Garvan Institute of Medical Research

Whole Exome Sequencing
at the Garvan Institute of Medical Research

Whole exome sequencing (WES) targets only the protein-coding regions of the genome and can be a more focused, cost-effective option than whole genome sequencing.

The Garvan Institute, through the Sequencing Laboratory in the Kinghorn Centre for Clinical Genomics, offers a NATA (ISO15189) accredited WES service from library preparation through to data delivery. Our team have extensive experience in genomic research and can help create a bespoke service that fits your project goals.

Why use whole exome sequencing?

Greater depth of coverage
Analysing only the exome allows for a *greater* depth of coverage to identify SNVs and de novo variants, compared to WGS

Manageable data set
Utilising WES in research can produce a manageable, easy to interpret dataset

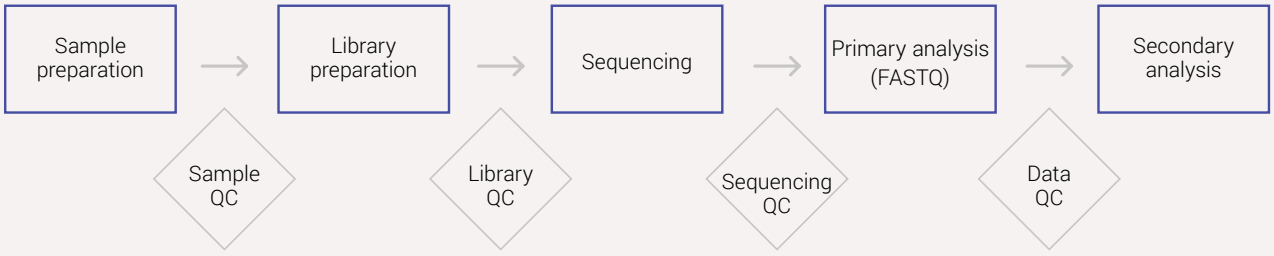
Applications across genomic research
WES can be used in gene discovery, clinical research, variant mapping, personalised medicine and agrigenomics

What we offer

The KCCG Sequencing Laboratory offers WES on the Illumina platform:

- Clinical grade end-to-end service, from library preparation through to data analysis
- ≥ 75% of bases above Q30 guaranteed
- Automated liquid handling and library preparations, with streamlined workflow for maximum efficiency
- Access our team of genomic research and bioinformatics experts for advice and support

How it works



Technical specifications

Sample	100µL of gDNA with a concentration between 10-40ng/µL and A260:A280 of >1.8
Sample type	Genomic DNA from blood, fresh frozen tissue, saliva, cell lines or single cells
Sequencing platforms	Illumina NovaSeq 6000 Illumina NextSeq 550Dx
Turn-around Time	Standard turn-around time (TAT) of 4-6 weeks from date of acceptance to date of data delivery (dependent on project size)*
Data specifications	≥75% bases above Q30
Data format	Raw data (FASTQ) delivered for all samples Alignments (Bam) and variants (VCF) available as secondary analysis through our bioinformatics service
Bioinformatics Services	Clinically accredited GATK pipeline using industry best practices and reference genome hs37d5 (1000 Genome Project GRCh37 plus decoy)
Delivery	Cloud-based data delivery including 90 days of hosting. Hard drive delivery available on request at an additional cost
Coverage	~70X or more for human samples Non-human samples will depend on the genomic size of the organism and what information the project is trying to extract

** Please contact the laboratory for any TAT concerns or specific requirements*

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