Short INDELS: genetic markers for adaptive divergence

Original aspects of the short INDELs project

- Divergent natural selection vs neutral processes
- Species with high diversity
- Systems with imperfect genomes can still contain useful functional information

INDEL-SNP comparisons

1. Outlier sharing
2. Clustering of (different types) markers
3. Derived allele frequencies (in progress and for now simply minor)
4. Distributions of cline parameters

1. Outlier sharing

Total number of Anja's SNP: 55106

Total number of SNP: 11225

Total number of INDEL: 1752

Proportion of SNP with significant clines.	Proportion of SNP with significant clines.	Proportion of INDEL with significant clines.
CZA left: 0.5122128	0.5317595	0.5296804
CZA right: 0.4238377	0.4457016	0.4549087
CZB left: 0.3201829	0.3277506	0.3413242
CZB right: 0.4114071	0.4244989	0.4092466
CZD left: 0.4393351	0.4473942	0.4737443
CZD right: 0.4732697	0.4823163	0.4834475
Proportions of SNP outliers that are shared.	Proportions of SNP outliers that are shared.	Proportions of INDEL outliers that are shared.
CZA left and right: 0.707804	0.6160714	0.7058824
CZB left and right: 0.5680581	0.5178571	0.4705882
CZD left and right: 0.6569873	0.6339286	0.6470588
CZA and CZB: 0.4650635	0.359375	0.3529412
CZA and CZD: 0.508167	0.4107143	0.4117647
CZB and CZD: 0.5426497	0.484375	0.4411765
Number of SNP outliers found in 1 hybrid zone(s): 602	142	24
Number of SNP outliers found in 2 hybrid zone(s): 258	66	7
Number of SNP outliers found in 3 hybrid zone(s): 137	29	7
Number of SNP outliers found in 4 hybrid zone(s): 117	27	5
Number of SNP outliers found in 5 hybrid zone(s): 95	25	1
Number of SNP outliers found in 6 hybrid zone(s): 139	13	3
Prop. of SNP outliers in inversions found in 1 zone(s): 0.648	0.556	0.625
Prop. of SNP outliers in inversions found in 2 zone(s): 0.698	0.636	0.57
Prop. of SNP outliers in inversions found in 3 zone(s): 0.912	0.862	0.86
Prop. of SNP outliers in inversions found in 4 zone(s): 0.923	0.889	1
Prop. of SNP outliers in inversions found in 5 zone(s): 0.979	0.92	1
Prop. of SNP outliers in inversions found in 6 zone(s): 1	1	1

Except this difference in the total number of SNPs, the proportions look quite similar but I have not run any statistical tests

Given the difference between SAMtools and GATK in the total number of SNPs, I have run some diagnostics

- Contigs after coverage filter (in progress)

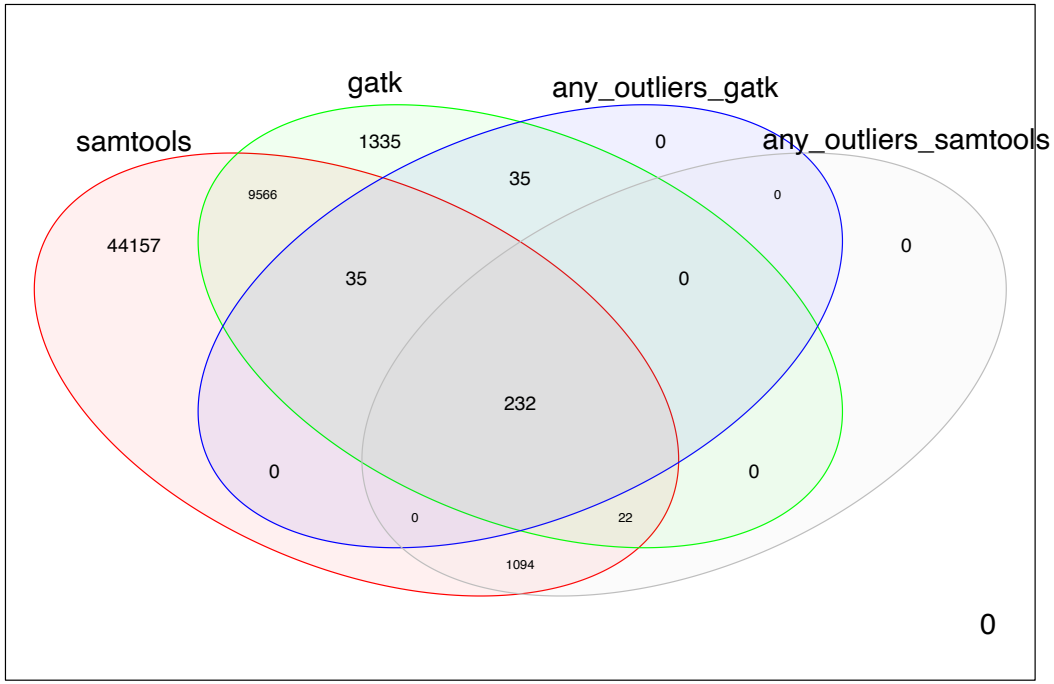
Given the difference between SAMtools and GATK in the total number of SNPs, I have run some diagnostics

- SNPs after all the filters (in progress)

Given the difference between SAMtools and GATK in the total number of SNPs, I have run some diagnostics

- SNPs after all the filters and cline/outlier analysis (i.e., clinal and non-clinal SNPs)

any_outliers = outlier in at least one CZ.



all_outliers = outlier shared by all six CZs (two per islands).

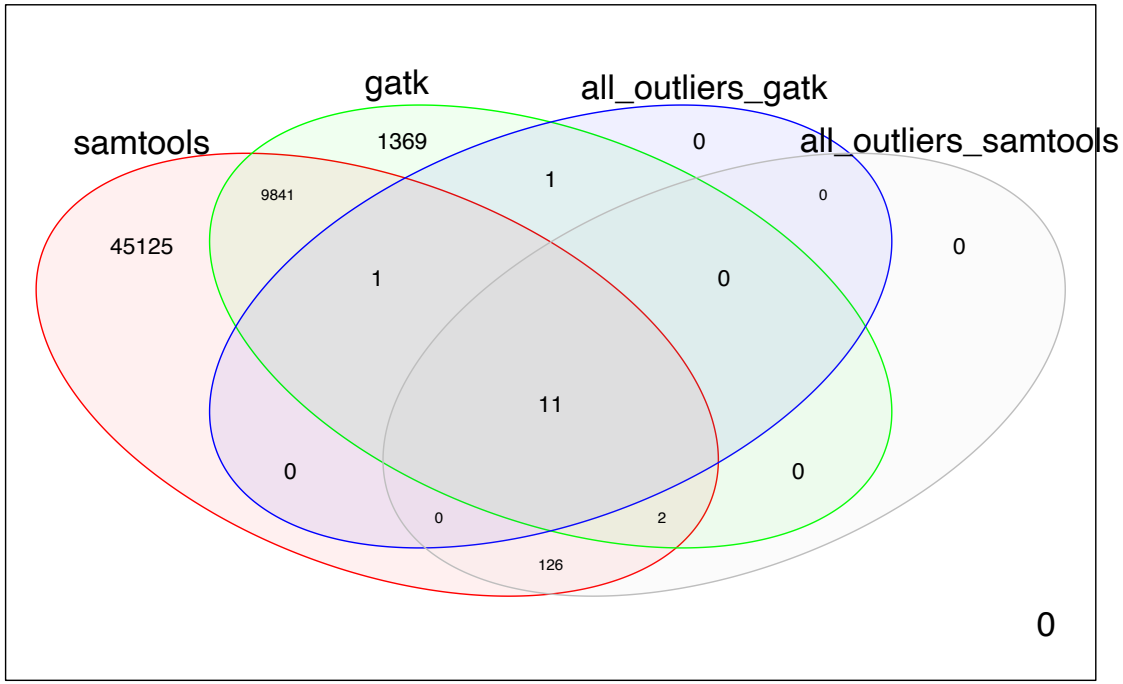
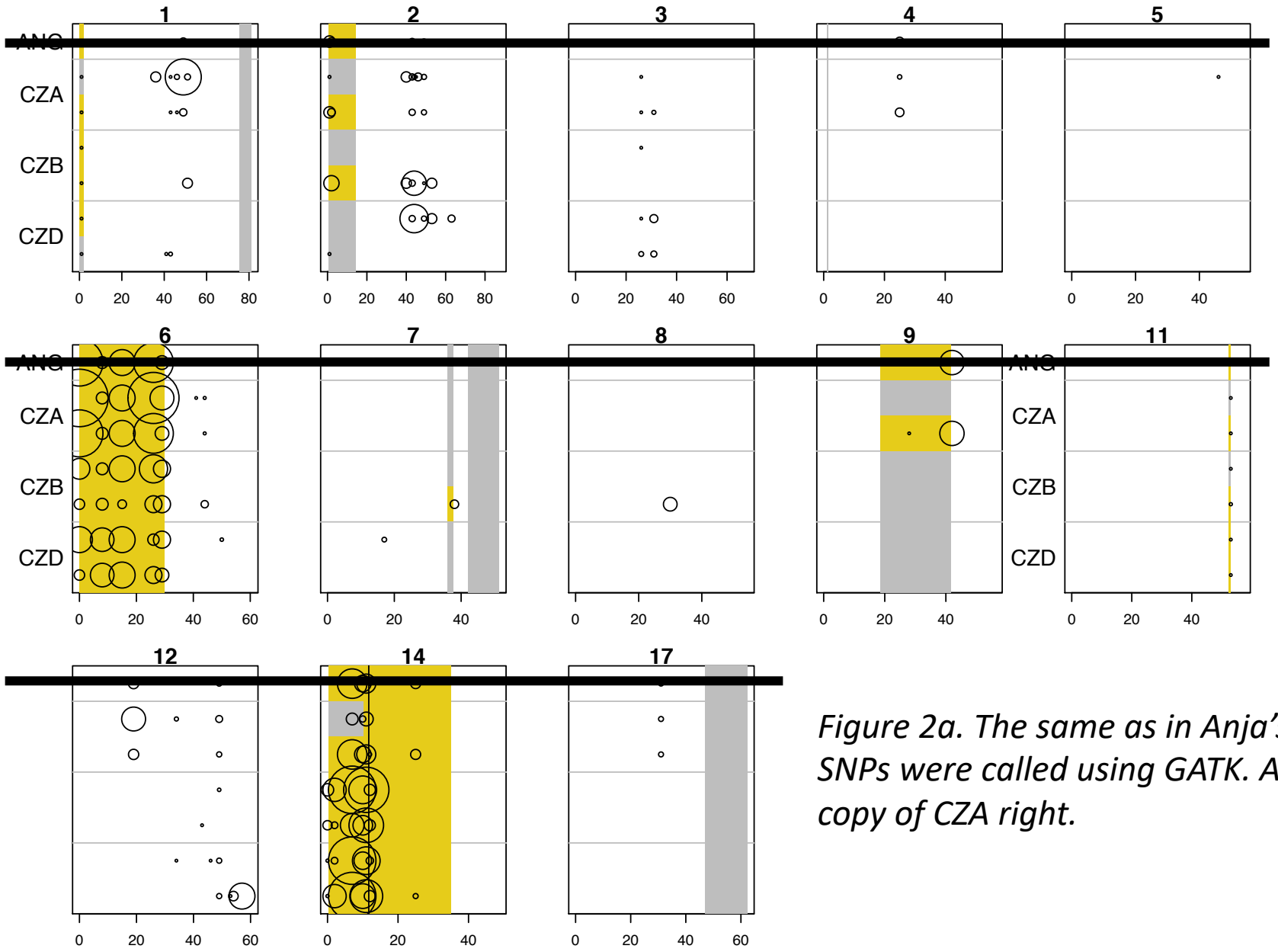


