SMALL SEGMENT ANALYSIS - JULIAN LAND'S PROCEDURE

After Andrew Millard partially corrected my lack of knowledge of reproductive biology in May 2025, my only way forward was to complete the analysis of all 1963 3cM segments generated from my 12-relative set by GEDmatch (at P = 3). From this my procedure for using small segments to prove family branch connections can now be documented. The following steps will help others do the same thing.

STEP 1

Learn the key - we are looking for rare segment boundary coincidences (RSBCs). These occur when 2 independent pairs of relatives have a coincident boundary (left or right). I found 46 of them - 25 on the left boundary, 21 on the right.

Because the number of these occurrences greatly exceeds the number expected at random (see **Appendix 1**), each RSBC is significant and thus conveys some genetic information. If pair A&B forms a RSBC with pair C&D, then either A matches C or D, or B matches C or D. Of course, A&B and C&D might also be genuine matches. Better genetic information can be obtained as follows.

STEP 2

Put all matches in order, using the left boundary, with each chromosome on its own (Excel) worksheet. Find each RSBC and colour it the same colour whichever chromosome it sits in.

STEP 3

Inspect the matches around each RSBC, looking for evidence that the RSBC will yield more genetic information. In my case I found that 5 RSBCs could not be improved leaving 4 possible interpretations, as explained above. I also found – sometimes after an iteration through the entire set of connections proven to that point - that one of the 4 possible interpretations had already been proven in which case no conclusion was possible, even if one of the 2 matches had been proven.

STEP 4

Some of the remaining RSBCs show just 2 possibilities eg A matches C or D. This can be useful where C and D have a known genetic or genealogical link. In my case I found one such case.

But much more progress can be made with the RSBCs which are amenable to logical deduction.

STEP 5

Here strict logic is required to reject any match around a RBSC which could be a false positive, whilst using any evidence to the contrary, usually in the form of linked 3-person Segment Boundary Coincidences (3pSBCs).

In my case, I was able to prove 21 genetic matches, quite enough to confirm connection between my family branches.

STEP 6

Display the results as in **Diagram 1,** so that the level of family connection between the 12 (in my case) relatives can be seen. The Diagram shows 11 are related via 21 proven family links. Missing are 45 links for there are 66 possible pairings. **Diagram 1** can also show the Step 3 and Step 4 cases.

Most pairs can be linked via 2 connections. Some relatives play a key role with more connections than others – Je, TP and RF have 6 connections each and Ju, SW and RM have 5. Those with the fewest connections are A, E and J having 1 connection each and MD none.

Diagram 2 shows how the uncertain connections can be resolved through genealogical knowledge (within a family branch). **Diagram 3** shows the known matches in a branch not picked up by our method.

CONCLUDING COMMENT

It is certainly true that small segments are a challenge for those who have to work with them. We describe a method of dealing with this challenge, starting with the set of Rare Segment Boundary Coincidences (RSBCs) which occurs amid any large set of small 3cM matches generated from a set of relatives.

Previous work indicated that a sample of relatives generated 5% more 3cM matches than the same-sized random sample. Here our 46 RSBCs comprise 2-3% of our 1963 matches. The number of proven family matches was found to be about 2% of 1963.

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APPENDIX 1 - PROBABILITY

The total number of coincidences expected at random involving 4 people from 12 in a set of N matches over each of 22 chromosomes would be 22*N*(N-1)100,000 = 1.7. We found 25 RSBCs (left) and 21 RSBCs (right). The 3-person Segment Boundary Coincidences (SBCs) occur more frequently. We suggest the excess incidence in both categories reflects the impact of family.

In this estimate of random occurrences, there were about (N=) 89 segment matches found per chromosome at the settings used (Qmatch 3cM P=3), and there are about 50,000 SNPs per chromosome.

The 3-person version involves one person occurring twice but not in the same match. The number of 4 person matches from M is:

$$M^*(M - 1)^*(M - 2)^*(M - 3)/24 = 495 \text{ when } M = 12$$

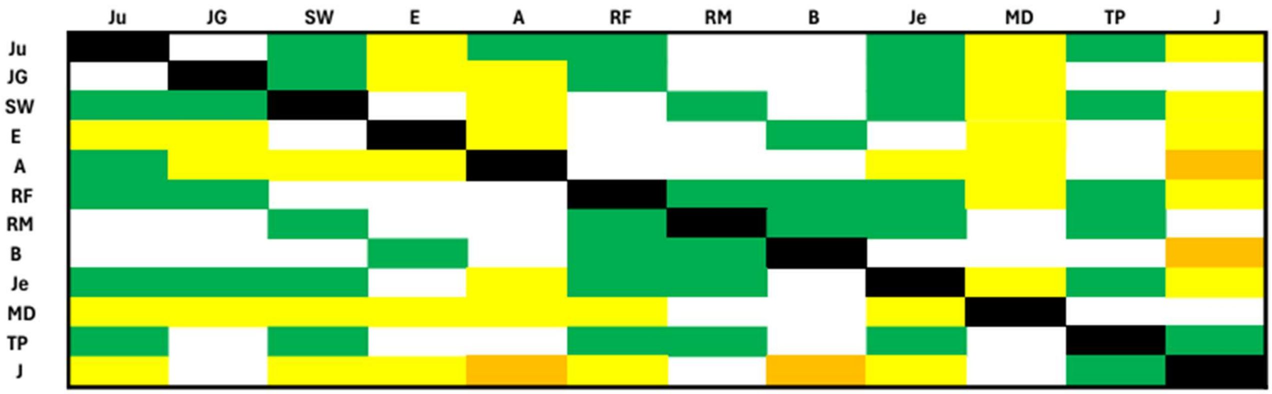
The number of 3-person matches from M is:

$$2/3^*3^*M^*(M - 1)^*(M - 2)/6 = 440 \text{ when } M = 12$$

That is, in our situation the number of 3-person SBCs should be smaller, but in fact is much larger. This large number of SBCs thus contains much family data. One’s initial temptation to dismiss them should be resisted, because they play a role in distilling the connections lying in the RSBCs.

