

The Medical Biochemical Genetics Milestone Project

A Joint Initiative of

The Accreditation Council for Graduate Medical Education

and

The American Board of Medical Genetics and Genomics



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The Milestones are designed only for use in evaluation of fellows 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 fellow 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 Biochemical Genetics Milestones

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Milestone Reporting

This document presents Milestones designed for programs to use in semi-annual review of fellow 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 from less to more advanced. They are descriptors and targets for fellow performance as a fellow moves from entry into fellowship through graduation. In the initial years of implementation, the Review Committee will examine Milestone performance data for each program's fellows as one element in the Next Accreditation System (NAS) to determine whether fellows overall are progressing.

For each period, review and reporting will involve selecting milestone levels that best describe each fellow's current performance and attributes. Milestones are arranged into numbered levels. Tracking from Level 1 to Level 5 is synonymous with moving from novice to expert in the subspecialty. These levels do not correspond with time in the educational program. Dependent upon previous education and experience, fellows may enter a program at varying points in the milestones.

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

Level 1: The fellow demonstrates milestones expected of an incoming fellow.

Level 2: The fellow is advancing and demonstrates additional milestones, but is not yet performing at a mid-fellowship level.

Level 3: The fellow continues to advance and demonstrate additional milestones, consistently including the majority of milestones targeted for fellowship.

Level 4: The fellow has advanced so that he or she now substantially demonstrates the milestones targeted for fellowship. This level is designed as the graduation target.

Level 5: The fellow has advanced beyond performance targets set for fellowship and is demonstrating "aspirational" goals which might describe the performance of someone who has been in practice for several years. It is expected that only a few exceptional fellows will reach this level.

Additional Notes

Level 4 is designed as the graduation *target* and *does not* represent a graduation *requirement*. Making decisions about readiness for graduation is the purview of the fellowship program director. Study of Milestone performance data will be required before the ACGME and its partners will be able to determine whether milestones in the first four levels appropriately represent the developmental framework, and whether Milestone data are of sufficient quality to be used for high-stakes decisions.

Examples are provided with some milestones. 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 fellow who performs a procedure independently must, at a minimum, be supervised through oversight.

Answers to Frequently Asked Questions about the Next Accreditation System and Milestones are posted on the Next Accreditation System section of the ACGME website.

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 fellow’s performance on the milestones for each sub-competency will be indicated by selecting the level of milestones that best describes that fellow’s performance in relation to those milestones.

Initial or Acute Management — Patient Care				
Level 1	Level 2	Level 3	Level 4	Level 5
With substantial guidance recognizes signs and symptoms of inborn errors of metabolism	With moderate guidance recognizes signs and symptoms of inborn errors of metabolism	With minimal guidance recognizes signs and symptoms of inborn errors of metabolism	Independently recognizes signs and symptoms of inborn errors of metabolism	Develops or contributes to acute management protocols or novel therapies
With substantial guidance recognizes signs and symptoms of acute metabolic decompensation	With moderate guidance recognizes signs and symptoms of acute metabolic decompensation	With minimal guidance recognizes signs and symptoms of acute metabolic decompensation	Independently recognizes signs and symptoms of acute metabolic decompensation	
With substantial guidance selects and orders diagnostic studies	With moderate guidance selects and orders diagnostic studies	With minimal guidance selects and orders diagnostic studies	Independently selects and orders diagnostic studies	
With substantial guidance provides supportive and disease-specific management	With moderate guidance provides supportive and disease-specific management	With minimal guidance provides supportive and disease-specific management	Independently provides supportive and disease-specific management	
<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments: Not yet achieved Level 1 <input type="checkbox"/>				

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).

