

# AMP Reference Material Forum

October 30, 2018

Location/ Room: Grand Hyatt, Travis AB, 3<sup>rd</sup> Level (600 E Market St, San Antonio, TX 78205)

## Agenda

- 8:00 SPOT/Dx Quality Assurance Pilot - Reference Samples for NGS Comparisons**  
*Barbara Zehnbauer PhD, Emory University, Atlanta, GA*
- 8:30 Defining the Somatic Mutation Gold Set for Cancer Reference Samples -- SEQC2 Consortium**  
*Li Tai Fang PhD, Roche Sequencing Solutions, Belmont, CA*
- 9:00 Mission Accomplished: A Panel of 108 Genomic DNA Reference Materials for HLA- A, B, C, DRB1, DRB3, DRB4, DRB5, DQA1, DQB1, DPA1 and DPB1 Loci**  
*Maria Bettinotti PhD, Johns Hopkins School of Medicine, Baltimore, MD*
- 9:30 Challenges in Microbiome Research – The Importance of Standardized Reference Materials**  
*Dev Mittar PhD, American Type Culture Collection (ATCC), Manassas, VA*
- 10:00-10:30 Break (on your own\*)**
- 10:30 An Update on NIST Viral Reference Materials and Somatic Mutations in the GIAB samples**  
*Megan Cleveland PhD, National Institute of Standards and Technology (NIST), Gaithersburg, MD*
- 11:00 Update on NIST/Early Detection Research Network Liquid Biopsy Projects for Cancer Genomic Measurements**  
*Hua-Jun He PhD, National Institute of Standards and Technology (NIST), Gaithersburg, MD*
- 11:30 Guidelines and Recommendations for Detection of CNVs in Cancer Samples with high Accuracy and Precision using digital PCR**  
*Toumy Guettouche PhD, Roche Sequencing Solutions, Pleasanton, CA*
- 12:00-1:30 Lunch (on your own\*)**
- 1:30 Using Reference Materials and Clinical Data to Rigorously Examine the Need to Confirm NGS-Detected Variants in Clinical Genetic Testing**  
*Stephen E. Lincoln, Invitae, San Francisco, CA*
- 2:00 Genome in a Bottle Benchmarks for Structural Variation**  
*Justin Zook PhD, National Institute of Standards and Technology (NIST), Gaithersburg, MD*
- 2:30 The MDIC Somatic Reference Standards Project: Introduction and Request for Input on Variant Prioritization**  
*JD Alvarez MD, PhD, Janssen, Spring House PA; Stayce Beck PhD, Food and Drug Administration, Silver Spring, MD; Timothy McDaniel PhD, Translational Genomics Research Institute, Phoenix AZ*
- 3:30-3:45 Break (on your own\*)**
- 3:45 Supporting Molecular Diagnostics: The Role of Digital PCR in the Value Assignment of Reference Materials**  
*Denise O'Sullivan PhD- National Measurement Laboratory, LGC, Middlesex UK*
- 4:15 State of the Caller 2018: Best-Practices Benchmark of Variant Callers for Whole-Genome Short-Read Sequencing Data**  
*Francisco De La Vega D.Sc, Stanford University, Stanford, CA*

\*For a list of dining options near the Hyatt, please refer to: [http://visitsanantonio.com/Microsites/New-Template-\(10\)/Dining](http://visitsanantonio.com/Microsites/New-Template-(10)/Dining) ; Participating restaurants that provide discounts with your official conference badge: <http://visitsanantonio.com/VisitSanAntonio/media/vsa-media/microsites/pdfs/SUYB-101818.pdf>