SOLUTIONS FOR CLINICAL GENOMICS

YOUR PARTNERS IN CLINICAL GENOMICS

Whole Genome Sequencing

Whole genome sequencing (WGS) provides the most comprehensive analysis of genome variance and structure. WGS enables accurate detection of single nucleotide variants (SNV), and structural variation of human genomes. AGRF offers a range of services for sequencing of small microbial genomes, large complex genomes and a clinical genome sequencing service for rare mendelian disorders, complex disease and cancer.

Our WGS Service

- ISO15189 Accreditation for Medical Testing
- Illumina PCR-Free DNA Library Preparation
- Illumina Sequencing : 150bp paired end reads
- Raw Coverage : 30x, 60x, 90x
- Germline variant calling

Deliverables: Alignment file, Variant call files, Genome coverage metrics, Contamination checks, Quality control statistics.

Whole Exome Sequencing

Whole exome sequencing (WES) is a powerful tool for investigation of genetic variance across the protein coding regions of the genome, providing high coverage and high confidence in variant calls for the study of rare mendelian disorders, complex diseases and cancer.

AGRF provides comprehensive solutions for WES, including bioinformatics using gold standard tools (BWA and GATK).

Our WES Service

- ISO15189 Accreditation for Medical Testing
- Agilent SureSelectXT Low Input Clinical Research Exome v2 (68Mb)
- Illumina Sequencing : 150bp paired end reads
- Mean On-Target Coverage >100x
- Germline variant calling

Deliverables: Alignment file, Variant call files, Target coverage metrics, Contamination checks, Quality control statistics.

Constitutional Cytogenetics

The identification of structural chromosomal aberrations can provide insight into causative relationships with complex phenotypes - including intellectual disability, developmental delay, and congenital anomalies. We use Illumina's CytoSNP-850k BeadChip cytogenetic microarray to reliably detect chromosomal imbalances of copy number and allelic homozygosity, which are commonly associated with genetic constitutional disorders.

Contact us: 1300 247 301
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Complex Disease Studies

Global Screening Array

Illumina's GSA contains 654,027 markers, of which 45,998 are ClinVar pathogenic and ClinVar likely pathogenic. The array also contains 301,888 markers for COSMIC genes for somatic mutations in cancer. The array can be used for pharmacogenomic applications and has 4,125 markers for PharmGKB which have applications in human genetic variation associated with drug responses.

Now available with booster content:
Multi-disease, Psych, Direct to Consumer, Cyto, Confluence (Breast Cancer)

Global Diversity Array

Illumina's GSA contains 1,831,442 markers and has a multi-ethnic backbone, with exceptional coverage and updated clinical research content

Variant Confirmation & Discovery

AGRF's clinical variant confirmation service can provide confirmation of SNVs and small indels using Sanger sequencing. Our service is NATA accredited to a clinical standard (ISO15189).

Options for translational research

RNA-Seq

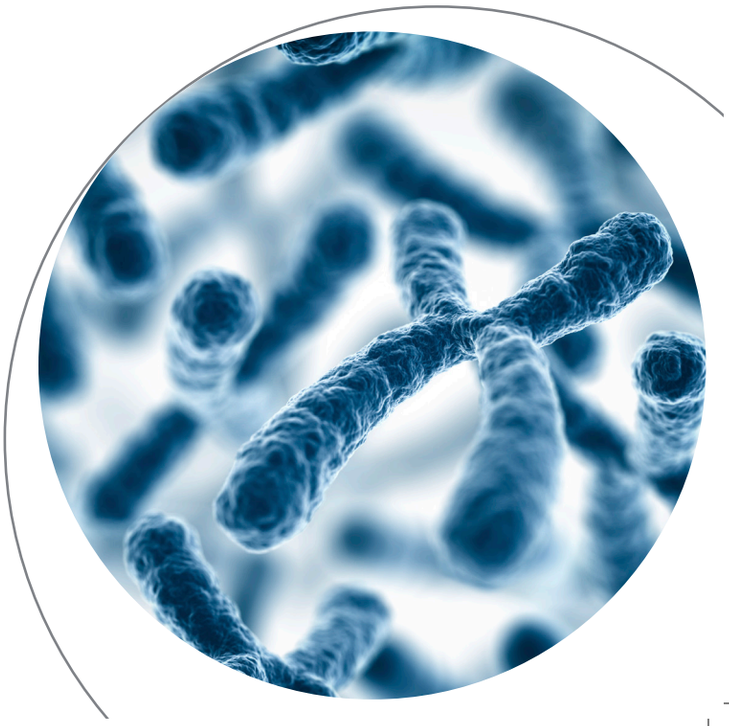
Exploration of gene expression and transcriptomes provides deep insights into the mechanisms of diseases. Use RNA-Seq to identify the dysregulation of gene transcription by putative pathogenic variants or detect fusion transcripts and expressed somatic mutations in cancer.

- mRNA-Seq: Capture Poly(A) transcript for the most sensitive option for detection of lowly expressed coding transcripts.
- Whole Transcriptome: captures both coding and noncoding RNA for a complete view of expression dynamics.

Methylation Profiling

We provide the option of using the Illumina MethylationEPIC array to characterise complex diseases and for tumour classification.

The EPIC Array differentiates >80 sub-entities in central nervous system tumours and approximately 60 sub-entities in sarcoma, as well as classifying epithelial cancer tumours.



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

NCRIS
National Research Infrastructure for Australia
An Australian Government Initiative

BIOPLATFORMS AUSTRALIA