Initial or Acute Management — Patient Care 1				
Level 1	Level 2	Level 3	Level 4	Level 5
With substantial guidance, recognizes signs and symptoms of inborn errors of metabolism	With moderate guidance, recognizes signs and symptoms of inborn errors of metabolism	With minimal guidance, recognizes signs and symptoms of inborn errors of metabolism	Independently recognizes signs and symptoms of inborn errors of metabolism	Develops or contributes to acute management protocols or novel therapies
With substantial guidance, recognizes signs and symptoms of acute metabolic decompensation	With moderate guidance, recognizes signs and symptoms of acute metabolic decompensation	With minimal guidance, recognizes signs and symptoms of acute metabolic decompensation	Independently recognizes signs and symptoms of acute metabolic decompensation	
With substantial guidance, selects and orders diagnostic studies	With moderate guidance, selects and orders diagnostic studies	With minimal guidance, selects and orders diagnostic studies	Independently selects and orders diagnostic studies	
With substantial guidance, provides supportive and disease-specific management	With moderate guidance, provides supportive and disease-specific management	With minimal guidance, provides supportive and disease-specific management	Independently provides supportive and disease-specific management	
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Chronic Management — Patient Care 2				
Level 1	Level 2	Level 3	Level 4	Level 5
With substantial guidance, provides disease-specific dietary, supportive, and medical management	With moderate guidance, provides disease-specific dietary, supportive, and medical management	With minimal guidance, provides disease-specific dietary, supportive, and medical management	Independently provides disease-specific dietary, supportive, and medical management	Develops or contributes to chronic management protocols or novel therapies
With substantial guidance, offers and provides disease modifying therapy (e.g., solid organ transplant, enzyme replacement therapy, cell-based therapy)	With moderate guidance, offers and provides disease modifying therapy (e.g., solid organ transplant, enzyme replacement therapy, cell-based therapy)	With minimal guidance, offers and provides disease modifying therapy (e.g., solid organ transplant, enzyme replacement therapy, cell-based therapy)	Independently offers and provides disease modifying therapy (e.g., solid organ transplant, enzyme replacement therapy, cell-based therapy)	
With substantial guidance, screens and provides referral for neurodevelopmental disorders	With moderate guidance, screens and provides referral for neurodevelopmental disorders	With minimal guidance, screens and provides referral for neurodevelopmental disorders	Independently screens and provides referral for neurodevelopmental disorders	
With substantial guidance, orders disease-specific monitoring evaluations and referrals	With moderate guidance, orders disease-specific monitoring evaluations and referrals	With minimal guidance, orders disease-specific monitoring evaluations and referrals	Independently orders disease-specific monitoring evaluations and referrals	
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Newborn Screening (NBS) — Patient Care 3				
Level 1	Level 2	Level 3	Level 4	Level 5
With substantial guidance, evaluates infants with abnormal NBS results	With moderate guidance, evaluates infants with abnormal NBS results	With minimal guidance, evaluates infants with abnormal NBS results	Independently evaluates infants with abnormal NBS results	Participates in state, regional, or national NBS program development or evaluation projects
With substantial guidance, selects and orders confirmatory laboratory studies	With moderate guidance, selects and orders confirmatory laboratory studies	With minimal guidance, selects and orders confirmatory laboratory studies	Independently selects and orders confirmatory laboratory studies	
With substantial guidance, provides timely management for infants with abnormal NBS results	With moderate guidance, provides timely management for infants with abnormal NBS results	With minimal guidance, provides timely management for infants with abnormal NBS results	Independently provides timely management for infants with abnormal NBS results	
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Molecular and Metabolic Mechanisms — Medical Knowledge 1				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>With substantial guidance, applies knowledge of molecular and metabolic pathogenesis of disease to diagnosis and treatment, including these disease groups:</p> <ul style="list-style-type: none"> • amino-acidopathies • disorders of carbohydrate metabolism • fatty acid oxidation disorders • lysosomal storage diseases • mitochondrial disorders • organic acidurias • urea cycle disorders 	<p>With moderate guidance, applies knowledge of molecular and metabolic pathogenesis of disease to diagnosis and treatment, including these disease groups:</p> <ul style="list-style-type: none"> • amino-acidopathies • disorders of carbohydrate metabolism • fatty acid oxidation disorders • lysosomal storage diseases • mitochondrial disorders • organic acidurias urea cycle disorders 	<p>With minimal guidance, applies knowledge of molecular and metabolic pathogenesis of disease to diagnosis and treatment, including these disease groups:</p> <ul style="list-style-type: none"> • amino-acidopathies • disorders of carbohydrate metabolism • fatty acid oxidation disorders • lysosomal storage diseases • mitochondrial disorders • organic acidurias urea cycle disorders 	<p>Independently applies knowledge of molecular and metabolic pathogenesis of disease to diagnosis and treatment, including these disease groups:</p> <ul style="list-style-type: none"> • amino-acidopathies • disorders of carbohydrate metabolism • fatty acid oxidation disorders • lysosomal storage diseases • mitochondrial disorders • organic acidurias urea cycle disorders 	<p>Expands understanding and publishes results of molecular and metabolic basis of pathogenicity or treatment of a disease or metabolic pathway</p>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<p>Comments:</p>			<p>Not yet achieved Level 1 <input type="checkbox"/></p>	

