

# BGI 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 multiple locations globally, to support biomedical research and to benefit small and large-scale clinical trials and other projects in pharmaceutical drug development.

Besides raw sequencing data output, BGI offers standard and custom bioinformatics services to suit your specific research needs.

## Sequencing Service Specification

BGI Whole Exome Sequencing services are executed with the Illumina HiSeq 4000 sequencing system.



### Sample preparation and services

- Agilent Sureselect, NimbleGen SeqCap or KAPA Hyper Prep kit for library construction and enrichment
- 100bp and 150bp paired-end sequencing options available
- Clean data and bioinformatics analysis are available in standard file formats
- Advanced and custom bioinformatics data analysis available
- Cloud-based data storage and delivery system



### Sequencing Quality Standard

- Guaranteed  $\geq 80\%$  of bases with quality score of  $\geq Q30$
- Standard sequencing coverage  $\geq 50X$ ;  $\geq 150X$  is recommended for cancer samples



### Turn Around Time

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

## Locations

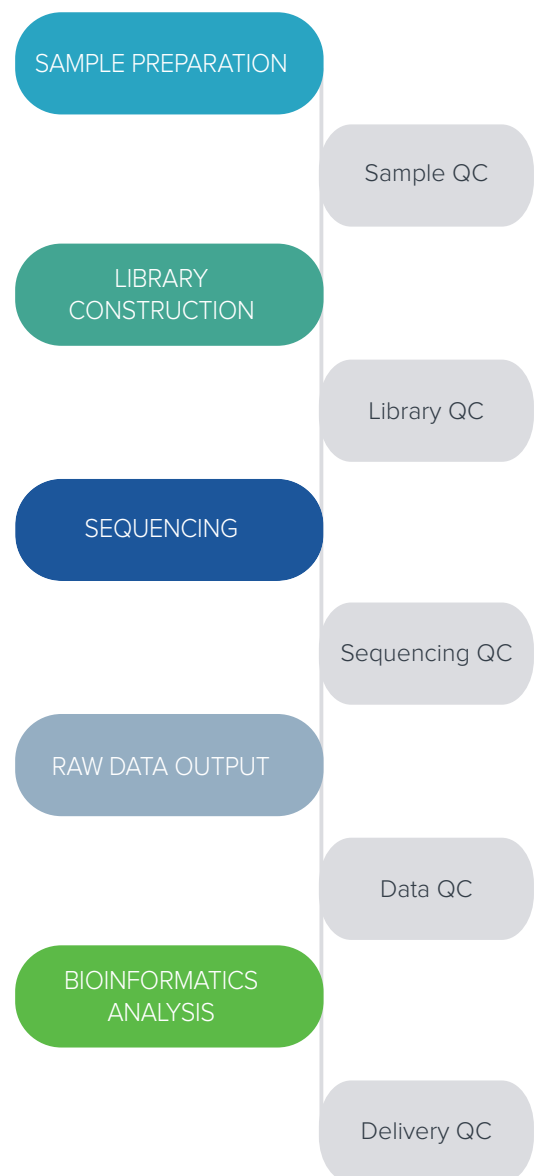
BGI's sequencing services are available from a variety of BGI service laboratory locations, including:

- Copenhagen, Denmark
- Philadelphia, USA
- Hong Kong, China

BGI also has a network of laboratories across mainland China.

## 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.



Quality Data



Fast TAT



Cost Effective

## Data Analysis

Besides raw data output, BGI offers a range of standard and customized bioinformatics pipelines for your exome sequencing project.

Reports and output data files are delivered in industry standard file formats: FASTQ, BAM, VCF, CNS and Excel.

### STANDARD 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, FFPE samples and single cell applications from human and rodents, with the following general requirements:

	DNA MASS AND CONCENTRATION	MINIMUM SAMPLE VOLUME
Regular Samples	Intact genomic DNA $\geq 1\mu\text{g}$ , Concentration $\geq 12.5 \text{ ng}/\mu\text{l}$	15 $\mu\text{l}$
Low Input Samples	Intact genomic DNA $\geq 200 \text{ ng}$ , Concentration $\geq 2.5 \text{ ng}/\mu\text{l}$	15 $\mu\text{l}$

## 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 BGI's scientific publications relating to Whole Exome Sequencing, sample shipping instructions or sample submission forms, please visit our website.

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[www.bgi.com](http://www.bgi.com)