ANG 6592 (Section 2B93)/ANT 4530 (Section 2B90) - SEMINAR IN MOLECULAR ANTHROPOLOGY

Prof. Connie J. Mulligan

Time: Tuesday, periods 4-6 (10:40 am – 1:40 pm), with a 15 min break

Place: 2334 Turlington

Web: Course is coordinated through e-Learning in Canvas (https://lss.at.ufl.edu)

Instructor: Dr. Connie J. Mulligan
Office hours: Tuesday, 8:30-10:30 in B119 Turlington, or by appointment in 409 Genetics Institute
Contact information: 409 Genetics Institute, 2033 Mowry Rd, (352) 273-8092, cmulligan@ufl.edu

Course summary: The purpose of this class is to examine current applications of molecular data to questions of human evolution and population genetics. Emergence of modern *Homo sapiens*, Neanderthal genetics, reconstruction of human migrations, and evidence of natural selection on the human genome are among the topics discussed in detail. Examples of ancient DNA studies, admixture, and molecular genetics of human disease are also briefly discussed. Data types include mitochondrial DNA, Y chromosome, nuclear DNA, and various –omic datasets, including genomic, exomic, transcriptomic, etc.

Course design: This course is intended for advanced undergraduate and graduate students who have an interest in human evolution as well as in molecular genetics. It is intended for students from all colleges and departments – in the past, students from anthropology, chemistry, history, molecular genetics and microbiology, and zoology have taken the class. 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 in this regard.

Course objectives and student goals: All students are expected to gain knowledge on the molecular genetic basis for various theories of human evolution and subsequent population movements. 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 evolution – in fact, this course may be the most challenging course you have ever taken with respect to developing expertise and familiarity with a wide range of scientific fields, materials, techniques, publications, etc. You are expected to do all of the required readings and to follow up with additional readings if you do not understand something. Additional readings are listed in the textbook and can be found in the references in the journal articles; furthermore, you should become familiar with searching PubMed or Google Scholar for supplementary, follow-up, or original readings.

Reading and course format: Reading material consists of one textbook (Human Evolutionary Genetics, 2nd edition, by Jobling, Hollox, Hurles, Kivisild and Tyler-Smith and a series of approximately 30 related journal articles plus 'news and views' type articles (available on the Canvas course webpage)). If students know of additional articles or topics that they would like to discuss, please contact me. The course meets once per week for three hours and the course format is mainly discussion with substantial student participation. Lectures will be supplementary only in order to provide necessary introductory and background material.

Grading: Final grades will be determined by the following five categories: 1) **participation** (100 pts), 2) weekly **summary/questions/comments** (100 pts), 3) 2-4 homework **exercises** (100 pts), 4) one **oral presentation** (100 pts), and 5) 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. Participation means speaking up in class and contributing ideas, questions, comments, etc. No credit will be given for simply showing up to class, although attendance is mandatory -.remaining silent in class means that the highest grade a student can receive is a B. Furthermore, punctuality is important and participation points will not be awarded to students who are late to class.
- At the beginning of each class, a 1-2 paragraph **summary** and 3 **questions** or **comments** for each journal article assigned for that day's class will be turned in. The summary/questions/comments are required only for the peer-reviewed journal articles, not for the news-and-views type articles that are included in the course pack to provide background or summary information. One question/comment should also incorporate information from the readings in the textbook. The summary/questions/comments are intended to ensure that each student has read the required materials for that day and is prepared to actively participate in class discussions.
- There will be a maximum of four homework **exercises** involving the analysis of hypothetical or actual molecular data. The exercises are intended to give students an opportunity to perform data 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 the class in which an exercise is assigned are expected to turn the exercise in on time, i.e. two weeks after the missed class.
- Each student will present one 15-20 min **oral presentation** based on journal articles from the course packet. The presentation must use MS Power Point. Regardless of whether a student is presenting an article, all students will read all articles in the course packet and be prepared to participate in class discussions of the articles. Students will sign up for articles for oral presentation on the second day of class (only one student may sign up for each article).
- There will be one **exam** that will consist of short answer and essay questions and questions similar to the homework exercises. Questions must be answered during the class period without reference to books or notes of any kind. Calculators may be used, but no cell phones, iPod, smart phones or similar devices will be allowed. Make-up exams will be scheduled only in extenuating circumstances and will require a doctor's note, police report, or similar supporting documentation.
- For advanced graduate students taking the class, there is an option to lecture on their dissertation research as it relates to topics in the class in exchange for part of the above assignments. Interested students should contact the instructor as soon as possible in order to make arrangements.

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

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, etc. Therefore, students are strongly encouraged to attend all classes and to arrive on time. Computers are allowed in class for taking notes, but are not allowed during exams. If computer use gets excessive, i.e. if no one is participating and I'm lecturing to a sea of laptop backs, then I will institute a 'no computers' policy. 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.

