DiscoSnp-RAD: de novo detection of small variants for population genomics

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ABSTRACT

Supplementary materials

Supplementary Table 1. *DiscoSnp-RAD* Computational resources on simulated data

# individuals	Wallclock Time (HH:MM:SS)	Memory Peak (GB)	Disk Peak (GB)
100	02:45:24	7.79	28.98
200	06:05:19	14.01	57.69
300	09:27:53	20.52	86.54
400	12:19:17	27.35	115.39
500	16:05:51	34.36	144.24
1000	41:36:29	71.84	288.49

Supplementary Table 2. Comparison of the number of loci identified by each tool

#		# output loci	
individuals	*DiscoSnp-RAD*	*IPyRAD*	*STACKS*
100	77,411	78,395	80,898
200	77,362	78,298	81,955
300	77,335	78,288	81,955
400	77,324	78,276	82,069
500	77,310	78,260	82,151
1000	77,264	78,193	82,511

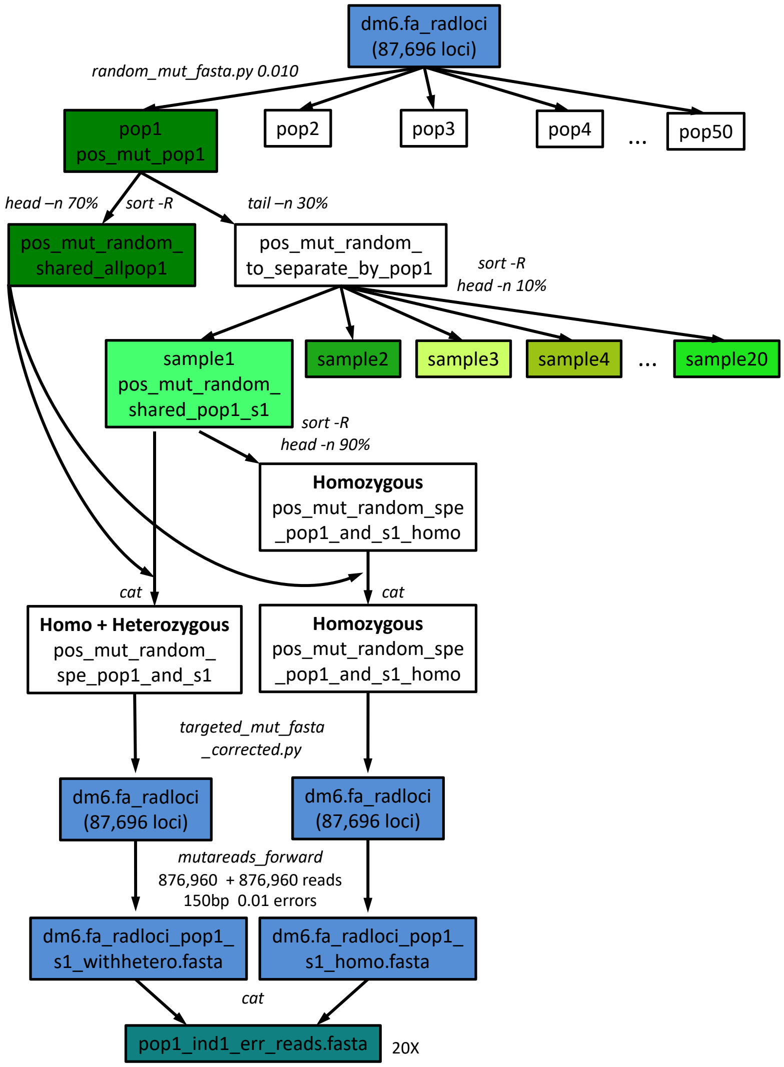
Supplementary Algorithm 2 (making use of Supplementary Algorithm 1) presents the detailed view of the SNPs and indels detection, including the symmetrical bubble detection and multiple SNPs per bubble.

Supplementary Algorithm 1 $bubble\_extension(path_1, path_2, nb\_sym\_branching, nb\_snps)$

