

All lectures at The Jackson Laboratory will be held in the C.C. Little Auditorium unless noted otherwise. These lectures will be live streamed. Evening lectures are at the Walsh House.



49th Annual Short Course on Medical and Experimental Mammalian Genetics

July 20-August 1, 2008

This schedule and is subject to change.

For more information, please contact Nancy Place, nancy.place@jax.org

Please note: All daytime lectures will be held in the C.C. Little Auditorium at The Jackson Laboratory. Evening lectures will be held in the Walsh House at The Bar Harbor Regency.

Sunday, July 20

4:00-7:00pm **Registration at the Walsh House, Bar Harbor Regency**

Monday, July 21

7:30- 8:10 am **Late Registration and Continental Breakfast**
(Main Lobby)

8:15-8:20 am **Overview of Course Evaluations**
Suzanne Hart, *University of Maine*

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

8:30 – 8:45 am **Course Overview**
Patsy Nishina, Ph.D., *The Jackson Laboratory*

8:45 - 9:30 am **History of Medical Genetics**
Victor McKusick, M.D., *Johns Hopkins University*

9:30-10:20a.m. **History of Mouse Genetics**
Ken Paigen, Ph.D., *The Jackson Laboratory*

10:20 -10:45a.m. **Break**

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- 10:45-11:40a.m. **Mendelian Inheritance in Humans**
Clair Francomano, M.D., *Harvey Institute for Human Genetics*
- 11:40-12:30p.m. **Mice as Models for Human Diseases**
Patsy Nishina, Ph.D., *The Jackson Laboratory*
- 12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory
- 2:00-4:00p.m. **Databases for mouse and human genetics**
(In Auditorium)
Introduction
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*
- 7:30-9:00p.m. **What is Genomics?**
Maynard V. Olson, Ph.D., *University of Washington*

Tuesday, July 22

- 7:45 - 8:25 am **Continental Breakfast**
(Main Lobby)
- 8:30-9:25a.m. **Principles & techniques of molecular genetics**
John A. Phillips, III, M.D., *Vanderbilt University School of Medicine*
- 9:25-10:15a.m. **Chromosome Structure, Function and Organization**
Lindsay Shopland, Ph.D., *The Jackson Laboratory*
- 10:15-10:45a.m. **Break**
- 10:45-11:40a.m. **Chromosomes in gametogenesis & in cell division**
David H. Ledbetter, Ph.D., *Emory University School of Medicine*
- 11:40-12:30p.m. **Chromosome aberrations: molecular cytogenetics**
David H. Ledbetter, Ph.D., *Emory University School of Medicine*
- 12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory

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- 2:00-4:00pm **Workshop: Cytogenetics**
(In Auditorium)
David H. Ledbetter, Ph.D., *Emory University School of Medicine*
Christa Martin, Ph.D., *Emory University School of Medicine*
- 7:30-9:00p.m. **Nature of Mutations**
Stylianos E. Antonarakis, M.D., Ph.D., *University of Geneva*
Medical School

Wednesday, July 23

- 7:45 - 8:25 am **Continental Breakfast**
(Main Lobby)
- 8:30-9:25a.m. **Deciphering cis-regulation in development and disease**
Andrew S. McCallion, Ph.D., *Johns Hopkins University*
School of Medicine
- 9:25-10:15a.m. **Post-transcriptional control of gene expression**
Harry C. Dietz, M.D., *The Johns Hopkins University*
School of Medicine
- 10:15-10:45a.m. **Break**
- 10:45-11:40a.m. **MicroRNAs in normal physiology and disease**
Joshua T. Mendell, M.D., Ph.D., *The Johns Hopkins University*
School of Medicine
- 11:40-12:30p.m. **Proteomics**
John R. Yates, Ph.D., *The Scripps Research Institute*
- 12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory
- 3:00-5:00p.m. **Poster Session**
At the Walsh House, Bar Harbor Regency
- 7:30-9:00p.m. **Genetics of Development- complex traits/cleft palate**
Jeffrey C. Murray, M.D., *University of Iowa*

