

Module 3 Quiz

8/10 points (80%)

Quiz, 10 questions

✓ Congratulations! You passed![Next Item](#)1 / 1
points

1.

Which of the following statements is FALSE:



SNV refers to a Single Nucleotide Variant.



SNP refers to a Single Non-defined Polymorphism

**Correct**

Structural variants include block deletions and insertions, among others.



SNVs encompass single nucleotide insertions, deletions and substitutions.

1 / 1
points

2.

Which of the following statements is FALSE:



The mpileup format has either 6 or 7 columns.



The VCF format shows the changes in amino acid resulting from the nucleotide mutation, in column 3.

**Correct**

The BAM format is a binary compressed representation for alignments of next generation sequencing reads.



The genotype fields in VCF provide information about the variant in each sample.

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points

3.

What program can be used to generate a list of candidate sites of variation in an exome data set:



mkdir



samtools

This should not be selected



bedtools



bcftools



0 / 1
points

4.

In a comprehensive effort to study genome variation in a patient cohort, you sequence and call variants in the exome, whole genome shotgun and RNA-seq data from each patient. Which of the following is FALSE when comparing these three types of resources:



Exome sequencing comprehensively captures variants in the 3' and 5' UTRs of genes.



Exome sequencing can capture variants in a pre-defined set of coding exons and their immediate surrounding area.



Exome sequencing cannot determine variants in novel polymorphic alternative splicing events.

This should not be selected



Exome sequencing captures fewer variants than whole genome sequencing.

4. Whole genome sequencing can potentially detect all variants that can be found with either exome sequencing or RNA-seq.

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5.

Which of the following options can be used to allow bowtie2 to generate partial alignments?



--local

**Correct**

-ignore-quals



-D



--sensitive

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points

6.

Select the correct interpretation for the snippet of 'mpileup' output below.

```
1  ``
2  Chr3 11700316 C 8 $. . . . . 8C@C;CB3
3  Chr3 11951491 G 16 AAAA, . . . . . aA..A C2@2BCBCCCAC2CC4
4  ``
```



Both sites show potential variation

the alternate letter for site 1 is \$, and for site 2 is G;

site 1 has 8 supporting reads, and site 2 has 16



Only site 2 shows potential variation;

the alternate letter for site 2 is A;

site 1 has 8 supporting reads, and site 2 has 16

**Correct**

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Both sites show potential variation;

the alternate letter for site 1 is \$, and for site 2 is A;

site 1 has 8 supporting reads, and site 2 has 16



Both sites show potential variation;

the alternate letter for site 1 is C, and for site 2 is G;

site 1 has 8 supporting reads, and site 2 has 7



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7.

Given the set of variants described in the VCF excerpt below, which of the following is FALSE?

```
1  ``
2  ##INFO=<ID=DP,Number=1,Type=Integer,Description="Raw read depth">
3  ##INFO=<ID=MQ,Number=1,Type=Integer,Description="Average mapping quality">
4  ##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
5  ##FORMAT=<ID=PL,Number=G,Type=Integer,Description="List of Phred-scaled genotype
    likelihoods">
6  Chr3 11966312 . G A 15.9 . DP=5;MQ=15 GT:PL 1/1:43,9,0
7  Chr3 11972108 . TAAAA TAAA 32.8 . INDEL;IDV=7;IMF=0.636364;DP=11;MQ=22 GT
    :PL 0/1:66,0,2
8  Chr3 13792328 rs145271872 G T 5.5 . DP=1;MQ=40 GT:PL 0/1:32,3,0
9  ``
```



The sample contains only the alternate allele for variant 3



Correct



The sample contains both alleles for variant 2



Average mapping quality for variant 3 is 40



The sample contains only the alternate allele for variant 1



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points

8.

What does the following code do:

```
1  ``
2  bowtie2 -x species/species -U in.fastq | grep -v "^@" | cut -f3 | sort | uniq -c
3  ``
```

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Run bowtie2 with a set of single-end reads, allowing for local matches;

then determine the number of exact matches on each genomic sequence



Run bowtie2 with a set of single-end reads, reporting the best alignment only;

then determine the number of matches on each genomic sequence



Correct



Run bowtie2 with a set of single-end reads, reporting the top 5 alignments for a read;

then determine the number of reads mapped reverse complemented



Run bowtie2 with a set of single-end reads, allowing for local matches;

then determine the number of matches with unmapped mates



1 / 1
points

9.

What does the following snippet of code do NOT do:

```
1  ```\n2  samtools mpileup -O -f genome.fa in.bam | cut -f7\n3  ```\n
```



Report in the intermediate mpileup output the qualities of all read bases aligned at that position



Produce a 7-column intermediate mpileup file that is piped to 'cut'



Require a sorted BAM file



Report an empty column



Correct

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What does the following code do NOT do:

```
1  '''  
2  bcftools call -v -c -O z -o out.vcf.gz in.vcf.gz  
3  '''
```



Skip indels

Correct



Use the 'consensus caller' model to call variants



Report output in compressed VCF format



Report variant sites only

