GNET/BIOL 621 Fall 2011

Course Policies

Lecture: Tues/Thurs 11:00 am - 12:15 pm
Recitation: Fridays, 3:00 - 03:50 pm

BIOL/GNET 621 is an upper-level genetics course intended for graduate students and advanced undergraduates. The course will cover 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 the textbooks used for BIOL 202 is appropriate.

Grading

Final grades will be based on:

- 45% Exams: two midterms and a comprehensive final
- 35% Problem sets
- 20% Recitation participation, including paper presentation

Exams

There will be two in-class midterm exams. Each will be 100 points and count for 10% 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 25% 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 at 10% of your grade.

When other students are presenting, you will be expected to pay attention and contribute to discussion by asking or answering questions, responding to comments by other students, etc. 10% 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 Blackboard with the Assignment tool. Problem sets will be graded and returned. Late problem sets will not be accepted. Problem sets count as 35% of your grade.

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Schedule of class meetings

Part I: Genetic Principles (Copenhaver)

Aug	23	Tues	Introduction, DNA & chromosome structure Duan, Z, *et al.* (2010) A three-dimensional model of the yeast genome. *Nature* 465: 363-7.
	25	Thurs	Meiosis & mitosis Bloom, K. and A. Joglekar (2010) Towards building a chromosome segregation machine. *Nature* 463:446-56.
	26	Fri	*Discussion (TAs present 1st paper)*
	30	Tues	Mendelian basics Hagstrom, S.A., *et al.* (1998) Recessive mutations in the gene encoding the tubby-like protein TULP1 in patients with retinitis pigmentosa. *Nature Genet.* 18: 174-6.
Sept	1	Thurs	Molecular biology basics Girisi, P.G., *et al.* (2007) FAIRE (Formaldehyde-Assisted Isolation of Regulatory Elements) isolates active regulatory elements from human chromatin. *Genome Res.* 17: 877-885.
	2	Fri	*Discussion*
	6	Tues	Recombination Baudat, F., *et al.* (2010) PRDM9 is a major determinant of meiotic recombination hotspots in humans and mice. *Science* 327:836-40. McVean, G. & S. Myers (2010) PRDM9 marks the spot. *Nature Genet* 42: 821-2. (review)
	8	Thurs	Chromosome aberrations Sasaki, M., J. Lange, & S. Keeney (2010) Genome destabilization by homologous recombination in the germ line. *Nature Reviews Mol. Cell Biol.* 11:182-95.
	9	Fri	*Discussion*
	13	Tues	Linkage and mapping
	15	Thurs	Pedigrees, tetrads & LODs Sobreira, N.L., *et al.* (2010) Whole-genome sequencing of a single proband together with linkage analysis identifies a Mendelian disease gene. *PLoS Genet.* 17: e1000991.
	16	Fri	NO DISCUSSION MEETING
	20	Tues	Association mapping
	22	**Thurs**	**Exam I**
	23	Fri	NO DISCUSSION MEETING

Part II: Genetic Analysis (Sekelsky)

Sept 27 Tues — Genetic Screens

Driever, W., *et al*. (1996) A genetic screen for mutations affecting embryogenesis in zebrafish. *Development* 123: 37-46.

29 Thurs — Mutations and Mutagenesis

Sivanantharajah L. and A. Percival-Smith (2009) 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. *Genetics* 182: 191-203.

30 Fri — *Discussion*

Oct 4 Tues — Complementation

Strathdee, C.A., A.M. Duncan, and M. Buchwald (1992) Evidence for at least four Fanconi anaemia genes including *FACC* on chromosome 9. *Nature Genet.* 1: 196-198.

6 Thurs — Complementation complexities

Yook, K.J., S.R. Proulx, & E.M. Jorgensen (2001) Rules of nonallelic noncomplementation at the synapse in *Caenorhabditis elegans*. *Genetics* 158: 209–220.

7 Fri — *Discussion*

11 Tues — Genetic interactions

Simon, M.A., *et al.* (1991) Ras1 and a putative guanine nucleotide exchange factor perform crucial steps in signaling by the sevenless protein tyrosine kinase. *Cell* 67: 701-16.

13 Thurs — Epistasis and pathway analysis

Conradt, B. & H.R. Horvitz (1999) The TRA-1A sex determination protein of *C. elegans* regulates sexually dimorphic cell deaths by repressing the *egl-1* cell death activator gene. *Cell*. 98: 317–327.

14 Fri — *Discussion*

18 Tues — Mosaicism

Choate, K.A. *et al.* (2010) Mitotic recombination in patients with ichthyosis causes reversion of dominant mutations in *KRT10*. *Science* 330: 94-97.

20 Thurs — FALL BREAK

22 Fri — FALL BREAK

25 Tues — Mosaic analysis

27 Thurs — Bacterial genetics

Eisenstein, B.I. *et al.* (1997) Conjugal transfer of the gonococcal penicillinase plasmid. *Science* 195: 998-1000.

Babic, A. *et al.* (2008) Direct visualization of horizontal gene transfer. *Science* 319: 1533-6.

28 Fri — *Discussion*

Nov 1 Exam 2

Part III: Non-Mendelian Genetics (Ahmed)

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