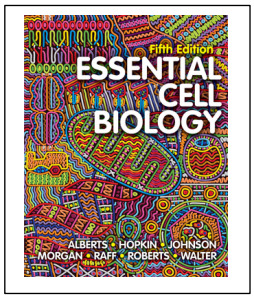
Biosc 1050: Cell and molecular biology of disease

Time: Tuesday/Thursday, 2:30-3:45
Location: G8 Cathedral of Learning
Instructor: Dr. Jeffrey Hildebrand
Email: jeffh@pitt.edu
Office: Rm 103 Life Sciences Annex

OFFICE HOURS: Wednesday 3pm or Friday at 1:30pm in Room L1 Clapp Hall in person. If that time does not work for you, please arrange an appointment, in person or via Zoom.

Texts:
Highly recommended:
Essential Cell Biology. Fifth edition. *Alberts, Bruce; Hopkin, Karen; Johnson, Alexander; Morgan, David; Raff, Martin; Roberts, Keith; Walter, Peter.*
Older editions are probably fine as are versions of Molecular Biology of the Cell (5th and 6th editions).



Other resources:

- Metabolic and Molecular Bases of Inherited Disease (MMBID) – 8th Edition – http://ommbid.mhmedical.com, on Pitt campus or through Pulse Secure VPN
- Online Mendelian Inheritance in Man, OMIM: https://www.omim.org/
- Genetics Home Reference: https://ghr.nlm.nih.gov/
- Gene Reviews: https://www.ncbi.nlm.nih.gov/books/NBK1116/
- Molecular Biology of the Cell (Alberts) 4th edition available from NCBI Bookshelf, https://www.ncbi.nlm.nih.gov/books/NBK21054/

I am committed to creating an environment that is welcoming and inclusive for every student and in which everyone feels a sense of belonging. If you ever have any questions, comments, requests, or concerns please don't hesitate to contact me. Your background, experiences, interests, identities, thoughts, and perspectives are an important part of creating a collaborative environment that enables the most effective, vibrant, and beneficial learning process for all. I value student contributions to the learning process and expect members of the class to treat others with respect. I strongly believe that everyone can learn the material and will do well if they put in the effort to keep up and actively engage in the course.

CLASS LEARNING OBJECTIVES
The advent of molecular biology and the Human Genome Project has dramatically increased our understanding of the mechanisms of human disease. The underlying molecular causes for many diseases have been elucidated. This course examines how abnormalities that occur at the molecular and cellular level manifest as pathologies affecting the structure and function of human tissues and organs. In addition, this course focuses on the pathophysiology of common human diseases and the environmental, genetic and epigenetic causes of specific disease types. As such, by the end of the course you should be able to, among other skills:

1. Discuss the structure, function, maintenance, and heritance of genetic material.
2. Understand and describe the relationship between DNA, RNA, and protein and how mutations can alter this flow of information.
3. Comprehend the structure and function relationship of proteins and the principles of enzymes and their regulation.
4. Apply the principles of genetics to family pedigree and to distinguish patterns of inheritance for single gene disorders linked to autosomes and sex chromosomes.
5. Demonstrate an understanding of cell structure, organelle function, and protein transport.
6. Discuss the mechanisms and regulation of cell communication.
7. Demonstrate an understanding of molecular pathways and how alterations lead to human disease.
8. Display familiarity with the experimental approaches used in cell and molecular biology and demonstrate the ability to analyze and interpret data and graphs.
9. Search for, read, and comprehend primary literature.
10. Demonstrate the ability to effectively communicate and work collaboratively to successfully address problems in molecular and cell biology.
11. Demonstrate professionalism by behaving in a professional, courteous and respectful manner when engaged in all course-related activities.

Grading

Finals grades will be determined using the following breakdown:

1. In class assessments: up to 1 point each, up to a total of 15 points.
2. Case studies (5): 10 points each for total of 50 points
3. Group presentation (1): 15 points
4. quizzes (6): 30 points each, total of 180 points

Final grade will be determined according to the grading scale as follows:

Final %	Grade	Final %	Grade
98	A+	72	C
92	A	70	C-
90	A-	67	D+
87	B+	62	D
82	B	60	D-
80	B-	≤59.9	F
77	C+		

Assessments:

1. **Quizzes** will be given IN CLASS via CANVAS. Therefore, you will be required to have a device capable of accessing Canvas and TopHat during class. Each quiz will be given DURING THE FIRST 45 MINUTES OF CLASS. You will be required to log on to TopHat for attendance prior to the quiz. These will consist of a mix of question types, including multiple choice, fill-in, and short answer. To provide some flexibility, the lowest quiz

score will be replaced with your highest quiz score. This does not apply in the case of an unexcused missed quiz.

