American Board of Medical Genetics and Genomics

Laboratory Genetics and Genomics 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 that 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		
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 type. Identify factors important for the transport of specimens, such as overnight delivery, appropriate transport media and containers, or recommended temperatures. Transport/ship specimens off-site using packaging that meets OSHA guidelines. Be aware of appropriate specimen and handling requirements.
	Assess acceptability of specimen for study	Check for appropriate labeling of specimen and requisition with at least two identifiers. Evaluate suitability of specimen for requested study, both for tissue type and amount/volume required. Judge quality of specimen. Assess for presence of interfering substances i.e.: presence of blood in amniotic fluid, blood clots or hemolysis in peripheral blood samples, syringe number and 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, etc. Describe methods for possible recovery of poor samples. Notify appropriate individuals of unsatisfactory samples and document such notification per laboratory policy and regulatory requirements.
	Accession specimen	Assign unique laboratory accession number to specimen. 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.

DOMAIN	OBJECTIVES	SKILLS
		Record notes of any special test requests, particularly those requiring transport of samples to other laboratories. Record chromosomal region of interest in high-resolution chromosomal studies, FISH, or DNA microarray and gene/sequence of interest for molecular studies.
	Tracking of specimen	Follow protocols to ensure proper identification of patient materials through the complete process, from accession through final report. Be able to track specimen through all aspects of the testing process.
	Appropriate documentation	Maintain necessary records and laboratory database, in logbooks or computers, as appropriate.
Appropriate culture techniques for submitted specimens	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.
	Prepare appropriate media for specimens, giving consideration to the clinical indication for the study	Choose appropriate medium 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 reagents.
	Employ appropriate culture techniques for specimen	Select culture equipment and vessels for closed or open culture systems. Select culture technique for specimen taking into account 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 solutions used in the procedures 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, incubator used, and mitogen, if used.
	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 inadequate specimens, or equipment failure, and describe corrective actions taken. Report findings of culture failure or growth inadequate for analysis to laboratory personnel, and, under supervision as needed, request new sample, if appropriate.
Principles and techniques for harvesting specimens or cell cultures	Determine optimal time sequence and method for harvest (manual or robotic)	Apply knowledge of cell cycle for various cell types and culture conditions (e.g., PHA stimulated lymphocytes, unstimulated leukemic cells, synchronized cultures) to time harvests.
	Use appropriate harvest procedures for specimen or culture	Understand the use synchronizing or intercalating agents, such as amethopterin, fluorodeoxyuridine, bromine deoxyuridine, ethidium bromide, or actinomycin D, at appropriate concentration, temperature, and duration. Use spindle fiber inhibitor (e.g., Colcemid, Velban) at correct concentration, temperature, and duration. Use recommended procedure 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.

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		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 that will produce high quality metaphases with optimum spreading (e.g., control variables such as wet or dry slides, air flow, humidity level and temperature to regulate slide drying rate). Employ techniques that control concentration and distribution of cellular and other debris on slides. Evaluate quality of slides with phase contrast microscope and adjust variables as necessary. Describe, document processes for determining acceptability for analysis. Employ techniques that control the aging of slides to produce optimal banding conditions (e.g., storing at various temperatures, such as 37°C, 60°C, 90°C, for various times, such as 20 min to 2 hours, or overnight; UV exposure or microwave). Use slide storage methods that best maintain chromosome quality for banding and staining procedures, with protection from humidity, light, chemicals, or mechanical damage.
Principles and techniques of chromosome banding and staining	Understand how to select banding and staining methods that permit identification of each chromosome pair, at an appropriate band level	Use G-banding pretreatment and staining methods, utilizing trypsin, 2XSSC, urea, or other chemicals. Evaluate quality of stained slides with phase contrast microscope and adjust variable as necessary. Understand the results for other specialized staining procedures when needed (e.g., DAPI/Distamycin A, sister chromatid exchange, etc.) and recognize the advantages and disadvantages of these methods.
	Select mounting materials, hydration/dehydration methods, and destaining techniques when necessary for multiple staining procedures on the same slide	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 both directly and indirectly labeled probes for interphase and metaphase FISH analyses on cultured and uncultured cells. Understand the use and interpretation of controls for FISH analysis.
	Select slide cleaning and storage methods that maintain quality of chromosome preparations for period of time required by regulatory agencies.	
	Troubleshoot unacceptable or unanalyzable results for all banding/staining procedures	
Maintenance and use of microscopes and computer-generated imaging techniques and equipment	Operate a standard compound microscope, inverted microscope, stereo microscope, and computerized karyotype equipment.	Clean, adjust, focus, and use appropriate illumination systems, eyepieces, objectives, condenser systems and filters, for bright field, fluorescent, and phase contrast microscopes. Operate microscopes and computerized image capture system equipment for optimal resolution of specimen. Demonstrate appropriate use of coverslips and immersion oil. Maintain computer image analysis equipment in optimal working order.
	Select methods that produce optimal chromosome images	Produce electronic images with clarity and appropriate contrast.

