

## Poster Listing

**Even numbered ePosters** will be attended by their authors on Tuesday, November 17, 1:00pm – 2:00pm Eastern

**Odd numbered ePosters** will be attended by their authors on Thursday, November 19, 1:00pm – 2:00pm Eastern.

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### GENETICS

**G01. Development and Validation of a High-Throughput Next-Generation Sequencing Assay from Buccal Cell DNA as a Cost-Effective Screening Method for Celiac Genetic Risk**

*S. Gunn*

**G02. Copy Number Variant Analysis Improves the Diagnostic Yield in a Cohort of Pediatric Patients with Previously Negative Constitutional Exome Sequencing Results**

*E. Hahn*

**G03. A Retrospective Study of Products of Conception with More Than 44,000 Specimens in 27 Years at a National Cytogenetic Reference Laboratory**

*H. Meng*

**G04. WITHDRAWN**

**G05. Reevaluation of Genomic Test Results for Germline Disorders: A Framework of Critical Considerations on Behalf of CLSI Document Development Committee (DDC) on Nucleic Acid Sequencing (MM09)**

*J. Ji*

**G06. Single Gene Transcript Analysis and 3D Modeling: An Integrated Approach to Variant Assessment**

*F. Vetrini*

**G07. Detection of Allelic Dropout in a Mass Array *HFE* Genotyping Assay**

*A. Campbell*

**G08. Result Interpretation for Clinical Exome and Genome Sequencing: On Behalf of CLSI Document Development Committee (DDC) on Nucleic Acid Sequencing (MM09)**

*J. Buchan*

**G09. Incidental Diagnosis of *NR5A1*-Related 46,XY Disorder of Testicular Development in Neonate with Mosaic Partial Trisomy 2q**

*S. Vallee*

**G10. Optimization and Validation of a Sanger Sequencing Clinical Assay for Germline *BRCA1/2* Gene Mutation Detection at King Hussein Cancer Center**

*W. Naser*

**G11. Mosaicism in Cancer Susceptibility Genes in Unselected Cancer Patients**

*D. Mandelker*

**G12. Presumed Germline Pathogenic Variants in Tumor-Only Sequencing: Frequency and Follow-Up**

*D. Toledo*

**G13. Comparison of Universal versus Traditional Genetic Testing Models for Cancer Patients**

*O. Ceyhan-Birsoy*

**G14. The Prevalence and Distribution of Germ-Line Inherited Cancer-Associated Variants**

*T. Huard*

**G15. Characterization of Reference Materials for Spinal Muscular Atrophy Genetic Testing: A GeT-  
RM Collaborative Project**

*T. Prior*

**G16. A Rare Single Nucleotide Variant Causing a False-Negative *HTT* CAG Repeat Expansion Result in the Evaluation of a Patient for Huntington Disease**

*F. El-Sharkawy*

**G17. Amplification-Free Targeted Enrichment Powered by CRISPR-Cas9 and Long-Read Single Molecule Real-Time Sequencing Can Efficiently and Accurately Sequence Challenging Repeat Expansion Disorders**

*J. Ekholm*

**G18. A Single-Assay Diagnostic Workflow for Genotyping and Phasing SNPs with Repeat Expansions for Allele-Selective Therapy in Huntington Disease**

*S. Statt*

**G19. Review of Analysis Methods for Repeat Expansion Diseases Using Capillary Electrophoresis Data**

*M. Avenarius*

**G20. Test Validation and Characterization of Reference Materials for *ADH5* Genotyping**

*A. Otsubo*

**G21. *CYP2D6* Guided Methadone Dosing in a Multi-Ethnic Population: A Pharmacogenomic Screen to Decrease Withdrawal Morbidity**

*C. Lum*

**G22. Developing *DPYD* Genotyping Method for Personalized 5-fluorouracil Therapy**

*B. Wong*

**G23. High-Throughput Fetal-Fraction Amplification Increases Analytical Performance of Noninvasive Prenatal Screening**

*D. Muzzey*

**G24. A Software Tool That Prevents Incorrect Estimations of Gestational Age and Maternal Age at Estimated Date of Delivery Reported by the College of American Pathologists NIPT Participant Summary**

*Y. Wang*

**G25. Genetic Insights and Incidental Findings from Maternal Cell Contamination Testing**

*N. Kopp*

**G26. The Relationship between Variant Type and Phenotype among Diseases Screened by the Foresight Expanded Carrier Screen**

*K. Karimi*

**G27. Two-Site Evaluation of a Rapid and Simple CFTR PCR/CE Assay and Software Targeting Mutations across Diverse Ethnic Groups**

*S. Filipovic-Sadic*

**G28. SMN1 and SMN2 Copy Number Distribution in 733 Clinical Cases of Carrier Screening for Spinal Muscular Atrophy**

*D. Toledo*

**G29. Proof-of-Concept for Single-Platform Trio Carrier Screening of FMR1, SMN1/2, and CFTR Variants Using PCR and Capillary Electrophoresis with Consolidated Workflows**

*W. Laosinchai-Wolf*

**G30. The Single-Tube SLIMamp NGS Assay for Detection of Mutations Associated with Thalassemia Is both Rapid and Robust**

*X. Wu*

**G31. Exploring Mosaic Mutations in Megalencephaly and Other Growth Disorders by Next-Generation Sequencing**

*N. Madkhali*

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**HEMATOPATHOLOGY**

**H01. Personalized Medicine in Practice: Comprehensive Genomic Profiling of a Lung Adenocarcinoma Leads to Reclassification of a Concurrent Lymphoma**

*P. Terraf*

**H02. WITHDRAWN**

**H03. Limitation in Confirming Low Allele Frequency Calls from Sensitive Cancer Assays: MSK Experience with the LiquidPlex cfDNA Panel on Hematologic Samples**

