MMBB422/522: Cellular and Molecular Basis of Disease

Objectives:
This course begins with an exploration of human genetics and the mechanism by which mutations arise in DNA, and how these mutations lead to human diseases. We also cover experimental methods used to detect and analyze these mutations. Part of the discussion focuses on the implications for individuals and societies that arise from the ability to predict the likelihood of disease based on DNA sequence. In the following sections of the course we examine the cellular defects that result due to mutations in specific genes, which leads to loss of expression of proteins or expression of altered proteins. In particular we discuss the development of different types of cancers, and the mechanisms of diseases such as cystic fibrosis, Alzheimer's disease and mad-cow or prion diseases.

Required Textbook: Thompson and Thompson Genetics in Medicine. 7th Edition.
Time and Place: TR 11-12:15 in Life Science South (LSS) Room 163

Office hours: Wed 10:30-11:30 or by appointment. MWF preferred.

Graduate Students presentations:
Graduate Students must give a 30-40 minute oral presentation on a genetic disease of their choice.
Undergraduate Students must either write a 3 page paper about a genetic disease of their choice,
Or give an oral presentation (if time allows). Details to Follow, but papers are generally due the first week of December.

Course grade based on Three Exams (100 pts each), and Presentation/Paper (50 pts), 3 quizzes or assignments (25 pts each). Class participation (25 points). 450 points total.

Course Materials:
Course Handouts will be posted prior to class on the Blackboard class site. All of you should receive an email about the course from Blackboard with instructions of how to access the class site.
https://www.blackboard.uidaho.edu/webct/entryPageIns.dowebct

Exams:
There will be three exams during the year, each covering the material in a portion of the lectures. The exams will be based on the material covered during class and will be closed books. The lecture notes, your class notes and the required readings should be sufficient to study for the exams. The material covered in student presentations will also be part of the third test. **There will be no retests, no extra credit or dropped exams.**
Only official UI excuses will be accepted for missing an exam (i.e. death in the family, orders from doctor or call to military service). In these cases, a retake will be administered during finals and <u>it will be a comprehensive exam</u>. Grading mistakes must be claimed within 1 week from when exam is returned.

Class ethics
Plagiarism and cheating as defined in the UI student manual are not acceptable. First time will result in a 0 in your exam, second time will be grounds for a failing final grade. Additional information about plagarism will be provided when I pass on details on the student papers/presentations.

Disability Support Services Reasonable Accommodations Statement:

Reasonable accommodations are available for students who have documented temporary or permanent disabilities. All accommodations must be approved through Disability Support Services located in the Idaho Commons Building, Room 306 in order to notify your instructor(s) as soon as possible regarding accommodation(s) needed for the course.

- ***885-6307***
- ***email at <dss@uidaho.edu>***
- ***website at <www.access.uidaho.edu>***

Students should present a completed and signed Accommodation Checklist for the current semester, from our office when requesting accommodations. Students should not present the checklist before or after class, if they do, request that they see you during your office hours. If they do not have a current checklist both completed and signed, please refer them to the Disability Support Services office (Idaho Commons, Room 306) to obtain one. If you have any questions regarding a student(s) with a disability(s), or how to best work with a particular student in class, please contact our office.

Syllabus for MMBB422/522: Cellular and Molecular Basis of Disease

Instructor: Jill Johnson

Course Outline

Week of	Tuesday	Thursday
August 22	Overview and implications of DNA age Chapters 1 and 20 and "consequences of genetic information" on blackboard	Chromosome and Gene Structure Chapters 2, 3 and 5 (up to p 69)
August 29	Tools of human molecular genetics. Chapter 4 and part of 5. Preview molecular biology animations in Blackboard folder	Discuss paper about mutation leading to cataracts. Reading in folder in Blackboard. **Assignment 1.**
September 5	Genetic Variation in Individuals. Chapter 9. Also look at extra reading about CODIS information.	Molecular biology terms and techniques Chapter 7. Patterns of single gene inheritance
September 12	End of Chapter 9 and Chapter 10. Gene mapping and Disease gene identification	Paper that demonstrates gene mapping, identification of disease causing mutation
September 19	**More on gene mapping. Review exam 1**	**Exam 1**
September 29	Chapter 11 Hemoglobinopathies	Chapter 12 enzyme defects
October 3	Chapter 12 Receptor proteins, LDL receptors	Paper discussing disease gene
October 10	Ch. 16 genetics and cancer	Ch 16 Genetics and Cancer
October 17	Chapter 12, Structural protein defects DMD, collagen, cystic fibrosis	More on cystic fibrosis, discuss PAPER
October 24	Review and/or another paper	**Exam 2**
October 31	Chapter 12 Alzheimer's disease	Chapter 12 Alzheimer's disease
November 7	Prion diseases	Prion diseases
November 14	Chapter 12 Huntington's disease	Huntington's disease
November 21	**THANKSGIVING BREAK**	**THANKSGIVING BREAK**
November 28	**Chapter 12 Other triplet repeat diseases**	**Paper/Presentation**
December 5	**Student Presentations**	**Student Presentations**
December 12	**Exam 3 Friday Dec. 16 10-12 am**	

Second half of course Syllabus subject to change, but exam dates are set and lectures up until Oct 8 are not expected to change.