

# Welcome to The Age of Personal Genomes

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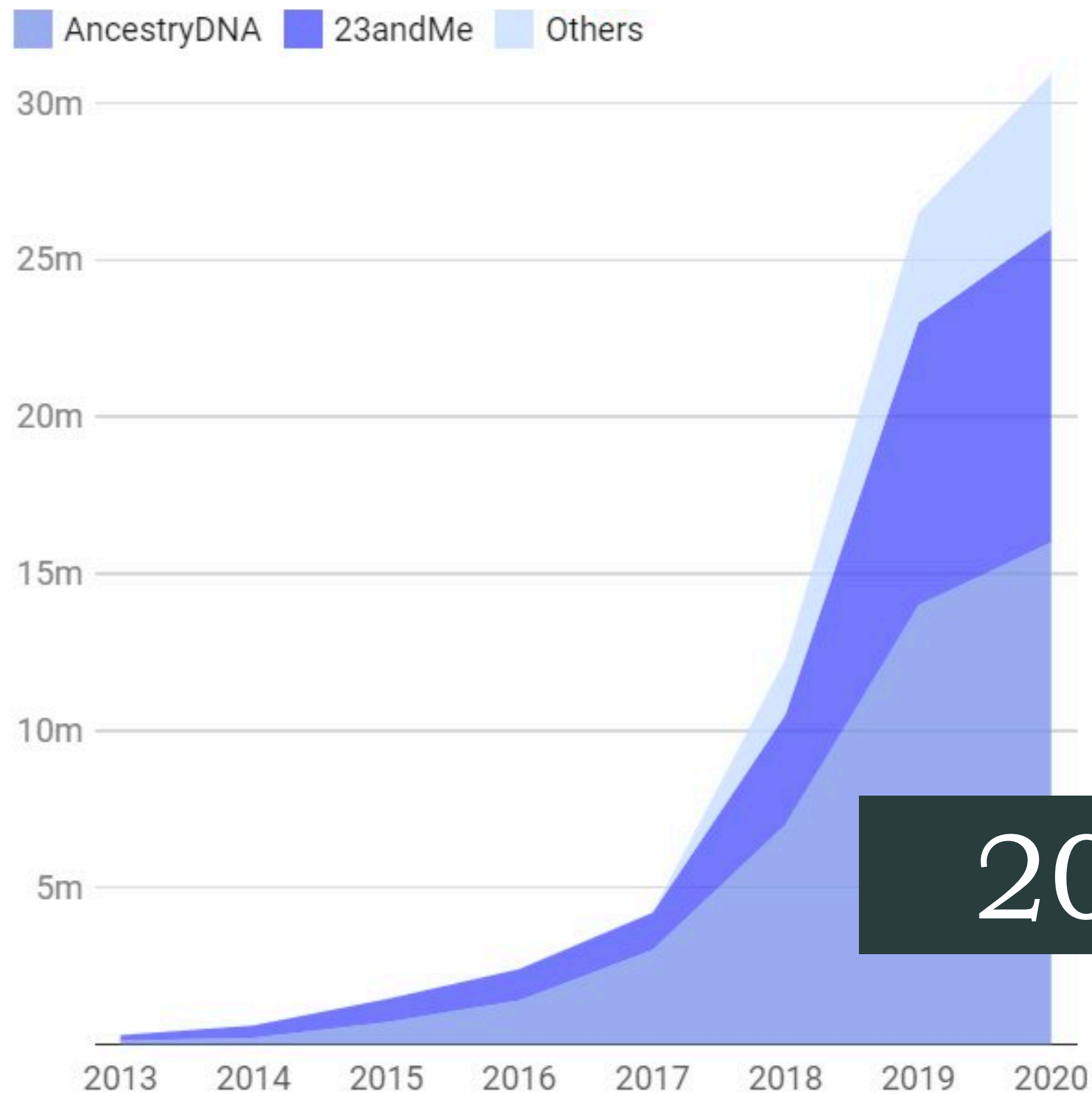
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1123	chrM	1	60	90S10M =	261
163	chrM	1	60	89S11M =	217
163	chrM	1	60	4S96M =	171
1123	chrM	1	60	93S7M =	194
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85879263  
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Carl Zimmer  
[carlzimmer.com](http://carlzimmer.com)

February 24, 2021, Yale MBB 452





2021 total: 37 million

Chart: MIT Technology Review • Source: Company reports, Leah Larkin, ISOGG  
• Created with Datawrapper



# 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.

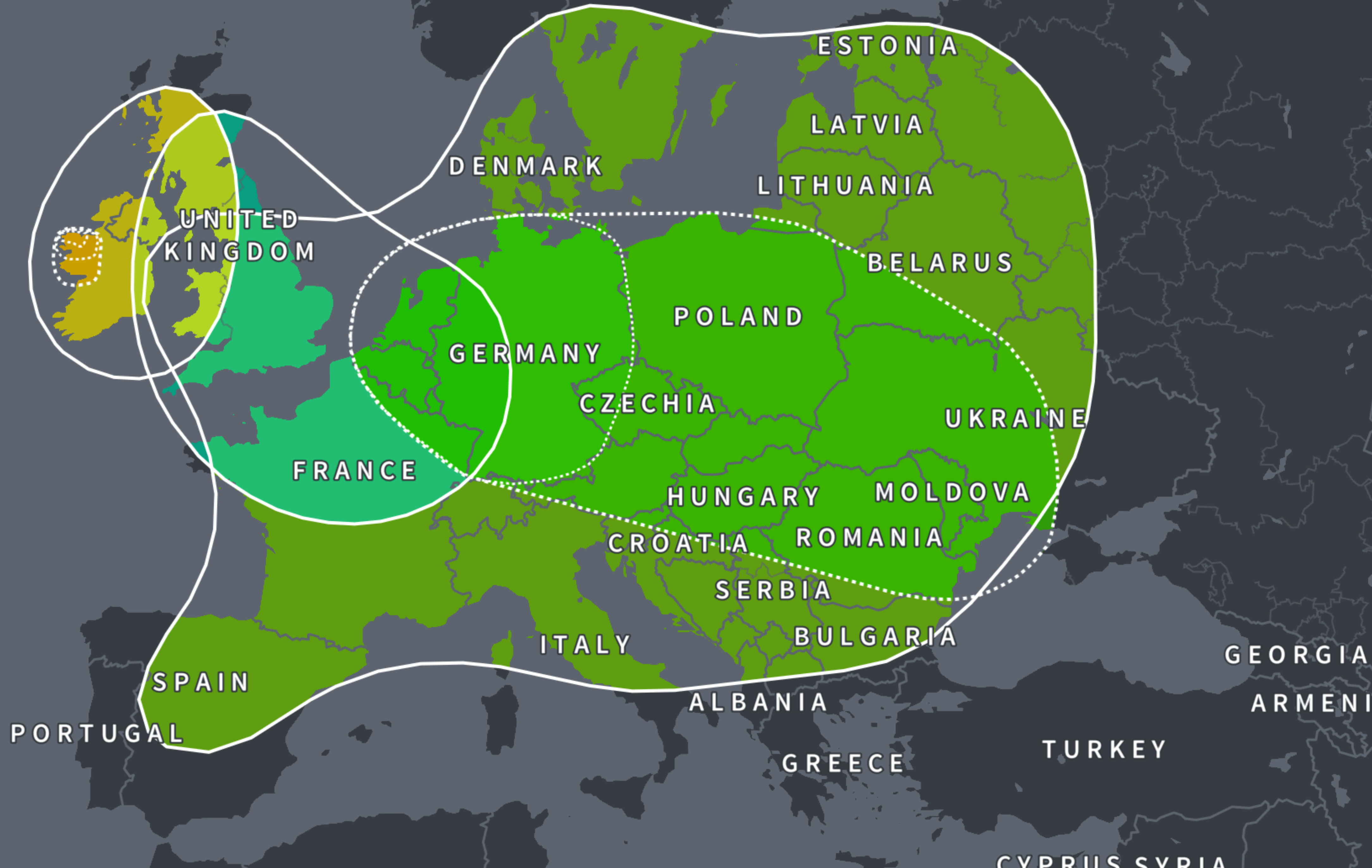
Jamie, you **do not have** the  $\epsilon 4$  variant we tested.

