Supplemental Guide:
Medical Biochemical Genetics

ACGME

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Milestones Supplemental Guide

This document provides additional guidance and examples for the Medical 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.

Some milestone descriptions include statements about performing independently. It is important to use this guide in conjunction with the ACGME specialty-specific Program Requirements. Specific language has been included that is best defined through the Program Requirements. One notable area within the requirements is VI.A.2.c) which includes the definitions for levels of supervision:

> Levels of Supervision
>
> To promote oversight of resident supervision while providing for graded authority and responsibility, the program must use the following classification of supervision:
>
> Direct Supervision – the supervising physician is physically present with the resident and patient.
>
> Indirect Supervision:
>
> with Direct Supervision immediately available – the supervising physician is physically within the hospital or other site of patient care, and is immediately available to provide Direct Supervision.
>
> with Direct Supervision available – the supervising physician is not physically present within the hospital or other site of patient care, but is immediately available by means of telephonic and/or electronic modalities, and is available to provide Direct Supervision.
>
> Oversight – the supervising physician is available to provide review of procedures/encounters with feedback provided after care is delivered

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: Initial or Acute Management **Overall Intent:** To diagnose and manage acutely presenting inborn errors of metabolism	
Milestones	**Examples**
Level 1 *With guidance, recognizes signs and symptoms of inborn errors of metabolism and identifies when further testing is needed*	● Recognizes initial presenting signs of metabolic emergencies like hyperammonemia, metabolic acidosis, and hypoglycemia
Selects and orders diagnostic tests and develops a rudimentary management plan for initial evaluation or implements the existing management plan	● Orders immediate (i.e., STAT) ammonia levels in appropriate acute clinical metabolic presentations ● Implements a previously established acute management plan for a patient with known metabolic disease
Level 2 *Independently recognizes signs and symptoms of inborn errors of metabolism and identifies when further testing is needed*	● Responds to consultation for hyperammonemia and orders plasma amino acids and urine organic acids
Develops and implements an initial management plan for common patient presentations of inborn errors of metabolism (e.g., organic aciduria, urea cycle)	● Responds to consultation for hyperammonemia and recommends starting intravenous (IV) dextrose fluids; considers use of scavenger therapy for management of hyperammonemia
Level 3 *Synthesizes the clinical context and pattern of laboratory results to identify the likelihood of a common inborn error of metabolism*	● Evaluates laboratory results like ammonia level and plasma amino acids to reach a specific urea cycle diagnosis and implement appropriate scavenger therapy ● In a patient with urea cycle disorder, establishes criteria for when to start dialysis when unresponsive to other therapies
Develops and implements an initial management plan for complex or rare patient presentations of inborn errors of metabolism (e.g., pyruvate dehydrogenase complex deficiency, carnitine-acylcarnitine translocase deficiency)	● Identifies that a patient with coarse facial features, joint stiffness, and increased glycosoaminoglycans has a lysosomal storage disease ● Develops a management plan for a patient presenting with pyruvate dehydrogenase deficiency that includes high-dose thiamine, decreasing IV dextrose administration, and increasing lipids
Level 4 *Synthesizes the clinical context and pattern of laboratory results to identify the likelihood of a complex or rare inborn error of metabolism*	● In a patient with hyperammonemia with normal metabolic labs, orders abdominal ultrasound to look for vascular shunts

Anticipates and creates contingency plan for an acute response and develops an acute secondary plan	● In a patient with urea cycle disorder, establishes criteria for when to refer for liver transplant evaluation
Level 5 *Leads development of institutional protocols for diagnosis and management of inborn errors of metabolism*	● Develops an institutional clinical practice guideline for acute evaluation of patients with metabolic emergencies
Assessment Models or Tools	● Case-based presentation ● Direct observation ● Faculty evaluations ● Medical record (chart) review
Curriculum Mapping	●
Notes or Resources	● American College of Medical Genetics and Genomics. Practice Resources. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Practice_Resources/ACMG/. 2021. ● Gene reviews ● Saudubray JM, van de Berghe G, Walter JH. *Inborn Metabolic Diseases: Diagnosis and Treatment*. 5th ed. Germany: Springer; 2011. ISBN:978-3642157196.

Patient Care 2: Chronic Management **Overall Intent:** To diagnose and longitudinally manage patients with inborn errors of metabolism	
Milestones	**Examples**
Level 1 *Takes a comprehensive genetic history for a patient with pertinent positive and negative findings; integrates the history with other data to develop a differential diagnosis*	● Takes comprehensive but inefficient patient histories, and includes elements that are unlikely to be relevant for biochemical genetics; makes broad differential diagnosis rather than focused and may only take into account one aspect of the presentation
Implements dietary and medical therapy for patients with a common inborn error of metabolism (e.g., carnitine uptake defect, medium-chain acyl-CoA dehydrogenase deficiency, dietary therapy for phenylketonuria (PKU))	● Accesses care guidelines for common conditions and implements care in accordance with them; develops straightforward care plan and does not entail significant risk
Provides symptomatic and supportive care to patients	● Provides general supportive care such as referring a child with developmental delay for developmental therapies and assessments
Level 2 *Takes an inborn error of metabolism focused history for a patient with an established common inborn error of metabolism (e.g., PKU)*	● For a patient with phenylketonuria, focuses the history on the details of development, behavior, diet and medication adherence, and barriers to care as opposed to a more general and superficial history
Synthesizes clinical and laboratory data to optimize complex dietary and medical therapy (e.g., urea cycle disorders)	● For a patient with a urea cycle disorder, assesses amino acids to consider both overtreatment and undertreatment
Provides routine disease-specific surveillance	● For a patient with Pompe disease, accesses professional practice guidelines and plans specific surveillance based on the recommendations
Level 3 *Takes a disease-specific history for a patient with a complex inborn error of metabolism with multisystemic manifestations (e.g., Hunter syndrome)*	● For a patient with Hunter Syndrome, takes a history focused on growth, development, behavior, recurrent otitis media, joint contractures, and cardiac valvular disease
Identifies a patient who, based on natural history, is a candidate for disease modifying therapy (e.g., solid organ transplant, enzyme replacement therapy, cell-based therapy)	● For a patient with methylmalonic acidemia, makes a referral for liver transplantation, and presents to the interdisciplinary team care conference to discuss the risks and benefits of that procedure for the patient

