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Maternal descendant of Elizabeth Wilborn

Human Migratory Path for the maternal ancestors of Elizabeth Wilborn based on mtDNA analysis.

Summary

Your maternal ancestry follows a typical pattern for Europeans—leaving Africa and settling in Asia for thousands of years before migrating to Europe after the last Ice Age <15,000 years ago. According to FTDNA, your haplogroup is H, but I believe this needs to be confirmed through additional testing. Haplogroup H is dispersed throughout the Europe, the Middle East and Asia as far east as India. It is the most common haplogroup in Europe. Since you have only tested HVR1, a small portion of your mitochondrial DNA, additional testing is required to find your ancestry for specifically.

Introduction

Below I have articulated the path your ancestors followed through ancient times to eventually link to your current genealogy. If you go back far enough, most humans descend from a common male ancestor, connecting us to a world family tree that is approximately 70,000 years old. A few African lines are much older, dating back to 140,000 years ago. The same is true for the maternal line, except the common convergence of all humans maternally is far older still, approximately 170,000 years ago.

New research is being added to further specify the complexities surrounding our common human origins—the beginning of this tree. In addition, new research is uncovering more specific SNPs (specific mutations) related to your place in the world family tree. The information I have included here is what is currently available as of June 2012.

Y-DNA Single Nucleotide Polymorphisms (SNPs)[1] are labeled according to their discovery in specific genetic laboratories with a letter followed by a set of numbers (for example, *V168*). Mitochondrial DNA SNPs are labeled according to their base-pair location (001-16559), with the

[1] SNP, or "single nucleotide polymorphism" is a unique mutation found in the DNA of an individual, haplogroup or species. SNPs can be arranged chronologically within a population by determining the scope and prominence of each in relation to a mutually-shared parent SNP. There are certain SNPs common to all humans. There are others that are exclusive to specific haplogroups, and others that are found among everyone except for specific haplogroups. The organization and distribution of haplogroups based on SNPs is among the most fascinating aspects of genetic genealogy and provides the most relevant information about one's deep ancestry.

former nucleotide mentioned prior to the location and the new nucleotide mentioned after (for example *G16153A*). A collection of individuals that exhibit the same SNPs is termed a "haplogroup[2]," which can be further defined into specific clades and subclades. Haplogroups are labeled according to their line of descent, alternating letters and numbers (for example, *R1b1a2*). SNP labels are permanent, while a haplogroup's subclade labels may change slightly as new research further specifies a more exact line of descent. All SNPs within a parent clade are found in a descendant population. Nature exhibits new SNPs at random times throughout the history of your specific lineage, and initially each new SNP is carried by one female individual and passed on to her children.

This process describes the changes within mitochrondrial DNA (mtDNA) since the first humans all the way to the present day. The process parallels that used with y-chromosomal DNA to test the paternal line of decent. Y-chromosomal DNA corresponds to surnames, providing an easier way to link DNA tested to genealogy. Tracing mitochrondrial DNA down to a specific family is difficult on two accounts: First, mtDNA is not attached to a specific surname. And second, mtDNA is limited to 16,559 base pairs.

While all SNP designations are unique, haplogroup designations are not, and must be qualified as to whether the haplogroup refers to mtDNA or yDNA. For example mtDNA haplogroup L1 is a completely different set than yDNA haplogroup L1.

By following the line of descent through the various SNPs specific to your haplogroup, one can compare your SNPs to those of current populations around the globe and develop the specific migratory path of your ancestors, including where and when you diverge from other global populations. With each subsequent SNP, one defines a more specific population and moves closer to the present day, and closer to connecting your genealogy (paper records) with your deep ancestry as revealed through DNA.

Some haplogroups and their subclades are defined by more than one SNP. These have yet to be fractured into individual subsequent mutations that will further specify your line of descent. Paternal (yDNA) lineages can be traced more specifically that maternal (mtDNA) lineages because yDNA is composed of approximately 58 million base pairs while mtDNA includes only around 16,500. SNPs within yDNA are more likely to be unique than SNPs with mtDNA.

