ANT 4530 (Section 4B31)/ ANG 6592 (Section 4B30) - GENETIC PERSPECTIVES ON HUMAN DIVERSITY

Instructor: Prof. Connie J. Mulligan

Time: Tuesday, periods 7-9 (1:55-4:55 pm), with a 15 min break

Place: 2333 Turlington

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

Instructor: Dr. Connie J. Mulligan
Office hours: Tuesday, 12:30-1:30 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 description: The purpose of this class is to examine current applications of molecular genetic data to investigate human diversity and human evolution. Our understanding of the human genome, emergence of modern *Homo sapiens*, Neanderthal genetics, reconstruction of human migrations around the world, and evidence of natural selection on the human genome are among the topics we will discuss. Examples of ancient DNA studies, admixture, and molecular genetics of human disease are also briefly discussed.

Prerequisites: A science background is not required to take this class, but students must have an interest in understanding the scientific basis of human evolution. The necessary science will be taught in the course and the course will be scientifically rigorous as befitting a 4000 level course. Students must also have an interest in thinking about how they construct arguments and discussing different ways to present information in a clear and compelling manner.

Course objectives and student goals: 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, biology, chemistry, history, and molecular genetics and microbiology 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 (there are no virtual or Zoom options for this class).

All students are expected to gain knowledge on the molecular genetic basis of human diversity and human evolution. 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 diversity and 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 (http://www.ncbi.nlm.nih.gov/pubmed) or Google Scholar (https://scholar.google.com/) for supplementary, follow-up, or original readings.

Critical thinking skills will be required throughout the course, both in evaluating the strengths/weaknesses of different arguments and also in thinking how to present such information. In class presentations, you will focus on presenting arguments and evaluating the evidence and rationale behind such arguments. It is important to point out that critical thinking is not just a way of thinking, but you first need material to inform your thinking and on which to think critically.

A student who completes this course will be able to:

- Identify key scientific concepts that underly the human genome and human genetic diversity

- Articulate and compare different types of molecular genetic data and how they help us understand human evolution and reconstruct population histories
- Apply critical thinking skills to develop and present arguments on human diversity and human evolution
- Create visual presentations to present peer-reviewed journal articles on human diversity and human evolution

Reading and course format: Reading material consists of one recommended 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 (pdfs are available on the e-Learning course webpage). The textbook is older than I'd like (but there are no better ones and I looked!) but it is a good option if you don't have a good background in genetics and molecular evolution and are struggling with reading the journal articles. If students know of additional articles or topics they would like to discuss, please contact me. The course meets once per week on Tuesdays for three hours and the course format is mainly discussion with substantial student participation. Lectures will be supplementary to provide necessary introductory and background material.

Grading: UF grading policies are at https://catalog.ufl.edu/ugrad/current/regulations/info/grades.aspx

Final grades will be determined by the following five categories (see detailed descriptions below):

1) **Participation** (50 pts)
2) **5 quizzes** (10 pts each, 50 pts total)
3) **1-2 homework exercises** (50 pts)
4) **4 team discussions** (25 pts each, 100 pts total)
5) **1 oral presentation** (100 pts each)
6) **1 exam** (100 pts)

Possible points total 450

- **Participation** is important to facilitate class discussions of the topics covered in class – in the past, many students have said they learned the most from these class discussions. Participation comments provide a demonstration that you have read and thought about the assigned material, and also to provide different perspectives on the topics being discussed. Participation is required of all students and will be based on each student's contribution of comments, questions, etc to the class. Students are not graded on the brilliance of their statements, but on their willingness to talk in class. Participation will be graded on comments made in class, not comments made to me personally after class, because the purpose of participation is to prompt class discussions. Punctuality is important and participation points will not be awarded to students who are habitually late to class. I will ensure that no student dominates the discussions and that there are opportunities for all students to participate. Approximately 85% of the participation grade will be based on comments made or questions asked in class and approximately 15% of the participation grade will be based on attendance.
- There will be six unscheduled **quizzes**, to be taken in class. The quizzes consist of multiple-choice 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, i.e. 5 quizzes will count towards your final grade. Missed quizzes cannot be made up. Quizzes must be taken in class because they are closed book and there are no safeguards to ensure that other material are not consulted outside of class, e.g. Honorlock.
- There will be 1-2 **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.
- There will be four **team 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 in class based on the week's reading and will present a short (~2-5 min) summary of their discussion to the class. Grades will

be based on the presentation as well as individual participation as assessed by team members. Missed team-based discussions cannot be made up.

