



The Senior Games Genome Project

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23andMe

9 April 2010



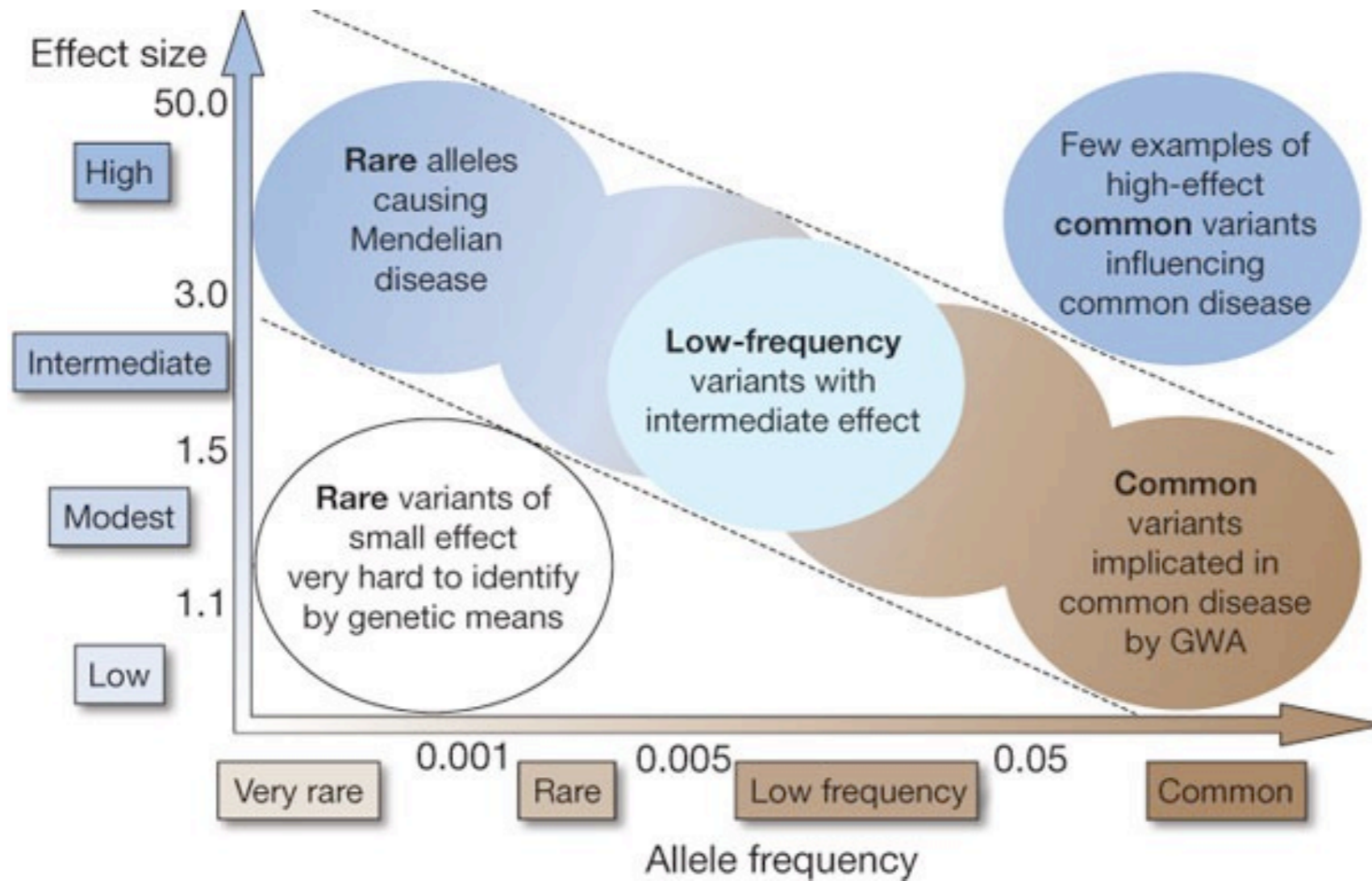
Genetics 101

Senior Games

Personalized Medicine

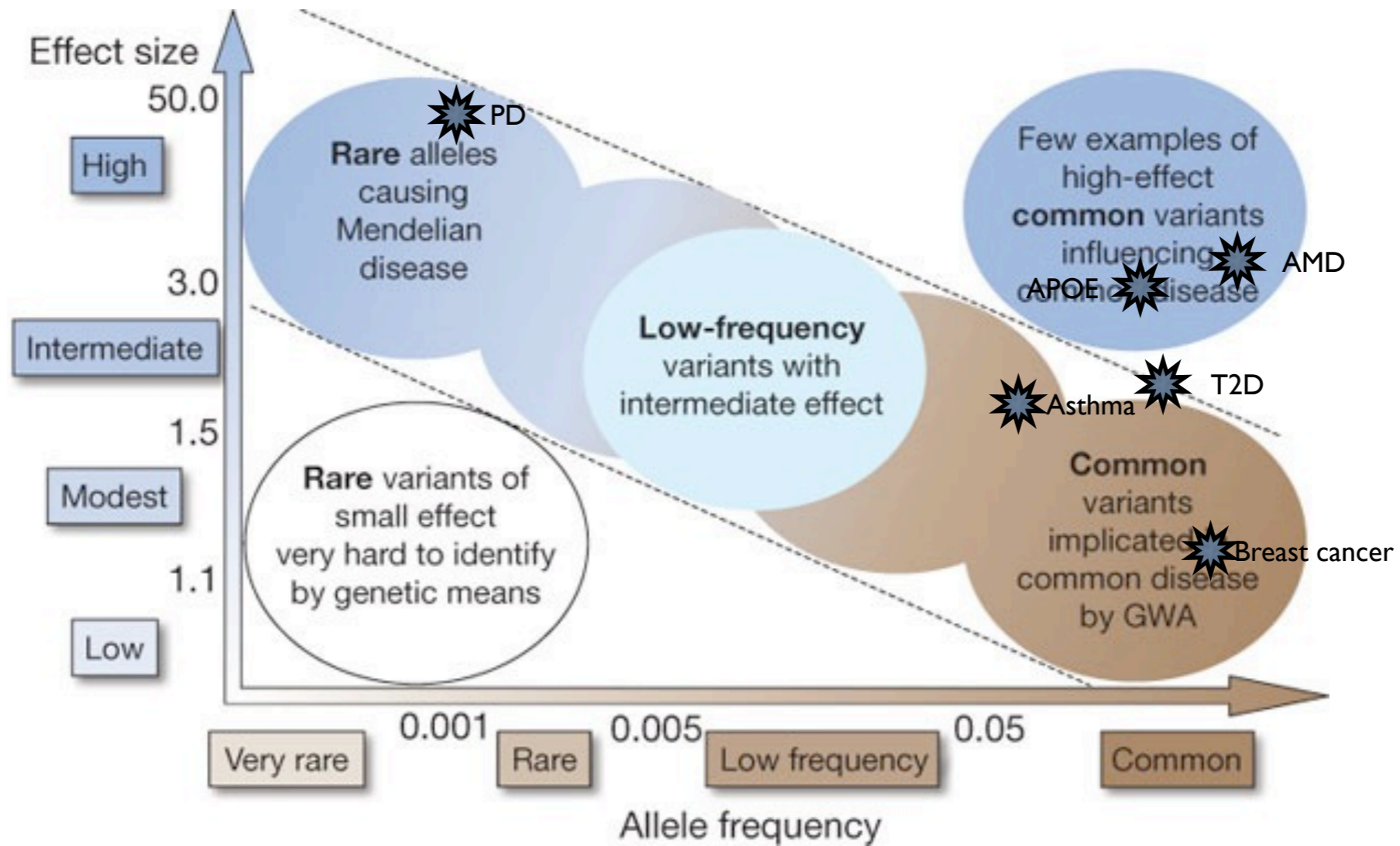
Research 2.0

Genetics and disease



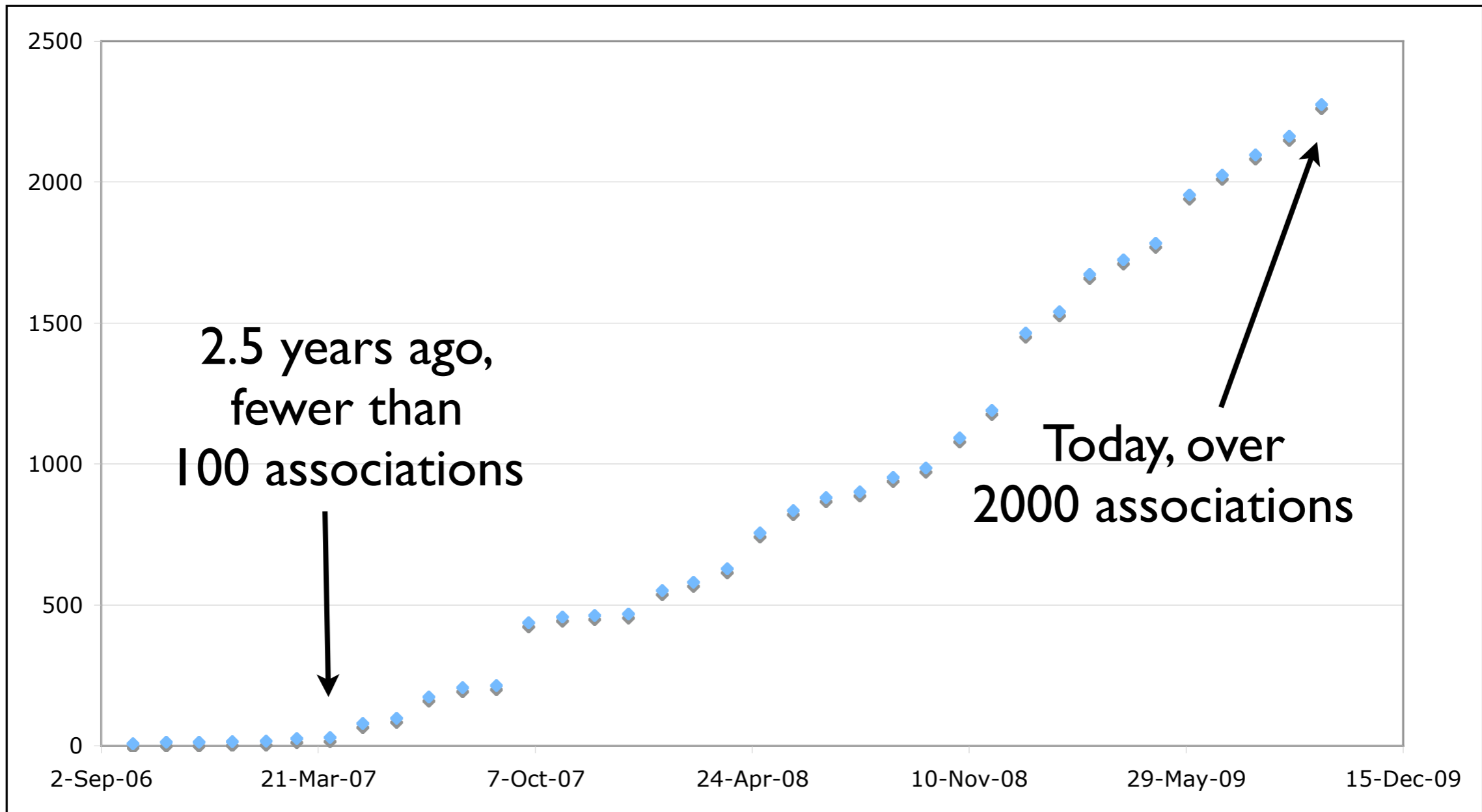
[TA Manolio *et al. Nature* **461**, 747-753 (2009)]

