Personal Genomic Testing and Precision Medicine

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Goals

- Define precision medicine
  - ≠ genomic medicine
  - Types & uses of genetic testing /genomic sequencing
  - Understand the benefits and limitations of precision medicine
- Discuss direct-to-consumer genetic testing
  - Help to inform decisions about its use
- Discuss some concerns about genetic info
  - Privacy, security, accuracy
- Question whether, when, & how info is empowering
‘Genetics’ refers to the study of genes and their roles in inheritance—in other words, the way that certain traits or conditions are passed down from one generation to another.

- Genetics involves scientific studies of genes and their effects.
- Genes (physical units of heredity) carry the instructions for making proteins, which direct the activities of cells and functions of the body.

‘Genomics’ is a more recent term that describes the study of all of a person's genes (the genome), including interactions of those genes with each other and with the person's environment.

- G x E — gene x environment interaction
- Uterine, physical, chemical, social, behavioral environments
“We used to think that our fate was in our stars, but now we know that, in large measure, our fate is in our genes.”
James Watson, 1989, quoted by Leon Jaroff

“I think it won’t be too many years before parents will be able to go home from the hospital with their newborn babies with a genetic map in their hands that will tell them, here’s what your child’s future will likely be like.”
President Bill Clinton, 1996
The New York Times

STUDENT OPINION

How Much Has Your ZIP Code Determined Your Opportunities?

To what extent has where you were born, and where you’ve grown up, affected who you are?
Human Genome Project (1990 – 2003), $2.7B

Reducing the Cost of Human Genome Sequencing

Moore’s Law

National Human Genome Research Institute

$1,000 GENOME
THE PRECISION MEDICINE INITIATIVE
THE PRECISION MEDICINE INITIATIVE
“Doctors have always recognized that every patient is unique, and doctors have always tried to tailor their treatments as best they can to individuals. You can match a blood transfusion to a blood type — that was an important discovery. What if matching a cancer cure to our genetic code was just as easy, just as standard? What if figuring out the right dose of medicine was as simple as taking our temperature?”

- President Obama, January 30, 2015

So what is Precision Medicine?

It's health care tailored to you.

In his 2015 State of the Union address, President Obama announced that he's launching the Precision Medicine Initiative — a bold new research effort to revolutionize how we improve health and treat disease.

Until now, most medical treatments have been designed for the “average patient.” As a result of this “one-size-fits-all” approach, treatments can be very successful for some patients but
Personalized, precision medicine

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— President Obama, January 30, 2015
Evidence-based
- Epidemiologically based
- Population-based
  - Association, not causation

Personalized
- Precision
- Stratified

conscientious, explicit, and judicious use of current best evidence in making decisions about the care of individual patients. The practice of evidence based medicine means integrating individual clinical expertise with the best available external clinical evidence from systematic research.

(BMJ 1996;312:71-72)

‘Personalized medicine’ refers to the tailoring of medical treatment to the individual characteristics of each patient. It does not literally mean the creation of drugs or medical devices that are unique to a patient, but rather the ability to classify individuals into subpopulations that differ in their susceptibility to a particular disease, in the biology and/or prognosis of those diseases they may develop, or in their response to a specific treatment. Preventive or therapeutic interventions can then be concentrated on those who will benefit, sparing expense and side effects for those who will not.

(President’s Council of Advisors on Science and Technology)
US experience with stratification

“... the Human Genome Project will bring to the forefront of human consciousness awareness of the range of variability not only among individuals but among groups. To acknowledge these differences, while insisting on their irrelevance for individual dignity and equality of right, is a challenge we shall have to face.”

Harold Edgar, 1992
Herman Shaw
Tuskegee Study Survivor

Do you have bad blood?
Free blood tests
Free treatment

The New York Times
Syphilis Victims in U.S. Study Went Untreated for 40 Years

WASHINGTON, July 28—For 40 years the United States Public Health Service has conducted a study in which it is claimed that the morality of the study, also say that it is too late to treat the

have serious doubts about the morality of the study, also say that it is too late to treat the

Herman Shaw
Tuskegee Study Survivor
Give your Supermom even more power over her health.

$50 OFF when you buy ONE Health + Ancestry kit

$150 OFF when you buy TWO Health + Ancestry kits

Shop now

Offer ends May 9
Three steps. It's simple.

All from home. No blood. No needles. Just a small saliva sample.

1. **Order**
   Choose from our Health + Ancestry or Ancestry + Traits services. Your saliva collection kit is the same for both services and typically arrives within 3 to 5 days. Express shipping is available.

2. **Spit**
   Follow kit instructions to spit in the tube provided – all from home. Register your saliva collection tube using the barcode so we know it belongs to you, and mail it back to our lab in the pre-paid package.

