



Human and Mammalian Genetics and Genomics: The 64th McKusick Short Course

July 17 – 28, 2023

All times listed are EDT (UTC – 4:00)

Schedule still subject to change.

Day 1: Monday, July 17th

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

- 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**
Gene Regulation, Selection, and the Genetic Architecture of Complex Traits
Jonathan Pritchard, Ph.D., *Stanford University*
- Transportation to Bar Harbor Regency – Stone House*
- 7:00 PM **Thomas H. Roderick Memorial Lecture Reception and Dinner**

Day 2: Tuesday, July 18th

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 **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-Cremens, Ph.D., *University of Pennsylvania*
- 12:30 – 12:45 PM Group Photo (Rain Date Wednesday, July 19th)
- 12:45 – 2:00 PM Lunch Break
- 2:00 – 2:30 PM **WORKSHOP: Intro to MGI**
Richard Baldarelli, Ph.D., *The Jackson Laboratory*
- 2:30 – 3:00 PM **WORKSHOP: Intro to OMIM**
Ada Hamosh, M.D., *Johns Hopkins University School of Medicine*

3:00 – 4:30 PM

WORKSHOP: All of US

Geoff Ginsburg, M.D., Ph.D., *National Institutes of Health & Genevieve Brandt, M.S, Broad Institute of MIT & Harvard**
**Presenting Remotely*

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM

Nature and Consequences of Genomic Variants – College of the Atlantic,
College of Human Ecology (CHE) Building
Stylianios Antonarakis, M.D., DSc, *University of Geneva Medical School*

Day 3: Wednesday, July 19th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM

Breakfast

8:00 – 9:00 AM

Advancing Genomic Technologies for Insights into Genome Organization and Transcription
Chia-Lin Wei, Ph.D., *The Jackson Laboratory for Genomic Medicine*

9:00 – 10:00 AM

gnomAD: Human Genome Variation & its Properties
Anne O'Donnell-Luria, M.D., Ph.D., *Broad Institute of MIT & Harvard*

10:00 – 10:30 AM

Coffee Break

10:30 – 11:30 AM

Every Biological Study will be Enlightened by an Accompanying Genome Sequence
Steven Scherer, Ph.D., *University of Toronto*

11:30 AM – 12:30 PM

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*

12:30 – 2:00 PM

Lunch Break

2:00 – 4:00 PM

WORKSHOP: The UCSC Genome Browser - Gateway to Genomic Data
Robert Kuhn, Ph.D., *UCSC (Retired)*

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM

Senior Lecturer – College of the Atlantic, CHE Building
Medicine, a Subspecialty of Genetics
Peter Byers, M.D., *UW Medicine*

Day 4: Thursday, July 20th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	Machine Learning for Regulatory Genomics: Basics and Pitfalls Michael Beer, Ph.D., <i>Johns Hopkins University</i>
9:00 – 10:00 AM	Probing Molecular Phenotypes with Quantitative Proteomics Devin Schweppe, Ph.D., <i>University of Washington</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	Mapping Complex Traits in Mice Gary Churchill, Ph.D., <i>The Jackson Laboratory</i>
11:30 AM – 12:30 PM	Model Organism Genetics and Human Disease: Models and Mechanisms to Therapies Phil Hieter, Ph.D., <i>University of British Columbia</i>
12:30 – 2:00 PM	Lunch Break
2:00 – 4:00 PM	WORKSHOP: RNA Sequencing Steven Munger, Ph.D., <i>The Jackson Laboratory</i>
4:00 – 5:30 PM	Poster Session

Transportation to COA/BH Regency

7:00 – 8:30 PM	Genetics and Genomics of Oxygen Homeostasis – College of the Atlantic, CHE Building Gregg Semenza, M.D., Ph.D., <i>Johns Hopkins University School of Medicine</i>
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Day 5: Friday, July 21st

