

```
bash          bash          samtools
C6WWDACXX:1:1101:405627:0    1123  chrM  1    60    2598M =    246    342    TGGATCACAGCTGTATGACCTATTAG
C6WWDACXX:1:1101:1460565:0    1123  chrM  1    60    37563M =    204    302    TAGCCCAACGGTTCCTTAAATAAG
C6WWDACXX:1:1101:1507057:0    81    chrM  1    60    4596M =    16298    16396    GATGGATCAGGGTGTATGACCTATT
C6WWDACXX:1:1102:34162:0    163    chrM  1    60    27573M =    169    267    GTTCCCTTAAATAGGCAACGATG
C6WWDACXX:1:1102:963540:0    161    chrM  1    3    48552M =    53    130    TAAAGCCTAAATAGGCAACGATG
C6WWDACXX:1:1103:141030:0    1187  chrM  1    60    33567M =    274    371    TTAGGGGCATAGGCECAATAGCC
C6WWDACXX:1:1103:213161:0    1123  chrM  1    60    4596M =    178    276    GATGGATCAGGGTGTATGACCTATT
1187  chrM  1    60    242    337    GCTAAAGTGAAGTGTATGACCTATT
```

# Welcome to The Age of Personal Genomes

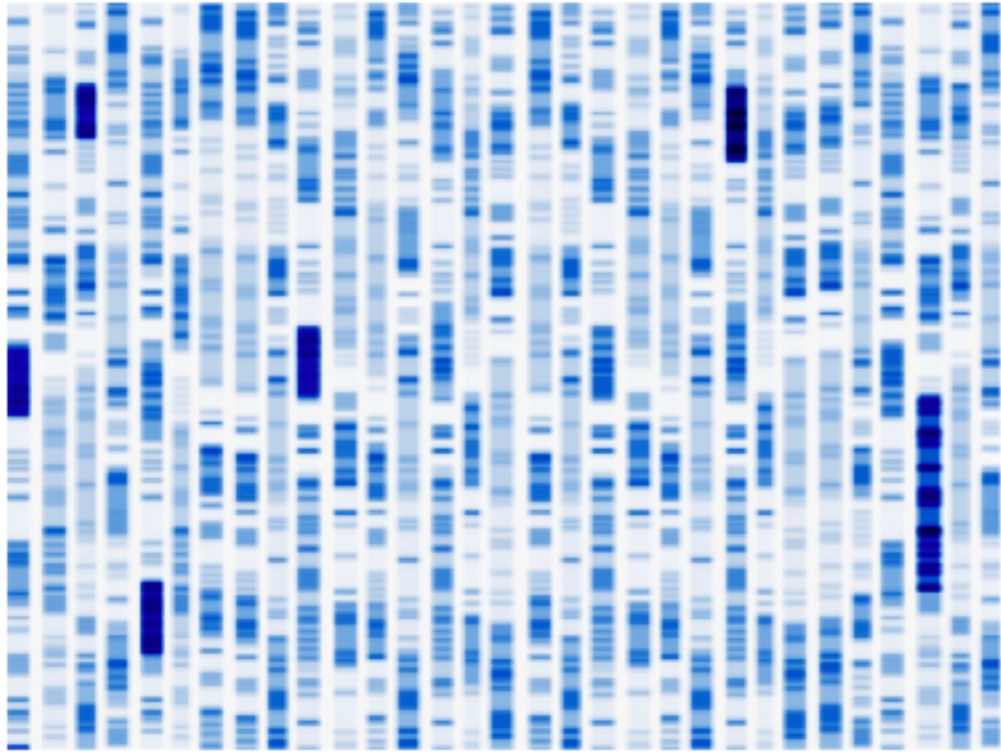
```
C6WWDACXX:1:1103:1512941:0    1187  chrM  1    60    14585M =    138    236    AGTTCAAGGCTGTATGACCTATT
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C6WWDACXX:1:1104:126593:0    1187  chrM  1    60    8592M =    107    205    GATTCCTAGTTCAGGCTGTATGAC
C6WWDACXX:1:1104:156772:0    1187  chrM  1    60    67533M =    129    227    TCCCTCACTCAGGCTGTATGAC
C6WWDACXX:1:1104:1188127:0    99    chrM  1    60    65535M =    128    226    AGTTCCTTAAATAGGCAACGATG
C6WWDACXX:1:1104:1910836:0    1187  chrM  1    60    28572M =    122    220    AGCCTAANTAGCCCAAGCTTCCTT
C6WWDACXX:1:1105:44874:0    1123  chrM  1    60    45555M =    100    198    CCTAAATAGCCCAAGCTTCCTTAA
C6WWDACXX:1:1105:343348:0    1187  chrM  1    60    43557M =    151    249    TAAAGCCTAAATAGGCAACGATG
C6WWDACXX:1:1105:528291:0    163    chrM  1    60    48552M =    248    345    EFCATAGGCTGTATGACCTATT
C6WWDACXX:1:1106:816521:0    163    chrM  1    60    52548M =    195    293    GATGGATCAGGGTGTATGACCTATT
C6WWDACXX:1:1106:858303:0    1187  chrM  1    60    4596M =    164    263    AAGTGAAGCTGTATGACCTATT
C6WWDACXX:1:1106:1498035:0    1123  chrM  1    60    90510M =    119    217    GATGGATCAGGGTGTATGACCTATT
C6WWDACXX:1:1106:1632446:0    163    chrM  1    60    4596M =    96    194    GGTAAAGTGAAGCTGTATGACCTATT
C6WWDACXX:1:1106:1960666:0    163    chrM  1    60    9357M =    141    239    AAAGTGAAGCTGTATGACCTATT
C6WWDACXX:1:1106:2132852:0    1123  chrM  1    60    90510M =    =    294    391    AAATAAGACATCAGGATGATGATG
C6WWDACXX:1:1107:197348:0    163    chrM  1    60    42557M1S =    199    297    GTTCCCTTAAATAGGCAACGATG
C6WWDACXX:1:1107:498216:0    1123  chrM  1    60    18582M =    172    270    TAAGACATCAGGATGATGATGATG
C6WWDACXX:1:1107:635534:0    1187  chrM  1    60    27573M =    =    382    480    GCCCACAGTTCCTTAAATAGGCAAC
C6WWDACXX:1:1107:970105:0    1123  chrM  1    60    15584M1S =    214    312    GGCATAAAGCCTAAATAGGCAACG
C6WWDACXX:1:1107:1075833:0    1123  chrM  1    60    35565M =    188    286    GTTCCCTTAAATAGGCAACGATG
C6WWDACXX:1:1107:1084924:0    1123  chrM  1    60    21579M =    129    227    TCCAGGCCATAAAGCCTAAATAGG
C6WWDACXX:1:1107:1214159:0    163    chrM  1    60    53547M =    71    169    AAGCCTAAATAGGCAACGATG
C6WWDACXX:1:1107:2132194:0    99    chrM  1    60    58542M =    172    270    CTAAAGTGAAGCTGTATGACCTATT
C6WWDACXX:1:1108:702977:0    163    chrM  1    60    46554M =    279    376    AAAGTGAAGCTGTATGACCTATT
C6WWDACXX:1:1108:970307:0    99    chrM  1    60    46554M =    93    191    AAAGTGAAGCTGTATGACCTATT
C6WWDACXX:1:1108:1229805:0    1187  chrM  1    60    9258M =    95    193    85879263
C6WWDACXX:1:1108:1590115:0    99    chrM  1    60    90510M =    85879165    204    TGTATCCGACATCTGTTCTACTTC
C6WWDACXX:1:1108:1741008:0    163    chrM  1    10    81519M =    106    204    TGTATCCGACATCTGTTCTACTTC
C6WWDACXX:1:1108:2227440:0    99    chrM  1    60    81519M =    106    204    TGTATCCGACATCTGTTCTACTTC
C6WWDACXX:1:1108:2313740:0    161    chrM  1    60    81519M =    106    204    TGTATCCGACATCTGTTCTACTTC
C6WWDACXX:1:1109:223979:0    1123  chrM  1    60    81519M =    106    204    TGTATCCGACATCTGTTCTACTTC
C6WWDACXX:1:1109:546465:0
C6WWDACXX:1:1109:586698:0
```