3. **Discover**
   In approximately 3-5 weeks, we will send you an email to let you know your reports are ready in your online account. Log in and start discovering what your DNA says about you.
Ancestry + Traits Service

The best DNA kit with the most comprehensive ancestry breakdown and 30+ trait reports.

- 80+ personalized reports
- Ancestry percentages (to the 0.1%)
- 2000+ geographic regions
- Health upgrade available

Add to cart $99
Health + Ancestry Service

Get personalized genetic insights and tools that can help make it easier for you to take action on your health.

- 150+ personalized reports
- Includes Ancestry + Traits Service
- Includes FDA-authorized reports
- FSA/HSA eligibility*

Add to cart $199 $149

There is a lot to consider with genetic testing. We encourage you to review relevant information about Carrier Status* and Genetic Health Risk* reports.

Important test info
2010: concerns about info accuracy

4 DTC companies, 5 “customers,” 15 diseases
- 4/5 results conflicted with actual disease
- Male received 1 risk of prostate CA

The U.S. Food and Drug Administration today allowed marketing of 23andMe Personal Genome Service Genetic Health Risk (GHR) tests for 10 diseases or conditions. These are the first direct-to-consumer (DTC) tests authorized by the FDA that provide information on an individual’s genetic predisposition to certain medical diseases or conditions, which may help to make decisions about lifestyle choices or to inform discussions with a health care professional.
Your health starts in your genes.

Invitae (“in-VEE-tay”) makes it easy to access your unique genetic information, so you can take control of your health.

Find out more  Register a kit

Build your blueprint for better health.
Have a health question? We have a test for you.

- **Pregnancy**
  Get helpful information to guide important health decisions before and during pregnancy.

- **Staying healthy**
  Get what you need to develop a personalized health plan for heart disease, cancer, and more.

- **Cancer**
  Understand your genes so you and your doctor can find the most effective treatments.

- **Genetic illness**
  Find a diagnosis or understand if a condition is hereditary so you can zero in on the right treatment.

Are you a healthcare provider? Visit our [provider site](#) for more information.

Considering genetic testing for the first time? [Take a short quiz](#) to find the right test for you.
Types or uses of genetic/genomic testing

- Diagnostic testing (single-gene, panel, or whole exome/genome sequencing)
  - Carrier screening (inform reproductive decisions)
  - Resolve a “diagnostic odyssey” or confirm diagnosis
- Predictive testing for susceptibility to disease
  - Variants in single genes (BRCA1/2, APOE, HTT (for HD))
  - Polygenic risk scores (PRS) where multiple genes each as a small effect
  - Clinically “actionable” or informs decisions
- Pharmacogenomic testing
- Tumor testing (to guide treatment or research)
Privacy – right to control access to / use of personal information about oneself

Why care about privacy
- Protect from harm
  - Reputation, relationship, economic interests
  - Effects on employability, insurability, marriageability
  - Protect family relationships, friendships
- Protect oneself from the “crippling public gaze”
  - Allow crafting of life plans & development of values, ideals, personality
  - Allow flourishing as an individual
    - Take risks, have a child, be an artist

Sources of risk to privacy
- Self-disclosure
- Privacy breach due to another’s
  - Carelessness
  - Malice
  - Non-malicious, but unwelcome seeking of info (insurers)
GINA does not apply to life, long-term care, disability, or auto insurance

Note the subpoena & court order exception
Security matters.

We have measures in place to prevent unauthorized access and ensure appropriate use of your genetic information.

- Separate databases keep your genetic information decoupled from your personally identifiable information, protecting your identity
- External firewalls restrict unauthorized connections to our databases
- All connections to our website are encrypted using SSL (Secure Sockets Layer) technology

Data is not anonymized, but info is “decoupled” from identifying info

Encryption & firewalls attempt to prevent hacking and unauthorized use
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<th>Records</th>
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Genetic information exposed in privacy breach at Massachusetts General Hospital

Genealogy Databases Enable Naming Of Anonymous DNA Donors

CAMBRIDGE, MASSACHUSETTS—One afternoon in March last year, Yaniv Erlich sat down at his computer to do an experiment. Before
#2—Who’s your (fore)daddy?
- Parentage
- Ancestry
- Who participated in studies → stratification and risk estimates
- Value of predictive genetic testing & precision medicine

#3—The “missing link” (for precision medicine) is behavioral, not genetic
- Genes x Environment
- Individualizing social problems
- What makes genetic info “actionable” and valuable?
- The right-to-know & not-to-know

#4—Genetic Enhancement & the Myth ofMerit
Problems of Playing God & Parenting