Monday, July 20th

8:00 – 9:00 am  Registration and Continental Breakfast

9:00 – 9:15 am  Overview of Course Evaluations

9:15 – 9:30 am  Course Welcome
Edison Liu, M.D., The Jackson Laboratory

9:30 – 10:30 am  History of Human Genetics
David Valle, M.D., Johns Hopkins University School of Medicine

10:30 – 11:00 am  Break

11:00 am – 12:00 pm  Mice as Models for Human Diseases
Greg Cox, Ph.D., The Jackson Laboratory

12:00 – 1:00 pm  Chromosome Structure and Function
Jeanne Lawrence, Ph.D., University of Massachusetts Medical School

1:00 – 2:00 pm  Lunch - Roscoe’s Dining Area

2:00 – 3:00 pm  Opportunities and Limits in the 21st Century Mouse Model System
Steve Munger, Ph.D., The Jackson Laboratory

3:00 – 4:00 pm  Molecular Genetics
John A. Phillips, III., M.D., Vanderbilt University School of Medicine
4:00 – 4:15 pm  Break
4:15 – 5:15 pm  **Workshop: Solving Molecular Genetics Problems**  
John A. Phillips, III., M.D., *Vanderbilt University School of Medicine*
5:15 – 6:00 pm  **Welcome Dinner - Roscoe’s Dining Area**

**Evening Lecture**

*Location: The Jackson Laboratory, 600 Main Street*

6:00 – 7:00 pm  **Thomas H. Roderick Lecture Reception – Roscoe’s Dining Area**
7:00 – 7:15 pm  **Introduction and Welcome**
7:15 – 8:15 pm  **Thomas H. Roderick Memorial Lecture**  
*Title TBD*  
Rudolf Jaenisch, M.D., *Whitehead Institute*

**Tuesday, July 21st**

7:45 – 8:30 am  **Continental Breakfast - Roscoe’s Dining Area**
8:30 – 9:25 am  TBD  
TBD
9:25 – 10:15 am  **Transcriptional Regulation of Gene Expression**  
Andrew McCallion, Ph.D., *Johns Hopkins University School of Medicine*
10:15 – 10:45 am  **Break - (GROUP PHOTO)**
10:45 – 11:40 am  **Single Cell Analysis in Normal and Disease Tissue**  
Loyal Goff, Ph.D., *Johns Hopkins University School of Medicine*
11:40 am – 12:30 pm  **Three-Dimensional Folding of Mammalian Genomes**  
Jennifer Phillips-Cremins, Ph.D., *University of Pennsylvania*
12:30 – 1:30 pm  **Lunch - Roscoe’s Dining Area**
1:30 – 2:30 pm  **Introduction to MGI/OMIM**  
David Shaw, M.S., *The Jackson Laboratory*  
Ada Hamosh, M.D., *The Johns Hopkins University School of Medicine*
2:30 – 4:30 pm  **RNA-seq Analysis Workshop**  
Steve Munger, Ph.D., *The Jackson Laboratory*
Evening Lecture

Location: The Bar Harbor Regency, 123 Eden Street, Bar Harbor, Maine

7:30 – 8:30 pm The Nature of Mutation
Stylianos Antonarakis, M.D., DSc, University of Geneva Medical School

Wednesday, July 22nd

7:45 – 8:30 am Continental Breakfast - Roscoe’s Dining Area

8:30 – 9:25 am Sequencing Technology and Application
Elaine Mardis, Ph.D., Nationwide Children’s Hospital

9:25 – 10:15 am CNVs in Human Disease
Charles Lee, Ph.D., The Jackson Laboratory for Genomic Medicine

10:15 – 10:45 am Break

10:45 – 11:40 am TBD

11:40 am – 12:30 pm Using Large-Scale Genomic Databases to Understand Human Gene Function
Anne O'Donnell-Luria, M.D., Ph.D., Broad Institute of MIT & Harvard

12:30 – 1:30 pm Lunch - Roscoe’s Dining Area

1:30 – 3:30 pm Workshop: UCSC Genome Browser in Clinical Medicine
Robert Kuhn, Ph.D., University of California, Santa Cruz

Poster Session

Location: The Jackson Laboratory, 600 Main Street, Bar Harbor, ME 04609

5:00 – 7:00 pm Poster Session

Evening Lecture

Location: The Jackson Laboratory, 600 Main Street, Bar Harbor, ME 04609

7:30 – 8:30 pm Victor A. McKusick Lecture
Regulation of Oxygen Homeostasis by Hypoxia-Inducible Factor I
Gregg Semenza, M.D., Ph.D., Johns Hopkins University

