



Fourth Annual **MidAtlantic Bioinformatics Conference** October 11, 2019



**Multi-Omics Data
Integration for
Biomedical Discovery:
Statistical and Machine
Learning Approaches
to Big Data**

October 11, 2019
Convene - Circa Centre
2929 Arch Street, Philadelphia

 **Children's Hospital
of Philadelphia™**
Department of Biomedical &
Health Informatics

 **Institute for
Biomedical
Informatics**

 **Perelman**
School of Medicine
UNIVERSITY OF PENNSYLVANIA

MidAtlantic Bioinformatics Conference

SCHEDULE

7:30 AM

REGISTRATION

8:15 AM

WELCOME

Bryan Wolf, MD, PhD

Chief Scientific Officer

Chair, Department of Biomedical & Health Informatics

Children's Hospital of Philadelphia

CONFERENCE INTRODUCTION

Yi Xing, PhD

Children's Hospital of Philadelphia

8:30 AM

KEYNOTE: GENOMIC VARIANTS IN PEDIATRIC CANCER

Jinghui Zhang, PhD

Member, St. Jude Faculty

Chair, Department of Computational Biology

St. Jude Endowed Chair in Bioinformatics

9:30 AM

MULTI-OMICS DATA BASED PATIENT SURVIVAL PREDICTION WITH DEEP-LEARNING

Lana Garmire, PhD

Associate Professor (with tenure)

Department of Computational Medicine and Bioinformatics

University of Michigan Medical School

10:00 AM

ACCESSIBLE ARTIFICIAL INTELLIGENCE FOR GENOMIC ANALYSIS

Jason Moore, PhD

Edward Rose Professor of Informatics

Director, Institute for Biomedical Informatics

University of Pennsylvania

10:30 AM

MORNING BREAK

10:45 AM

TRANSFER LEARNING FOR CROSS-SPECIES, CROSS-TISSUES, AND SPATIAL SINGLE CELL ANALYSIS

Elana J. Fertig, PhD

Associate Professor of Oncology, Biomedical Engineering, and Applied Mathematics and Statistics

Assistant Director of the Research Program in Quantitative Sciences

Johns Hopkins University, Sidney Kimmel Comprehensive Cancer Center

Multi-Omics Data Integration for Biomedical Discovery: Statistical and Machine Learning Approaches to Big Data

SCHEDULE

11:15 AM

POSTER LIGHTNING ROUND

Ruchi Lohia, Rutgers University
Zhen Miao, University of Pennsylvania
Yang Pan, PhD Children's Hospital of Philadelphia/UCLA
Jinchaun Xing, PhD Rutgers University
Jinli Zhang, PhD Drexel University/Beijing University of Technology

11:45 AM

LUNCH & POSTER SESSION

2:00 PM

KEYNOTE:

Lior Pachter, PhD
Bren Professor of Computational Biology
California Institute of Technology

3:00 PM

ML-BASED ANALYSES OF RARE DISEASES ARE ENHANCED BY TRAINING MODELS ON PUBLIC DATA COMPENDIA

Casey Greene, PhD
Associate Professor
Department of Systems Pharmacology & Translational Therapeutics
University of Pennsylvania Perelman School of Medicine

3:30 PM

AFTERNOON BREAK

3:45 PM

THE GTEX CONSORTIUM ATLAS OF GENETIC REGULATORY EFFECTS ACROSS HUMAN TISSUES

Tuuli Lappalainen, PhD
Core Faculty Member, New York Genome Center
Assistant Professor, Department of Systems Biology
Columbia University

4:15 PM

USING SYSTEMS BIOLOGY TO TRANSLATE FROM SINGLE CELL GENOMICS TO NEXT GENERATION PRECISION MEDICINE

Bruce Aronow, PhD
Co-director, Computational Medicine Center
Professor, Department of Pediatrics, University of Cincinnati
Cincinnati Children's Hospital Medical Center

4:45 PM

CLOSING REMARKS

KEYNOTE SPEAKERS



Jinghui Zhang, PhD
Member, St. Jude Faculty
Chair, Department of Computational Biology
St. Jude Endowed Chair in Bioinformatics

Dr. Zhang is a computational biologist focused on the integrative analysis of large-scale, multi-dimensional genomic data to understand the initiation and progression of diseases. Her research interest has been in the development of highly accurate and sensitive computational methods for analyzing large-scale genomic data, especially in the area of detecting and analyzing genetic variations and somatic mutations. Most recently her team developed a suite of algorithms for discovery of structural variations and copy number alterations in cancer genome at high accuracy. Dr. Zhang developed mathematical models for mapping clonal evolution trajectory from diagnosis to relapse for pediatric solid tumor and leukemia and has led major studies that have examined the nature of inherited genetic variants in both human and mouse genomes including describing single nucleotide variants in one of the first sequenced human genomes and prevalence of germline mutations causing cancer susceptibility in >1000 children with cancer. She earned her undergraduate degree from Fu Dan University, Shanghai, China and her Master's and PhD from the University of Connecticut.



