Personal Genomics

http://biochem158.stanford.edu/Personal%20Genomics.html


Doug Brutlag
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Stanford University School of Medicine
Genetic Penetrance of Inherited Diseases

- Many inherited diseases are Mendelian and highly penetrant
  - Sickle cell disease
  - Thalassemias
  - Huntington’s disease
  - Color blindness
  - Cystic fibrosis

- Most common diseases are complex (multifactorial - caused by multiple genes or multiple pathways as well as multiple environmental factors) and of low penetrance
  - Familial
  - Predisposition to disease
  - Very large environmental and/or behavioral component
    - Type I diabetes and other autoimmune diseases (lupus, rheumatoid arthritis, hyperthyroidism, Crohn’s disease, Celiac Sprue, irritable bowel disease etc.)
    - Type 2 diabetes
    - Coronary heart disease (atherosclerosis)
    - Asthma, COPD, pulmonary fibrosis
  - Many complex diseases can be avoided with diet, nutrition, exercise or behavioral modification
  - Many complex diseases can also be monitored by increased vigilance (another behavioral modification)
So What Can We Learn from Personal Genomics?

• Disease risk for common diseases
  – Genetic predisposition towards a disease (relative risk or odds ratio)
  – Genetic versus environmental contributions to disease (penetrance)
  – How to alter your environment and behavior to avoid or detect the disease

• Disease Carrier status
  – Premarital genetic counseling
  – Preimplantation genetic diagnosis
  – Neonatal diagnosis (amniocentesis, chorionic villus sampling, fetal DNA in mother)

• Familial traits, diseases and relationships
  – Known family diseases (breast cancer, colorectal cancer, lysosome storage diseases, etc.)
  – Paternity (10% of people do not know their true biological father)
  – Maternity (about 1% of people do not know their true biological mother)
  – Inbreeding and incest lead to increased homozygosity and recessive diseases
  – Orphans can find family relations

• Pharmacogenomics and Pharmacogenetics: Drug susceptibility
  – Efficacy of common drugs
  – Adverse reactions to common drugs

• Ancestry
  – One can follow maternal line using mitochondrial DNA SNPs
  – Males can follow paternal line using Y chromosome SNPs
  – Autosomal shared haplotype regions with recent relatives (up to 5th cousins)
23andMe can help you manage risk and make informed decisions...

Learn valuable health & ancestry information.

welcome to you®

23andMe DNA Spit Kit

$99

Order Now
Find out what your DNA says about you and your family.

- Learn what percent of your DNA is from populations around the world
- Contact your DNA relatives across continents or across the street
- Build your family tree and enhance your experience with relatives

order now  $99
23andMe Spit Kit
http://23andme.com/
Before providing your sample, register your kit at:

www.23andme.com/start

Your sample will NOT be processed unless it is registered.

1. Register
2. Fill line
3. Close funnel
4. Remove funnel
5. Screw on cap
6. Shake 5 seconds
7. Seal in bag
8. Mail in box
23andMe Spit Kit
http://23andme.com/
23andMe Spit Kit
http://23andme.com/
23andMe Spit Kit
http://23andme.com/
### Disease Risks (120, 1 locked report)

<table>
<thead>
<tr>
<th>Disease</th>
<th>Your Risk</th>
<th>Average Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prostate Cancer</td>
<td>22.4%</td>
<td>17.8%</td>
</tr>
<tr>
<td>Colorectal Cancer</td>
<td>7.1%</td>
<td>5.6%</td>
</tr>
<tr>
<td>Melanoma</td>
<td>6.0%</td>
<td>2.9%</td>
</tr>
<tr>
<td>Chronic Kidney Disease</td>
<td>4.2%</td>
<td>3.4%</td>
</tr>
<tr>
<td>Restless Legs Syndrome</td>
<td>2.5%</td>
<td>2.0%</td>
</tr>
</tbody>
</table>

### Carrier Status (49)

- Alpha-1 Antitrypsin Deficiency: Variant Absent
- Bloom's Syndrome: Variant Absent
- BRCA Cancer Mutations (Selected): Variant Absent
- Canavan Disease: Variant Absent
- Cystic Fibrosis: Variant Absent
- DPD Deficiency: Variant Absent
- Familial Dysautonomia: Variant Absent
- Factor XI Deficiency: Variant Absent

### Traits (57)

- Alcohol Flush Reaction: Does Not Flush
- Bitter Taste Perception: Can Taste
- Earwax Type: Wet
- Eye Color: Likely Brown
- Hair Curl: Straighter Hair on Average

### Drug Response (21)

- Captopril (Plavix®) Efficacy: Greatly Reduced
- Abacavir Hypersensitivity: Typical
- Alcohol Consumption, Smoking and Risk of Esophageal Cancer: Typical
- Fluorouracil Toxicity: Typical
- Response to Hepatitis C Treatment: Typical
### Disease Risk

**Show results for** 

- **23andWe Discoveries** were made possible by 23andMe members who took surveys.

**Locked Reports**

<table>
<thead>
<tr>
<th>Name</th>
<th>Confidence</th>
<th>Your Risk</th>
<th>Avg. Risk</th>
<th>Compared to Average</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alzheimer's Disease</td>
<td>🡫</td>
<td>✔️</td>
<td>✔️</td>
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</tr>
</tbody>
</table>

**Elevated Risk**

<table>
<thead>
<tr>
<th>Name</th>
<th>Confidence</th>
<th>Your Risk</th>
<th>Avg. Risk</th>
<th>Compared to Average</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prostate Cancer</td>
<td>🡫</td>
<td>22.4%</td>
<td>17.8%</td>
<td>1.26x</td>
</tr>
<tr>
<td>Colorectal Cancer</td>
<td>🡫</td>
<td>7.1%</td>
<td>5.6%</td>
<td>1.27x</td>
</tr>
<tr>
<td>Melanoma</td>
<td>🡫</td>
<td>6.0%</td>
<td>2.9%</td>
<td>2.10x</td>
</tr>
<tr>
<td>Chronic Kidney Disease</td>
<td>🡫</td>
<td>4.2%</td>
<td>3.4%</td>
<td>1.22x</td>
</tr>
<tr>
<td>Restless Legs Syndrome</td>
<td>🡫</td>
<td>2.5%</td>
<td>2.0%</td>
<td>1.25x</td>
</tr>
<tr>
<td>Exfoliation Glaucoma</td>
<td>🡫</td>
<td>2.2%</td>
<td>0.7%</td>
<td>2.90x</td>
</tr>
<tr>
<td>Abdominal Aortic Aneurysm</td>
<td>🡫</td>
<td>✔️</td>
<td>✔️</td>
<td></td>
</tr>
<tr>
<td>Ankylosing Spondylitis</td>
<td>🡫</td>
<td>✔️</td>
<td>✔️</td>
<td></td>
</tr>
<tr>
<td>Asthma</td>
<td>🡫</td>
<td>✔️</td>
<td>✔️</td>
<td></td>
</tr>
</tbody>
</table>
disease risk

Prostate Cancer

Prostate cancer is by far the most common cancer affecting men. (Women don’t have prostate glands and therefore cannot get prostate cancer, but can pass markers to their children.) About one in six men will develop prostate cancer over their lifetimes, according to the American Cancer Society. Fortunately, most prostate tumors grow slowly, and if detected early, treatment may help control their size. Until recently, the only well-known risk factors for prostate cancer were age, ethnicity, and family history. Although advanced age increases a person’s risk for any type of cancer, the involvement of ethnicity and family history suggests that there is a strong genetic component as well.

