Structural Variant detection

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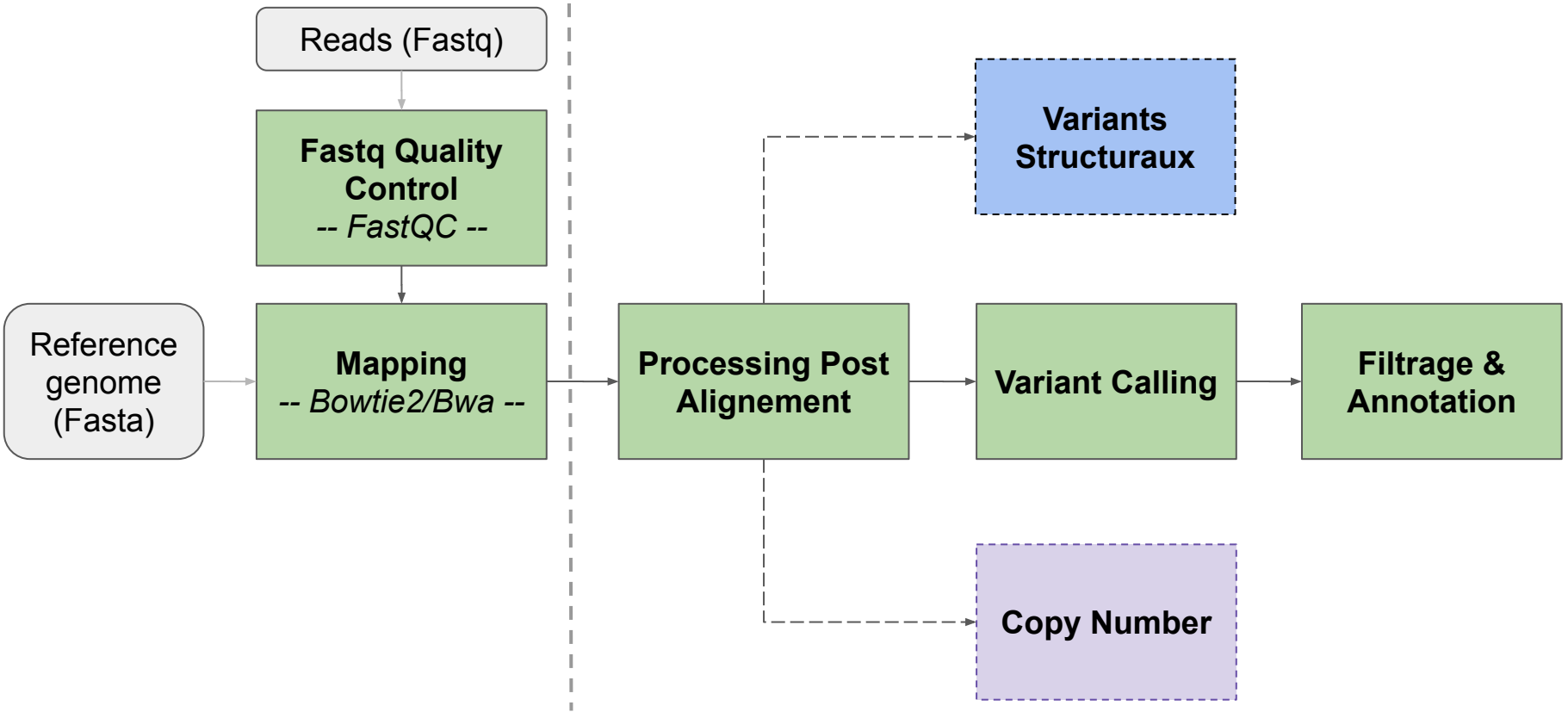
Workflow


Reads (Fastq)
Fastq Quality Control
-- FastQC --
Reference genome (Fasta)
Mapping
-- Bowtie2/Bwa --
Processing Post Alignement
Variants Structuraux
Variant Calling
Filtrage & Annotation
Copy Number

Définition

- Consensus actuel : Réarrangement génomique >50bp

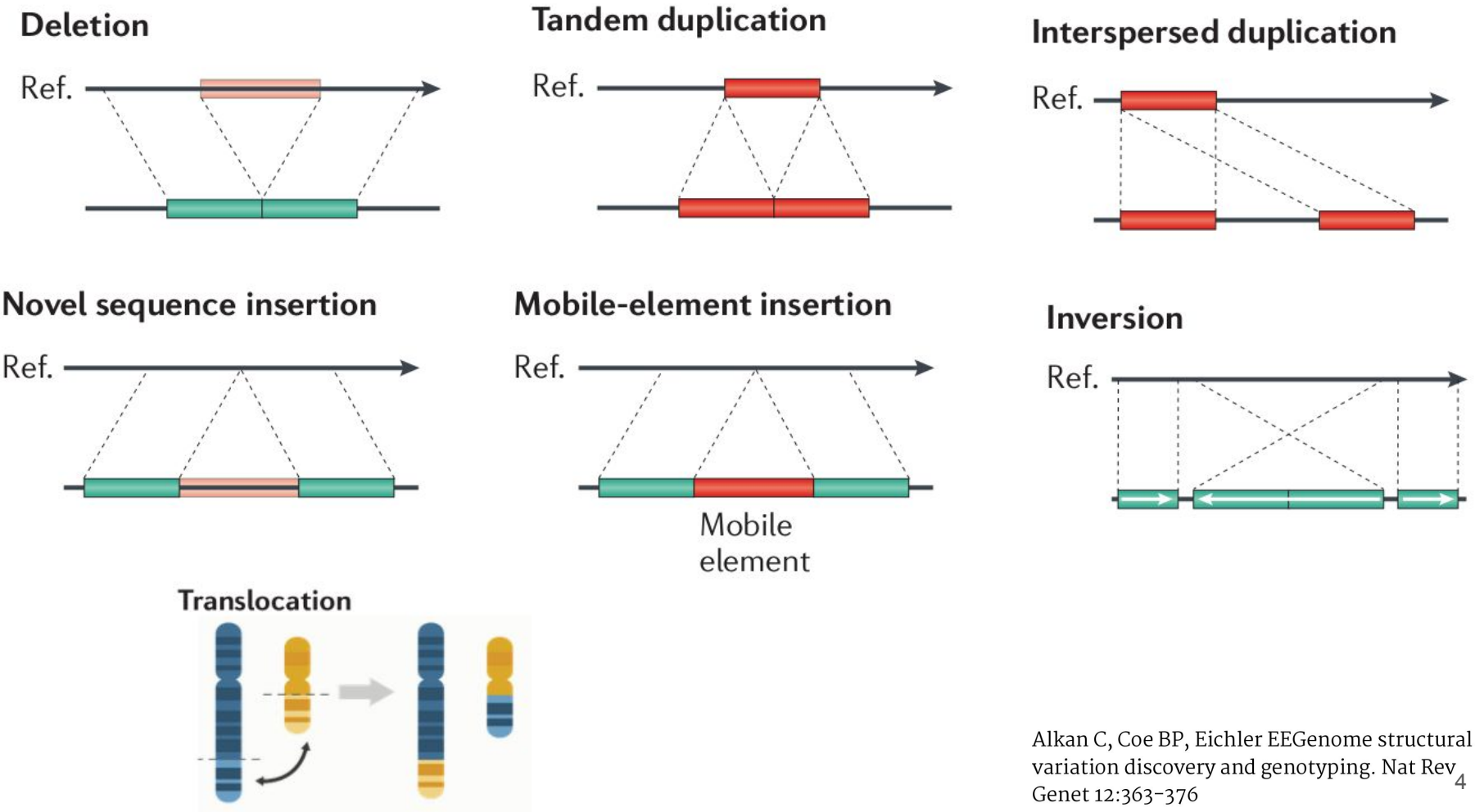
- Différents types de variants structuraux :