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

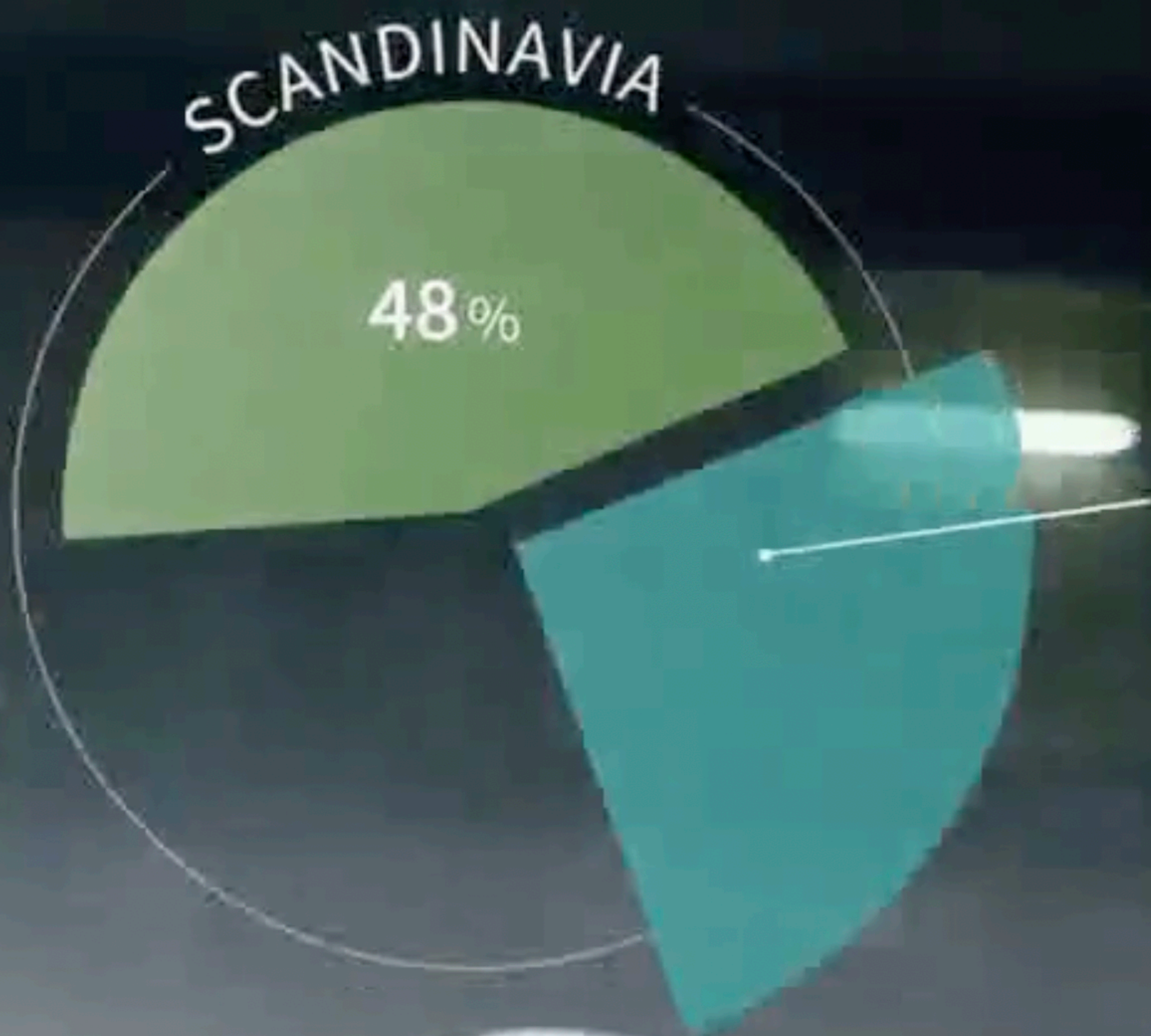


**0 variants detected**  
in the APOE gene









**27%**  
ASIA CENTRAL

<https://www.ispot.tv/ad/weoJ/ancestrydna-winter-sale-greatness>



The Powers, Perversions,  
*and Potential of Heredity*



SHE  
HAS HER  
MOTHER'S  
LAUGH

**"Magisterial."**

—THE ATLANTIC

**"Extraordinary."**

—THE NEW YORK TIMES

**"Engrossing."**

—WIRED

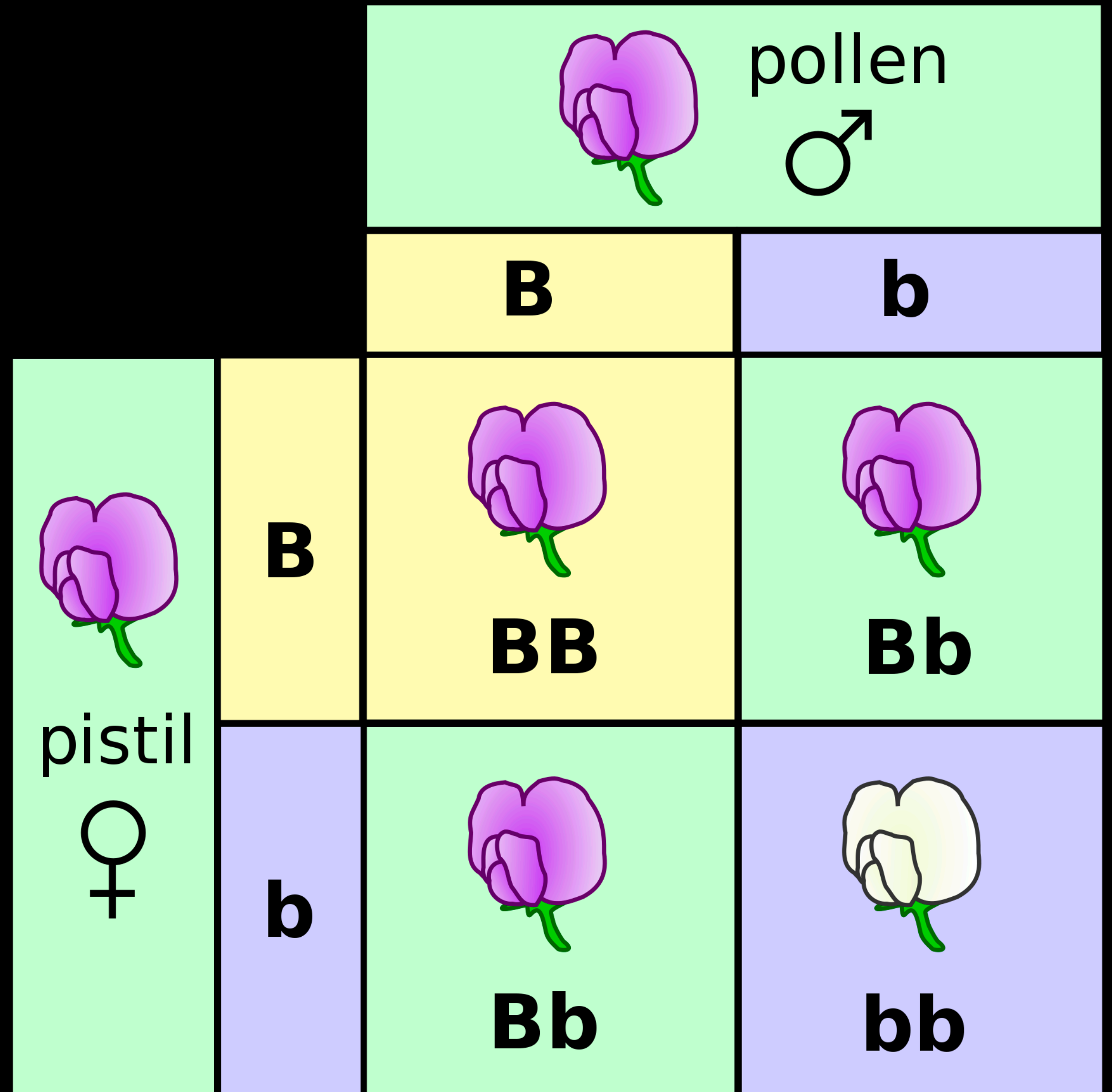
CARL ZIMMER

“SCIENCE BOOK OF THE YEAR.” —THE GUARDIAN

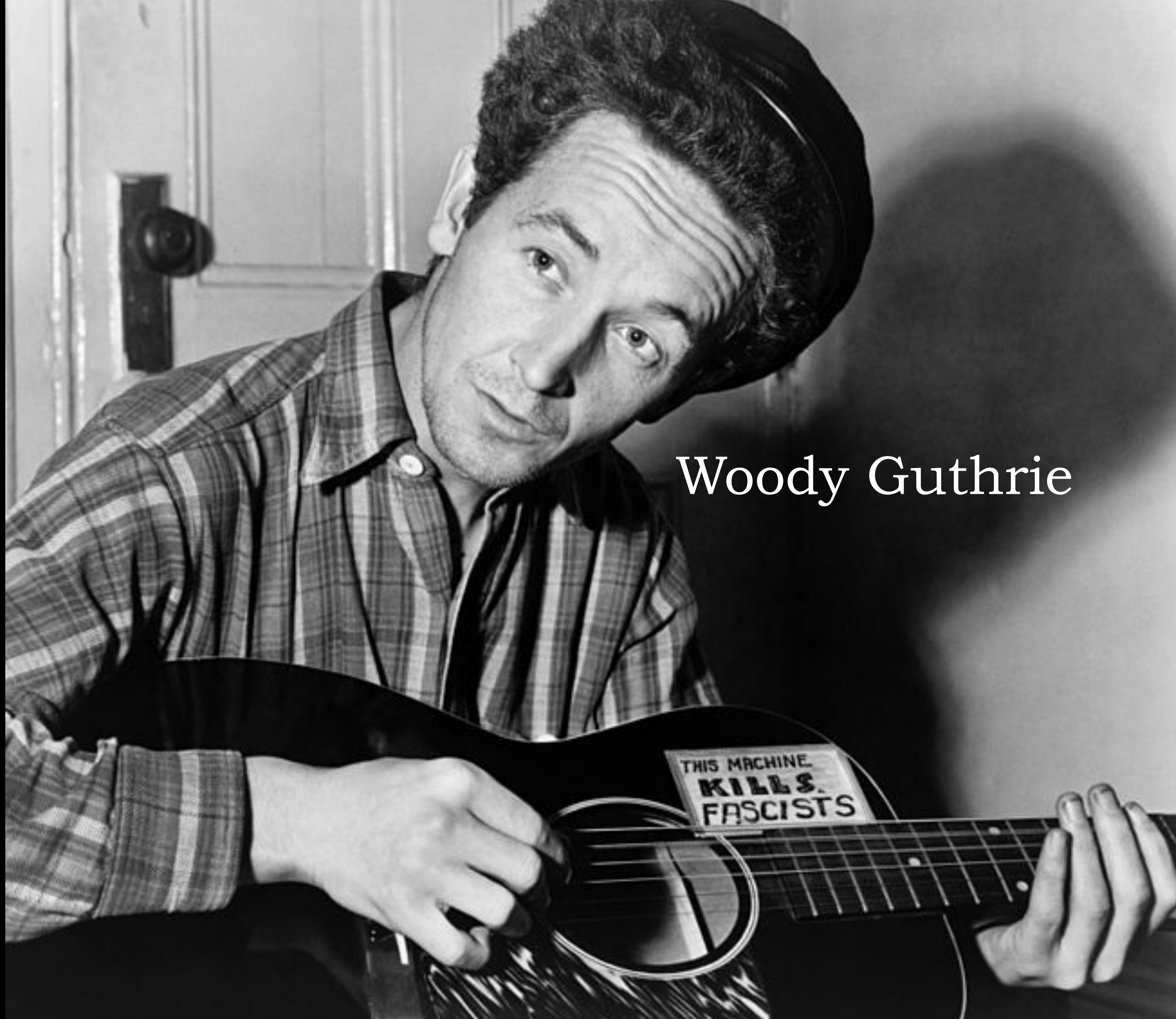




Gregor Mendel  
(1822-1884)