The following results are based on 4★ Established Research for 12 reported markers, updated November 4th, 2010.

Learn more about the biology of Prostate Cancer...
Major discoveries in Prostate Cancer...
23andMe Prostate Cancer Risks

Your Genetic Data

Show information for Douglas Brutlag assuming European ethnicity and an age range of 35–79.

What does the Odds Calculator show me?
Use the ethnicity and age range selectors above to see the estimated incidence of Prostate Cancer due to genetics for men with Douglas Brutlag's genotype. The 23andMe Odds Calculator assumes that a person is free of the condition at the lower age in the range. You can use the name selector above to see the estimated incidence of Prostate Cancer for the genotypes of other people in your account.

The 23andMe Odds Calculator only takes into account effects of markers with known associations that are also on our genotyping chip. Keep in mind that aside from genetics, environment and lifestyle may also contribute to one's risk for Prostate Cancer.

Genes vs. Environment

The heritability of prostate cancer is estimated to be 42–57%. This means that genetic and environmental factors contribute nearly equally to differences in risk for this condition. (If you are a woman, you have no chance of getting this type of cancer, but if you have sons, their risk may be affected by what they inherit from you.) Genetic factors that play a role in prostate cancer include both unknown factors and known factors such as the SNPs we describe. Other factors that can increase your risk include being older, having African ancestry, or living in North America, Northwestern Europe, Australia, or the Caribbean islands. The effect of nationality may be tied to diet, as a diet high in red meat and high-fat dairy products, and low in fruits and vegetables, may also put you at increased risk. (sources)
Marker Effects

What does this chart show?
The chart shows the approximate effects of the selected person's genotype at the 12 reported markers. Higher, red bars indicate increased risk from the average, while lower, green bars indicate decreased risk from the average. The light gray bars show the maximum possible effects for the possible genotypes at the marker.

Mouse over individual bars to view additional information about each marker. Click on a bar to view detailed information about that marker below. You can read more about all markers in the technical report.

Citations


Three SNPs in the same area of the genome have recently been found to be independently associated with prostate cancer risk. This region is called 8q24, because it lies within band 24 on the long arm (named the "q" arm) of chromosome 8. The three SNPs are not close to known genes (although there are others located farther away). But other studies have looked at DNA from prostate tumors and found that in the cancerous cells, this area of the genome often has unusual duplications, or extra copies of DNA.

The duplications might contribute to the progression of prostate cancer (for example, by increasing the number of genes related to cell growth), or they might simply be a side effect of the high mutation rate seen in all types of cancer cells. Similarly, the risk-associated versions of the SNPs in the 8q24 region might directly affect activity levels of genes involved in prostate cancer, or they might somehow make it easier for DNA duplications to occur. (And, they might only be linked to yet-unknown SNPs that are directly involved.)

One study has investigated this association in Japanese Americans. Although the SNP also appears to be associated with prostate cancer risk in this population, evidence suggests that the effect of this SNP on risk may differ between populations. Therefore, the exact association in populations with Asian ancestry still needs to be confirmed.

The genotyping services of 23andMe are performed in LabCorp’s CLIA-certified laboratory. The tests have not been cleared or approved by the FDA but have been analytically validated according to CLIA standards. The information on this page is intended for research and educational purposes only, and is not for diagnostic use.
## 23andMe Disease Risks

### Decreased Risk

<table>
<thead>
<tr>
<th>Name</th>
<th>Confidence</th>
<th>Your Risk</th>
<th>Avg. Risk</th>
<th>Compared to Average</th>
</tr>
</thead>
<tbody>
<tr>
<td>Type 2 Diabetes</td>
<td>⭐⭐⭐⭐⭐</td>
<td>19.2%</td>
<td>25.7%</td>
<td>0.75x</td>
</tr>
<tr>
<td>Age-related Macular Degeneration</td>
<td>⭐⭐⭐⭐⭐</td>
<td>2.9%</td>
<td>6.5%</td>
<td>0.44x</td>
</tr>
<tr>
<td>Rheumatoid Arthritis</td>
<td>⭐⭐⭐⭐⭐</td>
<td>1.2%</td>
<td>2.4%</td>
<td>0.52x</td>
</tr>
<tr>
<td>Esophageal Squamous Cell Carcinoma (ESCC)</td>
<td>⭐⭐⭐⭐⭐</td>
<td>0.29%</td>
<td>0.36%</td>
<td>0.80x</td>
</tr>
<tr>
<td>Crohn's Disease</td>
<td>⭐⭐⭐⭐⭐</td>
<td>0.26%</td>
<td>0.53%</td>
<td>0.50x</td>
</tr>
<tr>
<td>Multiple Sclerosis</td>
<td>⭐⭐⭐⭐⭐</td>
<td>0.20%</td>
<td>0.34%</td>
<td>0.59x</td>
</tr>
<tr>
<td>Stomach Cancer (Gastric Cardia Adenocarcinoma)</td>
<td>⭐⭐⭐⭐⭐</td>
<td>0.18%</td>
<td>0.23%</td>
<td>0.77x</td>
</tr>
<tr>
<td>Type 1 Diabetes</td>
<td>⭐⭐⭐⭐⭐</td>
<td>0.07%</td>
<td>1.02%</td>
<td>0.07x</td>
</tr>
<tr>
<td>Primary Biliary Cirrhosis</td>
<td>⭐⭐⭐⭐⭐</td>
<td>0.05%</td>
<td>0.08%</td>
<td>0.66x</td>
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<tr>
<td>Celiac Disease</td>
<td>⭐⭐⭐⭐⭐</td>
<td>0.03%</td>
<td>0.12%</td>
<td>0.28x</td>
</tr>
<tr>
<td>Atrial Fibrillation: Preliminary Research</td>
<td>⭐⭐⭐</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Type 2 Diabetes

The most common type of diabetes, type 2 diabetes mellitus occurs when chronically high blood sugar levels cause a breakdown of the body's natural response to eating sweets and starches. Left untreated, type 2 diabetes can result in kidney failure, blindness, and circulatory problems that increase the risk of heart attack or stroke. In the United States, almost 21 million children and adults have diabetes, but the rate of new diagnoses is increasing.

The following results are based on ★★★★★ Established Research for 11 reported markers, updated March 24th, 2011.

Learn more about the biology of Type 2 Diabetes...

Major discoveries in Type 2 Diabetes...

1 of 3. Smart choices about diet can help delay or prevent type 2 diabetes.
23andMe Type 2 Diabetes Risks

Your Genetic Data

Show information for Douglas Brutlag assuming European ethnicity and an age range of 20-79.

Douglas Brutlag
19.2 out of 100
men of European ethnicity who share Douglas Brutlag’s genotype will develop Type 2 Diabetes between the ages of 20 and 79.

Average
25.7 out of 100
men of European ethnicity will develop Type 2 Diabetes between the ages of 20 and 79.

What does the Odds Calculator show me?
Use the ethnicity and age range selectors above to see the estimated incidence of Type 2 Diabetes due to genetics for men with Douglas Brutlag’s genotype. The 23andMe Odds Calculator assumes that a person is free of the condition at the lower age in the range. You can use the name selector above to see the estimated incidence of Type 2 Diabetes for the genotypes of other people in your account.

The 23andMe Odds Calculator only takes into account effects of markers with known associations that are also on our genotyping chip. Keep in mind that aside from genetics, environment and lifestyle may also contribute to one’s risk for Type 2 Diabetes.

