

# GNET/BIOL 621 Fall 2017

## Course Policies

**Lecture:** Tues & Thurs 11:00 am - 12:15 pm, 311 Peabody

**Recitation:** Fridays, 2:30 - 3:20 pm, 311 Peabody

**Instructors:** See Sakai site

BIOL/GNET 621 is an upper-level genetics course intended for graduate students and advanced undergraduates. Undergraduates must have taken BIOL 202 or the equivalent; there are no pre-requisites for graduate students. The course covers genetic principles and tools through lectures, reading of research articles, and discussion. We have not assigned a textbook, but you may wish to consult one if you need to review introductory genetics. Any textbook used for BIOL 202 is okay.

### Grading

Final grades will be based on:

- 36% Exams: two midterms and a comprehensive final
- 40% Problem sets
- 24% Recitation (participation and paper presentation)

### Exams

There will be two in-class midterm exams. Each will be 100 points and count for 8% of your final grade. There will also be a comprehensive final exam. This exam will have 150 points from the final third of the course, plus 50 points from each of the first and second thirds of the course, and will be 20% of the final grade.

Exams will consist of questions similar to those on problem sets, and are meant to emphasize conceptual understanding of genetics. No makeup exams will be given; this includes the final! We are required to have a final exam at the time scheduled by the University. If your other courses decided to have an exam at some time of the instructors' choosing and it conflicts with this final, you must arrange for a different time with the other instructor.

### Paper presentation

One or two original research papers will be assigned as reading to accompany each lecture or topic. Each week, a group of 2-3 students will present one of these papers during recitation. Dates will be assigned/chosen at the first recitation meeting. Your presentation counts as 12% of your grade.

When other students are presenting, you will be expected to pay attention and contribute to discussion by asking or answering questions (yes – asking questions counts, too, even questions about not understanding parts of the article), responding to comments by other students, explaining figures or text from the article being discussed, etc. 12% of your grade will be based on your participation in recitation.

### Problem Sets

Problem sets will be assigned most weeks. These will include problems and questions about the lectures and reading. You are encouraged to work collaboratively to solve the problems, but each student must write and turn in his or her own answers. You can turn them in at class or post them to Sakai with the Assignment tool. Problem sets will be graded and returned. Late problem sets will not be accepted. Problem sets count as 40% of your grade.

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## Schedule of class meetings

### Part I: Genetic Principles (Copenhaver)

Aug	22	Tues	Introduction, DNA & chromosome structure Faffney, DJ, et al., (2012) Controls of nucleosome positioning in the human genome. PLOS Genetics 8(11): :e1003036. doi: 10.1371/journal.pgen.1003036
	24	Thurs	Meiosis & mitosis Lutes, AA, et al. (2010) Sister chromosome pairing maintains heterozygosity in parthenogenetic lizards. Nature 464(7286):283-6. doi: 10.1038/nature08818
	26	Fri	<i>No discussion this week</i>
	29	Tues	Mendelian basics Hagstrom, S.A., et al. (1998) <a href="#">Recessive mutations in the gene encoding the tubby-like protein TULP1 in patients with retinitis pigmentosa.</a> Nature Genet. 18: 174-6.
	31	Thurs	Molecular biology basics Long, C, et al. (2014) Prevention of muscular dystrophy in mice by CRISPR/Cas9-mediated editing of germline DNA. Science. 345(6201):1184-8. doi: 10.1126/science
Sept	1	Fri	<i>Discussion (TA presents first paper)</i>
	5	Tues	Recombination Baudat, F., et al. (2010) <a href="#">PRDM9 is a major determinant of meiotic recombination hotspots in humans and mice.</a> Science 327:836-40. McVean, G. & S. Myers (2010) <a href="#">PRDM9 marks the spot.</a> Nature Genet 42: 821-2. (review)
	7	Thurs	Chromosome aberrations Sasaki, M., J. Lange, & S. Keeney (2010) <a href="#">Genome destabilization by homologous recombination in the germ line.</a> Nature Reviews Mol. Cell Biol. 11:182-95.
	8	Fri	<i>Discussion</i>
	12	Tues	Linkage and mapping
	14	Thurs	Pedigrees, tetrads & LODs Sobreira, N.L., et al. (2010) <a href="#">Whole-genome sequencing of a single proband together with linkage analysis identifies a Mendelian disease gene.</a> PLoS Genet. 17: e1000991.
	15	Fri	<i>Discussion</i>
	19	Tues	Association mapping
	21	Thurs	<b>Exam I</b>
	22	Fri	<i>No discussion this week</i>

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## Part II: Genetic Analysis (Sekelsky)

