FALL 2017 HGEN501/BIOL530—Introduction to Human Genetics
Lectures: 10:00-11:20 a.m. Tuesday/Thursday, 8-036 Sanger

DATE	INSTRUC	TOPIC	READING
Modes of Inheritance & Risk Analysis			
n/a	Grotewiel	Pre-course review of key concepts	Chaps 1 & 2
8/24	Grotewiel	Introduction, classes of genetic disorders, peer advice	Chap 7 (p. 107-124)
8/29	Grotewiel	Mendelian genetics, pedigree analysis	Chap 7 (p. 107-124)
8/31	Grotewiel	Mendelian complexities, Mendelian risk analysis	Chap 7 (p. 107-124)
9/1	Grotewiel	Quiz 1 due (20 points, on Blackboard)	all to date
9/5	Grotewiel	Introduction to population genetics	Chap 9 (p. 155-166)
9/7	Grotewiel	Population genetics and risk analysis, problem session	Chap 9 (p. 155-166)
9/12	Grotewiel	Bayesian risk analysis	Supplemental & Chap 16 (p. 337-342)
9/14	Grotewiel	Problem session	all to date
9/15	Grotewiel	Quiz 2 due (20 points, on Blackboard)	since last Quiz
9/19	Pallante	Multifactorial disorders: no lecture, by video and review	Chap 8 (p. 133-137, 141-152)
9/21	Pallante	Mitochondrial & trinucleotide repeat genetics	Chap 7 (p. 124-131), Chap 12 (p. 246-254)
9/26	G/P	EXAM 1 (160 points)	All to date
Molecular & Biochemical Genetics			
n/a	Lloyd	Review of key molecular concepts	Supplemental Materials
9/28	Lloyd	Gene and genome structure & function	Chap 2, Chap 3
10/3	Lloyd	Determining the function of a gene	Chap 4, p. 61-64 (p. 271-272)
10/3	G/P	Exam 1 Debriefing 11:20 a.m.-12:00 p.m.	n/a
10/5	Lloyd	Human genome variation	Chap 4
10/6	Lloyd	Quiz 3 due (20 points, on Blackboard)	since last Exam
10/10	Lloyd	Gene expression	Chap 3 (p. 27-33)
10/12	Lloyd	Gene expression (continued)	Chap 14 (p. 298-307), Chap 13 (p. 263-270)
10/17	Lloyd	Epigenetics & imprinting	p. 33-41, p. 85-87, p. 134, p. 466-467
10/19	Pallante	Biochemical genetics I	Chap 11, Chap 18 (p.369-372)
10/20	L/P	Quiz 4 due (20 points, on Blackboard)	since last Quiz
10/24	Pallante	Biochemical genetics II	Chap 12 (pp 215-246) & Chap 13
10/26	L/P	EXAM 2 (160 points)	Since last Exam
Cytogenetics			
10/31	Jackson-Cook	Chromosome visualization, structure & organization	Chap 5, Chap 17 (p.361-364)
10/31	Lloyd, Pallante	Exam 2 Debriefing 11:20 a.m.-12:00 p.m.	n/a
11/2	Jackson-Cook	Chromosome visualization, structure & organization	Chap 5, Chap 17 (p.361-364)
11/7	Jackson-Cook	Autosomal aneuploidy & nondisjunction	Chap 6, Supplemental Reading Set
11/9	Jackson-Cook	Sex chromosome aneuploidy & X-inactivation	Supplemental Reading Set
11/14	Jackson-Cook	Translocations: formation & segregation	Supplemental Reading Set
11/16	Jackson-Cook	Problem session	Supplemental Reading
11/16	Jackson-Cook	Quiz 5 (20 points, in class)	since last Exam
11/21	Jackson-Cook	Other structural chromosomal abnormalities	Supplemental Reading Set
11/23	n/a	Thanksgiving Break	n/a
11/28	Jackson-Cook	Cancer cytogenetics & instability syndromes	none
11/30	Jackson-Cook	EXAM 3 (160 points)	Since last Exam
Genetic Counseling			
12/5	Gannaway	Genetic screening/genetic counseling	Chap 16, Chap 17, Chap 19 (p. 383-388)
12/5	Jackson-Cook	Exam 3 Debriefing 11:20 a.m.-12:00 p.m.	n/a
12/7	Vlangos	Clinical Genetic Diagnostics	t.b.a.
12/12	All	CUMULATIVE FINAL EXAM (200 points) 10:00 AM-12:50 PM, 8-036 Sanger	All to date