Lior Pachter, PhD
Bren Professor of Computational Biology
California Institute of Technology

Professor Pachter is a computational biologist working in genomics. His career began in comparative genomics, and he initially was interested in genome alignment, annotation, and the determination of conserved regions using phylogenetic methods. More recently he has focused on functional genomics, which includes answering questions about the function and interaction of DNA, RNA and protein products. He's particularly interested in applications of high-throughput sequencing to RNA biology. Genomics requires the development of algorithms, statistical methodology and mathematical foundations, and a major part of his research is therefore on methods. Dr. Pachter earned his undergraduate degree at California Institute of Technology and his PhD from Massachusetts Institute of Technology.

Multi-Omics Data Integration for Biomedical Discovery: Statistical and Machine Learning Approaches to Big Data

SPEAKERS



Bruce Aronow, PhD

Co-director, Computational Medicine Center

Professor, Department of Pediatrics, University of Cincinnati
Cincinnati Children's Hospital Medical Center

Dr. Aronow is a computational geneticist and developmental biologist. His group carries out analyses of a large variety of data, develops algorithms, and builds websites and databases. These tools allow researchers from varying disciplines and backgrounds to analyze genetic and genomic data to better understand, model, and carry out new research about normal development and disease. Recent areas of interest include large-scale clinical sample analyses, single cell-based dissection of developmental and disease tissues, and in vitro stem cell-based modeling of normal and disease-affected tissues including abnormal neurological, immunological, cardiac, and cancer tissues. The lab's recent efforts focus on predicting new therapeutic approaches based on disease mechanisms in the areas of inflammatory bowel disease, eosinophilic esophagitis, sickle cell anemia, cardiac development, and neurological and psychiatric diseases. His group is working on efforts to define the transcriptome of the developing kidney, lung and brain using stem cell-derived cells and organoids to dissect mechanisms that underlie organ development and function as well as oncogenesis. They are also working to infer novel disease indications for known drugs by semantically linking drug action and disease mechanism relationships. Dr. Aronow completed his undergraduate degree at Stanford University and his PhD at the University of Kentucky.



Elana Fertig, PhD

Associate Professor of Oncology, Biomedical Engineering,
and Applied Mathematics and Statistics

Assistant Director of the Research Program in Quantitative Sciences
Johns Hopkins University, Sidney Kimmel Comprehensive Cancer Center

Dr. Fertig is an Associate Professor of Oncology and Assistant Director of the Research Program in Quantitative Sciences at Johns Hopkins University, with secondary appointments in Biomedical Engineering and Applied Mathematics and Statistics, affiliations in the Institute of Computational Medicine, Center for Computational Genomics, Machine Learning, Mathematical Institute for Data Science, and the Center for Computational Biology. Prior to entering the field of computational cancer biology, she was a NASA research fellow in numerical weather prediction. Dr. Fertig's research is featured in numerous peer reviewed publications, R/Bioconductor packages, and independent research funding. She led the team that won the HPN-DREAM8 algorithm to predict phospho-proteomic trajectories from therapeutic response in cancer cells. Dr. Fertig's lab pursues research in the systems biology of cancer and therapeutic response. She develops computational methods for pattern detection from genomics data and integration of diverse high-throughput data modalities. These algorithms are applied to analyze data from diverse cancer types, with a primary focus on precision medicine and therapeutic resistance. Dr. Fertig completed her undergraduate degree at Brandeis University and earned her PhD from the University of Maryland.

MidAtlantic Bioinformatics Conference

SPEAKERS



Lana Garmire, PhD

Associate Professor (with tenure)

Department of Computational Medicine and Bioinformatics

University of Michigan Medical School

Dr. Lana Garmire is an Associate Professor (with tenure) and Faculty Director of Bioinformatics Core in the medical school of University of Michigan. Before joining University of Michigan, she rapidly rose to tenure at University of Hawaii Cancer Center, and has become a nationally recognized translational bioinformatics scientist leading a computational and experimental human genomics multidisciplinary team.

Dr. Garmire collaborates with a variety of top researchers nationally and internationally. She has published over 50 papers in high quality journals including Cell and Nature. She has mentored over 30 MD fellows, postdocs, graduate students and undergraduates of various academic backgrounds, in Biology, Mathematics, Physics, (bio)Statistics, Bioengineering, Computer Science and Electrical Engineering. She has served on various NIH study sections. She is an Associate Editor of BMC Bioinformatics and Guest Editor of PLoS Computational Biology. Dr. Garmire obtained the MA degree in Statistics and PhD degree in Comparative Biochemistry from University of California - Berkeley.