Figure 1. Venn diagrams of the number of SNPs after filtering and cline analysis. Left: SAMtools and GATK calls are intersected with the respective outliers that were at least present in one hybrid zone. Right: SAMtools and GATK calls are intersected with the respective outliers that were present in all six hybrid zones (two per islands).

GATK SNP call



Except this difference in the total number of SNPs, the map positions look quite similar but I have not run any statistics to test for significance.

Figure 2a. The same as in Anja's paper with the exception that the SNPs were called using GATK. ANG is crossed out because it was a copy of CZA right.

GATK INDEL call

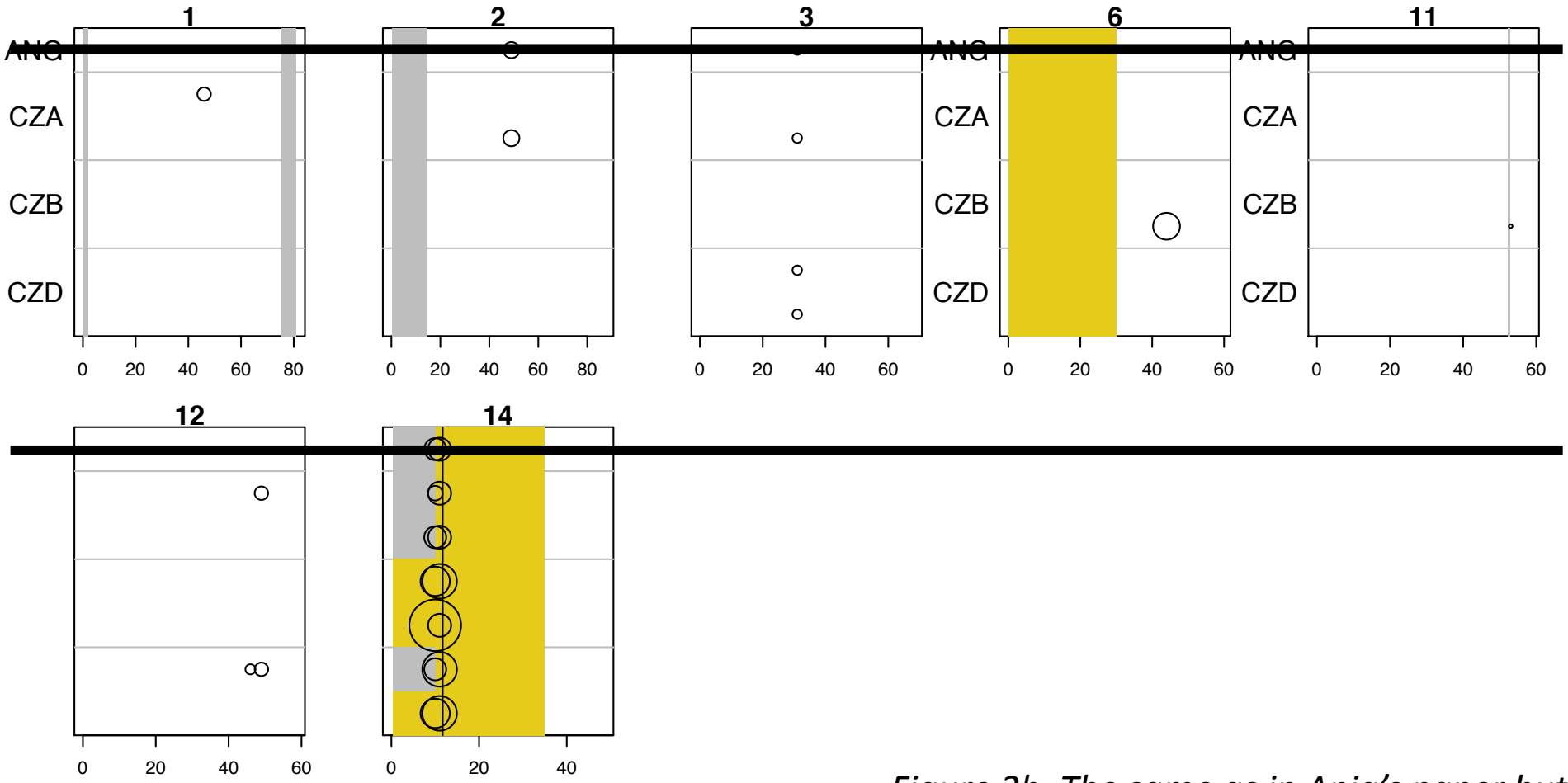


Figure 2b. The same as in Anja's paper but with INDELs. ANG is crossed out because it was a copy of CZA right.

2. Clustering of (different types) markers

- INDELs and SNPs after filtering and cline analysis.
- All six hybrid zones combined.

