

American Board of Medical Genetics and Genomics
 Clinical Biochemical Genetics Competencies
 December 2019

Introduction: These learning guides have been developed by the ABMGG to assist training program directors and trainees as they design, implement, monitor and evaluate the educational content of their ABMGG accredited training programs. The format of these learning guides reflects the common areas of knowledge and training that have been developed by the medical profession across the training spectra and are often referred to as the “Six Competencies.” The ABMGG has taken these areas of knowledge and experience and translated them into more specific content areas for ABMGG-accredited programs.

These learning guides are not presumed to be inclusive or exclusive. Thus, you will find that they mirror many other guiding principle documents from within the genetics community. Similarly, while they attempt to cover as many specific areas of training as possible, they cannot be viewed as the only areas of knowledge and expertise that are required to become a successful medical genetics professional. They are, as indicated, learning guides and are not rules or testing outlines. These guides are offered to the medical genetics educational community as one source of information concerning knowledge areas that may be useful in developing and evaluating the educational content of training programs.

DOMAIN	OBJECTIVES	SKILLS
1. Patient Care		
Preanalytic laboratory skills	Identify appropriate specimens for study and methods for collection, preservation and transport	Select appropriate containers, anticoagulants, and collection media for validated specimen types. Identify factors important for the transport of specimens including overnight delivery. Be aware of requirements for transporting/shipping specimens offsite using packaging which meets OSHA safety guidelines. Be aware of appropriate specimen handling requirements.
	Assess acceptability of specimen for study	Check for appropriate labeling of specimen and requisition. Evaluate suitability of specimen for requested study, both for type and amount obtained. Judge quality of specimen. Assess for presence of interfering substances. Describe methods for possible recovery of poor samples. Notify appropriate individuals of any unsatisfactory samples and document such notification.
	Accession specimen	Demonstrate knowledge in assigning laboratory accession number in database or Laboratory Information System (LIS). Be familiar with general accessioning procedures and required data fields. Be aware of specific data fields that may be required for a particular test (e.g., ethnicity). Be aware of special accessioning requirements for samples requiring transport to an outside laboratory.
	Track specimen	Follow protocols to ensure proper identification and location of patient materials through the complete process, from accession to final report. Be able to track specimen location through all aspects of the testing process.
	Appropriate documentation	Maintain necessary records and laboratory database, in logbooks or computers, as appropriate.
Preparation of blood, cell, urine and CSF samples for analysis	Understand principles of blood cell separation, tissue culture and harvesting	Demonstrate knowledge and skill in sample preparative procedures and laboratory practices that prevent cross-contamination between samples.

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Principles of chromatographic separation (liquid chromatography/gas chromatography)	Understand principles of chromatographic separation	Demonstrate knowledge in applying principles of liquid chromatography (selection of a column/mobile phase) to most common biochemical genetics analyses (amino acids, acylcarnitines, etc.) Demonstrate knowledge in applying principles of gas chromatography to organic acids analysis
Amino acid analysis	Understand principles and techniques of sample preparation, analysis and results reporting for quantitative amino acid analysis.	Perform amino acid analysis. Evaluate amino acid chromatograms or equivalent (including assessment of baseline, peak identification and co-eluting compounds). Troubleshoot common problems of amino acid analysis. Report results with proper units, control range, and clinically relevant interpretation.
Organic acid analysis	Understand principles and techniques of sample preparation, analysis and results reporting for organic acids by gas chromatography/mass spectrometry	Perform organic acid analysis. Evaluate organic acid chromatograms or equivalent (including peak identification, use of extracted ion chromatograms and peak subtraction). Troubleshoot common problems of organic acid analysis. Report results with a clinically relevant interpretation. Demonstrate knowledge in methods for organic acid quantitation and reporting of quantitative results.
Acylcarnitine analysis	Understand principles and techniques of sample preparation, analysis and results reporting for quantitative acylcarnitines analysis.	Perform acylcarnitine analysis. Evaluate acylcarnitine chromatograms or equivalent (including assessment of baseline, peak identification and co-eluting compounds). Troubleshoot common problems of acylcarnitine analysis. Report results with proper units, control range, and clinically relevant interpretation
Analysis by tandem mass spectrometry (MS/MS)	Understand principles and techniques of sample preparation, analysis and results reporting for MS/MS-based tests	Demonstrate knowledge in applications, limitations and interpretations of MS/MS-based results.
Enzyme assays	Understand principles and techniques of sample and substrate preparation, analysis and results reporting for enzyme-based testing	Demonstrate knowledge in assay conditions, use of positive and negative controls, limitations and data interpretation related to determination of affected and carrier status.
Single-analyte analysis	Understand the appropriate method and sample type for each analysis	Be familiar with assay conditions, use of appropriate controls, method limitations and data interpretation related to the evaluation of specific analytes and related disease states.
Other separation techniques: thin-layer chromatography, electrophoresis, etc.	Understand principles and techniques of sample preparation and analysis	Be familiar with use of appropriate matrix and conditions for analyzing the metabolite of interest (i.e. mucopolysaccharides), appropriate controls, method limitations and data interpretation related to disease states.

