Your Genes and Your Health

http://bio84.stanford.edu/

What We Can Learn from Personal Genomics

http://bio84.stanford.edu/06%20Personal%20Genomics.html

Doug Brutlag, Professor Emeritus of Biochemistry & Medicine (by courtesy) Stanford University School of Medicine

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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)
Common SNPs have Low Odds Ratio and Low Heritability

• Rare High Penetrance Variants Carry High Risk
• Common SNPs Carry Low Risk
• Multiple Variants May Increase Risk Synergistically
• Common SNPs Associated with Genes Containing High Risk Alleles
• Common SNPs Associations can Suggest Regions to Sequence in Cohorts or Trios or Subpopulations

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 the disease
  - How to increase your vigilance for symptoms of specific diseases

- Disease carrier status
  - Premarital genetic counseling
  - Preimplantation genetic diagnosis
  - Neonatal diagnosis
    - Amniocentesis
    - Chorion villus sampling (CVS)
    - Non-Invasive Prenatal Testing tests for fetal DNA in pregnant mother’s blood

- 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
  - Shared haplotypes with close relatives (up to 5th cousins)

- Familial traits, diseases and relationships
  - Known family diseases (breast cancers, 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
We bring the world of genetics to you.

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- Understand what your DNA says about your health, traits and ancestry
- Access interactive tools to share, compare and discover more with friends and family

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23andMe Spit Kit
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Before providing your sample, register your kit at:
www.23andme.com/start

Your sample will NOT be processed unless it is registered.
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
To register your kit, create an account or sign in.

Registration links your kit to your account.

Sign Up

Email

First Name

Last Name

Birthdate

Month

Day

Year

I accept 23andMe's Privacy Policy and Terms of Service. Required.

Security Check

Can't read this? Try another.

Enter the letters you see above

Sign Up

Have an account? Sign In
23andMe Spit Kit
http://23andme.com/
23andMe Login
http://23andme.com

RECOMMENDED FOR YOU

SCANDINAVIAN
ANCESTRY COMPOSITION

HEALTH OVERVIEW

CLOSE FAMILY
2nd & 3rd COUSINS
4th COUSINS
DISTANT COUSINS

DNA RELATIVES

534

NEANDERTHAL ANCESTRY

2.6%

FEATURED CONTENT

Surgical Complications

In the case of a surgical procedure, planned or unplanned, this set of your genetic results and health history information would be important to share with your doctor.

BASED ON YOUR 6 GENETIC REPORTS & 12 SURVEY ANSWERS

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# 23andMe Health Overview

http://23andme.com

## Health Overview

### 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>

See all 120 risk reports...

### 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

See all 49 carrier status...

### 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

See all 57 traits...

### Drug Response (21)

- Clopidogrel (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

See all 21 drug response...

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# 23andMe Disease Risks

## 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>
<td></td>
</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>
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 4-4-4 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**.

**Douglas Brutlag**

- **22.4 out of 100** men of European ethnicity who share Douglas Brutlag's genotype will develop Prostate Cancer between the ages of 35 and 79.

**Average**

- **17.8 out of 100** men of European ethnicity will develop Prostate Cancer between the ages of 35 and 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)

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8q24 (region 1)  

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 “c” 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.

Citations


Wang et al. (2007). “Two common chromosome 8q24 variants are associated with increased risk for prostate cancer.” Cancer Res 67(7):3447-50.


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>
</tr>
<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...
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


Marker Effects

2-fold Increased Risk

Average Risk

2-fold Decreased Risk

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.

MTNR1B

Marker: rs1387153

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](#) ➜

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

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

Alpha-1 Antitrypsin Deficiency

Like · 4 others like this

Your Data · How It Works · Resources · Technical Report · Community (5)

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 stars Established Research for 2 reported markers.

Learn more about the biology of Alpha-1 Antitrypsin Deficiency...

1 of 3. Low levels of alpha-1 antitrypsin can lead to COPD.
## 23andMe Drug Responses

### 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>
<td>★★★★★</td>
<td>Typical</td>
</tr>
<tr>
<td>Thiopurine Methyltransferase Deficiency</td>
<td>★★★★★</td>
<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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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®, Clopilert® 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...
### Show results for: Douglas Brutlag

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

<table>
<thead>
<tr>
<th>Trait</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>
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<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.
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.

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.

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.

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

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>
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 8th-century AD Chinese chieftain.