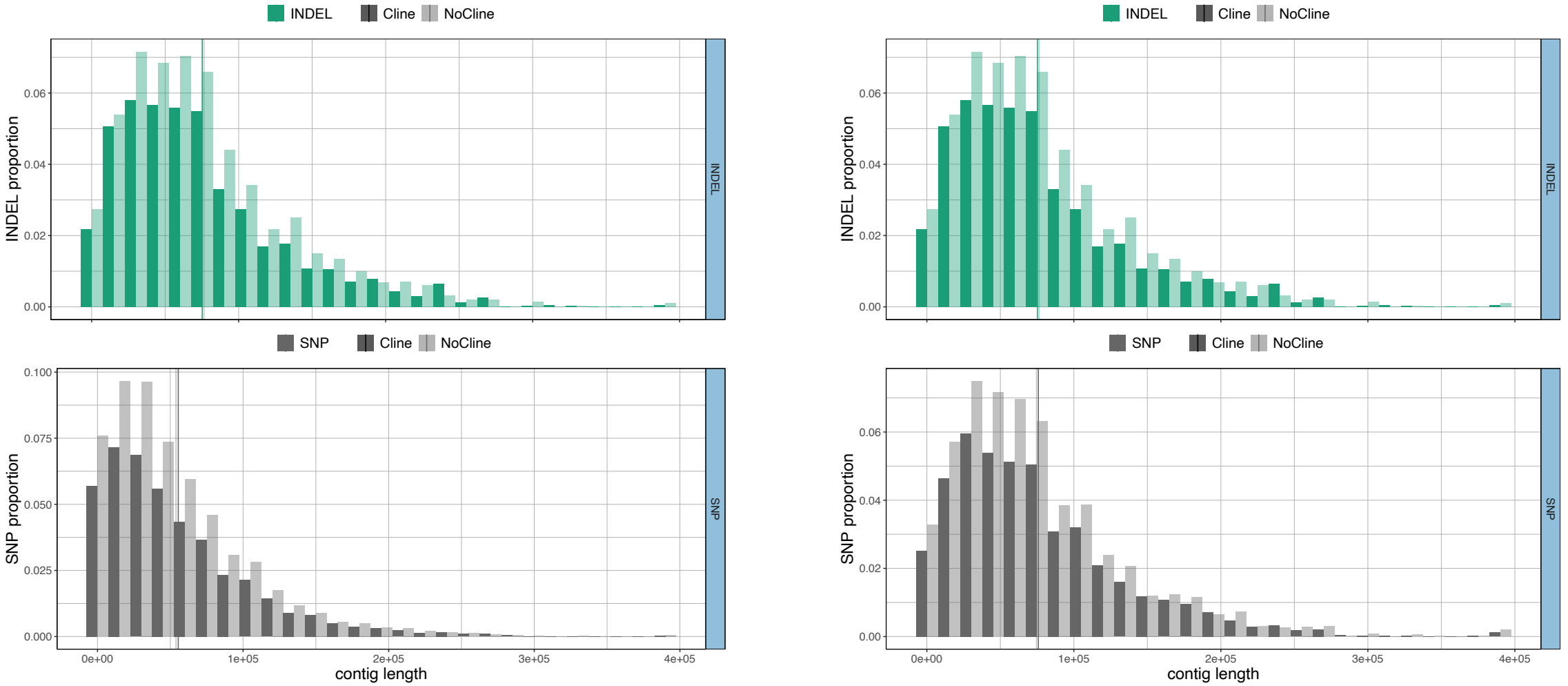


Figure 3a. Marker proportions over contig length. $Proportion = count/\Sigma\, count$ per marker type and bin width = 15000 base pairs. Clinal variants are dark coloured and non-clinal variants are light coloured. Left: SNP call using SAMtools and INDEL call using GATK. Right: both INDELs and SNPs were called with GATK.

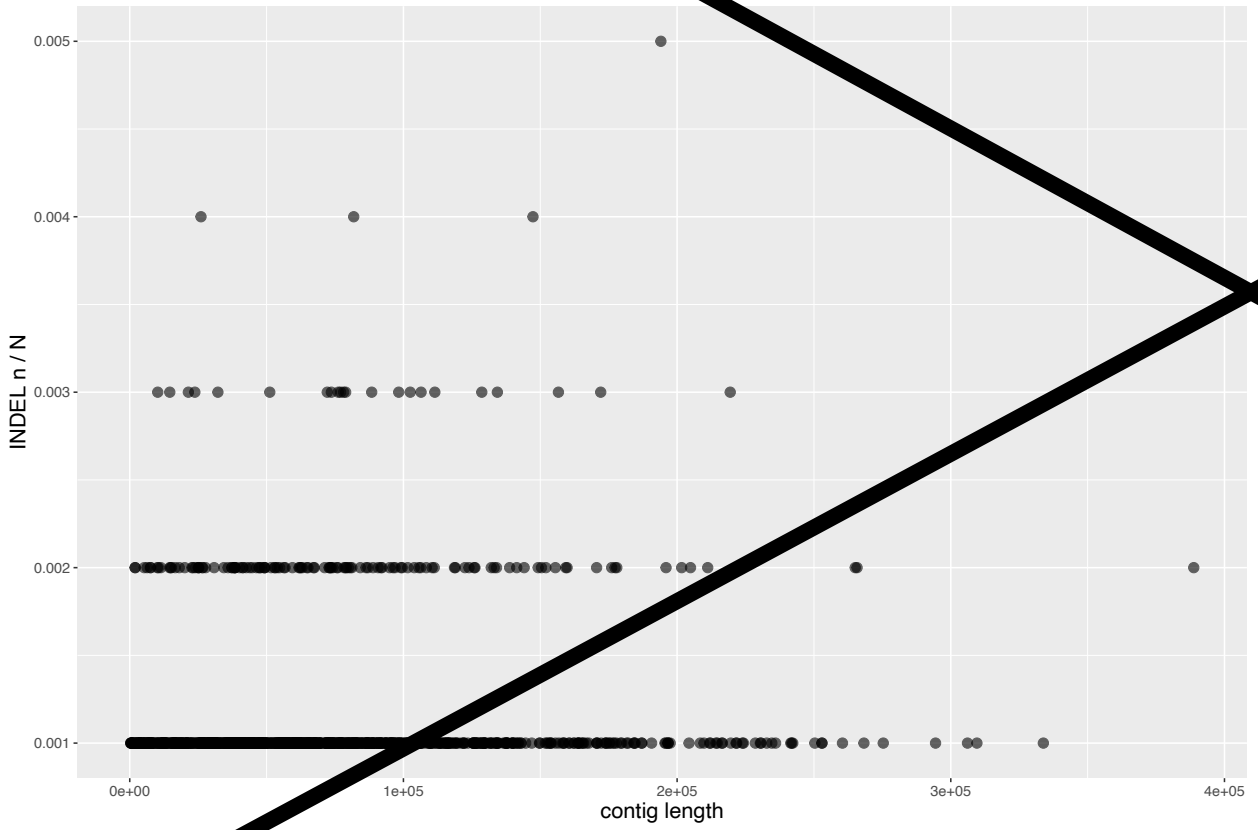
2. Clustering of (different types) markers

- All INDELs = clinal + non-clinal

$$n/N = \frac{n_i\ marker}{N\ marker}$$

n_i = number of INDELs in contig i

N = Total number of INDELs



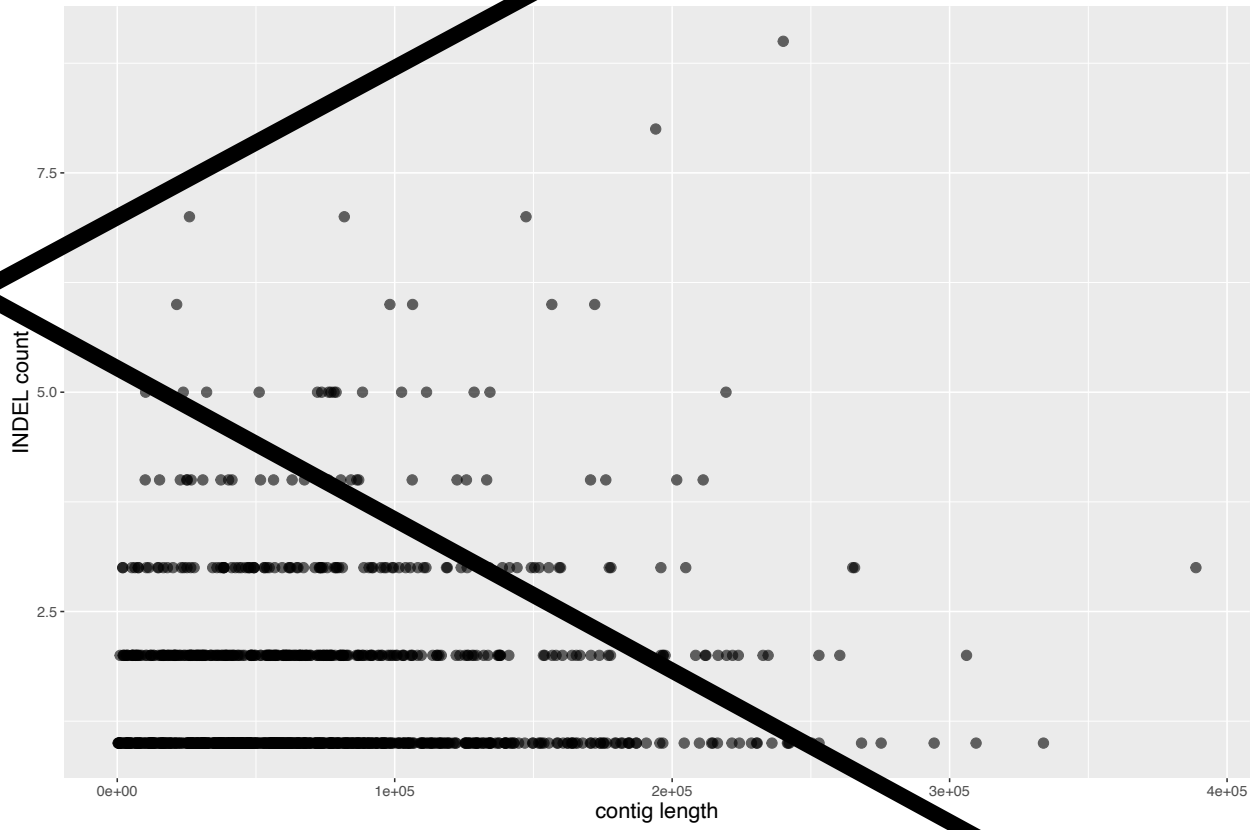


Figure 3a. Proportions (left) and counts (right) of INDELs per contig.

