Visual Phasing

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This originally appeared as a series of five blog posts at The Genetic Genealogist (www.TheGeneticGenealogist.com), between November 21-27, 2016:

1. **Part I** – Explaining visual phasing and identifying/labeling recombination points
2. **Part II** – Assigning segments of DNA
3. **Part III** – Using cousin matches to identify which grandparent provided the segments
4. **Part IV** – Mapping my own chromosome using the visually phased paternal chromosomes
5. **Part V** – Using the mapped DNA with new matches

Visual Phasing: An Example (Part 1 of 5) – November 21, 2016

This weekend, I spoke at a meeting of the New England chapter of the Association of Professional Genealogists, and it was a wonderful group. One of my talks was about "Chromosome Mapping." Unfortunately, since the talk was only an hour, we didn't have time to discuss "Visual Phasing," a chromosome mapping methodology. Instead, I promised to finish this blog post to explain the process. As I was writing, the blog post turned into a 5-part series!

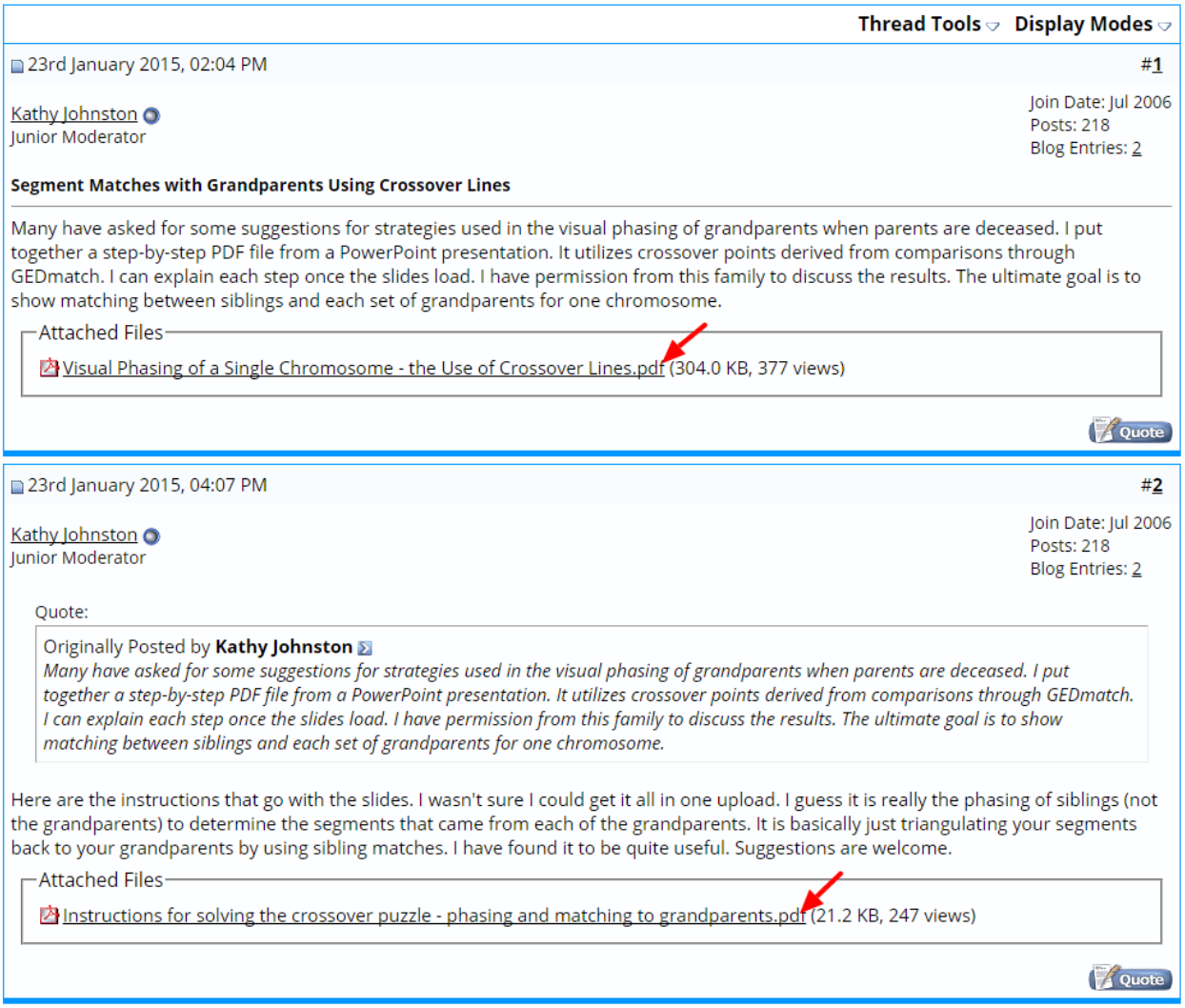
Quick Summary

- **What is it?** A method to assign segments of DNA to the test-taker's four grandparents.
- **Why use it?** To identify which grandparent gave the test-taker which segments of DNA (eliminating 75% of the family tree to search for MRCA).
- **What do you need?** Autosomal DNA of three siblings uploaded to GEDmatch.

Visual Phasing

Visual Phasing is a process by which the DNA of three siblings is assigned to each of their four grandparents using identified recombination points, without requiring the testing of either the parents or grandparents. Although the process does not automatically reveal which segment belongs to which of the four grandparents, matching with cousins provides this identification as a further step of the process.

Kathy Johnston developed this process some time ago, and first posted about it in the Family Tree DNA Forum (http://forums.familytreedna.com/showthread.php?t=36812). As shown in the figure below, there are two PDF documents available for download to explain the method with both images and text. However, note that you must be a registered member of the forum (free) to download the documents.



My understanding is that Randy Whited (https://about.me/rwhited) also independently developed this process. I attended his excellent lecture on visual phasing at SCGS Jamboree in June 2016, and an audio recording of that talk is available for

purchase here (Session# TH 023 entitled "Reconstructing Grandparent DNA Using Sibling Results" for $11.00) (http://www.myconferenceresource.com/products/45-01-scgs-genetic-dna-jamboree-conference-2016.aspx).

Visual Phasing is an incredibly valuable tool. Although requiring three siblings creates a considerable barrier for many people, it can be extremely valuable for genetic genealogists interested in chromosome mapping. For example, as we'll see, I have an adopted great-grandmother, and using visual phasing I can identify entire portions of my chromosomes that came from her, which could prove to be beneficial to my search.

NOTE: although visual phasing can potentially be performed with just two siblings and close cousin(s), it is considerably more challenging. I strongly recommend starting out with three siblings (either your own family or someone else's family).

Other Resources

In addition to Kathy's PDF documents and Randy's recording, there are several other resources. Joel Hartley has been publishing the results of his visual phasing (see "My Big Fat Chromosome 20") (http://www.jmhartley.com/HBlog/?p=462), as has Ann Raymont ("Chromosome Mapping with siblings – part 1" (https://dnasleuth.wordpress.com/2016/05/13/chromosome-mapping-with-siblings-part-1/) and "Chromosome Mapping with siblings – part 2") (https://dnasleuth.wordpress.com/2016/06/01/chromosome-mapping-with-siblings-part-2/). Ann's blog posts contain a lot of details about the whys and hows of visual phasing.

What You Need

You need three siblings who have done autosomal DNA testing and transferred their results to GEDmatch. The testing company doesn't really matter, even if you've tested all three at three different companies. Trust me!

I perform visual phasing in PowerPoint because it gives me a great deal of freedom to manipulate screenshots and add annotations, but it isn't perfect. I'd love for this process to be semi-automated, at least creating an output comparison for Step #1, below.

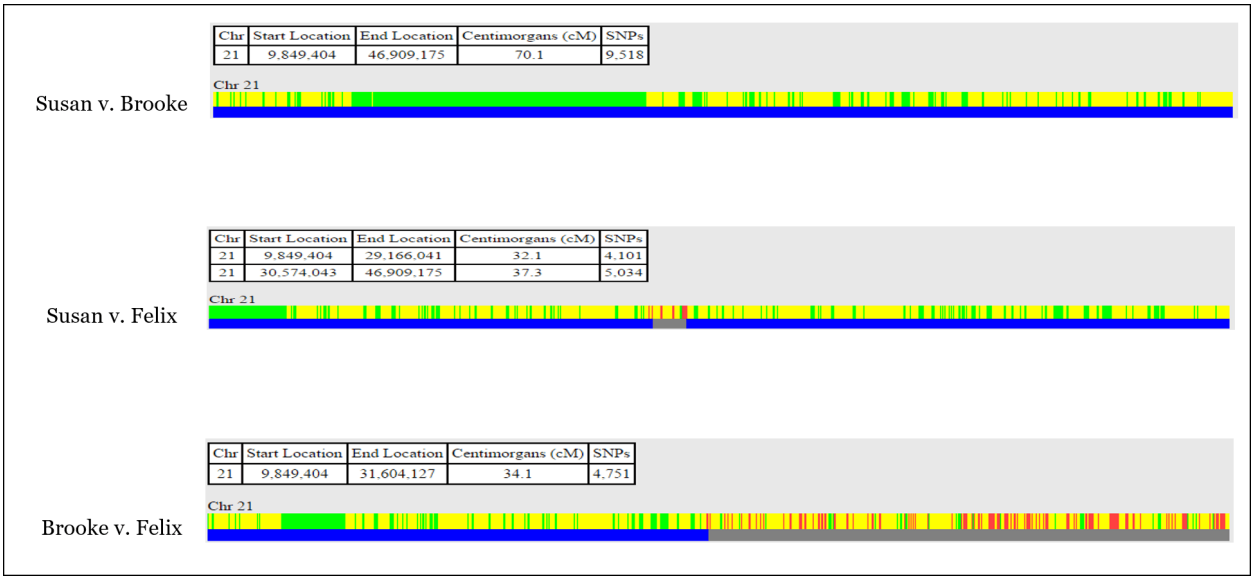
STEP 1 – Setting Up

Visual Phasing works by identifying recombination points in the DNA of the three siblings. As will become clear, a recombination event in one sibling will affect how she shares DNA with the other two siblings.

Accordingly, the first step in visual phasing is to compare the DNA of the three siblings to each other in GEDmatch using the One-to-One tool. We're going to work on **one chromosome at a time**, and I recommend starting with the X chromosome (especially if one of the siblings is male, since he'll only have one X chromosome) or one of the shorter chromosomes such as 20 through 22.

Capture a screenshot of the comparisons, and paste them into PowerPoint.

In this example, we're going to be looking at Chromosome 21 in three siblings, Brooke, Felix, and Susan:



With this information, you can identify most or all of the recombination that took place when the sperm and egg were created for each of the three siblings.

In One-to-One comparison, you'll usually see both half-identical (yellow) and fully-identical (green) sharing (but not on every chromosome). Remember that we're actually comparing TWO CHROMOSOMES of each person at each and every point, so sometimes full siblings will share DNA on only one of their chromosome pair, while they will also share DNA on both copies of their chromosome pair.

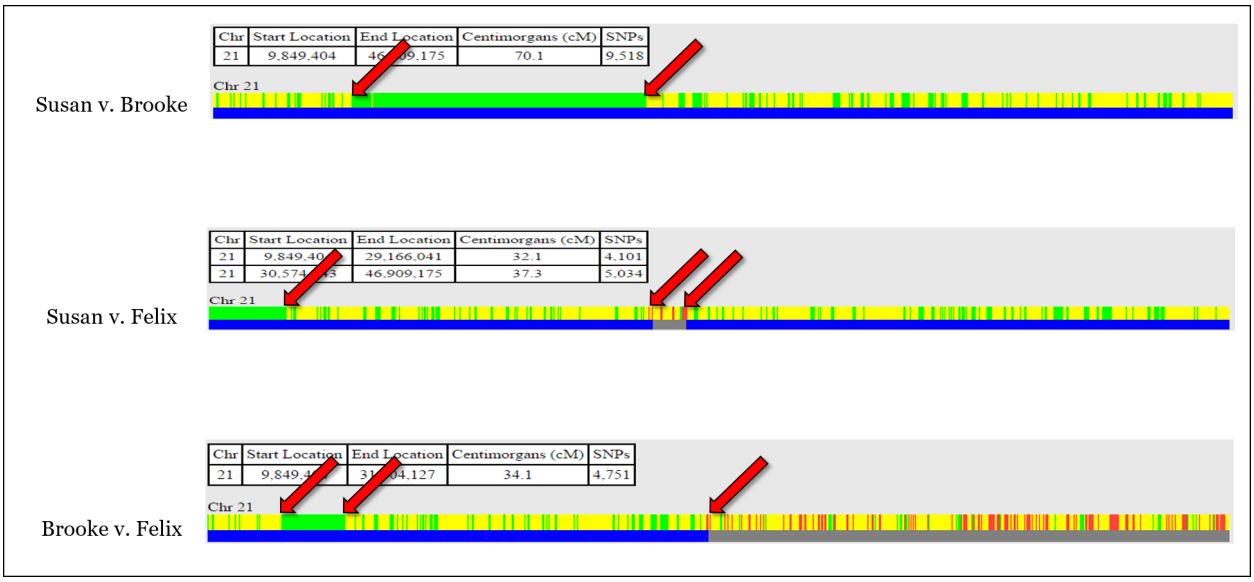
STEP 2 – Identify and Label the Recombination Points

Now we can identify and label the recombination points. Here is the first key point:

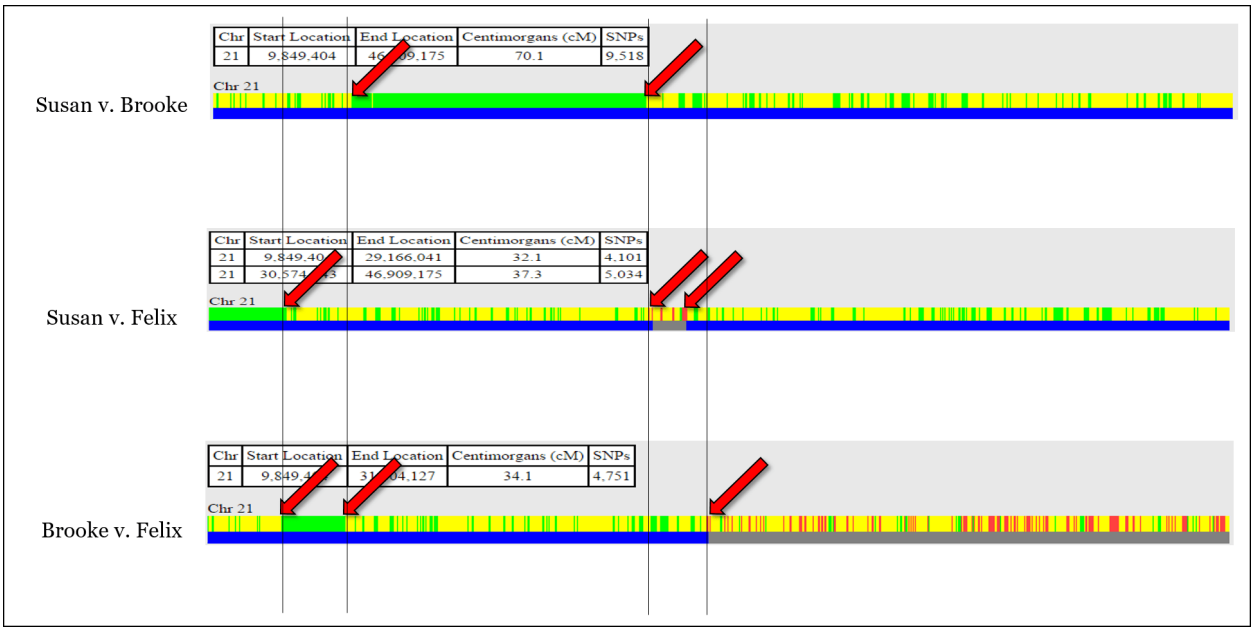
KEY POINT #1 *– Anywhere there is a change in the sharing status between two siblings, there must be a recombination event in at least ONE of the siblings (and sometimes both!).*

For example, a switch from sharing a yellow segment to sharing no DNA means there was recombination at that point in one or both siblings. A switch from a yellow segment to a full segment means there was recombination at that point in one or both siblings. And so on.

