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Examining genomics and your health, Part 1: Your genome and your health

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Genomics and Your Health

Part I: Your genome and your health: Personalized medicine

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What does genomics mean?

- According to the Merriam-Webster Dictionary:
 - a branch of biotechnology concerned with applying the techniques of genetics and molecular biology to the genetic mapping and DNA sequencing of sets of genes or the complete genomes of selected organisms, with organizing the results in databases, and with applications of the data (as in medicine or biology) — compare PROTEOMICS
 - First known use of the word, in 1987

The Human Genome Project

- Exploring our molecular selves (movie clip)
- http://www.youtube.com/watch?v=_EK3g6px7Ik

Personal Genomics in Medicine



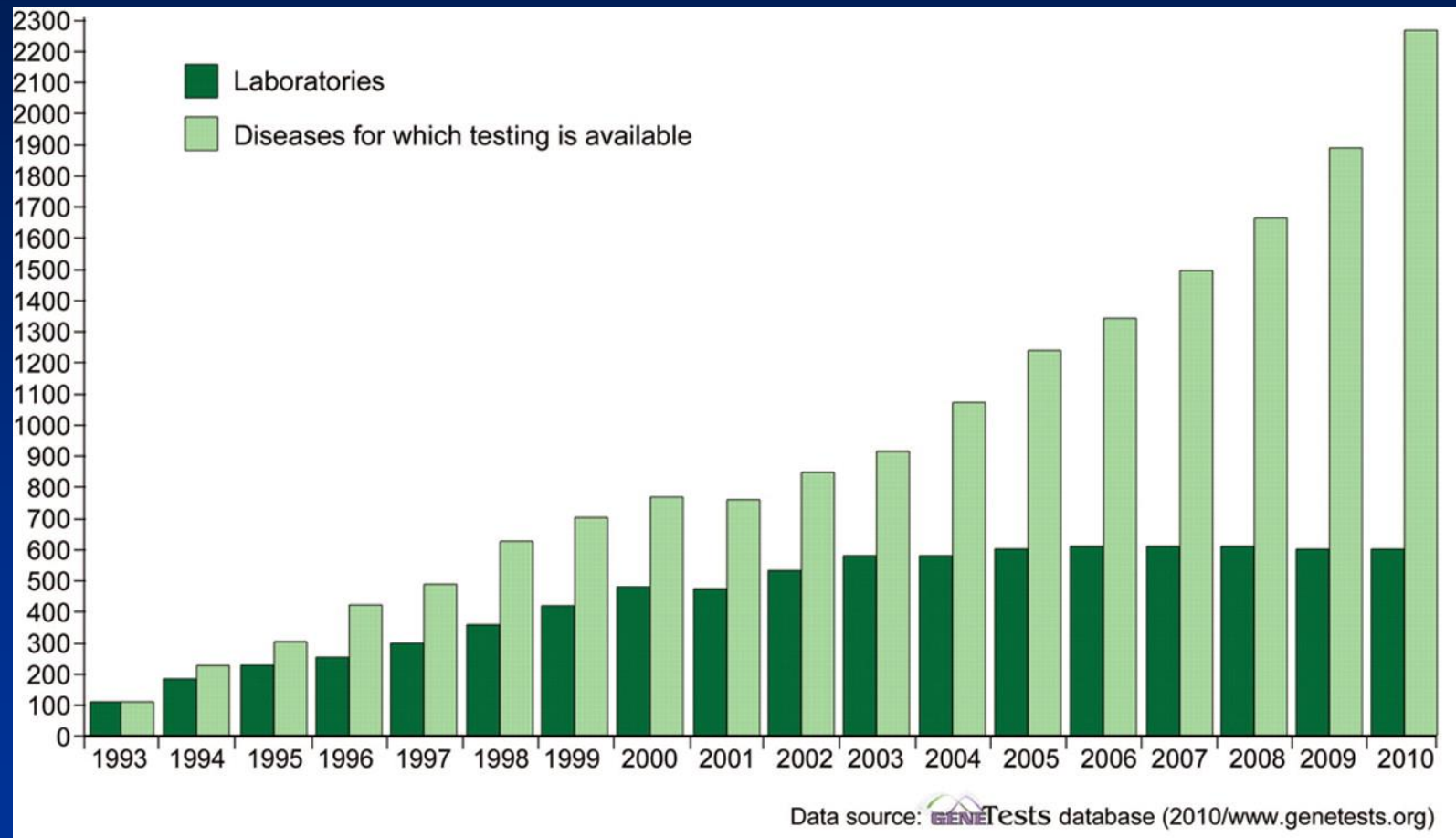
Taken from:

