American Board of Medical Genetics and Genomics

Laboratory 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: Pre-analytic laboratory skills	
Identify appropriate specimens for study and methods for collection, preservation, and transport	▪ Identify appropriate specimen age, containers, anticoagulants, collection media, antibiotics, type of glass slide, 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 tissue 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.,: presence of blood in amniotic fluid, blood clots or hemolysis in peripheral blood samples, presence of spicules in a bone marrow sample, quality of bone core, FFPE fixation methods, slide type and tissue thickness for FISH studies, quality of DNA from paraffin-embedded blocks, DNA with suboptimal A260/A280 ratios, fragmented DNA, acceptance of cheek swab and saliva samples, etc. ▪ Ensure tumor purity meets minimum requirements for the testing ordered. ▪ Describe methods for recovery of samples of suboptimal quality. ▪ Notify appropriate individuals of unsatisfactory samples and document such notification per laboratory policy and regulatory requirements.

Objectives	Skills
Accession specimen	▪ Assign unique laboratory accession number to specimen. ▪ Evaluate appropriateness of testing ordered based on information such as the reason for referral, demographic information, previous test history. ▪ Record related data, including patient's name and required and pertinent information. This could encompass patient medical record number, date of birth, sex, clinical history, indication for study, referring physician, etc. ▪ Record accurate and complete information concerning specimen including type of tissue, anatomic site of collection, amount/volume, appearance, collection date and time, anticoagulant, preservative, etc.; record priority status of specimen and identify, as appropriate. ▪ Record notes related to any special test requests, particularly those requiring transport of samples to other laboratories. ▪ When applicable, record chromosomal region of interest in chromosomal studies, FISH, or DNA microarray and gene/sequence/variant of interest for molecular studies.
Tracking of specimen and documentation	▪ Follow protocols to ensure proper identification of patient materials through the complete process, from accession through final report. ▪ Track specimens, as well as any aliquots and/or cell cultures and sub-cultures, through all aspects of the testing process. ▪ Maintain necessary records and laboratory database, in logbooks or computers, as appropriate.
Employ appropriate culture techniques for specimen, considering the clinical indication for the study	▪ Choose appropriate media and/or additives such as sera, antibiotics, buffers, mitogens, and growth factors depending on sample type and test ordered. ▪ Select appropriate methods of preparation and storage of media to maintain pH, sterility, and ability to support growth. ▪ Document processes to exclude expired or contaminated reagents. ▪ Select culture equipment and vessels for closed or open culture systems. ▪ Select culture technique for specimen, considering type of tissue, methods of initiation, type of culture, and purpose for study. ▪ Appraise the effect of cell density on rate of growth and adjust appropriately (e.g., cell count of leukemic specimens). ▪ Monitor and document the effectiveness of all assay solutions prior to use on diagnostic material. ▪ Record complete information for culture of specimen, including identification of technologist, lot numbers of media, sera, growth factors, and other reagents/plasticware, incubator used, and mitogen, if used. ▪ Understand when cell sorting is implicated and how to select for the appropriate cell (e.g., CD138+ sorting for multiple myeloma)

Objectives	Skills
Monitor cell growth and control variables	▪ Employ measures that will maintain optimal cell growth (e.g., feeding and centrifugation/concentration of specimens prior to culture to correct for depleted medium). ▪ Evaluate status of cultures using assessment of growth and mitotic activity, pH of medium, and turbidity. ▪ Identify and document probable causes of poor growth and culture failure, such as specimen inadequacy or equipment failure, and describe corrective actions taken. ▪ Report findings of culture failure or inadequate growth to laboratory personnel and, under supervision as needed, request new sample, if appropriate.
Use appropriate harvest procedures for specimen or culture	▪ Apply knowledge of cell cycle for various cell types and culture conditions (e.g., PHA stimulated lymphocytes, unstimulated leukemic cells, synchronized cultures) to timed harvests. ▪ Understand the use of synchronizing or intercalating agents, such as amethopterin, fluorodeoxyuridine, bromine deoxyuridine, ethidium bromide, or actinomycin D, at the appropriate concentration, temperature, and duration. ▪ Use spindle fiber inhibitor (e.g., Colcemid, Velban) at correct concentration, temperature, and duration. ▪ Use recommended procedures for removing cells from culture vessels. Use appropriate hypotonic solution (KCl or sodium citrate), at correct concentration, temperature, and duration. ▪ Use cell fixative (acetic acid/methanol) at correct concentration, temperature, and duration. ▪ Control mechanical damage to chromosomes by proper mixing, shaking, pipetting, centrifuging, or other handling of the cells. ▪ Record complete information for harvest of specimen including date, addition of spindle fiber inhibitor, intercalating or synchronizing agents, conditions used for harvest, and name of technologist processing the samples.
Prepare slides with analyzable metaphases	▪ Select method of slide preparation to optimize metaphase quality and spreading while minimizing debris (e.g., control variables such as wet or dry slides, air flow, humidity level and temperature to regulate slide drying rate). ▪ Evaluate quality of slides with phase contrast microscope and adjust variables, as necessary. ▪ Understand variables of slide aging and how they affect banding quality (e.g., temperatures, storage times, UV exposure or microwave). ▪ Use slide storage methods that best maintain chromosome quality for banding and staining procedures.

