

# 50<sup>th</sup> Annual Short Course

on Medical and Experimental Mammalian Genetics

July 19-July 31, 2009

For more information, please contact Erin McDevitt; [erin.mcdevitt@jax.org](mailto:erin.mcdevitt@jax.org)

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## **Sunday, July 19**

4:00-7:00pm      **Registration**  
*Bar Harbor Club - Ballroom*

## **Monday, July 20**

7:30- 8:10am      **Late Registration and Continental Breakfast**  
*The Jackson Laboratory – Main Lobby/Roscoe's*

8:10-8:20am      **Overview of Course Evaluations**  
Suzanne Hart, *University of Maine*  
*The Jackson Laboratory - Auditorium*

8:20-8:30am      **Welcome**  
Rick Woychik, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory - Auditorium*

8:30-8:45am      **Course History**  
Tom Roderick, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory - Auditorium*

8:45 - 9:30am      **History of Medical Genetics**  
David Valle, M.D., *Johns Hopkins University*  
*The Jackson Laboratory - Auditorium*

- 9:30-10:15am      **The Evolution of Mouse Genetics: Past, Present and Future**  
Ken Paigen, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory - Auditorium*
- 10:15 -10:45am      **Break**  
*The Jackson Laboratory – Roscoe’s*
- 10:45-11:40am      **Mendelian Inheritance in Humans**  
Clair Francomano, M.D., *Harvey Institute for Human Genetics*  
*The Jackson Laboratory - Auditorium*
- 11:40-12:30pm      **Mice as Models for Human Diseases**  
Patsy Nishina, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*
- 12:30-2:00pm      **Lunch**  
*The Jackson Laboratory – Roscoe’s*
- 2:00-4:00pm      **Mini Symposia: *Databases for mouse and human genetics***  
*The Jackson Laboratory – Auditorium*
- Introduction/Overview of Bioinformatics**  
Carol Bult, Ph.D., *The Jackson Laboratory*
- On-line Mendelian Inheritance in Man (OMIM)**  
Ada Hamosh, M.D., *Johns Hopkins University*
- Mouse Genome Informatics Database: Facilitating mouse as a Model for human disease**  
Paul Szauter, Ph.D., *The Jackson Laboratory*
- 7:30-9:00pm      **Regulatory Genomics and Epigenomics in Fly and Human**  
Manolis Kellis, Ph.D., *Broad Institute of MIT and Harvard*  
*Bar Harbor Club - Ballroom*

**Tuesday, July 21**

- 7:45 - 8:25am      **Continental Breakfast**  
*The Jackson Laboratory – Roscoe’s*
- 8:30-9:25am      **Principles & Techniques of Molecular Genetics**  
John A. Phillips, III, M.D., *Vanderbilt University School of Medicine*  
*The Jackson Laboratory - Auditorium*

- 9:25-10:15am      **Chromosome Structure and Function**  
Lindsay Shopland, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*
- 10:15-10:45am      **Break**  
*The Jackson Laboratory - Roscoe's*
- 10:45-11:40am      **Chromosomes in Gametogenesis and in Cell Division**  
David H. Ledbetter, Ph.D., *Emory University School of Medicine*  
*The Jackson Laboratory – Auditorium*
- 11:40-12:30pm      **Chromosome Aberrations: Molecular Cytogenetics**  
David H. Ledbetter, Ph.D., *Emory University School of Medicine*  
*The Jackson Laboratory – Auditorium*
- 12:30-2:00pm      **Lunch**  
*The Jackson Laboratory – Roscoe's*
- 2:00-4:00pm      **Workshop: Cytogenetics**  
David H. Ledbetter, Ph.D., *Emory University School of Medicine*  
Christa Martin, Ph.D., *Emory University School of Medicine*  
*The Jackson Laboratory – Auditorium*
- 7:30-9:00pm      **Nature of Mutations**  
Stylianos E. Antonarakis, M.D., Ph.D., *University of Geneva*  
*Medical School*  
*Bar Harbor Club - Ballroom*