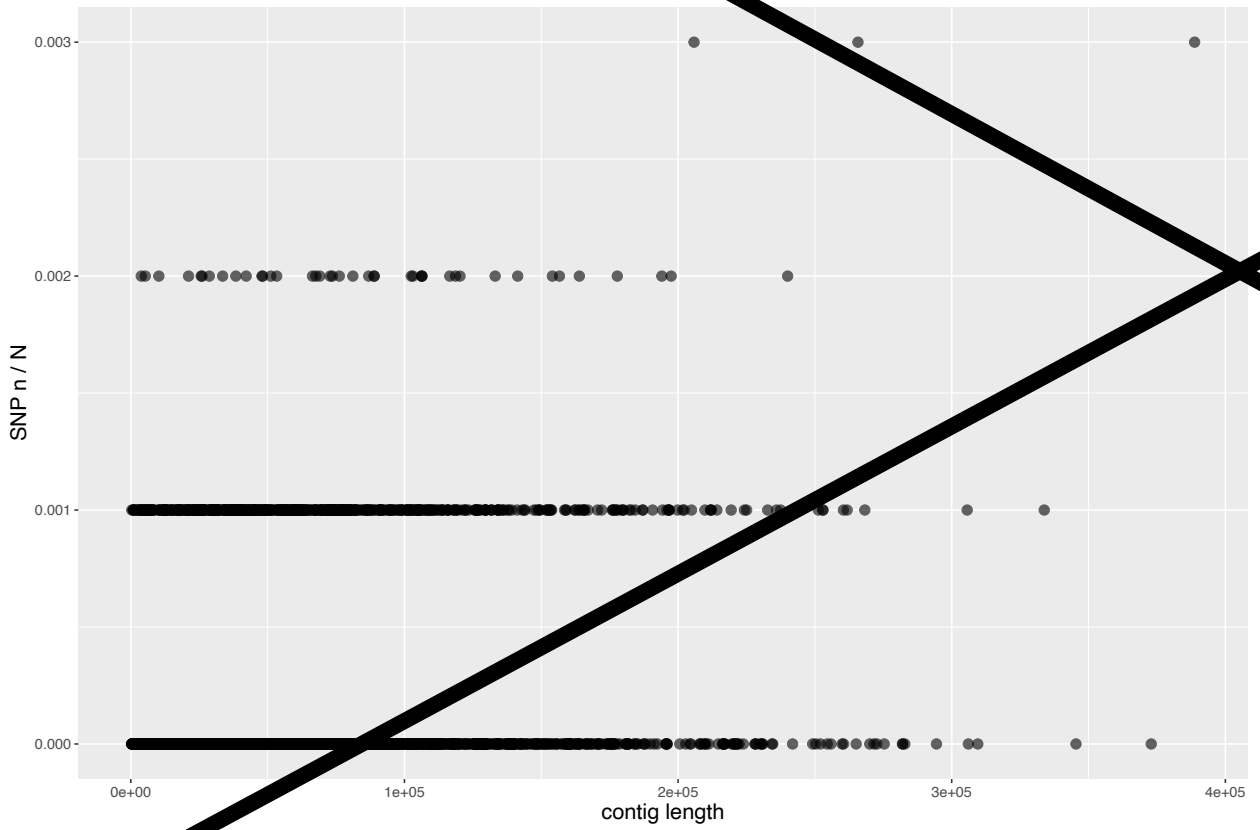
2. Clustering of (different types) markers

- All SNPs = clinal + non-clinal

$$n/N = \frac{n_i\ marker}{N\ marker}$$

n_i = number of SNPs in contig i

N = Total number of SNPs



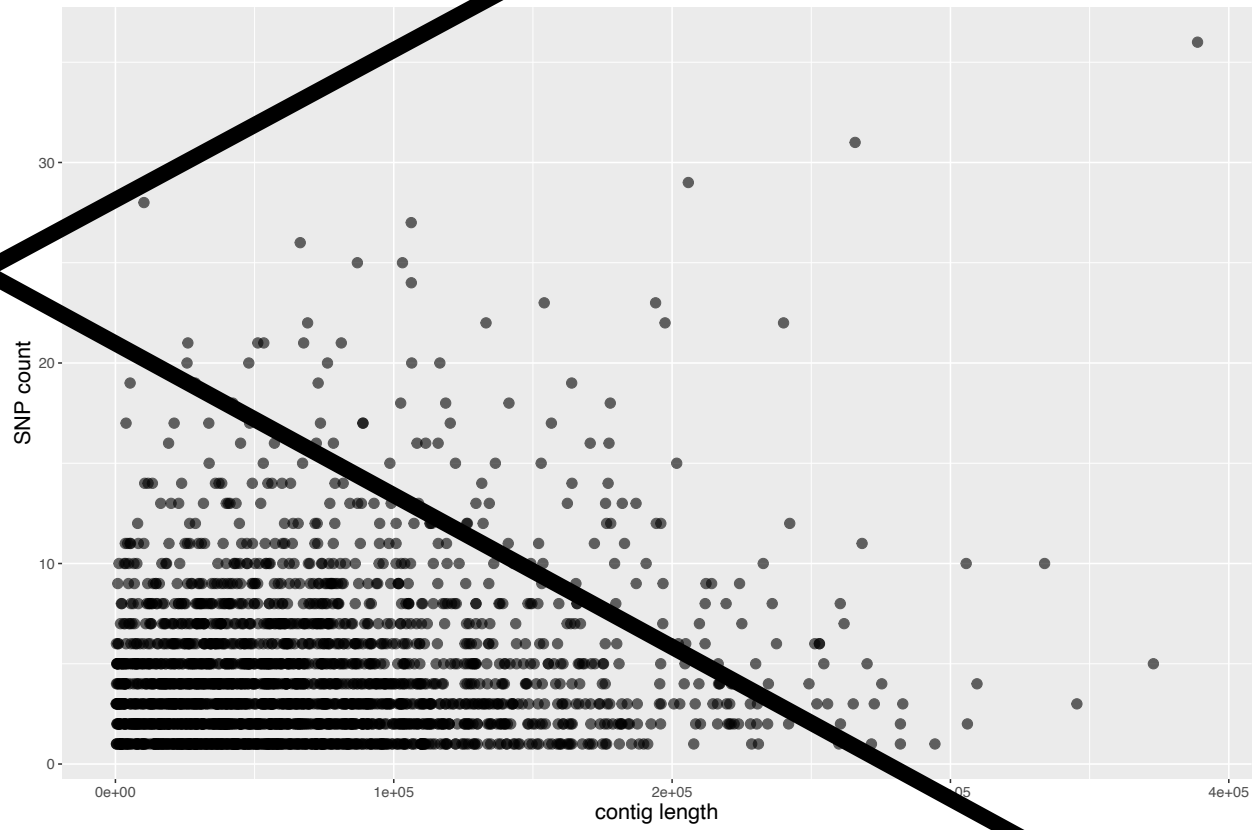


Figure 3b. Proportions (left) and counts (right) of SNPs per contig.

