PACE Accreditation Details

CGC 2024 Program Descriptions and Objectives

Sunday, August 4, 2024

Pre-Meeting Workshops

(Additional Registration Required)

Leadership Workshop: Influence without authority
Description of Session: "*If you want to go fast, go alone, if you want to go far, go with others" -African Proverb.*
But how would you work with others to bring about positive changes to the organization? We have been trained to make conclusions based on data with a $P < 0.05$ often following a sophisticated algorithm. For example, would it work the same way if we try to convince administration to budget for a half-a-million-dollar instrument and two extra FTE? How could you approach staff who do not seem to be as excited about the new assay as you are, or perhaps how could you convince IT to you let you have administrative rights to your computer?

In this session, we will examine how change agents get buy-in from key stakeholders--leadership, employees, and others. Using a metaphor of head, heart, and hands, we will look at how real people with little to no authority were able to get buy-in for major change initiatives.

Level of Instruction: Basic

Program Objectives:
At the end of the session, the participant will be able to:

1. Think about the changes you have brought in or are trying to bring to your organization. After the session, describe what the level of buy-in you have, why you succeeded, or what is missing.
2. Describe key actions you may take to get stakeholders to "buy-in" to your idea(s) or initiative.
3. Describe an example of a positive deviance approach to change. How does it differ from the traditional approach?

CONTACT HOURS: 1.5

Bioinformatics Workshop: Classifying variant oncogenicity from functional effect data
Description of Session: Clinical interpretation of genomic variants in patients requires the appropriate use of relevant genomic evidence resources supporting the role of a variant in driving tumor oncogenicity. One important evidence type is experimental data describing the functional consequences of gene variants. This bioinformatics workshop will provide an overview of the ClinGen/CGC/VICC criteria used to assess functional consequence data, with follow-along demonstrations of functional evidence in multiple platforms: the CIViC knowledge base, the MAVEdb evidence repository, and the Genomics 2 Proteins portal. This interactive workshop is intended for a general audience with varying expertise in bioinformatics and an interest in using these platforms to assist in evaluating functional consequence data

for variant interpretation. Participants are encouraged (but not required) to bring their own laptop computers to actively engage with these genomic evidence resources as demonstrated by the workshop presenters. Demonstrations will be led by platform experts, and questions are encouraged throughout the workshop.

Level of Instruction: Basic

Program Objectives:
At the end of the session, the participant will be able to:

1. Apply the ClinGen/CGC/VICC oncogenicity guidelines to classify variant oncogenicity.
2. Curate oncogenicity classifications in the CIViC resource.
3. Extract variant functional effect data from the MAVEdb resource.
4. Explore protein variant effects using the Genomics 2 Proteins portal.

CONTACT HOURS: 1.5

Keynote Presentation: Enabling clinical translation of high-throughput functional assay data

Alan Rubin*, Walter and Eliza Hall Institute*

Description of Session: This session will describe the generation and use of high-throughput functional assay data and its use for understanding protein missense variant function, including classifying variant pathogenicity.

Level of Instruction: Intermediate

Program Objectives:
At the end of the session, the participant will be able to:

1. Recognize the broad classes of high-throughput functional genomics experiments for assaying missense variants at scale.
2. Understand the importance of data standards and data sharing for applying high-throughput assay data in the clinic.
3. Comprehend the level of diversity and heterogeneity of experimental techniques being applied in this space.
4. Realize that careful application of functional assay data can resolve a high proportion of variants of uncertain significance in real-world clinical settings.
5. Appreciate how high-throughput functional assays can also be used to understand important biological mechanisms, such as resistance to chemotherapy.

CONTACT HOURS: 1.0

Speed Abstracts Session I: Precisive genetic diagnosis of hematological and other malignancies

Description of Session: This session will present information from laboratories on how to improve the precisive diagnosis of gene mutations/chromosomal aberrations in hematological and other malignancies, such as enrichment of specific tumor cells, quantitative detection of *MYD88 L265p* in LPL, validation of

OGM platforms, FISH automation, and detection of macrogenomic events (MGE) utilizing various platforms/methods.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:

1. Enrich their specific knowledge of improving laboratory performance by adding pre-analytical measurements.
2. Learn first-hand experience implementing and validating new platforms/methods such as OGM.
3. Comprehend FISH automation skills, even on amniocentesis specimens.

