

Cell and Molecular Biology 550 “GENETIC PRINCIPLES” Spring Semester 2011
Monday, Wednesday, Friday 10-11:30 am, 252 BRBII/III

This is a combined lecture and discussion course that surveys major concepts and approaches used in experimental 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
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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”
<http://www.ncbi.nlm.nih.gov:80/books/>

I. GENETIC CONCEPTS

	<u>Lecturer</u>	<u>Date</u>
1. Mendelian genetics	M. Sundaram DISCUSSION	Jan 12 Jan 14
MARTIN LUTHER KING’S BIRTHDAY – NO CLASS	-	Jan 17
2. Human pedigrees and inherited disease	D. Epstein DISCUSSION	Jan 19 Jan 21
3. Chromosome segregation and recombination	M. Sundaram	Jan 24
4. Linkage mapping and positional cloning	M. Sundaram DISCUSSION	Jan 26 Jan 28
5. Mutagenesis and forward genetic screens	T. Jongens	Jan 31
6. Determining how mutations affect gene function	M. Sundaram DISCUSSION	Feb 02 Feb 04
7. Genomics for Geneticists	K. Kaestner	Feb 07
8. Transposable elements	R. Bushman DISCUSSION	Feb 09 Feb 11
9. Quantitative traits	E. Brodtkin	Feb 14
1ST EXAM (IN CLASS)	- DISCUSSION	Feb 16 Feb 18

II. GENETICS OF MODEL ORGANISMS

	<u>Lecturer</u>	<u>Date</u>
1. <i>C. elegans</i>	D. Raizen	Feb 21
2. <i>Drosophila</i>	A. Ghabrial	Feb 23
	DISCUSSION	Feb 25
3. Mosaic analysis	M. Sundaram	Feb 28
4. Maternal effect and sterile mutants	T. Jongens	Mar 02
	DISCUSSION	Mar 04
		Mar 07
		Mar 09
		Mar 11
5. Epistasis and genetic modifier screens	M. Sundaram	Mar 14
6. RNAi and miRNAs	B. Gregory	Mar 16
	DISCUSSION	Mar 18
7. Mouse knockouts and transgenics	K. Kaestner	Mar 21
8. Forward genetics and genomics in the mouse	M. Bucan	Mar 23
	DISCUSSION	Mar 25

SPRING BREAK NO CLASS

2ND EXAM (TAKE HOME MAR 25 - MAR 30)**III. HUMAN GENETICS AND DISEASE**

1. Population genetics and human evolution	S. Tishkoff	Mar 28
2. Variation, linkage vs. association studies, p. I	V. Cheung	Mar 30
	DISCUSSION	Apr 01
3. Variation, linkage vs. association studies, p. II	V. Cheung	Apr 04
4. Mitochondrial genetics	M. Falk	Apr 06
	DISCUSSION	Apr 08
5. Chromosomal abnormalities	N. Spinner	Apr 11
6. X inactivation	M. Bartolomei	Apr 13
	DISCUSSION	Apr 15
7. Genetic diagnostic testing	A. Ganguly	Apr 18
8. Cancer genetics	D. George	Apr 20
	DISCUSSION	Apr 22

3RD EXAM (TAKE HOME APR 22 - APR 27)

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This is a combined lecture and discussion course that surveys major concepts and approaches used in experimental 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 – 2011

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