

LABORATORY GENETICS AND GENOMICS

GOALS AND OBJECTIVES

Patient care:

- To gather relevant clinical information from the patient chart to aid in interpretation of cytogenetics results
- To learn to integrate cytogenetics into patient care
- To learn to correlate morphologic and clinical history with test results
- To understand ethical and clinical implications of genetic testing
- To participate in the development of new diagnostic assays to enhance patient care, if applicable

Medical knowledge:

- To understand the principles and concepts of chromosome biology and cytogenetics techniques for diagnostic care
- To develop an investigatory and analytic thinking approach to clinical diagnostics

Interpersonal and communication skills:

- To use effective writing skills in preparing complex interpretive reports including use of proper cytogenetic nomenclature
- To develop effective oral communication and presentation skills by providing a continuing education presentation to laboratory personnel
- To develop leadership and teaching skills through interactions with residents and laboratory staff
- To develop proficiency in presentation of cytogenetic findings to pathologists, medical students, and clinicians including monthly Genetics Laboratory conference

Professionalism:

- To demonstrate respect and compassion for patients
- To complete written reports in a timely fashion
- To work effectively as a team player and treat fellow colleagues, technical and administrative staff with respect
- To enhance professional development by keeping abreast of new developments in cytogenetics
- To demonstrate respect, compassion, and integrity in observing and participating in counseling encounters in the VUMC Pediatrics-Genetics Division
- To demonstrate understanding of ethical issues involved in genetic testing

Systems-based practice:

- To understand the importance of proficiency testing in the Cytogenetics and understand how it is measured

- To evaluate in a critical manner the need for genetic testing
- To understand quality control and quality assurance issues in clinical diagnostics
- To understand the implications of clinical diagnosis as it relates to patient insurability
- To understand the managerial aspects of the clinical diagnostics laboratories, focusing on cost-effectiveness of testing and the clinical utility of an assay

Practice-based learning

- To use case-based learning as a tool for additional insight into the genetic basis of disease pathogenesis
- To locate, appraise and assimilate pertinent evidence from scientific studies and genomic databases
- To demonstrate effective problem-solving skills, using a wide variety of information resources

Molecular Diagnostics

Goals:

The goal of this fellowship is to understand the pathogenesis of a variety of common inherited diseases, recognize the utility of molecular profiling of acquired diseases for diagnosis and treatment and appreciate pharmacogenetics testing to enable precision medicine for appropriate drug therapy and dosing. The trainee will obtain extensive knowledge in laboratory techniques, results interpretation, and management skills in preparation as a Laboratory Director for a high complexity molecular diagnostics lab.

Objectives:

Patient care:

- To learn how molecular diagnostics testing is integrated into patient care
- To learn to correlate morphologic and clinical findings with DNA results
- To understand ethical and clinical implications of genetic testing

Medical knowledge:

- To understand laboratory assays used in molecular diagnostics
- To learn the specificity, sensitivity, limitations, and interpretations of each assay
- To understand the pathogenesis of inherited and acquired diseases
- To develop an investigatory and analytic thinking approach to molecular diagnostics

Interpersonal and communication skills:

- To use effective writing skills in preparing complex interpretive and NGS reports
- To develop effective oral communication and presentation skills by providing continuing education presentation to laboratory personnel, interesting case presentation to genetics professionals and 1 hour laboratory medicine rounds to CP faculty and residents
- To develop leadership and teaching skills through daily interactions with residents and laboratory staff

Professionalism:

- To demonstrate respect and compassion for patients
- To complete all tasks in an accurate, precise and timely manner
- To work effectively as a team player and treat fellow colleagues, technical and administrative staff with respect

- To enhance professional development by keeping abreast of new developments in molecular diagnostics

Systems-based practice:

- To understand the importance of proficiency testing in the Molecular Diagnostics lab and understand how it is measured
- To evaluate in a critical manner the need for genetic testing
- To understand quality control and quality assurance issues in molecular diagnostics
- To understand the implications of molecular diagnosis as it relates to patient insurability
- To understand the managerial aspects of the molecular diagnostics laboratory, focusing on cost-effectiveness of testing and the clinical utility of an assay

Practice-based learning

- To use case-based learning as a tool for additional insight into the molecular basis of disease pathogenesis
- To understand the ACMG/AMP guidelines for reporting of germline and somatic sequence variants
- To locate, appraise and assimilate pertinent evidence from scientific studies
- To demonstrate effective problem solving skills, using a wide variety of information resources

NEXT-GENERATION SEQUENCING (NGS)

Goals and Objectives:

Trainees will gain skills in the interpretation of NGS data for the diagnosis of inherited disorders and acquired diseases utilizing whole exome sequencing (WES) or exome-based NGS panel testing in the clinical genomics lab or a disease specific 50 gene myeloid panel. Trainees will actively participate in the interpretation of NGS results, variant classification and preparation of clinical reports.

