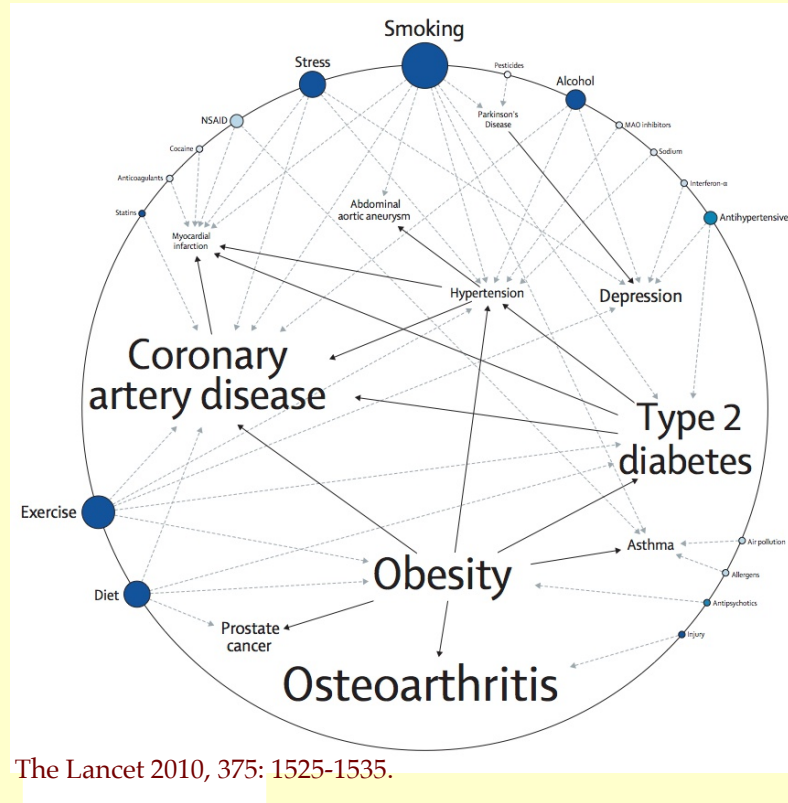


# Genomics, Bioinformatics & Medicine

<http://biochem158.stanford.edu/>

## Personal Genomics

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



Doug Brutlag

Professor Emeritus of Biochemistry & Medicine  
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 5<sup>th</sup> cousins)



# 23andMe

<http://23andme.com/>



sign in

register kit



welcome

ancestry

health

how it works

store

search

help

23andMe can help you manage risk and make informed decisions...



Ancestry

Connect to your past.



Health

Learn for the present.



Research

Participate for the future.

## Learn valuable health & ancestry information.



\$99

Order Now



# 23andMe

<http://23andme.com/>

sign in

register kit



0



welcome

ancestry

research

how it works

buy

help



23andMe provides ancestry-related genetic reports and uninterpreted raw genetic data. We no longer offer our health-related genetic reports. If you are a current customer please go to the [health page](#) for more information. [Close alert](#).



## 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/>





# 23andMe Spit Kit

<http://23andme.com/>



# 23andMe Spit Kit

<http://23andme.com/>

**Before providing your sample, register your kit at:**

**[www.23andme.com/start](http://www.23andme.com/start)**

**Your sample will NOT be processed unless it is registered.**





# 23andMe Spit Kit

<http://23andme.com/>





# 23andMe Spit Kit

<http://23andme.com/>





# 23andMe Spit Kit

<http://23andme.com/>





# 23andMe Login

<http://23andme.com>



HOME

MY RESULTS

FAMILY & FRIENDS

RESEARCH & COMMUNITY

DOUGLAS

## RECOMMENDED FOR YOU



NEANDERTHAL ANCESTRY

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

2    17    236    683

DNA RELATIVES



ANCESTRY OVERVIEW

## FEATURED CONTENT

### Migraines - The Wisdom of the Crowd

Did you know that shopping is a trigger for 22% of migraine sufferers? Learn what else the crowd can uncover about migraine triggers, symptoms, treatments and more.



### Your Diet, Your Health

Learn more about how your genes play a role in your diet and health. Share your findings away from caffeine or gluten?

# 23andMe Health Overview


<http://23andme.com>

## health overview

[Print my health overview](#) | [Share my health results](#)

Show results for

[See new and recently updated reports »](#)

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

### Disease Risks (120, 1 locked report) ?

 Elevated Risks	Your Risk	Average Risk
Prostate Cancer 	22.4%	17.8%
Colorectal Cancer	7.1%	5.6%
Melanoma	6.0%	2.9%
Chronic Kidney Disease	4.2%	3.4%
Restless Legs Syndrome	2.5%	2.0%

[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...](#)

## disease risk

Share my health results with family and friends

Show results for Douglas Brutlag

[See new and recently updated reports](#)

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

### Locked Reports ?

Name	Confidence	Your Risk	Avg. Risk	Compared to Average
Alzheimer's Disease	★★★★	🔒	🔒	🔒

### Elevated Risk ?

Name	Confidence	Your Risk	Avg. Risk	Compared to Average
Prostate Cancer ♂	★★★★	22.4%	17.8%	1.26x 📈
Colorectal Cancer	★★★★	7.1%	5.6%	1.27x 📈
Melanoma	★★★★	6.0%	2.9%	2.10x 📈
Chronic Kidney Disease	★★★★	4.2%	3.4%	1.22x 📈
Restless Legs Syndrome	★★★★	2.5%	2.0%	1.25x 📈
Exfoliation Glaucoma	★★★★	2.2%	0.7%	2.90x 📈
Abdominal Aortic Aneurysm	★★★			📈
Ankylosing Spondylitis	★★★			📈
Asthma	★★★			📈

🏠 [My Home](#)

Inbox (7)

[My Health](#)

- ▶ [Disease Risk](#)
- [Carrier Status](#)
- [Drug Response](#)
- [Traits](#)
- [Health Labs](#)
- [Family Health History](#)

[My Ancestry](#)

- [Maternal Line](#)
- [Paternal Line](#)
- [Relative Finder](#)
- [Ancestry Painting](#)
- [Global Similarity](#)
- [Ancestry Labs](#)

[Sharing & Community](#)

- [Compare Genes](#)
- [Family Inheritance](#)
- [23andMe Community](#)
- [Genome Sharing](#)

23andMe

## disease risk

[Next ▶](#)  
*Psoriasis*

### Prostate Cancer

[Like](#) - 0 others like this

[Share](#)

[Your Data](#)

[How It Works](#)

[Timeline](#)

[MD's Perspective](#)

[Resources](#)

[Technical Report](#)

[Community \(24\)](#)

### Prostate Cancer

 [Printable Version](#)

**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 ★★★★★ **Established Research** for 12 reported markers, updated [November 4th, 2010](#).

[Learn more about the biology of Prostate Cancer...](#)  
[Major discoveries in Prostate Cancer...](#)



1 of 3. Prostate cancer affects about 1 in 6 men. (Women don't have prostate glands and therefore cannot get prostate cancer.)

