Risk Calculation for Genetic Counseling
Human Genetics 2039

University of Pittsburgh Graduate School of Public Health
Spring 2020
1 credit
December 31, 2019 syllabus version

Instructor Information
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Class Meetings
Fridays 1:30 – 3:25, room 3121C
Class will not meet every week. Total contact hours for the semester will be approximately the 15 hours expected for a 1-credit course. A draft schedule is included in this document, but some changes may be necessary. Weeks marked "no class" will not change, however, and you should feel free to schedule other things during those times.

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) For each of the following, be able to discuss what they are, how they work, and strengths and weaknesses.

- Screening tests (including calculation of predictive value)
- Epidemiological models for risk prediction (Gail, Klaus, etc.)
- Risk prediction from sequencing data

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

5) non-consanguinous pedigrees with various affected and unaffected members
6) relatively common disorders with multiple mutations

7) genetic testing results
8) 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.

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

Calculations using linked markers

Be able to calculate risks based on linked markers for the following situations.

11) dominant disease with phase known (3-generation pedigree)
12) dominant disease with phase unknown (2-generation pedigree)
• recessive disease and x-linked disease will be discussed but not tested

Course Format

For each learning objective or set of learning objectives there will be a lecture (including extensive hands-on problem solving) to demonstrate the calculation methods, a homework set to practice solving problems, and a quiz consisting of problems very similar to those on the homework. The course is "competency-based," which means that students may re-take quizzes as many times as desired to achieve competency. Only the final grade on each quiz will count toward the course grade. Homeworks must be turned in, but will not be graded for correctness, only completion. Solutions will be posted on courseweb and discussed in class. Quizzes require a calculator and you may also use a one-page "cheat sheet" of notes.

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 courseweb.

Grading

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 12 competencies as follows.

A	24 or 23
A-	22
B+	21
B	20

Etc.

Academic Integrity Statement
I encourage you and your fellow students to work together and discuss all assignments (indeed that is a requirement of the course), but any final work you submit must be original and your own. All students are expected to adhere to the school's standards of academic honesty. 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.

Accommodation for Students with Disabilities
If you have any disability for which you may require accommodation, you are encouraged to notify both me and the Office of Disability Resources and Services (DRS), 140 William Pitt Union (voice or TTD 412-648-7890 or at www.studentaffairs.ptt.edu/drs, or by email at drsrecep@pitt.edu as early as possible in the term.

Diversity & Academic Civility
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).

Sexual Misconduct, Required Reporting, and Title IX

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

DRAFT Schedule (12/30/19 version).

Date	Topic	homework
January 10	**No class**	
January 17	Course intro Dominant inheritance (objectives 1 and 2)	
January 24	Genetic testing (objective 3) Other risk calculation paradigms (objective 4) Dominant practice problems	
January 31	**Possibly no class**	dominant homework due (objectives 1, 2, 3, 4)
February 7	Recessive inheritance (objectives 5, 6, 7, 8) recessive practice problems	
February 14	**No class**	recessive homework due (objectives 5, 6, 7, 8)
February 21	Quizzes on competencies 1 - 8	
February 28	X-linked lecture (objectives 9, 10) X-linked practice problems	
March 6	**No class**	X-linked homework due (objectives 9, 10)
March 13	**Spring break**	
March 20	Linked marker lecture (objectives 11, 12) Linked marker problems	
March 27	Repeat quizzes on competencies 1 - 8	linked marker homework due (objectives 11, 12)
April 3	Quizzes on competencies 9 - 12	
April 10	**Repeat quizzes**	
April 17	**Repeat quizzes**	
April 24	**No class**	

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