

Cooperation or Competition— How do health care and DTC Genetic Testing Coexist?

Patricia A. Ganz, M.D.
UCLA Schools of Public Health & Medicine
Jonsson Comprehensive Cancer Center

My Patient

- Mary is a 48 yo woman who just received 25 pages of genetic test results, listing specific SNPs & risk information
- FH of lymphoma, CLL & colon cancer
- Current health: fibromyalgia, hypothyroid, “chronic viral infection”
- Multiple medical consultants, treated with intravenous immunoglobulin
- Went to university for “DNA workup;” not done; she ordered through 23andMe

Doctor, can you please help...

- What do the DNA results mean?
 - Slightly increased odds of abdominal aortic aneurysm
 - Slightly increased odds of intracranial aneurysm
 - Typical odds of developing bladder cancer
 - Slightly higher odds of developing CLL

Overview

- Who is getting tested now?
- What do DTC genetic testing sites suggest about health care after testing?
- How prepared are physicians to receive info from DTC genetic testing sites?
- Will genetic test results motivate better health behavior?
- When is screening for disease (or risk of disease) acceptable in clinical medicine?
- Recommendations

The Hype and Hope of Genetic Testing.....

What we do

- Navigenics was founded on a simple yet powerful idea. If we can give people insight into their genes, they can take steps toward a healthier life.
- You can use this information to take action and focus on what matters most to your health. And before symptoms appear, you can take these steps now:
 - Make lifestyle changes.
 - Talk to your doctor.
 - Get early screening.
- ***We help you partner with your physician for earlier detection and better treatments, and ultimately, optimum wellness.***

- **Not just data. Insights.**
- We evaluate the latest medical literature to find options especially tailored to you. Your reports are personalized based on your level of risk. We help make the meaning clear.
- Your board-certified Genetic Counselor can help you understand your results. ***And we help you share your results with your doctor effectively.***

- **With this knowledge comes power**
- You have the power to take appropriate action. Focus on the right priorities. Take preventive measures. Be alert for early symptoms.
Schedule more detailed testing.
- You'll learn the real choices and chances open to you. And the better you can see down the road, the more prepared you'll be for life's challenges.

Navigenics testimonial....

- Personalized Prevention
- Taking charge of your health
- “My quick read was “relief” – no surprises – the conditions that I was at higher than normal risk for were ones that occurred in my family. My dad lived to age 87 and my mom to age 84; the conditions that I think I need to pay attention to were the same ones that they were afflicted with. But I have the chance to do something about it.”

Finding the ideal patients to target for genetic testing

The Navigenics services are most effective when targeted to those patients who stand to benefit most from the results. There are four main groups of patients that physicians have found benefit in testing:

1. Patients who require additional motivation to adhere to primary prevention strategies such as for cardiovascular or metabolic diseases
2. Patients seeking enhanced personalized screening programs such as for cancer or ophthalmic disease
3. Patients with difficult-to-diagnose diseases like Celiac disease
4. Patients with limited family histories

Who is using DTC
genetic testing now?

Limited Data Available

- Younger, well-educated, interested in genetics
- Perceived risk of disease may influence use
- Are social networkers and genome testers one in the same?
 - 23andMe focus on social networking
 - Internet savvy most-likely to uptake
 - Plan to communicate with their health care providers

Characteristics of users of online personalized genomic risk assessments: Implications for physician-patient interactions

McBride, et al. *Genetics in Medicine* 2009

- Early adopters interested in genetics
- Perceive they have health habits that can be changed

