7TH ANNUAL MIDATLANTIC BIOINFORMATICS CONFERENCE

AN IN-PERSON LIVE NETWORKING EVENT

COMPUTATIONAL APPROACHES TO DIFFERENTATION IN DISEASES

HOSTED BY

Children's Hospital of Philadelphia®

Penn Institute for Biomedical Informatics

Perelman SCHOOL OF MEDICINE UNIVERSITY of PENNSYLVANIA

AGENDA

8:30 AM	Welcome Yi Xing, PhD Executive Director, Department of Biomedical and Health Informatics Children's Hospital of Philadelphia Marylyn Ritchie, PhD Director of the Institute for Biomedical Informatics (IBI) University of Pennsylvania Perelman School of Medicine
8:45 AM - 9:30 AM	**Keynote Presentation #1: Karen Miga, PhD** Assistant Professor in the Biomolecular Engineering Department at UCSC Associate Director at the UCSC Genomics Institute University of Kiel *Discovering New Biology and Function in Complete, Telomere-to-Telomere (T2T) Genomes*
9:30 AM – 10:00 AM	Nilanjan Chatterjee, PhD Bloomberg Distinguished Professor Department of Biostatistics Bloomberg School of Public Health Department of Oncology School of Medicine Johns Hopkins University *Polygenic Risk Scores: Turning Dust into Gold?*
10:00 AM – 10:15 AM	Morning Break
10:15 AM – 10:45 AM	Gina Peloso, PhD Assistant Professor, Genetics Biostatistics Boston University *Implicating Genes, Pleiotropy, and Sexual Dimorphism at Blood Lipid Loci*
10:45 AM – 11:15 AM	Greg Bowman, PhD Professor Director of Folding@home Department of Bioengineering and Biochemistry University of Pennsylvania *Searching the Genome for Cryptic Pockets*

AGENDA CONT'D

11:15 AM – 11:45 AM	*Poster Lightning Round* *Presentations of 7 selected posters* *Sammy Hansali, Rutgers University* *Hongbo Liu, University of Pennsylvania* *Karleena Rybacki, Children's Hospital of Philadelphia* *Jason Liu, Rutgers University* *Alison Merikangas, Children's Hospital of Philadelphia* *Jacob Leiby, University of Pennsylvania* *Jennifer Guo, University of Pennsylvania*
11:45 PM – 1:45 PM	*Break for Lunch* *Poster Networking Session* *Roberts Center for Pediatric Research Lobby* *Posters will be on display in the main lobby and in the Level B lobby.*
1:45 PM – 2:30 PM	**Keynote Presentation #2: Bogdan Pasaniuc, PhD** Associate Professor Pathology and Laboratory Medicine, Human Genetics, and Computational Medicine University of California, Los Angeles *Polygenic Risk Scores for Individuals of Diverse Ancestries Mining*
2:30 - 3:00 PM	Iuliana Ionita-Laza, PhD Professor Department of Biostatistics Columbia University *Knockoff-based Statistics for the Identification of Putative Causal Loci in Genetic Studies*
3:00 PM – 3:15 PM	Afternoon Break
3:15 PM - 3:45 PM	Corey McMillan, PhD Associate Professor, Neurology Perelman School of Medicine University of Pennsylvania *Molecular Profiling of Alzheimer's Disease and Age-Related Neuorpathology*

3:45 PM - 4:15 PM

Duygu Ucar, PhD
Associate Professor
Epigenomics & Genomics
Jackson Laboratory for Genomic Medicine

Marylyn Ritchie, PhD
Director of the Institute for Biomedical Informatics (IBI)

4:15 PM - 4:30 PM

Closing Remarks and Presentation of Trainee Awards
Dokyoon Kim, PhD
Assistant Professor of Informatics in Biostatistics and Epidemiology
Senior Fellow,
Institute of Biomedical Informatics,
Associate Director of Informatics, Immune Health,
Perelman School of Medicine

THANK YOU CONFERENCE OPERATIONS

Ariana Downs, Outreach Specialist

Hannah Lazarus, Research Coordinator

Donna Vito, Outreach Manager

THANK YOU POSTER REVIEW COMMITTEE

Asif Chinwalla, Children's Hospital of Philadelphia

Noor Dawany, Children's Hospital of Philadelphia

Mark Porter, Children's Hospital of Philadelphia

Anurag Verma, University of Pennsylvania

Danielle Mowery, University of Pennsylvania

Joseph Daniel Romano, University of Pennsylvania

Shefali Setia Verma, University of Pennsylvania

KEYNOTE SPEAKERS

Karen Miga, PhD
Director
Human Pangenome Production Center
University of California, Santa Cruz
Named as “One to Watch” in Nature’s 10, 2020