Genetics and disease



[TA Manolio et al. *Nature* **461**, 747-753 (2009)]

SNP-disease associations

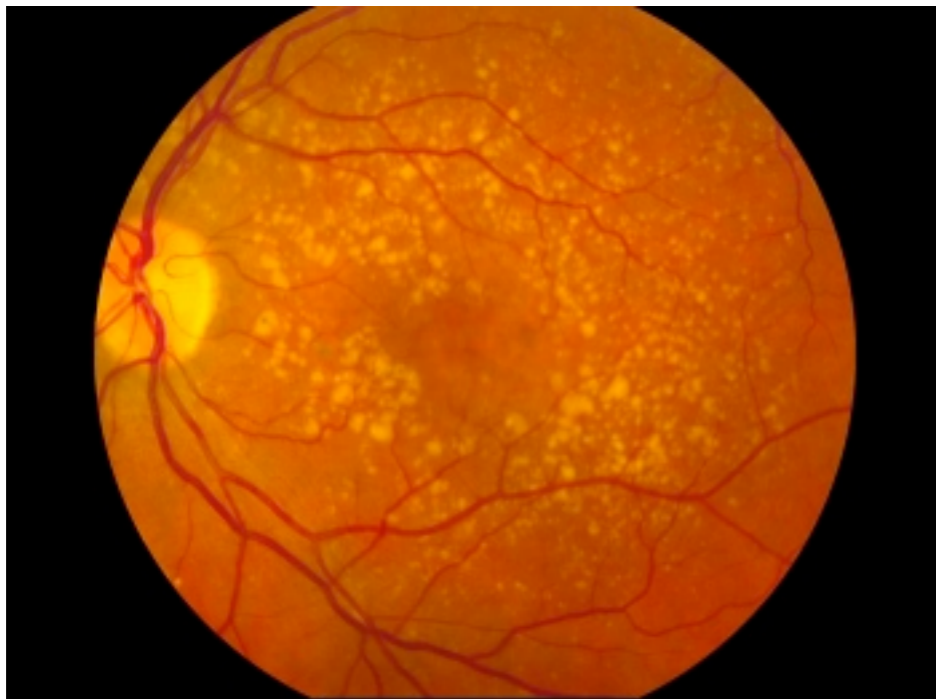


Growing weekly

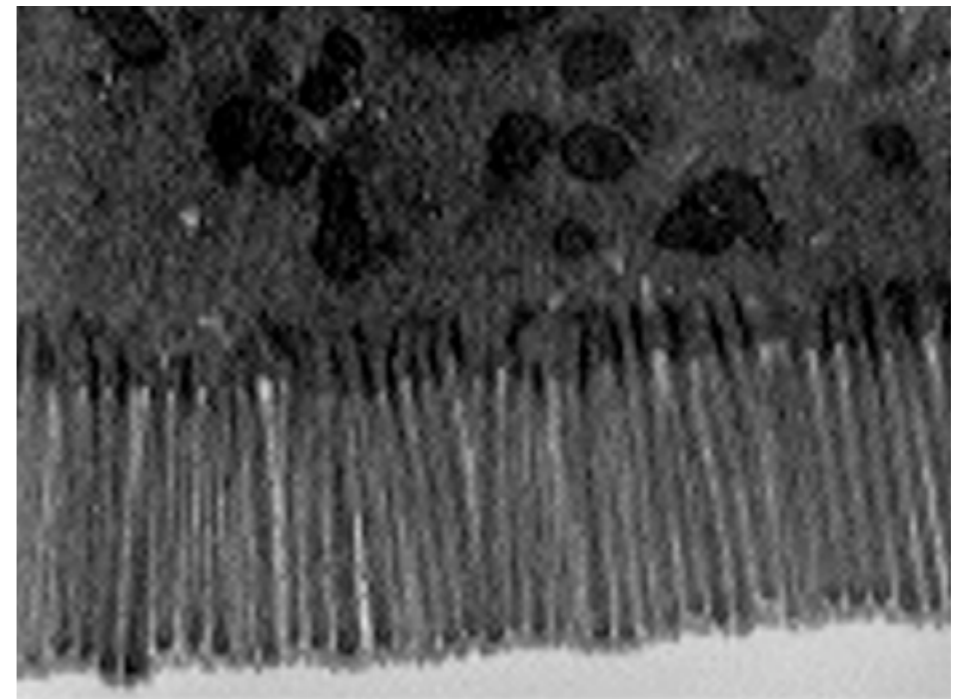
January	Glucose and Insulin response A set of metabolic traits Chronic Lymphocytic Leukemia Mood Disorders Ankylosing Spondylitis Electrocardiographic measures (3 studies) Fasting Glucose and Type 2 Diabetes
February	Atrial Fibrillation Dementia Pancreatic Cancer A set of hematological and biochemical traits COPD Telomere Length
March	Celiac disease Pediatric eosinophilic esophagitis Ulcerative colitis

[Nature Genetics, 2010]

Illuminating treatment



AMD - complement system



Crohn's - autophagy



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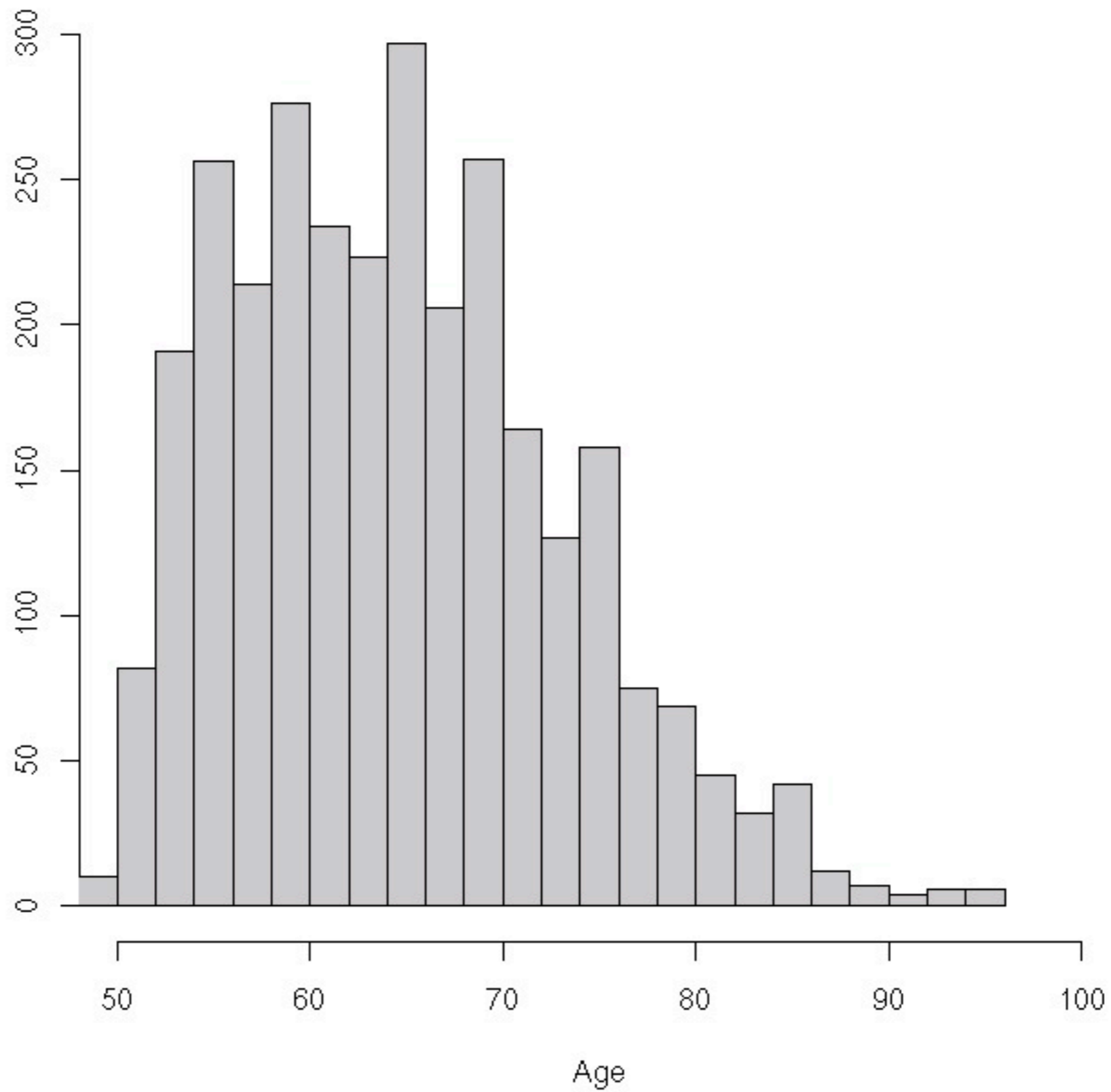
Research 2.0



Senior Games Genome Project

- 4535 people joined
- Over 70% participating in research
- 36,435 surveys completed!
- Contributed to over 400 studies
 - from agreeableness to zyrtec (dry mouth)