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Chromosome analysis	Select suitable metaphases/interphase cells for 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.
	Perform accurate microscopic counts and analyses of banded and non-banded chromosomes	Analyze chromosomes at the microscope and identify normal/abnormal karyotype. Document the analysis of distinct colonies on *in situ* amniotic fluid cultures.
	Record microscope identification, stage coordinates, and cell analysis data on all cells selected	Document analysis in an organized manner (e.g., patient information, modal number, sex chromosome constitution, aberrant chromosomes, slide identification, notation of Vernier coordinates, technologist name, and date of work). Use a method that allows rapid retrieval of any cell analyzed, on the same or another microscope, (e.g., use of a calibrated microscope stage, microlocator slide, conversion chart).
	Prepare accurate karyotypes from computer images.	Organize chromosomes according to a systematic and approved format (e.g., ISCN).
	Identify numerical and structural chromosome abnormalities, and relate their implications (e.g., phenotype and relationship to disease)	Determine numerical abnormalities of the autosomes and sex chromosomes. Differentiate between the presence of multiple cell lines, and random gain or loss of chromosomes in slide preparation of specimens and /or controls. Identify constitutional structural abnormalities such as translocations, deletions, inversions, ring chromosomes, isochromosomes, and fragile sites. Identity sporadic structural abnormalities such as chromatid breaks, chromatid exchanges, fragments, and endo-reduplication. Understand how to identify heteromorphic chromosomes with different variable regions, by band number, code letters, size, and banding intensity.
FISH Analysis	Select suitable metaphases/interphase cells for analysis Select appropriate regions of interest from paraffin sections	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 procedures for selection of areas for FISH analyses on FFPE slides. Understand when probe panels or individual probes are more appropriate in a given clinical circumstance. Recognize when FISH testing may be considered “STAT” (e.g., PML/RARA fusion in APML)
	Perform accurate microscopic counts and analyses	Recognize appropriate metaphases and/or interphase nuclei for FISH analysis. Recognize appropriate normal and abnormal signal patterns. Count the appropriate signal(s) and/or evaluate signal patterns. Document results of FISH evaluation appropriately. Understand the normal range or cut-off values for each probe / probe set. 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 microscope identification, stage coordinates, and analysis on selected cells	Document analysis in an organized manner (e.g., patient information, slide identification, notation of Vernier coordinates, technologist name, date of analysis). Use a method that allows rapid retrieval of any cell analyzed, on the same or another microscope, (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 ACMGG/CAP guidelines

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Microarray analysis	DNA extraction and purity	Extract DNA and determine purity and concentration. Be able to identify suboptimal specimens. Determine the appropriate amount of DNA necessary for microarray analysis, dependent upon platform type.
	DNA labeling of target and control samples	Fluorescently label DNA necessary for microarray analysis. Determine the specific activity and the yield.
	Microarray hybridization and washing	Determine the appropriate conditions for microarray hybridization and post-hybridization washing, dependent upon platform. Scan and analyze the data as per platform. Archive the appropriate data.
	Microarray data analysis	Use the appropriate software for data analysis, as well as proper use of the appropriate databases (e.g., UCSC Genome Browser, Database for Genomic Variants, DECIPHER, etc.). Explain relationships between microarray copy number and karyotype data. Perform relevant follow-up chromosomal studies to correlate with array findings. Understand principles of detecting copy number variation and genotyping data from microarrays and next-generation sequencing. • Intragenic and large multi-genic copy number variants • Homozygosity stretches in the genome indicative of IBD/UPD • Genotypes relevant to carrier screening • Concepts behind array probe design and relationship to data • Use of genome browsers to evaluate array designs and hybridization results • Reconciliation of microarray and exome/genome sequencing data
Appropriate techniques for nucleic acid isolation from submitted specimens	Use of techniques	Use Universal Precautions for protection against real or 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.
	Choose appropriate method for DNA/RNA isolation	Isolate DNA/RNA expediently, with consideration to specimen type and test requested. Choose appropriate type of solution (e.g., TE, water, etc.) for reconstitution of DNA/RNA. Choose appropriate amount of reconstitution solution for test being performed. Practice measures that prevent cross-contamination between samples. Monitor automated extraction instruments for reagent carry-over.
	Determine concentration of DNA/RNA, as appropriate	Options to estimate concentration and determine quality of DNA/RNA include: • spectrophotometry (determine optical density and nucleic acid/protein ratio of reconstituted DNA/RNA) • fluorimetry (estimate DNA concentration) • 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.
	Storage of DNA/RNA samples appropriately	Employ proper techniques for storage of DNA/RNA samples.

