

| Web-based genetics services

- | Elissa Levin, MS, CGC
Director, Genetic Counseling
Navigenics

| Institute of Medicine:
Innovations in Service Delivery
in the Age of Genomics

July 28, 2008



Navigenics

The Current Landscape |

Why People Seek Online Genetic Testing

- | Getting genetic testing can be difficult
 - No genetics service in region
 - Convenience: location, time preferences, time-off, childcare...
- | Provider awareness is not optimal
 - Lack of perceived value in genetic testing
- | More cost-effective
- | Privacy and confidentiality concerns
- | Proactive interest in genetic contribution to disease
 - | Time to adoption for new types of testing in clinical setting
 - | Media attention... increasing awareness about genetics

The Current Landscape

I A growing genetic testing market, but with major differences

1. Types of testing offered

Diagnostic

Predisposition

Recreational



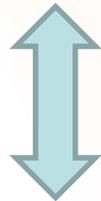
The Current Landscape

I A growing genetic testing market, but with major differences

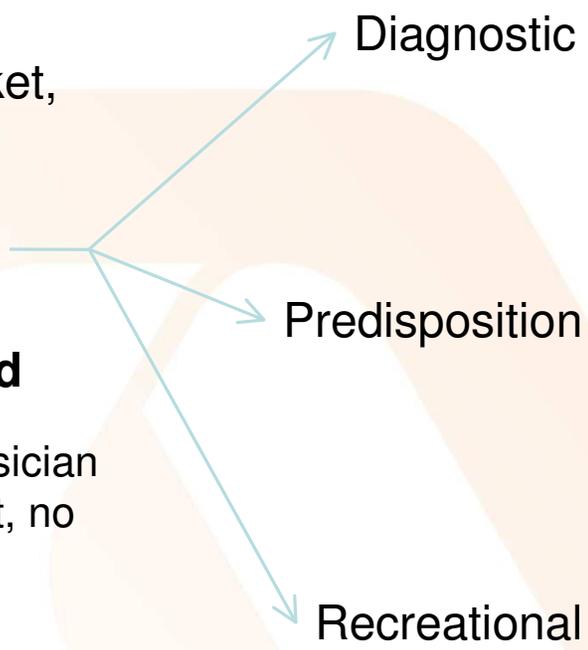
1. **Types of testing offered**

2. **Model of services offered**

Direct access testing: no physician ordering, no results in context, no professional support



“Virtual clinic”: mirrored after traditional clinical practice



Quick Checklist

Patients *and* professionals need to be able to scrutinize these services

- q What **types of genetic testing** is offered?
- q Is the **laboratory** utilized appropriately certified?
- q Is **educational information** provided?
- q What **level of services** are provided?
- q Who provides those services – **genetics professionals**?
- q Are **results interpretation** provided?
- q Will they **speak with your doctor**?
- q Are **costs** are clearly stated?
- q Are company **privacy policy** and standards available?

Examples of New Models |

New Models: Kimball Genetics

kimball genetics ■●

- A CLIA lab that provides access to testing and GC services
- Testing for genetic conditions that are “preventable or treatable”
- How it works:
 - Test ordered directly by patient, required to provide physician contact info
 - Results released directly to patient’s physician
 - Company provides interpretation reports, recommendations, and education
- GCs available by phone
 - For patients, their families, and physicians

New Models: DNA Direct

- Web-based testing modeled after clinical genetics services
- Pre-test education, questionnaires
 - What is the condition / disease?
 - How is it inherited?
 - What testing can or can't tell you?
 - Who is appropriate for testing?
 - What is the testing process?
 - What can you do with results?
 - Insurance coverage
 - Family history assessment
- Genetic counseling services
 - Condition-specific protocols (i.e. – BRCA)

The screenshot shows the DNA Direct website interface. The main heading is "What Causes Cystic Fibrosis?" and the site is identified as "DNA Direct A Genetic Testing Center of Excellence". The page features a navigation menu with options like "PATIENTS", "Tests We Offer", "Genetics & You", "Health Tools", and "About Us". A search bar is present with the text "Enter search words here...". Below the navigation, there is a section titled "What Causes Cystic Fibrosis" with a list of related topics such as "Alpha-1 Antitrypsin Deficiency", "Blood Clotting Disorders", "Breast & Ovarian Cancer", "Cystic Fibrosis", "Drug Metabolism (CYP2D6 Gene)", "Hemochromatosis", "Infertility", "PreGen-Plus Colon Cancer Screening", "Recurrent Pregnancy Loss", and "Sarcosin". A central diagram titled "Autosomal recessive" illustrates the inheritance pattern. It shows a carrier father and a carrier mother at the top, with lines leading to four children: an unaffected son, a carrier daughter, a carrier son, and an affected daughter. A legend indicates that blue shading represents "Unaffected", white represents "Affected", and a blue vertical bar represents a "Carrier". To the right of the diagram, there is a sidebar with a "Cystic Fibrosis testing" section, a "Price: \$260" box, and a "U.S. News" logo with a testimonial.