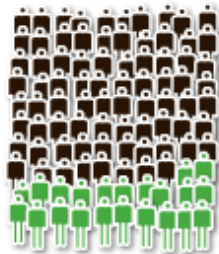
# 23andMe Prostate Cancer Risks

## Your Genetic Data

» Share your health results

Show information for  assuming  ethnicity and an age range of

[Where's mine?](#)



### 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

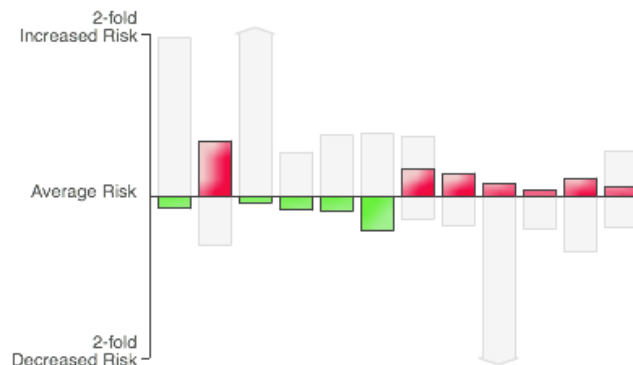
**42-57 %**  
Attributable to  
Genetics

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](#).

### 8q24 (region 1)

Marker: **rs1447295**

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.












### Citations

- [Amundadottir et al. \(2006\)](#) . "A common variant associated with prostate cancer in European and African populations." *Nat Genet* 38(6):652-8.
- [Freedman et al. \(2006\)](#) . "Admixture mapping identifies 8q24 as a prostate cancer risk locus in African-American men." *Proc Natl Acad Sci U S A* 103(38):14068-73.
- [Severi et al. \(2007\)](#) . "The common variant rs1447295 on chromosome 8q24 and prostate cancer risk: results from an Australian population-based case-control study." *Cancer Epidemiol Biomarkers Prev* 16(3):610-2.
- [Yeager et al. \(2007\)](#) . "Genome-wide association study of prostate cancer identifies a second risk locus at 8q24." *Nat Genet* 39(5):645-9.
- [Gudmundsson et al. \(2007\)](#) . "Genome-wide association study identifies a second prostate cancer susceptibility variant at 8q24." *Nat Genet* 39(5):631-7.
- [Wang et al. \(2007\)](#) . "Two common chromosome 8q24 variants are associated with increased risk for prostate cancer." *Cancer Res* 67(7):2944-50.
- [Schumacher et al. \(2007\)](#) . "A common 8q24 variant in prostate and breast cancer from a large nested case-control study." *Cancer Res* 67(7):2951-2956.
- [Suuriniemi et al. \(2007\)](#) . "Confirmation of a positive association between prostate cancer risk and a locus at chromosome 8q24." *Cancer Epidemiol Biomarkers Prev* 16(4):809-14.
- [Cheng et al. \(2008\)](#) . "8q24 and prostate cancer: association with advanced disease and meta-analysis." *Eur J Hum Genet* 16(4):496-505.
- [Zheng et al. \(2008\)](#) . "Cumulative association of five genetic variants with prostate cancer." *N Engl J Med* 358(9):910-9.



# 23andMe Disease Risks

## Decreased Risk

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



### Type 2 Diabetes

Like · 4 others like this

Share

Your Data

How It Works

Timeline

MD's Perspective

Resources

Technical Report

Community (24)

## Type 2 Diabetes

 [Printable Version](#)

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

» [Share your health results](#)

Show information for  assuming  ethnicity  
 and an age range of

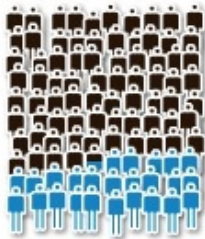
[Why are there limited choices of ethnicity in risk reports?](#)



### 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

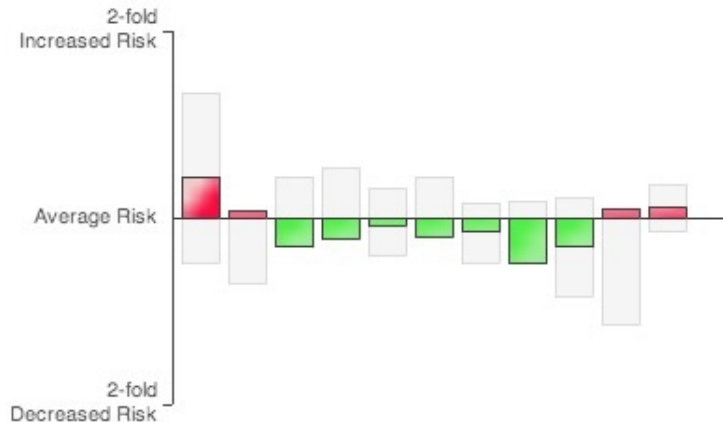
**26 %**  
 Attributable to  
 Genetics



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.

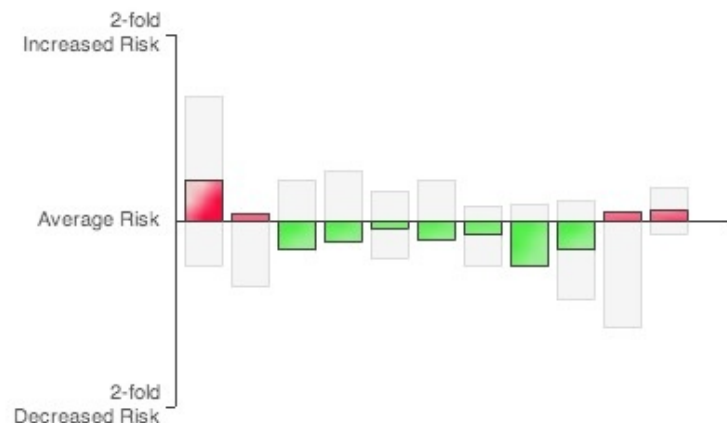
The T version of this SNP is also associated with increased odds of gestational diabetes, a form of diabetes that occurs only during pregnancy. Gestational diabetes can lead to complications for both mother— such as difficult delivery due to unusually large infant size— and baby, such as low blood sugar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

## Citations

- [Grant et al. \(2006\)](#) . "Variant of transcription factor 7-like 2 (TCF7L2) gene confers risk of type 2 diabetes." *Nat Genet* 38(3):320-3.
- [Saxena et al. \(2006\)](#) . "Common single nucleotide polymorphisms in TCF7L2 are reproducibly associated with type 2 diabetes and reduce the insulin response to glucose in nondiabetic individuals." *Diabetes* 55(10):2890-5.
- [Helgason et al. \(2007\)](#) . "Refining the impact of TCF7L2 gene variants on type 2 diabetes and adaptive evolution." *Nat Genet* 39(2):218-225.
- [Sladek et al. \(2007\)](#) . "A genome-wide association study identifies novel risk loci for type 2 diabetes." *Nature* 445(7130):881-5.
- [Saxena et al. \(2007\)](#) . "Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels." *Science* 316(5829):1331-6.
- [Zeggini et al. \(2007\)](#) . "Replication of genome-wide association signals in UK samples reveals risk loci for type 2 diabetes." *Science* 316(5829):1336-41.

# 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](#).

## 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

- Voight BF et al. (2010) . "Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis." *Nat. Genet.* 42(7):579-89.
- Bouatia-Naji N et al. (2009) . "A variant near MTNR1B is associated with increased fasting plasma glucose levels and type 2 diabetes risk." *Nat. Genet.* 41(1):89-94.
- Prokopenko I et al. (2009) . "Variants in MTNR1B influence fasting glucose levels." *Nat. Genet.* 41(1):77-81.
- Peschke E (2008) . "Melatonin, endocrine pancreas and diabetes." *J. Pineal Res.* 44(1):26-40.