→ Réarrangements déséquilibrés (variation du nombre de copie - CNV)

  - Délétion
  - Duplication

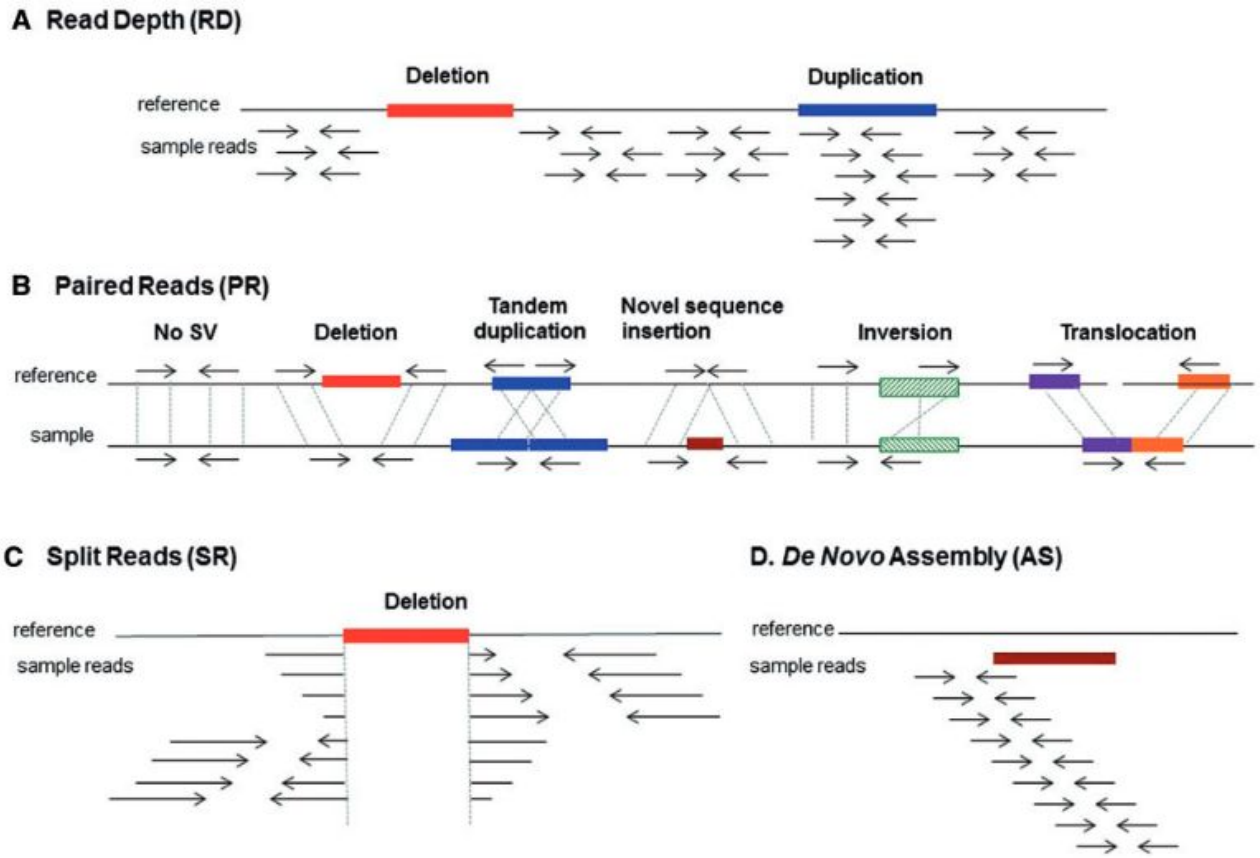
→ Réarrangements équilibrés

  - Insertion
  - Inversion
  - Translocation



Alkan C, Coe BP, Eichler EEGenome structural variation discovery and genotyping. Nat Rev Genet 12:363-376

Principe de détection des SVs



Review > Brief Funct Genomics. 2015 Sep;14(5):305-14. doi: 10.1093/bfgp/elv014. Epub 2015 Apr 15.

A decade of structural variants: description, history and methods to detect structural variation

Geòrgia Escaramís, Elisa Docampo, Raquel Rabionet

Short reads ou long reads?

Short reads (Illumina) : selon l'outil et la qualité des données

→ **faible recall** : 10 à 70% des SVs détectés
→ **faible précision** : jusqu'à 90% de Faux Positifs
→ Difficulté à caractériser des SVs complexes (alignement imprécis dans les régions répétées et faible résolution)

/!\ Un calling consensus avec plusieurs outils de détection peut être utile avec des données short reads /!\

Long reads (PacBio/MinION) :

→ Meilleure caractérisation des altérations des régions répétées
→ Une faible profondeur de couverture suffit (15-30x)

Sedlazeck, F. J. et al.Nat. Methods 15, 461–468 (2018).

Quel outil choisir ?

Critères de choix :

- Ai-je des données short reads ou long reads ?
- Ai-je de nombreux échantillons ?
- Quel type de SV est-ce que je recherche ?
- Est-ce que la profondeur de couverture est suffisante ?
- Que privilégier : sensibilité et / ou spécificité
- Quel est le format de sortie de l'outil ?

