BGISEQ-500 SERVICE OVERVIEW

Whole Exome Sequencing

Service Description

For many applications, Whole Exome Sequencing is gaining popularity as a viable and cost-effective alternative for Whole Genome Sequencing. BGI has performed professional sequencing services for many years at several locations around the world to support human, animal and plant research and to benefit small and large-scale clinical trials and pharmaceutical drug development projects.

Sequencing Service Specification

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

Sample Preparation and Services

- Agilent Sureselect, BGI exome V4 kit for library construction and enrichment
- 100bp Paired-end sequencing
- Clean data and bioinformatics analysis are available in standard file formats
- Standard and custom bioinformatics data analysis
- Available data storage and bioinformatics applications

Sequencing Quality Standard

- Guaranteed ≥80% of bases with quality score of ≥Q30
- Standard sequencing coverage ≥50X; ≥100x is recommended for cancer samples

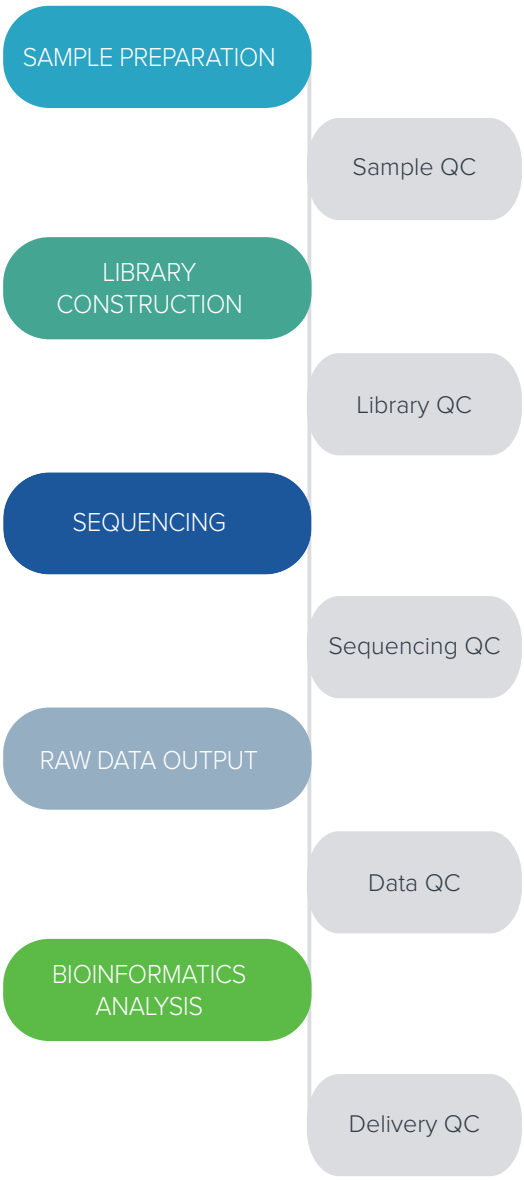
Turn Around Time (for pilot project)

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

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 exome 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

AVAILABLE ADVANCED ANALYSIS

- Cancer Somatic Mutation analysis
- Population genetics analysis
- Complex disease analysis
- Mendelian disease analysis
- *De novo* mutation analysis for family samples

CUSTOMIZED 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:

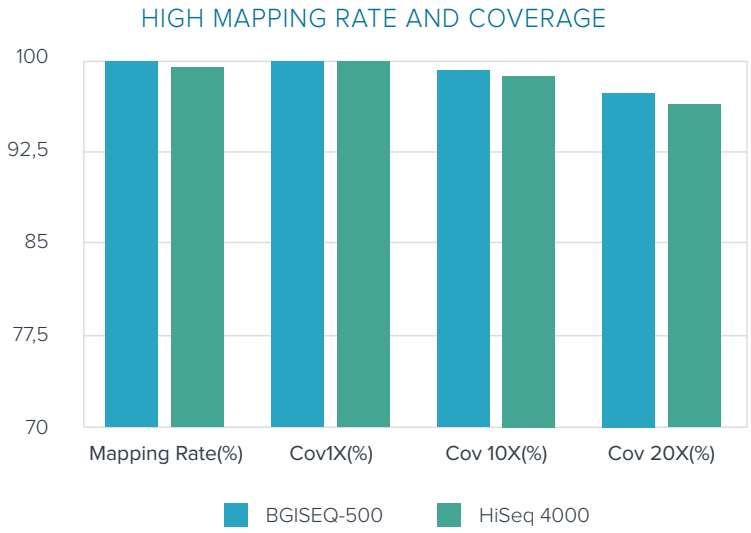
	DNA CONCENTRATION	MINIMUM SAMPLE VOLUME
Regular Samples	Intact genomic DNA ≥ 1μg, Concentration ≥ 12.5 ng/ μl	15 μl

BGISEQ-500 Sequencing Technology

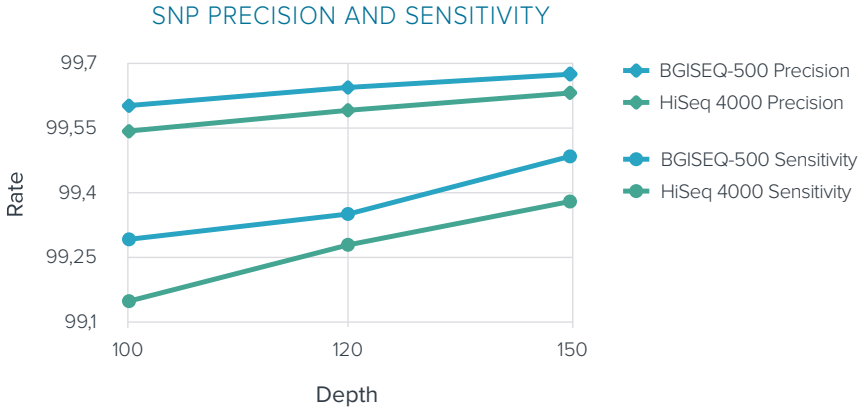
BGISEQ-500 is an industry leading high-throughput sequencing solution, powered by combinatorial Probe-Anchor Synthesis (cPAS) and improved DNA Nanoballs (DNB™) technology*. The cPAS chemistry works by incorporating a fluorescent probe to a DNA anchor on the DNB™, followed by high-resolution digital imaging.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

Data Performance



Bar-Graph showing the mapping rate and sequencing coverage of the samples using BGISEQ-500 and Illumina HiSeq 4000 platform of 100X WES.



Compared to the Illumina HiSeq4000 platform, BGISEQ-500 platform provides higher precision and sensitivity.

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.

info@bgi-international.com

International Head Offices

United States
One Broadway, 3rd Floor
Cambridge, MA 02142,
USA
Tel: **+1 617 500-2741**

Europe
Ole Maaløes Vej 3,
DK-2200 Copenhagen N,
Denmark
Tel: **+45 7026 0806**

Asia Pacific
16 Dai Fu Street,
Tai Po Industrial Estate,
New Territories, Hong Kong
Tel: **+852 36103510**

For BGISEQ-500 Whole Exome Sequencing sample shipping instructions or sample submission forms, please visit our website.

www.bgi.com

Published March 2017, version 1.0

Services are for research use only.