



Human and Mammalian Genetics and Genomics: The 66th McKusick Short Course

July 21 – August 1, 2025

All times listed are EDT (UTC – 4:00)

Schedule still subject to change.

Day 1: Monday, July 21st

Transportation to The Jackson Laboratory (See shuttle schedule in Canvas for full details)

8:00 – 9:30 AM	General Registration & Breakfast
9:30 – 9:45 AM	Course Announcements & General Housekeeping
9:45 – 10:00 AM	Welcome & Opening Remarks <i>Lon Cardon, Ph.D., FMedSci, The Jackson Laboratory</i>
10:00 – 11:00 AM	History of Human Genetics <i>David Valle, M.D., Johns Hopkins University School of Medicine</i>
11:00 – 12:00 PM	Mice as Models for Human Diseases <i>Greg Cox, Ph.D., The University of Maine</i>
12:00 – 1:00 PM	Lunch Break
1:00 – 2:00 PM	Chromosome Regulation and Nuclear Genome Organization <i>Jeanne Lawrence, Ph.D., University of Massachusetts Medical School</i>
2:00 – 3:00 PM	Genetic Recombination & Mutation <i>Beth Dumont, Ph.D., The Jackson Laboratory</i>
3:00 – 3:15 PM	Coffee Break
3:15 – 4:15 PM	Opportunities and Limits in the 21st Century Mouse Model System <i>Steven Munger, Ph.D., The Jackson Laboratory</i>

- 4:15 – 5:15 PM **Thomas H. Roderick Memorial Lecture Cocktail Reception**
- 5:15 – 5:30 PM **Introduction & Welcome – Thomas H. Roderick Memorial Lecture**
- 5:30 – 7:00 PM **Thomas H. Roderick Memorial Lecture – The Jackson Laboratory Auditorium**
The Evolution of Functional Genomics Technologies for Understanding Mammalian Biology and Disease Genetics
 John Schimenti, Ph.D., *Cornell University*

Transportation to COA/BH Regency

Day 2: Tuesday, July 22nd

Transportation to The Jackson Laboratory

- 7:30 – 8:30 AM Breakfast
- 8:30 – 9:30 AM **Navigating the Sea of Sequencing Technologies**
 Kimberly Doheny, Ph.D., *Johns Hopkins University School of Medicine*
- 9:30 – 10:30 AM **The Utility of Gapless Genome Assemblies in Understanding Biology**
 Rachel O’Neill, Ph.D., *Institute for Systems Genomics, University of Connecticut*
- 10:30 – 11:00 AM Coffee Break
- 11:00 – 12:00 PM **Single Cell Analysis in Development and Disease**
 Loyal Goff, Ph.D., *Johns Hopkins University School of Medicine*
- 12:00 – 1:00 PM **A Decade of Technology-Drive Insights into the 3D Genome's Structure-Function Relationship**
 Jennifer Phillips-Cremins, Ph.D., *University of Pennsylvania*
- 1:00 – 1:15 PM Group Photo (Rain Date Thursday, July 25th)
- 1:15 – 2:00 PM Lunch Break
- 2:00 – 2:45 PM **WORKSHOP: OMIM and Other Useful Genetic and Genomic Resources**
 Ada Hamosh, M.D., *Johns Hopkins University School of Medicine*
- 2:45 – 4:15 PM **WORKSHOP: The UCSC Genome Browser - Gateway to Genomic Data**
 Robert Kuhn, Ph.D., *University of California Santa Cruz (Retired)*

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM **Senior Lecturer – Stone House at Bar Harbor Regency**
Human Genomic Variants
Stylianos Antonarakis, M.D., DSc, *University of Geneva Medical School*

Day 3: Wednesday, July 23rd

Transportation to The Jackson Laboratory

7:30 – 8:30 AM Breakfast

8:30 – 9:30 AM **Transcriptional Regulation of Gene Expression**
Andrew McCallion, Ph.D., *Johns Hopkins University School of Medicine*

9:30 – 10:30 AM **Machine Learning for Regulatory Genomics: Basics and Pitfalls**
Michael Beer, Ph.D., *Johns Hopkins University*

10:30 – 11:00 AM Coffee Break

11:00 – 12:00 PM **The Landscape of Structural Variation and Dosage Sensitivity in the Human Genome**
Glennis Logsdon, Ph.D., *University of Pennsylvania*

12:00 – 1:00 PM **Systems Genetics and the Biology of the Cell**
Aravinda Chakravarti, Ph.D., *NYU Langone Health*

1:00 – 2:00 PM **Trainee Lunch**
Kimberly Doheny, Ph.D., *Johns Hopkins University School of Medicine*