*J. Jeon*

**H04. Somatic Mutation Testing for Pediatric Patients with Known or Suspected Inherited Bone Marrow Failure Syndromes**

*K. Fisher*

**H05. A Highly Reproducible Single-Day FISH Assay for Detection of t(11;14) in Multiple Myeloma Patient Samples**

*A. Prokhorova*

**H06. Comparison of Capture-Based Next-Generation Sequencing Designs in a Clinical Myeloid Neoplasm Panel**

*M. Dina*

**H07. Clinical Utility of a Custom-Designed Next-Generation Sequencing (NGS) Panel for Detection of Gene Fusions, Deletions, and Hotspot Mutations in Myeloid and Lymphoid Neoplasms**

*R. Starks*

**H08. Genomic Landscape of Primary Breast Lymphoma Diffuse Large B-Cell Lymphoma (PB-DLBCL)**

*L. Liu*

**H09. Evaluation of the Ion Torrent OncoPrint Myeloid Sequencing Panel**

*B. Houde*

**H10. Proteomics-Based Biomarkers in Squamous Cell Carcinoma: A Pilot Study Correlating Proteomic Profiles and Tumor Differentiation**

*Y. Chen Wongworawat*

**H11. Detection of Low-Frequency Variants for Minimal Residual Disease (MRD) Monitoring of Acute Myeloid Leukemia**

*N. Valencia*

**H12. A Next-Generation DNA Sequencing Assay for Detection of SNVs, Insertions, Deletions, and Copy Number Variants in 25 Lymphoma Genes in Samples**

*S. Roman*

**H13. High Throughput TRG Sequencing in a Clinical Laboratory: Analysis of Equivocal Results**

*V. Smith*

**H14. CloneRetriever: An Automated Algorithm to Identify Clonal Immunoglobulin Gene Rearrangements by Next-Generation Sequencing**

*E. Halper-Stromberg*

**H15. IGH V-Gene Somatic Hypermutation Assessment by Hybrid-Capture**

*E. Mahe*

**H16. Comparison of Next-Generation Sequencing-Based TRG and TRB Assays for the Diagnostic Evaluation of T Cell Lymphoid Malignancies**

*C. Ho*

**H17. Characterization of the Immunoglobulin Heavy- and Light-Chain Repertoires in a Single Reaction**

*G. Lowman*

**H18. Assessment of a High-Throughput Sequencing Assay for Measurable Residual Disease (MRD) Monitoring in Patients with T-Cell Malignancies**

*J. Tung*

**H19. Improved Clonality and Somatic Hypermutation Analysis of CLL with a Highly Multiplex IGHV Assay**

*M. Toro*

**H20. The Development of an NGS Assay of Immunoglobulin Heavy Variable Gene Somatic Hypermutation in CLL**

*G. Shi*

**H21. Validation of MYD88 L265P ddPCR Assay and Application in Assessment of Primary CNS Lymphoproliferative Disorders**

*M. Cantu*

**H22. An Artificial Intelligence System Applied to Recurrent Cytogenetic Aberrations and Derived Genetic Progression Scores Predicts MYC Rearrangements in Diffuse Large B-Cell Lymphoma.**

*R. Garcia*

**H23. Precise Detection of PDL1/PDL2 Copy Number Alterations in Classic Hodgkin Lymphoma Using Combined CD30 Immunophenotyping and FISH Analysis**

*Y. Zhang*

**H24. Clinical and Genetic Risk Factors Associated with Relapse of Hyperdiploid B-ALL: A Single Institution Review 2001-2019**

*J. Schubert*

**H25. Novel Fusion of *PVT1-RCOR1* in B-Cell Prolymphocytic Leukemia (BCPCLL) Producing False FISH Fusion of *MYC-IGH* with an Atypical Pattern**

*P. Koduru*

**H26. Identification of Clinical Molecular Targets for Childhood Burkitt Lymphoma**

*N. Zeng*

**H27. Characterization of *TP53* Mutations in Myeloid Neoplasms for Targeted Therapy**

*A. Mendiola Romero*

**H28. Chromosome Arm Gain or Loss by Next Generation Sequencing**

*M. Dabrowski*

**H29. Clinical Significance of *CEBPA* Double Mutants: Challenges in Variant Classification and Subtyping of Acute Myeloid Leukemia**

*J. Yoon*

**H30. Cytogenetic and Molecular Landscape in Hispanic Acute Myeloid Leukemia Patients from Puerto Rico**

*P. Deb*

**H31. Evaluation and Follow-up of *JAK2* V617F Positive Patients with Low Allele Burden: A Single-Center Experience**

*K. Reddy*

**H32. Number of Variants and Pathogenic Variants in *ASXL1*, *STAG2*, and *RUNX1* Correlate with High Ogata Score by Flow Cytometry in Myelodysplastic Syndromes: A National Reference Laboratory Experience**

*M. Williams*

**H33. Clinical Implementation of a Custom Myeloid NGS Assay and Overview of *NPM1* and *IDH1/IDH2* Mutation Status in a Clinical Cohort**

*M. Kluk*

**H34. Development of *FIP1L1-PDGFR* Real-time RT-PCR Assay**

*M. Mai*

**H35. *FLT3*-ITD Mutant Allelic Ratio: Impact of Using Non-standardized Published Calculations and Potential Correction Based on Marrow Blast Percentage**

*J. Reinartz*

**H36. Atypical CBFB FISH Signal Patterns Warrant Further Investigation for a True CBFB Rearrangement: An Analysis of 2,425 CBFB FISH Tests**

*R. Yang*

**H37. Curation of *FLT3* Variants in Acute Myeloid Leukemia by Clinical Genome Resource Somatic Hematologic Cancer Taskforce (ClinGen HCT)**

*X. Xu*

**H38. Persistent *IDH* Mutations in AML Patients in Remission on IDH Inhibitors**

*J. Xu*

H39. **Diagnostic Value of Molecular Markers in the Work-up of Myelodysplastic Syndromes**  
*R. He*

