Editor's Corner

Our Cup Runneth Over

Genetic Genealogists of today are like kids who just discovered a candy store. There is so much now available to us from the genetics testing companies. Just a year ago many of us were bemoaning the fact that Y-SNP tests were only available for the major markers and those at just one or two companies. We could then get 37 Y-STR markers from Family Tree DNA (FTDNA) or a somewhat different set of 43 from DNA Heritage (DNAH), Relative Genetics (RG), or (if you were willing to wait long enough) the Sorenson Molecular Genetics Foundation (SMGF). Or you could order both of the overlapping sets of markers and end up with a grand total of 48. We thought that we were in pretty good shape with respect to Y-STR markers, but the SNP situation was not very satisfactory.

Just in the last year, however, we have seen the two new companies DNA Fingerprint (DNAFP) and Ethnoancestry (EA) enter the genetic genealogy market with significant new test offerings. And, the established companies have brought forth their own new offerings. Now it is difficult to keep up with what is available. There seem to be new tests developed monthly for both SNPs and Y-STRs.

DNA Fingerprint (DNAFP), a spinoff of an existing medical and forensics company called Biotix, entered the fray early in the year. DNAFP is located in Germany and offers mtDNA, Y-STR, X-STR, and autosomal tests, but is perhaps best known for its active development of tests for new Y-STR markers not previously available commercially. During the last year tests for the following markers were developed at DNAFP: DYS413, DYS434, DYS643, DYS726, DYF371, DYF385S1, DYF399S1, and DYF401 (=DYS527), a total of 17 new markers if you count all the alleles in the multi-copy markers. DNAFP has also developed new versions of tests for two older markers, DYS385 and DYS464. The two alleles of DYS385 can now be distinguished, so that you can know whether, for example, a previously reported 11-14 is really 11-14 or 14-11. The new test for DYS464 shows which of the four alleles for R1b folks has the base "C" or a "G" associated with it in an adjoining sequence (in other haplogroups, all alleles are G-type). We also hear that more markers are in development, including DYS725.

Ethnoancestry came on the scene in mid-year with an extensive offering of SNP tests. EA has all of the major SNPs, but also offers subclade testing for Haplogroups G, I, J, and R1b. EA even says that if there's a SNP you want that isn't offered, talk to them and they'll discuss developing a test for it. EA uses a technique wherein the entire PCR product is sequenced, so that false negative results are much less likely. One of the principals in EA is Jim Wilson, a well-known population geneticist. Dr. Wilson is now developing commercial tests for many of the SNPs that he has discovered over the last several years. EA also has announced a panel of 18 STR markers, none of which are currently available commercially.

DNA Heritage also joined the fray in the SNP testing field at mid-year by offering multiplex SNP testing. In a multiplex series, tests are run on about a dozen SNPs at once. The first panel tests the major "backbone" markers, then a second panel is selected from among several possibilities, based on the results of the first panel, to refine the haplogroup. These multiplex tests typically test down to a medium level—not out to the most derived branches of the tree, but one or two levels past the major markers.

Near year's end, FTDNA announced the offering of several new subclade SNP test panels, including tests for Haplogroups E3b, G, I, J, Q, and R.

At this time last year a few folks in our community, by combining tests from different companies, could boast of knowing their results on 48 or 49 Y-STR markers. With the addition of the new markers from DNAFP, there are now a few of us with values for about 66 markers. When the first batch of results from EA's new Y-STR panel becomes available, possibly in January, a few of us test junkies

will have values on an amazing total of 84 markers.

FTDNA and DNAFP now offer sequencing of the entire mtDNA genome, all 16569 bases (give or take a few in individual cases). Having all of the information that exists on your mtDNA can usually show exactly where you fit into the mtDNA phylogenetic tree, but it can also, in rare cases, tell you that you may potentially suffer from a metabolic disease at some point in your life, either now or later. The health implications of whole-genome mtDNA testing are discussed in the review article by Ian Logan in this issue. You should be very sure that you really want to see your whole mtDNA sequence before ordering it. I now have my own complete sequence, but I am one of those people who want to know just what's going to happen, for better or worse. My complete sequence (done by FTDNA) showed 21 coding region differences from CRS, 16 of which "come with the territory" of being in Haplogroup U5a1a. Only five mutations appear to be unique to my particular lineage, and all of these appear to be quite benign. One of these five mutations, Ian Logan pointed out to me, is not exactly unique to me since it is shared by the wallaroo, which was very comforting.

FTDNA now offers a mtDNA Haplogroup H subclade test, which can determine which of 14 subgroups of H that a particular sample is in. FTDNA has stated that it plans to bring out mtDNA subclade tests for other haplogroups in the coming year.

With all of the new tests at our disposal, we amateurs can finally answer our own questions. Several members of our community are actively engaged in their own small, but quite important scientific studies. I am sensing that a watershed of sorts has been reached by our community, after which we will increasingly define our own agendas. We won't just have to wait impatiently for the next promised study to be published, but we may now be pushing the envelope ourselves in many areas of population genetics. Our cup truly runneth over.

Whit Athey