EXOME SEQUENCING

Empowering Australian Genomics

Focused, specific and cost effective.

Exome sequencing allows you to investigate the protein coding regions of the genome. It provides targeted, yet comprehensive re-sequencing which is suitable for screening the high value content of genomes economically.

Exome options

What is included

Exome sequencing at AGRF includes sample quality control, library preparation, exome capture and sequencing on the HiSeq 2500 with 2x100 bp paired-end reads.

We can also provide options for 50X and 100X mean coverage levels and low DNA input[i].

Agilent SureSelect Exomes are guaranteed to meet minimum specifications of the Agilent Certified Service Provider program[ii].

Reliable and consistent quality

Providing quality genomic services is our top priority

We operate to a unique set of quality standards: we are CPA, GLP/GCP accredited and we are also Agilent and Illumina CSPro certified.

Our Exome services have been accredited to the ISO/IEC 17025:2005 standard by the National Association of Testing Authorities (NATA) so you can be sure to receive industry leading data quality and service, consistently.

Human	Data/Sample	Minimum Submission	
Agilent SureSelect Human All Exon v5 (51Mb)	4Gb	3 samples	Superior performance at low cost.
Agilent SureSelect Human All Exon v5+UTR (75Mb)	6Gb	3 samples	Superior performance with greater genomic targets.
Agilent SureSelect Human All Exon v6 (58Mb)	5Gb	3 samples	Targets hard-to-capture and challenging multi-mapping regions.
Illumina Nextera Rapid Capture (37 Mb)	6Gb	12 samples	As chosen by the Melbourne Genomics Health Alliance[iii]
Mouse	**Data/Sample**	**Minimum Submission**	
Agilent SureSelect XT Mouse All Exon (50Mb)	4Gb	3 samples	Covers over 220,000 exons within 24,000 genes

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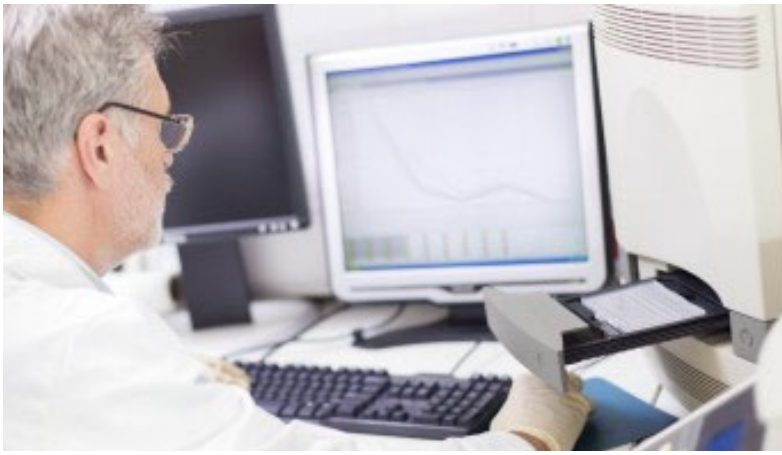
Advanced analysis

We know every project is unique. That is why we have a flexible approach to tailoring analysis to each project.

Optional data analysis includes:

- Structural analysis, including large deletions and translocations
- De novo mutation detection
- Somatic mutations
- Trio analysis
- Comparisons between tumor and matched normal samples.



Beyond exomes

As a complete genomics provider we offer a wide range of services, such as custom capture sequencing. This allows you to design custom captures to sequence specific regions of interest making it ideal for projects when you have a specific target in mind.

For whole human genome sequencing AGRF has partnered with the Kinghorn Centre for Clinical Genomics (KCCG) and the Illumina HiSeq X Ten system.

AGRF also has a large portfolio for verification or follow on genomic analysis, including SNP genotyping, methylation analysis and gene expression.

To find out more how we can match our expertise to your research goals please get in touch with us.

Notes:

i. Coverage is highly dependent on the sample quality, efficiency of hybridisation and PCR duplicate rate.

ii. Guaranteed 60% of reads in targeted regions, 75% of targeted bases with greater than or equal to 20 reads

iii. www.melbournegenomics.org.au

Specifications

Sample	High quality DNA: • Standard input: 5 µg requested • Low input: 500 ng requested
Inclusions	Exome analysis includes sample quality control, library preparation, exome capture, sequencing and basic analysis
Turn around time	4-6 weeks
Data format	Raw data, FASTQ files. Binary Alignment Map (BAM) file Variant Call Format (VCF) file
Data delivery	Delivered via secure FTP
Data analysis	Quality data trimming, alignment to reference, SNP and indel variant calling. Further analysis optional
Instrument specifications	Sequencing is performed on the HiSeq2500 system using paired-end (2x100 bp) protocol
Accreditation	NATA and ISO/IEC 17025:2005 Illumina Certified Service Provider (CSP pro) Agilent Certified Service Provider (NGS Target Enrichment)

Our funding partners

AGRF is a not-for-profit organisation supported by the Commonwealth Government infrastructure schemes administered through Bioplatforms Australia.

These schemes include NCRIS, EIF, Super Science Initiative CRIS and NCRIS 2.

NCRIS National Research Infrastructure for Australia
An Australian Government Initiative

BIOPLATFORMS AUSTRALIA

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