8TH 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: Mona Singh, PhD** Professor, Computer Science Lewis Sigler Institute for Integrative Genomics Princeton University
9:30 AM – 10:00 AM	Jian Zhou, PhD Assistant Professor, Bioinformatics Department of Biostatistics University of Texas Southwestern
10:00 AM – 10:15 AM	Morning Break
10:15 AM – 10:45 AM	Mingyao Li, PhD Professor, Biostatistics Chair, Graduate Program in Biostatistics Member, Graduate Group in Genomics and Computational Biology Dept. of Biostatistics, Epidemiology and Informatics University of Pennsyvania
10:45 AM – 11:15 AM	Obi Griffith, PhD Associate Professor Department of Medicine Department of Genetics McDonnell Genome Institute Washington University St. Louis

AGENDA CONT'D

11:15 – 11:45 AM	*Poster Lightning Round* *(3-minute Flash Presentations)* *1. Yonghyun Nam, University of Pennsylvania* *2. Carlos Castelan Angel, Columbia University* *3. Kyle Coleman, University of Pennsylvania* *4. Krishnamurthy Subramanian , Rutgers University* *5. Sora Yoon, University of Pennsylvania* *6. Da Wu,Children's Hospital of Philadelphia* *7. Annie Abioye, University of Pennsylvania*
11:45 AM – 2:00 PM	*Break for Lunch* *Poster Networking Session* *Colket Translational Research Building*
2:00 – 2:45 PM	**Keynote Presentation #2: Mark Gerstein, PhD** Professor Biomedical Informatics, Molecular Biophysics & Biochemistry, Computer Science, Statistics & Data Science Yale University
2:45 - 3:15 PM	Beth Cimini, PhD Senior Group Leader Cimini Lab Broad Institute Harvard University Massachusetts Institute of Technology
3:15 PM – 3:30 PM	Afternoon Break
3:30 PM - 4:00 PM	Li Shen, PhD Professor Informatics University of Pennsylvania Department of biostatistics, Epidemiology, and Informatics Perelman School of Medicine University of Pennsylvania

4:00 PM - 4:45 PM

Keynote Presentation: David Baker, PhD
Professor
Biochemistry
Director, Institute for Protein Design
University of Washington

4:45 PM - 5:00 PM

Closing Remarks and Presentation of Trainee Awards
Bo Li, PhD
Associate Professor
Computational Immunology
Perelman School of Medicine
University of Pennsylvania
Children's Hospital of Philadelphia

THANK YOU CONFERENCE OPERATIONS

Donna Vito, Outreach Manager

Ariana Downs, MS, Outreach Specialist

Hannah Lazarus, Research Coordinator

THANK YOU POSTER REVIEW COMMITTEE

Sharon Diskin, PhD, Children's Hospital of Philadelphia

Joseph Daniel Romano, PhD, Mphil, MA, University of Pennsylvania

Mark Porter, Children's Hospital of Philadelphia

Michael Xie, PhD, Children's Hospital of Philadelphia

Iain Matheson, PhD, University of Pennsylvania

Noor Dawany, PhD, Children's Hospital of Philadelphia

Asif Chinwalla, MS, MBA, Children's Hospital of Philadelphia

Ana Cristancho, MD, PhD, Children's Hospital of Philadelphia

Ramakrishnan Rajagopolan, Children's Hospital of Philadelphia

KEYNOTE SPEAKERS

Mona Singh, PhD
Professor
Computer Science
Lewis Sigler Institute for
Integrative Genomics

Mona Singh is a professor of computer science and the Lewis Sigler Institute for Integrative Genomics. She has been on the faculty at Princeton University since 1999. She received her A.B. and S.M degrees from Harvard University, and her Ph.D. from MIT, all three in computer science. She works broadly in computational molecular biology, as well as its interface with machine learning and algorithms. Much of her work is on developing algorithms to decode genomes at the level of proteins and she is especially interested in developing data-driven methods for predicting and characterizing protein sequences, functions, interactions and networks, both in healthy and disease contexts. Among her awards are the Presidential Early Career Award for Scientists and Engineers (PECASE) in 2001, and the Rheinstein Junior Faculty Award from Princeton's School of Engineering and Applied Science in 2003. She was named a Fellow of the ACM in 2019, and of the ISCB in 2018. (Source: Princeton University)

Mark Gerstein, PhD
Albert L Williams Professor of Biomedical Informatics
Professor of Molecular Biophysics & Biochemistry of Computer Science, and of Statistics & Data Science
Yale University

After graduating from Harvard with an A.B. in physics in 1989, Prof. Mark Gerstein earned a doctorate in theoretical chemistry and biophysics from Cambridge University in 1993. He did postdoctoral research in bioinformatics at Stanford University from 1993 to 1996. He came to Yale in 1997 as an assistant professor and in 2003 became co-director of the Yale Computational Biology and Bioinformatics Program. Gerstein has published appreciably in the scientific literature, with an H index of ~185 and >600 publications in total, including a number of them in prominent venues, such as Science, Nature, Cell, and Scientific American. His research is focused on biomedical data science, and he is particularly interested in machine learning, macromolecular simulation, human genome annotation & disease genomics, and genomic privacy. (Source: Yale University)

David Baker, PhD
Henrietta and Aubrey Davis Endowed Professor in Biochemistry
Director, Institute for Protein Design
University of Washington

Dr. Baker's research group is focused on the design of macromolecular structures and functions.

