

# **Human and Mammalian Genetics and Genomics: The 57<sup>th</sup> McKusick Short Course**

July 17 – July 29, 2016

## **EVENT SCHEDULE**

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

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### **Sunday, July 17**

*Location: The Jackson Laboratory, 600 Main Street*

5:00-7:00pm      **Welcome BBQ and Registration**

### **Monday, July 18**

*Location: The Jackson Laboratory, 600 Main Street*

7:30-8:00am      **Late Registration and Continental Breakfast**

8:00-8:20am      **Overview of Course Evaluations**

8:20-8:30am      **Course Welcome**  
*Edison Liu, M.D., The Jackson Laboratory*

8:30-9:25am      **History of Human Genetics**  
*David Valle, M.D., Johns Hopkins University School of Medicine*

9:25-10:15 am      **The Obligate Symbiosis Between Human and Mouse Genetics**  
*Kenneth Paigen, Ph.D., The Jackson Laboratory*

10:15-10:45am      **Break** - Roscoe's Dining Area

10:45-11:40am      **Chromosome Structure and Function**  
*Jeanne Lawrence, Ph.D., University of Massachusetts Medical School*

- 11:40-12:30pm      **3D Genome Organization Frames Chromatin Topology for Transcription Regulation**  
Yijun Ruan, Ph.D., *The Jackson Laboratory for Genomic Medicine*
- 12:30-1:30pm      **Lunch** - Roscoe's Dining Area
- 1:30-3:00pm      **Mendelian Inheritance in Humans**  
Clair Francomano, M.D., *Harvey Institute for Human Genetics*
- 3:00-4:00pm      **Mice as Models for Human Diseases**  
Greg Cox, Ph.D., *The Jackson Laboratory*

### **Evening Lecture**

*Location: The Jackson Laboratory, 600 Main Street*

- 6:00-7:00pm      **Thomas H. Roderick Lecture Reception** – Roscoe's Dining Area
- 7:00-7:20pm      **Introduction and Welcome**
- 7:20-8:15pm      **Thomas H. Roderick Memorial Lecture**  
**Genotype to Phenotype—A Difficult Problem**  
Gerry Fink, Ph.D., *Massachusetts Institute of Technology*

### **Tuesday, July 19**

*Location: The Jackson Laboratory, 600 Main Street*

- 7:45-8:25am      **Continental Breakfast** - Roscoe's Dining Area
- 8:30-9:25am      **Massively Parallel Sequencing: Technologies and Applications**  
Elaine Mardis, Ph.D., *The Genome Institute Washington University*
- 9:25-10:15am      **Transcriptional Regulation of Gene Expression**  
Andrew S. McCallion, Ph.D., *Johns Hopkins University School of Medicine*
- 10:15-10:45am      **Break** - Roscoe's Dining Area  
**(GROUP PHOTO)**
- 10:45-11:40am      **Post-transcriptional Control of Gene Expression**  
Harry C. Dietz, M.D., *Johns Hopkins University School of Medicine*
- 11:40-12:30pm      **CRISPR/Cas9-mediated Genome Editing**  
Haoyi Wang, Ph.D., *The Jackson Laboratory*
- 12:30-1:30pm      **Lunch** - Roscoe's Dining Area

- 1:30-2:30pm      **Molecular Genetics**  
John A. Phillips, III, M.D., *Vanderbilt University School of Medicine*
- 2:30-3:30pm      **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**

*Location: The Bar Harbor Club*

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

**Wednesday, July 20**

*Location: The Jackson Laboratory, 600 Main Street*

- 7:45-8:30am      **Continental Breakfast - Roscoe's Dining Area**
- 8:30-9:25am      **Genetics of Chromatin Readers and Writers**  
Hans Bjornsson, M.D., Ph.D., *Johns Hopkins University School of Medicine*
- 9:25-10:15am      **Epigenetics and DNA Methylation: Regulation and Reprogramming in Mammalian Development**  
Marisa Bartolomei, Ph.D., *University of Pennsylvania*
- 10:15-10:45am      **Break - Roscoe's Dining Area**
- 10:45-11:40am      **Immune Control of Tissue Regeneration**  
Nadia Rosenthal, 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*
- 12:30-1:30pm      **Lunch - Roscoe's Dining Area**

**ROTATING WORKSHOPS – 1 Hour Each**

**GROUP A:**

- 1:30-2:30pm      **RNA Sequencing Workshop**  
Steve Munger, Ph.D., *The Jackson Laboratory*

2:30-3:30pm                      **Exome Sequencing Workshop**  
Nara Sobreira, M.D., Ph.D., *Johns Hopkins University School of Medicine*

**GROUP B:**

1:30- 2:30pm                      **Exome Sequencing Workshop**  
Nara Sobreira, M.D., Ph.D., *Johns Hopkins University School of Medicine*

