

## HUMAN GENETICS (IB 141)

Summer 2019 Dr. Kelly Agnew

**Class meets:** VLSB 2060, M-Th 10:10am to 11:30am.      **email:** [agnewk@berkeley.edu](mailto:agnewk@berkeley.edu)

**Office hours:** M, W, VLSB 3007, 11:40am-12:30pm, or by appointment.

### TEXT:

- 1) Lewis, Ricki. 2015. *Human Genetics: Concepts and Applications*. 11<sup>th</sup> ed. McGraw Hill, New York.

This text is strongly recommended, but not required. I will be using the 11<sup>th</sup> edition, but there are other editions available. The page numbers may be different in other editions. The class schedule (see below) gives a rough outline of what chapters will be covered and when, but you do not need to know the extra material in the book that isn't presented in lecture. Use the book to review lecture topics and to clear up any confusion. Reviewing the text figures is a helpful way to study for exams.

- 2) I will also provide materials from the primary literature and from popular media, and will post them to bCourses. These may include readings, podcasts, or videos.

**GRADING:** A total of 360 points is possible for this course:

Best two of three exams during the semester	160 (80 X 2)
Comprehensive final exam	100
Questions over readings and problem sets	60
Best four of five unannounced quizzes	<u>40</u>
	360

The course grades are NOT curved. The grading scale will be as follows:

90% and above	A	(324 and above)
80%-89.9%	B	(288-323)
70%-79.9%	C	(252-287)
60%-69.9%	D	(216-251)
≤59.9%	F	(≤ 215)

Active discussion and participation may boost those individuals who are on grade borderlines to the next highest grade. Plus and minus letter grades will divide each grade bracket by thirds.

**EXAMS:** Exams are worth 80 points each and may include a variety of question types, including multiple choice, short answers, quantitative problems and essays. Some questions will require calculators; do not plan on using smart watches or your phones as a calculator. A standard calculator that is not internet-connected will be fine.

**MISSED CLASSES/EXAMS/QUIZZES/ASSIGNMENTS:** I take attendance in class and expect you to be there. Since only the two best exams during the semester count toward your grade, there are no makeup exams. I cannot change the date of the mandatory final exam for individual students.

If you practice a faith that does not allow you to attend class on high holidays, you must notify me by Wednesday, June 26<sup>th</sup> of any classes you will miss and make arrangements to complete the required work ahead of time.

Only your best four quiz scores will count toward your grade, so there are no makeups for missed quizzes, either. Quizzes are unannounced, and you will have about ten minutes to complete them.

Most of the assignments will be posted to bCourses, and they are only available for a set amount of time. Please make sure you give yourself plenty of time to complete the work; late assignments will not earn maximum points.

**CELL PHONES:** Please turn off your phones and put them away during class, unless I ask you to use them for quick calculations, photos, or quick polls.

I will probably ask one student in the front row to have their phone out, with volume off, in case we get campus-wide safety alerts that would require us to evacuate the building or shelter in place.

The class is 80 minutes long—we'll take a few minutes' break at some point during every non-exam class so you can get up and stretch, check your phones, etc.

If a student's cell phone use is distracting me or other students around them, I will have to start deducting points from the student's overall score.

You may use laptops or tablets for your reading material and to take notes, but I expect all students to be engaged with course material and to participate actively in the discussion.

**GENERAL INFORMATION:** Please help me learn to pronounce your name correctly, and forgive me and correct me if I get it wrong. If you prefer a personal pronoun (he, she, they) by which to be addressed, please let me know privately and I will do my best to model that for the entire class.

Genetics can be a really hard class. There is a lot of math, and a lot of learning vocabulary and complicated pathways. But Human Genetics is also full of issues at the intersection of ethics, technology, politics and society. Sometimes there isn't a "right answer."

Because the issues we will discuss in this class are sometimes extremely difficult and challenging, it is **ESSENTIAL** that students feel they are able to state their opinions freely. There will be times you strongly disagree with others in the class. In order to have a productive discussion, however, we will establish some "common ground" rules:

You can expect that your opinions will be questioned by others in the class. You must be prepared to defend an opinion, and be willing to accept that others will not agree.

You should also feel you are able to change your mind and restate your opinion without ridicule.

Additionally, I expect you to question others' opinions in the class, and you must be willing to accept that you may change your mind, or you will not agree, *but you will treat other persons in the class with respect and dignity.*

Occasionally, I may ask you to argue for an opinion with which you disagree in order to better understand the opposing position. Be prepared!

I expect you to adhere to the highest standards of academic honesty. Cheating on an exam or plagiarizing other people's work is unacceptable, may result in a failing grade, and will be dealt with by the Center for Student Conduct.