H40. **Clinical Validation of Mutant *IDH1* and *IDH2* Detection by Multiplex Digital Droplet PCR**  
*J. Racchumi*

H41. **Comparison of Targeted Myeloproliferative Subpanel versus Comprehensive Myeloid Panel in the Evaluation of Suspected *BCR-ABL1*-Negative Myeloproliferative Neoplasms**  
*D. Morlote*

H42. **Haplotype Phase of *CEBPA* Mutations in Acute Myeloid Leukemia**  
*S. Harley*

H43. **Identifying Non-canonical Mutations in Myeloproliferative Neoplasms: Our Experience with *JAK2* Sequencing**  
*L. Baugh*

H44. **Comparison of Whole Genome Sequencing (WGS) with Conventional Cytogenetics in Profiling Genome-Wide Large-Scale Copy Number and Structural Variations in Pediatric and Adolescent AML**  
*L. Wang*

H45. **Workflow Comparison between Two NCCN Guideline Recommended Myeloproliferative Neoplasms Screening Workup: A Single Institution's Experience**  
*N. Tabish*

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## INFECTIOUS DISEASES

ID01. **Multisite Evaluation of the ARIES MRSA Assay for the Detection of Methicillin-Resistant *Staphylococcus aureus* (MRSA) from Nasal Swabs**  
*B. Buchan*

ID02. **Comparison of a Cartridge-Based Host Gene Expression Test to a Manual Method for Use in the Diagnosis of Sepsis**  
*S. Cermelli*

ID03. **Comparison of Two Multiplex Real-Time PCR Assays for Detection of Tick-Borne Pathogens**  
*T. Uphoff*

ID04. **Development and Performance of a Multiplex Polymerase Chain Reaction (PCR)-Based Assay for Detection of Bacteria in Sterile Body Fluids**  
*C. Johnson*

ID05. **Automated Multiplex Real-Time PCR Detection of *Anaplasma phagocytophilum* and *Ehrlichia chaffeensis* Using the Panther Fusion Open Access System**  
*K. Stellrecht*

ID06. **Automated Real-Time PCR Detection of *Babesia microti* Using the Panther Fusion Open Access System**  
*K. Stellrecht*

ID07. **Evaluation of an Automated rRNA Quantitation System for Rapid AST in Clinical Lab Diagnostics**  
*D. Liu*

**ID08. Prospective Evaluation of a Multiplex HDPCR Tick-Borne Pathogen Panel**

*T. Uphoff*

**ID09. Development of a 29-mRNA Loop Mediated Isothermal Amplification Assay for the Rapid Diagnosis of Acute Infection and Sepsis**

*M. Remmel*

**ID10. *In silico* Performance of a Rapid Sepsis Test in Patients with Candidemia**

*D. Sampson*

**ID11. Development of ViroKey SARS-CoV-2 RT-PCR Test v2.0 for the Sensitive and Accurate Automated Detection of the SARS-CoV-2 Virus**

*I. Ng*

**ID12. Comparison of Four Commercial Molecular Diagnostic Kits for Detection of SARS-CoV-2: A Pilot Study**

*P. Chheda*

**ID13. Evaluation of Ion AmpliSeq SARS-CoV-2 NGS Research Panel**

*W. Liu*

**ID14. Rapid Detection of SARS-CoV-2 Virus via Novel Direct Amplification Methods**

*C. Knox*

**ID15. SARS-CoV-2 Cycle Number as a Metric for Population Trends in New Hampshire**

*E. Bradley*

**ID16. Verification of the Centers for Disease Control and Prevention Real-Time SARS-CoV-2 Assay for Emergency Use Authorization**

*K. Lancor*

**ID17. Analytical Validation of a SARS-CoV-2 Whole Genome Sequencing Method by Amplicon-Based NGS**

*S. Rosenthal*

**ID18. Developing Multiplex of Real-Time PCR Assays for Simultaneous Detection and Differentiation of COVID-19 Plus Flu A and Flu B in a Single Tube Format**

*C. Wang*

**ID19. Comparison of Test Performance of Two Rapid SARS-CoV-2 Viral Assays**

*R. Abdulbaki*

**ID20. Multi-Institutional Evaluation of the Performance of a Rapid Nucleic Acid Amplification Technology for Detecting SARS-CoV-2 on Nasal and Nasopharyngeal Swabs**

*S. Glogowski*

**ID21. Detecting Signatures of SARS-CoV-2 Using Clustered Regularly Interspaced Short Palindromic Repeats (CRISPR)**

*R. Barney*

**ID22. A Practical Comparison of Seven Molecular SARS-CoV-2 Methods**

*C. Gentile*

**ID23. Performance of SARS-CoV-2 Assay in Extraction-Free Method Compared to That of Conventional RNA Extraction Using Automated Instrument**

*S. Kim*

**ID24. The Combination Assay for SARS-CoV-2 and Other Respiratory Viruses in Symptomatic Patients and the Statistical Outcome Visualizing Metrics and Trends**  
*S. Lee*

**ID25. Temporal Spatial Heterogeneity of Immune Response to SARS-CoV-2 Lung Infection**  
*N. Desai*

**ID26. Clinical Performance of Six SARS-CoV-2 Nucleic Acid Amplification Assays in Symptomatic and Asymptomatic Pediatric and Maternal Patient Populations**  
*A. Rahman*

**ID27. Evaluating the Clinical Utility of Next-Generation Sequencing of Nasopharyngeal Specimens for SARS-CoV-2 in the COVID-19 Pandemic**  
*P. Velu*

**ID28. Validation of Saliva Testing for SARS-CoV-2 on Abbott m2000**  
*S. Amin*

**ID29. Validation of an Emergency Use Authorization RT-PCR Test for Detecting SARS-CoV-2 in Upper and Lower Respiratory Tract Specimens**  
*L. Cong*

**ID30. Development of a Multiplexed External Control for Monitoring Performance of a Qualitative Laboratory Nucleic Acid Testing Panel Used for Identification of Respiratory Infections, Including SARS-CoV-2**  
*M. Steffen*