Your SNPs follow the haplogroup, clade and subclade descent below. Your SNPs are indicated by the original base, followed by the position, followed by the new base. For example *G16153A* indicates that originally position 16,153 held a Guanine (G), but in your case it

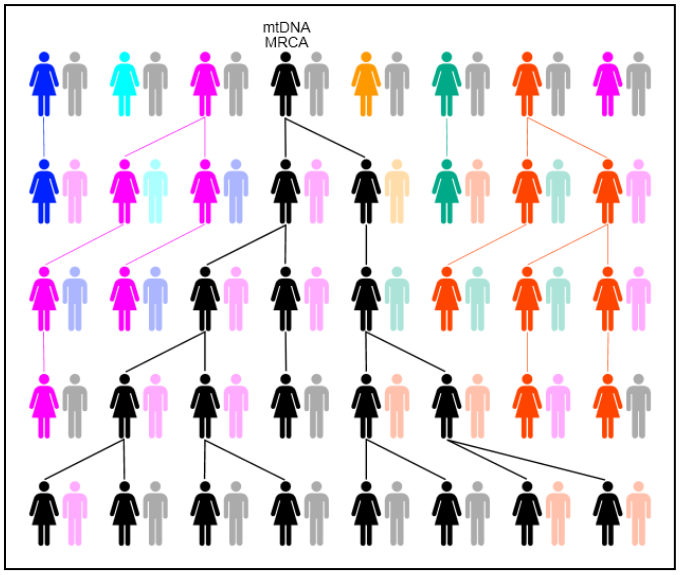
[2] A haplogroup is a term used in molecular biology to categorize a group of individuals who share a common ancestor through a "single nucleotide polymorphism" (SNP) mutation. Haplogroups based on Y-chromosomal DNA trace one's paternal ancestry. Haplogroups based on mitochondrial DNA trace one's maternal ancestry. This report only refers to mtDNA haplogroups. A haplogroup's nomenclature reflects its ancestry. For example, haplogroup R1b1a2a1a1b is a descendant of R1b1a2a1a1, which is a descendant of R1b1a2a1a, all the way back to R. Following the ancestral lines of human populations, all haplogroups find their ultimate ancestry with a common source in East Africa. Most European mitochondrial haplogroups descend through macrohaplogroup N.

changed to an Adenine (A). This mutation is defined chronologically by comparing people around the world who have this SNP in comparison to other SNPs. Some SNPs are common to all humans around the world. Some SNPs are found exclusively within large geographical populations. And some SNPs are found only in specific family groups. By arranging your SNPs chronologically, understanding that some SNPs are nested within others, we can develop the ancient migratory path of your ancestors.

The system used to give a specific time frame to the formation of successive SNPs is based on a combination of factors, including genetics, anthropology, archeology, cultural and social history, climatology and linguistics. Dates are approximate and subject to adjustment.

In humans, mtDNA consists of between 16,000 and 17,000 base pairs. There are two hypervariable control regions (HVR1 and HVR2) that are analyzed in a mtDNA test for genealogical purposes. HVR1 consists of about 440 base pairs. These 440 base pairs are then compared to the control regions of other individuals (either specific people or subjects in a database) to determine maternal lineage. Most often the comparison is made to the revised Cambridge Reference Sequence (CRS). As you can see when you log into the FTDNA website, your results are compared as differences from the CRS. The CRS contains 16,569 base pairs.

The CRS was established in 1981, long before the human genome project, and long before the large-scale DNA testing available today. However, the CRS is still used as the standard upon which all human mitochrondrial DNA is compared. The CRS belongs to haplogroup H2a2a1. If you fall into a different haplogroup than the CRS, you will have a long list of differences from the CRS upon which to compare. Those differences reveal your haplogroup and subclade.



Through random drift or selection the female-lineage will trace back to a single female, such as Mitochondrial Eve. In this example over 5 generations colors represent extinct matrilineal lines and black the matrilineal line descended from mtDNA

