Cancer Genomics Consortium, 14th Annual Meeting

August 13-16, 2023
Hyatt Regency at the Arch, St. Louis, Missouri

SUNDAY, AUGUST 13, 2023

Pre-Meeting Workshops

9:00 – 10:30 AM **Bioinformatics Workshop**

10:30 – 10:45 AM **Coffee Break for Workshop Attendees**

10:45 AM – 12:15 PM **Case-Based Workshop**

Opening of the 14th Annual Cancer Genomics Consortium

1:00 – 1:05 PM **Welcome**
Rashmi Kanagal-Shamanna, *MD Anderson Cancer Center*

1:05 - 1:15 PM **Presidential Address**
Obi Griffith, *Washington University School of Medicine*

1:15 – 2:15 PM **Keynote Presentation**
John DiPersio, *Washington University School of Medicine*

2:15 – 2:30 PM **Session 1: Applications of Emerging Technologies in Clinical Genomics 1**

TBD

Application of optical genome mapping to identify samples with homologous recombination deficiency
Alex Hastie, *Bionano Laboratories*

Clinical Validation of Plasma Whole Genome Sequencing for Detection of Minimal Residual Disease from Solid Tumours
Felix Beaudry, *Ontario Institute for Cancer Research*

Comprehensive next generation cytogenomics improves risk stratification of Acute Myeloid Leukemia
Stephen Eacker, *Phase Genomics/Fred Hutchinson Cancer Center*

3:15 – 4:00 PM **Coffee Break**

4:00 – 4:30 PM **Invited Speaker Presentation**
Robert Hasserjian, *Mass General Brigham*

4:30 – 5:30 PM **Session 2: Bioinformatics, Artificial Intelligence, Machine-Learning 1**

Overcoming challenges in semantic alignment of therapeutics knowledge using Therapy
James Stevenson, *Nationwide Children's Hospital*

Tracking Immunotherapy Response with Single Cell T Cell Receptor Profiling in Canine Models of Cancer
Obi Griffith, *Washington University School of Medicine*

AI-Based Algorithms for Neoplastic Metaphase Cells Boost Efficiencies in the Cytogenetics Laboratory
Bo Hong, *ARUP Laboratories*

Mapping variants from multiplex assays of variant effect (MAVEs) to human reference sequences
Jeremy Arbesfeld, *The Ohio State University*

5:30 – 6:00 PM **Speed Abstracts Session A**

Evaluation of Hi-C versus Optical Genome Mapping for Diagnosing Constitutional Genomic Structural Variants
He Fang, *University of Washington*

Personalized sequencing assays for cerebrospinal fluid liquid biopsies in children with brain tumors
Katherine Miller, *Nationwide Children's Hospital*

HPV forms chimeric virus-human transcripts that affect host gene expression in cervical tumors
Kay Jayachandran, *Washington University School of Medicine*

Concurrent Systemic Mastocytosis and T-Lymphoblastic Lymphoma Unified by a Novel Cryptic JAKMIP2::PDGFRB Rearrangement
Kevin Shopsowitz, *University of British Columbia*

Loss of MSH2 and MSH6 is frequently observed in prostate neoplasms with mismatch repair deficiency
Gokce Toruner, *The University of Texas MD Anderson Cancer Center*

6:00 – 6:15 PM **Platinum Vendor Showcase**
(No CME or CEUs available)

6:15 – 6:45 PM **Diamond Vendor Showcase**
(No CME or CEUs available)

6:45 – 7:00 PM **Platinum Vendor Showcase**
(No CME or CEUs available)

7:15 - 9:00 PM **Welcome Reception**
Exhibit Hall

MONDAY, AUGUST 14, 2023

8:00 – 9:00 AM **Session 3: Genomic resources for variant curation and standardization 1**

ClinGen Cancer Variant Interpretation (CVI) Committee: Pilot Guidance for Somatic Cancer Variant Curation Expert Panels
Deborah Ritter, *Baylor College of Medicine*

