Emerging Regulatory Challenges in Personal Genomics

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Direct-to-Consumer Genetic Testing:
A Cross-Academies Workshop
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Traditional paradigm towards genetic testing

- Genetic tests justified for “at risk” individuals for “actionable” conditions with available options
- Oversight by a trained healthcare professional
- Requirement for pre- and post-test genetic counseling
- High standards for quality assurance, validity and reliability
- Mechanisms for ensuring confidentiality and privacy
- Genetic information is special

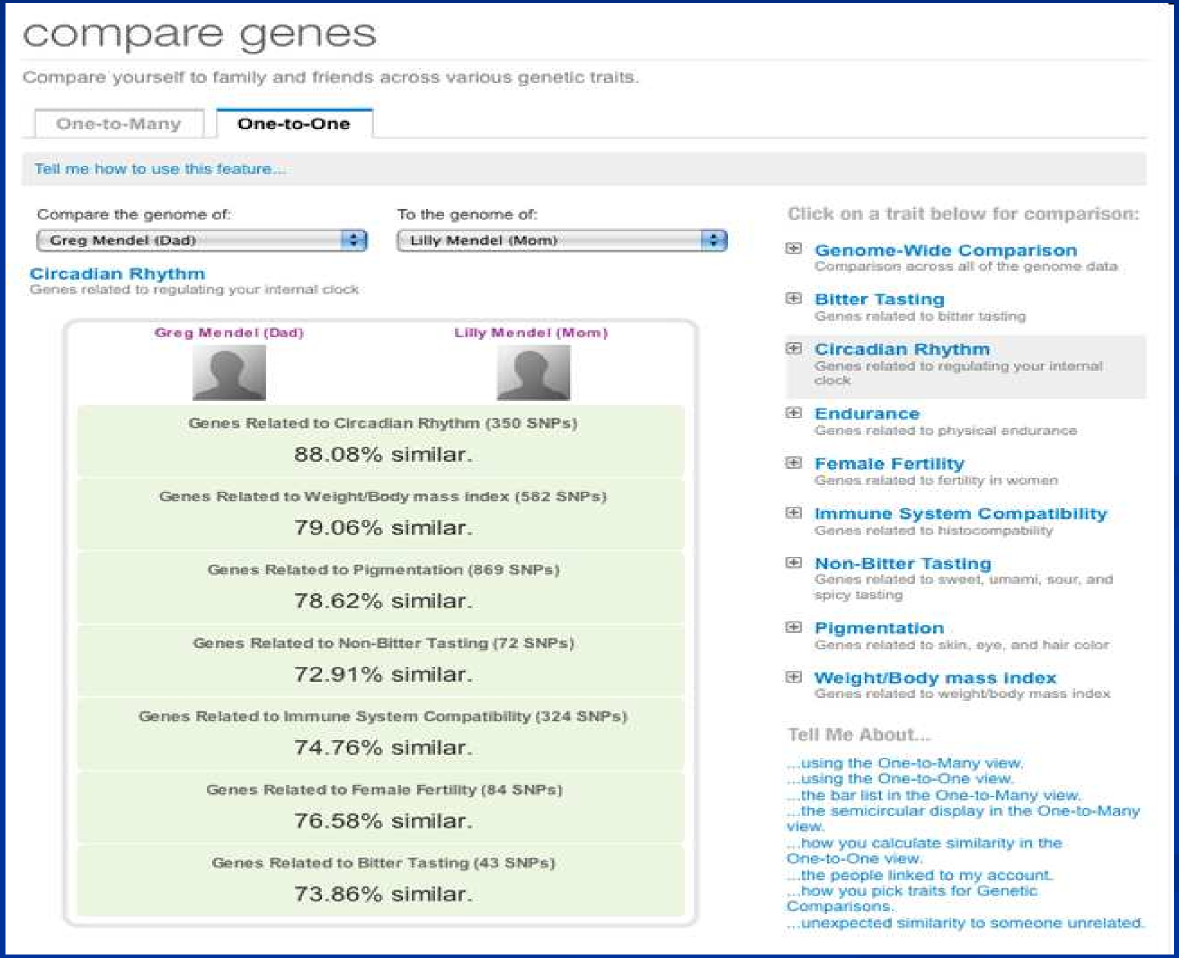
Shifting landscape

- Individual “right to information” without being “at risk”
- Diminished role of the health provider and broader category of genetic experts
- Proliferation of genetic tests for “non-actionable” traits and conditions
- Medical genetic testing is only part of the picture
- Lack of a “bright line” distinguishing categories of testing
- Genetics research as a catalyst for public advocacy and activism
- Corporate guardians of personal genetic information
- Genetic information remains “special”

Emerging Social Issues and Regulatory Challenges

- Social networking: privacy and protections
- Company sponsored research: consumer becomes human subject
- Ancestry testing: finding common language
- Behavioral genetics: testing for normal traits

Sharing your genome



compare genes

Compare yourself to family and friends across various genetic traits.

