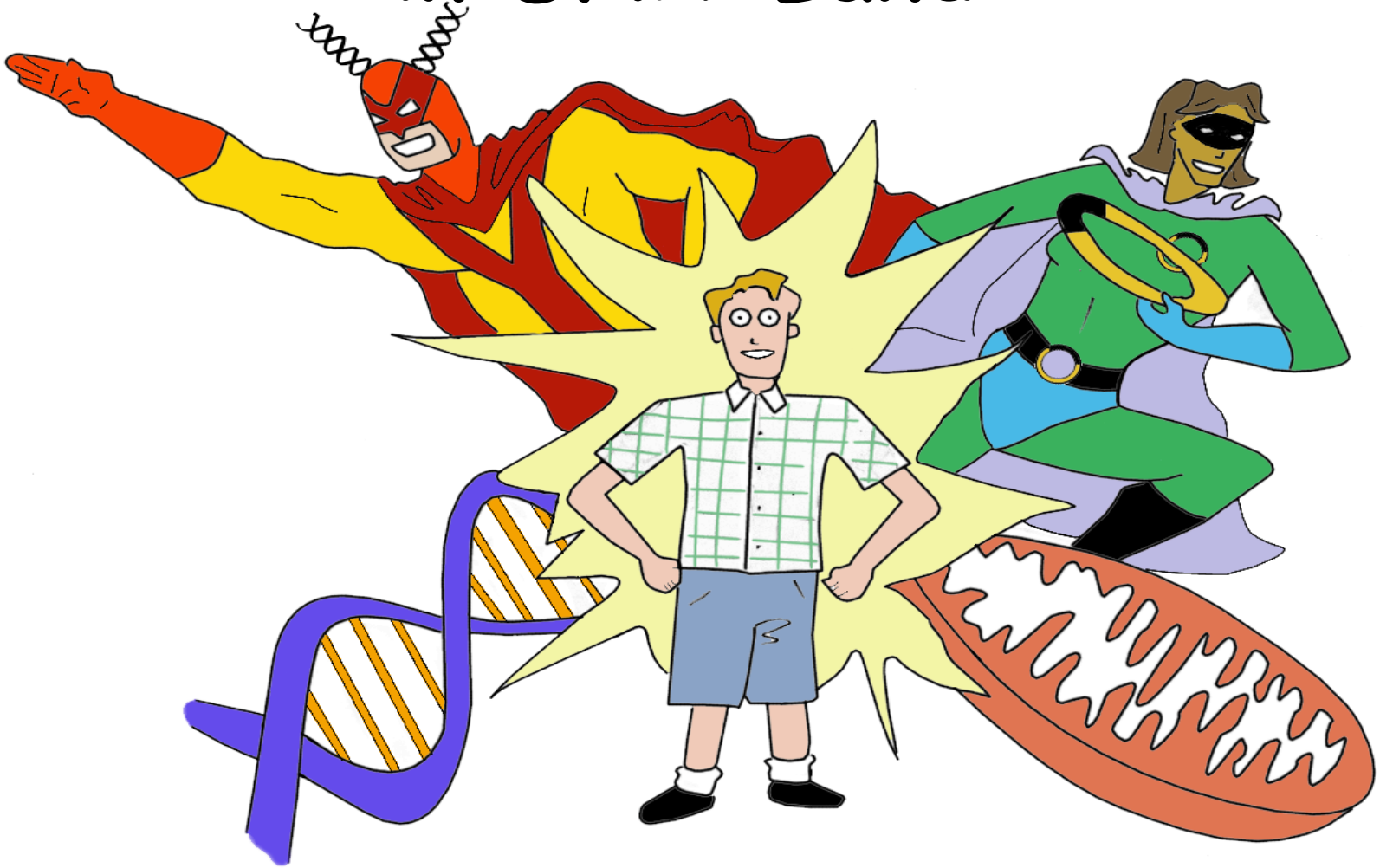
Ernie Ebayley's Adventure in DNA-Land



A Resource for Beginning Your Own Adventure into Genealogical Genetics

C.E. Smith Museum of Anthropology
College of Arts, Letters, and Social Sciences (CLASS)
California State University, East Bay
Hayward, CA 94542 (510) 885-3104
http://class.csueastbay.edu/cesmith/acesmith.html

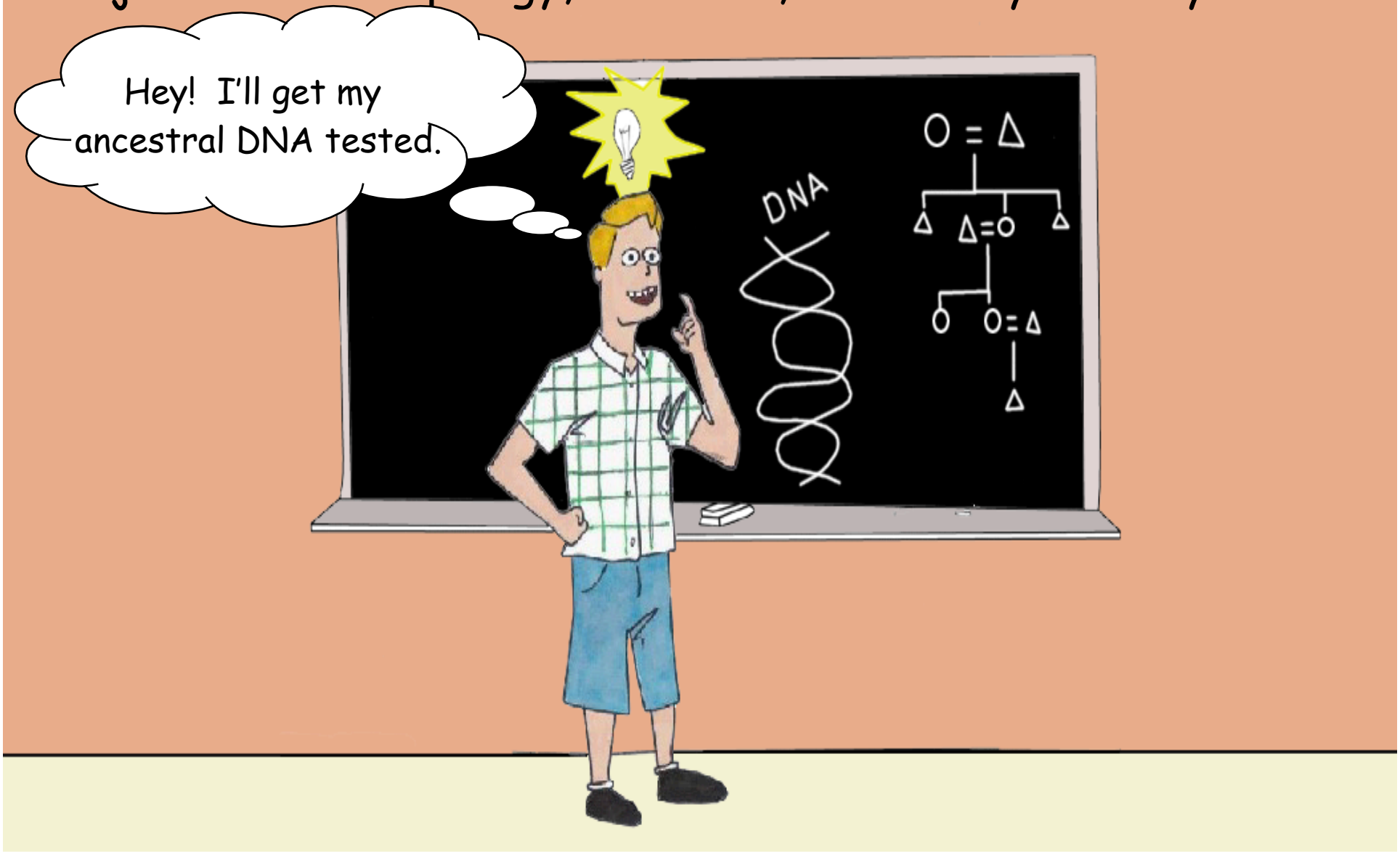
Text by George R. Miller, Ph.D.
Cartoons by Danny McNaughton and Gary Francis

Prepared for the exhibition

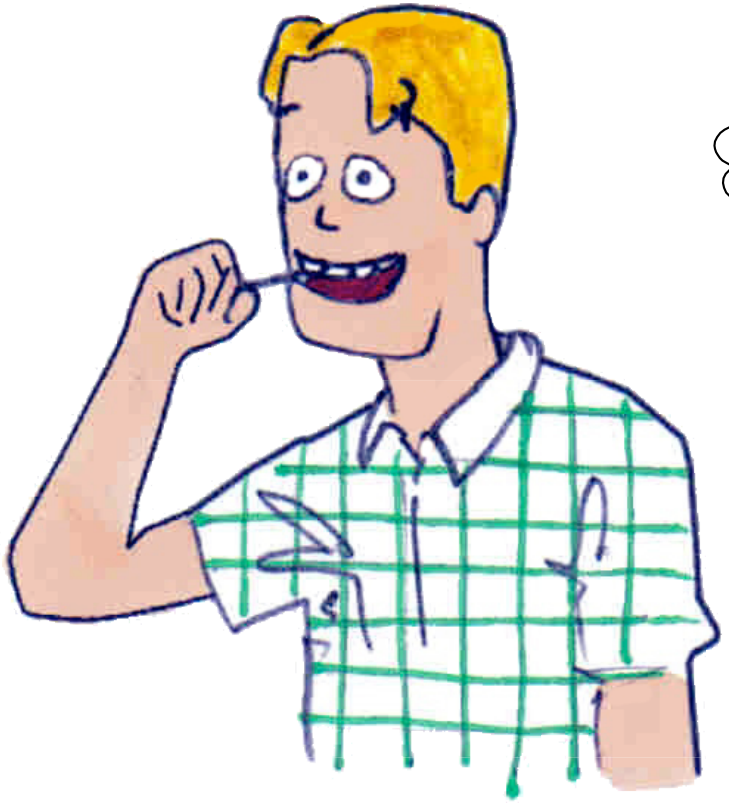
Immigrants All: Our MigrationTales and GeneticTrails

(February 24 - June 9, 2006)

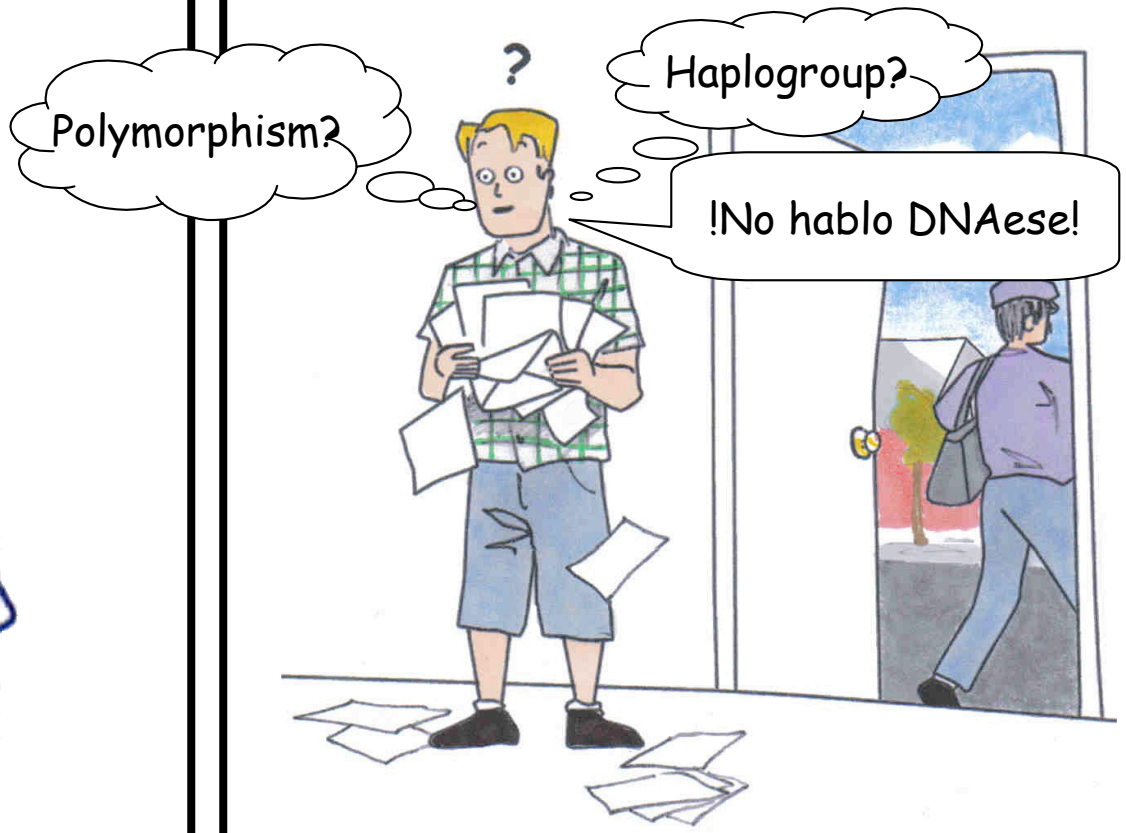
Finally, after fifteen quarters at CSUEB, Ernie Ebayley finds a way to tie together his fascination with his three favorite subjects - Anthropology, Genetics, and Family History.

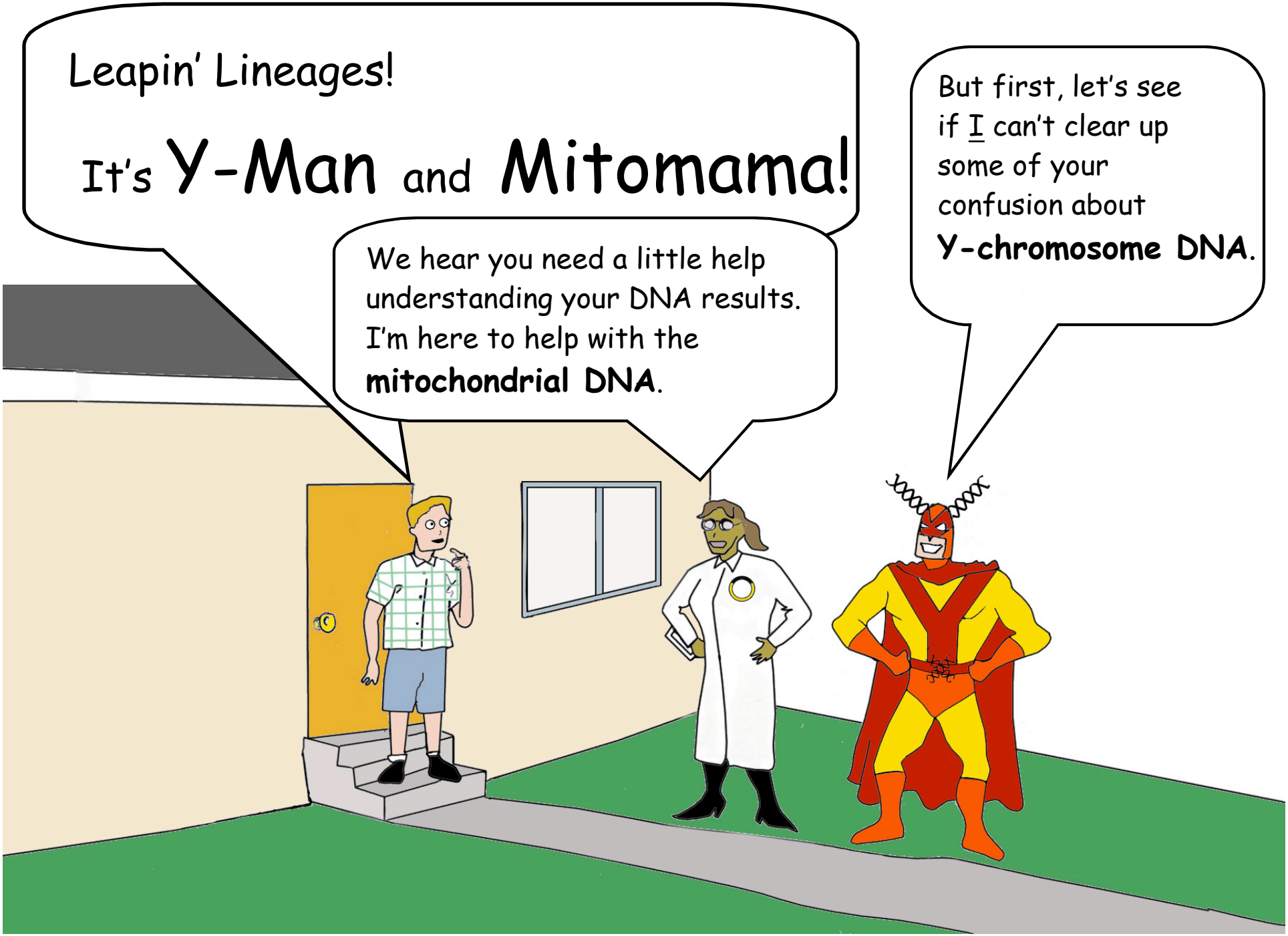


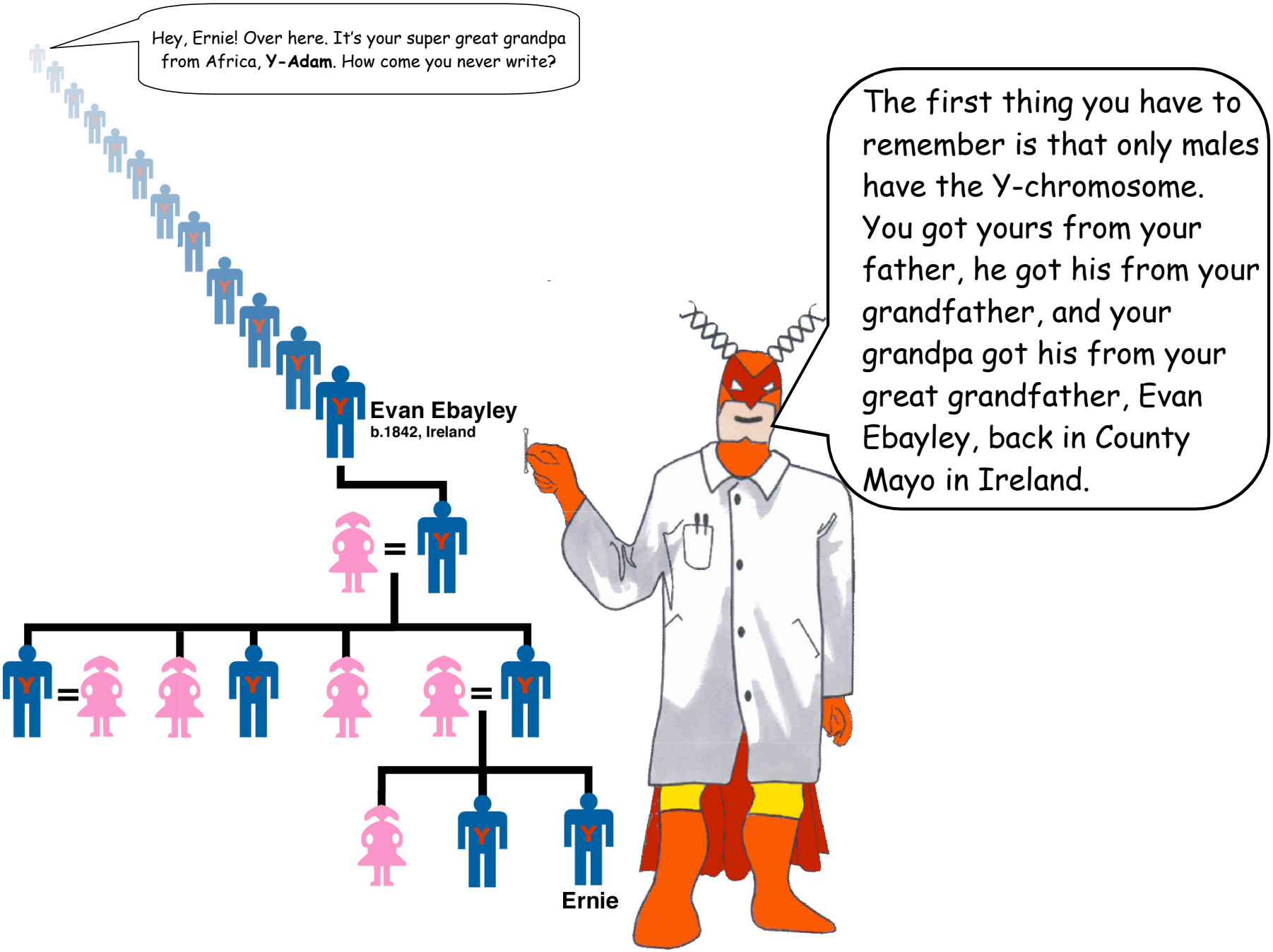
Ernie contacts the Family Tree DNA lab and sends off for a sampling kit. He spends a few minutes scraping the inside of his cheek, mails back the sampling vials, and waits.



