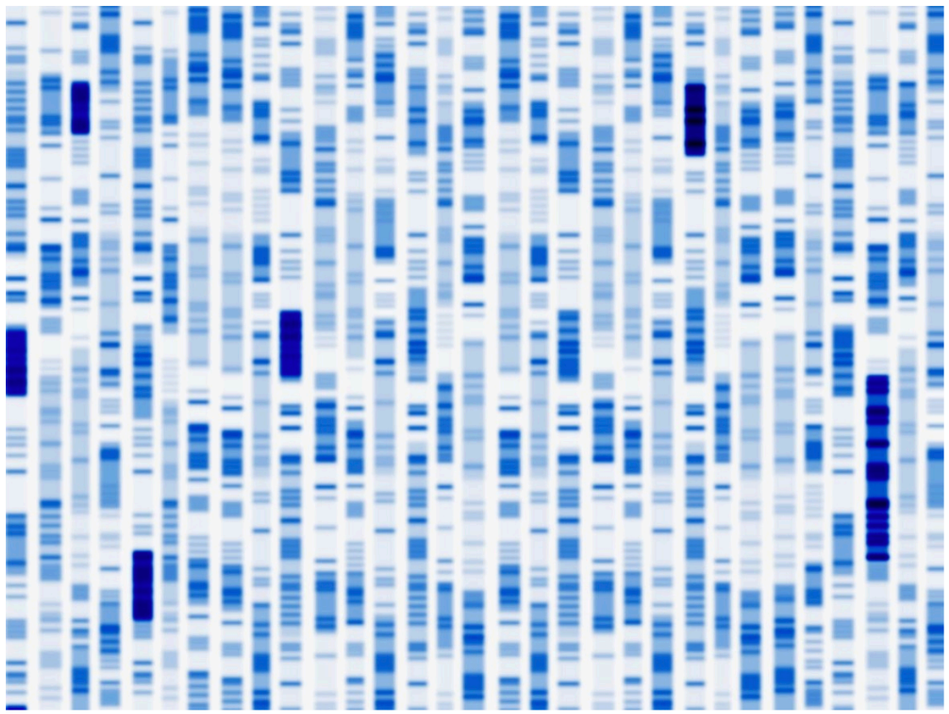
Welcome to The Age of Personal Genomes

Carl Zimmer

February 3, 2020, Yale MBB

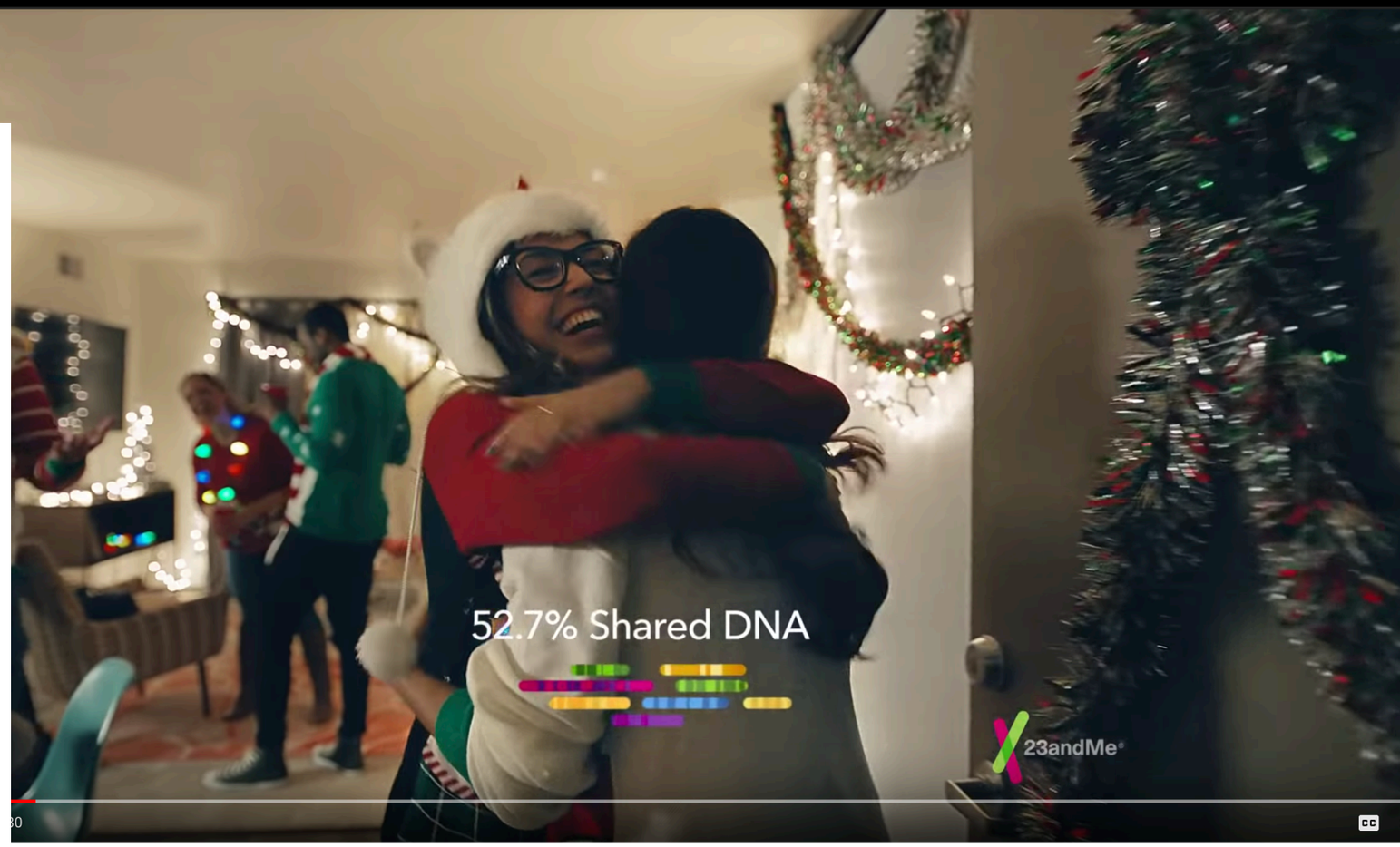
MEGAN MOLTENI SCIENCE 12.01.17 07:00 AM

ANCESTRY'S GENETIC TESTING KITS ARE HEADING FOR YOUR STOCKING THIS YEAR



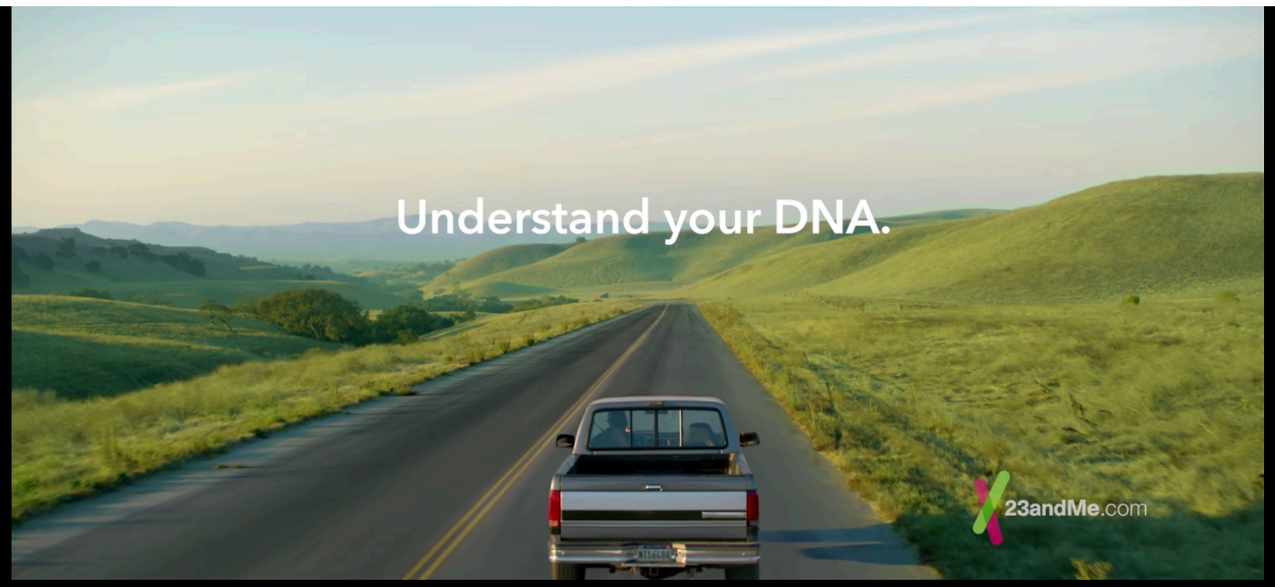
ALFRED PASIEKA/SCIENCE PHOTO LIBRARY/GETTY IMAGES

