ANG 6532 (2B73)/ ANT 4531 (2B74) MOLECULAR GENETICS OF DISEASE

Prof. Connie J. Mulligan

Class meets in 230 Florida Gymnasium Class time: Tuesday, periods 4-6, 10:40am-1:40pm, with a 15 min break

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Office hours: Tuesday, 8:30-10:30 in person in B119 Turlington, or in person/by Zoom in 409 Genetics Institute

Course summary: The availability of whole genome sequences, development of high-throughput sequencing platforms, and public databases of genetic variants have greatly accelerated the discovery of genes involved in disease, leading to breakthroughs in diagnosis and treatment. We will discuss single-gene recessive disorders (e.g. myotonic dystrophy) and complex diseases (e.g. diabetes and Alzheimer's disease). We will discuss methods used to isolate genes involved in disease as well as genetic testing and types of diagnosis and treatment. We will also discuss pharmacogenomics and epigenetics as well as the ethical, legal and social issues associated with genetic investigation of human health and disease.

Course design: This course is intended for advanced undergraduate and graduate students who have an interest in the molecular genetics of disease. Most human genetics courses spend only 1-2 weeks/semester discussing the genetics of disease, whereas this course focuses on disease for the entire semester. We will discuss the basics of the human genome and the basis of simple and complex diseases, including cause, inheritance, diagnosis, and treatment. We will discuss a number of diseases in detail, with the specifics determined by the state of research for a particular disease, i.e. certain eye diseases are now being treated by gene therapy so we will focus on gene therapies for that day, but will focus on the use of microarrays to diagnose different brain tumors on another day. Throughout the semester, guest lecturers (mainly from the medical school) will speak about their research on the diseases that we discuss in detail in class.

The course is intended for students from all colleges and departments – in the past, I have had students from anthropology, chemistry, English, molecular genetics and microbiology, pharmacology and zoology to name a few. A diverse audience makes for a more interesting class since everyone has different backgrounds, different perspectives and different interests to contribute to class discussions. Active participation is one of the strengths of the class. It also helps to have a class with undergraduate and graduate students together to bring diverse perspectives to the class. I keep separate sets of grades for the undergraduate and graduate classes so that each set of students is only compared to his/her cohort.

Course objectives and student goals: The class is fairly intense and demanding because knowledge across a broad range of fields is fundamental to an exploration of the molecular genetics of human disease – in fact, this course may be one of the most challenging courses you have taken with respect to developing expertise and familiarity with a wide range of materials, techniques, publications, etc. All students are expected to do all of the required readings and to follow up with additional readings if something is not understood. Additional readings are listed at the end of each chapter in the textbook and can be found in the references in the journal articles. Furthermore, students should become familiar with searching PubMed or Google Scholar for supplementary, follow-up, or original readings. Finally, there are suggested textbooks to provide additional and supplementary material. Although basic molecular biological concepts will be reviewed, some knowledge of Mendelian genetics and molecular biology (such as Intro Bio, BCS 2011) and a willingness to immerse yourself in the data are expected. Course objectives include:

- Learn about the molecular genetic basis of common simple and complex diseases
- Gain knowledge about the latest molecular genetic techniques to study, diagnose, and treat simple and complex diseases
- Learn about research being conducted at UF on the molecular genetics of certain diseases
- Discuss ethical, legal, and social issues associated with the diagnosis and treatment of diseases

Reading materials: There are two required reading sources - a textbook and journal articles (available on the E-Learning course website). The main textbook for the course is <u>Genetics and Genomics in Medicine</u> by Strachan, Goodship & Chinnery. Related journal articles and news-and-views articles will also be discussed and are available on the E-Learning course website. Supplemental texts (for additional background on molecular genetics and more information on genetic diseases, specifically cancer) include <u>How the Human Genome Works</u> by Edwin H. McConkey, <u>Medical Genetics</u> by Lynn Jorde et al. and <u>Medical Genetics at a Glance</u> (very brief and short-hand-written) by Pritchard and Korf. All textbooks are available at campus bookstores and through online sources such as amazon.com and half.com. In general, at least one journal article will be discussed for each disease that we discuss in detail. If students know of additional articles or diseases that they would like to discuss, please contact me.

