

Universidade do Minho Escola de Ciências

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# Mitochondrial DNA phylogeography of African lineages

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# Mitochondrial DNA phylogeography of African lineages

Dissertação de Mestrado Mestrado em Genética Molecular

Trabalho efetuado sob a orientação do Doutor Pedro Alexandre Dias Soares e da Doutora Teresa Sofia Teixeira Rito

# <span id="page-3-0"></span>DIREITOS DE AUTOR E CONDIÇÕES DE UTILIZAÇÃO DO TRABALHO

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# <span id="page-4-0"></span>**ACKNOWLEDGEMENTS**

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This master project is inserted in a project to investigate the genetic legacy on the slave trade ("From Portugal back to Africa: uncovering the African roots of present-day Portuguese" - TDC/SOC-ANT/30316/2017).

# <span id="page-5-0"></span>STATEMENT OF INTEGRITY

I hereby declare having conducted this academic work with integrity. I confirm that I have not used plagiarism or any form of undue use of information or falsification of results along the process leading to its elaboration.

I further declare that I have fully acknowledged the Code of Ethical Conduct of the University of Minho.

# <span id="page-6-0"></span>**ABSTRACT**

Africa is believed to be the birthplace of the first Anatomically Modern Human at 200 ka. This first form of Homo sapiens colonized Africa first and had one thing in common: the haplogroup of the mitochondrial DNA (mtDNA) L. Around 60 ka occur the Out-of-Africa migration, and from then on there were certain period of increased migration from Africa to mainly Europe and Southwestern Asia, mainly associated with optimal climatic conditions. More recently, with the globalization and the discovery of the New World, some European countries started to export slaves from Africa to Europe and the Americas. Since mtDNA is maternally inherited and is non-recombining, it is the "perfect" way to follow of lineages and migratory movements, allowing the study of the sources of displaced populations.

In order to attempt to understand the main source of population within Africa that migrated to Europe, Southwestern Asia, North and South America, a phylogeographic analysis of the L haplogroup was performed using 6658 human mtDNA sequences from Africa, Europe, Southwestern Asia, North and South America. Using phylogenetic reconstruction, a molecular mtDNA clock to date the events and Founder analysis we were able to reconstruct the history of the haplogroup L all over the world and visualize the peaks of migration from Africa to the other continents.

Two main events of migration from Africa occurred: one at 8000 years and the other at 500 years. Geographically, the source of populations at these time-frames are clearly different in some aspects, such as the appearance of South African lineages in Europe, Southwestern Asia and the Americas only in historical migrations (500 years ago). When associating this fact with world history, we can correlate this with the Trans-Atlantic Slave Trade route, a massive forced movement of over 10 million African from the  $16<sup>th</sup>$  century on.

Genetic studies associated with ancestry aid us in better comprehend history. In here, we were able to uncover a bit more about the L haplogroup's history, not only in Africa but all over the world, about the Trans-Atlantic Slave Trade and the genetic evidences left behind that are still present to this day.

Keywords: Africa, Founder analysis, mtDNA, Phylogeography, Transatlantic Slave Trade.

# <span id="page-7-0"></span>**RESUMO**

Acredita-se que Africa é o berço do Ser Humano anatomicamente moderno, há 200 000 anos. Esta primeira forma de Homo Sapiens colonizou Africa e tinha um facto em comum: o haplogrupo do ADN mitocondrial (mtDNA) L. Há cerca de 60 000 anos ocorreu a primeira migração para fora de Africa (" Out-of-Africa"), e a partir daí existiram períodos de maior migração de Africa para a Europa e o Sudoeste Asiático, principalmente associados com condições climáticas ótimas. Mais recentemente, com a globalização e a descoberta do Novo Mundo, alguns países europeus começaram a exportar escravos de Africa para a Europa e as Américas. Uma vez que o mtDNA é herdado por via materna e não sofre recombinação, é a forma "perfeita" de seguir linhagens e movimentos migratórios, permitindo o estudo da fonte das populações deslocadas.

Numa tentativa de compreender a principal fonte de população de África que migrou para a Europa, o Sudoeste Asiático, a América do Norte e do Sul, foi feita uma análise filogeográfica do haplogrupo L com 6658 sequências de mtDNA humano de Africa, Europa, Sudoeste Asiático, América do Norte e do Sul. Com reconstrução filogenética, relógio molecular de mtDNA para datar eventos e análise de Fundador, conseguimos reconstruir a história do haplogrupo L em todo o mundo e visualizar os picos de migração de Africa para outros continentes.

Ocorreram dois principais eventos de migração: um há 8000 anos e outro há 500 anos. Geograficamente, as populações-origem destes períodos são claramente distintas em alguns aspetos, como a presença de populações do sul de Africa nos restantes continentes apenas nas migrações históricas (500 anos). Quando associamos este facto com a história mundial, podemos relacioná-lo com a rota Transatlântica de tráfico de escravos, que movimentou à força mais de 10 milhões de africanos a partir do séc. XVI.

Os estudos genéticos associados à ancestralidade ajudam-nos a compreender melhor a história. Neste trabalho conseguimos revelar um pouco mais sobre o haplogrupo L, não só em Africa, mas em todo o mundo, sobre a rota Transatlântica de tráfico de escravos e as ligações genéticas deixadas que ainda hoje estão presentes.

Palavras-chave: Africa, Análise de Fundador, mtDNA, Filogeografia, Transatlântica de tráfico de escravos.

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# <span id="page-13-0"></span>**INTRODUCTION**

It is part of the Human nature to be curious about our origins and consequently to work as hard as possible to achieve answers. There are demographic and migratory events in Human history that need further explanation. With this in mind this thesis is organized in four chapters. On the first chapter an explanation is provided on the importance of mitochondrial DNA (mtDNA) in ancestry studies, the concept of phylogeography, the theories of the origin of modern humans and the expansion of mankind and the influence of slavery in the current mtDNA gene pool worldwide. The second chapter includes a list of all the methods used to answer the aims of this thesis, being the results shown on the third chapter. The last chapter is the discussion of the results: and that includes reviewing the results obtained and contextualising these results within the area of phylogeography. Also, the discussion contains the assessment of the goal's completion and future perspectives.

# <span id="page-14-0"></span>1. MITOCHONDRIAL DNA

### <span id="page-14-1"></span>1.1. THE MITOCHONDRIA

Mitochondria are organelles present in the majority of eukaryotic cell. Their main function is to produce metabolic energy [\(Alberts et al., 1994\)](#page-62-1).

Mitochondria were firstly observed in 1840, but only in 1925 they were implied in the respiratory chain [\(Ernster and Schatz, 1981\)](#page-65-0). Almost 30 years later, in 1957, mitochondria received the popular denomination "powerhouse of the cell" by the biologist Philip Siekevitz [\(Siekevitz, 1957\)](#page-69-0), due to their capability of generating most of the chemical energy needed to power the cell's biochemical reactions. Structure-wise, each mitochondrion is composed of an outer membrane and an inner membrane, with high motility and they have the capability of fusion and fission with other mitochondria [\(Chan, 2006;](#page-64-0) [Karbowski and Youle, 2003\)](#page-67-0). Mitochondria have a morphology that drastically changes according to the cell, including variation inside the same cell [\(Detmer and Chan,](#page-65-1)  [2007\)](#page-65-1), and they vary in number and location according to the cell type.

Functionally, mitochondria are linked to important roles such as signalling, cellular differentiation, apoptosis and autophagy, steroid and lipids synthesis, controlling the cell cycle and growth and regulation of the membrane potential and the cellular metabolism, beside the most notorious function of producing the cell's energy in the form of adenosine triphosphate (ATP) [\(McBride et al.,](#page-68-0)  [2006\)](#page-68-0).

#### <span id="page-15-0"></span>1.2. THE MITOCHONDRIAL GENOME

Mitochondrial DNA is the genetic material inside the mitochondria. The human mtDNA was the first mitochondrial genome to be completely sequenced in 1981 [\(Anderson et al., 1981\)](#page-62-2). A single mitochondrion can have up to 10 copies of mtDNA, each with 16569 base pairs, according to the revised Cambridge Reference Sequence (rCRS), compiled in an extremely compact circular double stranded conformation without histones [\(Andrews et al., 1999\)](#page-62-3).

The mtDNA sequence contains 37 genes, of which 28 are located on the H-strand (the "heavy" strand, rich in guanine) and 9 on the L-strand (the "light" strand, rich in cytosine by complementarity) distributed as shown in Figure 1. As mentioned previously, the mitochondria are extensible involved on the cell energy production, with 13 genes related with respiratory chain complexes, producing enzymes used on the oxidative phosphorylation, an ATP producing mechanism [\(Sharma](#page-69-1)  [and Sampath, 2019\)](#page-69-1). Of the other genes, 2 encode for rRNA and 22 for tRNA.

A major part of the mtDNA genome is the control region, with 1.1 kb, located between positions 576 and 16024 (Figure 1). This region contains the transcription and regulation factors, the origin of replication for the H-strand [\(Crews et al., 1979\)](#page-65-2) and, importantly, three hypervariable sections: HVS-I, HVS-II and HVS-III. These sections are fundamental in the study of human population genetics, mainly in the early stages of archaeogenetics, even though the last section (HVS-III) has a more focused application in forensic sciences and it is less variable [\(Brandstätter et al.,](#page-64-1)  [2004\)](#page-64-1). Although earlier archaeogenetic studies used exclusively HVS-I and HVS-II to genetically characterise the human populations, in the last two decades, the sequencing of the complete mtDNA genome allowed a much higher resolution and previous concepts about the history of worldwide populations were revisited.



<span id="page-16-0"></span>Figure 1 - Representation of the human mitochondrial genome.

mtDNA has 16569 bp compiled in a circular conformation. It has 37 genes organized in two strands: The H-strand ("heavy"), with 28 genes, and the L-strand ("light"), with 9 genes, all depicture in the figure. Emphasis on the control region and two of the hypervariable regions, HVS-I and HVS-II. Adaptation from [\(Jobling et al., 2014\)](#page-67-1)

### <span id="page-17-0"></span>1.3. MITOCHONDRIA IN POPULATION STUDIES

Mitochondrial DNA has several characteristics that make it the ideal subject for human population studies:

# 1.3.1.Heritage and Lack of recombination

<span id="page-17-1"></span>mtDNA is inherited almost exclusively by the maternal lineage, although some cases of paternal inherence have been reported [\(Luo et al., 2018;](#page-67-2) [Schwartz and Vissing, 2002\)](#page-69-2) but given the small amount of occurrences, these situations are considered extremely rare and possibly associated with abnormal processes. The male mtDNA, present in the sperm's mitochondria, is marked to be destroyed with a ubiquitin tag once inside the fertilized oocyte [\(Sutovsky et al., 1999\)](#page-70-0). So, even though mitochondria are capable of performing recombination, this has never been observed in the context of human evolution [\(Jobling et al., 2014\)](#page-67-1).

In general, being exclusively maternally inherited, presents a slight disadvantage, since mtDNA only presents the maternal side of the story. Thus, for a better picture of the evolutionary history, there is the need to integrate all the available genetic information with the mtDNA inferences, like using autosomes, Y-chromosome and incorporating other data from other fields of research (like archaeology and palaeontology) [\(Vibranovski and Long, 2016\)](#page-71-0).

## 1.3.2.Number of copies

<span id="page-17-2"></span>Unlike nuclear DNA where a single copy exists, each cell contains hundreds of mitochondria and thus, thousands of mtDNA copies. As a result, it is widely used on ancient DNA (aDNA) studies and forensic cases from degraded samples, where mtDNA is easier to find than nuclear DNA, that tends to be in very low concentrations [\(Pääbo, 1989\)](#page-68-1).

## 1.3.3.Mutation rate

<span id="page-17-3"></span>Just like nuclear DNA, mtDNA also suffers mutations (deletions, insertions and/or base substitutions), although in a faster mutation rate, up to 10 times faster, that allows the tracing of the maternal side of the human evolution more accurately in the timeline of most recents aspects of human history. This is often attributed to the high exposure of mitochondria to reactive oxygen species on the course of oxidative phosphorylation [\(Shokolenko et al., 2009\)](#page-69-3) and also the fact that mtDNA is not protected by histones. Besides the exposure to reactive oxygen species, the established mutational patterns can also be due from nutritional changes, alteration of life style and climate changes. It is known that there has been periodic migrations by archaic and modern humans, driven by climate changes and the lack of food, which contributed to the genetic diversity and variability of the different haplogroups [\(Rito et al., 2013\)](#page-69-4).

Heteroplasmy (the presence of more than one type of mtDNA genome inside the same cell [\(Stefano et al., 2017\)](#page-70-1)) occurs in 90% of the population, with up to 20% implied in certain mitochondrial diseases [\(Ye et al., 2014\)](#page-71-1), like cardiovascular diseases, diabetes and some forms of cancer [\(Dimauro and Davidzon, 2005\)](#page-65-3). The bottleneck effect is also an important mechanism of mutation management. As mentioned in the previously, heteroplasmy is a very common situation and it is associated with this mechanism, where the mutated or the wild-type (the one inherited by maternal lineage) sequences tend to be more or less replicated, according to its benefit for the cell and the individual [\(Zhang et al., 2018\)](#page-71-2).

The consecutive accumulated mutations in the germinal cells, where the mutated mtDNA is transmitted to the offspring, leads to new variants in the population and eventually to the creation of new haplogroups.

## <span id="page-18-0"></span>1.4. OTHER GENETIC STUDIES

Besides mtDNA, other genetic systems, for example Y-chromosome and autosomal data (at its highest level, whole genome data), can be used to study human population.

Y-chromosome presents the male side of the genetic history of a population, being inherited only by males and it also lacks recombination in most of the chromosome. Compared with mtDNA, it shows a much lower mutation rate. The difficulty to access the sequence diversity, associated with the low mutation rate and a very high number of repetitive regions, made it more challenging to characterize SNPs used in human population studies. In recent years though the Y-chromosome has been gaining a new breath, with more and more studies regarding its genetic history and

evolution [\(Cruciani et al., 2011\)](#page-65-4). Migration mainly dominated by males, often associated with pastoralism, are observed only in Y-chromosome variation, having left no signal on the rest of the genome or the mtDNA [\(Haak et al., 2015;](#page-66-0) [Henn et al., 2008\)](#page-66-1) .

Whole-genome analysis uses the nuclear genome information for population studies. Given the large amount of data implied in this type of studies, and the complexity, since 99.9% of the genome is shared by every human [\(Li et al., 2014\)](#page-67-3), genome-wide studies used to be very expensive and time-consuming. With the increasing interest in the past few years and the continuously decrease in sequencing prices, nowadays it is a much faster and more efficient process, with more data available to the public, that also led to the improvement of statistical method of analysis [\(Chakravarti, 2015\)](#page-64-2). When compared with Y-chromosome and mtDNA, genome-wide presents a disadvantage in the migratory records, since the nuclear DNA is subject to recombination, many population and migratory markers and other important artefacts are lost due to the admixture with the sink population [\(Rito et al., 2019\)](#page-69-5).

# <span id="page-20-0"></span>2. POPULATION STUDIES

#### <span id="page-20-1"></span>2.1. ARCHAEOGENETICS

Archaeogenetics is a relatively new discipline, that consists in the "application of molecular genetics to study the human past" with the contribution of both archaeology and genetics [\(Soares](#page-70-2)  [et al., 2010\)](#page-70-2). Although this field of study began in the 1960s with studies led by Cavalli-Sforza, observing traits like human blood groups and lactase persistence, known to be inherited characteristics, and correlating them to linguistic and ethnic groupings, it was only later that this discipline was named by the archaeologist Colin Renfrew [\(Sokal, 2001\)](#page-70-3).

The development of techniques like Polymerase Chain Reaction (PCR) and Sanger DNA sequencing in the 1980s and the next-generation sequencing techniques in the 2000s, combined with the advantages of the use of mtDNA, allowed a deep assessment of genetic variation. In recent years, it also allowed the sequencing of small and degraded DNA samples from archaeological sites (aDNA), providing a more in-depth study of ancient civilizations and giving a new strength to the archaeogenetics field [\(Marske, 2016\)](#page-67-4).

#### <span id="page-20-2"></span>2.2. PHYLOGEOGRAPHY

Phylogeography is the integration of phylogenetics and geography in the study of population history, by establishing lineages and their evolution throughout time and space [\(Avise, 2000\)](#page-63-0). The main tools used in phylogeography are phylogenetic trees, the geographical distribution of the established lineages and molecular clocks, in combination with other areas like archaeology, paleoanthropology, paleoclimatology and ethnology [\(Cruzan and Templeton, 2000\)](#page-65-5). Integrating all or some of these aspects, it is possible to shed light on the modern geographic distribution of a given species, by presenting its origin and dispersal patterns.

## 2.2.1.Phylogenetic trees

<span id="page-21-0"></span>A phylogenetic (or evolutionary) tree is a diagram that contains branches connected by nodes. The structure of the trees consists in three parts: root, node and branches. While the branches represent lineages, the nodes represent the common ancestor (real or theoretical), where the lineages started to diverge. The root, when present, is the representation of the common ancestor of all the individuals present on the tree, and it defines the directionality of it. At the end of each branch there is a terminal node, also known as leaf, that represent modern sampled sequences [\(Scott and Baum, 2016\)](#page-69-6).

As a whole, the tree is the visual representation of the evolutionary relationship between individuals. In phylogenetic trees these evolutionary relationships are based on genetic mutations or the lack of them [\(Choudhuri, 2014\)](#page-64-3).

## 2.2.2.Tree construction

<span id="page-21-1"></span>Trees can be rooted (Figure 2A) or unrooted (Figure 2B). Usually, the trees are rooted by using an outgroup: an individual that, although it is related closely to the data, it is far enough to not be represented in between the other branches. The same data can be presented without the root, but it will only show the relation between branches without indicating ancestrally [\(Choudhuri, 2014\)](#page-64-3).





There are two different methods to build the phylogenetic trees: distance-based and characterbased. For the distance-based approach, the distance between each sequence is calculated and the tree is formed. Methods such as UPGMA (unweighted pair-group method with arithmetic mean)

and Neighbour-joining (NJ) are employed. Trees generated by NJ clustering are the more common, since it generates the shortest sum of branch lengths. Even though this method has its benefits (e.g. faster calculations), character-based methods are preferable when it comes to phylogeography analysis. The character method is based on the aligned sequences organized by similarity of characters. This better presents the evolutionary processes, as it accounts for the occurrence of mutations and alterations in repetitive motifs. Maximum-Parsimony, Maximum-Likelihood (ML) and Bayesian methods are the most used character-based methods.

#### Maximum-Parsimony

In this method, the length of each branch is calculated according to the number of polymorphisms along it. The algorithm creates all possible trees and then chooses the one with the smallest branches, the one that minimizes the total number of character-state changes. This methodology requires a low computational power but tends to led to statistical inconsistencies called "long branched attraction", where two independent samples that have fast evolving characters are grouped together, regardless of their true evolutionary relationships [\(Freudenstein, 2016\)](#page-65-6).

#### Maximum-Likelihood

This method generates all the combinations to multiples unknown parameters and then, given the predefined evolutionary model, it estimates which tree is more likely to represent said data. The parameters can be branch length, sequence alignment, nucleotide substitution model, among others, and are what defines the models tested. This is a very demanding computational method [\(Dhar and Minin,](#page-65-7) 2016).

#### Bayesian Inference

This method, instead of only presenting one single tree, presents a set of trees and the probability of each, after repeated Markov Chain Monte Carlo (MCMC) simulations. This approach is also very demanding in terms of computational capacity and has been more and more used in phylogenetics and specifically in phylogeography [\(Yang, 2016\)](#page-71-3).

There are no perfect methods to create the most precise tree. For this reason, better results are obtained using an integration of various methodologies, since they complement each other and when used together may produce more reliable results.

## 2.2.3.Median Networks

<span id="page-23-0"></span>In many occasions, due to homoplasy (multiple occurrences of mutations in the same position of mtDNA), a single phylogenetic tree is not the most accurate representation of data, as loops or reticulations, can be generated, when multiple evolutionary equally parsimonious reconstructions are possible. To show the different possible evolutionary routes, structures called networks depict a better image. For mtDNA the most used are the median networks [\(Bandelt et al., 1995\)](#page-63-1).This type of network converts the variant sites to binary characters and then link the samples by their distance to each other. Reticulations can originate large hyperdimensional cubes when there is a large amount of data. To prevent this complication, a set of rules has been devised to eliminate some of the more likely occurrences, including a weighting scheme where fast sites are given a higher probability of recurrence [\(Nakhleh and Morrison, 2016\)](#page-68-2).



