# Visual Phasing

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## DNA Visual Phasing

- Visual phasing is a process by which the DNA of siblings is compared to determine all of the segments each received from all four of their grandparents
- Visual phasing is best done by comparing the DNA of three full siblings, but can be done with more or less
- Visual phasing can be done with half-siblings but only for the half that is in common
- Each person gets exactly half their DNA from mom and dad, but beyond that is random
- At any given point you have DNA from only two of your grandparents
- The places where your DNA switches from one grandparent to another are called recombination points
- Maternal DNA has about 70% more recombination points than paternal DNA

### Benefits of Visual Phasing

- Visual phasing allows you to determine which of your four grandparents a genetic match is related to
- This builds confidence in proof arguments and helps you to focus in on those whose DNA can be used to solve your brick walls
- Its really fun

### Phasing Platfoms

- Visual phasing requires comparing the sibling's DNA in a chromosome browser
- GedMatch is strongly preferred, 23andme will work ok if necessary
- FamilyTreeDNA and MyHeritage's chromosome browsers will not work because they do not distinguish FIR regions
- You will need software where the chromosomes can be compared side by side
- Microsoft Excel and PowerPoint are great options
- My case studies will use GedMatch and Excel

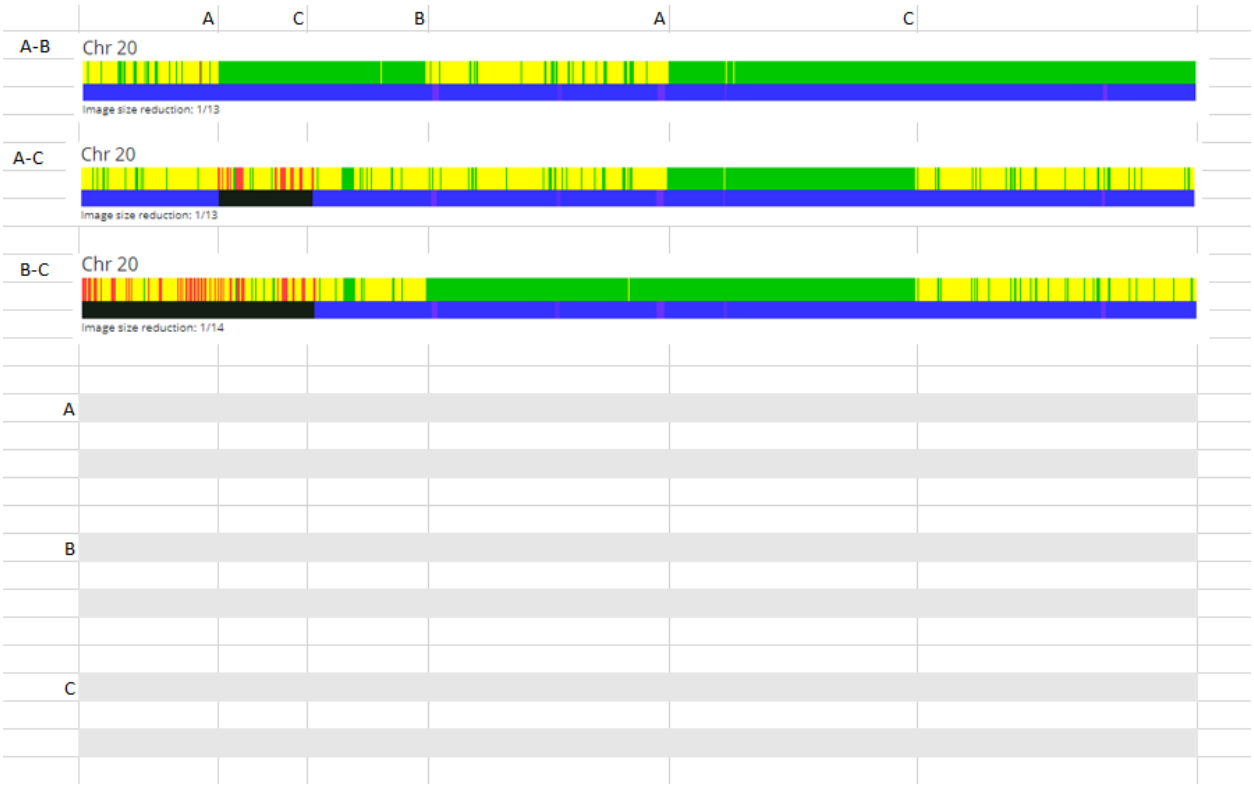
#### GedMatch's Chromosome Browser

- When comparing full siblings in GedMatch each chromosome will have segments highlighted with three colors. The three colors are green (full match), yellow (half match), and red (no match). The blue bar underneath simply highlights matches to make them easier to see.

- The places where the siblings inherited DNA from the same grandparents on both sides are gree, the places where the siblings inherited from the same grandparent on one side but opposite grandparents on the other side are yellow and the places where they inherited from opposite grandparents on both sides are red.
- Long green segments are usually only seen in full siblings unless there is endogamy. Other relations will only have yellow and red.
- Yellow often has lots of green static in it, and red often has lots of yellow static. Do not worry about that. Worry about the main colors.
- Green sometimes has thin yellow lines through it. Often these are because of incorrectly reported values along the genome. DNA tests are over 99% accurate but there are about 16 incorrect values per test. If you have several yellow lines next to each other it could actually be a small yellow segment sandwiched between two green segments. On the One-One Autosomal DNA comparison you can see the DNA more clearly by checking the “Full Resolution” box and then scrolling to the part in question. In the image further down in this handout, the green segments have some thin yellow bars. In this case they are of no concern.

