



The Jackson
Laboratory



Human and Mammalian Genetics and Genomics: The 65th McKusick Short Course

July 22 – August 2, 2024

All times listed are EDT (UTC – 4:00)

Schedule still subject to change.

Day 1: Monday, July 22nd

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

- 4:45 – 5:45 PM **Thomas H. Roderick Memorial Lecture Cocktail Reception**
- 5:45 – 6:00 PM **Introduction & Welcome – Thomas H. Roderick Memorial Lecture**
- 6:00 – 7:30 PM **Thomas H. Roderick Memorial Lecture** – The Jackson Laboratory Auditorium
Genetic Modifiers of mRNA Translation and Neurological Disorders
 Sue Ackerman, Ph.D., *University of California San Diego*

Transportation to COA/BH Regency

Day 2: Tuesday, July 23rd

Transportation to The Jackson Laboratory

- 7:30 – 8:00 AM Breakfast
- 8:00 – 9:00 AM **Navigating the Sea of Sequencing Technologies**
 Kimberly Doheny, Ph.D., *Johns Hopkins University School of Medicine*
- 9:00 – 10:00 AM **Transcriptional Regulation of Gene Expression**
 Andrew McCallion, Ph.D., *Johns Hopkins University School of Medicine*
- 10:00 – 10:30 AM Coffee Break
- 10:30 – 11:30 AM **Single Cell Analysis in Development and Disease**
 Loyal Goff, Ph.D., *Johns Hopkins University School of Medicine*
- 11:30 AM – 12:30 PM **A Decade of Technology-Drive Insights Into the 3D Genome's Structure-Function Relationship**
 Jennifer Phillips-Cremins, Ph.D., *University of Pennsylvania*
- 12:30 – 12:45 PM Group Photo (Rain Date Thursday, July 25th)
- 12:45 – 2:00 PM Lunch Break
- 2:00 – 3:00 PM **WORKSHOP: OMIM and Other Useful Genetic and Genomic Resources**
 Ada Hamosh, M.D., *Johns Hopkins University School of Medicine*
- 3:00 – 3:30 PM **WORKSHOP: Intro All of US**
 Anjené Musick, Ph.D., MPH, *National Institutes of Health*
- 3:30 – 4:00 PM **WORKSHOP: Intro to the UK Biobank**
 Megan Conroy, *UK Biobank*
 *Presenting Remotely

Participants responsible for transportation. Island Explorer is available.

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

Day 3: Wednesday, July 24th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM Breakfast

8:00 – 9:00 AM **The Utility of Gapless Genome Assemblies in Understanding Biology**
Rachel O’Neill, Ph.D., *Institute for Systems Genomics, University of Connecticut*

9:00 – 10:00 AM **Machine Learning for Regulatory Genomics: Basics and Pitfalls**
Michael Beer, Ph.D., *Johns Hopkins University*

10:00 – 10:30 AM Coffee Break

10:30 – 11:30 AM **The Landscape of Structural Variation and Dosage Sensitivity in the Human Genome**
Michael Talkowski, Ph.D., *Harvard Medical School, Mass General Hospital*

11:30 AM – 12:30 PM **Probing Molecular Phenotypes with Quantitative Proteomics**
Devin Schweppe, Ph.D., *University of Washington*

12:30 – 2:00 PM **Trainee Lunch: Women in Science**
Kimberly Doheny, Ph.D., *Johns Hopkins University School of Medicine*

2:00 – 3:00 PM **WORKSHOP: The UCSC Genome Browser - Gateway to Genomic Data**
Robert Kuhn, Ph.D., *University of California Santa Cruz (Retired)*

3:00 – 4:00 PM **WORKSHOP: Human Pangenome Workshop**
Karen Miga, Ph.D., *University of California Santa Cruz*

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM **Senior Lecturer** – Stone House at Bar Harbor Regency
Inherited Defects of Adaptive Immunity: Advances in Diagnosis and Treatment
Jennifer Puck, M.D., *University of California San Francisco*

Day 4: Thursday, July 25th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	gnomAD: Human Genome Variation & its Properties Anne O’Donnell-Luria, M.D., Ph.D., <i>Broad Institute of MIT & Harvard</i>
9:00 – 10:00 AM	Systems Genetics and the biology of the Cell Aravinda Chakravarti, Ph.D., <i>NYU Langone Health</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	TBD Elissa Chesler, Ph.D., <i>The Jackson Laboratory</i>
11:30 AM – 12:30 PM	The Study of Rare Pediatric Neurological Diseases Drives Discoveries in Common Diseases Hugo Bellen, D.M.V., Ph.D., <i>Baylor College of Medicine</i>
12:30 – 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>
4:15 – 5:45 PM	Poster Session

Transportation to COA/BH Regency

7:00 – 8:30 PM	Victor A. McKusick Memorial Lecture – Stone House at the Bar Harbor Regency Tackling the VUS Problem: An Open Debate and Discussion Heidi Rehm, Ph.D., <i>Massachusetts General Hospital & The Broad Institute</i>
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Day 5: Friday, July 26th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	Heterogeneity & Evolutionary Trajectory of Breast Cancer Olufumilayo Olopade, M.D., FACP, <i>UChicago Medicine</i> <i>*Presenting Remotely</i>