Figure 3 - A network diagram rendered with Network 10.0.1 and Network Publisher 1.1.0.7 software (www.fluxus-engineering.com). The yellow dots represent individuals and the red dots represent reticulations.

## 2.2.4.Founder Analysis

<span id="page-24-0"></span>Founder analysis is a quantitative method of phylogeographic analysis. It aims to identify, scale and date the migrations of a population to a new territory, by evaluating the diversity of lineages that came from a specific geographic population (the "source") and settled in a specific location (the "sink"), also dating the time that it took for this diversity to occur.

This method of analysis was firstly proposed by Richards and colleagues in 2000 [\(Richards et](#page-68-3)  [al., 2000\)](#page-68-3) and has since been used in the search of the founder lineages of the world. Although being a good method of analysis, the complex mathematical knowledge that its application requires, made it scarcely used. Recently, using the principles of the original methodology, a userfriendly program was developed, to facilitate the founder calculations [\(Vieira et al., 2020\)](#page-71-4).

Founder analysis aims to identify common sequence or clades between a hypothetical source and sink populations (Figure 4). Following that identification, the subclades in the sink are isolated and the diversity accumulated in the new regions is used to estimate the time of migration using a molecular clock.



#### Figure 4 - Representation of the Founder analysis process.

Common sequences (present or inferred) in a source and a sink population are detected in a phylogenetic tree or network. Diversity of each clade in the sink is used to estimate the time of migration.

Common mtDNA mutations and reverse gene flow can present a challenge to the founder analysis. As to surpass this problem, several criteria have been developed to select what can or cannot be considered a founder group.  $f0$  is the least restrictive criterium, as it considers all the samples in the data both source and sink. In  $f1$  and  $f2$  criteria, for a clade to be consider founder, the sequence type must display either one or two further derived branches, respectively, in the source populations.

Each founder will display a point estimate for the time of migration and a standard error for that estimate. Those parameters will allow each lineage to be statistically allocated to each predefined migration time (Figure 5) [\(Richards et al., 2000\)](#page-68-3). The migration patterns will be established through the statistical allocation of all founder clades. While the first methodology would allocate lineages into specific migrations (Figure 5.A), the researcher can also perform an unbiased scan by selection equally-distant migration times and by observing the distribution of lineages across time (figure 5.B) [\(Soares et al., 2012\)](#page-70-4).



Figure 5 - Statistical distribution of a founder type across:

A) three predefined migrations (M1 to M3) and, B) across multiple equally-spaced migration events.

## <span id="page-26-0"></span>2.2.5. Molecular Clock

The understanding of mutations and the time that they take to happen is pivotal in phylogeography, and for this a reliable molecular clock is essential since it allow us to date the divergences between branches and consequently the division of each clade and subclade. Theoretically, evolution occurs in a steady rate, making it possibly to use all the accumulated diversity to pinpoint certain occurrences in a temporal perspective [\(Bandelt et al., 2002\)](#page-63-2). Since molecular clocks allow us to estimate ages of lineages, they make possible to calculate the minimum arrival age of each founder cluster in the sink, using the before mentioned founder analysis [\(Richards et al., 2000\)](#page-68-3).

Most mathematical models behind the dating of evolutionary events assume that the mutation rate is known and constant. Most use either the coding or the non-coding region of mtDNA. One example of this is the estimated function  $1.80\times10<sup>7</sup>$  substitutions/nucleotide/year in the HVS-I section of the control region of mtDNA (non-coding), developed by Forster and co-authors [\(Forster et](#page-65-8)  [al., 1996\)](#page-65-8). Another example is the mutation rate estimated by Mishmar and colleagues of 1.26  $\times10^8$  substitutions/nucleotide/year in the coding region [\(Mishmar et al., 2003\)](#page-68-4). The approaches based on only a determined part of the mtDNA present many problems, for example, the mutation rate differs in the coding region compared to the non-coding region (the non-coding region has a ten folds higher mutation rate than the coding region) [\(Howell et al., 2007\)](#page-66-2). Other problem is that it presumes that the substitution rate is a linear relation between the accumulation of mutations and time, without taking to consideration the weight of the natural selection. It has also been shown that the oldest branches have a higher rate of synonymous mutations than younger branches. This is caused by the effect of purifying selection that occurs earlier on slightly pathogenic mutations (mostly non-synonymous) whose selective strength decreases later on as most deleterious mutations will hardly survive to older clades. This fact once again shows that a linear approach is not correct, since the short-term and the long-term mutation rate are not the same [\(Kivisild et al.,](#page-67-5)  [2006\)](#page-67-5).

A recalibrated molecular clock, that uses the entire mitochondrial genome (coding and noncoding regions), was presented in 2009 by Soares and colleagues [\(Soares et al., 2009\)](#page-70-5). They proposed a mutation rate of  $1.665 \times 10^8$  substitutions/nucleotide/year, that translates to approximately 1 mutation per 3624 years, that is further mathematically adjusted for the effect of purifying selection (Figure 6).

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Figure 6 - mtDNA mutation rate variation through time.

The black curve represents the mtDNA mutation rate and the grey line represents the interspecific (long term) mutation rate [\(Soares et al., 2009\)](#page-70-5).

# <span id="page-28-0"></span>3. AFRICAN MTDNA PHYLOGEOGRAPHY

#### <span id="page-28-1"></span>3.1. ORIGIN OF ANATOMICALLY MODERN HUMAN

The origin of the Anatomically Modern Human (AMH) has been subject of extensive studies from multiple areas, leading to the conjecture of many theories to explain the rise and dispersion of Homo sapiens.

One of the theories is known as the Out-of-Africa model. It proposes that AMH arose as a new species in 200-150 ka in Africa and later expanded to other parts of the world, replacing the existent populations of hominids. This theory implies that the archaic hominid forms did not have any genetic input in the AMH and so all human population ancestry can be traced back to Africa 200 000 years ago [\(Relethford, 2008\)](#page-68-5).

Another theory is the Multiregional evolution model, that suggests that the appearance of the AMH occurred in multiple events simultaneously across Europe, Asia and Africa, as the result of a coalescence of existing archaic populations [\(Relethford, 2008\)](#page-68-5). This theory was proposed attempting to explain how some human traits are present in all three continents without known gene flow between them, while others are clearly region-associated traits retained during the evolutionary process. To justify the apparent similarity between populations, the defenders of this model attribute it to an equilibrium of gene flow, selection and genetic drift [\(Relethford, 2001;](#page-68-6) [Wolpoff et al.,](#page-71-5)  [1984\)](#page-71-5).

The most consensual perspective of these theories is actually a combination of them, with an African origin and a variable degree of gene flow in and out of Africa during the migrations and repopulations. To truthfully pinpoint the moment that the *Homo sapiens* appeared is extremely difficult, since it was a gradual evolutionary process that lasted a few hundred thousand years, as the scientific studies prove it [\(Rito et al., 2019\)](#page-69-5).

The fossil findings of Omo I, dated 190 to 200 ka (Figure 7.1) [\(Fleagle et al., 2008;](#page-65-9) [McDougall](#page-68-7)  [et al., 2005\)](#page-68-7) and Herto, dated 154 to 160 ka in Ethiopia (Figure 7.2) [\(White et al., 2003\)](#page-71-6), among other less significant fossilised evidences, support the paleoanthropological and archaeological assumption that the AMH arose in eastern Africa, even though the oldest AMH fossil found were in fact the Jebel Irhoud remains, in Morocco, with approximately 315 ka (Figure 7.3) [\(Hublin et al.,](#page-66-3) 

[2017\)](#page-66-3). This discrepancy in data shows that the origin conclusion is yet unreliable and needs to be further studied.

In archaeogenetic analysis there is also a lack of consensus regarding the geographic location of the AMH, since different analytical methods and genetic markers present different results. Although all of them agree with the African origin, there is still a debate when it comes to specify a region in Africa or even a route for these first migratory events [\(Rito et al., 2013\)](#page-69-4) as represented in Figure 7.

Genome wide studies suggest a southern African origin [\(Henn et al., 2011;](#page-66-4) [Tishkoff et al.,](#page-70-6)  [2009\)](#page-70-6), congruent with the results of many autosomal genomic studies, that show an early divergence between Khoesan population (southern) and the rest of the african population, displaying a clear difference between northern and southern populations [\(Gronau et al., 2011;](#page-66-5) [Veeramah et al.,](#page-71-7)  [2012\)](#page-71-7). When it comes to gender-specific markers, the Y-chromosome, portrayer of the male lineage, albeit the small amount of data available, shows a central/western origin of the male lineage [\(Cruciani et al., 2011\)](#page-65-4) with a coalesce time of almost 350 ka [\(Mendez et al., 2013\)](#page-68-8), while the mtDNA, that represents the maternal lineages, has given ambiguous signals. The mtDNA lineages diverges in 2 branches: L1'6 and L0 [\(Rito et al., 2013\)](#page-69-4). L1'6 is in agreement with the origin of Ychromosome, or at least associated with the northern part of the continent, since there are no actual records of the north of Africa. This is due to depopulation that occurred 75-65 ka ago associated with climate changes, around the same time that L3 appeared and migrated out-of-Africa [\(Soares et al., 2012\)](#page-70-4). The region was repopulated again around 40-20ka from Eurasia [\(Macaulay](#page-67-6)  [et al., 1999;](#page-67-6) [Olivieri et al., 2006\)](#page-68-9). L0 shows a different story: the most probable origin of this haplogroup is in the south, where it still prevails in Khoesan populations and Bantu speakers [\(Barbieri et](#page-63-3) al., 2013; [Chen et al., 1995\)](#page-64-4). These differences make it still a mystery where AMH come from.

Mitochondrial Eve is the most recent common female-lineage ancestor and lived around 150- 200 ka ago, when the AMH population was about 10000 people. According to human genetics, Mitochondrial Eve is the point of convergence in the maternal tree of life, meaning the only past woman with an unbroken female lineage to this day [\(Cann et al., 1987\)](#page-64-5). The exact location of the Mitochondrial Eve is yet unknown due to the difficulty to pin-point it using phylogeography, since there is no deeper lineage, but the nucleotide diversity in the HVS-I region of mtDNA suggests a central African placement (Figure 7) [\(Soares et al., 2016\)](#page-70-7).



#### Figure 7 - Representation of the different hypothesis for the origin of Anatomically Modern Human.

Numbers 1, 2 and 3 represent the location of the major fossil findings. 1 - Omo I (Southestern Ethiopia); 2 - Herto (Northestern Ethiopia); 3 - Jebel Irhoud (Morocco). The circles represent the possible points of origin of AMH according to the different genetic analysis: Whole-genome and the mitochondrial haplogroup L0 point to a southern origin, while the Y-chromosome and the mitochondrial haplogroups L1'6 point a central/western origin, where it is presumed to be mitochondrial Eve's location.

#### <span id="page-31-0"></span>3.2. HAPLOGROUPS

An haplogroup consists in a group of similar haplotypes with a common ancestor. A haplotype is a set of DNA variations, or polymorphisms, that tend to be inherited together [\(Blumberg et al.,](#page-64-6)  [2016\)](#page-64-6). The first haplogroups to be classified were A, B C and D in Native Americans [\(Torroni et](#page-70-8)  [al., 1993\)](#page-70-8), and from then, the next haplogroups found were named in alphabetic order. Nomenclature of the haplogroups consists in capital letters and the subsequent subclades are named by alternation between numbers and lower-case letters. It is important to note that in some cases the haplogroup is an offshoot of another and does not obey the normal naming rules, for example the haplogroup M and N diverge from L3.

The most ancient haplogroups are located in Africa (LO'1'2'3'4'5'6) and all lineages found outside of Africa belong to L3, a strong argument for the out-of-Africa expansion theory. L3 divides in M and N outside the African continent, both present in Asia, Oceania and the Americas, but in Europe only N is present [\(Richards et al., 2016\)](#page-69-7). U is derived from R that is derived from haplogroup N and has currently a large representation in Europe and in North Africa, mainly due to the back-flow of populations [\(González et al., 2007\)](#page-65-10). In 2009, Soares et al. proposed a global mtDNA phylogenetic tree considering the updated molecular clock [\(Soares et al., 2009\)](#page-70-5). In it, it is possible to visualize the clear separation between within and outside African haplogroups.

#### <span id="page-31-1"></span>3.3. EVOLUTION AND MIGRATIONS

The different African haplogroups arose as a result of evolutionary forces, such as migrations, climate changes, and many others. Here they will be described according to their order of appearance, according to the estimated time of emergence for each clade: L0, L1, L5, L2, L6, L4, L3, clustered in L0 and L1'6, based on the works of Behar et al. [\(Behar et al., 2008\)](#page-64-7), Rito et al. (Rito [et al., 2013;](#page-69-4) [Rito et al., 2019\)](#page-69-5), Rosa and Brehem [\(Rosa and Brehem, 2011\)](#page-69-8), Salas et al. (Salas et [al., 2002\)](#page-69-9) and Soares *et al.* [\(Soares et al., 2012;](#page-70-4) [Soares et al., 2009;](#page-70-5) [Soares et al., 2016\)](#page-70-7). The phylogenetic tree of the Human mitochondrial DNA according to Soares et al. is represented in Figure 8.

The superhaplogroup L started to differentiate around 180-190 ka. Around 150 ka, L0 was the first branch to arise in sub-Saharan Africa. It comprises the subhaplogroups L0a'b'd'f'g'k. L0d was the first to appear, approximately 100 ka, and together with L0k, they make over half of the

maternal genetic pool. They are present mainly in Khoesan population of South Africa, but also in the click-speaking population of Tanzania and Angola. It was believed that L0f and L0a'b'g appeared in East Africa 80-90ka, but according to Rito et al. in 2019, this clade arose in the south and then migrated to East Africa around 70ka, where it remains to this day. Present at a smaller scale, L0a arose around 40-45ka in eastern Africa, although currently is spread all over the continent. This subhaplogroup has relatively short branches, indicating a recent evolution and population growth.

L1 separated from the remaining L1'6 between 140 and 150 ka and later branched in L1c and L1b. L1c coalesced 80-85 ka in central and western Africa. It has a 70% prevalence in the Pygmy maternal legacy and also 18-25% in the Bantu population in Angola, demonstrating a possible common ancestor between these populations within L1c. L1b has a similar origin point, although more turned to the coastal area. To note that this subhaplogroup has a very recent coalescence time, around 10 ka, indicative of a recent evolution and growth.

L5 has been previously classified as L1e. It appeared around 120 ka and it is observed in a very low frequency. It is originally from eastern Africa and has a noticeable gene flow to Pygmies and the Fali population located at the north of Cameroon.

L2 split from the main branch around 90 ka. Together with L3, it composes of 70% of the sub-Saharan maternal variation. This haplogroup subdivided in L2a'b'c'd'e. L2a is the most frequent and more widely spread haplogroup in Africa, going from the Tuareg population of Nigeria and Mali, through the Fali population, the Pygmies of Gabon and the Bantus of Mozambique. With a coalescence time of approximately 50 ka, L2a is hypothesised that it has a central African origin and then, starting 10ka migrated to the rest of the continent via the better conditions with the onset of the Holocene [\(Silva et al., 2015\)](#page-69-10), followed by the Bantu expansion and later the trans-Saharan slave trade. It is also the most common sub haplogroup present in the African American population, due to the Atlantic Slave Trade that occurred in the  $18<sup>th</sup>$  century, with slaves mainly from West Africa. The rest of the subhaplogroups coalesce around 60 ka and have a western central point of origin and still remain in this region.

L6 is a recent branch, with a coalescence time close to 20 ka, even though the divergence of the main branch is dated around 105 ka. It is mainly found in Ethiopia and Yemen, which leads to a probable point of origin in the eastern region, although it is still unknown.

L4 split from L3 around 85 ka, in north-eastern Africa. Nowadays it isn't very common. The subhaplogroup L4b coalescence time is very close to the haplogroup appearance, and it is mainly located in Tanzania and Ethiopia, much like L4a, also present in Ethiopia and in Sudan, but with a more recent coalescence time, close to 55 ka.

L3 appeared around 70 ka in eastern Africa and it is now widely spread in the entire continent. Some subhaplogroups of L3 can be linked to specific regions of Africa. L3h appeared 65 ka and is located in the northwest and the southeast of Africa. L3b'd appeared after, around 60 ka, and it is found mainly in central/south Africa, particularly Rwanda. L3f as a coalescence time of 50 ka and is found all over the continent, while L3e, with 40 ky, is found mainly in the central/south, but also has a small representation in the north of about 5%. This haplogroup is the origin of the other haplogroups present all over the world. Around the time of coalescence, it started to migrate outof-Africa through the Middle East, branching out in M (60 ka) and N (70 ka), that populated the rest of the planet. Due to backflow, M and N are also present in Africa.



#### Figure 8 - Human mitochondrial DNA phylogenetic tree.

The first Anatomically Modern Human (mitochondrial Eve) arose in Africa around 200 ka and colonized the rest of the world in a single exit out of Africa, carrying haplogroup L3. Regions are differentiated through different colours and shapes and years are expressed in ka [\(Soares et al., 2009\)](#page-70-9).

## 3.3.1.Causes and routes

<span id="page-35-0"></span>It is known that there were periodic migrations of AMH mainly due to climate changes. Climate affects and determines the existence of plants and animals, necessary for human sustenance and is often appointed as the main contributor for the spread of *Homo sapiens* and its diversification. These factors are also associated with a phenomenon called bottleneck, a sharp reduction in population size which leads to a drastic reduction of genetic diversity that have caused diversity to be reduced at some points in the past [\(Soares et al., 2016\)](#page-70-7).

Given the importance of the climatic changes, the migrations described next will be divided in periods known as MIS (Marine Isotopic Stage), based on the book *Africa from MIS 6-2* (Stewart [and Jones, 2016\)](#page-70-10), Blome *et al* [\(Blome et al., 2012\)](#page-64-8) and Rito *et al* [\(Rito et al., 2013\)](#page-69-4). These climate changes caused the intermittence of conditions in Africa, varying between the different regions, with periods of extreme drought to period of high levels of humidity and ice.

### MIS 6

This period occurred between 190 ka and 130 ka. It was probably in this MIS that the AMH and the mtEve appeared and the haplogroups L0 and L1'6 arose.

The first migration registered was from central Africa (assuming a point of origin of the mtEve) to south, were L0 arose, around 150 ka. While L1 stayed in western Africa, L2'6 migrated to east Africa, 130 ka (Figure 9.A).

This period is characterised as glacial, with refugees in west Africa and, also known as tropical, and in the south, where the climate changes were not as intense, with intermittent periods of both dry and wet conditions, as is represented in Figure 10.

#### MIS 5

Between 130 ka and 70 ka, this period was known as the Warm Interglacial Period. It was in this time frame that L2 arose in the west after a migration from the L2'6 cluster in the east ( $\tilde{ }$ 80ka) and the L5 branch diverged in the east (120 ka), according to Soares et al. [\(Soares et al.,](#page-70-7)  [2016\)](#page-70-7) (Figure 9.B).
The haplogroup L0 started to branch out, with the subhaplogroups L0a'b'f'k possibly traveling from the south to the east, starting in the beginning of this MIS, and only two of them (L0a'b) arriving to eastern Africa nearly at the end of this period (76 ka), according to [\(Soares et al., 2016\)](#page-70-0). This scenario was recently revised, however, with the migration from South to East Africa only taking place around 70 ka [\(Rito et al., 2019\)](#page-69-0).

The Saharan region was, at this time, a "green" environment, with favourable condition for the migratory periods. The eastern region was a very desirable destination, due to the increase of the temperature and a reduction of the moisture levels, creating a very suitable place for growing resources and for population growth (Figure 10).

### MIS 4

Lasting only 10 ky, between 70 ka and 60 ka, it was in this period that the out-of-Africa migration of L3 occurred, granting it a very important place in AMH history, as is shown in Figure 9.C. While the Out-of-Africa Migration involved only the East African L3 haplogroup, influences from the South (in the form of the before-mentioned migration of L0) might have been crucial in starting this expansion [\(Rito et al., 2019\)](#page-69-0).

