SERVICE OVERVIEW

BGISEQ-500 Whole Genome Sequencing

Service Description

Whole genome sequencing detects 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. Whole genome sequencing 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 to translational research to provide information on cancer and disease-associated mutations and is one of the most important approaches of precision medicine.

Sequencing Service Specification

BGI Human Genome Sequencing Service are performed with the BGISEQ-500 sequencing system, featuring cPAS and DNA Nanoballs (DNB™) technology for superior data quality.

- 100bp paired end sequencing
- Raw data, standard and customized data analysis
- Available data storage and bioinformatics applications

ACTGACT
TACTACT
GACTAC
CTGAGGT

Sequencing Quality Standard

- Guaranteed ≥80% of bases with quality score of ≥Q30
- Raw data is qualified

24/7

Turn Around Time

- Typical 40 working days from sample QC acceptance to filtered raw data availability
- Expedited services is available, contact your local BGI specialist for details

BGISEQ-500 Sequencing Technology

BGISEQ-500 is an industry leading high-throughput sequencing solution, powered by combinatorial Probe-Anchor Synthesis (cPAS) and improved DNA Nanoballs (DNB™) technology. The cPAS chemistry works by incorporating a fluorescent probe to a DNA anchor on the DNB™, followed by high-resolution digital imaging. This combination of linear amplification and DNB™ technology reduces the error rate while enhancing the signal. In addition, the size of the DNB™ is controlled in such a way that only one DNB™ is bound per active site. This patterned array technology not only provides sequencing accuracy, but it also increases the chip utilization and sample density.

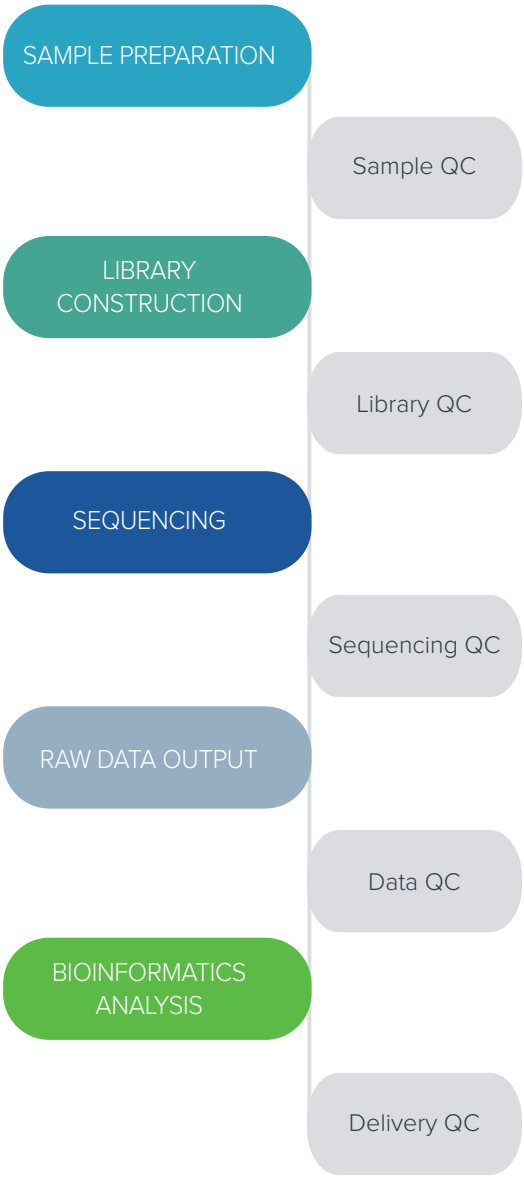
Sequencing Technology References

Drmanac R, Sparks AB, Callow MJ, Halpern AL, Burns NL, Kermani BG, Carnevali P, Nazarenko I, Nilsen GB, Yeung G, et al. Human genome sequencing using unchained base reads on self-assembling DNA nanoarrays. Science. 2010;327(5961):78–81.

Quality Data | Fast TAT | Cost Effective

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.



