UC San Diego
BIMM 110 Winter 2011
Syllabus

**Instructor:** Dr. Ella Tour **Time:** T/Th 8-9:20AM

Place: Warren Lecture Hall 2001

**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.

Prerequisites: BIMM100 (Molecular Biology), BICD100 (Genetics), BIBC102

(Metabolic Biochemistry)

Website for the course:

http://www.biology.ucsd.edu/classes/bimm110.WI11/

# 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. You 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 notigy 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.

# **Schedule of lectures**

Week 1. January 4 and 6. Lectures 1 and 2

The recent evolutionary and ecological history of humans has had a dramatic influence on the types of disease that afflict humankind. We will review some basics of human evolution, human genetics and pedigree analysis. Several examples of human genetic diseases will be discussed.

Basic background material can be found at:

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

 $\underline{http://www.bradshawfoundation.com/journey/}$ 

http://cogweb.ucla.edu/ep/Templeton\_02.html

#### http://en.wikipedia.org/wiki/Domestication

#### Week 2. January 11 and 13.

# Lecture 3: Human Mendelian genetics. Achodroplasia and Hemophilia.

Review on Achondroplasia: dowload 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.ncbi.nlm.nih.gov/books/NBK22589/

 $http://www.nhlbi.nih.gov/health/dci/Diseases/hemophilia/hemophilia\_treatments.html\\$ 

#### Lecture 4:

The conservation of animal regulatory pathways at the molecular level allows us to use model organisms to gain molecular understanding of human diseases. The most widely used model organisms and tools they offer to study human conditions.

#### Week 3. January 18 and 20. Lectures 5 and 6

Human Karyotype. Meiotic non-disjunction. Chromosomal numerical abnormalities. Down syndrome. X inactivation. Sex chromosomal abnormalities Resources:

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

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

#### Week 4. January 25 and 27. Lectures 7 and 8

# Epigenetics (imprinting) and diseases associated with abnormal imprinting: Angelman, Prader-Willi syndromes. Twin studies and what do they tell us about the influences of genome versus environment.

Background material for genetic imprinting and uniparental disomy at:

http://www.medgen.ubc.ca/wrobinson/mosaic/clinical/prenatal/upd.htm

Karotyping:

http://www.ncbi\_nlm.nih.gov/books/bv.fcgi?rid=hmg.section.188

Human twins: <a href="http://en.wikipedia.org/wiki/Twin">http://en.wikipedia.org/wiki/Twin</a> and twin studies and disease:

http://en.wikipedia.org/wiki/Minnesota Twin Family Study

#### Week 5. February 1 and 3. Lectures 9 and 10

# Single gene or single locus genetic diseases: Cystic fibrosis, FSHD muscular dystrophy.

#### Cardiovascular disease. Statins

Background sources on cardiovascular disease and statins:

http://en.wikipedia.org/wiki/Framingham\_Heart\_Study

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

http://www.spacedoc.net/statins\_a\_critical\_review.htm

#### Week 6.

**Tue, February 8 – MIDTERM EXAM (Lectures 1-10)** 

#### Feb. 10 Lecture 11

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

#### Resources:

Mechanism of action of cell wall-targeting antibiotics:

http://www.microbelibrary.org/images/spencer/spencer\_cellwall.html

#### Week 7. February 15 and 17. Lectures 12 and 13

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

Background on innate immunity.

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

#### Week 8. February 22 and 24. Lectures 14 and 15

The molecular biology of physiological disorders: diabetes and aging.

Current large studies of aging in humans:

The Long Life Family Study:

# http://www.nia.nih.gov/NewsAndEvents/SOAR/v2n1/DiscoveryInnovation/longlives.htm

#### THE NEW ENGLAND CENTENARIAN STUDY

http://www.bumc.bu.edu/centenarian/overview/

C. elegans researcher Cynthia Kenyon talks about the discovery of a genetic pathway involved in longevity:

http://www.ibioseminars.org/kenyon/kenyon1.shtml

#### 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.

 $\frac{\text{http://www.ncbi.nlm.nih.gov/sites/entrez?db=books\&cmd=Search\&term=cancer\%20AN}{D\%20mboc4[book]\&doptcmdl=TOCView\&log\%24=booksrch\&bname=mboc4}$ 

# 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

Neuhouser et al., (2009) Multivitamin Use and Risk of Cancer and Cardiovascular Disease in the Women's Health Initiative Cohorts *Arch Intern Med* 169(3):294-304

Week 10. March 8, 10. Lectures 18 and 19 The molecular biology of degenerative diseases of the human nervous system.

# **Depression and its treatments**

#### Resources:

Li et al. (2010) mTOR-Dependent Synapse Formation Underlies the Rapid Antidepressant effects of NMDA Antagonists. *Science* **329**, 959-964 Commentary on Li et al: Cryan and O'Leary (2010) *Science* **329**, 913-914

Final Exam: Thursday, March 17, 8:00-11:00AM. Location TBA