In this MIS began the Last Glacial Period (LGP), that lasted until MIS 2. As it is seen on Figure 10, the climate above the Sahara was extremely arid, causing this region to become desertified and depopulated. In contrast, the sub-Saharan regions had high moisture levels, with particular the southern part of the Nile Valley, close to the Horn of Africa, the point of exit for the out-of-Africa expansion, was considered a refugee, with permanent bodies of water. In the south, a region known as Namaqualand, in Namib, had high population density [\(Viehberg et al., 2018\)](#page-71-0).

#### MIS 3

Lasting from 60 ka to 30 ka, it was in this period that the AMH started to populate the rest of the world. In the Middle East (southwestern Asia), L3 started to branch into M and N, this one later diverging in R and U. These haplogroups, particularly sub-branches of M and U, M1 and U6 respectively, repopulated the north of Africa, in a process known as backflow, nearly at the end of MIS 3. Since these populations had some level of mtDNA differentiation to the rest of African haplogroups, there is a clear distinction between the sub-Saharan and the Mediterranean coast populations.

Climatic conditions in the west improved considerably, making it more suitable for human occupation, and L3b'c'd migrated to this area around 50ka, and later on L3e travelled the same route, 37ka (Figure 9.C).

#### MIS 2

This period began 30 ka and lasted until 15 ka. At the end of the LGP, it was at this time that the Last Glacial Maximum (LGM) was reached, meaning that the ice-sheets had achieved the maximum growth in Europe. In terms of migration this period seems to display little movement of people.

### MIS 1

This period began 15 ka and it lasts until the current days. The Holocene, the current geological period, started approximately 11,5 ka. It is characterized by a general improvement of weather and relative moisture conditions [\(Weldeab et al., 2007\)](#page-71-1). The Sahel Belt, located between the Saharan desert and the tropical forests, became a humid and fertile landscape at this time and until 6 ka, allowing the formation of migratory corridors, North/South and East/West [\(Triska et al.,](#page-70-1)  [2015\)](#page-70-1). In this period migrations of sub-Saharan lineages moved from West, Central and Eastern Africa into Northern Africa crossing the Sahel belt and ultimately reaching Europe.

The beginning of MIS 1 was a period of a large-scale populational expansion, not only in Africa, but all over the world, with particular importance in the Southwest of Asia and in Europe. The improvement of the climatic conditions let to a recolonization of these regions in the early Holocene, with the majority of the migrations being through the Middle East, except the Iberian Peninsula that received a large inflow of migration via the Strait of Gibraltar [\(Cerezo et al., 2012;](#page-64-0) [Hernández et](#page-66-0)  [al., 2015;](#page-66-0) [Skorecki and Behar, 2013\)](#page-70-2). In this same period, in the last few millennia, two major migrations to place in Arica: The Bantu expansion and the trans-Saharan slave trade.

The expansion of Bantu speakers is believed to have started from Nigeria/Cameroon where African populations developed agricultural practices. It is believed that they have taken two main routes to Southern Africa: through the West, along the coast towards the South, to Angola, South Africa and Botswana, around 3.5 ka; and to East, towards the Great Lakes, reaching Uganda approximately 2.5 ka and later Mozambique (1.8 ka) (Figure 9.E). This migration caused a forceful retreat of other populations: San, a population of hunter-gatherers known for their click consonants, went further south, into the Kalahari Desert, becoming even more isolated, and the Pygmies eventually adopted the Bantu language [\(Silva et al., 2015\)](#page-69-1).

L2 became widespread in the continent around this period, in West, Central and Southeast Africa associated with the Bantu expansion and in the northwest due to the trans-Saharan slave trade. The trans-Saharan slave trade happened after the repopulation of the Sahara, in early to mid-Holocene, when the weather conditions where at its best, and provided a significant flow between North/South and West/East Africa, also associated with the haplogroups L1b and L3e5 [\(Soares et al., 2016\)](#page-70-0) (Figure 9.D). Also known as the Arab trade, this route was used to transport, besides slaves, gold and salt between the North of Africa, the Persian Gulf and the sub-Saharan region [\(Kehinde, 2014\)](#page-67-0).

A part of the North-eastern African population later, 1500 years ago, migrated to Europe, in particular to the Iberian Peninsula, through the Strait of Gibraltar [\(Cerezo et al., 2012;](#page-64-0) [Skorecki](#page-70-2)  [and Behar, 2013;](#page-70-2) [Soares et al., 2010\)](#page-70-3). This major migration is also known as the Muslim Conquest, that left a major imprint in Iberia, not only genetic but also cultural [\(Botigué et al., 2013\)](#page-64-1). From then on, with the discovery and conquest of the New World, African migration as gained a whole new meaning. African populations were forcefully taken to Europe and the Americas as slaves around 500 years ago and, more recently, in search of better life conditions, migrated to the whole world.

The phylogeographic analysis across the last 200 thousand years allows for a complete understanding on the origin and geographic distribution of each of the African clades (Fig. 11) which will allow the tracing of each sequence outside Africa to an African region.





al. in 2019. Adapted from Soares et al. [\(Soares et al., 2016\)](#page-70-0)



Figure 10 - Summary of the African climate data from 150 to 30 ka, divided in the different regions. Adapted from [\(Blome et al., 2012\)](#page-64-2)



Figure 11 - Distribution of the major mtDNA haplogroups across Sub-Saharan Africa. Figure obtained from [\(Silva et al., 2015\)](#page-69-1).

### 4. AFRICAN INFLUENCES

### 4.1 SLAVERY

Enslavement became common around 11 ky [\(Hellie, 2020\)](#page-66-1) in ancient civilizations, such as the Ancient Egypt, Ancient Greece and Ancient China that made use of these people to build their great architectural endeavours among many other tasks. At this time, slavery was used as a form of punishment for crime and other reasons [\(Harris, 2012\)](#page-66-2).

Although not being a new practice, slavery gained new breath in the  $15<sup>m</sup>$  century, when Portugal began to explore the African coast, mainly the central and western regions. In the  $18<sup>th</sup>$  of June of 1452 the Pope Nicholas V issued a papal bull named *Dum Diversas* that granted the Portuguese King Afonso V the authorisation to consign people that did not believe in Catholicism to "perpetual servitude". This authorisation was later extended to other countries, including the Americas in 1493. This was the trigger to the begin of the Slave Trade and the Colonialism. With time, the African population in the colonizing countries increased immensely, for example 10% of the population living in Lisbon in the mid-16<sup>th</sup> century was of African ascendance, arriving via de Atlantic Slave Trade [\(Earle and Lowe, 2006\)](#page-65-0).

The first country to lawfully abolish slavery was Portugal in 1761 and later in its colonies, in 1869, but continued in many other countries (Figure 12). After this, in the  $19<sup>th</sup>$  century there was an effect called as "Back to Africa", where descendants of slaves decided to go back to the ancestor's countries, mainly coming from the Americas to western Africa.



Figure 12 - Vintage engraving of a mother and daughter sold at Slave Auction, Southern USA in the 19th Century.

From: https://www.history.com/topics/black-history/slavery

### 4.2 THE ATLANTIC SLAVE TRADE

The Atlantic Slave Trade took place between the  $16<sup>th</sup>$  and the  $19<sup>th</sup>$  Century and was responsible for the transportation of over 10 million enslaved Africans to the Americas and Europe. This route was also used to transport textiles, arms and wine from Europe to Africa and sugar and coffee from the Americas to Europe, as shown on Figure 13.

Portugal was the first country to use this route to transport both slaves and supplies between Europe and its main colony, Brazil, followed by the United Kingdom, the North America and many others. The route consisted of three points: Africa (in particular the central and western regions, and in a much smaller scale the region of Mozambique, also a Portuguese colony), Europe (where the main slave markets were, for example Lisbon and Faro) and the Americas (Brazil, North America and Caribbean) [\(Thornton, 1998\)](#page-70-4).



#### Figure 13 - Representation of the Atlantic Slave Trade.

Besides the transportation of slaves, this route also transported sugar, cotton, arms, textiles and wine, among other things, from and to the colonies.

The main ports used to carry out the slaves from Africa to the other continents were located in Western Africa, with two others in Southern Africa. Figure 14 represents the main ports locations, as well as the colonizing countries of the African regions.





The arrows point to the main ports used to carry out the slaves. Each colour represents a different colonizing country.

#### 4.3 GENETIC INFLUENCE OUT-OF-AFRICA

Although over 60% of the slaves transported to the Americas were male, previous studies have presented a sex bias towards female presence, due to the multiple accounts of sexual exploitation [\(Micheletti et al., 2020\)](#page-68-0). While in the former British colonies, as the Caribbean and the USA, slaves were forced to have multiple children, either to maintain the workforce to a maximum [\(Marable,](#page-67-1)  [2015\)](#page-67-1) or after given false promises of freedom [\(Deyle, 2005\)](#page-65-1), in South America the European women were encouraged, including via payment, to marry and have children with the more lightskinned slaves, as an effort to weaken the African traces of the population, a practice known as "Branqueamento" [\(Meade, 2016\)](#page-68-1). As a result, the African descendance is much more visible in the USA, also due to more recent events, such as the segregation period [\(LaVeist, 1993\)](#page-67-2), while in South America we can observe a distributed presence of african ancestry, for example in Brazil, 28% of the "mostly white" mtDNA sample is of African origin [\(Alves-Silva et al., 2000\)](#page-62-0).

When it comes to pinpoint the original African region from which the slaves were exported, there is vast evidence of mostly western-central descendance in countries such as Brazil, in agreement with the known trafficking routes. In the USA there is an overrepresentation of Nigerian descendance, mainly due to the migration within the Americas but also because slaves from this part of Africa were much more recently transported. The more expected origin would be western-central, as in South America, but since this was an earlier migration, with a high rate of mortality, there is a much smaller representation in the current population [\(Micheletti et al., 2020\)](#page-68-0).

In Europe there is an absence of a visible African ancestry, because it was only a negotiation place, not retaining a large number of slaves. In Portugal there is a prevalence of haplogroups associated with western Africa [\(Pereira et al., 2000\)](#page-68-2) and haplogroups from the north, such as U6, most likely linked to the geographic proximity between Iberia and Morocco, demonstrating the Arab influence in Portugal [\(Salas et al., 2004\)](#page-69-2).

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The work here presented had three objectives:

- 1) To reconstruct and update the African mitochondrial DNA tree, representing the female line of descent of the African populations around the world;
- 2) To determine the phylogeography of mtDNA in the African continent, taking into account the multiple processes with demographic impact
- 3) To understand the source and time of migration of the African lineages outside Africa and correlate this information with the literature on slavery and the Slave Trade market.

Ultimately, these three objectives combined, allow to determine the founders carried during the slave trade from Africa to Europe and the Americas and also to trace the most probable source in Africa of each founder, that end up being the major goal and outcome of this thesis.

### 1. DATASET

A dataset of both published and unpublished complete mtDNA sequences was compiled. This includes samples from NCBI and the 1000 Genome Project [\(Auton et al., 2015\)](#page-62-1) but also unpublished samples generated in collaboration with the University of Huddersfield. In total, the dataset had 6759 samples. A thousand and one samples were excluded due to low quality, to being generated from individuals with mitochondrial disorders or being part of clinical studies (e.g., cancer research). At the end, the dataset was comprised of 6658 L-type sequences samples. A full list of the dataset is presented in Table S1 of Supplementary File 1.

The variations in the whole mtDNA samples were scored as variants when aligned with the revised Cambridge Reference Sequence (rCRS), then the samples were assigned to the respective haplogroup, considering each sequence variation, using the online software HaploGrep 2.0 [\(Weissensteiner et al., 2016\)](#page-71-2).

### 2. PHYLOGENETIC RECONSTRUCTION

A phylogenetic tree was constructed using a reduced median algorithm of the software Network 10.0.1 and visualized in Network Publisher 1.1.0.7 add-on from Fluxus-Technology Ltd. In order to ease the visualization and reduce the amount of reticulations, mutations with high homoplasy were down-weighted. By default, Network 10.0.1 weights each position in 10, but positions 146, 150, 152, 195, 709, 16093, 16129, 16189, 16311 and 16362 were weighted down to 7. The polymorphisms 308-315, 310C, 3107N, 515-522, 16182C, 16183C, 16189, 16192C, 16518T and 16519C, along with insertions and deletions were removed, since they are described as unpredictable, very fast or poorly sequenced, meaning that they are unreliable for phylogenetic inferences.

To help make sense of the over-reticulated networks visualized, choices were made regarding the evolutionary paths: sequences were compared to a complete mtDNA tree available online, from Phylotree [\(van Oven and Kayser, 2009\)](#page-70-5), and positions were checked in a reference table for relative

frequency of each mutation from *Soares et al* [\(Soares et al., 2009\)](#page-70-6). Preference was given to rarer mutations in the basal branches. Other criteria, was considered, e.g. the number of individuals in each node, do define the pathway to follow.

After assembly, the trees were transcribed to Extensible Markup Language (XML), to be further analysed by founder analysis software [\(Vieira et al., 2020\)](#page-71-3).

### 3. FOUNDER ANALYSIS

After the XML transcription, the samples were assigned as source, sink or undetermined, according to the models to test. Samples were geographically classified as:

Iberia: Samples were included in "Iberia" if labelled as Portugal and Spain.

Europe: Samples were included in "Europe" if labelled as Bermuda, Bulgaria, Cyprus, Czech Republic, Denmark, England, France, Germany, Greece, Hungary, Ireland, Italy, Netherlands, Poland, Russia, Serbia, Slovenia, Sweden, Switzerland and Wales

North America: Samples were included in "North America" if labelled as Barbados, Puerto Rico and USA

South America: Samples were included in "South America" if labelled as Bolivia, Brazil, Colombia, Dominican Republic, Mexico, Paraguay and Peru

Southwestern Asia: Samples were included in "Southwestern Asia" if labelled as Iran, Israel, Jordan, Kuwait, Lebanon, Oman, Saudi Arabia, Syria, United Arab Emirates, Yemen, India and Pakistan

Africa: Samples were included in "Africa" and subdivided in regional sections if labelled as North Africa: Algeria, Egypt, Libya, Mauritania, Morocco, Sahrawi Arab, Tunisia; Western Africa: Benin, Burkina Faso, Cabo Verde, Ivory Coast, Gambia, Ghana, Guinea-Bissau, Guinea, Liberia, Mali, Niger, Nigeria, Senegal, Sierra Leone, Togo; *Eastern Africa:* Comoros, Djibouti, Ethiopia, Eritrea, Kenya, Madagascar, Mauritius, Rwanda, Seychelles, Somalia, South Sudan, Sudan, Tanzania, Uganda; *Central Africa*: Burundi, Cameron, Central African Republic, Chad, Congo, Democratic Republic of Congo, Equatorial Guinea, Gabon, São Tomé e Principe; South Africa: Angola, Botswana, Lesotho, Malawi, Mozambique, Namibia, South Africa, Swaziland, Zambia and Zimbabwe. The following migration models were considered for the founder analysis:

	Test 1		Test 2		Test 3		Test 4		Test 5	
	Source	Sink								
Africa	∧		↗		⋏				∧	
Europe		Χ			Χ					
Iberia		χ		Χ	Χ		∧			
<b>North America</b>										
<b>South America</b>										
Southwestern Asia					Χ					

Table 1 - Migration models used in Founder analysis. 5 tests were performed, each with a different source-sink combination.

The founder analysis was performed by an in-house developed software [\(Vieira et al., 2020\)](#page-71-3). Migrations were computed using the specific times of 500 years and 8000 years, the first correlated to the timeframe of the Slave Trade, and the other to accommodate all the other periods of migrations. The mutation rate was defined as 2643 years/mutation, based on studies of the African migrations to Iberia [\(Hernández et al., 2015\)](#page-66-0) and in agreement with the curve described by Soares et al [\(Soares et al., 2009\)](#page-70-6).

The founder analysis was run in two fashions. First, we considered an unbiased analysis where founders were statistically allocated to equally-spaced migration events from 0 to 20,000 years, with hypothetical migrations every 200 years. This will allow to perform a scan of probable periods of migrations by checking periods with an increased probability of multiples founders entering at that period.

Following this scan, a model of migration will be established with specific migration times. This model will take into account not only the migration scan performed previously but also combining with information from history, archaeology, anthropology and palaeoclimatology.

In order to visualize the geographic distribution outside Africa of L lineages associated with each of the migration in our demographic model, we constructed frequency distribution maps with Surfer® v.8 (Golden Software) using Kriging algorithm. We could only use datasets that corresponded to population data, and given this, the software mostly comprised data from the 1000

Genomes Project, the HGDP and the newly data generated in Huddersfield with a few other available cases [\(Derenko et al., 2013;](#page-65-2) [Matisoo-Smith et al., 2018\)](#page-67-3). A map of the datapoints is displayed in Figure 15.



Figure 15 - Datapoints to be used in the frequency maps.

## **RESULTS**

With the intent of understanding the timeframe of migration events from Sub-Saharan Africa into different regions, a Founder Analysis was performed scanning for the probabilistic distribution of founders across time (Figure 16). African population was assumed as the source population for all of the tests, with the addition of Southwestern Asia for Europe, Iberia, North and South America and Europe and Iberia for North and South America.





The plots show probabilistic distributions of founder clusters across migration times for population expansion of mtDNA L lineages into Southwestern Asia (A), Europe and Iberia (B) and North and South America (C). Africa is assumed as the source population against Southwestern Asia, Africa and Southwestern Asia against Europe and Iberia, and Africa, Southwestern Asia, Europe and Iberia against North and South America.



Figure 16 (continuation) - Founder analysis for mtDNA L haplogroup.

The plots show probabilistic distributions of founder clusters across migration times for population expansion of mtDNA L lineages into Southwestern Asia (A), Europe and Iberia (B) and North and South America (C). Africa is assumed as the source population against Southwestern Asia, Africa and Southwestern Asia against Europe and Iberia, and Africa, Southwestern Asia, Europe and Iberia against North and South America.

Southwestern Asia (Figure 16A) presents two very slight peaks, at 11000 years and at 5000 years and from 3500 years on the migratory probabilities increase exponentially till the present.

Europe and Iberia in specific (Figure 16B) have a similar dispersal between them as expected, with a peak at 6000 years and a more recent one, at approximately 1000 years, this last one more prominent in Iberia.

The Americas, both North and South, (Figure 16C) only present a rapid increase of migration probabilities in more recent years, which demonstrates that there was only recent migration of the haplogroup L to this continent.

Taking these results into consideration and taking into account known periods of population movements previously suggested from genetics, archaeology and palaeoclimatology, we selected two periods of migration in order to probabilistically part our data, one at 8000 years corresponding to the Holocene climatic optimum where movements likely occurred from Sub-Saharan Africa into North Africa and Europe, and one at 500 years, marking recent migrations, which includes the Atlantic Slave Trade. It is important to point out that more ancient migrations (before 10,000 years) as suggested before, will probabilistic be allocated into the 8000 years partition as well as more recent migrations older than 5,000 years as obtained in the scan for Arabia. In simpler terms, these partitions aim to split prehistoric migrations from historical ones.

Over 60% of L lineages found in Southwestern Asia are linked to recent migrations and only close to 40% are associated with Postglacial movements, as shown in Figure 17. Europe shows a very similar distribution of recent lineages and Postglacial while in Iberia it is possible to see a clear distinction between the recent migrations, that correspond to 67% of lineages, and the Postglacial movements. As expected, the Americas show a majority, over 85%, of recent lineages. The detection of postglacial lineages in North and South America is considered irrelevant, given that it is known that it was impossible at that time for migrations to occur between these continents and the percentage is mostly the results of statistical residues (large confidence intervals) from lineages that are mostly statistically allocated to modern periods.



Proportion of mtDNA Founder lineages

Figure 17 - Proportion of mtDNA L founder lineages in Southwestern Asia, Europe, Iberia, North America and South America.

Two migration periods were considered, taking into account the results of Figure 12: 500 years, that corresponds to recent migrations, and 8000 years, associated with the Postglacial Period.

As an attempt to understand where these lineages came from, a phylogeographic analysis of the source samples' locations was performed for founders statistically allocated to either recent or prehistoric migrations, with exception of North and South America, that only have recent migrations.

## POSTGLACIAL PERIOD



Figure 18 - Proportion of the point of origin of the lineages detected in the Founder Analysis at the Postglacial Period in Southwestern Asia (A), Europe (B) and Iberia (C).