Tuuli Lappalainen, PhD

Assistant Professor, Department of Systems Biology

Columbia University

Junior Investigator and Core Member

New York Genome Center

Tuuli Lappalainen is an Assistant Professor in the Department of Systems Biology at Columbia University and a Junior Investigator and Core Member at the New York Genome Center. Her research focuses on functional genetic variation in human populations and its contribution to traits and diseases. Her research group's work at New York Genome Center links computational and population genomics to experimental molecular biology. While their individual projects may focus on specific diseases, the overall goal is to uncover general rules of the genomic sources of human variation. Dr. Lappalainen also seeks to push the discoveries and methods from her research projects further towards clinical applications. She has pioneered the integration of large-scale genome and transcriptome sequencing data to understand how genetic variation affects gene expression, providing insight to cellular mechanisms underlying genetic risk for disease and has made an important contribution to several international research consortia in human genomics. She received her PhD from University of Helsinki, Finland in 2009, followed by postdoctoral research at University of Geneva, Switzerland and Stanford University.

Multi-Omics Data Integration for Biomedical Discovery: Statistical and Machine Learning Approaches to Big Data

SPEAKERS



Jason Moore , PhD

Edward Rose Professor of Informatics
Director, Institute for Biomedical Informatics
University of Pennsylvania

Jason Moore is the Edward Rose Professor of Informatics and Director of the Penn Institute for Biomedical Informatics. He also serves as Senior Associate Dean for Informatics and Chief of the Division of Informatics in the Department of Biostatistics, Epidemiology, and Informatics. He came to Penn in 2015 from Dartmouth where he was Director of the Institute for Quantitative Biomedical Sciences. He leads an active NIH-funded research program focused on the development of artificial intelligence and machine learning algorithms for the analysis of complex biomedical data. He is the author of the widely used multifactor dimensionality reduction (MDR) method and software that is the leading resource for discovering genetic interactions. His work has been communicated in more than 500 peer-reviewed paper, book chapters, and editorials. In addition to an active research program, Dr. Moore is committed to undergraduate and graduate education. He has trained more than 75 students and postdocs and has founded several interdisciplinary training programs. He is an elected fellow of the American Association for the Advancement of Science (AAAS), an elected fellow of the American College of Medical Informatics (ACMI), an elected fellow of the American Statistical Association (ASA), and was selected as a Kavli fellow of the National Academy of Sciences. Dr. Moore is currently a Penn Fellow and serves as Editor-in-Chief of the journal BioData Mining. He has a PhD in Human Genetics and an MS in Applied Statistics from the University of Michigan.



Casey Greene, PhD

Associate Professor
Department of Systems Pharmacology and Translational Therapeutics
Perelman School of Medicine at the University of Pennsylvania

Dr. Greene is an Associate Professor of Systems Pharmacology and Translational Therapeutics in the Perelman School of Medicine at the University of Pennsylvania. His lab develops deep learning methods that integrate distinct large-scale datasets to extract the rich and intrinsic information embedded in such integrated data. This approach reveals underlying principles of an organism's genetics, its environment, and its response to that environment. In addition to developing deep learning methods for extracting context, a core mission of his lab is bringing these capabilities into every molecular biology lab. Before starting the Integrative Genomics Lab in 2012, Dr. Greene earned his PhD for his study of gene-gene interactions in the field of computational genetics from Dartmouth College in 2009 and moved to the Lewis-Sigler Institute for Integrative Genomics at Princeton University where he worked as a postdoctoral fellow from 2009-2012. The overarching theme of his work has been the development and evaluation of methods that acknowledge the emergent complexity of biological systems.

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POSTERS

POSTER LIGHTNING ROUND

Physicochemical and Functional Effects of Disease Associated Mutations in Disordered Proteins

Ruchi Lohia, PhD¹, Kaitlin Bassi¹, Matt Hansen, PhD², Grace Brannigan, PhD¹ 1.Rutgers University; 2.University of Pennsylvania

Characterizing Cellular Communication in Human Central Nervous System by Single-Cell RNA Sequencing

Zhen Miao^{1,2}, Gang Hu², Kui Wang², Aivi Nguyen³, Yafei Lyu², Justin Lakkis², Christianne Strang⁴, Christine A. Curcio⁵, Dwight Stambolian⁶, Edward Lee³, Mingyao Li² 1.Graduate Group in Genomics and Computational Biology, University of Pennsylvania Perelman School of Medicine; 2. Department of Biostatistics, Epidemiology and Informatics, University of Pennsylvania Perelman School of Medicine; 3. Department of Pathology and Laboratory Medicine, University of Pennsylvania Perelman School of Medicine; 4. Department of Psychology, University of Alabama at Birmingham; 5. Department of Ophthalmology and Human Genetics, University of Pennsylvania Perelman School of Medicine; 6. Department of Ophthalmology and Visual Sciences, University of Alabama at Birmingham.

