Cell and Molecular Biology 550 "GENETIC PRINCIPLES" Spring Semester 2025
Monday, Wednesday, Friday 10:15-11:45 am, BRB251

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

Grading: 1/3 Class participation (Discussion of assigned problems)
2/3 Exams 1.5 hours in class

I. GENETIC SYSTEMS, CONCEPTS, AND TOOLS

	Lecturer	Date
1. Introduction to Genetics Principles	E. Joyce/Z. Gao	Jan 15
	DISCUSSION	Jan 17
2. Chromosome segregation and recombination	E. Joyce	Jan 22
	DISCUSSION	Jan 24
3. Mutagenesis and genetic mapping	M. Sundaram	Jan 27
4. Determining how mutations affect gene function	M. Sundaram	Jan 29
	DISCUSSION	Jan 31
5. Jumping genes: Transposable elements	A. Modzelewski	Feb 03
6. X-linked disease	M. Anguera	Feb 05
	DISCUSSION	Feb 07
7. RNAi and miRNAs	C. Conine	Feb 10
8. Ants, epigenetics, and emerging model systems	R. Bonasio	Feb 12
	DISCUSSION	Feb 14

9. Forward Genetics in *Drosophila*	E. Joyce	Feb 17
10. Reverse Genetics in *C. elegans*	M. Hart	Feb 19
	DISCUSSION	Feb 21
11. Mosaic analysis and conditional alleles	E. Joyce	Feb 24
12. Mouse genetic tools	E. Korb	Feb 26
	DISCUSSION	Feb 28

MIDTERM EXAM (in-class)---------- Mar 7

II. HUMAN GENETICS AND DISEASE

1. Human Genome and genetic variation	G. Logsdon	Mar 12
	DISCUSSION	Mar 14
2. Population genetics	I. Mathieson	Mar 17
3. Human evolution	I. Mathieson	Mar 19
	DISCUSSION	Mar 21
4. Quantitative genetics	Z. Gao	Mar 24
5. Genome-wide association studies	S. Grant	Mar 26
	DISCUSSION	Mar 28
6. Sequencing for Mendelian disease diagnosis	T. Drivas	Mar 31
7. Precision genetic therapies	K. Musunuru	Apr 02
	DISCUSSION	Apr 04
8. Cancer Genetics	D. Silverbush	Apr 07
9. Chromosome abnormalities	L. Conlin	Apr 09
	DISCUSSION	Apr 11
10. Complex traits genetics	Z. Gao	Apr 14
11. Biobank-based research	A. Verma	Apr 16
	DISCUSSION	Apr 18

FINAL EXAM (in-class) ---------- April 25

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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 two exams (100 points each) and Discussion participation (100 points), for a possible total of 300 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.

Both exams will be held in class to be completed within 1.5 hours. The exams will cover basic genetic concepts and will test your ability to design and interpret genetic experiments.

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 assigned specific weeks to lead the Discussion and answer questions.

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 – 2023

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