In the following figure, each of the recombination points (i.e., each change in sharing) is identified by an arrow:



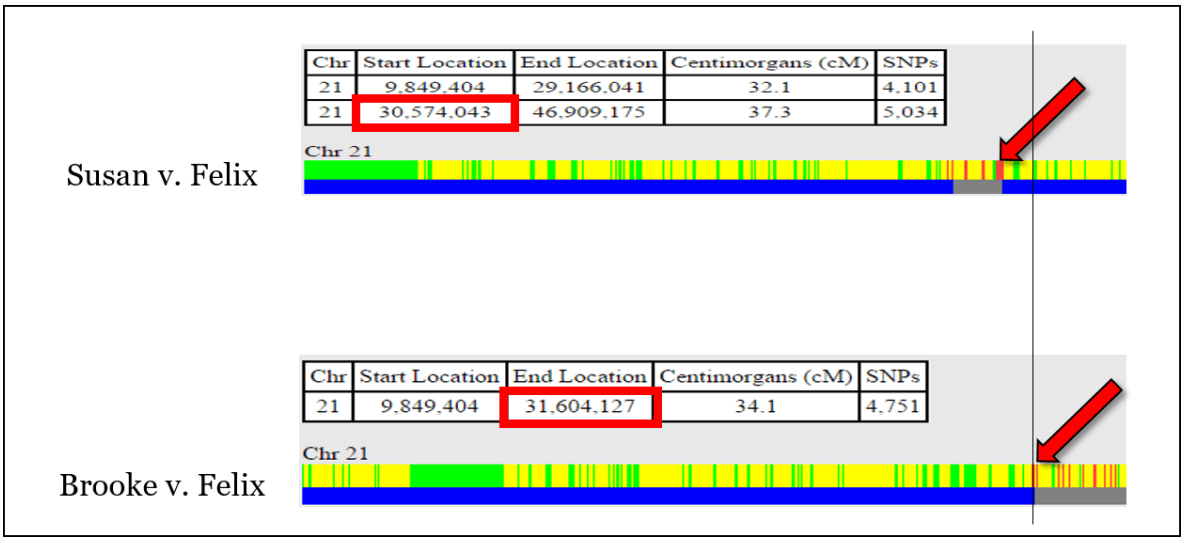
Now, in PowerPoint, draw a long vertical line through each recombination point. Each line should intersect at least two recombination points:



Often, this is where you're going to run into the first problem, namely that the line doesn't always seem to intersect at least two recombination points. This happens for a variety of reasons. Most commonly, the chromosome visualizations don't always line up perfectly. Second, sometimes the start and stop locations are fuzzy (Ann Raymont mentions this in her terrific blog post here: "Chromosome Mapping with siblings – part 2"). Third, sometimes there are recombination events in two siblings at one place, which can cause some difficulty.

KEY POINT #2 *– Do NOT get too stuck on recombination points. Trust me, getting frustrated with recombination points that don't line up can quickly derail a phasing project! "Close enough" is just fine when trying out the first few chromosomes.*

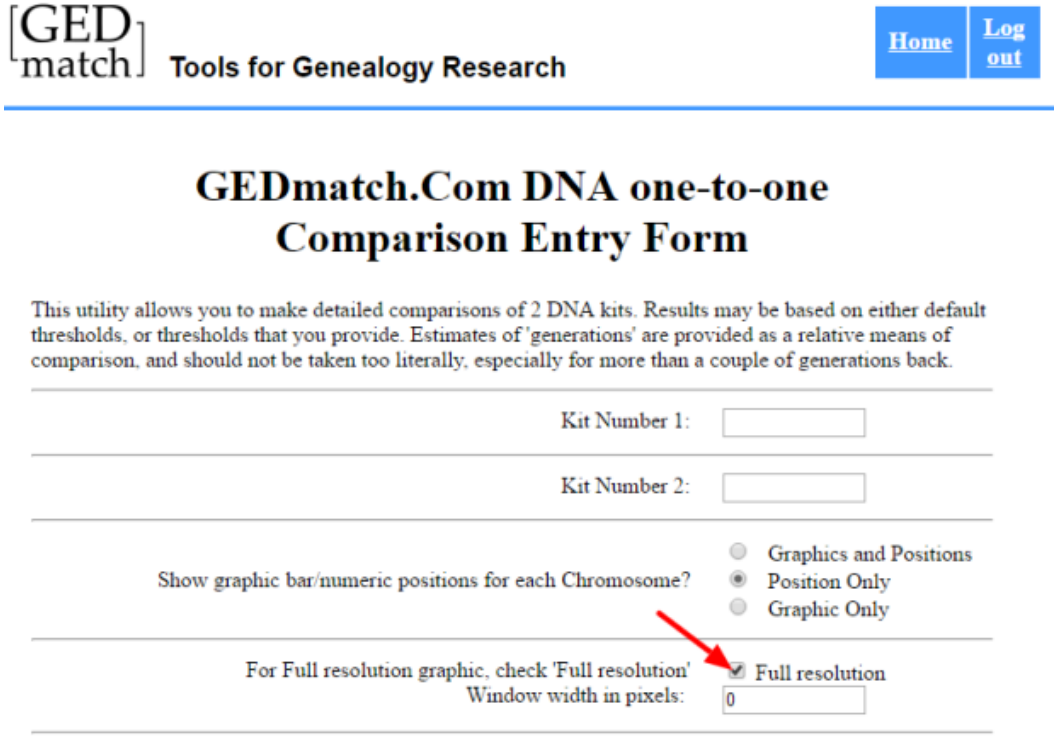
For example, there is an issue aligning the recombination event shown below, which is at position 30,574,043 according to the comparison of Susan v. Felix, but at position 31,604,127 according to the comparison of Brooke v. Felix. This is unlikely to be "fuzziness."



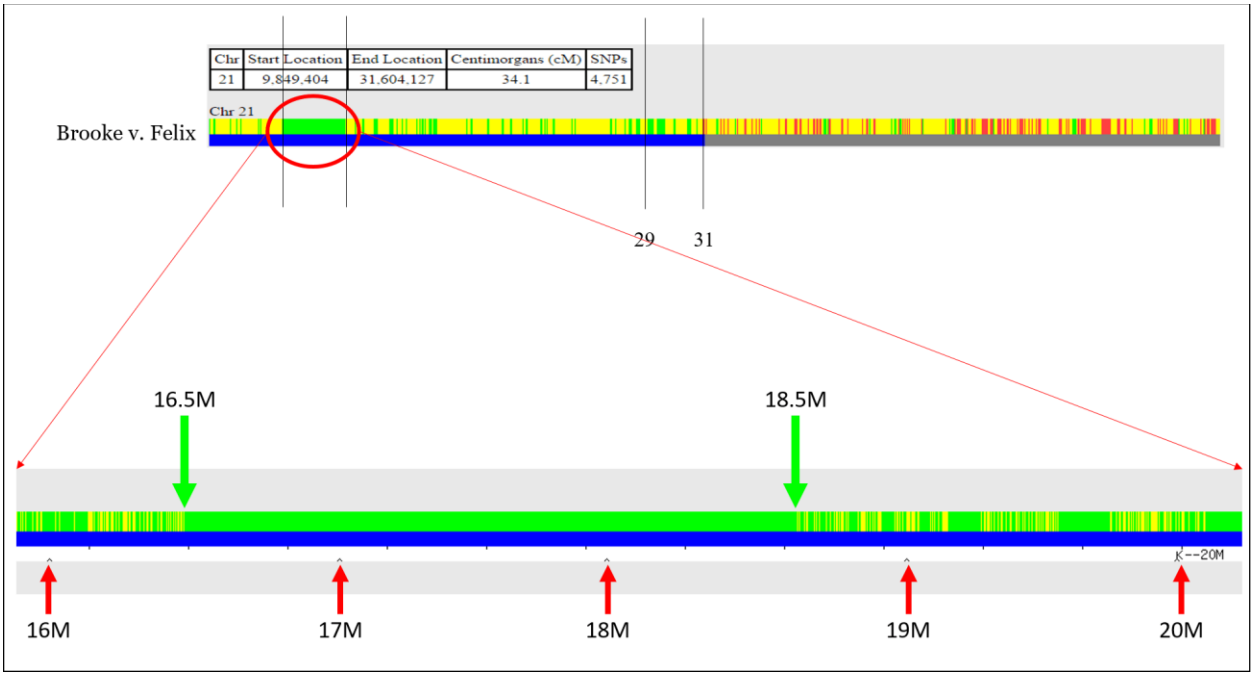
It's very easy at this point to throw your hands up and jump to another chromosome. But for now, we'll put a recombination point around 31,000,000 or so.

I also like to label the recombination points with a number for easy reference. The start and stop positions for each yellow segment (sharing on one chromosome) is provided by GEDmatch.

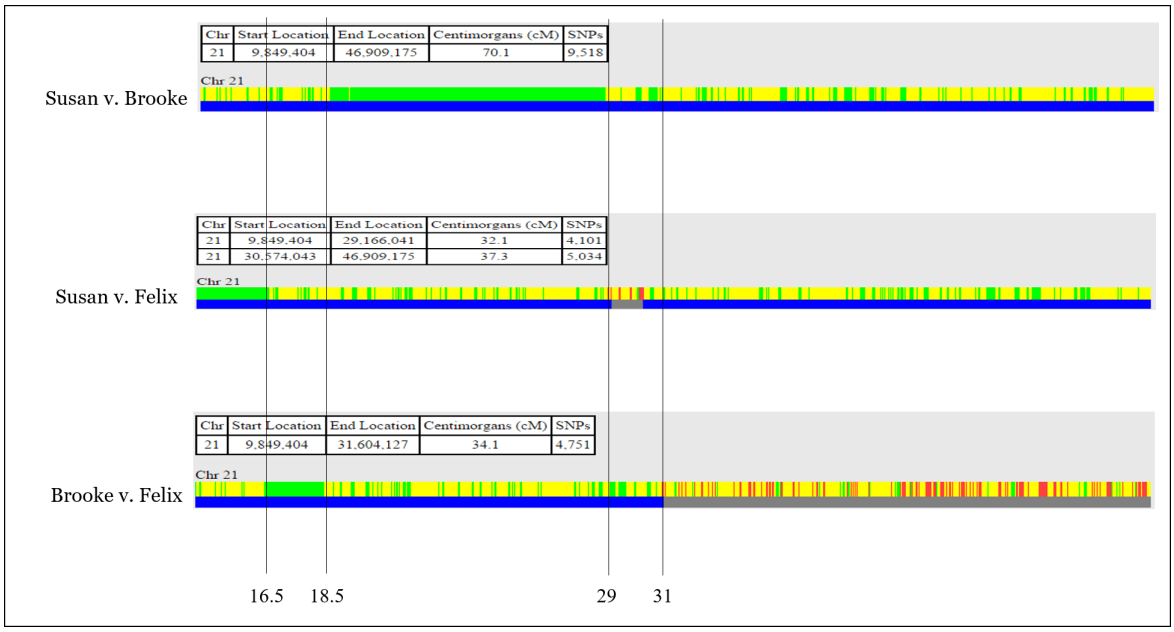
However, you'll need to take an extra step to get the start and stop point for green segments (sharing on both chromosomes). This is a great trick that I just learned recently (via Sue Griffith at "Obtaining FIR Boundaries on GEDmatch using the Little Tick Marks") is to perform a One-to-One comparison, but to click "Full resolution."



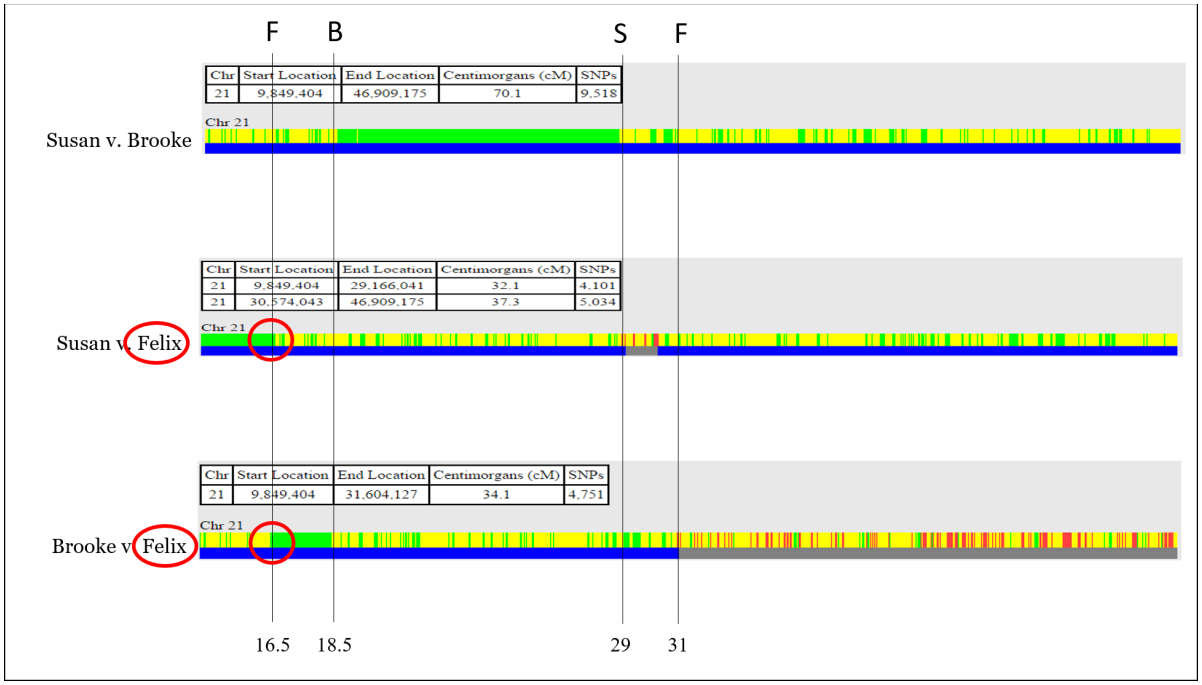
This will expand the chromosome and show megabase positions, and you can obtain the start or stop position:



So now we have positions for each of the recombination points:



Now, let's assign each recombination point to the person for which that recombination occurred. This is usually as straightforward as identifying which sibling has the recombination event twice. For example, Felix owns the first recombination point:



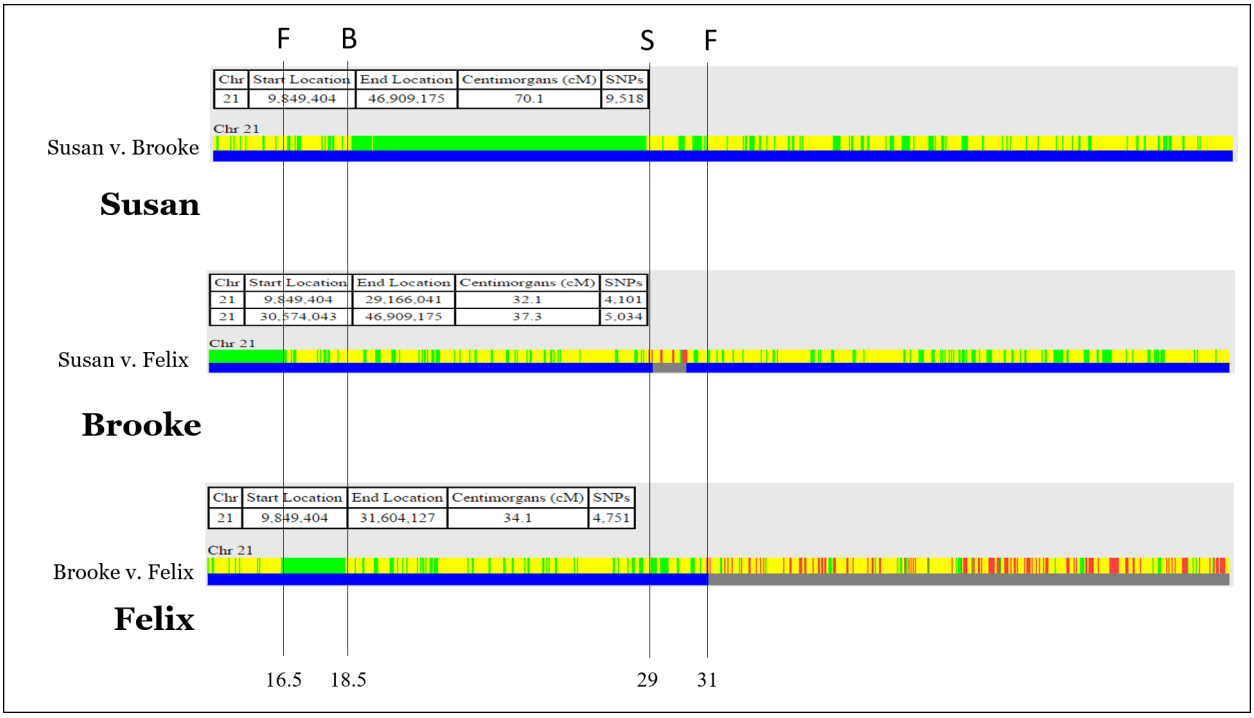
Now all recombination points are identified and labeled.

In the next post, we'll start assigning segments of DNA based on the identified and labeled recombination points.

