American Board of Medical Genetics and Genomics

Clinical Biochemical Genetics Competencies

July 2025

Introduction: This revised learning guide was created to assist program directors in designing, implementing, monitoring, and evaluating the educational content of ACGME-accredited programs in medical genetics and genomics; trainees may also find this to be a useful resource. Note that the content is not meant to be all-inclusive. While this learning guide covers a breadth of topics it comprises only a subset of the knowledge and expertise required of a practicing medical genetics professional.

OBJECTIVES	SKILLS
Patient Care: Pre-analytic laboratory skills	
Identify appropriate specimens for study and methods for collection, preservation, and transport	▪ Identify appropriate specimen age, containers, anticoagulants, collection media, or preservative(s) for validated specimen types. ▪ Identify factors important for the transport of specimens, such as overnight delivery, appropriate transport media and containers, or recommended temperatures. ▪ Understand how to transport/ship specimens off-site using packaging that meets OSHA guidelines. ▪ Be aware of appropriate specimen handling and storage requirements.
Assess acceptability of specimen for study	▪ Check for appropriate labeling of specimen and requisition with at least two identifiers. ▪ Evaluate suitability and quality of specimen for requested study, both for type and amount/volume required as well as correct collection tube (i.e., sodium heparin vs. EDTA). ▪ Assess for presence of interfering substances (i.e., hemolysis, lipemia, icterus). ▪ Describe methods for possible recovery of samples of suboptimal quality. ▪ Notify appropriate individuals of unsatisfactory samples and document such notification per laboratory policy and regulatory requirements.
Accession specimen	▪ Demonstrate knowledge in assigning laboratory accession number in database or Laboratory Information System (LIS), as applicable. ▪ Be familiar with data fields required for accessing a sample. ▪ Be aware of specific data fields that may be required for a particular test (e.g., age).
Tracking of specimen and documentation	▪ Follow protocols to ensure proper identification and location of patient materials through the complete process - from accession to final report. ▪ Track specimen location through all aspects of the testing process. ▪ Maintain necessary records and laboratory database, in logbooks or computers, as appropriate.

OBJECTIVES	SKILLS
Patient Care: Analytic laboratory skills	
Sample preparation	▪ Document processes to exclude expired or contaminated reagents. ▪ Monitor and document the effectiveness of all reagents prior to clinical use. ▪ Demonstrate knowledge and skill in sample preparative procedures and laboratory practices that prevent cross-contamination between samples.
Principles of chromatographic separation (liquid chromatography/gas chromatography)	▪ Demonstrate knowledge in applying principles of liquid chromatography (selection of a column/mobile phase) to most common biochemical genetics analyses (amino acids, etc.). ▪ Demonstrate knowledge in applying principles of gas chromatography to organic acids analysis.
Analysis by tandem mass spectrometry (MS/MS)	▪ Demonstrate knowledge in applications, limitations, and interpretations of MS/MS-based results.
Know and understand principles and techniques associated with amino acid analysis	▪ Perform amino acid analysis. ▪ Evaluate amino acid chromatograms or equivalent (including baseline review, peak identification and co-eluting compounds). ▪ Troubleshoot common problems of amino acid analysis. ▪ Report results with proper units, control range, and clinically relevant interpretation.
Know and understand principles and techniques associated with organic acid analysis	▪ 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 qualitative and/or quantitative analysis and reporting of organic acids.
Know and understand principles and techniques associated with acylcarnitine analysis	▪ Perform acylcarnitine analysis. ▪ Evaluate acylcarnitine profiles (including identification of different species, presence of isobaric or isomeric compounds, etc.). ▪ Troubleshoot common problems of acylcarnitine analysis. ▪ Report results with proper units, control range, and clinically relevant interpretation.
Know and understand principles and techniques associated with enzyme assays	▪ Demonstrate knowledge in assay conditions, use of positive and negative controls, limitations (e.g., pseudodeficiencies), and data interpretation related to determination of affected and carrier status.