# 23andMe Carrier Status

## carrier status

Share my health results with family and friends

Show results for

[See new and recently updated reports »](#)

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

Name	Confidence	Confidence ▾
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
Fanconi Anemia (FANCC-related)	★★★★★	Variant Absent
Familial Hypercholesterolemia Type B	★★★★★	Variant Absent
Familial Mediterranean Fever	★★★★★	Variant Absent
G6PD Deficiency	★★★★★	Variant Absent
Gaucher Disease	★★★★★	Variant Absent
Glycogen Storage Disease Type 1a	★★★★★	Variant Absent
Hemochromatosis (HFE-related)	★★★★★	Variant Absent
Limb-girdle Muscular Dystrophy	★★★★★	Variant Absent
Maple Syrup Urine Disease Type 1B	★★★★★	Variant Absent



# 23andMe Carrier Status for Alpha-1 Antitrypsin Deficiency



- 🏠 [My Home](#)
- Inbox (7)
- [My Health](#)
- Disease Risk
- ▶ [Carrier Status](#)
- Drug Response
- Traits
- Health Labs
- Family Health History
- [My Ancestry](#)
- Maternal Line
- Paternal Line
- Relative Finder
- Ancestry Painting
- Global Similarity
- Ancestry Labs
- [Sharing & Community](#)
- Compare Genes
- Family Inheritance
- 23andMe Community
- Genome Sharing

## carrier status

[Next ▶](#)  
Autosomal Recessive Po...

**Alpha-1 Antitrypsin Deficiency**
Like · 4 others like this
Share

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

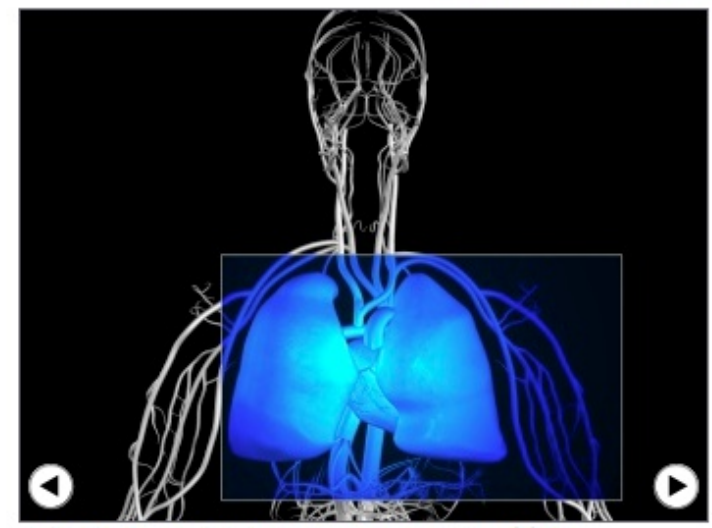
### Alpha-1 Antitrypsin Deficiency

[Printable Version](#)

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 ★★★★★ **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

## drug response

Share my health results with family and friends

Show results for

[See new and recently updated reports »](#)

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

Name	Confidence ▾	Status
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
Pseudocholinesterase Deficiency	★★★★★	Typical
Thiopurine Methyltransferase Deficiency	★★★★★	Typical
Warfarin (Coumadin®) Sensitivity	★★★★★	Typical
Oral Contraceptives, Hormone Replacement Therapy and Risk of Venous Thromboembolism ♀	★★★★★	Not Applicable
Caffeine Metabolism	★★★	Fast Metabolizer
Hepatitis C Treatment Side Effects	★★★	See Report
Metformin Response	★★★	Typical Odds of Positive Response
Antidepressant Response	★★	See Report

## My Home

Inbox (6)

## My Health

Disease Risk

Carrier Status

## Drug Response

Traits

Health Labs

## My Ancestry

Maternal Line

Paternal Line

Relative Finder

Ancestry Painting

Global Similarity

Ancestry Labs

## Sharing & Community

Compare Genes

Family Inheritance

23andMe Community

Genome Sharing

## 23andWe

Research Surveys (24)

Research Snippets

# drug response

## Clopidogrel (Plavix®) Efficacy ★★★★★ ?

share this

Established Research report on 5 reported markers.

Your Data

How It Works

Resources

Technical Report

Next ▶

Floxacin Toxicity

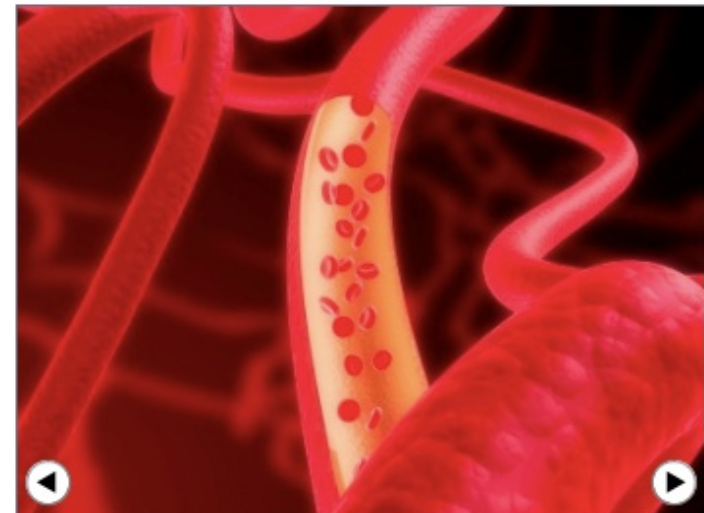
Only a medical professional can determine whether clopidogrel is the right medication for a particular patient. The information contained in this report should not be used to independently establish a clopidogrel regimen, or abolish or adjust an existing course of treatment.

### About Clopidogrel Efficacy

 [Printable Version](#)

**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...](#)



1 of 3. Clopidogrel keeps platelets from sticking together and prevents blood clots.

# Plavix Ad with Genetic Requirement



[My Home](#)

Inbox (3)

[My Health](#)

Disease Risk

Carrier Status

Drug Response

► [Traits](#)

Health Labs

**My Ancestry**

Maternal Line

Paternal Line

Relative Finder

Ancestry Painting

Global Similarity

Ancestry Labs

**Sharing & Community**

Compare Genes

Family Inheritance

23andMe Community

Genome Sharing

**23andWe**

Research Surveys (21)

Research Snippets

Research Initiatives

Research Discoveries

# traits

Share my health results with family and friends

Show results for

[See new and recently updated reports »](#)

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

Name	Confidence ▲	Outcome
<a href="#">Alcohol Flush Reaction</a>	★★★★★	Does Not Flush
<a href="#">Bitter Taste Perception</a>	★★★★★	Can Taste
<a href="#">Earwax Type</a>	★★★★★	Wet
<a href="#">Eye Color</a>	★★★★★	Likely Brown
<a href="#">Hair Curl</a> 🌟	★★★★★	Straighter Hair on Average
<a href="#">Lactose Intolerance</a>	★★★★★	Likely Tolerant
<a href="#">Malaria Resistance (Duffy Antigen)</a>	★★★★★	Not Resistant
<a href="#">Male Pattern Baldness</a> ♂	★★★★★	Decreased Odds
<a href="#">Muscle Performance</a>	★★★★★	Likely Sprinter
<a href="#">Non-ABO Blood Groups</a>	★★★★★	See Report
<a href="#">Norovirus Resistance</a>	★★★★★	Not Resistant
<a href="#">Resistance to HIV/AIDS</a>	★★★★★	Not Resistant
<a href="#">Smoking Behavior</a>	★★★★★	Typical
<a href="#">Adiponectin Levels</a> <span style="background-color: #f96;">new</span>	★★★	See Report
<a href="#">Asparagus Metabolite Detection</a> 🌟	★★★	Typical Odds of Detecting
<a href="#">Birth Weight</a>	★★★	See Report
<a href="#">Blood Glucose</a>	★★★	5.18 mmol/L on Average
<a href="#">Breastfeeding and IQ</a>	★★★	See Report
<a href="#">C-reactive Protein Level</a>	★★★	2.09 mg/L on Average

