Cell and Molecular Biology 550  “GENETIC PRINCIPLES” Spring Semester 2019
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.

Course Directors:  Meera Sundaram, 446a CRB, 573-4527, sundaram@pennmedicine.upenn.edu
Struan Grant, 1102D ARC, 215-200-0196, grants@email.chop.edu

Teaching Assistants:  Jennifer Cohen (Parts I&II): jencohen@pennmedicine.upenn.edu
Diana Cousminer (Part III): cousminerd@email.chop.edu
Office hours: Thursdays 3-5pm, Room 300 CRB

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

1. Beyond Mendel
   M. Sundaram  Jan 16
   DISCUSSION  Jan 18

   MARTIN LUTHER KING’S BIRTHDAY – NO CLASS

2. Chromosome segregation and recombination
   E. Joyce  Jan 23
   DISCUSSION  Jan 25

3. Mutagenesis and forward genetic screens
   T. Jongens  Jan 28

4. Determining how mutations affect gene function
   M. Sundaram  Jan 30
   DISCUSSION  Feb 01

5. Going from phenotype to gene in model organisms
   M. Sundaram  Feb 04

6. Going from phenotype to gene in human families
   M. Devoto  Feb 06
   DISCUSSION  Feb 08

7. Genomes and Genome Editing
   O. Shalem  Feb 11

8. Transposable elements
   R. Bushman  Feb 13
   DISCUSSION  Feb 15

9. RNAi and miRNAs
   B. Gregory  Feb 18
   DISCUSSION  Feb 20

1ST EXAM (IN CLASS)  Feb 22
II. GENETICS OF MODEL ORGANISMS

1. *C. elegans* genetics
   - D. Raizen
   - Date: Feb 25
2. Drosophila genetics
   - E. Joyce
   - Date: Feb 27
   - DISCUSSION
   - Date: Mar 01

SPRING BREAK MAR 04-08

3. Mosaic analysis and conditional alleles
   - M. Sundaram
   - Date: Mar 11
4. Maternal effect mutants in zebrafish
   - M. Mullins
   - Date: Mar 13
   - DISCUSSION
   - Date: Mar 15
5. Forward genetics and genomics in the mouse
   - Yana Kamberov
   - Date: Mar 18
6. Reverse genetics in the mouse
   - Maria Golson
   - Date: Mar 20
   - DISCUSSION
   - Date: Mar 22
7. Epistasis and Genetic modifiers
   - M. Sundaram
   - Date: Mar 25
8. Quantitative traits in the mouse
   - E. Brodkin
   - Date: Mar 27
   - DISCUSSION
   - Date: Mar 29

2nd EXAM (TAKE HOME MAR 29 – APR 05)

III. HUMAN GENETICS AND DISEASE

NO CLASS

1. Genome wide genetics for complex traits
   - S. Grant
   - Date: Apr 03
   - DISCUSSION
   - Date: Apr 05
2. Population genetics
   - I. Mathieson
   - Date: Apr 08
3. Human evolution
   - I. Mathieson
   - Date: Apr 10
   - DISCUSSION
   - Date: Apr 12
4. Expression QTL Analysis
   - C. Brown
   - Date: Apr 15
5. Chromosome abnormalities
   - L. Conlin
   - Date: Apr 17
   - DISCUSSION
   - Date: Apr 19
6. X chromosome inactivation
   - M. Bartolomei
   - Date: Apr 22
7. Translational Medicine
   - K. Musunuru
   - Date: Apr 24
   - DISCUSSION
   - Date: Apr 26
8. Cancer genetics and personalized medicine
   - A. Ganguly
   - Date: Apr 29
9. Mitochondrial genetics
   - R. Ganetzky
   - Date: May 01
   - DISCUSSION
   - Date: May 03

3rd EXAM (TAKE HOME MAY 03- MAY 10)
Cell and Molecular Biology 550 “GENETIC PRINCIPLES” Spring Semester 2019