Dr. Miga is an Assistant Professor in the Biomolecular Engineering Department UCSC, and an Associate Director of the UCSC Genomics Institute. In 2019, she co-founded the Telomere-to-Telomere (T2T) Consortium, an open, community-based effort to generate the first complete assembly of a human genome. Additionally, Dr. Miga is the Director of the Reference Production Center for the Human Pangenome Reference Consortium (HPRC). Central to Dr. Miga’s research program is the emphasis on satellite DNA biology and the use of long-read and new genome technologies to construct high-quality genetics and epigenetic maps of human peri/centromeric regions. (Source: University of California Santa Cruz)

Bogdan Pasaniuc, PhD
Department Vice Chair
Computational Medicine
Associate Professor, Pathology and Laboratory Medicine,Human Genetics,
and Computational Medicine
University of California Los Angeles

Bogdan Pasaniuc is an associate professor of Computational Medicine, Human Genetics and Pathology&Lab Medicine at UCLA. His group develops statistical and computational methods to understand the genetic basis of disease, focusing on under-represented populations, integrative genomics, and biobank studies. His group developed new methodologies to integrate epigenetic profiles within trans-ethnic studies to localize causal variants in post-GWAS studies. More recently, his group introduced transcriptome-wide studies based on gene expression imputation as a principled approach to localize causal genes for complex traits, and applied such approaches to identify new risk genes for multiple complex traits such as Schizophrenia or Ovarian Cancer.

SPEAKERS

Duygu Ucar, PhD
Associate Professor
Epigenomics & Genomics
Jackson Laboratory for Genomic Medicine

Dr. Ucar earned her B.S. degree in Computer Engineering from Bilkent University followed by her Ph.D. in Computer Science from the Ohio State University. She conducted postdoctoral studies at University of Iowa and Stanford University, during which she developed computational methods to discover cell-specific epigenomic patterns from ChIP-seq datasets. In 2013, she launched her laboratory at The Jackson Laboratory for Genomic Medicine (JAX-GM). The overarching goal of her lab is to uncover how epigenomic landscape of human cells -especially immune cells- are remodeled and disrupted with aging and aging-related diseases. She also studies how genetic variants impact epigenomic landscapes of human cells in the context of complex diseases, e.g., Type 2 Diabetes (T2D). Towards this goal, she combines advanced computational techniques with state-of-the art NGS approaches (e.g., ATAC-seq, CITE-seq).
(Source: The Jackson Laboratory)

Nilanjan Chatterjee, PhD
Bloomberg Distinguished Professor
Department of Biostatistics
Bloomberg School of Public Health

Dr. Chatterjee is a Bloomberg Distinguished Professor at the Department of Biostatistics, Bloomberg School of Public Health and Department of Oncology School of Medicine at the Johns Hopkins University. Prior to joining Johns Hopkins, he led the Biostatistics Branch of the Division of Cancer Epidemiology and Genetics of the US National Cancer Institute during 2008-2015. He is known for foundational and methodological contributions to multiple areas of modern biomedical data science, including large scale analysis of genetic associations, gene-environment interactions and predictive model building by synthesis of information from multiple data sources. His collaborative research has led to understanding of genetic architecture and role of gene-environment interactions in the etiology of a variety of cancers. He has received numerous prestigious national and international awards, including the Mortimer Spiegelman Award (2010) from the American Public Health Association, both the President's and the Snedecor Award from the Committee of the Presidents of the Statistical Society (2011), Myrto Lefkopoulou Distinguished Lecture (2013) from the Department of Biostatistics Harvard School of Public Health and Norman Breslow Distinguished Lecture (2017) from the Department of Biostatistics University of Washington, Seattle. He serves on the scientific advisory committee of the Radiation Effect Research Foundation, Hiroshima Japan and Population and Prevention Research Committee of the foundation of Cancer Research UK.

Gina Peloso, PhD
Associate Professor
Department of Biostatistics
Boston University

Gina Peloso, PhD, is an Associate Professor in the Department of Biostatistics. Prior to joining BU, she was a post-doctoral fellow in Human Genetics at Massachusetts General Hospital and the Broad Institute. Her primary research focus is statistical genetics. She has contributed to the identification of common genetic variants through genome-wide association studies (GWAS) with complex cardiovascular traits, particularly with plasma lipid levels. She also investigates the role of rare genetic variation using both exome and whole genome sequencing. Her goals for studying genetic variation of blood lipid levels include, first, making insights into the biology behind the traits: What are the underlying causes of high/low lipid levels? Second, to use genetic variants to answer clinically meaningful questions: Can we use genetic variants to predict risk?: Does genetics point to potential therapeutic targets? Finally, to make connections between diseases by shared genetic contributions: What is the relationship between the genetics of blood lipid levels and Alzheimer's disease (AD)? Gina is an active member in the Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) consortium and TOPMed Program, and a lead investigator in the Global Lipid Genetic Consortium (http://lipidgenetics.org/). (Source: Boston University)

