



Human and Mammalian Genetics and Genomics: The 59th McKusick Short Course

July 15 – July 27, 2018

The Jackson Laboratory, Bar Harbor, Maine

EVENT SCHEDULE

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

Sunday, July 15

Location: The Jackson Laboratory, 600 Main Street

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

Monday, July 16

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**

- 10:45-11:40am **Chromosome Structure and Function**
 Jeanne Lawrence, Ph.D., *University of Massachusetts Medical School*
- 11:40-12:30pm **Genetic Recombination: What it is, Why You Should Care, and How it Works**
 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*
- 2:30-3:30pm **Molecular Genetics**
 John A. Phillips, III., M.D., *Vanderbilt University School of Medicine*

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**
Bridging Mendelian and Common Disease
 Richard A. Gibbs, Ph.D., *Baylor College of Medicine*

Tuesday, July 17

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 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*
- 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* and
Narayana Raghupathy, Ph.D., *The Jackson Laboratory*

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*

Wednesday, July 18

Location: The Jackson Laboratory, 600 Main Street

- 7:45-8:30am **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25am **CNVs in Human Disease**
Charles Lee, Ph.D., *The Jackson Laboratory for Genomic Medicine*
- 9:25-10:15am **New Strategies for Accelerating Regeneration**
Nadia Rosenthal, Ph.D., *The Jackson Laboratory*
- 10:15-10:45am **Break**
- 10:45-11:40am **The Humanized Mouse in Biomedical Research**
Lenny Shultz, Ph.D., *The Jackson Laboratory*
- 11:40-12:30pm **Q&A Session with Course Faculty**
- 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 **Using the Medical Phenome with Genome to Drive Discovery and Translation in Human Disease**
Nancy Cox, Ph.D., *Vanderbilt Genetics Institute*

Thursday, July 19

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:40am **A Primer on Quantitative Systems Biology**
Aravinda Chakravarti, Ph.D., *NYU Langone Health*

9:40-10:15am **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 **Genetics of Oxygen Homeostasis**
Gregg Semenza, M.D., Ph.D., *Johns Hopkins University School of Medicine*

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

2017 Video of: Massively Parallel Sequencing: Technologies and Applications
Elaine Mardis, Ph.D., *Nationwide Children's Hospital*

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 **Determination of Retinal Cell Fate and Nanobodies as Regulators of Intracellular Activities**
Connie Cepko, Ph.D., *Harvard University and Howard Hughes Medical Institute*

Friday, July 20

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**

10:45-11:40am **Genetics of Autoimmunity**
David Serreze, Ph.D., *The Jackson Laboratory*

11:40- 12:30pm **The Tip of the ENCODE Iceberg**
David Bodine, Ph.D., *National Human Genome Research 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 Jackson Laboratory, 600 Main Street

7:30-9:00pm **Chen K. Chai Lecture**
The Origins of Cellular Life
Jack Szostak, Ph.D., *Harvard Medical School and Massachusetts General Hospital*

Saturday, July 21

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 | Interpretation of Genetic Variants using Massive-Scale Reference Data Sets
Annie O'Donnell-Luria, M.D., Ph.D., <i>The Broad Institute</i> |
| 10:15-10:45am | Break |
| 10:45-11:40am | Pan-African and Global Impact of the H3Africa Initiative on Biomedical (Genomic) Research
Charles Rotimi, Ph.D., <i>National Human Genome Research Institute</i> |
| 11:40-12:30pm | Evolution of Complex Traits in Africa
Sarah Tishkoff, Ph.D., <i>University of Pennsylvania</i> |
| 12:30pm | Boxed Lunches |

Sunday, July 22

No Scheduled Course Events

Monday, July 23

Location: The Jackson Laboratory, 600 Main Street

- 7:45-8:20am **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*
- 9:25-10:15 am **The Major Histocompatibility Complex in Health and Disease**
Derry Roopenian, Ph.D., *The Jackson Laboratory*
- 10:15-10:45am **Break**
- 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 **ClinGen and ClinVar Workshop**
Heidi Rehm, Ph.D., *Massachusetts General Hospital*

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*

Tuesday, July 24

Location: The Jackson Laboratory, 600 Main Street

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

10:15-10:45am

Break

10:45-11:40am

Genetic Analysis at Scale

Benjamin Neale, Ph.D., *Massachusetts General Hospital and The Broad Institute*

11:40-12:30

The Human Microbiome: from Metagenomes to Therapeutics

Julia Oh, 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*

Location: The Bar Harbor Regency

5:00-7:00pm

Lobster Bake

Evening Lecture

Location: The Bar Harbor Regency

7:30-9:00pm

Using Genomics to Unravel the Human Wiring Diagram

Peter Donnelly, FRS, *Wellcome Trust Centre for Human Genetics, University of Oxford, Genomics Plc.*

Wednesday, July 25

Location: The Jackson Laboratory, 600 Main Street

7:45-8:25am

Continental Breakfast - *Roscoe's Dining Area*

8:30-9:25am

The Genetics and Clinical Phenotypes of Inherited Skeletal Dysplasias

Julie Hoover-Fong, M.D., Ph.D., *Johns Hopkins University School of Medicine*

9:25-10:15am

Geisinger's Genomics and Precision Health Initiative

David Ledbetter, Ph.D., *Geisinger*

10:15-10:45am

Break

10:45-11:40am

Pharmacogenomics: Discovery, Translation and Implementation

Richard Weinshilboum, M.D., *Mayo Clinic*

- 11:40-12:30pm **DNA Forensics**
Phil Reilly, M.D., J.D., *Third Rock Ventures*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*
- 1:00-2:00pm **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**
Clinical Reasoning in the Genomics Era
Leslie Biesecker, M.D., *National Human Genome Research Institute*

Thursday, July 26

Location: The Jackson Laboratory, 600 Main Street

- 7:45-8:25am **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25 am **Proteomics**
Steven Gygi, Ph.D., *Harvard Medical School*
- 9:25-10:25am **Genetic and Pharmacologic Modifiers of CF**
Garry R. Cutting, M.D., *Johns Hopkins University School of Medicine*
- 10:25-10:45am **Break**
- 10:45-11:40am **Prenatal Diagnosis and Screening**
Ignatia B. Van den Veyver, M.D., *Baylor College of Medicine*
- 11:40-12:30pm **Biomedical Ethics in Genomics**
Debra Mathews, Ph.D., M.A., *Johns Hopkins Berman Institute of Bioethics, Johns Hopkins School of Medicine*
- 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

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 27

Location: The Jackson Laboratory, 600 Main Street

7:45-8:25am

Continental Breakfast - Roscoe's Dining Area

8:30-9:25am

Metabolomics in the Assessment of Human Diseases

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

IPS Cell Technology, Gene Editing and Disease Research

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

11:40-12:30pm

Course Summary & Wrap Up

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

12:30-1:30pm

Lunch - Roscoe's Dining Area

End of the course