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)
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<tr>
<th>Name</th>
<th>Department</th>
<th>Address</th>
<th>Phone</th>
<th>Email</th>
</tr>
</thead>
<tbody>
<tr>
<td>Marisa Bartolomei</td>
<td>Dept. of Cell &amp; Dev Biology</td>
<td>9-123 Smilow, 8-9063</td>
<td></td>
<td><a href="mailto:bartolom@pennmedicine.upenn.edu">bartolom@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Edward Brodkin</td>
<td>Dept. of Psychiatry</td>
<td>2220 TRL, 215-746-0118</td>
<td></td>
<td><a href="mailto:ebrodkin@pennmedicine.upenn.edu">ebrodkin@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Casey Brown</td>
<td>Dept of Genetics</td>
<td>538A CRB, 650-468-5731</td>
<td></td>
<td><a href="mailto:chrbro@upenn.edu">chrbro@upenn.edu</a></td>
</tr>
<tr>
<td>Rick Bushman</td>
<td>Dept. of Microbiology</td>
<td>426 Johnson Pavilion, 3-8732</td>
<td></td>
<td><a href="mailto:bushman@pennmedicine.upenn.edu">bushman@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Laura Conlin</td>
<td>CHOP, Division of Genomic Diagnostics</td>
<td>716C ARC, 267-426-7885</td>
<td></td>
<td><a href="mailto:conlinl@email.chop.edu">conlinl@email.chop.edu</a></td>
</tr>
<tr>
<td>Marcella Devoto</td>
<td>CHOP Division of Human Genetics</td>
<td>Abramson 1002, 267-426-0124</td>
<td></td>
<td><a href="mailto:devoto@email.chop.edu">devoto@email.chop.edu</a></td>
</tr>
<tr>
<td>Rebecca Ganetzky</td>
<td>CHOP Division of Human Genetics</td>
<td>Abramson 1002, 215-439-5375</td>
<td></td>
<td><a href="mailto:ganetzkyr@mail.chop.edu">ganetzkyr@mail.chop.edu</a></td>
</tr>
<tr>
<td>Arupa Ganguly</td>
<td>Dept of Genetics</td>
<td>415 Anatomy-Chemistry, 8-3122</td>
<td></td>
<td><a href="mailto:ganguly@pennmedicine.upenn.edu">ganguly@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Maria Golson</td>
<td>Dept of Genetics</td>
<td>12-164 Smilow, 8-8712</td>
<td></td>
<td><a href="mailto:mgolson@pennmedicine.upenn.edu">mgolson@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Struan Grant</td>
<td>CHOP, Center for Applied Genomics</td>
<td>Abramson 1216F, 267-426-2795</td>
<td></td>
<td><a href="mailto:grants@email.chop.edu">grants@email.chop.edu</a></td>
</tr>
<tr>
<td>Brian Gregory</td>
<td>Dept of Biology</td>
<td>131 Carolyn Lynch Laboratory</td>
<td></td>
<td><a href="mailto:bdgregor@sas.upenn.edu">bdgregor@sas.upenn.edu</a></td>
</tr>
<tr>
<td>Tom Jongens</td>
<td>Dept of Genetics</td>
<td>10-134 Smilow, 3-9332</td>
<td></td>
<td><a href="mailto:jongens@pennmedicine.upenn.edu">jongens@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Eric Joyce</td>
<td>Dept of Genetics</td>
<td>564 CRB, 8-1229ba</td>
<td></td>
<td><a href="mailto:erjoyce@pennmedicine.upenn.edu">erjoyce@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Yana Kamberov</td>
<td>Dept of Genetics</td>
<td>538A CRB</td>
<td></td>
<td><a href="mailto:yana2@pennmedicine.upenn.edu">yana2@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Ian Mathieson</td>
<td>Dept of Genetics</td>
<td>405B CRB, 3-1835</td>
<td></td>
<td><a href="mailto:mathi@upenn.edu">mathi@upenn.edu</a></td>
</tr>
<tr>
<td>Mary Mullins</td>
<td>Dept of Cell and Developmental Biology</td>
<td>1152 BRBII/III, 8-2644</td>
<td></td>
<td><a href="mailto:mullins@pennmedicine.upenn.edu">mullins@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Kiran Musunuru</td>
<td>Dept of Medicine</td>
<td>11-136 Smilow, 3-1214</td>
<td></td>
<td><a href="mailto:kmus@pennmedicine.upenn.edu">kmus@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>David Raizen</td>
<td>Dept. of Neurology</td>
<td>462 Stemmler</td>
<td></td>
<td><a href="mailto:raizen@pennmedicine.upenn.edu">raizen@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Ophir Shalem</td>
<td>CHOP Center for Cellular and Molecular Therapeutics &amp; Dept. of Genetics</td>
<td>5050 Colkett, 215-590-4168</td>
<td></td>
<td><a href="mailto:shalemo@pennmedicine.upenn.edu">shalemo@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Meera Sundaram</td>
<td>Dept of Genetics</td>
<td>446A CRB, 3-4527</td>
<td></td>
<td><a href="mailto:sundaram@pennmedicine.upenn.edu">sundaram@pennmedicine.upenn.edu</a></td>
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<td>TAs:</td>
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<tr>
<td>Jennifer Cohen, G&amp;E student</td>
<td></td>
<td>445 CRB, 3-4528</td>
<td></td>
<td><a href="mailto:jencohen@pennmedicine.upenn.edu">jencohen@pennmedicine.upenn.edu</a></td>
</tr>
<tr>
<td>Diana Cousminer, Postdoctoral Fellow</td>
<td></td>
<td>1103F, 267-9152</td>
<td></td>
<td><a href="mailto:cousminerd@email.chop.edu">cousminerd@email.chop.edu</a></td>
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Note: TAs: Jennifer Cohen, G&E student