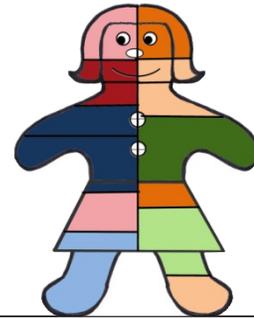


Chromosome Mapping: What It Is, Tools for Mapping, Why We Do It

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INTRODUCTION

Genetic genealogy is arguably the most exciting development in genealogy since original images of records became widely available online. Experienced genealogists look for the best ways to use our genetic data to extend our family trees and confirm research already done. Genealogists with scientific and mathematical skills devise new analysis techniques. Genealogists with programming skills automate those techniques and make them easier for the rest of us. This article introduces some of these tools, but is not a step-by-step guide for using any of the tools or techniques. Additional information on usage can be found in the sources cited in footnotes and in the referenced URLs.

WHAT IS CHROMOSOME MAPPING

Chromosome mapping assigns each segment of DNA to the test-taker's ancestor or an ancestral couple from whom that segment was inherited. Sometimes a segment can be assigned to a specific biogeographical or ethnic population. Most of the testing companies, except *AncestryDNA*,¹ provide tools called chromosome browsers that help us map segments of DNA shared with other test-takers in that database. *23andMe*² displays both the segments shared with other test-takers and a second browser shows which DNA segments have been assigned to a specific

1 All URLs were accessed 19 January 2018.

1 *AncestryDNA* (<https://www.ancestry.com/dna/>).

2 *23andMe* (<https://www.23andme.com/>).

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region or ethnicity. *Family Tree DNA*³ and *MyHeritage*⁴ provide browsers that display segments shared with other test-takers. *LivingDNA*⁵ plans to add test-taker matching and, presumably, a browser in the future.

All of these companies allow autosomal DNA raw data to be downloaded once a test is complete. Comma Separated Value (CSV) files can be downloaded from companies that supply a chromosome browser. That raw data file or the CSV files can then be uploaded to other third-party tools and sites—those not provided by a testing company—that offer segment analysis and chromosome mapping tools. *GEDmatch*⁶ is one of the most popular third-party tool sites, offering a chromosome browser for matching segments and several that will map a segment to a specific region or ethnicity. Some additional tools described below use the matching data from the testing companies and *GEDmatch* to create chromosome maps.

Triangulation is an analysis process that helps accurately map segments to a specific ancestral line.⁷ When autosomal DNA (chromosomes 1–22) first began to be used for genealogy the recommendation was to use segments of at least five centimorgans in length.⁸ Today a minimum length of ten centimorgans is more common. Smaller segments are more likely to be false positive matches or pseudo-segments. Our conclusion about which ancestor we inherited a specific DNA segment from are more credible with more cousins: (1) who share the same DNA segment; (2) who have a well-documented, accurate, and deep tree; and (2) who share no other ancestral lines. The triangulation information (naming a chromosome number and the start and end location of a shared DNA segment) is used to map a chromosome.

3 *Family Tree DNA* (<https://www.familytreedna.com/>).

4 *MyHeritage* (<https://www.myheritage.com/dna>).

5 *LivingDNA* (<https://www.livingdna.com/en-us>). As of this writing, *LivingDNA* does not yet offer a list of matches to other test-takers in their database. This company compares your DNA to determine which area of the British Isles your DNA most closely matches using the database of the People of the British Isles (POBI) Project (http://isogg.org/wiki/People_of_the_British_Isles).

6 *GEDmatch* (<https://www.gedmatch.com/>).

7 Debbie Parker Wayne, "Triangulating Autosomal DNA," *National Genealogical Society NGS Magazine* 42 (October-December 2016): 39-42; online at (http://debbiewayne.com/pubs/pub_NGSMag_201608_triangularion.pdf). See also Jim Bartlett, *segment-ology* (<https://segmentology.org/>).

8 "Centimorgan," *ISOGG Wiki* (<https://isogg.org/wiki/CentiMorgan>).