Age distribution for Sr Games cohort



Self-reported health

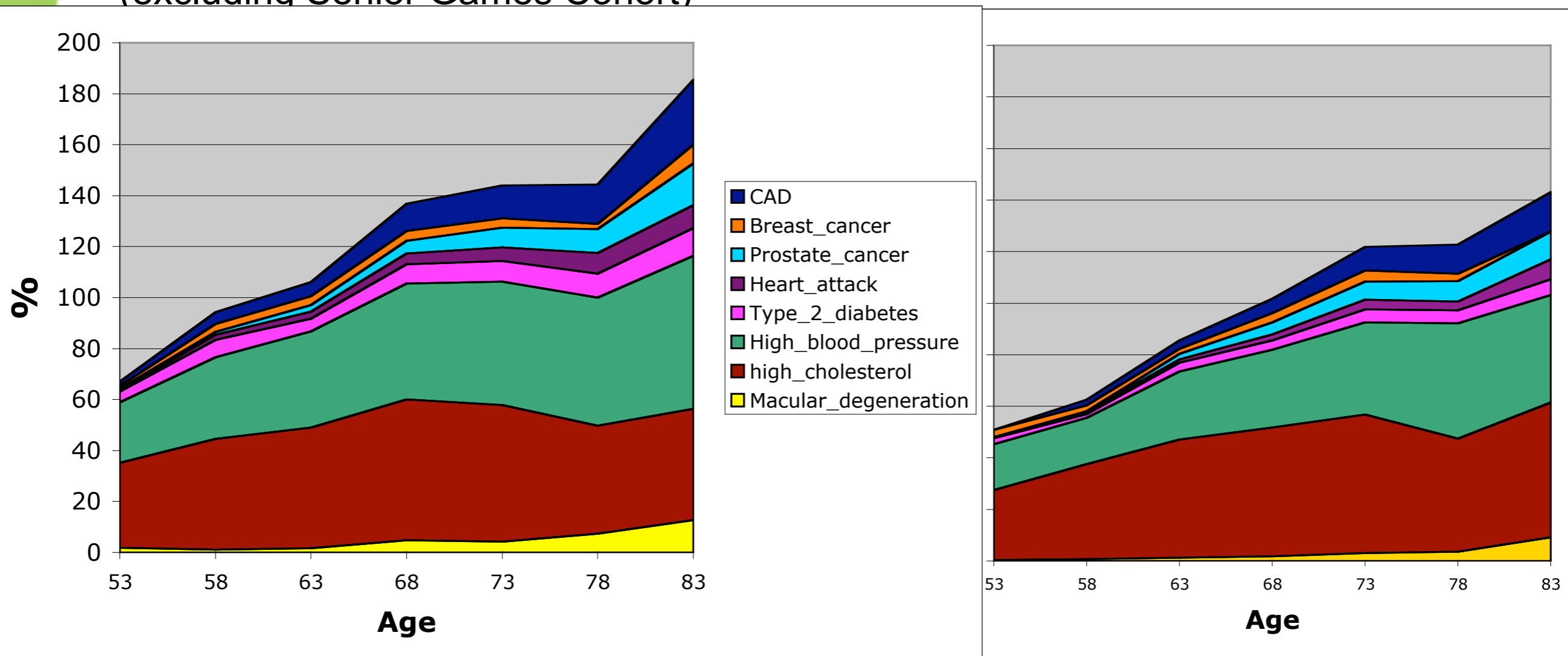
- Senior games participants had greater self-reported health measures than other over 50 year old customers.
- Factors impacting health in our study: lack of depression, higher levels of education, more physical activity, fewer diseases of aging, and less neuroticism.

A very healthy group

General Cohort

(excluding Senior Games Cohort)

Senior Games Cohort



*Total exceeds 100%; some individuals have multiple conditions



Genetics 101

Senior Games

Personalized Medicine

Research 2.0

What is Personalized Medicine?

“The application of **genomic** and molecular data to better target the delivery of health care, facilitate the discovery and clinical testing of new products, and help determine a person’s predisposition to a particular disease or condition.”

*Proposed Senate Bill S.976
Sponsored by Barack Obama in 2007*

“A form of medicine that uses information about a person’s **genes**, proteins, and environment to prevent, diagnose, and treat disease.”

National Cancer Institute website

“The use of **genetic** susceptibility or **pharmacogenetic** testing to tailor an individual's preventive care or drug therapy.”

Nature Genetics website

23andMe: Interwoven goals

Ancestry



Provide consumers with access to their genetic information with the tools needed to make sense of it.

Health and Traits



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Health and Traits



Research



Use public excitement about genetics to drive further research.

Sharing and Community

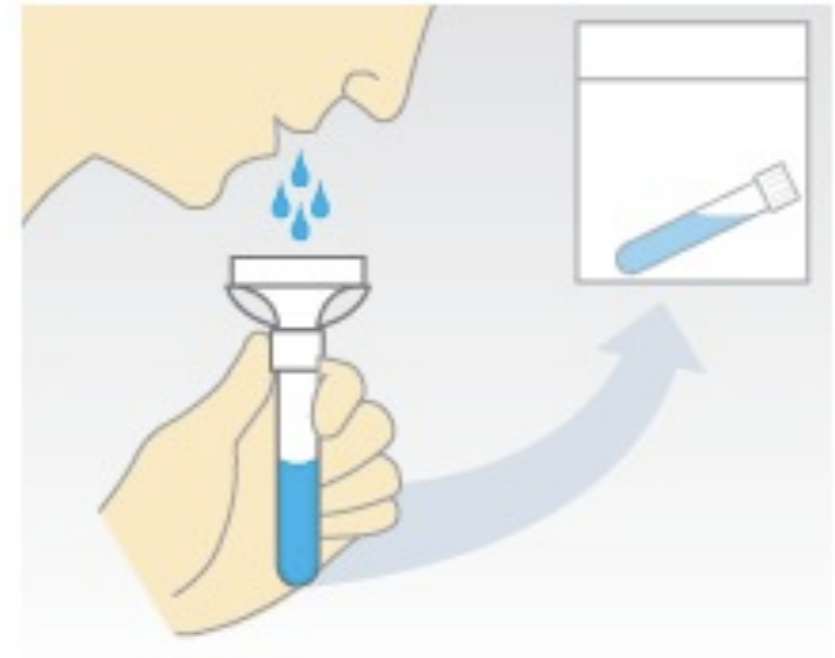


How it works



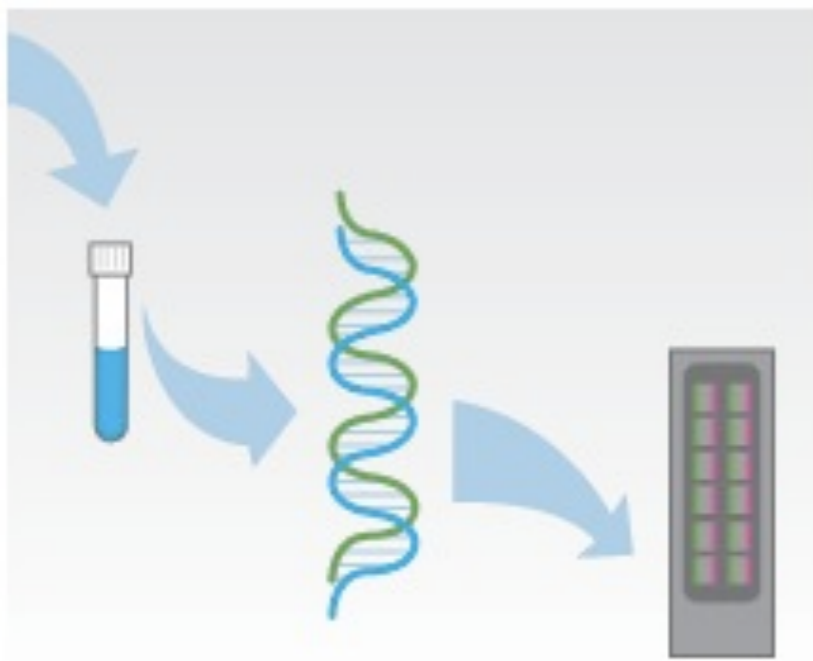
Order a kit via our web site.

2



Spit into tube & send tube to lab

3



Lab extracts DNA from your sample and analyzes it at about 600,000 locations.