DOMAIN	OBJECTIVES	SKILLS
Principles and techniques for polymerase chain reaction (PCR).	Know and understand principles and techniques associated with PCR analysis	Understand the principles of qualitative and quantitative PCR. 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. Utilize appropriate controls. Design an ongoing CQI (Continuous Quality Improvement) program for PCR based assays
	Know which primers are appropriate for disease/area of concern	Perform or be familiar with the development and design of new primers.
	Perform PCR to minimize carry over (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. "No template" controls in which target DNA is omitted (no product is expected) should be included in each run. Monitor liquid handlers to eliminate carry over.
Southern analysis	Understand principles and techniques associated with Southern blot	Understand aspects of the Southern blot procedure including: • digestion of DNA with appropriate restriction enzymes; • electrophoresis; • denaturation in alkali and transfer DNA to membrane; • preparation of probe (radioactive or chemiluminiscent label); • denaturation of probe and hybridization of membrane; • exposure to X-ray film and development of the autoradiograph.
Targeted mutation analysis	Understand principles and techniques associated with direct mutation detection	Understand a variety of methods for direct mutation detection, e.g., • restriction fragment length polymorphism analysis; • FRET analysis (Invader); • 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)
Gene scanning	Understand principles and techniques associated with gene scanning	Observe, perform, or be familiar with methods for gene scanning, e.g., • heteroduplex analysis; • melting curve analysis • MLPA
Sanger (dideoxy) sequencing	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.

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Next-generation sequencing	Know and understand principles and techniques associated with sequencing of gene panels, whole exomes, or whole genomes	Understand the technical details of next-generation sequencing – limitations and advantages of different methods of library preparation and sequencing. Understand applications, challenges, limitations, and advantages of sequencing 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., at repetitive sequences or GC-rich sequences. Determine components and concentrations for library preparation and sequencing. Optimize conditions for library preparation and sequencing. Troubleshoot failed or non-specific reactions. Utilize appropriate controls. Understand new test validation approaches for NGS-based tests and establishment of Qscores Demonstrate proper documentation and validation of changes to NGS pipelines
	Understand principles of exome or whole-genome analysis	Perform singleton and trio analyses of exome sequence 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 reinterpretation
Quantitative PCR	Know and understand principles and techniques associated with quantitative PCR	Observe, perform or be familiar with the use of quantitative PCR to quantify gene dosage and to assay for mutations and single nucleotide polymorphisms.
Array Analysis	Know and understand principles and techniques associated with microarray analysis	Observe, perform or be familiar with the use of microarrays in the laboratory to identify single nucleotide variants, single-gene intragenic deletions and duplications, or chromosomal copy number variants. Understand principles of genotyping and its applications, including carrier screening. Understand concepts of array probe design and relationship to data.
Identity testing	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., • paternity testing • forensics • zygosity • transplantation • maternal cell contamination
General laboratory skills, quality control, and quality assurance	Know how to prepare reagents	Prepare reagents at the proper concentration and pH, with proper labeling and dating, using required grades of water and chemicals.
	Know how to select, operate, clean, and maintain all	Monitor the need for service or repair on any equipment and report this to appropriate authority. Document usage of CO_2 tanks and how to replace when needed. Record equipment temperatures with reference thermometers and adjust controls if necessary.