Corey McMillan, PhD
Associate Professor, Neurology
University of Pennsylvania
Perelman School of Medicine

Corey McMillan, PhD is associate professor of neurology at the Perelman School of Medicine and a member of the Neuroscience Graduate Group and Institute for Translational Medicine and Therapeutics. Dr. McMillan holds a bachelor's degree in psychology and cognitive neuroscience from Temple University, a master's of science in psycholinguistics from the University of Edinburgh, and a PhD in psychology from the University of Edinburgh.

Dr. McMillan's research focuses on identifying cognitive and biological markers of neurodegenerative diseases like frontotempoeral lobar degeneration, amyotrophic lateral sclerosis, Alzheimer's disease, and Parkinson's disease. His cognitive research focuses on the social and decision-making mechanisms that contribute to language processing deficits associated with neurodegenerative diseases. His biological research uses neuroimaging, genetics, and biofluids in an effort to improve early diagnosis and predict which protein is causing a disease in patients. Dr. McMillan's biomarker research leverages sophisticated bioinformatic and statistical approaches to integrate multiple data sources in effort to identify precision medicine approaches for treating individual patients. Ultimately, Dr. McMillan intends to integrate cognitive and biological tests to develop powerful methods for identifying patients for entry into clinical trials and for measuring the efficacy of drug treatments in the context of clinical trials. (Source: University of Pennsylvania**)**

Iuliana Ionita-Laza, PhD
Professor
Department of Biostatistics
Columbia University

Dr. Ionita-Laza's main research interests lie at the interface between statistics and genomics. She is particularly interested in developing statistical and machine learning methods for the analysis of high-dimensional genetic and functional genomics data. Dr. Ionita-Laza is also involved in applications of such methods to understand the genetic basis of complex diseases and traits, including autism spectrum disorders, schizophrenia and Alzheimer's disease. (Source: Columbia University)

Greg Bowman PhD
Professor
Director of Folding@home
Department of Bioengineering and Biochemistry
University of Pennsylvania

Dr. Ionita-Laza's main research interests lie at the interface between statistics and Dr. Greg Bowman is a biophysicist who combines experiments with massive computer simulations and machine learning to understand protein dynamics, inform the development of new therapeutics, and interpret genetic variation. He also serves as the Director of the Folding@home distributed computing platform, which brings together hundreds of thousands of citizen scientists who volunteer to run simulations on their

LIGHTNING ROUND PRESENTERS

These top posters (in pink) received the highest rankings in the poster review and scoring and will be presented during the Lightning Round at 11:15 AM on Friday October 7, 2022. Attendees will vote for the best poster presentation and 3 will receive $250 Trainee Awards at the close of the conference day.

Causal impact of Mobile Elements (MEs) on neurodevelopmental disorders

Sammy Hansali, Anthony Wong, Anbo Zhou, Xiaolong Cao, Vaidhyanathan Mahaganapathy, Marco Azaro, Christine Gwin, Sherri Wilson, Steven Buyske, Christopher W. Bartlett, Judy F. Flax, Linda M. Brzustowicz, and Jinchuan Xing

Epigenomic and transcriptomic analyses define core cell types, genes and targetable mechanisms for kidney disease

Hongbo Liu1,2,3, Tomohito Doke1,2,3, Dong Guo4, Xin Sheng1,2,3, Ziyuan Ma1,2,3, Joseph Park1,3,5, Ha My T. Vy6,7, Girish N. Nadkarni6,7,8,9, Amin Abedini1,2,3, Zhen Miao1,2,3, Matthew Palmer10, Benjamin F. Voight2,3,11,12, Hongzhe Li13, Christopher D. Brown3, Marylyn D. Ritchie3,5, Yan Shu4 and Katalin Susztak1,2,3 1Department of Medicine, Renal Electrolyte and Hypertension Division, University of Pennsylvania, Philadelphia, PA, USA; 2Institute of Diabetes Obesity and Metabolism, University of Pennsylvania, Philadelphia, PA, USA; 3Department of Genetics, University of Pennsylvania, Philadelphia, PA, USA; 4Department of Pharmaceutical Sciences, School of Pharmacy, University of Maryland at Baltimore, Baltimore, MD, USA; 5Institute for Biomedical Informatics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, USA; 6Division of Nephrology, Department of Medicine, Icahn School of Medicine at Mount Sinai, New York, NY, USA; 7The Charles Bronfman Institute of Personalized Medicine, Icahn School of Medicine at Mount Sinai, New York, NY, USA; 8The Hasso Plattner Institute of Digital Health, Icahn School of Medicine at Mount Sinai, New York, NY, USA; 9The Mount Sinai Clinical Intelligence Center, Icahn School of Medicine at Mount Sinai, New York, NY, USA; 10Pathology and Laboratory Medicine at the Hospital of the University of Pennsylvania, Philadelphia, , USA; 11Department of Systems Pharmacology and Translational Therapeutics, University of Pennsylvania, Philadelphia, PA, USA; 12Institute of Translational Medicine and Therapeutics, University of Pennsylvania, Philadelphia, PA, USA; 13Department of Biostatistics, Epidemiology, and Informatics, and Center for Clinical Epidemiology and Biostatistics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, USA

