Medical Genetics and Genomics Milestones

The Accreditation Council for Graduate Medical Education

ACGME

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Medical Genetics and Genomics Milestones

The Milestones are designed only for use in evaluation of residents in the context of their participation in ACGME-accredited residency or fellowship programs. The Milestones provide a framework for the assessment of the development of the resident in key dimensions of the elements of physician competency in a specialty or subspecialty. They neither represent the entirety of the dimensions of the six domains of physician competency, nor are they designed to be relevant in any other context.

Medical Genetics and Genomics Milestones

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American Board of Medical Genetics and Genomics

Association of Professors of Human and Medical Genetics

ACGME Review Committee for Medical Genetics and Genomics

Understanding Milestone Levels and Reporting

This document presents Milestones designed for programs to use in semi-annual review of resident performance and reporting to the ACGME. Milestones are knowledge, skills, attitudes, and other attributes for each of the ACGME competencies organized in a developmental framework. The narrative descriptions are targets for resident performance throughout their training.

For each reporting period, the Clinical Competency Committee will review the completed evaluations to select the milestone levels that best describe each resident's current performance, abilities and attributes for each subcompetency. Milestones are arranged into levels. Tracking from Level 1 to Level 5 is synonymous with moving from novice to expert resident in the specialty.

These levels *do not* correspond with post-graduate year of education. Depending on previous experience, a junior resident may achieve higher levels early in training just as a senior resident may be at a lower level later in training. There is no predetermined timing for a resident to attain any particular level. A resident may also regress in their milestones. This may happen for many reasons, such as over scoring in the previous meeting, disjointed experience in a particular procedure, or significant act by the resident.

Selection of a level implies that the resident substantially demonstrates the milestones in that level, as well as those in lower levels (see the diagram on page v).

Additional Notes

For residents who have insufficient data/evaluations to assess, please choose “not yet assessable.”

Level 4 is designed as a graduation *goal* and *does not* represent a graduation *requirement*. Making decisions about readiness for graduation and unsupervised practice is the purview of the residency program director. Furthermore, Milestones 2.0 include revisions and changes that preclude using Milestones as a sole assessment in high stakes decisions (i.e. determination of eligibility for certification or credentialing). Level 5 is designed to represent an expert resident whose achievements in a subcompetency are greater than the expectation. Milestones are primarily designed for formative, developmental purposes to support continuous quality improvement for individual learners, training programs, and the specialty. ACGME and its partners will continue to evaluate and perform research on the Milestones 2.0 sets to assess their impact and value.

