Graduate School of Public Health
Department of Human Genetics
PRINCIPLES OF GENETIC COUNSELING
HUGEN 2035
Monday and Wednesday 2:00-3:25PM
3121C Public Health
Credit Hours: 3.0
Fall Semester 2022

Instructors and Contact Information

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Goal of the Course

The goal of this course is to provide you with a foundation in genetic counseling principles. We will expand upon these principles in HUGEN 2038—Intervention Skills in Genetic Counseling and HUGEN 2061—Cancer Genetic Counseling (Spring semester) in preparation for your clinical rotations.

Course Description

The purpose of this course is to provide you with the knowledge necessary to be an effective genetic counselor. Prior to entering your clinical rotations, you must learn and understand the genetic information commonly addressed in genetic counseling sessions. This course, and many of the other courses taught during the first two semesters, provide you with this information.

The first part of the course will address fundamental concepts important to genetic counseling including inheritance patterns, risk assessment, and pedigree analysis. The course will then address several topics important to pediatric genetic counseling and testing including childhood development, dysmorphology, metabolic conditions, and newborn screening.

In addition to pediatric genetic counseling, the course will cover adult genetic counseling topics and subspecialty areas such as cardiovascular genetics and neurogenetics. Throughout the course, several families will provide you with their perspectives on living with a genetic condition. To be an effective genetic counselor, you must try to understand the perspectives and needs of families coping with a genetic condition.

Course Learning Objectives

The learning objectives are grouped into three conceptual areas and describe what you will be able to do after completion of the course:

Genetic Counseling Skills

- Construct a complete pedigree and demonstrate proficiency in the use of pedigree symbols, standard notation, and nomenclature

- Identify inheritance patterns and apply Mendelian genetics and Bayesian analysis to conduct risk assessment
- Describe genetic/genomic screening and testing approaches and their application to genetic counseling practice
- Practice communicating key elements of genetic concepts based on the needs of the audience
- Identify, create, and present relevant and appropriate resources that play an important role in providing genetic counselors, other health care providers, and patients with up-to-date genetic information

Genetic Counseling Practice in Different Settings

- Explain normal development and the process of assessment for abnormal physical/mental development in a pediatric genetic counseling setting
- Recognize various categories of metabolic conditions, neurogenetic conditions, and cardiovascular genetic conditions and describe their genetic etiology, as well as applicable screening, testing, and management/treatment modalities
- Describe newborn screening, its public health significance, and the genetic counselor's role in the newborn screening process. Recognize the criteria for inclusion of conditions on the Recommended Uniform Screening Panel (RUSP), and challenges encountered in population screening

General Genetic Counseling Knowledge

- Describe the history of the genetic counseling profession and its impact on the current state of the profession
- Recognize families' experience with genetic disorders
- Identify aspects of practice related to the delivery of genetic counseling services in different settings and the ways in which legal, social, and cultural factors influence the provision of services

Required Texts

A Guide to Genetic Counseling, 2nd edition. WR Uhlmann ,JL Schuette, and BM Yashar (eds.). Wiley-Blackwell, 2009.

Human Genetics and Genomics. 4th edition. BR Korf. Blackwell Science, Inc., 2013.

Highly Recommended Texts

BS LeRoy, P McCarthy Veach, NP Callanan. *Genetic Counseling Practice, Advanced Concepts and Skills, 2nd Ed.* (2021) Wiley Blackwell.

P McCarthy Veach, BS LeRoy, NP Callanan. (2018). *Facilitating the Genetic Counseling Process, 2nd Ed.* Springer.

Course Format and Delivery

This class utilizes Canvas for class content and evaluation. Notifications will be sent regarding course content throughout the semester. Please make sure to set up your Canvas to receive email notifications from Canvas and please check for announcements and notifications on a regular basis. Should you need to communicate with the course instructor(s), please email them at their Pitt email addresses.

It is expected that class will take place in person. However, due to the shifting nature of the pandemic, it may be necessary to alter the delivery of the course. Any changes will be shared via announcements on

Canvas. Given the pandemic, it is important that students abide by public health regulations and University of Pittsburgh health standards and guidelines. For the most up-to-date information and guidance, please visit https://www.coronavirus.pitt.edu/. If you are sick, please do not come to class in-person. Please email the course instructors, and we will determine the most appropriate make-up plan for class.