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	Systems Biology of the Cell Aravinda Chakravarti, Ph.D., <i>NYU Langone Health</i>
9:00 – 10:00 AM	Inherited Susceptibility to Cancer William Foulkes, Ph.D., <i>McGill University</i>

10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	Using the Full Potential of Whole-Genome Sequencing in Cancer Genomics Serena Nik-Zainal, Ph.D., <i>University of Cambridge</i> <i>*Presenting Remotely</i>
11:30 AM – 12:30 PM	Cancer Biology Exposed Through Large-Scale Sequencing Victor Velculescu, 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: ClinGen, ClinVar and other Genomic Curation Databases Heidi Rehm, Ph.D., <i>Massachusetts General Hospital & The Broad Institute</i>
4:15 – 5:45 PM	Chen K. Chai Memorial Lecture – The Jackson Laboratory Auditorium Tech Policy for Geneticists Alondra Nelson, Ph.D., <i>Institute for Advanced Study</i> <i>Shuttle to Bar Harbor Regency Stone House</i>
6:00 – 9:00 PM	Chen K. Chai Memorial Lecture Reception and Dinner

Saturday, July 22nd & Sunday, July 23rd
No Scheduled Course Activities

Day 6: Monday, July 24th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
8:00 – 9:00 AM	Genetic and Genomic Analysis of Complex Traits and Diseases Benjamin Neale, Ph.D., <i>The Broad Institute and Massachusetts General Hospital</i>
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 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	Harnessing our Common African Genome to Improve Health Globally Ambroise Wonkam, M.D., Ph.D., <i>Johns Hopkins University</i>
11:30 AM – 12:00 PM	Post-traumatic Transgenerational Epigenetic Changes in Rwanda Leon Mutesa, M.D., Ph.D., <i>University of Rwanda</i>
12:00 – 12:30 PM	Complex Diseases Gene Mapping and Risk Prediction for African Populations Segun Fatumo, Ph.D., <i>London School of Hygiene and Tropical Medicine</i>
12:30 – 2:00 PM	Lunch Break
2:00 – 4:00 PM	WORKSHOP: WES/WGS Analysis Nara Sobreira, M.D., Ph.D., <i>Johns Hopkins University</i>

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM	Victor A. McKusick Memorial Lecture – College of the Atlantic, CHE Building Talk Title TBD Eric Olson, Ph.D., <i>UT Southwestern Medical Center</i>
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Day 7: Tuesday, July 25th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM	Breakfast
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- 8:00 – 9:00 AM **Genetics of Infectious Diseases**
Steven Holland, M.D., *National Institute of Allergy & Infectious Diseases, NIH*
- 9:00 – 10:00 AM **Genomics Characterization and Surveillance of Microbial Threats in West Africa**
Christian Happi, Ph.D., *Redeemer's University*
- 10:00 – 10:30 AM Coffee Break
- 10:30 – 11:30 AM **Inherited Defects of Adaptive Immunity: Advances in Diagnosis and Treatment**
Jennifer Puck, Ph.D., *University of California San Francisco*
**Presenting Remotely*
- 11:30 AM – 12:30 PM **The Human Genetic and Immunological Determinants of Life-Threatening COVID-19**
Jean-Laurent Casanova, M.D, Ph.D., *The Rockefeller University*
**Presenting Remotely*
- 12:30 – 2:00 PM Lunch Break
- Trainee Lunch & Learn – Staff Conference Room
International Outreach
Alice Matimba, Ph.D., *Wellcome Connecting Science*

2:00 – 4:00 PM **WORKSHOP: Mouse Genome Informatics**
Carol Bult, Ph.D., *The Jackson Laboratory*

Participants responsible for transportation. Island Explorer is available.