Strategies for success:

- It is important to complete all the readings on time and it is best to do 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. Start with the textbook and move onto the journal articles. For the textbook, do not skim the reading, but read every word. For the journal articles, you can focus on the Abstract, Introduction and Discussion. Continue reading though the week so that you have time to contemplate the readings and think of questions and comments to contribute during class. The amount of reading material is modest, particularly for an anthropology course. This is because it is expected that you will re-read (gasp!) 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 summary + questions you must turn in for every article.
- You have two weeks to work on the exercises. These exercises are most likely completely different from anything you've ever done and, thus, you may have lots of questions about how to proceed. 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.
- For the oral presentation, it is a good idea to practice your entire presentation without any stops the night before your scheduled presentation – this ensures your talk is the correct length of time and develops good practice for all public speaking.
- 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.

Strategies for learning: "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. Being prepared for class enables you to construct a knowledge base on which subsequent learning rests." Romack 2010, Enhancing Students' Readiness to Learn, Faculty Focus Special Report: 11 Strategies for Getting Students to Read What's Assigned.

Accommodations for students with disabilities: If you require accommodation due to a disability, please make an appointment 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:

January 10 Introduction
Lecture – background material
Video – The Gene Hunters (example of a study conducted under the Human Genome Diversity Program) or First Peoples (PBS special on human evolution)

January 17 **The human genome**
Reading material
- *Human Evolutionary Genetics* (HEG), Chpts 1 (An introduction to human evolutionary genetics), 2 (Organization and inheritance of the human genome) AND pp 98-100 (PCR amplification)
- Canvas website
  - "Science after the sequence", Nature, June 2010
  - Challenges in human genetic diversity: Demographic history and adaptation, Balaresque et al, 2007, Human Molecular Genetics, 16:R134-R139.
  - Multiple evidence strands suggest that there may be as few as 19 000 human protein-coding genes, Ezkurdia et al. 2014, Hum. Mol. Genet. 23: 5866-5878.

Lecture
- Background material
- Discussion of oral presentations

Sign up for oral presentation

January 24 **Human genetic diversity**
Reading material
- HEG – Chpts 3 (Human genome variation – read lightly), 4 (Finding and assaying genetic diversity – read lightly)
- Canvas website
  - A global reference for human genetic variation, The 1000 Genomes Project Consortium, 2015, Nature, 526:68-74 (don't read the Extended Data section at the end) AND N&V "The end of the start for population sequencing"
  - Novelty-seeking DRD4 polymorphisms are associated with human migration distance out-of-Africa after controlling for neutral population gene structure, Matthews and Butler, Am J Phys Anthropol, 2011, 145:382-389 (oral presentation)

Assign Exercise #1

January 31 **Processes that shape genetic diversity AND Making inferences from diversity**
Reading material
- HEG – Chpts 5 (Processes shaping diversity) and 6 (Making inferences from diversity)
- Canvas website
  - Worldwide genetic and cultural change in human evolution, Creanza and Feldman, 2016, Curr Opin Genet Dev, 41:85-92.
  - Sequencing of 50 human exomes reveals adaptation to high altitude, Yi et al., 2010, Science, 329:75-78 (oral presentation)

February 7 **Humans as apes**
Reading material
- HEG – Chpt 7 (Humans as apes)
- Canvas website
  - "Relative differences: The myth of 1%", Science, June 29, 2007

- Implications of natural selection in shaping 99.4% nonsynonymous DNA identity between humans and chimpanzees: Enlarging genus *Homo*, Wildman et al., 2003, Proc Natl Acad Sci USA, 100:7181-7188
- Parallel patterns of evolution in the genomes and transcriptomes of humans and chimpanzees, Khaitovich et al., Science, 2005, 309:1850-1854 (oral presentation)

Exercise #1 due

February 14 **What genetic changes have made us human?**

Reading material

- HEG – Chpt 8 (What genetic changes have made us human?)
- Canvas website
  - "Macaques vocally equipped to speak", 2016, Nature, 540: 319 (only read the short column on this article that is published elsewhere)
  - "Newly discovered gene could help to explain human evolution, say scientists" - http://www.independent.co.uk/news/science/newly-discovered-gene-could-help-to-explain-human-evolution-say-scientists-8316263.html (no pdf in Canvas – use the link)
  - Relaxed genetic control of cortical organization in human brains compared with chimpanzees, Gomez-Robles, et al. 2015, PNAS, 112:14799-14804 (oral presentation)

Assign Exercise #2

February 21 **Origins of modern humans**

Reading material

- HEG – Chpt 9, pp 283-307 (Origins of modern humans)
- Canvas website
  - Revising the human mutation rate: Implications for understanding human evolution, Scally and Durbin, 2012, Nature Reviews Genetics, 13:745-753.
  - Phonemic diversity supports a serial founder effect model of language expansion from Africa, Atkinson, Science, 2011, 332:346-349
  - Distance from sub-Saharan Africa predicts mutational load in diverse human genomes - http://www.pnas.org/content/113/4/E440 (oral presentation)
  - Optional reading (the most current human genome sequences) - The Simons Genome Diversity Project: 300 genomes from 142 diverse populations, Mallick et al. 2016, Nature, 538:201-206.

