FINAL 12-05-18

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
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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"
http://www.ncbi.nlm.nih.gov:80/books/

I. GENETIC CONCEPTS AND TOOLS

	Lecturer	Date
1. Beyond Mendel	M. Sundaram	Jan 16
	DISCUSSION	Jan 18
MARTIN LUTHER KING'S BIRTHDAY – NO CLASS	-	Jan 21
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	**Lecturer**	**Date**
1. *C. elegans* genetics	D. Raizen	Feb 25
2. Drosophila genetics	E. Joyce	Feb 27
	DISCUSSION	Mar 01
SPRING BREAK MAR 04-08		
3. Mosaic analysis and conditional alleles	M. Sundaram	Mar 11
4. Maternal effect mutants in zebrafish	M. Mullins	Mar 13
	DISCUSSION	Mar 15
5. Forward genetics and genomics in the mouse	Yana Kamberov	Mar 18
6. Reverse genetics in the mouse	Maria Golson	Mar 20
	DISCUSSION	Mar 22
7. Epistasis and Genetic modifiers	M. Sundaram	Mar 25
8. Quantitative traits in the mouse	E. Brodkin	Mar 27
	DISCUSSION	Mar 29
2ND EXAM (TAKE HOME MAR 29 – APR 05)		
III. HUMAN GENETICS AND DISEASE		
NO CLASS	-	Apr 01
1. Genome wide genetics for complex traits	S. Grant	Apr 03
	DISCUSSION	Apr 05
2. Population genetics	I. Mathieson	Apr 08
3. Human evolution	I. Mathieson	Apr 10
	DISCUSSION	Apr 12
4. Expression QTL Analysis	C. Brown	Apr 15
5. Chromosome abnormalities	L. Conlin	Apr 17
	DISCUSSION	Apr 19
6. X chromosome inactivation	M. Bartolomei	Apr 22
7. Translational Medicine	K. Musunuru	Apr 24
	DISCUSSION	Apr 26
8. Cancer genetics and personalized medicine	A. Ganguly	Apr 29
9. Mitochondrial genetics	R. Ganetzky	May 01
	DISCUSSION	May 03
3RD EXAM (TAKE HOME MAY 03- MAY 10)		

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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. <u>The take-home exams must be prepared independently without ANY outside consultation</u>.

Discussion guidelines

The homework problems and discussion are <u>the most important part</u> 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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CAMB 550 Lecturers – 2019

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