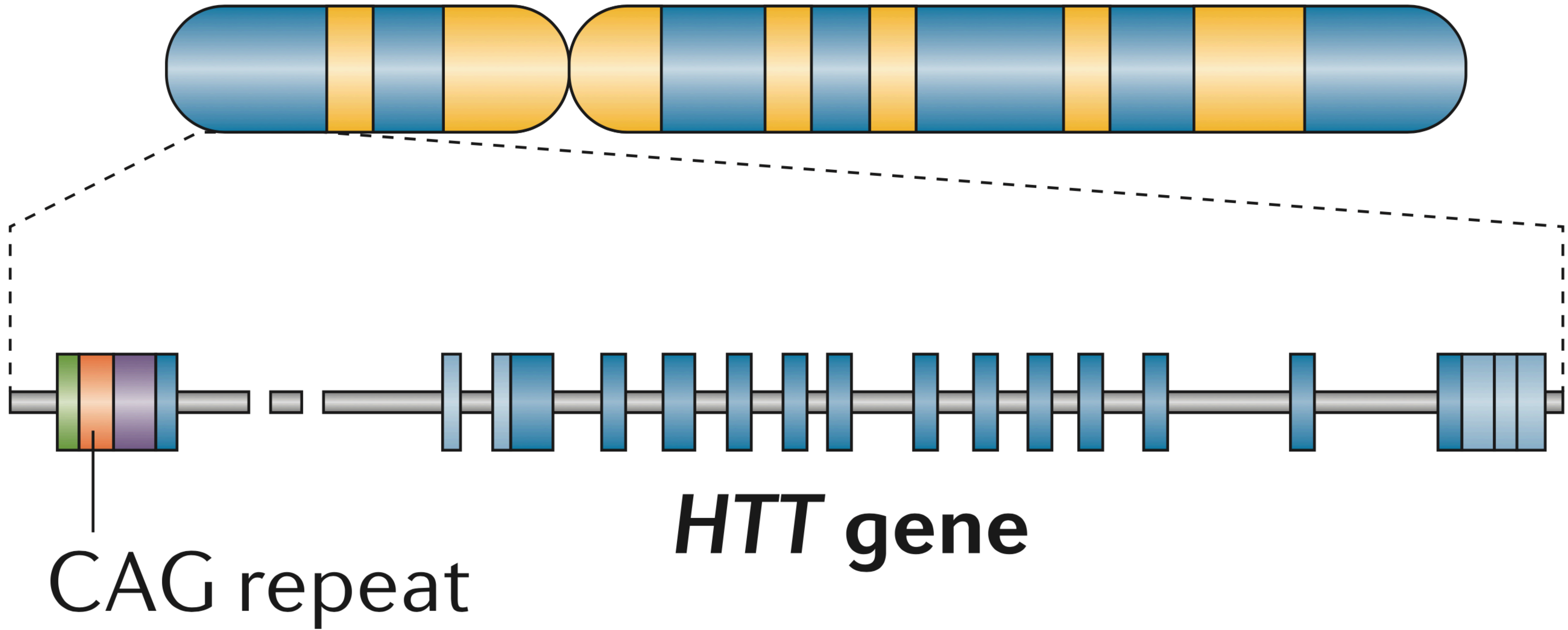




Woody Guthrie

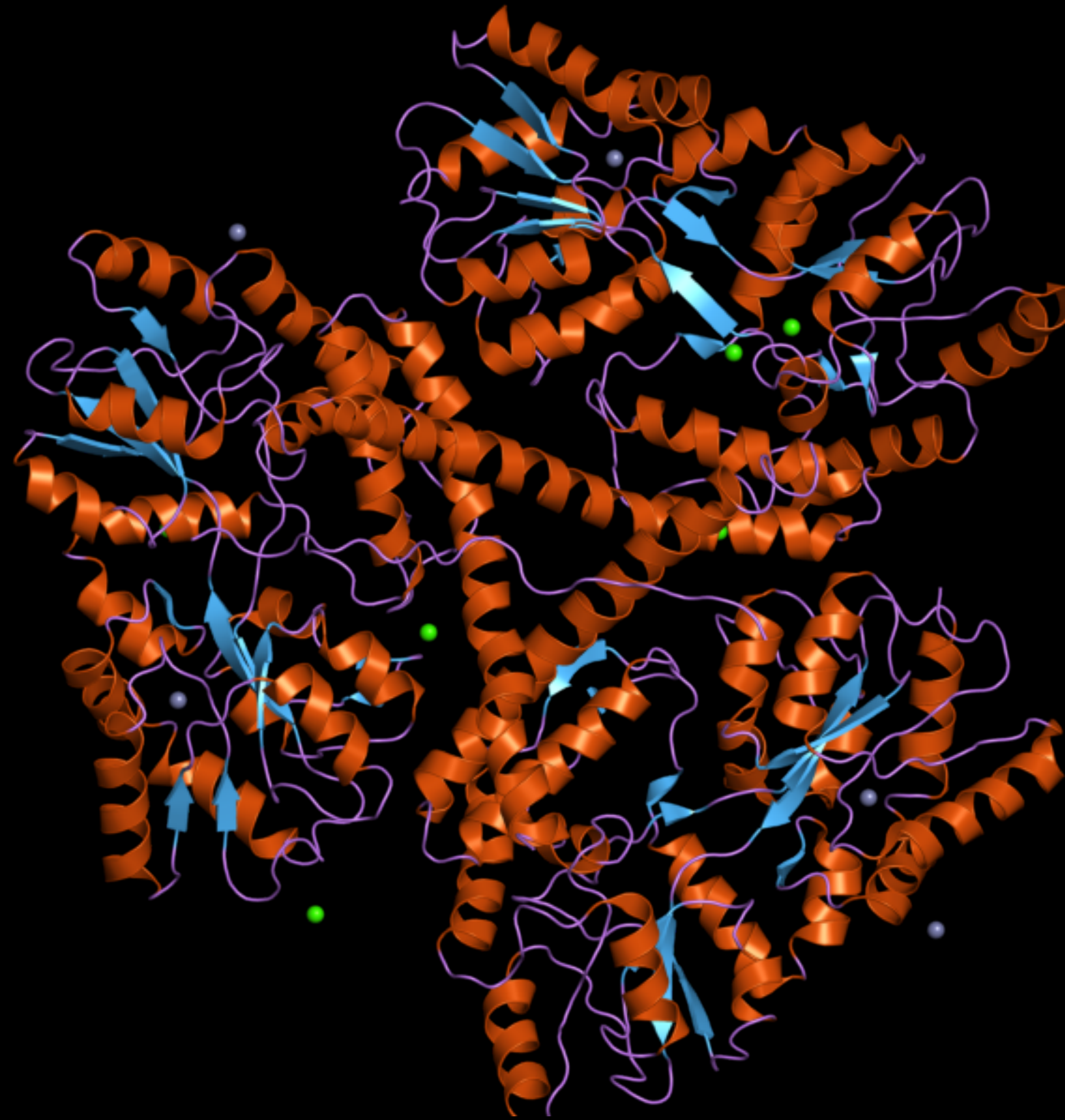


# Chromosome 4





# Huntingtin Protein







1. WHAT IS HD?

2. TESTING FOR HD

3. RESOURCES

[How Is It Done?](#)

[Prenatal Testing](#)

[Deciding To Test](#)

[Alternatives](#)

[Interpreting Results](#)

[Undergo Testing](#)



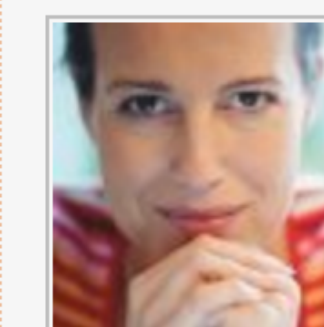
## What is Predictive Testing?

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

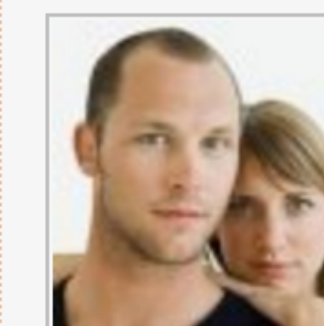


There are many other online websites and resources which provide information regarding HD in general, support

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 Human Genome



**NIH** National Human Genome  
Research Institute

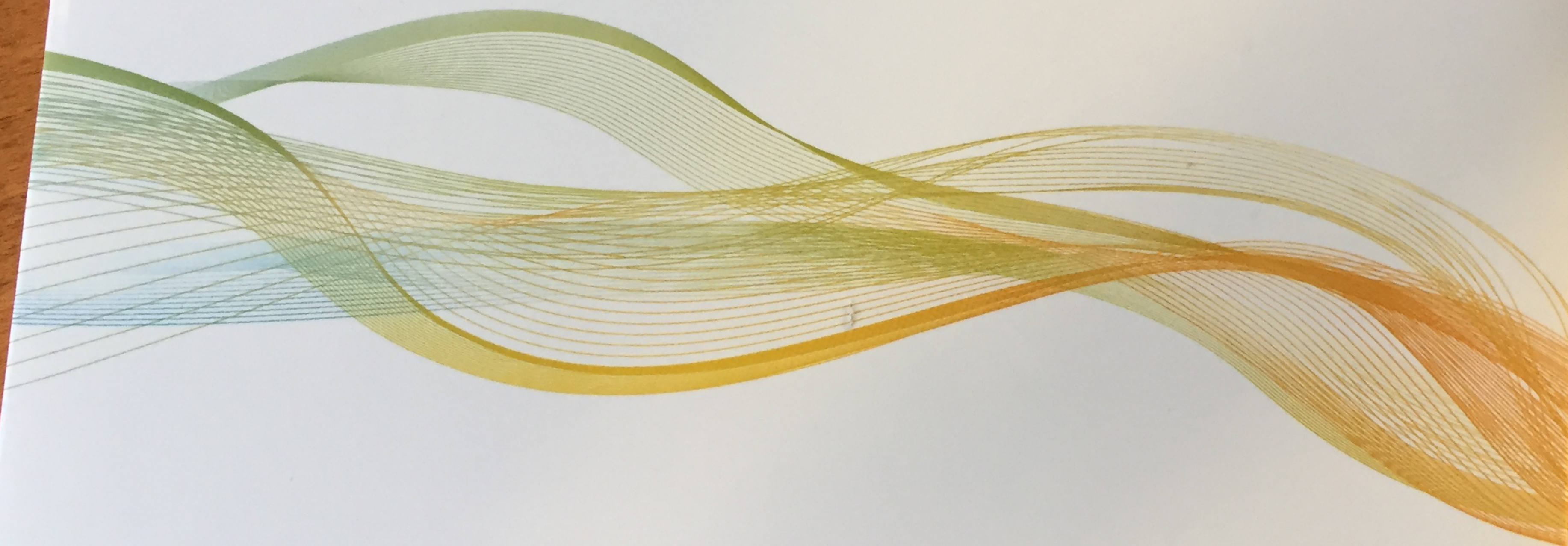
[genome.gov/sequencingcosts](http://genome.gov/sequencingcosts)

*Moore's Law*



UNDERSTAND YOUR GENOME®

IT STARTS WITH YOU



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





## 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.



# Chromosome 1



1 - 249,250,621

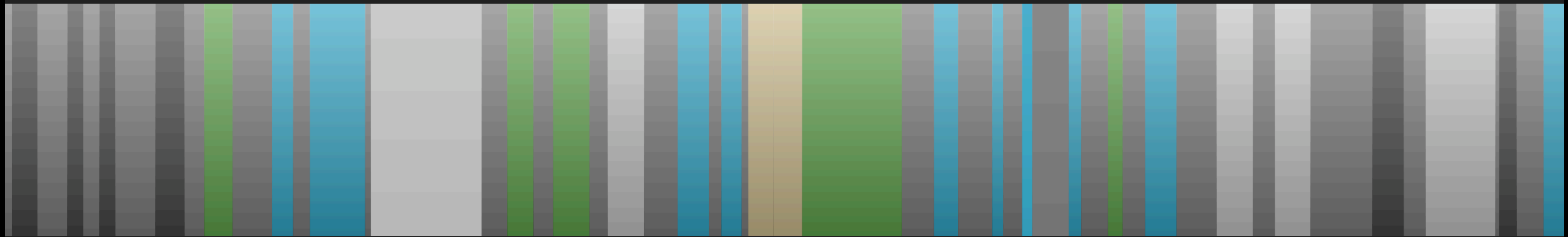


249M

0M 40M 60M 80M 100M 120M 140M 160M 180M 200M 220M

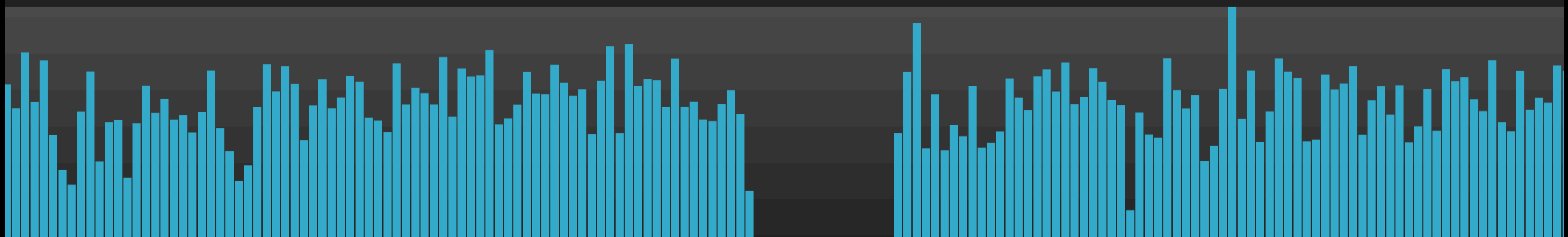
## REFERENCE GENOME

[+ EXPLAIN THIS](#)