Provides ongoing comprehensive surveillance of disease progression and care coordination	● For a patient with methylmalonic acidemia, ensures appointments with cardiology, nephrology, and ophthalmology
Level 4 *Efficiently takes a nuanced disease-specific history for a patient with an inborn error of metabolism*	● On a busy clinic day, successfully obtains history of present illness for several different metabolic diseases, in each case getting the relevant information in a relatively short amount of time
Guides a patient through shared decision making on novel or high-risk therapies; coordinates implementation of these therapies	● Guides a family of a patient with Niemann Pick Type B through considering whether to participate in a clinical trial of acid sphingomyelinase enzyme replacement therapy
Anticipates and creates contingency plans for highly pleotropic inborn errors of metabolism (e.g., mucopolysaccharidosis I)	● Takes a call from a family of a patient with Hurler syndrome and identifies that the patient is in respiratory distress due to a viral illness; proactively tells the emergency department physicians they will need to use fiber optic intubation techniques for the patient if intubation is required
Level 5 *Implements a clinical trial to improve management of patients with an inborn error of metabolism*	● Implements a trial with compassionate use of a drug for a patient
Implements a multi-disciplinary clinic for care of patients with an inborn error of metabolism	● Implements a clinic for patients transitioning from pediatric to adult care with dietician and social work support
Assessment Models or Tools	● Case-based presentation ● Direct observation ● Faculty evaluations ● Medical record (chart) review
Curriculum Mapping	●
Notes or Resources	● American College of Medical Genetics and Genomics. Practice Resources. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Practice_Resources/ACMG/. 2021. ● Gene reviews ● Saudubray JM, van de Berghe G, Walter JH. *Inborn Metabolic Diseases: Diagnosis and Treatment*. 5th ed. Germany: Springer; 2011. ISBN:978-3642157196.

Patient Care 3: Newborn Screening **Overall Intent:** To diagnose and manage patients with abnormal newborn screen results	
Milestones	**Examples**
Level 1 *Applies published (state or national) protocols to obtain follow-up testing based on initial newborn screen results* *Applies published (state or national) protocols for initial management based on the initial newborn screen result*	● Identifies the correct ACT algorithm and implements initial follow-up as recommended
Level 2 *Generates a differential diagnosis based on newborn screen results that includes diagnoses other than inherited metabolic diseases* *Develops a continuing management plan when initial testing is inconclusive*	● In the setting of a positive newborn screen (NBS) result for elevated complement 3, lists non-inborn errors that can lead to this result such as hyperbilirubinemia, maternal B-12 deficiency, etc. ● For a patient has an intermediate NBS result for very long-chain acyl-CoA dehydrogenase deficiency (VLCADD), instructs the family in appropriate precautions around fasting while the ongoing diagnostic evaluation is completed and provides appropriate written documentation for the family (e.g., an emergency letter)
Level 3 *Applies published protocols for diagnosis with modifications to account for patient specific factors* *Makes appropriate decisions about urgency of implementation of management based on initial clinical history*	● When a patient flags on NBS for elevated leucine, identifies that the child is premature, in the neonatal intensive care unit (NICU) and has multiple amino acid elevations; recommends follow-up testing is based on the low likelihood that this is a true positive result ● Refers the child of a vegan mother with elevated C3 to the nutrition clinic for follow-up of potential vitamin B-12 deficiency
Level 4 *Integrates data from multiple sources to arrive at an accurate diagnosis for the patient* *Develops a plan for management of a patient with incidental findings or results that indicate*	● Integrates deoxyribonucleic acid (DNA) and biochemical test results to confirm or rule out inborn error of metabolism ● When urine organic acid testing obtained per protocol shows elevated fumarate, correctly diagnoses the condition and implements care for both the patient as well as carriers for the condition ● Makes a diagnosis of succinate-CoA ligase ADP-forming subunit beta (SUCLA2) or formiminoglutamic acidemia (FIGLU) deficiency from elevated newborn screening results (complement 3 and complement 4 elevations, respectively)

concern for a disease that is not intended to be identified with newborn screening	
Level 5 *Participates in state, regional, or national newborn screening program policy development or evaluation projects*	● Works with the state laboratory to develop new screening protocols or modify existing protocols
Assessment Models or Tools	● Case-based presentation ● Direct observation ● Faculty evaluations ● Medical record (chart) review
Curriculum Mapping	●
Notes or Resources	● ACMG. ACT Sheets and Algorithms. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets_and_Algorithms.aspx. 2021. ● Health Resources and Services Administration (HRSA). Newborn Screening Information Center. https://newbornscreening.hrsa.gov/. 2021. ● New Steps. https://www.newsteps.org. 2021. ● Saudubray JM, van de Berghe G, Walter JH. *Inborn Metabolic Diseases: Diagnosis and Treatment*. 5th ed. Germany: Springer; 2011. ISBN:978-3642157196.

Medical Knowledge 1: Molecular and Metabolic Mechanisms **Overall Intent:** To demonstrate knowledge of the molecular and metabolic mechanisms underlying inborn errors of metabolism	
Milestones	**Examples**
Level 1 *Describes biochemical pathways and principles underpinning diagnosis and therapy for categories of inborn errors of metabolism (e.g., protein restriction, cofactor therapy, pathway modifying medications)*	● Describes why tyrosine supplementation is needed for phenylketonuria
Level 2 *Describes how manipulation of biochemical pathways through various therapeutic modalities (diet, medications, etc.) can be employed for management of specific inborn errors of metabolism (e.g., ornithine transcarbamylase deficiency versus argininosuccinate lyase deficiency; cystathionine beta-synthase deficiency versus cobalamin C disease)*	● Describes underlying biochemical mechanisms of urea cycle disorders and how arginine and citrulline can be used for specific urea cycle disorders for the therapeutic options
Level 3 *Demonstrates knowledge of the interconnectedness of biochemical pathways and employs these concepts in diagnosis and management*	● Describes why use of N-acetylglutamate may be beneficial in management of hyperammonemia in patients with propionic acidemia
Level 4 *Integrates knowledge of the complex nature of biochemical pathways to prioritize and employ the range of therapeutic options for a patient, based upon an individual's clinical and biochemical response to therapy*	● Describes how cytochrome P450 enzymes (CYP) metabolizer status impacts a patient's eligibility for Gaucher disease substrate reduction therapy
Level 5 *Expands understanding of underlying biochemical basis of inborn errors of metabolism through publication of new knowledge that impacts the diagnosis or treatment of a disease or category of inborn errors of metabolism*	● Is involved in a clinical research project on genomics of metabolic pathways, and drafts and publishes the result
Assessment Models or Tools	● Case-based presentation ● Direct observation ● Faculty evaluations ● Medical record (chart) review
Curriculum Mapping	●

Notes or Resources	● Kyoto Encyclopedia of Genes and Genomes (KEGG). KEGG PATHWAY Database. https://www.genome.jp/kegg/pathway.html. 2021. ● McGraw-Hill Medical. OMMBID Table of Contents. https://ommbid.mhmedical.com/. 2021.

