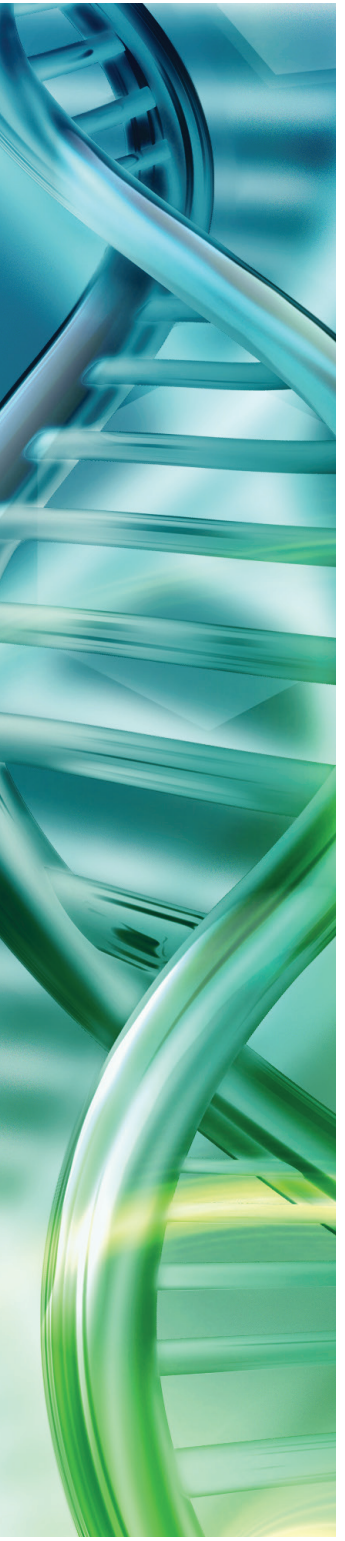

2017 SAGES

Symposium on Advances in Genomics, Epidemiology & Statistics

PROGRAM BOOKLET

Friday, June 9
9:00 a.m. - 6:00 p.m.

Arthur H. Rubenstein Auditorium
Smilow Center for Translational Research
3400 Civic Center Blvd.

CENTER FOR CLINICAL EPIDEMIOLOGY AND BIOSTATISTICS

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Center for Genetics and Complex Traits
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NIH

Welcome

Advances in technology and significant decrease in the associated costs are driving progress in genomic studies. Studies of whole exome and genome sequences of complex traits in large samples are becoming increasingly common. Other sources of high-dimensional information, including expression, epigenetic, metabolic and microbiomic data, are also being collected in disease and control samples. To fully understand the complex bases of human disease and trait variation, all of these factors should be properly considered in a unified analytical framework, together with epidemiological data on environmental exposures and other risk factors.

SAGES brings together an interdisciplinary group of scientists working in the fields of genomics, epidemiology, and statistics, to address these challenges. The forum provides an opportunity for scientists at all levels in their career to convene and review new developments in these areas of research. The symposium aims to facilitate exchange of ideas and promote interactions and collaborations among participants.

9:00-9:45 am	**REGISTRATION & BREAKFAST**
9:45-10:00 am	**Welcome and Opening Remarks:** **Harold Feldman,** *University of Pennsylvania*
10:00-10:30 am	**SESSION 1** Moderator: Hongzhe Li, University of Pennsylvania **Quantifying Genetic Regulatory Variation Affecting Each Gene in Human Populations** **Tuuli Lappalainen,** *New York Genome Center*
10:30-11:00 am	**Statistical Methods for Single-Cell RNAseq Experiments** **Christina Kendziorski,** *University of Wisconsin*
11:00-11:30 am	**Overcoming Bias and Batch Effects in Single Cell and Bulk RNAseq Data** **Rafael Irizarry,** *Harvard University*
11:30-12:30 pm	**LUNCH & POSTER SESSION 1** *(odd numbered posters)*
12:30-1:30 pm	**LUNCH & POSTER SESSION 2** *(even numbered posters)*
1:30-2:00 pm	**SESSION 2** Moderator: Joan Bailey-Wilson, National Human Genome Research Institute **Genome, Phenome, and What Happens in Between** **Haky Im,** *University of Chicago*
2:00-2:30 pm	**Leveraging 1000 Genomes to Improve Disease Gene Localization** **William C. L. Stewart,** *Nationwide Children's*
2:30-3:00 pm	**Challenges and New Approaches for Complex Trait Mapping in Ancestrally Diverse Populations** **Tim Thornton,** *University of Washington*
3:00-3:30 pm	**COFFEE BREAK**
3:30-4:00 pm	**SESSION 3** Moderator: Ingo Ruczinski, Johns Hopkins University **Metagenomic and Genomic Analysis of Host-Microbe Interactions of the Skin** **Elizabeth Grice,** *University of Pennsylvania*
4:00-4:30 pm	**Intersecting Pathology Images and Gene Expression Data to Understand Drivers of Complex Phenotypes** **Barbara Engelhardt,** *Princeton University*
4:30-5:30 pm	**CONCLUSION & COCKTAIL RECEPTION**