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TOOLS FOR MAPPING CHROMOSOMES

Many genealogists mapped chromosomes using spreadsheets when no other tools were available. Some still use spreadsheets. Images created from the numbers in spreadsheets help visual learners. Programmers who are also genealogists using DNA for research have provided most of our third-party tools. Newer tools are beginning to do more of the mechanical work for us, but the human brain is still one of the best tools we have available. Some of the tools to help with chromosome mapping are listed below.

Spreadsheets

Excel and other spreadsheet programs were the first applications used for genetic analysis by early genealogists. Jim Bartlett, Kitty Cooper, Roberta Estes, and Dr. Tim Janzen are genealogists who are very experienced in using spreadsheets for genetic analysis and who share their techniques online. If spreadsheets are your tool of choice, then check out Jim's *segment-ology* blog,⁹ Roberta's *DNAeXplained* blog,¹⁰ *Kitty Cooper's Blog*,¹¹ and Tim's publicly available technical papers and Powerpoint presentations from lectures.¹²

Kitty Cooper's Tools

Kitty Cooper offers tools to map one single chromosome¹³ and to display overlapping segments.¹⁴ These may help in the analysis phase of research. She also created one of the earliest image mapping tools for all chromosomes. A genealogist creates a CSV file as shown in figure 1 in a specific format specifying how a DNA segment is to be mapped: (1) the maternal or paternal side, (2) chromosome number, (3) start point for the

9 Jim Bartlett, *segment-ology* (<https://segmentology.org/>).

10 Roberta Estes, *DNAeXplained - Genetic Genealogy* (<https://dna-explained.com/>).

11 Kitty Cooper, *Kitty Cooper's Blog* (<http://blog.kittycooper.com/>).

12 See links to Tim Janzen's paper and presentations linked in many sections of the *ISOGG Wiki* including "Chromosome mapping," *ISOGG Wiki*

(https://isogg.org/wiki/Chromosome_mapping). Some can also be found by using Google to search for Tim's name in genetic genealogy mail list and forum archives.

13 Kitty Cooper, "One Chromosome Mapper - Make a graphic chromosome map from a CSV of overlapping segments," *Family History for Munson, Larkin, Thannhauser* (<http://kittymunson.com/dna/ChromosomeMapper.php>).

14 Kitty Cooper, "Overlapping Segment Mapper," *Kitty Cooper's Blog* (<http://blog.kittycooper.com/tools/my-graphing-or-mapping-tools/segment-mapper/>).

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shared segment, (4) end point for the shared segment, length of the shared segment in centiMorgans, and (5) Most Recent Common Ancestor (MRCA) from whom this DNA segment was inherited. Kitty's Chromosome Mapper webpage reads that file and creates a map.¹⁵ Figure 2 shows the output: a visual representation of the mapped segments with a different color used for each mapped ancestral *couple*. Moving the mouse (the black arrow) over a segment pops up a box with the ancestral couple name and the length, start point, and end point of the mapped segment.

side	Chro	Start	End	cMs	MRCA
m	1	105000000	157000000	28.1	5 - John Ira Ryan m. Mary Ann Otis
p	1	165000000	190000000	22.4	5 - Henry Parker Sr.
m	1	177000000	190000000	10.5	7 - Solomon Dearman
p	3	67000000	99000000	19.3	5 - Henry Parker Sr.
m	4	32000000	53000000	14.09	8 - John Parker m. Sarah Gordy

