



**The Jackson
Laboratory**



JOHNS HOPKINS
UNIVERSITY

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 is 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 Ada Hamosh, M.D., <i>Johns Hopkins University School of Medicine</i>
10:00 – 11:00 AM	Mice as Models for Human Diseases Greg Cox, Ph.D., <i>The University of Maine</i>
11:00 – 12:00 PM	Chromosome Regulation and Nuclear Genome Organization Jeanne Lawrence, Ph.D., <i>University of Massachusetts Medical School</i>
12:00 – 1:00 PM	Lunch Break
1:00 – 2:00 PM	Genetic Recombination & Mutation Beth Dumont, Ph.D., <i>The Jackson Laboratory</i>
2:00 – 3:00 PM	Opportunities and Limits in the 21st Century Mouse Model System Steven Munger, Ph.D., <i>The Jackson Laboratory</i>
3:00 – 3:15 PM	Coffee Break
3:15 – 3:30 PM	Introduction & Welcome – Thomas H. Roderick Memorial Lecture
3:30 – 5:00 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., <i>Cornell University</i>
5:00 – 6:00 PM	Thomas H. Roderick Memorial Lecture Reception

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., <i>Johns Hopkins University School of Medicine</i>
9:30 – 10:30 AM	The Utility of Gapless Genome Assemblies in Understanding Biology Rachel O'Neill, Ph.D., <i>Institute for Systems Genomics, University of Connecticut</i> *presenting remotely
10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	Single Cell Analysis in Development and Disease Loyal Goff, Ph.D., <i>Johns Hopkins University School of Medicine</i>
12:00 – 1:00 PM	A Decade of Technology-Drive Insights into the 3D Genome's Structure-Function Relationship Jennifer Phillips-Cremins, Ph.D., <i>University of Pennsylvania</i>
1:00 – 1:15 PM	Group Photo (Rain Date Thursday, July 25 th)
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., <i>Johns Hopkins University School of Medicine</i>
2:45 – 4:15 PM	WORKSHOP: The UCSC Genome Browser - Gateway to Genomic Data Robert Kuhn, Ph.D., <i>University of California Santa Cruz (Retired)</i>

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, <i>University of Geneva Medical School</i>
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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., <i>Johns Hopkins University School of Medicine</i>
9:30 – 10:30 AM	Machine Learning for Regulatory Genomics: Basics and Pitfalls Michael Beer, Ph.D., <i>Johns Hopkins University</i>

10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	Uncovering New Regions of the Human Genome using Long-Read Sequencing Glennis Logsdon, Ph.D., <i>University of Pennsylvania</i>
12:00 – 1:00 PM	Systems Genetics and the Biology of the Cell Aravinda Chakravarti, Ph.D., <i>NYU Langone Health</i>
1:00 – 2:00 PM	Trainee Lunch Kimberly Doheny, Ph.D., <i>Johns Hopkins University School of Medicine</i>
2:00 – 3:00 PM	WORKSHOP: gnomAD - Human Genome Variation & its Properties Kaitlin Samocha, Ph.D., <i>Massachusetts General Hospital</i>
3:00 – 4:30 PM	WORKSHOP: RNA Sequencing Steven Munger, Ph.D., <i>The Jackson Laboratory</i>

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., <i>Baylor College of Medicine</i>
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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 Ehrony, M.D., Ph.D., <i>NYU Langone Health</i>
9:30 – 10:30 AM	Human Structural Genomic Variation Charles Lee, Ph.D., <i>The Jackson Laboratory</i> *presenting remotely
10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	The Human Pangenome Project: Creating a Reference that Better Represents Human Global Genetic Diversity Karen Miga, Ph.D., <i>University of California Santa Cruz</i>
12:00 – 1:00 PM	The Power of Ancient and Modern Genomics in Reconstructing Human Population Histories Maanasa Raghavan, Ph.D., <i>The University of Chicago</i>
1:00 – 2:00 PM	Lunch Break
2:00 – 4:00 PM	WORKSHOP: Human Pangenome Workshop - An Introduction to the Human Pangenome Tools and Workflows Karen Miga, Ph.D., <i>University of California Santa Cruz</i>