http://www.dukehealth.org/health_library/health_articles/controversies_in_medicine_direct_to_consumer_genetic_testing

What might genomics mean for you?

- You can get your own entire genome sequenced for about a couple of thousand dollars, maybe less
- You can have your DNA screened for \$99 (about 200 health and ancestry genetic markers) companies like www.23andme.com

Genetic Testing is Growing



Taken from: <http://www.dementiatoday.com/neurogenetics-five-new-things/>

In May 2010 – Walgreens was going to start selling a personal genomics test kit, then stopped

- <http://www.thehealthculture.com/2010/05/genetic-testing-walgreens-says-will-then-wont/>
- News clip about the decision

Some potential problems with personalized genomics testing

- What do the results really mean?
- Do you really want to know?
- Just having one gene or not does not mean you will or won't get the gene.
- Importance of environmental factors
- Just because we know what a gene may do we likely do not have any treatment for it (yet?)
- Need to put in context of family history, if possible

Some beneficial examples of gene testing

- We are just scratching the surface of what personal genomics may be able to tell us
- In several cases, knowing your gene for something has led to better courses of treatment
- Knowing the genetic changes in the disease (such as a cancerous tumor) can help select more targeted medicine

Has a doctor ever asked you if you would like to have a genetic test?

- Screening tests exist for a variety of diseases and health conditions
 - Breast cancer
 - Diabetes
 - Cardiovascular disease (heart attack and stroke)
 - Alzheimer's disease
 - Ability to respond to particular medications
 - Many others

What will a doctor do with the information?

- My mom's experience with genetic testing and her apparently high cholesterol report
- She has a gene variant that makes diet and exercise much more effective (apparently) than the medicine (gene test not FDA approved) – part of Health Diagnostic Laboratory, Inc.
www.myhdl.com
- Doctor still prescribed the typical drug, Crestor.
- Mom is changing diet and exercise.
- What does this genetic test really mean?

Personalized Medicine

- Pharmacogenomics (PGx)
- The study of genetic variation and medication response
- Coriell Personalized Medicine Collaborative (and others)
- <http://cpmc1.coriell.org/personalized-medicine>

Many drugs/medicines work best if you have a particular gene change (mutation)

- Gleevec (imatinib); Iressa (gefitinib); Tarceva (erlotinib)
- Coumadin (warfarin)
- Plavix (clopidogrel)
- Tamoxifen
- Cetuximab (Erbitux)

Drugs with Pharmacogenomics Tests

(adapted from Singer, 2010)

Drug	Purpose	The gene factor
Atomoxetine HCl (Strattera)	ADHD treatment	CYP2D6 gene mutation → liver damage
Clopidogrel (Plavix)	Inhibits blood clots	Variant of CYP2C19 gene interferes with drug metabolism → ineffective
Cetuximan (Erbitux) and Panitumumab (Vectibix)	Colorectal cancer	Tumors must have a normal KRAS gene
Gefitinib (Iressa)	Lung cancer	Works best on tumors with a EGFR gene mutation
Irinotecan (Camptosar)	Colorectal cancer	Side effects due to fewer liver enzymes
Tamoxifen (Nolvadex)	Breast cancer	Variations in CYP2D6 → metabolize drug too quickly or not at all
Warfarin (Coumadin)	Blood thinner	Gene variants can lead to excessive bleeding instead of preventing clots → correct dose

Targeted Therapies Show Great Promise

- Charlie Rose Interviews Dr. Brian Druker and others about Gleevec

<http://www.youtube.com/watch?v=zurZqAFtX5Y>

- Targeted therapies help us with treating Chronic Myeloid Leukemia and other cancers.

