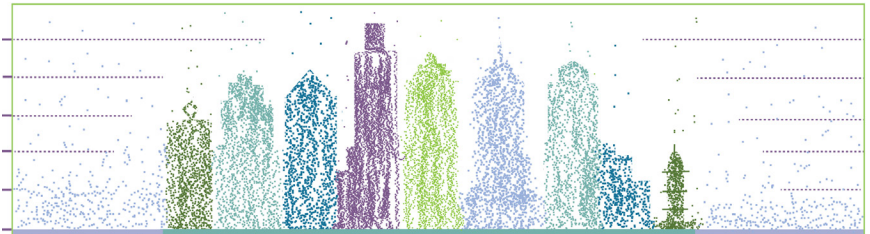

Third Annual **MidAtlantic Bioinformatics Conference** October 29, 2018

Challenges in Genetic Analysis of Complex Diseases

October 29, 2018
The Children's Hospital of Philadelphia
Colket Translational Research Building

Children's Hospital of Philadelphia℠
Department of Biomedical & Health Informatics

Institute for Biomedical Informatics

Perelman School of Medicine
University of Pennsylvania

SCHEDULE

7:30 AM — REGISTRATION OPENS

8:15 AM — WELCOME

Bryan Wolf, MD, PhD
Chief Scientific Officer
Chair, Department of Biomedical and Health Informatics
Children's Hospital of Philadelphia

8:30 AM — KEYNOTE: THE PRESENT AND THE FUTURE OF GENOMIC MEDICINE.

Wendy Chung MD, PhD
Associate Professor, Pediatrics
Columbia University

9:30 AM — DETECTION OF STRUCTURAL VARIANTS IN HUMAN GENOME BY LONG-READ SEQUENCING

Chia-Lin Wei, PhD
Director, Genome Technologies

10:00 AM — META-DIMENSIONAL ANALYSIS OF MULTI-OMICS DATA

Marylyn Ritchie, PhD
Director, Center for Translational Bioinformatics
Associate Director for Bioinformatics,
Institute for Biomedical Informatics (IBI)
Associate Director, Center for Precision Medicine
University of Pennsylvania

10:30 AM — MORNING BREAK

10:45 AM — PRESENTATION TITLE

Michael Talkowski, PhD
Associate Professor of Neurology (Genetics), Psychiatry, and Pathology. Harvard Medical School

11:15 AM — GABRIELLA MILLER KIDS FIRST DATA RESOURCE CENTER: ADVANCING GENETIC ANALYSES OF STRUCTURAL BIRTH DEFECTS AND CHILDHOOD CANCER

Deanne Taylor, PhD
Director, Bioinformatics,
Department of Biomedical & Health Informatics
Research Professor, Perelman School of Medicine

SCHEDULE

11:45 AM — LUNCH & POSTER SESSION
Poster session takes place in Colket Atrium

1:45 PM — KEYNOTE INTRODUCTION
Jason Moore, PhD.
Director, Institute for Biomedical Informatics
University of Pennsylvania

KEYNOTE: HUGE COHORTS, GENOMICS, AND CLINICAL DATA TO PERSONALIZE MEDICINE
Joshua Denny, M.D., M.S., FACMI
Professor of Biomedical Informatics, Professor of Medicine,
Director, Center for Precision Medicine
Vanderbilt University School of Medicine

2:45 PM — STUDYING NUCLEOSOME POSITIONING AND CHROMATIN ACCESSIBILITY AT SINGLE DNA MOLECULES BY NANOPORE SEQUENCING
Kin Fai Au, PhD
Associate Professor
Department of Internal Medicine/Department of Biostatistics
Ohio State University

3:15 PM — AFTERNOON BREAK

3:30 PM — IDENTIFICATION OF RELEVANT GENETIC ALTERATIONS IN CANCER USING TOPOLOGICAL DATA ANALYSIS
Pablo Gonzalez-Camara, PhD
Assistant Professor, Genetics
Perelman School of Medicine, University of Pennsylvania

4:00 PM — EPIGENOMIC STRATEGIES AT THE INTERFACE OF GENETICS AND PERINATAL EXPOSURES IN AUTISM
Janine Lasalle, PhD
Professor; Associate Director of Genomics Department of Microbiology and Immunology; M.I.N.D. Institute
University of California at Davis Genome Center