Détection de SV pour données **short reads**

	SV Callers	SV Types						Data	Anomalously Mapped Reads Used										Techniques				BP Resolution (Y/N)	References
									Discovery Stage					Validation Stage										
		CNV	INS	DEL	DUP	INV	TRA		RD	SC	PR	OEA	UM	RD	SC	PR	OEA	UM	CL	SA	CA	ST		
CNV	BIC-seq	x						PE;SE	x					x								x	N	[110]
	cn.MOPS	x						PE;SE	x					x								x	N	[44]
	cnD	x						PE	x					x								x	N	[88]
	CNVeM	x						PE	x					x								x	N	[105]
	CNVnator	x						PE;SE	x					x								x	N	[3]
	CNV-seq	x						PE;SE	x					x								x	N	[111]
	JointSLM	x						PE;SE	x					x								x	N	[59]
	RDXplorer	x						SE	x					x								x	N	[115]
	SegSeq	x						PE;SE	x					x								x	N	[15]
	CNVer	x						PE			x			x					x			x	N	[62]
SV	LUMPY			x	x	x	x	PE	x	x	x			x	x	x			x	x			N	[50]
	MetaSV		x	x	x	x	x	PE	x	x	x			x	x	x			x	x	x		Y	[65]
	SVM2		x	x				PE	x		x			x		x			x			x	N	[16]
	Breakpointer		x	x				SE	x					x	x					x		x	N	[95]
	Meerkat		x	x	x	x	x	PE		x	x	x			x		x		x	x			Y	[112,113]
	Scalpel		x	x				PE		x	x	x									x		Y	[68]
	SVMerge		x	x	x	x	x			x	x	x			x	x	x		x	x	x		Y	[109]
	SoftSV			x	x	x	x	PE		x	x				x	x			x	x			Y	[9]
	BreaKmer		x	x	x	x	x	PE		x		x				x					x		Y	[2]
	ClipCrop		x	x	x	x	x	PE		x					x				x	x			Y	[97]
	CREST			x	x	x	x	PE;SE		x					x						x		Y	[104]
	Gustaf			x	x	x	x	PE;SE		x					x					x			Y	[99]
	Socrates			x	x	x	x	PE;SE		x					x				x	x			Y	[86]
	Bellerophon						x	PE			x				x	x			x	x			Y	[30]
	BreakDancer		x	x	x	x	x	PE			x								x			x	N	[14]
	CLEVER		x	x				PE			x					x			x			x	N	[60]
	DELLY			x	x	x	x	PE			x				x		x		x	x			Y	[80]
	FACTERA			x			x	PE			x				x				x	x			Y	[69]
	GASV		x	x	x	x	x	PE			x								x				N	[90]
	GASVPro		x	x	x	x	x	PE			x			x		x			x			x	N	[91]
	GenomeSTRiP			x				PE			x					x			x			x	N	[29]
	HYDRA			x	x	x	x	PE			x					x			x				Y	[78]
	HYDRA-Multi			x	x	x	x	PE			x					x			x				Y	[58]
	inGAP-SV		x	x	x	x	x	PE			x			x									N	[76]
	MoDIL		x	x				PE			x					x						x	N	[51]
	PEMer		x	x	x	x	x	PE			x								x				N	[45]
	PeSV-Fisher			x	x	x	x	PE;MP			x			x					x				N	[21]
	PRISM		x	x	x	x	x	PE			x						x		x	x			Y	[37]
	RetroSeq		x					PE			x				x				x	x			Y	[40]
	SVDetect		x	x	x	x	x	PE;MP			x					x			x				N	[116]
	SVMiner		x	x		x		PE			x			x		x			x			x	N	[31]
	Ulysses		x	x	x	x	x	MP			x			x		x			x			x	N	[25]
	VariationHunter		x	x				PE			x					x			x				N	[32]
	NovelSeq		x					PE				x	x						x		x		Y	[27]
	PINDEL		x	x				PE				x								x			Y	[114]
	SLOPE		x	x			x	PE;SE				x					x		x	x			Y	[1]
	SOAPindel		x	x				PE				x			x	x	x				x		Y	[55]
	Splitread		x	x				PE				x								x			Y	[39]
	BreakSeq		x	x				PE							x					x			Y	[47]
	SMUFIN		x	x		x	x	PE											x		x		Y	[66]

P. Guan, W.-K. Sung / Methods 102 (2016) 36–49

Outils en long reads

- PBHoney, 2014
- SMRT-SV, 2015
- Hysa, 2016 (hybrid avec short reads)
- NanoSV, 2017
- Sniffles, 2018

Partie TP

Data : souche de *Zymoseptoria tritici* séquencées à la fois en Illumina et en MinION.

→ chaque set de reads a été aligné sur le génome de référence avec les outils dédiés

→ les données ont été réduites aux premiers 500kb du chr10

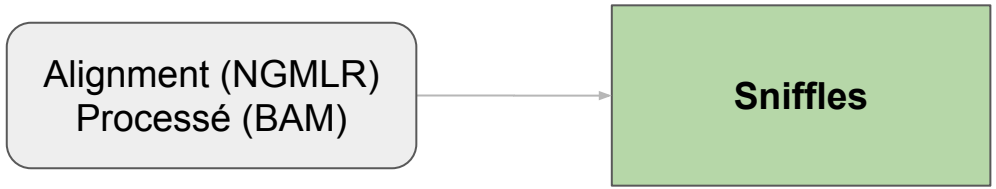
Tools :

- **Delly** *(Bioinformatics, Volume 28, Issue 18, 15 September 2012, Pages i333-i339, https://doi.org/10.1093/bioinformatics/bts378)*
- **Sniffles** *(Nature Methods volume 15, pages 461-468 (2018) , https://www.nature.com/articles/s41592-018-0001-7)* with **NGMLR** mapping

Workflow - Variants Structuraux

Short Reads (Illumina)

Long Reads (MinION)



Préparation des données

```
# Copie des données SV
$ cp -R /shared/projects/ebai2019/atelier_variant/data/sv ~/tp_sv
$ cd ~/tp_sv

# Indexation des fichiers
$ module load samtools/1.10

$ srun samtools index mapping_illumina_chr10_500kb.bam
$ srun samtools index mapping_minion_chr10_500kb.bam
$ srun samtools faidx Zymoseptoria_tritici.fa

$ ls -l
```

```
13812904 Oct 28 14:41 mapping_illumina_chr10_500kb.bam
    1720 Oct 28 14:51 mapping_illumina_chr10_500kb.bam.bai
43323244 Oct 28 14:41 mapping_minion_chr10_500kb.bam
    9040 Oct 28 14:51 mapping_minion_chr10_500kb.bam.bai
40348870 Oct 28 14:41 Zymoseptoria_tritici.fa
     606 Oct 28 14:44 Zymoseptoria_tritici.fa.fai
```

Delly

```
$ mkdir -p delly/logs
$ cd delly

$ module load delly/0.8.3
$ delly      # (v0.8.3)
$ delly call

Usage: delly call [OPTIONS] -g <ref.fa> <sample1.sort.bam> <sample2.sort.bam> ...

Generic options:
  -? [ --help ]                    show help message
  -t [ --svtype ] arg (=ALL)       SV type to compute [DEL, INS, DUP, INV,
                                   BND, ALL]
  -g [ --genome ] arg              genome fasta file
  -x [ --exclude ] arg             file with regions to exclude
  -o [ --outfile ] arg (="sv.bcf")  SV BCF output file
```

Delly

```
$ sbatch -J delly -o logs/delly.out -e logs/delly.err --mem=8G --wrap=" \
    delly call -g ~/tp_sv/Zymoseptoria_tritici.fa \
    -o SV_calling_illumina.bcf ~/tp_sv/mapping_illumina_chr10_500kb.bam"
```

```
$ less SV_calling_illumina.bcf
# "delly/SV_calling_illumina.bcf" may be a binary file.  See it anyway? n
```

```
# Conversion en fichier vcf
$ module load bcftools/1.10.2

$ sbatch -J bcf_to_vcf -o logs/bcf_to_vcf.out -e logs/bcf_to_vcf.err --wrap=" \
    bcftools view SV_calling_illumina.bcf > SV_calling_illumina.vcf"

$ less -S SV_calling_illumina.vcf      # “Q” pour quitter
```