We intend to record most classes to make them available on Canvas. It is possible that some guest lecturers may prefer to not be recorded and we will communicate this information during the semester. Recordings will only be available for the semester and should not be distributed for non-class purposes.

Office Hours

Office hours will take place on Mondays from 3:25 to 3:50 in room 3121C. Should class need to take place remotely due to changes in the COVID pandemic, office hours will take place at the same time via Zoom. The course instructors are available to meet at other times by appointment.

Course Grading

The goal of the course is to provide you with skills and knowledge that are applicable to your future career as a genetic counselor. The assignments and activities are designed to help students achieve the course learning objectives. The course structure and grading are based on adult learning theory (andragogy) which suggests that adults learn best in a flexible, interesting, and challenging environment. The genetic counseling topics covered in the course are interesting and challenging. The course grading is flexible and allows students to pursue assignments in ways that are customized to their needs.

With the exception of the two exams, the risk/inheritance problem set, and genetic testing cases, all assignments for the class are graded as either satisfactory or unsatisfactory and have a rubric that describe the requirements for satisfactory completion. To receive a satisfactory grade on the risk/inheritance problem set and genetic testing cases, a student must get 70% of the questions correct. The midterm and final exam are graded using a 100-point system. The table below outlines the course components that must be completed to earn a particular letter grade. Students can decide how much to do to achieve the grade they desire. The course learning objectives are addressed at each level but students who earn higher grades demonstrate greater mastery of the learning objectives.

Course Component	Requirements for Letter Grade of A (A bundle)	Requirements for Letter Grade of B (B bundle)	Requirements for Letter Grade of C (C bundle)
Absences	1 unexcused absence	2 unexcused absences	3 unexcused absences
Discussion Board - Original Post	1 satisfactory original discussion post	1 satisfactory original discussion post	1 satisfactory original discussion post
Discussion Board - Response Post	4 satisfactory responses to discussion posts	3 satisfactory responses to discussion posts	2 satisfactory responses to discussion posts
Group Project	Satisfactory group project evaluation Creation of a satisfactory condition-specific engagement activity Creation of a satisfactory condition-specific study guide	Satisfactory group project evaluation Creation of a satisfactory condition-specific engagement activity	Satisfactory group project evaluation Creation of a satisfactory condition-specific engagement activity
Risk/Inheritance	Satisfactory (≥70%)	Satisfactory (≥70%)	Satisfactory (≥70%)

Problem Set	completion of a 15 question problem set	completion of a 12 question problem set	completion of a 10 question problem set
Pedigree Exemplar	Satisfactory completion of the Pedigree Exemplar Satisfactory completion of the Pedigree Reflection-3 pedigrees	Satisfactory completion of the Pedigree Exemplar Satisfactory completion of the Pedigree Reflection-1 pedigree	Satisfactory completion of the Pedigree Exemplar
Genetic Concept Explanation	Satisfactory completion of the narrative component of the Genetic Concept Explanation assignment Satisfactory completion of a video recording of verbal explanation of two genetic concepts	Satisfactory completion of the narrative component of the Genetic Concept Explanation assignment Satisfactory completion of a video recording of verbal explanation of one genetic concept	Satisfactory completion of the narrative component of the Genetic Concept Explanation assignment
Genetic Testing Cases	Satisfactory (≥70%) completion of cases #1-10	Satisfactory (≥70%) completion of cases #1-8	Satisfactory (≥70%) completion of a cases #1-6
Exams (midterm and final)	≥90% average	≥80% average	≥70% average

Each grade bundle will be further delineated based on the combined average of the midterm and final exam. More specifically, within the A bundle, students who earn 97-100% will receive an A+, those who earn 93-96% will receive an A, and those who earn 90-92% will receive an A-. For the B bundle, students who earn 87-89% will receive a B+, those who earn 83-86% will receive a B grade, and those who earn 80-82% will receive a B-. For the C bundle, students who earn 77-79% will receive a C+, those who earn 73-76% will receive a C grade, and those who earn 70-72% will receive a C-. Students who do not meet the requirements for a C grade will receive a F grade.

Assignments

All assignments should be turned in on Canvas unless otherwise instructed by the due dates indicated in Canvas. *All file names should follow the format LASTNAME.ASSIGNMENT (i.e. Vento.GeneticTestingCases.doc). When possible, documents should be in Word or PowerPoint format (it is harder to add comments in pdf documents)*. A brief description of the assignments is provided below but please see Canvas for more detailed information.