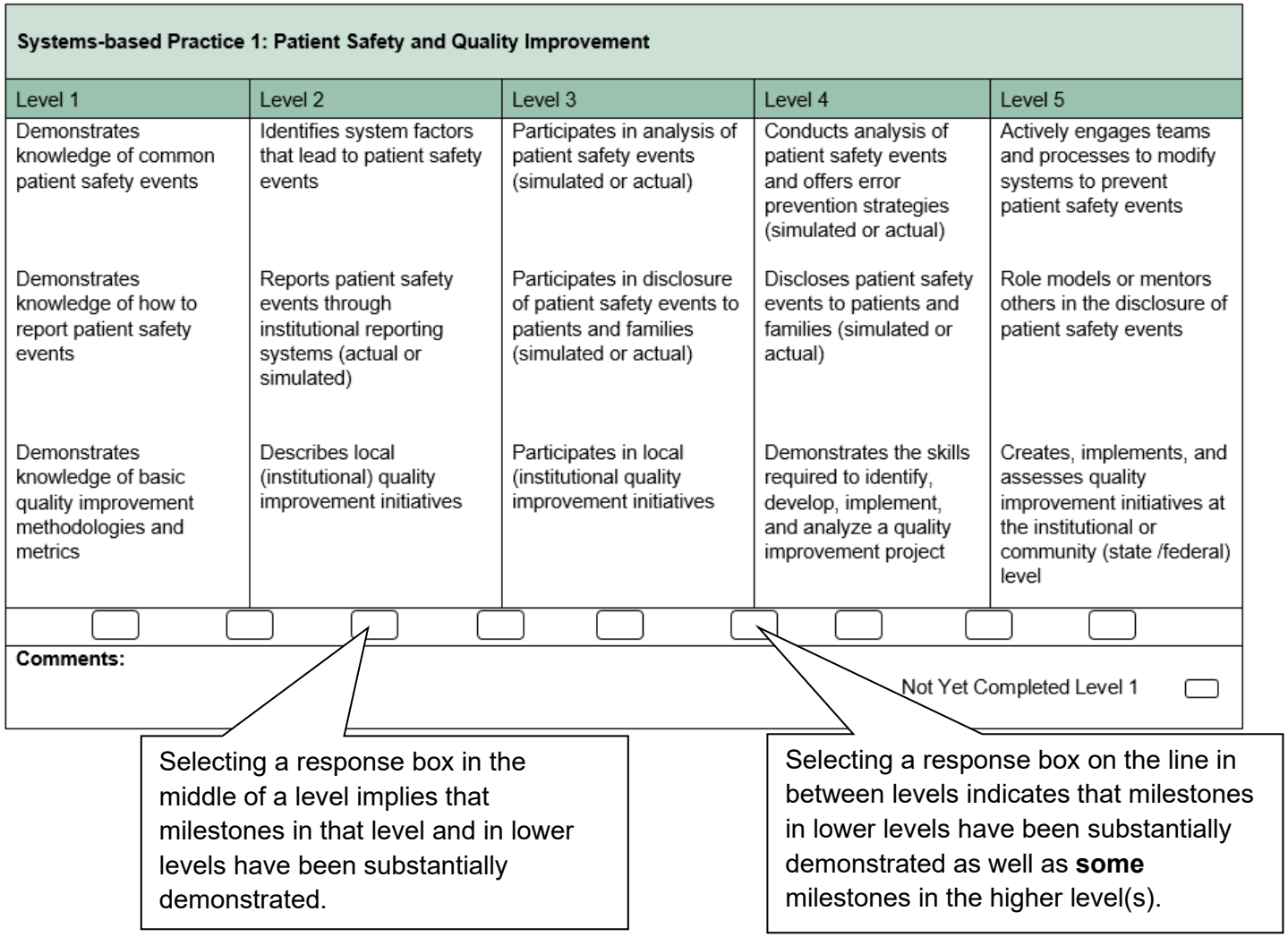
Examples are provided for some milestones within this document. Please note that the examples are not the required element or outcome; they are provided as a way to share the intent of the element.

Some milestone descriptions include statements about performing independently. These activities must occur in conformity to the ACGME supervision guidelines, as well as to institutional and program policies. For example, a resident who performs a procedure independently must, at a minimum, be supervised through oversight.

A Supplemental Guide is also available. This Guide provides the intent of each subcompetency, examples for each level, assessment methods or tools, and other resources that are available. This Guide, like examples contained within the Milestones, was designed to assist the program director and Clinical Competency Committee and is not meant to demonstrate any required element or outcome.

Supplemental Guides and other resources are available on the Milestones page of each specialty section of the ACGME website. On www.acgme.org, choose the applicable specialty under the “Specialties” menu, then select the “Milestones” link in the lower navigation bar.

The diagram below presents an example set of milestones for one sub-competency in the same format as the ACGME Report Worksheet. For each reporting period, a resident's performance on the milestones for each sub-competency will be indicated by selecting the level of milestones that best describes that resident's performance in relation to those milestones.



Systems-based Practice 1: Patient Safety and Quality Improvement				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates knowledge of common patient safety events	Identifies system factors that lead to patient safety events	Participates in analysis of patient safety events (simulated or actual)	Conducts analysis of patient safety events and offers error prevention strategies (simulated or actual)	Actively engages teams and processes to modify systems to prevent patient safety events
Demonstrates knowledge of how to report patient safety events	Reports patient safety events through institutional reporting systems (actual or simulated)	Participates in disclosure of patient safety events to patients and families (simulated or actual)	Discloses patient safety events to patients and families (simulated or actual)	Role models or mentors others in the disclosure of patient safety events
Demonstrates knowledge of basic quality improvement methodologies and metrics	Describes local (institutional) quality improvement initiatives	Participates in local (institutional quality improvement initiatives	Demonstrates the skills required to identify, develop, implement, and analyze a quality improvement project	Creates, implements, and assesses quality improvement initiatives at the institutional or community (state /federal) level
☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐				
Comments: Not Yet Completed Level 1 ☐				

Selecting a response box in the middle of a level implies that milestones in that level and in lower levels have been substantially demonstrated.