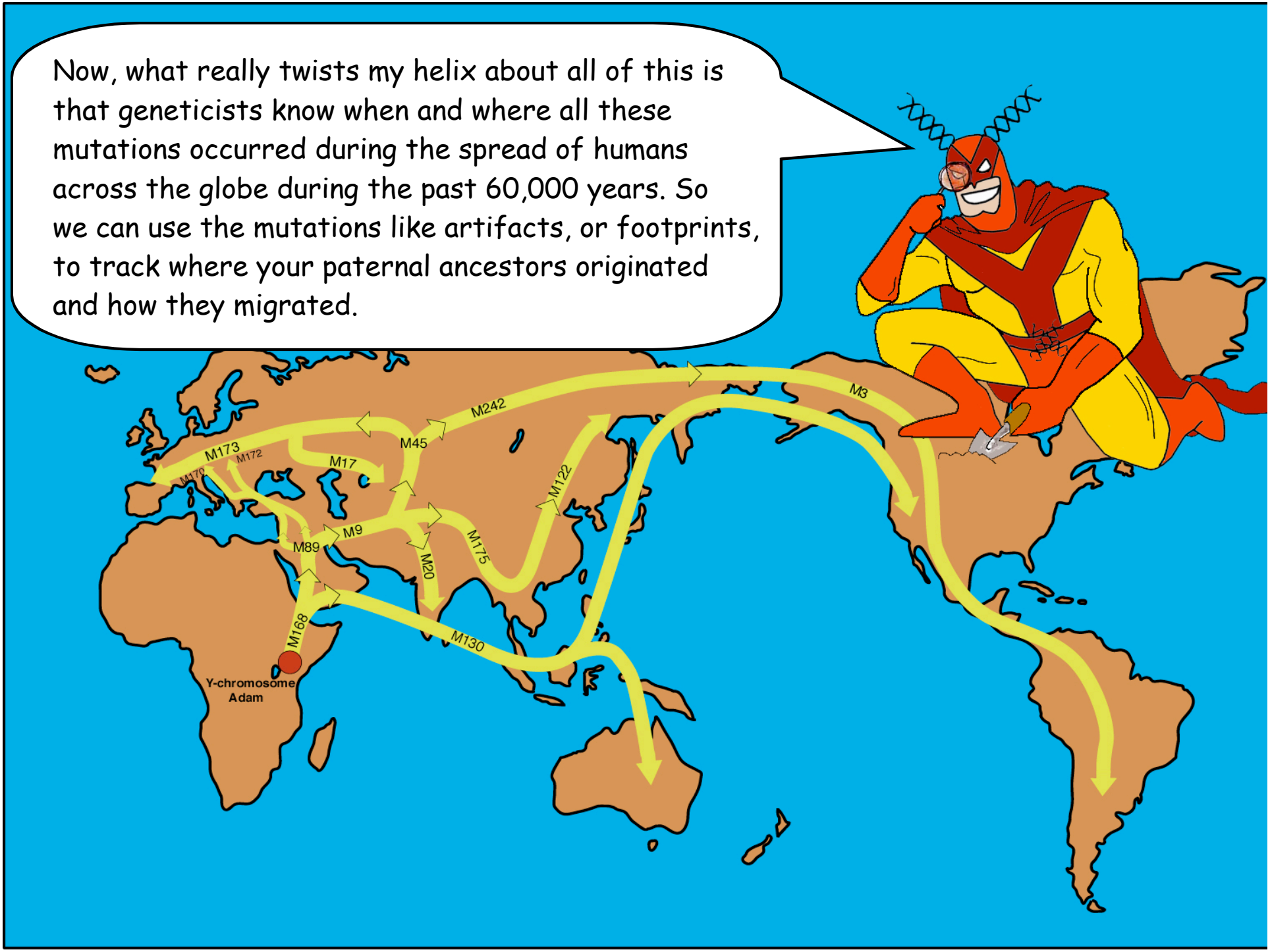
Six weeks later he receives a list of his DNA markers along with some other very confusing data. Fortunately, his DNA certificate comes with a special customer support offer, so soon he receives a visit from …



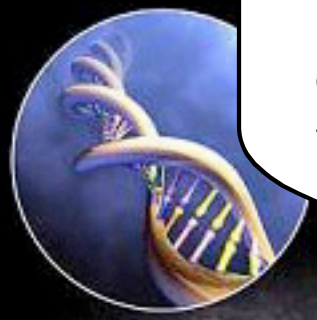

Leapin' Lineages!
It's Y-Man and Mitomama!
We hear you need a little help understanding your DNA results. I'm here to help with the **mitochondrial DNA**.
But first, let's see if I can't clear up some of your confusion about **Y-chromosome DNA**.

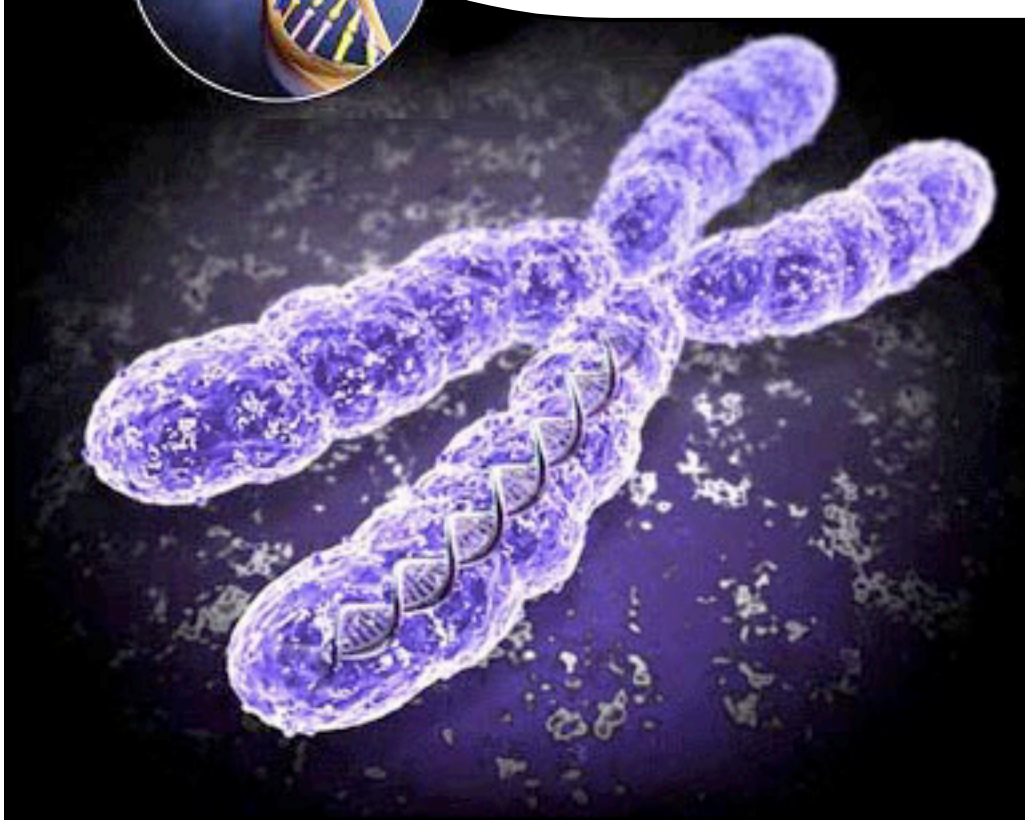

Hey, Ernie! Over here. It's your super great grandpa from Africa, **Y-Adam**. How come you never write?
Evan Ebayley
b.1842, Ireland
Ernie
The first thing you have to remember is that only males have the Y-chromosome. You got yours from your father, he got his from your grandfather, and your grandpa got his from your great grandfather, Evan Ebayley, back in County Mayo in Ireland.

In essence, your Y-chromosome test shows you all the mutations that have accumulated on the Y-chromosomes of your direct paternal ancestors, going back hundreds, even thousands of generations into the remote past - all the way back to a man that lived in Africa over 100,000 years ago. Geneticists call him **Y-chromosome Adam**.


Now, what really twists my helix about all of this is that geneticists know when and where all these mutations occurred during the spread of humans across the globe during the past 60,000 years. So we can use the mutations like artifacts, or footprints, to track where your paternal ancestors originated and how they migrated.
M3
M242
M45
M173
M172
M170
M17
M122
M9
M89
M175
M20
M168
M130
Y-chromosome
Adam

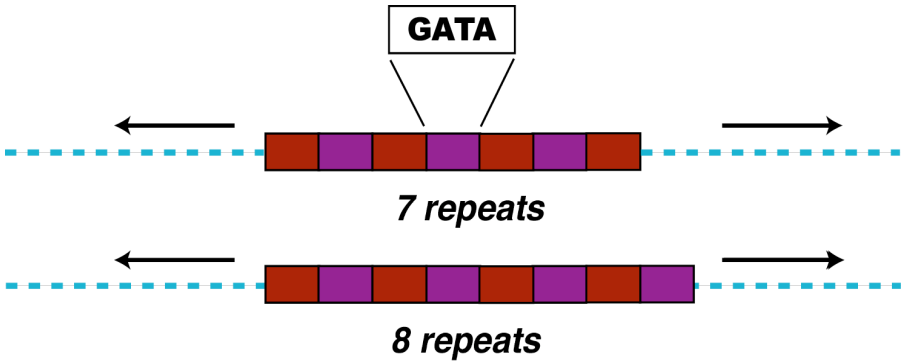
Here's how it works. The Y-Chromosome is a strand of DNA, approximately 60,000 **base pairs** (**nucleotides**) long, with only one fundamental purpose - to determine your maleness. Most of the Y-chromosome does not code for anything and is sometimes called **junk DNA.** The non-coding region of the Y-chromosome, however, has accumulated a tremendous number of benign mutations or **markers** over the millennia and they are the genetic artifacts that we use to decipher your ancestral history.





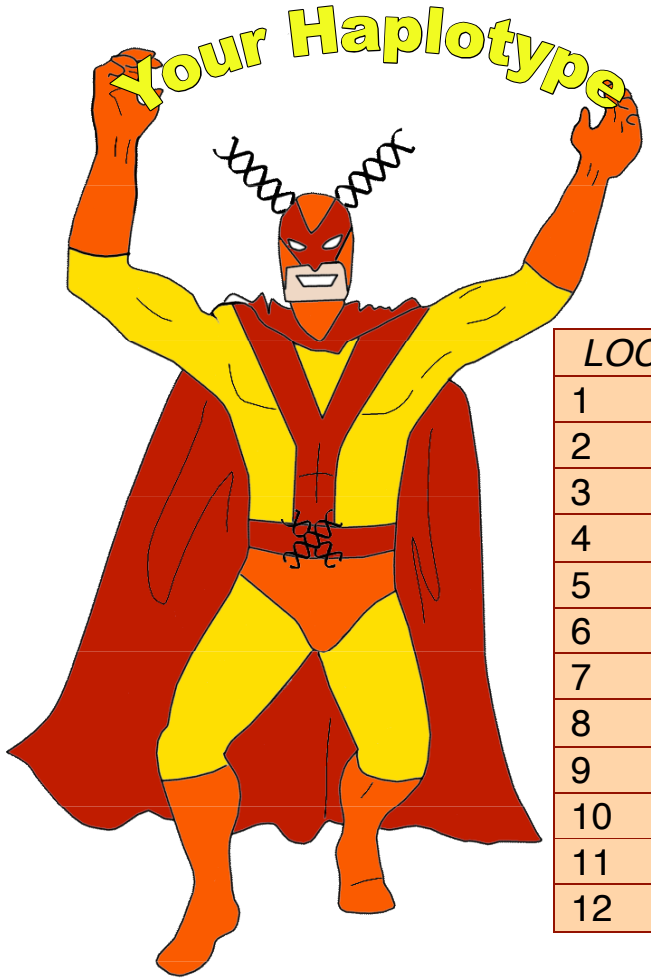
Geneticists have identified hundreds of markers on the Y-chromosome. Some are very rare and probably occurred only once in human history, but they are difficult and expensive to identify, so we first look for ancestral clues in a more common type of marker, called **Short Tandem Repeats**.

Short Tandem Repeats (STRs) consist of segments of the Y-chromosome where a short pattern of nucleotides stutters, or repeats itself.

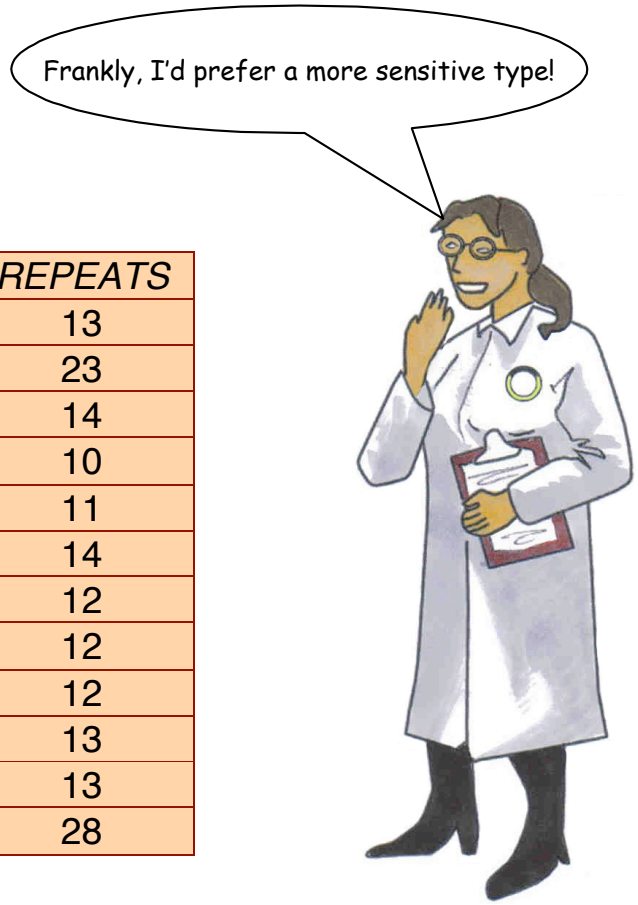


For example, at a particular location on your Y-chromosome the four-base pattern GATA (Guanine, Adenine, Thymine, Adenine) might be repeated seven times - GATA GATA GATA GATA GATA GATA GATA - while some other guy might have eight or nine repeats at the same location.

Your Y-Chromosome DNA was tested for STRs at 12 separate marker locations. On this chart you see your entire set, called your **haplotype,** and the number of repeats you have at each of the 12 locations. This is the profile we use to check how closely you are related to other males, whether they might be long-lost cousins with the same surname or just some random guy you meet on the street.



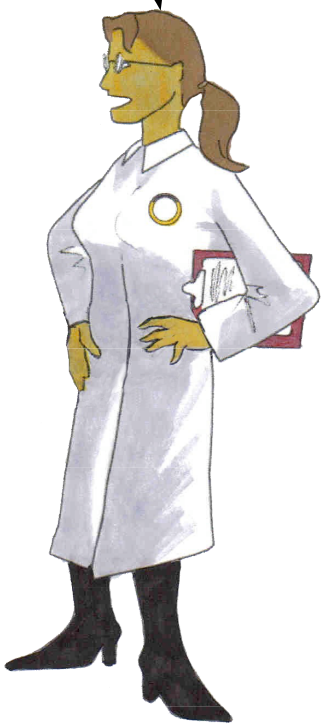
LOCUS	*DYS#*	*REPEATS*
1	393	13
2	390	23
3	19	14
4	391	10
5	385a	11
6	385b	14
7	426	12
8	388	12
9	439	12
10	389-1	13
11	392	13
12	389-2	28

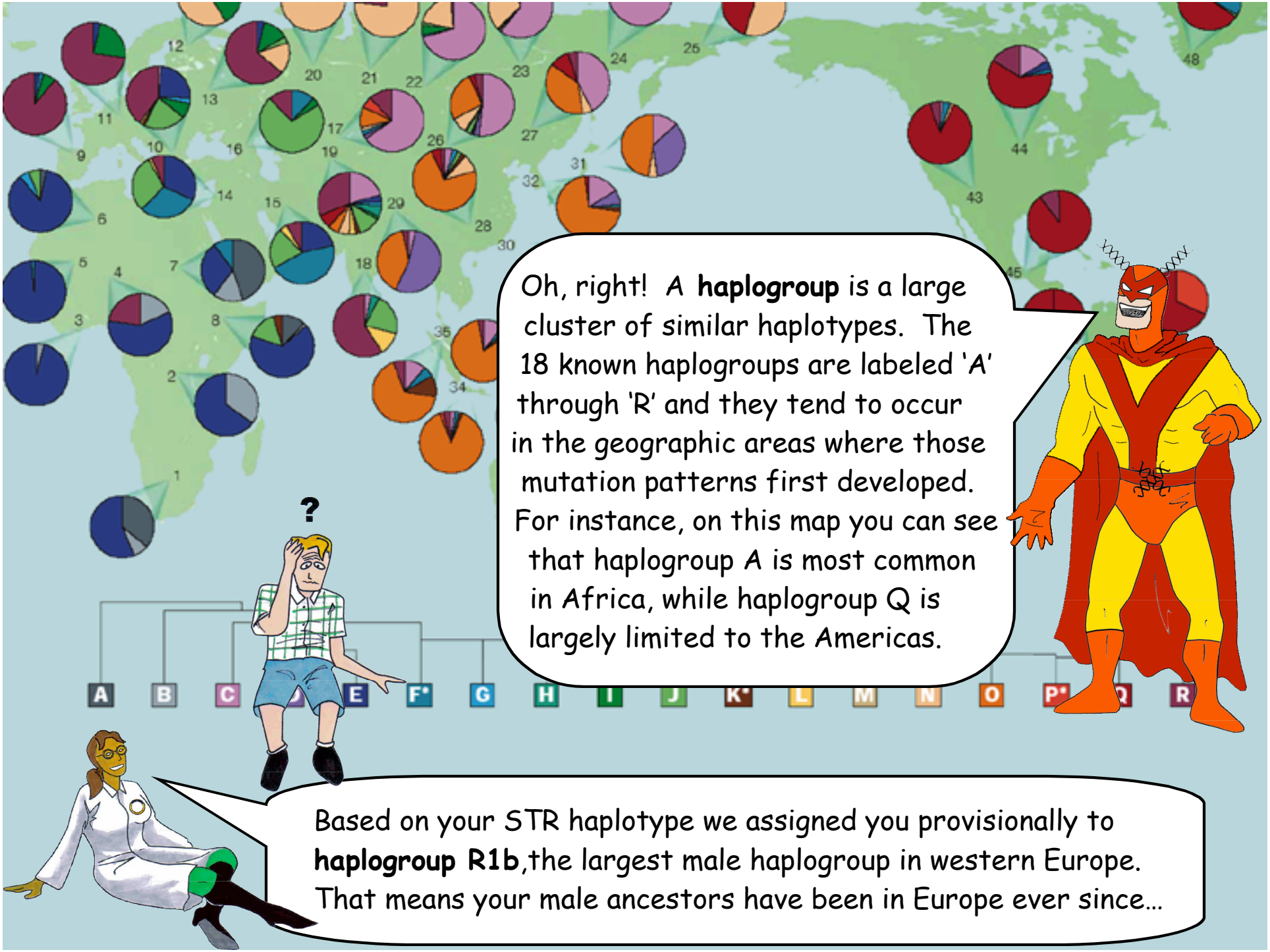


Let's compare your STR results with those of a few of the guys in your museum class. Your results are quite similar to those of Mike, with just a two mutation difference between the two of you on the 12 markers. If the two of you had the same surname, we would calculate a 29% probability that you and Mike had a common male ancestor in the past 500 years. On the other hand, there is much greater genetic distance between you and James, and even more between you and Ravi, whose grandfather was from India.