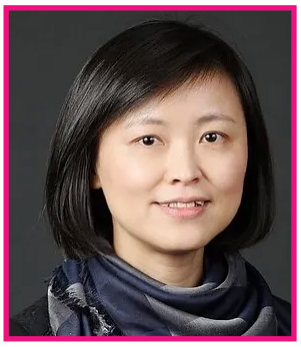
He received his Ph.D. in biochemistry with Randy Schekman at the University of California, Berkeley, and did postdoctoral work in biophysics with David Agard at UCSF. Dr. Baker has published over 550 research papers, been granted over 100 patents, and co-founded 17 companies. Over 70 of his mentees have gone on to independent faculty positions. Dr. Baker is a member of the National Academy of Sciences and the American Academy of Arts and Sciences. He is also a project leader with The Audacious Project.

Dr. Baker has received awards from the National Science Foundation, the Beckman Foundation, and the Packard Foundation. He is the recipient of the Breakthrough Prize in Life Sciences, The Wiley Prize in Biomedical Sciences, Irving Sigal and Hans Neurath awards from the Protein Society, the Overton Prize from the ISCB, the Feynman Prize from the Foresight Institute, the AAAS Newcomb Cleveland Prize, the Sackler Prize in Biophysics, and the Centenary Award from the Biochemical Society. (Source: University of Washington)

SPEAKERS

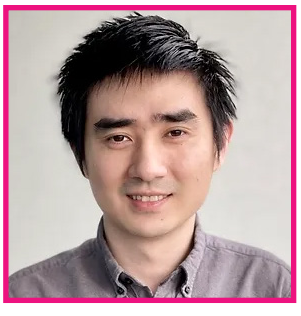
Obi Griffith, PhD
Associate Professor
Department of Medicine
Department of Genetics
McDonnell Genome Institute
Washington University St. Louis

Dr. Griffith is Associate Professor of Medicine (Oncology) and Genetics and Assistant Director of the McDonnell Genome Institute at Washington University. Dr. Griffith's research is focused on the development of personalized medicine strategies for cancer. He develops bioinformatics tools and methods for the analysis of high throughput sequence data and identification of biomarkers for diagnostic, prognostic and drug response prediction. Dr. Griffith is President of the Cancer Genomics Committee, on the Steering Committee for the Global Alliance for Genomic Health, and co-chair of the Variant Interpretation for Cancer Consortium. He has published over 100 studies, received numerous research awards and honors and held several large grants from the NIH including a K22 Transition Career Development Award, U01/U24 for Development of Informatics Technologies for Cancer Research, and others.
He has mentored more than 50 bioinformatics trainees and taught hundreds more as an Instructor for Cold Spring Harbor Laboratories and the Canadian Bioinformatics Workshops.
(Source: Washington University St. Louis)



Mingyao Lee, PhD
Professor of Biostatistics
Chair, Graduate Program in Biostatistics
Member, Graduate Group in Genomics and Computational Biology

Dr. Mingyao Li joined the Biostatistics faculty in 2006. She is also a faculty member of the Genomics and Computational Biology graduate program. Her main research area is statistical genetics and genomics. The central theme of her current research is to use statistical and machine learning approaches to understand cellular heterogeneity in human disease relevant tissues, to characterize gene expression diversity across cell types, and to study the patterns of cell state transition and crosstalk of various cells using data generated from single-cell transcriptomics studies. In addition to methods development, Dr. Li is also interested in collaborating with researchers seeking to identify complex disease susceptibility genes. Her collaborative research includes cardiometabolic diseases, age-related macular degeneration, Alzheimer's disease, chronic kidney disease, type 1 diabetes, and cancer. Findings from her research will seed cell-specific functional studies, in vivo modeling, and precision therapeutic targeting of human diseases. Dr. Li actively serves in the scientific community. She is an Associate Editor of Statistics in Biosciences, and was a regular member of the Genomics, Computational Biology and Technology study section and a member of the review committee of the Center for Inherited Disease Research of the NIH. (Source; University of Pennsylvania)



Jian Zhou, PhD
Assistant Professor
Bioinformatics
Lupe Murchison Foundation Scholar in Medical Research

Since November 2019, Jian Zhou has been appointed as Assistant Professor in the Lyda Hill Department of Bioinformatics. He is a Lupe Murchison Foundation Scholar in Medical Research and is a Scholar of the Cancer Prevention and Research Institute of Texas (CPRIT). Prior to joining UTSW, he was a Flatiron Research Fellow at the Center of Computational Biology at Flatiron Institute, New York. He received his B.S. in Biological Sciences from Peking University and Ph.D. in Quantitative and Computational Biology from Princeton University.