Visual Phasing: An Example (Part 2 of 5) – November 22, 2016

Originally Appeared at:
http://thegeneticgenealogist.com/2016/11/22/visual-phasing-an-example-part-2-of-5/

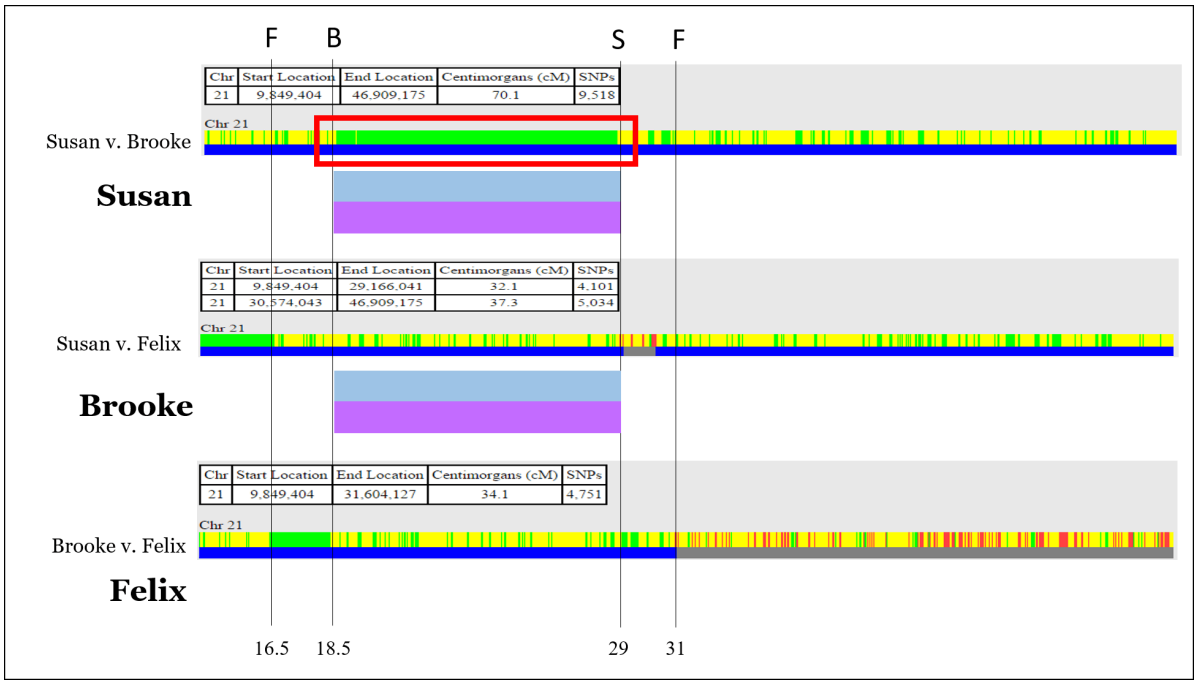
In "Visual Phasing: An Example (Part I)," we identified and labeled all of the recombination points in the three siblings, Susan, Brooke, and Felix:



In Part 2 of the series, we'll use the identified and labeled the recombination points to assign segments of DNA to the four grandparents.

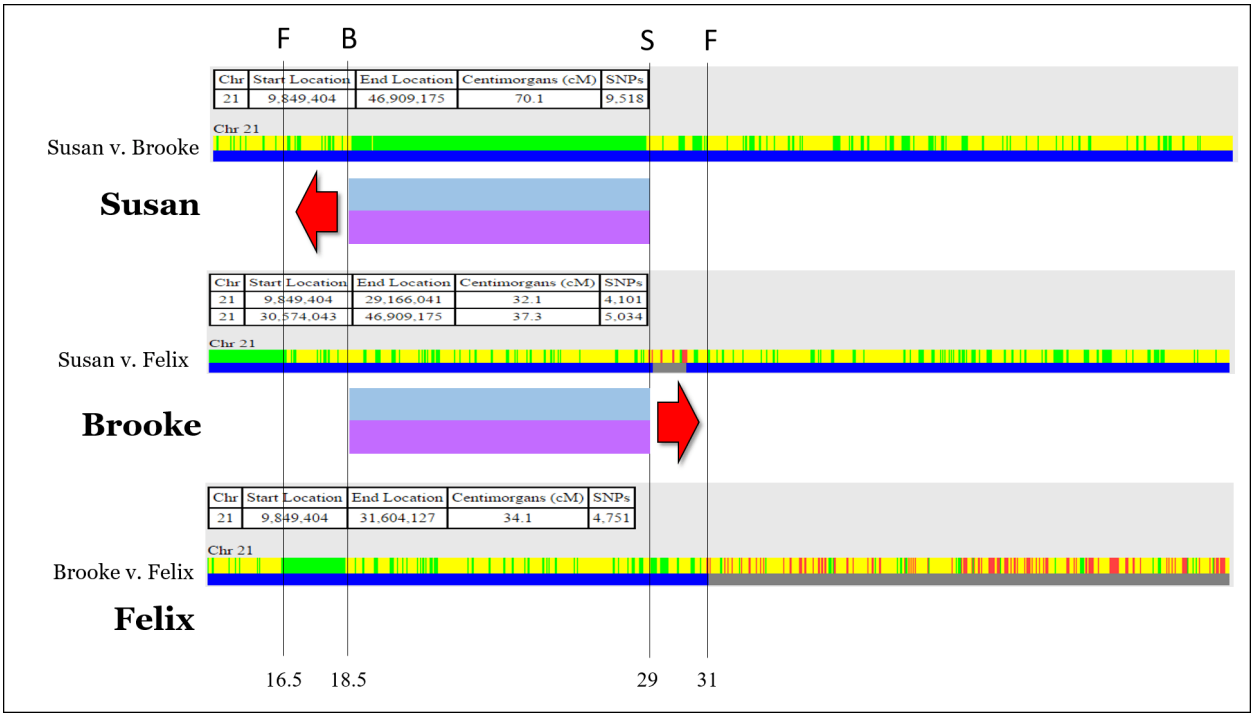
Step 3 – Fill in Chromosomes

The next step is to fill in the chromosomes using the identified recombination points. We're going to start with the fully identical region shared by Susan and Brooke, shown in the red square in the next image. We can fill into both chromosomes for Susan and Brooke (the blue and purple segments):

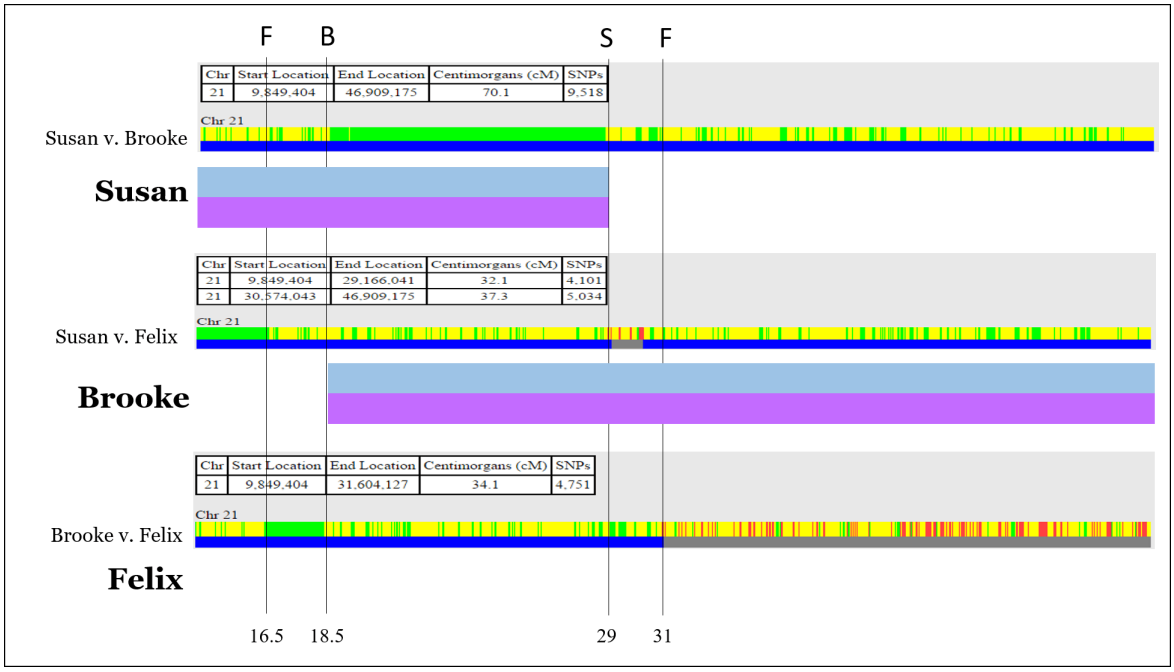


Each of the blue and purple segments is one of the following segments: (i) paternal grandfather; (ii) paternal grandmother; (iii) maternal grandfather; or (iv) maternal grandmother. We'll try to figure out which is which later.

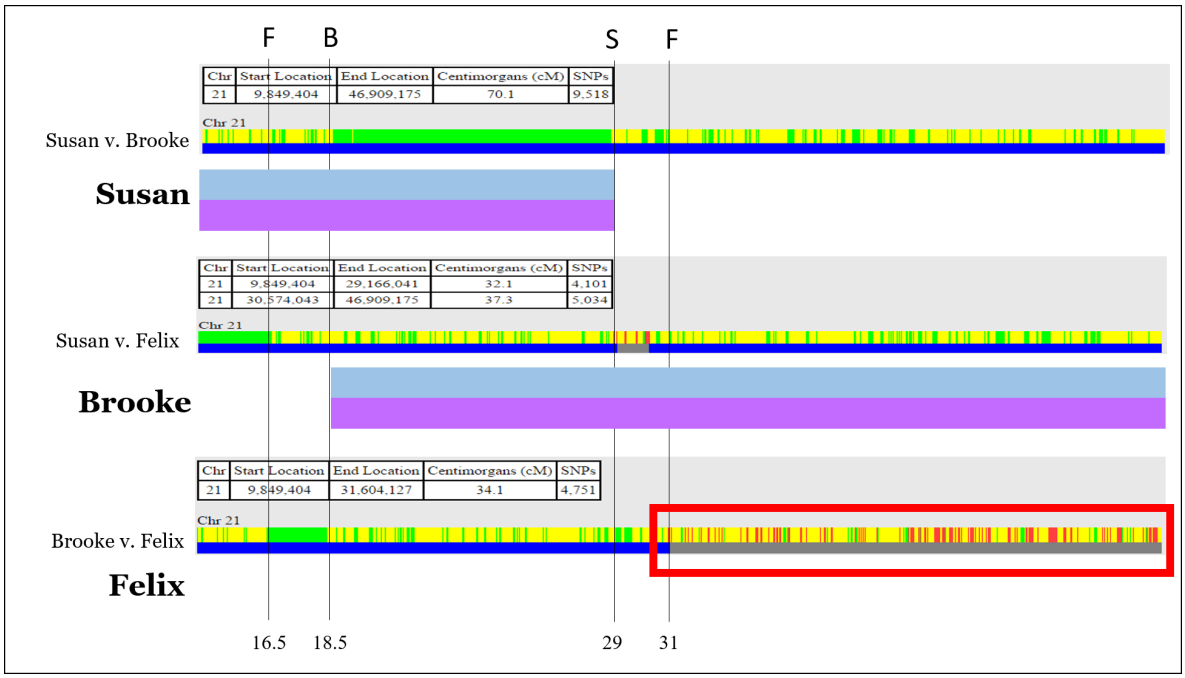
We do know that a segment will continue until it reaches a recombination point. So we can extend Susan and Brooke's segments outward in each direction to either the end of the chromosome or the next recombination point:



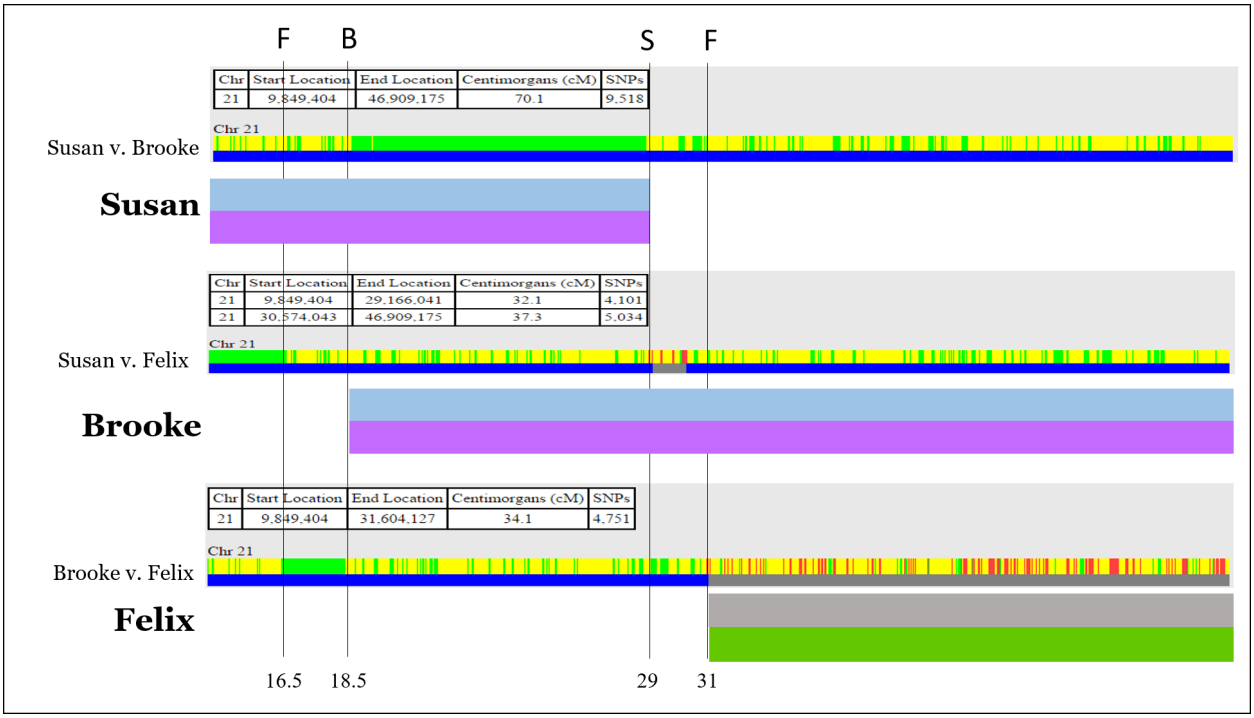
Susan's segments have a recombination point directly to the right (the line marked "S"), but can be extended to the left all the way to the end of the chromosome. Brooke's segments have a recombination point directly to the left (the line marked "B"), but can be extended to the right all the way to the end of the chromosome:



Now that we have Brooke's entire chromosome to the right end, we can focus on the comparison of Brooke and Felix. As shown in the red square in the next image, Felix and Brooke share no DNA on the right half of the chromosome, from Felix's recombination point to the end:



Since we know that Brooke's chromosomes at that location are blue and purple, Felix's chromosomes must be the two other colors we are going to use. Now we can fill those in, and we have all four colors (for paternal grandfather, paternal grandmother, maternal grandfather, and maternal grandmother, although we don't know which is which right now):



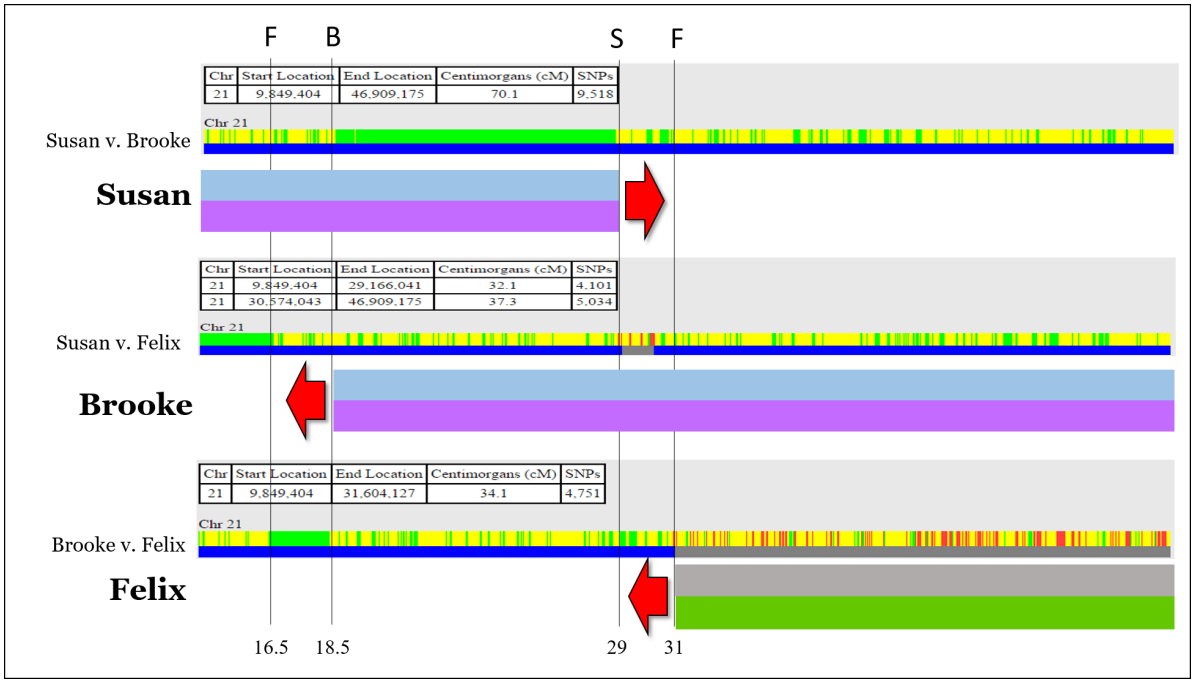
But now we're stuck. For the most part (except for the left end of Felix's chromosomes, which are identical to Susan's), the GEDmatch One-to-One comparisons don't help us fill out the remainder of the chromosomes.