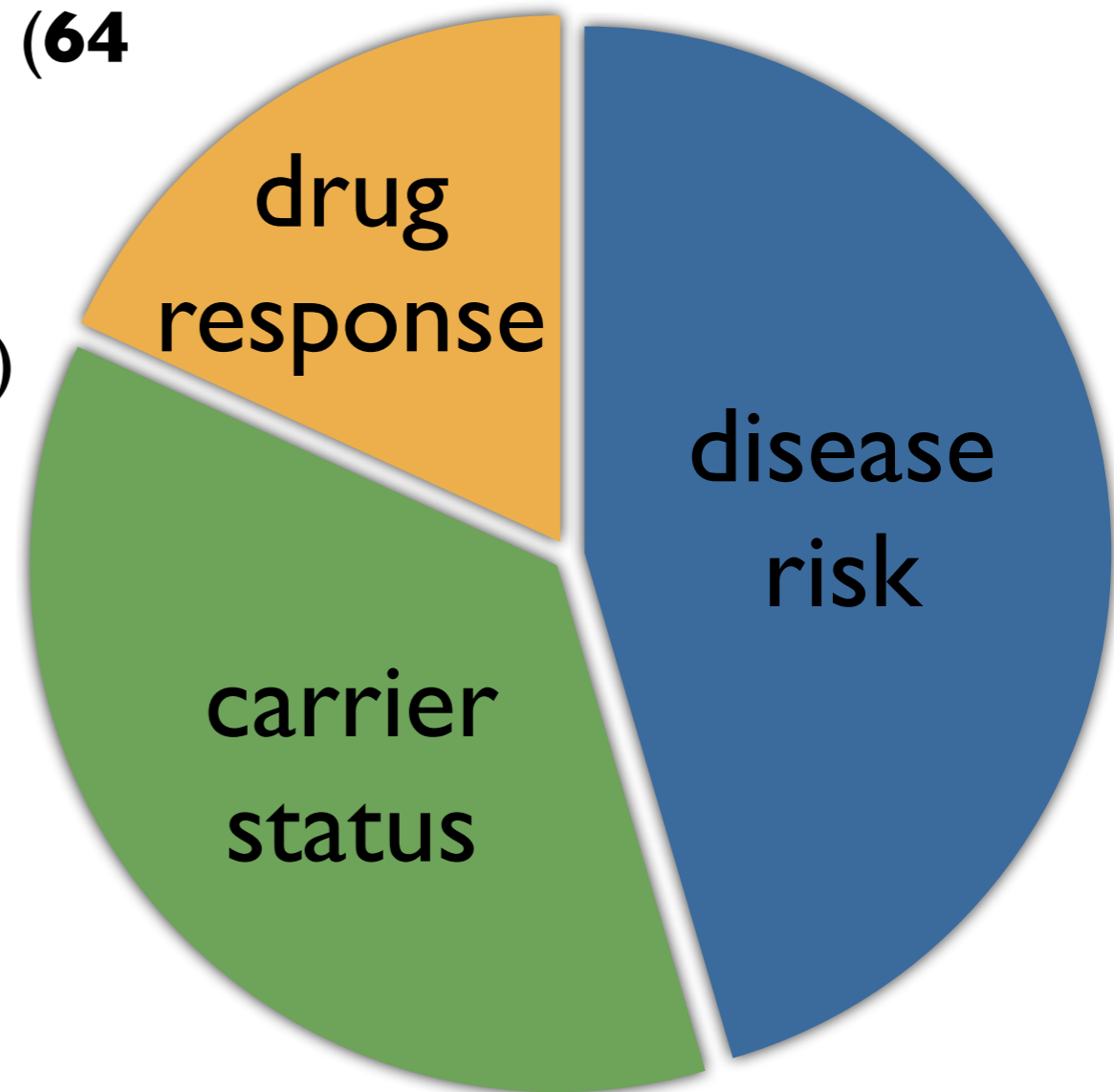
4



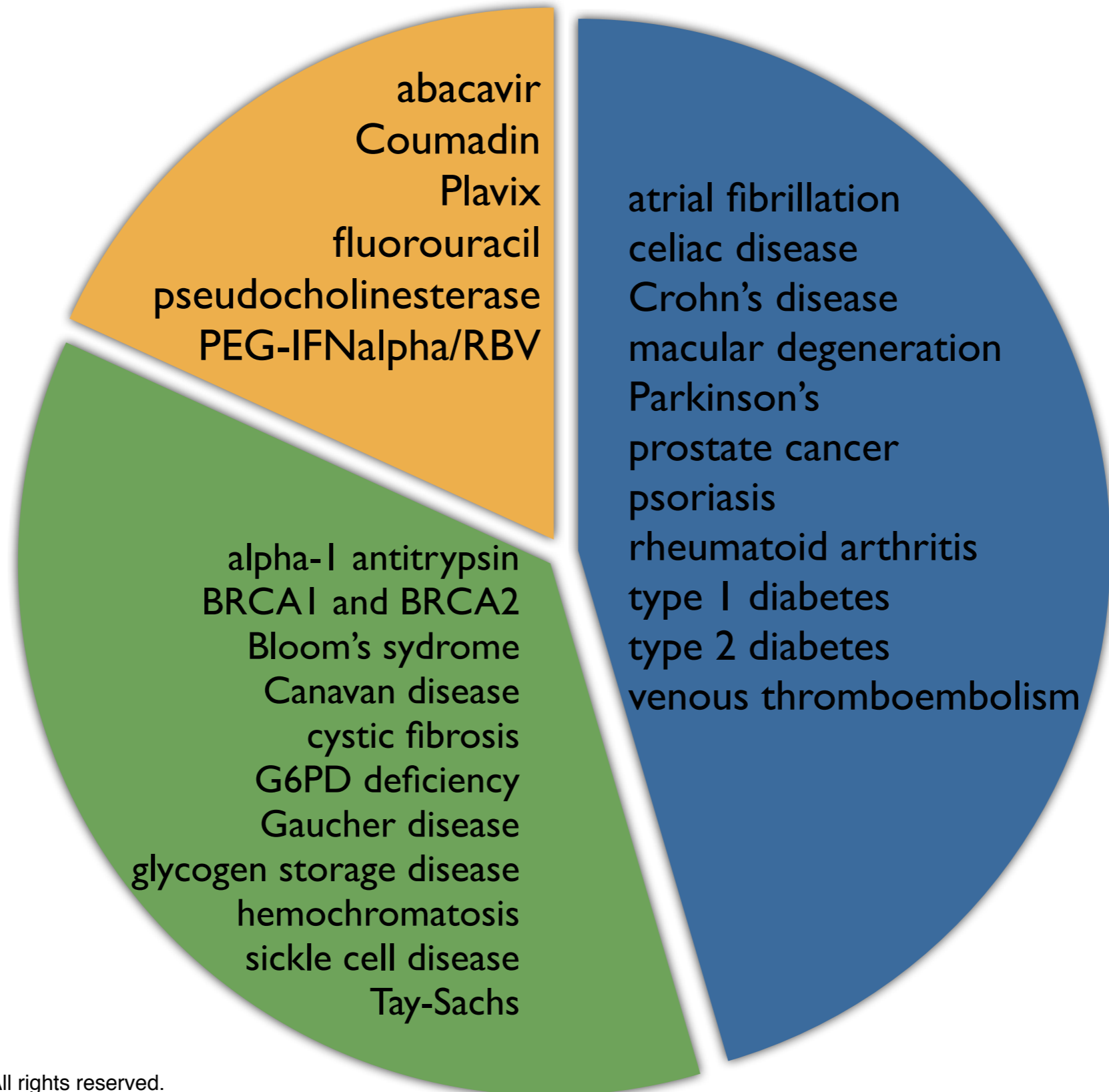
Log in and explore your genetic data.

Health information

- In total, 140+ reports on health and traits (**64 are of very high confidence**, and growing)
 - 24 **carrier status** (recessive disease) reports
 - 8 **drug response** reports
 - 22 **disease risk** reports
 - 10 traits



Health information



Who learns what?

Venous Thromboembolism	F5 or F2 mutation	8.2%
AMD	4-fold average risk (29%)	5.6%
Cystic Fibrosis	delta F508	2.5%
Prostate cancer	double average risk (36%)	1.0%
Psoriasis	double average risk (30%)	0.8%
Parkinson's disease	5-fold average risk	1%
Crohn's	5-fold average risk	5%
Plavix	Greatly reduced efficacy	3%

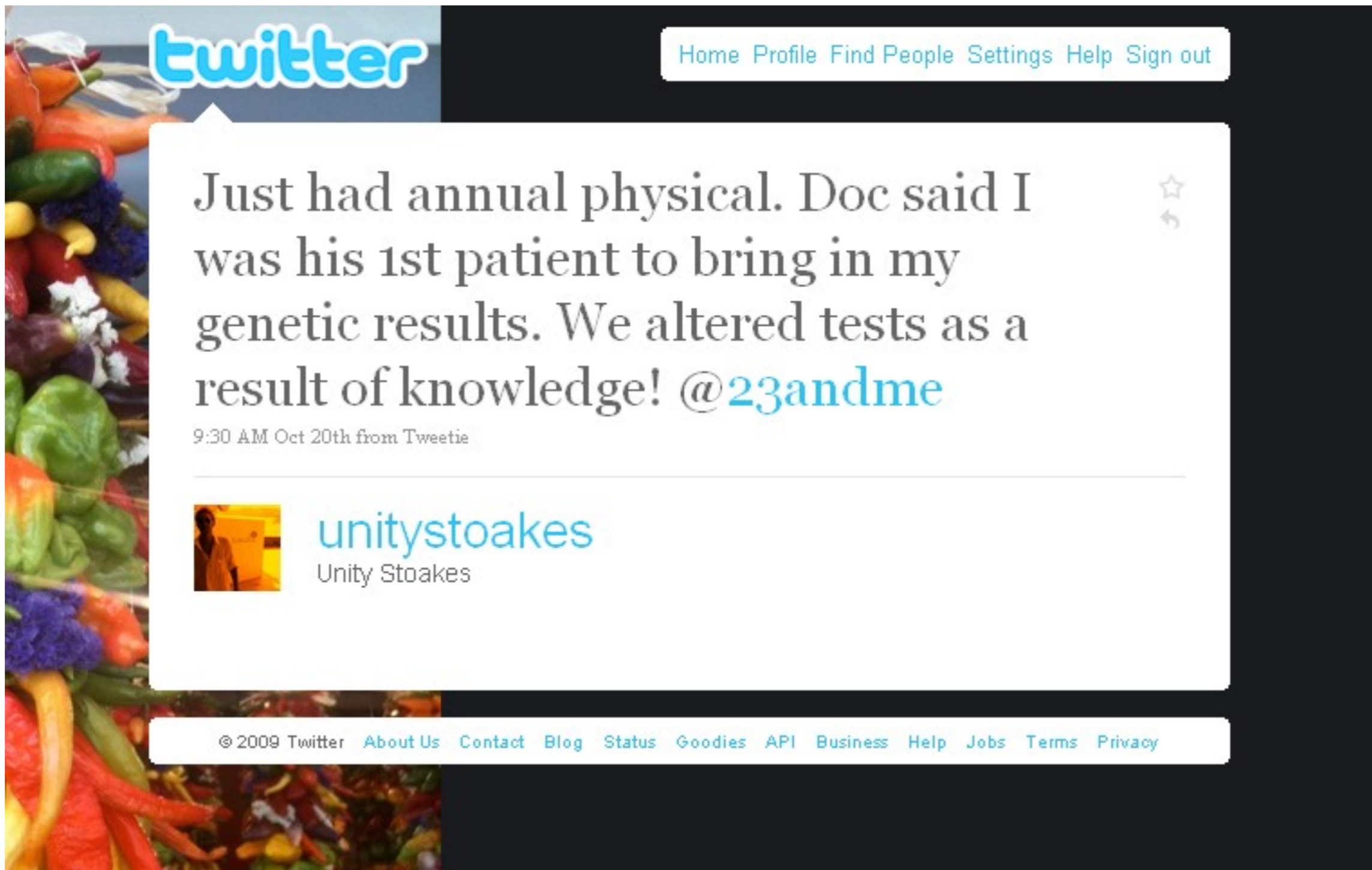
Real-life example

Jeff Gulcher, CSO of deCODE

- Got his genetic data from a deCODEme test
- PSA test for prostate cancer at the age of 48
- Exploratory biopsy - aggressive form of cancer
- Underwent surgery right away

