

## Balancing research, health and patient privacy in the genomics era Reading list

### Mandatory readings:

- DNA variants:  
Browse the 1000 genomes project site:  
<http://www.1000genomes.org/>
- Informed consent issues:  
Points to consider for informed consent for genome/exome sequencing. (2013). ACMG Board of Directors. Genetics in Medicine 15, 748–749  
doi:10.1038/gim.2013.94 <http://www.nature.com/gim/journal/v15/n9/full/gim201394a.html>
- De-identification and privacy issues:  
Gymrek M, McGuire AL, Golan D, Halperin E, Erlich Y. (2013) Identifying Personal Genomes by Surname Inference. Science. Jan 18;339(6117):321–  
4. <http://www.sciencemag.org/content/339/6117/321>

Also read the commentaries on this article. <http://www.sciencemag.org/content/339/6123/1032.2.full.pdf>

- Malin B, Loukides G, Benitez K, Clayton EW. (2011) Identifiability in biobanks: models, measures, and mitigation strategies. Hum Genet. Sep;130(3):383-92. doi: 10.1007/s00439-011-1042-5. <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3621020/>
- Commentary on 23 and me:  
  
<http://www.scientificamerican.com/article/23andme-is-terrifying-but-not-for-reasons-fda/>
- Genetic discrimination:  
Watch the youtube trailer from the GATTACA movie:  
[https://www.youtube.com/watch?v=ZppWok6SX88&list=PLN-SnMXbRhkMkyOCE1-wYq0V8Q6I\\_YMT](https://www.youtube.com/watch?v=ZppWok6SX88&list=PLN-SnMXbRhkMkyOCE1-wYq0V8Q6I_YMT)  
<http://www.wired.com/2008/05/the-genetic-inf/>
- Sequencing your own genome  
listen to the NPR story:  
<http://www.npr.org/blogs/health/2012/09/19/160955379/scientists-see-upside-and-downside-of-sequencing-their-own-genes>  
or read the transcript:  
<http://www.npr.org/templates/transcript/transcript.php?storyId=160955379>
- Clinical care implications of genome medicine:  
ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing  
[https://www.acmg.net/docs/ACMG\\_Releases\\_Highly-Anticipated\\_Recommendations\\_on\\_Incidental\\_Findings\\_in\\_Clinical\\_Exome\\_and\\_Geno](https://www.acmg.net/docs/ACMG_Releases_Highly-Anticipated_Recommendations_on_Incidental_Findings_in_Clinical_Exome_and_Geno)

[me Sequencing.pdf](#)

- Burke W, Trinidad SB, and Clayton EW. Seeking Genomic Knowledge: The Case for Clinical Restraint. *Hastings Law J.* 2013; 64(6): 1650-1654.  
<http://www.hastingslawjournal.org/wp-content/uploads/2014/03/Burke-Trinidad-Clayton-64.6.pdf>

#### Optional readings:

- Kahn SD. (2011) On the future of genomic data. *Science.* Feb 11;331(6018):728-9. doi: 10.1126/science.1197891. <http://www.sciencemag.org/content/331/6018/728.full>
- Clayton, Ellen Wright. (2003) Ethical, legal, and social implications of genomic medicine." *New England Journal of Medicine* 349.6: 562-569.  
<http://www.nejm.org/doi/full/10.1056/NEJMra012577>
- Sethi P, and Theodos, K. (2009) Translational Bioinformatics and Healthcare Informatics: Computational and Ethical Challenges. *Perspect Health Inf Manag.* <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2804463/>
- Kaye J. (2012) The tension between data sharing and the protection of privacy in genomics research. *Annu Rev Genomics Hum Genet.* 13:415-31. doi: 10.1146/annurev-genom-082410-101454. Epub 2012 Mar 9.  
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4337968/>
- Downing NR, Williams JK, Daack-Hirsch S, Driessnack M, Simon CM. (2013) Genetics specialists' perspectives on disclosure of genomic incidental findings in the clinical setting. *Patient Educ Couns.* Jan;90(1):133–8.  
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3522795/?report=classic>
- Review the content on this website:  
<http://www.vanderbilt.edu/or/cores/biovu-vanderbilt-dna-databank/>
- Medicine's Problem of 'Incidental Findings' (not specific to genomics, but a nice perspective on modern medicine issues) <http://www.theatlantic.com/health/archive/2013/12/medicines-problem-of-incidental-findings/282294/>
- Illumina's DNA Supercomputer Ushers in the \$1,000 Human Genome  
<http://www.businessweek.com/articles/2014-01-14/illuminas-dna-supercomputer-ushers-in-the-1-000-human-genome>
- Baby DNA Analysis Ushers in Brave New World of Treatment <http://www.bloomberg.com/news/2014-01-16/baby-dna-analysis-ushers-in->

[brave-new-world-of-treatment-health.html](http://brave-new-world-of-treatment-health.html)

- Illumina and a Billionaire Want to Jump-Start Genomics  
Upstarts <http://www.businessweek.com/articles/2014-02-17/illumina-and-billionaire-yuri-milner-to-aid-genomics-startups>
- Read about DNAdigest <http://dnadigest.org/frequently-asked-questions/>