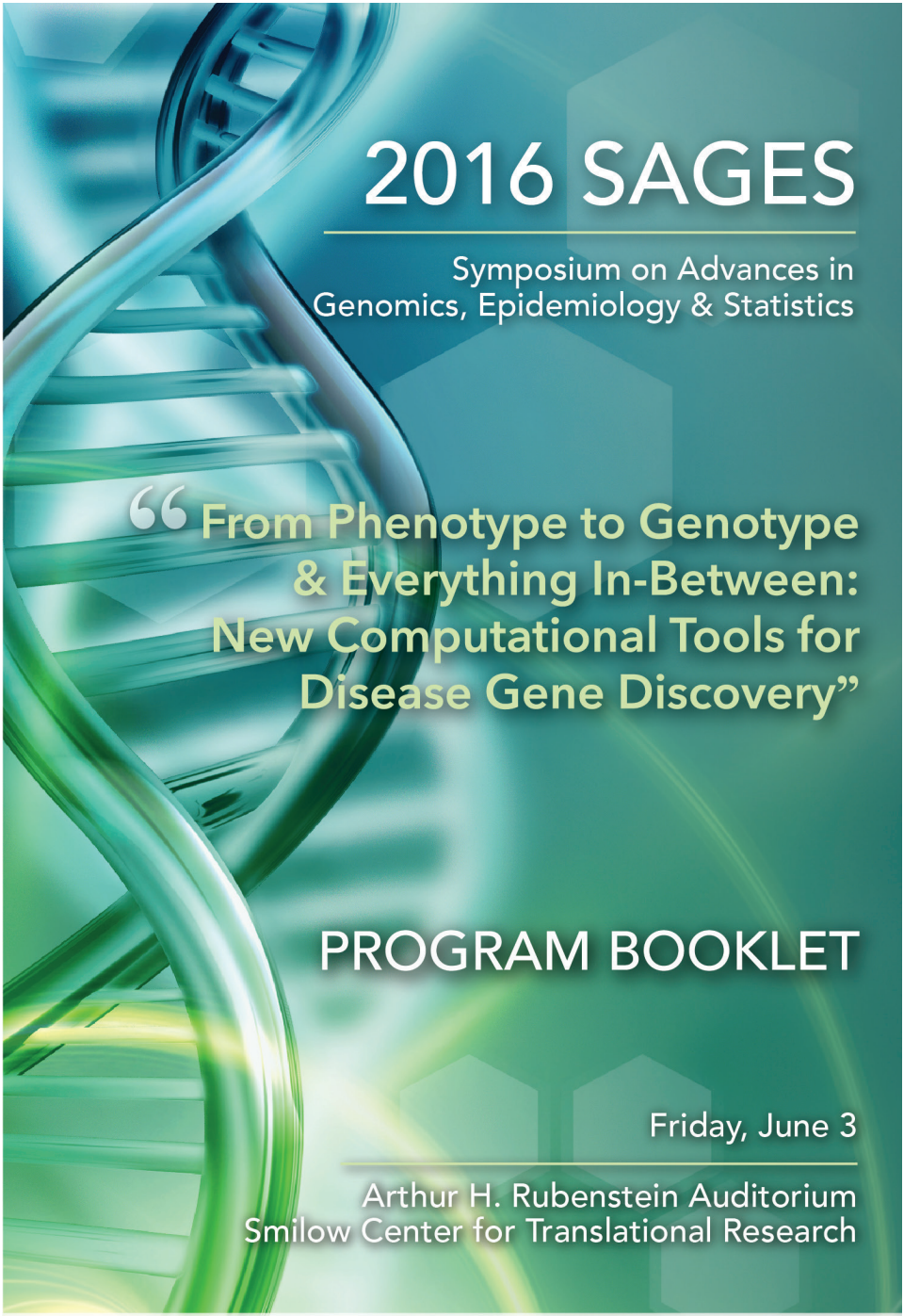

# 2016 SAGES

## Symposium on Advances in Genomics, Epidemiology & Statistics

## "From Phenotype to Genotype & Everything In-Between: New Computational Tools for Disease Gene Discovery"

## PROGRAM BOOKLET

Friday, June 3

Arthur H. Rubenstein Auditorium
Smilow Center for Translational Research

CGACTatgctaggatctatacatcacgactcgcccgca
Center for Genetics and Complex Traits
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The Children's Hospital *of* Philadelphia
RESEARCH INSTITUTE

CCEB

NIH
National Institutes of Health

# Welcome

The Center for Genetics and Complex Traits (CGACT) of the Perelman School of Medicine at the University of Pennsylvania will hold a Symposium on Advances in Genomics, Epidemiology and Statistics (SAGES) on June 3, 2016 in the Arthur H. Rubenstein Auditorium of the Smilow Center for Translational Research in Philadelphia, PA.