New Models: DNA Direct

- Post-test interpretation of results
 - Comprehensive web-based report
 - Customized to family & medical history
 - Includes lab report, physician & family letter
 - Helps build an action plan for next step

- 34% have tested positive



- *People are testing appropriately!*

Breast Ovarian Cancer Report 2008-02-14

1 Your Test Results

You have been tested for an inherited condition known as hereditary breast and ovarian cancer, or HBOC. The test you took, called **full sequencing**, looks at the full DNA of the two genes most commonly associated with hereditary breast and ovarian cancer. These genes are called BRCA1 and BRCA2 (short for "breast cancer"). This test is a comprehensive analysis, which can identify the majority of changes believed to cause HBOC.

It is estimated that 1 in every 500 to 750 people in the general population carry a genetic change that gives them a higher-than-average lifetime risk for breast and ovarian cancer. Both men and women carry these genetic changes with equal frequency.

Your test found that you have a gene change associated with hereditary breast and ovarian cancer. This gene change is a mutation called 3036de14 located on the BRCA2 gene.

Your Test Results Explained

You told DNA Direct that:

- You were diagnosed with breast cancer at age 35 years and had a right mastectomy in 1996.
- You have a significant family history of cancer.
- Your sister was diagnosed with bilateral breast cancer.
- Your mother was diagnosed with breast cancer at age 40 and died after the cancer spread to her liver at age 55.
- Your maternal aunt was diagnosed with ovarian cancer.
- Your maternal first cousin was diagnosed with ovarian cancer at age 50.
- Your maternal ethnic background is German and English.
- You do not have a paternal history of cancer.
- Your paternal ethnic background is Irish, Swedish, and English.

Because you carry the 3036de14 mutation, you have an increased risk to develop another cancer. Your personal risks are summarized in the table below:

Type of Cancer	Risk
Second Breast Cancer	50-60%
Ovarian Cancer	10-20%

New Models: Navigenics

| Vision:

To **improve individuals' health** across the population by educating, empowering and motivating people to take action to prevent the onset of disease or lessen its impact.

| Product Description:

The Navigenics™ Health Compass is an innovative service which informs individuals of their **genetic predisposition** for a variety of **common diseases** and provides **guidance** and **information** on how to delay or prevent the onset of those diseases and focus healthcare strategies.

| Key Company Standards:

- Quality science with a clinical basis
- CLIA lab, guarantee 100% call rates
- Health and wellness partnerships
- Genetic counseling support
- Security and privacy
- External advisory boards

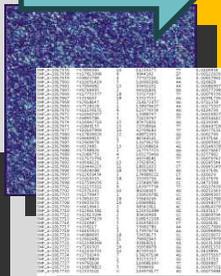
New Models: Navigenics

CLIA-Certified Lab

Receive & process biological samples



Data



Navigenics

Scientific and clinical curation of literature



Genetic Counselors



Personalized, secure web portal



Data analysis & risk estimation algorithms

$$\frac{p(D)}{p(D|NN)} = f_{RR}\lambda_{RR} + f_{RN}\lambda_{RN} + f_{NN}$$

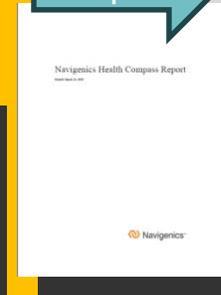
Physicians

Order tests

Release results

Provide follow-up

Report



Collaborators:



Collaborators:



MayoClinic.com



Collaborators:



Scientific Criteria

- | The association is replicated and published in top-tier journals
 - >60% of independent studies
- | Reasonable sample size to detect weak effects
 - OR < 1.5 needs at least 250 cases/250 controls (most studies have 1000's)
- | Statistically significant result after correction for multiple testing
- | Adjustment for population stratification
- | Supporting evidence:
 - Adjustment for confounders
 - Functional data
- | Well designed study and sound lab practice

