Practical chromosome mapping: gaining insights from segments of DNA

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This talk is intended for those who have mapped their chromosomes or are interested in doing so. A basic understanding of chromosomes and recombination is helpful but not essential.

I hope that after listening you will go away with some fresh ideas on how to apply segments in your DNA research.

Types of segments

A segment of DNA normally refers to a sequence on a chromosome where two people match each other continuously for a significant distance. However, there are also other types of segment that can help us in our quest to identify matches and map our DNA.

HIRs and FIRs

In almost all cases, you will share DNA with your matches on just one copy of the chromosome: a half-identical region (HIR).

When you match a full sibling, some of the DNA you share will match on both maternal and paternal copies of the chromosome: a fully identical region (FIR).

Segment length

Some matching segments of DNA reported by testing companies can be false: a weaving together of maternal and paternal DNA to produce a pseudosegment. Segments of more than 7cM are more likely to be genuine, and some use a larger threshold. This doesn't mean that all segments beneath the threshold are false; it just means that they are far less likely to be traceable and therefore may not be worth your time to investigate. It's also important to remember that

even 15 or 20cM segments can date back to long before the genealogical timeframe.

Ancestral segments

Ancestral segments are based not on where you match someone else, but on how each entire chromosome is divided up by crossovers (also known as recombination points).

When each of us was created, we inherited a maternal and a paternal copy of each chromosome. The DNA on each of these copies was handed out via the process of recombination. Each copy starts with DNA from one grandparent, and there are then crossovers where they may switch to the other grandparent on that side and back.

This produces **ancestral segments**, which are like jigsaw pieces that snap together to make a complete chromosome. These segments don't immediately leap out at us; we have to use other information – such as segments shared with relatives – to try and figure out where they begin and end.

Each ancestral segment contains further ancestral segments from previous generations.

Shared segments

By contrast, a segment you share with a match:

- Is often just a fragment of one of these ancestral segments
- One or both ends might coincide with the start or end of an ancestral segment

These **shared segments** can therefore provide clues that can help identify ancestral segments and add detail to your map.

Inferred segments

Inferred segments are areas on your chromosome where you've been able to establish that you definitely **didn't** inherit DNA from one ancestor. While this can help you advance your map, since these segments are based on negative information, they can help you narrow things down only as far as the ancestor you've inferred.

You can now get segments from AncestryDNA, 23andMe and FamilyTreeDNA that correspond to your ancestral composition (aka ethnicity estimate). These **ancestral composition segments** can sometimes provide clues about ancestral segments

What can we learn?

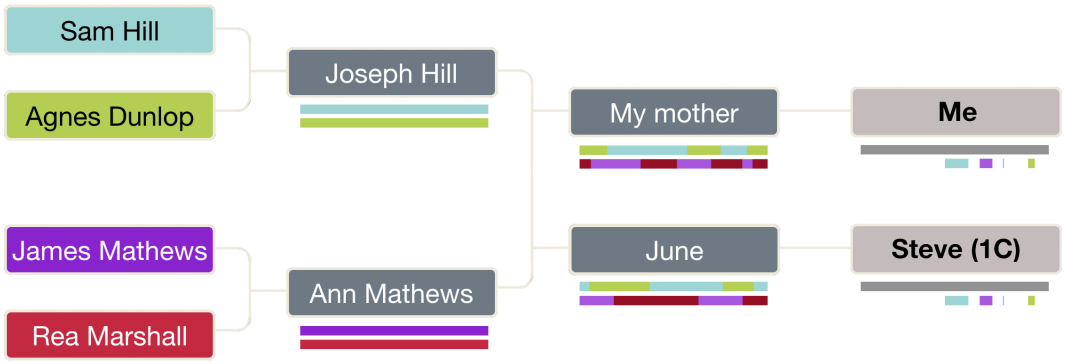
A child inherits DNA from each parent via the process of recombination. This means that in any given spot on a chromosome, the child can only inherit DNA from one of their two grandparents on that side.

