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Whole Genome Sequencing Workflow Guide

A high-level map from DNA sequencing through coverage assessment, variant discovery and interpretation.

Workflow

Step	Purpose	Typical outputs
1. DNA and library QC	Confirm sample and library quality.	QC metrics
2. Sequencing	Generate genome-wide reads.	FASTQ
3. Read QC	Assess quality and contamination.	QC reports
4. Alignment	Map reads to a reference genome.	BAM/CRAM
5. Alignment QC	Evaluate mapping, depth and coverage.	Coverage metrics
6. Variant discovery	Call SNVs, indels and other variant classes as needed.	VCF and other callsets
7. Annotation	Add functional and population annotations.	Annotated variants
8. Interpretation	Connect genomic findings to the biological question.	Report / prioritized findings

WGS is broad: SNVs/indels, structural variants, copy-number variants, mitochondrial variants and repeat expansions may require different tools and validation strategies.