HGEN501/BIOL530 Introduction to Human Genetics
Tues/Thurs 10:00-11:20, 8-036 Sanger Hall
Fall 2017

Course directors: **Dr. Joyce Lloyd**
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Course Instructors: Dr. Grotewiel, Ms. Pallante, Dr. Lloyd, Dr. Jackson-Cook, Dr. Chris Vlangos, and Ms. Gannaway

Teaching Assistant: Ms. Kaitlyn Riley

Course overview: The primary focus of this course is to provide a strong background in Mendelian and non-Mendelian inheritance, risk analysis, molecular genetics, cytogenetics, genome structure and function, and biochemical genetics. To excel in the course, it is imperative that students attend all lectures and review sessions, work the assigned problem sets, prepare for and take the quizzes and keep up with the flow of material. This is a graduate course, so the pace is rapid and the amount of material covered is rather large. Students are therefore strongly encouraged to read and study daily so that they do not get behind. On average, previous students have spent ~12 hours per week on this course. Note that many of the questions on the exams involve working and solving problems, which makes cramming for the exams in this course wholly ineffective.

Prerequisites: This course is a graduate genetics course that focuses on human genetics. Students are expected to have taken and done well in upper-level undergraduate genetics and chemistry courses. Additionally, students are expected to have previously mastered basic concepts in genetics, cell biology, and biochemistry and to have some grasp of molecular biology techniques.

Text: The required textbook for this course is Thompson & Thompson Genetics in Medicine, Edition 8, 2016 (ISBN: 9781437706963). This text will serve as an excellent reference source. Supplemental reading materials will also be provided. Some notes and possibly other resources may be available to students online.

Lectures: All lectures are on Tuesdays and Thursdays from 10:00-11:20 a.m. in room 8-036 Sanger Hall. Most PowerPoint lectures will be available on Blackboard; regardless of the availability of slides, all enrolled students are expected to attend all lectures. Students are encouraged to use the lecture slides and other materials provided in whatever manner works best for them. Many students find that bringing printouts of the slides to the lecture helps them to follow along, whereas others find it better to take written notes during class and then fill in details with the information from slides posted on Blackboard.

Teaching Assistant: The Graduate Teaching Assistant (TA) will have office hours twice per week (times to be announced), and will be available after class lectures most days. The TA will take questions about course material from students and will work problems with students, as requested.

The TA will also be available for questions about material covered on the quizzes or exams. Prepare for meeting with the TA by identifying questions or other points of confusion that arise while you are attending lecture or studying.

Problem sessions: Some problem sessions are indicated in the lecture schedule. Those and others will be integrated into lectures throughout the course. Understanding the problems discussed in these sessions is critical to learning the material and excelling in the course.

Problem sets: Problem sets will be available on Blackboard**.** Students should REPEATEDLY work the problems to understand the material. Students may work in groups or alone, whichever is more productive. The problem sets will be discussed during the scheduled problem sessions and by the TA during review sessions. The problem sets will emphasize the major topics discussed in the lecture and will represent the essence of the material the students will be expected to master for the exams. Be prepared. You may be asked to work a problem in class. Problem sets are not graded.

Quizzes: Quizzes will cover the topics discussed in lectures, reading assignments and problem sets, and therefore represent an excellent opportunity for students to identify areas of misunderstanding. Students will take quizzes online via Blackboard or in class. There will be five quizzes during the semester, each quiz will be worth 20 points, and each quiz will have its own deadline for completion. Unless otherwise indicated by individual instructors, all quizzes are open book. Once started, all Blackboard and in-class quizzes must be completed in the allotted time. Importantly, each student must take the quizzes without the help of others and collaboration between students on quizzes is strictly prohibited. Any and all reproduction of quizzes or their components is strictly prohibited. This moratorium on reproduction of quizzes includes, but is not limited to, taking of pictures, making videos, hand-scribing, emailing, posting to web pages, taking screen shots, electronic clipping, and verbal retelling.