## SEQUENCED GENOME

[+ EXPLAIN THIS](#)

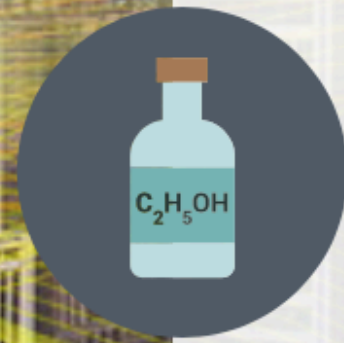




# Here is what we see in your DNA...



**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.**





illumina®

Seagate



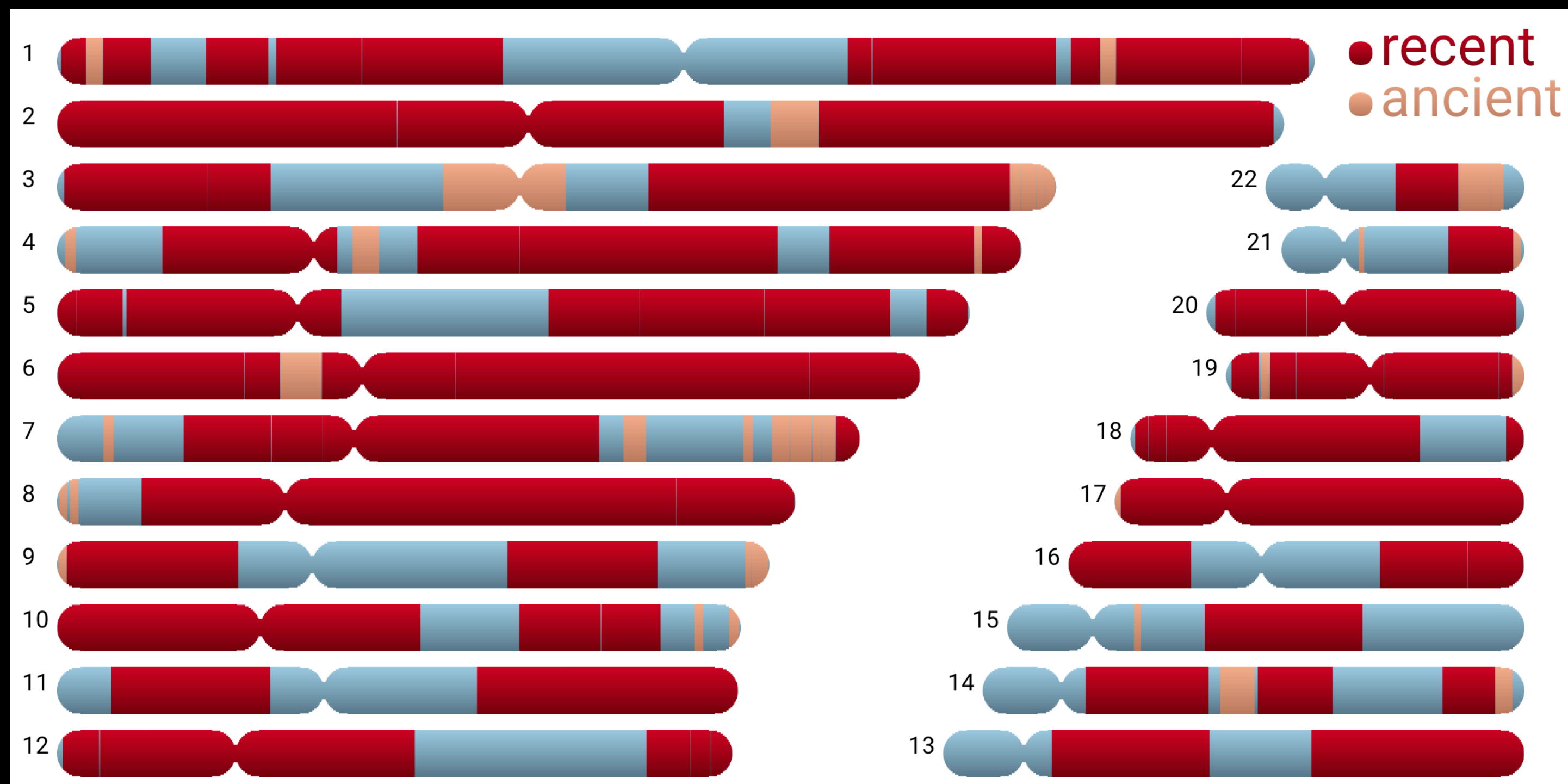
My variant: rs11209026  
Gene: IL23R



Ali Torkamani,  
Scripps Translational Science  
Institute







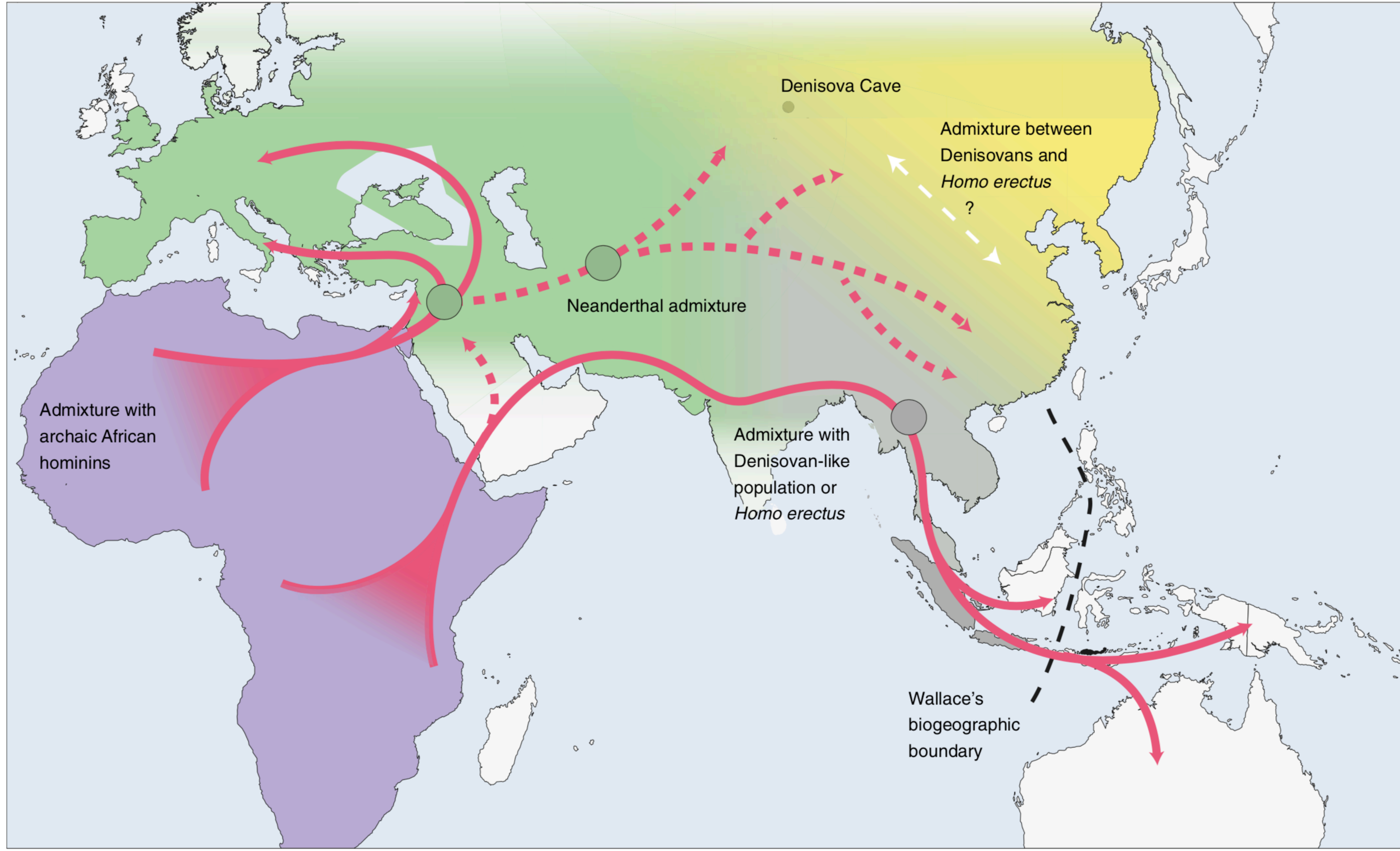
DNA.Land





Image: Viktor Deak

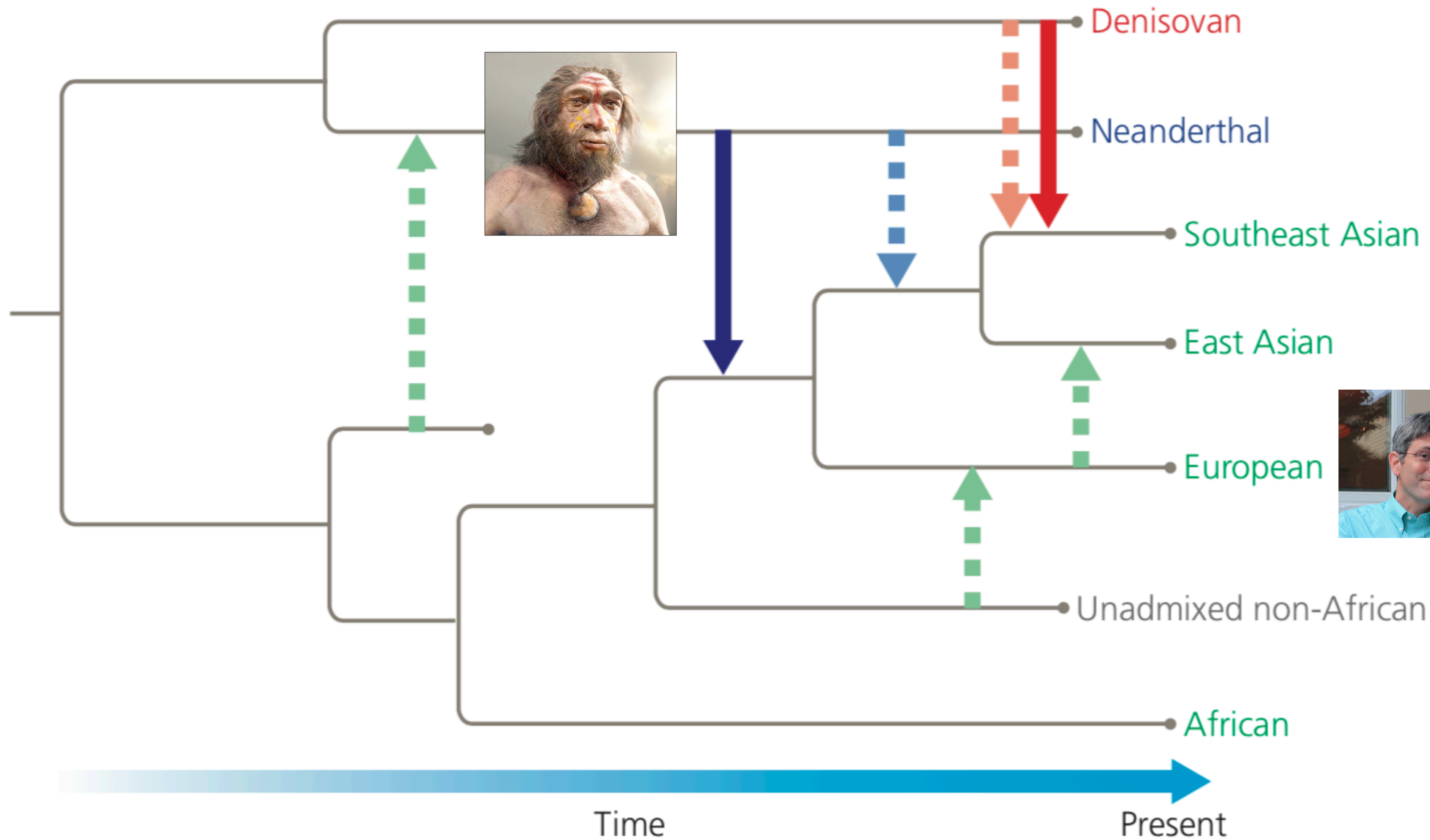




Possible ranges of archaic forms

<span style="display: inline-block; width: 15px; height: 15px; background-color: #90EE90; border: 1px solid black; margin-right: 5px;"></span> Neanderthals	<span style="display: inline-block; width: 15px; height: 15px; background-color: #FFFF00; border: 1px solid black; margin-right: 5px;"></span> Denisovans	<span style="display: inline-block; width: 15px; height: 15px; background-color: #D3D3D3; border: 1px solid black; margin-right: 5px;"></span> <i>Homo erectus</i>	<span style="display: inline-block; width: 15px; height: 15px; background-color: #9370DB; border: 1px solid black; margin-right: 5px;"></span> Archaic African hominins	<span style="display: inline-block; width: 15px; height: 15px; background-color: #000000; border: 1px solid black; margin-right: 5px;"></span> <i>Homo floresiensis</i>	<span style="display: inline-block; width: 15px; height: 15px; background-color: #FF0000; border: 1px solid black; margin-right: 5px;"></span> <i>Homo sapiens</i> dispersals inferred	<span style="display: inline-block; width: 15px; height: 15px; background-color: #FF0000; border: 1px dashed black; margin-right: 5px;"></span> <i>Homo sapiens</i> dispersals speculative
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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



Analysis courtesy of Joshua Akey  
& Selina Vattathil, Princeton

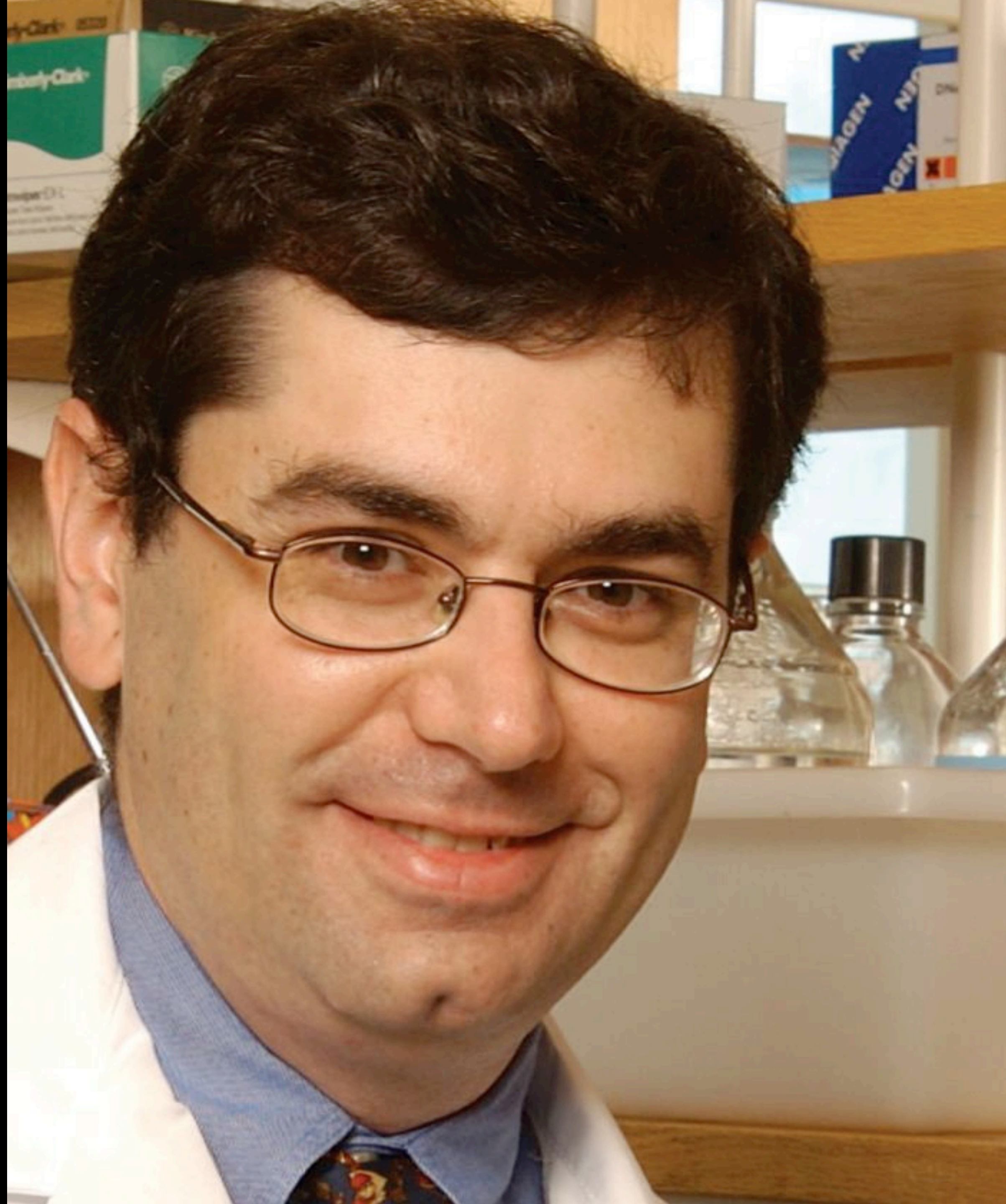


Charles Byrne (1761–1783)

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

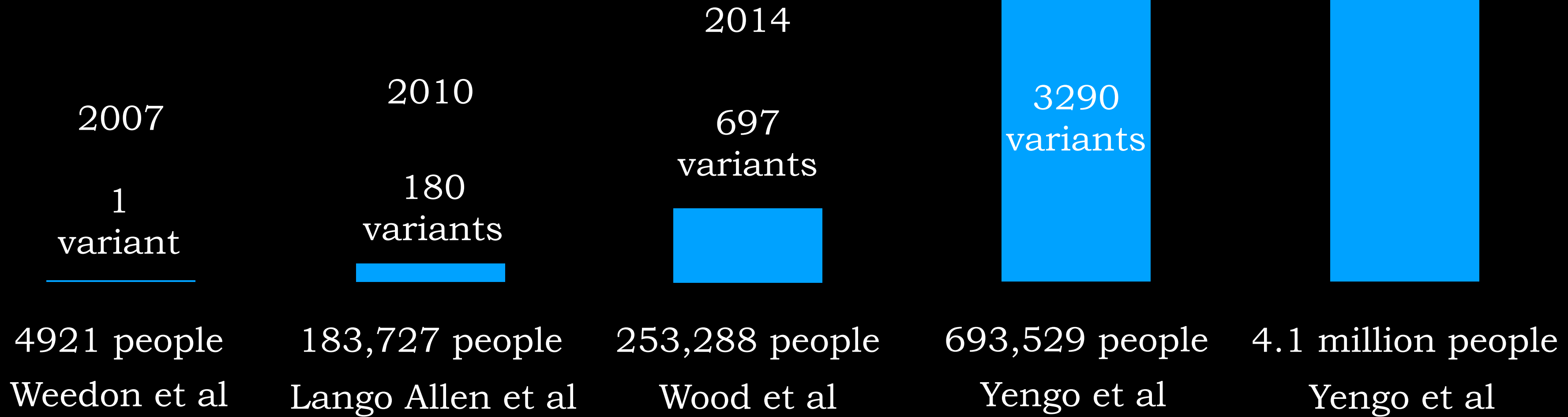






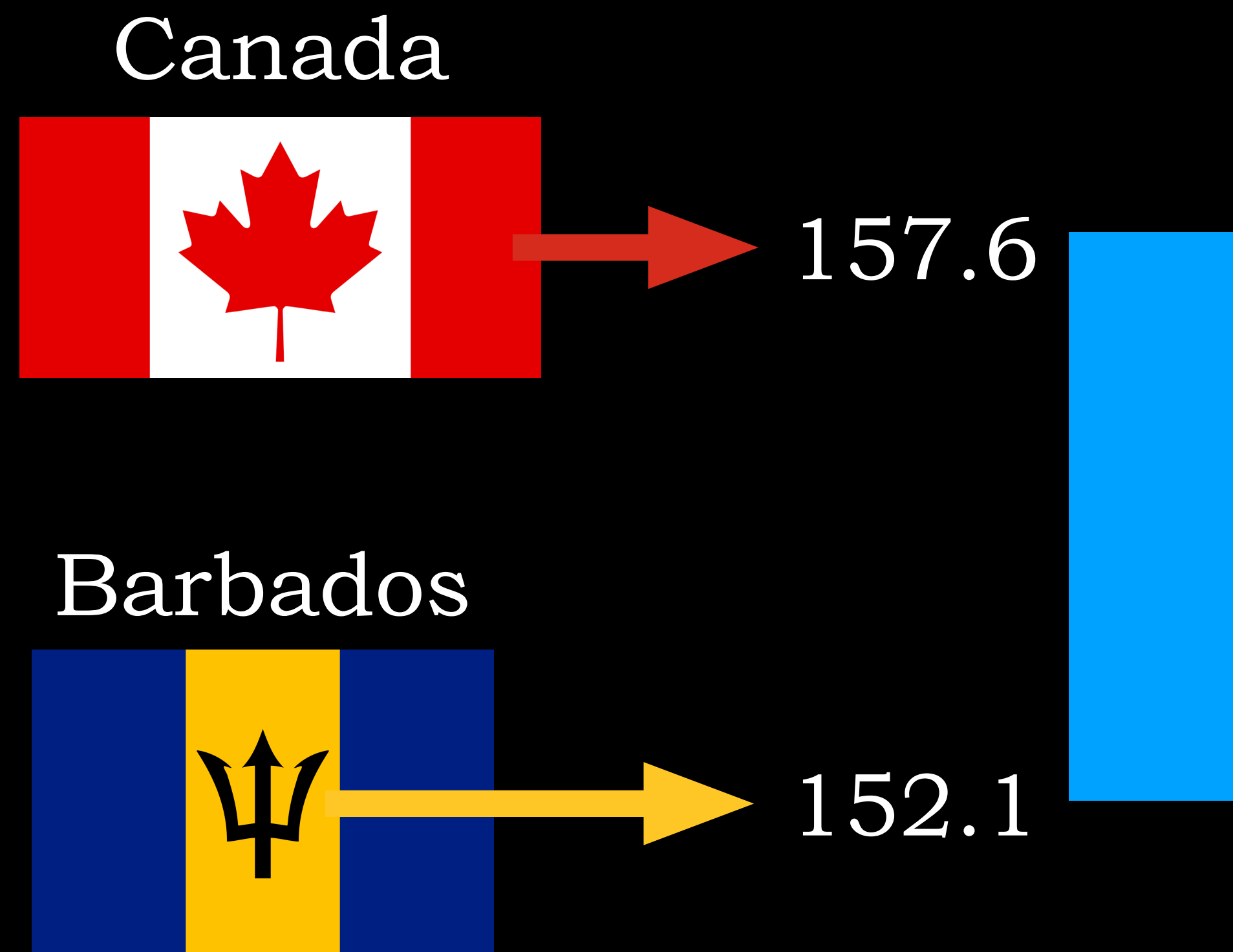
Joel Hirschhorn  
Harvard Medical School