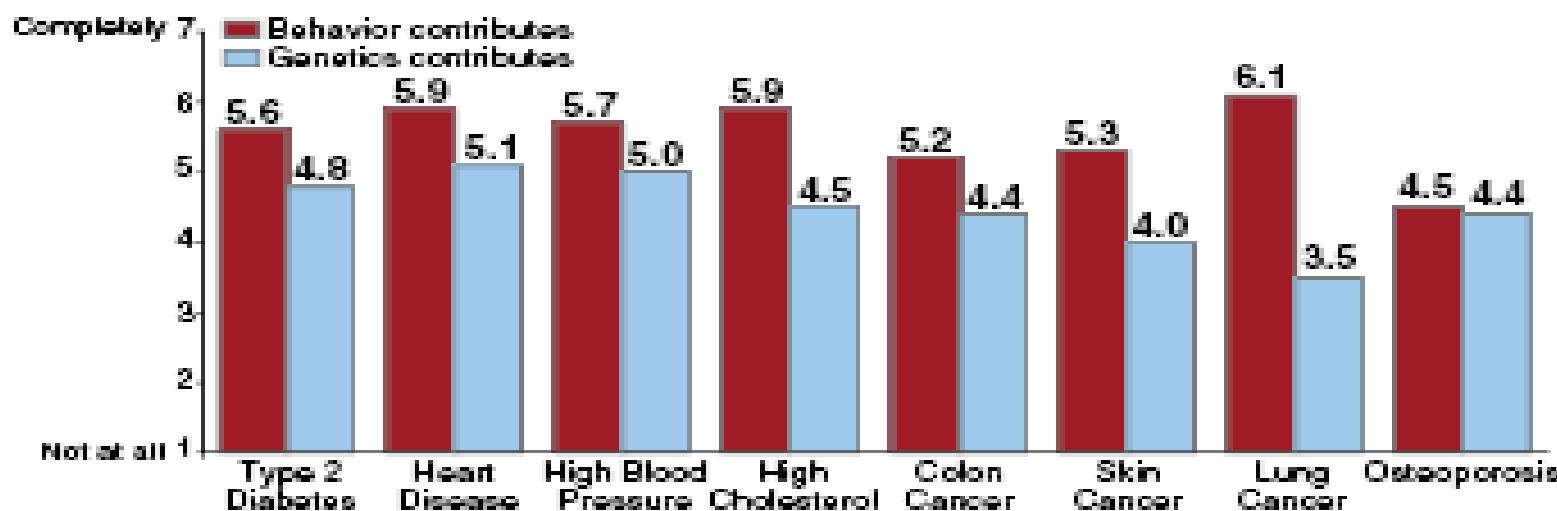


Fig. 2. Participant ratings of factors contributing to eight health conditions on the multiplex genetic susceptibility test.

What do DTC genetic testing sites suggest about health care after testing?

- **Personalized prevention:** We are surrounded by more health advice than ever. With your genetic test results, you can discover which health measures are really right for you. [Learn more. >](#)
- **Working with your doctor:** When you and your doctor examine your DNA profile together with your family history, medical history, and lifestyle, your health picture becomes even more complete. Find out how Navigenics' genetic testing services can help you and your doctor customize your preventive strategies. [Learn more. >](#)
- **Truly personalized health:** Your genetic test results can point you toward optimal health in ways more tailored to you than ever before, letting you make the difference between your DNA and your destiny. [Learn more](#)

23andMe accessed 8/16/09

Service disclaimer

- **Description of What the Services Are and Are Not:** 23andMe Service Is **For Research and Educational Use Only**. We Do Not Provide Medical Advice, And The Services Cannot Be Used For Health Ascertainment or Disease Purposes
- The genetic information provided by 23andMe is for **research and educational use only**. This means two things. **First, the genetic information you receive from 23andMe is based on scientific research, and cannot be relied upon at this point for diagnostic purposes. Genetic discoveries that we report have not, for the most part, been clinically validated, and the technology the laboratory uses the same technology used by the research community has also not yet been validated for clinical utility.** **Second, by your participation in the 23andMe service you contribute your genetic information to our research effort to study various aspects of human genetics in an attempt to better understand the human genome. In addition our service enables you to contribute other personal information towards research as well.**

deCODEme Complete Scan

- Explore your genetic risk factors and keep a **vigilant eye** on your prospects for prolonged health. Learn about your ancestry at the same time. By analyzing an unparalleled one million genetic variants, the Complete Scan is the most accurate, advanced and comprehensive test of its kind.
- The Complete Scan *focuses on medical conditions that can either be improved by altering your lifestyle or have better treatment options if caught early.*
- You will receive updates to your profile as new genetic variants are discovered.

- **Lifestyle changes to prevent or slow down Alzheimer's Disease**

The latest medical research suggests that the best hope for preventing or slowing down Alzheimer's Disease is to **adopt a healthy lifestyle** that includes protecting your head from injury at all times (e.g. by wearing a seatbelt and using a helmet when biking or skiing) and exercising or stimulating your brain regularly (e.g. by taking a class, learning a new language, playing memory games, or doing crosswords).

- Individuals at high risk for Alzheimer's may **benefit from regular screening for early symptoms of the disease** by their primary care provider. **Early, active medical management**, through available treatment options and utilization of programs and support services, can improve quality of life through all stages of the disease for diagnosed individuals and their caregivers.

Key Messages

- Testing will identify risk for diseases that can be prevented
- Changes in behavior, lifestyle, or use of medications will reduce your risk of developing disease
- Your health care provider will be able to help you determine how to use this information!

How prepared are physicians to receive info from DTC genetic testing sites?

MDs Have Limited Knowledge of Genetics

- When tests have known clinical value in clinical medicine, they are ordered e.g. prenatal testing, bleeding disorders
- For known genetic syndromes, e.g. cancer predisposition, physicians have limited skills to interpret test results
- Knowledge/expertise related to results from association studies is even more limited

Genetic Testing for Breast and Ovarian Cancer Susceptibility: Evaluating Direct-to-Consumer Marketing --- Atlanta, Denver, Raleigh-Durham, and Seattle, 2003

- “in all four cities, providers often lacked knowledge to advise patients about inherited BOC and testing. These findings underscore the need for evidence-based recommendations on appropriate use of genetic tests and education of providers and the public to achieve maximum individual and public health benefit from genetic testing.”
- MMWR July 2004

Will genetic test results motivate
better health behavior?