Compare the genome of: Greg Mendel (Dad)

To the genome of: Lilly Mendel (Mom)

Circadian Rhythm

Genes related to regulating your internal clock

Genes Related to Circadian Rhythm (350 SNPs)
88.08% similar.

Genes Related to Weight/Body mass index (582 SNPs)
79.06% similar.

Genes Related to Pigmentation (869 SNPs)
78.62% similar.

Genes Related to Non-Bitter Tasting (72 SNPs)
72.91% similar.

Genes Related to Immune System Compatibility (324 SNPs)
74.76% similar.

Genes Related to Female Fertility (84 SNPs)
76.58% similar.

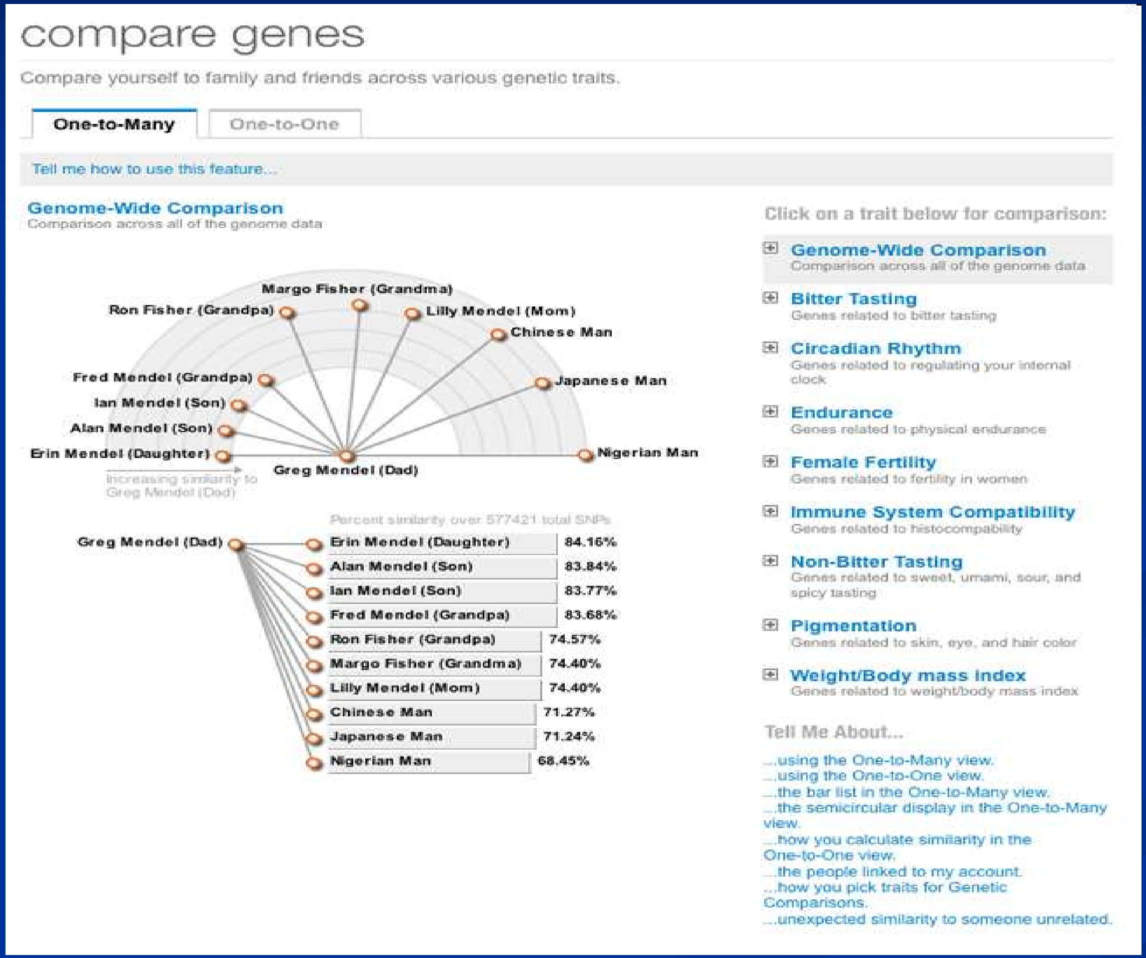
Genes Related to Bitter Tasting (43 SNPs)
73.86% similar.

Click on a trait below for comparison:

- ⊞ **Genome-Wide Comparison** Comparison across all of the genome data
- ⊞ **Bitter Tasting** Genes related to bitter tasting
- ⊞ **Circadian Rhythm** Genes related to regulating your internal clock
- ⊞ **Endurance** Genes related to physical endurance
- ⊞ **Female Fertility** Genes related to fertility in women
- ⊞ **Immune System Compatibility** Genes related to histocompatibility
- ⊞ **Non-Bitter Tasting** Genes related to sweet, umami, sour, and spicy tasting
- ⊞ **Pigmentation** Genes related to skin, eye, and hair color
- ⊞ **Weight/Body mass index** Genes related to weight/body mass index

Tell Me About...