The Zhou lab works at the intersection of machine learning and genomics. The lab develops computational methods to improve our understanding of genome-based regulation and the genetic basis of human health and diseases. Advancing machine learning and AI methods for science, especially genomics is a long-term goal of the lab. Visit the lab website for more details https://zhoulab.io. (Source: UT Southwestern)

Beth Cimini, PhD
Professor
Department of Biostatistics
Columbia University

Beth Cimini leads the Cimini Lab within the Imaging Platform of the Broad Institute. Her team works with biologists to help them create image analysis workflows and makes the open-source image analysis software CellProfiler. In 2020, she was named a CZI Imaging Scientist for her work on making open-source image analysis tools more accessible to the bioimaging community and for her creation of a postdoctoral training program in bioimage analysis.

Cimini's lab started at the Broad in 2021, after 5 years as a postdoc then computational biologist in Anne Carpenter's lab at the Broad Institute. She holds a PhD in biochemistry and molecular biology from the Blackburn Lab at the University of California-San Francisco and a BA in biochemistry and molecular biology from Boston University.

Dr. Cimini's laboratory is best known for our popular open source software CellProfiler and CellProfiler Analyst. (Source: Broad Institute)

Li Shen PhD
Professor
Director of Folding@home
Department of Bioengineering and Biochemistry
University of Pennsylvania

Dr. Li Shen is a Professor of Informatics in the Department of Biostatistics, Epidemiology and Informatics at the Perelman School of Medicine in the University of Pennsylvania. He also holds a secondary appointment in the Department of Radiology. He is a Senior Fellow at the Penn Institute for Biomedical Informatics and the Leonard Davis Institute of Health Economics. He obtained his Ph.D. degree in Computer Science from Dartmouth College.

Dr. Shen's research interests include medical image computing, biomedical informatics, machine learning, network science, imaging genomics, multi-omics and systems biology, Alzheimer's disease, and big data science in biomedicine. He has authored over 300 peer-reviewed articles in these fields. His work has been continuously supported by the NIH and NSF. His current research program is focused on developing and applying informatics, computing and data science methods for discovering actionable knowledge from complex biomedical and health data (e.g., genetics, omics, imaging, biomarker, outcome, EHR, health care), with applications to complex disorders such as Alzheimer's disease.

Dr. Shen has served on a variety of scientific journal editorial boards, grant review committees, and organizing committees of professional meetings in medical image computing and biomedical informatics. He served as the Executive Director of the Medical Image Computing and Computer Assisted Intervention (MICCAI) Society between 2016 and 2019. He is a fellow of the American Institute for Medical and Biological Engineering (AIMBE), a distinguished member of the Association for Computing Machinery (ACM), and a distinguished contributor of the IEEE Computer Society. (Source: University of Pennsylvania

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

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

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

Shubha Vasisht (1), Yuanchao Zhang (1), Deanne Taylor (1,2) 1. Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia 2. Perelman School of Medicine, University of Pennsylvania

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

Kaishu Mason, Nancy Zhang, University of Pennsylvania, Professor of Statistics Anuja Sathe, Stanford University, Postdoctoral Fellow

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

Ene Belleh, Children's Hospital of Philadelphia; Jennae Luecke, Children's Hospital of Philadelphia; Allison Olsen, Children's Hospital of Philadelphia; Julianna Pakstis, Children's Hospital of Philadelphia

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

Megha Lal, PhD1, Ravi Gautam, PhD1, Zoe Mrozek, BS1, Yusen Zhou, PhD3, Jarad Beers, MS1, Margaret C. Carroll, MS1, Melanie A. Ruffner, MD, PhD1,2. 1Division of Allergy and Immunology, Children's Hospital of Philadelphia 2Department of Pediatrics, Perelman School of Medicine at University of Pennsylvania 3Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia

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

Ben Stear (1), MS; Taha Mohseni Ahooyi (1), PhD; Shubha Vasisht (1); Jonathan Silverstein (3,4), MD, MS, FACS, FACMI; Tiffany Callahan (5), PhD; Deanne Taylor (1,2), PhD. 1. Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia 2. Perelman School of Medicine, University of Pennsylvania 3. Health Sciences and Institute for Precision Medicine, University of Pittsburgh 4. Department of Biomedical Informatics, University of Pittsburgh 5. Anschutz Medical Campus, University of Colorado Denver