Header du vcf de Delly

```
##fileformat=VCFv4.2
##FILTER=<ID=PASS,Description="All filters passed">
##fileDate=20200804
##ALT=<ID=DEL,Description="Deletion">
##ALT=<ID=DUP,Description="Duplication">
##ALT=<ID=INV,Description="Inversion">
##ALT=<ID=BND,Description="Translocation">
##ALT=<ID=INS,Description="Insertion">
##FILTER=<ID=LowQual,Description="Poor quality and insufficient number of PEs and SRs.">
##INFO=<ID=CIEND,Number=2,Type=Integer,Description="PE confidence interval around END">
##INFO=<ID=CIPOS,Number=2,Type=Integer,Description="PE confidence interval around POS">
##INFO=<ID=CHR2,Number=1,Type=String,Description="Chromosome for POS2 coordinate in case of an inter-chromosomal translocation">
##INFO=<ID=POS2,Number=1,Type=Integer,Description="Genomic position for CHR2 in case of an inter-chromosomal translocation">
##INFO=<ID=END,Number=1,Type=Integer,Description="End position of the structural variant">
##INFO=<ID=PE,Number=1,Type=Integer,Description="Paired-end support of the structural variant">
##INFO=<ID=MAPQ,Number=1,Type=Integer,Description="Median mapping quality of paired-ends">
##INFO=<ID=SRMAPQ,Number=1,Type=Integer,Description="Median mapping quality of split-reads">
##INFO=<ID=SR,Number=1,Type=Integer,Description="Split-read support">
##INFO=<ID=SRQ,Number=1,Type=Float,Description="Split-read consensus alignment quality">
##INFO=<ID=CONSENSUS,Number=1,Type=String,Description="Split-read consensus sequence">
##INFO=<ID=CE,Number=1,Type=Float,Description="Consensus sequence entropy">
##INFO=<ID=CT,Number=1,Type=String,Description="Paired-end signature induced connection type">
##INFO=<ID=SVLEN,Number=1,Type=Integer,Description="Insertion length for SVTYPE=INS.">
##INFO=<ID=IMPRECISE,Number=0,Type=Flag,Description="Imprecise structural variation">
##INFO=<ID=PRECISE,Number=0,Type=Flag,Description="Precise structural variation">
##INFO=<ID=SVTYPE,Number=1,Type=String,Description="Type of structural variant">
##INFO=<ID=SVMETHOD,Number=1,Type=String,Description="Type of approach used to detect SV">
##INFO=<ID=INSLEN,Number=1,Type=Integer,Description="Predicted length of the insertion">
##INFO=<ID=HOMLEN,Number=1,Type=Integer,Description="Predicted microhomology length using a max. edit distance of 2">
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
##FORMAT=<ID=GL,Number=G,Type=Float,Description="Log10-scaled genotype likelihoods for RR,RA,AA genotypes">
##FORMAT=<ID=GQ,Number=1,Type=Integer,Description="Genotype Quality">
##FORMAT=<ID=FT,Number=1,Type=String,Description="Per-sample genotype filter">
##FORMAT=<ID=RC,Number=1,Type=Integer,Description="Raw high-quality read counts or base counts for the SV">
##FORMAT=<ID=RCL,Number=1,Type=Integer,Description="Raw high-quality read counts or base counts for the left control region">
##FORMAT=<ID=RCR,Number=1,Type=Integer,Description="Raw high-quality read counts or base counts for the right control region">
##FORMAT=<ID=CN,Number=1,Type=Integer,Description="Read-depth based copy-number estimate for autosomal sites">
##FORMAT=<ID=DR,Number=1,Type=Integer,Description="# high-quality reference pairs">
##FORMAT=<ID=DV,Number=1,Type=Integer,Description="# high-quality variant pairs">
##FORMAT=<ID=RR,Number=1,Type=Integer,Description="# high-quality reference junction reads">
##FORMAT=<ID=RV,Number=1,Type=Integer,Description="# high-quality variant junction reads">
##reference=Zymoseptoria_tritici.fa
##contig=<ID=chr_1,length=6088797>
```

Delly : comptage du nombre de SVs

```
# Combien de variants ?
$ grep -v -c "^#" SV_calling_illumina.vcf
```

```
# Combien de variants de bonne qualité ?
$ grep -v "^#" SV_calling_illumina.vcf | grep -v -c "LowQual"
```

Delly : comptage du nombre de SVs

```
# Combien de variants de bonne qualité de type Deletion…

$ grep -v "^#" SV_calling_illumina.vcf | grep -v "LowQual" | grep -c "<DEL>"

$ grep -v "^#" SV_calling_illumina.vcf | grep -v "LowQual" | grep -c "<DUP>"

$ grep -v "^#" SV_calling_illumina.vcf | grep -v "LowQual" | grep -c "<INV>"

$ grep -v "^#" SV_calling_illumina.vcf | grep -v "LowQual" | grep -c "<BND>"

$ grep -v "^#" SV_calling_illumina.vcf | grep -v "LowQual" | grep -c "<INS>"
```

Delly : extraction des informations

```
$ grep -v "^#" SV_calling_illumina.vcf | grep -v "LowQual" | grep "<DEL>"
```

```
chr_10  29522   DEL00000002     A       <DEL>   1200    PASS    PRECISE;SVTYPE=DEL;SVMETHOD=EMBL.DELLYv0.8.
3;END=29580;PE=0;MAPQ=0;CT=3to5;CIPOS=-3,3;CIEND=-3,3;SRMAPQ=60;INSLEN=0;HOMLEN=2;SR=20;SRQ=1;CONSENSUS=AAG
TGTCTCGACCAGGTCGAGAGGGAAACGTAGAAGGGCGAAGTGGATGAGGAGAGGAGAAGGAAGAGGAGGCTTCTGCAAAGTCTGAGTCCGTGGTCAAGGTCTTCCAA
CGGTACTGTCACGGGCTCGCCAGATGTTCATGAATTTCAGACCCCGATGTACGTGAATTCTATTTACGAAGAACTACCAGTCTTGCAAGACTCCAACCTAA;CE=1.
98003       GT:GL:GQ:FT:RCL:RC:RCR:CN:DR:DV:RR:RV   1/1:-109.497,-9.02787,0:90:PASS:612:32:745:0:0:0:0:30
chr_10  32733   DEL00000003     C       <DEL>   1200    PASS    PRECISE;SVTYPE=DEL;SVMETHOD=EMBL.DELLYv0.8.
3;END=32783;PE=0;MAPQ=0;CT=3to5;CIPOS=-2,2;CIEND=-2,2;SRMAPQ=60;INSLEN=0;HOMLEN=1;SR=20;SRQ=1;CONSENSUS=ATG
CACAACGCAGACTCGTGCAGCCGCTACACTGGCAACACCGACAGGAAAACGTTCTTTACATAGACCAGTCGTGTTCGGCATCTACCCGGCCGTTTTCTGTAATCATC
CTAGCCGTTTCCCGTATGGCTCGAGGGCTTTTTCTGGATCTTGGGCGTTTTCCATATGGCTTGCCGTTGTCCCTATGGCTGGATGG;CE=1.97989      GT:G
L:GQ:FT:RCL:RC:RCR:CN:DR:DV:RR:RV   1/1:-152.993,-12.937,0:129:PASS:686:41:792:0:0:0:0:43
```