Figure 18 presents the percentage of lineages from the different source regions that were detected at the Postglacial Period, 8000 years ago. Figure 18A shows the Southwestern Asia sink population, with source population from Africa, and has an overwhelming percentage of 82,1% came from Eastern Africa, 7,4% from Central Africa, 7,4% from Central Africa and 3,2% from Western Africa. Most common lineages include L0a2a2a+16188G, L2a1+16189C+143A+16309G+ 16192T, L3d1a1a, L3e3a and L3f1b+16292T. Figure 18B shows Europe sink population, with source populations from Africa and Southwestern Asia. Western Africa represents 40,3%, North Africa 29%, Central Africa 19,4%, Eastern Africa 6,5% and Southwestern Asia 4,8%. It mostly includes L1b1a lineages (L1b1a+185C+14016A, L1b1a+189G, L1b1a6 and L1b1a+1462A), L3f1b and L3d1b1. Figure 18C shows the Iberian sink population with source populations from Africa and Southwestern Asia. The majority of lineages came from North Africa, 53,3%, 40% from Western Africa and at last the Southwestern Asia with 6,7%, mostly represented by L1b1a6 and L3f1b. To note that none of the figures above show lineages from South Africa.

### RECENT



Figure 19 - Proportion of the point of origin of the lineages detected in the Founder Analysis at the Recent migrations (500 years ago) in Southwestern Asia (A), Europe (B), Iberia (C), North (D) and South (E) America.



Figure 19 (continuation) - Proportion of the point of origin of the lineages detected in the Founder Analysis at the Recent migrations (500 years ago) in Southwestern Asia (A), Europe (B), Iberia (C), North (D) and South (E) America.

Figure 19 presents the percentage of lineages from the different source regions that were detected at the Recent Migration Period, 500 years ago, mostly associated with the Slave Trade. Figure 19A shows the Southwestern Asia sink population, with source population from Africa, and, as in the Postglacial period, the majority of the lineages is from Eastern Africa, 58,1%. Western Africa represents 12,4%, Northern Africa 11,6%, Central Africa 9,3% and Southern Africa 8,5%. No lineage displays a substantially higher frequency. Figure 19B shows the European sink population, with African and Southwestern Asia populations as source. Western African lineages are the most represented, with 54,2% followed by Northern Africa, with 14,5%, Southwestern Asia, with 9,6%, Eastern and Southern Africa with 8,4% each and Central Africa with 4,8%. The most common lineage at around 10% is L2a1l2+143A. Figure 19C shows the Iberian sink population, with Africa

and Southwestern Asia as source populations. Western Africa represents 58,3% of the lineages, while Southern Africa represents 16,7%, North Africa 14,6% and Eastern Africa 10,4%. No specific lineage shows an increased frequency. Figure 19D shows the North American sink population with Africa, Southwestern Asia, Europe and Iberia as the source populations. Western Africa has the most lineage representation, with 73,4%, followed by Southern Africa, with 13,9%, then Eastern Africa, with 7%. Europe represents 2,4% of the lineages, Central Africa 1,6%, Northern Africa 1,4% and Southwestern Africa 0,4%. Most common lineages are L1c3b2+16086C+16104T+14929T, L2a1f+16192T, L2a1f1, L3e2b+152C and L3f1b1a, all from a probable Western African source. Figure 19E shows the South American sink population, with Africa, Southwestern Asia, Europe and Iberia as the source populations. Southern Africa represents 41,2% of the lineages, Western Africa represents 35,3%, Northern Africa represents 8,8%, Iberia and Central Africa represent 5,9% each and Eastern Africa represents 2,9%. Most common lineages include L1c3b1a and L0a1b+5563A, most common in Angola and Zambia in the African dataset.



Figure 20 – Maps of geographic distribution frequencies of L lineages outside Africa associated with each of the migration in our demographic model.

(A) Prehistorical arrivals, at 8000 years and (B) Historical arrivals, at 500 years.

In terms of frequencies across Europe and Asia (Figure 20), it is possible to observe that considering both prehistorical (Figure 20A) and historical (Figure 20B) migration, the higher frequency is in Southwestern Asia. The Iberian Peninsula shows substantial frequencies of L sequences from both periods. However, the remaining Mediterranean Europe mostly shows lineages associated with prehistoric migration.

## **DISCUSSION**

This worked aimed to apply a phylogeographic approach to understand the presence of mtDNA lineages of African lineages outside Africa. In particular, we applied a methodology called founder analysis, that aims at establishing migration episodes and quantify the number of lineages involved in those migratory episodes. By studying the movement of sub-Saharan mtDNA L lineages into several parts of the world, namely North America, South America, Europe, Southwestern Asia and South Asia, it is possible to understand the performance of the founder analysis methodology, as in some particular cases, mostly North and South America, the migratory model is mostly known, related with the transatlantic slave trade, all related with the last few centuries.

In the Postglacial period, more specific at 8000 years ago, the climatic conditions in Africa were at the optimal point, particularly in the Sahel Belt. This region, which includes the Sahara Desert, became a migratory corridor, allowing populations from the sub-Saharan region to arrive to the North of Africa, and from there to Europe and Iberia [\(Triska et al., 2015\)](#page-70-1). This phenomenon explains the majority of the results shown above in Figure 18. The high prevalence of Eastern African L lineages in Southwestern Asia (Figure 18A) is associated with the geographical proximity, where gene flow has occurred in both directions for most prehistorical and historical periods (Skorecki [and Behar, 2013\)](#page-70-2). In the case of Europe (Figure 18B) there is a high percentage of lineages that trace to Northern Africa but mostly from Western Africa and from Central Africa. In general, while not all lineages left traceable descendants in North Africa, the three are basically representing the same migration event - during the Holocene improvements of the conditions in the African Sahel, when populations moved from Western and Central Africa into North Africa [\(Rosa](#page-69-3)  [and Brehem, 2011\)](#page-69-3) and then into Europe. The higher percentage of Western lineages reflects the easier crossing into Iberia, due to short distance in the Strait of Gibraltar. This trend is highlighted in the analysis considering only Iberia as the sink where all lineages basically fall into Western and Northern Africa (Figure 18C). Nevertheless, African lineages are observed on this period across most of Mediterranean Europe, leaving small traces of those events until the present [\(Cerezo et](#page-64-0)  [al., 2012\)](#page-64-0).

Recent lineages, that in the Founder Analysis were represented by a migration at 500 years, are the result of migrations and gene flow at a time of globalization, with easier means of transporttation and most importantly transportation through the land was not the most probable means of travel in most cases. It was at this time that occurred the Trans-Atlantic Slave trade, that carried over 10 million people out of Africa into the Americas and, in a smaller percentage, to Europe [\(Earle and Lowe, 2006;](#page-65-0) [Salas et al., 2004;](#page-69-2) [Thornton, 1998\)](#page-70-4). As so, most of the lineages found are associated with the origin of the slaves and the place where they embarked. It is known that most ports were located in the Western coast of Africa, and only two were located in the South, more particularly in Angola and Mozambique (Figure 14), hence the majority of the lineages found in Europe, Iberia and the Americas having their founder located in the West of Africa. As observed in the Postglacial Period, the geographical proximity of Eastern and Northern Africa still has a large impact in many lineages found in Southwestern Asia, Europe and Iberia, respectively, as it is expected that continuous gene flow occurred in the last few thousand years [\(Botigué et al.,](#page-64-1) 2013; [Pereira et al., 2000\)](#page-68-2). Nevertheless, the Western source dominates the analyses showing that the Slave Trade was likely the major source of L lineages in Europe and Iberia.

One source that was absent in the analyses for Postglacial lineages but emerges in recent migrations is Southern Africa, mainly associated with the Slave Trade, since European countries had colonies there that were used as a source of slaves. Iberia, in particular, has a higher percentage of South African lineages when compared to Europe as a whole. This is linked to the fact that the Portuguese colonies in Africa, Angola and Mozambique, located in South Africa, with the exception of Guinea-Bissau, in the West. The main ports of slave commercialization in Europe were in Portugal - Lisbon and Faro - leaving behind an important imprint of lineages in the Iberian population [\(Earle and Lowe, 2006;](#page-65-0) [Hernández et al., 2015;](#page-66-0) [Pereira et al., 2000\)](#page-68-2). Again, it is important to point out that slave trade involved transportation of people through sea allowing the "jumping" Southern Africa into Europe and the Americas. Lineages in South America are mostly related with a source in Angola.

Taking into account the countries that colonized the Americas and Africa, we can see a correlation between the lineages found North and South America and the colonizing country that carried them. While in North America (Figure 19D), primarily colonized by Great Britain and Spain, we can see over 70% of Western lineages, where the ports in Africa were located, in South America (Figure 19E) there is a distribution of lineages between West and South Africa. South America was colonized mainly by Portugal and Spain and since Spain had almost no colonies in Africa, the slaves either came from the Portuguese colonies, located in the South, or the Western colonies, first of Portuguese rule and later most of which belonged to France as evident in Figure 14.

# **CONCLUSION**

Founder Analysis has the purpose of identifying, scaling and dating the migration of a certain population to a new territory, taking into account the diversity of lineages originally from a specific geographic location, identified as the source, and that later settled in a new location, identified as the sink. In this work, we performed this analysis to verify the time-frames of major migrations involving Sub-Saharan African lineages to Southwestern Asia, Europe, Iberia, North and South America. After confirming that there was a clear distinction between pre-historical migrations, around 8000 years ago, and historical movements, around 500 years ago - in here primarily linked to Trans-Atlantic Slave Trade - we were able to pinpoint the general location of the lineages in the source, giving us a clear picture from where the mtDNA L lineages found nowadays in the sink populations were carried out from and possibly the reason why.

As expected, not considering the statistical errors and a few undetected source lineages, there are no pre-historical migrations to the Americas, opposed to the clear signals observed in Europe, Iberia and Southwestern Asia. After taking into account the historical records of the Trans-Atlantic Slave Trade, it was also expected the results here shown – most L lineages found in North America where carried from the West of Africa, where the main slave ports were located.

It was observed, in agreement with other genetic studies, that there were clear signs of prehistorical migrations to Europe, Iberia and Southwestern Asia, following the geographical proximity. Meaning that we observed lineages that arrived to Southwestern Asia from Eastern Africa, and to Iberia arrived from Western and Northern Africa and to Europe in general from Eastern, Central and Western African. This also supports the previously introduced theory that the Sahel Belt and North Africa were migratory corridors to enter Europe and Iberia following the Glacial Period.

In historical lineages, it was observed a major influence of South Africa both in Iberia and South America. As mentioned before, Portugal colonized Angola and Mozambique - located in the South of Africa - where there were two important ports for slave exportation, and Brazil, in South America. At the time, Portugal was one of the most influential nations of the world, alongside Spain, controlling not only the main potencies of the New World but also the most important Slave Trade markets, Lisbon and Faro. As a consequence, most of the slaves carried from Angola and Mozambique either came to Portugal for trading or went directly to Brazil, leaving this way an important imprint in L lineages in Iberia and South America.

To conclude, the genetic studies associated with ancestry, such as Founder Analysis, help us to better understand what happened long before us. This work in particular gave a new insight to lesser-known facts related to the Trans-Atlantic Slave Trade and the genetic evidences left behind that are still present to this day.

## **REFERENCES**

- Alberts B, Bray D, Lewis J and Raff M (1994) Molecular biology of the cell, Garland Science, New York, NY.
- <span id="page-62-0"></span>Alves-Silva J, da Silva Santos M, Guimarães PE, Ferreira AC, Bandelt HJ, Pena SD and Prado VF (2000) The ancestry of Brazilian mtDNA lineages. The American Journal of Human Genetics **67**:444-461.
- Anderson S, Bankier AT, Barrell BG, de Bruijn MH, Coulson AR, Drouin J, Eperon IC, Nierlich DP, Roe BA, Sanger F, Schreier PH, Smith AJ, Staden R and Young IG (1981) Sequence and organization of the human mitochondrial genome. Nature 290:457-465.
- Andrews RM, Kubacka I, Chinnery PF, Lightowlers RN, Turnbull DM and Howell N (1999) Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. Nature Genetics 23:147-147.
- <span id="page-62-1"></span>Auton A, Abecasis GR, Altshuler DM, Durbin RM, Abecasis GR, Bentley DR, Chakravarti A, Clark AG, Donnelly P, Eichler EE, Flicek P, Gabriel SB, Gibbs RA, Green ED, Hurles ME, Knoppers BM, Korbel JO, Lander ES, Lee C, Lehrach H, Mardis ER, Marth GT, McVean GA, Nickerson DA, Schmidt JP, Sherry ST, Wang J, Wilson RK, Gibbs RA, Boerwinkle E, Doddapaneni H, Han Y, Korchina V, Kovar C, Lee S, Muzny D, Reid JG, Zhu Y, Wang J, Chang Y, Feng Q, Fang X, Guo X, Jian M, Jiang H, Jin X, Lan T, Li G, Li J, Li Y, Liu S, Liu X, Lu Y, Ma X, Tang M, Wang B, Wang G, Wu H, Wu R, Xu X, Yin Y, Zhang D, Zhang W, Zhao J, Zhao M, Zheng X, Lander ES, Altshuler DM, Gabriel SB, Gupta N, Gharani N, Toji LH, Gerry NP, Resch AM, Flicek P, Barker J, Clarke L, Gil L, Hunt SE, Kelman G, Kulesha E, Leinonen R, McLaren WM, Radhakrishnan R, Roa A, Smirnov D, Smith RE, Streeter I, Thormann A, Toneva I, Vaughan B, Zheng-Bradley X, Bentley DR, Grocock R, Humphray S, James T, Kingsbury Z, Lehrach H, Sudbrak R, Albrecht MW, Amstislavskiy VS, Borodina TA, Lienhard M, Mertes F, Sultan M, Timmermann B, Yaspo M-L, Mardis ER, Wilson RK, Fulton L, Fulton R, Sherry ST, Ananiev V, Belaia Z, Beloslyudtsev D, Bouk N, Chen C, Church D, Cohen R, Cook C, Garner J, Hefferon T, Kimelman M, Liu C, Lopez J, Meric P, O'Sullivan C, Ostapchuk Y, Phan L, Ponomarov S, Schneider V, Shekhtman E, Sirotkin K, Slotta D, Zhang H, McVean GA, Durbin RM, Balasubramaniam S, Burton J, Danecek P, Keane TM, Kolb-Kokocinski A, McCarthy S, Stalker J, Quail M, Schmidt JP, Davies CJ, Gollub J, Webster T,

Wong B, Zhan Y, Auton A, Campbell CL, Kong Y, Marcketta A, Gibbs RA, Yu F, Antunes L, Bainbridge M, Muzny D, Sabo A, Huang Z, Wang J, Coin LJM, Fang L, Guo X, Jin X, Li G, Li Q, Li Y, Li Z, Lin H, Liu B, Luo R, Shao H, Xie Y, Ye C, Yu C, Zhang F, Zheng H, Zhu H, Alkan C, Dal E, Kahveci F, Marth GT, Garrison EP, Kural D, Lee W-P, Fung Leong W, Stromberg M, Ward AN, Wu J, Zhang M, Daly MJ, DePristo MA, Handsaker RE, Altshuler DM, Banks E, Bhatia G, del Angel G, Gabriel SB, Genovese G, Gupta N, Li H, Kashin S, Lander ES, McCarroll SA, Nemesh JC, Poplin RE, Yoon SC, Lihm J, Makarov V, Clark AG, Gottipati S, Keinan A, Rodriguez-Flores JL, Korbel JO, Rausch T, Fritz MH, Stütz AM, Flicek P, Beal K, Clarke L, Datta A, Herrero J, McLaren WM, Ritchie GRS, Smith RE, Zerbino D, Zheng-Bradley X, Sabeti PC, Shlyakhter I, Schaffner SF, Vitti J, Cooper DN, Ball EV, Stenson PD, Bentley DR, Barnes B, Bauer M, Keira Cheetham R, Cox A, Eberle M, Humphray S, Kahn S, Murray L, Peden J, Shaw R, Kenny EE, Batzer MA, Konkel MK, Walker JA, MacArthur DG, Lek M, Sudbrak R, Amstislavskiy VS, Herwig R, Mardis ER, Ding L, Koboldt DC, Larson D, Ye K, Gravel S, The Genomes Project C, Corresponding a, Steering c, Production g, Baylor College of M, Shenzhen BGI, Broad Institute of MIT, Harvard, Coriell Institute for Medical R, European Molecular Biology Laboratory EBI, Illumina, Max Planck Institute for Molecular G, McDonnell Genome Institute at Washington U, Health USNIo, University of O, Wellcome Trust Sanger I, Analysis g, Affymetrix, Albert Einstein College of M, Bilkent U, Boston C, Cold Spring Harbor L, Cornell U, European Molecular Biology L, Harvard U, Human Gene Mutation D, Icahn School of Medicine at Mount S, Louisiana State U, Massachusetts General H, McGill U and National Eye Institute NIH (2015) A global reference for human genetic variation. Nature 526:68-74.

