



Human and Mammalian Genetics and Genomics: The 67th McKusick Short Course

July 20 – 31, 2026

All times listed are EDT (UTC – 4:00)

Schedule is subject to change.

Day 1: Monday, July 20th

Transportation to The Jackson Laboratory (See shuttle schedule 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., The Jackson Laboratory</i>
10:00 – 11:00 AM	History of Human Genetics <i>Dave 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 – 4:00 PM	Opportunities and Limits in the 21st Century Mouse Model System <i>Steven Munger, Ph.D., The Jackson Laboratory</i>
4:00 – 4:45 PM	Thomas H. Roderick Memorial Lecture Reception
4:45 – 5:00 PM	Introduction & Welcome – Thomas H. Roderick Memorial Lecture
5:00 – 6:30 PM	Thomas H. Roderick Memorial Lecture – The Jackson Laboratory Auditorium Title TBD <i>Patricia Wittkopp, Ph.D., University of Michigan</i>

Transportation to COA/BH Regency

Day 2: Tuesday, July 21st

Transportation to The Jackson Laboratory

8:30 – 9:00 AM	Breakfast
9:00 – 10:00 AM	A Field Guide to Modern Sequencing: From Molecules to Methods Winston Timp, Ph.D., <i>Johns Hopkins University</i>
10:00 – 11:00 AM	Genomic Innovation on the Short Arms of Acrocentric Chromosomes Adam Phillippy, Ph.D., <i>National Genome Research Institute</i>
11:00 – 11:30 AM	Coffee Break
11:30 – 12:30 PM	An Introduction to the Human Pangenome Tools Karen Miga, Ph.D., <i>University of California, Santa Cruz</i>
12:30 – 12:45 PM	Group Photo (Rain Date: Thursday, July 23)
12:45 – 1:30 PM	Lunch Break
1:30 – 2:30 PM	WORKSHOP: OMIM and Other Useful Genetic and Genomic Resources Ada Hamosh, M.D., <i>Johns Hopkins University School of Medicine</i>
2:30 – 4:00 PM	WORKSHOP: gnomAD - Human Genome Variation & its Properties Katherine Chao, M.A., <i>Broad Institute of Harvard and MIT</i>

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM	Human Genomic Variants – The Stone House at the Bar Harbor Regency Stylianios Antonarakis, M.D., DSc, <i>University of Geneva Medical School</i>
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Day 3: Wednesday, July 22nd

Transportation to The Jackson Laboratory

8:30 – 9:00 AM	Breakfast
9:00 – 10:00 AM	Transcriptional Regulation of Gene Expression Andrew McCallion, Ph.D., <i>Johns Hopkins University School of Medicine</i>
10:00 – 11:00 AM	Machine Learning for Regulatory Genomics: Basics and Pitfalls Michael Beer, Ph.D., <i>Johns Hopkins University</i>
11:00 – 11:30 AM	Coffee Break
11:30 – 12:30 PM	Genotype-to-Phenotype Mapping with AI Nadav Brandes, Ph.D., <i>NYU Grossman School of Medicine</i>

12:30 – 1:30 PM Lunch

1:30 – 2:30 PM **Systems Genetics and the Biology of the Cell**
Aravinda Chakravarti, Ph.D., *NYU Langone Health*

2:30 – 4:30 PM **WORKSHOP: ClinGen, ClinVar, and other Genomic Curation Databases**
Heidi Rehm, Ph.D., *Massachusetts General Hospital & The Broad Institute*

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM **Senior Lecture: Understanding the Genetic Architecture of Psoriasis** – Stone House at the Bar Harbor Regency
Anne Bowcock, Ph.D., *Icahn School of Medicine at Mount Sinai*

Day 4: Thursday, July 23rd

Transportation to The Jackson Laboratory

8:30 – 9:00 AM Breakfast

9:00 – 10:00 AM **The Landscape of Structural Variation and Dosage Sensitivity in the Human Genome**
Michael Talkowski, Ph.D., *Harvard Medical School, Mass General Hospital*

10:00 – 11:00 AM **Mendelian Disorders of the Epigenetic Machinery: Genetic Disorders with Epigenomics Consequences**
Jill Fahrner, Ph.D., *Johns Hopkins University School of Medicine*