Diagnostic gene fusion detection in cancer by adaptation of short-read CHOP Fusion Panel to Oxford Nanopore long-read sequencing

Karleena Rybacki, BS, 1,2 Li Fang, PhD, 2 Feng Xu, PhD, 3 Yu Hu, PhD, 2 Mian Umair Ahsan, MS, 2 Marilyn Li, MD, 3 Kai Wang, PhD, 1,2 1 University of Pennsylvania 2 Children's Hospital of Philadelphia (CHOP) 3 Division of Genomic Diagnostics at Children's Hospital of Philadelphia (CHOP)

Predicting embryonic aneuploidy rate and identifying candidate genes in IVF patients using synonymousvariants

Jason Liu Siqi Sun Jinchuan Xing Rutgers University

A combined polygenic risk score provides a data-driven biomarker for metabolic syndrome

Leiby, Jacob S.; Choe, Eun Kyung; Kim, Dokyoon University of Pennsylvania School of Medicine

Is common genetic risk for psychiatric disorders associated with traumatic experiences in youth?

Alison K. Merikangas PhD, MPH (1,2,3), Laura M. Schultz PhD (2,3), Zoe Rapisardo-Drigot (1,4), PNC Collaborators (3,5), Laura Almasy PhD (1,2,3) 1. Department of Biomedical and Health Informatics, Children'sHospital of Philadelphia, Philadelphia, PA 2. Department of Genetics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA 3. Lifespan Brain Institute of the Children's Hospital of Philadelphia and the University of Pennsylvania, Philadelphia, PA 4. Department of Psychology andNeuroscience, University of Colorado Boulder, Boulder, CO 5. Department of Psychiatry, Neuropsychiatry Section, Perelman School of Medicine, University of Pennsylvania,

Investigation of Cell-Free DNA Methylation as a Biomarker of Malignant vs. Benign Lung Nodules

Jennifer Guo1; Jacob E. Till, MD, PhD1; Thrusha Puttaraju1; Aseel Abdalla1; Zhouyang Wang1; Danielle Shoshany1; Wanding Zhou, PhD2; Jeffrey Thompson, MD3*; Erica L. Carpenter, MBA, PhD1*; 1Division of Hematology-Oncology, Department of Medicine, Perelman School of Medicine, University of Pennsylvania; 2Center for Computational and Genomic Medicine, The Children's Hospital of Philadelphia; 3Division of Pulmonary, Allergy, and Critical Care Medicine, Department of Medicine, Perelman School of Medicine, University of Pennsylvania; *Co-Senior Author

Seamless integrative pipeline for QTL datasets enhance the discovery of putative causal variants for Alzheimer's Disease.

Jeffrey Cifello(1), Pavel P Kuksa(1), Li-San Wang(1), Yuk Yee Leung(1) 1 Penn Neurodegeneration Genomics Center, Department of Pathology and Laboratory Medicine, University of Pennsylvania

Cladebreaker: Using proteomic novelty to test clonality in outbreaks and epidemics

A Feder1, AM Moustafa2,3, PJ Planet1,3 1- Division of Pediatric Infectious Diseases, Children's Hospital of Philadelphia, Philadelphia, PA, 19104, USA 2- Division of Gastroenterology, Hepatology & Nutrition, Children's Hospital of Philadelphia, Philadelphia, PA, 19104, USA 3- Department of Pediatrics, Perelman College of Medicine, University of Pennsylvania, Philadelphia, PA, 19104, USA

Single-cell Genomics Study of Fontan-associated Liver Disease

Po Hu, B.S., Juanjuan Zhao, Ph.D., Wenbao Yu, Ph.D., Benjamin Wilkins, M.D., Ph.D., Aidan Bauer, B.S., Kai Tan, Ph.D., Jack Rychik, M.D., Liming Pei, Ph.D.

