

Module 3 Quiz

9/10 points (90%)

Quiz, 10 questions

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points

1.

Which of the following statements is FALSE:

- ☐ SNVs encompass single nucleotide insertions, deletions and substitutions.
- ☒ SNP refers to a Single Non-defined Polymorphism

**Correct**

- ☐ SNV refers to a Single Nucleotide Variant.
- ☐ Structural variants include block deletions and insertions, among others.

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points

2.

Which of the following statements is FALSE:

- ☐ 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.
- ☐ 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

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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

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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:



RNA editing can confound the detection of variants from RNA-seq data.



RNA-seq allows detection of intronic variants.



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



All of the three methods can identify variants located in the introns, albeit to different degrees.

**This should not be selected**

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5.

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



--local

**Correct**

-ignore-quals



--sensitive



-D

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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 2 shows potential variation;

the alternate letter for site 2 is A;

the alternate allele for site 2 is supported by 9 reads



Only site 2 shows potential variation;

the alternate letter for site 2 is G;

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

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Only site 2 shows potential variation;

the alternate letter for site 2 is '.';

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

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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 both alleles for variant 2
- ☒ The sample contains only the alternate allele for variant 3

Correct

- ☐ The sample contains only the alternate allele for variant 1
- ☐ Average mapping quality for variant 3 is 40



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8.

What does the following code do:

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

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

then determine the number of matches on each genomic sequence

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Correct

- ☐ Run bowtie2 with a set of single-end reads, reporting up to 5 alignments per read;
- then determine the number of matches with unmapped mates
- ☐ Run bowtie2 with a set of single-end reads, reporting the top 5 alignments for a read;
- then list the number of matches containing insertions and deletions, respectively
- ☐ Run bowtie2 with a set of single-end reads, allowing for local matches;
- then determine the number of matches with unmapped mates
-



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points

9.

What does the following snippet of code do NOT do:

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

- ☒ Report an empty column

**Correct**

- ☐ Require a sorted BAM file
- ☐ Produce a 7-column intermediate mpileup file that is piped to 'cut'
- ☐ Report in the intermediate mpileup output the qualities of all read bases aligned at that position
-



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10.

What does the following code do NOT do:

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

- ☐ Call variants in a single sample
- ☐ Report variant sites only
- ☐ Report output in compressed VCF format
- ☒ Skip indels

Correct