
VIRTUAL:
Human and Mammalian Genetics and Genomics:
The 62nd McKusick Short Course

July 19 - 29, 2021

Schedule is Subject to Change

Pre-Recorded Content (4 hours)

The following pre-recorded, introductory lectures will be available on the Canvas site two weeks prior to the course. These are intended to be supplementary primers or refreshers, however, we heavily encourage you to watch these prior to the course.

Course Welcome (10 min)

Edison Liu, M.D., *The Jackson Laboratory*

An Introduction to Mouse Genome Informatics (30 min)

David Shaw, M.S., *The Jackson Laboratory*

Intro to OMIM (30 min)

Ada Hamosh, M.D., *Johns Hopkins University School of Medicine*

Molecular Genetics (1 hr)

John A. Phillips, III., M.D., *Vanderbilt University School of Medicine*

Epigenetics (1 hr)

Hans Bjornsson, M.D., Ph.D., *Johns Hopkins University School of Medicine, University of Iceland*

Basic Mendelian Genetics (1 hr)

Carolyn Applegate, MGC, CGC, *Johns Hopkins University Institute of Genetic Medicine*

Day 1: Monday, July 19th
History of Mammalian Genetics and the Evolution of Concepts

Morning Moderator: *Nadia Rosenthal, The Jackson Laboratory*

10:00 am EDT **History of Human Genetics**
David Valle, M.D., *Johns Hopkins University School of Medicine*

10:50 am EDT **Mice as Models for Human Diseases**
Greg Cox, Ph.D., *The Jackson Laboratory*

11:40 pm EDT **Opportunities and Limits in the 21st Century Mouse Model System**
Steve Munger, Ph.D., *The Jackson Laboratory*

12:30 pm EDT **Lunch Break (30 min)**

Afternoon Moderator: *Charles Lee, The Jackson Laboratory*

1:00 pm EDT **Genetic Recombination & Mutation**
Beth Dumont, Ph.D., *The Jackson Laboratory*

1:50 pm EDT **Chromosome Structure and Regulation**
Jeanne Lawrence, Ph.D., *University of Massachusetts Medical School*

2:40 pm EDT **Break (20 min)**

3:00 pm EDT **Panel Discussion + Q&A**
David Valle, Greg Cox, Steve Munger, Beth Dumont, & Jeanne Lawrence

Evening Moderator: *Aravinda Chakravarti, NYU Langone Health*

4:00 – **Thomas H. Roderick Memorial Lecture**
5:30 pm EDT **Mapping the Human Gastrointestinal Tract Through Space and Time**
Sarah Teichmann, F.Med.Sci, FRS, *Wellcome Sanger Institute*

Day 2: Tuesday, July 20th
DNA and its Variation

8:30 am EDT **WORKSHOP: UCSC Genome Browser**
Robert Kuhn, Ph.D., *University of California Santa Cruz*

Morning Moderator: Aravinda Chakravarti, *NYU Langone Health*

10:00 am EDT **gnomAD: Human Genome Variation & its Properties**
Anne O'Donnell-Luria, M.D., Ph.D., *Broad Institute of MIT & Harvard*

10:50 am EDT **Human Structural Variation**
Charles Lee, Ph.D., *The Jackson Laboratory for Genomic Medicine*

11:40 am EDT **Telomere-to-Telomere Chromosome Assemblies: New Insights into Genome Biology and Structure**
Karen Miga, Ph.D., *University of California, Santa Cruz*

12:30 pm EDT **Lunch Break (30 min)**

Afternoon Moderator: Greg Cox, *The Jackson Laboratory*

1:00 pm EDT **The ENCODE Project**
John Stamatoyannopoulos, M.D., *Altius Institute for Biomedical Sciences and Dept. of Genome Sciences University of Washington*

1:50 pm EDT **Organizing Principles of the 3D Epigenome**
Jennifer Phillips-Cremins, Ph.D., *University of Pennsylvania*

2:40 pm EDT **Break (20 min)**

3:00 pm EDT **Panel Discussion + Q&A**
Anne O'Donnell-Luria, Charles Lee, Karen Miga, John Stamatoyannopoulos & Jennifer Phillips-Cremins

Evening Moderator: Greg Cox, *The Jackson Laboratory*

4:00 –
5:30 pm EDT **Nature and Consequences of Human Genome Variants**
Stylianos Antonarakis, M.D., DSc, *University of Geneva Medical School*