Thursday, July 23rd

7:45 – 8:30 am  
Continental Breakfast - Roscoe’s Dining Area

8:30 – 9:25 am  
A Primer on Quantitative Systems Biology
Aravinda Chakravarti, Ph.D., NYU Langone Health

9:25 – 10:15 am  
TBD
TBD

10:15 – 10:45 am  
Break

10:45 – 11:40 am  
Mapping Complex Traits in Mice
Gary Churchill, Ph.D., The Jackson Laboratory

11:40 am – 12:30 pm  
TBD
Jose Florez, M.D., Ph.D., Massachusetts General Hospital, Harvard Medical School, & The Broad Institute

12:30 – 1:30 pm  
Lunch - Roscoe’s Dining Area

Evening Lecture

Location: The Bar Harbor Regency, 123 Eden Street, Bar Harbor, Maine

7:30 – 8:30 pm  
TBD
Evan Eichler, Ph.D., University of Washington

Friday, July 24th

7:45 – 8:30 am  
Continental Breakfast - Roscoe’s Dining Area

8:30 – 9:25 am  
Genetic Epidemiology of Breast and Ovarian Cancer
Larry Brody, Ph.D., National Human Genome Research Institute
9:25 – 10:15 am  Cancer Biology Exposed by Large-Scale Sequencing  
Victor Velculescu, M.D., Ph.D., Johns Hopkins University

10:15 – 10:45 am  Break

10:45 – 11:40 am  Inborn Errors of the Epigenetic Apparatus: Genetics of Chromatin Readers and Writers  
Hans Bjornsson, M.D., Ph.D., Johns Hopkins University School of Medicine

11:40 am – 12:30 pm  TBD  
TBD

12:30 – 1:30 pm  Lunch - Roscoe’s Dining Area

1:30 – 3:30 pm  Workshop: ClinGen & ClinVar  
Heidi Rehm, Ph.D., Massachusetts General Hospital & Broad Institute

Evening Lecture

Location: The Bar Harbor Regency, 123 Eden Street, Bar Harbor, Maine

7:00 pm  Introduction and Welcome

7:30 – 8:30 pm  Chen K. Chai Lecture  
TBD

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

Saturday, July 25th

7:45 – 8:30 am  Continental Breakfast - Roscoe’s Dining Area

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

9:25 – 10:15 am  African Evolutionary Genomics  
Sarah Tishkoff, Ph.D., University of Pennsylvania

10:15 – 10:45 am  Break

10:45 – 11:40 am  Genetic Medicine of the African Population: Promise and Prospects  
Ambroise Wonkam, M.D., D.Med.Sci, Ph.D., University of Cape Town

11:40 am – 12:30 pm  TBD  
Eimear Kenny, Ph.D., Icahn School of Medicine at Mount Sinai
12:30 pm  Lunch – Boxed lunch provided to-go

**Monday, July 27th**

7:45 – 8:30 am  **Continental Breakfast - Roscoe’s Dining Area**

8:30 – 9:25 am  **Genetics of Autoimmunity**
David Serreze, Ph.D., *The Jackson Laboratory*

9:25 – 10:15 am  TBD

10:15 – 10:45 am  Break

10:45 – 11:45 am  **Immunodeficiency**
Steven Holland, M.D., *National Institutes of Health*

11:40 am – 12:30 pm  **Inborn Errors of Metabolism**
Hilary Vernon, M.D., Ph.D., *Johns Hopkins University*

12:30 – 1:30 pm  **Lunch - Roscoe’s Dining Area**

1:30 – 3:30 pm  **Workshop: WES/WGS Analysis**
Nara Sobreira, M.D., Ph.D., *Johns Hopkins University*

**Evening Lecture**

*Location: The Bar Harbor Regency, 123 Eden Street, Bar Harbor, Maine*

7:30 – 8:30 pm  **TGF Beta Signaling and the Marfan Syndrome**
Harry “Hal” Dietz, *Johns Hopkins University School of Medicine*

**Tuesday, July 28th**

7:45 – 8:30 am  **Continental Breakfast - Roscoe’s Dining Area**

8:30 – 9:25 am  TBD

9:25 – 10:15 am  **Genomics and the Inference of Human Population History**
Andrew Clark, Ph.D., *Cornell University*