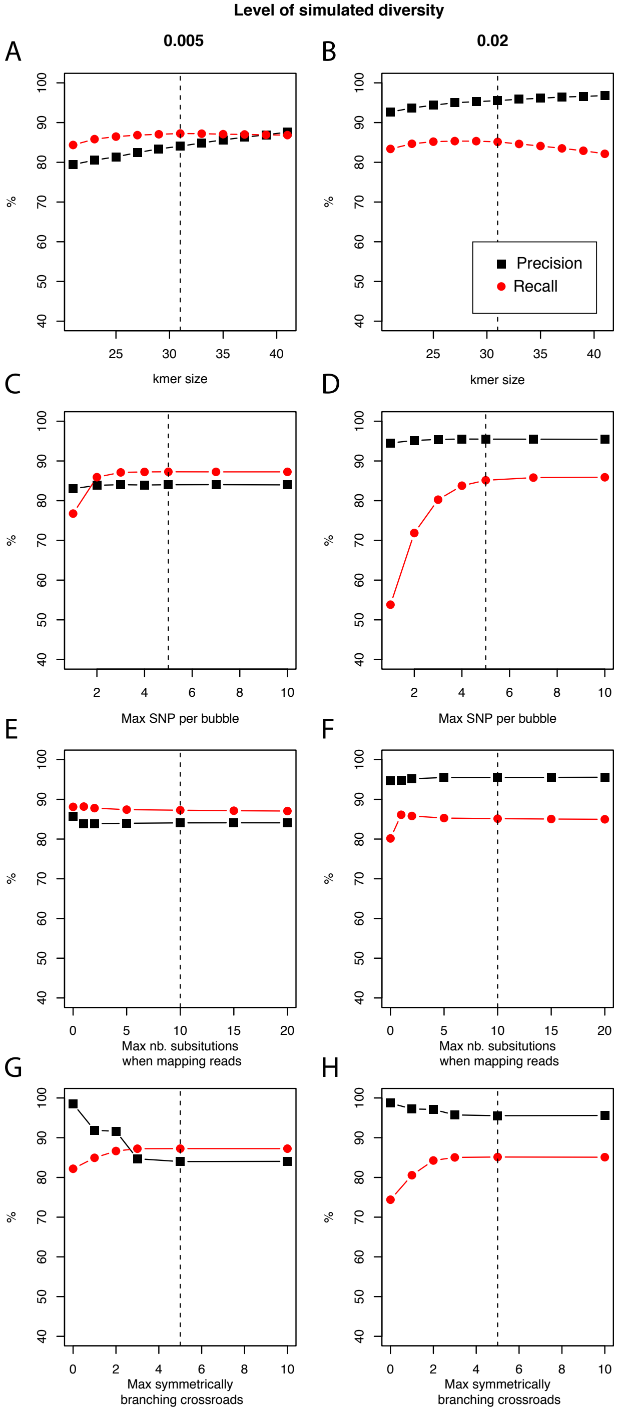
```
$kmer_1 =$ last $k$ nucleotides of $path_1$
$kmer_2 =$ last $k$ nucleotides of $path_2$
if $kmer_1$ equals $kmer_2$ then
    Output $path_1$,$path_2$
    return
$e_1 =$ set of last character of each children of $kmer_1$ ($e_1 \in \{A, C, G, T\}$)
$e_2 =$ set of last character of each children of $kmer_2$ ($e_2 \in \{A, C, G, T\}$)
if $|e_1| = 0$ and $|e_2| = 0$ then
    if last 3 characters from $kmer_1$ and $kmer_2$ are equal then
        Output $path_1$,$path_2$
        return
if $|e_1 \cap e_2| = 1$ then
    $bubble\_extension(path_1 + e_1 \cap e_2, path_2 + e_1 \cap e_2, nb\_sym\_branching, nb\_snps)$
    return
if $|e_1 \cap e_2| > 1$ then
    if $nb\_sym\_branching + 1 \geq max\_branching\_nodes$ then
        return
    for All $\alpha \in e_1 \cap e_2$ do
        $bubble\_extension(path_1 + \alpha, path_2 + \alpha, nb\_sym\_branching + 1, nb\_snps)$
    return
if $|e_1| = 1$ and $|e_2| = 1$ (thus $e1 \neq e2$) then
    if $nb\_snps + 1 \geq max\_snps$ then
        return
    $bubble\_extension(path_1 + e_1, path_2 + e_2, nb\_sym\_branching, nb\_snps + 1)$
    return
```

Supplementary Algorithm 2 *DiscoSnp-RAD* bubble detection for SNPs and indels detection