4:30 PM — CLOSING

KEYNOTE SPEAKERS

Wendy Chung, MD, PhD

Kennedy Family Professor of Pediatrics and Medicine
Director of Clinical Research of SFARI at the Simons Foundation
Columbia University

A world leader in the diagnosis and treatment of rare diseases, Dr. Wendy Chung has identified more than 41 new genes for human diseases. She is experienced in both the molecular and clinical characterization of the disorders associated with these gene mutations, as well as the integration of these discoveries into clinical practice. Dr. Chung directs NIH funded research programs in human genetics of autism, neurodevelopmental disorders, birth defects including congenital diaphragmatic hernia, esophageal atresia, and congenital heart disease, pulmonary hypertension, cardiomyopathy, and breast cancer. She leads the Precision Medicine Resource in the Irving Institute at Columbia University and serves on the Genomics and Pediatrics Advisory Committees for the All of Us Precision Medicine Initiative and Council for the National Human Genome Research Institute.

Joshua Denny, MD, MS, FACMI

Professor of Biomedical Informatics and Medicine
Director of the Center for Precision Medicine
Vice President of Personalized Medicine
Vanderbilt University Medical Center

Dr. Denny 's research interests include natural language processing, accurate phenotype identification from electronic medical record data, and using the electronic medical record to discover genome-phenome associations to better understand disease and drug response, including the development of the EMR-based phenome-wide association (PheWAS). At Vanderbilt, he is part of the PREDICT (Pharmacogenomic Resource for Enhanced Decisions in Care and Treatment) program, which prospectively genotypes patients to tailor drugresponse. He is PI of the Data and Research Center of the "All of Us" Research Program, which will eventually enroll at least 1 million Americans in an effort to understand the genetic, environmental, and behavioral factorsthat influence human health and disease. He is also PI for Vanderbilt sites in the ElectronicMedical Records and Genomics (eMERGE) Network, Pharmacogenomics Research Network(PGRN), and the Implementing Genomics Into Practice (IGNITE) Network.

SPEAKERS

Kin Fai Au, PhD
Associate Professor
Department of Biomedical Informatics
Ohio State University

Dr. Kin Fai Au is an associate professor of Biomedical Informatics at The Ohio State University. Dr. Au is interested in developing novel statistical and computational/algorithmic methods for analyzing high-throughput biological data, including but not limited to transcriptome and epigenetics sequencing data. For example, Dr. Au's group has developed a set of high-impact statistical and computational methods for analyzing Third Generation Sequencing data (e.g., PacBio and Oxford Nanopore sequencing). Au lab is also interested in stem cell biology, developmental biology and cancer research. We focus on making meaningful advances to understand stem cell biology , developmental biology and cancer, using cutting edge technologies (such as PacBio and Oxford Nanopore Technologies) to create authentic models and gain novel biological insights in transcriptome and epigenetics level.

Pablo Gonzalez-Camara, PhD
Assistant Professor, Genetics
Perelman School of Medicine, University of Pennsylvania

After receiving his PhD in theoretical physics in 2006 from Universidad Autónoma de Madrid, Dr. Cámara performed research in string theory at Ecole Polytechnique, the European Organization for Nuclear Research (CERN), and University of Barcelona. He shifted his research focus into problems in quantitative biology in 2014. Dr. Cámara has devised novel approaches to the inference of ancestral recombination, human recombination mapping, the study of cancer heterogeneity, and the analysis of single-cell RNA-sequencing data from dynamic and heterogeneous cellular populations. His current research focus is on the development and application of computational approaches to the study of cellular heterogeneity and its role in disease using high-throughput single-cell technologies.

Janine Lasalle, PhD
Professor; Associate Director of Genomics Department of Microbiology and Immunology; M.I.N.D. Institute
UC Davis Genome Center

Dr. LaSalle's research is on epigenetics of neurodevelopmental disorders. Her laboratory uses genomic and epigenomic technologies to investigate the role of DNA methylation and MeCP2 in the pathogenesis of Rett syndrome and autism spectrum disorders. Dr. LaSalle's lab also takes integrative genetic and epigenomic approaches to investigate the role of persistent organic pollutants and other environmental factors on the placental and developing brain methylome. Dr. LaSalle recently served as chair of the Genes in Health and Disease study section for the NIH. She also serves on the editorial board of the journals "Human Molecular Genetics", "Molecular Autism", and "Environmental Epigenetics" and is on the Scientific Advisory boards of the International Rett Syndrome Foundation and the Dup15q Alliance.