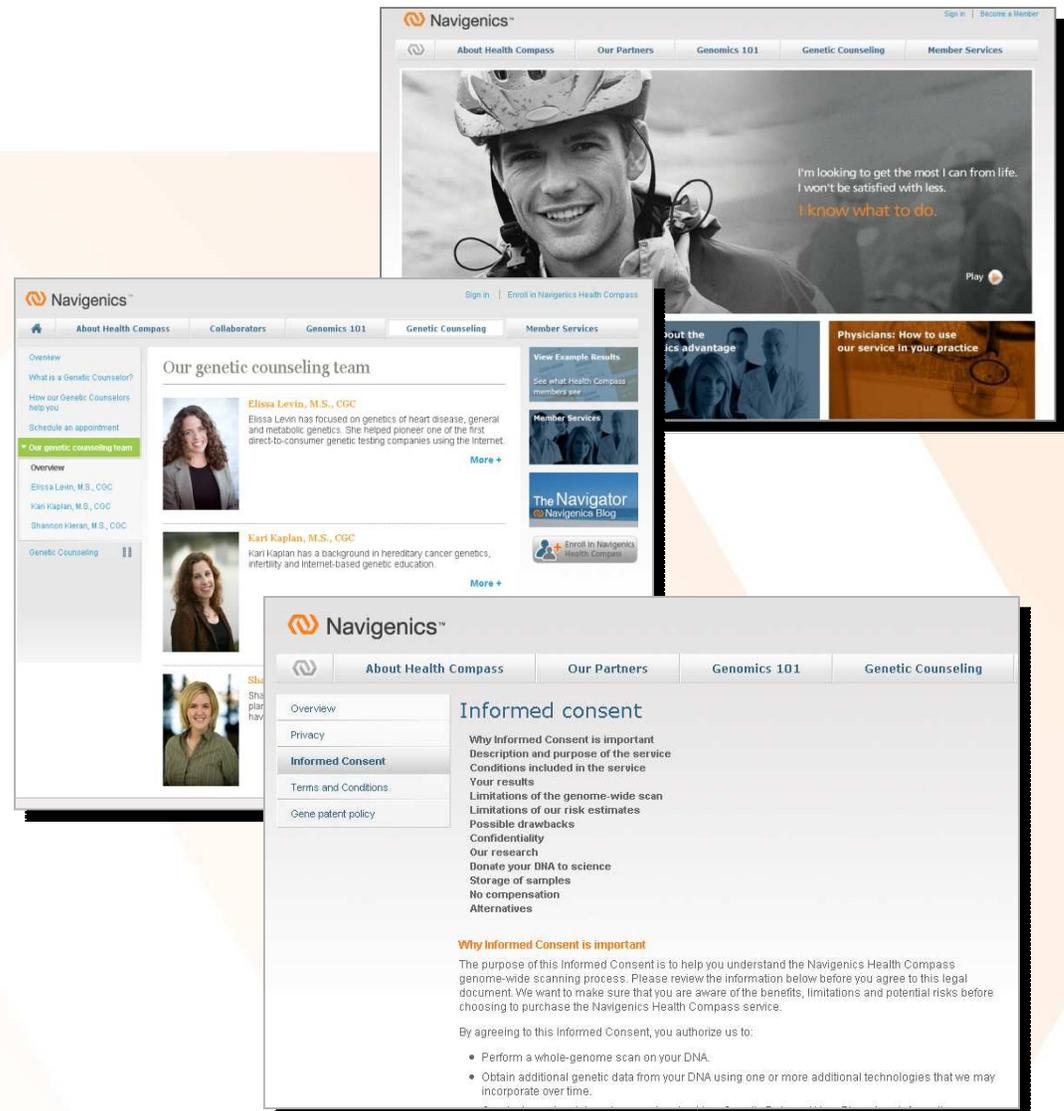
Clinical Criteria

- | The condition affects > one in 1,000 Americans
- | The disease/condition must be clinically relevant and “actionable”
 - Preventable
 - Early screening makes a difference
 - Lifestyle makes a difference
 - Diet makes a difference
 - Medication/surgery makes a difference
 - Affects multiple organ systems
 - Impacts other diseases
- | The risk information must be clinically/socially responsible
 - No IQ, athletic propensity, sticky earwax,

>95% of studies fail to meet our criteria

Online Education

- | Transparent
- | Set expectations
- | Information/audiovisual tools
 - Genetics 101
 - How the process works
 - Behind the science
 - How GCs can help
 - Working with your doctor
 - About Navigenics
 - Genetic discrimination
 - Privacy
- | Informed Consent
- | Access to genetic counselors



Genetic Risk Assessment and Communication

1

Sample Acquisition



2

CLIA Laboratory



3

Bioinformatics

SNP_A-1917680	rs1783662
SNP_A-1917683	rs6669915
SNP_A-1917685	rs2909678
SNP_A-1917686	rs632683
SNP_A-1917688	rs7598926
SNP_A-1917689	rs2086753
SNP_A-1917691	rs1717276
SNP_A-1917697	rs2019535
SNP_A-1917698	rs1756405
SNP_A-1917701	rs1115531
SNP_A-1917702	rs2371333
SNP_A-1917705	rs1379947
SNP_A-1917707	rs2850232
SNP_A-1917708	rs9933070
SNP_A-1917709	rs6419643

2,000 pages

4

Private Web Portal



5

Ongoing Updates



GC Services

Receiving Results

1. Secure login to personal web portal
2. Introduction tutorial
3. Select conditions (active opt-in)

Current list of conditions:

- Abdominal Aneurysm
- Alzheimer's Disease
- Atrial Fibrillation
- Breast Cancer
- Celiac Disease
- Colon Cancer
- Crohn's Disease
- Diabetes Type 2
- Glaucoma
- Grave's Disease
- Heart Attack
- Lung Cancer
- Lupus
- Macular Degeneration
- Multiple Sclerosis
- Obesity
- Osteoarthritis
- Psoriasis
- Prostate Cancer
- Restless Leg Syndrome
- Rheumatoid Arthritis

