

# WELCOME TO *YOUR* STORY

**Patient Name**

Barcode ID:

Date of birth: 00/00/0000

Reporter date: 00/00/0000



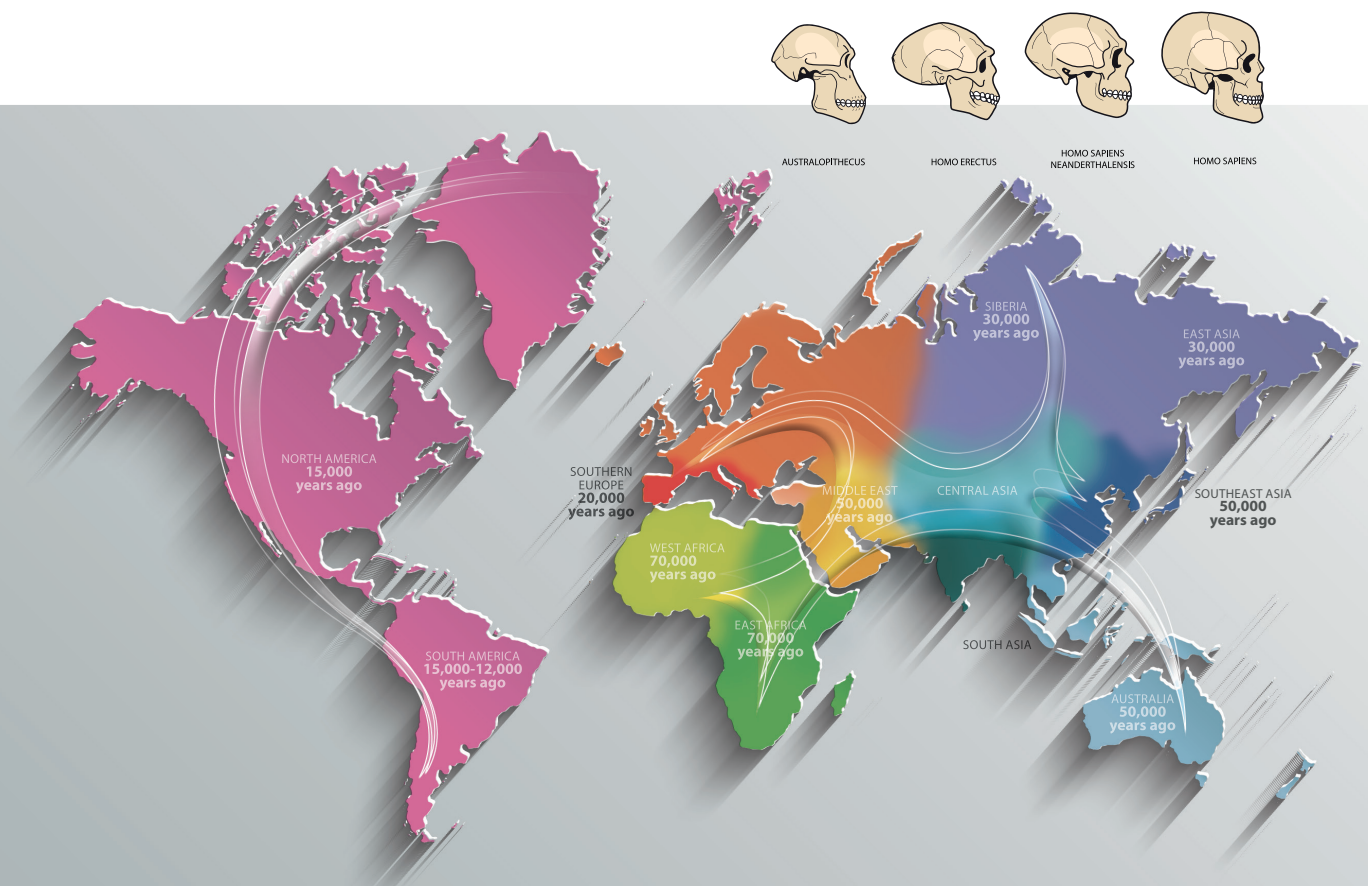
## Once upon a time...

