

Module 3 Quiz

9/10 points (90%)

Quiz, 10 questions

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

1.

Which of the following statements is FALSE:



Different versions of a gene resulted from genomic mutations are called alleles.



SNP refers to a Single Non-defined Polymorphism

**Correct**

SNV refers to a Single Nucleotide Variant.



Differences in the genomes of individuals are strong contributors to their phenotypic variations.

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points

2.

Which of the following statements is FALSE:



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

**Correct**

VCF stands for Variant Call Format.



The BCF format is a binary compressed version of VCF.



The VCF INFO lines describe characteristics of the variant, included in column 8.

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3.

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



samtools mpileup

**Correct**

samtools depth



samtools view



bowtie2

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:



Whole genome sequencing can comprehensively identify variants in all protein-coding genes.



RNA-seq will only capture variants in the expressed genes.

**This should not be selected**

RNA-seq can systematically identify variants in gene regulatory regions.



RNA-seq allows detection of intronic variants.



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

`--local`**Correct**`-l``--very-fast``-ignore-quals`

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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   ...
```



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**

Only site 1 shows potential variation;

the alternate letter for site 1 is \$;

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



Only site 1 shows potential variation;

the alternate letter for site 1 is C;

site 1 has 9 supporting reads, and site 2 has 16



Both sites show potential variation;

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the alternate letter for site 1 is '.', and for site 2 is A;

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site 1 has 9 supporting reads, and site 2 has 16



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points

7.

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

```
1  ```\n2  ##INFO=<ID=DP,Number=1,Type=Integer,Description="Raw read depth">\n3  ##INFO=<ID=MQ,Number=1,Type=Integer,Description="Average mapping quality">\n4  ##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">\n5  ##FORMAT=<ID=PL,Number=G,Type=Integer,Description="List of Phred-scaled genotype\n    likelihoods">\n6  Chr3  11966312  .  G A 15.9  . DP=5;MQ=15  GT:PL  1/1:43,9,0\n7  Chr3  11972108  .  TAAAA TAAA 32.8  .  INDEL;IDV=7;IMF=0.636364;DP=11;MQ=22  GT\n    :PL 0/1:66,0,2\n8  Chr3  13792328  rs145271872 G T 5.5  . DP=1;MQ=40  GT:PL 0/1:32,3,0\n9  ```\n
```



The sample contains only the alternate allele for variant 3

**Correct**

The alternate allele for variant 1 is A



The alternate allele for variant 3 is T



The quality values for the three calls are 15.9, 32.8 and 5.5



1 / 1

points

8.

What does the following code do:

```
1  ```\n2  bowtie2 -x species/species -U in.fastq | grep -v "^@" | cut -f3 | sort | uniq -c\n3  ```\n
```



Run bowtie2 with a set of single-end reads, reporting up to 5 alignments per read; then determine the number of matches on each genomic sequence



Run bowtie2 with a set of paired-end reads, allowing up to 10 matches per read;

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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 paired-end reads, allowing for local matches;

then report the numbers of alignments containing insertions and deletions, respectively;



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9.

What does the following snippet of code do NOT do:

```
1  ```
2  samtools mpileup -O -f genome.fa in.bam | cut -f7
3  ```
```



Generate intermediate output in uncompressed BCF format

**Correct**

Generate intermediate output in mpileup format



Take in the input BAM file in.bam



Require a sorted BAM file



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10.

What does the following code do NOT do:

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

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Report output in compressed VCF format



Call variants in a single sample



Skip indels



Correct



Report variant sites only

