American Board of Medical Genetics and Genomics

Medical 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	
Recognize and initiate care for a patient with a suspected inherited metabolic disorder	▪ Recognize the signs and symptoms of acute metabolic decompensation (altered mental status). ▪ Know the common triggers for a metabolic crisis (fever, infection). ▪ Take an accurate and specific medical history and document pertinent positives and negatives (family history, episodic nature of illness, vomiting, diarrhea). ▪ Perform an appropriate physical examination and document pertinent findings. ▪ Recognize abnormal laboratory values (ammonia, lactate). ▪ Follow protocols to ensure proper management of the patient and know when to escalate care to the ICU. ▪ Determine appropriate diagnostic work-up and coordinate with laboratory services to ensure it is done in a timely manner. ▪ Review, interpret, and discuss results with attending physician, primary service, and patient/family. ▪ Understand the principles of dietary and pharmacological therapies, as they apply to metabolic disorders. ▪ Suggest appropriate interventions (e.g., IV fluids with dextrose, carnitine, protein restriction). ▪ Communicate recommendations effectively with primary team or other health care providers. ▪ Devise appropriate long-term management plans.

OBJECTIVES	SKILLS
Know and understand the principles and processes associated with newborn screening (NBS)	▪ Be familiar with national recommendations on 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 (e.g., second tier, use of ratios, use of databases, algorithms, etc.). ▪ Formulate a differential diagnosis based on NBS results and determine appropriate diagnostic work-up. ▪ Recognize critical results (e.g., , abnormal screen suggestive of VLCAD deficiency or GA1) and appropriately manage the patient and the diagnostic testing. ▪ Know procedures for communicating abnormal results and coordinating follow-up testing and effectively communicate with the primary care team and patient/family.
Apply knowledge of biochemistry and metabolism to the diagnosis and treatment of inherited metabolic disorders	▪ Recognize the different signs and symptoms of inherited metabolic disorders, including urea cycle disorders, amino acidopathies, organic acidemias, fatty acid oxidation disorders, disorders of carbohydrate metabolism, mitochondrial disorders, peroxisomal disorders, and lysosomal disorders. ▪ Identify appropriate diagnostic and monitoring testing. ▪ Recognize the strength and limitations of the different testing methodologies. ▪ Order and evaluate results of diagnostic testing and formulate a diagnosis. ▪ Formulate a treatment/management plan based on the diagnosis. ▪ Understand principles of enzyme replacement/substitution and gene therapy and consider these options when determining treatment, as applicable. ▪ Determine changes to dietary/pharmacological management based on monitoring testing results, as applicable. ▪ Determine if a patient needs critical care. ▪ Identify the need for emergency protocol letters and draft them. ▪ Recognize recurrence risk and the need for testing of family members, as applicable.
Biochemical genetics results interpretation	▪ Be familiar with the patterns observed for the most common biochemical genetics tests (e.g., amino acids, organic acids, acylcarnitines, MPS screening, enzyme assays). ▪ Recognize clinically significant metabolite patterns and distinguish from common causes of abnormal findings (e.g., physiologic ketosis and/or ketogenic diet; pattern of dicarboxylic aciduria secondary to MCT oil in diet vs. MCAD or MADD deficiency; etc.). ▪ Determine the clinical significance and the limitations of enzyme results (affected or carrier status as appropriate; pseudodeficiency). ▪ Integrate results from other studies and/or clinical findings. ▪ Recognize when additional testing is needed.
Software	▪ Utilize appropriate tools such as gnomAD, ClinVar, Decipher, and biochemical databases (IEMbase, HMBD, etc.). ▪ Become familiar with electronic health records (EHR), including ordering tests, medications, and imaging studies.

