

# 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.

BGI offers a comprehensive range of WGS services for many sample types and coverage levels.

### Sequencing Service Specification

BGI Human Whole Genome Sequencing Services are performed with BGI's 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
- 100bp and 150bp 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 ≥80% of bases with quality score of ≥Q30

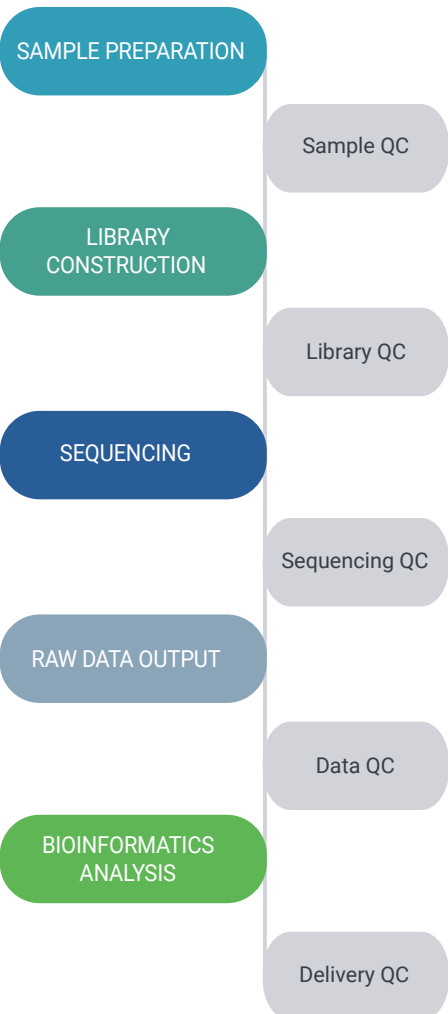


#### Turnaround Time

- 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 your local BGI specialist for details

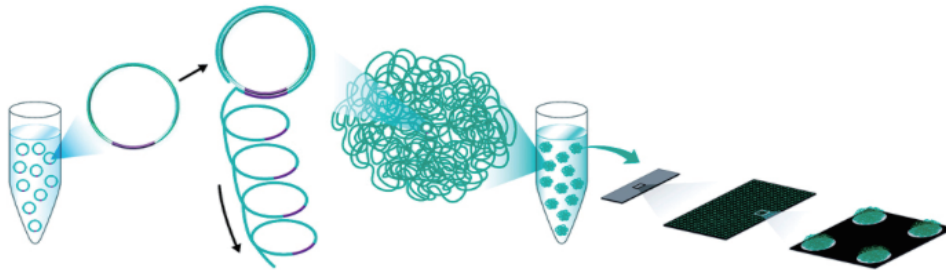
### Project Workflow

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-Ancor 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, BGI offers 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

### CUSTOM ANALYSIS

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

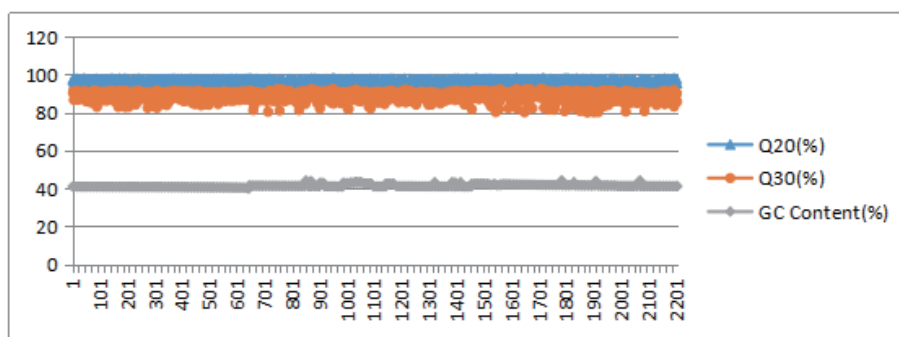
## Sample Requirements

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

	Library type	DNA Amount and Concentration	Minimum Sample Volume
Regular Samples	PCR	Intact genomic DNA $\geq 1\mu\text{g}$ Concentration $\geq 12.5\text{ng}/\mu\text{l}$	15 $\mu\text{l}$
	PCR-free	Intact genomic DNA $\geq 2\mu\text{g}$ Concentration $\geq 12.5\text{ng}/\mu\text{l}$	15 $\mu\text{l}$
Low Input Samples	PCR	Intact genomic DNA $\geq 200\text{ng}$ Concentration $\geq 2.5\text{ng}/\mu\text{l}$	15 $\mu\text{l}$

## Stable and High-Quality Data Performance

2,206 Samples were randomly selected from over 20,000 samples that were sequenced at BGI's laboratories over a period of 6 months. The data output and data quality remained stable over that period. The average Q20 and Q30 scores were 96.78% and 88.81% respectively. The average GC content was 41.25% without obvious base bias.

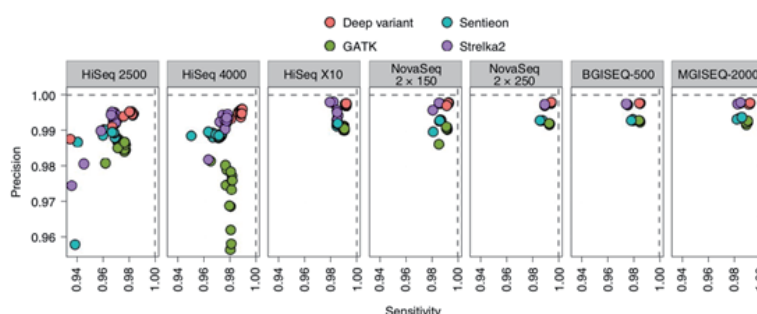


Stable Data quality scores and GC content from our DNBSEQ™ production line

## Superior SNP/InDel Detection

Here the Association of Biomolecular Resource Facilities (ABRF) Next-Generation Sequencing Study<sup>[1]</sup> 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 your BGI account representative for the most affordable rates in the industry and to discuss how we can meet your specific project requirements or for expert advice on experiment design, from sample to bioinformatics.

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We Sequence, You Discover