



56th Annual Short Course on Medical and Experimental Mammalian Genetics

July 19 – July 31, 2015

EVENT SCHEDULE

For more information, please contact Erin McDevitt; erin.mcdevitt@jax.org

Sunday, July 15

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

4:00-6:00pm **Registration**

Monday, July 20

Location: The Jackson Laboratory, 600 Main Street

7:30- 8:00am **Late Registration and Continental Breakfast**

8:00-8:20am **Overview of Course Evaluations**

8:20-8:40am **Course Welcome**
Edison Liu, M.D., The Jackson Laboratory

8:40 - 9:25am **History of Human Genetics**
David Valle, M.D., Johns Hopkins University School of Medicine

9:25 – 10:15 am **The Evolution of Mouse Genetics: Past, Present and Future**
Kenneth Paigen, Ph.D., The Jackson Laboratory

10:15 -10:45am **Break** - *Roscoe's Dining Area*

10:45-11:40am **Chromosome Structure and Function**
Jeanne Lawrence, Ph.D., University of Massachusetts Medical School

- 11:40-12:30pm **TBD**
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*
- 1:30 – 3:00pm **Mendelian Inheritance in Humans**
Clair Francomano, M.D., *Harvey Institute for Human Genetics*
- 3:00-4:00pm **Mice as Models for Human Diseases**
Greg Cox, Ph.D., *The Jackson Laboratory*

Evening Lecture

Location: The Jackson Laboratory, 600 Main Street

- 6:00-7:00pm **Thomas H. Roderick Lecture Reception** – *Roscoe's Dining Area*
- 7:00-7:20pm **Introduction and Welcome**
- 7:20-8:15pm **Thomas H. Roderick Memorial Lecture**
Title TBD
Terry Magnuson, Ph.D., *UNC School of Medicine*

Tuesday, July 21

Location: The Jackson Laboratory, 600 Main Street

- 7:45 - 8:25am **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25am **Massively Parallel Sequencing: Technologies and Applications**
Elaine Mardis, Ph.D., *The Genome Institute Washington University*
- 9:25-10:15am **Transcriptional Regulation of Gene Expression**
Andrew S. McCallion, Ph.D., *Johns Hopkins University School of Medicine*
- 10:15-10:45am **Break** - *Roscoe's Dining Area*
(GROUP PHOTO)
- 10:45-11:40am **Post-transcriptional Control of Gene Expression**
Harry C. Dietz, M.D., *The Johns Hopkins University School of Medicine*
- 11:40-12:30pm **MicroRNAs in Normal Physiology and Disease**
Joshua T. Mendell, M.D., Ph.D., *UT Southwestern Medical Center*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*
- 1:30 -2:30pm **Molecular Genetics**
John A. Phillips, III, M.D., *Vanderbilt University School of Medicine*

2:30 – 3:30pm **Introduction to MGI/OMIM**
Joanne Berghout, *The Jackson Laboratory* and
Ada Hamosh, M.D., *The Johns Hopkins University School of Medicine*

Evening Lecture

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

7:30-9:00pm **The Nature of Mutation**
Stylianos Antonarakis, M.D., DSc, *University of Geneva Medical School*

Wednesday, July 22

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:25am **Genetics of Chromatin Readers and Writers**
Hans Bjornsson, M.D., Ph.D., *Johns Hopkins University School of Medicine*

9:25-10:15am **Epigenetics**
Andrew Feinberg, M.D., MPH, *Johns Hopkins University*

10:15 -10:45am **Break - Roscoe's Dining Area**

10:45-11:40am **Transcriptional Responses to Hypoxia and Ischemia**
Gregg Semenza, M.D., Ph.D., *Johns Hopkins University School of Medicine*

11:40-12:30pm **Genetics of Obesity and Type II Diabetes**
Jürgen Naggert, Ph.D., *The Jackson Laboratory*

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

1:30 – 2:30pm **RNA Sequencing Workshop**
Steve Munger, Ph.D., *The Jackson Laboratory*

2:30-3:30pm **Exome Sequencing Workshop**
Nara Sobreira, M.D., Ph.D., *Johns Hopkins University School of Medicine*

Poster Session

Location: Bar Harbor Regency Hotel – Ballroom, 123 Eden Street

5:00-7:00pm **Poster Session**

Evening Lecture

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

7:30-9:00pm **TGF Beta Signaling and Marfan Syndrome**
Hal Dietz, M.D., Johns Hopkins University