WARNING! Now comes the most confusing part of visual phasing!

This took me a while to figure out and become comfortable with, and it can be difficult to explain. Essentially, we have to make an arbitrary decision at this point, but this it the only arbitrary decision we can make. Once we make it, we're stuck with our decision.This effectively cements a certain color combination as 'maternal" and the other color combination as "paternal," although I don't yet know which is which.

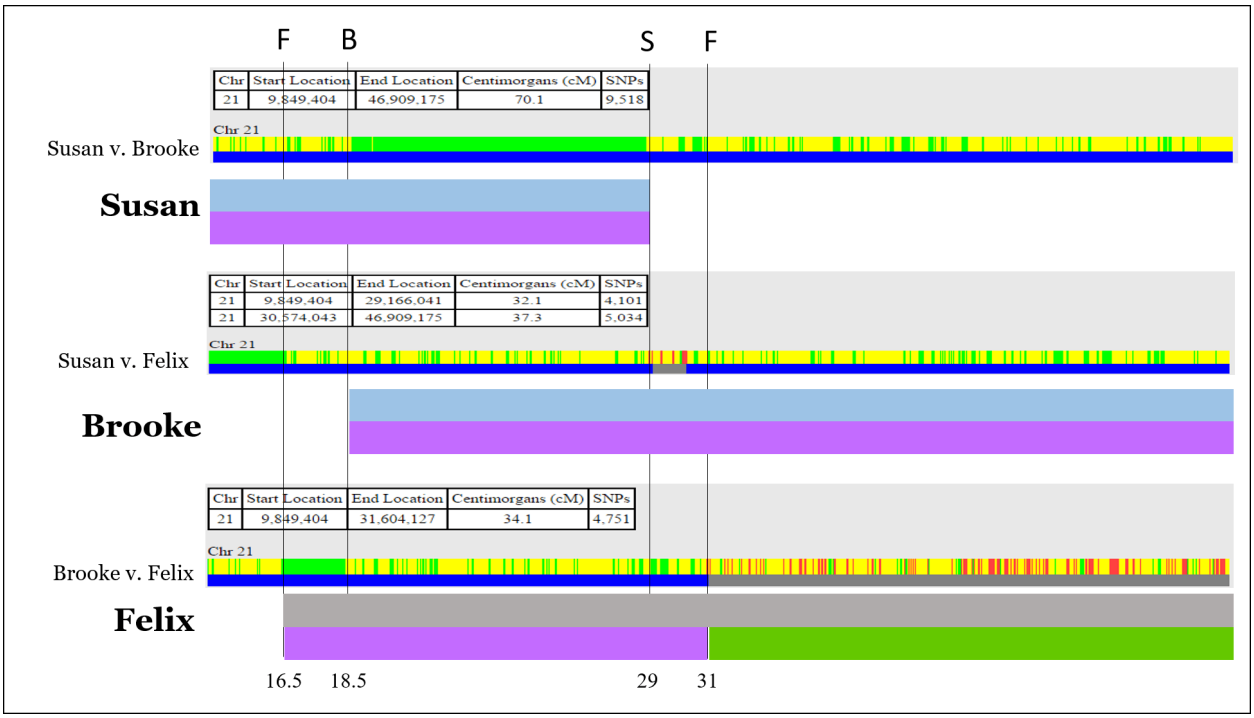
Each of Susan, Brooke, and Felix experience a recombination event at this point. We're going to chose one person, and one chromosome, to extend. Once we do that, the relationship of all the colored segments to each other is fixed, and we won't be able to arbitrarily decide which chromosomes to extend from the other two people.

So we have to extend a chromosome at one of the following red arrows:



KEY TIP: This decision point is a good place to come back to if you think you've made a mistake, or if the chromosome you decided to extend wasn't helpful. For example, in the image above, extending Brooke's segment and finishing her chromosomes might not allow us to complete the entire puzzle.

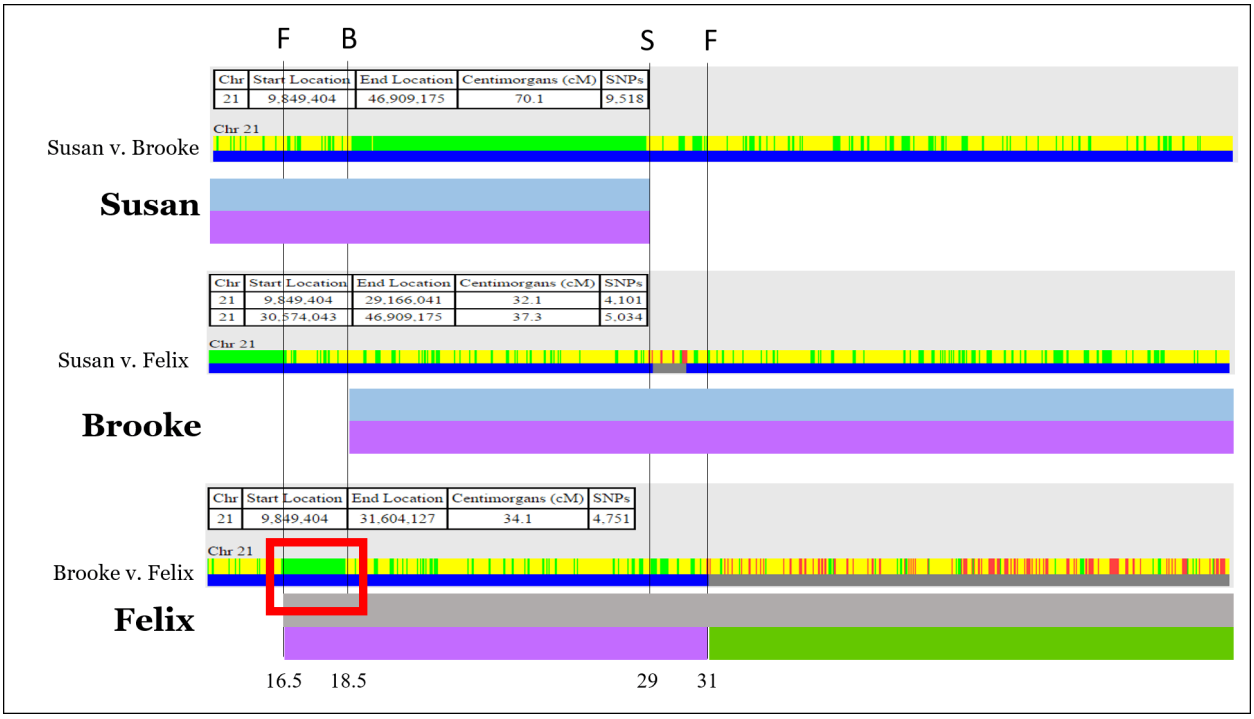
For example, let's extend Felix's chromosome gray chromosome, which means that the green segment has the recombination point and switches to the complementary color, purple. We can extend both of those out to Felix's next recombination point:



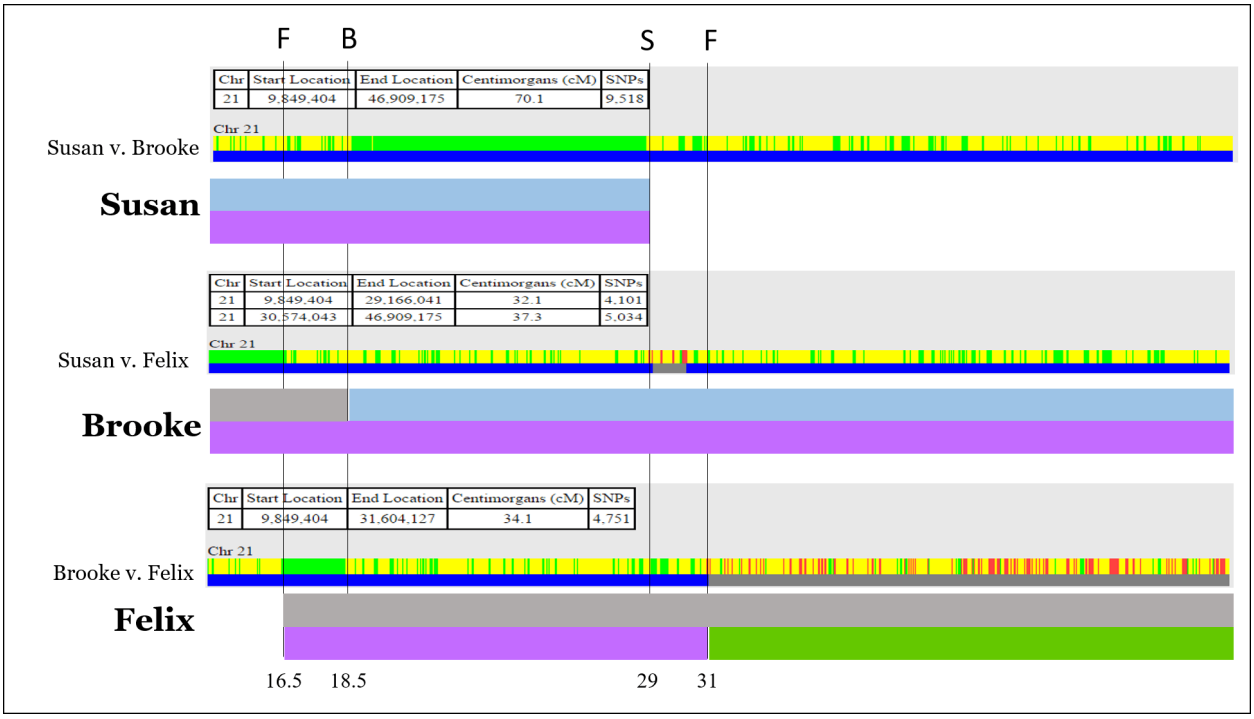
Before that decision, purple could have been either paternal or maternal, and blue could have been either paternal or maternal. They were interchangeable. Now, however, although we still don't know which is which, we know that the purple and green chromosomes underwent recombination in either the mother or father at that point.

So we can't arbitrarily decide which segments to extend in Brooke and Susan, instead we have to go back to the GEDmatch One-to-One comparisons.

We see that Brooke and Felix share on both chromosomes in the region shown by the red box in the next figure:

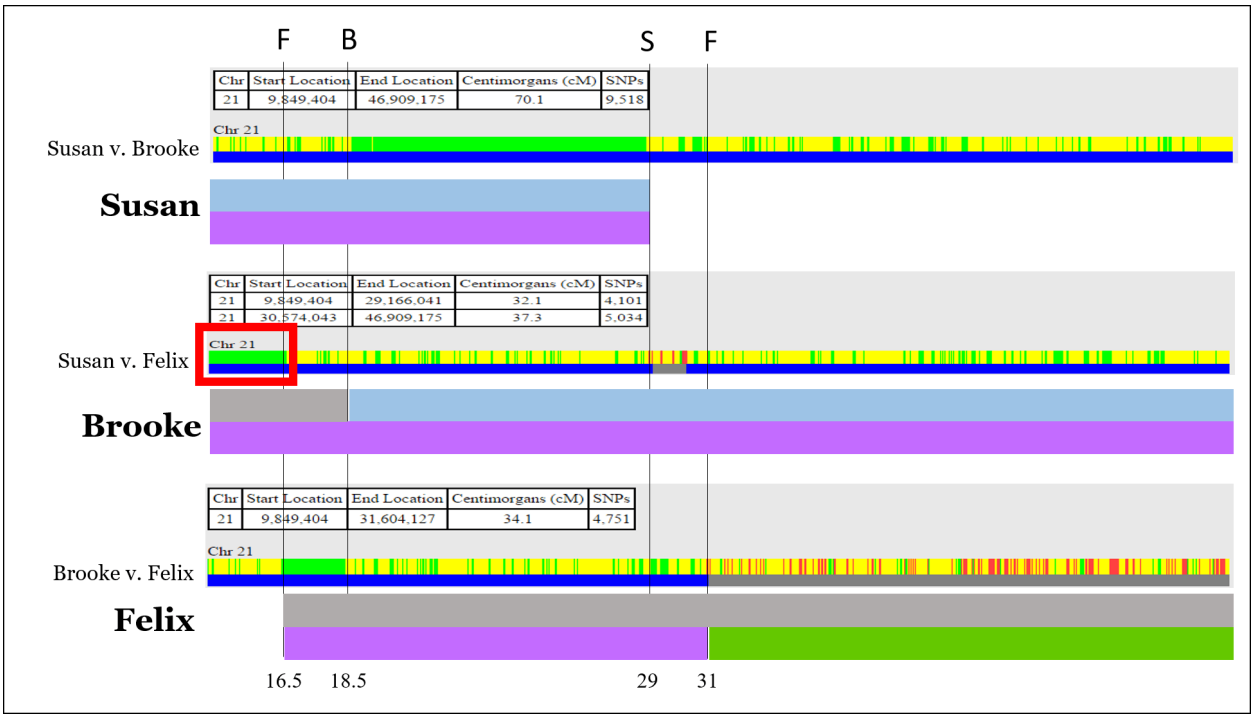


Since we know what Felix's chromosomes 'look' like at that region, we can fill in Brooke's chromosomes at that region, and we know which extends and which experienced recombination. And, we can extend that out to the end of Brooke's chromosome:

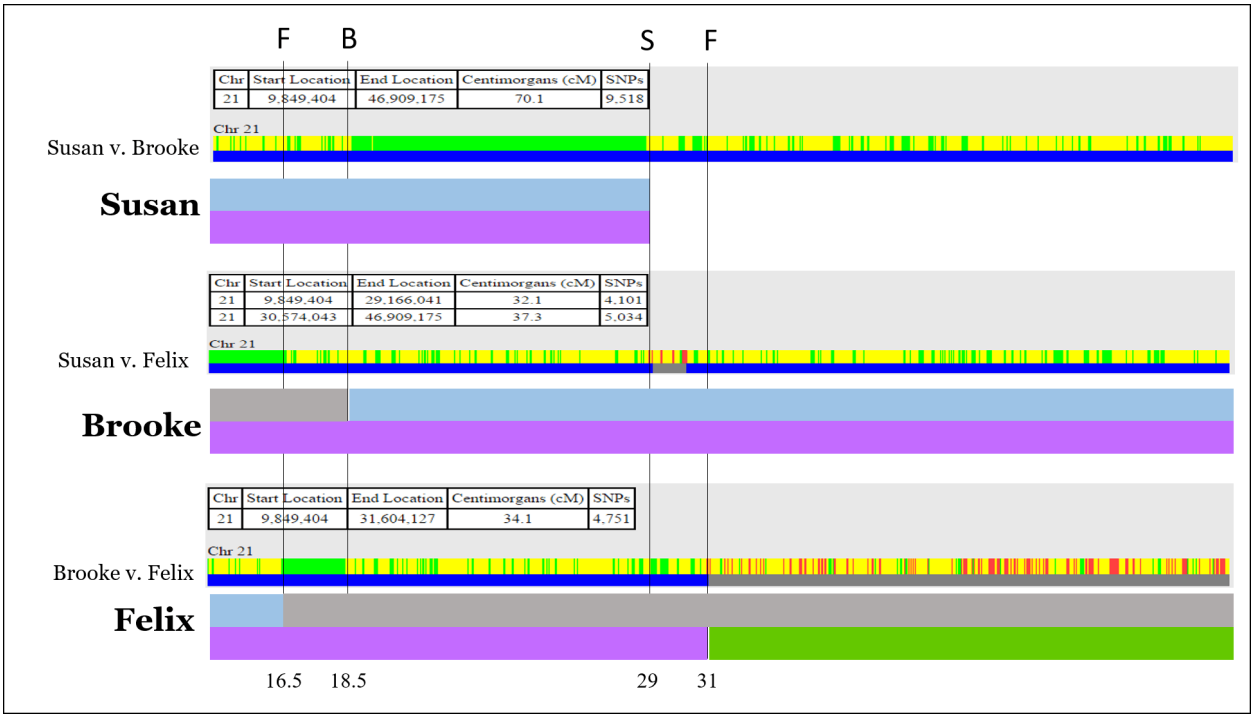


We've now finished Brooke's chromosome entirely!