SPEAKERS

Marylyn Ritchie, PhD

Director, Center for Translational Bioinformatics,
Associate Director for Bioinformatics,
Institute for Biomedical Informatics (IBI)
Associate Director, Center for Precision Medicine
University of Pennsylvania

Dr. Marylyn Ritchie is a statistical and computational geneticist with a focus on understanding genetic architecture of complex human disease. She has expertise in developing novel bioinformatics tools for complex analysis of big data in genetics, genomics, and clinical databases, in particular in the area of pharmacogenomics. Some of her methods include Multifactor Dimensionality Reduction (MDR), the Analysis Tool for Heritable and Environmental Network Associations (ATHENA), and the Biosoftware suite for annotating/filtering variants and genomic regions as well as building models of biological relevance for gene-gene interactions and rare-variant burden/dispersion tests. Dr. Ritchie has over 15 years of experience in the analysis of complex data, has authored over 250 publications and is the recipients of several national awards and honors for her distinguished work including being named one of the most highly cited researchers in her field by Thomas Reuters (in 2014).

Michael Talkowski, PhD

Harvard Medical School
Desmond and Ann Heathwood Associate Professor
Center for Genomic Medicine
Departments of Neurology, Psychiatry, and Pathology
Massachusetts General Hospital, Harvard Medical School, and Broad

Dr. Talkowski's research focuses on understanding the genetic etiology of complex neurodevelopmental and neuropsychiatric disorders. His lab integrates molecular and computational approaches to explore the functional consequences of genomic variation, with a particular interest in the relationship between genome structure and function studying disorders affecting prenatal, neonatal, and early childhood development, particularly autism spectrum disorder (ASD) and related neurodevelopmental and psychiatric disorders. Dr. Talkowski received his Ph.D. in human genetics and trained in genomics as a postdoctoral fellow at Harvard Medical School, Massachusetts General Hospital (MGH) and the Broad Institute.

SPEAKERS

Deanne Taylor, PhD

Director, Bioinformatics
Department of Biomedical and Health Informatics
Research Professor
Perelman School of Medicine, University of Pennsylvania

Dr. Taylor's research focuses on the development of mathematical and computational methods to better understand biological variation and the genetic contribution to disease, coupling clinical information with high-dimensional biomedical data from next-gen sequencing, microarray, PCR, and proteomics experiments. She is currently diretcor of bioinformatics with the Department of Biomedical and Health Informatics at Children's Hospital of Philadelphia. Previously, she was Director of Bioinformatics with Reproductive Medicine Associates and Assistant Professor with the Department of Obstetrics, Gynecology and Reproductive Medicine at Rutgers Robert Wood Johnson Medical School. Prior to that she worked for Harvard School of Public Health as a Research Scientist and served several years as the Program Director of the Graduate Program in Bioinformatics at Brandeis University. Her background is in biophysics, bioinformatics, computational biology and structural biology with emphasis on human genetics and translational medicine.

Chia-Lin Wei, PhD

Director, Genome Technologies
The Jackson Laboratory

Dr. Wei joined the Jackson Laboratory in 2016 as the Director of Genomic Technologies, where she actively engages the development and application of new sequencing based genomic technologies to explore genome structure and function. Dr. Wei gother Ph.D. (Microbiology) from University of California at Davis. Prior to that she was at the Joint Genome Institute (JGI), joining in 2010. She was responsible for Department of Energy (DOE) user facility genomic sequencing operation as well as sequencing related technology development e ort within the Genomic Technology Department. Dr. Wei moved to JGI after an eight-year stint at Genome Institute of Singapore (GIS) and was responsible for establishing a sequencing technology platform and genome biology program.

POSTERS

Risk Variants Disrupting Enhancers of TH1 and TREG Cells in Type 1 Diabetes

Yasin Uzun, PhD, Peng Gao, PhD, Bing He, PhD, Sarah E. Salamati, Julie K.M. Coffey, Eva Tsalikian, Kai Tan, PhD - Children's Hospital of Philadelphia

Identifying Noncoding Risk Variants Using Disease-relevant Gene Regulatory Networks/Role of Structural Variants and Non-coding Variantss

