



54th Annual Short Course on Medical and Experimental Mammalian Genetics

July 21 – August 2, 2013

EVENT SCHEDULE

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

Sunday, July 21

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

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

Monday, July 22

Location: The Jackson Laboratory, 600 Main Street

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

8:00-8:20am **Overview of Course Evaluations**
Donna Doherty, M.S., *University of Maine*

8:20-8:30am **Welcome**
Edison Liu, M.D., *The Jackson Laboratory*

8:30 - 9:25am **History of Human Genetics**
David Valle, M.D., *Johns Hopkins University*

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 **Chromosomes in Gametogenesis and in Cell Division**
Christa Martin, Ph.D., *Emory University*
- 11:40-12:30pm **Chromosome Structure and Function**
Jeanne Lawrence, Ph.D., *University of Massachusetts Medical School*
- 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 Bioinformatics**
Aravinda Chakravarti, Ph.D., *The Johns Hopkins University School of Medicine* and Ada Hamosh, M.D., *Johns Hopkins University School of Medicine*

Evening Lecture

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

- 7:30-9:00pm **Nature and Consequences of Mutations**
Stylianios E. Antonarakis, M.D., Ph.D., *University of Geneva Medical School*

Tuesday, July 23

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 **Mendelian Inheritance in Humans**
Clair Francomano, M.D., *Harvey Institute for Human Genetics*
- 2:30 – 3:30pm **Mice as Models for Human Diseases**
Greg Cox, Ph.D., *The Jackson Laboratory*

Evening Lecture

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

- 7:30-9:00pm **Embracing Heterogeneity, Noise and Healthcare Data for Biomedical Discovery**
Isaac Kohane, Ph.D., M.D., *Harvard University*

Wednesday, July 24

Location: The Jackson Laboratory, 600 Main Street

- 7:45 - 8:25am **Continental Breakfast** - *Roscoe's Dining Area*
- 8:30-9:25am **Genetics of Obesity and Type II Diabetes**
Jürgen Naggert, Ph.D., *The Jackson Laboratory*
- 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 **Genetics of Chromatin Readers and Writers**
Hans Bjornsson, M.D., Ph.D., *Johns Hopkins University School of Medicine*
- 11:40-12:30pm **Transcriptional Responses to Hypoxia and Ischemia**
Gregg Semenza, M.D., Ph.D., *Johns Hopkins University School of Medicine*

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

1:30 – 3:30pm **Bioinformatics**
Deanna Church, Ph.D., *National Center for Biotechnology Information*

Poster Session

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

5:30-7:30pm **Poster Session**

Evening Lecture

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

7:30-9:00pm **Found in Translation: New Insights into the Pathogenesis and Treatment of Marfan Syndrome and Related Disorders**
Harry C. Dietz, B.S.E., M.D., *Johns Hopkins University School of Medicine*

Thursday, July 25

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:25am **The 1000 Genomes Project**
Goncalo Abecasis, Ph.D., *The University of Michigan*

9:25-10:15am **Population Genetics & Genomics**
Aravinda Chakravarti, Ph.D., *The Johns Hopkins University School of Medicine*

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

10:45-11:40am **Methods for Linkage & Association Studies**
Goncalo Abecasis, Ph.D., *The University of Michigan* and Aravinda Chakravarti, Ph.D., *The Johns Hopkins University School of Medicine*

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 **Bioinformatics**
Deanna Church, Ph.D., *National Center for Biotechnology Information*

Evening Lecture

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

7:30-9:00pm **Pharmacogenomics: Development, Science and Translation**
Richard M. Weinshilboum, M.D., *Mayo Clinic*

Friday, July 26

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 **Genetics of Infectious Diseases**
Steven M. Holland, M.D., *National Institutes of Health*

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

10:45-11:40am **African Genomics: Implications for Studies of Human Evolution and Disease**
Sarah Tishkoff, Ph.D., *University of Pennsylvania*

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 **Bioinformatics**
Deanna Church, Ph.D., *National Center for Biotechnology Information*

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

5:30-7:30pm **Lobster Dinner**

Evening Lecture

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

7:30-9:00pm **Chen K. Chai Lecture**
RNA Biology and Cancer
Phillip Sharp, Ph.D., *Massachusetts Institute of Technology*

Saturday, July 27

Location: The Jackson Laboratory, 600 Main Street

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

8:30-9:25am **Regulation of Genes**
Oliver Rando, Ph.D., M.D., *University of Massachusetts Medical School*

9:25-10:15am **Nature of Selection**
Molly Przeworski, Ph.D., *University of Chicago*

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

10:45-11:40am **Copy Number Variation and Pediatric Disease**
Evan Eichler, Ph.D., *University of Washington*

11:40 – 12:30pm **Pecking at the Origin of Natural Variation: Insights from Darwin's Finches and Other Birds**
Arhat Abzhanov, Ph.D., *Harvard University*