Objectives	Skills
Understand how to select banding and staining methods that permit identification of each chromosome pair, at an appropriate band level	▪ Perform G-banding pretreatment and staining methods according to laboratory protocols. ▪ Evaluate quality of stained slides with a microscope and adjust variables, as necessary. ▪ Understand the results for other specialized staining procedures, when needed, (e.g., DAPI/Distamycin A, AgNOR, etc.) and recognize the advantages and disadvantages of these methods. ▪ Understand the appropriate destaining method necessary for re-banding or re-staining of a previously banded/stained slide. ▪ For FISH analysis, select the appropriate type of specimen and probe type for both interphase and metaphase FISH analyses. ▪ Perform appropriate slide pretreatment, denaturation, dehydration, hybridization, and detection for interphase and metaphase FISH analyses on cultured and uncultured cells. ▪ Understand the use and interpretation of controls for FISH analysis. ▪ Troubleshoot unacceptable or unanalyzable results for all banding/staining procedures.
Choose appropriate method for DNA/RNA isolation	▪ Isolate DNA/RNA expediently and as may vary with specimen type and test requested. ▪ Choose appropriate solution (e.g., TE, water, etc.) and volumes for reconstitution of DNA/RNA. ▪ Practice measures that prevent cross-contamination between samples. ▪ Monitor automated extraction instruments for reagent carry-over. ▪ Employ proper techniques for storage of DNA/RNA samples.
Determine concentration of DNA/RNA	▪ Measure quality and/or quantity of nucleic acid prior to testing, when appropriate. Options to estimate concentration and determine quality of DNA/RNA include: ○ Spectrophotometry ○ Fluorimetry ○ Direct visualization by gel electrophoresis
Understand probable causes of poor or failed DNA/RNA isolation	▪ Identify, evaluate, and document probable causes of poor or failed DNA/RNA isolation, such as inadequate specimen or reagent failure. ▪ Document corrective actions taken to address suboptimal DNA/RNA isolations or yields, as appropriate.
Patient Care: Analytic Laboratory Skills	
Select suitable metaphases/interphase cells for karyotype analysis	▪ Select metaphases according to morphology, spreading, length, and banding detail. Assess difficulties in microscopic analysis and computer imaging posed by overlapping chromosomes, debris, poor stain, etc.