Poster Numbers & Titles

1	Genetic risk factors for PANDAS and Sydenham's chorea, two pediatric autoimmune diseases of the brain following Streptococcus infection *I Agalliu, M Gonzalez, F Mentch, D Agalliu, J Frankovich, H Hakonarson, T Cutforth*
2	Integrative analysis identifies immune-related enhancers and lncRNAs perturbed by genetic variants associated with Alzheimer's disease *A Amlie-Wolf, M Tang, J King, B Dombroski, Y Chou, E Mlynarski, G Schellenberg, L Wang*
3	SCnorm: A quantile-regression based approach for robust normalization of single-cell RNA-seq data *R Bacher, L Chu, N Leng, A Gasch, J Thomson, R Stewart, M Newton, C Kendziorski*
4	The missing landscape of human genomic diversity in the Arabian Peninsula *N Bakhsh, L Jackson, C Cross, F Jackson*
5	Regulation of Keratinocyte Gene Expression by the Skin Microbiome *C Bartow-McKenney, J Meisel, J Horwinski, E Grice*
6	Linear Discriminant Analysis Predicts Extension in Patients with Juvenile Idiopathic Arthritis *A Brescia, M Simonds, S McCahan, H Bunnell, K Sullivan, C Rose*
7	Cloudy with a Chance of Reproducibility: Interactive Quality Control for Genetic Studies *B Cole, J Moore*
8	Defiant: (DMRs: Easy, Fast, Identification and ANnoTation) Identifies Differentially Methylated Regions from Iron-Deficient Rat Hippocampus *D Condon, P Tran, Y Lien, J Schug, M Georgieff, R Simmons, K Won*
9	A Bayesian Allele Specific Expression Model for Large Scale Genetic Expression studies using a Sparse Overdispersed Poisson Generalized Linear Models *G Gliner, Y Park, B Lamarche, C Brown, B Engelhardt*
10	Bayesian Hierarchical Modeling of Genic Sub-Region Intolerance *T Hayeck, N Stong, D Goldstein , A Allen*
11	Violence and Allostatic load in African American young adults *L Jackson, M Shestov, F Saadatmand, J Wright*
12	Integrative Deep Models for Alternative Splicing *A Jha, M Gazzara, Y Barash*
13	eQTL analysis of megakaryocytes derived from induced pluripotent stem cells *K Kammers, M Taub, I Ruczinski, J Martin, L Yanek, A Frazee, Y Gao, D Hoyle, N Faraday, D Becker, L Cheng, Z Wang, J Leek, L Becker, R Mathias*
14	Genetic and Phenotypic Heterogeneity of Mood Disorders in a Large Multigenerational Pedigree *R Kember, L Hou, X Ji, L Andersen, L Estrella, F McMahon, C Brown, M Bucan*
15	Modified Random Forest for Trio Data with Alternative Splitting Criterion to allow for Missing Genotypes *Q Li, E Holzinger, J Bailey-Wilson*

Poster Numbers & Titles

16	Rare Copy Number Variants in Over 100,000 Subjects Reveal Novel Disease Associations *R Li, J Glessner, B Coe, J Li, X Chang, C Kao, A Cederquist, C Kim, M Khan, F Mentch, M Garris, D Abrams, G Otieno, P Sleiman, E Eichler, H Hakonarson*
17	A comparison of methods for identification of genetic variants related to age-of-onset of Cystic fibrosis related diabetes *H Ling, P Zhang, EW Pugh, M Atalar, S Blackman*
18	Risks of familial breast cancer associated with known and proposed breast cancer susceptibility genes *K Maxwell, T Slavin, J Lilyquist, J Vijai, S Neuhausen, S Hart, V Ravichandran, T Thomas, A Maria, K Schrader, R Moore, C Hu, B Wubbenhorst, B Wenz, K D'Andrea, S Domchek, M Robson, P Peterlongo, P Radice, J Ford, J Garber, C Szabo, K Offit, F Couch, J Weitzel, K Nathanson*
19	Determining and inducing gene expression patterns underlying cell identity *I Mellis, W Yang, P Shah, R Jain, A Raj*
20	Urinary Epidermal Growth Factor and Monocyte Chemoattractant Protein-1 as Biomarkers of Renal Involvement in ANCA-Associated Vasculitis *C Najem, W Ju, H Gore, V Nair, D Cuthbertson, R Rhee, L Mariani, S Carette, G Hoffman, N Khalidi, C Koening, C Langford, C McAlear, P Monach, L Moreland, C Pagnoux, P Seo, U Specks, A Sreih, S Ytterberg, J Krischer, M Kretzler, P Merkel*
21	Outlier detection for improved differential splicing quantification from RNA-Seq experiments with replicates *S Norton, J Vaquero-Garcia, Y Barash*
22	The genomic landscape of matched primary and metastatic breast cancer tumors *M Paul, L Chodosh*
23	Identification of epistatic interactions between the human RNA demethylases FTO and ALKBH5 with gene set enrichment analysis informed by differential methylation *E Piette, J Moore*
24	Change in ancestry related assortative mating in the United States and implications for genetic studies *R Sebro, G Peloso, J Dupuis, N Risch*
25	Genes & Geography: A comprehensive study of geographical effects on addiction and immunity in populations *M Shestov, L Jackson*
26	DNA methylation markers associated with injection drug use status and HIV infection among chronic injection drug users in the ALIVE study *C Shu, K Bakulski, K Benke, A Jaffe, S Wang, S Sabunciyan, S Mehta, G Kirk, B Maher*
27	QRank: A novel quantile regression tool for eQTL discovery *X Song, G Li, Z Zhou, X Wang, I Ionita-Laza, Y Wei*
28	Stem cell divisions, somatic mutations, cancer etiology, and cancer prevention *C Tomasetti, L Li, B Vogelstein*
29	Assessing the Geospatial Distribution of Asthma Exacerbations in Philadelphia Using Electronic Health Record (EHR)-Derived Data *S Xie, R Greenblatt, M Levy, A Apter, M Ross, B Himes*
30	Testing for genetic association in case-control studies incorporating multivariate disease characteristics *H Zhang, T Ahearn, M Garcia-Closas, N Chatterjee*

2017 SAGES
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Notes

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