We see that Susan and Felix share on both chromosomes at the region shown by the red box in the next figure:



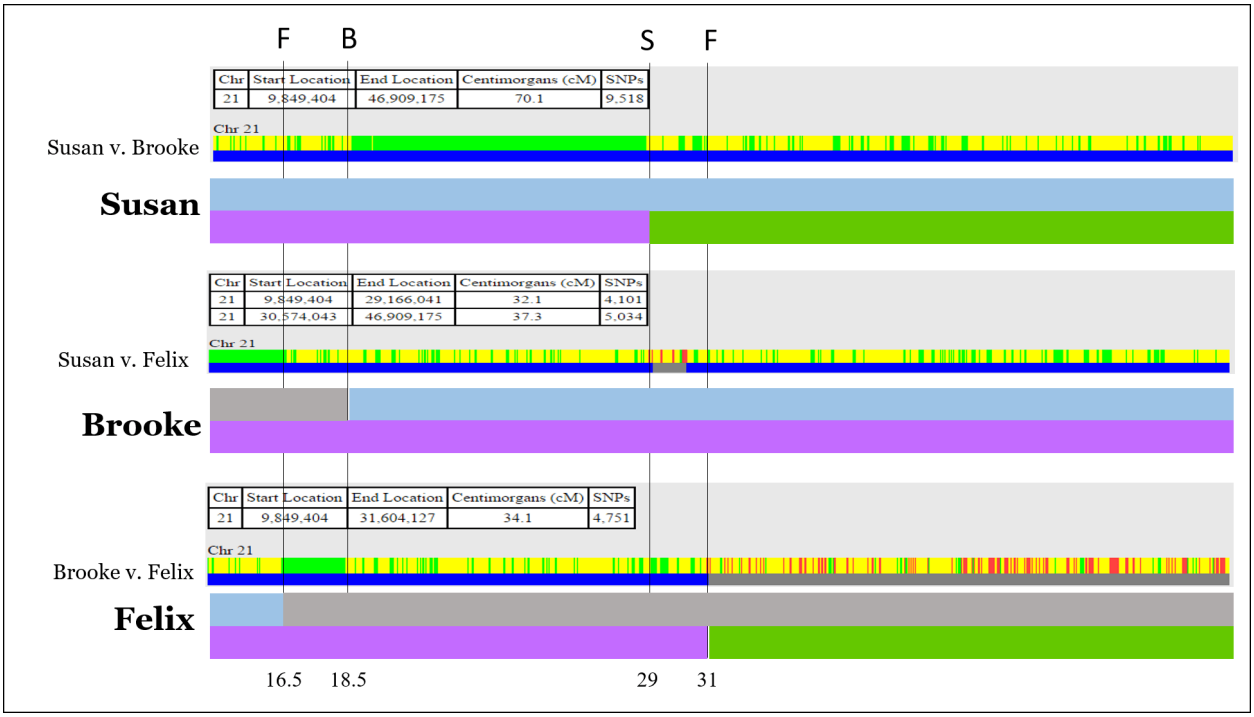
So we can fill that out for Felix:



We've now finished Felix's chromosome entirely!

We see that Susan and Felix share NO DNA on either chromosome at the region shown by the red box in the next figure:

So we can fill in that region for Susan, using the colors opposite Felix at that region (Felix has gray and purple, so Susan must have blue and green), and we can extend that all the way to the end of the chromosome:



We've completed all three chromosomes!

Now, note that I intentionally selected a chromosome that I could complete fairly easily. It is VERY common to not be able to solve an entire chromosome. Sometimes it can be solved by bringing in another sibling or cousin matches, but even then sometimes it cannot be completely solved. And that's fine, you can work with a chromosome map that isn't completely solved.

I know that blue and gray are one side of the family, and that purple and green are the other side of the family. But I don't know, yet, which is which. In the next part, we'll use cousin matches to deduce which combination is maternal and which is paternal, and hopefully we'll determine which segments belonged to which grandparent.

NOTES/TIPS:

- VERY IMPORTANT! Each chromosome is its own individual project. The color combinations can be completely opposite from one chromosome to the next. Here, where blue/gray might mean maternal, they might mean paternal in the next chromosome. Once I determine the source, if I can, I'll change around the colors to be consistent from chromosome to chromosome. But think of each chromosome

as its own project, do NOT carry other preconceptions about colors to the next chromosome!

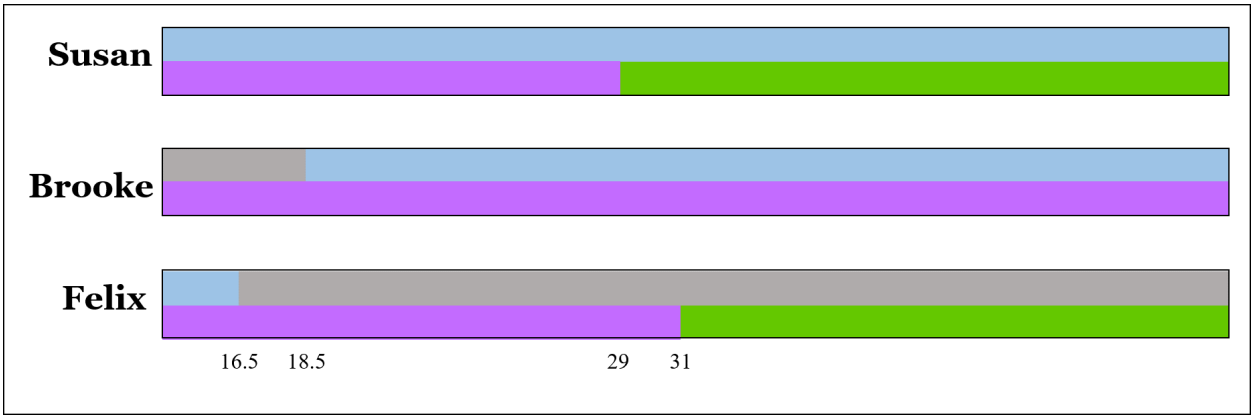
- We can already make some interesting conclusions here:
  - For example, we see that the entire blue, gray, and purple chromosomes are represented in these three siblings. The green chromosome is not, however. So if there are no other siblings, that green chromosome from 0 Mb to 29 Mb has been lost to all descendants.
  - As another example, only Felix inherited DNA from all four grandparents on chromosome 21. Brooke and Susan only received DNA from three grandparents, and mostly from the purple and blue grandparents!

- Eventually, I would like to be able to tell a computer program to recreate the blue and gray chromosomes in a new file in their entirety for the first parent (either mother or father), and to recreate the entire purple and partial green chromosomes in a new file for the other parent (the other of the mother or father).

Visual Phasing: An Example (Part 3 of 5) – November 25, 2016

Originally Appeared at:
http://thegeneticgenealogist.com/2016/11/25/visual-phasing-an-example-part-3-of-5/

In "Visual Phasing: An Example (Part 1)," we identified and labeled all of the recombination points in the three siblings, Susan, Brooke, and Felix. Then, in Part 2 of the series, we used the identified and labeled recombination points to assign segments of DNA to the four grandparents (the blue grandparent and grey grandparent pair, and the purple grandparent and green grandparent pair):

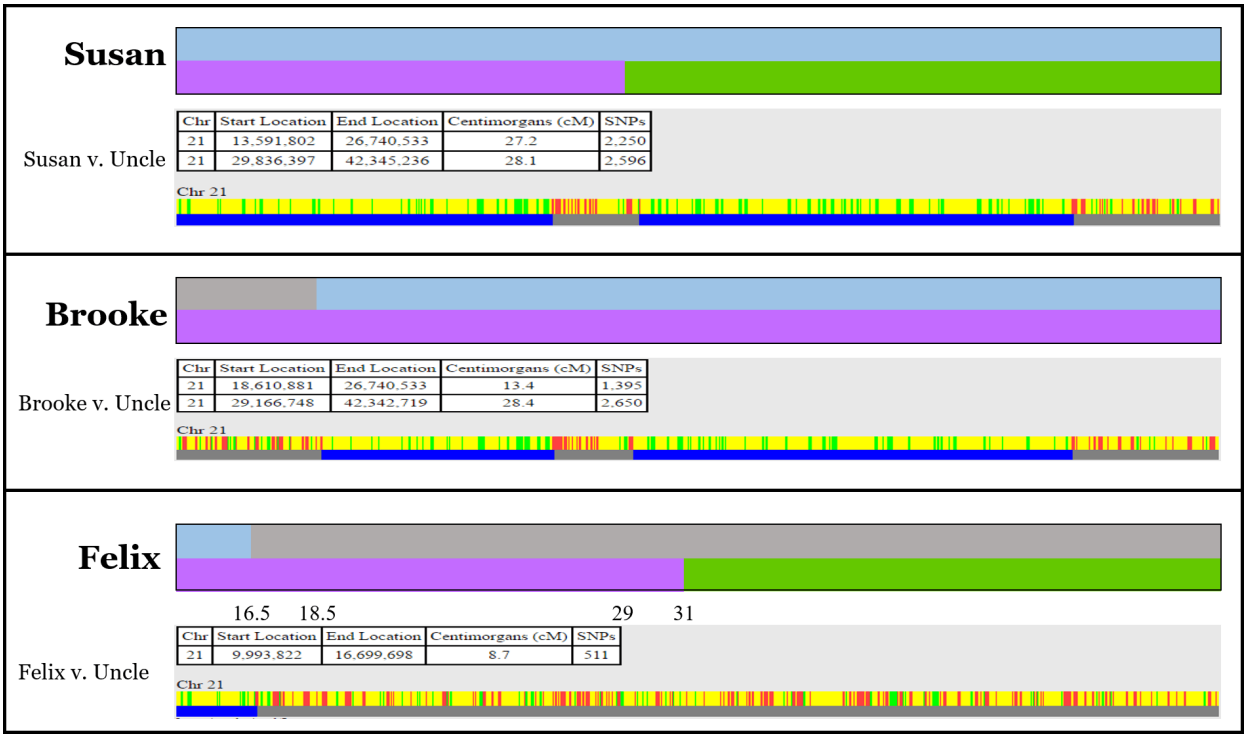


But without more information, we don't know to which grandparent each segment belongs.

Today, we'll use cousin matching (defining "cousin" to mean anyone other than the siblings) to first identify the maternal pair of chromosomes, and the paternal pair of chromosomes. Hopefully, in the process, we'll be able to assign the segments to each of the four grandparents, rather than just maternal/paternal.

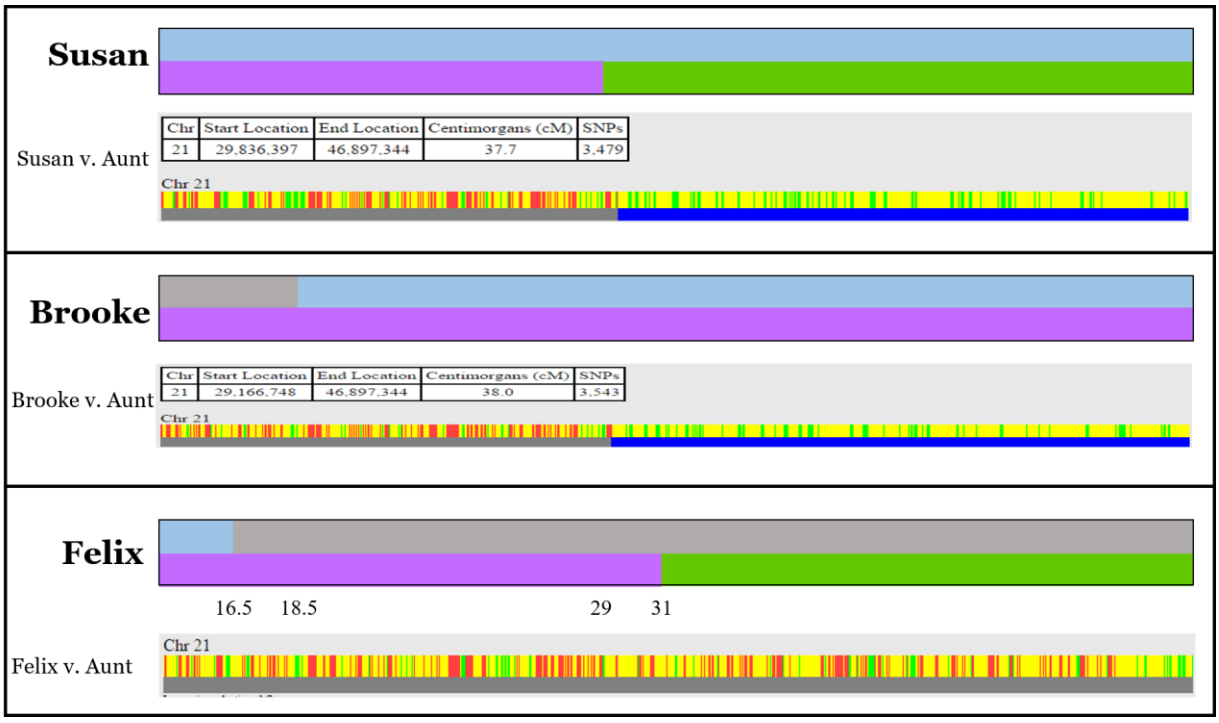
In this example, I have a maternal aunt and a maternal uncle to whom I can compare the three siblings. This won't allow me to identify particular grandparents, but it will allow me to determine maternal versus paternal.

In this first comparison, we use a maternal uncle:



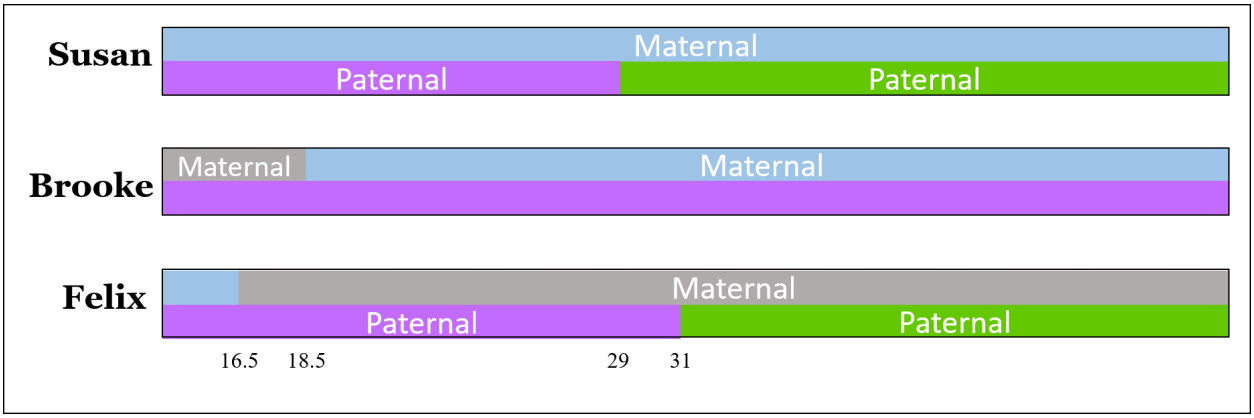
Looking at Felix's comparison to the maternal uncle, we see that the uncle must be matching the blue segment at the left end of chromosome 21; if he were matching the purple, he would also match Brooke at that region. So this suggests that the blue and gray chromosomes are maternal.

