BIMM110: Molecular Basis of Human Disease **Spring 2013**
Dr. Bill McGinnis, Dr. Ella Tour **MWF 8-8:50 AM, WLH 2001**

COURSE SYLLABUS

#	DATE	TOPIC	READING	HOMEWORK
1	Mon. 4/1	Overview of course procedures and the syllabus. Recent evolutionary and ecological history of humans and its dramatic influence on the types of disease that afflict humankind.	Supplementary reading on Human evolution: http://tolweb.org/tree/ http://anthropology.si.edu/humanorigins/faq/Encarta/encarta.htm http://www.bradshawfoundation.com/journey/ http://en.wikipedia.org/wiki/Domestication	
2	Wed. 4/3	**Part 1: Genetic diseases** Human pedigree analysis Achondroplasia: misregulation of Receptor Tyrosine Kinase (RTK) pathway in bones	Supplementary reading: *Pedigree analysis*: Strachan, Tom. Human Molecular Genetics, 4th Edition. Garland Science, Ch. 3, pp. 61-78 or any textbook on Genetics: types of inheritance, symbols used in pedigree analysis	
3	Fri. 4/5	Molecular pathways that control bone elongation: RTK signaling, role of FGFR3 Mouse model of Achondroplasia	Mandatory reading: http://depts.washington.edu/bonebio/ASBMRed/growth.html Supplementary reading: Wang et al. (1999) A mouse model for Achondroplasia produced by targeting fibroblast growth factor receptor 3	Homework #1 on Ted, based on the mandatory reading, due before class
4	Mon. 4/8	Use of model organisms to understand the mechanisms of human disease: gene knock-out and gene knock-in	Supplementary reading: Molecular Cell Biology. 4th edition by Lodish, et al http://www.ncbi.nlm.nih.gov/books/NBK21632/#A198 (focus on Gene Targeting Makes It Possible to Produce Transgenic Mice That Are Missing Specific Genes) **or** Molecular Biology of the Cell. 4th edition. Alberts B, Johnson A, Lewis J, et al., focus on Figures 8-70 and 8-71: http://www.ncbi.nlm.nih.gov/books/NBK26818/#A1654	Problem Set 1 discussed in sections this week
5	Wed. 4/10	Hemophilia and the molecular mechanisms of the blood clotting	Supplementary reading on the clotting cascade: Biochemistry. 5th edition. Berg JM, Tymoczko JL, Stryer L. New York: W H Freeman; 2002, online: http://www.ncbi.nlm.nih.gov/books/NBK22589/	
6	Fri. 4/12	Human Karyotype. Meiotic non-disjunction. Chromosomal numerical abnormalities.	Required reading: Human chromosomes, Parts 2.3 – 2.6 in http://www.ncbi.nlm.nih.gov/books/bv.fcgi?highlight=xxy&rid=hmg.section.196#207	Complete homework #2 on Ted before class

7	Mon. 4/15	Down syndrome. Mouse models of DS and study of patients with DS caused by translocations	Data from the following paper discussed: Korbel et al., (2009) The genetic architecture of Down syndrome phenotypes revealed by high-resolution analysis of human segmental trisomies. PNAS 106 (29), 12031–12036	Problem Set 2 discussed this week in sections
8	Wed. 4/17	Sex chromosomal abnormalities. X inactivation.	**Mandatory reading:** X inactivation, Part 2.2.3 http://www.ncbi.nlm.nih.gov/books/bv.fcgi?highlight=xxy&rid=hmg.section.144#152	Complete homework #3 on Ted before class
9	Fri. 4/19	X inactivation and epigenetic regulation of genes expression. Review		
10	**Mon 4/22**	**MIDTERM 1**	Will cover lectures 1-9, will include problems similar to problem sets 1 and 2 and in-class clicker and group discussion questions	
11	Wed. 4/24	Part 2: **Multifactorial diseases – interactions of genes and environment** Diabetes		
12	Fri. 4/26	Type 2 diabetes, obesity and metabolic syndrome	Supplementary reading: Is sugar toxic? http://www.nytimes.com/2011/04/17/magazine/mag-17Sugar-t.html?pagewanted=all&_r=0 **Watch** *Sugar: The Bitter Truth* http://www.youtube.com/watch?v=dBnniua6-oM	
13	Mon. 4/29	Investigations of causes of multifactorial diseases: Epidemiological studies		Problem Set 3 discussed in sections this week
14	Wed. 5/1	Cancer Guest speaker: Reuben Shaw		
15	Fri. 5/3	Cancer, contd. Guest speaker: Reuben Shaw		
16	Mon. 5/6	Cancer: misregulation of cell cycle Cancer treatments		Problem set 4 discussed in sections this week
17	Wed. 5/8	Genome sequencing and personalized medicine Critical thinking: evaluating evidence		

