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# Human and Mammalian Genetics and Genomics: The 65<sup>th</sup> 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 22<sup>nd</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	<b>Course Announcements &amp; General Housekeeping</b>
9:45 – 10:00 AM	<b>Welcome &amp; Opening Remarks</b> <i>Lon Cardon, Ph.D., FMedSci, The Jackson Laboratory</i>
10:00 – 11:00 AM	<b>History of Human Genetics</b> <i>David Valle, M.D., Johns Hopkins University School of Medicine</i>
11:00 AM – 12:00 PM	<b>Mice as Models for Human Diseases</b> <i>Greg Cox, Ph.D., The University of Maine</i>
12:00 – 1:30 PM	Lunch Break
1:30 – 2:30 PM	<b>Nuclear Genome Organization: Chromosome Structure and Regulation</b> <i>Jeanne Lawrence, Ph.D., University of Massachusetts Medical School</i>
2:30 – 3:30 PM	<b>Genetic Recombination &amp; Mutation</b> <i>Beth Dumont, Ph.D., The Jackson Laboratory</i>
3:30 – 3:45 PM	Coffee Break
3:45 – 4:45 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>

- 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 23<sup>rd</sup>

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*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 25<sup>th</sup>)
- 12:45 – 2:00 PM Lunch Break
- 2:00 – 3:00 PM **WORKSHOP: Intro to OMIM**  
 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: TBD**  
 Speaker TBD

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

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

## Day 3: Wednesday, July 24<sup>th</sup>

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*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      **TBD**  
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: Pangenome**  
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, Ph.D., *University of California San Francisco*

## Day 4: Thursday, July 25<sup>th</sup>

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*Transportation to The Jackson Laboratory*

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	<b>gnomAD: Human Genome Variation &amp; its Properties</b> Anne O’Donnell-Luria, M.D., Ph.D., <i>Broad Institute of MIT &amp; Harvard</i>
9:00 – 10:00 AM	<b>Systems Biology of the Cell</b> Aravinda Chakravarti, Ph.D., <i>NYU Langone Health</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	<b>TBD</b> Elissa Chesler, Ph.D., <i>The Jackson Laboratory</i>
11:30 AM – 12:30 PM	<b>TBD</b> 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	<b>WORKSHOP: WES/WGS Analysis</b> Nara Sobreira, M.D., Ph.D., <i>Johns Hopkins University</i>

4:00 – 5:30 PM Poster Session

*Transportation to COA/BH Regency*

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

**Day 5: Friday, July 26<sup>th</sup>**

*Transportation to The Jackson Laboratory*

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	<b>TBD</b> Jill Fahrner, Ph.D., <i>Johns Hopkins University School of Medicine</i>
9:00 – 10:00 AM	<b>TBD</b> Larry Brody, Ph.D., <i>National Institutes of Health</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	<b>TBD</b>

Olufumilayo Olopade, M.D., FACP, *UChicago Medicine*

11:30 AM – 12:30 PM **Cancer Biology Exposed Through Large-Scale Sequencing**  
Victor Velculescu, M.D., Ph.D., *Johns Hopkins University School of Medicine*

12:30 – 2:00 PM Lunch Break

2:00 – 4:00 PM **WORKSHOP: ClinGen, ClinVar and other Genomic Curation Databases**  
Heidi Rehm, Ph.D., *Massachusetts General Hospital & The Broad Institute*

4:15 – 5:45 PM **Chen K. Chai Memorial Lecture – The Jackson Laboratory Auditorium**  
**Tech Policy for Geneticists**  
Nancy Cox, Ph.D., *Vanderbilt University Medical School*

*Transportation to COA/ BH Regency*

**Saturday, July 27<sup>th</sup> & Sunday, July 28<sup>th</sup>**

*No Scheduled Course Activities*

## Day 6: Monday, July 29<sup>th</sup>

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*Transportation to The Jackson Laboratory*