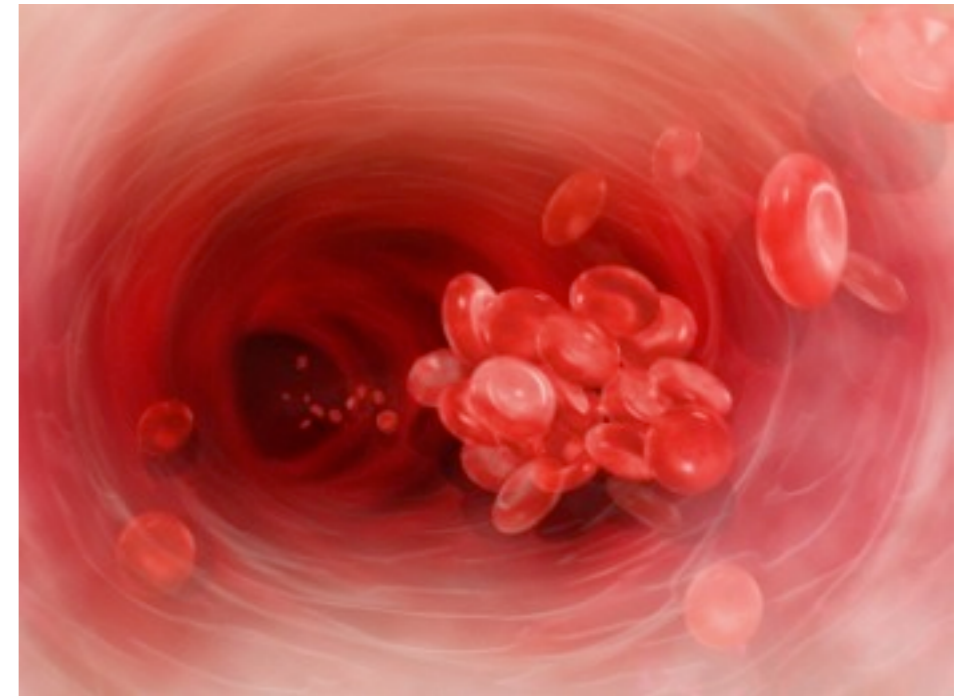
deCODEme blog, July 25, 2008

Real-life example



Pharmacogenetics

Warfarin: FDA recommends a genetic test.



5-FU: toxic in about 30% of people, many because of a genetic deficiency

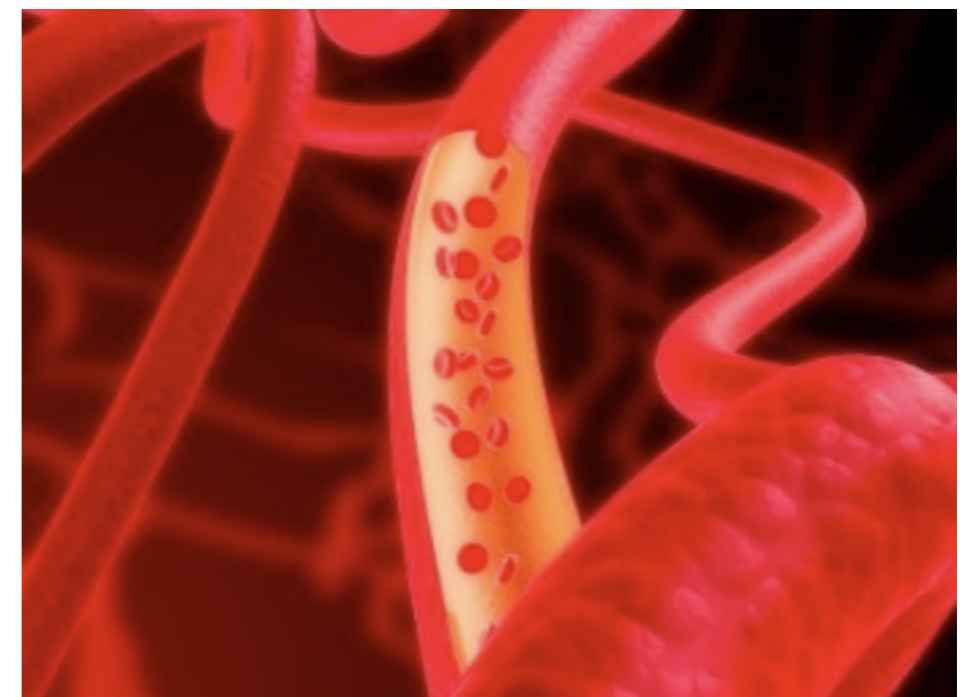


Pharmacogenetics

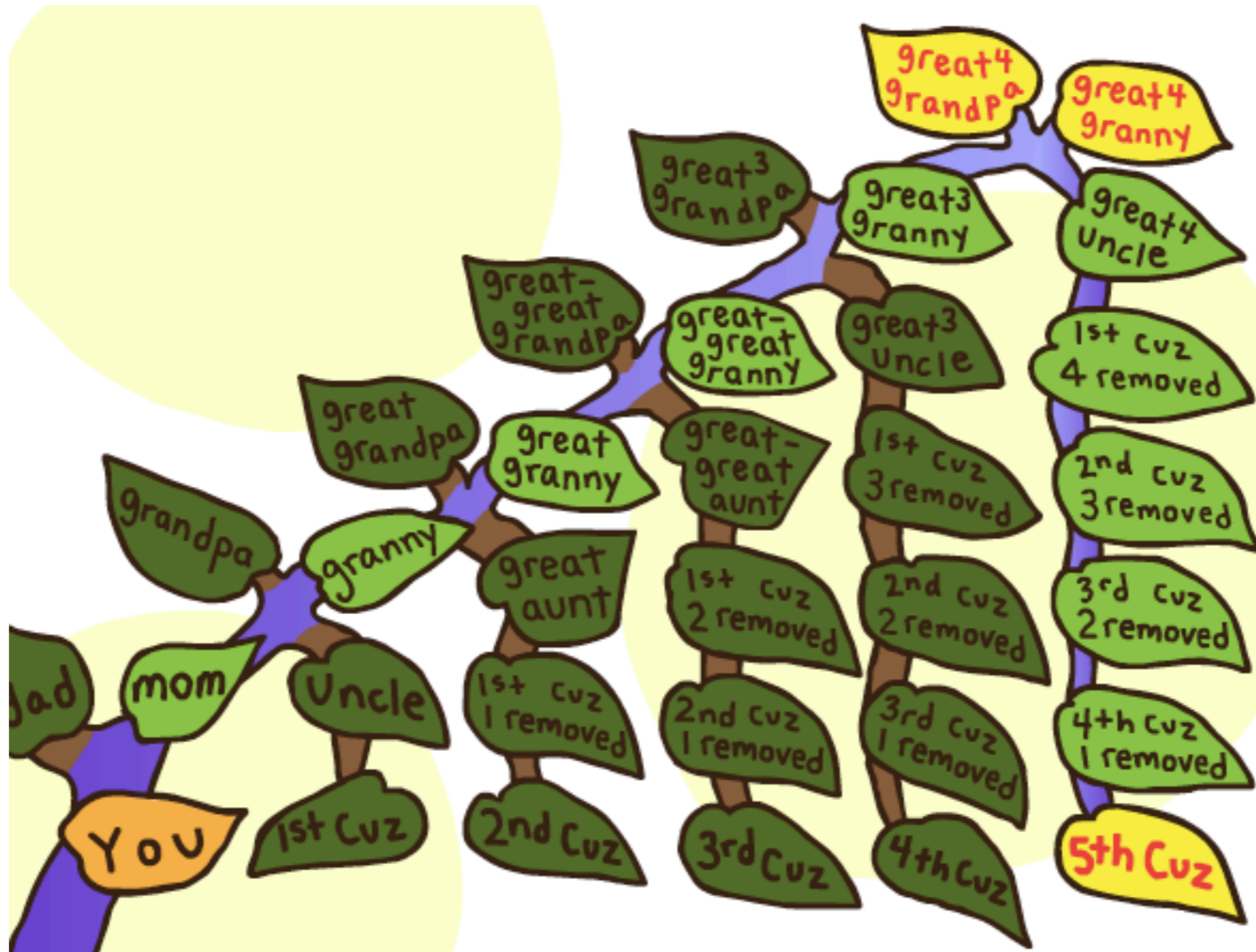
Hepatitis C: IL28B gene influences success of interferon-alpha and ribavirin therapy



Plavix: genetically, 21% of people have reduced efficacy, 3% greatly reduced



Relative Finder: Discover your cousins!



Relative Finder: Discover your cousins!

relative finder^{beta}



Video: Just What Is A Fifth Cousin?

List View **Discovery View**

Filter Predicted Relatives: Results 1 – 100 of 124

Parent or Child	Sibling	Aunt/Uncle, Nephew/Niece, or Half-Sibling	4th Cousin	5th Cousin			

Ancestry

Paternal Haplogroup: I1

Locations of haplogroup I1 circa 500 years ago, before the era of intercontinental travel.



Haplogroup I1 can be found at levels of 10% and higher in many parts of Europe, due to its expansion with men who migrated northward after the end of the Ice Age about 12,000 years ago. It reaches its highest levels in Denmark and the southern parts of Sweden and Norway.

Maternal Haplogroup: V

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



Haplogroup V originated in Iberia during the Ice Age. After a last burst of cold conditions roughly 12,000 years ago, migrations carried the haplogroup northward along the Atlantic coast and through central Europe to Scandinavia. Today it is found in a wide variety of populations from the Basques of Spain to the Saami of Finland.

