



eHEALTH INITIATIVE
Real Solutions. Better Health.

Policy Perspectives: Bioinformatics, Genomics & the Future of Health Care

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Agenda

2:00 – 2:05 PM Welcome and Introductions

- Moderator: Ticia Gerber, Senior Advisor, *eHealth Initiative*

2:05 – 2:20 PM Legislative Update and Q & A

- Elizabeth Farrar, JD, Health Counsel, *Office of the Honorable Diana DeGette (D-CO)*

2:20 – 2:35 PM Genomics and Policy Today

- Grant M. Wood, Senior Information Technology Strategist, Clinical Genetics Institute, *Intermountain Healthcare*; Chair, *HL7 Clinical Genomics Workgroup*

2:35-2:50 PM Towards Precision Medicine: Policies Pending

- Jessica Tenenbaum, PhD, Associate Director for Bioinformatics, *Duke Translational Medicine Institute*; Chair, *AMIA Genomics and Translational Bioinformatics Working Group*

2:50 – 3:00 PM Q&A and Conclusion (Moderator: Ticia Gerber)





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Legislative Update: 21st Century Cures

Elizabeth Farrar, JD

**Office of the Honorable Diana
DeGette (D-CO)**

Q&A

Submit a question or a comment through the chat box.





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Genomics and Policy Today

Grant M. Wood
Intermountain Clinical Genetics
Institute



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Toward Precision Medicine: Policies Pending

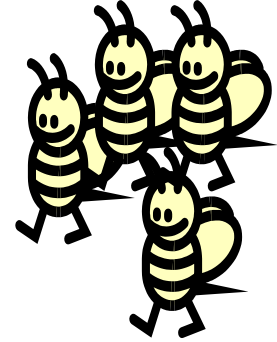
Jessica Tenenbaum, PhD
Duke Translational Medicine
Institute

Talk Outline

- N of 7 billion: Precision medicine, the learning healthcare system, and patient consent
- N of 1: Direct to consumer genetic testing, GINA, and the FDA



P* medicine



- Personalized medicine
- Precision medicine
- P4: predictive, preventive, personalized, and participatory (Hood)
- Aka stratified, genomic, individualized...
- Each a different emphasis, but fundamentally: use more data to deliver
 - the right treatment
 - for the right patient
 - at the right time



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Need Larger N

- High dimensional data → curse of dimensionality, multiple hypothesis testing
- With increased numbers of parameters, need larger sample numbers to detect signal
- Larger sample, larger cost
- Various large consortia



Large-scale Biorepositories

- Millions Veterans Project
- UK Biobank, Genomes England, Genes and Health East London
- The Framingham Study
- US National Children's Study (on hold indefinitely)
- National Biobank of Korea (NBK)
- China Kadoorie Biobank (CKB)
- Regeneron/Geisinger collaboration
- Kaiser/UCSF Research Program on Genes, Environment and Health
- eMERGE Consortium sites
- The MURDOCK Study
 - Tenenbaum et al. *Amer J Trans Res.* 2012



The learning health care system as a living laboratory

- Each patient encounter becomes a data point
- Enables data mining, hypothesis generation
- Bi-directional translation
 - Research informs clinical care, care informs research
- What kind of consent is needed?
 - Informed consent, as in traditional research studies? \$\$\$
 - Option on permission to treat form
 - Opt out ok? Ethical issues.
 - Regional differences

Every Patient a
Subject
JJ Kulynych, H
Greely
Slate.com
Dec 30, 2014



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Direct to Consumer (DTC) Genetic Testing

Personal Genome Service™

Get to know your DNA. All it takes is a little bit of spit.

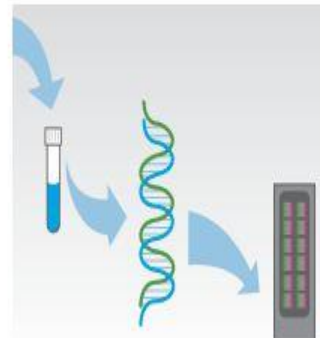
Here's what you do:



1. Order a kit from our [online store](#).



2. [Register your kit](#), spit into the tube, and send it to the lab.



3. Our CLIA-certified lab analyzes your DNA in 6-8 weeks.



4. [Log in](#) and start exploring your genome.

<http://www.singularityweblog.com/23andme-dna-test-review-its-right-for-me-but-is-it-right-for-you/>

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DTC Genetic Testing

- 2013- 23andMe was last one standing in US
- FDA ordered them to stop advertising and offering health-related information (ancestry ok)
- Bringing back one test at a time with FDA approval
- Ethical, Legal, Social Issues (ELSI)
 - Regulated by government?
 - Are consumers ready?
 - Are providers ready?



Case

- 35 years old female
- Heterozygous prothrombin gene mutation (rs1799963) aka factor 2
 - Present in ~2% of population
- Pregnant with twins
- Mutation validated through doctor- part of medical record
- Recommendation: anti-coagulant throughout pregnancy



Other factors for consideration

- No personal or family history
- Infertility
- “Advanced maternal age”
- Economic status
- Professional expertise
- Fear of needles





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23andMe cont.

- Patient applied for life insurance
- Annual rate >2x what it would have been without the prothrombin mutation in medical record
- GINA- Genetic Information Nondiscrimination Act
 - covers employment and health insurance, NOT life, disability, or long term care insurance
- Is that wrong?



Issues and challenges arising from bioinformatics capabilities

- Data governance, provenance
 - Funders, publishers demand reproducible research
- Data sharing
 - Reproducible research, maximum bang for buck
- Privacy
 - Genomic data can't be truly de-identified
- Training and education
 - More quantitative training, more genetic counselors



Increased need for...

- Data standards development and adoption
- Probability, uncertainty, and numeracy
- Decision support, clinical actionability
- Expectation setting



Q&A

Submit a question or a comment through the chat box.





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2015 Annual Conference & Member Meeting

Interoperability: Building Consensus Through the 2020 Roadmap

February 3-5 | Washington D.C.



eHealth Initiative's upcoming **2015 Annual Conference and Government Affairs Retreat** will focus on creating interoperable systems for new care delivery models, with a strong emphasis on supporting transitions of care.

Don't Miss These Exciting Discussion Topics:

Congress and the Administration: Reports from the Front

Getting to Yes: Solutions for Getting Consensus on Standards

Patient Matching: Why You Should Care

Debate: Methods for Managing Privacy Preferences Across the Ecosystem

Working with CEOs on Data Security

Catering to the Consumer

Wearables: Changing the Way We Think About Our Healthcare

Telemedicine: The Next Frontier

Debate on Consolidation in the Health IT Market: Is Bigger Better?

Case Studies Using Analytics in Public Health: Syndromic Surveillance

Register NOW by going to
www.ehdc.org and click on "Events."



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