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: Exploring the Clinical Interpretation Resource Landscape**
Moderators: Alex Wagner, *Nationwide Children's Hospital*

Presenters:
Beth Pitel, *Mayo Clinic*
Brendan Reardon, *Dana Farber Cancer Center*
Moriel Singer-Berk, *Broad Institute*

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

10:45 AM – 12:15 PM **Case-Based Workshop: Assessment of Genomic Changes in Clinical Cases: Things to Think About and How We Approached Them**
Moderators: Teresa Smolarek, *Cincinnati Children's Hospital Medical Center and* Celeste Eno, *Cedars Sinai*

Presenters:
Lisa Brailey, *Mt. Sinai School of Medicine*
Celeste Eno, *Cedars-Sinai*
Jinbo Fan, *University of Virginia*
Elena Repnikova, *University of Missouri-Columbia*
Teresa Smolarek, *Cincinnati Children's Hospital Medical Center*

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: CAR-T Therapies for T-cell Malignancies
John DiPersio, *Washington University School of Medicine*
Introduction: Obi Griffith*, Washington University School of Medicine*

2:15 – 3:15 PM

Session 1: Applications of Emerging Technologies in Clinical Genomics

Moderators: Jane Houldsworth, *Icahn School of Medicine at Mount Sinai* and Vivek, Gupta, *Government Institute of Medical Sciences, India*

Clinical validation of plasma whole genome sequencing for detection of minimal residual disease from solid tumours
Felix Beaudry, *Ontario Institute for Cancer Research*

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

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

Comprehensive next generation cytogenomics improves risk stratification of Acute Myeloid Leukemia
Stephen Eacker, *Phase Genomics*

3:15 – 4:00 PM

Coffee Break
Science Café Presentations:

- **Agilent**
- **LOXO@Lilly**
- **Genome Insight, Erin Strong**

4:00 – 4:30 PM

Invited Presentation: *NPM1* Mutation in Myeloid Malignancies: Advancing Precision Medicine Through Molecular Testing
Robert Hasserjian, *Mass General Brigham*
Introduction: Kristin Deeb, *Emory University*

4:30 – 5:30 PM

Session 2: Advances in Cancer Informatics
Moderators: Alex Wagner, *Nationwide Children's Hospital and*
Brendan Reardon*, Dana-Farber Cancer Institute*

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

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*
Trainee Award

5:30 – 6:15 PM **CGC Scientific and Program Updates**
Moderator: Rashmi Kanagal-Shamanna, *MD Anderson Cancer Center*

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*

Current State of Diagnostic Testing in Pediatric Sarcoma: Practical Solutions to Diagnostic Challenges
Kathleen Schieffer, *Nationwide Children's Hospital*

Early Career Program
Rebecca Smith, *Vanderbilt University Medical Center*

CGC Education Program
Teresa Smolarek, *Cincinnati Children's Hospital Medical Center*

6:15 – 6:45 PM **Speed Abstracts Session I: Use of Different Molecular Techniques to Diagnose Cancer**
Moderator: Paulo Campregher, *Hospital Israelita Albert Einstein*
(No CME or CEUs available)

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

Endothelial cells are a key target of IFN-g during response to combined PD-1/CTLA-4 ICB treatment in bladder cancer
Sharon Freshour, *Washington University School of Medicine*

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*

6:45 – 7:00 PM **Platinum Vendor Showcase: Pacific BioSciences (PacBio)**
A New Age in Cancer Genomics: From Single Cell Transcriptomics to Liquid Biopsy
Jonathan Bibliowicz, *Pacific BioSciences*
(No CME or CEUs available)

7:00 - 8:30 PM **Welcome Reception**
CGC Exhibit Hall, Grand Ballroom ABCD

MONDAY, AUGUST 14, 2023

8:00 – 9:00 AM

Session 3: Genomic Resources for Variant Curation and Standardization
Moderators: Melissa Cline, *UC Santa Cruz Genomics Institute* and Kilannin Krysiak, *Washington University School of Medicine*

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: Heterogeneity of breast cancer genomes: Going beyond therapy to risk assessment and prevention
Olufunmilayo Olopade, *University of Chicago Medicine*
Introduction: Gordana Raca, *Children's Hospital Los Angeles*

