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Milestones Supplemental Guide

This document provides additional guidance and examples for the Laboratory Genetics and Genomics 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 targeted BRCA1 mutation testing for a patient where the familial variant is known
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 targeted BRCA1 mutation testing for a patient where the familial variant is known
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 an ordering algorithm in the EHR for familial BRCA1 testing
Creates a new protocol to assist the laboratory in the appraisal of specimen issues	 Creates a Laboratory Information System 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
Independently develops a new laboratory workflow	 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 Laboratory Information System 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. Molecular Diagnostics Textbooks, e.g., Molecular Diagnostics: Fundamentals, Methods, and Clinical Applications 3rd ed. by Buckingham PhD MB DLM(ASCP), Lela (Author)

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	Discusses the underlying principles of polymerase chain reaction (PCR) and karyotype analysis
Describes quality control (QC) in the clinical laboratory	•Identifies the need for appropriate positive and negative controls for PCR
Level 2 Performs assays, with substantial assistance	Performs PCR following the approved standard operating procedures
Identifies QC failures	Recognizes when quality control results are outside of established limits
Level 3 Performs assays, with minimal assistance	Performs karyotype analysis following the standard operating procedures
Explains possible sources of QC failures	 Explains how contamination can be a source of QC failures Explains how suboptimal probe hybridization is a source of QC failures
Level 4 Independently performs assays	Demonstrates competence in karyotype analysis
Investigates QC failures and proposes resolution	Identifies hybridization temperature as a source of QC failures and makes appropriate adjustments
Level 5 Develops new, alternate, or improved assay	Researches, designs, and implements a new cell enrichment protocol
Identifies and implements a new QC approach for a clinical test	Researches and implements an informatics QC in conjunction with the wet lab QC
Assessment Models or Tools	Direct observation
	 Lab-specific competency assessment Trend reports
Curriculum Mapping	•
Notes or Resources	ABMGG Learning Guides
	CAP checklists <u>www.cap.org</u> . Accessed 2019.
	CLSI. http://clsi.edaptivedocs.biz/Login.aspx . Accessed 2019.
	ACMG. Medical Genetics Practice Resources. https://www.acmg.net/ACMG/Medical- 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. • AGT Cytogenetics Laboratory Manual (Wiley publisher). https://onlinelibrary.wiley.com/doi/book/10.1002/9781119061199. 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	Correctly identifies a normal male or female karyotype Correctly identifies noncarrier for sickle cell anemia
Discusses the importance of patient's clinical history to test interpretation	Recognizes that a female karyotype for a male patient may not be due to a laboratory error
Level 2 Interprets simple results, with assistance	 Correctly identifies t(9;22) as a common finding in a patient with chronic myeloid leukemia (CML)
	 Correctly identifies a BRCA1 missense variant on DNA sequencing in a patient with familial breast cancer
Gathers pertinent elements of the clinical history to aid in interpretation	Recognizes that a female karyotype for a male patient may be due to transplant
Level 3 Interprets complex results, with assistance	Correctly identifies a BRCA1 indel variant on DNA sequencing in a patient with familial breast cancer
Integrates results with the clinical history to develop a final interpretation, with assistance	Incorporates bone marrow biopsy findings with karyotype and fluorescence in situ hybridization (FISH) results to diagnose acute myeloid leukemia (AML)
Level 4 Independently interprets results Independently integrates results with the clinical history to develop a final interpretation	Reconciles discordant karyotype and FISH results that suggest a myeloid disease process with pathology results that suggest a lymphoid disease process
Level 5 Develops an improved result interpretation workflow	Creates an enhancement in the bioinformatics pipeline for variant assessment
Identifies novel correlations between results and clinical history	Publishes a unique gene or pathogenic variant association for intellectual disability
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.