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.

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|---|---|
| 9:30 - 10:20 am | **Registration & Light Breakfast** |
| 10:20 - 10:30 am | **Welcome & Opening Remarks** |
| | **SESSION 1**<br>*Moderator: Nandita Mitra, University of Pennsylvania* |
| 10:30 - 11:00 am | "Replicability and winner's curse in genome-wide association studies"<br>***Itsik Pe'er, Columbia University*** |
| 11:00 - 11:30 am | "Robust analysis of secondary phenotypes in case-control genetic association studies"<br>***Andrew Allen, Duke University*** |
| 11:30 - 12:00 pm | **"Substructure in whole-genome sequencing studies: detection and adjustment procedures"**<br>***Christoph Lange, Harvard University*** |
| 12:00 - 1:30 pm | **Lunch & Poster Session** |
| | **SESSION 2**<br>*Moderator: Peter Kanetsky, Moffitt Cancer Center* |
| 1:30 - 2:00 pm | "Reconstructing the starts of human tumors"<br>***Kimberly Siegmund, University of Southern California*** |
| 2:00 - 2:30 pm | "Advances in brain tumor research: Leveraging BIG data for BIG discoveries"<br>***Jill Barnholtz-Sloan, Case Western University*** |
| 2:30 - 3:00 pm | "Understanding etiology of lung and common cancers"<br>***Chris Amos, Dartmouth College*** |
| 3:00 - 3:30 pm | **Coffee Break** |
| | **SESSION 3**<br>*Moderator: Blanca Himes, University of Pennsylvania* |
| 3:30 - 4:00 pm | "Genetics of gene expression: from rare to common, from ubiquitous to cell specific"<br>***Christopher Brown, University of Pennsylvania*** |
| 4:00 - 4:30 pm | "Understanding the impact of rare regulatory variation"<br>***Alexis Battle, Johns Hopkins University*** |
| 4:30 - 5:30 pm | **Conclusion & Cocktail Reception** |

# Poster Numbers & Titles

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|---|---|
| 1 | **A large de novo germline sequencing study of bilateral RB patients finds no higher oncogenic potential of nonsense mutations at CpG sites in RB1 gene.**<br>**V Aggarwala, A Ganguly, BF Voight* |
| 2 | **Identified structural variants associated with multiple phenotypes of COPDGene African American Study Cohort.**<br>**F Begum, I Ruczinski, S Li, MM Parker, J Hetmanski, TH Beaty, E Silverman, J Crapo, and COPDGene Investigators* |
| 3 | **Synovial fluid proteins differentiate patients with oligoarticular juvenile idiopathic arthritis who are destined to extend from those who will remains persistent in course.**<br>*AC Brescia, MM Simonds, KE Sullivan, CD Rose* |
| 4 | **Genome Wide Association Study Identifies EFEMP1 as a New Candidate Biliary Atresia Susceptibility Gene.**<br>**Y Chen, CM Grochowski, M Gilbert, R Rajagopalan, KM Loomes, NB Spinner, M Devoto* |
| 5 | **Identifying functionally relevant Mental Health hotspots and polymorphisms in African Americans.**<br>**C Cross, L Jackson, M Shestov* |
| 6 | **Exploring Tissue-Specific Effects of Rare Non-Coding Variants.**<br>**F Damani, Y Kim, X Li, J Davis, E Tsang, C Chiang, Z Zappala, B Strober, The GTEx Consortium, I Hall, SB Montgomery, A Battle* |
| 7 | **Causal Effects of Blood Pressure Components on Brain Atrophy and White Matter Integrity: A Mendelian Randomization Analysis Based On Individual Participant Data.**<br>**J Ding, O Meirelles, LJ Launer* |
| 8 | **PheWAS study using research participants' self -reported data provides insight intoTh17/IL-17 pathway.**<br>*MG Ehm, JL Aponte, MN Chiano, LM Yerges-Armstrong, JN Barker, SF Cook, A Gupta, DA Hinds, L Li, T Johnson, MA Simpson, C Tian, LC McCarthy, D Rajpal, DM Waterworth* |
| 9 | **Prioritize Risk Genetic Variants in Regulatory DNA Sequences Using Disease-relevant Gene Regulatory Networks.**<br>*L Gao, Y Uzun, X Ma, J Wang, B He, K Tan* |
| 10 | **Assessing the role of genetic variation in amplified musculoskeletal pain syndromes (AMPS) in pediatric populations.**<br>**MV Gonzalez, SA Gmuca, J Tress, FD Mentch, R Chiavacci, P Sleiman, GA Barr, DD Sherry, H Hakonarson* |
| 11 | **r2VIM: Variable selection method for identifying interaction effects.**<br>**E Holzinger, S Szymcak, J Malley, A Dasgupta, Q Li, J Bailey-Wilson* |
| 12 | **Gender Differences in the Experience of Violence, Discrimination, and Stress Hormone in African Americans: Implications for Public Health.**<br>**L Jackson, M Shestov, M Abbas, F Saadatmand* |
| 13 | **Increased burden of deleterious variants in essential genes in autism spectrum disorder.**<br>**X Ji, RL Kember, CD Brown, M Bucan* |

