

Cell and Molecular Biology 5500 “GENETIC PRINCIPLES” Spring Semester 2026
Monday, Wednesday, Friday 10:15-11:45 am, BRB251

This is a combined lecture and discussion course that surveys major concepts and approaches in model organism and human genetics. Discussions are problem-based and emphasize practical aspects of generating, analyzing, and interpreting genetic data.

Course Directors: Eric Joyce, 564 CRB, 898-1229, erjoyce@upenn.edu
 Ziyue Gao, 411B CRB, ziyuegao@pennmedicine.upenn.edu

Teaching Assistants: Atharva Dhamale Atharva.Dhamale@Pennmedicine.upenn.edu
 Timothy Cater trcater@pennmedicine.upenn.edu
 Office hours: Thursday TBD

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

Grading: 1/3 Class participation
 2/3 Exams (2 take-home exams)

I. GENETIC CONCEPTS, TOOLS, and SYSTEMS

	<u>Lecturer</u>	<u>Date</u>
1. Introduction to Genetics Principles	E. Joyce	Jan 14
	DISCUSSION	Jan 16
2. Chromosome segregation and recombination	E. Joyce	Jan 21
	DISCUSSION	Jan 23
3. Mutagenesis and genetic mapping	M. Sundaram	Jan 26
4. Determining how mutations affect gene function	M. Sundaram	Jan 28
	DISCUSSION	Jan 30
5. RNAi and miRNAs	C. Conine	Feb 02
6. CRISPR screening	O. Shalem	Feb 04
	DISCUSSION	Feb 06
7. X-inactivation and imprinting	M. Anguera	Feb 09
8. Ants, epigenetics, and emerging model systems	R. Bonasio	Feb 11
	DISCUSSION	Feb 13
9. Forward Genetics in <i>Drosophila</i>	E. Joyce	Feb 16
10. Reverse Genetics in <i>C. elegans</i>	M. Hart	Feb 18
	DISCUSSION	Feb 20
11. Epigenetics Institute Symposium	-----	Feb 23
12. Mouse genetic tools	Y. Kamberov	Feb 25
	DISCUSSION	Feb 27

MIDTERM EXAM (Take-home)-----Feb 27-Mar 06

SPRING BREAK-----Mar 07-Mar 15

II. HUMAN GENETICS AND DISEASE

1. Human genome and genetic variation	G. Logsdon	Mar 16
2. Selfish genetic elements	M. Levine	Mar 18
	DISCUSSION	Mar 20
3. Chromosome abnormalities	L. Conlin	Mar 23
	DISCUSSION	Mar 27
4. Population genetics I	I. Mathieson	Mar 30
5. Population genetics II	I. Mathieson	Apr 1
	DISCUSSION	Apr 3
6. Functional genomes or functional annotation	K. Wang	Apr 06
7. Sequencing for Mendelian disease diagnosis	T. Drivas	Apr 08
	DISCUSSION	Apr 10
8. Quantitative genetics	Z. Gao	Apr 13
9. Genome-wide association studies	S. Grant	Apr 15
	DISCUSSION	Apr 17
10. Complex traits genetics	Z. Gao	Apr 20
11. Genetic approaches to causal inference	Z. Gao	Apr 22
	DISCUSSION	Apr 24
12. Cancer genetics	D. Silverbush	Apr 27
	DISCUSSION	May 1

FINAL EXAM (Take-home) -----April 25-May 1

Cell and Molecular Biology 5500 “GENETIC PRINCIPLES” Spring Semester 2026

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

The final grade will be a composite of two exams (33 points each) and participation (34 points), for a total of 100 points. Letter grades will be assigned on a curve relative to the class mean:

- Scores above the mean: A range (A+, A, A-)
- Scores below the mean: B range (B+, B, B-)
- Scores more than two standard deviations below the mean: C or below.

Historically, the mean score has been 75-80, with a standard deviation of 10-15.

Participation grades

There are 23 lectures and 14 discussion sessions. Students may earn up to 34 participation points based on attendance and engagement. For lectures, one point will be awarded for each session attended. For discussion sessions, active participation (e.g., serving as a group representative, asking and answering questions) is expected to receive full credit. Please sign the attendance sheet at each lecture and discussion session to ensure your attendance is recorded. A maximum of 34 points across the 37 scheduled sessions will go toward your final grade.

Discussion guidelines and participation

The problem sets and discussion are central to learning 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 problems before the Friday discussion and may consult TAs during office hours. It is fine to work collaboratively in a “study group”. Problem sets will NOT be collected.

Each discussion session will last for 1.5 hours, covering the content of one or two lectures in the same week. At the beginning of each discussion session, students will be divided into groups and assigned specific questions to discuss among themselves for 10-15 minutes. Then, each group will send representatives to present their answer to the whole class.

Exams

Both exams will be take-home (open book) and must be prepared independently without outside consultation. The exams will cover core genetic concepts and will test your ability to design and interpret genetic experiments and analyses. Use of internet and AI is allowed, but students must demonstrate understanding of the solution process.

CAMB 5500 Lecturers – 2026

Montserrat Anguera

Dept. of Biomedical Sciences
anguera@vet.upenn.edu

Roberto Bonasio

Dept. of Cell & Dev Biology
roberto@bonasiolab.org

Colin Conine

Dept. of Genetics
conine@upenn.edu

Laura Conlin

CHOP, Division of Genomic Diagnostics
conlinl@email.chop.edu

Theodore Drivas

Dept of Medicine
theodore.drivas@pennmedicine.upenn.edu

Struan Grant

Dept of Genetics
CHOP, Divisions of Human Genetics and Endocrinology
grants@pennmedicine.upenn.edu

Ziyue Gao

Dept of Genetics
ziyuegao@pennmedicine.upenn.edu

Mike Hart

Dept of Genetics
hartmic@pennmedicine.upenn.edu

Eric Joyce

Dept of Genetics
erjoyce@upenn.edu

Yana Kamberov

Dept of Genetics
yana2@pennmedicine.upenn.edu

Erica Korb

Dept of Genetics
ekorb@pennmedicine.upenn.edu

Glennis Logsdon

Dept of Genetics
glogsdon@pennmedicine.upenn.edu

Iain Mathieson

Dept of Genetics
mathi@upenn.edu

Dana Silverbush

Dept of Cancer Biology
dana.silverbush@pennmedicine.upenn.edu

Ophir Shalem

CHOP Center for Cellular and Molecular Therapeutics
& Dept. of Genetics
shalemo@pennmedicine.upenn.edu

Meera Sundaram

Dept of Genetics
446A CRB, 3-4527
sundaram@pennmedicine.upenn.edu