ACMG. Technical Standards and Guidelines. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx . Accessed 2019. CAP checklists https://www.acmg.net/ACMG/Medical-genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx . Accessed 2019.		Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx. Accessed 2019.
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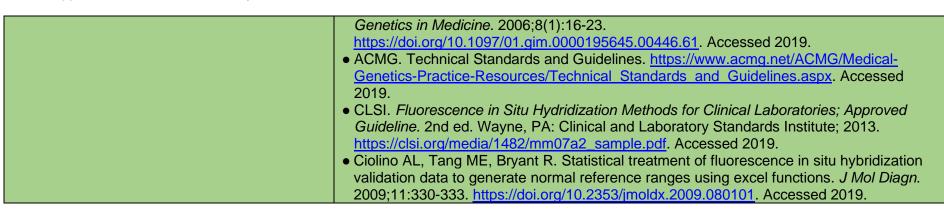
Patient Care 4: Reports	
Overall Intent: To generate effective clinical genetics reports for both simple and complex cases while using accurate terminology/nomenclature and providing appropriate recommendations	
Milestones	Examples
Level 1 Identifies the elements of a laboratory report	In a report identifies sample type, date of collection, test name, clinical indication, test results, and interpretation as key elements of a report
Identifies that reports can be revised	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	Drafts a report for normal chromosomal microarray (CMA)
Identifies when to correct, amend, or addend a report based on the type of alteration required	 Identifies that a wrong date of birth requires a corrected report Identifies that a change in variant classification requires an amended report and possible
	recontacting 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 report for CMA with both terminal copy number loss and terminal copy number gain that suggests a possible unbalanced translocation
Drafts a revised report, with assistance	 Drafts an amended report for nomenclature changes Drafts an addended report for additional test results
Level 4 Independently generates concise reports for complex cases	Generates a report for CMA with both terminal copy number loss and terminal copy number gain that suggests a possible unbalanced translocation and recommends additional testing
Independently generates a revised report	Generates an amended report that updates nomenclature Generates an addended report to include results of additional testing
Level 5 Develops a new reporting template for original or revised reports	 Develops a template to integrate the cytogenetic and molecular findings in complex cancer cases 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 <u>www.cap.org</u> . Accessed 2019.
	ACMG. Technical Standards and Guidelines. https://www.acmg.net/ACMG/Medical-
	Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx. Accessed
	2019.
	International System for Cytogenetic Nomenclature (ISCN)
	Human Genome Variation Society (HGVS)

Milestones	Examples
Level 1 Defines relevant medical terminology	Demonstrates understanding of most common clinical signs and symptoms of genetic conditions
Explains basic structure and function of the genome	Describes chromosome structure; also describes similarities and differences between DNA and RNA
	Describes mitosis and meiosis
Describes the basic mechanisms of germline	Describes Mendelian patterns of inheritance
and somatic disease development	Describes the two-hit hypothesis for cancer development
Level 2 Describes genetic conditions using accurate medical terminology	Describes features of common microdeletion syndromes
Identifies the basic genomic alterations	Describes basic types of genetic findings, such as aneuploidies, structural variants, single
underpinning abnormal lab results	nucleotide variants (SNV), copy number variations (CNV), and uniparental disomy
Recognizes the pathogenic mechanisms for	Explains non-Mendelian inheritance patterns
well-described conditions	Explains haploinsufficiency
Level 3 With assistance, assimilates and	Discusses the implications of consanguinity in a pedigree, the risk for autosomal
integrates medical information to generate a differential diagnosis	recessive conditions, and laboratory test results that may suggest consanguinity
Correlates genomic alterations and phenotypic information to guide laboratory testing and interpretation, with assistance	Recommends using methylation testing for Prader-Willi/Angelman syndrome (PWS/AS) versus FISH testing
Applies understanding of pathogenic mechanisms to interpretation of test results, with assistance	Discusses the role of a Robertsonian translocation in familial Down syndrome
Level 4 Independently assimilates and integrates medical information to generate a differential diagnosis	Based on apparent X-linked inheritance, identifies Duchenne and Emery-Dreifuss syndrome in the differential diagnosis for a boy with muscular dystrophy

Independently correlates genomic alterations and phenotypic information to guide laboratory testing and interpretation	Describes the occurrence of SNVs or CNVs involving <i>ELN</i> and their laboratory interpretation
Independently applies understanding of pathogenic mechanisms to interpretation of test results	Recognizes the clinical urgency to identify t(15;17) in a patient with acute promyelocytic leukemia (APML)
Level 5 Mentors others in the process of integration of clinical and laboratory findings to generate a differential diagnosis	Leads working groups with fellows/residents/graduate students to teach the process of developing a differential diagnosis
Presents a new clinical genomic/phenotypic correlation	Presents work at national meeting(s) and/or publishes work in peer-reviewed journal(s)
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. Textbooks