How might one use personalized medicine?

- John's story from:
- Coriell Personalized Medicine Collaborative

<http://cpmc1.coriell.org/personalized-medicine/meet-john>

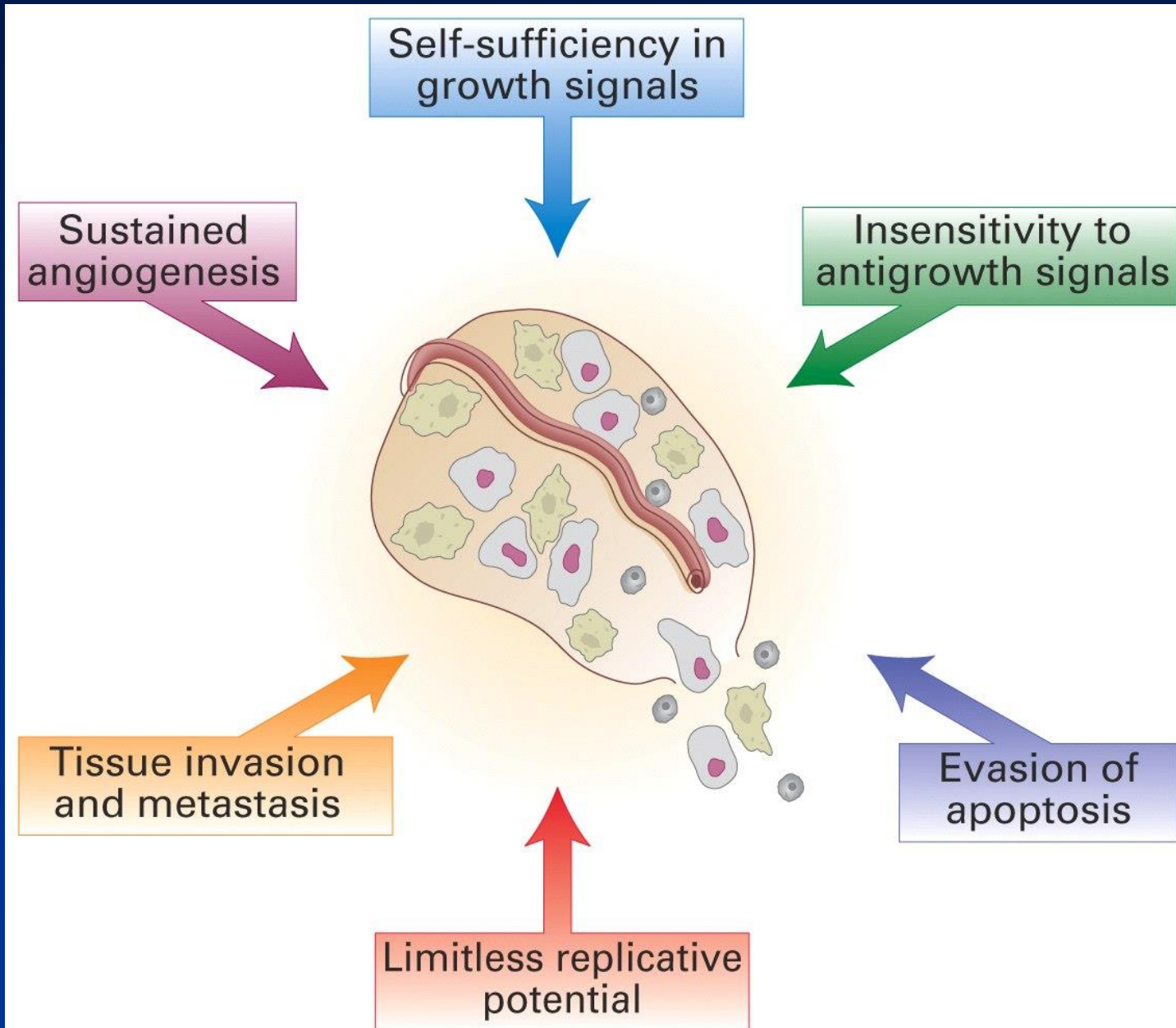
The Cancer Genome Atlas

Taken from <http://cancergenome.nih.gov/cancersselected>

Cancers selected for study were chosen based on specific criteria that include:

- Poor prognosis and overall public health impact
- Availability of human tumor and matched-normal tissue samples that meet TCGA standards for patient consent, quality and quantity

Changes in cells that cause cancer



Taken from:
Lodish et al., 2012

The idea behind tumor gene sequencing

- Find out which genes are specifically turned on or off (affected) in the tumor
- Select a drug that specifically targets that change
- First patient to use this approach, having his own tumor sequenced and compared against his own healthy cells
 - Retired physician (fall 2008)
 - Adenocarcinoma of the tongue; metastasized to the lungs
 - Drugs chosen using the specific tumor genome model
 - Staved off tumor growth for several months
 - Patient died of the cancer one year later

Gene Sequencing will lead to better cancer treatments!

- <http://cancergenome.nih.gov/newsevents/multimedialibrary/videos/dnasequencing>
- Video clip explaining the significance of The Cancer Genome Atlas (TCGA)

Genome sequencing may also
help you before you get sick...



The story of Michael Snyder



iPOP – Integrative Personal Omics Profiling –
The Snyderome

<http://snyderome.stanford.edu/>

The story of Michael Snyder

- 2010, 54 year old healthy molecular geneticist, Stanford scientist
- Sequenced his genome, made public, many medical tests
- Genetic predisposition to type 2 diabetes, a surprise
- 14 month study, glucose levels spiked following a respiratory infection
- He was able to reverse onset of diabetes through diet and exercise (*Cell*, March 16, 2012)

Others are studying nutrigenomics

- How nutrition can influence the expression of genes ultimately affecting a person's health (Sardi, 2011)