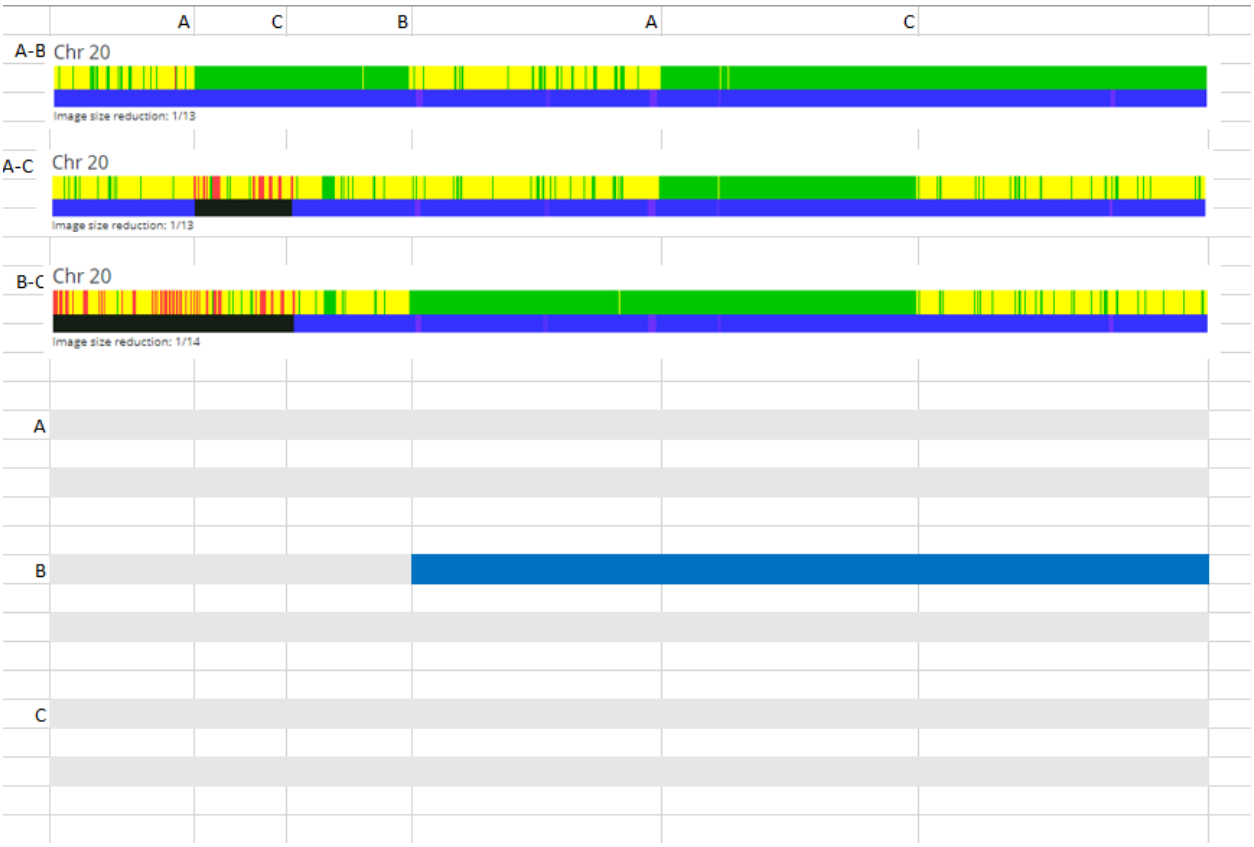
## The Set Up

- Compare siblings A and B, A and C, and B and C in GedMatch’s One-to-one Autosomal DNA comparison.
- Leave the default settings the same. If you do change something I like to put a 3 in the box next to the “Minimum segment cM size
- to be included in total:” box. Many segments smaller than 7 cM are false positive and I end up excluding them but it can be nice to at least have the small matches highlighted for the times they are real
- Each chromosome must be done individually. Take screenshots of one of the chromosomes and put the comparisons in excel. Line up the chromosomes as best you can and try to have them all the same size
- Click and drag the boundaries of the cell columns and line them up with the recombination points. The recombination points are where the chromosomes switch from one color to another.
- Now you need to identify which sibling owned each recombination. This is usually the sibling who switches from one color to another in both of his comparisons. For example, in the image below at the first point siblings A and B go from yellow to green in the same spot that siblings A and C go from yellow to red. Therefore, sibling A probably owns that recombination point.
- Put initials of the siblings or something to help you remember who owns each one above each recombination point.
- Next I recommend coloring in six bars below the chromosomes with a neutral color. This is where you will fill in the solution. See the image below.
- Finally select four colors. It doesn’t matter what colors you choose put pick two for the top chromosomes for each child and two for the bottom. Be consistent for all three siblings.

