

All lectures at The Jackson Laboratory will be held in the LCC Main Auditorium unless noted otherwise. These lectures will be live streamed.



## **48<sup>th</sup> Annual Short Course on Medical and Experimental Mammalian Genetics** *July 15-27, 2007*

For more information, please contact Toni Joerres, [toni.joerres@jax.org](mailto:toni.joerres@jax.org)

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### **Sunday, July 15**

4:30-6:30p.m.      **Event Registration/ Workshop Signup**  
*Regency Hotel, Walsh House*

### **Monday, July 16**

7:15-7:45a.m.      **Late Registration/ Workshop Signup**  
*The Jackson Laboratory, LCC Main Lobby*

8:25-8:30a.m.      **Overview of Course Evaluations**  
Suzanne Hart, University of Maine  
*The Jackson Laboratory*

8:30-8:45a.m.      **Welcome/ Course Overview**  
Rick Woychik, Ph.D., The Jackson Laboratory  
Patsy Nishina, Ph.D., Course Co-Organizer, The Jackson  
Laboratory  
*The Jackson Laboratory*

8:45-9:35a.m.      **History of Medical Genetics**  
Victor McKusick, M.D., Course Co-Organizer, Johns Hopkins  
University  
*The Jackson Laboratory*

9:35-10:15a.m.      **History of Mouse Genetics**  
Kenneth Paigen, Ph.D., The Jackson Laboratory  
*The Jackson Laboratory*

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- 10:15-10:45a.m.      **Coffee Break**  
*The Jackson Laboratory*
- 10:45-11:40a.m.      **Mendelian Inheritance in Humans**  
Clair Francomano, M.D., Greater Baltimore Medical Center  
*The Jackson Laboratory*
- 11:40-12:30p.m.      **Chromosomes in Gametogenesis and in Cell Division**  
David Ledbetter, Ph.D., Emory University School of Medicine  
*The Jackson Laboratory*
- 12:30-2:00p.m.      **Welcome Lunch**  
*Roscoe's, The Jackson Laboratory*
- 1:30-4:30p.m.      **Workshop: Databases for mouse and human genetics**  
**Introduction**  
Carol Bult, Ph.D., The Jackson Laboratory  
**On-line Mendelian Inheritance in Man (OMIM)**  
Ada Hamosh, M.D., Johns Hopkins University  
*The Jackson Laboratory*  
**Mouse Genome Informatics Database: Facilitating mouse as a model for human disease**  
MGI User Support Staff, The Jackson Laboratory
- 7:30-9:00p.m.      **Genomics and the Future of Medicine**  
Eric Lander, Ph.D., The Broad Institute  
*Regency Hotel, Proctor Ballroom*

## **Tuesday, July 17**

- 8:15-8:30a.m.      **March of Dimes Birth Defects Foundation**  
Michael Katz, M.D., Senior Vice President for Research & Global Programs  
*The Jackson Laboratory*
- 8:30-9:25a.m.      **Extreme Comparative Genomics Deciphering Gene Regulation**  
Eddy Rubin, M.D., Ph.D., Joint Genome Institute, Lawrence Berkeley National Laboratory  
*The Jackson Laboratory*
- 9:25-10:15a.m.      **Chromosome Aberrations: Molecular Cytogenetics**  
David Ledbetter, Ph.D., Emory University School of Medicine  
Christa Lese Martin, Ph.D., Emory University School of Medicine  
*The Jackson Laboratory*

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- 10:15-10:45a.m.      **Coffee Break**  
*The Jackson Laboratory*
- 10:45-11:40a.m.      **Chromosome Structure and Function**  
Ethylin Wang Jabs, M.D., Johns Hopkins University  
*The Jackson Laboratory*
- 11:40-12:30p.m.      **Chromosomal Organization**  
Lindsay Shopland, Ph.D., The Jackson Laboratory  
*The Jackson Laboratory*
- 12:30-2:00p.m.      **Lunch**  
*Roscoe's, The Jackson Laboratory*
- 2:00-4:00p.m.      **Workshop:**  
**Cytogenetics**  
David Ledbetter, Ph.D., Emory University School of Medicine  
Christa Lese Martin, Ph.D., Emory University School of Medicine  
*The Jackson Laboratory*
- 7:30-9:00p.m.      **Nature of mutations**  
Stylianos Antonarakis, M.D., Ph.D., University of Geneva Medical School  
*Regency Hotel, Proctor Ballroom*