Exams: Exams will cover the topics discussed in the lectures, textbook and on-line reading assignments, and problem sets. There will be 3 regular exams during the semester as indicated on the course schedule. Each regular exam will be worth 160 points for a total of 480 points. The final exam, worth 200 points, will cover the lectures after the third regular exam and also be cumulative for the semester as a whole. All exams are closed book. Most exam questions will require CONSIDERABLE problem solving. Questions on exams will be a combination of multiple choice, written answer and matching. Students should be prepared to QUICKLY solve and answer questions on the exams. Exams in this course can be rather lengthy.

Exams will not be returned to students. Students will have the opportunity to review their exams after they are graded; however, students must return their exams to the instructor prior to leaving the room. Exams will be retained by the course directors until the end of the term, then shredded and discarded. Students that want to see their exams (1-3) prior to and in preparation for the final exam or at any other time during the semester—WHICH IS A VERY GOOD IDEA—should make arrangements by contacting the course directors or TA. Any and all reproduction of exams or their components is strictly prohibited. This moratorium on reproduction of exams includes, but is not limited to, taking of pictures, making videos, hand-scribing, emailing, posting to web pages, taking screen shots, electronic clipping, and verbal retelling.

Students taking exams can have pens or pencils and stand-alone calculators (i.e. not cell phones) at their seats. All other items including, but not limited to book bags, books, notes, note cards, notebooks, computers, phones, coats, hats, water or other beverages, containers of any kind, snacks, etc. are strictly forbidden. Any items that students believe they need during an exam (e.g.

tissues for respiratory illness or clipboard for seating accommodation) must be approved by the instructor or exam proctor before the exams are distributed. Emergencies will be handled on a case-by-case basis.

In addition to pencils, pens and calculators, students reviewing exams with the TA or instructors may have paper for taking notes. Otherwise, the restrictions on acceptable materials during review of exams are the same as for taking exams. It is acceptable to make notes regarding concepts or ways to solve problems while reviewing exams, but otherwise the moratorium on reproducing exams, exam questions, or their components applies.

Grading system: Together, your exam and quiz scores will determine your final point total and therefore your course grade. In all cases, final grades will be determined at the end of the semester by the total points accumulated by students on their exams and quizzes throughout the course. The grading scale is 90% of total points is an A, 80% is a B, 70% is a C, 60% is a D and <60% is an F. The distribution of final course grades may be curved slightly (normalized) at the end of the semester.

Getting help with course material: The TA and the primary lecturer are students' best sources for addressing questions regarding course material. The TA will be available during review sessions and by appointment. Instructors generally prefer that you email them to make an appointment. If requested, tutors can be provided or recommended who are very knowledgeable and have previously taken this course. If you are having difficulty with course material, contact the TA or relevant instructor right away. Students that are unable to resolve points of confusion via the TA are strongly encouraged to make an appointment with the individual instructor for the lecture or with the course directors. This will allow all students to get the help they need. Please note that if you do not perform as you wanted to on the first exam, you should immediately seek help (i.e. that day).

Getting help with administrative questions: For information regarding course content, grading system, assignment of grades, and other global issues, students should contact one of the course directors.

Absences: Virginia Commonwealth University policy will be followed with regard to health or medical problems that cause students to miss an exam or other assigned work. If a student knows that there will be a problem attending an exam, that student is required to inform one of the course directors prior to the exam and as early as possible. Emergencies will be handled on a case-by-case basis. Approved excuses for missing an exam include: medical (requires a doctor's note specifying that the severity of the illness requires absence from exam or inability to perform the work), death in the immediate family (requires an obituary announcement from newspaper), travel on official University business (requires a note from a VCU representative/supervisor), or time away from class due to interviews for graduate/professional school (requires copy of itinerary from interviewer). You must have an approved absence to make up any work. If you plan to be absent from class due to observance of a religious holiday, you must make arrangements with the instructor prior to the holiday. If arrangements are not made in advance, the absence will be considered unexcused. Unauthorized absences from an examination or quiz will result in a score of zero (0) for the missed work.