Carl Zimmer  
February 4, 2019, Yale MBB 452

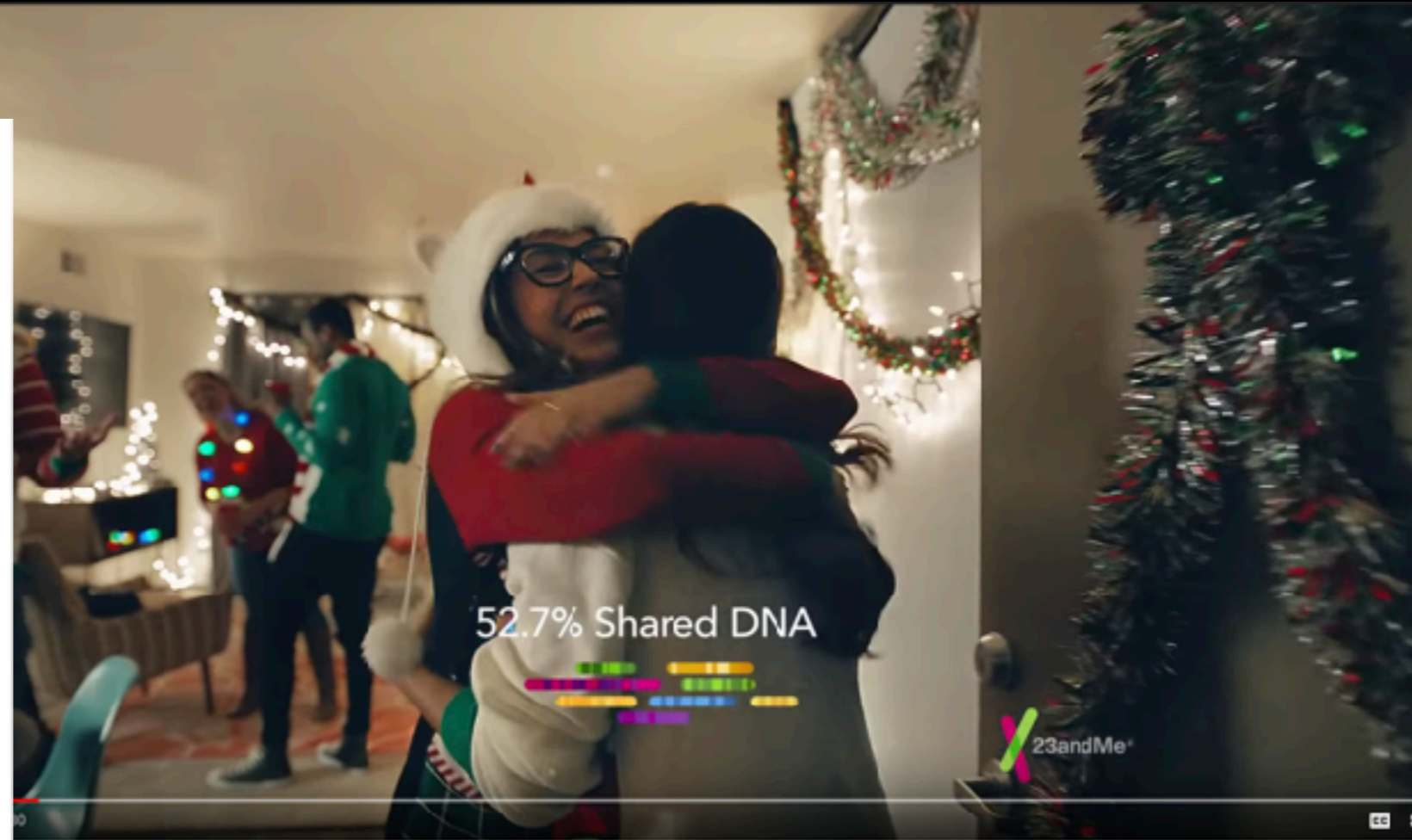


MEGAN MOLTENI SCIENCE 12.01.17 07:00 AM

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



ALFRED PASIEKA/SCIENCE PHOTO LIBRARY/GETTY IMAGES



For Everyone You Love!

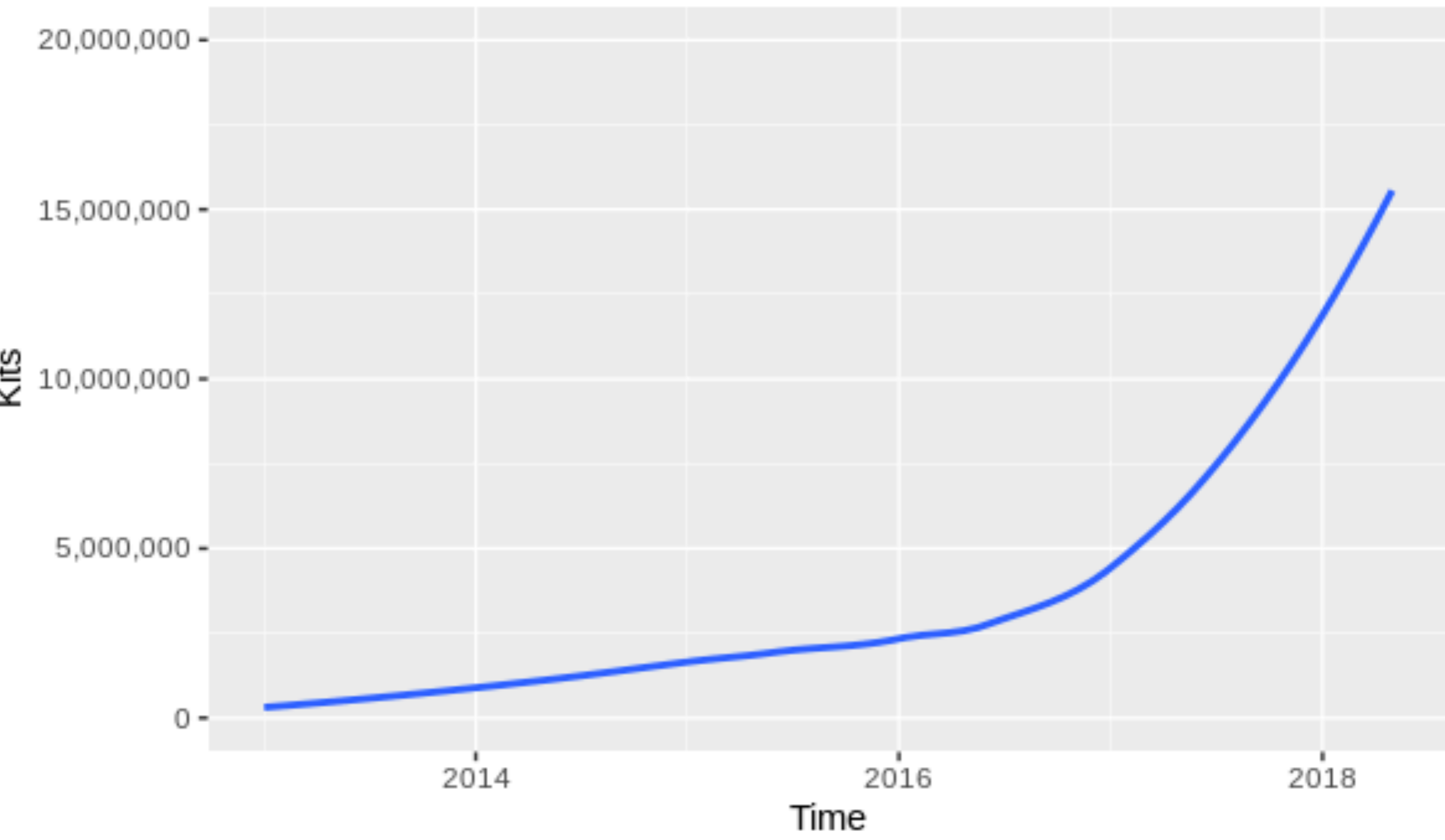
Up next



[ispot.tv/ad/wHZ4/23andme-one-body-one-mind](https://ispot.tv/ad/wHZ4/23andme-one-body-one-mind)

[wired.com/story/ancestrys-genetic-testing-kits-are-heading-for-your-stockings-this-year/](https://wired.com/story/ancestrys-genetic-testing-kits-are-heading-for-your-stockings-this-year/)