CONTACT HOURS: 0.5

Session 1: Diagnostic and prognostic impact of molecular profiling across hematological neoplasms

Description of Session: This session will present molecular studies pertinent to haematological malignancies that will assist in the diagnostic setting

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:
1. Understand the different clinical outcomes for those patients with myeloid neoplasms and *D816KIT* mutations as compared to those with non-*D816 KIT* variants.
2. Know what is required to implement gene expression profiling (GEP)-based classification for paediatric B-lymphoblastic leukemias.
3. Appreciate the differences in subclone diversity in clonal cytopenias of undetermined significance as compared to that in myelodysplastic syndrome.
4. Be aware of the FISH best practice guidelines for testing and reporting in multiple myeloma.

CONTACT HOURS: 1.0

Speed Abstracts Session II: Standardization, bioinformatics workflows, and data integration for personalized healthcare

Description of Session: This session will cover innovative advancements in clinical genomics, highlighting the standardization of gene fusion detection, the integration of therapeutic context into drug-gene interactions, the identification and analysis of focal amplifications in cancer genomes, and the development of robust bioinformatics workflows for personalized neoantigen vaccine clinical trials.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:

1. Understand the development and application of the FUSOR package for standardizing and validating gene fusion calls in clinical genomics, and how it addresses gaps in current fusion detection standards.
2. Gain insights into the enhancement of drug-gene interaction databases with therapeutic context using large language models and FDA label data, and how this improved context can inform clinical decision-making.
3. Learn about the identification and classification of focal amplifications in cancer genomes using the AmpliconSuite tools, and the significance of these amplifications in cancer progression and treatment.
4. Explore the integration of robust bioinformatics workflows for designing personalized neoantigen vaccines in clinical trials, focusing on the strategies and tools used to ensure accurate neoantigen prediction and validation.

CONTACT HOURS: 0.5

Monday, August 5, 2024

Keynote Presentation: The grand challenge of cancer disparities

Melissa B. Davis, *Morehouse School of Medicine*

Description of Session: This session will present information on disparities in cancer care in racial and ethnic minorities and other underserved populations.

Level of Instruction: Basic

Program Objectives:
At the end of the session, the participant will be able to:

1. Understand the challenges and barriers that affect access to cancer care in underserved populations.
2. Gain insights into how social determinants of health can affect cancer outcome.
3. Appreciate how health disparities can account for significant healthcare costs to government and communities.
4. Comprehend how community engagement can be a key strategy for reducing health inequities, including cancer disparities.

CONTACT HOURS: 1.0

Session 2: Genomic equity and inclusivity: Bridging the gap in diagnostics and personalized care across diverse populations

Description of Session: During this exciting phase of genomic advancements, it is critical to ensure fair and inclusive application of genomic research and technologies across all populations, regardless of socioeconomic, ethnic, or geographic background. This session and included panel discussion will explore the importance of

understanding genomic variations across diverse populations and their implications in design and development of clinical assays, with focus on optimizing testing strategies for underserved and resource-limited settings.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participants will be able to:

1. Understand the importance of genomic variations across different populations and impact on the effectiveness of clinical assays.
2. Identify strategies for optimizing genomic testing approaches to effectively serve underserved and limited resource populations.
3. Develop methods to ensure equitable access to genomic testing.
4. Evaluate the importance of inclusivity in genomics research and biomarker driven clinical trials.

CONTACT HOURS: 1.0

Invited Speaker Presentation: Current and future integrations of genomics and AI

Chad Vanderbilt, *Memorial Sloan Kettering Cancer Center*

Description of Session: This session will present information covering the ways deep learning methods are being used in genomic medicine.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:

1. Identify how deep learning techniques are utilized to detect microsatellite instability (MSI) in cancers.
2. Describe the applications of natural language processing in genomic research and clinical practice.
3. Recognize the advancements in AI-driven genomic medicine.
4. Understand the limitations and considerations in integrating AI in genomic studies.
5. Discuss the potential and challenges of AI in future genomic medicine applications.

CONTACT HOURS: 0.5

Speed Abstracts Session III: Integrated sample processing and genomic analysis approaches that improve diagnostic yield for hematologic malignancies

Description of Session: This session will present cases that exemplify the strengths of integrated sample processing and genomic analysis in the diagnostic workup of hematologic malignancies.

Level of Instruction: Advanced

Program Objectives:

At the end of the session, the participant will be able to:

1. Contrast the strengths and limitations of routine genetic tests and whole genomic assays such as microarray, RNA-Seq, and optical genome mapping.
2. Understand the significance of FIP1L1::KIT gene fusion for the diagnosis of peripheral T-cell lymphoproliferative neoplasm.
3. Appreciate the utility of cell sorting via flow cytometry in the workup for hematologic malignancies.
4. Enumerate key genes encompassed by the intrachromosomal amplification of chromosome 21 and the significance of iAMP21 in acute myeloid leukemia.