Objectives	Skills
Perform accurate microscopic counts and analyses of banded and non-banded chromosomes	▪ Analyze chromosomes and identify normal/abnormal karyotype. Document the analysis of distinct colonies on *in situ* amniotic fluid cultures. ▪ Document analysis in an organized manner (e.g., patient information, modal number, sex chromosome constitution, aberrant chromosomes, slide identification, Vernier coordinates or image number, technologist name, and date of work). ▪ Use a method that allows rapid retrieval of any cell analyzed (e.g., use of a calibrated microscope stage, micro-locator slide, conversion chart).
Prepare accurate karyotypes from computer images.	▪ Organize chromosomes according to a systematic and approved format (e.g., current ISCN). ▪ Produce electronic images with clarity and appropriate contrast.
Identify numerical and structural chromosome abnormalities, and relate their implications (e.g., phenotype and relationship to disease)	▪ Identify chromosomes by number, chromosome group, size, band number, banding intensity. ▪ Determine numerical abnormalities of the autosomes and sex chromosomes. ▪ Differentiate between the presence of multiple clones and/or subclones and random events identified during a conventional chromosome analysis. ▪ Identify structural abnormalities such as translocations, deletions, inversions, ring chromosomes, marker chromosomes, chromatic breaks, endoreduplication, etc. ▪ Be informed about chromosome heteromorphisms and be able to distinguish these variants from clinically relevant structural findings. ▪ Distinguish abnormalities that are culture artifacts from disease-associated abnormalities.
Fluorescent *in situ* hybridization (FISH) analysis	▪ Understand concepts of probe design and relationship to data interpretation. ▪ Select the appropriate specimen type, probe type, and interphase versus metaphase FISH assay. ▪ Determine the appropriate conditions for pretreatment, dehydration, hybridization, washing and counter-staining of FISH slides. ▪ Understand how to mark FFPE slides for FISH analysis. ▪ Understand whether probe panels or individual probes are more appropriate in each clinical circumstance. ▪ Recognize when FISH testing may be considered “STAT” (e.g., PML::RARA fusion in APL).
Perform accurate microscopic counts and analyses for FISH	▪ Recognize appropriate metaphases and/or interphase nuclei for FISH analysis. ▪ Recognize appropriate normal and abnormal signal patterns. ▪ Analyze metaphase and interphase cells using fluorescence microscopy. ▪ Document results of FISH evaluation appropriately. ▪ Understand how to determine and apply normal ranges or cut-off values for each probe/probe set.

Objectives	Skills
	▪ Document QA monitoring of FISH probes, including periodic correlation of results with those from orthologous testing, such as karyotype studies, NGS results, or sequence-based fusion testing.
Record equipment and identifiers (e.g., microscope; stage coordinates; image station; image numbers) for FISH analysis on selected cells	▪ Document analysis in an organized manner (e.g., patient information, slide identification, Vernier coordinates/image numbers, technologist name, date of analysis). ▪ Use a method that allows rapid retrieval of any cell analyzed (e.g., use of a calibrated microscope stage, micro-locator slide, conversion chart).
Prepare correct number of images	▪ Prepare correct number of FISH images as recommended by the ACMG/CAP guidelines.
Be familiar with microarray technical processing	▪ Process extracted nucleic acid in preparation for microarray hybridization. ▪ Determine the appropriate conditions for microarray hybridization and post-hybridization washing, dependent upon platform. ▪ Scan and analyze data. ▪ Archive the appropriate data.

Objectives	Skills
Know and understand principles and techniques associated with microarray analysis	▪ Use the appropriate software and databases for data analysis. ▪ Explain relationships between microarray and other genomic data (i.e., understand mechanisms underlying aberrations). ▪ Understand principles of detecting copy number variation and genotyping data from microarrays and other technologies such as next-generation sequencing. ▪ Understand the concepts, strengths and weaknesses of: o Intragenic and large multigenic copy number variants o Homozygosity stretches (cnLOH) in the genome indicative of IBD/UPD/cnLOH o Genotypes relevant to carrier screening o Concepts behind array probe design and relationship to data o Use of genome browsers to evaluate array designs and hybridization results o Reconciliation of microarray and exome/genome sequencing data ▪ Observe, perform or be familiar with the use of microarrays to identify single nucleotide variants, single-gene intragenic deletions and duplications, or chromosomal copy number variants. ▪ Understand principles of genotyping and its applications. ▪ Understand concepts of array probe design and relationship to data interpretation.
Know and understand principles and techniques associated with PCR analysis	▪ Understand the principles of qualitative and quantitative PCR. ▪ Understand the process of designing primers for PCR reactions and how to optimize. ▪ Determine components and concentrations for a particular reaction(s). ▪ Assemble reagents for master mix. ▪ Calculate primer dilutions. ▪ Optimize conditions for amplification. ▪ Troubleshoot failed or non-specific reactions.
Understand principles and techniques associated with gene scanning methodologies	▪ Observe, perform, or be familiar with methods for gene scanning, such as heteroduplex analysis and melting curve analysis.

