DNBSEQ™ SERVICE OVERVIEW
Low-Pass Human Whole Genome Sequencing

Service Description

Low-pass Whole Genome Sequencing (WGS) provides an accurate and cost-effective solution to measuring genome-wide genetic variation. Low-pass WGS is an increasingly-popular high-throughput tool for large-scale genomics projects that traditionally have used legacy technology like genotyping arrays. This technology outperforms genotyping arrays by providing an order of magnitude more data, greater statistical power, and enhanced variant discovery capabilities. Applications include genome-wide association studies, biobank profiling, and pharmacogenomics. In addition, low-pass WGS can be used to build custom reference panels to improve imputation of future samples from a specific population or disease group. The unique combination of BGI DNBSEQ sequencing and Gencove's ImputeSeq low-pass imputation pipeline enables researchers to efficiently obtain fully-imputed, analysis-ready VCF files from DNA in a single service.

DNBSEQ™ Low-pass Sequencing	+	ImputeSeq™ Imputation and Data Analysis

Sequencing Service Specification

BGI Human Genome Sequencing Services are performed with BGI's DNBSEQ sequencing technology, featuring DNA Nanoballs, linear Rolling Circle Replication and cPAS technology for superior data quality.

- PCR and PCR-free library methods are available
- 100bp or 150bp Paired end sequencing
- Choice of sequencing depth: low pass WGS (1x, 4x, or custom)
- CAP/CLIA laboratory services available

Sequencing Quality Standard

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

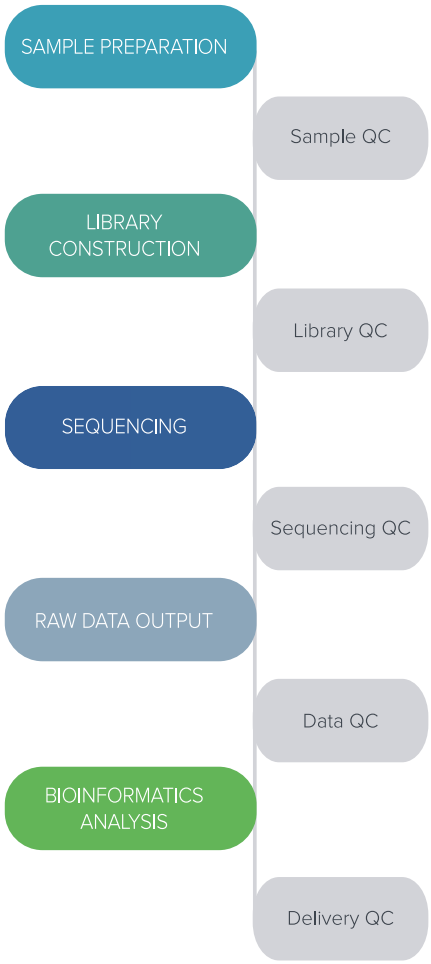
Turnaround Time

- Typical 18 working days from sample QC acceptance to data delivery
- Expedited services are available, contact your local BGI specialist for details

gencove

Project Workflow

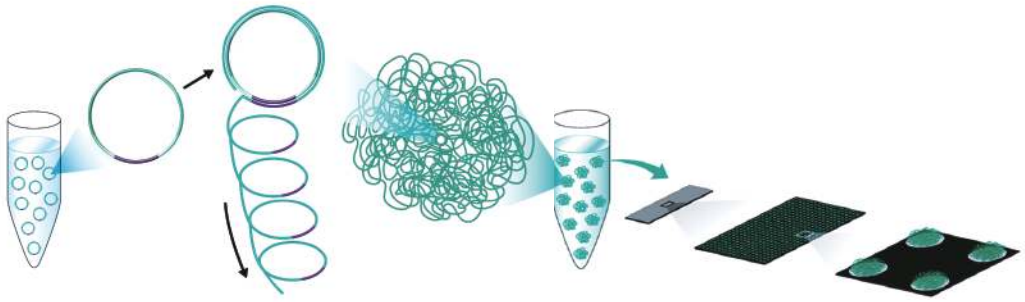
We care for your samples from the start through to the result reporting. Highly experienced laboratory professionals follow strict quality procedures to ensure the integrity of your results.



