**Cell and Molecular Biology 550  “GENETIC PRINCIPLES” Spring Semester 2017**  
*Monday, Wednesday, Friday 10-11:30 am, Room 251 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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**Teaching Assistant:** Suzi Shapira, 12-180 Smilow, 215-746-0551, sshapira@mail.med.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 (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

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<th>Lecturer</th>
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<tr>
<td>M. Sundaram</td>
<td>Jan 11</td>
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<td>DISCUSSION</td>
<td>Jan 13</td>
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**MARTIN LUTHER KING’S BIRTHDAY – NO CLASS**

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2. Chromosome segregation and recombination

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<tr>
<td>E. Joyce</td>
<td>Jan 18</td>
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<td>DISCUSSION</td>
<td>Jan 20</td>
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3. Mutagenesis and forward genetic screens

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<td>T. Jongens</td>
<td>Jan 23</td>
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4. Determining how mutations affect gene function

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<td>M. Sundaram</td>
<td>Jan 25</td>
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<td>DISCUSSION</td>
<td>Jan 27</td>
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5. Going from phenotype to gene in model organisms

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<td>M. Sundaram</td>
<td>Jan 30</td>
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6. Linkage mapping in human pedigrees

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<td>M. Devoto</td>
<td>Feb 01</td>
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<td>DISCUSSION</td>
<td>Feb 03</td>
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7. Genomes and Genome Editing

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<td>D. Epstein</td>
<td>Feb 06</td>
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8. RNAi and miRNAs

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<td>B. Gregory</td>
<td>Feb 08</td>
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<tr>
<td>DISCUSSION</td>
<td>Feb 10</td>
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9. Transposable elements

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<td>R. Bushman</td>
<td>Feb 13</td>
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<td>DISCUSSION</td>
<td>Feb 15</td>
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**1ST EXAM (IN CLASS)**

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II. GENETICS OF MODEL ORGANISMS

1. *C. elegans* genetics  
   Lecturer: D. Raizen  
   Date: Feb 20

2. Drosophila genetics  
   Lecturer: Rachel Monyak  
   Date: Feb 22

3. Mosaic analysis  
   Lecturer: M. Sundaram  
   Date: Feb 27

4. Maternal effect mutants in zebrafish  
   Lecturer: M. Mullins  
   Date: Mar 01

DISCUSSION  
Mar 03

SPRING BREAK  (REVIEW SESSIONS AS WARRANTED)  
Mar 04-12

5. Forward genetics and genomics in the mouse  
   Lecturer: M. Bucan  
   Date: Mar 13

6. Reverse genetics in the mouse  
   Lecturer: Maria Golson  
   Date: Mar 15

DISCUSSION  
Mar 17

7. Epistasis and Genetic modifiers  
   Lecturer: M. Sundaram  
   Date: Mar 20

8. Quantitative traits in the mouse  
   Lecturer: E. Brodkin  
   Date: Mar 22

DISCUSSION  
Mar 24

2ND EXAM (TAKE HOME MAR 24-MAR 31)

III. HUMAN GENETICS AND DISEASE

1. Family-based analyses and exome sequencing  
   Lecturer: S. Grant  
   Date: Mar 27

2. Genome wide genetics for complex traits  
   Lecturer: S. Grant  
   Date: Mar 29

DISCUSSION  
Mar 31

3. Population genetics  
   Lecturer: S. Tishkoff  
   Date: Apr 03

4. Human evolution  
   Lecturer: S. Tishkoff  
   Date: Apr 05

DISCUSSION  
Apr 07

5. Chromosome abnormalities  
   Lecturer: L. Conlin  
   Date: Apr 10

6. Expression QTL Analysis  
   Lecturer: C. Brown  
   Date: Apr 12

DISCUSSION  
Apr 14

7. Mitochondrial genetics  
   Lecturer: M. Falk  
   Date: Apr 17

8. X-inactivation  
   Lecturer: M. Bartolomei  
   Date: Apr 19

DISCUSSION  
Apr 21

9. Cancer genetics and personalized medicine  
   Lecturer: A. Ganguly  
   Date: Apr 24

10. Translational Medicine  
    Lecturer: K. Musunuru  
    Date: Apr 26

DISCUSSION  
Apr 28

3RD EXAM (TAKE HOME APR 28- MAY 05)
Cell and Molecular Biology 550 “GENETIC PRINCIPLES” Spring Semester 2017

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 – 2017

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