2:30-3:30pm                      **RNA Sequencing Workshop**  
Steve Munger, Ph.D., *The Jackson Laboratory*

**Poster Session**

*Location: The Bar Harbor Club*

5:00-7:00pm                      **Poster Session**

**Evening Lecture**

*Location: The Bar Harbor Club*

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

**Thursday, July 21**

*Location: The Jackson Laboratory, 600 Main Street*

7:45-8:25am                      **Continental Breakfast - Roscoe's Dining Area**

8:30-9:40am                      **The Genetics of Age Related Macular Degeneration**  
Goncalo Abecasis, Ph.D., *The University of Michigan*

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

10:15-10:45am                      **Break** - *Roscoe's Dining Area*

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

11:40-12:30pm                      **A primer on Quantative Systems Biology**  
Aravinda Chakravarti, Ph.D., *Johns Hopkins University School of Medicine*

12:30-1:30pm                      **Lunch** - *Roscoe's Dining Area*

1:30-3:30pm                      **Human Genomics Database Resources**  
Melissa Landrum, Ph.D., *NCBI/NLM/NIH*

**Evening Lecture**

*Location: The Bar Harbor Club*

7:30-9:00pm                      **Global Analysis of Genetic Interaction Networks in Yeast**  
Charles Boone, Ph.D., *University of Toronto*

**Friday, July 22**

*Location: The Jackson Laboratory, 600 Main Street*

7:45-8:25am                      **Continental Breakfast** - *Roscoe's Dining Area*

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*

10:15-10:45am                      **Break** - *Roscoe's Dining Area*

10:45-11:40am                      **Interpretation of Genetic Variants using Massive-Scale Reference Data Sets**  
Daniel MacArthur, Ph.D., *Massachusetts General Hospital*

11:40- 12:30pm                      **Genomic Mechanisms of Gene Regulation and Chromatin Organization**  
Frank Pugh, Ph.D., *Penn State University*

12:30-1:30pm                      **Lunch** - *Roscoe's Dining Area*

1:30 – 3:30pm                      **Mouse Genomic Resources Workshop**  
Carol Bult, Ph.D., *The Jackson Laboratory*

*Location: The Bar Harbor Club*

5:00-7:00pm                      **Lobster Dinner**

**Evening Lecture**

*Location: The Bar Harbor Club*

7:30-9:00pm

**Chen K. Chai Lecture**

**Telomerase and the Consequences of Telomere Dysfunction**

Carol Greider, Ph.D., *Johns Hopkins University School of Medicine*

**Saturday, July 23**

*Location: The Jackson Laboratory, 600 Main Street*

7:45- 8:25am

**Continental Breakfast** - *Roscoe's Dining Area*

8:30-9:25am

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

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

9:25-10:15am

**Genetic Recombination: Why, how and mapping**

Ken Paigen, Ph.D., *The Jackson Laboratory*

10:15-10:45am

**Break** - *Roscoe's Dining Area*

10:45-11:40am

**CNVs in Human Disease**

Charles Lee, Ph.D., *The Jackson Laboratory for Genomic Medicine*

11:40-12:30pm

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

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

12:30pm

Boxed Lunches

**Sunday, July 24**

No Scheduled Course Events

## **Monday, July 25**

*Location: The Jackson Laboratory, 600 Main Street*

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|-----------------|--|
| 7:45-8:20am     | <b>Continental Breakfast</b> - <i>Roscoe's Dining Area</i>   |
| 8:30-9:25am     | <b>The Major Histocompatibility Complex in Health and Disease</b><br>Derry Roopenian, Ph.D., <i>The Jackson Laboratory</i>     |
| 9:25-10:15 am   | <b>Genetics of Autoimmunity</b><br>David Serreze, Ph.D., <i>The Jackson Laboratory</i>   |
| 10:15-10:45am   | <b>Break</b> - <i>Roscoe's Dining Area</i>   |
| 10:45-11:40am   | <b>Genetics of Infectious Diseases</b><br>Steven M. Holland, M.D., <i>National Institutes of Health</i>                        |
| 11:40 – 12:30pm | <b>Innate Immunity and IBD</b><br>Judy Cho, M.D., <i>Icahn School of Medicine at Mount Sinai</i>                               |
| 12:30-1:30pm    | <b>Lunch</b> - <i>Roscoe's Dining Area</i>   |
| 1:30- 2:30pm    | <b>Inborn Errors</b><br>David Valle, M.D., <i>Johns Hopkins University School of Medicine</i>                                  |
| 2:30-3:30pm     | <b>Workshop: Methods of Molecular Genetics</b><br>John A. Phillips, III, M.D., <i>Vanderbilt University School of Medicine</i> |

### **Evening Lecture**

*Location: The Bar Harbor Club*

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| 7:30-9:00pm | <b>TBD</b> |
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## **Tuesday, July 26**