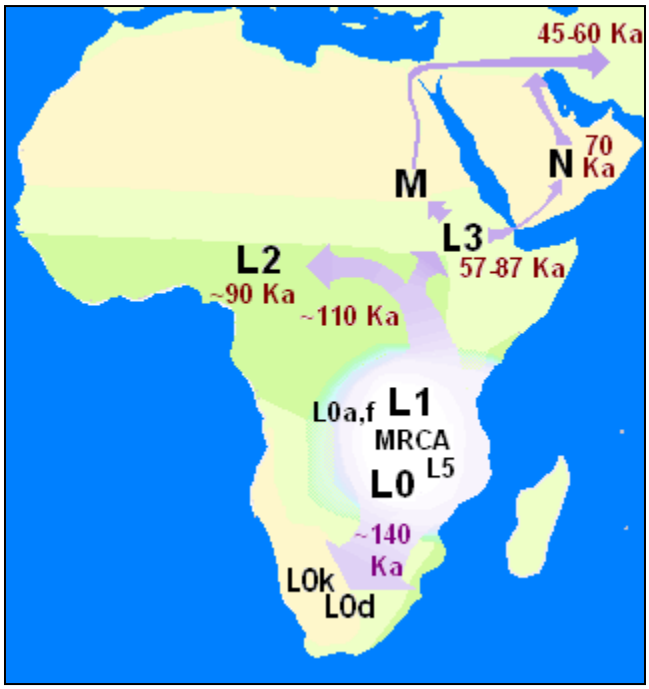
More recently the RSRS (Reconstructed Sapiens Reference Sequence) has been established to track phylogenetic changes from the root of the mtDNA tree. Your report follows the RSRS from the earliest humans in East Africa to the point where I've included all your mutations within the tree.

This outline begins with "Mitochondrial Eve," the common maternal ancestor of all humans alive today. However she was not the only woman alive at that time. She was part of a group of humans, possibly numbering around 10,000, that lived in East Africa. The others in her clan are also our ancestors, but she is the only woman to have an unbroken maternal line of descent

to everyone in the world today. The common maternal ancestor of all people alive a thousand years ago would have been earlier in time than the mitochondrial Eve of today, because lines of descent frequently go extinct. We find this phenomenon frequently upon analyzing ancient DNA in human remains.

We can follow the changes in your SNPs down through prehistory and through historic human populations to end up connecting to your genealogy. Your deep ancestry begins the same as all humans alive today. I use www.phylotree.org as my SNP guide to writing this report. Mutations within the hypervariable regions HVR1 (16001-16568) and HVR2 (001-574) are labeled in blue. Mutations in the coding region (575-16000) are labeled in black. Back mutations are labeled with an exclamation point (!). Back mutations are frequent in mitochondrial DNA and also need to be tracked because they may help place you in a more specific subclade.

mtDNA SNP line of descent



Map showing the location of some of the diversity of the L haplogroup in Africa, and the supposed migratory paths of M and N out of Africa between 60,000 and 70,000 years ago.

1 Mitochondrial Eve (East Africa)

All humans have a common maternal ancestor approximately 170,000 years ago in East Africa. This is determined by tracing mutations in mitochondrial DNA back through time. Mitochondrial Eve was not the only woman alive at that time, but she was the only one to be the direct maternal ancestor of everyone alive today. Mitochondrial Eve is traced to East Africa because East African populations have the most DNA variance, including the oldest known haplogroups. The RSRS begins with Mitochondrial Eve. Two branches form from the earliest humans: haplogroups L0 and L1'6. Each has a distinctive SNP signature. L0 is found in sub-Saharan Africa among the Khosian people of Namibia, Botswana and South Africa. L0 contains mutations at G263A C1048T C3516a T5442C T6185C C9042T A9347G G10589A G12007A A12720G. Everyone not part of L0 descends through macrohaplogroup L1'6, including you.

2 L1'6, East Africa C146T C182T T4312C T10664C C10915T A11914G G13276A G16230A

Between 140,000 and 170,000 years ago SNPs formed that are common in all humans except for those in haplogroups L0. These are categorized as macrohaplogroup L1'6, which later progressively divided into L1 and L2'6. Haplogroup L1 is found typically among the Mbenga Pygmies of Cameroon. The side branch L1 is defined by sequence changes at loci G3666A A7055G T7389C T13789C T14178C G14560A. You likely have all the above mutations that represent L1'6 except

C182T, which mutated back to its original state downstream at "generation" #5 (L3'4) below. Everyone not falling within L0 or L1 descends through haplogroup L2'6 below, including you.

