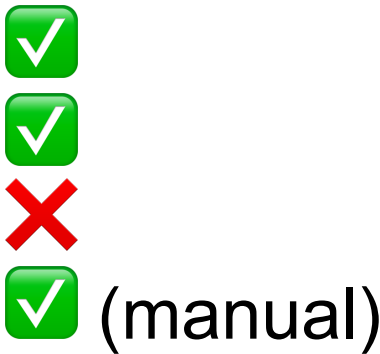


Pipeline modules - WGS

Phase 1 – Testing ACMGuru package

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



Phase 2 – Testing pipeline automation

- GATK pipeline optimise
- Storage and costs
- ACMGuru reports



Phase 3 – Automated run



Goals

WGS turnaround

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

Varian association tests

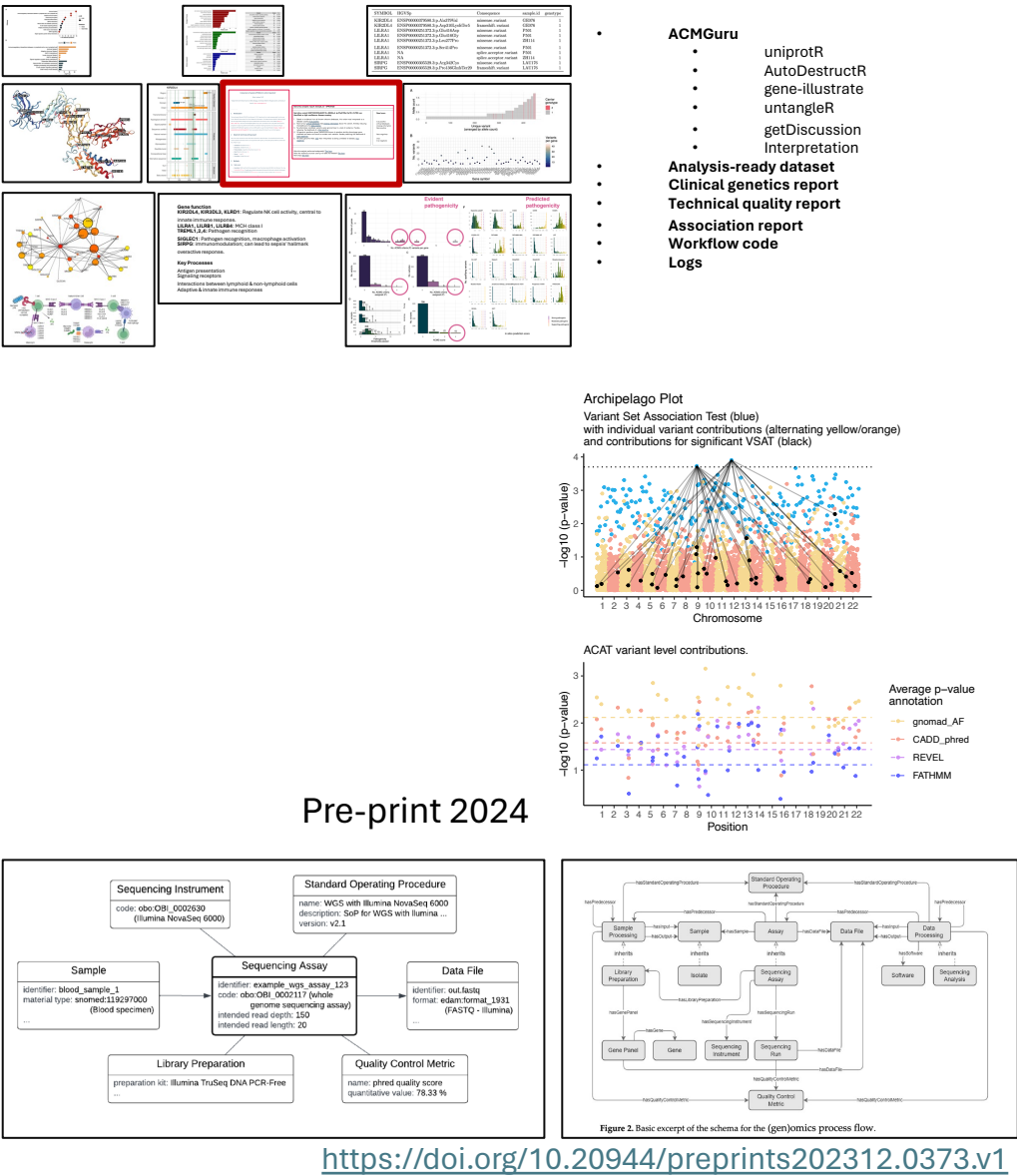
- Genetic association with outcome including multi-omic input

SPHN RDF Schema

- Output directly

Formal test

- genomebottle pangenome



Future