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Newborn screening	Understand principles and techniques of population-based screening, including composition of newborn screening panels, sample preparation, analysis and results reporting	Demonstrate knowledge in State protocols for sample collection. Know testing methods, limitations, results and interpretations of screening procedures. Know procedures for communicating abnormal results and coordinating follow up testing.
Results interpretation and reporting	Know how to interpret results	Recognize clinically significant metabolite patterns. Determine the clinical significance of enzyme results (affected or carrier status as appropriate). Integrate results from other studies and/or clinical findings. Make recommendations for additional testing. Assess the need to report results to appropriate authority.
	Know how to report results	Draft neat, accurate written reports summarizing the findings and interpretation. Include all relevant patient information and clinical and laboratory data in the report. Communicate results clearly to all levels of healthcare providers. Document conversations when giving oral results.
General laboratory skills, quality control, and quality assurance	Know how to prepare and store reagents	Prepare reagents at the proper concentration and pH, using required grades of water and chemicals. Follow procedures for proper labeling and storing of reagents and chemicals. Be familiar with procedures for maintaining current Material Safety Data Sheets for all chemicals used in the laboratory procedures.
	Select, operate, clean, and maintain all laboratory equipment and instruments, as appropriate	Be aware of regulatory requirements for preventative maintenance of equipment and documentation of equipment repairs. Perform temperature monitoring as required. Be familiar with procedures for performing and documenting regular instrument function checks.
	Practice established procedures for laboratory safety	Use Universal Precautions as established by the Centers for Disease Control (CDC) and individual state or local governments. Use appropriate procedures for laboratory emergencies (e.g., fire, accident/injury, natural disaster, chemical spill, or power failure). Use correct procedures for storage, handling, and disposal of hazardous materials and waste: biological and chemical, volatile or stable, radioactive, sharps and glass.
	Understand laboratory quality control, quality assurance and quality management in all areas and comply with all regulatory requirements	Demonstrate knowledge in the use of quality control samples. Know how to evaluate quality control data. Follow and document proper corrective action procedures when necessary. Follow procedures for quality assurance and quality management. Know how to validate new tests according to CLIA standards. Demonstrate knowledge in policies and procedures for all laboratory tests and administrative activities. Follow required procedures for ensuring confidentiality and security of patient records. Assist in reviewing and revising laboratory policies and procedures. Participate in laboratory proficiency testing, as appropriate.

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Bioinformatics	Software	<p>Use and understand software packages for clinical lab processing, data analysis and storage, and for report writing:</p> <ul style="list-style-type: none"> • Understand implications of using electronic record keeping with respect to private health information (PHI). • Understand the informatics processes that connect sample requisition to wet lab processes, data analysis, report writing, and transmission of final reports to referring physicians.
2. Genetics Knowledge [also refer to Content Outline]		
General principles of biology and genetics	Understand principles of general biology and genetics as it relates to biochemical genetics	<p>Understand DNA structure (base sequence, pairing, replication, and packaging into chromosomes). Explain transcription, splicing, translation, and variation of gene expression between tissues. Explain genomic organization and gene structure. Understand core technologies for allele discrimination and mutation detection. Understand protein translation, modification, transport and regulation of these processes. Understand enzyme kinetics. Understand biochemical pathways and how they interrelate. Understand the effect of mutational change on protein activity and pathway interactions.</p>
	Understand principles and techniques of biochemical genetics	<p>Understand abnormalities of cell metabolism including but not limited to:</p> <ul style="list-style-type: none"> ○ Enzymopathies: single or multiple pathway effects ○ Receptor, transporter, structural protein (e.g., hemoglobin) ○ Disorders of development (e.g., cholesterol metabolism disorders) ○ Cellular structure disorders (e.g., lysosome formation disorders) ○ Mitochondrial disorders ○ Contiguous gene syndromes ○ Abnormalities of cell division ○ Abnormalities of cell differentiation ○ Abnormalities of cell migration <p>Understand treatment options for all biochemical disorders. Understand newborn screening and responses required to assist primary care physician as well as acute care situations.</p>
3. Interpersonal and Communication Skills		
Professional communication	Know how to communicate with colleagues	<p>Maintain comprehensive, timely and legible medical records. Communicate information to health professionals one-on-one or in groups. Effectively discuss test results and interpretations with physicians. Initiating referrals to clinical geneticists or other professionals as appropriate.</p>
	Effectively communicate complex, difficult or challenging information	<p>Effectively communicate errors, complications, adverse events, and bad news</p>