...using the One-to-Many view.
...using the One-to-One view.
...the bar list in the One-to-Many view.
...the semicircular display in the One-to-Many view.
...how you calculate similarity in the One-to-One view.
...the people linked to my account.
...how you pick traits for Genetic Comparisons.
...unexpected similarity to someone unrelated.



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Tell me how to use this feature...

Genome-Wide Comparison
Comparison across all of the genome data

Percent similarity over 577421 total SNPs

Greg Mendel (Dad)

Erin Mendel (Daughter)	84.16%
Alan Mendel (Son)	83.84%
Ian Mendel (Son)	83.77%
Fred Mendel (Grandpa)	83.68%
Ron Fisher (Grandpa)	74.57%
Margo Fisher (Grandma)	74.40%
Lilly Mendel (Mom)	74.40%
Chinese Man	71.27%
Japanese Man	71.24%
Nigerian Man	68.45%

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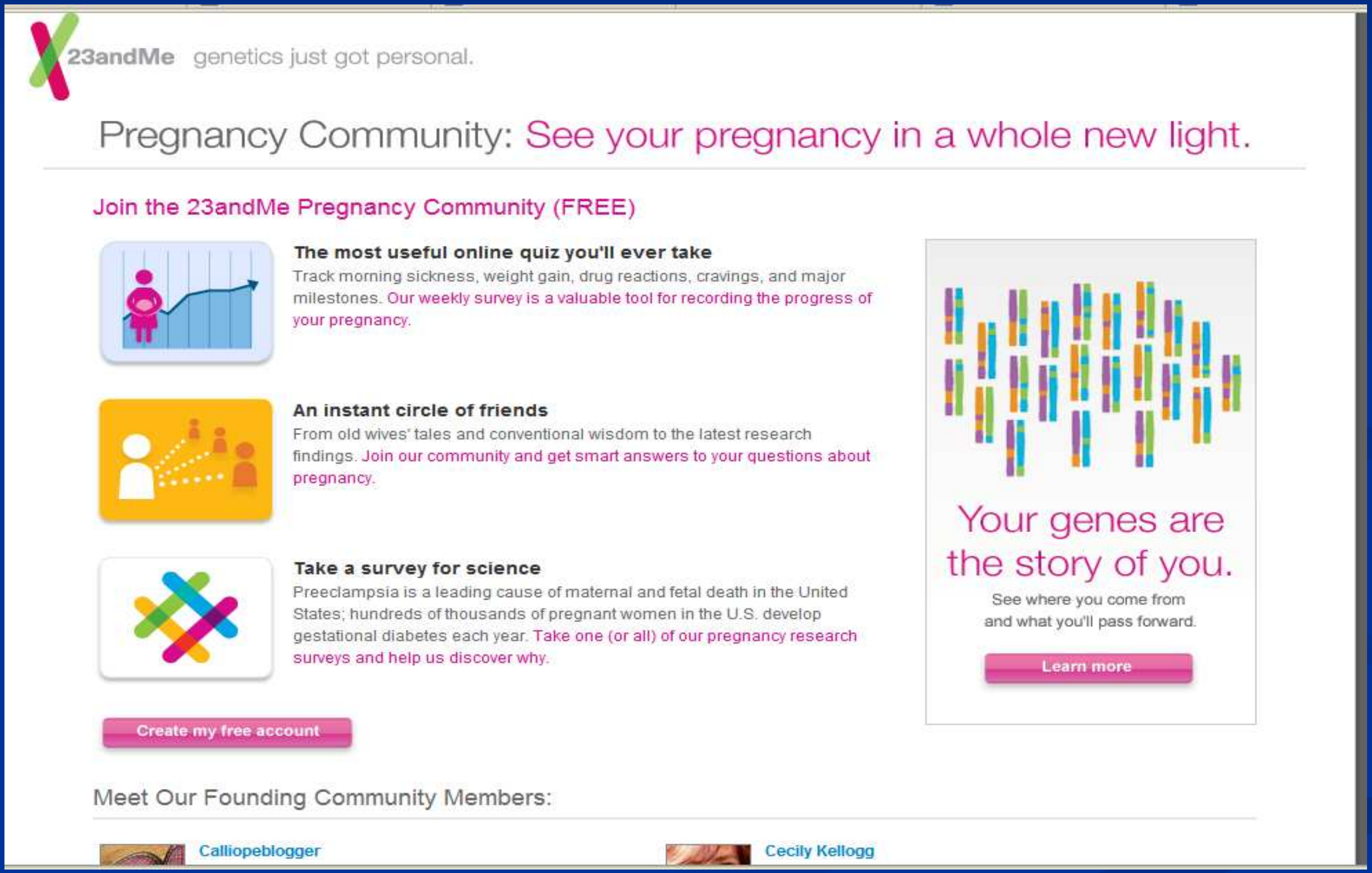
“An instant circle of friends”



Pregnancy Community: See your pregnancy in a whole new light.