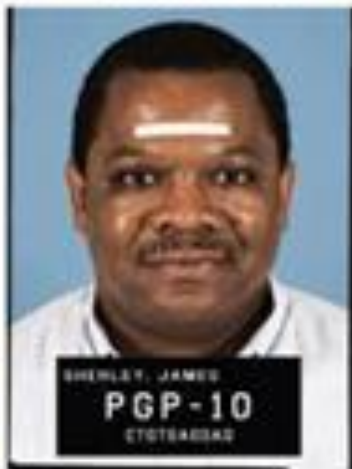
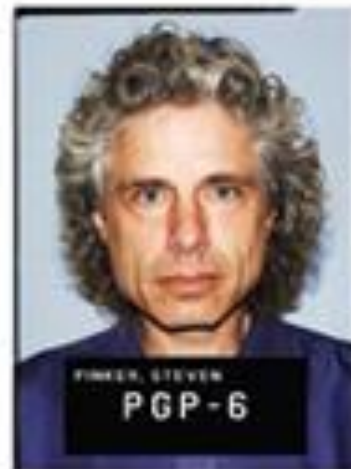
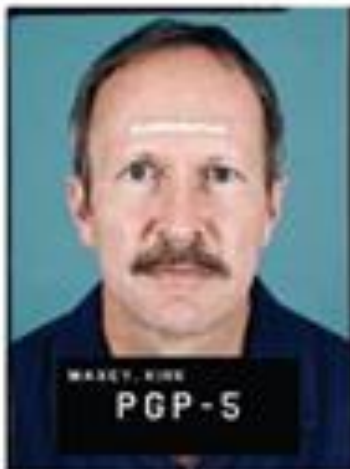
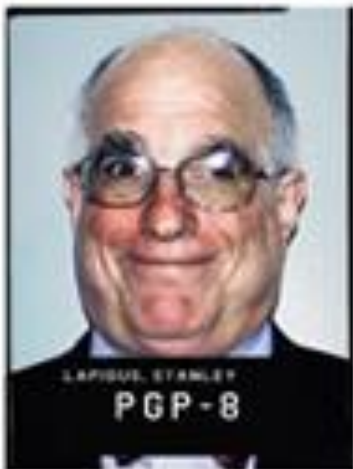
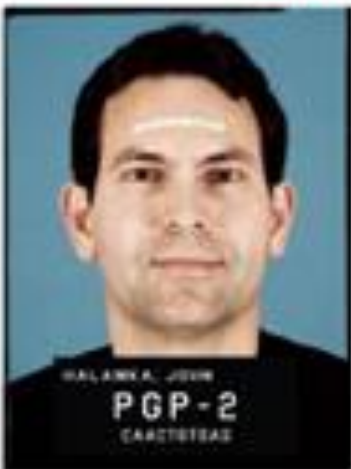
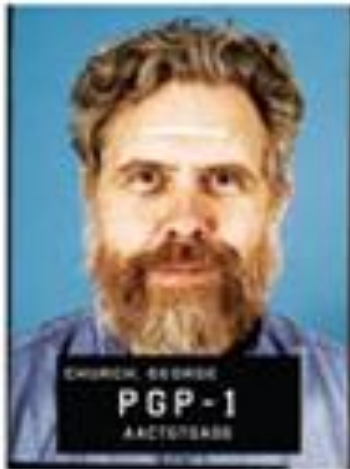
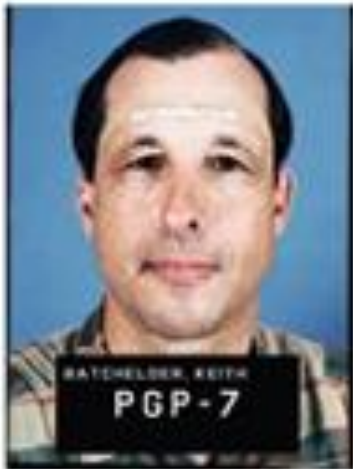


Taken from:
<http://www.healthymeal-experts.com/category/healthy-food-for-kids/>

Landmark Project

- The Personal Genome Project (PGP)
- Started by Dr. George Church, Harvard Medical School in 2007
- 9 more people joined him
- 10,000 people are signed up; project will expand to 100,000 people!
- What will this mean for the future of medicine?

The PGP-10



Personal Genome Project

Steven Pinker, Ph.D. Harvard Psychologist

Pinker, 2009

http://www.nytimes.com/2009/01/11/magazine/11Genome-t.html?_r=0



But, What does whole genome sequencing mean to us?

- Gene screening for particular genes known to cause disease; carrier screening (Tay-Sachs, Huntington's, Cystic Fibrosis, etc.) available from companies such as Counsyl <https://www.counsyl.com/> “The first step to a healthy pregnancy”.
- “Geno’s Paradox – personal genomics will be more recreational than diagnostic for some time to come.”
- What does it mean to be 15% more likely to be predisposed to Alzheimer’s Disease, for example...

Putting genomics in perspective

- We all have about 25,000 known genes; 1% of the genome; termed the exome.
- Need to know family history.
- Will whole genome sequencing tell us a lot of what we could have already guessed?
- There are relatively few “high profile genes”.
- What do the rest of the genes do?
- Will you want to post your genome on facebook?
- What will your relatives think about that (it is partly their genome too)?

