ANG 6532 (2G41)/ ANT 4531 (2G33) MOLECULAR GENETICS OF DISEASE

Prof. Connie J. Mulligan

Class meets in 120 Fine Arts C
Class time: Wednesday, periods 5-7, 11:45-2:45, with a 15 min break

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Course summary: This course will examine the molecular genetics of disease in humans. The completion of the human genome sequence, the availability of high-throughput, 'next-generation' 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 a range of genetic disorders from single-gene recessive defects (e.g. myotonic dystrophy and alpha 1-antitrypsin deficiency) to complex diseases (e.g. diabetes). We will discuss the methods used to isolate genes involved in disease (e.g. GWAS and whole exome or genome sequencing) as well as genetic testing and types of diagnosis and treatment (e.g. microarrays and gene therapy). Other topics such as pharmacogenomics and epigenetics will be discussed. We will also discuss 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. Contrary to what one might think, undergrads are often as well prepared and as insightful as graduate students. 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: All students are expected to gain knowledge on the molecular genetic basis of simple and complex diseases in general and the specific genetic basis for a number of particular human diseases. 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.

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 Genetics and Genomics in Medicine 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 How the Human Genome Works by Edwin H. McConkey, Medical Genetics by Lynn Jorde et al. and Medical Genetics at a Glance (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 format: The course meets once a week for three hours and the course format is lecture (by me and by guest lecturers) plus discussion with substantial class participation. Evaluation of student performance is based on class participation, written assignments, 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, 90-93%=A-, 87-90%=B+, 83-87%=B, 80-83%=B-, 77-80%=C+, 73-77%=C, 70-73%=C-, 67-70%=D+, 63-67%=D, 60-63%=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 several multiple choice and/or short answer questions 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 can not be made up.
- There will be 2-4 **team-based discussion** exercises. Teams will be determined by the instructor and the discussion exercise will take place in class. Teams will be given discussion points or questions 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 approximately 2-4 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 short answer questions and exercises (similar to the homework exercises). Questions must be answered during the exam period without reference to books or notes of any kind. Calculators may be used, but no personal communication devices will be allowed.

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 eventual 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 an exponential way. During your reading, you should take copious notes and these notes will form the basis of the commentary/questions you will turn in. 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://lss.at.ufl.edu – access to e-Learning course website
http://www.uflib.ufl.edu - UF database of available electronic journals (may only be available from a campus computer)
http://www.ncbi.nlm.nih.gov/PubMed - National Library of Medicine database of over 11 million journal articles dating back to the 1960s
http://www.genome.gov/glossary.cfm – NIH-maintained glossary of genetic terms
http://www.genomesonline.org/ - status of genome sequencing projects
www.hgmd.cf.ac.uk/ac/index.php – Human Gene Mutation Database, database of genetic variants that cause disease, maintained by Cardiff University, UK
www.pitt.edu/~super1 – more than 2600 free lectures on public health and disease
http://cgap.nci.nih.gov/Chromosomes/Mitelman - Database of chromosome aberrations in cancer
http://www.genome.gov/LegislativeDatabase - 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 my lecture, etc. Therefore, students are strongly encouraged to attend all classes. 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. Plus it can be very off-putting for me or a guest lecturer to lecture to a sea of laptop backs. Class discussions/lectures cannot be recorded in any manner without special permission. All cell phones must be turned off during class and should not be seen, i.e. no texting, checking the time, etc.

Top Hat: We will be using the Top Hat (www.tophat.com) classroom response system in class. You will be able to submit answers to in-class questions using Apple or Android smartphones and tablets, laptops, or through text message. You can visit the Top Hat Overview (https://success.tophat.com/s/article/Student-Top-Hat-Overview-and-Getting-Started-Guide) within the Top Hat Success Center which outlines how you will register for a Top Hat account, as well as providing a brief overview to get you up and running on the system. An email invitation will be sent to you by email. Top Hat will require a paid subscription, and a full breakdown of all subscription options available can be found here: www.tophat.com/pricing. Should you require assistance with Top Hat at any time, due to the fact that they require specific user information to troubleshoot these issues, please contact their Support Team directly by way of email (support@tophat.com), the in app support button, or by calling 1-888-663-5491.