wired.com/story/ancestrys-genetic-testing-kits-are-heading-for-your-stocking-this-year/

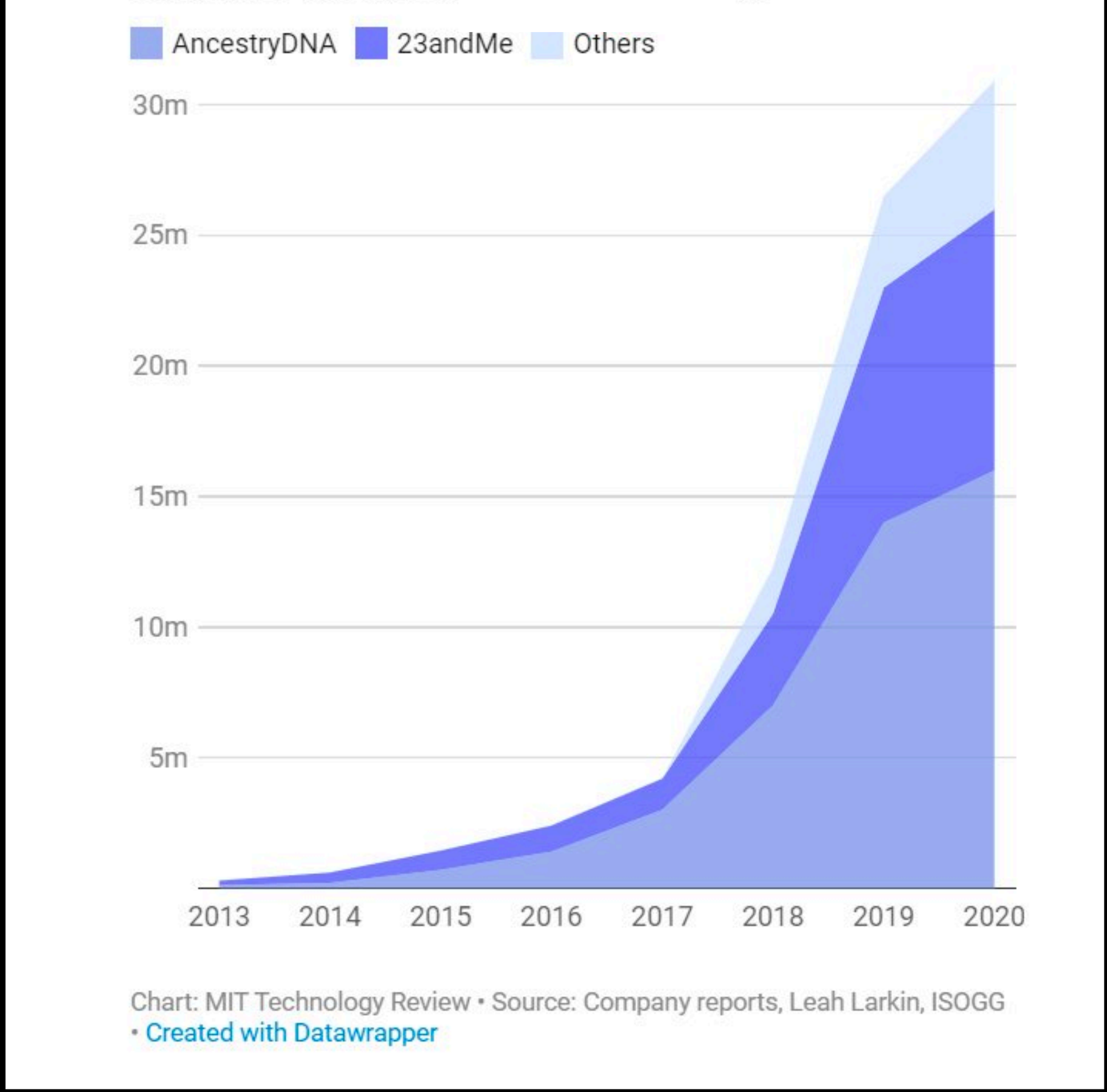


For Everyone You Love!

Up next



ispot.tv/ad/wHZ4/23andme-one-body-one-mind



https://twitter.com/antonioregalado/status/1220466266361815043/photo/1

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.

