RT1748 Developing a DNA Testing Plan

Paul Woodbury
Outreach Manager at Legacy Tree Genealogists • Course Developer at Salt Lake Community College
pwoodbury@legacytree.com • 801-590-0534

Introduction

Development of a strong testing plan requires development of a robust research question. Whereas some research questions can be answered with a single DNA test, many others will require additional testing and considerations. Therefore, once you have defined what you are trying to investigate, determine **who** you will test, **what** types of tests, a timeline or workflow for **when** you will perform testing, the companies **where** you will test, **why** you have chosen these options and **how** you will execute your plan.

Developing a DNA testing plan also relates back to the five-step research process as presented by Brigham Young University:

1. Identify What You Know About Your Family
2. Decide What You Want to Learn About Your Family
3. Select Records to Search
4. Obtain and Search the Record
5. Use the Information

In this context, DNA is the record; performing a DNA test just to find out more about your own ancestry is an excellent starting point. It is the first step of the process of identifying what you know about yourself and your family. It constitutes the initial discovery of your genetic genealogy. However, after you test, you should use the information gained to continually expand your research discoveries. If you find that your DNA test results were a "waste of money." The fault lies not with the test but with your failure to:

1. Thoroughly analyze your own results (identify what you know)
2. Develop a specific research question (decide what you want to learn)
3. Identify who you would like to test (select records to search)
4. Recruit additional individuals to test (obtain and search the record)
5. Use other's test results to expand your research (Use the information).

What you gain from DNA testing will likely depend on the effort you put into planning it. You could be lucky; you probably won't. You could break through your persistent thirty-years-in-the-working brick wall on your first DNA test, but that would be the exception rather than the rule. Most researchers who are successful with genetic genealogy investigation have had to plan and execute additional testing. If your initial test has not met your research needs, plan more testing.

WHO should I test?

Who you test will depend largely on the question, "*Who has the DNA that will help me with my research problem?*" As you answer this question, consider the different types of DNA and their unique inheritance patterns.

Y-DNA is inherited intact along the direct paternal line. Occasional mutations distinguish unique Y-chromosome lineages. If you are concerned about the possibility of recent mutations in your Y-chromosome line, consider testing older relatives. Once you have tested one relative, their Y-chromosome will be representative of their known male relatives who carry the same surname. If you struggle to find a descendant to test, consider searching Y-DNA surname projects at Family Tree DNA to determine if one of the direct line descendants of your ancestor may have already tested. Y-DNA signatures may vary from the expected if there was a "non-paternal event" like an illegitimacy or undocumented adoption. If you suspect a situation like this in your own family, test another direct paternal relative from another line to confirm or refute this hypothesis. If you are attempting to determine the paternity of an ancestor and there is at least one paternal candidate, test a direct paternal descendant or relative of that candidate.

Mitochondrial DNA likewise is inherited intact from generation to generation, but along the direct maternal line. Occasional mutations distinguish different mitochondrial DNA lineages. Therefore, one individual can represent a large number of relatives. Mitochondrial DNA mutates at a much slower rate. Though recent mutations are certainly possible, they are rarer than recent Y-DNA mutations, though it still might be a good idea to test older relatives for reasons outlined below.

Autosomal DNA has a very different inheritance pattern which is more random and subject to chance. Each individual inherits 50% of their DNA from each parent, the percentage of DNA inherited from previous generations can only be approximated due to recombination which randomly shuffles autosomal DNA every generation. As a result, each individual shares about 25% of their DNA with their grandparents and approximately half the previous amount for each preceding generation. When using autosomal DNA tests consider the following:

- Each individual will inherit some DNA from each of their fourth great grandparents. Prior to this, they may have some genealogical ancestors who did not contribute genetic material to their genome.
- Autosomal DNA tends to recombine less through paternal lines than through maternal lines.
- Collateral descendants of a common ancestor will inherit different portions of their ancestors DNA; though they may share DNA with the common ancestor they may not share DNA with each other.
- Relatives closer than the level of second cousins should share detectable amounts of DNA with each other. Third cousin and more distant relationships are sometimes undetectable through DNA testing.
- The DNA inherited from a specific ancestor passed through all ancestors in between, so if a child's parent has been tested, there is little reason to test the child unless the other parent is also a descendant of the ancestor of interest.
- Tests for sibling sets, first cousins and second cousins can be helpful in reconstructing the genomes of deceased ancestors since the segments of DNA they share in common and the segments of DNA they do NOT share in common can all be used to draw conclusions.
- The likelihood of identifying additional relatives by testing known relatives varies based on their relationship and can be assessed in the AncestryDNA help menus.[1]

[1] AncestryDNA, *Should other family members get tested?* AncestryDNA Help menus, table 2, dna.ancestry.com, accessed January 2016.

- Since the amount of autosomal DNA shared with an ancestor decreases by approximately 50% each generation, prioritization of autosomal DNA testing should be given to the closest generational descendant and not necessarily the oldest living descendant.
- Search for closest generational descendants among the youngest children of the youngest children of the ancestor. These individuals will have the longest generation times.