In this next comparison, we'll use a maternal aunt:

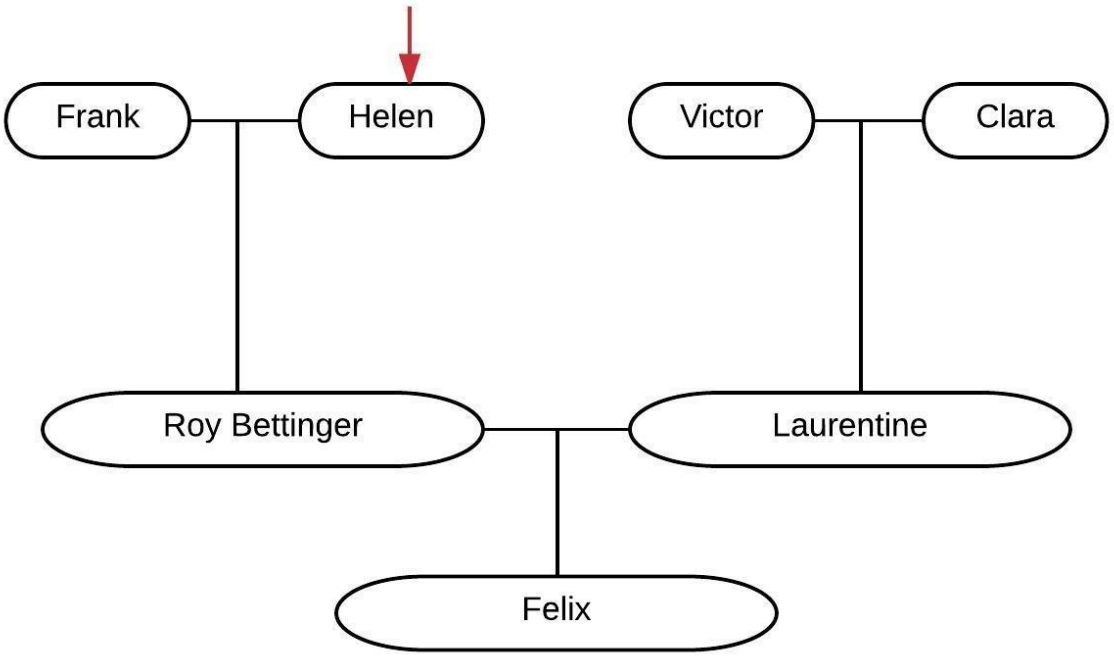


Looking at Susan's comparison to the maternal aunt, we see that the aunt must be matching the blue segment at the right half of chromosome 21; if she were matching the green segment, then she would also match Felix at that region. This again suggests that the blue and gray chromosomes are maternal.

So both comparisons suggest that the blue and gray chromosomes are maternal, and thus that the green and purple chromosomes are paternal:



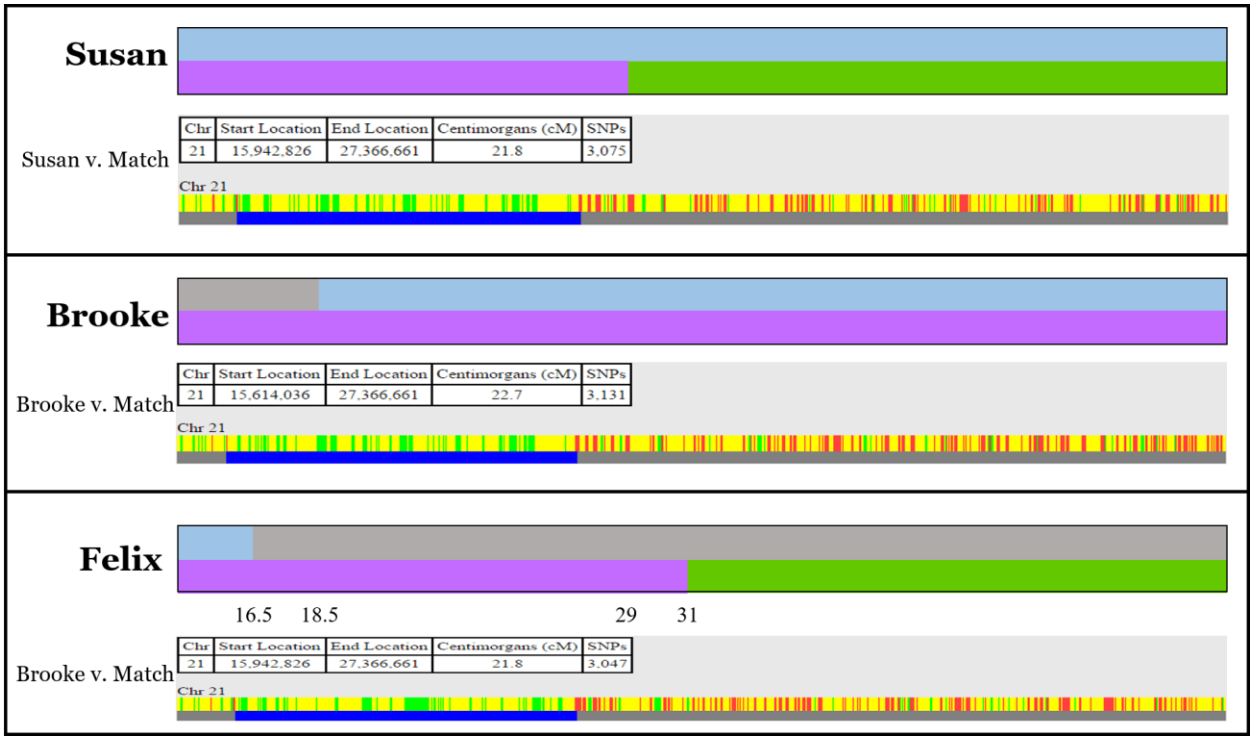
Let's see if we can break down "maternal" and "paternal" into each of the four grandparents. Here is the family tree for Brooke, Susan, and Felix:



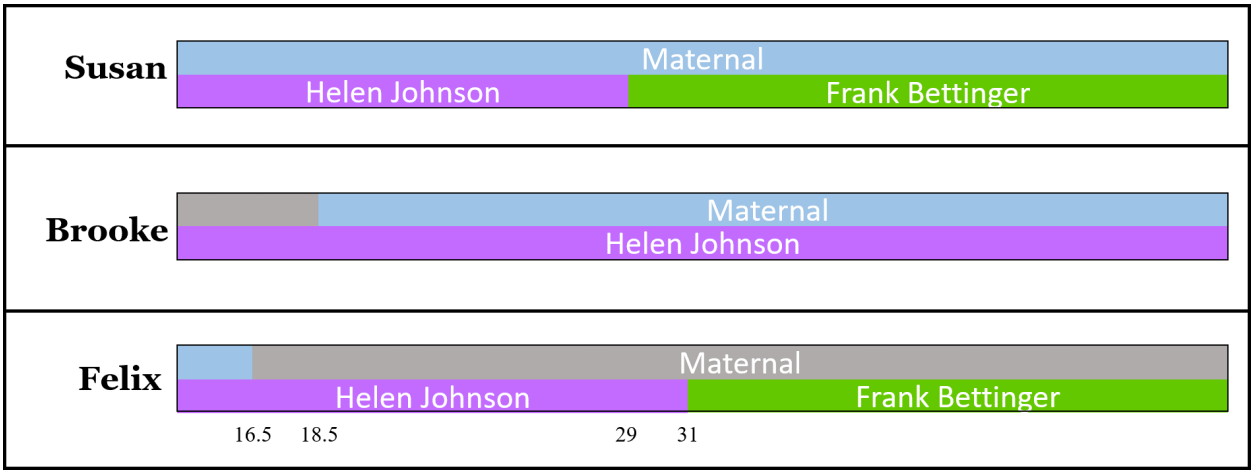
We know that the blue and gray chromosomes are from Victor and Clara, and that the green and purple chromosomes are from Frank and Helen.

I have a handful of matches that I know are through Helen. Thus, if I can use that match to figure out whether Helen is the green or purple grandparent, I will also be able to determine Frank's segments as well (by process of elimination).

Here is the comparison of Brooke, Susan, and Felix to the match:



The match is clearly matching the purple segment, so Helen Johnson is the purple grandparent and Frank Bettinger is the green grandparent.



I don't have a match that clearly identifies the source of the blue versus gray chromosomes, so that will require either targeted testing or waiting for a random match.

This is where the process of visual phasing ends, although we'll want to break down the maternal chromosomes in the future, if we can. Now, for example, when a new match comes in that matches the right half of chromosome 21 with Susan and Felix, we know that the match is via Frank Bettinger, narrowing the search down to 25% from 100% of the family tree. Or, if someone matches only Brooke on the right half of chromosome 21, the match has to be through Helen Johnson.

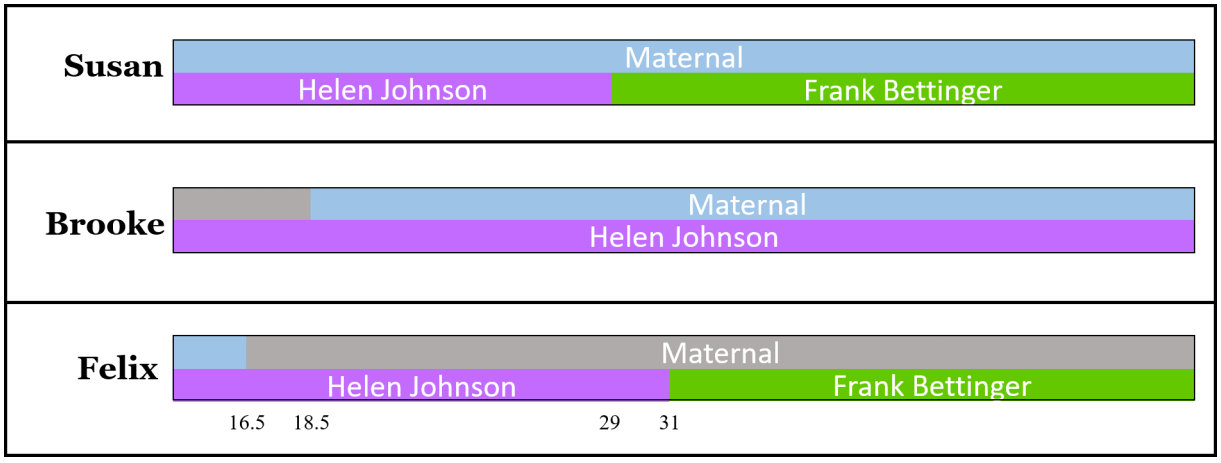
In Part 4 of the series, we'll use these three phased and labeled chromosomes, which belong to my father and two aunts, to characterize my own paternal chromosome.

Visual Phasing: An Example (Part 4 of 5) – November 26, 2016

Originally Appeared at:
http://thegeneticgenealogist.com/2016/11/26/visual-phasing-an-example-part-4-of-5/

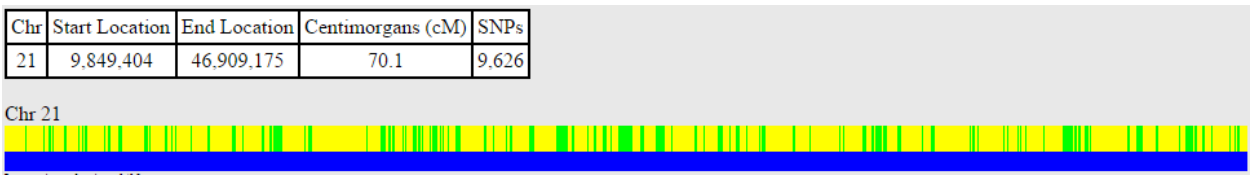
In "Visual Phasing: An Example (Part 1)," we identified and labeled all of the recombination points in the three siblings, Susan, Brooke, and Felix. Then, in Part 2 of the series, we used the identified and labeled recombination Visual Phasing: An Example (Part 4 of 5)

In "Visual Phasing: An Example (Part 1)," we identified and labeled all of the recombination points in the three siblings, Susan, Brooke, and Felix. Then, in Part 2 of the series, we used the identified and labeled recombination points to assign segments of DNA to the four grandparents (the blue grandparent and grey grandparent pair, and the purple grandparent and green grandparent pair). In Part 3 of the series, we used cousin matching to identify the grandparental source of the chromosomal segments. After Part 3, we had the following for Brooke, Susan, and Felix:



Since Felix is my father and Susan and Brooke are my aunts, I might be able to compare my DNA to my aunts in order to identify which of the segments I obtained from my father.

Of course, when I compare my father to myself, I get the following:

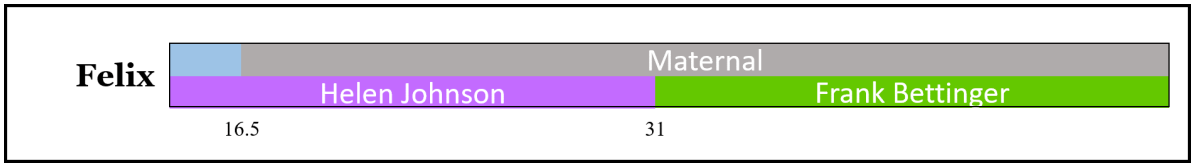


Chr	Start Location	End Location	Centimorgans (cM)	SNPs
21	9,849,404	46,909,175	70.1	9,626

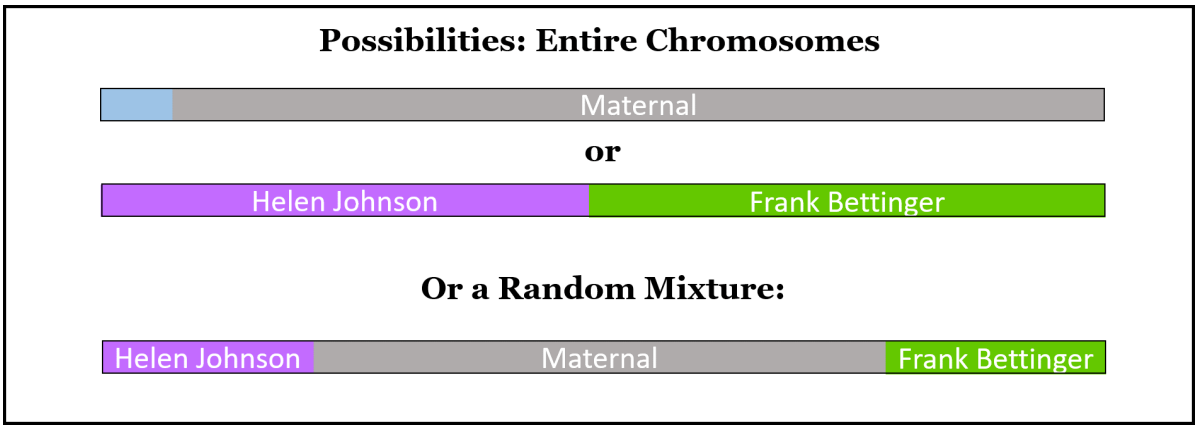
Chr 21

So that comparison isn't going to be helpful at all.

What I do know, however, is that this is the ENTIRE universe of DNA that I could possibly obtain from my father on chromosome 21:

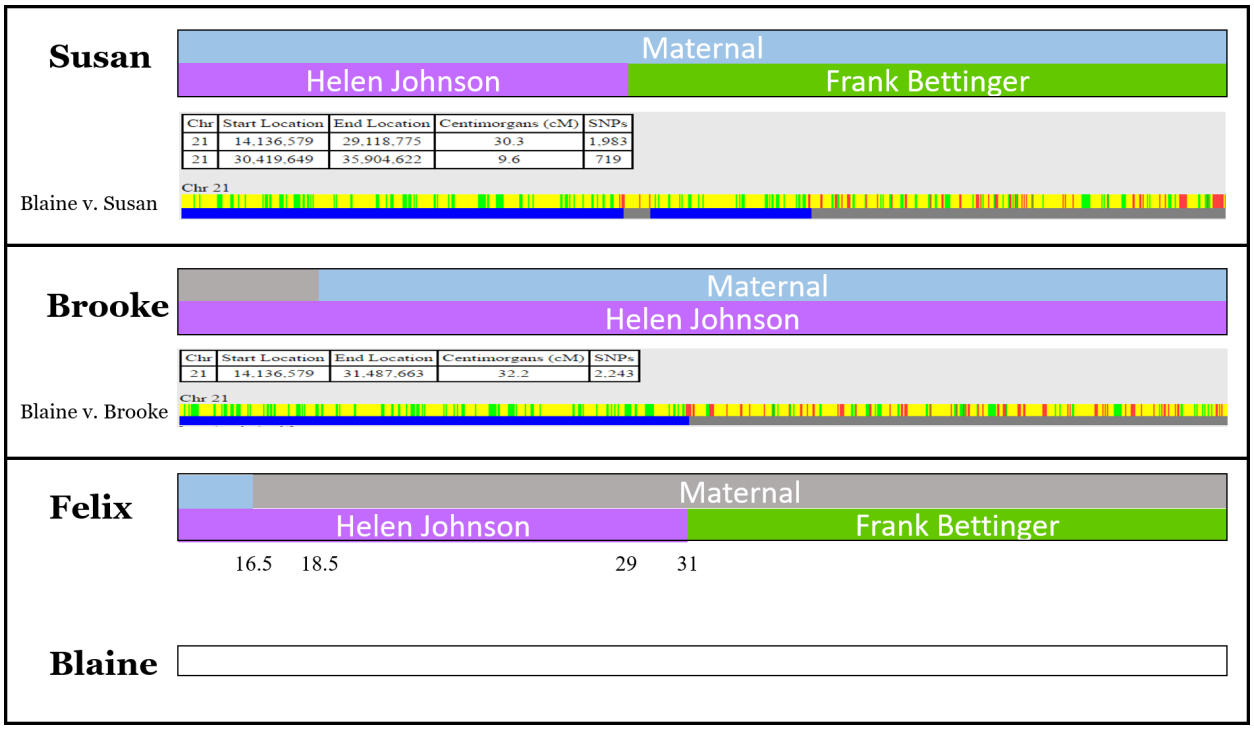


So some possible chromosomes that I could have inherited include full copies of either the maternal or paternal chromosomes (meaning no recombination took place), or a recombined mixture of the maternal AND paternal chromosomes:



The "Random Mixture" chromosome is just one of many different examples I could have created. As we'll see below, I did indeed inherit a random mixture chromosome, but not the one shown above.

