MITOCHONDRIAL DNA – mtDNA

Via Direct Female Line

Haplogroup: K1b2b

Introduction: Mitochondria are small bacteria - like cell inclusions that function as little energy packets for the cell. Mitochondria have their own DNA, separate from the nuclear DNA where the chromosomes are located. Mitochondria are passed from females to their children, but it is only females who can pass this form of DNA on to the next generation. Therefore this DNA provides an indication of the deep maternal ancestry in both males and females.

As with the male Y chromosome, over the generations, and in each geographic location certain sites of "junk DNA" have mutated and these are the markers which are measured and serve to differentiate families and wider groups (e.g., Native American and African). Therefore you should have the same pattern of mitochondrial mutations as your ancestors say 2000 years ago. Mutations could happen at any time, but they would be rare within the period since surnames were adopted. Unfortunately it is often difficult to obtain a clear documentary record of female ancestors since their names (among Europeans) change with each marriage. The usefulness of mitochondrial DNA studies to genealogy is debatable - although it can serve to rule out candidates if their mitochondrial DNA does not match.

Specific Results:

HVR1 & 2 (Hypervariable Regions 1 & 2) Haplogroup K ("Clan Katrine according" to Dr. Sykes of Oxford University).

HVR1 Mutations: 16224C 16311C 16320T 16519C

HVR2 Mutations: 73G; 146C; 195C; 263G; 309.1C; 315.1C; 524.1C; 524.2A

A full genome sequence of the entire mtDNA has been completed by Family Tree DNA and confirms the specific **K1b2b** placement. In addition there are mutations that are specific to my closest relatives (even if many hundreds of years ago). These are:

309.1C, 315.1C, 522.1A, 522.3A, 522.4C, A9896G, and C16320Y. The latter is interesting in that some of my cells have this mutation, and some do not. This is known as heteroplasmy, a form of mosaicism. The genesis of this anomaly is unknown at this time.

This pattern is somwhat rare and there is as yet no clear geographical association.

The K haplotype appears to have originated about 16,000 years ago and may have taken part in the pre – Neolithic expansion into Europe from Western Asia or a glacial refugium in Europe.

As to K1b2b it is only by turning to ancient DNA research that we can get a sense of where and how long ago the specific K haplogroup emerged. In a database of thousands of ancient DNA mtDNA samples from across the world it is clear that K1b2b is extremely rare:

1) Two samples dated 2570 BC from a cemetery in Althausen, Germany.
2) A sample dated 900 AD from Gnezdova, Russia associated with the Vikings (likely from Sweden).
3) A sample dated 1225 AD from Nordby, Denmark.
4) Two samples, one dated 1350 AD from St. Mary Graces, and another dated 1445 AD from St. Mary Spital from England show that the haplogroup was in that country by Medieval times.

In terms of modern samples, the database of Family Tree DNA shows that the haplogroup was widespread in Europe, but at extremely low levels (typically less than .1%. Those with slightly higher numbers include France, then Sweden then Scotland – but none reaches beyond the base rate of .3%.

Further work needs to be done before we are able to conclude that my specific motif came from native Scots (e.g., Pictish or Scotti) or via Angles or the Norse Vikings.

In the case of the author, the lineage goes from Mary Brash to Elizabeth Muir to Mary Bain to Annie McCormack to Eva Fern Dawson to Violet May Williamson to David K. Faux.

The earliest known maternal ancestor is **Mary Brash** who at the time of her marriage in 1800 was residing in Kirkintilloch, Dunbarton, **Scotland**.

Joseph William Dawson and Annie McCormack with three daughters L to R Eva Fern, Hazel Mae, Pearl

Mary Bain and William McCormack

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