For many years now, researchers have been using DNA to infer the origin of our species. Whilst this is still a relatively new science, and the story is constantly being updated, there are several facts that are well established.

*Homo sapiens sapiens*, also known as the modern human, had a unique origin in Africa some 200 000 - 300 000 years ago and was largely restricted to this continent until as recently as 60 000 - 70 000 years ago.

At that time, a small group of East Africans migrated beyond Africa's borders and populated the other continents. When your first ancestors travelled beyond the African continent and began mixing with other population groups, their genetic make-up began to transform. As time passed, variations in their DNA were passed on through the generations; to be inherited by you. Even today, the average European DNA contains approximately 2.3% Neanderthal DNA, arising from the mixing between these two population groups. With the genetic information we have collated on a diverse range of anthropological groups or haplogroups, we can now provide you with information regarding your ancestors.

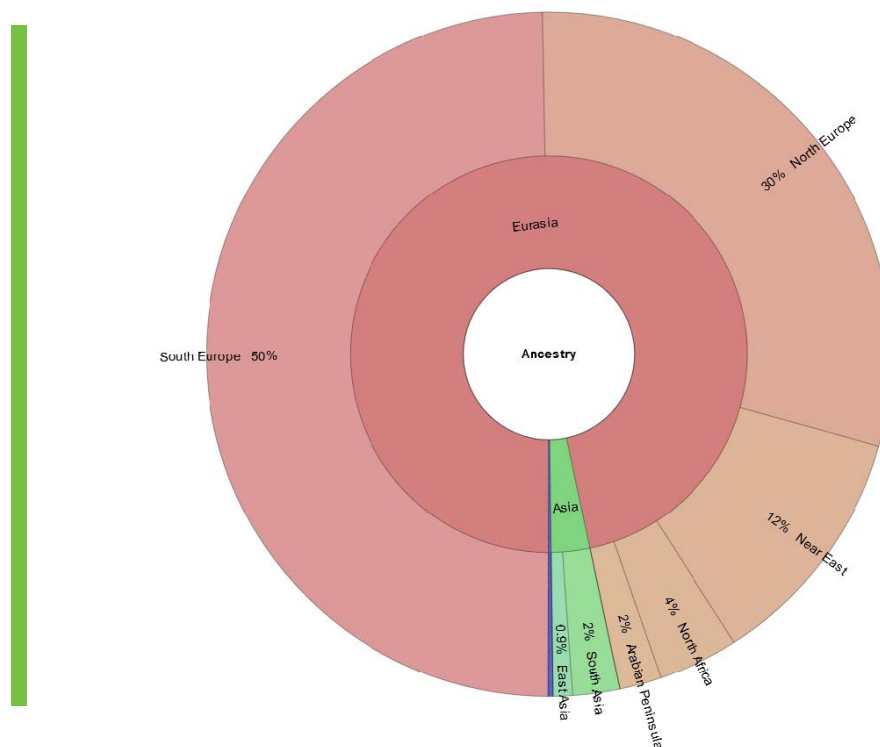
The image below shows the movement of the modern human, *Homo sapiens sapiens*, out-of-Africa.



## Your Genetic Ancestry

Each human has 23 pairs of **chromosomes**; 22 of which, known as the **autosomes**, comprise the largest part of our genome. All of our ancestors have contributed to the information carried in these 22 chromosome pairs. By meticulously analysing and comparing the genomes of different population groups, we can pinpoint the ancestral group to which you are most closely genetically related. For the purpose of this comparison, 10 ancestral groups were used: West Africa, East Africa, North Africa, Near East, Arabian Peninsula, South Europe, North Europe, South Asia, East Asia and Native American.

This graph represents the percentage of each of these populations within your own genome.



