

# Incidental Findings: A 23andMe perspective

Joanna Mountain, PhD  
Senior Director of Research

April 30<sup>th</sup>, 2013



# What is 23andMe?

A company that provides:

- ▣ A service to give consumers **access** to their genetic data (+ features and tools to make that data useful and interesting)
- ▣ A platform for genetic **research**

# People care about:

- What might I learn about myself if I sign up for 23andMe?
- What might another person/organization learn about me if I sign up for 23andMe?
- What might another person learn about him- or herself if I sign up for 23andMe?
- What might a third person/organization learn about another person if I sign up for 23andMe?



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# 23andMe's Personal Genome Service

The screenshot shows the 23andMe website interface for user Joanna. The top navigation bar includes links for HOME, MY RESULTS, FAMILY & FRIENDS, and RESEARCH & COMMUNITY, along with a search bar labeled SEARCH 23ANDME. The user's name, Joanna, is displayed in the top right corner. Below the navigation bar, the main content area is divided into two sections: RECOMMENDED FOR YOU and FEATURED CONTENT.

**RECOMMENDED FOR YOU**

- ANCESTRY COMPOSITION:** A circular progress indicator shows 25.8% for BRITISH AND IRISH ancestry.
- HEALTH OVERVIEW:** A light blue square with icons representing various health aspects like a heart, ear, and person.
- SURVEYS FUEL RESEARCH DISCOVERIES:** A green button labeled TAKE SURVEYS is positioned below a document icon with a checkmark.
- NEA ANC:** A partially visible card on the right with a classical figure illustration.

**FEATURED CONTENT**

- Surgical Complications:** A red-bordered card with a red cross icon and a pair of scissors. The text reads: "In the case of a surgical procedure, planned or unplanned, this set of your genetic results and health history information would be important to share with your doctor." Below the text, it states: "BASED ON YOUR 6 GENETIC REPORTS & 12 SURVEY ANSWERS".
- Breast Cancer and You:** A white-bordered card with a red border. The text reads: "Breast cancer touches hundreds of thousands of individuals every year. Men women, old, young – no one is fully exempt. We will guide you through the factors that are most relevant to you." Below the text, it states: "BASED ON YOUR 3 GENETIC REPORTS & 11 SURVEY".

# 23andMe's Personal Genome Service



## Health Overview

 PRINT  
 SH

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

SHOW RESULTS FOR Joanna Mountain [SEE NEW AND RECENTLY UPDATED REPORTS >](#)

### Health Risks (120) ?

**↑ ELEVATED RISKS**

	YOUR RISK	AVERAGE RISK
Lung Cancer	8.6%	6.2%
Restless Legs Syndrome	5.2%	4.2%
Exfoliation Glaucoma	2.9%	1.0%
Scleroderma (Limited Cutaneous Type)	0.20%	0.16%
Esophageal Squamous Cell Carcinoma (ESCC)	0.09%	0.07%

[See all 120 risk reports...](#)

### Inherited Conditions (50) ?

REPORT	RESULT
Alpha-1 Antitrypsin Deficiency	Variant Present
Hemochromatosis (HFE-related)	Variant Present
Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)	Variant Absent
Autosomal Recessive Polycystic Kidney Disease	Variant Absent
ARSACS	Variant Absent
Beta Thalassemia	Variant Absent
Bloom's Syndrome	Variant Absent
BRCA Cancer Mutations (Selected)	Variant Absent

[See all 50 carrier status...](#)

### Traits (57) ?

REPORT	RESULT
Alcohol Flush Reaction	Does Not Flush
Bitter Taste Perception	Unlikely to Taste
Earwax Type	Wet
Eye Color	Likely Brown
Hair Curl ✦	Slightly Curlier Hair on Average

[See all 57 traits...](#)

### Drug Response (21) ?

REPORT	RESULT
Abacavir Hypersensitivity	Typical
Alcohol Consumption, Smoking and Risk of Esophageal Cancer	Typical
Clopidogrel (Plavix®) Efficacy	Typical
Oral Contraceptives, Hormone Replacement Therapy and Risk of Venous Thromboembolism ♀	Typical
Fluorouracil Toxicity	Typical

[See all 21 drug response...](#)

# 23andMe's Personal Genome Service

## Health Risks

### Venous Thromboembolism

#### Venous Thromboembolism

**Venous thromboembolism** (VTE) encompasses two related conditions: **deep vein thrombosis** or DVT, is the formation of a blood clot in the body, usually in the legs. The second, **pulmonary embolism**, is a blood clot that breaks free and travels through the circulatory system to the lungs. PE always precedes PE. It is estimated that about 250,000 people

are diagnosed with venous thromboembolism in the United States each year, but the incidence is probably much higher as many cases go undiagnosed. Pulmonary embolism is potentially life threatening if prompt medical attention is not received. Therefore, recognizing the symptoms of venous thromboembolism and avoiding risk factors is of paramount importance.

The following results are based on ★★★★★ **Established Research** for 3 reported markers, updated **March 29th, 2012**.



1 of 3. The ability of blood to clot prevents it from flowing out of your body uncontrollably in the event of an injury.

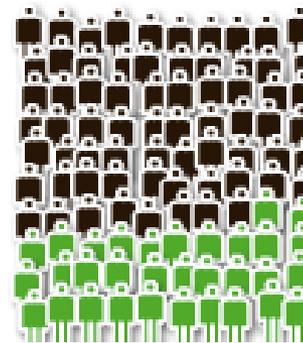
# 23andMe's Personal Genome Service

## Health Risks

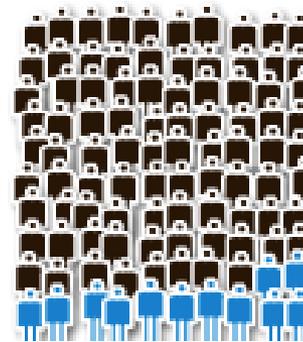
### Venous Thromboembolism

#### Genes vs. Environment

The **heritability** of **venous thromboembolism** is estimated to be 55%. This means that genetics (including unknown factors and known ones such as the SNPs we describe here) and environment play nearly equal roles in this condition. There are a number of **environmental factors** of various strengths that contribute to venous thromboembolism. Strong risk factors include hip or leg fractures, hip or knee replacement, major surgery or trauma, and spinal cord injury or surgery. Moderate risk factors include arthroscopic knee surgery, having central venous lines, congestive heart or respiratory failure, hormone replacement or oral contraceptive use, cancer, pregnancy, paralytic stroke, previous venous thromboembolism, and thrombophilia. Weak risk factors include bed rest for more than three days, immobility due to sitting (such as a long car or plane trip), specific types of chemotherapy, increasing age, laparoscopic surgery, obesity, and varicose veins. ([sources](#))

