

Biology 861P: Human Genetics

Instructor: Dr. Kimberly A. Carlson
Phone: 865-1554

Office: Bruner Hall of Science Room 201D
E-mail: carlsonka1@unk.edu

Contact Hours: M-F: 9:00 am to 5:00 pm central standard time (CST). I do not check email after 5:00 pm CST and rarely on weekends.

Course Materials:

- Required: *Human Genetics* (12th edition, Domestic version) by Ricki Lewis (ISBN ##978-1-259-70093-4)
- Announcements, assignments, tests, and quizzes: All information for this class will be available on Canvas. You should check it regularly.
- E-mail: All students MUST use their UNK email account so that I can reach them. All emails to Dr. Carlson should be sent from your email account and **not** through blackboard.

Course description: The course focuses on contemporary human genetics with emphases on genetic diseases. Some class topics may be upsetting in terms of ethics, human relationships, or religion. Talk to me beforehand if you have any questions or concerns.

Course objectives:

1. Understand the analysis of genetic transmission in families and populations.
2. Understand the genes, DNA, mutation, and control of gene expression.
3. Understand population genetics.
4. Understand the role of genetics in human disease.
5. Discuss the role of human genetics in medicine and society.
6. Understand genetic technologies.
7. Develop & reinforce writing skills through case studies & discussions.

Students will achieve these objectives through lectures, online quizzes/tests, and discussion board.

Expectations: Grades are based on participation and quality of discussion, quizzes, tests, and case study workbook answers. Students must participate online for all discussion topics. You are expected to post on blackboard weekly on the case study(ies) assigned. The quality of the discussion is exhibited by the contribution of original ideas, respect for differing opinions, and evidence of knowledge on the subject matter.

Grading Scheme:	Points	Due Date
Exam #1	100	Friday, September 28 th by 11:55 pm CST
Exam #2	100	Friday, November 2 nd by 11:55 pm CST
Exam #3	100	Wednesday, December 12 th by 8:00 am CST
Quizzes (15 @ 10 pts each)	150	Weekly from Monday 8:00 am CST to Friday 11:55 pm
Case Study Discussion Board (11 @ 10 points each; includes introduction & syllabus – 5 pts each)	110	11:55 pm CST on the Friday of the week when discussion board ends
Group Participation (2 @ 20 points each)	40	8:00 am CST on 10/5 and 12/11
Total Points	600	

Grade assignment: Grades will be assigned using the standard grading scale for the Department of Biology, as follows: A (93-100%), A- (90-92%), B+ (88-89%), B (83-87%), B- (80-82%), C+ (78-79%), C (73-77%), C- (70-72%), D+ (68-69%), D (63-67%), D- (60-62%), and F (below 60%). FYI, I do not round up grades. Therefore, if you have an 82.6%, you have earned a letter grade of a B-, not a B. Also, 1% = 6 points not 1 point. Grade assignments are final unless there is a calculation error.

Lecture topics: Chapters 1 (to Precision Medicine on page 8), 2 (to The Human Microbiome on page 36), 3 (to Disorders that Resemble Accelerated Aging on page 61), part of Chapter 4 (to 4.2 Single-Gene Inheritance is Rare on page 71), Chapter 9 (entire chapter) & Chapter 10 (entire chapter) are considered a review of material that you should have had in a basic biology course. You are solely responsible for this material. You will be given a 100 point Review test over this material. This schedule is subject to change. Small

portions of chapters may not be covered. The parts of CH 1, 2, and 3 that are not part of the review will be discussed in the Topic: Why is Genetics important and interesting and will be on the first test. The remainder of Chapter 4 will be discussed as listed below. My PowerPoint slides will **not** be made available to the class for the topics listed below. They will be available for the review material. I will be placing fillable outlines on Canvas for you to use while attending the lectures. You are solely responsible for this material. You will be given a timed 10-point quiz (5 minutes/5 questions/2 pts each) over each review chapter for a total of 60 points. These quizzes can be taken at any time during the semester, but must be completed by September 2nd at 11:55 pm CST. At this time, they will be removed from Canvas. I have indicated times in the semester when you may want to take these quizzes. This schedule is subject to change. Small portions of chapters may not be covered. My PowerPoint slides will **not** be made available to the class. They are copyrighted by the book publisher and can't be distributed. All Lectures will be available on Canvas.

