

CASE STUDY

Fast and cost-effective whole-genome analysis



Objectives

To develop cost efficient whole-genome variant calling analysis pipeline that processes large batches of 50X samples and discover associations by aggregated rare variants.

Our approach

- Read alignment, germline SNV/Indel/SV/CN variant calling
- Joint genotyping and extensive QC
- Pipeline is Hg38 HLA and ALT-aware
- Broad Institutes GATK best practices

Results

We helped the client built a pipeline which annotates up to 150 50X whole genome sequencing samples.