12:30 Boxed Lunches

Sunday, July 28

No Scheduled Course Events

Monday, July 29

Location: The Jackson Laboratory, 600 Main Street

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| 7:45 - 8:20am | Continental Breakfast - <i>Roscoe's Dining Area</i> |
| 8:20-8:30am | March of Dimes Birth Defects Foundation
Joe Leigh Simpson, M.D., <i>Senior Vice President for Research & Global Programs, March of Dimes</i> |
| 8:30-9:25am | The Major Histocompatibility Complex in Health and Disease
Derry Roopenian, Ph.D., <i>The Jackson Laboratory</i> |
| 9:25 -10:15 am | Genetics of Autoimmunity
David Serreze, Ph.D., <i>The Jackson Laboratory</i> |
| 10:15 -10:45am | Break - <i>Roscoe's Dining Area</i> |
| 10:45-11:40am | Genetic Disorders of the Immune System in Humans
Jennifer Puck, M.D., <i>University of California San Francisco</i> |
| 11:40 – 12:30pm | Genetics of Schizophrenia
Dimitrios Avramopoulos, M.D., Ph.D., <i>Johns Hopkins University</i> |
| 12:30-1:30pm | Lunch - <i>Roscoe's Dining Area</i> |
| 1:30 – 2:30pm | Inborn Errors of Metabolism
David Valle, M.D., <i>Johns Hopkins University School of Medicine</i> |
| 2:30-3:30pm | Workshop: Methods of Molecular Genetics
John A. Phillips, III, M.D., <i>Vanderbilt University School of Medicine</i>
and Jürgen Naggert, Ph.D., <i>The Jackson Laboratory</i> |

Evening Lecture

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

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| 7:30-9:00pm | Lessons Learned from Genomics and Gene Regulation
Eddy Rubin, M.D., Ph.D., <i>Joint Genome Institute, Lawrence Berkeley National Laboratory</i> |
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Tuesday, July 30

Location: The Jackson Laboratory, 600 Main Street

Updated July 16, 2013

7:45 - 8:25am	Continental Breakfast - <i>Roscoe's Dining Area</i>
8:30-9:25am	KOMP Bob Braun, Ph.D., <i>The Jackson Laboratory</i>
9:25 -10:15 am	Pulmonary Hypertension John A. Phillips, III, M.D., <i>Vanderbilt University School of Medicine</i>
10:15 -10:45am	Break - <i>Roscoe's Dining Area</i>
10:45-11:40am	DNA Forensics Phillip Reilly, M.D., J.D., <i>Venture Partner at Third Rock Ventures</i>
11:40-12:30	Cancer biology exposed by large-scale sequencing Victor Velculescu, M.D., Ph.D., <i>The Johns Hopkins University School of Medicine</i>
12:30-1:30pm	Lunch - <i>Roscoe's Dining Area</i>
1:30 – 3:30pm	Genetic Analysis of Complex Traits I Aravinda Chakravarti, Ph.D., <i>The Johns Hopkins University School of Medicine</i> and Goncalo Abecasis, Ph.D., <i>The University of Michigan</i>

Evening Lecture

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

7:30-9:00pm	Lecture Title TBD Barbara Wold, Ph.D., <i>California Institute of Technology</i>
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Wednesday, July 31

Location: The Jackson Laboratory, 600 Main Street

7:45 - 8:25am	Continental Breakfast - <i>Roscoe's Dining Area</i>
8:30-9:25am	Genetic Modifiers of CF Garry R. Cutting, M.D., <i>Johns Hopkins University School of Medicine</i>

- 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 **Chromosome Instability and Cancer**
Kevin Mills, Ph.D., *The Jackson Laboratory*
- 11:40-12:30pm **Genomic Analysis of Cancer**
Edison Liu, M.D., *The Jackson Laboratory*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*

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

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

12:30-2:00pm **Lunch** – *Eden Restaurant*

- 2:00-4:00pm **Human Genetics Clinic Presentations**
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 – Evolution and Cancer**
David Botstein, Ph.D., *Princeton University*

Thursday, August 1st

Location: The Jackson Laboratory, 600 Main Street

- 7:45 - 8:25am **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 – 10:15am **Metabolomics and Metabolic Flux Analysis**
Ralph DeBeradinis, M.D., Ph.D., *UT Southwestern*

- 10:15-10:25 a.m. **Description of the Mouse Clinic**
Leah Rae Donahue, Ph.D., *The Jackson Laboratory*
- 10:25-10:45am **Break** - *Roscoe's Dining Area*
- 10:45am-11:40pm **Individualized Medicine**
Bob Nussbaum, M.D., *University of California, San Francisco*
- 11:40-12:30pm **Biomedical Ethics in Genomics**
Deborah Matthews, Ph.D., M.A., *Johns Hopkins University School of Medicine*
- 12:30-1:30pm **Lunch** - *Roscoe's Dining Area*
- 2:00-3:30pm **Mouse Genetics Clinic**
Leah Rae Donahue, Ph.D., *The Jackson Laboratory*
The Jackson Laboratory – Tent in Visitor Parking Lot
- Lab Tours**
(Departing from Under the Tent every 15 minutes)

Evening Lecture

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

- 7:30-9:00pm **ELSI Case Studies**
Ada Hamosh, M.D.
Clair A. Francomano, M.D.
John Phillips, III, M.D.
Deborah Matthews, Ph.D., M.A.,

Friday, August 2

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 **Reawakening Fetal Hemoglobin for Therapy: An Old Problem with New Prospects**
Stuart Orkin, M.D., *Harvard Stem Cell Institute*
- 10:25-10:45am **Break** - *Roscoe's Dining Area*

10:45-11:40am	Gene Therapy for Genetic Disease Katherine High, M.D., <i>The Children's Hospital of Philadelphia</i>
11:40-12:30pm	Course Wrap Up – Senior Lecturer David Botstein, Ph.D., <i>Princeton University</i>
12:30-1:30pm	Lunch - <i>Roscoe's Dining Area</i>
End of the course	