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
Edison Liu, M.D., The Jackson Laboratory

9:30-10:25am  History of Human Genetics
David Valle, M.D., Johns Hopkins University School of Medicine

10:25-11:00am  Break

11:00–11:55am  Opportunities for Using Mouse Models in the 21st Century
Gary Churchill, Ph.D., The Jackson Laboratory

11:55-12:50pm  Chromosome Structure and Function
Jeanne Lawrence, Ph.D., University of Massachusetts Medical School
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, Ill., 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., 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

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**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*

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**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*

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**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