CONTACT HOURS: 0.5

Session 3: Advances in molecular profiling for tumor risk assessment and management

Description of Session: This session with included panel discussion will highlight the application of state-of-the-art molecular diagnostic technologies to improve tumor risk assessment and management of prostate cancer, benign and malignant intracranial tumors, and clonal hematopoiesis.

Level of Instruction: Advanced

Program Objectives:
At the end of this session the participant will be able to:

1. Evaluate the potential superiority of methylation profiling of meningiomas to predict the recurrence risk compared to conventional WHO grading in routine practice.
2. Understand how a neural network machine learning algorithm that integrates microRNA biomarkers in prostate cancer may predict prognosis and guide effective monitoring strategies.
3. Consider the applicability of whole exome sequence analysis performed on plasma cell-free DNA to identify somatic mutations in common and rare intracranial tumors.
4. Understand the confounding effect of cell-type proportions on variant allele fraction (VAF) measurements in clonal hematopoiesis (CH) and assess a cost-effect effective targeted enzymatic DNA methylation sequencing assay that may overcome this limitation.

CONTACT HOURS: **1.5**

Invited Speaker Presentation: Interpretable and context-free deconvolution of multi-scale transcriptomic lung cancer data

Robert Sebra, *Icahn School of Medicine at Mount Sinai*

Description of Session: This session will present information from adaptation of emerging genomic technology perspectives where whole-transcriptome spatial platforms are used for lung cancer specimens.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:

1. Explain the analytic strategies for spatial transcriptomic data.
2. Appreciate the algorithms to deconvolve cell type fractions from the transcriptomic data.
3. Explain the training and validation processes for the machine-learning based deconvolve algorithms.
4. Appreciate the new findings in the biology of lung cancers by spatial transcriptomic data.

CONTACT HOURS: 0.5

Session 4: Global initiatives in enhancing interpretation of the somatic genome

Description of Session: This session will present information from initiatives ongoing in different parts of the world to enhance the curation and interpretation of somatic oncogenic variants.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:
1. Appreciate the role of open-access knowledge in precision oncology and somatic variant interpretation.
2. Comprehend the strengths and challenges associated with systematic application of the ClinGen/CGC/VICC Oncogenicity guidelines in a high-throughput pediatric cancer genomics clinical laboratory setting.
3. Be informed of genomic testing and variant curation practices in diagnostic laboratories performing somatic testing in Australia and New Zealand.
4. Understand the addition of non-gene features such as tumor biomarkers to the CIViC data model.

CONTACT HOURS: 1.0

Tuesday, August 6, 2024

Invited Speaker Presentation: To a carpenter, every problem is a nail: The FDA brings a hammer to diagnostic medicine

Dara Aisner, *University of Colorado Anschutz Medical Campus*

Description of Session: This session will present the background and current status of the FDA proposed regulation of laboratory developed tests (LDTs).

Level of Instruction: Intermediate

Program Objectives:
At the end of the session, the participant will be able to:
1. Understand the background of the FDA regulation of diagnostic medicine.

2. Appreciate the history of the FDA trying to regulate LDTs.
3. Comprehend the current status of the FDA proposed regulation of LDTs.
4. Appreciate the efforts of scientific organizations to provide guidance for an alternative pathway to regulate LDTs.

CONTACT HOURS: 1.0

Session 5: Development of resources for the reimbursement and clinical reporting standardization of cancer genomic testing

Session Description: This session with included panel discussion will present efforts to develop resources to assess the current reimbursement landscape for molecular assays and resources to standardize the clinical reporting of cancer genomic testing.

Level of Instruction: Intermediate

Program Objectives:
At the end of this session, the participant will be able to:
1. Understand the current reimbursement landscape for molecular assays.
2. Appreciate the different clinical reporting practices of next-generation sequencing test results.
3. Comprehend the efforts aimed at standardizing the clinical reporting of next-generation sequencing test results.
4. Understand the current initiatives to align the definitions and descriptions of gene fusions in clinical lab reports.

