

**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 2021**

Instructors and Contact Information

Robin E. Grubs, PhD, LCGC (she/her)

Office: 3138 Public Health

Phone: 412-624-4695 (work)

412-260-7460 (cell)

Email: rgrubs@pitt.edu

Office hours: By appointment

Jodie M. Vento, MGC, CGC (she/her)

Office: 3136 Public Health

Phone: 412-624-3136 (work)

412-913-1932 (cell)

Email: ventojm@pitt.edu

Office hours: By appointment

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, dysmorphism, 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.

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 grading. 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.

For the first two weeks of the semester, students will have the option of attending class in person or using Zoom for remote course access. It is expected that after this two-week period, 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 with students 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 example, at this time, face coverings are required indoors for everyone regardless of vaccination status. 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 depending on the content.

We plan on recording most classes to make them available on Canvas. We recognize 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.

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](#).

Student Performance Evaluation

Your grade for the course is based upon the grades you receive on the assignments, midterm exam, and final exam. Each exam is 100 points and the total number of points for the assignments is 177. Therefore, there are a total of 377 points. Since you are not graded on a curve, you will not be competing with one another for a grade. Therefore, we encourage you to help each other achieve the best work you are capable of producing. Working with one another to strive for greater understanding will help you learn the material with more ease and enjoyment.

Grading Scale

97-100%	A+	73-76%	C
93-96%	A	70-72%	C-
90-92%	A-	67-69%	D+
87-89%	B+	63-66%	D
83-86%	B	60-62%	D-
80-82%	B-	<60	F
77-79%	C+		

Course Learning Objectives

After completion of this course, the student will be able to:

1. Describe the history of the genetic counseling profession and utilize skills important to the current practice of genetic counseling.
2. 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.
3. Construct a complete pedigree and demonstrate proficiency in the use of pedigree symbols, standard notation, and nomenclature.
4. Recognize the ways in which some families cope with genetic disorders.
5. Explain normal development and the process of assessment for abnormal physical/mental

development in a pediatric genetic counseling setting.

6. Describe genetic/genomic screening and testing approaches and their application to genetic counseling practice.
7. Recognize aspects 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.

Assignments

All assignments should be turned in on Canvas unless otherwise instructed by the due dates indicated in the class schedule. Should a due date be problematic, please let us know as there is some flexibility in the scheduling of the due dates. All file names should follow the format LASTNAME.ASSIGNMENT (i.e. Vento.Definitions.doc). When possible, documents should be in Word or PowerPoint format (it is harder to add comments in pdf documents).

Discussion Board 15 points

Learning Objective 1: Describe the history of the genetic counseling profession and utilize skills important to the current practice of genetic counseling.

Learning Objective 2: 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.

Learning Objective 3: Construct a complete pedigree and demonstrate proficiency in the use of pedigree symbols, standard notation, and nomenclature.

Learning Objective 4: Recognize the ways in which some families cope with genetic disorders.

Learning Objective 5: Explain normal development and the process of assessment for abnormal physical/mental development in a pediatric genetic counseling setting.

Learning Objective 6: Describe genetic/genomic screening and testing approaches and their application to genetic counseling practice.

Learning Objective 7: Recognize aspects 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.

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 at least four additional posts on the weeks in which you are not assigned as the original poster. Therefore, you will have a total of five posts (one original post & four responses). The original

post is due before the corresponding class and the responses need to be completed by midnight on the Saturday after class. Each post is worth 3 points. Since the discussion board will be used throughout the entire semester, it addresses all learning objectives for the course.

Risk Analysis & Inheritance Pattern Problem Set **20 points**

Learning Objective 1: Describe the history of the genetic counseling profession and utilize skills important to the current practice of genetic counseling.

You will be assigned a 10-question problem set on risk assessment and inheritance patterns where you will utilize advanced knowledge of inheritance patterns in families as well as 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.

Pedigree Assignment **15 points**

Learning Objective 3: Construct a complete pedigree and demonstrate proficiency in the use of pedigree symbols, standard notation, and nomenclature.

You will draw your family pedigree including three generations. You will turn this in for assessment of the correct use of pedigree symbols and inclusion of all pertinent components of a medical family pedigree. You will verbally take and draw three additional family pedigrees (you can ask family members, friends, your fellow classmates – please make sure everyone is over 18). These three pedigrees 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 you or any of the individuals you work with to take the three additional pedigrees do not wish to reveal personal medical information, you/they may fictionalize your/their health history. This assignment will begin to give you an understanding of the skills necessary for eliciting a family history.

Definition Assignment **50 points**

Learning Objective 1: Describe the history of the genetic counseling profession and utilize skills important to the current practice of genetic counseling.

Learning Objective 2: 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.

Learning Objective 5: Explain normal development and the process of assessment for abnormal physical/mental development in a pediatric genetic counseling setting.