Objectives	Skills
Understand principles and techniques associated with direct variant detection	▪ Understand a variety of methods for direct mutation (SNV and CNV) detection, such as: ○ Restriction fragment length polymorphism analysis ○ FRET analysis ○ Allele-specific oligonucleotide dot blot hybridization ○ Allele-specific PCR amplification (ARMS) ○ Pyrosequencing ○ Exon-focused array CGH ○ Molecular inversion probe ○ Multiplex ligation-dependent probe amplification (MLPA) ○ Single nucleotide extension ○ Mass Spectrometry (MassArray) ○ Droplet-digital PCR ○ TaqMan probes ○ Other
Know and understand principles and techniques associated with dideoxy sequencing of single genes or exons	▪ Be familiar with concepts behind Sanger dideoxy sequencing. ▪ Perform direct DNA sequencing. ▪ Understand technical pitfalls of Sanger sequencing (allele drop-out, etc.)
Know and understand principles and techniques associated with sequencing of single genes, gene panels, whole exomes, or whole genomes	▪ Understand the technical details of next-generation sequencing and limitations and advantages of different methods of library preparation and sequencing. ▪ Understand applications, challenges, limitations, and advantages of sequencing single genes, gene panels, exome, and the whole genome. ▪ Understand the assay design process, including selecting capture baits for hybridization or primers for microdroplet PCR. ▪ Understand the refinement steps of assay design to optimize analysis of difficult genomic regions, e.g., repetitive sequences or GC-rich sequences. ▪ Determine components and concentrations for library preparation and sequencing. ▪ Optimize conditions for library preparation and sequencing. ▪ Demonstrate proper documentation and validation of changes to NGS pipelines.

Objectives	Skills
Understand principles of exome and genome analysis	▪ Perform singleton and duo/trio analyses of exome or whole genome sequencing data. ▪ Apply principles of homozygosity mapping and search for recessive disease mutations. ▪ Apply principles of using phenotype information to isolate gene lists for analysis. ▪ Apply modeling inheritance modes (dominant, recessive, X-linked) based on pedigree of tested individual and create priority gene lists for analysis. ▪ Create and use virtual gene panels for analysis based on disease phenotype information. ▪ Understand limitations of sequence depth coverage and implications for diagnostic testing. ▪ Use workflow for analyzing and reporting incidental findings. ▪ Understand principles of exome and whole genome re-analysis and re-interpretation.
Know and understand principles and techniques associated with identity testing	▪ Perform identity testing using the analysis of polymorphic genetic markers (NOT gene mutations associated with disease), e.g., ○ Parentage testing ○ Forensics ○ Zygosity ○ Maternal cell contamination ○ Transplantation ○ Uniparental disomy
Patient Care: Post-analytic laboratory skills	
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 (PHI). ▪ Understand the informatics processes that connect sample requisition to wet lab processes, data analysis, report writing, and transmission of final reports to referring physicians. ▪ Understand processes associated with exome and genome data storage in the electronic health record (EHR).

Objectives	Skills
Variant calling	▪ Learn to use software for next-generation sequencing for reading alignment, variant calling, and confirmations. ▪ Use and understand genome sequence alignment software (e.g., BWA, DRAGEN). ▪ Know how to visualize single nucleotide variants and copy number variation. ▪ Understand and use variant calling algorithms (e.g., GATK, DRAGEN). ▪ Identify artifacts and trouble spots in genome sequence data. ▪ Understand biostatistical analysis of sequencing data (depth of coverage, read quality, Q cores, mapping quality, etc.). ▪ Analyze and integrate data from orthogonal confirmation methods.
Analysis of genomic sequence data	▪ Observe and understand how to use: ○ Genome browsers (e.g., UCSC, IGV, ENSEMBL) ○ Human genome variation databases (e.g., ClinVar, 1000Genomes, ExAC, gnomAD, DGV, DECIPHER) ○ Variant analysis software, if available (custom software or vendor software, e.g., Alamut, Ingenuity, Agilent Cartagenia, Affymetrix CytoScan HD, etc.) ○ Basic biostatistical concepts – case-control studies, odds ratios, use of different statistical measurements, outcomes of population studies, variant allele frequencies
Variant interpretation: SNV	▪ Use ACMG/AMP/ASCO guidelines to interpret sequence variants. ▪ Understand how and when to use ClinGen Consortium variant interpretation specification guidelines. ▪ Pull together relevant evidence from genomics databases, published literature on case studies and functional analyses, clinician-provided phenotype information, and *in silico* algorithms to classify variant using the relevant classification guidelines. ▪ Understand how to use *in silico* algorithms for prediction of effects of missense changes and evolutionary conservation (PolyPhen, SIFT, REVEL, GERP, PhyloP, etc.). ▪ Use ClinVar and other databases to review classifications from other sources and consider reasons for any discordance.
Variant interpretation: CNV	▪ Use appropriate databases to help classify chromosomal copy number variants (e.g., UCSC Genome Browser, Database for Genomic Variants, DECIPHER, ClinGen, gnomAD). ▪ Use ACMG/AMP/CAP/ASCO guidelines to interpret copy number variants. ▪ Collect relevant evidence from genomics databases, published literature on case studies, and clinician-provided phenotype information to classify microarray results using the ACMG recommendations, where relevant. ▪ Apply principles of chromosomal structure, rearrangement, and meiotic/mitotic behavior to interpret chromosomal imbalances.

