

**Special Topics 3895: *The Molecular Genetics of Inherited Human Disease, and the use of Computational Techniques to identify Potential Therapeutic Agents***  
**(Fall 2021)**

**Monday and Wednesday, (2:30-3:20PM)**  
**Torrey Life Science, Rm. 263**

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Text: An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases

By: Jack J. Pasternak-Please note, this text will not be made available through the UConn Book Store

COURSE Goal I: Foster-an appreciation for the causal relationship between the Human genome/cell physiology and Disease, and Demonstrate-how an understanding of the genetic/molecular nature of the cell has and will continue to enhance development of effective therapeutics.

Goal II: This course aims to link molecular biological processes to observable genetic disease phenotypes in humans. In the process opportunities to leverage advanced computational tools for the purposes of drug discovery and student publication may present themselves. The semester-long projects will be detailed in HuskyCT.

**Objectives:**

- 1. Attend weekly lectures/discussions**
- 2. Student develop research projects demonstrating the ability to use various Computational Techniques in resolving potential therapeutics**
- 3. Complete quizzes relevant to course of study designed to demonstrate mastery of important concepts**

**ASSIGNMENTS/ASSESSMENTS:**

*Quizzes*

Student researched oral and written presentations:

Student Problem-Ligand Binding

**ORGANIZATION OF CLASS PERIODS:**

- Monday-Wednesday: Lecture/Discussion/Student Research Projects

## CLASS SCHEDULE

### Part I. The Molecular Genetics of Inherited Human Disease

<b>Week/Day/Month</b>	<b>Topics</b>	<b>Chapter Readings</b>
Mon. 8/30	Human Genetic Disease Genes and Phenotypes	Chapter 1: (pp. 1-9) Chapter 1: (pp. 9-17)
Wed. 9/01	Human Chromosomes and The Meiotic Process	Chapter 2: (pp. 19-22)
Mon. 9/06	<b>Labor Day-No classes</b>	
Wed. 9/08	Characterizing Human Chromosomes And Chromosome Abnormalities	Chapter 2: (pp. 23-36)
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— Mon. 9/13	<b>3D-Protein Modeling/Pymol</b>	Lysozyme
Wed. 9/15	<b>Introduction to Pymol Sample Problems</b>	Discussion
Mon. 9/20	<b>Pymol Continued: Introduction to Student Presentations</b>	Discussion
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— Wed. 9/22	Mendal's Laws of Inheritance and Genetic Linkage	Chapter 3: (pp. 37-47)
Mon. 9/27	Constructing Genetic Maps Multiple Alleles and Human Genetics	Chapter 3: (pp. 47-54) Chapter 3: (pp. 54-71)
Wed. 9/29	Decoding Genetic Information, Mutations of Structural Genes, Dominant Mutations and Genetic Disorders	Chapter 4: (pp. 79-95) Chapter 4: (pp. 95-105)
Mon. 10/ 04	<b>Chapter Quiz</b>	
Wed. 10/06	Restriction Endonucleases and Cloning Vectors Screening DNA Constructs	Chapter 5: (pp. 107-122)

Mon. 10/11	DNA Hybridization-Rodent Somatic Cell Hybrids DNA Libraries, Chemical Synthesis of DNA Human Genetic Files	Chapter 5: (pp. 122-139) Chapter 5: (pp. 140-149)
Wed. 10/13	Genetic Mapping of Human Chromosomes-Mapping of Genetic Disease Loci	Chapter 6: (pp. 153-159) Chapter 6: (pp. 159-171)
Mon. 10/18	Genotyping Single -Nucleotide Polymorphisms, Physical Mapping	Chapter 6: (pp. 172-184)
Wed. 10/20	Cloning Human Disease Genes, Detecting Mutations in Human Genes	Chapter 7: (pp. 189-199)
Mon. 10/25	<b>Chapter Quiz</b>	
Wed. 10/27	Similarity Search of a DNA Data Base Functional Genomics, Proteomics	Chapter 8: (pp. 203-211) Chapter 8: (pp. 212-220)
Fri. 10/29	<b>Research Project (1) is due</b>	
Mon. 11/01	Phenotypic Variation of Monogenic Disorders Oligogenic Disorders and Polygenic Inheritance	Chapter 10: (pp. 295-303) Chapter 10: (pp. 304-325)
Wed. 11/03	Parent of Origin Effect, Prader -Willi and Angelman Syndromes	Chapter 11: (pp. 333-345)
Mon. 11/08	<b>Chapter Quiz</b>	