2. Clustering of (different types) markers

- All INDELs and SNPs in the same contigs

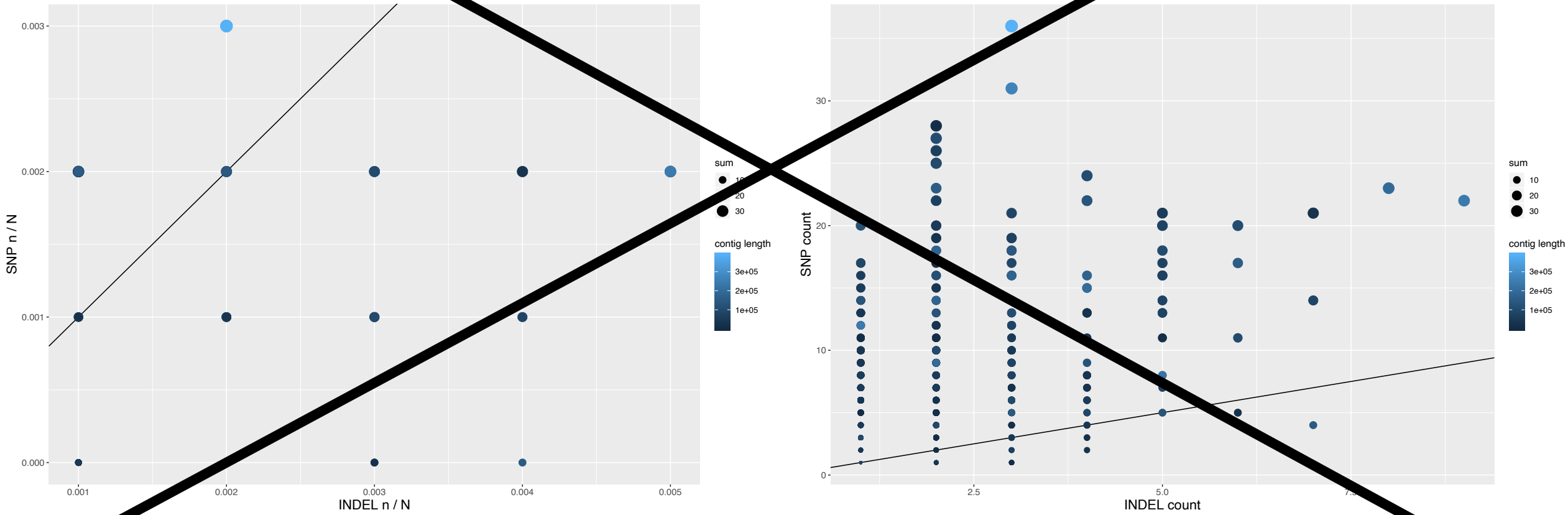


Figure 3c. Relationship between SNPs and INDELs with respect to their proportions (left) and their counts (right) per contig. Proportions and counts of SNPs and INDELs are the same as in Fig. 1-2.

3. Derived allele frequencies (in progress; for now minor allele frequencies Fig. 4-5)

- Ancestral state was inferred from called genotypes:

    1. Reference allele = ancestral allele
       compressa is homo for the reference allele (0)

    2. Alternative allele = ancestral allele
       compressa is homo for the alternative allele (2)

    3. Unknown ancestry
       compressa is het (1)

3. Minor allele frequencies (GATK call)

- INDELs and SNPs after filtering but before cline analysis.
- All six hybrid zones combined.

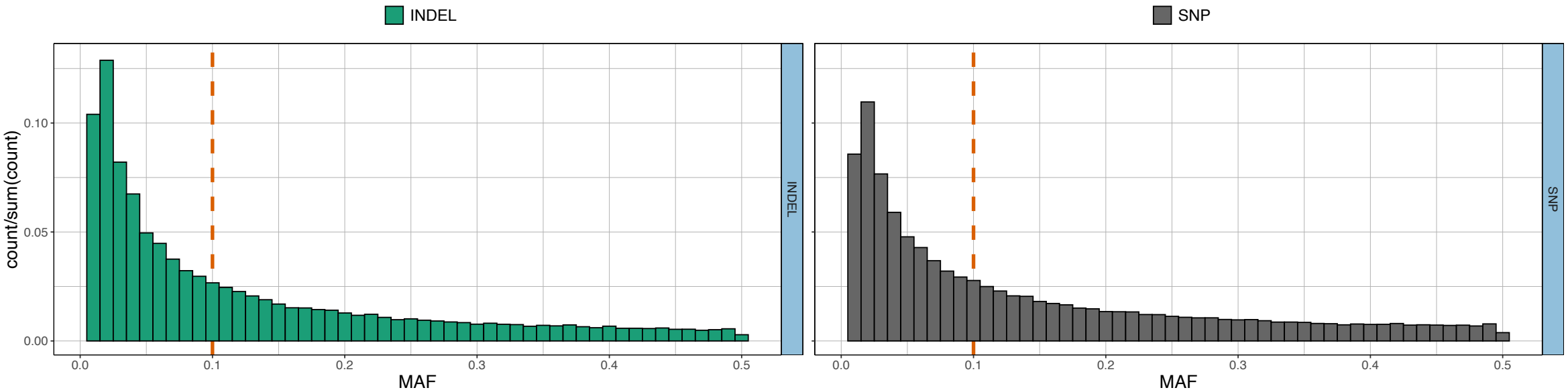


Figure 4. Proportions of minor allele frequencies of INDELs (left) and SNPs (right) after filtering but before cline analysis. Bin width is 0.01 and orange dashed line marks the maf filter in the cline analysis (0.1).

3. Minor allele frequencies (GATK call)

For the joint AFS between INDELs and SNPs, see (attached) file short_indels.nb.html

- INDELs and SNPs after filtering and cline analysis.
- All six hybrid zones combined.

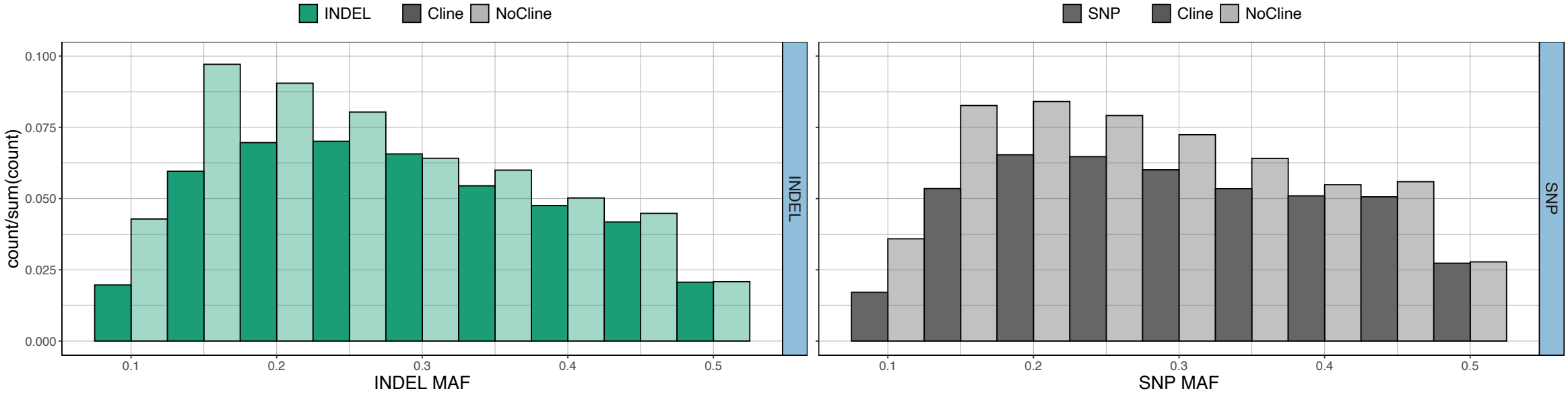


Figure 5. Proportions of minor allele frequencies of INDELs (left) and SNPs (right) after filtering and cline analysis. Bin width is 0.05. Clinal variants are dark coloured and non-clinal variants are light coloured.

3. Derived allele frequencies - INDELs

- Ancestral state was inferred from called genotypes:

    1. Reference allele = ancestral allele = ref_anc
    compressa is homo for the reference allele (0)

    2. Alternative allele = ancestral allele = alt_anc
    compressa is homo for the alternative allele (2)

    3. Unknown ancestry = het
    compressa is het (1)

Table 1. Count of INDELs and SNPs for each combination of possible allelic states given one outgroup (L. compressa) with two samples (NE and W). There are two combinations in which the allelic state is concordant in both samples (in green), eight in which the allelic state can only be retrieved from one sample (in yellow) and finally, five in which the allelic state cannot be inferred (in red).

NE_Lcomp	W_Lcomp	INDEL	SNP
alt_anc	alt_anc	5305	27188
alt_anc	het	528	3543
alt_anc	NA	245	1097
alt_anc	ref_anc	511	2195
het	alt_anc	2231	12439
het	het	1577	9691
het	NA	151	627
het	ref_anc	3120	17198
NA	alt_anc	158	765
NA	het	33	267
NA	ref_anc	449	1831
ref_anc	alt_anc	693	3292
ref_anc	het	1422	7462
ref_anc	NA	1003	3675
ref_anc	ref_anc	38884	178715

3. Derived allele frequencies (GATK call)

- INDELs and SNPs after filtering but before cline analysis.
- All six hybrid zones combined.