The screenshot shows the Navigenics web portal interface. At the top, there is a navigation bar with the Navigenics logo and the text 'Welcome, elevin74 | Sign out | Manage Profile'. Below the navigation bar, there are several tabs: 'Your Results', 'Learn More', 'Sharing Results', 'Genetic Counseling', and 'Member Services'. The main content area features a large blue banner with the text 'Welcome back!' and a prominent orange button labeled 'View your results' with a 'Next >' arrow. To the right of the banner, there is a section titled 'Understanding your results' which displays a grid of disease risk cards. The cards include: 'Disease' (You: 0.55%, Avg: 0.58%), 'Alzheimer's disease' (You: 2.4%, Avg: 9%), 'Glaucoma' (You: 0.21%, Avg: 1.4%), and 'Restless leg syndrome' (You: 4.1%, Avg: 1.1%). A 'Watch Video' button is located below these cards. Below the banner, there is a 'Latest headlines' section with a list of links: 'Our look at DNA and lung cancer', 'FAQ for physicians', 'Navigenics on PBS', 'Bringing genes into breast cancer detection', 'Navigenics in the American Journal of Human Genetics', 'In Aspen, a focus on prevention', and 'My genes need counseling?'. At the bottom of the page, there is a footer with links for 'About Us', 'Blog', 'Scientific Details', 'For Physicians', 'Policies', and 'Help', along with the text 'Questions? Call us at (866) 522-1585'.

In-Depth Condition Reports

Condition summary

Summary

Your odds are 0.55% (Your odds are 0.55% higher than the average for the rest of the population)

What you can do

66% There's no proven way to prevent celiac disease. However, you can avoid celiac disease by eating a gluten-free diet. If you have celiac disease, you should avoid gluten. If you have celiac disease, you should avoid gluten. If you have celiac disease, you should avoid gluten.

What does it mean?

Genes are only part of the story. Environmental and lifestyle factors also play a role. Studies of twins show how genes and environment interact to cause disease. If you have celiac disease, you should avoid gluten. If you have celiac disease, you should avoid gluten. If you have celiac disease, you should avoid gluten.

Action steps

What you can do

66% There's no proven way to prevent celiac disease. However, you can avoid celiac disease by eating a gluten-free diet. If you have celiac disease, you should avoid gluten. If you have celiac disease, you should avoid gluten. If you have celiac disease, you should avoid gluten.

Points to remember

Your Navigenics Health Compass is not a diagnostic test and cannot tell you whether you have celiac disease. Only your doctor can do that. Don't wait to call your doctor — that could prevent your doctor from diagnosing you correctly.

Related links

- ▶ [Dietary results with your doctor](#)
- ▶ [Having results with your doctor](#)
- ▶ [Protecting your privacy](#)

In-depth condition information

Introduction

Celiac disease is a digestive condition triggered by consumption of the protein gluten, which is found in wheat, grain, cereals, rye, barley, and malt. When a person with celiac disease eats foods containing gluten, their immune system reacts and damages the lining of the small intestine and its ability to absorb certain nutrients from food.

Related links

- ▶ [Dietary results with your doctor](#)
- ▶ [Having results with your doctor](#)
- ▶ [Protecting your privacy](#)

Marker details

What we found for celiac disease

We looked at three places on your genome where a one-letter difference in the genetic code raises your odds of celiac disease. At each location, there are two markers, for a total of six possible risk markers. You have five of the six risk markers we looked for.

Your genetic markers

Gene or location	Risk marker	Your markers	Odds ratio	Source
L2-IL22 loci	C	CC	2.02	Nature Genetics, 2007
HLA-DQ2.5	T	TC	7.04	Nature Genetics, 2007
CTLA4	T	TT	1.54	European Journal of Human Genetics, 2005

What it means

Everyone has some chance of developing celiac disease. Each risk marker increases your odds of developing celiac disease a little, some a lot. The "odds ratio" column in the chart above shows their relative impact.

Sharing with physician

A report for your physician

Your personal Navigenics Health Compass report file can be saved to your computer or emailed to your physician. The report will include your odds of celiac disease and the genetic markers we looked for. The odds ratio column in the chart above shows their relative impact.

Include only my elevated risk conditions

- Celiac disease
- Adherence's disease
- Lupus
- Breast cancer
- Multiple sclerosis
- Crohn's disease
- Obesity
- Osteoarthritis
- Diabetes, Type 2
- Psoriasis
- Restless legs syndrome
- Glaucoma
- Rheumatoid arthritis
- Graves' disease

A genome-wide association study for celiac disease identifies risk variants in the region harboring IL2 and IL21

The researchers found a number of variants that map to the key areas of the genome connected to celiac disease.

Why it is important

Celiac disease is a common condition, present in about 1 percent of people, when inflammation in the small intestine is induced by dietary intake of the proteins wheat, barley and rye (collectively "gluten") ingested outside of the naturally-associated HLA-A*02:01 variants, just the most common in healthy individuals. 2007 research found that the variant contributes to, but is not sufficient for, disease development. The researchers wanted to find out what other genetic factors could contribute to celiac disease. They found 766 SNPs (single nucleotide polymorphisms) that were associated with celiac disease and 1,427 healthy controls.