Jamie, you **do not have** the ε4 variant we tested.

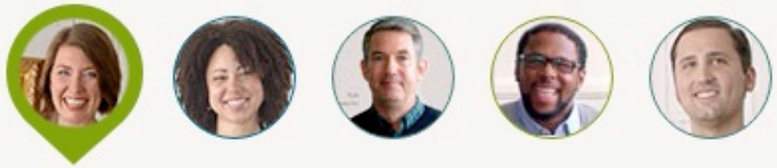
Your risk for Alzheimer's disease also depends on other factors, including lifestyle, environment, and genetic variants not covered by this test.

0 variants detected

in the APOE gene

23andme.com/dna-reports-list/

Stories from Our Members



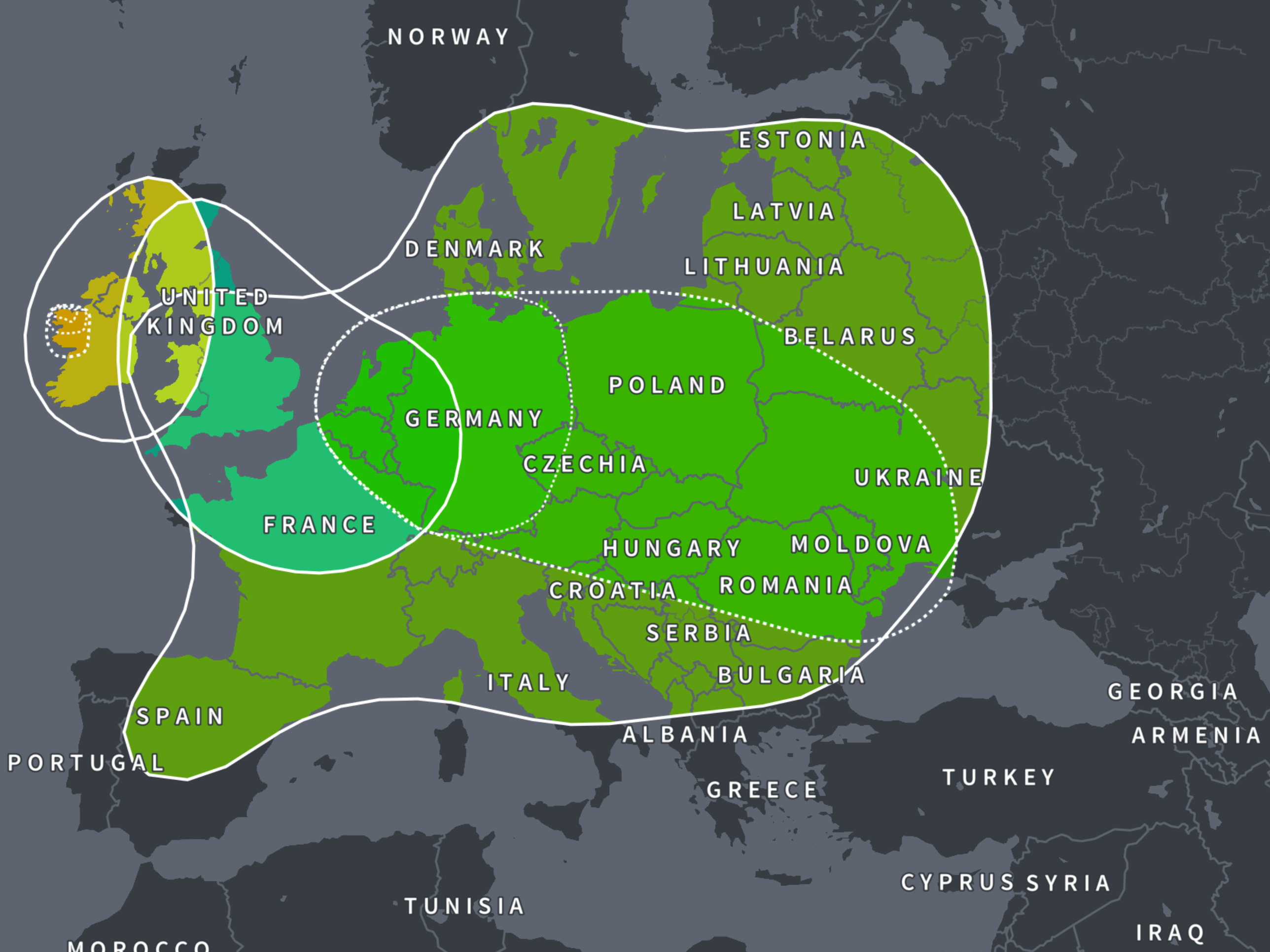
“Holy crow! I'm related to George Washington.”

Just days after beginning her family history search, Emily discovered a truly legendary ancestor. With the help of Ancestry Hints, she traced her family all the way back to her ten-times great grandmother, who just so happened to also be George Washington’s aunt.

Emily found a presidential cousin—who could be hiding in your family tree?

Find your story

ispot.tv/ad/AZbe/ancestry-com-ancestry-testimonial-emily


NORWAY
ESTONIA
LATVIA
DENMARK
LITHUANIA
UNITED KINGDOM
BELARUS
POLAND
GERMANY
CZECHIA
UKRAINE
FRANCE
HUNGARY
MOLDOVA
CROATIA
ROMANIA
SERBIA
BULGARIA
ITALY
GEORGIA
SPAIN
ARMENIA
ALBANIA
PORTUGAL
TURKEY
GREECE
CYPRUS
SYRIA
TUNISIA
IRAQ
MOROCCO