DIAGRAM 1

Here we show the family matches generated by 46 RSBCs.

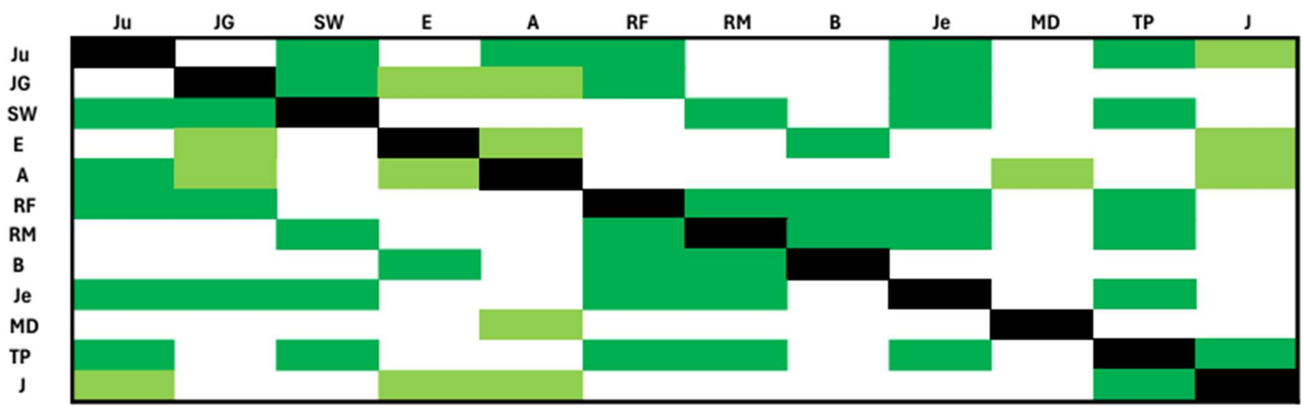


The 21 proven matches are shown in green. J matches A or B as shown in orange, each case having an a priori probability of 50%. In 5 cases we were left with 4 possibilities (shown in yellow) - but with 2 of the 20 possibilities duplicating themselves, we are left with 18 possible connections with a probability of 25%.

Those uncertain matches can be resolved through genealogy as shown in Diagram 2.

DIAGRAM 2

Here, all the matches shown as uncertain in Diagram 1 can be upgraded due to genealogical certainty. With one (or sometimes 2) of each set of possibilities confirmed, the others fall away, hence no yellow or orange entries.

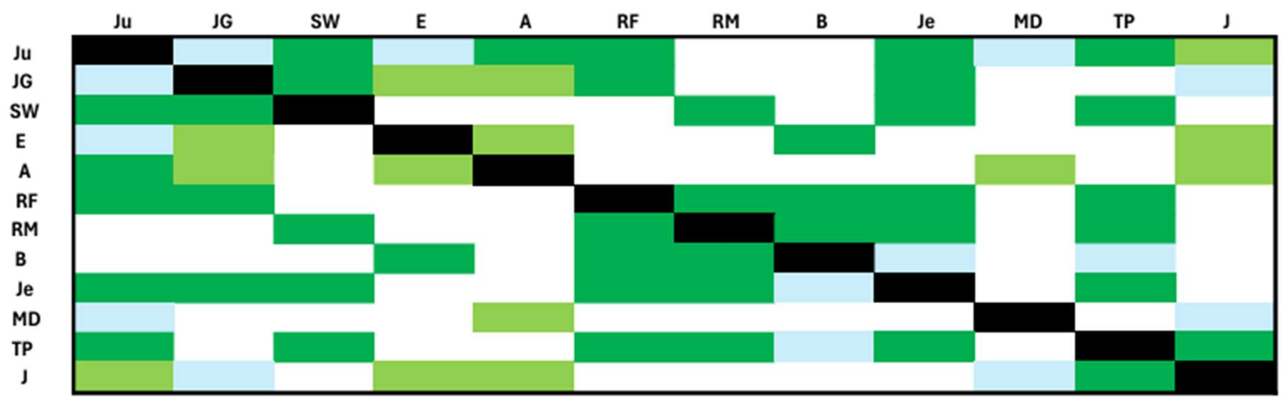


The lighter shade of green shows where we can upgrade the certainty of the match.

Some of the matches not found with our small match procedure are in fact known through genealogy. These are shown in Diagram 3.

DIAGRAM 3

Here we can show where matches not found by our small match procedure are in fact known.



The blue colour marks those connections which are known by genealogy alone.

The 31 white entries perhaps indicate that we are near the limits of technology, for there are 66 possible pairings. We may have been somewhat fortunate to have found 6 of our 12 relatives having cross-branch proven connections.