- Avise JC (2000) Phylogeography: The History and Formation of Species, Harvard University Press, Cambridge, UK.
- Bandelt HJ, Forster P, Sykes BC and Richards MB (1995) Mitochondrial portraits of human populations using median networks. Genetics 141:743-753.
- Bandelt HJ, Macaulay V and Richards M (2002) What molecules can't tell us about the spread of languages and the Neolithic, in *Examining the Farming/Language Dispersal Hypothesis* pp 99-107, Cambridge.
- Barbieri C, Vicente M, Rocha J, Mpoloka SW, Stoneking M and Pakendorf B (2013) Ancient substructure in early mtDNA lineages of southern Africa. The American Journal of Human Genetics 92:285-292.
- Behar DM, Villems R, Soodyall H, Blue-Smith J, Pereira L, Metspalu E, Scozzari R, Makkan H, Tzur S, Comas D, Bertranpetit J, Quintana-Murci L, Tyler-Smith C, Wells RS and Rosset S (2008) The dawn of human matrilineal diversity. The American Journal of Human Genetics 82:1130-1140.
- <span id="page-64-2"></span>Blome MW, Cohen AS, Tryon CA, Brooks AS and Russell J (2012) The environmental context for the origins of modern human diversity: a synthesis of regional variability in African climate 150,000-30,000 years ago. Journal of human evolution 62:563-592.
- Blumberg A, Barshad G and Mishmar D (2016) Mitochondrial and Nuclear Genome Coevolution, in *Encyclopedia of Evolutionary Biology* (Kliman RM ed) pp 19-26, Academic Press, Oxford.
- <span id="page-64-1"></span>Botigué LR, Henn BM, Gravel S, Maples BK, Gignoux CR, Corona E, Atzmon G, Burns E, Ostrer H, Flores C, Bertranpetit J, Comas D and Bustamante CD (2013) Gene flow from North Africa contributes to differential human genetic diversity in southern Europe. Proceedings of the National Academy of Sciences 110:11791-11796.
- Brandstätter A, Peterson CT, Irwin JA, Mpoke S, Koech DK, Parson W and Parsons TJ (2004) Mitochondrial DNA control region sequences from Nairobi (Kenya): inferring phylogenetic parameters for the establishment of a forensic database. International Journal of Legal Medicine **118**:294-306.
- Cann RL, Stoneking M and Wilson AC (1987) Mitochondrial DNA and human evolution. Nature 325:31-36.
- <span id="page-64-0"></span>Cerezo M, Achilli A, Olivieri A, Perego UA, Gómez-Carballa A, Brisighelli F, Lancioni H, Woodward SR, López-Soto M, Carracedo A, Capelli C, Torroni A and Salas A (2012) Reconstructing ancient mitochondrial DNA links between Africa and Europe. Genome research 22:821-826.
- Chakravarti A (2015) Perspectives on Human Variation through the Lens of Diversity and Race. Cold Spring Harbor perspectives in biology 7:a023358.
- Chan DC (2006) Mitochondrial fusion and fission in mammals. Annual review of cell and developmental biology **22**:79-99.
- Chen Y-S, Torroni A, Excoffier L, Santachiara-Benerecetti AS and Wallace DC (1995) Analysis of mtDNA variation in African populations reveals the most ancient of all human continentspecific haplogroups. The American Journal of Human Genetics 57:133.
- Choudhuri S (2014) Chapter 9 Phylogenetic Analysis, in *Bioinformatics for Beginners* (Choudhuri S ed) pp 209-218, Academic Press, Oxford.
- Crews S, Ojala D, Posakony J, Nishiguchi J and Attardi G (1979) Nucleotide sequence of a region of human mitochondrial DNA containing the precisely identified origin of replication. Nature 277:192-198.
- Cruciani F, Trombetta B, Massaia A, Destro-Bisol G, Sellitto D and Scozzari R (2011) A revised root for the human Y chromosomal phylogenetic tree: the origin of patrilineal diversity in Africa. The American Journal of Human Genetics 88:814-818.
- Cruzan MB and Templeton AR (2000) Paleoecology and coalescence: phylogeographic analysis of hypotheses from the fossil record. Trends in ecology & evolution 15:491-496.
- <span id="page-65-2"></span>Derenko M, Malyarchuk B, Bahmanimehr A, Denisova G, Perkova M, Farjadian S and Yepiskoposyan L (2013) Complete Mitochondrial DNA Diversity in Iranians. PLOS ONE **8:**e80673.
- Detmer SA and Chan DC (2007) Functions and dysfunctions of mitochondrial dynamics. Nature Reviews Molecular Cell Biology 8:870-879.
- <span id="page-65-1"></span>Deyle S (2005) *Carry me back: the domestic slave trade in American life*, Oxford University Press on Demand.
- Dhar A and Minin VN (2016) Maximum Likelihood Phylogenetic Inference, in *Encyclopedia of* Evolutionary Biology (Kliman RM ed) pp 499-506, Academic Press, Oxford.
- Dimauro S and Davidzon G (2005) Mitochondrial DNA and disease. Annals of medicine 37:222-232.
- <span id="page-65-0"></span>Earle TF and Lowe KJP (2006) Black Africans in Renaissance Europe. *The American Historical* Review 111:286-287.
- Ernster L and Schatz G (1981) Mitochondria: a historical review. The Journal of cell biology 91:227s-255s.
- Fleagle JG, Assefa Z, Brown FH and Shea JJ (2008) Paleoanthropology of the Kibish Formation, southern Ethiopia: Introduction. Journal of human evolution 55:360-365.
- Forster P, Harding R, Torroni A and Bandelt HJ (1996) Origin and evolution of Native American mtDNA variation: a reappraisal. The American Journal of Human Genetics 59:935-945.
- Freudenstein JV (2016) Parsimony Methods in Phylogenetics, in *Encyclopedia of Evolutionary* Biology (Kliman RM ed) pp 220-224, Academic Press, Oxford.
- González AM, Larruga JM, Abu-Amero KK, Shi Y, Pestano J and Cabrera VM (2007) Mitochondrial lineage M1 traces an early human backflow to Africa. *BMC genomics* 8:223.
- Gronau I, Hubisz MJ, Gulko B, Danko CG and Siepel A (2011) Bayesian inference of ancient human demography from individual genome sequences. Nature Genetics 43:1031.
- Haak W, Lazaridis I, Patterson N, Rohland N, Mallick S, Llamas B, Brandt G, Nordenfelt S, Harney E, Stewardson K, Fu Q, Mittnik A, Bánffy E, Economou C, Francken M, Friederich S, Pena RG, Hallgren F, Khartanovich V, Khokhlov A, Kunst M, Kuznetsov P, Meller H, Mochalov O, Moiseyev V, Nicklisch N, Pichler SL, Risch R, Rojo Guerra MA, Roth C, Szécsényi-Nagy A, Wahl J, Meyer M, Krause J, Brown D, Anthony D, Cooper A, Alt KW and Reich D (2015) Massive migration from the steppe was a source for Indo-European languages in Europe. Nature 522:207-211.
- <span id="page-66-2"></span>Harris WV (2012) Demography, Geography and the Sources of Roman Slaves. *Journal of Roman* Studies 89:62-75.
- <span id="page-66-1"></span>Hellie R (2020) Slavery, in *Encyclopedia Britannica*, https:/[/www.britannica.com/topic/slavery](http://www.britannica.com/topic/slavery-sociology)[sociology.](http://www.britannica.com/topic/slavery-sociology)
- Henn BM, Gignoux C, Lin AA, Oefner PJ, Shen P, Scozzari R, Cruciani F, Tishkoff SA, Mountain JL and Underhill PA (2008) Y-chromosomal evidence of a pastoralist migration through Tanzania to southern Africa. Proceedings of the National Academy of Sciences of the United States of America 105:10693-10698.
- Henn BM, Gignoux CR, Jobin M, Granka JM, Macpherson JM, Kidd JM, Rodríguez-Botigué L, Ramachandran S, Hon L, Brisbin A, Lin AA, Underhill PA, Comas D, Kidd KK, Norman PJ, Parham P, Bustamante CD, Mountain JL and Feldman MW (2011) Hunter-gatherer genomic diversity suggests a southern African origin for modern humans. *Proceedings of* the National Academy of Sciences of the United States of America 108:5154-5162.
- <span id="page-66-0"></span>Hernández CL, Soares P, Dugoujon JM, Novelletto A, Rodríguez JN, Rito T, Oliveira M, Melhaoui M, Baali A, Pereira L and Calderón R (2015) Early Holocenic and Historic mtDNA African Signatures in the Iberian Peninsula: The Andalusian Region as a Paradigm. PLOS ONE 10:e0139784.
- Howell N, Elson JL, Howell C and Turnbull DM (2007) Relative Rates of Evolution in the Coding and Control Regions of African mtDNAs. *Molecular Biology and Evolution* **24**:2213-2221.
- Hublin J-J, Ben-Ncer A, Bailey SE, Freidline SE, Neubauer S, Skinner MM, Bergmann I, Le Cabec A, Benazzi S, Harvati K and Gunz P (2017) New fossils from Jebel Irhoud, Morocco and the pan-African origin of Homo sapiens. Nature 546:289-292.
- Jobling M, Hollox E, Hurles M, Kivisild T and Tyler-Smith C (2014) Human Evolutionary Genetics, Garland Science, USA.
- Karbowski M and Youle RJ (2003) Dynamics of mitochondrial morphology in healthy cells and during apoptosis. Cell Death & Differentiation 10:870-880.
- <span id="page-67-0"></span>Kehinde M (2014) Trans-Saharan Slave Trade, in *Encyclopedia of Migration* (Bean FD and Brown SK eds) pp 1-4, Springer Netherlands, Dordrecht.
- Kivisild T, Shen P, Wall DP, Do B, Sung R, Davis K, Passarino G, Underhill PA, Scharfe C, Torroni A, Scozzari R, Modiano D, Coppa A, de Knijff P, Feldman M, Cavalli-Sforza LL and Oefner PJ (2006) The role of selection in the evolution of human mitochondrial genomes. *Genetics* 172:373-387.
- <span id="page-67-2"></span>LaVeist TA (1993) Segregation, Poverty, and Empowerment: Health Consequences for African Americans. The Milbank Quarterly 71:41-64.
- Li R, Montpetit A, Rousseau M, Wu SY, Greenwood CM, Spector TD, Pollak M, Polychronakos C and Richards JB (2014) Somatic point mutations occurring early in development: a monozygotic twin study. Journal of medical genetics 51:28-34.
- Luo S, Valencia CA, Zhang J, Lee N-C, Slone J, Gui B, Wang X, Li Z, Dell S, Brown J, Chen SM, Chien Y-H, Hwu W-L, Fan P-C, Wong L-J, Atwal PS and Huang T (2018) Biparental Inheritance of Mitochondrial DNA in Humans. Proceedings of the National Academy of Sciences **115**:13039-13044.
- Macaulay V, Richards M, Hickey E, Vega E, Cruciani F, Guida V, Scozzari R, Bonné-Tamir B, Sykes B and Torroni A (1999) The emerging tree of West Eurasian mtDNAs: a synthesis of controlregion sequences and RFLPs. The American Journal of Human Genetics 64:232-249.
- <span id="page-67-1"></span>Marable M (2015) How capitalism underdeveloped Black America: Problems in race, political economy, and society, Haymarket Books, Chicago.
- Marske KA (2016) Phylogeography, in *Encyclopedia of Evolutionary Biology* (Kliman RM ed) pp 291-296, Academic Press, Oxford.
- <span id="page-67-3"></span>Matisoo-Smith E, Gosling AL, Platt D, Kardailsky O, Prost S, Cameron-Christie S, Collins CJ, Boocock J, Kurumilian Y, Guirguis M, Pla Orquín R, Khalil W, Genz H, Abou Diwan G, Nassar J and Zalloua P (2018) Ancient mitogenomes of Phoenicians from Sardinia and Lebanon: A story of settlement, integration, and female mobility. PLOS ONE 13:e0190169.
- McBride HM, Neuspiel M and Wasiak S (2006) Mitochondria: more than just a powerhouse. Current biology 16:R551-560.
- McDougall I, Brown FH and Fleagle JG (2005) Stratigraphic placement and age of modern humans from Kibish, Ethiopia. Nature 433:733-736.
- <span id="page-68-1"></span>Meade TA (2016) History of modern Latin America: 1800 to the present, John Wiley & Sons, New York.
- Mendez FL, Krahn T, Schrack B, Krahn AM, Veeramah KR, Woerner AE, Fomine FL, Bradman N, Thomas MG, Karafet TM and Hammer MF (2013) An African American paternal lineage adds an extremely ancient root to the human Y chromosome phylogenetic tree. The American Journal of Human Genetics **92**:454-459.
- <span id="page-68-0"></span>Micheletti SJ, Bryc K, Ancona Esselmann SG, Freyman WA, Moreno ME, Poznik GD, Shastri AJ, Beleza S and Mountain JL (2020) Genetic Consequences of the Transatlantic Slave Trade in the Americas. The American Journal of Human Genetics 107:265-277.
- Mishmar D, Ruiz-Pesini E, Golik P, Macaulay V, Clark AG, Hosseini S, Brandon M, Easley K, Chen E, Brown MD, Sukernik RI, Olckers A and Wallace DC (2003) Natural selection shaped regional mtDNA variation in humans. Proceedings of the National Academy of Sciences 100:171-176.
- Nakhleh L and Morrison DA (2016) Phylogenetic Networks, in *Encyclopedia of Evolutionary Biology* (Kliman RM ed) pp 264-269, Academic Press, Oxford.
- Olivieri A, Achilli A, Pala M, Battaglia V, Fornarino S, Al-Zahery N, Scozzari R, Cruciani F, Behar DM and Dugoujon J-M (2006) The mtDNA legacy of the Levantine early Upper Palaeolithic in Africa. Science 314:1767-1770.
- Pääbo S (1989) Ancient DNA: extraction, characterization, molecular cloning, and enzymatic amplification. Proceedings of the National Academy of Sciences 86:1939.
- <span id="page-68-2"></span>Pereira L, Prata MJ and Amorim A (2000) Diversity of mtDNA lineages in Portugal: not a genetic edge of European variation. Annals of human genetics 64:491-506.
- Relethford JH (2001) Genetics and the search for modern human origins, Wiley-Liss/John Wiley & Sons, New York.
- Relethford JH (2008) Genetic evidence and the modern human origins debate. *Heredity* 100:555-563.
- Richards M, Macaulay V, Hickey E, Vega E, Sykes B, Guida V, Rengo C, Sellitto D, Cruciani F, Kivisild T, Villems R, Thomas M, Rychkov S, Rychkov O, Rychkov Y, Gölge M, Dimitrov D,

Hill E, Bradley D, Romano V, Calì F, Vona G, Demaine A, Papiha S, Triantaphyllidis C, Stefanescu G, Hatina J, Belledi M, Di Rienzo A, Novelletto A, Oppenheim A, Nørby S, Al-Zaheri N, Santachiara-Benerecetti S, Scozari R, Torroni A and Bandelt HJ (2000) Tracing European founder lineages in the Near Eastern mtDNA pool. The American Journal of Human Genetics 67:1251-1276.

- Richards Martin B, Soares P and Torroni A (2016) Palaeogenomics: Mitogenomes and Migrations in Europe's Past. Current Biology 26:R243-R246.
- Rito T, Richards MB, Fernandes V, Alshamali F, Cerny V, Pereira L and Soares P (2013) The First Modern Human Dispersals across Africa. PLOS ONE 8:e80031.
- <span id="page-69-0"></span>Rito T, Vieira D, Silva M, Conde-Sousa E, Pereira L, Mellars P, Richards MB and Soares P (2019) A dispersal of Homo sapiens from southern to eastern Africa immediately preceded the out-of-Africa migration. Scientific reports 9:4728.
- <span id="page-69-3"></span>Rosa A and Brehem A (2011) African human mtDNA phylogeography at-a-glance. Journal of Anthropological Sciences 89:25-58.
- Salas A, Richards M, De la Fe T, Lareu MV, Sobrino B, Sánchez-Diz P, Macaulay V and Carracedo A (2002) The making of the African mtDNA landscape. The American Journal of Human Genetics **71**:1082-1111.
- <span id="page-69-2"></span>Salas A, Richards M, Lareu MV, Scozzari R, Coppa A, Torroni A, Macaulay V and Carracedo A (2004) The African diaspora: mitochondrial DNA and the Atlantic slave trade. The American Journal of Human Genetics 74:454-465.
- Schwartz M and Vissing J (2002) Paternal inheritance of mitochondrial DNA. The New England journal of medicine **347**:576-580.
- Scott AD and Baum DA (2016) Phylogenetic Tree, in *Encyclopedia of Evolutionary Biology* (Kliman RM ed) pp 270-276, Academic Press, Oxford.
- Sharma P and Sampath H (2019) Mitochondrial DNA Integrity: Role in Health and Disease. Cells 8.
- Shokolenko I, Venediktova N, Bochkareva A, Wilson GL and Alexeyev MF (2009) Oxidative stress induces degradation of mitochondrial DNA. Nucleic acids research 37:2539-2548.
- Siekevitz P (1957) Powerhouse of the Cell. Scientific American 197:131.
- <span id="page-69-1"></span>Silva M, Alshamali F, Silva P, Carrilho C, Mandlate F, Jesus Trovoada M, Černý V, Pereira L and Soares P (2015) 60,000 years of interactions between Central and Eastern Africa documented by major African mitochondrial haplogroup L2. Scientific reports 5:12526.
- <span id="page-70-2"></span>Skorecki K and Behar DM (2013) North Africans traveling north. Proceedings of the National Academy of Sciences of the United States of America 110:11668-11669.
- <span id="page-70-3"></span>Soares P, Achilli A, Semino O, Davies W, Macaulay V, Bandelt H-J, Torroni A and Richards MB (2010) The Archaeogenetics of Europe. Current Biology 20:R174-R183.
- Soares P, Alshamali F, Pereira JB, Fernandes V, Silva NM, Afonso C, Costa MD, Musilová E, Macaulay V and Richards MB (2012) The expansion of mtDNA haplogroup L3 within and out of Africa. Molecular biology and evolution 29:915-927.
- <span id="page-70-6"></span>Soares P, Ermini L, Thomson N, Mormina M, Rito T, Röhl A, Salas A, Oppenheimer S, Macaulay V and Richards MB (2009) Correcting for purifying selection: an improved human mitochondrial molecular clock. Am J Hum Genet 84:740-759.
- <span id="page-70-0"></span>Soares P, Rito T, Pereira L and Richards MB (2016) A Genetic Perspective on African Prehistory, in Africa from MIS 6-2 (Jones SC and Stewart BA eds) pp 383-405, Springer, London, UK.
- Sokal RR (2001) Archaeogenetics: DNA and the Population Prehistory of Europe. The American Journal of Human Genetics 69:243-244.
- Stefano GB, Bjenning C, Wang F, Wang N and Kream RM (2017) Mitochondrial Heteroplasmy. Advances in experimental medicine and biology **982**:577-594.
- Stewart B and Jones S (2016) Africa from MIS 6-2: Population Dynamics and Paleoenvironments, Springer, London, UK.
- Sutovsky P, Moreno RD, Ramalho-Santos J, Dominko T, Simerly C and Schatten G (1999) Ubiquitin tag for sperm mitochondria. Nature 402:371-372.
- <span id="page-70-4"></span>Thornton J (1998) Africa and Africans in the Making of the Atlantic World, 1400–<sup>1800</sup>, Cambridge University Press, Cambridge.
- Tishkoff SA, Reed FA, Friedlaender FR, Ehret C, Ranciaro A, Froment A, Hirbo JB, Awomoyi AA, Bodo J-M and Doumbo O (2009) The genetic structure and history of Africans and African Americans. Science 324:1035-1044.
- Torroni A, Schurr TG, Cabell MF, Brown MD, Neel JV, Larsen M, Smith DG, Vullo CM and Wallace DC (1993) Asian affinities and continental radiation of the four founding Native American mtDNAs. The American Journal of Human Genetics 53:563-590.
- <span id="page-70-1"></span>Triska P, Soares P, Patin E, Fernandes V, Cerny V and Pereira L (2015) Extensive Admixture and Selective Pressure Across the Sahel Belt. Genome biology and evolution 7:3484-3495.
- <span id="page-70-5"></span>van Oven M and Kayser M (2009) Updated comprehensive phylogenetic tree of global human mitochondrial DNA variation. Human mutation 30:E386-394.
- Veeramah KR, Wegmann D, Woerner A, Mendez FL, Watkins JC, Destro-Bisol G, Soodyall H, Louie L and Hammer MF (2012) An early divergence of KhoeSan ancestors from those of other modern humans is supported by an ABC-based analysis of autosomal resequencing data. Molecular biology and evolution **29**:617-630.
- Vibranovski MD and Long M (2016) Gene Origin, Sex Chromosomes and, in *Encyclopedia of* Evolutionary Biology (Kliman RM ed) pp 117-126, Academic Press, Oxford.
- <span id="page-71-0"></span>Viehberg FA, Just J, Dean JR, Wagner B, Franz SO, Klasen N, Kleinen T, Ludwig P, Asrat A, Lamb HF, Leng MJ, Rethemeyer J, Milodowski AE, Claussen M and Schäbitz F (2018) Environmental change during MIS4 and MIS 3 opened corridors in the Horn of Africa for Homo sapiens expansion. *Quaternary Science Reviews* **202**:139-153.
- <span id="page-71-3"></span>Vieira D, Almeida M, Richards MB and Soares P (2020) An Efficient and User-Friendly Implementation of the Founder Analysis Methodology., in *Practical Applications of* Computational Biology and Bioinformatics, 13th International Conference, Springer.
- <span id="page-71-2"></span>Weissensteiner H, Pacher D, Kloss-Brandstätter A, Forer L, Specht G, Bandelt HJ, Kronenberg F, Salas A and Schönherr S (2016) HaploGrep 2: mitochondrial haplogroup classification in the era of high-throughput sequencing. Nucleic acids research 44:W58-63.
- <span id="page-71-1"></span>Weldeab S, Lea DW, Schneider RR and Andersen N (2007) 155,000 years of West African monsoon and ocean thermal evolution. Science 316:1303-1307.
- White TD, Asfaw B, DeGusta D, Gilbert H, Richards GD, Suwa G and Howell FC (2003) Pleistocene Homo sapiens from Middle Awash, Ethiopia. Nature 423:742-747.
- Wolpoff M, Wu X and Thorne A (1984) Modern Homo sapiens origins: a general theory of hominid evolution involving the fossil evidence from East Asia, [in:] Origins of modern humans: a world survey of the fossil evidence, pp 411-483, Alan Liss New York.
- Yang Z (2016) Bayesian Phylogenetic Methods, in *Encyclopedia of Evolutionary Biology* (Kliman RM ed) pp 137-140, Academic Press, Oxford.
- Ye K, Lu J, Ma F, Keinan A and Gu Z (2014) Extensive pathogenicity of mitochondrial heteroplasmy in healthy human individuals. Proceedings of the National Academy of Sciences of the United States of America 111:10654-10659.
- Zhang H, Burr SP and Chinnery PF (2018) The mitochondrial DNA genetic bottleneck: inheritance and beyond. *Essays in biochemistry* **62**:225-234.
## SUPPLEMENTARY FILE 1

Table S1 – Complete dataset analysed for mtDNA studies including the respective haplogroup and country

## Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country.



