Human Genetics

The University of Toledo
Department of Biological Sciences, College of Natural Sciences and Mathematics

BIOL4110-001, 3 credit hours

Fall 2015

Wolfe Hall 1240
M/W/F 11:00-11:50; CRN: 58318

INSTRUCTOR:
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OFFICE HOURS

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

COURSE/CATALOG DESCRIPTION

A systematic survey of genetic variation in man with emphasis on modern research methodology.

COURSE OVERVIEW

In this course we will study basic principles of human genetics and genomics 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 first be 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, immunogenetics, pharmacogenetics and gene therapy will also be discussed. 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.

STUDENT LEARNING OUTCOMES
Upon completion of this course, the student will be able to

1. Describe the structures of DNA and chromosomes using proper terms.
2. Understand the features of human genome.
3. Understand mutations at DNA levels and numerical and structural aberrations of chromosomes.
4. Read and interpret cytogenetic results.
5. Understand the laws governing genetic inheritance and perform basic estimation of genetic risks using probability rules.
6. Understand molecular biology foundation of genes, how their activities are regulated at different levels, and how mutations could affect gene activities and lead to specific phenotypes.
7. Have a basic understanding about multifactorial inheritance and population genetics and realize the importance of environment in shaping the phenotypes of individuals or populations.
8. Apply the genetics principles to understand specific inheritable diseases in human beings.
9. Appreciate the contribution of genomics in promoting personalized medicine and precision medicine.
10. Understand some key techniques used in human genetics and genomics.

PREREQUISITES AND COREQUISITES

BIOL 3030 WITH Minimum GRADE OF D-.

REQURIED TEXTS AND ANCILLARY MATERIALS

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 (ISBN: 978-0-07-352530-3) and "*Genetics and Genomics in Medicine*" by Tom Strachean, Judith Goodship and Patrick Chinnery (ISBN: 978-0-8153-4480-3) will be useful references.

The following websites will help students obtain materials for further reading or preparing their presentations.

Book references:

http://www.synapses.co.uk/genetics/index.html

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

Course websites:

http://www2.umdnj.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/

TECHNICAL RQUIREMENTS

Course materials including the syllabus, homework and other supplementary materials will be deposited on the course website hosted at UT BLACKBOARD. Announcements from the instructor and submissions of students' works will also be through BLACKBOARD.

UNIVERSITY POLICIES

Policy Statement on Non---Discrimination on the basis of Disability (ADA)

The University is an equal opportunity educational institution. **Please read** The University's Policy Statement on Nondiscrimination on the Basis of Disability Americans with Disability Act Compliance.

Academic Accommodations

The University of Toledo is committed to providing equal access to education for all students. If you have a documented disability or you believe you have a disability and would like information regarding academic accommodations/adjustments in this course please contact the Student Disability Services Office.

GRADING

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 "short answer" type of questions. Exam III will count for 30% of final grade and mainly contain "short answer" and "essay" types of 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 if class size is too big to accommodate all solo presentations. 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.

The total points will be converted into % and letter grades will be given according to the following table.

% of available marks	Grade	Standard
≥93	A	Achievement of outstanding quality
≥90	A-	Achievement of slightly less than outstanding quality
≥87	B+	Achievement of slightly more than high quality
≥83	B	Achievement of high quality
≥80	B-	Achievement of slightly less than high quality
≥77	C+	Work of slightly more than acceptable quality
≥73	C	Work of acceptable quality
≥70	C-	Work of slightly less than acceptable quality

≥67	D+	Work slightly below the quality expected
≥63	D	Work below the quality expected
≥60	D-	Barely above failing
<60	F	Fail

IMPORTANT DATES

Aug. 24- First class
Oct. 2-EXAM I
Nov. 4-EXAM II
Nov. 30-Dec. 11-Students' Presentations
Dec. 14-EXAM III

TENTATIVE CLASS SCHEDULE

Aug. 24 Course introduction/syllabus
26 DNA structure and function: chemical composition, structure and replication
28 DNA structure and function: transcription and translation
31 DNA structure and function: gene inactivation, imprinting and epigenetics
Sept. 2 Cell Division and Chromosomes: basics
4 Cell Division and Chromosomes: numerical chromosome abnormalities
7 Labor day, No class
9 Cell Division and Chromosomes: structural chromosome abnormalities
11 Genetic Variations: DNA sequence variants
14 Genetic Variations: Detection of DNA sequence variants/polymorphisms
16 Basic principles of heredity: Mendel and Morgan's discoveries
18 Patterns of inheritance: Pedigree analysis
21 Patterns of inheritance: autosomal dominant and recessive inheritance
23 Patterns of inheritance: sex-linked inheritance
25 Patterns of inheritance: penetrance, expressivity and other topics
28 Patterns of inheritance: mitochondrial inheritance
30 review/Students' presentations
Oct. 2 **Exam I**
5 Fall Break, No Class
7 The human genome: from mapping to sequencing
9 The human genome: genome features and impact on genetic research
12 Multifactorial inheritance
14 Population Genetics
16 Population Genetics
19 Cancer Genetics: oncogenes
21 Cancer Genetics: tumor suppressors
23 Cancer Genetics: genetic background and environment
26 Immunogenetics: blood groups
28 Immunogenetics: MHC and T cell
30 Immunogenetics: Antibody and B cell

Nov. 2 review/Students' presentations
4 **Exam II**
6 Chromosome abnormalities
9 Newborn screening
11 Veteran's Day, No class
13 Developmental Genetics
16 Carrier Screening
18 Genetic Risk Assessment
20 Pharmacogenetics
23 Gene Therapy
25 No Class Thanksgiving
27 No Class Thanksgiving
30 Case studies and Students' presentation
Dec. 2 Case studies and Students' presentation
4 More case studies and Students' presentation
7 More case studies and Students' presentation
9 More case studies and Students' presentation
11 More case studies and Students' presentation
14 **Exam III**

Note: Both the class schedule and covered topics may be adjusted at the instructor's discretion.

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.