

## BIMM 110 Molecular Basis of Human Disease, Spring 2012

### Syllabus

Location – Peterson 108

Time - 11:00 AM to 12:20 PM on Tuesdays and Thursdays.

Instructor: Dr. Ella Tour

Website for the course:

<http://classes.biology.ucsd.edu/bimm110.SP12>

Ted: only for clicker registration, grades, discussion forums, paper pdf's:

<https://ted.ucsd.edu/webapps/login/>

Office hours: TBA

### Course 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. Since this is an upper level undergraduate class, it is expected that students who take BIMM 110 already have a good background in molecular biology, metabolic biochemistry, and genetics.

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. In addition, helpful websites or pdfs with background material for course subjects will be posted on the website or WebCT. Please check the website frequently for updates on the posted material.

### Reference textbooks:

1. Molecular Biology of the Cell, 5th Edition, by Bruce Alberts et al. provides useful background information for many subjects in this course. A searchable online edition of the 4th edition of this textbook can be found at:  
<http://www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.View..ShowTOC&rid=mboc4.TOC&depth=10>
2. For some topics on human genetics:  
Human Molecular Genetics (3rd Edition), T. Strachan & A.P. Read, 2004. Available at BML.
3. Wikipedia is a searchable reference website with explanations for nearly all of the specialized terminology used in the course.

Discussion Sections (starting on the second week of classes)

Schedule of the sections and TA's office hours will be posted on the Website within a couple of days. Activities in the discussion sections: TA's will explain the more difficult parts of the material and answer questions; you will solve problems (example exam questions), You can attend any discussion sections.

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. iClicker is a response system that will allow you to solve problems in-class and get immediate feedback from the instructor, gauge your understanding of a topic relative to the rest of the class, and provide your feedback to improve my teaching methods. Your Clicker participation will contribute to 5 pts out of 185 total pts. You will be graded based on your participation, regardless whether your answer was right or wrong. To get full credit, you will need to participate (=click) at least once 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.

Register your iClicker on WebCT by Thursday, January 13 (beginning of the 4th class) to get the first 1 pt for clicker participation. Rest of the clicker credits will be determined as follows:

If you clicked at least once in 75-100% of the lectures - 4 pts

50-75% - 3 pts

25-50% - 2 pts

10- 25% - 1 pt

How to register your iClicker:

Log into WebCT using your UCSD username and password (don't know which username/password to use? [click here](#)). Select this class (BILD1- 2011 Tour). In the Course content/Home page, select and click on Register your iClicker button. To register, locate your iClicker Remote ID - the series of numbers (and perhaps letters). The Remote ID can be found on the back of your remote (there is a helpful picture on WebCT showing where to find it). You should get a notification that you have successfully registered your iClicker remote ID with the system. Now your class participation will be recorded and linked to your student ID. Questions about iClicker? Please see FAQ in the Announcement page of this site.

Grading

The Midterm will count 80 points

The final exam will count 100 points

Clicker participation - 5 points

All exams will be closed book/closed computer. The format of the exams will be similar, i.e. short answers to short questions. Questions will be derived from lecture material and homework reading of papers.

## Schedule of lectures

Week 1. April 3 and 5. Lectures 1 and 2.

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.

Breast cancer and myopia as examples of diseases that are influenced by modern lifestyles. Introduction to epidemiological studies.

Basic background material can be found at:

<http://tolweb.org/tree/>

<http://anthropology.si.edu/humanorigins/faq/Encarta/encarta.htm>

<http://www.bradshawfoundation.com/journey/>

<http://en.wikipedia.org/wiki/Domestication>

### **Part 1: Human Genetic Diseases**

Week 2. April 10 and 12. Lectures 3 and 4.

Human Mendelian genetics. Pedigree analysis.

Achondroplasia and Hemophilia as examples of diseases caused by a mutation in one gene. Mouse models of Achondroplasia: gene knock-out and gene knock-in.

Supplementary readings:

Pedigree analysis: Strachan, Tom. Human Molecular Genetics, 4th Edition. Garland Science, Ch. 3, pp. 61-78

Review on Achondroplasia: [download here](#)

Great resource on how bones grow, how growth is regulated (with animations)

<http://depts.washington.edu/bonebio/ASBMRed/growth.html>

Hemophilia resources:

[http://www.mhhe.com/biosci/esp/2002\\_general/Esp/folder\\_structure/tr/m1/s7/trm1s7\\_3.htm](http://www.mhhe.com/biosci/esp/2002_general/Esp/folder_structure/tr/m1/s7/trm1s7_3.htm)

<http://www.ncbi.nlm.nih.gov/books/NBK22589/>

[http://www.nhlbi.nih.gov/health/dci/Diseases/hemophilia/hemophilia\\_treatments.html](http://www.nhlbi.nih.gov/health/dci/Diseases/hemophilia/hemophilia_treatments.html)

Week 3. April 17 and 19. Lectures 5 and 6.