Join the 23andMe Pregnancy Community (FREE)

The most useful online quiz you'll ever take

Track morning sickness, weight gain, drug reactions, cravings, and major milestones. Our weekly survey is a valuable tool for recording the progress of your pregnancy.

An instant circle of friends

From old wives' tales and conventional wisdom to the latest research findings. Join our community and get smart answers to your questions about pregnancy.

Take a survey for science

Preeclampsia is a leading cause of maternal and fetal death in the United States; hundreds of thousands of pregnant women in the U.S. develop gestational diabetes each year. Take one (or all) of our pregnancy research surveys and help us discover why.

Your genes are the story of you.

See where you come from and what you'll pass forward.

Meet Our Founding Community Members:

Calliopeblogger

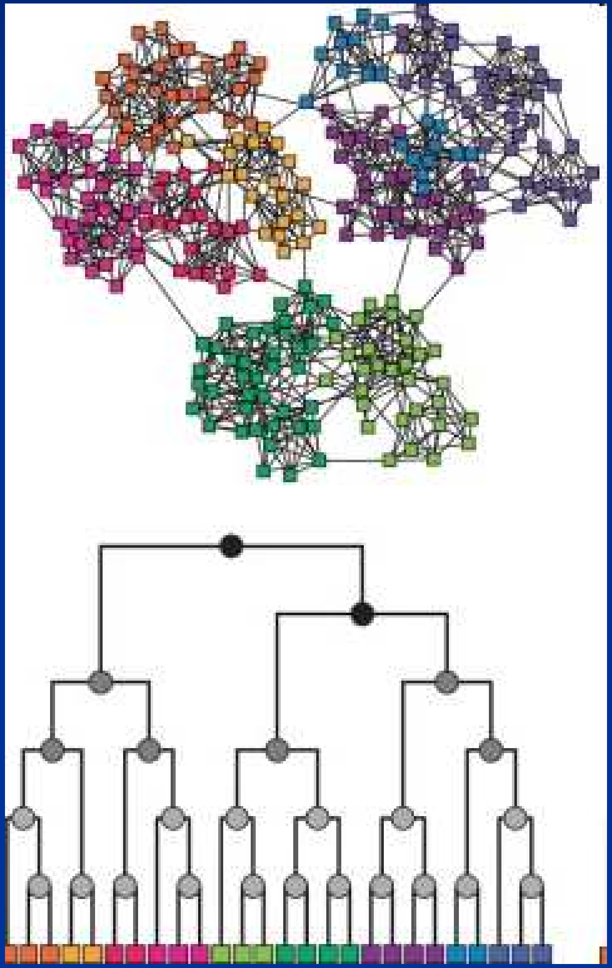
Cecily Kellogg

Biosociality

“DTC genomic companies emerge as new regimes of biosociality where PGI reconstitutes human identity and yet builds on existing networks of social relationships. Social networking around PGI has the potential to create biosocial groups that ultimately serve to overcome the ‘nature/culture split’”

American Journal of Bioethics. 2009: 38

Social Networks through the Genome



- Commercial infrastructure for networking
- Personal genomic information as a medium for connections
- Communities coalescing around population based genomic research

“Patients helping patients live better everyday”

patientslikeme™ Patients helping patients live better every day.

Share Your Experience »

See how easy it is...

Find Patients Like You »

Try it out...

Learn From Others »

Discover the power of many...

Already a member? Log in

Our Current Communities

Neurological Conditions
- MS (Multiple Sclerosis)
- Parkinson's Disease
- ALS (Amyotrophic Lateral Sclerosis) / MND (Motor Neuron Disease)
- PLS (Primary Lateral Sclerosis)
- PMA (Progressive Muscular Atrophy)
- PSP (Progressive Supranuclear Palsy)
- CBD (Corticobasal Degeneration)
- MSA (Multiple System Atrophy)
- Devic's Neuromyelitis Optica

Neuroendocrine Conditions
- Fibromyalgia **(New!)**

Mood Conditions
- Depression
- Anxiety
- Bipolar
- OCD (Obsessive-Compulsive Disorder)
- PTSD (Post-Traumatic Stress Disorder)

Immune Conditions
- HIV/AIDS

If we have a community for your condition, join now! Don't see your condition? Request it here.

Highlights

Fibromyalgia Community
Introducing our new community for Fibromyalgia patients like you. **Learn more...**

Lithium & ALS Study
See how ALS patients taking lithium are doing in real-time. **Learn more...**

Clinical Trial Awareness
Two of our clinical trial awareness partners are seeking patients for clinical trials. **Find out more...**

"Most informative site! truly innovative"
— A PatientsLikeMe patient Read more...