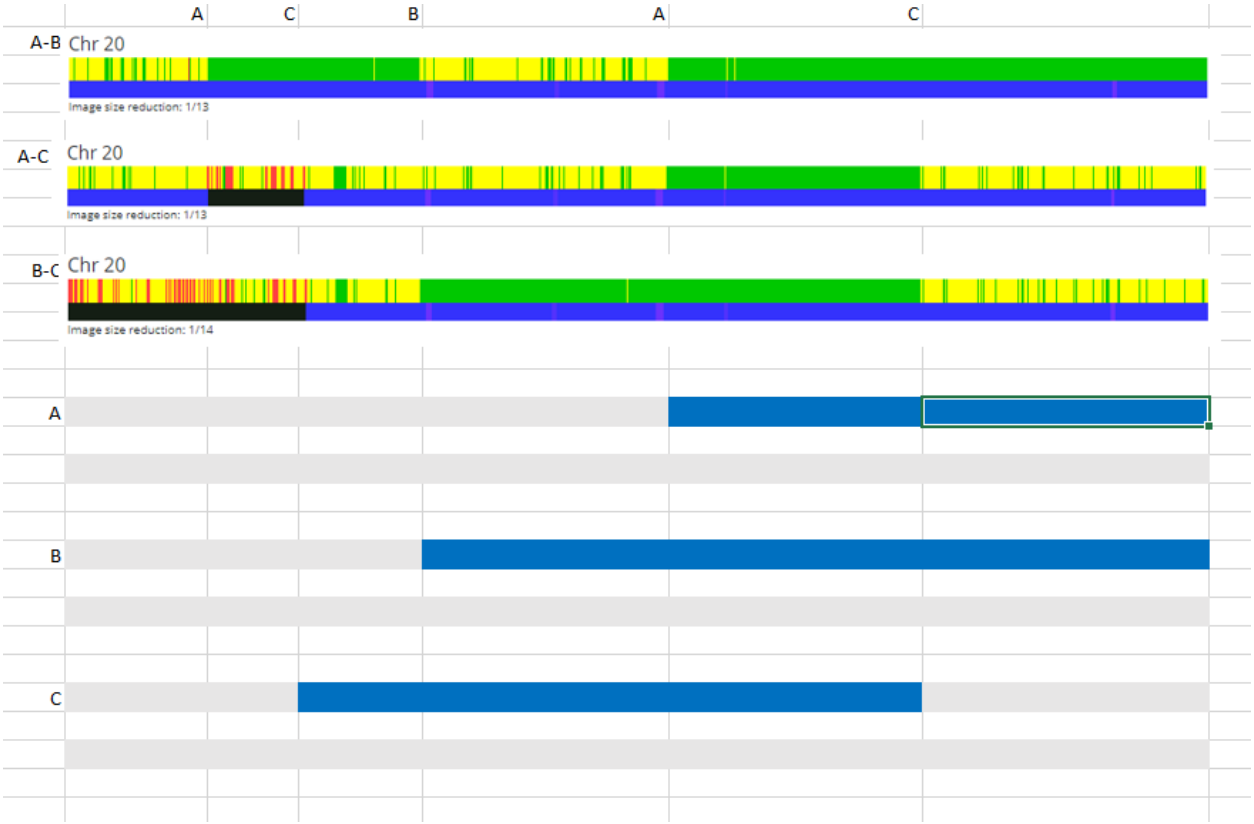



**Solving It**

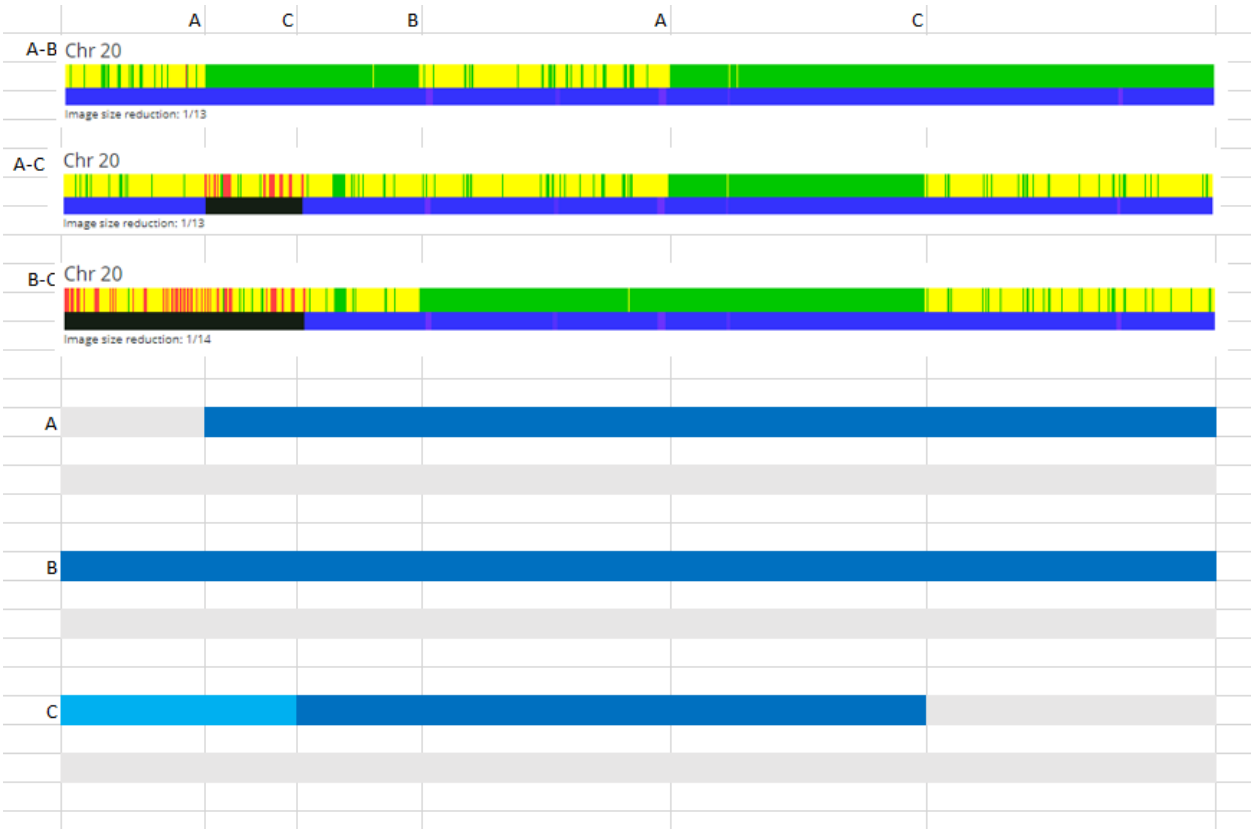
- Every book I have read or video I have seen on visual phasing says to start filling in the colors and then use cousins and other relations to determine which chromosomes in your solution are paternal and which are maternal. I like to pretermine the top to be the paternal and the bottom the maternal and I have standard colors I like to use for each grandparent. Both ways work, but the second way will only work if you have a cousin or other relation matching at least one of the siblings on the desired chromosome to compare against before you fill in anything. Therefore I will demonstrate the traditional method because it does not depend as much as on having the right cousins in the database.
- Pick a spot on the chromosome and start filling it in for one of the siblings at a spot of your choosing. Then continue to fill in to the left and right until you either come to a recombination point for that sibling or the end of the chromosome. Here is an example:



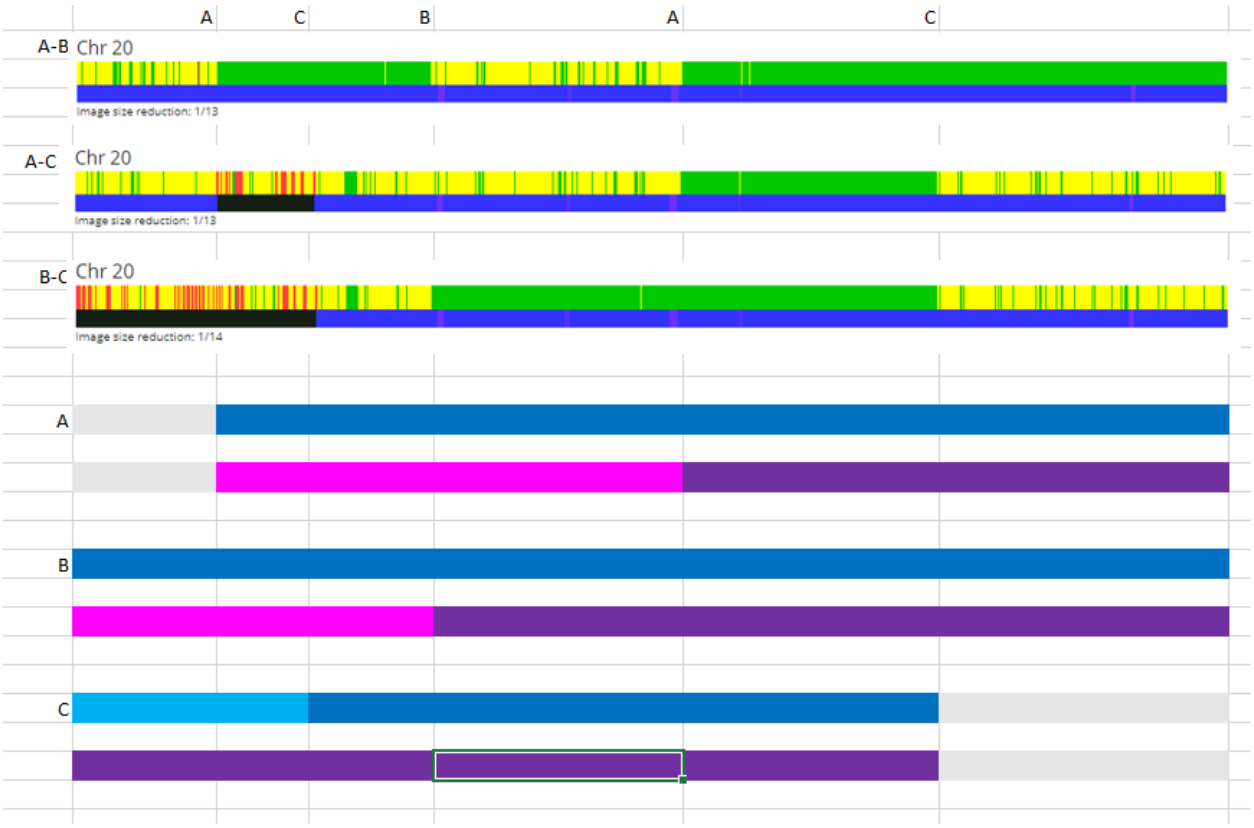

- Now fill in the other siblings in their green and red segments that overlap where you filled in the one sibling as best you. Continue with them until they have recombination points.




- At this point the colors are arbitary meaning each color can represent any grandparent. You can try out different starting points and pick one that gives you as much as possible filled in initially. The moment I cross a recombination point for a sibling or start to fill in farther, the colors are no longer arbitrary and I must be careful to not fill anything in without evidence. I recommend choosing a point that will allow you to fill in as much as possible.