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 Level 1 Defines the basic components of a test validation and verification	• Defines sensitivity, specificity, positive/negative predictive values, and reproducibility
Describes various methodologies used in a clinical genetics laboratory	Identifies the procedures that require tissue culture and those that require nucleic acid extraction
Level 2 Describes methods and data used in establishing test performance characteristics for validation and verification	Identifies the need for appropriate known normal and abnormal specimens for determining the test reference range or precision
Describes the strengths and limitations of a genetic laboratory test	 Explains why Sanger sequencing may miss deletions Describes limitations for next-generation DNA sequencing
Level 3 Determines test performance characteristics, with assistance	Calculates sensitivity, specificity, positive/negative predictive value, and reproducibility
Determines the optimal technologies and platforms for genetic tests, with assistance	Identifies that a balanced rearrangement should be characterized by karyotyping rather than by CMA
Level 4 Independently calculates test performance characteristics	Calculates sensitivity, specificity, positive/negative predictive value and reproducibility
Independently determines the optimal technologies and platforms for genetic tests	• Identifies that atypical BCR-ABL1 fusions may yield discrepant results across karyotype, FISH, and PCR
Level 5 Designs a test validation/verification and establishes QC metrics	Designs a test validation strategy for plasma to be an acceptable specimen for a laboratory test that is currently only being performed on fibroblasts
Independently designs a new test for a genetic condition	Designs a test to detect multiple partners of a frequently rearranged gene
Assessment Models or Tools	Direct observation In-training exam and/or in-house exam Portfolio
Curriculum Mapping	•
Notes or Resources	• Wiktor AE, Van Dyke DL, Stupca PJ, Ketterling RP, Thorland EC, Shearer B., Dewald GW. Preclinical validation of fluorescence in situ hybridization assays for clinical practice.



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 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	Assumes a leadership role at the departmental or institutional level for patient safety
Develops innovative protocols to detect and report safety events	Conducts a simulation or internal mock challenge for early identification of patient safety risk
Develops and assesses QI initiatives at the institutional or community level	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/gapi/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	 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
Explains the importance of effective transitions of care and hand-offs	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	Documents communication regarding ongoing cases when handing off pagers or ending a rotation
Describes examples of safe and effective transitions of care/hand-offs	Describes the communication for a pending critical specimen/case
Level 3 Coordinates care with the clinical and laboratory team, with assistance	Communicates critical results directly to providers
Performs safe and effective transitions of care/hand-offs	Emails successor when rotating off service about an instrument problem and the steps taken to resolve
Level 4 Coordinates care with the clinical and	Communicates critical results directly to providers
laboratory team	Coordinates multiple test requests on a single specimen
Models and advocates for safe and effective transitions of care/hand-offs	Prior to going on vacation, proactively prepares a plan and informs the covering fellow/resident/staff 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 his/her 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 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
Identifies basic types of medical reimbursement	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	Understands the impact of health plans on testing workflow and reimbursement
Identifies testing documentation related to billing and reimbursement	Identifies the importance of international classification of diseases (ICD)/common procedural technology (CPT) code for insurance billing
Describes the financial components related to the laboratory operation	Explains the types of direct and indirect costs
Level 3 Collaborates with the other members of the health care system, with assistance	Contacts ordering provider when requisition is missing information or inappropriate tests are ordered
Identifies opportunities for cost-effective patient care	Reviews worksheets to identify cases of duplicate testing
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)	Gathers information regarding usage of stock reagents to determine if volumes are too high (reagent expires before use leading to waste)
Level 4 Independently collaborates with the other members of the health care system	Contacts ordering provider to suggest alternate test for optimal patient care
Practices cost-effective patient care	Meets with other members of the health care team to improve testing algorithms for specific clinical indications
Independently drafts the assessment of a laboratory fiscal metric for director review	 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	 Works with EHR staff to create a pop-up box displaying a patient's Cytochrome P450 2C9 (CYP2C9) alleles and dosing recommendations when ordering warfarin 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
Participates in a local or national committee related to fiscal issues in genetic testing	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. https://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 **Examples Milestones** Level 1 Identifies laboratory accreditation and • Describes the roles of College of American Pathologists (CAP) and Clinical Laboratory Improvement Amendments (CLIA) in clinical testing licensing agencies Defines terminology related to laboratory quality Defines the terms quality management, quality assurance, quality control, quality improvement, and proficiency testing Level 2 Describes the accreditation process, its • Describes regulatory requirements and compliance requirements and necessary documentation Assesses trends of harvest failure or growth failure cytogenetic/molecular testing Interprets quality data and charts and trends, including proficiency testing results, with Evaluates daily instrument quality control and proficiency testing result summaries assistance **Level 3** Participates in review of laboratory • Participates in departmental quality assurance/quality control meetings, conferences, and practice to assure compliance with accreditation accreditation/regulatory summation meetings requirements Independently evaluates quality indicators, Assesses harvest failure or growth failure for cytogenetic/molecular testing including proficiency testing results Evaluates monthly and daily instrument quality control and proficiency testing reports **Level 4** Actively participates in the laboratory • Performs mock or self-inspections using a CAP checklist self-inspection Formulates a response for a proficiency test Assists in developing a strategy for handling quality control or proficiency testing failures failure (actual or simulated) Level 5 Participates in the inspection of an • Participates an inspection of another laboratory external laboratory • Serves on a committee for a regional or national accreditation agency Reviews the quality management plan to • Identifies alternate assessment options for laboratory proficiency testing identify areas for improvements • Audit of proficiency testing response draft Assessment Models or Tools 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. <u>www.cap.orq</u> . 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-
	<u>Guidance/Legislation/CLIA/index.html?redirect=/CLIA/05_CLIA_Brochures.asp</u> . 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
Identifies the various resources including software, tools and databases used for classification of variants	Locates the most common web-based genomics resources
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
Uses resources to classify variants in the context of the phenotype, with assistance	Uses ACMG guideline, ClinGen (https://clinicalgenome.org/) or ClinVar to classify variants
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
Independently uses resources to perform routine variant classification using professional standards and guidelines	Classifies a CFTR variant according to current professional standards and guidelines
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
Independently uses resources to reclassify ambiguous variants	Uses available resources to reclassify a previously ambiguous CFTR variant
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