* Eligible for poster award

# Poster Numbers & Titles

| | |
|---|---|
| 14 | **Accounting for technical batch effects in single-cell RNA sequencing analysis.**<br>**C Jia, M Li, NR Zhang* |
| 15 | **Assessing intra-tumor heterogeneity and tracking longitudinal and spatial clonal evolution by next-generation sequencing.**<br>**Y Jiang , Y Qiu, AJ Minn, NR Zhang* |
| 16 | **Integrity of induced pluripotent stem cell (iPSC) derived megakaryocytes as assessed by genetic and transcriptomic analysis.**<br>**K Kammers, M Taub, I Ruczinski, J Martin, LR Yanek, A Frazee, Y Gao, D Hoyle, N Faraday, DM Becker, L Cheng, ZZ Wang, J Leek, LC Becker, RA Mathias* |
| 17 | **Epidemiology and Clinical studies related to Elongated Styloid Process (ESP) causing head and neck pain.**<br>**HM Khan, AD Fraser, M Mupparapu* |
| 18 | **Training a Type 2 Diabetes Specific Functional Sequence Predictor.**<br>**KM Lorenz, BF Voight* |
| 19 | **Theoretical constraints imposed by the transcriptome on genotype-phenotype maps.**<br>**IA Mellis, A. Raj* |
| 20 | **Linkage Analyses Reveals Significant Association for Myopia.**<br>**AM Musolf, CL Simpson, F Murgia, L Portas, JE Bailey-Wilson, D Stambolian* |
| 21 | **Differential Expression Analysis of Gene and Transcript Abundance for Single Cell RNA-Seq Data using STAR and HISAT Aligners.**<br>**JS Ngwa, M Liu, R Wojciechowski , T Beaty, D Zack, I Ruczinski* |
| 22 | **Multivariate models from RNA-Seq SNVs yield candidate molecular targets for biomarker discovery: SNV-DA.**<br>**MR Paul, NP Levitt, DE Moore, PM Watson, RC Wilson, CE Denlinger, DK Watson, PE Anderson* |
| 23 | **A Chemogenomics Approach to Identify Bacterial Metabolites with Immune-modulatory Effects via Human Host Receptors.**<br>**S Saha, C Jayawickreme, JP Marino, A Benowitz, DK Rajpal, JR Brown* |
| 24 | **Gene-based analysis identified the gene ZNF248 is associated with late-onset asthma in African Americans.**<br>**L Wang, YD Salinas, AT DeWan* |
| 25 | **The Power of Population Diversity in Probing the Biology of Positive Selection in Asthma Disparities.**<br>**B Wilson, TE Mason, L Ricks-Santi, GM Dunston* |
| 26 | **A Powerful Procedure for Pathway-based Meta-Analysis Using Summary Statistics Identifies 43 Pathways Associated with Type II Diabetes in European Populations.**<br>**H Zhang, W Wheeler, PL Hyland, Y Yang, J Shi, N Chatterjee, K Yu* |

* Eligible for poster award

# 2016 SAGES Organizing Committee

**Marcella Devoto**
*Chair, University of Pennsylvania,*
*The Children's Hospital of Philadelphia*

**Joan Bailey-Wilson**
*National Human Genome Research Institute*

**Iuliana Ionita-Laza**
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**Peter Kanetsky**
*Moffitt Cancer Center*

**Hongzhe Li**
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**Nandita Mitra**
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**Adam Naj**
*University of Pennsylvania*

**Timothy Rebbeck**
*Harvard University*

**Ingo Ruczinski**
*Johns Hopkins University*

# Notes

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