Selecting a response box on the line in between levels indicates that milestones in lower levels have been substantially demonstrated as well as **some** milestones in the higher level(s).

Patient Care 1: History and Physical Examination				
Level 1	Level 2	Level 3	Level 4	Level 5
Takes a general medical and family history	Takes a basic genetics-focused history and completes a basic pedigree	Takes a genetics-focused history with some pertinent positive and negative findings; completes an accurate pedigree	Takes a comprehensive genetic history with pertinent positive and negative findings; integrates the history with other data to develop a differential diagnosis	Makes a nationally recognized contribution by describing a new genetic disorder or expanding the phenotype of a known syndrome or disorder
Completes a general physical examination	Completes a basic genetics-focused physical examination; identifies normal and abnormal phenotypic features and/or anomalies	Completes a genetics-focused physical examination; identifies and accurately describes common phenotypic features and/or anomalies; recognizes common syndromes or disorders	Identifies and accurately describes phenotypic features and/or anomalies using standardized nomenclature; recognizes complex syndromes or disorders	
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments:				
				Not Yet Completed Level 1 ☐ Not Yet Assessable ☐

Patient Care 2: Selecting Tests, Interpreting Results, and Management of Genetic Conditions				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies the variety of testing modalities for genetic conditions	Identifies basic testing options for common genetic disorders	Identifies strengths and limitations of testing methodologies in order to select first tier tests	Selects and prioritizes testing options across a broad spectrum of complex disorders and inheritance patterns/ mechanisms	Contributes to the knowledge base for the refinement of ambiguous test results
Identifies the components of the genetics test result	Identifies resources to facilitate interpretation of positive, negative, and uncertain test results	Uses resources to interpret diagnostic test results in the context of the phenotype	Uses resources to interpret ambiguous test results in the context of the phenotype	
Recognizes the availability of intervention for some genetic conditions	Identifies resources and guidelines for treatment and management of common genetic conditions	Implements treatment and/or surveillance plans for common genetic conditions	Implements treatment and/or surveillance plans for complex genetic conditions	Creates evidence-based guidelines for management
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Comments:				
				Not Yet Completed Level 1 ☐ Not Yet Assessable ☐

Patient Care 3: Pre- and Post-Test Genetic Counseling				
Level 1	Level 2	Level 3	Level 4	Level 5
Participates in pre-test counseling	Explains the rationale for the recommended testing	Conveys the impact and limitations of disorder-specific targeted testing while obtaining informed consent	Clearly conveys the impact and limitations of complex untargeted testing while obtaining informed consent	Participates in the development of professional practice guidelines regarding testing and return of results
Participates in post-test counseling	Explains the results of the test	Conveys the impact and limitations of diagnostic and non-diagnostic results	Conveys the impact and limitations of unexpected and ambiguous results	
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Comments:				
				Not Yet Completed Level 1 ☐ Not Yet Assessable ☐

Medical Knowledge 1: Foundations of Genetics and Genomics				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates basic medical knowledge of embryology, inheritance, and genetic mechanism of disease	Applies knowledge of embryology, inheritance, and genetic mechanism of disease to identify a differential diagnosis	Applies advanced knowledge of embryology, inheritance, and genetic mechanism of disease to make a diagnosis	Applies advanced knowledge of embryology, inheritance, and genetic mechanism of disease to diagnostic and therapeutic interventions	Contributes to peer-reviewed resources addressing genetic mechanism of disease
Demonstrates basic medical knowledge of gene and genome structure and function	Applies knowledge of gene and genome structure and function to identify a differential diagnosis	Applies advanced knowledge of gene and genome structure and function to make a diagnosis	Applies advanced knowledge of gene and genome structure and function to diagnostic and therapeutic interventions	Recognized as a national expert in diagnosis and management of genetic disease
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Comments:				
				Not Yet Completed Level 1 ☐ Not Yet Assessable ☐

Medical Knowledge 2: Clinical Genetics and Genomics				
Level 1	Level 2	Level 3	Level 4	Level 5
Recognizes syndromic and non-syndromic etiologies	Identifies syndromic and non-syndromic etiologies	Demonstrates knowledge of syndromic and non-syndromic etiologies and the impact on diagnosis and management	Applies knowledge of syndromic and non-syndromic etiologies to diagnosis and management	Serves as an expert resource for syndromic and/or non-syndromic etiologies
Recognizes that phenotypes evolve across the lifespan	Identifies the changes of phenotypes across the lifespan	Demonstrates knowledge of the changes in phenotypes across the lifespan and how it impacts diagnosis and management	Applies knowledge of the changes in phenotypes across the lifespan and how it impacts diagnosis and management	Contributes to peer-reviewed resources addressing natural history of genetic disease
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Comments:				
				Not Yet Completed Level 1 ☐ Not Yet Assessable ☐