PEGASAS: A Pathway-guided Approach for Analyzing Pre-mRNA Alternative Splicing During Cancer Progression

John W Phillips, Yang Pan, Brandon L Tsai, Zhijie Xie, Levon Demirdjian, Wen Xiao, Harry T Yang, Yida Zhang, Chia-Ho Lin, Donghui Cheng, Douglas L Black, Owen N Witte, Yi Xing 1.University of California, Los Angeles; 2. Center for Computational and Genomic Medicine, The Children's Hospital of Philadelphia

Whole Exome Sequencing Links Mutations in Centrosomal Genes to Human Aneuploid Conception Risk

Katarzyna M. Tyc,¹ Warif El Yakoubi,¹ Aishee Bag,¹ Jessica Landis,² Yiping Zhan,² Nathan R. Treff,³ Richard T. Scott, Jr.³, Xin Tao², Karen Schindler¹, Jinchuan Xing¹ 1. Department of Genetics, Rutgers University; 2. Foundation for Embryonic Competence; 3. Reproductive Medicine Associates of New Jersey

Predicting Disease-related Associations Based on A Novel Graph Adversarial Network

Jinli Zhang^{1,2}, Xiaohua Tony Hu², Zongli Jiang¹, Bo Song², Wei Quan² 1. Beijing University of Technology; 2. Drexel University College of Informatics

Integrative Single-Cell and Bulk RNA-Seq Analysis in Human Retina Identified Cell Type-Specific Composition and Gene Expression Changes for Age-Related Macular Degeneration

Yafei Lyu, Randy Zauhar, Nico Dana, Christianne Strang, Kui Wang, Zhen Miao, Paul Gamlin, Christine Curcio, Dwight Stambolian, Mingyao Li University of Pennsylvania

Genome-wide Network Analysis Reveals Perturbations in the Immune System, Synaptic, and Hippo Signaling in Association with Cognitive Decline in Late-onset Alzheimer's Disease

John Stephen Malamon, MSE, PhD Candidate, Andres Kriete, PhD Drexel University

PhENominal: Design and Implementation of an EHR-integrated Phenotype Annotation Tool

Bimal Desai, MD, MBI¹, Malar Singaravelu, MS², Livija Medne, MS, LCGC³, Division of Human Genetics, Ian Krantz, MD³ 1. Division of General Pediatrics, Children's Hospital of Philadelphia; 2. Digital Health, Children's Hospital of Philadelphia Division of Human Genetics; 3. Children's Hospital of Philadelphia

Multi-Omics Data Integration for Biomedical Discovery: Statistical and Machine Learning Approaches to Big Data

POSTERS

Dnmt3a and Dnmt3b-decommissioned Fetal Enhancers Are Linked to Kidney Disease

Yuting Guan, Hongbo Liu, Ziyuan Ma, Szu-Yuan Li, Jihwan Park, Xin Sheng, Katalin Susztak, University of Pennsylvania

An Integrative View of the Regulatory and Transcriptional Landscapes in Mouse Hematopoiesis

Guanjue Xiang¹, Cheryl A. Keller¹, Elisabeth Heuston², Belinda M. Giardine¹, Lin An¹, Alexander Q. Wixom¹, Amber Miller¹, April Cockburn¹, Jens Lichtenberg², Berthold Göttgens³, Qunhua Li¹, David Bodine², Shaun Mahony¹, James Taylor⁴, Gerd A. Blobel⁷, Mitchell J. Weiss⁶, Yong Cheng⁶, Feng Yue¹, Jim Hughes⁵, Douglas R. Higgs⁵, Yu Zhang¹, Ross C. Hardison¹ 1. Department of Biochemistry and Molecular Biology, The Pennsylvania State University; 2. NHGRI Hematopoiesis Section, National Institutes of Health; 3. University of Cambridge, Cambridge, UK; 4. Johns Hopkins University, Baltimore; 5. Oxford University; 6. St. Jude Children's Research Hospital; 7. Children's Hospital of Philadelphia

Incorporating Single-cell RNA-Seq Data to Infer Allele-specific Expression

Jiaxin Fan, Rui Xiao, Mingyao Li, Department of Biostatistics, Epidemiology and Informatics, University of Pennsylvania Perelman School of Medicine

Markov Affinity Proteogenomic Signal Diffusion (MAPSD) Reveals Dynamics of Multi-omics Data Integration in Brain Disorders

Abolfazl Doostparast Torshizi¹, Kai Wang, PhD^{1,2} 1. Center for Cellular and Molecular Therapeutics Children's Hospital of Philadelphia; 2. Department of Pathology and Laboratory Medicine, University of Pennsylvania Perelman School of Medicine

LinkedSV: Detection of Mosaic Structural Variants from Linked-read Exome and Genome Sequencing Data

Li Fang¹, Charly Kao², Michael V Gonzalez², Fernanda A Mafra², Renata Pellegrino da Silva², Mingyao Li³, Sören Wenzel⁴, Katharina Wimmer⁴, Hakon Hakonarson^{2,5}, Kai Wang^{1,6} 1. Raymond G. Perelman Center for Cellular and Molecular Therapeutics, Children's Hospital of Philadelphia; 2. Center for Applied Genomics, Children's Hospital of Philadelphia; 3. Department of Biostatistics, University of Pennsylvania; 4. Section for Human Genetics, Department of Medical Genetics, Molecular and Clinical Pharmacology, Medical University Innsbruck; 5. Department of Pediatrics, University of Pennsylvania; 6. Department of Pathology and Laboratory Medicine, University of Pennsylvania

