

# BGI Human 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. Microarray-based genotyping studies have gradually moved to whole genome sequencing with the falling costs of sequencing technology. 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 Whole Genome Sequencing services are executed with the Illumina HiSeq X Ten sequencer sequencing system.



### Sample Preparation and Services

- 150bp paired end sequencing
- Clean data, standard and customized data analysis
- Cloud-based data storage and delivery system



### Sequencing Quality Standard

- Guaranteed  $\geq 75\%$  of bases  $\geq Q30$
- Standard sequencing coverage  $\geq 30X$ ;  $\geq 60X$  is recommended for cancer samples



### Turn Around Time

- Typical 40 working days from sample QC acceptance to raw data availability

## Locations

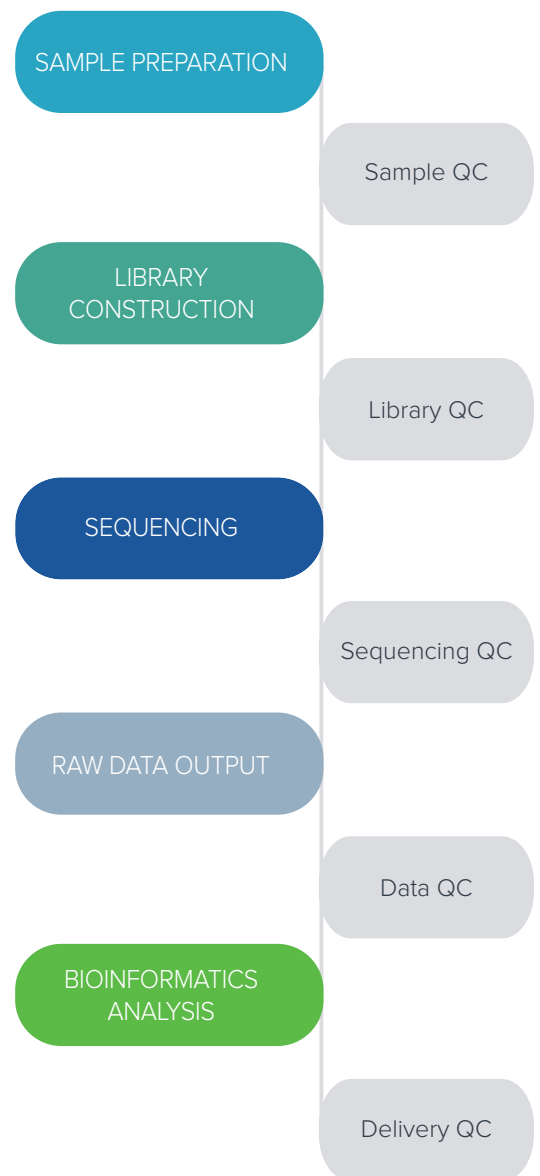
BGI's sequencing services are available from a variety of BGI service laboratory locations, including:

- 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.



## Data Analysis

In addition to 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: 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
- CNV calling and annotation
- SV calling and annotation

### 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 and FFPE samples and single-cell applications, with the following general requirements:

	DNA CONCENTRATION	MINIMUM SAMPLE VOLUME
Regular Samples	Intact genomic DNA $\geq 1\mu\text{g}$ , Concentration $\geq 50 \text{ ng}/\mu\text{l}$	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 Human Genome Sequencing, sample shipping instructions or sample submission forms, please visit our website.



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