<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
DQ304925	L2a1a1	<b>USA</b>	DQ304963	L2a1f1	<b>USA</b>
DQ304926	L2a1a	<b>USA</b>	DQ304964	L2a1f1	<b>USA</b>
DQ304927	L2ala2c	<b>USA</b>	DQ304965	L2a1f	<b>USA</b>
DQ304928	L2a1a	<b>USA</b>	DQ304966	L2a1f	<b>USA</b>
DQ304929	L2a1e1	<b>USA</b>	DQ304967	L2a1f1	<b>USA</b>
DQ304930	L2a1e1	<b>USA</b>	DQ304968	L2a1a2	<b>USA</b>
DQ304931	L2a1e1	<b>USA</b>	DQ304969	L2ala2ala	<b>USA</b>
DQ304932	L2a1a	<b>USA</b>	DQ304970	L2ala2ala	<b>USA</b>
DQ304933	L2a1a1	<b>USA</b>	DQ304971	L2a1a2	<b>USA</b>
DQ304934	L2a1f1a	<b>USA</b>	DQ304972	L2a1a2b	<b>USA</b>
DQ304935	L2a1f	<b>USA</b>	DQ304973	L2a1a2b	<b>USA</b>
DQ304936	L2a1f	<b>USA</b>	DQ304974	L2ala2ala	<b>USA</b>
DQ304937	L2a1f1	<b>USA</b>	DQ304975	L2a1a2a	<b>USA</b>
DO304938	L2a1m1a	<b>USA</b>	DQ304976	L2ala2ala	<b>USA</b>
DQ304939	L2a111a	<b>USA</b>	DQ304977	L2a1a2	<b>USA</b>
DQ304940	L2a1m1	<b>USA</b>	DQ304978	L2b1a3	<b>USA</b>
DQ304941	L2a1n	<b>USA</b>	DQ304979	L2b1a3	<b>USA</b>
DQ304942	L2a1c4a	<b>USA</b>	DQ304980	L2b1a3	<b>USA</b>
DQ304943	L2a1c4a	<b>USA</b>	DQ304981	L2b1a	<b>USA</b>
DQ304944	L2a1c	<b>USA</b>	DQ304982	L2b1a3	<b>USA</b>
DQ304945	L2a1e	<b>USA</b>	DQ304983	L2b1a3	<b>USA</b>
DQ304946	L2a1e1	<b>USA</b>	DQ304984	L2b1a3	<b>USA</b>
DQ304947	L2a1e1	<b>USA</b>	DQ304985	L2b1a2	<b>USA</b>
DQ304948	L2a1a3b	<b>USA</b>	DQ304986	L2c2	<b>USA</b>
DQ304949	L2a1c	<b>USA</b>	DQ304987	L2c2a1	<b>USA</b>
DQ304950	L2a1c4a1	<b>USA</b>	DQ304988	L2c2a	<b>USA</b>
DQ304951	L2a1c4a1	<b>USA</b>	DQ304989	L2c2	<b>USA</b>
DQ304952	L2a1f	<b>USA</b>	DO304990	L3b1a3	<b>USA</b>
DQ304953	L2a1f	<b>USA</b>	DQ304991	L3b1a11	<b>USA</b>
DQ304954	L2a1f	<b>USA</b>	DQ304992	L3bla11	<b>USA</b>
DQ304955	L2a1f1	<b>USA</b>	DQ304993	L3b1a5	<b>USA</b>
DQ304956	L2a1f	<b>USA</b>	DQ304994	L3b1a1a	<b>USA</b>
DQ304957	L2a1f2	<b>USA</b>	DQ304995	L3b1a1a	<b>USA</b>
DQ304958	L2a1f1a	<b>USA</b>	DQ304996	L3b1a	<b>USA</b>
DQ304959	L2a1f	<b>USA</b>	DQ304997	L3b1a	<b>USA</b>
DQ304960	L2a1f1	<b>USA</b>	DQ304998	L3e2b+152	<b>USA</b>
DQ304961	L2a1f1	<b>USA</b>	DQ304999	L3e2b+152	<b>USA</b>
DQ304962	L2a1f	<b>USA</b>	DQ305000	L3e2b4	<b>USA</b>

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)





<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
EF184599	L <sub>Of2a</sub>	Tanzania	EF556171	L3x1a	Israel
EF184600	L0f	Tanzania	EF556173	L5a1a	Israel
EF184601	L0a1c1	Tanzania	EF556174	L0a2c	Israel
EF184602	L0a2	Tanzania	ESP0167	L3e4a	Spain
EF184603	L0a2a1a	Tanzania	ESP0191	L3d1b3a	Spain
EF184604	L0a2	Tanzania	<b>ESP0225</b>	L2a1c1	Spain
EF184605	L0a2a2a	Tanzania	<b>ESP0297</b>	L3e5a	Spain
EF184606	L0a2	Tanzania	ESP0313	L2a1c3a	Spain
EF184607	L0a2	Tanzania	ESP0413	L4b2b	Spain
EF184608	L0a2	Tanzania	ESP0702	L1b1a+189	Spain
EF184609	L0kla1	South Africa	ESP0714	L3b1a+@16124	Spain
EF184610	LOklalb	South Africa	ESP0813	L1b1a6	Spain
EF184611	LOklala	South Africa	<b>ESP0922</b>	L3b1b1	Spain
EF184612	L1c3bla	Tanzania	<b>ESP0926</b>	L2c5	Spain
EF184613	L1cla2b	Cameroon	<b>ESP0928</b>	L2a1c+16129	Spain
EF184614	L1cla2b	Cameroon	<b>ESP0954</b>	L1b1a14	Spain
EF184615	L1cla2b	Cameroon	<b>ESP0969</b>	L1b1a6	Spain
EF184616	Llclalala	Cameroon	<b>ESP0975</b>	L3e2b	Spain
EF184617	L2a1c5	Tanzania	ESP1024	L3e1f	Spain
EF184618	$L2a1+143$	Tanzania	EU092658	L2a1+143+16189	Israel
EF184619	$L2a1+143$	Tanzania	EU092659	L2a1+143+16189	Israel
EF184620	$L2a1+143$	Tanzania	EU092660	L3c	Israel
EF184621	L2dla	Tanzania	EU092661	L2b3c	Israel
EF184622	L3h1a2	Tanzania	EU092662	L4b2a2a	Israel
EF184623	L3d1a1a	Tanzania	EU092663	L2a1c3a	Israel
EF184624	L3h1a2	Tanzania	EU092664	L2b1a2	Israel
EF184625	L3a1a	Tanzania	EU092665	L0a1a	Israel
EF184627	L4b2a2	Tanzania	EU092666	$L3x1+16311$	Israel
EF184628	L3dlalal	Tanzania	EU092667	L1b1a16	Israel
EF184629	L4b2a1	Tanzania	EU092668	L0f2a1	Israel
EF184630	L3a1a	Tanzania	EU092669	L3b1a2	Israel
EF184631	L3h1a2a1	Tanzania	EU092670	L0a1d	Israel
EF184632	L3h1a2a1	Tanzania	EU092671	L2a1p	Israel
EF184633	L4b2a2c	South Africa	EU092672	L1b1a2	Israel
EF184639	L4b2a2	Tanzania	EU092673	L6b	Israel
EF184640	L4b2a2b	Tanzania	EU092674	L2a1+143+16189	Israel
EF184641	L3dlala	Tanzania	EU092675	L3e1b1	Israel
EF556166	L3h1a2a1	Israel	EU092676	L2a1h	Israel

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
EU092677	L3e3a	Israel	EU092715	L1bla4a	Guinea-Bissau
EU092678	L4ala	Israel	EU092716	L1b1a6	Guinea-Bissau
EU092679	L2a1+143+16189	Israel	EU092717	L1c1c	Guinea-Bissau
EU092680	L3f1b+16292	Israel	EU092718	L1c3a1a	Guinea-Bissau
EU092681	L3e1b1	<b>Israel</b>	EU092719	L <sub>2a1</sub> i	Guinea-Bissau
EU092682	L3b1b1	Israel	EU092720	L2a1c3b1	Guinea-Bissau
EU092683	L2a1c+16129	<b>Israel</b>	EU092721	L2a112	Guinea-Bissau
EU092684	L3x2a1a	<b>Israel</b>	EU092722	L2b1a2	Guinea-Bissau
EU092685	L3e1	<b>Israel</b>	EU092723	L2c	Guinea-Bissau
EU092686	L6b	Israel	EU092724	L2e	Guinea-Bissau
EU092687	L2a1l2a	<b>Israel</b>	EU092725	L3b2a	Guinea-Bissau
EU092688	L0a1b1a1	Mozambique	EU092726	L3b1a	Guinea-Bissau
EU092689	L1c3b1b	Mozambique	EU092727	L3b1a9a	Guinea-Bissau
EU092690	L2a1b1a	Mozambique	EU092728	L3d1b1	Guinea-Bissau
EU092691	L2a1a2	Mozambique	EU092729	L3e2a3	Guinea-Bissau
EU092692	L2b2	Mozambique	EU092730	L3e2b	Guinea-Bissau
EU092693	L3e1d1a	Mozambique	EU092731	L3e4a1	Guinea-Bissau
EU092694	L3b1a11	Mozambique	EU092732	L3f1b+16292	Guinea-Bissau
EU092695	L3e4a	Mozambique	EU092733	L2a1c	Guinea-Bissau
EU092696	L3f1b1a	Mozambique	EU092734	L2b3b	Guinea-Bissau
EU092697	L2c2	Mozambique	EU092735	L3d2b	Guinea-Bissau
EU092698	L3e1b2	Mozambique	EU092736	L3h1b2	Guinea-Bissau
EU092699	L5a2	Mozambique	EU092737	L1b1a9	Syria
EU092700	L0d2a1	Mozambique	EU092738	L1c2b2	Syria
EU092701	L0a2a2a	Mozambique	EU092739	L2a1o	Syria
EU092702	L3d1a1a	Mozambique	EU092740	L3e1c	Syria
EU092703	L1c3a	Mozambique	EU092741	L3f1b+16292	Syria
EU092704	L3f1b4c	Mozambique	EU092742	L3d4a	Syria
EU092705	L2a1b1a	Mozambique	EU092743	L4b2a2a	Syria
EU092706	L3e3a	Mozambique	EU092744	L3b1a2	Syria
EU092707	L1c2a1a	Mozambique	EU092745	L0a2a2a	Saudi Arabia
EU092708	L0d2c1	Mozambique	EU092746	L0a1b2a	Saudi Arabia
EU092709	L3ela3a	Mozambique	EU092747	L2b1	Saudi Arabia
EU092710	L2c2b1b	Netherlands	EU092748	L4ala	Saudi Arabia
EU092711	L2a1a3	Portugal	EU092749	L3elala	Saudi Arabia
EU092712	L1c2b1c	Portugal	EU092750	L4b2a2	Saudi Arabia
EU092713	L1bla3a	Portugal	EU092751	L3f1b+16292	Lebanon
EU092714	L0alal	Guinea-Bissau	EU092752	L3e4	Lebanon

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)









<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
EU092905	L2ala3a	Chad	EU092943	L5a1b	Ethiopia
EU092906	L0a4	Chad	EU092944	L <sub>3x2a</sub>	Ethiopia
EU092907	L3e3a	Chad	EU092945	L0a1c1	Ethiopia
EU092908	L3dlala	Kenya	EU092946	$L2a1+143$	Ethiopia
EU092909	L0a1bla1	Kenya	EU092947	L3h1b1a	Ethiopia
EU092910	L2a1b1a	Kenya	EU092948	L1b1a2a	Ethiopia
EU092911	L0a2a1a2	Kenya	EU092949	L4a2	Ethiopia
EU092912	L3f1b4a1	Kenya	EU092950	L0a1d	Ethiopia
EU092913	L0a2d	Kenya	EU092951	L4b2a2b	Ethiopia
EU092914	L2a1h	Kenya	EU092952	L1b1a2a	Ethiopia
EU092915	L3elel	Kenya	EU092953	L3f1b2a	Ethiopia
EU092916	L2ala	Kenya	EU092954	L2a1c1a2	Ethiopia
EU092917	L3e2b	Kenya	EU092955	L2c2a	Unknown
EU092918	$L3b1a+152$	Kenya	EU092956	L1c3a1b	Unknown
EU092919	L2a1bla	Jordan	EU092957	L <sub>2</sub> c <sub>2a</sub>	Unknown
EU092920	L3x2a1a	Jordan	EU092958	L3b1a6	Unknown
EU092921	L0d3a	Kuwait	EU092959	L3e5b	Unknown
EU092922	L2a1+143+@16309	Kuwait	EU092960	L3e1a3b	Gambia
EU092923	L3i1b	Kuwait	EU092961	L2a1f1	Unknown
EU092924	L6a	Saudi Arabia	EU092962	L3b1a5	Unknown
EU092925	L0a2a2a	Yemen	EU092963	L0a1b2	Unknown
EU092926	L3e2a2	Yemen	EU092964	L0f <sub>2</sub> a	Unknown
EU092927	L2a1d1	Oman	EU092965	L0d1b2b1a	Unknown
EU092928	L1b1a5	Tunisia	EU092966	L0klala	Unknown
EU092929	L1b1a5	Egypt	EU200759	L3d1b1b	Poland
EU092930	L3blala	Cyprus	EU200760	L2a1k	Czech Rep
EU092931	L3blala	Cyprus	EU200761	L3b1b1	Russia
EU092932	L3dlalal	Pakistan	EU200762	L2a1c3a	Slovenia
EU092933	L2a1a2	Pakistan	EU200763	L2a1k	Slovenia
EU092934	L5b1a	Pakistan	EU200764	L1b1a12b	Russia
EU092935	L4a2	Pakistan	EU273476	L1c1a2b	Cameroon
EU092936	L <sub>0</sub> b	Ethiopia	EU273477	Llclalala	Cameroon
EU092937	L2a1m1	Ethiopia	EU273478	L1clalalb1	Cameroon
EU092938	L4b2a2a	Ethiopia	EU273479	L1cla2a1	Gabon
EU092939	L2a1d1	Ethiopia	EU273480	L1clalalb	Gabon
EU092940	L1bla2a	Ethiopia	EU273481	L1clalalb	Gabon
EU092941	L3a2a	Ethiopia	EU273482	L1clalalb1	Gabon
EU092942	L4b2a1	Ethiopia	EU273483	Llclalala	Gabon

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

















<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
HG03133	L1c2b1c	Nigeria	HG03291	L2b1a	Nigeria
HG03135	L2a1f1	Nigeria	HG03294	L3b1b	Nigeria
HG03136	L1b1a3	Nigeria	HG03295	L1b1a	Nigeria
HG03139	L4b2b	Nigeria	HG03297	L2a1f1	Nigeria
HG03157	L2c2a1	Nigeria	HG03298	L0a1a2	Nigeria
HG03159	L2a1f	Nigeria	HG03300	L1c2b1a'b	Nigeria
HG03160	L3e2b8	Nigeria	HG03301	L3e2a1b1	Nigeria
HG03162	L3f1b+16292+150	Nigeria	HG03303	L3f1b4c	Nigeria
HG03163	L2a1f1	Nigeria	HG03304	L3e2b8	Nigeria
HG03166	L1b1a3b	Nigeria	HG03311	L1b1a	Nigeria
HG03168	L1c2a1	Nigeria	HG03313	L1c2a2	Nigeria
HG03169	L2a1a3c	Nigeria	HG03343	L1c3a	Nigeria
HG03172	L3f1b1a	Nigeria	HG03351	L1b1a	Nigeria
HG03175	L3e2a1b1	Nigeria	HG03352	L3e2bla1	Nigeria
HG03189	L4b2b	Nigeria	HG03354	L2c2bla	Nigeria
HG03190	L3ela	Nigeria	HG03363	L1c2b1b	Nigeria
HG03193	L3f1b1a	Nigeria	HG03366	L3d1b2	Nigeria
HG03195	L3e2b1	Nigeria	HG03367	L2e1a	Nigeria
HG03196	L3e1	Nigeria	HG03369	L3e3b	Nigeria
HG03198	L3d5a	Nigeria	HG03370	L3b1a	Nigeria
HG03199	L2a1f	Nigeria	HG03372	L3e3b	Nigeria
HG03202	L2a1b1	Nigeria	HG03376	L2c	Sierra Leone
HG03209	L <sub>2e</sub>	Sierra Leone	HG03378	L3e3b	Sierra Leone
HG03212	L3e4a1	Sierra Leone	HG03380	L1c1c	Sierra Leone
HG03224	L3d2	Sierra Leone	HG03382	L2a111b	Sierra Leone
HG03225	L2c	Sierra Leone	HG03385	L1c1c	Sierra Leone
HG03240	L2e	Gambia	HG03388	L2c2	Sierra Leone
HG03241	L3d1a1b	Gambia	HG03391	L1c1c	Sierra Leone
HG03246	L3b1a+@16124	Gambia	HG03394	L3b1a6	Sierra Leone
HG03247	L3d2a	Gambia	HG03397	L1c1d	Sierra Leone
HG03259	L4bla	Gambia	HG03401	L2a1	Sierra Leone
HG03265	L3d5a	Nigeria	HG03410	L2a1a1	Sierra Leone
HG03267	L0a1a2	Nigeria	HG03419	L3e2b+152	Sierra Leone
HG03268	L0a1a2	Nigeria	HG03428	L2a111b	Sierra Leone
HG03270	L1b1a18	Nigeria	HG03432	L2a1i	Sierra Leone
HG03271	L1c2b1b	Nigeria	HG03433	L2c	Sierra Leone
HG03279	L2d1	Nigeria	HG03436	L1bla14	Sierra Leone
HG03280	L3e2b8	Nigeria	HG03437	L2d1	Sierra Leone

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)







## Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)





<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
JN655773	L3x1a2	Ethiopia	JN655811	L3e1d1	Somalia
JN655774	L3a1b	Ethiopia	JN655812	L3e3a	Somalia
JN655775	L3h1b1a	Ethiopia	JN655813	L3a1a	Somalia
JN655776	L3x2a	Ethiopia	JN655814	L3i2	Somalia
JN655777	L3f1b+16292	Ethiopia	JN655815	L3h2	Somalia
JN655778	L3i2	Ethiopia	JN655816	L3i2	Somalia
JN655779	L3x1a2	Ethiopia	JN655817	L3f1a1	Somalia
JN655780	L3i1a	Ethiopia	JN655818	L3x1b	Somalia
JN655781	L3x1b	Ethiopia	JN655819	L3f2a1a	Somalia
JN655782	L3x1b	Ethiopia	JN655820	L3h2	Yemen
JN655783	L3f1b+16292	Ethiopia	JN655821	L3h2	Yemen
JN655784	L3f	Ethiopia	JN655822	L <sub>3</sub> e <sub>5</sub>	Sudan
JN655785	L3i2	Ethiopia	JN655823	L3d1b1b	Sudan
JN655786	$L3x1+16311$	Ethiopia	JN655824	L3h1a2a1	Sudan
JN655787	L3i2	Ethiopia	JN655825	L3h2	Sudan
JN655788	L3h1a2b	Ethiopia	JN655826	L3b1a+@16124	Sudan
JN655789	L3k1	Chad	JN655827	L3b1a2	Sudan
JN655790	$L3b1a+152$	Chad	JN655828	$L3e5+195$	Sudan
JN655791	L3b1a9	Chad	JN655829	L3x2a1a	Sudan
JN655792	L3b1a+@16124	Chad	JN655830	L3h1a1	Sudan
JN655793	L3d2b	Chad	JN655831	L3f3	Sudan
JN655794	L3e1b1	Chad	JN655832	L3f2a1	Sudan
JN655795	L3e2b6	Chad	JN655833	L3f1b+16292	Sudan
JN655796	L3e3b	Chad	JN655834	L3d4a	Sudan
JN655797	L3d3b	Chad	JN655835	L3f1b+16292	Sudan
JN655798	L3e5a1a	Chad	JN655836	L3f1b+16292	Sudan
JN655799	L3h1b1a	Chad	JN655837	$L3x1+16311$	Sudan
JN655800	L3dlala	Somalia	JN655838	L3h1b1a	Sudan
JN655801	L3h2	Somalia	JN655839	L3h1b1a	Sudan
JN655802	L3x2a	Somalia	JN655840	L3h1a2a	Sudan
JN655803	$L3a+709$	Somalia	JN655841	L3f2a1	Sudan
JN655804	L3i2	Somalia	JN655842	L3f1a1	Sudan
JN655805	L3a2a	Somalia	JN858955	L2a1c+16129	Cameroon
JN655806	L3b1a+@16124	Somalia	JN858956	L2a1d2	Benin
JN655807	L3elala	Somalia	JN989561	L2a1f1	unknown
JN655808	L3dla1	Somalia	JQ044792	L1b1a4a	<b>Burkina Faso</b>
JN655809	L3f1a1	Somalia	JQ044793	L1b1a10	Burkina Faso
JN655810	L3x1b	Somalia	JQ044794	L3e2bla1	Burkina Faso