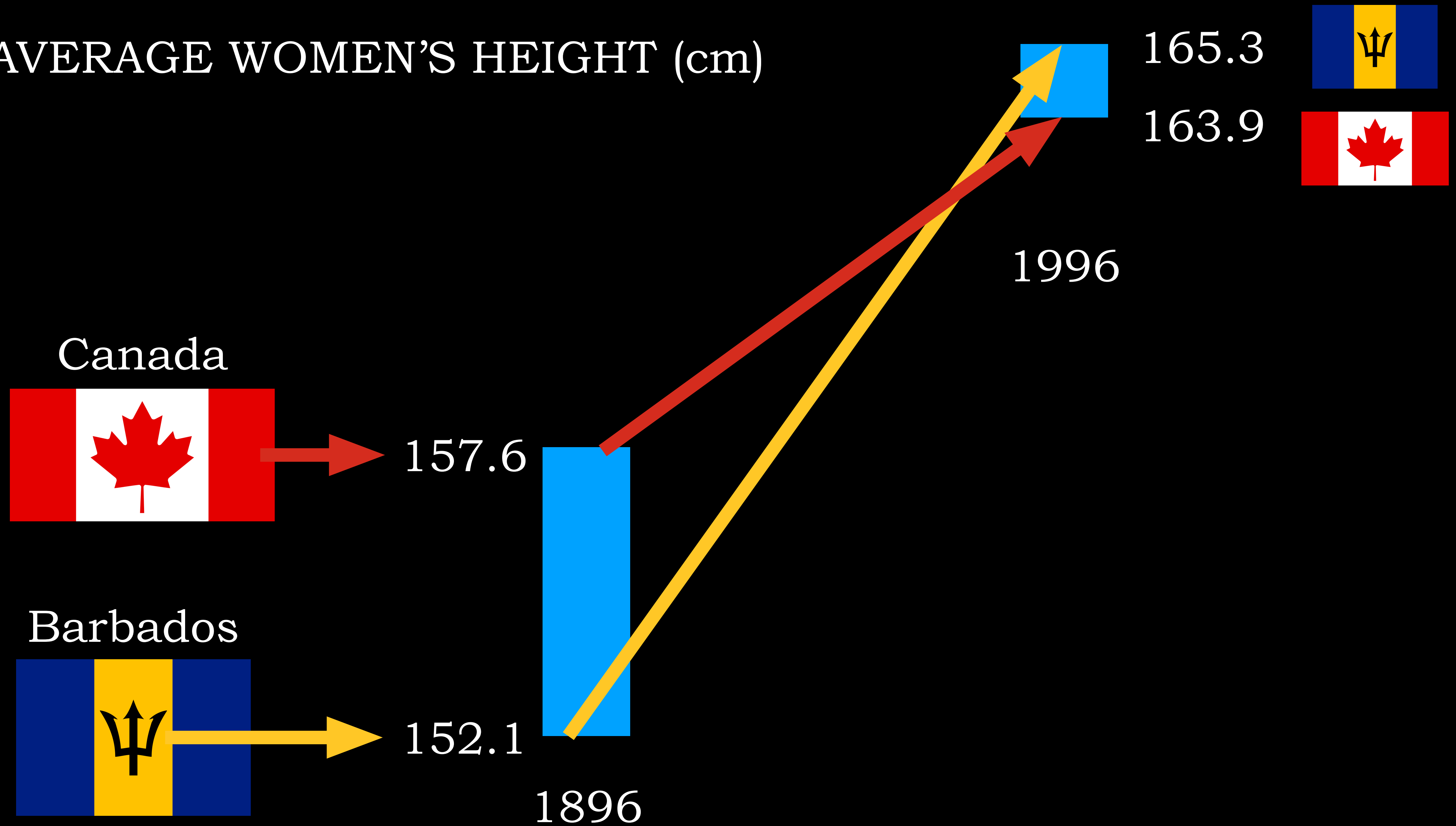


# AVERAGE WOMEN'S HEIGHT (cm)





# AVERAGE WOMEN'S HEIGHT (cm)





CNS TV

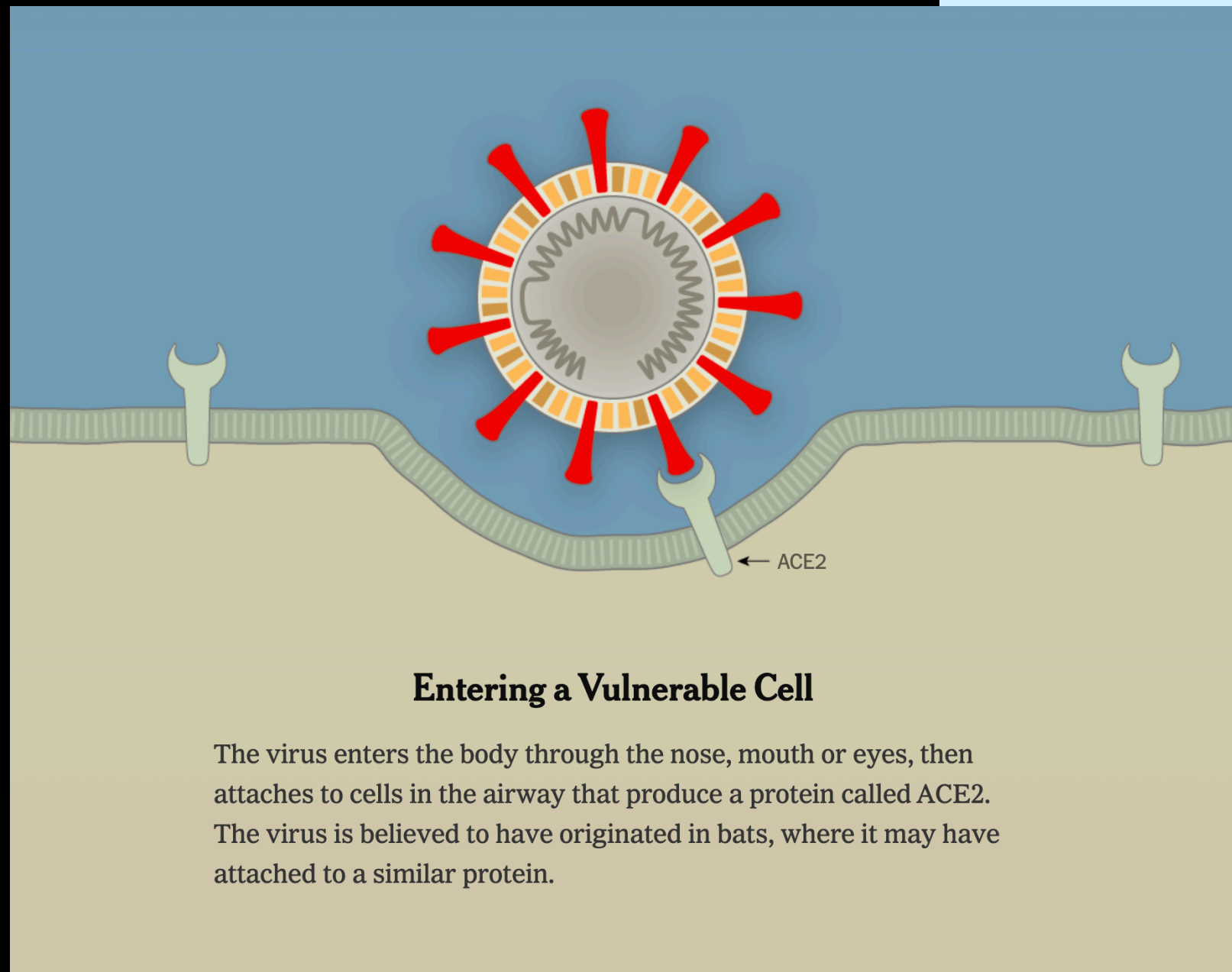
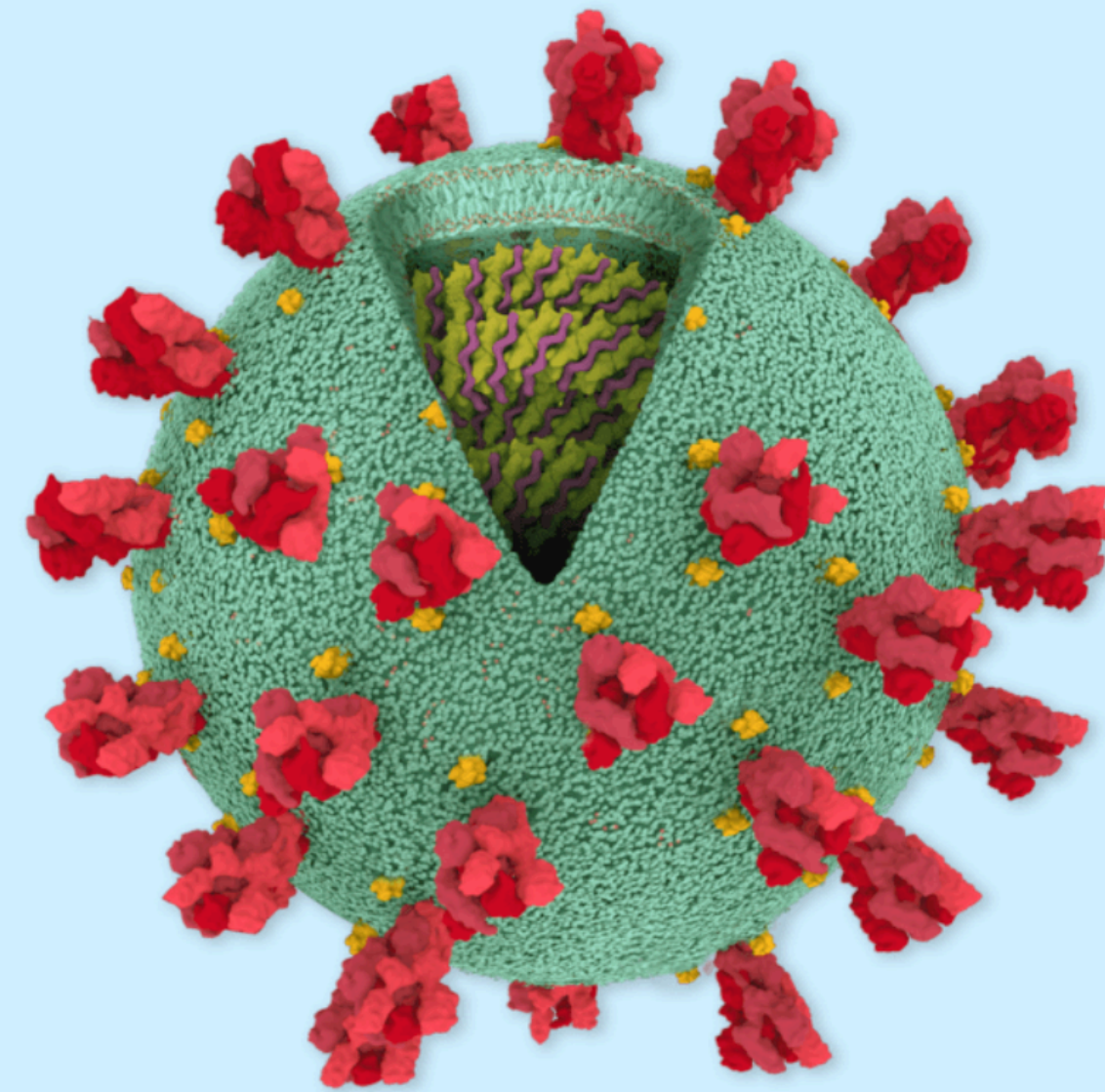
武汉站欢迎您