Quality Data | Fast TAT | € Cost Effective

DNBSEQ Sequencing Technology

DNBSEQ is an innovative high-throughput sequencing technology, developed by BGI's Complete Genomics subsidiary in Silicon Valley. The system is powered by combinatorial Probe-Anchor Synthesis (cPAS), linear isothermal Rolling-Circle Replication and DNA Nanoballs (DNB™) technology, followed by high-resolution digital imaging.
The combination of linear amplification and DNB technology reduces the error rate while enhancing the signal.
The size of the DNB is controlled in such a way that only one DNB is bound per active site on the flow cell. This densely patterned array technology provides optimal sequencing accuracy and increases flow cell utilization.



Gencove ImputeSeq Data Analysis

Gencove's ImputeSeq pipeline enables highly-accurate variant calling from low-pass sequencing data. Imputation accuracy outperforms genotyping arrays at common and rare variations. In addition, ImputeSeq returns ancestry and microbiome analysis for every sample.

Output data files are delivered in industry standard BAM and VCF files.

STANDARD ANALYSIS

- Read alignment
- Variant calling by imputation (reference panel: 1000 Genomes Phase 3)
- Ancestry inference
- Microbiome profiling

gencove
ImputeSeq Analysis

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, saliva, blood, fresh frozen tissue, cell pellets and FFPE samples, with the following general requirements:

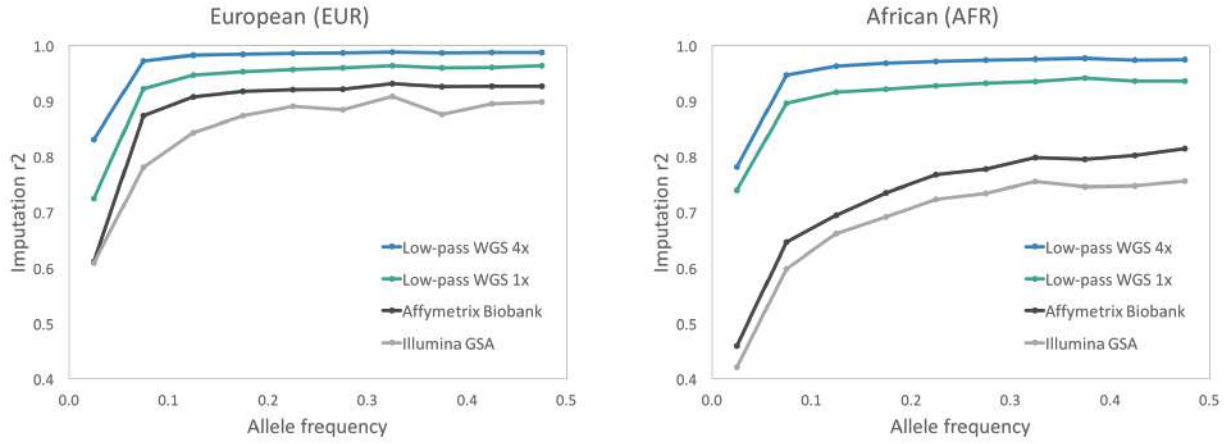
	DNA CONCENTRATION	MINIMUM SAMPLE VOLUME
Regular Samples	Intact genomic DNA ≥ 1μg, Concentration ≥ 12.5ng/ μl	15μl
PCR-free Samples	Intact genomic DNA ≥ 2μg, Concentration ≥ 12.5ng/ μl	15μl
Low Input Samples	Intact genomic DNA ≥ 200ng, Concentration ≥ 2.5ng/μl	15μl

Data Performance

SNP calls from imputed low-pass genome sequences are highly concordant with gold standard genotypes in the Genome in a Bottle (GIAB) sample. We sequenced the NA12878 sample to 4x coverage, performed down-sampling and imputation at different coverage levels, then compared the concordance at sites in the 1000 Genomes reference panel where the GIAB gold standard showed at least one non-reference allele.

Coverage	SNP Concordance with deep WGS at known variants
4x	99.5%
2x	99.4%
1x	99.1%

We additionally compared imputation quality from two commonly-used arrays (Affymetrix Precision Medicine Research Array and Illumina Global Screening Array) and low-pass sequencing.



Imputation accuracy from low-pass sequencing and two commonly used genotyping arrays in European and African populations

Request for Information or Quotation

Contact your BGI account representative for the most affordable rates in the industry and to discuss how we can meet your specific project requirements or for expert advice on experiment design, from sample to bioinformatics.

info@bgi.com

www.bgi.com

BGI Americas

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

BGI Europe

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

BGI Asia-Pacific

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

BGI Australia

L6, CBCRC, 300 Herston
Road, Herston, Brisbane,
Queensland 4006, Australia
Tel: +61 (07) 3362 0475

BGI Genomics BGI_Genomics

We Sequence, You Discover