AMP 2019 Molecular Pathology Outreach Course

AMPlicons: A Practical Molecular Toolkit and Case Studies

Wednesday, November 6, 2019

Hilton Holiday Ballroom 1/2

Baltimore, Maryland

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*	**-**
8:20am	**Molecular testing advancements and hot topics on the global front**	**Marilyn Li, MD** *Children's Hospital of Philadelphia*	**-**
9:05am	**PRE-ANALYTICAL CONSIDERATIONS** This session will help in understanding the tissue and sample requirements in the process of molecular testing including possible limitations that may be present during the process prior to obtaining a molecular result. Methods of improving the chances of obtaining a molecular result in scant tissue specimens will also be discussed. Preanalytic considerations for tissue specimens, hematologic (blood/bone marrow) specimens, DNA and RNA, as well as microbiological specimens will be reviewed. • Describe key issues in sample collection, transport, and storage for molecular specimens. • Discuss rationale for specimen selection for molecular testing • Discuss strategies to maximize material for testing with limited specimens.	**Rashmi Goswami, MD, PhD** *Sunnybrook Health Sciences Centre, University of Toronto* **Susan Hsiao, MD, PhD** *Columbia University Medical Center*	**0.5**
9:40am	**BREAK**		
	Lectures		
10:00am	**Signal Amplification-based Methodologies & Clinical Applications; PCR Methodologies and Specialized PCR Applications** The goals of the session are to provide a concise review of the mechanisms involved in PCR and discuss the many variations of this basic concept as they are employed in clinical assays. In addition, brief descriptions of various new techniques and their potential in the clinical molecular diagnostics laboratory will be discussed. • Describe the PCR and the most common approaches to PCR. • 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.	**Barbara Anderson, MS, MB(ASCP)^{CM}** *Duke University Health Systems*	**0.5**
10:25am	**Q&A Session**		
10:30am	**Micro Array and Hybridization-based Technology and its Clinical Application** This session is a description of hybridization-based microarray technologies and its role in oncology and infectious disease. • Describe microarray technology • Applications of microarray technology • Interpret microarray results from case examples	**Yassmine Akkari, PhD** *Legacy Health*	**0.5**
10:55am	**Q&A Session**		
11:00am	**Introduction to NGS Platform & Clinical Applications** In the NGS introduction lecture, we will review different next-generation sequencing (NGS) platform, basic steps of NGS library preparation and variations on the chemistry. We will also review some clinical applications for NGS during this introductory talk. • 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. • Give examples of clinical applications for NGS.	**Cecilia C.S. Yeung, MD**	**0.5**
11:25am	**Q&A Session**		

11:30am	**Introduction to Bioinformatics Pipeline & Data Analysis** This session is an introduction to Bioinformatics and its role in molecular diagnostics next generation sequencing (NGS) assay development. • Describe the basics about the role of bioinformatics in a molecular test. • Construct a simplified bioinformatics process for variant calling and variant annotation. • Describe the overall components of a bioinformatics pipeline for a molecular test, without necessarily understanding the technical details.	***Sabah Kadri, PhD*** *Ann & Robert H. Lurie Children's Hospital of Chicago*	**0.5**
11:55am	**Q&A Session**		
12:00pm	**LUNCH**		
	Break-out Sessions		
1:00pm	**Introduction and Overview to NGS Workshop**	**Cecilia C.S. Yeung, MD**	**-**
1:05pm	**BREAK-OUT Sessions** (See below for break-out titles, speakers, and descriptions)		**1.0**
2:10pm	**Break**		
2:20pm	**BREAK-OUT Sessions (Continued)**		**1.0**
3:25pm	**Questions, Closing Remarks, and Evaluations**	**Cecilia C. S. Yeung, MD**	**-**
3:45pm	**Adjourn**		

BREAK-OUT Session Descriptions

NGS Case Study 1 – Infectious Diseases This is a 30-minute interactive case presentation that walks learners through the basic methods behind and applications of metagenomic next-generation sequencing for infectious disease diagnosis. • Describe the basic principles of metagenomic next generation sequencing. • Identify scenarios in which mNGS would be clinically useful. • Discuss some limitations of mNGS testing.	***Roberta Sitnik, MSC, PhD*** *Hospital Israelita Albert Einstein, Brazil* **Erin H. Graf, PhD** *Mayo Clinic Arizona* **Preeti Pancholi, PhD** *Ohio State University Wexner Medical Center*
NGS Case Study 2 – Solid Tumors The pathogenesis of cancer is marked by deregulation of multiple intracellular signaling pathways, often due to genetic mutation. Next-generation sequencing provides a cost-effective method to assay for these multiple abnormalities. Although this would be impossible without high-performance computing and sophisticated algorithms, human expertise remains indispensable. Not all findings will be appropriate for clinical reporting, for instance, and automated data analysis may give rise to interpretive discrepancies. In particular, complex multinucleotide alterations often require pathologists' judgment, as explored in the following cases. • Review the molecular pathogenesis of colorectal carcinoma. • Explain a general approach to annotating genetic variants discovered by next-generation sequencing experiments/assays. • Describe several challenges in detecting and reporting complex insertions and deletions.	**Christian A. Kunder, MD, PhD** *Stanford University School of Medicine* **Joshua Coleman, MD** *University of Utah/ARUP Labs*
NGS Case Study 3 – Hemepathology This session will review how next-generation sequencing (NGS) testing can be used in the interpretation of myeloid malignancies. We will walk through an example from the initial clinical presentation through the morphologic features of the bone marrow biopsy, and will teach you how to correlate all of this data with the NGS myeloid panel results for an appropriate interpretation. We will discuss the spectrum of myeloid malignancies from Clonal Hematopoiesis of Indeterminate Potential (CHIP) to acute myeloid leukemia. We will also review the strengths and the limitations of using a myeloid NGS panel in conjunction with a bone marrow biopsy. • List some advantages and disadvantages of using NGS in the evaluation of bone marrow biopsies and myeloid malignancies. • List some of the mutational patterns of CHIP, MDS, AML and other myeloid malignancies. • Interpret NGS mutation profiles and describe which patterns have a higher positive predictive value of leading to a myeloid malignancy.	***Kristin Hunt Karner, MD*** *University of Utah/ARUP Labs* **Rashmi Goswami, MD, PhD**
NGS Case Study 4 – Genetics This session will review the identification of germline variants in tumor-only sequencing, with clinical follow up and confirmation. • Evaluate the likelihood that a variant detected on tumor-only sequence variants may represent germline variants • Interpret the role of variant allele fraction in determining whether a variant is likely to be germline	***Sabah Kadri, PhD*** **Alanna Church, MD** *Boston Children's Hospital*