# Supplemental Guide:
# Medical Genetics and Genomics

ACGME

August 2019

## Milestones Supplemental Guide

This document provides additional guidance and examples for the Medical 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.

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
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> To promote oversight of resident supervision while providing for graded authority and responsibility, the program must use the following classification of supervision:
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> Direct Supervision – the supervising physician is physically present with the resident and patient.
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> Indirect Supervision:
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> 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.
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> 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.
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> Oversight – the supervising physician is available to provide review of procedures/encounters with feedback provided after care is delivered

| **Patient Care 1: History and Physical Examination**<br>**Overall Intent:** To ensure the resident takes a medical and family history and performs a physical examination to identify salient genetic features and develops a differential diagnosis | |
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| **Milestones** | **Examples** |
| **Level 1** *Takes a general medical and family history*<br><br>*Completes a general physical examination* | ● Does a routine history and physical exam, but does not include a genetic history or dysmorphic features |
| **Level 2** *Takes a basic genetics-focused history and completes a basic pedigree*<br><br>*Completes a basic genetics-focused physical examination; identifies normal and abnormal phenotypic features and/or anomalies* | ● Asks if other family members have similar features as part of the history<br><br>● For a patient with possible neurofibromatosis type 1 (NF1), looks for café au lait macules during the physical exam but does not look for additional NF1-related features |
| **Level 3** *Takes a genetics-focused history with some pertinent positive and negative findings; completes an accurate pedigree*<br><br>*Completes a genetics-focused physical examination; identifies and accurately describes common phenotypic features and/or anomalies; recognizes common syndromes or disorders* | ● Acquires a detailed family genetic history and draws a three-generation pedigree using standard symbols<br><br>For a patient with possible NF1:<br>● History includes hypertension, learning disabilities, and psychiatric conditions<br>● Looks for and counts the size and number of café au lait macules, presence of neurofibromas, and axillary and inguinal freckling |
| **Level 4** *Takes a comprehensive genetic history with pertinent positive and negative findings; integrates the history with other data to develop a differential diagnosis*<br><br>*Identifies and accurately describes phenotypic features and/or anomalies using standardized nomenclature; recognizes complex syndromes or disorders* | For a patient with café au lait macules:<br>● During history, asks about cancer diagnosis in the patient and/or family<br>● Looks for the presence or absence of Noonan syndrome facial characteristics or features of other diagnoses on the differential |
| **Level 5** *Makes a nationally recognized contribution by describing a new genetic disorder or expanding the phenotype of a known syndrome or disorder* | ● Contributes to the identification of a new gene/syndrome associated with café au lait macules |
| Assessment Models or Tools | ● Direct observation<br>● Faculty evaluations |

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| | ● In-training exam<br>● Medical record (chart) audit<br>● Multisource feedback |
| Curriculum Mapping | ● |
| Notes or Resources | ● Nussbaum RL, McInnes RR, Williard HF. *Thompson & Thompson Genetics in Medicine.* 7th ed. Philadelphia, PA: Saunders; 2007.<br>● Gene Reviews<br>● Jones KL, Jones MC, del Campo M. *Smith's Recognizable Patterns of Malformations*. 7th ed. Philadelphia, PA: Saunders; 2013.<br>● Bennett R, Steinhaus French K, Resta R, Doyle DL. Standardized human pedigree nomenclature: update and assessment of the recommendations of the National Society of Genetic Counselors. *J Genet Couns*. 2008 Oct;17(5):424-33. |

| **Patient Care 2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions**<br>**Overall Intent:** To select correct genetic tests, know how to read and interpret genetic test reports, and provide appropriate care for patients with genetic conditions | |
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| **Milestones** | **Examples** |
| **Level 1** *Identifies the variety of testing modalities for genetic conditions* | ● Knows the difference between chromosomal microarray and a karyotype |
| *Identifies the components of the genetics test result* | ● Knows genetic test reports should include header, result summary, interpretation, recommendations, methodology, limitations, references, and contact information |
| *Recognizes the availability of intervention for some genetic conditions* | ● Knows phenylketonuria (PKU) is a genetic condition that can be treated |
| **Level 2** *Identifies basic testing options for common genetic disorders* | ● Orders chromosomal microarray analysis (CMA) for patient with multiple congenital anomalies |
| *Identifies resources to facilitate interpretation of positive, negative, and uncertain test results* | ● Lists ClinVar, Online Mendelian Inheritance in Man (OMIM), GeneReviews as potential resources for interpretation of test reports |
| *Identifies resources and guidelines for treatment and management of common genetic conditions* | ● Lists PubMed, American College of Medical Genetics (ACMG) Standards and Guidelines as possible resources to provide management |
| **Level 3** *Identifies strengths and limitations of testing methodologies in order to select first tier tests* | ● Orders karyotype instead of CMA for patient suspected of having Down syndrome |
| *Uses resources to interpret diagnostic test results in the context of the phenotype* | ● Uses Database of Genomic Variants (DGV), University of California Santa Cruz database, Miami acquired loss of heterozygosity (AOH) Tool to further interpret test reports |
| *Implements treatment and/or surveillance plans for common genetic conditions* | ● For a patient with PKU, places patient on metabolic formula, attempts KUVAN trial |
| **Level 4** *Selects and prioritizes testing options across a broad spectrum of complex disorders and inheritance patterns/ mechanisms* | ● Differentiates between when to order sequencing panel versus whole exome sequencing |
| *Uses resources to interpret ambiguous test results in the context of the phenotype* | ● Contributes case to GeneMatcher |

