

# Human and Mammalian Genetics and Genomics: The 59<sup>th</sup> McKusick Short Course

July 16 – July 27, 2018

## WEBCAST EVENT SCHEDULE

For more information, please contact Erin McDevitt; [erin.mcdevitt@jax.org](mailto:erin.mcdevitt@jax.org)

**NOTE: All times listed on this schedule are in Eastern Standard Time**

---

---

### **Monday, July 16**

8:20-8:30am	<b>Course Welcome</b> Edison Liu, M.D., <i>The Jackson Laboratory</i>
8:30-9:25am	<b>History of Human Genetics</b> David Valle, M.D., <i>Johns Hopkins University School of Medicine</i>
9:25–10:15 am	<b>Opportunities for Using Mouse Models in the 21<sup>st</sup> Century</b> Gary Churchill, Ph.D., <i>The Jackson Laboratory</i>
10:45-11:40am	<b>Chromosome Structure and Function</b> Jeanne Lawrence, Ph.D., <i>University of Massachusetts Medical School</i>
11:40-12:30pm	<b>Recombination</b> Kenneth Paigen, Ph.D, <i>The Jackson Laboratory</i>
1:30-2:30pm	<b>Mice as Models for Human Diseases</b> Greg Cox, Ph.D., <i>The Jackson Laboratory</i>
2:30-3:30pm	<b>Molecular Genetics</b> John A. Phillips, III., M.D., <i>Vanderbilt University School of Medicine</i>

### Evening Lecture

7:00-8:15pm      **Thomas H. Roderick Memorial Lecture**  
**Title TBD**  
Richard A. Gibbs, Ph.D., *Baylor College of Medicine*

### Tuesday, July 17

8:30-9:25am      **Epigenetics**  
Hans Bjornsson, M.D., Ph.D., *Johns Hopkins University School of Medicine*

9:25-10:15am      **Transcriptional Regulation of Gene Expression**  
Andrew S. McCallion, Ph.D., *Johns Hopkins University School of Medicine*

10:45-11:40am      **Single Cell RNA-Seq**  
Loyal Goff, Ph.D., *Johns Hopkins University School of Medicine*

11:40-12:30pm      **3D Genome Reconfiguration in Mammalian Brain Development and Disease**  
Jennifer Phillips-Cremins, Ph.D., *University of Pennsylvania*

1:30-2:30pm      **Introduction to MGI/OMIM**  
MeiYee Law, Ph.D., *The Jackson Laboratory* and  
Ada Hamosh, M.D., *The Johns Hopkins University School of Medicine*

### Evening Lecture (Not Available Live)

7:30-9:00pm      **The Nature of Mutation**  
Stylianos Antonarakis, M.D., DSc, *University of Geneva Medical School*

### Wednesday, July 18

8:30-9:25am      **Massively Parallel Sequencing: Technologies and Applications**  
Elaine Mardis, Ph.D., *Nationwide Children's Hospital*

9:25-10:15am      **CNVs in Human Disease**  
Charles Lee, Ph.D., *The Jackson Laboratory for Genomic Medicine*

10:45-11:40am      **Immune Control of Tissue Regeneration**  
Nadia Rosenthal, Ph.D., *The Jackson Laboratory*

11:40-12:30pm      **The Humanized Mouse in Biomedical Research**  
Lenny Shultz, Ph.D., *The Jackson Laboratory*

**Evening Lecture (Not Available Live)**

7:30-9:00pm      **TBD**

**Thursday, July 19**

8:30-9:40am      **A Primer on Quantitative Systems Biology**  
Aravinda Chakravarti, Ph.D., *Johns Hopkins University School of Medicine*

9:40-10:15am      **Methods for Genetic Association Studies in Humans**  
Goncalo Abecasis, Ph.D., *The University of Michigan*

10:45-11:40am      **Mapping Complex Traits in Mice**  
Gary Churchill, Ph.D., *The Jackson Laboratory*

11:40-12:30pm      **Transcriptional Responses to Hypoxia and Ischemia**  
Gregg Semenza, M.D., Ph.D., *Johns Hopkins University School of Medicine*

1:30- 2:30pm      **Inborn Errors**  
David Valle, M.D., *Johns Hopkins University School of Medicine*

**Evening Lecture (Not Available Live)**

7:30-9:00pm      **Title TBD**  
Connie Cepko, Ph.D., *Harvard University*

**Friday, July 20**

8:30-9:25am      **Genetic Epidemiology of Breast and Ovarian Cancer**  
Larry Brody, Ph.D., *National Human Genome Research Institute*

9:25-10:15am      **Cancer Biology Exposed by Large-Scale Sequencing**  
Victor Velculescu, M.D., Ph.D., *Johns Hopkins University School of Medicine*

10:45-11:40am      **Inborn Errors of the Epigenetic Apparatus: Genetics of Chromatin Readers and Writers**  
Hans Bjornsson, M.D., Ph.D., *Johns Hopkins University School of Medicine*

11:40- 12:30pm      **Gene Regulation and ENCODE**  
David Bodine, Ph.D., *National Human Genome Research Institute*

