

DATE	TOPIC	HOMEWORK READING	HOMEWORK: SUBMIT	SECTIONS
Fri. 10/3	Introduction, course logistics	None	None	
Mon. 10/6	Hemophilia	Supplementary reading on the clotting cascade: Biochemistry. 5th edition. Berg JM, Tymoczko JL, Stryer L. New York: W H Freeman; 2002, online: <a href="http://www.ncbi.nlm.nih.gov/books/NBK22589/">http://www.ncbi.nlm.nih.gov/books/NBK22589/</a> Starting with Chapter 10.5.5	Complete CURE survey this week, link will be send by email	Week 1: section attendance strongly recommended . Get to know your TA and other members of your section
Wed. 10/8	Hemophilia: genetic treatment in humans?	Paper: Nathwani, Ch, et al. (2011) Adenovirus-associated virus vector-mediated gene transfer in Hemophilia B (find this paper on PubMed: <a href="http://www.ncbi.nlm.nih.gov/pmc/">http://www.ncbi.nlm.nih.gov/pmc/</a> or on Google Scholar. Connect via UCSD web to download this paper for free – you should never pay for papers in this class!) Further reading (optional): High, Nathwani, et al., (2014) Current status of haemophilia gene therapy		
Fri. 10/10	Cystic fibrosis	Read: basic mechanisms Atul Gawande “Better” Ch. The Bell Curve (download the pdf from <a href="http://reserves.ucsd.edu/eres/courseindex.aspx?error=&amp;page=dept">http://reserves.ucsd.edu/eres/courseindex.aspx?error=&amp;page=dept</a> Password: ET110	Online reflection	
Mon., 10/13	Cystic fibrosis	Treatments: paper TBA		Week 2 <b>Section attendance mandatory:</b> groups form and pick their Group name. Submit the names of the students in the group and the name of the group to your TA
Wed. 10/15	Mouse models of single-gene diseases	Supplementary reading: Molecular Cell Biology. 4th edition by Lodish, et al <a href="http://www.ncbi.nlm.nih.gov/books/NBK21632/#A198">http://www.ncbi.nlm.nih.gov/books/NBK21632/#A198</a> (focus on Gene Targeting Makes It Possible to Produce Transgenic Mice That Are Missing Specific Genes) <b>or</b> Molecular Biology of the Cell. 4th edition. Alberts B, Johnson A, Lewis J, et al., focus on Figures 8-70 and 8-71:		

		<a href="http://www.ncbi.nlm.nih.gov/books/NBK26818/#A1654">http://www.ncbi.nlm.nih.gov/books/NBK26818/#A1654</a>		
Fri. 10/17	Achondroplasia (short-limbed dwarfism)	Video lecture: RTK signaling in Achondroplasia <u>Supplemental</u> : How long bones grow: <a href="http://depts.washington.edu/bonebio/ASBMRed/growth.html">http://depts.washington.edu/bonebio/ASBMRed/growth.html</a>		
Mon. 10/20	Normal RTK signaling	Laederich and Horton (2010) Achondroplasia: pathogenesis and implications for future treatment	Complete sections paper 1	<p>Week 3 <b>Sections mandatory: discussion of Paper 1</b></p> <p>Monsonogo-Ornan et al., (2000) Posted on Ted</p> <p>Submit the hard copy of your Section paper in the beginning of the section</p>
Wed. 10/22	Mouse models of Achondroplasia	Wang et al., (1999) A mouse model for achondroplasia produced by targeting fibroblast growth factor receptor 3.		
Fri. 10/24	Wrap-up: single-gene diseases	TBA		
Mon. 10/27	An inexplicable disease	None	Online reflection: Most difficult aspects of scientific papers	<p>Week 4 In sections: review</p>
Wed. 10/29	An inexplicable disease: solution Introduction to Alzheimer's	Watch <a href="http://video.nytimes.com/video/2010/06/01/health/1247467822745/fear-grips-a-family.html?ref=research">http://video.nytimes.com/video/2010/06/01/health/1247467822745/fear-grips-a-family.html?ref=research</a> <a href="http://www.hhmi.org/biointeractive/molecular-mechanism-synaptic-function">http://www.hhmi.org/biointeractive/molecular-mechanism-synaptic-function</a>		
Fri. 10/31	<b>MIDTERM 1</b>	Includes material up to and including Achondroplasia		