Course communications: I can be reached by email and through the messaging ("Inbox") tool in E-Learning

Course format: The course meets once a week for three hours and the course format is in-person lecture (by me and by guest lecturers) plus discussion with substantial class participation. Attendance via Zoom is not available. Evaluation of student performance is based on class participation, weekly quizzes, team discussions, written exercises, and an exam.

Grading: Final grades will be determined by the following five categories: participation (100 pts), weekly quizzes (100 pts), team-based discussions (100 pts), homework exercises (100 pts total), and one exam (100 pts). Possible points total 500. Grades will be based on the following point percentages: 93-100%=A, 90-92.99%=A-, 87-89.99%=B+, 83-86.99%=B, 80-82.99%=B-, 77-79.99%=C+, 73-76.99%=C, 70-72.99%=C-, 67-69.99%=D+, 63-66.99%=D, 60-62.99%=D-, <60%=E. The university grading policy can be found at https://catalog.ufl.edu/ugrad/current/regulations/info/grades.aspx.

- Participation is required of all students and will be based on each student's contribution of original comments, questions, etc. to the class. Simply showing up for class does not constitute participation. Punctuality is important and participation points will not be awarded to students who are late to class.
- There will be weekly **quizzes**, to be taken in class, during the semester. The quizzes consist of multiple choice questions administered through E-Learning and are intended to ensure that you are doing the readings every week and to help prepare you for the exam. The lowest score on your quizzes will be dropped. Missed quizzes cannot be made up.
- There will be 2-4 team-based discussions. Teams will be determined by the instructor and the discussion exercise will take place in class. Teams will be given issues or questions to discuss based on the week's reading and will present a short (~2 min) summary of their discussion to the class. Grades will be based on participation as determined by team members. Missed team-based discussions cannot be made up.
- There will be 2 homework **exercises** involving the analysis of hypothetical or real molecular data. The exercises are intended to give students an opportunity to perform analyses similar to those conducted in the discussion papers and will be graded on effort and accuracy. Exercises must be turned in two weeks after they are assigned. Students who miss a class in which an exercise is assigned are expected to make up any missed material and to turn in exercises on time, i.e. two weeks after the exercise was assigned.
- There will be 1 **exam** that will consist of essay and multiple choice questions. Questions must be answered during the exam period without reference to books or notes of any kind.

Strategies for success:

- "Learning is not a spectator sport. Fundamentally, the responsibility to learn is yours and yours alone. For learning to happen in any course, you must take an active role in the process. For our class, you are expected to come to class 'prepared' and 'ready to learn', which requires you to read and to study the assigned reading before you come to class." Excerpted from Romack 2010, Enhancing Students' Readiness to Learn, Faculty Focus Special Report: 11 Strategies for Getting Students to Read What's Assigned.
- Furthermore, to get the most out of class, you must arrive on time (5 minutes late is not on time) and you must not use computers for non-class-related activities or use cell phones during class. While you may think that you can multi-task and follow the lecture while playing on your phone, you will definitely learn less than if you give the class your undivided attention. Finally, punctuality is a show of respect for your instructor and classmates and is important not just in class but in a job and your future career.
- It is important to complete all the readings on time and it is best to read the readings throughout the week. In this way, you have time to think about and process the information during the week and in between different readings. Ideally, you would read some every night of the week. The amount of reading material is very modest, particularly for an anthropology course. This is because it is expected that you will re-read anything you do not understand the first time. I often re-read journal articles and find myself discovering things that I completely missed on the first read. Or new items will come to light after having read a different, but related article. This is the intent of the readings that they relate to each other and increase your knowledge and expertise in a synergistic way. During your reading, you should take notes and these notes give you material to ask questions and contribute to class discussions. Use the quizzes at the end of each chapter in the textbook to ensure you have understood the material.
- You have two weeks to work on the exercises. The intent is that you will use the first week to start on the exercise and come to class the next week (i.e. the week before the exercise is due) with any questions you have about the exercise. Do not wait until the second week to start the exercise. I am always available by email to ask questions about the exercises.
- Finally, you should review material throughout the course rather than trying to cram everything in the night before the exam. Ask for help in taking notes, comprehending the material, or preparing any of the written or oral assignments I am available during office hours, you can schedule an appointment outside of office hours, and I am always available by email.