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	Exhibit appropriate ethical and professional standards at all times	Demonstrate an attitude of responsibility and respect toward the patient, a respectful and cooperative attitude toward professional colleagues and an honest, forthright manner in carrying out professional task. Demonstrate knowledge of the institution's policies and procedures regarding communication with patients. Use appropriate language to explain laboratory testing to members of the healthcare team, including patients.
	Know how to teach and supervise	Educate, mentor, and assess progress and skills, and provide appropriate feedback and appraisal.
4. Practice-Based Learning and Improvement		
Standards of care	Knowledge of relevant practice guidelines or consensus statements	Compare own laboratory practices and outcomes to accepted practice/guidelines and national or peer-reviewed data. Identify areas for practice improvement.
Ongoing learning	Know how to keep up to date in common biochemical genetics topics	Participate in educational activities including local seminars, and regional and national meetings. Critique research evidence for applicability to laboratory practice. Use appropriate bioinformatics resources. Seek feedback from others. Recognize the importance of continuing Certification programs.
Quality improvement	Know quality metrics	Change practice behaviors in response to feedback from others and review of own practice; apply new skills or knowledge to laboratory service. Exhibit willingness to change and to adapt.
5. Professionalism		
Responsibility	Understand the responsibility to the ordering physician and patient/family	Complete tasks required to provide laboratory services effectively in a careful and thorough manner.
Practices within ability	Recognize limits of his/her abilities	Seek consultation when appropriate. Exercise authority accorded by position and/or experience. Recognize cognitive, legal and ethical limitations of credentials.
Patient diversity	Recognize differences (cultural, educational, etc.)	Recognize each patient's unique needs and characteristics. Provide equitable services regardless of patient culture or socioeconomic status. Respectful and sensitive to issues related to patient culture, age, gender and disabilities.
Integrity and ethical behavior	Recognize ethical dilemmas and potential conflicts of interest. Be knowledgeable about the elements of informed consent, privacy, confidentiality, duty to warn, and HIPAA compliance	Take responsibility for actions; admit mistakes, try to address ethical dilemmas and conflicts of interest. Demonstrate commitment to ethical principles pertaining to: (1) patient privacy and autonomy, (2) provision or withholding of test results, (3) confidentiality of patient information, (4) informed consent, (5) conflict of interest, and (6) business practices.
Health professional relationships	Know how to interact with health professionals	Be courteous and respectful when relating with peers and referring healthcare providers.
Leadership	Know teamwork and leadership skills and know how to teach and supervise	Provide direction to staff. Educate and mentor. Assess progress and skills, provide appropriate feedback and appraisal.

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6. Systems-Based Practice		
Service coordination	Know how to provide comprehensive and integrated service	Coordinate services with other providers. Provide timely service.
Evidence-based medicine	Knowledge of evidence-based guidelines and appropriate billing	Determine cost and cost components of tests and understand reimbursement issues. Provide cost-conscious services. Consider cost and benefits of test. Follow accepted laboratory guidelines. Use appropriate billing codes.
Evidence-based medicine	Understand research principles	Critically read and interpret scientific publications. Have awareness of policy implications.
Health services	Knowledgeable about system resource utilization; understand different healthcare delivery systems and medical practices	Interface with laboratory information systems, electronic health records, and billing systems.
Health services	Information access	Conduct literature review and database searches. Identify resources for the patient/family and referring healthcare provider.