Mon. 11/3	Alzheimer's Guest speaker: Dr. Jonathan Aow	Read review: Mattson (2004) Pathways towards and away from Alzheimer's disease		Week 5 <b>Mandatory sections:</b> <b>Paper 2 is discussed:</b> <a href="http://scienceintheclassroom.org/research-papers/deep-clean-your-brain-one-easy-step/university">http://scienceintheclassroom.org/research-papers/deep-clean-your-brain-one-easy-step/university</a>
Wed. 11/5	Studying memory and memory loss	<a href="http://www.hhmi.org/biointeractive/mapping-memory-brain">http://www.hhmi.org/biointeractive/mapping-memory-brain</a> <a href="http://www.hhmi.org/biointeractive/building-brains-molecular-logic-neural-circuits">http://www.hhmi.org/biointeractive/building-brains-molecular-logic-neural-circuits</a>		Submit the hard copy of your Section paper in the beginning of the section
Fri. 11/7	Chronic Traumatic Encephalopathy	<a href="http://www.sportsonearth.com/article/75487104/football-concussions-traumatic-brain-injuries-nfl">http://www.sportsonearth.com/article/75487104/football-concussions-traumatic-brain-injuries-nfl</a> Paper TBA	Online reflection	
Mon. 11/10	Diseases caused by interactions of multiple genes and environment: Diabetes	Watch online lecture: insulin signaling Obesity related health problems, animation: <a href="http://bcove.me/3tvu5v6q">http://bcove.me/3tvu5v6q</a>		6
Wed. 11/12	Diabetes, contd.	Paper TBA		Midterm review in sections
Fri. 11/14	Obesity: genes and molecular mechanisms	Watch: <a href="http://www.ibiology.org/ibiomagazine/jeffrey-friedman-causes-obesity-discovery-leptin.html">http://www.ibiology.org/ibiomagazine/jeffrey-friedman-causes-obesity-discovery-leptin.html</a> Obesity - cancer connection		
Mon. 11/17	The science of fat and sugar	The Science of Fat: watch these two lectures Supplementary reading: Is sugar toxic? <a href="http://www.nytimes.com/2011/04/17/magazine/mag-17Sugar-t.html?pagewanted=all&amp;r=0">http://www.nytimes.com/2011/04/17/magazine/mag-17Sugar-t.html?pagewanted=all&amp;r=0</a> <b>Watch</b> <i>Sugar: The Bitter Truth</i> <a href="http://www.youtube.com/watch?v=dBnniua6-oM">http://www.youtube.com/watch?v=dBnniua6-oM</a>		7 <b>Mandatory sections:</b> <b>Paper 3 is discussed:</b> <a href="http://scienceintheclassroom.org/research-papers/new-diagnostic-strategy-prostate-cancer/university">http://scienceintheclassroom.org/research-papers/new-diagnostic-strategy-prostate-cancer/university</a>
Wed. 11/19	Review, problem solving			Submit the hard copy of your Section

				paper in the beginning of the section
Fri 11/21	<b>MIDTERM</b>	Will cover material from 10/27 – 11/12		
Mon 11/24	Interactions between pathogens and our immune system	<a href="http://www.ibiology.org/ibioseminars/microbiology/stanley-falkow-part-1.html">http://www.ibiology.org/ibioseminars/microbiology/stanley-falkow-part-1.html</a>  <a href="http://www.hhmi.org/biointeractive/cells-immune-system">http://www.hhmi.org/biointeractive/cells-immune-system</a> (if you can't open it, try another browser, like Firefox) Make sure to see this animation from the link above: <a href="http://media.hhmi.org/biointeractive/click/immunology_primer/09-vid.html">http://media.hhmi.org/biointeractive/click/immunology_primer/09-vid.html</a>  <a href="http://www.ibiology.org/ibioeducation/exploring-biology/microbiology-ed/viruses/influenza-virus-infection.html">http://www.ibiology.org/ibioeducation/exploring-biology/microbiology-ed/viruses/influenza-virus-infection.html</a>		Week 8 Review
Wed. 11/26	Flu and other viruses	<a href="http://www.nytimes.com/video/world/africa/100000003107917/dying-of-ebola-at-the-hospital-door.html?playlistId=1194811622283">http://www.nytimes.com/video/world/africa/100000003107917/dying-of-ebola-at-the-hospital-door.html?playlistId=1194811622283</a>		
<b>Fri. 11/28</b>		No class, Happy Thanksgiving!		
Mon. 12/1	Dr. Guichard: Cholera – the disease	Electronic course reserves		<b>Week 9: Mandatory sections: Paper 4:</b> Cohen, Zhang et al. (2013) Influenza A penetrates host mucus by cleaving sialic acids with neuraminidase
Wed. 12/3	Dr. Guichard: studying cholera in flies and human cells	Paper TBA		
Fri. 12/6	Malaria	<a href="http://www.ibiology.org/ibioseminars/microbiology/joseph-derisi-part-1.html">http://www.ibiology.org/ibioseminars/microbiology/joseph-derisi-part-1.html</a> <a href="http://www.hhmi.org/biointeractive/malaria-human-host">http://www.hhmi.org/biointeractive/malaria-human-host</a> <a href="http://www.hhmi.org/biointeractive/malaria-mosquito-host">http://www.hhmi.org/biointeractive/malaria-mosquito-host</a>		

