

Course Syllabus – *Draft 20191219*
Department of Human Genetics
HUGEN 2060: Chromosomes - Structure and Function
HUMAN GENETICS
Spring Semester 2020 – 2 credits
Wednesday and Friday, 9:00-10:25AM, Room A216 Public Health

The instructor reserves the right to make changes to the schedule.

Instructor:

Candy Kammerer, PhD
Room: 3120 Public Health
Phone: 412-624-7265 (work)
E-Mail: cmk3@pitt.edu
Office hours: Before and after class, and by appointment

Course Description:

Chromosomes are the primary means of biologically organizing and manipulating nuclear DNA within the cell, across cell generations, and across sexual generations. In this course we will investigate how differences in chromosome structure and function affect the roles chromosomes play as dynamic “megamolecules” in reproduction, development, health and disease.

Course Goal

The goal of this course is to provide a thorough grounding in the field of cytogenetics by conveying key concepts about the dynamic relationship between chromosome structure and function, and its impact on cellular maintenance, recombination, genetic and epigenetic transmission of genetic material across generations, and evolution. Historical and current examples from humans and other species will be used to gain insights into cytogenetic phenomena and mechanisms. The course will also include discussion of several methodologies, e.g., high-C chromosome capture, used to discern the relationship between structure, function, and dysfunction, aka, disease. A few specific topics include meiosis, mitosis, recombination, chromatin, imprinting, cistromes, topologically associated domains (TADs), sex determination, reproduction/infertility, chromosome stability/instability, and cancer genetics. Overall, the course will include a comparative cytogenetics approach because “Nothing in biology makes sense, except in the light of evolution”, Theodosius Dobzhansky (1973).

Learning Objectives:

After completion of this course, the student will be able to:

- Compare and contrast the chromosome mechanisms (structure and function) involved in mitosis and meiosis
- Interpret cytogenetic nomenclature
- Compare and contrast imprinting mechanisms across species
- Describe effects of chromosome constitution on sex determination, reproduction, and infertility
- Describe the role of chromosomal instability and copy number variants in development of cancer and other disorders

Teaching Philosophy:

This course emphasizes active participation, critical thinking, and continued learning. Because we all know different things and have experienced different events, all questions and viewpoints are encouraged and respected in the classroom and within groups. Science advances by observing, asking questions, and listening.

Text:

The Principles of Clinical Cytogenetics. SL Gersen, MB Keagle. 2013 SpringerLink
Available online free for University of Pittsburgh from ULS & HSLs
<https://link.springer.com/book/10.1007/978-1-4419-1688-4>

Additional articles and handouts will be posted on Courseweb.

Grading:

The grading for this course is based on multiple homework exercises, quizzes, in-class activities, and two exams. Your grade for the course is based upon your work as follows:

In-class activities/discussion	10%
Homework	15%
Quizzes	15%
Midterm exam (in-class)	30%
Final exam (take-home)	30%

Except for the exams and in-class quizzes, students may help each other to achieve the best work you are capable of producing. Working with one another to achieve mastery will help you learn the material with greater ease and enjoyment.

Grading scale:

- 94- 100% = A
- 90-93% = A-
- 87-89% = B+
- 84-86% = B
- 80-83% = B-
- 70-79% = C
- Below 70% = F

Homework

Several short homework assignments (5-8) will be assigned throughout the semester. Students are encouraged to work cooperatively, but each student must submit their own work. Homework assignments will be assessed for completeness and effort. Late homework assignments will not be accepted unless the instructor gives prior approval.

All homework assignments will be posted on Courseweb.

Quizzes

Several quizzes (at least five) will be given at the beginning of class throughout the semester and will comprise 1-3 questions each (total 5-10 minutes) and will cover the same material as the homework assignments. They will be graded for correctness. The lowest quiz grade will be dropped.