Tissue Specificity of Gene Expression Impacted TWAS Gene Prioritization Power

Binglan Li,BS¹, Yogasudha Veturi, PhD², David W. Haas MD, PhD³, Marylyn D. Ritchie PhD⁴ 1. Genomics and Computational Biology Program, University of Pennsylvania; 2. Department of Genetics, University of Pennsylvania; 3. Departments of Medicine, Pharmacology, Pathology, Microbiology & Immunology, Vanderbilt University School of Medicine; Department of Internal Medicine, Meharry Medical College; 4. Institute for Biomedical Informatics, University of Pennsylvania

Interpretable and Integrative Deep Learning for Survival Analysis Using Histopathological Images and Genomic Data

Jie Hao PhD¹, Sai C. Kosaraju MS², Nelson Z. Tsaku MS³, Dae Hyun Song MD⁴, Mignon Kang PhD² 1. Department of Biostatistics, Epidemiology and Informatics, University of Pennsylvania School of Medicine; 2. Department of Computer Science, University of Nevada, Las Vegas; 3. Department of Computer Science, Kennesaw State University; 4. Department of Pathology, Gyeongsang National University Changwon Hospital

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POSTERS

Expanding Polygenic Risk Scores to Include Automatic Genotype Encodings and Gene-gene Interactions

Trang T. Le, PhD¹, Hoyt Gong¹, Patryk Orzechowski, PhD², Elisabetta Manduchi, PhD², Jason Moore, PhD²
1. Department of Biostatistics, Epidemiology and Informatics, University of Pennsylvania; 2. Institute for Biomedical Informatics, University of Pennsylvania

A Single Cell RNA-SEQ Based Pooled CRISPR Screen (CROP-SEQ) to Identify Molecular Drivers in the Nucleic Acid Sensing Pathway

Mugdha Khaladkar, Amit Grover, Aidan MacNamara, Ketil Tvermosegaard, Klio Maratou, GlaxoSmithKline

Aberrant Splicing in B-cell acute Lymphoblastic Leukemia

Ammar S. Naqvi, PhD^{1,2}, Kathryn Black, PhD^{2,3}, Mukta Asnani, PhD^{2,3}, PA Katharina E. Hayer^{1,2,3}, Scarlett Yang^{4,5}, Elisabeth Gillespie, PhD⁴, Asen Bagashev, PhD^{2,3}, Vinodh Pillai, MD/PhD⁴, Sarah K. Tasian, MD⁶, Matthew Gazzara⁶, Martin Carroll, MD⁷, Deanne Taylor, PhD^{1,10}, Kristin W. Lynch, PhD^{5,8}, Yoseph Barash, PhD^{6,9}, Andrei Thomas-Tikhonenko, PhD^{2,4,5,10} 1. Department of Biomedical & Health Informatics, Children's Hospital of Philadelphia; 2. Department of Pathology and Laboratory Medicine, Children's Hospital of Philadelphia; 3. Division of Cancer Pathobiology, Children's Hospital of Philadelphia; 4. Department of Pathology, Children's Hospital of Philadelphia; 5. Immunology Graduate Group, University of Pennsylvania; 6. Department of Genetics, Perelman School of Medicine, University of Pennsylvania; 7. Department of Medicine, Perelman School of Medicine; 8. Department of Biochemistry & Biophysics, Perelman School of Medicine, University of Pennsylvania; 9. Department of Computer and Information Science, School of Engineering and Applied Science, University of Pennsylvania; 10. Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania

MicroRNA Frequently Utilize Both Mature Arms to Cooperatively Regulate Tumorigenic Processes Across Cancers

Ramkrishna Mitra, PhD, Clare M. Adams, PhD, Christine M. Eischen, PhD, Thomas Jefferson University

GWAS of Hippocampal Subfield and Neighboring Cortical Structure Volumes Identifies an ERC1 Locus Using ADNI High-Resolution MRI Data

Shan Cong, PhD¹, Xiaohui Yao, PhD², Shannon L. Risacher, PhD³, Kwangsik Nho, PhD³, Andrew J. Saykin, PhD³, Li Shen, PhD² 1. Purdue University; 2. University of Pennsylvania; 3. Indiana University

NITECAP: A Novel Method and Interface for the Identification of Circadian Behavior in Highly Parallel Time-course Data

Thomas G. Brooks, PhD¹, Cris W. Lawrence, PhD¹, Nicholas F. Lahens, PhD¹, Soumyashant Nayak, PhD¹, Dimitra Sarantopoulou, MSc¹, Garret A. FitzGerald, MD², Gregory R. Grant, PhD³ 1. ITMAT Bioinformatics, University of Pennsylvania; 2. ITMAT, Department of Pharmacology, University of Pennsylvania; 3. Department of Genetics, University of Pennsylvania

Tourette Syndrome Multiplex Family Gene Candidate Analysis

Justin Koesterich¹, Yeting Zhang, PhD¹, Robert King, MD³, Jay Tischfield, PhD^{1,2}, Gary Heiman, PhD^{1,2}, Jinchuan Xing, PhD^{1,2} 1. Department of Genetics, Rutgers University; 2. Human Genetics Institute of New Jersey; 3. Child Study Center, Yale School of Medicine

