
Human and Mammalian Genetics and Genomics: The 63rd McKusick Short Course

VIRTUAL SCHEDULE

Schedule subject to change – Times are listed in EDT – Zoom links accessible via Canvas

Day 1: Monday, July 18th

10:15 – 10:30 AM	Welcome & Opening Remarks Lon Cardon, Ph.D., FMedSci, <i>The Jackson Laboratory</i>
10:30 – 11:20 AM	History of Human Genetics David Valle, M.D., <i>Johns Hopkins University School of Medicine</i>
11:20 AM – 12:10 PM	Mice as Models for Human Diseases Greg Cox, Ph.D., <i>The Jackson Laboratory</i>
12:10 – 1:00 PM	Opportunities and Limits in the 21st Century Mouse Model System Steven Munger, Ph.D., <i>The Jackson Laboratory</i>
1:00 – 2:00 PM	Lunch Break
2:00 – 2:50 PM	Nuclear Genome Organization: Chromosome Structure and Regulation Jeanne Lawrence, Ph.D., <i>University of Massachusetts Medical School</i>
2:50 – 3:00 PM	Break
3:00 – 3:45 PM	Panel Discussion + Q&A David Valle, Greg Cox, Steven Munger & Jeanne Lawrence
3:45 – 4:00 PM	Coffee Break
4:00 – 5:30 PM	Thomas H. Roderick Memorial Lecture Genetics Basis of Adaptation in (Wild) Mice Hopi Hoekstra, Ph.D., <i>Harvard University</i> <i>Introduction by Lon Cardon, Ph.D., FMedSci, The Jackson Laboratory</i>

Day 2: Tuesday, July 19th

- 10:30 – 11:20 AM **Discovering New Biology and Function in Complete, Telomere-to-Telomere (T2T) Genomes**
Karen Miga, Ph.D., *University of California, Santa Cruz*
- 11:20 AM – 12:10 PM **Transcriptional Regulation of Gene Expression**
Andrew McCallion, Ph.D., *Johns Hopkins University School of Medicine*
- 12:10 – 1:00 PM **Single Cell Analysis in Development and Disease**
Loyal Goff, Ph.D., *Johns Hopkins University School of Medicine*
- 1:00 – 2:00 PM Lunch Break
- 2:00 – 2:50 PM **A Decade of Technology-Drive Insights Into the 3D Genome's Structure-Function Relationship**
Jennifer Phillips-Cremins, Ph.D., *University of Pennsylvania*
- 2:50 – 3:00 PM Break
- 3:00 – 3:45 PM **Panel Discussion + Q&A**
Karen Miga, Andrew McCallion, Loyal Goff & Jennifer Phillips-Cremins
- 3:45 – 4:00 PM Coffee Break
- 4:00 – 5:30 PM **Human Genome Variants**
Stylianos Antonarakis, M.D., DSc, *University of Geneva Medical School*

Day 3: Wednesday, July 20th

- 10:30 – 11:20 AM **gnomAD: Human Genome Variation & its Properties**
Anne O'Donnell-Luria, M.D., Ph.D., *Broad Institute of MIT & Harvard*
- 11:20 AM – 12:10 PM **Human Structural Variation**
Charles Lee, Ph.D., *The Jackson Laboratory for Genomic Medicine*
- 12:10 – 1:00 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*
- 1:00 – 2:00 PM Lunch Break
- 2:00 – 2:50 PM **Machine Learning for Regulatory Genomics: Basics and Pitfalls**
Michael Beer, Ph.D., *Johns Hopkins University*

2:50 – 3:00 PM	Break
3:00 – 3:45 PM	Panel Discussion + Q&A Anne O’Donnell-Luria, Charles Lee, Hans Bjornsson & Michael Beer
3:45 – 4:00 PM	Coffee Break
4:00 – 5:30 PM	Genetics and Genomics of Oxygen Homeostasis Gregg Semenza, M.D., Ph.D., <i>Johns Hopkins University</i>