Week	Chapter (Ch.)	Quiz/Test	Case Study Discussion Board
#1 8/20-24	Why is Human Genetics Important and Interesting?	Overview quizzes	Introductions & Syllabus
#2 8/27-8/31	13: Chromosomes	Overview quizzes	
#3 9/3-7	4: Single Gene Inheritance	Timed quiz #1 over Chapter 13	CS: Forensics Focus Question #2 for CH 4
#4 9/10-14	5: Beyond Mendelian's Law	Timed quiz #2 over Chapter 4 (p73-85)	CS: Case Study #1 for CH 5
#5 9/17-21	6: Matters of Sex	Timed quiz #3 over Chapter 5	CS: Case Study #4 for CH 6
#6 9/24-9/28		Exam #1-Chapters 13 & 4-6 Due 9/28 by 11:55 pm CST	
#7 10/1-5	7: Multifactorial Traits	<i>Group Participation Scores Due by 8:00 am CST on 10/5</i>	CS: Case Study #6 for CH 7
#8 10/8-12	8: Genetics of Behavior	Timed quiz #4 over Chapter 7	CS: Case Study #2 for CH 8
#9 10/15-19	11: Gene Expression and Epigenetics	Timed quiz #5 over Chapter 8	No Case Study – Fall Break
#10 10/22-26	12: Gene Mutation	Timed quiz #6 over Chapter 11	CS: Case Study #3 (a&b) for CH 12
#11 10/29-11/2		Exam #2-Chapters 7,8,11, &12 Due 11/2 by 11:55 pm CST	
#12 11/5-9	14: Constant Allele Frequencies		CS: Case Study #2 (a&b) for CH 14
#13 11/12-16	15: Changing Allele Frequencies	Timed quiz #7 over Chapter 14	CS: Case Study #1 for CH 15
#14 11/19-23	19: Genetic Technologies: Amplifying, Modifying, & Monitoring DNA	Timed quiz #8 over Chapter 15	No Case Study – Thanksgiving Break
#15 11/28-12/4	20: Genetic Testing & Treatment	Timed quiz #9 over Chapter 19	CS: Case Study #7 for CH 19
#16 12/5-11		<i>Group Participation Scores Due by 8:00 am CST on 12/11</i>	CS: Case Study #2 (a&b) for CH 20
#17 12/12-14	Finals week	Exam #3: Chapters 14, 15, 19, & 20 Due 12/14 by 8:00 am CST	

Exams: Exams may be composed of multiple choice, true/false, application problems, case studies, and/or short essays. I believe that genetics and science exams in general should be an application of the material

learned, and not simply rote memorization. The exams are not comprehensive, in that, the questions asked will specifically address material covered since the last exam or in the case of the first exam, material since the first day of class. The material on the test will include lecture material only, not the Case Studies for Discussion Board. The multiple choice/true or false portion of the exam will be taken as a timed online portion. The amount of time allotted will be announced at the time of the test. If you run out of time while taking the test, you will not be given extra time. If you have a problem while taking your test, send me an email and I will deal with it as soon as possible. The computer displays a message to me that tells me what kind of problem you had, whether you took too much time or there was a computer problem. I will be available M-F from 9:00 am to 5:00 pm CST, not constantly as I do have other teaching and research responsibilities. **You must take all three exams to receive full credit. If you miss an exam, you will automatically fail and receive a letter grade of an "F".** The dates for the exams are listed on the schedule. The exam, both online and take-home portions, will be made available at 8:00 am CST on Monday of the week listed and taken off at 11:55 pm CST on Friday of the week listed. For example, the first test will be put on blackboard at 8:00 am CST on Monday, September 24th and you have until 11:55 pm CST Friday, September 28th to take the online portion and email the take-home portion to me. **THE DEADLINE IS FINAL! THE TESTS WILL BE REMOVED AT 11:55 pm CST ON THE FRIDAY DUE DATE. LATE PAPERS WILL NOT BE ACCEPTED! MAKEUP EXAMS WILL BE GIVEN ONLY UNDER EXTREME OR EXTENUATING CIRCUMSTANCES, WHICH CAN BE DOCUMENTED! In any and all cases, the final validity and acceptability of any excuse for absence are at my sole discretion.**

Quizzes: Throughout the semester, you will be given 15 timed quizzes worth 10 points each. Of these, six are over the review material and must be taken by the deadline listed. The other nine quizzes will cover the book/lecture material not the Case Workbook. For all quizzes, you will be allotted 5 minutes to answer 5 multiple choice or true/false questions. You will have only one chance to answer the question and will not be allowed to go back and change answers. Therefore, make sure to answer carefully. If we have done calculations in the lecture for a certain week, make sure to have a calculator with you at the time of the test. For quizzes with calculations, you will be allotted extra time. If you run out of time while taking the quiz, you will not be given extra time. If you have a problem while taking your quiz, send me an email and I will deal with it as soon as possible. The computer displays a message that tells us what kind of problem you had, whether you took too much time or there was a computer problem I will be available M-F from 9:00 am to 5:00 pm CST, not constantly as I do have other teaching and research responsibilities. The dates for the quizzes are listed on the schedule and will be made available at 8:00 am CST on the Monday of the week listed. You have until 11:55 pm CST on Friday of the week listed to take the quiz. For example, the first regular (not review) quiz will be put on blackboard at 8:00 am CST on Monday, September 3rd and you have until 11:55 pm CST Friday, September 7th to take the quiz. **THE DEADLINE IS FINAL! THE QUIZ WILL BE REMOVED AT 11:55 pm CST ON THE FRIDAY DUE DATE.**

General Discussion Board: For this class, there will be two discussion boards. One will be for introductions, syllabus discussion, general questions, and questions over lecture. With the exception of the introductions and syllabus forums, the general discussion board will **not be graded** and there will be no groups established. If you have questions about lecture material, please post it here. In my experience, if you have a question about the material, so does everyone else! Do not post questions about the quizzes or tests here. If you have a question about a quiz or test you have taken, email me directly. If it is something that should be shared with the rest of the class, I will post it.