**ID31. Development of a Synthetic External Control for Rapid Detection of SARS-CoV-2 for Use on Xpert Xpress SARS-CoV-2**  
*M. Amadei*

**ID32. Development of a Multiplex Respiratory Panel and a Singleplex SARS-CoV-2 External Control for Use in a Rapid Nucleic Acid Amplification Detection System**  
*J. Salem*

**ID33. Comparison of Two High-Throughput qPCR Assays for SARS-CoV-2**  
*S. Turner*

**ID34. Development of a Multiplexed Synthetic Control for Rapid Detection of SARS-CoV-2 and Other Respiratory Pathogens Using a Nucleic Acid Syndromic Testing Panel**  
*T. Schleicher*

**ID35. Comparison of Nasopharyngeal Swabs and Saliva Samples for the Detection of SARS-Cov-2 RNA**  
*T. McMillen*

**ID36. Strategy for Analysis of Human ACE2 Putative Variants Linked to Protein Structure and Stability: Implications for ACE2 Receptor Binding to SARS-CoV-2**  
*E. Hughes*

**ID37. Comparison of Oral Rinses and Nasopharyngeal Swabs for the Detection of SARS-CoV-2 RNA**  
*T. McMillen*

**ID38. Lung Injury Due to COVID-19 Relative to Influenza and Non-viral ARDS and Normal Controls**  
*A. Borczuk*



**ID39. Evaluation of Sample Pooling for the Detection of SARS-CoV-2 RNA Using the Cobas SARS-CoV-2 Test**

*T. McMillen*

**ID40. RT-PCR Detection of SARS-CoV-2 Infection in Formalin-Fixed, Paraffin-Embedded Tissue Sections in Autopsy Cases**

*D. Berman*

**ID41. The Evaluation of Oropharyngeal Swabs and Saliva Samples for the Detection of SARS-CoV-2 RNA**

*T. McMillen*

**ID42. Evaluation of a Sample-to-Answer Cartridge-Based SARS-CoV-2 Assay**

*J. Lefferts*

**ID43. Viral Sequencing Suggesting Transmission of SARS-CoV-2 from a Patient with False-Negative Molecular Results to Health Care Providers**

*D. Green*

**ID44. Digital Droplet PCR to Detect Low-Titer SARS-CoV-2 in Nasopharyngeal, Nasal, and Salivary Specimens**

*J. Xu*

**ID45. Leveraging Clinical Metagenomic Testing against SARS-CoV-2**

*B. Briggs*

**ID46. A Systematic Review of the Genomic Diversity of SARS-CoV-2 Virus Detected in Dartmouth-Hitchcock Hospital**

*D. Green*

**ID47. Evaluation of Saliva as an Alternative Sample Type for SARS-CoV-2 Detection Using the Hologic Panther Aptima EUA Assay**

*W. Rehrauer*

**ID48. Automated, High-Throughput Testing Using the RealTime SARS-CoV-2 Assay**

*M. Johnston*

**ID49. The Utility of Repeat Testing for Severe Acute Respiratory Syndrome-Coronavirus-2 by Reverse Transcriptase-Polymerase Chain Reaction in Improving Diagnostic Accuracy**

*S. Fathima*

**ID50. Clinical Performance of GenMark ePlex SARS-CoV-2 Test Compared to a Laboratory Developed Procedure**

*J. Laudadio*

**ID51. Evaluation of the SARS-CoV-2 Chromacode EUA Assay**

*L. Thompson*

**ID52. Design and Optimization of Novel ITS2-28s rRNA Gene Primers for Fungal Species Detection from Formalin-Fixed, Paraffin-Embedded Tissues with a Targeted Next-Generation Sequencing Assay**

*G. Wang*

**ID53. Non-invasive Microbial Cell-Free DNA Sequencing Detects Invasive Mold Infections in Immunocompromised Patients with Pneumonia**

*T. Blauwkamp*

**ID54. A Comparative Study of qPCR to a NGS Metagenomics Assay to Detect and Quantify DNA Viruses in Pediatric Bone Marrow Transplant Patients**

*L. Cooper*

**ID55. Investigating Targeted Next-Generation Sequencing of 16S RNA as a Tool for Detecting Shiga Toxin-Producing *E. coli* and *Salmonella* in Ground Beef**

*J. Au-Young*

**ID56. Subtyping of Human Papillomavirus (HPV) Using Next-Generation Sequencing (NGS) Data in Cervical Cancer: A Feasibility Study with Comparison to Conventional Clinical Assays**

*J. Chen*

**ID57. Analytical Performance Characteristics of Galileo ONE: An End-to-End Metagenomics Assay for the Unbiased Sequencing and Bioinformatics Analysis of Microbial DNA and RNA Directly from EDTA Plasma**

*M. Carpenter*

**ID58. Application of Whole-Genome Sequencing for Bacterial Strain Typing in Investigating Hospital Infections**

*K. Park*

**ID59. Validation of ddPCR-Quantified Standards for Use in Viral Load Measurements by NGS**

*D. Hoerres*

**ID60. Performance Evaluation of Abbott Alinity m System to Detect HBV, HCV, and HIV-1 Infections: Comparison with Hologic Panther Aptima System**

*J. Han*

**ID61. Characterization and Evaluation of AcroMetrix HIV, HBV, and HCV Whole Process Quality Controls for Molecular Diagnostic Tests Using Cobas 6800 System**

*H. Wang*

**ID62. WITHDRAWN**

**ID63. Evaluation of a Novel VZV Molecular Assay for Detection of VZV from CSF and Swabs**

*A. Cruz*

**ID64. A Multi-Lab Collaboration for Quantitative BK Virus Test Development on the Fully Automated Cobas 6800/8800 OMNI Utility Channel**

*K. Lebel*

**ID65. Utilization of Digital PCR Assay for the Detection of HPV-16 in Cell-Free DNA in Patients with Head and Neck Cancer at an Oncology Center**