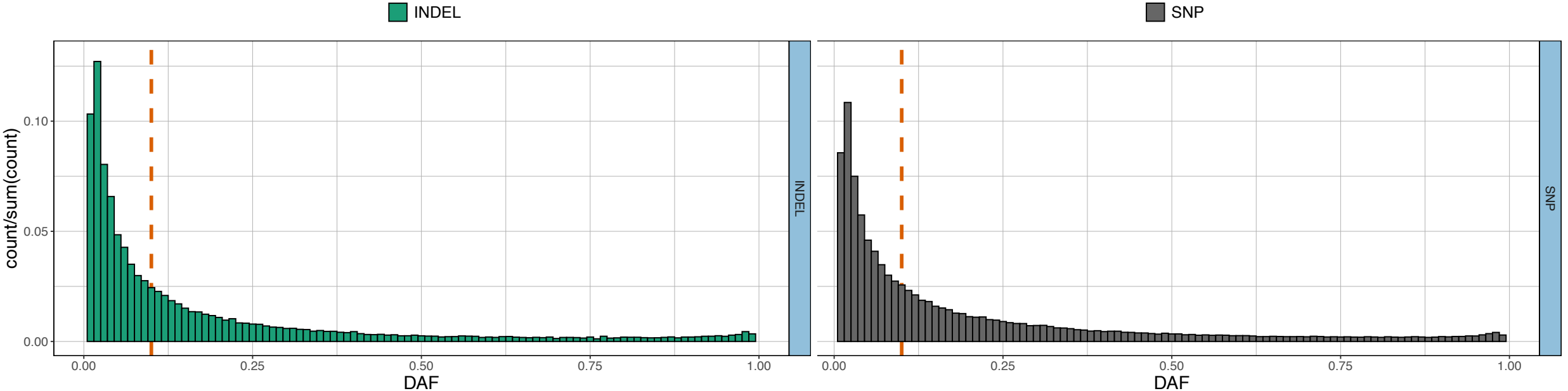


Figure 6. Proportions of derived allele frequencies of INDELs (left) and SNPs (right) after filtering but before cline analysis. Bin width is 0.01 and orange dashed line marks the maf filter in the cline analysis (0.1).

3. Derived allele frequencies (GATK call)

- INDELs and SNPs after filtering but before cline analysis.
- All six hybrid zones combined.

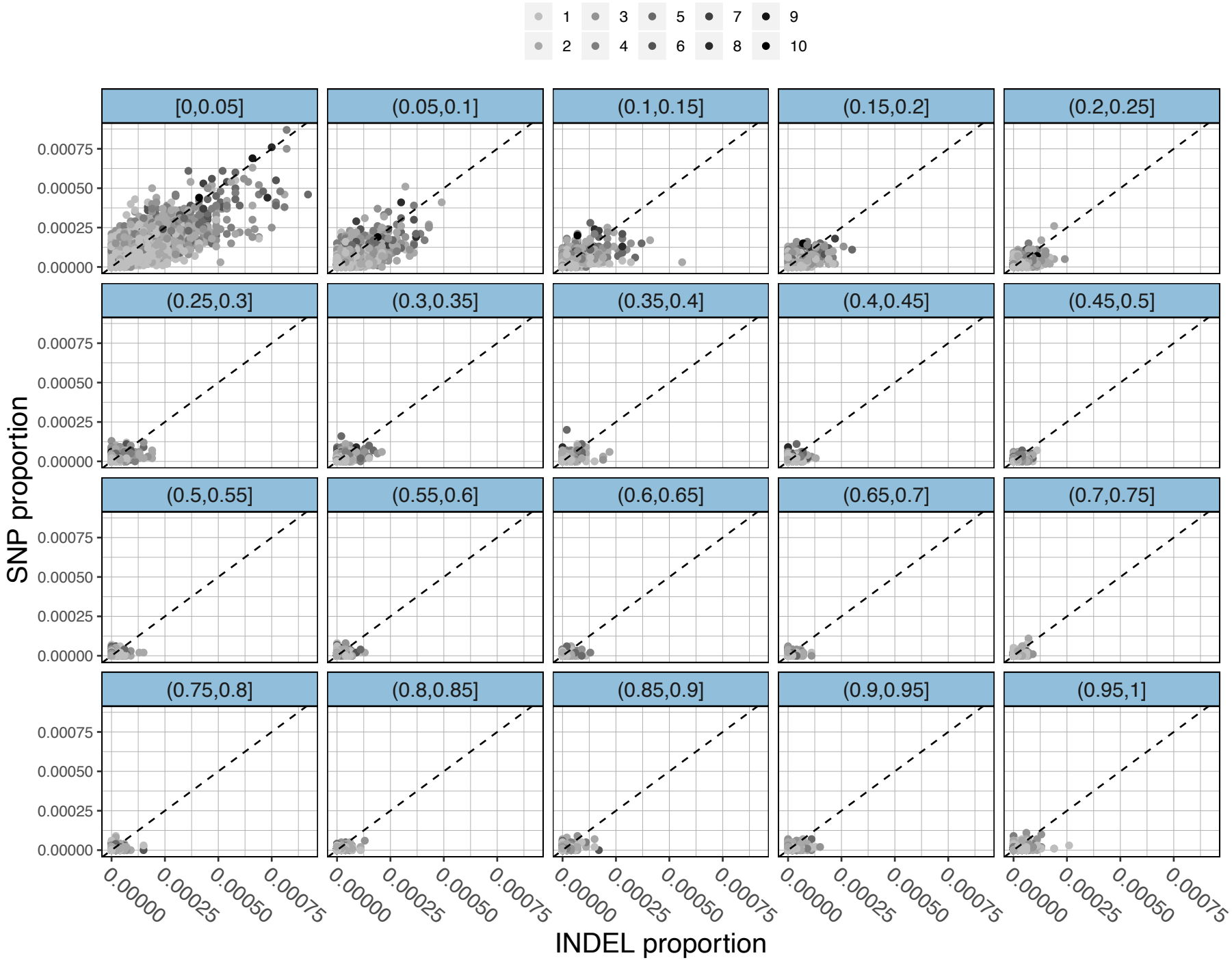


Figure 7. Proportions of SNPs against proportions of INDELs per contig and per derived allele frequency class. Contigs were grouped by length into ten bins of size = 50000 bp (from bin 1 in grey of range 0-50000 bp to bin 10 in black of range 450000-500000 bp). The derived frequency spectrum was divided into 20 classes of 0.05 frequency difference (facets).

3. Derived allele frequencies (GATK call)

- INDELs and SNPs after filtering but before cline analysis.
- All six hybrid zones combined.

- From the simple comparison between proportions of filtered SNPs and filtered INDELs (Fig. 7), we can further group variants by genomic location (coding vs. non-coding) and fitness effect (e.g., low impact for synonymous SNPs and intergenic INDELs and high impact for nonsynonymous SNPs and frameshift INDELs). In progress ...

- Similarly, we can compare INDELs-SNPs proporitons with respect to the cline parameters:
  - Centre (Fig. 8)
  - Width (Fig. 9)
  - Slope (Fig. 10)
  - Crab-Wave frequency difference (p_diff) (Fig. 11)
  - Variance explained (Var.Ex) (Fig. 12)

4. Distributions of cline parameters - centres (GATK call)

- Clinal INDELs and SNPs.
- All six hybrid zones combined.