### **Wednesday, July 18**

- 8:30-9:25a.m.      **Principles and Techniques of Molecular Genetics**  
John Phillips, III, M.D., Vanderbilt University  
*The Jackson Laboratory*
- 9:25-10:15a.m.      **Transcriptional Regulation of Gene Expression**  
Gregg Semenza, M.D., Ph.D., Johns Hopkins University  
*The Jackson Laboratory*
- 10:15-10:45a.m.      **Coffee Break**  
*The Jackson Laboratory*
- 10:45-11:40a.m.      **Post-Transcriptional Control of Gene Expression**  
Harry Dietz, M.D., Johns Hopkins University  
*The Jackson Laboratory*

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- 11:40-12:30p.m.      **Proteomics**  
John Yates, Ph.D., The Scripps Research Institute  
*The Jackson Laboratory*
- 12:30-2:00p.m.      **Lunch**  
*Roscoe's, The Jackson Laboratory*
- 2:00-4:00p.m.      **Poster Session**  
*Regency Hotel, Walsh House*
- 7:30-9:00p.m.      **Interactome Networks and Human Disease**  
Marc Vidal, Ph.D., Center for Cancer Systems Biology, Dana-Farber Cancer Institute and the Center for Cancer Systems Biology  
<http://ccsb.dfci.harvard.edu>  
*Regency Hotel, Proctor Ballroom*

### **Thursday, July 19**

- 8:30-9:25a.m.      **Population Genetics**  
Aravinda Chakravarti, Ph.D., Johns Hopkins University  
*The Jackson Laboratory*
- 9:25-10:15a.m.      **Linkage Analysis of Mendelian Traits**  
Gonçalo Abecasis, Ph.D., University of Michigan  
*The Jackson Laboratory*
- 10:15-10:45a.m.      **Coffee Break – Group Photo** (Rain date July 20<sup>th</sup>)  
*The Jackson Laboratory*
- 10:45-11:40a.m.      **Genetics of Analysis of Complex Traits**  
Aravinda Chakravarti, Ph.D., Johns Hopkins University  
Gonçalo Abecasis, Ph.D., University of Michigan  
*The Jackson Laboratory*
- 11:40-12:30p.m.      **Complex Traits in Mice**  
Sharon Tsaih, Ph.D., The Jackson Laboratory  
*The Jackson Laboratory*
- 12:30-2:00p.m.      **Lunch**  
*Roscoe's, The Jackson Laboratory*

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2:00-4:00p.m.      **Workshops:**  
**Statistical Genetics**  
Aravinda Chakravarti, Ph.D., Johns Hopkins University  
Gonçalo Abecasis, Ph.D., University of Michigan  
*The Jackson Laboratory*

7:30-9:00p.m.      **Genetic Epidemiology in Admixed Populations**  
Neil Risch, Ph.D., University California San Francisco  
*Regency Hotel, Proctor Ballroom*

## **Friday, July 20**

8:30-9:25a.m.      **A Mitochondrial Paradigm for Metabolic and Degenerative Diseases, Aging and Cancer: Why Do We Still Have a Maternally Inherited Mitochondrial DNA?**  
Douglas Wallace, Ph.D., University of California, Irvine  
*The Jackson Laboratory*

9:25-10:15a.m.      **Population Genetics: The Iceland Experiment**  
Jeffrey Gulcher, M.D., Ph.D., Decode Genetics  
*The Jackson Laboratory*

10:15-10:45a.m.      **Coffee Break**  
*The Jackson Laboratory*

10:45-11:40a.m.      **MircoRNA's**  
Josh Mendell, M.D., Ph.D., Johns Hopkins University School of Medicine  
*The Jackson Laboratory*

11:40-12:30p.m.      **Genetics of Schizophrenia and Related Neuropsychiatric disorders**  
Daniel Weinberger, Ph.D., Johns Hopkins University School of Medicine  
*The Jackson Laboratory*

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

1:30-4:30p.m.      **Workshop: Databases for mouse and human genetics**  
**Introduction**  
Carol Bult, Ph.D., The Jackson Laboratory  
**On-line Mendelian Inheritance in Man (OMIM)**  
Ada Hamosh, M.D., Johns Hopkins University  
*The Jackson Laboratory, LCC Staff Conference Room*

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**Mouse Genome Informatics Database: Facilitating mouse as a model for human disease**

MGI User Support Staff, The Jackson Laboratory

**Methods of Molecular Genetics Workshop**

John Phillips, III, M.D., Vanderbilt University  
Jürgen Naggert, Ph.D., Course Co-Organizer, The Jackson Laboratory

*The Jackson Laboratory, LCC Main Auditorium*

7:30-9:00p.m.