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| *Implements treatment and/or surveillance plans for complex genetic conditions* | ● For an adult patient with PKU, considers pegvaliase to maintain phe level of 120-360 umol/l (2-6 mg/dl) |
| **Level 5** *Contributes to the knowledge base for the refinement of ambiguous test results*<br><br>*Creates evidence-based guidelines for management* | ● Publishes findings on a variant reclassification<br><br>● Resident updates GeneReviews for PKU |
| Assessment Models or Tools | ● Direct observation<br>● Faculty member evaluations<br>● In-training exam<br>● Medical record (chart) audit |
| Curriculum Mapping | ● |
| Notes or Resources | ● University of California Santa Cruz. Genome Browser. https://genome.ucsc.edu/. 2018.<br>● Miami AOH Tool. http://firefly.ccs.miami.edu/cgi-bin/ROH/ROH_analysis_tool.cgi. 2018.<br>● GeneMatcher. https://www.genematcher.org/. 2018 |

| **Patient Care 3: Pre- and Post-Test Genetic Counseling**<br>**Overall Intent:** To understand and convey the reasoning for and possible results of genetic testing | |
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| **Milestones** | **Examples** |
| **Level 1** *Participates in pre-test counseling*<br><br>*Participates in post-test counseling* | ● Observes others providing pre- and post-testing counseling to patients and families with a diagnosis of intellectual disability |
| **Level 2** *Explains the rationale for the recommended testing*<br><br>*Explains the results of the test* | ● Explains to patient/family why CMA is a first-tier test for genetic evaluation of intellectual disability<br><br>● Communicates that a test was diagnostic or non-diagnostic to the patient/family |
| **Level 3** *Conveys the impact and limitations of disorder-specific targeted testing while obtaining informed consent*<br><br>*Conveys the impact and limitations of diagnostic and non-diagnostic results* | ● Explains to a family the possible need for testing in additional family members<br>● Explains that exome sequencing may not reliably detect triplet repeat disorders<br><br>● Communicates the difference between clinical and molecular diagnosis in the context of non-diagnostic result |
| **Level 4** *Clearly conveys the impact and limitations of complex untargeted testing while obtaining informed consent*<br><br>*Conveys the impact and limitations of unexpected and ambiguous results* | ● Resident effectively communicates possibility of identifying unexpected results including ACMG secondary findings, consanguinity, misattributed parentage, and/or variant of uncertain significance (VUS) |
| **Level 5** *Participates in the development of professional practice guidelines regarding testing and return of results* | ● Participates in ClinVar variant resolution<br>● Is a member of ACMG practice guidelines committee |
| Assessment Models or Tools | ● Direct observation<br>● Medical record (chart) audit<br>● Multisource feedback<br>● Resident self-reflection |
| Curriculum Mapping | ● |
| Notes or Resources | ● Kalia SS, Adelman K, Bale SJ, et al. Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. *Genet Med.* 2017 Feb;19(2):249-255. https://www.ncbi.nlm.nih.gov/pubmed/27854360.<br>● Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of |

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| | Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med.* 2015 May;17(5):405-24. https://www.ncbi.nlm.nih.gov/pubmed/25741868.<br>● ACMG. Practice guidelines. https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Practice-Guidelines.aspx. 2018.<br>● Uhlmann WR, Schuette JL, Yashar BM. *A Guide to Genetic Counseling*. 2nd ed. Danvers, MA: John Wiley & Sons, Inc; 2009. |

| **Medical Knowledge 1: Foundations of Genetics and Genomics**<br>**Overall Intent:** To progressively incorporate basic science knowledge into patient care | |
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| **Milestones** | **Examples** |
| **Level 1** *Demonstrates basic medical knowledge of embryology, inheritance, and genetic mechanism of disease*<br><br>*Demonstrates basic medical knowledge of gene and genome structure and function* | ● Demonstrates ability to differentiate autosomal dominant, recessive, X-linked, and mitochondrial inheritance<br>● Describes heart looping in development<br><br>● Describes basic types of genetic variants such as aneuploidies, single nucleotide variants (SNV), and copy number variations (CNV) |
| **Level 2** *Applies knowledge of embryology, inheritance, and genetic mechanism of disease to identify a differential diagnosis*<br><br>*Applies knowledge of gene and genome structure and function to identify a differential diagnosis* | ● Identifies consanguinity in a pedigree and recognizes risk for autosomal recessive conditions<br>● Describes the increased risk for fetal aneuploidies with advancing maternal age<br><br>● Identifies Fragile X and other X-linked disorders in the differential for a boy with developmental delay<br>● Describes mechanism of pleiotropy in genetic diseases |
| **Level 3** *Applies advanced knowledge of embryology, inheritance, and genetic mechanism of disease to make a diagnosis*<br><br>*Applies advanced knowledge of gene and genome structure and function to make a diagnosis* | ● In a child with retinoblastoma, tests for sporadic or inherited *RB1* variants<br><br>● Understands tumor suppressor mechanism and two-hit hypothesis of disease for a child with retinoblastoma |
| **Level 4** *Applies advanced knowledge of embryology, inheritance, and genetic mechanism of disease to diagnostic and therapeutic interventions*<br><br>*Applies advanced knowledge of gene and genome structure and function to diagnostic and therapeutic interventions* | ● For a patient with ovarian cancer and a pathogenic loss of function *BRCA1/2* variant, recognizes the implications for treatment with a PARP inhibitor<br><br>● For a patient with spinal muscular atrophy, recognizes the implications for treatment with nusinersen |
| **Level 5** *Contributes to peer-reviewed resources addressing genetic mechanism of disease*<br><br>*Recognized as a national expert in diagnosis and management of genetic disease* | ● Co-authorship on a peer-reviewed publication on forkhead stalling and template switching as a mechanism of genetic disease<br><br>● Contributes to a practice guideline in diagnosis or management of hereditary breast and ovarian cancer |