# Kits of all major companies

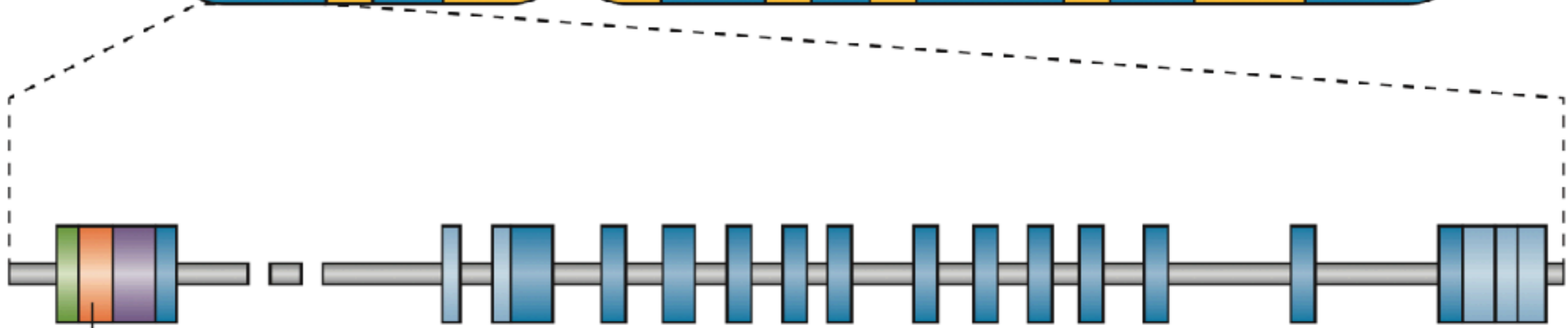


# Woody Guthrie





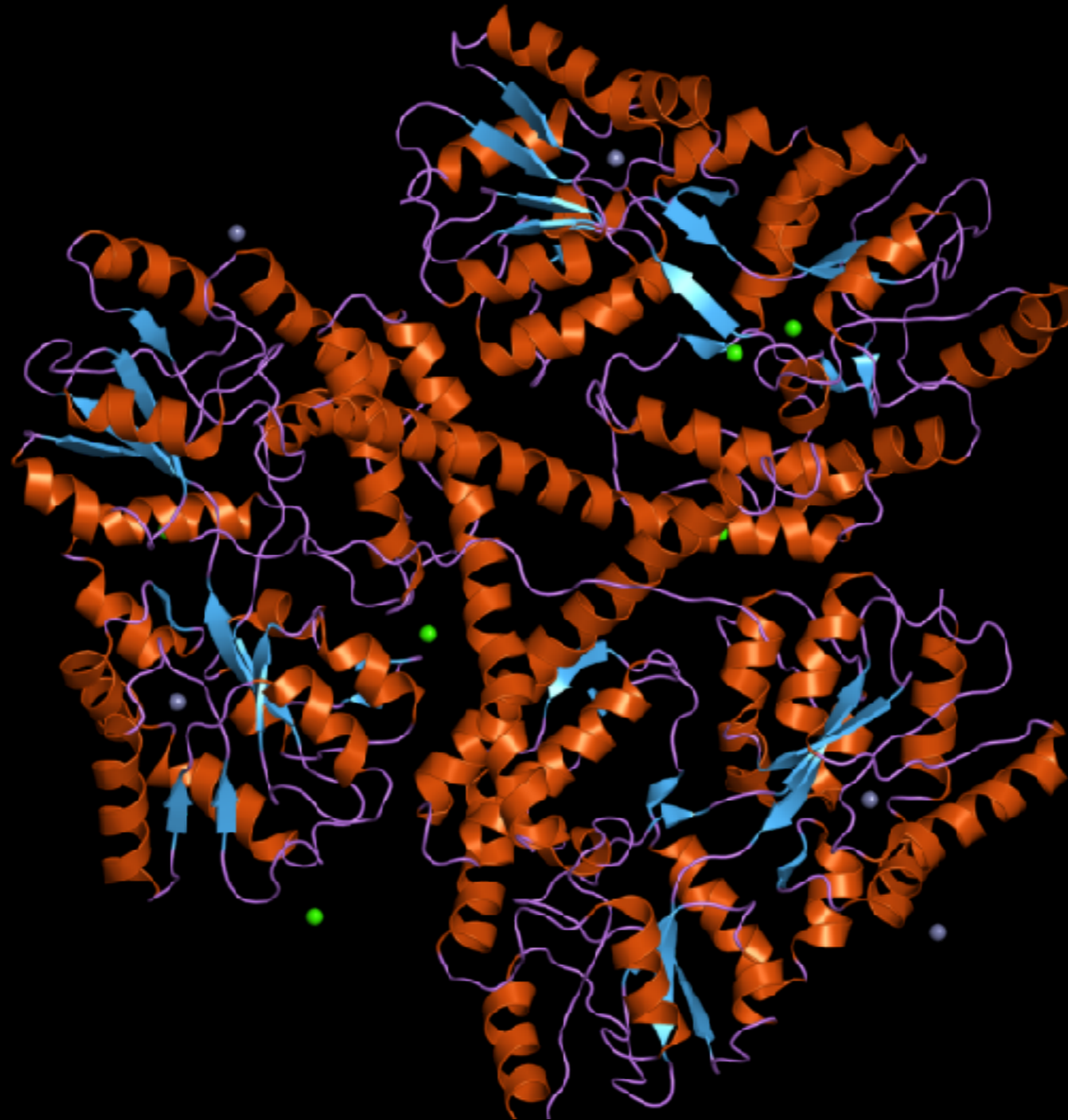
# Chromosome 4



CAG repeat

*HTT* gene

# Huntingtin

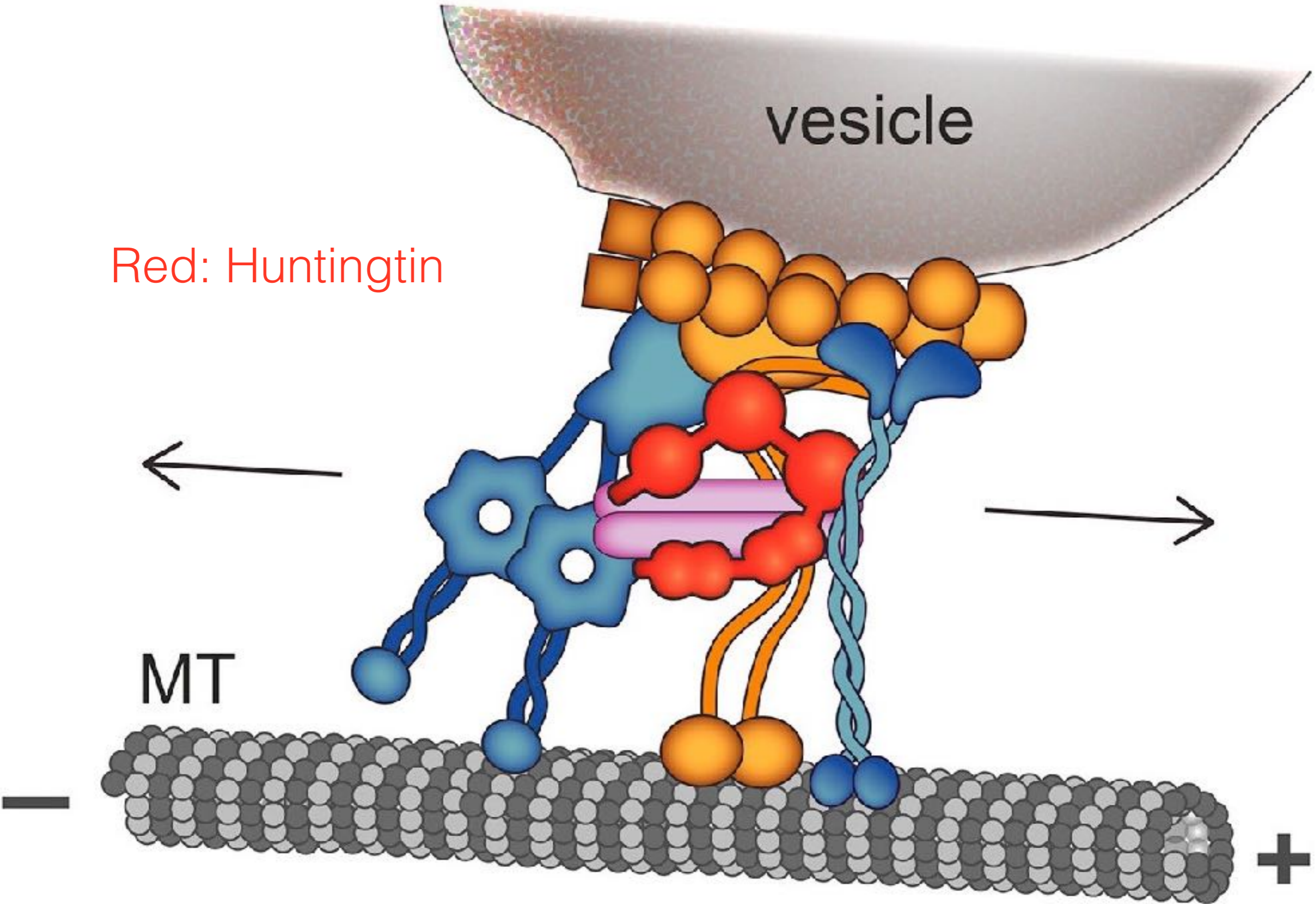




Red: Huntingtin

vesicle

MT





1. WHAT IS HD?

2. TESTING FOR HD

3. RESOURCES

[How Is It Done?](#)

[Prenatal Testing](#)

[Deciding To Test](#)

[Alternatives](#)

[Interpreting Results](#)

[Undergo Testing](#)



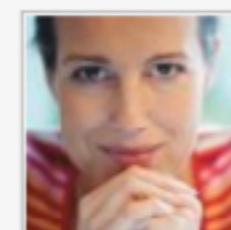
## What is Predictive Testing?

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

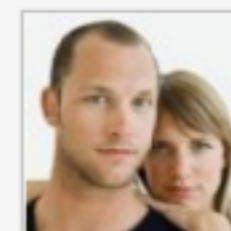
## HD Resources



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](#)

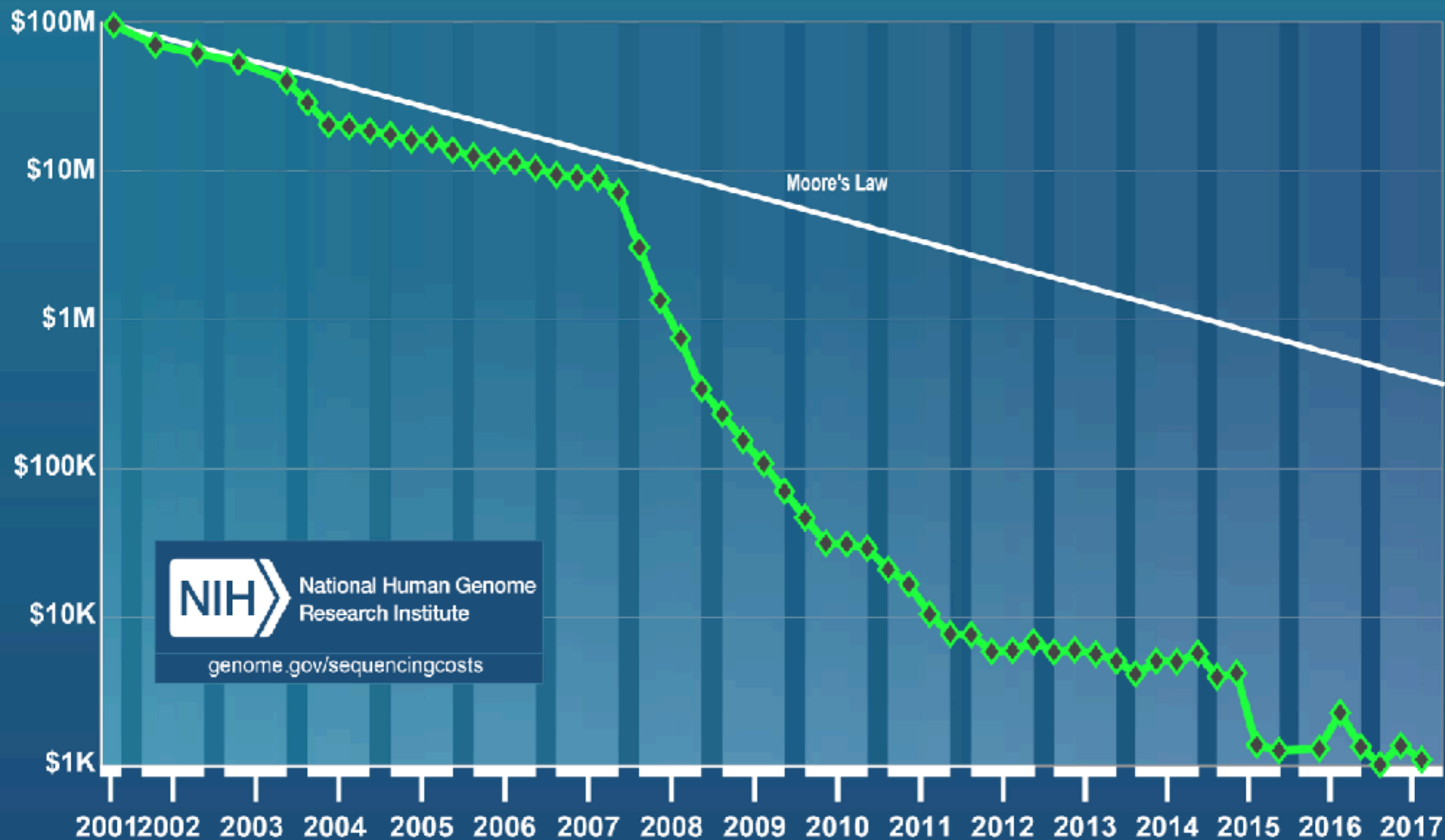
## Our Stories



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



CARL ZIMMER



The Powers, Perversions,  
*and* Potential of Heredity



UNDERSTAND YOUR GENOME®

IT STARTS WITH YOU



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



## **TruGenome Predisposition Screen**

### **Clinical Report**





## **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.



## TruGenome Predisposition Screen Clinical Report

**No pathogenic or likely pathogenic variants** were found in the 1,691 genes evaluated that are expected to be associated with the disease. 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.