Objectives	Skills
Summarizing results	▪ Correctly interpret results of all laboratory assays to determine normal/affected/carrier status. ▪ Correlate results with other laboratory results and/or clinical information to develop an appropriate interpretation of the laboratory results.
Patient Care: Reports	
Explain the results and report to the appropriate provider	▪ Draft a comprehensive, accurate report using standard nomenclature, summarizing the findings in understandable text, and incorporating the patient identification, and all relevant clinical and laboratory data. ▪ Document oral and preliminary reports on final written report when applicable. ▪ Recognize and avoid difficulties or hazards of oral reporting of results. ▪ Report on the need for additional studies to complete the diagnosis, when appropriate. ▪ Understand how bioinformatics pipelines can be used to prepare primary reports and issue amended reports. ▪ Use proper nomenclature as standardized by HGVS to describe molecular results and ISCN to describe cytogenetic results. ▪ Understand when and how to amend or addend a report.

Objectives	Skills
Medical Knowledge: Foundations of Medical Genetics and Genomics	
Understand principles of cell biology, molecular biology, and genetics	▪ Describe cell structure and function. ▪ Summarize the stages of the cell cycle, and of mitosis and meiosis (both spermatogenesis and oogenesis). ▪ Describe DNA structure (base sequence, complementarity, repetitive vs. unique sequence, gene structure, etc.), function (genetic code, replication, transcription and translation, and mutations), and chromosome ultrastructure (telomeres, centromeres, nucleosomes, histones, loop domains, scaffolding, DNA packaging, etc.). ▪ Review basic embryology and the origin of various tissues, such as blood, skin, CVS, and amniotic fluid. ▪ Describe basic principles of inheritance (dominant or recessive, autosomal or sex linked, multifactorial, polygenic, Lyon hypothesis, imprinting, trinucleotide repeat, polygenic etc.). ▪ Describe mutagenicity and principles of genetic toxicology. ▪ Describe etiology of specific chromosomal abnormalities, such as anaphase lag, non-disjunction, dispermy, breakage and repair, uniparental disomy, and understand the influences on these processes by variables such as maternal age, clastogens, inherited breakage syndromes, and imprinting. ▪ Understand basic principles of genetic counseling, including pedigree analysis and risk calculations for inherited conditions. ▪ Discuss basic principles of cancer cytogenetics, including hematopoiesis, clonal evolution, disease remission, and relapse. ▪ Be familiar with clinical features of common constitutional and acquired genetic disorders including those caused by aneuploidies, microdeletion/microduplication, SNVs, and cardinal genetic features of hematologic neoplasms and solid tumors. ▪ Understand intragenic and large multi-genic copy number variants and mechanisms for their formation. ▪ Understand the mechanisms for and implications of large regions of homozygosity in the genome (i.e., UPD, consanguinity, IBD). ▪ Explain transcription, splicing, translation, and variation of gene expression between tissues. ▪ Explain mechanisms of pathogenicity at cellular level (dominant-negative, recessive). ▪ Describe different classes of mutations (e.g., missense, nonsense, deletion, insertion, splice-site, triplet repeat expansion). ▪ Perform Bayesian risk analysis. ▪ Describe risk factors for mutations (advanced maternal age and nondisjunction, advanced paternal age and new autosomal dominant mutations, mutagens and carcinogens).

