Visual Phasing Workshop

By Stephanie Saylor

DNA and Chromosomes

DNA is a recipe for making you and every other living thing. Much of our DNA that we use in genetic genealogy is organized into chromosomes.

Chromosomes are like the recipe for life written on a very long pieces of tickertape wound tightly, to keep it from tangling around a series of spools. You get one set of chromosomes (1-22 and a sex chromosome) from your mother and another complete set from your father.

Because DNA is inherited from a person's ancestors, people who share your DNA are generally your relatives.

> **DNA Theory**
>
> If you and another person test the same type of DNA, and you "match" each other (share DNA with each other), than you are genetic relatives and share a common ancestor

Chromosome Browser

A chromosome bowser is a way to visualizes the amount of DNA you share with a DNA match and where that DNA is shared on the chromosomes. A variety of chromosome browsers are available for you to use for chromosome mapping.
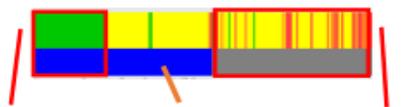
Fully-identical   Half Identical   No match

Visual Phasing

Visual phasing is a type of chromosome mapping that is preformed by comparing 3 siblings' DNA to each other to find the points of change (recombination points/crossover points) and create a map corresponding to those points of change, eventually showing the DNA all three siblings inherited from their 4 grandparents.

Specific colors assigned to segments on a chromosome can be labeled for ancestry belonging to a specific grandparent. Each chromosome must be individually labeled on this map. This can be less time consuming than other forms of chromosome mapping when grandparents are not available for targeted DNA testing.

The development of the visual phasing technique is attributed to Kathy Johnston, and was made widely popular by Blaine Bettinger. Kathy and Blaine both have released PDF's explaining how to use this technique to build a maps of all autosomal DNA chromosomes and the X-Chromosomes for 3 siblings simultaneously.

- Kathy's PDF explaining the process

- Blaine's PDF explaining the process

To preform visual phasing you will need access to a chromosome browser which reveals if you and a genetic relative share a segment of DNA on one (half-identical match) or both (fully-identical match) your chromosomes.

GEDmatch's chromosome browser is most commonly used for visual phasing as it can give you specific measurements for where your fully-identical and half-identical shared segments begin & end, as well as standardized chromosome lengths for all autosomal chromosomes (they are actually different lengths) eliminating some of the need to resize the browser results. An example of how to read GEDmatch results is to the bottom left.

Principle of mapping

Use what you know from the chromosome browser results to fill in the map for the sibling's grandparents using 4 colors

1. Notice which sibling underwent recombination
- No changes happen in a persons chromosome without recombination
2. Look at similarities and differences:
- Fully identical= 2 siblings share the same results for both chromosomes
- Half identical=2 siblings share DNA on only one chromosome
- No match= 2 siblings have completely opposite results

Step 1: **Find 3 full siblings to DNA test**

This technique can *only* be used if you can DNA test at least 3 siblings and have the ability to compare the 3 sibling to the other 2 siblings.

- Sibling 1 compared to sibling 2
- Sibling 1 compared to sibling 3
- Sibling 2 compared to sibling 3

Step 2: **Decide what software you would like to use to create your chromosome map.**

Many people use Microsoft PowerPoint but any document editing software where you can insert shapes and pictures will work. I have used Word and PowerPoint in the past for this purpose.

Step 3: **Find or create a template**

No matter what software you use you will need to repeat this process for the 22 autosomal pairs and the X-chromosome. Having a template prepared in advance will significantly speed up your work.

I have a link to my PowerPoint template for your reference below. You should use whatever template you feel most comfortable with.

- [Stephanie's PowerPoint template](#)

Using find and replace tools can help speed up customization of your template.