Though there are no specific DNA tests for the X chromosome, it does have a unique inheritance pattern that can justify a different research plan. When passed through males, the X-chromosome does not recombine; when passed through females it may recombine. Therefore it is equally likely that a female individual will inherit some X-DNA from her father's mother's father's mother's father's mother (4th great grandparent) as it is that they will inherit some X-DNA from their mother's mother's mother (great grandparent). Searching for descendants who follow the father-mother descent line can increase the chances of matching an unknown distant relative on the X-chromosome.

Finally, when deciding who to test, it may be wise to maximize your time and test older relatives.

WHAT type of test should I use?

As with all elements of your plan, what test you use will depend upon the nature of your research question. When possible, use as many types of tests as possible that are pertinent to the research question. If you are exploring the ethnicity of an ancestor, both Y-DNA and mtDNA haplogroups can be specific to geographic localities. If the ancestor is a fairly recent relative, you may be able to answer ethnicity questions with an autosomal DNA test as well.

For specific research questions regarding the identity of an ancestors parents, testing choices might depend on the closeness of the relationship. If the research subject is the parent, grandparent, or great grandparent of the individual, prioritize testing the individual and her immediate family members. If the ancestor is a more distant relative, prioritize testing direct line paternal or maternal relatives. If no such relatives are available, able or willing to test, resort to autosomal testing along several unique descent lines. When possible use, a combination of test types to anchor and filter your DNA test results.

All autosomal DNA tests are essentially equivalent to each other and none of the companies offers different levels of autosomal DNA testing. However, with direct line tests there are several options. With direct line tests, begin with lower marker levels and work higher. If you have matches at the lower marker levels who have already tested at higher levels, upgrade your test. If you have promising matches at equal or lower marker levels who have not upgraded, offer to help them purchase higher levels of testing. If you do not have any matches, wait. If you have particularly promising matches who have not used other test types, invite them to perform an autosomal test.

WHEN should I test?

DNA test planning is a cyclical trial and error process. Unlike the majority of genealogical records which, if they were available today, were likely available yesterday and will likely be available tomorrow, DNA test results are constantly changing, thus requiring a delicate balance between waiting for more results and moving forward with additional testing and analysis. The timeline of your DNA testing plan will likely be progressive in nature: waiting to perform additional testing until after the results of the first test have arrived. In some cases where a research question is time

sensitive (i.e. I need to find my biological family before they die or before I die). Waiting between test results may not be feasible. Deciding the timeline of a project will also require prioritization of who you will test first based on who is still living.

WHERE should I test?

The big question! Where should I test? The answer: it depends. Each company has its advantages and shortcomings.

There are some easy answers to this if you have specific questions. If you require Y-DNA or mtDNA analysis, use Family Tree DNA. If you are interested mostly in ethnicity, use 23andMe. The answers to other questions depend upon the research company.

23andMe: Large database (1 million+) • Best ethnicity results • Excellent ancestry tools • Includes Y-DNA and mtDNA haplogroups • Worldwide customer database • Strong privacy • Medical information • *Complicated and cumbersome invitation, sharing and messaging system • Low response rate • Outlawed in France, Germany, and Maryland • Low level of genealogical experience • most expensive option ($199 per kit)*

AncestryDNA: Large database (1 million +) • Fastest growing database • DNA Circles, New Ancestor Discoveries, Matches in Common and Shared cM values• Expanding to UK, Ireland, Canada, Australia, and NZ • Many matches have attached trees • Moderate to high level of genealogical experience • *Until recently, almost exclusively American • No chromosome browser • Few organizational and analysis tools • Additional $49 cost for full access*

Family Tree DNA: Also offers Y-DNA and mtDNA tests • Sample storage for additional testing (25 years) • Cheek swab rather than spit tube • Accepts autosomal transfers from AncestryDNA and 23andMe (prior to Nov 2013) • Advanced analysis and interpretation • Direct contact with matches • Higher response rate • Strong representation of Jewish samples • Group projects • *Smallest atDNA database • Difficult to navigate • Lowest amount of privacy • Stringent matching parameters*

WHY should I test?

As you craft your plan ensure that you are making wise decisions by writing out your reasoning. The Genealogical Proof Standard offers compelling evidence for why DNA testing should be at least considered as part of a reasonably exhaustive search, but it also suggests that conclusions should be coherently written. As part of your research, your testing plan reasoning should be coherently communicated as well. This will assist you in communicating and convincing relatives to test

HOW will I execute my plan?

There are many other considerations to take into account as you carry out your plan. Convincing relatives to test may require salesmanship, creativity and compromise. In many cases you will need to pay the expenses or arrange a trade-off. Plan how you will sell your research efforts. Regularly update recruited relatives regarding progress. If they decline participation, respect their choices. Budgeting for large number of tests can be a considerable financial burden, consider recruiting interested family members who are willing to help, but who do not have the DNA you need. Search for heritage and family associations who might be willing to host your project. Spread the word, recruit and report your findings and enthusiasm will spread once others see your results. Make your DNA tests a success by crafting a personalized testing plan today!