# 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

Share

Map

History

Haplogroup Tree

Community

### 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

<a href="#">A2</a>	Samantha Hill
<a href="#">D4e2</a>	Japanese Person
<a href="#">D5a2a'c</a>	Chinese Person
<a href="#">H3</a>	Lilly Mendel (Mom), Erin Mendel (Daughter), Alan Mendel (Son), Ian Mendel (Son), Margo Fisher (Grandma)
<a href="#">H4a1</a>	Ron Fisher (Grandpa)
<a href="#">K1a1b1a</a>	Benjamin Brutlag, Pauline Brutlag, Simone Brutlag
<a href="#">L3e2b2</a>	Nigerian Person
<a href="#">M35b</a>	renu heller

## Paternal Haplogroup: E1b1b1a2\*

Share

Map

History

Haplogroup Tree

Community

### 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.

**Haplogroup:** E1b1b1a, a subgroup of [E1b1b](#)

**Age:** 23,000 years

**Region:** Northern Africa, Southern Europe

**Populations:** Berbers, Iberians, Balkans

**Highlight:** Two different migrations brought E1b1b1a into Europe.

### Your Family and Friends

[D2a1b](#) Japanese Person

[E1b1a8a1...](#) Nigerian Person

[E1b1b1a2...](#) Douglas Brutlag, Benjamin Brutlag

[G2a](#) Brian Becker

[I1\\*](#) Greg Mendel (Dad), Alan Mendel (Son), Ian Mendel (Son), Fred Mendel (Grandpa)



# 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.

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

2    0    60    443



## From Your Ancestry Expert

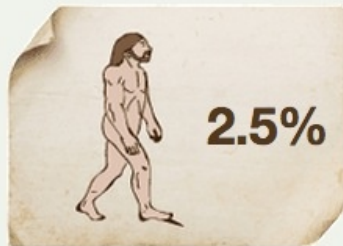
It's remarkable what you can discover from a little saliva. On this page are the highlights of what we've learned about your ancestry, based just on your DNA. Enjoy!

*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.*

## 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.



## AS SEEN ON



A N D E R S O N

[Ancestry Help](#)

[Send Feedback](#)

## Top Relative Surnames

Surname	Count	Enrichment
Anderson	5	10
Smith	5	1



# Ancestry Composition

ancestry composition

Douglas Brutlag

Standard Estimate

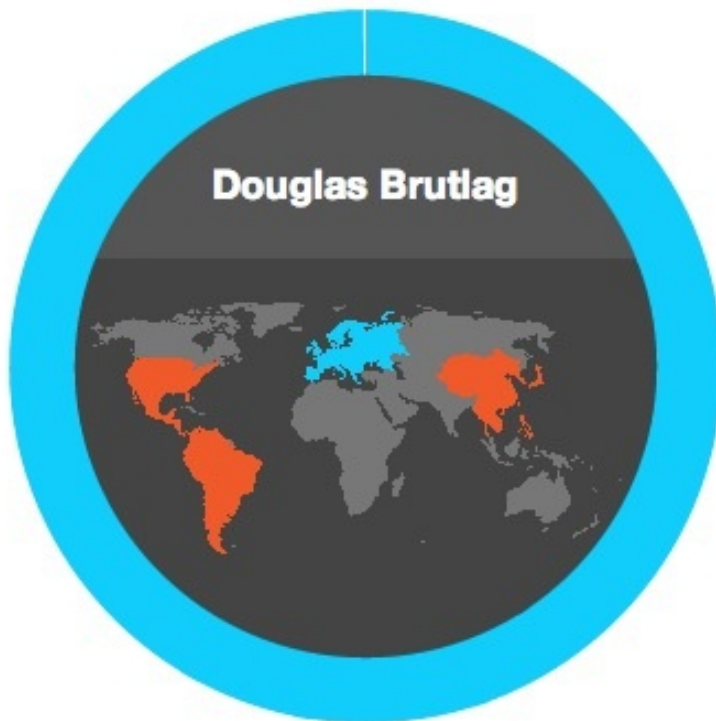


Map View

Global Resolution



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

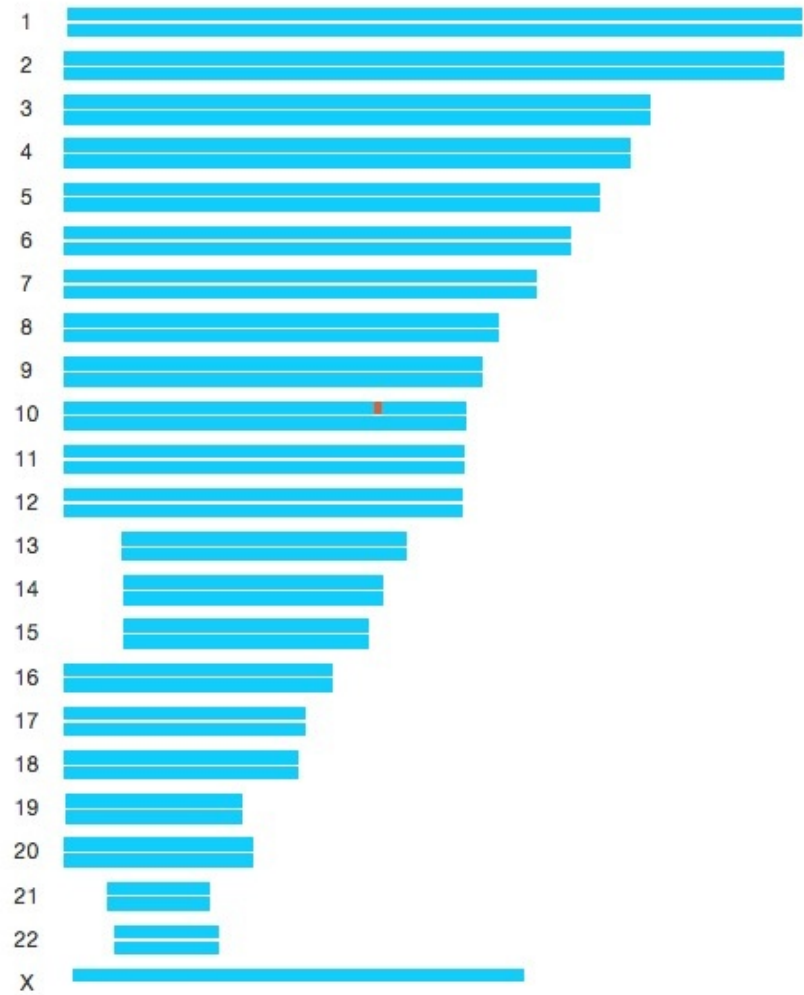
Douglas Brutlag

Speculative Estimate



Chromosome View

Global Resolution



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.