Outside the HLA area, they found four novel variants in a block of DNA on chromosome 10. The researchers found that the variant on chromosome 10 was independently confirmed the association of two further variants with celiac disease. The researchers also found a variant on chromosome 10 that was independently confirmed the association of two further variants with celiac disease. The researchers also found a variant on chromosome 10 that was independently confirmed the association of two further variants with celiac disease.

Genetic Counseling Service

- | Access to certified GCs:
 - Core component, cost included with testing
 - Pre-test, post-test, additional access as new data emerges
- | GCs have specific training in genome-wide screening and risk assessment for common, multifactorial conditions
- | Continuity of care: GC assigned to each tester
- | Convenient scheduling, low barriers to engagement
- | Identify in-person services and additional testing, as appropriate

Schedule genetic counseling

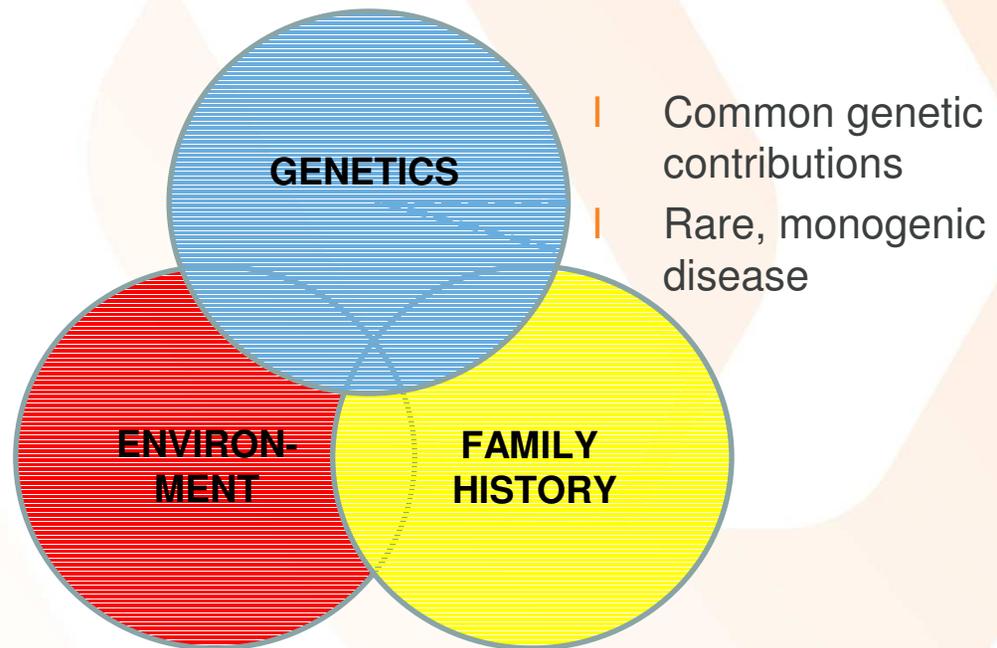
You can schedule a time to speak with one of our genetic counselors at any point. Enter date and time below:

Putting Results In Context

- | Phone-based genetic counseling:
 - Conditions of most concern – based on results and patient interest
 - Multi-generation family history collection and assessment
 - Understanding results and review next step options

- | Need to consider many components of risk and disease contribution



How Are People Handling It?

