

Mitochondrial DNA and the ancient organisms in your cells

This is the third of three articles about DNA and its use in genealogical research. The first article dealt with the amazing world of DNA and what it does. The second article was about Y-DNA and why it is so useful in genealogical research. The third article is about mitochondrial DNA (shortened to mtDNA) and how it is passed only down the female line. As we shall see, it is not as useful as Y-DNA in genealogical research which is why



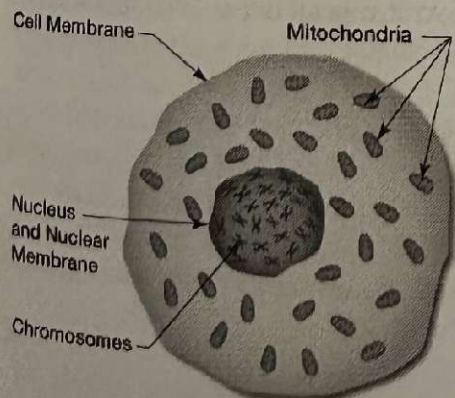
As before, you can contact me at: ronald.east@btinternet.com if there are any points you would like to discuss.

First, a recap:

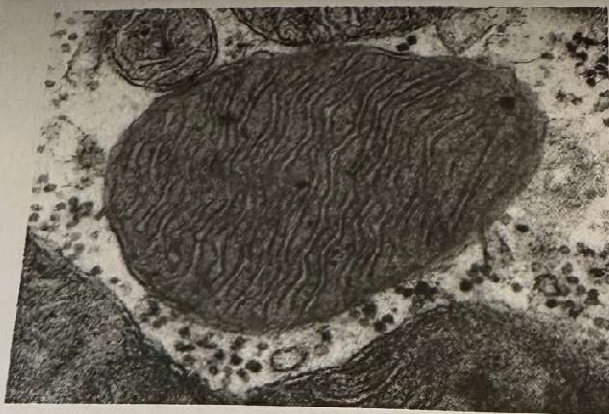
Nearly every cell in your body contains an exact copy of 46 chromosomes in its nucleus that were formed as a set when your mother's egg (with 23 chromosomes) was fertilised by your father's sperm (also with 23 chromosomes). The chromosomes are made from DNA which is itself made up of just four chemicals called bases which we labelled A, G, C and T. Scattered along the chromosomes are genes which determine who and what we are; they are our inheritance but between the genes are vast strings of junk DNA that do nothing at all. About 95% of the 3.2

billion base pairs in our DNA is junk DNA but it is in this junk DNA on the Y-chromosome (which all men carry) that clues are found about our family history.

All of our inherited DNA is contained in the nucleus but surrounding it in the cell is a chemical soup called cytoplasm. It is in this cytoplasm that we find mitochondria (singular: mitochondrion).



Mitochondria are the powerhouses of the cell for it is in these mitochondria that energy is released from a chemical reaction between the nutrients we obtain from food and the oxygen that we breathe in. Nearly all cells contain several mitochondria but some, like the hard working muscle cells, will contain many hundreds of mitochondria.



A close up picture of a mitochondrion.

Now comes the scary bit.

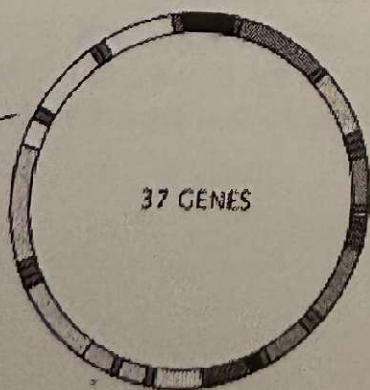
Mitochondria have their own DNA. It is a helix, just like nuclear DNA but tiny, only 16569 base pairs in length. It is shaped into a ring and, furthermore, there are several copies of the DNA in each mitochondrion (remember, there is only one copy of the DNA in the nucleus). Note that mitochondrial DNA will be written as *mtDNA* in the rest of this article

Does this mean that each cell in our body has been invaded by another kind of living organism with its own DNA? Well, in a sense, yes. Scientists have puzzled over the origins of mitochondria. They look very similar to bacteria and have the same ring-shaped DNA. Perhaps billions of years ago mitochondria were once free-living bacteria that had discovered how to use oxygen to release energy from nutrients. Other cells may have tried to digest them but preferred the extra energy supply instead. These cells then evolved to live in a mutually beneficial partnership with the energy producing bacteria they had engulfed – and eventually evolved into the cells with built-in energy supplies from which we are made!