Genes vs. Environment

The heritability of type 2 diabetes is estimated to be 26%. This means that environmental factors contribute more to differences in risk for this condition than genetic factors. Genetic factors that play a role in type 2 diabetes include both unknown factors and known factors such as the SNPs we describe here. Environmental factors include obesity, gestational diabetes, giving birth to at least one baby weighing nine pounds or more, high blood pressure, abnormal cholesterol levels, physical inactivity, polycystic ovarian syndrome, other clinical conditions associated with insulin resistance, a history of impaired glucose tolerance or impaired fasting glucose, and a history of cardiovascular disease. (sources)
23andMe Type 2 Diabetes Risks

Marker Effects

What does this chart show?
The chart shows the approximate effects of the selected person's genotype at the 11 reported markers. Higher, red bars indicate increased risk from the average, while lower, green bars indicate decreased risk from the average. The light gray bars show the maximum possible effects for the possible genotypes at the marker.

Mouse over individual bars to view additional information about each marker. Click on a bar to view detailed information about that marker below. You can read more about all markers in the technical report.

TCF7L2

Marker: rs7903146

This SNP is located in the TCF7L2 gene, which encodes a protein involved in cell signalling. How TCF7L2 affects the development of type 2 diabetes is not completely understood. TCF7L2 has been shown to be involved in the development of pancreatic islets, which contain insulin-producing beta cells. Studies suggest that the T version of this SNP is associated with impaired baseline insulin secretion.

The T version of this SNP is also associated with increased odds of gestational diabetes, a form of diabetes that occurs only during pregnancy. Gestational diabetes can lead to complications for both mother—such as difficult delivery due to unusually large infant size—and baby, such as low blood sugar and breathing problems.

Citations


**MTNR1B**

This SNP is located near the MTNR1B gene, which encodes a pancreatic beta cell protein that interacts with a hormone called melatonin. In healthy individuals, insulin secretion follows a circadian rhythm with peaks during the day and troughs at night. Melatonin levels have the opposite pattern being highest during the night and thus may inhibit insulin secretion, possibly through the MTNR1B protein. Studies have shown that melatonin receptors like MTNR1B are overexpressed in pancreatic islets of individuals with type 2 diabetes compared to non-diabetic individuals.

Multiple studies have confirmed this association in populations with European ancestry. This association has not been studied in Asian or African populations.

**Citations**

# 23andMe Carrier Status

## Carrier Status

**Show results for** [Douglas Brutlag](#)  
**See new and recently updated reports** »

*23andWe Discoveries were made possible by 23andMe members who took surveys.*

<table>
<thead>
<tr>
<th>Name</th>
<th>Confidence</th>
<th>Confidence</th>
<th>Status</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alpha-1 Antitrypsin Deficiency</td>
<td>⭐⭐⭐⭐⭐</td>
<td></td>
<td>Variant Absent</td>
</tr>
<tr>
<td>Bloom’s Syndrome</td>
<td>⭐⭐⭐⭐⭐</td>
<td></td>
<td>Variant Absent</td>
</tr>
<tr>
<td>BRCA Cancer Mutations (Selected)</td>
<td>⭐⭐⭐⭐⭐</td>
<td></td>
<td>Variant Absent</td>
</tr>
<tr>
<td>Canavan Disease</td>
<td>⭐⭐⭐⭐⭐</td>
<td></td>
<td>Variant Absent</td>
</tr>
<tr>
<td>Cystic Fibrosis</td>
<td>⭐⭐⭐⭐⭐</td>
<td></td>
<td>Variant Absent</td>
</tr>
<tr>
<td>DPD Deficiency</td>
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<td></td>
<td>Variant Absent</td>
</tr>
<tr>
<td>Familial Dysautonomia</td>
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<td></td>
<td>Variant Absent</td>
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<tr>
<td>Factor XI Deficiency</td>
<td>⭐⭐⭐⭐⭐</td>
<td></td>
<td>Variant Absent</td>
</tr>
<tr>
<td>Fanconi Anemia (FANCC-related)</td>
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</tr>
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<tr>
<td>Familial Mediterranean Fever</td>
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<tr>
<td>G6PD Deficiency</td>
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</tr>
<tr>
<td>Gaucher Disease</td>
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<td></td>
<td>Variant Absent</td>
</tr>
<tr>
<td>Glycogen Storage Disease Type 1a</td>
<td>⭐⭐⭐⭐⭐</td>
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<td>Variant Absent</td>
</tr>
<tr>
<td>Hemochromatosis (HFE-related)</td>
<td>⭐⭐⭐⭐⭐</td>
<td></td>
<td>Variant Absent</td>
</tr>
<tr>
<td>Limb-girdle Muscular Dystrophy</td>
<td>⭐⭐⭐⭐⭐</td>
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<tr>
<td>Maple Syrup Urine Disease Type 1B</td>
<td>⭐⭐⭐⭐⭐</td>
<td></td>
<td>Variant Absent</td>
</tr>
</tbody>
</table>

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23andMe Carrier Status for Alpha-1 Antitrypsin Deficiency

Alpha-1 Antitrypsin Deficiency

The alpha-1 antitrypsin (AAT) protein protects the body, especially fragile lung tissues, from the damaging effects of a powerful enzyme called neutrophil elastase that is released from white blood cells. In AAT deficiency, a genetic mutation reduces levels of the protective protein in the bloodstream. AAT deficiency can lead to chronic obstructive pulmonary disease (COPD), specifically emphysema, and liver disease. Smoking, which can inhibit what little AAT protein an affected person does have, increases the risk of lung disease.

The following results are based on 4★☆☆☆ Established Research for 2 reported markers.

Learn more about the biology of Alpha-1 Antitrypsin Deficiency...
## 23andMe Drug Responses

### drug response

Show results for **Douglas Brutlag**

23andWe Discoveries were made possible by 23andMe members who took surveys.

<table>
<thead>
<tr>
<th>Name</th>
<th>Confidence</th>
<th>Status</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clopidogrel (Plavix®) Efficacy</td>
<td>⭐️⭐️⭐️⭐️⭐️</td>
<td>Greatly Reduced</td>
</tr>
<tr>
<td>Abacavir Hypersensitivity</td>
<td>⭐️⭐️⭐️⭐️⭐️</td>
<td>Typical</td>
</tr>
<tr>
<td>Alcohol Consumption, Smoking and Risk of Esophageal Cancer</td>
<td>⭐️⭐️⭐️⭐️⭐️</td>
<td>Typical</td>
</tr>
<tr>
<td>Fluorouracil Toxicity</td>
<td>⭐️⭐️⭐️⭐️⭐️</td>
<td>Typical</td>
</tr>
<tr>
<td>Response to Hepatitis C Treatment</td>
<td>⭐️⭐️⭐️⭐️⭐️</td>
<td>Typical</td>
</tr>
<tr>
<td>Pseudocholinesterase Deficiency</td>
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<td>Typical</td>
</tr>
<tr>
<td>Thiopurine Methyltransferase Deficiency</td>
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<td>Typical</td>
</tr>
<tr>
<td>Warfarin (Coumadin®) Sensitivity</td>
<td>⭐️⭐️⭐️⭐️⭐️</td>
<td>Typical</td>
</tr>
<tr>
<td>Oral Contraceptives, Hormone Replacement Therapy and Risk of Venous Thromboembolism</td>
<td>⭐️⭐️⭐️⭐️⭐️</td>
<td>Not Applicable</td>
</tr>
<tr>
<td>Caffeine Metabolism</td>
<td>⭐️⭐️⭐️</td>
<td>Fast Metabolizer</td>
</tr>
<tr>
<td>Hepatitis C Treatment Side Effects</td>
<td>⭐️⭐️⭐️</td>
<td>See Report</td>
</tr>
<tr>
<td>Metformin Response</td>
<td>⭐️⭐️⭐️</td>
<td>Typical Odds of Positive Response</td>
</tr>
<tr>
<td>Antidepressant Response</td>
<td>⭐⭐</td>
<td>See Report</td>
</tr>
</tbody>
</table>

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drug response

Clopidogrel (Plavix®) Efficacy ★★★★

Established Research report on 5 reported markers.

