FEATURED ABSTRACT

5:30 PM - 5:50 PM

Session #8: Featured Plenary Abstract Session I — Hall B - Level 1/Convention Center

PgmNr 3: Exome sequencing of 25,000 schizophrenia cases and 100,000 controls implicates 10 risk genes, and provides insight into shared and distinct genetic risk and biology with other neurodevelopmental disorders.

Tarjinder Singh

POSTER TALKS

6:30 PM - 7:30 PM

Poster Talks I: Describing Populations and Screening Applications — Room 360D - Level 3/Convention Center

PgmNr 1975/T: Meta-analysis of >1.4 million individuals for human height. **Sailaja Vedantam**

PgmNr 2396/F: Mapping annual fine-scale genetic structure of Finland through the 20th century. **Sini Kerminen**

PgmNr 3019/T: Defining and characterizing pleiotropy across 57 common diseases and complex traits. **Luke O'Connor**

BROAD INSTITUTE
GENOMIC SERVICES

ASHG19 CONFERENCE AGENDA | WED / OCT 16

BOOTH #714

10am - 4:30pm Genomic Sequencing
Jane Wilkinson & Andy Hollinger

10am - 11am Single Cell Sequencing
Tera Bowers

10am - Noon Hail Data Analysis Demo
Hail Development Team

Noon - 2pm GATK 4 Q&A
Geraldine Van der Auwera

2pm - 3pm Heidi Rehm
Medical Director, Clinical Research Sequencing Platform

3pm - 4pm T2D Portal Demo
Maria Costanzo

7:30pm - 10:30pm Broad Institute Reception

PRESENTATIONS

4:15 PM - 4:30 PM
Session #29: Single Cell Transcriptomics of the Brain to Inform the Genetics of Neurological Disorders
PgmNr 52: Single-cell dissection of brain circuitry across 800+ individuals including AD, schizophrenia, bipolar, ALS, HD, and controls.
Grand Ballroom B - Level 3/Convention Center
Manolis Kellis

4:15 PM - 4:30 PM
Session #30: Analyses Utilizing Biobanks
PgmNr 58: Global Biobank Meta-analysis Initiative: Powering genetic discovery across human diseases.
Grand Ballroom C - Level 3/Convention Center
Wei Zhou

4:15 PM - 4:30 PM
Session #33: Improved Structural Variation Detection Leads to New Insights into Disease and DevelopmentPgmNr 76: Analyses of 1,268 breakpoints from balanced chromosomal abnormalities reveals patterns of three-dimensional reorganization associated with human developmental anomalies.
Room 361D - Level 3/Convention Center
Chelsea Lowther

4:45 PM - 5:00 PM
Session #29: Single Cell Transcriptomics of the Brain to Inform the Genetics of Neurological Disorders
PgmNr 54: Human single-cell transcriptomes identify cell-types and states relevant to brain disorders.
Grand Ballroom B - Level 3/Convention Center
Sherif Gerges

4:45 PM - 5:00 PM
Session #35: Statistical Methods for GWAS Interpretation with Gene Expression Data
PgmNr 90: Calculating principled gene priors for genetic association analysis.
Room 371A - Level 3/Convention Center
Lokendra Thakur

5:00 PM - 5:15 PM
Session #30: Analyses Utilizing Biobanks
PgmNr 61: Evaluating the age-of-onset-dependent genetic architecture of complex disorders in the UK Biobank.
Grand Ballroom C - Level 3/Convention Center
Yen-Chen Anne Feng

5:00 PM - 5:15 PM
Session #35: Statistical Methods for GWAS Interpretation with Gene Expression Data
PgmNr 91: Transcriptomic-LD-score regression (tLDSC).
Room 371A - Level 3/Convention Center
Christos Chatzinakos

POSTERS

2:00 PM - 3:00 PM

Expo Hall

Bioinformatics and Computational Approaches
PgmNr 1629/W: Integration of best practice RNA-Seq workflows into cloud-based translational analysis platform. **Micah Rickles-Young**

Molecular Phenotyping and Omics Technologies
PgmNr 1815/W: Somatic mosaicism and mono-allelic expression across six brain cell types from single-cell RNA-seq in 48 AD individuals. **Maria Kousi**