Global Regulation of Chromatin Accessibility, Enhancer and Super-enhancer Architecture in Leukemia by Ikaros Tumor Suppressor

Yali Ding, Bo Zhang, Jonathon L. Payne, Feng Yue, Sinisa Dovat, Penn State University College of Medicine

Multi-Omics Data Integration for Biomedical Discovery: Statistical and Machine Learning Approaches to Big Data

POSTERS

BEERS 2: The Next Generation of RNA-Seq Simulator

Nicholas F. Lahens¹, Thomas G. Brooks¹, Dimitra Sarantopoulou¹, Soumyashant Nayak¹, Cris Lawrence¹, Anand Srinivasan², Jonathan Schug^{3,4}, Garret A. FitzGerald^{1,5}, John B. Hogenesch⁶, Yoseph Barash⁴, Gregory R. Grant^{1,4} 1. Institute for Translational Medicine and Therapeutics, University of Pennsylvania Perelman School of Medicine; 2. PMACS Enterprise Research Applications and High-Performance Computing, University of Pennsylvania Perelman School of Medicine; 3. Institute for Diabetes, Obesity, and Metabolism, University of Pennsylvania Perelman School of Medicine; 4. Department of Genetics, University of Pennsylvania Perelman School of Medicine; 5. Department of System Pharmacology and Translational Therapeutics, University of Pennsylvania Perelman School of Medicine; 6. Division of Human Genetics, Department of Pediatrics, Center for Chronobiology Cincinnati Children's Hospital Medical Center

Genetic Variation and miRNAs Fine-tune A-to-I RNA Editing in Human Tissues

Eddie Park, PhD¹, Lili Hao, PhD², Zhang Zhang, PhD², Yi Xing, PhD^{1,3} 1. Center for Computational and Genomic Medicine, Children's Hospital of Philadelphia; 2. National Genomics Data Center & BIG Data Center & CAS Key Laboratory of Genome Sciences and Information, Beijing Institute of Genomics, Chinese Academy of Sciences; 3. Department of Pathology and Laboratory Medicine, University of Pennsylvania

Whole Genome Detection of Short Tandem Repeats by Deep Convolutional Neural Network on Oxford Nanopore Sequencing Signal Data

Qian Liu, PhD¹, Kai Wang, PhD^{1,2} 1. Raymond G. Perelman Center for Cellular and Molecular Therapeutics, Children's Hospital of Philadelphia; 2. Department of Pathology and Laboratory Medicine, Perelman School of Medicine, University of Pennsylvania

Genetics of Congenital Heart Defect Differences between 22q11.2 Deletion Syndrome Patients

Nhat Duong^{1,2}, Steven Pastor¹, Deanne Taylor¹, Bernice E. Morrow³, Michael Xie¹, Beverly S. Emanuel^{4,5} 1. Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia; 2. School of Biomedical Engineering, Sciences and Health Systems, Drexel University; 3. Department of Genetics, Albert Einstein College of Medicine 4. Division of Human Genetics, Children's Hospital of Philadelphia; 5. Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania

Single-Cell Transcriptomic Analysis of mIHC Images via Antigen Mapping

Kiya Govek, Emma Troisi, Steven Woodhouse, PhD, Pablo G. Camara, PhD, Department of Genetics and Institute for Biomedical Informatics, Perelman School of Medicine, University of Pennsylvania

Interactive VCF Parser for Graphical Display of Structural Variants

James J. Kelley, MS^{1,2,3}, Omkar Asawale^{1,3}, Andrey Grigoriev, PhD^{1,2} 1. Department of Biology, Rutgers University; 2. Center of Computational and Integrative Biology, Rutgers University; 3. Department of Computer Science, Rutgers University

Inference of Epistasis and Entrenchment Leading to Drug Resistance in HIV-1 Subtype B/ Epistasis of Genetic Variants, Computational Systems Biology and Biological Network Modeling

Avik Biswas, MS^{1,2}, Allan Haldane, PhD^{1,2}, Eddy Arnold, PhD^{3,4}, Ronald M Levy, PhD^{1,2} 1. Center for Biophysics and Computational Biology, Temple University; 2. Department of Physics, Temple University; 3. Center for Advanced Biotechnology and Medicine, Rutgers University; 4. Department of Chemistry and Chemical Biology, Rutgers University

Database Characterization of BAP1 Loss in Cancer

Timothy J. Purwin, MS¹ and Andrew E. Aplin, PhD^{1, 2} 1. Department of Cancer Biology, Thomas Jefferson University; 2. Sidney Kimmel Cancer Center, Thomas Jefferson University

