

## NGS\_21\_QC\_Variant\_Calling

## Quiz template

Add questions, at least two answer alternatives and choose correct answers (at least one). Have fun creating your awesome quiz!  
See an example question below (don't forget to overwrite this with your first question!)

Question	Answer 1	Answer 2	Answer 3	Answer 4	Correct answer(s) - choose at least one
Which of the following is NOT a type of variant that can be identified by a variant calling algorithm	SNPs/SNVs The probability of the base being an error is 1	Indels The probability of the base being an error is 1 in 10	MNPs <b>The probability of the base being an error is 1 in 1000</b>	CpGs The probability of the base being an error is 1 in 10000	4
A base call with a Q score of 30 indicates					3
A pileup file contains as many lines in the file as there are:	Reads in the FASTQ file	Reads in the alignment file	<b>Bases that align that align</b>	Bases in the FASTQ file	3
Refer back to your out.vcf file created during your assignment session (exercise 2.3.1): Locate variant at position 10059938. The ref/alt alleles, allele depth, depth of coverage and mapping quality is:	G/A; 57,0; 57; 59	<b>G/A; 0,94; 99; 60</b>	C/T; 0,94; 99; 45	A/G; 94,0;94; 36	2
Refer back to your out.vcf file created during your assignment session (exercise 2.3.1): Locate variant at position 10059938. The genotype of this variant in this sample is:	<b>A/A</b>	0/0	G/A	A/G	1
Refer back to your out.vcf file created during your assignment session (exercise 2.3.1): What type of variant is found at position 10975593	SNP/SNV	Mutation	CNV	<b>INDEL</b>	4
Refer back to your out.vcf file created during your assignment session (exercise 2.3.1): How many SNPs have a quality score of greater than or equal to 30, together with an alternative allele depth of greater than or equal to 70		108	<b>128</b>	107	111 2
Refer back to your out.vcf file created during your assignment session (exercise 2.3.1): Use any command to determine the number of indels in the file with a quality score of greater than or equal to 50		<b>9</b>	575	612	527 1
Filtering of variants called from NGS data is important because:	variants called may be false	variants called may be sequenced	<b>all of the options</b>	there may be mapping errors	3
How big must a genetic variant be to qualify as a Structural Variant?	50000 basepairs	<b>50 basepairs</b>	25 basepairs	1000 basepairs	2
What is the name of a transposon (or "Jumping Gene") present in human genomes?	TP53	Gypsy	<b>Alu</b>	Junk	3
What technique(s) would be useful for identifying a translocation?	Chromosome banding	Short Read WGS	Long Read WGS	<b>all of the options</b>	4
In the ERR1015121.breakdancer.out file created in Exercise 3.1, what type of structural variation is present at position II:202502? (Hint: use grep)	Insertion	Duplication	<b>Deletion</b>	Inversion	3
What is the size of the structural variation present at position II:202502 in the ERR1015121.breakdancer.out file created in Exercise 3.1?	203040		50	2	<b>440</b> 4
What is the score of the structural variation present at position II:202502 in the ERR1015121.breakdancer.out file created in Exercise 3.1?	203040		<b>50</b>	2	440 2
In the ERR1015069.vcf file created by lumpy in Exercise 3.3.1, what type of structural variation is present at position IV:383993 ?	<b>Duplication</b>	Deletion	Insertion	Inversion	1
What is the size of the structural variation present at position IV:383993 in the ERR1015069.vcf file created in Exercise 3.3.1?	38399		84464	326	<b>471</b> 4
Using IGV and the files created in exercise 4.4, go to the region the CHR:IV Position:127636 - 128998 and click on the feature there to bring up the feature information box. What specific type of structural variation type is defined?	Duplication	<b>Deletion</b>	Insertion	Inversion	2
How many reads support the feature observed at CHR:IV Position:127636 - 128998?		10	12	<b>15</b>	19 3