Newborn Screening (NBS)— Medical Knowledge 2				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates basic knowledge of NBS With substantial guidance, generates a differential diagnosis based on NBS results	Demonstrates knowledge of principles and processes of NBS Demonstrates knowledge of variations in NBS programs With moderate guidance, generates a differential diagnosis based on NBS results	Demonstrates knowledge of positive predictive values of NBS tests With minimal guidance, generates a differential diagnosis based on NBS results Demonstrates knowledge of triage for individuals with abnormal NBS results	Utilizes positive predictive values in clinical decision making Demonstrates knowledge of causes of false positive and false negative results Independently generates a differential diagnosis based on NBS results	Contributes to generalizable medical knowledge of NBS
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Diagnostic Testing — Medical Knowledge 3				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>Understands the technology and utilization of diagnostic testing including these disease groups:</p> <ul style="list-style-type: none"> • amino-acidopathies • disorders of carbohydrate metabolism • fatty acid oxidation disorders • lysosomal storage diseases • mitochondrial disorders • organic acidurias • urea cycle disorders 	<p>With substantial guidance, identifies best methods for diagnosis and subsequent laboratory monitoring in these disease groups:</p> <ul style="list-style-type: none"> • amino-acidopathies • disorders of carbohydrate metabolism • fatty acid oxidation disorders • lysosomal storage diseases • mitochondrial disorders • organic acidurias urea cycle disorders <p>With substantial guidance, interprets and applies the information obtained from testing</p>	<p>With minimal guidance, identifies best methods for diagnosis and subsequent laboratory monitoring in these disease groups:</p> <ul style="list-style-type: none"> • amino-acidopathies • disorders of carbohydrate metabolism • fatty acid oxidation disorders • lysosomal storage diseases • mitochondrial disorders • organic acidurias urea cycle disorders <p>With minimal guidance, interprets and applies the information obtained from testing</p>	<p>Independently identifies best methods for diagnosis and subsequent laboratory monitoring in these disease groups:</p> <ul style="list-style-type: none"> • amino-acidopathies • disorders of carbohydrate metabolism • fatty acid oxidation disorders • lysosomal storage diseases • mitochondrial disorders • organic acidurias urea cycle disorders <p>Independently interprets and applies the information obtained from testing</p>	<p>Develops polices or practice guidelines for testing</p> <p>Participates in assay development</p>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Systems Approach — Systems-based Practice 1				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>Recognizes how health care systems influence individual practice and patient care</p> <p>Sensitive to cost-effective care</p>	<p>With substantial guidance, functions effectively within different systems</p> <p>With substantial guidance, advocates for genetics services to enhance cost-effective care</p> <p>With substantial guidance, recognizes and manages the variation in access to testing and therapies for inborn errors of metabolism</p> <p>With substantial guidance, participates in identifying system errors and implementing potential system solutions (e.g., emergency protocol letter)</p>	<p>With minimal guidance, functions effectively within different systems</p> <p>With minimal guidance, advocates for genetics services to enhance cost-effective care</p> <p>With minimal guidance, recognizes and manages the variation in access to testing and therapies for inborn errors of metabolism</p> <p>With minimal guidance, participates in identifying and anticipating system errors and implementing potential system solutions</p>	<p>Independently functions effectively within different systems</p> <p>Independently advocates for genetics services to enhance cost-effective care</p> <p>Independently recognizes and manages the variation in access to genetic testing and therapies for inborn errors of metabolism</p> <p>Independently identifies and anticipates system errors and implements potential system solutions</p>	<p>Leads a quality improvement project to improve quality of care or access to resources</p>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Newborn Screening Systems — Systems-based Practice 2				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>With substantial guidance, collaborates with the primary provider, birth hospital, nurses, dieticians, and other members of the health care team to act upon results in a timely manner</p> <p>With substantial guidance, collaborates with the NBS program and confirmatory laboratory</p>	<p>With moderate guidance, collaborates with the primary provider, birth hospital, nurses, dieticians, and other members of the health care team to act upon results in a timely manner</p> <p>With moderate guidance, collaborates with the NBS program and confirmatory laboratory</p>	<p>With minimal guidance, collaborates with the primary provider, birth hospital, nurses, dieticians, and other members of the health care team to act upon results in a timely manner</p> <p>With minimal guidance, collaborates with the NBS program and confirmatory laboratory</p>	<p>Independently collaborates with the primary provider, birth hospital, nurses, dieticians, and other members of the health care team to act upon results in a timely manner</p> <p>Independently collaborates with the NBS program and confirmatory laboratory</p>	<p>Develops a program to improve collaboration and/or care between health care providers and systems</p> <p>Participates in NBS registries and/or evaluates the data</p>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Transitions of Care— Systems-based Practice 3				
Level 1	Level 2	Level 3	Level 4	Level 5
Recognizes that diagnosis and management have implications for care at the hospital, in the home, and in the community across the lifespan of the patient	With substantial guidance, facilitates management and transitions of care teams at the hospital, in the home, and in the community across the lifespan of the patient	With minimal guidance, facilitates management and transitions of care teams at the hospital, in the home, and in the community across the lifespan of the patient	Independently facilitates management and transitions of care teams at the hospital, in the home, and in the community across the lifespan of the patient	Participates in the development of a system or resources to improve transitions in care
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Self-Directed Learning — Practice-based Learning and Improvement 1				
Level 1	Level 2	Level 3	Level 4	Level 5
Acknowledges gaps in personal knowledge and expertise	With substantial guidance, assesses self-performance and develops a learning plan	With minimal guidance, assesses self-performance and develops a learning plan	Independently assesses self-performance and develops a learning plan	Develops nationally-recognized materials and approaches for education of others
Demonstrates computer literacy and basic computer skills	With substantial guidance, critically evaluates and utilizes information from diverse sources (e.g., professional practice guidelines)	With minimal guidance, critically evaluates and utilizes information from diverse sources	Independently evaluates critically and utilizes information from diverse sources	
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Process Improvement and Patient Safety — Practice-based Learning and Improvement 2				
Level 1	Level 2	Level 3	Level 4	Level 5
Demonstrates awareness of common sources of error in patient care and laboratory testing	Consistently demonstrates work habits that minimize error; consistently and promptly communicates discrepancies to health care team	Initiates practice change based on an identified error or systematic problem	Able to develop and lead a process improvement exercise (e.g., root-cause analysis, work-flow analysis)	Participates in an institutional quality improvement team
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Giving and Receiving Feedback — Professionalism 1				
Level 1	Level 2	Level 3	Level 4	Level 5
Receives feedback constructively	Accepts feedback constructively and modifies practice in response to feedback	Provides constructive feedback	Exemplifies giving and receiving of constructive feedback; encourages and actively seeks feedback to improve performance	Models giving and receiving of constructive feedback; encourages and actively seeks feedback to improve performance
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Cultural Competency — Professionalism 2				
Level 1	Level 2	Level 3	Level 4	Level 5
Understands the importance of caring, honesty, and sensitive behavior with diverse patient populations	Demonstrates caring, honesty, and sensitive behavior in common and uncomplicated situations	Demonstrates caring, honesty, and sensitive behavior in complex situations	Demonstrates caring, honesty, and sensitive behavior in all relationships and situations	Demonstrates leadership and mentoring regarding these principles
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Accountability and Integrity — Professionalism 3				
Level 1	Level 2	Level 3	Level 4	Level 5
Completes assigned tasks on time	Dependably completes assigned tasks in a timely manner; assists team members when requested; respects assigned schedules Acknowledges personal near-misses and errors, and puts the needs of patients first Engages in ethical behavior	Anticipates team needs and steps in to assist as needed Demonstrates accountability with all members of the health care team Identifies, communicates, and corrects errors	Anticipates team needs and takes leadership role to independently implement solutions Is viewed by members of the health care team as a role model in accepting personal responsibility, and in always putting the needs of the patient above his or her own interests	Exemplifies effective management of multiple competing tasks, with reliable follow-up Is a source of support/guidance to other members of health care team
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>