Step 4: **Use a chromosome browser that shows fully-identical regions as well as half-identical regions, like the 'One to One' compare tool at GEDmatch, to compare each sibling to each other**

You will have to run 3 comparisons to compare each sibling to every other sibling in our sample.

| Sibling	GEDmatch number/Name	Sibling	GEDmatch number/Name
1		2	
1		3	
2		3	

Step 5: **Insert clips of each chromosome in your templates for chromosomes 1-22 and X**
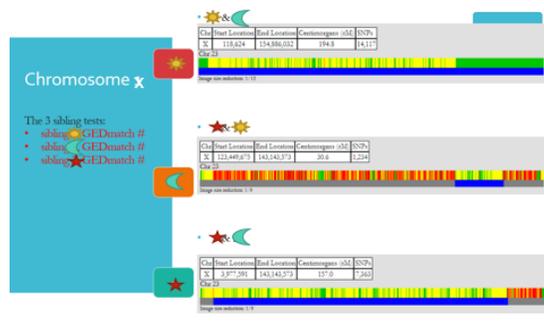
You will repeat this process for every comparison of the 3 siblings.

## *Step* 6: **Mark the recombination points with a red line**

Recombination points will be evident when there is a change from half, full, or no match to another
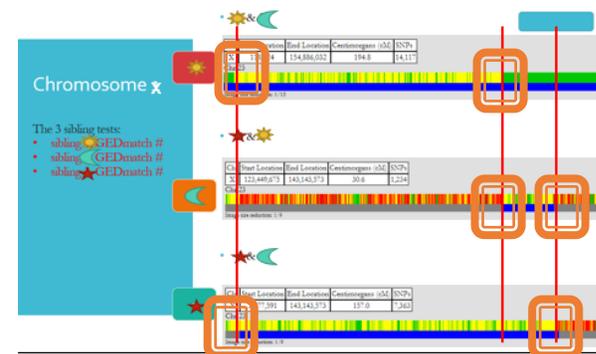
## *Step* 7: **Label the recombination points**

Change should always occur in 2 of the 3 comparisons you preformed on your 3 siblings. The sibling that had a change in both comparisons had recombination occur at that point.

Mark the sibling who had recombination at that point of change crossover with a single letter or symbol associated with them.
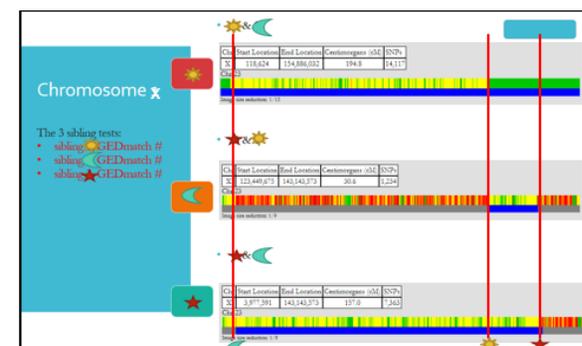
Step 8: Fill in the map

Start with a comparison containing a larger fully-identical region. In this example, the Sun and Moon are identical on right I know the 2 siblings compared in that test have the same DNA results there.
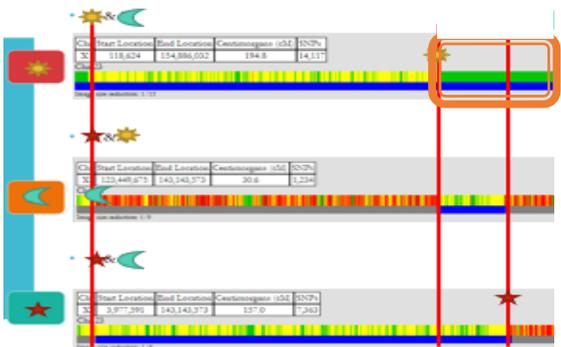

Since a sibling will only change from inheriting DNA from one grandparent to another at the points of recombination assigned to that sibling I can extend the chromosome results out to that siblings point(s) of recombination
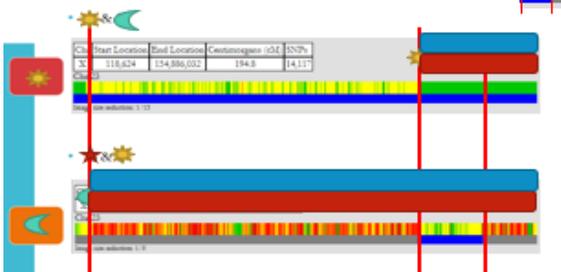