Djerba: A Modular System to Generate Clinical Genome Interpretation Reports for Cancer
Iain Bancarz, *Ontario Institute for Cancer Research*

Investigation of pathogenic and truncated variants of RUNX1 and DDX41 in All of Us
Huan Mo, *National Human Genome Research Institute*

Developing a generalized model for variants in CIViC
Arpad Danos, *Washington University in St. Louis*

9:00 – 10:00 AM **Keynote Presentation**
Olufunmilayo Olopade, *University of Chicago*

10:00 – 10:45 AM **Coffee Break**

10:45 – 11:45 AM **Session 4: Solid Tumors 1**

Comparative analysis of RNA expression identifies druggable targets in difficult-to-treat pediatric solid tumors
Yvonne Vasquez, *University of California, Santa Cruz*

Comprehensive 'Omic' Profiling Reveals 'Atypical Oligodendrogliomas' which Challenge CNS Diagnostic Classification
Adrian Dubuc, *Brigham and Women's Hospital*

Cell-free DNA genomic and epigenomic analysis to predict survival in mCRPC patients treated with AR-directed therapy
Predeep Chauhan, *Washington University School of Medicine*

Optical Genome Mapping Reveals New Insights into ZFTA Fusion in Supratentorial Ependymomas
Jianling Ji, *Children's Hospital Los Angeles, USC*

11:45 – 12:15 PM **Speed Abstracts Session B**

Clinical Utility of Optical Genome Mapping: Comparison with Standard Cytogenomics Work-up for Hematological Malignancies
Gokce Toruner, *The University of Texas MD Anderson Cancer Center*

High-risk genetic variants underlie unfavorable prognosis of B-lymphoblastic leukemia patients of Hispanic ethnicity

Wengyn Maximilian, *Children's Hospital Los Angeles*

Improving interoperability of therapeutics and their targets for clinical and precision medicine applications
Matthew Cannon, *Nationwide Children's Hospital*

Five-year Experience of Evaluating Individuals At-risk for Underlying Genetic Predisposition to Hematologic Malignancy
Min Fang, *Fred Hutchinson Cancer Center*

Analytical validation of an optical genome mapping assay for structural variant detection in hematologic malignancies
Trilochan Sahoo, *Bionano Laboratories*

12:15 – 12:30 PM **Platinum Vendor Showcase**
(No CME or CEUs available)

12:30 – 1:00 PM **Diamond Vendor Showcase**
(No CME or CEUs available)

1:00 – 2:00 PM **Buffet Lunch**
(Exhibit Hall)

2:00 – 3:00 PM **Session 5: Hematological Malignancies 1**

Real-World Analysis of Cytopenic Patients for Identification of Clonal Cytopenia(s) of Undetermined Significance (CCUS)
Anwar Iqbal, *University of Rochester Medical Center*

Chromosomal Microarray Analysis Work-up for Hypocellular MDS Patients with Inconclusive Cytogenetics
Ha Nguyen, *Northwestern Medicine*

Prognostic significance of copy number gain of MYC detected by FISH analysis in large B-cell lymphoma
Madina Sukhanova, *Northwestern Medicine Feinberg School of Medicine*

Prognostic significance of copy number gain of MYC detected by FISH analysis in large B-cell lymphoma
Victória Tomaz, *Hospital Israelita Albert Einstein*

3:00 – 3:30 PM **Invited Speaker Presentation**
Francesc Sole, *Josep Carreras Leukaemia Research Institute*

3:30 – 4:00 PM **Session 6: Others (Bioinformatics and Genomic Resources)**

Optical Genome Mapping identifies additional cytogenetic abnormalities in patients with hematologic malignancies
Sachin Jadhav, *Healthcare Global*

Comprehensive Genomic Characterization of Infantile Cancers Reveals High Yield of Therapeutically Targetable Alterations
Mariam Mathew, *Nationwide Children's Hospital*