*T. McMillen*

**ID66. Detection of Adenovirus Serotype 7 in a Cancer Patient Population**

*R. Sumner*

**ID67. Retrospective Review of Seasonality of Human Parainfluenza Virus Subtypes at an Oncology Center**

*T. McMillen*

**ID68. Validation of the RealStar Adenovirus Reagents on Plasma and Stool Samples and Comparison to a Laboratory-Developed Test Using the MultiCode Adenovirus Reagents**

*C. Lee*

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## INFORMATICS

I01. **Assessment of RAS Dependency for *BRAF* Mutations Using Real-World Evidence Databases**  
*G. Zheng*

I02. **CarrierSeq, an Expanded Carrier Screening Product Using Next-Generation Sequencing Technology**  
*T. Fahland*

I03. **Evaluation of Roche NAVIFY Mutation Profiler for NGS Variant Annotation and Reporting**  
*P. Ward*

I04. **Optimizing the Detection of Insertions and Deletions Using Next-Generation Sequencing in the Clinical Laboratory**  
*K. Craven*

I05. **Optimizing Reference Mixture Samples for Bioinformatics Pipeline Assessment on Variant Calling Detection for Cancer Diagnostics and Treatment**  
*C. Laing*

I06. **Use and Feasibility of Multi-Algorithmic Consensus-Based Bioinformatics Pipelines in the Detection of Fusions in FFPE Treated Samples**  
*V. Williamson*

I07. **Highly Scalable and Automated Approach to Gut Microbiome Profiling and Quantification Using a New Ion Torrent Next-Generation Sequencing Assay**  
*S. Sarda*

I08. **Microhaplotype Locus-Based Workflow for Sample Contamination Detection in Multiplexed Next-Generation Sequencing (NGS) Assays**  
*J. Balan*

I09. **Prediction of DDR and Other Mutation Signatures Using Panel-Based Sequencing**  
*A. Chellappan*

I10. **Accurate Detection and Quantification of *FLT3* Internal Tandem Duplications in Clinical Hybrid Capture Next-Generation Sequencing Data**  
*J. Tung*

I11. **Identification of Large Deletions Affecting *CTNNB1* Exon 3 in Solid Tumors**  
*Z. Zhang*

I12. **Development of a Clinical Bioinformatics Pipeline for the Comprehensive Genomic Profiling of Patient-Derived Xenograft Tumors**  
*S. Turner*

I13. **Comprehensive Single-Nucleotide, Indel, Structural, and Copy-Number Variant Detection in Human Genomes with PacBio HiFi Reads**  
*W. Rowell*

I14. **Look before You Leap: A Toolkit for Moving Clinical Panels to GRCh38**  
*A. Skol*

**I15. A Novel Machine Learning Approach to Characterize Cancer Signatures for Improved Clinical Reporting**

*S. Shams*

**I16. Pindel as a Back-up INDEL Caller to a GATK4 Mutect2-Based in-House Developed Somatic Secondary Analysis Bioinformatics Pipeline for a Custom Clinical Cancer NGS Panel**

*S. Harada*

**I17. Classification Methods for Germline and Somatic Single Nucleotide Variant (SNV) in Circulating Tumor DNA (ctDNA) of Small Cell (SCLC) and Non-small Cell Lung Cancer (NSCLC)**

*C. Wöstmann*

**I18. Evaluating Machine Learning Methods for Accurate Variant Calling Detection on Acute Myeloid Mutation Analysis**

*C. Laing*

**I19. Noise Reduction Using a Positional Variant-Dependent Error Model for the Detection of Low Frequency Variants in a Pan-Cancer Next-Generation Sequencing Panel**

*C. Laing*

**I20. Clinical Cancer Genomics: Artificial Intelligence Assisted Data Re-analysis to Improve Detection of Potentially Actionable Mutations**

*C. Fischer*

**I21. Capturing and Visualizing Cancer Genomic Data with Category Variants in the JAX Clinical Knowledgebase (JAX-CKB)**

*T. Yin*

**I22. MPath STAR-QC: Automated Quality Control Application for Contamination and Sample Swap Detection Using Short Tandem Repeat Testing**

*S. Lachhander*

**I23. Many NGS-Based Assays, One Platform: Ensuring a High-Quality Case Review and Sign-out Process with NGS Reporter (NGSR)**

*A. Sboner*

**I24. Database for Managing Results of High-Throughput Sequencing Clonality Assays in Clinical Laboratories**

*C. Ho*

**I25. MPath Lab QC: A Centralized Assay Agnostic Approach to Store, Review, and Finalize Laboratory QC for NGS-Based Genomic Clinical Tests**

*A. Agarunov*

**I26. MPath Results PCR: An Integrated Approach to Programmatically Load, Curate and Report Non-NGS Germline Results**

*R. Murray*

**I27. mrLab: Leveraging Mixed Reality in a Precision Medicine Laboratory to Increase Safety and Productivity of Healthcare Workers during the COVID-19 Pandemic**

*A. Sigaras*

**I28. Building a Comprehensive Teaching Repository of Whole Slide Images**

*E. Iriabho*

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## **OTHER (e.g., Education)**

**OTH01. Effect of Implementation of a Medium-Sized NGS Panel and Organ-Specific Subpanels on Send-out Testing: Experiences of a Small, Hospital-Based Molecular Diagnostics Lab**

*R. Kumar*

**OTH02. Establishment of a Multidisciplinary Precision Medicine Lymphoma Tumor Board Incorporating Results of Massively Parallel Sequencing**

*N. Gupta*

**OTH03. Educating in a Pandemic: Rapid Changes to Molecular Genetic Pathology Graduate Medical Education Training during COVID-19**

*F. El-Sharkawy Navarro*

**OTH04. Economic, Operational, and Clinical Considerations in Deploying Rapid NGS for Lung Cancer**

*C. Sande*

**OTH05. Study of the Critical Role Denials, Appeals, and Patient Engagement Play in the Financial Health of Pathology Practices and Molecular Laboratories**