9:00 – 10:00 AM	Genetic Epidemiology of Breast and Ovarian Cancer Larry Brody, Ph.D., <i>National Institutes of Health</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	Cancer Biology Exposed Through Large-Scale Sequencing Victor Velculescu, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</i>
11:30 AM – 12:30 PM	Mendelian Disorders of the Epigenetic Machinery: Genetic Disorders with Epigenomics Consequences Jill Fahrner, Ph.D., <i>Johns Hopkins University School of Medicine</i>
12:30 – 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>
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 Nancy Cox, Ph.D., <i>Vanderbilt University Medical School</i>

Transportation to COA/ BH Regency

Saturday, July 27th & Sunday, July 28th

No Scheduled Course Activities

Day 6: Monday, July 29th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	Human Evolution and Adaptation in Africa Sarah Tishkoff, Ph.D., <i>University of Pennsylvania, Perelman School of Medicine</i>
9:00 – 10:00 AM	Harnessing our Common African Genome to Improve Health Globally Ambroise Wonkam, M.D., Ph.D., <i>Johns Hopkins University</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	Leveraging African Genetic Diversity for Novel Gene Discovery & Genetic Risk Prediction Segun Fatumo, Ph.D., <i>London School of Hygiene & Tropical Medicine</i>
11:30 AM – 12:30 PM	Genetic Medicine for Sickle Cell Disease Dan Bauer, M.D., Ph.D., <i>Harvard Stem Cell Institute</i>
12:30 – 2:00 PM	Trainee Lunch: Cultivating Trainee Skills and Professional Development Alice Matimba, Ph.D., <i>Wellcome Connecting Science</i>
2:00 – 4:00 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	Genetic Diversity of the Human Y Chromosome – Stone House at Bar Harbor Regency Charles Lee, Ph.D., <i>The Jackson Laboratory</i>
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Day 7: Tuesday, July 30th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 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>

9:00 – 10:00 AM	Inborn Errors of Immunity: One Gene, Multiple Phenotypes Luigi Notarangelo, M.D., <i>National Institutes of Health</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 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>
11:30 AM – 12:30 PM	Inborn Errors of Metabolism Hilary Vernon, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</i>
12:30 – 2:00 PM	Lunch Break
2:00 – 4:00 PM	WORKSHOP: Mouse Genome Informatics Richard Balderelli, Ph.D., <i>The Jackson Laboratory</i> Cynthia Smith, Ph.D., <i>The Jackson Laboratory</i>

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM	Genetics and Genomics of Oxygen Homeostasis – Stone House at Bar Harbor Regency Gregg Semenza, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</i>
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Day 8: Wednesday, July 31st

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	Essential Genes Mary Dickinson, Ph.D., <i>The Jackson Laboratory</i>
9:00 – 10:00 AM	Prenatal Screening Diagnosis & Therapy Ignatia Van den Veyver, M.D., <i>Baylor College of Medicine</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	After the Diagnosis: Mobilizing the Preclinical Pipeline for Rare Disease Cat Lutz, Ph.D., M.B.A., <i>The Jackson Laboratory</i>
11:30 AM – 12:30 PM	Ethics and Policy Issues Raised by Genetics and Genomics Debra Mathews, Ph.D., M.A., <i>Johns Hopkins University</i>
12:30 – 2:00 PM	Lunch Break

2:00 – 2:30 PM **What Happens in a Human Genetics Clinic**
Ada Hamosh, Ph.D., *Johns Hopkins*

2:30 – 4:00 PM **Human Genetics Clinic Presentations**
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 Bar Harbor Regency
Harry ‘Hal’ Dietz, M.D., *Johns Hopkins University School of Medicine*

Day 9: Thursday, August 1st

Transportation to The Jackson Laboratory

7:30 – 8:00 AM Breakfast

8:00 – 9:00 AM **Metabolomics and Human Disease Phenotypes**
Ralph DeBerardinis, M.D., Ph.D., *University of Texas, Southwestern*

9:00 – 10:00 AM **Pharmacogenomics to Pharmaco-omics**
Richard Weinshilboum, M.D., *Mayo Clinic*

10:00 – 10:30 AM Coffee Break

10:30 – 11:30 AM **Antisense Modulation of RNA Splicing for Rare Disease Therapy**
Adrian Krainer, Ph.D., *Cold Spring Harbor Laboratory*

11:30 AM – 12:30 PM **The Expanding Use of Gene Targeting Therapeutics in Neurological Disease**
Charlotte Sumner, M.D., *Johns Hopkins University School of Medicine*

12:30 – 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*
Debra Mathews, Ph.D., MA, *Johns Hopkins University*
Louise Wilkins-Haug, M.D., Ph.D., *Boston Children’s Hospital*

Day 10: Friday, August 2nd

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	Integrating Molecular Genetics Testing into Mainstream Medicine: View from a Commercial Laboratory Robert Nussbaum, M.D., <i>Invitae</i>
9:00 – 10:00 AM	mRNA Therapy for Cardiac Fibrosis Jonathan Epstein, M.D., University of Pennsylvania Medical School
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	Precision Genome Editing Tools Greg Newby, Ph.D., <i>Johns Hopkins University School of Medicine</i>
11:30 AM – 12:30 PM	Genetic & Pharmacologic Modifiers of Cystic Fibrosis Garry Cutting, M.D., <i>Johns Hopkins University School of Medicine</i>
12:30 – 1:30 PM	Course Summary and Wrap-Up
1:30 – 2:30 PM	Lunch

Participants responsible for transportation. Island Explorer is available.