Complex Traits and Polygenic Disorders
PgmNr 2001/W: Complement contributes to sex differences in risk of lupus and schizophrenia. **Nolan Kamitaki**
PgmNr 2175/W: Applying reverse linkage as a method in 1,589 Finnish migraine families. **Kumar Veerapen**
PgmNr 2199/W: Polygenic risk scores for intelligence and schizophrenia jointly contribute to community functioning in patients with a psychotic disorder. **Ari Ahola-Olli**
PgmNr 2277/W: Rare missense variants in intracerebral hemorrhage. **Jaeyoon Chung**
PgmNr 2283/W: Cross-population fine-mapping of 50 complex traits and diseases in ~675,000 individuals across three global biobanks. **Masahiro Kanai**
PgmNr 2289/W: Cross-population fine-mapping to identify shared and population-specific causal effects. **Zachary McCaw**

Cardiovascular Phenotypes
PgmNr 2775/W: Electrocardiographic P-wave duration reveals diverse genetic mechanisms of atrial fibrillation. **Lu-Chen Weng**

Statistical Genetics and Genetic Epidemiology
PgmNr 2817/W: Large scale GWAS of hypothyroidism and health risks in treated vs. untreated hypothyroidism. **Mary Pat Reeve**
PgmNr 2889/W: Tractor: A framework for well-calibrated genomic analysis of complex traits in admixed individuals. **Elizabeth Atkinson**
PgmNr 2991/W: Computational gene prioritization from GWAS using local and polygenic signal. **Elle Weeks**

Molecular Effects of Genetic Variation
PgmNr 3093/W: Autoimmune disease-associated MHC polymorphisms influence T cell receptor repertoire. **Kazuyoshi Ishigaki**

Epigenetics and Gene Regulation
PgmNr 3237/W: Allele-specific expression changes dynamically during T cell activation in HLA and other autoimmune loci. **Maria Gutierrez-Arcelus**

3:00 PM - 4:00 PM

Molecular Phenotyping and Omics Technologies
PgmNr 1770/W: Genomic landscape of subependymal giant cell astrocytomas: Unique gene expression profile with few somatic events. **Krinio Giannikou**

Complex Traits and Polygenic Disorders
PgmNr 2226/W: Expression and alternative splicing QTLs integrated with GWAS reveal novel genetic associations and causal genes for glaucoma. **Andrew Hamel**

Cardiovascular Phenotypes
PgmNr 2652/W: Testing of a genome-wide polygenic score for coronary artery disease in individuals with ancestries outside of Europe. **Akl Fahed**

Statistical Genetics and Genetic Epidemiology
PgmNr 2994/W: Imputation of variable number tandem repeat variants reveals VNTRs driving GWAS loci. **Ronen Mukamel**
PgmNr 3042/W: Polygenic prediction via Bayesian regression and continuous shrinkage priors. **Tian Ge**
PgmNr 3054/W: Estimating the autocorrelation of causal minor allele effect sizes as a function of genomic distance. **Armin Schoech**

Epigenetics and Gene Regulation
PgmNr 3252/W: Optimal strategies for linking disease-associated SNPs to genes. **Steven Gazal**

Precision Medicine, Pharmacogenomics, and Genetic Therapies
PgmNr 486/W: Mapping impact of plasma proteins on dental diseases: A Mendelian randomization study. **Dmitry Shungin**

Bioinformatics and Computational Approaches
PgmNr 1494/W: Dissecting phenotypic variation in PTSD using genetically-regulated transcriptional variation. **Nikolaos Daskalakis**

BROAD INSTITUTE
GENOMIC SERVICES

ASHG19 CONFERENCE AGENDA | THR / OCT 17

BOOTH #714

10am - 4:30pm Genomic Sequencing
Jane Wilkinson & Andy Hollinger

10am - 11am Stacey Gabriel
Senior Director, Genomics Platform

10am - 12:30pm Genomic Data Analysis and GATK 4
Bhanu Gandham & Kris Cibulskis

11am - Noon Single Cell Sequencing
Tera Bowers

Noon - 1pm Liquid Biopsy
Carrie Cibulskis

12:30pm - 1:30pm Cromwell and Cloud Computing
Robert Title

1:30pm - 2:30pm High Throughput Exome and Genome
Tom Howd

1:30pm - 2:30pm gnomAD New Release Q&A
Daniel MacArthur & Laurent Francioli

2:30pm - 3:00pm Broad Genomics CoLab Talk Booth #345
Niall Lennon & Jane Wilkinson