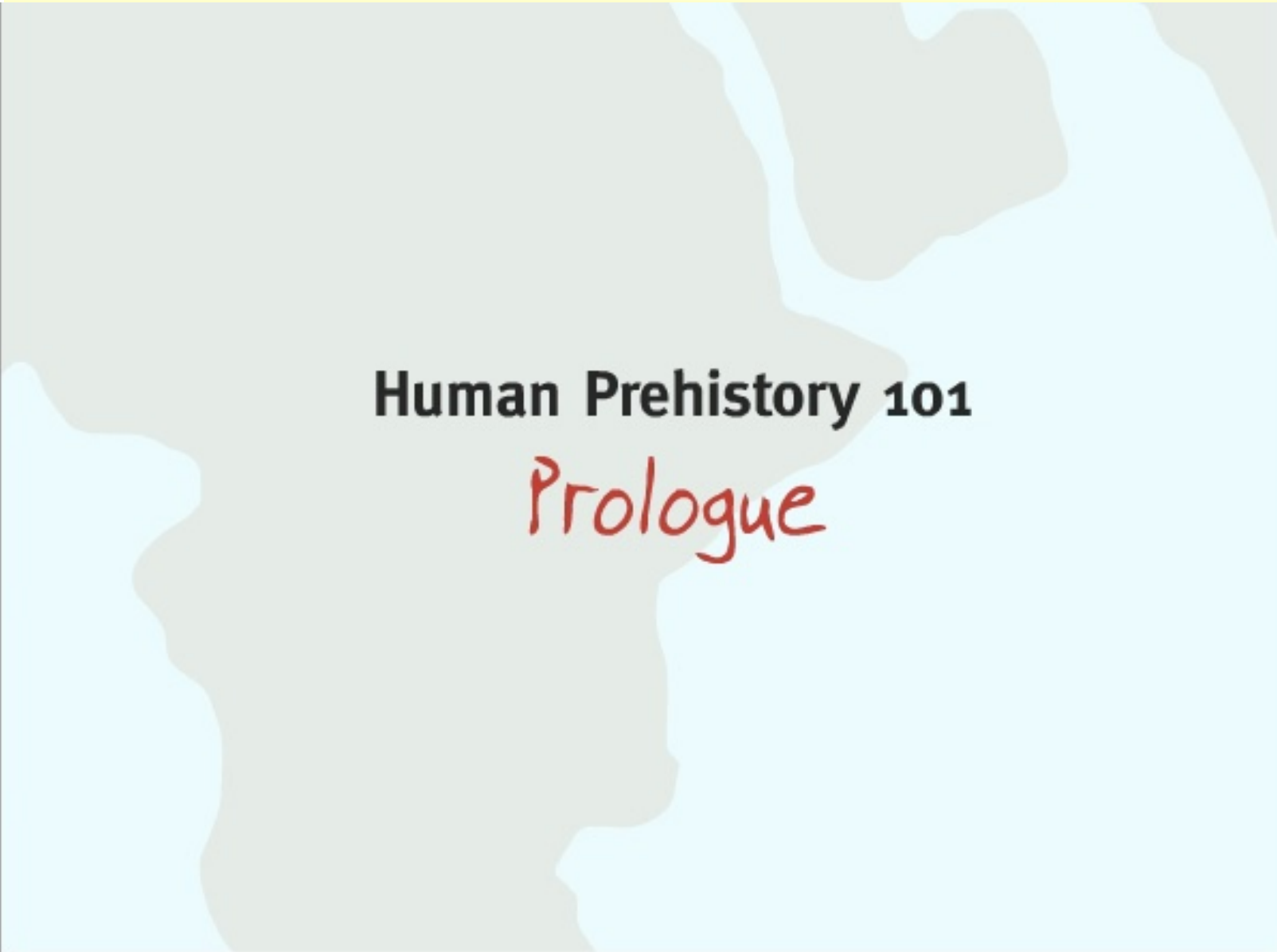
99.9% European

0.1% East Asian & Native American

100.0% Douglas Brutlag

[show all populations](#)

# History of Man

A large, light gray silhouette of a human head and neck, facing right, set against a light blue background. The silhouette is centered on the page.

**Human Prehistory 101**









*Prologue*

# 23andMe Relative Finder List View

## relative finder

? List view Map view Surname view

Q search matches Show: both sides Sort: relationship 25 per page 1 - 25 of 504

	Male	You	U5b... E1b1b1...	<a href="#">Update Your Profile</a>
	<b>Benjamin Brutlag</b> Male, b. 1980	Son 47.7% shared, 22 segments	United Sta... Southern Euro... K1a1b... E1b1b1...	Sharing Genomes <a href="#">Send a Message</a>
	<b>Pauline Brutlag</b> Female	Daughter 53.1% shared, 25 segments	United Sta... Northern Euro... K1a1b...	Sharing Genomes <a href="#">Send a Message</a>
	Male	3rd to 4th Cousin 0.77% shared, 3 segments	... I2...	<a href="#">Send an Introduction</a>
	Male	3rd to 5th Cousin 0.47% shared, 3 segments	... R1a...	<a href="#">Send an Introduction</a>
	<b>Larry Vongroven</b> Male	3rd to 5th Cousin 0.54% shared, 2 segments	United Sta... Alen, Nor... Haltalen, Nor... Voss, Nor... 8 m... Northern Euro... Vongroven (Vongrav... Bakk... Good... 11 m... U4b1... R1a...	Introduction Received <a href="#">Respond</a> <a href="#">View Family Tree</a>
	<b>Carolyn Otterness</b> Female, b. 1941	3rd to 5th Cousin 0.47% shared, 2 segments	United Sta... Otsego, Wisconsin, Dodge County, C... Northern Euro... Ottern... Brandsn... Gjerne... 5 m... K1a...	Public Match <a href="#">Send a Message</a>
	<b>Gale Enger</b> Male, b. 1925	3rd to 5th Cousin 0.41% shared, 2 segments	United Sta... Norway, Denmark, Minnesota, Wisco... Northern Euro... En... Lars... Mest... 6 m... K1... ...	Introduction Received <a href="#">Respond</a>



