## BIOL4110 Human Genetics Fall 2011, 3 credit hours

M/W/F 11:00-11:50 in Bowman-Oddy 1031.

### **INSTRUCTOR:**

Dr. Song-Tao Liu WO3223B Tel: 419-530-7853 Email: Song-Tao.Liu@utoledo.edu

#### **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 including population genetics, specific topics 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 taken BIOL3030 CELL BIOLOGY.

#### **REQUIRED TEXT**

A textbook by Bruce R. Korf, "*Human Genetics and Genomics*" (3<sup>rd</sup>. ed.) 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"* (9<sup>th</sup>. ed.) by Ricki Lewis will be a useful reference. 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.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/

## **IMPORTANT DATES**

Aug. 22- First class Sept. 23-Last Day to Withdraw Sept. 30-EXAM I Nov. 2-EXAM II Dec. 5-9-Students' Presentations Dec. 12-EXAM III

## STUDENT EVALUATION

Evaluation will be based on attendance/quizzes (10%), three exams (80% total) and one oral presentation (10%).

Exam I and II each counts for 25% 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  $\sim 15$  min presentation on a specific type of genetic variation/disease or other appropriate topics related to the course. The presentation will count for 10% of final grade. Some questions in Exam III will come from students' presentations.

Assigning 10% of final grade to attendance/quizzes is mainly to encourage students to ask questions in class, either during the instructor's lectures or during other students' presentation of their papers. Quizzes will be held without prior notice.

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.

% of available marks	Grade	Standard
<u>≥90</u>	A	Achievement of outstanding quality
≥88	A-	Achievement of slightly less than outstanding quality
≥85	B+	Achievement of slightly more than high quality
≥78	В	Achievement of high quality
≥75	В-	Achievement of slightly less than high quality
≥72	C+	Work of slightly more than acceptable quality
≥64	С	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 GRADING SCALE**

### CLASS SCHEDULE

Aug. 22 Course introduction/syllabus

24 DNA structure and function: chemical composition, structure and replication

26 DNA structure and function: transcription and translation

29 DNA structure and function: gene inactivation, imprinting and epigenetics

31 Cell Division and Chromosomes: basics

# Sept. 2 Cell Division and Chromosomes: numerical chromosome abnormalities

5 <u>Labor day, No class</u>

7 Cell Division and Chromosomes: structural chromosome abnormalities 9 Genetic Variations: DNA sequence variants

12 Genetic Variations: Detection of DNA sequence variants/polymorphisms

14 Basic principles of heredity: Mendel and Morgan's discoveries

16 Patterns of inheritance: Pedigree analysis

19 Patterns of inheritance: autosomal dominant and recessive inheritance

21 Patterns of inheritance: sex-linked inheritance

23 Patterns of inheritance: penetrance, expressivity and other topics

26 Patterns of inheritance: mitochondrial inheritance

28 review

30 Exam I

Oct. 3 The human genome: from mapping to sequencing

5 The human genome: genome features and impact on genetic research

7 Multifactorial inheritance

10 Population Genetics

12 Population Genetics

14 Cancer Genetics: oncogenes

17 Fall Break, No Class

19 Cancer Genetics: tumor suppressors

21 Cancer Genetics: genetic background and environment

24 Immunogenetics: blood groups

26 Immunogenetics: MHC and T cell

28 Immunogenetics: Antibody and B cell

31 review

## Nov. 2 Exam II

4 Chromosome abnormalities

7 Molecular diagnoses

9 Newborn screening

11 Veteran's Day, No class

14 Developmental Genetics

16 Carrier Screening

18 Genetic Risk Assessment

21 Risk of Cancer

23 No Class Thanksgiving

25 No Class Thanksgiving

28 Pharmacogenetics

30 Gene Therapy

Dec. 2 Bio-ethics in the post-genomic era
5 Case studies and Students' presentation
7 More case studies and Students' presentation
9 More case studies and Students' presentation

12 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.