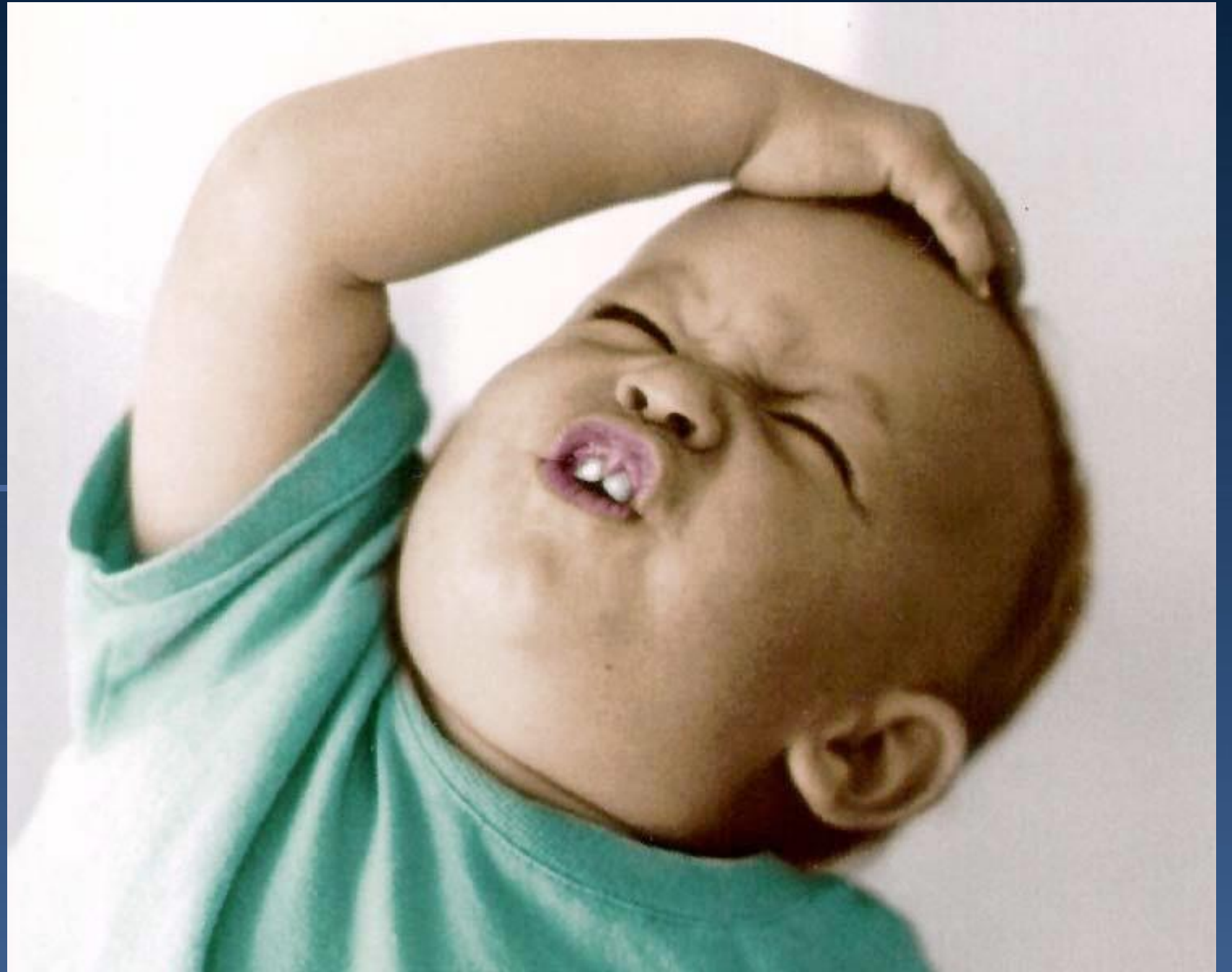


Tests I Wish You'd Never Ordered 2021 *“Here We Go Again!!!”*

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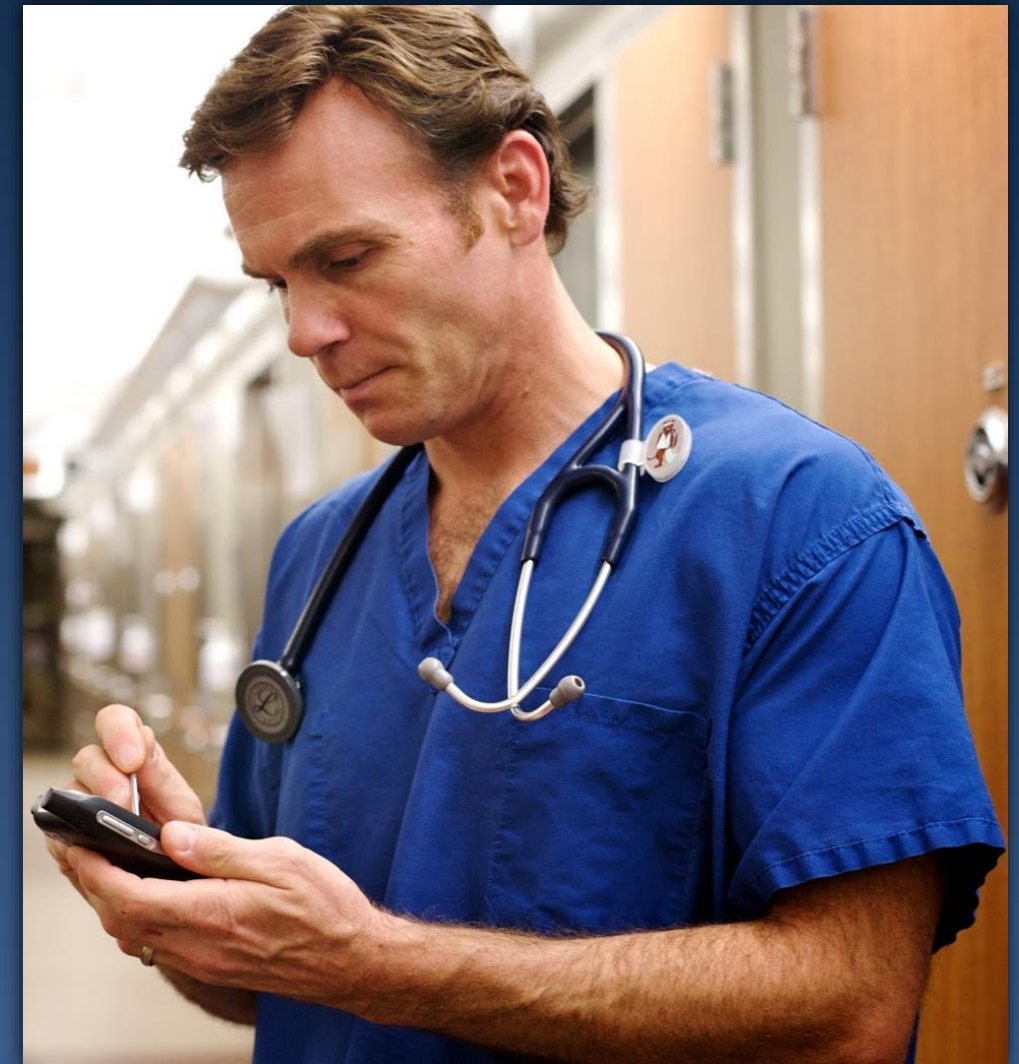
Disclosure of Financial Relationships



I have no conflict of interest with the information presented herein.

Case History

- You know it's going to be a bad day when your colleague calls you on the phone and the conversation begins with...“I'm really sorry, but...”



