Cell and Molecular Biology 550 "GENETIC PRINCIPLES" Spring Semester 2022
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
Kai Wang, 5004 CTRB, 267-425-9573, wangk@chop.edu

Teaching Assistants: Office hours: Thursday 3-5PM
(Parts I&II): Bailey Warder Bailey.Warder@pennmedicine.upenn.edu
(Part III): James Havrilla havrillaj@chop.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 (1 in-class, 2 take-home exams)

I. GENETIC CONCEPTS AND TOOLS

	Lecturer	Date
1. Beyond Mendel	E. Joyce	Jan 12
	DISCUSSION	Jan 14
MARTIN LUTHER KING'S BIRTHDAY – NO CLASS	-	Jan 17
2. Chromosome segregation and recombination	E. Joyce	Jan 19
	DISCUSSION	Jan 21
3. Determining how mutations affect gene function	M. Sundaram	Jan 24
4. Going from phenotype to gene in model systems	M. Sundaram	Jan 26
	DISCUSSION	Jan 28
5. RNAi and miRNAs	C. Conine	Jan 31
6. CRISPR Genome Editing	O. Shalem	Feb 02
	DISCUSSION	Feb 04
7. Jumping genes: Transposable elements	R. Bushman	Feb 07
	DISCUSSION	Feb 09
1ST EXAM (in class, 1.5 hours)		Feb 11

II. GENETICS OF MODEL ORGANISMS	**Lecturer**	**Date**
1. *Drosophila* genetics	E. Joyce	Feb 14
2. Mosaic analysis and conditional alleles	E. Joyce	Feb 16
	DISCUSSION	Feb 18
3. *C. elegans* genetics	M. Hart	Feb 21
4. Maternal effect mutants in zebrafish	M. Mullins	Feb 23
	DISCUSSION	Feb 25
5. Forward genetics and genomics in the mouse	Y. Kamberov	Feb 28
6. Reverse genetics in the mouse	E. Korb	Mar 02
	DISCUSSION	Mar 04

SPRING BREAK MAR 05-13

7. Epistasis and Genetic modifiers	M. Sundaram	Mar 14
8. Ants, epigenetics, and emerging model systems	R. Bonasio	Mar 16
	DISCUSSION	Mar 18

2ND EXAM (TAKE HOME MAR 18 –25)

III. HUMAN GENETICS AND DISEASE

NO CLASS	-	Mar 21
1. Genome wide genetic studies for human diseases	K. Wang	Mar 23
	DISCUSSION	Mar 25
2. Population genetics	I. Mathieson	Mar 28
3. Basics of quantitative genetics	Z. Gao	Mar 30
	DISCUSSION	Apr 01
4. Expression QTL Analysis	C. Brown	Apr 04
5. Chromosome abnormalities	L. Conlin	Apr 06
	DISCUSSION	Apr 08
6. X chromosome inactivation	M. Bartolomei	Apr 11
7. Cancer Genetics	M. Li	Apr 13
	DISCUSSION	Apr 15
8. Translational & personalized medicine	D. Rader	Apr 18
9. Mitochondrial genetics	R. Ganetzky	Apr 20
	DISCUSSION	Apr 22

3RD EXAM (TAKE HOME April 22 - April 29)

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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.

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. These two exams will be take-home (open book) and must be prepared independently without ANY outside consultation.

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)

CAMB 550 Lecturers – 2022

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

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

Casey Brown
Dept. of Genetics
casey6r0wn@gmail.com

Rick Bushman
Dept. of Microbiology
bushman@pennmedicine.upenn.edu

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

Mary Mullins
Dept. of Cell and Developmental Biology
mullins@pennmedicine.upenn.edu

Dan Rader
Dept. of Genetics
rader@pennmedicine.upenn.edu

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

Meera Sundaram
Dept of Genetics
sundaram@pennmedicine.upenn.edu

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

TAs:

Bailey Warder, G&E student
Bailey.Warder@pennmedicine.upenn.edu

James Havrilla, Postdoc
havrillaj@chop.edu