**TruGenome Predisposition Screen**  
**Clinical Report**

**Findings Regarding Carrier Status**

<b>Variant</b>	<b>Interpretation</b>	<b>Associated Condition</b>	<b>Mode of Inheritance</b>	<b>Zygoty</b>
<b>MEFV</b> c.2177T>C (p.Val726Ala)	Pathogenic	Familial Mediterranean Fever	Autosomal Recessive	Heterozygous
<b>MBL2</b> c.154C>T (p.Arg52Cys)	Likely Pathogenic	Mannose-Binding Protein Deficiency	Autosomal Recessive	Heterozygous

# Chromosome 1



1 - 249,250,621



+ 249M



## 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.**



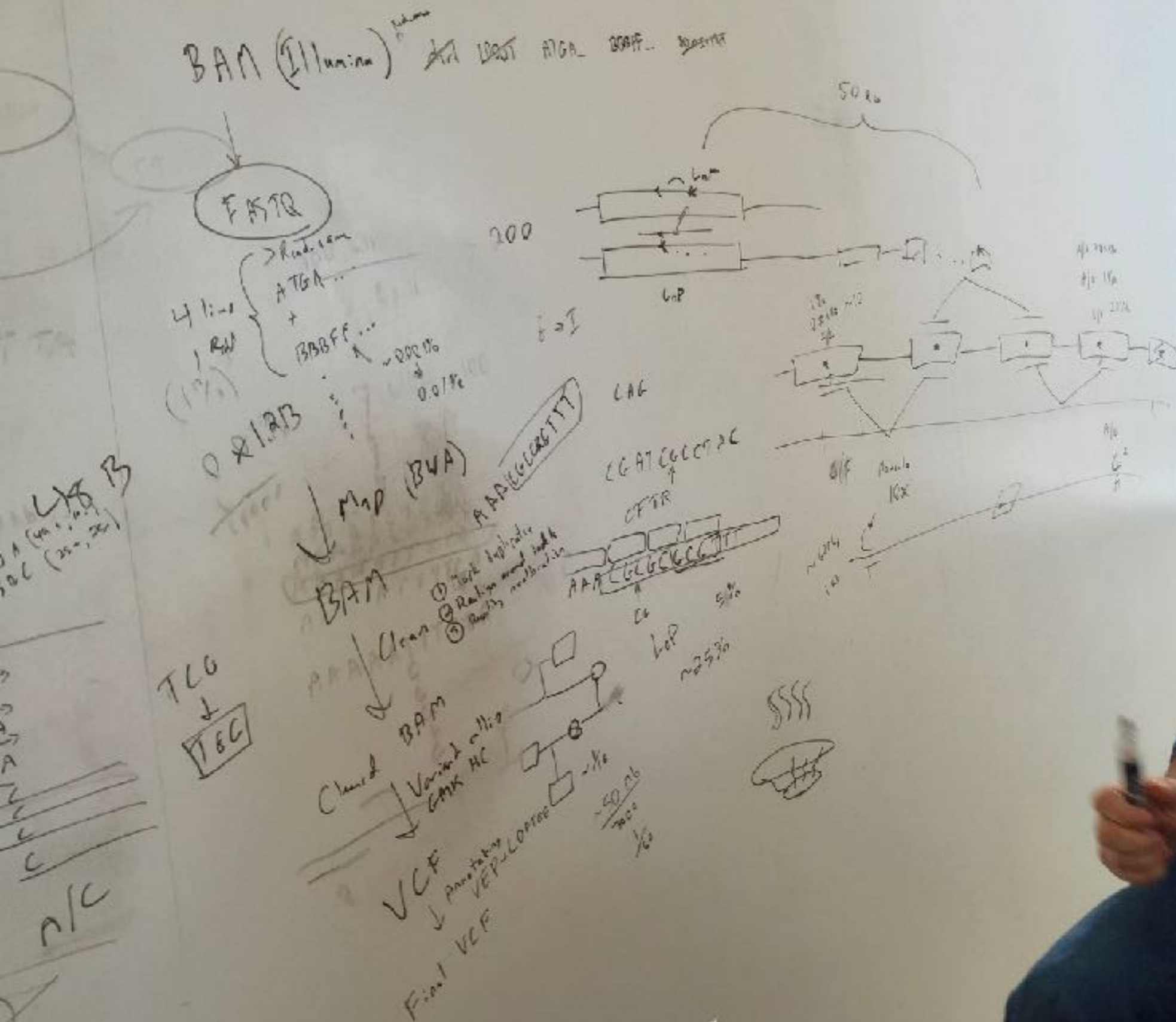








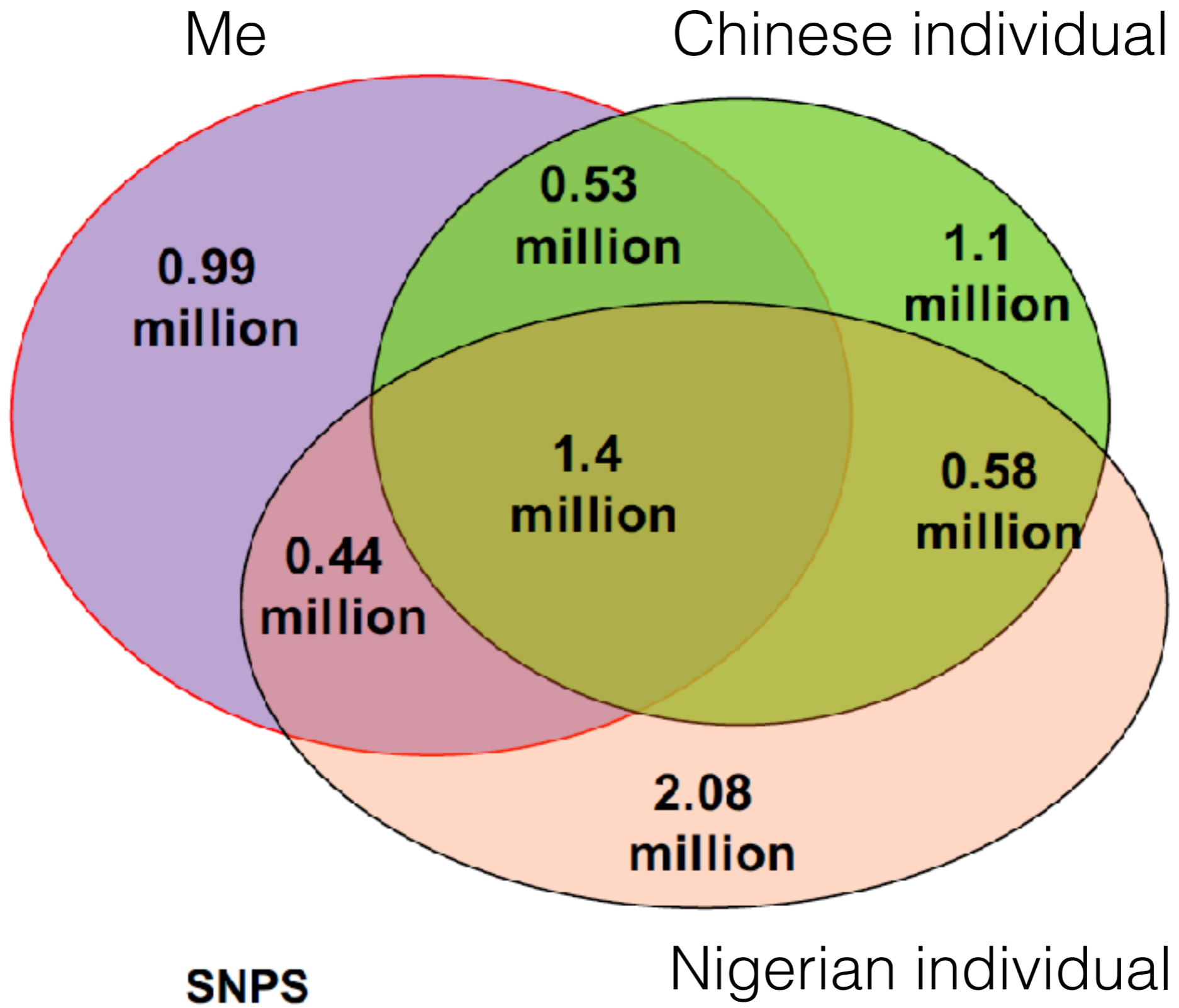
# Konrad Karczewski, Broad Institute





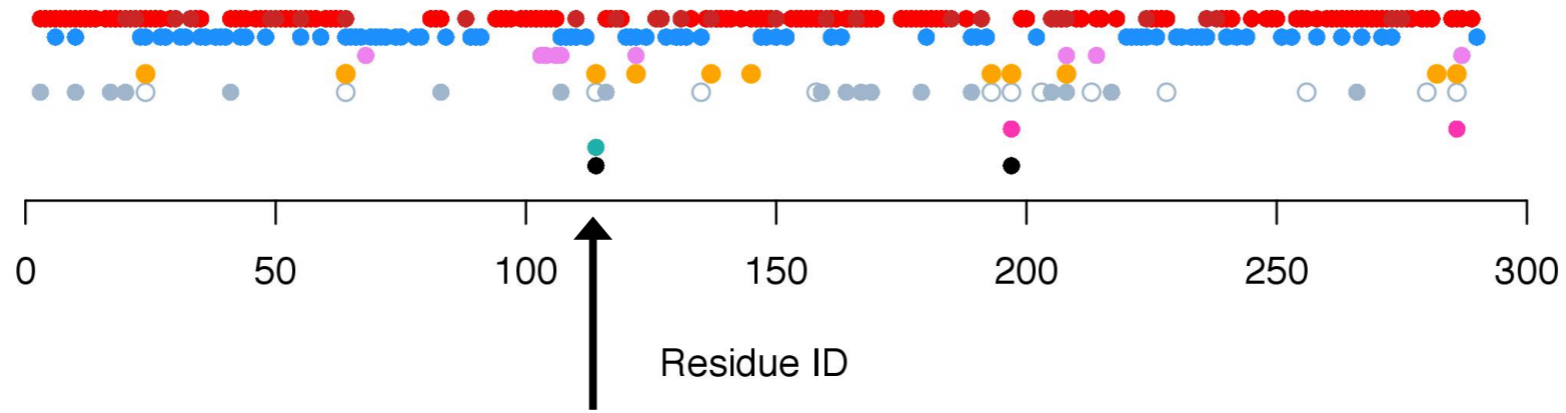




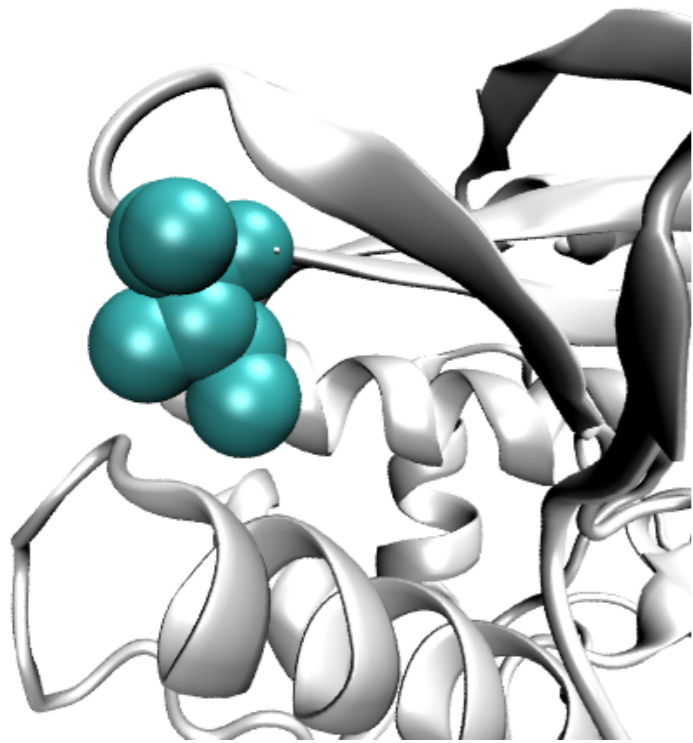


Analysis courtesy of Sushant Kumar, Yale University

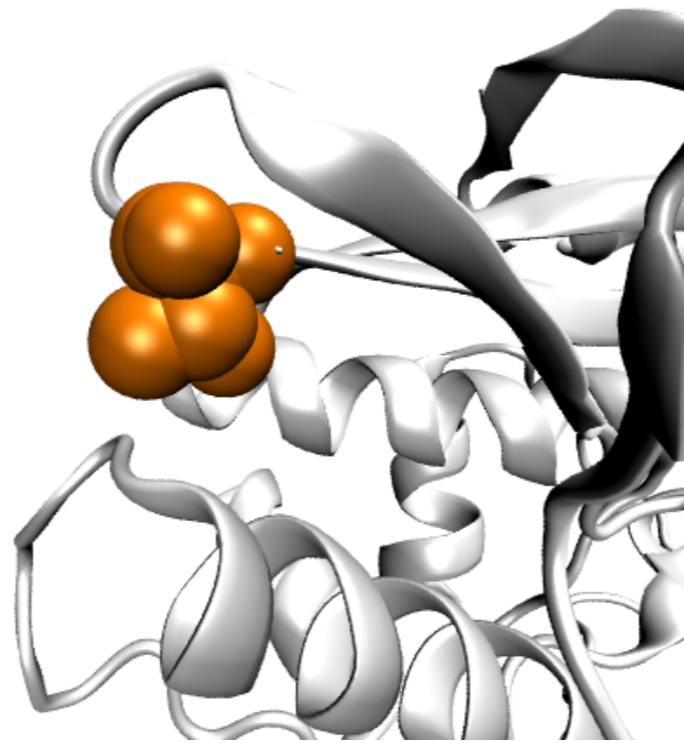
# Arylamine N-acetyltransferase 2 (2PFR\_A: gene = NAT2)