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POSTERS

Regional Variation of Splicing QTLs in Human Brain

Yida Zhang¹, Harry Yang¹, Kate Kadash-Edmondson², Yang Pan¹, Zhicheng Pan¹, Beverly L. Davidson^{4,5}, Yi Xing^{1,2,3,5}. 1. Bioinformatics Interdepartmental Graduate Program, University of California, Los Angeles; 2. Center for Computational and Genomic Medicine, Children's Hospital of Philadelphia; 3. Department of Microbiology, Immunology & Molecular Genetics, University of California, Los Angeles; 4. The Raymond G Perleman Center for Cellular and Molecular Therapy, Children's Hospital of Philadelphia; 5. Department of Pathology and Laboratory Medicine, University of Pennsylvania

Digital Preservation of Biological and Health Data

Christiana Dobrzynski MS/MA, Perry Evans PhD, Nicole Feldman, Stephanie Huang, Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia

GADB: Large-scale, Curated Functional Genomics Annotation Database

Pavel P Kuksa¹, Prabhakaran Gangadharan¹, Chien-Yueh Lee¹, Yi-Fan Chou¹, Emily Greenfest-Allen^{1,2}, Han-Jen Lin¹, Ž. Katanić¹, Otto Valladares¹, Yuk Yee Leung¹, Li-San Wang¹. 1. Penn Neurodegeneration Genomics Center, Department of Pathology and Laboratory Medicine, Perelman School of Medicine, University of Pennsylvania; 2. Department of Genetics, Perelman School of Medicine, University of Pennsylvania

SparkINFERNO: Scalable Spark-based Framework for Inferring Dysregulated Enhancer and Noncoding RNAs for WGS and GWAS data

C.-Y. Lee¹, P.P. Kuksa¹, A. Amlie-Wolf¹, E.E. Mlynarski¹, Y.-F. Chou¹, H.-J. Lin¹, E. Greenfest-Allen¹, Z. Katanić¹, O. Valladares¹, A. Kuzma¹, A. Najz¹, G.D. Schellenberg¹, Y.Y. Leung¹, L.-S. Wang¹, Alzheimer's Disease Sequencing Project. 1. Penn Neurodegeneration Genomics Center, Department of Pathology and Laboratory Medicine, Perelman School of Medicine, University of Pennsylvania; 2. Department of Biostatistics, Epidemiology, and Informatics/Center for Clinical Epidemiology and Biostatistics, Perelman School of Medicine, University of Pennsylvania

A Pathway Activity Inference on Multi-omics Data Using Directed Random Walks for Predicting Survival in Bladder Cancer

So Yeon Kim^{1,2}, Eun Kyung Choe¹, Manu Shivakumar¹, Dokyoon Kim^{1,3}, Kyung-Ah Sohn². 1. Department of Biostatistics, Epidemiology & Informatics, Perelman School of Medicine, University of Pennsylvania; 2. Department of Software and Computer Engineering, Ajou University; 3. Institute for Biomedical Informatics, University of Pennsylvania

The Impact of Undesired Technical Variability on Large-scale Data Compendia

Alexandra J. Lee^{1,2}, YoSon Park², Georgia Doing³, Deborah Hogan³, Casey S. Greene². 1. Genomics and Computational Biology Graduate Program, University of Pennsylvania; 2. Department of Systems Pharmacology and Translational Therapeutics, University of Pennsylvania; 3. Department of Microbiology and Immunology, Geisel School of Medicine, Dartmouth University

Reusing Label Functions to Extract Multiple Types of Relationships from Biomedical Abstracts at Scale

David N. Nicholson^{1,2}, Daniel S. Himmelstein², Casey S. Greene². 1. Genomics and Computational Biology Graduate Program, University of Pennsylvania; 2. Department of Systems Pharmacology and Translational Therapeutics, University of Pennsylvania

The Impact of Mobile Element on Gene Expression and Splicing

Xiaolong Cao, Yeting Zhang, Hannah Lords, Jinchuan Xing. Department of Genetics, Rutgers University

Multi-Omics Data Integration for Biomedical Discovery: Statistical and Machine Learning Approaches to Big Data

POSTERS

NOPE: Neural Network Oriented for Prediction of Epitopes

Karami, A¹, Huuki, L.¹, Kaplan, D² 1. Temple University; 2. Johnson & Johnson

A Framework to Track Complex Participant Familial Relationships for a Birth Defects Biorepository

Suzanne Gerace¹, Alex S. Felmeister, PhD¹, Monica Molina, MS¹, Alexander Gonzalez, MS¹, Edward Krause, MS¹, Rebecca L. Linn, MD¹, Vivek Narayanan Padmanabhan, MS¹, Janine Pappas¹, Lillie Rizack, MSN, CNM¹, Erica M. Schindewolf, MS, LCGC¹, Donna Stephan, RN BSN¹, Remo M. S. Williams, MS¹, Stacy Woyciechowski, MS, LCGC¹, Julie S. Moldenhauer, MD, FACOG, FACMG^{1,2}, J. William Gaynor, MD^{1,2}
1. Children's Hospital of Philadelphia; 2. Perelman School of Medicine at the University of Pennsylvania