Contributes to the knowledge base for the refinement of ambiguous test results and	Participates on a national level to create standards and guidelines for variant classification
variant classifications	Publishes work on test result classification in a peer-reviewed journal
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	 UCSC. Genome browser. https://genome.ucsc.edu/training/. Accessed 2019. ClinGen. Training Modules. https://clinicalgenome.org/curation-activities/variant-pathogenicity/training-materials/. 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 1: Evidence-Based and Scholarship 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 guidelines for analyzing the correct number of cells and using appropriate cell culture conditions
Level 3 Identifies and applies the best available evidence and/or clinical laboratory standards/guidelines to guide diagnostic evaluation of complex cases	Uses ClinVar or other databases to guide results interpretation
Level 4 Critically appraises and applies evidence to guide lab-based recommendations, even in the face of conflicting data	 Assesses the primary literature to identify genotype-phenotype correlations or conditions associated with variable expressivity/reduced penetrance to guide the need for reflex or parental testing and to aid in the interpretation of a result
Level 5 Mentors others to critically appraise and apply evidence for complex cases; and/or participates in the development of laboratory standards/guidelines	Leads teaching on application of ACMG guidelines in the interpretation of variants of uncertain significance
Assessment Models or Tools	 Direct observation Oral or written examinations Presentation evaluation Review of drafted reports Scholarly portfolio
Curriculum Mapping	•
Notes or Resources	 US 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.