I'll begin this process by comparing my chromosome 21 to my aunts' chromosome 21:



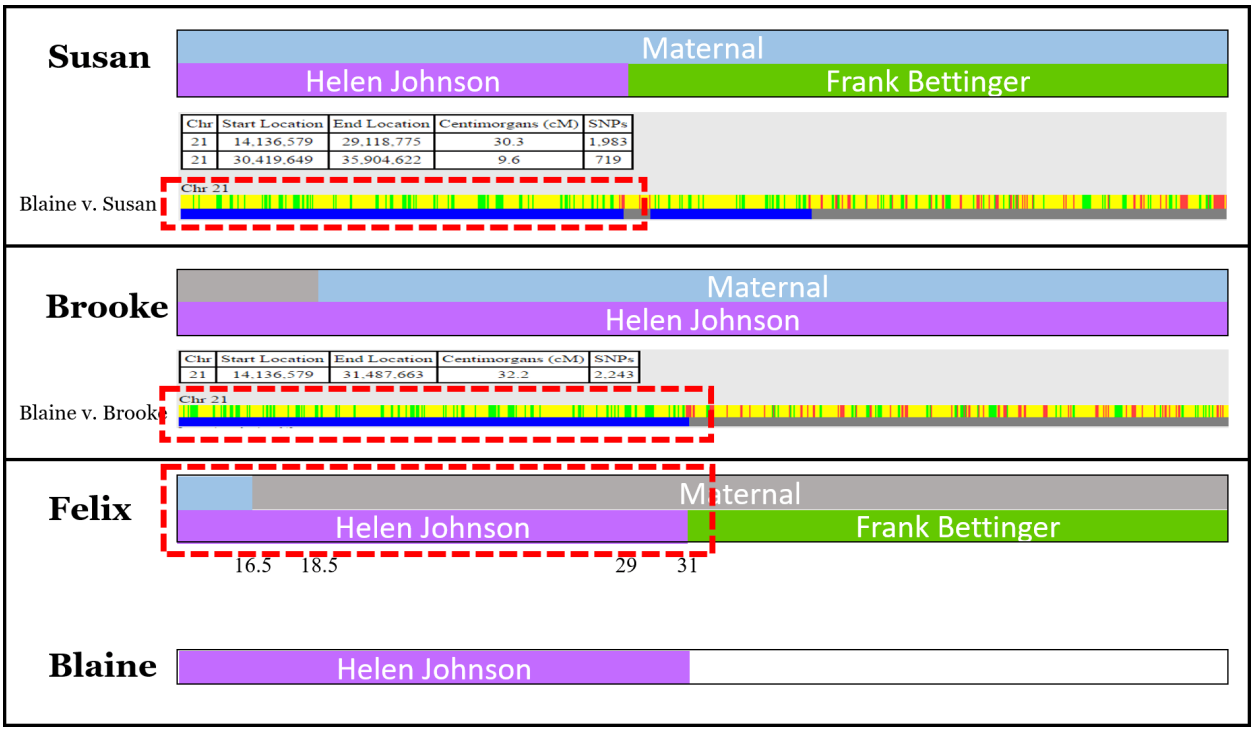
So I share one or more great segments with both aunts on chromosome 21.

Let's focus on the left end of the chromosome where I share segments with both aunts. Remember that Felix's chromosome is the possible universe from which I can pull DNA. That means that for most of the segment shown in the red boxes, I could NOT have inherited the maternal copy of the chromosome (blue), since my father didn't have that portion after 16.5 Mb.

And if you compare the start to 16.5 Mb, you see that I share that segment with Brooke. Since Brooke and Felix only share purple at that region, I must have inherited purple as well.

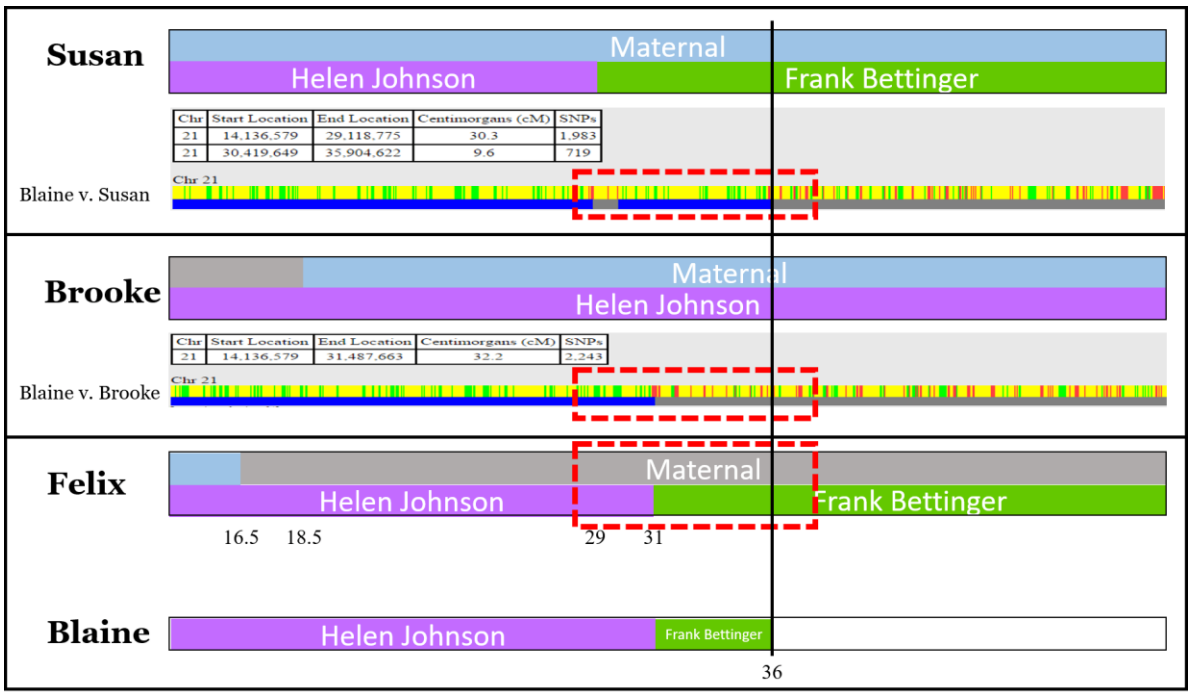
From about 29 Mb to about 31 Mb, I share DNA entirely with Brooke (meaning it must be purple), since Felix doesn't share the blue grandparent with Brooke at that region.

As a result, on my copy of chromosome 21, I've filled out purple from the start of the chromosome to position 31 Mb:



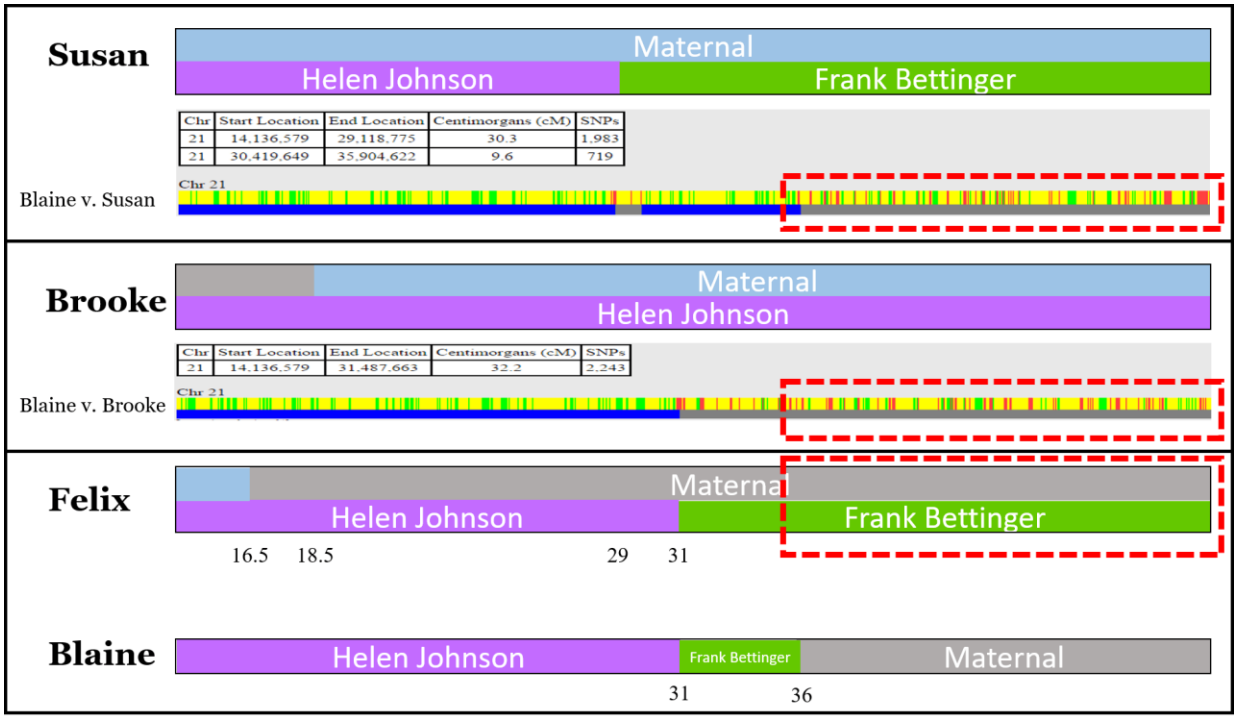
Let's focus on the next segment I share with aunt Susan, from about 30-31 Mb to 36 Mb (in the dotted red box). Remember again that Felix's chromosome is the possible universe from which I can pull DNA. That means that for the segment shown in the red boxes from 30/31 to 36 Mb, it can either be green or gray.

Susan doesn't have gray DNA at that region, so it must be green. That makes sense, since Brooke doesn't match me at that region, and she doesn't have any green DNA.

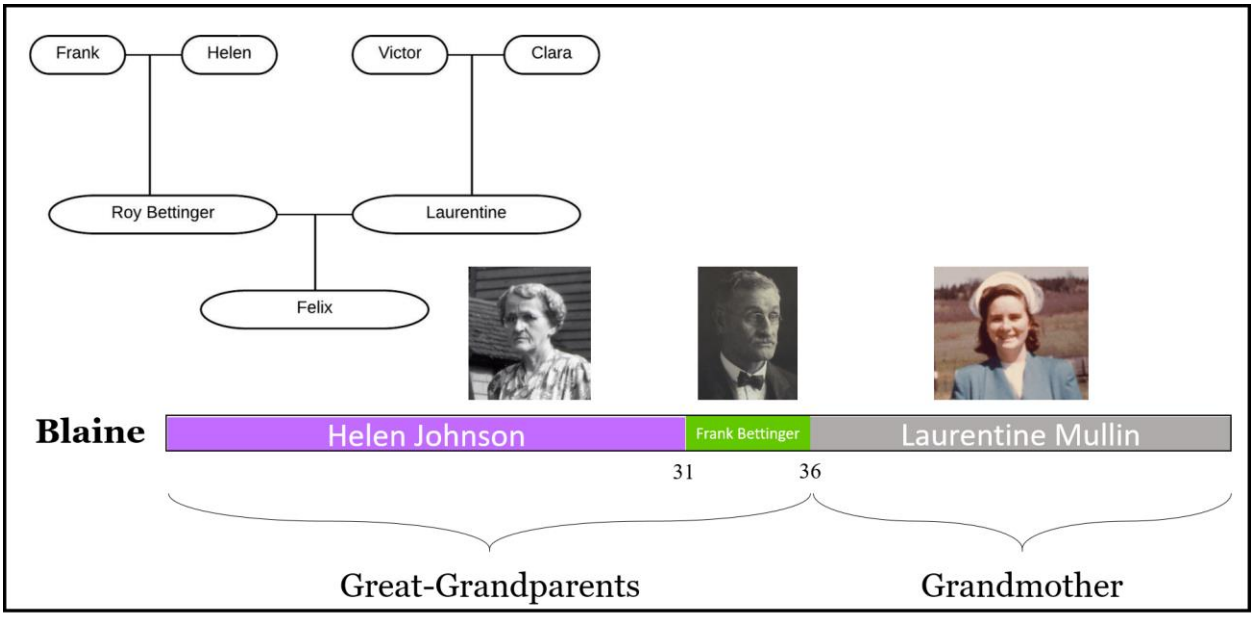


But you see that at 36 Mb, something changes. After that, even though both Felix and Susan have green DNA from 36 Mb to the end, I don't share any more DNA with Susan.

That means that there must have been recombination at that point, switching from the paternal chromosome to the maternal chromosome (gray). Again that makes sense, as neither Susan nor Brooke have any gray DNA at that region, and I don't share any DNA with them:



And this is one of the many reasons why I love Visual Phasing. Without having tested ANY of my grandparents or great-grandparents, I've been able to map my entire chromosome 21 back to them:



This is fascinating in its own right, tracking segments of DNA through both time and space. However, it also has some very important implications for matching! If a new match comes along that shares DNA with me on the paternal copy of chromosome 21, I know exactly which line of my family tree to search for a shared ancestor. In the case of Laurentine Mullin, I can narrow my tree down to 25%, and in the case of the rest of the chromosome, I can narrow the search down to 12.5% of my entire family tree!

In Part 5 of the series, we'll look at ways to use the mapped chromosome 21 to explore new matches and other techniques.

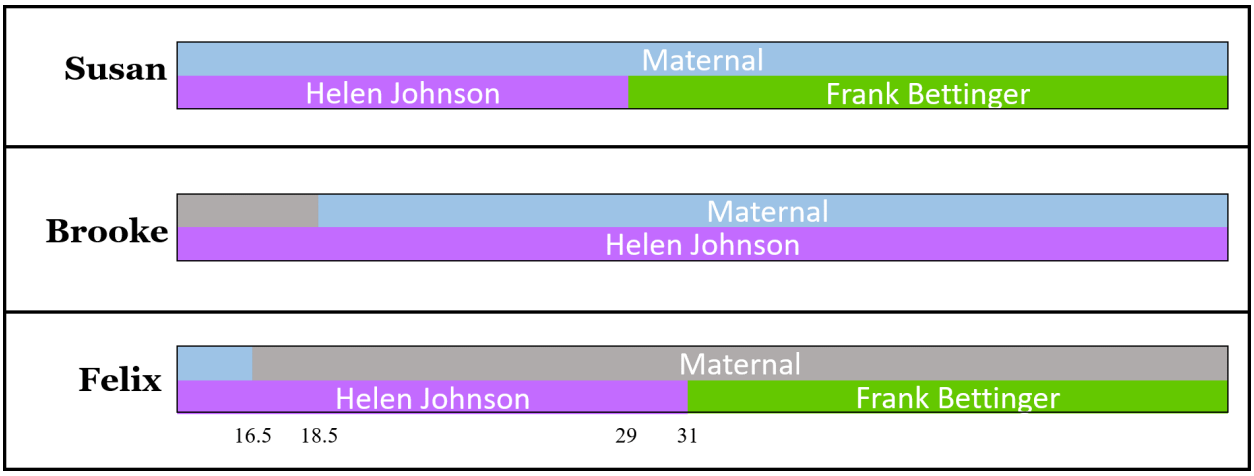
NOTE: This process does NOT always work, when the sharing with the aunts does not line up to allow you do deduce which segments you obtained. Here, for example, is the entire map of chromosome 17 that I'm able to do at the current time, using the process described above:

Visual Phasing: An Example (Part 5 of 5) – November 27, 2016

Originally Appeared at:
http://thegeneticgenealogist.com/2016/11/27/visual-phasing-an-example-part-5-of-5/

In "Visual Phasing: An Example (Part 1)," we identified and labeled all of the recombination points in the three siblings, Susan, Brooke, and Felix. Then, in Part 2 of the series, we used the identified and labeled recombination points to assign segments of DNA to the four grandparents (the blue grandparent and grey grandparent pair, and the purple grandparent and green grandparent pair). In Part 3 of the series, we used cousin matching to identify the grandparental source of the chromosomal segments. And finally, in Part 4 of the series, we characterized my paternal chromosome.

