

Genetic Testing and Genealogy

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There are basically three types of DNA tests offered that have applicability to genealogy:

- Y-DNA testing, identifying the haplogroup and subclades of the direct male line from son to father to the earliest human beings;
- mtDNA testing, identifying the haplogroup and subclades of the direct female line from daughter to mother back to the earliest human beings; and
- Autosomal DNA testing, identifying strings of DNA among the other 22 chromosomes (i.e., leaving out the Y chromosome) that one has inherited from one's parents, grandparents, great grandparents, etc.

Y-DNA testing

Y-DNA testing for males will tell him what haplogroup and subclade he is in. It won't work for females, since they don't have a y chromosome.

Y-DNA testing will eventually tell males what family they are members of. By determining the order in which mutations in the SNP's of their y-dna occurred (called sequencing), the mutation pattern will identify first the haplogroup (the deep ancestry) and then its subclades (branches) down to the present day and, presumably, the route their ancestors took to get here today.

For example, I have been tested in the J haplogroup and down the string of sequenced mutations to a mutation at position J-L70. The first male having the J-identifying mutation occurred in Mesopotamia about 10,000 years ago, and all of his descendants, including me, carry that mutation. The J-L70 mutation occurred about 3,000 years ago somewhere on the coast of Turkey or Lebanon, and some of us haplogroup J's, including me, carry that mutation as well. Eventually, they will find the terminal mutation that only we Leonards have, and we will be able to trace where the mutations occurred leading up to that final one.

Y-DNA testing also tells males who else is in their family and/or how distant someone with similar test results is from them. This is measured through a different kind of mutation, the y-Single Tandem Repeat (y-str). These mutations occur in the hundreds of years, rather than thousands.

For example, almost all descendants of James and Henry Leonard test in the same y-str pattern of mutations at 37 or 67 markers. Test results within 3 mutations at 37 markers or 4 mutations at 67 markers indicates to a 99+% likelihood that we are related within 16 to 20 generations. These mutations indicate genetic distance: 3 at 37 or 4 at 67. The closest relative to me who has been tested is a genetic distance of 1 from me. He and I share a direct male great-great-great grandfather. A distant cousin over in England with whom I share a great grandfather 15 or 16 generations back is a genetic distance of 4 from me.

mtDNA testing

Mitochondrial DNA testing works for both men and women much like y-dna for men. Men have mtDNA from their mothers, but do not pass on to their children. For both men and women, mtDNA identifies the haplogroup their mother's mother's mother's...back hundreds of generations belongs to. Because mutations occur less often in mtDNA (think: 50 generations on an average), there is less identification of clades and subclades within the haplogroup.

For example, my mtDNA puts me in haplogroup T1a1, which I share with a lot of people in Scandinavia and England and going back in Russia (Tsar Alexander of Russia, a Romanov, was supposedly a T1a1). I have something over a thousand people who share my T1a1 and mutation pattern on FTDNA, but none that I can identify as relatives. 50 generations is a long time. The furthest back any of my genealogical research has taken me is 40 generations to a local duke on the coast of Norway in 740 AD.

Autosomal DNA testing

Autosomal dna testing covers the 22 autosomal chromosomes, all but the y chromosome, shared by both men and women. It looks for patterns of mutations over given segments of dna and matches those patterns against all the other people in their data base. As mentioned, one receives half of one's dna from one parent and half from the other, and so on back generation after generation. But also, which half one receives is left to chance: it

might include more from one great grandparent and less from another. And, obviously, the contribution of each great grandparent as one goes back in generations is likely to be less. Beyond ten generations back, the segments one receives are usually too short to identify the contributor.

For example, I've had my autosomal dna tested by 23andme, FTDNA, AncestryDNA, and the National Geographic Project. We'll skip the result that says I'm 1.8% Neanderthal (that's less than most people). According to all, I have over 1,000 cousins (different with each testing company, since their base of comparison people is different). The closest relatives who've been tested are second cousins (i.e., we share a common set of great grandparents). I share a little over 2% of my autosomal dna with my second cousins, about 10 segments that are long enough for identification of forebears. One more distant cousin who responded lives over in Sweden (my grandmother emigrated from Sweden), but we couldn't identify a common ancestor. In another case, a Swede who was very knowledgeable about his ancestry did identify a common ancestor 7 generations back.

Summary

DNA testing can be helpful in family research, but will not always yield the results you expect. Learning your haplogroup can eliminate possibilities. For example, Leonards occur in 10 or more different haplogroups, and you can focus on the one that's yours. Finding someone who shares a segment of DNA can sometimes help identify that common branch in your family trees.

But sometimes the DNA results may produce an unanticipated result. For example, the Y-DNA results may not verify the haplogroup your genealogical research indicated you should be in to match what you thought were your ancestors. This result may come from an adoption back in your family tree (parental deaths and orphaned children were more common in earlier generations) or a "nonmarital event" (an affair outside of marriage) somewhere along the chain of descent.

If you know you were adopted and want to find your birth parents or if you know there was a "nonmarital event" somewhere along the chain of descent, DNA testing can be of some limited assistance in helping to identify and then verify the birth parent. But one should anticipate a lot of additional research, not an easy answer.

DNA testing for genealogical purposes is in its early stages. It will become more helpful as more people are tested and the results compiled.