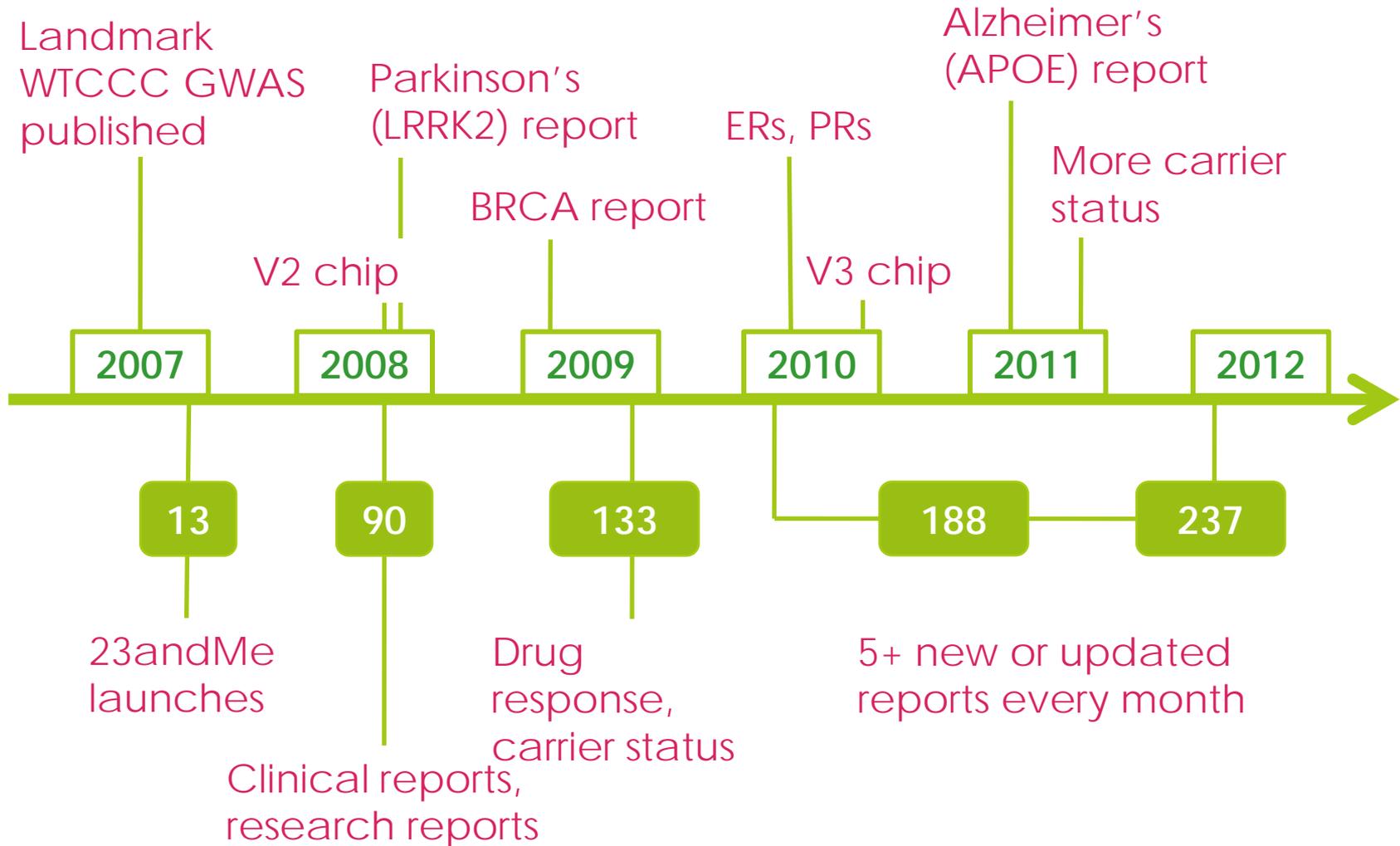


**32.5 out of 100**  
men of European ethnicity who share \_\_\_\_\_ genotype will develop Venous Thromboembolism between the ages of 0 and 79.



**Average**  
**12.3 out of 100**  
men of European ethnicity will develop Venous Thromboembolism between the ages of 0 and 79.

# Health and traits reports over time



# 23andMe's Terms of Service



## 5. Risks and Considerations Regarding 23andMe Services

**Once you obtain your Genetic Information, the knowledge is irrevocable.** You should not assume that any information we may be able to provide to you, whether now or as genetic research advances, will be welcome or positive. You should also understand that as research advances, in order for you to assess the meaning of your DNA in the context of such advances, you may need to obtain further services from 23andMe or from your physician or other health care provider.

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# Who learns what?

Disease	Effect/result	Percentage
Venous thromboembolism*	F5 or F2 mutation	8.2%
AMD	4-fold average risk (28%)	5.6%
Cystic Fibrosis*	DeltaF508 mutation	2.5%
Prostate cancer	double average risk (36%)	1.0%
Alzheimer's disease*	At least one APOE e4 copy	25%
Parkinson's*	LRRK2 G2019S mutation	0.5%
Crohn's	5-fold average risk	5%
Fluorouracil*	toxic reaction	1.5%
Plavix*	greatly reduced efficacy	5%

\* 23andMe's custom chip only

# “Locked” reports

## Health Risks (20, 2 locked reports) ?

### ↑ ELEVATED RISKS

	YOUR RISK	AVERAGE RISK
Type 2 Diabetes	32.3%	25.7%
Restless Legs Syndrome	2.5%	2.0%
Exfoliation Glaucoma	2.2%	0.7%
Ulcerative Colitis	1.2%	0.8%
Esophageal Squamous Cell Carcinoma (ESCC)	0.43%	0.36%

## Inherited Conditions (50, 2 locked reports) ?

### REPORT

### RESULT

Hemochromatosis (HFE-related)	Variant Present
Alpha-1 Antitrypsin Deficiency	Variant Absent
Bloom's Syndrome	Variant Absent

# “Locked” reports

The screenshot shows a 'Health Risks' section with a sub-header '(120, 2 locked reports)'. Below this, there is a red arrow icon followed by the text 'ELEVATED RISKS'. A list of conditions follows: 'Type 2 Diabetes', 'Restless Legs Syndrome', 'Exfoliation Glaucoma', 'Ulcerative Colitis', and 'Esophageal Squamous C (ESCC)'. A tooltip box is overlaid on the right side of the list, titled 'Locked Reports'. The tooltip contains the following text: 'Established Research reports for the following conditions will not initially display your results, **regardless of whether they indicate your risk is high, low, or typical.** Click the name of the report to read it without seeing your results.' Below this text, two conditions are listed: 'Alzheimer's Disease' and 'Parkinson's Disease'. At the bottom of the tooltip, it says 'The report will tell you how to make your results visible'.