The Powers, Perversions, *and* Potential *of* Heredity

"Magisterial."
—*THE ATLANTIC*

"Extraordinary."
—*THE NEW YORK TIMES*

"Engrossing."
—*WIRED*

CARL ZIMMER

"SCIENCE BOOK OF THE YEAR."—*THE GUARDIAN*

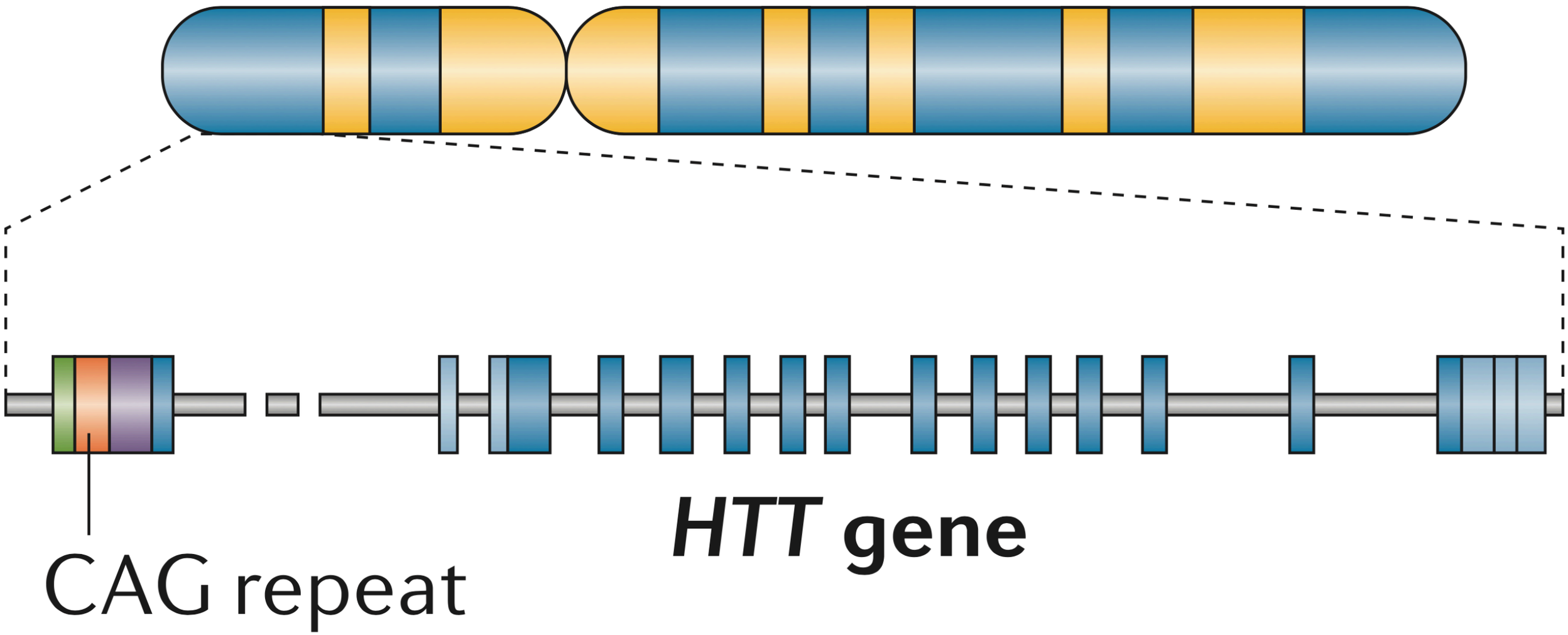
pistil ♀ \ pollen ♂	**B**	**b**
B	**BB**	**Bb**
b	**Bb**	**bb**

Gregor Mendel
(1822-1884)

Wikipedia

Woody Guthrie

Chromosome 4



Bates et al, *Nature Reviews Disease Primers* 2015

Huntingtin

Bates et al, *Nature Reviews Disease Primers* 2015

Predictive Testing for HUNTINGTON DISEASE

Healthcare Provider Area Login

Search:

1. WHAT IS HD? | 2. TESTING FOR HD | 3. RESOURCES

How Is It Done? | Prenatal Testing | Deciding To Test | Alternatives | Interpreting Results | Undergo Testing

What is Predictive Testing?

This section is intended to help the individual considering testing for HD reflect on some of the issues involved in testing and in dealing with the test results.

Family, friends and professional support people may also find this material useful in supporting those considering testing.

In 1983, genetic markers closely linked to the Huntington disease (HD) gene were identified. This discovery, together with the identification of additional genetic markers, led to the development of predictive testing

HD Resources

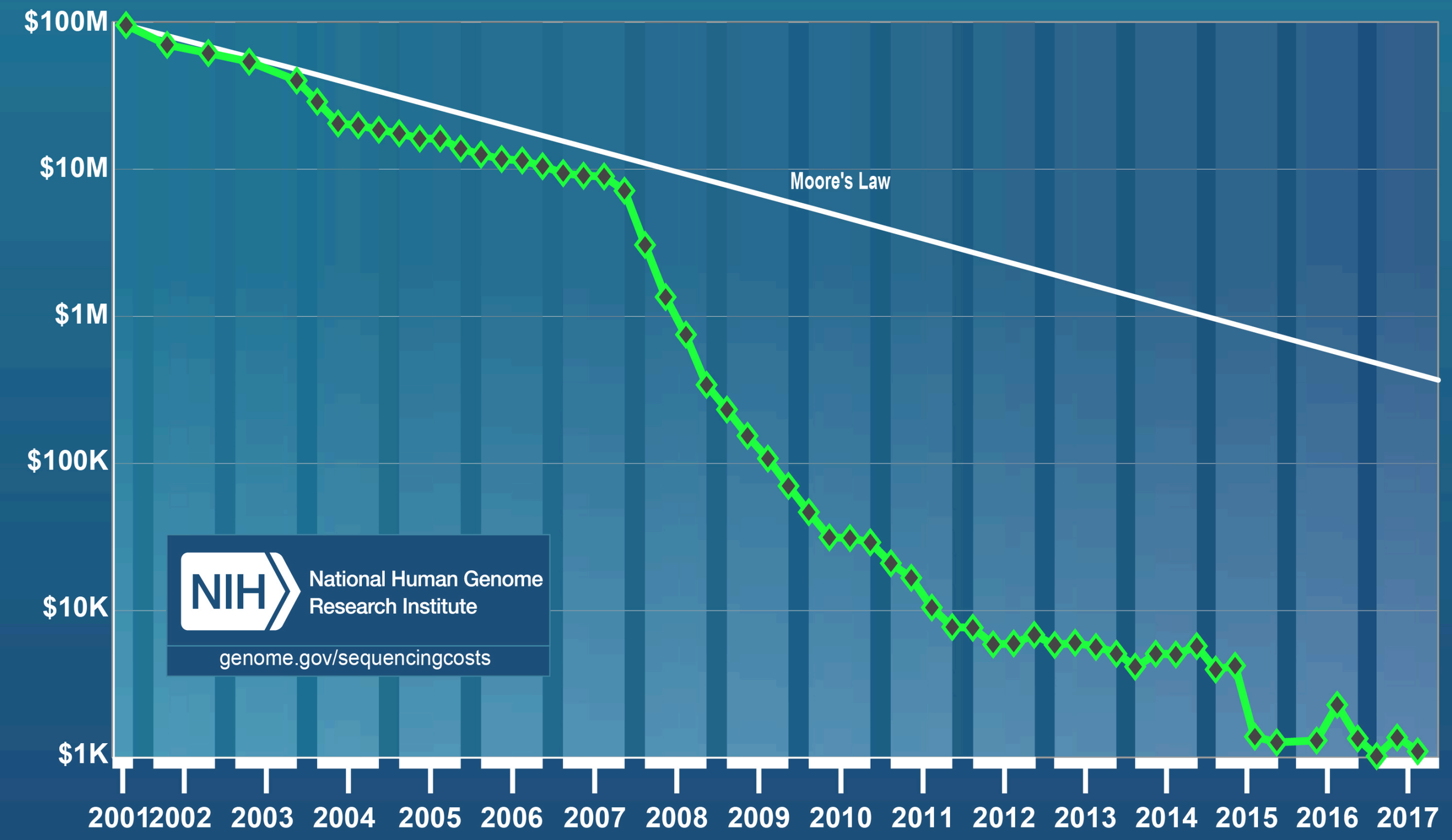
There are many other online websites and resources which provide information regarding HD in general, support groups in your area, research updates and opportunities to be involved in clinical trials.