CONTACT HOURS: 1.0

Invited Speaker Presentation: Cell-free DNA and AI technology for liquid biopsy detection of cancer early and prediction of cancer treatment response

Aadel Chaudhuri, *Mayo Clinic*

Description of Session: This session will present an overview of liquid biopsy strategies for early cancer detection and prediction of response to treatment using Cell-free DNA and AI technology.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:
1. Be informed of liquid biopsies as an emerging class of techniques for noninvasive tumor profiling.
2. Appreciate the use of liquid biopsy technologies to find cancer earlier.
3. Understand the application of cell-free DNA analysis to predict responses to cancer treatment and to enable more precise response-adapted treatment strategies.
4. Realize the potential of AI technology powered liquid biopsy methods in guiding precision oncology.

CONTACT HOURS: 0.5

Speed Abstracts Session IV: Innovative tools and challenges in genetic variant analysis

Description of Session: This session will present information on advanced computational tools for genetic variant analysis, including the prediction and prioritization of alternative splicing neoantigens, variant interpretation workflow customization, and the identification of challenges in variant normalization.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:

1. Utilize pVACsplice to predict and prioritize alternative splicing neoantigens.
2. Customize their variant interpretation workflow using OpenCRAVAT to enhance genetic analysis.
3. Identify and address common challenges in variant normalization to improve data accuracy.
4. Integrate advanced computational tools into genomic research and clinical practice for more effective variant analysis.

CONTACT HOURS: 0.5

Keynote Presentation: Precision interception in multiple myeloma and its precursor conditions

Irene Ghobrial, *Dana Farber Cancer Institute*

Description of Session: This session will present information about the plasma cell neoplasm, multiple myeloma, and the early detection of its precursor conditions, monoclonal gammopathy of undetermined significance and smoldering multiple myeloma.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:

1. Comprehend that multiple myeloma evolves from a precursor state, monoclonal gammopathy of undetermined significance (MGUS) and/or smoldering multiple myeloma (SMM), but not all individuals with MGUS or SMM evolve to develop multiple myeloma.
2. Understand the biological and clinical differences between the precursor conditions and multiple myeloma.
3. Appreciate that there may be benefits to early screening for precursor conditions prior to the development of multiple myeloma.
4. Realize that effective treatment of multiple myeloma may benefit from recognizing which patients will progress from the precursor condition to multiple myeloma.

CONTACT HOURS: 1.0

Wednesday, August 7, 2024

Session 6: Genomic analyses of solid tumors: Arrays, sequencing and developing a gene list

Description of Session: This session will present different approaches used to characterize genomic changes in solid tumors as well as an approach used to develop a gene list used to evaluate CNS tumors.

Level of Instruction: Advanced

Program Objectives:
At the end of the session, the participant will be able to:

1. Identify clinically important genomic changes in meningioma.
2. Identify common genomic changes in Wilms tumor.
3. Describe the clinical utility of a commercially available kit used to detect genomic changes in solid tumors.
4. Identify an approach to develop a gene list to be used in the evaluation of genomic changes relevant to a specific tumor subtype.

CONTACT HOURS: 1.0

Session 7: Updates in oncogenicity guidelines and classification rules for prioritizing variants in somatic disease

Description of Session: This session will present updates from various taskforces of the ClinGen Somatic Variant Curation Working Groups across a range of adult and pediatric cancers.

Level of Instruction: Intermediate

Program Objectives:
At the end of the session, the participant will be able to:

1. Appreciate proposed modifications to ClinGen/AMP/ASCO/CAP guidelines for the interpretation of histone H3 specific variants in glioma.
2. Differentiate disease-defining and disease-supportive diagnostic variants for curation prioritizing in pediatric malignancies.
3. Understand somatic curation and classification rules piloted against a set of new variants targeting internal tandem duplications and fusions.
4. Realize challenges in fusion-specific classifications using the NTRK oncogenicity guidelines as a model to improve workflows and clinical validity guidance.

CONTACT HOURS: 1.0

Session 8: Advancements in cancer-adjacent genetic diagnostics: Guidelines, assays, and case studies

Description of Session: This session will explore diagnostic guidelines, assays, and case studies relevant to cancer genomics in non-cancer conditions.

Level of Instruction: Intermediate

Program Objectives:

At the end of the session, the participant will be able to:

1. Identify recurrent structural and sequence variants associated with bone marrow failure syndromes, developmental delay, and vascular anomalies.
2. Understand the use of UMI-based NGS panels and cytogenomic SNP-A microarrays for precise diagnosis of non-cancerous genetic conditions.
3. Recognize the importance of developing and applying condition-specific guidelines for accurate variant interpretation in non-cancer somatic disease.
4. Discuss the translation of diagnostic methods and frameworks between cancer and cancer-adjacent diseases.

CONTACT HOURS: 1.0