Reports on genetic variants that have particularly strong impact on disease risk

# “Locked” reports

## Your Genetic Data

Show information for



assuming

European

ethnicity

and an age range of

30-79

Why are there limited choices of ethnicity in risk reports?

Your results do not affect whether you see the text below. Everyone must view this information before choosing whether to view their results for this report.

**Parkinson's Disease** is a serious disease with no known cure for which strong genetic factors have been established. Consider the following before choosing whether to view your genetic data regarding Parkinson's Disease:

- **Genetics can substantially affect your Parkinson's risk:** This report includes information on a relatively rare mutation in the LRRK2 gene associated with significantly increased risk in European populations, in addition to other variants with relatively smaller effects in both European and Asian populations.
- **Your family history affects your chances of having the LRRK2 mutation:** Though rare in the general population, this mutation is much more common in families with European ancestry and a history of Parkinson's.
- **These genetic variants cannot predict definitively whether you will develop Parkinson's:** Genes and environment both contribute to a person's chances of developing Parkinson's. Many people who have the risk-associated versions of the genetic variants in this report will never get the disease. Conversely, lacking these versions does not substantially reduce one's Parkinson's risk below average.
- **This information may have implications for your relatives:** Because you are genetically similar to your relatives, anything you learn about your own genes may have implications for them as well.
- **The significance of your genetic information could change:** The development of new treatments or cures could substantially change the implications of this information. New discoveries could refine our understanding of the risks associated with certain genotypes or link them to additional diseases or conditions.

I understand, please show me my results

# “Locked” reports

## disease risk

### Alzheimer's Disease ★★★★★ ?

Established Research report on 1 reported marker.

*Alzheimer's Disease is a serious disease with no known cure for which a strong genetic factor has been established. [View the informational video](#) below and consider the following before choosing whether to view your genetic data regarding Alzheimer's disease:*

I understand, please show me my results

VIDEO: The APOE Gene and Alzheimer's Disease

[Close video](#)



# “Locked” reports

Inherited Conditions (50, 2 locked reports) ?

## Locked Reports

Established Research reports for the following conditions will not initially display your results, **regardless of whether they indicate your risk is high, low, or typical.**

Click the name of the report to read it without seeing your results.

[BRCA Cancer Mutations \(Selected\)](#)

[TTR-Related Familial Amyloid](#)

[Polyneuropathy](#)

The report will tell you how to make your results visible

Reports on genetic variants that have particularly strong impact on disease risk

# “Locked” reports

Learn more about the biology of Breast/Ovarian Cancer...

## Your Genetic Data

Who	What It Means
	<p>Has not opted in to view results for this report. <a href="#">Click here to learn more.</a></p>
	<p>Has the 185delAG BRCA1 mutation. Lifetime risk of breast cancer for women is increased from 12% to about 60% and risk of ovarian cancer is increased from less than 2% to about 40%. May significantly increase risk of prostate cancer in men. There is also an increased risk for breast cancer in men.</p>
	<p>Has the 5382insC BRCA1 mutation. Lifetime risk of breast cancer for</p>

# “Locked” reports

PeerJ

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PDF



Introduction

Participants and Methods

Results

Discussion

Supplemental Information

Additional Information and Declarations

Peer Review history

Subject areas

Genetics

Genomics

Medical Genetics

Ethical Issues

1,716

Unique visitors

2,451

Pageviews

## Dealing with the unexpected: consumer responses to direct-access *BRCA* mutation testing

Uta Francke <sup>1,2</sup>, Cheri Dijamco<sup>1</sup>, Amy K. Kiefer<sup>1</sup>, Nicholas Eriksson<sup>1</sup>, Bianca Moiseff<sup>1</sup>, Joyce Y. Tung<sup>1</sup>, Joanna L. Mountain<sup>1</sup>

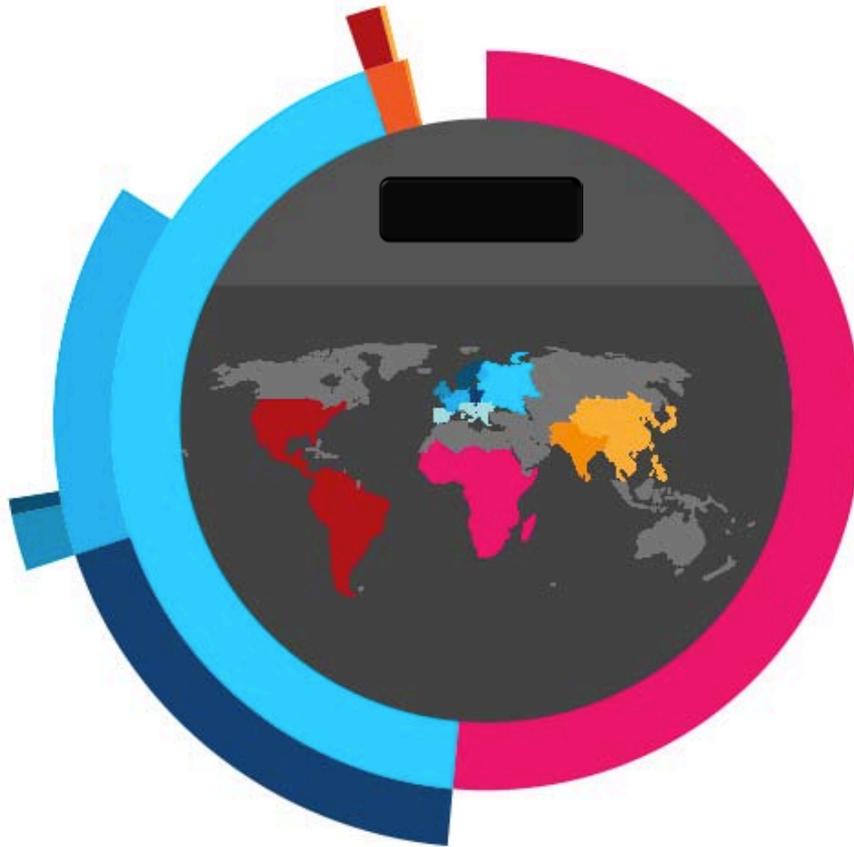
› Author and article information

▼ Abstract

**Background.** Inherited *BRCA* gene mutations convey a high risk for breast and ovarian cancer, but current guidelines limit *BRCA* mutation testing to women with early-onset cancer and relatives of mutation-positive cases. Benefits and risks of

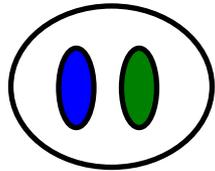
Francke et al. (2013) Dealing with the unexpected: consumer responses to direct-access *BRCA* mutation testing. *PeerJ* 1:e8  
<http://dx.doi.org/10.7717/peerj.8>

