

# Cancer Genomics Consortium, 12<sup>th</sup> Annual Meeting August 1 - 4, 2021

The CGC 2021 Virtual Annual Meeting will be presented in the eastern daylight times (EDT) shown below.

After each presentation, recordings will be available on the virtual meeting platform for those registrants in different time zones, and will be posted later on the CGC website for on demand access for CGC members.

#### Sunday, August 1, 2021

### Opening of the 12<sup>th</sup> Annual Cancer Genomics Consortium

11:30 AM Welcome

Patricia M. Miron, UMass Memorial Medical Center

11:35 AM – 12:35 PM Keynote Presentation

Deciphering oncohistone pathogeniesis in cancer

Nada Jabado, McGill University

Introduction: Patricia M. Miron, UMass Memorial Medical Center

Speaker Q & A

12:35 – 1:20 PM Diamond Vendor Showcase: Illumina

(No CME available)

DNA Methylation, CNV Analysis, and Case Studies

Andreas von Deimling, University Clinic Heidberg and DKFZ

Eric Vail, Cedars-Sinai

David Solomon, University of California San Francisco

Speaker Q & A

1:20 – 1:25 PM Break: Announcements

1:25 - 2:25 PM

## **CGC Session 1: Brain Tumors – Molecular Landscape and Emerging Targets**

#### **Moderators:**

Jacklyn Biegel, Children's Hospital Los Angeles Vera Paulson, University of Washington

Clinicopathologic analysis of gliomas harboring ROS1 gene rearrangements

Linda Cooley, UMKC

TERT and MGMT RNA sequencing gene expression correlation with mutational/methylation promoter status in brain tumors

Christiane Ida, Mayo Clinic

Optical Genome Mapping reveals novel structural variants in in pediatric high grade gliomas

Miriam Bornhorst, Children's National Hospital

Glioblastomas with MAPK pathway alterations show low grade histologic features and present novel therapeutic targets

David Meredith, Brigham and Women's Hospital

Session Q & A

2:25 – 2:50 PM Platinum Vendor Showcase: Qiagen

(No CME available)

Homologous recombination repair (HRR) and deficiency (HRD):

The role of DNA damage repair (DRR) Vikas Gupta and Raed Samara, *Qiagen* 

Speaker Q & A

2:50 PM Gold Vendor Showcase: BioDot

(no CME available)

**BioDot CellWriter** Matt Sergent, *BioDot* 

2:55 – 3:00 PM Break: Announcements

### **Sunday Workshops**

#### Additional Information available online.

There is an additional registration fee to attend the workshops.

3:00 – 4:30 PM CGC Workshop

Visualization and interpretation tools/resources for structural

genomic variants

**Moderator:** 

Deborah Ritter, Baylor College of Medicine

#### Speakers:

Debyani Chakravarty, *Memorial Sloan Kettering Cancer Center* Jianjiong Gao, *Memorial Sloan Kettering Cancer Center* Jason Saliba, *Washington University in St. Louis* Ahmet Zehir, *Memorial Sloan Kettering Cancer Center* 

4:30 – 4:40 PM Break: Announcements

4:40 – 6:10 PM CGC Workshop

Case-based discussion of updates in cytogenic and molecular nomenclature, interpretation and reporting standards, and interpretation resources for cancer variants

#### **Moderator:**

Gordana Raca, Children's Hospital Los Angeles

#### Speakers:

Jean Mcgowan-Jordean, CHEO Genetics Laboratory/ University of Ottawa Beth Pitel, Mayo Clinic

Teresa Smolarek, Cincinnati Children's Hospital Medical Center Gordana Raca, Children's Hospital Los Angeles Svetlana Yatsenko, University of Pittsburgh Medical Center

Ravindra Kohle, Augusta University

6:15 – 7:15 PM Early Career/New Director Event

#### Monday, August 2, 2021

10:00 - 10:10 AM Presidential Address

Yassmine Akkari, Legacy Health

10:10 – 11:10 AM CGC Session 2: New Strides in Technology

#### **Moderators:**

Fabiola Quintero-Rivero, *University of California Irvine* Cinthya Zepeda, *ARUP* 

Clinical laboratory validation of TCR-seq for detection of clonality Jane Houldsworth, *Icahn, Mt. Sinai* 

