8:00am
Welcome and Introductions | Marjorie David, MD, MS *University of Texas Health Science Center at San Antonio*

8:05am
To PCR and Beyond: Molecular Tools for Infectious Disease Detection | Cecilia Thompson, MS, PhD, D(AB-MM) *Ann & Robert H. Lurie Children's Hospital of Chicago*

This session will review molecular approaches for infectious disease screening and diagnosis. The session will provide an overview of target amplification methods that include polymerase chain reaction (PCR), non-PCR and isothermal amplification techniques such as transcription-mediated amplification, and highly complex molecular techniques such as targeted and untargeted sequencing. The limitations of molecular techniques for infectious disease diagnosis will be discussed.

Learning Objectives
· Review the diverse molecular techniques used in infectious disease screening and diagnosis.
· Highlight the utility of molecular based assays in the diagnosis of some common infectious diseases.
- Describe the limitations of molecular approaches in infectious diseases' screening and diagnosis.

8:30am
Introduction to Bioinformatics Pipeline and Data formats | Weiwei Zhang, PhD *University of Nebraska Medical Center*

The session will introduce a bioinformatics pipeline for comprehensive genomic profiling assays, describe three major components within the pipeline, and review main steps in each component. The session will also cover the most applied data formats involved with examples through the pipeline.

Learning objectives
· Introduce a general bioinformatics pipeline
· List major components of the bioinformatics pipeline
· Describe common data formats used in bioinformatics

9:00am
Clinical Utilities of Optical Genomic Mapping at a Clinical Cytogenetic Laboratory | Ying Zou, MD, PhD *Johns Hopkins University*

The session will describe technical components of optical genome mapping (OGM), review clinical utilities from OGM literature, go through OGM nomenclature, and mention OGM regulator issues associated with clinical practices. The session will also share three clinical cases to demonstrate clinical utilities in a clinical cytogenetic laboratory.

Learning objectives
· To understand technical components of optical genome mapping (OGM)
· To be familiar with clinical utilities of OGM in patients with constitutional diseases and various cancers including hematological malignancies and solid tumors.
· To understand nomenclature and regulator issues associated with OGM adoption into clinical practices.

9:20am Break

9:30am
Concurrent BREAK-OUT Sessions (Each case study session with be 30 minutes)

Case Study 1 – Genetics
Yang Cao, PhD *Washington University in St. Louis, School of Medicine* **and Eli S. Williams, PhD** *University of Virginia School of Medicine*

Join us as we dive into genetics case studies, which will showcase the power of advanced genomic technologies in unraveling genetic mysteries. In this session, we'll explore real-world genetic puzzles while providing insights into current and emerging molecular modalities, data analysis and interpretation, and the impact that genomic medicine can have for our patients.

Learning Objectives
· Gain a comprehensive understanding of molecular genetic testing modalities, including their principles, workflows, and applications in genomics.
· Learn the essential steps and tools involved in molecular genetic data analysis, from raw data processing to variant calling, and understand how to interpret genomic findings.
· Explore the clinical significance of molecular testing results by examining how they impact patient diagnosis, treatment decisions, and genetic counseling, while also addressing ethical considerations.
- Develop the ability to apply advanced molecular technology effectively to solve complex genetic mysteries through the in-depth examination of a real-life case study, gaining practical insights into its potential applications in research and clinical practice.

Case Study 2 – Hematopathology
Kevin E. Fisher, MD, PhD *Baylor College of Medicine* **and Jo Conant, MD** *University of Vermont Medical Center and College of Medicine*

Next-generation sequencing (NGS) has become a core component of the work up of many hematologic malignancies. However, there are instances in which NGS testing, in conjunction with conventional karyotype and FISH analysis, may not fully characterize genomic alterations that can impact diagnostic classification and prognostication. In this session, we will showcase additional genomic methodologies including optical genome mapping (OGM) and microarray analysis that supplement initial NGS testing.

Learning Objectives
· Recognize which hematopathology situations may benefit from additional diagnostic testing such as OGM or microarray.
· Identify diagnostic OGM data patterns.
· Interpret microarray data in the context of positive NGS findings.
- Assess diagnostic yield of OGM and microarray in select scenarios.

Case Study 3 – Infectious Diseases
Yi Ding, MD, PhD *Geisinger Medical Center* **and** **Rebecca Yee PhD, D(ABMM)** *The George Washington University School of Medicine and Health Sciences*

Using different case studies, we will showcase the diversity of molecular approaches from PCR to NGS used for infectious diseases diagnosis. We will discuss the utility and challenges of the different molecular technologies and their role in the clinical management of infectious diseases. We will also cover implementation of quality metrics to ensure accurate and reliable test results.

Learning Objectives
· Differentiate between qualitative and quantitative PCR approaches and their respective roles in diagnosing infections.
· Explain how advanced molecular technologies, such as sequencing, are implemented into current microbiology workflows.
· Describe tools and processes to ensure good molecular laboratory practice in infectious diseases.

Case Study 4 - Solid Tumors
Alexander J. Neil, MD, PhD *Brigham and Women's Hospital* **and** **Anna Matynia, MD** *University of Utah*

We will review a short series of solid tumor cases tested by next-generation sequencing (NGS) assays with tricky mutation patterns. These cases will highlight the importance of through clinico-morphologic correlation in providing proper interpretation and recommendations and the role of molecular pathologists in that process. We will have interactive discussions of potential pitfalls of inaccurate correlations, review of basic tumor biology, as well as confirmatory approaches (including other molecular methodologies).

Learning Objectives
· Perform basic NGS data interpretation.
· Review basic tumor biology in selected solid tumor types.
· Highlight the importance of accurate clinico-morphologic correlation of molecular results for proper patient management.

11:30am Closing Remarks | **Marjorie David, MD, MS,** *University of Texas Health Science Center at San Antonio*