2:30pm - 4:00pm Clinical Genome Interpretation
Deanna Brockman

2:30pm - 4:30pm Genomic Data Analysis and GATK 4
Sushma Chaluvaldi

PRESENTATIONS

9:00 AM - 9:15 AM
Session #43: Genetic Risk Factors for Cardiovascular Diseases
PgmNr 114: Association of exome sequence variation with blood lipids in 170,000 individuals across multiple ancestries.
Grand Ballroom C - Level 3/Convention Center
George Hindy

9:30 AM - 9:45 AM
Session #40: Methods and Resources in Large-scale Population Data
PgmNr 98: Public platform with 42,291 exome control samples enables association studies without genotype sharing.
Hall B - Level 1/Convention Center
Mykyta Artomov

9:45 AM - 10:00 AM
Session #41: Somatic Mosaicism in Affected and Unaffected Individuals
PgmNr 105: Mosaic copy number variants are associated with autism spectrum disorder.
Grand Ballroom A - Level 3/Convention Center
Maxwell Sherman

9:45 AM - 10:00 AM
Session #44: Genetic Regulatory Variants and Complex Trait Associations
PgmNr 123: Causal mediation analysis identifies a comprehensive map of gene-level polygenicity and pleiotropy across 43 traits in 49 tissues.
Room 310A - Level 3/Convention Center
Yongjin Park

9:45 AM - 10:00 AM
Session #48: Causal Genes in Skeletal Development
PgmNr 147: From GWAS to causal variants: Separate regulatory base pairs at GDF5-UQCC1 underlie common knee osteoarthritis risk and developmental dysplasia of the hip.
Room 371A - Level 3/Convention Center
Terence Capellini

10:00 AM - 10:15 AM
Session #47: Genetic Mechanisms of Autism and Related Disorders
PgmNr 142: Insights into the genetic architecture of autism from exome and genome sequencing of over 60,000 individuals.
Room 370A - Level 3/Convention Center
Frederick Kyle Satterstrom

BROAD INSTITUTE
GENOMIC SERVICES

ASHG19 CONFERENCE AGENDA | THR / OCT 17

PRESENTATIONS

11:00 AM - 11:15 AM
Session #54: Evolutionary Mechanisms Underlying Phenotypic Change
PgmNr 180: Population-specific causal disease effect sizes at loci impacted by negative selection.
Room 360D - Level 3/Convention Center
Huwenbo Shi

11:15 AM - 11:30 AM
Session #51: Chromatin Accessibility and Spatial Genome Organization in Disease
PgmNr 163: Mechanistic dissection of chromatin topology disruption as an indirect, strong effect driver of neurodevelopmental disorders.
Grand Ballroom B - Level 3/Convention Center
Kiana Mohajeri

11:15 AM - 11:30 AM
Session #55: Genetic Effects on Transcriptome and Genome Traits
PgmNr 187: Exon-skipping regulation in complex disease.
Room 361D - Level 3/Convention Center
Ruize Liu

11:30 AM - 11:45 AM
Session #50: Dominant and Recessive: Not that Simple? Lessons from Clinics and Cohorts
PgmNr 158: Known disease variants in a population-wide analysis of 135,638 Finns.
Grand Ballroom A - Level 3/Convention Center
Henrike Heyne

11:45 AM - 12:00 PM
Session #57: Solving the Unsolved: Strategies for Increasing Diagnostic Yield
PgmNr 201: Identifying diagnoses beyond the exome: Lessons from challenging cases with compelling clinical phenotypes.
Room 371A - Level 3/Convention Center
Anne O'Donnell-Luria

4:30 PM - 4:45 PM
Session #66: Heritability and Dominance in Complex Traits
PgmNr 237: Estimating heritability and its enrichment in tissue-specific gene sets in admixed populations.
Room 371A - Level 3/Convention Center
Xinyi Li

5:00 PM - 5:15 PM
Session #61: Dissecting Molecular Pathways in Schizophrenia
PgmNr 219: From genetic risk variants to convergent protein networks: An integrative approach to elucidate the causal molecular mechanisms of schizophrenia.
Grand Ballroom C - Level 3/Convention Center
April Kim