So, if a segment of DNA is shared by a known match, we can use the rules of recombination to help us narrow down its possible source:

Each segment you share with a cousin will have been inherited from just one of the common ancestors' parents

To illustrate, let's consider my maternal first cousin Steve:

- Our mothers were sisters
- Our common ancestors are our maternal grandparents
- My mother and Steve's mother each inherited different ancestral segments, but share about 50% of their DNA across half-and fully identical regions
- Me and Steve share DNA on just our maternal copies of the chromosomes, so in any one place, we can share DNA From just one great-grandparent on our maternal side



Here's a table summarizing the conclusions we can draw from segments shared with different relatives:

Relationship	**What you can say about a shared segment**
Cousins	Each segment you share with a cousin will have been inherited from **just <u>one</u> of the common ancestral couple's parents**
Half-relatives	You share just one common ancestor with a half-relative. Each segment will therefore have been inherited from **just <u>one</u> of the common ancestor's parents**
Close relatives	***Caution***: Each segment you share with a sibling, ancestor, or ancestor's full sibling could match them across **a mix of both copies** of their chromosomes

An easy version of the rule to remember

Each segment you share with an *Nth* cousin will have been inherited from just <u>one</u> of your *Nth* great-grandparents

(For removed cousins, use the older person's Nth great-grandparents)

Complications

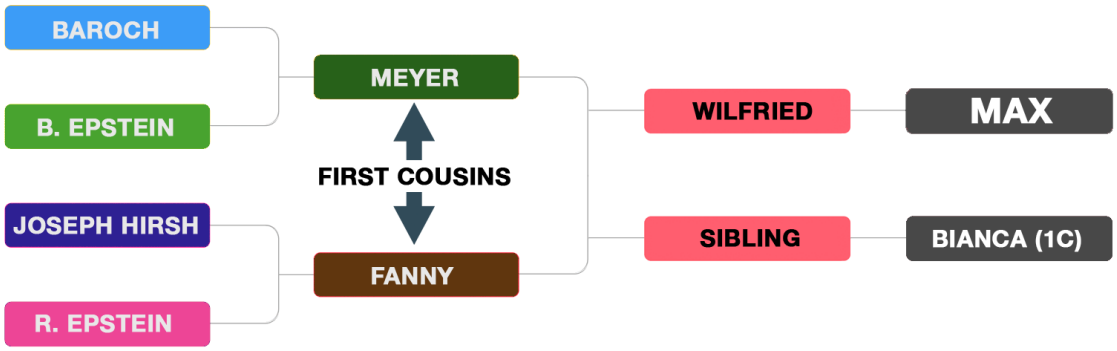
It's important to remember that any time you assign a segment of DNA to an ancestor, that assignment is always provisional. It's always possible that some evidence will come along to contradict your previous assertions. As I said in my first ever webinar for FamilyTreeWebinars, "Always be prepared to be wrong"

Specifically:

1) **Close relatives**: As noted above, segments shared with siblings, ancestors or the full siblings of ancestors may span both of their copies of a chromosome, so this rule only works for cousins and half-relatives
2) **Near-identical crossovers**: Sometimes two siblings will have crossovers that are close enough that two distinct segments are merged
3) **False segments**: Occasionally the testing company's algorithm

will create a ‘matching’ segment that is not real

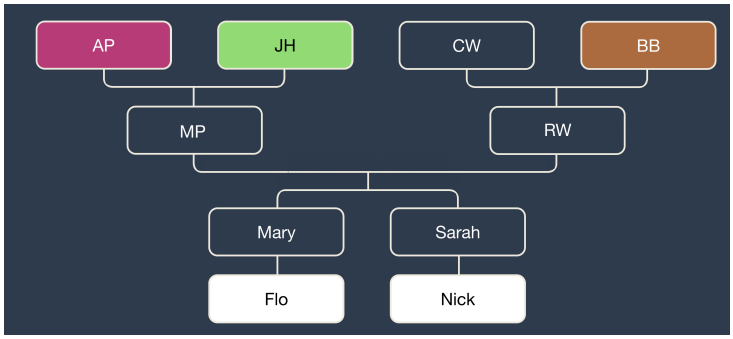
4) **Inferred segments**: These weren’t created by recombination, and therefore you aren’t able to learn any more than what you have already inferred (see links below).
5) **Pedigree collapse**: If you have ancestors who were for example first cousins, this will mean you can’t narrow down segments to the same degree. In the diagram below, each segment that Max and Bianca share could have been inherited via either Baroch, Joseph, or [one of B. and R. Epstein] – it won’t be possible to distinguish between the Epstein sisters.



