## 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.

Course Directors: Eric Joyce, 564 CRB, 898-1229, [erjoyce@upenn.edu](mailto:erjoyce@upenn.edu)

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Teaching Assistants: Office hours: Thursday TBD

Sections 1&2: May Wai [maywai@pennmedicine.upenn.edu](mailto:maywai@pennmedicine.upenn.edu)

Section 3: Winter Bruner [brunerw@chop.edu](mailto:brunerw@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 (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

DISCUSSIONMar 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**

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 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)

CAMB 550 Lecturers – 2023

**Marisa Bartolomei**

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**Mary Mullins**

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