Communicates with Patients and Families — Interpersonal and Communication Skills 1				
Level 1	Level 2	Level 3	Level 4	Level 5
<p>With substantial guidance, provides timely and effective communication with patients and families</p> <p>Develops a positive relationship based on mutual trust with patients and families in uncomplicated situations</p>	<p>With moderate guidance, provides timely and effective communication to patients and families, including education and genetic counseling</p> <p>With moderate guidance, develops and sustains relationships with patients and families</p> <p>With moderate guidance, manages conflicts with patients and families</p>	<p>With minimal guidance, provides timely and effective communication to patients and families, including education and genetic counseling</p> <p>With minimal guidance, develops and sustains relationships with patients and families</p> <p>With minimal guidance, manages conflicts with patients and families</p>	<p>Effectively communicates complex, difficult, or challenging information (e.g., errors, complications, adverse events, bad news)</p> <p>Independently provides comprehensive, effective patient and family education and genetic counseling</p> <p>Independently develops and sustains relationships with patients and families</p> <p>Independently manages conflicts with patients and families</p>	<p>Serves as a role model for effective, compassionate, and professional communication to patients and families</p>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<p>Comments:</p>				<p>Not yet achieved Level 1 <input type="checkbox"/></p>

Communication and Conflict Management within the Health Care Team — Interpersonal and Communication Skills 2				
Level 1	Level 2	Level 3	Level 4	Level 5
With substantial guidance, provides timely and effective communication to the health care team	With moderate guidance, provides timely and effective communication to the health care team	With minimal guidance, provides timely and effective communication to the health care team	Independently provides timely and effective communication to the health care team	Serves as a role model for effective and professional communication to the health care team
Recognizes conflicts within the health care team	With moderate guidance, participates in team-based care	With minimal guidance, participates in team-based care	Independently anticipates and manages conflicts within the health care team	Develops models/approaches to managing difficult communications
Effectively utilizes the electronic health record	With moderate guidance, manages conflicts within the health care team	With minimal guidance, manages conflicts within the health care team	Independently participates in team-based care	
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comments:				Not yet achieved Level 1 <input type="checkbox"/>