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| Assessment Models or Tools | ● Direct observation<br>● Faculty evaluations<br>● In-training exam<br>● Medical record (chart) audit |
| Curriculum Mapping | ● |
| Notes or Resources | ● Nussbaum RL, McInnes RR, Williard HF. *Thompson & Thompson Genetics in Medicine.* 7th ed. Philadelphia, PA: Saunders; 2007.<br>● National Cancer Comprehensive Network Practice Guideline. 2014. https://www.nccn.org/professionals/physician_gls/default.aspx.<br>● Valle D, Beaudet AL, Vogelstein B, et al. The online metabolic and molecular bases of inherited disease. The McGraw-Hill Companies, Inc. 2018. https://ommbid.mhmedical.com/book.aspx?bookid=971<br>● Gardner RJM, Sutherland GR, Shaffer LG. *Chromosome Abnormalities and Genetic Counseling*. 4th ed. New York, NY: Oxford University Press; 2012.<br>● Erickson RP, Wynshaw-Boris AJ. *Epstein's Inborn Errors of Development: The Molecular Basis of Clinical Disorders of Morphogenesis*. 3rd ed. New York, NY: Oxford University Press; 2016.<br>● Coleman WB, Tsongalis GJ. *The Molecular Basis of Human Cancer.* 2nd ed. New York, NY: Springer Science+Business, Media: 2017. |

| **Medical Knowledge 2: Clinical Genetics and Genomics**<br>**Overall Intent:** To recognize and diagnose genetic syndromes, including genotype/phenotype relationships | |
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| **Milestones** | **Examples** |
| **Level 1** *Recognizes syndromic and non-syndromic etiologies*<br><br>*Recognizes that phenotypes evolve across the lifespan* | ● Understands that there are genetic (chromosomal) and non-genetic causes of intellectual disability (ID)<br><br>● Appreciates that children with Down syndrome have different concerns at different ages |
| **Level 2** *Identifies syndromic and non-syndromic etiologies*<br><br>*Identifies the changes of phenotypes across the lifespan* | ● Identifies genetic causes of ID (trisomy 21) and non-genetic causes of ID (fetal alcohol syndrome)<br><br>● For a patient with Down syndrome, appreciates that newborn concerns include hypotonia, feeding, and cardiac issues, whereas adults are at risk for Alzheimer's disease |
| **Level 3** *Demonstrates knowledge of syndromic and non-syndromic etiologies and the impact on diagnosis and management*<br><br>*Demonstrates knowledge of the changes in phenotypes across the lifespan and how it impacts diagnosis and management* | ● Recognizes that a patient with an isolated cleft lip and palate may not need ongoing genetic surveillance<br>● Recognizes the need for evaluation and/or surveillance of multiple systems in a patient with a Down syndrome diagnosis<br>● Recognizes that management of feeding for a patient with Prader-Willi syndrome is age dependent |
| **Level 4** *Applies knowledge of syndromic and non-syndromic etiologies to diagnosis and management*<br><br>*Applies knowledge of the changes in phenotypes across the lifespan and how it impacts diagnosis and management* | ● Orders flexion/extension cervical spine radiographs for a five-year-old with Down syndrome<br><br>● Discusses pre-implantation genetic testing with a 30-year-old diagnosed with a *BRCA1/2* pathogenic variant |
| **Level 5** *Serves as an expert resource for syndromic and/or non-syndromic etiologies*<br><br>*Contributes to peer-reviewed resources addressing natural history of genetic disease* | ● Publishes a peer-reviewed publication on risk of premature ovarian insufficiency in women who are premutation carriers of Fragile X |
| Assessment Models or Tools | ● Direct observation<br>● Faculty evaluation<br>● In-training exam<br>● Medical record (chart) audit |

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| | ● Multisource feedback<br>● Resident self-reflection |
| Curriculum Mapping | ● |
| Notes or Resources | ● Nussbaum RL, McInnes RR, Williard HF. *Thompson & Thompson Genetics in Medicine.* 7th ed. Philadelphia, PA: Saunders; 2007.<br>● GeneReviews<br>● Online Mendelian Inheritance in Man. An online catalog of human genes and genetic disorders. https://www.omim.org. 2018.<br>● ACMG and other professional practice guidelines for diagnosis and surveillance of genetic conditions |

| **Medical Knowledge 3: Clinical Reasoning**<br>**Overall Intent:** To integrate information obtained to generate a differential diagnosis and evaluation plan | |
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| **Milestones** | **Examples** |
| **Level 1** *Demonstrates a basic framework for clinical reasoning*<br><br>*Identifies appropriate resources to inform clinical reasoning* | ● Evaluates a patient with developmental delay, notes pertinent findings, and generates a differential diagnosis, but is unable to prioritize<br><br>● In the evaluation of a patient with cleft lip and palate, articulates that there are Mendelian and non-Mendelian causes |
| **Level 2** *Demonstrates clinical reasoning to determine relevant information*<br><br>*Selects relevant resources based on scenario to inform decisions* | ● In a patient with cleft lip and palate, looks for other major and minor birth defects associated with syndromic forms of cleft lip and palate<br><br>● Uses resources like OMIM, GeneReviews and facial recognition software to support a differential diagnosis |
| **Level 3** *Synthesizes information to inform clinical reasoning, with assistance*<br><br>*Seeks and integrates evidence-based information to inform diagnostic decision making in complex cases, with assistance* | ● In a patient with cleft lip and palate, prioritizes a differential diagnosis based on history and physical exam findings<br><br>● Generates a genetic testing plan based on the differential diagnosis and relevant practice diagnostic guidelines |
| **Level 4** *Independently synthesizes information to inform clinical reasoning in complex cases*<br><br>*Independently seeks out, analyzes and applies relevant original research to diagnostic decision making in complex clinical cases* | ● Analyzes genetic testing results in setting of the patient presentation<br>● Integrates non-diagnostic genetic testing results to re-evaluate and formulate a new plan<br><br>● Finds and integrates information from recent peer-reviewed journal publications to support the diagnosis |
| **Level 5** *Develops a novel approach for the assessment of complex cases* | ● Identifies novel biomarkers for diagnosis of hypermobile Ehlers-Danlos syndromes (EDS) |
| Assessment Models or Tools | ● Direct observation<br>● Faculty evaluations<br>● In-training exam<br>● Medical record (chart) audit |
| Curriculum Mapping | ● |
| Notes or Resources | ● Online Mendelian Inheritance in Man. An online catalog of human genes and genetic disorders. https://www.omim.org. 2018.<br>● Genereviews. www.genereviews.org. 2018.<br>● Exome Aggregation Consortium (ExAC). http://exac.broadinstitute.org/. 2018. |

