AMP 2018 Molecular Pathology Outreach Course

AMPlicons: A Practical Molecular Toolkit and Case Studies

Wednesday, October 31, 2018

Grand Hyatt Presidio ABC 3rd Floor, San Antonio, Texas

DETAILED AGENDA

TIME	SESSION	SPEAKER	CE Hours
7:30am	**Registration and Continental Breakfast**		
8:15am	**Welcome and Introductions**	**Cecilia C. S. Yeung, MD** *Fred Hutchinson Cancer Research Center*	**NA**
8:20am	**HOT TOPICS in Molecular Diagnostics**	**Kojo S. J. Elenitoba-Johnson, MD** *University of Pennsylvania, Perelman School of Medicine*	**NA**
9:05am	**Molecular Testing Landscape – Pre-analytical Considerations** • Compare main types of samples submitted for molecular testing highlighting key criteria for specimen selection. • Determine the rationale for specimen requirements for molecular testing. • Assess requirements for the sample adequacy in relation to the test performance characteristics.	**Anna Yemelyanova, MD** *University of Alabama at Birmingham*	**0.5**
9:40am	**BREAK**		

Lectures and Break-out Sessions			CE Hours
10:00am	**Non-PCR based Methodologies and Clinical Applications** • Explain the mechanism of the cleavase probe signal amplification methodology. • Describe the differences between signal amplification and target amplication and identify a clinical application of the methodology.	**Cynthia L. Jackson, PhD** *Rhode Island Hospital and Brown University Alpert Medical School*	**0.75**
10:15am	**Non-PCR Case Studies and Break-out Session** ***Hybridization based nucleic acid counting*** • Recognize the utility of molecular testing in breast cancer prognosis. • Identify and appraise the characteristics of the NanoString Prosigna assay. • Identify and select the appropriate tissue specimens for use with the Prosigna assay. ***Fluorescence in situ hybridization (FISH) for the detection of structural and copy number variation*** • Explain common appropriate use of FISH in clinical diagnostics. • Interpret common ISCN nomenclature for FISH results. • Interpret common FISH images.	**Rashmi Goswami, MD PhD** *University of Toronto* **Kristy R. Crooks, PhD** *University of Colorado, Denver*	
10:35am	**Q&A Session**		
10:40am	**PCR Methodologies and Specialized PCR Applications** • Assess which method of detecting PCR amplification is most suitable for a given clinical assay. • Describe clinical situations which call for a single amplicon versus multiplex PCR design.	**Jeffrey Gagan, MD PhD** *UT Southwestern University*	**0.75**
10:55am	**PCR Case Studies and Break-out Session** ***Nested Multiplex PCR for Multi-Pathogen Meningitis Detection*** • Describe the clinical utility of multiplex syndromic panels. • Explain the advantages and limitations of the meningitis syndromic panel testing. ***Specialized PCR Applications in Cystic Fibrosis*** • Describe the principles of Multiplex Ligation-dependent Probe Amplification (MLPA) for copy number detection. • Evaluate Cystic Fibrosis carrier screening data and select cases where MLPA is an important adjunct to routine methods.	**Preeti Pancholi, PhD** *Ohio State University* **Mark D. Ewalt, MD** *University of Colorado, Denver*	
11:15am	**Q&A Session**		

11:20am	**Micro Array and Hybridization-based Technology and its Clinical Application** • Describe microarray technology. • Apply microarray technology.	**Yassmine Akkari, PhD** *Legacy Health*	**0.75**
11:35am	**Array Case Studies and Break-out Session** ***Microarrays in Oncology: Case Studies*** • Apply microarray technology in oncology case studies. ***Oligo Hybridization-based methodology: Microarray in the Infectious Diseases Diagnostic Laboratory*** • Explain how microarray technology is used in the Clinical Microbiology Laboratory. • Discuss the impact of microarray technology on patient care.	**Yassmine Akkari, PhD** **Sophie S. Arbefeville, MD** *University of Minnesota Medical Center*	
11:55am	**Q&A Session**		
12:00pm	**LUNCH**		**NA**

	Lectures and Break-out Sessions		
1:15pm	**Introduction to NGS Platform & Clinical Applications** • Describe the two main steps of a library preparation and some of the different variations that are possible. • Compare different next-generation sequencing platforms and understand the basic chemical differences. • Provide examples of clinical applications for NGS.	**Cecilia C. S. Yeung, MD**	**1.0**
	Introduction to Bioinformatics Pipeline & Data Analysis • Select the appropriate database to research a given genomic variant. • Arrange the essential steps of a DNA next-generation sequencing bioinformatics pipeline.	**Joshua Coleman, MD** *University of Utah*	
2:15pm	**BREAK**		**NA**
2:30pm	**NGS Case Studies and Break-out Session** ***Using Next Generation Sequencing in Constitutional Genetics: Glycogen Storage Disease*** • Discuss indications, diagnosis, and typing of glycogen storage disease and the benefits of using next generation sequencing. • Describe NGS sequencing and analysis in constitutional genetics using the Ion Torrent PGM. • Discuss criteria for identification and reporting of variants found. ***Solid Tumor NGS case study*** • Describe molecular alterations in GIST. • Discuss limitations of NGS technology. ***Next Generation Sequencing in Hematologic Malignancies*** • Describe a fusion gene and possible testing modalities to identification of gene fusion. • Identify common and significant fusion genes common in myeloid neoplasms. ***Whole Exome Sequencing and Complex Phenotypes*** • Show the importance of Whole Exome Sequencing (WES) for identification of known and new genes in families segregating with Mendelian diseases. • Identify the appropriate applications to WES testing. • Evaluate the usefulness and pitfalls of the technique for investigation of cases with mixed / complex phenotypes not described in the literature.	**Barbara Anderson, BS, MS** *Duke Molecular Diagnostic Laboratory* **Susan Hsiao, MD** *Columbia University* **Brittany Coffman, MD** *University of New Mexico* **Roberta Sitnik, MSc PhD** *Hospital Israelita Albert Einstein*	**1.0**

3:30pm	**Closing Remarks, Questions, and Evaluations**	**Cecilia C. S. Yeung, MD**	**NA**
3:45pm	**Adjourn**		