Discussion Board

Learning Objectives:

Since the discussion board will be used throughout the entire semester, it addresses all learning objectives for the course.

The course will utilize a discussion board to further facilitate class discussion. This will provide an opportunity for thoughtful reflection about assigned readings and course material. There will be 12 discussion board topics throughout the semester, and each week will have an assigned student to start the discussion. You will create an original post that is based on the readings/other materials (see Canvas for your assigned week and further details). You will also respond to a certain number of additional posts on the weeks in which you are not assigned as the original poster. The number of responses depends on the grade

bundle you are trying to achieve. The original post is due before the corresponding class and the responses need to be completed by midnight on the Saturday after class.

Risk Analysis & Inheritance Pattern Problem Set
Learning Objectives:

- Construct a complete pedigree and demonstrate proficiency in the use of pedigree symbols, standard notation, and nomenclature
- Identify inheritance patterns and apply Mendelian genetics and Bayesian analysis to conduct risk assessment

You will be assigned a problem set on risk assessment and inheritance patterns where you will utilize advanced knowledge of inheritance patterns, basic Bayesian statistics, coefficient of relationship and coefficient of inbreeding. This problem set will help you to practice the risk analysis content presented in class and to prepare you for calculating risks during your clinic rotations. For the A grade bundle, students need to complete 15 questions, for the B grade bundle, students need to complete 12 questions and for the C grade bundle, students need to complete 10 questions.

Pedigree Exemplar and Reflection
Learning Objectives:

- Construct a complete pedigree and demonstrate proficiency in the use of pedigree symbols, standard notation, and nomenclature

Pedigree exemplar:
You will be provided with a family history narrative and will draw a pedigree using standardized nomenclature based on this family history.

Pedigree reflection:
For students who are striving for an A grade or B grade course bundle you will complete a pedigree reflection. For the reflection, you will verbally take and draw one or more family pedigrees (you can ask family members, friends, your fellow classmates – please make sure everyone is over 18). Students working towards the A grade bundle are required to collect and draw three pedigrees for this assignment and students working towards the B grade bundle are required to collect and draw one pedigree for this assignment. The pedigree(s) do not need to be turned in. Rather, submit a one-page document to summarize your experiences that includes the following for each pedigree: a) The length of time that it took you to take the pedigree, b) Two things that went well, c) Two things you would like to improve upon, and d) Any questions that arose from taking the pedigree that you would like answered. If any of the individuals you work with to take the pedigree do not wish to reveal personal medical information, they may fictionalize their health history. This assignment will begin to give you an understanding of the skills necessary for eliciting a family history.

Genetic Concept Explanation
Learning Objectives:

- Practice communicating key elements of genetic concepts based on the needs of the audience
- Identify, create, and present relevant and appropriate resources that play an important role in providing genetic counselors, other health care providers, and patients with up-to-date genetic information
- Explain normal development and the process of assessment for abnormal physical/mental development in a pediatric genetic counseling setting

You will write an explanation of genetic concepts and inheritance patterns as if you are speaking with a patient who has limited health and genetic literacy. Based on the grade bundle you will strive for, you may also choose to video record 1-2 explanations. The goal is for your explanations to be straightforward and complete. Describing complex medical/genetic information to patients in understandable ways is one of the more difficult tasks a counselor faces in their daily work. This assignment will help you develop the skills necessary to perform this task successfully.

Genetic Testing Cases
Learning Objectives:

- Describe genetic/genomic screening and testing approaches and their application to genetic counseling practice
- Recognize various categories of metabolic conditions, neurogenetic conditions, and cardiovascular genetic conditions and describe their genetic etiology, as well as applicable screening, testing, and management/treatment modalities

You will be assigned a problem set with various clinical genetic testing cases. The purpose of this assignment is to practice applying learned concepts about metabolic and molecular testing methodologies to clinical cases. For the A grade bundle, students need to complete 10 questions, for the B grade bundle, students need to complete 8 questions and for the C grade bundle, students need to complete 6 questions.