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

























<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
JX303768	L1cla2	Zambia	JX303806	L2a1d2	Zambia
JX303769	L1b1a10b	Zambia	JX303807	L2b2a	Zambia
JX303770	L1b2a	Zambia	JX303808	L3e1d1	Zambia
JX303771	L1b1a3	Zambia	JX303809	L1b2a	Zambia
JX303772	L0a2a2a2	Zambia	JX303810	L3d3a1	Zambia
JX303773	L1c2b1b1	Zambia	JX303811	L1c2b1b1	Zambia
JX303774	L3f1bla1	Zambia	JX303812	L1b1a10b	Zambia
JX303775	L1c2ala	Zambia	JX303813	L1b1a10b	Zambia
JX303776	L3e1	Zambia	JX303814	L1b1a10b	Zambia
JX303777	L1b1a	Zambia	JX303815	L3d3a1b	Zambia
JX303778	L0a2a2a	Zambia	JX303816	L1c2b1b1	Zambia
JX303779	L1b1a	Zambia	JX303817	L0a1bla	Zambia
JX303780	L2a1f1	Zambia	JX303818	L0d1b2a2	Zambia
JX303781	L3e1e1	Zambia	JX303819	L1b1a10b	Zambia
JX303782	L1c2a3a	Zambia	JX303820	L1c2b1b1	Zambia
JX303783	L1c2a1a	Zambia	JX303821	L3d1b3a	Zambia
JX303784	L0a2a1b	Zambia	JX303822	L3e2bla2	Zambia
JX303785	L1c3b1a	Zambia	JX303823	L0a1b1a	Zambia
JX303786	L0a2a2a2	Zambia	JX303824	L1c3a	Zambia
JX303787	L1b1a3	Zambia	JX303825	L2a1d2	Zambia
JX303788	L0d2b2	Zambia	JX303826	L0a2a2a	Zambia
JX303789	L3e2bla2	Zambia	JX303827	L3e1a3a	Zambia
JX303790	L1c2b1b1	Zambia	JX303828	L1c2a1a	Zambia
JX303791	L0d1c2a	Zambia	JX303829	L2a5	Zambia
JX303792	L2c2a1	Zambia	JX303830	L0a2a1b	Zambia
JX303793	L1c2a1a	Zambia	JX303831	L0a2a2a1	Zambia
JX303794	L1c2b1b1	Zambia	JX303832	L2a1f	Zambia
JX303795	L2a1d2	Zambia	JX303833	L1c2b1b1	Zambia
JX303796	L0a1bla1	Zambia	JX303834	L3d3a1b	Zambia
JX303797	L1c5	Zambia	JX303835	L0a2a2a1	Zambia
JX303798	L2a1g	Zambia	JX303836	L3d3a1b	Zambia
JX303799	L3e3b2	Zambia	JX303837	L3e1a3a	Zambia
JX303800	L1c2b1b1	Zambia	JX303838	L2a1d2	Zambia
JX303801	L1b1a10b	Zambia	JX303839	L3d3a1b	Zambia
JX303802	L3d3a1b	Zambia	JX303840	L1b1a10b	Zambia
JX303803	L3d3a1b	Zambia	JX303841	L2b2a	Zambia
JX303804	L3f1b1a1	Zambia	JX303842	L3f1b1a1	Zambia
JX303805	L2a1f3	Zambia	JX303843	L3f2a1	Zambia

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
JX303844	L3e2b	Zambia	JX303882	L2b1a3	Zambia
JX303845	L1c2b2	Zambia	JX303883	L1c2b1b1	Zambia
JX303846	L1b1a10b	Zambia	JX303884	L3e1f1a	Zambia
JX303847	L3e1d1	Zambia	JX303885	L3e1e1	Zambia
JX303848	L1c2a1a	Zambia	JX303886	L3e1d1a	Zambia
JX303849	L3e1d1a	Zambia	JX303887	L3e1a3a	Zambia
JX303850	L3e1d1	Zambia	JX303888	L3f1b1a1	Zambia
JX303851	L1c2b1b1	Zambia	JX303889	L3e1d1a	Zambia
JX303852	L2a1c1	Zambia	JX303890	L3blala	Zambia
JX303853	L2a1i1	Zambia	JX303891	L3e1d1	Zambia
JX303854	L3e1a3a	Zambia	JX303892	L1b1a3	Zambia
JX303855	L1c2a1a	Zambia	JX303893	L3e1a3a	Zambia
JX303856	L0k2a1a	Zambia	JX303894	L3f2a1	Zambia
JX303857	L2a1i1	Zambia	JX303895	L0k2a1a	Zambia
JX303858	L2a1b1a	Zambia	JX303896	L1b2a	Zambia
JX303859	L1b1a10b	Zambia	JX303897	L0d2b1a1	Zambia
JX303860	L1b1a10b	Zambia	JX303898	L1b1a3	Zambia
JX303861	L0k1b	Zambia	JX303899	L0d2b1a1	Zambia
JX303862	L2a1d2	Zambia	JX303900	L1c2b1b1	Zambia
JX303863	L2c2a1	Zambia	JX303901	L1b1a3	Zambia
JX303864	L3d3a1	Zambia	JX303902	L3e1d1	Zambia
JX303865	L0k1b	Zambia	JX303903	L0d2b1a1	Zambia
JX303866	L1b1a10b	Zambia	JX303904	L0a2a2a	Zambia
JX303867	L0k1b	Zambia	JX303905	L1b2a	Zambia
JX303868	L0k2a1a	Zambia	JX303906	L2a1g	Zambia
JX303869	L0a1a2	Zambia	JX303907	L1c2a1a	Zambia
JX303870	L2a1q	Zambia	JX303908	L3e1a3a	Zambia
JX303871	L1c3a1b	Zambia	JX303909	L2a1i1	Zambia
JX303872	L3e1a3a	Zambia	JX303910	L1c2b1b1	Zambia
JX303873	L2a1d2	Zambia	JX303911	L0a1a2	Zambia
JX303874	L2a1b1a	Zambia	JX303912	L1c2a1a	Zambia
JX303875	L3e1d1	Zambia	JX303913	L1c2a1a	Zambia
JX303876	L3b1a3	Zambia	JX524225	L2a1c1a2	<b>Brazil</b>
JX303877	L1c2a1a	Zambia	JX666328	L3b1a7	<b>USA</b>
JX303878	L3e1a2	Zambia	KC152939	L1c2bla1	Unknown
JX303879	L1c2a1a	Zambia	KC257334	L1c3b2	<b>USA</b>
JX303880	L2a1b1a	Zambia	KC257335	L1c3b2	<b>USA</b>
JX303881	L3ela3a	Zambia	KC257336	L1c3b2	<b>USA</b>

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
KC257337	L1c3b2	<b>USA</b>	KC345794	L0k1a2	Botswana
KC257338	L1c3b2	<b>USA</b>	KC345795	L0k1a2	Botswana
KC257339	L1c3b2	<b>USA</b>	KC345796	L0k1a2	Botswana
KC257340	L1c3b2	<b>USA</b>	KC345797	L0kla2	Botswana
KC257341	L1c3b2	<b>USA</b>	KC345798	L0d1b1a1	Botswana
KC257342	L1c3b2	<b>USA</b>	KC345799	LOd1b1a1	Botswana
KC257343	L1c3b2	<b>USA</b>	KC345800	LOd1b1a1	Botswana
KC257344	L1c3b2	<b>USA</b>	KC345801	L0d1b1a1	Botswana
KC345764	L0d1c	Angola	KC345802	LOd1b1a1	Botswana
KC345765	L0d1b1b1	Angola	KC345803	L0k1a2	Botswana
KC345766	L0d1b1b1	Angola	KC345804	L0kla2	Botswana
KC345767	L0d1b1b1	Angola	KC345805	L0k1a2	Botswana
KC345768	L0d1b1b1	Angola	KC345806	L0kla2	Botswana
KC345769	L0dlalbla	Angola	KC345807	L0d1b2a1	Botswana
KC345770	L0dlalbla	Angola	KC345808	LOd1c2a	Botswana
KC345771	L0d1b1b1	Angola	KC345809	L0d1b2b2a	Botswana
KC345772	L0d1b1b1	Angola	KC345810	LOdlclalal	Botswana
KC345773	L0dlalbla	Angola	KC345811	LOd1clala	Botswana
KC345774	L0d1b1b1	Angola	KC345812	LOd1c1a1a	Botswana
KC345775	L0dlalbla	Angola	KC345813	LOdlclalal	Botswana
KC345776	L0dlalbla	Angola	KC345814	L0d1c	Botswana
KC345777	L0d2a1a	Angola	KC345815	LOd1c2a1	Botswana
KC345778	L0d1c2	Angola	KC345816	L0d1c	Botswana
KC345779	L0d1c	Angola	KC345817	LOd1c1a1a	Botswana
KC345780	L0d1b1+@152	Angola	KC345818	L0d1c	Botswana
KC345781	L0d1b2b2	Angola	KC345819	L0d2a2	Botswana
KC345782	L0dlalbla	Angola	KC345820	L0d2a2	Botswana
KC345783	L0d1b1b1	Angola	KC345821	L0k1a2	Botswana
KC345784	L0d2a1a	Angola	KC345822	LOdlclala2	Botswana
KC345785	L0k1a2	Angola	KC345823	LOdlclalal	Botswana
KC345786	L0d1b2a	Botswana	KC345824	LOd1c1a2	Botswana
KC345787	L0d1b2a1	Botswana	KC345825	L0dlalal	Botswana
KC345788	LOd1b1a1	Botswana	KC345826	LOd1c1a1a	Botswana
KC345789	LOd1b1a1	Botswana	KC345827	L0d1c2a	Botswana
KC345790	L0d1b1a1	Botswana	KC345828	L0d1c2a	Botswana
KC345791	L0d1a	Botswana	KC345829	LOdlclala2	Botswana
KC345792	L0d1c2	Botswana	KC345830	LOd1c1a1a	Botswana
KC345793	LOd1bla1	Botswana	KC345831	LOdlalal	Botswana

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)




































<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
KC911364	L5b1a	Iran	KF055328	L3elala	<b>USA</b>
KC911395	L2a1f3	Iran	KF055329	L3f1b1a	<b>USA</b>
KC911506	L3e3a	Iran	KF055330	L2c	<b>USA</b>
KC911529	L3d1a1a	Iran	KF055331	L1c4b	<b>USA</b>
KC911533	L3d1a1a	Iran	KF055332	L2a1c4a1	<b>USA</b>
KF011502	L3f1b	Spain	KF055869	L2c	Spain
KF011503	L3f1b	Spain	KF055870	L2c	Spain
KF055291	L1b1a3	<b>USA</b>	KF161500	L3blala	Denmark
KF055293	L3e2b8	<b>USA</b>	KF162786	L4b	Denmark
KF055296	L3d1a1a	<b>USA</b>	KF179062	L1b1a8	<b>USA</b>
KF055297	L3d1b3	<b>USA</b>	KF255394	L2a111a	Dominican Republic
KF055298	L1c3a	<b>USA</b>	KF358472	L3e5e	Cameroon
KF055299	L2a1i1	<b>USA</b>	KF358473	L3e5c	Cameroon
KF055300	L3e3b	<b>USA</b>	KF358474	L3e5c	Cameroon
KF055302	L0a1a+200	<b>USA</b>	KF358475	L3e5ala	Cameroon
KF055303	L1b1a10	<b>USA</b>	KF358476	L3e5b	Niger
KF055304	L3e2b+152	<b>USA</b>	KF358477	L3e5ala	Cameroon
KF055305	L1b1a	<b>USA</b>	KF358478	L <sub>3</sub> e <sub>5</sub>	Cameroon
KF055306	L3b1a	<b>USA</b>	KF358479	L <sub>3</sub> e <sub>5</sub>	Chad
KF055307	L3e2b+152	<b>USA</b>	KF358480	L3e5f	Chad
KF055308	L3e1e	<b>USA</b>	KF358481	L3e5f	Chad
KF055309	L3dlalal	<b>USA</b>	KF358482	L3e5c	Nigeria
KF055310	L2c	<b>USA</b>	KF358483	L3e5b	Nigeria
KF055311	L2c2a	<b>USA</b>	KF358484	L3e5d	Nigeria
KF055313	L1b1a	<b>USA</b>	KF358485	L3e5	Cameroon
KF055314	L1b1	<b>USA</b>	KF358486	L3e5b	Cameroon
KF055315	L3e1e	<b>USA</b>	KF358487	L3e5d	Cameroon
KF055317	L1c2b2	<b>USA</b>	KF358488	L3e5b	Cameroon
KF055318	L2a1a1	<b>USA</b>	KF358489	L3e5b	Cameroon
KF055319	L2c	<b>USA</b>	KF358490	L3e5e	Cameroon
KF055320	L3e2bla1	<b>USA</b>	KF358712	L1c3b1a	Puerto Rico
KF055321	L3e4a	<b>USA</b>	KF450887	L1b1a3a	Pakistan
KF055322	L1b1a18	<b>USA</b>	KF450890	L2a1f3	Pakistan
KF055323	L2a1c+16129	<b>USA</b>	KF450894	L3dlala	Pakistan
KF055324	L3b1a4	<b>USA</b>	KF450895	L2a1g	Pakistan
KF055325	L2c1	<b>USA</b>	KF450901	L0a1bla1	Pakistan
KF055326	L2a1a2	<b>USA</b>	KF450910	L2a1a2	Pakistan
KF055327	L2ale1	<b>USA</b>	KF450917	L1c2b2	Pakistan

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)













<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
KJ185427	L2a5	Zambia	KJ185465	L1c1	Zambia
KJ185428	L2a1d2	Zambia	KJ185466	L1c2a2	Zambia
KJ185429	L2a1f	Zambia	KJ185467	L1c2b1b	Zambia
KJ185430	L0a1a2	Zambia	KJ185468	L2a1c5	Zambia
KJ185431	L0a1bla1	Zambia	KJ185469	L3blala	Zambia
KJ185432	$L0a1+16293$	Zambia	KJ185470	L3b1a11	Zambia
KJ185433	L0a2a1a	Zambia	KJ185471	L3e1e1	Zambia
KJ185434	L1b1a	Zambia	KJ185472	L3e1a3a	Zambia
KJ185435	L1c2b1b1	Zambia	KJ185473	L3e1	Zambia
KJ185436	L1c2b2	Zambia	KJ185474	L3e2b1a2	Zambia
KJ185437	L1cla2	Zambia	KJ185475	L3e1e1	Zambia
KJ185438	L1c2b1b1	Zambia	KJ185476	L0a2a1b	Angola
KJ185439	L1c2b1b1	Zambia	KJ185477	L0a2a2a	Angola
KJ185440	L2a1a2	Zambia	KJ185478	L0a2a2a	Angola
KJ185441	L2a5	Zambia	KJ185479	L0a2a2a	Angola
KJ185442	L2a1b1a	Zambia	KJ185480	L0a2a1b	Angola
KJ185443	L2b1a3	Zambia	KJ185481	L1c1b	Angola
KJ185444	L2b1b	Zambia	KJ185482	L1c2b2	Angola
KJ185445	L3e2b	Zambia	KJ185483	L1c2b1b1	Angola
KJ185446	L3e1	Zambia	KJ185484	L1c2b1b	Angola
KJ185447	L3e4a	Zambia	KJ185485	L1c3a1a	Angola
KJ185448	L3elala	Zambia	KJ185486	L2a1c+16129	Angola
KJ185449	L3e1a3a	Zambia	KJ185487	L2a1c4a1	Angola
KJ185450	L1c2a1a	Zambia	KJ185488	L2a1c4a1	Angola
KJ185451	L1c3a	Zambia	KJ185489	L2b1a3	Angola
KJ185452	L2a1b1a	Zambia	KJ185490	L3d1a1a	Angola
KJ185453	L1c3a1b	Zambia	KJ185491	L3d3a1	Angola
KJ185454	L1c2a1a	Zambia	KJ185492	L3e1e1	Angola
KJ185455	L2ala2ala	Zambia	KJ185493	L3e2b	Angola
KJ185456	L3e3b1	Zambia	KJ185494	L0a2a1b	Angola
KJ185457	L <sub>Of1</sub>	Zambia	KJ185495	L0a2a1b	Angola
KJ185458	L0a2a2a2	Zambia	KJ185496	L0a2a1b	Angola
KJ185459	L2a1d2	Zambia	KJ185497	L0a1b2	Angola
KJ185460	L2b1a3	Zambia	KJ185498	L0a2a1b	Angola
KJ185461	L0a2a1b	Zambia	KJ185499	L0a2a1b	Angola
KJ185462	L0a2a1b	Zambia	KJ185500	L0a2a1b	Angola
KJ185463	L0a2d	Zambia	KJ185501	L0a2a1b	Angola
KJ185464	L1c2b2	Zambia	KJ185502	L0a1b2	Angola

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)





<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
KJ185579	L1c2b1b1	Zambia	KJ185617	L3e1d1a	Zambia
KJ185580	L1c2b2	Zambia	KJ185618	L3e1d1	Zambia
KJ185581	L1c2ala	Zambia	KJ185619	L3e3b	Zambia
KJ185582	L2a1f3	Zambia	KJ185620	L3e1a3a	Zambia
KJ185583	L2a1f3	Zambia	KJ185621	L3ela3a	Zambia
KJ185584	L2a1c5	Zambia	KJ185622	L3e1d1a	Zambia
KJ185585	L2a1c5	Zambia	KJ185623	L3e1e1	Zambia
KJ185586	L2a1d2	Zambia	KJ185624	L3e2b	Zambia
KJ185587	L2a1bla	Zambia	KJ185625	L3e1d1a	Zambia
KJ185588	L2a1b1a	Zambia	KJ185626	L3e1a3a	Zambia
KJ185589	L2a1a1	Zambia	KJ185627	L3e1d1	Zambia
KJ185590	L2a1d2	Zambia	KJ185628	L3e1a3a	Zambia
KJ185591	L2a1d2	Zambia	KJ185629	L3e1a3a	Zambia
KJ185592	L2a5	Zambia	KJ185630	L3e1e1	Zambia
KJ185593	L2a5	Zambia	KJ185631	L3e2b	Zambia
KJ185594	L2a5	Zambia	KJ185632	L3e2a1b1	Zambia
KJ185595	L2a1c1	Zambia	KJ185633	L3e1a3a	Zambia
KJ185596	L2a1f3	Zambia	KJ185634	L3e1d1	Zambia
KJ185597	L2a1i1	Zambia	KJ185635	L3e2b	Zambia
KJ185598	L2a1i1	Zambia	KJ185636	L3e3b	Zambia
KJ185599	L2b1a3	Zambia	KJ185637	L3e3b2	Zambia
KJ185600	L2b1a3	Zambia	KJ185638	L3e3b1	Zambia
KJ185601	L2c2a1	Zambia	KJ185639	L3e1a3a	Zambia
KJ185602	L2c2b1b	Zambia	KJ185640	L3e3a	Zambia
KJ185603	L2c2a1	Zambia	KJ185641	L3e1d1a	Zambia
KJ185604	L2c2a1	Zambia	KJ185642	L3e1a3a	Zambia
KJ185605	L2c2a1	Zambia	KJ185643	L3e1a3a	Zambia
KJ185606	L2ela	Zambia	KJ185644	L3e1d1a	Zambia
KJ185607	L2e1a	Zambia	KJ185645	L3e1a3a	Zambia
KJ185608	L2e1a	Zambia	KJ185646	L3e2b	Zambia
KJ185609	L3bla11	Zambia	KJ185647	L3f1b4a	Zambia
KJ185610	L3bla11	Zambia	KJ185648	L5a2	Zambia
KJ185611	L3d3a1	Zambia	KJ185649	L5a2	Zambia
KJ185612	L3d3a1b	Zambia	KJ185650	L1c3bla	Zambia
KJ185613	L3d3a1	Zambia	KJ185651	L1c2a1a	Zambia
KJ185614	L3d4	Zambia	KJ185652	L3e1d1	Zambia
KJ185615	L3d3a1	Zambia	KJ185653	L0a2a1b	Zambia
KJ185616	L3e2bla2	Zambia	KJ185654	L0a2a1b	Zambia

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)





