

CSAT 5025
Genetics
Spring 2023

CLASS DAYS and TIME: Genetics is scheduled to meet on Monday, Wednesday, and Friday, 8:30 - 10:00.

CLASSROOM: The Genetics course is currently scheduled to meet in person in ALTC 2.215. If the graduate school determines that the course should be held “virtually” due to the pandemic, Zoom links will be forwarded to students prior to each session.

COURSE FACULTY: Ellen Kraig, Ph.D.
Christi Walter, Ph.D.
Robin Leach, Ph.D.
Şerife Tekin, Ph.D., Director of Medical Humanities, UTSA (guest lecturer)

OFFICE LOCATION and HOURS: For general aspects of the course, please contact Dr. Kraig. For more specific questions, you may contact any of the faculty instructors directly. We are available for meetings by scheduled appointment; please phone or email to identify a mutually convenient time. Our offices are located on the Long Campus: 4.013V-1 (Kraig), 225D (Walter), and 552C-3 (Leach).

EMAIL: kraig@uthscsa.edu, walter@uthscsa.edu, leach@uthscsa.edu, serife.tekin@utsa.edu

TELEPHONE: Kraig: (210) 367-3171, Walter: (210) 567-3800, Leach: (210) 567-6947

READ THIS DOCUMENT CAREFULLY - YOU ARE RESPONSIBLE FOR ITS CONTENTS

COURSE DESCRIPTION AND OBJECTIVES

This course is designed to provide an overview of current topics in genetics with a focus on mammalian systems. Topics to be discussed include: cytogenetics and chromosome dynamics, mitochondrial genetics, mutagenesis and genomic instability, programmed gene rearrangements and transposable elements, imprinting, genetic variation, linkage and methods for analyzing, population genetics, gene/cell-based therapies, and ethical dilemmas in genetics.

Pre-requisites – this course is designed for students who have already taken Fundamentals of Biomedical Sciences IBMS 5000. In certain situations, the prerequisite may be waived; please consult Dr. Kraig if you wish to petition for enrollment without having taken IBMS 5000.

Semester credit hours – 1 credit hour

By the end of this course, each student should be able to:

- Read, interpret, critically evaluate, and discuss peer reviewed primary scientific papers relevant to genetics, including publications that employ genetic techniques and others studying genetic disorders.
- Demonstrate foundational knowledge of genetics through written exercises, problem sets, oral and written presentations.

COURSE ORGANIZATION

The main teaching modalities used in this course include:

- 1) didactic lectures to deliver foundational and factual information about genetics
- 2) self-study activities and online review to prepare for upcoming lectures and activities
- 3) student presentations on genetic topics of interest
- 4) problem sets and paper reviews

Materials – The required textbook, *Thompson & Thompson Genetics in Medicine*, 8th edition, by Nussbaum, McInnes, and Willard, is digitally available at the link below through the Clinical Key resource.

<https://www-clinicalkey-com.libproxy.uthscsa.edu#!/browse/book/3-s2.0-C2009059798X>

Additional readings may be posted to CANVAS prior to individual lectures.

Computer Access – Various materials will be posted to CANVAS so students will require access to a computer with internet capabilities. *Also, since this course will likely be taught using Zoom, in order to facilitate student participation in class and presentations, you will need to have both a functional camera and microphone on your computer.*

Reading Assignments – It is expected that students will already have a working knowledge of basic genetic principles (including concepts like “recessive/dominant”, “P1, F1, F2”, “meiosis, mitosis”, “linkage”, “pedigree”, “penetrance/expressivity”, etc.). Any basic genetics text should suffice for reviewing these fundamental concepts. If any of these terms are unfamiliar and you would like additional reading material, please contact Dr. Kraig.

Thompson & Thompson Genetics in Medicine, 8th edition, is the required textbook for this course; relevant sections that would prepare students for the individual lectures are noted on the attached class schedule. Additional reading assignments, such as primary scientific papers, may also be posted on CANVAS.

NOTE: Students will each need to complete three karyotypes from an online location (link below), take screen shots of the completed karyotypes and send the screen shots to Dr. Walter (walter@uthscsa.edu) before class on January 11, 2023. The first lecture on chromosomes will prepare the student for the independent karyotyping activity.