Sperm cells contain mitochondria in their tails; they need the energy that the mitochondria supply to swim to the egg. Egg cells also contain mitochondria to supply enough energy to keep the egg alive for a few days. However, when a sperm enters the egg to fertilise it, the sperm leaves its swimming tail behind – and, with it, all the mitochondria. The fertilised egg, which will develop into a human being, therefore contains only mitochondria from the egg – in other words from the mother. Both sons and daughters have mtDNA from their mother but it is only passed on through the female line.

There are only 37 genes in mtDNA (compared with about 25000 genes in nuclear DNA) and there is no junk DNA! If you recall, information about family history on the male side is found in the junk DNA of the Y-chromosome. This junk DNA contains sequences of bases that 'stutter' and can be read to give information about family history.

In mtDNA there is no junk DNA but there are two areas where the bases can change. They are called Hypervariable Region 1 (HVR1) and Hypervariable Region 2 (HVR2) and are located in the black sector at 12 o'clock in the diagram.



MITOCHONDRIAL DNA

Thirty years ago, the order of the 16569 bases in a sample of mtDNA was worked out by a group of scientists in Cambridge. The sequence they found is called the Cambridge Reference Sequence or

CRS and it is against this original sequence that all mtDNA is now compared.

Here is a small part of the CRS in HVR1 from 16130 bases to 16139 bases:

Location	16130	16131	16132	16133	16134	16135	16136	16137	16138	16139
CRS	T	A	T	T	G	T	A	C	G	G

So what might happen if a sample of your mtDNA is examined (do not forget that men also carry their mother's mtDNA but they cannot pass it on to their offspring)?

One possibility is that, compared to the CRS, one base in your mtDNA has been replaced. Instead of having base A at position 16136, your mtDNA has base C; this is an example of a *mutation*:

Location	16130	16131	16132	16133	16134	16135	16136	16137	16138	16139
CRS	T	A	T	T	G	T	A	C	G	G
Sample	T	A	T	T	G	T	C	C	G	G

This mutation is written **16136C** which means that the base at position 16136 in the CRS has been replaced by base C in your sample of mtDNA.

A second kind of mutation is the insertion of an extra base between two others in the sequence. Here, for example, an extra copy of base C has been inserted at position 315 in HVR2:

Location	310	311	312	313	314	315		316	317	318	319
CRS	T	C	C	C	C	C		G	C	T	T
Sample	T	C	C	C	T	C	C	G	C	T	T