OBJECTIVES	SKILLS
	▪ Demonstrate familiarity with clinical laboratory information system, test ordering, and retrieval of results. ▪ Recognize how laboratory results are integrated into the EHR. ▪ Understand the implications of using electronic record keeping with respect to private health information.
Document relevant information for health professionals, patients, and families	▪ Write consultation reports, progress notes, and letters that include relevant information about the diagnosis, test options and results, risk assessment, and management plan.
Medical Knowledge	
Understand principles of biology and genetics	▪ Understand basic principles of general biology, genetics, and genomics including: o Basis of inheritance o Chromosome and gene structure and function o Population genetics o Disorders of growth and development o Mutation and inheritance o Single gene disorders o Multifactorial/complex genetic disorders o Cytogenetics o Epigenetics o Biochemical Genetics o Exome and genome sequencing o Direct to consumer genetic testing o Genomic medicine o Gene environmental interactions o RNA biology o Principles of gene therapy ▪ Understand abnormalities of cell metabolism including but not limited to: o Enzymopathies: single or multiple pathway effects; role of co-factors and their ability to cause multiple pathway effects o Receptor, transporter, structural protein (e.g., hemoglobin, amino acid transporters) o Disorders of development (e.g., cholesterol metabolism disorders) o Cellular structure disorders (e.g., lysosome formation disorders) o Mitochondrial disorders o Peroxisomal disorders

OBJECTIVES	SKILLS
	○ Contiguous gene syndromes ○ Abnormalities of cell division ○ Abnormalities of cell differentiation ○ Abnormalities of cell migration ○ Abnormalities in cell trafficking ▪ Understand treatment options for all biochemical disorders. ▪ Understand newborn screening and responses required.
Interpersonal and Communication Skills	
Ability to communicate effectively with colleagues and patients	▪ Maintain comprehensive, timely, and legible medical records. ▪ Effectively communicate errors, complications, adverse events, and unanticipated results. ▪ Effectively addresses patients' concerns; provide psychosocial support, help families and individuals recognize and cope with their emotional and psychological needs. ▪ Communicate information to health professionals one-on-one or in groups. ▪ Be familiar with and use of the most appropriate method of communication (pager, text message, telephone call, in-person, electronic health record) for transfer of care and documentation of care. ▪ Initiate referrals to 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 care plans to members of the healthcare team, and 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 patient care. ▪ Use appropriate bioinformatics resources. ▪ Recognize the importance of Continuing Certification programs.

OBJECTIVES	SKILLS
Receiving and incorporating feedback	▪ Compare own practices, treatment, 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.
Professionalism	
Practice within ability and recognize 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. ▪ Practice with humility.
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 care for patients effectively in a timely, careful, and thorough manner. ▪ Take responsibility for actions, admit mistakes, try to address ethical dilemmas and conflicts of interest. ▪ Act with integrity. ▪ Demonstrate knowledge 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 staff. ▪ Assess progress and skills and provide appropriate feedback and appraisal. ▪ Take leadership role in family/team meetings.

OBJECTIVES	SKILLS
Well-being awareness	▪ Identify signs of fatigue/burn-out in self (and others) and be aware of resources for well-being.
Systems-Based Practice	
Knowledge of evidence-based guidelines and appropriate billing	▪ Provide cost-conscious care. ▪ Consider the costs and benefits associated with testing and treatment. ▪ Be familiar with the cost and insurance coverage for genetic testing and treatment. ▪ Follow accepted patient care pathways. ▪ Use appropriate international classification of diseases (ICD) and billing (CPT) codes. ▪ Be aware of studies/data supporting an approach to patient management and apply them as appropriate.
Understand research principles/evidence-based medicine	▪ Critically read and interpret scientific publications. ▪ Be familiar with resources to refer interested patients to clinical trials. ▪ Consider implications for clinical practice, and stakeholders in implementation.
Understand system resource utilization, different healthcare delivery systems, and medical practices	▪ Ensure patient awareness of, and advocate for, patient access to available care options. ▪ Recognize and manage variations in access to genetic/genomic services and testing. ▪ Make appropriate referrals to community resources. ▪ Participate in systems to identify errors and implement potential system solutions.
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 care with a range of health care providers, including MDs, genetic counselors, primary care providers, specialists, nurses, nutritionists, clinic personnel, lab personnel, and in the multidisciplinary clinic setting. ▪ Provide timely care. ▪ Document essential elements of a genetics encounter to enhance the transfer of information and patient safety, utilizing decision support tools as needed.
Awareness of pertinent public policies	▪ 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.