Case Study Discussion Board: The second discussion board will be the Case Study Discussion Board and this discussion board **will be graded**. In this discussion board, you will be separated into groups and asked to work together. This is a really important practice in the field of Human Genetics, especially in Genetic Counseling because it takes multiple people from different disciplines to make a diagnosis on a genetic disorder or disease at times. This is a good practice for "real world" situations, in that, in science we rarely work alone. The Case Studies are found in your textbook at the end of the chapter. As a group, you will need to turn in one answer for the question(s) posed. These are listed in the lecture outline above. You are to discuss the case during the week and turn in the final document to me at carlsonka1@unk.edu. Do NOT submit through Canvas or Google Docs. You are to format the document as follows with 1" margins all around, 12pt Arial font, Double spacing, Group name and contributing students in the *right hand corner of the header*, Case Study Question # and Chapter in *left hand corner of the header*. The document should be **no longer than 1 page and must contain 2 peer reviewed primary science research articles**. You do not have a limit to the number of references used,

but you must have a minimum of **2 peer-reviewed scientific primary research** journal articles. You may also want to consult OMIM (www.ncbi.nlm.nih.gov/sites/entrez?db=omim), NCBI's Genes and Disease (www.ncbi.nlm.nih.gov/disease/), & Genetic and Rare Conditions Site (www.kumc.edu/gec/support/) for your answer. These will not count as your 2 peer reviewed references, but you can use them and they should be cited as used. These are the only two websites allowed! Books and other websites are not counted as references. In the text, cite references should be by superscripted number¹. In your reference list, use the following format (note punctuation, capitalization, and abbreviation). The references are listed in the order they are cited in the paper, not in alphabetical or chronological order. They should be single spaced and this is the only part of the paper that is single spaced.

Journal articles:

1. Sinoway MP, Kitagawa K, Timsit S, Hashim GA, Colman DR. 1994. Proteolipid protein interactions in transfectants: implications for myelin assembly. *J Neurosci Res* 37:551-562.

Web page:

2. Converse PJ. 2006. Zinc finger protein 175; ZNF175. *OMIM*. Retrieved on August 1, 2018 from <https://omim.org/entry/601139>.

This is a good practice in having an **informed** opinion. Your answer(s) must be typed and sent to me by email. I will accept only one set of answers per group. I would **highly** recommend that you assign a different group leader for each week and have that person compile all of the answers for the group. All of your discussions to answer these questions should be made over Canvas so that I can witness the interactions among the group. I will not be grading these discussions, but will be grading the answers you turn in to me. Your group answers **must** be emailed to me by 11:55 pm CST on the Sunday of the week when discussion board ends. For example, the first Case Study is due on Friday September 7th by 11:55 pm CST. This is for the Chapter 4 case. **LATE PAPERS WILL NOT BE ACCEPTED UNDER ANY CIRCUMSTANCES!** You will have a chance to grade your interactions/participation with each other as part of this process twice during the semester. Halfway through the semester and at the end of the semester, you will score yourself and everyone in your group on a 20-point scale using a rubric I will provide to you. I will compile the scores and the comments each of you make. The comments and scores will be returned to each of you, but in an anonymous fashion, so that problems do not arise. If you do not turn in the Group Participation Scores for yourself and your group mates by the due date, you will receive 0 out of 20 points as your score.

Bonus Points or Extra Credit: I do not give Bonus Points or Extra Credit to Graduate Students, please don't ask.

Students with Disabilities

It is the policy of the University of Nebraska at Kearney to provide flexible and individualized reasonable accommodation to students with documented disabilities. To receive accommodation services for a disability, students must be registered with UNK Disabilities Services Coordinator, David Brandt, in the Academic Success Office, 163 Memorial Student Affairs Building, 308-865-8214 or by email unkdso@unk.edu

Students Who are Pregnant

It is the policy of the University of Nebraska at Kearney to provide flexible and individualized reasonable accommodation to students who are pregnant. To receive accommodation services due to pregnancy, students must contact Cindy Hayes in Student Health, 308-865-8218. The following link provides information for students and faculty regarding pregnancy rights. <http://www.nwlc.org/resource/pregnant-and-parenting-students-rights-faqs-college-and-graduate-students>

Reporting Student Sexual Harassment, Sexual Violence or Sexual Assault

Reporting allegations of rape, domestic violence, dating violence, sexual assault, sexual harassment, and stalking enables the University to promptly provide support to the impacted student(s), and to take appropriate action to prevent a recurrence of such sexual misconduct and protect the campus community. Confidentiality will be respected to the greatest degree possible. Any student who believes she or he may be the victim of sexual misconduct is encouraged to report to one or more of the following resources:

Local Domestic Violence, Sexual Assault Advocacy Agency 308-237-2599; **Campus Police (or Security)** 308-865-8911; **Title IX Coordinator** 308-865-8655

Retaliation against the student making the report, whether by students or University employees, will not be tolerated.