**CSV Input to
Kitty Cooper Chromosome Mapping Tool**

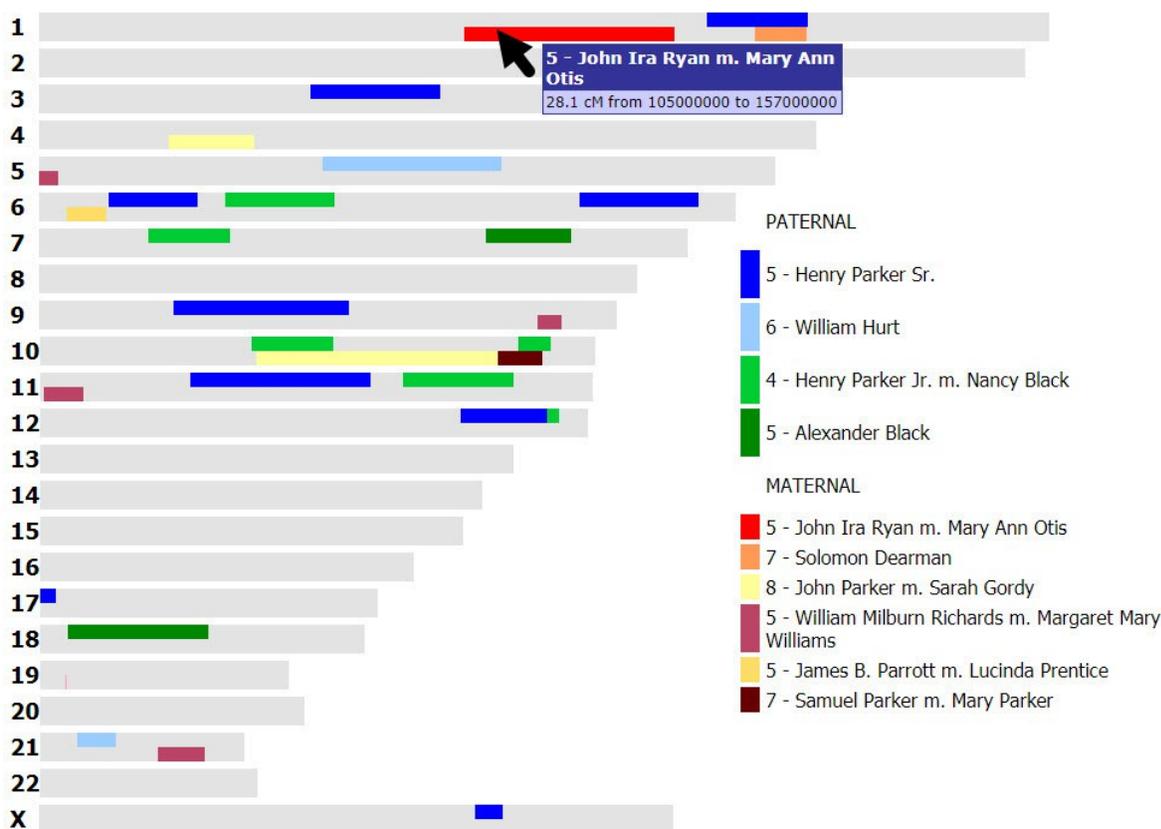
Figure 1. CSV Input to Kitty Cooper Chromosome Mapping Tool.

15 Kitty Cooper, "Chromosome Mapper - Make a graphic chromosome map from a CSV of your ancestral segments," *Family History for Munson, Larkin, Thannhauser* (<http://kittymunson.com/dna/ChromosomeMapper.php>).

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Chromosome Map - Jan 19, 2018 - 11:35 AM CST



Chromosome Mapping Tool by Kitty Cooper at <http://kittymunson.com/dna/ChromosomeMapper.php>

Figure 2. Map from Kitty Cooper's Chromosome Mapping Tool.

Genome Mate Pro [replaced in 2020 with *GDAT - Genealogical DNA Analysis Tool*]

Genome Mate Pro,¹⁶ developed by Rebecca Walker, is a full-blown analysis tool that includes a segment mapper. The display is similar to the Kitty Cooper mapping tool. Figure 3 shows the output: a visual representation of the mapped segments with a different color used for each mapped ancestral

16 Rebecca Walker, *Genome Mate Pro: A Tool for Managing DNA Comparisons* (<https://www.getgmp.com/>). This site includes links to download the program for Windows, Mac, and Linux systems; Quick Start Guides and a User's Guide; videos for getting started with the program; and other information.

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generation. Moving the mouse over a segment displays at the bottom of the window the chromosome number, start point and end point of the mapped segment, ancestral couple names, and a code assigned by the user to the couple.

