

**Graduate School of Public Health  
Department of Human Genetics  
PRINCIPLES OF GENETIC COUNSELING  
HUGEN 2035  
Monday and Wednesday 2:00-3:25PM  
A425 Public Health  
Credit Hours: 3.0  
Fall Semester 2019**

**Instructors and Contact Information:**

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**Course Description:**

This course addresses fundamental concepts important to genetic counseling principles and practice.

We will expand upon these principles in Human Genetics 2038- Intervention Skills (Spring semester) in preparation for your clinical rotations.

**Goal of the Course:**

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 this course will address fundamental concepts important to genetic counseling including inheritance patterns, risk assessment, taking a family history, and a review of normal childhood development. The course will then address several topics important to pediatric genetic counseling and testing including dysmorphology, metabolic conditions, newborn screening, variant interpretation and whole exome sequencing.

The second part of the course will cover a variety of sub-specialty areas of genetic counseling. We will review psychiatric, cardiovascular, and coagulation disorders in addition to teratology, and ethnocultural issues in genetic counseling. The last part of the course will focus on prenatal genetics including prenatal diagnosis, ultrasonography and maternal serum screening. In the first part of Human Genetics 2038, you will be assigned terms covered in these prenatal lectures. Throughout the course, several families will provide you with their perspectives on coping with some of the genetic conditions we will discuss. 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, 2<sup>nd</sup> edition. WR Uhlmann ,JL Schuette, and BM Yashar (eds.). Wiley-Blackwell, 2009.

Human Genetics and Genomics. 4<sup>th</sup> edition. BR Korf. Blackwell Science, Inc., 2013.

**Highly Recommended Texts:**

Genetic Counseling Practice: Advanced Concepts and Skills by Bonnie S. LeRoy, Patricia M. Veach, and Dianne M. Bartels. Wiley-Blackwell. 2010

Facilitating the Genetic Counseling Process. P McCarthy Veach, BS LeRoy, D Bartels. Springer, 2003.

**CourseWeb/BlackBoard Instruction:**

This class utilizes CourseWeb for class content and grading. Notifications will be sent when course content is updated.

**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.

**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, 140 William Pitt Union, 412-648-7890 as early as possible in the term.

A comprehensive description of the services of that office can be obtained at [www.drs.pitt.edu](http://www.drs.pitt.edu).

**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:

[www.titleix.pitt.edu/report/confidentiality](http://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](http://www.titleix.pitt.edu/report-0)

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. 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](mailto: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).

### **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 207. Therefore, there are a total of 407 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+
93-96%	A
90-92%	A-
87-89%	B+
83-86%	B
80-82%	B-
77-79%	C+
73-76%	C
70-72%	C-
67-69%	D+
63-66%	D
60-62%	D-
<60	F

**Schedule of Sessions:**

<b>Date</b>	<b>Class Topic</b>	<b>Lecturer(s)</b>
Monday, 8/26	Introduction to genetic counseling – definition and historical perspective	Robin Grubs, MS, PhD Andrea Durst, MS, DrPH
Wednesday, 8/28	From Mendelian Genetics to Complex Disease: An Overview	Robin Grubs, MS, PhD Kara Levine, MS
Monday, 9/2	<b>No Class – Labor Day</b>	
Wednesday, 9/4	Pedigree Analysis	Andrea Durst, MS, DrPH
Monday, 9/9	Pedigree Analysis II	Andrea Durst, MS, DrPH
Wednesday, 9/11	Introduction to risk assessment and Bayesian statistics	Nadene Henderson, MS
Monday, 9/16	Providing Information about Genetic Conditions to Patients	Robin Grubs, MS, PhD Andrea Durst, MS, DrPH
Wednesday, 9/18	Ethnocultural Genetic Counseling	Andrea Durst, MS, DrPH
<b>Risk Analysis &amp; Inheritance Pattern Problem Set Due 9/18 by 5pm</b>		
Monday, 9/23	Families with Duchenne Muscular Dystrophy	Terri Ellsworth, Billy Ellsworth
Wednesday, 9/25	Normal Childhood Development & an Approach to a Child with Intellectual Disability	Laura Jenkins, MS Evgenia Sklirou, MD
Monday, 9/30	An approach to a Child with Dysmorphology	Damara Ortiz, MD
<b>Pedigree Assignment Due 9/30 by 5pm</b>		
Wednesday, 10/2	Whole Exome Sequencing	Jessica Sebastian, MS
Monday, 10/7	Relevant Tools for Genetic Counseling Practice (Variant Classification)	Natalie Carter, MS
<b>Definition Assignment Due 10/7 by 5pm</b>		
Wednesday, 10/9	An approach to a child with a metabolic condition- I	Nadene Henderson, MS
Monday 10/14	An approach to a child with a metabolic condition-II	Cate Walsh Vockley, MS

