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# **VIRTUAL**

## **Human and Mammalian Genetics and Genomics: The 61<sup>st</sup> McKusick Short Course**

**July 20<sup>th</sup> – 30, 2020**

*Registered Attendees will receive access to our Canvas LMS approximately two weeks in advance of the course. Pre-Recorded content will be available on initial log in to Canvas. Each day of the course, additional pre-recorded lectures will be uploaded with live afternoon sessions for Q&A Sessions with speakers, Keynote Lectures and Workshops. The times of all live sessions are shown in Eastern Standard Time, and will be available on-demand 48 hours after session. Each pre-recorded lecture will be approximately 60 minutes in length.*

***Schedule is Subject to Change***

### **Pre-Recorded Content**

#### **Course Welcome**

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

#### **History of Human Genetics**

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

#### **Mice as Models for Human Diseases**

Greg Cox, Ph.D., *The Jackson Laboratory*

#### **Opportunities and Limits in the 21<sup>st</sup> Century Mouse Model System**

Steve Munger, Ph.D., *The Jackson Laboratory*

#### **Molecular Genetics**

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

#### **Introduction to MGI**

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

#### **OMIM**

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

#### **Skippy Lane Memorial Mouse Genetics Clinic**

Greg Cox, Ph.D., *The Jackson Laboratory*

## **Monday, July 20<sup>th</sup>**

### PRE-RECORDED CONENT

#### **Chromosome Structure and Function**

Jeanne Lawrence, Ph.D., *University of Massachusetts Medical School*

#### **Using Large-Scale Genomic Databases to Understand Human Gene Function**

Anne O'Donnell-Luria, M.D., Ph.D., *Broad Institute of MIT & Harvard*

#### **CNVs in Human Disease**

Charles Lee, Ph.D., *The Jackson Laboratory for Genomic Medicine*

#### **Sequencing Technology and Application**

Elaine Mardis, Ph.D., *Nationwide Children's Hospital*

### LIVE SESSIONS

11:15 am

#### **Q&A Panel Session**

*David Valle, Greg Cox & Steve Munger*

11:45 am

#### **Q&A Panel Session**

*John Phillips, Ada Hamosh & David Shaw*

1:15 pm

#### **Workshop: Solving Molecular Genetics Problems**

*John A. Phillips, III., M.D., Vanderbilt University School of Medicine & Greg Cox, Ph.D., The Jackson Laboratory*

3:15 pm

#### **Thomas H. Roderick Memorial Lecture Title TBD**

*Rudolf Jaenisch, M.D., Whitehead Institute*

6:00 pm

#### **Virtual Happy Hour Meet & Greet**

## **Tuesday, July 21<sup>st</sup>**

### PRE-RECORDED CONENT

#### **Transcriptional Regulation of Gene Expression**

Andrew McCallion, Ph.D., *Johns Hopkins University School of Medicine*

#### **Inborn Errors of the Epigenetic Apparatus: Genetics of Chromatin Readers and Writers**

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

#### **Organizing Principles of the 3D Genome**

Jennifer Phillips-Cremins, Ph.D., *University of Pennsylvania*

#### **Machine Learning for Regulatory Genomics: Basics and Pitfalls**

Michael Beer, Ph.D., *Johns Hopkins University School of Medicine*

### LIVE SESSIONS

11:15 am

#### **Q&A Panel Session**

*Jeanne Lawrence & Charles Lee*

11:45 am

#### **Q&A Panel Session**

*Elaine Mardis & Anne O'Donnell*

1:15 pm

#### **Workshop: Mouse Genomic Resources**

*Carol Bult, Ph.D., The Jackson Laboratory &  
Cynthia Smith, Ph.D., The Jackson Laboratory*