February 28 **Origins of modern humans**

Reading material

- HEG – Chpt 9, pp 307-317 (Origins of modern humans - Neanderthals, Denisovans, archaics)
- Canvas website
  - Genetic time travel, Krause and Pääbo, 2016, Genetics, 203:9-12.
  - "Revolution in human evolution", Ann Gibbons, 2015, Science, 349:362-366
  - Archaic admixture in human history, Wall and Brandt, 2016, Curr Opin Genet Dev, 41:93-97
  - Excavating Neandertal and Denisovan DNA from the genomes of Melanesian individuals, Vernot et al., 2016, Science, 352:235-239.
  - "Neanderthals had outsize effect on human biology", 2015, Nature, 523:512-513.
  - Altitude adaptation in Tibetans caused by introgression of Denisovan-like DNA, Huerta-Sánchez et al., 2014, Nature, 512:194-197 (don't read the Extended Data sections) (oral presentation)

- Neanderthal-human hybrid unearthed, 2015, The Scientist - http://www.the-scientist.com//?articles.view/articleNo/43354/title/Neanderthal-Human-Hybrid-Unearthed/ (no pdf on Canvas – use the link)
- What do archaeologists think? "Bone technique redrafts prehistory", 2014, Nature, 512:242

Exercise #2 due

March 7 **Spring break**

March 14 **The distribution of diversity AND The colonization of the Old World and Australia**

Reading material

- HEG – Chpts 10 (The distribution of diversity) and 11 (The colonization of the Old World and Australia)
- Canvas website
  - A comparison of worldwide phonemic and genetic variation in human populations, Creanza et al., 2015, PNAS, 112: 1265-1272 (oral presentation)
  - Evidence for gradients of human genetic diversity within and among continents, Serre and Paabo, Genome Res, 2004, 14:1679-1685

March 21 **Agricultural expansions**

Reading material

- HEG – Chpt 12 (Agricultural expansions)
- Canvas website
  - The genetic history of Ice Age Europe, Fu et al. 2016, Nature, 534:200-205.
  - Genome-wide patterns of selection in 230 ancient Eurasians, Mathieson et al., 2015, Nature, 528:499-503 (oral presentation)

Assign Exercise #3

March 28 **Peopling of the Americas and the Pacific**

Reading material

- HEG – Chpt 13 (Into new-found lands)
- Canvas website
  - A three-stage colonization model for the peopling of the Americas, Kitchen et al., PLoS ONE, 2008, 3:e1596
  - A time transect of exomes from a Native American population before and after European contact, Lindo et al., 2016, Nature Comm, 7:13175 (oral presentation)

April 4 **Admixture and Individual Identification**

Reading material

- HEG – Chpts 14 (What happens when populations meet?) and 18 (Identity and identification)
- Canvas website
  - The genetic ancestry of African Americans, Latinos, and European Americans across the United States, Bryc et al., AJHG, 2015, 96:37-53 (grad student oral presentation)
  - "In our Blood", Newsweek, Feb 6, 2006 - http://www.newsweek.com/our-blood-113321 (no pdf on Canvas, use the link)
  - "Who has your DNA, or wants it", Kaiser, Science, 2015, 349:147
  - "Familial DNA testing scores a win in serial killer case", Science, July 16, 2010

- "High-tech, high-risk forensics", NYT, July 24, 2013 - http://www.nytimes.com/2013/07/25/opinion/high-tech-high-risk-forensics.html?nl=todaysheadlines&emc=edit_th_20130725 (no pdf on Canvas – use the link)

Video – African American Lives
Exercise #3 due

April 11 **Phenotypic variation/adaptation**

Reading material

- HEG – Chpt 15 (Understanding the past and future of phenotypic variation)
- Canvas website
  - "Are humans still evolving?", Science, July 8, 2005
  - Origin of clothing lice indicates early clothing use by anatomically modern humans in Africa, Toups et al., Mol Biol Evol, 2011, 28:29-32.
  - Evidence for evolution in response to natural selection in a contemporary human population, Milot et al., 2011, PNAS, 108:17040-17045 (oral presentation)

April 18 **Genetics of disease**

Reading material

- HEG – Chpts 16 (Evolutionary insights into simple genetic diseases) and 17 (Evolution and complex diseases)
- Canvas website
  - "Parsing the genetics of behavior", Science, Nov 7, 2008
  - "Deflating the genomic bubble", Science, Feb 18, 2011
  - Interaction of *Alu* polymorphisms and novel measures of discrimination in association with blood pressure in African Americans living in Tallahassee, FL, Boulter et al., 2016, Hum Biol, 87:295-305.
  - Mitochondrial dysfunction in schizophrenia: An evolutionary perspective, Goncalves et al., 2015, Hum Genet, 134:13-21 (oral presentation)

Review for exam

Final exam – Monday, April 24, 12:30-2:30 (Group 24C)