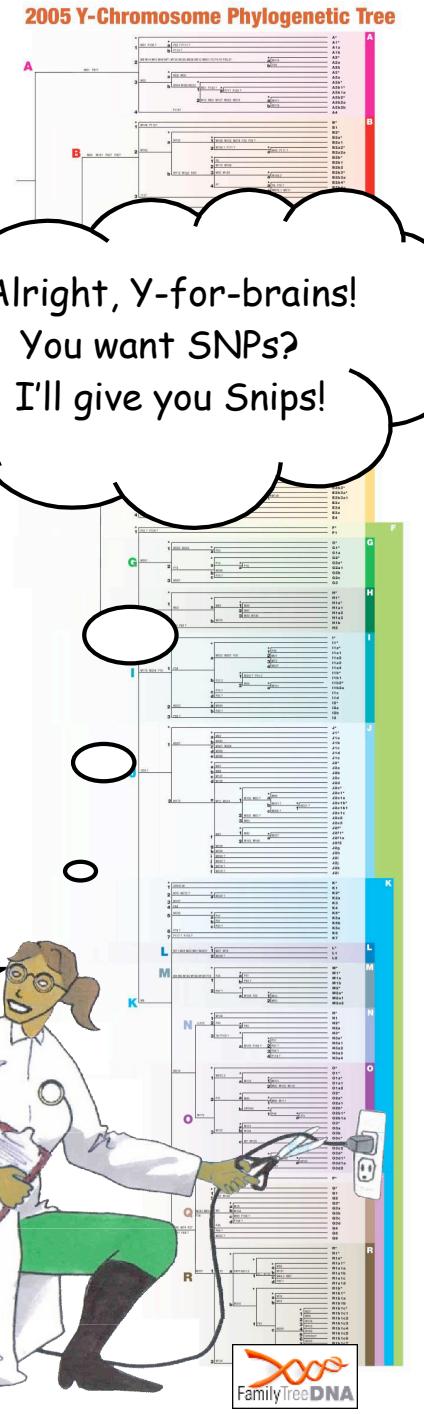
Holy Haplogroups, Y-Man!
Don't just show him numbers!
Ernie wants to know where his ancestors <u>came</u> from.

LOCUS	*DYS#*	Ernie	Mike *Swiss*	James *Bosnian*	Ravi *Indian*
1	393	13	13	13	12
2	390	23	23	23	22
3	19	14	14	15	16
4	391	10	11	10	10
5	385a	11	11	14	15
6	385b	14	14	14	16
7	426	12	12	11	11
8	388	12	12	12	12
9	439	12	12	11	11
10	389-1	13	13	12	13
11	392	13	13	11	11
12	389-2	28	29	27	30
Haplogroup		R1b	R1b	I	H
Genetic Distance			2	9	16




Oh, right! A **haplogroup** is a large cluster of similar haplotypes. The 18 known haplogroups are labeled 'A' through 'R' and they tend to occur in the geographic areas where those mutation patterns first developed. For instance, on this map you can see that haplogroup A is most common in Africa, while haplogroup Q is largely limited to the Americas.
?
A B C E F* G H I J K* L M N O P* Q R
Based on your STR haplotype we assigned you provisionally to **haplogroup R1b**,the largest male haplogroup in western Europe. That means your male ancestors have been in Europe ever since...

Wait, Ernie! We don't know that for sure. First we need to do a **Single Nucleotide Polymorphisms** (SNP) test. SNPs (pronounced "snips") are absolutely unique and totally cool Y-chromosome markers that allow us to both construct a phylogenetic tree showing how one haplogroup is related to another, and to track your ancestors' genetic footprints clear back to Africa. SNPs are my favorite! They're a little more expensive to test for, but I know you're going to love 'em. Here - let's take a look at this Y-chromosome tree to see if we can't trace the SNPs on your Y-chromosome. Your list of SNPs is M168, M89, M9, M45, M173, and M343. Sounds complicated, huh? But I can explain it. After all, I'm **Y-man** and I graduated Summa Cum Whydah. . . . .

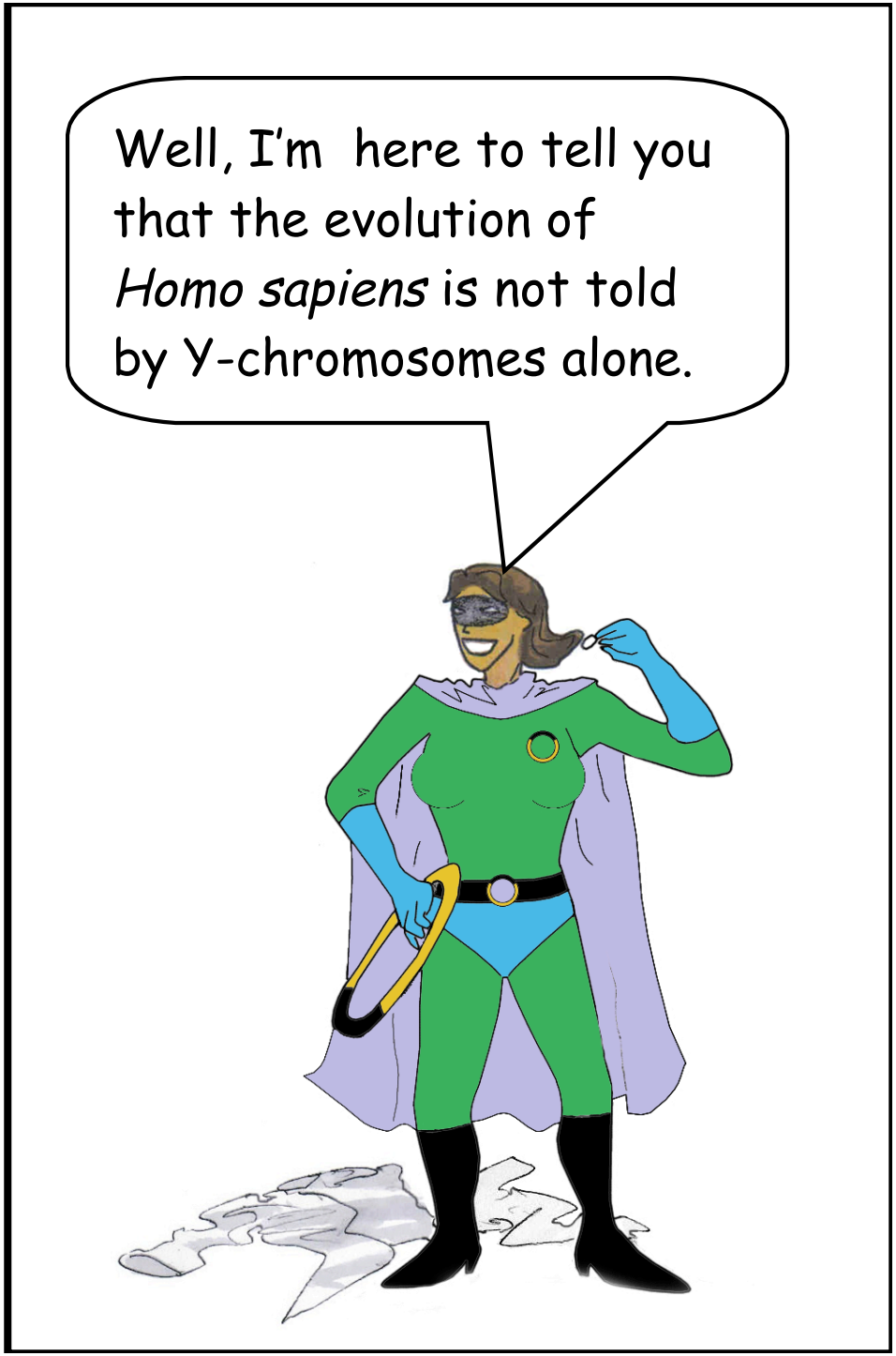


Alright, Y-for-brains! You want SNPs? I'll give you Snips!

Sorry, everyone! If you want to learn more About SNPs, turn to the back of the book.

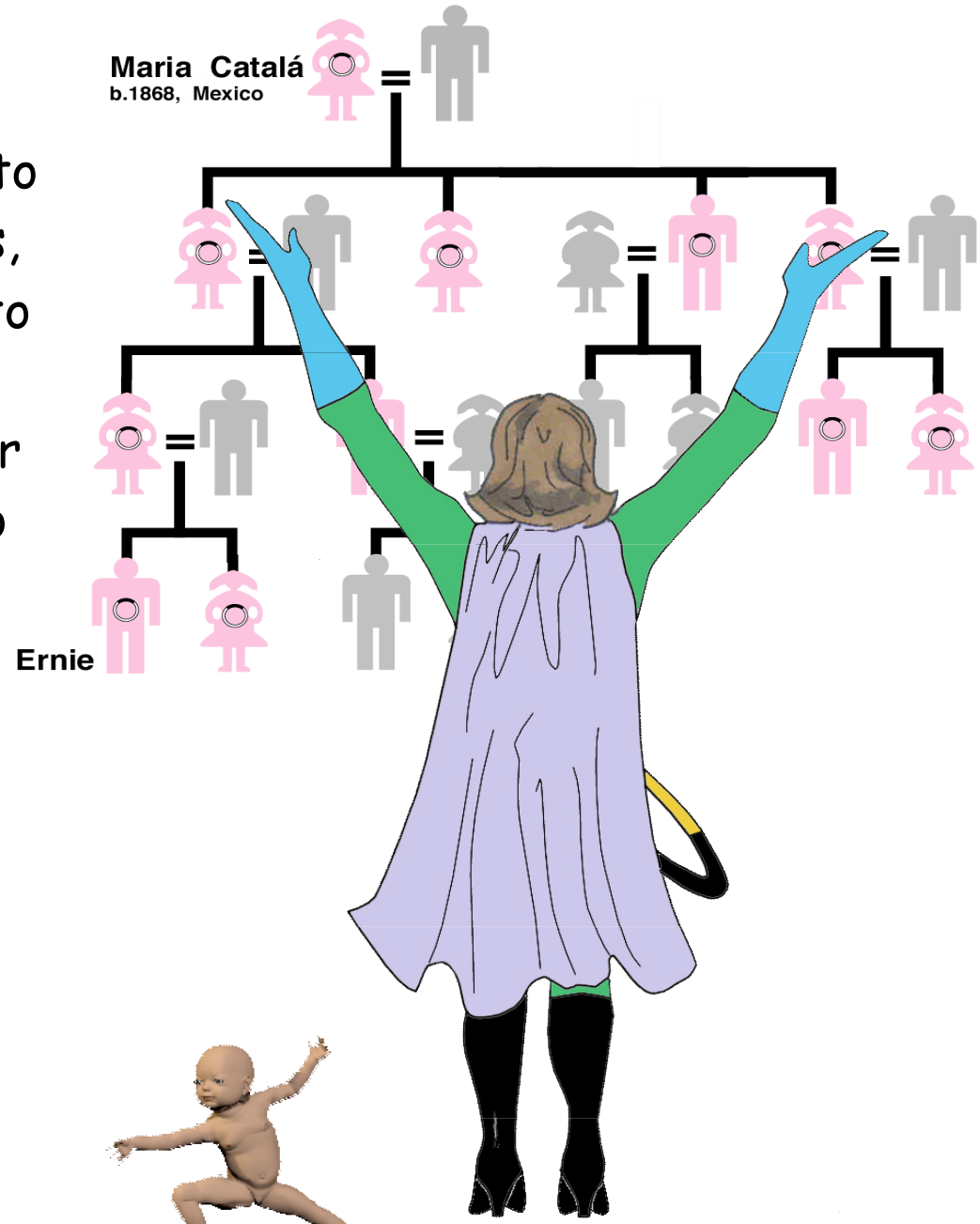

OK, I give up… but, Ernie I just wanted to tell you that you've only been Irish for a few thousand years. Your DNA shows that at the end of Ice Age your ancestors were Magdalenian cave artists in Spain - before that they were mammoth hunters on the Ukrainian steppes - and long before that they were some of the first Africans to venture across the Arabian Peninsula into Central Asia.
M173
M170
M172
M17
M45
M9
M89
M20
M168
Y-chromosome
Adam


Oh, brother! Just leave it to a man to turn a simple description of the tiniest little chromosome in the human body into some kind of a **grand epic journey** involving glacial ice, ferocious beasts, and all that other macho stuff.

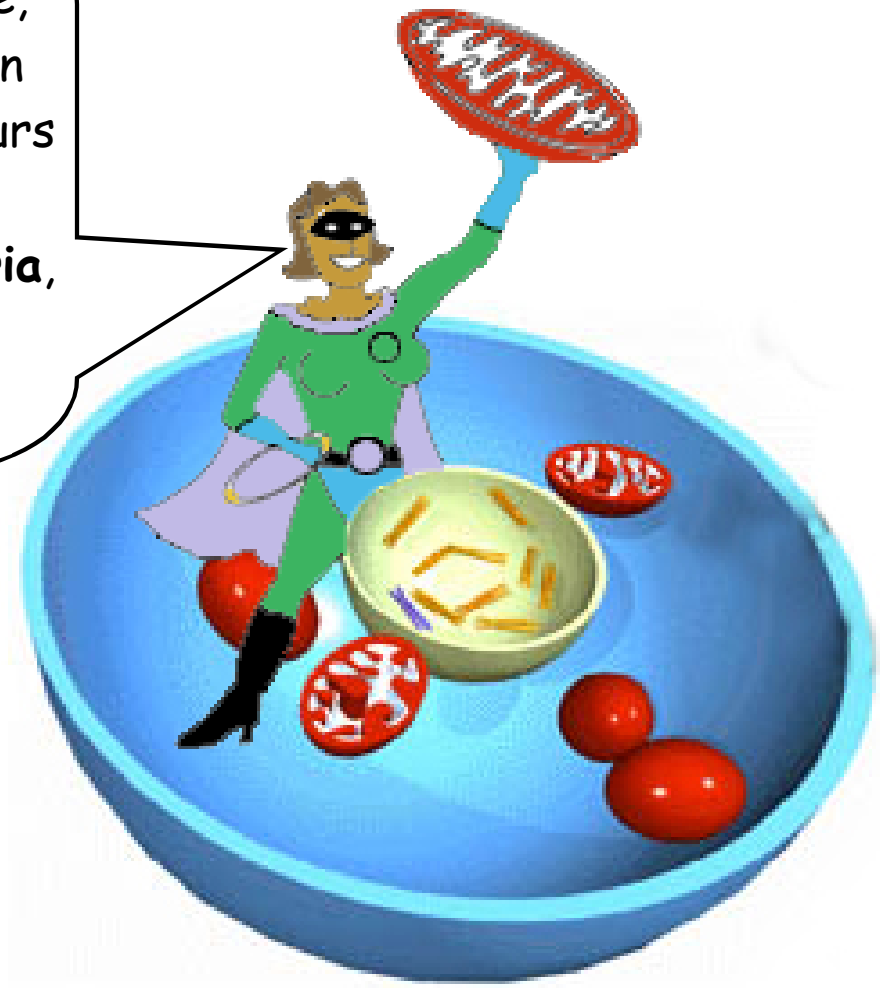

Well, I'm here to tell you that the evolution of *Homo sapiens* is not told by Y-chromosomes alone.

We women control another kind of DNA, called **mitochondrial DNA**, which we give generously to both our sons and our daughters, and those daughters pass it on to the next generation.

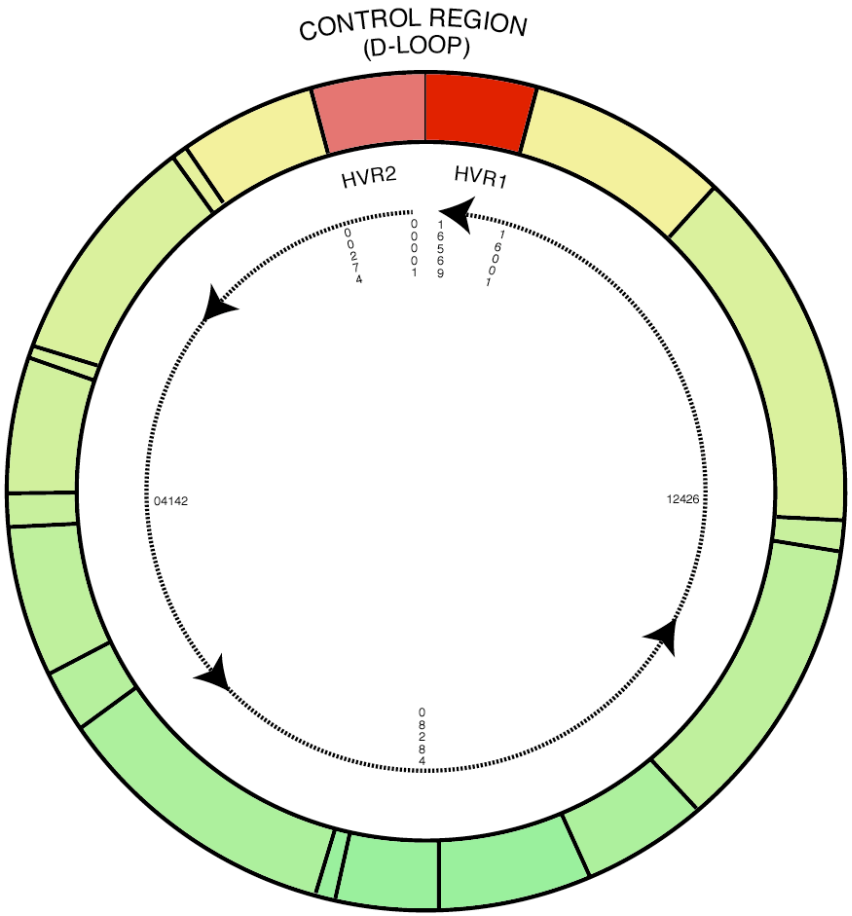
Your mtDNA traces back to your great grandmother from Mexico and ultimately to a woman that lived in Africa over 100,000 years ago that we call **Mitochondrial Eve**.



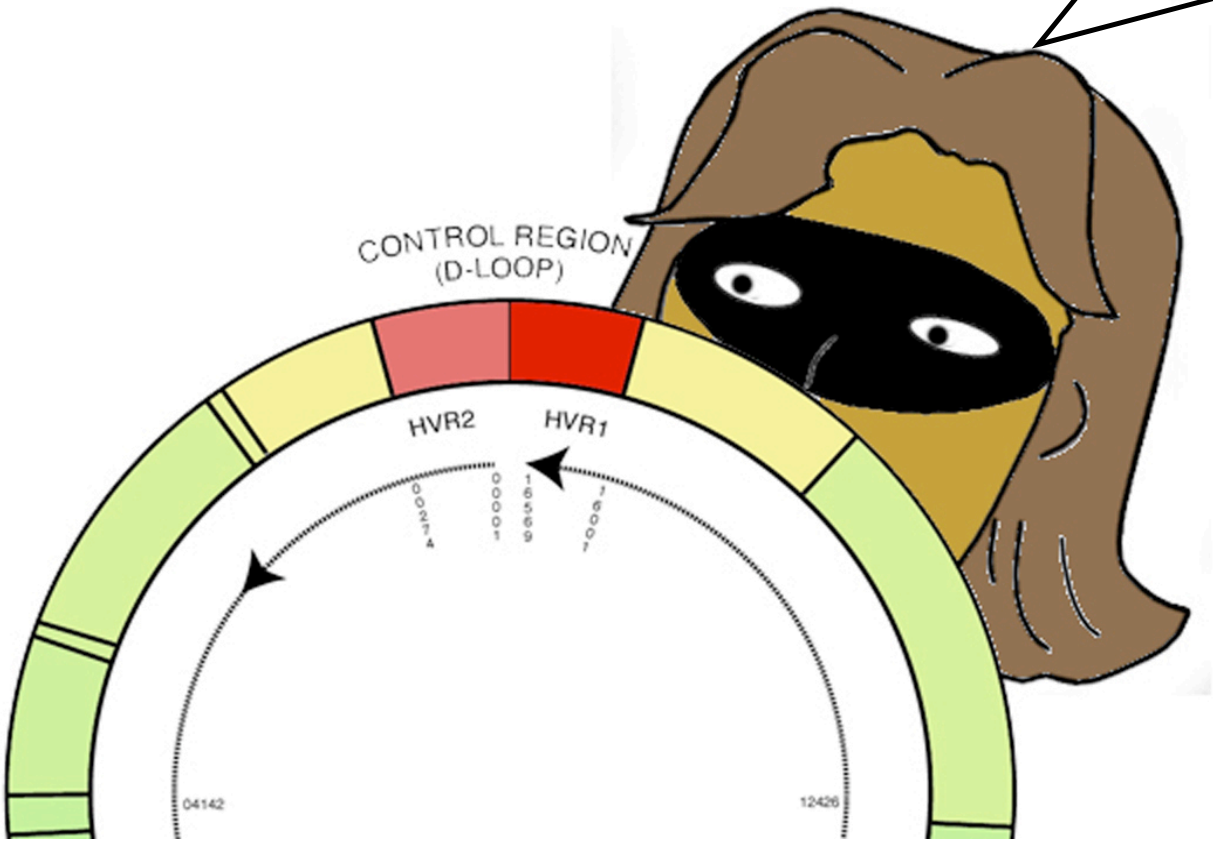
In contrast to the Y-chromosome, mitochondrial DNA is not found in the cell's nucleus. Instead it occurs within the cellular cytoplasm in tiny organelles called **mitochondria**, which are the powerhouses of cellular activity.