*D. Richard*

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## **SOLID TUMORS**

**ST01. Clinical Application of oncoMonitor: A Simple ctDNA Assay for Liquid Biopsy Monitoring of Treatment and Assessment of Therapy in Colorectal and Lung Cancers**

*M. Minarik*

**ST02. Validation and Performance of Fusion Gene Panel for MiT Family Translocation Renal Cell Carcinomas: Quality of RNA Is Important for Fusion Detection**

*S. Harada*

**ST03. Testing for *CDKN2A* Loss in Infiltrating Gliomas Using Targeted Amplicon-Based Sequencing**

*E. Hissong*

**ST04. Long Mononucleotide Repeat Markers Improve Detection of Microsatellite Instability in Non-colorectal Cancers**

*J. Lin*

**ST05. Comprehensive Coverage of Lung Cancer Somatic Mutations by IntelliPlex Lung Cancer Panel**

*L. Felicioni*

**ST06. Assessment of Microsatellite Instability on a Multi-Racial Cohort of High Grade Prostate Cancer Using Idylla MSI Test**

*M. Rodriguez Pena*

**ST07. CANTRK: A Canadian Multi-Centre *NTRK* Gene Fusion Testing Validation in Solid Tumors Project**

*S. Martins-Filho*

**ST08. De-stained Cytology Smears Can Be Used for Detection of *KRAS* Mutations Using the Biocartis Idylla PCR-Based Molecular Diagnostic Assay**

*Q. Wei*

**ST09. A Next-Generation Sequencing Assay for Comprehensive Genomic Profiling and Identification of Microbial Signatures in Tumor Samples**

*M. Yee*

**ST10. Assessment of PD-L1 Expression in Gastric Tumor Samples**

*P. Scorer*

**ST11. Targeted Mutational Analysis of Predictive and Prognostic Biomarkers in Colorectal Carcinoma**

*G. Huang*

**ST12. Simultaneous Detection of Genetic and Copy-Number Variations in *BRCA1/2* Genes**

*L. Georgieva*

**ST13. Analytical Performance Evaluation of TruSight Oncology 500 (TSO500) ctDNA Kit: A Commercial Next-Generation Sequencing Liquid Biopsy Platform**

*S. Verma*

**ST14. Genomic Test Utilization for Neuroblastoma Risk Classification: A Quality Improvement Project**

*H. Jung*

**ST15. Validation of a Comprehensive, Targeted Next-Generation Sequencing Panel for Solid Tumors**

*E. Barrie*

**ST16. MammaPrint and Blueprint Next-Generation Sequencing (NGS) Results Are Robust and Accurate for Patients with Early Stage Breast Cancer**

*D. Kingma*

**ST17. Evaluation of Three RNA Quantification Methods for Next-Generation Sequencing of Formalin-Fixed, Paraffin-Embedded Tumor Samples**

*D. Chan*

**ST18. An Exome- and Transcriptome-Based NeXT Dx Test Enables Therapy Selection for Cancer Patients and Offers Insight into Emerging Composite Biomarkers for Immunotherapy**

*J. Saldivar*

**ST19. A Comprehensive Approach for Detection of Known and Novel Gene Fusions with RNA Sequencing**

*A. Marcovitz*

**ST20. Cancer-Testis Antigen Detection by Targeted RNA Sequencing**

*J. Conroy*

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**ST22. *FGFR* Gene Mutation Analysis in Urothelial Cancer Using the theascreen *FGFR* RGQ Assay in FFPE Specimen Type**

*L. Cai*

**ST23. Development and Validation of the OncoScreen RNA Panel for the Detection of Gene Fusions and Splice Variants in Tumors**

*B. Li*

**ST24. Benefits of Rapid Genotyping of *KRAS* Mutations versus NGS in Pancreatic Cyst Fluids**  
*A. Farahani*

**ST25. *PIK3CA* Gene Mutation Analysis in Breast Cancer Using the theascreen *PIK3CA* RGQ Assay in FFPE Specimen Type**  
*L.Cai*

**ST26. Detection of Microsatellite Instability Using Anchored Multiplex PCR and Next-Generation Sequencing**  
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**ST27. Clinical and Analytical Validation of the ONCO/Reveal Dx Lung and Colon Cancer Assay (O/RDx-LCCA)**  
*N. Lodato*

**ST28. Noninvasive Genomic Profiling of 113 Patients with Advanced Renal Cell Carcinoma**  
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**ST29. Utilization of a Targeted Next-Generation Sequencing Assay for Assessment of Tumor Cellularity, and Genome-Wide and Gene-Specific Loss of Heterozygosity (LOH)**  
*M. Gupta*

**ST30. Highly Sensitive and Specific Analysis of *PIK3CA* Mutations in Formalin-Fixed, Paraffin-Embedded (FFPE) Samples Using MALDI-TOF Mass Spectrometry**  
*A. Sartori*

**ST31. Internal Validation and Performance Characteristics Using the Oncomine Precision Assay to Detect Multiple Variant Types from Solid and Liquid Biopsy Samples**  
*J. Schageman*

**ST32. Somatic Variant Analysis Using a Pan-Solid Tumor Expanded Gene Panel**  
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**ST33. Evaluation of a Mass Spectrometry-Based *PIK3CA* Mutation Assay for Predictive Breast Cancer Therapeutic Decision Making**  
*A. Box*

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*N. Willard*

**ST35. Identifying Prognostic and Predictive Gene Alterations in Metastatic Prostate Cancer**  
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*J. Huang*

**ST37. Clinical Validation of an Automated 170 Gene Panel Workflow in a CAP/CLIA Laboratory for Solid Tumors**  
*S. Deharvengt*