5:00 PM - 5:15 PM
Session #66: Heritability and Dominance in Complex Traits
PgmNr 239: The role of dominance in complex traits in the UK Biobank.
Room 371A - Level 3/Convention Center
Duncan Palmer

ASHG19 CONFERENCE AGENDA | THR / OCT 17

POSTERS

2:00 PM - 3:00 PM

Expo Hall

Bioinformatics and Computational Approaches

PgmNr 1453/T: A scalable framework for collaborative rare disease diagnosis and gene discovery. **Hana Snow**
PgmNr 1615/T: Surfing petabytes of genomic data from your laptop. **Alex Kotlar**
PgmNr 1657/T: Systematic comparison of different evidence sources for predicting for predicting GWAS effector genes. **Benjamin Alexander**
PgmNr 1741/T: Machine learning-based prediction of gene expression modifiers prioritizes non-coding variants causal for rare disease. **Qingbo Wang**

Complex Traits and Polygenic Disorders

PgmNr 1897/T: Comprehensive genomic analysis of data-driven dietary patterns in UK Biobank identifies novel loci and reveals causal associations. **Joanne Cole**
PgmNr 1975/T: Meta-analysis of >1.4 million individuals for human height. **Sailaja Vedantam**
PgmNr 2167/T: A trans-ethnic two-stage polygenetic scoring analysis detects genetic correlation between osteoporosis and schizophrenia. **Feng Zhu**
PgmNr 2185/T: De novo missense mutations revealed by whole-exome sequencing in monozygotic twins are associated with schizophrenia. **Lili Guan**
PgmNr 2209/T: Identifying cognitive genes for nootropic drug repurposing. **Max Lam**
PgmNr 2287/T: Estimation of heritability mediated by assayed gene expression levels elucidates gene architecture of complex traits. **Douglas Yao**

Molecular and Cytogenetic Diagnostics

PgmNr 2611/T: Is likely pathogenic really 90% likely? A look at the data. **Steven Harrison**

Cardiovascular Phenotypes

PgmNr 2671/T: A genome-wide polygenic score for coronary artery disease to stratify risk among South Asians. **Minxian Wang**

Statistical Genetics and Genetic Epidemiology

PgmNr 3019/T: Defining and characterizing pleiotropy across 57 common diseases and complex traits. **Luke O'Connor**

Molecular Effects of Genetic Variation

PgmNr 3097/T: Genetic regulation of immune cell activation in subjects with immune-related disease. **Nikolaos Patsopoulos**

Precision Medicine, Pharmacogenomics, and Genetic Therapies

PgmNr 433/T: Loss-of-function variants in >4 million humans suggest that partial LRRK2 inhibition is a safe therapeutic strategy for Parkinson's disease. **Nicola Whiffin**

3:00 PM - 4:00 PM

Bioinformatics and Computational Approaches

PgmNr 1426/T: Algorithm for gene regulatory network inference recovers biological insights from large-scale gene expression data. **Aleksandr Loboda**
PgmNr 1576/T: Broad Institute's genomic platform portfolio leads support the research community. **Tera Bowers**

Complex Traits and Polygenic Disorders

PgmNr 2104/T: Characterization of focal and generalized epilepsy polygenic burden in 630,603 individuals. **Costin Leu**
PgmNr 2200/T: Biobank-based genome-wide analysis for post-traumatic stress disorder. **Chia-Yen Chen**

Cardiovascular Phenotypes

PgmNr 2776/T: Mendelian randomization reveals a causal role for obesity in stroke that varies across subtypes. **Sandro Marini**

Statistical Genetics and Genetic Epidemiology

PgmNr 2872/T: Sex-specific genetic drivers in common diseases with susceptibility sex bias: The case of multiple sclerosis. **Zhou Fang**
PgmNr 3046/T: Partitioning genetic correlation by annotation using LD score regression. **Daniel Kassler**
PgmNr 3058/T: Approximately LD independent regions for biobank scale genetic analyses. **Jacob Ulirsch**

Molecular Effects of Genetic Variation

PgmNr 3130/T: Tissue-specific transcriptional signatures in reciprocal genomic disorders: Insights from mouse brain and human neuronal models. **Rachita Yadav**

