

Canadian Bioinformatics Workshops

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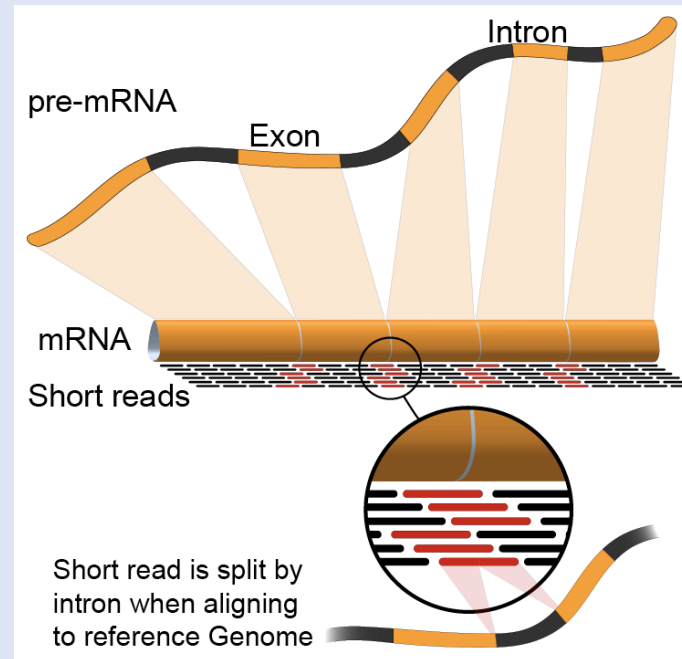
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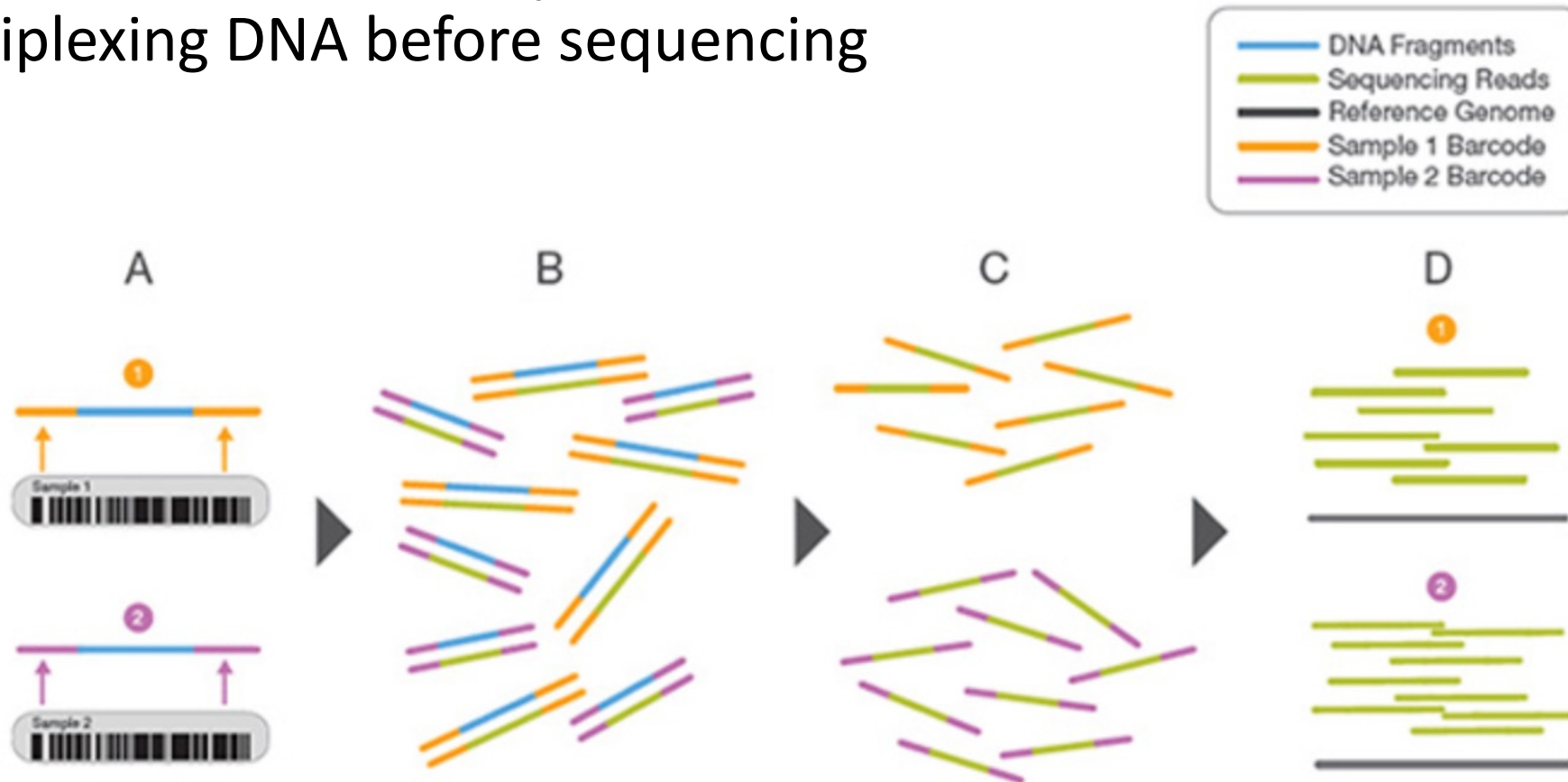
RNA-Seq Module 1: Indexing