Medical Knowledge 2: Diagnostic Testing **Overall Intent:** To demonstrate knowledge of diagnostic testing for inborn errors of metabolism	
Milestones	**Examples**
Level 1 *Describes the technology and use of diagnostic testing for inborn errors of metabolism*	● Identifies and interprets the methods for initial diagnostic testing for acid-base disorder, hyperammonemia and hypoglycemia
Demonstrates knowledge of the differences between newborn screening and diagnostic testing	● Knows the state specific NBS program, and compares and contrasts NBS results from confirmatory diagnostic test results
Level 2 *Identifies possible methods for diagnosis and subsequent laboratory monitoring for inborn errors of metabolism*	● Identifies plasma amino acids, urine organic acids, lactate-pyruvate as tests for IEM ● Explains how Tandem-MS is involved and how it works in diagnostic evaluations
Interprets common metabolic testing (e.g., plasma amino acids) for inborn errors of metabolism	● Describes the pattern of amino acid elevations in maple syrup urine disease
Describes causes of false positive/negative rates and how these factors can impact newborn screening test interpretation	● Describes why there is an increased risk of false-positives observed in premature infants and infants with sickness
Level 3 *Identifies best methods for diagnosis and subsequent laboratory monitoring for common inborn errors of metabolism*	● Describes strengths and limitations of enzyme and urine glycosaminoglycan testing in the diagnosis of mucopolysaccharidoses
Interprets complex metabolic testing (e.g., very long chain fatty acids) for inborn errors of metabolism	● Interprets the results of very long chain fatty acids in peroxisomal disorders
Demonstrates knowledge of positive predictive values of newborn screening tests and tools for improved discrimination of positive and negative results	● Explains how two-tier testing reduces rate of false positives based on higher pre-test probability for the follow up test

Level 4 *Identifies best methods for diagnosis and subsequent laboratory monitoring for complex inborn errors of metabolism*	● Identifies categories of congenital disorders of glycosylation and which disorders have abnormal N- or O-linked glycosylation patterns
Integrates results of metabolic and genomic testing to arrive at a diagnosis	● Uses enzyme testing, urine hex4 and α-glucosidase (GAA) variant to diagnose late-onset Pompe disease
Integrates data from multiple sources to identify how likely an individual newborn screening test is a true positive	● Incorporates data from biochemical testing and DNA testing, including population databases, to identify likelihood of pseudo-deficiency of MPS1 (Mucopolysaccharidosis Type I)
Level 5 *Develops polices or practice guidelines for diagnostic testing of inborn errors of metabolism*	● Is involved in projects that develop state- and institution-specific guidelines
Participates in new assay development or gene discovery	● Describes a new biomarker for follow-up evaluation of a metabolic disorder
Contributes to generalizable medical knowledge of newborn screening tests	● Participates in the development of practice guidelines for newborn screening
Assessment Models or Tools	● Case-based presentation ● Direct observation ● Faculty evaluations ● Medical record (chart) review
Curriculum Mapping	●
Notes or Resources	● Garg U, Smith LD, Heese BA. *Laboratory Diagnosis of Inherited Metabolic Diseases*. Washington: AACC Press; 2012. ISBN:978-1-594-25140-5. ● Garg U, Smith LD. *Biomarkers in Inborn Errors of Metabolism: Clinical Aspects and Laboratory Determination.* 1st ed. Cambridge, MA: Elsevier; 2017. ISBN:978-0128028964.

Systems-Based Practice 1: Patient Safety and Quality Improvement (QI) **Overall Intent:** To identify, report, analyze, and disclose patient safety events and participate in a QI project	
Milestones	**Examples**
Level 1 *Demonstrates knowledge of common patient safety events*	● Acknowledges risks associated with prescribing the incorrect diet for patients with metabolic conditions
Demonstrates knowledge of how to report patient safety events	● Identifies the safety event reporting mechanism for their institution
Demonstrates knowledge of basic quality improvement methodologies and metrics	● Describes the components of a Plan, Do, Study, Act (PDSA) cycle
Level 2 *Identifies system factors that lead to patient safety events*	● Identifies transitions of care as a system risk factor contributing to metabolic decompensation
Reports patient safety events through institutional reporting systems (simulated or actual)	● Enters a safety event report after discovering the inadvertent administration of the wrong medication or IV fluid
Describes local (institutional) quality improvement initiatives	● Describes a current QI project to improve timely access to clinic appointments
Level 3 *Participates in analysis of patient safety events (simulated or actual)*	● Participates in a simulated root cause analysis related to a sodium benzoate/sodium phenylacetate overdose in the hospital
Participates in disclosure of patient safety events to patients and families (simulated or actual)	● In collaboration with the attending, discloses the erroneous administration of IV fluid to a patient/caregiver
Participates in local (institutional) quality improvement initiatives	● Participates in a QI project with ancillary staff members to reduce false positive ammonia results from improper blood collection
Level 4 *Conducts analysis of patient safety events and offers error prevention strategies (simulated or actual)*	● Collaborates with patient safety committee to analyze a medication error
Independently discloses patient safety events to patients and families (simulated or actual)	● Independently discloses the erroneous administration of IV fluid to a patient/caregiver

Demonstrates the skills required to identify, develop, implement, and analyze a quality improvement project	● Plans and starts a PDSA cycle related to improved timely access to clinic appointments
Level 5 *Actively engages teams and processes to modify systems to prevent patient safety events* *Mentors others in the disclosure of patient safety events* *Creates, implements, and assesses quality improvement initiatives at the institutional or community (state/federal) level*	● Leads an initiative to reduce risk of medication errors during transitions of care ● Coaches a resident on disclosure of a safety event related to a medication error ● Completes and shares outcomes of a full PDSA cycle related to improved access to clinic appointments
Assessment Models or Tools	● Direct observation ● Institutional patient safety e-module multiple choice tests ● Medical record (chart) audit ● Portfolio
Curriculum Mapping	●
Notes or Resources	● American Academy of Family Physicians. Basic of Quality Improvement. https://www.aafp.org/practice-management/improvement/basics.html. 2021. ● Institute of Healthcare Improvement. http://www.ihi.org/Pages/default.aspx. 2021.

Systems-Based Practice 2: System Navigation for Patient-Centered Care **Overall Intent:** To navigate the health care system to adapt care to a specific patient population to ensure high-quality patient outcomes	
Milestones	**Examples**
Level 1 *Demonstrates knowledge of care coordination* *Identifies key elements for safe and effective transitions of care and hand-offs* *Demonstrates knowledge of population and community health needs*	● Identifies the members of the interprofessional team and describes their roles, but is not yet routinely using team members or accessing resources ● Recognizes the essential components of an effective sign-out ● Identifies components of social determinants of health and their impact on the delivery of patient care
Level 2 *Coordinates care of patients in routine clinical situations effectively using the roles of the interprofessional teams, including non-physician patient caregivers* *Performs safe and effective transitions of care/hand-offs in routine clinical situations* *Identifies specific population and community health needs for their local population*	● Contacts interprofessional team members and consultants for necessary referrals for a patient with lysosomal storage disorders ● Performs a basic sign-out, but still needs guidance for anticipated events ● Knows which patients are at high risk for metabolic decompensation related to health literacy concerns and insurance status
Level 3 *Coordinates care of patients in complex clinical situations effectively using the roles of the interprofessional teams* *Performs safe and effective transitions of care/hand-offs in complex clinical situations* *Uses local resources effectively to meet the needs of a patient population and community*	● Coordinates with primary care provider, dietician, and social worker for the care of a newly diagnosed metabolic patient ● Provides anticipatory guidance for unstable patients including recommendations for how to escalate treatments for patients with uncontrolled ammonia levels ● Works with the social worker/health navigator to ensure patients with low literacy understand how to access resources over time
Level 4 *Models effective coordination of patient-centered care among different disciplines and specialties including referrals and testing*	● Educates other learners on engagement of appropriate interprofessional team members to ensure the necessary resources have been arranged