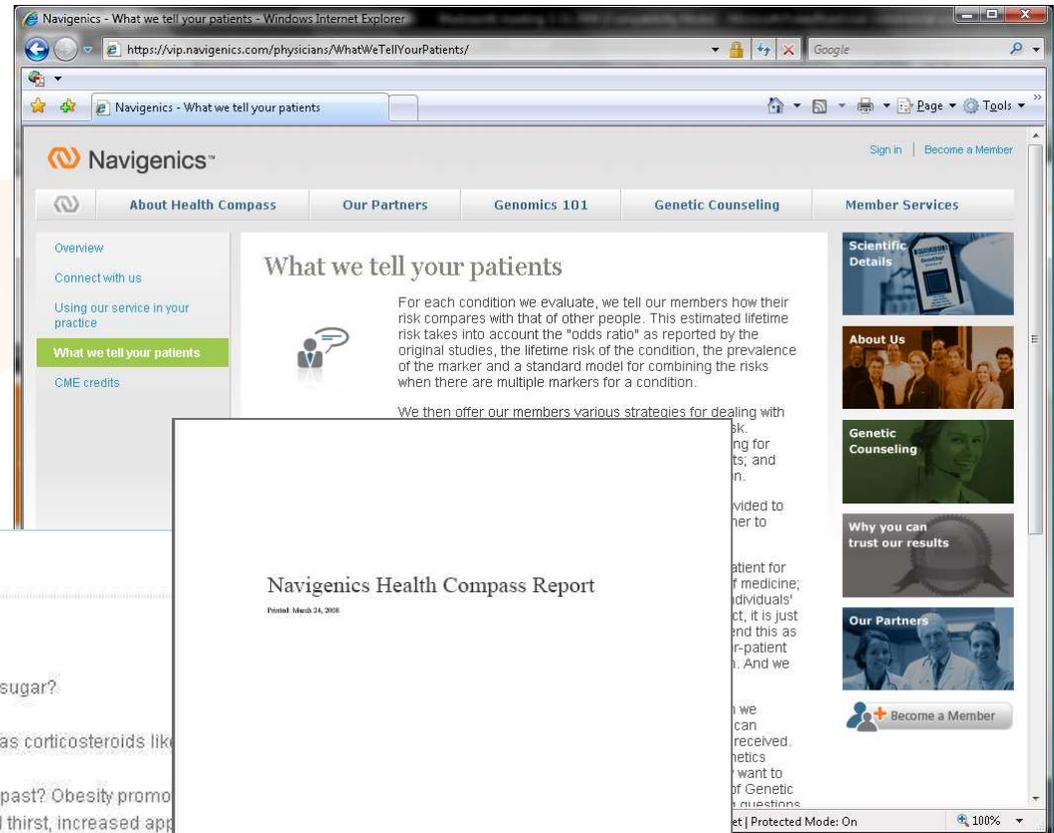
- | Early reports = no extreme reactions to date
 - People can understand concept of risk factors
 - Those who are initially anxious report returning to baseline
 - Many further report feeling empowered and informed



- | 46% of early testers took action after testing
 - Lifestyle/behavior modification
 - Screening or interventions in conjunction with healthcare provider
- | *Additional research is needed to further understand the long-term impact of genomic risk information on health behaviors*

Engaging Providers

- | How to talk to your doctor
- | Physician Report
- | Provider access to GCs
- | Web portal for providers (education, ordering...)



 **Talking to your doctor**

What should I tell my doctor?

- Do you have a family history of diabetes?
- Has anyone ever mentioned that you had a high or borderline blood sugar?
- If you have been pregnant, did you have gestational diabetes?
- Are you taking any medication that can raise your blood sugar, such as corticosteroids like prednisone?
- Are you under extreme stress, which can elevate your blood sugar?
- What is your current weight in comparison to what it has been in the past? Obesity promotes insulin resistance.
- Do you have any symptoms of possible diabetes, such as increased thirst, increased appetite, increased urination, blurred vision or fatigue?

What can my doctor do?

- Order blood tests to get a baseline of your blood sugar, probably including a fasting blood sugar and possibly an oral glucose tolerance test.
- Advise you about starting a weight loss and exercise program.
- Perform a baseline exam and lab tests to check organs that can be affected by diabetes: eyes, heart, and kidneys.
- Be attentive to even modest elevations in blood pressure or cholesterol, as these are affected by diabetes.
- Advise you to get a glucometer so you can periodically check your blood sugar on your own.

Navigenics Health Compass Report

Printed: March 24, 2008

 Navigenics™

Physician Education



- I CME article authored by Dr. Geoff Ginsburg of Duke
- I Sponsored by Navigenics (no role in creation)
- I Currently has generated about **6000** Medscape member readers, **2500** applied for credit (MDs, nurses, other HCPs)

Log In | Register
eMedicine | The Medscape Journal

Latest | News | CME | Conferences | Resource Centers | Journals & Reference | Experts & Viewpoints

[Email This](#)

A Primer on Genomic and Personalized Medicine: How Will It Affect Your Practice? CME/CE

[Author Information and Disclosures](#)

Release Date: February 29, 2008; **Valid for credit through March 1, 2009**

Credits Available

Physicians - maximum of 1.0 *AMA PRA Category 1 Credit(s)*™ for physicians;
Nurses - 1.0 nursing contact hours (None of these credits is in the area of pharmacology)

To participate in this internet activity: (1) review the target audience, learning objectives, and author disclosures; (2) study the education content; (3) take the post-test and/or complete the evaluation; (4) view/print certificate [View details](#).

Supported by an independent educational grant from

Supported by an independent educational grant from

CME/CE Information

[Earn CME/CE Credit »](#)

Contents of This CME/CE Activity

[A Primer on Genomic and Personalized Medicine: How Will It Affect Your Practice?](#)

Geoffrey S. Ginsburg, MD, PhD

[Go to CME/CE Test Questions](#)

Legal Disclaimer

The material presented here does not necessarily reflect the views of Medscape or companies that support educational programming on [www.medscape.com](#). These materials may discuss therapeutic products that have not been approved by the FDA for their intended use. The professional should be consulted before using any therapeutic product described in this educational activity.

CME/CE Test

A Primer on Genomic and Personalized Medicine: How Will It Affect Your Practice?

Please answer the test questions below. Some activities require you to meet a certain passing score to earn credit. Questions answered incorrectly will be highlighted.