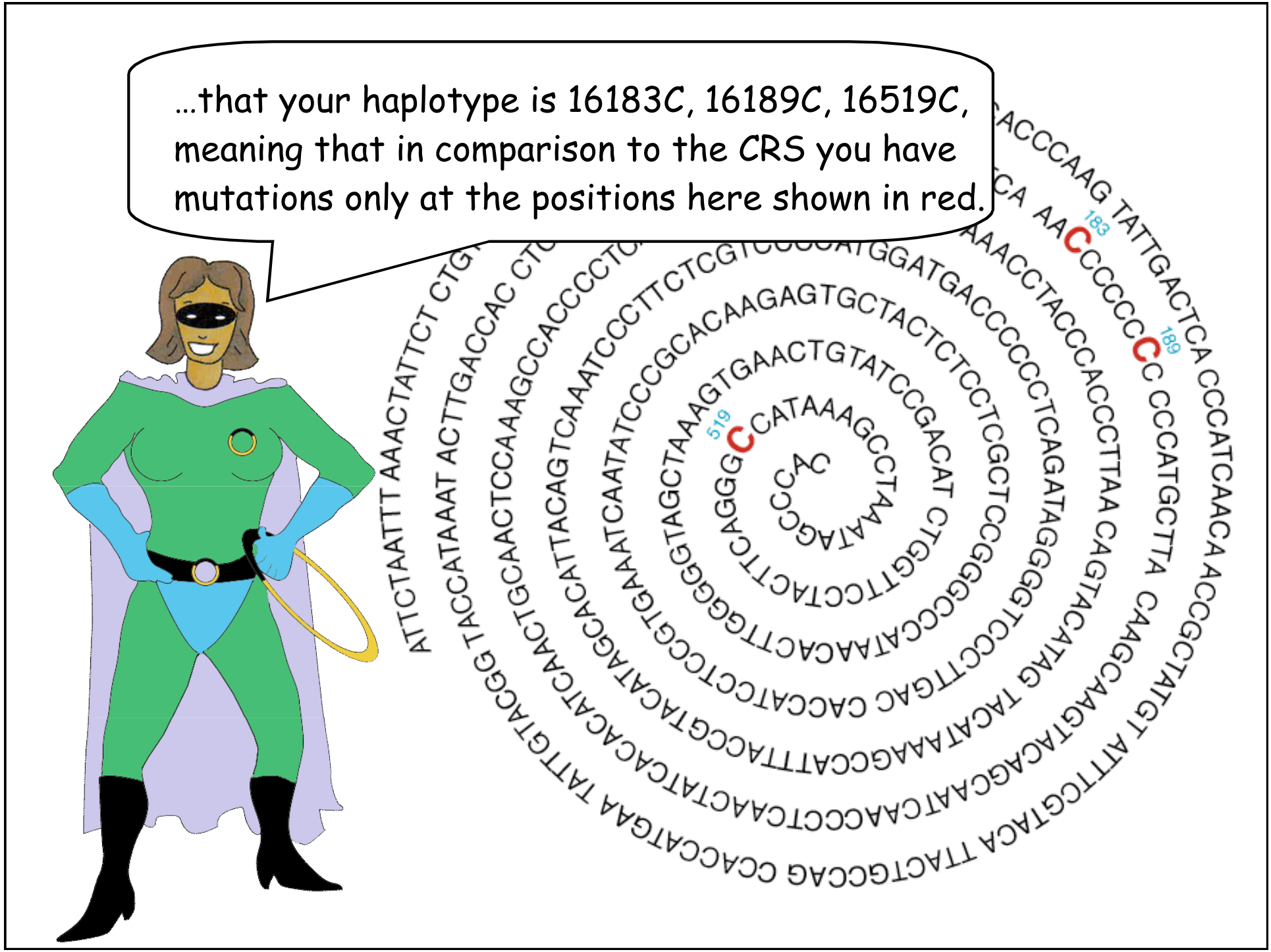


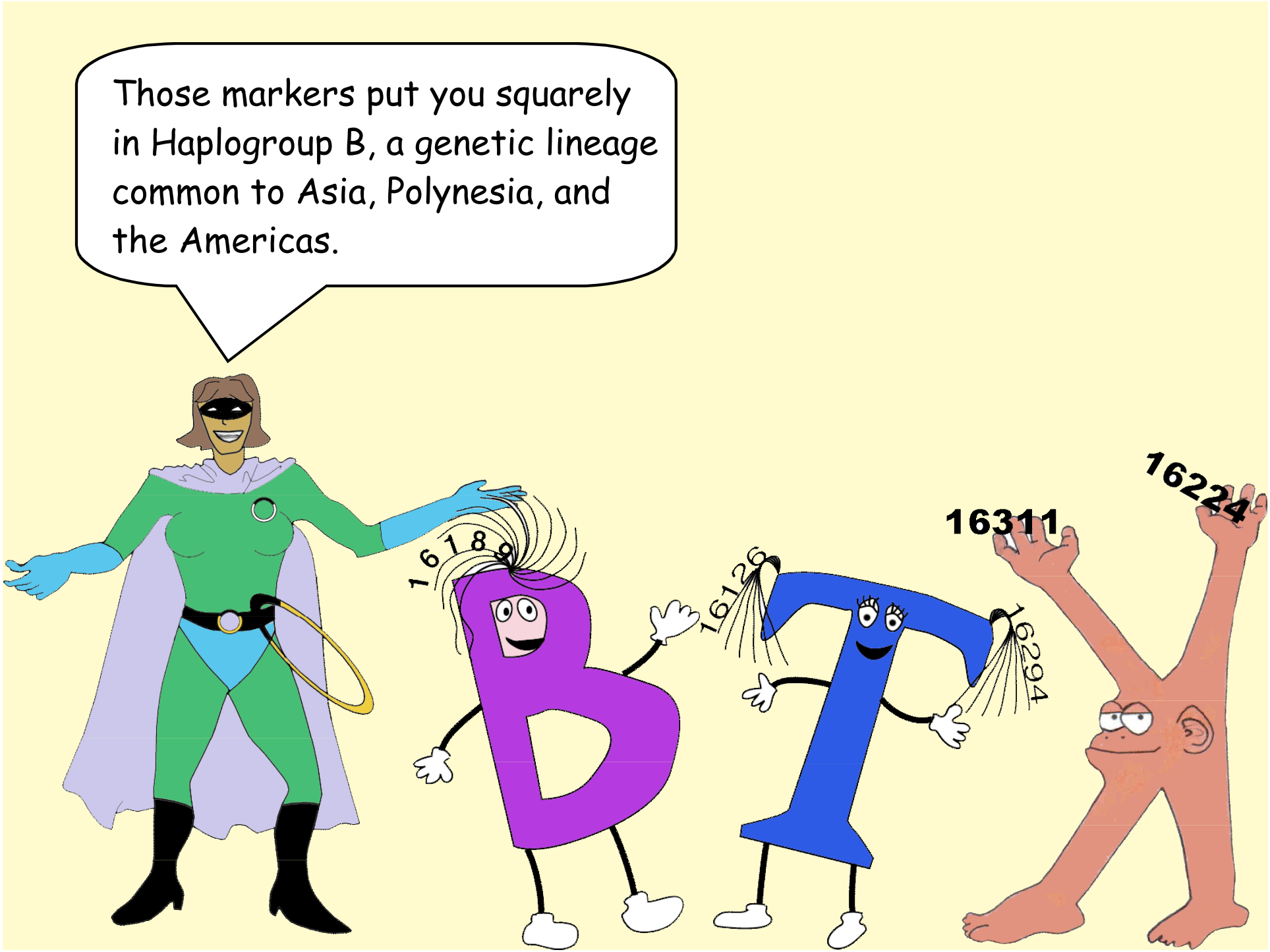
The basic form of mtDNA is a ring consisting of 16,569 base pairs. The entire sequence was first recorded at Cambridge University using an English woman's DNA, so is known as the **Cambridge Reference Sequence (CRS).** We use the CRS as a standard baseline and look for differences between it and <u>your</u> mitochondrial sequence.



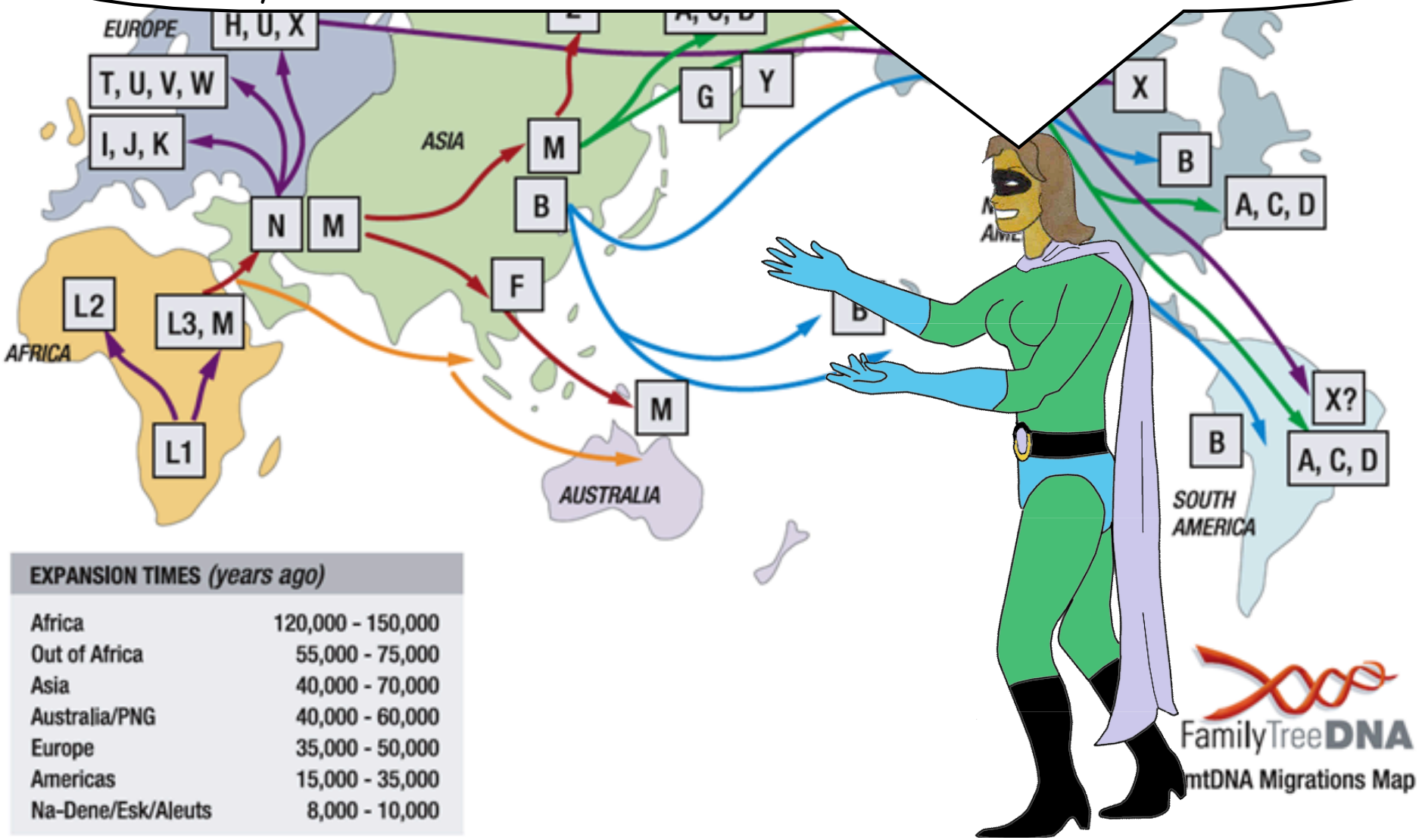
Actually we examined only a portion of the total CRS sequence called the **Hypervariable Region 1 (HVR1)** in the control region. We looked at every base pair between position 16001 and position 16569 in that section of your mtDNA and we found.....



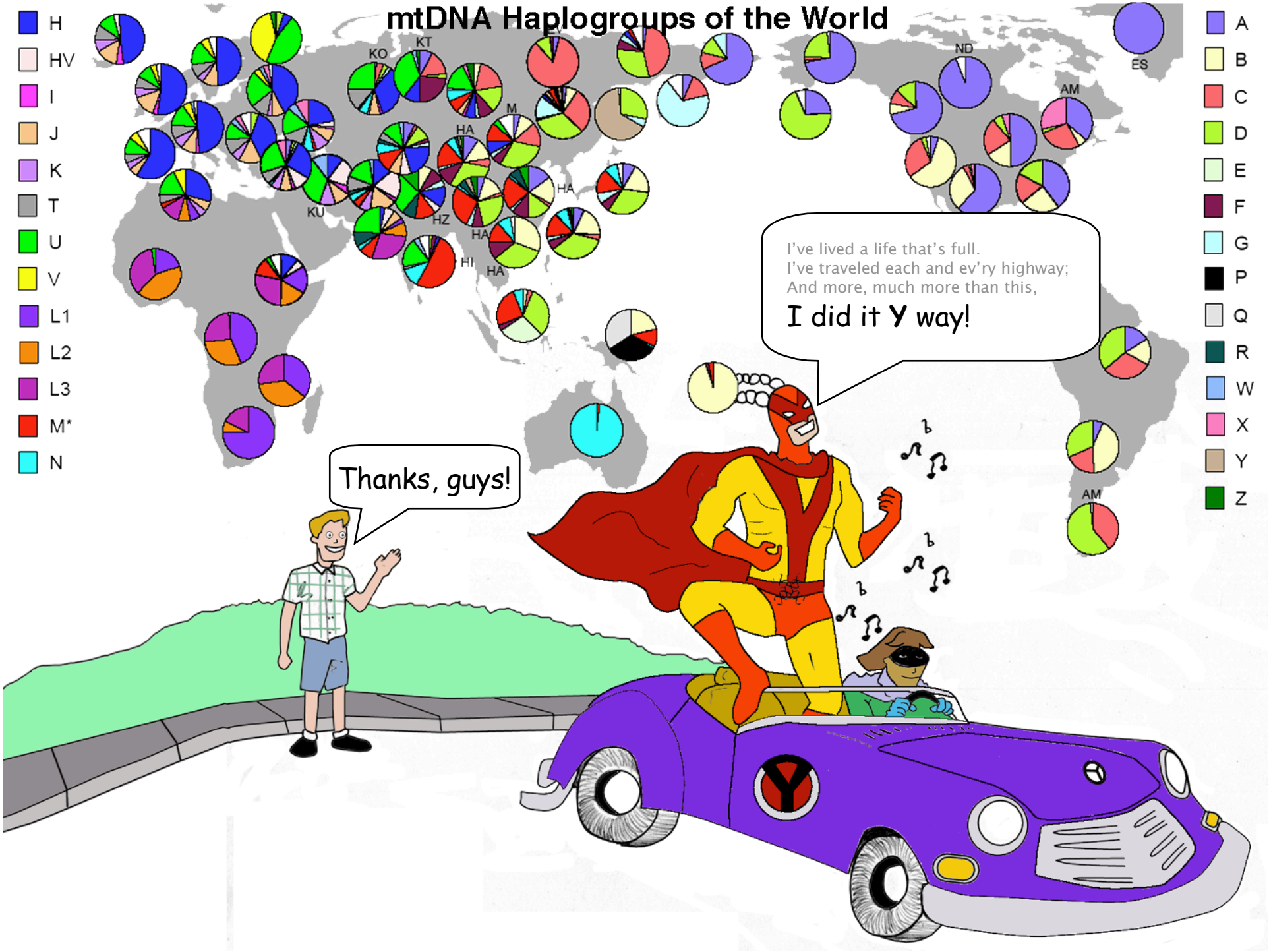

…that your haplotype is 16183C, 16189C, 16519C, meaning that in comparison to the CRS you have mutations only at the positions here shown in red.
183
189
519


Those markers put you squarely in Haplogroup B, a genetic lineage common to Asia, Polynesia, and the Americas.
16189
16126
16294
16311
16224

And that all fits. Your great grandmother, Maria, was from Mexico but everyone said that she had an Asian or Polynesia look about her. Her maternal ancestors could have arrived in Mexico in several ways - 10 to 20,000 years ago as one of the early Paleoindian colonists to the New World - or as much more recent immigrant from Asia or Polynesia.



EXPANSION TIMES *(years ago)*	
Africa	120,000 - 150,000
Out of Africa	55,000 - 75,000
Asia	40,000 - 70,000
Australia/PNG	40,000 - 60,000
Europe	35,000 - 50,000
Americas	15,000 - 35,000
Na-Dene/Esk/Aleuts	8,000 - 10,000


mtDNA Haplogroups of the World
H
HV
I
J
K
T
U
V
L1
L2
L3
M*
N
A
B
C
D
E
F
G
P
Q
R
W
X
Y
Z
KO
KT
M
HA
KU
HZ
HI
ND
AM
ES
I've lived a life that's full.
I've traveled each and ev'ry highway;
And more, much more than this,
I did it Y way!
Thanks, guys!

Additional Resources

BOOKS (Introductory)

Fitzpatrick, Colleen and Andrew Yeiser
2005 *DNA & Genealogy*. Rice Book Press.

Cavalli-Sforza, Luigi Luca
2001 *Genes, Peoples, and Language*. University of California Press, Berkeley.

Olson, Steve
2002 *Mapping Human History: Genes, Race, and Our Common Origins*.
Houghton Mifflin Company, Boston.

Oppenheimer, Stephen
2004 *The Real Eve: Modern Man's Journey Out of Africa*. Carroll & Graf.

Smolenyak, Megan and Ann Turner
2003 *Trace Your Roots with DNA: Using Genetic Tests to Explore Your Family Tree.*
Rodale Books, N.Y.

Sykes, Bryan
2001 *The Seven Daughters of Eve: The Science That Reveals Our Genetic Ancestry.* W.W. Norton & Company, N.Y.

Wells, Spencer
2002 *The Journey of Man: A Genetic Odyssey*. Princeton University Press, Princeton.

BOOKS (Technical)

Jobling, Mark A., Mathew Hurles, and Chris Tyler-Smith
2004 *Human Evolutionary Genetics: Origins, Peoples and Disease.* Garland Science, N.Y.

Renfrew, Colin and Katie Boyle, eds
2000 *Archaeogenetics: DNA and the Population Prehistory of Europe.* McDonald Institute Monographs, Oxford.

DVDs

The Journey of Man. PBS Home Video.
The Real Eve. Discovery Channel Video.
Race: The Power of an Illusion. PBS Home Video.