## Evening Lecture

7:30-9:00pm

### **Chen K. Chai Lecture**

**Title TBD**

Jack Szostak, Ph.D., *Harvard Medical School and Massachusetts General Hospital*

## Saturday, July 21

8:30-9:25am

### **Genomics and the Inference of Human Population History**

Andrew Clark, Ph.D., *Cornell University*

9:25-10:15am

### **Interpretation of Genetic Variants using Massive-Scale Reference Data Sets**

Daniel MacArthur, Ph.D., *Massachusetts General Hospital*

10:45-11:40am

### **H3Africa**

Charles Rotimi, Ph.D., *National Human Genome Research Institute*

11:40-12:30pm

### **African Genomics: Implications for Studies of Human Evolution and Disease**

Sarah Tishkoff, Ph.D., *University of Pennsylvania*

## Sunday, July 22

No Scheduled Course Events

## **Monday, July 23**

- 8:30-9:25am      **The Major Histocompatibility Complex in Health and Disease**  
Derry Roopenian, Ph.D., *The Jackson Laboratory* –
- 9:25-10:15 am      **Genetics of Autoimmunity**  
David Serreze, Ph.D., *The Jackson Laboratory*
- 10:45-11:40am      **Genetics of Infectious Diseases**  
Steven M. Holland, M.D., *National Institutes of Health*
- 11:40 – 12:30pm      **Innate Immunity and IBD**  
Judy Cho, M.D., *Icahn School of Medicine at Mount Sinai*

### **Evening Lecture (Not Available Live)**

- 7:30-9:00pm      **TGF Beta Signaling and the Marfan Syndrome**  
Harry C. Dietz, M.D., *Johns Hopkins University School of Medicine*

## **Tuesday, July 24**

- 8:30-9:25am      **The Genetics of Skin Diseases and Psoriasis**  
Anne Bowcock, Ph.D., *Icahn School of Medicine at Mount Sinai*
- 9:25-10:15 am      **Modeling Behavior in the Laboratory Mouse**  
Vivek Kumar, Ph.D., *The Jackson Laboratory*
- 10:45-11:40am      **Genetics of Schizophrenia**  
Benjamin Neale, Ph.D., *Massachusetts General Hospital*
- 11:40-12:30      **Human Microbiome in Health and Disease**  
Julia Oh, Ph.D., *The Jackson Laboratory for Genomic Medicine*

### **Evening Lecture (Not Available Live)**

- 7:30-9:00pm      **Title TBD**  
Peter Donnelly, FRS, Wellcome Trust Centre for Human Genetics

## **Wednesday, July 25**

- 8:30-9:25am            **Genetic and Pharmacologic Modifiers of CF**  
Garry R. Cutting, M.D., *Johns Hopkins University School of Medicine*
- 9:25-10:15am        **The Genetics and Clinical Phenotypes of Inherited Skeletal Dysplasias**  
Julie Hoover-Fong, M.D., Ph.D., *Johns Hopkins University School of Medicine*
- 10:45-11:40am      **Personalized Medicine**  
David Ledbetter, Ph.D., *Geisinger Health System*
- 11:40-12:30pm      **Pharmacogenomics: Development, Science and Translation**  
Richard Weinshilboum, M.D., *Mayo Clinic*

### **Evening Lecture (Not Available Live)**

- 7:30-9:00pm        **Senior Lecturer**  
**Title TBD**  
Leslie Biesecker, M.D., *National Human Genome Research Institute*

## **Thursday, July 26**

- 8:30-9:25 am        **Proteomics**  
Steven Gygi, Ph.D., *Harvard Medical School*
- 9:25-10:25am      **Prenatal Diagnosis and Screening**  
Ignatia B. Van den Veyver, M.D., *Baylor College of Medicine*
- 10:45-11:40am     **Biomedical Ethics in Genomics**  
Debra Mathews, Ph.D., M.A., *Johns Hopkins Berman Institute of Bioethics*
- 11:40-12:30pm     **DNA Forensics**  
Phil Reilly, M.D., J.D., *Third Rock Ventures*

### **Evening Lecture (Not Available Live)**

- 7:30-9:00pm        **ELSI Case Studies and Editing the Human Genome**  
John A. Phillips, III., M.D., *Vanderbilt University School of Medicine*,  
Debra Mathews, Ph.D., M.A., *Johns Hopkins Berman Institute of Bioethics*,  
Ada Hamosh, M.D., M.P.H., *Johns Hopkins University School of Medicine*,

## **Friday, July 27**

8:30-9:25am

### **Metabolomics and Metabolic Flux Analysis**

Ralph DeBeradinis, M.D., Ph.D., *UT Southwestern*

9:25-10:15am

### **Treatment of Genetic Disease**

David Valle, M.D., *Johns Hopkins University School of Medicine*

10:45-11:40am

### **IPS Cell Technology, Gene Editing and Disease Research**

Rudolf Jaenisch, M.D., *Whitehead Institute*

11:40-12:30pm

### **Course Wrap Up**

#### **Title TBD**

Leslie Biesecker, M.D., *National Human Genome Research Institute*

**End of the course**