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Accommodations for students with disabilities: If you require accommodation due to a disability, please make an appointment or visit during my office hours so that we may discuss your needs. Students requesting classroom accommodation must first register with the Dean of Students Office. The Dean of Students Office will provide documentation to the student who must then provide this documentation to the Instructor when requesting accommodation.

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."

An excellent website that discusses plagiarism, correct citing of references and correct use of quotes is http://mediasite.video.ufl.edu/mediasite/Viewer/?peid=adaa44500eaf460a84f238e6b9a558f9. All students should read this material at least once. Remember that the university considers self-plagiarism to be plagiarism.

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. Student Mental Health, Student Health Care Center, 392-1171, personal counseling
3. Sexual Assault Recovery Services (SARS), Student Health Care Center, 392-1161, sexual assault counseling
4. Career Resource Center, Reitz Union, 392-1601, career development assistance and counseling
5. Additionally, student web-based resources on sexual harassment are available at http://www.ufsa.ufl.edu/students/sh/sexualharassment.shtml

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

umatter@ufl.edu so that the U Matter, We Care Team can reach out to the student in distress. A nighttime and weekend crisis counselor is available by phone at 352-392-1575. 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)**:**

August 23 – First day of classes
Introduction
Plagiarism
Video – Cracking your Genetic Code, NOVA PBS - http://ufl.kanopystreaming.com/video/nova-cracking-your-genetic-code
Reading assignments:

- Arthritis is the price for our ancestors surviving the ice age, say scientists, The Telegraph, July 3, 2017, https://uk.news.yahoo.com/arthritis-price-ancestors-surviving-ice-150000950.html
- "Why can some kids handle pressure while others fall apart?" Bronson & Merryman, NYT, Feb 6, 2013, http://www.nytimes.com/2013/02/10/magazine/why-can-some-kids-handle-pressure-while-others-fall-apart.html

August 30 – Week 2 – Basics of genetics
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:
  - Ancestry and disease in the age of genomic medicine, Rotimi and Jorde, 2010, NEJM, 363:1551-1558
  - Our fallen genomes, Macosko & McCarroll, Science, 2013, 342:564-565
  - Harmful mutations can fly under the radar, Science, 2014, 345:1438-1439, http://www.sciencemag.org/content/345/6203/1438.full
  - "The continuing evolution of genes", NYT, April 28, 2014, https://www.nytimes.com/2014/04/29/science/the-continuing-evolution-of-genes.html
  - "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-thesis.html
  - Optional - Human genetic basis of interindividual variability in the course of infection, Casanova, 2015, PNAS, 112:E7118-E7127, http://www.pnas.org/content/112/51/E7118.full (really interesting article on infectious disease)

Lecture

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

Sept 6 – Week 3 – Methods – DNA cloning, sequencing and microarrays

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 3 (Principles underlying core DNA technologies)
- e-Learning:
  - The origins, determinants, and consequences of human mutations, Shendure and Akey, 2015, Science, 349:1478-1483
  - Optional – A clinician's perspective on clinical exome sequencing, O'Donnell-Luria and Miller, 2016, Hum Genet, 135:643-654.