Pan-Omic Analysis of Diffuse Intrinsic Pontine Glioma from Children Enrolled in the PNOC003 Precision Medicine Trial Identifies Opportunities and Challenges in Clinical Implementation of a Multi-omics Sequencing Approach

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Investigating the Role of miRNAs in Human Female Fertility

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Detecting Allele-specific Alternative Splicing from Population-scale RNA-seq Data

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Evaluating the Impact of Multiple Simultaneous Mutations on Evolutionary Rate Inference

Alexander G. Lucaci, Sadie Wisotsky, PhD, Sergei Pond, PhD, Temple University

Investigating the Genetic Architecture of Psychiatric Disorders and their Medical Comorbidity

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Secure Molecular Epidemiological Analysis Through Encryption

Jordan Zehr, Steven Weaver, Sergei Pond PhD, Temple University

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GRAMc, an Unbiased Genome-scale Reporter Assay Method, Identifies Alu Elements as a Major Contributor of the Human cis-regulatory Genome

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annoFuse: An R Package to Annotate and Prioritize Putative Oncogenic RNA Fusions

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Benchmarking Somatic Simple Variants to Generate Consensus Calls

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Impact of Utilizing Default Parameters in Copy Number Pipelines

David Curley¹, Miguel Brown¹, Yuankun Zhu¹, Krutika Gaonkar^{1,2}, Adam Resnick^{1,2,3}, Jo Lynne Rokita^{1,2} 1. Center for Data-Driven Discovery in Biomedicine, Children's Hospital of Philadelphia; 2. Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia; 3. Division of Neurosurgery, Children's Hospital of Philadelphia

Non-coding Regulatory Landscape of Alzheimer's Disease Variants using GWAS of 63,926 Individuals

Yuk Yee Leung, Pavel Kuksa, Chien-Yueh Lee, Yi-Fan Chou, Alex Amlie-Wolf, Gerard Schellenberg, Li-San Wang, University of Pennsylvania Neurodegeneration Genomics Center

Clues Into the Pathogenesis of Juvenile Onset Aniridic Cataract from RNAseq Data

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Cancer Survival Analysis Using RNA Sequencing and Clinical Data

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Serum Proteomics Analysis Identifies Early Indicators of Response to Anti-IL6 Therapy in Idiopathic Multicentric Castelman Disease

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OpenPBTA: An Open Pediatric Brain Tumor Atlas

Contributors of The OpenPBTA Project

Multi-Omics Data Integration for Biomedical Discovery: Statistical and Machine Learning Approaches to Big Data

POSTERS

Accelerating Next Generation Sequencing Data Analysis Via Optimized GATK Algorithms, IBM POWER9, and Nvidia GPUs

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Shared Genetic Influences on Comprehensive Health Check-up Database Across 10,349 Korean Population

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Can Synonymous Mutations Cause Cancer?

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TADCompare: an R Package for Differential Analysis and Characterization of Topologically Associated Domains

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Gene Scoring Algorithms with Partial Graph Fusions for Identifying Disease Candidate Genes

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Corneal Keratopathy Due to Pax6 Mutation May Result from a Transcriptomic Shift Towards Genes Specifying Conjunctival Characteristics

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Cholesterol Associated with Copy Number Variation Burden in a Study Using 10,000 Participants from the eMERGE Network

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Gene-based Burden Tests Suggest Involvement of Phagosome Pathway in Response to Infliximab in Patients with Very Early Onset Inflammatory Bowel Disease

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Epigenetic Regulation of Oas1 Amplifies Tumor Intrinsic Type I Interferon Signaling to Drive Resistance to Immunotherapy

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Identification of Immunotherapeutic Targets Driven by the Core Regulatory Circuit in Neuroblastoma.

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ClinVar Workflow: a Tool for Variant Clinical Data Analysis & Visualization

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Identifying Reported (likely) Pathogenic Single Nucleotide Variants for Rare Genetic Diseases in Germline Genomic Data from Pediatric Brain Cancer Patients: a Pilot Study in CBTTCC

Yiran Guo, PhD, Yuankun Zhu, BSc, Adam Resnick, PhD, Center for Data Driven Discovery in Biomedicine, Children's Hospital of Philadelphia.

Natural Selection and Demographic History Influence the Distributions of Ages of Rare Variants

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Comparison of Feature Selection Methods for Prediction of CHD

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Identification of Differentially Expressed Proteins in Mice Models of Down Syndrome Using Bipartite Networks

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Multi-Omics Data Integration for Biomedical Discovery: Statistical and Machine Learning Approaches to Big Data

POSTERS

Elucidating Dynamics and Regulation of Alternative Splicing Using Time-course RNA-seq Data

Yuan Yuan Wang, Rene Chun, Samir Adhikari, Christopher M. Lopez, Lan Lin, John Adams, Yi Xing,
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Improved Prediction Modeling of Alzheimer's Disease Using Knowledge-based Multiomic Data Integration

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Patient Adipose Stem Cell-Derived Adipocytes Reveal Genetic Variation that Predicts Antidiabetic Drug Response

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Image Processing on the Brain: 3D Printing MRI Images for Further Observation

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