### **Wednesday, July 22**

- 7:45 - 8:25am      **Continental Breakfast**  
*The Jackson Laboratory – Roscoe's*
- 8:30-9:25am      **Transcriptional regulation of gene expression**  
Andrew S. McCallion, Ph.D., *Johns Hopkins University*  
*School of Medicine*  
*The Jackson Laboratory – Auditorium*
- 9:25-10:15am      **Post-transcriptional control of gene expression**  
Harry C. Dietz, M.D., *The Johns Hopkins University*  
*School of Medicine*  
*The Jackson Laboratory – Auditorium*

- 10:15-10:45am      **Break**  
*The Jackson Laboratory – Roscoe’s*
- 10:45-11:40am      **MicroRNAs in Normal Physiology and Disease**  
Joshua T. Mendell, M.D., Ph.D., *The Johns Hopkins University*  
*School of Medicine*  
*The Jackson Laboratory – Auditorium*
- 11:40-12:30pm      **Discovering Protein-Protein Interactions through Mass Spectrometry**  
John R. Yates, Ph.D., *The Scripps Research Institute*  
*The Jackson Laboratory – Auditorium*
- 12:30-2:00pm      **Lunch**  
*The Jackson Laboratory – Roscoe’s*
- 3:00-5:00pm      **Poster Session**  
*Bar Harbor Club – Ballroom*
- 7:30-9:00pm      **Interactome Networks and Human Disease**  
Marc Vidal, Ph.D., *Dana-Farber Cancer Institute*  
*Bar Harbor Club - Ballroom*

**Thursday, July 23**

- 7:45 - 8:25am      **Continental Breakfast**  
*The Jackson Laboratory – Roscoe’s*
- 8:30-9:25am      **Population Genetics in the Genomic Age**  
Aravinda Chakravarti, Ph.D., *The Johns Hopkins University*  
*School of Medicine*  
*The Jackson Laboratory – Auditorium*
- 9:25-10:15am      **Mapping Genes Contributing to Mendelian and Complex Traits**  
Goncalo Abecasis, Ph.D., *University of Michigan*  
*The Jackson Laboratory – Auditorium*
- 10:15-10:45am      **Break**  
*The Jackson Laboratory – Roscoe’s*

- 10:45-11:40am      **Genetic Analysis of Complex Traits**  
Aravinda Chakravarti, Ph.D., *The Johns Hopkins University School of Medicine*  
Goncalo Abecasis, Ph.D., *University of Michigan*  
*The Jackson Laboratory – Auditorium*
- 11:40-12:30pm      **Complex Traits in Mice**  
Gary Churchill, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*
- 12:30-2:00pm      **Lunch**  
*The Jackson Laboratory – Roscoe’s*
- 2:00-4:00pm      **Statistical Genetics Workshop**  
Aravinda Chakravarti, Ph.D., *The Johns Hopkins University School of Medicine*  
Goncalo Abecasis, Ph.D., *University of Michigan*  
*The Jackson Laboratory – Auditorium*
- 7:30-9:00pm      **The Role of Microbial Communities in Health and Disease**  
Claire Fraser-Liggett, Ph.D., *University of Maryland School of Medicine*  
*Bar Harbor Regency – Walsh House*

**Friday, July 24**

- 7:45 - 8:25am      **Continental Breakfast**  
*The Jackson Laboratory – Roscoe’s*
- 8:30-9:25am      **Mitochondrial Medicine**  
Douglas C. Wallace, Ph.D., *University of California, Irvine*  
*The Jackson Laboratory – Auditorium*
- 9:25-10:15am      **Genetics of Obesity and Type II Diabetes**  
Juergen Naggert, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*
- 10:15-10:45am      **Break**  
*The Jackson Laboratory – Roscoe’s*