Guest speaker – Dr. Henry Baker, Professor and Chair, Dept of Molecular Genetics and Microbiology – Use of microarray data to diagnose brain tumors

September 13 – Week 4 – Genetic variation

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 4 (Principles of genetic variation)
- e-Learning:
  - "Human genetics: Genomes on prescription", Nature, 2011, 478:22-24
  - News Feature: Genetic mutations you want, 2016, PNAS, http://www.pnas.org/content/113/10/2554.full
  - Parental somatic mosaicism is underrecognized and influences recurrence risk of genomic disorders, Campbell et al, 2014, Am J Hum Genet, 95(2):173-182
  - Mitochondrial dysfunction in schizophrenia: an evolutionary perspective, Goncalves et al, 2015, Hum Genet, 134:13-21, http://link.springer.com/article/10.1007/s00439-014-1491-8/fulltext.html?wt_mc=alerts:TOCjournals (Interesting article on schizophrenia, evolution and mitochondrial/energy demands of the human brain)

Team-based discussion #1

September 20 – 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:
  - The genetic basis of Mendelian phenotypes: Discoveries, challenges, and opportunities, Chong et al., 2015, AJHG, 97:199-215, http://www.cell.com/ajhg/pdf/S0002-9297%2815%2900245-1.pdf
  - Optional – Mitochondrial DNA copy number in peripheral blood cells declines with age and is associated with general health among the elderly, Mengel-From et al, 2014, Hum Genet, 133(9):1149-1159
  - Alpha1-antitrypsin deficiency, Greene et al. 2016, Nature Reviews, 2:1-17 (Guest speaker article)
  - Optional - A systematic characterization of genes underlying both complex and Mendelian diseases, Jin et al, 2012, Hum Mol Genet, 21(7):`611-1624

Guest lecturer - Dr. Mark Brantly, Professor, Dept of Medicine and Molecular Genetics and Microbiology – alpha 1-antitrypsin deficiency – Confirmed

September 27 – 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 at the epicenter of modern medicine, Feinberg, JAMA, 2008, 299:1345-1350.
  - "A patchwork mind: How your parents' genes shape your brain", Scientific American, 2009, http://www.scientificamerican.com/article.cfm?id=a-patchwork-mind (Guest speaker article)

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

October 4 – Week 7 – Complexities with monogenic disorders

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 7 (Genetic variation producing disease-causing abnormalities in DNA and chromosomes)
- e-Learning:
  - The unstable repeats – Three evolving faces of neurological disease, Nelson et al., 2013, Neuron, 77:825-843

Team-based discussion #2

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

October 11 – Week 8 – Complex diseases, part I

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 8, pp 247-281 (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

Exercise 1 due

October 18 – Week 9 – Complex diseases, part II

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 8, pp 281-308 (Identifying disease genes and genetic susceptibility to complex diseases)

- e-Learning:
  - New concerns raised over the value of genome-wide disease studies, Nature, 2017
  - "Deep genealogy and dilution of risk", Roberts RG, 2013, PLoS Biology, 11(9): e1001660
  - The link between mental illness and physical illness, The Daily Dose, Apr 11, 2017, http://www.ozy.com/acumen/the-link-between-mental-illness-and-physical-illness/76671?utm_source=dd&utm_medium=email&utm_campaign=04112017&variable=7484b1138a1020da7084a14dc20bdfa5
  - Optional - In search of low-frequency and rare variants affecting complex traits, Panoutsopoulou et al., 2013, Hum Mol Genet, 22: R16-R21

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

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

Oct 25 – Week 10 - FL 2017 Genetics Symposium (students must attend 12:45-2:45 and should stay for the 2:45-3:15 talk if possible. Other events are optional). By end of the day, all students must submit (via email) a one paragraph summary of one talk or poster (it counts as a quiz).

Session 1 – Precision Genetics (Chair: Lari Cavallari, Keynote chair: Maurice Swanson)

- Arrive at 12:45 to get seated before the speakers begin
- 1-1:15: Opening remarks, UFGI Director Pat Concannon
- 1:15-2pm: A multi-ethnic, multi-omics approach to cardiovascular disease, Stephen Rich, University of Virginia
- 2-2:45pm: The miracle of genome editing in plants has arrive. Now what", Zach Lippman, Cold Spring Harbor Laboratory
- 2:45-3:15pm: Advancing patient care through genetically-guided drug therapy: The UF Health Personalized Medicine Program, Julie Johnson, University of Florida
- 3:15-5:15pm: Poster session and reception
- 5:30-6:30pm: Keynote speaker – Nusinersen (SpinrazaTM): The first FDA-approved treatment for SMA, Adrian Krainer, Cold Spring Harbor Laboratory