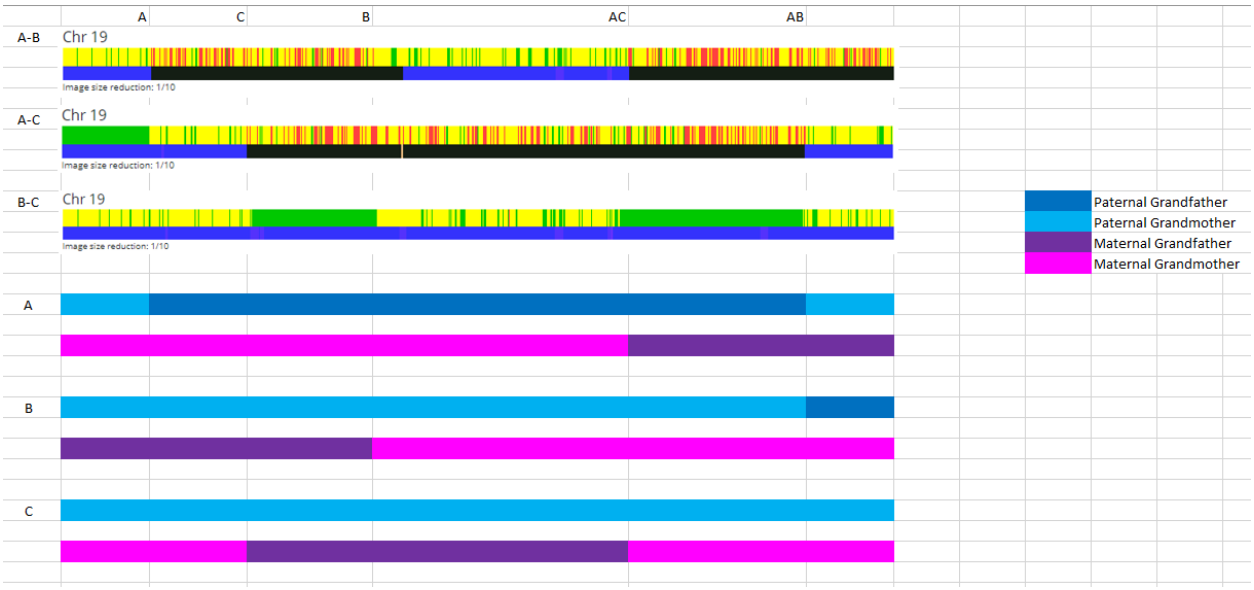

- Start filling in the bottom chromosomes. Pick a place and pick a color. Notice that some of the identified recombinations have already been filled past. In some cases, the top copy switch colors and in other cases it did not. Assume there is only one recombination per point. Therefore if you have already changed colors at one on the top chromosome you do not need to do so again on the bottom. Conversley if you did not switch colors at a recombination on the top then you need to do so on the bottom. In this example I chose to fill in the highlighted cell purple and the rest was logically deduced from that.




- This is as far as I was able to fill in without either testing a fourth sibling or getting more evidence from cousins. The left tip of sibling A is not filled in because there are two solutions and the right tip of sibling C is not filled in because there are also two solutions there. Notice the unsolved places are where the desired siblings are yellow in both of their comparisons. Yellow segments are generally the hardest to solve.
- To solve this I needed one cousin who matched at least one sibling in each grey box and I need to know which grandparent that cousin is related to. I like to compare the siblings against the cousins and then put those comparisons below the solution in excel. Doing so yielded the following solution. In this case the top chromosomes are paternal and the bottom maternal. That will not always be the case and the colors could belong to different grandparents too. After solving it you could repaint it and standardize the colors if you wish.


## Common Problems When Doing Visual Phasing

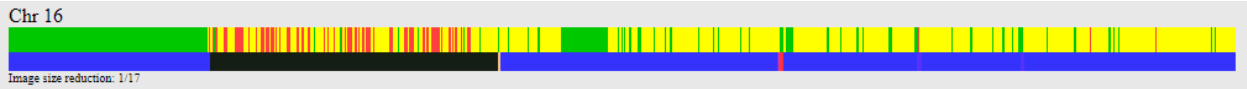
- The last example was fairly straight forward but visual phasing is not always that way. Sometimes two or more siblings will have a recombination in the same or nearly the same spot or one sibling will have two recombinations in the same spot. Sometimes this will be obvious other times it will be hidden and only come to lot when comparisons against cousins does not match up with the solution you created.
- Here is the soluation to an example of two siblings that had a recombination that was not obvious initially:




- At the first point it initially looks like sibling B has a recombination but the truth is A and C

do but not B. At the second point it looks like sibling C has the recombination, but the truth is A and B do but not C.

- Remember that a recombination will usually be for the one that it appears to be for but it could be that the other two siblings have recombinations in that spot instead
- Comparing against cousins can help prove the places these spots exist or adding a fourth sibling to the mix can help unless the fourth sibling also has a recombination in the same spot which is unlikely.
- Here is a more obvious example of double recombination:




- The only way the siblings could go from green to red is with two recombinations in the same place
- Other times you may need to look at a segment under full resulotion to help you determine if a yellow line in a green segment or a red line in a yellow segment is an incorrectly called SNP or a true recombination