



Human and Mammalian Genetics and Genomics: The 58th McKusick Short Course

July 16 – July 28, 2017

EVENT SCHEDULE

For more information, please contact Erin McDevitt; erin.mcdevitt@jax.org

Sunday, July 16

Location: The Jackson Laboratory, 600 Main Street

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

Monday, July 17

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 **Opportunities for Using Mouse Models in the 21st Century**
Gary Churchill, 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 **Recombination**
Kenneth Paigen, Ph.D, *The Jackson Laboratory*

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

1:30-2:30pm **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**
Advancing NHLBI Discovery Science for Public Health Impact
Gary Gibbons, M.D., *National Heart, Lung and Blood Institute*

Tuesday, July 18

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., *Nationwide Children's Hospital*

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 **Long noncoding RNAs: Mechanisms and insights into RNA Biology**
Loyal Goff, Ph.D., *Johns Hopkins University School of Medicine*

11:40-12:30pm **Epigenome Folding Principles During Lineage Commitment and Reprogramming**
Jennifer Phillips-Cremins, Ph.D., *University of Pennsylvania*

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 Regency

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

Wednesday, July 19

Location: The Jackson Laboratory, 600 Main Street

7:45-8:30am **Continental Breakfast - Roscoe's Dining Area**

8:30-9:25am **Inborn Errors of the Epigenetic Apparatus: Genetics of Chromatin Readers and Writers**
Hans Bjornsson, M.D., Ph.D., *Johns Hopkins University School of Medicine*

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

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 Regency

5:00-7:00pm

Poster Session

Evening Lecture

Location: The Bar Harbor Regency

7:30-9:00pm

Genetic Risk, Adherence to a Healthy Lifestyle, and Coronary Disease

Sekar Kathiresan, M.D., *Massachusetts General Hospital, Broad Institute, Harvard Medical School*

Thursday, July 20

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

Mapping Complex Traits in Mice

Gary Churchill, Ph.D., *The Jackson Laboratory*

11:40-12:30pm

A Primer on Quantitative Systems Biology

Aravinda Chakravarti, Ph.D., *Johns Hopkins University School of Medicine*

12:30-1:30pm

Lunch - Roscoe's Dining Area

1:30- 2:30pm

Inborn Errors

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

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

Evening Lecture

Location: The Bar Harbor Regency

7:30-9:00pm **Systems Biology Through Single Cells**
Dana Pe'er, Ph.D., *Memorial Sloan Kettering Cancer Center*

Friday, July 21

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:25am **Interpretation of Genetic Variants using Massive-Scale Reference Data Sets**
Daniel MacArthur, Ph.D., *Massachusetts General Hospital*

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 **Epigenetics**
Danny Reinberg, Ph.D., *NYU School of Medicine*

11:40- 12:30pm **Gene Regulation and ENCODE**
Ross Hardison, 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*

Evening Lecture

Location: The Jackson Laboratory, 600 Main Street

7:30-9:00pm **Chen K. Chai Lecture**
Why do we (still) want to be scientists?
Harold Varmus, M.D., *Weill Cornell Medicine*

Saturday, July 22

Location: The Jackson Laboratory, 600 Main Street

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| 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., <i>Cornell University</i> |
| 9:25-10:15am | Drosophila as a Model for Human Genetic Disease
Andrew Clark, Ph.D., <i>Cornell University</i> |
| 10:15-10:45am | Break - <i>Roscoe's Dining Area</i> |
| 10:45-11:40am | Genetic Epidemiology of Breast and Ovarian Cancer
Larry Brody, Ph.D., <i>National Human Genome Research Institute</i> |
| 11:40-12:30pm | African Genomics: Implications for Studies of Human Evolution and Disease
Sarah Tishkoff, Ph.D., <i>University of Pennsylvania</i> |
| 12:30pm | Boxed Lunches |

Sunday, July 23

No Scheduled Course Events

Monday, July 24

Location: The Jackson Laboratory, 600 Main Street

- 7:45-8:20am **Continental Breakfast** - *Roscoe's Dining Area*
- 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*
- 10:15-10:45am **Break** - *Roscoe's Dining Area*
- 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*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*
- 1:30- 3:30pm **ClinVar Workshop**
Heidi Rehm, Ph.D., *BWH and Harvard Medical School*

Evening Lecture

Location: The Bar Harbor Regency

- 7:30-9:00pm **The Resolution of Phenotypic, Genetic & Cellular Heterogeneity in Hirschsprung Disease**
Aravinda Chakravarti, Ph.D., *Johns Hopkins University School of Medicine*

Tuesday, July 25

Location: The Jackson Laboratory, 600 Main Street

- 7:45-8:25am **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25am **DNA Forensics**
Phil Reilly, M.D., J.D., *Third Rock Ventures*
- 9:25-10:15 am **Modeling Behavior in the Laboratory Mouse**
Vivek Kumar, Ph.D., *The Jackson Laboratory*

- 10:15-10:45am **Break** - *Roscoe's Dining Area*
- 10:45-11:40am **Genetics of Schizophrenia**
Dimitri Avramopoulos, M.D., Ph.D., *Johns Hopkins University School of Medicine*
- 11:40-12:30 **Human Microbiome in Health and Disease**
Elizabeth Grice, Ph.D., *University of Pennsylvania*
- 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*

Location: The Bar Harbor Regency

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

Evening Lecture

Location: The Bar Harbor Regency

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

Wednesday, July 26

Location: The Jackson Laboratory, 600 Main Street

- 7:45-8:25am **Continental Breakfast** - *Roscoe's Dining Area*
- 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 Centers for Mendelian Genomics: Lessons Learned in the First Five Years**
Jennifer Posey, M.D., Ph.D., *Baylor College of Medicine*
- 10:15-10:45am **Break** - *Roscoe's Dining Area*
- 10:45-11:40am **Title TBD**
Alan Shuldiner, M.D., *University of Maryland School of Medicine*
- 11:40-12:30pm **Pharmacogenomics: Development, Science and Translation**
Richard Weinshilboum, M.D., *Mayo Clinic*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*

1:00-2:00pm **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 Regency

7:30-9:00pm **Senior Lecturer**
Epigenetic Inheritance Using Mouse Models
Anne Ferguson-Smith, Ph.D., *University of Cambridge*

Thursday, July 27

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:25 am **Proteomics**
Edward Huttlin, 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 **The Humanized Mouse in Biomedical Research**
Lenny Shultz, Ph.D., *The Jackson Laboratory*

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 Regency

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*,
Jeffrey Kahn, Ph.D., M.P.H., *Johns Hopkins Berman Institute of Bioethics*
Rudolf Jaenisch, M.D., *Whitehead Institute*

Friday, July 28

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

IPS Cell Technology, Gene Editing and Disease Research

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

11:40-12:30pm

Course Wrap Up

Anne Ferguson-Smith, Ph.D., *University of Cambridge*

12:30-1:30pm

Lunch - *Roscoe's Dining Area*

End of the course