- Each student will present one 15-20 min **oral presentation** based on journal articles from the course packet. 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. Questions must be answered during the class period without reference to books or notes of any kind. Make-up exams will be scheduled only in extenuating circumstances and will require a doctor's note, police report, or similar supporting documentation.
- Final grades will be based on the following point percentages:
  - 100 - 93% = A
  - 92.99 - 90% = A-
  - 89.99 - 87% = B+
  - 86.99 - 83% = B
  - 82.99 - 80% = B-
  - 79.99 - 77% = C+
  - 76.99 - 73% = C
  - 72.99 - 70% = C-
  - 69.99 - 67% = D+
  - 66.99 - 63% = D
  - 62.99 - 60% = D-
  - Less than 60% = E

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
http://www.uflib.ufl.edu - UF database of available electronic journals (may only be available from a campus computer or through a vpn)
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.genomesonline.org/ - status of genome sequencing projects

Attendance and punctuality: Requirements for class attendance and make-up exams, assignments, and other work in this course are consistent with university policies that can be found at: https://catalog.ufl.edu/ugrad/current/regulations/info/attendance.aspx. Because the class meets only one time per week and because the class format is mainly discussion and class participation, it is very difficult to make up missed classes by borrowing notes, watching lectures, etc. Therefore, students are strongly encouraged to attend all classes and to arrive on time. Punctuality is important because I summarize important logistical items at the beginning of class and because punctuality demonstrates professionalism (if you cannot make it to class on time because of class or work conflicts, please let me know). 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 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 student to speak to a sea of laptop backs. Cell phones can be used to research topics we are discussing in class, but should be used sparingly and not be used for texting, checking the time, etc.

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.

Copyright information: Publication of any course materials 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 Honor Code and Student Conduct Code.

Accommodations: UF is committed to achieving full accessibility for people with disabilities, and I am committed to making this classroom accessible to you. If there are ways in which the overall structure of the course and general classroom interactions could be adapted to facilitate improved participation, please do not hesitate to raise your ideas with me: Your comments and suggestions about the format of readings, lectures, and class discussions are always welcome.

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, 392-1261, 202 Peabody Hall. The Dean of Students Office will provide documentation to the student who must then provide this documentation to the instructor when requesting accommodation.

Evaluations: Students are expected to provide feedback on the quality of instruction in this course by completing online evaluations at https://evaluations.ufl.edu. Evaluations are typically open during the last two or three weeks of the semester, but students will be given specific times when they are open. Summary results of these assessments are available to students at https://evaluations.ufl.edu/results/.

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." On all work submitted for credit by students at the University of Florida, the following pledge is either required or implied: "On my honor, I have neither given nor received unauthorized aid in doing this assignment." The Honor Code (https://www.dso.ufl.edu/sccr/process/student-conduct-honor-code/) specifies a number of behaviors that are in violation of this code and the possible sanctions – prohibited behavior includes the use of AI applications to write assignments. Furthermore, you are obligated to report any condition that facilitates academic misconduct to appropriate personnel. If you have any questions or concerns, please consult with the instructor in this class.

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

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.

Use of AI, ChatGPT and other LLMs. Generative AI can be useful and will surely develop more utility in the coming years. You may be tempted to use it in this course as a short-cut on assignments or to understand the readings, but I ask that you do not. The readings, assignments, and discussions in this course are intended to develop your own analytical and critical thinking skills, and use of ChatGPT and other LLMs can short-circuit that process.

Critical thinking and freedom of speech: Students are encouraged to employ critical thinking and to rely on data and verifiable sources to interrogate all assigned readings and subject matter in this course as a way of determining whether they agree with their classmates and/or their instructor. No lesson is intended to espouse, promote, advance, inculcate, or compel a particular feeling, perception, viewpoint, or belief.

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:

August 29/Week 1 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)

September 5/Week 2 The human genome (Team discussion)

Reading material

- eLearning website
  - Worldwide genetic and cultural change in human evolution, Creanza & Feldman, 2016, *Current Opinion in Genetics & Development*.
  - How a scholarly spat shaped genetic research (a book review of Disputed Inheritance: The Battle over Mendel and the Future of Biology), Hall, 2023, Nature
  - Genetic determinism, essentialism and reductionism: Semantic clarity for contested science, Harden, 2023, *Nature Genetics Reviews.*
  - Genetic and environmental contributions to individual differences in sustainable working life—A Swedish twin cohort study, Ropponen et al., 2023, *PLoS ONE.*
- *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)

