American Board of Medical Genetics and Genomics

Medical Biochemical Genetics Competencies

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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		
Initial or Acute Management	Recognizes signs and symptoms of inborn errors of metabolism	Takes accurate and specific history Documents important pertinent positives and negatives (family history, episodic nature of illness)
	Recognizes signs and symptoms of acute metabolic decompensation	Takes accurate and specific history and documents important pertinent positives and negatives (vomiting, diarrhea, fever)
	Selects and orders diagnostic studies	Determines a diagnostic work-up Effectively communicates work up with primary team or other health care providers Ensures work up is done in timely manner Reviews, interprets and discusses results with attending physician, primary service and family.
	Provides supportive and disease-specific management	Follows protocols to ensure proper management of the patient Devises long-term management plans
Chronic management	Assesses clinical outcomes of patient-specific treatment and therapies	Understand the principles of dietary and pharmacological therapies as they apply to metabolic disorders
	Monitors treatment with appropriate tests	Reviews, interprets and discusses results with family and makes appropriate changes to therapy as needed.

DOMAIN	OBJECTIVES	SKILLS
Documentation	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
Newborn screening	Identifies principles and processes of newborn screening Selects appropriate follow-up for abnormal newborn screen results	Recognize the limitations and strengths of methodologies used for NBS Is familiar with the disorders included in NBS panel nationwide and in the region of practice Formulate a differential diagnosis based on NBS results and determine appropriate diagnostic work-up Recognize critical results (for example abnormal screen suggestive of VLCAD deficiency or GA 1) and appropriately manages the patient and the diagnostic testing Effectively communicates with primary care team and families
Disorders affecting amino acid metabolism (amino acidopathies, organic acidemias)	Applies knowledge of biochemistry and metabolism to the diagnosis and treatment of amino acidopathies and organic acidemias	Recognize the different signs and symptoms of amino acidopathies and organic acidemias Identifies the appropriate diagnostic and monitoring testing Evaluate results of diagnostic tests and formulate a diagnosis Determine changes to dietary/pharmacological therapy based on monitoring test results Determine if a patient needs critical care Identify the need for emergency protocol letters
Fatty acid oxidation disorders	Applies knowledge of biochemistry and metabolism to the diagnosis and treatment of fatty acid oxidation disorders	Recognize the different signs and symptoms of fatty acid oxidation disorders Identify the appropriate diagnostic and monitoring tests Evaluate results of diagnostic tests and formulate a diagnosis Determine changes to dietary/pharmacological therapy based on monitoring test results Determine if a patient needs critical care Identifies the need for emergency protocol letters
Urea cycle disorders	Applies knowledge of biochemistry and metabolism to the diagnosis and treatment of urea cycle disorders	Recognize the different signs and symptoms of urea cycle disorders Identifies the appropriate diagnostic and monitoring testing Evaluate results of diagnostic tests and formulate a diagnosis Determine changes to dietary/pharmacological therapy based on monitoring test results Determine if a patient needs critical care Identify the need for emergency protocol letters
Storage disorders	Applies knowledge of biochemistry and metabolism to the diagnosis and treatment of storage disorders	Recognize the different signs and symptoms of storage disorders Selects the most appropriate diagnostic strategy Recognize the limitations and the strengths of the different testing methodologies Evaluate results of diagnostic tests and formulate a diagnosis Determine the most appropriate treatment for specific storage disorders Determine if a patient needs critical care Identifies principles of enzyme replacement and gene therapy
Disorders of carbohydrate metabolism	Applies knowledge of biochemistry and metabolism to the diagnosis and treatment of disorders of carbohydrate metabolism	Recognize the different signs and symptoms of disorders of carbohydrate metabolism Identifies the appropriate diagnostic and monitoring testing Evaluate results of diagnostic tests and formulate a diagnosis Determine changes to dietary/pharmacological therapy based on monitoring test results Determine if a patient needs critical care Identifies the need for emergency protocol letters