Case History

- 45 year-old woman with a history of anxiety and hypertension who had a DNA test run by a national company. She presents for further “counseling” for her DNA results.
- “My doctor says you know more about DNA than anyone he knows of. So...”

“...I really need to introduce him to some new friends...”

Case History

- Her family history was negative
- Her personal history was otherwise negative; she had mild anemia while pregnant with both of her children and she had hypertension and anxiety
- She wanted testing done to see what she might “have”
- The list: coronary artery disease, hyperlipidemia, hypertension, diabetes mellitus, chronic kidney disease, difficulty metabolizing anticonvulsants, a couple of possible glycogen storage disorders, dementia, HFE gene positivity, and MTHFR gene mutations

Case History

- My questions...
 - Why did you get the test?
 - What do you want to do with the results you have?
 - How does this information help you in managing your health?

DNA Testing

- Definitions
 - Whole genome sequencing (WGS)
 - Sequences the entire human genome
 - Whole exome sequencing (WES)
 - Sequences only the gene-coding portion of the genome (exome)

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- Muscle Composition

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DATA YOU MIGHT ALREADY TRACK

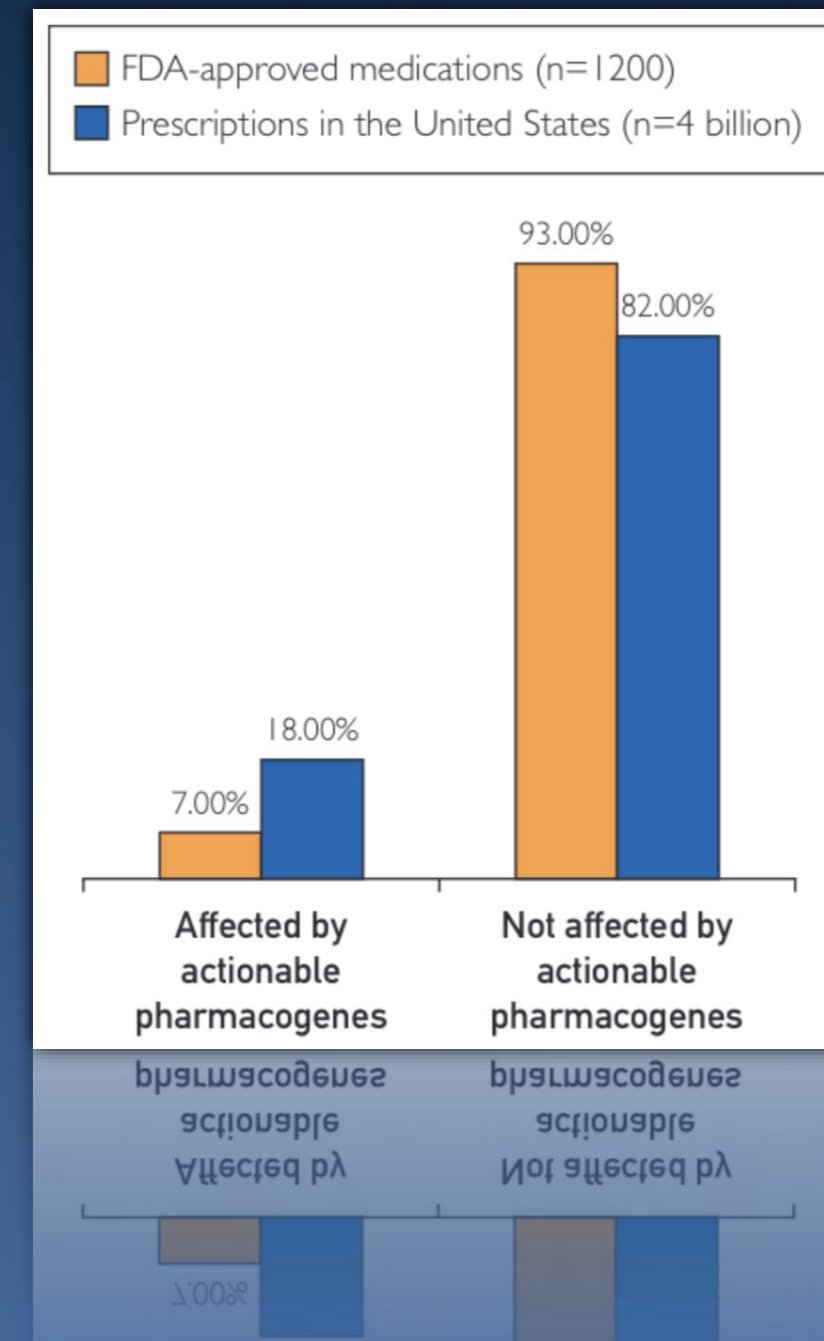
- Number of steps
- Hours slept
- Family history
- Lifestyle choices