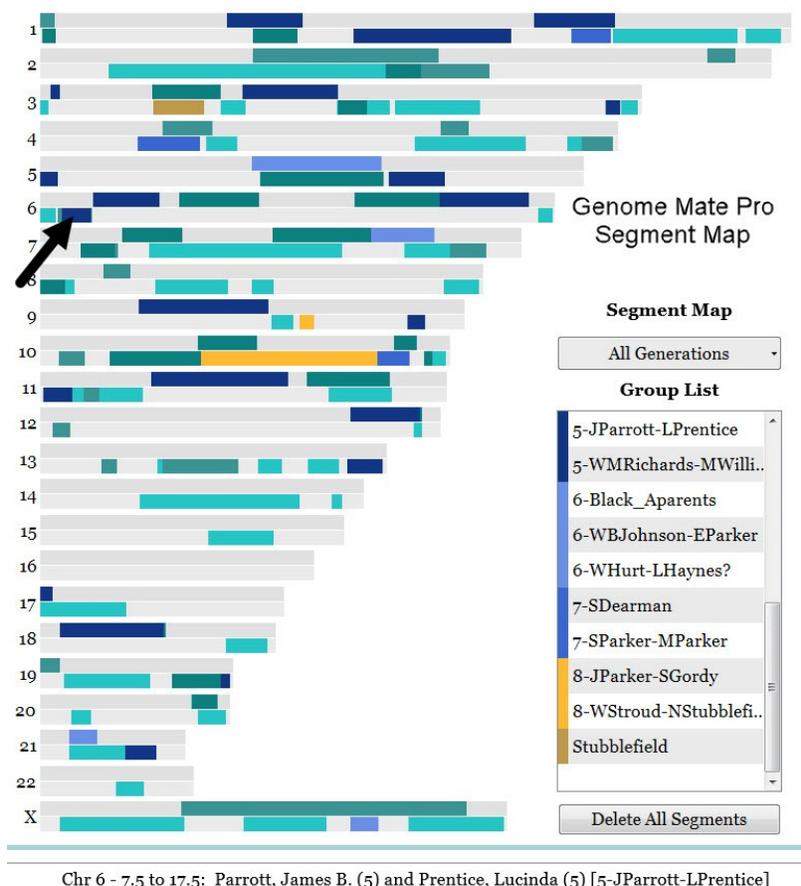


Figure 3. Genome Mate Pro Segment Map.

The learning curve is high for *Genome Mate Pro*, but it is a powerful tool allowing analysis of our genetic data on our own computer. *Genome Mate Pro* imports data from the major testing companies and major third-party tool sites. The researcher analyzes the matching DNA segments and ancestral lines of a person with whom a test-taker shares DNA. One downside for some researchers is that support for and information on the

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program is primarily provided through *Facebook*.¹⁷ A comprehensive user's guide provides help for those who prefer not to use *Facebook*.

DNA Painter

Jonny Perl makes several useful tools available on his website, one of which is a chromosome mapper, *DNA Painter*.¹⁸ With *DNA Painter* you manually enter matching segment data into a web page. The data can be copied and pasted directly from a testing company, *GEDmatch*, or from your saved CSV file. You can enter the data then print a chromosome map without saving any data on the website. If you want to save the map for future access you must register (at no cost), login, and save the data. Figure 4 illustrates the output which is a variation of that seen in Kitty Cooper's Chromosome Mapper and the *Genome Mate Pro* Segment Mapper.