Thursday, July 23

Location: The Jackson Laboratory, 600 Main Street

7:45 - 8:25am **Continental Breakfast** - *Roscoe's Dining Area*

8:30-9:40am **Population Genomics**
Aravinda Chakravarti, Ph.D., *Johns Hopkins University School of Medicine*

9:40-10:15am **Methods for Linkage & Association Studies**
Goncalo Abecasis, Ph.D., *The University of Michigan*

10:15 -10:45am **Break** - *Roscoe's Dining Area*

10:45-11:40am **Methods for Linkage & Association Studies continued**
Goncalo Abecasis, Ph.D., *The University of Michigan*

11:40-12:30pm **Complex Traits in Mice**
Gary Churchill, Ph.D., *The Jackson Laboratory*

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

1:30-3:30pm **UCSC Genome Browser**

Evening Lecture

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

7:30-9:00pm **TBD**

Friday, July 24

Location: The Jackson Laboratory, 600 Main Street

7:45 - 8:25am **Continental Breakfast** - *Roscoe's Dining Area*

8:30-9:25am **Genetic Epidemiology of Breast and Ovarian Cancer**
Lawrence C. Brody, Ph.D., *National Human Genome Research Institute*

9:25-10:15am **Cancer biology exposed by large-scale sequencing**
Victor Velculescu, M.D., Ph.D., *Johns Hopkins University School of Medicine*

10:15 -10:45am **Break** - *Roscoe's Dining Area*

10:45-11:40am **Genetics of Schizophrenia**
Dimitrios Avramopoulos, M.D., Ph.D., *Johns Hopkins University*

11:40 – 12:30pm **Proteomics**
Steven Gygi, Ph.D., *Harvard Medical School*

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

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

Location: Bar Harbor Regency Hotel – Ballroom, 123 Eden Street

5:00-7:00pm **Lobster Dinner**

Evening Lecture

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

7:30-9:00pm **Chen K. Chai Lecture**
Fishing for the secrets of evolutionary change in sticklebacks and humans
David Kingsley, Ph.D., *Stanford University*

Saturday, July 25

Location: The Jackson Laboratory, 600 Main Street

7:45 - 8:25am **Continental Breakfast** - *Roscoe's Dining Area*

8:30-9:25am **Human Population History and Genetics**
Andrew Clark, Ph.D., *Cornell University*

9:25-10:15am **African Genomics: Implications for Studies of Human Evolution and Disease**
Sarah Tishkoff, Ph.D., *University of Pennsylvania*

10:15 -10:45am **Break** - *Roscoe's Dining Area*

10:45-11:40am **CRISPR/Cas9-mediated Genome Editing**
Haoyi Wang, Ph.D., *The Jackson Laboratory*

11:40 – 12:30pm **iPS Cell Technology, Gene Editing and Disease Research**
Rudolf Jaenisch, M.D., *Whitehead Institute*

12:30 Boxed Lunches

Sunday, July 26

No Scheduled Course Events

Monday, July 27

Location: The Jackson Laboratory, 600 Main Street

- 7:45 - 8:20am **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25am **The Major Histocompatibility Complex in Health and Disease**
Derry Roopenian, Ph.D., *The Jackson Laboratory*
- 9:25 -10:15 am **Genetics of Autoimmunity**
David Serreze, Ph.D., *The Jackson Laboratory*
- 10:15 -10:45am **Break** - *Roscoe's Dining Area*
- 10:45-11:40am **Immune Dysfunction from Common and Rare Variants**
Judy Cho, M.D., *Mount Sinai Hospital*
- 11:40 – 12:30pm **Genetics of Infectious Diseases**
Steven M. Holland, M.D., *National Institutes of Health*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*
- 1:30 – 2:30pm **Inborn Errors**
David Valle, M.D., *Johns Hopkins University School of Medicine*
- 2:30-3:30pm **Workshop: Methods of Molecular Genetics**
John A. Phillips, III, M.D., *Vanderbilt University School of Medicine* and
Juergen Naggert, Ph.D., *The Jackson Laboratory*

Evening Lecture

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

- 7:30-9:00pm **Lessons Learned from Genomics and Gene Regulation**
Eddy Rubin, M.D., Ph.D., *Joint Genome Institute, Lawrence Berkeley National Laboratory*