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<b>Week/Day/Month</b>	<b>Topics</b>	<b>Discussion</b>
<b>Research Project (1)/Presentations</b>		
Wed. 11/10	Start of Student Presentations	
Mon. 11/15	Student Presentations	
Wed. 11/17	Student Presentations	

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**Mon. 11/22-Sun. 11/28 Thanks Giving Recess**

<b>Week/Day/Month</b>	<b>Topics</b>	<b>Discussion</b>
Mon. 11/29	<b>Research Project (2)</b> is due/Start Student Presentations	
Wed. 12/01	Student Presentations	
Mon. 12/06	Student Presentations	
Wed. 12/08	Student Presentations, completed, Semester Summary	

**MASKING:** Until further notice, to ensure a safe learning environment for everyone, masks/face coverings must be worn at all times when inside buildings, including in the classroom, regardless of vaccination status. If a student is not wearing a mask/face covering, they will be asked by the instructor to put one on immediately or leave the classroom. Repeatedly failing to follow this expectation will result in a referral to Community Standards. Activities that involve temporarily removing the mask, such as eating or drinking are not allowed. Please leave the classroom for such activities. If an instructor is not wearing a mask/face covering, students should feel comfortable asking the instructor to put one on immediately. More information about proper usage of masks is available from UConn Environmental Health and Safety at this [link](#).

Although social distancing will not be required inside classrooms for vaccinated individuals, please be respectful of the wishes of others who prefer to maintain social distancing. For their own protection, unvaccinated individuals are requested to maintain 6 feet social distancing from others.

### **DISABILITIES**

Any student with disabilities that he/she would like the faculty to be aware of should communicate that information in confidence to the faculty and any issues arising will be addressed in accordance with the policy of the University.

### **ACADEMIC MISCONDUCT STATEMENT:**

“Academic misconduct in any form is in violation of the University of Connecticut Student Conduct Code and will not be tolerated. This includes, but is not limited to: copying or sharing answers on tests or assignments, plagiarism, and having someone else do your academic work. Depending on the act, a student could receive an F grade on the test/assignment, F grade for the course, or could be suspended or expelled.”

### **Policy On Plagiarism**

Plagiarizing is defined as “**To steal and pass off (the ideas or words of another) as one's own: use (another's production) without crediting the source**”(www.Merrian-Webster.com, 2005)

**Plagiarism violates the Academic Misconduct section of “The Student Code” of the University of Connecticut (<http://web.uconn.edu/mcb201/misconduct.html>) and will not be tolerated in MCB courses. The instructors of MCB 241W will adhere to the guidelines laid out in “The Student Code”; therefore, students should read and understand these policies and the consequence of violations. The definition of plagiarism extends to all aspects of evaluated work in this course. Copying another**

**student's work is plagiarism. Failure to give full and proper citation to other people's work is plagiarism.** Full and proper citation includes putting quotation marks around any quoted passage, including a correct citation to the publication from where the ideas originated and a complete reference to that publication in the "literature cited" section. This applies to all forms of communication including websites or personal communication from someone, such as would occur in verbal discussions of scientific data. Direct quotations are appropriate when the original statements would lose clarity or intent. However, your assignment should not include multiple direct quotations. **Paraphrasing of other authors' work is acceptable given that the ideas contained in the paraphrased passage are properly attributed to the author and the ideas are re-worded into the student's own original language.**

There are many resources available to students:

PLEASE COMPLETE THE PLAGIARISM MODULE IN HUSKYCT.

Should you need additional information the following web sites may be of help:

<http://www.lib.uconn.edu/using/tutorials/LILT/plagiarism.htm>

[http://owl.english.purdue.edu/handouts/print/research/r\\_plagiar.html](http://owl.english.purdue.edu/handouts/print/research/r_plagiar.html)

The grading policies for Special Topics 3895 are as follows:

Late Assignments: 5 point/day deduction; after seven days = 0

**Grade Cutoffs:**

Range	Letter Grade
> 93	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-
≤ 59	F

**Course grade:**

Assessment	Number of Points	Total
(3) Unit Quizzes	10 x 3	30
(1) PyMol Assignment	20	20
(2) Research Modeling Projects	60 x 2	120
(2) Research Presentations	15 x 2	30

Total=200pts

**Have a great Semester**