Useful websites:

Although basic molecular biological concepts will be reviewed, some knowledge of DNA structure, Mendelian genetics, and molecular biology is expected (such as Intro Bio, BCS 2011). Useful information is provided by several websites:

https://elearning.ufl.edu - access to E-Learning course website

<u>http://www.uflib.ufl.edu</u> - UF database of available electronic journals (may only be available from a campus computer)

https://pubmed.ncbi.nlm.nih.gov/ - National Library of Medicine database of over 11 million journal articles dating back to the 1960s

<u>https://gold.jgi.doe.gov/</u> - Genome OnLine Database, database of genome sequencing projects <u>http://www.genome.gov/LegislativeDatabase</u> - Database of federal and state laws focused on genetic issues, such as genetic testing and counseling, insurance and employee discrimination, etc.

Class attendance policy: Because the class meets only one time per week and because the class format is mainly discussion, it is very difficult to make up missed classes by borrowing notes, watching lectures, etc. Therefore, students are strongly encouraged to attend all classes. Attendance via Zoom is not available. If you do miss class, you will only be allowed to make up work if you provide documentation of the reason you missed class (e.g. ER visit, family emergency) and only at my discretion. Computers should be used sparingly in class. In a seminar format, it is more important to participate in class discussions than to record everything on your

computer. Additionally, it can be very off-putting for me or a guest lecturer to lecture to a sea of laptop backs. All cell phones must be turned off during class and should not be seen, i.e. no texting, checking the time, etc.

In Class Recording: Students are allowed to record video or audio of class lectures. However, the purposes for which these recordings may be used are strictly controlled. The only allowable purposes are: (1) for personal educational use, (2) in connection with a complaint to the university, or (3) as evidence in, or in preparation for, a criminal or civil proceeding. All other purposes are prohibited. Specifically, students may not publish recorded lectures without the written consent of the instructor. A "class lecture" is an educational presentation intended to inform or teach enrolled students about a particular subject, including any instructor-led discussions that form part of the presentation, and are delivered by any instructor hired or appointed by the University of Florida, or by a guest instructor, as part of a University of Florida course. A class lecture does not include student presentations, academic exercises involving solely student participation, assessments (quizzes, tests, exams), private conversations between students in the class or between a student and the faculty or lecturer during a class session. Publication without permission of the instructor is prohibited. To "publish" means to share, transmit, circulate, distribute, or provide access to a recording, regardless of format or medium, to another person (or persons), including but not limited to another student within the same class section. Additionally, a recording, or transcript of a recording, is considered published if it is posted on or uploaded to, in whole or in part, any media platform, including but not limited to social media, book, magazine, newspaper, leaflet, or third party note/tutoring services. A student who publishes a recording without written consent may be subject to a civil cause of action instituted by a person injured by the publication and/or discipline under UF Regulation 4.040 Student.

Copyright information: The contents of the syllabus, lectures, lecture outlines, and handouts for this course are copyrighted and intended for the private use of students registered in ANG 6532/ANT 4531. These materials, therefore, cannot legally be reproduced, in part or in whole, by any commercial enterprise or for any commercial purposes.

Accommodations for students with disabilities: Students requesting accommodation for disabilities must first register with the Dean of Students Office (DSO). The DSO will provide documentation to the student who must then provide this documentation to the instructor when requesting accommodation. The student must submit this documentation prior to submitting assignments or taking the quizzes. Accommodations are **not retroactive**; therefore, students should contact the office as soon as possible in the term for which they are seeking accommodations.

University policy on academic misconduct: Academic honesty and integrity are fundamental values of the University of Florida community. Students should be sure that they understand the UF Student Honor Code. **Students caught cheating or plagiarizing on any assignment in this class will be reported to the Dean of Students and receive a zero for the assignment.**

Academic honesty: As a result of completing the registration form at the University of Florida, every student has signed the following statement: "I understand that the University of Florida expects its students to be honest in all their academic work. I agree to adhere to this commitment to academic honesty and understand that my failure to comply with this commitment may result in disciplinary action up to and including expulsion from the University."

The latest student honor code and student conduct code can be found at: <u>https://sccr.dso.ufl.edu/policies/student-honor-code-student-conduct-code/</u>

The following presentation from the University of Florida Writing Center has essential information concerning plagiarism and properly citing sources. Be sure to note that the University of Florida does consider self-plagiarism to be plagiarism.

http://mediasite.video.ufl.edu/mediasite/Viewer/?peid=adaa44500eaf460a84f238e6b9a558f9 . All students enrolled in this course are expected to view this presentation and refer back to it as questions arise. Please let me know if you have any questions.