18	Fri. 5/10	Cardiovascular disease		
19	Mon. 5/13	Cardiovascular disease and twin studies		Problem Set 5 discussed in sections this week
20	Wed. 5/15	Summary of Part 2 Critical thinking questions		
21	Fri. 5/17	MIDTERM 2	Will cover lectures 11-20, will include problems similar to problem sets 1 and 2 and in-class clicker and group discussion questions	
22	Mon. 5/20	**Part 3: Infectious Diseases** Microbial and viral diseases		
23	Wed. 5/22	Infectious diseases: microbial and viral		Problem Set 6 discussed in sections this week
24	Fri. 5/24	The innate immune system and defenses against pathogens	Background on innate immunity. http://www.ncbi.nlm.nih.gov/books/NBK26846/	
	Mon. 5/28	**Memorial Day – No class**		
25	Wed. 5/29	The adaptive immune system and its defenses against pathogens	On adaptive immunity: B cells and antibodies: http://www.ncbi.nlm.nih.gov/books/NBK26884/ T cells and MHC proteins: http://www.ncbi.nlm.nih.gov/books/NBK26926/	
26	Fri. 5/31	**Part 4: Neurodegenerative diseases of the brain.**		
27	Mon. 3/7	Neurodegenerative diseases of the brain. Stem cells		Problem Set 7 discussed in sections this week
28	Wed. 3/9	TBD		
29	Fri. 3/11	Summary		
	06/14 8-11AM	**FINAL EXAM Location TBA**	**Comprehensive**	*TA's Review Session TBD*

DESCRIPTION
This course presents 1) genetic, biochemical, and molecular biological approaches used to identify the molecular basis of human diseases; 2) current understanding of selected major human diseases at the molecular and cellular levels; 3) successful and possible therapeutic treatments of these diseases. It is expected that students who take BIMM 110 already have a good background in molecular biology, metabolic biochemistry, and genetics.

COURSE WEBSITE http://classes.biology.ucsd.edu/bimm110.SP13

REQUIRED MATERIALS:
iClicker - a small handheld radio frequency device that you will use to answer questions posed in class each day. Only the iClicker brand will work. New or used iClickers can be purchased at the bookstore. If purchasing iClicker imposes significant financial burden, another option is to sit close to one of the TA's and submit your answers to him or her at the same time as the class is voting via clickers. Clicker participation will contribute to 2% of your final grade. You will be graded based on your participation, regardless whether your answer was right or wrong. To get full credit, you will need to answer (=click) to at least half of the questions in 75% of the lectures. There is no need to notify the instructor or the TA's if you forgot your clicker or ran out of battery - as long as you've participated in 75% of the lectures, you are fine.

TEXTBOOK There is no required course textbook. All lecture slides will be posted on the website and are available for download. The lectures will be podcasted.

OFFICE HOURS
Ella Tour (until May 17): Wed, 9-9:50AM at Mandeville coffee cart, Friday, 1-2PM in York 2300
Bill McGinnis (after May 17): Friday, 1-3PM NSB 6205, Dean's conference room, 6th floor
TA's office hours will be posted on the Contact Info page of the website

GRADING:

2 Midterms, 20% each	40% of the final grade
Final exam	55%
Homework assignments on Ted	3%
Clicker participation	2%

All exams will be closed book/closed computer. Questions will be derived from the lectures, problem sets, homeworks, and questions discussed in class.

Overall course letter grades will be assigned using the following scheme:

87-100%	A (A-, A, A+)
77-86%	B (B-, B, B+)
67-76%	C (C-, C, C+)
50-66%	D
0-49%	F

STUDENTS WITH DISABILITIES Reasonable accommodations will be provided for qualified students with disabilities. If you have any disability that may impair your ability to complete the course successfully, please contact me during the first week of the course.

ACADEMIC INTEGRITY
Absolutely no cheating will be tolerated. UCSD Policies on Academic integrity will be enforced
For further information:
http://blink.ucsd.edu/Blink/External/Topics/Policy/0,1162,19400,00.html

All work must be done by the student to whom it is assigned, without any unauthorized aid of any kind. For the homework assignments, you can discuss the questions with your peers and your TA's, but you must write it on your own, in your own words.