Pipeline modules - WGS

Phase 1 – Testing ACMGuru package

- Prepare analysis-ready datasets
- Qualifying variant selection
- Tailored filtering
- Result interpretation





✓ ✓ × ✓ (manual)

Phase 2 – Testing pipeline automation

- GATK pipeline optimise
- Storage and costs
- ACMGuru reports





(template done)



Phase 3 – Automated run

Goals

WGS turnaround

- Single sample turn-around time
- Joint cohort analysis time / cost

Varian association tests

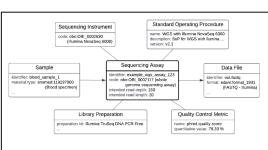
Genetic association with outcome including multi-omic input

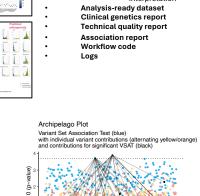
SPHN RDF Schema

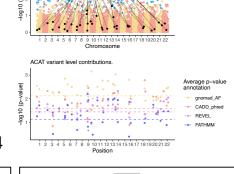
Output directly

Formal test

genomebottle pangenome true positive critical databases missing information table positive Max this analysis performed adequately? <u>See here</u> More the evidence sources used up-to-sters and reliable? <u>See here</u> New steps <u>See here</u>





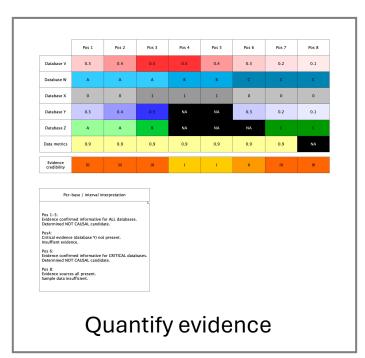


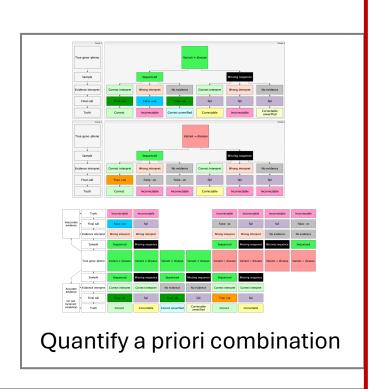


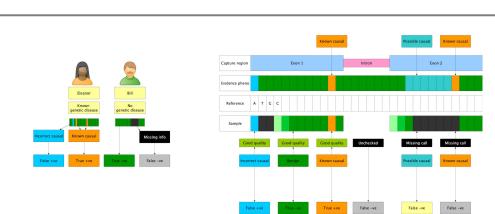
https://doi.org/10.20944/preprints202312.0373.v1

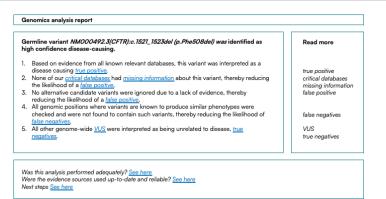
Future











Bayesian confidence diagnostic rate