UF Counseling Services: On-campus services are available for students having personal problems or lacking clear career and academic goals. They include:

- 1. University Counseling Center, 301 Peabody Hall, 392-1575, personal and career counseling
- 2. Career Resource Center, Reitz Union, 392-1601, career development assistance and counseling Student Mental Health, Student Health Care Center, 392-1171, personal counseling
- 3. Counseling and Wellness Center, (352) 392-1575, 3190 Radio Rd, personal counseling
- 4. Sexual Assault Recovery Services (SARS), Student Health Care Center, 392-1161, sexual assault counseling
- 5. Additionally, student web-based resources on sexual harassment are available at <u>http://www.ufsa.ufl.edu/students/sh/sexualharassment.shtml</u>

U Matter We Care

Your well-being is important to the University of Florida. The U Matter, We Care initiative is committed to creating a culture of care on our campus by encouraging members of our community to look out for one another and to reach out for help if a member of our community is in need. If you or a friend is in distress, please contact <u>umatter@ufl.edu</u> or 352-294-CARE (2273) so that the U Matter, We Care Team can reach out to the student in distress. The U Matter, We Care Team can help connect students to the many other helping resources available including, but not limited to, Victim Advocates, Housing staff, and the Counseling and Wellness Center. Please remember that asking for help is a sign of strength. In case of emergency, call 9-1-1.

Course schedule (some guest speakers and associated readings are still being confirmed):

January 11 – Week 1 - First day of classes

Introduction

Plagiarism

- "The baby was gravely ill. Rapid sequencing turned around a diagnosis in 13 hours and pointed to a treatment", Joseph, 2021 STAT, <u>https://www.statnews.com/2021/07/22/rapid-sequencing-baby-diagnosis-13-hours/</u>
- "Arthritis is the price for our ancestors surviving the ice age, say scientists", The Telegraph, July 3, 2017, <u>https://uk.news.yahoo.com/arthritis-price-ancestors-surviving-ice-150000950.html</u>
- "Why can some kids handle pressure while others fall apart?" Bronson & Merryman, NYT, Feb 6, 2013, <u>http://www.nytimes.com/2013/02/10/magazine/why-can-some-kids-handle-pressure-while-others-fall-apart.html</u>
- "The link between mental illness and physical illness", The Daily Dose, Apr 11, 2017, <u>http://www.ozy.com/acumen/the-link-between-mental-illness-and-physical-</u> <u>illness/76671?utm_source=dd&utm_medium=email&utm_campaign=04112017&variable=</u> <u>7484b1138a1020da7084a14dc20bdfa5</u>
- "Senator's thesis turns out to be remix of others' work, uncited", NYT, July 23, 2014, https://www.nytimes.com/2014/07/24/us/politics/montana-senator-john-walsh-plagiarized-

<u>thesis.html</u>. ***Walsh's degree was ultimately revoked by the Army War College and he dropped out of the US Senate race because of the plagiarism charges.

January 18 - Week 2 - Introduction to genetics

- Lecture
 - Background material
 - How to read a scientific article
 - PubMed search

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpts 1 & 2 (Fundamentals of DNA, Chromosomes, and Cells AND Fundamentals of Gene Structure, Gene Expression, and Human Genome Organization)
- E-Learning:
 - *The influence of evolutionary history on human health and disease, Benton et al. 2021, *Nature Reviews Genetics*, 22:269-283.
 - *The legacy of the human genome project, Rood and Regev, 2021, *Science*, 373:1442-1443

January 25 – Week 3 – Methods – DNA cloning, sequencing and microarrays Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 3 (Principles underlying core DNA technologies some old stuff at the beginning of the chapter)
- E-Learning:
 - *Linking genome variants to disease: Scalable approaches to test the functional impact of human mutations, Findlay, 2021, *Hum Molec Genet*, 30:R187-R197.\
 - Is a racially-biased algorithm delaying health care for one million Black people? Madhusoodanan, 2020, *Nature*, <u>https://www.nature.com/articles/d41586-020-03419-6?WT.ec_id=NATURE-20201224&utm_source=nature_etoc&utm_medium=email&utm_campaign=20201224&sap-outbound-id=18BF30523AC94F238436868F4202EF1246922458
 </u>

Guest speaker – Dr. Kiley Graim, Assistant professor, Dept of Computer & Information Science & Engineering, College of Engineering – bioinformatics & personalized medicine – invited

