

Read Alignment

Alignment Process

Reference:

ATCGATCGATCGTAGCTAGCTAGCTAGCTAGCTAGCTAGCTAGCTAGCTAGCTAGCT...

Read 1: ATCGATCGATCG

✓ Perfect match

Read 2:

TAGCTAGCTAGC

! Mismatch allowed

Read 3:

TAGCTAGCTA

Gap/Indel

- Map sequencing reads to reference genome
- Find best matching position for each read
- Allow for mismatches and gaps (indels)
- Handle multi-mapping and unique reads

BWA

DNA-seq

Burrows-Wheeler Aligner

Bowtie2

DNA-seq

Fast, gapped alignment

STAR

RNA-seq

Splice-aware aligner

Key Considerations

- Read length
- Error rate
- Computational resources
- Paired-end vs single-end

Quality Metrics

- Mapping rate (>80% good)
- Properly paired (%)
- Coverage uniformity
- Duplicate rate