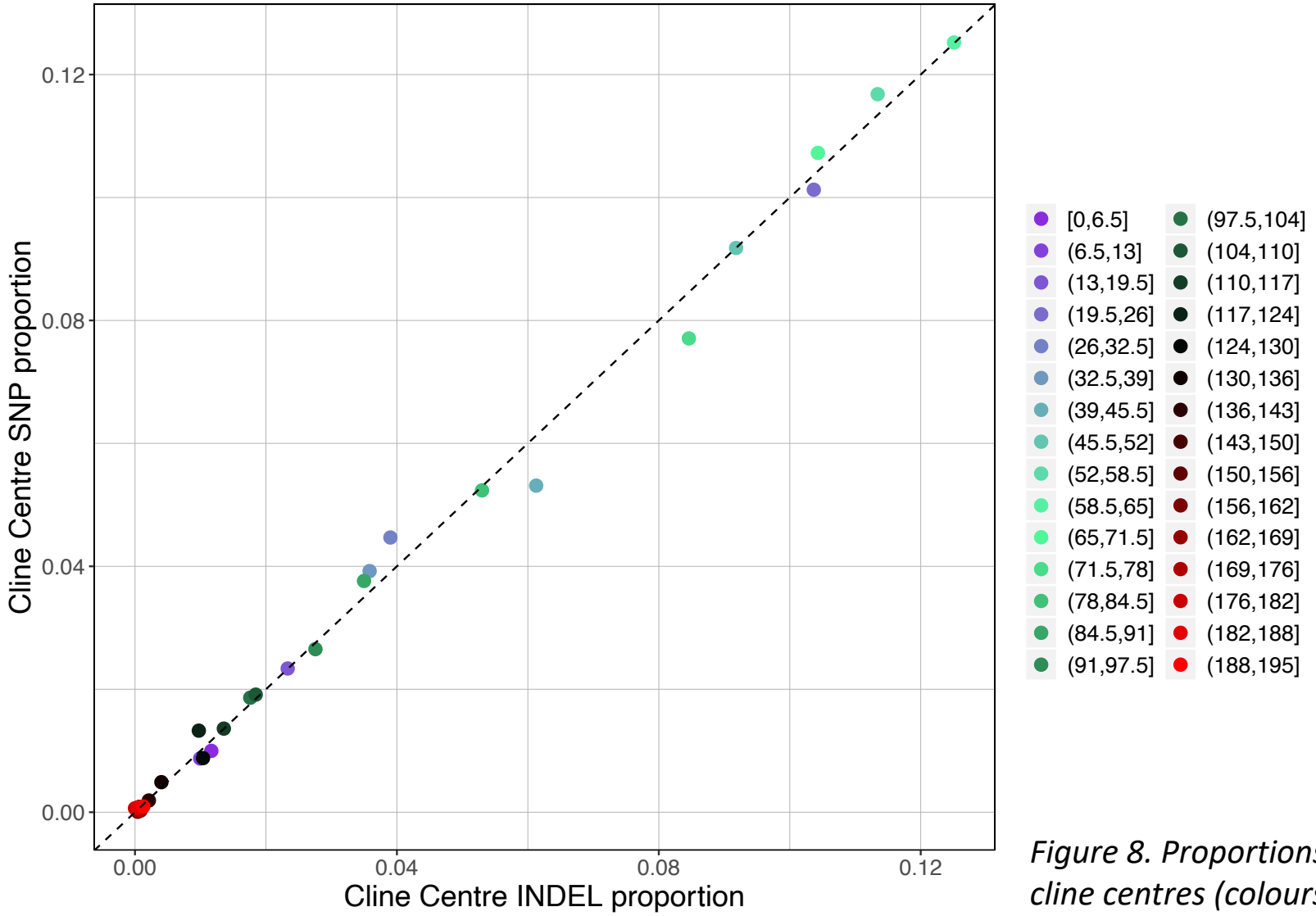


Figure 8. Proportions of SNPs vs INDELs for a given range of cline centres (colours).

4. Distributions of cline parameters - width (GATK call)

- Clinal INDELs and SNPs.
- All six hybrid zones combined.

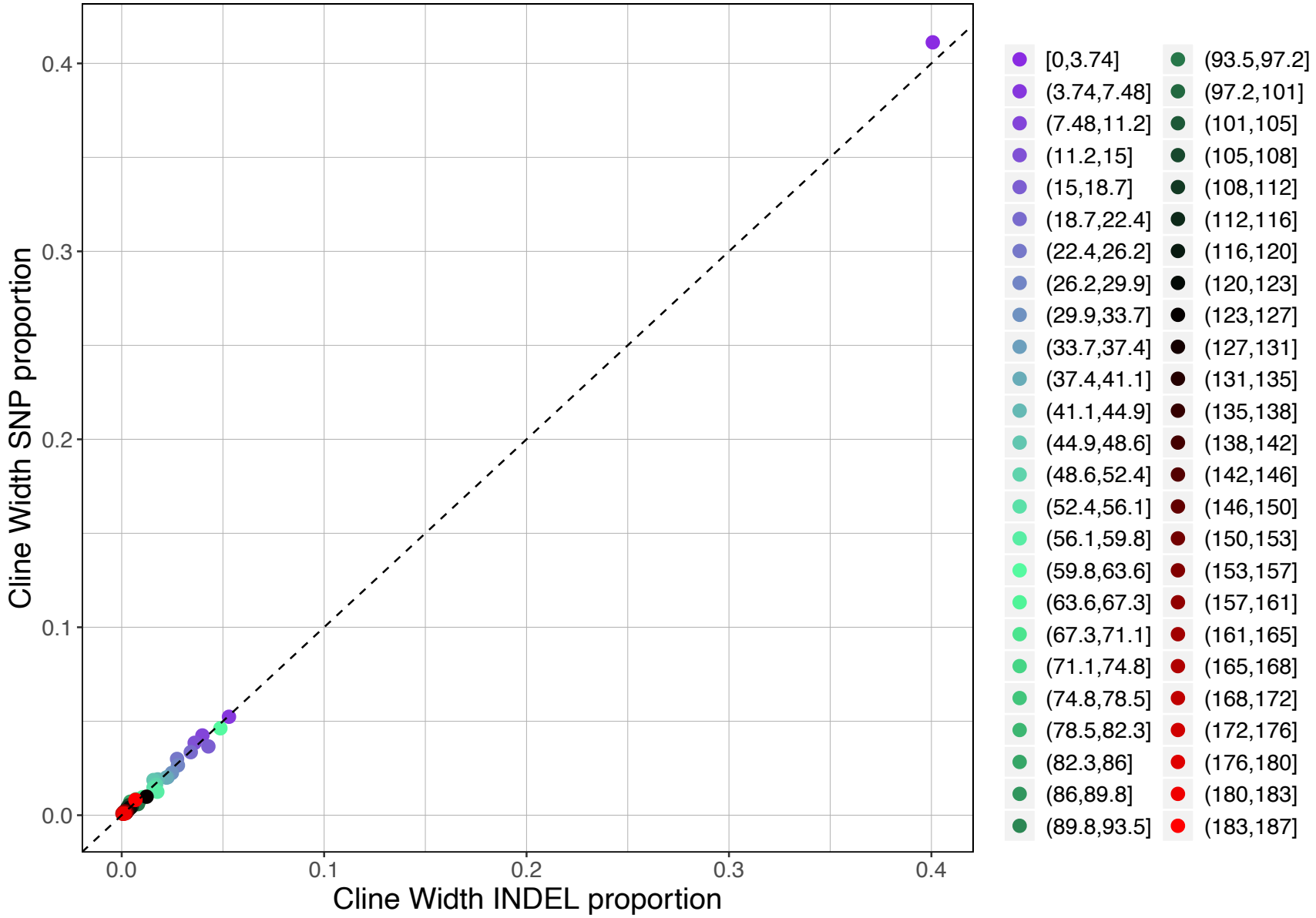


Figure 9. Proportions of SNPs vs INDELs for a given range of cline widths (colours).

4. Distributions of cline parameters - slope (GATK call)

- Clinal INDELs and SNPs.
- All six hybrid zones combined.

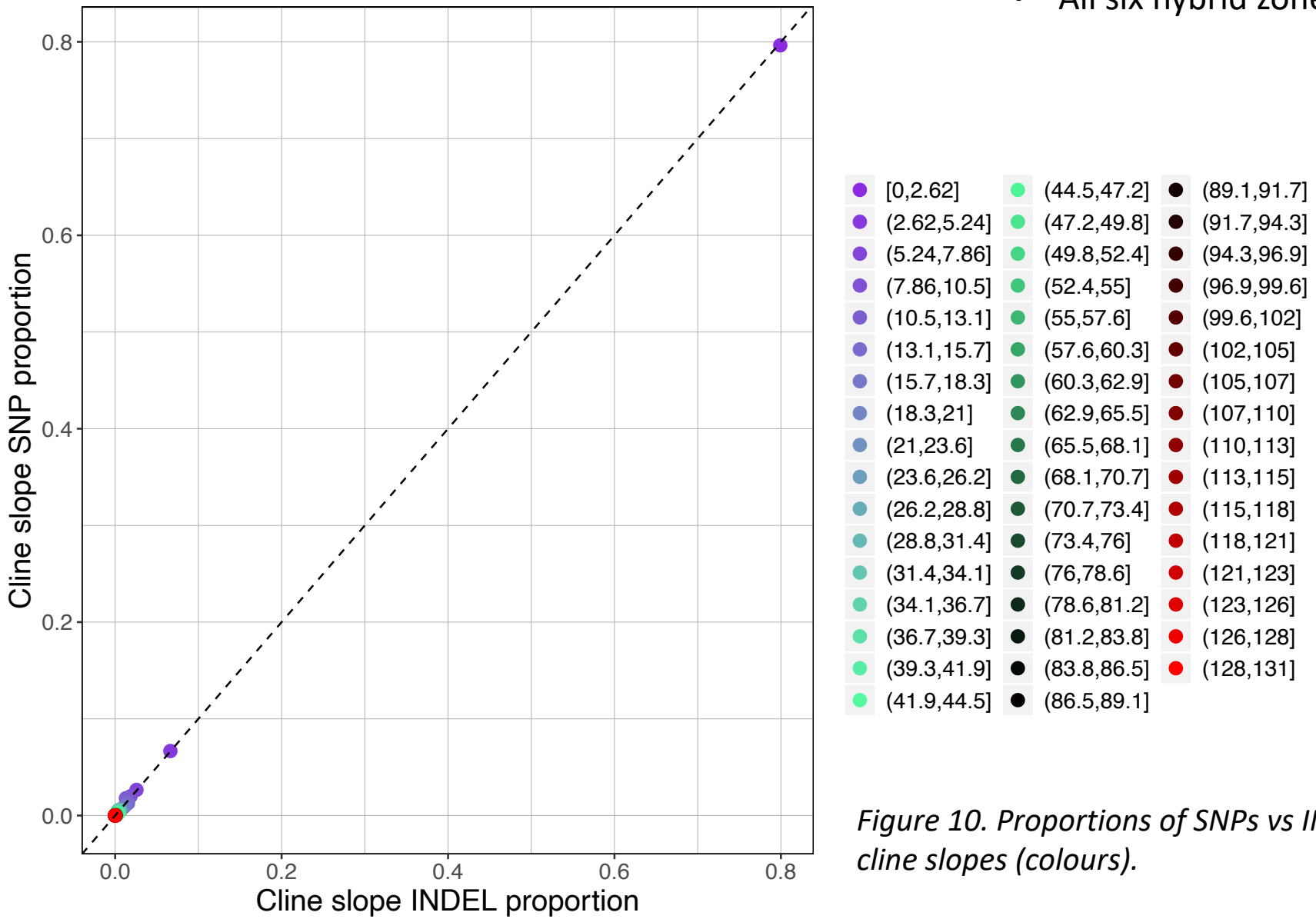


Figure 10. Proportions of SNPs vs INDELs for a given range of cline slopes (colours).