Genetic Heterogeneity of Four Deep Learning-derived MCI/AD Dimensions via Genome-wide Tiling Associations

Jiong Chen1,2,3, Junhao Wen, PhD1,2, Zhijian Yang1,2, Yuhan Cui1,2, Jingxuan Bao4, Brian N Lee2, Guray Erus, PhD1,2, Sarah Wait Zaranek, PhD5, Alexander Wait Zaranek, PhD5, Yong Fan, PhD1,2, Andrew J. Saykin, MS, PsyD6, Paul M. Thompson, PhD7, Li Shen, PhD4, Haochang Shou, PhD1,8, Ilya M. Nasrallah, MD, PhD1,2, Christos Davatzikos, PhD1,2,9,10 1Center for Biomedical Image Computing and Analytics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, USA 2Department of Radiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, USA 3Department of Bioengineering, School of Engineering and Applied Science, University of Pennsylvania, Philadelphia, USA 4Department of Biostatistics, Epidemiology and Informatics, University of Pennsylvania, Philadelphia, USA 5Curii Corporation, Somerville, MA, USA 6Center for Neuroimaging, Department of Radiology and Imaging Sciences, and the Indiana Alzheimer's Disease Research Center, Indiana University School of Medicine, Indianapolis, USA 7Keck School of Medicine, University of Southern California, Los Angeles, CA, USA 8Penn Statistics in Imaging and Visualization Center, Department of Biostatistics, Epidemiology, and Informatics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, USA 9For the Alzheimer's Disease Neuroimaging Initiative 10For the AI4AD consortium

Translational Bioinformatics for Heterogenous Longitudinal Data in Pre-Clinical Models of Neurological Injury: From Collection to Therapeutic Target DiscoveryHunter

A. Gaudio1,2, Viveknarayanan Padmanabhan3, Gerard Laurent4, Ryan W. Morgan1,2, Julia Slovis1,2, Frank Mi3, Helen Shi3, Luiz Eduardo Silva3, Wesley B. Baker2,4, Fuchiang Tsui3, Todd J. Kilbaugh1,2, Tiffany S. Ko1,2 1 Department of Anesthesiology and Critical Care Medicine, Children's Hospital of Philadelphia, Philadelphia, PA 2 The Resuscitation Science Center, Children's Hospital of Philadelphia Research Institute, Philadelphia, PA 3 Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia, Philadelphia, PA 4 Division of Neurology, Department of Pediatrics, Children's Hospital of Philadelphia, Philadelphia, PA

Using combinatorial hash-joins to identify SARS-CoV-2 variants.

S Haag1, A Feder2, AM Moustafa3,4, PJ Planet2,4 1-Arcus, Children's Hospital of Philadelphia, Philadelphia, PA, 19104, USA 2- Division of Pediatric Infectious Diseases, Children's Hospital of Philadelphia, Philadelphia, PA, 19104, USA 3- Division of Gastroenterology, Hepatology & Nutrition, Children's Hospital of Philadelphia, Philadelphia, PA, 19104, USA 4- Department of Pediatrics, Perelman College of Medicine, University of Pennsylvania, Philadelphia, PA, 19104, USA

SNAF: Comprehensive prediction of splicing neoantigens for targeted cancer immunotherapy
Guangyuan Li, BS, Cincinnati Children's Hospital Medical Center 2. Anukana Bhattacharjee, PhD, Cincinnati Children's Hospital Medical Center 3. Gloria M. Sheynkman, PhD, University of Virginia 4. Nathan Salomonis, PhD, Cincinnati Children's Hospital Medical Center

Deep Learning of Videourodynamics to Classify Bladder Dysfunction Severity
Weaver JK, Martin-olenski M, Logan J, Broms R, Antony M, Van Batavia J, Weiss D, Long CJ, Smith AL, Zderic SA, Yong F, Tasian GE

A 3D Anatomical and Molecular Map of Cardiac Vagal Motor Neurons
Hornung E, Robbins S, Srivastava A, Achanta S, Schwaber J, and Vadigepalli R

Best Practices for COVID-19 Dashboards
Malkani, Dillon, Bullis School; Malkani, Melina, Bullis School; Singh, Neel, Landon School; Madan, Eesha, BASIS Independent School McLean