Team discussion #1

Sign up for oral presentations

September 12/Week 3 Human genetic diversity

Reading material

- eLearning website (*** not available on eLearning, click link to access article)
  - A draft human pangenome reference, Liao et al., 2023, *Nature* (you don't have to read past page 12)
    - N&V - A collective human reference genome, Massarat & Gymrek, 2023, *Nature.*
    - N&V – First human 'pangenome' aims to catalog diversity, Liverpool, 2023, *Nature.*
  - African American genomes don't capture Africa's genetic diversity, Fatumo & Choudhury, 2023, *Nature.*
  - Genetic ancestry in precision medicine is reshaping race debate, Krainc & Fuentes, 2022, *PNAS*.
  - The functional and evolutionary impacts of human-specific deletions in conserved elements, Xue et al., 2023, *Science.*
    - N&V Seeing humans through an evolutionary lens, Romero, 2023, *Science*.
    - ***'Deletions' from the human genome may be what made us human, Hathaway, 2023, YaleNews.
  - Optional – So many Nigerians: why is Nigeria overrepresented as the ancestral genetic homeland of Legacy African North Americans, Jackson, 2021, *AJHG.*
- HEG – Chpts 3 (Human genome variation – read lightly), 4 (Finding and assaying genetic diversity – read lightly)

Team discussion #2

September 19/Week 4 Processes that shape genetic diversity AND Making inferences from diversity

Reading material

- eLearning website
  - The genomics of human local adaptation, Rees et al., 2020, *Trends in Genetics.*
  - A unified genealogy of modern and ancient genomes, Wohns et al., 2022, *Science*.
    - Inferring human evolutionary history, Rees & Andrés, 2022, *Science*.
  - The genetic legacy of African Americans from Catoctin Furnace, Harney et al., 2023, Science
    - Forging connections: Cemetery DNA links enslaved people, Curry, 2022, *Science*.
- HEG – Chpts 5 (Processes shaping diversity) and 6 (Making inferences from diversity)

Assign Exercise #1

September 26/Week 5 Humans as apes AND What genetic changes have made us human?

Reading material

- eLearning website
  - "Relative differences: The myth of 1%", Cohen, 2007, *Science*.
  - Mutations help genes emerge from aimless DNA, Pennisi, 2023, *Science*.
  - New copies of old gene drove brain expansion, Pennisi, 2018, *Science*.
  - Newly evolved genes in the human lineage are functional, Papadopoulos & Albà, 2022, *Trends in Genetics*.

- "Macaques vocally equipped to speak", 2016, *Nature* (just read the short summary, under Primatology, of this article that is published elsewhere)
- Relaxed genetic control of cortical organization in human brains compared with chimpanzees, Gomez-Robles, et al. 2015, *PNAS*. (Oral presentation)

- HEG – Chpt 7 (Humans as apes) and 8 (What genetic changes have made us human?)

Oral presentation

October 3/Week 6 Origins of modern humans

Reading material

- eLearning website
  - Tracing the peopling of the world through genomics, Nielsen, et al., 2017, *Nature*.
  - Origins of modern human ancestry, Berstrőm et al., 2020, *Nature*.
  - Khoe-San genomes reveal unique variation and confirm the deepest population divergence in Homo sapiens, Schlebusch et al., 2020, *Mol Biol Evol*. (Oral presentation)
- HEG – Chpt 9, pp 283-307 (Origins of modern humans)

Exercise #1 due
Oral presentation

October 10/Week 7 Origins of modern humans – ancient DNA, introgression with Neanderthals, Denisovans, and other archaic hominins, and archaic genetic variation

Reading material

- eLearning website
  - Archaic hominin introgression into modern human genomes, Gokcumen, 2020, *Yrbk Phys Anthropol*.
  - 'Truly gobsmacked' Ancient human genome count passes 10,000, Callaway, 2023, *Nature*.
  - Genetic insights into the social organization of Neanderthals, Skov et al., 2023, *Nature*. (Oral presentation)
    - The first genomic portrait of a Neanderthal family, Cassidy, 2023, *Nature*.
- HEG – Chpt 9, pp 307-317 (Origins of modern humans - Neanderthals, Denisovans, archaics)

Oral presentation

October 17/Week 8 Origins of modern humans - functionality of archaic introgression

Reading material

- eLearning website (these are challenging articles)
  - How genes from Neanderthals predispose people to severe COVID-19, Dasgupta, 2023, *TheScientist.*
  - Impact and evolutionary determinants of Neanderthal introgression on transcriptional and post-transcriptional regulation, Silvert et al., 2019, *Am J Hum Genet*.
  - Denisovan introgression has shaped the immune system of present-day Papuans, Verpasiani et al., 2022, *PLoS Genetics*. (Oral presentation)

Oral presentation

October 24/Week 9 **The distribution of diversity AND The colonization of the Old World and Australia**