Emerging clinically-relevant applications of ultra-sensitive mutation detection

Lei Wei, Roswell Park

Adoption off Optical Genome Mapping in Clinical Cancer Cytogenetics Laboratory: A Stepwise Approach Victoria Stinnett, *The Johns Hopkins University* 

Rapid Clinical BRAF V600 mutation testing using Idylla Platform Meenakshi Mehrotra, *Icahn School of Medicine at Mount Sinai* 

Session Q & A

11:10 AM Gold Vendor Showcase: FujiFilm Irvine Scientific

(no CME available)

Cell culture media specialists: Providing solutions for cytogentics in

clinical and research applications Shaun Waters, FujiFilm Irvine Scientific

11:15 – 11:35 AM Break Announcements

11:35 AM - 12:35 PM Keynote Presentation

Global views of cell lineage in metastasis and development

Jay Shendure, University of Washington

Introduction: Cinthya Zepeda, ARUP

Speaker Q & A

12:35 – 1:20 PM Diamond Vendor Showcase: Bionano Genomics

(No CME available)

1:20 – 2:35 PM Lunch Break / Exhibit Hall Open

2:35 – 3:20 PM Invited Speaker

The next generation of cytogenetics and molecular genetics in

leukemia diagnostics

Torsten Haferlach, Munich Leukemia Laboratory

Introduction: Rashmi Kanagal-Shamann, MD Anderson

Speaker Q & A

3:20 PM Gold Vendor Showcase: ADS Biotec

(no CME available)

**Enabling Seamless Automation in Cytogenetics** 

John McCloskey, ADS Biotec

3:25 – 4:10 PM CGC Session 3: New Molecular Insights into Myeloid Hematologic

**Malignancies** 

**Moderators:** 

Gordana Raca, Children's Hospital Los Angeles

Lisa Lansdon, Children's Mercy Hospital

ETV6-PDGFRA fusions detected by FISH in acute myeloid leukemia

with translocation t(4;12)(q12;p13) are false positive

Sarah Mueller, Massachusetts General Hospital

First Place Trainee Abstract Award

SNP microarray analysis of over 7,500 myeloid patients: Implications,

importance and suggestions for standard of care

Stuart Schwartz, Laboratory Corporation of America

## Optical Genome Mapping for Chromosomal Structural Variants Analysis in Hematological Malignancies

Rashmi Kanagal-Shamanna, MD Anderson Cancer Center

Session Q & A

4:10 - 4:35 PM

Platinum Vendor Showcase: SOPHiA Genetics

(No CME available)

The SOPHiA DDMTM Platform: Deep dive on how we support solid tumor detection

Emily Paul, Mikhail Pertziger, and Marian Novak, SOPHiA Genetics

4:35 - 5:40 PM

**Exhibit Hall Open** 

5:40 - 6:25 PM

**CGC Session 4: Integrating Genomics into Constitutional Analysis** 

#### **Moderators:**

Patricia Miron, *UMass Memorial Medical Center* Rebecca Smith, *Genetics Associates, Inc.* 

Cancer Cytogenomic Array analysis reveals origin of ovarian teratoma and reproductive lessons learned from it

Lina Shao, University of Michigan

Next-generation cytogenetics: Proposal for a cost-effective approach for comprehensive testing of prenatal cases

Nikhil Sahajpal, Augusta University

Deletions resulting in Brachydactyly Mental Retardation Syndrome (BDMR): Multiple Contributing Regions in Distal 2q37?

Aiko Otsubo, Indiana University

Session Q & A

6:25 PM

**Gold Vendor Showcase: Global Alliance for Genomics & Health (GA4GH)** 

(no CME available)

Presenting the Global Alliance for Genomics & Health (GA4GH)

**Gold Vendor Showcase: Michigan Medical Laboratories** *(no CME available)* 

6:30 - 7:00 PM

Panel-Based Discussion: Navigating the evolution of cytogentics Yassmine Akkari, *Legacy Health*P. Alan Lennon, *PathGroup* 

Karen Rabin, Texas Children's Cancer Center, Baylor College of Medicine

**Q & A with Speakers** 

#### Tuesday, August 3, 2021

10:00 - 11:00 AM

**CGC Session 5: Database Curation and Mining** 

**Moderators:** 

Kilannin Krysiak, *Washington University School of Medicine* Beth Pitel, *Mayo Clinic* 