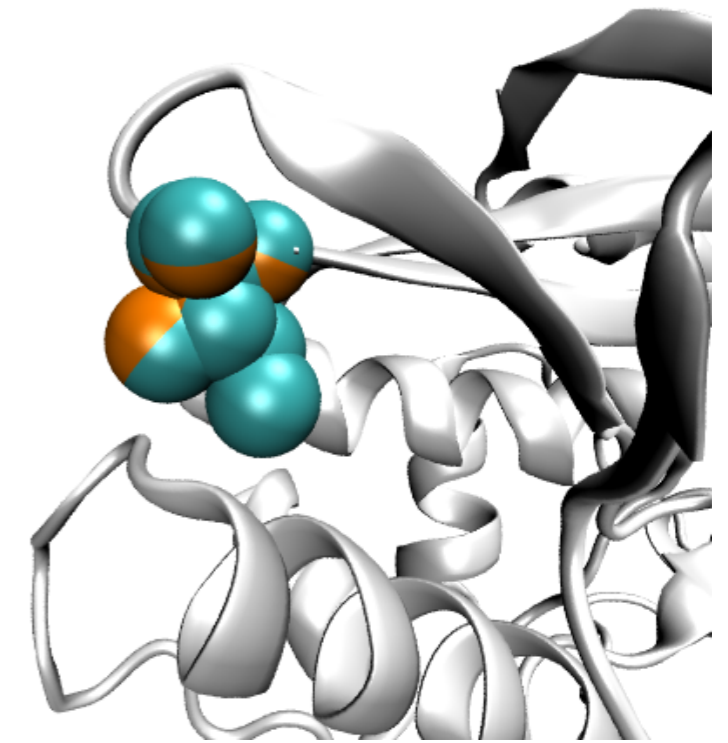
**114: I->T**



**Wild-type**



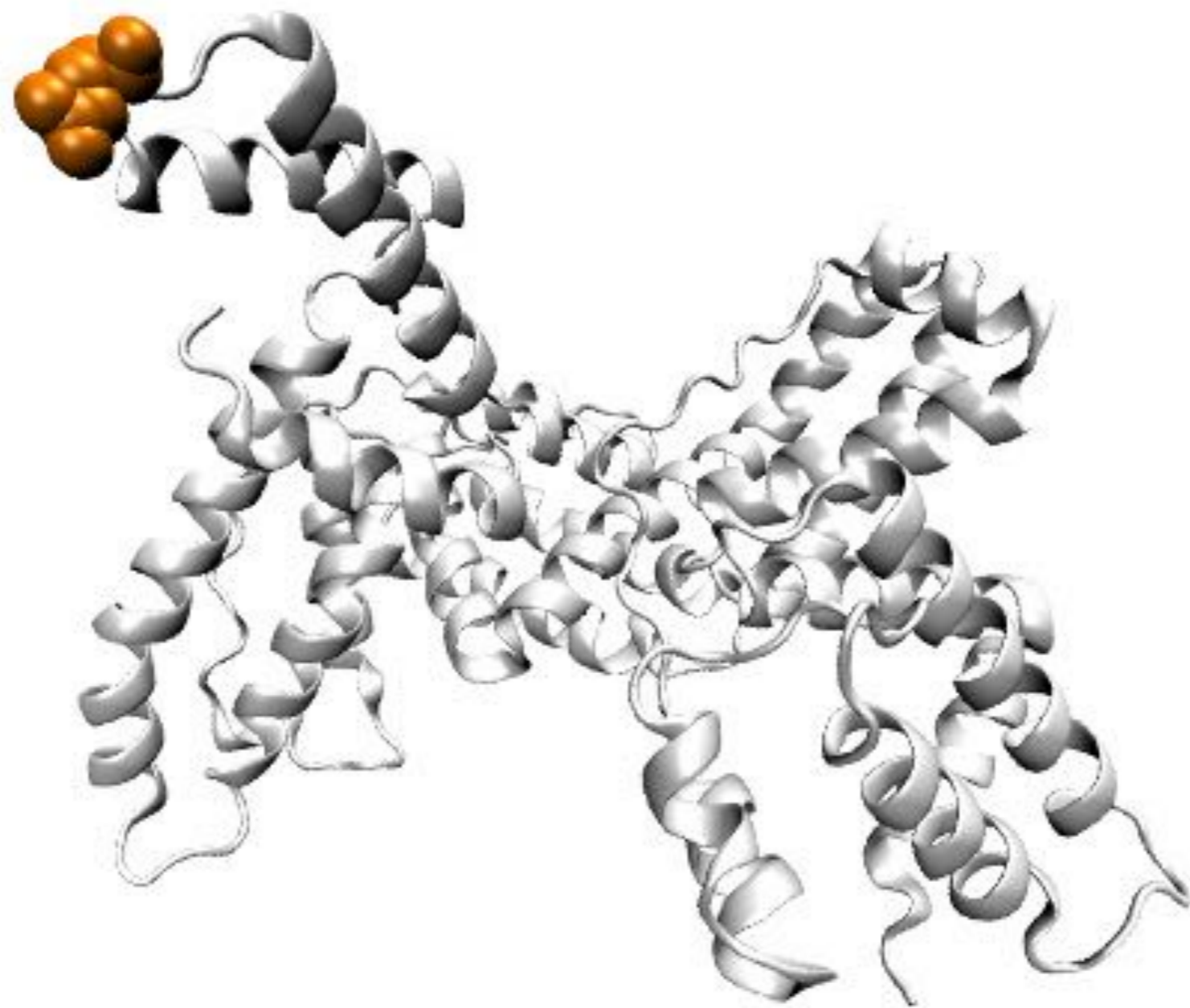
**Mutated**



**(superimposed)**

Analysis courtesy of Declan Clarke, Yale University







Ali Torkamani,  
Scripps Translational  
Science Institute

My variant: rs11209026  
Gene: IL23R



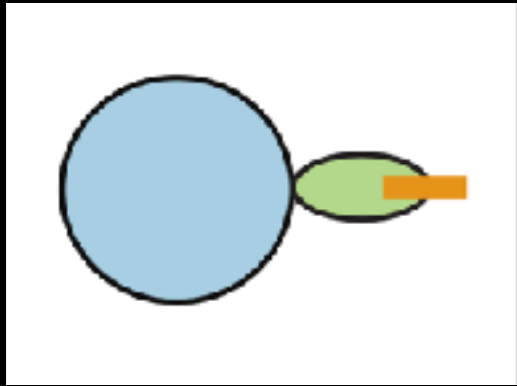




Bob Handsaker,  
Broad Institute

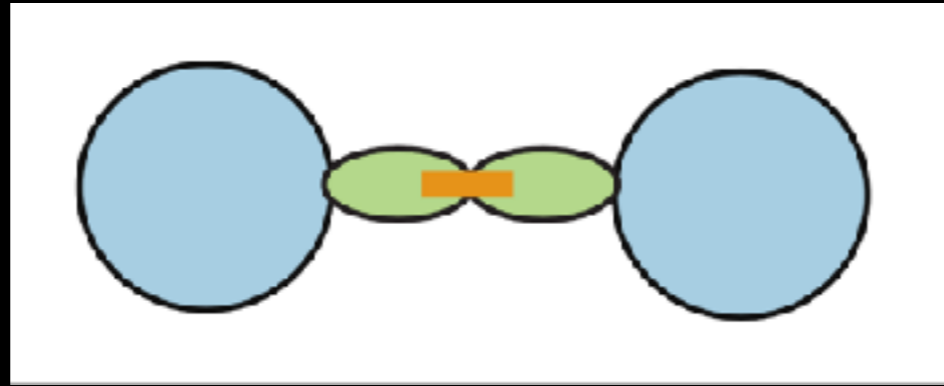
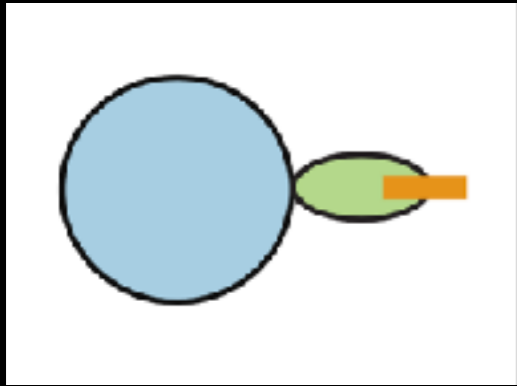


Haptoglobin