# 23andMe Relative Finder Map View

## relative finder

List view Map view Surname view

Search your matches

Total results: 193

Clustering: Off On

### 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

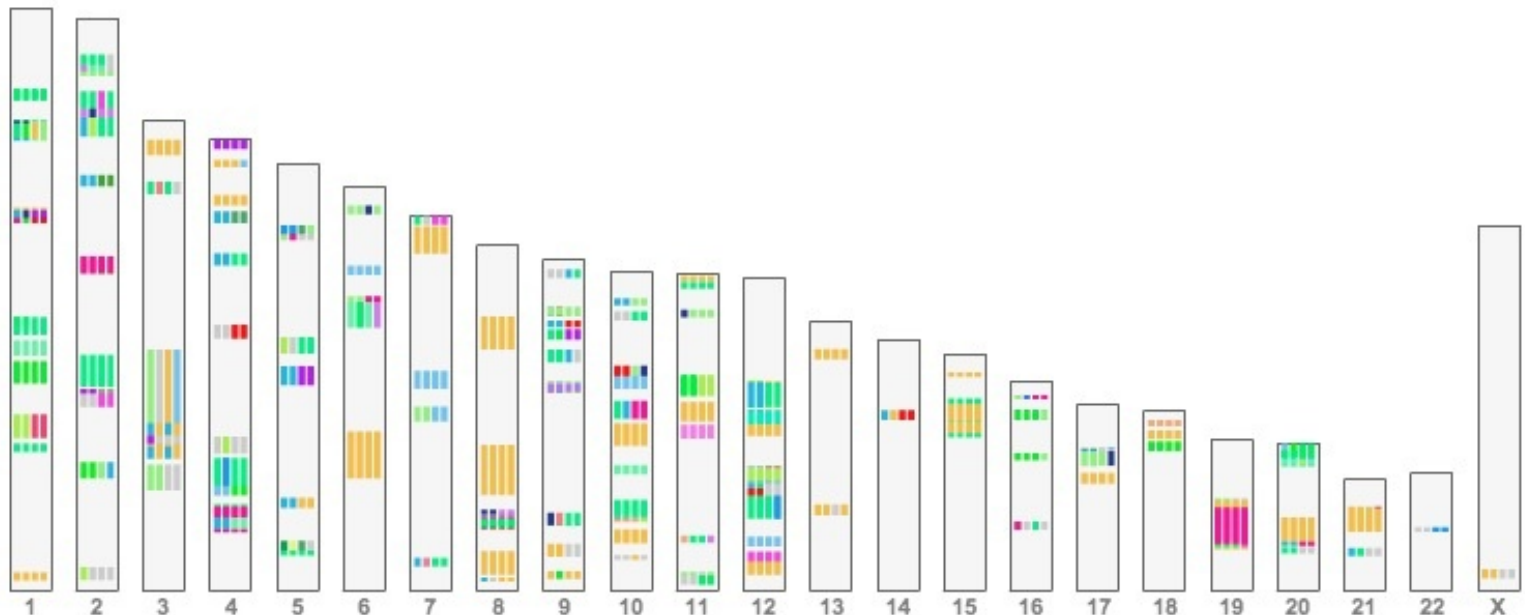


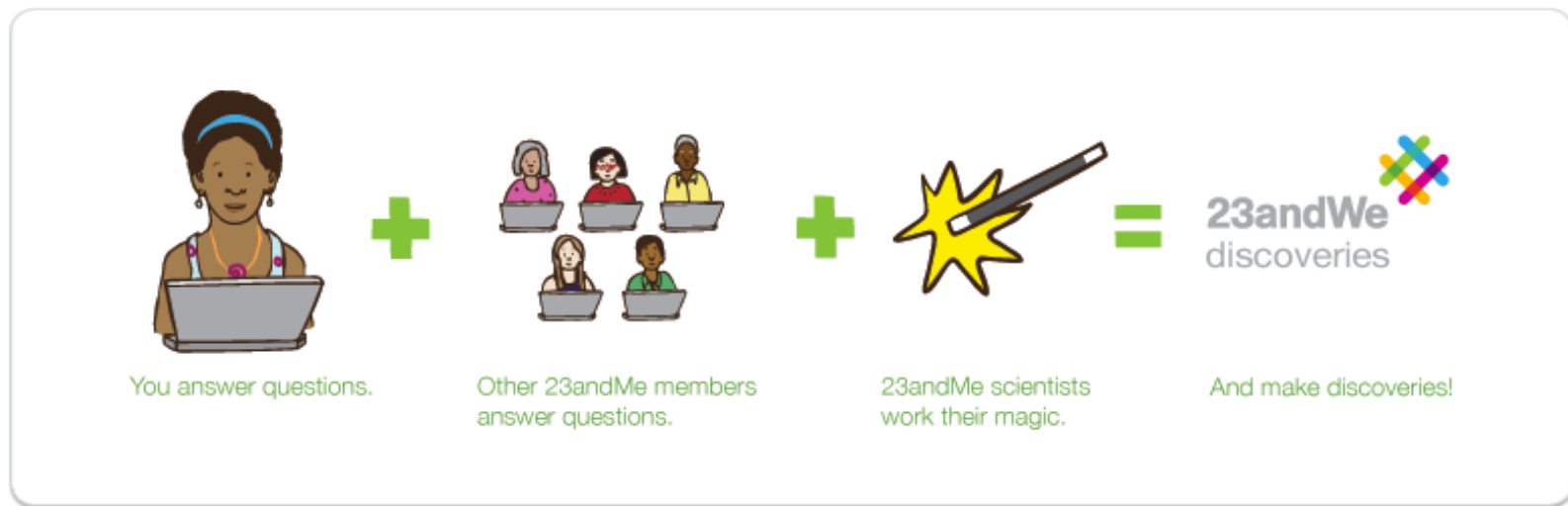
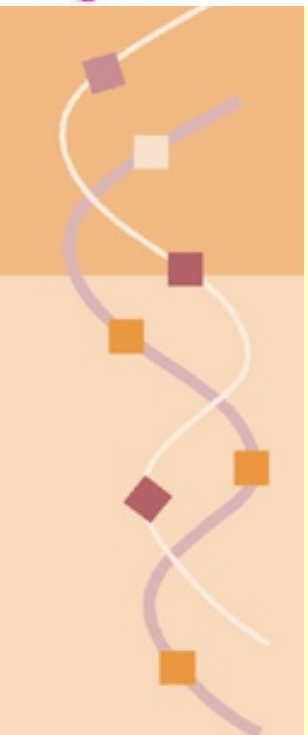
# What is a Fifth Cousin?

**So You're**

# 23andMe Ancestry Finder





Country	Color	Percent of Douglas Brutlag's Genome Covered
 Norway		3.6% – 5.4%
 Germany		1.3% – 4.2%
 Ireland		0.7% – 1.4%
 Sweden		0.5% – 1.2%
 Denmark		0.4% – 1.7%
 Russia		0.4% – 0.8%
 Netherlands		0.3% – 0.7%
 Finland		0.3% – 0.7%





## 23andWe community contributions

You haven't taken the surveys that led to these discoveries (yet!)


-  **Asparagus Metabolite Detection**  
This report is based on "Ten Things About You".  
[View this report](#) [get involved!](#)
-  **Freckling**  
This report is based on "Pigmentation".  
[View this report](#) [get involved!](#)
-  **Hair Curl**  
This report is based on "Ten Things About You".  
[View this report](#) [get involved!](#)
-  **Photic Sneeze**  
This report is based on "Ten Things About You".  
[View this report](#) [get involved!](#)





## A new paradigm for genetic research.



 [Read our open letter to the science community](#)

23andMe 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.

23andMe 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 web site.

▶▶ [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.

Call: (800) 975-4819

[For Patients](#)

[For Providers](#)

[For Hospitals](#)

[For Health Plans](#)

Connecting  
Patients to  
the Power  
of Genetics

## InformedDNA

### Individualized Counseling by Genetics Experts.

Convenient access to independent genetic experts who will guide you or your patients through the complexities of family health history, genetics and genetic testing.

I'm a Patient →

[REQUEST YOUR APPOINTMENT →](#)

[ACCESS MY SECURE MESSAGES →](#)

I'm a Provider →

[REFER A PATIENT →](#)

Call InformedDNA Today: (800) 975-4819





## 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

[REFER A PATIENT →](#)

### Access to Experts

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

[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 →](#)

# Navigenics

[Member Login](#) | [Physician Login](#) | [Request Information](#)

(866) 522-1585 / +1 (650) 585-7743



[What We Offer](#)

[Genetics & Health](#)

[For Physicians](#)

[About Us](#)

[Try Demo](#)

## Overview

[DNA basics](#)

[Family history](#)

[Taking action](#)

[FAQs](#)

[Terminology](#)

## Receive our newsletter

email address

[Sign Up](#)

### 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.

[LEARN MORE >](#)

## 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.

### Understand your DNA

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

[MORE >](#)

### Get a more complete picture

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

[MORE >](#)

### Start a personalized approach

Find out how your genetic test results can help point you toward better health and well-being.