Single cell long read sequencing and perturbation analysis reveals splicing heterogeneity and logic
Yuntian Fu:Graduate Group in Genomics and Computational Biology, University of Pennsylvania, Philadelphia, PA, USA Heonseok Kim, Susan M. Grimes, Billy T. Lau, Anuja Sathe,:Division of Oncology, Department of Medicine, Stanford University School of Medicine, Stanford, CA, USA

rMATS-turbo: An efficient and flexible computational tool for alternative splicing analysis of large-scale RNA-seq data
Yuanyuan Wang 1,2,†, Zhijie Xie 2,†, Eric Kutschera 2, Jenea I. Adams 2,3, Kathryn E. Kadash-Edmondson 2, Yi Xing 2,4,5,* 1. Bioinformatics Interdepartmental Graduate Program, University of California, Los Angeles, Los Angeles, CA 90095, USA 2. Center for Computational and Genomic Medicine, The Children's Hospital of Philadelphia, Philadelphia, PA, 19104, USA 3. Genomics and Computational Biology Graduate Program, University of Pennsylvania, Philadelphia, PA, 19104, USA 4. Department of Pathology and Laboratory Medicine, University of Pennsylvania, Philadelphia, PA, 19104, USA 5. Department of Biomedical and Health Informatics, The Children's Hospital of Philadelphia, Philadelphia, PA, 19104, USA *Corresponding author. Phone: (215) 590-0280 †These authors contributed equally to this work

Modules of genotypic variance reflect heterogeneity across TDP-43 proteinopathies
Barbara E Spencer, PhD; David J. Irwin, MD; Vivianna M Van Deerlin, MD, PhD; Eddie B Lee, MD, PhD; Lauren Elman, MD; Colin Quinn, MD; Murray Grossman, MD, EdD; David A. Wolk, MD and Corey T McMillan, PhD University of Pennsylvania, Philadelphia, PA, USA

The Codon Statistics Database: a Database of Codon Usage Bias
Krishnamurthy Subramanian1,2,‡, Bryan Payne1,‡, Felix Feyertag1 and David Alvarez-Ponce1 1 Biology Department, University of Nevada, Reno, Reno, NV, 89557. 2 Department of Genetics, Rutgers, The State University of New Jersey, Piscataway, NJ, 08854

LongReadSum: A fast and flexible quality control tool for long-read sequencing data
[1] Jonathan Elliot Perdomo, BA. Children's Hospital of Philadelphia. [2] Mian Umair Ahsan, MS. Children's Hospital of Philadelphia. [3] Qian Liu, Ph.D. Children's Hospital of Philadelphia. [4] Li Fang, Ph.D. Children's Hospital of Philadelphia. [5] Kai Wang, Ph.D. Children's Hospital of Philadelphia.

Detecting Coupled-Gene Clusters In scRNA-Seq Data Using Deep Learning
Alicia Petrany, Yong Chen PhD.

Functional Impact of Copy Number Variants in Autism Probands
Rohan Alibutud, Vaidhyanathan Mahaganapathy, Xiaolong Cao, Marco Azaro, Christine Gwin, Sherri Wilson, Steven Buyske, Christopher W. Bartlett, Judy F. Flax, Linda M. Brzustowicz, Jinchuan Xing

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Krishnamurthy Subramanian1,2,‡, Bryan Payne1,‡, Felix Feyertag1 and David Alvarez-Ponce1 1 Biology Department, University of Nevada, Reno, Reno, NV, 89557. 2 Department of Genetics, Rutgers, The State University of New Jersey, Piscataway, NJ, 08854

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Analysis of variation in epigenetic age prediction in Domestic dog samples (Canis lupus familiaris)

Nirali Desai1,2, Matteo Pellegrini2 1College of Science, Northeastern University, Boston, MA 25000D Terasaki Life Sciences Building, Molecular, Cell & Developmental Biology Department, University of California, Los Angeles, California Corresponding Author Matteo Pellegrini 5000D Terasaki Life Sciences Building, Molecular, Cell & Developmental Biology Department, University of California, Los Angeles, California

Medulloblastoma subtype single sample predictor built on multiple gene expression platforms

Steven M. Foltz, PhD 1,2, Casey S. Greene, PhD 1,3, Jaclyn N. Taroni, PhD 2 1 Department of Systems Pharmacology and Translational Therapeutics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, USA 2 Childhood Cancer Data Lab, Alex's Lemonade Stand Foundation, Wynnewood, PA, USA 3 Center for Health AI, University of Colorado School of Medicine, Aurora, CO, USA

Distinct cellular trajectories diverge between cancerous and non-cancerous pathologies in the murine esophageal epithelium