Day 4: Thursday, July 21st

10:30 – 11:20 AM	Genetic and Genomic Analysis of Complex Traits and Diseases Benjamin Neale, Ph.D., <i>The Broad Institute and Massachusetts General Hospital</i>
11:20 – 11:35 AM	Break
11:35 AM – 12:25PM	Population Genetics & Genomics Guy Sella, Ph.D., <i>Columbia University</i>
12:25 – 1:15 PM	Genetic Dissection of Complex Traits in Mice Greg Carter, Ph.D., <i>The Jackson Laboratory</i>
1:15 – 2:00 PM	Lunch Break
2:00 – 2:50 PM	Genetics of Infectious Diseases Steven Holland, M.D., <i>National Institute of Allergy & Infectious Diseases, NIH</i> <i>*Presenting Remotely</i>
2:50 – 3:00 PM	Break
3:00 – 3:45 PM	Panel Discussion + Q&A Guy Sella, Greg Carter, and Steven Holland
3:45 – 4:00 PM	Coffee Break
4:00 – 5:30 PM	Senior Lecturer: The Genetics of Hearing: More Than a Thousand Ways to be Deaf or Hard-of-Hearing Cynthia Morton, Ph.D., <i>Harvard Medical School & Brigham and Women’s Hospital</i>

Day 5: Friday, July 22nd

- 10:30 – 11:20 AM **Systems Biology of the Cell**
Aravinda Chakravarti, Ph.D., *NYU Langone Health*
- 11:20 AM – 12:10 PM **The Human Genetic and Immunological Determinants of Life-Threatening COVID-19**
Jean-Laurent Casanova, M.D, Ph.D., *The Rockefeller University*
**Presenting remotely*
- 12:10 – 12:25 PM Break
- 12:25 – 1:15 PM **SARS-CoV-2 Genomic Integration in Human and Bat PSC-Derived Cell Types**
Rudolf Jaenisch, M.D., *Whitehead Institute, MIT*
- 1:15 – 2:00 PM Lunch Break
- 2:00 – 2:50 PM **Pharmacogenomics to Pharmaco-omics, AI and ML**
Richard Weinshilboum, M.D., *Mayo Clinic*
- 2:50 – 3:00 PM Break
- 3:00 – 3:45 PM **Panel Discussion + Q&A**
Aravinda Chakravarti, Jean-Laurent Casanova, Rudolf Jaenisch & Richard Weinshilboum
- 3:45 – 4:00 PM Coffee Break
- 4:00 – 5:30 PM **Chen K. Chai Memorial Lecture**
Archaic Genomics
Svante Pääbo, Ph.D., *Max Planck Institute for Evolutionary Anthropology*
**Presenting remotely*
Introduction by Aravinda Chakravarti, Ph.D., NYU Langone Health

Day 6: Saturday, July 23rd

- 8:00 – 8:50 AM **Precision Medicine**
Dan Roden, M.D., *Vanderbilt University Medical Center*
- 8:50 – 9:40 AM **NHLBI Trans-Omics for Precision Medicine: Lessons and Opportunities**
Rasika Mathias, Sc.D., *John Hopkins University School of Medicine*
- 9:40 – 10:10 AM Coffee Break

- 10:10 – 11:00 AM **Cancer Biology Exposed by Genomic Sequencing**
Victor Velculescu, M.D, Ph.D, *John Hopkins University School of Medicine*
- 11:00 – 11:15 AM Break
- 11:15 AM – 12:05 PM **Using the Full Potential of Whole-Genome Sequencing in Cancer Genomics**
Serena Nik-Zainal, Ph.D., *University of Cambridge*
**Presenting Remotely*

Day 7: Monday, July 25th

- 10:00 – 10:50 AM **Human Evolution and Adaptation in Africa**
Sarah Tishkoff, Ph.D., *University of Pennsylvania, Perelman School of Medicine*
- 10:50 – 11:40 AM **Human Genetic Diversity Across Latin America and the Pacific**
Andrés Moreno-Estrada, M.D, Ph.D., *Cinvestav UGA-Langebio*
- 11:40 AM – 12:30 PM **Investigation of Genetic Diseases in North Africa: Lessons Learnt and Perspectives**
Sonia Abdelhak, Ph.D., *Institut Pasteur de Tunis*
**Presenting remotely*
- 12:30 – 1:30 PM Lunch Break
- 1:30 – 2:20 PM **Genomics Characterization and Surveillance of Microbial Threats in West Africa**
Christian Happi, Ph.D., *Redeemer's University*
- 2:20 – 2:30 PM Break
- 2:30 – 3:45 PM **Diversity in Genetics Panel Discussion**
Ambroise Wonkam, M.D., Ph.D., *Johns Hopkins University*
Christian Happi, Ph.D., *Redeemer's University*
Laura Arbour, MSc, MD, FRCPC, FCCMG, *University of Victoria*
Andrés Moreno-Estrada, M.D, Ph.D., *Cinvestav UGA-Langebio*
Ghada El-Kamah, M.D, Ph.D., *National Research Center, Cairo, Egypt*
Guida Landoure, Ph.D., *University of Science, Technique and Technology of Bamako, Mali*
- Introduction:
Harnessing African Genomic Variations to Improve Generic Medicine Globally
Ambroise Wonkam, M.D., Ph.D., *Johns Hopkins University*

