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# **Human and Mammalian Genetics and Genomics: The 66<sup>th</sup> 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 21<sup>st</sup>**

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

*Transportation to COA/BH Regency*

## Day 2: Tuesday, July 22<sup>nd</sup>

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### *Transportation to The Jackson Laboratory*

7:30 – 8:30 AM	Breakfast
8:30 – 9:30 AM	<b>Navigating the Sea of Sequencing Technologies</b> Kimberly Doheny, Ph.D., <i>Johns Hopkins University School of Medicine</i>
9:30 – 10:30 AM	<b>The Utility of Gapless Genome Assemblies in Understanding Biology</b> 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	<b>Single Cell Analysis in Development and Disease</b> Loyal Goff, Ph.D., <i>Johns Hopkins University School of Medicine</i>
12:00 – 1:00 PM	<b>A Decade of Technology-Drive Insights into the 3D Genome's Structure-Function Relationship</b> Jennifer Phillips-Cremins, Ph.D., <i>University of Pennsylvania</i>
1:00 – 1:15 PM	Group Photo (Rain Date Thursday, July 25 <sup>th</sup> )
1:15 – 2:00 PM	Lunch Break
2:00 – 2:45 PM	<b>WORKSHOP: OMIM and Other Useful Genetic and Genomic Resources</b> Ada Hamosh, M.D., <i>Johns Hopkins University School of Medicine</i>
2:45 – 4:15 PM	<b>WORKSHOP: The UCSC Genome Browser - Gateway to Genomic Data</b> 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	<b>Senior Lecturer – Stone House at Bar Harbor Regency</b> <b>Human Genomic Variants</b> Stylianios Antonarakis, M.D., DSc, <i>University of Geneva Medical School</i>
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## Day 3: Wednesday, July 23<sup>rd</sup>

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### *Transportation to The Jackson Laboratory*

7:30 – 8:30 AM	Breakfast
8:30 – 9:30 AM	<b>Transcriptional Regulation of Gene Expression</b> Andrew McCallion, Ph.D., <i>Johns Hopkins University School of Medicine</i>
9:30 – 10:30 AM	<b>Machine Learning for Regulatory Genomics: Basics and Pitfalls</b> Michael Beer, Ph.D., <i>Johns Hopkins University</i>

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

*Participants responsible for transportation. Island Explorer is available.*

7:00 – 8:30 PM	<b>The Study of Rare Pediatric Neurological Diseases Drives Discoveries in Common Diseases</b> – 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 24<sup>th</sup>

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*Transportation to The Jackson Laboratory*

7:30 – 8:30 AM	Breakfast
8:30 – 9:30 AM	<b>Mutations and their Origins</b> Gilad Evrony, M.D., Ph.D., <i>NYU Langone Health</i>
9:30 – 10:30 AM	<b>Human Structural Genomic Variation</b> Charles Lee, Ph.D., <i>The Jackson Laboratory</i> *presenting remotely
10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	<b>The Human Pangenome Project: Creating a Reference that Better Represents Human Global Genetic Diversity</b> Karen Miga, Ph.D., <i>University of California Santa Cruz</i>
12:00 – 1:00 PM	<b>The Power of Ancient and Modern Genomics in Reconstructing Human Population Histories</b> Maanasa Raghavan, Ph.D., <i>The University of Chicago</i>
1:00 – 2:00 PM	Lunch Break
2:00 – 4:00 PM	<b>WORKSHOP: Human Pangenome Workshop - An Introduction to the Human Pangenome Tools and Workflows</b> Karen Miga, Ph.D., <i>University of California Santa Cruz</i>

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 25<sup>th</sup>

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*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**  
Susan Bello, Ph.D., *The Jackson Laboratory*

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., *University of Wisconsin Law School*

*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

## Saturday, July 26<sup>th</sup> & Sunday, July 27<sup>th</sup>

*No Scheduled Course Activities*

## Day 6: Monday, July 28<sup>th</sup>

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### *Transportation to The Jackson Laboratory*

7:30 – 8:30 AM	Breakfast
8:30 – 9:30 AM	<b>Human Evolution and Adaptation in Africa</b> Sarah Tishkoff, Ph.D., <i>University of Pennsylvania, Perelman School of Medicine</i>
9:30 – 10:30 AM	<b>Harnessing African Genomes to Enhance Health Globally</b> Ambroise Wonkam, M.D., Ph.D., <i>Johns Hopkins University</i>
10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	<b>Advancing Rare Diseases Genomics from Africa</b> Aime Lumaka, M.D., Ph.D., <i>University of Kinshasa</i>
12:00 – 1:00 PM	<b>Brazilian Population Diversity: Its Contribution to Human Genetics</b> Maria Rita Passos-Bueno, Ph.D., <i>University of São Paulo</i>
1:00 – 2:00 PM	Lunch
2:00 – 3:00 PM	<b>WORKSHOP: Introduction to Exome and Genome Analysis Workshop</b> Nara Sobreira, M.D., Ph.D., <i>Johns Hopkins University</i>
3:30 – 4:15 PM	<b>Fireside Chat with Francis Collins – Roscoe’s</b> Francis Collins, M.D., Ph.D., <i>National Human Genome Research Institute</i> Moderator: TBD
4:15 – 5:15 PM	<b>Fireside Chat Reception – Roscoe’s Patio</b>