Day 3: Wednesday, July 21st *Gene Expression & Regulation*

8:30 am EDT **WORKSHOP: WES/WGS**
Nara Sobreira, M.D., Ph.D., *Johns Hopkins University*

Morning Moderator: Aravinda Chakravarti, *NYU Langone Health*

10:00 am EDT **Transcriptional Regulation of Gene Expression**
Andrew McCallion, Ph.D., *Johns Hopkins University School of Medicine*

10:50 am EDT **Single Cell Analysis in Development and Disease**
Loyal Goff, Ph.D., *Johns Hopkins University School of Medicine*

11:40 am EDT **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 pm EDT **Lunch Break (30 min)**

Afternoon Moderator: David Valle, *Johns Hopkins University*

1:00 pm EDT **Colliding Ribosomes at the Heart of Cellular Signaling Pathways**
Rachel Green, Ph.D., *Johns Hopkins University School of Medicine*

1:50 pm EDT **Mapping Dynamic Proteomes**
Devin Schweppe, Ph.D., *University of Washington*

2:40 pm EDT **Break (20 min)**

3:00 pm EDT **Panel Discussion + Q&A**
Andrew McCallion, Loyal Goff, Hans Bjornsson, Rachel Green, and Devin Schweppe

Evening Moderator: David Valle, *Johns Hopkins University*

4:00 –
5:30 pm EDT **Victor A. McKusick Memorial Lecture**
Genetics of Neuropsychiatric Disorders: Lessons from Rett Syndrome and Other MECP2 Disorders
Huda Zoghbi, M.D., *Baylor College of Medicine; HHMI; Texas Children's Hospital Jan and Dan Duncan Neurological Research Institute*

Day 4: Thursday, July 22nd
Population Genetics & Genetic Architecture

8:30 am EDT **WORKSHOP: Standardizing Phenotyping: Human, Mouse & Cross-Species Comparison**
Peter Robinson, M.D., M.S., *The Jackson Laboratory*

Morning Moderator: Greg Cox, *The Jackson Laboratory*

10:00 am EDT **Systems Biology of the Cell**
Aravinda Chakravarti, Ph.D., *NYU Langone Health*

10:50 am EDT **Population Genetics & Genomics**
Guy Sella, Ph.D., *Columbia University*

11:40 am EDT **Genome Analysis of Diverse Populations**
Neil Hanchard, M.D., Ph.D., *National Human Genome Research Institute*

12:30 pm EDT **Lunch Break (30 min)**

Afternoon Moderator: Nadia Rosenthal, *The Jackson Laboratory*

1:00 pm EDT **Genetic Dissection of Complex Traits in Mice**
Gary Churchill, Ph.D., *The Jackson Laboratory*

1:50 pm EDT **Machine Learning for Regulatory Genomics: Basics and Pitfalls**
Michael Beer, Ph.D., *Johns Hopkins University*

2:40 pm EDT **Break (20 min)**

3:00 pm EDT **Panel Discussion + Q&A**
Aravinda Chakravarti, Guy Sella, Neil Hanchard, Michael Beer & Gary Churchill

Evening Moderator: Charles Lee *The Jackson Laboratory*

4:00 –
5:30 pm EDT **Genetics and Genomics of Oxygen Homeostasis**
Gregg Semenza, M.D., Ph.D., *Johns Hopkins University*

Day 5: Friday, July 23rd
Human Evolution & its Principles

Morning Moderator: Ada Hamosh, *Johns Hopkins University*

8:30 am EDT **African Genetics**
Sarah Tishkoff, Ph.D., *University of Pennsylvania*

9:30 am EDT **African Genetics Panel - Six short talks, followed by a panel discussion**

African Genomic Variations: The Next Frontier of Global Genetic Medicine
Ambroise Wonkam, Ph.D., *University of Cape Town*

Opportunity for Rare Disease Research in Africa
Guida Landoure, M.D., Ph.D., *USTTB*

Genomic Characterization and Surveillance of Microbial Threats in West Africa
Christian Happi, Ph.D., *ACEGID & Redeemer's University*

Overview of Novel Gene Discoveries in Africa
Sonia Abdelhak, Ph.D., *Institut Pasteur de Tunis*