Epigenetics and Gene Regulation

PgmNr 3334/T: Chromatin state annotations across 833 tissue/cell types help annotate thousands of additional GWAS loci. **Carles Boix**

BROAD INSTITUTE
GENOMIC SERVICES

ASHG19 CONFERENCE AGENDA | FRI / OCT 18

BOOTH #714

10am - 3:30pm Genomic Sequencing
Jane Wilkinson & Andy Hollinger

10am - 11am T2D Portal Demo
Maria Costanzo

11am - Noon Genomic Data Analysis and GATK 4
Ruchi Munshi

1pm - 2pm RNA Analysis Pipelines
Micah Rickles-Young

1:30pm - 2:30pm Rare Disease, CMG and gnomAD
Anne O'Donnell Luria

PRESENTATIONS

9:15 AM - 9:30 AM
Session #73: Alternative Methods for Evaluating Variant Pathogenicity
PgmNr 261: Protein function-specific structural insights into the effect of Mendelian disease variants in 1,330 human genes.
Room 360D - Level 3/Convention Center **Sumaiya Iqbal**

9:30 AM - 9:45 AM
Session #70: Fast Methods for Genome Analysis
PgmNr 250: Efficient approximation of GWAS resampling for method validation on massive datasets.
Grand Ballroom B - Level 3/Convention Center **Nikolas Baya**

9:30 AM - 9:45 AM
Session #75: Reproductive Fitness: Genetic Insights into Fertility
PgmNr 270: Aneuploidy and recombination across chromosomes, gametes, and individuals from large-scale single-sperm sequencing.
Room 370A - Level 3/Convention Center **Avery Bell**

POSTERS

1:00 PM - 2:00 PM

Bioinformatics and Computational Approaches
PgmNr 1517/F: Characterizing deleterious variation using sub-population allele frequency information. **Konrad Karczewski**
PgmNr 1655/F: Improving the informativeness of Mendelian disease pathogenicity scores for common diseases and complex traits. **Samuel Kim**

Complex Traits and Polygenic Disorders
PgmNr 1907/F: Understanding the genetic basis of type 2 diabetes risk: Whole genome sequence analysis of glycemic traits from the NHLBI's TOPMed Program. **Alisa Manning**
PgmNr 1973/F: A large-scale whole genome sequencing to identify less common and rare variants associated with osteoporosis and fracture: The NHLBI TopMed Study. **Yi-Hsiang Hsu**
PgmNr 2009/F: Cross-population polygenic risk score predictions improved by prioritization of variants in predicted cell-type-specific regulatory elements. **Tiffany Amariuta**
PgmNr 2153/F: The influence of common and rare variation on risk for autism spectrum disorders. **Celia van der Merwe**
PgmNr 2213/F: Nightmares: Genetic variants and risk of psychiatric disorders. **Hanna Ollila**
PgmNr 2297/F: Genetic architecture of phenome-wide latent factors in the UK Biobank. **Caitlin Carey**

Statistical Genetics and Genetic Epidemiology
PgmNr 2873/F: Epilepsy and epilepsy subsyndromes: Findings from the Epi25 Consortium GWASs. **Alexander Smith**
PgmNr 2891/F: Most rare and high-risk CNV carriers do not have major health, cognitive, or socioeconomic consequences. **Elmo Saarentaus**
PgmNr 2957/F: Shrinkage methods utilising the estimation of error quantity to improve GWAS and polygenic risk score. **Yunfeng Ruan**
PgmNr 2993/F: Characterizing the block-jackknife based significance testing used in stratified LD-score regression. **Katherine Tashman**
PgmNr 3029/F: A catalog of likely-causal, coding variant associations from imputed UK Biobank exome data. **Alison Barton**

Molecular Effects of Genetic Variation
PgmNr 3161/F: Convergent pathways of 22q11.2 deletion and schizophrenia in human neurons. **Olli Pietilainen**
PgmNr 3203/F: Manual curation of homozygous predicted loss-of-function variants in 125,000 exomes reveals 1,752 human genes tolerant to inactivation. **Eleanor Seaby**

Cancer Genetics
PgmNr 827/F: Development of a large scale liquid biopsy platform for classification of microsatellite instability. **Carrie Cibulskis**

2:00 PM - 3:00 PM

Molecular Phenotyping and Omics Technologies
PgmNr 1868/F: Application of lean manufacturing methodologies in high throughput genomic sequencing. **Tom Howd**