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	<b>Human Evolution and Adaptation in Africa</b> Sarah Tishkoff, Ph.D., <i>University of Pennsylvania, Perelman School of Medicine</i>
9:00 – 10:00 AM	<b>Harnessing our Common African Genome to Improve Health Globally</b> Ambroise Wonkam, M.D., Ph.D., <i>Johns Hopkins University</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	<b>TBD</b> Segun Fatumo, Ph.D., <i>London School of Hygiene &amp; Tropical Medicine</i>
11:30 AM – 12:30 PM	<b>TBD</b> Dan Bauer, M.D., Ph.D., <i>Harvard Stem Cell Institute</i>
12:30 – 2:00 PM	<b>Trainee Lunch: Topic TBD</b> Alice Matimba, Ph.D., <i>Wellcome Connecting Science</i>
2:00 – 4:00 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>TBD</b> Charles Lee, Ph.D., <i>The Jackson Laboratory</i>
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## Day 7: Tuesday, July 30<sup>th</sup>

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*Transportation to The Jackson Laboratory*

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	<b>Genomics Characterization and Surveillance of Microbial Threats in West Africa</b> Christian Happi, Ph.D., <i>Redeemer's University</i>
9:00 – 10:00 AM	<b>TBD</b> Luigi Notarangelo, M.D., <i>National Institutes of Health</i>

10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	<b>Genetics of Infectious Diseases</b> Steven Holland, M.D., <i>National Institute of Allergy &amp; Infectious Diseases, NIH</i>
11:30 AM – 12:30 PM	<b>Inborn Errors of Metabolism: A Primer</b> 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	<b>WORKSHOP: Mouse Genome Informatics</b> Carol Bult, Ph.D., <i>The Jackson Laboratory</i>

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

7:00 – 8:30 PM	<b>Victor A. McKusick Memorial Lecture</b> – Stone House at the Bar Harbor Regency <b>TBD</b> Mary Claire King, Ph.D., <i>University of Washington</i>
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## Day 8: Wednesday, July 31<sup>st</sup>

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*Transportation to The Jackson Laboratory*

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	<b>TBD</b> Mary Dickinson, Ph.D., <i>Baylor College of Medicine</i>
9:00 – 10:00 AM	<b>Prenatal Screening Diagnosis &amp; Therapy</b> Ignatia Van den Veyver, M.D., <i>Baylor College of Medicine</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	<b>TBD</b> Cat Lutz, Ph.D., M.B.A., <i>The Jackson Laboratory</i>
11:30 AM – 12:30 PM	<b>Ethics and Policy Issues Raised by Genetics and Genomics</b> Debra Mathews, Ph.D., M.A., <i>Johns Hopkins University</i>
12:30 – 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      **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 1<sup>st</sup>

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*Transportation to The Jackson Laboratory*

7:30 – 8:00 AM      Breakfast

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

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

10:00 – 10:30 AM      Coffee Break

10:30 – 11:30 AM      **TBD**  
Adrian Krainer, Ph.D., *Cold Spring Harbor Laboratory*

11:30 AM – 12:30 PM      **TBD**  
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 2<sup>nd</sup>

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*Transportation to The Jackson Laboratory*

7:30 – 8:00 AM      Breakfast



8:00 – 9:00 AM	<b>Integrating Molecular Genetics Testing into Mainstream Medicine: View from a Commercial Laboratory</b> Robert Nussbaum, M.D., <i>Invitae</i>
9:00 – 10:00 AM	<b>TBD</b>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	<b>Precision Genome Editing Tools to Address Genetic Disease</b> Greg Newby, Ph.D., Broad Institute of MIT & Harvard
11:30 AM – 12:30 PM	<b>Genetic &amp; Pharmacologic Modifiers of Cystic Fibrosis</b> Garry Cutting, M.D., <i>Johns Hopkins University School of Medicine</i>
12:30 – 1:30 PM	<b>Course Summary and Wrap-Up</b>
1:30 – 2:30 PM	Lunch

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