**Genetics of Type II Diabetes**

Karen Mohlke, Ph.D., University of North Carolina  
*Regency Hotel, Proctor Ballroom*

**Saturday, July 21**

8:30-9:25a.m.

**Genetics of Age-Related Macular Degeneration**

Rando Allikmets, Ph.D., Columbia University  
*The Jackson Laboratory*

9:25-10:15a.m.

**Genetic Epidemiology of Breast and Ovarian Cancer**

Lawrence Brody, Ph.D., National Human Genome Research Institute

*The Jackson Laboratory*

10:15-10:45a.m.

**Coffee Break**

*The Jackson Laboratory*

10:45-11:40a.m.

**Genomic Analysis of Human Cancer**

Victor Velculescu, M.D., Ph.D., Johns Hopkins University School of Medicine

*The Jackson Laboratory*

11:40-12:30p.m.

**Pharmacogenomics: Development, Science and Translation**

Richard Weinshilboum, M.D., Mayo Clinic

*The Jackson Laboratory*

12:30-2:00p.m.

**Barbecue**

*Roscoe's, The Jackson Laboratory*

**Sunday, July 22**

*No Scheduled Course Events*

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## **Monday, July 23**

- 8:30-9:25a.m.      **MHC**  
Derry Roopenian, Ph.D., The Jackson Laboratory  
*The Jackson Laboratory*
- 9:25-10:15a.m.      **Genetics of Autoimmunity**  
David Serreze, Ph.D. The Jackson Laboratory  
*The Jackson Laboratory*
- 10:15-10:45a.m.      **Coffee Break**  
*The Jackson Laboratory*
- 10:45-11:40a.m.      **Genetic Disorders of the Human Immune System**  
Jennifer Puck, M.D., National Human Genome Research Institute  
*The Jackson Laboratory*
- 11:40-12:30p.m.      **Genetics of Infectious Diseases**  
Steven Holland, M.D., National Institutes of Health  
*The Jackson Laboratory*
- 12:30-1:45p.m.      **Lunch**  
*Roscoe's, The Jackson Laboratory*
- 1:45-2:15p.m.      **Mice as Models for Human Diseases**  
Patsy Nishina, Ph.D., Course Co-Organizer, The Jackson  
Laboratory  
*The Jackson Laboratory*
- 2:15-4:15p.m.      **Workshops: Case studies**  
**Mouse Gene Mapping**  
Lucy Rowe, B.A., The Jackson Laboratory  
*The Jackson Laboratory*  
**In Depth Bioinformatics**  
Benjamin King, M.S., The Jackson Laboratory  
*The Jackson Laboratory, LCC Staff Room*
- 7:30-9:00p.m.      **Inflammatory Bowel Disease**  
Judy Cho, M.D., University of Yale Medical School  
*Regency Hotel, Proctor Ballroom*

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## **Tuesday, July 24**

- 8:30-9:25a.m.      **Inborn Errors of Metabolism**  
David Valle, M.D., Course Co-Organizer, Johns Hopkins University  
*The Jackson Laboratory*
- 9:25-10:15a.m.      **Development**  
Kyuson Yun, Ph.D., The Jackson Laboratory  
*The Jackson Laboratory*
- 10:15-10:45a.m.      **Coffee Break**  
*The Jackson Laboratory*
- 10:45-11:40a.m.      **Molecular Genetics of Human Anomalies**  
Ethylin Wang Jabs, M.D., Johns Hopkins University  
*The Jackson Laboratory*
- 11:40-12:30p.m.      **Congenital Anomalies in Humans—Clinicians Approach**  
Judith Hall, M.D., UBC & BC's Children's Hospital  
*The Jackson Laboratory*
- 12:30-1:45p.m.      **Lunch**  
*Roscoe's, The Jackson Laboratory*
- 1:45-2:15p.m.      **A physiological systems approach to mouse phenotyping**  
Anthony Nicholson, Ph.D., The Jackson Laboratory  
*The Jackson Laboratory*
- 2:15-4:15p.m.      **Workshops: solution to case studies**  
**Mouse Gene Mapping**  
Lucy Rowe, B.A., The Jackson Laboratory  
*The Jackson Laboratory*  
**In Depth Bioinformatics**  
Benjamin King, M.S., The Jackson Laboratory  
*The Jackson Laboratory, LCC Staff Room*
- 7:30-9:00p.m.      **Genetics of Development-Complex Traits/Cleft Palate**  
Jeffrey Murray, M.D., University of Iowa  
*Regency Hotel, Proctor Ballroom*