Long Gao PhD[2], Yasin, Uzun PhD[1], Peng Gao, PhD[1], Bing He PhD[1], Xiaoke Ma, PhD[3], Xidian Jiahui Wang, MS[3], ShiZhong Han PhD[4], Kai Tan PhD[1] 1)Children's Hospital of Philadelphia; 2)University of Pennsylvania; 3)The Jackson Lab; 4)Johns Hopkins University

CRISPRKat - A Tool to Summarize and Prioritize Results from Multiple Analyses of CRISPR-Cas9 Based Functional Genomics Screens

Katharina E. Hayer[1,2], Abby M. Green[3,4,5], Matthew D. Weitzman1[4,5] ,Deanne M. Taylor [2,3,5] 1)Departmen of Pathology and Laboratory Medicine, Division of Cancer Pathobiology, Children's Hospital of Philadelphia; 2) Department of Biomedical and Health Informatics Children's Hospital of Philadelphia; 3) Department of Pediatrics, Perelman School of Medicine,University of Pennsylvania; 4) Center for Childhood Cancer Research, Children's Hospital of Philadelphia; 5) Perelman School of Medicine, University of Pennsylvania

Optical Mapping Reveals Structures Involved in Non-Allelic Homologous Recombination in 22q11 Deletion Syndrome

Steven Pastor[1], Andrea Jin[1], Oanh Tranh[1], Ming Xiao[2], Beverly S. Emanuel[1,3] 1)The Children's Hospital of Philadelphia; 2)Drexel University; 3)Perelman School of Medicine University of Pennsylvania

Investigating the Genetic Architecture of Psychiatric Disorders and their Medical Comorbidity from a Developmental Perspective

Alison K. Merikangas MPH, PhD[1], Rachel L. Kember MSc, PhD[2,3], Kosha Ruparel MSE[4], Monica E. Calkins PhD[4], Ruben C. Gur PhD[4], Raquel E. Gur MD, PhD[4], Laura Almasy PhD[1,2] 1)Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia; 2) Department of Genetics, Perelman School of Medicine, University of Pennsylvania; 3)Crescenz VA Medical Center; 4) Department of Psychiatry, Neuropsychiatry Section, Perelman School of Medicine, University of Pennsylvania

Scedar: A scalable Python Package for Single-cell RNA-seq Exploratory Data Analysis

Yuanchao Zhang, PhD[1,2] , Deanne Taylor[1,3], 1)Department of Genetics, Rutgers University; 2)Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia; 3) Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania

Dissecting Autism Genetic Risk Using Single-cell Transcriptomics Data

Siying Chen, PhD candidate, Yufeng Shen, PhD, Department of System Biology, Columbia University

Statistical Inference Relief (STIR) Feature Selection

Trang T. Le[1], Ryan J. Urbanowicz[1], Jason H. Moore[1], Brett A. McKinney[2,3] 1)Institute for Biomedical Informatics, University of Pennsylvania; 2)Department of Mathematics, University of Tulsa; 3)Tandy School of Computer Science, University of Tulsa

Ribosomal DNA Variation in Human Genomes

Lingyu Guan, Spyros Karaiskos, Andrey Grigoriev, PhD, Rutgers University

POSTERS

Correlation of the Metabolome and Microbiome of VEO-IBD Patients

Tanaya Jadhav[1,2], Maire Conrad, MD[1], Kyle Bittinger, PhD[1], Yue Ren[1], Frederic Bushman, PhD[1], Marcella Devoto, PhD[1], Robert Baldassano, MD[1], Judith Kelsen, MD[1], Noor Dawany, PhD[1]
1)Children's Hospital of Philadelphia; 2)Temple Univeristy

Pan-cancer Analysis of Causal Noncoding Mutations in Pediatric Cancers

Bing He, PhD, Peng Gao, PhD, Chia-Hui Chen, MS, Yang-Yang Ding, MD, Hannah Kim, MS, Sarah K. Tasian, MD, Stephen P. Hunger, MD, Kai Tan, PhD - Children's Hospital of Philadelphia

Inferring Enhancer and Noncoding RNA Dysregulation Underlying 2,419 UK Biobank Phenotypes

A. Amlie-Wolf[1,2], L. Qu[2], E.E. Mlynarski[2], P.P. Kuksa[2], Y.Y. Leung[2], C.D. Brown[2,3], G.D. Schellenberg[2,3], L.S. Wang[2,3] 1) Penn Genomics and Computational Biology Graduate Group; 2) Penn Neurodegeneration Genomics Center, Department of Pathology and Laboratory Medicine; 3) Department of Genetics - Perelman School of Medicine, University of Pennsylvania