### *Transportation to COA/BH Regency*

7:00 – 8:30 PM	<b>Navigating the Unknown: Improving Rare Disease Diagnosis into the Noncoding Genome – Stone House at the Bar Harbor Regency</b> Anne O’Donnell-Luria, M.D., Ph.D., <i>Broad Institute of MIT &amp; Harvard</i>
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## Day 7: Tuesday, July 29<sup>th</sup>

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### *Transportation to The Jackson Laboratory*

7:30 – 8:30 AM	Breakfast
8:30 – 9:30 AM	<b>Genomics Surveillance and Characterization of Microbial Threats Facilitates Early Detection and Containment of Disease Outbreaks in West Africa</b> Christian Happi, Ph.D., <i>Redeemer’s University</i>
9:30 – 10:30 AM	<b>Genetics of Infectious Diseases: Where the Rubber Hits the Road of Life</b> Steven Holland, M.D., <i>National Institute of Allergy &amp; Infectious Diseases, NIH</i>
10:30 – 11:00 AM	Coffee Break

11:00 – 12:00 PM	<b>When Lymphocytes go South, Very Very South</b> Kate Sullivan, M.D., Ph.D., <i>The Children's Hospital of Philadelphia</i>
12:00 – 1:00 PM	<b>Genetics and Genomics of Oxygen Homeostasis</b> 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	<b>What Happens in a Human Genetics Clinic</b> Ada Hamosh, Ph.D., <i>Johns Hopkins</i>
2:30 – 4:00 PM	<b>Human Genetics Clinic Presentations</b>

*Participants responsible for transportation. Island Explorer is available.*

7:00 – 8:30 PM	<b>Victor A. McKusick Memorial Lecture – Stone House at the Bar Harbor Regency</b> <b>A Diagnosis for all Rare Genetic Diseases: Collaborative Approaches to Discovery and Translation</b> Kym Boycott, M.D., Ph.D., <i>Children's Hospital of Eastern Ottawa</i>
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## Day 8: Wednesday, July 30<sup>th</sup>

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*Transportation to The Jackson Laboratory*

7:30 – 8:30 AM	Breakfast
8:30 – 9:30 AM	<b>Prenatal Screening Diagnosis &amp; Therapy</b> Ignatia Van den Veyver, M.D., <i>Baylor College of Medicine</i>
9:30 – 10:30 AM	<b>Building Precision Mouse Models of Rare Disease</b> Steve Murray, Ph.D., <i>The Jackson Laboratory</i>
10:30 – 11:00 AM	Coffee Break
11:00 – 12:00 PM	<b>Inborn Errors of Metabolism</b> Hilary Vernon, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</i>
12:00 – 1:00 PM	<b>Ethics and Policy Issues Raised by Genetics and Genomics</b> Debra Mathews, Ph.D., M.A., <i>Johns Hopkins University</i>
1:00 – 2:00 PM	Lunch Break
2:00 – 4:00 PM	<b>WORKSHOP: ClinGen, ClinVar, and other Genomic Curation Databases</b> Heidi Rehm, Ph.D., <i>Massachusetts General Hospital &amp; 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 31<sup>st</sup>

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*Transportation to The Jackson Laboratory*

7:30 – 8:30 AM      Breakfast

8:30 – 9:30 AM      **Precision Genome Editing Tools**  
Greg Newby, 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 – 12:00 PM      **Pharmacogenomics to Pharmaco-omics**  
Richard Weinshilboum, M.D., *Mayo Clinic*

12:00 – 1:00 PM      **Developing a DIA-Based Proteomics Approach to Decipher Protein Degradation and Synthesis Dynamics**  
Tian Zhang, Ph.D., *University of Virginia*

1:00 – 2:00 PM      Lunch Break

2:00 – 4:00 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      **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 1<sup>st</sup>

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*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**  
Stuart Orkin, M.D., *Harvard Medical School*

10:30 – 11:00 AM      Coffee Break

11:00 – 12:00 PM	<b>Implications of the First Successful Personalized Gene Editing Therapy</b> 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	<b>Genetic &amp; Pharmacologic Modifiers of Cystic Fibrosis</b> Garry Cutting, M.D., <i>Johns Hopkins University School of Medicine</i>
1:00 – 2:00 PM	<b>Course Summary and Wrap-Up</b>
2:00 – 2:30 PM	Lunch

*Participants responsible for transportation. Island Explorer is available*