# Other discoveries: ancestry

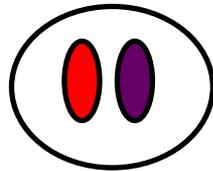


51.4%	Sub-Saharan African
43.3%	European
18.5%	Ashkenazi
	Northern European
1.9%	British and Irish
0.5%	Scandinavian
11.8%	Nonspecific Northern Europ...
	Southern European
< 0.1%	Nonspecific Southern Europ...
10.6%	Nonspecific European
1.8%	East Asian & Native American
1.3%	Native American
0.2%	East Asian
0.2%	Nonspecific East Asian & Nativ...
0.2%	South Asian
3.4%	Unassigned
100%	

# Other discoveries: family relationships

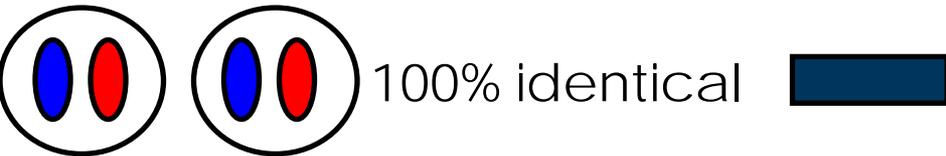


mother

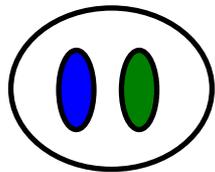


father

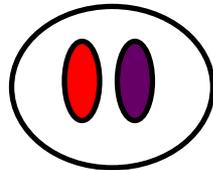
siblings



# Other discoveries: family relationships

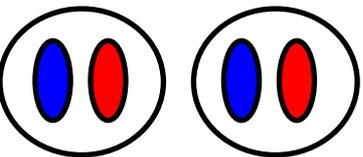


mother

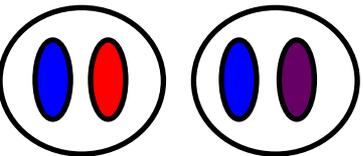


father

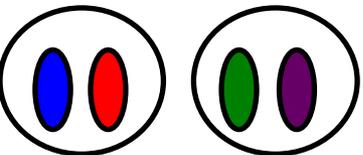
siblings



100% identical



50% identical



0% identical



Compare the genome of:

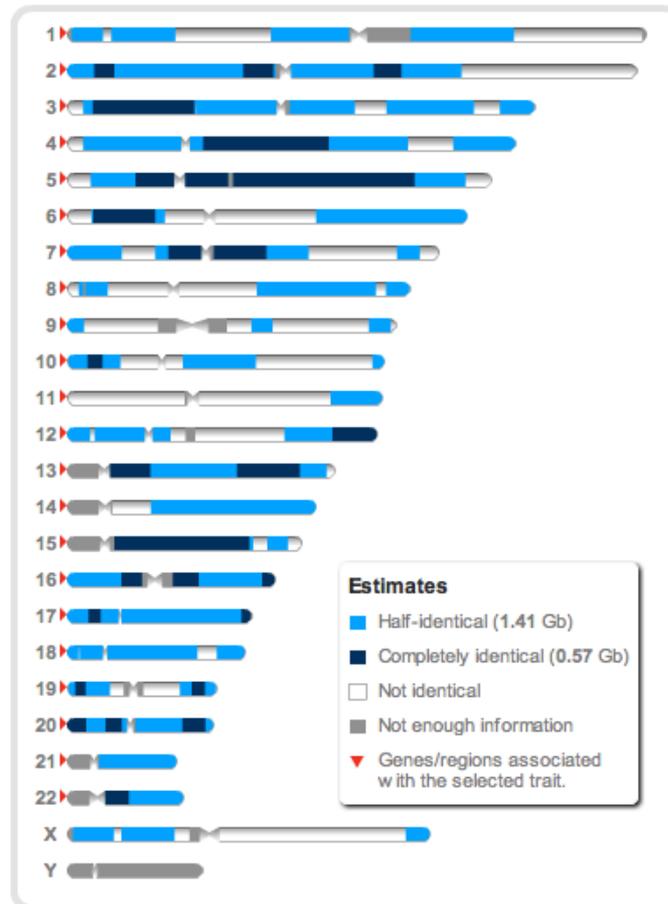
Erin Mendel (Daughter)

To the genome of:

Alan Mendel (Son)

## Genome-Wide Comparison

Comparison across all of the genome data



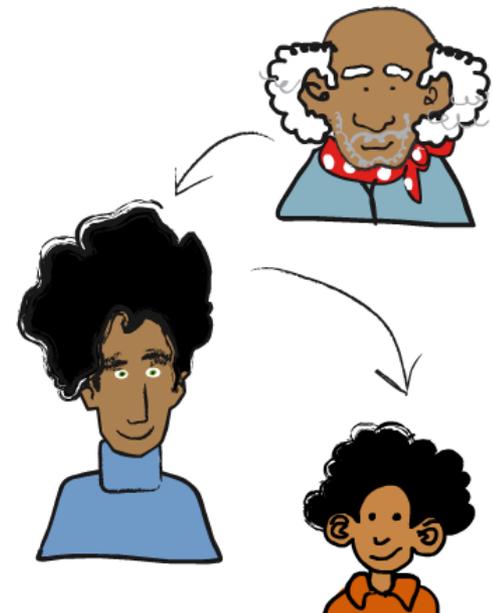
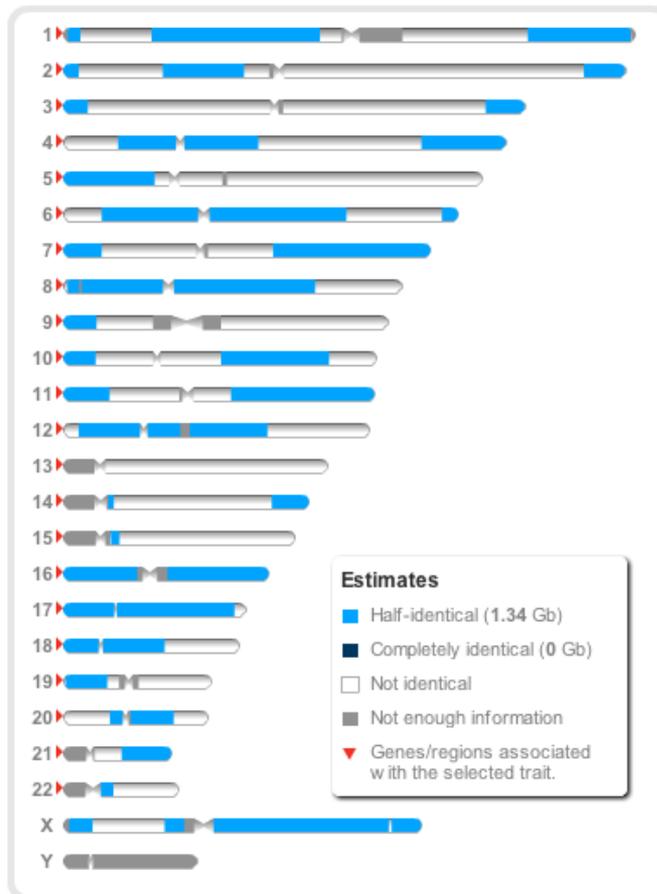
# Other discoveries: family relationships