Your Family and Friends

A2
Samantha Hill

D4e2
Japanese Person

D5a2a'c
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
23andMe Paternal Inheritance

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

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

Neanderthal Ancestry
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</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
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
### 23andMe Relative Finder

#### Relative Finder

<table>
<thead>
<tr>
<th></th>
<th>Male</th>
<th>You</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td><strong>Benjamin Brutlag</strong></td>
<td><strong>Son</strong></td>
<td></td>
<td>United States</td>
</tr>
<tr>
<td></td>
<td>Male, b. 1980</td>
<td>47.7% shared, 22 segments</td>
<td></td>
<td>Southern Europe</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>K1a1b...</td>
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<tr>
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<td></td>
<td></td>
<td>E1b1b1...</td>
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<td>Sharing Genomes</td>
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<td>Send a Message</td>
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<tr>
<td></td>
<td><strong>Pauline Brutlag</strong></td>
<td><strong>Daughter</strong></td>
<td></td>
<td>United States</td>
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<tr>
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<td>Female</td>
<td>53.1% shared, 25 segments</td>
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<td>Northern Europe</td>
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<td>K1a1b...</td>
</tr>
<tr>
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<td>Sharing Genomes</td>
</tr>
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<td>Send a Message</td>
</tr>
<tr>
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<td>Send an Introduction</td>
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<tr>
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<td><strong>3rd to 4th Cousin</strong></td>
<td></td>
<td>United States</td>
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<tr>
<td></td>
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<td>0.77% shared, 3 segments</td>
<td></td>
<td>Aien, Norway</td>
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<td></td>
<td></td>
<td></td>
<td>Haltalen, Norway</td>
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<tr>
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<td></td>
<td></td>
<td>Voss, Norway</td>
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<td></td>
<td>8 m...</td>
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<tr>
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<td></td>
<td>Northern Europe</td>
</tr>
<tr>
<td></td>
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<td></td>
<td></td>
<td>Vorgroven (Vorgraven)</td>
</tr>
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<td>Bakk...</td>
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<td>Good...</td>
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<td>11 m...</td>
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<td>U4b1...</td>
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<td>Introduction Received</td>
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<tr>
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<td></td>
<td></td>
<td>Respond</td>
</tr>
<tr>
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<td>View Family Tree</td>
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<tr>
<td></td>
<td><strong>Larry Vorgroven</strong></td>
<td><strong>3rd to 5th Cousin</strong></td>
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<td>0.54% shared, 2 segments</td>
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</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Otterness</td>
</tr>
<tr>
<td></td>
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<td></td>
<td>Brandsen...</td>
</tr>
<tr>
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<td></td>
<td></td>
<td>Gjørne...</td>
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<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>5 m...</td>
</tr>
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<td>K1a...</td>
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<td>Introduction Received</td>
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<td></td>
<td>Respond</td>
</tr>
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<td></td>
<td>Public Match</td>
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<td>Send a Message</td>
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<tr>
<td></td>
<td><strong>Carolyn Otterness</strong></td>
<td><strong>3rd to 5th Cousin</strong></td>
<td></td>
<td>United States</td>
</tr>
<tr>
<td></td>
<td>Female, b. 1941</td>
<td>0.47% shared, 2 segments</td>
<td></td>
<td>Norway, Denmark, Minnesota, Wisconsin</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Northern Europe</td>
</tr>
<tr>
<td></td>
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<td>En...</td>
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<tr>
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<td></td>
<td>Lars...</td>
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<td>Mest...</td>
</tr>
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<td></td>
<td></td>
<td></td>
<td>6 m...</td>
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<td></td>
<td>K1a...</td>
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<td>Introduction Received</td>
</tr>
<tr>
<td></td>
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<td></td>
<td></td>
<td>Respond</td>
</tr>
</tbody>
</table>
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
### Advanced Controls

Number of grandparents from the same country 1+ Minimum Segment Size 5cM 10cM 15cM Include matches primarily from US, Canada, Australia, New Zealand & South Africa

Indicate segments declared to be of Ashkenazi Jewish ancestry. Only show segments belonging to public individuals.

Learn more about Advanced Controls

<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>3.4% – 5.2%</td>
</tr>
<tr>
<td>Germany</td>
<td>🇩🇪</td>
<td>1.6% – 6.1%</td>
</tr>
<tr>
<td>Ireland</td>
<td>🇮🇪</td>
<td>0.7% – 1.5%</td>
</tr>
<tr>
<td>Denmark</td>
<td>🇩🇰</td>
<td>0.4% – 1.7%</td>
</tr>
<tr>
<td>Russia</td>
<td>🇷🇺</td>
<td>0.4% – 0.9%</td>
</tr>
<tr>
<td>Sweden</td>
<td>🇸🇪</td>
<td>0.3% – 1.0%</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>
What is a Fifth Cousin?