Defining the Impact of DNA Accessibility on CRISPR-Cas9 Cleavage Efficiency

Cheng-Han Chung[1,2], Alex Allen[1,2], Neil Sullivan[1,2], Andrew Atkins[1,2], Michael R. Nonnemacher[1,2,3], Brian Wigdahl[1,2,3], and Will Dampier[1,2,4] 1)Department of Microbiology and Immunology, Drexel University College of Medicine; 2)Center for Molecular Virology and Translational Neuroscience, Institute for Molecular Medicine and Infectious Disease, Drexel University College of Medicine; 3)Sidney Kimmel Cancer Center, Thomas Jefferson University; 4)School of Biomedical Engineering, Science, and Health Systems, Drexel University

MAJIQ-HET Robustly Detects Changes in RNA Splicing Between Large Heterogeneous Sample Groups

Scott Norton[1,2], Jordi Vaquero-Garcia[2,3], Christopher Green[2], Yoseph Barash[2,3] 1)Biomedical Graduate Studies, Perelman School of Medicine, University of Pennsylvania; 2)Department of Genetics, Perelman School of Medicine, University of Pennsylvania; 3)Department of Computer and Information Sciences, School of Engineering, University of Pennsylvania

Differences Between AUDIT-C and AUD Phenotypes Revealed by Genome-wide

Rachel L. Kember, PhD[1,2], Hang Zhou, PhD[3,4], Amy C. Justice, MD, PhD[3,4,5], Rachel V. Smith, PhD[2], Janet P. Tate, ScD[4], William C. Becker, MD[3,4], John Concato, MD[3,4], David A. Fiellin, MD[3,5], Ke Xu, MD[3,4], Hongyu Zhao, PhD[3,5], Joel Gelernter, MD[3,4], and Henry R. Kranzler, MD[1,2] 1)University of Pennsylvania Perelman School of Medicine; 2)VISN 4 MIRECC, Crescenz Veterans Administration Medical Center; 3)Yale School of Medicine; 4)Veterans Affairs Connecticut Healthcare System; 5)Yale School of Public Health

Integration of Genomic, Proteomic, and Electronic Health Record Data to Assess Polygenic Cardiovascular Risk

Yi-An Ko[1], Dawn Marchadier[1], Aditya Munshi[2], Rachel Kember[1], Renae L. Judy[3], Nick Hand[1], Scott M. Damrauer[3], Daniel J. Rader[1] 1)Institute for Translational Medicine and Therapeutics, University of Pennsylvania School of Medicine; 2)Department of Internal Medicine, Thomas Jefferson University Hospitals; 3) Department of Surgery, Perelman School of Medicine, University of Pennsylvania

Investigating Sequencing Platform Variant Call Biases

Brian Ennis BS, Children's Hospital of Philadelphia Perry Evans PhD, Zalman Vaksman PhD Children's Hospital of Philadelphia

POSTERS

Discovering and Visualizing Physical Activity Patterns and Disease Exacerbation Risks for COPD Patients in Real-time Digital Monitoring Data

Yuan An, PhD[1], Matthew Marschall[1], Siling Chen[1], Rachel Fisher[1], Parth Sharma[1]
Russell P. Bowler, MD, PhD[2], Matthew Allinder, MS[3], Nicholas Locantore, PhD[3]
1)CCI at Drexel University; 2)National Jewish Health; 3)GSK Research and Development

Phenome-wide Study Identifies Long-range LD Hotspots that Infer Epistasis in Alzheimer's Disease

Shefali S. Verma[1], Pankhuri Singhal[1], David Fasel[2], Sarah Pendergrass[3], Daniel Schaid[4], Iftikhar Kullo[4], Ozan Dikilitas[4], Chunhua Weng[2], Hakon Hakonarson[5], Patrick Sleiman[5], Murray Brilliant[6], Scott Hebbring[6], Steven Schrodi[6], Alex Frase[1], Scott Dudek[1], Anurag Verma[1], Marylyn D. Ritchie[1]
1)University of Pennsylvania; 2)Columbia University; 3)Geisinger Medical Center; 4)Mayo Clinic; 5)Children's Hospital of Pennsylvania; 6)Marshfield Clinic

Harnessing Longitudinal Data to Derive a New Genetic Risk Score for Childhood Obesity

