VARIANT DISCOVERY

Germline & somatic analysis at scale

An end-to-end variant-calling platform built on the MPEG-G stack — from raw reads to annotated, clinically ready variant reports.

WHAT IT IS

Variant Discovery orchestrates alignment, deduplication, recalibration and calling (DeepVariant / FreeBayes) on a reproducible Snakemake backbone, then annotates and prioritises findings.

WHY IT MATTERS

A single, auditable pipeline replaces brittle hand-wired scripts. MPEG-G storage keeps intermediate and final artefacts compact and addressable across a lab's whole cohort.

KEY CAPABILITIES

- Germline & somatic workflows
- DeepVariant and FreeBayes callers
- WGS / WES / targeted panels
- ANNOVAR-based annotation & filtering
- Reproducible Snakemake execution
- Interactive IGV-based review

Inputs	**FASTQ · BAM · CRAM · MPEG-G**
Outputs	**VCF · annotated reports**
Callers	**DeepVariant · FreeBayes**
Scaling	**Workstation to cluster**