3 L2'6, East Africa C152T A2758G C2885T G7146A T8468C

Between 114,000 and 126,000 years ago SNPs defining haplogroup L5 split away from L2'6. L5 is defined by sequence changes at loci 459.1C T3423C A7972G C12432T A12950G C16148T A16166G. L5 is found among Mbuti pygmies from the Congo, the Sandawe of Tanzania, and a scattering in Kenya, Ethiopia, Sudan, Nubia, Egypt and Saudi Arabia. You likely have all the above mutations that represent L2'6. You and everyone else not found in L0, L1 or L5 descends through macrohaplogroup L2'3'4'6 below.

4 L2'3'4'6, East Africa C195T A247G A825t T8655C A10688G C10810T G13105A T13506C G15301A A16129G T16187C C16189T

Between 85,000 and 111,000 years ago SNPs defining haplogroup L2 split away from L2'3'4'6. L2 is a very common haplogroup among Africans, accounting for approximately 1/3 of current Africans and about 19% of African Americans. It is defined by ten downstream sequence changes. You have all the mutations that represent L2'3'4'6 above. You descend through macrohaplogroup L3'4'6 below.

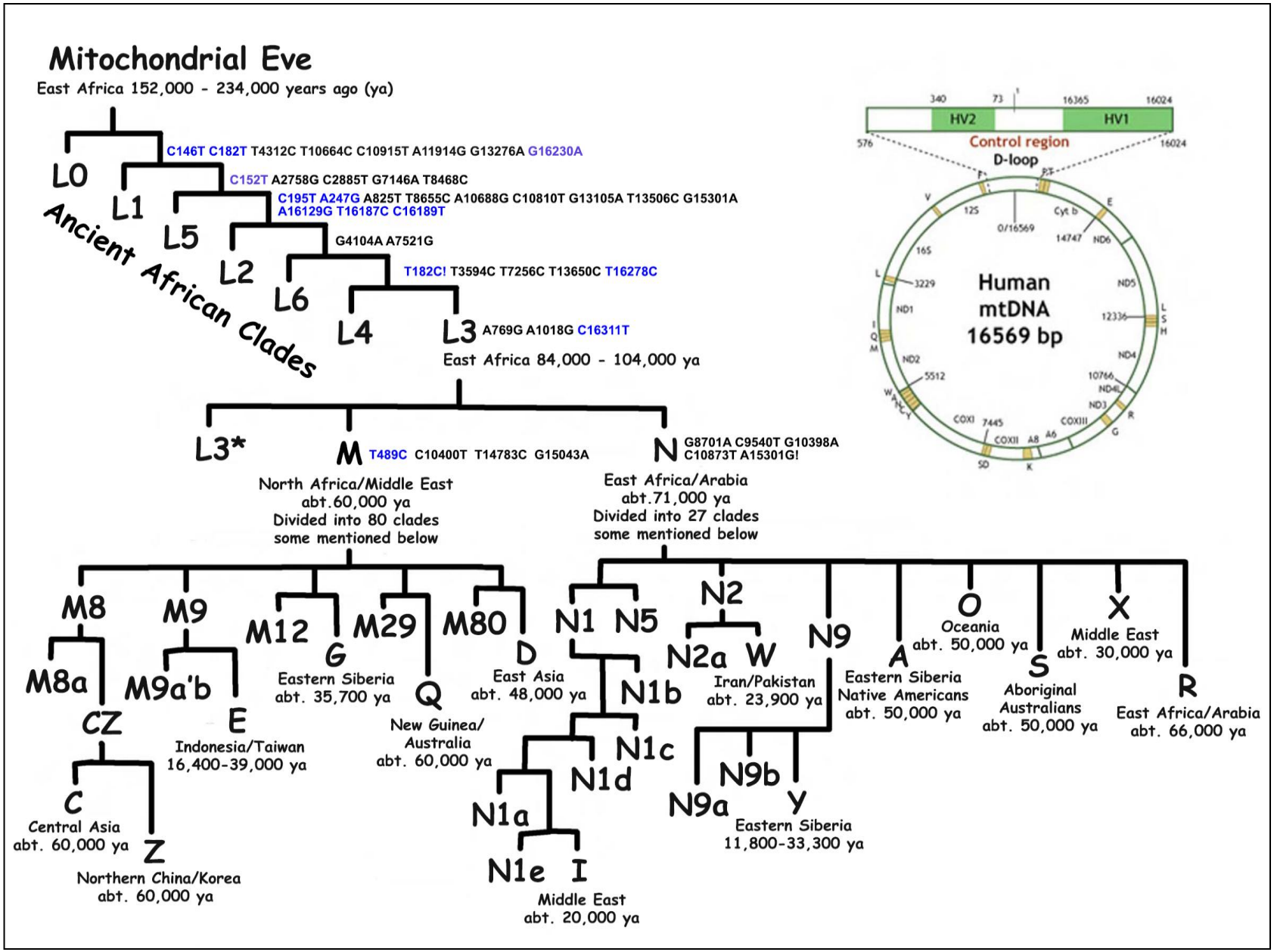
5 L3'4'6, East Africa G4104A A7521G

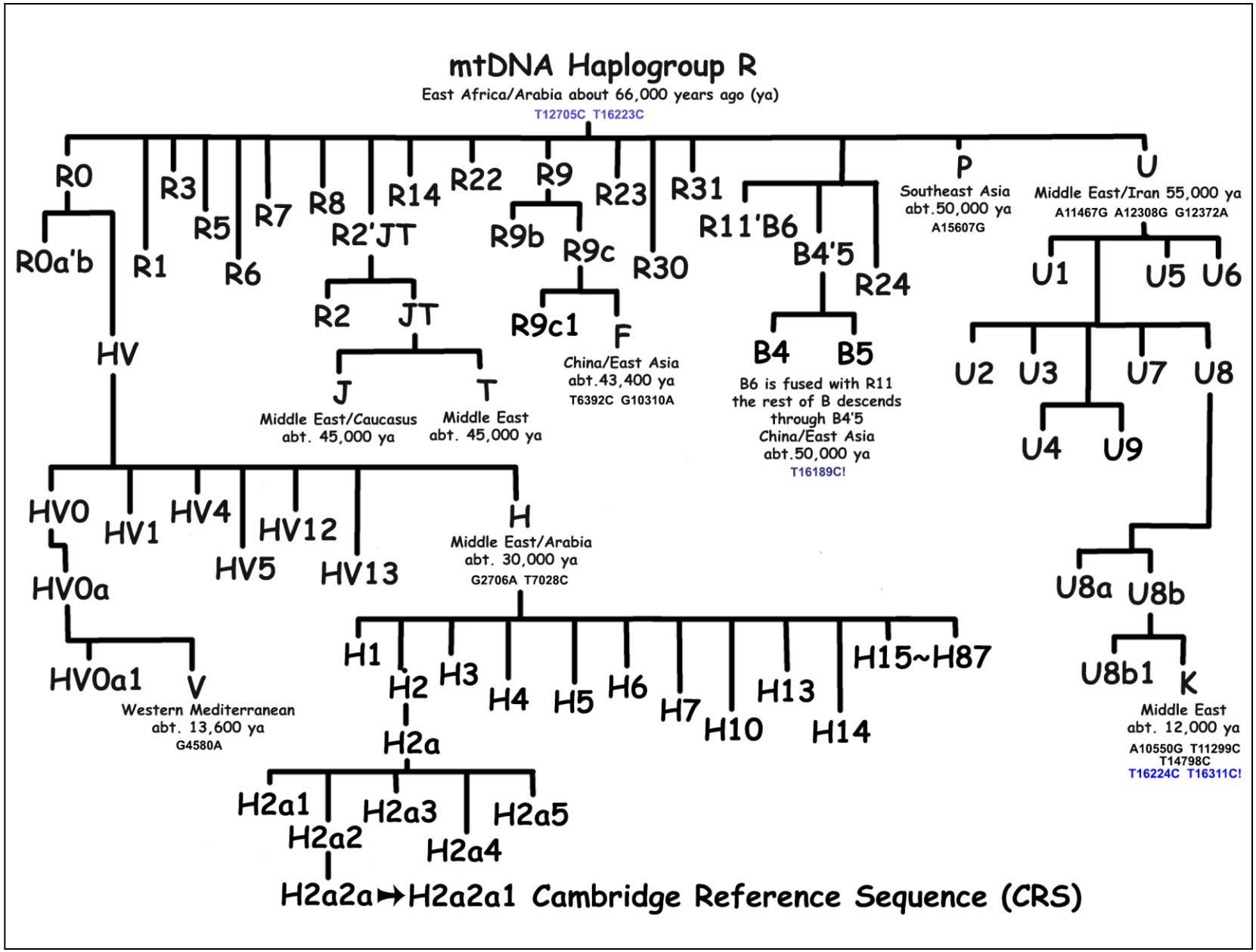
Sometime soon after the above divergence, around 80,000 years ago, haplogroup L6 forms from L2'3'4'6 in East Africa. Haplogroup L6 is defined by 24 downstream sequence changes. It is a small African haplogroup, found predominately in Ethiopia but also has a presence in Yemen. You likely have the two mutations that represent haplogroup L3'4'6 above. Almost all Eurasian populations, including you, descend through L3'4 below:

6 L3'4, East Africa T182C! T3594C T7256C T13650C T16278C

Note that in L1'6 above ("generation" #2), loci 182 converts from a C to a T. Here that position is reversed—the T reverts back to a C. This is an example of a "back mutation," that occurs occasionally in genetics. Mitochondrial DNA is more susceptible to back mutations than y-chromosomal DNA, and therefore these back mutations need to often be tracked in order to determine your specific subclade.

Around 70,000 years ago, the small African haplogroup L4 splits from L3'4, defined by mitochondrial DNA sequence changes at loci T195C! G5460A T16362C. Note that L4 has another back mutation! L4 is found in East Africa, particularly the Horn of Africa. Its highest frequencies are among the Hadza people (60-83%) and the Sandawe people (48%) of Tanzania. You do not belong to L4, but have all the mutations representing L3'4 above. You and almost all Eurasian populations descend through L3 below.


Mitochondrial Eve
East Africa 152,000 - 234,000 years ago (ya)
L0
L1
L5
L2
L6
L4
L3
Ancient African Clades
C146T C182T T4312C T10664C C10915T A11914G G13276A G16230A
C152T A2758G C2885T G7146A T8468C
C195T A247G A825T T8655C A10688G C10810T G13105A T13506C G15301A
A16129G T16187C C16189T
G4104A A7521G
T182C! T3594C T7256C T13650C T16278C
A769G A1018G C16311T
East Africa 84,000 - 104,000 ya
L3*
M T489C C10400T T14783C G15043A
N G8701A C9540T G10398A C10873T A15301G!
North Africa/Middle East
abt.60,000 ya
Divided into 80 clades
some mentioned below
East Africa/Arabia
abt.71,000 ya
Divided into 27 clades
some mentioned below
HV2
HV1
Control region
D-loop
Human
mtDNA
16569 bp
M8
M9
M12
G
M29
Q
M80
D
M8a
M9a'b
E
CZ
C
Central Asia
abt. 60,000 ya
Z
Northern China/Korea
abt. 60,000 ya
Indonesia/Taiwan
16,400-39,000 ya
Eastern Siberia
abt. 35,700 ya
New Guinea/
Australia
abt. 60,000 ya
East Asia
abt. 48,000 ya
N1 N5
N1b
N1c
N1d
N1a
N1e I
Middle East
abt. 20,000 ya
N2
N2a W
Iran/Pakistan
abt. 23,900 ya
N9
N9b
N9a y
Eastern Siberia
11,800-33,300 ya
A
Eastern Siberia
Native Americans
abt. 50,000 ya
O
Oceania
abt. 50,000 ya
S
Aboriginal
Australians
abt. 50,000 ya
X
Middle East
abt. 30,000 ya
R
East Africa/Arabia
abt. 66,000 ya


mtDNA Haplogroup R
East Africa/Arabia about 66,000 years ago (ya)
T12705C T16223C
R0
R0a'b
HV
HV0
HV0a
HV0a1
V
Western Mediterranean
abt. 13,600 ya
G4580A
HV1
HV4
HV5
HV12
HV13
H
Middle East/Arabia
abt. 30,000 ya
G2706A T7028C
H1
H2
H2a
H2a1
H2a2
H2a2a➡H2a2a1 Cambridge Reference Sequence (CRS)
H3
H4
H5
H6
H7
H10
H13
H14
H15~H87
R1
R3
R5
R6
R7
R8
R14
R2'JT
R2
JT
J
Middle East/Caucasus
abt. 45,000 ya
T
Middle East
abt. 45,000 ya
H2a3
H2a4
H2a5
R22
R9
R9b
R9c
R9c1
F
China/East Asia
abt.43,400 ya
T6392C G10310A
R23
R30
R31
R11'B6
B4'5
B4
B5
B6 is fused with R11
the rest of B descends
through B4'5
China/East Asia
abt.50,000 ya
T16189C!
R24
P
Southeast Asia
abt.50,000 ya
A15607G
U
Middle East/Iran 55,000 ya
A11467G A12308G G12372A
U1
U5
U6
U2
U3
U7
U8
U4
U9
U8a
U8b
U8b1
K
Middle East
abt. 12,000 ya
A10550G T11299C
T14798C
T16224C T16311C!