Craig SJC, PhD[1,2], Kenney AM[3], Lin J PhD[3], Paul IM MD, MSc[2,4], Birch LL PhD[5], Savage JS PhD[6,7], Marini ME[7], Chiaromonte F PhD[2,3], Reimherr M PhD[2,3], Makova KD PhD[1,2] 1) Biology Department, Penn State; 2) Center for Medical Genomics, Penn State; 3) Department of Statistics, Penn State; 4) Department of Pediatrics, Penn State College of Medicine; 5) Department of Foods and Nutrition, University of Georgia; 6) Department of Nutrition Sciences, Penn State; 7) Center for Childhood Obesity Research, Penn State

SV-INFERNO: a Spark Based Pipeline for INFERring the Molecular Mechanisms of NOncoding Structural Variants

Elisabeth E. Mlynarski[1, 2, 3], Alexandre Amlie-Wolf[1, 2, 3, 4] , Pavel P. Kuksa[1, 2, 3], Otto Valladares[1, 2, 3], Gerard D. Schellenberg[1,3], Li-San Wang[1, 2, 3] 1)Penn Neurodegeneration Genomics Center; 2)Institute for Biomedical Informatics; 3)Department of Pathology and Laboratory Medicine; 4) Genomics & Computational Biology Graduate Group - Perelman School of Medicine University of Pennsylvania

Modeling the Spatiotemporal Dynamics of EGFR Activation in the Follicular Epithelium

Nicole T. Revaitis[1], Nastassia Pouradier Duteil, PhD[1], Matthew Niepielko, PhD[1], Benedetto Piccoli, PhD[1,2], Nir Yakoby, PhD[1,3] 1)Center for Computational and Integrative Biology; 2)Mathematics Department; 3)Biology Department - Rutgers University

Prediction of HIV-1 LTR Transcriptional Activity Using Deep Convolutional Neural Networks

Robert W. Link[1], Michael R. Nonnemacher[2,3,4], Brian Wigdahl[2,3,4], and Will Dampier[1,2,3]
1)School of Biomedical Engineering, Science and Health Systems, Drexel University; 2) Department of Microbiology and Immunology, Drexel University College of Medicine; 3)Center for Molecular Virology and Translational Neuroscience, Institute for Molecular Medicine and Infectious Disease, Drexel University College of Medicine; 4)Sidney Kimmel Cancer Center, Thomas Jefferson University

Visualizing the Determinants of Health

Edwin C., MA, Juhan S., BA - GoInvo

POSTERS

Utilizing Convolutional Neural Networks to Predict HIV-1 Tat Biological Function

Angela Tomita[1], Anthony R. Mele[2,3], Brian Wigdahl[2,3,4], Michael R. Nonnemacher[2,3,4], Will Dampier[1,2,3]
1)School of Biomedical Engineering, Science and Health Systems, Drexel University; 2)Department of Microbiology and Immunology, Drexel University College of Medicine; 3)Center for Molecular Virology and Translational Neuroscience, Institute for Molecular Medicine and Infectious Disease, Drexel University College of Medicine; 4)Sidney Kimmel Cancer Center, Thomas Jefferson University

Understanding the Role of Ciliary Genes in Gongenital Heart Disease Pathogenesis.

Kylia Williams[1], Abha Bais, PhD[1], Wenjuan Zhu[2], Madhavi Ganapthiraju, PhD[3],
Dennis Kostka, PhD[1], Dorothy Hammond, PhD[4], Michael Xie, PhD[4], Laura Mitchell, PhD[5],
Lisa Martin, PhD[6], Deanne Taylor, PhD[4], Elizabeth Goldmuntz, MD[7], Cecilia Lo, PhD[1]
1) University of Pittsburgh, Department of Developmental Biology; 2) The Chinese University of Honk Kong; 3) University of Pittsburgh, Department of Biomedical Informatics; 4) Children's Hospital of Philadelphia, Department of Biomedical and Health Informatics; 5) University of Texas Health Science Center, Division of Epidemiology, Human Genetics, and Environmental Science; 6) Cincinnati Children's Hospital Medical Center; 7) Children's Hospital of Philadelphia

Toward Clinical-grade In Silico Prediction: A disease-specific Machine Learning Model Derived from Manually Curated Dataset Peaks Accuracy of Nonsynonymous Variant Impact Prediction in Hearing Loss Genes

Fengxiao Bu[1,2], Yumei Wang[3], Jing Cheng[1], Hela Azaiez[2], Xiaofei Xu[1], Yu Lu[1], Xiarong Li[3],
Richard Smith[2], Huijun Yuan[1] 1)Medical Genetics Center, Southwest Hospital, Chongqing, China; 2)Molecular Otolaryngology and Renal Research Laboratories, University of Iowa; 3) GeneDock Co. Ltd.

