By Debbie Parker Wayne, cg, cgL DNA BASICS



This column is a series on using DNA for genealogical research. There are several types of DNA tests offered for genealogical purposes. Researchers must understand that only like tests can be compared: Y-DNA to Y-DNA, mitochondrial DNA (mtDNA) to mtDNA, autosomal DNA (atDNA) to atDNA, and X-DNA to X-DNA. To use DNA to solve a problem, an understanding of DNA inheritance and the limits of the evidence is paramount. This article covers X-DNA and builds on the atDNA article in the last issue. Specialized X-DNA short tandem repeat (STR) tests are not covered in this article.

arlier articles have shown how to research our patrilineal line using Y-DNA, which is passed only from a father to his sons; our matrilineal line using mtDNA, which is passed only from a mother to all of her children, with only daughters passing it to the next generation; and all of our recent ancestral lines using atDNA inherited from all of the ancestors on our pedigree chart. X-DNA follows a unique inheritance pattern that is different in males and females and includes only some of our ancestral lines.

#### What can you DO with X-DNA?

For most genealogical problems, X-DNA alone is not useful. It is used in correlation with other DNA evidence to support a theory. For example, atDNA might support a theory that two people are descended from a common ancestor, while X-DNA provides evidence for the ancestral line that common ancestor is part of. X-DNA focuses research on the most likely ancestral lines on which you may be related to a person and excludes other lines as a possibility. Because of random recombination, the absence of an X-DNA match does not prove you are not related on a particular line, but the existence of an X-DNA match of significant size indicates you are related on an ancestral line through which X-DNA is inherited. Charts help determine which ancestral lines may have contributed to a person's X-DNA and are discussed later.

X-DNA is usually tested along with other chromosomes as part of an atDNA test. Until recently X-DNA analysis tools were only available as third-party tools and at 23andMe. Even with access to the X-DNA data, the lack

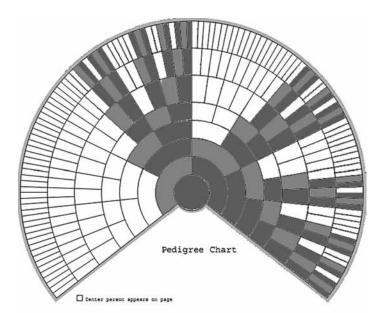


Figure 1. Female X-DNA inheritance chart.

of tools and the different inheritance pattern for X-DNA have caused many genealogists to ignore X-DNA data when it can narrow down the lines to be searched, allowing for efficient use of our research time.

### What is X-DNA and how is it inherited?

Each cell of our body usually has twentythree pairs of chromosomes in the nucleus. Chromosomes one through twenty-two are the autosomes. The twenty-third pair of chromosomes defines gender: an X-Y pair in males, a pair of Xs in females.<sup>1</sup>

A male child inherits a Y chromosome from his father and an X chromosome from his mother. The X is a recombination of the two X chromosomes the mother inherited from her parents. When a male tester has a match on X-DNA the entire paternal half of his family tree and portions of the maternal half can be excluded as a source of the X-DNA match.

A female child inherits an X chromosome from her father and an X chromosome from her mother. The X chromosome from her father is the one the father inherited from his mother; no recombination occurs before it is passed to the female child. The X chromosome from the child's mother is a recombination of the two X chromosomes the mother inherited from her

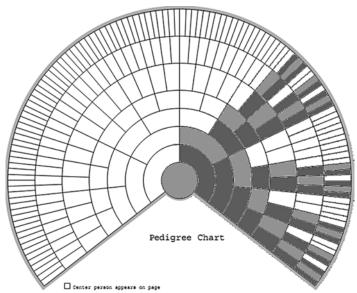


Figure 2. Male X-DNA inheritance chart.

parents. When a female tester has a match on X-DNA one half of the paternal half of her family tree and portions of the rest of the tree can be excluded as a source of the X-DNA match.

As with autosomal DNA, recombination of the X chromosome may divide and shorten the segments whenever recombination is part of the process. With autosomal DNA the larger matching segments usually indicate closer relationships. With X-DNA we are seeing unexpected recombination patterns; we still have much to discover about exactly how the X chromosomes recombine. Until more information is available, genetic genealogists must be careful how we interpret matching segment sizes when analyzing the X chromosome. See the endnote for blog posts with some detailed examples.<sup>2</sup>

The pattern of X inheritance is most clearly illustrated using charts. Blaine Bettinger of *The Genetic Genealogist* blog created colored fan chart images that can be used to determine which ancestors may have contributed to the X chromosome of a person. See figure 1 for a female X-DNA inheritance chart and figure 2 for a male X-DNA inheritance chart.<sup>3</sup> The fan chart images are beautiful when printed in color where pink represents female and blue represents male ancestors. Ancestral names can be hand-written on a printed chart or added with an image editor. With Bettinger's permission, the charts were recreated as a word processor document so ancestor names could be added and easily changed during research. These charts also indicate the average percentage of the X-DNA that could have been contributed by each ancestor. See figure 3.4 A Creative Commons license was included that allows others to add their own enhancements and make the charts available for all to use and improve. This shows the power of collaboration between members of the genealogy community as others quickly made further enhancements to the charts.<sup>5</sup> Several blog posts illustrate how some have used the X inheritance charts.<sup>6</sup> Some charts have ahnentafel numbers included and some include the percent of the X chromosome inherited from each ancestor.