10:15 – 10:45 am  Break
10:45 – 11:40 am  **New Strategies for Accelerating Regeneration**  
Nadia Rosenthal, Ph.D., *The Jackson Laboratory*

11:40 am – 12:30 pm  **Prenatal Diagnosis and Screening**  
Ignatia B. Van den Veyver, M.D., *Baylor College of Medicine*

12:30 – 1:30 pm  **Lunch - Roscoe’s Dining Area**

1:30 – 3:30 pm  **Workshop: Mouse Genomic Resources**  
Carol Bult, Ph.D., *The Jackson Laboratory*  
Cynthia Smith, Ph.D., *The Jackson Laboratory*

**Evening Lecture**

*Location: The Bar Harbor Regency, 123 Eden Street, Bar Harbor, Maine*

7:30 – 8:30 pm  TBD  
TBD

**Wednesday, July 29th**

7:45 – 8:30 am  **Continental Breakfast - Roscoe’s Dining Area**

8:30 – 9:25 am  **The Humanized Mouse in Biomedical Research**  
Lenny Shultz, Ph.D., *The Jackson Laboratory*

9:25 – 10:15 am  **The Genetics and Clinical Phenotypes of Inherited Skeletal Dysplasias**  
Julie Hoover-Fong, M.D., Ph.D., *Johns Hopkins University School of Medicine*

10:15 – 10:45 am  **Break**

10:45 – 11:40 am  **Genetic and Pharmacologic Modifiers of Cystic Fibrosis**  
Garry Cutting, M.D., Johns Hopkins University

11:40 am – 12:30 pm  **Pharmacogenomics: Discovery, Translation and Implementation**  
Richard Weinshilboum, M.D., *Mayo Clinic*

12:30 – 1:30 pm  **Lunch - Roscoe’s Dining Area**

1:00 – 2:00 pm  **Human Genetics Clinic Presentations**  
*(Selected Participants and Invited Guests Only)*  
Ada Hamosh, M.D., M.P.H., *Johns Hopkins University*

2:00 – 4:00 pm  **Human Genetics Clinic Presentations**  
*(Open to All Attendees)*
Evening Lecture

Location: The Bar Harbor Regency, 123 Eden Street, Bar Harbor, Maine

5:30 pm Dinner
7:00 pm Introduction and Welcome
7:30 – 8:30 pm Senior Lecturer
Title TBD
David FitzPatrick, M.D., The University of Edinburgh Institute of Genetics

Thursday, July 30th

7:45 – 8:30 am Continental Breakfast - Roscoe’s Dining Area
8:30 – 9:25 am Proteomics as a Biological Assay
Steven Gygi, Ph.D., Harvard Medical School
9:25 – 10:25 am Metabolomics in the Assessment of Human Diseases
Ralph DeBerardinis, M.D., Ph.D., University of Texas Southwestern
10:25 – 10:45 am Break
10:45 – 11:40 am Biomedical Ethics in Genomics
Debra Mathews, Ph.D., M.A., Johns Hopkins University
11:40 am – 12:30 pm TBD
TBD
12:30 – 1:30 pm Lunch - Roscoe’s Dining Area
1:30 – 3:30 pm Skippy Lane Memorial Mouse Genetics Clinic
Greg Cox, Ph.D., The Jackson Laboratory
5:00 – 7:00 pm Harbor Cruise - Advance sign up required

Evening Lecture

Location: The Bar Harbor Regency, 123 Eden Street, Bar Harbor, Maine

7:30 – 9:00 pm ELSI Case Studies
Clair Francomano, M.D., Indiana University
Friday, July 31st

7:45 – 8:30 am  **Continental Breakfast - Roscoe’s Dining Area**

8:30 – 9:25 am  **Treatment of Genetic Disease**
David Valle, M.D., *Johns Hopkins University School of Medicine*

9:25 – 10:15 am  **Title TBD**
Stuart Orkin, M.D., Harvard Medical School

10:15 – 10:45 am  **Break**

10:45 – 11:40 am  **TBD**
TBD

11:40 am – 12:30 pm  **Course Summary & Wrap Up**
David FitzPatrick, M.D., *The University of Edinburgh Institute of Genetics*

12:30 – 1:30 pm  **Lunch – Boxed lunch provided to-go**

- End -