- 7:00 – 8:30 PM **Engineering T cells in vivo with mRNA** – College of the Atlantic, CHE Building
Jonathan Epstein, M.D., *University of Pennsylvania*

Day 8: Wednesday, July 26th

Transportation to The Jackson Laboratory

- 7:30 – 8:00 AM Breakfast
- 8:00 – 9:00 AM **Prenatal Screening Diagnosis & Therapy**
Ignatia Van den Veyver, M.D., *Baylor College of Medicine*

9:00 – 10:00 AM **MyCode: Enabling Genomic Discoveries and Moving Genomics into Everyday Care**
Juliann Savatt, M.S., C.G.C., *Geisinger*

10:00 – 10:30 AM Coffee Break

10:30 – 11:30 AM **Ethics and Policy Issues Raised by Genetics and Genomics**
Debra Mathews, Ph.D., MA, *Johns Hopkins University*

11:30 AM – 12:30 PM **GENOMICS FOR EVERYONE? AN INDIGENOUS PERSPECTIVE**
Krystal Tsosie, Ph.D., *Arizona State University*

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 **Panel: Using Population Descriptors in Genetics & Genomics Research –**
College of the Atlantic, Gates Auditorium
Aravinda Chakravarti, Ph.D., *NYU Langone Health*
Debra Mathews, Ph.D., MA, *Johns Hopkins University*
Sarah Tishkoff, Ph.D., *University of Pennsylvania, Perelman School of Medicine*

Day 9: Thursday, July 27th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM Breakfast

8:00 – 9:00 AM **Inborn Errors of Metabolism: A Primer**
Hilary Vernon, M.D., Ph.D., *Johns Hopkins University School of Medicine*

9:00 – 10:00 AM **Metabolomics and Human Diseases**
Ralph DeBerardinis, M.D., Ph.D., *University of Texas, Southwestern*

10:00 – 10:30 AM Coffee Break

10:30 – 11:30 AM **Pharmacogenomics to Pharmaco-omics, AI and ML**
Richard Weinshilboum, M.D., *Mayo Clinic*

11:30 AM – 12:30 PM **Integrating Molecular Genetics Testing into Mainstream Medicine: View from a Commercial Laboratory**
Robert Nussbaum, M.D., *Invitae*
**Presenting Remotely*

12:30 – 1:00 PM **The Jackson Laboratory Rare Disease Translational Center**
Cat Lutz, Ph.D., M.B.A., *The Jackson Laboratory*

1:00 – 2:00 PM Lunch Break

1:00 – 3:00 PM **Skippy Lane Memorial Mouse Clinic**
Greg Cox, Ph.D., *The Jackson Laboratory*

Participants responsible for transportation. Island Explorer is available.

7:00 – 8:30 PM **ELSI Case Studies** – College of the Atlantic, Gates Auditorium
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., *Harvard University*

Day 10: Friday, July 28th

Transportation to The Jackson Laboratory

7:30 – 8:00 AM Breakfast

8:00 – 9:00 AM **Gene Therapy Approaches to Hemoglobinopathies**
Stuart Orkin, M.D., *Harvard Medical School*

9:00 – 10:00 AM **Early Forays in Individualized Genetic Medicine**
Timothy Yu, M.D., Ph.D., *Harvard Medical School & Boston Children's Hospital*
**Presenting Remotely*

10:00 – 10:30 AM Coffee Break

10:30 – 11:30 AM **Precision Genome Editing Tools to Address Genetic Disease**
Greg Newby, Ph.D., *Broad Institute of MIT & Harvard*

11:30 AM – 12:30 PM **Genetic & Pharmacologic Modifiers of Cystic Fibrosis**
Garry Cutting, M.D., *Johns Hopkins University School of Medicine*

12:30 – 2:00 PM Lunch Break

2:00 – 3:00 PM

**Insights into the Pathogenesis and Treatment of Vascular Connective
Tissue Disorders**

Harry 'Hal' Dietz, M.D., *Johns Hopkins University School of Medicine*

3:00 – 4:00 PM

Course Summary and Wrap-Up

Participants responsible for transportation. Island Explorer is available.