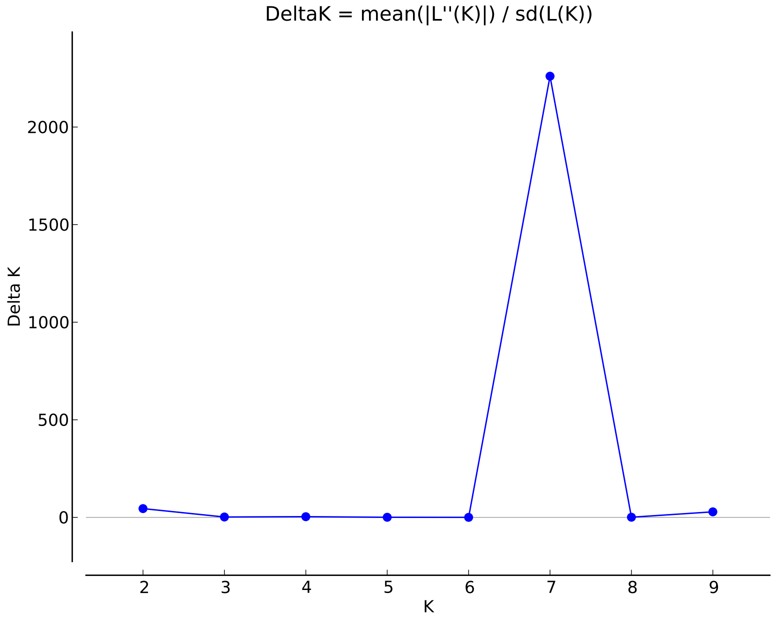
```
Create a de Bruijn Graph from all (any number $\geq 1$) read set(s)
for Each right branching $k$-mer in the graph $start$ do
    for each couple of successor $kmer_1, kmer_2$ of $k$-mer $start$ do
        //Snp detection
        $bubble\_extension(kmer_1, kmer_2, 0, 1)$
        //Indel detection from $kmer_1$
        for $d$ in [1, $max\_index\_size$] do
            Extend $kmer_1$ with $d$ nucleotides
            if last nucleotide of $kmer_1$ equals last nucleotide of $kmer_2$ then
                $bubble\_extension(kmer_1, kmer_2, 0, 1)$ with $max\_snps = 1$ (no close SNP with indels)
        //Indel detection from $kmer_2$
        for $d$ in [1, $max\_index\_size$] do
            Extend $kmer_2$ with $d$ nucleotides
            if last nucleotide of $kmer_2$ equals last nucleotide of $kmer_1$ then
                $bubble\_extension(kmer_1, kmer_2, 0, 1)$ with $max\_snps = 1$ (no close SNP with indels)
```



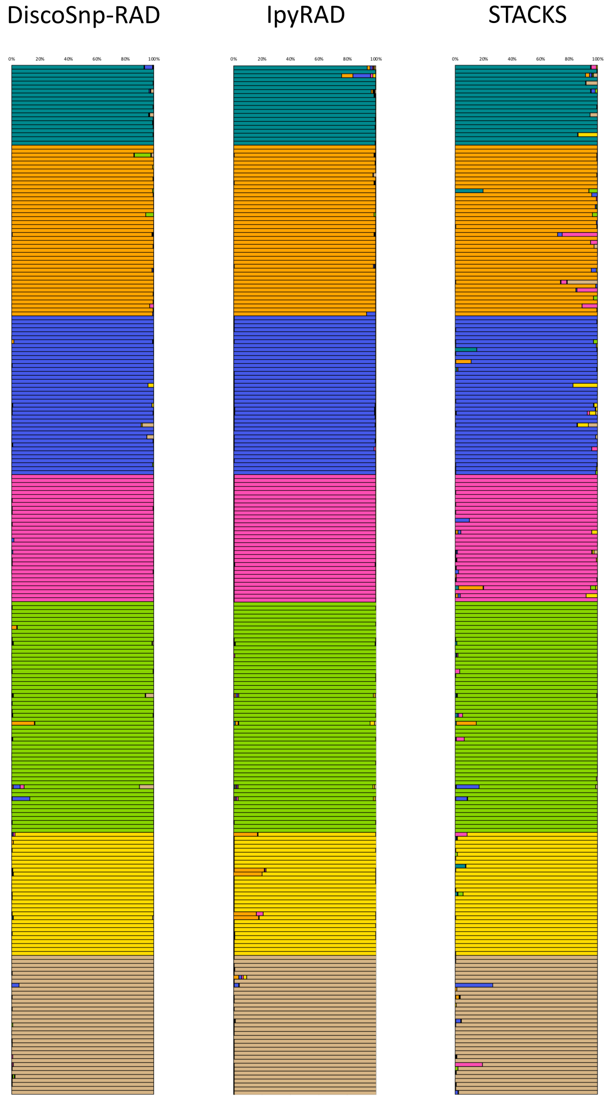
Supplementary Figure 1. Schematic representation of the pipeline designed and used to simulate RAD-Seq data from *D. melanogaster* genome. This pipeline includes RAD-Seq loci extraction, mutation simulation for various samples and populations and Illumina sequencing simulation.



Supplementary Figure 2. Recall and precision on simulated data of 100 samples with two levels of simulated diversity (0.5% and 2%) using *DiscoSnp-RAD* with respect to **A. B.** *k*-mer sizes, **C. D.** maximal number of authorized SNP per bubble, **E. F.** maximal number of authorized substitutions while mapping reads on predicted variants sequences, and **G. H.** maximal number of symmetrically branching crossroads. Dashed vertical line represents on each plot the chosen default value.



Supplementary Figure 3. Graph of the DeltaK distribution for all tested *K*, i.e. number of clusters.



Supplementary Figure 4. Comparison of STRUCTURE assignations for K=7 using SNPs obtained with *DiscoSnp-RAD*, *IPyRAD* and *STACKS* on all samples from the seven *Chiastocheta* species.