

**MCB 149 - The Human Genome**  
**UC Berkeley**  
**Spring 2014**

This is an upper division course for majors in MCB with an interest in an in-depth exploration of the forces that shape the human genome and the human population, as well as the ways that human genetic information can be used in medicine, ancestry and forensics. The course will combine lectures and discussion of research papers.

**Lectures:**

M,W,F at 1:00 in Barker Hall 101

**Faculty:**

Michael Eisen - mbeisen+mcb149@gmail.com - Stanley Hall 304B  
Dan Rokhsar  
Barbara Meyer

**Sections:**

1. Monday 2-3 in 125 Li Ka Shing
2. Friday 12-1 in 182 Dwinelle

**TA:**

Shannon Hateley - [shateley@berkeley.edu](mailto:shateley@berkeley.edu)

**Grading:**

There will be three take home midterm exams, each worth 25% of the final grade. All students will present a research paper, which will be worth 15% of the final grade. The remaining 10% will be based on mini quizzes in section and class participation.

**Textbook:**

None. There are no good, up-to-date, textbooks on the human genome. You will instead be assigned readings from the primary literature every week.

**Topics:**

- Gross structure of the human genome
- Content of the human genome
  - Protein-coding and non-coding genes
  - Complexity of splicing, splicing regulation, diversity of isoforms
  - Telomeres and centromeres
  - Transposable elements
  - Regulatory sequences and control of gene expression

- Genetic variation among humans
  - Origins of variation
  - Spread of variation through population
  - Genetic variation across the human population
    - SNPs, indels and structural variants
  - Geographic distribution of variation
    - What is “race”?
    - Admixture
  - Recombination and haplotype structure of human genome
  - Reconstruction of recent and distant history with genetics
    - mitochondria and Y chromosomal history
    - Genghis Kahn
  - Use of DNA in forensics
- Human evolution
  - Comparison of human genome to other primates and mammals
  - Archaic humans
    - Neanderthals and Denisovans
    - Interbreeding between modern and archaic humans
  - Human-specific traits
  - Selection along human lineage
- Phenotype and Disease
  - allelic variation linked to disease, OMIM
  - inferring phenotype from genotype: what’s possible, what are the limitations
  - mitochondrial diseases, somatic variation/heteroplasmy in mtDNA
  - sex-linked traits, dosage compensation and phenotypic variation
- Mapping
  - GWAS
  - Common vs. rare variation in human disease and its impact on mapping
  - “missing” heritability of traits
- Cancer genomics
  - extent of somatic variation
  - types of variation seen in cancer
  - targeted therapeutics
- Personal genomics