*** Students must register for the symposium

November 1 – Week 11 – Genetic treatments, Part I

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 9, pp 309-317 AND 336-371 (Genetic approaches to treating disease)
- e-Learning:
  - Retinal gene therapy using adeno-associated viral vectors: Multiple applications for a small virus, Hauswirth, Hum Gene Therap, 2014, 25:671-678.
  - Gene therapy for Leber congenital amaurosis caused by *RPE65* mutations: Safety and efficacy in 15 children and adults followed up to 3 years, Jacobson et al, 2012, Arch Opthalmology, 130:9-24 (can skim pp 13, starting with Immune response assays – 21, but be sure to read Comment section on pp 21-23)

Exercise 2 due

Guest speaker – Prof. William Hauswirth, Professor, Dept of Ophthalmology and Molecular Genetics – Gene therapy and eye disease - Confirmed

November 8 – 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
  - 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-human-embryos.html?emc=edit_th_20170803&nl=todaysheadlines&nlid=55234026

Exercise 3 assigned – due November 21 (1 day before Thanksgiving break)

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

November 15 – Week 13 - Cancer

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 10 (Cancer genetics and genomics)
- e-Learning:
  - Somatic mutation in cancer and normal cells, Martincorena and Campbell, 2015, Science, 349:1483-1489
  - 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, http://www.motherjones.com/politics/2015/10/faulty-research-behind-mammograms-breast-cancer/
  - Optional - Deciphering intratumor heterogeneity using cancer genome analysis, Ryu et al. 2016, Hum Genet, 135:635-642
  - Optional - 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.

November 22 - Week 14 - Thanksgiving break

***Exercise 3 due on November 21

November 29 – Week 15 – Genetic testing

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 11, pp 431-474 (Genetic testing from genes to genomes, and the ethics of genetic testing and therapy)
- e-Learning:

- FDA authorizes direct-to-consumer access for multiple 23andMe health risk tests - https://www.genomeweb.com/regulatory-news/fda-authorizes-direct-consumer-access-multiple-23andme-health-risk-tests?utm_source=Sailthru&utm_medium=email&utm_campaign=GWDN%20Thurs%20PM%202017-04-06&utm_term=GW%20Daily%20News%20Bulletin
- "A geneticist's research turns personal", NYT, June 2, 2012
- "A spot of trouble", Carmichael, Nature, 2011
- "Unknown significance", genetic testing from a science writer's perspective - http://www.sciencemag.org/content/346/6214/1167.full
- Preimplantation genetic diagnosis: State of the ART 2011, Harper & SenGupta, 2012, Hum Genet, 131:175-186
- Who has your DNA, or wants it, Science, 2015, 349:1475 - http://www.sciencemag.org/content/349/6255/1475.full?utm_campaign=email-sci-toc
- Optional - Prospective comparison of the cost-effectiveness of clinical whole-exome sequencing with that of usual care overwhelmingly supports early use and reimbursement, Stark et al. 2017, Genetics in Medicine, 19:867-874, http://www.nature.com/gim/journal/vaop/ncurrent/full/gim2016221a.html

Team discussion #3

December 6 - Week 16 – Ethics

Reading assignments:

- *Genetics and Genomics in Medicine* (GGM), Chpt 11, pp 474-488 (Genetic testing from genes to genomes, and the ethics of genetic testing and therapy)
- e-Learning
  - "Researchers to return blood samples to the Yamomamo", Science, June 4, 2010
  - "Taking the least of you", The New York Times, April, 2006
  -

Review for exam

Guest speaker – Bill Allen, Director, Program in Bioethics, Law and Medical Professionalism – Ethical, legal and social issues – Confirmed

Final exam – Thursday, December 14, 7:30-9:30am, regular class room