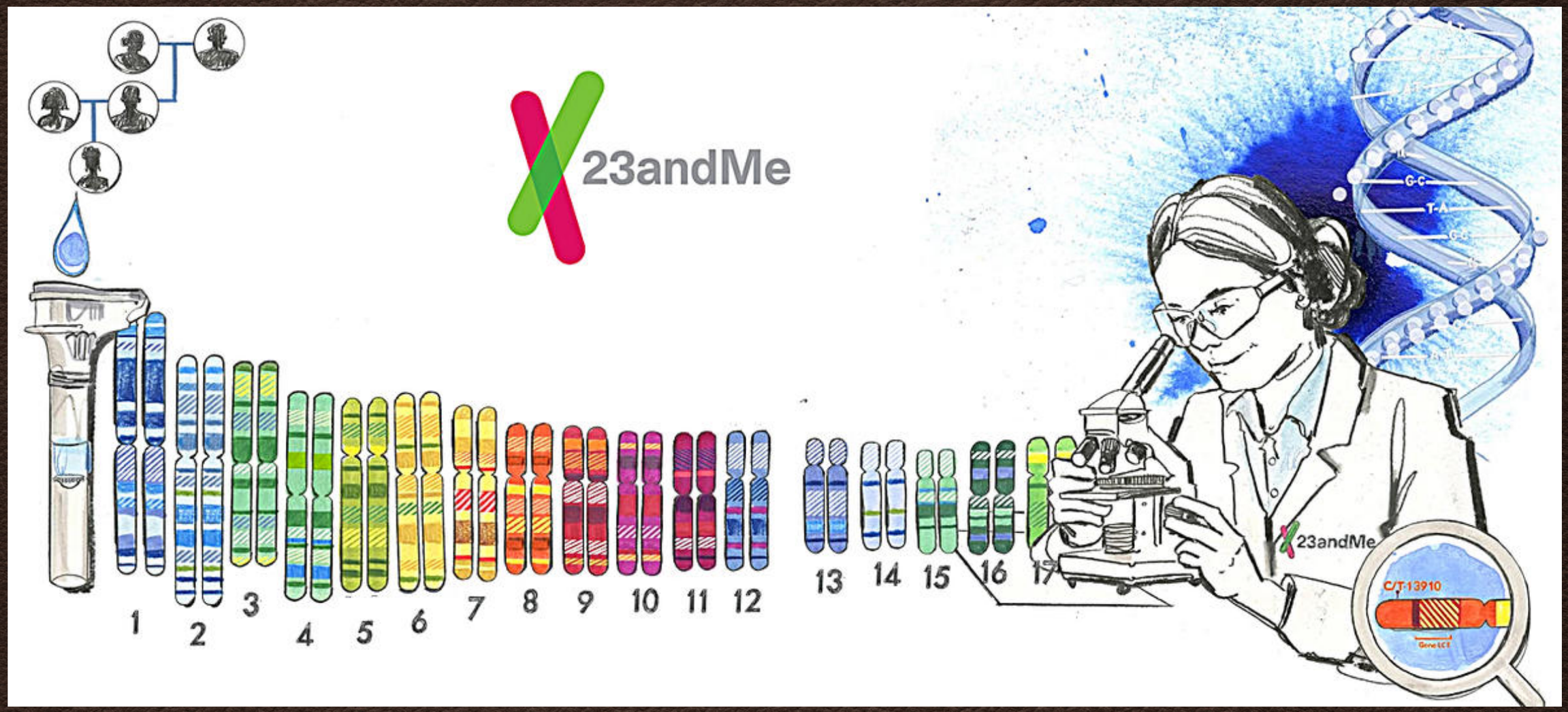
# 23andMe - DNA (Medical)




By Lee Macklin

2018 AHSGR and GRHS National Conventions

# 23andMe

- 23andMe is one of the four largest companies where consumers can have their DNA tested. They provide two types of DNA results
  - Family History DNA (Ethnicity, DNA Matches)
  - Medical (Risks, Traits, Genes)
- This presentation ***primarily*** focuses on the Medical DNA results

# Genealogy - Ethnicity

## Why are my results different?

- Number of Reference Panels (or Reference Populations)
  - People who have taken the DNA test **and** have their actual family history very well documented (**and** validated) for many generations.

- 23andme https://customercare.23andme.com/hc/en-us/articles/212169298-Reference-Populations

- Ancestry.com https://www.ancestry.com/cs/dna-help/ethnicity/reference-panel

- Independent Comparison https://cruwys.blogspot.com/2015/05/comparing-admixture-results-from.html

# Genealogy - Ethnicity

## Why are my results different?

- Geography Breakdown
- 23andMe - 1,000 Locations
- Ancestry - 350 Locations

# 23andMe - 1,000 Regions

## Discover More About Your Ancestry Than Ever Before


Our scientists are committed to continually improving the accuracy and specificity of your ancestry results. To celebrate the new year, we're providing the most refined view of your ancestry we've ever been able to offer.