Wednesday, 10/16	Genetic counseling in cardiovascular genetics	Emily James, MS
Monday, 10/21	<b>Midterm Exam</b>	
Wednesday, 10/23	GINA, HIPAA, and the ADA Alternative Service Delivery Models	Andrea Durst, MS, DrPH
Monday, 10/28	Group Projects (Groups 1 and 2)	Students
Wednesday, 10/30	Group Projects (Group 3 and review activities)	Students
Monday, 11/4	Class rescheduled-Attend the 2 <sup>nd</sup> Annual Pitt Genetics Retreat	
<b>Tuesday 11/5 12:30-1:55PM</b>	An Approach to a Child with Down Syndrome <b>HUGEN 3140</b>	Kishore Vellody, MD
	Fact Sheet Evaluation Assignment Due 11/4 by 5pm	
Wednesday, 11/6	Newborn Screening	Cate Walsh Vockley, MS
Monday, 11/11	Families with Down syndrome	Meredith Peterson, Family Panel
Wednesday, 11/13	Teratology	Robin Grubs, MS, PhD
	Fact Sheet Assignment Due 11/13 by 5pm	
Monday, 11/18	Prenatal Diagnosis I <b>Magee Womens Hospital – Room 2700</b>	Luanne Fraer, MS
Wednesday, 11/20	Prenatal Diagnosis II (Ultrasound) <b>Magee Womens Hospital – Room 2700</b>	TBD
Monday, 11/25	<b>No Class—Thanksgiving Break</b>	
Wednesday, 11/27	<b>No Class—Thanksgiving Break</b>	
Monday, 12/2	Prenatal Diagnosis III <b>Magee Womens Hospital – Room 2700</b>	TBD
Wednesday, 12/4	Prenatal Diagnosis IV <b>Magee Womens Hospital – Room 2700</b>	TBD
	Teratogen Assignment Due 12/4 by 5pm	
Monday, 12/9	Psychiatric Genetic Counseling	Robin Grubs, MS, PhD
Wednesday, 12/11	<b>Final Exam – Take home exam</b>	

## **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, evaluate, 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 prenatal screening/testing procedures as well as other genetic/genomic testing approaches including their application and associated risks.
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 via email to both Robin and Andrea 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. Durst.Definitions.doc). When possible, documents should be in Word or PowerPoint format (it is harder to add comments in pdf documents).

### **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, etc.). 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, evaluate, 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 terms. These exercises will help you develop the skills necessary to perform this task successfully.

## Group Projects 52 points

Learning Objective 2: Identify, evaluate, 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 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 emailed to Robin, Andrea, and your classmates.
- **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 Evaluation Assignment**  
**15 points**

Learning Objective 2: Identify, evaluate, 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 be paired with a classmate and provided with a patient-facing piece of literature regarding a genetic condition. Based on information reviewed during class about providing information to patients about genetic conditions, you will evaluate this piece and submit a one page critique of the layout, readability, appropriateness of the piece for a patient audience, etc., as well as a bulleted list of the five most important changes you would make to the document.

**Fact Sheet Assignment**  
**25 points**

Learning Objective 2: Identify, evaluate, 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 be given a list of several conditions to look up in the databases/resources reviewed in class. You will write a fact sheet on the condition you are assigned. These resources are valuable assets for genetic counselors and this assignment will help you become familiar with them.

**Teratogen Letter Assignment**  
**30 points**

Learning Objective 2: Identify, evaluate, 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 6: Describe prenatal screening/testing procedures as well as other genetic/genomic testing approaches including their application and associated risks.

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 description of an assigned teratogenic agent. The description will be in a format of a patient letter. Your patient is a high school graduate who has never heard about the concerns of prenatal exposure to the agent. You need to include the critical time of exposure, the risk associated with the agent, the effect of the agent (associated malformations), and possible testing options to identify associated malformations. After you complete the description, you will provide a copy to a fellow classmate. Your classmate will review the description and try to imagine that he/she is a patient reading the letter. Your classmate will try to identify areas that may need improvement or clarification. The job of the reviewer will be to critique the paper, not to criticize the writer. Once the critique is completed, the writer will revise the paper based upon the reviewer's comments and then hand in the paper for grading.