



**47th Annual Short Course on Medical and
Experimental Mammalian Genetics
*July 16-28, 2006***

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

Sunday, July 16

4:30-6:30p.m. **Event Registration/ Workshop Signup**
Proctor Ballroom, Bar Harbor Regency

Monday, July 17

7:15-7:45a.m. **Late Registration/ Workshop Signup**
Proctor Ballroom, Bar Harbor Regency

8:25-8:30a.m. **Overview of Course Evaluations**
Suzanne Hart, University of Maine
Gates Auditorium, College of the Atlantic

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
Gates Auditorium, College of the Atlantic

8:45-9:25a.m. **History of Medical Genetics**
Victor McKusick, M.D., Course Co-Organizer, Johns Hopkins
University
Gates Auditorium, College of the Atlantic

9:25-10:15a.m. **History of Mouse Genetics**
Kenneth Paigen, Ph.D., The Jackson Laboratory
Gates Auditorium, College of the Atlantic

- 10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic
- 10:45-11:40a.m. **Mendelian inheritance in humans**
Clair Francomano, M.D., Greater Baltimore Medical Center
Gates Auditorium, College of the Atlantic
- 11:40-12:30p.m. **Mice as models for human diseases**
Patsy Nishina, Ph.D., Course Co-Organizer, The Jackson
Laboratory
Gates Auditorium, College of the Atlantic
- 12:30-2:00p.m. **Welcome Lunch**
Course Tent, College of the Atlantic
- 2:00-4:00p.m. **Mini-Symposia:**
Overview Bioinformatics
Carol Bult, Ph.D., The Jackson Laboratory
Computerized Databases: OMIM
Ada Hamosh, M.D., Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 7:30-9:00p.m. **Primate Evolution**
Svante Paabo, Ph.D., Max Planck Institute for Evolutionary
Anthropology
Proctor Ballroom, Bar Harbor Regency

Tuesday, July 18

- 8:15-8:30a.m. **March of Dimes Birth Defects Foundation**
Michael Katz, M.D., Senior Vice President for Research & Global
Programs
Gates Auditorium, College of the Atlantic
- 8:30-9:25a.m. **Chromosome structure and function**
Ethylin Wang Jabs, M.D., Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 9:25-10:15a.m. **Chromosome Organization**
Lindsay Shopland, Ph.D., University of Maine/ The Jackson
Laboratory
Gates Auditorium, College of the Atlantic
- 10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic

- 10:45-11:40a.m. **Chromosomes in gametogenesis and in cell division**
David Ledbetter, Ph.D., Emory University School of Medicine
Gates Auditorium, College of the Atlantic
- 11:40-12:30p.m. **Chromosome aberrations: molecular cytogenetics**
David Ledbetter, Ph.D., Emory University School of Medicine
Christina Lese Martin, Ph.D., Emory University School of
Medicine
Gates Auditorium, College of the Atlantic
- 12:30-2:00p.m. **Lunch with Q&A Session**
Course Tent, College of the Atlantic
- 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
Proctor Ballroom, Bar Harbor Regency
- 7:30-9:00p.m. **Mitochondrial genetics**
Douglas Wallace, Ph.D., University of California, Irvine
Proctor Ballroom, Bar Harbor Regency

Wednesday, July 19

- 8:30-9:25a.m. **Principles and techniques of molecular genetics**
John Phillips, III, M.D., Vanderbilt University
Gates Auditorium, College of the Atlantic
- 9:25-10:15a.m. **Transcriptional regulation of gene expression**
Gregg Semenza, M.D., Ph.D., Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic
- 10:45-11:40a.m. **Post-transcriptional control of gene expression**
Harry Dietz, M.D., Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 11:40-12:30p.m. **Proteomics**
John Yates, Ph.D., The Scripps Research Institute
Gates Auditorium, College of the Atlantic

- 12:30-2:00p.m. **Lunch with Q&A Session**
Course Tent, College of the Atlantic
- 2:00-4:00p.m. **Workshop: Methods of Molecular Genetics Workshop**
John Phillips, III, M.D., Vanderbilt University
Jüergen Naggert, Ph.D., Course Co-Organizer, The Jackson
Laboratory
Proctor Ballroom, Bar Harbor Regency
Computerized Databases: MGI
Gates Auditorium, College of the Atlantic
- 7:30-9:00p.m. **Nature of mutations**
Stylianos Antonarakis, M.D., Ph.D., University of Geneva Medical
School
Proctor Ballroom, Bar Harbor Regency

Thursday, July 20

- 8:30-9:25a.m. **Population genetics**
Aravinda Chakravarti, Ph.D., Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 9:25-10:15a.m. **Linkage analysis of Mendelian traits**
Gonçalo Abecasis, Ph.D., University of Michigan
Gates Auditorium, College of the Atlantic
- 10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic
- 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
Gates Auditorium, College of the Atlantic
- 11:40-12:30p.m. **Complex traits in mice**
Gary Churchill, Ph.D., The Jackson Laboratory
Gates Auditorium, College of the Atlantic
- 12:30-2:00p.m. **Lunch with Q&A Session**
Course Tent, College of the Atlantic

2:00-4:00p.m.

