American Board of Medical Genetics and Genomics

Clinical Genetics and Genomics 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: History and Physical Examination	
Conduct a thorough and effective genetics-focused medical history interview	▪ Obtain information about the patient's medical history in an objective, non-judgmental, culturally relevant, and gender sensitive 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, pleasant effects,, and nonjudgmental tone.
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.
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.
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 and respect for patient privacy. ▪ Recognize different patterns of dysmorphology and normal variants.

OBJECTIVES	SKILLS
Patient Care: Selecting Tests, Interpreting Results, and Management of Genetic Conditions	
Recognize benefits and limitations of testing including analytic validity, clinical validity, and clinical utility of tests. **Distinguish between diverse types of genetic and genomic tests and different types of genetic variants** **Recognize analytic techniques used in genetic and genomic tests**	▪ Identify best testing strategy and interpret 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 distinct types of prenatal testing. ▪ Recognize germline and somatic mutations in cancer and appropriate testing to order. ▪ Identify when specific genomic testing is appropriate. ▪ Utilize online and machine learning to assess variants of unknown significance. ▪ Consider ethical, legal, and social implications of testing. ▪ Provide appropriate pre- and post-test counseling for genetic and genomic tests, including implications of secondary findings on genomic testing.
Exercise proper clinical judgment in assessing condition	▪ Obtain sufficient information from the 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.
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. ▪ Incorporate polygenic risk scores in risk assessment.
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); the 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 provide chronic management for patients with inborn errors of metabolism. ▪ Describe options for early detection and screening for risk factors. ▪ Be familiar with resources for management of common genetic conditions, as supported by national care guidelines from national organizations (i.e., American Academy of Pediatrics, American College of Medical Genetics and Genomics, National Cancer Comprehensive Network). ▪ Address patient's concerns and preferences. ▪ Be aware of environmental exposures that increase risks (i.e., cancer syndromes, smoking, teratogens, QT prolongation medications). ▪ Create a plan for newly discovered pharmacological and gene-based therapies for genetic conditions. ▪ Be able to provide patient/family with resources such as disease-oriented support organizations and access to treatment or clinical trials.

OBJECTIVES	SKILLS
	▪ Create a plan for how to utilize pharmacogenomic information in patient care.
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.
Perform appropriate procedures to aid in diagnosis or management	▪ Perform skin biopsy, lumbar puncture, obtain and document informed consent, clinical photography, use of machine learning tools/models to improve patient care, etc.
Medical Knowledge	
Understand principles of biology, genetics, and genomics	▪ Understand basic principles of general biology and genetics, 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 Polygenic risk assessment o Cytogenetics /Molecular genetics/ Laboratory Genetics and Genomics o Epigenetics o Biochemical Genetics o Metabolomics o Emerging technologies (i.e., long read sequencing, etc.) o Exome and genome sequencing o Direct to consumer genetic testing o Genomic Medicine o Gene environmental interactions o RNA biology o Pharmacogenomics o Emerging Therapies (Principles of Gene Therapy; cell-based therapies, small molecule therapies o Clinical trials (referrals, understanding the types of trials, off-label usage of treatments)

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	▪ Obtain experience in genetic subspecialty area, including: o Prenatal diagnosis - identify high and low risk patients, principles of prenatal screening and when, who to screen o Cancer genetics - differentiate among high risk, moderate risk, and average risk patients, o Genetic screening o Genetic counseling
Understand principles of clinical genetics and genomics	▪ 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: o Pediatric genetics and dysmorphology o Biochemical genetics and newborn screening (including dietary management) o Preconception, prenatal, and preimplantation diagnosis and fetal dysmorphology o Neurogenetics o Adult/cancer genetics o Genomic medicine o Public health/population-based genetics practice o o Some examples may include: o Cancer (common familial and rare syndromes) o Cardiac disorders (e.g., atherosclerosis, cardiomyopathies, conduction defects, acropathies) o Congenital abnormalities (single and multiple malformations, deformations and disruptions, fetal and neonatal presentations); patterns of birth defects o Connective tissue disorders (e.g., Marfan and Ehlers Danlos syndromes) o Cystic fibrosis o Chromosomal disorders (sporadic and familial numerical and structural) o Deafness (isolated and syndromic) o Dysmorphic syndromes (common and rare) o Fragile X syndrome and other intellectual disability syndromes o Hematological disorders (hemoglobinopathies, hemophilia, thrombophilia, hemochromatosis) o Huntington disease and other adult-onset hereditary neurodegenerative disorders o Learning disability (familial and syndromic causes) o Metabolic disorders (acute decompensation, chronic dietary management) o Mitochondrial disorders (myopathies/encephalopathies, mitochondrial depletion, and Leber optic atrophy) o Multifactorial disorders (neural tube defects, epilepsies, and common adult-onset disorders)