Mon. 12/8	Vaccination	Paper TBA		Week 10: Review
Wed. 12/10	Disease of your choice		CURE survey	
Fri. 12/12	Last class: overview		Online reflection: Most difficult aspects of scientific papers	

**BIMM110: Molecular Basis of Human Disease**  
**Dr. Ella Tour**

**Fall 2014**  
**Mon/Wed/Fri 3-3:50 PM, CNTR 119**

### **COURSE SYLLABUS**

#### **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 GOALS: AT THE END OF THIS COURSE YOU SHOULD BE ABLE TO**

1. Explain the molecular causes of several representative diseases we will examine in this class
2. Current limitations of transgenic approaches in humans
3. Explain how model organisms can be used to understand the mechanisms of diseases: explain the basics of transgenic, knock-out, and knock-in approaches. Be able to come up with a basic experimental design using these approaches.
4. Interpret data from experiments similar to those we examined in class and in sections
5. Demonstrate skills in interpreting primary literature
6. Demonstrate skills of talking about science in public
7. Demonstrate skills of teamwork

**COURSE WEBSITE** [Ted](#)

#### **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 10% of your final grade. The specific breakdown is shown below. 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, starting from week 3 (Oct. 20), you are fine (please see more information below). If getting a clicker device imposes hardship, please talk to the instructor about alternatives.

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

#### **OFFICE HOURS**

Dr. Tour: Thursday, 12:30-1:50 PM, York 3010  
 Also, Dr. Tour is happy to answer questions before and after class

**SECTIONS:**

TA	Email	Section #	Section, day	Section, time	Section, place	Section, room	OH
Yvonne Pao	ypao@ucsd.edu	A01	M	5:00p-5:50p	MANDE	B-104	
Yvonne Pao	ypao@ucsd.edu	A02	M	6:00p-6:50p	MANDE	B-104	
Peter Kim	d1kim@ucsd.edu	A03	Tu	6:00p-6:50p	CENTR	217A	Thu, 3-4pm, Hi Thai
Alisha Caliman	acaliman@ucsd.edu	A05	W	5:00p-5:50p	HSS	2154	Tue, 11am -1pm, Biomedical Library room 227
Alisha Caliman	acaliman@ucsd.edu	A06	W	6:00p-6:50p	HSS	2154	Tue, 11am -1pm, Biomedical Library room 227
Anusha Pasumarthi	apasumar@ucsd.edu	A09	F	4:00p-4:50p	CENTR	207	Mon, 4-5pm at Peet's Coffee (by RIMAC)

**ENROLLING IN SECTIONS:**

You must enroll in one of the sections. Sections in weeks 2, 3, 5, 7, and 9 are mandatory (please see the schedule of classes for details). Enrollment will be open between **Monday, Oct. 6, 10AM to Sunday, Oct. 12, 12PM**. We encourage you to attend multiple sections in week 1 to determine which works best for you.

Enroll on [sections.ucsd.edu](https://sections.ucsd.edu)

Username: bimm110afa14

Password: BIMMJKFA14

**GRADING:**

2 Midterms, 10% each	20% of the final grade
Final exam (cumulative, all material covered)	at least 55% (please see below)
Homework assignments on Ted	5%
Sections papers and participation	10%
Clicker (orange box)	5%
Clicker participation (green box)	5%

**GRADING, IN MORE DETAIL:**

**1. Exams:** Questions on exams will be derived from lectures, problem sets, and papers we'll examine in class and in sections. If your final exam grade is higher than the Midterms, that Midterm grades will be dropped. That grade will be replaced with your final exam score. There will be no re-grades on the Midterms, except when points were added incorrectly. For the Final exam, re-grades can be submitted during exam-viewing sessions.

