5th Human Genetics in NYC Conference

Keynote Address:

"Insights into the Genetics of Autism"

Evan E. Eichler, PhD

Associate Member, New York Genome Center
Professor, Genome Sciences, University of Washington
Investigator, Howard Hughes Medical Institute

Friday, February 2, 2018

New York Genome Center
101 Avenue of the Americas
1st Floor Auditorium
New York, NY 10013

5th Human Genetics in NYC Conference

Conference Leaders/Moderators:

Tom Maniatis, PhD, New York Genome Center & Columbia University
Bruce D. Gelb, MD, Icahn School of Medicine at Mount Sinai
Jean-Laurent Casanova, MD, PhD, The Rockefeller University & Howard Hughes Medical Institute
David Goldstein, PhD, New York Genome Center, Columbia University & Weill Cornell Medicine

PROGRAM

8:30 AM - 9:00 AM	**Registration & Breakfast**
9:00 AM - 9:10 AM	**Opening Remarks:** **Cheryl A. Moore** President & Chief Operating Officer, New York Genome Center **Tom Maniatis, PhD** Scientific Director & Chief Executive Officer, New York Genome Center Director, Columbia Precision Medicine Initiative Isidore S. Edelman Professor, Biochemistry Chair, Department of Biochemistry & Molecular Biophysics, Columbia University **Jean-Laurent Casanova, MD, PhD** Professor, St. Giles Laboratory of Human Genetics of Infectious Diseases The Rockefeller University Investigator, Howard Hughes Medical Institute
9:10 AM - 10:00 AM	**Keynote Address: Evan E. Eichler, PhD** Associate Member, New York Genome Center Professor, Genome Sciences, University of Washington Investigator, Howard Hughes Medical Institute **"Insights into the Genetics of Autism"**
10:00 AM - 10:30 AM	**Break & Poster Session**
10:30 AM - 11:00 AM	**Eimear Kenny, PhD** Assistant Professor, Genetics and Genomic Sciences Icahn School of Medicine at Mount Sinai **"Population Genetics in an Era of Precision Medicine"**
11:00 AM - 11:30 AM	**Olga Troyanskaya, PhD** Professor, Department of Computer Science & Lewis-Sigler Institute for Integrative Genomics, Princeton University Deputy Director for Genomics, Flatiron Institute, Simons Foundation **"From Genome to Clinical Outcome: Decoding the Impact of Noncoding Mutations in Human Disease"**

11:30 AM - 12:00 PM **Molly Przeworski, PhD**
Professor, Department of Biological Sciences
Department of Systems Biology, Columbia University

"Sex and Age Effects on Human Germline Mutation"

12:00 PM - 1:00 PM **Lunch, Poster Session & Tours**

1:00 PM - 1:30 PM **Molly Gale Hammell, PhD**
Assistant Professor, Cold Spring Harbor Laboratory

"Mobile Genomics: TDP-43 Links Mobile Element Control to Neurodegenerative Disease"

1:30 PM - 2:00 PM **Philip L. De Jager, MD, PhD**
Weil-Granat Professor of Neurology, Taub Institute for Research on Alzheimer's Disease and the Aging Brain, and the Precision Medicine Initiative
Director, Center for Translational & Computational Neuro-Immunology
Director, Multiple Sclerosis Clinical Care & Research Center
Columbia University Medical Center

"The Genetic Architecture of Human Microglial Traits: Grounding Network Models & Clinical Translation"

2:00 PM - 2:15 PM **Daniel Felsky, PhD**
Postdoctoral Research Assistant, Center for Translational and Computational Neuroimmunology, Department of Neurology, Columbia University Medical Center

"Neuropathological Correlates and Molecular-Genetic Causes of Microglial Activation in Elderly Human Brain"

2:15 PM - 2:30 PM **Cristen J. Willer, PhD**
Associate Professor, Division of Cardiovascular Medicine, Department of Internal Medicine, Department of Human Genetics, Department of Computational Medicine and Bioinformatics, University of Michigan

"Genome-Wide Association Study of 1 Million People Identifies 111 Loci for Atrial Fibrillation"

2:30 PM - 2:45 PM **Break & Poster Session**

2:45 PM - 3:15 PM **Matthew T. Maurano, PhD**
Assistant Professor, Institute for Systems Genetics, NYU Langone Health

"Decoding Regulatory Variation"

3:15 PM - 3:30 PM **Ansuman Satpathy, MD, PhD**
Instructor, Department of Pathology, Stanford University School of Medicine

"Enhancer Connectome in Primary Human Cells Reveals Target Genes of Disease-Associated DNA Elements"

