Biology 425 is an upper-level genetics course. An introductory course in genetics and molecular biology is a pre-requisite. If you did not do well in introductory genetics, you should consider carefully whether this course is appropriate for you. We will assume an understanding of basic genetic and molecular principles; you will be responsible for reviewing appropriate topics on your own before lectures, using pre-class lectures posted to SAKAI throughout the semester, online sources, your intro genetics text, or any of the texts currently used in BIOL 202.

The goals of this course are to learn the core principles of Human Genetics and how they are applied to study human disease; to gain an appreciation for human genetics research and how research informs approaches to disease treatment; and to learn to critically evaluate (using higher levels of thinking on the “Bloom’s Taxonomy” hierarchy) the scientific literature to learn how scientists apply core concepts to solving new and increasingly complex problems. This course should be an extension and introduce further applications of what you have been exposed to in your introductory genetics courses. This course and corresponding syllabus are based on the BIOL 425 course taught by Drs. J. Sekelsky and G. Copenhaver during the Fall semester.

**Location**
Genome Sciences Building Room 1374

**What to bring to class**
- Preparedness: working on pre-class activities will ensure you are up to speed on necessary background before delving into the new material presented during class
- A willingness to participate in class discussions including paper discussions and answering questions throughout the class period
- A package of 3 x 5 notecards, lined or unlined

**Contact Info**
   **Office:** Wilson Hall, Rm 237
   **Office Hours:** Monday, 9:30-11 AM; Friday 3-4:30 PM; and by appointment
   I will only check my email (morrislp@email.unc.edu) between 9AM and 5PM, Sunday-Friday

**Course Website:** [www.SAKAI.unc.edu](http://www.SAKAI.unc.edu)
This site will have postings including lecture outlines, supplemental material, pre-class lectures/assignments, and readings. I will also post announcements on this site. *It is your responsibility to check it regularly.*

**Textbook:**
*A Short Guide to Writing About Biology*, Jan A. Pechenik, Pearson Longman, 7th addition (if you have a 6th edition copy, that’s fine.)
**Grading**

Final grades will be based on:
- 20% Exam I
- 20% Exam II
- 20% Exam III (Final exam)
- 15% Problem sets
- 15% Group projects
- 10% Class participation

**Exams**

There will be three exams: Exams I and II will be in class and Exam III will be given during the scheduled final exam period (either Monday July 27 or Tuesday July 28). Please note the dates of each exam, as make-up exams (including early exams) will not be given. Exams will consist of questions similar to those on problem sets, which emphasize conceptual understanding of human genetics. You will not need to know details from assigned articles, but you may be given a reminder of the essential data from an article and asked what main conclusions were drawn from the data, or you may be asked how an assigned reading fit into the principles being discussed at the time.

**Problem Sets**

Problem sets will be assigned approximately every 1.5 weeks, and will include problems and questions related to in-class discussions and reading. The goals with problem sets are to both reinforce and apply the material that was covered and, in some cases, extend beyond what we covered in class. Problem sets will involve a substantial effort. You may work collaboratively to solve the problems, but each student must write and turn in his or her own answers. Late problem sets will not be accepted or graded.

