Cell and Molecular Biology 550  “GENETIC PRINCIPLES” Spring Semester 2013
Monday, Wednesday, Friday 10-11:30 am, 1301 BRBII/III

This is a combined lecture and discussion course that surveys major concepts and approaches used in model organism and human genetics. Discussions are problem-based and emphasize practical aspects of generating and interpreting genetic data.

Course Directors: Meera Sundaram, 446a CRB, 573-4527, sundaram@mail.med.upenn.edu
Struan Grant, 1216F ARC, 267-426-2795, grants@email.chop.edu

Format: Monday and Wednesday, 1 - 1.5-hour lectures
Friday, 1.5 hour discussion of assigned problem sets

Grading: 25% Class participation (Discussion of assigned problems)
75% Exams (1 in-class exam and 2 take-home exams)

Supplementary textbooks available online: Griffiths et al. “Introduction to Genetic Analysis”
Strachan and Read, “Human Molecular Genetics”

I. GENETIC CONCEPTS

1. Beyond Mendel
   Lecturer: M. Sundaram
   Date: Jan 09
   DISCUSSION: Jan 11

   MARTIN LUTHER KING’S BIRTHDAY – NO CLASS
   Date: Jan 14

2. Chromosome segregation and recombination
   Lecturer: M. Sundaram
   Date: Jan 16
   DISCUSSION: Jan 18

3. Linkage, WGS and positional cloning in model organisms
   Lecturer: M. Sundaram
   Date: Jan 21

4. Linkage mapping in human pedigrees
   Lecturer: M. DeVoto
   Date: Jan 23
   DISCUSSION: Jan 25

5. Determining how mutations affect gene function
   Lecturer: M. Sundaram
   Date: Jan 28

6. Mutagenesis and forward genetic screens
   Lecturer: T. Jongens
   Date: Jan 30
   DISCUSSION: Feb 01

7. Quantitative traits
   Lecturer: E. Brodkin
   Date: Feb 04

8. Genomics for Geneticists
   Lecturer: K. Kaestner
   Date: Feb 06
   DISCUSSION: Feb 08

9. Transposable elements
   Lecturer: R. Bushman
   Date: Feb 11
   DISCUSSION: Feb 13

1ST EXAM (IN CLASS)

Date: Feb 15
II. GENETICS OF MODEL ORGANISMS

1. C. elegans
   Lecturer: J. Parry   Date: Feb 18
2. Drosophila
   Lecturer: A. Ghabrial   Date: Feb 20
   DISCUSSION   Date: Feb 22

3. Mosaic analysis
   Lecturer: M. Sundaram   Date: Feb 25
4. Epistasis and genetic modifier screens
   Lecturer: M. Sundaram   Date: Feb 27
   DISCUSSION   Date: Mar 01

5. Maternal effect and sterile mutants
   Lecturer: T. Jongens   Date: Mar 11
6. RNAi and miRNAs
   Lecturer: B. Gregory   Date: Mar 13
   DISCUSSION   Date: Mar 15

7. Mouse knockouts and transgenics
   Lecturer: K. Kaestner   Date: Mar 18
8. Forward genetics and genomics in the mouse
   Lecturer: M. Bucan   Date: Mar 20
   DISCUSSION   Date: Mar 22

SPRING BREAK   NO CLASS

2ND EXAM (TAKE HOME MAR 22 - MAR 29)

III. HUMAN GENETICS AND DISEASE

1. Population genetics
   Lecturer: J. Lachance   Date: Mar 25
2. Human evolution
   Lecturer: J. Lachance   Date: Mar 27
   DISCUSSION   Date: Mar 29

3. Family-based analyses and exome sequencing
   Lecturer: S. Grant   Date: Apr 01
4. Genome wide genetics for complex traits
   Lecturer: S. Grant   Date: Apr 03
   DISCUSSION   Date: Apr 05

5. Chromosomai abnormalities
   Lecturer: N. Spinner   Date: Apr 08
6. X inactivation
   Lecturer: M. Bartolomei   Date: Apr 10
   DISCUSSION   Date: Apr 12

7. Mitochondrial genetics
   Lecturer: M. Falk   Date: Apr 15
8. Cancer genetics and personalized medicine
   Lecturer: A. Ganguly   Date: Apr 17
   DISCUSSION   Date: Apr 19

3RD EXAM (TAKE HOME APR 19 - APR 24)
Cell and Molecular Biology 550  “GENETIC PRINCIPLES” Spring Semester 2013

This is a combined lecture and discussion course that surveys major concepts and approaches used in model organism and human genetics.

