

Course Syllabus  
Department of Human Genetics, School of Public Health  
HUGEN 2060: Chromosomes - Structure and Function  
Spring Semester 2023 – 2 credits  
Friday, 9:00-10:50 AM,  
A115, Pitt Public Health

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

Course Director:

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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                        | 40% |
| Midterm exam                               | 25% |
| Final exam                                 | 25% |
| 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. Exams will be in class and open book/ open notes.

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: 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: Friday, 9:00-10:50 AM, A115, Pitt Public Health

| Date            | Topic   | Lecturer | GM, PCG Chapters; Readings are online |
|-----------------|---|----------|---------------------------------------|
|                 | <b><i>Schedule Topics Subject to Change</i></b>   |          |                                       |
| Jan 13          | Introduction to course; history of chromosomes, Chromosome facts and structure, Chromatin organization, Centromere. | QSP      | PCG-1,2 & GM-2                        |
| Jan 20          | Telomeres, Chromosome organization, TADs  | QSP      | PCG-1,2 & GM-2                        |
| Jan 27          | Intro to cell cycle, Mitosis  | QSP      | GM-3, PCG-2, Readings                 |
| Feb. 3          | Meiosis, Recombination, numerical abnormalities   | QSP      | GM-3, PCG-8,9 & GM-5,6                |
| Feb. 10         | Cytogenetic Nomenclature/in-class exercises   | MA       | PCG-9 & GM-5,6                        |
| Feb. 17         | Chromosome abnormalities – Numerical, Rearrangement   | QSP      | PCG-3, ISCN (2020)                    |
| Feb. 24         | Chromosome abnormalities – Structural abnormalities, Contiguous gene deletions                                      | QSP      | PCG-3, ISCN (2020)                    |
| March 3         | <b>Exam 1</b> (covers material through Feb 17)  |          |                                       |
| <b>March 10</b> | <b>Spring Break</b>   |          |                                       |
| March 17        | Sex determination/Dosage compensation and disorders   | SY       | <i>PCG-10 &amp; GM-6</i>              |
| March 24        | Epigenetics/Uniparental disomy/Imprinting -XCI development  | MA       | <i>PCG-10, 20 &amp; GM-6</i>          |

| Date     | Topic  | Lecturer | GM, PCG Chapters; Readings are online |
|----------|--|----------|---------------------------------------|
| March 31 | Fragile X – A family of disorders  | MDF      | <i>PCG-19 &amp; GM-7</i>              |
| April 7  | DNA Repair Defects; Chromosomal Breakage Syndromes; Cancer Predisposition  | PLO      | Readings                              |
| April 14 | Catch-up/Follow-up/Review  | QSP      |                                       |
| April 21 | Cancer Genetics 1. The Basics: Oncogenes/Tumor suppressor genes/ Mismatch Repair Genes/ DNA Repair Genes/ Inherited Cancer Predisposition. | PLM      | Readings                              |
| April 28 | <b>Final Exam</b> (covers material through April 21)   | QSP      |                                       |

**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](http://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-ixcompliance/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.]