Development of Core Gene Regulatory Networks in Lens Development and Disease

Deepti Anand, PhD[1], Atul Kakrana, PhD[2], Salil A. Lachke, PhD[2,3] 1)Department of Biological Sciences; 2) Center for Bioinformatics and Computational Biology; 3)Department of Biological Sciences - University of Delaware

Identifying Genetic Factors that Contribute to Human Female Infertility

Katarzyna M. Tyc[1], Warif El Yakoubi[1], Xin Tao[2], Jessica Landis[2], Yiping Zhan[2], Nathan Treff[3], Richard T. Scott, Jr.[3], Karen Schindler[1], Jinchuan Xing[1] 1)Department of Genetics, Human Genetics Institute of New Jersey, Rutgers University; 2)Foundation for Embryonic Competence; 3)Reproductive Medicine Associates of New Jersey

A Genome-first Approach to Aggregating Rare Genetic Variants in LMNA for Association with Electronic Health Record Phenotypes

Joseph Park[1,2], Michael Levin[2], Renae Judy[3], Rachel L. Kember[1], Nosheen Reza[2,4], Regeneron Genetics Center[5], Anjali T. Owens[2,4], Scott M. Damrauer[3], Daniel J. Rader[1,2,6] 1)Department of Genetics, Perelman School of Medicine, University of Pennsylvania; 2)Department of Medicine, Perelman School of Medicine, University of Pennsylvania; 3)Department of Surgery, Perelman School of Medicine, University of Pennsylvania; 4)Center for Inherited Cardiovascular Disease, Division of Cardiovascular Medicine, Hospital of the University of Pennsylvania; 5)Regeneron Genetics Center, Regeneron Pharmaceuticals; 6) Institute for Translational Medicine and Therapeutics, Perelman School of Medicine

Predicting Pathogenicity of Missense Variants Using Deep Learning

Haicang Zhang, Hongjian Qi, Chen Chen, Wendy K. Chung, Yufeng Shen Columbia University

POSTERS

Computational Prediction of Chromatin Accessibility QTLs Reveals Functional Ompact of Rare and Cryptic Variants on Transcriptional Regulation

Avantika Diwadkar, MS[1], Daniel F. Simola, PhD[1] 1)Computational Biology, Target Sciences, GlaxoSmithKline

Pseudomonas Latent Space Transformation Using Variational Autoencoder (VAE)

Alexandra J. Lee[1,2], Georgia Doing[3], Deborah Hogan[3], Casey S. Greene[2] 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

Association of Chromosomal Deletion del(20q12) with Histological Transformation of Nodal Marginal Zone Lymphoma.

Lei Qian, PhD[1], Richard Baxter, PhD[1] and Agata Bogusz, MD, PhD[2] 1)Dept. of Medical Genetics & Molecular Biochemistry, Lewis Katz School of Medicine at Temple University; 2)Department of Pathology and Laboratory Medicine, Hospital of the University of Pennsylvania,

Whole-genome DNA modification detection using Nanopore sequencing and Deep Neural Network

Qian Liu1, Li Fang[1], Kai Wang[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

Genome Analysis of Polar Bears and Brown Bears

James J. Kelley, Ryan Snow, Sean D. Smith, Joseph K. Kawash, Andrey Grigoriev
Department of Biology, Center for Computational and Integrative Biology, Rutgers University

Variational Auto Encoders for Dimensionality Reduction in Single-cell RNAseq Datasets

Ben Stear, Yuanchao Zhang, PhD, Deanne M. Taylor, PhD
Department of Biomedical and Health Informatics, Children's Hospital of Philadelphia

Structural Variation Detection From Linked-read Sequencing

Li Fang, PhD[1], Charlly Kao, PhD[2], Michael V Gonzalez[3], Renata Pellegrino da Silva, PhD[3,5], Mingyao Li, PhD[4], Hakon Hakonarson, PhD[3,5], Kai Wang, PhD[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)Center for Applied Genomics, Children's Hospital of Philadelphia 4)Department of Biostatistics, University of Pennsylvania 6)Department of Pathology and Laboratory Medicine, University of Pennsylvania; 5)Department of Pediatrics, University of Pennsylvania