2:00 – 3:00 PM **WORKSHOP: gnomAD - Human Genome Variation & its Properties**
Kaitlin Samocha, Ph.D., *Massachusetts General Hospital*

3:00 – 4:30 PM **WORKSHOP: RNA Sequencing**
Steven Munger, Ph.D., *The Jackson Laboratory*

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM **The Study of Rare Pediatric Neurological Diseases Drives Discoveries in Common Diseases – Stone House at the Bar Harbor Regency**
Hugo Bellen, D.M.V., Ph.D., *Baylor College of Medicine*

Day 4: Thursday, July 24th

Transportation to The Jackson Laboratory

7:30 – 8:30 AM Breakfast

- 8:30 – 9:30 AM **Mutations and their Origins**
Gilad Evrony, M.D., Ph.D., *NYU Langone Health*
- 9:30 – 10:30 AM **Human Structural Genomic Variation**
Charles Lee, Ph.D., *The Jackson Laboratory*
- 10:30 – 11:00 AM Coffee Break
- 11:00 AM – 12:00 PM **Title TBD**
Karen Miga, Ph.D., *University of California Santa Cruz*
- 12:00 – 1:00 PM **Title TBD**
Maanasa Raghavan, Ph.D., *The University of Chicago*
- 1:00 – 2:00 PM Lunch Break

2:00 – 4:00 PM **WORKSHOP: Human Pangenome Workshop**
Speaker TBD

- 4:15 – 5:45 PM Poster Session

Transportation to COA/BH Regency

- 7:00 – 8:30 PM **Multiomic Integration in Complex Traits: Beyond Expert Curation and Toward an AI Future**— Stone House at the Bar Harbor Regency
Judy Cho, M.D., *Mount Sinai*

Day 5: Friday, July 25th

Transportation to The Jackson Laboratory

- 7:30 – 8:30 AM Breakfast
- 8:30 – 9:30 AM **Inborn Errors of the Epigenetic Apparatus: Genetics of Chromatin Readers & Writers**
Hans Bjornsson, M.D., Ph.D., *Johns Hopkins University School of Medicine, University of Iceland*
- 9:30 – 10:30 AM **Genetic Epidemiology of Breast and Ovarian Cancer**
Larry Brody, Ph.D., *National Institutes of Health*
- 10:30 – 11:00 AM Coffee Break
- 11:00 – 12:00 PM **Heterogeneity & Evolutionary Trajectory of Breast Cancer**
Olufunmilayo Olopade, M.D., FACP, *UChicago Medicine*

12:00 – 1:00 PM

Cancer Biology Exposed Through Large-Scale Sequencing

Victor Velculescu, M.D., Ph.D., *Johns Hopkins University School of Medicine*

1:00 – 2:00 PM

Trainee Lunch

Alice Matimba, Ph.D., *Wellcome Connecting Science*

2:00 – 4:00 PM

WORKSHOP: Mouse Genome Informatics

Speaker TBD

4:15 – 4:30 PM

Introduction and Welcome to Chen K. Chai Memorial Lecture

4:30 – 6:00 PM

**Chen K. Chai Memorial Lecture – The Jackson Laboratory Auditorium
The Surprising Ways that the Genetic Component to Laboratory Values
Impact Clinical Decisions**

Pilar Ossorio, Ph.D., *University of Wisconsin Law School*

Transportation to COA/ BH Regency

Saturday, July 26th & Sunday, July 27th

No Scheduled Course Activities

Day 6: Monday, July 28th

Transportation to The Jackson Laboratory

7:30 – 8:30 AM	Breakfast
8:30 – 9:30 AM	Human Evolution and Adaptation in Africa Sarah Tishkoff, Ph.D., <i>University of Pennsylvania, Perelman School of Medicine</i>
9:30 – 10:30 AM	Harnessing African Genomes to Enhance Health Globally Ambroise Wonkam, M.D., Ph.D., <i>Johns Hopkins University</i>
10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	Title TBD TBD
12:00 – 1:00 PM	Brazilian Population Diversity: Its Contribution to Human Genetics Maria Rita Passos-Bueno, Ph.D., <i>University of São Paulo</i>
1:00 – 2:00 PM	Lunch
2:00 – 4:00 PM	WORKSHOP: Exome and Genome Analysis Workshop Nara Sobreira, M.D., Ph.D., <i>Johns Hopkins University</i>

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM	Title TBD – Stone House at the Bar Harbor Regency Anne O’Donnell-Luria, M.D., Ph.D., <i>Broad Institute of MIT & Harvard</i>
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Day 7: Tuesday, July 29th