Compare the genome of:

To the genome of:

## Genome-Wide Comparison

Comparison across all of the genome data



# Other discoveries: family relationships

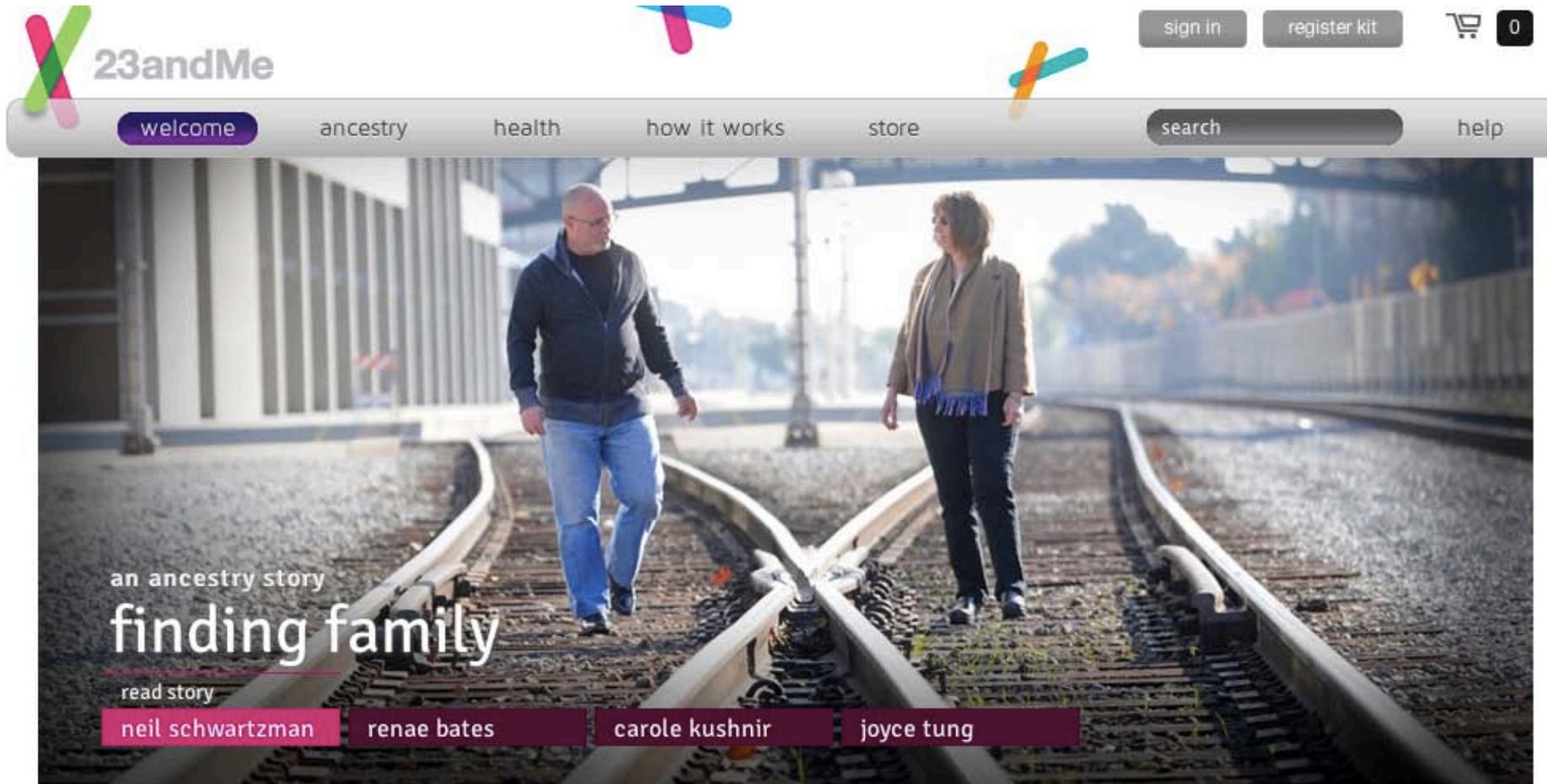
Paraphrasing:

"I'm the family genealogist - I spend all my time looking up old records, and I'm at this reunion of my Dad's family, recording information, thinking, I'm not even related to any of these people! And my mom still isn't talking to me ..."

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# Other discoveries: family relationships



The image shows a screenshot of the 23andMe website. At the top left is the 23andMe logo, which consists of a stylized 'X' made of three overlapping lines in green, pink, and blue. To the right of the logo is the text '23andMe'. Further right are two buttons: 'sign in' and 'register kit'. On the far right is a shopping cart icon with the number '0' next to it.

Below the logo and buttons is a navigation bar with several links: 'welcome', 'ancestry', 'health', 'how it works', and 'store'. To the right of these links is a search bar with the word 'search' inside, and a 'help' link on the far right.

The main content area features a large photograph of a man and a woman walking on a set of railroad tracks. The man is on the left, wearing a dark jacket and blue jeans. The woman is on the right, wearing a light-colored jacket and dark pants. They are walking away from the camera towards the horizon. The tracks curve to the right in the distance.

Overlaid on the bottom left of the photograph is the text 'an ancestry story' in a small font, followed by 'finding family' in a large, bold font. Below this is a 'read story' link. At the bottom of the photograph are four colored boxes containing the names of the authors: 'neil schwartzman' (pink), 'reanae bates' (purple), 'carole kushnir' (purple), and 'joyce tung' (purple).

23andMe Stories: <https://www.23andme.com/stories/2/>

# 23andMe's Terms of Service



## 5. Risks and Considerations Regarding 23andMe Services

...

**Genetic research is not comprehensive.** While we measure many hundreds of thousands of data points from your DNA, only a small percentage of them are known to be related to human traits or health conditions. The research community is rapidly learning more about genetics, and an important mission of 23andMe is to conduct and contribute to this research. In addition, many ethnic groups are not included in genetic studies. Because interpretations provided in our service rely on these published studies, some interpretations may not apply to you. Future scientific research may change the interpretation of your DNA. In the future, the scientific community may show previous research to be incomplete or inaccurate.

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# Health and traits reports over time

The 23andMe Team donotreply@23andme.com [via](#) mail24.us1.rsgsv.net 8/30/12 ☆  
to me ▾

Health and Traits Updates from 23andMe [Is this email not displaying correctly? View it in your browser.](#)



Dear Joanna,

New discoveries have recently been made about your DNA\*!