Epigenetics & African Genomics
Neil Hanchard, M.D., Ph.D., *National Human Genome Research Institute*

Addressing Equity in Genomic Research in Africa
Charmaine Royal, Ph.D., *Duke University*

11:30 am EDT **Lunch Break (1hr 30 min)**

Afternoon Moderator: Greg Cox, *The Jackson Laboratory*

1:00 pm EDT **Ancient DNA & Human Evolution**
David Reich, Ph.D., *Harvard Medical Institute*

1:50 pm EDT **Global Human Diversity**
Graham Coop, Ph.D., *University of California, Davis*

2:40 pm EDT **Break (20 min)**

3:00 pm EDT **Panel Discussion + Q&A**
David Reich, Sarah Tishkoff & Graham Coop

Evening Moderator: Aravinda Chakravarti, *NYU Langone Health*

4:00 – **Chen K. Chai Lecture**

5:30 pm EDT **Moving From Hammer to Nail: Genomics and Health Outcomes in 2021**
Katrina Armstrong, M.D., *Massachusetts General Hospital*

Weekend Pre-Recorded Content (4.5 hours)

The following pre-recorded lectures will be available on the Canvas site upon login. They are intended to be watched on Saturday, July 24th, or before Monday, July 26th.

Treatment of Genetic Disease (1 hr)

David Valle, M.D., *Johns Hopkins University School of Medicine*

Sequencing for Deciphering Mendelian Disease (1 hr)

Gilad Evrony, M.D., Ph.D., *NYU Langone Health*

Organoids to Model Human Disease (1 hr)

Hans Clevers, M.D., Ph.D., *Hubrecht Institute*

What happens in a Medical Genetics Clinic? (30 min)

Ada Hamosh, M.D., MPH, *Johns Hopkins University School of Medicine*

Julie Hoover-Fong, M.D., Ph.D., *Johns Hopkins University School of Medicine*

Genetics of Autoimmunity (1 hr)

David Serreze, Ph.D., *The Jackson Laboratory*

Day 6: Monday, July 26th
Cellular & Molecular Basis of Disease

8:30 am EDT **WORKSHOP: RNA Sequencing**
Steven Munger, Ph.D., *The Jackson Laboratory*

Morning Moderator: Nadia Rosenthal, *The Jackson Laboratory*

10:00 am EDT **Immune Responses to SARS-CoV-2**
Akiko Iwasaki, Ph.D., *Yale School of Medicine & Howard Hughes Medical Institute*

10:50 am EDT **Genetics of Infectious Diseases**
Steve Holland, M.D., *National Institute of Allergy & Infectious Diseases*

11:40 am EDT **Genetic Dissection of IBD, its Pathogenesis & Treatment**
Judy Cho, M.D., *Icahn School of Medicine at Mount Sinai*

12:30 pm EDT **Lunch Break (30 min)**

Afternoon Moderator: Ada Hamosh, *Johns Hopkins University*

1:00 pm EDT **Genetics and Genetic Epidemiology of Breast and Ovarian Cancer**
Lawrence Brody, Ph.D., *National Human Genome Research Institute*

1:50 pm EDT **Cancer Biology Exposed Through Large-Scale Sequencing**
Victor Velculescu, M.D., Ph.D., *Johns Hopkins University School of Medicine*

2:40 pm EDT **Break (20 min)**

3:00 pm EDT **Panel Discussion + Q&A**
Akiko Iwasaki, Steve Holland, Judy Cho, Lawrence Brody & Victor Velculescu

Evening Moderator: Ada Hamosh, *Johns Hopkins University*

4:00 – **Variation, Standardization & Data Sharing**
5:30 pm EDT Heidi Rehm, Ph.D., *Massachusetts General Hospital & The Broad Institute*

Day 7: Tuesday, July 27th
Genotype-Phenotype Correlations: Pathophysiology

8:30 am EDT **WORKSHOP: ClinGen & ClinVar**
Heidi Rehm, Ph.D., *Massachusetts General Hospital & The Broad Institute*

Morning Moderator: Ada Hamosh, *Johns Hopkins University*

10:00 am EDT **Inborn Errors of Metabolism: A Primer**
Hilary Vernon, M.D., Ph.D., *Johns Hopkins University*