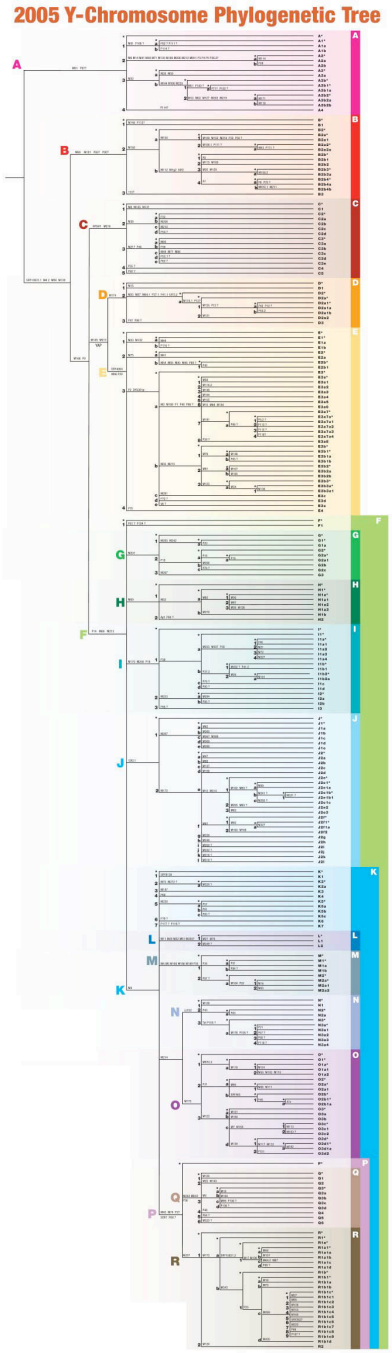
WEB SITES

Family Tree DNA
http://www.familytreedna.com
Genographic Project
https://www3.nationalgeographic.com/genographic/index.html
African Ancestry
http://africanancestry.com
DNA Heritage
http://www.dnaheritage.com
Journey of Mankind
http://www.bradshawfoundation.com/journey
MitoSearch
http://www.mitosearch.org
Oxford Ancestors
http://www.oxfordancestors.com
A (Personal) mtDNA View of the Peopling of the World by *Homo sapiens*
http://www.mcdonald.cam.ac.uk/genetics/mtDNAworld/one.html
Roots Project
http://www.uml.edu/dept/biology/rootsproject
Sorenson Molecular Molecular Genealogy Foundation
http://www.uml.edu/dept/biology/rootsproject
Yseearch
http://www.ysearch.org

Appendix 1

SNPs Made Simple

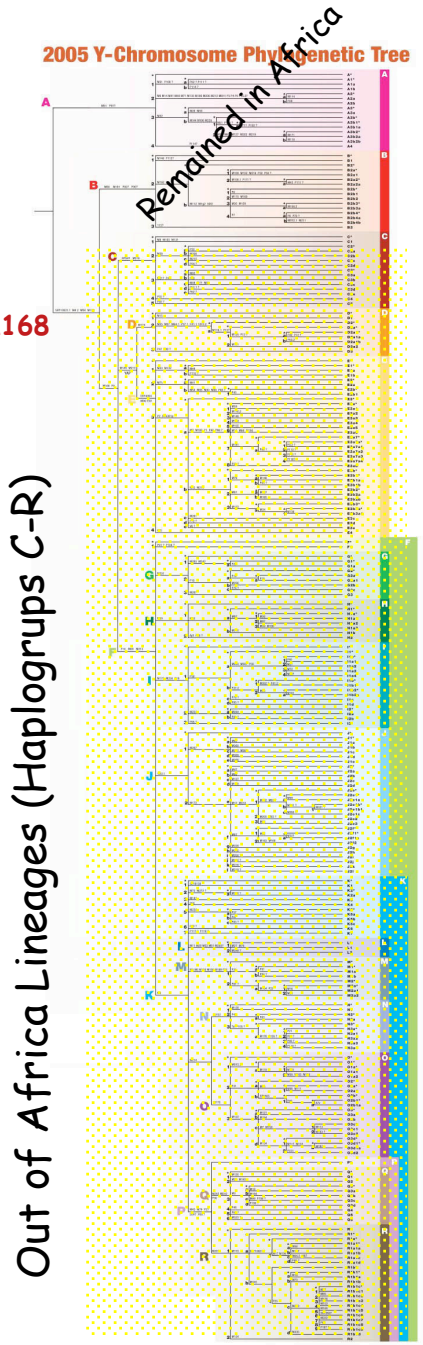
The ancestor of all *Homo sapiens* men alive today lived in Africa more than 100,000 years ago. Geneticists call him **Y-Adam** and all men carry his genetic marker.



FamilyTreeDNA

Check it out! M168 is my mark. It separates my descendants (yellow) from those that stayed in Africa.

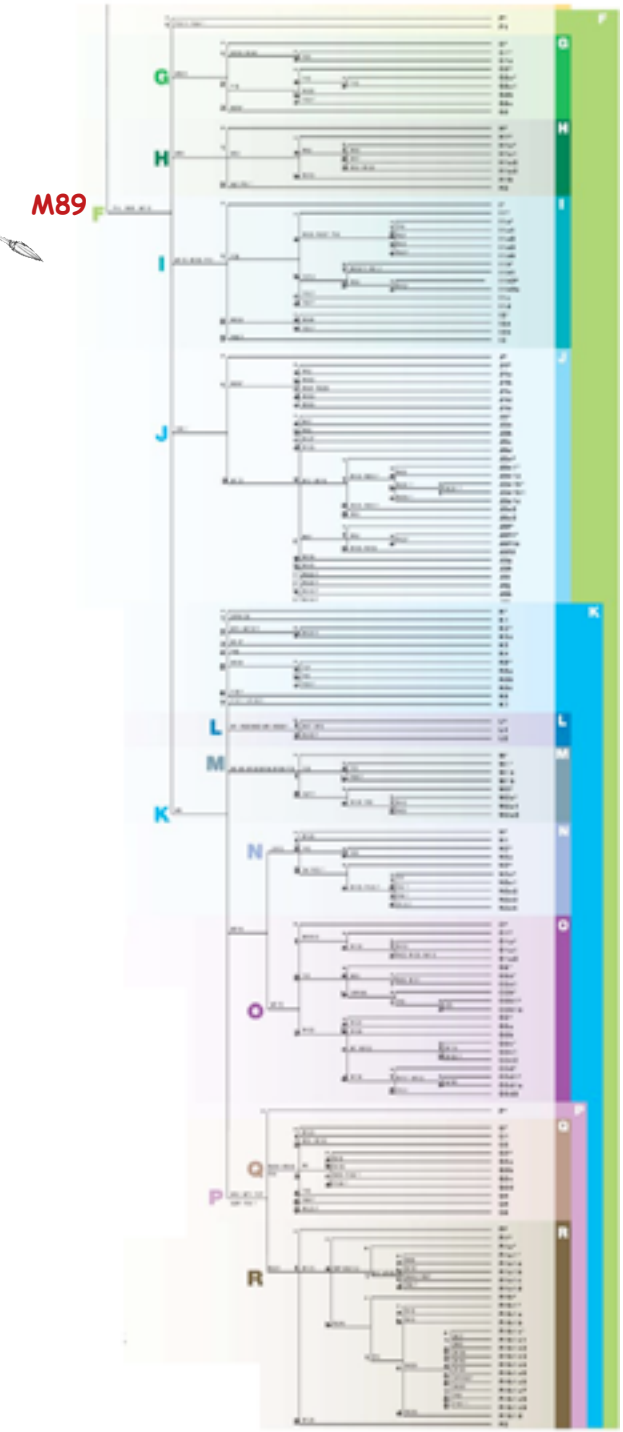
M168



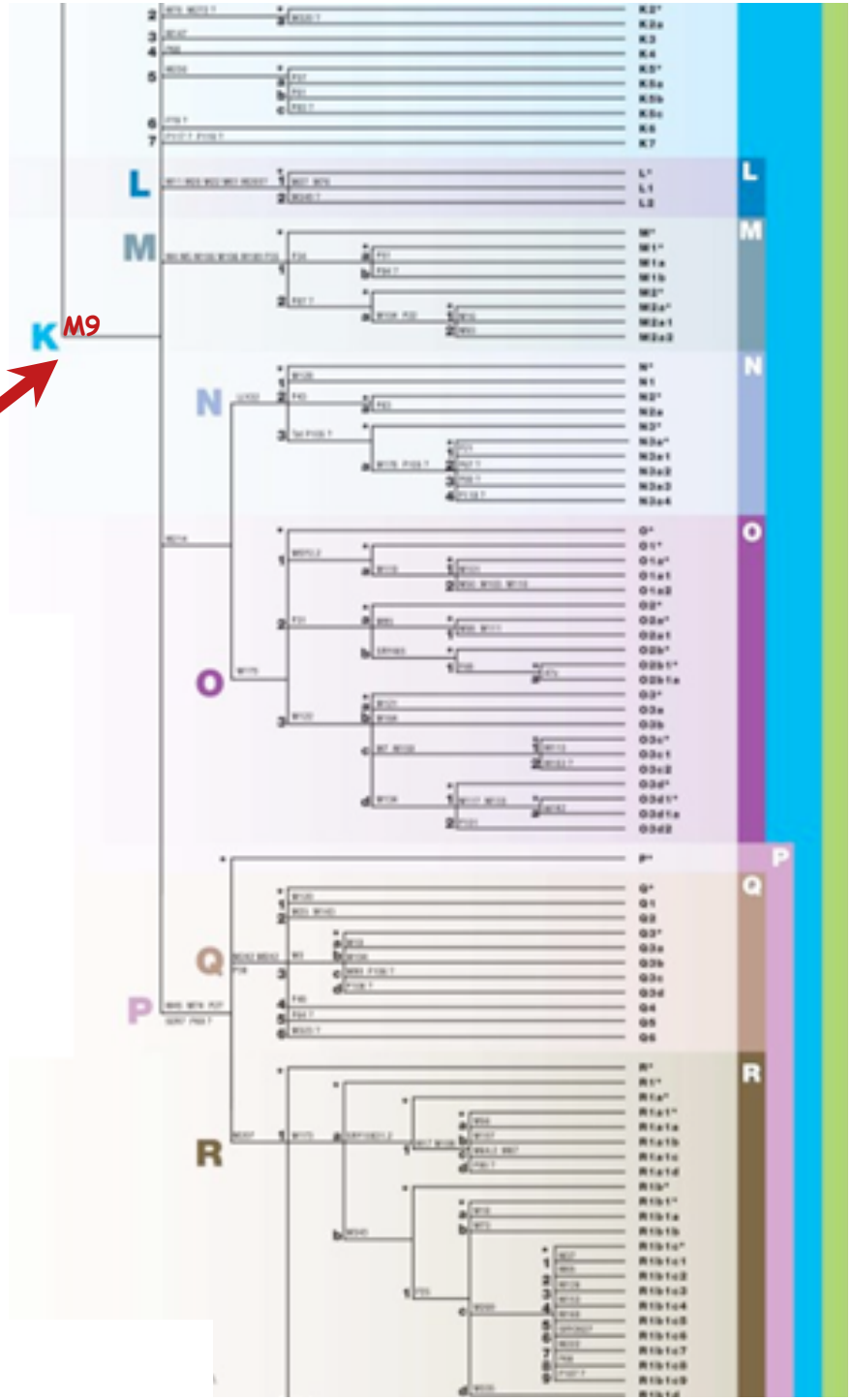
Somewhere around 60,000 years ago a mutation (SNP) called **M168** occurred on the Y-chromosome of a man in this Afican population. This man, who we will call **Out of Africa Adam**, was the ancestor of a branch of the African population that began to migrate out of Africa about 50,000 years ago, carrying with it the M168 marker of their great-great…grandfather. The M168 marker is carried by all living men, except the descendants of the two lineages that remained in Africa.

FamilyTreeDNA

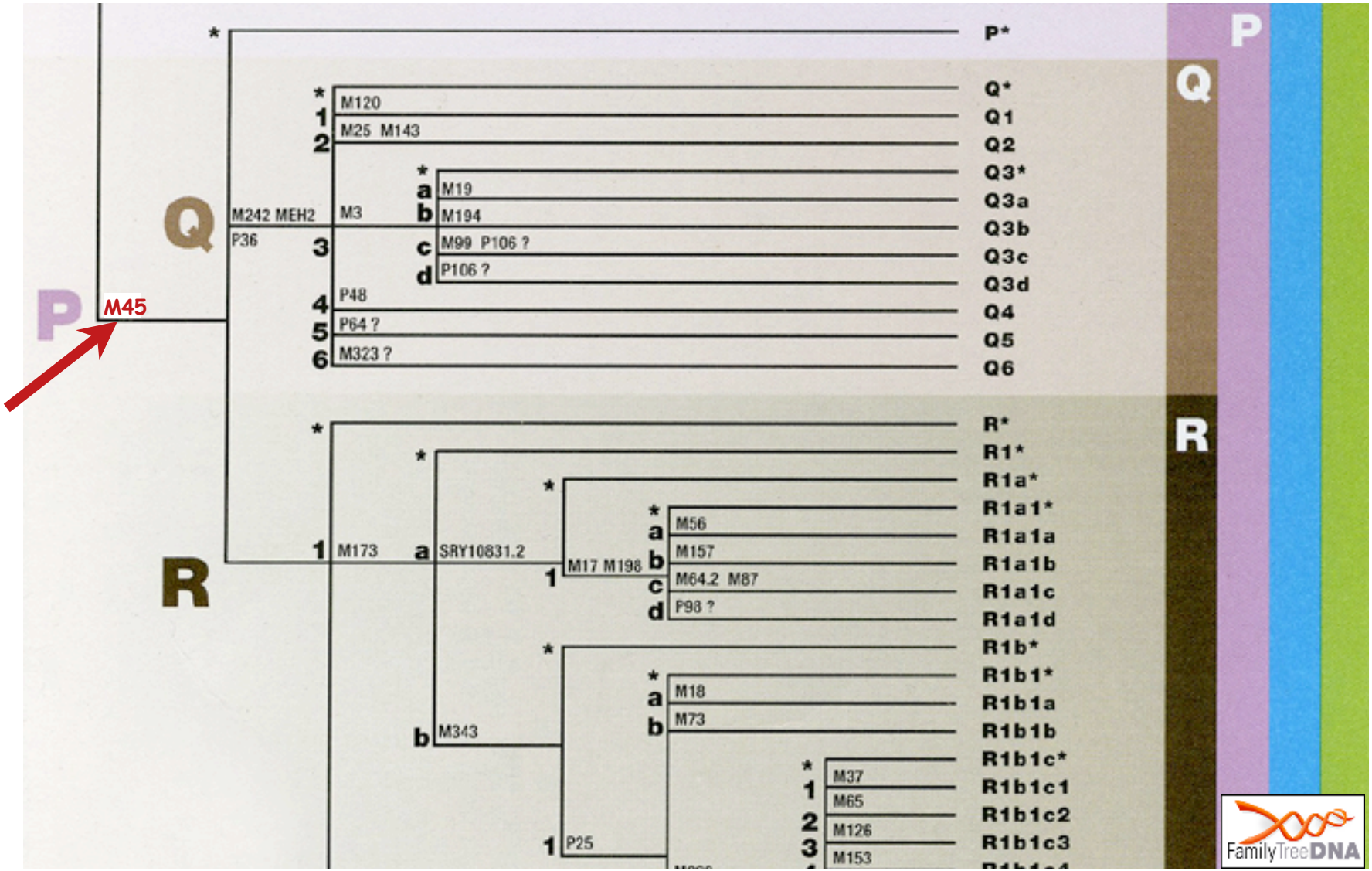
As the Out of Africa lineage migrated across northeastern Africa and the Arabian Peninsula another mutation occurred on the Y-chromosome of one of M168's male descendants. This marker, called **M89**, arose about 45,000 years ago, probably in modern-day Iraq and is carried by all Eurasian and Native American men (Haplogroups G through R).



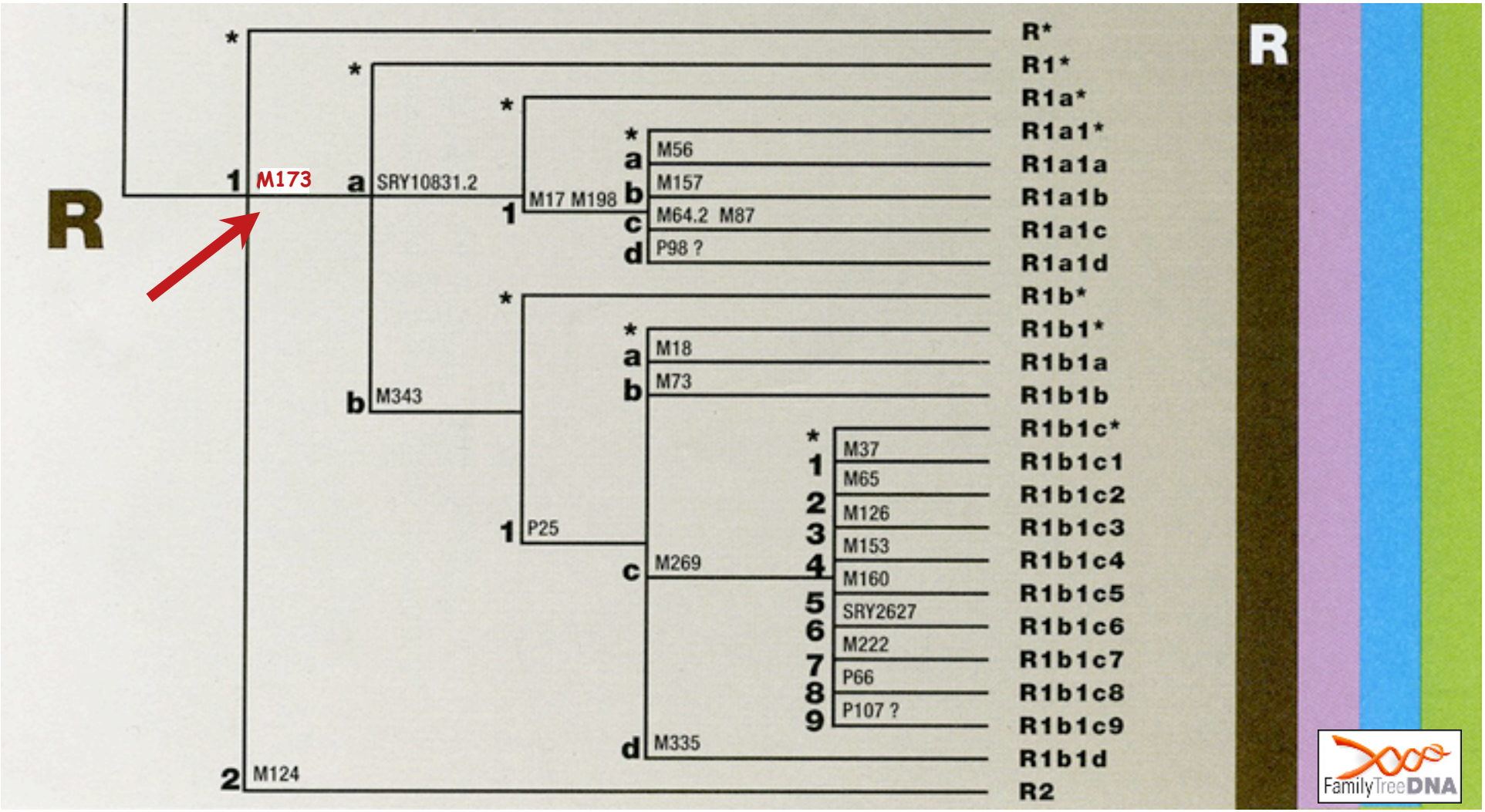
Some 5,000 years later in the vicinity of Iran another SNP occurred on the Y-chromosome of one of these M89 men. This marker, called **M9**, defines the main branch of the Eurasian clan and includes the haplogroups K through R.