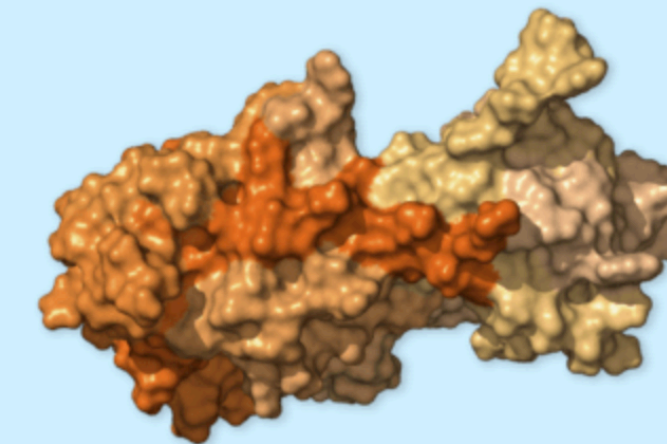


# Bad News Wrapped in Protein: Inside the Coronavirus Genome

By Jonathan Corum and Carl Zimmer April 3, 2020



**Protein Scissors · NSP5**



This protein makes most of the cuts that free other NSP proteins to carry out their own jobs.

<https://www.nytimes.com/interactive/2020/03/11/science/how-coronavirus-hijacks-your-cells.html>

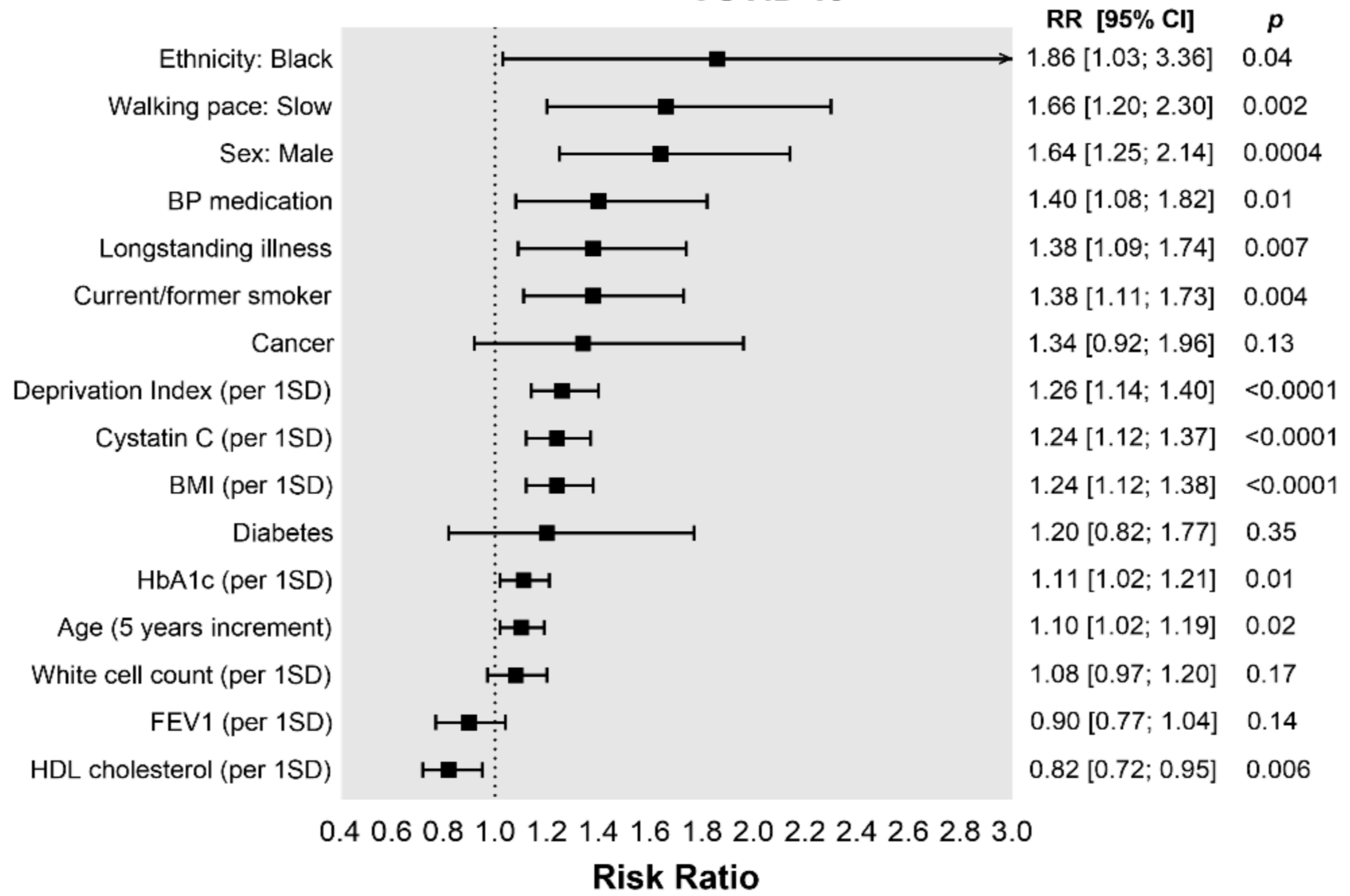
<https://www.nytimes.com/interactive/2020/04/03/science/coronavirus-genome-bad-news-wrapped-in-protein.html>



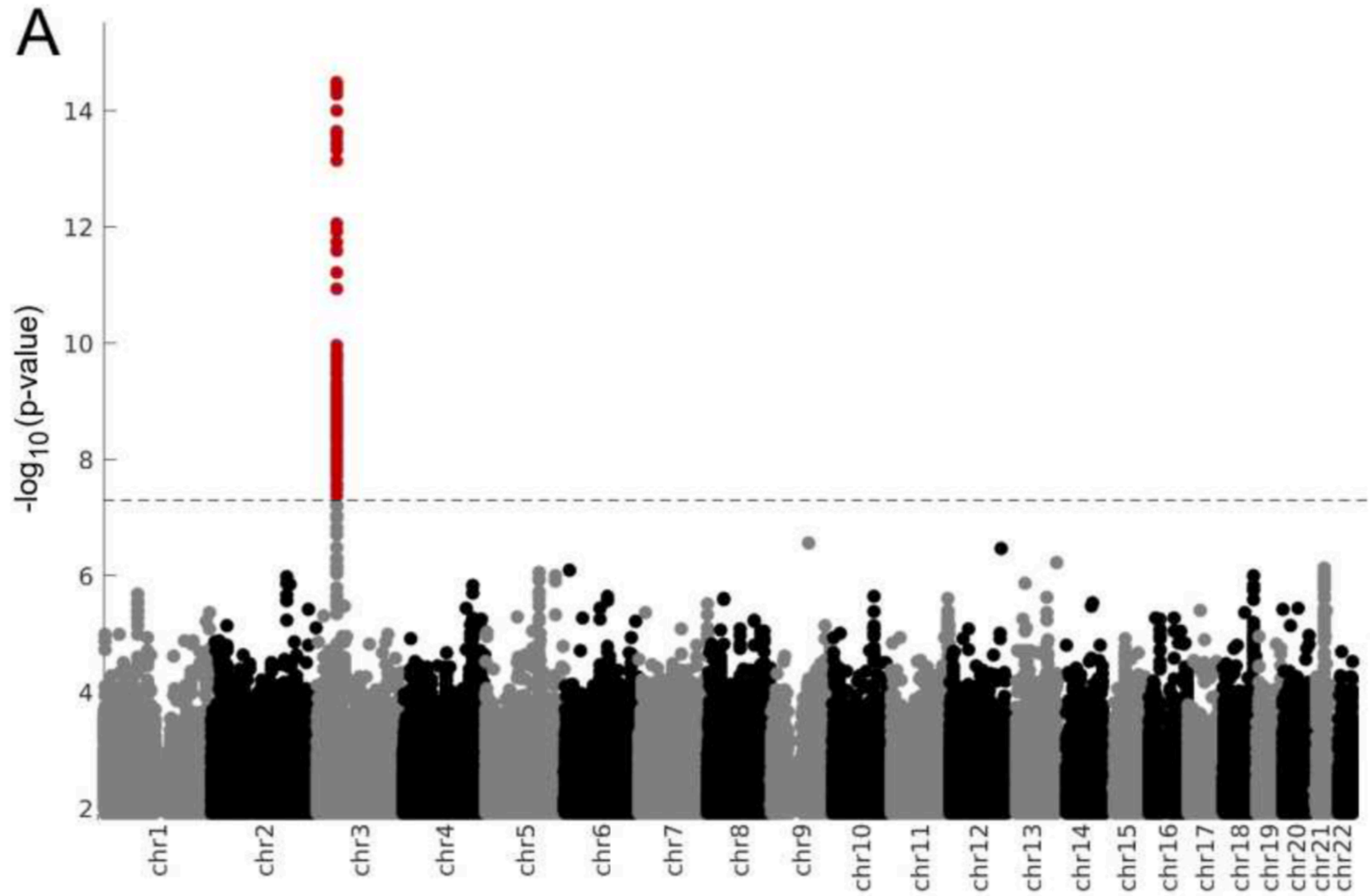




# COVID-19









Article | [Published: 30 September 2020](#)

# The major genetic risk factor for severe COVID-19 is inherited from Neanderthals

[Hugo Zeberg](#)  & [Svante Pääbo](#) 

*Nature* **587**, 610–612(2020) | [Cite this article](#)



# ***DNA Inherited From Neanderthals May Increase Risk of Covid-19***

The stretch of six genes seems to increase the risk of severe illness from the coronavirus.