*Location: The Jackson Laboratory, 600 Main Street*

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| 7:45-8:25am   | <b>Continental Breakfast</b> - <i>Roscoe's Dining Area</i>   |
| 8:30-9:25am   | <b>Humanized Mouse in Biomedical Research</b><br>Lenny Shultz, Ph.D., <i>The Jackson Laboratory</i>      |
| 9:25-10:15 am | <b>Modeling Behavior in the Laboratory Mouse</b><br>Elissa Chesler, Ph.D., <i>The Jackson Laboratory</i> |

- 10:15-10:45am      **Break** - *Roscoe's Dining Area*
- 10:45-11:40am      **Genetics of Schizophrenia**  
Dimitri Avramopoulos, Ph.D., *Johns Hopkins University School of Medicine*
- 11:40-12:30      **The Real Poop on the Microbiome**  
George Weinstock, Ph.D., *The Jackson Laboratory for Genomic Medicine*
- 12:30-1:30pm      **Lunch** - *Roscoe's Dining Area*
- 1:30- 3:30pm      **UCSC Genome Browser**  
Robert Kuhn, Ph.D., *University of California, Santa Cruz*

### **Evening Lecture**

*Location: The Bar Harbor Club*

- 7:30-9:00pm      **Genetics, Environment and Coronary Artery Disease**  
Sekar Kathiresan, M.D., *Massachusetts General Hospital, Broad Institute, Harvard Medical School*

### **Wednesday, July 27**

*Location: The Jackson Laboratory, 600 Main Street*

- 7:45-8:25am      **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25am      **Genetic Modifiers of CF**  
Garry R. Cutting, M.D., *Johns Hopkins University School of Medicine*
- 9:25-10:15am      **Molecular Bases of Malformation Syndromes**  
David FitzPatrick, M.D., FRCD(Edin), *Medical Research Council*
- 10:15-10:45am      **Break** - *Roscoe's Dining Area*
- 10:45-11:40am      **Precision Medicine Initiative**  
David Ledbetter, Ph.D., FACMG, *Geisinger Health System*
- 11:40-12:30pm      **Pharmacogenomics to Pharmaco-omics: Genomics and Drug Response**  
Richard Weinshilboum, M.D., *Mayo Clinic*
- 12:30-1:30pm      **Lunch** - *Roscoe's Dining Area*

*Location: The Bar Harbor Club*

- 12:30-2:00pm      **Lunch** - *La Bella Vita Ristorante*



***(Selected Participants and Invited Guests Only)***

**Human Genetics Clinic Presentations**

***(Selected Participants and Invited Guests Only)***

Ada Hamosh, M.D., M.P.H., *Johns Hopkins University School of Medicine*

2:00-4:00pm

**Human Genetics Clinic Presentations**

***(Open to All Attendees)***

Ada Hamosh, M.D., M.P.H., *Johns Hopkins University School of Medicine*

**Evening Lecture**

*Location: The Bar Harbor Club*

7:30-9:00pm

**Senior Lecturer**

**Decoding the *C. elegans* Genome**

Robert Waterston, M.D., Ph.D., *University of Washington Genome Sciences*

**Thursday, July 28**

*Location: The Jackson Laboratory, 600 Main Street*

7:45-8:25am

**Continental Breakfast** - *Roscoe's Dining Area*

8:30-9:25 am

**Proteomics**

Steve Gygi, Ph.D., *Harvard Medical School*

9:25-10:25am

**Metabolomics and Metabolic Flux Analysis**

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

10:25-10:45am

**Break** - *Roscoe's Dining Area*

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*

12:30-1:30pm

**Lunch** - *Roscoe's Dining Area*

1:30-3:30pm

**Mouse Genetics Clinic**

Greg Cox, Ph.D., *The Jackson Laboratory*

### **Evening Lecture**

*Location: The Bar Harbor Club*

7:30-9:00pm                      **ELSI Case Studies**

### **Friday, July 29**

*Location: The Jackson Laboratory, 600 Main Street*

7:45-8:25am                      **Continental Breakfast - Roscoe's Dining Area**

8:30-9:25am                      **Prenatal Diagnosis and Screening**  
*Ignatia B. Van den Veyver, M.D., Baylor College of Medicine*

9:25-10:15am                      **Treatment of Genetic Disease**  
*David Valle, M.D., Johns Hopkins University School of Medicine*

10:25-10:45am                      **Break - Roscoe's Dining Area**

10:45-11:40am                      **Epigenetics, Stem Cells and Disease Research**  
*Rudolf Jaenisch, Ph.D., MIT*

11:40-12:30pm                      **Course Wrap Up**  
*Robert Waterston, M.D., Ph.D., University of Washington Genome Sciences*

12:30-1:30pm                      **Lunch - Roscoe's Dining Area**

**End of the course**