Find out more

Our Stories

We understand that learning that someone in your family has HD can be devastating. It can leave you with questions, concerns, and no idea where to turn next. Find about more about what others have done in your situation – you are not alone.

Cost per Genome



UNDERSTAND YOUR GENOME®

IT STARTS WITH YOU

THINK ME. THINK WE. *think* **BIG**

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TruGenome Predisposition Screen

Clinical Report

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TruGenome Predisposition Screen

Clinical Report

No pathogenic or likely pathogenic variants were found in the 1,691 genes evaluated that are expected to be clinically significant for the patient. However, this screen only detects single nucleotide substitutions and insertions and deletions of up to seven base pairs. Other types of genetic variants, including but not limited to larger insertions or deletions, copy number variants and trinucleotide repeats are not reported in this screening test. Further, the coverage of each gene is less than 100%. Therefore, clinically significant variants could exist in this genome that are not detected with this test. The coverage for each gene is provided in the Gene-Disease appendix.

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TruGenome Predisposition Screen

Clinical Report

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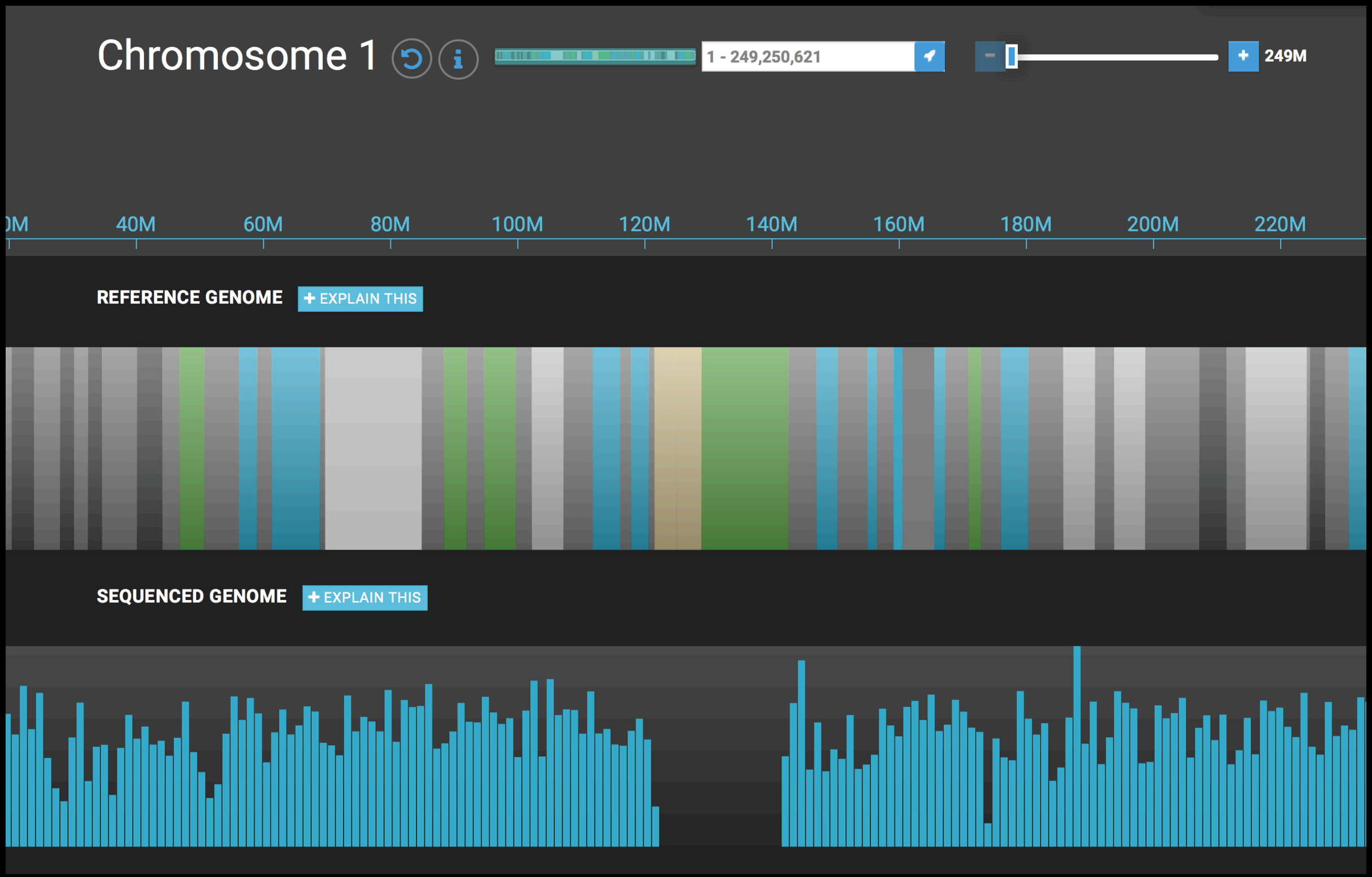
TruGenome Predisposition Screen

Clinical Report

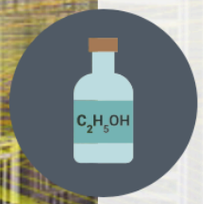
Findings Regarding Carrier Status				
Variant	**Interpretation**	**Associated Condition**	**Mode of Inheritance**	**Zygosity**
MEFV c.2177T>C (p.Val726Ala)	Pathogenic	Familial Mediterranean Fever	Autosomal Recessive	Heterozygous
MBL2 c.154C>T (p.Arg52Cys)	Likely Pathogenic	Mannose-Binding Protein Deficiency	Autosomal Recessive	Heterozygous



understandyourgenome.com

Here is what we see in your DNA...