A researcher excavating a Neanderthal skeleton last year. Gailan Haji/EPA-EFE/REX



By **Carl Zimmer**

Published July 4, 2020 Updated July 8, 2020



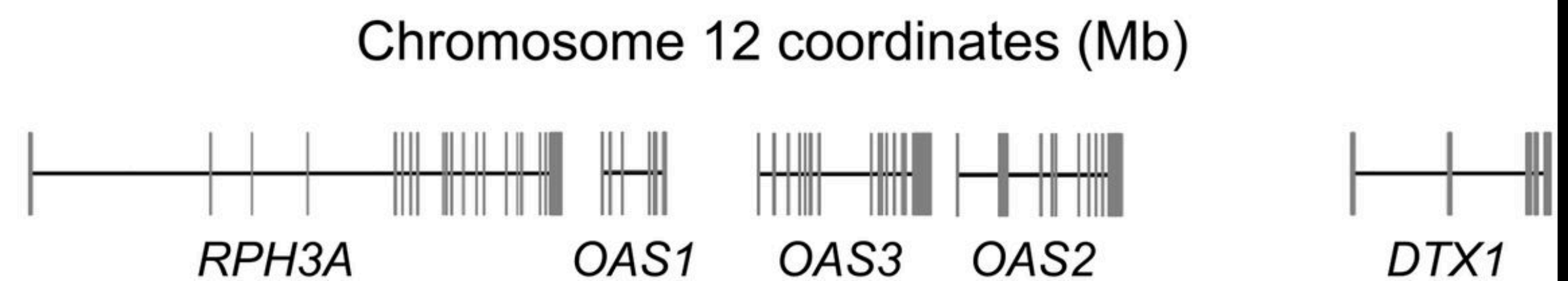
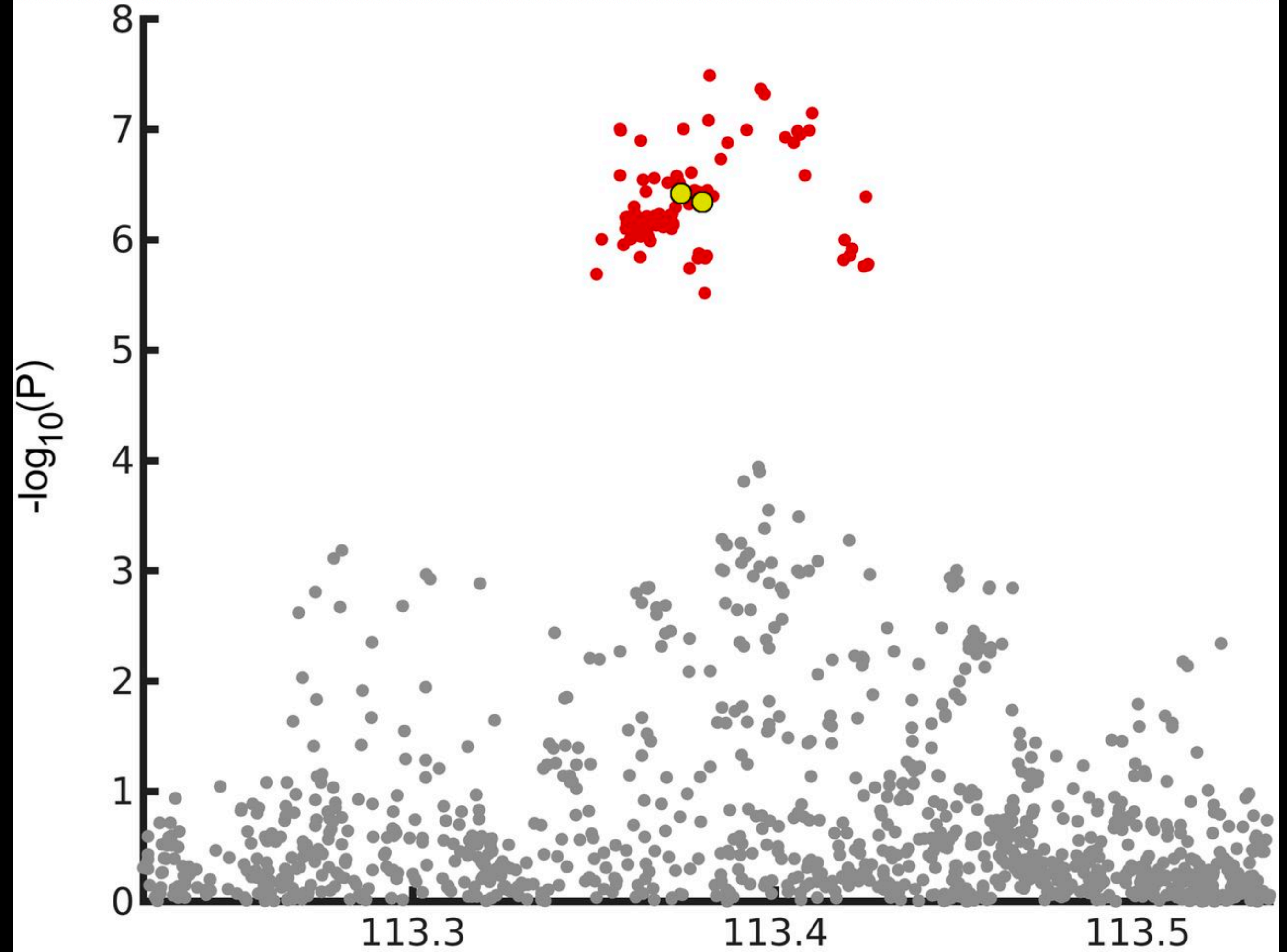
A stretch of DNA [linked to Covid-19](#) was passed down from Neanderthals 60,000 years ago, according to a new study.





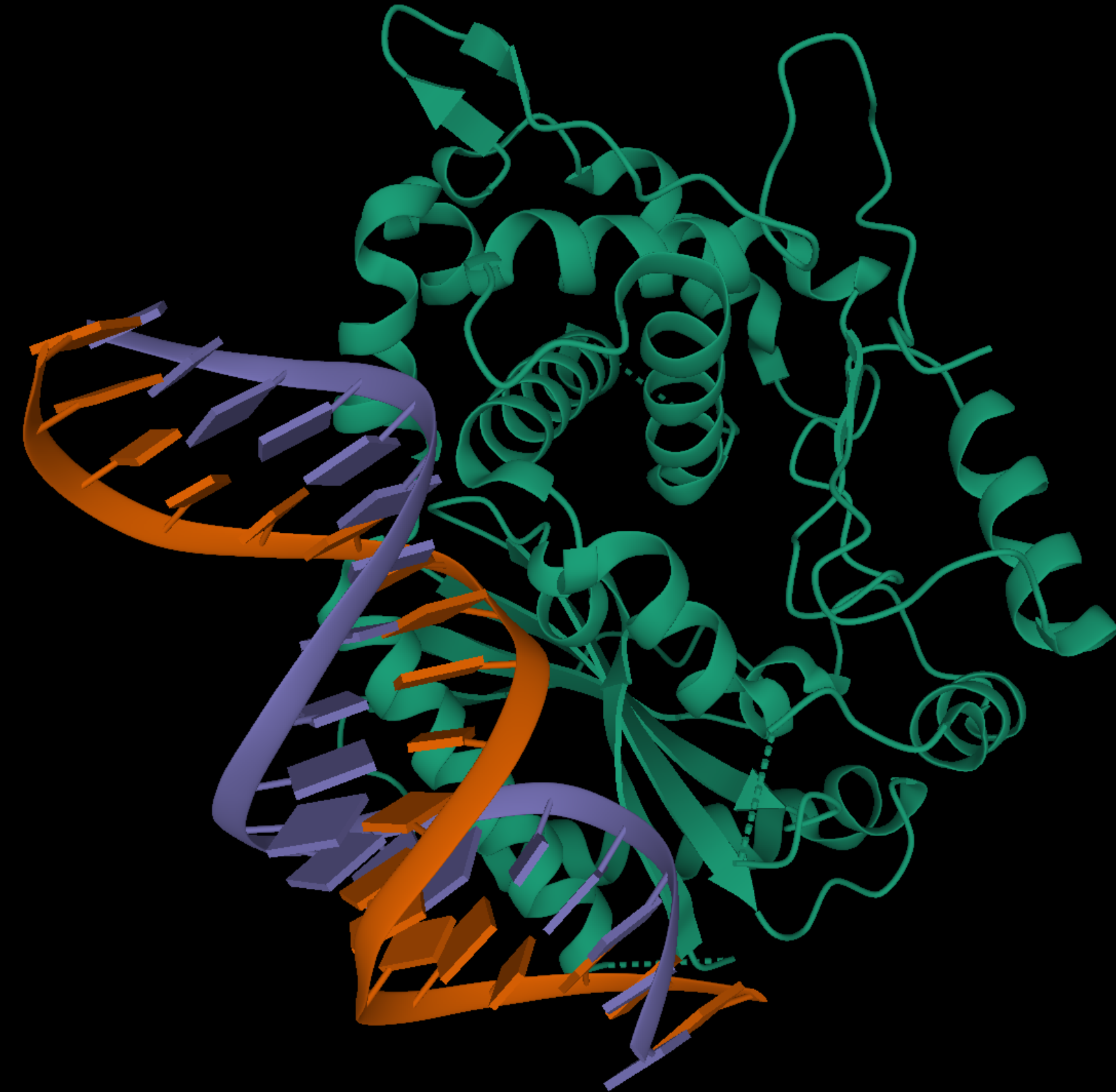
# A genomic region associated with protection against severe COVID-19 is inherited from Neandertals

Hugo Zeberg<sup>a,b,1</sup> and Svante Pääbo<sup>a,c,1</sup>



OAS3 (green)

Double-stranded RNA (orange and purple)



[pnas.org/content/118/9/e2026309118](https://www.pnas.org/content/118/9/e2026309118)  
<https://www.ebi.ac.uk/pdbe/>









Alleles for increased risk on  
chromosome 13: NONE

Alleles for decreased risk on  
chromosome 12: Two  
heterozygous SNPs

On balance, a good  
inheritance?

Courtesy of Jiahao Gao



Thank you  
For more  
information,  
please visit  
[carlzimmer.com](http://carlzimmer.com)

 @carlzimmer

The Powers, Perversions,  
*and Potential of Heredity*



**“Magisterial.”**  
—THE ATLANTIC

**“Extraordinary.”**  
—THE NEW YORK TIMES

**“Engrossing.”**  
—WIRED

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“SCIENCE BOOK OF THE YEAR.” —THE GUARDIAN