

HGEN 7380: Biochemical Genetics

Credit Hours: 3.0

Contact Information

Name	Position	Phone	Email
Faculty - TBD		801-581-8629	UUGPGC@utah.edu

Course Information

Brief Description of Course

Clinical Biochemical Genetics applies the knowledge of biochemistry to the diagnosis and treatment of human disorders. It translates advances in medical diagnosis research into clinical practice. It requires the strict interaction between the laboratory and the clinician to achieve the best possible patient care. Although the discipline has been applied mostly to inborn errors of metabolism, it is becoming evident that several common disorders, such as diabetes and hypertension, have biochemical bases correctable by diet or other medical intervention. The goal of this course is to provide the scientific bases for such interventions. This will be accomplished by providing an overview of biochemical pathways, practical experience on how the biochemical pathways can be studied in vivo and in vitro, the molecular bases of common metabolic problems, the mechanism of inheritance including recurrence risk, and how to rationally treat metabolic blocks.

Course Objectives

At the completion of the course, students will be able to:

1. Provide an overview of inborn errors of metabolism
2. Identify inheritance pattern and recurrence risk
3. Define tests needed for the diagnosis of inborn errors of metabolism
4. Discuss therapy of inborn errors of metabolism

The ACGC Practice-Based Competencies Addressed in this course:

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

Course Format & Schedule

Timeline

One semester

Educational and Instructional Modalities

This course is held in person for one 120 minutes and one 60 minutes sessions a week.

Role of the Student in this Course

Students will be expected to engage in a variety of learning activities including reading, homework and case based discussions.

Suggested Textbook

- Kyriakie Sarafoglou, Georg Hoffmann, and Karl Roth. **Pediatric Endocrinology and Inborn Errors of Metabolism, Second Edition.** McGraw Hill, (2017) ISBN-13: 978- 0071773140, ISBN-10: 0071773142
- Additional reading assignments and sources will be found in Canvas under each individual class.

Assessment & Grading

Evaluation Methods

The final grade will be based upon midterm and final examinations, homework assignments and discussions, participation in class. **Students must earn at least B- to pass the course.**

Grading System

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

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.

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