4:00 – 4:45 PM **Coffee Break**

4:45 – 5:00 PM **Platinum Vendor Showcase**
(No CME or CEUs available)

5:00 – 5:15 PM **Platinum Vendor Showcase**
(No CME or CEUs available)

5:15 – 5:45 PM **CGC Scientific and Program Updates**

Implementation survey of the ACMG/CGC standards for interpretation of acquired CNAs and CN-LOH in neoplastic disorders
Fady Mikhail, *University of Alabama at Birmingham*

Implementation survey of the ACMG/CGC standards for interpretation of acquired CNAs and CN-LOH in neoplastic disorders
Kathleen Schieffer, *Nationwide Children's Hospital*

Early Career / CCGA / Webinar Overview

5:45 – 6:15 PM **Spotlight Symposium: ISCN Nomenclature**
Rosalind Hastings, *ISCN Standing Committee Chair & GenQA Consultant Oxford University Hospitals NHS Foundation Trust*

6:15 – 7:45 PM **Poster Viewing Session**
Exhibit Hall

Early Career Social
For attendees in training or recently out of training

TUESDAY, AUGUST 15, 2023

8:00 – 9:00 AM **Session 7: Bioinformatics, Artificial Intelligence, Machine Learning 2**

Cell-free DNA fragmentation profiling as a method for tumor fraction assessment and treatment monitoring in NSCLC
Zachary Skidmore, *Delfi Diagnostics*

Resolving Ambiguities in Copy Number Variation Representation
Kori Kuzma, *Nationwide Children's Hospital*

Automated Deep Aberration Detection from Chromosome Karyotype Images
Min Fang, *Fred Hutchinson Cancer Research Center*

Fusion Curation Interface: an educational tool to explore a unified framework for representing & curating gene fusions
Kathryn Stahl, *Nationwide Children's Hospital*

9:00 – 10:00 AM **Keynote Presentation**
Guillermo Garcia-Manero, *MD Anderson Cancer Center*

10:00 – 10:45 AM **Coffee Break**

10:45 – 11:45 AM **Session 8: Hematological Malignancies 2**

Frequency and Etiology of Cytogenetically Cryptic Oncogenic Fusions in Pediatric AML
Gordana Raca, *Children's Hospital Los Angeles*

Clonal Hematopoiesis in Childhood Cancer Survivors
Irenaeus Chan, *Washington University in St. Louis – School of Medicine*

Integrative Cytogenetic and Molecular Studies Unmasks 'Chromosomal Mimicry' in Hematologic Malignancies
Adrian Dubuc, *Brigham and Women's Hospital*

Optical genome mapping in hematological malignancy: Clinical outcomes in a 2-year follow-up retrospective study
Nikhil Sahajpal, *Greenwood Genetic Center*

11:45 – 12:15 PM **Speed Abstracts Session C**

Evolution of a variant curation procedures in the open-access cancer variant interpretation knowledgebase CIViC
Kilannin Krysiak, *Washington University School of Medicine*

Gene Normalizer: a tool to resolve genetic ambiguity through data harmonization
Anastasia Smith, *The Ohio State University*

Cell-type-specific genetic-to-epigenetic relationships in the human breast
Axel Hauduc, *University of British Columbia*

Assessment of TRG and TRB Clonality by NGS of Dermatologic Specimens is Impacted by Biopsy Type, DNA and Amplicon Sizes
Jane Houldsworth, *Icahn School of Medicine at Mount Sinai*

Genomic microarray analysis reveals heterogeneity in high hyperdiploid B-cell acute lymphoblastic leukemia
Julie Feusier, *ARUP Laboratories – Phoenix Children's*

12:15 – 12:30 PM **Platinum Vendor Showcase**
(No CME or CEUs available)

12:30 – 1:00 PM **Diamond Vendor Showcase**
(No CME or CEUs available)

1:00 – 2:15 PM **Round Table Discussions with Lunch**
Please sign up for table topics during conference registration.

