The Genetics of Evolution and Adaptation in Human Populations

Time and location: TBA, 2 times weekly for 1.5 hours

Course Instructor: Jenn Coughlan, (PhD Candidate, Duke University)
Email: jennifer.coughlan@duke.edu * emails will be answered within 24 hours
Office: FFSC 3332
Office Hours: TBA in class (by popular vote), or half-hour meetings by request

Course Overview:

Change through time is inevitable in all populations, including *Homo sapiens*. Understanding the evolutionary mechanisms which underlay these changes are important, because it not only helps us to understand our own evolutionary past, but also to make important and informed decisions about how to apply evolution to our everyday lives. In this class, we will review the basics of evolutionary genetics as they apply to humans, explore how neutral and demographic processes shape population structure in modern day humans, and discover how natural and sexual selection have shaped phenotypic variation in modern human populations. The class will be split into mini-lecture and seminar style discussion sections (once per week each), and students will be in charge of leading discussion. We will also work throughout the semester on developing experiments to test the evolutionary mechanism(s) responsible for potential adaptations in human populations, with the overall goal of writing a miniature grant proposal. This process will include an oral presentation, as well as providing and obtaining peer-review comments on proposal drafts. While we will be reviewing some basic evolutionary genetics, BIOLOGY 202 (Genetics and Evolution) is a pre-requisite for this course. Any student wishing to take this course without the appropriate requirement will need special permission from myself before enrolling.

Learning Objectives:

1) Identify cases of evolution in human populations and describe the evolutionary mechanism by which change has occurred

2) Interpret population and quantitative genetic data to determine what evolutionary forces are at work, and be able to apply the patterns seen in one population to other populations of humans worldwide.

3) Explain how experimental designs test a specific hypothesis and be able to design a hypothetical experiment to test if a human population is evolving and why.

4) Be able to assess and critique the quality of an experimental design and commonly used methodologies in evolutionary genetics.

Readings:
While this class will have no required textbook, we will read and discuss peer-reviewed primary literature every week. Articles will be available on Sakai.

Other Useful Resources:

Books

Authors	Title	Links
Falconer and Mackay	Introduction to Quantitative Genetics	Duke Library Amazon
Hartl and Clark	Principles of Population Genetics	Duke Library Amazon

Websites and Blogs		
Topic	**Authors**	**Links**
Notes on Population Genetics	Graham Coop, UC Davis	https://gcbias.org/population-genetics-notes/
Notes on Quantitative Genetics	Phillip McClean, NDSU	https://www.ndsu.edu/pubweb/~mcclean/plsc431/quantgen/qgen1.htm
Notes on Evolutionary Genetics	Mark Rausher, Duke University	http://sites.biology.duke.edu/rausher/lecout.htm
Helpful genetics definitions	University of Wyoming	http://www.uwyo.edu/dbmcd/popecol/maylects/popgengloss.html
Duke Writing Center		

Assignments and Grading:

Participation (10%)- *based on active class discussion. Points need not be numerous, simply of substance.*

Pre-class memos (5%)- *1 page overview of the assigned readings. The goal of these memos is to synthesize and summarize the readings. Questions such as- what was the big question? How did they authors answer it? What were the main findings? And What is the main take home?- should be addressed in each memo. Memos are due at 5pm before the first class of the week. Top 10 of 12 will count towards grade.*

Post- class reflection papers + class discussion lead (12.5% x 2 each= 25% total)- *For two classes, you will be responsible for leading discussion. Students should have a series of discussion questions ready, plus be able to give a brief synopsis of the readings. After class, students are responsible for a 2 page review and synopsis of the material discussed that week in class based on the paper(s) read in class, plus 2 additional peer-review articles on the subject. Students should touch on the broad*

themes discussed in the readings, as well as provide suggestions for future areas of research in the discussed field.

Grant Proposal (20% or 25%*)- *Students are responsible for a 2 page mock-grant proposal in which they test whether genetic variation in humans is adaptive, and a potential selective agent. This mock experiment can be completed at any level (i.e. population, group of populations, latitudinal gradient, whole species), and could test any type of selection. Final proposal will be due the final week of class.*

Presentation (20% or 25%*)- *Students will give a brief (15 minute+ questions) proposal overview to the class, outlining the big question and the proposed methods. Presentations will occur for 2 weeks during regular class time.*

Peer Review (15%)- *students are responsible for providing constructive critiques to their peers. Each presentation will receive anonymous reviews from 3 students, plus myself. Feedback should include a brief synopsis of the proposed experiment, any potential methodological flaws, any points of confusion, and a 'fundability' rating. They should also include at least 2 things that were appreciated, or that the reviewer felt were very interesting. We will practice giving constructive feedback in class. These reviews will be brief- a one page fillable sheet will be available on Sakai- and will be due 24 hours after the observation takes place so students have enough time to incorporate reviews into their final projects.*

*The cumulative grade of the written grant proposal and grant proposal presentation will be 45%. Students may choose which of these projects is worth 25%, and which is worth 20%.

