BME 580.689 Computational Personal Genomics. This course is cross-listed in Computer Science where it counts as a regular CS course.

Professor: Steven Salzberg, http://ccb.jhu.edu/people/salzberg
Times: Tuesdays and Thursdays, 9:00am – 10:15am
Location: Malone Hall, room 228 *[exception: meeting on Feb 19 in Malone 107]*
Textbook: None, but readings will be required throughout the semester and announced in class or on this syllabus.
GRADING POLICY: Four laboratory assignments are worth 20% each. The in-class presentation is worth 10%, and the final exam is worth 10%.

SYLLABUS

(Note: this is a new course and this schedule will change during the semester; refresh your browser for updates. Readings will be posted here or announced in class.)

Week 1: 27-29 January
Tuesday: Introduction to the course. Biology and genomics background, genome sequencing technology. Course logistics.
Thursday: Motivating example: the discovery of the Huntington's Disease gene. Review of pairwise sequence alignment algorithms.

Week 2: 3-5 February
Tuesday: Sequencing technology, whole genome sequencing, and the first bacterial genome.
Thursday: The human genome project, 1989-2003. The F1000 review site.

Week 3: 10-12 February
Tuesday: Alignment of whole genomes, alignment of a short query against a genome.
Thursday: Methods for SNP discovery, from anthrax to human.

Week 4: 17-19 February *[NOTE: meeting on Feb 19 in Malone Hall, room 107]*
Tuesday: Personal genomics: sequencing your genome. 23andMe: a case study
Thursday: RNA-Seq analysis: reads to transcripts to quantification. Part 1, spliced alignment. Analysis of cancer genomes to find fusion genes.

Week 5: 24-26 February
Tuesday: ***Lab 1 due.*** RNA-Seq analysis continued: assembly and quantification with Cufflinks and StringTie.
Thursday: Annotation: a brief history of gene finding and the human gene count. Introduction to computational gene finding.

Week 6: 3-5 March
Tuesday: The status of the human reference genome today: assembly, annotation, and single nucleotide polymorphisms (SNPs). The UCSC Genome Browser.
Thursday: Data mining with IGV. Systematic sequencing errors and the problems they cause. RNA-DNA differences: RNA editing and the perils of large-scale genome data.

Week 7: 10-12 March
Tuesday: Using genome data to find relatedness between people. Session on ethics: gene patents and the BRCA1 and BRCA2 genes.
Thursday: ***Lab 2 due***. What makes people live longer? Sequencing the very old to find variants related to longevity.

Spring break 16-20 March

Week 8: 24-26 March
Tuesday: In-depth analysis of a paper: M. Meyer *et al.*, A High-Coverage Genome Sequence from an Archaic Denisovan Individual**,** *Science* 12 October 2012, pp 222-226.
http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3617501
Thursday: Exome analysis: exome sequencing to discover the causes of disease. The DIAMUND algorithm.

Week 9: 31 March – 2 April
Tuesday: Genome assembly: introduction to the problem and the main algorithms. Assembly of short reads and hybrid assembly of short and long reads.
Thursday: Student presentations of selected papers. (10-15 minutes per presentation, 5 per class).

Week 10: 7-9 April
Tuesday: ***Lab 3 due.*** Student presentations of selected papers (10-15 minutes per presentation, 5 per class).
Thursday: Student presentations of selected papers (10-15 minutes per presentation, 5 per class).

Week 11: 14-16 April
Tuesday: The human microbiome: introduction. Biological results: obesity-related microbes.
Thursday: 16S sequencing data and methods for analysis.

Week 12: 21-23 April
Tuesday: Fast k-mer counting and the Kraken algorithm.
Thursday: The 2014 Ebola story.

Week 13: 28-30 April
Tuesday: ***Lab 4 due.*** Topic TBD.
Thursday: In-class final exam, open book/open laptop.