Workshops:

Statistical genetics

Aravinda Chakravarti, Ph.D., Johns Hopkins University

Gonçalo Abecasis, Ph.D., University of Michigan

Walsh House, Bar Harbor Regency

Optional mouse gene mapping workshop, lecture session

Lucy Rowe, B.A., The Jackson Laboratory

Proctor Ballroom, Bar Harbor Regency

7:30-9:00p.m.

Extreme comparative genomics

Eddy Rubin, M.D., Ph.D., Joint Genome Institute, Lawrence

Berkeley National Laboratory

Proctor Ballroom, Bar Harbor Regency

Friday, July 21

8:30-9:25a.m.

Genetic epidemiology of breast and ovarian cancer

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

Gates Auditorium, College of the Atlantic

9:25-10:15a.m.

MicroRNAs in health and disease

Joshua Mendell, M.D., Ph.D., Johns Hopkins University

Gates Auditorium, College of the Atlantic

10:15-10:45a.m.

Coffee Break

Porch, College of the Atlantic

10:45-11:40a.m.

**Human genome sequence variation and the inherited basis of
disease**

David Altshuler, M.D., Ph.D., Massachusetts General Hospital

Gates Auditorium, College of the Atlantic

11:40-12:30p.m.

**Using the mouse and bioinformatics to find human disease
genes**

Beverly Paigen, Ph.D., The Jackson Laboratory

Gates Auditorium, College of the Atlantic

12:30-2:00p.m.

Lunch with Q&A Session

Course Tent, College of the Atlantic

2:00-4:00p.m. ***Faculty and Participant Poster Session***
Proctor Ballroom, Bar Harbor Regency

7:30-9:00p.m. **Interactome networks and human disease**
Marc Vidal, Ph.D., Dana-Farber Cancer Institute and the Center
for Cancer Systems Biology
Proctor Ballroom, Bar Harbor Regency

Saturday, July 22

8:30-9:25a.m. **General principles of human evolution and how they relate to disease**
Kenneth Weiss, Ph.D., Penn State University
Gates Auditorium, College of the Atlantic

9:25-10:15a.m. **Segmental duplication and structural polymorphisms**
Evan Eichler, Ph.D., University of Washington
Gates Auditorium, College of the Atlantic

10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic

10:45-11:40a.m. **Human evolution, genetics, and the concept of race**
Lynn Jorde, Ph.D., University of Utah
Gates Auditorium, College of the Atlantic

11:40-12:30p.m. **Population Genetics: The Iceland Experiment**
Jeffrey Gulcher, M.D., Ph.D., deCODE
Gates Auditorium, College of the Atlantic

12:30-2:00p.m. **Barbecue**
Course Tent, College of the Atlantic

Sunday, July 23

No Scheduled Course Events

Monday, July 24

- 8:30-9:25a.m. **MHC**
Derry Roopenian, Ph.D., The Jackson Laboratory
Gates Auditorium, College of the Atlantic
- 9:25-10:15a.m. **Genetics of autoimmunity**
David Serreze, Ph.D. The Jackson Laboratory
Gates Auditorium, College of the Atlantic
- 10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic
- 10:45-11:40a.m. **Immunological disorders in humans**
Jennifer Puck, M.D., National Human Genome Research Institute
Gates Auditorium, College of the Atlantic
- 11:40-12:30p.m. **Genetics of infectious diseases**
Steve Holland, M.D., National Institutes of Health
Gates Auditorium, College of the Atlantic
- 12:30-2:00p.m. **Lunch with Q&A Session**
Course Tent, College of the Atlantic
- 2:00-4:00p.m. **Workshops:**
Mouse gene mapping
Lucy Rowe, B.A., The Jackson Laboratory
Proctor Ballroom, Bar Harbor Regency
In Depth Bioinformatics Case Studies
Benjamin King, M.S., The Jackson Laboratory
Gates Auditorium, College of the Atlantic
- 7:30-9:00p.m. **Cytokines and disease**
James Ihle, Ph.D., St. Jude's Research Hospital
Proctor Ballroom, Bar Harbor Regency

Tuesday, July 25

- 8:30-9:25a.m. **Inborn errors of metabolism**
David Valle, M.D., Course Co-Organizer, Johns Hopkins
University
Gates Auditorium, College of the Atlantic
- 9:25-10:15a.m. **Development**
Kyuson Yun, Ph.D., The Jackson Laboratory
Gates Auditorium, College of the Atlantic