[View results >>](#)

Best regards,  
The 23andMe Team

\*Results reported for ethnicities in underlying studies.

# 23andMe's Personal Genome Service

## health overview

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

Show results for Joanna Mountain

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

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

### Disease Risks (90) ?

Elevated Risks	Your Risk	Average Risk
<a href="#">Age-related Macular Degeneration</a>	11.3%	7.0%
<a href="#">Lung Cancer</a>	8.6%	6.2%
<a href="#">Restless Legs Syndrome</a>	5.2%	4.2%
<a href="#">Exfoliation Glaucoma</a>	2.9%	1.0%
<a href="#">Type 1 Diabetes</a>	1.5%	1.0%

[See all 90 risk reports...](#)

### Carrier Status (24) ?

<a href="#">Alpha-1 Antitrypsin Deficiency</a>	Variant Present
<a href="#">Bloom's Syndrome</a>	Variant Absent
<a href="#">Canavan Disease</a>	Variant Absent
<a href="#">Cystic Fibrosis</a>	Variant Absent
<a href="#">Familial Dysautonomia</a>	Variant Absent
<a href="#">Factor XI Deficiency</a>	Variant Absent
<a href="#">Fanconi Anemia (FANCC-related)</a>	Variant Absent
<a href="#">Familial Hypercholesterolemia Type B</a>	Variant Absent

[See all 24 carrier status...](#)

### Traits (42) ?

<a href="#">Alcohol Flush Reaction</a>	Does Not Flush
<a href="#">Bitter Taste Perception</a>	Unlikely to Taste
<a href="#">Earwax Type</a>	Wet
<a href="#">Eye Color</a>	Likely Brown
<a href="#">Hair Curl</a>	Slightly Curlier Hair on Average

[See all 42 traits...](#)

### Drug Response (18) ?

<a href="#">Abacavir Hypersensitivity</a>	Typical
<a href="#">Alcohol Consumption, Smoking and Risk of Esophageal Cancer</a>	Typical
<a href="#">Clopidogrel (Plavix®) Efficacy</a>	Typical
<a href="#">Oral Contraceptives, Hormone Replacement Therapy and Risk of Venous Thromboembolism</a>	Typical
<a href="#">Fluorouracil Toxicity</a>	Typical

[See all 18 drug response...](#)

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.

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[See all 21 drug response...](#)

# What DNA data does 23andMe provide?

- Genotypes for about 1,000,000 single nucleotide polymorphisms, or SNPs, and a few insertion/deletions
- Illumina 730k OmniExpressPlus with an additional ~300,000 SNPs custom-selected by 23andMe
- SNPs distributed across entire genome: chromosomes 1-22, X & Y, as well as mitochondrial DNA

# What DNA data does 23andMe provide?

Settings 1 | HOME MY RESULTS FAMILY & FRIENDS RESEARCH & COMMUNITY Joanna Mountain | HELP

 **Browse Raw Data** [download raw data](#)

**NOTE:** This is an advanced view of all the uninterpreted SNP data from your chip. To see how these raw data relate to 248 traits and conditions, go to [Health and Traits](#).

The data from 23andMe's "Browse Raw Data" feature is for research and informational use only. While this data has undergone a chip-wide review process, it has not gone through the same rigorous review process as the data from the 23andMe Health Reports.

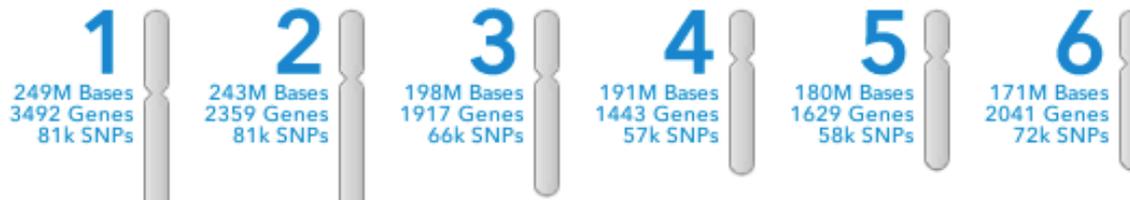
Most of the DNA inside each of your body's cells is divided into pieces called chromosomes, with the remaining DNA found in tiny loops inside your cells' mitochondria. Click below on any chromosome or the mitochondrial loop to see the genes and SNPs it contains. [Learn more about how to use this feature.](#)

Jump to a gene:

Go

a SNP:

Go



# What DNA data does 23andMe provide?

23andMe logo | HOME | MY RESULTS | FAMILY & FRIENDS | RESEARCH & COMMUNITY | Joanna Mountain | HELP

[download raw data](#)

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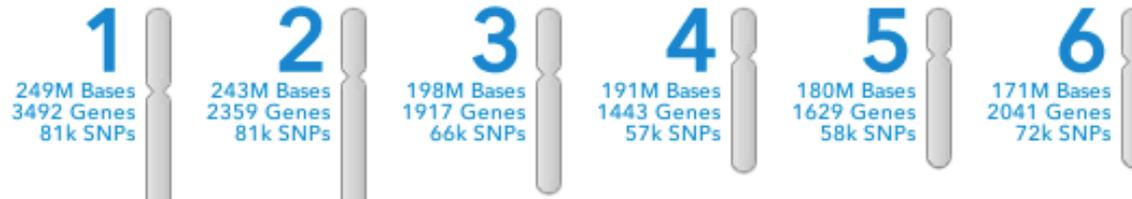
Most of the DNA inside each of your body's cells is divided into pieces called chromosomes, with the remaining DNA found in tiny loops inside your cells' mitochondria. Click below on any chromosome or the mitochondrial loop to see the genes and SNPs it contains. [Learn more about how to use this feature.](#)

Jump to a gene:

Go

a SNP:

Go



# External tools for interpreting 23andMe raw data

## [Promethease](#) Report

**Version: 0.1.153**

**Generated: 2013-04-28 22:32**

**Infile:** [http---genomealberta.ca-files-Personal\\_Genotype\\_Files-deCODEme\\_scan\[1\].csv](http---genomealberta.ca-files-Personal_Genotype_Files-deCODEme_scan[1].csv), [http---genomealberta.ca-files-Personal\\_Genotype\\_Files-23andMe\\_V3\\_genome\\_mikesgene\\_Full.zip](http---genomealberta.ca-files-Personal_Genotype_Files-23andMe_V3_genome_mikesgene_Full.zip)

**Reference Population CEU**

**19096 genotypes annotated**

genotype [conflicts](#) detected

[UI version 2](#) interactive report

**Good news**

[...more...](#)

**Bad news**

[...more...](#)

**These seem interesting, but have not been flagged as clearly Good or Bad**

[...more...](#)

[Medicines](#)

[Medical Conditions](#)

[Topics](#)

[Help](#) interpreting your results.