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“Index” has many different meanings

- Indexes can refer to unique barcodes used for multiplexing DNA before sequencing



<https://www.illumina.com/science/technology/next-generation-sequencing/multiplex-sequencing.html>

Indexing in bioinformatics/CS enables rapid access

- Indexing is a recurring theme in genome analysis
- Files are *big* - scanning through them can take a long time
- Indexing builds a table-of-contents so that we can jump directly to specific positions
- Indexing may require significant compute/time but typically only occurs once
- Each application may require a different indexing strategy

What's inside a fasta's index file? (.fai)

| contig name | bases in contig | byte index of the file where the contig begins | bases per line | | bytes per line |
|-------------|-----------------|--|----------------|----|----------------|
| | | | | | |
| chr1 | 248956422 | 6 | 60 | 61 | |
| chr2 | 242193529 | 253105708 | 60 | 61 | |
| chr3 | 198295559 | 499335802 | 60 | 61 | |
| chr4 | 190214555 | 700936293 | 60 | 61 | |
| chr5 | 181538259 | 894321097 | 60 | 61 | |
| chr6 | 170805979 | 1078885000 | 60 | 61 | |
| chr7 | 159345973 | 1252537752 | 60 | 61 | |
| chr8 | 145138636 | 1414539498 | 60 | 61 | |
| chr9 | 138394717 | 1562097118 | 60 | 61 | |
| chr10 | 133797422 | 1702798421 | 60 | 61 | |

Example index applications and associated files

| Source file | Indexed file | Indexing tool | Use case |
|-------------|--------------|----------------|--|
| .bam | .bai | samtools index | Visualize bam in IGV |
| .fasta | .fai | faidx | Extract specific sequences from ref genome |
| .vcf | vcf.gz.tbi | bgzip/tabix | Pull out specific variants |
| .bed | .bed.gz.tbi | bgzip/tabix | extract specific genomic regions |

Indexing is also essential for alignment

- Finding out where to place a read in the genome is impractical unless matches can be quickly found
- All read aligners use some kind of indexing
- These indices must be “built” once for a reference genome, but can then be used every time the aligner is run
- Different aligners use different indexing schemes that are not compatible

We are on a Coffee Break & Networking Session

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