GSSCC - - DNA SIG - - handout
Guide to DNA Testing and Genetic Genealogy book (second edition)
Part Two
by Blaine Bettinger

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compiled by Gail Burk

Part Three of Blaine Bettinger's *Guide* covers analysis and application of DNA test results. Chapter 8 in this section reviews third party tools for autosomal DNA analysis. Of course, this second edition of the book was published in 2019, so these tools have undergone revision, and new tools have since been developed. Third party tools are independent of the DNA testing companies and they offer additional capabilities. Most importantly, third party tools allow you to compare raw data from the DNA company where you tested to raw data of your DNA matches who tested at other companies. Now, though, an easy way to make such comparisons is to upload your raw data from Ancestry or 23 and Me to the other DNA sites that accept data upload (Family Tree DNA; MyHeritage; and Living DNA).

One widely-used third party site is DNA Painter, where you can do chromosome mapping, which can help determine which segments of your DNA come from which ancestor. Chromosome mapping can help you to confirm or reject hypotheses about relationships with genetic cousins. Another popular feature at DNA Painter is the "What Are the Odds" ("WATO") tool, which just this year has been revised to WATO+. WATO can help to calculate probable relationships among DNA cousins. Blaine Bettinger's Shared Centimorgan tool is also found at DNA Painter. This helpful tool and the accompanying histograms enable you to narrow down the range of possible relationships by examining the quantity of shared DNA with your match. The current version of the Shared Centimorgan tool also allows you to compare two matches simultaneously. DNA Painter offers limited free versions of its tools and more robust applications by subscription. The Shared Centimorgan tool is entirely free.

GEDmatch is a popular third party tool where users can compare test results with shared matches who have tested at other companies. At GEDmatch, you can review one-to-many matches and compare one-to-one matches. A GEDmatch subscription enables users to access a clustering tool, multiple kit analysis, and matching segment searches. A subscription also aids triangulation by comparing your top 500 one-to-many matches with each other, which can be a huge advantage in working on brick wall research. Because GEDmatch uses e-mail addresses as its user communication protocol, you may be able to find contact information for a shared match of interest from another site. GEDmatch also has a feature that indicates start and end locations of shared DNA segments by table or graphic illustration. The chromosome browser at GEDmatch can show the precise

location of shared DNA segments, which in turn can help link a segment to a specific ancestor. The free version of GEDmatch offers the basic tools; additional applications are available by subscription.

Since publication of the second edition of Blaine's *Guide*, additional third party tools and sites have been developed or become more widely utilized, such as DNA GEDcom.com, Borland Genetics, and the new BanyanDNA.

Part Three of the book also has a chapter about ethnicity estimates. Bettinger explains in detail how the different DNA companies calculate their ethnicity estimates using reference populations and various processes and algorithms to parse each test. The comparisons described among the different DNA companies on exactly how each company calculates ethnicity estimates is especially informative, as is the sidebar about AncestryDNA's genetic communities. This chapter also touches on genealogical application of ethnicity estimates and their obvious limitations.

In Chapter 10, Bettinger discusses some ways we can explore complex genealogical issues using DNA analysis. Such analyses can help us to confirm or reject hypotheses based on our document research. The perpetual monkeywrench in DNA analysis (as well as in document research) is the ever-present possibility of misattributed parentage (NPE). Early pages in the chapter review the potential usefulness and limitations of using Y-DNA and mt-DNA analyses to help break down genealogical brick walls. The chapter touches briefly on the utility of joining a Y-DNA surname project at Family Tree DNA. If a Y-DNA tester does not match anyone in his assumed surname group, that does not always mean that he is not a member of that surname group, though. In the case of common surnames, it may only mean that no one else in that particular branch has tested yet. Thus, it is best, if possible, to test at least two descendants in the surname line of interest.

The bulk of Chapter 10 focuses on how to use autosomal DNA matches to research brick wall ancestors. The review of which DNA cousins to test in order to maximize potential analysis is quite helpful, because in order to obtain the most information about an ancestral couple, you need to test multiple descendants of that couple. This chapter emphasizes how important clustering shared autosomal matches is as an analytical tool. This method, also called shared match triangulation, helps to identify a cluster of shared matches whose trees (if available) can be reviewed for clues. This method requires us, of course, to do the genealogical research necessary to verify that our match's tree is correct, and to build out the tree and connect our shared matches to a shared ancestral network. As more people test and link trees, shared match clustering becomes an ever-more useful tool for researching genealogical brick walls. Thus, it's important to revisit the brick wall issue periodically, because some new test-taker may hold the key that finally helps to resolve the question!