[Show Everything](#)

# External tools for interpreting 23andMe raw data

## Bad news

[\(hide\)](#)

[Show Everything](#)

[rs4363657\(C:C\)](#)

17x increased myopathy risk for statin users...[more...](#)

Magnitude: 4

Frequency: 0.9%

Count: 2

Repute:Bad

References:8

[rs738409\(G:G\)](#)

higher odds of alcoholic liver disease, increased liver fat While found in 55%+ of all people, alcohol seems to be 3x more damaging to your liver than typical. Higher risk for developing fatty liver, fibrosis, and fibrosis progression, with a per allele odds ratio of 2.55, 3.13 and 2.64, respectively. news

Magnitude: 4

Frequency: 55.4%

Repute:Bad

References:62

[...more...](#)

[ambig](#)

[gs141](#)

You carry one APOE-ε3 allele and one APOE-ε4 allele. This results in 2x increased relative risk of Alzheimer's disease. For non-caucasians the risk is increased, but SNPedia has not yet seen any reliable estimates. This is based on \*rs429358(C;T) \*rs7412(C;C)

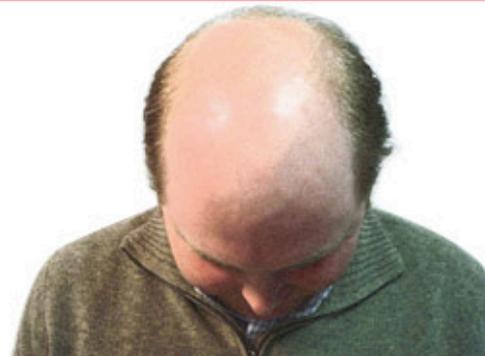
Magnitude: 3.5

Repute: Bad

[gs122](#)

Magnitude: 3.1

Repute: Bad



7x risk of baldness according to an article in Nature. That site may require paid access; the abstract at is accessible.

# 23andMe's Terms of Service



In addition, if you choose to provide your Genetic and/or Self-Reported Information to third parties - whether individuals to whom you facilitate access, intentionally or inadvertently, or to third parties for diagnostic or other purposes - you agree to defend and hold harmless 23andMe, its employees, contractors, successors, and assigns from any and all liability arising from such disclosure or use of your Genetic and/or Self-Reported Information.

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# Our most recent publication



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521

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12

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RESEARCH ARTICLE

## Genome-Wide Analysis Points to Roles for Extracellular Matrix Remodeling, the Visual Cycle, and Neuronal Development in Myopia

Amy K. Kiefer, Joyce Y. Tung, Chuong B. Do, David A. Hinds, Joanna L. Mountain, Uta Francke, Nicholas Eriksson 

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Acknowledgments

### Abstract

Myopia, or nearsightedness, is the most common eye disorder, resulting primarily from excess elongation of the eye. The etiology of myopia, although known to be complex, is poorly understood. Here we report the largest ever genome-wide association study (45,771 participants) on myopia in Europeans. We performed a survival analysis on age of myopia onset and identified 22 significant associations ( $p < 5 \cdot 10^{-8}$ ), two of which are replications of earlier associations with refractive error. Ten of the 20 novel associations identified replicate in

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# List of 23andMe publications



- 23andMe Research
- 23andMe Research Findings
- Physician Community
- Scientific Community

## for the scientific community

Just a few years ago the technology to perform genome-wide association studies did not even exist. Now we are at the forefront of a revolution in personalized genetics and medicine, thanks to a community of geneticists and other scientists, technologists, physicians and innovators. We realize that only through a community effort will this field continue to advance.

We want our service, our methods, and our goals to be transparent to our colleagues in the scientific community. Explore our scientific publications and white papers below to find out more. We welcome your comments and suggestions.

### Publications

Learn what 23andMe researchers have discovered with the help of 23andMe customers.

Kiefer AK, Tung JY, Do CB, Hinds DA, Mountain JL, Francke U, and N. Eriksson. [Genome-wide analysis points to roles for extracellular matrix remodeling, the visual cycle, and neuronal development in myopia.](#) PLoS Genet. 2013. 9(2): e1003299.

Francke U, Dijamco C, Kiefer AK, Eriksson N, Moiseff BR, Tung JY, Mountain JL. [Dealing with the unexpected: Consumer responses to direct-access BRCA mutation testing.](#) PeerJ. 2013 1:e8.

Eriksson N, Wu S, Do CB, Kiefer AK, Tung JY, Mountain JL, Hinds DA, Francke U. ["A genetic variant near olfactory receptor genes influence cilantro preference."](#) Flavour. 2012 Dec;1(22).

Heilmann S, Kiefer AK, Fricker N, Drichel D, Hillmer AM, Herold C, Tung JY, Eriksson N, Redler S, Betz RC, Li R, Karason A, Nyholt DR, Song K, Vermeulen SH, Kanoni S, Dedoussis G, Martin NG, Kiemeny LA, Mooser V, Stefansson K, Richards JB, Becker T, Brockschmidt FF, Hinds DA, Nothen MM. ["Androgenetic alopecia: identification of four new genetic risk loci and evidence for the contribution of WNT-signaling to its etiology."](#) J Invest Dermatol. 2013 Jan 28.

Do CB, Hinds DA, Francke U, Eriksson N. ["Comparison of Family History and SNPs for Predicting Risk of Complex Disease."](#) PLoS Genet. 2012 Oct;8(10):e1002973.

# Updating customers regarding research

The screenshot shows the 23andMe website interface. At the top, there is a navigation bar with the 23andMe logo, a search bar, and links for 'welcome', 'ancestry', 'health', 'how it works', 'store', and 'help'. On the right side of the navigation bar, there are buttons for 'sign in', 'register kit', and a shopping cart icon with a '0' next to it. Below the navigation bar, there is a sidebar on the left with a list of links: '23andMe Research', '23andMe Research Findings', 'Physician Community', and 'Scientific Community'. The main content area is titled '23andMe Research Findings' and features a list of research articles. Each article has a small image, a title, a date, and a brief description. The articles are: 'Cilantro as a matter of taste' (September 2012), 'Parkinson's, Cholesterol, Type 2 Diabetes and BMI' (April 2012), 'Parkinson's Disease, Back Pain and Joint Replacement' (April 2012), 'Parkinson's Disease and Personality' (April 2012), and 'Rapid replication of research in MPN' (March 2012). To the right of the articles, there is a blue box with a 'Thank you' message and a red button that says 'Add to Cart »'. At the bottom of the page, there is a copyright notice: 'Copyright © 2013 23andMe, Inc. All rights reserved.'