Only a medical professional can determine whether clopidogrel is the right medication for a particular patient. The information contained in this report should not be used to independently establish a clopidogrel regimen, or abolish or adjust an existing course of treatment.

About Clopidogrel Efficacy

Clopidogrel (sold under the trade names Plavix®, Iscover®, Clopilet® and Ceruvin®) is a drug commonly prescribed in combination with aspirin to help prevent blood clots that can block blood flow and cause a heart attack or stroke. However, clopidogrel doesn't inhibit clotting to the same extent in everyone. For some people, genetic variations that prevent the drug from being converted into its active form in the body are the cause. Studies have shown that people who are taking clopidogrel who have these genetic variations may have reduced protection from heart attacks, strokes and death from cardiovascular causes.

Learn more about the biology of Clopidogrel Efficacy...
Plavix Ad with Genetic Requirement
# 23andMe Traits

## Show results for Douglas Brutlag

*23andMe Discoveries were made possible by 23andMe members who took surveys.*

<table>
<thead>
<tr>
<th>Name</th>
<th>Confidence</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alcohol Flush Reaction</td>
<td>★★★★☆☆☆☆☆</td>
<td>Does Not Flush</td>
</tr>
<tr>
<td>Bitter Taste Perception</td>
<td>★★★★☆☆☆☆☆</td>
<td>Can Taste</td>
</tr>
<tr>
<td>Earwax Type</td>
<td>★★★★☆☆☆☆☆</td>
<td>Wet</td>
</tr>
<tr>
<td>Eye Color</td>
<td>★★★★☆☆☆☆☆</td>
<td>Likely Brown</td>
</tr>
<tr>
<td>Hair Curl</td>
<td>★★★★☆☆☆☆☆</td>
<td>Straighter Hair on Average</td>
</tr>
<tr>
<td>Lactose Intolerance</td>
<td>★★★★☆☆☆☆☆</td>
<td>Likely Tolerant</td>
</tr>
<tr>
<td>Malaria Resistance (Duffy Antigen)</td>
<td>★★★★☆☆☆☆☆</td>
<td>Not Resistant</td>
</tr>
<tr>
<td>Male Pattern Baldness</td>
<td>★★★★☆☆☆☆☆</td>
<td>Decreased Odds</td>
</tr>
<tr>
<td>Muscle Performance</td>
<td>★★★★☆☆☆☆☆</td>
<td>Likely Sprinter</td>
</tr>
<tr>
<td>Non-ABO Blood Groups</td>
<td>★★★★☆☆☆☆☆</td>
<td>See Report</td>
</tr>
<tr>
<td>Norovirus Resistance</td>
<td>★★★★☆☆☆☆☆</td>
<td>Not Resistant</td>
</tr>
<tr>
<td>Resistance to HIV/AIDS</td>
<td>★★★★☆☆☆☆☆</td>
<td>Not Resistant</td>
</tr>
<tr>
<td>Smoking Behavior</td>
<td>★★★★☆☆☆☆☆</td>
<td>Typical</td>
</tr>
<tr>
<td>Adiponectin Levels</td>
<td>★★★☆☆☆☆☆☆☆</td>
<td>See Report</td>
</tr>
<tr>
<td>Asparagus Metabolite Detection</td>
<td>★★★☆☆☆☆☆☆☆</td>
<td>Typical Odds of Detecting</td>
</tr>
<tr>
<td>Birth Weight</td>
<td>★★★☆☆☆☆☆☆☆</td>
<td>See Report</td>
</tr>
<tr>
<td>Blood Glucose</td>
<td>★★★☆☆☆☆☆☆☆</td>
<td>5.18 mmol/L on Average</td>
</tr>
<tr>
<td>Breastfeeding and IQ</td>
<td>★★★☆☆☆☆☆☆☆</td>
<td>See Report</td>
</tr>
<tr>
<td>C-reactive Protein Level</td>
<td>★★★☆☆☆☆☆☆☆</td>
<td>2.09 mg/L on Average</td>
</tr>
</tbody>
</table>
Choice of GWAS Studies

• Common traits of broad interest
  – Prevalence of > 1%
  – Report Mendelian traits when possible
  – Focus on drug responses

• Avoid false discoveries
  – Large case-control studies > 750 cases
  – Highly significant expectation values (<0.01 errors)
  – Published in reputable journals
  – Studies that have been replicated

• May impute highly linked missing SNPs

• Calculate likelihood and odds ratio using customers ethnicity as detected

• Distinguish preliminary studies (non-replicated or smaller sample sizes) from established research.
Maternal Haplogroup: U5b2a

U5b2a is a subgroup of U5, which is described below.

Locations of haplogroup U5 circa 500 years ago, before the era of intercontinental travel.

Haplogroup U5 arose among early colonizers of Europe around 40,000 years ago; maternal descendants of those early colonizers persist in the region to this day. After the last ice Age two subgroups of U5 expanded across Europe and into northern Africa and the Near East. Today, one subgroup, U5b1b, is shared by groups as diverse as the northern African desert-dwelling Berbers and the Scandinavian Arctic-dwelling Saami, also known as the Lapps.

Human Prehistory Videos

Human Prehistory: Prologue

Out of (Eastern) Africa

Haplogroup: U5, a subgroup of R
Age: 40,000 years
Region: Europe, Near East, North Africa
Populations: Basques, Saami (Lapps) of northern Scandinavia
Highlight: Though primarily a European haplogroup, U5 was recently found in mitochondrial DNA extracted from the remains of a 6th-century AD Chinese chieftain.

Your Family and Friends

A2, Samantha Hill
D4e2, Japanese Person
D5a1a, Chinese Person
H3, Lily Mendel (Mom), Erin Mendel (Daughter), Alan Mendel (Son), Ian Mendel (Son), Margo Fisher (Grandma)
H4a1, Ron Fisher (Grandpa)
K1a1b1a, Benjamin Brutlag, Pauline Brutlag, Simone Brutlag
L3e2b2, Nigerian Person
M35b, renu heller

© Doug Brutlag 2015
Paternal Haplogroup: E1b1b1a2*

E1b1b1a2* is a subgroup of E1b1b1a, which is described below.

Locations of haplogroup E1b1b1a circa 500 years ago, before the era of intercontinental travel.

E1b1b1a is most common in northern Africa and southern Europe. It arose about 23,000 years ago in eastern Africa and spread into the Mediterranean region after the Ice Age. E1b1b1a, a subgroup of E1b1b, expanded out of the Near East 8,000 years ago into northern Africa and southern Europe. Today it is one of the most common haplogroups in those regions.
ancestry overview

Your Father's Line
Your father's line was likely in eastern Africa 50,000 years ago. Today that line is still found primarily in Africa.

