Joseph S. Takahashi, Ph.D.

Joseph Takahashi is the Loyd B. Sands Distinguished Chair in Neuroscience, an Investigator of the Howard Hughes Medical Institute, and Chair of the Department of Neuroscience at the University of Texas Southwestern Medical Center in Dallas. His research interests are the molecular mechanism of circadian clocks, neuroscience, and the genetic basis of behavior. Dr. Takahashi pioneered the use of genetics in the mouse as a tool for discovery of genes underlying neurobiology and behavior.

Monica J. Justice, Ph.D.

Monica Justice is Senior Scientist and Head of the Program in Genetics and Genome Biology at the Toronto Hospital for Sick Children, and Professor of Molecular Genetics at the University of Toronto. A pioneer in the field of mouse mutagenesis, her research aims to merge mouse modeling with clinical genetics to understand the basis for human diseases, and to use mouse models to ameliorate disease symptoms. Current efforts focus on a genetic suppressor screen in a model for Rett Syndrome to identify alternative pathways for therapeutic intervention.
The 6th Annual Genetics Symposium

NEUROGENETICS

8:30  Breakfast

9:00  WELCOME REMARKS
     Daniel J. Rader, M.D.
     Chair, Department of Genetics
     J. Larry Jameson, M.D.
     EVP, University of Pennsylvania for the Health System
     Dean, Raymond and Ruth Perelman School of Medicine

9:10  KEYNOTE ADDRESS
     Joseph S. Takahashi, Ph.D.
     Investigator, Howard Hughes Medical Institute
     Chair, Department of Neuroscience, UT Southwestern
     Genetic analysis of circadian and complex behavior

10:00  Amita Sehgal, Ph.D.
       Professor of Neuroscience
       Circadian control of rest:activity behavior

10:25  Thomas A. Jongens, Ph.D.
       Associate Professor of Genetics
       Fragile X and the insulin pathway

10:50  Coffee Break

11:10  David R. Lynch, M.D., Ph.D.
       Professor of Neurology
       Robert B. Wilson, M.D., Ph.D.
       Professor of Pathology & Laboratory Medicine
       Advances in the understanding of Friedreich Ataxia

11:45  Eric D. Marsh, M.D., Ph.D.
       Assistant Professor of Neurology, CHOP
       Manipulating ARX to elucidate network dysfunction in the early onset severe epilepsies

12:10  TOM KADESCH PRIZE PRESENTATION
       Judy I-Ting Wang
       Genetic dissection of CDKL5 disorder

12:35  Lunch and Poster Viewing

2:00  Ian D. Krantz, M.D.
     Professor of Pediatrics
     Russ P. Carstens, M.D.
     Associate Professor of Medicine
     Douglas J. Epstein, Ph.D.
     Professor of Genetics
     ESRP1 mutations link alternative splicing, auditory development, and deafness

2:35  Mariella De Biasi, Ph.D.
     Associate Professor of Psychiatry
     Functional studies of nicotine dependence

3:00  Gerard D. Schellenberg, Ph.D.
     Professor of Pathology & Laboratory Medicine
     Multiple approaches to resolving neurodegenerative disease genetics

3:25  Alice S. Chen-Plotkin, M.D.
     Assistant Professor of Neurology
     What to do after the GWAS: TMEM106B in frontotemporal dementia

3:50  Coffee Break and Poster Viewing

4:10  POSTER AWARD ANNOUNCEMENTS

4:15  KEYNOTE ADDRESS
     Monica J. Justice, Ph.D.
     Senior Scientist and Head, Program in Genetics and Genome Biology,
     Toronto Hospital for Sick Children
     A mouse genetic suppressor screen points to metabolic defects in Rett Syndrome

5:10  Conclusion
28  **Yingtao Zhao** (Zhou Lab)
    Locus- and cell type-specific epigenomic reconfiguration during cellular differentiation

29  **Hyun Mi Kang** (Susztak Lab)
    Defective fatty acid oxidation in renal tubular epithelial cells plays a key role in kidney fibrosis development

30  **Jenny Russ** (Lee Lab)
    C9orf72 promoter hypermethylation is a clinical disease modifier in frontotemporal degeneration

31  **Amanda Zacharias** (Murray Lab)
    Transmitotic persistence of nuclear beta-catenin diversifies gene expression in C. elegans embryos

32  **Robert Bauer** (Rader Lab)
    Tribbles1 regulates hepatic lipogenesis in mice through post-translational regulation of C/EBPα

33  **Jonathan Mitchell** (Grant & Zemel Labs)
    Sex and maturation unmask the role of genome wide association study implicated adult bone mass loci in childhood and adolescence

