What Is This DNA Stuff and Why Should I Care?

Part 6: X-Chromosome DNA (X-DNA) Testing

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In the article on Y-chromosome DNA (Part 4) we discussed the male determining Y-DNA allosome which is passed exclusively down the paternal line. In this article, we will discuss the other allosome, X-chromosome DNA (X-DNA), and its corresponding test.

As a reminder, the allosomes are the sex-determining pair of chromosomes (labeled pair 23 in the human genome) and they are found in the nucleus of the cells. Men have one X-chromosome copy from their mother and women have two copies, one from their mother and one from their father.
The X-chromosome is over 150,000,000 base pairs in length, which is nearly 3 times the size of the Y-chromosome, and it contains about 2000 genes. X-DNA testing is done using SNP analysis although the SNP density is lower than the autosomal DNA. All four of the major testing companies test for X-DNA but, only as part of their autosomal DNA test, i.e., you cannot order a separate X-DNA test because X-DNA displays a unique inheritance pattern that varies for men and women and makes interpretation difficult. As part of the atDNA tests, Ancestry tests 28,892 X-DNA SNPs (labeled chromosome 23), FTDNA tests 18,091 SNPs, MyHeritage tests 17,889 SNPs, and 23&Me tests 16,530 SNPs. FTDNA is the only company that will indicate an X-DNA match (X-match) in their cousin matching list, which means you share some X-DNA with that person, but there is no indication of how much X-DNA is shared. However, if you transfer your results to GedMatch, you can see X-DNA matching information from any of the testing companies, but only matches that share 10 cM or more should be considered X-DNA matches.

As mentioned above, the inheritance pattern for X-DNA is unique which makes it difficult to use for cousin matching or direct maternal or paternal line determination. Fathers always pass down their X-DNA without any recombination and they only pass it on to their daughters; no X-DNA is passed down from father to son because fathers only pass their Y-DNA down to their sons. On the other hand, mothers pass down their X-DNA to all their children (male and female), either with or without recombination.

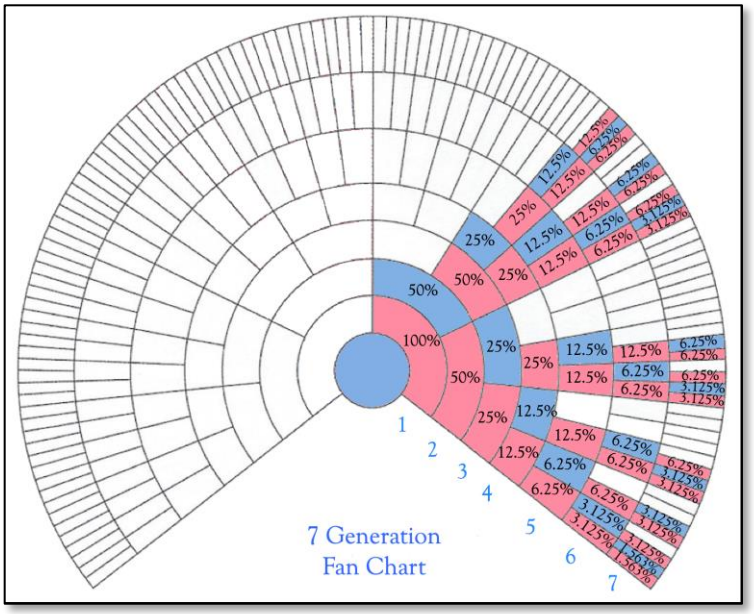

Figure 1. Male Inheritance Pattern for X-DNA.
Source: Generated by the author from similar fan charts in
"Guide to DNA Testing and Genetic Genealogy" by Blaine T. Bettinger, 2016

Figure 1 shows the unique inheritance pattern for a male in which the male and his male ancestors only receive their X-DNA from their mothers, but all of the female ancestors receive X-DNA from both of their parents. Due to this unique inheritance pattern, the fan chart shows that at 4 generations, only 5 of 16 great-great grandparents are represented in the males X-DNA, all from the maternal side, and at 7 generations, only 8 male and 13 female ancestors out of 128 total ggggg-grandparents can theoretically contribute to one's X-DNA. Note, the percentages shown in the fan chart are the theoretical amounts of X-DNA from each possible ancestor at a particular generation, however, the actual amounts will be based on the amount of recombination that occurs at each generation and some contributions will likely be lost.

On the other hand, females receive half their X-DNA from their mother and half from their father (one X-chromosome from the mother and one from the father). Figure 2 shows the unique inheritance pattern for a female in which she and her male ancestors received their X-DNA from their mothers, but all of the female ancestors received their X-DNA from both of their parents. At 4 generations only 8 of 16 great-great grandparents are represented and at 7 generations, only 13 male and 21 female ancestors out of 128 total ggggg-grandparents can contribute their X-DNA.

