

## **BCB 722 (Spring 2016): Foundations of population genomics**

### **Basic course information:**

Duration: 03/22/16 – 04/21/16

When: Tuesdays/Thursdays, 2:30 pm – 3:45 pm

Where: Bondurant 2020

Materials: All course materials will be posted on “Sakai” at <https://sakai.unc.edu>

### **Instructor:**

Dr. Praveen Sethupathy  
Department of Genetics  
120 Mason Farm Road  
5091 Genetic Medicine Building  
E-mail: [praveen\\_sethupathy@med.unc.edu](mailto:praveen_sethupathy@med.unc.edu)  
Tel: 919-966-6387

### **Teaching assistant:**

Sherif Farag  
Bioinformatics & Computational Biology Curriculum  
E-mail: [farags@email.unc.edu](mailto:farags@email.unc.edu)

### **Course description:**

This course will cover the fundamental principles of population genomics. We will address such questions as: How frequently do mutations arise? What are the evolutionary forces that have shaped the genetic diversity we see today? Can we distinguish one such force from another? Can we estimate their relative strengths? Is any of this relevant for studying the role of genetics in complex disease? The goal of the course is to equip students with foundational knowledge in the theory of evolution, which serves as the bedrock of modern biology.

### **Course prerequisites:**

Students should have a grasp of undergraduate level genetics/biology. The course is open to all graduate students within the Biological and Biomedical Sciences Program (BBSP) at UNC.

### **Course requirements:**

To obtain full credit, students must attend 80% of the lectures, complete all reading/homework assignments, and achieve a passing grade overall. If a student believes that he/she may have to miss more than 20% of the lectures, he/she must discuss this with the lead instructor in advance.

### **Grading rubric:**

- In-class participation: 20%
- In-class assignments: 25%
- Homework assignments: 45%
- End-of-module survey completion: 10%

## Course syllabus:

All homework assignments will be uploaded onto Sakai immediately following the lecture. Optional reading assignments will also be uploaded onto Sakai along with the required reading. Each time materials are deposited onto the course site the lead instructor will send an e-mail notification.

Please also note that the lead instructor may modify the course syllabus at any time, and will formally inform the students both in lecture and through Sakai if/when this happens.

Every lecture will start with brief discussion of ~2-3 concepts in population genomics, largely based on questions that students submitted on the first day of class. (Questions will be selected for discussion on the basis of their relevance to the lecture topic and the overall course material.)

### Lecture content and reading assignments:

Date	Content	Reading for next lecture
03/22	<ul style="list-style-type: none"><li>➤ Laying the foundation<ul style="list-style-type: none"><li>• Introducing concepts in population genomics and molecular evolution</li></ul></li></ul>	<ul style="list-style-type: none"><li>➤ Crow.pdf: <i>The origins, patterns and implications of human spontaneous mutation</i></li><li>➤ Manolio.pdf: <i>A HapMap harvest of insights into the genetics of common disease</i></li></ul>
03/24	<ul style="list-style-type: none"><li>➤ Evolutionary forces that shape genetic diversity<ul style="list-style-type: none"><li>• Emphasis on genetic drift and natural selection</li></ul></li></ul>	<ul style="list-style-type: none"><li>➤ Bamshad.pdf: <i>Signatures of natural selection in the human genome</i></li></ul>
03/29	<ul style="list-style-type: none"><li>➤ Inferring natural selection part I<ul style="list-style-type: none"><li>• Tests of neutrality from each of the following categories:<ul style="list-style-type: none"><li>○ Between-species tests</li><li>○ Within-species tests</li><li>○ Between- and within-species tests</li></ul></li></ul></li></ul>	<ul style="list-style-type: none"><li>➤ Sabeti.pdf: <i>Positive natural selection in the human lineage</i></li></ul>
03/31	<ul style="list-style-type: none"><li>➤ Inferring natural selection part II<ul style="list-style-type: none"><li>• Additional tests of neutrality<ul style="list-style-type: none"><li>○ <math>F_{st}</math> test</li><li>○ Extended haplotype heterozygosity (EEH) test</li></ul></li></ul></li></ul>	<ul style="list-style-type: none"><li>➤ Drake.pdf: <i>Conserved noncoding sequences are selectively constrained and not mutation cold spots</i></li><li>➤ Chen.pdf: <i>Natural selection on human microRNA binding sites inferred from SNP data</i></li></ul>
04/05	<ul style="list-style-type: none"><li>➤ Application of tests of neutrality part I<ul style="list-style-type: none"><li>• Small group discussion of two papers that apply within-species and between-species tests of neutrality to infer modes of selection on regulatory regions of the genome</li></ul></li></ul>	<ul style="list-style-type: none"><li>➤ Mu.pdf: <i>Analysis of genomic variation in non-coding elements using population-scale sequencing data from the 1000 Genomes Project</i></li><li>➤ Tishkoff.pdf: <i>Convergent adaptation of human lactase persistence in Africa and Europe</i></li></ul>
04/07	<ul style="list-style-type: none"><li>➤ Application of tests of neutrality part II<ul style="list-style-type: none"><li>• Small group discussion of two papers that apply within-species and between-species tests of neutrality to infer modes of selection on regulatory regions of the genome</li></ul></li></ul>	<ul style="list-style-type: none"><li>➤ TBD</li></ul>

04/12	➤ Introduction to the coalescent	➤ CC.pdf: <i>The Genome Architecture of the Collaborative Cross Mouse Genetic Reference Population</i>
04/14	• Faculty guest lecture (TBD)	
04/19	• Student guest lectures (TBD)	
04/21	• Review of major concepts	