Your odds of developing male pattern baldness are increased if you are Caucasian.



You are less likely to have flush reaction if you drink alcohol.

Your muscle fibers are built for power.

You are likely to perceive bitter tastes.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Seagate

Charles Byrne (1761–1783)

“Mr. O'Brien the Irish Giant the Tallest Man in the Known World Being Near Nine Feet High”

whitney.med.yale.edu

Joel Hirschhorn
Harvard Medical School

2007

1
variant

4921 people

Weedon et al

2007

2010

1
variant

4921 people

Weedon et al

180
variants

183,727 people

Lango Allen et al

2007

2010

2014

1
variant

4921 people

Weedon et al

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697
variants

253,288 people

Wood et al

2007

1
variant

4921 people

Weedon et al

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180
variants

183,727 people

Lango Allen et al

2014

697
variants

253,288 people

Wood et al

2018

3290
variants

693,529 people

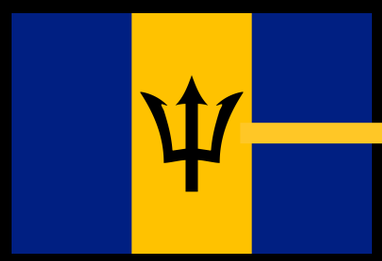
Yengo et al

AVERAGE WOMEN'S HEIGHT (cm)

Canada

157.6

Barbados



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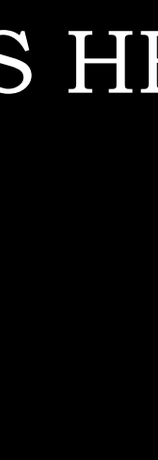


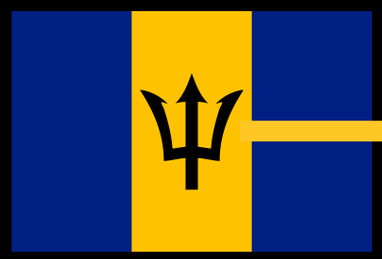
http://ncdrisc.org/height-mean-ranking.html

AVERAGE WOMEN'S HEIGHT (cm)

Canada

157.6

Barbados



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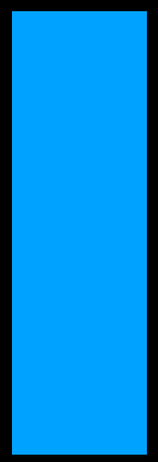

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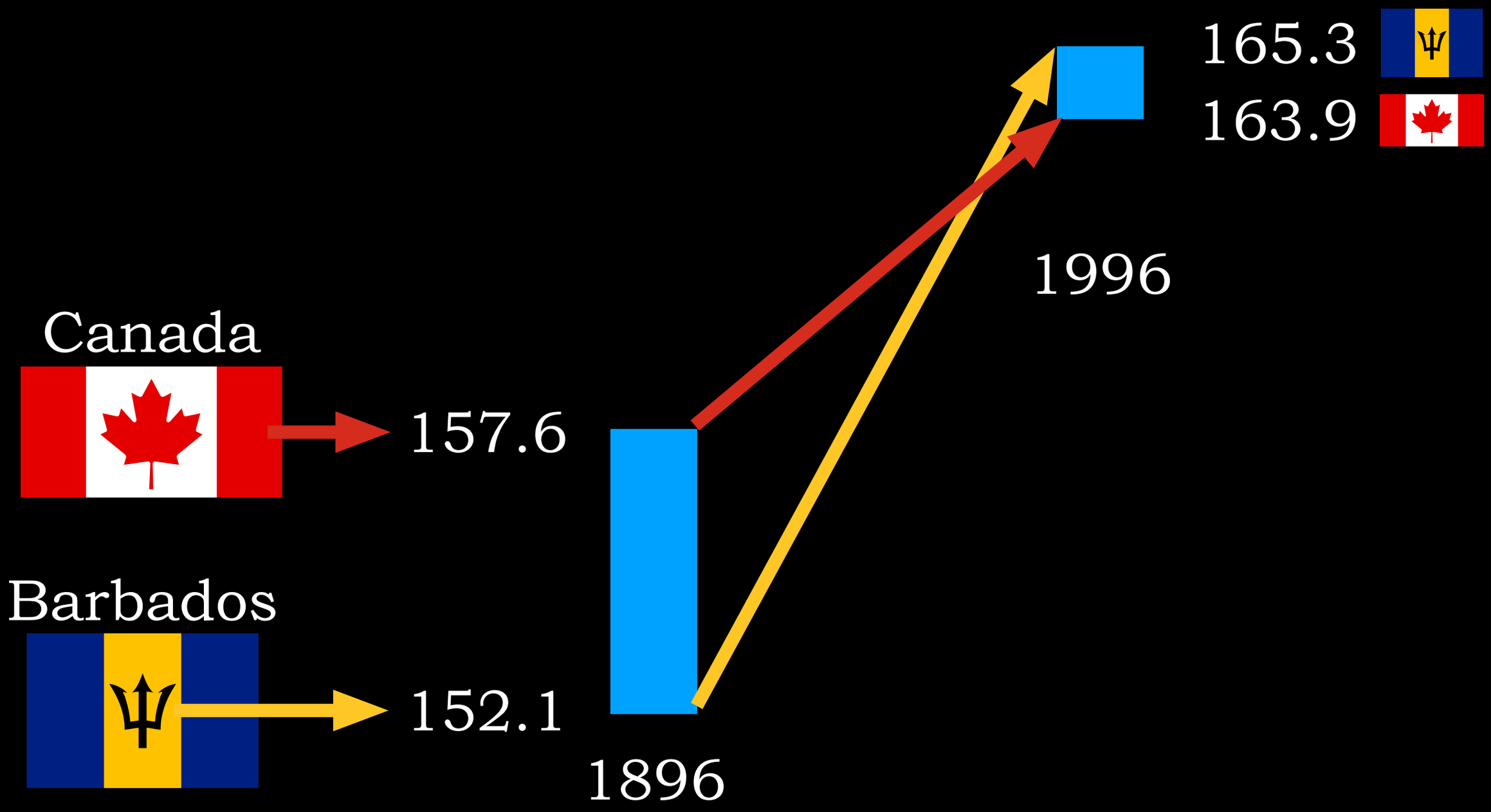
http://ncdrisc.org/height-mean-ranking.html