A Machine Learning Model for Identification of Sequencing Artifacts in Molecular Tumor Profiling

Chao Wu, PhD[1], Xiaonan Zhao, PhD[2], Mark Welsh, BS[1], Kelliane Costello, BS[1], Kajia Cao, PhD[1], Marilyn Li, MD[1,2,3], Mahdi Sarmady PhD[1,2] 1)Division of Genomic Diagnostics, Department of Pathology and Laboratory Medicine, Children's Hospital of Philadelphia 2)Department of Pathology and Laboratory Medicine, Perelman School of Medicine, University of Pennsylvania; 3)Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania

POSTERS

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

Shihao Shen[1#], Levon Demirdjian[1#], Yang Pan[1], Shayna Stein[2], Zhijie Xie[3], Eddie Park[1], Ying Nian Wu[4], Yi Xing[1*] 1)Center for Computational and Genomic Medicine, Children's Hospital of Philadelphia; 2)Department of Biostatistics, Harvard University; 3)Department of Microbiology, Immunology & Molecular Genetics, UCLA 4) Department of Statistics, UCLA #Co-first authors. *Corresponding author

Cloud Based Genomic Harmonization Effort Across Birth Defect Diseases and Pediatric Cancers for NIH Kids First Program

Yuankun Zhu, BS[1], Bo Zhang, BS[1], Miguel, MS[1], Allison Heath, PhD[1], Deanne Taylor, PhD[1], Michele Mattioni, PhD[2], Bogdan Gavrilovic, PhD[2], Nikola Skundric, PhD[2], Kyle Hernandez, PhD[3], Maarten Leerkes, PhD[4], Daniel Miller, PhD[4], Adam Resnick, PhD[1] 1) Center for Data Driven Discovery in Biomedicine, Children's Hospital of Philadelphia; 2) Seven Bridges Genomics; 3)University of Chicago; 4) National Institutes of Health

A Bioinformatics Pipeline for Clinical Mitogenome Diagnostics

Jayaraman, Pushkala, MS[1], Balciuniene, Jorune, PhD[1], Cao, Kajia, PhD[1], Gonzalez, Michael, A, PhD[1], Wu, Chao, PhD[1], Jung, Hou-Sung, PhD[1], Trioani, Juliana[1], Sarmady, Mahdi, PhD[1,4,5], Wilson, Robert, MD, PhD[1,3,4,5], Wallace, Douglas, C, MD, PhD[3,4], Falk, Marni, J, MD, PhD[2,4,6], Dulik, Matthew, C, PhD[1,4] 1)Division of Genomic Diagnostics, Department of Pathology and Laboratory Medicine, Children's Hospital of Philadelphia; 2)Division of Human Genetics, Mitochondrial Medicine Frontier Program, Children's Hospital of Philadelphia; 3)Center for Mitochondrial and Epigenomic Medicine, Children's Hospital of Philadelphia; 4)Perelman School of Medicine, University of Pennsylvania; 5)Division of Pathology Informatics, Children's Hospital of Philadelphia 6)Division of Human Genetics, Department of Pediatrics, Children's Hospital of Philadelphia

Transcriptome Analysis of Epithelial Cells in IBD Reveals New Markers of Disease Activity

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Using Simulations to Evaluate the Effect Genetic Architecture Has on Neural Network-based Predictions

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A Comparative Study of Medical Processes of Normal Rapid Sequence Intubation and Trauma Intubation

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Chromatin digestion by the Chemotherapeutic Agent Bleomycin Produces Nucleosome and TF Footprinting Patterns Similar to Micrococcal Nuclease

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Machine Learning for Early Detection of Lung Cancer

Rehman Qureshi PhD, Andrew Kossenkov PhD, Louise Showe PhD - Wistar Institue

POSTERS

Text mining Can Improve the Detection of Disease-causing Genes and Mutations in Rare Genetic Disorders

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Discovery of allele-specific protein-RNA interactions in human transcriptomes

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Developmental Time-series Analysis of Sexual Identity in Very Early Mammalian Embryogenesis

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Population and Allelic Variation of A-to-I RNA Editing in Human Transcriptomes

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Detecting Potential Pleiotropy Across Cardiovascular and Neurological Diseases

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