3:15 pm

#### **The Nature of Mutation**

*Stylianos Antonarakis, M.D., DSc, University of Geneva Medical School*

4:45 pm

#### **Poster Presentations**

## **Wednesday, July 22<sup>nd</sup>**

### PRE-RECORDED CONTENT

#### **CRISPR**

Albert Cheng, Ph.D., *The Jackson Laboratory for Genomic Medicine*

#### **Single Cell Analysis in Normal and Disease Tissue**

Loyal Goff, Ph.D., *Johns Hopkins University School of Medicine*

#### **A Primer on Quantitative Systems Biology**

Aravinda Chakravarti, Ph.D., *NYU Langone Health*

#### **Mapping Complex Traits in Mice**

Gary Churchill, Ph.D., *The Jackson Laboratory*

### LIVE SESSIONS

11:15 am

#### **Q&A Panel Session**

*Andy McCallion & Michael Beer*

11:45 am

#### **Q&A Panel Session**

*Jennifer Phillips-Cremins & Hans Bjornsson*

1:15 pm

#### **RNA-seq Analysis Workshop**

Steve Munger, Ph.D., *The Jackson Laboratory*

3:15 pm

#### **Victor A. McKusick Lecture**

#### **Regulation of Oxygen Homeostasis by Hypoxia-Inducible Factors**

Gregg Semenza, M.D., Ph.D., *Johns Hopkins University*

## **Thursday, July 23<sup>rd</sup>**

### PRE-RECORDED CONTENT

#### **Complex Disease Genetics and Drug Discovery**

Jeffrey Barrett, DPhil, *Wellcome Sanger Institute*

#### **Lecture Title TBD**

Jose Florez, M.D., Ph.D., *Massachusetts General Hospital, Harvard Medical School, & The Broad Institute*

