**Data Basics & Alignment workshop: question sheet**

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| **Q1** | Use this website (<http://grand-prismatic.blogspot.co.uk/2013/02/fastq-quality-score-convesion-table.html>) to determine the error probability of the first base pair of read 1. | | | | | |
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| **Q2** | How many reads do the files contain? | | | | | |
| No. reads in WES01\_chr22\_R1.fastq | | | No. reads in WES01\_chr22\_R2.fastq | | |
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| **Q3** | How long are the reads (bp)? | | | | | |
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| **Q4** | Has either file failed any of the sequence quality checks? | | | | | |
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| **Q5** | What number and percentage of reads were removed (>10% bases with phred <20)? | | | | | |
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| **Q6** | Use the Flagstat output to determine the percentage of mapped reads | | | | | |
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| **Q7** | Use the Flagstat outputs to calculate the number of reads that were filtered out (difference in total read count between the raw and filtered BAM files). | | | | | |
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| **Q8** | Use this website (http://broadinstitute.github.io/picard/explain-flags.html) to decode the flag (147) of the read in table 1 and determine which strand of the reference genome it is aligned with. | | | | | |
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| **Q9** | Use the IdxStats output to calculate the percentage of reads mapping to chromosome 22. | | | | | |
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| **Q10** | View the tabular output and record: | | | | | |
| Mean insert size (bp) | | | Standard deviation | | |
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| **Q11** | What is the mean coverage and percentage of target bases covered by 15 or more reads? | | | | | |
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| **Q12** | What does the coverage/depth look like in exons and introns and is this expected? | | | | | |
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| **Q13** | Mouse over the coverage track for the SNV at 24,167,513bp and record: | | | | | |
| alleles | No. ref reads | No. alt. reads | Gene | Amino acid | rsID |
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| **Q14** | Is the variant at 24,167,513bp likely to contribute to patients symptoms? | | | | | |
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