Objectives	Skills
Understand principles and applications of population genetics	▪ Be familiar with various types of screening assays (such as carrier screening, newborn screening, cell-free fetal DNA screening) and understand how they differ from diagnostic assays. ▪ Understands concepts of heritability, inheritance patterns, variability, heterogeneity, penetrance, and the epidemiology/natural history of a condition. ▪ Explain genetics concepts clearly and be able to identify family members at risk. ▪ Ability to use Bayesian and Hardy-Weinberg for risk reduction calculation. ▪ Understand the principles of incomplete penetrance and variable expressivity.
Interpersonal and Communication Skills	
Ability to communicate effectively with colleagues	▪ Maintain comprehensive, accurate, timely, and legible medical records. ▪ Communicate appropriate information to health professionals one-on-one or in group settings, under clinical supervision, as appropriate. ▪ Communicate results verbally to ordering provider, his or her designee, or genetic counselor, as appropriate. ▪ Understand and adhere to HIPAA guidelines. ▪ Document all communication with providers, particularly as it relates to laboratory testing plan.
Consistently maintain appropriate ethical and professional standards	▪ Demonstrate an attitude of responsibility and respect for the patient, a respectful and cooperative attitude toward professional colleagues, and an honest, forthright manner in conducting professional tasks.
Learn how to teach and supervise effectively	▪ Educate students or junior trainees, mentor, assess progress and skills, and provide appropriate feedback and appraisal (oral and written).
Practice-Based Learning and Improvement	
Know how to keep up to date in common clinical genetics topics.	▪ Thoroughly research topics when needed. ▪ Critiques research evidence for applicability to laboratory practice. ▪ Apply new skills or knowledge to laboratory service. ▪ Recognize the importance of Continuing Certification Programs.
Receiving and incorporating feedback	▪ Seek feedback from others and exhibit willingness to change and to adapt. ▪ Be receptive to feedback received from peers and mentors. ▪ Change practice behaviors in response to feedback from others and review of own practice.
Professionalism	

Objectives	Skills
Practices within ability and recognize limits of one's abilities	▪ Seek consultation, when appropriate. ▪ Exercise authority accorded by 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	▪ 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 teach and supervise effectively.	▪ 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.
Systems-Based Practice	
Knowledge of evidence-based guidelines and appropriate billing	▪ Understand how to determine operating costs and cost components of tests. ▪ Understand how laboratory test reimbursement works. ▪ Provide cost-conscious services. ▪ Consider the costs and benefits of the test. ▪ Follow accepted laboratory 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.

Objectives	Skills
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 referring healthcare provider.
Knows how to provide comprehensive and integrated service	▪ Coordinate services with other providers, 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 sample 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.
Perform PCR to minimize carryover (false positive results)	▪ Utilize unidirectional workflow. ▪ Utilize adequate physical separation of pre- and post-amplification samples to avoid amplicon contamination. ▪ Change gloves frequently during processing. ▪ Use dedicated pipettes (positive displacement type or with aerosol barrier tips). ▪ Manipulations must minimize aerosolization. ▪ Monitor liquid handlers to eliminate carry over.
Sample storage	▪ Select slide cleaning and storage methods that maintain quality of chromosome preparations for the period required by regulatory agencies. ▪ Understand methods of nucleic acid storage and follow retention policies, as required by regulatory agencies.
Assay Controls	▪ Understand the purpose of using appropriate positive, negative, and no-template controls when performing and interpreting laboratory testing.
Next-Generation sequencing quality metrics	▪ Understand a variety of quality metrics specific to next-generation sequencing, including depth of coverage, Qscores, ROI coverage, GC bias.

Objectives	Skills
Laboratory Accreditation	▪ Be familiar with the requirements of regulatory agencies such as 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.
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 labs must follow with respect to the type and frequency of PT that must be performed.
Sample ID testing	▪ Be familiar with the concept of intra-laboratory sample identity testing for quality purposes and how to manage possible incidental findings associated with such testing.
Safety	
Laboratory and data safety	▪ Complete institutional safety training and be familiar with safety protocols pertaining to both laboratory and patient safety practices. ▪ Identify specimen and reagent disposal needs in compliance with safety, chemical, and biosafety guidelines. ▪ Identify personal protection practices (gloves, gowns, eyewear) and equipment indicated for processing of lab specimens/reagents. ▪ Identify cybersecurity risks and follow practices to minimize potential breaches.