Improving Genomic Data Diversity Using Few-shot Generative Domain Adaptation

Chen Song, Computer and Information Science Department, Temple University Emily Thyrum, Computer and Information Science Department, Temple University Dr. Xinghua Shi, Computer and Information Science Department, Temple University

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

Wenkang Zhan, Department of Computer & Information Science, Temple University Chen Song, Department of Computer & Information Science, Temple University Xinghua Shi, PhD, Department of Computer & Information Science, Temple University

Cardiovascular Events Associated with PCOS Diagnosis in Large Longitudinal Cohort

Cherlin T , PhD (1), Morse C, PT, DPT, MSA (2), Lee ITL , MD (3), Dokras A, MD, PhD (3), Verma SS, PhD (1) 1. Dept. of Pathology and Laboratory Medicine, University of Pennsylvania, Philadelphia, PA 2. Penn Medicine Biobank, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA 3. Perelman School of Medicine, Division of Reproductive Endocrinology and Infertility, University of Pennsylvania, Philadelphia, PA

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

Pankhuri Singhal1 (BS), Lindsay Guare1 (BS), Anastasia Lucas1,2 (BS), Colleen Morse1 (DPT), Marta Byrska-Bishop3 (PhD), Marie A. Guerraty1 (MD), Dokyoon Kim1 (PhD), Anurag Verma1,2 (PhD) , and Marylyn D. Ritchie1 (PhD), 1 University of Pennsylvania Perelman School of Medicine, Philadelphia, PA 2Corporal Michael Crescenz VA Medical Center, Philadelphia, PA 3New York Genome Center, New York, NY

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

Yu Hu, PhD 1, Joe Chan, MS 1, Kai Wang, PhD 1,2 1: Raymond G. Perelman Center for Cellular and Molecular Therapeutics, Children's Hospital of Philadelphia, Philadelphia, PA 19104, USA 2: Department of Pathology and Laboratory Medicine, University of Pennsylvania, Philadelphia, PA 19104, USA

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

Erin Nesmith(1), Michelle Gong(1), Deanne Taylor (1,2) DBHI, CHOP Dept Pediatrics, UPenn Perelman School of Medicine

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

Dr. Kevin Lin*, Dr. Nancy R. Zhang* *Wharton Statistics and Data Science, University of Pennsylvania

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

Wesley V Wilson (1), Fei Miao, Alfred L Garfall (2), Adam D Cohen(2), Michael C.Milone (1) 1 - Center for Cellular Immunotherapies, Perelman School of Medicine At the Univ. of Pennsylvania, Philadelphia, PA 2 - Abramson Cancer Center, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA

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

Arnav Lal 1, Andries Feder 2, Ahmed Moustafa 3,4, and Paul J. Planet 2,3,5 1 School of Arts and Sciences, University of Pennsylvania, Philadelphia, PA 19104, USA 2 Division of Pediatric Infectious Diseases, Children's Hospital of Philadelphia, Philadelphia, PA 19104, USA 3 Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA 19104, USA 4 Division of Gastroenterology, Hepatology & Nutrition, Children's Hospital of Philadelphia, Philadelphia, PA 19104, USA 5 Institute for Comparative Genomics, American Museum of Natural History, New York, NY 10024, USA

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

Dongyuan Song, PhD candidate in Bioinformatics, UCLA Qingyang Wang, PhD student in Statistics, UCLA Guanao Yan, PhD candidate in Statistics, UCLA Tianyang Liu, Master of Applied Statistics, UCLA Jingyi Jessica Li, Associate Professor in the Department of Statistics, Department of Human Genetics, Department of Computational Medicine and Department of Biostatistics, UCLA

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

Van Q. Truong1-5, Joseph D. Romano2, Allison R. Greenplate4-6, Scott M. Dudek2,3, E. John Wherry4-6, Marylyn D. Ritchie2,3,7 1 Graduate Group in Genomics & Computational Biology, Perelman School of Medicine, University of Pennsylvania 2 Institute for Biomedical Informatics, Perelman School of Medicine, University of Pennsylvania 3 Biomedical and Translational Informatics Laboratory, Perelman School of Medicine, University of Pennsylvania 4 Immune Health Project, Perelman School of Medicine, University of Pennsylvania 5 Institute for Immunology, Perelman School of Medicine, University of Pennsylvania 6 Department of Pharmacology & Translational Therapeutics, Perelman School of Medicine, University of Pennsylvania 7 Department of Genetics, Perelman School of Medicine, University of Pennsylvania

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