

DNBSEQ™ SERVICE OVERVIEW

Human Whole Genome Sequencing



Service Description

Whole Genome Sequencing (WGS) determines the complete human genome sequence at one time and provides the most comprehensive collection of an individual's genetic variation based on the human reference genome.

WGS can be applied to human genetics and evolution studies to detect genome-wide genetic variations, pathogenic and susceptibility genes, and to enable genetic diversity and evolution analysis. It can also be applied in translational research to provide information on cancer and other disease-associated mutations and is one of the most important tools for precision medicine.

We offer a comprehensive range of WGS services for many sample types and coverage levels.

Sequencing Service Specification

Our Human Whole Genome Sequencing Services are performed with DNBSEQ™ sequencing technology, featuring DNA Nanoballs, linear Rolling Circle Replication and cPAS technology for superior data quality.



- PCR and PCR-free library methods are available
- 100 bp and 150 bp Paired end sequencing options
- Choice of sequencing depth: standard (~30X), deep (~60X) and low pass (less than 10X)
- Raw data, standard and customized bioinformatics analysis available
- Available data storage and bioinformatics applications



Sequencing Quality Standard

- Guaranteed ≥85% of bases with quality score of ≥Q30

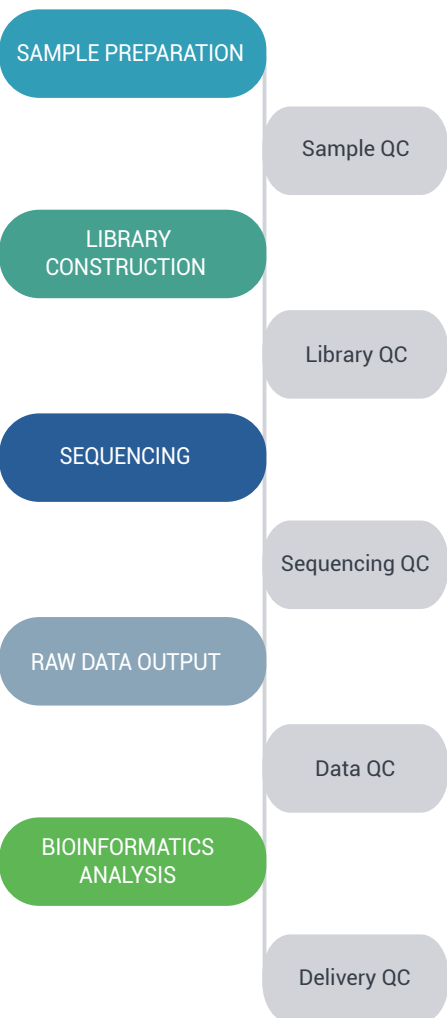


Turnaround Time

- Standard Turnaround: typical 18 working days from sample QC acceptance to filtered data availability
- Rapid Turnaround: 10 working days from sample QC acceptance to filtered data availability
- Expedited services are available for all WGS sequencing options, contact our specialist for details

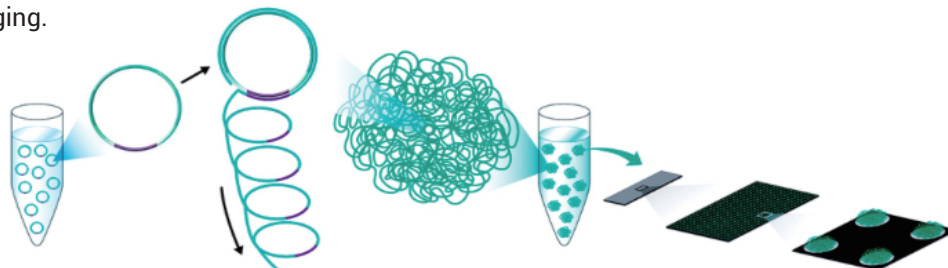
Project Workflow

We care for your samples from the start through to the result reporting. Highly experienced laboratory professionals follow strict quality procedures to ensure the integrity of your results.



DNBSEQ™ Sequencing Technology

DNBSEQ™ is an innovative high-throughput sequencing solution, developed by BGI's Complete Genomics subsidiary in Silicon Valley. The system is powered by combinatorial Probe-Anchor Synthesis (cPAS), linear isothermal Rolling-Circle Replication and DNA Nanoballs (DNB™) technology, followed by high-resolution digital imaging.



The combination of linear amplification and DNB technology reduces the error rate while enhancing the signal. The size of the DNB is controlled in such a way that only one DNB is bound per active site on the flow cell. This densely patterned array technology provides optimal sequencing accuracy and increases flow cell utilization.