So You’re
23andWe Discoveries

You answer questions. Other 23andMe members answer questions. 23andMe scientists work their magic. And make discoveries!

Your contributions

New Genetic Factors for Hypothyroidism
✓ Thanks! You took a survey that fueled this discovery.

Ancestry and Disease Risk
✓ Thanks! You took a survey that fueled this discovery.

Genes and Geography
✓ Thanks! You took a survey that fueled this discovery.
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
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 genetist 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.
The End of Illness  David B. Agus
DNA Direct brings the power of personalized medicine to payors, providers and patients.

Our Customers
- Health Plans
- Employers
- Hospitals
- Physicians
- Consumers

Our Products
- Policy & Benefit Support Program
- Coverage Management
- Clinical Testing Programs
- Decision Support Program
- Home Biometrics
- Genomic Medicine Network

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.

2. www.fda.gov/drugs/scienceresearch/researchareas/pharmacogenetics/ucm083378.htm
Personalis

Genome Services for Researchers and Clinicians.

Personalis combines world class expertise in the technology of genome sequencing with interpretation.

With Enrollment at 200K, VA's Million Veteran Program Inks Contracts for Genetic Analysis

Contact Us | SEARCH

Services | Technology | Applications | Publications | News & Events | Company
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. →

http://www.stationxinc.com/
Deploy accurate NGS testing with Omicia’s Opal Clinical™

The variant interpretation and reporting platform of choice for the UK 100,000 Genomes Project, LabCorp and clinical labs worldwide.

Accurately report on causative variants from gene panels, exomes, and whole genomes.

Omicia’s Opal Clinical system is a robust, scalable platform developed in collaboration with leading testing labs to accelerate the clinical interpretation of NGS data.

Learn how your lab can optimize report turnaround time and increase diagnostic yield with NGS testing. Schedule a free Opal demo today.
Omicia Movie
http://www.omicia.com/
Sure Genomics
http://www.suregenomics.com/

Our Story
Watch our video to learn more about Sure Genomics' full DNA sequencing process and platform.

Your Full DNA Sequence
We are enabling consumers to obtain, access and review their full Genomic DNA sequence through a system we call Get Look Plan™
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.
Genome Voyager DX from Genos Research
https://voyager-staging.genosresearch.com/index.html#case/13356/sample/33/workspace
NM_013339.3(ALG6): c.391T>C (p.Y131H)
chr1:63,872,031-63,872,032
*Pathogenic*

NM_000362.2(AMPD1): c.242C>T (p.P81L)
chr1:115,231,253-115,231,254
*Pathogenic*

NM_000362.2(AMPD1): c.133C>T (p.Q45*)
chr1:115,236,056-115,236,057
*Pathogenic*

NM_001002294.2(FMO3): c.472G>A (p.E158K)
chr1:171,076,965-171,076,966
*Pathogenic*

NM_001002294.2(FMO3): c.769G>A (p.V257M)
chr1:171,080,079-171,080,080
*Pathogenic*

NM_001002294.2(FMO3): c.923A>G (p.E308G)
chr1:171,083,241-171,083,242
*Pathogenic*
**Genome Voyager DX**

**Genomic Coordinates**
- Chromosome: chr1
- Position: 63,872,031 - 63,872,032
- Cytoband: 1p31.3
- Reference Genome: GRCh37

**DNA Change**
- Reference Sequence: T
- Called Sequence: C
- Small Variation Type: snv
- Zygosity: Heterozygous-Ref

**Allele Frequencies**
- 1000 Genomes Allele Freq: 2.000% (2184)
- ESP6500 Allele Freq: 2.922% (13006)
- Complete Genomics Allele Freq: 1.852% (108)
- Wellcome Allele Freq: Not Found
NM_013339.3(ALG6): c.391T>C (p.Y131H)

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ESP6500 Allele Freq: 2.922% (13006)
Complete Genomics Allele Freq: 1.852% (108)
Wellderley Allele Freq: Not Found

Gene Annotations
Gene Symbol: ALG6
Associated KB Conditions: Congenital disorder of glycosylation, type Ic

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<th>mRNA Acc (Transcript)</th>
<th>Nucleotide Change</th>
<th>Protein Change</th>
<th>Gene Region</th>
<th>Functional Impact</th>
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</thead>
<tbody>
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<td>p.Y131H</td>
<td>CDS, Exon(6)</td>
<td>Missense</td>
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Cross References
dbSNP ID: rs35383149
DGV ID: Not Found
pFam ID: Not Found
mirBase ID: Not Found
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