View your results →

### 1000+ Geographic Regions Now Included in Ancestry Composition

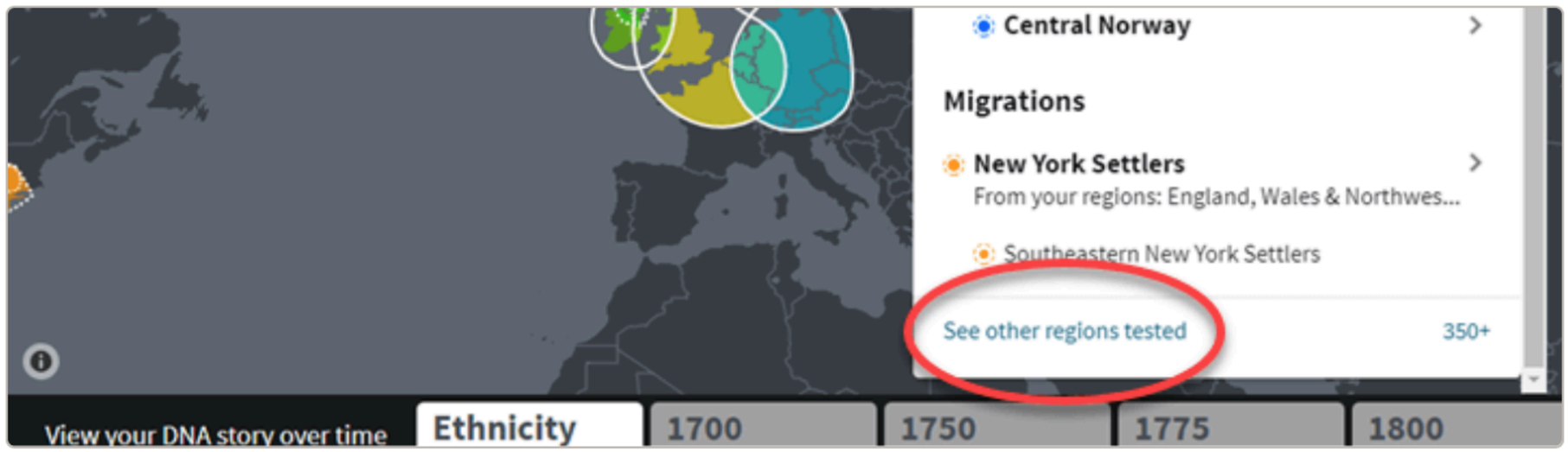
With 33 new Ancestry Detail Reports, we've dramatically increased the coverage of the popular Ancestry Composition analysis. View interactive maps of your ancestry by region, beautiful photographs, and stories about culture, language, food, and more.

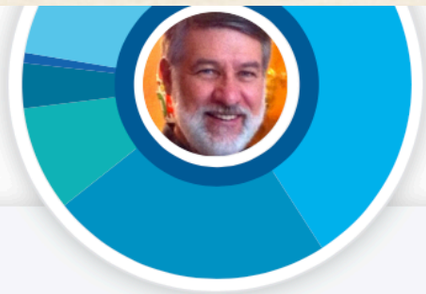
# Ancestry - 350 Regions

## List of DNA Regions

More than 350 global regions make up the ethnicities displayed in our DNA test. As DNA science improves, the number of regions we test for (and the countries covered in each region) may change.

This article lists each region, but to see which areas of the globe are included in the regions, you'll need to view the list from your DNA Story page (which will highlight an area of the map when you click a region). To see all the regions, click **See other regions tested** at the bottom of your ethnicity estimate and click on a region on the next page.





## Lee Macklin 100%

**European 99.3%**

- French & German 40.9%
  - Germany
- British & Irish 23.4%
  - United Kingdom, Ireland
- Eastern European 8.7%
  - Poland
- Scandinavian 3.7%
- Italian 0.8%
- Broadly Northwestern European 17.6%
- Broadly Southern European 0.4%
- Broadly European 3.7%

23andMe

## Ethnicity Estimate

Updates

- England, Wales & Northwestern Europe 40%
- Germanic Europe 30%
  - Germany & the Midwestern United States
    - Northwest Germany & the Midwest
- Eastern Europe & Russia 10%
- Norway 8%
- Baltic States 5%
- Ireland & Scotland 4%
- Sweden 3%

### Migrations

- Ohio River Valley, Indiana, Illinois & Iowa Settlers
  From your regions: England, Wales & Northweste…
  - Western Ohio, Indiana, Illinois & Southeast Iowa Settlers

Ancestry

# DNA Matches

**Ancestry family trees are integrated with DNA Matches.**

**23andMe has a partnership with MyHeritage.**

MyHeritage

**Welcome back Lee,**

You are a member of 2 family sites. Which one do you want to go to?

**MyHeritage Trees NOT integrated with 23andMe DNA Matches**

**Ramsey Web Site**

Managed by Mark Ramsey
26 members, 343 photos
One family tree with 27183 people
48 visits
Last visited today

Go to this site

**Macklin Family Site (23andMe)**

Managed by Cynthia Macklin
2 members, one photo
One family tree with 57 people
2 visits
Last visited 220 days ago

Go to this site

☐ Don't ask me this again, next time after login go directly to the site I will choose now

# Ancestry Accuracy Over Time

| Location | Genealogy % | Ancestry 2012 | Ancestry % V2 2017 | Ancestry 2018 | 2018 % Range | Actual to 2018 |
|---|---|---|---|---|---|---|
| England, Wales and NE Europe | | | | 73% | 68-100 | 9% high |
| Great Britain | | | 3 | | | |
| Europe West | | | 79 | | | |
| British Isles | 1.56 | 80 | | | | |
| British Isles Inferred | 32.76 | | | | | |
| England and Wales | 9.36 | | | | | |
| Netherlands | 14.04 | | | | | |
| France | 6.24 | | | | | |
| Ireland and Scotland | | | | 2 | 0-4 | 5% low |
| Ireland | 1.56 | | 2 | | | |
| Scottish | 5.46 | | | | | |
| German | 24.96 | | | 25 | 24-26 | exact |
| French and German | | | | | | |
| Italy/Greece | 0 | | 2 | | | |
| Scandinavia | 0 | 12 | 11 | | | |
| North Africa | | | 1 | | | |
| Native | <1 | | <1 | | | missing |
| East Asia | | | <1 | | | |
| Unknown/Uncertain | 3.9 | 8 | | | | |
| Total | 99.84 | | 98 | 100 | | |
| | | | | | | |
| Yellow - England, Wales, NE Europe | 63.96 | 80 | 82 | 73 | | |
| Blue - Ireland & Scotland | 7.02 | 0 | 2 | 2 | | |
| Green - Germanic | 24.96 | | 0 | 25 | | |
| Red - Missing or Reassigned Today | 3.9 | 20 | 14+ | 0 | | |