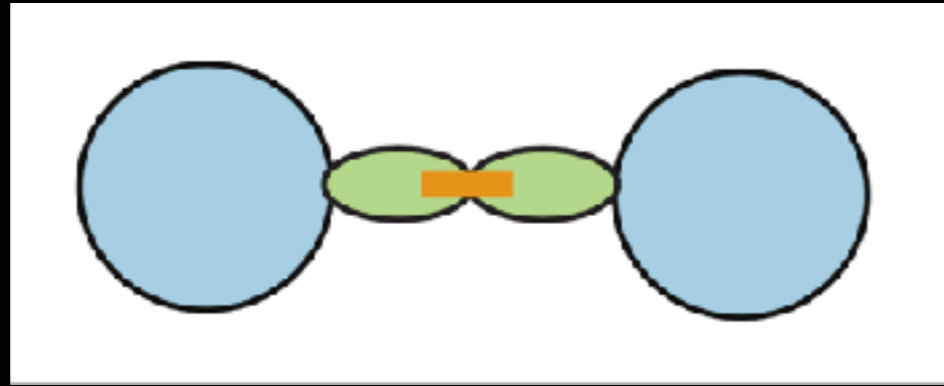
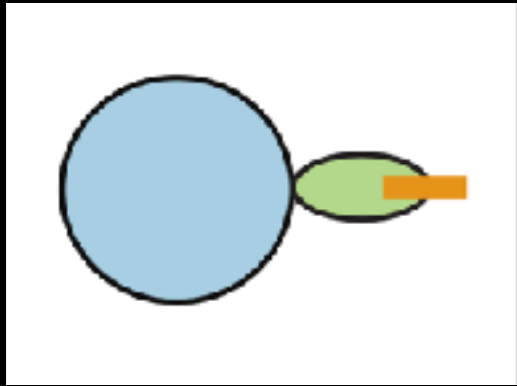


Haptoglobin





Haptoglobin

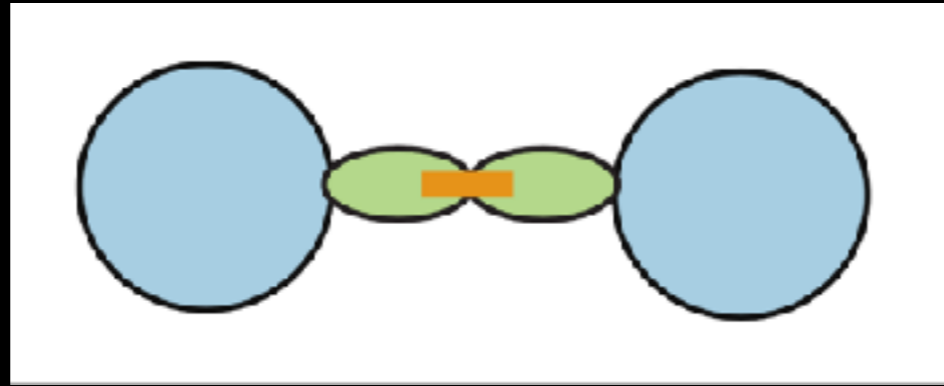
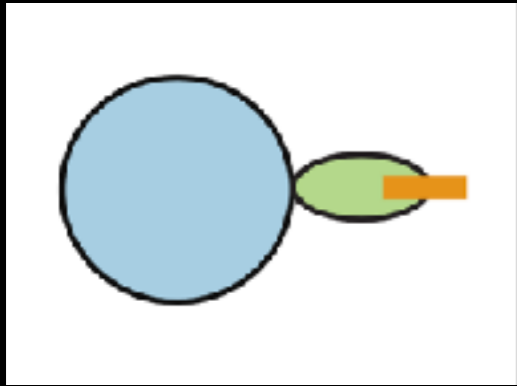


Lower cholesterol



Haptoglobin



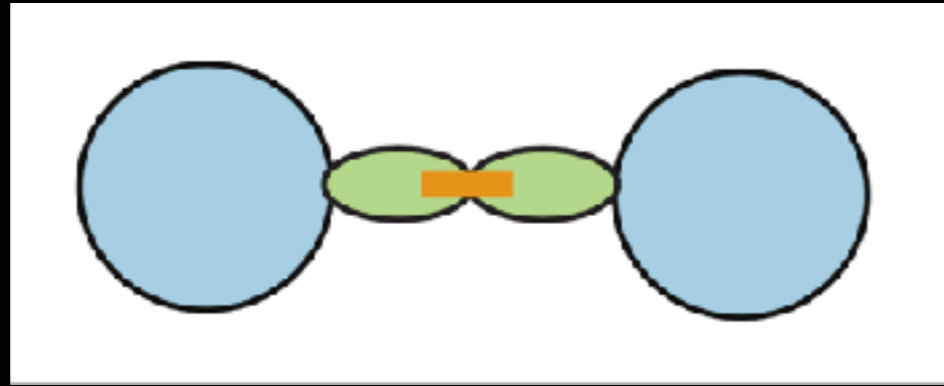
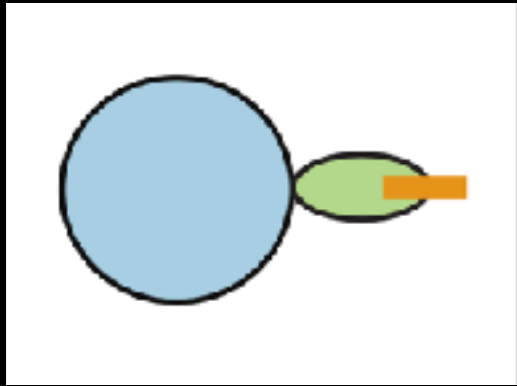


Lower cholesterol



Haptoglobin

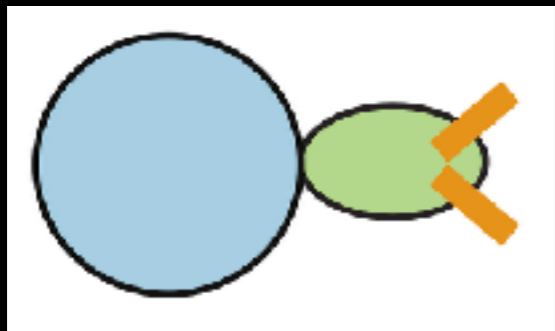


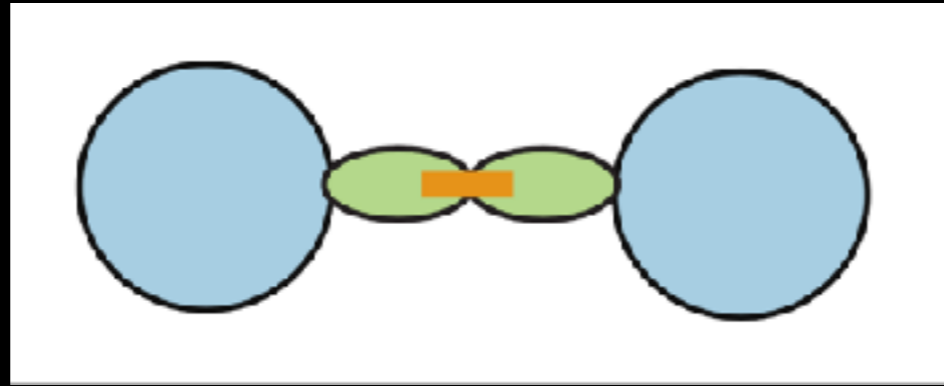
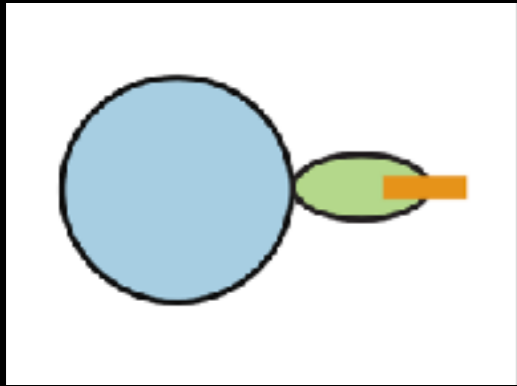