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## **Wednesday, July 25**

- 8:30-9:25a.m.      **Genetics of Obesity and Type II Diabetes**  
Jürgen Naggert, Ph.D., Course Co-Organizer, The Jackson Laboratory  
*The Jackson Laboratory*
- 9:25-10:15a.m.    **Genetics of Bardet-Biedl Syndrome**  
Nicholas Katsanis, Ph.D., Johns Hopkins University  
*The Jackson Laboratory*
- 10:15-10:45a.m.   **Coffee Break**  
*The Jackson Laboratory*
- 10:45-11:40a.m.   **Genetics of Parkinson Disease**  
Robert Nussbaum, M.D., National Human Genome Research Institute  
*The Jackson Laboratory*
- 11:40-12:30p.m.   **Marfan Syndrome: Disease Mechanisms and New Ideas About Treatment**  
Harry Dietz, M.D., Johns Hopkins University  
*The Jackson Laboratory*
- 12:30-2:00p.m.    **Lunch**  
*Roscoe's, The Jackson Laboratory*
- Lunch**  
Human Genetic Invited Guests  
*Regency- Eden Restaurant*
- 2:30-4:30p.m.      **Workshop:**  
**Human Genetics Clinic**  
Ada Hamosh, M.D., Johns Hopkins University  
Clair Francomano, M.D., Greater Baltimore Medical Center  
*Regency Hotel, Proctor Ballroom*
- 7:30-9:00p.m.      **TBA**  
**Senior Lecturer:** Peter Goodfellow, Ph.D., deCODE Genetics  
*Regency Hotel, Proctor Ballroom*

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## **Thursday, July 26**

- 8:30-9:25a.m.      **Treatment of Genetic Disease**  
David Valle, M.D., Course Co-Organizer, Johns Hopkins University  
*The Jackson Laboratory*
- 9:25-10:00a.m.      **Genetics and Personalized Medicine**  
Robert Nussbaum, M.D., National Human Genome Research Institute  
*The Jackson Laboratory*
- 10:00-10:15a.m.      **Introduction to Mouse Genetics Clinic**  
Leah Rae Donahue, Ph.D., The Jackson Laboratory  
*The Jackson Laboratory*
- 10:15-10:45a.m.      **Coffee Break**  
*The Jackson Laboratory*
- 10:45-11:40a.m.      **Social and Legal Implications of DNA Sequence Information**  
Philip Reilly, M.D.,J.D, Interleukin Genetics, Inc.  
*The Jackson Laboratory*
- 11:40-12:30p.m.      **Ethics and Policy in Medical Genetics**  
Wylie Burke, M.D., Ph.D., University of Washington  
*The Jackson Laboratory*
- 12:30-2:00p.m.      **Lunch**  
*Roscoe's, The Jackson Laboratory*
- 2:00-3:30p.m.      **Workshop:**  
**Mouse Genetics Clinic**  
Leah Rae Donahue, Ph.D., The Jackson Laboratory  
*The Jackson Laboratory, LCC Guest Parking Lot*  
**Tours of The Jackson Laboratory**  
*(please see shuttle schedule)*
- 7:30-9:00p.m.      **Ethics in medical and public health genetics/ ELSI case studies**  
Wylie Burke, M.D., Ph.D., University of Washington  
Clair Francomano, M.D., Greater Baltimore Medical Center  
Ada Hamosh, M.D., Johns Hopkins University  
Judith Hall, M.D., UBC & BC's Children's Hospital  
Laird G. Jackson, M.D., Drexel University College of Medicine  
John Phillips, III, M.D., Vanderbilt University  
Philip Reilly, M.D., Interleukin Genetics, Inc.  
*Regency Hotel, Proctor Ballroom*

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**Friday, July 27**

- 8:30-9:25a.m.      **Prenatal Diagnosis**  
Laird Jackson, M.D., Drexel University College of Medicine  
*The Jackson Laboratory*
- 9:25-10:15a.m.    **DNA and Diagnosis and Screening**  
Garry Cutting, M.D., Johns Hopkins University  
*The Jackson Laboratory*
- 10:15-10:45a.m.   **Coffee Break**  
*The Jackson Laboratory*
- 10:45-11:40a.m.   **Gene Transfer and Novel Genetic Therapies for Genetic Disease**  
Katherine High, M.D., The Children's Hospital of Philadelphia/HHMI  
*The Jackson Laboratory*
- 11:40-12:30p.m.   **Course Wrap Up**  
*Senior Lecturer:* Peter Goodfellow, Ph.D., deCODE Genetics  
*The Jackson Laboratory*
- 12:30-2:00p.m.    **Lunch**  
*Roscoe's, The Jackson Laboratory*

*END OF COURSE*