6) **Multiple relationships**: If you are related to a match on two different branches, this increases the possible sources for each segment. For example, you might have two sets of common ancestors with a double 3rd cousin, meaning each segment shared is from one of the eight parents of these two couples.
7) **Endogamy**: It might be necessary for those with endogamous ancestry to use a higher segment threshold

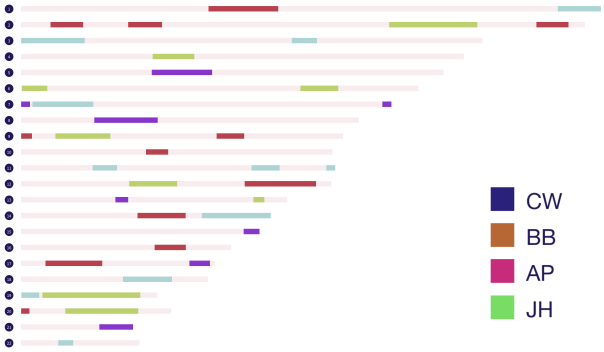
Practical examples

1. Flo and Nick



- Flo and Nick are first cousins, so they share two grandparents and four great-grandparents
- Three of their four common great-grandparents have tested
    - I can use the segments they share with these three to infer the segments they must have shared with the untested great-grandparent

Flo and Nick share 40 segments, and I was able to confirm that each was inherited from just one of their shared great-grandparents.

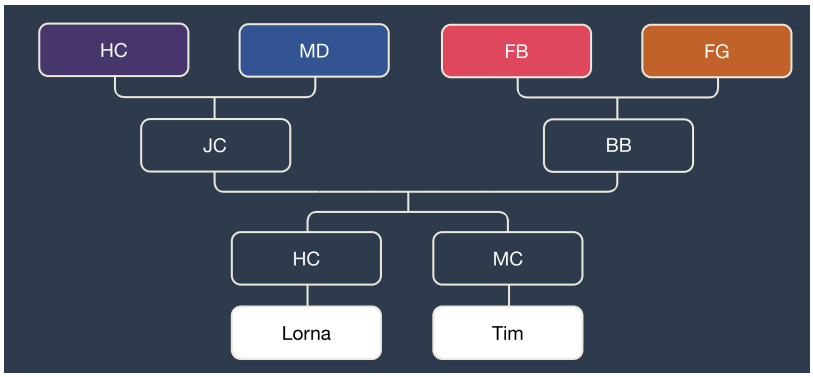


Notes

- It's important to remember that the start and end positions are not precise
- There were also two instances where segments had been incorrectly merged by the testing company. These were both in places where Flo and Nick's mothers had crossovers in very similar positions.

2. Lorna and Tim

Lorna and Tim are also first cousins, but unlike Flo and Nick, none of their ancestors have tested.



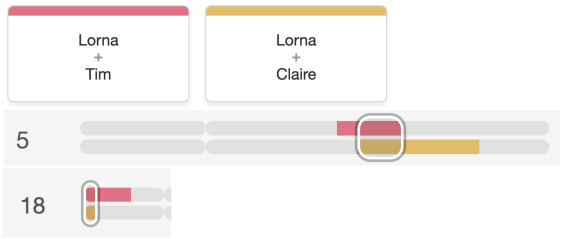
Based on the rules of recombination, I can see that each of the 27 segments they share will be from one of **HC**, **MD**, **FB**, or **FG**.

Although none of these people have tested, there are other testers who are related to just one of these people. I can use these to start classifying which great-grandparent each segment came from.

Claire is a known 3rd cousin who descends from the sister of Frank B. Two segments triangulate with those that Lorna shares with Tim.