Models and advocates for safe and effective transitions of care/hand-offs within and across health care delivery systems including outpatient settings, referrals, and testing *Participates in changing and adapting practice to provide for the needs of specific populations including advocating for a patient's genetic testing coverage*	● Proactively calls the outpatient clinicians to communicate status updates and goals of care ● Independently drafts letters of medical necessity for genetic testing or metabolic formulas to advocate for their patients
Level 5 *Analyzes the process of care coordination and leads in the design and implementation of quality improvements* *Improves quality of transitions of care within and across health care delivery systems to optimize patient outcomes* *Leads innovations and advocates for populations and communities with specific health care needs at the state or federal level*	● Creates order set for patients with metabolic disorders presenting to the emergency department ● Develops protocols for pre- and intra-transplant patients with urea cycle disorder and measures patient outcomes ● Collaborates with key stakeholders at the state level to ensure patients with phenylketonuria receive access to metabolic formula throughout the life span
Assessment Models or Tools	● Direct observation ● Medical record (chart) audit ● Multisource feedback ● Review of written sign-out/hand-off tools
Curriculum Mapping	●
Notes or Resources	● Agency for Healthcare Research and Quality (AHRQ). Handoffs and Signouts. https://psnet.ahrq.gov/primer/handoffs-and-signouts. 2021. ● IPASS. Patient Safety Institute. https://ipassinstitute.com. 2021. ● Wohlauer MV, Arora VM, Bass EJ, et al. The patient handoff: a comprehensive curricular blueprint for resident education to improve continuity of care. *Acad Med.* 2012 Apr; 87(4): 411-418. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3409830/pdf/nihms-395982.pdf. 2021.

Systems-Based Practice 3: Physician Role in Health Care Systems **Overall Intent:** To navigate the health care system to improve patient care and the health system's performance	
Milestones	**Examples**
Level 1 *Identifies key components of the complex health care system (e.g., hospital, skilled nursing facility, finance, personnel, technology)*	● Recognizes the many factors that may impact a patient's hospital length of stay
Describes basic health payment systems (e.g., government, private, public, uninsured care) and practice models	● Demonstrates knowledge of payment systems, such as Medicare, Medicaid, the VA, and commercial third-party payers for authorization of genetic testing
Identifies basic knowledge for effective transition to practice (e.g., information technology, legal, billing and coding, financial, personnel)	● Recognizes the use of ICD10 and CPT codes in billing and ordering genetic testing
Level 2 *Describes how components of a complex health care system are interrelated, and how this impacts patient care*	● Recognizes how early genetic/metabolic consultation can impact hospital length of stay
Delivers care with consideration of each patient's payment model (e.g., insurance type) and access to genetic testing or formula	● Describes how genetic services are covered by different payment systems
Demonstrates use of information technology required for medical practice (e.g., electronic health record, documentation required for billing and coding)	● Produces documentation necessary for billing and reimbursement
Level 3 *Discusses how individual practice affects the broader system (e.g., access to genetic testing and treatments, testing advocacy)*	● Discusses how a diagnostic genetic and biochemical test results may lead to additional subspecialty consultations and further testing or screening
Engages with patients in shared decision making, often informed by each patient's payment models	● Counsels patients on genetic testing options and coverage for metabolic medications, formulas and medical food depending upon insurance coverage, co-payments, and deductibles

Describes core administrative knowledge needed for transition to practice (e.g., contract negotiations, malpractice insurance, government regulation, compliance)	● Is familiar with resources available for contract negotiations when evaluating future job opportunities
Level 4 *Manages various components of the complex health care system to provide efficient and effective patient care and transition of care* *Advocates for patient care needs (e.g., community resources, patient assistance resources) with consideration of the limitations of each patient's payment model, including genetic testing through research* *Analyzes individual practice patterns and professional requirements in preparation for practice*	● Manages transition from hospital to outpatient treatment for a patient with metabolic disorder ● If insurance denies genetic testing and coverage for metabolic medications, formulas and medical food for a patient, discuss alternatives such as research protocols, clinical trials, charity funding, and self-payment ● Develops a professional development plan for the first year after training
Level 5 *Advocates for or leads systems change that enhances high-value, efficient, and effective patient care and transition of care* *Participates in health policy advocacy activities* *Educates others to prepare them for transition to practice*	● Presents institution-specific data and incorporates rapid evaluation for hyperammonemia in neonates undergoing sepsis evaluation with metabolic alkalosis to allow early diagnosis of urea cycle disorders ● Develops e-consults or telehealth services to increase access to genetic/metabolic services for rural and underserved patient populations ● Counsels residents on transition to practice
Assessment Models or Tools	● Direct observation ● Medical record (chart) audit ● Multisource feedback ● Resident self-reflection
Curriculum Mapping	●
Notes or Resources	● ACMG. Policy Statements. https://www.acmg.net/ACMG/Advocacy/Policy-Statements/ACMG/Advocacy/Policy-Statements.aspx. 2021. ● ACMG. Salary Survey. https://www.acmg.net/PDFLibrary/2019%20ACMG%20Salary%20Survey%20Report%20Now%20Available.final.pdf. 2021.

	● AHRQ. Measuring the Quality of Physician Care. https://www.ahrq.gov/professionals/quality-patient-safety/talkingquality/create/physician/challenges.html. 2021. ● AHRQ. Major Physician Measurement Sets. https://www.ahrq.gov/talkingquality/measures/setting/physician/measurement-sets.html. 2021. ● American Medical Association. Tips for negotiating employee contracts. https://www.ama-assn.org/residents-students/career-planning-resource/tips-negotiating-employment-contracts. 2021. ● Institutional templates for letters of medical necessity ● NEJM. Navigating the Transition from Residency to Physician Practice. https://www.nejmcareercenter.org/article/navigating-the-transition-from-residency-to-physician-practice/. 2021.

