

Bioinformatics

at the Garvan Institute of Medical Research

The Garvan Institute, through the Sequencing Laboratory in the Kinghorn Centre for Clinical Genomics, specialises in providing an end-to-end service – from sequencing through to bioinformatics and data delivery.

With our award-winning variant calling and structural variation pipelines, our expert team can help you get the most from your sequencing data.

Award-winning analytics

Our GATK best practice pipeline was awarded the highest SNP precision in the precisionFDA Truth Challenge in 2016

Advanced software

Detect clinically relevant copy number variations in WGS data with ClinSV, our proprietary structural variation pipeline

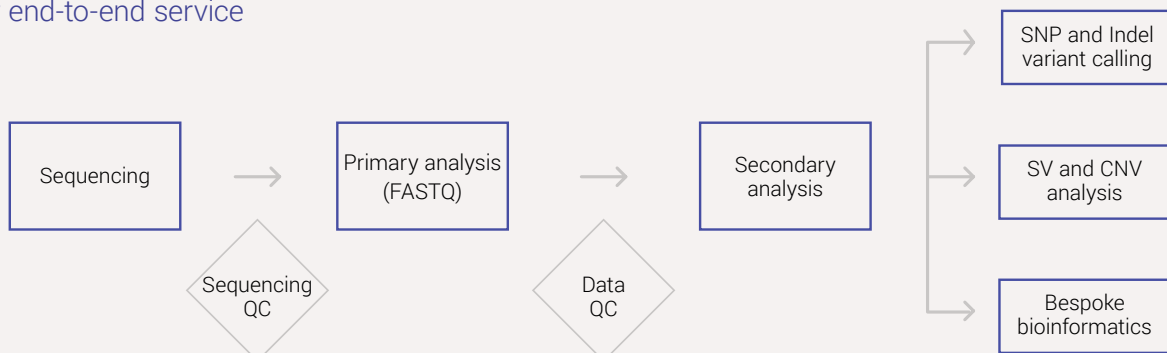
Rigorous quality control

All samples pass through our quality control pipeline to ensure the delivery of high quality data. Pipeline includes running FASTQC, Picard tools and more

Custom service

Scalable analytics that can be tailored to data from whole genome sequencing, whole exome sequencing or RNA sequencing

Our end-to-end service



Our secondary analysis

Best practice variant calling pipeline

GATK pipeline using industry best practice and reference genome hs37d5 (1000 Genome Project GRCh37 plus decoy)

- Analysis and delivery via DNAnexus platform
- Includes hosting of data on DNAnexus for 90 days
- Option to expand to SV and CNV analysis at additional cost (WGS data only)
- Files delivered:
 - Raw data (FASTQ)
 - Analysis ready alignment file (BAM)
 - Genomic VCF per sample (GVCF for joint calling)
 - VCF of joint called cohort (per submitted batch, annotated using VEP)

Structural variation (SV) and copy number variation (CNV) analysis (WGS data only)

ClinSV pipeline offers robust detection of clinically relevant structural and copy number variations

- Integration of three CNV signals: depth of coverage, split and spanning reads
- Extensive quality attributes and QC report
- Gene and phenotype annotation of each SV
- Multiple population allele frequency measures to help identify rare variants

Cohort Sequencing

End to end solution for your cohort sequencing needs including:

- Managing of genomic data archiving
- Best practices variant calling pipeline (GATK)
- Variant annotation
- Access to "Variant Atlas Platform": a genome analysis platform build by Garvan's Data Sciences Platform for visualising and filtering through your genomic data.

For a demo of the high coverage genomes in the 1000 Genomes Project Cohort on Variant Atlas please visit variantatlas.org.au

Bespoke bioinformatics

Our team can help with a range of custom bioinformatics and data analysis. Contact us to find out more or discuss your specific project needs

Location

KCCG Sequencing Laboratory is part of the Garvan Institute of Medical Research in Darlinghurst, NSW, Australia.

We receive samples from all over the world – visit garvan.org.au/sequencing for international sample requirements, or contact our team to discuss further.

Contact

To find out more or discuss your specific project needs, contact the KCCG Sequencing Laboratory team at kccgseq@garvan.org.au



Garvan Institute
of Medical Research