7 L3 East Africa A769G A1018G C16311T

Around 70,000 years ago haplogroup L3 and haplogroup L4 differentiate from each other. L3 further splits into multiple clades L3a through L3k, and M and N. The L3 clades L3a through L3k largely remain in Africa, but M and N are largely found outside of Africa. M and N are either found immediately outside of Africa (in Arabia), or formed in Africa immediately prior to the "Out of Africa" migration of modern humans. L3 is the halpogroup from which all modern humans outside of recent African origin derive. It represents the original "Out of Africa" migration 60,000-70,000 years ago, crossing the southern end of the Red Sea into Arabia. You have all three mutations represented by L3 above, and you descend through haplogroup N below.

8 N Arabia G8701A C9540T G10398A C10873T A15301G!

Note that haplogroup N contains another back mutation at A15301G. This mutation first formed in "generation" #4, L2'3'4'6, and reversed. Approximately 65,000 years ago haplogroup N differentiates from its parent clade L3 and its sister clade M. Haplogroup M is found predominately on the Indian subcontinent, while haplogroup N is distributed throughout Europe, Asia, Oceania, and the Americas. Haplogroup N splits further into the following:

1) haplogroup R - Most common in Western Eurasia, but also distributed in Southern Asia, Iran, Arabia, Pakistan and India. About 89% of Europeans descend through haplogroup R.
2) haplogroup A - northern and central Asia, Siberia. 8-15% of Koreans, and 7% of Japanese, 2% of Turkish peoples, and a scattering among Native Americans
3) haplogroup I - found in 2-3% of northern Europeans, but also found in Kenya (due to back migration), Pakistan, Iran and Azerbaijan. (Haplogroup I is actually a descendant of the subclade N1e).
4) haplogroup S - Aboriginal Australians
5) haplogroup W - Found throughout Europe but also in Northern Pakistan (haplogroup W descends from N2)
6) haplogroup X - Found among 2% of Europeans, in the Near East and North Africa.
7) haplogroup Y - Descends through subclade N9 and is found through coastal regions of East Asia, from the Malay Archipelago through Kamchatka.

Haplogroup R, above leads to the CRS (Cambridge Reference Sequence). Here you follow haplogroup R and from there you descend through haplogroup H. You can trace your descent from Mitochondrial Eve to clade H13 using the charts on pages 6-7.

9 R Arabia T12705C T16223C

Approximately 55,000 years ago haplogroup R formed from haplogroup N in the Arabian Peninsula and spread through Western Eurasia. About 89% of Europeans follow a stream through this haplogroup. Many Swiss families follow subgroup HV or subgroup R2'JT.

Haplogroup R has various subclades as follows below:

1) R0 - Arabian peninsula with a scattering across North Africa, the Horn of Africa, South Asia and Europe. HV is a subset of R0, found in the Middle East and Iran.

2) R1 - India, Caucasus and Central Asia. Found in 9% of peoples from Turkmenistan
3) R2'JT - Found from India/Pakistan through the Caucasus and Turkey into Europe, includes about 11% of Europeans
4) R3 – Armenia
5) R5 – Indian subcontinent (found in Madhya Pradesh at 17%)
6) R6'7 - India and Pakistan (among 10% 0f those speaking Austro-Asiatic languages)
7) R8 - East India among the Orissa people at 12%
8) R9 - Southeast Asia, Indonesia, Malaysia
9) R11'B - China, especially the Lahu people (12.5%), also found in South Asia, coastal Asian regions, Polynesia, and Madagascar.
10) R12'21 - Found in Malaysia and among Australian Aborigines.
11) R14 - Found in Papua New Guinea and teh Nicobar Islands
12) R22 – Eastern Indonesia, especially the Lesser Sundra Islands (8%)
13) R23 - On Bali and Sumba in Indonesia
14) R30 - India, Tharu people from Nepal, and Sinhalese people from Sri Lanka
15) R31 - India
16) P – Australia, Melanesia, Philippines and Eastern Indonesia
17) U - Middle East and Caucasus, including about 11% of Europeans.