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| | ● Genome Aggregation Database (GnomAD). https://gnomad.broadinstitute.org/. 2018.<br>● ClinVar. https://www.ncbi.nlm.nih.gov/clinvar/. 2018.<br>● London Dysmorphology Database. https://www.face2gene.com/lmd-library-london-medical-database-dysmorphology/. 2018. |

| **Systems-Based Practice 1: Patient Safety and Quality Improvement (QI)**<br>**Overall Intent:** To identify, report, analyze, and disclose patient safety events and participate in a QI project | |
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| **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 |
| *Discloses patient safety events to patients and families (simulated or actual)* | ● Independently discloses the erroneous administration of IV fluid to a patient/caregiver |

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| *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*<br><br>*Role models or mentors others in the disclosure of patient safety events*<br><br>*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<br><br>● Coaches a resident on disclosure of a safety event related to a medication error<br><br>● Completes and shares outcomes of a full PDSA cycle related to improved access to clinic appointments |
| Assessment Models or Tools | ● Direct observation<br>● Institutional patient safety e-module multiple choice tests<br>● Medical record (chart) audit<br>● Portfolio |
| Curriculum Mapping | ● |
| Notes or Resources | ● Institute of Healthcare Improvement website, which includes multiple choice tests, reflective writing samples, and more. http://www.ihi.org/Pages/default.aspx. 2018.<br>● American Academy of Family Physicians. Basic of Quality Improvement. https://www.aafp.org/practice-management/improvement/basics.html. 2018. |

| **Systems-Based Practice 2: System Navigation for Patient-Centered Care**<br>**Overall Intent:** To navigate the health care system to adapt care to a specific patient population to ensure high-quality patient outcomes | |
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| **Milestones** | **Examples** |
| **Level 1** *Demonstrates knowledge of care coordination*<br><br>*Identifies key elements for safe and effective transitions of care and hand-offs*<br><br>*Demonstrates knowledge of population and community health needs and disparities* | ● Identifies the members of the interprofessional team and describes their roles, but is not yet routinely using team members or accessing resources<br><br>● Recognizes the essential components of an effective sign-out<br><br>● 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*<br><br>*Performs safe and effective transitions of care/hand-offs in routine clinical situations*<br><br>*Identifies specific population and community health needs and inequities for the local population* | ● Contacts interprofessional team members and consultants for necessary referrals for a patient with Down syndrome<br><br>● Performs a basic sign-out, but still needs guidance for anticipated events<br><br>● 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*<br><br>*Performs safe and effective transitions of care/hand-offs in complex clinical situations*<br><br>*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<br><br>● Provides anticipatory guidance for unstable patients including recommendations for how to escalate treatments for patients with uncontrolled ammonia levels<br><br>● Works with the social worker/health navigator to ensure patients with low literacy understand how to access resources over time |
| **Level 4** *Role 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 |

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| *Role 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* | ● Proactively calls the outpatient clinicians to communicate status updates and goals of care |
| *Participates in changing and adapting practice to provide for the needs of specific populations including advocating for a patient's genetic testing coverage* | ● 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 improvements* | ● Creates order set for patients with metabolic disorders presenting to the emergency department |
| *Improves quality of transitions of care within and across health care delivery systems to optimize patient outcomes* | ● Develops protocols for pre- and intra-transplant patients with urea cycle disorder and measures patient outcomes |
| *Leads innovations and advocates for populations and communities with health care inequities at the state or federal level* | ● Collaborates with key stakeholders at the state level to ensure patients with PKU receive access to metabolic formula throughout the life span |
| Assessment Models or Tools | ● Direct observation<br>● Medical record (chart) audit<br>● Multisource feedback<br>● Review of written sign-out/hand-off tools |
| Curriculum Mapping | ● |
| Notes or Resources | ● Agency for Healthcare Research and Quality. Patient Safety Network. Handoffs and signouts. January 2019. https://psnet.ahrq.gov/primers/primer/9/resource.aspx?resourceID=18439.<br>● 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.<br>● IPASS. Patient Safety Institute. https://ipassinstitute.com. 2018. |

| **Systems-Based Practice 3: Physician Role in Health Care Systems**<br>**Overall Intent:** To navigate the health care system to improve patient care and the health system's performance | |
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| **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 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 test result 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 depending upon insurance coverage, co-payments, and deductibles |

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| *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 |
| **Level 4** *Manages various components of the complex health care system to provide efficient and effective patient care and transition of care*<br><br>*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*<br><br>*Analyzes individual practice patterns and professional requirements in preparation for practice* | ● Manages transition from hospital to outpatient treatment for a patient with metabolic disorder<br><br>● If insurance denies genetic testing for a patient, discuss alternatives such as research protocols, clinical trials, charity funding, and self-payment<br><br>● 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*<br><br>*Participates in health policy advocacy activities*<br><br>*Educates others to prepare them for transition to practice* | ● Presents institution-specific data to show rapid exome sequencing reduces neonatal intensive care unit length of stay<br><br>● Develops e-consults or telehealth services to increase access to genetic services for rural and underserved patient populations<br><br>● Counsels residents on transition to practice |
| Assessment Models or Tools | ● Direct observation<br>● Medical record (chart) audit<br>● Multisource feedback<br>● Resident self-reflection |
| Curriculum Mapping | ● |
| Notes or Resources | ● Agency for Healthcare Research and Quality (AHRQ): The Challenges of Measuring Physician Quality https://www.ahrq.gov/professionals/quality-patient-safety/talkingquality/create/physician/challenges.html. 2018. |

