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'SATIABLE CURIOSITY: STACKING THE DECK: MUTATION RATE IN THE MTDNA CODING REGION

Author(s): Ann Turner

'SATIABLE CURIOSITY

Stacking the Deck: Mutation Rate in the mtDNA Coding Region

'Satiable Curiosity **is a column dedicated to the proposition that genetic genealogists are an untapped resource for resolving questions about DNA behavior -- how DNA changes over the course of a few or many generations and how DNA patterns are distributed around the world. Some questions are so broad that it could take decades to arrive at a conclusion, yet others are narrow enough to answer in a shorter time frame, perhaps even within a semester or two for a student research project. The results may nonetheless be of considerable genealogical utility and scientific interest, worthy of publication in a technical journal.**

The scientific method emphasizes the importance of obtaining random, representative samples before drawing conclusions about results. However, pilot studies may look for the best opportunity to observe a phenomenon. Genetic genealogists, who are obtaining full-sequence mtDNA tests in increasing numbers, are in a position to provide a "biased sample" for the study of the mutation rate in the coding region.

The more orthodox approach would be to study a very large number of mother/child pairs and divide the number of mutations by the number of transmission events, but mutations are rare, and the cost would be prohibitive with current technology. Adding a genealogical component, testing descendants of a common ancestor, augments the efficiency of the effort: two people can represent a larger number of transmission events.

So far, the method does not violate the random selection requirement. But what if we stacked the deck by picking some people who may have had a recent mutation, then testing their matrilineal relatives? This may seem like an impossible quest – how can one detect a recent mutation by looking at *one* person's sequence? "Recent" is a relative term – we can't tell exactly when a mutation happened – but we *can* tell which mutations in a person's sequence arose after the clan mother of a haplogroup or cluster lived. For instance, Behar (2006) has diagrammed the tree structure of Haplogroup K down to a very derived level. **Figure 1** shows the K1c2 section with a sequence (GenBank accession DQ830736) from a genetic genealogist grafted on to it.[1] It shows that four more mutations have occurred after the mutation defining K1c2. Perhaps a distant cousin of DQ830736 would lack one of those, demonstrating that a mutation occurred in DQ830736's line after their Most Recent Common Ancestor (MRCA) lived.

As of this writing, 47 genetic genealogists have submitted their sequences to GenBank.[2] Many of those records have "private" mutations, which can easily be identified by positioning them on a master phylogenetic tree at MITOMAP.[3] A call for volunteers who have obtained their full sequence and who can locate a distant matrilineal cousin would doubtless bring a larger sample onboard.

The results of this pilot study would be of great interest to genetic genealogists, who wonder how useful a full sequence test might be in identifying branch points in descendancy charts. They would also be helpful to the scientific community, since the results would place an upper bound on the mutation rate and provide guidance

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[1] GenBank http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?db=nucleotide&val=110351027

[2] Ron Scott keeps a running tally on his website http://freepages.genealogy.rootsweb.com/~ncscotts/

[3] http://www.mitomap.org/mitomap-phylogeny.pdf

about the size of a sample necessary to conduct a randomized study.

Suggestions for future columns are welcome. E-mail: DNACousins (at) aol.com.

Ann Turner

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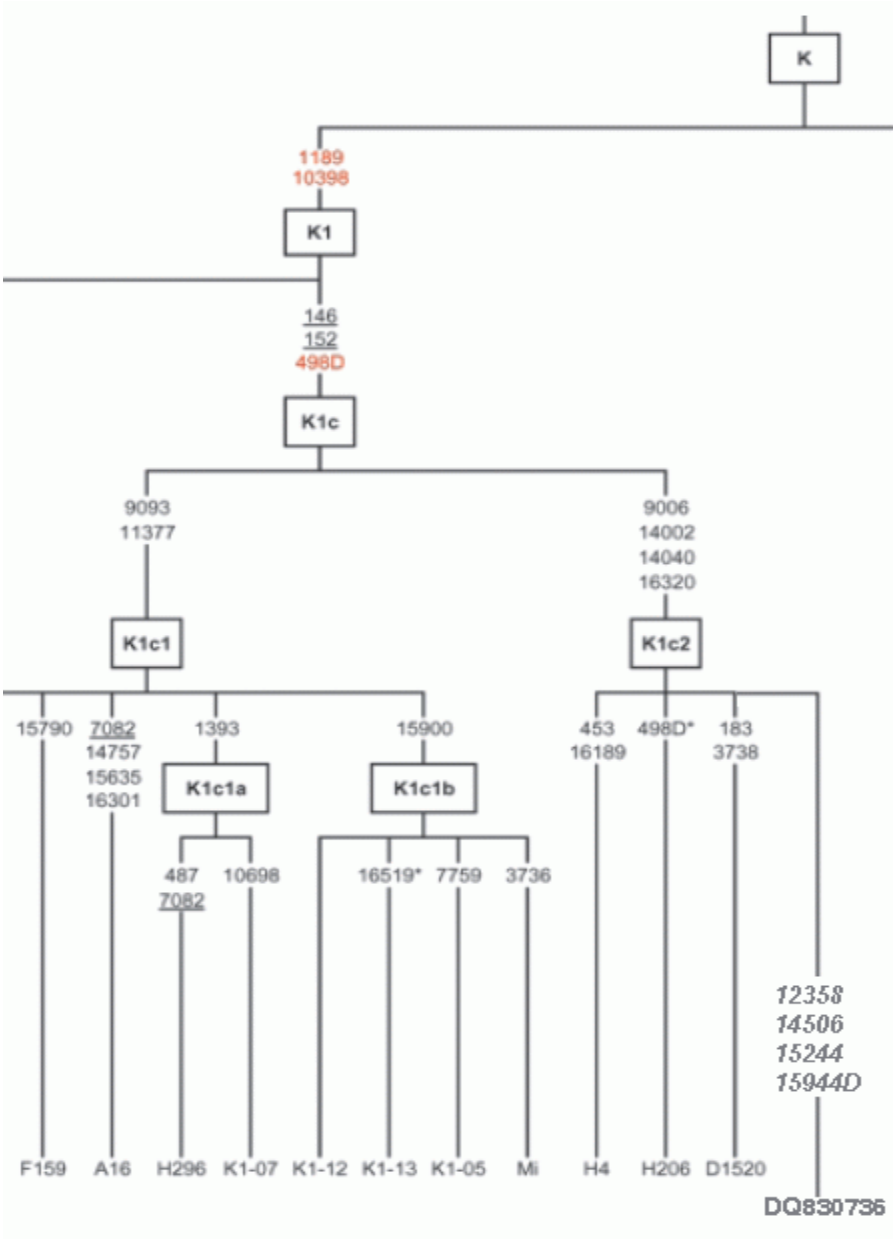

Figure 1 Part of the Phylogenetic Tree for Haplogroup K (Behar 2006)