Link to karyotypes:

http://www.sih.wisc.edu/wp-content/uploads/articulate_uploads/KARYOTYPER-Chromosome-Identification/story.html

ATTENDANCE

Attendance at every session is expected to achieve the expected level of competency. If a student has an allowable absence (i.e., an out-of-town scientific conference), he/she should contact Dr. Kraig as soon as he/she becomes aware of the conflict so that accommodations can be made. Attendance and participation at the student presentations are mandatory. If a student becomes ill or misses a class for any other reason, he/she must contact Dr. Kraig and apprise her of the situation. If there are chronic or unexplained absences, the student’s grade will be lowered.

GRADING POLICIES AND EXAMINATION PROCEDURES

The student’s grade will be based on a composite score comprised of his/her performance on written exercises, class participation, and student presentations. Grading is on a letter scale with the following minimal requirements: A ≥ 90%, B ≥ 80%, C ≥ 70%, D ≥ 60%, F < 60%. Fractions are rounded to the nearest whole number for the final grade. Passing requires a grade of A or B.

Exam: There will be NO comprehensive exam over the course. There will be problem sets and exercises associated with the individual lectures and some of these will count towards the grade.

Student presentations: Each student will prepare a presentation on a genetic disease chosen by the student from a list provided in class, in consultation with the faculty instructors. The grade determined by the instructors will be based on the student’s performance, including the scholarliness of the material selected (e.g. primary peer reviewed scientific articles in credible scientific journals), the care in preparation of the slides, and the clarity of the oral presentation. In addition, the student will prepare a brochure (in lay language) covering his/her selected disease, its underlying genetic cause, treatment options, and other important and interesting information.

The final grade will be comprised of the following components:

Final student presentation/brochure (February 3 rd and 6 th)	50%
Problem set associated with Dr. Leach’s classes; due 1/28/23	15%
Karyotype exercise associated with Dr. Walter’s class; due 1/11/23	10%
Exercises associated with remaining classes; see schedule 4 x 5%	20%
Incorporation of gene therapy into final presentation	5%

REQUESTS FOR ACCOMMODATIONS FOR DISABILITIES

In accordance with policy 4.2.3, **Request for Accommodation Under the ADA and the ADA Amendments Act of 2008 (ADAAA)**, any student requesting accommodation must submit the appropriate request for accommodation under the American with Disabilities Act (ADA, form 100). to his/her appropriate Associate Dean of their School and a copy to the ADA Coordinator. Additional information may be obtained at <http://uthscsa.edu/eo/request.asp>.

ACADEMIC INTEGRITY AND PROFESSIONALISM

Any student who commits an act of academic dishonesty is subject to discipline as prescribed by the UT System Rules and Regulations of the Board of Regents. Academic dishonesty includes, but is not limited to, cheating, plagiarism, collusion, the submission for credit of any work or materials that are attributable in whole or in part to another person, taking an exam for another person, signing attendance sheets for another student, and any act designed to give unfair advantage to a student or the attempt to commit such an act. Additional information may be obtained at <http://catalog.uthscsa.edu/generalinformation/generalacademicpolicies/academicdishonestypolicy/>

TITLE IX AT UTHSCSA

Title IX Defined:

Title of the Education Amendments of 1972 is a federal law that prohibits sex discrimination in education. It reads “no person in the United States shall, on the basis of sex, be excluded from participation in, be denied the benefits of, or be subjected to discrimination under any education program or activity receiving Federal financial assistance.”

University of Texas Health Science Center San Antonio’s Commitment:

University of Texas Health Science Center San Antonio (UTHSCSA) is committed to maintaining a learning environment that is free from discriminatory conduct based on gender. As required by Title IX, UTHSCSA does not discriminate on the basis of sex in its education programs and activities, and it encourages any student, faculty, or staff member who thinks that he or she has been subjected to sex discrimination, sexual harassment (including sexual violence) or sexual misconduct to immediately report the incident to the Title IX Director.

In an emergency, victims of sexual abuse should call 911. For non-emergencies, they may contact UPD at 210-567-2800. Additional information may be obtained at <http://students.uthscsa.edu/titleix/>

EMAIL POLICY

As a matter of University Policy, official communications between students and faculty occur using the student’s university assigned “livemail” email address. Students are expected to check their university email on a daily basis. Missed communication due to inadequate monitoring of university email is not a valid excuse for failing to perform expected activities. Students are welcome to email the instructors at any time.