Data Analysis

Besides clean data output, BGI offers a range of standard and customized bioinformatics pipelines for your whole genome sequencing project.

Reports and output data files are delivered in industry standard file formats: BAM, .xls, .png.

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.

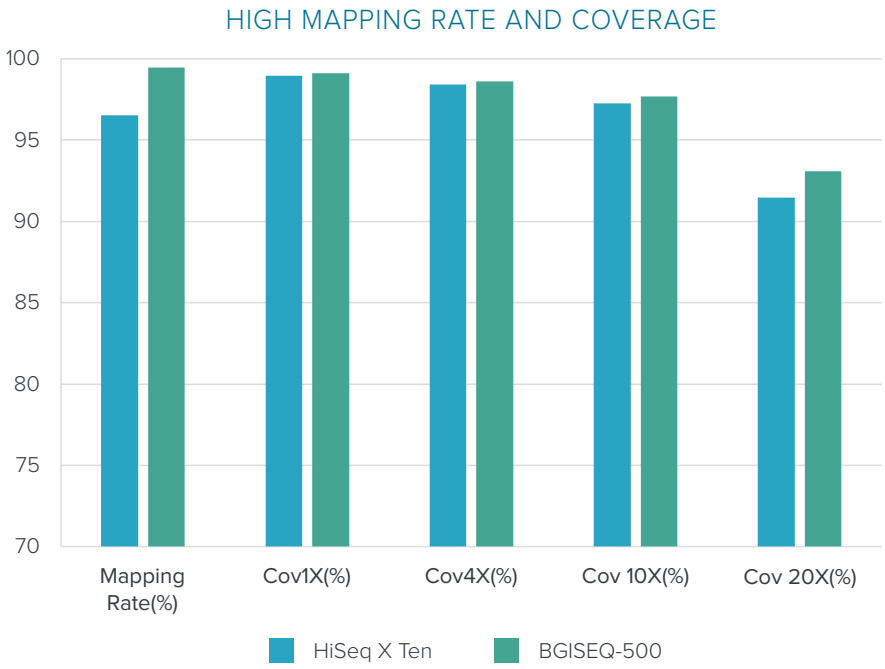
Sample Requirements

We can process your gDNA, Blood, Cell line, Fresh frozen tissue samples from a variety of species, with the following general requirements:

Sample Types	Concentration and Total Mass		Volume	Quality	Comment
Genome DNA	m≥2μg	c≥12.5ng/μL	15~100μL	No degradation or slight degradation	Strongly Recommended
	1μg≤m<2μg				Required

Data Performance

Following is an example of typical BGISEQ-500 data output for a 30X WGS project with standard sample NA12878, compared with data from the Illumina HiSeq X Ten system.



Bar-Graph showing the mapping rate and sequencing coverage of the samples using BGISEQ-500 and Illumina HiSeq X Ten platform of 30X WGS.

	BGISEQ-500	HiSeq X Ten
Clean Reads	1001630550	732165210
Clean Bases (Mb)	100163	110083
Mapping Rate	99.47%	96.52%
Unique Rate	94.33%	85.14%
Duplicate Rate	1.77%	11.76%
Mismatch Rate	0.53%	0.56%
Average Sequencing Depth	33.02	31.57
Coverage	99.10%	98.95%
Coverage at least 4X	98.62%	98.43%
Coverage at least 10X	97.68%	97.24%
Coverage at least 20X	93.09%	91.45%

Table showing comparison of relevant data from BGISEQ-500 and Illumina HiSeq X Ten platforms. SNP consistency is about 94.06%, the BGISEQ-500 platform provides lower false positive ratio and higher precision. As a whole, BGISEQ-500 platform provides comparable results to the Illumina HiSeq X Ten platform.

Request Information or Quotation

Contact your BGI account representative for the most affordable rates in the industry, 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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For BGISEQ-500 Whole Genome sample shipping instructions or sample submission forms, please visit our website.

www.bgi.com

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Services are for research use only.