4:15 – 5:45 PM	Poster Session
<i>Transportation to COA/BH Regency</i>	
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., <i>Mount Sinai</i>

Day 5: Friday, July 25th

	<i>Transportation to The Jackson Laboratory</i>
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., <i>Johns Hopkins University School of Medicine, University of Iceland</i>
9:30 – 10:30 AM	Genetic Epidemiology of Breast and Ovarian Cancer Larry Brody, Ph.D., <i>National Institutes of Health</i>
10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	Heterogeneity & Evolutionary Trajectory of Breast Cancer Olufunmilayo Olopade, M.D., FACP, <i>UChicago Medicine</i>
12:00 – 1:00 PM	Cancer Biology Exposed Through Large-Scale Sequencing Victor Velculescu, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</i>
1:00 – 2:00 PM	Trainee Lunch Alice Matimba, Ph.D., <i>Wellcome Connecting Science</i>
2:00 – 4:00 PM	WORKSHOP: Mouse Genome Informatics Susan Bello, 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 Responsible Use of Shared Data Pilar Ossorio, Ph.D., <i>University of Wisconsin Law School</i>

Transportation to BH Regency

6:45 – 8:00 PM	Chen K. Chai Memorial Lecture Dinner – Stone House at the Bar Harbor Regency *Cash bar available
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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	Advancing Rare Diseases Genomics from Africa Aime Lumaka, M.D., Ph.D., <i>University of Kinhasa</i>
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 – 3:00 PM	WORKSHOP: Introduction to Exome and Genome Analysis Workshop Nara Sobreira, M.D., Ph.D., <i>Johns Hopkins University</i>
3:30 – 4:15 PM	Fireside Chat with Francis Collins – Roscoe's Francis Collins, M.D., Ph.D., <i>National Human Genome Research Institute</i> Moderator: TBD
4:15 – 5:15 PM	Fireside Chat Reception – Roscoe's Patio

Transportation to COA/BH Regency

7:00 – 8:30 PM	Navigating the Unknown: Improving Rare Disease Diagnosis into the Noncoding Genome – 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 – 12:00 PM	When Lymphocytes go South, Very Very South 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 A Diagnosis for all Rare Genetic Diseases: Collaborative Approaches to Discovery and Translation 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	Building Precision Mouse Models of Rare Disease Steve Murray, Ph.D., <i>The Jackson Laboratory</i>
10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	Inborn Errors of Metabolism Hilary Vernon, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</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., <i>Johns Hopkins University School of Medicine</i>
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Day 9: Thursday, July 31st

Transportation to The Jackson Laboratory

7:30 – 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	Metabolomics and Human Disease Phenotypes Ralph DeBerardinis, M.D., Ph.D., <i>University of Texas, Southwestern</i>
10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	Pharmacogenomics to Pharmaco-omics Richard Weinshilboum, M.D., <i>Mayo Clinic</i>
12:00 – 1:00 PM	Developing a DIA-Based Proteomics Approach to Decipher Protein Degradation and Synthesis Dynamics Tian Zhang, Ph.D., <i>University of Virginia</i>
1:00 – 2:00 PM	Lunch Break
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	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> Louise Wilkins-Haug, M.D., Ph.D., <i>Brigham and Women's Hospital</i>
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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., <i>Boston Children's Hospital</i>
9:30 – 10:30 AM	Gene Therapy Approaches to Hemoglobinopathies Stuart Orkin, M.D., <i>Harvard Medical School</i>
10:30 – 11:00 AM	Coffee Break

11:00 – 12:00 PM	Implications of the First Successful Personalized Gene Editing Therapy 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
12:00 – 1:00 PM	Genetic & Pharmacologic Modifiers of Cystic Fibrosis Garry Cutting, M.D., <i>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