**Standard Procedure for the Curation and Maintenance of Cancer-Specific Gene Lists** 

Beth A. Pitel, Mayo Clinic

First Place Technologist Abstract Award

Enhancement of Pediatric Cancer Curation and Representation through expert-guided data mining and ontology refinement Jason Saliba, *Washington University School of Medicine* 

Multi-consotria initiative to standardize the representation and curation of oncogenic fusions

Alex Wagner, Nationwide Children's Hospital

Progenetix – An open reference resource for copy number variation data in cancer

Qingyao Huang, Universität Zürich

Session Q & A

11:00 AM

**Gold Vendor Showcase: The Jackson Laboratory** (no CME available)

JAX Clinical Knowledgebase (CKB) Cara Statz, The Jackson Laboratory

11:05 - 11:50 AM

**Invited Speaker** 

Al Applications to drive precision oncology clinical trials

Subha Madhavan, Georgetown University

Introduction: Obi Griffith, Washington University

Speaker Q & A

11:50 AM - 12:35 PM

Diamond Vendor Showcase: Agilent Technologies

(No CME available)

One workflow – endless options: NGS workflow solutions

Brigette Brown-Kipphut, Agilent Technologies

K.C. Varva, Agilent Technologies

12:40 - 1:40 PM

**Roundtable Breakout Sessions** 

Topic list available <u>online</u>. Please make your selection during registration. Only 16 spaces are available in each discussion topic.

1:40 – 2:15 PM Lunch Break / Exhibit Hall Open

2:15 – 3:15 PM Keynote Presentation

Towards Population genomic screening for cancer susceptability Leslie G. Biesecker, *National Human Genome Research Institute* 

Introduction: P. Alan Lennon, PathGroup

Speaker Q & A

3:15 PM Gold Vendor Showcase: Geneoscopy (no CME available)

Presenting Geneoscopy Erica Barnell, Geneoscopy

3:20 – 4:05 PM CGC Session 6: Genetic Spectrum of Vascular Anomalies and Overgrowth Syndromes

Moderators:

Min Fang, Fred Hutchinson Cancer Research Center Alaa Koleilat, Mayo Clinic

**Genomic structural variations in lymphatic anomalies** Thuy Phung, *University of South Alabama* 

Diagnostic utility and lessons learned from deep sequencing vascular malformations

Candace Myers, Seattle Children's Hospital

Co-existence of two activating variants in somatic overgrowth and vascular anomalies: 7 years' findings at a single center Yang Cao, Washington University School of Medicine

Session Q & A

4:05 – 4:30 PM Platinum Vendor Showcase: Natera (No CME available)

Informed by the tumor: The power of a personalized ctDNA Assay for Treatment Monitoring in HER2

Angel Rodriguez and John Simmons, Natera

4:30 – 5:30 PM Exhibit Hall Open

5:30 – 6:15 PM CGC Session 7: Hematologic Malignancies

**Moderators:** 

Lina Shao, *University of Michigan*Marilena Melas, *Nationwide Children's Hospital* 

Characterization of atypical iAMP21 observed in B-Lymphoblastic Leukemia (B-ALL): a retrospective study from Mayo Clinic

Alaa Koleilat, Mayo Clinic

**Second Place Abstract Trainee Award** 

Complementarity of RNA sequencing and optical genome mapping in detection of rare fusions in pediatric B-ALL

Gordana Raca, Children's Hospital Los Angeles

Archer system copy number alteration caller: user experience

Celeste Eno, Cedars-Sinai Medical Center

Session Q & A

6:15 - 6:35 PM Platinum Vendor Showcase: Roche (No CME available)

HyperPETE: A Novel Primer Extension Target Enrichment Solution for

**Rapid and Reliable Variant Detection Results** 

Brian Godwin, Roche Sequencing Solutions

6:40 - 7:10 PM From oncology to pathology and genomics and back again: a 360-

degree perspective on a unique B-cell leukemia

Adrian Dubuc, Jacqueline Garcia, and Gabriel Griffin, Brigham and

Women's Hospital

7:15 - 8:30 PM **After Hours Social** 

#### Wednesday, August 4, 2021

10:00 - 11:00 AM **CGC Session 8: Hereditary Cancer/Cancer Predisposition** 

**Moderators:** 

Xinjie Xu, Mayo Clinic

Angela Lager, University of Chicago

Large scale analysis in Von Hippel-Lindau disease

Kristen Farncombe, University Health Network

Co-occurrence of rosette-forming glioneuronal tumors with Noonan

svndrome

Marilena Melas, Nationwide Children's Hospital

10-year retrospective analysis of BRCA germline mutation in multiethnic gynecologic patients form a U.S. cancer center