#### **Lecture Title TBD**

Eimear Kenny, Ph.D., *Icahn School of Medicine at Mount Sinai*

#### **Genetic Epidemiology of Breast and Ovarian Cancer**

Larry Brody, Ph.D., *National Human Genome Research Institute*

#### **Cancer Biology Exposed by Large-Scale Sequencing**

Victor Velculescu, M.D., Ph.D., *Johns Hopkins University*

### LIVE SESSIONS

11:15 am

#### **Q&A Panel Session**

*Loyal Goff & Albert Cheng*

11:45 am

#### **Q&A Panel Session**

*Aravinda Chakravarti & Gary Churchill*

1:15 pm

#### **Workshop: UCSC Genome Browser in Clinical Medicine**

Robert Kuhn, Ph.D., *University of California, Santa Cruz*

3:15 pm

#### **Lecture Title TBD**

Evan Eichler, Ph.D., *UW Genome Sciences*

## **Friday, July 24<sup>th</sup>**

### PRE-RECORDED CONENT

#### **Genetics of Autoimmunity**

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

#### **Inherited Immunodeficiencies: Models for Newborn Screening and Gene Therapy**

Jennifer Puck, M.D., *University of California San Francisco*

#### **Immunodeficiency**

Steven Holland, M.D., *National Institutes of Health*

#### **The Humanized Mouse in Biomedical Research**

Lenny Shultz, Ph.D., *The Jackson Laboratory*

### LIVE SESSIONS

11:15 am

#### **Q&A Panel Session**

*Jeffrey Barrett, Jose Florez & Eimear Kenny*

11:45 am

#### **Q&A Panel Session**

*Lawrence Brody & Victor Velculescu*

1:15 pm

#### **Workshop: ClinGen & ClinVar**

*Heidi Rehm, Ph.D., Massachusetts General Hospital & Broad Institute*

3:15 pm

#### **Chen K. Chai Lecture**

##### **Lecture Title TBD**

*David Scadden, M.D., Harvard Medical School, Massachusetts General Hospital*

## **Monday, July 27<sup>th</sup>**

### PRE-RECORDED CONTENT

#### **Genomics and the Inference of Human Population History**

Andrew Clark, Ph.D., *Cornell University*

#### **African Evolutionary Genomics**

Sarah Tishkoff, Ph.D., *University of Pennsylvania*

#### **Out of Africa, Into Genomics – An Overview of African Genomic Diversity**

Neil Hanchard, M.D., Ph.D., *Baylor College of Medicine*

#### **Genetic Medicine of the African Population: Promise and Prospects**

Ambroise Wonkam, M.D., D.Med.Sci, Ph.D., *University of Cape Town*

### LIVE SESSIONS

11:15 am

#### **Q&A Panel Session**

*Steven Holland & Jennifer Puck*

11:45 am

#### **Q&A Panel Session**

*David Serreze & Lenny Shultz*

1:15 pm

#### **Workshop: WES/WGS Analysis**

*Nara Sobreira, M.D., Ph.D., Johns Hopkins University*

3:15 pm

#### **Inborn Errors of Immunity to Sars-CoV-2**

*Jean-Laurent Casanova, M.D., Ph.D., The Rockefeller University*

## **Tuesday, July 28<sup>th</sup>**

### PRE-RECORDED CONTENT

#### **The Genetics and Clinical Phenotypes of Inherited Skeletal Dysplasias**

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

#### **Genetic and Pharmacologic Modifiers of Cystic Fibrosis**

Garry Cutting, M.D., *Johns Hopkins University*

#### **Pharmacogenomics: Discovery, Translation and Implementation**

Richard Weinshilboum, M.D., *Mayo Clinic*

#### **Mouse Models for COVID-19**

Nadia Rosenthal, Ph.D., *The Jackson Laboratory*

#### **Biomedical Ethics in Genomics**

Debra Mathews, Ph.D., M.A., *Johns Hopkins University*

### LIVE SESSIONS

11:15 am

#### **Q&A Panel Session**

*Andrew Clark & Neil Hanchard*

11:45 am

#### **Q&A Panel Session**

*Sarah Tishkoff & Ambroise Wonkam*

1:15 pm

#### **Poster Presentations**

3:15 pm

#### **TGF Beta Signaling and the Marfan Syndrome**

Harry "Hal" Dietz, *Johns Hopkins University School of Medicine*



## **Wednesday, July 29<sup>th</sup>**

### PRE-RECORDED CONTENT

#### **Proteomics as a Biological Assay**

Steven Gygi, Ph.D., *Harvard Medical School*

#### **Metabolomics in the Assessment of Human Diseases**

Ralph DeBerardinis, M.D., Ph.D., *University of Texas Southwestern*

#### **Inborn Errors of Metabolism**

Hilary Vernon, M.D., Ph.D., *Johns Hopkins University*

#### **Treatment of Genetic Disease**

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

#### **Prenatal Genetic Diagnosis and Screening**

Ignatia B. Van den Veyver, M.D., *Baylor College of Medicine*

### LIVE SESSIONS

11:15 am

#### **Q&A Panel Session**

*Julie Hoover-Fong, Garry Cutting & Richard Weinshilboum*

11:45 am

#### **Q&A Panel Session**

*Nadia Rosenthal & Debra Mathews*

1:15 pm

#### **Senior Lecturer**

#### **The Genetic Basis of Human Developmental Disorders**

*David FitzPatrick, M.D., The University of Edinburgh Institute of Genetics*

**Thursday, July 30<sup>th</sup>**

PRE-RECORDED CONTENT

**Gene Therapy**

TBA

**Remembrance of Things Past: Reactivation of Fetal Hemoglobin for Therapy**

Stuart Orkin, M.D., *Harvard Medical School*

**Milasen: An Unexpected Case Study in Hyperindividualized Medicine**

Timothy Yu, M.D., Ph.D., *Boston Children's Hospital*

LIVE SESSIONS

11:15 am

**Q&A Panel Session**

*Steven Gygi & Ralph DeBerardinis*

11:45 am

**Q&A Panel Session**

*Hillary Vernon, David Valle & Ignatia Van den Veyver*

1:15 pm

**Q&A Panel Session**

*Stuart Orkin & Timothy Yu*

3:15 pm

**Course Wrap-Up & Poster Awards**

- End -