

BIOL4110
Human Genetics
Fall 2013, 3 credit hours
M/W/F 11:00-11:50 in Wolfe Hall 1240

INSTRUCTOR:

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OFFICE HOURS

Mon. 4:00- 5:00 pm or by appointment

COURSE DESCRIPTION

In this course we will study basic principles of human genetics and discuss their applications in medical practice. For the basic principles of human genetics, classical patterns of inheritance, chromosome transmission and cytogenetics, molecular-level understandings about DNA structure, function and genetic variations will be first introduced. Then we will learn about past achievements and current status of the human genome project and take a glimpse at how the knowledge about human genome is re-shaping the field of human genetics. More specific topics including population genetics, cancer genetics and immunogenetics will also be presented. A problem-based approach (PBA) will be adopted to explore how to apply basic principles in medical practice. Students are supposed to have already passed BIOL3030 CELL BIOLOGY with a minimum Grade set by the Department of Biological Sciences.

REQUIRED TEXT

A textbook by Bruce R. Korf & Mira B. Irons, "*Human Genetics and Genomics*" (4th ed.) (ISBN: 978-0-4706-5447-7) will provide the framework of lecture materials, but online sources and new developments will be incorporated for up-to-date information. Lecture slides are available for registered students at the course website through UT Blackboard.

"*Human Genetics: Concepts and Applications*" (10th ed.) by Ricki Lewis will be a useful reference (ISBN: 978-0-07-352530-3). The following websites will help students obtain materials for further reading or preparing their presentations.

Book references:

<http://www.synapses.co.uk/genetics/genwebs.html>

<http://www.blackwellpublishing.com/korfgenetics/default.asp>

Course websites:

<http://www2.umdj.edu/~genetics/hg-1.htm>

<http://www.uic.edu/classes/bms/bms655/>

<http://www.kumc.edu/gec/prof/genecour.html>

<http://biology.ucsd.edu/classes/bimm110.SP07/>

<http://www.cs.columbia.edu/~itsik/CHG/CompHumanGen.htm>

Human genetics and human genome:

<http://www.sanger.ac.uk/humgen/>

http://www.ornl.gov/sci/techresources/Human_Genome/home.shtml

<http://www.ncbi.nlm.nih.gov/omim/>

IMPORTANT DATES

Aug. 19- First class

Sept. 20-Last Day to Withdraw

Sept. 27-EXAM I

Oct. 30-EXAM II

Dec. 2-6-Students' Presentations

Dec. 9-EXAM III

STUDENT EVALUATION

Evaluation will be based on attendance/homework/quizzes (20%), three exams (70% total) and one oral presentation (10%).

Exam I and II each counts for 20% of final grade. These two exams will mainly test basic principles in human genetics and include mostly multiple-choice questions. They may also contain a few essay type questions. Exam III will count for 30% of final grade and mainly contain essay type questions to cover applications of human genetics principles in medical practice.

Students are also required to prepare a ~15 min presentation on a specific type of genetic variation/disease or other appropriate topics approved by the instructor. Two students can form a group due to increase in class size. The presentation will count for 10% of final grade. Some questions in Exams will derive from students' presentations.

Assigning 5% of final grade to attendance is mainly to encourage students to ask questions in class, either during the instructor's lectures or during other students' presentations. Quizzes will be held without prior notice. Some homework will be assigned. Quizzes and homework together will count 15% of final grade.

Students arriving more than 10 minutes late for an exam will not be allowed to take the exam. In addition, under no circumstances will students be able to take an exam once other students have completed the exam and left the room.

Bring 2-3 sharpened number 2 pencils with good erasers to the exam.

Students must present a picture I.D. to the instructor or proctors when turning in exams.

If an exam is missed, the instructor must be notified within 48 hours and documentation of the reason for missing the exam must be provided. Acceptable excuses include a death in the immediate family and illness of the student.

Make-up exams will be given at the discretion of the instructors and will consist primarily of essay type questions. Because of this, it is likely that make-up exams will be more difficult than the exam taken in class.

Exams will be based on materials from lectures and assigned textbook readings, however material covered in the lectures will be emphasized so students should attend class and take detailed notes. The instructors will not provide lecture notes, so if you miss a class, be sure to get notes from other students.