Data Analysis

In addition to clean data output, we offer a range of standard and customized bioinformatics pipelines for your human WGS project.

Reports and output data files are delivered in industry standard FASTQ, BAM. Excel formats with publication-ready tables and figures.

STANDARD BIOINFORMATICS ANALYSIS

- Filtering
- Alignment
- SNP calling and annotation
- SNP validation and comparison
- SNP functionality and conservation prediction
- SNP Statistics per functional element
- InDel calling and annotation
- InDel validation and comparison
- InDel statistics per functional element
- CNV calling and annotation
- SV calling and annotation

ADVANCED ANALYSIS

Advance Analysis	Contents
Cancer Analysis Paired samples are required (Normal tissue and Tumor tissue from the same person)	Somatic Mutation Calling 1 Somatic SNV calling and annotation; 2 Somatic InDel calling and annotation; 3 Somatic SV calling and annotation; 4 Somatic CNV calling and annotation; Cancer Classical Analysis; 1 Purity and Ploidy; 2 Homology Test; 3 Cancer Susceptibility Genes; 4 Drug Targeted Annotation; 5 Prediction of Driver Genes; 6 Identifying Significant Mutated Genes; 7 Deciphering Mutational Signature; 8 CN-neutral Loss of Heterozygosity; 9 Hyper-mutated Sample Classification; 10 Clone Analysis;

ADVANCED ANALYSIS	
Advance Analysis	Contents
Mendelian Disorders Analysis	Public group AF annotation; Scoring 12 harmful or conservative prediction tools; Signaling pathway annotation; OMIM annotation; Normal tissue protein expression annotation; Data screening and interpretation 1 Screening according to ACMG genetic variation classification criteria and guidelines 2 Screening by threshold 3 <i>De novo</i> mutation screening (optional, Parental samples are required) 4 Analysis of family co-separation 5 RoH analysis (optional, pedigree is consanguineous union)

CUSTOM ANALYSIS

Further customization of Bioinformatics analysis to suit your unique project is available: Please contact your technical representative.

Sample Requirements

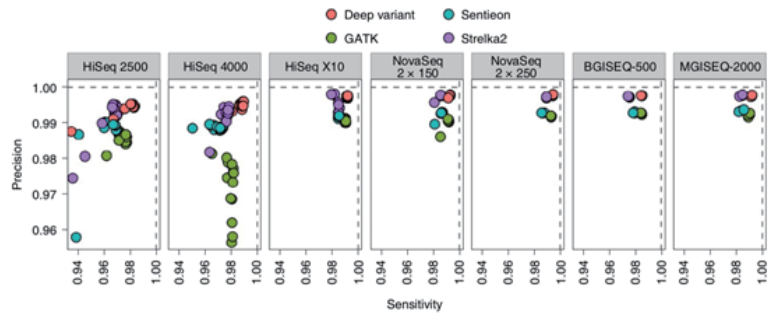
We can process your gDNA, saliva, blood, fresh frozen tissue, cell pellets and FFPE samples, with the following general requirements:

DNA Sample	Library type	Mass	Concentration	Integrity (AGE)	Sample Purity
Regular Samples	PCR	≥200 ng (Recommend ≥400 ng)	≥8 ng/μL	The band shown on gel electrophoresis has little degradation, or of fragment size greater than 20 kb.	No contamination with RNA, protein or salt ions; colorless and transparent; non-sticky
	PCR-free	≥1 μg (Recommend ≥2 μg)	≥12.5 ng/μL		
Low Input/ FFPE Samples	PCR	≥50 ng	≥2.5 ng/μL	Fragment size greater than 500 bp.	

Superior SNP/InDel Detection

Here the Association of Biomolecular Resource Facilities (ABRF) Next-Generation Sequencing Study^[1] benchmarks the performance of a set of sequencing instruments on human reference DNA samples. It is further confirmed that the DNBSEQ™ platform has greater advantages in error rate and SNP/InDel detection.

- Among short-read instruments, the DNBSEQ™ platform provided the lowest sequencing error rates.
- For SNP/InDel, the DNBSEQ™ platform had the greatest sensitivity and precision.



Common germline haplotype variant callers were compared for each sequencing platform

References

[1]. Foox, J., Tighe, S.W., Nicolet, C.M. et al. Performance assessment of DNA sequencing platforms in the ABRF Next-Generation Sequencing Study. *Nat Biotechnol* 39, 1129–1140 (2021).

Request for Information or Quotation

Contact a BGI Genomics representative to discuss how we can meet your specific needs or for expert advice on experiment design, from sample to bioinformatics.

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