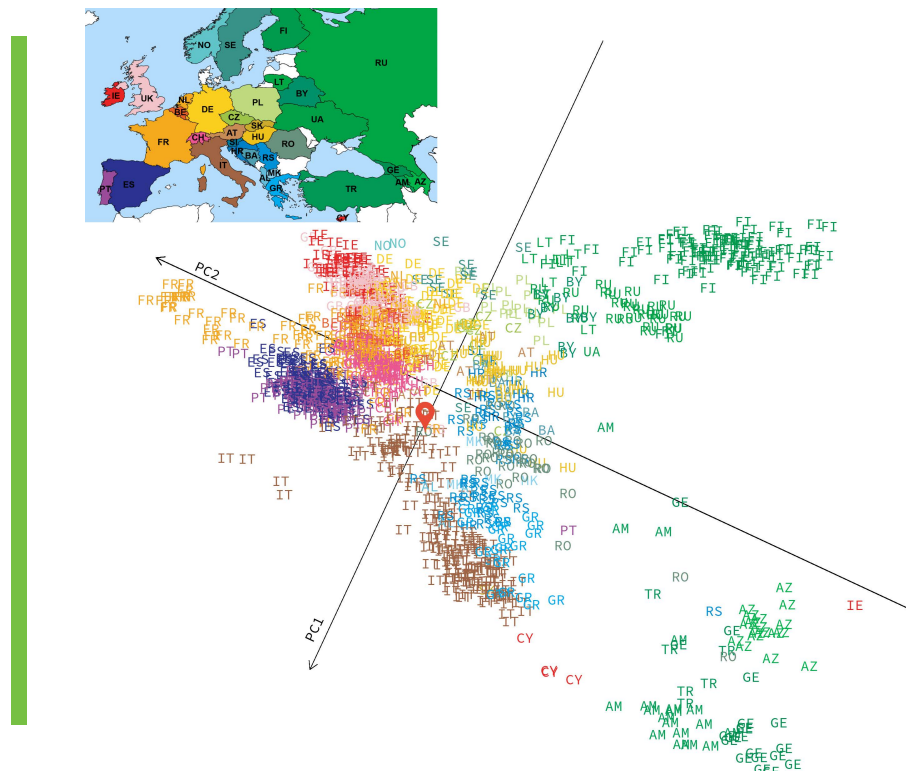
This is a hierarchical representation: the outside circle represents the 10 ancestral groups while the inner circle represents the grouping of the three main human population groups: Africa (Sub-Saharan), Asia and Eurasia.

Geographically, some of these groupings may seem odd, but it is important to remember that they indicate **genetic ancestral grouping**. For example, in this instance, "North Africa" falls under "Eurasia" because this population was mainly founded by individuals from the Near East.

## Projection of your genetic profile on main ancestral group

To provide you with more information on the possible origin of your primary ancestral background, we perform an analysis called Principle Component Analysis (PCA). PCA is a clustering technique that allows us to group genetically similar individuals together in a two-dimensional representation. Given that genetic diversity will increase with geographic distance (as it is more probable to mate with close by neighbours than with distant neighbours), this analysis can estimate the geographic origin of an individual based on their genetic results.

The below plot is created by overlaying the genetic profiles of established reference samples on a map of the relevant continent.



Your highest scoring ancestral group was Europe.

The above image illustrates the projection of your genetic diversity on European reference samples, with each colour representing a different country or region on the map. Individuals with closer relationships have been plotted closer to each other, as their genetic diversity is less. The further away an individual travels from their origin, and reproduces, the greater the diversity will be in subsequent generations.

YOUR GENETIC ANCESTRY IS INDICATED BY THE RED SYMBOL, SHOWING THAT YOU ARE GENETICALLY MOST SIMILAR TO THE NEARBY POPULATION GROUPS

