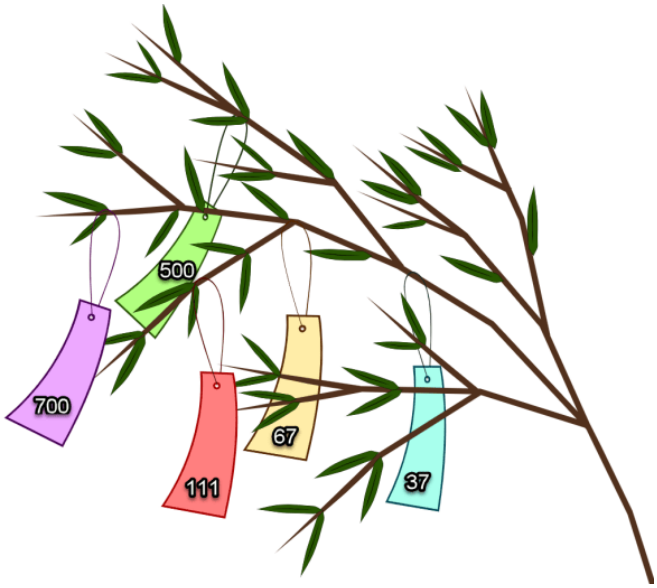
UPDATE – DNA FOR AGS

By Martin Brady, AGS Member

The Power of Big Y-700

The Y chromosome (Y chr) is passed down relatively unchanged from father to son, generation after generation. It is therefore ideal for researching surnames and patrilineal heritage. The Big Y-700 (only offered at FamilyTreeDNA.com) tests over 700 Short Tandem Repeats (STRs) and well over 100,000 new Single Nucleotide Polymorphisms (SNPs). STRs define the haplotype, SNPs define the haplogroup. SNPs are more stable than STRs. The FTDNA website states "The Big Y test is intended for expert users with an interest in advancing science…it is not a test for matching you to one or more men with the same surname in the way that our other Y STR tests do."



According to the FTDNA White Paper on the Big Y-700 (available on the FTDNA blog site) "customers can expect to receive 40% more STRs and 50% more high quality SNPs than they did with Big Y (previously offered by FTDNA)." Most autosomal tests use DNA microarrays to target known SNPs. The Big Y-700 uses 2nd/next generation sequencing (NGS, aka massively parallel sequencing) to discover new, previously unknown SNPs. SNPs that begin with the prefix "FT" were discovered by the Big Y-700 test. I recently noticed that in August and September hundreds of new SNPs had been added to the SNP index (ISOGG website) from the Big Y-700 test. As more people test, the haplotree will become better defined. This should more clearly define timelines for migration patterns from YAdam to present. Currently, there are 47,747 branches recorded in the GeneticHomeland.com database.

With NGS the DNA is first fragmented into small segments (the segments in my Big Y-700 results were ~150 base pairs). Each fragment is then sequenced at the same time (in parallel). As many as 1 billion fragments can be sequenced in parallel in a single run (thereby massively parallel). The segment sequence data must be reassembled into a chromosome using computers.

Big Y-700 results: Private variants are SNPs that haven't been entered on the haplotree. Named variants have been entered on the haplotree. A private variant stays private until a second person carries it. The variant then gets entered on the haplotree. I recently noticed that my terminal SNP

(my named variant that is the farthest out on the tree branch - downstream) had been changed from R-Z16284 to FT14437. Researchers at FTDNA had previously assigned R-Z16284 and FT14437 as equivalent SNPs (i.e., they defined the same branch on the haplotree) and recently noticed (as more people have tested) that not everyone who carries Z16284 also carries FT14437, but everyone who carries FT14437 also carries R-Z16284. So, these two SNPs were separated into two different nodes on the haplotree with FT14437 becoming a subgroup of R-Z16284. SNPs occur roughly every 80 to 140 years. So, this may move my terminal SNP branchpoint from about 800 BC to 720 to 660 BC. It is my hope that as more people test, my 12 private variants will find a place on the haplotree and I will be able to trace my patrilineal heritage to a more recent time.

Big Y-700 only tests about 40% of the Y chr. Some-day the highly repetitive region of the Y chr that is inaccessible to NGS may be accessible with the use of 3rd generation sequencing techniques. But more on that at a later time.