My Use of DNA in Genealogy

Mitochondrial SNP Tests

I was introduced to the subject of DNA testing by a book called **Out of Eden** by Stephen Oppenheimer. The subject of the book was what I now know to be a new 'science' called phylogeography. This combines phylogenetics with traditional archaeology to study the ancient migrations of peoples. It focused primarily on mitochondria, though the Y-chromosome was included. The startling conclusion of the book was that all non-Africans in the world are descended from a small group of humans that left Africa 80,000 years ago.

I then came across two books by Professor Bryan Sykes, **The Seven Daughters of Eve**, and **Adam's Curse**. These books have very readable discussions of the science, and lots of interesting anecdotes.

In summary mitochondria has been used to divide the world population into 36 haplotypes, 13 in Africa. Ninety-five percent of Europeans fall into 7 haplotypes which are discussed in detail in **The Seven Daughters of Eve**.

Perhaps the most startling assertion, though it is logical when you think about it, is that each mitochondrial clan must be descended from a single woman. The copying error (SNP) occurred just once, so everyone carrying the error must be descended from the first woman to carry the error. Not only that, but by linking the clans in a tree, one clan becomes the source of all the others. Not unnaturally the origin of this clan is called Eve. This doesn't mean that Eve was not one of a population of similar early humans, but that no descendants of her contemporaries exist today.

Professor Sykes had set up a testing company called **Oxford Ancestors** to allow people to get their DNA tested, so back in 2006 I got my first test test. It cost me £400 for a basic mitochondrial test.

Mitochondria is a small piece of DNA having approximately 16,000 bases. Of these 2 regions, called Hyper Variable Regions, or HVR1 and HVR2 are used to define haplogroups.

HVR1, containing about 400 bases, is sufficient to determine your haplogroup and this is what **Oxford Ancestors** tested. I had 3 Single Nucleotide Polymorphisms (SNPs) reported in this sequence. The one at position 126 is found in two haplogroups, J and T, but that at position 69 is specific to haplogroup J. The SNP at position 366 put me in the main group of Js, not in one of a number of sub-clans that had been defined.

J is a complex haplotype with several defined sub-groups. The other six main haplogroups found in Europe are all believed to have existed, or derived from groups that existed in Europe before the last Ice Age. Mitochondrial Clan J however originated in the Near East (possibly the Caucasus) and only moved into Europe after the last Ice Age when Neolithic farmers began to move into Europe 10,000 years ago. It followed 2 tracks, one of which followed the coast around the Mediterranean, and eventually up to Britain, whilst the other moved through central Europe.

The presence of haplogroup J in Europe, but in relatively small numbers (10% of the population) alongside the descendants of the earlier hunter-gatherer population answers an old historical argument. Did the gradual adoption of farming across Europe represent the migration of an idea, or the migration of farming peoples who replaced the indigenous population? The answer lies between the two. There was a migration of people, but they did not replace the existing population and the idea of farming spread into the indigenous

population. My SNP at position 366 was not found in the Near East so appears to have originated in Europe.

In 2013 I took the **Geno 2.0** test. This is used by **The Genographic Project** run by **National Geographic**. My main objective was the Y-chromosome SNP test which I will discuss later, but it includes both a mitochondrial SNP test and an autosomal test (all for $200).

The **Geno 2.0** test covered both HRV1, and HRV2 an additional 576 bases. My result classified me as J1c2, defined by a SNP at location 188, which occurred about 9,800 years ago in Europe. Today it is found in North and West Europe, reaching 10% of the population of Scotland. King Richard III, whose body was identified from a mitochondrial match with descendants of his maternal line, was also a J1c2.

One feature of the **Geno 2.0** test is you can transfer your result, and your sample to **Family Tree DNA** the company that hosts my **Warburton DNA Project**. In fact I believe the same laboratory does testing for **Family Tree DNA** and **The Genographic Project**. **Family Tree DNA** host a large number of special interest projects, including surname projects and haplogroup projects, including the **J-mtDNA** project. To fully exploit **Family Tree DNA**'s mtDNA facilities I would need to purchase a further upgrade.

Y-Chromosome Tests

Y-chromosome tests include Short Tandem Repeat (STR) and SNP tests. I began my own experience of STR testing in 2006 with a 10 marker test from **Oxford Ancestors.** This was sufficient to predict that my Y-chromosome haplogroup is R1b. It also showed that I share the Atlantic Modal Haplotype (AMH) values for 6 of the markers. The AMH is the most frequently occurring haplotype amongst human males in Atlantic Europe with its highest frequencies in the Iberian Peninsula, and in Great Britain and Ireland.

However the test wasn't much use for anything else. After more research I realised I needed more markers, and people to compare with. I also began to investigate my own family tree and then conceived the idea of doing a Warburton One-Name Study. As this seemed to fit so neatly with exploring Warburton DNA, the **Warburton DNA Project** became a part of the one-name study.

The Y-chromosome is much larger than mitochondria and so offers much more detail. I selected **DNA Heritage** as the company for the **Warburton DNA Project** tests, and began by testing myself, using their 43 marker STR test.

STR results come with an estimate of your SNP based haplogroup, but they cannot identify your more recent SNPs.

My result classified me as R1b3*. This indicated that I have SNPs at markers M207 (which defines me as haplogroup R), M343, and M269. M269 is the most recent SNP that an STR test can predict with confidence. It occurred about 10,000 years ago either in Europe or Western Asia. The classification of haplogroups has changed over the years and now simply incorporates your most recent known SNP. R1b3* is now classified as R-M269.

