1 Structural Variation Calling - Solutions

No questions in this section.

2 Looking at Structural Variants in VCF

2.1 Exercises

1. What does the CIPOS format tag indicate? **Confidence interval around POS for imprecise variants**

2. What does the PE tag indicate? **Number of paired-end reads supporting the variant across all samples**

3. What tag is used to describe an inversion event? **INV**

4. What tag is used to describe a duplication event? **DUP**

5. How many deletions were called in total? (**Hint:** DEL is the info field for a deletion. The -c option of the grep command can be used to return a count of matches.) **31, try**

```
grep -c DEL ERR1015121.vcf
```

6. What type of event is predicted at IV:437148? What is the length of the SV? How many paired-end reads and split-reads support this SV variant call? **Deletion -370 20 PE 21 split**

7. What is the total number of SV calls predicted on the IV chromosome? **10, try**

```
grep -c "IV" ERR1015121.vcf
```

3 Calling Structural Variants

Q: mean=454.87 std=86.29

3.1 Breakdancer

3.1.1 Exercises

```
grep "III:83065" ERR1015121.breakdancer.out
```

1. Inversion

2. -116,

3. 31

```
grep "II:258766" ERR1015121.breakdancer.out
```

4. Deletion (7325, 99)

5. grep DEL | awk OFS= breakdancer.dels.bed | awk '{print $1"\t"$2"\t"$5"\t"$7"\t"$9}' > breakdancer.dels.bed

3.2 Inspecting SVs with IGV

3.2.1 Exercises

1. Yes, a deletion (view as paired, sort by insert size, squish).

2. There are very few reads mapping of low mapQ and it has a SV score = 99

3. Size?

Was the deletion at II:258766 also called by the other structural variant software?

5. Yes, SVTYPE=DEL, SVLEN=-7438

6. DEL called by breakdancer (score=34). Not found by other caller.

7. Yes, 2 reads support (red).

3.3 Lumpy

3.3.1 Exercises

1. The `-F` option in `samtools view` excludes reads matching the specified flag

2. reads in proper pair | read unmapped | mate unmapped | not primary alignment | PCR optical duplicate

3. Duplication 471

4. Deletion -38

4 Calling Structural Variants from Long Reads

4.0.1 Align the reads with minimap and convert to bam

```
cd ref            minimap2 -d Saccharomyces_cerevisiae.R64-1-1.dna.toplevel.fa.mmi
Saccharomyces_cerevisiae.R64-1-1.dna.toplevel.fa              cd ../exercises4
minimap2 -t 2 -x map-pb -a ../ref/Saccharomyces_cerevisiae.R64-1-
1.dna.toplevel.fa.mmi YPS128.filtered_subreads.10x.fastq.gz | samtools view -b
-o YPS128.filtered_subreads.10x.fastq.bam -
```

4.0.2 Sort the bam

```
samtools sort -o YPS128.filtered_subreads.10x.fastq.sorted.bam -@ 4
YPS128.filtered_subreads.10x.fastq.bam
```

4.0.3 Index the sorted bam

```
samtools index YPS128.filtered_subreads.10x.fastq.sorted.calmd.bam
```

4.0.4 Call SVs with sniffles

```
sniffles -m YPS128.filtered_subreads.10x.fastq.sorted.calmd.bam -v
YPS128.filtered_subreads.10x.fastq.vcf
```

4.0.5   Exercises

1. What sort of SV was called at on chromosome 'Mito' at position 29295? __Duplication_

2. What is the length of the SV? **17511**

3. How many reads are supporting the SV? **14 (RE tag)**

4. From a visual inspection of the SV in IGV, can you determine how accurate is the breakpoint of the called SV compared to what you see in IGV?

5   Bedtools

5.1   Exercises

1. How many SVs found in `ERR1015069.dels.vcf` overlap with a gene? (**Hint:** Use bedtools intersect command) **18, try**

```
bedtools intersect -u -a ERR1015069.dels.vcf -b Saccharomyces_cerevisiae.R64-1-
1.82.genes.gff3  | wc -l
```

2. How many SVs found in `ERR1015069.dels.vcf` do not overlap with a gene? (**Hint:** note the -v parameter to bedtools intersect) **9, try**

```
bedtools intersect -v -a ERR1015069.dels.vcf -b Saccharomyces_cerevisiae.R64-1-
1.82.genes.gff3  | wc -l
```

3. How many SVs found in `ERR1015069.dels.vcf` overlap with a more strict definition of 50%? **14, try**

```
bedtools intersect -u -f 0.5 -a ERR1015069.dels.vcf -b Saccharomyces_cerevisiae.R64-
1-1.82.genes.gff3  | wc -l
```

4. What is the closest gene to the structural variant at IV:384220 in `ERR1015069.dels.vcf`? **YDL037C, try**

```
bedtools closest -d -a ERR1015069.dels.vcf -b Saccharomyces_cerevisiae.R64-1-
1.82.genes.gff3| grep IV | grep 384220
```

5. How many SVs overlap between the two files `ERR1015069.dels.vcf` and `ERR1015121.dels.vcf`? **27, try**

```
bedtools intersect -u -a ERR1015069.dels.vcf -b ERR1015121.dels.vcf | wc -l
```

6. How many SVs have a 90% reciprocal overlap between the two files `ERR1015069.dels.vcf` and `ERR1015121.dels.vcf` (**Hint:** first find the option for reciprocal overlap by typing: bedtools intersect -h) **24, try**

```
bedtools intersect -u -r -f 0.9 -a ERR1015069.dels.vcf -b ERR1015121.dels.vcf |
wc -l
```