One or more chromosome could have had recombination at any crossover point but usually we see a single chromosome undergo change at the recombination point so try extending only one of the chromosomes past the recombination post for the sibling that underwent recombination at that point.
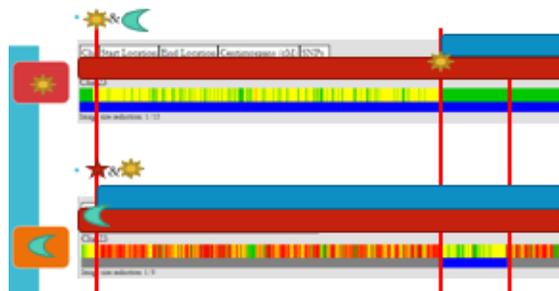

The other chromosome had a change so the color of the chromosome present will change on the chromosome which underwent recombination. This completes our examples map of the X-chromosome for the Moon sibling.
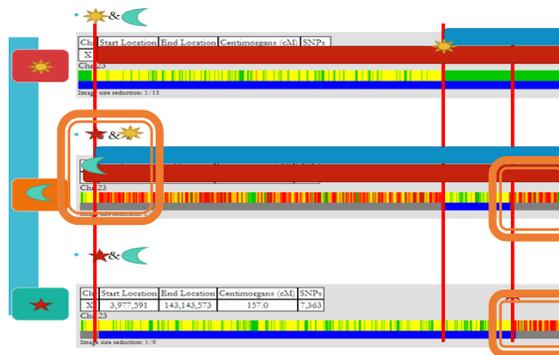

The 2 tests involving the star are both no matches on the far right. This means that the star is completely opposite from the Sun and Moon at this location.
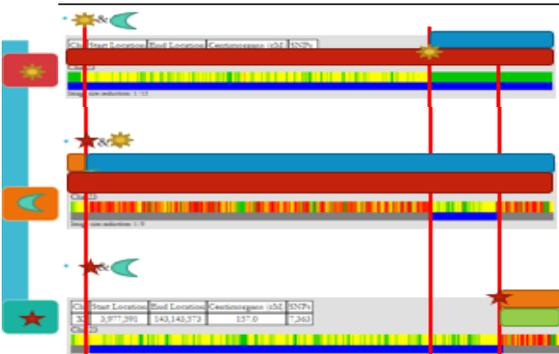
You can again generally assume only one chromosome underwent recombination at a single crossover point allowing you to continue to fill in the corresponding colors
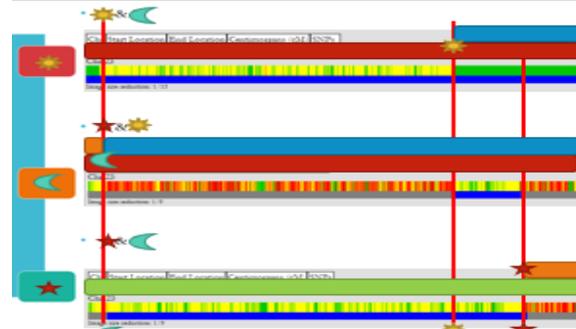
Verify that change in a siblings chromosomes only occurred at a recombination point for the sibling
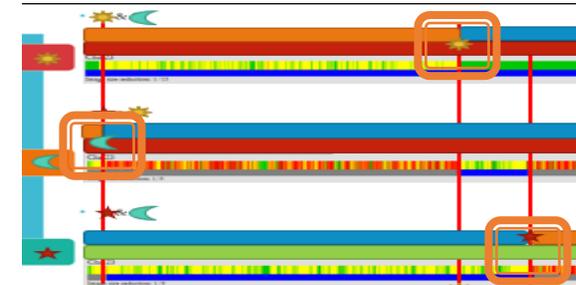
Verify that the 3 siblings are 1/2 identical , fully identical and not matching at the correct locations based on the chromosome browser results for those siblings.