Delly : extraction des informations des délétions

```
#Récupération du start des variants
$ grep -v "^#" SV_calling_illumina.vcf | grep -v "LowQual" | grep "<DEL>" | \
    cut -f1,2 > delly_del_start.txt
```

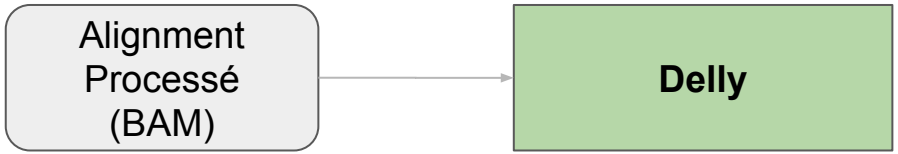
```
#Récupération des autres informations
$ grep -v "^#" SV_calling_illumina.vcf | grep -v "LowQual" | grep "<DEL>" | \
    cut -f8 | cut -d ";" -f1,4,5,13 | sed "s/;/\t/g" > delly_del_info.txt
```

```
#Fusion des deux fichiers
$ paste -d '\t' delly_del_start.txt delly_del_info.txt > delly_del.txt
```

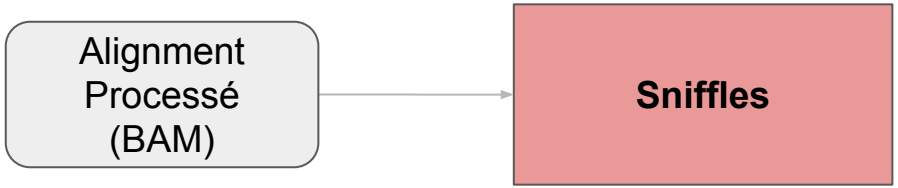
```
#Formattage et ménage
$ awk '{print $1"\t"$2"\t"$4"\t"$3"\t"$5"\t"$6}' delly_del.txt | sed "s/END=//g" \
    > delly_del.csv
$ rm delly_del_info.txt delly_del_start.txt delly_del.txt
```

Workflow - Variants Structuraux

Short Reads (Illumina)



Long Reads (MinION)



Détection de données long reads avec Sniffles

```
$ module load sniffles/1.0.11
$ sniffles --help
Usage: sniffles [options] -m <sorted.bam> -v <output.vcf>
Version: 1.0.11
Contact: fritz.sedlazeck@gmail.com

Input/Output:
     -m <string>,  --mapped_reads <string>
     (required)  Sorted bam File
     -v <string>,  --vcf <string>
     VCF output file name []
     -b <string>,  --bedpe <string>
          bedpe output file name []
     --Ivcf <string>
     Input VCF file name. Enable force calling []
     --tmp_file <string>
     path to temporary file otherwise Sniffles will use the current directory. []
( -l <int>,  --min_length <int>
     Minimum length of SV to be reported. [30] )
```

Sniffles

```
$ mkdir -p ~/tp_sv/sniffles/logs
$ cd ~/tp_sv/sniffles

$ sbatch -J sniffles -o logs/sniffles.out -e logs/sniffles.err --mem=8G --wrap=" \
    sniffles -l 100 -m ~/tp_sv/mapping_minion_chr10_500kb.bam \
    -v SV_calling_minion.vcf"

$ less -S SV_calling_minion.vcf
```

Header du vcf de Sniffles

```
##fileformat=VCFv4.3
##source=Sniffles
##fileDate=20191028
##contig=<ID=chr_1,length=6088797>
##contig=<ID=chr_2,length=3860111>
##contig=<ID=chr_3,length=3505381>
##contig=<ID=chr_4,length=2880011>
##contig=<ID=chr_5,length=2861803>
##contig=<ID=chr_6,length=2674951>
##contig=<ID=chr_7,length=2665280>
##contig=<ID=chr_8,length=2443572>
##contig=<ID=chr_9,length=2142475>
##contig=<ID=chr_10,length=1682575>
##contig=<ID=chr_11,length=1624292>
##contig=<ID=chr_12,length=1462624>
##contig=<ID=chr_13,length=1185774>
##contig=<ID=chr_14,length=773098>
##contig=<ID=chr_15,length=639501>
##contig=<ID=chr_16,length=607044>
##contig=<ID=chr_17,length=584099>
##contig=<ID=chr_18,length=573698>
##contig=<ID=chr_19,length=549847>
##contig=<ID=chr_20,length=472105>
##contig=<ID=chr_21,length=409213>
##ALT=<ID=DEL,Description="Deletion">
##ALT=<ID=DUP,Description="Duplication">
##ALT=<ID=INV,Description="Inversion">
##ALT=<ID=INVDUP,Description="InvertedDUP with unknown boundaries">
##ALT=<ID=TRA,Description="Translocation">
##ALT=<ID=INS,Description="Insertion">
##INFO=<ID=CHR2,Number=1,Type=String,Description="Chromosome for END coordinate in case of a translocation">
##INFO=<ID=END,Number=1,Type=Integer,Description="End position of the structural variant">
##INFO=<ID=MAPQ,Number=1,Type=Integer,Description="Median mapping quality of paired-ends">
##INFO=<ID=RE,Number=1,Type=Integer,Description="read support">
##INFO=<ID=IMPRECISE,Number=0,Type=Flag,Description="Imprecise structural variation">
##INFO=<ID=PRECISE,Number=0,Type=Flag,Description="Precise structural variation">
##INFO=<ID=UNRESOLVED,Number=0,Type=Flag,Description="An insertion that is longer than the read and thus we cannot predict the full size.">
```