[MORE >](#)



- Overview**
- Our services
- FAQs
- Tools
- Set up your practice
- Request information

Physician newsletter

[Sign Up](#)

**What other physicians say:**

“We believe that Navigenics’ preventive genomics service has the potential to be an innovation that could significantly enhance patient care.”

-Edward Goldman, M.D.,  
Former CEO, MDVIP, Inc.

# 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.



[▶ Play Video](#)

## 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.

- Resources**
- Downloads**
- [Applying Preventive Genomic Medicine in Clinical Practice](#)
  - [The Science Behind the Navigenics Health Compass Service](#)
  - [Conditions We Cover](#)
- Quick links**
- [Medications Wallet Card](#)
  - [Our testing services](#)
  - [Educational webinars](#)
  - [Physician FAQs](#)
  - [Case studies](#)

[View Demo](#) [Request Info](#)

**Free Information Kit**



Navigenics physician kit

[Order](#)



# Navigenics

[What We Offer](#)
[Genetics & Health](#)
[For Physicians](#)
[About Us](#)
[Try Demo](#)
[Overview](#)
[How it works](#)
[Our genetic analysis](#)
[Why Navigenics](#)
[Genetic counseling](#)
[Conditions & medications](#)
[Our policies](#)
[FAQs](#)
[Success stories](#)
[Request information](#)

## Receive our newsletter

[Sign Up](#)

## 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

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

### Medications

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



**Health Conditions** **Medications**

[Print this page](#)

# 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.

0 - 1%	>1 - 10%	>10 - 25%	>25 - 50%	>50 - 100%
<b>Brain aneurysm</b> You: 0.91% Avg: 0.90% >>	<b>Alzheimer's disease</b> You: 10% Avg: 17% >>	<b>Breast cancer</b> You: 20% Avg: 13% >>	<b>Diabetes, type 2</b> You: 36% Avg: 30% >>	You have no results in this range
<b>Macular degeneration</b> You: 0.78% Avg: 3.1% >>	<b>Lung cancer</b> You: 5% Avg: 6% >>	<b>Atrial fibrillation</b> You: 19% Avg: 23% >>	<b>Heart attack</b> You: 26% Avg: 25% >>	
<b>Sarcoidosis</b> You: 0.55% Avg: 1.0% >>	<b>Deep vein thrombosis</b> You: 4.2% Avg: 3.6% >>		<b>Osteoarthritis</b> You: 26% Avg: 47% >>	
<b>Glaucoma</b> You: 0.47% Avg: 2.4% >>	<b>Colon cancer</b> You: 4.1% Avg: 5% >>		<b>Obesity</b> You: 25% Avg: 32% >>	

## Key to your results

**Condition name**

**Diabetes type 2**

**Your results**  
You: 45%

**Population Average**  
Avg: 30%

[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.

[View Report](#)

[Example Results](#) This is an example Navigenics Health Compass report.
**Health Conditions****Medications** [Print this page](#)

# 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

Drug	Your Risk	Side Effect
<a href="#">Fluorouracil (Efudex®)</a> Used to treat many types of cancer	High Risk	Severe, potentially fatal toxicity
<a href="#">Simvastatin (Vytorin®, Zocor®)</a> Used to treat high cholesterol and help prevent heart disease	Moderate Risk	Muscle pain and damage
<a href="#">Abacavir (Ziagen®)</a> Used to treat HIV infection	Low Risk	Severe allergic reaction, including fever, rash, and nausea

**Be Prepared**
**Talk to your doctor about updating your medical records.**

Carry a wallet card with your Medications profile.

[Print Wallet Card](#)
**Printable report**

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

[View Report](#)

# 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

Overview

How it works

Our genetic testing

Why Navigenics

Genetic counseling

Conditions we cover

Our policies

FAQs

Success stories

Request information

Tell a friend

Try Demo

Order Now

Receive our newsletter

email address

Sign Up

## 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 going to be.

## Conditions we cover

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

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

### 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:

# DNAdirect: Clinical Genetic Testing









## 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
- » About Personalized Medicine
- » Newsroom
- » About Us

DNA Direct brings the power of personalized medicine to payors, providers and patients.

 <b>THE RIGHT PERSON</b>	 <b>THE RIGHT TEST</b>	 <b>THE RIGHT INTERPRETATION</b>
Finding the right people to benefit from genomic medicine can improve disease management and lower healthcare costs.	Getting the wrong test can misinform medical decisions and increase healthcare costs.	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.

## About Personalized Medicine

### Our Customers

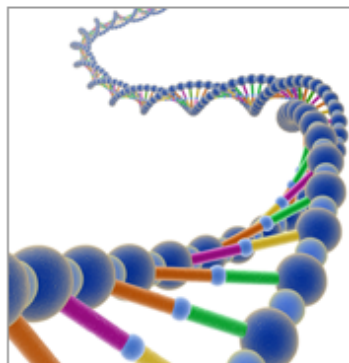
- » Health Plans
- » Employers
- » Hospitals
- » Physicians
- » Consumers

### Our Products

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

### About Personalized Medicine

- » Newsroom
- » About Us



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 year<sup>1</sup> and the Food and Drug Administration (FDA) states that more than 100 medications have pharmacogenomic information included in their drug labels<sup>2</sup>. 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 virology<sup>3</sup>. 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](http://www.fda.gov/drugs/scienceresearch/researchareas/pharmacogenetics/ucm083378.htm)

3. *Personalized Medicine Is Playing A Growing Role In Development Pipelines* November/December 2010 Tufts CSDD Impact Report ; Vol12:6

 Knome®

The Human Genome Interpretation Company™



knoSYS™100



knomeDISCOVERY



knomeBASE

## Genome Voyager™ ALPHA



[LEARN MORE HERE](#)

### 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.

BGI-Shenzhen  
Completes  
Acquisition of  
Complete Genomics

[Learn More](#)

**nature**  
Read the  
ground  
breaking  
Nature paper  
on LFR  
Technology

[Read More](#)



## Personalis Genome Services for Researchers and Clinicians.



[Applications](#) >

[Services](#) >

PERSONALIS COMBINES WORLD CLASS EXPERTISE IN THE TECHNOLOGY OF GENOME SEQUENCING WITH INTERPRETATION.



# 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. →

# BaseHealth and Genophen

<http://www.basehealth.com/>



[Home](#) [Team](#) [Press](#) [Contact](#)

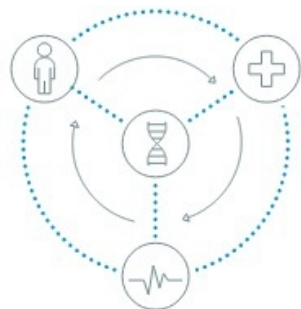
Login

Sign Up

## 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.

[LEARN MORE](#)



INTEGRATED DATA



COMPREHENSIVE ANALYSIS



PERSONALIZE YOUR PRACTICE

# Personal Genomics References

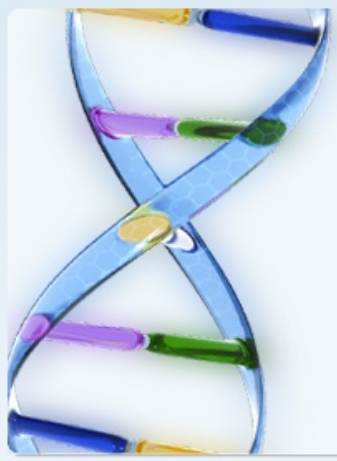
---

- Clinical Assessment Incorporating a Personal Genome. Ashley, E. et al. (2010) *Lancet* 375, 1525-1535.
- Emerging genomic applications in coronary artery disease. Damani SB, Topal EJ, *JACC Cardiovasc. Intervention* (2011). 4:473-482.
- Clinical applicability of sequence variations in genes related to drug metabolism. Stojiljkovic M, Patrinos GP, Pavlovic S. (2011) *Curr Drug Metab.* 1;12(5):445-54.
- Clinical pharmacogenetics and potential application in personalized medicine. Zhou et al., (2008) *Curr Drug Metab.* 9(8):738-84.
- Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. Cooper et al (2010) *Hum Mutat.* 31(6):631-55.
- Web-based, participant-driven studies yield novel genetic associations for common traits. Eriksson et al. (2010) *PLoS Genetics* 6, e1000993.