11:00 – 11:30 AM Coffee Break

11:30 – 12:30 PM **Evidence of Intergenerational Transmission of Epigenetic Inheritance in PTSD**
Leon Mutesa, M.D. Ph.D. *University of Rwanda*

12:30 – 1:30 PM Lunch

1:30 – 2:30 PM **The Power of Ancient and Modern Genomics in Reconstructing Human Population Histories**
Maanasa Raghavan, Ph.D., *The University of Chicago*

2:30 – 4:15 PM **WORKSHOP: The UCSC Genome Browser - Gateway to Genomic Data**
Robert Kuhn, Ph.D., *University of California Santa Cruz (Retired)*

4:15 – 6:00 PM Poster Session

Transportation to COA/BH Regency

7:00 – 8:30 PM **From Genomes to Mechanisms to Medicine: AI-Enabled Discovery in Human Genetics**
Olga Troyanskaya, Ph.D., *Princeton University*

Day 5: Friday, July 24th

Transportation to The Jackson Laboratory

8:30 – 9:00 AM	Breakfast
9:00 – 10:00 AM	Measuring, Modeling, and Reprogramming Tissue Organization Fei Chen, Ph.D., Broad Institute of MIT and Harvard
10:00 – 11:00 AM	Cancer Biology Exposed Through Large-Scale Sequencing Victor Velculescu, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</i>
11:00 – 11:30 AM	Coffee Break
11:30 – 12:30 PM	Heterogeneity & Evolutionary Trajectory of Breast Cancer Olufunmilayo Olopade, M.D., FACP, <i>UChicago Medicine</i>
12:30 – 1:30 PM	Lunch
1:30 – 2:30 PM	Intrinsic and Extrinsic Drivers of Stem Cell Fitness During Aging Jennifer Trowbridge, Ph.D., <i>The Jackson Laboratory</i>
2:30 – 4:30 PM	WORKSHOP: RNA Sequencing Steven Munger, Ph.D., <i>The Jackson Laboratory</i>
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 Out of the Mist: The Role of Long Noncoding RNA in Human Development and Disorders John Mattick, Ph.D., <i>University of New South Wales Sydney</i>

Saturday, July 25th & Sunday, July 26th

No Scheduled Course Activities

Day 6: Monday, July 27th

Transportation to The Jackson Laboratory

8:30 – 9:00 AM	Breakfast
9:00 – 10:00 AM	Human Evolution and Adaptation in Africa Sarah Tishkoff, Ph.D., <i>University of Pennsylvania, Perelman School of Medicine</i>
10:00 – 11:00 AM	Harnessing African Genomes to Enhance Health Globally Ambroise Wonkam, M.D., Ph.D., <i>Johns Hopkins University</i>

- 11:00 – 11:30 AM Coffee Break
- 11:30 – 12:30 PM **Uncovering New Regions of the Human Genome using Long-Read Sequencing**
Solomon Ofori-acquah, Ph.D., *Emory School of Medicine*
- 12:30 – 1:30 PM Lunch
- 1:30 – 2:30 PM **Consanguineous Populations as Engines of Discovery in Genomic Medicine**
Fowzan Alkuraya, Ph.D., *Alfaisal University*

2:30 – 4:30 PM **WORKSHOP: Exome and Genome Analysis Workshop**
Nara Sobreira, M.D., Ph.D., *Johns Hopkins University*

Participants responsible for transportation. Island Explorer is available.

- 7:00 – 8:30 PM **Genetics and Genomics of Oxygen Homeostasis – The Stone House at the Bar Harbor Regency**
Gregg Semenza, M.D., Ph.D., *Johns Hopkins University School of Medicine*

Day 7: Tuesday, July 28th

Transportation to The Jackson Laboratory

- 8:30 – 9:00 AM Breakfast
- 9:00 – 10:00 AM **Genomics Surveillance and Characterization of Microbial Threats Facilitates Early Detection and Containment of Disease Outbreaks in West Africa**
Christian Happi, Ph.D., *Redeemer's University*
- 10:00 – 11:00 AM **Genetics of Infectious Diseases: Where the Rubber Hits the Road of Life**
Steven Holland, M.D., *National Institute of Allergy & Infectious Diseases, NIH*
- 11:00 – 11:30 AM Coffee Break
- 11:30 – 12:30 PM **Title TBD**
Troy Torgerson, M.D., Ph.D., *Allen Institute*
- 12:30 – 1:30 PM **Title TBD**
Cathleen Lutz, Ph.D., M.B.A., *The Jackson Laboratory*
- 1:30 – 2:30 PM Lunch
- 2:30 – 3:00 PM **What Happens in a Human Genetics Clinic**
Ada Hamosh, Ph.D., *Johns Hopkins*
- 3:00 – 4:30 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**