- Which of the following genetic variations is thought to be largely responsible for complex human diseases?
 - Chromosomal deletions
 - Chromosomal insertions
 - Repetitive DNA elements
 - Single nucleotide polymorphisms
 - Large chromosomal rearrangements
- Which of the following statements is false?
 - Microarrays are the major tool used to measure genome wide changes in gene expression
 - Reverse transcriptase polymerase chain reaction is rarely used to measure transcription of discrete gene sets
 - A proteomic assay measuring C-reactive protein, B-type natriuretic peptide, and cardiac troponin I can be used for risk stratification of patients with acute coronary syndrome
 - Advances in high-throughput mass spectrometry or nuclear magnetic resonance technologies allow detection, analysis, and identification of a range of metabolites
- Which of the following statements is false? Genome-wide association studies have:
 - Identified a SNP locus for coronary artery disease at 9p21
 - Not identified several SNPs for diabetes susceptibility
 - Identified a susceptibility gene for obesity
 - Identified genes for Crohn's disease and rheumatoid arthritis
- Which of these statements about pharmacogenomics tests is false?
 - Genotyping for VKORC1 and CYP2C3 can help account for the variability in warfarin dosing requirements
 - Homozygosity for defective thiopurine methyltransferase gene can lead to a high risk for Emercitapone toxicity
 - Quantifying expression of HER2/neu to identify patients with breast cancer who are most likely to benefit from trastuzumab is an example of a targeted therapy
 - Cardiac troponin I (cTnI) measurement does not enable a targeted therapeutic approach

[Save and Proceed](#)

Setting Industry Standards |

Professional Statements on DTC

- | ACMG
- | ASHG
- | NSGC

**NSGC POSITIONS STATEMENT
DIRECT TO CONSUMER GENETIC TESTING**

Genetic testing is currently available to consumers without the involvement of healthcare providers. A growing number of "Direct to Consumer" testing companies offer tests that may diagnose genetic disorders. In addition, DTC genetic analysis is available for purposes such as ancestry, marketing of "nutrigenomic" products, and other consumer-oriented testing.

The National Society of Genetic Counselors (NSGC) recognizes the need to increase access to medical testing services for some individuals seeking genetic testing without input from a healthcare provider. The purpose and application of these results for themselves and their families is complex. There is limited regulatory oversight to increase the likelihood that a DTC service undertaking a direct-to-consumer company has addressed the following issues:

1. Are consumer-friendly professionals with expertise for individuals seeking genetic testing available?
2. Is information disclosed in a timely, valid and accurate manner?
3. Will results be given in a clear explanation of the test and providing appropriate counseling?
4. Are patients encouraged to consult their healthcare providers for interpretation of their results?
5. Are consumer referrals available, either on-site or after testing to assure appropriate counseling as needed?
6. Is there a process for handling complaints consistent with accreditation standards?
7. What safeguards are in place to protect the privacy of the consumer's genetic information?

* Approved by the ASHG Board of Directors in January 2007.

ASHG Statement* on Direct-to-Consumer Genetic Testing in the United States

Direct-to-consumer (DTC) genetic testing has been gaining prominence over the past several years.¹ Proponents of DTC testing cite benefits that include increased consumer access to testing, greater consumer autonomy and empowerment, and enhanced privacy of the information obtained. Critics of DTC genetic testing have pointed to the risks that consumers will choose testing without adequate context or counseling, will receive tests from laboratories of dubious quality, and will be misled by unproven claims of benefit.

Currently, DTC genetic testing is permitted in about half the states² and is subject to little oversight at the federal level. In July 2006, the Government Accountability Office issued a report documenting troubling marketing practices by some DTC testing companies,³ and the Federal Trade Commission (FTC) issued a consumer alert cautioning consumers to be skeptical about claims made by some DTC companies.⁴ Internationally, several countries have issued reports cautioning against its use,^{5,6} and several European countries have banned or are considering banning it entirely.

DTC testing has emerged during a period of rapid growth in the number of genetic tests. Today, there are more than 1,100 genetic tests available clinically, and several hundred more are available in research settings. Although most genetic testing is currently available only through a health care provider, an increasing variety of tests are being offered DTC, often without any health care provider involvement or counseling. The range of tests available DTC is broad, from tests for single-gene disorders, such as cystic fibrosis, to tests for predisposition to complex, multifactorial diseases, such as depression and cardiovascular disease. In addition to providing test results DTC, some companies also make recommendations regarding lifestyle changes on the basis of these results, such as changes in diet or use of nutritional supplements.