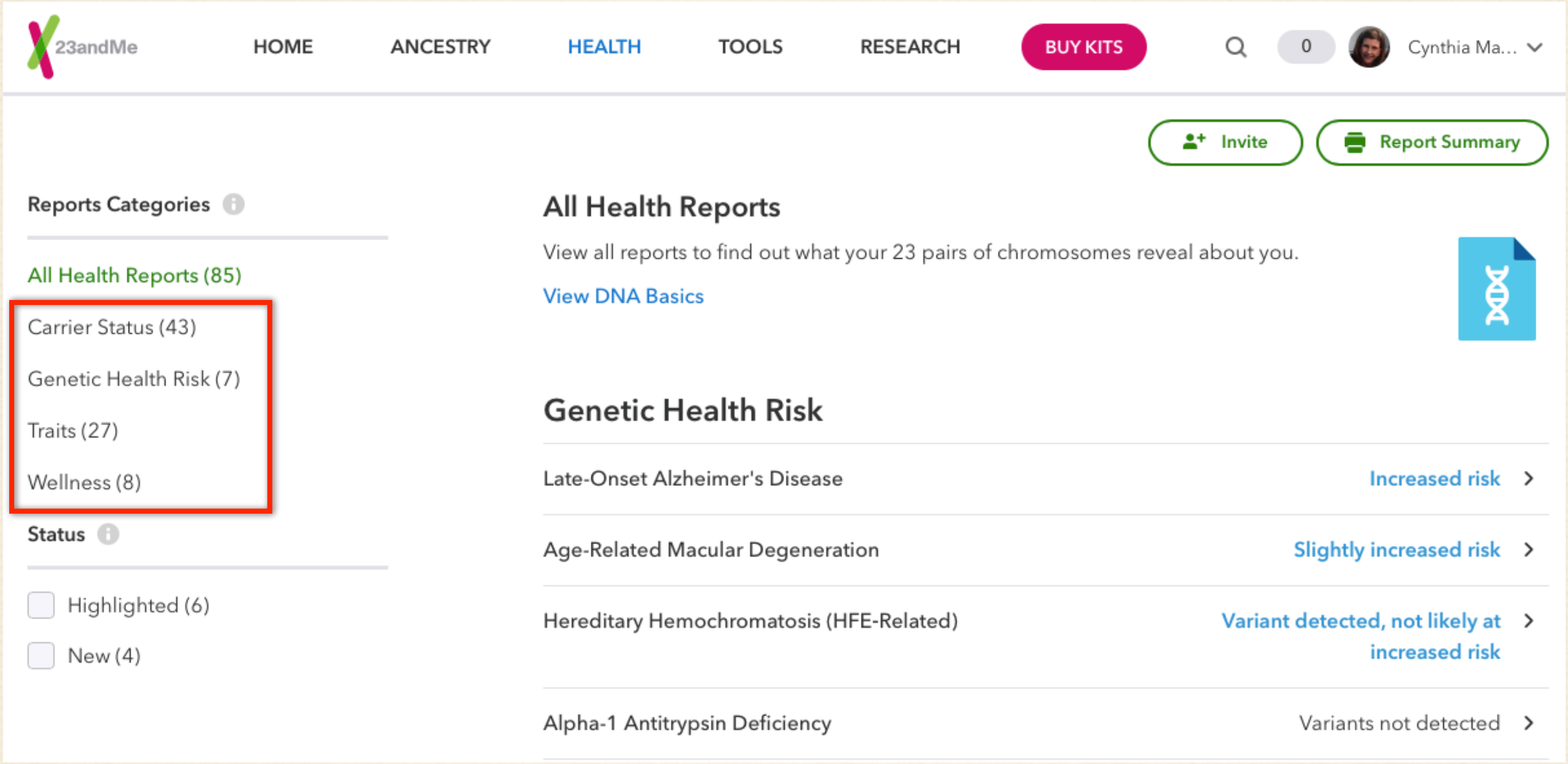
**The unknown 3.9% is most likely split across the British Isles (blue and yellow)**

**Source: DNAeXplained - Genetic Genealogy https://dna-explained.com/2018/09/13/ancestry-2018-ethnicity-update/**

# 23andMe - Medical DNA

- **Health Reports**:
  - Genetic Health Risks (you are at risk for these diseases)
  - Carrier Status (you may pass these mutated genes to your children)
  - Wellness (how your DNA may affect your body's response to diet, exercise and sleep)
  - Traits (the genetics behind your appearances and senses)

# 23andMe - Health Tab




Note: the number of health reports changes frequently as 23andMe expands their medical DNA offerings.

# 23andMe
# Carrier Status Reports

Which specific genetic variants that may not affect your health, but could affect your children's health

## Carrier Status

| | |
|---|---|
| ARSACS | Variant not detected > |
| Agenesis of the Corpus Callosum with Peripheral Neuropathy | Variant not detected > |
| Autosomal Recessive Polycystic Kidney Disease | Variant not detected > |
| Beta Thalassemia and Related Hemoglobinopathies | Variant not detected > |
| Bloom Syndrome | Variant not detected > |
| Canavan Disease | Variant not detected > |
| Congenital Disorder of Glycosylation Type 1a (PMM2-CDG) | Variant not detected > |
| Cystic Fibrosis | Variant not detected > |
| D-Bifunctional Protein Deficiency | Variant not detected > |
| Dihydrolipoamide Dehydrogenase Deficiency | Variant not detected > |

| | |
|---|---|
| Familial Dysautonomia | Variant not detected > |
| Familial Hyperinsulinism (ABCC8-Related) | Variant not detected > |
| Fanconi Anemia Group C | Variant not detected > |
| GRACILE Syndrome | Variant not detected > |
| Gaucher Disease Type 1 | Variant not detected > |
| Glycogen Storage Disease Type Ia | Variant not detected > |
| Glycogen Storage Disease Type Ib | Variant not detected > |
| Hereditary Fructose Intolerance | Variant not detected > |
| Herlitz Junctional Epidermolysis Bullosa (LAMB3-Related) | Variant not detected > |
| Leigh Syndrome, French Canadian Type | Variant not detected > |
| Limb-Girdle Muscular Dystrophy Type 2D | Variant not detected > |
| Limb-Girdle Muscular Dystrophy Type 2E | Variant not detected > |

| | |
|---|---|
| Limb-Girdle Muscular Dystrophy Type 2I | Variant not detected > |
| MCAD Deficiency | Variant not detected > |
| Maple Syrup Urine Disease Type 1B | Variant not detected > |
| Mucolipidosis Type IV | Variant not detected > |
| Neuronal Ceroid Lipofuscinosis (CLN5-Related) | Variant not detected > |
| Neuronal Ceroid Lipofuscinosis (PPT1-Related) | Variant not detected > |
| Niemann-Pick Disease Type A | Variant not detected > |
| Nijmegen Breakage Syndrome | Variant not detected > |
| Nonsyndromic Hearing Loss and Deafness, DFNB1 (GJB2-Related) | Variant not detected > |
| Pendred Syndrome and DFNB4 Hearing Loss (SLC26A4-Related) | Variant not detected > |
| Phenylketonuria and Related Disorders | Variant not detected > |

# Maple Syrup Urine Disease Type 1B

MSUD 1B is a rare genetic disorder. It is characterized by poor growth and feeding, slowed mental and physical processes, and urine with a distinct, sweet odor. A person must have two variants in the BCKDHB gene in order to have this condition.