<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
KJ185731	L2a1c1	Zambia	KJ185769	L2a1a2	Angola
KJ185732	L2ala2ala	Zambia	KJ185770	L2a1a3c	Angola
KJ185733	L2ala2ala	Zambia	KJ185771	L2a1a3c	Angola
KJ185734	L2a5	Zambia	KJ185772	L2b1a3	Angola
KJ185735	L2a1bla	Zambia	KJ185773	L2c3	Angola
KJ185736	L3blala	Zambia	KJ185774	L2c2b1b	Angola
KJ185737	L3b1a1	Zambia	KJ185775	L2c2b1b	Angola
KJ185738	L3b1a11	Zambia	KJ185776	L3blala	Angola
KJ185739	L3e1a3a	Zambia	KJ185777	L3b1a1a	Angola
KJ185740	L3e1d1	Zambia	KJ185778	L3b1a10	Angola
KJ185741	L3e4a	Zambia	KJ185779	L3dlala	Angola
KJ185742	L3e1a3a	Zambia	KJ185780	L3d3a1	Angola
KJ185743	L3e1a3a	Zambia	KJ185781	L3e2b	Angola
KJ185744	L3e4a	Zambia	KJ185782	L3e1	Angola
KJ185745	L3e4a	Zambia	KJ185783	L3e4a	Angola
KJ185746	L3e4a	Zambia	KJ185784	L3e1e1	Angola
KJ185747	L3e1f1a	Zambia	KJ185785	L3e3b1	Angola
KJ185748	L5a2	Zambia	KJ185786	L3e1a2	Angola
KJ185749	L0a2a2a	Angola	KJ185787	L3e2bla2	Angola
KJ185750	L0a1b2	Angola	KJ185788	L3e3b1	Angola
KJ185751	L0a2a1a	Angola	KJ185789	L3e1e1	Angola
KJ185752	L0a2a1a	Angola	KJ185790	L3e2b	Angola
KJ185753	L0a1b1a	Angola	KJ185791	L3e2bla2	Angola
KJ185754	L0a1a2	Angola	KJ185792	L3e1e1	Angola
KJ185755	L0a2a1b	Angola	KJ185793	L3elala	Angola
KJ185756	L0a1b1a	Angola	KJ185794	L3e2b1a2	Angola
KJ185757	L0a1b1a	Angola	KJ185795	L3f1b4a	Angola
KJ185758	L0a1b2	Angola	KJ185796	L3f1b4a	Angola
KJ185759	L0a1b1a	Angola	KJ185797	L3f1b4a	Angola
KJ185760	L1b1a15	Angola	KJ185798	L3f1b4a	Angola
KJ185761	L1c2b1b	Angola	KJ185799	L3f1b1a	Angola
KJ185762	L1c1b	Angola	KJ185800	L3f1b4a	Angola
KJ185763	L1c2b1b1	Angola	KJ185801	L3f1b4a	Angola
KJ185764	L1c2a3	Angola	KJ185802	L0a2a2a	Angola
KJ185765	L1c2a2	Angola	KJ185803	L0a2a2a1	Angola
KJ185766	L1c3b1a	Angola	KJ185804	L0a2a2a	Angola
KJ185767	L1cla2	Angola	KJ185805	L0a1b2a	Angola
KJ185768	L2a1a	Angola	KJ185806	L0a1a2	Angola

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)



## Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)



## Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

















<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
KM101583	L3f1b1a	<b>USA</b>	KM101626	L2a1b	<b>USA</b>
KM101585	L3f1b1a	<b>USA</b>	KM101627	L2c	<b>USA</b>
KM101586	L3b2b	<b>USA</b>	KM101628	L1c5	<b>USA</b>
KM101587	L3f1b1a	<b>USA</b>	KM101629	L3e1	<b>USA</b>
KM101588	L1c3a	<b>USA</b>	KM101630	L2a1f	<b>USA</b>
KM101589	L3f1b+16292+150	<b>USA</b>	KM101631	L3b1a6	<b>USA</b>
KM101590	L2a1b1	<b>USA</b>	KM101632	L1c2b1c	<b>USA</b>
KM101591	L3e2b+152	<b>USA</b>	KM101634	L3f1b+16292+150	<b>USA</b>
KM101592	L2c	<b>USA</b>	KM101635	$L2a1b+143$	<b>USA</b>
KM101593	L2c	<b>USA</b>	KM101636	L2b2a	<b>USA</b>
KM101594	L2a1mla	<b>USA</b>	KM101637	L3d1b2	<b>USA</b>
KM101595	L1b1a	<b>USA</b>	KM101638	L3f1b4	<b>USA</b>
KM101596	L3e2a1b1	<b>USA</b>	KM101639	L <sub>2a1i</sub>	<b>USA</b>
KM101597	L3b3	<b>USA</b>	KM101640	L1b1a3a	<b>USA</b>
KM101598	L0a1a2	<b>USA</b>	KM101642	L2a1a3	<b>USA</b>
KM101599	L2c2	<b>USA</b>	KM101643	L2ala2ala	<b>USA</b>
KM101600	L3b2	<b>USA</b>	KM101644	L0a1a2	<b>USA</b>
KM101601	L2a1f	<b>USA</b>	KM101645	L3f1b4c	<b>USA</b>
KM101602	L3e2b	<b>USA</b>	KM101646	L3d1'2'3'4'5'6	<b>USA</b>
KM101603	L2a1f	<b>USA</b>	KM101647	L3e2a1b	<b>USA</b>
KM101604	L2a1c3a1	<b>USA</b>	KM101648	L3e2a1b1	<b>USA</b>
KM101606	L3e2b	<b>USA</b>	KM101649	L1b1a4	<b>USA</b>
KM101608	L3e3b	<b>USA</b>	KM101650	L1c2a1a	<b>USA</b>
KM101609	L2a1f	<b>USA</b>	KM101651	L2a1c	<b>USA</b>
KM101610	L2a1b1	<b>USA</b>	KM101652	L3b1a4	<b>USA</b>
KM101611	L2a1f2	<b>USA</b>	KM101653	L2a1c5	<b>USA</b>
KM101612	L3e2a1b1	<b>USA</b>	KM101654	L3k1	<b>USA</b>
KM101613	L3e2a1b3	<b>USA</b>	KM101655	L2a1a1	<b>USA</b>
KM101614	L3f1b+16292+150	<b>USA</b>	KM101656	L1b1a6	<b>USA</b>
KM101615	L2b2	<b>USA</b>	KM101657	L2a1c2a	<b>USA</b>
KM101616	L1b1a7a	<b>USA</b>	KM101658	L2a1a	<b>USA</b>
KM101617	L3e4a	<b>USA</b>	KM101659	L3b1a6	<b>USA</b>
KM101618	L2a1c5	<b>USA</b>	KM101660	L2c	<b>USA</b>
KM101619	L2c2	<b>USA</b>	KM101661	L3b3	<b>USA</b>
KM101620	L1b1a6	<b>USA</b>	KM101662	L0a1b2	<b>USA</b>
KM101621	L1bla7	<b>USA</b>	KM101663	L2d1a	<b>USA</b>
KM101622	L3e2a1b1	<b>USA</b>	KM101664	L2a1f	<b>USA</b>
KM101625	L1bla3a	<b>USA</b>	KM101665	L3e3a	<b>USA</b>

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)
































































































<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
MF621094	L3x2b	Spain	MF695865	L0a1blala	Kenya
MF621095	L3x2a1a	Jordan	MF695866	LOf	Kenya
MF621096	L3h1a1	Sudan	MF695867	L0a2a2a	Kenya
MF621097	L3h1a1	Sudan	MF695868	L3b1a11	Kenya
MF621098	L3h1a1	Sudan	MF695869	L0a1d	Kenya
MF621099	L3h1a2a1	Tunisia	MF695870	L3b1a1a	Kenya
MF621100	L3h1a2a1	Kenya	MF695871	L0a2a1b	Kenya
MF621101	L3h1a2a1	Sudan	MF695872	L1c1	Kenya
MF621102	L3h1a2a1	Kenya	MF695874	L1c2a3	Kenya
MF621103	L3h1a2a1	Sudan	MF695875	L0a2	Kenya
MF621104	L3h1a2b	Sudan	MF695876	L0a2a2a	Kenya
MF621105	L3h1a2b	Sudan	MF695877	L0a1'4	Kenya
MF621106	L3h1b1a	Saudi Arabia	MF695878	L0a	Kenya
MF621107	L3h1b2	Saudi Arabia	MF695879	L2ala2	Kenya
MF621108	L4ala	Saudi Arabia	MF695880	L0a2a2a	Kenya
MF621109	L4a2	Saudi Arabia	MF695881	LOf	Kenya
MF621110	L4a2	Rwanda	MF695882	L2a1a2	Kenya
MF621111	L4bla	Ivory Coast	MF695883	L <sub>0f1</sub>	Kenya
MF621112	L4b1	Sudan	MF695885	L3blala	Kenya
MF621113	L4b1	Sudan	MF695886	L3e3a	Kenya
MF621114	L4b1	Kenya	MF695887	L3e3a	Kenya
MF621115	L4b1	Sudan	MF695888	L0a2a2a	Kenya
MF621116	L4b1	Sudan	MF695889	L3e3a	Kenya
MF621117	L4b1	Sudan	MF695890	L3d1a1a	Kenya
MF621118	L4b2a1	Saudi Arabia	MF695891	L <sub>0f1</sub>	Kenya
MF621119	L4b2b	Equatorial Guinea	MF695892	L2d1a	Kenya
MF621120	L2a1c2a	Ghana	MF695893	L0a1d	Kenya
MF621121	L2a1c3b	Sudan	MF695894	L0f2a1	Kenya
MF621122	L2a1+143+16189	Saudi Arabia	MF695895	L0a2	Kenya
MF621123	L2b1a3	Nigeria	MF695896	L1b1a	Kenya
MF621124	L2d+16129	Mauritania	MF695897	L0a2a2a	Kenya
MF621125	L5a1a	Tanzania	MF695898	L0f <sub>2</sub> a	Kenya
MF621127	L1b2a	Nigeria	MF695899	L0a	Kenya
MF621128	L1clalalb1	<b>Equatorial Guinea</b>	MF695900	L2a1h	Kenya
MF621129	L1c2a1b	Nigeria	MF695901	L4b2a2	Kenya
MF621130	L0a1b1	Morocco	MF695902	L3e1e	Kenya
MF695863	L3e3a	Kenya	MF695903	L4b2a2	Kenya
MF695864	L0a2a1b	Kenya	MF695904	L3dlalal	Kenya

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)





















<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
NA18917	L1b1a+189	Nigeria	NA19114	L3ela3b	Nigeria
NA18921	L2b2	Nigeria	NA19116	L2a1e1	Nigeria
NA18923	L1b1a3	Nigeria	NA19117	L2ala2ala	Nigeria
NA18924	L1b1a3a	Nigeria	NA19118	L3e2a1b	Nigeria
NA18933	L1b1a15	Nigeria	NA19119	L3b1a7a	Nigeria
NA18934	L3e2a1b1	Nigeria	NA19121	L3d5a	Nigeria
NA19017	L3blala	Kenya	NA19122	L2c1a	Nigeria
NA19019	L5a1b	Kenya	NA19124	L3b1a4	Nigeria
NA19020	L3e2a	Kenya	NA19125	L2a1f	Nigeria
NA19022	L3blala	Kenya	NA19129	L2a1b1	Nigeria
NA19023	L2a1f	Kenya	NA19130	L2a1c3b2	Nigeria
NA19024	L2b1a3	Kenya	NA19131	L1b1a	Nigeria
NA19026	L3blala	Kenya	NA19133	L1b1a3	Nigeria
NA19027	L0a1a+200	Kenya	NA19135	L1b2a	Nigeria
NA19028	L2a5	Kenya	NA19137	L0a1a2	Nigeria
NA19030	L3dlalal	Kenya	NA19138	L3e2b3	Nigeria
NA19031	L <sub>0</sub> b	Kenya	NA19141	L3e1	Nigeria
NA19035	L3e1a3a	Kenya	NA19143	L3b1a7a	Nigeria
NA19036	L3b1a1a	Kenya	NA19144	L3d5a	Nigeria
NA19037	L3h1a1	Kenya	NA19146	L <sub>2ela</sub>	Nigeria
NA19038	L3h1a2a1	Kenya	NA19147	L3e2b6	Nigeria
NA19039	L0a3	Kenya	NA19149	L2a1c5	Nigeria
NA19041	L1b1a	Kenya	NA19150	L3e2b8	Nigeria
NA19042	L0a1a+200	Kenya	NA19153	L3d2b	Nigeria
NA19043	L5b2	Kenya	NA19156	L0a1a+200	Nigeria
NA19044	L3blala	Kenya	NA19157	L2a1f	Nigeria
NA19045	L2a5	Kenya	NA19159	L3f1b4c	Nigeria
NA19046	L2a5	Kenya	NA19160	L3e2b2	Nigeria
NA19092	L3e2a1b	Nigeria	NA19162	L3e2a2	Nigeria
NA19093	L2a1c5	Nigeria	NA19163	L1c3b2	Nigeria
NA19095	L2a1a2	Nigeria	NA19166	L3e1b1	Nigeria
NA19096	L2a1c3b2	Nigeria	NA19168	L3e1	Nigeria
NA19098	$L3b1a+152$	Nigeria	NA19171	L3e3b	Nigeria
NA19099	L2a1mla	Nigeria	NA19175	L3b1a8	Nigeria
NA19102	L2a1a1	Nigeria	NA19181	L3b1a8	Nigeria
NA19107	L3b2a	Nigeria	NA19182	L3f1b1a	Nigeria
NA19108	L2ela	Nigeria	NA19184	L3e1b1	Nigeria
NA19113	L3e2b+152	Nigeria	NA19185	L2b1a3	Nigeria

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
NA19187	L4b2b1	Nigeria	NA19309	L3e2b	Kenya
NA19189	L2a1f	Nigeria	NA19310	L3blala	Kenya
NA19190	L1b1a15	Nigeria	NA19311	L0a1a+200	Kenya
NA19195	L3e2a1b2	Nigeria	NA19312	L0a2a2a	Kenya
NA19196	L3e2a1b1	Nigeria	NA19315	L3e3b1	Kenya
NA19197	L2b2	Nigeria	NA19316	L3d1a	Kenya
NA19198	L1b1a15a	Nigeria	NA19317	L5a1c	Kenya
NA19200	L3e3b	Nigeria	NA19318	L5b2	Kenya
NA19201	L2e1	Nigeria	NA19319	L3e3b2	Kenya
NA19204	L3e2a	Nigeria	NA19320	L3b1a1a	Kenya
NA19207	L1b1a3	Nigeria	NA19321	L3blala	Kenya
NA19209	L2b1b	Nigeria	NA19323	L1c2a1a	Kenya
NA19210	L1b1a3	Nigeria	NA19324	L3b1a1a	Kenya
NA19213	L3f1b3	Nigeria	NA19327	LOf	Kenya
NA19214	L3e2a1b1	Nigeria	NA19328	L0a2a2a	Kenya
NA19216	L0a1a2	Nigeria	NA19331	L3b1a1a	Kenya
NA19217	L3e1	Nigeria	NA19332	L5b2	Kenya
NA19220	L3d1	Nigeria	NA19334	L3blala	Kenya
NA19222	L3b1a8	Nigeria	NA19338	L3b1a1a	Kenya
NA19223	L1b1a3	Nigeria	NA19346	L3blala	Kenya
NA19228	L2a1c2a	Nigeria	NA19347	L3blala	Kenya
NA19229	L2c2	Nigeria	NA19350	L0a1a+200	Kenya
NA19235	L3e2b+152	Nigeria	NA19351	L2a1a2	Kenya
NA19236	L3e2b+152	Nigeria	NA19352	L3blala	Kenya
NA19238	L3e2b5	Nigeria	NA19355	L3blala	Kenya
NA19239	L2a1p	Nigeria	NA19359	L4b2a2	Kenya
NA19247	L2b	Nigeria	NA19360	L3blala	Kenya
NA19248	L2b3a	Nigeria	NA19371	L3e1a2	Kenya
NA19250	L1bla3a1	Nigeria	NA19372	L3blala	Kenya
NA19253	L2b2a	Nigeria	NA19373	L5b1	Kenya
NA19256	L1c3b2	Nigeria	NA19374	L5b1	Kenya
NA19257	L3b1a10	Nigeria	NA19375	L3b1a1a	Kenya
NA19259	L4b2b	Nigeria	NA19376	L2a4b	Kenya
NA19260	L3e3b	Nigeria	NA19377	L3h1a2a1	Kenya
NA19262	L3d1b2	Nigeria	NA19378	L1c2b1a	Kenya
NA19266	L2a1a1	Nigeria	NA19379	L0a1a+200	Kenya
NA19307	L3blala	Kenya	NA19380	L3blala	Kenya
NA19308	L5a1a	Kenya	NA19381	L2a1q	Kenya

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>	<b>Sample ID</b>	<b>Haplogroup</b>	<b>Country</b>
NA19382	L0a1blala	Kenya	NA19455	L5b2	Kenya
NA19383	L4b1	Kenya	NA19456	L3b1a1a	Kenya
NA19384	L3h1b1a	Kenya	NA19457	L3h1a1	Kenya
NA19385	L1c1d	Kenya	NA19461	L5b2	Kenya
NA19390	L1c3b1a	Kenya	NA19462	L1b1a15	Kenya
NA19391	L3ele	Kenya	NA19463	L4b2a2b	Kenya
NA19392	L2b1a3	Kenya	NA19466	L0a1blala	Kenya
NA19393	L2a4b	Kenya	NA19467	L0a1c1	Kenya
NA19395	L2a1f	Kenya	NA19468	L3b1a1a	Kenya
NA19396	L3blala	Kenya	NA19469	L2a2b1	Kenya
NA19397	L3blala	Kenya	NA19470	L2a2b1	Kenya
NA19398	L3f2a1	Kenya	NA19471	L1b1a3	Kenya
NA19399	L3e1e	Kenya	NA19472	L3blala	Kenya
NA19401	L3x1a2	Kenya	NA19473	L3blala	Kenya
NA19402	L0a2a2a	Kenya	NA19474	L <sub>0</sub> b	Kenya
NA19404	L5b1b	Kenya	NA19475	L3blala	Kenya
NA19428	L3blala	Kenya	NA19625	$L2a1b+143$	<b>USA</b>
NA19429	L3blala	Kenya	NA19700	L3e2b+152	<b>USA</b>
NA19430	L0a1a2	Kenya	NA19703	L0a1b1a	<b>USA</b>
NA19431	L1bla	Kenya	NA19704	L3e2ala	<b>USA</b>
NA19432	L4b2a2c	Kenya	NA19707	L3e2a	<b>USA</b>
NA19434	L4b2a2c	Kenya	NA19711	L3e3b	<b>USA</b>
NA19435	L4b2a2c	Kenya	NA19712	L1c2b1c	<b>USA</b>
NA19436	L3blala	Kenya	NA19713	L0a2a2a1	<b>USA</b>
NA19437	L3i1	Kenya	NA19818	L3e1a3a	<b>USA</b>
NA19438	L3a2	Kenya	NA19819	L3e2b+152	<b>USA</b>
NA19439	L3blala	Kenya	NA19834	L1b1a	<b>USA</b>
NA19440	L0a1c	Kenya	NA19835	L2c5	<b>USA</b>
NA19443	L2a2b1	Kenya	NA19900	L3e3b	<b>USA</b>
NA19444	L4b2a2c	Kenya	NA19901	L3f1b1a	<b>USA</b>
NA19445	L4b2a2c	Kenya	NA19904	L0a2a2a	<b>USA</b>
NA19446	L3x1a1	Kenya	NA19908	L2a1c	<b>USA</b>
NA19448	L0a1blala	Kenya	NA19909	L1b2	<b>USA</b>
NA19449	L0a1blala	Kenya	NA19913	L3b1a+@16124	<b>USA</b>
NA19451	L3blala	Kenya	NA19914	L1c3a1b	<b>USA</b>
NA19452	L2a1a2	Kenya	NA19916	L1b1a9	<b>USA</b>
NA19453	L4b2a2c	Kenya	NA19917	L1c1b	<b>USA</b>
NA19454	L <sub>0f1</sub>	Kenya	NA19920	L3h1b2	<b>USA</b>

Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

## Table S1 - Complete dataset analysed for mtDNA studies including the respective haplogroup and country. (Continuation)

