Cell and Molecular Biology 550 "GENETIC PRINCIPLES" Spring Semester 2023
Monday, Wednesday, Friday 10:15-11:45 am, BRB253

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, Room BRB 1201
Bailey Warder Bailey.Warder@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)

I. GENETIC CONCEPTS AND TOOLS

	Lecturer	Date
1. Beyond Mendel	E. Joyce	Jan 11
	DISCUSSION	Jan 13
MARTIN LUTHER KING'S BIRTHDAY – NO CLASS	-	Jan 16
2. Chromosome segregation and recombination	E. Joyce	Jan 18
	DISCUSSION	Jan 20
NO CLASS	-	Jan 23
3. Mutagenesis and genetic mapping (recorded)	M. Sundaram	Jan 25
	DISCUSSION	Jan 27
4. Determining how mutations affect gene function (recorded)	M. Sundaram	Jan 30
5. CRISPR Genome Editing	O. Shalem	Feb 01
	DISCUSSION	Feb 03
6. RNAi and miRNAs	C. Conine	Feb 06
7. Jumping genes: Transposable elements	R. Bushman	Feb 08
	DISCUSSION	Feb 10

1st EXAM (TAKE HOME Feb 10 – 17)

II. GENETICS OF MODEL ORGANISMS	**Lecturer**	**Date**
1. *Drosophila* genetics	E. Joyce	Feb 20
2. *C. elegans* genetics	M. Hart	Feb 22
	DISCUSSION	Feb 24
3. Mosaicism and maternal effect mutants in zebrafish	M. Mullins	Feb 27
4. Ants, epigenetics, and emerging model systems	R. Bonasio	Mar 01
	DISCUSSION	Mar 03

SPRING BREAK MAR 04-12

5. Mouse Genomics	Y. Kamberov	Mar 13
6. Reverse genetics in the mouse	E. Korb	Mar 15
	DISCUSSION	Mar 17

2nd EXAM (TAKE HOME Mar 17 - 24)

III. HUMAN GENETICS AND DISEASE		
1. Genome wide genetic studies for human diseases	K. Wang	Mar 27
2. Sequencing for Mendelian disease diagnosis	K. Wang	Mar 29
	DISCUSSION	Mar 31
3. Population genetics	I. Mathieson	Apr 03
4. Basics of quantitative genetics	Z. Gao	Apr 05
	DISCUSSION	Apr 07
5. X chromosome inactivation	M. Bartolomei	Apr 10
6. Chromosome abnormalities	L. Conlin	Apr 12
	DISCUSSION	Apr 14
7. Mitochondrial genetics	R. Ganetzky	Apr 17
8. Cancer Genetics	M. Li	Apr 19
	DISCUSSION	Apr 21

3RD EXAM (TAKE HOME April 21 - 28)

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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 11/09/22

CAMB 550 Lecturers – 2023

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