American Board of Medical Genetics and Genomics

Clinical Genetics and Genomics 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 [includes: pediatric genetics, dysmorphology; biochemical genetics; preconception, prenatal and preimplantation diagnosis and fetal dysmorphology; neurogenetics; adult genetics, cancer genetics; genomic medicine; public health/population-based genetics practice]		
Medical history	Conduct a thorough and effective genetics-focused medical interview	Obtain information about the patient's medical history in an objective, non-judgmental way. Note pertinent positives and negatives. Record pertinent habits, exposures, medications, allergies, menstrual, contraceptives and pregnancy history, and prevention and screening activities. Build rapport through appropriate eye contact, relaxed body language and pleasant affect.
Family history	Obtain and record a genetics-focused family history using standard pedigree nomenclature	Obtain information from patient/family to construct at least a three generation pedigree (all 1st and 2nd degree relatives at a minimum) using standard symbols to depict donor gametes, adoption, and gender identity Document vital status, current age or age at death, cause of death if deceased, disease status/conditions including age at diagnosis/onset, and pregnancy/reproductive history. Document ancestry/grandparents' country of origin, consanguinity, twin and adoption status.
Social history	Gather a complete social history	Gather information about the patient's living situation, educational history, employment history and family dynamics. Elicit smoking history, alcohol history and medication or drug use relevant to the reason for visit.
Physical examination	Conduct an appropriate physical exam relevant to the chief complaint, reason for referral and/or genetic condition	Conduct appropriate physical exam in a logical and efficient sequence. Note pertinent positives and negatives. Display sensitivity to patient comfort, respect for patient privacy. Recognize different patterns of dysmorphology and normal variants.

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Ordering and Interpreting laboratory tests, imaging studies, and procedures relevant to genetic diseases	Recognize benefits and limitations of testing including analytic validity, clinical validity, and clinical utility of tests.	Identify best testing strategy and interprets results in the context of medical and family history, physical examination and other pertinent data. Select and prioritize diagnostic studies including interpreting laboratory data generated from biochemical genetic, cytogenetic and molecular genetic analyses. Interpret and guide collection of appropriate confirmatory testing for newborn screening (NBS) results. Recognize and interpret the different types of prenatal testing. Recognize germline and somatic mutations in cancer and appropriate testing to order. Identify when genomic testing (array comparative genomic hybridization, exome sequencing or genome sequencing) is appropriate. Utilize tools such as gnomAD, ClinVar, Decipher, to assess variants of unknown significance. Consider ethical, legal and social implications of testing. Provides appropriate pre- and post-test counseling for genetic and genomic tests, including implications of secondary findings on genomic testing.
	Distinguish between different types of genetic and genomic tests and different types of genetic variants	
	Recognize analytic techniques used in genetic and genomic tests	
Diagnosis	Exercise proper clinical judgment in assessing condition	Obtain sufficient information from interview (personal medical, family and social history), physical examination, procedures and laboratory results to formulate a differential diagnosis and a plan to reach the most likely diagnosis and to exclude relevant conditions that are in the differential diagnosis.
Risk Assessment/ pedigree analysis	Integrate genetic, genomic, and non-genetic data in risk assessment	Recognize and use relevant medical, family history, and genetic/genomic laboratory data pertinent to disease occurrence or recurrence for a patient, and the implications for family members.
Management, prevention, anticipatory care, and continuing care	Understand the importance of an appropriate treatment/care plan, including influence of environmental and lifestyle factors and screening for early detection and risk factors	Develop a plan that is appropriate for the condition, including pediatric, adult (adult-onset and cancer disorders) and prenatal disorders (of mother and fetus); plan should reflect a good understanding of the current, accepted medical genetics and genomics practice in the context of environmental and lifestyle factors. Manage acute metabolic crises and provides chronic management for patients with inborn errors of metabolism. Describe options for early detection and screening for risk factors. Familiarity with resources for management of Down syndrome, NF1, hereditary breast ovarian cancer syndrome from national organizations: e.g. American Academy of Pediatrics, American College of Medical Genetics and Genomics, National Cancer Comprehensive Network. Address patient's concerns and preferences. Describe the exposures and behaviors that interact with a genetic diagnosis or susceptibility. Be able to provide patient/family with resources such as disease-oriented support organizations and access to treatment or clinical trials.
Documentation	Communicate relevant information to health professionals and patients; Utilize technology to accomplish safe health care delivery	Write consultation reports, progress notes, and letters that include relevant information about the diagnosis, risk assessment, pedigree analysis, genetic/genomic test options and results, assessment and plan. Enhance transfer of information using EHR and utilize decision support tools.
Clinical procedures	Perform appropriate procedures to aid in diagnosis or management	Perform, skin biopsy, lumbar puncture, obtaining and documenting informed consent, clinical photography, etc.

