

# Syllabus, Molecular Basis of Human Disease, BIMM 110, winter 2012

**Time:** 3:30 PM to 4:50 PM, Tuesdays and Thursdays

**Instructor:** Professor Dong-Er Zhang, email: d7zhang@ucsd.edu

**Website for the course:** TED (WebCT) with your own username and password

## Course Description:

This course presents 1) genetic, biochemical, and molecular biological approaches used to identify the molecular basis of human diseases; 2) the current understanding of selected major human diseases at molecular and cellular levels with resulted physiological consequences; 3) successful and possible therapeutic treatment of these human diseases. 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.** Lecture slides will be posted on the website and are available for download.

Reference textbooks:

**1. An Introduction to Human Molecular Genetics (2nd Edition), J.J. Pasternak, 2005.**

**2. Human Molecular Genetics (4th Edition), Tom Strachan & Andrew Read, 2010.** One copy is available in our biomedical library; 3<sup>rd</sup> edition textbooks should be available.

**3. Molecular Biology of the Cell (5th Edition), B. Alberts et al., 2008.**

These three textbooks are also on reserve at BioMedical libraries.

[Wikipedia](#) is a searchable reference website with explanations for nearly all of the specialized terminology used in the course.

## Week 1:

January 10, Lecture 1: Diseases, genes, cell cycles, and chromosomes

January 12, Lecture 2: Human disease pedigree and hemophilia

## Week 2:

January 17, Lecture 3: Gene expression, mutation, and diseases of red blood cells

January 19, Lecture 4: Epigenetics in gene expression, human diseases, and X-inactivation

## Week 3:

January 24, Lecture 5: Meiotic disjunction and chromosomal numerical abnormalities (by PhD graduate TA, Russ DeKelver)

January 26, Lecture 6: Identification of disease genes by analyzing human genome

## Week 4:

January 31, Lecture 7: Cell lines and animal models to study human diseases

February 2, Lecture 8: Muscle disorders

**Week 5:**

February 7, Lecture 9: Cystic fibrosis

February 9, Lecture 10: Diabetes mellitus, an overview (guest lecture, Steven Chessler, MD-PhD)

**Week 6:**

February 14, Midterm exam

February 16, Lecture 11: Cancer and oncogenes

**Week 7:**

February 21, Lecture 12: Signal transduction in cancer and metabolism (guest lecture, Reuben Shaw, PhD)

February 23, Lecture 13: Cell cycle and apoptosis related to cancer/tumor suppressors

**Week 8:**

February 28, Lecture 14: Human Mitochondrial Diseases

March 1, Lecture 15: Neurodegenerative diseases

**Week 9:**

March 6, Lecture 16: Genetic Characteristics of Ophthalmologic Diseases and Treatment (guest lecture, Kang Zhang, MD-PhD)

March 8, Lecture 17: Dynamic mutations and human disease (guest lecture, Albert La Spada, MD-PhD)

**Week 10:**

March 13, Lecture 18: Telomeres, genome stability and aging (guest lecture, Jan Karlseder, PhD)

March 15, Lecture 19: Stem cells and gene therapy

**Class attendance:** Students are expected to attend all lectures. Keep cell phone off or on vibrate mode.

**Discussion sections and office hours of Teaching Assistants (start from week 2):**

**Course grading**

**MIDTERM EXAM:** February 14, 3:30 - 4:50 PM, location will be announced later. The midterm exam will account for 40% of the final grade. No make-up exams.

**FINAL EXAM:** March 20, 3:00 - 5:59 PM, location will be announced later. The final **comprehensive** exam (all lectures) will account for 60% of the final grade.

Both exams will be closed book/closed computer/no any electronics. There will be zero tolerance to any cheating behavior. The format of midterm and final exams will be similar, i.e. short answers to short questions. All questions on both exams will be derived from lecture material.

The midterm and final exam questions with answers from last year are at the WebCT site to assist you to prepare for the exams.

Overall course letter grades will be calculated:  $\text{midterm} \times 40\% + \text{final} \times 60\% = \text{score}$

90-100 - A

78-89 - B

65-77 - C

53-64 - D

0-52 - F