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| | ● AHRQ. Major physician performance sets: https://www.ahrq.gov/talkingquality/measures/setting/physician/measurement-sets.html. 2018.<br>● Institutional templates for letters of medical necessity<br>● ACMG Policy Guidelines https://www.acmg.net/ACMG/Advocacy/Policy-Statements/ACMG/Advocacy/Policy-Statements.aspx. 2018.<br>● NEJM. Navigating the Transition from Residency to Physician Practice. 2016. https://www.nejmcareercenter.org/article/navigating-the-transition-from-residency-to-physician-practice/. 2018.<br>● American Medical Association. Tips for negotiating employee contracts. https://www.ama-assn.org/tips-negotiating-employment-contracts. 2018. |

| **Practice-Based Learning and Improvement 1: Evidence-Based and Informed Practice**<br>**Overall Intent:** To evaluate and incorporate evidence and patient values into clinical practice | |
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| **Milestones** | **Examples** |
| **Level 1** *Demonstrates how to access and use available evidence, and incorporate patient preferences and values in order to take care of a routine patient* | ● Identifies clinical practice guideline for evaluation of a patient with Turner syndrome<br>● Understands that patient values affect care |
| **Level 2** *Articulates clinical questions and elicits patient preferences and values in order 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 genetic 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 genetic disorders |
| Assessment Models or Tools | ● Direct observation<br>● In-training exam<br>● Faculty evaluations<br>● Multisource feedback<br>● Resident self-reflection |
| Curriculum Mapping | ● |
| Notes or Resources | ● Pubmed search<br>● Online Mendelian Inheritance in Man. An online catalog of human genes and genetic disorders. https://www.omim.org. 2018.<br>● GeneReviews. www.genereviews.org. 2018.<br>● Professional Practice Guidelines<br>● Cochrane Library. Cochrane Database of Systematic Reviews. https://www.cochranelibrary.com/cdsr/about-cdsr. 2018. |

| **Practice-Based Learning and Improvement 2: Reflective Practice and Commitment to Personal Growth**<br>**Overall Intent:** To seek clinical performance information to improve care and develop objectives and goals for improvement | |
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| **Milestones** | **Examples** |
| **Level 1** *Accepts responsibility for personal and professional development by establishing goals*<br><br>*Identifies the factors which contribute to gap(s) between expectations and actual performance*<br><br>*Actively seeks opportunities to improve* | ● Sets a personal practice goal of documenting use of the revised Ghent criteria for evaluation of patients for possible Marfan syndrome<br><br>● Identifies gaps in knowledge of metabolic pathways<br><br>● Asks for feedback from patients, families, and patient care team members |
| **Level 2** *Demonstrates openness to performance data (feedback and other input) in order to inform goals*<br><br>*Analyzes and reflects on the factors which contribute to gap(s) between expectations and actual performance*<br><br>*Designs and implements a learning plan, with prompting* | ● Integrates feedback to adjust the documentation of the revised Ghent criteria for evaluation of patients for possible Marfan syndrome<br><br>● Assesses time management skills and how it impacts timely completion of clinic notes and literature reviews<br><br>● When prompted, develops individual education plan to improve their evaluation of VUS |
| **Level 3** *Seeks performance data episodically, with adaptability and humility*<br><br>*Analyzes, reflects on, and institutes behavioral change(s) to narrow the gap(s) between expectations and actual performance*<br><br>*Independently creates and implements a learning plan* | ● Does a chart audit to determine the percent of patients evaluated for possible Marfan syndrome which documents all components of revised Ghent criteria<br><br>● Completes a comprehensive literature review prior to patient encounters<br><br>● Using web-based resources, creates a personal curriculum to improve his/her evaluation of VUS |
| **Level 4** *Seeks performance data consistently with adaptability and humility* | ● Completes a quarterly chart audit to ensure documentation of the revised Ghent criteria for evaluation of patients for possible Marfan syndrome |

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| *Challenges assumptions and considers alternatives in narrowing the gap(s) between expectations and actual performance*<br><br>*Uses performance data to measure the effectiveness of the learning plan and when necessary, improves it* | ● 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<br><br>● Performs a chart audit on personal documentation of their evaluation of VUS |
| **Level 5** *Serves as a role model in seeking performance data with adaptability and humility*<br><br>*Mentors others on reflective practice*<br><br>*Facilitates the design and implementing learning plans for others* | ● Models practice improvement and adaptability<br><br>● Develops educational module for collaboration with other patient care team members<br><br>● Assists first-year residents in developing their individualized learning plans |
| Assessment Models or Tools | ● Direct observation<br>● Medical record (chart) audit<br>● Mentored review of individualized learning plan<br>● Multisource feedback |
| Curriculum Mapping | ● |
| Notes or Resources | ● Hojat M, Veloski JJ, Gonnella JS. Measurement and correlates of physicians' lifelong learning. *Acad Med.* 2009. Aug;84(8):1066-74. doi: 10.1097 /ACM. 0b013e 3181acf25f. NOTE: Contains a validated questionnaire about physician lifelong learning.<br>● Lockspeiser TM, Schmitter PA, Lane JL, et al. Assessing fellows' written learning goals and goal writing skill: validity evidence for the learning goal scoring rubric. *Acad Med*. 2013. 88 (10)<br>● Burke AE, Benson B, Englander R, Carraccio C, Hicks PJ. Domain of competence: practice-based learning and improvement. *Academic Pediatrics* 2014. 14: S38-S54.<br>● https://journals.lww.com/academicmedicine/FullText/2016/10000/The_Relationship_Between_Academic_Motivation_and.28.aspx.<br>● Hauer J, Quill T. Educational needs assessment, developing learning objectives, and choosing a teaching approach. *Journal of Palliative Medicine*. 2011. Volume 14 Number 4. Doi: 10.1089/jpm.2010.0232. |

| **Professionalism 1: Professional Behavior and Ethical Principles**<br>**Overall Intent:** To demonstrate ethical and professional behavior and identify and manage lapses in self and others | |
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| **Milestones** | **Examples** |
| **Level 1** *Demonstrates compassion, sensitivity, honesty and 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 and 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, and integrity and serves as a role model to others* | ● Models empathy for a patient and family making end-of-life care decisions |