#### X-DNA test results

The X chromosome is tested along with chromosomes 1 through 22 during an autosomal DNA test. The X-DNA test results consist of raw DNA data. Some companies include the X-DNA data in the same file as the autosomal raw data. Some companies, such as Family Tree DNA, put the X-DNA data in a separate file. When downloading raw DNA data to your own device, be sure you get all of the files if the data is in multiple files. Keep in mind company policies can change over time so always verify current practice to ensure that you have all of your data.

Raw X-DNA data look very much like the raw atDNA data except the chromosome is listed as X or 23 instead of 1 through 22. AncestryDNA uses 23 to designate the X chromosome; Family Tree DNA and 23andMe use X. Past and future policies may differ. See table 1 on the following page for sample raw data. As with atDNA, the raw data include a list of marker names, chromosome designation and locations on that chromosome, and the chemical found on each chromosome at that location. The chemicals are Adenine, Cytosine, Guanine, and Thymine, each usually represented by the first letter of the

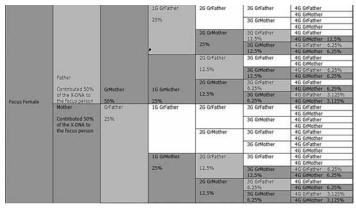


Figure 3. Word processor X-DNA inheritance chart.



Figure 4. Family Tree DNA match list showing X-Match.



*Figure 5. Family Tree DNA Chromosome Browser showing X-Match.* 

name: A, C, G, or T. Everything else we get from X-DNA data is based on analysis of the data and comparing it to other testers.

When a list of atDNA matches is viewed, an indication as to whether this person also matches on the X chromosome can be seen. At Family Tree DNA the down-arrow under a person's photo can be clicked to display the X match status on the match list page. See figure 4 showing the display after the down-arrow has been clicked. When viewing matches in the chromosome browser, the X chromosome is now displayed at Family Tree DNA. See figure 5. The display at 23andMe in the chromosome browser is very similar. AncestryDNA does not display this data in a chromosome browser format and does not supply matching segment data that lists start and stop points for matches on individual chromosomes.

In-depth analysis of X-DNA matches using the segment data (the chromosome number and start

Marker name	Chromosome	Position	Genotype
rs2840722	Х	116155511	AG
rs5909828	Х	116155870	СТ
rs2819637	Х	116189668	AA

Table 1. Raw Data: Raw X-DNA data is usually downloaded as CSV, XLS, or ZIP file.

and stop points of matching segments for each person) is done the same way as described in the atDNA article. The focus is on the ancestral lines identified in the X inheritance charts. Spreadsheets and third-party analysis tools help organize the segment data for analysis and comparison with others.

#### **Using X-DNA test results**

The basic steps for using X-DNA are similar to those for atDNA, but an X-DNA inheritance chart focuses the researcher on the ancestral lines that may have contributed to a person's X-DNA.

Complete all lines of your pedigree as far back as possible. Including collateral lines may help determine who a common ancestor may be. Document this to share with DNA matches looking for a common ancestor. List your ancestral names, dates, and geographic origins. The more information included, the easier it will be to determine when a person is common to two family trees.

Create a privatized pedigree chart. For example, list information on your earliest known ancestors down to a great-grandparent or a recent generation that is no longer living. Include geographic locations and dates for comparisons.

Create an X inheritance chart for each person who has taken an autosomal DNA test. When the person matches on both autosomal and X-DNA, search for a common ancestor on the ancestral lines identified on the X inheritance chart.

Review any ancestral information shared by your DNA matches and contact the person for more information. If a common ancestor cannot be identified by name, look for patterns that provide additional research clues, such as geographic locales, spouses' names, and so on. Matches may not have posted everything they know online. Some people don't respond to contacts, but an attempt should be made. Be patient; the person may respond months after an initial query.

#### Resources

This article is a short introduction to X-DNA. For information on tests offered by different companies see each vendor's website and the International Society of Genetic Genealogists (ISOGG) Wiki pages.<sup>7</sup>

This article on X-DNA completes the series of basic introductory DNA articles. Anyone who needs more study on the basics may find Kelly Wheaton's *Beginners Guide to Genetic Genealogy* useful.<sup>8</sup> Future articles will delve deeper into some DNA topics, illustrate some of the thirdparty tools as well as tools provided by the testing companies, and show how different types of DNA testing can be combined with documentary research.

## Notes

All URLs accessed 17 May 2014.

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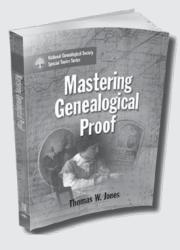
8. Kelly Wheaton, *Beginners Guide to Genetic Genealogy* (https://sites.google.com/site/ wheatonsurname/beginners-guide-to-genetic -genealogy).

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