Cell and Molecular Biology 550 "GENETIC PRINCIPLES" Spring Semester 2021
Monday, Wednesday, Friday 10-11:30 am,

Zoom links to class can be found on CANVAS

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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Teaching Assistants: Office hours: Thursday 3-5PM
(Parts I&II): Randi Isenhart isenhart@pennmedicine.upenn.edu
(Part III): Elizabeth Burton Elizabeth.Burton2@pennmedicine.upenn.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 (3 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 20
	DISCUSSION	Jan 22
2. Chromosome segregation and recombination	E. Joyce	Jan 25
3. Transposable elements	R. Bushman	Jan 27
	DISCUSSION	Jan 29
4. RNAi and miRNAs	C. Conine	Feb 01
5. CRISPR Genome Editing	O. Shalem	Feb 03
	DISCUSSION	Feb 05
6. Determining how mutations affect gene function	M. Sundaram	Feb 08
7. Going from phenotype to gene in model organisms	M. Sundaram	Feb 10
	DISCUSSION	Feb 11 (4-5PM)

1ST EXAM (TAKE HOME Feb 11-19)

II. GENETICS OF MODEL ORGANISMS

	Lecturer	Date
1. *C. elegans* genetics	M. Hart	Feb 15

2. *Drosophila* genetics	E. Joyce	Feb 17
	DISCUSSION	Feb 19
3. Maternal effect mutants in zebrafish	M. Mullins	Feb 22
4. Epistasis and Genetic modifiers	M. Sundaram	Feb 24
	DISCUSSION	Feb 26
5. Forward genetics and genomics in the mouse	Y. Kamberov	Mar 01
6. Reverse genetics in the mouse	E. Korb	Mar 03
	DISCUSSION	Mar 05

SPRING BREAK MAR 08-12

7. Ants and other emerging model organisms	R. Bonasio	Mar 17
	DISCUSSION	Mar 19

2ND EXAM (TAKE HOME MAR 19 –26)

III. HUMAN GENETICS AND DISEASE

1. Genome wide genetics for complex traits	S. Grant	Mar 22
2. X chromosome inactivation	M. Bartolomei	Mar 24
	DISCUSSION	Mar 26
3. Population genetics	I. Mathieson	Mar 29
4. Forensic and personal genetics	I. Mathieson	Mar 31
	DISCUSSION	Apr 02
5. Human mutation rates and recombination patterns	Z. Gao	Apr 05
6. Basics of quantitative genetics	Z. Gao	Apr 07
	DISCUSSION	Apr 09
NO CLASS	-	Apr 12
7. Expression QTL Analysis	C. Brown	Apr 14
	DISCUSSION	Apr 16
8. Mitochondrial genetics	R. Ganetzky	Apr 19
9. Translational Medicine	D. Rader	Apr 21
	DISCUSSION	Apr 23
10. Cancer genetics and personalized medicine	Kristopher Bosse	Apr 26
11. Chromosome abnormalities	L. Conlin	Apr 28
	DISCUSSION	Apr 30

3RD EXAM (TAKE HOME April 30 - MAY 07)

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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.

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

Discussion guidelines

The homework problems and discussion are the most important part 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)

FINAL 1-4-21

CAMB 550 Lecturers – 2021

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