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| *Recognizes and uses appropriate resources for managing and resolving ethical dilemmas as needed*<br><br>*Demonstrates consistently professional behavior with regard to conflicts of interest relevant to presentations, publishing, consulting, and service* | ● Collaborates with the ethics committee to address 50-year-old woman with sickle cell disease who wants to use assisted reproductive technologies<br><br>● Respects a families desire to not be included in a research publication |
| **Level 5** *Coaches others when their behavior fails to meet professional expectations*<br><br>*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<br><br>● Develops a patient-centered guideline for addressing non-beneficial treatments |
| Assessment Models or Tools | ● Direct observation<br>● Institutional ethics and conflict of interest modules<br>● Institutional reporting of conflict of interest<br>● Multisource feedback<br>● Resident self-reflection<br>● Simulation |
| Curriculum Mapping | ● |
| Notes or Resources | ● American Society of Human Genetics Code of Ethics. https://www.ashg.org/about/ethics.shtml. 2018.<br>● American Medical Association Code of Ethics. https://www.ama-assn.org/delivering-care/ama-code-medical-ethics. 2018.<br>● American Board of Internal Medicine; 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. https://abimfoundation.org/wp-content/uploads/2015/12/Medical-Professionalism-in-the-New-Millenium-A-Physician-Charter.pdf<br>● 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<br>● Levinson W, Ginsburg S, Hafferty FW, Lucey CR. *Understanding Medical Professionalism*. McGraw-Hill Education, 2014. |

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| | ● 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. 2018.<br>● Jonsen AR. *Clinical Ethics, A Practical Approach to Ethical Decisions in Clinical Medicine*. 8th ed. McGraw-Hill. 2015.<br>● Living with Grief: Ethical Dilemmas at the End of Life. Kenneth Doka. Quality Books. 2005.<br>● CITI Training Modules, University of Miami. https://about.citiprogram.org/en/homepage/. 2018. |

| **Professionalism 2: Accountability/Conscientiousness**<br>**Overall Intent:** To take responsibility for one's actions and the impact on patients and other members of the health care team | |
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| **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 |
| *Recognizes situations that may impact his/her own ability to complete tasks and responsibilities in a timely manner* | ● Recognizes that personal sudden illness may interfere with assigned tasks |
| *Demonstrates a professional appearance, daily demeanor, and conduct* | ● Dresses professionally<br>● Treats 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 |
| *Sets a standard for appearance, daily demeanor, and conduct as a professional* | ● Meets institutional requirements and expectations for conduct and dress |

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| **Level 4** *Recognizes and addresses situations that may impact others' ability to complete tasks and responsibilities in a timely manner*<br><br>*Promotes professional appearance, demeanor, and conduct in their peers and associates* | ● Collaborates with the team and recognizes overburdened associates and assists with patient care<br><br>● 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<br>● Direct observation<br>● Multisource feedback<br>● Resident self-evaluation<br>● Rotation evaluations |
| Curriculum Mapping | ● |
| Notes or Resources | ● ABIM Foundation, ACP-ASIM Foundation, and European Federation of Internal Medicine. Medical professionalism in the new millennium: a physician charter. *Ann Intern Med*. 2002;136(3):243-6.<br>● Institutional Code of Conduct |

| **Professionalism 3: Self-Awareness and Help-Seeking**<br>**Overall Intent:** To identify, use, manage, improve, and seek help for personal and professional well-being for self and others | |
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| **Milestones** | **Examples** |
| **Level 1** *Recognizes status of personal and professional well-being, with assistance*<br><br>*Recognizes limits in the knowledge/skills of self or team, with assistance* | ● Acknowledges own response to patient's fatal genetic diagnosis<br><br>● Receives feedback on missed emotional cues after a family meeting |
| **Level 2** *Independently recognizes status of personal and professional well-being*<br><br>*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<br><br>● 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*<br><br>*With assistance, proposes a plan to remediate or improve limits in the knowledge/ skills of self or team* | ● With the multi-disciplinary team, develops a reflective response to deal with personal impact of difficult patient encounters and disclosures<br><br>● Integrates feedback from the multi-disciplinary 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*<br><br>*Independently develops a plan to remediate or improve limits in the knowledge/skills of self or team* | ● Independently identifies ways to manage personal stress<br><br>● 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<br>● Works with multi-disciplinary team to develop a feedback framework for learners around family meetings |
| Assessment Models or Tools | ● Direct observation<br>● Group interview or discussions for team activities<br>● Participation in institutional well-being programs<br>● Resident self-reflection<br>● Review of learning plan |

| Curriculum Mapping | ● |
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| Notes or Resources | ● Local resources, including Employee Assistance Program<br>● Institutional GME guidelines regarding resident wellness<br>● ACGME Tools and Resources for Resident and Faculty Member Well-Being https://www.acgme.org/What-We-Do/Initiatives/Physician-Well-Being/Resources. 2018.<br>● Stanford Medicine. WELLMD https://wellmd.stanford.edu<br>● American Academy of Pediatrics. Resilience Curriculum, Part D: Resilience in the face of grief and loss. https://www.aap.org/en-us/advocacy-and-policy/aap-health-initiatives/hospice-palliative-care/Pages/Resilience-Curriculum.aspx. 2018. |