2:15 – 3:15 PM **Session 9: Applications of Emerging Technologies in Clinical Genomics 2**

Targeted RNA-Seq on fresh frozen and methanol/acetic acid fixed cells in diagnostic workup of hematologic malignancies
Xiaoyu Qu, *Fred Hutchinson Cancer Center*

A novel method for detection of loss of heterozygosity using B-allele frequency from optical genome mapping data
Aliz Raksi, *Bionano Laboratories – Tesa Consulting*

Analysis of Gene Rearrangements in Neoplasms with Hi-C Sequencing Using Fresh-Frozen and FFPE Specimens
Yajuan Liu, *University of Washington-Seattle*

Rare SRY-positive derivative X chromosome in female fetus with apparently normal development
Casey Brewer, *Cincinnati Children's Hospital Medical Center*

3:15 – 3:45 PM **Invited Speaker Presentation**
Joseph Khoury, University of Nebraska Medical Center

3:45 – 4:30 PM **Coffee Break**

4:30 – 5:00 PM **Diamond Vendor Showcase**
(No CME or CEUs available)

5:00 – 5:15 PM **Platinum Vendor Showcase**
(No CME or CEUs available)

5:15 – 5:45 PM **Speed Abstracts Session D**

ClinGen Pediatric Cancer Taskforce initiatives to advance pediatric clinical interpretations through expert curation
Jason Saliba, *Washington University School of Medicine*

Uveal Melanoma - The New Zealand Perspective
Amanda Dixon-McIver, *IGENZ*

VMD4Kids: A highly sensitive NGS panel to detect low-level mosaic variants in vascular anomalies & overgrowth disorders.
Avinash Dharmadhikari, *Children's Hospital Los Angeles*

Clinical validation and Implementation of exome, transcriptome and whole genome sequencing for pediatric cancers
Alexandre Rouette, *CHU Sainte-Justine – Molecular Diagnostic Lab*

Whole-exome sequencing identifies somatic mutations penile squamous cell carcinoma
Kelly Duarte, *University of Sao Paulo*

5:45 – 6:15 PM **Spotlight Symposium: GOAL Consortium**
Dara Aisner, *University of Colorado*
Jeremy Segal, *University of Chicago*

6:00 – 9:00 PM **Optional After Hours Social Activities**

WEDNESDAY, AUGUST 16, 2023

8:30 – 9:30 AM **Session 10: Genomic resources for variant curation and standardization 2**

Curating Variants of Established Clinical Significance
Mariam Khanfar, *Washington University School of Medicine*

Implementing the ClinGen/CGC/VICC Oncogenicity Guidelines in a Pediatric Variant Classification Workflow
Wesley Goar, *Nationwide Children's Hospital*

Oncogenic assessment of FLT3 Variants by the ClinGen FLT3 Somatic Cancer Variant Curation Expert Panel
Jason Saliba, *Washinton University School of Medicine*

Feasibility, Accuracy and usability analysis of MapAML, a first-in-class app for integrated diagnosis in AML
Thais Moyen, *Hospital Israelita Albert Einstein*

9:30 – 10:30 AM **Keynote Speaker**
David Beck, *Columbia University*

10:30 – 11:15 AM **Coffee Break**

11:15 – 12:15 PM **Session 11: Solid Tumors 2**

A cell-free DNA 5-hydroxymethylcytosine marker predicts immunotherapy response in lung cancer
Zejuan Li, *Houston Methodist*

Profiling PIK3CA Variants - a highlight of C2 domain variants in Disorders of Somatic Mosaicism
Yang Cao, *Washington University in St. Louis*

A female-specific chimeric RNA with differential expression in COVID patients
Xinrui Shi, *University of Virginia*

Utilizing Rapid Molecular Testing to Reduce Disparites in Pediatric Cancer in Sub-Saharan Africa
Julie Gastier-Foster, *Baylor College of Medicine/Texas Children's Hospital*

12:15 – 12:45 PM **CGC 2023 Business Meeting**