2. **Case studies** will consist of a set questions regarding an assigned paper. You will be required to answer a set of questions regarding each paper These must be turned in prior to the day on which the paper will be discussed. Each will be worth 10 points. I encourage you to discuss the paper with your peers, but you are expected to submit your own work. These will be turned in via Canvas.
3. **Presentations.** For each case study, a number of students will be selected at random to present certain aspects of the paper to the class. I will provide a serious of presentation prompts for each figure/topic to be presented to the class. Student that are presenting will not be required to turn in a case study assignment for that module.
4. **In class questions.** There will be the opportunity for students to earn points in class, via TopHat. These will review material from the previous class or will be pertain to material discussed in class that day. You will receive 0.5 points for attempting the question and 0.5 points for the correct answer. You can receive up to 15 points for the semester. These points will be added to the final point total. Yes, these are essentially extra-credit points.

There are no make-up quizzes or assignments in this course. If you miss a quiz or assignment and do not have a valid reason, you will receive no credit for the quiz or assignment. Acceptable reasons for missing an assessment include illness, injury, or a death in your immediate family. To receive an excused absence, you must submit in writing, no later than 3 business days after the due date: your name, the reason for missing, and date you missed. In some cases, documentation may be requested to validate of absence. If you are granted an excused missed assessment, you will be given an extension to complete the assessment. If you know in advance that you may not be able to meet a specific deadline, contact Dr. Hildebrand immediately. You can submit assignments prior to the due date, which avoids the possibility of technical or other complications.

Academic Integrity
Students in this course will be expected to comply with the University of Pittsburgh's Policy on Academic Integrity. This includes cheating on exams and plagiarism. A student suspected of violating this obligation for any reason during the semester will be required to participate in the procedural process, initiated at the instructor level, as outlined in the University Guidelines on Academic Integrity. This may include, but is not limited to, the confiscation of the examination of any individual suspected of violating University Policy. At a minimum, students will receive a 0 for the assessment in question. Furthermore, no student may bring any unauthorized materials to an exam. To learn more about Academic Integrity, visit the Academic Integrity Guide for an overview of the topic.

Disability Services
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 (DRS), 140 William Pitt Union, (412) 648-7890, drsrecep@pitt.edu, (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.

Email Communication
Each student is issued a University e-mail address (username@pitt.edu) upon admittance. This e-mail address may be used by the University for official communication with students. Students are expected to read e-mail sent to this account on a regular basis. Failure

to read and react to University communications in a timely manner does not absolve the student from knowing and complying with the content of the communications. The University provides an e-mail forwarding service that allows students to read their e-mail via other service providers (e.g., Hotmail, AOL, Yahoo). Students that choose to forward their e-mail from their pitt.edu address to another address do so at their own risk. If e-mail is lost as a result of forwarding, it does not absolve the student from responding to official communications sent to their University e-mail address.

Daily schedule

MODULE	Date	**Topic**	**Resources**
Module 1	10 Jan	Course overview and expectations. Genetic material and review of genetics	Chapter 19
	12 Jan	Genetic manipulations and "-omics". Genes and genomes	Chapter 9 and 10
	17 Jan	The structure/function relationship of proteins.	Chapter 4
	19 Jan	Case study: Limb development DNA structure	https://doi.org/10.1038/gim.2018.18 Chapter 5
	24 Jan	Quiz 1	
Module 2	24 Jan	DNA replication and repair	Chapter 6
	26 Jan	RNA transcription	Chapter 7
	31 Jan	Regulation of gene expression	Chapter 8
	02 Feb	Case study: Uncombable hair Protein translation	https://doi.org/10.1016/j.ajhg.2016.10.004 Chapter 7
	07 Feb	Mitochondria and energy	Chapters 13 and 14
	09 Feb	Quiz 2	
Module 3	09 Feb	Membrane structure and function	Chapter 11
	14 Feb	Membrane structure and function	Chapter 12
	16 Feb	Protein trafficking	Chapter 15
	21 Feb	Case study: Progerin and the nucleus Cytoskeleton	https://doi.org/10.1073/pnas.0511133103 Chapter 17
	23 Feb	Cellular interactions	Chapter 20
	28 Feb	Quiz 3	
Module 4	28 Feb	Cellular and tissue organization	Chapter 20
	02 March	Case Study: In vivo editing	https://doi.org/10.1016/j.omtn.2022.07.024
07 March and 09March SPRING BREAK			

Module 4	14 march	Cell division	Chapter 18
	16 March	Cell division, part 2	Chapter 18
	21 March	Quiz 4	
Module 5	21 March	Cell signaling I	Chapter 16
	23 March	Cell signaling II	Chapter 16
	28 March	Cell signaling III	Chapter 18
	30 March	Case Study 5: escaping CAR-T Cell death	https://doi.org/10.1038/s41467-022-31035-7 Chapter 18
	04 April	Cancer	Chapter 20
	06 April	Quiz 5	
Module 6	06 April	Developmental biology	Lecture materials
	11 April	The mammalian embryo	Lecture materials
	13 April	Stem cells	Chapter 20 and lecture
	18 April	Genome modification	Chapter 10 and lecture
	20 April	Case study 6: iPS, organoids, and gene repair	https://doi.org/10.1038/s41467-020-17954-3
	April 26 8AM	Quiz 6	