Supplemental Material

Figure S1

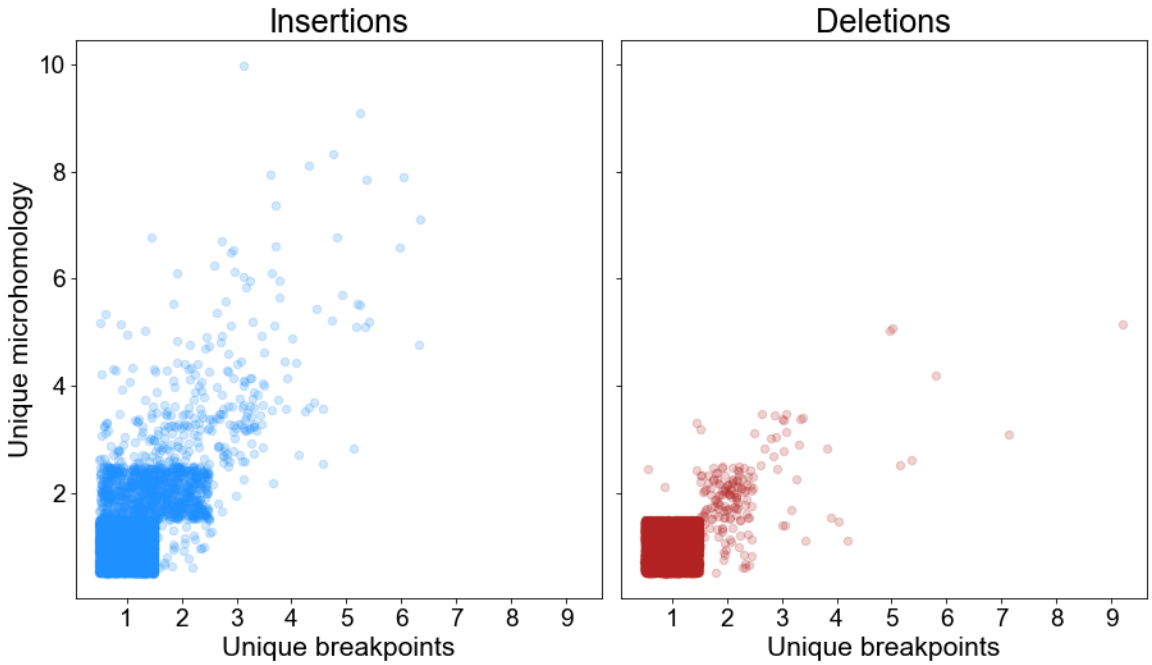

Figure S1: Breakpoint changes alter microhomology. The number of unique breakpoints (horizontal axis) a variant has across haplotypes has a dramatic impact on the number of unique microhomology annotations (violet line: best-fit least-squares regression line). Transparency and jittering (± 0.5) separates points falling on integers.

503 Figure S2

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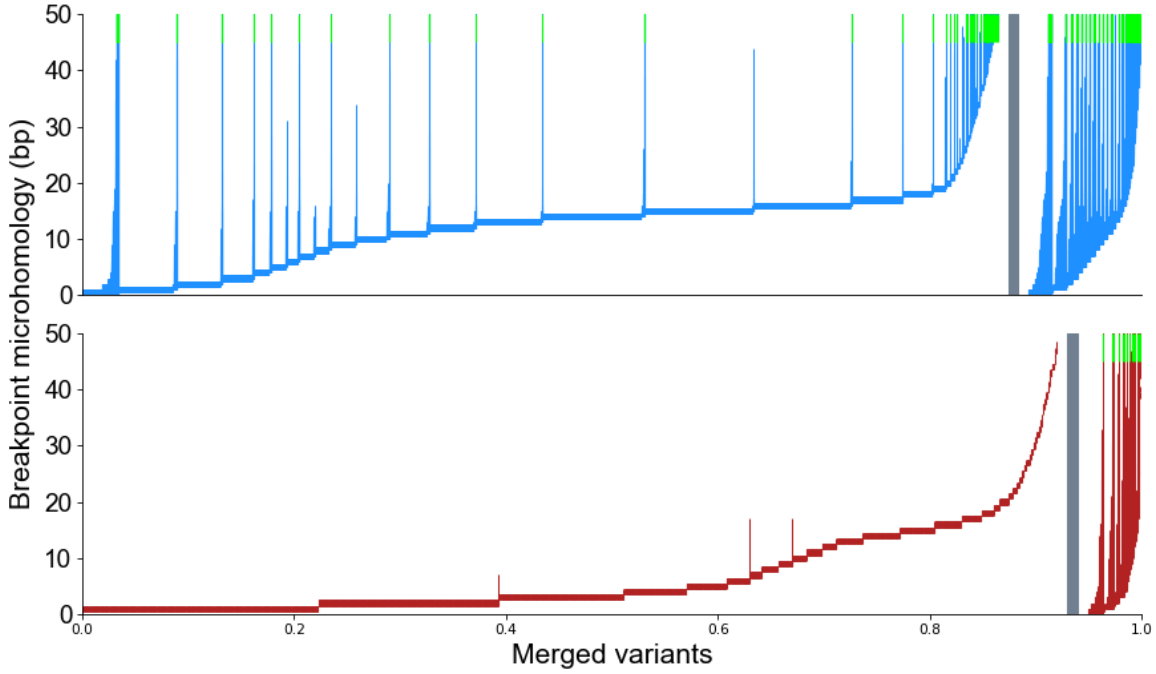