Neanderthal Ancestry
You have an estimated 2.5% Neanderthal DNA, which puts you in the 39th percentile among Northern European 23andMe members.

Your Mother's Line
Along your mother's line, you have ancestry in Europe/the Near East. In the past few hundred years, that traces back to eastern Africa around 50,000 years ago.

Your Extended DNA Family
Guess what? If you have a large piece of identical DNA in common with someone, then you're related. You have 505 DNA relatives in 23andMe. Explore their info to learn more about your own ancestry.

Close Family
1st-2nd 3rd-4th Distant Cousins Cousins cousins

2 0 60 443

From Your Ancestry Expert
It's remarkable what you can discover from a little saliva. On this page are the highlights of what we've learned about your ancestry, based just on your DNA. Enjoy!

Dr. Joanna Mountain, PhD
Joanna Mountain is 23andMe's Senior Director of Research. A former Stanford professor, she has traveled the world studying genetics and human history.

AS SEEN ON

ANDERSON

Ancestry Help

Send Feedback

Top Relative Surnames

<table>
<thead>
<tr>
<th>Surname</th>
<th>Count</th>
<th>Enrichment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anderson</td>
<td>5</td>
<td>10</td>
</tr>
<tr>
<td>Smith</td>
<td>5</td>
<td>1</td>
</tr>
</tbody>
</table>
Ancestry Composition

Ancestry Composition tells you what percent of your DNA comes from each of 22 populations worldwide. The analysis includes DNA you received from all of your ancestors, on both sides of your family. The results reflect where your ancestors lived 500 years ago, before ocean-crossing ships and airplanes came on the scene.

- 99.9% European
- < 0.1% East Asian & Native American
- 0.1% Unassigned

100.0% Douglas Brutlag

show all populations
Ancestry Composition

Ancestry Composition tells you what percent of your DNA comes from each of 22 populations worldwide. The analysis includes DNA you received from all of your ancestors, on both sides of your family. The results reflect where your ancestors lived 500 years ago, before ocean-crossing ships and airplanes came on the scene.

- **99.9%** European
- **0.1%** East Asian & Native American

100.0% Douglas Brutlag

[Show all populations]
History of Man

Human Prehistory 101

Prologue
<table>
<thead>
<tr>
<th>Male</th>
<th>You</th>
<th>U5b...</th>
<th>E1b1b1...</th>
</tr>
</thead>
<tbody>
<tr>
<td>Benjamin Brutlag</td>
<td>Son</td>
<td>United Sta...</td>
<td>Southern Euro...</td>
</tr>
<tr>
<td>Male, b. 1980</td>
<td>47.7% shared, 22 segments</td>
<td>K1a1b...</td>
<td>E1b1b1...</td>
</tr>
<tr>
<td>Pauline Brutlag</td>
<td>Daughter</td>
<td>United Sta...</td>
<td>Northern Euro...</td>
</tr>
<tr>
<td>Female</td>
<td>53.1% shared, 25 segments</td>
<td>K1a1b...</td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>3rd to 4th Cousin</td>
<td>...</td>
<td>I2...</td>
</tr>
<tr>
<td>Male</td>
<td>3rd to 5th Cousin</td>
<td>...</td>
<td>R1a...</td>
</tr>
<tr>
<td>Larry Vongroven</td>
<td>3rd to 5th Cousin</td>
<td>United Sta...</td>
<td>Aien, Nor...</td>
</tr>
<tr>
<td>Male</td>
<td>0.54% shared, 2 segments</td>
<td>Haltalen, Nor...</td>
<td>Voss, Nor...</td>
</tr>
<tr>
<td></td>
<td></td>
<td>8 m...</td>
<td>Northern Euro...</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Vongroven (Vongrav...</td>
<td>Bakk...</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Good...</td>
<td>11 m...</td>
</tr>
<tr>
<td></td>
<td></td>
<td>R1a...</td>
<td></td>
</tr>
<tr>
<td>Carolyn Otterness</td>
<td>3rd to 5th Cousin</td>
<td>United Sta...</td>
<td></td>
</tr>
<tr>
<td>Female, b. 1941</td>
<td>0.47% shared, 2 segments</td>
<td>Otsego, Wisconsin, Dodge County, C...</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Northern Euro...</td>
<td>Ottern...</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Brandsn...</td>
<td>Gjorne...</td>
</tr>
<tr>
<td></td>
<td></td>
<td>5 m...</td>
<td></td>
</tr>
<tr>
<td>Gale Enger</td>
<td>3rd to 5th Cousin</td>
<td>United Sta...</td>
<td></td>
</tr>
<tr>
<td>Male, b. 1925</td>
<td>0.41% shared, 2 segments</td>
<td>Norway, Denmark, Minnesota, Wisco...</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Northern Euro...</td>
<td>En...</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Northern Euro...</td>
<td>Lars...</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Mest...</td>
<td>6 m...</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
23andMe Relative Finder Map View

relative finder

Search your matches

Total results: 193

Top Locations
- California, USA (7)
- Germany (6)
- Chicago, IL, USA (5)
- Virginia, USA (5)
- Norway (5)
- Poland (3)
- Pennsylvania, USA (3)
- Peoria, IL, USA (3)

Jump to Region
- United States
- North America
- South America
- Europe
- Africa
- Asia
- Eastern Hemisphere

Clustering: Off On
What is a Fifth Cousin?

So You’re
### 23andMe Ancestry Finder

<table>
<thead>
<tr>
<th>Country</th>
<th>Color</th>
<th>Percent of Douglas Brutlag's Genome Covered</th>
</tr>
</thead>
<tbody>
<tr>
<td>Norway</td>
<td></td>
<td>3.6% – 5.4%</td>
</tr>
<tr>
<td>Germany</td>
<td></td>
<td>1.3% – 4.2%</td>
</tr>
<tr>
<td>Ireland</td>
<td></td>
<td>0.7% – 1.4%</td>
</tr>
<tr>
<td>Sweden</td>
<td></td>
<td>0.5% – 1.2%</td>
</tr>
<tr>
<td>Denmark</td>
<td></td>
<td>0.4% – 1.7%</td>
</tr>
<tr>
<td>Russia</td>
<td></td>
<td>0.4% – 0.8%</td>
</tr>
<tr>
<td>Netherlands</td>
<td></td>
<td>0.3% – 0.7%</td>
</tr>
<tr>
<td>Finland</td>
<td></td>
<td>0.3% – 0.7%</td>
</tr>
</tbody>
</table>

The image also shows a visual representation of DNA segments for each chromosome, which corresponds to the countries and their percentages.
23andWe discoveries

You answer questions. + Other 23andMe members answer questions. + 23andMe scientists work their magic. = 23andWe discoveries

23andWe community contributions

Asparagus Metabolite Detection
This report is based on "Ten Things About You".
View this report

Freckling
This report is based on "Pigmentation".
View this report

Hair Curl
This report is based on "Ten Things About You".
View this report

Photic Sneeze
This report is based on "Ten Things About You".
View this report
research initiatives

A new paradigm for genetic research.

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.

23andWe involves our customers in research as collaborators, advisers and contributors by conducting studies that correlate their responses to online surveys with their genetic data. The idea is to enable large studies that would be infeasible using current methods, which typically involve recruiting patients through physicians' practices and other means. We plan to share the results of our research and show you how your contributions are making an impact by posting regular updates on this website.

Next: How does research work at 23andMe?

Join a research community

Parkinson's Disease
Recent discoveries suggest that genetics plays a greater role in Parkinson's disease than was previously thought. You can advance research into the genetic roots of Parkinson's disease.

