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, 6004 CTRB, 267-425-9573, wangk@chop.edu

**Teaching Assistants:** Office hours: Thursday 3-5PM, Room BRB 1201  
Bailey Warder Bailey.Warder@pennmedicine.upenn.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

<table>
<thead>
<tr>
<th>Topic</th>
<th>Lecturer</th>
<th>Date</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Beyond Mendel</td>
<td>E. Joyce</td>
<td>Jan 11</td>
</tr>
<tr>
<td>DISCUSSION</td>
<td></td>
<td>Jan 13</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>MARTIN LUTHER KING’S BIRTHDAY – NO CLASS</td>
<td></td>
<td>Jan 16</td>
</tr>
<tr>
<td>2. Chromosome segregation and recombination</td>
<td>E. Joyce</td>
<td>Jan 18</td>
</tr>
<tr>
<td>DISCUSSION</td>
<td></td>
<td>Jan 20</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>NO CLASS</td>
<td></td>
<td>Jan 23</td>
</tr>
<tr>
<td>3. Mutagenesis and genetic mapping (recorded)</td>
<td>M. Sundaram</td>
<td>Jan 25</td>
</tr>
<tr>
<td>DISCUSSION</td>
<td></td>
<td>Jan 27</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4. Determining how mutations affect gene function (recorded)</td>
<td>M. Sundaram</td>
<td>Jan 30</td>
</tr>
<tr>
<td>5. CRISPR Genome Editing</td>
<td>O. Shalem</td>
<td>Feb 01</td>
</tr>
<tr>
<td>DISCUSSION</td>
<td></td>
<td>Feb 03</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>6. RNAi and miRNAs</td>
<td>C. Conine</td>
<td>Feb 06</td>
</tr>
<tr>
<td>7. Jumping genes: Transposable elements</td>
<td>R. Bushman</td>
<td>Feb 08</td>
</tr>
<tr>
<td>DISCUSSION</td>
<td></td>
<td>Feb 10</td>
</tr>
</tbody>
</table>

**1st EXAM (TAKE HOME Feb 10 – 17)**
II. GENETICS OF MODEL ORGANISMS

1. *C. elegans* genetics
   Lecturer: M. Hart
   Date: Feb 20

2. *Drosophila* genetics
   Lecturer: E. Joyce
   Date: Feb 22

3. Mosaicism and maternal effect mutants in zebrafish
   Lecturer: M. Mullins
   Date: Feb 27

4. Ants, epigenetics, and emerging model systems
   Lecturer: R. Bonasio
   Date: Mar 01

DISCUSSION
Feb 24

SPRING BREAK MAR 04-12

5. Mouse Genomics
   Lecturer: Y. Kamberov
   Date: Mar 13

6. Reverse genetics in the mouse
   Lecturer: E. Korb
   Date: Mar 15

DISCUSSION
Mar 17

2nd EXAM (TAKE HOME Mar 17 - 24)

III. HUMAN GENETICS AND DISEASE

1. Genome wide genetic studies for human diseases
   Lecturer: K. Wang
   Date: Mar 27

2. Sequencing for Mendelian disease diagnosis
   Lecturer: K. Wang
   Date: Mar 29

   DISCUSSION
   Mar 31

3. Population genetics
   Lecturer: I. Mathieson
   Date: Apr 03

4. Basics of quantitative genetics
   Lecturer: Z. Gao
   Date: Apr 05

   DISCUSSION
   Apr 07

5. X chromosome inactivation
   Lecturer: M. Bartolomei
   Date: Apr 10

6. Chromosome abnormalities
   Lecturer: L. Conlin
   Date: Apr 12

   DISCUSSION
   Apr 14

7. Mitochondrial genetics
   Lecturer: R. Ganetzky
   Date: Apr 17

8. Cancer Genetics
   Lecturer: M. Li
   Date: Apr 19

   DISCUSSION
   Apr 21

3RD EXAM (TAKE HOME April 21 - 28)
Cell and Molecular Biology 550  “GENETIC PRINCIPLES” Spring Semester 2023

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

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

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

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