Introductory Offer

Case History

- Well, maybe...
 - 7% of 1200 FDA approved medications are affected by actionable pharmacologic genomics
 - Cost effective?
 - Whole genome - no
 - Focused testing - maybe?

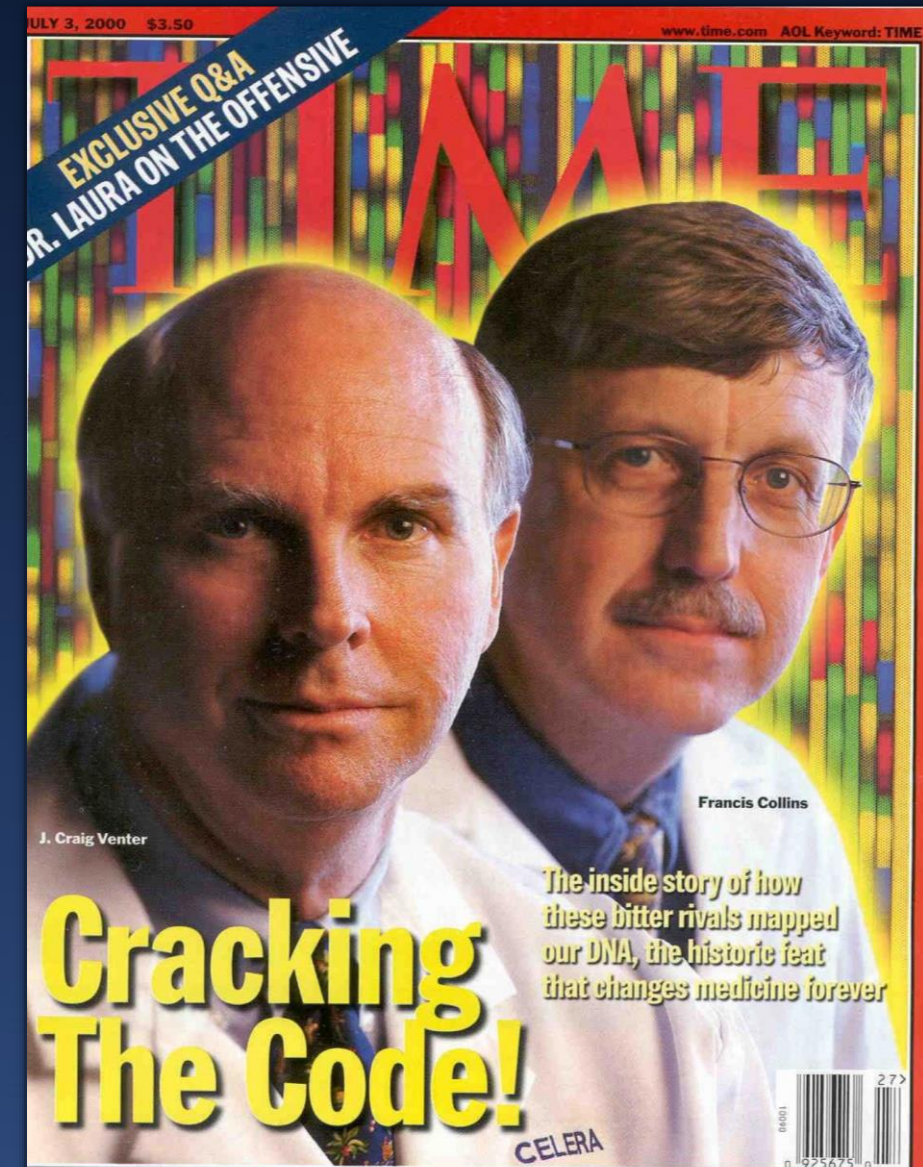
Source: United States FDA



History of Genetic Screening

“I would be willing to make a prediction that within 10 years, we will have the potential of offering any of you the opportunity to find out what particular genetic conditions you may be at increased risk for, based upon the discovery of genes involved in common illnesses like diabetes, hypertension, heart disease, and so on.”

~Francis Collins, June 26, 2000



DNA Testing - Benefits

- Can provide some insight to risk for illness in later life
- Fairly inexpensive (< \$1000 in many cases)
- Effective Use for Patients and Physicians
 - Carpenter MJ, et al., *Ann Behav Med.* 2007;33(1):22
 - 199 smokers, patients homozygous for a severely deficient, alpha-1-antitrypsin null allele were significantly more likely to attempt to quit smoking (59%) than either mutation carriers (34%) or individuals with a normal genotype (26%)

DNA Testing - Benefits

- Effective Use for Patients and Physicians
 - Sanderson SC, Michie S, *Clin Genet.* 2007;71(6):501
 - Study of 261 smokers asked to consider one of two randomly assigned hypothetical scenarios, subjects who were informed of having genetic risk factors for heart disease were more likely to report an inclination to quit smoking than individuals in a high-risk (but non-genetic) group
 - Decisions were motivated by the belief that smoking cessation would translate to reductions in heart disease

DNA Testing - Benefits

- Effective Use for Patients and Physicians
 - Huijgen R, et al., *PLoS One*. 2010;5(2):e9220
 - Study of 781 individuals who were carriers of mutations that cause familial hypercholesterolemia (ascertained through an affected relative), cholesterol-lowering medication use increased from 51% to 81% two years after learning genotype status