Practice-Based Learning and Improvement 1: Evidence-Based and Informed Practice **Overall Intent:** To evaluate and incorporate evidence and patient values into clinical practice	
Milestones	**Examples**
Level 1 *Demonstrates how to access and use available evidence, and incorporate patient preferences and values to take care of a routine patient*	● Identifies clinical practice guideline for evaluation of a patient with urea cycle disorder ● Understands that patient values affect care
Level 2 *Articulates clinical questions and elicits patient preferences and values to guide evidence-based care*	● Asks questions to determine patient and family preferences regarding evaluation, testing, and treatment
Level 3 *Locates and applies the best available evidence, integrated with patient preference, to the care of complex patients*	● Synthesizes available evidence to make a recommendation for treatment of newborn with severe hyperammonemia considering patient and family preferences
Level 4 *Critically appraises and applies evidence even in the face of uncertainty and conflicting evidence to guide care, tailored to the individual patient*	● Recognizes gaps in high-level evidence and incorporates other case reports or non-clinical studies (animal models) to guide recommendation for treatment of rare metabolic disorders
Level 5 *Mentors others to critically appraise and apply evidence for complex patients; and/or participates in the development of guidelines*	● Develops standardized journal club format for critical appraisal of available evidence and its application to patients with metabolic disorders
Assessment Models or Tools	● Direct observation ● In-training exam ● Faculty member evaluations ● Multisource feedback ● Resident self-reflection
Curriculum Mapping	●
Notes or Resources	● American College of Medical Genetics and Genomics. Practice Resources. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Practice_Resources/ACMG/. 2021. ● Cochrane Library. Cochrane Database of Systematic Reviews. https://www.cochranelibrary.com/cdsr/about-cdsr. 2021. ● GeneReviews. www.genereviews.org. 2021. ● Online Mendelian Inheritance in Man (OMIM). An Online Catalog of Human Genes and Genetic Disorders. https://www.omim.org/. 2021. ● PubMed search

Practice-Based Learning and Improvement 2: Reflective Practice and Commitment to Personal Growth **Overall Intent:** To seek clinical performance information to improve care and develop objectives and goals for improvement	
Milestones	**Examples**
Level 1 *Demonstrates openness to performance data (feedback and other input) 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 prompting*	● Integrates feedback to adjust the documentation of diet, medical history and plan for patients with phenylketonuria ● Assesses time management skills and how it impacts timely completion of clinic notes and literature reviews ● When prompted, develops individual education plan to improve their understanding of biochemical testing
Level 2 *Seeks performance data episodically, with adaptability and humility* *Analyzes, reflects on, and institutes behavioral change(s) to narrow the gap(s) between expectations and actual performance* *Independently creates and implements a learning plan*	● Does a chart audit to determine the percent of patients with phenylketonuria with detailed dietary history and analysis of phenylalanine and tyrosine levels ● Completes a comprehensive literature review prior to patient encounters ● Using web-based resources, creates a personal curriculum to improve his/her understanding of biochemical testing
Level 3 *Seeks performance data consistently with adaptability and humility* *Challenges assumptions and considers alternatives in narrowing the gap(s) between expectations and actual performance* *Uses performance data to measure the effectiveness of the learning plan and when necessary, improves it*	● Completes a quarterly chart audit to ensure a detailed dietary history for patients with phenylketonuria ● After patient encounter, debriefs with the attending and other patient care team members to optimize future collaboration in the care of the patient and family ● Performs a chart audit on personal documentation of their understanding of biochemical testing
Level 4 *Models seeking performance data with adaptability and humility* *Mentors others on reflective practice*	● Models practice improvement and adaptability ● Develops educational module for collaboration with other patient care team members

Facilitates the design and implementing learning plans for others	● Assists first-year residents in developing individualized learning plans
Level 5 *Develops evaluations or education resources/tools for learners* *Participates in the development of courses for the education of students or other physicians*	● Authors a book chapter on metabolic disorders ● Serves on a meeting program committee ● Serves as a member of the Milestones Development Committee
Assessment Models or Tools	● Direct observation ● Medical record (chart) audit ● Mentored review of individualized learning plan ● Multisource feedback
Curriculum Mapping	●
Notes or Resources	● Burke AE, Benson B, Englander R, Carraccio C, Hicks PJ. Domain of competence: practice-based learning and improvement. *Acad Pediatr.* 2014;14: S38-S54. https://www.academicpedsjnl.net/article/S1876-2859(13)00333-1/fulltext. 2020. ● Hauer J, Quill T. Educational needs assessment, developing learning objectives, and choosing a teaching approach. *Journal of Palliative Medicine*. 2011;14(4):503-508. https://pubmed.ncbi.nlm.nih.gov/21438708/. 2021. ● Hojat M, Veloski JJ, Gonnella JS. Measurement and correlates of physicians' lifelong learning. *Academic Medicine.* 2009;84(8):1066-1074. https://journals.lww.com/academicmedicine/fulltext/2009/08000/Measurement_and_Correlates_of_Physicians__Lifelong.21.aspx. 2021. ● Lockspeiser TM, Schmitter PA, Lane JL, Hanson JL, Rosenberg AA, Park YS. Assessing residents' written learning goals and goal writing skill: validity evidence for the learning goal scoring rubric. *Academic Medicine*. 2013;88(10):1558-1563. https://journals.lww.com/academicmedicine/fulltext/2013/10000/Assessing_Residents__Written_Learning_Goals_and.39.aspx. 2021. ● Sockalingam S, Wiljer D, Yufe S, et al. The relationship between academic motivation and lifelong learning during residency: A study of psychiatry residents. *Academic Medicine*. 2016;91(10):1423-1430. https://journals.lww.com/academicmedicine/FullText/2016/10000/The_Relationship_Between_Academic_Motivation_and.28.aspx. 2021.

Professionalism 1: Professional Behavior and Ethical Principles **Overall Intent:** To demonstrate ethical and professional behavior and identify and manage lapses in self and others	
Milestones	**Examples**
Level 1 *Demonstrates compassion, sensitivity, honesty, integrity, and identifies potential triggers for professionalism lapses*	● Recognizes that fatigue may lead to unprofessional behavior
Demonstrates knowledge of the ethical principles underlying patient care	● Describes beneficence, non-maleficence, justice, and autonomy
Demonstrates basic knowledge of conflict of interest	● Understands what a conflict of interest is
Level 2 *Demonstrates compassion, sensitivity, honesty, integrity, and takes responsibility for own professionalism lapses*	● Acknowledges when actions are inappropriate without becoming defensive, making excuses, or blaming others
Analyzes straightforward situations using ethical principles	● Supports a patient who declines prenatal testing despite partner's insistence
Identifies different types of conflicts of interest, knows guidelines for interactions with vendors	● Recognizes that holding stocks in the company conducting a clinical trial at the institution must be disclosed
Level 3 *Demonstrates compassion, sensitivity, honesty, and integrity in complex/stressful situations*	● Exhibits empathy for a patient and family making end-of-life care decisions
Recognizes need to seek help in managing and resolving complex ethical situations	● Seeks further guidance when a patient with a *BRCA* pathogenic variant refuses to inform at-risk family members
Identifies resources for managing and resolving conflicts of interest	● Consults institutional legal team regarding a potential conflict of interest
Level 4 *Demonstrates compassion, sensitivity, honesty, integrity, and serves as a role model to others*	● Models empathy for a patient and family making end-of-life care decisions

