pediatrics/105/Supplement_3/973.full.pdf. Accessed 2019. ● Braddock CH, Edwards KA, Hasenberg NM, Laidley TL, Levinson W. Informed decision making in outpatient practice: time to get back to basics. *JAMA*. 1999;282:2313-2320. https://doi.org/10.1001/jama.282.24.2313. Accessed 2019.

Interpersonal and Communication Skills 3: Communication within Health Care Systems **Overall Intent:** To ensure the fellow effectively communicates using a variety of modalities	
Milestones	**Examples**
Level 1 *Protects patient personal health information by following institutional policies* *Identifies institutional and departmental procedures for communication of issues*	● Shreds patient list after case conference ● Actively logs off the computer and keeps protected health information (written or electronic) locked at all times ● Identifies the location of the on-call schedule and emergency contact numbers ● Is able to find written policies in binders, computers, intranet or any institutional resources
Level 2 *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*	● Identifies that provider should be called or paged immediately when a critical result is identified ● Knows the chain of command and escalating procedures ● Contacts the service representative about an instrument malfunction
Level 3 *Effectively and securely communicates clinical information, with guidance* *Uses institutional structure to effectively communicate clear and constructive suggestions*	● Contacts provider when a critical result is identified ● Knows when to direct concerns locally, departmentally, or institutionally via appropriate escalation
Level 4 *Independently communicates clinical information* *Initiates conversations on difficult subjects with appropriate stakeholders to improve the system*	● Immediately contacts provider when a critical result is identified and appropriately documents the communication ● Organizes a discussion with clinical and laboratory stakeholders following the release of erroneous laboratory results
Level 5 *Models effective communication of clinical information* *Facilitates dialogue regarding systems issues among larger community stakeholders*	● Develops a simulation project to improve communication skills between junior fellow and ordering providers ● Develops an electronic barrier so that only authorized providers can review sensitive test results
Assessment Models or Tools	● Direct observation ● Medical record (chart) audit ● Multisource feedback ● Simulation

Curriculum Mapping	●
Notes or Resources	● Institutional policies and procedures ● Bierman JA, Hufmeyer KK, Liss DT, Weaver AC, Heiman HL. Promoting responsible electronic documentation: validity evidence for a checklist to assess progress notes in the electronic health record. *Teach Learn Med.* 2017;29(4):420-432. https://doi.org/10.1080/10401334.2017.1303385. Accessed 2019.