Chapter 10 also includes a discussion of segment triangulation. Triangulation is a time-consuming, advanced technique whereby segment analysis can determine which ancestor or ancestral couple passed down a specific segment of DNA, which can then aid in concretely identifying common ancestors shared with new genetic matches. Segment triangulation can be plotted on a spreadsheet or third party tool. Segment data can be downloaded from 23 and Me, Family Tree DNA, MyHeritage and GEDmatch. Ancestry DNA does not share segment data with test-takers. Bettinger explains how to obtain segment data from GEDmatch, and he shows an example of a segment data spreadsheet, including a list of categories to include in order to get the most out of the sorted data. This chapter also explains how to identify triangulation groups; that is, groups of three or more people who are known to share a particular segment of DNA in common. This section explains which tools to use at each testing company to find triangulation groups.

It is important to remember that neither shared match clustering nor segment triangulation is error-proof. If our trees have missing ancestral branches at the generation of connection, we are limited in how sure we can be about any shared match or shared segment conclusions. Smaller shared segments may be "identical by state;" that is, widely shared by the population, rather than identical by actual descent.

Chapter 11 in Part Three shows how DNA testing can help adoptees and genealogists with stubborn brick walls find these hidden ancestors. Bettinger recommends that researchers hoping to resolve adoption, NPE, and brick wall questions begin with an autosomal test. Y-DNA testing can sometimes, but not always, help males find their biological surnames. Y-DNA testers should use a 37-marker or higher Y-DNA test. Due to how slowly it mutates, mt-DNA is generally not as helpful as Y-DNA in solving brick-wall issues, although mt-DNA can conceivably help point a researcher in the direction of her or his maternal origins. Mitochondrial DNA matches should be sought among autosomal matches in order to find the most useful mitochondrial DNA cousins. In both cases, joining a Y-DNA or mt-DNA research project can be helpful. Combining Y-DNA and mt-DNA analysis with surname clues found among the earliest known paternal or maternal ancestors listed by Y-DNA and mt-DNA matches can go a long way in helping to solve these cases. Of course, it's always possible that the most recent common ancestor of a Y-DNA or mt-DNA match is so far back in time that it is beyond the reach of genealogical records.

The most powerful DNA tool in breaking down genealogical brick walls is autosomal DNA testing. Due to the composition of the accumulated data in the various testing companies' databases, the highest success rate is in the United States. In 2017, more than 80% of adoptees in the U.S. had a half-second cousin or closer on their shared match list. Working with matches that are second cousins or closer will almost always lead to successful conclusions. Working with more distant matches, such as fourth cousins, will

be much more difficult and time-consuming, and may not ultimately be successful. Shared match clustering and triangulation are very useful tools in working out brick wall questions.

Chapter 11 mentions, but in my opinion does not sufficiently emphasize, the importance of doing descendancy research on the trees of shared matches in the suspected brick wall ancestral lines. Recommended strategies in the *Guide* also detail the importance of finding a geographic nexus between the suspected ancestors in the mystery line and your own brick wall forebears. Researchers hoping to solve brick wall cases should have their DNA data at all the DNA sites, because some people only test at a single site.

Bettinger concludes by predicting some future developments in DNA testing and analysis, such as affordable whole-genome autosomal testing. He hypothesizes about the possibility of reconstructing the genomes of long-dead ancestors and even the potential for creating composite images of these far-back ancestors from the DNA signatures found among their descendants. The potential to create DNA-only trees of unnamed long-ago ancestors is also proposed, as is the use of epigenetic testing. Bettinger suggests that epigenetic data might be useful in differentiating close DNA matches into the correct relationships. He mentions fascinating studies that suggest that people who experience trauma as a child, or who had a parent or grandparent who experienced trauma, have different epigenetic profiles from people did not experience similar trauma. Another intriguing possibility for the future is artifact testing, which at the moment is expensive and still in its infancy. Bettinger also predicts that, in the future, lineage societies may be more accepting of genetic data as part of a submitter's application for membership.