Lower cholesterol



Haptoglobin

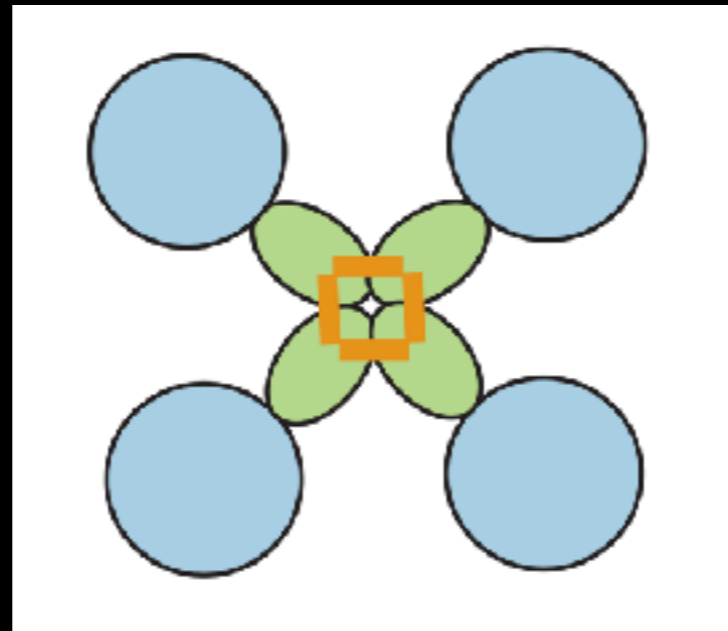
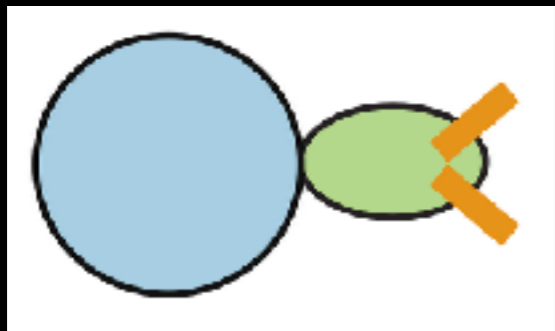




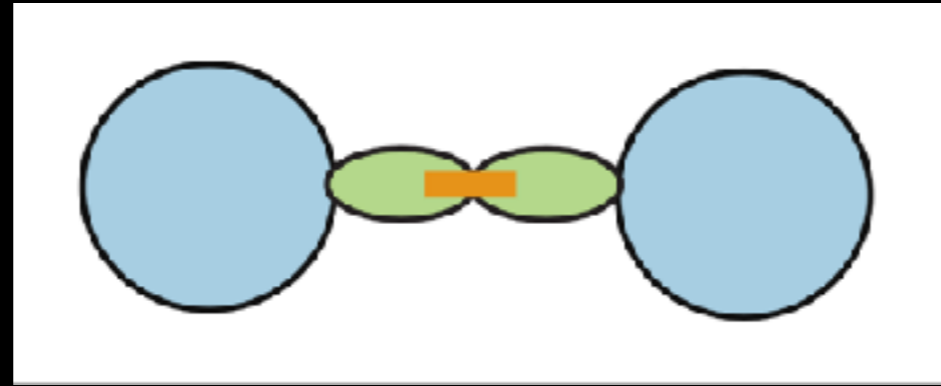
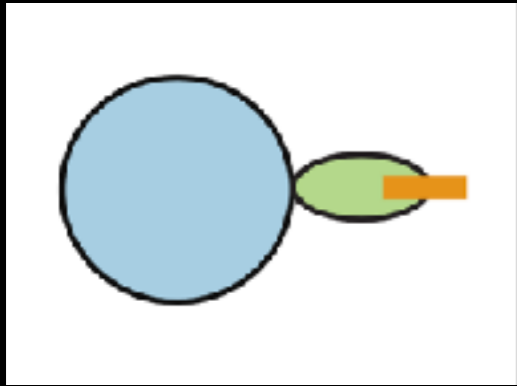
Lower cholesterol



Haptoglobin



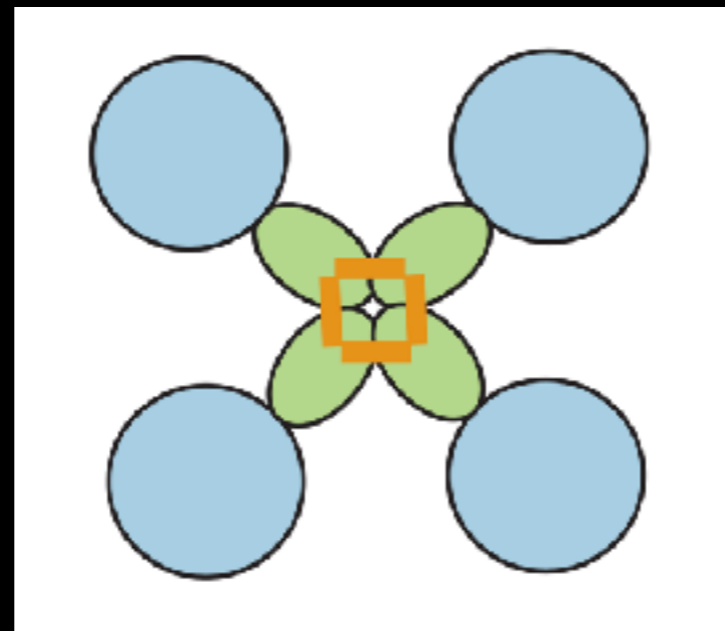
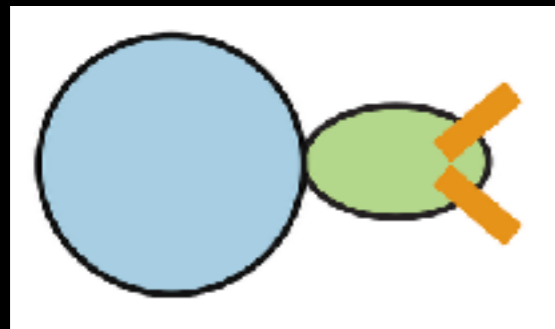




Lower cholesterol



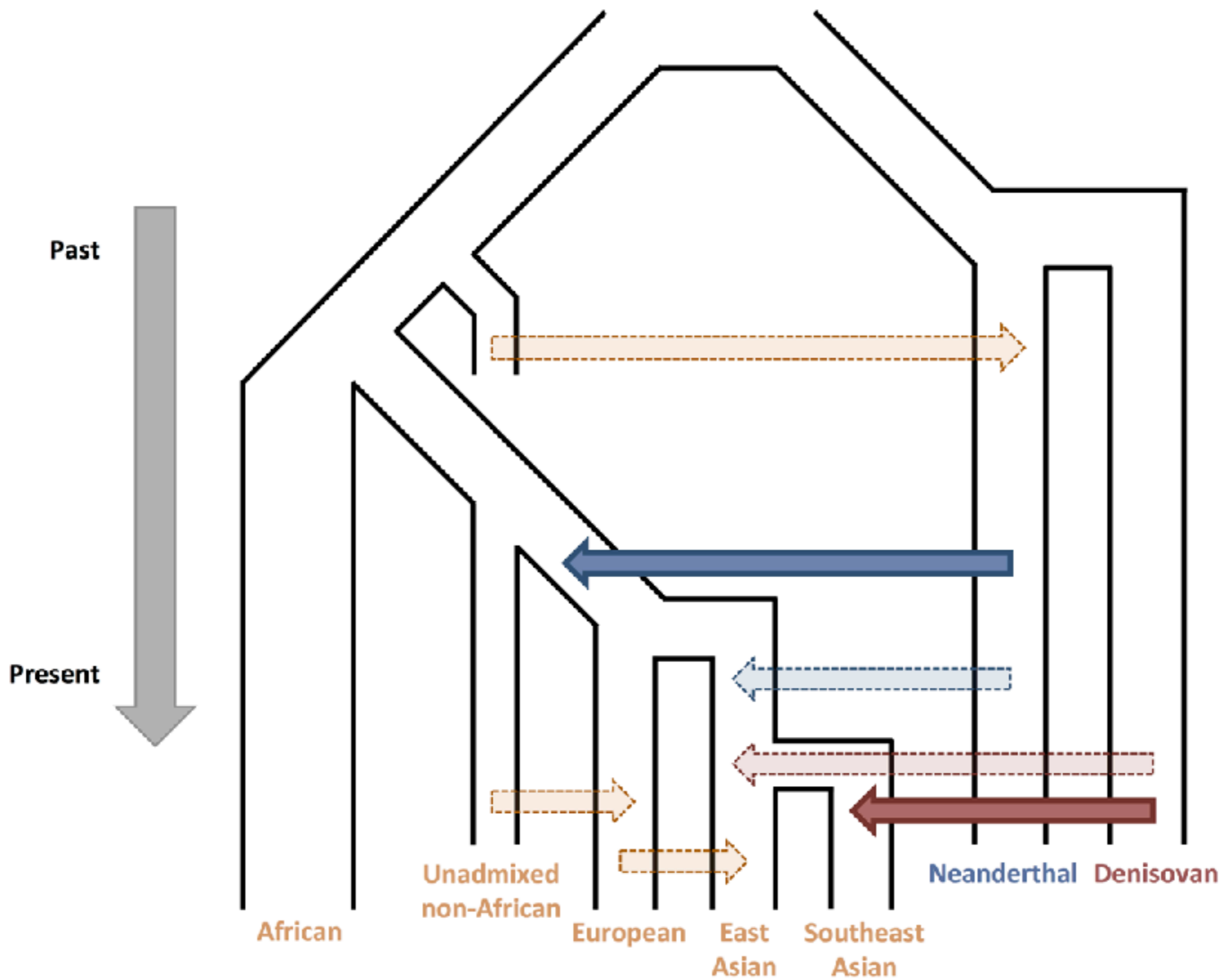
Haptoglobin



Higher cholesterol



Image: Viktor Deak



Wolf, Aaron B., and Joshua M. Akey. "Outstanding questions in the study of archaic hominin admixture." *PLoS genetics* 14.5 (2018): e1007349.