| **Interpersonal and Communication Skills 1: Patient and Family-Centered Communication**<br>**Overall Intent:** To employ listening, language, behaviors, and self-awareness to form a therapeutic relationship that facilitates effective communication | |
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| **Milestones** | **Examples** |
| **Level 1** *Uses language and nonverbal behavior to demonstrate respect and establish rapport*<br><br>*Identifies common barriers to effective communication while accurately communicating own role within the health care system*<br><br>*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 him/herself to the patient/family and states their role in the care of the patient<br><br>● Reflects on how the use of silence and active listening assists in establishing patient/caregiver rapport<br>● Identifies the need for an interpreter for a patient/caregiver who is non-English speaking<br><br>● Understands that different patients may have different goals for a clinic visit |
| **Level 2** *Establishes a therapeutic relationship in straightforward encounters using active listening and clear language*<br><br>*Identifies complex barriers to effective communication*<br><br>*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 a family history of breast cancer, develops shared goals (contracting)<br>● Refrains from the use of overly complicated terminology<br><br>● Demonstrates therapeutic relationship with appropriate use of silence and normalizing emotional responses<br><br>● Identifies non-English-speaking patient who prefers to defer decision-making to their caregiver as a potential communication challenge |
| **Level 3** *Establishes a therapeutic relationship in challenging patient encounters*<br><br>*When prompted, reflects on personal biases while attempting to minimize communication barriers*<br><br>*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<br><br>● Identifies and reflects on personal bias towards patient autonomy over cultural preferences in decision making<br><br>● Delivers sensitive medical information to patients/families privately<br>● With guidance, collects and incorporates patient and family values into the medical decision-making process |

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| **Level 4** *Establishes therapeutic relationships, with attention to patient/family concerns and context, regardless of complexity*<br><br>*Recognizes personal biases while attempting to proactively minimize communication barriers*<br><br>*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 are in disagreement about the therapeutic management of their child<br><br>● Discusses the option of pregnancy termination despite conflict with the learner's personal values<br><br>● 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*<br><br>*Role models self-awareness practice while identifying teaching a contextual approach to minimize communication barriers*<br><br>*Role 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<br><br>● Coaches a learner to acknowledge personal bias and successfully manage communication with non-English-speaking patient |
| Assessment Models or Tools | ● Direct observation<br>● Faculty evaluations<br>● Multisource feedback<br>● Resident self-reflection |
| Curriculum Mapping | ● |
| Notes or Resources | ● Uhlmann WR, Schuette JL, Yashar BM. *A Guide to Genetic Counseling*. 2nd ed. Danvers, MA: John Wiley & Sons, Inc; 2009.<br>● Veach PC, LeRoy BS, Callanan NP. *Facilitating the Genetic Counseling Process: Practice Based Skills*. 2nd ed. Cham, Switzerland: Spring International Publishing, AG; 2018.<br>● 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. 2018. https://www.acmg.net/PDFLibrary/Ethical-Policy-Issues-Genetic-Screening-Children.pdf. |

| **Interpersonal and Communication Skills 2: Interprofessional and Team Communication**<br>**Overall Intent:** To communicate with the interdisciplinary team and other health care providers | |
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| **Milestones** | **Examples** |
| **Level 1** *Respectfully requests a consultation*<br><br>*Respectfully receives a consultation request*<br><br>*Uses language that values all members of the health care team* | ● When asking for a cardiology consultation for a patient with Marfan syndrome, respectfully relays the diagnosis and need to assess the aortic root diameter<br>● Receives consult request for a patient with Down syndrome, asks clarifying questions politely, and expresses gratitude for the consult<br>● Acknowledges the contribution of each member of the metabolic team to the patient |
| **Level 2** *Clearly and concisely requests a consultation*<br><br>*Clearly and concisely responds to a consultation request*<br><br>*Communicates information effectively with all health care team members* | ● Communicates diagnostic evaluation recommendations clearly and concisely in an organized and timely manner<br><br>● Sends a message in electronic health record to the dietician of a metabolic patient to increase the protein restriction |
| **Level 3** *Checks own understanding of consultant recommendations*<br><br>*Checks requestor's understanding of recommendations when providing consultation*<br><br>*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<br><br>● When receiving treatment recommendations from an attending physician, repeats back the plan to ensure understanding |
| **Level 4** *Coordinates recommendations from different members of the health care team to optimize patient care*<br><br>*Provides information to the primary care team regarding rationale for recommendations*<br><br>*Models active listening to other health care team members* | ● Initiates a multidisciplinary meeting to developed shared care plan for a patient with 22q11.2 deletion syndrome<br><br>● Explains rationale for chromosome analysis instead of chromosome microarray analysis as the preferred diagnostic test for suspected Down syndrome<br><br>● Asks other members of the health care team to repeat back recommendations to ensure understanding |

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| **Level 5** *Role 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<br>● Faculty evaluation<br>● Medical record (chart) audit<br>● Multisource feedback<br>● Resident self-reflection |
| Curriculum Mapping | ● |
| Notes or Resources | ● 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.<br>● 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<br>● Youngwerth J, Twaddle M. Cultures of interdisciplinary teams: how to foster good dynamics. *J Palliat Med*. 2011;14(5):650-654. |