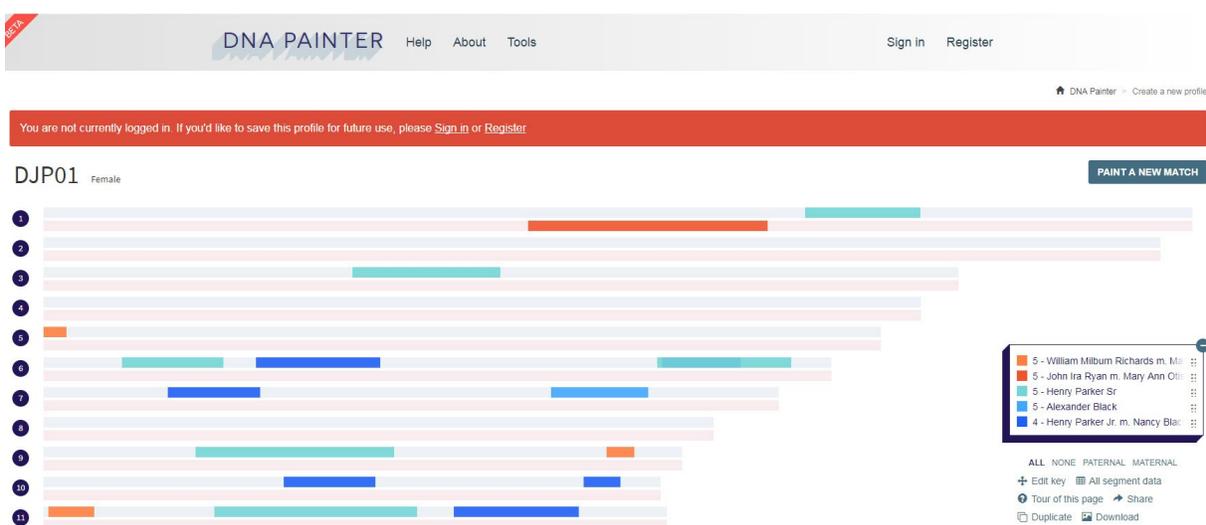


Figure 4. DNA Painter Segment Map.

17 "Genome Mate Pro," Public Group, *Facebook* (<https://www.facebook.com/groups/GenomeMatePro/>).

18 Jonny Perl, *DNA Painter* (<https://dnapainter.com/>) and *Facebook* (<https://www.facebook.com/dnapainter/>).

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Phasing

The simplest form of chromosome mapping is fairly easily done. Every person inherits one copy of each of the autosomes (chromosomes 1-22) from each parent, mitochondrial DNA from the mother, an X chromosome from the mother, and either an X or a Y chromosome from the father. Phased DNA data is sorted so that the DNA inherited from each of our parents is identified. True phasing is done when a child's DNA data is compared to both parents. The majority of the tested DNA locations can be phased with the child and one parent, but some locations will be undetermined.

Many genealogists are old enough that they no longer have living parents to test. Workarounds have been developed for this situation. DNA data can be pseudo-phased by comparing a test-taker's DNA with a population database reference (as is done by *AncestryDNA*). Visual phasing helps those of us without the proprietary tools of a large company to do a different kind of "phasing"—assigning portions of our DNA to the grandparents from whom that DNA was inherited.

Visual Phasing

Dr. Kathy Johnston pioneered visual phasing¹⁹—a method that allows phasing and assigning segments to our four grandparents when three or more siblings have been tested.²⁰ Dr. Blaine T. Bettinger²¹ and others teach this methodology.

In the beginning, this technique required a lot of manual work to create chromosome comparisons for the sibling pairs and paste them into a document, carefully lining up the images, for analysis. Researchers create

19 "Visual phasing," *ISOGG Wiki* (https://isogg.org/wiki/Visual_phasing).

20 Posted with permission of Kathy Johnston by Baron Lewis, "The Use of Crossover Lines Among Siblings to Determine Segment Matches with Grandparents—Visual Phasing," *DNA Genealogy* blog (<https://dnagenealogy.tumblr.com/post/137722603308/the-use-of-crossover-lines-among-siblings-to>).

21 Blaine T. Bettinger, PhD, JD, "Visual Phasing: Mapping DNA to Your Grandparents" *Virtual Institute of Genealogical Research* (<http://vigrgenealogy.com/store/bettinger-phasing/>).

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the chromosome comparisons using the chromosome browser on *GEDmatch* and paste the images into a Powerpoint, Word, or Excel document.

The chromosome images are analyzed to find the crossover points—the places where the chromosomes recombined before the parent passed them to the child. Those crossover points are then assigned to the child whose DNA holds that crossover. The result of this process is shown in the top of figure 5 where full sisters I, J, and D are compared on the X chromosome. Using the green Fully-Identical Regions (FIR) and yellow Half-Identical Regions (HIR) and matches to other test-takers who are related to only one of the four grandparents, the researcher can map the chromosomes of all three sisters to the grandparent from whom the X chromosome segment was inherited. The bottom of figure 5 shows the sisters all share the entire paternal X chromosome (blue) which their father inherited from his mother—the sisters’ paternal grandmother. The sisters have a different mix of the X chromosomes of their maternal grandparents—pink from the maternal grandmother and green from the maternal grandfather.

