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: Meera Sundaram, 446a CRB, 573-4527, sundaram@mail.med.upenn.edu
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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>
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</tbody>
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1. Beyond Mendel

MARTIN LUTHER KING’S BIRTHDAY – NO CLASS

2. Chromosome segregation and recombination
   M. Sundaram Jan 21
   DISCUSSION Jan 23

3. Mutagenesis and forward genetic screens
   T. Jongens  Jan 26

4. Determining how mutations affect gene function
   M. Sundaram Jan 28
   DISCUSSION Jan 30

5. Going from phenotype to gene in model organisms
   M. Sundaram Feb 02

6. Linkage mapping in human pedigrees
   M. Devoto  Feb 04
   DISCUSSION Feb 06

7. Genomes and Genome Editing
   D. Epstein  Feb 09

8. RNAi and miRNAs
   B. Gregory  Feb 11
   DISCUSSION Feb 13

9. Transposable elements
   R. Bushman  Feb 16
   DISCUSSION Feb 18

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

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

| 5. Epistasis and genetic modifiers                                   | M. Sundaram         | Mar 16 |
| 6. Mouse knockouts and transgenics                                   | K. Kaestner         | Mar 18 |
| DISCUSSION                                                          |                     | Mar 20 |
| 7. Forward genetics and genomics in the mouse                        | M. Bucan            | Mar 23 |
| 8. Quantitative traits in the mouse                                  | E. Brodkin          | Mar 25 |
| DISCUSSION                                                          |                     | Mar 27 |

**2ND EXAM (TAKE HOME MAR 27-APR 06), NO CLASS SESSIONS WEEK OF MAR 30**

# III. HUMAN GENETICS AND DISEASE

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