February 1 – Week 4 – Genetic variation

- Genetics and Genomics in Medicine (GGM), Chpt 4 (Principles of genetic variation)
- E-Learning:
 - *The role of the microbiota in human genetic adaptation, Suzuki and Ley, 2020, *Science*, 370: eaaz6827
 - *Common gene variants found among psychiatric disorders, Olena, 2018, The Scientist, <u>https://www.the-scientist.com/news-opinion/common-gene-variants-found-among-psychiatric-disorders--64244</u>
 - *Genetic mutations you want, Williams, 2016, PNAS, 113:2554-2557

• *Exploring people's thoughts about the causes of ethnic stereotypes, Schmalor et al., 2021, *PLoS ONE*, 16(1): e0245517

Team-based discussion #1

February 8 - Week 5 - Monogenic disorders

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 5 (Single-gene disorders: Inheritance patterns, phenotype variability, and allele frequencies
- E-Learning:
 - *A genetic revolution in rare disease medicine, Alkuraya, 2021, *Nature*, 590:218-219
 - *A brief history of human disease genetics, Claussnitzer et al., 2019, *Nature*, 577:179-189
 - *Alpha1-antitrypsin deficiency, Greene et al. 2016, *Nature Reviews*, 2:1-17

Guest lecturer - Dr. Mark Brantly, Professor, Dept of Medicine and Molecular Genetics and Microbiology – alpha 1-antitrypsin deficiency – confirmed at 10:40

February 15 – Week 6 – Gene regulation and epigenetics Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 6 (Principles of gene regulation and epigenetics)
- E-Learning:
 - *Epigenetics of aging and disease: A brief overview, Pagiatakis et al. 2021, Aging Clin Exp Res, 33:737-745
 - "A patchwork mind: How your parents' genes shape your brain", Scientific American, 2009, <u>http://www.scientificamerican.com/article.cfm?id=a-patchwork-mind</u>

Exercise 1 assigned – due in 2 weeks

Guest speaker – Dr. Jim Resnick, Professor, Dept of Molecular Genetics and Microbiology – imprinting, genetics of Prader-Willi and Angelman syndromes – confirmed

February 22 – Week 7 – Complexities with monogenic disorders

- *Genetics and Genomics in Medicine* (GGM), Chpt 7 (Genetic variation producing diseasecausing abnormalities in DNA and chromosomes)
- E-Learning:
 - *Longitudinal increases in somatic mosaicism of the expanded CTG repeat in myotonic dystrophy type 1 are associated with variation in age-at-onset, Morales et al., 2020, *Hum Mol Genet*, 29:2496-2507
 - "24-year-old researches treatment for her own crippling disease: "I'm in a race against time", People, Dec 2, 2021, <u>https://people.com/health/24-year-oldresearches-treatment-for-her-own-crippling-disease-im-in-a-race-against-time/</u>
 - Friedreich ataxia fact sheet <u>https://www.ninds.nih.gov/Disorders/Patient-</u> Caregiver-Education/Fact-Sheets/Friedreichs-Ataxia-Fact-Sheet#3070_4

Guest speaker - Dr. Maury Swanson, Professor, Dept of Molecular Genetics and Microbiology – myotonic dystrophy – invited

March 1 - Week 8 - Complex diseases, part I

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 8, pp 247-278 + Box 8.5, pp 290-291 (Identifying disease genes and genetic susceptibility to complex diseases)
- E-Learning:
 - *Deep genealogy and dilution of risk, Roberts, 2013, *PLoS Biology*, 11(9): e1001660
 - *New concerns raised over the value of genome-wide disease studies, Nature, 2017
 - *Costly Genes, Karayiorgou, 2020, PLOS Genetics, https://doi.org/10.1371/journal.pgen.1008889
 - *Overcoming translational barriers impeding development of Alzheimer's disease modifying therapies, Golde, 2016, J Neurochemistry, 139:224-236

Guest speaker – Dr. Todd Golde, Director, Center for Translational Research in Neurodegenerative Disease; Professor, Dept of Neuroscience – Alzheimer's disease - invited