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Mitochondrial disorders	Applies knowledge of biochemistry and metabolism to the diagnosis and treatment of mitochondrial disorders	Recognize signs and symptoms of mitochondrial disorders Identifies the appropriate diagnostic testing Evaluate results of diagnostic tests and formulate a diagnosis Formulate a treatment plan based on diagnosis Recognize recurrence risk based on molecular diagnosis
Biochemical genetics results interpretation	Familiarize with the patterns observed for the most common biochemical genetics tests (amino acids, organic acids, acylcarnitines, MPS screening, enzyme assays)	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 Recognize when additional testing is needed
Bioinformatics	Software	Become familiar with electronic medical records, including ordering tests, medications, and imaging studies Demonstrates familiarity with clinical laboratory information system, test ordering, and retrieval of results Identify implications of using electronic record keeping with respect to private health information (PHI) Recognize how laboratory results are integrated in the HER Utilize appropriately tools such as gnomAD, ClinVar, Decipher, and biochemical databases (IEMbase, HMBD, etc.)
2. Medical Knowledge		
General principles of biology, genetics and genomics	Demonstrate understanding of principles of general biology, genetics, and genomics	Apply 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 environment interactions o RNA biology o Principles of gene therapy)
	Understand principles and techniques of biochemical genetics	Understand abnormalities of cell metabolism including but not limited to: o Enzymopathies: single or 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 o Contiguous gene syndromes

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		○ Abnormalities of cell division ○ Abnormalities of cell differentiation ○ Abnormalities of cell migration • Understand treatment options for all biochemical disorders • Understand newborn screening and responses required to assist primary care physician, as well as acute care situations
3. Interpersonal and Communication Skills		
Professional communication	Communicates effectively with colleagues	Maintain comprehensive, timely and legible medical records Communicate information to health professionals one-on-one or in groups Familiarity 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
	Effectively communicate complex, difficult or challenging information	Effectively addresses patients' concerns; provide psychosocial support, help families and individuals recognize and cope with their emotional and psychological needs Recognizes situations requiring psychiatric referral Display a responsive, attentive, and reassuring demeanor, as appropriate
	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
	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 and implementation of relevant practice guidelines or consensus statements	Develop method to assess practice and impact of care Compare own practice treatment/care outcomes to accepted practice/guidelines and national or peer-reviewed data Identify areas for practice improvement
Ongoing learning	Up-to-date clinical genetics/ genomics and biochemical genetics knowledge	Participate in educational activities including local seminars, and regional and national meetings Critique research evidence for applicability to patient care Use appropriate bioinformatics resources Seek feedback from others Recognize the importance of Maintenance of Certification programs. Critically read and interpret scientific literature Participate in education of patients, families, students, residents and other health professionals
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 patient care Exhibit willingness to change and to adapt

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5. Professionalism		
Responsibility	Exhibit responsibility to patient/family and other health-care professionals	Complete tasks required to care for patients effectively in a careful and thorough manner. Recognize challenging situations regarding maintaining personal, emotional and physical health.
	Maintain personal, emotional, physical and mental health of individual professional and colleagues	Recognize fatigue and sleep deprivation Recognize signs of physician impairment Ask for help when needed Maintain professional appearance Recognition of signs of stress or impairment in colleagues, and knowing what resources are available
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 Practice with humility
Patient diversity	Recognize values and priorities of diverse populations	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 Is 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 Acts with integrity 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	Is courteous and respectful when relating with peers and referring healthcare providers
Leadership	Demonstrate 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
6. Systems-Based Practice		
Care coordination	Provide comprehensive and integrated care, transitions of care as patients age	Coordinate care with a range of health care providers, including MDs, genetic counselors, primary care providers, specialists, nurses, nutritionists, clinic personnel, and in the multidisciplinary clinic setting Provide timely care Document essential elements of a genetics encounter to enhance transfer of information and patient safety, utilizing decision support tools as needed
Evidence-based medicine	Use evidence-based guidelines, up-to-date clinical genetics literature, and appropriate billing	Provide cost-conscious care Consider costs & benefits of test and treatments Be familiar with cost and insurance coverage for genetic testing and treatment Follow accepted patient care pathways Use appropriate billing codes

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Translational medicine	Apply new research principles and knowledge in clinical genetics/ genomics care	Critically read and interpret scientific publications Consider implications for clinical practice, and stakeholders in implementation
Health services	Understand system resource utilization, different healthcare delivery systems and medical practices; recognize potential errors in systems	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, implement potential system solutions
Information access	Utilize appropriate genetic/ genomic information resources	Conduct literature review and database searches as well as identification of resources for the patient/family and referring healthcare provider