Medical Knowledge 3: Clinical Reasoning				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates a basic framework for clinical reasoning Identifies appropriate resources to inform clinical reasoning	Demonstrates clinical reasoning to determine relevant information Selects relevant resources based on scenario to inform decisions	Synthesizes information to inform clinical reasoning, with assistance Seeks and integrates evidence-based information to inform diagnostic decision making in complex cases, with assistance	Independently synthesizes information to inform clinical reasoning in complex cases Independently seeks out, analyzes and applies relevant original research to diagnostic decision making in complex clinical cases	Develops a novel approach for the assessment of complex cases
☐	☐	☐	☐	☐ ☐ ☐ ☐ ☐
Comments:				
Not Yet Completed Level 1 ☐ Not Yet Assessable ☐				

Systems-Based Practice 1: Patient Safety and Quality Improvement				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates knowledge of common patient safety events	Identifies system factors that lead to patient safety events	Participates in analysis of patient safety events (simulated or actual)	Conducts analysis of patient safety events and offers error prevention strategies (simulated or actual)	Actively engages teams and processes to modify systems to prevent patient safety events
Demonstrates knowledge of how to report patient safety events	Reports patient safety events through institutional reporting systems (simulated or actual)	Participates in disclosure of patient safety events to patients and families (simulated or actual)	Discloses patient safety events to patients and families (simulated or actual)	Role models or mentors others in the disclosure of patient safety events
Demonstrates knowledge of basic quality improvement methodologies and metrics	Describes local (institutional) quality improvement initiatives	Participates in local (institutional) quality improvement initiatives	Demonstrates the skills required to identify, develop, implement, and analyze a quality improvement project	Creates, implements, and assesses quality improvement initiatives at the institutional or community (state/federal) level
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Comments:				
				Not Yet Completed Level 1 ☐

Systems-Based Practice 2: System Navigation for Patient-Centered Care				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates knowledge of care coordination	Coordinates care of patients in routine clinical situations effectively using the roles of the interprofessional teams, including non-physician patient caregivers	Coordinates care of patients in complex clinical situations effectively using the roles of the interprofessional teams	Role models effective coordination of patient-centered care among different disciplines and specialties including referrals and testing	Analyzes the process of care coordination and leads in the design and implementation of improvements
Identifies key elements for safe and effective transitions of care and hand-offs	Performs safe and effective transitions of care/hand-offs in routine clinical situations	Performs safe and effective transitions of care/hand-offs in complex clinical situations	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	Improves quality of transitions of care within and across health care delivery systems to optimize patient outcomes
Demonstrates knowledge of population and community health needs	Identifies specific population and community health needs for the local population	Uses local resources effectively to meet the needs of a patient population and community	Participates in changing and adapting practice to provide for the needs of specific populations including advocating for a patient's genetic testing coverage	Leads innovations and advocates for populations and communities with specific health care needs at the state or federal level
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Comments:				
				Not Yet Completed Level 1 ☐

Systems-Based Practice 3: Physician Role in Health Care Systems				
Level 1	Level 2	Level 3	Level 4	Level 5
Identifies key components of the complex health care system (e.g., hospital, skilled nursing facility, finance, personnel, technology)	Describes how components of a complex health care system are interrelated, and how this impacts patient care	Discusses how individual practice affects the broader system (e.g., access to genetic testing and treatments, testing advocacy)	Manages various components of the complex health care system to provide efficient and effective patient care and transition of care	Advocates for or leads systems change that enhances high-value, efficient, and effective patient care and transition of care
Describes basic health payment systems (e.g., government, private, public, uninsured care) and practice models	Delivers care with consideration of each patient's payment model (e.g., insurance type) and access to genetic testing or formula	Engages with patients in shared decision making, often informed by each patient's payment models	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	Participates in health policy advocacy activities
Identifies basic knowledge for effective transition to practice (e.g., information technology, legal, billing and coding, financial, personnel)	Demonstrates use of information technology required for medical practice (e.g., electronic health record, documentation required for billing and coding)	Describes core administrative knowledge needed for transition to practice (e.g., contract negotiations, malpractice insurance, government regulation, compliance)	Analyzes individual practice patterns and professional requirements in preparation for practice	Educates others to prepare them for transition to practice
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Comments:

Not Yet Completed Level 1 ☐

Practice-Based Learning and Improvement 1: Evidence-Based and Informed Practice				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates how to access and use available evidence, and incorporate patient preferences and values in order to take care of a routine patient	Articulates clinical questions and elicits patient preferences and values in order to guide evidence-based care	Locates and applies the best available evidence, integrated with patient preference, to the care of complex patients	Critically appraises and applies evidence even in the face of uncertainty and conflicting evidence to guide care, tailored to the individual patient	Mentors others to critically appraise and apply evidence for complex patients; and/or participates in the development of guidelines
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Comments:				
Not Yet Completed Level 1 ☐				

Practice-Based Learning and Improvement 2: Reflective Practice and Commitment to Personal Growth				
Level 1	Level 2	Level 3	Level 4	Level 5
Accepts responsibility for personal and professional development by establishing goals	Demonstrates openness to performance data (feedback and other input) in order to inform goals	Seeks performance data episodically, with adaptability and humility	Seeks performance data consistently with adaptability and humility	Serves as a role model in seeking performance data with adaptability and humility
Identifies the factors which contribute to gap(s) between expectations and actual performance	Analyzes and reflects on the factors which contribute to gap(s) between expectations and actual performance	Analyzes, reflects on, and institutes behavioral change(s) to narrow the gap(s) between expectations and actual performance	Challenges assumptions and considers alternatives in narrowing the gap(s) between expectations and actual performance	Mentors others on reflective practice
Actively seeks opportunities to improve	Designs and implements a learning plan, with prompting	Independently creates and implements a learning plan	Uses performance data to measure the effectiveness of the learning plan and when necessary, improves it	Facilitates the design and implementing learning plans for others
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Comments:				
Not Yet Completed Level 1 ☐				

Professionalism 1: Professional Behavior and Ethical Principles				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates compassion, sensitivity, honesty and integrity, and identifies potential triggers for professionalism lapses	Demonstrates compassion, sensitivity, honesty and integrity, and takes responsibility for own professionalism lapses	Demonstrates compassion, sensitivity, honesty, and integrity in complex/stressful situations	Demonstrates compassion, sensitivity, honesty, and integrity and serves as a role model to others	Coaches others when their behavior fails to meet professional expectations
Demonstrates knowledge of the ethical principles underlying patient care	Analyzes straightforward situations using ethical principles	Recognizes need to seek help in managing and resolving complex ethical situations	Recognizes and uses appropriate resources for managing and resolving ethical dilemmas as needed	Identifies and seeks to address system-level factors that induce or exacerbate ethical problems or impede their resolution
Demonstrates basic knowledge of conflict of interest	Identifies different types of conflicts of interest, knows guidelines for interactions with vendors	Identifies resources for managing and resolving conflicts of interest	Demonstrates consistently professional behavior with regard to conflicts of interest relevant to presentations, publishing, consulting, and service	
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Comments:				
				Not Yet Completed Level 1 ☐

Professionalism 2: Accountability/Conscientiousness				
Level 1	Level 2	Level 3	Level 4	Level 5
Takes responsibility for failure to complete tasks and responsibilities, identifies potential contributing factors, and describes strategies for ensuring timely task completion in the future	Performs tasks and responsibilities in a timely manner with appropriate attention to detail in routine situations	Performs tasks and responsibilities in a timely manner with appropriate attention to detail in complex or stressful situations	Recognizes and addresses situations that may impact others' ability to complete tasks and responsibilities in a timely manner	Volunteers to improve and takes ownership of system outcomes
Responds promptly to requests or reminders to complete tasks and responsibilities	Recognizes situations that may impact his/her own ability to complete tasks and responsibilities in a timely manner	Proactively implements strategies to ensure that the needs of patients, teams, and systems are met		
Recognizes the role of appearance, daily demeanor and conduct in the role of a professional	Demonstrates a professional appearance, daily demeanor, and conduct	Sets a standard for appearance, daily demeanor, and conduct as a professional	Promotes professional appearance, demeanor, and conduct in their peers and associates	
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Comments:				
				Not Yet Completed Level 1 ☐