## Cynthia, you **do not have the variants** we tested.

You could still have a variant not covered by this test.

**0 variants detected**

in the BCKDHB gene

| | |
|---|---|
| Primary Hyperoxaluria Type 2 | Variant not detected › |
| Rhizomelic Chondrodysplasia Punctata Type 1 | Variant not detected › |
| Salla Disease | Variant not detected › |
| Sickle Cell Anemia | Variant not detected › |
| Sjögren-Larsson Syndrome | Variant not detected › |
| Tay-Sachs Disease | Variant not detected › |
| Tyrosinemia Type I | Variant not detected › |
| Usher Syndrome Type 1F | Variant not detected › |
| Usher Syndrome Type 3A | Variant not detected › |
| Zellweger Syndrome Spectrum (PEX1-Related) | Variant not detected › |

# 23andMe
# Genetic Health Risk Reports

Specific genetic variances that can influence your risk for certain health conditions

## Genetic Health Risk

| | |
|---|---|
| Late-Onset Alzheimer's Disease | Increased risk > |
| Age-Related Macular Degeneration | Slightly increased risk > |
| Hereditary Hemochromatosis (HFE-Related) | Variant detected, not likely at increased risk > |
| Alpha-1 Antitrypsin Deficiency | Variants not detected > |
| Celiac Disease | Variants not detected > |
| G6PD Deficiency | Variant not detected > |
| Hereditary Thrombophilia | Variants not detected > |

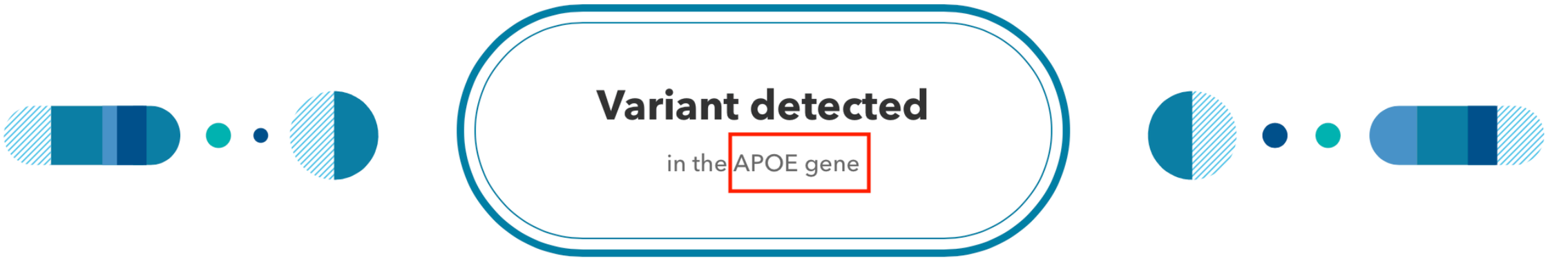
# Late-Onset Alzheimer’s Detected

## Late-Onset Alzheimer's Disease

Alzheimer's disease is characterized by memory loss, cognitive decline, and personality changes. Late-onset Alzheimer's disease is the most common form of Alzheimer's disease, developing after age 65. Many factors, including genetics, can influence a person's chances of developing the condition. This test includes the most common genetic variant associated with late-onset Alzheimer's disease.

Cynthia, you have **two copies** of the ε4 variant we tested.

People with this result have an increased risk of developing late-onset Alzheimer's disease. Lifestyle, environment, and other factors can also affect your risk.



# Other Risk Factors

## Lifestyle and other factors can also influence the chances of developing late-onset Alzheimer's disease.

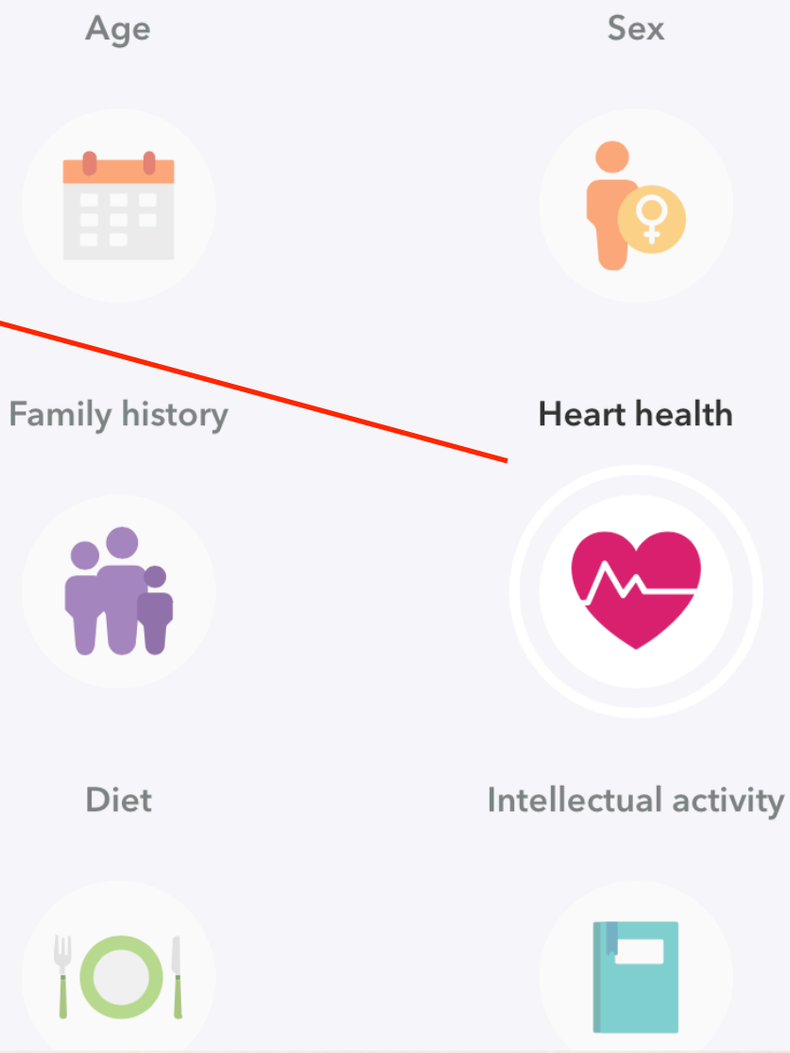
Consult with a healthcare professional before making any major lifestyle changes.