3:30 PM - 3:45 PM	**Julia TCW, PhD** Druckenmiller Fellow, New York Stem Cell Foundation Postdoctoral Fellow, Department of Neuroscience, Ronald M. Loeb Center for Alzheimer's Disease, The Friedman Brain Institute Icahn School of Medicine at Mount Sinai **"Cell Autonomous Effects of APOE ε4/ε4 on Human iPSC-Derived Astrocytes and Microglia"**
3:45 PM - 4:00 PM	**Sahin Naqvi** Graduate Student, Whitehead Institute for Biomedical Research **"A Multi-Tissue Program of Sex-Biased Gene Expression Conserved Across Mammals"**
4:00 PM - 4:15 PM	**Ronan Chaligne, PhD** Postdoctoral Research Associate, New York Genome Center & Weill Cornell Medicine **"Epigenetic Evolution and Lineage Histories of Chronic Lymphocytic Leukemia"**
4:15 PM - 4:45 PM	**Christina Leslie, PhD** Member, Computational Biology Program, SKI, Memorial Sloan Kettering Cancer Center **"Decoding Immune Cell States in Development and Disease"**
4:45 PM - 5:15 PM	**Marcin Imielinski, MD, PhD** Core Faculty Member, New York Genome Center Assistant Professor of Computational Genomics, Assistant Professor of Pathology & Laboratory Medicine, Weill Cornell Medicine **"Signatures of Complex Rearrangements Across Thousands of Human Cancer Genomes"**
5:15 PM	**Closing Remarks: Tom Maniatis, PhD**
5:30 PM	**Tours**

POSTER SESSION

Listed in alphabetical order according to first author's last name

1. Peter A. Andrews, Ivan Iossifov, Jude Kendall, Steven Marks, Lakshmi Muthuswamy, Zihua Wang, Dan Levy, Michael Wigler – *Mumdex: MUM-Based Structural Variation Detection Plus Copy Number and Visualization Software*

2. Julie M. Behr, Marcin Imielinski – *Phasing of Cancer Genome Rearrangements with Aneuploid Loci*

3. Evan Biederstedt, Aaron Dornbrand-Lo, Khagay Nagdimov, Marcin Imielinski – *bxBam: Software Package for Querying Barcoded-Reads with 10x Data*

4. Margot Brandt, Ana Vasileva, Tuuli Lappalainen – *Characterizing Causal Cis-Regulatory Variants Using Computational Approaches and CRISPR/Cas9 Genome Editing*

5. Stephane Castel, Alejandra Cervera, Pejman Mohammadi, Ana Vasileva, Tuuli Lappalainen – *Modified Penetrance of Coding Variants by Cis-Regulatory Variation Contributes to Disease Risk*

6. Sinead F. Cullina, Gillian M. Belbin, Genevieve L. Wocjik, Elena P. Sorokin, Matthew Levin, Christopher R. Gignoux, Eimear E. Kenny – *Exploring Population-Specific Incidence of Disease in a Multi-Ethnic Health System Reveals Native American Haplotype Underlying Peripheral Artery Disease in Dominicans*

7. Tyler Cutforth, Ilir Agalliu, Michael V. Gonzalez, Tanya Murphy, Hakon Hakonarson, Dritan Agalliu – *Genetic Risk Factors for Post-Infectious Basal Ganglia Encephalitis, A Pediatric Autoimmune Disease of the Brain Caused by Streptococcus Pyogenes and Other Infections*

8. Aditya S. Deshpande, Kenneth W. Eng, Andrea Sboner, Himisha Beltran, Olivier Elemento, Mark A. Rubin, Marcin Imielinski – *Towards the Eradication of Noisy Coverage in Cancer Exomes and Genomes*

9. Delphine Fagegaltier, Phaedra Agius, Erin G. Conlon, Isabel Hubbard, Kristy Kang, Duyang Kim, James Gregory, Nadia Propp, Giuseppe Narzisi, Samantha Fennessey, Heather Geiger, Nicolas Robine, The Target ALS Consortium, The NYGC ALS Consortium, James L. Manley, Neil Shneider, Hemali Phatnani – *Identifying Splicing Signatures in ALS and FTD*

10. Laura Ferrè, Tina Roostaei, Daniel Felsky, Hans-Ulrich Klein, Galit Alter, Philip L. De Jager – *Association of Multiple Sclerosis Risk Variants and NK Cell Markers Expression*

11. Erin Flaherty, Shijia Zhu, Alesia Antoine, Esther Cheng, Robert Sebra, Gang Fang, Kristen Brennand – *Cataloging and Manipulating NRXN1α Transcriptional Variants in hiPSC Neurons from Psychosis Patients with Heterozygous NRXN1 Deletions*

12. Heather Geiger, Evan Biederstedt, Christian Stolte – *FusionViz – Intuitive Visualization of Gene Fusion Events*

13. Zoran Gajic, Kevin Hadi, Evan Biederstedt, Kenneth Eng, Andrea Sboner, Himisha Beltran, Olivier Elemento, Mark Rubin, Marcin Imielinski – *Real-Time Cancer Driver Nomination with FishHook*

14. Kofi Gyan, Huasong Tian, Marlon Stoeckius, Peter Smibert, Shaham Beg, Juan Miguel Mosquera, Marcin Imielinski – *Transcriptional Dynamics of Intra-Tumor Heterogeneity in the Lung Epithelium Using Single-Cell RNA-seq*