 - Significant reductions in LDL-C levels were noted, although normalized levels were not achieved in most subjects

DNA Testing - Burdens

- Genetic screening may have disastrous results...
 - Prospective spouse finds something in the history of self or other that is alarming
 - Concern about having children for fear of passing on “undesirable” traits
 - Unintended disclosure - are you required to disclose information that you didn't order?
 - Psychological impact - the patient and the physician!



DNA Testing

- Having the gene vs having the disease
 - Many genetic alterations occur in non-genetic portions of the genome
 - Having a risk for something does not mean getting something
 - Some relative risks for disease development may fit well into a patient's lifestyle and not result in a change of lifestyle habits

From the “Choosing Wisely” campaign...

- Don't order a duplicate genetic test for an inherited condition unless there is uncertainty about the validity of the existing test result.
- Don't order APOE genetic testing as a predictive test for Alzheimer disease.
- Don't order MTHFR genetic testing for the risk assessment of hereditary thrombophilia.

From the “Choosing Wisely” campaign...

- Don't order HFE genetic testing for a patient without iron overload or a family history of HFE-associated hereditary hemochromatosis.
- Don't order exome or genome sequencing before obtaining informed consent that includes the possibility of secondary findings.

Help For Frustrated Internists

- The American College of Medical Genetics and Genomics (www.acmg.net)
 - Site has lots of resources, including practice resources, development of guidelines for testing
 - Contributor to the ABIM's Choosing Wisely Campaign

“To find health should be the object of the doctor. Anyone can find disease.”

~A.T. Still MD, DO Philosophy of Osteopathy