Alzheimer's Disease
More than 5 million Americans have Alzheimer's Disease. 23andMe and Genentech have teamed up to find out how genetics might protect against Alzheimer's Disease. This research could lead to new scientific knowledge or possibly a drug that could prevent or slow Alzheimer's Disease.
INFORMED Genetic Counselors

About InformedDNA

Our nationwide network of board-certified genetic counselors provide genetic expertise to patients, physicians, and organizations across all fifty states in the USA, and are available internationally.

Genetic Expertise

- Cancer Genetics
- Reproductive Genetics
- Cardiac Genetics
- Ocular Genetics
- Neurogenetics
- Adult Genetics

Access to Experts

- Convenient Accessible Scheduling
- Ample Appointment Availability
- Insurance Authorization
- Genetic Test Coordination
- Expert Test Interpretation
- Personalized Healthcare Reports

REFER A PATIENT

REQUEST YOUR APPOINTMENT
About Genetic Counseling

Genetic counseling can benefit individuals with a family history of an inherited condition, individuals who are pregnant or considering a pregnancy, and individuals with a family history of cancer. Learn more about genetic counseling, please visit:

- Connecting Patients to the Power of Genetics
- Our online library
- Genetics and genetic testing
- Become familiar with our counseling process
- Visit our learning center

REQUEST YOUR APPOINTMENT ➔ REFER A PATIENT ➔
Specialty Genetic Services

Our unique nationwide team of telephone-based genetics experts sub-specialize to ensure that you are working with a genetics professional who is expertly trained in the area of concern for you or your patients. Our specialty disciplines include cancer, cardiac, reproductive, ocular, pediatric, neurogenetics, pharmacogenetics, and genomic medicine.

For Patients
- SCHEDULE APPOINTMENT

For Providers
- REFER A PATIENT

For Health Plans
- OPTIMIZE YOUR BENEFITS
A new look at a healthier future

Your family history may play an important role in your genetic makeup, but your DNA is unique. By understanding your genetic predispositions, you can start looking at your health in a new way. You can also learn if certain medications work with your genetic makeup.

Myth:
Most people don’t have any genetic mutations, so why bother?

Truth:
Everyone has some genetic mutations, even people who live to 110. What’s important is the specific nature of your own DNA and how that correlates to the likelihood of developing a specific condition. Navigenics will give you that information.

Understand your DNA
Learn about DNA, how it affects your health, and how genetic testing reveals the answers your DNA holds.

Get a more complete picture
Learn how family history and your DNA insights can give you a comprehensive view of your health.

Start a personalized approach
Find out how your genetic test results can help point you toward better health and well-being.
We care about the science, your patients, and you

Make personalized genomic medicine and pharmacogenomics part of your practice. And provide your patients with a powerful tool for change.

Your patients trust you; you can trust us.

- Founders, practicing physician David Agus, M.D., and geneticist Dietrich Stephan, Ph.D., came together so that they could create a powerful new tool for personalized medicine.
- Focused on prevention, pharmacogenomics, and longitudinal health outcome studies.

We can help answer your questions.

- Medical education programs, resources and board-certified genetic counseling.
- Specifics on our Medications Wallet Card, including background information on each medication result presented on the card.

Partner with the leader in genomic health, just as we partner with the leaders in medicine.

- We collaborate with Mayo Clinic, Scripps Genomic Medicine, Duke, and others.
Conditions and medication responses

Navigenics analyzes your DNA for genetic risk markers associated with a wide variety of important health conditions and medication responses

Health Conditions

- Abdominal aneurysm
- Alzheimer's disease
- Atrial fibrillation
- Brain aneurysm
- Breast cancer
- Celiac disease
- Colon cancer
- Crohn's disease
- Deep vein thrombosis
- Diabetes, type 2
- Glaucoma
- Graves' disease
- Heart attack
- Hemochromatosis, HFE-related
- Lactose intolerance
- Lung cancer
- Lupus
- Macular degeneration
- Melanoma
- Multiple sclerosis
- Obesity
- Osteoarthritis
- Prostate cancer
- Psoriasis
- Restless legs syndrome
- Rheumatoid arthritis
- Sarcoidosis
- Stomach cancer, diffuse

Medications

- Abacavir
- Beta blockers
- Carbamazepine
- Clopidogrel
- Floxacillin
- Fluorouracil
- Irinotecan
- Simvastatin
- Statins
- Succinylcholine
- Thiopurines
- Warfarin

Success story:

"I think it's important to know as much as you can, so you can make decisions that will enable you to control your life, how long you're going to live, and especially what the quality of your life is."
## Health Conditions

### Your estimated lifetime risk

Click anywhere on the colored boxes below to access in-depth information about each health condition, your genetic predispositions, what you can do, your specific genetic markers, and much more.

<table>
<thead>
<tr>
<th>Condition</th>
<th>You</th>
<th>Avg</th>
<th>Population Average</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Diabetes type 2</strong></td>
<td></td>
<td></td>
<td>45%</td>
</tr>
<tr>
<td><strong>Brain aneurysm</strong></td>
<td>0.91%</td>
<td>0.90%</td>
<td>1.3%</td>
</tr>
<tr>
<td><strong>Alzheimer's disease</strong></td>
<td>10%</td>
<td>17%</td>
<td>4%</td>
</tr>
<tr>
<td><strong>Breast cancer</strong></td>
<td>20%</td>
<td>13%</td>
<td>1%</td>
</tr>
<tr>
<td><strong>Diabetes, type 2</strong></td>
<td>36%</td>
<td>30%</td>
<td>10%</td>
</tr>
<tr>
<td><strong>Macular degeneration</strong></td>
<td>0.78%</td>
<td>3.1%</td>
<td>1.7%</td>
</tr>
<tr>
<td><strong>Lung cancer</strong></td>
<td>5%</td>
<td>6%</td>
<td>1%</td>
</tr>
<tr>
<td><strong>Atrial fibrillation</strong></td>
<td>10%</td>
<td>23%</td>
<td>3%</td>
</tr>
<tr>
<td><strong>Heart attack</strong></td>
<td>26%</td>
<td>25%</td>
<td>5%</td>
</tr>
<tr>
<td><strong>Sarcoidosis</strong></td>
<td>0.55%</td>
<td>1.0%</td>
<td>0.2%</td>
</tr>
<tr>
<td><strong>Deep vein thrombosis</strong></td>
<td>4.2%</td>
<td>3.6%</td>
<td>2%</td>
</tr>
<tr>
<td><strong>Osteoarthritis</strong></td>
<td>26%</td>
<td>47%</td>
<td>20%</td>
</tr>
<tr>
<td><strong>Glaucoma</strong></td>
<td>0.47%</td>
<td>2.4%</td>
<td>5%</td>
</tr>
<tr>
<td><strong>Colon cancer</strong></td>
<td>4.1%</td>
<td>5%</td>
<td>1%</td>
</tr>
<tr>
<td><strong>Obesity</strong></td>
<td>25%</td>
<td>32%</td>
<td>15%</td>
</tr>
</tbody>
</table>

*Note: Key to your results:*

- **Diabetes type 2**
  - You: 45%
  - Avg: 36%
  - Population Average: 10%
  - Why orange & gray boxes?
  - Video: Understanding your results
  - Tutorial: Review the tutorial

*Printable report*

You can print or save a PDF version of this report for your personal use.