To excel in the course, students should:

- Attend lectures—During the lecture, the material indicated in the course schedule will be discussed at the level of detail expected from students on exams. Additionally, the logic

behind a multitude of example problems will be described and it is one of the best opportunity for students to ask instructors questions regarding course material.

- Participate in the problem sessions and discussions during class.
- Work the problem sets—The problem sets will give you key insight into the nature of the material and questions on exams. Working through the problem sets is the primary mechanism by which many students will learn the material. However, it is not sufficient to casually work the problem sets and not become intimately familiar with underlying logic. You should EXTENSIVELY and REPEATEDLY work the problem sets and solution paths prior to each exam.
- Prepare for and take the exams and quizzes—The exams and quizzes will account for all of your points, which in turn, will determine your course grade. Students should anticipate vigorously preparing for each of the regular exams and the comprehensive final exam. Students should be prepared to quickly solve problems on all exams and quizzes.
- Seek help from the TA, course directors, and other instructors whenever you have questions—The bulk of the material in this course lays the foundation for subsequent material. Too often, students will misunderstand key concepts early on and then will not be able to understand the more advanced material that follows. Discuss misunderstandings with the TA and course instructors as they arise.
- Study course material daily—It is recommended that your routine includes reviewing lecture slides before each class, reviewing lecture slides on the same day after class, and taking time to study the material each day. This will ensure that you keep pace with the current material and therefore understand the subsequent material. When studying genetics, it is critical that students actively engage in the learning process. In the past, students have spent an average of 3 hours reviewing and learning the material for each lecture hour.

Preparing for and Taking Exams: Since different people learn in different ways, it is difficult to give specific advice that fits the learning processes of all students. However, below is guidance on excelling on exams that works for the VAST majority of students. Prioritize your learning/study time and be prepared to perform as follows:

- Read the assigned text and other material to get an overview of the topics covered BEFORE each lecture.
- Attend lecture, take notes, ask questions, and participate in the classroom discussion.
- Rewrite all lecture notes within 24 hours of the lecture. Make sure all concepts are either clear to you or that you clarify all points of confusion with the course instructors or TA the same week as the relevant lectures.
- Work all problem sets until you are intimately aware of the solution path or manner in which to solve all questions.
- Take all of the quizzes. Your performance on quizzes will give you some insight into the concepts you understand and those that are confusing. Address confusing concepts right away by talking to the TA or the relevant course instructors. If you do not take a quiz during the scheduled time, there is no mechanism available to make up a quiz.
- In your final preparations for all exams, you should vigorously review/rehearse/rewrite your notes, review/rewrite all lecture slides, and rework all of the problem sets until you have an in-depth understanding of all concepts and the solutions to all problems. Consult the textbook, instructor-recommended websites, TA, or relevant instructors for any topics that remain confusing.
- When taking the exams, use a calculator/pencils and write legibly. Be prepared to budget your time wisely. Quickly answer all the questions for which you are confident, then go back

to address those questions for which you have less confidence. This will maximize your score.

If you can quickly answer all of the questions on the problem sets, you have an active knowledge of all of the concepts covered in the lectures, and you heed the exam-taking advice above, you will maximize your exam performance.

Please see http://go.vcu.edu/syllabus for important information regarding:

1. Campus emergency information
2. Class registration required for attendance
3. Honor System: upholding academic integrity
4. Important dates
5. Managing Stress
6. Mandatory responsibility of faculty members to report incidents of sexual misconduct
7. Military short-term training or deployment
8. Student conduct in the classroom
9. Student email policy
10. Student financial responsibility
11. Students representing the university – excused absences
12. Students with disabilities
13. Withdrawal from classes