### Heart health

Research shows that high blood pressure and high cholesterol are both associated with an increased risk for late-onset Alzheimer's disease.

Maintaining normal blood pressure, keeping a healthy weight, and exercising regularly are a few things you can do to promote and maintain your heart health.

See Scientific Details for more information



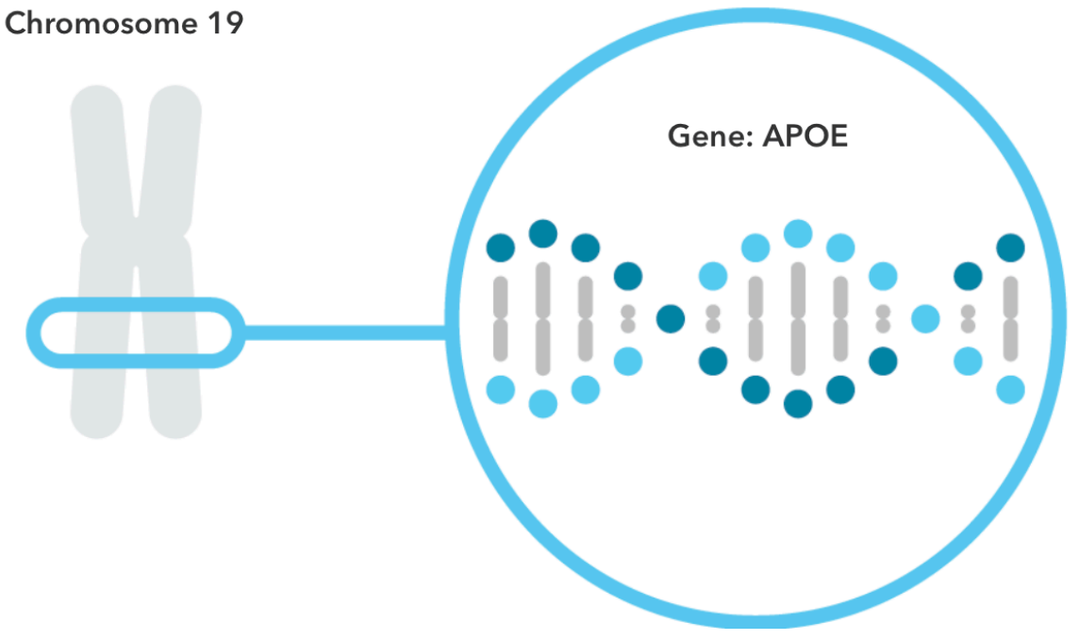
# APOE Gene Purpose

The ε4 variant in the APOE gene is the most common genetic factor associated with late-onset Alzheimer's disease.

APOE

The APOE gene contains instructions for making a protein called apolipoprotein E. This protein helps control the levels of cholesterol and fats in the blood. It is not known exactly how the ε4 variant increases the risk of late-onset Alzheimer's disease.

**Read more at** Genetics Home Reference



# APOE Variant Details

You have two copies of the ε4 variant we tested.

Variants Detected | View All Tested Markers

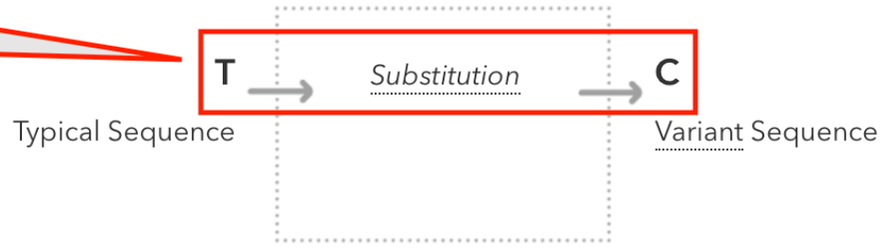
| Marker Tested | Genotype* | Additional Information |
|---|---|---|
| **ε4**<br>**Gene:** APOE<br>**Marker:** rs429358 | **C** Variant copy from one of your parents<br>**C** Variant copy from your other parent | |

**Biological explanation**

The variant tested is a change from a T to a C in the DNA sequence of the APOE gene. Two genetic markers (rs429358 and rs7412) in the APOE gene are often used together to define three variants of the APOE gene called ε2, ε3, and ε4. However, the rs429358 marker by itself can be used to identify the ε4 variant.

**NOTE: there are 7 possible types of mutations**

**Typical vs. variant DNA sequence(s)**



Scientific Details | Frequently Asked Questions

| Genotype | Sex | Age 65 | Age 75 | Age 85 |
|---|---|---|---|---|
| General population | Men | <1% | 3% | 11% |
| General population | Women | <1% | 3% | 14% |
| No ε4 variants | Men | <1% | 1-2% | 5-8% |
| No ε4 variants | Women | <1% | 1-2% | 6-10% |
| One copy of ε4 variant | Men | 1% | 4-7% | 20-23% |
| One copy of ε4 variant | Women | <1% | 5-7% | 27-30% |
| Two copies of ε4 variant | Men | 4% | 28% | 51% |
| Two copies of ε4 variant | Women | 2% | 28% | 60% |

# Stanford University Alzheimer's Research Study

- 23and Me shared Cindy's DNA results with this Stanford University group (DNA settings)

- They contacted Cindy stating they have an active research program underway developing a new Alzheimer's drug and invited Cindy to participate in the five year program

# 23andMe
# Wellness Reports

## How your DNA may affect your body's response to diet, exercise, and sleep

## Wellness

| | |
|---|---|
| Alcohol Flush Reaction | Unlikely to flush › |
| Caffeine Consumption | Likely to consume less › |
| Deep Sleep | Less likely to be a deep sleeper › |
| Genetic Weight | Predisposed to weigh less than average › |
| Lactose Intolerance | Likely tolerant › |
| Muscle Composition | Common in elite power athletes › |
| Saturated Fat and Weight | Likely similar weight › |
| Sleep Movement | Likely more than average movement › |

# Caffeine Consumption

What makes some people caffeine fanatics, while others go easy on the java? Genetic factors help explain how much caffeine people tend to consume.