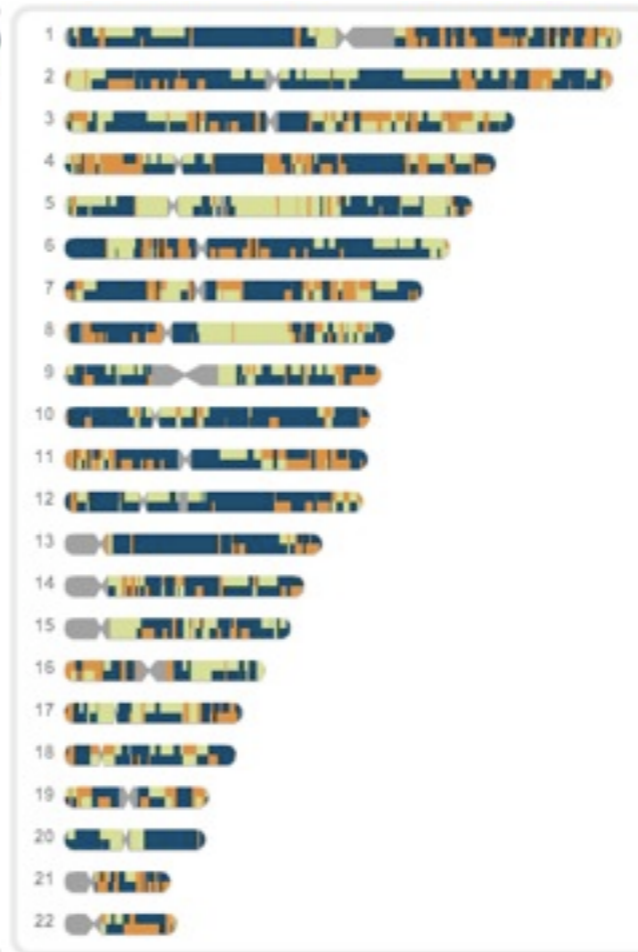
Ancestry painting



European



African-American



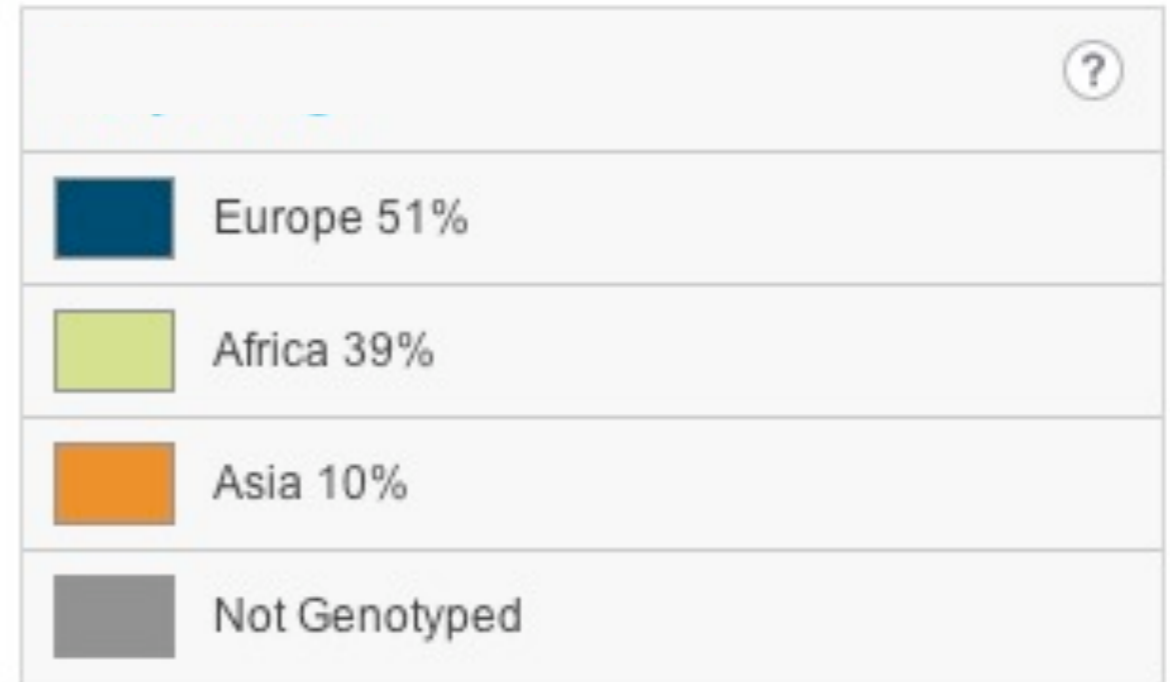
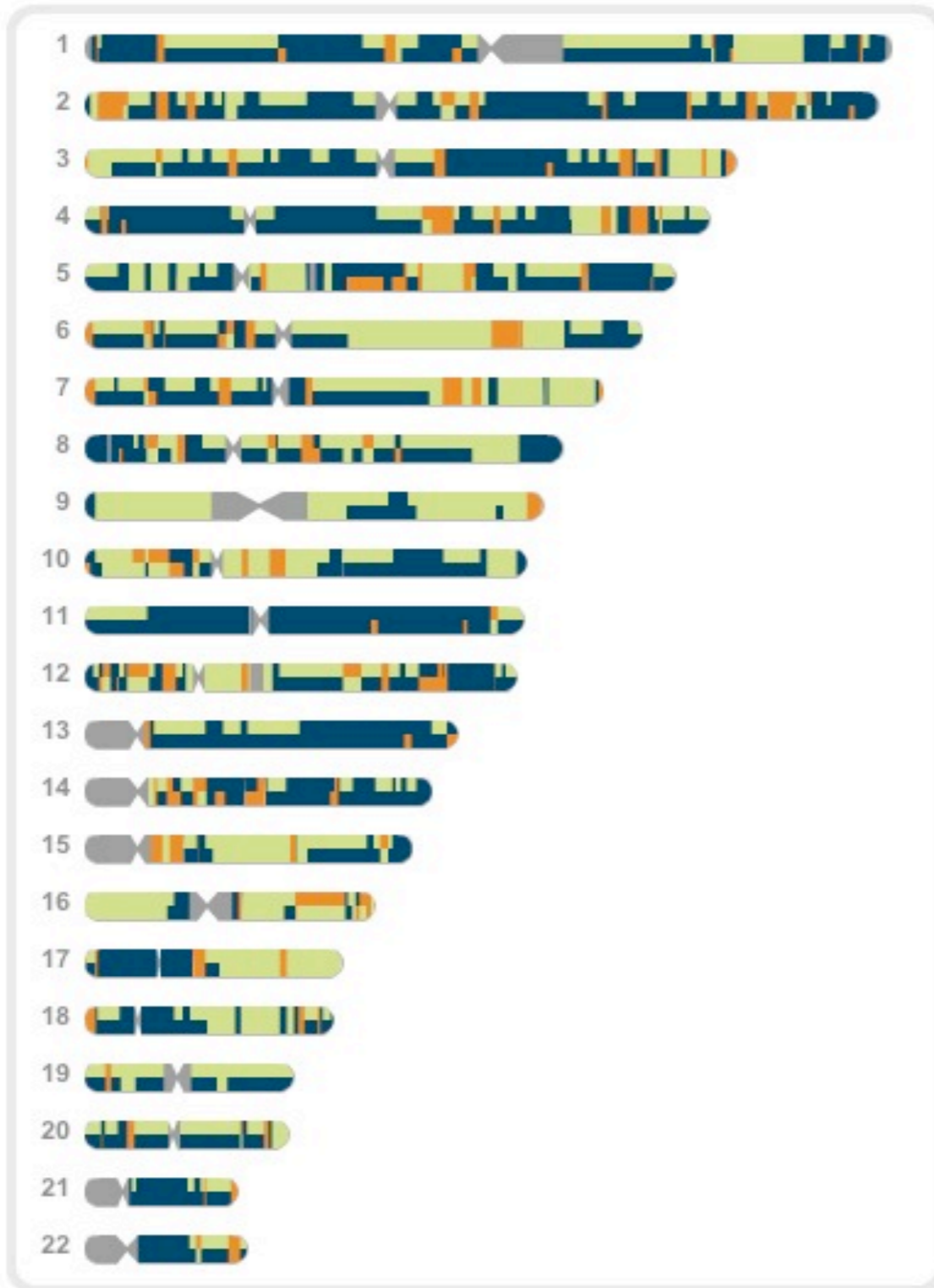
Colombian



European and Asian parents

■ Europe ■ Asia ■ Africa


Chromosomes 1-22



Worldwide Examples

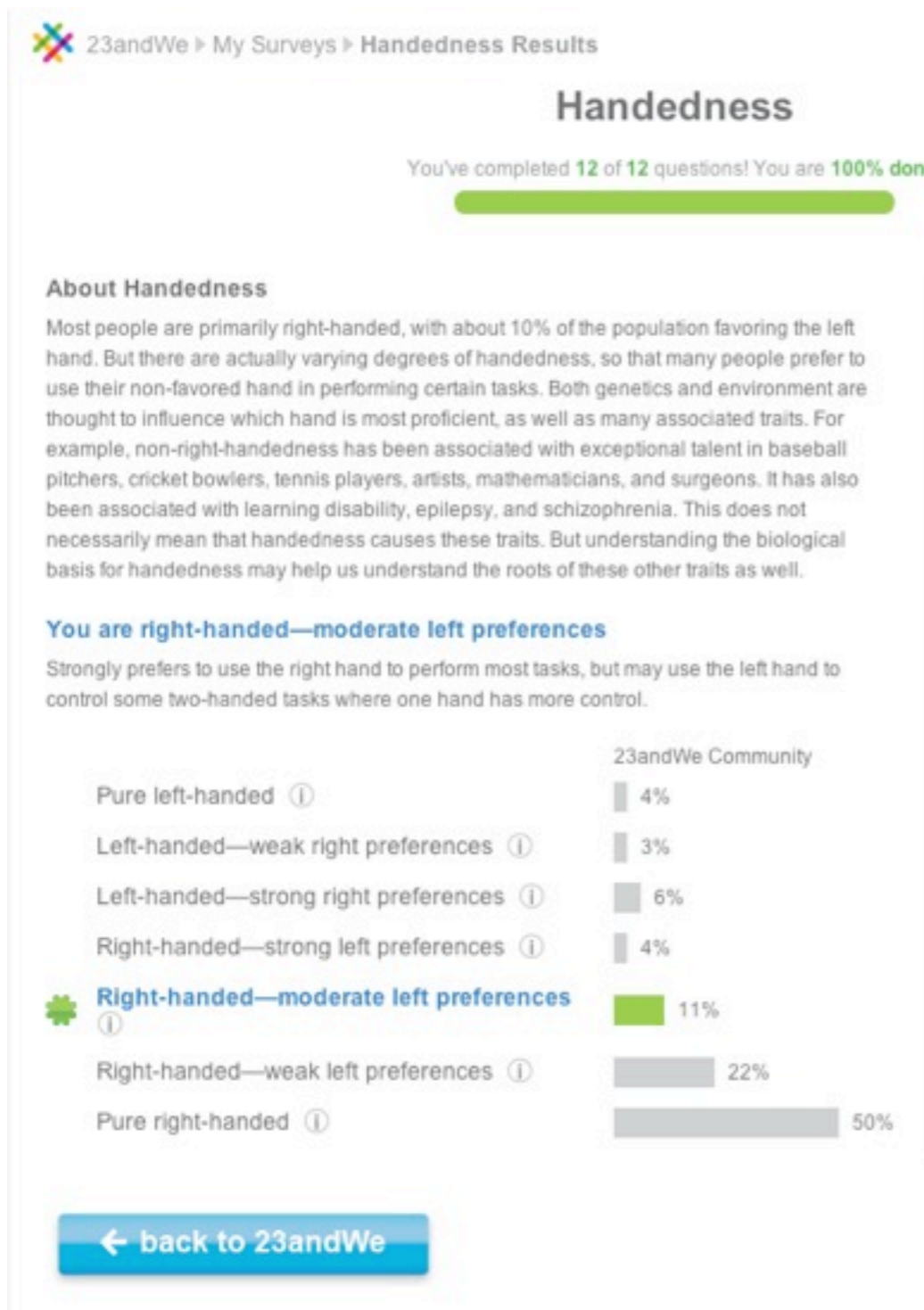
Click on the icons in the map below to see sample paintings of individuals from across the globe.





