

Course Syllabus
Department of Human Genetics
HUGEN 2060: Chromosomes - Structure and Function
HUMAN GENETICS
Spring Semester 2022 – 2 credits
Wednesday and Friday, 9:00-10:25 AM,
A115, Pitt Public Health (or virtual)

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



Course Director:

Quasar Padiath (QSP), MBBS, PhD



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Teaching assistant: - Jonathan Chernus

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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, function and chromatin organization, and its impact on cellular maintenance, recombination, genetic and epigenetic transmission of genetic material across generations. Historical and current examples from humans will be used to gain insights into cytogenetic phenomena and mechanisms. The course will also include an overview of several methodologies, e.g., G-banding, aCGH, Chip-Seq, high-C chromosome capture, used to discern the relationship between structure, function, and disease. A few specific topics are: chromosome structure, chromatin organization, meiosis, mitosis, recombination, topologically associated domains (TADs) structural and numerical chromosomal abnormalities, chromosome nomenclature, imprinting, sex determination, reproduction/infertility, chromosome stability/instability, and cancer genetics.

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.

Understand the basic concepts underlying chromatin organization.

Understand mechanisms of numerical and structural chromosomal aberrations and diseases associated with them. 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, questioning, listening, and discussing.

Texts:

Thompson & Thompson *Genetics in Medicine, (GM), Eighth Edition*

Available online free for University of Pittsburgh from ULS & HSLS

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

SL Gersen, MB Keagle. *The Principles of Clinical Cytogenetics (PCG)*. 2013 SpringerLink
On Canvas and Box

Available online free for University of Pittsburgh from ULS & HSLS

This text is older, but it will provide background for some basic information.

Additional articles and handouts will be posted on Canvas

Grading:

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

Assignments/Quizzes	30%
Midterm exam	30%
Final exam	30%
Class participation (attendance/questions)	10%

Except for the exams and 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 homework assignments or in-class activities will be assigned throughout the semester. Students are encouraged to work cooperatively, but each student must submit their own work, unless advised otherwise. 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.

Course Agenda

Guest lecturers for the course are listed below and noted on the course schedule by their initials.

Guests Lecturers: Melanie Babcock (MB), Ph.D., DABMGG
 Mahmoud Aarabi (MA), M.D., Ph.D.
 Svetlana Yatsenko (SY), M.D.
 Marie DeFrancis (MDF), M.D., Ph.D.
 Patricia L. Opresko, (PLO), Ph.D.
 Phuong L. Mai, (PLM), M.D.

Class Schedule: Wednesday and Friday, 9:00-10:25 AM, A115 (or virtual), Pitt Public Health

Date	Topic	Lecturer	GM, PCG Chapters; Readings are online
	<i>Schedule Topics Subject to Change</i>		
Jan 12 (Virtual)	Introduction to course; history of chromosomes, Chromosome facts and structure, Chromatin organization	QSP	<i>PCG-1,2 & GM-2</i>
Jan 14 (Virtual)	Centromere, Telomeres, TADs	QSP	<i>PCG-2 & GM-2, Readings</i>
Jan 19 (Virtual)	Intro to cell cycle, Mitosis	QSP	<i>GM-3, Readings</i>
Jan 21 (Virtual)	Meiosis, Recombination	QSP	<i>GM-3 & PCG-2</i>
Jan 26 (Virtual)	Recombination, numerical abnormalities	MB	<i>PCG-8 & GM-5,6</i>
Jan 28	Chromosome rearrangement-Structural abnormalities	MB	<i>PCG-9 & GM-5,6</i>
Feb 2	Contiguous gene deletions	MB	<i>PCG-9 & GM-5,6</i>
Feb 4	Nomenclature/in-class exercises	MA	<i>PCG-3, ISCN (2020)</i>
Feb 9	Catch-up/Follow-up/Review		Readings
Feb 11	Exam 1 (covers material through Feb 9)		

Date	Topic	Lecturer	GM, PCG Chapters; Readings are online
Feb 16	Prenatal testing-Detection of chromosomal abnormalities	SY	Readings
Feb 18	Sex determination/Dosage compensation and disorders	SY	PCG-10 & GM-6
Feb 23	Epigenetics/Uniparental disomy/Imprinting -XCI development	SY	PCG-10, 20 & GM-6
Feb 25	Fragile X – A family of disorders	MDF	PCG-19 & GM-7
Mar 2	Chromosomal abnormalities in hematological cancers	MA	PCG-15, Readings
Mar 4	DNA Repair Defects; Chromosomal Breakage Syndromes; Cancer Predisposition	PLO	Readings
Mar 9	SPRING BREAK		
Mar 11	SPRING BREAK		
Mar 16 - PM	Cancer Genetics 1. The Basics: Oncogenes/Tumor suppressor genes/ Mismatch Repair Genes/ DNA Repair Genes/ Inherited Cancer Predisposition.	PLM	Readings
Mar18	Cancer Genetics 2. More in depth of cancer genetics Familial Cancer Syndromes, and Predisposing Genes. Family History Essentials, Counseling, and Testing.	PLM	Readings
Mar 23	Catch-up/Follow-up/Review		
Mar 25	FINAL EXAM – In person		

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 and Inclusion*](#)
- the University's Office of Diversity and Inclusion at 412-648-7860 or <https://www.diversity.pitt.edu/civil-rights-title-ix-compliance/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: :

<https://www.diversity.pitt.edu/civil-rights-title-ix-compliance/make-report/confidentiality-and-retaliation>

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: <https://www.diversity.pitt.edu/civil-rights-title-ix-compliance/make-report>

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.