	mprovement 2: Reflective Practice and Commitment to Personal Growth formation with the intent to improve care; to reflect on all domains of practice, personal
	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	Adopts the ACGME Milestones as personal study guide to perform periodic self- assessment in one or multiple areas
Identifies the gap(s) between expectations and actual performance	Uses ABMGG Learning Guides to identify gaps in knowledge
Actively seeks opportunities to improve	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	Identifies areas for improvement using the ACGME Milestones when performing periodic self-assessment in one or multiple areas
Analyzes and reflects on the factors which contribute to gap(s) between expectations and actual performance	 Assesses time management skills to achieve competency in a laboratory process Works with mentors to create or adapt a career development plan and to seek additional professional and personal growth opportunities
Designs and implements a learning plan, with assistance	When prompted, develops individual learning plan to improve skills in variant classification
Level 3 Seeks performance data and feedback with respect	Performs a monthly review of learner's case interpretations with the program director
Institutes behavioral change(s) to narrow the gap(s) between expectations and actual performance	Executes plans for improvement in weak areas/gaps identified when using the ACGME Milestones or ABMGG Learning Guides to perform periodic self-assessment
Independently creates and implements a learning plan	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	Seeks additional rotation(s) in weak areas/gaps identified when using the ACGME Milestones and ABMGG Learning Guides
Uses performance data to measure the effectiveness of the learning plan and improves it when necessary	Uses in-training exam scores and monthly case reviews to ensure readiness for independent practice
Level 5 Coaches others in personal and professional development	Coaches first-year fellows in how to use the ABMGG Learning Guides
Facilitates the design and implementation of learning plans for others	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 Milestones	and professional dilemmas Examples
Level 1 Demonstrates knowledge of the ethical	Understands that being tired can contribute to lapses in professionalism
principles underlying laboratory testing	Understands that being late can have an adverse effect on patient care and on professional relationships
Describes how to report professionalism lapses, including strategies for addressing common barriers	Articulates how the principle of "do no harm" applies to laboratory testing
Level 2 Analyzes straightforward situations using ethical principles	Refrains from discussing a case when in public places
Demonstrates insight into professional behavior in routine situations; takes responsibility for own professionalism lapses	Notifies appropriate supervisor when another learner appears to be impaired
Level 3 Recognizes the need for and uses appropriate resources to seek solutions in managing and resolving complex ethical situations	After noticing a colleague's inappropriate social media post, reviews policies related to posting of content and seeks guidance
Demonstrates professional behavior in complex or stressful situations	Remains calm when confronted by a provider who is upset or frustrated
Level 4 Manages complex ethical situations	Models respect for patients and promotes the same from colleagues when lab errors are identified and required correction
Recognizes situations that may trigger professionalism lapses and intervenes to prevent lapses in self and others	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 and creates a performance improvement plan to prevent recurrence
Coaches others when their behavior fails to meet professional expectations	 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
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	 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. <i>Medical Professionalism Best Practices</i>. Menlo Park, CA: Alpha Omega Alpha Honor Medical Society; 2015. https://alphaomegaalpha.org/pdfs/2015MedicalProfessionalism.pdf. Levinson W, Ginsburg S, Hafferty FW, Lucey CR. <i>Understanding Medical Professionalism</i>. 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. <i>Arch Pathol Lab Med</i>. 2017;141:215-219. https://doi.org/10.5858/arpa.2016-2017-CP. Accessed 2019. Bynny RL, Paauw DS, Papadakis MA, Pfeil S. <i>Medical Professionalism Best Practices: Professionalism in the Modern Era</i>. Menlo Park, CA: Alpha Omega Alpha Honor Medical Society; 2017. https://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, • Has timely attendance at laboratory meetings, grand rounds and clinical conferences requests, or reminders to complete tasks and • Completes administrative tasks, safety training documentation, and procedure review by responsibilities specified due date **Level 2** Takes ownership and performs tasks • Completes evaluations by specified due date and responsibilities in a timely manner • Completes assigned tasks before taking scheduled leave Level 3 Anticipates situations that may impact • Notifies director of errors, testing delays, complex results that require more time own ability to meet responsibilities and • Asks for assistance from director, lab staff members, or faculty members as needed describes the impact on team • Arranges coverage for assigned tasks and notifies appropriate individuals in preparation for scheduled leave Level 4 Shares responsibility for system • Takes responsibility for inadvertently omitting key diagnostic information from a report outcomes as a member of the team **Level 5** Designs new strategies to ensure that After soliciting input from providers, designs a testing algorithm for a specific disease the needs of patients, teams, and systems are met **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** ABMGG Professionalism Guidelines Notes or Resources 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 **Examples Milestones** • With assistance, acknowledges own response to patient's fatal genetic diagnosis **Level 1** Describes common indicators of • Recognizes the need for time away from work for personal care personal or professional well-being Level 2 Independently recognizes status of • Independently identifies and communicates impact of a personal family tragedy on one's personal and professional well-being and seeks ability to be productive help when needed • Recognizes a pattern of agitation and works to handle appropriately Level 3 Proposes a plan to optimize personal • With a mentor, develops a reflective response to deal with personal impact of difficult and professional well-being 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 • Independently identifies and implements ways to manage personal stress and professional well-being • Starts a book club for members of the laboratory team • Assists in organizational efforts to address laboratorian well-being after an adverse **Level 5** Coaches others to develop and implement plans to optimize personal and patient outcome resulting from a laboratory error professional well-being • 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** • Local resources, including Employee Assistance, HR Notes or Resources • 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.