Exercise 1 due

March 8 – Week 9 - Spring break

March 15 – Week 10 – Complex diseases, part II

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 8, pp 278-308 (Identifying disease genes and genetic susceptibility to complex diseases)
- E-Learning:
 - *Genetic loci and novel discrimination measures associated with blood pressure variation in African Americans living in Tallahassee, Quinlan et al, 2016, *PLoS ONE*
 - *Methylation changes at NR3C1 in newborns associate with prenatal stress exposure and newborn birth weight, Mulligan et al, 2012, *Epigenetics*, 7:853-857
 - *Genetic variation across phenotypic severity of autism, Toma, *Trends in Genetics*, 36:228-231
 - *Sex differences in immune responses to viral infection, Offord, 2021, The Scientist, <u>https://www.the-scientist.com/features/sex-differences-in-immune-responses-to-viral-infection-68466</u>

Team-based discussion #2

March 22 (AABA meeting) - Week 11 - Genetic treatments, Part I

- *Genetics and Genomics in Medicine* (GGM), Chpt 9, pp 309-317 AND 336-371 (Genetic approaches to treating disease)
- E-Learning:

- *Tweaking genes with CRISPR or viruses fixes blood disorders, Kaiser, 2020, *Science*, 370:1254-1255.
- *Mutation-independent rhodopsin gene therapy by knockdown and replacement with a single AAV vector, Cideciyan et al. 2018, *PNAS*, doi/10.1073/pnas.1805055115
- "In breakthrough, scientists edit a dangerous mutation from genes in human embryos", NY Times, Aug 2, 2017, https://www.nytimes.com/2017/08/02/science/gene-editing-humanembryos.html?emc=edit_th_20170803&nl=todaysheadlines&nlid=55234026

Guest speaker – Prof. Al Lewin, Professor, Dept of Molecular Genetics and Microbiology, College of Medicine – Gene therapy and eye disease - Confirmed

March 29 – Week 12 – Genetic treatments, Part II

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 9, pp 317-336 (Genetic approaches to treating disease)
- E-Learning:
 - *Pharmacogenomics in the clinic, Relling and Evans, 2015, Nature, 526:343-350
 - *For better drugs, diversify clinical trials, Bumpus, 2021, *Science*, <u>371:570-571</u>

Exercise 2 (LOD scores) assigned - due in 2 weeks

Guest speaker - Dr. Larisa Cavallari, Associate Professor, Dept of Pharmaceutical Sciences and Director, UF Center for Pharmacogenomics – Pharmacogenomics – invited

April 5 – Week 13 - Cancer

Reading assignments:

- Genetics and Genomics in Medicine (GGM), Chpt 10 (Cancer genetics and genomics)
- E-Learning:
 - A cancer legacy, Once viewed as tragic anomalies, many childhood cancers may have their roots in inherited mutations, Couzin-Frankel, 2016, *Science* 351: 440-443
 - Relative risk: Mutations in BRCA genes predispose women to cancer, but outside influences shape the ultimate risk, Velasquez-Manoff, 2015, *Nature*, 327:5116-5117
 - "What if everything your doctors told you about breast cancer was wrong?", Mother Jones, Aschwanden, 2015, <u>http://www.motherjones.com/politics/2015/10/faulty-research-behind-mammograms-breast-cancer/</u>
 - Optional Deciphering intratumor heterogeneity using cancer genome analysis, Ryu et al. 2016, *Hum Genet*, 135:635-642

Team discussion #3

April 12 - Week 14 - Genetic testing

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 11 (skim methods, like pp 435-450, 455-457) (Genetic testing from genes to genomes, and the ethics of genetic testing and therapy)

- E-Learning:
 - *Uncertainty in genomics impacts precision medicine, Pyeritz, 2021, *Trends in Genetics*, 37:711-716.
 - *Project Baby Bear: Rapid precision care incorporating rWGS in 5 California children's hospitals demonstrates improved clinical outcomes and reduced costs of care, Dimmock et al., 2021 *Am J Hum Genet*, 108:1231-1238.
 - *Screening embryos for complex genetic traits called premature, Kaiser, 2019, *Science*, 366:405-406.
 - *Who has your DNA, or wants it?", Kaiser, 2015, *Science*, 349:1475.
 - "A geneticist's research turns personal", NYT, June 2, 2012, <u>https://www.nytimes.com/2012/06/03/business/geneticists-research-finds-his-own-diabetes.html</u>
 - "Researchers to return blood samples to the Yamomamo", Science, June 4, 2010

Team discussion #4

Exercise 2 due

April 19 - Week 15 – Exam