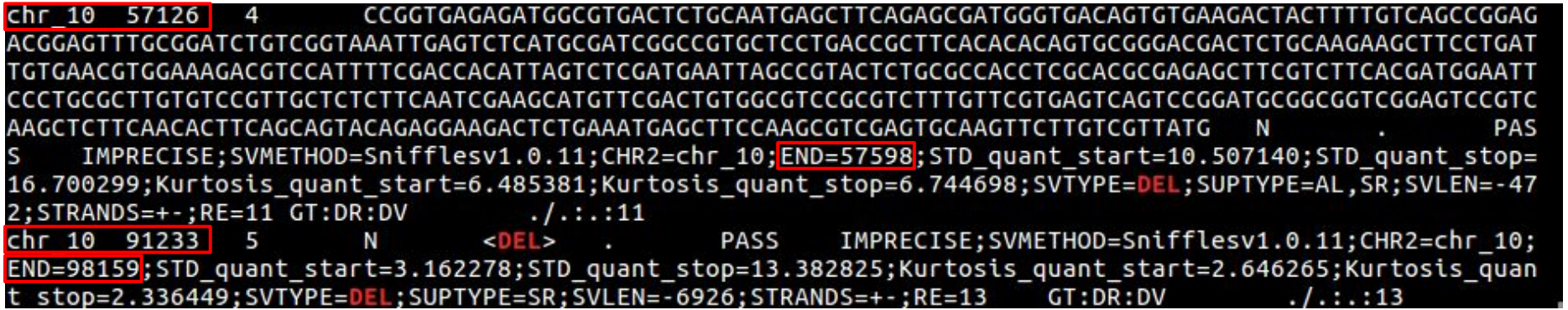
Sniffles : comptage du nombre de SVs

```
$ cat SV_calling_minion.vcf | grep ^chr_10 | wc -l
```

```
$ cat SV_calling_minion.vcf | grep ^chr_10 | grep "DEL" | wc -l

$ cat SV_calling_minion.vcf | grep ^chr_10 | grep "DUP" | wc -l

$ cat SV_calling_minion.vcf | grep ^chr_10 | grep "INV" | wc -l

$ cat SV_calling_minion.vcf | grep ^chr_10 | grep "TRA" | wc -l

$ cat SV_calling_minion.vcf | grep ^chr_10 | grep "INS" | wc -l
```

Sniffles : extraction des positions des délétions

```
$ cat SV_calling_minion.vcf | grep ^chr_10 | grep "DEL"
```



Sniffles : extraction des positions des délétions

revoir les commandes, DEL peuvent apparaître différemment

```
$ cat SV_calling_minion.vcf | grep ^chr_10 | grep "DEL" | cut -f -2 \
    > sniffles_del_start.txt
```

```
$ cat SV_calling_minion.vcf | grep ^chr_10 | grep "DEL" | cut -d ";" -f 4 | \
    cut -d "=" -f 2 > sniffles_del_stop.txt
```

```
$ cat SV_calling_minion.vcf | grep ^chr_10 | grep "DEL" | cut -f 8 | \
    cut -d ";" -f 1 > sniffles_del_infos.txt
```

```
$ paste sniffles_del_start.txt sniffles_del_stop.txt sniffles_del_infos.txt \
    > sniffles_del.csv
```

```
$ rm sniffles_del_start.txt sniffles_del_stop.txt sniffles_del_infos.txt
```

Comparaison des résultats de Delly et Sniffles

```
$ cd ~/tp_sv
$ cat delly/delly_del.csv
$ cat sniffles/sniffles_del.csv
```

Delly (Illumina)				
Start	End	precision	PairEnd	Split Reads
29522	29580	PRECISE	0	20
32733	32783	PRECISE	0	20
57127	57600	PRECISE	3	16
80015	80622	PRECISE	15	20
90255	90309	PRECISE	0	7
90309	101040	IMPRECISE	8	0
111021	111676	IMPRECISE	20	0
191291	191343	PRECISE	0	20
-	-	-	-	-
264986	265063	PRECISE	0	12
267829	267857	PRECISE	0	19
-	-	-	-	-
360628	361052	PRECISE	0	20
383682	477911	IMPRECISE	7	0
425686	426624	IMPRECISE	28	0
459094	459124	PRECISE	0	12
465858	466080	PRECISE	0	20
468192	468342	PRECISE	0	20
477523	479732	PRECISE	41	20
496882	496919	PRECISE	0	20

Sniffles (Minion)		
Start	End	precision
-	-	-
-	-	-
57126	57598	IMPRECISE
-	-	-
-	-	-
91233	98159	IMPRECISE
111020	111655	PRECISE
-	-	-
257001	257165	IMPRECISE
-	-	-
-	-	-
343161	343273	PRECISE
360638	361061	PRECISE
383681	477805	IMPRECISE
425682	426487	IMPRECISE
-	-	-
-	-	-
468192	468341	PRECISE
477525	479731	PRECISE
-	-	-