Christina Wei, City of Hope National Medical Center

Identification of TP53 germline variants in pediatric patients

undergoing tumor testing

Minjie Luo, Children's Hospital of Philadelphia

Session Q & A

11:00 AM **Gold Vendor Showcase: Oxford Nanopore** 

(no CME available)

11:05 - 11:35 AM **Break: Announcements** 

11:35 AM - 12:35 PM **Keynote Presentation**  Proteogenomics as a driver for discovery of biomarkers and therapeutic targets in cancer

Kojo S. J. Elenitoba-Johnson, University of Pennsylvania

Introduction: Yassmine Akkari, Legacy Health

Speaker Q & A

12:35 - 1:20 PM **Diamond Vendor Showcase: Thermo Fisher Scientific** 

(No CME available)

Biomarker discovery through tumor profiling

Mary Napier, Thermo Fisher Scientific

Improved risk stratification for B-ALL patients using chromosome genomic array testing and targeted RNA sequencing

Min Fang, Fred Hutchinson Cancer Center Research Center and University of Washington, Seattle

Speaker Q & A

1:20 - 2:35 PM **Lunch Break / Exhibit Hall Open** 

2:35 PM **Gold Vendor Showcase: BioDiscovery** 

(no CME available)

NxClinical's Approach to HRD Analysis

2:40 - 3:30 PM **CGC Session 9: CGC Updates** 

**Moderators:** 

Yassmine Akkari, Legacy Health

Kilannin Krysiak, Washington University School of Medicine

Launching a CGC initiative to support trainees and early career members: Survey results and analysis

Kilannin Krysiak, Washington University School of Medicine

Perspectives in genetic counseling

Lindsey Byrne, The Ohio State University

Sheila Solomon, Guardant Health

The Compendium of Cancer Genome Aberrations (CCGA): Becoming

a global hub for clinical interpretation

Jennelle Hodge, Indiana University

**CGC PACC Report: New Initiatives and future directions** 

Rashmi Kanagal-Shamanna, MD Anderson Cancer Center

3:30 - 3:55 PM Platinum Vendor Showcase: Oxford Gene Technology

(No CME available)

Transitioning CLL FISH analysis into the molecular biomarker era

Tracy Tucker, BC Cancer

**CGC Session 10: Bioinformatics** 3:55 - 4:25 PM

#### Moderator:

Obi Griffith, Washington University School of Medicine

A cost-effective bioinformatics triage strategy for testing PMS2 shortread next generation sequencing

Wei Shen, Mayo Clinic

Computer-aided cytogenomic classification of renal cell carcinoma

Soheil Shams, BioDiscovery

4:25 PM Gold Vendor Showcase: Twist Bioscience (no CME available)

Writing the future if cancer research

Melina Mathur, Twist Bioscience

4:30 – 5:30 PM Exhibit Hall Open

5:30 – 6:30 PM CGC Session 11: Unraveling Solid Tumors with Genomic Technologies

#### Moderators:

Teresa Smolarek, Cincinnati Children's Hospital Medical Center Niroshi Senaratne, University of California Los Angeles

Clinical utility and feasibility of adopting optical genome mapping for chromosomal characterization of solid tumors

Nikhil Sahajpal, *Augusta University* Second Place Technologist Abstract Award

Tumor cellularity estimation in a targeted NGS panel of tumor-only specimens expands analytical utility

Stephanie Siegmund, Brigham and Women's Hospital

A molecular and clinicopathologic analysis of primary intracranial sarcomas

Zahra Aldawood, Harvard School of Dental Medicine

Atypical FISH patterns clarified by RNAseq in solid tumor specimens Beth A. Pitel, *Mayo Clinic* 

Session Q & A

Yassmine Akkari, CGC President, *Legacy Health*Xinjie Xu, CGC Treasurer, *Mayo Clinic*Patricia M. Miron, CGC President-Elect, *UMass Memorial Medical Center* 

**Business Meeting Q & A** 

**Closing Remarks**