Reading material

- eLearning website (*** not available on eLearning, click link to access article)
  - Community partnerships are fundamental to ethical ancient DNA research, Kowal et al., 2022, *Hum Genet Genome Adv.*
  - Ancient genome studies expose Africa's past, Callaway, 2017, *Nature*.
  - ***Ancient humans traveled half the world to Asia before main migration out of Africa, Price, 2023, Science, https://www.science.org/content/article/ancient-humans-traveled-half-world-asia-main-migration-out-africa
  - ***The first Europeans weren't who you might think, Curry, 2019, National Geographic, https://www.nationalgeographic.com/culture/article/first-europeans-immigrants-genetic-testing-feature
  - Ancient genomes and West Eurasian history, Arbuckle and Schwandt, 2022, *Science*.
  - Evidence for gradients of human genetic diversity within and among continents, Serre and Pääbo, 2004, *Genome Research*.
- HEG – Chpts 10 (The distribution of diversity) and 11 (The colonization of the Old World and Australia)

Team discussion #3

October 31/Week 10 **Agricultural expansions**

Reading material

- eLearning website
  - Ancient Rome: A genetic crossroads of Europe and the Mediterranean, Antonio et al, 2019, *Science*. (Oral presentation)
  - The mystery of early milk consumption in Europe, Wilkin, 2022, *Nature*.
  - A molecular investigation of human self-domestication, Wilkins, 2020, *Trends in Genetics*.
- HEG – Chpts 12 (Agricultural expansions)

Assign Exercise #2
Oral presentation

November 7/Week 11 **2023 Florida Genetics Symposium**

The class will attend the opening session of the 2023 Florida Genetics Symposium, located in the auditorium of the Genetics Institute, 2033 Mowry Rd, 1-3pm, plus poster session at 3:15-5:15pm

The opening session is on Pangenomics and features the following speakers (read their bios and peruse their webpages before the symposium):

- Erik Garrison, Assistant professor, University of Tennessee, Health Science Center, http://hypervolu.me/~erik/erik_garrison.html
- Erin Molloy, Assistant professor, Dept of Computer Science, University of Maryland, https://www.cs.umd.edu/people/ekmolloy
- Meixia Zhao, Assistant professor, Dept of Microbiology and Cell Science, University of Florida, https://microcell.ufl.edu/people/meixia-zhao/

November 14/Week 12 **Admixture and Individual Identification**

Reading material

- eLearning website (*** not available on eLearning, click link to access article)

- Human genetic admixture, Korunes & Goldberg, 2021, *PLoS Genetics*.
- What is ancestry? Mathieson & Scally, 2020, *PL0S Genetics*.
- ***"In our Blood", Newsweek, Feb 6, 2006 - http://www.newsweek.com/our-blood-113321
- Sample Genetic ancestry report, intended for an African American audience
- Familial DNA testing scores a win in serial killer case, Miller, 2010, *Science*.
- ***"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
- ***Environmental human DNA offers new opportunities for public good, UF News, Aug 2023, https://news.ufl.edu/2023/08/environmental-dna-opportunities/
- "Who has your DNA, or wants it", Kaiser, Science, 2015, 349:147
- An innovative transfer DNA experimental design and qPCR assay: Protocol and pilot study, McCrane & Mulligan, 2023, *Journal of Forensic Sciences*.

- HEG – Chpts 14 (What happens when populations meet?) and 18 (Identity and identification)

Video – African American Lives

Exercise #2 due
Team discussion #4

November 21/Week 13 Phenotypic variation/adaptation

Reading material

- eLearning website (*** not available on eLearning, click link to access article)
  - "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., 2011, *Mol Biol Evol*.
  - How the Black Death left its mark on immune system genes, Gibbons, 2022, *Science*.
  - Apolipoprotein E4 is associated with improved cognitive function in Amazonian forager-horiculturalists with a high parasite burden, Trumble et al. 2017, *FASEB J.* (Oral presentation)
    - ***New study shows cognitive decline may be influenced by interaction of genetics and…worms, 2016, The Science Explorer, http://thescienceexplorer.com/brain-and-body/new-study-shows-cognitive-decline-may-be-influenced-interaction-genetics-and-worms)
- HEG – Chpt 15 (Understanding the past and future of phenotypic variation)

Oral presentation

November 28/Week 14 Genetics of disease

Reading material

- eLearning website
  - The ancient history of kissing, Arbøll & Rasmussen, 2023, *Science*.
  - "Parsing the genetics of behavior", Science, Nov 7, 2008
  - Low birthweight is associated with epigenetic age acceleration in the first 3 years of life, Quinn et al., 2023, *Evolution, Medicine, & Public Health*.
  - The gut microbiota links disease to human genome evolution, Quan et al., 2023, *Trends in Genetics*.

- HEG – Chpts 16 (Evolutionary insights into simple genetic diseases) and 17 (Evolution and complex diseases)

Review for exam

December 5/Week 15 Exam