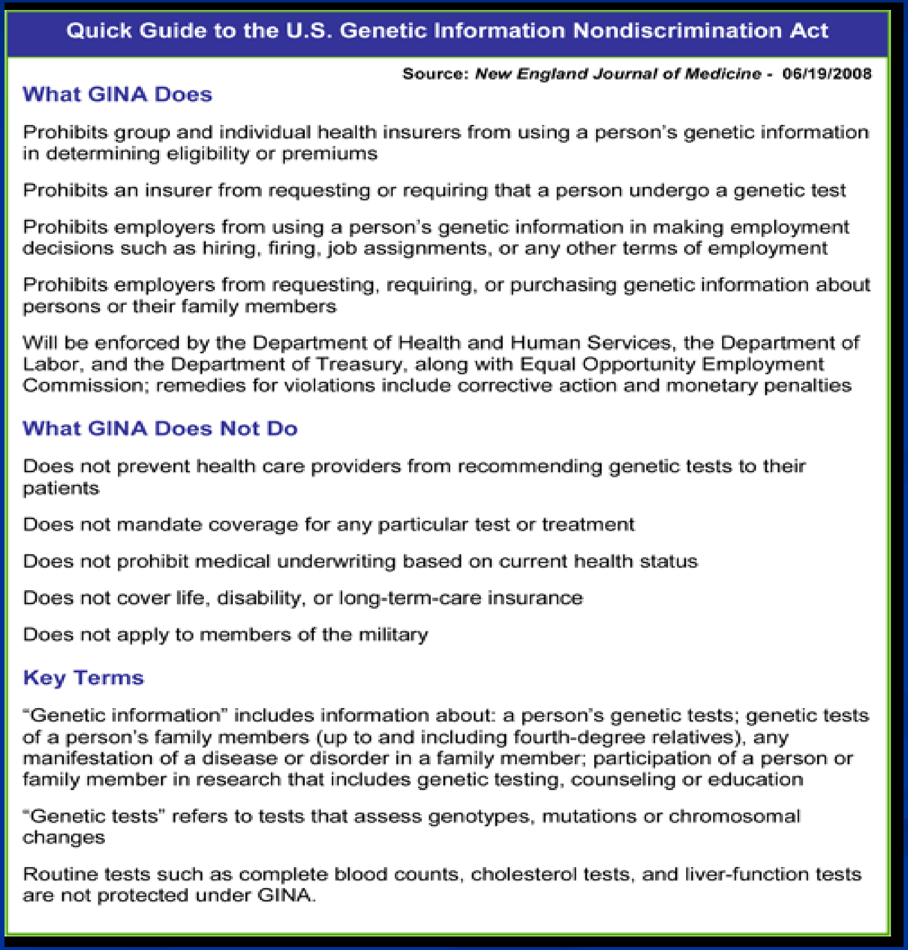
Openness Philosophy

“the Internet can democratize patient data and accelerate research like never before. Furthermore, we believe data belongs to you the patient to share with other patients, caregivers, physicians, researchers, pharmaceutical and medical device companies, and anyone else that can help make patients’ lives better.”

PatientsLikeMe

http://www.patientslikeme.com/about/openness

GINA Protections



Quick Guide to the U.S. Genetic Information Nondiscrimination Act

Source: *New England Journal of Medicine* - 06/19/2008

What GINA Does

Prohibits group and individual health insurers from using a person's genetic information in determining eligibility or premiums

Prohibits an insurer from requesting or requiring that a person undergo a genetic test

Prohibits employers from using a person's genetic information in making employment decisions such as hiring, firing, job assignments, or any other terms of employment

Prohibits employers from requesting, requiring, or purchasing genetic information about persons or their family members

Will be enforced by the Department of Health and Human Services, the Department of Labor, and the Department of Treasury, along with Equal Opportunity Employment Commission; remedies for violations include corrective action and monetary penalties

What GINA Does Not Do

Does not prevent health care providers from recommending genetic tests to their patients

Does not mandate coverage for any particular test or treatment

Does not prohibit medical underwriting based on current health status

Does not cover life, disability, or long-term-care insurance

Does not apply to members of the military

Key Terms

"Genetic information" includes information about: a person's genetic tests; genetic tests of a person's family members (up to and including fourth-degree relatives), any manifestation of a disease or disorder in a family member; participation of a person or family member in research that includes genetic testing, counseling or education