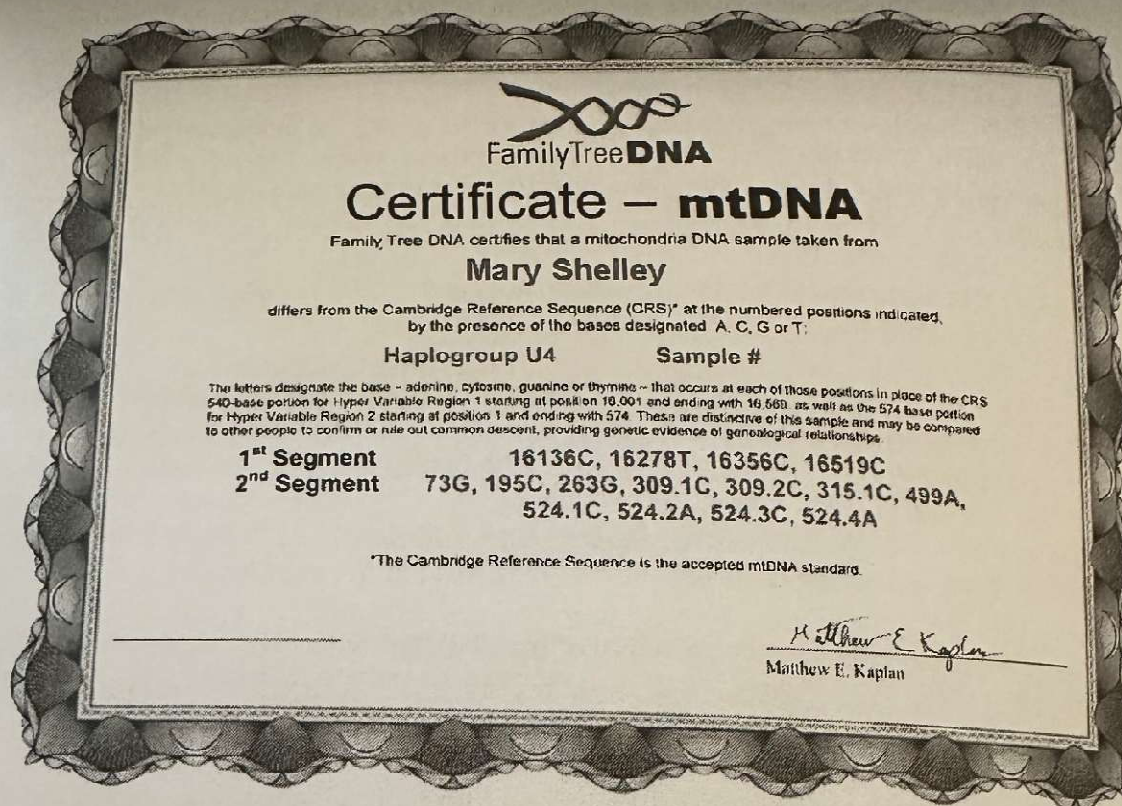
This mutation is written **315.1C** where .1C means that one copy of base C has been inserted at position 315 compared to the CRS. If two copies of base C had been inserted, this would be written as 315.2C.

A deletion is also possible. A common deletion is of base A at position 523. This would be written as **523-**.

Location	520	521	522	523	524	525	526	527	528	529
CRS	C	A	C	A	C	C	G	C	T	G
Sample	C	A	C		C	C	G	C	T	G

You can now read the mtDNA certificate below which is, in fact, a collection of mutations (compared to the CRS) and is known as the haplotype.

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The Haplogroup

A group of people who share a similar haplotype (a similar set of mutations) is called a haplogroup. These people are direct descendents of a single person (the common ancestor) in whom a specific mutation occurred many generations ago. This mutation has since been passed on to their descendents.

Haplogroup U4 is a sub-clan of the clan 'Ursula' (see The Seven Sisters of Eve by Bryan Sykes). The common ancestor of U4 is called 'Ulrike' by Bryan Sykes. This haplogroup had its origin in the Upper Paleolithic Age, about 25,000 years ago (the Upper Paleolithic Age or Late Stone Age extended from about 35,000 years BC to about 8,500 years BC).

The haplogroup is small but is widely distributed in Europe following the expansion of modern humans into Europe after the last Ice Age. Populations are found in Siberia (where up to 25% of the population belong to this haplogroup), the Volga-Ural region of western Russia and around the Baltic and Atlantic coasts in countries such as Estonia, Finland, Sweden and even France and Britain (1.6% of the population in England and Wales).

Mutations in mtDNA are very slow compared to those in Y-DNA and happen only about once in a thousand generations. They can be traced back over thousands of years but many women will share the same mtDNA.

Mutations in Y-DNA happen more often and are more varied and so are more suited to tracing family history which is usually traced back over hundreds of years. The same Y-DNA is usually only shared by very close (such as grandfather to father to son) male relatives.