Recognizes and uses appropriate resources for managing and resolving ethical dilemmas as needed *Demonstrates consistently professional behavior regarding conflicts of interest relevant to presentations, publishing, consulting, and service*	● Collaborates with the ethics committee to address a 50-year-old woman with sickle cell disease who wants to use assisted reproductive technologies ● Respects a family's desire to not be included in a research publication
Level 5 *Coaches others when their behavior fails to meet professional expectations* *Identifies and seeks to address system-level factors that induce or exacerbate ethical problems or impede their resolution*	● Coaches colleagues to correct unprofessional behavior and appearance in a respectful manner ● Develops a patient-centered guideline for addressing non-beneficial treatments
Assessment Models or Tools	● Direct observation ● Institutional ethics and conflict of interest modules ● Institutional reporting of conflict of interest ● Multisource feedback ● Resident self-reflection ● Simulation
Curriculum Mapping	●
Notes or Resources	● American Academy of Pediatrics. Case Based Teaching Guides for Resident Training. https://www.aap.org/en-us/continuing-medical-education/Bioethics-Cased-Based-Teaching-Guides/Pages/Bioethics-Case-Based-Teaching-Guides.aspx. 2021. ● ABIM Foundation; American Board of Internal Medicine, ACP-ASIM Foundation, American College of Physicians-American Society of Internal Medicine, European Federation of Internal Medicine. Medical professionalism in the new millennium: a physician charter. *Ann Intern Med*. 2002;136:243-246. http://abimfoundation.org/wp-content/uploads/2015/12/Medical-Professionalism-in-the-New-Millenium-A-Physician-Charter.pdf. 2021. ● American Medical Association. Ethics. https://www.ama-assn.org/delivering-care/ethics. 2021. ● American Society of Human Genetics. Code of Ethics. https://www.ashg.org/about/ethics.shtml. 2021. ● Byyny RL, Papadakis MA, Paauw DS. Medical professionalism best practices. Menlo Park, CA: Alpha Omega Alpha Medical Society; 2015. https://alphaomegaalpha.org/pdfs/2015MedicalProfessionalism.pdf. 2021.

	● CITI Program. Training Courses. https://about.citiprogram.org/en/homepage/. 2021. ● Doka KJ, Jennings B, Corr CA. *Living with Grief: Ethical Dilemmas at the End of Life*. Hospice Foundation of America; 2015. ISBN:978-1893349063. ● Jonsen AR, Siegler M, Winslade W. *Clinical Ethics: A Practical Approach to Ethical Decisions in Clinical Medicine*. 8th ed. McGraw-Hill; 2015. ISBN:978-0071845069. ● Levinson W, Ginsburg S, Hafferty FW, Lucey CR. *Understanding Medical Professionalism*. 1st ed. New York, NY: McGraw-Hill Education; 2014.

Professionalism 2: Accountability/Conscientiousness **Overall Intent:** To take responsibility for one's actions and the impact on patients and other members of the health care team	
Milestones	**Examples**
Level 1 *Takes responsibility for failure to complete tasks and responsibilities, identifies potential contributing factors, and describes strategies for ensuring timely task completion in the future*	● Recognizes that patient load may delay timely completion of documentation
Responds promptly to requests or reminders to complete tasks and responsibilities	● Completes Case Logs after a reminder from the coordinator
Recognizes the role of appearance, daily demeanor and conduct in the role of a professional	● Recognizes appropriate behavior and dress code
Level 2 *Performs tasks and responsibilities in a timely manner with appropriate attention to detail in routine situations*	● Accurately documents patient encounters in a timely manner ● Completes case logs and clinical work hour logs without being reminded
Recognizes situations that may impact his/her own ability to complete tasks and responsibilities in a timely manner	● Recognizes that sudden personal illness may interfere with assigned tasks
Demonstrates a professional appearance, daily demeanor, and conduct	● Dresses professionally ● Treat co-workers with respect
Level 3 *Performs tasks and responsibilities in a timely manner with appropriate attention to detail in complex or stressful situations*	● Plans ahead and completes notes prior to leaving for vacation
Proactively implements strategies to ensure that the needs of patients, teams, and systems are met	● Informs schedulers of pending absence from clinic and reassignment of patients in a timely manner ● Informs supervisors, program director, program administrator, and fellow learners of absence in a timely manner
Sets a standard for appearance, daily demeanor, and conduct as a professional	● Meets institutional requirements and expectations for conduct and dress

Level 4 *Recognizes and addresses situations that may impact others' ability to complete tasks and responsibilities in a timely manner* *Promotes professional appearance, demeanor, and conduct in their peers and associates*	● Collaborates with the team and recognizes overburdened associates and assists with patient care ● Helps others recognize departure from expected behavior and dress
Level 5 *Volunteers to improve and takes ownership of system outcomes*	● Assists outpatient clinic to develop streamlined processes for completion of prior authorizations of genetic testing
Assessment Models or Tools	● Compliance with deadlines and timelines ● Direct observation ● Multisource feedback ● Resident self-evaluation ● Rotation evaluations
Curriculum Mapping	●
Notes or Resources	● ABIM Foundation; American Board of Internal Medicine, ACP-ASIM Foundation, American College of Physicians-American Society of Internal Medicine, European Federation of Internal Medicine. Medical professionalism in the new millennium: a physician charter. *Ann Intern Med*. 2002;136:243-246. http://abimfoundation.org/wp-content/uploads/2015/12/Medical-Professionalism-in-the-New-Millenium-A-Physician-Charter.pdf. 2021. ● Institutional Code of Conduct

Professionalism 3: Self-Awareness and Help-Seeking **Overall Intent:** To identify, use, manage, improve, and seek help for personal and professional well-being for self and others	
Milestones	**Examples**
Level 1 *Recognizes status of personal and professional well-being, with assistance* *Recognizes limits in the knowledge/skills of self or team, with assistance*	● Acknowledges own response to patient's fatal genetic diagnosis ● Receives feedback on missed emotional cues after a family meeting
Level 2 *Independently recognizes status of personal and professional well-being* *Independently recognizes limits in the knowledge/ skills of self or team and demonstrates appropriate help-seeking behaviors*	● Independently identifies and communicates impact of a personal family tragedy ● Recognizes a pattern of missing emotional cues during family meetings and asks for feedback
Level 3 *With assistance, proposes a plan to optimize personal and professional well-being* *With assistance, proposes a plan to remediate or improve limits in the knowledge/ skills of self or team*	● With the multidisciplinary team, develops a reflective response to deal with personal impact of difficult patient encounters and disclosures ● Integrates feedback from the multidisciplinary team to develop a plan for identifying and responding to emotional cues during the next family meeting
Level 4 *Independently develops a plan to optimize personal and professional well-being* *Independently develops a plan to remediate or improve limits in the knowledge/skills of self or team*	● Independently identifies ways to manage personal stress ● Self-assesses and seeks additional feedback on skills responding to emotional cues during a family meeting
Level 5 *Coaches others when emotional responses or limitations in knowledge/skills do not meet professional expectations*	● Assists in organizational efforts to address clinician well-being after patient diagnosis/prognosis/death ● Works with multidisciplinary team to develop a feedback framework for learners around family meetings
Assessment Models or Tools	● Direct observation ● Group interview or discussions for team activities ● Participation in institutional well-being programs ● Resident self-reflection ● Review of learning plan