USE OF RECORDING DEVICES

Course policy allows the use of recording devices, if given permission by the presenter.

ELECTRONIC DEVICES

Cell phones shall not be used during class (unless requested to do so by the instructors). Use of social media or email via any devices is not allowed during class.

TENTATIVE CLASS SCHEDULE
CSAT 5025 Genetics Spring 2023

DATE	TOPIC	READING	ASSIGNMENT	
Jan 9	Chromosomes, Cytogenetics, Breakage, Aneuploidy, Syndromes	<p><u>Chapter 2</u>: Structure of Human Chromosomes; Organization of the Human Genome, Mitosis, The Human Karyotype; Meiosis, Medical Relevance of Mitosis and Meiosis</p> <p><u>Chapter 5</u>: all</p> <p><u>Chapter 6</u>: all</p>	<p>http://www.slh.wisc.edu/wp-content/uploads/articulate_uploads/KAR_YOTYPER-Chromosome-Identification/story.html</p> <p>show completed karyotypes (screen shots) for chromosome matching sets (# 4), for sets 1a, 2a and 4a and name the abnormality when present.</p> <p>Karyotype assignment is due <u>before class</u> on January 11, 2023</p>	
Jan 11	Mitochondrial genetics & diseases	<p><u>Chapter 2</u>: the Mitochondrial Chromosome</p> <p><u>Chapter 7</u>: Maternal Inheritance of Disorders Caused by Mutations in the Mitochondrial Genome and subsections,</p> <p><u>Chapter 11</u>: The mt DNA Genome and the Genetics of mt DNA diseases</p>	<p>Complete required reading before class.</p> <p>Homework due <u>by noon</u> on 1/13/23</p>	Walter
Jan 13	Mutagenesis & triple repeats	<p><u>Chapter 4</u>: The Origin and Frequency of Different types of Mutations, Regional Mutations, Gene Mutations, Sex Difference and Age Effects on Mutation Rates, Types of Mutations and Their Consequences (all subsections), Impact of Mutation and Polymorphism</p> <p><u>Chapter 7</u>: Dynamic Mutations: Unstable Repeat Expansions (all subsections)</p>	<p>Complete reading before class</p> <p>Homework due <u>by noon</u> on 1/17/23</p>	
Jan 16	No class (MLK Day)			
Jan 18	Genetic variation (including CNVs), linkage, QTLs	<p><u>Chapter 4</u>: <u>The Nature of Genetic Variation, Inherited Variants and Polymorphisms in DNA</u></p> <p>Chapter 10: Identifying the Genetic Basis for Human Disease – Full Chapter</p>	<p>Practice set for population genetics will be distributed after class on the 1/20/23 and will be reviewed on 1/23/23.</p> <p>Homework will be distributed on 1/23/23 after class and is due on 1/30/23.</p>	Leach
Jan 20	Population genetics and risk assessment	<u>Chapter 9</u> : Genetic Variations in Populations – Full chapter		

Jan 23	Imprinting and X chromosome inactivation	<p><u>Chapter 3:</u> Epigenetics and Epigenomics Aspects of Gene Expression; Allelic Imbalance in Gene Expression.</p> <p><u>Chapter 6:</u> Disorders Associated with Genomic Imprinting,</p> <p><u>Chapter 7:</u> X-Linked Inheritance</p>	Problem set due by 9:00 am on 1/27/23	Leach
Jan 25	Programmed gene rearrangements and transposable elements	Reviews will be provided in CANVAS; textbook is very limited in these areas.	Exercise due by 9:00 am on 2/1/23	Kraig
Jan 27	Gene therapy and cell based therapies	<u>Chapter 13:</u> Treatment of genetic disease (entire chapter)	<p>No specific assignment</p> <p>Gene therapy must be incorporated in your final presentation</p>	
Jan 30	Ethical dilemmas in genetics			Tekin
Feb 1	<u>No class:</u> time to work on presentations		Reference lists due to all three instructors by 5:00 pm 2/1/23	
Feb 3	Student presentations/ brochures			
Feb 6	Student presentations/ brochures			