**STUDENTS WITH DISABILITIES:** If you need special accommodations, please make sure your documentation is on file with the DSP office, and that I've made contact with you during the first week of class to discuss accommodations.

**COURSE CAPTURE, SLIDES AND LECTURE NOTES:** We don't have video course capture for this course in the summer sessions. I try to post my slides a day or two after the lecture in which they were given; you can find them under the "Files" section of the course website on bCourses. I will use bCourses to email students and to post announcements, so check the site regularly. I don't post my lecture notes; students who take their own notes during class have done better in the course.

**COURSE/TEACHING EVALUATIONS:** At the end of the semester, you will have an opportunity to provide *anonymous* feedback online about the course and my teaching. These evaluations help me improve the course. Although I can't see the responses until after the semester is over, I *can* see the *response rate*. If the overall class response rate exceeds 80%, everybody in the class will earn an extra 2 points.

**TENTATIVE LECTURE SCHEDULE:** Dates and topics may change!

Date	General Topics	Assignments
Jun 24 <sup>th</sup>	Introduction, Ch.1 Why Genetics is Important	Assessment
Jun 25 <sup>th</sup>	Ch. 1 Genes, genomes, important terms	
Jun 26 <sup>th</sup>	Ch. 2 Cell biology review, cell cycle, mitosis	
Jun 27 <sup>th</sup>	Ch. 3 Meiosis, development, homeotic genes	Probability problems
Jul 1 <sup>st</sup>	Ch. 4 Mendelian Genetics	
Jul 2 <sup>nd</sup>	Ch. 4 Pedigrees, probability	
Jul 3 <sup>rd</sup>	Ch. 5 Lethal alleles, codominance, epistasis, pleiotropy, penetrance	Pedigree problems
Jul 4 <sup>th</sup>	<b>NO CLASS; INDEPENDENCE DAY!</b>	
Jul 8 <sup>th</sup>	mtDNA, Linkage, intro to GWAS	
Jul 9 <sup>th</sup>	Ch. 6 Sex Chromosomes, sex linkage, sex-influenced traits, mosaicism, imprinting	
Jul 10 <sup>th</sup>	Ch. 7 Polygenetic traits, GWAS	
Jul 11 <sup>th</sup>	<b>EXAM I</b>	
Jul 15 <sup>th</sup>	Ch. 9 DNA structure, history of discovery, Replication	
Jul 16 <sup>th</sup>	Ch. 9 PCR, sequencing; Ch. 10 Transcription, RNA types, RNA processing	
Jul 17 <sup>th</sup>	Ch. 10 Translation, the Code, protein processing, prions	DNA to protein problems
Jul 18 <sup>th</sup>	Ch. 11 Epigenetics, non-protein-coding and non-human DNA in our genomes	

Jul 22 <sup>nd</sup>	Ch. 12 Point mutations, mutation rate, indels, pseudogenes, DNA repair	
Jul 23 <sup>rd</sup>	Ch. 13 Chromosomal mutation, nondisjunction, telomeres, translocations, inversions	
Jul 24 <sup>th</sup>	<b>EXAM II</b>	
Jul 25 <sup>th</sup>	Ch. 14 Population Genetics, Hardy-Weinberg Equilibrium	Hardy-Weinberg
Jul 29 <sup>th</sup>	DNA Profiling, genetic privacy, case studies of Thomas Jefferson, Golden State Killer	
Jul 30 <sup>th</sup>	Ch. 15 Evolutionary Genetics, nonrandom mating, gene flow, genetic drift, natural selection	Huntington's Chorea
Jul 31 <sup>st</sup>	Ch. 15 Balanced polymorphism, Eugenics	
Aug 1 <sup>st</sup>	Ch. 16 Human Evolution: archaic humans, Neanderthals, Denisovans, mitochondrial Eve, human migrations	
Aug 5 <sup>th</sup>	Ch. 18 Genetics of Cancer	
Aug 6 <sup>th</sup>	Ch. 19 Genetic Technologies: patents, microarrays, transgenic organisms, CRISPR	CRISPR podcast
Aug 7 <sup>th</sup>	Ch. 20 Genetic counseling, gene therapy	
Aug 8 <sup>th</sup>	<b>EXAM III</b>	
Aug 12 <sup>th</sup>	Ch. 21 Savior Siblings, reproductive technologies	
Aug 13 <sup>th</sup>	Ch. 22 Genomics	
Aug. 14 <sup>th</sup>	Review, Evaluations	
Aug. 15 <sup>th</sup>	<b>COMPREHENSIVE FINAL EXAM</b>	

### Ethical Issues in Human Genetics

This is a list of some of the topics we may cover during the course. Read through it and note those that you find particularly interesting. I'll ask you early in the class for your favorites, and I'll try to prioritize those topics.

