# Mitochondrial DNA Survey by Dr Peter Forster, Molecular Genetics Laboratory McDonald Institute for Archaeological Research, Cambridge

#### What we did

We analysed your mitochondrial DNA and compared it with samples in a database containing all published mitochondrial DNA sequences - about 20,000 sequences of which about 2,000 are African. This database is increasing all the time, but unfortunately doesn't yet give exact matches for everyone in the survey.

#### What is mitochondrial DNA?

Mitochondrial DNA (mtDNA) is unlike any of the other DNA which you inherit from your parents in that it is contained in small particles called mitochondria found in most of the cells of our bodies. Mitochondria are often called the cell's energy factories, because they are where most of our energy is converted from our food. They contain their own DNA, and are inherited only from our mothers, so mtDNA is a way of tracking our maternal ancestry - our mothers, our mother's mother and so on.

#### Mitochondrial DNA and history

About 20 years ago, the mtDNA of a European woman in Cambridge, was sequenced and hers is known as the Cambridge reference sample. All results today, including yours, are compared with that sample. As a general rule, the more differences your sequence has from the reference sample, the more distant in time your connections are, on your mother's side, from a modern European. On average

scientists have calculated that there is about one change in every 10,000 years from the first modern human - referred to as "mitochondrial Eve" who lived approximately 150,000 years ago in Africa.

Soon afterwards, an early expansion of modern humans populated Africa and left its genetic footsteps in the mitochondrial DNA (L1 types) found particularly in the KhoiSan people today. This early expansion appears not to have left Africa. Probably Neanderthals, Homo erectus and the harsh ice-age climate prevented successful colonisation of Eurasia. But tens of thousands of years later, an east African re-expansion (60-80,000 years ago) repopulated Africa (L2 and L3 types), and ultimately led to a migration out of African of at least one mtDNA subtype (L3a). L3 is the most widespread African type today.

At the McDonald Institute in Cambridge we date the out-of-Africa migration of the ancestral mtDNA types that founded all Eurasian lineages at 54,000 years ago. Then around 30,000 years ago, the east Asian, Indian and European mtDNA pools seem to have separated from each other into small founder populations. The east Asian expansion entered America about 25,000 years ago but was then driven back on both sides of the Pacific to more southerly latitudes during the Last Glacial Maximum around 20,000 years ago. Repopulation of northern Asian latitudes occurred after the Last Glacial Maximum, obscuring the ancestral Siberian gene pool of native Americans. All this happened before the onset of agriculture 10,000 years ago. These populations are shown schematically in the diagram below.

The earliest mtDNA sequences arose in Africa, as L1, from which L2 and L3 emerged. People with L3 mtDNA sequences left and types M and N and late R arose. Other non-African populations have emerged from these, for example F,G, M1, Y and Z in Asia, A, B, C, D and X in the Americas and so on.

## **Interpreting Your Results**

The part of the mtDNA we have sequenced is a section called HVS1 (hypervariable sequence 1) because we find this is where there is most variation.

We list the variations of your mtDNA from the reference sample, and also the samples in the database which they're closest to. These variations are not connected in any way to health or disease.

## Here's a key to how to read it, taking F103 as an example

Results of search within HVSI Characters under consideration within HVSI: from np 16093 to np 16362 This is the region of the mtDNA we sequenced

Mutations of the search sequence within HVSI 16209.C 16223.T 16292.T 16295.T 16311.C

F103's mtDNA differs from the Cambridge Reference Sample in the five positions shown above, with position 16209 being substituted for a C (cytosine) 16223 with a T (thymine) and so on.

Best matches at distance 0 Frequency of best matches 1

## The best match in the database was one single exact ("distance 0") match

RAN TUK 176 This is an abbreviation of the scientific paper the sample came from Frequency: 1 There was one person with this result Mutations in HVSI: 16209.C 16223.T 16292.T 16295.T 16311.C These are the differences between this sample and the Cambridge Reference Sample Tukulor tribe, Senegal This is where the sample in the database came from, i.e. from the Tulukor people of Senegal 13.00N 1300W This is the geographical co-ordinate of the sample, i.e. the place it was taken

So F103 now knows that someone belonging to the Tukulor people living in present day Senegal shares common maternal ancestry with her - more recently than anyone else we currently know about.

2nd best matches at distance 10 Frequency of 2nd best matches 10

There were 10 sequences in the database with just one difference from F103s ("distance 10" in scientific)

KRI NUB 21 Frequency 1 Mutations in HVSI: 16037.N 16209.C 16292.T 16311.C Nubia 20.00N 30.00E

KRI NUB 27 Frequency 1 Mutations in HVSI: 16209.C 16223.T 16292.T 16311.C Nubia 20.00N 30.00E

Ironically, those of you who have the most variations from the Cambridge Reference Sample will often find the fewest matches. This is a reflection of the shortage of African samples currently held in the database.

On this website, you will find a map showing where the matches closest to your sample originate.