Displaying 6 of 13 total results.
# How medications affect you

Welcome to your Medications results! This new Navigenics feature provides personalized genetic information to help you understand which drugs work best for you, starting with your responses to 12 medications. Many of these medications, such as statins, are taken by millions of people each day.

Click on the name of any medication below to access in-depth information about your risk of side effects or the drug’s effectiveness for you. You’ll also learn what you can do, details about your specific genetic markers, what your results may mean for your family, and much more.

Even if your results for particular medications appear low risk or typical, this information is still helpful. Should you ever need one of these drugs, you and your doctor can use your genetic results to help make medication decisions tailored for you.

You can also click on the Health Conditions tab above to see your genetic results for important health risks.

## Side Effects

<table>
<thead>
<tr>
<th>Drug</th>
<th>Your Risk</th>
<th>Side Effect</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fluorouracil (Efudex®)</td>
<td>High Risk</td>
<td>Severe, potentially fatal toxicity</td>
</tr>
<tr>
<td>Used to treat many types of cancer</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Simvasatin (Vytorin®, Zocor®)</td>
<td>Moderate Risk</td>
<td>Muscle pain and damage</td>
</tr>
<tr>
<td>Used to treat high cholesterol and help prevent heart disease</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Abacavir (Ziagen®)</td>
<td>Low Risk</td>
<td>Severe allergic reaction, including fever, rash, and nausea</td>
</tr>
<tr>
<td>Used to treat HIV infection</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
The End of Illness  David B. Agus

#1 NEW YORK TIMES BESTSELLER

THE END of ILLNESS

$12 download on Amazon

David B. Agus, MD
Navigenics Conditions Covered

Conditions we cover
Navigenics analyzes your DNA for genetic risk markers associated with a wide variety of important health conditions

- Abdominal aneurysm
- Alzheimer's disease
- Atrial fibrillation
- Brain aneurysm
- Breast cancer
- Celiac disease
- Colon cancer
- Crohn's disease
- Deep vein thrombosis
- Diabetes, type 2
- Glaucoma
- Graves' disease
- Heart attack
- Hemochromatosis
- Lactose intolerance
- Lung cancer
- Lupus
- Macular degeneration
- Melanoma
- Multiple sclerosis
- Obesity
- Osteoarthritis
- Prostate cancer
- Psoriasis
- Restless legs syndrome
- Rheumatoid arthritis
- Sarcoidosis
- Stomach cancer, diffuse

A health investment that continues to grow
Navigenics Health Compass is an extensive, health-focused set of genetic testing services, offering genetic insights now and in the future. Our testing platform includes all of the conditions listed above, and then enables ongoing updates to bring you new genetic results for as long as you subscribe. At any step in the process, you'll be able to consult with our board-certified Genetic Counselors, health professionals who can help you understand your results and decide on next steps.

Genetic science continues to advance at a rapid rate. As new links between genetic markers and health are discovered, we add new conditions to our genetic testing services. It's a key benefit of Health Compass: Your genetic test results will be continually updated with new health information as long as you subscribe.

Learn more about the genetic testing services offered with Navigenics Health Compass >

How we choose
We use rigorous standards for deciding which health conditions to include in your genetic test, and we focus on conditions that you can do something about. To be added to our genetic testing services, a health condition and the science behind it must meet our strict guidelines:
DNA Direct brings the power of personalized medicine to payors, providers and patients.

- **The Right Person**
  Finding the right people to benefit from genomic medicine can improve disease management and lower healthcare costs.

- **The Right Test**
  Getting the wrong test can misinform medical decisions and increase healthcare costs.

- **The Right Interpretation**
  Delivers the full value of genetic information and enables physicians to make appropriate management decisions.

### Hospital Plan Webinar
**Strategies to Optimize Personalize Medicine: How to Integrate Genomic Services into Your Hospital Community**
Dr. Derek Kelly, Vice President, Medical Management at Swedish Covenant Hospital in Chicago discusses integrating genomic services into their clinical care.

### Health Plan Webinar
**How a Health Plan Successfully Integrated Genomic Services into Its System**
Dr. Charles Stemple, Medical Director, Personalized Medicine/Genomics at Humana discusses their genetic guidance program.

http://www.dnadirect.com/
About Personalized Medicine

Personalized medicine, also referred to as genomic medicine, is changing the landscape of healthcare. By harnessing the power of genetic testing, physicians can make more informed healthcare decisions and better target treatments and drug therapies. The result is better healthcare outcomes.

Genetic tests are used in all areas of medicine – from prevention and screening to diagnosis and treatment. G2 Intelligence estimated that the market was $14.3B in 2010 and growing rapidly at 16% per year¹ and the Food and Drug Administration (FDA) states that more than 100 medications have pharmacogenomic information included in their drug labels². Research by the Tufts Center for the Study of Drug Development indicates that oncology leads other therapeutic areas in the number of targeted therapies on the market as well as in the pipeline, with the expectation that within the decade all oncology drugs will have a related diagnostic. Other key therapeutic areas in which personalized medicine is impacting clinical decision-making include cardiovascular, neurologic, and immunologic therapies, whereas personalized medicine development is just getting started for metabolic and respiratory therapies, as well as virology³. With the advent of all of this new technology and information available to healthcare professionals and consumers, it will be critical to stay abreast of the new developments.

Low-cost whole genome sequencing (WGS) is on the horizon as well, adding a profound new dimension to the personalized medicine arsenal. Healthcare providers and consumers will be challenged with how best to interpret the information available to them.

As advances in personalized medicine continue, patients benefit from the deeper knowledge that genomics brings to healthcare decision making and outcomes.

¹. G2 Intelligence: Lab Industry Strategic Outlook 2011; Market Trends & Analysis
². www.fda.gov/drugs/scienceresearch/researchareas/pharmacogenetics/ucm083378.htm
Complete Genomics
http://www.completegenomics.com/

Accurate Whole Human Genome Sequencing & Analysis

Complete Genomics is a leader in accurate whole human genomic sequencing. Using our proprietary sequencing instruments, chemistry, and software, we have sequenced more than 15,000 whole human genomes for our research customers over the past three years. Our mission is to provide the technology for sequencing one million human genomes, enabling researchers and clinicians to improve human health through the prevention, diagnosis, and treatment of genetic diseases and conditions.
Personalis
http://www.personalis.com/
StationX
http://www.stationxinc.com/

Genomes
Decoded and Delivered

Got DNA?
Learn how we make your clinical R&D more efficient →

Software and services that simplify the analysis and visualization of genome-scale data in clinical research and development. →
70% of diseases are preventable.

BaseHealth™ makes an integrated health management platform called Genophen. We leverage the broad appeal of genomics to engage patients with their physicians in a highly-collaborative way.
Personal Genomics References


Gene-ius.
A smart way to look at your health.

Navigenics is the leading provider of clinically guided genetic analysis. Our goal is to empower you with genetic insights to help motivate you to improve your health. We also put a premium on privacy, keeping you in control of your genetic information.

New: Your genes, your medications
Will a new medication be effective for you? Will a treatment cause serious side effects? Now, genetic insights from Navigenics can help you and your doctor select medications that may be right for your genetic makeup.

Find a physician
Find a physician in your area who offers the Navigenics genetic testing services, so you can focus your health plan on prevention.