Figure S2: Breakpoint placement leads to large microhomology differences. For each merged SVs (insertions top/blue, deletions bottom/red), vertical lines extend from the minimum microhomology to the maximum microhomology across haplotypes. A gray bar separates SVs with consistent breakpoints (left) from SVs called at different breakpoints across haplotypes (right). Green tips denote lines that extend past the top of the figure.

Figure S3

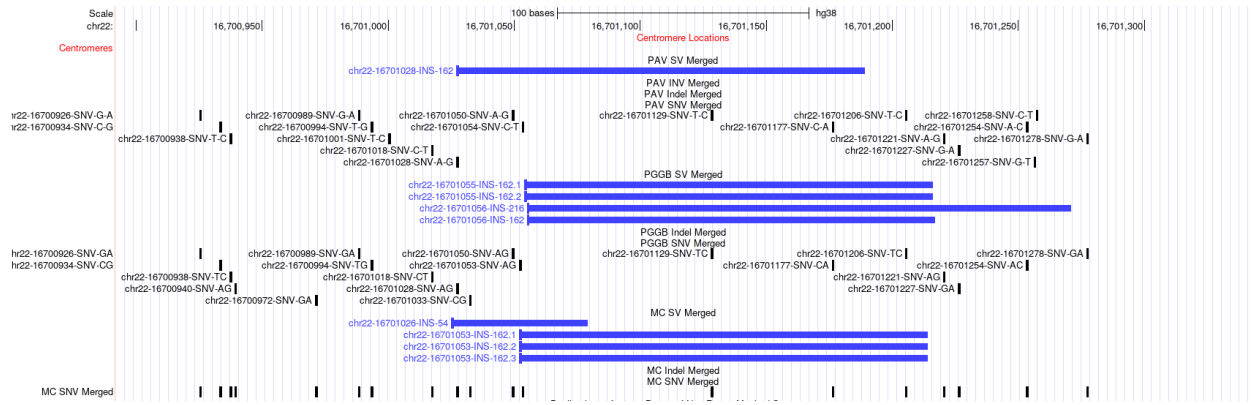

Figure S3: Ambiguous breakpoints for SVs in degenerate tandem repeats. The true breakpoint for this 162 bp expansion is difficult to identify even though tandem repeats in this locus were too diverged or too small to yield a tandem annotation. Despite this divergence, breakpoints were still not consistently placed, and the optimal location is difficult to identify and all three methods chose different breakpoints.

Table S1

Table S1. Offsets per haplotype pair. Average effect of shifted breakpoints on each pair of haplotypes (n = 2,016 combinations of 64 haplotypes). TR: Tandem Repeat, SD: Segmental Duplication, N: Number of variants, Diff: Variants with different offsets in the pair of haplotypes.

	Insertions			Deletions		
	N	Diff	Diff %	N	Diff	Diff %
No TR/SD	1,235	54	4.4%	391	7	1.7%
SD, no TR	58	6	9.8%	22	2	8.8%
All	4,862	967	20.0%	2,614	382	14.7%