From this point forward, I cannot completely determine your path, since you have only tested the HVR1 region of mtDNA. If you test more of your mtDNA, the path will be clearer. You have two additional mutations within the HVR1 region that is not accounted for above. T16189C and C16218T are found in several locations downstream from R, but not together, except in your DNA. More testing is needed to sort out your exact path. The options are as follows:

Options that are positive for T16189C

1) R0a1b – C26T T16093C T16189C!
2) HV1b2 – A3547G G6023A T16189C!
3) H1a6 – C151T A11893G T16189C!
4) Unnamed clade directly upstream from H1b, H1f, H1g, H1k, H1y, H1z, H1aa, H1ab, H1ac, H1ad – T16189C!
5) H1c3b – T16189C! T16362C
6) H1ap1 – T152C! G5780A T16189C!
7) H3av – T16189C!

Options that are positive for C16218T

1) H1ag1a – G12940A C16218T
2) H1aq1 -- C15304a C16218T
3) H20 – C16218T

Based on the above downstream options, you are almost certainly part of haplogroup H. And within H, you are probably part of H1. I expand this line to H1 below:

10 R0 Arabia G73A A11719G

Approximately 40,000 years ago (range between 23,600 to 54,900 years ago), R0 formed from R on the Arabian peninsula. Its highest frequency is on the island of Socotra (38%) in the Indian Ocean and among the Kalash (23%) of Pakistan. It is found all over the Arabian peninsula. RO later splits to form R0a'b (Arabian Peninsula) and HV (Middle East/Iran). You likely descend through HV.

11 HV Turkey/Iraq/Iran/Caucasus Mountains T14766C

Approximately 25,000 years ago HV forms from R0 in southwestern Turkey, Iraq, Iran or the Caucasus and spreads across the Caucasus to southern Russia and Georgia. It is also found among Sudanese Arabs due to back-migration. These ancestors represent the second wave of modern human migrations to Europe (the first, over 30,000 years ago, belongs to mtDNA haplogroup U5). These continual migrations sounded the end of the era of the Neandertals. In 2003, Cro-Magnon bones in southern Italy dated at 24,000 years ago were found to be part of mtDNA HV.

HV splits into the following clades: HV0 (from which V descends), HV1, HV4, HV5, HV12, HV13, and H. You descend through H.

12 H Turkey/Caucasus Mountains G2706A T7028C

Approximately 20,000 years ago H forms from HV and eventually becomes the most dominate mtDNA haplogroup in Europe, including about half of all Europeans. Haplogroup H expanded quickly into central and northern Europe following the last Ice Age. In the Far East and the Caucasus, H reaches 20%, and in Iran, 17%. H has been divided into nearly 40 subclades, most of which have a European component. H1 and H3 are the most dominant European haplogroups. You likely descend through the major clade H1. It is here where you diverge from the CRS (Cambridge Reference Sequence), as the CRS falls within H2a2a1.

13 H1 Turkey/Caucasus Mountains G3010A

Approximately 17,000 years ago H1 forms from H and eventually becomes a dominate mtDNA haplogroup in Western Europe, and with significant distribution in Eastern Europe, North Africa and the Middle East.

Next Steps

You should upgrade your test to include at least HVR2 or the full sequence which includes the coding region of mtDNA. When more markers are tested, I can determine your line of decent more completely.

Let me know if you have any questions.

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Maps in this report are derived from maps.google.com or www.worldatlas.com.
Photographs of ancient objects are derived from the public domain databases of www.wikipedia.org.
Ancestral charts appearing in this report were created using Adobe Photoshop Elements 7.0.
The information found in this report is subject to modification and revaluation as more people have their DNA tested.

To learn more about DNA visit www.lmhs.org or the DNA discussion forum at https://discuss.lmhs.org.