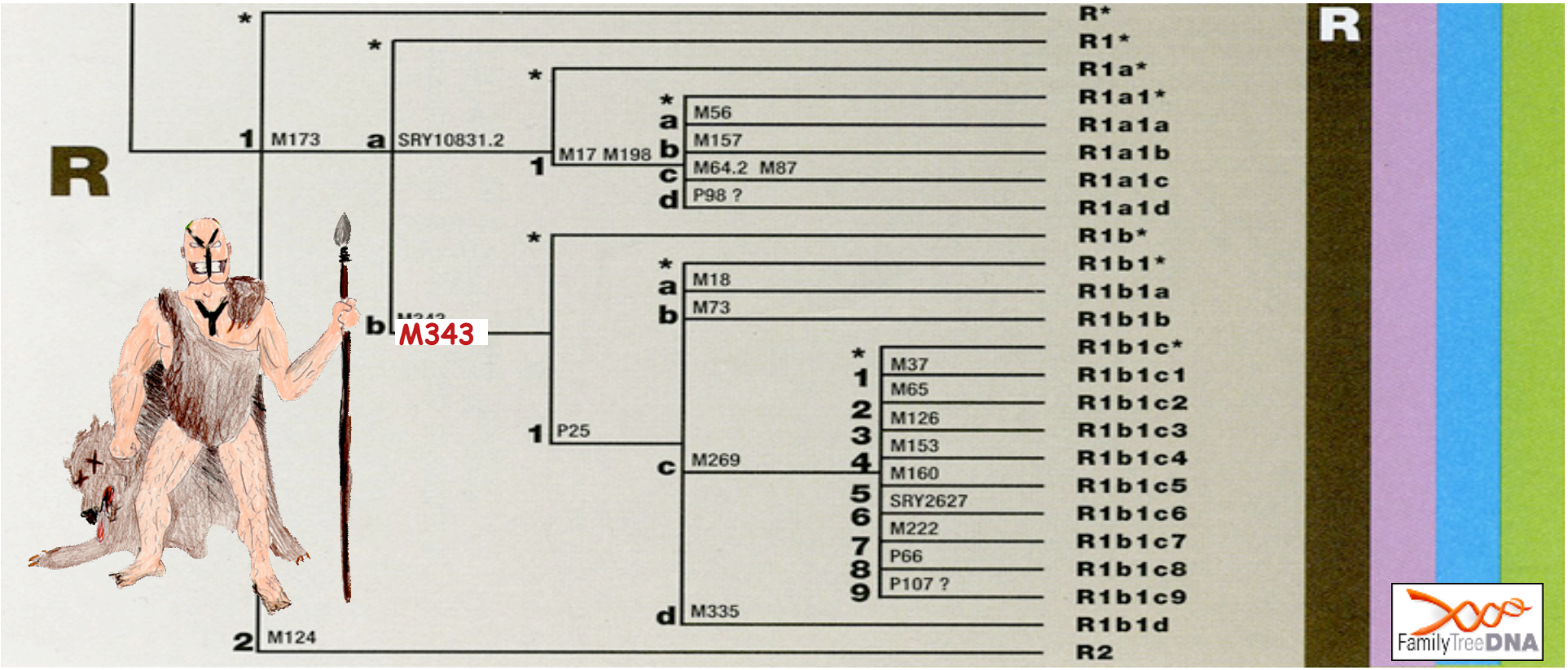
At the beginning of the Upper Paleolithic, about 35,000 years ago, one branch of the Eurasian clan moved into Central Asia in pursuit of the game animals that abounded there and begat yet another SNP marker, **M45**. This marker is carried on the Y-chromosomes of men of both the R haplogroup (principally European and Indian) and the Q haplogroup (Native American).



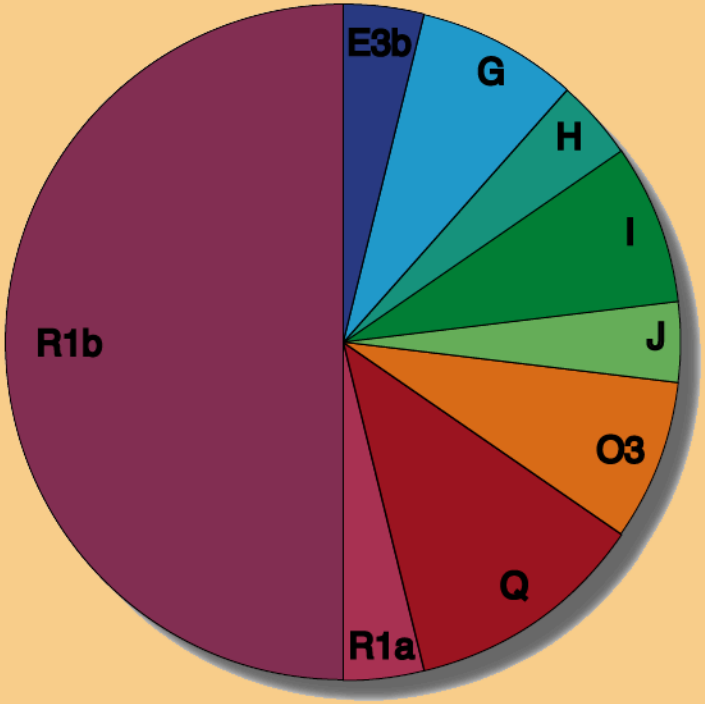
The SNP that marks the entrance of your Upper Paleolithic ancestors into Europe about 30,000 years ago, is **M173**. You share this marker with over 40% of European men as well as many men from Iran and India, all of whom belong to haplogroup R.



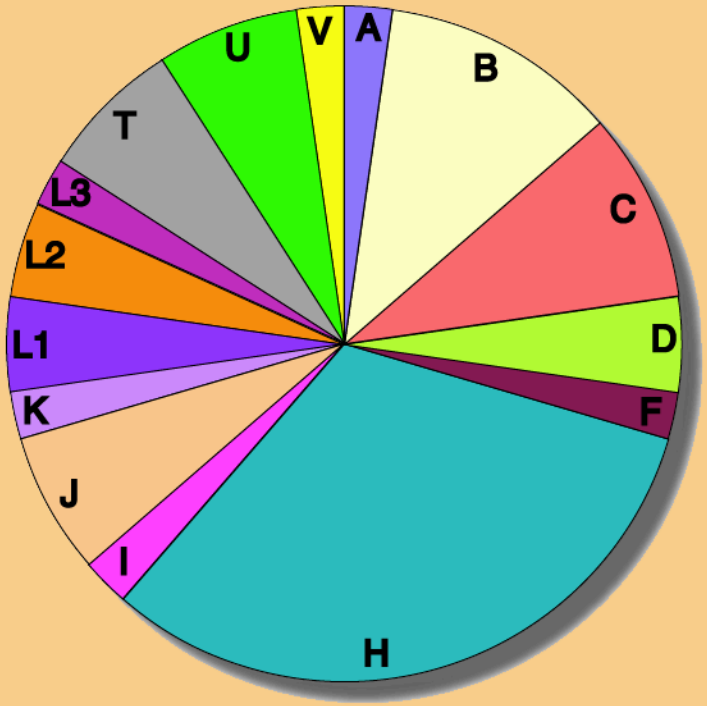
Finally, the marker that defines your membership in sub-haplogroup R1b is the SNP called **M343**. This mutation probably occurred around 20,000 years ago in a western European population. Somewhere around 13,000 BC at the end of Last Glacial Maximum R1b men carrying the M343 marker are known to have expanded northward from Iberia to recolonize central and northern Europe. The R1b lineage dominates modern European populations and 98% of your male ancestors from western Irish are from this group.



Appendix 2
Samples from the Exhibition

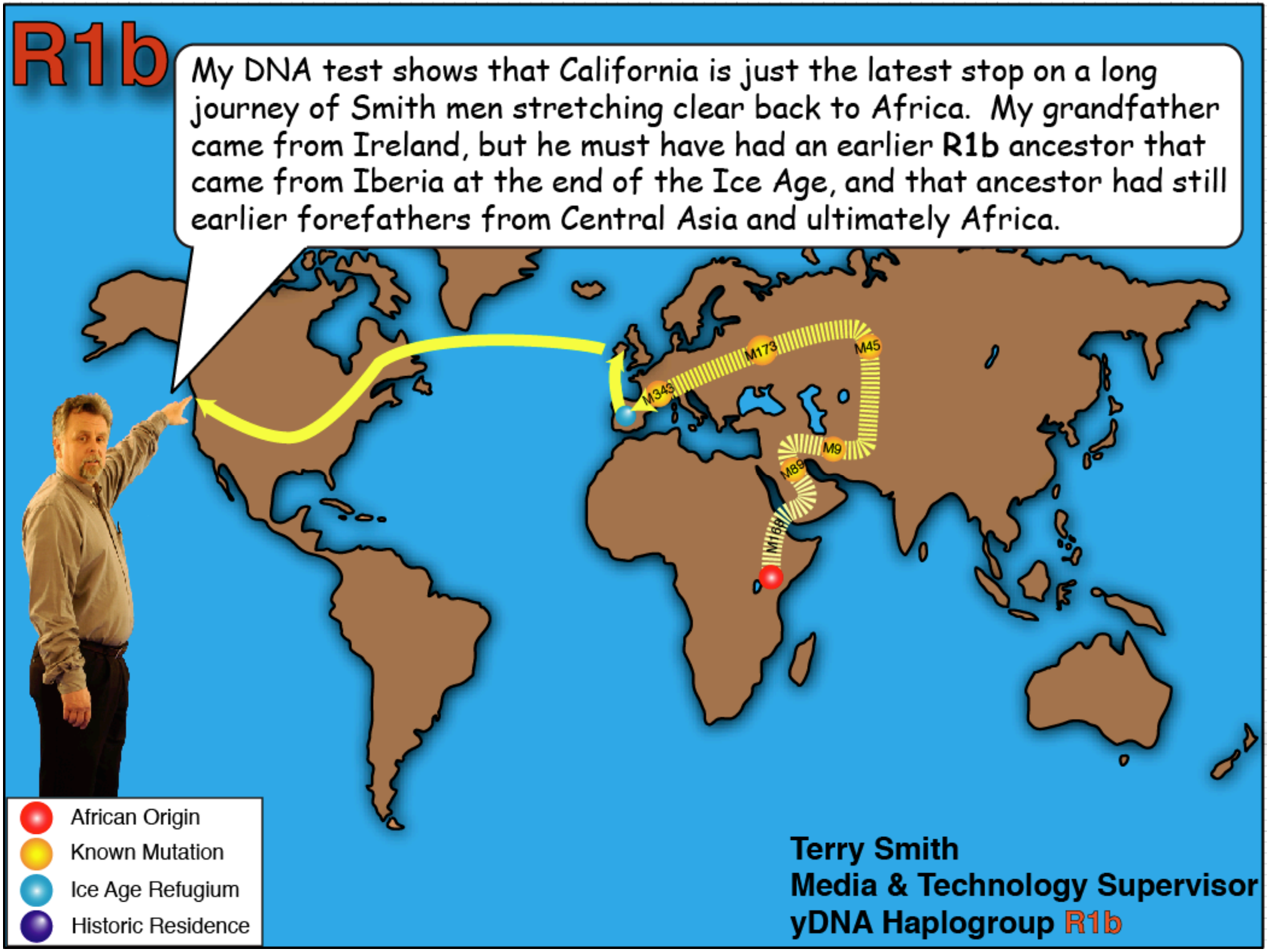


Y-chromosome Haplogroups



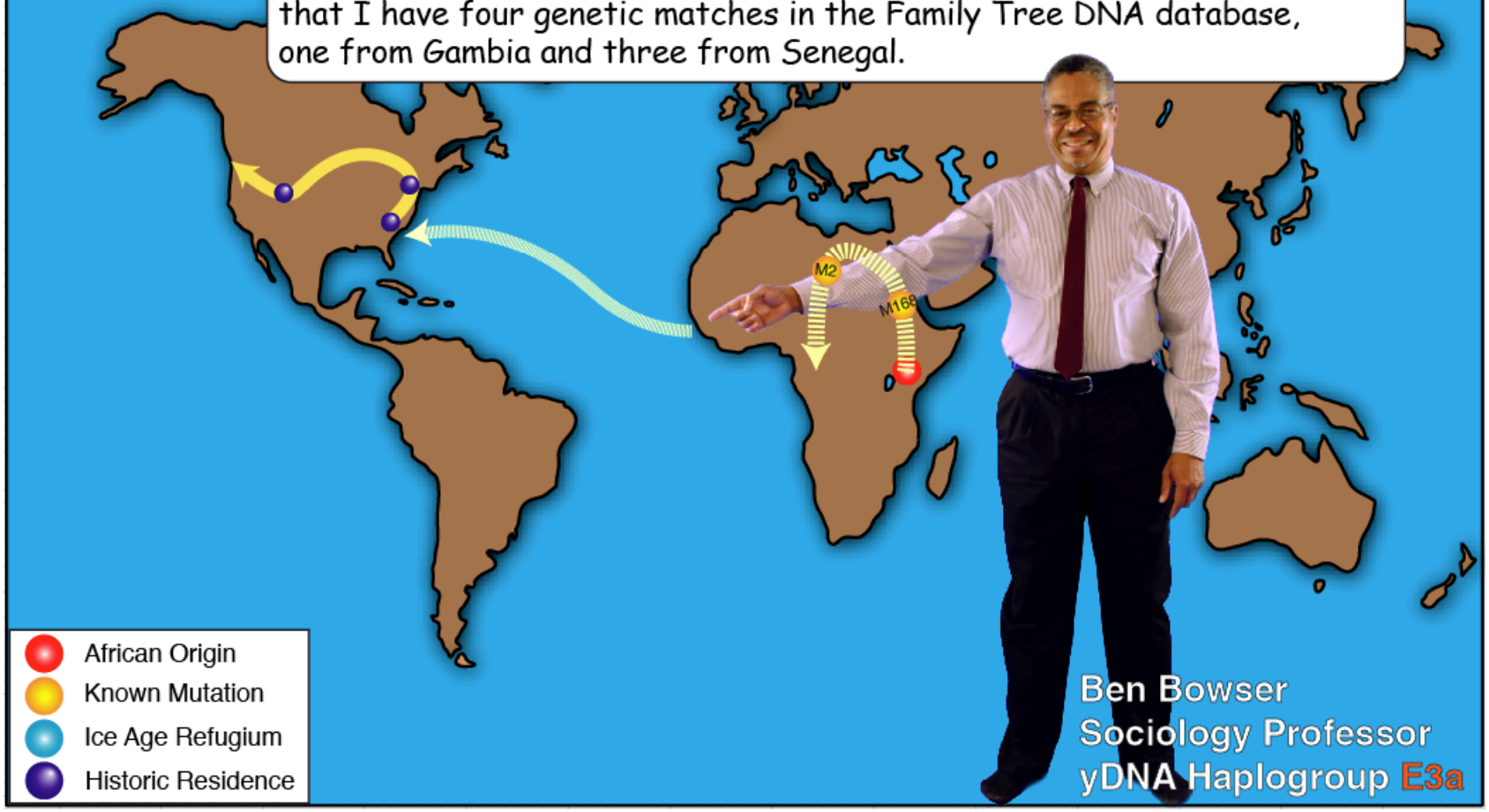
mtDNA Haplogroups

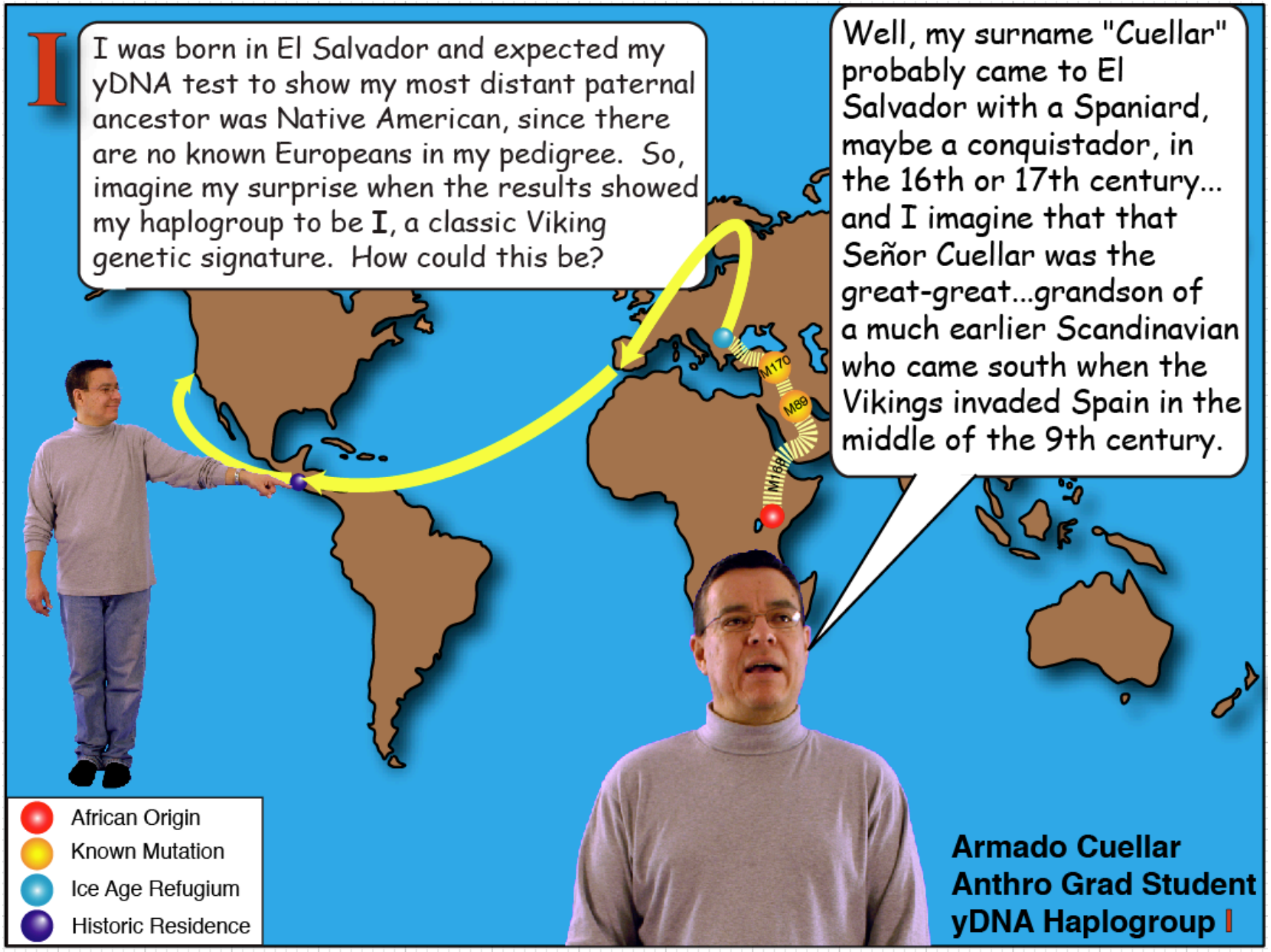
Since initiating the *Immigrants All* project in November, 2004 we have received the results of 73 DNA tests from the Family Tree DNA lab (27 12 marker Y-chromosome tests and 44 HVR1 mtDNA tests). The haplogroup assignments from these tests are displayed in percentage form in the graphs above. Thirteen examples of the individual historic and genetic migration trails from a total of 32 participants highlighted in the exhibition appear on the pages that follow. The testing of 67 additional participants has not been completed at the time of the opening of the exhibition on February 24, 2006. The picture reported here will surely change as these results come in and we expand our study during 2006-2007.