DOMAIN	OBJECTIVES	SKILLS
	laboratory equipment and instruments, as appropriate	Monitor centrifuge speed using a tachometer and know how to adjust, if necessary. Be aware of regulatory requirements for preventative maintenance of equipment and documentation of equipment repairs. Be aware of the need for regular instrument function checks and how this is documented in the laboratory.
	Understand principles of sterilization and decontamination procedures	Able to use disinfectants, steam, dry heat, gas, U.V. irradiation, and membrane filtration appropriately.
	Understand how to stock laboratory supplies and chemicals	Maintain adequate stocks of laboratory supplies and chemicals. Employ limits on stock usage imposed by shelf life and expiration dates.
	Practice established procedures for general laboratory safety	Employ appropriate cleaning procedures for laboratory glassware and instruments. Use Universal Precautions as established by Centers for Disease Control (CDC) and individual state or local governments. Use appropriate procedures for laboratory emergencies (e.g., fire, accidental injury, natural disaster, chemical spill, or power failure). Use correct procedures for storage, handling, and disposal of different types of materials and waste: biological and chemical, volatile or stable; radioactive; sharps and glass.
	Maintain a system to ensure laboratory quality control in all areas, to comply with all regulatory requirements	Maintain a system to: (1) ensure accuracy of chromosomal results, including appropriate documentation, throughout all steps of laboratory procedures; (2) ensure accuracy of molecular tests, including appropriate documentation, throughout all steps of laboratory procedures; (3) ensure confidentiality and security of patient records; (4) appropriately label, store, and monitor shelf life, sterility, and quality of all media, sera, reagents and chemicals. Maintain an easily accessible collection of current Material Safety Data Sheets (MSDS) for all chemicals used in the laboratory procedures. Maintain a system of records for equipment and instruments (serial numbers, date of purchase, maintenance checks, gauge readings, dates and type of service or repair). Practice the techniques, procedures and policies used in the laboratory, as documented in the laboratory manual. Assist in reviews and revising the laboratory manual. Participate in laboratory proficiency testing, as appropriate.
Bioinformatics	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 to collect information from multiple individuals in the process between sample accessioning to final report. • Understand processes associated with exome and genome data storage in EHR
	Variant calling	Learn to use software for next-generation sequencing for read alignment, variant calling, and confirmations.

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		• Genome sequence alignment software (e.g., BWA) • Variant calling algorithms (e.g., GATK) • Identifying artifacts and trouble spots in genome sequence data • Visualizing single nucleotide and copy number variation • Basic biostatistical analysis of sequencing data (depth of coverage, read quality, Q cores, mapping quality, etc.) • Analyzing and integrating data from orthogonal confirmation methods • Understand the utility of and how to use in silico prediction algorithms (e.g., Polyphen, SIFT, splice site predictors)
	Analysis of genomic sequence data	Observe and understand how to use: • Genome browsers (UCSC, IGV, ENSEMBL) • Human genome variation databases (e.g., ClinVar, 1000Genomes, ExAc, DGV, DECIPHER) • Variant analysis software, if available (custom software or vendor software, e.g., Alamut, Ingenuity, Agilent Cartagenia, Affymetrix CytoScan HD, etc.) • in silico algorithms for prediction of effects of missense changes and evolutionary conservation (PolyPhen, SIFT, GERP, PhyloP, etc.) Understand basic biostatistical concepts – case-control studies, odds ratios, use of different statistical measurements, outcomes of population studies.
Post-analytic laboratory skills	Molecular variant interpretation	Use ACMG/AMP guidelines to interpret sequence variants 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 ACMG 5-tier system Use ClinVar and other databases to review classifications from other sources and consider reasons for any discordance Use proper HGVS nomenclature
	Cytogenetic variant interpretation	Use ACMG guidelines to interpret chromosomal copy number variants Pull together relevant evidence from genomics databases, published literature on case studies, and clinician-provided phenotype information to classify microarray results using the ACMG 5-tier system, where relevant. Apply principles of chromosomal structure, rearrangement, and meiotic/mitotic behavior to interpret abnormal chromosomal imbalances detected by FISH/karyotype Use proper ISCN nomenclature
	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.
	Explain the results and report to the appropriate authority	Recognize and avoid hazards implied in oral reporting of results. Draft a neat, accurate report using standard nomenclature, summarizing the findings in understandable text and incorporating the patient identification, and all relevant clinical and laboratory data; forward to the appropriate individual for review and signature. Document oral and preliminary reports on final written report.
	Understand additional studies needed to make a diagnosis	Report the need for additional studies to complete the diagnosis: repeat the culture, perform additional staining techniques, analyze other tissues, request family studies, FISH, microarray, DNA extraction from a new sample, confirmation by other molecular method, RNA analysis, biochemical studies, etc.