Curriculum Mapping	●
Notes or Resources	● This subcompetency is not intended to evaluate a fellow's well-being, but to ensure each fellow has the fundamental knowledge of factors that impact well-being, the mechanisms by which those factors impact well-being, and available resources and tools to improve well-being. ● American Academy of Pediatrics. Resilience Curriculum: Resilience in the face of grief and loss. Part D: Introduction to Personal Wellness. https://www.aap.org/en-us/advocacy-and-policy/aap-health-initiatives/hospice-palliative-care/Pages/Resilience-Curriculum.aspx. 2021. ● ACGME. "Well-Being Tools and Resources." https://dl.acgme.org/pages/well-being-tools-resources. 2021. ● Institutional GME guidelines regarding resident wellness ● Local resources, including Employee Assistance Program ● Stanford Medicine. WELLMD. https://wellmd.stanford.edu. 2021.

Interpersonal and Communication Skills 1: Patient- and Family-Centered Communication **Overall Intent:** To employ listening, language, behaviors, and self-awareness to form a therapeutic relationship that facilitates effective communication	
Milestones	**Examples**
Level 1 *Uses language and nonverbal behavior to demonstrate respect and establish rapport* *Identifies common barriers to effective communication while accurately communicating own role within the health care system* *Identifies the need to adjust communication strategies based on assessment of patient/family expectations and understanding of their health status and treatment options*	● Learner formally introduces self to the patient/family and states the learner's role in the care of the patient ● Reflects on how the use of silence and active listening assists in establishing patient/caregiver rapport ● Identifies the need for an interpreter for a patient/caregiver who is non-English-speaking ● Understands that different patients may have different levels of understanding and needs ● Aware of cultural and socioeconomic norms/differences in patient populations
Level 2 *Establishes a therapeutic relationship in straightforward encounters using active listening and clear language* *Identifies complex barriers to effective communication* *Organizes and initiates communication with patient/family by introducing stakeholders, setting the agenda, clarifying expectations, and verifying understanding of the clinical situation*	● In a patient referred for abnormal newborn screen results, develops shared goals (contracting) ● Refrains from the use of overly complicated terminology ● Demonstrates therapeutic relationship with appropriate use of silence and normalizing emotional responses ● Employs active listening/repeat back and written resources to ensure patient understands the medical plan
Level 3 *Establishes a therapeutic relationship in challenging patient encounters* *When prompted, reflects on biases while attempting to minimize communication barriers* *With guidance, sensitively and compassionately delivers medical information, elicits patient/family values, goals, and preferences, and acknowledges uncertainty and conflict*	● Successfully maintains therapeutic relationship in the context of patient's/caregiver's expression of anger at health care system ● Identifies and reflects on bias towards patient autonomy over cultural preferences in decision making ● Delivers sensitive medical information to patients/families privately ● With guidance, collects and incorporates patient and family values into the medical decision making process

Level 4 *Establishes therapeutic relationships, with attention to patient/family concerns and context, regardless of complexity* *Recognizes biases while attempting to proactively minimize communication barriers* *Uses shared decision making to align patient/family values, goals, and preferences with treatment options to make a personalized care plan*	● Navigates situations where parents disagree about the therapeutic management of their child ● Discusses end-of-life care that may conflict with the learner's personal values and opinions ● Independently collects and incorporates patient and family values into the medical decision-making process
Level 5 *Mentors others in situational awareness and critical self-reflection to consistently develop positive therapeutic relationships* *Models self-awareness practice while identifying teaching a contextual approach to minimize communication barriers* *Models shared decision making in patient/family communication including those with a high degree of uncertainty/conflict*	● Teaches a model for consistent family meeting debriefing ● Coaches a learner to acknowledge bias and successfully manage communication with non-English-speaking patient
Assessment Models or Tools	● Direct observation ● Faculty member evaluations ● Multisource feedback ● Resident self-reflection
Curriculum Mapping	●
Notes or Resources	● Ross LF, Saal HW, David KL, Anderson RR, American Academy of Pediatrics, American College of Medical Genetics and Genomics. Technical report: ethical and policy issues in genetic testing and screening of children. *Genetics in Medicine*. 2013;15(3):234-245. https://www.acmg.net/PDFLibrary/Ethical-Policy-Issues-Genetic-Screening-Children.pdf. 2021. ● Uhlmann WR, Schuette JL, Yashar BM. *A Guide to Genetic Counseling*. 2nd ed. Danvers, MA: John Wiley & Sons, Inc; 2009. ISBN:978-0470179659. ● Veach PC, LeRoy BS, Callanan NP. *Facilitating the Genetic Counseling Process: Practice Based Skills*. 2nd ed. Cham, Switzerland: Spring International Publishing, AG; 2018. ISBN:978-3-319-74798-9.

Interpersonal and Communication Skills 2: Interprofessional and Team Communication **Overall Intent:** To communicate with the interdisciplinary team and other health care providers	
Milestones	**Examples**
Level 1 *Clearly and concisely requests a consultation* *Clearly and concisely responds to a consultation request* *Communicates information effectively with all health care team members*	● Communicates diagnostic evaluation recommendations clearly and concisely in an organized and timely manner ● Sends a message in electronic health record to the dietician of a metabolic patient to increase the protein restriction
Level 2 *Checks own understanding of consultant recommendations* *Checks requestor's understanding of recommendations when providing consultation* *Uses active listening to adapt communication style to fit team needs*	● After a consultation has been completed, communicates with the primary care team to verify they have received and understand the recommendations ● When receiving treatment recommendations from an attending physician, repeats back the plan to ensure understanding
Level 3 *Coordinates recommendations from different members of the health care team to optimize patient care* *Provides information to the primary care team regarding rationale for recommendations* *Models active listening to other health care team members*	● Initiates a multidisciplinary meeting to developed shared care plan for a patient with a new diagnosis of an inborn error of metabolism ● Explains rationale for therapeutic recommendations in managing acute hyperammonemia ● Asks other members of the health care team to repeat back recommendations to ensure understanding
Level 4 *Leads an interprofessional metabolic team to optimize patient care* *Leads interactions between the primary care and metabolic team regarding rationale for recommendations*	● Leads decision making in a team of nurses, dieticians and other physicians and healthcare professionals in implementing a plan of management for a patient with glycogen storage disease ● Directly communicates with inpatient primary care teams in the collaborative management of a patient admitted for metabolic acidosis