Professionalism 3: Self-Awareness and Help-Seeking				
Level 1	Level 2	Level 3	Level 4	Level 5
Recognizes status of personal and professional well-being, with assistance	Independently recognizes status of personal and professional well-being	With assistance, proposes a plan to optimize personal and professional well-being	Independently develops a plan to optimize personal and professional well-being	Coaches others when emotional responses or limitations in knowledge/skills do not meet professional expectations
Recognizes limits in the knowledge/skills of self or team, with assistance	Independently recognizes limits in the knowledge/ skills of self or team and demonstrates appropriate help-seeking behaviors	With assistance, proposes a plan to remediate or improve limits in the knowledge/ skills of self or team	Independently develops a plan to remediate or improve limits in the knowledge/skills of self or team	
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Comments:				
Not Yet Completed Level 1 ☐				

This subcompetency is not intended to evaluate a resident's well-being. Rather, the intent is to ensure that each resident has the fundamental knowledge of factors that affect well-being, the mechanisms by which those factors affect well-being, and available resources and tools to improve well-being.

Interpersonal and Communication Skills 1: Patient and Family-Centered Communication				
Level 1	Level 2	Level 3	Level 4	Level 5
Uses language and nonverbal behavior to demonstrate respect and establish rapport	Establishes a therapeutic relationship in straightforward encounters using active listening and clear language	Establishes a therapeutic relationship in challenging patient encounters	Establishes therapeutic relationships, with attention to patient/family concerns and context, regardless of complexity	Mentors others in situational awareness and critical self-reflection to consistently develop positive therapeutic relationships
Identifies common barriers to effective communication while accurately communicating own role within the health care system	Identifies complex barriers to effective communication	When prompted, reflects on biases while attempting to minimize communication barriers	Recognizes biases while attempting to proactively minimize communication barriers	Role models self-awareness practice while identifying teaching a contextual approach to minimize communication barriers
Identifies the need to adjust communication strategies based on assessment of patient/family expectations and understanding of their health status and treatment options	Organizes and initiates communication with patient/family by introducing stakeholders, setting the agenda, clarifying expectations, and verifying understanding of the clinical situation	With guidance, sensitively and compassionately delivers medical information, elicits patient/family values, goals and preferences, and acknowledges uncertainty and conflict	Uses shared decision making to align patient/family values, goals, and preferences with treatment options to make a personalized care plan	Role models shared decision making in patient/family communication including those with a high degree of uncertainty/conflict
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Comments:				Not Yet Completed Level 1 ☐

Interpersonal and Communication Skills 2: Interprofessional and Team Communication				
Level 1	Level 2	Level 3	Level 4	Level 5
Respectfully requests a consultation	Clearly and concisely requests a consultation	Checks own understanding of consultant recommendations	Coordinates recommendations from different members of the health care team to optimize patient care	Role models flexible communication strategies that value input from all health care team members, resolving conflict when needed
Respectfully receives a consultation request	Clearly and concisely responds to a consultation request	Checks requestor's understanding of recommendations when providing consultation	Provides information to the primary care team regarding rationale for recommendations	
Uses language that values all members of the health care team	Communicates information effectively with all health care team members	Uses active listening to adapt communication style to fit team needs	Models active listening to other health care team members	
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Comments:				
				Not Yet Completed Level 1 ☐

Interpersonal and Communication Skills 3: Communication within Health Care Systems				
Level 1	Level 2	Level 3	Level 4	Level 5
Accurately records information in the patient record	Demonstrates organized diagnostic and therapeutic reasoning through notes in the patient record	Concisely reports diagnostic and therapeutic reasoning in the patient record	Communicates clearly, concisely, timely, and in an organized written form, including anticipatory guidance	Models feedback to improve others' written communication
Safeguards patient personal health information	Uses documentation shortcuts accurately, appropriately and in a timely manner Documents required data in formats specified by institutional policy	Appropriately selects direct (e.g., telephone, in-person) and indirect (e.g., progress notes, text messages) forms of communication based on context	Achieves written or verbal communication (e.g., patient notes, e-mail) that serves as an example for others to follow	Guides departmental or institutional communication around policies and procedures
☐	☐	☐	☐	☐

Comments:

Not Yet Completed Level 1 ☐