Genomic Newborn Screening

Wendy Chung, M.D., Ph.D., *Boston Children's Hospital*

Day 8: Wednesday, July 29th

Transportation to The Jackson Laboratory

8:30 – 9:00 AM	Breakfast
9:00 – 10:00 AM	Prenatal Screening Diagnosis & Therapy Ignatia Van den Veyver, M.D., <i>Baylor College of Medicine</i>
10:00 – 11:00 AM	Aneuploidy in Human Embryos Rajiv McCoy, Ph.D., <i>Johns Hopkins University</i>
11:00 – 11:30 AM	Coffee Break
11:30 – 12:30 PM	From Disease Mechanisms to Therapeutic Targets for tRNA Synthetase-Associated Inherited Peripheral Neuropathies Robert Burgess, Ph.D., <i>The Jackson Laboratory</i>
12:30 – 1:30 PM	Lunch
1:30 – 2:30 PM	Ethics and Policy Issues Raised by Genetics and Genomics Debra Mathews, Ph.D., M.A., <i>Johns Hopkins University</i>
2:30 – 4:30 PM	WORKSHOP: Mouse Genome Informatics Carol Bult, Ph.D., <i>The Jackson Laboratory</i>

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., <i>Vanderbilt University School of Medicine</i> Ada Hamosh, M.D., <i>Johns Hopkins University School of Medicine</i> Debra Mathews, Ph.D., M.A., <i>Johns Hopkins University</i> Louise Wilkins-Haug, M.D., Ph.D., <i>Brigham and Women's Hospital</i>
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Day 9: Thursday, July 30th

Transportation to The Jackson Laboratory

8:30 – 9:00 AM	Breakfast
9:00 – 10:00 AM	Inborn Errors of Metabolism Hilary Vernon, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</i>
10:00 – 11:00 AM	Metabolomics and Human Disease Phenotypes Ralph DeBerardinis, M.D., Ph.D., <i>University of Texas, Southwestern</i>

11:00 – 11:30 AM	Coffee Break
11:30 – 12:30 PM	Pharmacogenomics to Pharmaco-omics Liewei Wang, M.D., Ph.D., <i>Mayo Clinic</i>
12:30 – 1:30 PM	Lunch
1:30 – 2:30 PM	TBD

2:30 – 4:30 PM **Skippy Lane Memorial Mouse Clinic – Tentative**

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., <i>Johns Hopkins University School of Medicine</i>
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Day 10: Friday, July 31st

Transportation to The Jackson Laboratory

8:00 – 8:30 AM	Breakfast
8:30 – 9:30 AM	Precision Genome Editing Tools Greg Newby, Ph.D., <i>Johns Hopkins University School of Medicine</i>
9:30 – 10:30 AM	Genetic Medicine for Sickle Cell Disease Daniel Bauer, M.D., Ph.D., <i>Harvard Stem Cell Institute</i>
10:30 – 10:45 AM	Coffee Break
10:45 – 11:45 AM	The First Successful Personalized Gene Editing Therapy – Follow-up from 2025 Kiran Musunuru, M.D., Ph.D., M.P.H., M.L, M.R.A., <i>University of Pennsylvania, Perelman School of Medicine</i> Rebecca Ahrens-Nicklas, M.D., Ph.D., <i>Children’s Hospital of Philadelphia</i> *presenting remotely
11:45 – 12:45 PM	Genetic & Pharmacologic Modifiers of Cystic Fibrosis Garry Cutting, M.D., <i>Johns Hopkins University School of Medicine</i>
12:45 – 1:15 PM	Course Summary and Wrap-Up
1:15 – 1:45 PM	Lunch

Participants responsible for transportation. Island Explorer is available