The grades will be assigned on the standard scale (no curving), so theoretically, everyone can get an A if they score 87% or higher. 100% will be set as the average of 10 highest grades. 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

Missing exams: you can miss one or two Midterm – and have those 20% come from your final exam. However, I strongly recommend taking all midterms, because it's a great low-stress practice. Since it takes several days to write an exam, we will not be able to offer make-up exams. Please check your schedule and make sure that you are available on

the date of the final exam. If you have a conflict with the final exam in another class, please drop this or the other class.

If you are having a family or medical emergency during the final exam, please provide documentation (e.g., emergency room paperwork) and contact the instructor as soon as you can to schedule an oral exam.

**2. Homework assignments:** surveys, reflections on your learning – will be announced in class and by email. Submit at least 75% to get an A in this category.

**3. Sections papers and participation:** Section attendance is mandatory during the sections where Papers 1-4 are discussed. **4%** of your grade will come from your active participation in paper discussions (if you actively participate in your section in other weeks, it will also count). The other **6%** will come from section papers. Guidelines for section papers will be provided. The papers are due in the beginning of the section. They will be graded as follows:  
2 = (S) Satisfactory, 1 = (I) Improvement needed, 0 = (N) No credit

If your total section papers grade is 6 or higher, then 6% of your grade is an automatic A. If your Section papers score is between 3 and 5, then 3% of your grade is an automatic A, the other 3% will be the same as your final exam score. If your Section papers score is less than 3, then all 6% will be the same as your final exam score.

**4. Clicker questions: Orange box questions, 5%.** Each class section will start with several clicker questions that will be based on your understanding of the homework reading. These questions will be framed in orange box and will contribute to 5% of your grade. Answer at least 75% of them correctly in 75% of the lectures (the count will start on Oct. 20, but you can start accumulating points starting week 1) and 5% of your grade is an automatic A  
**Green box questions, 5%.** These are scored based on participation (not whether you answered them correctly). To get full credit, you need to answer (click) to at least half of the green box questions in 75% of the lectures (the count will start on Oct. 20, but you can start accumulating points starting week 1)

**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**

We take academic integrity very seriously. Cheating undermines honest effort and hard work by other students. It will not be tolerated. Cheating on exam, submitting someone else's work as your own, clicking in for another student, copying all or parts of someone else section paper are all examples of academic dishonesty. Please talk to the instructor or the TA immediately if you learn of any incidents of academic dishonesty

UCSD Policy of Academic Integrity, student's responsibilities:

Students are expected to complete the course in compliance with the instructor's standards. No student shall engage in an activity that involves attempting to receive a grade by means other than honest effort; for example:

No student shall knowingly procure, provide, or accept any unauthorized material that contains questions or answers to any examination or assignment that is being, or will be, administered.

No student shall complete, in part or in total, any examination or assignment for another person.

No student shall knowingly allow any examination or assignment to be completed, in part or in whole, for himself or herself by another person.

No student shall plagiarize or copy the work of another person and submit it as his or her own work.

No student shall employ aids excluded by the instructor in undertaking course work or in completing any exam or assignment.

No student shall alter graded class assignments or examinations and then resubmit them for regrading.

No student shall submit substantially the same material in more than one course without prior authorization.

### **Consequences of cheating:**

Cases of cheating will be reported to the Office of Academic Integrity, who will forward them to the Dean of the student's college. In addition, the grade for the assignment in which the cheating occurred will be an 'F'. Cheating on exam will result in 'F' in the course, as well as in administrative consequences. To learn more, please read:

## HOW TO SUCCEED IN THIS CLASS

- ❖ Do the assigned reading. Serious engagement with the material before class will lead to significantly higher gains in class
- ❖ Be proactive, reach out and get help! If you are having troubles with any part of the course material, talk to me or the TA's and come to our office hours. Please don't wait! We care about the success of each and every student and we want to help.
- ❖ Scientific articles are hard. Work with your group or form a study team, and put your collective intelligence to work. Come to my and TA's office hours (and sections) and ask questions. Don't be discouraged if you don't understand everything: I don't understand 100% of every article either!
- ❖ Plan ahead. If you anticipate that you'll need help with a paper or with exam prep, allow yourself enough time to attend office hours and get your questions answered. I or the TA's will not be able to answer last minute questions emailed to us few hours before exam. To get best help, see us in person.
- ❖ Attend classes and sections. Do the section papers and in class activities. It takes time to build up knowledge and skills, don't leave it to the last minute. Cramming the night before the exam will not work in this class.

**Good luck! We want all of you to succeed!**