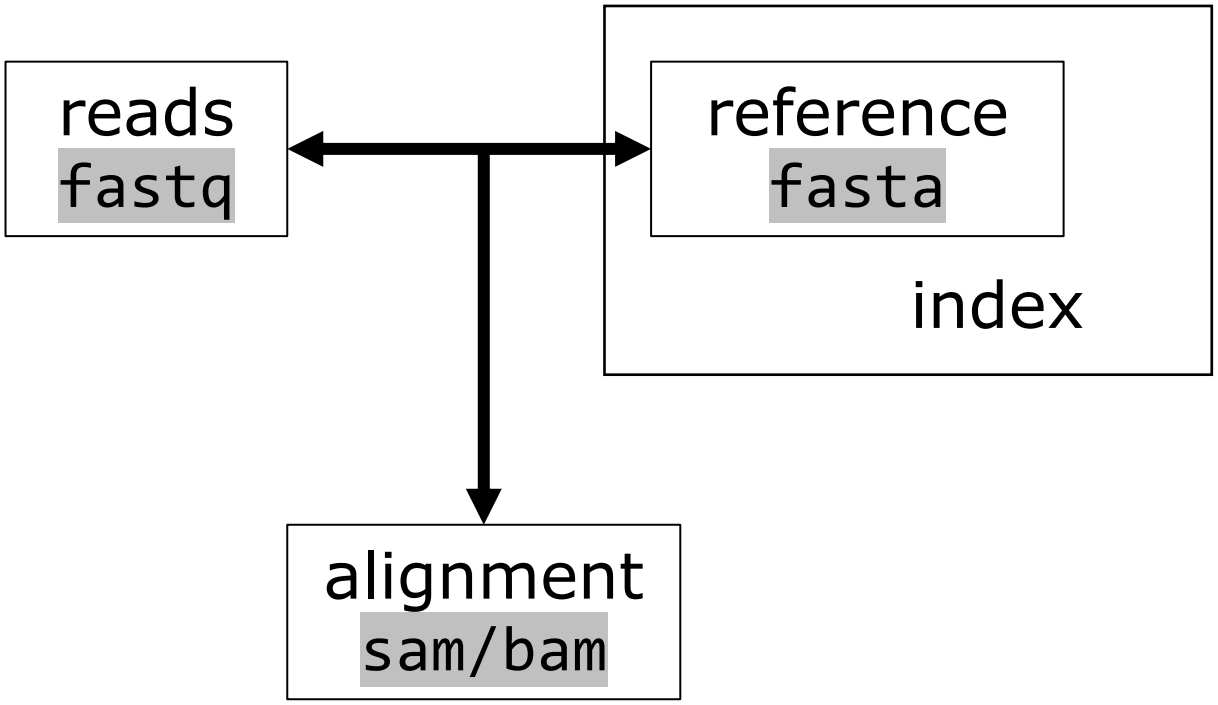
NGS - quality control, alignment, visualisation

Read alignment

Read alignment

- Find location of reads on reference



Software

- Basic alignment:
  - `bowtie2`
  - `bwa-mem`
- Splice-aware (RNA-seq):
  - `hisat2`
  - `STAR`
- Long reads + short reads + splice-aware:
  - `minimap2`

Mapping quality

$$MAPQ$$
$$= -10log_{10}\Pr\{mapping\ position\ is\ wrong\}$$

$$-10log_{10}(0.01) = 20$$
$$-10log_{10}(0.5) = 3$$