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Thursday, July 24

- 7:45 - 8:25 am **Continental Breakfast**
(Main Lobby)
- 8:30-9:25a.m. **Population Genetics**
Aravinda Chakravarti, Ph.D., *The Johns Hopkins University*
School of Medicine
- 9:25-10:15a.m. **Linkage Analysis of Mendelian Traits**
Goncalo Abecasis, Ph.D., *University of Michigan*
- 10:15-10:45a.m. **Break**
- 10:45-11:40a.m. **Genetic Analysis of Complex Traits**
Aravinda Chakravarti, Ph.D., *The Johns Hopkins University*
School of Medicine
Goncalo Abecasis, Ph.D., *University of Michigan*
- 11:40-12:30p.m. **Complex Traits in Mice**
Gary Churchill, Ph.D., *The Jackson Laboratory*
- 12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory
- 2:00-4:00p.m. **Statistical Genetics Workshop**
(In Auditorium)
Aravinda Chakravarti, Ph.D., *The Johns Hopkins*
University School of Medicine
Goncalo Abecasis, Ph.D., *University of Michigan*
- 7:30-9:00p.m. **Senior Lecturer**
Genomic disorders: mechanisms for copy-number variation (CNV) and clinical implementation of high-resolution genome analysis.
James R. Lupski. M.D., Ph.D., *Baylor College of Medicine*

Friday, July 25

- 7:45 - 8:25 am **Continental Breakfast**
(Main Lobby)
- 8:30-9:25a.m. **Deconstructing Complex Traits**
Joseph H. Nadeau, Ph.D., *Case Western Reserve University*

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- 9:25-10:15a.m. **Genetic Epidemiology of Breast and Ovarian Cancer**
Lawrence C. Brody, Ph.D., *National Human Genome
Research Institute*
- 10:15-10:45a.m. **Break**
- 10:45-11:40a.m. **Pharmacogenomics: Development, Science and Translation**
Richard M. Weinshilboun, M.D., *Mayo Clinic*
- 11:40-12:30p.m. **Individualized Medicine**
Robert Nussbaum, M.D., *University of California,
San Francisco*
- 12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory
- 2:00-4:00p.m. **Workshop: Methods of Molecular Genetics**
(*In Auditorium*)
John A. Phillips, III, M.D., *Vanderbilt University School of
Medicine*
Juergen Naggert, Ph.D., *The Jackson Laboratory*
- 7:30-9:00p.m. **Extreme Comparative Genomics for Deciphering Gene
Regulation**
Eddy Rubin, M.D., Ph.D., *Joint Genome Institute
Lawrence Berkeley National Laboratory*

Saturday, July 26

- 7:45 - 8:25 am **Continental Breakfast**
(*Main Lobby*)
- 8:30-9:25a.m. **Genetics of Obesity and Type II Diabetes**
Juergen Naggert, Ph.D., *The Jackson Laboratory*
- 9:25-10:15a.m. **MHC**
Derry Roopenian, Ph.D., *The Jackson Laboratory*
- 10:15-10:45a.m. **Break**
- 10:45-11:40a.m. **Genetics of Autoimmunity**
David Serreze, Ph.D., *The Jackson Laboratory*
- 11:40-12:30p.m. **Genetics of Infectious Diseases**
Steven M. Holland, M.D., *National Institutes of Health*

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12:30-2:30p.m. **Lunch (barbeque)**
Roscoe's, The Jackson Laboratory

Sunday, July 27

No Lectures Today

Monday, July 28

7:45 - 8:25 am **Continental Breakfast**
(Main Lobby)

8:30-9:25a.m. **Regulation of Oxygen Homeostasis by HIF-1**
Gregg L. Semenza, M.D., Ph.D., *Johns Hopkins University*
School of Medicine

9:25-10:15a.m. **WNT Signaling**
Nicholas Katsanis, Ph.D., *Johns Hopkins University*

10:15-10:45a.m. **Break**

10:45-11:40a.m. **NO regulation in the cardiovascular system**
Charles Lowenstein, M.D., *Johns Hopkins University*

11:40-12:30p.m. **Mitochondrial Genetics**
Douglas C. Wallace, Ph.D., *University of California, Irvine*

12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory

2:00-4:00p.m. **In Depth Bioinformatics Case Studies**
(In Auditorium)
Benjamin King, M.S., *The Jackson Laboratory*

7:30-9:00p.m. **Circadian Biology**
Joseph Takahashi, Ph.D., *Northwestern University,*
Howard Hughes Medical Institute