Adam Karami, MS(1); Mohammed Faujul Kabir, PhD(1); Alena Klochkova, MS(1); Anbin Mu(1); Yinfei Tan, PhD(2); Andres Klein-Szanto, MD, PhD(2); Kelly A. Whelan, PhD(1) (1)Fels Cancer Institute for Personalized Medicine, Temple University, Philadelphia, PA (2)Department of Pathology and Cancer Biology Program, Fox Chase Cancer Center, Philadelphia, PA

JOnTAD: joint hierarchical TAD caller for high resolution, single cell and bulk cell Hi-C data

Qiuhai Zeng, BS, Pennsylvania State University; Guanjue Xiang, PhD, CAMP4 Therapeutics; Yu Zhang, PhD, Two Sigma; Qunhua Li, PhD, Pennsylvania State University

Linkreg: a Bayesian framework for linking candidate cis-regulatory elements to target genes

Qiuhai Zeng, BS, Pennsylvania State University; Guanjue Xiang, PhD, CAMP4 Therapeutics; Ross Hardison, PhD, Pennsylvania State University; Xiang Zhu, PhD, Pennsylvania State University; Qunhua Li, PhD, Pennsylvania State University

Identifying and Validating Recurrent Structural Variants Affecting Tumor Genomes using GROM and VN

Syeda Aiman Nadeem [1] James J. Kelley, MS [1,2], Andrey Grigoriev, PhD [1,2] 1. Dept of Biology, Rutgers University, Camden, NJ 2. Center of Computational and Integrative Biology, Rutgers University, Camden, NJ

RNA-seq reveals condition-dependent global transcriptional effects of the msf gene in Haemophilus influenzae biofilms

Evangeline Williams, BA; Laura Anastor-Walters, MS; Bhaswati Sen, PhD; Steven Lang, BS; Sergey Balashov, PhD; Rachel L. Ehrlich, MS; Garth D. Ehrlich, PhD; Benjamin Janto, PhD; Joshua Chang Mell PhD AFFILIATIONS: Drexel University College of Medicine, Center for Advanced Microbial Processing, Institute for Molecular Medicine and Infectious Disease

Network-based cross-phenotype risk scoring models for compositing multiple disease risks using biobank-scaled PheWAS data

Yonghyun Nam 1, Vivek Sriram 1, Sang-Hyuk Jung 1,2, Brenda Xiao 1, Manu Shivakumar 1, Anurag Verma 3*, Dokyoon Kim 1,4* 1Department of Biostatistics, Epidemiology & Informatics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA 19104, USA 2Samsung Advanced Institute for Health Sciences and Technology (SAIHST), Sungkyunkwan University, Samsung Medical Center, Seoul 06351, Republic of Korea 3Division of Translational Medicine and Human Genetics, Department of Medicine, University of Pennsylvania, Philadelphia, PA 19104, USA 4Institute for Biomedical Informatics, University of Pennsylvania, Philadelphia, PA 19104, USA

Mapping PTBP splicing in human brain identifies targets for therapeutic splice switching including SYNGAP1

Jennine M. Dawicki-McKenna, PhD 1* Alex J. Felix, PhD 1* Elisa A. Waxman, PhD 2 Congsheng Cheng, PhD 2 Defne A. Amado, MD, PhD 2 Paul T. Ranum, PhD 2 Alexey Bogush, PhD 1 Lea V. Dungan 2 Elizabeth A. Heller, PhD 3 Deborah L. French, PhD 2,4 Beverly L. Davidson, PhD 2,4 Benjamin L. Prosser, PhD 1 * These authors contributed equally to the work. 1 - Department of Physiology, Pennsylvania Muscle Institute, University of Pennsylvania Perelman School of Medicine 2 - Center for Cellular and Molecular Therapeutics, Children's Hospital of Philadelphia 3 - Department of Systems Pharmacology and Translational Therapeutics, University of Pennsylvania Perelman School of Medicine 4 - Department of Pathology and Laboratory Medicine, University of Pennsylvania Perelman School of Medicine

A Novel Genetic Correlation Disease-Disease Network for the Improved Identification of Associated Phenotypes

Jakob Woerner(1,2), Vivek Sriram(1,2), Yonghyun Nam(2), Dokyoon Kim(2,3) 1Genomics and Computational Biology Graduate Group, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA 19104, USA 2Department of Biostatistics, Epidemiology & Informatics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA 19104, USA 3Institute for Biomedical Informatics, University of Pennsylvania, Philadelphia, PA 19104, USA

Repairing the neutral set in codon evolutionary models

Hannah Verdonk (1), Sergei L. Kosakovsky Pond, PhD (1), and Jody Hey, PhD (2) 1. Institute for Genomics and Evolutionary Medicine, Department of Biology, Temple University, Philadelphia, Pennsylvania, USA 2. Center for Computational Genetics and Genomics, Department of Biology, Temple University, Philadelphia, Pennsylvania, USA

