

**MEDICAL GENETICS TRAINING PROGRAM**  
*Mount Sinai School of Medicine*

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**GOALS & OBJECTIVES: Molecular Genetics Laboratory**

The overall goal of the Clinical Molecular Genetics Training Program is to provide the trainee with the skills they will need to direct a molecular diagnostic laboratory. This includes gaining expertise in test design and validation; interpretation, troubleshooting, reporting and communication of results to providers; proper quality control and assurance and to direct a fellowship program in the future. In addition, the trainee will perform research in molecular genetic testing.

The specific skills to master are outlined below:

**Patient Care and Laboratory Skills:**

1. Preanalytical Laboratory Skills

- Accurately select appropriate laboratory testing based on diagnosis and/or clinical features or appropriate screening testing
- Identify appropriate specimen type for testing performed in the laboratory
- Knowledgeable in specimen collection, transport, storage, stability, common interferences, acceptability, accessioning, specimen tracking, and appropriate documentation

2. Analytical Laboratory Skills

Competent in practicing general lab quality management procedures through all lab rotations

- Perform reagent controls with all assays including tracking critical reagents lots, testing new critical reagents prior to use, appropriate labeling of all reagents
- Perform quality control with all assays, compile QC record, track QC trend
- Perform regular calibration procedures
- Demonstrate knowledge in setting up and implementing necessary quality measures to assure and improve testing quality
- Get involved with preparation for lab inspections
- Receive training on all aspects of nucleic acid isolation to ensure that the samples are not contaminated and adequate DNA/RNA is obtained for analysis
  - Use of aseptic techniques
  - Choice of appropriate method of isolation
  - Determine concentration or integrity when needed
  - Troubleshoot poor or failed isolations
  - Proper storage of samples and extracted nucleic acid
- Master all aspects of polymerase chain reaction amplification (PCR)
  - Principles of PCR
  - Primer design and assay development
  - Techniques for minimizing PCR contamination
  - Proper controls
  - Troubleshooting of results
- Southern blot analysis
  - Understand principles and techniques used in Southern blotting
  - Troubleshooting of results
- Targeted mutation analysis
  - Understand principles of targeted mutation analyses
    - Multiplex allele specific primer extension and bead arrays
    - Restriction fragment length polymorphism analysis
    - FRET analysis

- Allele specific hybridization
    - Pyrosequencing
  - Gene Scanning analysis
    - Understand principles and techniques used for gene scanning
      - dHPLC
      - MLPA
    - Troubleshooting of results
  - Gene sequencing
    - Understand principles and techniques used in gene sequencing
    - Knowledgeable about different databases and assay design
    - Troubleshooting of results
  - Quantitative PCR
    - Understand principles and techniques used in quantitative PCR
    - Troubleshooting and interpretation of results
  - Array Analysis
    - Understand principles and techniques used in array analysis
    - Troubleshooting of results
  - Identity testing
    - Understand principles and techniques used in identity testing
      - Zygoty
      - Maternal cell contamination studies
    - Troubleshooting of results
3. Postanalytical Laboratory Skills
- Results interpretation and reporting
    - Competent in result interpretation. Recognize normal/carrier/affected status, determine the clinical significance, correlate with clinical findings and recent literature, make recommendations for additional testing.
    - Competent in drafting accurate clinical reports
    - Communicate results clearly with healthcare professionals (residents and genetic counselors)
4. Rotations in Cytogenetics/Cytogenomics and Biochemical Genetics non-specialty laboratories
- Understand the indications for performing cytogenetic and biochemical testing
  - Understand the technologies employed and the technical workflow
  - Understand the quality control measures implemented in the laboratory
  - Understand the interpretation and reporting of results
  - Understand the limitations of the technologies employed

### **Medical Genetic Knowledge:**

1. Understand the principles of biology and genetics- covered by departmental and divisional seminars, journal clubs and case conferences and medical genetics course
2. Understand principles of molecular genetics
  - a. Mode of inheritance
  - b. Different classes of mutations
  - c. Describe mutations based on standard nomenclature
  - d. Bayesian risk analysis
  - e. Significance of novel variations

### **Practice-Based Learning**

1. Can review literature to obtain information on molecular genetics techniques and their use in clinical medicine
2. Participate in presenting cases at division case conference

### **Interpersonal & Communication Skills**

1. Can obtain clinical information to assist the lab in appropriate test selection
2. Can explain clinical findings and their significance to laboratory staff
3. Can communicate laboratory results in a appropriate manner to patients & their families, and referring physicians and genetic counselors

### **Professionalism**

1. Demonstrates respect for patient confidentiality
2. Considers sensitivity to patients of diverse backgrounds when communicating lab results
3. Become sensitive to ethical issues in genetics, particularly those raised by molecular genetic testing

### **System-Based Practice**

1. Work effectively with various members of the clinical and laboratory team to facilitate accurate and appropriate patient testing and reporting of results
2. Is aware of the cost and risks/benefits of obtaining samples and providing testing
3. Participate in lab quality management process including quality control, quality assurance and quality improvement
4. Familiarize with other lab management process including laboratory information systems, billing and reimbursement issues and policy implications

### **Research-Related Activities**

1. Learn IRB and HIPAA regulations, protocols, and consent process
2. Development of new laboratory tests and validation of assays for clinical use
3. Participate in ongoing translational research projects related to molecular genetics