

Variant Calling Workflow Guide

A practical reference for moving from FASTQ reads to a filtered and annotated variant set.

Workflow

Step	Purpose	Typical outputs
1. Raw-read QC	Check FASTQ quality and sequencing artifacts.	QC reports
2. Read preprocessing	Trim or clean reads only when justified.	Clean FASTQ
3. Reference alignment	Map reads to the appropriate reference genome.	SAM/BAM
4. BAM processing	Sort, index and inspect alignments.	Sorted BAM + BAI
5. Variant calling	Detect candidate SNVs and indels.	Raw VCF
6. Variant filtering	Apply quality filters or calibrated thresholds.	Filtered VCF
7. Annotation	Add gene, consequence and population information.	Annotated VCF/table
8. Interpretation	Evaluate evidence in biological or clinical context.	Prioritized variants

Do not skip validation: Variant quality, coverage, caller choice, reference build and sample relationships can change conclusions. For clinical work, follow validated laboratory and interpretation standards.