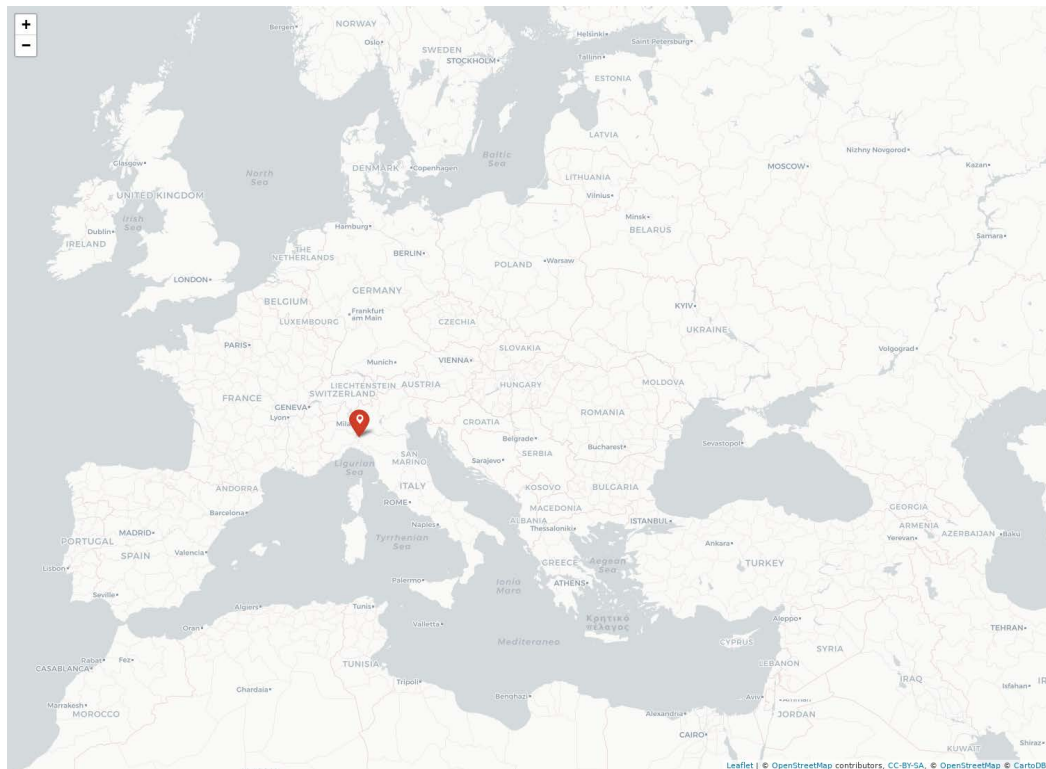


## Genetic GPS

Genetic GPS uses the Spatial Ancestry Analysis (SPA) tool to analyse your genetic profile, taking into account reference genetic profiles and their geographic coordinates.

This tool provides an estimate of the geographic coordinates of the origin of your ancestors.

