



Molecular Diagnostics and Cytogenetics Course

UNC Department of Pathology and Laboratory Medicine
September 2019 (1-month duration)

Goal: To instill expertise in molecular diagnostics and cytogenetics so that our residents and fellows become competent clinical consultants on using genetic technology in a wide variety of clinical settings. Trainees develop technical, clinical, communication, management, and judgment skills. A fundamental understanding of genetic technologies prepares trainees to critically evaluate the medical literature now and as new methods are introduced in future years. Training is provided in a structured environment via didactic seminars, laboratory workshops under the supervision of expert faculty, and interaction with clinicians, counselors, and laboratory scientists. A project is completed on a topic of choice.

Objectives:

1. Gain a working knowledge of molecular technologies including polymerase chain reaction, *in situ* hybridization, microarrays, and sequencing (Sanger, NextGen, pyrosequencing). Learn about clinical applications in patients with cancer, heritable disease, and infectious disease, and about applications in HLA typing, parentage, forensics, and pharmacogenetics.
2. Gain a working knowledge of cytogenetics including terminology and methods (karyotype, FISH, SNP chip). Learn clinical applications in prenatal diagnosis, congenital disorders and cancer diagnosis and monitoring.
3. Interpret molecular and cytogenetic data in correlation with clinical, morphologic, and immunophenotypic findings.
4. Discuss quality assurance, assay validation, ethics, regulatory issues, and lab administration.

Resident Duties and Responsibilities: The resident attends didactic sessions, reads articles, delivers case-based and scholarly presentations, and interprets sample results. Recommended resources include:
<https://www.amp.org/education/education-resources/>

Coleman W, Tsongalis G: *Diagnostic Molecular Pathology*, 1e, Elsevier (2016)

Leonard D: *Molecular Pathology in Clinical Practice*, 2e, Springer (2016)

Burtis CA: *Tietz Textbook of Clinical Chemistry & Molecular Diagnostics*, 6e. Saunders Elsevier (2017)

Gerson SL and Keagle MB: *The Principles of Clinical Cytogenetics*, 3e, Springer (2013)

Arsham MS: *The AGT Cytogenetics Laboratory Manual*, 4e, Wiley (2017)

Sarafoglou K: *Pediatric Endocrinology and Inborn Errors of Metabolism*, 2e, McGraw Hill (2017)

Resident Evaluation: Participation is monitored in scheduled laboratory and didactic sessions overseen by faculty and staff. The resident delivers three oral case-based presentations, and a fourth end-of-rotation presentation on a topic of their choice. The course director oversees training and evaluates performance with input from colleagues.

Course Directors: Margaret L. Gulley, MD margaret_gulley@med.unc.edu

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Faculty: Berg, Booker, Calikoglu, Eron, Farber, Forman, Gulley, Kaiser-Rogers, King, Lai-Goldman, Merker, Miller, Montgomery, Nelson, Powell, Schmitz, Schwartz, Thorne, Trembath, Weck, Weimer, fellows, technologists, and others.

Registration: Training occurs at UNC Hospitals from ~9-5pm weekdays in September 2019. The course is required for Anatomic & Clinical Pathology Residents and some Fellows at UNC. Participants from other departments / institutions may register for \$1200 each. The deadline for registering is Aug 1, 2019.

For information contact Dr. Gulley at margaret_gulley@med.unc.edu.