

Visual Phasing (part 1)

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History of Visual Phasing - Kathy Johnston posted the process in the FTDNA forums on January 23, 2015.

What is Visual Phasing? - The process of assigning segments of DNA among three or more siblings to shared grandparents using the siblings' recombination points.

Why Visually Phase? – It's difficult to explain until you understand the outcome of visual phasing. Essentially, through visual phasing we can assign much more DNA to our ancestors than standard matches alone allow.

Caveat

You may not like Visual Phasing if.....

- You are a perfectionist.
- You want easy answers.
- You don't like making mistakes.

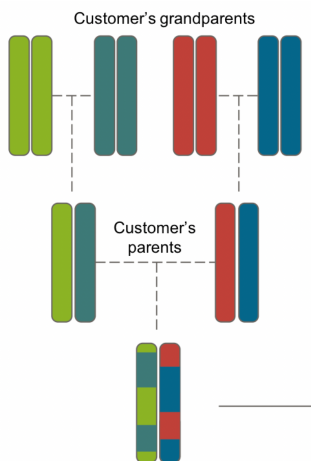
Be prepared to completely redo your Phasing if new information comes to light.

Recombination background

Essentially, all cells in the human body (somatic cells – soma is Greek for body) contain two copies of each autosomal (non-sex) chromosome. When sperms and eggs unite in a process known as fertilization the resulting cell also has two copies of each autosomal chromosome. So, each sperm and egg should only contain one copy of each chromosome so that when they unite, they produce a cell with only 2 copies of each autosomal chromosome.

Humans therefore go through a process of reducing the number of copies of chromosomes when making gametes (i.e., eggs and sperms). This process is called **Meiosis**. Meiosis means diminution in Greek. In meiosis the DNA is re-shuffled or re-distributed so that essentially no two gametes contain exactly the same DNA. This redistribution is called **Recombination**. There are 2 kinds of Recombination that take place in meiosis.

Intrachromosomal recombination changes the DNA along a particular chromosome creating segments of grandma's and grandpa's DNA along the same chromosome through crossover events. **Interchromosomal recombination** changes which of grandma's and grandpa's chromosomes get passed on to the grandchild through a process of independent assortment. For example, if chromosome **1** from grandma is passed on to the grandchild it has no effect (it is independent) on whether the grandchild inherits chromosome **2** from grandma. Each chromosome is independently sorted to the grandchild. However, because intrachromosomal recombination occurs before interchromosomal recombination, the chromosomes are no longer strictly grandma's or grandpa's. There are segments of grandma's DNA on grandpa's chromosomes and vice versa. The main thing to note is that the 2 kinds of recombination combine to yield an incredible amount of diversity in the DNA of each sibling.



To the left is a graphic from Ancestry's White Paper on Ethnicity Estimates. It gives the reader a good representation of how the customer's parents inherit half of their DNA from each of their parents (customer's grandparents). The grandparent's DNA is then scrambled by the parents through recombination and given to the grandchildren (the customer in this case).

The purpose of Visual Phasing is to determine which segment in the grandchild's DNA comes from which grandparent. Once we know which segment was inherited from which grandparent, we can begin to discover how certain DNA cousins match or what trait came from which grandparent.

The Visual Phasing Process

Visual Phasing works best if you have the DNA results of at least 3 siblings to compare. We start the Visual Phasing process by uploading our DNA data to GEDmatch. The next step is to make sure we have the DNA of at least two siblings and some cousins uploaded to GEDmatch. Visual Phasing works best by starting with 3 sibling comparisons. Four sibling comparisons can complicate the data when you are starting out, but may be useful at a later time. Since larger chromosomes have more recombination points, it is recommended to start with the smaller chromosomes first. Chromosomes 21 and 22 have far fewer recombination points than chromosome 1. The next step is to make screenshots of your comparison data and paste them into Excel, Powerpoint or Word. Next you can identify the recombination points in the comparisons and define in which sibling the recombination occurred. Once you have identified the recombination points and who owns them, you can begin filling in segment data.

It is very difficult to describe the whole process in words, so I recommend watching the video. Blaine says there is no perfect way to start, but you can start by filling in the colors on the largest fully identical region (FIR). At this point, the colors you choose are completely arbitrary and have no special connection to any grandparent or other chromosome. You can extend those colors for siblings that don't have recombination points in the extension area. The next thing to color might be fully unidentical regions. Fully identical and fully unidentical regions have fewer variables than half identical regions and we want to have as few variables as possible. Once all the apparent extensions are done, you are at a decision point.

One time and one time only for each chromosome, you can semi-arbitrarily extend one color on one segment. Make it one that will be most informative. This may allow you to complete the filling in for that chromosome or it may not. You may have to wait to compare cousins or aunts and uncles before you can complete the filling in process. You will certainly need cousin comparisons to assign which grandparent contributed which segment. We will talk about that process in the next part.

Homework Assignments to prepare for Part 2:

1. Get your DNA results.
2. Upload to GEDmatch.
3. Set up Visual Phasing workspace on either Excel, Powerpoint or Word.
4. Try the process on at least one chromosome.

It may also help to watch Andy Lee's and Blaine Bettinger's videos on the subject.

Resources

Videos

The cost of Blaine Bettinger's series of videos on Visual Phasing is \$25 via PayPal. You can email Blaine at (blainebettinger@gmail.com) for the password, and you'll have an unlimited amount of time to watch these recordings at your leisure.

<https://vimeo.com/showcase/6781046>

Andy Lee's videos on Visual Phasing and Segment Phasing are available on Youtube and the GEDmatch website. Andy's videos are free.

You can use a screengrabber to copy images of the comparisons. Blaine Bettinger recommends www.AwesomeScreenshot.com.

Additional Resources are available at: goo.gl/LfvXSP

Excel template is available at: goo.gl/99h4Ih