Group Project
Learning Objectives:

- Describe genetic/genomic screening and testing approaches and their application to genetic counseling practice
- Practice communicating key elements of genetic concepts based on the needs of the audience
- Identify, create, and present relevant and appropriate resources that play an important role in providing genetic counselors, other health care providers, and patients with up-to-date genetic information
- Recognize families' experience with genetic disorders
- Identify aspects of practice related to the delivery of genetic counseling services in different settings and the ways in which legal, social, and cultural factors influence the provision of services

You will be assigned to a group and each group is required to research a genetic condition in a collaborative manner. You and your group will give a 20-minute presentation on the assigned genetic condition to the class. For your presentation, include information that your group believes is important to research as part of preparation for a clinical case. For example, prior to counseling a patient it would be important to be familiar with the features of the condition, its inheritance pattern, management, and treatment, as well as other important aspects. This assignment is intended to help you critically think about relevant areas to research in preparation for a case.

After your presentation, there will be a 5-minute question and answer session. You need to distribute your slides to the class during the class period before the group presentation day so that your classmates have time to develop possible questions. Each student is expected to come prepared with two questions regarding each condition for the question and answer session.

- **The three group presentations will take place on day one.**

Each group will also present an engagement activity on day two of group presentations. This activity should be approximately 15 minutes in length and its purpose is to engage the audience and reinforce the key concepts from your presentation.

- **All three groups will present their engagement activity on day two of group presentations.**

For those who want to achieve the A grade bundle, the last element of the project is a condition-specific study guide. This guide should be a complete, yet concise, summary of the main aspects of the assigned genetic condition and is intended to be used to study for course exams, the comprehensive exam that is taken during the second year of the program, and the ABGC certification exam. This document is due the day of the group presentation and should be one page in length.

There are two types of evaluation for this project including an intragroup and intergroup evaluation.

Please see the group project instructions on Canvas for further information.

Midterm Exam and Final Exam
Learning Objectives:
The exams cover all learning objectives.

Two exams, each worth 100 points, will be administered during the semester. The midterm exam will be given approximately halfway through the semester and will cover course content addressed to that point. The final exam will be a take home exam and is cumulative covering all course content but focusing primarily on course content addressed during the second half of the semester.

The exams will be delivered via Canvas. The midterm will take place in the classroom and you will need a laptop to take the exam (please let the course instructor(s) know if you do not have access to a laptop). For the midterm there is an opportunity to earn two points for a question that you did not earn full credit with the use of a token. To earn these two points, you need to submit a narrative explanation (approximately one paragraph) of what you missed on the question and what you learned from reexamining the question. *To earn these two points, this short narrative explanation needs to be submitted within one week of receiving your graded exam.*

The final exam will be a take home and open book exam. For the final, you will be able to access the exam during a certain period of time via Canvas.

Tokens
At the start of the semester, each student will have three tokens that can be used in the following ways:

- Resubmit work that is unsatisfactory. The assignment must be resubmitted within one week of receiving the assignment evaluation.
- To extend the deadline of an assignment. To use this token, the student must contact the instructor ahead of the deadline and both the student and instructor must agree upon the new date for the assignment.
- One token can be used to miss a class.
- One token can be used to earn two points on the midterm. To use this token, you need to submit a narrative explanation (approximately one paragraph) of what you missed on the question and what you learned from reexamining the question within one week of receiving your graded exam.

Late Work Policy

Each assignment has a corresponding due date. Should an assignment be turned in late, then it will receive an unsatisfactory grade. As noted in the "Tokens" section, students can use a token to extend the deadline of an assignment if the student contacts the instructor(s) prior to the deadline. Should a student encounter an emergency or extenuating circumstance, then they should contact the instructor(s) as soon as possible, prior to the deadline, when possible, to discuss a possible extension. This will be evaluated on a case-by-case basis.

The course instructor(s) recognize that the semester can get busy at certain times and assignment deadlines may conflict with due dates for other classes. We are willing to consider an adjustment to an assignment deadline for the entire class when such concerns are brought to our attention and when they impact the majority of the class.

Absence/Participation

Each class covers critical material related to the practice of genetic counseling. Therefore, your attendance and participation are important to your professional development and to the success of the course. We expect that each student will read the assigned materials and participate in class discussion. As noted above, unexcused absences are part of the grading structure for the course. Missing class due to illness or an unanticipated emergency are considered excused absences. Please notify the course instructor(s) about a missed class and a make-up plan can be determined.

Your Wellbeing Matters

Graduate school can be an exciting and challenging time for students. Taking time to maintain your well-being and seek appropriate support can help you achieve your goals and lead a fulfilling life. It can be helpful to remember that we all benefit from assistance and guidance at times, and there are many resources available to support your well-being while you are at Pitt. You are encouraged to visit Thrive@Pitt to learn more about well-being and the many campus resources available to help you thrive.

