



Human and Mammalian Genetics and Genomics: The 60th McKusick Short Course

July 15 – July 26, 2019

The Jackson Laboratory, Bar Harbor, Maine

EVENT SCHEDULE

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

Monday, July 15

Location: The Jackson Laboratory, 600 Main Street

8:00-9:00am	Registration and Continental Breakfast
9:00-9:15am	Overview of Course Evaluations
9:15-9:30am	Course Welcome <i>Edison Liu, M.D., The Jackson Laboratory</i>
9:30-10:25am	History of Human Genetics <i>David Valle, M.D., Johns Hopkins University School of Medicine</i>
10:25-11:00am	Break
11:00–11:55am	Opportunities for Using Mouse Models in the 21st Century <i>Gary Churchill, Ph.D., The Jackson Laboratory</i>
11:55-12:50pm	Chromosome Structure and Function <i>Jeanne Lawrence, Ph.D., University of Massachusetts Medical School</i>

- 12:50-2:00pm **Lunch** - *Roscoe's Dining Area*
- 2:00-2:55pm **Genetic Recombination: What it is, Why You Should Care, and How it Works**
Kenneth Paigen, Ph.D., *The Jackson Laboratory*
- 2:55-3:50pm **Mice as Models for Human Diseases**
Greg Cox, Ph.D., *The Jackson Laboratory*
- 3:50-4:15pm **Break**
- 4:15-5:10pm **Molecular Genetics**
John A. Phillips, III., M.D., *Vanderbilt University School of Medicine*
- 5:10-6:00pm **Social Hour** - *Roscoe's Dining Area*
- 6:00-7:00pm **Welcome Dinner** - *Roscoe's Dining Area*

Tuesday, July 16

Location: The Jackson Laboratory, 600 Main Street

- 7:45-8:25am **Continental Breakfast** - *Roscoe's Dining Area*
- 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*
- 10:15-10:45am **Break**
(GROUP PHOTO)
- 10:45-11:40am **Single Cell Analysis in Normal and Disease Tissue**
Loyal Goff, Ph.D., *Johns Hopkins University School of Medicine*
- 11:40-12:30pm **Three-Dimensional Folding of Mammalian Genomes**
Jennifer Phillips-Cremins, Ph.D., *University of Pennsylvania*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*
- 1:30-2:30pm **Introduction to MGI/OMIM**
David Shaw, M.S., *The Jackson Laboratory* and
Ada Hamosh, M.D., *The Johns Hopkins University School of Medicine*

2:30-4:30pm **RNA-seq Analysis Workshop**
Steve Munger, 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**
An Epigenomic Regulatory System
Kenneth Paigen Ph.D., *The Jackson Laboratory*

Wednesday, July 17

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:25am **Sequencing Technology and Application**
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*

10:15-10:45am **Break**

10:45-11:40am **New Strategies for Accelerating Regeneration**
Nadia Rosenthal, Ph.D., *The Jackson Laboratory*

11:40-12:30pm **TBD**
Albert Cheng, Ph.D., *The Jackson Laboratory for Genomic Medicine*

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

1:30- 3:30pm **WES/WGS Analysis Workshop**
Nara Sobreira, M.D., Ph.D., *Johns Hopkins University School of Medicine*

Poster Session

Location: The Bar Harbor Regency

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

Evening Lecture

Location: The Bar Harbor Regency

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

Thursday, July 18

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:25am **A Primer on Quantitative Systems Biology**
Aravinda Chakravarti, Ph.D., *NYU Langone Health*

9:25-10:15am **Methods for Genetic Association Studies in Humans**
Goncalo Abecasis, Ph.D., *The University of Michigan*

10:15-10:45am **Break**

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

11:40-12:30pm **Regulation of Oxygen Homeostasis by Hypoxia-Inducible Factor 1**
Gregg Semenza, M.D., Ph.D., *Johns Hopkins University School of Medicine*

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

1:30- 2:30pm **Inborn Errors of Metabolism**
Hilary Vernon, M.D., Ph.D., *Johns Hopkins University School of Medicine*

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

Evening Lecture

Location: The Bar Harbor Regency

7:30-9:00pm **Insights from Genetic Disorders of Cranial Motor Neuron Development**
Elizabeth Engle, M.D., *Boston Children's Hospital*