In 2013 the Y-chromosome part of the **Geno 2.0** test added further SNPs, the most important being at U106 and Z306. In fact I was defined as R-Z306.

One of my reasons for taking the **National Genographic Project Geno 2.0** test was that I had discovered STR matches with a group of Duttons and one of this group (Mike Dutton) had taken the **Geno 2.0** test. The Y-chromosome element of this test checks over 12,000 known SNPs and Mike and I have identical mutations or positives. The most recent mutation we shared was on marker Z306 so we were officially classed as haplogroup R1b-Z306. Z306 lies below U106 which is important as there is an **Rb1-U106 Project** at **Family Tree DNA,** so we are now both members.

About 25% of R1b in Europe have the U106 SNP, and it is most common in the Netherlands and Northern Germany. It is currently dated around 3100 BC.

U106 is subdivided by additional SNPs. Only Z306 was reported in my **Geno 2.0** results, but this means I also have Z381, Z156, and probably Z307, and Z305. Z306 occurred about 2700 BC, probably in Northern Germany.

Mike Dutton and I then tested for a more recent SNP called DF98, and were positive. DF98 is identified with **The King's Cluster**, which is maintained by Iain McDonald of the **Rb1-U106 Project.** His impressive phylogenetic tree and distribution map can be seen in **The King's Cluster Tree,** and **The King's Cluster Geography** on the **Warburton** website**.**

In the meantime **The King's Cluster** has been given more definition by using the results of 67 or 111 marker STR tests. DYF359S1 equal to 16-16 (it's a double marker) and DYS557 equal to 15 are the key STR identifiers for the **King's Cluster** and these are both covered by a 67 marker test.

In all 23 43-marker STR tests were conducted at **DNA Heritage.** One milestone was my first match to a genetic cousin, Clive. This lead to an interesting search for our common ancestor, which is described in **The Cheshire Group** on the Warburton website.

DNA Heritage ceased trading in 2011 and I switched to the **Family Tree DNA** 37 marker test, transferring the **DNA Heritage** results. The project has lived here ever since.I am now one of 11 matching results within the Cheshire Group.

In 2013 I expanded my STR result to 111 markers, and took the Big Y next generation sequencing (NGS) test. In the past year the Big Y-500 test has become available, incorporating 500 STR markers (including the original 111). Older BigY results have been upgraded to this level. My haplogroup is now defined as R-FGC17094, a SNP which occurred sometime between 1300 AD and 1600 AD. I also have two further SNPs which are unique to me.

There are now3 Cheshire Group Big Y results, and two Dutton ones. These show the Warburtons and Duttons share about 30 SNPs, including FGC13446. The last of this sequence is dated between 550 AD and 1550 AD, with the most likely date around 1150 AD

This fits neatly with the historical record that in the latter part of the 13th century Sir Piers de Dutton built a manor house at Werberton and began to style himself de Werberton. It would seem that Sir Piers is the ancestor of the Cheshire Group.

Autosomal Tests

My 2013 **Geno 2.0** test included an autosomal test. I was determined to be 41% Northern European, 40% Mediterranean, and 18% Southwest Asian. This most closely resembled the German reference population which has 46% North European, 36% Mediterranean, and 17% Southwest Asian. The British reference population has an even higher North European content with 50%, and just 33% Mediterranean, and 17% Southwest Asian.

I was also reported to be 2.2% Neanderthal and 2.0% Denisovan. This is because early modern human populations met the descendants of earlier humanoid migrations from Africa and interbred. A small amount of genetic material from these earlier species is found in most non-African populations, suggesting it conferred advantages that resulted in the descendants of this interbreeding coming to dominate modern populations.

In 2015 I took the **Family Tree DNA** Family Finder test. This reported me to be 89% British Isles, 4% West and Central European, and the rest trace elements including Finland, North Africa, Asia Minor and the Middle East. I can only assume that the difference between this

and the **Geno 2.0** result is a question of how deep into the past the analysis is looking. The Family Finder result also included an Ancient Origins breakdown which reported 44% Hunter gatherer, 44% Farmer and 12% Metal Age Invader. The descriptions of the Metal Age Invaders ties closely with my R-M269 Y-chromosome story, whilst the Farmers represent the neolithic farmer migrations represented by my J haplogroup mitochondria. The Hunter gatherers were an earlier migration into Europe from the Near East. They all passed through Europe to reach the British Isles. I guess my 89% British Isles from Family Finder simply means most of my line have been in the British Isles for some time.

Family Finder also show a large number (2500+) of potential matches, including 54 2nd-4th cousins and about 250 3rd-5th cousins. I have done little to verify any of these cousins, though I have loaded my details onto the gedmatch.com cross supplier matching site. However as the AncestryDNA database has 10 million results whilst both Family Tree DNA and Gedmatch have little more than a million each, I should probably take the Ancestry test if I want to do some serious cousin searching.

Reading List

1. Stephen Oppenheimer, *Out of Eden: The Peopling of the World* (Robinson Publishing, 29 Jul 2004)

2. Professor Bryan Sykes, *Seven Daughters of Eve* (Corgi, 1 Sep 2004)

3. Professor Bryan Sykes, *Adam's Curse: A Future Without Men* (Corgi, 1 Sep 2004)

4. Chris Pomery and Steve Jones, *DNA and Family History: How Genetic Testing Can Advance Your Genealogical Research* (PRO Publications. Sep 2004)

5. Professor Bryan Sykes, *Blood of the Isles* (Corgi, 3 Sep 2007)

6. Stephen Oppenheimer, *The Origins of the British: A Genetic Detective Story* (Robinson Publishing, 12 Apr 2007)