GRADING SCALE

% of available marks	Grade	Standard
≥90	A	Achievement of outstanding quality
≥88	A-	Achievement of slightly less than outstanding quality
≥85	B+	Achievement of slightly more than high quality
≥78	B	Achievement of high quality
≥75	B-	Achievement of slightly less than high quality
≥72	C+	Work of slightly more than acceptable quality
≥64	C	Work of acceptable quality
≥62	C-	Work of slightly less than acceptable quality
≥61	D+	Work slightly above the quality expected
≥52	D	Work below the quality expected
≥50	D-	Work slightly below the quality expected

TENTATIVE CLASS SCHEDULE

- Aug. 19 Course introduction/syllabus
21 DNA structure and function: chemical composition, structure and replication
23 DNA structure and function: transcription and translation
26 DNA structure and function: gene inactivation, imprinting and epigenetics
28 Cell Division and Chromosomes: basics
30 Cell Division and Chromosomes: numerical chromosome abnormalities
- Sept. 2 Labor day, No class
4 Cell Division and Chromosomes: structural chromosome abnormalities
6 Genetic Variations: DNA sequence variants
9 Genetic Variations: Detection of DNA sequence variants/polymorphisms
11 Basic principles of heredity: Mendel and Morgan's discoveries
13 Patterns of inheritance: Pedigree analysis
16 Patterns of inheritance: autosomal dominant and recessive inheritance
18 Patterns of inheritance: sex-linked inheritance
20 Patterns of inheritance: penetrance, expressivity and other topics
23 Patterns of inheritance: mitochondrial inheritance
25 review/Students' presentations
27 **Exam I**
30 Fall Break, No Class
- Oct. 2 The human genome: from mapping to sequencing
4 The human genome: genome features and impact on genetic research
7 Multifactorial inheritance
9 Population Genetics
11 Population Genetics
14 Cancer Genetics: oncogenes
16 Cancer Genetics: tumor suppressors
18 Cancer Genetics: genetic background and environment
21 Immunogenetics: blood groups
23 Immunogenetics: MHC and T cell
25 Immunogenetics: Antibody and B cell
28 review/Students' presentations
30 **Exam II**
- Nov. 1 Chromosome abnormalities
4 Molecular diagnoses
6 Newborn screening
8 Developmental Genetics
11 Veteran's Day, No class
13 Carrier Screening
15 Genetic Risk Assessment
18 Risk of Cancer
20 Pharmacogenetics
22 Gene Therapy
25 Bio-ethics in the post-genomic era/Students' presentations
27 No Class Thanksgiving

29 No Class Thanksgiving

Dec. 2 Case studies and Students' presentation

4 More case studies and Students' presentation

6 More case studies and Students' presentation

9 **Exam III**

STATEMENT OF ACADEMIC DISHONESTY

Department of Biological Sciences

Academic dishonesty by students enrolled in undergraduate and graduate courses and programs offered by the Department of Biological Sciences will not be tolerated. Academic dishonesty includes but is not limited to:

1. Obtaining assistance from another individual during an examination.
2. Giving assistance to another individual during an examination.
3. The unauthorized use of study material or textbooks during an examination.
4. Changing answers on an examination after it has been returned and then submitting it for regrading.
5. Plagiarizing written assignments. Plagiarizing includes but is not limited to: a) Copying laboratory reports from previous years, b) copying or paraphrasing reports, term papers, or these prepared by other students, c) unauthorized collaboration in the preparation of reports, term papers, or theses, and d) use of another author's materials without appropriate acknowledgement through quotation and citation.
6. Attempting to bribe or otherwise induce an instructor to alter either a grade or examination score.
7. Obtaining or attempting to obtain a copy of an examination prior to its administration.

In accordance with policies presented in The Student Handbook and The University Catalog, Instructors have the responsibility and right to report cases of alleged dishonesty to departmental, college, and university administrative units. Students involved in academic dishonesty may expect to receive a grade of F on specific assignments as well as in the course where the assignment was made. In addition, disciplinary action may be recommended through appropriate college and university disciplinary committees. Please consult your instructor for instructions on the implementation of this policy.