| **Interpersonal and Communication Skills 3: Communication within Health Care Systems**<br>**Overall Intent:** To communicate through established institutional pathways using a variety of methods | |
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| **Milestones** | **Examples** |
| **Level 1** *Accurately records information in the patient record*<br><br>*Safeguards patient personal health information* | ● Accurately documents a telephone communication encounter with a patient<br><br>● Logs off computer when leaving clinical workstation |
| **Level 2** *Demonstrates organized diagnostic and therapeutic reasoning through notes in the patient record*<br><br>*Uses documentation shortcuts accurately, appropriately and in a timely manner*<br><br>*Documents required data in formats specified by institutional policy* | ● Documents a differential diagnosis and justifies recommendations<br><br>● Accurately edits “Copy Forward” notes<br><br>● Documents a telephone communication encounter within 24 hours |
| **Level 3** *Concisely reports diagnostic and therapeutic reasoning in the patient record*<br><br>*Appropriately selects direct (e.g., telephone, in-person) and indirect (e.g., progress notes, text messages) forms of communication based on context* | ● Documents rationale for progression of testing in the work-up of a patient with developmental delay<br><br>● Understands when communication of results are better delivered in person as opposed to by telephone |
| **Level 4** *Communicates clearly, concisely, timely, and in an organized written form, including anticipatory guidance*<br><br>*Achieves written or verbal communication (e.g., patient notes, e-mail) that serves as an example for others to follow* | ● Provides a printed after visit summary for the patient outlining recommendations<br><br>● Develops a template for a metabolic emergency letter |
| **Level 5** *Models feedback to improve others’ written communication*<br><br>*Guides departmental or institutional communication around policies and procedures* | • Provides education for hospital policy related to acute metabolic decompensation and emergency visits |
| Assessment Models or Tools | ● Direct observation |

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| | ● Faculty evaluation<br>● Medical record (chart) audit<br>● Multisource feedback<br>● Resident self-reflection |
| 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 Oct-Dec;29(4):420-432.<br>● Institutional policies on documentation and communication |

In an effort to aid programs in the transition to using the new version of the Milestones, the original Milestones 1.0 have been mapped to the new Milestones 2.0. Where the subcompetencies are similar between versions has been noted below. These are not necessarily exact matches, but are areas that include some similar elements. Note that 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: Obtain and interpret medical, social, and family histories, as well as physical exam findings necessary for the evaluation of patients with or at-risk for genetic disorders | PC1: History and Physical Examination |
| PC2: Incorporate genetic tests into patient management | PC2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions |
| PC3: Incorporate whole genome or exome tests into patient management | PC2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions<br>PC3: Pre- and Post-Test Counseling<br>MK1: Foundations of Genetics and Genomics<br>MK3: Clinical Reasoning |
| PC4: Diagnose and manage patients with inborn errors of metabolism | PC2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions<br>PC3: Pre- and Post-Test Counseling<br>MK1: Foundations of Genetics and Genomics<br>MK2: Clinical Genetics and Genomics<br>MK3: Clinical Reasoning |
| PC5: Evaluates infants with abnormal newborn screens in a cost-effective and sensitive manner and educates community providers | PC2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions<br>PC3: Pre- and Post-Test Counseling<br>MK1: Foundations of Genetics and Genomics<br>MK2: Clinical Genetics and Genomics<br>MK3: Clinical Reasoning |
| PC6: Develop proficiency in cancer genetics | PC2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions<br>PC3: Pre- and Post-Test Counseling<br>MK1: Foundations of Genetics and Genomics<br>MK2: Clinical Genetics and Genomics<br>MK3: Clinical Reasoning |

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| PC7: Evaluate and manage patients with single malformations, multiple congenital anomalies, developmental disabilities, and growth abnormalities by utilizing knowledge of embryology, teratology, developmental pathways, pathophysiology, and etiologic mechanisms | PC2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions<br>PC3: Pre- and Post-Test Counseling<br>MK1: Foundations of Genetics and Genomics<br>MK2: Clinical Genetics and Genomics<br>MK3: Clinical Reasoning |
| PC8: Develop proficiency in prenatal risks assessment, screening, diagnosis, and counseling | PC1: History and Physical Examination<br>PC2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions<br>PC3: Pre- and Post-Test Counseling<br>MK1: Foundations of Genetics and Genomics<br>MK2: Clinical Genetics and Genomics<br>MK3: Clinical Reasoning |
| PC9: Provide longitudinal management and reproductive counseling in pregnancies with known or suspected genetic conditions in the mother or fetus | PC1: History and Physical Examination<br>PC2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions<br>PC3: Pre- and Post-Test Counseling<br>MK1: Foundations of Genetics and Genomics<br>MK2: Clinical Genetics and Genomics<br>MK3: Clinical Reasoning |
| MK1: Apply knowledge of anatomy, development, pathophysiology, natural history, clinical history, and inheritance to provide counseling, anticipatory guidance, and longitudinal management to patients with multisystem genetic disorders | MK1: Foundations of Genetics and Genomics<br>MK2: Clinical Genetics and Genomics<br>MK3: Clinical Reasoning |
| MK2: Assess and participate in a clinical translational research study or clinical trial involving patients with or at-risk for a genetic disorder | None |
| SBP1: Function effectively within the larger context of health care systems, practice cost-effective medicine | SBP2: System Navigation for Patient-Centered Care<br>SBP3: Physician Role in the Health Care Systems |
| SBP2: Use technology to accomplish safe health care delivery | ICS3: Communication within Health Care Systems |
| PBLI1: Self-Directed Learning | PBLI1: Evidence-Based and Informed Practice<br>PBLI2: Reflective Practice and Commitment to Personal Growth |
| PBLI2: Implement a quality improvement project | SBP1: Patient Safety and Quality Improvement |

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| PROF1: Is sensitive and responsive to diverse patient populations with respect to gender, age, culture, race, religion, disabilities, and sexual orientation | PROF1: Professional Behavior and Ethical Principles<br>ICS1: Patient- and Family-Centered Communication |
| PROF2: Adhere to the ethical principles to the practice of medicine | PROF1: Professional Behavior and Ethical Principles |
| PROF3: Demonstrate personal responsibility to maintain emotional, physical, and mental health and accountability to patients, society, and the profession | PROF2: Accountability/Conscientiousness<br>PROF3: Self-Awareness and Help-Seeking |
| ICS1: Relationship building, teamwork, and conflict management | ICS1: Patient- and Family-Centered Communication<br>ICS2: Interprofessional and Team Communication |
| ICS2: Information gathering and sharing | ICS1: Patient- and Family-Centered Communication<br>ICS2: Interprofessional and Team Communication<br>ICS3: Communication within Health Care Systems |