Personal Genome Project

- <http://www.personalgenomes.org/>
- An interview with Dr. George Church
Founder; 10 participants
- [http://www.youtube.com/watch?v=mVZI7
NBgcWM](http://www.youtube.com/watch?v=mVZI7NBgcWM)

NOVA Documentary

- Premiered in March 2012
- *Cracking your genetic code*
- <http://video.pbs.org/video/2195620092/> (trailer)
- <http://www.pbs.org/wgbh/nova/body/cracking-your-genetic-code.html> (the whole 60 minute program)

TED Talk about Genomics

- Richard Resnick – Welcome to the genomic revolution.
- http://www.ted.com/talks/richard_resnick_welcome_to_the_genomic_revolution.html

Can genes be patented?

- Most of the human genome already is, many times...
- Supreme Court case and Myriad Genetics – April 15, 2013
- BRCA1 and 2 (Breast cancer genes)
- Myriad Genetics invested \$500 million to develop BRCA testing
- Company made \$405.5 million last year (mostly from BRCA testing)

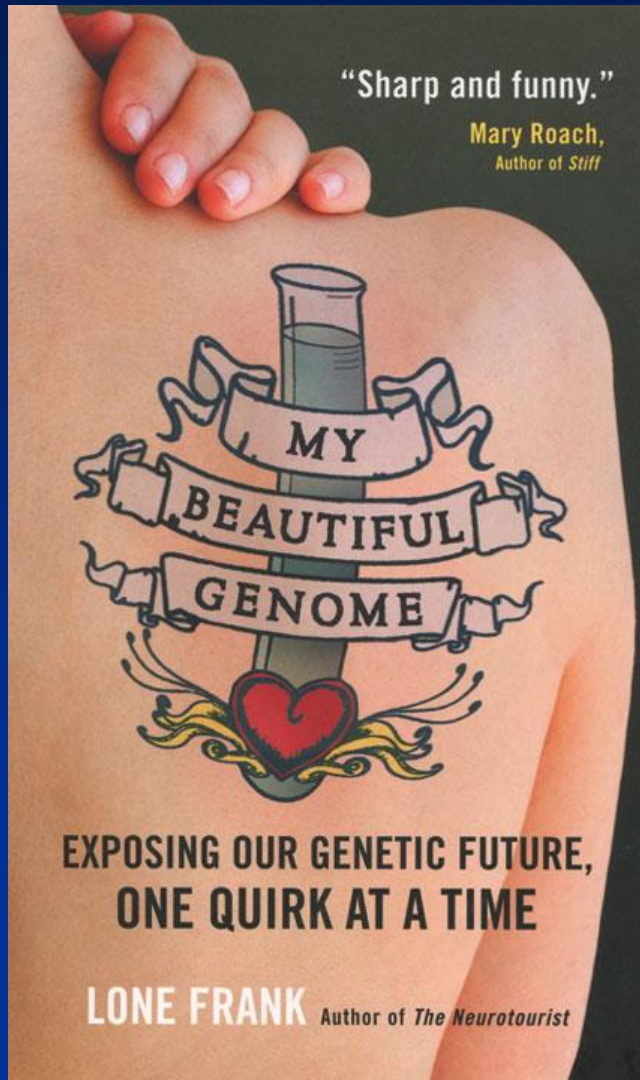
A patient's breast cancer was missed

- Case file for the Supreme court in 2009
- Technology has advanced since then
- Plaintiff in the case; tested negative for breast cancer
- Myriad refused to let other researchers (Yale Cancer Center) look for mutations in this patient or others that they suspected were missed; insurance would not pay until later
- Myriad released additional genetic tests
- Patient's daughter already developed cancer in the meantime

What about this?

- Costs less money to sequence the entire genome than to pay \$4000 to Myriad for just 2 gene tests (BRCA1 and BRCA2)
- Whole genome sequencing might not infringe on patents on isolated genes?
- Stay tuned for developments...