- 10:45-11:40am      **Genetic Epidemiology of Breast and Ovarian Cancer**  
Lawrence C. Brody, Ph.D., *National Human Genome  
Research Institute*  
*The Jackson Laboratory – Auditorium*
- 11:40-12:30pm      **Genetic Modifiers of CF**  
Garry R. Cutting, M.D., *Johns Hopkins University  
School of Medicine*  
*The Jackson Laboratory – Auditorium*
- 12:30-2:00pm      **Lunch**  
*The Jackson Laboratory – Roscoe’s*
- 2:00-4:00pm      **Workshop: Methods of Molecular Genetics**  
Juergen Naggert, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*
- 7:30-9:00pm      **Comparative Genomics at the Extremes**  
Eddy Rubin, M.D., Ph.D., *Joint Genome Institute  
Lawrence Berkeley National Laboratory*  
*Bar Harbor Regency – Walsh House*

## **Saturday, July 25**

- 7:45 - 8:25am      **Continental Breakfast**  
*The Jackson Laboratory – Roscoe’s*
- 8:30-9:25am      **MHC**  
Derry Roopenian, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*
- 9:25-10:15am      **Genetics of Autoimmunity**  
David Serreze, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*
- 10:15-10:45am      **Break**  
*The Jackson Laboratory – Roscoe’s*
- 10:45-11:40am      **Immunological Disorders in Humans**  
Jennifer Puck, M.D., *University of California, San Francisco*  
*The Jackson Laboratory – Auditorium*

11:40-12:30pm      **Mapping Complex Traits in Mice Using Outbred Stocks**  
Jonathan Flint, M.D., *Oxford University*  
*The Jackson Laboratory – Auditorium*

12:30-2:30pm      **BBQ**  
*The Jackson Laboratory – Roscoe's*

### **Sunday, July 26**

No Scheduled Course Events

### **Monday, July 27**

7:45 - 8:25am      **Continental Breakfast**  
*The Jackson Laboratory – Roscoe's*

8:30-9:25am      **Short Telomeres and Age-related Disease**  
Mary Armanios, M.D., *Johns Hopkins University*  
*The Jackson Laboratory – Auditorium*

9:25-10:15am      **Human Genome Structural Variation and Disease**  
Evan Eichler, Ph.D., *University of Washington*  
*The Jackson Laboratory – Auditorium*

10:15-10:45am      **Break**  
*The Jackson Laboratory – Roscoe's*

10:45-11:40am      **Genetics of Type 2 Diabetes in Humans: Where to from here?**  
David Altschuler, M.D., Ph.D., *Massachusetts General Hospital*  
*The Jackson Laboratory – Auditorium*

11:40-12:30pm      **Transcriptional Responses to Hypoxia**  
Greg Semenza, M.D., Ph.D., *Johns Hopkins University*  
*School of Medicine*

12:30-2:00pm      **Lunch**  
*The Jackson Laboratory – Roscoe's*

2:00-4:00pm      **In Depth Bioinformatics Case Studies**  
Benjamin King, M.S., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*

7:30-9:00pm **Extreme Outliers Identify Rate Determining Steps in Complex Systems: Genetic Dissection of Hypertension**  
Richard Lifton, M.D., Ph.D., *Yale University School of Medicine*  
*Bar Harbor Club – Ballroom*

**Tuesday, July 28**

7:45 - 8:25am **Continental Breakfast**  
*The Jackson Laboratory – Roscoe’s*

8:30-9:25am **Inborn Errors of Metabolism**  
David Valle, M.D., *Johns Hopkins University*  
*School of Medicine*  
*The Jackson Laboratory – Auditorium*

9:25-10:15am **Genetics of Infectious Diseases**  
Steven M. Holland, M.D., *National Institutes of Health*  
*The Jackson Laboratory – Auditorium*

10:15-10:45am **Break**  
*The Jackson Laboratory – Roscoe’s*

10:45-11:40am **Congenital Anomalies in Humans – Clinician’s Approach**  
Judith Hall, M.D., *University of British Columbia and*  
*British Columbia’s Children’s Hospital*  
*The Jackson Laboratory – Auditorium*

11:40-12:30pm **Molecular Genetics of Human Anomalies**  
Ethylin Wang Jabs, M.D., *Mount Sinai School of Medicine*  
*The Jackson Laboratory – Auditorium*