10:50 am EDT **Metabolomics and Disease Phenotypes**
Ralph DeBerardinis, M.D., Ph.D., *University of Texas, Southwestern*

11:40 am EDT **Genetic & Pharmacologic Modifiers of Cystic Fibrosis**
Garry Cutting, M.D., *Johns Hopkins University*

12:30 pm EDT **Lunch Break (30 min)**

Afternoon Moderator: Aravinda Chakravarti, *NYU Langone Health*

1:00 pm EDT **Medical Consequences of Human Diversity**
David Goldstein, Ph.D., *Columbia University*

1:50 pm EDT **Host Genetics and Infectious Disease**
Priya Duggal, Ph.D., *Johns Hopkins University School of Medicine*

2:40 pm EDT **Break (20 min)**

3:00 pm EDT **Panel Discussion + Q&A**
Hilary Vernon, Ralph DeBerardinis, Garry Cutting, David Goldstein & Priya Duggal

Moderated by Aravinda Chakravarti & Ada Hamosh

Evening Moderator: David Valle, *Johns Hopkins University*

4:00 – **How de novo Mutations Disrupt Developmental Programs**
5:30 pm EDT David FitzPatrick, M.D., *The University of Edinburgh Institute of Genetics*

Day 8: Wednesday, July 28th
Genome Variation, Precision Medicine & Society

8:30 am EDT **WORKSHOP: Mouse Genomics**
Richard Baldarelli, Ph.D., Carol Bult, Ph.D., and Martin Ringwald, Ph.D., *The Jackson Laboratory*

Morning Moderator: David Valle, *Johns Hopkins University*

10:00 am EDT **Precision Medicine**
Dan Roden, M.D., *Vanderbilt University Medical Center*

10:50 am EDT **Pharmacogenomics**
Richard Weinshilboum, M.D., *Mayo Clinic*

11:40 am EDT **Prenatal Screening Diagnosis & Therapy**
Ignatia Van den Veyver, M.D., *Baylor College of Medicine*

12:30 pm EDT **Lunch Break (30 min)**

Afternoon Moderator: David Valle, *Johns Hopkins University*

1:00 pm EDT **Maximizing the Usability of Genomics For All**
Vence Bonham Jr., J.D., *National Human Genome Research Institute*

1:50 pm EDT **CRISPR: Ethics & Governance of Germline Genome Editing**
Debra Mathews, Ph.D., MA, *Johns Hopkins University*

2:40 pm EDT **Break (20 min)**

3:00 pm EDT **Panel Discussion + Q&A**
Dan Roden, Richard Weinshilboum, Vence Bonham Jr., Ignatia Van den Veyver & Debra Mathews

Evening Moderator: Charles Lee, *The Jackson Laboratory*

4:00 –
5:30 pm EDT **Humanizing Drug Discovery**
David Altshuler, M.D., Ph.D., *Vertex Pharmaceuticals*

Day 9: Thursday, July 29th
Genetic Therapies

8:30 am EDT **WORKSHOP: Skippy Lane Memorial Mouse Clinic**
Greg Cox, Ph.D., *The Jackson Laboratory*

Morning Moderator: Charles Lee, *The Jackson Laboratory*

10:00 am EDT **Antisense Modulation of Splicing for Disease**
Adrian Krainer, Ph.D., *Cold Spring Harbor Laboratory*

10:50 am EDT **Early Forays in Individualized Medicine**
Timothy Yu, M.D., Ph.D., *Harvard Medical School & Boston Children's Hospital*

11:40 am EDT **In Situ Gene Therapy**
Beverly Davidson, Ph.D., *University of Pennsylvania, Children's Hospital of Philadelphia*

12:30 pm EDT **Lunch Break (30 min)**

Afternoon Moderator: Greg Cox, *The Jackson Laboratory*

1:00 pm EDT **Gene Therapy Approaches to Hemoglobinopathies**
Stuart Orkin, M.D., *Harvard Medical School*

2:00 pm EDT **Panel Discussion + Q&A**
Adrian Krainer, Timothy Yu, Beverly Davidson, Stuart Orkin & Kay Davies

3:00 PM EDT **Insights into the Pathogenesis and Treatment of Vascular Connective Tissue Disorders**
Hal Dietz, M.D., *Johns Hopkins University*

4:30pm EDT **Course Summary & Wrap Up**