- 3:45 – 4:00 PM Coffee Break
- 4:00 – 5:30 PM **Silent Genomes: Building the Path to Equitable Genomics Care for Indigenous Patients in Canada**
Laura Arbour, MSc, MD, FRCPC, FCCMG, *University of Victoria*

Day 8: Tuesday, July 26th

- 10:30 – 11:20 AM **Metabolomics and Disease Phenotypes**
Ralph DeBerardinis, M.D., Ph.D., *University of Texas, Southwestern*
- 11:20 – 11:35 AM Break
- 11:35 AM – 12:25 PM **Inborn Errors of Metabolism: A Primer**
Hilary Vernon, M.D., Ph.D., *Johns Hopkins University*
- 12:25 – 1:15 PM **Probing Molecular Phenotypes with Quantitative Proteomics**
Devin Schweppe, Ph.D., *University of Washington*
- 1:15 – 2:00 PM Lunch Break
- 2:00 – 2:50 PM **Drug Discovery for Rare Diseases**
Lon Cardon, Ph.D., FMedSci, *The Jackson Laboratory*
- 2:50 – 3:00 PM Break
- 3:00 – 3:45 PM **Panel Discussion + Q&A**
Hilary Vernon, Ralph DeBerardinis, Devin Schweppe & Lon Cardon
- 3:45 – 4:00 PM Coffee Break
- 4:00 – 5:30 PM **Victor A. McKusick Memorial Lecture**
Learning from Communities
Wylie Burke, M.D., Ph.D., *University of Washington*
Introduction by David Valle, M.D., Johns Hopkins University

Day 9: Wednesday, July 27th

- 10:30 – 11:20 AM **Prenatal Screening Diagnosis & Therapy**
Ignatia Van den Veyver, M.D., *Baylor College of Medicine*
- 11:20 AM – 12:10 PM **The Genetics and Clinical Phenotypes of Inherited Skeletal Dysplasias**
Julie Hoover-Fong, M.D., Ph.D., *Johns Hopkins University School of Medicine*

12:10 – 1:00 PM **CRISPR: Ethics and Governance of Germline Genome Editing**
Debra Mathews, Ph.D., MA, *Johns Hopkins University*

Day 10: Thursday, July 28th

10:30 – 11:20 AM **Gene Therapy Approaches to Hemoglobinopathies**
Stuart Orkin, M.D., *Harvard Medical School*

11:20 AM – 12:10 PM **Early Forays in Individualized Genetic Medicine**
Timothy Yu, M.D., Ph.D., *Harvard Medical School & Boston Children's Hospital*
**Presenting remotely*

12:10 – 1:00 PM **Precision Genome Editing Tools to Address Genetic Disease**
Greg Newby, Ph.D., *Broad Institute of MIT & Harvard*

1:00 – 2:00 PM Lunch Break

2:00 – 2:50 PM **Genetic & Pharmacologic Modifiers of Cystic Fibrosis**
Garry Cutting, M.D., *Johns Hopkins University*
**Presenting remotely*

2:50 – 3:00 PM Break

3:00 – 3:45 PM **Panel Discussion + Q&A**
Stuart Orkin, Timothy Yu, Greg Newby and Garry Cutting

3:45 – 4:00 PM Coffee Break

4:00 – 5:30 PM **Insights into the Pathogenesis and Treatment of Vascular Connective Tissue Disorders**
Harry 'Hal' Dietz, M.D., *Johns Hopkins University*

5:30 – 5:45 PM **Course Summary and Wrap-Up**
Cynthia Morton, Ph.D., *Harvard Medical School & Brigham and Women's Hospital*