Class Schedule:

Date	Topic	G&K Chapters; Readings are online
	MODULE 1: Concepts and Background	
Jan 8	Introduction to course; history of chromosomes, mitosis	1
Jan 10	Cell cycle, replication, mechanics of chromosome segregation, chromosome chemistry and packaging	2, Readings
Jan 15	Basic cytogenetic methods, chromosome nomenclature	3, 4; ISCN(2016)
Jan 17	Karyotypes, comparative cytogenetics, evolution	Readings
Jan 22	Chromosome interactions, Chromosome capture conformation (hi-C) methods	Readings
Jan 24	Structural chromosome variations (aneuploidy, rearrangements)	8,9
Jan 29	Meiosis	Readings
Jan 31	Recombination, mechanisms	Readings
Feb 5	Polyploidy, chromosomes and evolution	Readings
Feb 7	Sex Determination, X and Y chromosomes	10, Readings
Feb 12	Exam 1 (covers material through Feb 5)	
Feb 14	Sex determination systems and disorders	10
Feb 19	X-inactivation, imprinting, development	20, Readings
Feb.21	Chromosome Instability, copy number variants	14
Feb 26	DNA Repair Defects; Chromosomal Breakage Syndromes; Cancer Predisposition Guest Lecturer: Patricia L. Opresko, Ph.D.	Readings
Feb 28	Fragile X – A family of disorders and review	19
Mar 4	Cancer Genetics 1. The Basics: Oncogenes/Tumor suppressor genes/ Mismatch Repair Genes/ DNA Repair Genes/ Inherited Cancer Predisposition. Guest Lecturer: Phuong L. Mai, M.D	Readings
Mar 6	Cancer Genetics 2. More in depth of cancer cytogenetics	Readings

Date	Topic	G&K Chapters; Readings are online
	Familial Cancer Syndromes, and Predisposing Genes. Family History Essentials, Counseling, and Testing. Guest Lecturer: Phuong L. Mai, M.D	
Mar 11-13	Spring Recess	
Mar 18	Topic of Student choice	
Mar 20	FINAL EXAM – take home	

Diversity:

The University of Pittsburgh Graduate School of Public Health considers the diversity of its students, faculty, and staff to be a strength and critical to its educational mission. Pitt Public Health is committed to creating and fostering inclusive learning environments that value human dignity and equity. Every member of our community is expected to be respectful of the individual perspectives, experiences, behaviors, worldviews, and backgrounds of others. While intellectual disagreement may be constructive, no derogatory statements, or demeaning or discriminatory behavior will be permitted.

If you feel uncomfortable or would like to discuss a situation, please contact any of the following:

- the course instructor;
- the Pitt Public Health Associate Dean for Diversity at 412-624-3506 or nam137@pitt.edu;
- the University’s Office of Diversity and Inclusion at 412-648-7860 or <https://www.diversity.pitt.edu/make-report/report-form> (anonymous reporting form).

Disabilities:

If you have a disability for which you are or may be requesting an accommodation, you are encouraged to contact both your instructor and [Disability Resources and Services](#), 140 William Pitt Union, 412-648-7890 as early as possible in the term.

Academic Integrity:

All students are expected to adhere to the school’s standards of academic honesty. Cheating/plagiarism will not be tolerated. The Graduate School of Public Health’s policy on academic integrity, which is based on the University policy, is available online in the Pitt Public Health Academic Handbook www.publichealth.pitt.edu/home/academics/academic-requirements. The policy includes obligations for faculty and students, procedures for adjudicating violations, and other critical information. Please take the time to read this policy.

Sexual Misconduct, Required Reporting and Title IX Statement:

The University is committed to combatting sexual misconduct. As a result, you should know that University faculty and staff members are required to report any instances of sexual misconduct, including harassment

and sexual violence, to the University's Title IX office so that the victim may be provided appropriate resources and support options. What this means is that as your professor, I am required to report any incidents of sexual misconduct that are directly reported to me, or of which I am somehow made aware.

There are two important exceptions to this requirement about which you should be aware:
A list of the designated University employees who, as counselors and medical professionals, do not have this reporting responsibility and can maintain confidentiality, can be found here: www.titleix.pitt.edu/report/confidentiality

An important exception to the reporting requirement exists for academic work. Disclosures about sexual misconduct that are shared as part of an academic project, classroom discussion, or course assignment, are not required to be disclosed to the University's Title IX office.

If you are the victim of sexual misconduct, Pitt encourages you to reach out to these resources:

- Title IX Office: 412-648-7860
- SHARE @ the University Counseling Center: 412-648-7930 (8:30 A.M. TO 5 P.M. M-F) and 412-648-7856 (AFTER BUSINESS HOURS)

If you have a safety concern, please contact the University of Pittsburgh Police, 412-624-2121.
Other reporting information is available here: www.titleix.pitt.edu/report-0

Statement from the Department of Gender, Sexuality, and Women's Studies

[This statement was developed by Katie Pope, Title IX Coordinator, in conjunction with GSWS instructors.]