10:00 – 10:45 AM

Coffee Break with Exhibitors
Science Café Presentation: BioCartis

10:45 – 11:45 AM

Session 4: Clinical Utility of Genomic Testing for Identification and Potential for Therapy Selection in Solid Tumors
Moderators: Teresa Smolarek, *Cincinnati Children's Hospital Medical Center* and Sadif Saba, *Cincinnati Children's Hospital Medical*

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
Pradeep 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 AM – 12:15 PM

Speed Abstracts Session II
Moderator: Emilie Lalonde, *London Health Sciences Center*
(No CME or CEUs available)

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: Thermo Fisher Scientific**
Using OncoScan to Address Challenges Calling Genomic Instability in Solid Tumor Samples.
Caleb Kidwell, *Quantigen BioSciences*
Mary Napier, *Thermo Fisher Scientific*
(No CME or CEUs available)

12:30 – 1:00 PM **Diamond Vendor Showcase: Bionano Genomics**
Unleashing Genomic Insights that Matter with Optical Genome Mapping: An Interactive Panel Discussion
Moderator: Alka Chaubey, *Bionano Genomics*
Panelists:
Yassmine Akkari, *Nationwide Children's Hospital*
Ravindra Kolhe, *Augusta University*
Sachin Jadhav, *HealthCare Global*
(No CME or CEUs available)

1:00 – 2:00 PM **Buffet Lunch**
Exhibit Hall and Foyer

2:00 – 3:00 PM **Session 5: Hematological Malignancies**
Moderator: Gordana Raca, *Children's Hospital Los Angeles and*
Patricia Hernandez, *Washington University in St. Louis*

Real-World Analysis of Cytopenic Patients for Identification of Clonal Cytopenia(s) of Undetermined Significance (CCUS)
Rashmi Kanagal-Shamanna, *MD Anderson Cancer Center*

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

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*

Whole transcriptome sequencing as a diagnostic tool for AML
Victória Tomaz, *Hospital Israelita Albert Einstein*
Trainee Award

3:00 – 3:30 PM **Invited Speaker Presentation: Cytogenetics is Still Useful in the Era of Genomics**
Francesc Sole, *Josep Carreras Leukaemia Research Institute*
Introduction: Brynn Levy, *Columbia University School of Medicine*

3:30 – 4:00 PM **Session 6: Integration of Novel Technologies in the Clinical Cancer Genomics Laboratory for Improving Patient Care**
Moderator: Angela M. Lager, *University of Chicago*

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 with Exhibitors**
Science Café Presentations:

- **Oxford Gene Technology (OGT),** Jace Doshier
- **Velsera**
- **Abbvie**

4:45 – 5:00 PM **Platinum Vendor Showcase: Novartis**
The Growing Role of Molecular Diagnostics in Cancer Care
Roger Bishop, *Novartis Precision Medicine Liaison*
(No CME or CEUs available)

5:00 – 5:30 PM **Speed Abstracts Session III: Analytic and Post Analytic Improvements in Genomics for Establishment of Efficient Clinical Workflows and Management in Patient Care**
Moderator: Meenakshi Mehrota, *Mount Sinai Hospital*
(No CME or CEUs available)

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*

5:30 – 6:00 PM **Spotlight Symposium: ISCN 2024 update including a preview of the new genomic mapping nomenclature**
Rosalind Hastings, *ISCN Standing Committee Chair & GenQA Consultant Oxford University Hospitals NHS Foundation Trust*
Introduction: Alex Wagner, *Nationwide Children's Hospital*

6:00 – 7:00 PM **Poster Viewing Session**
Exhibit Hall

7:00 – 8:00 PM **Early Career Social**
For attendees in training or recently out of training
Park View Room, Fourth Floor

8:00 – 11:00 PM **CGC Social: Celebrating YOU – The Stars of Genomics**
Supported by Bionano
All CGC 2023 attendees are invited to join the festivities on the rooftop of the Hyatt Regency overlooking St. Louis, the Mississippi River, and the Gateway Arch.