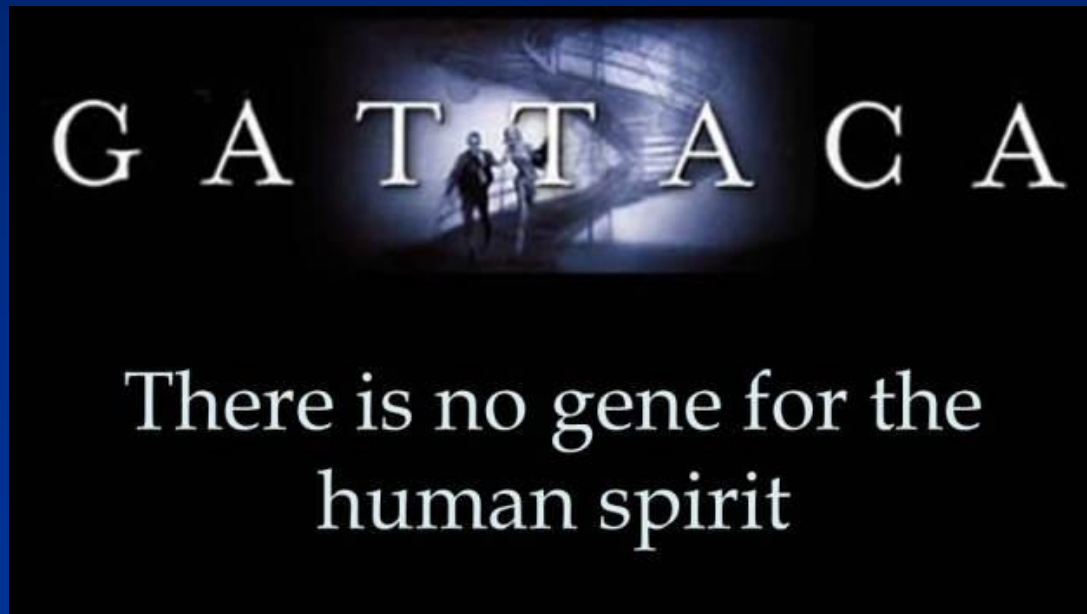
Great Book/Overview of Personal Genomics



*My Beautiful Genome:
Exposing our genetic future
one quirk at a time* by Dr.
Lone Frank, neuroscientist

http://www.sciencenews.org/view/generic/id/337601/description/BOOK_REVIEW_My_Beautiful_Genome_Exposing_Our_Genetic_Future_One_Quirk_at_a_Time_by_Lone_Frank

Great Movie (cautionary tale)
about a futuristic world where we
are define by our genetic code
Gattaca, 1997



- <http://explorersofthemind.wordpress.com/>

Personal Genomics: Do you want to know?



Wellcome Images

Image taken from: (left) <http://dailycloudinfo.com/2013%E2%80%B2s-pandoras-box-the-cyberattack/>

(right) http://www.medgadget.com/2009/02/a_long_difficult_future_foreseen_for_personalized_genetic_medicine.html

More Resources

- *Curing Cancer*, PBS documentary episode 4 is about gene targeted therapy for CML and breast cancer specifically

<http://www.pbs.org/wnet/dna/episode4/>

References

- <http://explorersofthemind.wordpress.com/>

What do you think? What would you do?

- In small discussion groups, consider some scenarios from the book; something to discuss with your family and friends?
- *Your Genes, Your Choices* by Catherine Baker published by the American Association for the Advancement of Science, AAAS, 1997.

*Coming next week:
Genomics and Your Health*

90% of your cells are not even human... 9 out of 10 cells in your body are not you!!!!

Let's explore the genomics of them...
the aliens inside of us.

Coming next week:
Genomics and Your Health

Part II: Genomics of your
microbiome and your health

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