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Tuesday, July 29

- 7:45 - 8:25 am **Continental Breakfast**
(Main Lobby)
- 8:30-9:25a.m. **Unfolded Protein Response**
Susan Ackerman, Ph.D., *The Jackson Laboratory,*
Howard Hughes Medical Institute
- 9:25-10:15a.m. **Cohesin Biology and the Cohesinopathies**
Ian Krantz, M.D., *The Children's Hospital of Philadelphia*
- 10:15-10:45a.m. **Break**
- 10:45-11:40a.m. **mTOR Signaling**
Ken Inoki, M.D., Ph.D., *University of Michigan*
- 11:40-12:30p.m. **Congenital Anomalies in Humans – Clinician's Approach**
Judith Hall, M.D., *University of British Columbia and*
British Columbia's Children's Hospital
- 12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory
- 2:00-4:00p.m. **Workshop: Mouse Gene Mapping**
(In Staff Conference Room)
Lucy Rowe, B.A., *The Jackson Laboratory*
- In Depth Bioinformatics Case Studies- Follow Up, Q&A**
(In Auditorium)
Benjamin King, M.S., *The Jackson Laboratory*
- 7:30-9:00p.m. **Genetic Architecture: one hundred, and one hundred million years of color variation**
Greg Barsh, M.D., Ph.D., *Stanford University Medical Center*

Wednesday, July 30

- 7:45 - 8:25 am **Continental Breakfast**
(Main Lobby)
- 8:30-9:25a.m. **Notch Signaling**
Thomas Gridley, Ph.D., *The Jackson Laboratory*

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- 9:25-10:15a.m. **TGFbeta Signaling and Marfan Syndrome**
Harry C. Dietz, M.D., *The Johns Hopkins University*
School of Medicine
- 10:15-10:45a.m. **Break**
- 10:45-11:40a.m. **Molecular Genetics of Human Anomalies**
Ethylin Wang Jabs, M.D., *Mount Sinai School of Medicine*
- 11:40-12:30p.m. **Inborn Errors of Metabolism**
David Valle, M.D., *Johns Hopkins University*
School of Medicine
- 12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory
- 12:30-2:00p.m. **Lunch**
Human Genetics Clinic Selected Participants and Invited Guests
Bar Harbor Regency- Eden Restaurant
- 2:00-4:00p.m. **Human Genetics Clinic**
(*At the Walsh House, Bar Harbor Regency*)
Ada Hamosh, M.D., M.P.H., *Johns Hopkins University School of*
Medicine
- 7:30-9:00p.m. **Interactome Networks and Human Disease**
Marc Vidal, Ph.D., *Center for Cancer Systems Biology*
Dana-Farber Cancer Institute and
Harvard Medical School

Thursday, July 31

- 7:45 - 8:25 am **Continental Breakfast**
(*Main Lobby*)
- 8:30-9:25a.m. **Treatment of Genetic Disease**
David Valle, M.D., *Johns Hopkins University*
School of Medicine
- 9:25-10:05a.m. **Immunological Disorders in Humans**
Jennifer Puck, M.D., *University of California, San Francisco*
- 10:05-10:15a.m. **Description of the Mouse Clinic**
Leah Rae Donahue, Ph.D., *The Jackson Laboratory*
- 10:15-10:45a.m. **Break**

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- 10:45-11:40a.m. **DNA Forensics**
Philip R. Reilly, M.D., J.D., *Interleukin Genetics, Inc.*
- 11:40-12:30p.m. **Ethics and Policy in Medical Genetics**
Wylie Burke, M.D., Ph.D., *University of Washington*
- 12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory
- 1:00-2:00p.m. **Workshop: Mouse Gene Mapping, Follow Up, Q & A**
(In Staff Conference Room)
Lucy Rowe, B.A., *The Jackson Laboratory*
- 2:00-3:30 p.m. **Mouse Genetics Clinic**
(Under the Tent)
Leah Rae Donahue, Ph.D., *The Jackson Laboratory*
- Lab Tours**
(Departing from Under the Tent, please sign up in advance)
- 7:30-9:00p.m. **Ethics in Medical and Public Health Genetics**
Wylie Burke, M.D., Ph.D., *University of Washington*
- 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.

Friday, August 1

- 7:45 - 8:25 am **Continental Breakfast**
(Main Lobby)
- 8:30-9:25a.m. **Genetic Modifiers of CF**
Garry R. Cutting, M.D., *Johns Hopkins University*
School of Medicine
- 9:25-10:15a.m. **Prenatal Diagnosis**
Laird G. Jackson, M.D., *Drexel University College of Medicine*
- 10:15-10:45a.m. **Break**
Roscoe's, The Jackson Laboratory

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- 10:45-11:40a.m. **Gene Therapy**
Katherine A. High, M.D., *Howard Hughes Medical Institute,*
The Children's Hospital of Philadelphia
- 11:40-12:30p.m. **Course Wrap-up**
James R. Lupski, M.D., Ph.D., *Baylor College of Medicine*
- 12:30-2:00p.m. **Lunch**
Roscoe's, The Jackson Laboratory

End of Course