Cell and Molecular Biology 550  “GENETIC PRINCIPLES” Spring Semester 2015  
Monday, Wednesday, Friday 10-11:30 am, 253 BRBII/III

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:**  
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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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<tbody>
<tr>
<td>M. Sundaram</td>
<td>Jan 14</td>
</tr>
<tr>
<td>DISCUSSION</td>
<td>Jan 16</td>
</tr>
<tr>
<td>T. Jongens</td>
<td>Jan 23</td>
</tr>
<tr>
<td>M. Sundaram</td>
<td>Jan 28</td>
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<tr>
<td>DISCUSSION</td>
<td>Jan 30</td>
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<td>M. Devoto</td>
<td>Feb 04</td>
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<td>DISCUSSION</td>
<td>Feb 06</td>
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<td>D. Epstein</td>
<td>Feb 09</td>
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<td>B. Gregory</td>
<td>Feb 11</td>
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<td>DISCUSSION</td>
<td>Feb 13</td>
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<td>R. Bushman</td>
<td>Feb 16</td>
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<tr>
<td>DISCUSSION</td>
<td>Feb 18</td>
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1. Beyond Mendel
2. Chromosome segregation and recombination
3. Mutagenesis and forward genetic screens
4. Determining how mutations affect gene function
5. Going from phenotype to gene in model organisms
6. Linkage mapping in human pedigrees
7. Genomes and Genome Editing
8. RNAi and miRNAs
9. Transposable elements

1ST EXAM (IN CLASS)
II. GENETICS OF MODEL ORGANISMS

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<thead>
<tr>
<th></th>
<th>Lecturer</th>
<th>Date</th>
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<tbody>
<tr>
<td>1.</td>
<td><em>C. elegans</em></td>
<td>D. Raizen Feb 23</td>
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<td>2.</td>
<td>Drosophila</td>
<td>A. Ghabrial Feb 25</td>
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<td>DISCUSSION</td>
<td>Feb 27</td>
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<td>3.</td>
<td>Mosaic analysis</td>
<td>M. Sundaram Mar 02</td>
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<td>4.</td>
<td>Maternal effect and sterile mutants</td>
<td>T. Jongens Mar 04</td>
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<td>DISCUSSION</td>
<td>Mar 06</td>
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SPRING BREAK (REVIEW SESSIONS AS WARRANTED) Mar 09-13

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<tr>
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<th>Lecturer</th>
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<tr>
<td>5.</td>
<td>Epistasis and genetic modifiers</td>
<td>M. Sundaram Mar 16</td>
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<td>6.</td>
<td>Mouse knockouts and transgenics</td>
<td>K. Kaestner Mar 18</td>
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<tr>
<td></td>
<td>DISCUSSION</td>
<td>Mar 20</td>
</tr>
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<td>7.</td>
<td>Forward genetics and genomics in the mouse</td>
<td>M. Bucan Mar 23</td>
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<td>8.</td>
<td>Quantitative traits in the mouse</td>
<td>E. Brodkin Mar 25</td>
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<td>DISCUSSION</td>
<td>Mar 27</td>
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2ND EXAM (TAKE HOME MAR 27-APR 06), NO CLASS SESSIONS WEEK OF MAR 30

III. HUMAN GENETICS AND DISEASE

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<thead>
<tr>
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<th>Lecturer</th>
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<tbody>
<tr>
<td>1.</td>
<td>Chromosomal abnormalities</td>
<td>N. Spinner Apr 06*</td>
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<td>2.</td>
<td>X inactivation</td>
<td>M. Bartolomei Apr 08</td>
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<td>DISCUSSION</td>
<td>Apr 10</td>
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<td>3.</td>
<td>Population genetics</td>
<td>Sarah Tishkoff Apr 13</td>
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<td>4.</td>
<td>Human evolution</td>
<td>Sarah Tishkoff Apr 15</td>
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<tr>
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<td>DISCUSSION</td>
<td>Apr 17</td>
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<td>5.</td>
<td>Family-based analyses and exome sequencing</td>
<td>S. Grant Apr 20</td>
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<td>6.</td>
<td>Genome wide genetics for complex traits</td>
<td>S. Grant Apr 22</td>
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<td>DISCUSSION</td>
<td>Apr 24</td>
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<td>7.</td>
<td>Expression QTL Analysis</td>
<td>C. Brown Apr 27</td>
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<td>8.</td>
<td>Mitochondrial genetics</td>
<td>M. Falk Apr 29</td>
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<td>DISCUSSION</td>
<td>May 01</td>
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<td>9.</td>
<td>Cancer genetics and personalized medicine</td>
<td>A. Ganguly May 04</td>
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<td>10.</td>
<td>Translational Medicine</td>
<td>D. Rader May 06</td>
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<tr>
<td></td>
<td>DISCUSSION</td>
<td>May 08</td>
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3RD EXAM (TAKE HOME MAY 01- MAY 08)

*All students should also attend 4pm Cohen lecture by Jeannie Lee
Cell and Molecular Biology 550  “GENETIC PRINCIPLES” Spring Semester 2015

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 provide assigned homework problems on the day of their lecture. 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 – 2015

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