Your Wellness Result

Cynthia, based on your genetics, you are likely to drink **slightly less caffeine than average**, if you drink caffeine at all.

23andMe research participants with your genetic result who consume caffeine regularly tend to drink the equivalent of about an eighth of a cup of coffee (17 mg of caffeine) less than average per day. Of course, not everyone chooses to consume caffeine, but for those who do, their genetics may play a role in the amount they consume.

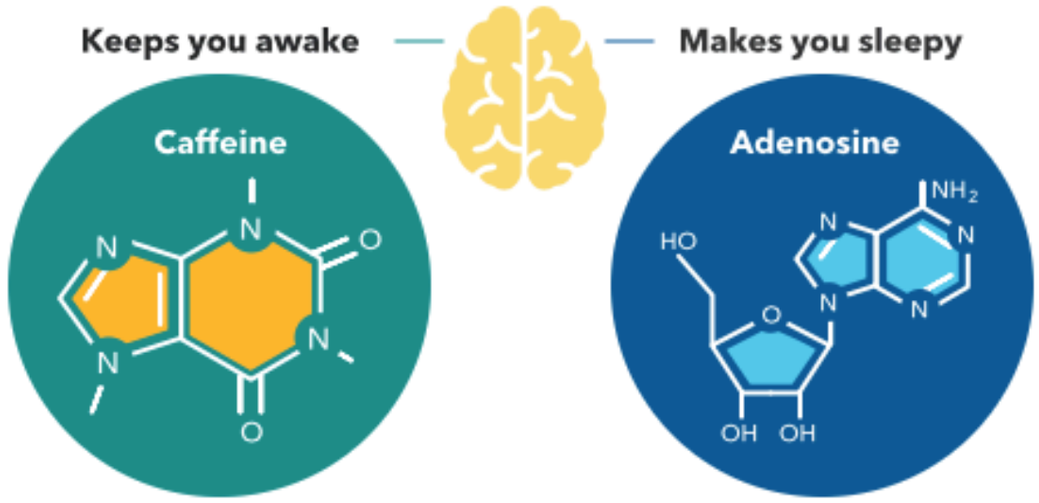
# Genetics and Caffeine

## Genetics

This report is based on genetic variants near two genes that play a role in how your body handles caffeine. The first gene, CYP1A2, contains instructions for an enzyme that breaks down 95% of the caffeine you consume. The second gene, AHR, contains instructions for a protein that ramps up production of the CYP1A2 enzyme. Variants in these genes may affect how quickly the body breaks down and clears away caffeine.


## How does caffeine keep you awake?

Caffeine interferes with the brain system that causes sleepiness. A molecule called adenosine acts as a signal between brain cells to bring on sleepiness. Caffeine blocks adenosine's signals, making you feel more alert. This is also why caffeine can make it hard to fall asleep and interfere with deep sleep.



## Does caffeine enhance performance?

It can feel like coffee makes you smarter, but some research suggests this may be an illusion. People with a daily caffeine habit may actually perform worse than other people on mental and physical tasks – that is, until they get their morning fix. As caffeine is cleared from the body overnight, daily caffeine drinkers start to experience caffeine withdrawal. This leads to worse performance until they have their morning coffee or tea, which reverses the withdrawal. Meanwhile, for people who aren't used to consuming caffeine every day, caffeine may not improve performance much, if at all.

**Side effects of caffeine withdrawal**

Trouble focusing | Headaches | Sleepiness

## How much is too much?

Moderate levels of daily caffeine consumption aren't associated with increased health risks and may even lower the risk for some diseases. If you consume caffeine regularly, current guidelines recommend that healthy adults limit themselves to 400 mg of caffeine per day or less. That's the equivalent of about three 12-oz cups of coffee or eight 8-oz cups of black tea. Keep in mind the exact amount of caffeine in coffee and tea – even decaf – can vary widely depending on how they're made.

**Maximum healthy amount: 400 mg per day**

# 23andMe - Traits Reports

Explore the genetics behind your appearance and senses

## Traits

| | |
|---|---|
| New Ability to Match Musical Pitch | Less likely to be able to match a musical pitch › |
| New Fear of Heights | More likely than average to be afraid of heights › |
| New Hair Photobleaching | More likely to experience hair photobleaching › |
| New Mosquito Bite Frequency | Likely bitten as often as others › |
| Asparagus Odor Detection | *Not determined* › |
| Bitter Taste | Likely can't taste › |
| Cheek Dimples | Likely no dimples › |
| Cilantro Taste Aversion | Slightly higher odds of disliking cilantro › |
| Cleft Chin | Likely no cleft chin › |

| | |
|---|---|
| Earlobe Type | Likely detached earlobes › |
| Earwax Type | Likely wet earwax › |
| Eye Color | Likely blue or green eyes › |
| Finger Length Ratio | Likely ring finger longer › |
| Freckles | Likely little freckling › |
| Hair Texture | Likely straight or wavy › |
| Hair Thickness | Less likely to have thick hair › |
| Light or Dark Hair | Likely light › |
| Misophonia | Average odds of hating chewing sounds › |
| Newborn Hair | Likely little baby hair › |
| Photic Sneeze Reflex | Likely no photic sneeze reflex › |
| Red Hair | Likely no red hair › |

| | |
|---|---|
| Skin Pigmentation | Likely lighter skin › |
| Sweet vs. Salty | Likely prefers sweet › |
| Toe Length Ratio | Likely second toe longer › |
| Unibrow | Likely no unibrow › |
| Wake-Up Time | Likely to wake up around 7:00 am › |
| Widow's Peak | Likely no widow's peak › |

Light color hair, Blue/Green eyes
Exactly what her DNA Traits show!!