R1b
My DNA test shows that California is just the latest stop on a long journey of Smith men stretching clear back to Africa. My grandfather came from Ireland, but he must have had an earlier **R1b** ancestor that came from Iberia at the end of the Ice Age, and that ancestor had still earlier forefathers from Central Asia and ultimately Africa.
M173
M45
M343
M9
M89
M168
African Origin
Known Mutation
Ice Age Refugium
Historic Residence
Terry Smith
Media & Technology Supervisor
yDNA Haplogroup R1b

E3a

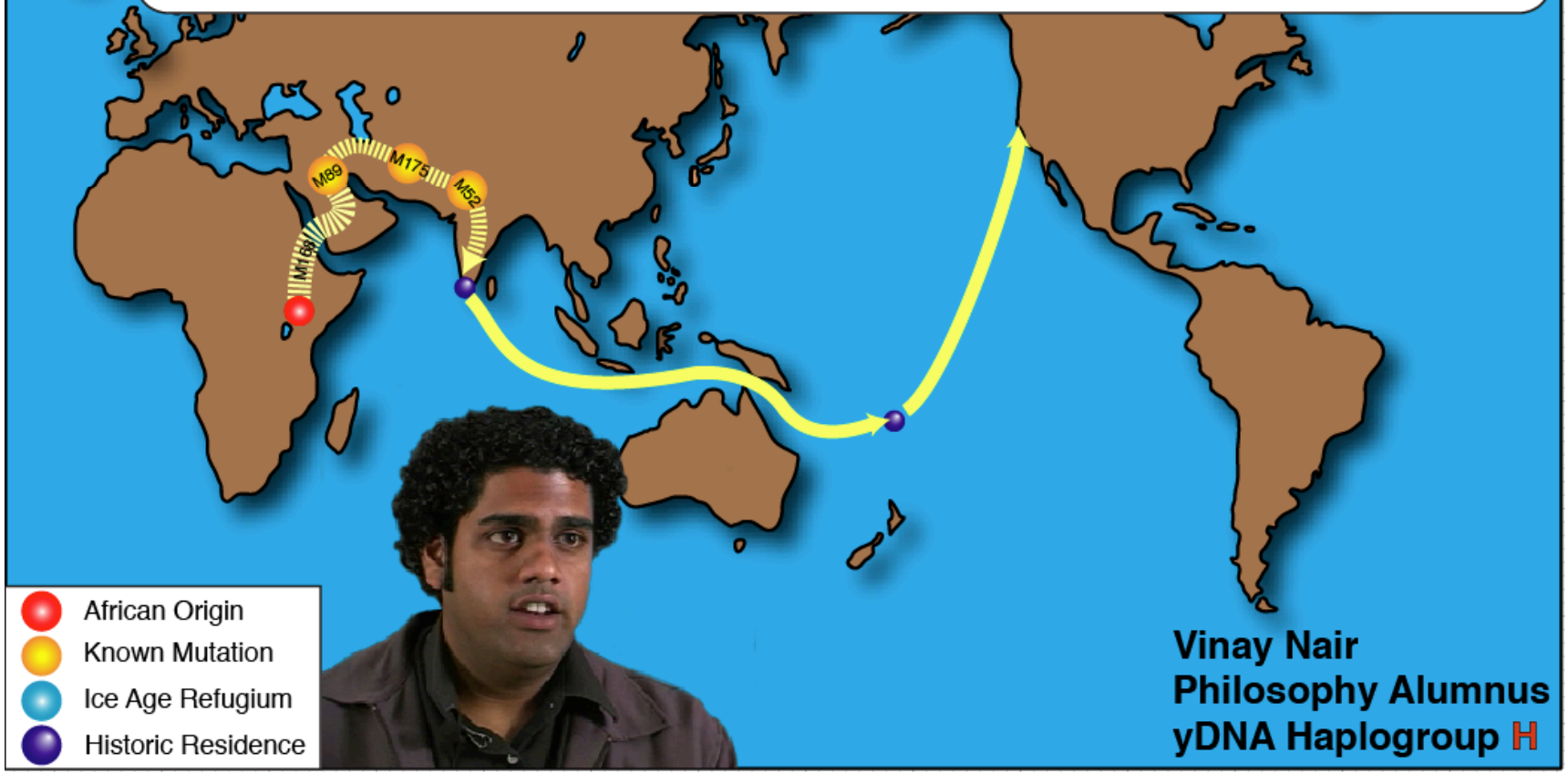
Bowsers of African ancestry can be traced back to 1650 in Virginia and then as freedmen after 1731 in coastal North Carolina. The Bowsers were major agitators for abolishing slavery; 34 Bowser men fought in the Civil War as Union soldiers and Mary Bowser was a maid for Robert E. Lee and an important Union spy. My father often spoke of a family oral history that we were from the area of Gambia and Gabore in West Africa. It is amazing to now learn that my yDNA haplogroup is **E3a** and to find that I have four genetic matches in the Family Tree DNA database, one from Gambia and three from Senegal.



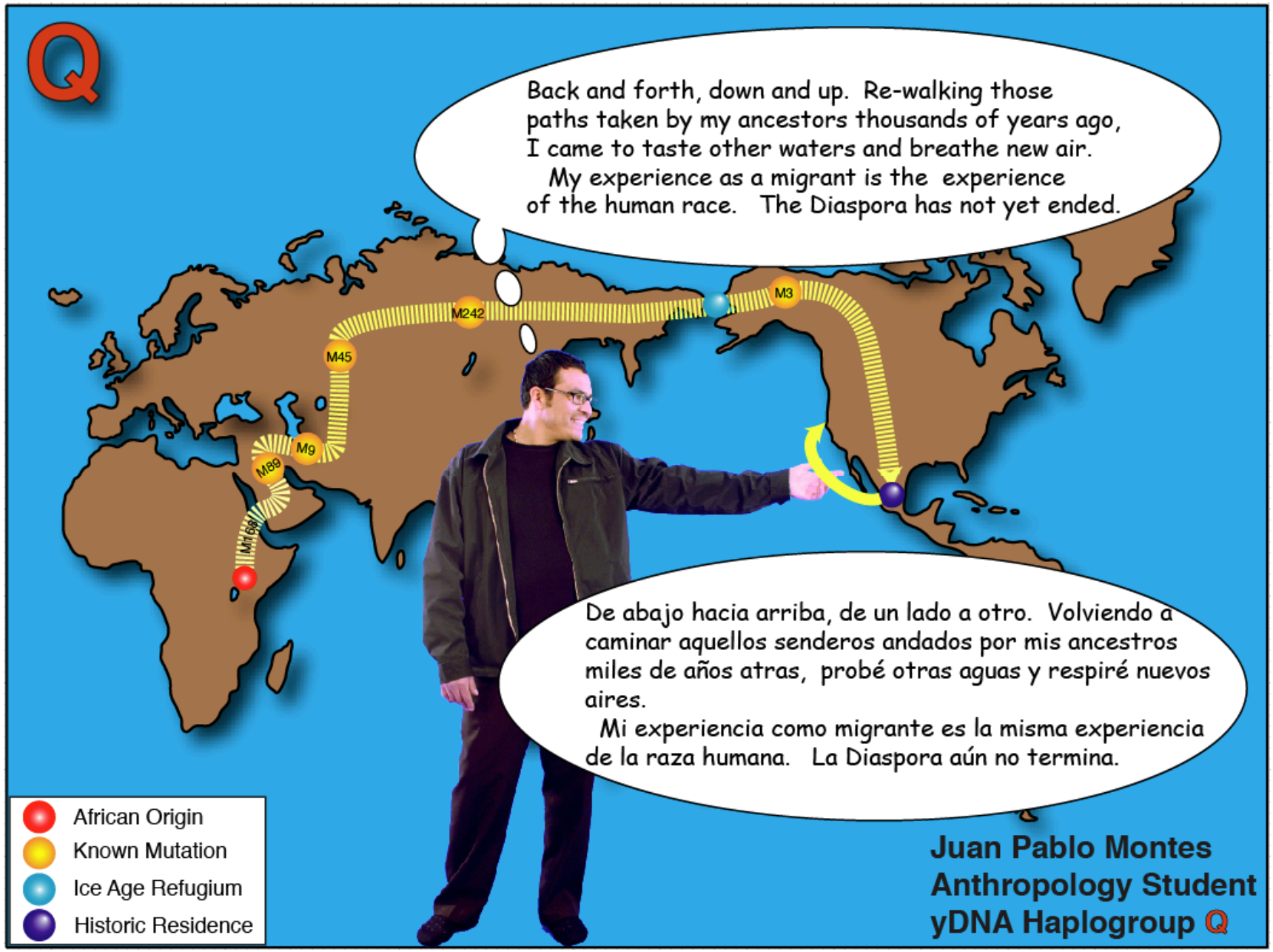

I
I was born in El Salvador and expected my yDNA test to show my most distant paternal ancestor was Native American, since there are no known Europeans in my pedigree. So, imagine my surprise when the results showed my haplogroup to be I, a classic Viking genetic signature. How could this be?
Well, my surname "Cuellar" probably came to El Salvador with a Spaniard, maybe a conquistador, in the 16th or 17th century... and I imagine that that Señor Cuellar was the great-great...grandson of a much earlier Scandinavian who came south when the Vikings invaded Spain in the middle of the 9th century.
M170
M89
M181
African Origin
Known Mutation
Ice Age Refugium
Historic Residence
Armado Cuellar
Anthro Grad Student
yDNA Haplogroup I

H

My parents and grandparents were born in Fiji, but my great grandparents came from southern India from the state of Kerala. I was interested to find that I am from the y-chromosome haplogroup H. The H haplogroup means that my ancestors were one of the earliest groups of humans to settle in India after the Out of Africa migration, somewhere around 60,000 years ago. Through subsequent migrations and invasions into India, the majority of this haplogroup was driven further and further into southern India and Sri Lanka.

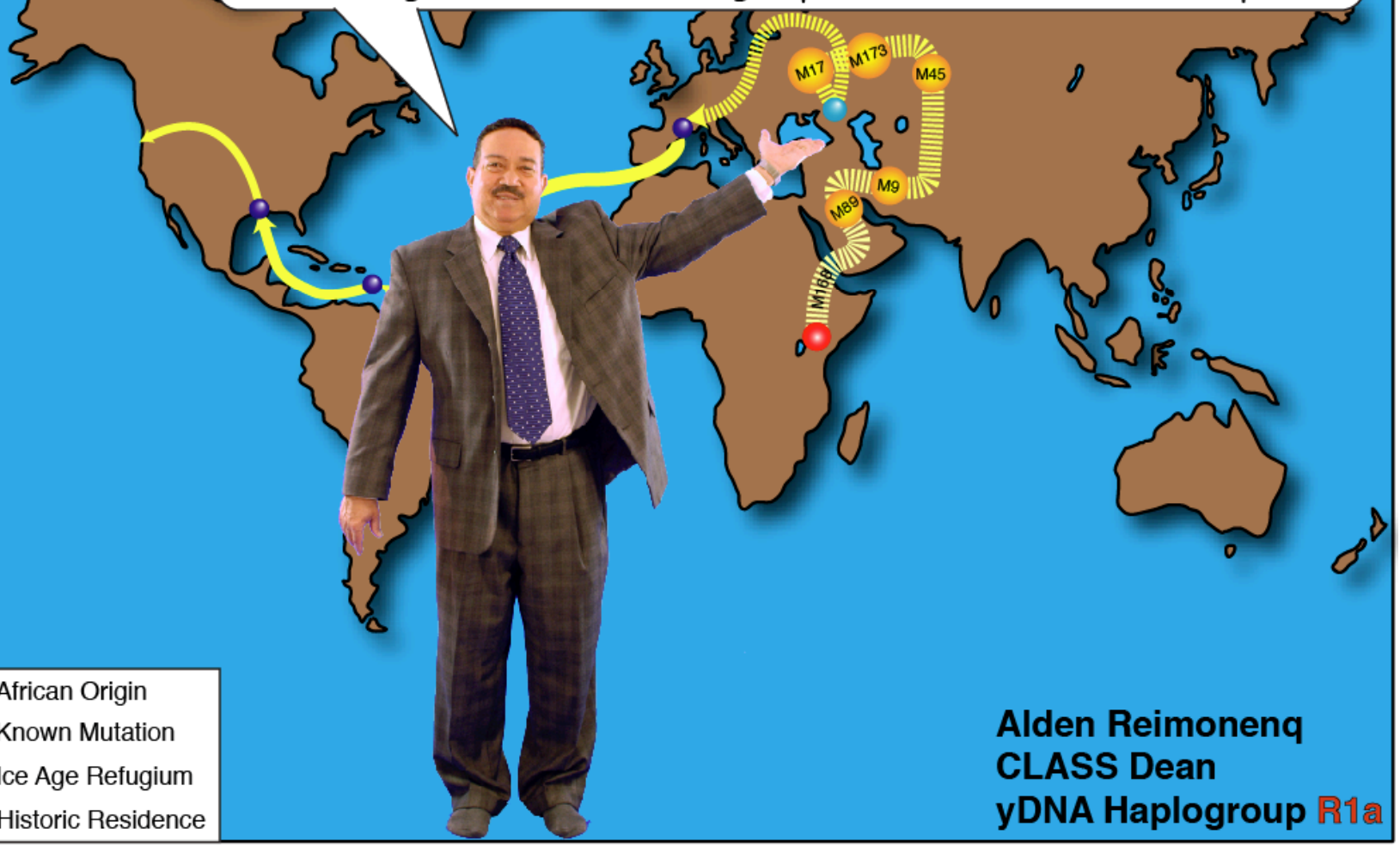


Vinay Nair
Philosophy Alumnus
yDNA Haplogroup H


Q
Back and forth, down and up. Re-walking those paths taken by my ancestors thousands of years ago, I came to taste other waters and breathe new air.
My experience as a migrant is the experience of the human race. The Diaspora has not yet ended.
M242
M3
M45
M9
M89
M168
De abajo hacia arriba, de un lado a otro. Volviendo a caminar aquellos senderos andados por mis ancestros miles de años atras, probé otras aguas y respiré nuevos aires.
Mi experiencia como migrante es la misma experiencia de la raza humana. La Diaspora aún no termina.
African Origin
Known Mutation
Ice Age Refugium
Historic Residence
Juan Pablo Montes
Anthropology Student
yDNA Haplogroup Q

R1a

When I was growing up, my paternal grandfather used to tell us stories of his great grandfather, "The Russian," who had blue eyes and red hair. We have traced our family genealogy from southern France to the Caribbean island of Guadeloupe to New Orleans, where I was born. So hearing about "the Russian" was just family legend to us. Now, guess what? My paternal **R1a** DNA signature is a match to groups in Russia and Eastern Europe!!!



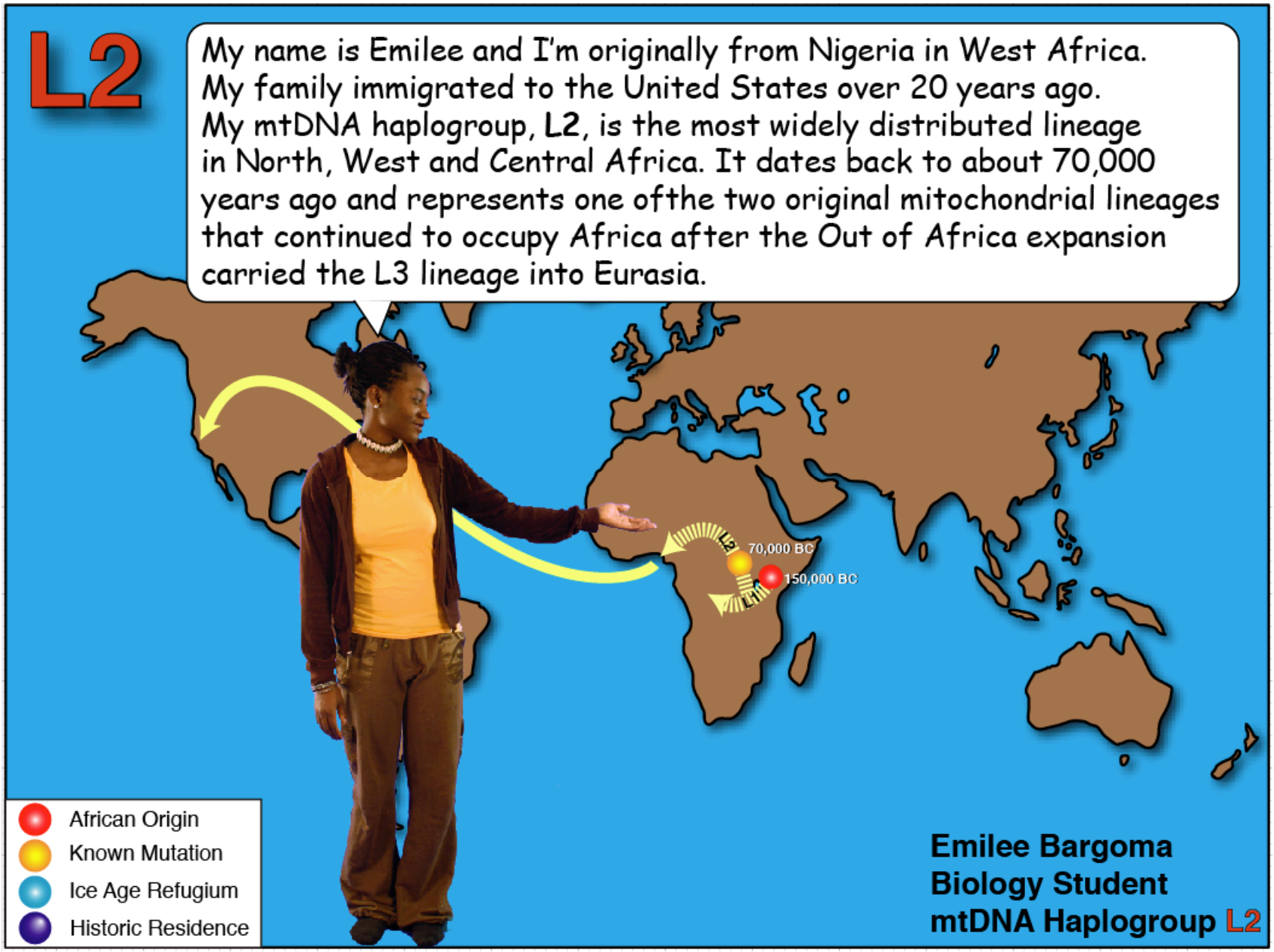
African Origin
Known Mutation
Ice Age Refugium
Historic Residence

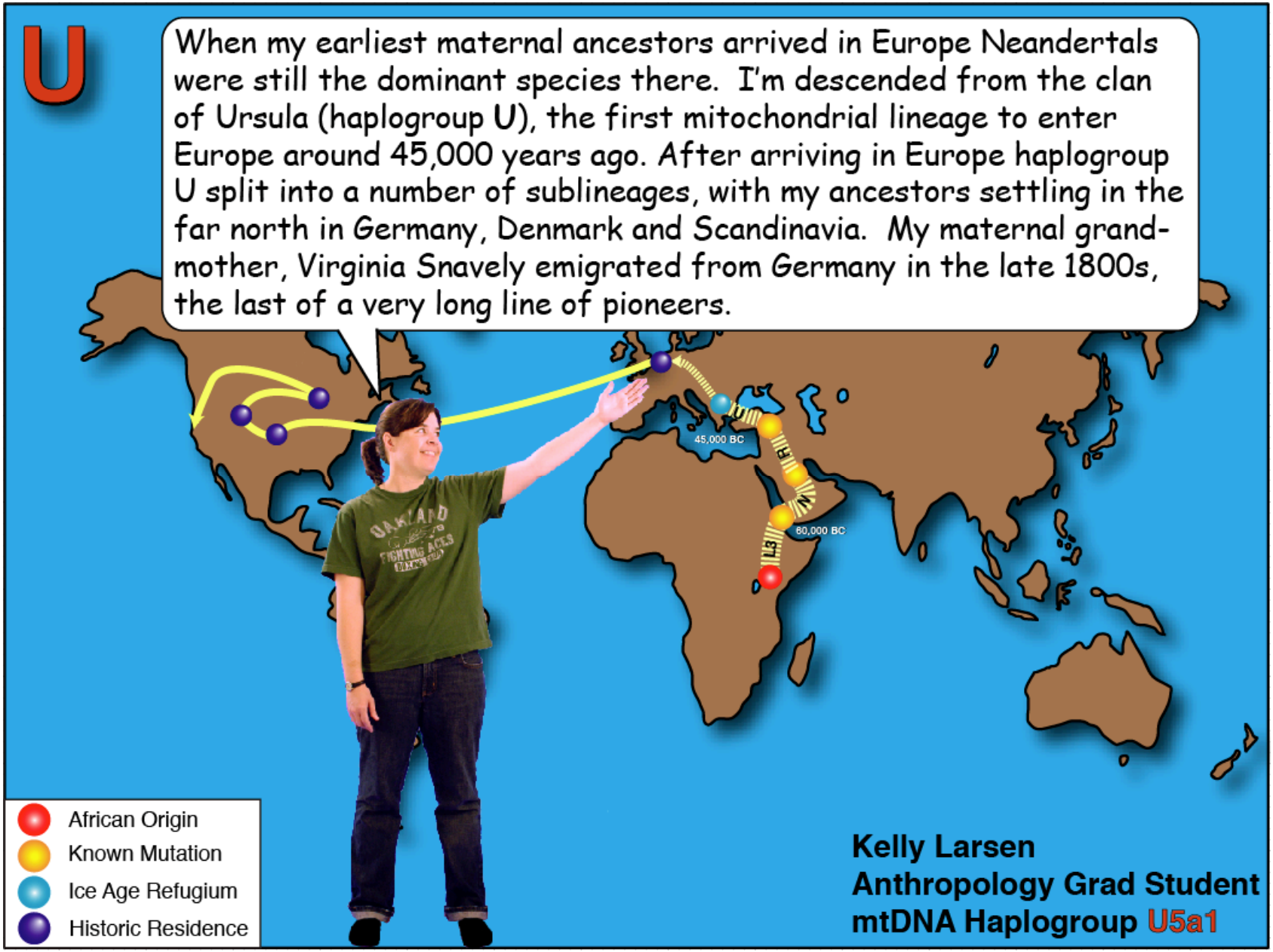
Alden Reimonenq
CLASS Dean
yDNA Haplogroup R1a

A

People always ask if I am Italian because of my last name, but I was born in a small village in Nayarit, Mexico. My family had lived in Mexico for at least three generations, but my father always said one of our forefathers came from Spain. I now know that that is likely true because my yDNA results are haplogroup R1b, the most common European male lineage. My mtDNA is from haplogroup A, which originated in Northeastern Siberia and spread across Beringia about 17,000 years ago.