# 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.

[Learn More](#)



### Success Stories



"We hear a lot of different – and sometimes conflicting – opinions about how to take care of our health. I'm very excited about receiving only the most relevant information to me, based on my DNA."

[More Success Stories](#)

### 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.

[Find a physician now >](#)

#### 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



[View all collaborators >](#)

**Latest Headlines** [Video: Dr. Vanier joined by Dr. Eric Topol to discuss NEJM study data](#)

[Read our blog >](#)

# Navigenics

What We Offer

Genetics & Health

For Physicians

About Us

Try Demo

Overview

DNA basics

Understanding DNA

Genetic markers

DNA analysis

DNA & destiny

Family history

Taking action

FAQs

Terminology

## 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 >](#)

Receive our newsletter

email address

Sign Up

Success story:

I wouldn't be getting early care to ward off macular degeneration without the empowering results my genetic test provided me.

— Patrick,  
Internet executive



## DNA Basics

You can step through this simple tutorial to learn the essentials of DNA.

Next

[Overview](#)
[DNA basics](#)
[Family history](#)
[Inheriting DNA](#)
[Family history & DNA](#)
[Uncertain history](#)
[Complete picture](#)
[Taking action](#)
[FAQs](#)
[Terminology](#)

Receive our newsletter

[Sign Up](#)

### Success story:

“I assumed that I knew my family history, but after I saw my high risk for diabetes, I realized that I didn't know as much as I thought.”

-Sarah,  
Marketing consultant

## 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 >](#)



## Family History

Start learning about family history

[Next](#)

[What We Offer](#)
[Genetics & Health](#)
[For Physicians](#)
[About Us](#)
[Try Demo](#)
[Overview](#)
[DNA basics](#)
[Family history](#)
[Taking action](#)
[Prevention](#)
[Work with your doctor](#)
[Personalized health](#)
[FAQs](#)
[Terminology](#)

Receive our newsletter



From our blog:

"If everyone with a risk factor for heart attack or stroke – 78 percent of the American adult population– got serious about prevention, it would boost the average life expectancy by 1.3 years."

[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

What We Offer

Genetics & Health

For Physicians

About Us

Order Now

Overview

How it works

Our genetic testing

Why Navigenics

Genetic counseling

Conditions we cover

Our policies

FAQs

Success stories

Request information

Tell a friend



"Crohn's disease was really high on my Navigenics results, so I made an appointment with a physician. I've made changes to my diet and now I feel better than I have in years."

*Sarah, Teacher*

## Comprehensive genetic testing services: \$999

Get Started Now

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.

Receive our newsletter

email address

Sign Up


### Success story:

"With this test, I learned what medical information out there really applies to me."

-Aaron,  
Internet entrepreneur

# DNAdirect: Clinical Genetic Testing





*Guidance & Decision Support  
for Genomic Medicine*

[Register or Sign in](#)

Consumers

Healthcare Providers


Healthcare Plans


Laboratories

About Us

**Tests & Services:**

- Screening Tests
- Testing for Genetic Disorders
- Testing for Drug Response
- Genetic Consultation Services
- DNA Storage
- Paternity & Family Tests


 [Contact Customer Support](#)




00:00 / 00:00

**PATIENT STORIES**


Listen to real patients talk about their experience with DNA Direct



**Lisa**  
At risk for Breast & Ovarian Cancer?



**Ken Graham**  
El Camino Hospital CEO



**Millie**  
Is Hemochromatosis Hereditary?

**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](mailto:expert@dnadirect.com)) or call 1-877-646-0002 to speak with a genetic counselor to find out

**FEATURED TOPIC**

Breast & Ovarian Cancer Risk

**RELATED ARTICLES:**

- Who Should Consider Testing
- What Testing Tells You
- How Testing Works
- If You Are At Risk
- If You Have a Known Family Mutation

**ORDER RELATED TESTING:**

- Order Testing

# DNAdirect: Clinical Genetic Testing

**DNAdirect**

*Guidance & Decision Support  
for Genomic Medicine*

Register or Sign in

[Consumers](#) [Healthcare Providers](#) [Healthcare Plans](#) [Laboratories](#) [About Us](#)

[home](#) / [about-dna-direct](#) / [our-solutions](#)



[About DNA Direct](#)

[Our Staff & Advisors](#)

[Our Board of Directors](#)

[Our Standards](#)

[Clinical Labs We Use](#)

[Professional Guidelines](#)

[Our Solutions](#)

[Our Experts](#)

[Our Partners](#)

[DNA Direct in the News](#)

[Press Releases & Statements](#)

[Employment Opportunities](#)

[Privacy Policy](#)

[Customer Comments](#)

## 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.

# Genetic Disorders

**DNAdirect**

Guidance & Decision Support  
for Genomic Medicine

Register or Sign in Search the Website

Consumers

Healthcare Providers

Healthcare Plans

Laboratories

About Us

**Tests & Services:**

Screening Tests

Testing for Genetic Disorders

- › Alpha-1 Antitrypsin Deficiency
- › Ashkenazi Jewish Carrier Testing
- › Blood Clotting Disorders
- › Breast & Ovarian Cancer Risk
- › Cystic Fibrosis
- › Hemochromatosis
- › Infertility
- › Recurrent Pregnancy Loss

Testing for Drug Response

Genetic Consultation Services

DNA Storage

Paternity & Family Tests

Contact Customer

Support

**Testing for Genetic Disorders**

home / testing for genetic disorders



FEATURED ARTICLE

## 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:

A HUMAN TOUCH

### Our Genetic Experts

Genetic tests personalized to you backed by our expert insights.



Meet Stephen Brown, M.D.

Medical Director

[+ Learn More](#)

OUR VOICE

### DNA Direct Blog

DNA Direct's Founder and CEO Reflects on Company's Growth

DNA Spotlight

DNA Archive™

# DNA Direct Clinical Labs

**DNAdirect**

Guidance & Decision Support  
for Genomic Medicine

Register or Sign in

Search the Website

Consumers

Healthcare Providers

Healthcare Plans

Laboratories

About Us

home / about-dna-direct / clincallab-dna.dnadirect



About DNA Direct

Our Staff & Advisors

Our Board of Directors

Our Standards

Clinical Labs We Use

Professional Guidelines

Our Solutions

Our Experts

Our Partners

DNA Direct in the News

Press Releases & Statements

Employment Opportunities

Privacy Policy

Customer Comments

Contact Us

## 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); 34DO906071 (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 LM00049HP; 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: 34DO954530
- American Association of Blood Banks 00030; American Society of Histocompatibility and Immunogenetics 07-3-NC-10-1; Florida 800020370; Forensic Quality Services for Convicted Offender DNA Databasing Laboratories 04-TR-46; Forensic Quality Services International ISO/IEC 04-TR-46; Maryland 1282; 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: 34DO655205