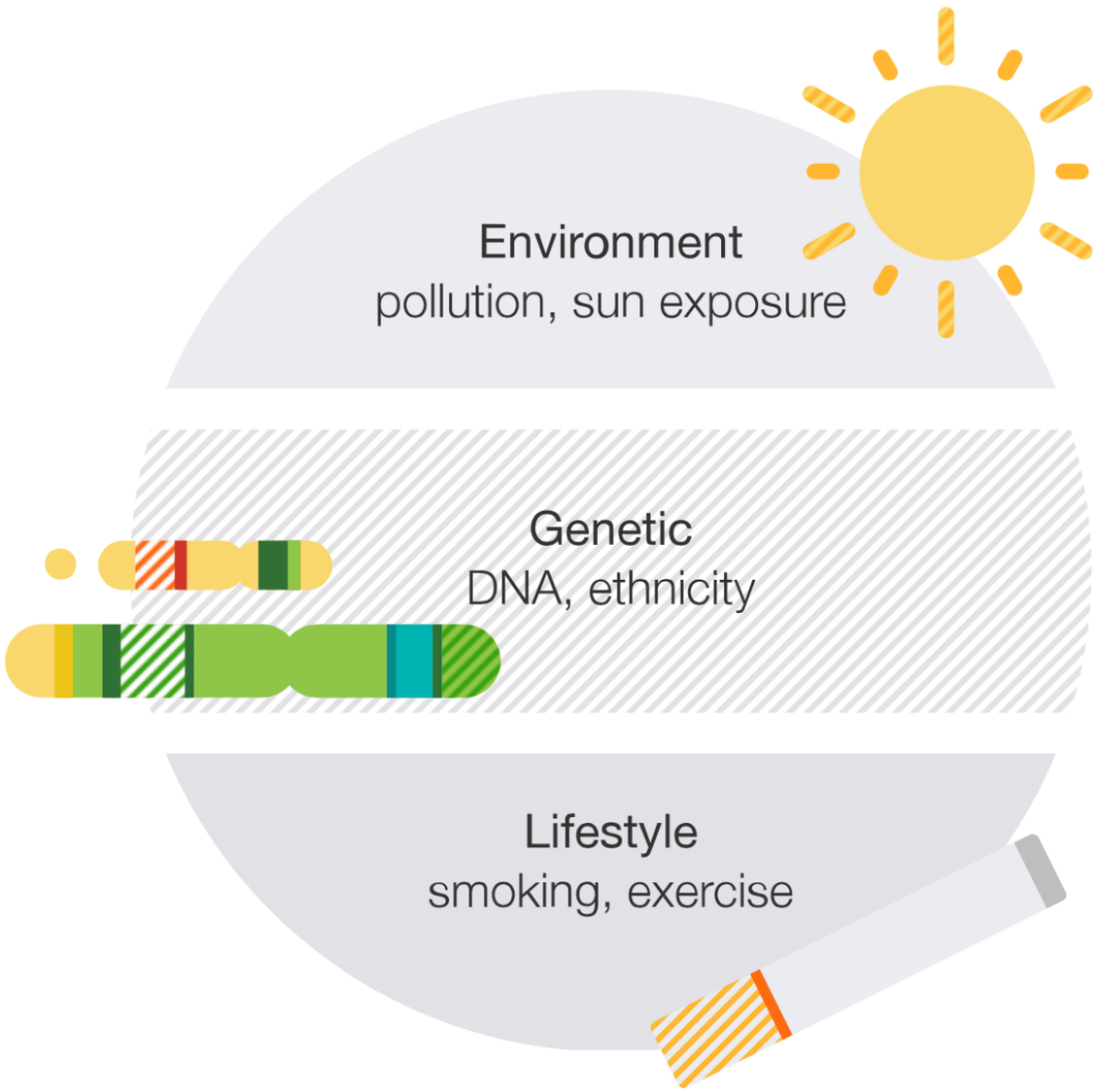
Cindy Macklin

# Consumer DNA Testing Risks

- Should people only learn about their DNA from a doctor or genetic counselor to ensure that the results are clearly and accurately explained?

- Should people only learn about risks for disease that are "medically actionable"? With screening, therapeutics, or lifestyle change may prevent or treat the disease.

# All the Risk Factors




## Why genetics is only part of the story.

When it comes to your health and traits, DNA is only part of the story. Other variables come into play, including non-genetic factors, such as your environment and lifestyle.

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Genetics Home Reference | Your Guide to Understanding Genetic Conditions

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Health Conditions | Genes | Chromosomes & mtDNA | Resources | Help Me Understand Genetics

## Genes

Explore the normal functions of human genes and the health implications of genetic changes.

A B C D E F G H I J K L M N O P Q R

S T U V W X Y Z

AAAS: aladin WD repeat nucleoporin

AASS: aminoadipate-semialdehyde synthase

ABAT: 4-aminobutyrate aminotransferase

ABCA1: ATP binding cassette subfamily A member 1

ABCA3: ATP binding cassette subfamily A member 3

ABCA4: ATP binding cassette subfamily A member 4

ABCA12: ATP binding cassette subfamily A member 12

ABCB4: ATP binding cassette subfamily B member 4

ABCB7: ATP binding cassette subfamily B member 7

ABCB11: ATP binding cassette subfamily B member 11

### Learn More about Genes

What is DNA?

What is a gene?

How do genes direct the production of proteins?

How can gene mutations affect health and development?

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Genetics Home Reference

Your Guide to Understanding Genetic Conditions

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Health Conditions | Genes | Chromosomes & mtDNA | Resources | Help Me Understand Genetics

## Health Conditions

Explore the signs and symptoms, genetic cause, and inheritance pattern of various health conditions.