15. Kevin Hadi, Marcin Imielinski – *Towards a Simulation of Rearrangements in Cancers*

16. Yun Hao, Nicholas P. Tatonetti – *Identifying G-Protein Coupled Receptors for Targeted Cancer Therapy*

17. Silva Kasela, Krista Fischer, Lili Milani, Andres Metspalu – *Translating DNA Methylation Data into Feedback for Biobank Participants*

18. Sarah Kim-Hellmuth, Matthias Bechheim, Benno Pütz, Pejman Mohammadi, Johannes Schumacher, Veit Hornung, Tuuli Lappalainen – *Genome-Wide Analysis of Transcriptional and Cytokine Response Variability in Activated Human Immune Cells*

19. Hans-Ulrich Klein, David A. Bennett, Philip L. De Jager – *Epigenome-Wide Study Uncovers Tau Pathology-Driven Changes of Chromatin Organization in the Aging Human Brain*

20. Ana Mesquita, Andreas Jenny – *Genetic Regulation of Endosomal Microautophagy in Drosophila*

21. Jacqueline Odgis, Toby Bloom, George Diaz, John Greally, Bruce Gelb, Carol Horowitz, Nicole Kelly, Michelle Ramos, Sabrina Suckiel, Eimear E. Kenny – *NYCKidSeq: Implementing Genomic Medicine in Children from Underrepresented Populations in Harlem and the Bronx*

22. Marika L. Osterbur Badhey, Alexander C. Bertalovitz, Thomas V. McDonald – *Synonymous Modifications Affect hERG Synthesis through Multiple Mechanisms and the First 51 Nucleotides of the hERG CDS Are Largely Responsible for Translation Efficiency*

23. Yiyi Ma, Charles C. White, David A. Bennet, Philip L. De Jager – *Preliminary Study of Mapping RNA Editing Events in Alzheimer's Disease*

24. Adriana Munoz, Boris Yamrom, Yoon-ha Lee, Peter Andrews, Steven Marks, Kuan-Ting Lin, Zihua Wang, Adrian R. Krainer, Bob Darnell, Michael Wigler, Ivan Iossifov – *Role of De Novo Intronic Indels in Autism*

25. Shilpa Sonti, Andrew Sproul, Ismael Santa-Maria, Damian Williams, Elan Louis, Lorraine Clark – *Functional Studies of Candidate Genes Identified in Essential Tremor Families*

26. Christian Stolte, Dimitrije Jevremovic, Weichu Shen, Nina Lapchyk, Avinash Abhyankar, Fred Criscuolo, Toby Bloom – *MetroNome -Aa Genomic Data Laboratory with Visualization Tools to Empower Researchers to Make Discoveries*

27. Arielle S. Strasser, Tin Leong, Ethylin W. Jabs, Meng Wu – *Defective Vasculogenesis in a Cohesinopathy: Roberts Syndrome Mouse Model*

28. Netha Ulahannan, Julie Behr, Xiaotong Yao, Marcin Imielinski – *Chromatin Perturbation Landscapes of Cancer Rearrangements*

29. Nicole Vo, Corry Rillahan, Thomas Buttrick, Anthony Khairallah, Wassim Elyaman, Elizabeth Bradshaw, Philip De Jager – *CD45 and CD33: The Untold Story in Alzheimer's Disease*

30. Stephane Wenric, Janina M. Jeff, Thomas Joseph, Kristina Slivinski, Muh-ching Yee, Gillian M. Belbin, Aniwaa Owusu Obeng, Stephen B. Ellis, Erwin P. Bottinger, Omri Gottesman, Matthew A. Levin, Eimear E. Kenny – *Capturing Drug Response During Surgery for Pharmacogenomic Discoveries*

31. Alexandre Yahi, Paul Hoffman, Pejman Mohammadi, Nicholas P. Tatonetti, Tuuli Lappalainen – *EdiTyper: A Scalable and Accurate Software for Genotyping of Samples from CRISPR/Cas Genome Editing*

32. Xiaotong Yao, Charalampos Xanthopoulakis, Evan Biederstedt, Kevin Hadi, Marcin Imielinski – *Graph Representation of Structural Variations in Cancer Genomes and Karyotype Reconstruction to Characterize Complex Rearrangement Events*

33. Kenny Ye, Boris Yomrom, Matthew Wroten, Ivan Iossifov, Dan Levy, Michael Wigler – *A Bias in Sibling Chromosomal Sharing in Autism Cohorts*

34. Raymond Yeh, Susan Hsiao, Mahesh Mansukhani – *Confirmation of Copy Number Variants (CNVs) Detected by Next Generation Sequencing (NGS) with Droplet Digital PCR*

35. Dejian Zhao, Deyou Zheng – *SMARTcleaner: Identify and Clean Off Target Signals in SMART ChIP-seq Libraries*