12:30-2:00pm **Lunch**  
*The Jackson Laboratory – Roscoe’s*

1:30-2:30pm **Follow-Up In Depth Bioinformatics Case Studies**  
Benjamin King, M.S., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*

2:30-4:00pm **Mouse Gene Mapping**  
Lucy Rowe, B.A., *The Jackson Laboratory*  
*The Jackson Laboratory – Staff Conference Room*

7:30-9:00pm      **Human Developmental Disorders and Complex Traits**  
Jeffrey Murray, M.D., University of Iowa  
*Bar Harbor Club - Ballroom*

**Wednesday, July 29**

7:45 - 8:25am      **Continental Breakfast**  
*The Jackson Laboratory – Roscoe’s*

8:30-9:25am      **TGFbeta Signaling and Marfan Syndrome**  
Harry C. Dietz, B.S.E., M.D., *The Johns Hopkins University*  
*School of Medicine*  
*The Jackson Laboratory – Auditorium*

9:25-10:05am      **Individualized Medicine**  
Robert Nussbaum, M.D., *University of California, San Francisco*  
*The Jackson Laboratory – Auditorium*

10:05-10:15am      **Introduction to Mouse Genetics Clinic**  
Leah Rae Donahue, Ph.D., *The Jackson Laboratory*  
*The Jackson Laboratory – Auditorium*

10:15-10:45am      **Break**  
*The Jackson Laboratory – Roscoe’s*

10:45-11:40am      **DNA Forensics**  
Philip R. Reilly, M.D., J.D., *Third Rock Ventures*  
*The Jackson Laboratory – Auditorium*

11:40-12:30pm      **Public Policy Challenges in Genetics**  
Kathy Hudson, *Johns Hopkins University*  
*The Jackson Laboratory – Auditorium*

12:30-2:00pm      **Lunch**  
*The Jackson Laboratory – Roscoe’s*

12:30-2:00pm      **Lunch**  
Human Genetics Clinic Selected Participants and Invited Guests  
*Bar Harbor Regency- Eden Restaurant*

1:00-2:00pm      **Follow-Up Mouse Gene Mapping**  
Lucy Rowe, B.A., *The Jackson Laboratory*  
*The Jackson Laboratory – Staff Conference Room*

2:00-4:00pm      **Human Genetics Clinic**  
Ada Hamosh, M.D., M.P.H., *Johns Hopkins University School of Medicine*  
*Bar Harbor Regency- Walsh House*

7:30-9:00pm      **ELSI Case Studies**  
Ada Hamosh, M.D.  
Judith Hall, M.D.  
Laird G. Jackson, M.D.  
John Phillips, III, M.D.  
Philip Reilly, M.D.  
*Bar Harbor Club – Ballroom*

### **Thursday, July 30**

7:45 - 8:25am      **Continental Breakfast**  
*The Jackson Laboratory – Roscoe’s*

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

9:25-10:15am      **Pharmacogenomics: Development, Science and Translation**  
Richard M. Weinshilboum, M.D., *Mayo Clinic*  
*The Jackson Laboratory – Auditorium*

10:15-10:45am      **Break**  
*The Jackson Laboratory – Roscoe’s*

10:45-11:40am      **Prenatal Diagnosis and Screening**  
Laird G. Jackson, M.D., *Drexel University College of Medicine*  
*The Jackson Laboratory – Auditorium*

11:40-12:30pm      **Gene Therapy**  
Katherine A. High, M.D., *Howard Hughes Medical Institute, The Children's Hospital of Philadelphia*  
*The Jackson Laboratory – Auditorium*

12:30-2:00pm      **Lunch**  
*The Jackson Laboratory – Roscoe’s*

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)*

7:30-9:00pm

**Synapses, Genetics and Prospects for Drug Treatment of  
Intellectual Disability**

Rod McInnes, M.D., Ph.D., *The Hospital for Sick Children, Toronto*  
*Bar Harbor Club - Ballroom*

**Friday, July 31**

SEE SYMPOSIUM SCHEDULE