**ST38. An RNA Sequencing Panel for Detection of Fusions and Splice Site Variants in Solid Tumors**  
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**ST39. Uncovering Subsets of Non-small Cell Lung Cancer (NSCLC) Enriched in Mutations in Cytoskeletal Dynamics and DNA Repair Genes: Additive Value of Large Gene Panels for Clinical Tumor Profiling**

*H. Tu*

**ST40. Rapid qPCR Testing in the NGS Era Enables Same-Day Resulting of *EGFR* Mutant NSCLC**

*N. Z. Georgantas*

**ST41. Rapid Assessment of Microsatellite Instability across a Spectrum of Tumor Types Using the Idylla System**

*A. Momeni-Boroujeni*

**ST42. DNA Methylation Profiling of DNA Extracted from Archived Stained Tissue Slides for Central Nervous System Tumor Diagnostics**

*Z. Abdullaev*

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*M. Lin*

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*S. Harada*

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*R. Ondrasik*

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*A. Reddy*

**ST48. Tumor Microbiome in Colorectal Carcinoma: Bacterial Enrichment Is Associated with Oncogenic Variants within Specific Signaling Pathways**

*C. Beech*

**ST49. *NKX2-1* Gene Variants in Solid Tumors: The Spectrum and Potential Impact in Surgical Pathology Diagnosis**

*F. El-Sharkawy Navarro*

**ST50. Comprehensive Genomic Profiling of Different Subsets of Merkel Cell Carcinoma: Insights on Pathogenetic Pathways**

*R. DeCoste*

**ST51. Correlation between MMR IHC and MSI Testing for Detection of MSI-High Solid Tumors**

*M. Shirazi*

**ST52. Circulating Tumor DNA Genomic and Methylation Profiling in Advanced Non-small Cell Lung Cancer Patients**

*J. Qin*

**ST53. Systemic Review of the Clinical Utility of Fluorescence *in situ* Hybridization (FISH) Testing**

*Y. Lo*

**ST54. Commercial Tissue-Based Genomic Profiling on Breast Cancer and Its Impact on Clinical Decision Making: A Single Institution Experience**

*J. Chen*



**ST55. Comprehensive Genomic Profiling in Patients with Advanced Cancer in a Large US Healthcare System**

*B. Piening*

**ST56. Development of Quality Control Reference Materials for Microsatellite Instability (MSI) Testing**

*C. Huang*

**ST57. Mutated Allele Frequency and *NRAS* Mutational Status Are Significantly Associated with High-Risk Prognosis by 31-Gene Expression Profile**

*F. Monzon*

**ST58. Genomic Profiling Uncovers Mutation Signatures That Differentiate Pediatric Rhabdomyosarcoma (RMS) Subgroups and Predict Clinical Outcomes**

*F. Lin*

**ST59. Detection of Actionable Alterations in Breast and Ovarian Tumor Tissues by Testing with a 50-gene NGS Panel**

*C. Ma*

**ST60. Microsatellite Instability Testing for Lynch Syndrome Screening in Colorectal Adenomas**

*A. Javanbakht*

**ST61. Detection and Interpretation of Canonical and Cryptic Splice Sites in Solid Tumors and Their Relevance to FDA Approved Therapies and Clinical Trials**

*E. Bogdanova*

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*H. Chen*

**ST63. Gene Expression Profile of Sex Cord Stromal Cell Tumors and Their Relevance to Prognosis**

*P. Bhattacharyya*

**ST64. Expression Profiling Reveals Novel Molecular Signature in Pleomorphic Lobular Carcinoma *in situ***

*E. Makhoul*

**ST65. Aberrant *PAX3* (*Paired Box Gene 3*) RNA Splicing Is a Potential Marker for Diagnosis of Melanoma**

*I. Kasago*

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*L. Hess*

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*M. Marques*

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*E. Rudzinski*

**ST69. Biomarker Testing and Overall Survival among Patients Diagnosed with Advanced or Metastatic Non-small Cell Lung Cancer**

*L. Hess*

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*P. Quon*

**ST71. Clinical Characteristics of *RET*- and *NTRK*-Rearranged Tumors in a Single Tertiary Cancer Center**

*T. Vougiouklakis*

**ST72. Incidence of T790M Mutation by ddPCR in Patients Progressing on First- and Second-Generation TKIs and Clinical Outcomes on Osimertinib**

*S. Nathany*

**ST73. Frequency of *EGFR* Mutations and ALK Expression in NSCLC in the North of México**

*B. Montaña Miyagui*

**ST74. Tissue Requirements of a Novel 27-Gene Immuno-Oncology Algorithm Measuring Tumor Microenvironment to Predict Response to Immunotherapies**

*T. Nielsen*

**ST75. Pan-Cancer Liquid Biopsy Assay for Mutation Profiling in 61 Genes by Low-Depth Sequencing**

*P. Hao*

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**ST77. Spatially Resolved Gene Expression Profiles in Human Glioblastoma**

*A. Hartnett*

**ST78. Spatially Resolved Molecular Interrogation of Triple Negative Breast Cancer**

*S. Williams*

**ST79. Quantitative Assessment of Functional Activity of Multiple Signaling Pathways in Recurrent Breast Cancer with Low to Intermediate 21 Gene Recurrence Score**

*L. Lin*

**ST80. Prospective Study Using Virtual Enrollment to Assess an RNA-FIT Assay for Non-invasive Detection of Colorectal Cancer, Advanced Adenomas, and Other Precancerous Adenomas**

*E. Barnell*

**ST81. *NAB2-STAT6* Gene Fusions to Identify Primary/Metastasis Hemangiopericytoma/Solitary Fibrous Tumors**

*N. Singh*

**ST82. Development and Performance of Formalin Compromised FFPE Reference Materials**

*O. Clement*

**ST83. CNV Detection from a Multi-Cancer NGS Panel: A Single-Tube, Multiplex-PCR Based NGS with 309 Tiled Amplicons**

*A. LaBonte*

**ST84. Rapid Isolation of High-Quality Ultra-High Molecular Weight Genomic DNA from Blood, Bone Marrow Aspirates, and Fresh Frozen Human Tumors**

