BioMG 4870 Human Genomics

Instructor: Prof. Andrew G. Clark
227 Biotechnology Bldg.
Office hours: Tues, Thur 12:30 – 1:30 PM
ac347@cornell.edu 255-0527

Teaching assistant: Manisha Munasinghe, mam737@cornell.edu
Office: 101 Biotech
Office Hours: Mondays 1 – 3 PM

Meeting time and place: Tues and Thurs 8:40-9:55 AM, 173 Warren Hall
Prerequisites: BioMG 2810

Course description

The latest in applications of genomics technologies will be united with fundamental concepts of transmission, population, and molecular genetics to understand the molecular basis of genetic disorders. Emphasis will be placed on the role of full genome knowledge in expediting the process of gene discovery. Quantitative concepts will be developed, including the role of statistical inference in interpreting genomic information in inferences from studies designed to test genetic associations. Population genetics, and the central role of variation in the human genome in mediating variation in disease risk, will be examined. Methods such as homozygosity mapping, linkage disequilibrium mapping, and admixture mapping will be studied. We will explore methods for modeling genetic and environmental interactions in mediating complex phenotypes, including disease risk, and critically examine the prospects for progress in disease prediction based on genomic information. The course will be conducted as a series of lectures with classroom discussion of current primary literature.

Readings:

Each lecture will be associated with a reading assignment of current papers posted on the Blackboard website. Background material can come from textbooks like Strachan and Read's *Human Molecular Genetics* or Korf and Irons' *Human Genetics and Genomics*. See also Gibson and Muse *A Primer of Genome Science* and Gibson *A Primer of Human Genetics.*

Assignments and exams.

There will be a series of five problem sets, one mid-term, one final exam and a term paper. Each problem set will be handed out in class and will be due one week later. There will be a midterm exam, and the final exam will be take-home. The term paper will be on a topic in human genomics of your choosing. More info on the term paper will be provided. Problem sets will be handed out Sept 11, Sept 25, Oct 16, Oct 30, and Nov 13. The term paper is due on the last day of lecture, and typically the final is due a week later.

Grading:

The term paper will be worth 250 points, the problem sets/reading assignment write-ups will be worth 250 points, class participation 50 points, the midterm will be worth 200 points and the final exam will be worth 250 points, for a total of 1000 points.

III. Lecture schedule (* = problem set handed out)

Date	Topic
Aug 23	**Survey of the human genome** 1. The scope of human genomics
Aug 28	2. Structure of the human genome
Aug 30	3. The X and Y chromosomes
Sept 4	4. Aneuploidies and chromosome disorders
Sept 6	5. Copy Number Variation
Sept 11*	6. Epigenetics and disease
Sept 13	**Variation and Human Population Genetics** 7. Human variation and massive free datasets
Sept 18	8. Mutation-drift balance – the null model
Sept 20	9. Pathogenic mutations and mutation-selection balance
Sept 25*	10. Linkage disequilibrium and the HapMap project
Sept 27	**Disease genetics** 11. Discovering monogenic disorder genes in humans
Oct 2	12. Molecular basis of disease: CF and PKU
Oct 4	13. midterm exam
Oct 11	14. Mouse and Rat as models for human biology
Oct 16*	15. Drosophila as a model for human disease
Oct 18	16. Personal genomics
Oct 23	17. Human genetic engineering
Oct 25	**Complex trait genetics** 18. Complex traits – overview of the issues
Oct 30*	19. Linkage disequilibrium mapping (GWAS)
Nov 1	20. Homozygosity and Admixture mapping
Nov 6	21. Genetic basis of cancer
Nov 8	22. Genetic basis of resistance to infectious disease
Nov 13*	23. Cardiovascular disease
Nov 15	24. Diabetes and metabolic syndrome
Nov 20	25. Genetics of variation in gene expression
Nov 27	**Human Evolutionary Genetics** 26. Ancestry and the structure of human genetic variation
Nov 29	27. Inference of human origins and Neanderthal genome
Dec 4	28. Which genetic changes make us human?