If you or anyone you know experiences overwhelming academic stress, persistent difficult feelings and/or challenging life events, you are strongly encouraged to seek support. In addition to reaching out to friends and loved ones, consider connecting with a faculty member you trust for assistance connecting to helpful resources.

The University Counseling Center is also here for you. You can call 412-648-7930 at any time to connect with a clinician. If you or someone you know is feeling suicidal, please call the University Counseling Center at any time at 412-648-7930. You can also contact Resolve Crisis Network at 888-796-8226. If the situation is life threatening, call Pitt Police at 412-624-2121 or dial 911.

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.

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(s) and Disability Resources and Services, 140 William Pitt Union, (412) 648-7890, (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.

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 professors, we are required to report any incidents of sexual misconduct that are directly reported to us, or of which we are 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/make-report/report-form

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.]

Diversity Statement
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 and promote social justice. 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 director or course instructors;
- the Pitt Public Health Associate Dean responsible 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/make-report/report-form (anonymous reporting form)

Copyright Notice

Course material may be protected by copyright. United States copyright law, 14 USC section 101, et sec., in addition to University policy and procedures, prohibit unauthorized duplication or retransmission of course materials. See Library of Congress Copyright Office and the University Copyright Policy.

Schedule of Sessions

Date	Class Topic	Lecturer(s)
Monday, 8/29	Introduction to genetic counseling – definition and historical perspective	Robin Grubs, MS, PhD Jodie Vento, MGC
Wednesday, 8/31	From Mendelian genetics to complex disease, Part 1	Robin Grubs, MS, PhD
Monday, 9/5	No Class—Labor Day	
Wednesday, 9/7	From Mendelian genetics to complex disease, Part 2	Robin Grubs, MS, PhD Kara Levine, MS
Monday, 9/12	Pedigree analysis I	Jodie Vento, MGC
Wednesday, 9/14	Introduction to risk assessment and Bayesian statistics	Nadene Henderson, MS
Monday, 9/19	Pedigree analysis II	Jodie Vento, MGC
Wednesday, 9/21	An approach to a child with dysmorphology	Damara Ortiz, MD
Monday, 9/26	Normal childhood development & an approach to a child with intellectual disability	Laura Jenkins, MS Evgenia Sklirou, MD
Wednesday, 9/28	An approach to genetic/genomic testing in a pediatric setting	Charlotte Skinner, MS, MPH
Monday, 10/3	Newborn screening	Cate Walsh Vockley, MS
Wednesday, 10/5	An approach to a child with a metabolic condition-I	Sarah Drewes, MS
Monday, 10/10	An approach to a child with a metabolic condition-II	Cate Walsh Vockley, MS

Wednesday, 10/12	An approach to a child with a metabolic condition-III	Nadene Henderson, MS
Monday, 10/17	An approach to a child with Down syndrome	Kishore Vellody, MD
Wednesday, 10/19	Families with Down syndrome	Erin Kelly and Family Panel
Monday, 10/24	Families with Duchenne muscular dystrophy	Terri Ellsworth
Wednesday, 10/26	**Midterm Exam**	
Monday, 10/31	Pediatric neurogenetics-mitochondrial disorders	Jodie Vento, MGC
Wednesday, 11/2	Group Projects (Presentations)	Students
Monday, 11/7	Group Projects (Engagement Activities)	Students
Wednesday, 11/9	Adult neurogenetics	Abby Peffer, MS Chris Munro, MS, MPH
Monday, 11/14	Cardiovascular genetics	Emily James, MS
Wednesday, 11/16	No Class NSGC	
Monday, 11/21 & Wednesday, 11/23	No Class Thanksgiving Break	
Monday, 11/28	GINA, HIPAA, and the ADA alternative service delivery models	Andrea Durst, MS, DrPH -
Wednesday, 11/30	No Class Human Genetics Retreat	
Monday, 12/5	Communicating with patients	Robin Grubs, MS, PhD
Wednesday, 12/7	Palliative care and early intervention	Robin Grubs, MS, PhD Jodie Vento, MGC
Monday, 12/12	Working with diverse populations	Caitlin Lavin, MS
Wednesday, 12/14	Families with HD	Tammy Makoul, MSW
	Final Exam – Take home exam	Take home on 12/13 and due 12/15