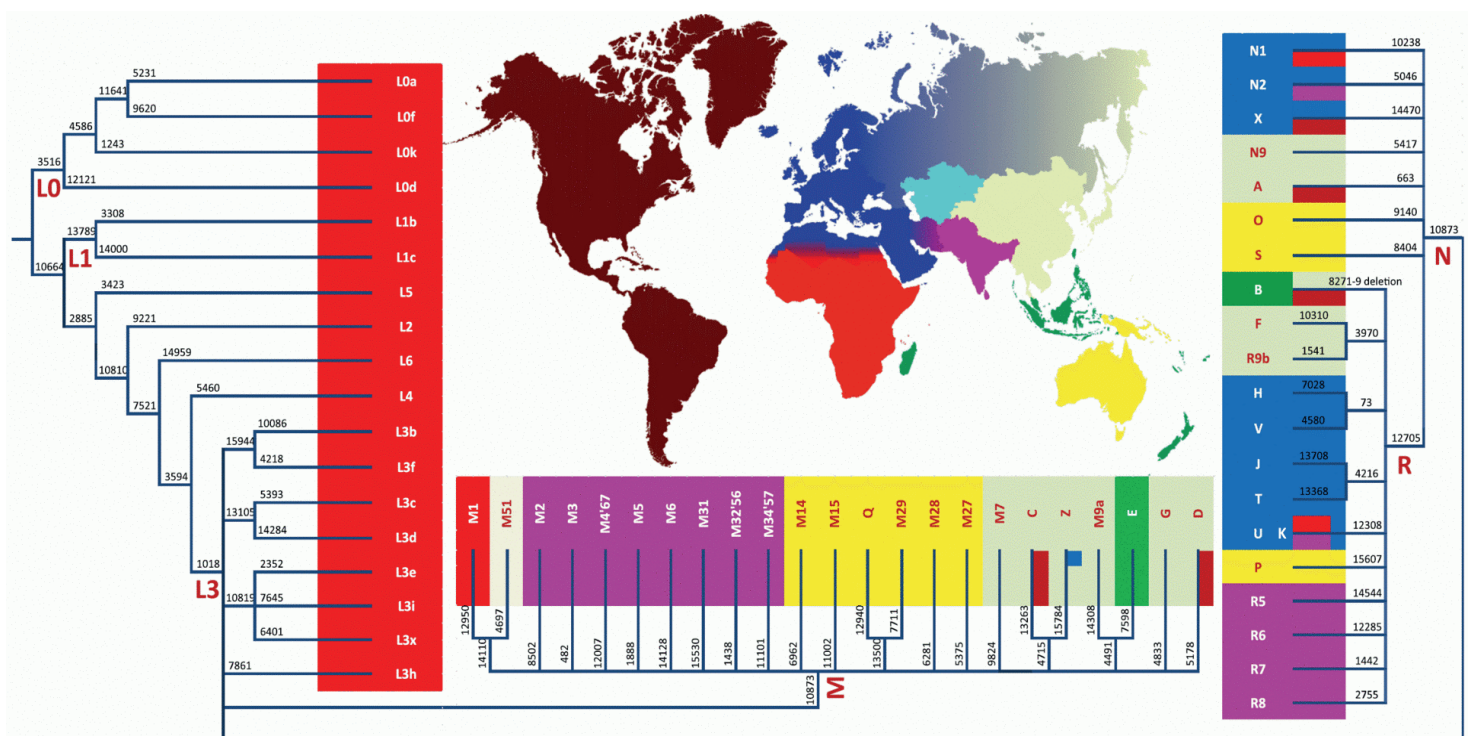
Please allow for an error margin of  $\pm 200\text{km}$ .



## Maternal Lineage

In addition to our nuclear DNA (our 23 pairs of chromosomes), humans also have mitochondrial DNA. This DNA can be found in the mitochondria and is present in many copies. Both males and females have mitochondrial DNA, however, it is passed down, almost unchanged, solely from a mother to her children; establishing the maternal lineage.

The image below represents a phylogenetic tree of mitochondrial DNA haplogroups. A haplogroup is a group of individuals that have the same maternal ancestor, sharing a group of genetic variants specific to a geographical region. Haplogroups are identified using a naming system of letters and numbers. In this tree, the older groups are indicated in red and originate in sub-Saharan Africa. The most recent large branches, N and M, are dispersed in Eurasia and East Asia. The brown colour in America indicates only the Native Americans.



The above image shows only the primary branches of the phylogenetic Tree. In actual fact, these haplotypes can be further sub-divided multiple times to give rise to many more branches.

## Your Results

**MITOCHONDRIAL HAPLOTYPE** 73G 263G 709A 1438G 2706G 4216C 4769G 4917G  
7028T 8860G 10463C 11251G 11812G 14233G 14766T 14905A 15326G 15452A  
15607G 15928A