The Single-cell Pediatric Cancer Atlas: Open-source data and tools for single-cell transcriptomics of pediatric tumors

Allegra G. Hawkins, PhD. Alex's Lemonade Stand Foundation. Joshua A. Shapiro, PhD. Alex's Lemonade Stand Foundation. Chante Bethell, BS. Alex's Lemonade Stand Foundation. David S. Mejia. HSD. Alex's Lemonade Stand Foundation. Deepa Prasad, MS. Alex's Lemonade Stand Foundation. Nozomi Ichihara, AS. Alex's Lemonade Stand Foundation. Arkadii Yakovets, MS. Alex's Lemonade Stand Foundation. Kurt Wheeler, BS. Alex's Lemonade Stand Foundation (former); Reify Health (current). Steven Foltz PhD. Alex's Lemonade Stand Foundation; University of Pennsylvania. Jennifer O'Malley, MA. Alex's Lemonade Stand Foundation. Stephanie J. Spielman, PhD. Alex's Lemonade Stand Foundation. Jaclyn N. Taroni, PhD. Alex's Lemonade Stand Foundation.

RNA-seq with and without rRNA depletion and varying library prep protocol parameters

Thomas G Brooks 1 Nicholas F Lahens 1 Antonijo Mrčela 1 Shaon Sengupta 1, 2 Peter S Choi 3 Gregory R Grant 1, 4 1. Institute for Translational Medicine and Therapeutics, University of Pennsylvania 2. Division of Neonatology, Children's Hospital of Philadelphia 3. Division of Cancer Pathobiology, Children's Hospital of Philadelphia 4. Department of Genetics, University of Pennsylvania

Comparing Metabolic Pathway Fluxes of Ground-Bound and Space-Flown Mice Using Single-cell RNA-sequencing and a Constraint-based Metabolic Model

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Niche differential gene expression analysis in spatial transcriptomics data identifies context-dependent cell-cell interactions

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Using Decision Trees to Predict the Clinical Isolation Source of Haemophilus influenzae Based on Pan-Genomic Diversity

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Integrating common and rare variants into a genetic risk score for Alzheimer's disease risk prediction

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Intuitive intracellular communication deconvolution and ranking for improved discovery of context-dependent signaling via TrokaChat

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Gene Signature Reveals Decreased SOX10 Activity in Malignant Cells from Immunotherapy-Resistant Cutaneous Melanoma Tumors

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Gene-environment interaction of coffee with body mass index in multiple populations

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Quantification of 3′ UTR isoform expression from scRNA-seq reveals substantial changes in differentiation

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What Can Bioinformatics Scientists Learn from Librarians? Using Library and Archival Methodologies to Preserve and Reuse Omics Data

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Understanding Interferon-γ-Induced Esophageal Epithelial Barrier Dysfunction and Apoptosis using Integrative Analysis of In Vivo and Model Gene Expression

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Identification of genetic loci associated with the risk of aneuploidy with maternal origin using PGT-A sequences

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Evaluating the frequency and the impact of pharmacogenetic variants in an ancestrally diverse Biobank population

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Susceptibility of human glycogenes to functional variations

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ADE: an integrated bioinformatics web server for neurodegenerative disease exploration, omics data analysis, and drug discovery

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Petagraph: A biomedical knowledge graph built into the UMLS

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Improving Genomic Data Diversity Using Few-shot Generative Domain Adaptation

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Leveraging Graph Neural Networks for Interpretable Prediction of Pathological Stages in Prostate Cancer

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Cardiovascular Events Associated with PCOS Diagnosis in Large Longitudinal Cohort

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Identifying longitudinal disease trajectories and their clinical associations in 146,000 individuals with hypertension from Penn Medicine Electronic Health Records

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MultiNEP: disease-specific Multi-omics Network Enhancement for Prioritizing disease genes and metabolites

Zhuoran Xu, Brian Lee, Luigi Marchionni, Shuang Wang

Benchmark study of similarity measures from query phenotypic abnormalities to diseases based on the human phenotype ontology

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Prioritizing de novo variants using phenotype selection and an annotated data knowledge graph

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Understanding common and distinct information in paired multiomic data with Tilted-CCA

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Multi-modal single cell analysis of Multiple myeloma patient bone marrow over time reveal mechanisms

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Sensitive and Efficient Pangenome Construction through Alignment-Free Residue Pangenome Analysis (ARPA)

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A unified framework for realistic in silico data generation and statistical model inference in single-cell and spatial omics

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Structuring information via an immune-focused ontology enables the construction of a high-quality knowledge graph for the study of autoimmune diseases

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