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	Report	Prepare and generate reports (including pre-written and *de novo* results and interpretations) incorporating all relevant clinical and laboratory data. Understand how bioinformatics pipelines can be used to prepare primary reports and issue amended reports Use nomenclature as standardized by HGVS to describe molecular results or ISCN to describe cytogenetic results.
	Communication	Communicate results verbally to ordering physician, his or her designee, or genetic counselor as appropriate. Understand and adhere to HIPAA guidelines.
2. Genetics Knowledge		
General principles of biology and genetics	Understand principles of general biology and genetics that relate to cytogenetics	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, etc.), and function (genetic code, replication, transcription and translation, and mutations) chromosome ultrastructure: telomeres, centromeres, nucleosomes, histones, loop domains, scaffolding, DNA packing, etc. Review basic embryology and the origin of various tissues: 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. Understand genome structure – gene structure, low-copy and high-copy repeat sequences, chromosomal structure, unstable regions, conserved regions.
	Understand principles of clinical cytogenetics	Describe etiology of chromosomal abnormalities such as anaphase lag, non-disjunction, dispermy, breakage and repair, uniparental disomy, and the influence of these processes of maternal age effect, 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. Correlate molecular genetic results with cytogenetics for prenatal diagnosis, family studies, and cancer cytogenetics. Be familiar with clinical features of common constitutional and acquired cytogenetic disorders including aneuploidies, microdeletion syndromes, chromosome breakage syndromes, and 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 indicative of uniparental disomy, consanguinity, and identity by descent.
	Understand principles of general biology and genetics that relate to molecular genetics	Understand DNA structure (base sequence, pairing, replication and packaging into chromosomes). Explain transcription, splicing, translation, and variation of gene expression between tissues. Explain genomic organization and gene structure. Understand core technologies for allele discrimination and mutation detection.

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	Understand principles of molecular genetics	Explain mode of inheritance at level of organism (dominant, co-dominant, recessive, autosomal, sex-linked, multifactorial, polygenic, inheritance of imprinted genes). Explain action of gene at cellular level (dominant-negative, recessive). Describe different classes of mutations (e.g. missense, nonsense, deletion, insertion, splice-site, triplet repeat expansion). Explain gene expression at cellular level (dominant, dominant-negative, or negative). Discuss basic principles of genetic counseling including pedigree analysis. 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). Correlate molecular genetic results with cytogenetic results for prenatal diagnosis, family studies, and cancer diagnostics or cancer risk assessment and any other pre-analytic clinical information.
3. Interpersonal and Communication Skills		
Inheritance/risk counseling	Understands concepts of heritability, inheritance patterns, variability, heterogeneity, penetrance and the epidemiology/natural history of a condition.	Transmit pertinent information in a comprehensible way. Explain genetics concepts and identify family members at risk.
Professional communication	Know how to communicate with colleagues	Maintain comprehensive, timely and legible medical records. Communicate appropriate information to health professionals one-on-one or in groups.
	Exhibit appropriate ethnical and professional standards at all times	Demonstrate an attitude of responsibility and respect to the patient, a respectful and cooperative attitude toward professional colleagues and an honest, forthright manner in carrying out professional tasks.
	Know how to teach and supervise	Educate, mentor, assess progress and skills, and provide appropriate feedback and appraisal.
4. Practice-Based Learning and Improvement		
Ongoing learning	Know how to keep up to date in common clinical cytogenetics topics.	Seek feedback from others; can research topics when needed; critiques research evidence for applicability to laboratory practice; uses bioinformatics resources. Be receptive to feedback. Participate in ABMGG Continuing Certification.
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 laboratory service. Exhibit willingness to change and to adapt.
5. Professionalism		
Responsibility	Understand the responsibility to the patient/family	Complete the tasks required to provide laboratory services effectively in a careful and thorough manner.

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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.
Patient diversity	Recognize differences (cultural, educational, etc.)	Recognize each patient's unique needs and characteristics. Provide equitable services regardless of patient culture or socioeconomic status. Is 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. Knowledgeable about the elements of informed consent, privacy, confidentiality, duty to warn, and is HIPAA compliant.	Take responsibility for actions; admits mistakes; tries to address ethical dilemmas and conflicts of interest. Demonstrate a commitment to ethical principles pertaining to (1) patient privacy and autonomy, (2) the provision or withholding of test results, (3) confidentiality of patient information, (4) informed consent, (5) conflict of interest, and (6) business practices that are in conflict with stated principles of professionalism.
Health professional relationships	Know how to interact with health professionals	Courteous and respectful when relating with peers and referring healthcare providers.
Leadership	Know teamwork and leadership skills. Knows how to teach and supervise.	Provide direction to staff. Educates and mentors, can assess progress and skills and provide appropriate feedback and appraisal.
6. Systems-Based Practice		
Service coordination	Know how to provide comprehensive and integrated service	Coordinate services with other providers, specialty clinics; provide timely service.
Evidence-based medicine	Knowledge of evidence-based guidelines and appropriate billing	Determine cost and cost components of tests and understand reimbursement issues; provide cost-conscious services; consider costs & benefits of test; follows accepted laboratory guidelines; uses appropriate billing codes.
	Understand research principles	Critically read and interpret scientific publications. Be aware of policy implications.
Health services	Understand system resource utilization; understand different healthcare delivery systems and medical practices.	Interface with laboratory information systems, electronic health records, and billing systems.
	Information access	Conduct literature review and database searches. Identify resources for the patient/family and referring healthcare provider.