**MATERNAL HAPLOGROUP** T2

**GEOGRAPHICAL REGION** Eurasia

### BRIEF DESCRIPTION OF THE HAPLOGROUP

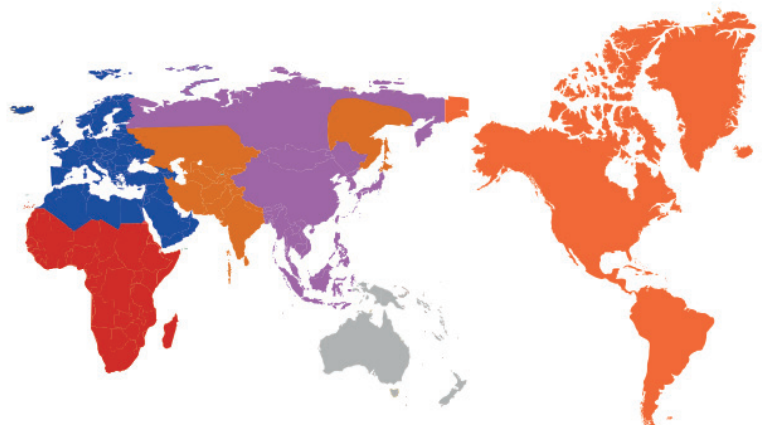
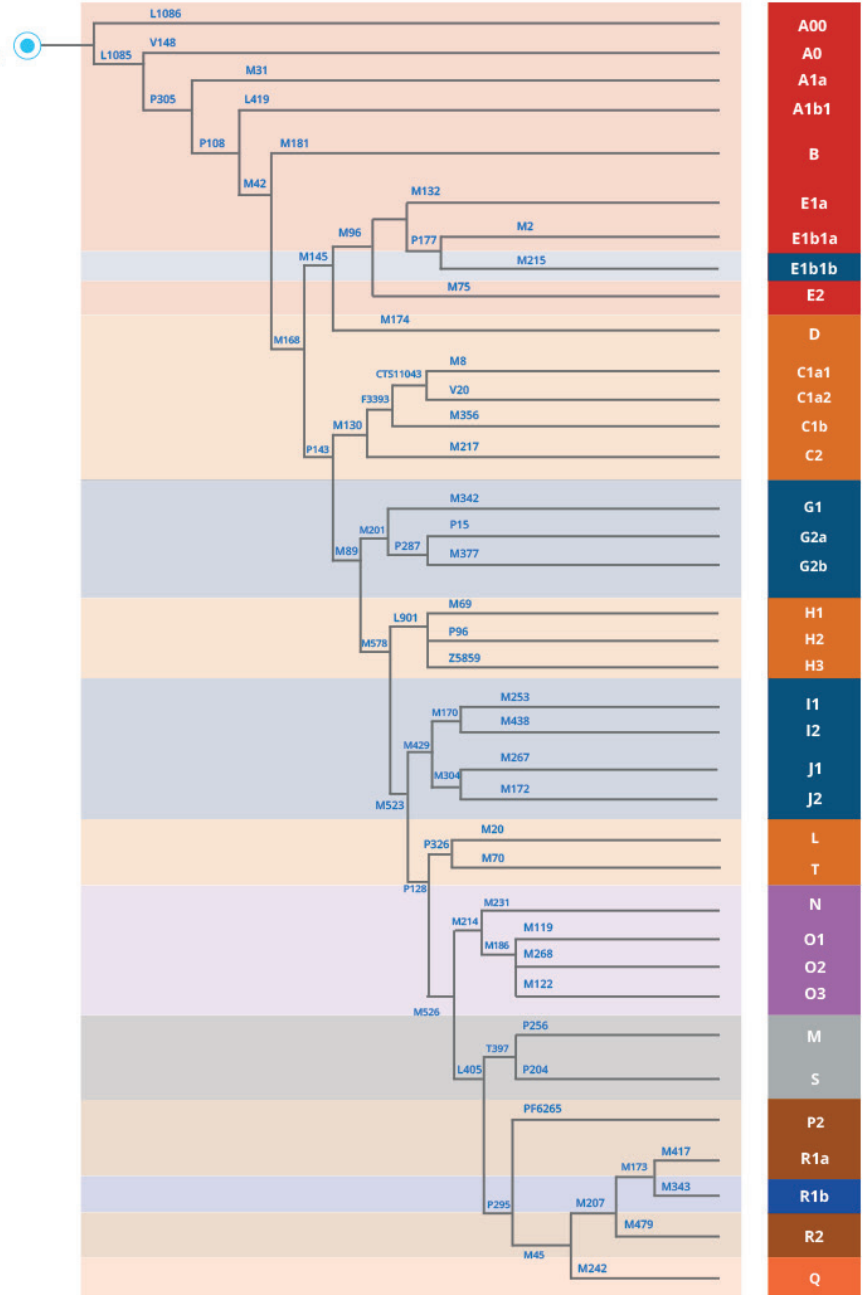
J and T haplogroups share an ancestral in the Near East at around 50,300 years ago, and geneticists thought they were introduced to Europe with the Neolithic. However, improvements in the molecular clock, showed that the entrance of J and T lineages in Europe must have occurred earlier, during the glacial period, and that they were already in Europe when the agriculture knowledge was introduced. The subgroup T2 is around 78,700 years old, and it is more frequent in the Netherlands (72%), Sardinia (70%), Iceland (70%), Switzerland (9.5%), Hungary (8.5%) and Ukraine (8.5%), as well as among many ethnic groups around the Caucasus such as the Kumyks (70%), Azeri (9.5%) and Georgians (9%).