34  **Rebecca Ganetzky** (Sondheimer Lab)
    Loss of function mutations in EGFR cause a novel progeroid syndrome

35  **Varun Aggarwala** (Voight Lab)
    A Bayesian framework to model the rate of single nucleotide polymorphism in humans

36  **Kriti Gaur** (Susztak Lab)
    Role of DNA methylation in Chronic Kidney Disease

37  **Alessandra Chesi** (Grant Lab)
    A trans-ethnic GWAS identifies gender specific loci influencing pediatric aBMD and BMC at the distal radius

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**The 6th Annual Genetics Symposium**

**POSTER SESSION**

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**GRADUATE STUDENTS**

01  **Ian M. Silverman** (Gregory Lab)
    Insights into primate mRNA regulation and conservation from transcriptome-wide RNA structure analysis

02  **Katie Wood** (Zhou Lab)
    Interpreting DNA methylation in the brain: The role of Methyl-CpG binding domain protein 2 (Mbd2)

03  **Reina Aoki** (Kaestner Lab)
    Foxl1 defines the intestinal stem cell niche

04  **Ellie Weisz** (Jongens Lab)
    Assessment of metabolism in a drosophila model of Fragile X Syndrome

05  **Chris Hsiung** (Raj & Blobel Labs)
    Structure and regulation of the mitotic genome

06  **Julia Kieckhaefer** (Kaestner Lab)
    The Foxa transcription factors sustain murine intestinal health by regulating Crohn’s Disease risk loci Igpm1 and Fut2
07 **Alex Rohacek** (Epstein Lab)
*Investigating roles for Esrps in inner ear development*

08 **Kate Palozola** (Zaret Lab)
*Post-mitotic transcription reactivation: insights into maintaining cell identity*

09 **Mike Gallagher** (Chen-Plotkin Lab)
*From GWAS to functional variants at the TMEM106B locus: implications for neurodegeneration*

10 **Rob Plasschaert** (Bartolomei Lab)
*CTCF’s role in the imprinted of Grb10 expression during neuronal commitment*

11 **Sumeet Khetarpal** (Rader Lab)
*Coronary artery disease-protective A43T variant in APOC3 alters circulating ApoC-III levels in vivo*

12 **Christine Dubowy** (Sehgal Lab)
*Genetic and molecular characterization of a drosophila mutant with reduced sleep rebound*

13 **Rose Li** (Hakonarson Lab)
*The genomic landscape of pediatric autoimmune diseases*

14 **Jennifer Myers** (Bartolomei Lab)
*Exploring the role of Tet1 in genomic imprinting*

15 **Lauren Brady** (Koumenis Lab)
*RNA sequencing of hypoxic tumors reveals alternative splicing as a way to regulate microenvironment*

16 **Yi-An Ko** (Susztak Lab)
*Functional annotation of SNPs associated with Chronic Kidney Disease*

17 **Suzanne Shapira** (Seale Lab)
*Regulation of brown adipogenesis by the mSWI/SNF chromatin remodeling complex*

18 **Samantha Falk** (Black Lab)
*CENP-C stabilizes CENP-A nucleosomes at the centromere*

19 **Seth Johnson** (Roegiers Lab)
*Numb diverts notch from recycling to late endosomes in drosophila sensory organ precursor*

20 **Joyce Lee** (Wellen Lab)
*Acetyl-CoA regulation of histone acetylation and gene expression in cancer cell*

21 **Aaron Leichty** (Poethig Lab)
*Developing a model system for studying the evolution of developmental timing and leaf morphogenesis*

22 **Lee Vandivier** (Gregory Lab)
*DICER-LIKE processing of protein-coding mRNAs in arabidopsis thaliana*

23 **Jeremy Grevet** (Blobel Lab)
*Mechanisms and therapeutic potential of forced chromatin looping for the treatment of Sickle Cell Anemia*

24 **Allison Jamieson-Lucy** (Mullins Lab)
*The Balbiani body proteome in zebrafish*

**POSTDOCTORAL STUDENTS**

25 **Elizabeth Bhoj** (Zackai Lab)
*Evolutionarily-conserved imprinting between mouse and human orthologs identifies 17 novel candidate genes for human imprinting disorders*

26 **Janine Lamonica** (Zhou Lab)
*Exploring novel treatment strategies for Rett Syndrome*

27 **Yao Yao** (Epstein Lab)
*A transcription factor collective governs Sonic hedgehog expression in the evolution of a vertebrate brain-signaling center*