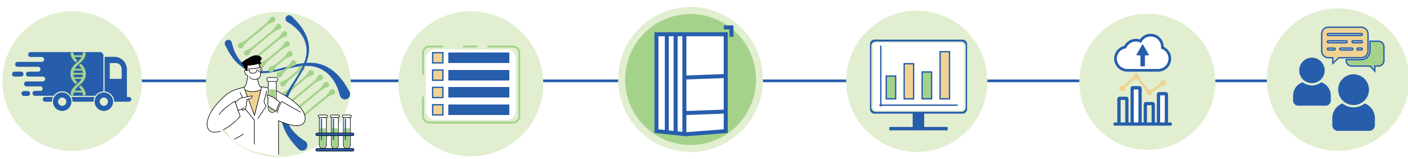
BMKGENE

Human Whole Exome Sequencing

Whole exome sequencing (WES) is a cost-effective strategy for identifying disease-causing mutations. Exons, though only about 1.7% of the genome, directly encode protein sequences, representing the core of cellular functions. In human genome, it is reported that over 85% of disease related mutations occur in protein coding region. BMKGENE offers comprehensive and flexible human WES services with different exon capturing strategies to meet various research goals.

Service Workflow



Sample delivery — DNA extraction — Library construction — Sequencing — Data analysis — Data delivery — After-sale services

Bioinformatics

Tumor Paired with Healthy Tissue

1.Sequencing Data Quality Control

2.Reference Genome Alignment

3.SNP/Somatic SNP Identification and Annotation

4.Small InDels/Somatic InDels Ide-ntification and Annotation

5. Advanced Analysis

- Susceptibility gene analysis (based on germline mutation)
- Mutation signatures analysis
  - Mutation spectrum analysis
  - Mutation feature NMF analysis
- Driver gene analysis
  - Predictive driver gene analysis
  - Known driver gene analysis
  - High-frequency mutated gene pathway enrichment analysis
- Genome-wide somatic variant circos plot

Hereditary Disease

1.Sequencing Data Quality Control
2.Reference Genome Alignment
3.SNP Identification and Annotation
4.Small InDels Identification and Annotation

5.Advanced analysis

- Pathogenic variant screening
- Incorporating phenotype analysis (family history of illness, inheritance pattern, etc.)

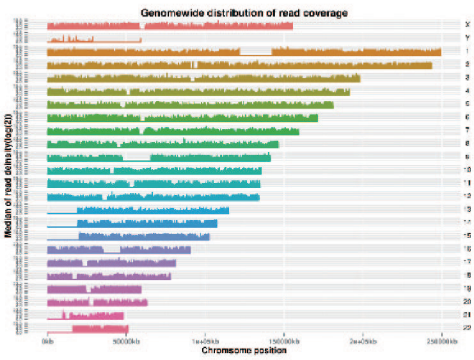
Service Specifications

Platform	Library	Exon Capture Strategy	Recommend Sequencing Strategy
Illumina NovaSeq	PE150	Agilent SureSelect Human All Exon V6 IDT xGen Exome Hyb Panel V2	10 Gb 5 Gb

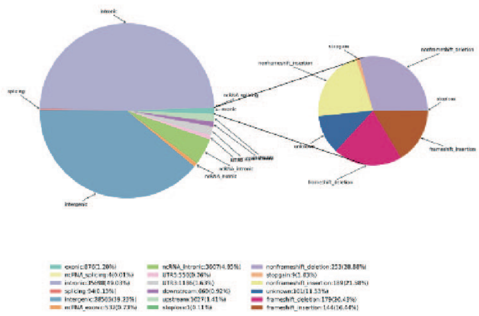
Sample Requirements

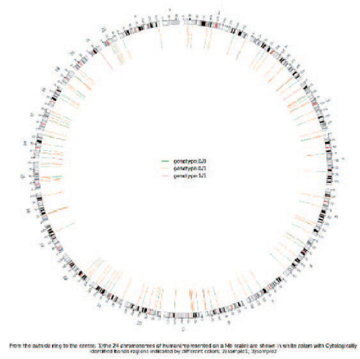
Sample Type	Amount	Volume	Concentration (Qubit®)	Quality
Genomic DNA	≥ 50 ng	≥ 15 μL	≥ 6 ng/μL	No or limited degradation or contamination

Demo Results

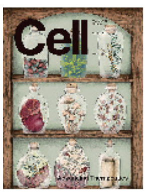


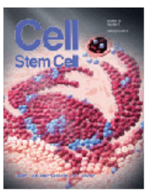
Chromosome coverage depth distribution

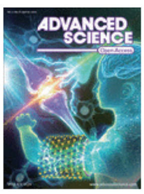

Variant annotation

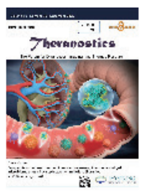

Genome-wide variants distribution

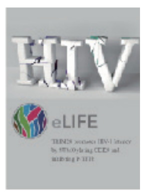
Featured Publications











BMKGENE

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