Goals of the course

Students will be able to:

• Recognize and understand the molecular basis for different patterns of inheritance
• Understand the factors that generate and shape patterns of genetic variation
• Understand basic principles and approaches for forward genetics in model organisms and humans - how can you go from a phenotype to a molecular understanding of the causative variant(s)?
• Understand basic principles and approaches for reverse genetics in model organisms - given a gene of known sequence, how can you use genetic approaches to determine its biological functions?
• Be comfortable accessing genetic information from the primary literature and online databases
• Understand the difference between necessity and sufficiency
• Understand the difference between association and causality

Grading Policy

Grades will be based on three exams (100 points each) and Discussion participation (100 points), for a possible total of 400 points. Letter grading will be based on a curve. Those with scores above the mean will usually receive some sort of an “A” (A+, A or A-), while those with scores below the mean will receive some sort of a “B”. Those with scores more than two standard deviations below the mean will receive a C or below.

The first exam will be in-class (closed book) and covers basic genetic concepts that are the foundation for the rest of the course. The second and third exam will be in take-home (open book) format; these exams will test your ability to design and interpret genetic experiments. The take-home exams must be prepared independently without ANY outside consultation.

Discussion guidelines

The homework problems and discussion are the most important part of this course. Each lecturer will provide assigned homework problems on the day of their lecture. Students are expected to complete the homework problems prior to Friday discussion; it is fine to work collaboratively in a “study group”. Homework will NOT be collected. However, students will be randomly chosen to answer questions during Discussion.

Discussion grades will be based on:

• attendance
• preparation (e.g. ability to answer questions when called upon)
• engagement (e.g. voluntary participation in discussion)
CAMB 550 Lecturers – 2013

Marisa Bartolomei
Dept. of Cell & Dev Biology
363 CRB, 8-9063
bartolom@mail.med.upenn.edu

Edward Brodkin
Dept. of Psychiatry
2220 TRL, 215-746-0118
ebrodkin@mail.med.upenn.edu

Maja Bucan
Dept of Genetics
528 CRB, 8-0020
bucan@pobox.upenn.edu

Rick Bushman
Dept. of Microbiology
426 Johnson Pavilion, 3-8732
bushman@mail.med.upenn.edu

Marcella DeVoto
CHOP Division of Human Genetics
Abramson 1002, 267-426-0124
devoto@email.chop.edu

Marni Falk
CHOP Division of Human Genetics
Abramson 1002c, 215-590-4564
falkm@mail.chop.edu

Arupa Ganguly
Dept of Genetics
415 Anatomy-Chemistry, 8-3122
ganguly@mail.med.upenn.edu

Amin Ghabrial
Dept of Cell & Dev. Biology
1214 BRBII/III, 8-7805
ghabrial@mail.med.upenn.edu

Struan Grant
CHOP, Center for Applied Genomics
Abramson 1216F, 267-426-2795
grants@email.chop.edu

Brian Gregory
Dept of Biology
131 Carolyn Lynch Laboratory
bdgregor@sas.upenn.edu

Tom Jongens
Dept of Genetics
10-134 TRC, 3-9332
jongens@mail.med.upenn.edu

Klaus Kaestner
Dept of Genetics
12-126 TRC, 8-8759
kaestner@mail.med.upenn.edu

Joe Lachance
Postdoc, Tishkoff lab
Dept. of Genetics
430 CRB,
lachance.joseph@gmail.com

Jean Parry
Postdoc, Sundaram lab
Dept. of Genetics
445 CRB, 3-4528
jparry@mail.med.upenn.edu

Nancy Spinner
CHOP Division of Human Genetics
Abramson 1007a, 215-590-4177
spinner@mail.med.upenn.edu

Meera Sundaram
Dept of Genetics
446A CRB, 3-4527
sundaram@mail.med.upenn.edu