Cell and Molecular Biology 550 "GENETIC PRINCIPLES" Spring Semester 2024
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.

<u>Course Directors:</u>	Eric Joyce, 564 CRB, 898-1229, erjoyce@upenn.edu
	Struan Grant, 1102D ARC, 215-200-0196, grants@email.chop.edu
<u>Teaching Assistants:</u>	Office hours: Thursday TBD
	Sections 1&2: May Wai maywai@pennmedicine.upenn.edu
	Section 3: Winter Bruner brunerw@chop.edu
<u>Format:</u>	Monday and Wednesday, 1 - 1.5-hour lectures
	Friday, 1.5 hour discussion of assigned problem sets
<u>Grading:</u>	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 22
2. Chromosome segregation and recombination	E. Joyce	Jan 24
	DISCUSSION	Jan 26
3. Mutagenesis and genetic mapping	M. Sundaram	Jan 29
4. Determining how mutations affect gene function	M. Sundaram	Jan 31
	DISCUSSION	Feb 02
5. CRISPR Genome Editing	O. Shalem	Feb 05
6. RNAi and miRNAs	C. Conine	Feb 07
	DISCUSSION	Feb 09
7. Jumping genes: Transposable elements	A. Modzelewski	Feb 12
	DISCUSSION	Feb 14

1st EXAM (TAKE HOME Feb 16 – 23)

II. GENETICS OF MODEL ORGANISMS

	Lecturer	Date
1. Ants, epigenetics, and emerging model systems	R. Bonasio	Feb 26
2. *Drosophila* genetics	E. Joyce/B. Warder	Feb 28
	DISCUSSION	Mar 01
3. *C. elegans* genetics	M. Hart	Mar 04
4. Mosaic analysis and conditional alleles	E. Joyce	Mar 06
	DISCUSSION	Mar 08
5. Mouse Genomics	Y. Kamberov	Mar 11
6. Reverse genetics in the mouse	E. Korb	Mar 13
	DISCUSSION	Mar 15
7. Maternal effect mutants in zebrafish	M. Mullins	Mar 18
	DISCUSSION	Mar 20

2nd EXAM (TAKE HOME Mar 22 - 29)

III. HUMAN GENETICS AND DISEASE

	Lecturer	Date
1. Genome wide genetic studies for human diseases	S. Grant	Apr 01
2. Sequencing for Mendelian disease diagnosis	K. Wang	Apr 03
	DISCUSSION	Apr 05
3. Population genetics	I. Mathieson	Apr 08
4. Basics of quantitative genetics	I. Mathieson	Apr 10
	DISCUSSION	Apr 12
5. Chromosome abnormalities	L. Conlin	Apr 15
6. X chromosome inactivation	M. Bartolomei	Apr 17
	DISCUSSION	Apr 19
7. Mitochondrial genetics	R. Ganetzky	Apr 22
8. Cancer Genetics	M. Li	Apr 24
	DISCUSSION	Apr 26

3RD EXAM (TAKE HOME April 26 – May 03)

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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 <u>must be prepared independently without ANY outside consultation</u>. 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 <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)

CAMB 550 Lecturers – 2023

Marisa Bartolomei
Dept. of Cell & Dev Biology
bartolom@pennmedicine.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

Rebecca Ganetzky
CHOP Division of Human Genetics
ganetzkyr@email.chop.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

Marilyn Li
CHOP, Division of Genomic Diagnostics
& Dept. of Pathology
lim5@chop.edu

Iain Mathieson
Dept of Genetics
mathi@upenn.edu

Andrew Modzelewski
Vet Med
amodz@vet.upenn.edu

Mary Mullins
Dept. of Cell and Developmental Biology
mullins@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

Kai Wang
CHOP, Center for Cellular and Molecular Therapeutics
& Department of Pathology
wangk@chop.edu