## Paternal Lineage

The final pair of chromosomes we are interested in, are the sex chromosomes. In women, these are the same (XX) whereas they differ in men (XY), with the X chromosome deriving from the mother and the Y chromosome from the father. Because the Y chromosome only exists in males, this allows for the trace of your paternal lineage.

As with the map for maternal lineage, the image on the right shows the haplogroups of the Y chromosome – identified using a naming system of letters and numbers. A haplogroup is a group of individuals that have the same paternal ancestor, sharing a group of genetic variants specific to a geographical region.

Y-DNA  
"ADAM"





## Your Results

**DERIVED VARIANT IN THE Y CHROMOSOME TREE** J-M92

**PATERNAL HAPLOGROUP** J2a7b7

**GEOGRAPHICAL REGION** Eurasia

### BRIEF DESCRIPTION OF THE HAPLOGROUP

The paternal J2 haplogroup may have appeared somewhere in the Near East by the end of the last glaciation, between 75,000 and 22,000 years ago. The oldest known J2a samples were identified in remains from northern Iran, dating from 9700-8600 BCE, and from Georgia, dating from 7940-7600 BCE. The worldwide highest J frequencies are found in Caucasus, followed Cyprus (37%), Crete (34%), northern Iraq (28%), Lebanon (26%), Turkey (24%), Greece (23%), Central Italy (23%), Sicily (23%), South Italy (27.5%), and Albania (79.5%), as well as among Jewish people (79 to 25%). Several common Italian J2a subclades are found mainly in the south of Italy (including J-M92) and are likely to be of Greek origin.

## Admixture with Neanderthal

When modern humans migrated out of Africa, they interbred with other human groups such as Neanderthals. Current European and Asian genomes have an approximate 2.3% Neanderthal genetic input. African genomes do not contain Neanderthal input as this group was never present on the African continent.

ADMIXTOOLS allows us to infer the Neanderthal input into your genetic profile.

**YOUR PERCENTAGE VALUE IS**    2.47 %

## Glossary

ADMIXTURE	The result of interbreeding between two or more previously isolated populations within a species.
ANCESTRAL HAPLOGROUP	A genetic population group of people who share a common ancestor. Haplogroups are assigned letters of the alphabet, and refinements consist of additional number and letter combinations.
AUTOSOME	Any chromosome that is not a sex chromosome.
CHROMOSOME	A threadlike structure of chromatin, found in the nucleus of most living cells, carrying genetic information in the form of genes.
MITOCHONDRIAL DNA	Mitochondrial DNA is the DNA housed inside the mitochondria; an organelle found within the cell. Mitochondrial DNA is inherited solely from the mother.
NEANDERTHALS	An extinct species or subspecies of archaic humans who lived in Eurasia until about 40 000 years ago.
NUCLEAR DNA	The DNA contained within the nucleus of a cell, stored in 23 pairs of chromosomes.
PHYLOGENETICS	The study of evolutionary relationships among biological entities – often species, individuals or genes.
Y CHROMOSOME	The male sex chromosome passed on from father to son.



From the laboratories of



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