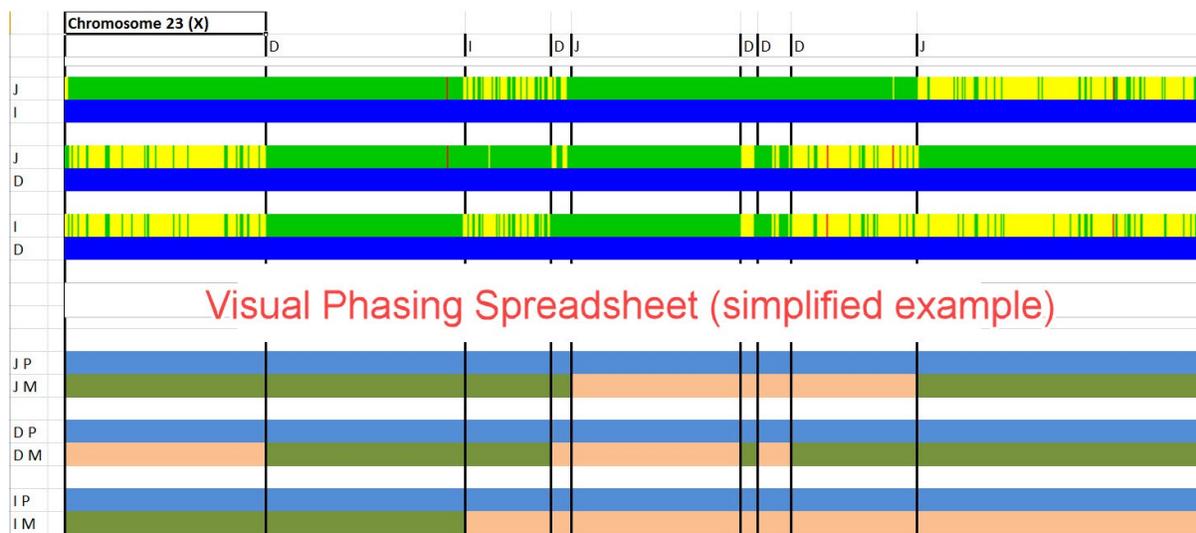


Figure 5. Visual Phasing Spreadsheet for X Chromosome.

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Steven Fox and David Love created a macro-enabled Excel spreadsheet that automates gathering the chromosome images and analysis.²² The tool and the methodology have matured to handle relationships other than three full siblings, but the best results are seen with three or more full siblings. You must join a *Facebook* group to download the tool.

GEDmatch Admixture Tools to Map Regions and Ethnicity

Whether or not the testing company maps DNA segments to a region or ethnicity, anyone can upload the raw DNA data to *GEDmatch*. There we can use any of several admixture tools to map DNA segments to a specific region or ethnicity as shown in figure 6. If those same segments of DNA have been mapped to a specific ancestral line we should search for the indicated ethnicity in that ancestral line.

It may also be necessary to apply knowledge of history to properly interpret the estimates. For example, the Asian predictions shown in figure 6 could be Asian if the test-taker has ancestors from Japan or Korea. If the test-taker only has European ancestry and the family has been in the Americas for hundreds of years, this might indicate Native American ancestry. At times Asian and Native American ancestry can be similar as the first Americans came over the Bering Strait from Asia to the Americas.

22 "The Visual Phasing Working Group," Closed Group, *Facebook* (<https://www.facebook.com/groups/visualphasing/>).