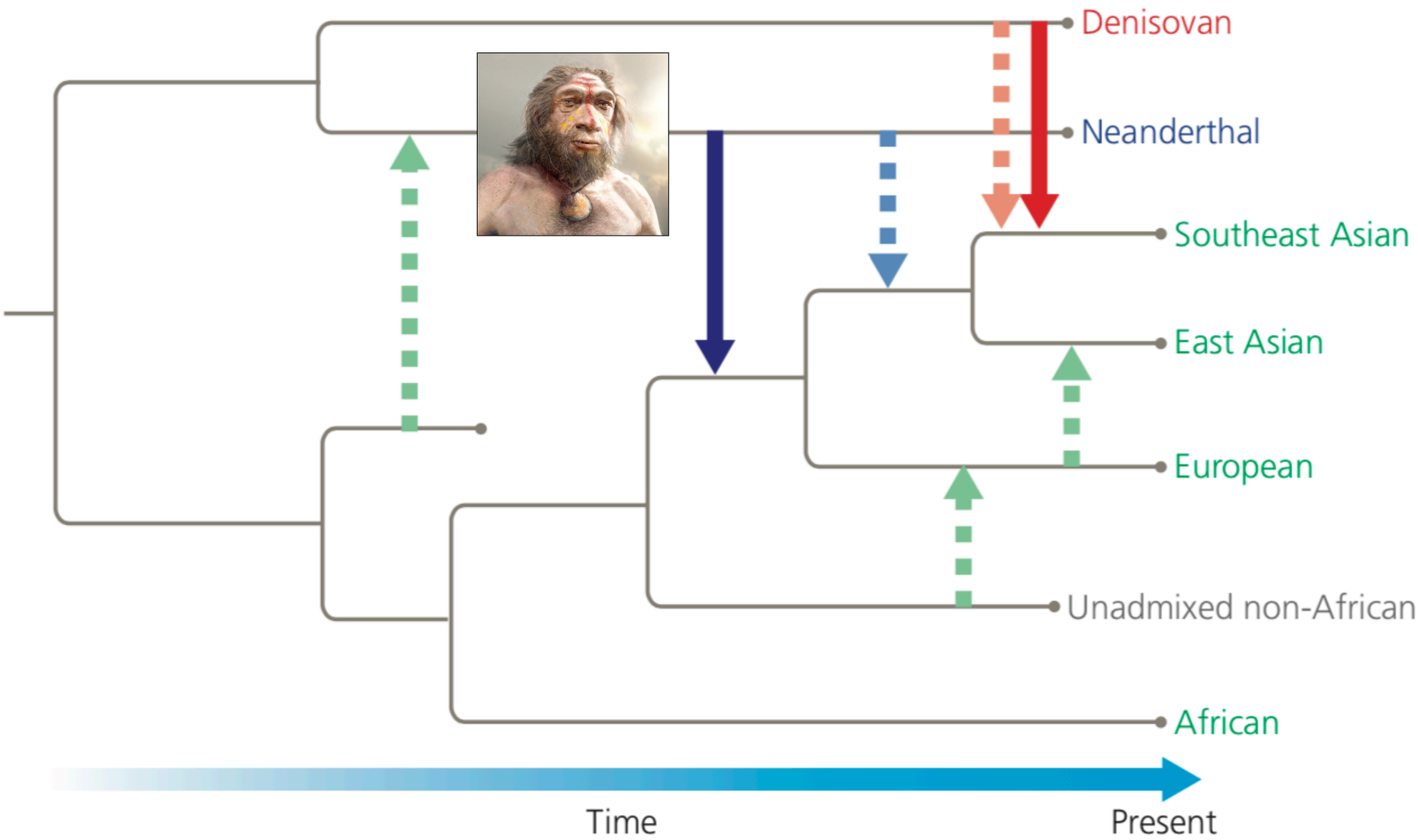
AVERAGE WOMEN'S HEIGHT (cm)



http://ncdrisc.org/height-mean-ranking.html

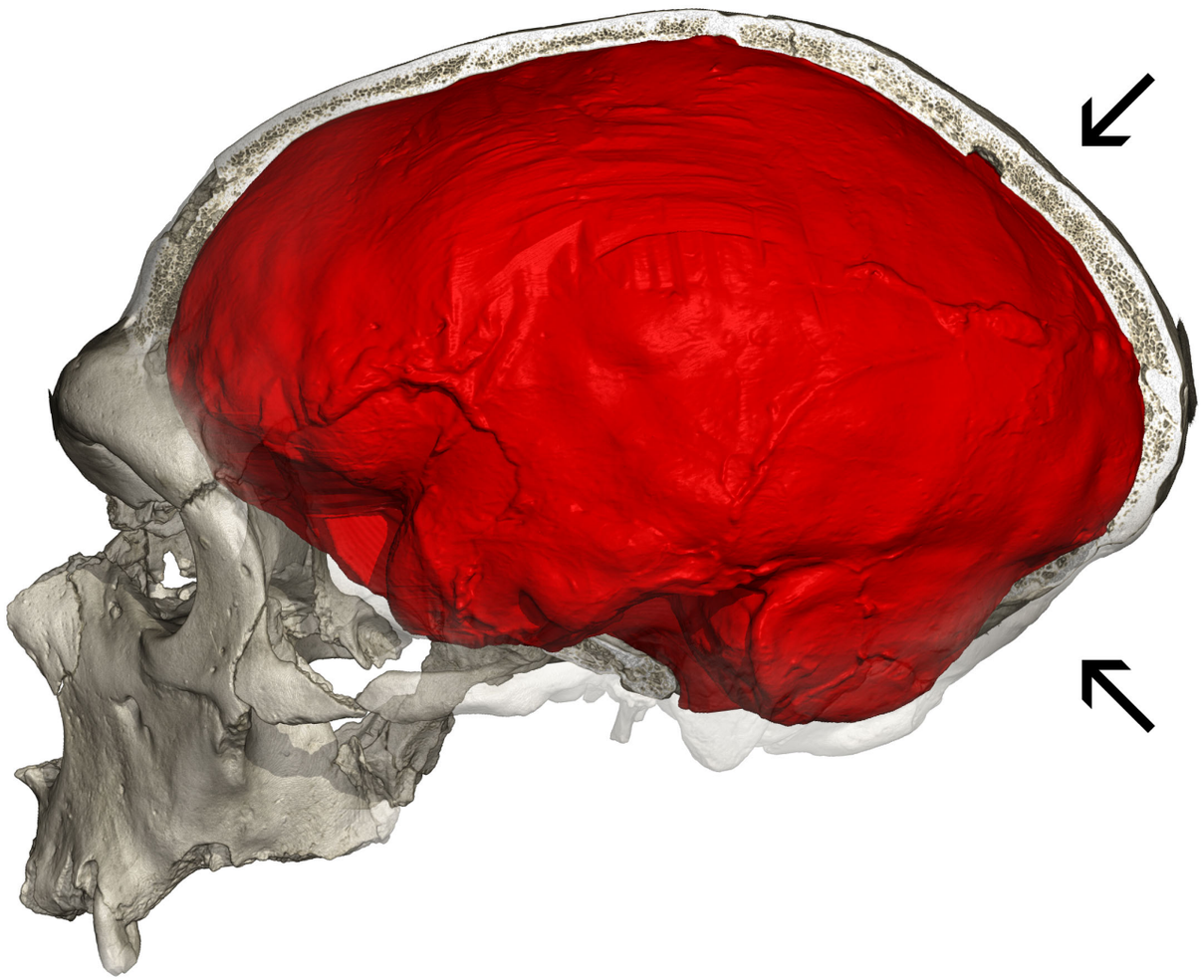
Ali Torkamani,
Scripps Translational
Science Institute

