FINAL 11-11-19

Cell and Molecular Biology 550 "GENETIC PRINCIPLES" Spring Semester 2020
Monday, Wednesday, Friday 10-11:30 am, Room 251 BRB

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: Eric Joyce, 564 CRB, 898-1229, erjoyce@upenn.edu
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Office hours: Thursday 3-5PM CRB300

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 AND TOOLS

	Lecturer	**Date**
1. Beyond Mendel	E. Joyce	Jan 15
	DISCUSSION	Jan 17
MARTIN LUTHER KING'S BIRTHDAY – NO CLASS	-	Jan 20
2. Chromosome segregation and recombination	E. Joyce	Jan 22
	DISCUSSION	Jan 24
3. Mutagenesis and forward genetic screens	T. Jongens	Jan 27
4. Determining how mutations affect gene function	M. Sundaram	Jan 29
	DISCUSSION	Jan 31
5. Going from phenotype to gene in model organisms	M. Sundaram	Feb 03
6. Going from phenotype to gene in human families	M. Devoto	Feb 05
	DISCUSSION	Feb 07
7. Genomes and Genome Editing	O. Shalem	Feb 10
8. Transposable elements	R. Bushman	Feb 12
	DISCUSSION	Feb 14
9. RNAi and miRNAs	B. Gregory	Feb 17
	DISCUSSION	Feb 19
1ST EXAM (IN CLASS)		Feb 21

II. GENETICS OF MODEL ORGANISMS	**Lecturer**	**Date**
1. *C. elegans* genetics	M. Hart	Feb 24
2. Drosophila genetics	S. Little	Feb 26
	DISCUSSION	Feb 28
3. Mosaic analysis and conditional alleles	M. Sundaram	Mar 02
4. Maternal effect mutants in zebrafish	M. Mullins	Mar 04
	DISCUSSION	Mar 06

SPRING BREAK MAR 09-13

5. Forward genetics and genomics in the mouse	Y. Kamberov	Mar 16
6. Reverse genetics in the mouse	E. Korb	Mar 18
	DISCUSSION	Mar 20
7. Epistasis and Genetic modifiers	M. Sundaram	Mar 23
8. Quantitative traits in the mouse	E. Brodkin	Mar 25
	DISCUSSION	Mar 27

2ND EXAM (TAKE HOME MAR 27 – APR 03)

III. HUMAN GENETICS AND DISEASE

NO CLASS	-	Mar 30
1. Genome wide genetics for complex traits	S. Grant	Apr 01
	DISCUSSION	Apr 03
2. Population genetics	I. Mathieson	Apr 06
3. Forensic and personal genetics	I. Mathieson	Apr 08
	DISCUSSION	Apr 10
4. Expression QTL Analysis	S. Ramdas	Apr 13
5. Mitochondrial genetics	R. Ganetzky	Apr 15
	DISCUSSION	Apr 17
6. X chromosome inactivation	M. Bartolomei	Apr 20
7. Translational Medicine	D. Rader	Apr 22
	DISCUSSION	Apr 24
8. Cancer genetics and personalized medicine	Kristopher Bosse	Apr 27
9. Chromosome abnormalities	L. Conlin	Apr 29
	DISCUSSION	May 01

3RD EXAM (TAKE HOME MAY 01- MAY 08)

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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 and cells - 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 and Exams

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. <u>The take-home exams must be prepared independently without ANY outside consultation</u>.

Discussion guidelines

The homework problems and discussion are <u>the most important part</u> of this course. Each lecturer will assign homework problems for the week of their lecture (these will be posted on Canvas). 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)

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CAMB 550 Lecturers – 2020

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