1. *Gene therapy/germ line therapy*—How does gene therapy work? How are the genes introduced into the body? What's the difference between somatic therapy and germ line therapy? What is the current status of gene therapy trials and treatments in the United States?

2. *Stem cells*—Where do stem cells come from? Why are they so powerful and what promise do they hold? Why is there a debate about stem cell lines? Which states in the U.S. are pushing stem cell therapies forward and what has it meant for research in those states? What are other nations doing?

3. *Genetic information privacy*—Should insurance companies have access to your genetic information? Should your employer? If you are part of a study group, like the man who was discovered to be immune to HIV, do you own your genetic information? Who gets to patent

your genes? Some nations, like Iceland, have generated huge genetic databases of their citizens. Should companies have access to that information? Did the citizens know they were giving information that could be used for research companies? How should we ensure consent has been given?

4. *Federal and state criminal DNA databases/ inmate exoneration*—some death row and life – sentence inmates have been exonerated through DNA evidence. In Louisiana, appeals courts gave a man who had been in prison for twenty years access to DNA testing ONLY if he waived the right to sue the state for wrongful incarceration if he were found innocent. The DNA evidence excluded the prisoner as the criminal. How powerful is DNA evidence? What safeguards need to be taken to ensure it is reliable? How is DNA evidence used to identify the remains of soldiers who have been dead thirty years, or victims of genocide in Guatemala and Rwanda?

5. *Direct-to-Consumer DNA tests*—are they accurate and what can they tell you? What are the companies doing with your data? Do biological parents of adopted children have the right to remain anonymous? Should law enforcement agencies be allowed to use data to capture and convict genetic relatives suspected of crimes? (The Golden State Killer case and the UC Berkeley employee case will be discussed in class).

6. *Specific gene cloning, human therapeutic cloning and human whole embryo cloning*—What is the difference between cloning a gene for insulin to provide drugs for diabetics, cloning human stem cells for tissue transplants, and cloning a whole human being? What are the pitfalls of whole-organism cloning? What nations have already cloned or are pursuing human cloning?

7. *Trans-species cloning*—Is it ethical to put flounder genes into a strawberry so that the fruit is more frost-tolerant? What about human genes into a pig to produce a human protein for hemophiliacs?

8. *The role of pre-natal genetic sex and genetic disease testing and the social consequences*—How does sex-choosing technology work? How much does it cost and should insurance cover the cost? Should it be allowed when parents know they carry a male-linked disease, like hemophilia? Should society bear the financial responsibility for children who are born to couples knowing that the child will be born with severe mental and physical defects? When should genetic screening be allowed? For disease? For height?

9. *CRISPR/Cas and gene editing*—How does the CRISPR/Cas system work? What are the potential applications of the system, and why does it raise concerns among the scientific community? How do major scientific journals handle submissions on papers that raise ethical concerns?

10. *Eugenics*—What is the history of eugenics in the United States and abroad? What legislation was passed, and in some cases, remains on the books, to promote eugenics? How do modern countries view eugenics and how are those views shaped by history? Mathematically, does eugenics work? How many generations are needed to remove “undesirable” alleles? Is there such a thing as an undesirable allele?

11. *Assisted reproduction*—How do these technologies work? Is it responsible for IVF clinics to re-implant more than 2 fertilized eggs? What happens to the embryos that aren't implanted? Do the genetic parents of embryos have joint custody? What happens if couples divorce? Is it responsible to use IVF technology to help a 55-year old woman conceive a child? Should insurance companies cover the costs?

12. *Assisted reproduction, cont.* —“Wombs for rent” and social justice issues. Is paying women prepared to be surrogate mothers unacceptable exploitation? Should governments play a role in regulating this practice when the parents and the surrogate are in different countries? What if the woman's country or society doesn't approve of the genetic parents because of their race, nationality, or sexual orientation?

13. *The human animal* —Is race biologically meaningful? How do we test that? Is prejudice genetic? How does cooperation evolve? What can science tell us about the evolution of prejudice, sexual attraction, aggression, and vengeance? What are potential problems with some of the studies that claim to explain some human behaviors?

14. *Bio-ownership*: who owns the stuff of life? What are immortal cell lines? *HeLa* cells and Henrietta Lacks. The *Mo* cell line and the University of California lawsuit. Bioprospecting vs. Biopiracy. Patents on BRCA1 and BRCA2, and “Round-Up Ready” traits.