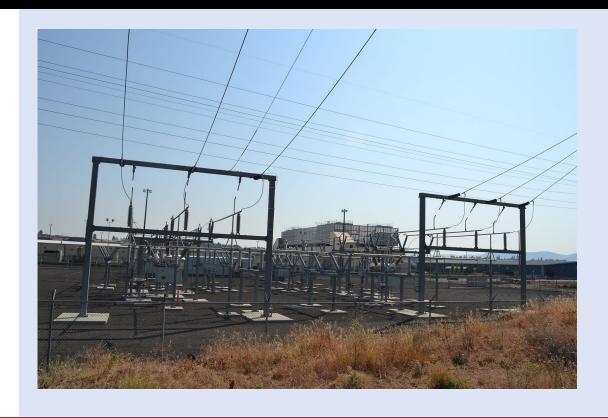


Cold Spring Harbor Laboratory

RNA-Seq Module 1 Indexing

Kelsy Cotto, Felicia Gomez, Obi Griffith, Malachi Griffith, Huiming Xia Advanced Sequencing Technologies & Applications November 5- 16, 2019

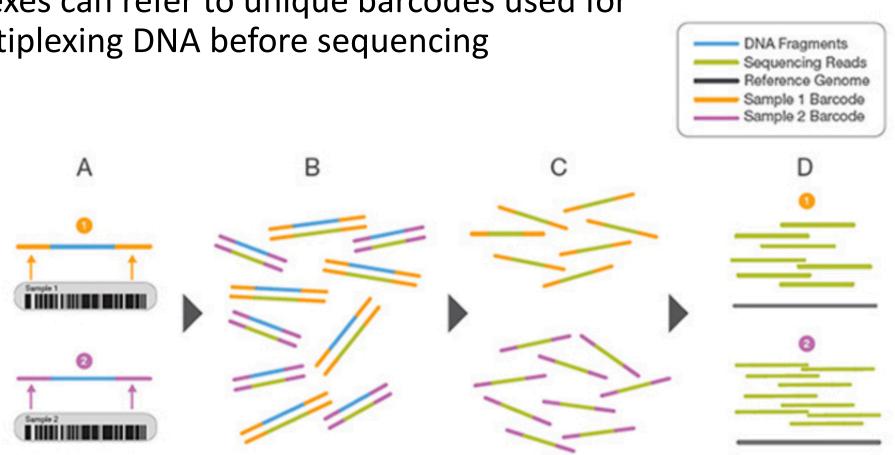




Module 1 rnabio.org

"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 ir	n contig	file	e index of the where the ntig begins		bases	s per line	bytes per	· line
C	chr1	248956422	2	6		60	61		
C	chr2	242193529)	253105708		60	61		
C	chr3	198295559)	499335802		60	61		
C	chr4	190214555	5	700936293		60	61		
C	chr5	181538259)	894321097		60	61		
C	chr6	170805979)	1078885000)	60	61		
C	chr7	159345973	3	1252537752	2	60	61		
C	chr8	145138636	5	1414539498	3	60	61		
C	chr9	138394717	7	1562097118	3	60	61		
C	chr10	133797422	2	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