Sept	26	Tues	Genetic Screens Driever, W., <i>et al.</i> (1996) <a href="#">A genetic screen for mutations affecting embryogenesis in zebrafish.</a> <i>Development</i> 123: 37-46.
	28	Thurs	Mutations and Mutagenesis Sivanantharajah L. and A. Percival-Smith (2009) <a href="#">Analysis of the sequence and phenotype of Drosophila Sex combs reduced alleles reveals potential functions of conserved protein motifs of the Sex combs reduced protein.</a> <i>Genetics</i> 182: 191-203.
	29	Fri	<i>Discussion</i>
Oct	3	Tues	Complementation Strathdee, C.A., A.M. Duncan, and M. Buchwald (1992) <a href="#">Evidence for at least four Fanconi anaemia genes including FACC on chromosome 9.</a> <i>Nature Genet.</i> 1: 196-198.
	5	Thurs	Complementation complexities & Genetic interactions Yook, K.J., S.R. Proulx, & E.M. Jorgensen (2001) <a href="#">Rules of nonallelic noncomplementation at the synapse in Caenorhabditis elegans.</a> <i>Genetics</i> 158: 209–220.  Simon, M.A., <i>et al.</i> (1991) <a href="#">Ras1 and a putative guanine nucleotide exchange factor perform crucial steps in signaling by the sevenless protein tyrosine kinase.</a> <i>Cell</i> 67: 701-16.
	6	Fri	<i>Discussion</i>
	10	Tues	Epistasis and pathway analysis Conradt, B. & H.R. Horvitz (1999) <a href="#">The TRA-1A sex determination protein of C. elegans regulates sexually dimorphic cell deaths by repressing the egl-1 cell death activator gene.</a> <i>Cell.</i> 98: 317–327.
	12	Thurs	No class: University Day
	13	Fri	<i>Discussion</i>
	17	Tues	Mosaicism Choate, K.A. <i>et al.</i> (2010) <a href="#">Mitotic recombination in patients with ichthyosis causes reversion of dominant mutations in KRT10.</a> <i>Science</i> 330: 94-97.
	19	Thurs	No class: Fall Break
	20	Fri	<i>No discussion this week</i>
	24	Tues	Mosaic analysis
	26	Thurs	Bacterial genetics Eisenstein, B.I. <i>et al.</i> (1997) <a href="#">Conjugal transfer of the gonococcal penicillinase plasmid.</a> <i>Science</i> 195: 998-1000.  Babic, A. <i>et al.</i> (2008) <a href="#">Direct visualization of horizontal gene transfer.</a> <i>Science</i> 319: 1533-6.
	27	Fri	<i>Discussion</i>
	31	Tues	Genetic interactions Simon, M.A., <i>et al.</i> (1991) <a href="#">Ras1 and a putative guanine nucleotide exchange factor perform crucial steps in signaling by the sevenless protein tyrosine kinase.</a> <i>Cell</i> 67: 701-16.
Nov	2	Thurs	<b>Exam 2</b>

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## Part III: Non-Mendelian Genetics (Ahmed)

Nov	8	Tues	DNA transposons Ivics, Z. <i>et al.</i> (1997) <a href="#">Molecular reconstruction of Sleeping Beauty, a Tc1-like transposon from fish, and its transposition in human cells.</a> <i>Cell</i> 91: 501-510.
	10	Thurs	Retrotransposons Brennecke, J. <i>et al.</i> (2007) <a href="#">Discrete small RNA-generating loci as master regulators of transposon activity in Drosophila.</a> <i>Cell</i> 128: 1089-1103.
	11	Fri	<i>Discussion</i>
	15	Tues	Gene targeting Sandler, J.D. <i>et al.</i> (2011) <a href="#">Selection-free zinc-finger-nuclease engineering by context-dependent assembly (CoDA).</a> <i>Nature Methods</i> 8: 67-69.
	17	Thurs	RNAi I Fire, A. <i>et al.</i> (1998) <a href="#">Potent and specific genetic interference by double-stranded RNA in <i>Caenorhabditis elegans</i>.</a> <i>Nature</i> 391: 806-811.
	18	Fri	<i>Discussion</i>
	22	Tues	RNAi II Boutros, M. <i>et al.</i> (2008) <a href="#">The art and design of genetic screens: RNA interference.</a> <i>Nature Reviews Genetics</i> 9: 554-566.
	24	Thurs	THANKSGIVING
	25	Fri	THANKSGIVING
	29	Tues	Non-Mendelian inheritance Starr, D.J. and T. Cline (2002) <a href="#">A host parasite interaction rescues Drosophila oogenesis defects.</a> <i>Nature</i> 418: 76-79.
Dec	1	Thurs	Epigenetics I Gottschling, D.E. <i>et al.</i> (1990) <a href="#">Position effect at <i>S. cerevisiae</i> telomeres: reversible repression of Pol II transcription.</a> <i>Cell</i> 63: 751-762.
	2	Fri	<i>Discussion</i>
	6	Thurs	Epigenetics II Avner, P. and E. Heard (2004) <a href="#">X-chromosome inactivation: counting, choice and initiation.</a> <i>Nature Reviews Genetics</i> 2: 59-67.  Bartolomei, M.S. <i>et al.</i> (1991) <a href="#">Parental imprinting of the mouse H19 gene.</a> <i>Nature</i> 351: 153-155.
	15	Thurs	<b>CUMMULATIVE FINAL EXAM 12:00 – 3:00 pm</b>