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EDITOR'S CORNER: A FUNNY THING HAPPENED ON THE WAY TO RETIREMENT

Author(s): T. Whit Athey

Editor's Corner

A Funny Thing Happened on the Way to Retirement

In this space in the last issue of JoGG, I announced my retirement as editor. But, here I am, still on the job. My successor-to-be resigned in late December, so I have agreed to stay on for a while longer. Please continue to submit any articles for publication to me.

I am pleased to announce that David Wilson has agreed to join the JoGG Board of Directors and also accept a position as Associate Editor. David brings to the board experience in yet another area that has occasional application in studies of human history—linguistics. David is also the administrator of the large Wilson project. I am sure that many participants in the GENEALOGY-DNA List have seen his thoughtful postings.

The field of genetic genealogy has been relatively quiet for the past six months, at least compared to the lively pace of announcements from the testing labs in similar previous periods. We do have one new company offering mtDNA testing services, Argus Biosciences, and we welcome their entry into our marketplace. However, there has been no blockbuster announcement from the major companies of the field. Perhaps it is an appropriate time for consolidating past advances—getting the existing "trains" to run on time.

Before any company introduces tests for any new Y-STR markers, I certainly hope that steps will be taken to avoid the nomenclature issues that came up with the offering of the new 30-marker panel by FTDNA. FTDNA had six markers in their new panel that were already available from Ethnoancestry, but they apparently made no effort to understand why there were differences in their values from those of Ethnoancestry.

To their credit, the staff at the University of Arizona, who developed the tests for FTDNA, had sequenced the PCR products for all of the new markers they planned to offer. This allowed them to see the underlying repeat structure on each marker and to understand the options for scoring them. However, instead of investigating any further their differences from Ethnoancestry, which had previously stated that their nomenclature was based on the article by Kayser et al (2004) where the discovery of the markers was announced, they simply chose to insist that they had done it right and there was no need to look further.

Now I have no doubt that the University of Arizona group is perfectly capable of accurately sequencing the PCR products and counting the number of repeats that they find. However, there are guidelines available, developed by the International Society for Forensic Genetics (ISFG), with the participation of the U.S. National Institute for Standards and Technology (NIST), that explains how to score a Y-STR marker when the repeat structure is complex.

Nomenclature differences are a large problem for our field, and this one could have easily been avoided. FTDNA and Ethnoancestry, to their credit, have pledged to work toward eliminating such differences. FTDNA has announced that it has submitted its data on several markers to NIST for an opinion, and also pledged to abide by whatever opinion may be forthcoming. However, NIST has been slow to respond, and the issue is still pending.

A company may sometimes decide upon a reasonable approach to scoring a new marker, only to have later standards or publications appear or modifications/ updates of existing standards/publications that would affect the scoring.

This has occurred recently where a second publication (Lim, 2007) has appeared on the nomenclature of the markers in the Kayser (2004) article, including a recalibration of the Ethnoancestry markers in question. Since Ethnoancestry had originally based their reporting scheme on the first article, and since there had been no response forthcoming from NIST, they recently adjusted the results for several markers on their own, which seems appropriate.

Has this move eliminated the nomenclature differences? Unfortunately, the situation now is only a little better.

Originally, FTDNA and EA differed on four markers, DYS481, DYS490, DYS531, and DYF406. EA has now made adjustments to DYS490 (add 1), DYS531 (subtract 1), and DYS406 (add 7), resulting in an agreement with FTDNA on these three markers. EA has also adjusted its results on DYS481 upward by three units, but this still leaves FTDNA's values one unit higher.

Originally, EA and FTDNA agreed on DYS594, but one of the changes that EA has made, again, based upon Lim (2007), reduces the repeat count by one unit on this marker. Now, EA and FTDNA disagree on the reporting of this marker.

Therefore, it appears that FTDNA and EA now disagree on two markers, DYS481 and DYS594, instead of four, so I guess we have to consider this to represent progress.

While FTDNA and EA now agree on DYS531, it would appear that Lim (2007) may have failed to follow the ISFG guidelines on this marker. This marker has the following repeat structure:

$$(AAAT)_nA(AAAT)_1$$

Both FTDNA and EA are apparently not counting that last AAAT because of the intervening A. However, the ISFG guidelines address the issue of intervening bases, and they recommend counting *both* parts as one marker if the intervening sequence is not longer than the basic repeat motif (Gusmão, 2006). But, since we at least have agreement between the two companies on this marker, it is probably best to leave well enough alone.

There is no commercial lab at the present time besides FTDNA that tests for DYS450, but the way that FTDNA reports this marker appears to differ from that used in NIST's own internal studies by one unit. It will be interesting to see how NIST responds to FTDNA concerning this marker, assuming that FTDNA even asked about it and that NIST will eventually respond.

Everyone seems to agree that a difference in nomenclature for Y-STR markers is a bad thing. Why, then, do we have such differences? It appears to me that it should be a fairly easy procedure just to follow the ISFG guidelines. They are not very complicated, even as they apply to complex markers. While most professionals in this field are quite capable of counting to 12 (or whatever) with a good degree of accuracy, it also seems not too much to ask that they consult the guidelines to see just what they should be counting.

Another principle that seems well worth following, and which is also suggested in the ISFG guidelines, is that considerable weight should be given to whatever approach was taken by the first lab to score a new marker. If all of our genetic genealogy companies followed this bit of guidance, it would appear that differences in nomenclature would rarely arise.

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