Tuesday, July 28

Location: The Jackson Laboratory, 600 Main Street

- 7:45 - 8:25am **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25am **KOMP**
Bob Braun, Ph.D., *The Jackson Laboratory*

- 9:25 -10:15 am **Modeling Behavior in the Laboratory Mouse**
Elissa Chesler, Ph.D., *The Jackson Laboratory*
- 10:15 -10:45am **Break** - *Roscoe's Dining Area*
- 10:45-11:40am **CNVs in Human Disease**
Charles Lee, Ph.D., *The Jackson Laboratory for Genomic Medicine*
- 11:40-12:30 **Chromosome Instability and Cancer**
Kevin Mills, Ph.D., *The Jackson Laboratory*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*
- 1:30 – 3:30pm **Human Genomes and Genomic Resources Workshop**
Deanna Church, Ph.D., *Personalis*

Evening Lecture

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

- 7:30-9:00pm **Reawakening Fetal Hemoglobin for Therapy: An Old Problem with New Prospects**
Stuart Orkin, M.D., *Harvard Stem Cell Institute*

Wednesday, July 29

Location: The Jackson Laboratory, 600 Main Street

- 7:45 - 8:25am **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25am **Genetic Modifiers of CF**
Garry R. Cutting, M.D., *Johns Hopkins University School of Medicine*
- 9:25-10:15am **Molecular Bases of Malformation Syndromes**
David FitzPatrick, M.D., FRCD(Edin), *Medical Research Council Human Genetics Unit*
- 10:15 -10:45am **Break** - *Roscoe's Dining Area*
- 10:45-11:40am **Genetic Disorders of Dietary Excess: Getting to the Heart of the Matter**
Helen Hobbs, M.D., HHMI, *UT Southwestern Medical Center*
- 11:40-12:30pm **Pharmacogenomics: Development, Science and Translation**
Richard M. Weinshilboum, M.D., *Mayo Clinic*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

12:30-2:00pm **Lunch** - *La Bella Vita Ristorante*
(Selected Participants and Invited Guests Only)

Human Genetics Clinic Presentations
(Selected Participants and Invited Guests Only)
Ada Hamosh, M.D., M.P.H., *Johns Hopkins University School of Medicine*

2:00-4:00pm **Human Genetics Clinic Presentations**
(Open to All Attendees)
Ada Hamosh, M.D., M.P.H., *Johns Hopkins University School of Medicine*

Evening Lecture

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

7:30-9:00pm **Senior Lecturer**
Mouse Models and Stem Cell Models of Human Disease
Janet Rossant, PhD, FRS, FRSC, *SickKids*

Thursday, July 30

Location: The Jackson Laboratory, 600 Main Street

7:45 - 8:25am **Continental Breakfast** - *Roscoe's Dining Area*

8:30-9:25 am **Humanized Mice in Research**
Michael Brehm, Ph.D., *University of Massachusetts Medical School*

9:25 – 10:25am **Metabolomics and Metabolic Flux Analysis**
Ralph DeBeradinis, M.D., Ph.D., *UT Southwestern*

10:25-10:45am **Break** - *Roscoe's Dining Area*

10:45am-11:40pm **Biomedical Ethics in Genomics**
Debra Mathews, Ph.D., M.A., *Johns Hopkins University School of Medicine*

11:40-12:30pm **DNA Forensics**
Phil Reilly, M.D., J.D., *Third Rock Ventures*

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

1:30-3:30pm **Mouse Genetics Clinic**
Greg Cox, Ph.D., *The Jackson Laboratory*

Evening Lecture

Location: Bar Harbor Regency Hotel – Stone House, 123 Eden Street

7:30-9:00pm **ELSI Case Studies**

Friday, July 31

Location: The Jackson Laboratory, 600 Main Street

7:45 - 8:25am **Continental Breakfast** - *Roscoe's Dining Area*

8:30-9:25am **Prenatal Diagnosis and Screening**
Laird G. Jackson, M.D., *Drexel University College of Medicine*

9:25-10:15am **Treatment of Genetic Disease**
David Valle, M.D., *Johns Hopkins University School of Medicine*

10:25-10:45am **Break** - *Roscoe's Dining Area*

10:45-11:40am **Gene Therapy for Genetic Disease**
Katherine High, M.D., *Spark Therapeutics*

11:40-12:30pm **Course Wrap Up**
Janet Rossant, PhD, FRS, FRSC, *SickKids*

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

End of the course