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<tr><th colspan="3">2. Genetics Knowledge</th></tr>
<tr><th>DOMAIN</th><th>OBJECTIVES</th><th>SKILLS</th></tr>
<tr><td rowspan="2">General principles of biology, genetics and genomics</td><td>Demonstrate understanding of principles of general biology, genetics and genomics</td><td rowspan="2">Apply basic principles of general biology and 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 /Molecular genetics/ Laboratory Genetics and Genomics
Epigenetics
Biochemical Genetics
Exome and genome sequencing
Direct to consumer genetic testing
Genomic Medicine;
Gene environment interactions
RNA biology
Principles of Gene Therapy)
Obtain experience in genetic subspecialty area, including:
Prenatal diagnosis -identify high and low risk patients, principles of prenatal screening and when, who to screen
Cancer genetics - identify high and low risk patients, genetics of common cancer
Genetic screening
Genetic counseling
[Refer to General and Specialty Content Outlines and other resources for details.]</td></tr>
<tr><td>Demonstrate understanding of principles of embryology and normal growth and development</td></tr>
<tr><td>Medical knowledge</td><td>Demonstrate understanding and principles of clinical genetics and genomics</td><td>Know, critically evaluate, and effectively use current medical information and scientific evidence for patient care, including results from genetics/genomics laboratory tests, quantitative risk assessment and available bioinformatics.
Develop an appropriate differential diagnosis and know the appropriate treatment/care and plan/options for the spectrum of genetic conditions including:
pediatric genetics and dysmorphology
biochemical genetics and newborn screening
preconception, prenatal and preimplantation diagnosis and fetal dysmorphology
neurogenetics
adult/cancer genetics
genomic medicine
public health/population-based genetics practice</td></tr>
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DOMAIN	OBJECTIVES	SKILLS
		Specific categories and examples include: ○ cancer (common familial and rare syndromes) ○ cardiac disorders (e.g., atherosclerosis, cardiomyopathies, conduction defects) ○ congenital abnormalities (single and multiple malformations, deformations and disruptions, fetal and neonatal presentations); patterns of birth defects ○ connective tissue disorders (e.g., Marfan and Ehlers Danlos syndromes) ○ cystic fibrosis ○ chromosomal disorders (sporadic and familial numerical and structural) ○ deafness (isolated and syndromic) ○ dysmorphic syndromes (common and rare) ○ Fragile X syndrome and other intellectual disability syndromes ○ hematological disorders (hemoglobinopathies, hemophilia, thrombophilia, hemochromatosis) ○ Huntington disease and other adult onset hereditary neurodegenerative disorders ○ inborn errors of metabolism ○ learning disability (familial and syndromic causes) ○ mitochondrial disorders (myopathies/encephalopathies, mitochondrial depletion and Leber optic atrophy) ○ multifactorial disorders (neural tube defects, epilepsies and common adult onset disorders) ○ neurogenetic disorders (spinal muscular atrophy, spinocerebellar ataxias, hereditary neuropathies, hereditary spastic paraplegia, epilepsies, leukodystrophies, movement disorders, dementias, motor neuron diseases ○ neuromuscular disorders (myotonic dystrophy, Duchenne, Becker, limb girdle, FSH and Emery-Dreifuss muscular dystrophies) ○ neurocutaneous disorders (neurofibromatosis, tuberous sclerosis); ophthalmic genetic disorders (retinitis pigmentosa) ○ pharmacogenetic disorders (malignant hyperthermia and G6PD deficiency) ○ renal disorders (adult and infantile polycystic kidney disease) ○ skeletal dysplasias (achondroplasia, osteogenesis imperfecta, spondyloepiphyseal dysplasia) ○ teratogens (alcohol and anticonvulsants)
3. Interpersonal and Communication Skills		
Inheritance/risk counseling	Effectively utilize concepts of heritability, inheritance patterns, variability, heterogeneity, penetrance and the epidemiology/natural history of a condition	Transmit pertinent information in a comprehensible way that is relevant to the patient and family Explain genetic/genomic concepts and identify family members at risk. Identify relevant exposures and behaviors that might interact with a genetic risk. Recommend appropriate screening strategies for early detection and risk factor modification. Display a responsive, attentive, and reassuring demeanor, as appropriate. Provide appropriate resources for health care providers and patients/families. Respects privacy and follows HIPPA guidelines

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Psychosocial support/counseling	Develop a plan for patient and family support and counseling	Engage/contract with the client. Address patient's concerns; incorporate patient's preferences including their cultural values and the social context into treatment/care plan. 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.
	Recognize and manage conflicts	
Professional communication	Communicate effectively with colleagues; team-based care	Perform comprehensive, timely and legible medical record documentation and oral communication with care providers at all levels. 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. Acts as a member of a care team.
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. Reflect on areas of uncertainty to identify improvement needs and implement effective changes in practice.
Ongoing learning	Up-to-date clinical genetics/ genomics knowledge	Seeks feedback from others and receives feedback appropriately Research topics when needed. Critique research evidence for applicability to patient care. Use bioinformatics resources. Participate in ABMGG Continuing Certification. Identify strengths and deficiencies and set individual learning goals. Use IT resources. Critically read and interpret scientific literature. Participate in education of patients, families, students, residents and other health professionals.
Quality improvement	Utilize quality metrics in clinical genetics/genomics care	Change practice behaviors in response to Quality Improvement efforts and review of own practice; apply new skills or knowledge to patient care.
5. Professionalism		
Responsibility	Exhibit responsibility to patient/family; Maintain personal, emotional, physical and mental health of individual professional and colleagues	Complete tasks required to care for patients effectively in a careful and thorough manner. Recognize challenging situations regarding maintaining personal, emotional and physical health. 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.

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Practice within ability	Recognize limits of his/her abilities	Refer patients when appropriate; exercise authority accorded by position and/or experience. Practice with humility.
Patient diversity	Recognize values and priorities of diverse populations	Respond to each patient's unique needs and characteristics. Provide equitable care regardless of patient culture or socioeconomic status. Display respect and sensitivity to issues related to patient culture, age, gender and disabilities.
Integrity and ethical behavior	Recognize ethical dilemmas and potential conflicts of interest	Takes responsibility for actions; admits mistakes; addresses 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 clinical care, (3) confidentiality of patient information, (4) informed consent, (5) conflict of interest, and (6) business practices.
	Understands elements of informed consent, privacy, confidentiality, duty to warn and is HIPAA compliant	
Health professional relationships	Interact appropriately with health professionals	Display courtesy and respect when relating with peers and referring healthcare providers.
Leadership	Demonstrate teamwork, leadership and supervisory skills	Provide direction to staff. Educate and mentor. Assess progress and skills and 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.
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.