Visualisation sous IGV

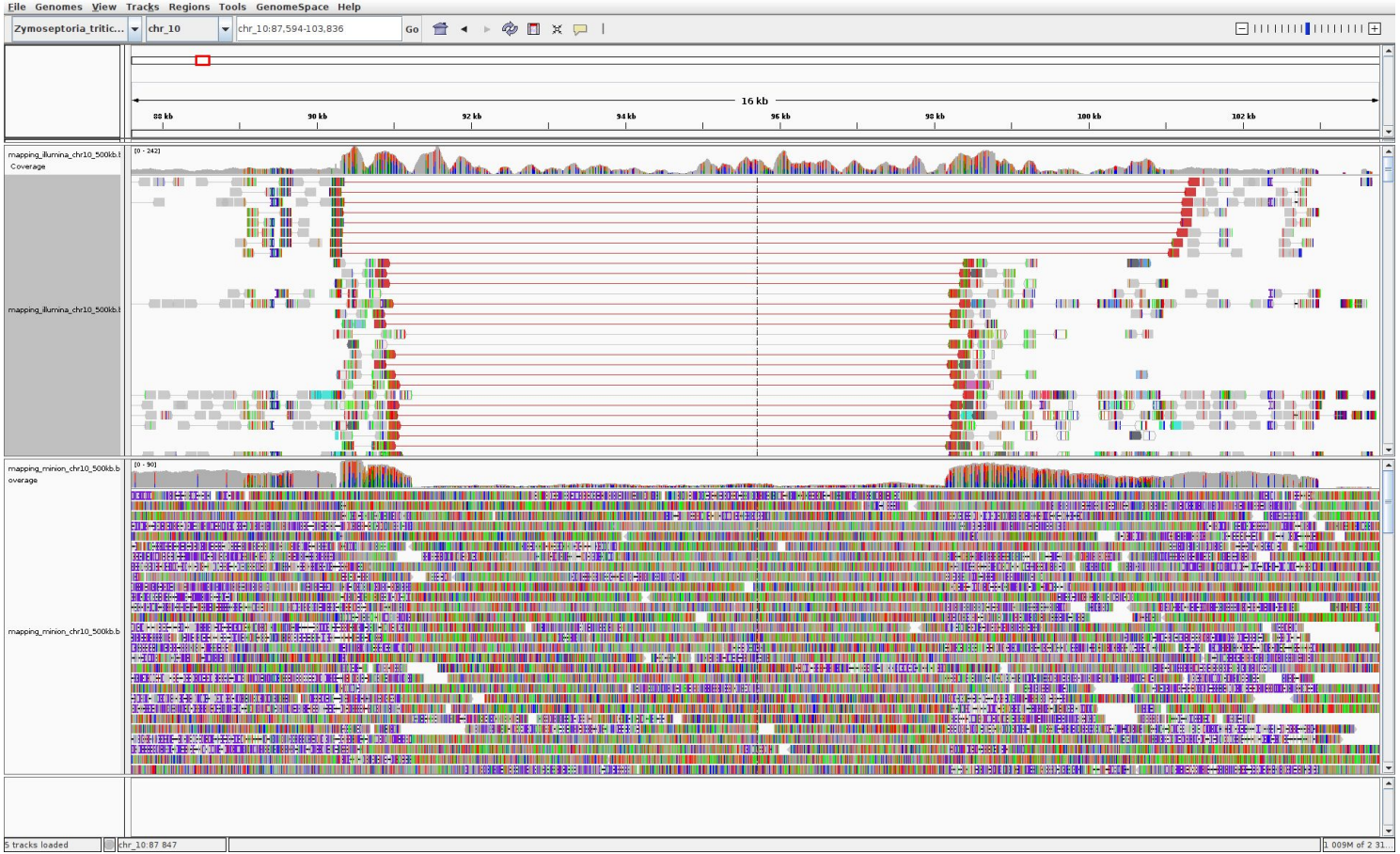
- Chargement du génome de référence

  → `~/tp_sv/Zymoseptoria_tritici.fa`

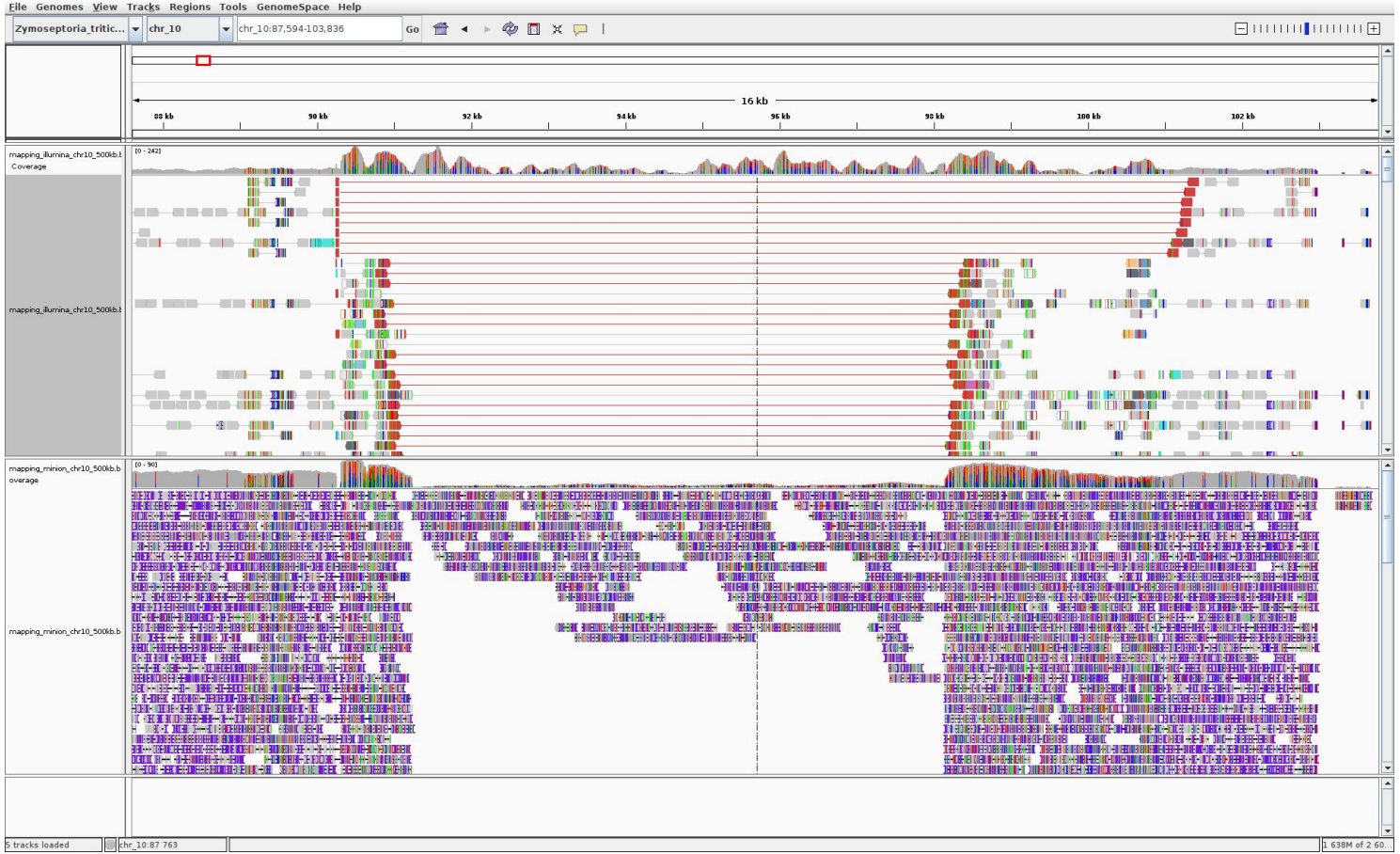
- Ouvrez à partir d'un fichier les fichiers BAM correspondant aux deux analyses (short et long reads) :

  → `~/tp_sv/mapping_illumina_chr10_500kb.bam`
  → `~/tp_sv/mapping_minion_chr10_500kb.bam`

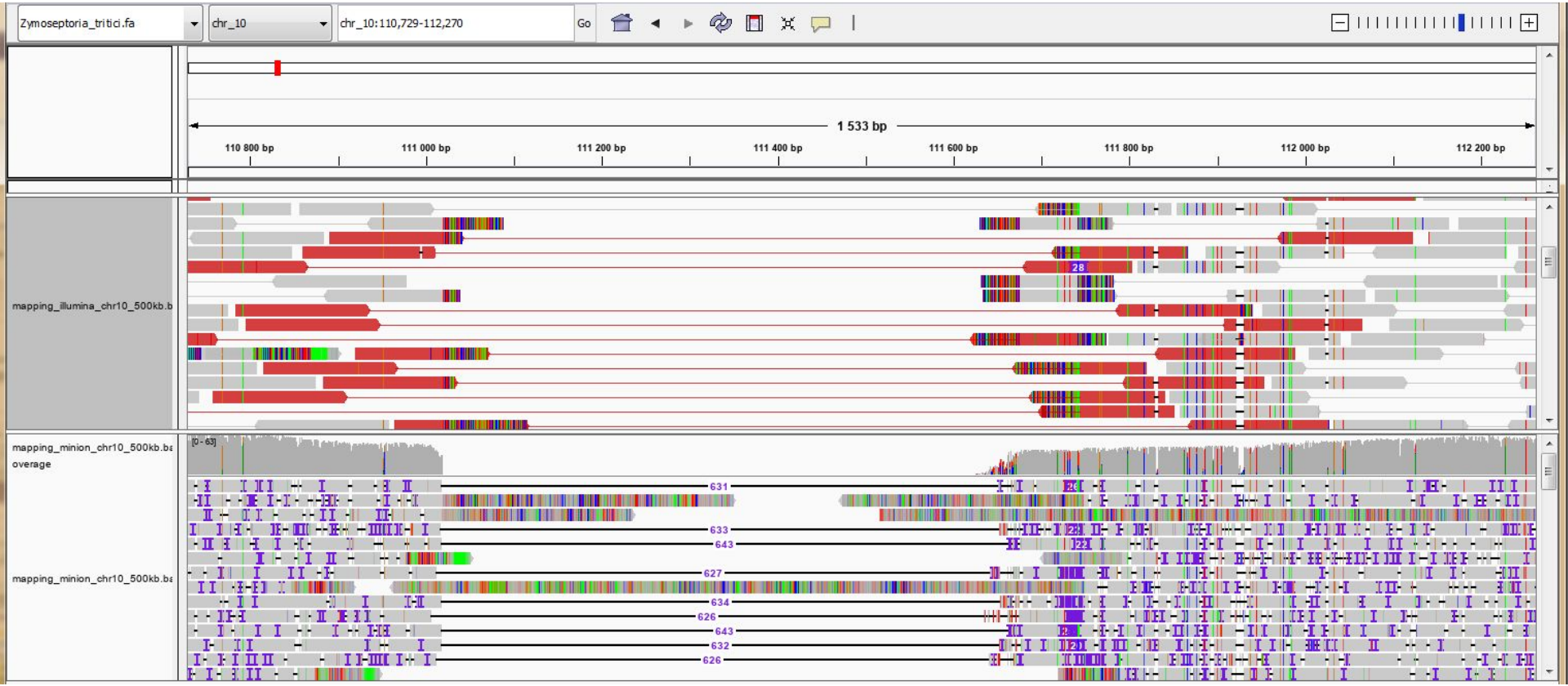
deletion 90309-101040 (illumina), 91233-98159 (Minion)