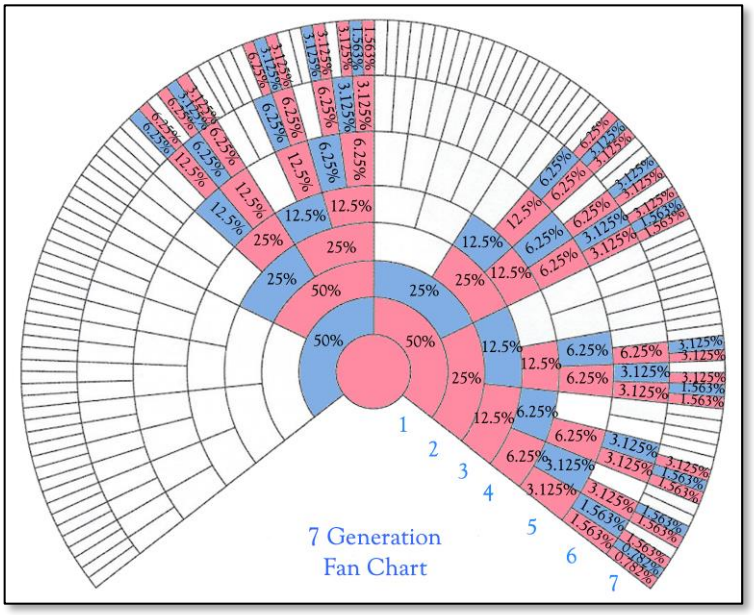

Figure 2. Female Inheritance Pattern for X-DNA.
Source: Generated by the author from similar fan charts in
"Guide to DNA Testing and Genetic Genealogy" by Blaine T. Bettinger, 2016

One of the reasons that X-DNA is not used as a stand-alone test is due to these distinctive and different inheritance patterns. Figure 3 gives an interesting example whereby an individual does not match the X-DNA of their sibling. To begin, paternal grandfather (David) has no daughters, so his X-DNA chromosome (blue) is not passed down to the next generation. Paternal grandmother (Justine) passes one of her X-chromosomes to her son (Benji) without any recombination (green). Note, it could have been the yellow-colored X-chromosome but for this example, the green X-DNA chromosome was passed down. Maternal grandfather (Nathan) has a single copy of the X-chromosome and he passes this down completely unchanged to Susan (red). Finally, maternal grandmother (Cara) recombines her two X-chromosomes and passes down a mixed chromosome copy to Susan (gray & pink).

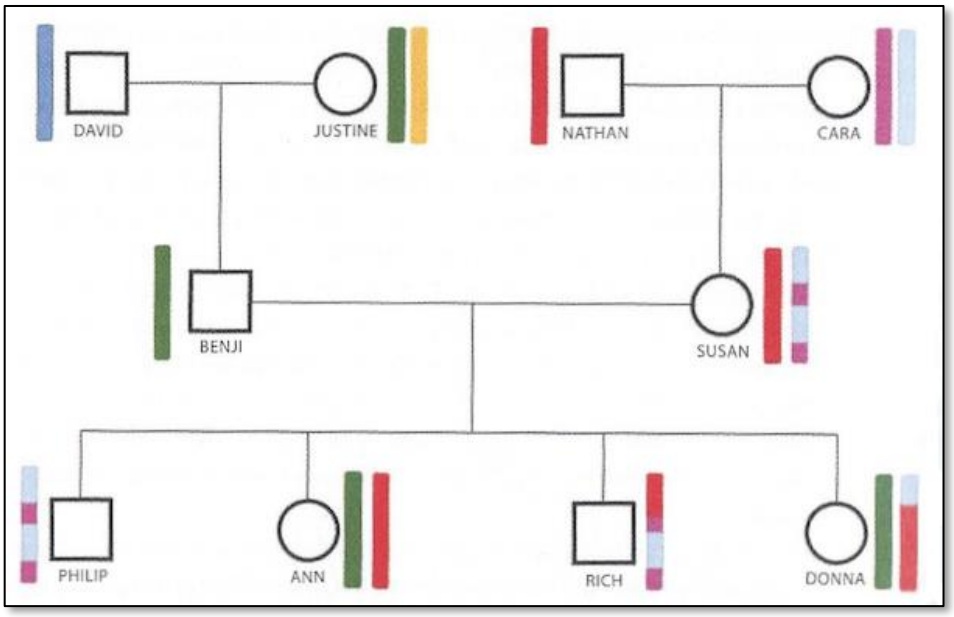

Figure 3. X-DNA Inheritance Example
Source: "Guide to DNA Testing and Genetic Genealogy" by Blaine T. Bettinger, 2016

Susan, therefore, has X-DNA from 3 of her 4 grandparents, while her husband Benji has a single X-chromosome from his mother. In the third generation, Ann and Donna receive an unchanged X-chromosome directly from their father Benji (green). Philip and Ann receive an X-chromosome from their mother without any recombination. Rich receives his X-chromosome with recombination from his mother Susan. Looking at the grandchildren, sisters Ann and Donna both share a full chromosome from their father (green) and their paternal grandmother (Justine). Ann also receives a full X-chromosome from her maternal grandfather (red), but Donna only receives a portion of her X-chromosome from her maternal grandfather (red). Philip shares some X-DNA with Rich and Donna (purple or gray) but not with Ann. It is not uncommon for siblings to share no X-DNA let alone be a perfect X-DNA match. Based on this example, it is clear that X-DNA testing is limited in its usefulness as a stand-alone DNA test used for comparing family members or looking for cousins.

In summary, the X chromosome is one of the two sex chromosomes. Men have one copy from their mother and women have two copies, one from their mother and one from their father. X-DNA testing is done using SNP analysis although the SNP density is lower than autosomal DNA testing and only segments that are 10 cM or more should be considered matches. The unique inheritance pattern for males and females are different, fathers always pass down their X-DNA without any recombination and only to their daughters, whereas mothers pass down their X-DNA to all of their children either with or without recombination. For this reason, the testing companies only test X-DNA as part of their autosomal DNA test.

This concludes our discussion of X-chromosome DNA and of the four main types of DNA tests that are available for genealogical investigations. The next article (Part 7) will compare the four major testing companies to help you decide which one(s) are best suited to help you with your family history pursuits whether you are interested in finding out your ethnicity origins, finding related family cousins, learning more about potential health issues or traits, or trying to find a lost relative. Until then, happy genealogical hunting!