BIOS 234 Reading List

1. Genetics & Health
   a) “Human genetics: One gene, twenty years” Nature 2009
   b) “Charting a course for genomic medicine from base pairs to bedside” Nature 2011
      http://www.nature.com/nature/journal/v470/n7333/full/nature09764.html
   c) “Genomics reaches the clinic: From basic discoveries to clinical impact” Cell 2011
   d) “Cutting the Gordian Helix – Regulating Genomic Testing in the Era of Precision Medicine” NEJM 2015

2. The Human Genome Project
   a) “Initial sequencing and analysis of the human genome” Nature 2001
      http://www.nature.com/nature/journal/v409/n6822/full/409860a0.html
   b) Interview with Eric Green, 2013:
      http://www.smithsonianmag.com/science-nature/the-work-is-only-beginning-on-understanding-the-human-genome-89390748/?no-ist

3. Next Generation DNA Sequencing
   a) “Next-Generation DNA Sequencing Methods” Annual Reviews 2008
4. DNA Sequence Analysis

4.0 Misc

a) “Analysis of genetic inheritance in a family quartet by whole-genome sequencing” Science 2010


b) “Accurate and comprehensive sequencing of personal genomes” Genome Research 2011

http://genome.cshlp.org/content/21/9/1498.long

c) “SpeedSeq: ultra-fast personal genome analysis and interpretation” Nature Methods 2015

http://www.nature.com/nmeth/journal/v12/n10/full/nmeth.3505.html
4.1 Alignment

d) “Fast and accurate short read alignment with Burrows-Wheeler transform” Bioinformatics 2009

e) “Aligning sequence reads, clone sequences and assembly contigs with BWA-MEM”

f) “A survey of sequence alignment algorithms for next-generation sequencing” Briefings in Bioinformatics 2010
   http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2943993/

4.2 SNPs

  g) “Genotype and SNP calling from next-generation sequencing data” Nature 2011
     http://www.nature.com/nrg/journal/v12/n6/full/nrg2986.html

  h) “Low concordance of multiple variant-calling pipelines: practical implications for exome and genome sequencing” Genome Medicine 2013
     http://genomemedicine.com/content/5/3/28

4.3 INDELs


  j) “Small insertions and deletions (INDELs) in human genomes” Human Molecular Genetics 2010
     http://hmg.oxfordjournals.org/content/19/R2/R131.abstract
4.4 Copy Number Variants

k) “Human Copy Number Variation and Complex Genetic Disease” Annual Review of Genetics 2011


l) “Computational tools for copy number variation (CNV) detection using next-generation sequencing data: features and perspectives” BMC Bioinformatics 2013

http://www.biomedcentral.com/1471-2105/14/S11/S1

4.5 Structural Variants

m) “Genome structural variation discovery and genotyping” Nature Reviews Genetics 2011

http://www.nature.com/nrg/journal/v12/n5/full/nrg2958.html

n) “Detection of structural DNA variation from next generation sequencing data: a review of informatics approaches” Cancer Genetics 2013


4.6 Prioritization & Interpretation

o) “A survey of tools for variant analysis of next-generation genome sequencing data” Briefings in Bioinformatics 2013

http://bib.oxfordjournals.org/content/early/2013/01/21/bib.bbs086.full

p) “Genic Intolerance to Functional Variation and the Interpretation of Personal Genomes” PLoS Genetics 2013

http://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1003709

q) “Sequence to Medical Phenotypes: A Framework for Interpretation of Human Whole Genome DNA Sequence Data” PLoS Genetics 2015

http://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1005496
5. Large Scale Projects

a) “An integrated map of genetic variation from 1,092 human genomes” Nature 2012


   http://www.nature.com/nature/journal/v526/n7571/full/nature15393.html

   http://www.nature.com/nature/journal/v526/n7571/full/nature15394.html

e) “Analysis of protein-coding genetic variation in 60,706 human” bioRxiv
   http://biorxiv.org/content/early/2015/10/30/030338

f) “Integrating human sequence datasets provides a resource of benchmark SNP and indel genotype calls” Nature Biotechnology 2014
   http://www.nature.com/nbt/journal/v32/n3/full/nbt.2835.html

6. Clinical Genomes

a) “Clinical Assessment Incorporating a Personal Genome” The Lancet 2010
   http://www.thelancet.com/journals/lancet/article/PIIS0140-6736%2810%2960452-7/abstract

b) “Clinical Interpretation and Implications of Whole Genome Sequencing” JAMA 2014
c) “Personal Omics Profiling Reveals Dynamic Molecular and Medical Phenotypes” Cell 2012
   http://www.cell.com/abstract/S0092-8674(12)00166-3

d) “Molecular Diagnosis of long QT syndrome at 10 days of life by rapid whole genome sequencing” Heart Rhythm 2014
   http://www.heartrhythmjournal.com/article/S1547-5271%2814%2900692-4/pdf

e) “Evolution of an adenocarcinoma in response to selection by targeted kinase inhibitors” Genome Biology 2010
   http://genomebiology.com/2010/11/8/r82

f) “Points to consider in the clinical application of genome sequencing” Genetics in Medicine 2012
   http://www.nature.com/gim/journal/v14/n8/full/gim201274a.html

g) “ACMG clinical laboratory standards for next-generation sequencing” Genetics in Medicine 2013
   http://www.nature.com/gim/journal/v15/n9/full/gim201392a.html

f) “A research roadmap for next-generation sequencing informatics” Science Translational Medicine 2016
   http://stm.sciencemag.org/content/8/335/335ps10.full.pdf+html

7. Pharmacogenomics


b) “Genetic variant in folate homeostasis is associated with lower warfarin dose in African Americans” Blood 2014
   http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4183989/
8. Prenatal sequencing

a) “Maternal Plasma DNA Sequencing Reveals the Genome-Wide Genetic and Mutational Profile of the Fetus” Science Translational Medicine 2010

http://stm.sciencemag.org/content/2/61/61ra91.abstract

b) “Noninvasive Whole-Genome Sequencing of a Human Fetus” Science Translational Medicine 2012

http://stm.sciencemag.org/content/4/137/137ra76.abstract

9. Ethical Concerns

a) “Identifying Personal Genomes by Surname Inference” Science 2013

http://www.sciencemag.org/content/339/6117/321

b) “The uneasy ethical and legal underpinnings of large-scale genomic biobanks” Annual Reviews 2007