Provides constructive feedback on active listening to health care team members	● Discusses with genetic residents how repeating back specific therapy plans can be done to improve patient safety
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 health care team
Assessment Models or Tools	● Direct observation ● Faculty evaluation ● Medical record (chart) audit ● Multisource feedback ● Resident self-reflection ● Peer to peer evaluation
Curriculum Mapping	●
Notes or Resources	● 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. 2021. ● François, J. Tool to assess the quality of consultation and referral request letters in family medicine. *Can Fam Physician*. 2011 May;57(5), 574-575. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3093595/pdf/0570574.pdf. 2021. ● Youngwerth J, Twaddle M. Cultures of interdisciplinary teams: how to foster good dynamics. *J Palliat Med*. 2011;14(5):650-654. https://www.liebertpub.com/doi/10.1089/jpm.2010.0395?url_ver=Z39.88-2003&rfr_id=ori%3Arid%3Acrossref.org&rfr_dat=cr_pub++0pubmed&. 2021.

Interpersonal and Communication Skills 3: Communication within Health Care Systems **Overall Intent:** To communicate through established institutional pathways using a variety of methods	
Milestones	**Examples**
Level 1 *Demonstrates organized diagnostic and therapeutic reasoning through notes in the patient record*	● Documents a differential diagnosis and justifies recommendations
Uses documentation shortcuts accurately, appropriately and in a timely manner	● Accurately edits "copy/forward" notes ● Documents phone communication encounters within 24 hours
Level 2 *Concisely reports diagnostic and therapeutic reasoning in the patient record*	● Documents rationale for progression of testing in the work-up of a patient with abnormal newborn screen results
Appropriately selects direct (e.g., telephone, in-person) and indirect (e.g., progress notes, text messages) forms of communication based on context	● Understands when communication of results is better delivered in person as opposed to by phone
Level 3 *Communicates clearly, concisely, timely, and in an organized written form, including anticipatory guidance*	● Provides a printed after visit summary for the patient outlining recommendations
Achieves written or verbal communication (e.g., patient notes, email) that serves as an example for others to follow	● Develops a template for a metabolic emergency letter
Level 4 *Provides feedback to more junior learners on reporting diagnostic and therapeutic reasoning in the patient record*	● Critiques and edits genetic residents' notes to improve the clarity of the therapy plan and rationale for selecting from different therapeutic options for a patient with Gaucher disease
Mentors more junior learners in written and verbal communication	● Provides real-time feedback to genetics residents when a metabolic patient does not understand instructions a resident is providing
Level 5 *Models feedback to improve others' written communication*	● Provides education for hospital policy related to acute metabolic decompensation and emergency visits
Guides departmental or institutional communication around policies and procedures	
Assessment Models or Tools	● Direct observation ● Faculty member evaluation

	● Medical record (chart) audit ● Multisource feedback ● Resident self-reflection ● Peer-to-peer evaluation
Curriculum Mapping	●
Notes or Resources	● 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://www.scholars.northwestern.edu/en/publications/promoting-responsible-electronic-documentation-validity-evidence-. 2021. ● Institutional policies on documentation and communication

To help programs transition to the new version of the Milestones, the ACGME has mapped the original Milestones 1.0 to the new Milestones 2.0. Indicated below are where the subcompetencies are similar between versions. These are not exact matches but are areas that include similar elements. Not all subcompetencies map between versions. Inclusion or exclusion of any subcompetency does not change the educational value or impact on curriculum or assessment.

Milestones 1.0	Milestones 2.0
PC1: Initial or Acute Management	PC1: History and Physical Examination
PC2: Chronic Management	PC2: Chronic Management
PC3: Newborn Screening	PC3: Newborn Screening
MK1: Molecular and Metabolic Mechanisms	MK1: Molecular and Metabolic Mechanisms
MK2: Newborn Screening	
MK3: Diagnostic Testing	MK2: Diagnostic Testing
SBP1: Systems Approach	SBP3: Physician Role in the Health Care Systems
SBP2: Newborn Screening Systems	PC3: Newborn Screening
SBP3: Transitions of Care	SBP2: System Navigation for Patient-Centered Care
PBLI1: Self-Directed Learning	PBLI1: Evidence-Based and Informed Practice
PBLI2: Process Improvement and Patient Safety	SBP1: Patient Safety and Quality Improvement
PROF1: Giving and Receiving Feedback	PBLI2: Reflective Practice and Commitment to Personal Growth
PROF2: Cultural Competency	PROF1: Professional Behavior and Ethical Principles
PROF3: Accountability and Integrity	PROF2: Accountability/Conscientiousness
	PROF3: Self-Awareness and Help-Seeking
ICS1: Communicates with Patients and Families	ICS1: Patient- and Family-Centered Communication
ICS2: Communication and Conflict Management within the Health Care Team	ICS2: Interprofessional and Team Communication
	ICS3: Communication within Health Care Systems

Available Milestones Resources

Milestones 2.0: Assessment, Implementation, and Clinical Competency Committees Supplement, 2021 - *https://meridian.allenpress.com/jgme/issue/13/2s*

Milestones Guidebooks: https://www.acgme.org/milestones/resources/

- *Assessment Guidebook*
- *Clinical Competency Committee Guidebook*
- *Clinical Competency Committee Guidebook Executive Summaries*
- *Implementation Guidebook*
- *Milestones Guidebook*

Milestones Guidebook for Residents and Fellows: https://www.acgme.org/residents-and-fellows/the-acgme-for-residents-and-fellows/

- Milestones Guidebook for Residents and Fellows
- Milestones Guidebook for Residents and Fellows Presentation
- Milestones 2.0 Guide Sheet for Residents and Fellows

Milestones Research and Reports: https://www.acgme.org/milestones/research/

- *Milestones National Report*, updated each fall
- *Milestones Predictive Probability Report,* updated each fall
- *Milestones Bibliography*, updated twice each year

Developing Faculty Competencies in Assessment courses - https://www.acgme.org/meetings-and-educational-activities/courses-and-workshops/developing-faculty-competencies-in-assessment/

Assessment Tool: Direct Observation of Clinical Care (DOCC) - https://dl.acgme.org/pages/assessment

Assessment Tool: Teamwork Effectiveness Assessment Module (TEAM) - https://team.acgme.org/

Improving Assessment Using Direct Observation Toolkit - https://dl.acgme.org/pages/acgme-faculty-development-toolkit-improving-assessment-using-direct-observation

Remediation Toolkit - https://dl.acgme.org/courses/acgme-remediation-toolkit

Learn at ACGME has several courses on Assessment and Milestones - https://dl.acgme.org/