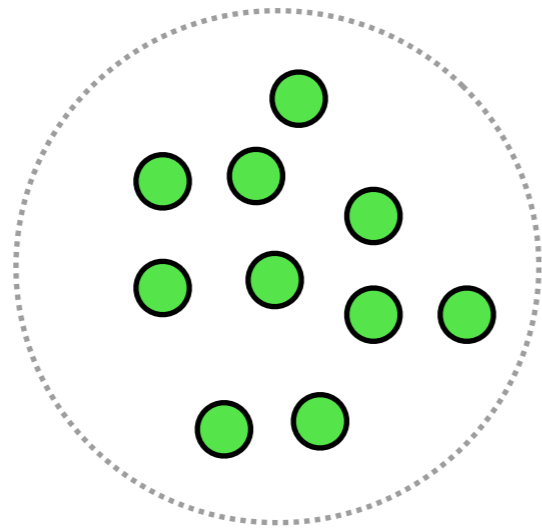
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Web-based methods



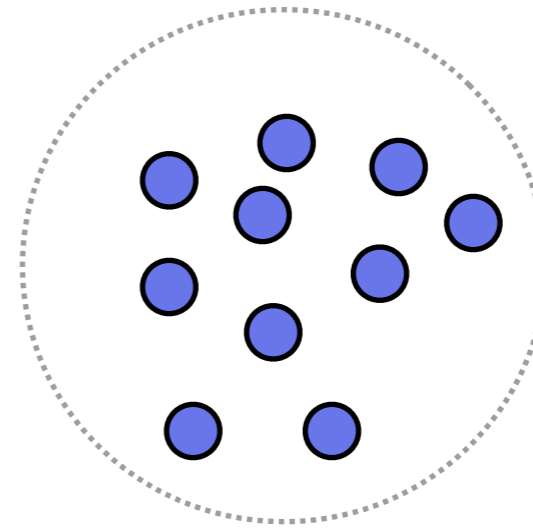
- We can ask questions on topics it is **difficult to get funding** for
- We can **recontact** easily
- We can engage consumers in research. Users can even **form groups** to try to learn more about their disease of interest.
- We can take advantage of the **interactivity of the web**

Typical genetic association study



1000

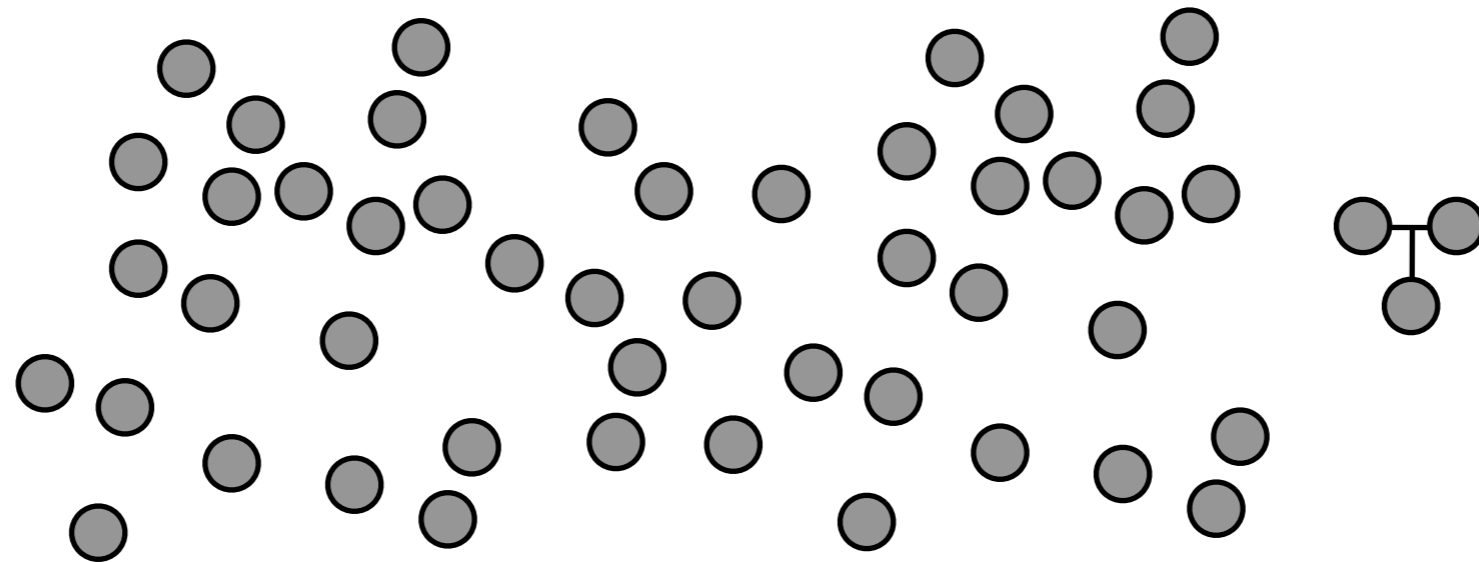
patients with
type 2 diabetes



1000

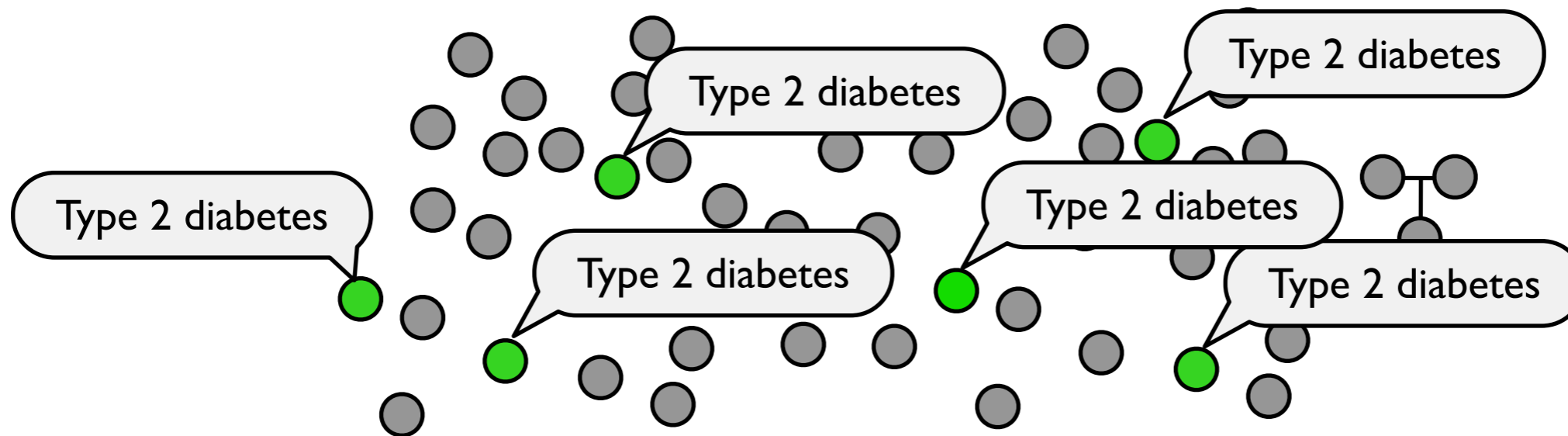
healthy controls

23andMe association study



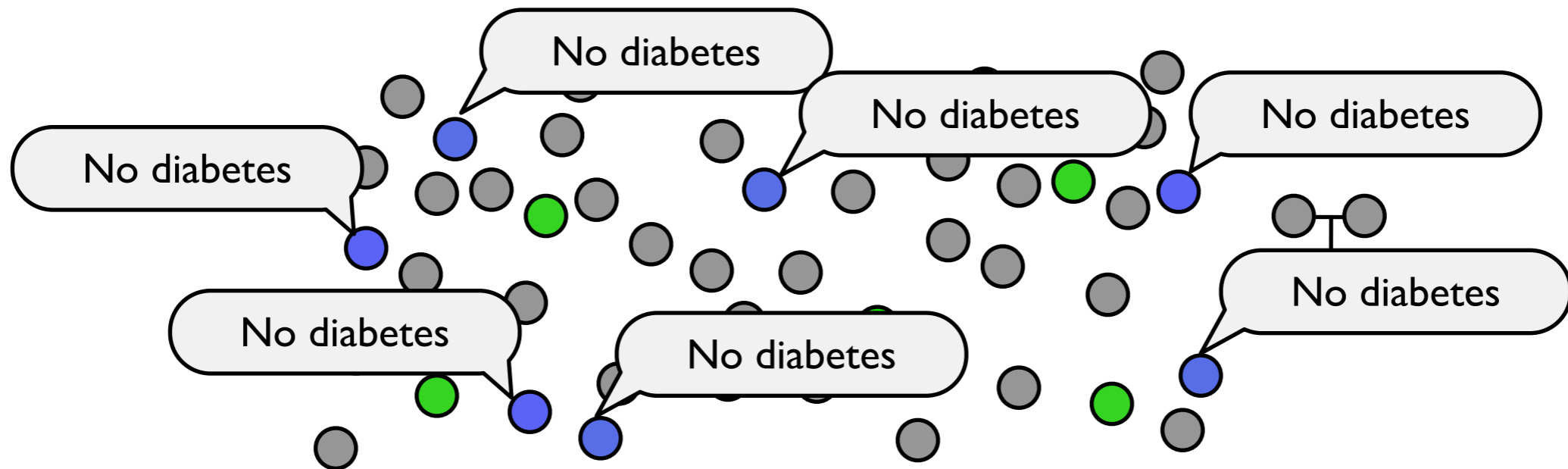
A heterogeneous population
including families

