

2025 Spring Symposium: Emerging Frontiers in Cancer Genetics

Day One: April 8, 2025

9:00am – 10:00am: Deciphering Noncoding Regulation

- 9:00am – 9:10am *Decoding CDH1: the hidden role of regulatory noncoding sequences in hereditary diffuse gastric cancer*
Celina São José, Instituto de Investigação e Inovação em Saúde (i3S)
- 9:15am – 9:25am *Modulation of Oncogenic Regulatory Networks by Exosomal miRNAs in Patients with Non-Small Cell Lung Cancers*
Abeer Hassanin, Texas A&M Health
- 9:30am – 9:40am *Reannotation of cancer mutations based on expressed RNA transcripts reveals novel functional non-coding mutations in melanoma*
Xander Janssens, KU Leuven
- 9:45am – 9:55am *Genome-Wide Mapping of Regulatory Elements and Clusters of Non-Coding Somatic Mutations in Liver Cancer Genomes*
Todd Johnson, RIKEN Center for Integrative Medical Sciences (IMS)

10:00am – 11:00am: Genomics-Aided Precision Oncology

- 10:00am – 10:10am *Integrated multi-omics drive precision cancer care for pediatric CNS tumors: An institutional study of 582 tumors*
Derek Wong, Children's Hospital of Philadelphia
- 10:15am – 10:25am *Pathways to precision cancer care: integrating genetic services into a nationwide cancer program at scale*
Scott Topper, Color Health
- 10:30am – 10:40am *Precision Oncology Workflow Modifications in a Community Cancer Center Led to Greater Adoption of Genetic Counseling Referral and Testing in Patients Diagnosed with Ovarian Cancer*
Sourat Darabi, Precision Medicine
- 10:45am – 10:55am *Defining actionable co-mutations and mutually exclusive genes in NF1-altered sporadic breast cancer*
Mehrshid Faraji Zonooz, Van Andel Institute

11:00am – 12:00pm: Refining Variant Classification in Cancer

- 11:00am – 11:10am *Integrating Germline and Somatic Data to Resolve Variants of Uncertain Significance (VUS) in Colorectal Cancer (CRC) Patients who Self-Identified as Hispanic/Latino*
Jonathan (Yonatan) Amzaleg, City of Hope
- 11:15am - 11:25am *Development and application of MUTYH-specific ACMG/AMP variant classification criteria*
Emily Nadeau, University of Vermont
- 11:30am – 11:40am *Saturation genome editing based functional evaluation and clinical classification of BRCA2 single nucleotide variants*
Fergus Couch, Mayo Clinic
- 11:45am – 11:55am *Single cell RNA sequencing-based modeling of the mismatch repair pathway to support clinical variant classification for Lynch Syndrome*
Rakesh Pinninti, Basavatarakam Indo-American Cancer Hospital and Research Institute

Day Two: April 9, 2025

9:00am – 10:00am: Cancer Versus the Immune System

- 9:00am – 9:10am *AlphaMissense pathogenicity scores predict response to immunotherapy and enhances the predictive capability of tumor mutational burden*
David Adeleke, North Dakota State University
- 9:15am – 9:25am *Genetic regulation of TERT splicing contributes to reduced or elevated cancer risk by altering cellular replicative potential*
Oscar Florez-Vargas, National Cancer Institute
- 9:30am – 9:45am *Immune surveillance and cancer risk*
Miriam Saffern, Icahn School of Medicine at Mount Sinai
- 9:45am – 10:00am *Identification of Gene Fusions Using RNA-Seq in a Large Cohort of CLL Tumors*
Numrah Fadra, Mayo Clinic

10:00am – 11:00am: Innovations in Cancer Genomics and Prediction

- 10:00am – 10:10am
- 10:15am – 10:25am *Targeted cell-free DNA methylation haplotype signatures in prostate cancer progression*
Jodie Wong, Moffitt Cancer Center
- 10:30am – 10:40am *Multiomic 6-base data from cell-free DNA enhances the performance of liquid biopsy classifiers*
Tom Charlesworth, biomodal Ltd
- 10:45am – 10:55am *Multi-ancestry transcriptome-wide association analyses on single-cell deconvoluted expression data to identify cell-type specific risk genes for colorectal cancer*
Qing Li, Vanderbilt University Medical Center

11:00am – 12:00pm: Polygenic Models of Cancer Risk

11:00am – 11:10am *Translating genomic risk models to clinical trials in a learning health system: the Prostate Cancer integrated Risk Evaluation (P-CARE) model*

Jason Vassy, Harvard Medical School at VA Boston Healthcare System

11:15am - 11:25am *Multi-Ancestry Analysis Identifies Susceptibility Variants and Improves Polygenic Risk Scores for Breast Cancer Subtypes*

Haoyu Zhang, National Cancer Institute

11:30am – 11:40am *Implementation of breast cancer polygenic risk scores in a personalized screening trial*

Yiwey Shieh, Weill Cornell Medicine

11:45am – 11:55am *Population-enriched germline variants in Finns pinpoint shared mechanisms shaping hematopoietic mosaic chromosomal alterations and diverse solid tumors*

Aoxing Liu, Broad Institute & Massachusetts General Hospital