After Part 3, we had the following for Brooke, Susan, and Felix:

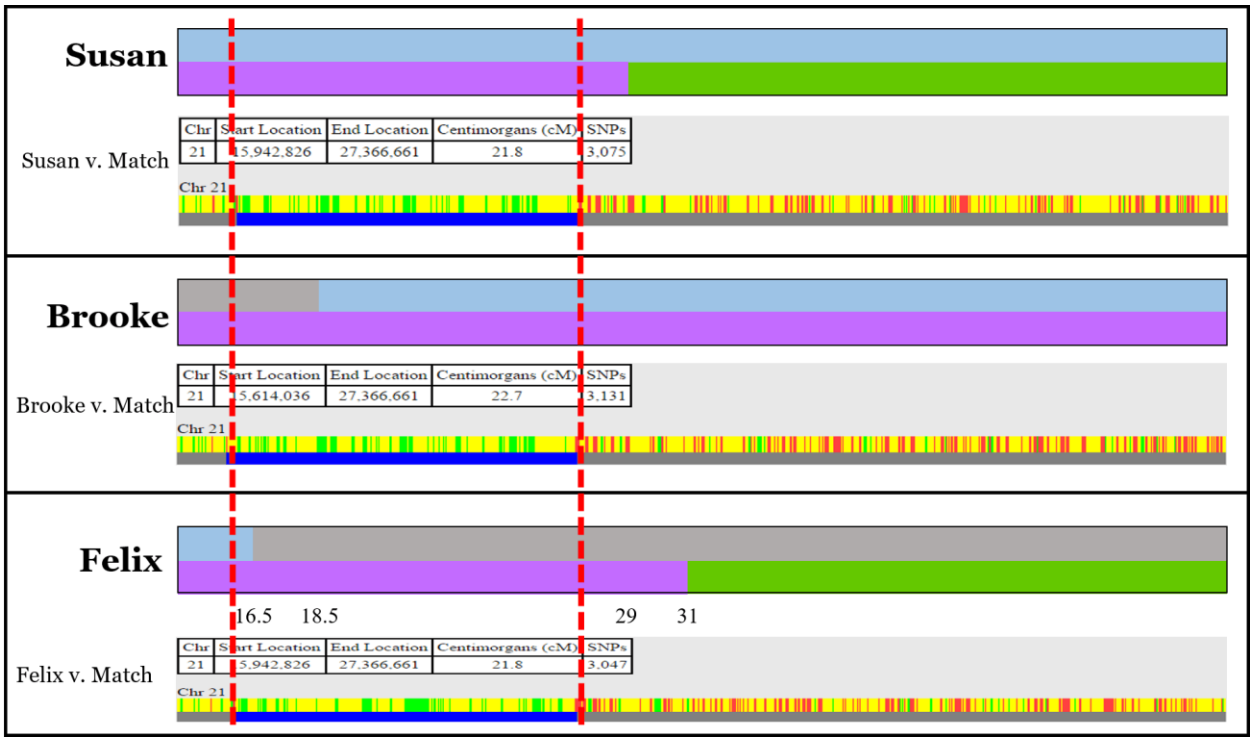


Now that we have this information, let's see if we can use that to explore new matches with Brooke, Susan, and Felix.

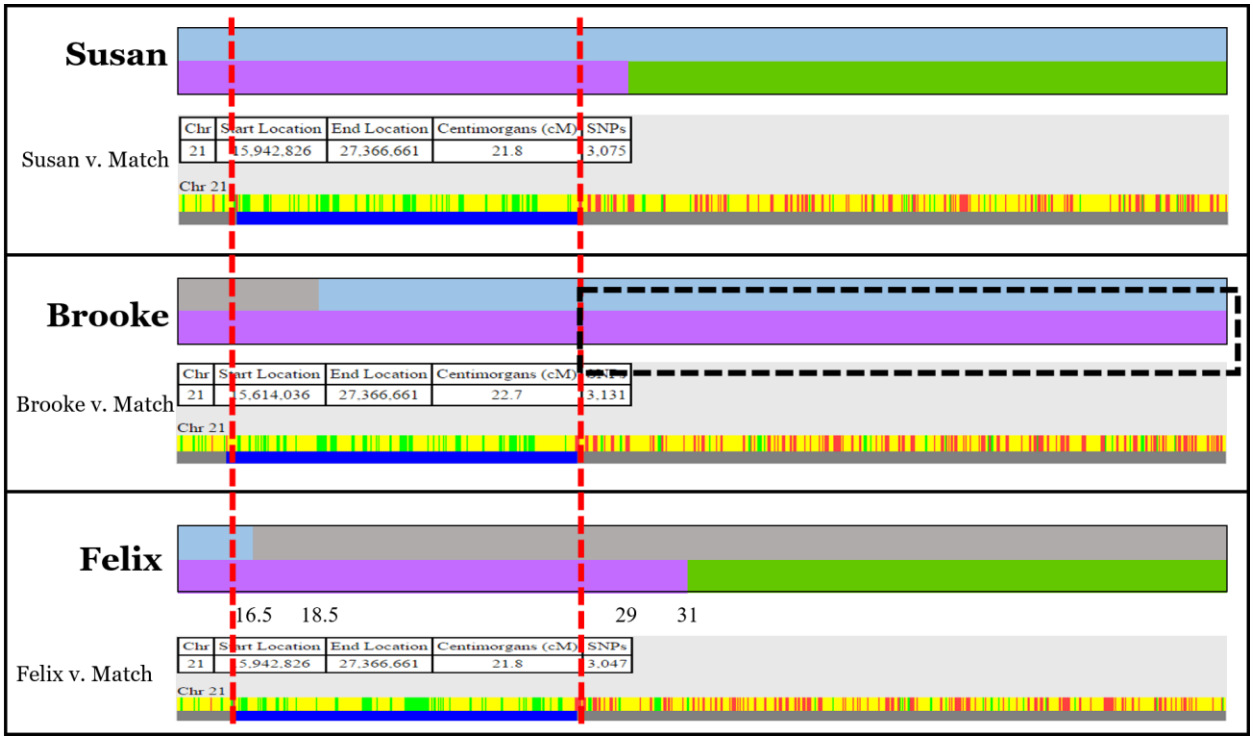
I'm particularly interested in the DNA from Helen Johnson, my great-grandmother (and grandmother of the three siblings), who was born and adopted in 1889. I have used DNA to help identify Helen Johnson's biological mother, but I do not yet know who her biological father was.

If I can identify matches to Brooke, Susan, and Felix that are definitively matching through Helen Johnson, they will help me identify Helen's biological father.

Some of this, of course, I could have done previously. I knew at least one segment that came from Helen Johnson to each of the three siblings, the segment they shared with the match to Helen's mother's family. Anyone that matches Brooke, Susan, AND Felix at the region shared with this match (between the dotted red lines), is shared via Helen Johnson's mother:

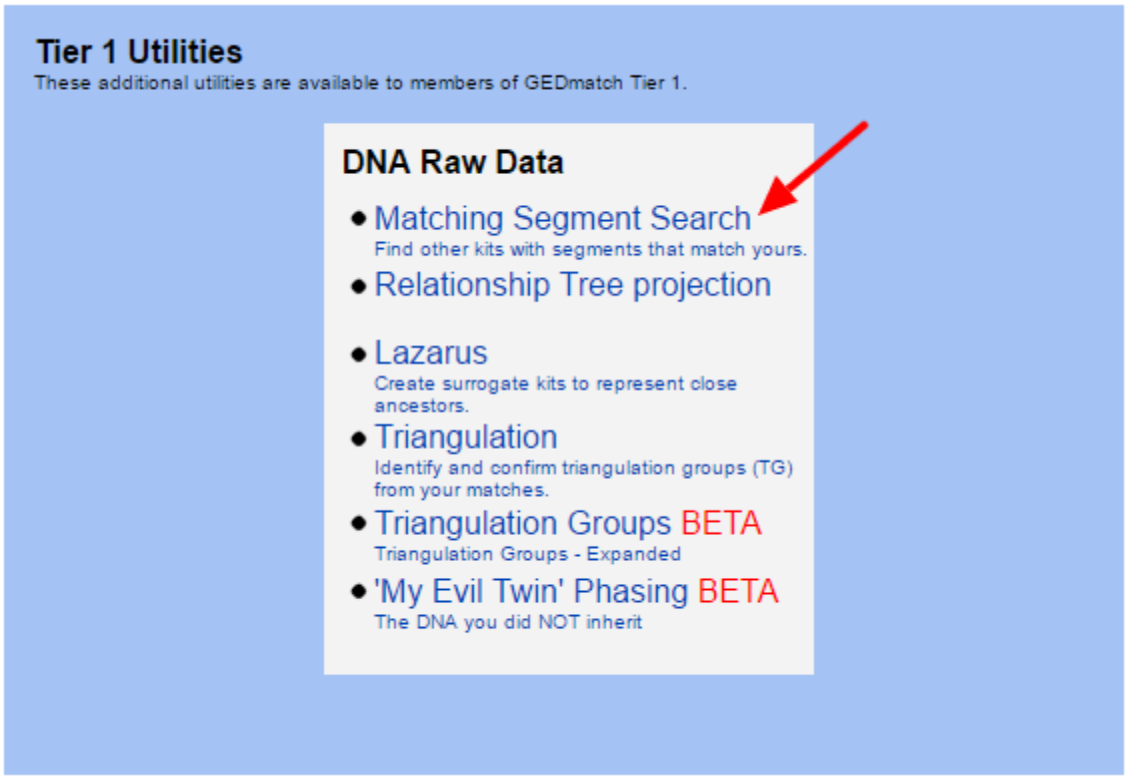


But before visual phasing, I was limited to that segment. I had no idea how far that segment extended outward on each of the three siblings. Solely from visual phasing, I've learned that Brooke has an entire chromosome from Helen Johnson. The area shown in the dotted black lines is now an area of interest:



If I identify matches from 31 Mb to the end of chromosome 21 that are shared by Brooke and neither Susan nor Felix, I will have identified matches that share ancestry with Helen Johnson.

There are several ways to do this. For example, I can use the Matching Segment Search at GEDmatch to find segments on chromosome 21 from 31 Mb to the end, shared by Brooke but neither Susan nor Felix. I'll run the Matching Segment Search for each of them, take the results from chromosome 21 (31 Mb to end) and combine them into a spreadsheet.



This will allow me to identify matches shared by Brooke but neither Susan nor Felix. The following is a screenshot of the spreadsheet, showing two segments in rows 8 and 9 that are shared with Brooke but neither Susan nor Felix:

	A	B	C	D	E	F	G	H	I	J
1	Felix	A0	21	30,809,529	36,740,941	11.6	1,599	*	F	L t.net
2	Felix	A1	21	31,384,852	35,271,495	7	759	K	F	k
3	Felix	M8	21	31,411,628	36,270,350	9.4	1,061	*	M	a
4	Brooke	T7	21	32,136,224	42,236,178	25	3,290	A	M	lr om
5	Susan	T7	21	32,136,224	42,256,842	25.1	3,190	A	M	lr om
6	Brooke	M2	21	32,142,892	42,637,976	26.5	1,787	*	F	a
7	Susan	M2	21	32,142,892	42,637,976	26.5	1,762	*	F	a
8	Brooke	A80XXXX	21	32,421,702	39,041,800	13.2	1,887	Match Name #1	U	matchname1@gmail.com
9	Brooke	A89XXXX	21	32,449,069	38,757,156	12.4	1,140	Match Name #2	M	matchname2@gmail.com
10	Felix	A7	21	32,880,218	39,418,349	13	1,910	*	F	m
11	Felix	A1	21	33,165,007	37,746,525	9.2	1,301	*	F	D
12	Susan	A2	21	33,200,101	37,501,784	8.8	1,263	*	F	D

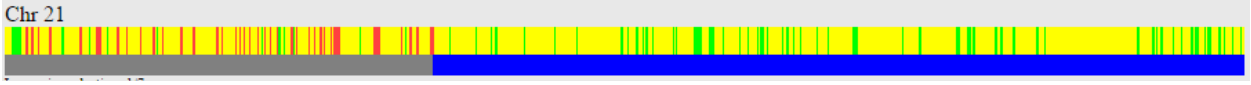
The segment shared with A80XXXX shows nicely in a One-to-One comparison:

Chr	Start Location	End Location	Centimorgans (cM)	SNPs
21	32,421,702	39,041,800	13.1	1,887

Unfortunately these are very small segments at 13.2 and 12.4 cM, but they illustrate the process. And I may find larger segments on other chromosomes.

Now, if A80XXXX and A89XXXX are indeed matches via Helen Johnson's DNA (and thus match Brooke on a single chromosome), then they MUST share that segment in common with each other. A One-to-One comparison of the two kits at GEDmatch confirms that:

Chr	Start Location	End Location	Centimorgans (cM)	SNPs
21	26,321,396	46,897,344	43.8	4,120



I can contact the owners of kits A80XXXX and A89XXXX, and I can review their trees to find people who were in the right place at the right time.

Although I've focused on Helen Johnson's DNA, I can also use the results for any of the other grandparents. For example, the spreadsheet above shows a very promising group of matches shared by Susan and Brooke but not with Felix. They share a much larger 25 cM segment, which must be shared via their mother and the blue grandparent (whichever of their maternal grandparents this turns out to be).

This is just one way to use the information from visual phasing!

The Future

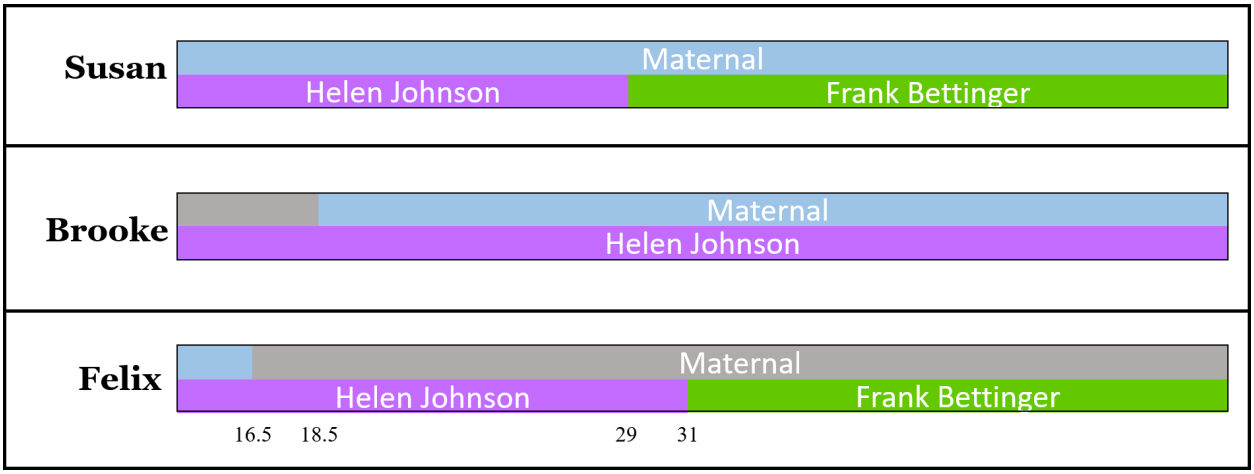
I envision two major developments with Visual Phasing, although the uses of this output is limited only by our creativity.

1. Visual Phasing Automation. Visual Phasing will be partially or entirely automated. The data from three siblings could be fed into an algorithm that determines recombination points and fills in the chromosomes. Then, you could feed known matches into the algorithm to have it automatically identify which segments came from which grandparent.

2. Grandparent Reconstruction. The data from Visual Phasing, particularly from people lucky enough to have multiple siblings (4 or more) tested, could be used to recreate incredibly complete and accurate reconstructions of our grandparents. For example, we could extract the Visually Phased segments from one or more of the grandparents to create a grandparent kit. And since we're using entire portions of chromosomes beyond

matching segments, this will allow for greater reconstruction than tools like Lazarus which rely only on matching segments.

Using our visually phased chromosome 21 as an example, from just these 3 siblings we have an ENTIRE copy of chromosome 21 for Helen Johnson and both maternal grandparents (blue grandparent and gray grandparent). We have roughly half of Frank Bettinger's chromosome 21:



Final Words

I hope you've enjoyed this series on Visual Phasing. I'd like to thank Kathy Johnston and Randy Whited for sharing their knowledge with all of us. I'd also like to thank others who have blogged or posted about their experiences with Visual Phasing. I think Visual Phasing is a terrific example of how existing data can give rise to new tools and new information!

Best of luck with your own Visual Phasing project!