This weeks discussion sections: discuss Achondroplasia mouse model paper (can be downloaded from Ted)

Lecture 5: Single gene genetic diseases, continued: Cystic fibrosis

Lecture 6: Human Karyotype. Meiotic non-disjunction. Chromosomal numerical abnormalities. Down syndrome.

Resources:

<http://www.ncbi.nlm.nih.gov/books/bv.fcgi?highlight=xxy&rid=hmg.section.196#207>.

Karotyping:

<http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=hmg.section.188>

Week 4. April 24 and 26. Lectures 7 and 8.

**Lecture 7: Quiz 1**

X inactivation. Sex chromosomal abnormalities

Supplementary reading:

X inactivation

<http://www.ncbi.nlm.nih.gov/books/bv.fcgi?highlight=xxy&rid=hmg.section.144#152>

**Part 2: Infectious Diseases**

Lecture 8, April :

Infectious diseases, past and present. The molecular biology of microbial pathogens and antibiotics.

The molecular biology of passive and active defenses against infectious diseases, barriers, innate immunity and adaptive immunity. Are vaccinations harmful?

Resources:

Mechanism of action of cell wall-targeting antibiotics:

[http://www.microbelibrary.org/images/spencer/spencer\\_cellwall.html](http://www.microbelibrary.org/images/spencer/spencer_cellwall.html)

Background on innate immunity.

[http://www.ncbi.nlm.nih.gov/sites/entrez?db=books&cmd=Search&term=innate%20immunity%20AND%20mboc4\[book\]&doptcmdl=TOCView](http://www.ncbi.nlm.nih.gov/sites/entrez?db=books&cmd=Search&term=innate%20immunity%20AND%20mboc4[book]&doptcmdl=TOCView)

On adaptive immunity

[http://www.ncbi.nlm.nih.gov/sites/entrez?db=books&cmd=Search&term=antibody%20AND%20mboc4\[book\]&doptcmdl=TOCView](http://www.ncbi.nlm.nih.gov/sites/entrez?db=books&cmd=Search&term=antibody%20AND%20mboc4[book]&doptcmdl=TOCView)

Week 5, Lectures 9 and 10, May 1 and 3

Global threats: malaria and tuberculosis. Potential terrorist weapon: Anthrax -research and historical perspective.

The molecular biology of viral pathogens.

In sections: Discuss Guichard et al., (2010) Anthrax toxins cooperatively inhibit endocytic recycling by the Rab11/Sec15 exocyst.

**Part 2: Multifactorial diseases**

Week 6. May 8 **Quiz 2**

and 10 Lectures 11 and 12.

Week 7. May 15 and 17. Lectures 12 and 13.

**Lecture 12 May 15:** Cardiovascular disease.

Background sources on cardiovascular disease and statins:

[http://en.wikipedia.org/wiki/Framingham\\_Heart\\_Study](http://en.wikipedia.org/wiki/Framingham_Heart_Study)

<http://en.wikipedia.org/wiki/Statin>

[http://www.spacedoc.net/statins\\_a\\_critical\\_review.htm](http://www.spacedoc.net/statins_a_critical_review.htm)

Lecture 13: The molecular biology of diabetes

Week 8. May 22 and 24. Lectures 14 and 15.

Week 9. March 1 and 3. Lectures 16 and 17.

Cancer – the known molecular mechanisms. The influence of diet, multivitamin intake, etc. on the incidence of cancer.

Useful background material on cancer.

[http://www.ncbi.nlm.nih.gov/sites/entrez?db=books&cmd=Search&term=cancer%20AND%20mboc4\[book\]&doptcmdl=TOCView&log%24=booksrch&bname=mboc4](http://www.ncbi.nlm.nih.gov/sites/entrez?db=books&cmd=Search&term=cancer%20AND%20mboc4[book]&doptcmdl=TOCView&log%24=booksrch&bname=mboc4)

Papers to read (pdf's will be posted on WebCT) and discuss in sections:

Larsson et al. (2004) Fruit and vegetable consumption in relation to ovarian cancer incidence: the Swedish mammography cohort. *British Journal of Cancer*. 90, 2167 – 2170

Health Initiative Cohorts *Arch Intern Med* 169(3):294-304

Neuhouser et al., (2009) Multivitamin Use and Risk of Cancer and Cardiovascular Disease in the Women's

Week 9 Aging

Week 10. March 8, 10. Lectures 18 and 19.

The molecular biology of degenerative diseases of the human nervous system.

Alzheimer's association video: <http://www.youtube.com/watch?v=Z6lA1P2tF0o>

Depression and its treatments

**Final Exam: Thursday, June 12, 11:30AM-2:30 PM. Location TBA**