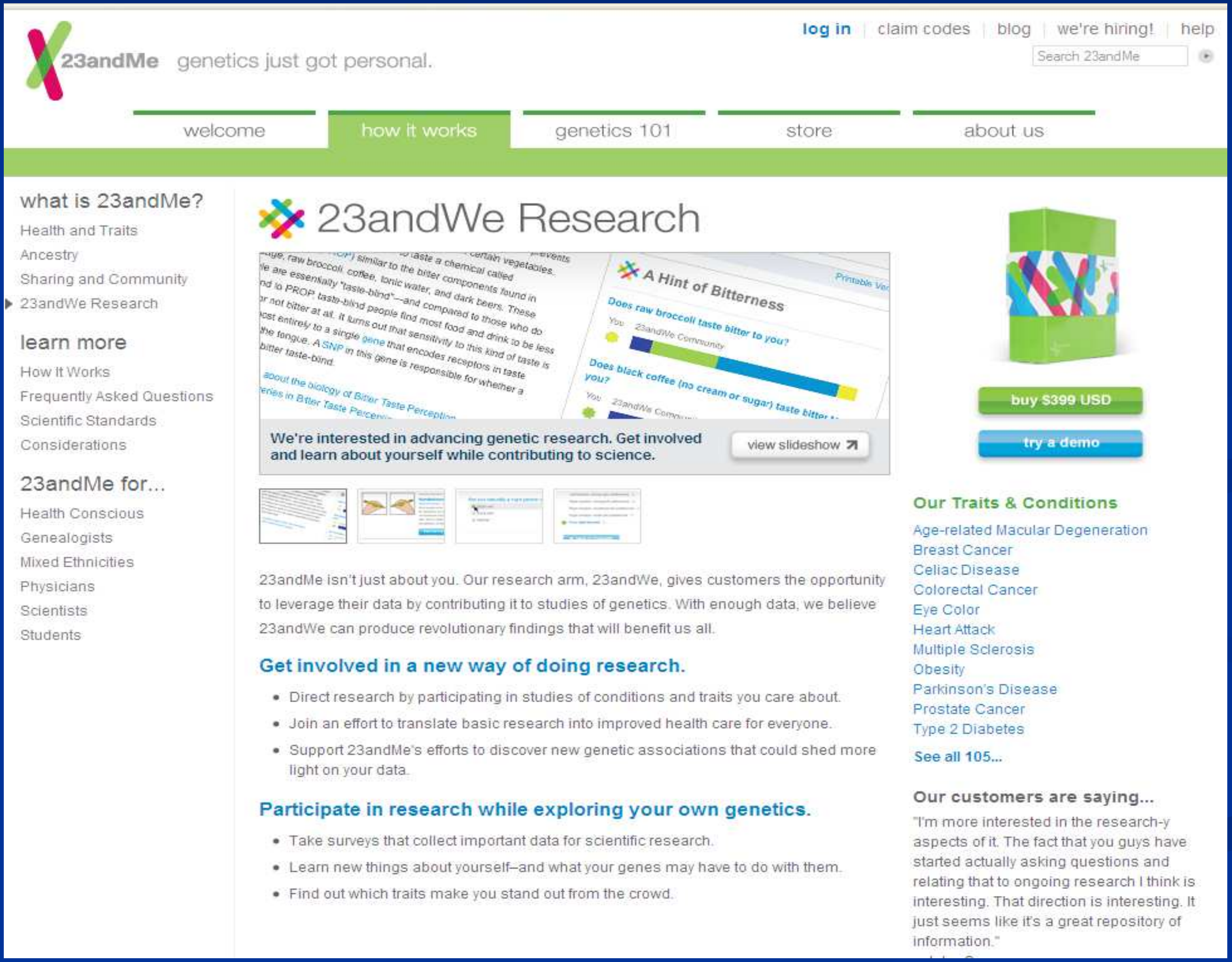
"Genetic tests" refers to tests that assess genotypes, mutations or chromosomal changes

Routine tests such as complete blood counts, cholesterol tests, and liver-function tests are not protected under GINA.

Exceptions to GINA

- Obtaining information during the Family and Medical Leave Act (FMLA) certification process;
- Obtaining genetic information as part of an employer-sponsored voluntary wellness program;
- The acquisition of genetic information through publicly-available information;
- Conducting genetic monitoring of the biological effects of toxic substances in the workplace;
- Requiring employees to provide genetic information as a quality control marker, but only for those employers engaged in conducting genetic testing for law enforcement purposes; and
- Inadvertently obtaining genetic information.

DTC PGx Research



what is 23andMe?

- Health and Traits
- Ancestry
- Sharing and Community
- 23andWe Research

learn more

- How It Works
- Frequently Asked Questions
- Scientific Standards
- Considerations

23andMe for...

- Health Conscious
- Genealogists
- Mixed Ethnicities
- Physicians
- Scientists
- Students

23andWe Research

We're interested in advancing genetic research. Get involved and learn about yourself while contributing to science.

23andMe isn't just about you. Our research arm, 23andWe, gives customers the opportunity to leverage their data by contributing it to studies of genetics. With enough data, we believe 23andWe can produce revolutionary findings that will benefit us all.

Get involved in a new way of doing research.

- Direct research by participating in studies of conditions and traits you care about.
- Join an effort to translate basic research into improved health care for everyone.
- Support 23andMe's efforts to discover new genetic associations that could shed more light on your data.

Participate in research while exploring your own genetics.