OBJECTIVES	SKILLS
Know and understand principles and techniques associated with single-analyte 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.
Know and understand principles and techniques associated with other separation techniques: thin-layer chromatography, electrophoresis, etc.	▪ 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.
Know and understand principles and techniques associated with newborn screening (NBS)	▪ Be familiar with national recommendations for NBS panels, sample collection, turn-around-time, follow-up, etc. ▪ Know testing methods, limitations, results, and interpretations of screening procedures. ▪ Be familiar with methods to improve performance of NBS (second tier, use of ratios, use of databases, algorithms, etc.) ▪ Know procedures for communicating abnormal results and coordinating follow up testing.
Patient Care: Post-analytic laboratory skills	
Results interpretation	▪ Recognize clinically significant metabolite patterns. ▪ Determine the clinical significance of enzyme results (affected or carrier status, as appropriate; pseudodeficiency). ▪ Integrate results from other studies and/or clinical findings. ▪ Make recommendations for additional testing. ▪ Assess the need to report results to appropriate provider/health care team.
Reporting	▪ 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. ▪ Recognize when critical results need to be communicated promptly to the healthcare provider. ▪ Document conversations when giving oral results. ▪ Understand when and how to amend or addend a report.
Software	▪ Use and understand software packages for clinical lab processing, data analysis and storage, and for report writing. ▪ Understand implications of using electronic record keeping with respect to private health information.

OBJECTIVES	SKILLS
	▪ Understand the informatics processes that connect sample requisition to wet lab processes, data analysis, report writing, and transmission of final reports to referring physicians .
Medical Knowledge	
Understand principles of biology and genetics	▪ Understand principles of general biology and genetics as it relates to biochemical genetics including: ○ Basis of inheritance ○ Chromosome and gene structure and function ○ Population genetics ○ Disorders of growth and development ○ Mutation and inheritance ○ Single gene disorders ○ Multifactorial/complex genetic disorders ○ Cytogenetics ○ Epigenetics ○ Biochemical Genetics ○ Exome and genome sequencing ○ Direct to consumer genetic testing ○ Genomic medicine ○ Gene environmental interactions ○ RNA biology ○ Principles of gene therapy ▪ Understand abnormalities of cell metabolism including but not limited to: ○ Enzymopathies: single or multiple pathway effects; role of co-factors and their ability to cause multiple pathway effects ○ Receptor, transporter, structural protein (e.g., hemoglobin, amino acid transporters) ○ Disorders of development (e.g., cholesterol metabolism disorders) ○ Cellular structure disorders (e.g., lysosome formation disorders) ○ Mitochondrial disorders ○ Peroxisomal disorders ○ Contiguous gene syndromes ○ Abnormalities of cell division ○ Abnormalities of cell differentiation ○ Abnormalities of cell migration ○ Abnormalities of cell trafficking

OBJECTIVES	SKILLS
	▪ Understand treatment options for all biochemical disorders. ▪ Understand newborn screening and responses required to assist primary care physician as well as acute care situations.
Interpersonal and Communication Skills	
Ability to communicate effectively with colleagues	▪ Maintain comprehensive, timely and legible medical records. ▪ Effectively communicate errors, complications, adverse events, and unanticipated results. ▪ Effectively discuss test results and interpretations with physicians. ▪ Communicate information to health professionals one-on-one or in groups. ▪ Understand and adhere to HIPAA guidelines. ▪ Recommend referrals to clinical geneticists or other professionals, as appropriate.
Consistently maintain appropriate ethical and professional standards	▪ Demonstrate an attitude of responsibility and respect toward the patient, a respectful and cooperative attitude toward professional colleagues, and an honest, forthright manner in conducting professional tasks. ▪ 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 and their families.
Learn how to teach and supervise effectively	▪ Educate, mentor, and assess progress and skills, and provide appropriate feedback and appraisal.
Practice-Based Learning and Improvement	
Know how to keep up to date in biochemical genetics topics	▪ Participate in educational activities including local seminars and regional and national meetings. ▪ Critique research evidence for applicability to laboratory practice. ▪ Apply new skills or knowledge to laboratory service. ▪ Use appropriate bioinformatics resources. ▪ Recognize the importance of Continuing Certification Programs.
Receiving and incorporating feedback	▪ Compare own laboratory practices and outcomes to accepted practice/guidelines and national or peer-reviewed data. ▪ Identify areas for practice improvement. ▪ Seek feedback from others and exhibit willingness to change and to adapt. ▪ Change practice behaviors in response to feedback from others and review of own practice.

