Cell and Molecular Biology 550  “GENETIC PRINCIPLES” Spring Semester 2020
Monday, Wednesday, Friday 10-11:30 am, Room 251 BRB

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.

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**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 exam and 2 take-home exams)

**Supplementary textbooks available online:** Griffiths et al. “Introduction to Genetic Analysis”
Strachan and Read, “Human Molecular Genetics”

### I. GENETIC CONCEPTS AND TOOLS

<table>
<thead>
<tr>
<th>Lecturer</th>
<th>Date</th>
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</thead>
<tbody>
<tr>
<td><strong>1. Beyond Mendel</strong></td>
<td></td>
</tr>
<tr>
<td>E. Joyce</td>
<td>Jan 15</td>
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<tr>
<td>DISCUSSION</td>
<td>Jan 17</td>
</tr>
</tbody>
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MARTIN LUTHER KING’S BIRTHDAY – NO CLASS

| **2. Chromosome segregation and recombination** |        |
| E. Joyce       | Jan 22 |
| DISCUSSION     | Jan 24 |

| **3. Mutagenesis and forward genetic screens** |        |
| T. Jongens     | Jan 27 |

| **4. Determining how mutations affect gene function** |        |
| M. Sundaram    | Jan 29 |
| DISCUSSION     | Jan 31 |

| **5. Going from phenotype to gene in model organisms** |        |
| M. Sundaram    | Feb 03 |

| **6. Going from phenotype to gene in human families** |        |
| M. Devoto      | Feb 05 |
| DISCUSSION     | Feb 07 |

| **7. Genomes and Genome Editing** |        |
| O. Shalem       | Feb 10 |

| **8. Transposable elements** |        |
| R. Bushman       | Feb 12 |
| DISCUSSION       | Feb 14 |

| **9. RNAi and miRNAs** |        |
| B. Gregory       | Feb 17 |
| DISCUSSION       | Feb 19 |

**1ST EXAM (IN CLASS)**

Feb 21
## II. GENETICS OF MODEL ORGANISMS

<table>
<thead>
<tr>
<th>Topic</th>
<th>Lecturer</th>
<th>Date</th>
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</thead>
<tbody>
<tr>
<td>1. <em>C. elegans</em> genetics</td>
<td>M. Hart</td>
<td>Feb 24</td>
</tr>
<tr>
<td>2. Drosophila genetics</td>
<td>S. Little</td>
<td>Feb 26</td>
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<tr>
<td>DISCUSSION</td>
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<td>Feb 28</td>
</tr>
<tr>
<td>3. Mosaic analysis and conditional alleles</td>
<td>M. Sundaram</td>
<td>Mar 02</td>
</tr>
<tr>
<td>4. Maternal effect mutants in zebrafish</td>
<td>M. Mullins</td>
<td>Mar 04</td>
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<tr>
<td>DISCUSSION</td>
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<td>Mar 06</td>
</tr>
</tbody>
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### SPRING BREAK MAR 09-13

5. Forward genetics and genomics in the mouse                        | Y. Kamberov     | Mar 16  |
6. Reverse genetics in the mouse                                     | E. Korb         | Mar 18  |
| DISCUSSION                                                          |                 | Mar 20  |

7. Epistasis and Genetic modifiers                                    | M. Sundaram     | Mar 23  |
8. Quantitative traits in the mouse                                   | E. Brodkin      | Mar 25  |
| DISCUSSION                                                          |                 | Mar 27  |

### 2ND EXAM (TAKE HOME MAR 27 – APR 03)

### III. HUMAN GENETICS AND DISEASE

<table>
<thead>
<tr>
<th>Topic</th>
<th>Lecturer</th>
<th>Date</th>
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</thead>
<tbody>
<tr>
<td>NO CLASS</td>
<td>-</td>
<td>Mar 30</td>
</tr>
<tr>
<td>1. Genome wide genetics for complex traits</td>
<td>S. Grant</td>
<td>Apr 01</td>
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<tr>
<td>DISCUSSION</td>
<td></td>
<td>Apr 03</td>
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<tr>
<td>2. Population genetics</td>
<td>I. Mathieson</td>
<td>Apr 06</td>
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<tr>
<td>3. Forensic and personal genetics</td>
<td>I. Mathieson</td>
<td>Apr 08</td>
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<tr>
<td>DISCUSSION</td>
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<td>Apr 10</td>
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<td>4. Expression QTL Analysis</td>
<td>S. Ramdas</td>
<td>Apr 13</td>
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<tr>
<td>5. Mitochondrial genetics</td>
<td>R. Ganetzky</td>
<td>Apr 15</td>
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<tr>
<td>DISCUSSION</td>
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<td>Apr 17</td>
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<tr>
<td>6. X chromosome inactivation</td>
<td>M. Bartolomei</td>
<td>Apr 20</td>
</tr>
<tr>
<td>7. Translational Medicine</td>
<td>D. Rader</td>
<td>Apr 22</td>
</tr>
<tr>
<td>DISCUSSION</td>
<td></td>
<td>Apr 24</td>
</tr>
<tr>
<td>8. Cancer genetics and personalized medicine</td>
<td>Kristopher Bosse</td>
<td>Apr 27</td>
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<tr>
<td>9. Chromosome abnormalities</td>
<td>L. Conlin</td>
<td>Apr 29</td>
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<tr>
<td>DISCUSSION</td>
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<td>May 01</td>
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### 3RD EXAM (TAKE HOME MAY 01- MAY 08)
Cell and Molecular Biology 550  “GENETIC PRINCIPLES” Spring Semester 2020

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 will be in-class (closed book) and covers basic genetic concepts that are the foundation for the rest of the course. The second and third exam will be in take-home (open book) format; these exams will test your ability to design and interpret genetic experiments. The take-home exams 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 – 2020

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