



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

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 – Gene Regulation, Selection, and the Genetic Architecture of Complex Traits <i>Jonathan Pritchard, Ph.D., Stanford University</i>

Day 2: Tuesday, July 18th

- 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-Cremins, Ph.D., *University of Pennsylvania*
- 7:00 – 8:30 PM **Nature and Consequences of Genomic Variants –**
Stylianos Antonarakis, M.D., DSc, *University of Geneva Medical School*
This will not be live streamed. It will be recorded and posted on canvas within a week of airing.

Day 3: Wednesday, July 19th

- 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*

7:00 – 8:30 PM **Senior Lecturer**
Medicine, a Subspecialty of Genetics
Peter Byers, M.D., *UW Medicine*
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Day 4: Thursday, July 20th

8:00 – 9:00 AM **Machine Learning for Regulatory Genomics: Basics and Pitfalls**
Michael Beer, Ph.D., *Johns Hopkins University*

9:00 – 10:00 AM **Probing Molecular Phenotypes with Quantitative Proteomics**
Devin Schweppe, Ph.D., *University of Washington*

10:00 – 10:30 AM Coffee Break

10:30 – 11:30 AM **Mapping Complex Traits in Mice**
Gary Churchill, Ph.D., *The Jackson Laboratory*

11:30 AM – 12:30 PM **Model Organism Genetics and Human Disease: Models and Mechanisms to Therapies**
Phil Hieter, Ph.D., *University of British Columbia*

7:00 – 8:30 PM **Genetics and Genomics of Oxygen Homeostasis**
Gregg Semenza, M.D., Ph.D., *Johns Hopkins University School of Medicine*
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Day 5: Friday, July 21st

8:00 – 9:00 AM **Systems Biology of the Cell**
Aravinda Chakravarti, Ph.D., *NYU Langone Health*

9:00 – 10:00 AM **Inherited Susceptibility to Cancer**
William Foulkes, Ph.D., *McGill University*

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., *University of Cambridge*
**Presenting Remotely*

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*

4:15 – 5:45 PM **Chen K. Chai Memorial Lecture –
Tech Policy for Geneticists**
Alondra Nelson, Ph.D., *Institute for Advanced Study*

Saturday, July 22nd & Sunday, July 23rd

No Scheduled Course Activities

Day 6: Monday, July 24th

8:00 – 9:00 AM **Genetic and Genomic Analysis of Complex Traits and Diseases**
Benjamin Neale, Ph.D., *The Broad Institute and Massachusetts General Hospital*

9:00 – 10:00 AM **Human Evolution and Adaptation in Africa**
Sarah Tishkoff, Ph.D., *University of Pennsylvania, Perelman School of Medicine*

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., *Johns Hopkins University*

11:30 AM – 12:00 PM **Post-traumatic Transgenerational Epigenetic Changes in Rwanda**
Leon Mutesa, M.D., Ph.D., *University of Rwanda*

12:00 – 12:30 PM **Complex Diseases Gene Mapping and Risk Prediction for African Populations**
Segun Fatumo, Ph.D., *London School of Hygiene and Tropical Medicine*

7:00 – 8:30 PM **Victor A. McKusick Memorial Lecture**
Talk Title TBD
Eric Olson, Ph.D., *UT Southwestern Medical Center*
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Day 7: Tuesday, July 25th

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*
- 7:00 – 8:30 PM **Engineering T cells in vivo with mRNA**
Jonathan Epstein, M.D., *University of Pennsylvania*
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Day 8: Wednesday, July 26th

- 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*

Day 9: Thursday, July 27th

- 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., <i>University of Texas, Southwestern</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	Pharmacogenomics to Pharmaco-omics, AI and ML Richard Weinshilboum, M.D., <i>Mayo Clinic</i>
11:30 AM – 12:30 PM	Integrating Molecular Genetics Testing into Mainstream Medicine: View from a Commercial Laboratory Robert Nussbaum, M.D., <i>Invitae</i> <i>*Presenting Remotely</i>
12:30 – 1:00 PM	The Jackson Laboratory Rare Disease Translational Center Cat Lutz, Ph.D., M.B.A., <i>The Jackson Laboratory</i>

Day 10: Friday, July 28th

8:00 – 9:00 AM	Gene Therapy Approaches to Hemoglobinopathies Stuart Orkin, M.D., <i>Harvard Medical School</i>
9:00 – 10:00 AM	Early Forays in Individualized Genetic Medicine Timothy Yu, M.D., Ph.D., <i>Harvard Medical School & Boston Children's Hospital</i>
10:00 – 10:30 AM	Coffee Break
10:30 – 11:30 AM	Precision Genome Editing Tools to Address Genetic Disease Greg Newby, Ph.D., <i>Broad Institute of MIT & Harvard</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 – 2:00 PM	Lunch Break
2:30 – 3:30 PM	Insights into the Pathogenesis and Treatment of Vascular Connective Tissue Disorders Harry 'Hal' Dietz, M.D., <i>Johns Hopkins University School of Medicine</i>
3:30 – 4:30 PM	Course Summary and Wrap-Up

