

Risk Calculation for Genetic Counseling

Human Genetics 2039

University of Pittsburgh School of Public Health

Summer 2023

1 credit

May 9 syllabus version

Instructor Information

Eleanor Feingold

feingold@pitt.edu

Class Meetings

Thursdays 3:00 – 5:00 with a short break in the middle

A draft schedule is included in this document.

Course Description (from PeopleSoft)

This course provides hands-on training in calculating risk of disease or carrier status in a variety of typical genetic counseling situations, as well as discussion of the limitations of those calculation methods.

Learning Objectives/Competencies

This course provides hands-on training in calculating risk of disease or carrier status in a variety of typical genetic counseling situations, as well as discussion of the limitations of those calculation methods. Detailed (numbered) learning objectives/competencies are below.

Dominant inheritance

Be able to infer Mendelian inheritance patterns and obligate carrier status and calculate risks in a pedigree, considering the following factors.

- 1) various affected and unaffected pedigree members with fixed penetrance
- 2) age-dependent penetrance
- 3) genetic testing results

Comparison of risk calculation paradigms

4) Describe how risk calculation in pedigrees relates to other risk calculation paradigms in genetics, including qualitative information on how each paradigm works and the strengths and weaknesses of each. Quantitatively calculate predictive value of screening tests.

Recessive inheritance

Be able to infer carrier status and calculate Mendelian risks in a pedigree, considering the following factors.

- 5) non-consanguineous pedigrees with various affected and unaffected members and/or genetic testing results
- 6) relatively common disorders with multiple mutations
- 7) consanguinity (with rare or common disease alleles)

X-linked inheritance

Know and/or be able to calculate risks for common pedigree situations such as mother of an isolated case, mother of two cases, sister of an isolated case, etc., and be able to calculate Mendelian risks in a pedigree, considering the following factors in addition to those listed for dominant and recessive inheritance.

- 8) various configurations of affected and unaffected relatives in a 2-generation pedigree
- 9) various configurations in a 3-generation pedigree

Textbook and Course Materials

You may want to have a copy of *Risk Calculation in Genetic Counseling* by Ian Young, 3rd edition (purple cover, available electronically or on paper). It is not required, but some students find it useful to have a reference other than what we do in class. All required course materials will be posted on Canvas.

Course Format and Assignments

For each topic (dominant, recessive, etc.) we will follow a similar format:

- About an hour of lecture (including extensive participation)
- About an hour of (in-class) problem solving practice
- Practice problems to do on your own time. These assignments are not graded. Solutions will be posted, and issues can be discussed in class or in separate meetings with me. Problems will be generally be assigned after each class and “due” (solutions posted) just before the following week’s class.
- Assessment consisting of problems very similar to those done in class and in practice.

Assessment and Grading

The course is “competency-based,” which means that students may re-do assessments as many times as desired to achieve competency. Only the final grade on each assessment will count toward the course grade. Assessments will be closed book, with a calculator and a one-page summary sheet.

Each learning objective on each quiz will be scored as full competency (2 points), partial competency (1 point), or inadequate competency (0 points). Final grades at the end of the course after quizzes are re-taken as many times as desired will be based on the total score for 9 competencies as follows.

A	18
A-	17
B+	16
B	15
Etc.	

Academic Integrity Statement

I encourage you and your fellow students to work together and discuss all practice problems, but the work for assessments must be your own.

Students in this course will be expected to comply with the [University of Pittsburgh’s Policy on Academic Integrity](#). Any student suspected of violating this obligation for any reason during the semester will be required to participate in the procedural process, initiated at the instructor level, as outlined in the University Guidelines on Academic Integrity. This may include, but is not limited to, the confiscation of the examination of any individual suspected of violating University Policy. Furthermore, no student may bring any unauthorized materials to an exam, including dictionaries and programmable calculators.

To learn more about Academic Integrity, visit the [Academic Integrity Guide](#) for an overview of the topic. For hands-on practice, complete the [Understanding and Avoiding Plagiarism tutorial](#).

Accommodation for Students with 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 \(DRS\)](#), 140 William Pitt Union, (412) 648-7890, drsrecep@pitt.edu, (412) 228-5347 for P3 ASL users, as early as possible in the term. DRS will verify your disability and determine reasonable accommodations for this course.

Equity, Diversity, and Inclusion

The University of Pittsburgh does not tolerate any form of discrimination, harassment, or retaliation based on disability, race, color, religion, national origin, ancestry, genetic information, marital status, familial status, sex, age, sexual orientation, veteran status or gender identity or other factors as stated in the University's Title IX policy. The University is committed to taking prompt action to end a hostile environment that interferes with the University's mission. For more information about policies, procedures, and practices, visit the [Civil Rights & Title IX Compliance web page](#).

I ask that everyone in the class strive to help ensure that other members of this class can learn in a supportive and respectful environment. If there are instances of the aforementioned issues, please contact the Title IX Coordinator, by calling 412-648-7860, or e-mailing titleixcoordinator@pitt.edu. Reports can also be [filed online](#). You may also choose to report this to a faculty/staff member; they are required to communicate this to the University's Office of Diversity and Inclusion. If you wish to maintain complete confidentiality, you may also contact the University Counseling Center (412-648-7930).

Sexual Misconduct, Required Reporting, and Title IX

If you are experiencing sexual assault, sexual harassment, domestic violence, and stalking, please report it to me and I will connect you to University resources to support you.

University faculty and staff members are required to report all instances of sexual misconduct, including harassment and sexual violence to the Office of Civil Rights and Title IX. When a report is made, individuals can expect to be contacted by the Title IX Office with information about support resources and options related to safety, accommodations, process, and policy. I encourage you to use the services and resources that may be most helpful to you.

As your professor, I am required to report any incidents of sexual misconduct that are directly reported to me. You can also report directly to Office of Civil Rights and Title IX: 412-648-7860 (M-F; 8:30am-5:00pm) or via the Pitt Concern Connection at: [Make A Report](#)

An important exception to the reporting requirement exists for academic work. Disclosures about sexual misconduct that are shared as a relevant 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 wish to make a confidential report, Pitt encourages you to reach out to these resources:

- The University Counseling Center: 412-648-7930 (8:30 A.M. TO 5 P.M. M-F) and 412-648-7856 (AFTER BUSINESS HOURS)
- Pittsburgh Action Against Rape (community resource): 1-866-363-7273 (24/7)

If you have an immediate safety concern, please contact the University of Pittsburgh Police, 412-624-2121

Any form of sexual harassment or violence will not be excused or tolerated at the University of Pittsburgh.

For additional information, please visit the [full syllabus statement](#) on the Office of Diversity, Equity, and Inclusion webpage.

DRAFT Schedule (5/9/23 version)

Date	Topic
May 18	Dominant inheritance (competencies 1 and 2)
May 25	Genetic testing (competency 3) Other risk calculation paradigms (competency 4)
June 1	Recessive inheritance (competencies 5, 6, 7)
June 8	NO CLASS
June 15	NO CLASS
June 22	NO CLASS
June 29	In-class practice problems to get back into the swing of things
July 6	Assessments (competencies 1 – 7)
July 13	X-linked inheritance (competencies 8, 9)
July 20	Assessments (competencies 8, 9) Repeat assessments (competencies 1 – 7)
July 27	More repeat assessments (optional)
August 3	NO CLASS

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