Complex Traits and Polygenic Disorders
PgmNr 1958/F: The genetic architecture of liver cirrhosis: A multi-trait genome-wide association analysis. **Connor Emdin**
PgmNr 2036/F: Unexplained familial hypercholesterolemia in African Americans: No detectable mutation does not necessarily imply a polygenic basis. **Onuralp Soylemez**

Evolution and Population Genetics
PgmNr 2396/F: Mapping annual fine-scale genetic structure of Finland through the 20th century. **Sini Kerminen**

Molecular and Cytogenetic Diagnostics
PgmNr 2612/F: Whole genome sequencing versus standard-of-care genetic testing in patients with a suspected genetic disorder. **Deanna Brockman**

Statistical Genetics and Genetic Epidemiology
PgmNr 2804/F: The functional impacts of rare coding variants in 46,000 individuals on 23 quantitative phenotypes. **Peter Dornbos**
PgmNr 3038/F: Modeling imputation uncertainty in fine-mapping improves accuracy. **Carlos Albors**
PgmNr 3050/F: Multi-ancestry meta-analysis of genome-wide association studies. **Alicia Martin**

Molecular Effects of Genetic Variation
PgmNr 3110/F: Colocalizing gene-lifestyle interaction associations with molecular signatures of smoking and alcohol consumption. **Timothy Majarian**

Epigenetics and Gene Regulation
PgmNr 3308/F: Explore the value of circRNA-miRNA multimolecular regulatory network in the diagnosis of early onset schizophrenia. **Sha Liu**

Cancer Genetics
PgmNr 842/F: Enrichment of rare germline variants in DNA repair and cell cycle genes in chronic lymphocytic leukemia. **Nicholas Moore**

Evolution and Population Genetics
PgmNr 2384/F: Standardized statistics to detect balancing selection utilizing substitution data. **Katherine Siewert**

PRESENTATIONS

8:30 AM **8:45 AM**
Session #100: Uncovering Genome Complexity and Function with Long-read Sequencing
PgmNr 311: Long-read transcriptome sequencing in over 60 human tissue samples reveals isoform diversity.
Room 371A - Level 3/Convention Center **Beryl Cummings**

8:45 AM **9:00 AM**
Session #96: Computational Methods for Genetic Data
PgmNr 296: Public programmatic access to GWAS summary statistics and analytical methods.
Room 310A - Level 3/Convention Center **Marcin von Grotthuss**

9:15 AM **9:30 AM**
Session #96: Computational Methods for Genetic Data
PgmNr 298: Estimating assortative mating and its changes over time in samples of unrelated individuals.
Room 310A - Level 3/Convention Center **Patrick Turley**

9:45 AM **10:00 AM**
Session #107: Methods and Resources for Improved Genomic Variant Interpretation
PgmNr 351: gnomAD-SV: An open resource of structural variation for medical and population genetics.
Room 361D - Level 3/Convention Center **Ryan Collins**

10:30 AM **10:45 AM**
Session #105: Mechanisms of Immune Cell Phenotypes and Clonal Hematopoiesis
PgmNr 342: Multimodal single-cell analysis of 70,000 human memory T cells characterizes genetic associations with immune cell states and gene expression in a Peruvian tuberculosis progression cohort.
Room 310A - Level 3/Convention Center **Aparna Nathan**

10:45 AM **11:00 AM**
Session #102: Darwin's Tumor: Mutation, Selection, and Evolution in Cancer Genomes
PgmNr 325: Detecting cancer vulnerabilities through gene networks under purifying selection.
Grand Ballroom A - Level 3/Convention Center **Heiko Horn**

10:45 AM **11:00 AM**
Session #105: Mechanisms of Immune Cell Phenotypes and Clonal Hematopoiesis
PgmNr 343: Monogenic and polygenic inheritance become instruments for clonal selection.
Room 310A - Level 3/Convention Center **Po-Ru Loh**

12:10 PM **12:30 PM**
Session #110: Featured Plenary Abstract Session IV
PgmNr 371: Genomic analyses in 3 million individuals identify genetic determinants of questionnaire response bias and study participation with potential implications for GWAS interpretation.
Hall B - Level 1/Convention Center **Andrea Ganna**