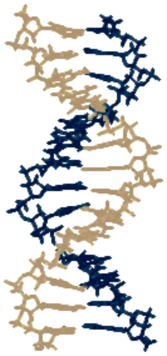
BIOL 4803/8803 Special Topics: **Human Evolutionary Genomics**



Spring Semester 2016, 3 credits
MWF – 10:05am-10:55am
Location: Cherry Emerson 204

Instructor

Dr. **Joseph Lachance**, Assistant Professor in the School of Biology
Email: joseph.lachance@biology.gatech.edu
Office: 2103 Engineered Biosystems Building
Office hours: Tuesdays 1:00pm-2:30pm, or by appointment

Overview

This is an advanced course where students will discuss primary literature and use computational tools to investigate how evolution has shaped global patterns of human genetic variation. It is geared towards graduate students (MS or PhD) and experienced undergraduate students. This class integrates genetics, evolutionary biology, anthropology, computational biology, and bioinformatics. Each week students will be introduced to:

- ***concepts*** of evolutionary genetics
- ***papers*** of cutting edge research in human genomics
- ***exercises*** where computational tools are applied to real-world datasets

Prerequisites

Undergraduates taking BIOL 4803 are required to have previously taken BIOL 1510 or BIOL 1511, and previous coursework in genetics and evolutionary biology is advised (e.g. BIOL 2344 and BIOL 3600). It is assumed that you are comfortable using a command line.

Learning outcomes

As a result of taking this course, you will be able to:

1. Understand how different evolutionary forces shape human genomic variation. This will enable you to propose alternative hypotheses to explain observed patterns of human genetic diversity.
2. Critically read and discuss the scientific literature. You will be able understand how a given paper advances our knowledge of human evolutionary genomics and be able to critically evaluate both the approaches taken and the conclusions of the authors.
3. Use computational tools to analyze genome-scale datasets of human genetic variation. You will become comfortable working with large datasets and develop a familiarity with a wide-array of computational tools. This hands-on experience will serve as a springboard for your own research in the future.

Instructional format

Class time will be equally divided among lectures, discussion of the primary literature, and in class exercises.

Lectures (Mondays): Each Monday a different topic will be introduced to the class. To get the most out of these in-class exercises it is highly advised that students read the assigned chapter in the textbook ahead of time. Concepts introduced during lectures will be relevant to the paper discussed at midweek and the in-class exercise at the end of the week.

Discussion of the primary literature (Wednesdays): Each Wednesday we will discuss a different paper. During the first week of class we will create a schedule of who will lead each paper. Discussion leaders will meet with the course instructor prior to the discussion they will lead. All students are required to read each week's paper and actively contribute to all discussions. Prior to the start of each journal club each student will submit one question about the paper via email (for credit and to stimulate discussion).

In-class exercises (Fridays): Each Friday we will analyze real world datasets of human genetic variation. Datasets will be chosen to illustrate the concepts introduced early in the week. During most weeks this will involve using computational tools from the paper that was discussed midweek. Students will work in small 2-4 person groups, with at least one member of each group bringing a laptop to class. At the end of each in-class exercise each student will turn in a worksheet describing what he or she learned.

Course policies and assignments

Participation: You are expected participate in both the discussion of the primary literature (journal club) and in the weekly in-class exercises. If you are unable to attend class due to illness or another type of excused absence please email the course instructor. Note that in-class use of cell phones or computers for purposes unrelated to course activities is not allowed.

Exams: There will be three take-home exams in this course. Hard copies of your answers will be due at the beginning of deadline. In addition, .pdf versions of your answers will need to be uploaded to t-square. Students are allowed to research their answers over the internet and discuss questions with each other (science is collaborative after all). However, exams must represent the work of individual students. In other words, make sure that your answers are your own. Late take-home exams will be accepted up to 5 days late, with 10% deducted per 24-hour period that it is late.

Evaluation

Journal club participation	25%
(15% for leading discussion and 10% for active participation during other weeks)	
In-class exercises	30%
(2% for each week)	
Exam 1 (take-home)	15%
Exam 2 (take-home)	15%
Exam 3 (take-home)	15%

Textbook and class webpage

Required textbook: *Human Evolutionary Genetics (2nd edition)* by Jobling et al. (Garland, 2013)
The Lachance Lab has a copy of *Human Evolutionary Genetics* in EBB 2101 (note, however that the book needs to stay in the room!).
Additional online resources: http://tsquare.gatech.edu

Learning Accommodations

If needed, we will make classroom accommodations for students with disabilities. These accommodations must be arranged in advance and in accordance with the ADAPTS office (http://www.adapts.gatech.edu).

Academic integrity

Although you are allowed to discuss answers to take-home exams with your classmates, it is important that you submit your own work. There is a zero-tolerance policy re: plagiarism. If you have any questions about what constitutes plagiarism, please email the course instructor. Students are reminded of the obligations and expectations associated with the Georgia Tech Academic Honor Code, available online at: http://catalog.gatech.edu/rules/18b.php. Any violations of the GT Honor Code will result in referral to the Office of Student Integrity with a penalty ranging from no credit for the assignment in question, to a grade of “F” for the class.

Syllabus

Spring 2016	**Topic**	**Text**	**Paper**	**In-class exercise**
Jan 11-15	Human genetic variation	Ch 1	1KG Project Consortium 2015	Introduction to online databases (UCSC browser, GGV)
Jan 18	*No class (MLK day)*			
Jan 20-22	Mutation	Ch 2	Scally and Durbin 2012	Estimating TMRCA
Jan 25-29	Introduction to theoretical population genetics	Ch 3	Myles et al 2008	F_{ST} calculations and tests of Hardy-Weinberg Equilibrium
Feb 1-5	Genotyping technologies and ascertainment bias	Ch 4	Lachance and Tishkoff 2013	Compare SNP array to whole genome sequencing data in R
Feb 8-12	Linkage and recombination	Ch 5	Pratto et al 2014	Calculate LD using 1KG data
Assigned: Feb 12 **Due: Feb 19**	**EXAM 1**			
Feb 15-19	Out-of-Africa migration	Ch 11	Henn et al 2015	Model different types of bottlenecks
Feb 22-26	Human adaptation	Ch 15	Grossman et al 2013	CMS browser
Feb 29-Mar 4	Diseases of modernity	Ch 12	Ayub et al 2014	Compare variants between hunter-gatherers and farmers
Mar 7-11	GWAS and disease	Ch 16	Gibson 2012	PLINK
Assigned: Mar 11 **Due: Mar 18**	**EXAM 2**			
Mar 14-18	Disease risk across populations	Ch 17	Corona et al 2013	Catalog of Published GWAS + GGV
Mar 21-25	*No class (spring break)*			
Mar 28-Apr 1	Geography and genetics	Ch 10	Novembre et al 2008	PCA
Apr 4-8	Demographic inference	Ch 6	Li and Durbin 2011 Gutenkunst et al 2009	PSMC
Apr 11-15	Neanderthal introgression	Ch 9	Green et al. 2010 *(subject to change)*	ABBA/BABA test
Apr 12-16	*American Association of Anthropological Genetics meets in Atlanta* *(Guest lectures and chance to meet with outside researchers who study ancient admixture)*			
Apr 18-22	Adaptive introgression and human uniqueness	Ch 8	Dannemann et al 2016 Bustamante et al 2005	MK tests
Assigned: Apr 22 **Due: May 2**	**EXAM 3**			
Apr 25	Ancestry and admixture *(discussion on Monday)*	Ch 14	Hellenthal et al 2014 Raj et al 2014	STRUCTURE *(optional exercise)*