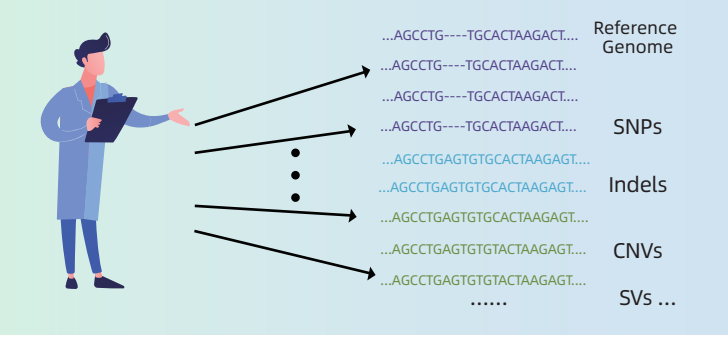
BMKGENE

Human Whole Genome Sequencing

Whole genome sequencing delivers a comprehensive collection of small and large genetic variations across the entire human genome. It provides fundamental data resources for characterizing genetic markers associ-ated with a variety of biological processes, which paves the way for new insights into diseases, cancers, population evolution, etc. BMKGENE offers human whole genome sequencing services on both Next-generation sequencing and Third-generation sequencing platforms to tackle different scientific questions.



Service Workflow

Experiment design → Sample delivery → DNA extraction → Library construction → Sequencing → Data analysis → After-sale services

Bioinformatics

❶ Raw data quality control
❷ Alignment with reference genome
❸ SNP identification and annotation
❹ Small InDel identification and annotation
❺ SV, CNV identification and annotation (Long read sequencing or high depth)
❻ Distribution of variations on genome
❼ Function annotation on variation (NR, SwissProt, GO, KEGG, COG, KOG, Pfam)
❽ Variant pathogenicity screening and functional enrichment analysis (Long read sequencing)

Service Advantages

Detects multiple types of variations	Whole genome sequencing detects various types of variations, including SNP, InDel, SV, CNV, etc.
Comprehensive genome information	Whole genome sequencing covers the entire genome, including exonic, intronic, non-coding, and intergenic regions, providing comprehensive detection.
Diverse platform options	BMKGENE offers PacBio, Nanopore long read sequencing, and NGS Illumina sequencing platforms.
Rigorous quality control process	BMKGENE provides real-time quality control tracking for DNA extraction, library quality, sequencing base quality, sequence alignment, etc.
Professional team	BMKGENE has professional experimental, analysis, and service teams.

Service Specifications

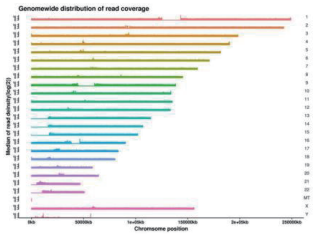
Platform	Library	Sequencing Depth
Illumina NovaSeq	PE150	30 -50 X
Nanopore PromethION 48	Nanopore - 8 Kb	≥30 X
PacBio Revio	CCS-HiFi	≥10 X

Sample Requirements

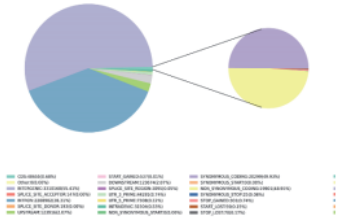
Platform	Conc. (ng/μl)	Amount (μg)	OD 260/280	
Illumina NovaSeq	≥1	≥0.2	1.6-2.5	Limited degradation and protein or RNA contamination
Nanopore PromethION 48	≥20	≥2	1.7-2.2	
PacBio Revio	≥50	≥10	1.7-2.2	

Featured Publications

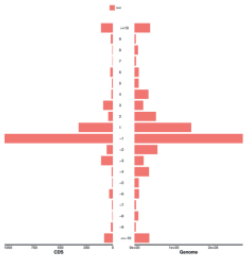
Demo Results



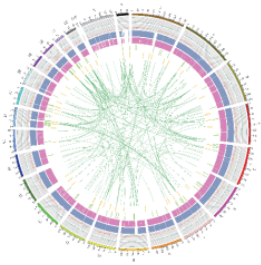
Distribution of chromosome coverage depth



SNP/InDel Annotation



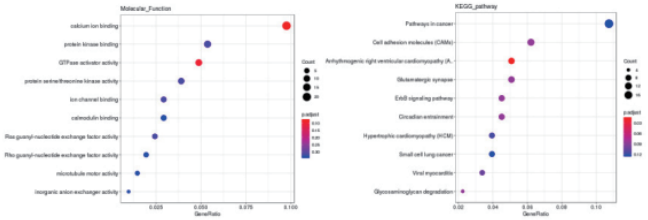
InDel length distribution



Circos plot of variation distribution

#Variant Region	All	test
Total	795,299	795,299
Pathogenic	18	
Likely pathogenic	528	528
VUS	296,436	296,436
Likely benign	178	178
Benign	498,139	498,139

Statistics of variant pathogenicity classification



GO/KEGG Functional Enrichment

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