There should only be one combination of colors that can fulfill these requirements. If a different arrangement of the chromosomes seems to be able to work recheck your work. for mistakes.

If everything checks out you are done with the mapping portion.

Step 9: Label the grandparents

Once all the chromosomes are mapped or your chromosome of interest is mapped you can start labeling your chromosomes for what grandparent belongs to what color for each chromosome separately. There are some tricks to do this and we will gloss over a few things to look out for.

Compare a match related to the siblings through a specific grandparent (tested close cousins make this part easier). Matches on more than 1 sibling can help you identify what chromosome the match is on. In the example below: both Moon and the Star had half-matched a match in the same location.
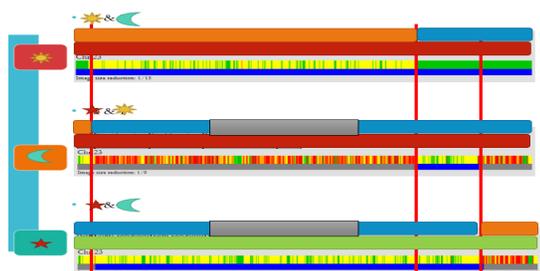

This let us know that that match is on the blue chromosome and all blue chromosomes are DNA we received from that specific grandparent who is related to the match. In this case we will say the match is on the paternal grandfathers side. *Label all the blue sections of only this chromosome the name of their paternal grandfather.*
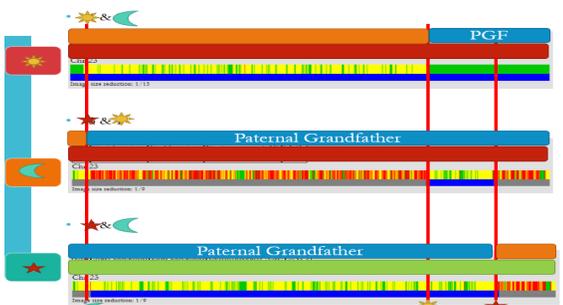
Your father's parents passed on a combination of their parents DNA to you. Put together this is your paternal DNA and it constitutes an entire chromosome.

After labeling one of the colors as the paternal grandfather, you can then label the complimentary color (orange) on the top, the paternal grandmother. All the DNA on the top chromosome is now labeled.
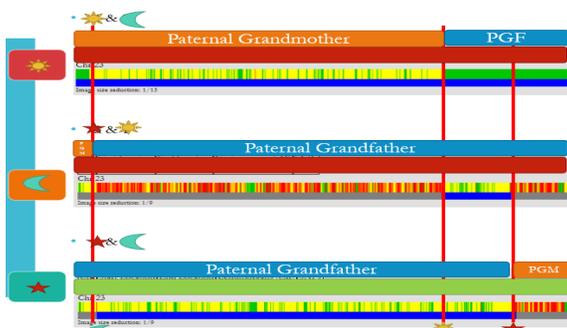
If the orange and blue sections of chromosome were part of the chromosome copy the siblings inherited from their father (paternal) than the green and red would be sections of chromosome inherited from their mother (maternal.).

You compare a match related to your through a maternal grandmother to all 3 siblings . In this case the match only shared a segment with Star on this chromosome (so it is a match on the green segment). Allowing you to label all green segments maternal grandmother, and all red segments from the siblings maternal grandfather.

Label your start and stop points using the positional data on the GEDmatch chromosome browser.

You can type in the points to the segment, or use call out boxes, others put the information in a table separate from the map itself. Decide for yourself what you like.

Remove the chromosome browser reports and the lines indicating the recombination points as you no longer need them. You can now dress up your report to make it look fancier by giving the chromosomes depth or changing other details. The examples below for the completed X chromosome.

Repeat step 9 for each chromosome individually. After labeling the chromosomes you may wish to standardize the colors so that orange is the grandparent Abigale Adams for every chromosome mapped.