deletion 90309-101040 (illumina), 91233-98159 (Minion)

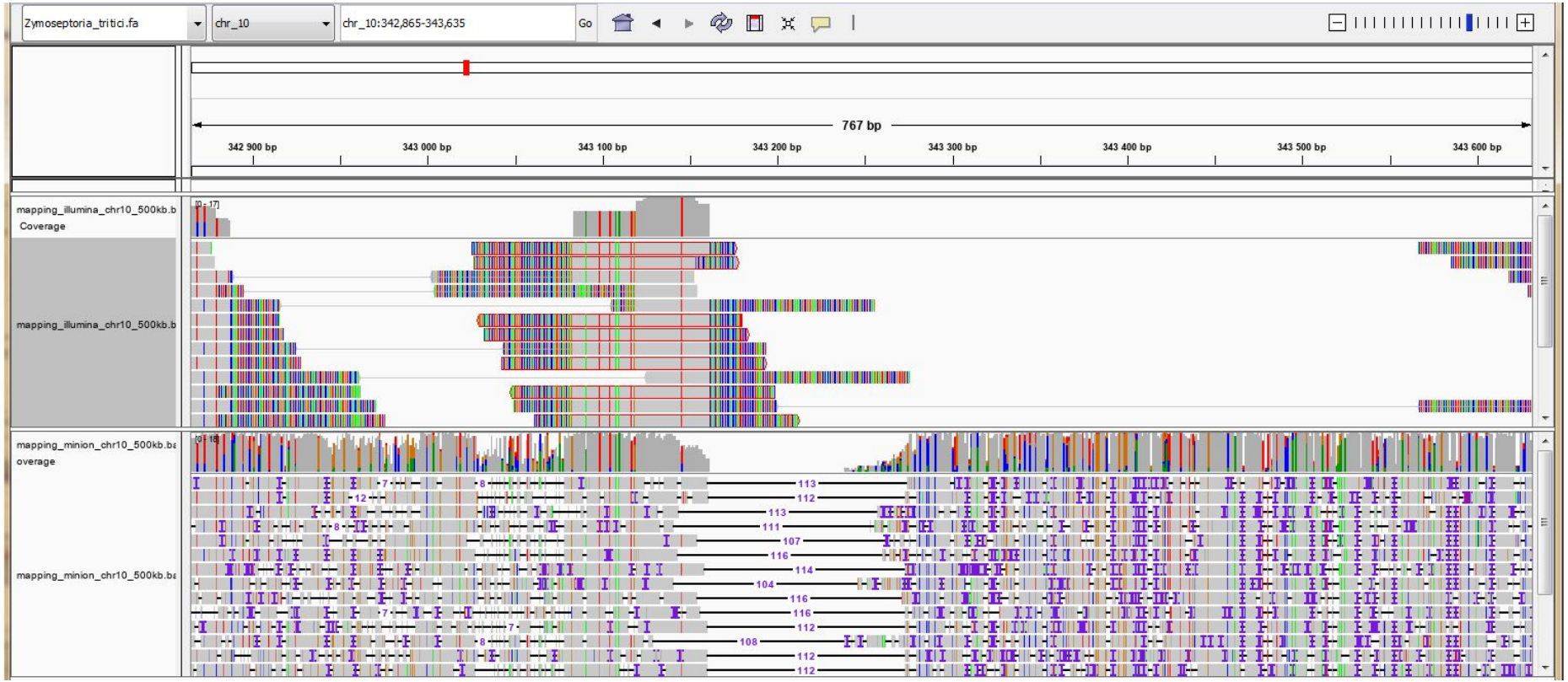


deletion 111021-111676



deletion 191291-191343

deletion 343161-343273



Comparaison des résultats de Delly et Sniffles

Delly (illumina)				
start	stop	precision	PE	SR
29522	29580	PRECISE	0	20
57127	57600	PRECISE	3	16
80015	80622	PRECISE	15	20
90255	90309	PRECISE	0	7
90309	101040	IMPRECISE	8	0
111021	111676	IMPRECISE	20	0
191291	191343	PRECISE	0	18
-	-	-	-	-
264986	265063	PRECISE	0	12
-	-	-	-	-
360628	361052	PRECISE	0	20
383682	477911	IMPRECISE	7	0
425686	426624	IMPRECISE	28	0
465858	466080	PRECISE	0	20
468192	468342	PRECISE	0	20
477523	479732	PRECISE	0	20
477526	479732	IMPRECISE	41	0

Sniffles (Minion)		
start	stop	precision
-	-	-
57126	57598	IMPRECISE
-	-	-
-	-	-
91233	98159	IMPRECISE
111020	111655	PRECISE
-	-	-
257001	257165	IMPRECISE
-	-	-
343161	343273	PRECISE
360638	361061	PRECISE
383681	477805	IMPRECISE
425682	426487	IMPRECISE
-	-	-
468192	468341	PRECISE
477525	479731	PRECISE
-	-	-

IGV OK

IGV ~OK

IGV doubt

IGV NO

Conclusion

- La détection des SVs manque de précision et engendre des faux positifs et faux négatifs

  → **Nécessité de croiser différents outils/technologies**

  → **Nécessité de bien utiliser les métriques des outils**

  → **Nécessité d'une bonne profondeur (variant hétérozygote)**

- Vérifier visuellement les résultats sur IGV permet d'augmenter la confiance dans les SVs détectés