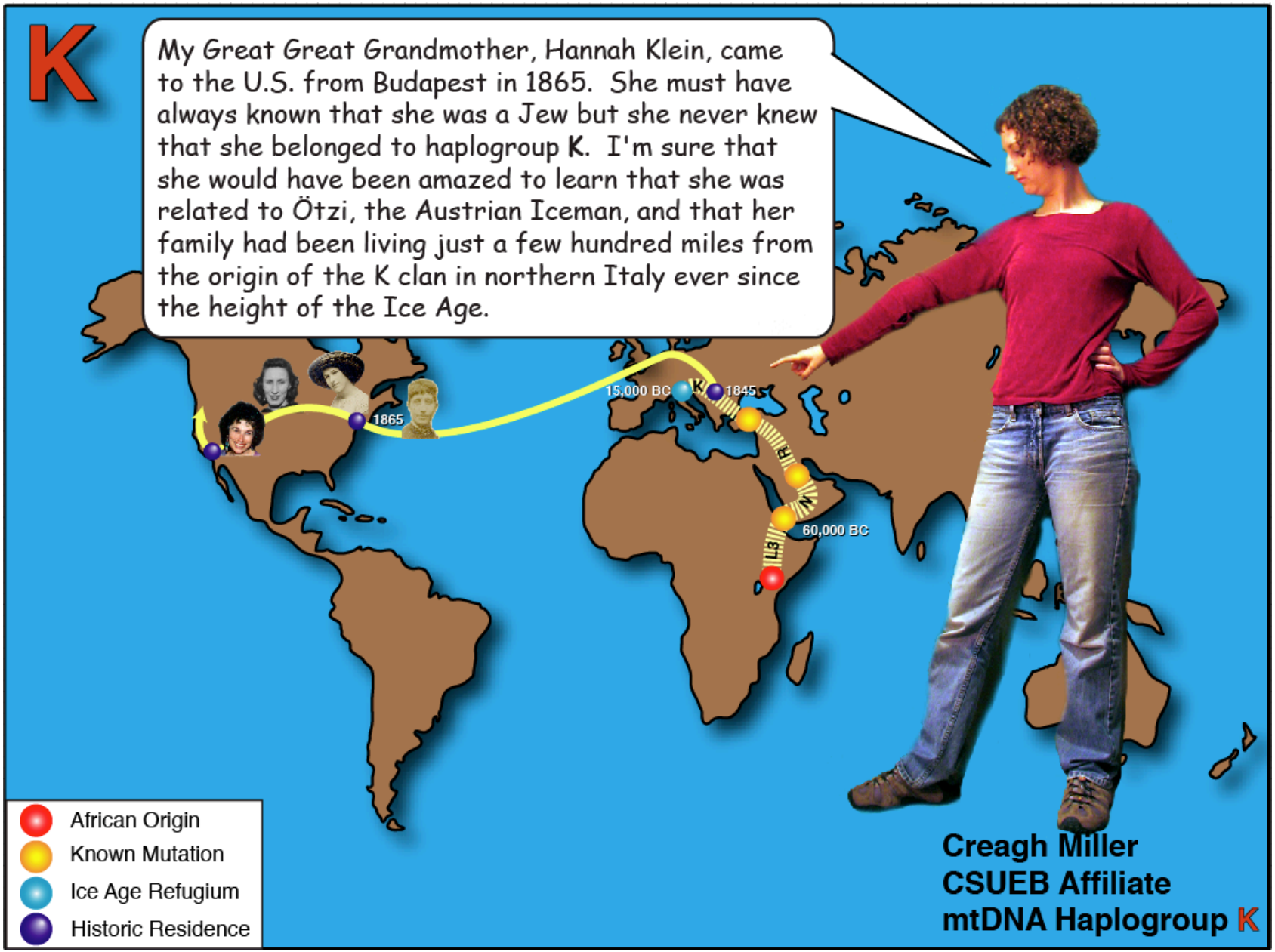
Cecilio Gudina
Anthropology Student
mtDNA Haplogroup A

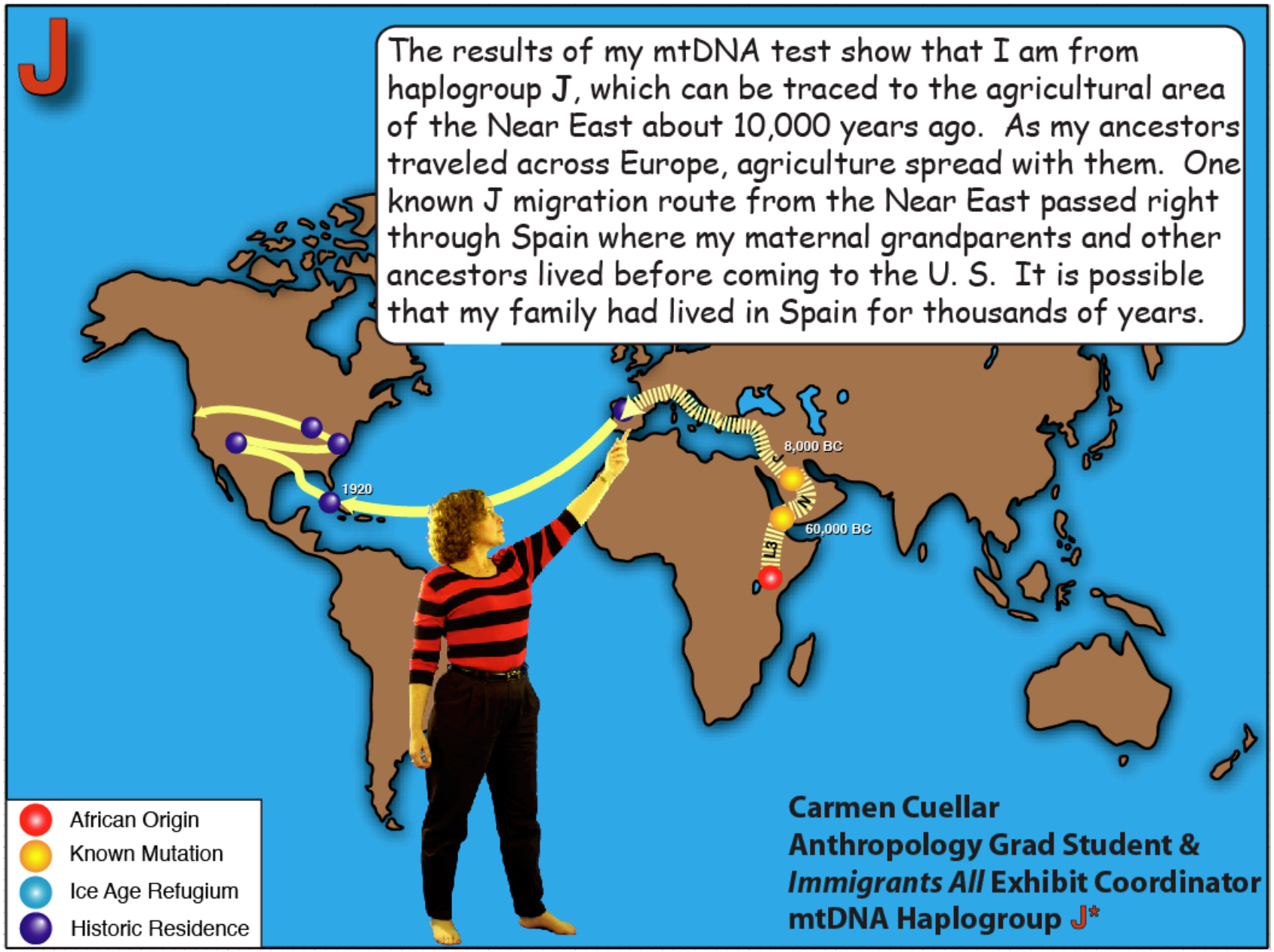

L2
My name is Emilee and I'm originally from Nigeria in West Africa. My family immigrated to the United States over 20 years ago. My mtDNA haplogroup, L2, is the most widely distributed lineage in North, West and Central Africa. It dates back to about 70,000 years ago and represents one ofthe two original mitochondrial lineages that continued to occupy Africa after the Out of Africa expansion carried the L3 lineage into Eurasia.
L2
70,000 BC
150,000 BC
L3
African Origin
Known Mutation
Ice Age Refugium
Historic Residence
Emilee Bargoma
Biology Student
mtDNA Haplogroup L2

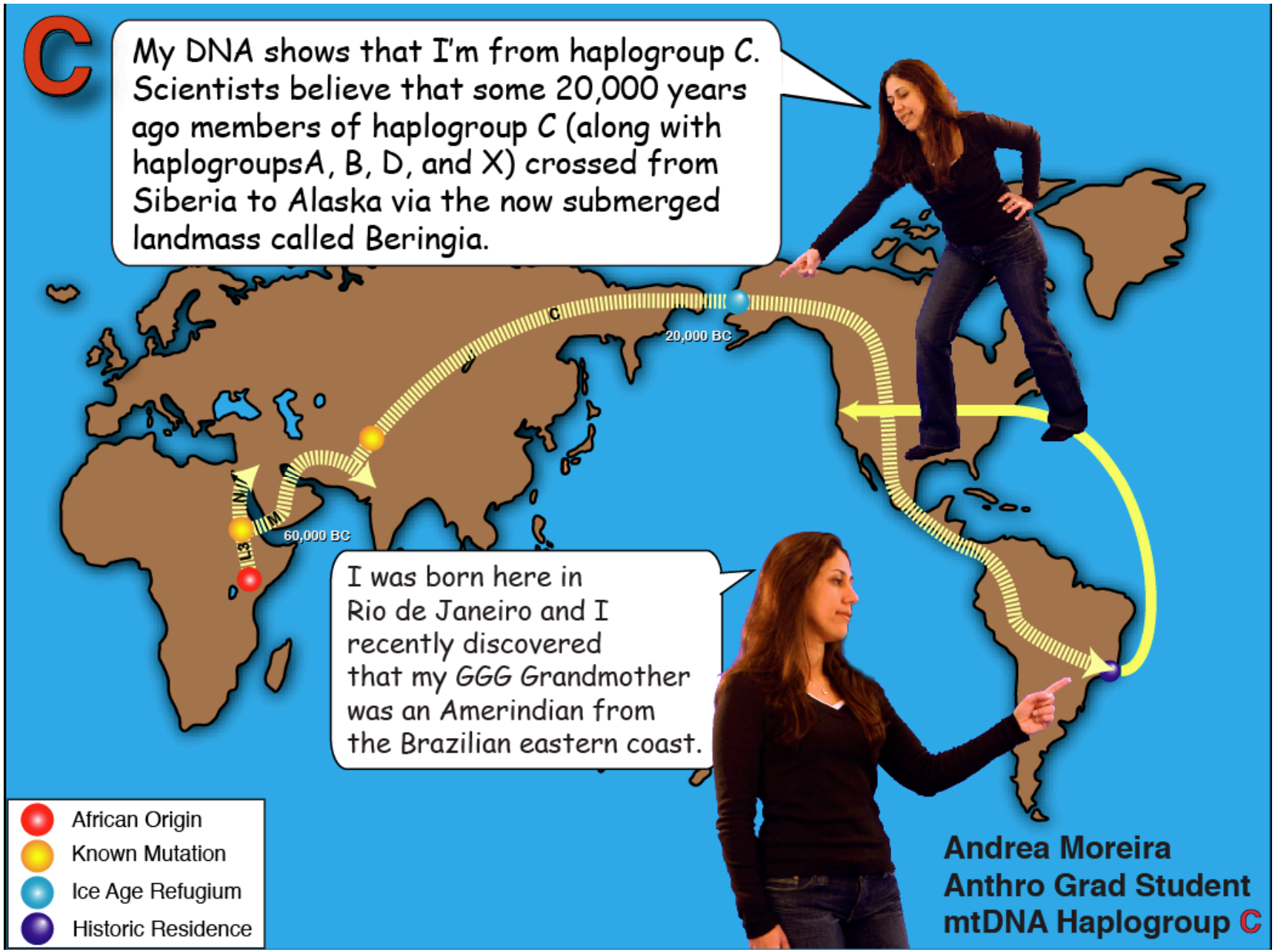

U
When my earliest maternal ancestors arrived in Europe Neandertals were still the dominant species there. I'm descended from the clan of Ursula (haplogroup U), the first mitochondrial lineage to enter Europe around 45,000 years ago. After arriving in Europe haplogroup U split into a number of sublineages, with my ancestors settling in the far north in Germany, Denmark and Scandinavia. My maternal grand-mother, Virginia Snavely emigrated from Germany in the late 1800s, the last of a very long line of pioneers.
45,000 BC
80,000 BC
African Origin
Known Mutation
Ice Age Refugium
Historic Residence
Kelly Larsen
Anthropology Grad Student
mtDNA Haplogroup U5a1

I was born in St. Petersburg, Russia and according to my family we come from a long line of Cossacks. But I guess our ancestors have not always lived in Russia since my mtDNA results show that I belong to haplogroup **I**, which originated in Turkey about 30,000 years ago. Members of haplogroup I are fairly rare today and they are spread throughout Europe from Russia and Transylvania in the east to Spain and Britain in the west.

Lisa Kosheleva
Biology Alumna
mtDNA Haplogroup I


K
My Great Great Grandmother, Hannah Klein, came to the U.S. from Budapest in 1865. She must have always known that she was a Jew but she never knew that she belonged to haplogroup K. I'm sure that she would have been amazed to learn that she was related to Ötzi, the Austrian Iceman, and that her family had been living just a few hundred miles from the origin of the K clan in northern Italy ever since the height of the Ice Age.
1865
15,000 BC
K
1845
R
N
60,000 BC
L3
African Origin
Known Mutation
Ice Age Refugium
Historic Residence
Creagh Miller
CSUEB Affiliate
mtDNA Haplogroup K

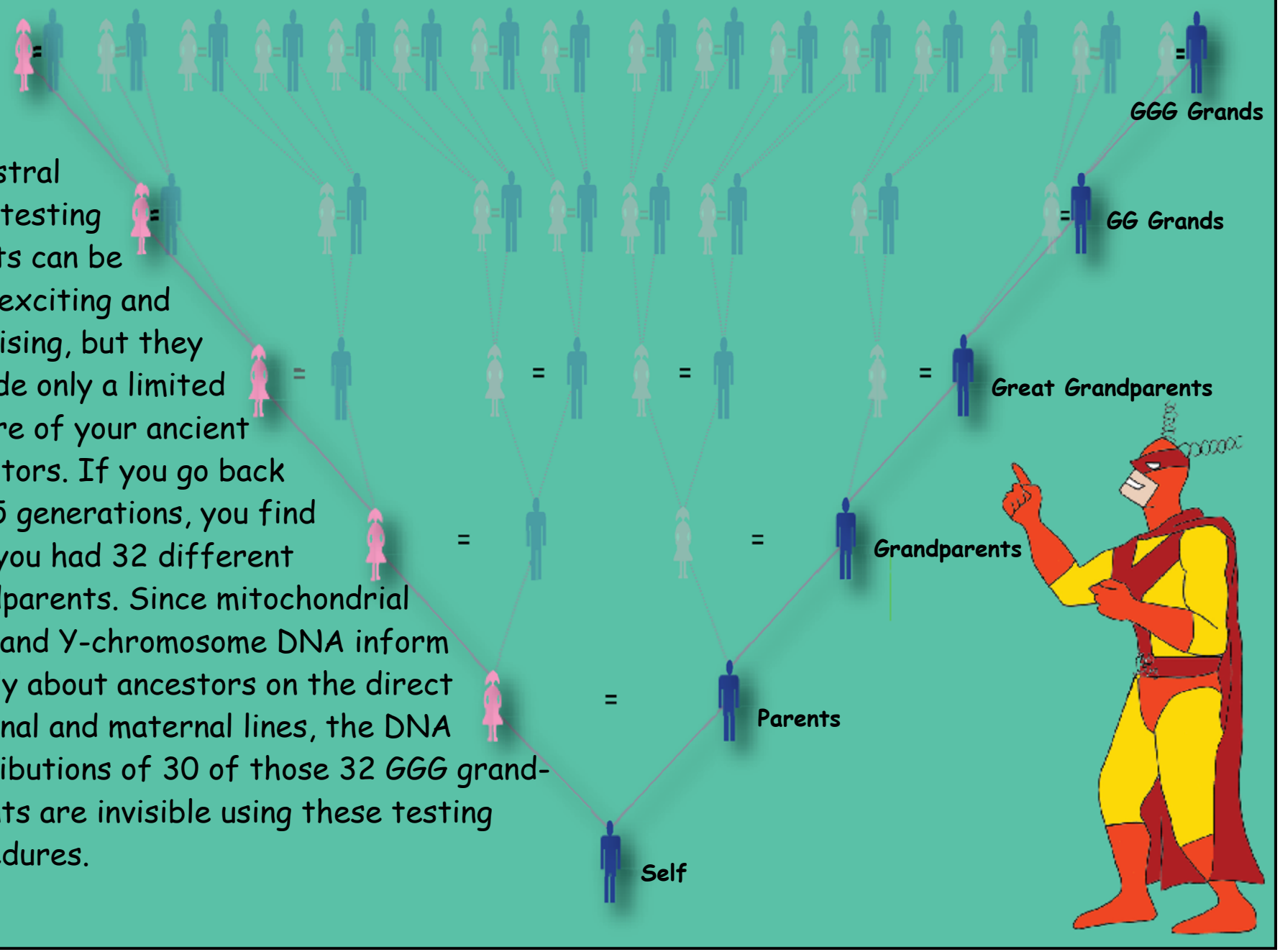

J
The results of my mtDNA test show that I am from haplogroup J, which can be traced to the agricultural area of the Near East about 10,000 years ago. As my ancestors traveled across Europe, agriculture spread with them. One known J migration route from the Near East passed right through Spain where my maternal grandparents and other ancestors lived before coming to the U. S. It is possible that my family had lived in Spain for thousands of years.
8,000 BC
60,000 BC
1920
African Origin
Known Mutation
Ice Age Refugium
Historic Residence
Carmen Cuellar
Anthropology Grad Student &
Immigrants All Exhibit Coordinator
mtDNA Haplogroup J*


C
My DNA shows that I'm from haplogroup C. Scientists believe that some 20,000 years ago members of haplogroup C (along with haplogroupsA, B, D, and X) crossed from Siberia to Alaska via the now submerged landmass called Beringia.
C
20,000 BC
N
M
L3
60,000 BC
I was born here in Rio de Janeiro and I recently discovered that my GGG Grandmother was an Amerindian from the Brazilian eastern coast.
African Origin
Known Mutation
Ice Age Refugium
Historic Residence
Andrea Moreira
Anthro Grad Student
mtDNA Haplogroup C

CREDITS

p.7 - modification of Herto Man image, *Nature* 12 June 2003
p.8 - based on Figure 10, *The Journey of Man* (2002) by Spencer Wells
p.9 - chromosome images from http://smi-web.stanford.edu
p.10 - chromosome image from http://www.epilepsyfoundation.org
p.14 - 2005 Y-Chromosome Phylogenetic Tree by Family Tree DNA
p.23 - mtDNA Migration Map by Family Tree DNA
p.24 - mtDNA Haplogroups of the World map by J.D. McDonald, ©2004
p.28 - Herto Man image, *Nature* 12 June 2003 and 2005 Y-Chromosome Phylogenetic Tree by Family Tree DNA
pp.29-34 - 2005 Y-Chromosome Phylogenetic Tree by Family Tree DNA

The Limits of Maternal/Paternal DNA Testing



Ancestral DNA testing results can be both exciting and surprising, but they provide only a limited picture of your ancient ancestors. If you go back just 5 generations, you find that you had 32 different grandparents. Since mitochondrial DNA and Y-chromosome DNA inform us only about ancestors on the direct paternal and maternal lines, the DNA contributions of 30 of those 32 GGG grand-parents are invisible using these testing procedures.