Lung cancer

- **Fact:** Lung cancer is the leading cause of cancer deaths in the United States. It claims more lives each year than colon, prostate, lymphatic and breast cancers combined.
- **Proportion of risk that's in your genes:** 14 percent.
- **What you can do:** Particularly if you're a current or former smoker, knowing from a genetic test whether you have an increased risk of lung cancer can encourage you to make lifestyle changes to improve your health and reduce your risk of developing the disease. Quitting smoking is the most important change you can make. Getting regular exercise and eating fruits and vegetables may also help prevent lung cancer.
- **Did you know?** While smoking cigarettes increases your risk of lung cancer, so may drinking too much alcohol. Women should limit themselves to no more than one drink a day, while men should stop at two drinks

Lung Cancer (cont'd)

- Knowing from a gene test that you are at increased genetic risk can help you and your doctor be alert to the symptoms of lung cancer. Your genetic test results could also spur you to come up with a plan for lowering the risk that is due to your behavior and the environment.
- Protecting yourself from exposure to asbestos, radon gas and secondhand smoke reduces your risk. Regular exercise and a healthy diet full of fruits and vegetables can also help. If you're a smoker, quitting now will also reduce your risk of lung cancer, even if you've smoked for years.

What is the evidence?



Marteau & Lerman, BMJ 322:1056, 2001

What is the evidence re: behavior change after genetic testing?

- Mixed results
- More likely for highly penetrant genes (e.g. *BRCA1/2*, *MEN*, *FAP*)
- Low risk genes from GWAS may have limited impact on behavior
- Unclear if genetic information added to family history improves behavior change

When is screening for disease (or
risk of disease) acceptable in
clinical medicine?

Principles of Screening

- The condition should be an important health problem.
- There should be a treatment for the condition.
- Facilities for diagnosis and treatment should be available.
- There should be a latent stage of the disease.
- There should be a test or examination for the condition.
- The test should be acceptable to the population.
- The natural history of the disease should be adequately understood.
- There should be an agreed policy on who to treat.
- The total cost of finding a case should be economically balanced in relation to medical expenditure as a whole.
- Case-finding should be a continuous process, not just a "once and for all" project.

WHO 1968

Additionally...

- Screening test should be sensitive, specific, and have strong positive/negative predictive value for disease or condition
- Similarly, if a test is negative, it may give a false sense of security that the disease will not occur, which often is not the case

DeCODE genetic disease testing 2008

- Abdominal aortic aneurysm
- Age-related macular degeneration
- Alzheimer's disease
- Asthma
- Arterial fibrillation
- Breast cancer
- Coeliac disease
- Colorectal cancer
- Crohn's disease
- Exfoliation glaucoma
- Heart attack
- Haemochromatosis
- Intracranial aneurysm
- Lactose intolerance
- Lung cancer
- Multiple sclerosis
- Obesity
- Peripheral arterial disease
- Prostate cancer
- Psoriasis
- Thromboembolism

What is the quality of these tests and the setting of testing?

- Many are based on variations in SNPs and association studies
- Some are from GWAS studies, with no application to clinical populations
- Limited family information may lead to uninformative negative tests
- None or limited pre-test genetic counseling

Risks to the Clinical Encounter

- Demand for screening tests which have no proven value
- Physician may seem unsympathetic or lacking in knowledge when presented with output from genetic screening report
- Finding of “low risk” may give a false sense of security, when effective behavioral recommendations are warranted in spite of “negative genetic test”

Strategies/Recommendations

- *This is part of the future:* physicians need to improve their understanding of genetics and be able to discuss the pros/cons/limitations of these tests
- Need scientific guidance on tests that can be linked to effective screening strategies—for the benefit of the public and the medical community

Need More Research...

- On communication of risk information
- On the positive/negative consequences for patients on learning these genetic results
- On whether or not behavior change is enhanced through genetic information
- On the role of physicians and the health care system in managing expectations of patients who pursue testing

Conclusion

“We currently lack the knowledge to define when or how genetic risk information might motivate healthy behavior. Lacking that knowledge, we are unable to define appropriate health care uses, impacts on health care resources of consumer tests or parameters for truthful advertising of direct-to-consumer tests. Identifying the settings in which genomic risk can motivate healthy behavior, and perhaps the individuals most likely to respond to such information, is an important policy concern.”

Henrikson, Bowen, & Burke,
Genomic Medicine 2009