23andMe

welcome ancestry health **how it works** store search help

sign in register kit 0

23andMe Research  
▶ 23andMe Research Findings  
Physician Community  
Scientific Community

## 23andMe Research Findings ?

**Cilantro as a matter of taste** SEPTEMBER 2012

 For many people fresh cilantro is a beloved ingredient in their favorite dishes. Others find the herb practically inedible, likening the taste to soap, mold, and dirt. The story behind this love-hate reaction is partly genetic. [Show Details »](#)

**Parkinson's, Cholesterol, Type 2 Diabetes and BMI** APRIL 2012

 People with Parkinson's disease suffer from a number of motor symptoms but it appears that they may have better than average metabolic health. [Show Details »](#)

**Parkinson's Disease, Back Pain and Joint Replacement** APRIL 2012

 Researchers at 23andMe have found that along with impacting a person's motor skills Parkinson's disease is also associated with back pain and the need for joint replacement surgery for some patients. [Show Details »](#)

**Parkinson's Disease and Personality** APRIL 2012

 Researchers at 23andMe have found that people with Parkinson's disease tend to have certain personality traits and preferences. [Show Details »](#)

**Rapid replication of research in MPN** MARCH 2012

 We replicate an association between inherited variation in the JAK2 gene and rare blood cancers linked to another JAK2 mutation that can appear later in life. [Show Details »](#)

*Our research is driven by our community. A big thank you to our customers who make our research possible.*

Thank you

Help us fuel more research

**Add to Cart »**

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# 23andMe's Personal Genome Service



June 16, 2011

## 23andMe Research Findings: From You, Back to You

Published by 23andMe under 23andMe Research, announcements

It's been nearly a year since we published our first scientific paper. Titled "[Web-based, Participant-Driven Studies Yield Novel Genetic Associations for Common Traits](#)," the paper demonstrated that 23andMe, our revolutionary new research model, works.



Since then we've been making exciting progress, applying the research framework to dozens of common medical conditions, studies of drug response, infectious diseases, and more physical traits. Our database is also growing larger every day, making this one of the largest collaborative research projects in the world, and certainly the largest one where every research participant has access to his or her own genetic data.

NEW AND RECENTLY UPDATED REPORTS

	RESULT
	Variant Present
	Variant Present
with Peripheral	Variant Absent
kidney Disease	Variant Absent
	Variant Absent

[See all 50 carrier status...](#)

Alcohol Flush Reaction	Does Not Flush	Abacavir Hypersensitivity	Typical
Bitter Taste Perception	Unlikely to Taste	Alcohol Consumption, Smoking and Risk of Esophageal Cancer	Typical
Earwax Type	Wet	Clopidogrel (Plavix®) Efficacy	Typical
Eye Color	Likely Brown	Oral Contraceptives, Hormone Replacement Therapy and Risk of Venous Thromboembolism ♀	Typical
Hair Curl ✖	Slightly Curlier Hair on Average	Fluorouracil Toxicity	Typical

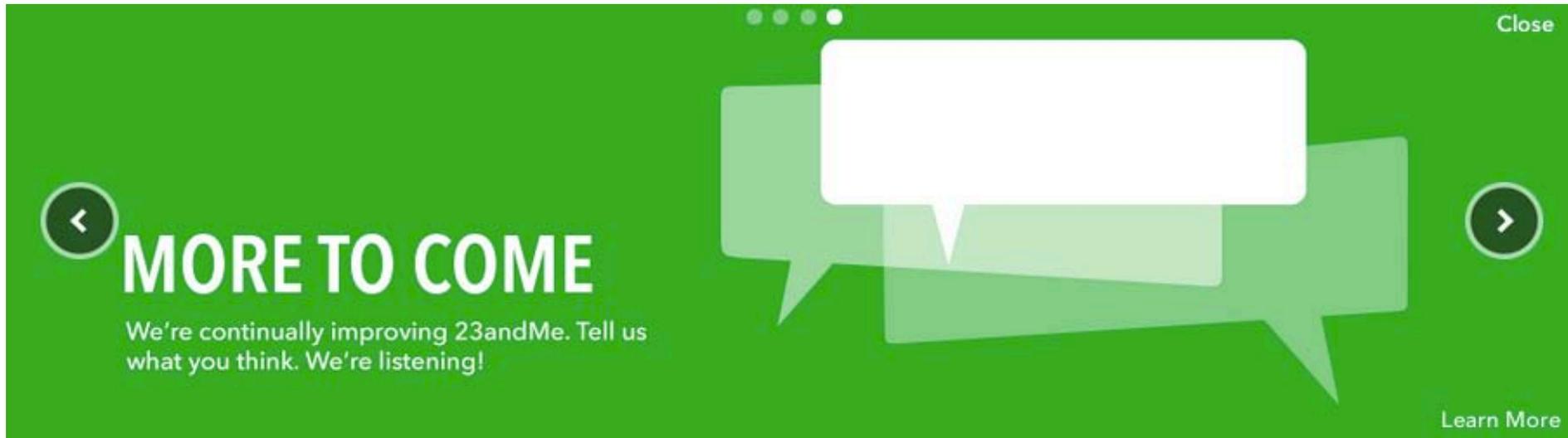
[See all 57 traits...](#)

[See all 21 drug response...](#)

# Some steps we have taken ...

- Stated risks around new personal discoveries very explicitly in Terms of Service
- Moved from “opt in” step for all health reports to “opt in” step for a subset of four health reports
- Provided extensive educational & background information
  - Provided in-depth background information on diseases plus associated genetic and non-genetic factors
  - Provided educational videos for APOE and LRRK2 reports
- Provided genetic counseling option through partner
- Provided customers with options for sharing different sets of results with other customers
  - Provided options of sharing with or without health reports
  - Excluded four “locked” reports from both sharing options
- Provided new reports based on research supported by customers

# Importance of customer feedback



Close

<

## MORE TO COME

We're continually improving 23andMe. Tell us what you think. We're listening!

>

Learn More

The banner features a central graphic of three overlapping speech bubbles in shades of green and white. At the top center, there are four small white dots. On the right side, there is a 'Close' button. On the left side, there is a circular button with a left-pointing arrow. On the right side, there is a circular button with a right-pointing arrow. At the bottom right, there is a 'Learn More' link.

# Acknowledgements

- Colleagues at 23andMe
  - Anne Wojcicki, Brian Naughton, Ashley Gould, Nick Eriksson, David Hinds, Chuong Do, Amy Kiefer, Uta Francke, Joyce Tung, Emily Drabant, Cheri Djamco, Meghan Mullins, Kim Barnholt, Samara Mohammed, Mike Macpherson, Eric Durand, Arnab Chowdry, Shirley Wu, and many more
- 23andMe's Advisors
- 23andMe's customers who have provided feedback regarding the service, as well as survey responses and genotypic data for research