OBJECTIVES	SKILLS
Professionalism	
Practices within ability and recognizes limits of one's abilities	▪ Seek consultation, when appropriate. ▪ Exercise authority according to position and/or experience. ▪ Recognize cognitive, legal, and ethical limitations of credentials.
Awareness of patient diversity	▪ Recognize each patient's unique needs and characteristics. ▪ Provide equitable services regardless of patient culture or socioeconomic status. ▪ Be respectful and sensitive to issues related to patient culture, age, gender, and disabilities.
Demonstrate integrity and ethical behavior	▪ Complete tasks required to provide laboratory services effectively in a timely and thorough manner. ▪ Take responsibility for actions; admit mistakes, try to address ethical dilemmas and conflicts of interest. ▪ Demonstrate knowledge of and commitment to ethical principles pertaining to: ○ Patient privacy and autonomy ○ The provision or withholding of test results ○ Confidentiality of patient information ○ Informed consent ○ Conflict of interest ○ Business practices that conflict with stated principles of professionalism ▪ Recognize ethical dilemmas and potential conflicts of interest.
Know how to interact with health professionals	▪ Be courteous and respectful when relating with peers and referring healthcare providers.
Demonstrate teamwork and leadership skills and effectively teach and supervise	▪ Provide direction to staff. ▪ Educate and mentor other trainees and laboratory staff. ▪ Assess progress and skills and provide appropriate feedback and appraisal.
Well-being awareness	▪ Identify signs of fatigue/burn-out in self (and others) and be aware of resources for well-being.

OBJECTIVES	SKILLS
Systems-Based Practice	
Knowledge of evidence-based guidelines and appropriate billing	▪ Understand how to determine operating cost and cost components of tests. ▪ Understand how laboratory test reimbursement generally works.

OBJECTIVES	SKILLS
	▪ Provide cost-conscious services. ▪ Consider the costs and benefits of the test. ▪ Follow accepted laboratory guidelines (e.g., ACMG standards and guidelines). ▪ Understand appropriate use of billing (CPT) and international classification of diseases (ICD) codes.
Understand research principles/evidence-based medicine	▪ Critically read and interpret scientific publications.
Understand system resource utilization, different healthcare delivery systems, and medical practices	▪ Interface with laboratory information systems, electronic health records, and billing systems.
Ability to access pertinent information	▪ Conduct comprehensive literature review and database searches. ▪ Identify resources for the patient/family and the referring healthcare provider.
Know how to provide comprehensive and integrated services	▪ Coordinate services with other providers and specialty clinics. ▪ Provide timely service.
Awareness of public policies pertinent to clinical testing	▪ Stay informed about current legislation and policies and understand how they can impact the regulation of genetic testing. ▪ Have familiarity with research/clinical boundaries and understand situations in which IRB approval is needed.
Quality Control	
Use of aseptic techniques	▪ Use Universal Precautions for protection against potential exposure to infectious agents (e.g., protective clothing, gloves and masks, containers for same delivery and waste disposal, biological safety cabinets). ▪ Use and document methods to detect, identify, control, and eliminate microbial or chemical contamination. ▪ Practice measures that prevent cross-contamination between samples.
General laboratory skills	▪ Select, operate, clean, and maintain all laboratory equipment and instruments, as appropriate. ▪ Know how to prepare and store reagents and specimens.

OBJECTIVES	SKILLS
Assay controls	▪ Understand the purpose of using appropriate positive, negative, and blank controls when performing and interpreting laboratory testing.
Assay validation	▪ Understand the principles of assay validation or equivalency assessments and be familiar with technical guidelines for the development of clinical assays.
Proficiency testing	▪ Understand the role of proficiency testing (PT) and regulations clinical laboratories must follow with respect to the type and frequency of PT that must be performed.
Laboratory accreditation	▪ Be familiar with the requirements of regulatory agencies such as the College of American Pathologists (CAP), Clinical Laboratory Improvement Amendments (CLIA), Joint Commission (JC), and have awareness of any additional state-level regulations that impact clinical laboratories.
Quality	▪ Understand laboratory quality control, quality assurance, and quality management in all areas and comply with all regulatory requirements.
Safety	
Laboratory and data safety	▪ Complete institutional safety training and be familiar with safety protocols pertaining to both laboratory and safety practices. ▪ Identify specimen and reagent disposal needs in compliance with safety, chemical, and biosafety guidelines. ▪ Identify personal protection practices (e.g., gloves, gowns, eyewear) and equipment indicated for processing of laboratory specimens/reagents. ▪ Identify cybersecurity risks and follow practices to minimize potential breaches.