Traditional

Selective
Some bias

Patient

WGS

Evidence priors → Tabular 1..n variants → 0 Clinical grade Clinical genetics report / Research grade ...cal ...etics ...ort

Improved

Automated input / output
No bias

Evidence priors

Guru

Tabular 1..n variants

Mechanism evidence viz, stats, etc.

1 Clinical grade Clinical genetics report

2 Research New disease mechanism

Cohort-level Statistical genomics ML / DL

3 Research New disease mechanism Cohort

4 Research in context Gene Phenotype Treatment Age Cohort

5 Clinical grade in context Gene Phenotype Treatment Age

Clinical data warehouse

SPHN RDF / Structured database ontologies

Improved

Structured database omics results → Clinical data warehouse → Results in context Gene Phenotype Treatment Age Sex

Clinical data warehouse

We have developed new advances over the traditional best-practices in genomics. There are 5 distinct outputs from
the new genomics protocols compared to a single output of traditional clinical/reseach genomics.
(0) The tradtional approach focuses on annotation and filtering to find a genetic
cause of disease which often allows strong selective bias resulting in false positives.
The new approach relise on automated and non-biased approach that is uniform across all patients.
(1) A patient-specific clinical genetics report using standardisede guidelines and currently the best-practice.
(2) The same pipeline outputs evidence supporting new disease menaisms for research.
(3) The same pipeline outputs cohort-level data for statistical genomics, machine learning and deep learning projects.
(4) Results of those cohort-level projects included database-ready content including metadata added during the pipeline.
(5) Database ready results are suitable for clinical data warehouse for reporting and reuse.