Cancer Genomics Consortium, 12th 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 12th Annual Cancer Genomics Consortium**

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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</thead>
<tbody>
<tr>
<td>11:30 AM</td>
<td>Welcome</td>
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<tr>
<td></td>
<td>Patricia M. Miron, UMass Memorial Medical Center</td>
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<tr>
<td>11:35 AM – 12:35 PM</td>
<td><strong>Keynote Presentation</strong></td>
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<td></td>
<td>Deciphering oncohistone pathogenesis in cancer</td>
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<td>Nada Jabado, McGill University</td>
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<td><strong>Introduction:</strong> Patricia M. Miron, UMass Memorial Medical Center</td>
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<tr>
<td>12:35 – 1:20 PM</td>
<td><strong>Diamond Vendor Showcase: Illumina</strong></td>
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<td></td>
<td>(No CME available)</td>
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<td></td>
<td>DNA Methylation, CNV Analysis, and Case Studies</td>
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<td></td>
<td>Andreas von Deimling, University Clinic Heidelberg and DKFZ</td>
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<td>Eric Vail, Cedars-Sinai</td>
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<td>David Solomon, University of California San Francisco</td>
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<td></td>
<td><strong>Speaker Q &amp; A</strong></td>
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<td>1:20 – 1:25 PM</td>
<td><strong>Break: Announcements</strong></td>
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</table>
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 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 Mccowan-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**

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**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 of 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 cytogenetics 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 cytogenetics
Yassmine Akkari, Legacy Health
P. Alan Lennon, PathGroup
Karen Rabin, Texas Children’s Cancer Center, Baylor College of Medicine

Q & A with Speakers
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

AI 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 online. 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 syndrome
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

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

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

3:55 – 4:25 PM

**CGC Session 10: Bioinformatics**
Moderator:
Obi Griffith, *Washington University School of Medicine*

A cost-effective bioinformatics triage strategy for testing PMS2 short-read 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

6:30 – 7:00 PM

**CGC 2021 Business Meeting**

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