Friday, July 19

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 **Epigenetic Regulation in Development, Aging and Disease**
Rudolf Jaenisch, M.D., *Whitehead Institute*

10:15-10:45am **Break**

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 **Using Large-Scale Genomic Databases to Understand Human Gene Function**
Daniel MacArthur., Ph.D., *The Broad Institute*

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 Bar Harbor Regency

7:30-9:00pm **Chen K. Chai Lecture**
Systems Genomics in Breast Cancer
Edison Liu, M.D., *The Jackson Laboratory*

Saturday, July 20 – 60th Anniversary Symposium

Location: The Bar Harbor Club

Please see [Symposium Schedule](#)

Sunday, July 21

No Scheduled Course Events

Monday, July 22

Location: The Jackson Laboratory, 600 Main Street

- 7:45-8:20am **Continental Breakfast - Roscoe's Dining Area**
- 8:30-9:25am **Genetics of Autoimmunity**
David Serreze, Ph.D., *The Jackson Laboratory*
- 9:25-10:15 am **Genetic Disorders of Immunity**
Jennifer Puck, M.D., *University of California San Francisco*
- 10:15-10:45am **Break**
- 10:45-11:40am **Immunodeficiency**
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 **ClinGen and ClinVar Workshop**
Heidi Rehm, Ph.D., *Broad Institute of MIT and Harvard*

Evening Lecture

Location: The Bar Harbor Regency

- 7:30-9:00pm **Case Studies in Genomic Medicine**
Heidi Rehm, Ph.D., *Massachusetts General Hospital*

Tuesday, 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*
- 9:25-10:15 am **African Evolutionary Genomics**
Sarah Tishkoff, Ph.D., *University of Pennsylvania*
- 10:15-10:45am **Break**

- 10:45-11:40am **Genetic and Pharmacologic Modifiers of Cystic Fibrosis**
Garry R. Cutting, M.D., *Johns Hopkins University School of Medicine*
- 11:40-12:30pm **Clinical Reasoning in the Genomic Era**
Leslie Biesecker, M.D., *National Human Genome Research Institute*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*
- 1:30- 3:30pm **UCSC Genome Browser in Clinical Medicine**
Robert Kuhn, Ph.D., UCSC

Location: The Bar Harbor Regency

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 and Howard Hughes Medical Institute*

Wednesday, July 24

Location: The Jackson Laboratory, 600 Main Street

- 7:45-8:25am **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25am **The Humanized Mouse in Biomedical Research**
Lenny Shultz, Ph.D., *The Jackson Laboratory*
- 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*
- 10:15-10:45am **Break**
- 10:45-11:40am **Genetic Medicine of African Population: Promise and Prospects**
Ambroise Wonkam, Ph.D., *University of Cape Town*
- 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**
Genetic Testing: View from the Front Lines
Robert Nussbaum, M.D., *Invitae*

Thursday, July 25

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:25 am **Proteomics as a Biological Assay**
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*

10:25-10:45am **Break**

10:45-11:40am **Biomedical Ethics in Genomics**
Debra Mathews, Ph.D., M.A., *Johns Hopkins Berman Institute of Bioethics, Johns Hopkins School of Medicine*

11:40-12:30pm **Antisense Therapy for Genetic Diseases**
Adrian Krainer, Ph.D., *Cold Spring Harbor Laboratory*

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

1:30-3:30pm **Skippy Lane Memorial Mouse Genetics Clinic**
Greg Cox, Ph.D., *The Jackson Laboratory*

5:00-7:00pm **Harbor Cruise** - *Advance sign up required*

Evening Lecture

Location: The Bar Harbor Regency

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

Friday, July 26

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:25am **Metabolomics and Human Disease**
Ralph DeBerardinis, M.D., Ph.D., *UT Southwestern*

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

10:25-10:45am **Break**

10:45-11:40am **Cancer Biology Exposed by Large-Scale Sequencing**
Victor Velculescu, M.D., Ph.D., *Johns Hopkins University School of
Medicine*

11:40-12:30pm **Course Summary & Wrap Up**
Robert Nussbaum, M.D., *Invitae*

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

End of the course