Ensuring adequate information, high-quality laboratories, and accurate claims and interpretation of test results is important for all genetic tests, including those provided DTC. At the same time, a one-size-fits-all approach is not appropriate for DTC tests, because the types of tests being offered are heterogeneous, and their consequences are wide ranging. A test may be used to diagnose disease, to predict risk of future disease, to determine the risk of passing on a disease to one's offspring, to aid in therapy selection, or to guide "lifestyle" choices such as diet and skin care. Different possible actions may result from different types of tests. For example, tests to determine whether someone is a carrier of a mutation for a particular disease may affect the choice of whether or whom to marry, whether to have children, and whether to terminate a pregnancy. Thus, the level of evidence required before a test is offered DTC, and the safeguards appropriate to ensure adequate consumer protection, will differ depending on what is being tested for and what the foreseeable consequences of testing are. Whereas the DTC model may be contraindicated for certain types of tests, the availability of other tests in the absence of a

health care provider may not compromise, and may even foster, patient health. This policy statement does not attempt to set the dividing line between those tests that should be offered DTC and those that should not; rather, it sets forth principles that should govern all health-related genetic tests that are offered DTC.

Scope of this Statement

While DTC testing also encompasses paternity and ancestry testing, this policy statement addresses solely those genetic tests that make health-related claims or that directly affect health care decision making. In addition, although "DTC" is sometimes used to refer to tests advertised but not sold DTC, this policy statement focuses on tests that can be ordered directly by a consumer and whose results are reported DTC without an independent health care provider—one not employed by the testing company—serving as an intermediary.

Context

DTC genetic testing differs from traditional genetic testing in that consumers order tests and receive test results without an independent provider serving as an intermediary. Whether a company is permitted to provide DTC genetic-testing services is a matter of state law. Currently, about half the states permit DTC genetic testing.⁷ Additionally, although some states require a provider to order a test on behalf of a patient, this requirement can generally be fulfilled by a physician employed by the laboratory. Some DTC companies offer genetic counseling, while others do not.

DTC tests are typically advertised and sold over the Internet. After the consumer orders the test, the testing company sends a sample-collection kit (e.g., buccal swab or blood-spot collection). The consumer sends back the sample, and the company performs the test and sends a test report via the Internet or the mail. This context has led to the concern that consumers will not receive adequate counseling—either in advance, to ensure that the test is appropriate, or on receipt of test results, to ensure that consumers comprehend the complex information and understand the consequences of testing for themselves and their family members.

Quality

Because of the fragmented regulatory environment for genetic testing in general, there is concern that the quality of the tests offered DTC may be inadequate. For a test to be of good quality, the laboratory performing it must be able to obtain the correct answer reliably, meaning that it detects a particular genetic variant when it is present and does not detect the variant when it is absent. A test's accuracy is referred to as "analytic validity." Further, there must be adequate scientific evidence to support the



- Officers**
 Joe Leigh Simpson, MD
 President
 Marilyn C. Jones, MD
 Past President
 Bruce Korff, MD
 President-Elect
 Louanne Hudgins, MD
 VP Clinical Genetics
 Thomas W. Prior, PhD
 VP Laboratory Genetics
 Elaine B. Spector, PhD
 Secretary
 Lisa Shaffer, PhD
 Treasurer

ACMG Media Contact: Kathy Beal, 301-238-4382
 kbeal@acmg.net

STATEMENT ON DIRECT-TO-CONSUMER GENETIC TESTING

Genetics Professionals Should Be Part of Genetic Testing Process Says American College of Medical Genetics

BETHESDA, MD – September 24, 2007— More genetic tests are available than ever before, ranging from home DNA test kits, tests that can help determine a person's predisposition to certain diseases, and even home paternity tests. Along with greater availability of genetic tests and increased Direct-to-Consumer (DTC)

SOCIETY NEWS

posed risks to the public. The American College of Medical Genetics and Genomics (ACMG) urges consumers to involve a genetics expert in the appraisal efforts to educate the public about resources to help identify and manage those at risk, but it is absolutely critical that the public be part of the process. Genetic testing should be used for genetic counseling, both in the consideration of a patient's interpretation of test results. Trained genetic counselors, PhD geneticists and certified genetic counselors perform this important role," says Joe Leigh Simpson, PhD, FACMG, Executive Director of the American College of Medical Genetics. "Genetic testing is a highly complex specialty. A genetics expert such as a genetic counselor can help people to sort out family history factors and what test results may mean for consumers made through the numerous tests available. Many tests have not met standards of scientific validity or have not met standards of scientific rigor," says Joe Leigh Simpson, PhD, FACMG, Executive Director of the American College of Medical Genetics.

-more-

Setting Industry Standards

- **Validity** of scientific and clinical information provided
- **Accuracy and quality** of testing in a CLIA-certified lab
- **Clinical relevance** of information provided
- **Actionable** next steps for health conditions assessed
- **Access to genetic counseling** by qualified professionals
- **Security and privacy** including consistence with HIPAA regulations
- **Ownership of genetic information** belongs to the individual
- **Physician education and engagement**
- **Transparency** at all levels of the service
- **Measurement** to assess impact on health outcomes

The Personalized Medicine Coalition is leading an industry standards setting event with Navigenics and other industry leaders and stakeholders (Dec. 2008)

Questions?

Please contact me at any point with questions!

Elissa Levin, MS, CGC

Director, Genetic Counseling Program

650-585-7714

elevin@navigenics.com