BMKGENE

Human Whole Exome Sequencing

Whole exome sequencing (WES) is a cost-effective strategy for identifying disease -causing mutations. Although exons only take up approximately 1.7% of the gen -ome, it represents the profile of total protein functions directly. In human genome, it was 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

Disease

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
  - Genomic somatic cell variation circos plot

Tumor

1.Sequencing Data Quality Control

2.Reference Genome Alignment

3.SNP Identification and Annotation

4.Small InDels Identification and Annotation

5.Advanced analysis

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

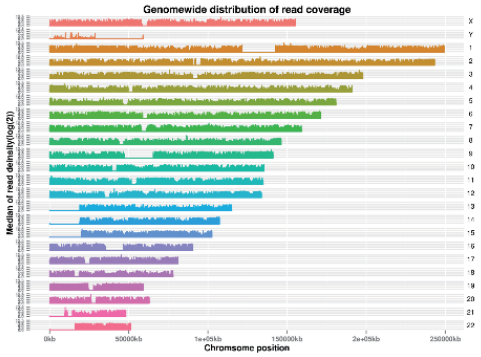
Service Specifications

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

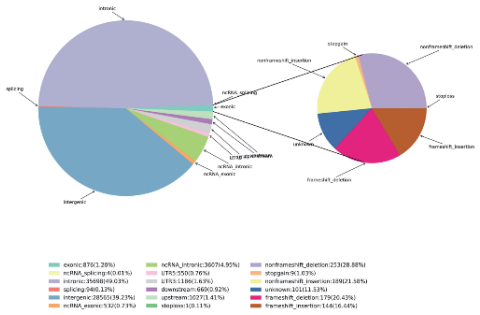
Sample Requirements

Sample Type	Amount(Qubit®)	Volume	Concentration	Purity(NanoDrop™)
Genomic DNA	≥ 300 ng	≥ 15 μL	≥ 20 ng/μL	OD260/280=1.8-2.0 no degradation， no contamination

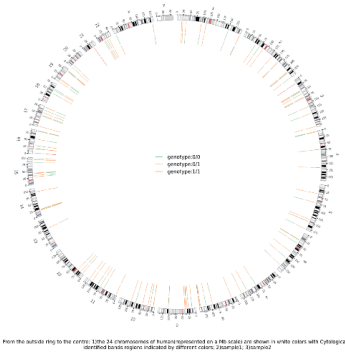
Demo Results



Chromosome coverage depth distribution map



Variant annotation



Genome-wide distribution of InDel

BMKGENE

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