

Module Planning template

NGS Bioinformatics - Remote classrooms

Module Title: Alignment to Reference

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→ **Summary or objectives** – what you plan to cover for this module

→ **List of learning outcomes specific for this module**

(Please list 3 - 5 Learning Outcomes here; refer to How to writing learning outcomes)

- *Understanding the need for a reference sequence*
- *Understanding the concept of alignment (local alignment, seed based alignment, suffix tries)*
- *Understanding alignment limitations and mapping qualities*
- *Understanding the costs involved in doing large scale alignments*
- *Being aware of additional steps involved in doing alignment improvement*

→ **Total number of hours:** 3 hours

→ **Tools/software and resources**

◆ Software

- bwa
- samtools
- picard
- igv

◆ Description of datasets to be used (if known)

- We will align genomic sequence (from Whole-Genome Sequencing) from a mouse embryo which has been mutagenised while the one-cell stage is manipulated using CRISPR-Cas9 and a gRNA targeting an exon of the Tyr gene. The successful mutation of the gene will delete one or both alleles. A bi-allelic null Tyr mouse will be albino, but otherwise healthy.

◆ Other resources, or readings

→ **Overview of activities and exercises**

(list specific practical activities which participants will do)

Introduction lecture:

Exercise/Activity 1: Align mouse sequences against a reference and run mark duplicates on that. Gather some statistics.

Exercise 2: Go through features in IGV and inspect variants in your data.

Exercise 3: Align two lanes of sequence data, merge and mark duplicates. Inspect in IGV.

[Appendix](#) (include any guideline documents related to content development such as breaking down content for remote classroom format, how to write LO's, designing assignments etc...)

[See links to other guidelines](#)

-Overall trainer guideline and template documents will include information on lecture formats and recordings, contact sessions schedules, checklists, assignments and assessments, quizzes, feedback, audio-video conferencing, learning management platform, teaching assistants