*H. Sadowski*

**ST85. Novel Amplicon-Based NGS Library Preparation Protocols Compared and Evaluated across Two Sequencing Technologies**

*E. Petrilli*

**ST86. Chromosomal Microarray Analysis of Benign Mesenchymal Tumors with *RB1* Deletion**

*A. Dusenbery*

**ST87. A Novel Nanoparticle-Based Approach to Improve Extraction of Circulating Tumor DNA (ctDNA)**

*E. Williams*

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**TT01. Dimensionality Reduction for Noise Filtering of Big Data Sets**

*E. Mahe*

**TT02. Molecular Profiling in Challenging Oncology Research Samples Using a Novel Library Preparation Chemistry**

*H. Huang*

**TT03. Droplet Digital-PCR (ddPCR) as Confirmatory Method for Low Allelic Frequency Variants Detected by Manual Review of Data in Clinical NGS Testing**

*W. Song*

**TT04. Performance Validation of Magnis BR: A Full-Automatic Capture-Based Library Preparation Platform for Next-Generation Sequencing (NGS)**

*X. Wang*

**TT05. From Plasma to Variants: A Fully Automated Workflow Solution for Low-Frequency Variant Detection in Cell-Free DNA**

*T. Barnes*

**TT06. Concordance of Variant Detection between the MoCha ctDNA Assay and Matched Tissue Biopsy in Non-small Cell Lung Cancer**

*C. Karlovich*

**TT07. Reproducibility of Allelic Fractions of Genomic Variants from Colorectal and Lung Cancer Tissue Downstream of DNA Extraction**

*M. Javey*

**TT08. Flexible and Complete Exome Next-Generation Sequencing (NGS) Solution for Variants Detection with Improved Human Exome Panel**

*M. Hong*

**TT09. DNA Samples with Low Concentration Can Benefit from Speed Vacuum Concentration in NGS Testing**

*W. Song*

**TT10. Comparative Study of Three Assays: Target Capture Sequencing, MassARRAY and Real-Time qPCR for Testing Somatic Mutations in Plasma Cell-Free Circulation Tumour DNA of Non-small Cell Lung Cancer**

*L. Zhang*

**TT11. Workflow Evaluation: Impact of Specimen Storage and Transport on ccfRNA Multiplex Analysis in Dedicated Blood Collection Tubes**

*T. Voss*

**TT12. Reference Materials for Measurable Residual Disease (MRD) Monitoring in Circulating Cell-Free DNA (ccfDNA)**

*Y. Konigshofer*

**TT13. Automation of Fluorescence *in situ* Hybridization Processing and Digital Analysis**

*M. Azim*

**TT14. A Modified Vendor Extraction Protocol Better Preserves the Structural Integrity of Genomic DNA Extracted from FFPE Tissue**

*C. Artymiuk*

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**TT16. Detection of the Mutational Status in Colorectal Cancer from Formalin-Fixed, Paraffin-Embedded (FFPE) Tissue**

*E. Haenssler*

**TT17. Centrifugation and RBC Lysis-Free Preparation of Blood Samples in less than 30 Minutes**

*C. Barr*

**TT18. A Complete Yet Flexible Workflow for Library Preparation and Analysis with Enhanced Error Correction for Low Input FFPE Tissue Biopsy and Circulating Tumor DNA Samples**

*S. Lee*

**TT19. Next-Day Analysis from Specimen to Variant Calling with the Genexus System**

*J. Gioia*

**TT20. Pushing the Limits of Cancer Research: An Integrated and Automatic Workflow on Ion Torrent Genexus System from Nucleic Acid Extraction to Next-Generation Sequencing**

*R. Cao*

**TT21. Analytical Performance Testing of the MoCha Circulating Tumor DNA Assay**

*R. Harrington*

**TT22. Digital PCR Paired with High-Speed AFM for Quantitation and Length Analysis of DNA Length Polymorphisms**

*S. Koebley*

**TT23. Focused-Ultrasonication Driven High-Quality DNA and RNA Extraction and Purification from FFPE Samples**

*K. Amirault*

**TT24. Evaluating Effects of PCR Instruments and Temperature Ramp Rates to Base-Composition Bias in TruSight Oncology 500 (TSO500) Panel**

*W. Song*

**TT25. Evaluation of the Biocartis Idylla ctEGFR Mutation Assay on Samples with DNA Concentrations Insufficient for Next-Generation Sequencing (NGS)**

*W. Keegan*

**TT26. Analysis of Simple and Complex Variants and Biomarkers for Comprehensive Genomic Profiling (CGP) of Solid Tumors and Hematologic Malignancies Using a Single NGS Workflow from FFPE and cfDNA Samples**

*R. Samara*

**TT27. Matrix and DNA Source of Reference Material Significantly Affect Extraction Recovery, Drift in qPCR Quantitation, Assay Precision, and Limit of Detection (LOD) in Validation of ctDNA Assay**

*Y. Lu*

**TT28. Archival FFPE and DNA Quality: Optimal Storage Time and Predictive Metrics for Next-Generation Sequencing**

*V. Parimi (Parini)*

**TT29. Confirmation of Fusions Detected with Sequencing**

*D. Jones*

**TT30. Development of a Universal Probe System for Droplet Digital PCR**

*D. Jones*

**TT31. Targeting Clinically Significant “Dark” Regions of the Human Genome with High-Accuracy Long-Read Sequencing**

*C. Heiner*

**TT32. Performance of GeoMx CTA and WTA, High-Plex, Spatial Gene Expression Profiling Tools**

*K. Sorg*

**TT33. Standardizing Plasma ctDNA Measurements Using SNAQ-SEQ ONCO1LB Internal Controls**

*S. Deharvengt*

**TT34. Optical Mapping Enables High-Throughput Analysis of Pathogenic Repeats**

*E. Lam*