- 10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic
- 10:45-11:40a.m. **Molecular genetics of human anomalies**
Ethylin Wang Jabs, M.D., Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 11:40-12:30p.m. **Human congenital anomalies**
Judith Hall, M.D., UBC & BC's Children's Hospital
Gates Auditorium, College of the Atlantic
- 12:30-2:00p.m. **Lunch with Q&A Session**
Course Tent, College of the Atlantic
- 2:00-4:00p.m. **Mouse Phenome Database**
Molly Bogue, Ph.D. The Jackson Laboratory
Proctor Ballroom, Bar Harbor Regency
A physiological systems approach to mouse phenotyping
Anthony Nicholson, Ph.D., The Jackson Laboratory
The Jackson Laboratory (see shuttle schedule)
- 7:30-9:00p.m. **Genetics of development and organogenesis**
Brigid Hogan, Ph.D., Duke University Medical Center
Proctor Ballroom, Bar Harbor Regency

Wednesday, July 26

- 8:30-9:25a.m. **Marfan Syndrome**
Harry Dietz, M.D., Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 9:25-10:15a.m. **Genetics of Parkinson Disease**
Robert Nussbaum, M.D., National Human Genome Research
Institute
Gates Auditorium, College of the Atlantic
- 10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic
- 10:45-11:40a.m. **Genetics of obesity and Type II diabetes**
Jürgen Naggert, Ph.D., Course Co-Organizer, The Jackson
Laboratory
Gates Auditorium, College of the Atlantic

- 11:40-12:30p.m. **Genetics of Bardet-Biedl syndrome**
Nicholas Katsanis, Ph.D., Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 12:30-2:00p.m. **Lunch with Q&A Session**
Course Tent, College of the Atlantic
- 2:00-4:00p.m. **Workshop:**
Human Genetics Clinic
Ada Hamosh, M.D., Johns Hopkins University
Proctor Ballroom, Bar Harbor Regency
- 7:30-9:00p.m. **Senior Lecturer: The Genetics of Vision: From Molecules to Mechanisms**
Roderick McInnes, M.D., Ph.D., University of Toronto
Proctor Ballroom, Bar Harbor Regency

Thursday, July 27

- 8:30-9:25a.m. **Treatment of genetic disease**
David Valle, M.D., Course Co-Organizer, Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 9:25-10:05a.m. **Skeletal dysplasias**
Julie Hoover-Fong, M.D., Johns Hopkins University
Gates Auditorium, College of the Atlantic
- 10:05-10:15a.m. **Introduction to Mouse genetics clinic**
Leah Rae Donahue, Ph.D., The Jackson Laboratory
Gates Auditorium, College of the Atlantic
- 10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic
- 10:45-11:40a.m. **Ethical, legal and social implications of the genetic revolution**
Jeffrey Botkin, M.D., M.P.H., University of Utah
Gates Auditorium, College of the Atlantic
- 11:40-12:30p.m. **Social and legal implications of DNA sequence information**
Phil Reilly, M.D.,J.D, Interleukin Genetics, Inc.
Gates Auditorium, College of the Atlantic

- 12:30-2:30p.m. **Lunch with Q&A Session**
Course Tent, College of the Atlantic
- 2:00-4:00p.m. **Workshop:**
Mouse Genetics Clinic
 Leah Rae Donahue, Ph.D., The Jackson Laboratory
The Jackson Laboratory (please see shuttle schedule)
Tours of The Jackson Laboratory
(please see shuttle schedule)
- 7:30-9:00p.m. **Ethics in medical and public health genetics/ ELSI case studies**
 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.
Proctor Ballroom, Bar Harbor Regency

Friday, July 28

- 8:30-9:25a.m. **Prenatal diagnosis**
 Laird Jackson, M.D., Drexel University College of Medicine
Gates Auditorium, College of Atlantic
- 9:25-10:15a.m. **DNA and diagnosis and screening**
 Garry Cutting, M.D., Johns Hopkins University
Gates Auditorium, College of Atlantic
- 10:15-10:45a.m. **Coffee Break**
Porch, College of the Atlantic
- 10:45-11:40a.m. **Gene therapy**
 Katherine High, M.D., The Children's Hospital of
 Philadelphia/HHMI
Gates Auditorium, College of Atlantic
- 11:40-12:30p.m. **Course Wrap Up**
 Roderick McInnes, M.D., Ph.D., University of Toronto
Gates Auditorium, College of Atlantic
- 12:30-2:30p.m. **Lunch with Q&A Session**
Course Tent, College of the Atlantic

END OF COURSE