Class Attendance and Participation Policies:
Students are expected to attend class each week, to come prepared to discuss the assigned readings, and to participate in class discussion. Participation need not be lengthy, but rather substantive. Students may have a maximum of 2 unexcused absences, but any unexcused absences after are each subject to a 1% loss of the final grade (to be taken from the participation grade).

Policy on missed or late assignments:
Late assignments will receive a grade penalty of 10% per day late, unless prior extensions have been granted. Assignments later than 1 week will receive a 0%. Any extension requests

must come at a reasonable time in advance of the due date (i.e. 48 hours or more), unless extenuating circumstances apply.

Policy on Academic integrity, cheating and classroom decorum:
Students are expected to engage respectfully in discussion and adhere to the Duke Community Standard:

1. I will not lie, cheat, or steal in my academic endeavors;
2. I will conduct myself honorably in all my endeavors; and
3. I will act if the Standard is compromised.

Any suspicion of academic dishonesty or cheating will be reported to the Office of Student Conduct

Policy on Americans with Disabilities Act

Any persons with disabilities are encouraged to contact the Duke Disability Access Office as soon as possible to ensure classroom accessibility.

Tentative Schedule (subject to change based on snow days/ cancelled classes)

Date	**Course Topics**	**Readings**	**Presenters/ Assignments Due**
Week 1: Aug. 24	What can evolutionary genetics teach us about humans?	Botstein and Risch. 2003. Discovering genotypes underlying human phenotypes: past successes for Mendelian diseases, future approaches for complex disease.	1 pg. Memo
	Transmission genetics, determining phase, Mendelian genetics – simple overview, classic examples in humans (discussion)		Presenter: JC
Week 2: Aug. 31	Quantitative genetics- basics and practice	Allen *et al.* 2010. Hundreds of variants clustered in genomic loci and biological pathways affect human height	1 pg. Memo
	What do we know about complex traits in humans? *Case study of height* (discussion)		Presenter:
Week 3: Sept. 7	HWE and departures there of (inbreeding + selection)	McQuillan et al. 2012. Evidence for inbreeding depression on human height	1 pg. Memo
	Case study of inbreeding in human populations (discussion)		Presenters:

Week 4: Sept. 14	Genetic Drift and random sampling- crazy things that have happened to humans based only on sampling error	Gattepaille *et al.* 2012. Inferring population size changes with sequence and SNP data: lessons from human bottlenecks	1 pg. Memo
	Island migration and founder effects (discussion)		Presenters:
Week 5: Sept. 21	The Coalescent and inferring population relatedness	Conrad *et al.* 2006. A worldwide survey of haplotype variation and linkage disequilibrium in the human genome. Nature	1 pg. Memo
	Using PCA to determine human ancestry (discussion)		Presenters:
Week 6: Sept. 28	Demography: migration + bottlenecks in humans	Li *et al.* 2008. Worldwide human relationships inferred from genome-wide patterns of variation	1 pg. Memo
	tracking migratory routes out of Africa (discussion)		Presenters:
Week 7: Oct. 5	Balancing school vs Neutral Theory- what is it, where have we come since then, general take homes and assessments	Bamshad and Wooding. 2003. Signatures of natural selection in the human genome.	1 pg. Memo
	Tracking the relative importance of different classes of mutations/selection in human populations (discussion)		Presenters:
Week 8: Oct. 12	Selection Part I- The major role of purifying selection + detecting selection using popgen tools	Henn *et al.* 2016. Distance from sub-Saharan Africa predicts mutational load in diverse human genomes.	1 pg. Memo
	Looking at genetic load in human populations out of Africa (discussion)		Presenters:
Week 9: Oct. 19	Selection Part II- Local vs global adaptations- case studies in humans	Robinson *et al.* 2015. Population genetic differentiation of height and body mass index across Europe	1 pg. Memo
	Case study- selection on human height (discussion)		Presenters:
Week 10: Oct.26	Selection Part III- sexual selection in humans	Courtiol *et al.* 2011. Natural and sexual selection in a monogamous historical human population	1 pg. Memo
	Natural vs Sexual selection in humans (discussion)		Presenters:

Week 11: Nov. 2	Selection Part IV- trade offs + levels of selection	Jones and Tuljapurkar. 2014. Measuring selective constraint on fertility in human life-histories	1 pg. Memo
	Case studies in life history theory of humans - discussion		Presenters:
Week 12: Nov. 9	Sources of Genetic Variation in humans- standing genetic variation, de novo mutation, inter-specific introgression	Huerta-Sanchez *et al.* 2014. Altitude adaptation in Tibet caused by introgression of Denisovian-like DNA.	1 pg. Memo
	Case study- Adaptive introgression from Denisovians into humans, and adaptation to high altitude		Presenters:
Week 13: Nov. 16			Student Presentations
THANKS GIVING	NO CLASS	NO CLASS	
Week 14: Nov. 30			Student Presentations
Finals Week	NO CLASS	NO CLASS	Final Papers