23andMe association study



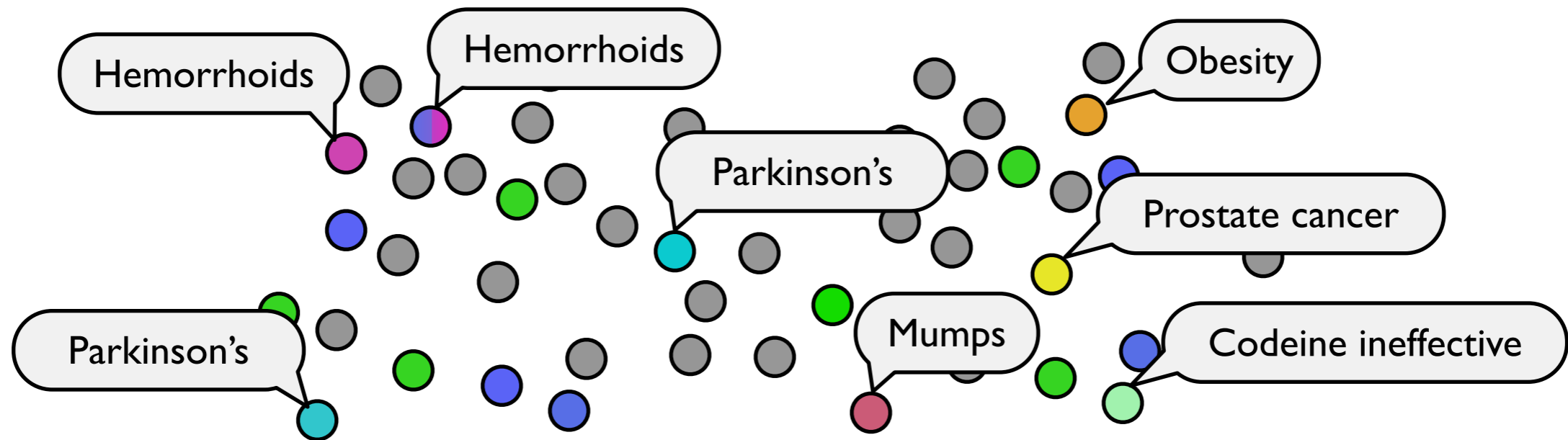
Participants declare having
type 2 diabetes

23andMe association study



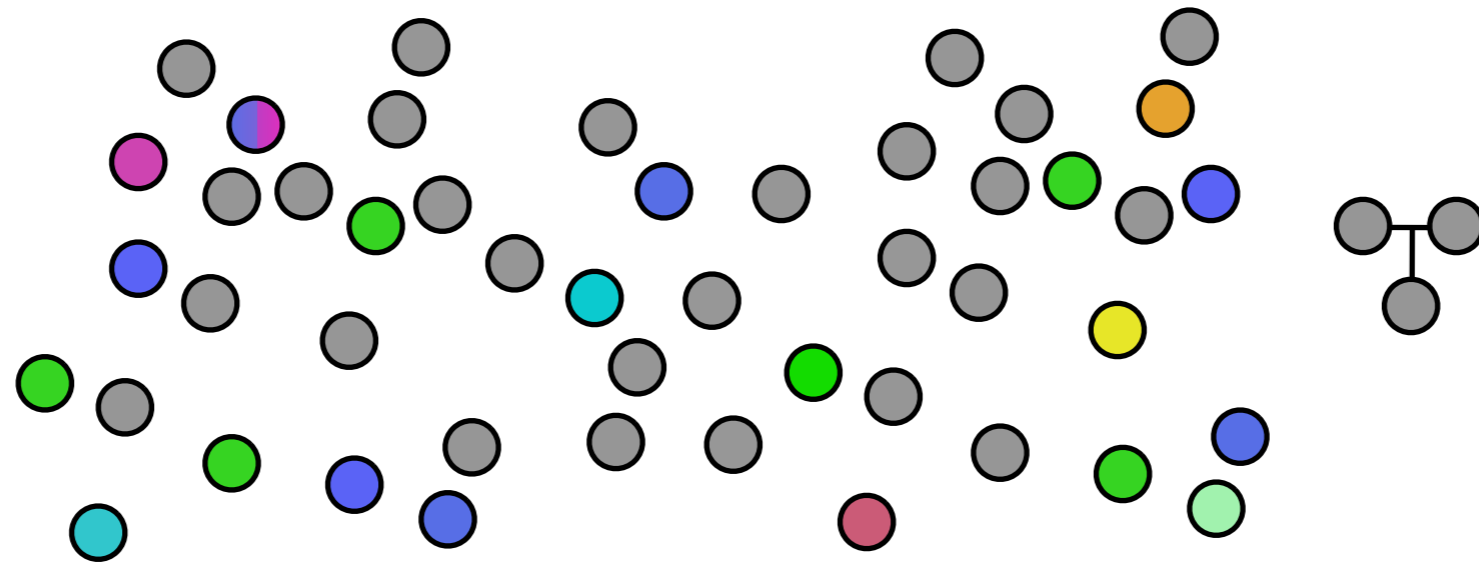
Some participants declare not having
type 2 diabetes.
Others are *population controls*.

23andMe association study

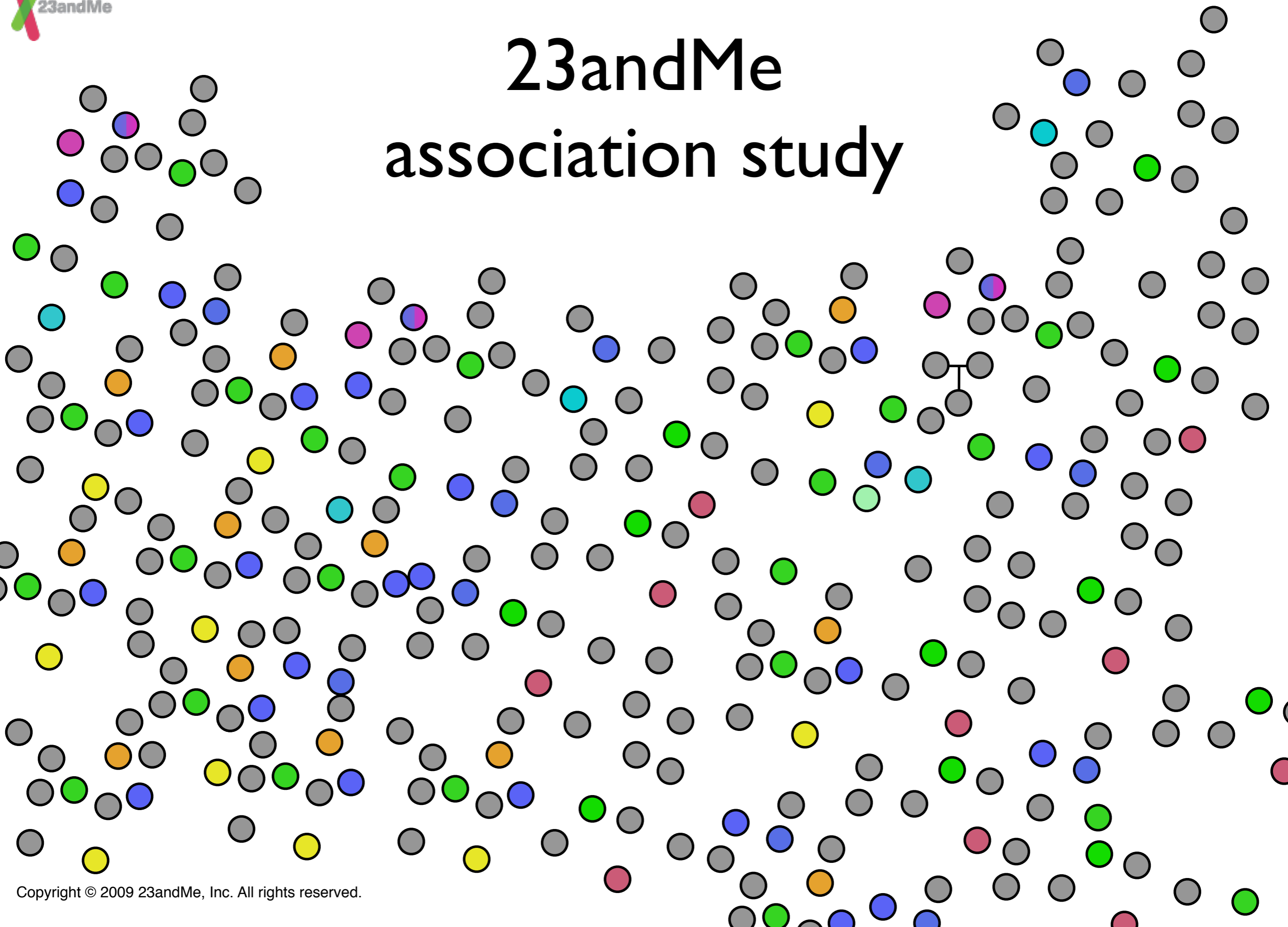


Participants can have
other diseases

23andMe association study



23andMe association study



Two projects so far looking at healthy aging

- Longevity survey: how long did your relatives live?
- Sports injury survey: what sports did you play and what did they do to you?
- These are the largest studies so far searching for a genetic basis to longevity and sports injuries.

Longevity survey

- Over 4500 responses
- over 200 with a centenarian in family
- over 2400 with a nonagenarian in family
- almost 500 people over 60, without major disease, and physically active
 - over 125 of them over 70



Sports injuries

- Almost 3000 responses
- ACL tear (200+ people)
- Tendinitis (1000+ people)
- Rotator cuff tear (350+ people)

Parkinson's disease

4000 patients enrolled in a year.

The Senior Games cohort has contributed heavily as
(generally) unaffected controls



Parkinson's results

- Have identified four genes known to be involved in PD: *LRRK2*, *SNCA*, *GBA*, and *MAPT*.
- We are validating three to five more genes.
- Could lead to major advances in PD research.

Many other contributions

- Senior games cohort has contributed to new discoveries about the genetic basis of
- hair curl, asparagus anosmia, photic sneeze reflex, attached earlobes, and many more traits and conditions
- Multiple papers to come in the next 6 months...



Community

“As for this site, it makes me feel like I am not alone, there is someone out there I can talk to, even if it is just to compare symptoms. Even though I have become actively involved in 2 PD groups, These conversions really help. Thank you to everone who takes the time to read and reply.”