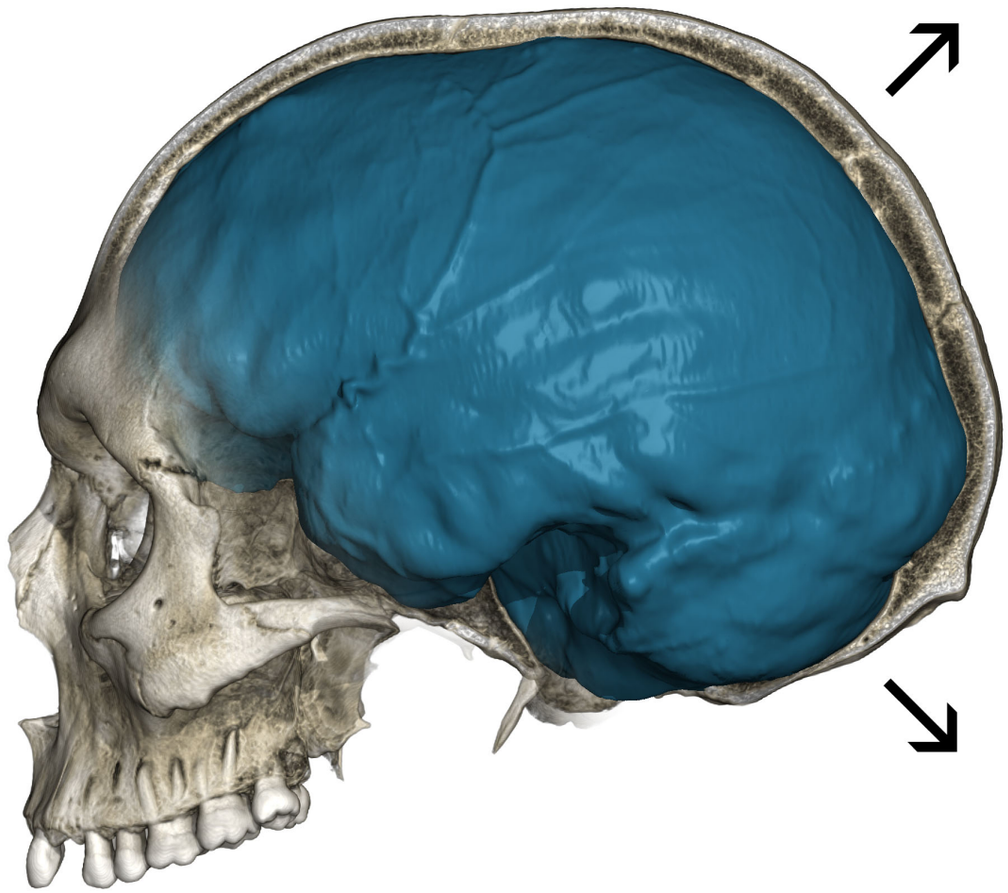
My variant: rs11209026
Gene: IL23R



hlen & Zimmer, *Evolution: Making Sense of Life* (Macmillan, 2019)

gene	chrom	gene_start	gene_end
MIR7846	chr1	12226999	12227095
MIR4632	chr1	12251769	12251830
TNFRSF1B	chr1	12227059	12269277
TNFRSF8	chr1	12123433	12204264
LRRC38	chr1	13801444	13840242
C1orf64	chr1	16330730	16333190
HSPB7	chr1	16340522	16345285
ZBTB17	chr1	16268363	16302627
CLCNKA	chr1	16348485	16360545
LDLRAD2	chr1	22138757	22151714
HSPG2	chr1	22148724	22263790
USP48	chr1	22004791	22109688
FGR	chr1	27938800	27961727
AKIRIN1	chr1	39456915	39471737
PABPC4	chr1	40026484	40042521
HEYL	chr1	40089102	40105348
OXCT2	chr1	40235196	40237020
PPIE	chr1	40204516	40229586
BMP8B	chr1	40223902	40254533
SMAP2	chr1	40839377	40888998
ZFP69B	chr1	40916336	40929390
C1orf168	chr1	57184476	57285369
LOC1019275	chr1	84041470	84326679
MIR548AP	chr1	84259597	84379059
LOC1019275	chr1	84267198	84326229
NTNG1	chr1	107682539	108027521
RPL31P11	chr1	161653494	161655042
FCGR2B	chr1	161632904	161648444
FCRLA	chr1	161676761	161684142
FCRLB	chr1	161691333	161697933
DUSP12	chr1	161719557	161726954
OLFML2B	chr1	161952981	161994255
ATF6	chr1	161736033	161933860
LINC00970	chr1	168873142	169056243
LINC01142	chr1	170240545	170253349
FAM163A	chr1	179712297	179785333
TOR1AIP1	chr1	179851176	179889212
TOR1AIP2	chr1	179809101	179846941
CEP350	chr1	179923907	180084015
FLJ23867	chr1	180167143	180169859
QSOX1	chr1	180123967	180167169





Gunz et al.: “Neandertal introgression sheds light on modern human endocranial globularity”
cell.com/current-biology/fulltext/S0960-9822(18)31470-2

Thank you

For more information, please visit carlzimmer.com

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The Powers, Perversions, *and* Potential *of* Heredity

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SHE HAS HER MOTHER'S LAUGH

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CARL ZIMMER

"SCIENCE BOOK OF THE YEAR." —*THE GUARDIAN*