

# HGEN 6470: Clinical Genetics Across the Lifespan

---

**Credit Hours: 3.0**

## Contact Information

Name	Position	Phone	Email
Faculty TBD			UUGPGC@utah.edu

## Course Information

### Brief Description of Course

This course consists of lectures on the etiology, natural history, molecular genetics, available treatment, inheritance patterns, medical concerns across the lifespan including care transition and counseling issues of a wide variety of genetic conditions or categories of conditions. Prerequisite: University of Utah Graduate Program in Genetic Counseling (UUGPGC) students or permission of instructor; undergraduate course in genetics.

### Course Objectives

At the conclusion of this course students will be able to:

1. Explain the etiology, clinical features, and natural history of several common genetic conditions.
2. Expand their foundation and background in clinical medical genetics essential for effective genetic counseling.
3. Identify and address the medical and genetic issues involved in diagnosing and managing individuals with genetic conditions across the lifespan.
4. Distinguish between age-specific management and genetic counseling issues in adults and children with genetic conditions.
5. Recognize and identify indications for and methods of genetic diagnosis including physical assessment, laboratory, and other diagnostic studies.
6. Discuss the impact of genetic conditions on families and recognize the needs of families coping with these conditions.

### The ACGC Practice-Based Competencies Addressed in this course:

- Genetics and Genomics Expertise: 1; 1.a; 1.b; 1.c
- Risk Assessment: 2; 2.a; 2.c; 2.d
- Counseling 3, 3.c.
- Communication: 4
- Healthcare Systems: 6, 6.a, 6.c, 6.d.
- Professional Identity: 7; 7.a; 7.b; 7.c

## Course Format & Schedule

### Timeline

One semester

### Educational and Instructional Modalities

This course is held in person for two 90-minute sessions a week. There will be some asynchrony work and occasional recorded lectures within class discussion.

### Role of the Student in this Course

You will be expected to engage in a variety of learning activities including reading, viewing videos, sharing with peers, writing and creating educational presentations.

**Required Textbooks/Readings**

Optional Textbook: Jorde L B, Carey JC, and Bamshad MJ. (2020) Medical Genetics (6th edition).

Optional Textbooks: Reed and Donai (2021) New Clinical Genetics (4rd edition)

Assigned Readings, Optional Readings, Resources:

Readings will be found in Canvas under each individual class and may be printed or downloaded on your home computer. Make sure you read the headers and footers, as they contain textbook chapter information and websites that are required reading.

**Assessment & Grading**

**Evaluation Methods**

1. Exams: two examinations will be given (midterm and final examination) each worth 25% of the final grade. All students must take both exams. Exams cover lecture and homework content as well as readings from the textbook and assigned articles. The final examination is comprehensive but will focus primarily on material from the second half of the course. A student will be given an early or late exam only in the event of an extreme emergency.
2. Quizzes and Assignments: five quizzes will be given during the class and one cumulative assignment ("condition spreadsheet") will be worth 30% of the final grade. Please make sure that all of your questions are answered during lectures to make sure you can complete the quiz questions. On the due date, homework should be submitted electronically or in person by the specified time. These assignments will cover lecture and reading materials. \*One point, per day will be deducted for any assignments turned in late.
3. Syndrome Review Assignment: Each student will be assigned a genetic condition at the beginning of the semester and will present on that condition at the beginning of the relevant lecture based on personal research and knowledge gained during this course. This assignment will count for 10% of the overall grade. Students will be expected to present on the syndrome, in detail, create a quick reference hand out for their peers, and write two quiz questions about the condition at a reasonable difficulty level.
4. Participation: Up to 10% of the final grade will be given for active participation. This means attending lectures, being prepared for class, and initiating and participating in class discussions. In-person attendance is mandatory. Exceptions will be made on a case-by-case basis and must be preapproved by a course director.

**Grading System**

Students will receive a final letter grade. To pass this course you must achieve an overall score of at least 80% (a B- equivalent) as required by the Department of Human Genetics.

Grading Scale

A	95-100
A-	90-94
B+	86-89
B	82-85
B-	79 – 81
C+	76-78
C	73-75
C-	70-72
D+	67-69
D	64-66
D-	60-63
F	Below 60

## Standard Policies

### **The Americans with Disabilities Act.**

The University of Utah seeks to provide equal access to its programs, services, and activities for people with disabilities. If you will need accommodations in this class, reasonable prior notice needs to be given to the Center for Disability Services, 162 Olpin Union Building, 801-581-5020. CDS will work with you and the instructor to make arrangements for accommodation. All written information in this course can be made available in an alternative format with prior notification to the Center for Disability Services.

### **Addressing Sexual Misconduct.**

Title IX makes it clear that violence and harassment based on sex and gender (which includes sexual orientation and gender identity/expression) is a civil rights offense subject to the same kinds of accountability and the same kinds of support applied to offenses against other protected categories such as race, national origin, color, religion, age, status as a person with a disability, veteran's status or genetic information. If you or someone you know has been harassed or assaulted, you are encouraged to report it to the Title IX Coordinator in the Office of Equal Opportunity and Affirmative Action, 135 Park Building, 801-581-8365, or the Office of the Dean of Students, 270 Union Building, 801-581-7066. For support and confidential consultation, contact the Center for Student Wellness, 426 SSB, 801-581-7776. To report to the police, contact the Department of Public Safety, 801-585-2677(COPS).

### **Campus Safety.**

The University of Utah values the safety of all campus community members. To report suspicious activity, call campus police at 801-585-COPS (801-585-2677). You will receive important emergency alerts and safety messages regarding campus safety via text message. For more information regarding safety and to view available training resources, including helpful videos, visit [safeu.utah.edu](http://safeu.utah.edu).

### **Alternate Name and/or Personal Pronoun**

Class rosters are provided to the instructor with the student's legal name as well as 'Preferred' first name (if previously entered by you in the Student Profile section of your CIS account). While CIS refers to this as merely a preference, we will honor you by referring to you with the name and pronoun that feels best for you in class, on papers, exams, group projects, etc. Please advise us of any name or pronoun changes (and please update CIS) so we can help create a learning environment in which you, your name, and your pronoun will be respected.

### **Current COVID-19 Campus Guidelines**

COVID-19 Guidelines change rapidly. Please access the most current information COVID-19 Central @ The U. [coronavirus.utah.edu](http://coronavirus.utah.edu)