BGISEQ-500 SERVICE OVERVIEW

Human Whole Genome Sequencing

Service Description

Whole genome sequencing 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. 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 in 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 bioinformatics analysis available
- Available data storage and bioinformatics applications

Sequencing Quality Standard

- Guaranteed ≥80% of bases with quality score of ≥Q30

Turn Around Time

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

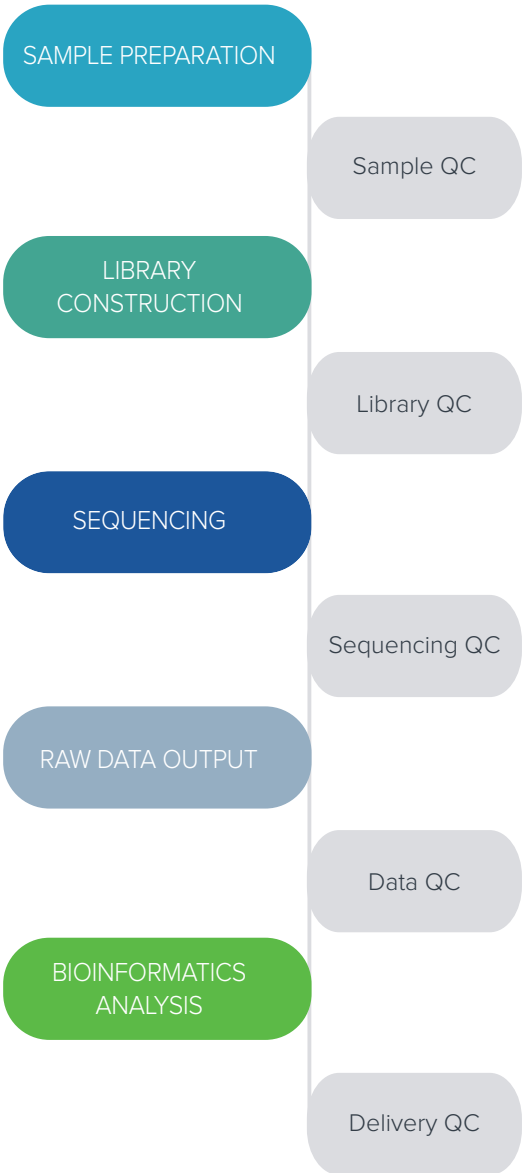
BGISEQ-500 Sequencing Technology

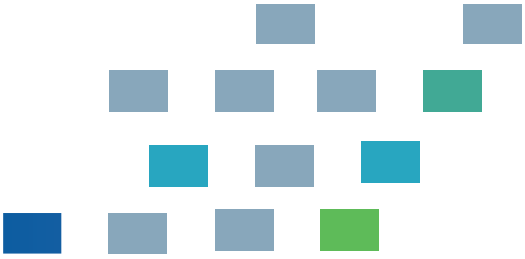
BGISEQ-500 is an industry leading high-throughput sequencing solution, powered by combinatorial Probe-Anchor Synthesis (cPAS) and improved DNA Nanoballs (DNB™) technology*. The cPAS chemistry works by incorporating a fluorescent probe to a DNA anchor on the DNB™, followed by high-resolution digital imaging. 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.

* 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.

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

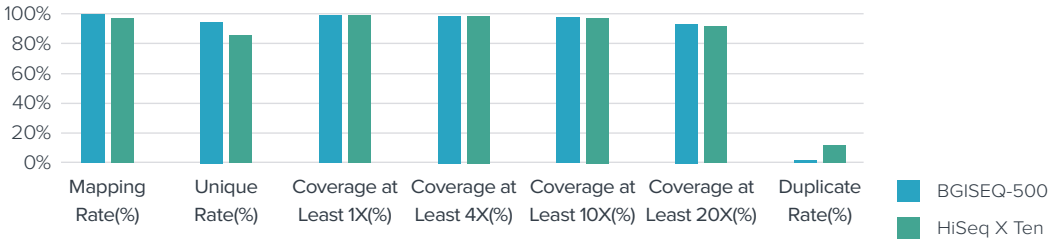
We can process your gDNA, blood, cell line, fresh frozen tissue samples from a variety of species, with the following general requirements:

	DNA CONCENTRATION	MINIMUM SAMPLE VOLUME
Regular Samples	Intact genomic DNA ≥ 1μg, Concentration ≥ 12.5 ng/ μl	15 μl
Low Input Samples	Intact genomic DNA ≥ 200 ng, Concentration ≥ 2.5 ng/ μl	15 μl

Data Performance

Mapping Rate and Coverage

The data below shows significantly lower duplicate rate, higher unique rate and comparable coverage in BGISEQ-500 PE100 data compared to the Illumina HiSeq X Ten platform, with 30X coverage WGS of the NA12878 standard sample.

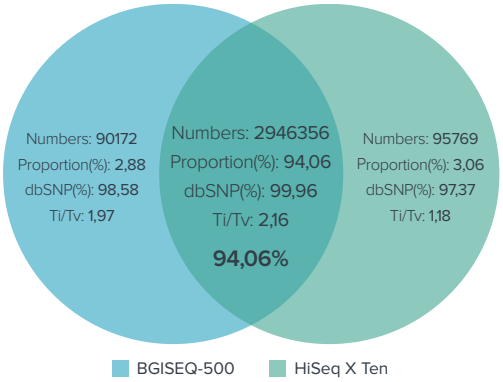


Item	BGISEQ-500	HiSeq X Ten
Clean Reads	1,001,630,550	732,165,210
Clean Bases (Mb)	100,163	110,083
Mapping Rate	99.47%	96.52%
Unique Rate	94.33%	85.14%
Duplicate Rate	1.77%	11.76%
Mismatch Rate	0.53%	0.56%
Average Sequencing Depth	33.02	31.57
Coverage	99.10%	98.95%
Coverage at least 4X	98.62%	98.43%
Coverage at least 10X	97.68%	97.24%
Coverage at least 20X	93.09%	91.45%

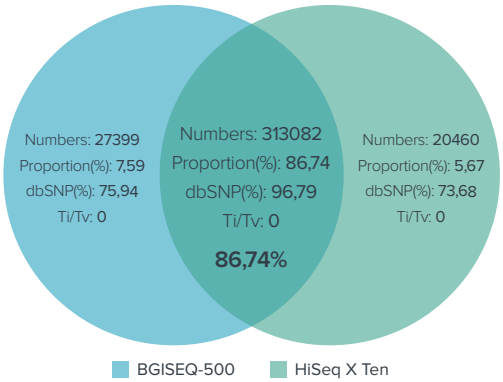
Correlation, Precision, Sensitivity

The BGISEQ-500 platform shows comparable precision and sensitivity compared to the HiSeq X Ten PE150 data, using the high confident variations previously identified in NA12878 and provided by GIAB (Genome In A Bottle), using the methods provided by GIAB.

Comparison of data from BGISEQ-500 and Illumina HiSeq X Ten platforms shows equivalent sequencing results. SNP and InDel consistency exceeds 94% and 86% respectively.



SNP ANALYSIS	BGISEQ-500	HiSeq X Ten
Precision	99.94%	99.89%
Sensitivity	96.21%	96.34%



INDEL ANALYSIS	BGISEQ-500	HiSeq X Ten
Precision	97,62%	96.55%
Sensitivity	93.23%	90.32%

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 Human Whole Genome Sequencing sample shipping instructions or sample submission forms, please visit our website.

www.bgi.com

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