I can assign these segments to **Frank B.** in Lorna's map.

Look at triangulated segments



3. Segments shared with a 2nd cousin

These segments can be assigned to one of four great-great-grandparents (the parents of the great-grandparent couple who were our common ancestors). I can use more distant matches to try and determine which of these four people was the source of each segment.

Important: Just because you can assign part of a segment to a more distant ancestor, it doesn't follow that the entire 2nd cousin segment can be assigned to that ancestor. An intermediate ancestor may have had a crossover.

Make a note of your reasoning: as you can imagine, it's quite easy to lose track of things. I therefore highly recommend editing segments on an individual basis to make a note of your reasoning.

4. X-DNA segments

Due to the X-DNA inheritance path, you can potentially rule out certain ancestors from having been the source of an X-segment.

When working with X-DNA segments, be sure to check the X-DNA inheritance works for both you and your match.

5. Using partially identified matches

You can make use of segments where you haven't figured out your connection to the match.

The lighter segments above are segments that I share with unknown matches, but which triangulate with the known segment.

The fact that these people share DNA from a position right at the beginning of the chromosome implies that my ancestral segment probably extends all the way leftwards.

6. Use with segments shared with clusters of matches

The *Cluster Auto Painter* tool lets you map the segments you share with members of clusters made at Gedmatch, Genetic Affairs and DNAGedcom.

Segments associated with clusters could potentially be helpful:

- You could use the trees and locations associated with people in the cluster to try and determine which ancestor might have been the source the segment
- Once you've narrowed down which of the four ancestors was the source, you could then use that information to focus your attempts to identify clusters.

Segment availability

This has changed since 2023. For more info, please see my blogpost 'Segment data: what's going on' linked below.

Selecting matches to research based on shared segments

Once you've mapped out your DNA in detail, it's tempting to browse your segment lists for people who share this identified area of DNA. But in practice you will probably have more success by selecting people for further research based on a combination of the following:

- The total amount of DNA they share with you
- Shared ancestral places and/or names
- Your map can then provide additional evidence

Links

Why map your chromosomes?
https://dnapainter.com/blog/why-map-your-chromosomes/

Your First Chromosome Map: Using your DNA Matches to Link Segments to Ancestors
https://www.youtube.com/watch?v=tzd5arHkv64

Getting Started with DNA Painter
https://www.familysearch.org/rootstech/session/getting-started-with-dna-painter

How to Use Chromosome Browsers for Genealogy (MyHeritage)
https://education.myheritage.com/article/how-to-use-chromosome-browsers-for-genealogy/

Why don't the segments triangulate? (Nicole Dyer)
https://familylocket.com/why-dont-the-segments-triangulate/

Chromosome mapping and endogamy (Lara Diamond)
https://dnapainter.com/blog/chromosome-mapping-and-endogamy/

Genetic Genealogy and the Single Segment (Steve Mount)
https://ongenetics.blogspot.com/2011/02/genetic-genealogy-and-single-segment.html

Inferred Chromosome Mapping: Maximize your DNA Matches
https://www.youtube.com/watch?v=8RrWvOx3gxY

Painting your DNA with inferred matches
https://dnapainter.com/blog/painting-your-dna-with-inferred-matches/

More tips for inferred chromosome mapping
https://dnapainter.com/blog/more-tips-for-inferred-chromosome-mapping/

Painting your populations
https://dnapainter.com/blog/painting-your-populations/

Cluster Auto Painter: unravel your DNA test results
https://dnapainter.com/blog/cluster-auto-painter-unravel-your-dna-test-results/

Segment data: What's going on?
https://dnapainter.com/blog/what-is-going-on-with-segment-data/

Advanced DNA Techniques: Using Phasing to Test DNA Segments
(Blaine Bettinger)
https://familytreewebinars.com/webinar/advanced-dna-techniques-using-phasing-to-test-dna-segments/

Advanced DNA Techniques: Deductive Chromosome Mapping (Blaine Bettinger)
https://familytreewebinars.com/webinar/advanced-dna-techniques-deductive-chromosome-mapping/