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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		
Examples		
Identifies that language (verbal and also 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		
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		
Tarrives on time and remains present for the entire patient encounter		
• 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		
Wallitaine by a bornable and opposite and buy to patiente with intellection alloadimited		
• Creates an educational brochure to explain the different types of DNA variants found by		
molecular testing		
Explains an abnormal karyotype result for a patient with newly diagnosed Trisomy 21		
• Explains an abhornal karyotype result for a patient with newly diagnosed misorny 21		

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) Objective structured clinical examination (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. <i>Med Teach</i>. 2011;33(1):6-8. Makoul G. Essential elements of communication in medical encounters: The Kalamazoo consensus statement. <i>Acad Med</i>. 2001;76:390-393. Makoul G. The SEGUE Framework for teaching and assessing communication skills. <i>Patient Educ Couns</i>. 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. <i>BMC Med Educ</i>. 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. <i>Pediatrics</i>. 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. <i>Am J Med Genet A</i>. 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** • Acknowledges the contribution of each member of the laboratory team Level 1 Uses language that values all members of the health care team, including clinical and laboratory professionals Describes the utility of constructive feedback • Identifies that constructive feedback benefits the entire team Level 2 Adapts communication style to fit team • Communicates diagnostic evaluation results clearly and concisely in an organized and needs timely manner Solicits feedback on personal performance as a • Asks the laboratory team how they can improve explanation of test results member of the laboratory team • Asks senior fellow how to improve timeliness for completion of assay Level 3 Communicates information effectively • After a test has been interpreted, communicates with the primary care team to verify they with all health care team members, including have received and understand the results clinical and laboratory professionals, with assistance Integrates feedback from team members to • Shares information gained regarding the request for a STAT result with the lab team and improve own communication and other skills 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 • Explains rationale for chromosome analysis instead of chromosome microarray analysis information effectively with all health care team as an initial diagnostic test for suspected Down syndrome members, including clinical and laboratory • Asks other members of the health care team to repeat back recommendations to ensure professionals understanding Facilitates regular health care team-based • Participates in multidisciplinary meetings to discuss triage strategy for bone marrow communications and feedback in complex specimens having multiple genetic tests situations Level 5 Models flexible communication • Mediates a conflict resolution between different members of the laboratory team strategies that value input from all health care team members, resolving conflict when needed 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. <i>Med Teach</i>. 2018;21:1-4. https://doi.org/10.1080/0142159X.2018.1481499. Accessed 2019. Green M, Parrott T, Cook G. Improving your communication skills. <i>BMJ</i>. 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. <i>Med Teach</i>. 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. <i>MedEdPORTAL</i>. 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. <i>Pediatrics</i>. 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. <i>JAMA</i>. 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	Shreds patient list after case conference Actively logs off the computer and keeps protected health information (written or electronic) locked at all times	
Identifies institutional and departmental procedures for communication of issues	 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	• Identifies that provider should be called or paged immediately when a critical result is identified	
Uses institutional structure to effectively communicate clear and constructive suggestions, with assistance	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	Contacts provider when a critical result is identified	
Uses institutional structure to effectively communicate clear and constructive suggestions	Knows when to direct concerns locally, departmentally, or institutionally via appropriate escalation	
Level 4 Independently communicates clinical information	Immediately contacts provider when a critical results is identified and appropriately documents the communication	
Initiates conversations on difficult subjects with appropriate stakeholders to improve the system	Organizes a discussion about the inappropriate release of pharmacogenetics testing results in the EHR	
Level 5 Models effective communication of clinical information	Develops a simulation project to improve communication skills between junior fellow and ordering providers	
Facilitates dialogue regarding systems issues among larger community stakeholders	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.