- Take surveys that collect important data for scientific research.
- Learn new things about yourself–and what your genes may have to do with them.
- Find out which traits make you stand out from the crowd.

Our Traits & Conditions

- Age-related Macular Degeneration
- Breast Cancer
- Celiac Disease
- Colorectal Cancer
- Eye Color
- Heart Attack
- Multiple Sclerosis
- Obesity
- Parkinson's Disease
- Prostate Cancer
- Type 2 Diabetes

See all 105...

Our customers are saying...

"I'm more interested in the research-y aspects of it. The fact that you guys have started actually asking questions and relating that to ongoing research I think is interesting. That direction is interesting. It just seems like it's a great repository of information."

Scaling Up

“23andWe is a new, more efficient way of doing genetic research. Even though new technologies have made it possible to link genes to diseases, traits and conditions more effectively than ever before, collecting the data for this research can be a costly, time-consuming and logistically difficult process. Progress is hindered by the fact that these studies require both genetic and personal information from thousands – sometimes tens of thousands – of people.”

Collaboration as Empowerment

"23andWe intends to create another kind of collaboration. By connecting consenting 23andMe users who are interested in participating in research - and their genotype data - with the research community, we eliminate the need for inefficient recruitment procedures and distribute the cost of genotyping. We believe connecting people with scientists empowers everyone to accelerate the pace of research."

Research Revolution

The 23andMe Research Revolution

« Go to Research Revolution home

What is genetic research?

The benefits of participating in 23andMe research.

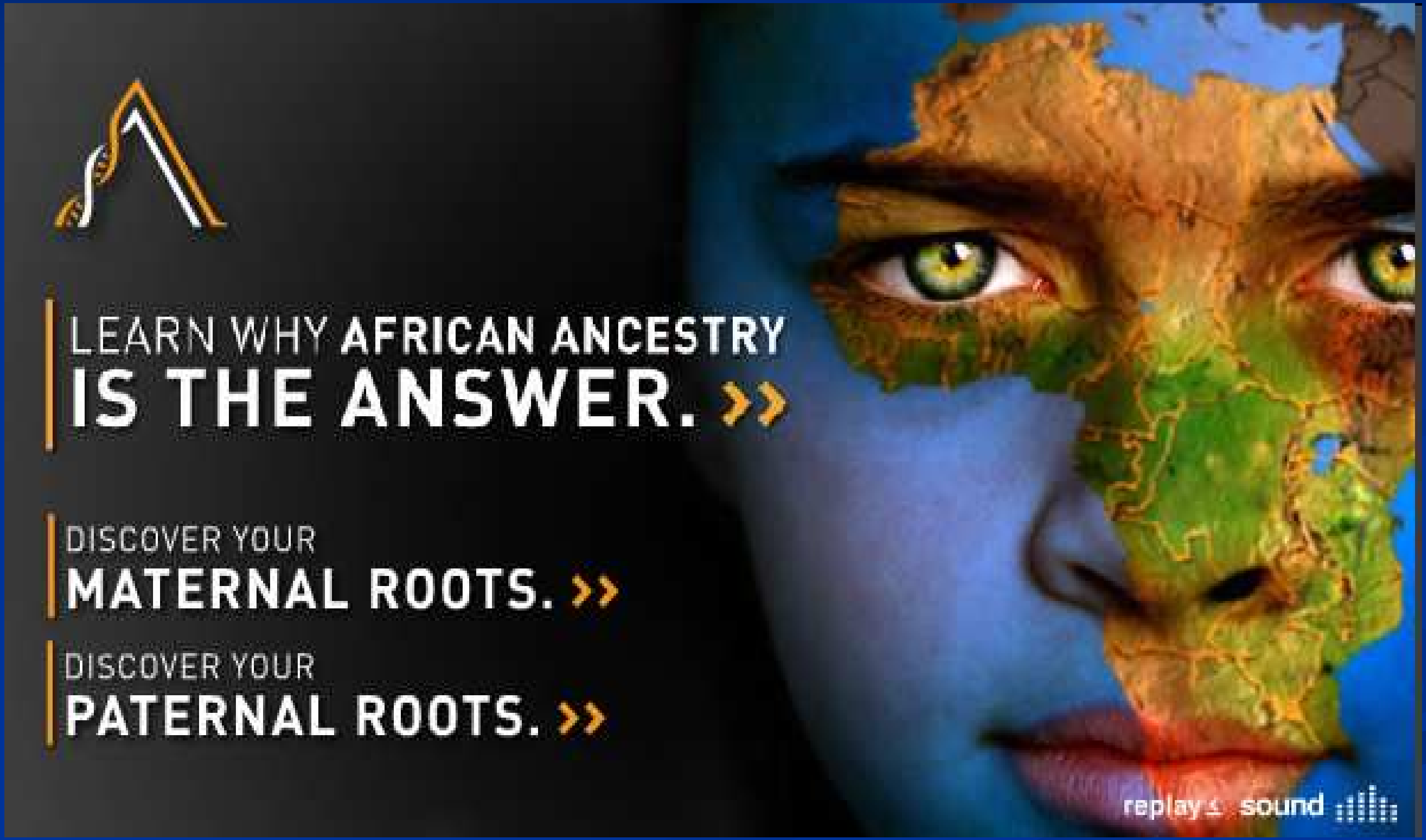
By participating in research, you help 23andMe find new associations between genes and diseases that could lead to better healthcare for everyone. But it also makes your own genetic data more valuable by giving it more meaning.