Once we have reviewed all the types of inheritance patterns, you will write a paragraph describing each inheritance pattern as you would describe them to a patient who has a high school degree and who has a very limited knowledge of genetics. You will also be given a list of genetic terms to describe in the same manner. One of the most difficult tasks a counselor faces is describing complex medical/genetic information to patients in understandable ways. These exercises will help you develop the skills necessary to perform this task successfully.

Group Projects

52 points

Learning Objective 2: 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.

Learning Objective 4: Recognize the ways in which some families cope with genetic disorders.

Learning Objective 6: Describe genetic/genomic screening and testing approaches and their application to genetic counseling practice.

Learning Objective 7: Recognize aspects 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.

Presentation and Interactive Activity:

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 30-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 10-minute question and answer session. You need to distribute your slides to the class during the class period before your presentation 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.

- **Two groups will give their presentations on day one of group presentations and the third group will present on day two.** Remember that your slides are due at least one class period prior to your presentation date. These should be uploaded to the corresponding Canvas assignment.
- **All three groups will present their interactive activity on day two of group presentations during the remaining 40 minutes of class. Therefore, each group will**

have about 10 minutes (with a couple of minutes for set up).

Evaluations: There will be two types of evaluation for this project including an intragroup and intergroup evaluation. Please see the group project instructions for further information.

**Fact Sheet Assignment
25 points**

Learning Objective 2: 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.

Learning Objective 7: Recognize aspects 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 write a fact sheet on a genetic condition or topic that will be assigned to you early in the semester. When developing your fact sheet, you should focus on providing information in a patient friendly format that explains the condition or topic in a way that can be understood by the general public. You should utilize multiple resources to gather information to include in your fact sheet. A list of commonly used resources in genetic counseling will be provided, and this assignment will help you become familiar with them.

Schedule of Sessions

Date	Class Topic	Lecturer(s)
Monday, 8/30	Introduction to genetic counseling – definition and historical perspective	Robin Grubs, MS, PhD Jodie Vento, MGC
Wednesday, 9/1	From Mendelian genetics to complex disease: An overview	Robin Grubs, MS, PhD Kara Levine, MS
Monday, 9/6	No Class—Labor Day	
Wednesday, 9/8	Pedigree analysis I	Jodie Vento, MGC
Monday, 9/13	Pedigree analysis II	Jodie Vento, MGC
Wednesday, 9/15	Introduction to risk assessment and Bayesian statistics	Nadene Henderson, MS
Monday, 9/20	Normal childhood development & an approach to a child with intellectual disability	Laura Jenkins, MS Evgenia Sklirou, MD
	Pedigree Assignment Due 9/20 by 5pm	
Wednesday, 9/22	No Class—NSGC Conference	

Monday, 9/27	Newborn screening	Cate Walsh Vockley, MS
Wednesday, 9/29	An approach to genetic/genomic testing in a pediatric setting	Charlotte Skinner, MS, MPH
	Risk Analysis & Inheritance Pattern Problem Set Due 9/29 by 5pm	
Monday, 10/4	An approach to a child with a metabolic condition-I	Sarah Drewes, MS
Wednesday, 10/6	An approach to a child with a metabolic condition-II	Cate Walsh Vockley, MS
Monday, 10/11	An approach to a child with a metabolic condition-III	Nadene Henderson, MS
	Definition Assignment Due 10/11 by 5pm	
Wednesday, 10/13	An approach to a child with dysmorphology	Damara Ortiz, MD
Monday, 10/18	An approach to a child with Down syndrome	Kishore Vellody, MD
Wednesday, 10/20	Families with Down syndrome	Erin Kelly and Family Panel
Monday, 10/25	Families with Duchenne muscular dystrophy	Terri Ellsworth
Wednesday, 10/27	Midterm Exam	
Monday, 11/1	Pediatric neurogenetics-mitochondrial disorders	Jodie Vento, MGC
Wednesday, 11/3	Group Projects (Groups 1 and 2)	Students
Monday, 11/8	Group Projects (Group 3 and review activities)	Students
Wednesday, 11/10	Pediatric cardiovascular genetics	Jessica Sebastian, MS
Monday, 11/15	Adult cardiovascular genetics	Emily James, MS
Wednesday, 11/17	Adult neurogenetics	Abby Pepper, MS Chris Munro, MS, MPH
Monday, 11/22 and Wednesday, 11/24	No Class—Thanksgiving Break	
Monday, 11/29	GINA, HIPAA, and the ADA alternative service delivery models	Andrea Durst, MS, DrPH

Wednesday, 12/1	Communicating with patients	Robin Grubs, MS, PhD
Monday, 12/6	Palliative care and early intervention	Robin Grubs, MS, PhD Jodie Vento, MGC
	Fact Sheet Assignment Due 12/6 by 5pm	
Wednesday, 12/8	Working with diverse populations	Peggy Cottrell, MS, Caitlin Lavin, MS, and Emily Lancaster, MS
Monday, 12/13	Families with HD	Tammy Makoul, MSW
Wednesday, 12/15	Final Exam – Take home exam	