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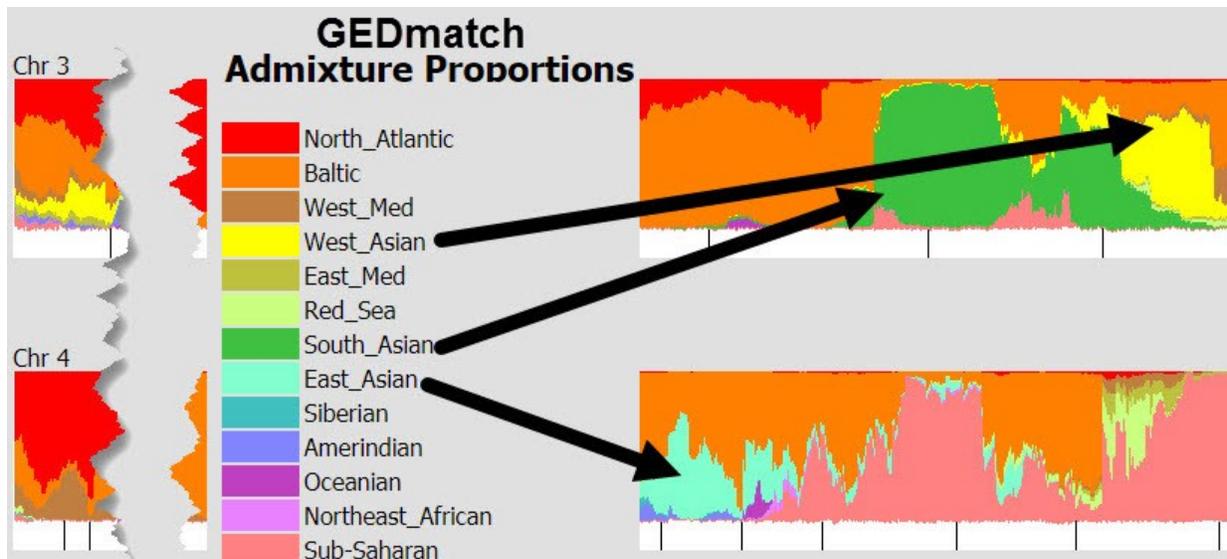


Figure 6. GEDmatch Admixture Tool Mapping Ethnicity.

WHY SHOULD GENEALOGISTS MAP OUR CHROMOSOMES

All of these pretty pictures are cool, but they can also help with your genealogical research. When a new DNA match appears in your list, compare the DNA segments shared with this new person to those segments you have mapped. If the new person shares a segment already mapped then they should share the ancestral line that segment is mapped to.

Confirmation of Genealogical Conclusions

Sharing DNA segments with multiple cousins who descend from the same ancestral couple corroborates our genealogical conclusions linking those people into our family tree. The more DNA matches you have with test-takers descended from multiple children of an ancestral couple, the more credible the conclusion that these are really your ancestors. Of course, if five test-takers match each other on DNA and all have copied the same wrong ancestor into their tree, those researchers may assume the DNA confirms the ancestor. The more credible conclusions are the result of a large number of test-takers who all match each other and who all have well-

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researched and accurate trees, preferable individually researched as opposed to copied from elsewhere.

Focus New Genealogical Research Effectively

A shared DNA segment with multiple test-takers who all name the same ancestor or ancestral couple provides clues that you should look for a link to that family. Your ancestor could be one of the siblings, descendants, or ancestors of a matching test-taker's ancestor. Somewhere in that lineage you will find a common ancestor, but it may be further back than you or the other test-taker have researched.

Even if you have not mapped any DNA segments further than to grandparents through visual phasing, that chromosome map can eliminate one-half or three-fourths of your tree as a likely place to search for a common ancestor with a new DNA match. That can save time and money.

Maps Provide a Visual Representation That May Interest Non-Genealogists

Chromosome maps may catch the interest of a cousin who has not been interested in genealogy before. Showing a child a map illustrating which DNA was inherited from each of his grandparents might stir an interest in genealogy or science or both. For those willing to do some scientific research, we can determine the placement of genes for eye color, hair color, or even a medical condition, and determine which ancestral line passed that trait down.

CONCLUSION

Chromosome mapping provides clues to focus our genealogical research more efficiently and effectively. Sometimes it may be our map that provides a clue for someone else. At other times someone else's map may provide a vital clue for us. Any time we have images we are more likely to interest a non-genealogist who may become the genealogist who carries our work forward.

For additional information see "Chromosome mapping," *ISOGG Wiki* (https://isogg.org/wiki/Chromosome_mapping).

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Debbie Parker Wayne, CG, CGL, is experienced using DNA analysis as well as more traditional techniques for genealogical research in Texas, the South and the West. She coordinated the first genetic genealogy courses at the three major U.S. genealogy institutes and is the Texas State Genealogical Society's DNA Project director. She wrote the NGS *Continuing Genealogical Studies: Autosomal DNA* course and co-authored the first genetic genealogy workbook, *Genetic Genealogy in Practice*, published by NGS. See <http://debbiewayne.com/> for more information.

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