Vertical lines=Neanderthal segments  
 Circles=centromeres

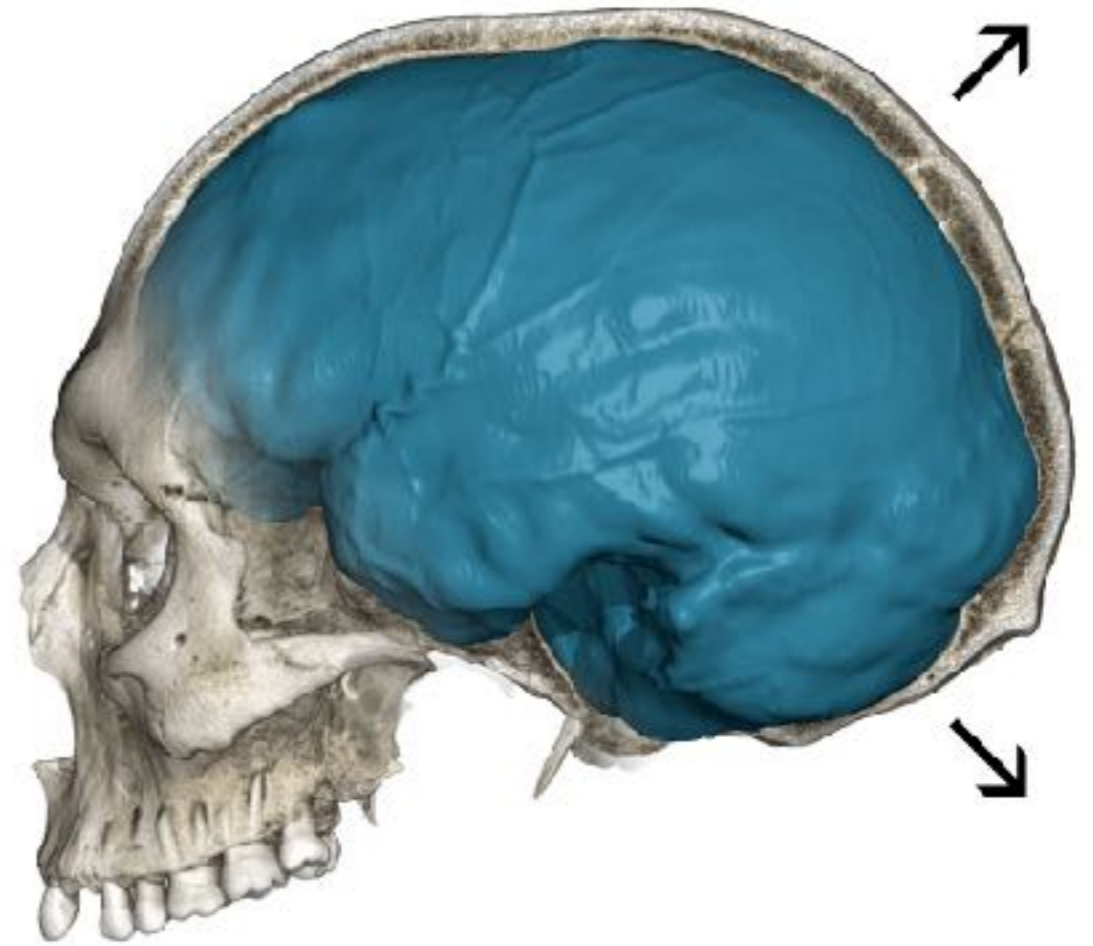
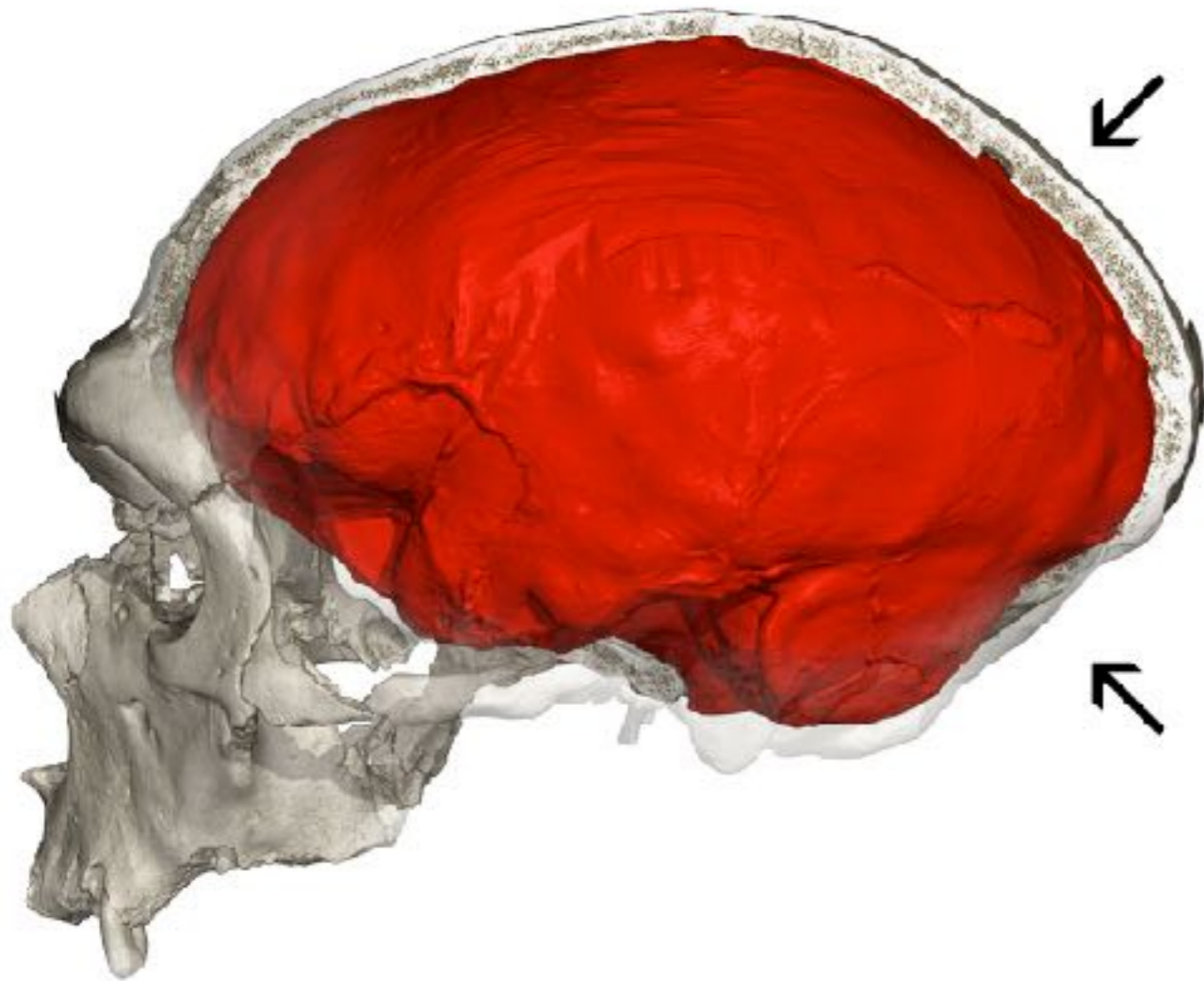


Analysis courtesy of Joshua Akey  
 & Selina Vattathil, Princeton

gene	chrom	gene_start	gene_end
MIR7846	chr1	12226999	12227095
MIR4632	chr1	12251769	12251830
TNFRSF1B	chr1	12227059	12269277
TNFRSF8	chr1	12123433	12204264
LRR38	chr1	13801444	13840242
C1orf64	chr1	16330730	16333190
HSPB7	chr1	16340522	16345285
ZBT617	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
QSOK1	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](https://doi.org/10.1016/j.cell.2018.05.011)





- Main
- Blood
- PGx

### rs12203592(C;T)

Primarily in Europeans; slightly lighter hair and eye color, less tanning ability

plos rs12203592 showed the largest allele frequency difference between the Irish individuals and those individuals of Northern, Central European and Eastern European descent associated with hair color rs12896399 rs12203592 for freckling was previously associated with hair color, eye color, and tanning response to sunlight. A genome-wide association scan in admixed Latin Americans identifies loci influencing facial and scalp hair features Squamous Cell Carcinoma [PharmGKB:Non-Curated GWAS Results: A Genome-Wide Association Study Identifies Novel Alleles Associated with Hair Color and Skin Pigmentation (Initial Sample Size: 2,287 women; Replication Sample Size: up to 8,465 individuals; Risk/trait: Allele: rs12203592-T). This variant is associated with Black vs. red hair color.] [GW...  
[more info](#)

2	Magnitude
Other	ClinVar Significance
26.5%	Frequency
0.05372	GMAF
22	References
<a href="#">IRF4</a>	Gene
6	Chromosome
395321	Position
2	Max Magnitude
20150831	R <sub>s</sub> time
plus	Stabilized
plus	Orientation

- Topics: Appearance Eye color Freckling

ClinVar Other Skin/hair/eye pigmentation  
 {MLEAC: [1], 'AC': [1], 'BaseQRankSum': 0.996, 'MLEAF': [0.5], 'culprit': 'QD', 'AF': [0.5], 'VQSLOD': 1.94, 'POSITIVE\_TRAIN\_SITE': True, 'AN': 2, 'MQ0': 0, 'FS': 4.773, 'MQ': 59.45, 'ClippingRankSum': -0.155, 'MQRankSum': 2.439, 'PrometheaseInferredRnum': True, 'ReadPosRankSum': 0.578, 'DP': 39, 'QD': 17.59}

### rs8085664(A;C)

0.7Bx reduced risk of Male Pattern Baldness. Discovered by 23andMe based on customer surveys, and considered preliminary research.

This is the 23andMe discovery: 'Male Pattern Baldness: Preliminary Research'. It affects appearance.  
[more info](#)

Good	Repute
2	Magnitude
40.7%	Frequency
0.3012	GMAF
<a href="#">SLC14A2</a>	Gene
18	Chromosome
45234191	Position
2.1	Max Magnitude
20150106	R <sub>s</sub> time

# CARL ZIMMER'S GAME OF GENOMES



*Illustration: Molly Ferguson for STAT;*

## Supplementary Materials

Welcome to the companion site for Carl Zimmer's "[Game of Genomes](#)" series on [STAT](#). Here you will find data and analysis from some of the scientists who examined Zimmer's genome.

### Part 1

[Gerstein lab](#)

### Part 2

[Torkamani lab](#)

[zimmerome.gersteinlab.org](http://zimmerome.gersteinlab.org)



CARL ZIMMER



The Powers, Perversions,  
*and Potential of Heredity*

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