

Read Alignment

Alignment Process

Reference:

Read 1: ATCGATCGATCG

✓ Perfect match

Read 2:

TAGCTAGCAAGC

! Mismatch allowed

Read 3:

TAGCT⁺-AGCTA

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