**Readings**

Readings will consist of handouts posted to SAKAI corresponding to each topic and other various handouts. Many lectures will have an assigned reading in the form of a published research article. Assigned articles will be discussed in class. You should bring your copy of the article to class and be prepared to contribute to discussions. To facilitate participation, each student must turn in a response paper (about one-half page, single-spaced) for each article that we will discuss, by 9 am before class on the day the article is being discussed. Do not summarize the article in your response! Instead, I'm interested in your reactions. Did understand that questions and approaches? Do you agree with the conclusions? Did you think the study had any flaws? What do you think is the significance of the results? How does the article relate to issues we’ve discussed in class? What additional questions were left unanswered or raised by the results? These are merely examples of questions you might choose to answer; you don't need to answer all of them all of the time, or there may be different elements of your response that you'd like to discuss. Response papers will count for a portion of your class participation grade. Each response paper will be graded on a scale of 0-5. A response that shows only that you read the article will get a 3. If I can't even tell you read it, you will be given a lower score. Responses that show that you were conscious, engaged, and thinking while reading (which doesn't mean you have to understand it all) will get 4 or 5. When reading, keep in mind that research articles are not merely reports of results. Authors are trying to convince the reader that their results are solid and interpretations are supported. For class discussion, you should think about the following questions: • Why was the study undertaken? • How were the experiments performed or the data gathered? • What are the strengths and weaknesses of the approach taken? • What are the results? You should be able to explain or ask questions about each figure and table. • How did the authors interpret their results? • What are the main conclusions? • Do you agree with these conclusions? • What questions are unanswered or raised by the results?
Group Project
Enrolled students will be placed into groups of 3-4 during the second week of class. Each group will conduct a research project and turn in a paper reporting on the research conducted. Students are expected to work on the projects outside of class, but there will be some class time dedicated to working in your groups. At the end of the semester, each group will present to the class their research project in chalk-talk format. More information will be posted to SAKAI once groups are assigned.

I strongly encourage you to

Other Policies
- All aspects of the UNC Honor Code will be enforced.
- Re-grade requests must be made in writing within one week of receiving a grade (returned problem set, exam, etc.). Only errors in grading are considered, not requests for additional partial credit.
- ALL COURSE MATERIALS ARE COPYRIGHTED. It is a violation of the honor code to distribute course materials outside of the classroom without written permission from the instructors. This includes depositing in fraternity or sorority files or contributing to online repositories.
- Recording of lectures is prohibited unless explicit permission has been granted by the instructor.
  Use of cell phones is prohibited. Google Glass may not be worn during class. Computers are allowed, but only for taking notes or viewing PowerPoints and readings during discussion. Students caught using computers for other tasks (email, surfing, other reading, etc.) will lose computer privileges for the remainder of the semester.
- The schedule provided below is subject to change! Also, since this is in part a discussion-based course, we often have to make small adjustments in response to discussions that are longer than anticipated.
HUMAN HEREDITY IN PEDIGREES

Mon June 22  Introduction; Pedigrees, Simple inheritance patterns
Watch pre-class lecture reviewing pedigree analysis

Tues June 23  Complex Inheritance Patterns

HUMAN CHROMOSOMES

Wed June 24  Mitosis/Meiosis, and Chromosome number abnormalities
Complete pre-class review assignment on meiosis and nondisjunction

Thurs June 25  Chromosome Structure abnormalities

THE HUMAN GENOME

Fri June 26  Sequencing the human genome

Mon June 29  Variation and Genome annotation
Reading: TBD

Tues June 30  Human Evolution, Mitochondrial genetics/ Exam review session
Reading: TBD

Wed July 1  Exam I

IDENTIFYING CAUSES OF HUMAN GENETIC DISEASES

Thurs July 2  Recombination and Introduction to mapping
Reading: TBD

Guest Lecturer: Talia H.

Mon July 6  Segregation mapping

Tues July 7  Association mapping
Reading: TBD
Wed July 8  **Identifying human disease genes**  

**SPECIAL TOPICS IN HUMAN GENETICS**

Thurs July 9  **Epigenetics***/Exam review session  
*Reading:* TBD  
*Guest Lecturer: Greg Z.*

Fri July 10  **Exam II**

Mon July 13  **Ethics**

Tues July 14  **Cancer Genetics***/  
*Guest Lecturer: D. Rogers*  

Wed July 15  **Pharmacogenetics***/  
*Reading:* TBD  
*Guest Lecturer: Greg Z.*

Thurs July 16  **Student choice:** Gene therapy, Reproductive technologies, Immunogenetics OR Genetics of behavior

Fri July 17  Work on group presentations

Mon July 20 through Thursday July 23 **STUDENT PRESENTATIONS** (schedule TBD)

Thursday July 23  Exam III review session

Monday July 27/Tuesday July 28 **Exam III**