4. Distributions of cline parameters – p_diff (GATK call)

- Clinal INDELs and SNPs.
- All six hybrid zones combined.

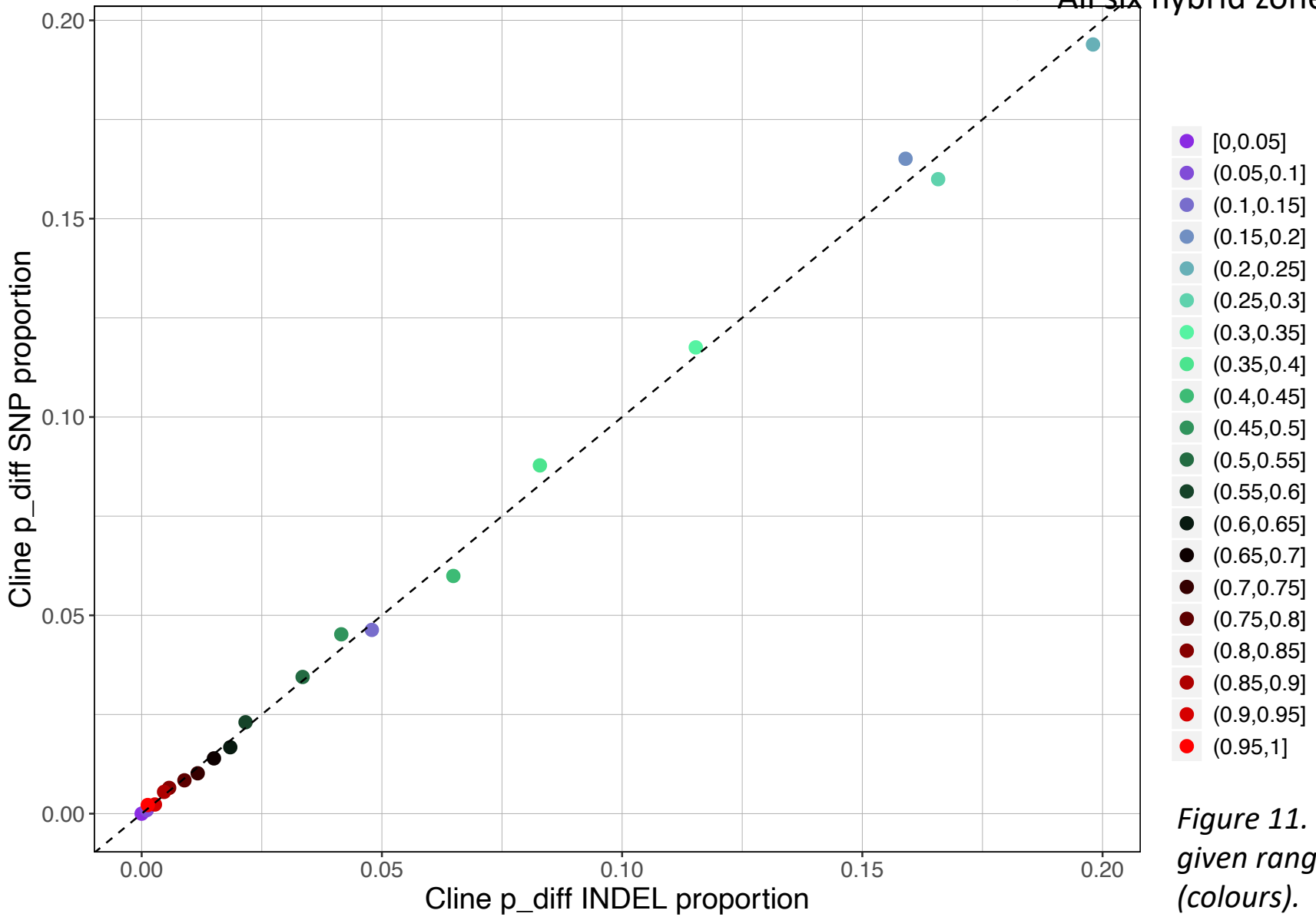


Figure 11. Proportions of SNPs vs INDELs for a given range of cline end frequencies difference (colours).

4. Distributions of cline parameters – Var.Ex (GATK call)

- Clinal INDELs and SNPs.
- All six hybrid zones combined.

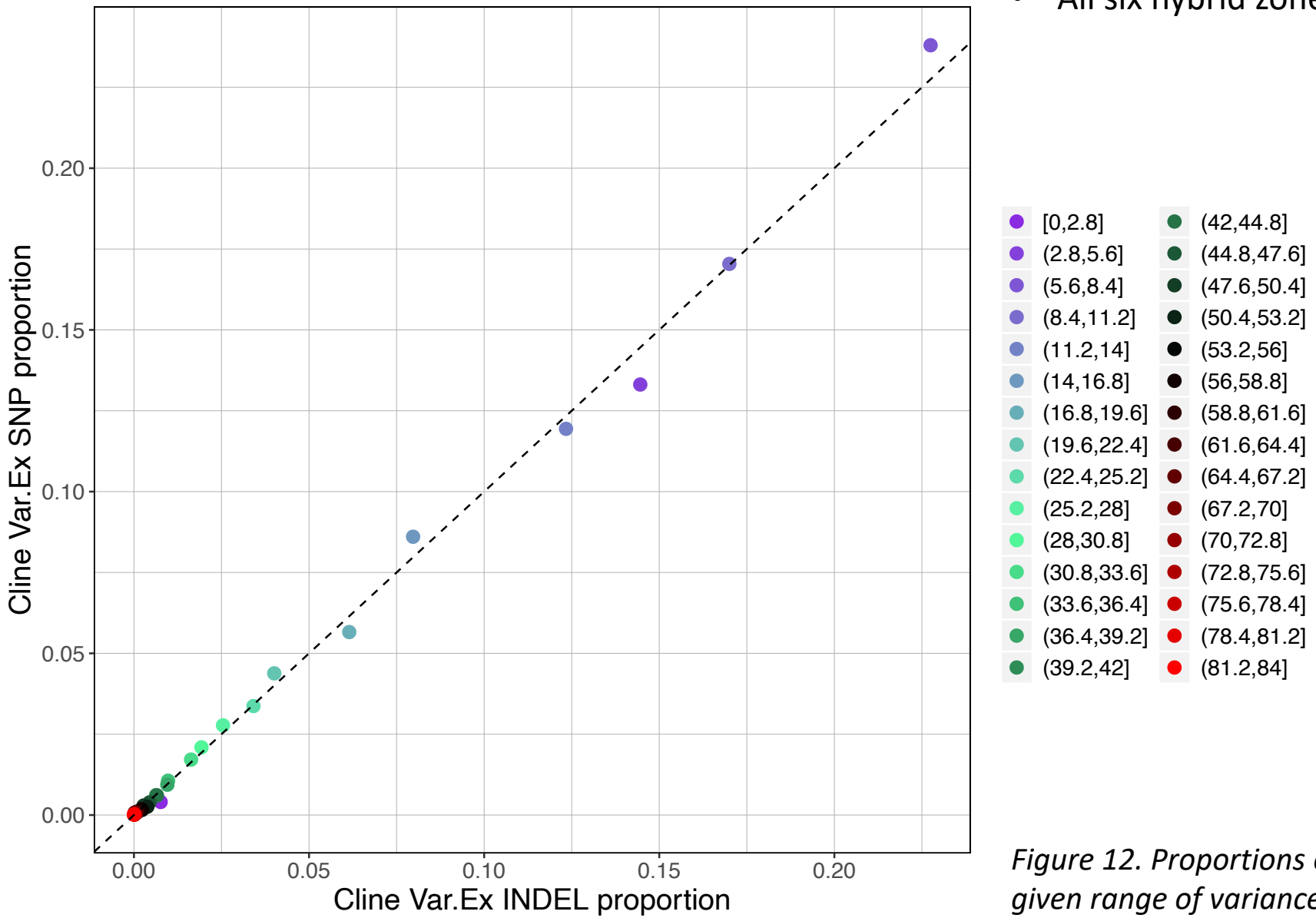


Figure 12. Proportions of SNPs vs INDELs for a given range of variance explained (colours).