TUESDAY, AUGUST 15, 2023

8:00 – 9:00 AM **Session 7: Bioinformatics, Artificial Intelligence, Machine Learning**
Moderators: Cate Paschal, *Seattle Children's Hospital and*
Jeanine Ruggeri, *University of Colorado*

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*
Technologist Award

9:00 – 10:00 AM **Keynote Presentation: Incorporating Genomic Information in the Treatment of MDS**

Guillermo Garcia-Manero*, MD Anderson Cancer Center*
Introduction: Rashmi Kanagal-Shamanna, *MD Anderson Cancer Center*

10:00 – 10:45 AM **Coffee Break with Exhibitors**
Science Café Presentations:

- **nRichDX,** Nafiseh Jafari
- **GT Molecular,** Stephanie Barbari
- **Metasystems**

10:45 – 11:45 AM **Session 8: Value of Novel Technologies for the Identification of Clonal Aberrations different from Standard of Care Findings in Hematologic Malignancies**
Moderators: Min Fang, *Fred Hutchinson Cancer Center* and
Xiaoyu Qu, *Fred Hutchinson Cancer Center*

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
Samuel Brody, *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 AM – 12:15 PM **Spotlight Symposium: WHO/IARC Overview and CGC Collaboration**
Dilani Lokuhetty, *International Agency for Research on Cancer (IARC)*
Jennelle Hodge, *Indiana University School of Medicine*
Introduction: Rashmi Kanagal-Shamanna, *MD Anderson Cancer Center*
(No CME or CEUs available)

12:15 – 12:30 PM **Platinum Vendor Showcase: Imagia Canexia Health**
Machine Learning Method for Identifying Microsatellite Instability (MSI) DNA Samples without Matched Normal
Vincent Funari, *Imagia Canexia Health*
(No CME or CEUs available)

12:30 – 12:45 PM **Platinum Vendor Showcase: Qiagen**
(No CME or CEUs available)

12:45 – 2:00 PM **Round Table Discussions with Lunch**
Table topic selection was during conference registration.
Hyatt Regency Fourth Floor Foyer and Mills Meeting Rooms

2:00 – 3:00 PM **Session 9: Applications of Emerging Technologies in Clinical Genomics**
Moderators: Patricia Miron, *UMass Memorial Medical Center* and
Barbara Nelson, *Vanderbilt University Medical Center*

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 Genomics*

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*

3:00 – 3:30 PM

Invited Speaker Presentation: TP53 Alterations in Myelodysplastic Neoplasms and Acute Myeloid Leukemia
Joseph Khoury, University of Nebraska Medical Center
Introduction: Panieh Terraf, *Memorial Sloan Kettering Cancer Center*

3:30 – 4:15 PM

Coffee Break with Exhibitors
Science Café Presentations

- **PGDx / LabCorp**
- **Constantiam BioSciences,** Nicholas Schafer
- **Tempus**

4:15 – 4:30 PM

Platinum Vendor Showcase: SOPHiA Genetics
Liquid Biopsy – From Individual Markers to Tumor Signal
Florian Klemm, *SOPHiA Genetics*
(No CME or CEUs available)

4:30 – 5:00 PM

Speed Abstracts Session IV
Moderator: Sara Akhavanfard, *University Hospitals Cleveland Medical Center*
(No CME or CEUs available)

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*

5:00 – 5:30 PM **Spotlight Symposium: GOAL Consortium**
Jeremy Segal, *University of Chicago*
Introduction: Angela Lager, *University of Chicago*

5:30 – 9:00 PM **Departure for Pre-Registered Social Activities**
On-site registration may be available – please check with the meeting registration desk if you are interested in attending one of these events. Participants pick up tickets at the CGC 2023 Registration Desk.

- **St. Louis Riverboat Cruise:**
  Meet at 5:30 PM in the Hyatt Regency Lobby to walk to the river entrance on the far side of the arch. Boat loads at 5:45 PM and departs promptly at 6:00 PM. Cruise is one hour in length.
- **St. Louis Cardinals vs. Oakland Athletics:**
  Meet at 6:00 PM in Hyatt Regency Lobby to walk to the event together or join the group at Busch Stadium. Game starts at 6:45PM.

WEDNESDAY, AUGUST 16, 2023

8:30 – 9:30 AM **Session 10: Understanding Genomic Variants in Clinical Practice**
Moderators: Huan Mo, *NHGRI* and
Ngonidzashe Faya, *Cincinnati Children's Hospital Medical Center*

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 Presentation: Somatic Mutations and their Contribution to Bone Marrow Failure and Inflammation**
David Beck, *New York University School of Medicine*
Introduction: Celeste Eno, *Cedars-Sinai Medical Center*

10:30 – 11:15 AM **Coffee Break with Exhibitors**

11:15 AM – 12:15 PM **Session 11: Solid Tumors**
Moderators: Thuy Phung. *University of South Alabama* and
Casey Brewer, *Cincinnati Children's Hospital Medical Center*

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*
Trainee Award

Utilizing rapid molecular testing to reduce disparities 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**