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	○ 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, abortifacients, and anti-seizure medications)
Interpersonal and Communication Skills	
Ability to communicate effectively with colleagues and patients	▪ Maintain comprehensive, timely, and legible medical record documentation. ▪ Document oral communication with care providers at all levels. ▪ Be familiar with and the 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. ▪ Present pertinent healthcare information in a comprehensible way that is relevant to the patient and family. ▪ Explain genetic/genomic concepts including heritability, inheritance patterns, variability, heterogeneity, and penetrance, and identify family members at risk. ▪ Discuss the epidemiology/natural history of a condition. ▪ Identify relevant exposures and behaviors that might interact with genetic risk. ▪ Recommend appropriate screening strategies for early detection and risk factor modification. ▪ Be responsive, attentive, and reassuring, as appropriate. ▪ Provide appropriate resources for health care providers and patients/families. ▪ Respect privacy and follow HIPAA guidelines.
Develop a plan for patient and family support and counseling and recognize and manage conflicts	▪ Engage/contract with the client. ▪ Address the patient's concerns; incorporate patient's preferences including their cultural values and the social context into treatment/care plan. ▪ Provide psychosocial support to help families and individuals recognize and cope with emotional and psychological needs. ▪ Initiate referrals to other professionals, including psychiatrists, as appropriate. ▪ Display a responsive, attentive, and reassuring demeanor, as appropriate while maintaining appropriate patient-physician boundaries.

OBJECTIVES	SKILLS
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. ▪ Maintain professional interactions that respect physician/colleague's boundaries in the healthcare setting. ▪ 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 clinical genetics topics	▪ Participate in educational activities including local seminars, and regional and national meetings. ▪ Participate in education of patients, families, students, residents, and other health professionals. ▪ Identify strengths and deficiencies and set individual learning goals. ▪ Reflect on areas of uncertainty to identify improvement needs and implement effective changes in practice. ▪ Identify appropriate resources and research topics, when needed. ▪ Critique research evidence for applicability to patient care. ▪ Apply machine learning to clinical care, as appropriate. ▪ Recognize the importance of Continuing Certification programs.
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. ▪ Change practice behaviors in response to feedback from others and review of own practice. ▪ Pursue feedback from others and exhibit willingness to change and to adapt.
Professionalism	
Practice within ability and recognize limits of one's abilities	▪ Seek consultation, when appropriate. ▪ Exercise authority according to position and/or experience. ▪ Maintain professional credentials (i.e., licensure, hospital credentialing, DEA, board certification, etc.) to provide care for patients. ▪ Practice with humility.
Awareness of patient diversity	▪ Recognize each patient's unique needs and characteristics (i.e., interpreter usage, pronoun preferences, physical needs). ▪ Provide equitable care regardless of patient culture or socioeconomic status.

OBJECTIVES	SKILLS
	▪ 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. ▪ Engage in patient safety event reporting and subsequent analysis, as appropriate. ▪ Demonstrate knowledge and commitment to ethical principles pertaining to: o Patient privacy and autonomy o The provision or withholding of clinical care o Confidentiality of patient information o Informed consent o Conflict of interest o Business practices that conflict with stated principles of professionalism
Know how to interact with health professionals	▪ Be courteous and respectful when relating with peers and referring to healthcare providers. ▪ Respect the role of the health care team in providing optimal care (i.e., primary care, other specialists, genetic counseling, dietitians).
Demonstrate teamwork and leadership skills and effectively teach and supervise	▪ Provide guidance to the healthcare team. ▪ Educate and mentor other trainees and team members. ▪ Assess progress and skills and provide appropriate feedback and appraisal. ▪ Take leadership role in family/team meetings.
Well-being awareness	▪ Identify signs of fatigue/sleep deprivation/burn-out in self (and others) and be aware of resources for well-being. ▪ Recognize challenging situations regarding maintaining personal, emotional, and physical health. ▪ Ask for help, when needed. ▪ Recognize signs of stress or impairment in colleagues.
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 protocols and guidelines. ▪ 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.

OBJECTIVES	SKILLS
Understand research principles/evidence-based medicine	▪ Critically read and interpret scientific publications. ▪ Be familiar with resources to refer interested patients to clinical trials. ▪ Understand the role of implementing recent technologies and treatments in an expanding clinical field of practice.
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 (i.e., social work, case management, hospice, advocacy groups, family groups, etc.). ▪ Participate in systems to identify errors and implement potential system solutions.
Ability to access pertinent information	▪ Conduct comprehensive literature reviews 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 professionals, including MDs, genetic counselors, primary care providers, specialists, nurses, nutritionists, clinic personnel, lab personnel, and in the multidisciplinary clinic setting. ▪ Provide timely care. ▪ Provide appropriate transitions of care as patients age. ▪ Document essential elements of a genetics encounter to enhance 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.