0 – 9 **A** B C D E F G H I J K L M N O P

Q R S T U V W X Y Z

A-alphalipoprotein Neuropathy, *see* Tangier disease

A-T, *see* Ataxia-telangiectasia

AA, *see* Alopecia areata

AAA, *see* Triple A syndrome

AAA syndrome, *see* Triple A syndrome

AADC deficiency, *see* Aromatic l-amino acid decarboxylase deficiency

Aarskog syndrome, *see* Aarskog-Scott syndrome

Aarskog-Scott syndrome

AAS, *see* Aarskog-Scott syndrome

AASA dehydrogenase deficiency, *see* Pyridoxine-dependent epilepsy

### Learn More about Health Conditions

What does it mean if a disorder seems to run in my family?

What are the different ways in which a genetic condition can be inherited?

What are complex or multifactorial disorders?

What does it mean to have a genetic predisposition to a disease?

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Health Conditions | Genes | Chromosomes & mtDNA | Resources | Help

ALSP, *see* Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia

Alstrom syndrome, *see* Alström syndrome

Alstrom-Hallgren syndrome, *see* Alström syndrome

Alström syndrome

Alternating hemiplegia of childhood

alternating hemiplegia syndrome, *see* Alternating hemiplegia of childhood

alveolar capillary dysplasia, *see* Alveolar capillary dysplasia with misalignment of pulmonary veins

Alveolar capillary dysplasia with misalignment of pulmonary veins

ALX, *see* Alexander disease

alymphoid cystic thymic dysgenesis, *see* T-cell immunodeficiency, congenital alopecia, and nail dystrophy

Alzheimer dementia (AD), *see* Alzheimer disease

Alzheimer disease

Alzheimer sclerosis, *see* Alzheimer disease

Alzheimer syndrome, *see* Alzheimer disease

Alzheimer's Disease, *see* Alzheimer disease

Alzheimer-type dementia (ATD), *see* Alzheimer disease

AMACR deficiency, *see* Alpha-methylacyl-CoA racemase deficiency

amaurosis, Leber congenital, *see* Leber congenital amaurosis

AMCD1, *see* Distal arthrogryposis type 1

AMCX1, *see* X-linked infantile spinal muscular atrophy

AMD, *see* Age-related macular degeneration

AMD, *see* Pompe disease

Amelogenesis imperfecta

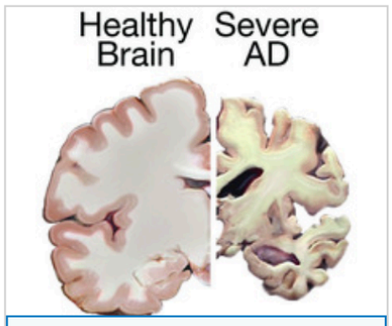
Aminoacylase 1 deficiency

# Alzheimer disease



## Description

Alzheimer disease is a degenerative disease of the brain that causes dementia, which is a gradual loss of memory, judgment, and ability to function. This disorder usually appears in people older than age 65, but less common forms of the disease appear earlier in adulthood.




Enlarge

Memory loss is the most common sign of Alzheimer disease. Forgetfulness may be subtle at first, but the loss of memory worsens over time until it interferes with most aspects of daily living. Even in familiar settings, a person with Alzheimer disease may get lost or become confused. Routine tasks such as preparing meals, doing laundry, and performing other household chores can be challenging. Additionally, it may become difficult to recognize people and name objects. Affected people increasingly require help with dressing, eating, and personal care.

As the disorder progresses, some people with Alzheimer disease experience personality and behavioral changes and have trouble interacting in a socially appropriate manner. Other common symptoms include agitation, restlessness, withdrawal, and loss of language skills. People with this disease usually require total care during the advanced stages of the disease. Affected individuals usually survive 8 to 10 years after the appearance of symptoms, but the course of the disease can range from 1 to 25 years. Death usually results from pneumonia, malnutrition, or general body wasting (inanition).

Alzheimer disease can be classified as early-onset or late-onset. The signs and symptoms of the early-onset form appear before age 65, while the late-onset form appears after age 65. The early-onset form is much less common than the late-onset form, accounting for less than 5 percent of all cases of Alzheimer disease.

## Causes

Most cases of early-onset Alzheimer disease are caused by gene mutations that can be passed from parent to child. Researchers have found that this form of the disorder can result from mutations in one of three genes: *APP*, *PSEN1*, or *PSEN2*. When any of these genes is altered, large amounts of a toxic protein fragment called amyloid beta peptide are produced in the brain. This peptide can build up in the brain to form clumps called amyloid plaques, which are characteristic of Alzheimer disease. A buildup of toxic amyloid beta peptide and amyloid plaques may lead to the death of nerve cells and the progressive signs and symptoms of this disorder.

Some evidence indicates that people with Down syndrome have an increased risk of developing Alzheimer disease. Down syndrome, a condition characterized by intellectual disability and other health problems, occurs when a person is born with an extra copy of chromosome 21 in each cell. As a result, people with Down syndrome have three copies of many genes in each cell, including the *APP* gene, instead of the usual two copies. Although the connection between Down syndrome and Alzheimer disease is unclear, the production of excess amyloid beta peptide in cells may account for the increased risk. People with Down syndrome account for less than 1 percent of all cases of Alzheimer disease.

The causes of late-onset Alzheimer disease are less clear. The late-onset form does not clearly run in families, although clusters of cases have been reported in some families. This disorder is probably related to variations in one or more genes in combination with lifestyle and environmental factors. A gene called *APOE* has been studied extensively as a risk factor for the disease. In particular, a variant of this gene called the e4 allele seems to increase an individual's risk for developing late-onset Alzheimer disease. Researchers are investigating many additional genes that may play a role in Alzheimer disease risk.

**Learn more about the genes associated with Alzheimer disease**

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## APOE gene

apolipoprotein E

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### Normal Function

The *APOE* gene provides instructions for making a protein called apolipoprotein E. This protein combines with fats (lipids) in the body to form molecules called lipoproteins. Lipoproteins are responsible for packaging cholesterol and other fats and carrying them through the bloodstream. Maintaining normal levels of cholesterol is essential for the prevention of disorders that affect the heart and blood vessels (cardiovascular diseases), including heart attack and stroke.

There are at least three slightly different versions (alleles) of the *APOE* gene. The major alleles are called e2, e3, and e4. The most common allele is e3, which is found in more than half of the general population.

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# Questions?

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