▶▶ Get involved!

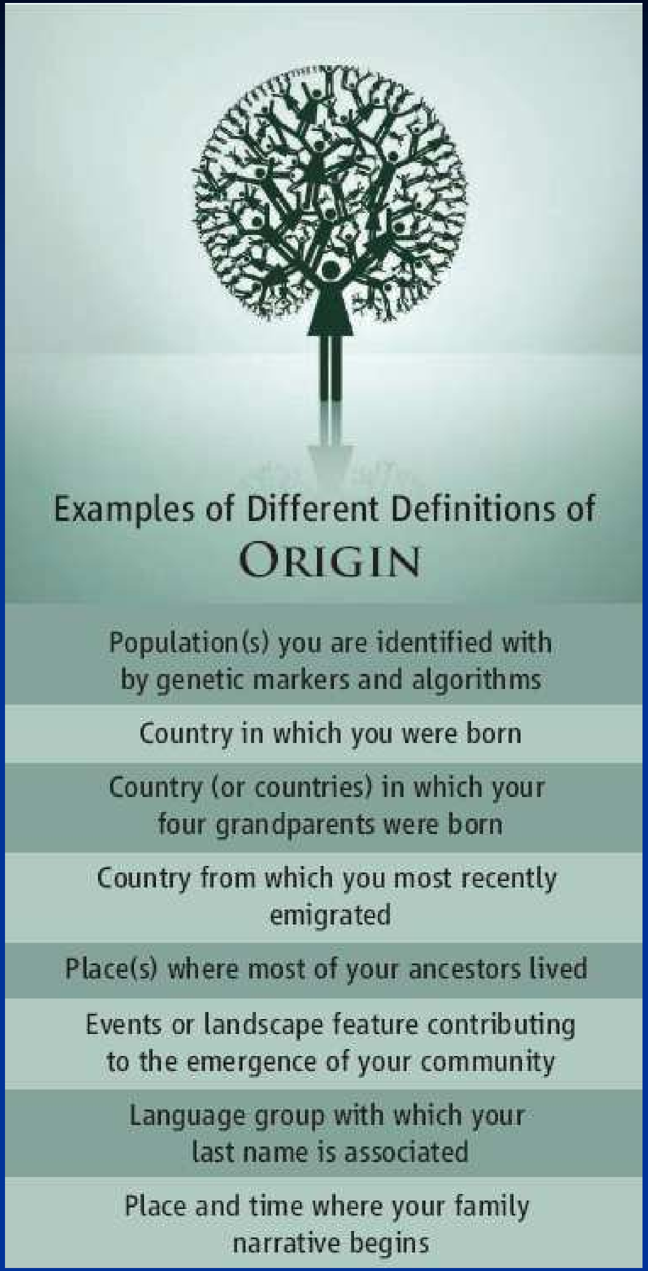
Emerging Questions:
New Paradigm?

- What does “democratization of the genome” mean for the future of large scale population based biomedical research?
- What are the implications of the blurring boundaries between consumer and research participant ?
- How will consumer genetic information travel?
- What are the rights and responsibilities of companies and consumers?

Expectations of Genetic Ancestry



Finding Common Language



Examples of Different Definitions of ORIGIN

- Population(s) you are identified with by genetic markers and algorithms
- Country in which you were born
- Country (or countries) in which your four grandparents were born
- Country from which you most recently emigrated
- Place(s) where most of your ancestors lived
- Events or landscape feature contributing to the emergence of your community
- Language group with which your last name is associated
- Place and time where your family narrative begins

“A first step may be joint creation of a vernacular that characterizes key concepts like probability, association, origin, and ancestry to help minimize variability that exists in how such concepts are understood across fields, communities, and governmental and commercial entities with different vantage points.”

Science 2009. 325:38-9

Testing of Normal Traits

- Ear wax type
- Eye color
- Avoidance of errors
- Breastfeeding and IQ
- Hair color
- Hair Thickness

- Memory
- Food preference
- Pain sensitivity
- Height
- Intelligence

Summer Camp 2.0

"Nowadays, competition in the world is about who has the most talent..We can give Chinese children an effective, scientific plan at an early age."

Chang 2009

Where do we go from here?

- Regulatory protection for the sharing of personal genetic information through social networks
- Need for a genetic vernacular
- Acknowledge the false divide between “recreational” and “medical” genetics
- Focused scrutiny of genetic testing of normal traits
- Guidelines on testing children