Next Steps
- I'm new to Navigenics
- Adding to family history
- Genetic testing: Myths and truths
- Genetic knowledge can help you

For Physicians
- Free educational webinars
- More personalized care
- Genetic counselors for patients and you
- Foundation that rests on strong science

Our Collaborators
- PositScience®
- Scripps Health Foundation

Latest Headlines
Video: Dr. Vanier joined by Dr. Eric Topol to discuss NEJM study data
DNA Basics

What's in your DNA? Each of us is born with our own genetic code, a unique set of instructions stored inside our cells. These instructions tell our bodies how to function over the course of our lifetimes. Learn more >

What are genetic markers? Think of genetic risk markers as bits of DNA that vary from person to person. These markers are part of what makes each of us unique. They also reveal patterns in your DNA that relate to certain health conditions and medication responses. Learn more >

How do we look at your DNA? Genetic science has come a long way in recent years. Now, genetic tests based on a simple saliva sample can tell you about key points in your genetic code and how they affect your risk of many health conditions. Learn more >

Is your DNA your destiny? Most of your genes work together with the environment around you, including your behaviors and the influences you are exposed to on a regular basis. That means the choices you make can shape how much your genes affect your health. Learn more >
Family history

How DNA is inherited: Each of your parents gave you half your DNA, and your children receive half of yours. Which half? That's completely unpredictable. Learn more >

Adding to family history: Family history can be an important piece of your health puzzle. Adding genetic information to that knowledge gives us a powerful combination for understanding our health. Learn more >

What if I don't know my family history? Many of us are adopted. Others have lost contact with a parent. Gaps in family history leave many of us without important information about our health, but now genetic testing can bring essential knowledge to light. Learn more >

Completing your health picture: The knowledge you gain through genetic analysis does more than add to family history. It can also sharpen your awareness of health risks you already thought you understood. Learn more >
A new way to look at your health

**Personalized prevention:** We are surrounded by more health advice than ever. With insights from your genetic analysis, you and your doctor can consider which health measures and prescription medications are right for you. Learn more >

**Working with your doctor:** When you and your doctor examine your DNA results together with your family history, medical history, and lifestyle, your health picture becomes more complete. Find out how Navigenics can help you and your doctor customize your preventive strategies and medication choices. Learn more >

**Truly personalized health:** Your genetic results can help you consider more personalized ways to stay healthy, helping you make the difference between your DNA and your destiny. Learn more >

Taking action with your results
Navigenics Compass Program

Comprehensive genetic testing services: $999

Knowing your genetic predispositions for important health conditions is an investment in your health. By gaining insight into these predispositions, you have the power to help delay the onset of conditions, detect them earlier, or prevent them altogether.

Our genetic testing services uncover your genetic predispositions for important health conditions and give you resources to take action and build a lifetime of better health.

When you order Navigenics Health Compass, you will receive:

- An analysis of your genetic predispositions for a variety of health conditions that meet stringent scientific criteria.
- A customized test for each genetic marker associated with the select health conditions, carried out by a CLIA-certified laboratory that complies with federal regulations.
- Access to our board-certified Genetic Counselors to help explain what your genetic test results mean and support you in knowing how to take next steps.
- A health investment that continues to grow, with ongoing, personalized updates for an entire year, bringing you new health condition predispositions, genetic risk markers, clinical therapies and wellness strategies.
- Easy-to-use, relevant health information, developed by our physicians and the Mayo Clinic to help you understand each health condition and know what to do next.

Order today! After your first year, you can opt to maintain an ongoing subscription at the rate of $199 per year for additional conditions and updates.
DNA Direct exists to help you, one-to-one, with genetics and your most important health questions. Our dedicated team of medical specialists can help you understand genetics, your family and medical history, and the genetic tests that may be of benefit to you.

**HOW OUR PROCESS WORKS**

1. **Identify a Genetic Test or Counseling Service.** DNA Direct provides online tools and education to help you discover if a genetic consultation or test is right for you based on your family and medical history. You can email an expert (expert@dnadirect.com) or call 487-410-0000 to speak with a genetic counselor to find out.
Our Solutions

To increase access to genetic expertise in healthcare, DNA Direct has created industry leading guidance and decision support solutions for genetics that deliver the value of genetic testing through personalized expert guidance both pre- and post-testing, along with web-based tools that help individuals and healthcare providers use genetic information to guide healthcare decisions.

Guidance and Decision Support for Genetics

Pre-Test Questionnaires:

Web-based tool to help providers and their patients identify whether testing is appropriate.

Personalized Reports:

Comprehensive patient and physician reports complete with test results, detailed clinical information, guidelines and references available on a secure, personal website.

Prenatal Primer:

Web-based, interactive guide to prenatal testing options, offered exclusively through physicians. Pregnant patients and couples may access the tool for education and decision-making support, based on their physician’s recommendation. Access to CLIA-certified Lab Testing Convenient, secure and clinically valid genetic testing and counseling services conducted by CLIA-certified lab partners with appropriate state licenses to complete clinical testing.

Genetics Call Center:

Genetic counseling services are delivered by phone through DNA Direct’s Genetic Call Center. When a patient places a call, it is routed through our custom telephony system to one of our nationwide genetic counselors. DNA Direct’s counselors are located in our San Francisco office as well as locations nationwide, and they deliver consistent levels of service by operating off of a common workflow and clinical protocols.

http://www.dnadirect.com/web/
Genetic Disorders

Testing for Genetic Disorders

Who Should Consider Genetic Testing for Recurrent Pregnancy Loss?

People have different reasons for choosing genetic testing for recurrent pregnancy loss. For example:

Factors can occur in both women and men that contribute to pregnancy loss. Past experience may suggest that one partner is more at risk for reproductive difficulties. For example:

http://www.dnadirect.com/web/
DNA Direct Clinical Labs

Clinical Labs We Use

The quality of our lab partners is important to DNA Direct. DNA Direct only uses clinical laboratories that are CLIA-certified, have appropriate State licenses, and have lab directors who are qualified to interpret complex test results.

LabCorp Holdings

Regional Lab and Center for Esoteric Testing (CET)
1447 York Court
Burlington, NC 27215

- CLIA Identification No: 34DO655059 (Regional Lab and CET); 34DO866071 (Cytology); 34DO867865 (Histology)
- Additional Licenses/Accreditation: California HIV 068; California C0S 800058; AABB; ASHI; CDC; DOJ; OSHA; Connecticut: CL-0456; Florida: 800004811; Iowa HIV; Maryland 283; New Hampshire Blood Lead PB005; New York: 2502; North Carolina HIV/Cytology L000049HP; Ohio Blood Lead C10028; Pennsylvania: 21885; Rhode Island: 182; U.S. FDA Blood Establishment 105834; U.S. FDA Human Cells, Tissue, and Cellular and Tissue-Based Products; West Virginia: HIV-RL16 CAP Participant No: 13969-01 (Regional Lab and CET), 13969-02 (Histology) CAP Participant No: 13969-04 (Cytology)

DNA Identification Division
1440 York Court
Burlington, NC 27215

- CLIA Identification No: 34DO964630
- American Association of Blood Banks 00030; American Society of Histocompatibility and Immunogenetics 07-3-NC-10-1; Florida 80000370; Forensic Quality Services for Convicted Offender DNA Databasing Laboratories 04-TR-48; Forensic Quality Services International ISO/IEC 04-TR-48; Maryland 1292; New York: 2502; Rhode Island LC000443; Texas Department of Public Safety Crime Laboratory: U.S. FDA Human Cells, Tissue, and Cellular and Tissue-Based Products CAP 13969-05

Center for Molecular Biology and Pathology (CMBP1)
1912 Alexander Drive
Research Triangle Park, NC 27709
- CLIA Identification No: 34DO865205

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