Transportation to The Jackson Laboratory

7:30 – 8:30 AM	Breakfast
8:30 – 9:30 AM	Genomics Surveillance and Characterization of Microbial Threats Facilitates Early Detection and Containment of Disease Outbreaks in West Africa Christian Happi, Ph.D., <i>Redeemer’s University</i>
9:30 – 10:30 AM	Genetics of Infectious Diseases: Where the Rubber Hits the Road of Life Steven Holland, M.D., <i>National Institute of Allergy & Infectious Diseases, NIH</i>

10:30 – 11:00 AM	Coffee Break
11:00 AM – 12:00 PM	Title TBD Kate Sullivan, M.D., Ph.D., <i>The Children’s Hospital of Philadelphia</i>
12:00 – 1:00 PM	Genetics and Genomics of Oxygen Homeostasis Gregg Semenza, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</i>
1:00 – 2:00 PM	Lunch Break
2:00 – 2:30 PM	What Happens in a Human Genetics Clinic Ada Hamosh, Ph.D., <i>Johns Hopkins</i>
2:30 – 4:00 PM	Human Genetics Clinic Presentations

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM	Victor A. McKusick Memorial Lecture – Stone House at the Bar Harbor Regency Title TBD Kym Boycott, M.D., Ph.D., <i>Children’s Hospital of Eastern Ottawa</i>
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Day 8: Wednesday, July 30th

Transportation to The Jackson Laboratory

7:30 – 8:30 AM	Breakfast
8:30 – 9:30 AM	Prenatal Screening Diagnosis & Therapy Ignatia Van den Veyver, M.D., <i>Baylor College of Medicine</i>
9:30 – 10:30 AM	Essential Genes Mary Dickinson, Ph.D., <i>The Jackson Laboratory</i>
10:30 – 11:00 AM	Coffee Break
11:00 AM – 12:00 PM	Title TBD Steve Murray, Ph.D., <i>The Jackson Laboratory</i>
12:00 – 1:00 PM	Ethics and Policy Issues Raised by Genetics and Genomics Debra Mathews, Ph.D., M.A., <i>Johns Hopkins University</i>
1:00 – 2:00 PM	Lunch Break
2:00 – 4:00 PM	WORKSHOP: ClinGen, ClinVar, and other Genomic Curation Databases Heidi Rehm, Ph.D., <i>Massachusetts General Hospital & The Broad Institute</i>

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM **Insights into the Pathogenesis and Treatment of Vascular Connective Tissue Disorders** – Stone House at the Bar Harbor Regency
Harry ‘Hal’ Dietz, M.D., *Johns Hopkins University School of Medicine*

Day 9: Thursday, July 31st

Transportation to The Jackson Laboratory

7:30 – 8:30 AM Breakfast

8:30 – 9:30 AM **Inborn Errors of Metabolism**
Hilary Vernon, M.D., Ph.D., *Johns Hopkins University School of Medicine*

9:30 – 10:30 AM **Metabolomics and Human Disease Phenotypes**
Ralph DeBerardinis, M.D., Ph.D., *University of Texas, Southwestern*

10:30 – 11:00 AM Coffee Break

11:00 AM – 12:00 PM **Pharmacogenomics to Pharmaco-omics**
Richard Weinshilboum, M.D., *Mayo Clinic*

12:00 – 1:00 PM **TBD**
Tian Zhang, Ph.D., *University of Virginia*

1:00 – 2:00 PM Lunch Break

1:00 – 3:00 PM **Skippy Lane Memorial Mouse Clinic**

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM **ELSI Case Studies** – Stone House at the Bar Harbor Regency
John A. Phillips, III., M.D., *Vanderbilt University School of Medicine*
Ada Hamosh, M.D., *Johns Hopkins University School of Medicine*
Louise Wilkins-Haug, M.D., Ph.D., *Brigham and Women’s Hospital*

Day 10: Friday, August 1st

Transportation to The Jackson Laboratory

7:30 – 8:30 AM Breakfast

8:30 – 9:30 AM **Genomic Newborn Screening**
Wendy Chung, M.D., Ph.D., *Boston Children’s Hospital*

9:30 – 10:30 AM	Gene Therapy Approaches to Hemoglobinopathies <i>Stuart Orkin, M.D., Harvard Medical School</i>
10:30 – 11:00 AM	Coffee Break
11:00 AM – 12:00 PM	Precision Genome Editing Tools <i>Greg Newby, Ph.D., Johns Hopkins University School of Medicine</i>
12:00 – 1:00 PM	Genetic & Pharmacologic Modifiers of Cystic Fibrosis <i>Garry Cutting, M.D., Johns Hopkins University School of Medicine</i>
1:00 – 2:00 PM	Course Summary and Wrap-Up
2:00 – 2:30 PM	Lunch

Participants responsible for transportation. Island Explorer is available