Rotation Faculty: Laura Lee, M.D., Ph.D., Interim Medical Director of Clinical Genomics

PATIENT CARE

- To learn to incorporate germline and somatic clinical NGS testing into patient care.
- To learn to correlate clinical and morphologic findings with NGS results.
- To understand ethical and clinical implications of NGS testing.

MEDICAL KNOWLEDGE

- To understand the laboratory techniques used for NGS on the Illumina platform.
- To understand the importance of levels of evidence for the interpretation of NGS results.
- To gain experience with the application of ACMG/AMP criteria for the classification of germline and somatic NGS variants.
- To understand the importance of computer processing of NGS data.

INTERPERSONAL AND COMMUNICATION SKILLS

- To prepare detailed, accurate, and clearly written NGS reports using the Genosity portal.
- To actively participate in Clinical Genomics Lab group huddles.
- To present at least two interesting cases at biweekly and/or monthly conferences attended by clinical colleagues.

PROFESSIONALISM

- To demonstrate respect and compassion for patients and other team members.
- To review patient results and prepare clinical NGS reports in a timely fashion.
- To work effectively as a member of a diverse team that includes pathologists, genetic counselors, variant scientists, clinical geneticists, medical technologists, and business/finance managers.

SYSTEMS-BASED PRACTICE

- To maintain clear communication about cases with the clinical team as appropriate.

- To gain familiarity with issues surrounding insurance pre-authorization for clinical germline NGS testing by attending at least two regularly held meetings with the finance team.
- To attend a monthly Anatomic Pathology Molecular Improvement Team meeting to observe efforts to streamline somatic NGS workflow processes from pre-analytic to post-analytic phases.

PRACTICE-BASED LEARNING

- To review and learn from NGS interesting case files.
- To become familiar with sources of valid and useful information for the assessment of NGS data.
- To learn how to investigate and interpret possible unexpected findings.

SITUATIONS IN WHICH EVEN AN EXPERIENCED RESIDENT MUST CALL AN ATTENDING:

- When there is contact by an attorney, a relative, or any party not secured by HIPAA requesting information.
- When there is contact by an upset clinician.
- When a possible specimen loss or misidentification has occurred.
- When a preliminary or final diagnosis is requested.
- When unusual results are observed, and the resident is unsure how to troubleshoot the situation to avoid delays in repeat testing.
- When there may be a conflict involving the resident and another laboratory professional.
- When the resident is unsure how to process a unique laboratory specimen.
- When important data processing systems unexpectedly fail.

CLINICAL GENETICS

Goals:

The goal of this rotation is to provide a working knowledge of medical genetics including dysmorphology, inborn errors of metabolism, adult late onset genetic disease, prenatal diagnosis, and cancer genetics.

Rotation Faculty: Angela Grochowski (Peds) and Georgia Wiesner (Cancer- LGG only) and Martha Dudek (Prenatal-LGG only)

Objectives:

Patient Care

- To understand how to evaluate the family and clinical history of the patient, perform an examination to develop a differential diagnosis for the patient and to order appropriate laboratory testing to assist in the analysis
- To learn how to review biochemical, cytogenetics and/or molecular genetic results to establish a diagnosis
- To understand how to calculate risk assessment based on family history and laboratory data
- To observe (LGG only) and learn how testing for metabolic diseases is performed

Medical Knowledge

- To understand how to identify and distinguish between mendelian and mitochondrial inheritance, chromosomal abnormalities and multifactorial disorders
- To understand principles of genetic disease including, but not limited to: genetic anticipation, penetrance, imprinting, genetic and allelic heterogeneity and X inactivation
- To understand the pathogenesis of genetic diseases
- To understand the mechanism of disease by understanding the causative effects of missense, nonsense, frameshift, duplication, deletions, promoter and splice site variants

Interpersonal and Communication Skills

- To understand the various components in patient reports
- To develop effective oral communication skills when dealing with patients and their families

To develop oral presentations skills by presenting interesting cases

Professionalism

- To demonstrate respect and compassion for patients
- To work effectively as a team player and treat fellow colleagues, technical and administrative staff with respect

Systems-based Practice

- To utilize on-line web based genetic data bases to understand genetic diseases for information on diagnostics and management guidelines
- To utilize on-line in house data bases for patient chart review and laboratory results

Practice-based learning

- To use case-based learning as a tool to assimilate pertinent data from genetic databases and the scientific literature to formulate a differential diagnosis
- To demonstrate effective problem solving skills

Clinic Attended	
Pediatric and Biochemical Genetics	Maintain Log of Cases
Hereditary Cancer	Maintain Log of Cases
Prenatal	Maintain Log of Cases
